[A001] Why adaptive walks can reach high-ranking peaks—a simplified Rough Mount Fuji model as a minimal example

Kye Hunter[1,2], Nora S. Martin[1]
[1]CRG (Barcelona Collaboratorium for Modelling and Predictive Biology), , Spain, [2]Facultat de Física, Universitat de Barcelona (UB), , Spain

Adaptive evolution can be modeled using a network of possible genotypes connected according to possible mutations, with a fitness value assigned to each genotype. Simple models of selection can use such fitness landscapes to simulate populations moving through this network in fitness-increasing steps. These walks end when reaching a genotype whose fitness exceeds that of all its neighbors—a local peak—and so important questions include whether these peaks have high or low fitness, and if populations are likely to be trapped at low-fitness peaks. In evolutionary simulations on several empirical landscapes, Li & Zhang (Mol. Biol. Evol. 2025) found that such fitness-increasing walks are likely to reach the globally-highest-ranked peaks among a large number of peaks, despite only using local information. They also found similar results in a mathematical model of fitness landscapes, the Rough Mount Fuji model.

We explain this by dividing the landscape into high- and low-fitness regions, and examine the number of peaks relative to the total number of genotypes in each region, finding that the low-fitness region can contain both most peaks, and a lower density of peaks than the high-fitness region. Using a simplified Rough Mount Fuji model, we find simple analytical explanations for how most populations can reach the highest-ranked peaks in that model. We then show that those same arguments can be successfully applied to the same empirical landscapes identified by Li & Zhang.

[A002] Bingo! Dino DNA: Chromosome-scale genome assembly of a multi-gigabase dinoflagellate protist fills a Tree-of-Life gap

Thomas Harvey[1], Gareth Gillard[1], Ole Kristian Tørresen[2], Morten Skage[2], Kjetill Jakobsen[2]
[1]Norwegian University Of Life Sciences, Ås, Norway, [2]University of Oslo, Oslo, Norway

Dinoflagellates represent a pivotal but under-sampled branch of eukaryotic diversity. They are abundant unicellular protists in marine and freshwater ecosystems, spanning free-living, symbiotic, and harmful bloom-forming lifestyles, and they play roles in global primary production and biogeochemical cycling. Despite this ecological importance, their exceptionally large, repeat-rich genomes and unconventional biology had long limited the availability of chromosome-scale references.

Here we have generated the first chromosome-scale genome assembly for the heterotrophic dinoflagellate Crypthecodinium cohnii, an industrially important DHA-producing strain. Using Oxford Nanopore long reads combined with Illumina Hi-C scaffolding, we assembled a 4.6 Gb genome comprising 122 chromosomes. The assembly showed strong continuity (contig N50 15 Mb; scaffold N50 42 Mb) and reached reference-level standards aligned with the Earth BioGenome Project. Our analyses captured hallmark dinoflagellate genome features, including extreme repeat expansion, extended unidirectional gene arrays, and domain-scale 3D organization.

This represents the largest and most complete dinoflagellate genome produced to date. It demonstrates that, with current sequencing and assembly technologies, even multi-gigabase, highly repetitive eukaryotic genomes are now obtainable, and illuminates a mysterious and under-sampled branch of the eukaryotic Tree of Life.

[A003] MeQTL Discovery in Admixed Human Genomes to Estimate Epistasis

Gillian Meeks[1], Oshiomah Oyageshio[1], Dr Rasika Mathias[2,3], Dr Dana Al-Hindi[1,4], Justin Myrick[1], Dr Caitlin Uren[5], Dr Marlo Möller[5], Dr Brenna Henn[1]

[1]University of California Davis, Davis, United States, [2]National Institute of Allergy and Infectious Diseases, Bethesda, United States, [3]Johns Hopkins Schools of Medicine and Public Health, Baltimore, United States, [4]University of California San Francisco, San Francisco, United States, [5]Stellenbosch University, Stellenbosch, South Africa

The extent to which epistatic interactions contribute to ancestry-specific genetic effect sizes in humans—and thereby limit the transferability of genetic prediction—remains unresolved. Progress has been constrained by two challenges: cross-continental comparisons confound epistasis with gene-by-environment effects, and prior estimates rely on too few traits to achieve statistical precision. We overcome these limitations using a newly generated dataset from an admixed South African population (n = 366). By comparing genetic effects across local ancestries within a shared environment, we directly estimate epistatic effects. This population comprises Khoe-San (the most divergent human ancestry), non-Khoe-San African, and out-of-Africa ancestries, enabling estimation of three pairwise ancestry-specific effect correlations across a range of genetic divergences. We integrate genome-wide CpG methylation data with paired DNA sequence data, treating methylation at individual CpGs as molecular phenotypes, yielding hundreds of thousands of traits to estimate the genome-wide genetic effect correlation. To validate the parameter space over which we have power to detect deviations from equivalent ancestry effect sizes, we use ancestry-matched simulated admixed genomes and simulate phenotypes calibrated to cis-methylation genetic architecture. Across simulations, we accurately recover ancestry effect correlations spanning the full range from 0 to 1, with a mean absolute error of only 6%. Finally, applying CARMA-X, a fine-mapping method designed for admixed populations, we achieve 98% power to recover the true causal variant across all simulated ancestry genetic effect correlation regimes. Together, this framework and our newly generated multi-omic dataset enable robust genome-wide estimation of ancestry effect correlations and fine-mapping of loci with ancestry-specific effects.

[A004] Convergence in responses to selection across two major plant pathogens

Jigisha Jigisha[1,2], Fabrizio Menardo[2], Daniel Croll[1]
[1]University of Neuchatel, , Switzerland, [2]University of Zurich, , Switzerland
Plant pathogenic fungi, characterized by large population sizes, mixed reproductive modes, and high mutation rates, show a high evolutionary potential to adapt to new environments. In agricultural settings, strong and recurrent selection imposed by resistant host cultivars, fungicide use, and local climate can further accelerate pathogen evolution. Comparing how different species experiencing similar conditions respond to these selective pressures can reveal the extent to which adaptation is convergent and help predcit evolutionary trajectories of pathogens. We analysed whole-genome sequence data from nearly 4,000 isolates of two major fungal pathogens of wheat, Blumeria graminis f. sp. tritici and Zymoseptoria tritici, the causal agents of powdery mildew and septoria leaf blotch, respectively. Using a global panel spanning four decades, we found broadly concordant continent-level population structure across species with marked differences at finer spatial scales. We then assessed population- and species-specific genomic responses to multiple sources of selection, including climate, fungicide exposure, and host resistance and found evidence for convergence in adaptive strategies, despite differences in the life history and genomic architecture of the two species. Our findings highlight convergence as a pervasive feature of pathogen evolution under strong selection, and have broader implications for disease management.

[A005] Effect of incubation temperature on transposon activity in the African house snake (Boaedon fuliginosus)

Mariela Tenorio[1], Tomáš Pšenička[1], Sabrina Carli[1], Michail Rovatsos[1]
[1]Department of Ecology, Faculty of Science, Charles University, Prague, Czech Republic
Incubation temperature is a major environmental factor influencing embryonic development, survival, and hatching phenotype in oviparous squamate reptiles. Despite the recent efforts to understand the effect of incubation temperature on organisms and populations, we still lack in-depth information considering the impact of egg incubation temperature on various aspects of genome regulation and evolution. Among amniotes, oviparous reptiles are an ideal model group for thermosensitivity studies, as the majority of the species lack parental care and embryonic development is exposed to ambient conditions in the nest. Repetitive elements represent a major fraction of reptilian genomes and their activity is often regulated by epigenetic mechanisms. There is accumulating evidence that ambient temperature can affect transposon activity in reptiles. In this project, we investigate the effect of incubation temperature on transposon activity in hatchlings of the African house snake (Boaedon fuliginosus). In total, 12 eggs were separated in two groups and incubated at 26°C and 30°C, respectively. Total RNA was isolated from blood and liver from hatchlings and sequenced using Illumina platform to explore the effect of incubation temperature on genome regulation, with a particular focus on repetitive elements and transposons, which tend to accumulate on differentiated sex chromosomes. Our study revealed novel insights into the impact of temperature as a variable affecting evolutionary process and may help extrapolate predictions for understanding the genome evolution in reptiles.

[A006] An evolutionary ghost hunt: hybridisation between island endemic bird species

Abby Williams

Hybridisation between species was once considered an uncommon occurrence, but is now recognised to occur frequently across many different taxa. It can result in homogenisation of previously distinct forms (a potential conservation issue), but can also act as a catalyst for diversification through introgression of favourable genes. Repeated rounds of island colonisation followed by speciation result in secondary sympatry, with the potential for hybridisation between early and late arrivers. In the southwest Pacific, this situation has arisen in the avian family Zosteropidae (the white-eyes). Here we use whole genome sequencing of live birds and historical specimens to characterise hybridisation between three white-eye species on Norfolk Island: two island endemics, Zosterops tenuirostris and the now-extinct Zosterops albogularis, and Zosterops lateralis, which colonised the island in 1904. Despite over two million years of divergence between Z. lateralis and the two endemics, we provide genomic evidence of their hybridisation. First, we confirm the identities of three Z. lateralis x Z. tenuirostris hybrids and additionally identify one Z. lateralis x Z. albogularis hybrid. We also report asymmetric, genome-wide introgression from both endemics into Z. lateralis, with introgressing regions of the genome enriched for a range of potential functions. However, despite this introgression, species boundaries have been maintained, and the extant endemic Z. tenuirostris does not appear to be at risk of genetic extinction. This study sheds light on the genomic outcomes of secondary sympatry and its potential consequences for single-island endemics.

[A007] Extremotolerance Meets Genetic Accessibility in a Non-Model Microalga from Cuatro Ciénegas Basin

Abril Beltran[1], Mr. Even Ramos[1], Dr. Hamlet Avilés[1], Dr. Susana De la Torre[1]
[1]Institute of Biotechnology, Universidad Autónoma de Nuevo León, San Nicolás de los Garza, México

Extremotolerant microalgae from isolated environments represent promising yet underexplored systems for molecular and evolutionary research. Microalgae from the Cuatro Ciénegas Basin, a geographically isolated system of ponds, springs, and rivers within the Chihuahuan Desert in northern Mexico, provide a unique opportunity to study biological resilience in non-model lineages. However, their use in experimental frameworks is often limited by the assumption that physiological resilience is linked to reduced genetic accessibility, excluding many extremotolerant taxa from molecular studies.

Here, we evaluated an extremotolerant microalgal strain isolated from the Cuatro Ciénegas Basin as a candidate for genetic transformation. Prior to genetic manipulation, the wild-type strain (Churince FTE 3) was characterized across multiple stress conditions, including growth under acidic to highly alkaline pH values, elevated temperatures, and increased salinity, encompassing parameters not directly associated with its native environment. This wide stress tolerance suggests a high degree of physiological plasticity and supported its selection as a robust non-model system. Transformation assays were performed using electroporation with the pChlamy4 plasmid, followed by selection on zeocin-containing media.

Putative transformant colonies were recovered under selective conditions and showed sustained growth, indicating successful genetic integration. Although transgene expression and post-transformation extremotolerance were not assessed, the recovery of transformants demonstrates that genetic accessibility is not inherently precluded by prior physiological resilience.

Overall, these findings indicate that extremotolerant microalgae from the Cuatro Ciénegas Basin are viable candidates for genetic manipulation and highlight the role of physiological plasticity in shaping environmental resilience and genetic accessibility in non-model lineages.

[A008] The influence of chromosomal inversions on local adaptation of Atlantic silversides, Menidia menidia

Adam Ziegler[1], Dr. Maria Akopyan[2], Dr. Nina Overgaard Therkildsen[3], Hannes Baumann[4], Jessica Rick[5]
[1]Genetics Graduate Interdisciplinary Program, University Of Arizona, Tucson, United States, [2]Department of Evolution, Ecology, and Organismal Biology, University of California, Riverside, Riverside, United States, [3]Department of Natural Resources and the Environment, Cornell University, Ithaca, United States, [4]Department of Marine Sciences, University of Connecticut, Storrs, United States, [5]Department of Natural Resources and the Environment, University of Arizona, Tucson, United States

The Atlantic silverside, Menidia menidia, is a prominent species in estuarine ecosystems along the eastern United States and Canada, ranging from Florida to Nova Scotia. This expansive range covers varying climates and season lengths, resulting in locally adapted populations that show morphological and physiological differences. Recent work with silversides at opposite ends of their range has discovered numerous chromosomal inversions in the silverside genome, with three large inversions showing significant association with observed phenotypic differences between northern and southern populations. Here, we explore these three inversions (on chromosomes 11, 18, and 24) in silversides collected from Massachusetts (n=44) and North Carolina (n=67) and their lab-reared offspring (n=2,015), to characterize the influence of inversion karyotypes on phenotypes and the interaction between inversion karyotype and rearing temperature. We use a genotyping-in-thousands-by-sequencing approach to sequence 360 targeted loci. As expected, we found linkage disequilibrium within inversions to be greater than outside of the inversions. Additionally, haplotype diversity within inversions suggests that recombination does still occur within these inverted regions. Thus, while recombination is reduced in inversions, it is not completely suppressed. We examine these patterns and explore the mechanisms of recombination within inversions, leveraging this diversity to identify candidate loci underlying adaptive traits. Finally, we utilize GWAS-like analyses to illuminate the influence these inversions have on silverside local adaptation. This work provides important context to better understand the ecological role inversions play in the silverside's ability to adapt to its local environment, as well as the mechanisms involved in inversion-related adaptation more generally.

[A009] Evolution of ecological interactions in Pseudomonas aeruginosa across different stress regimes

Aditee Ashar[1], Professor Rees Kassen[1]
[1]McGill University, Department of Biology, Montreal, Canada

Species interactions shape the structure and function of ecological communities, yet we know little about how these interactions evolve as populations adapt to their environments. The Stress Gradient Hypothesis (SGH) predicts that interactions shift from competition under benign conditions to facilitation under severe stress, but experimental tests of whether facilitative strategies evolve de novo under stress are lacking. Here, I address when and why facilitative interactions evolve and how genomic changes within populations drive these shifts.

I combine experimental evolution, quantitative interaction mapping, and population genomics to investigate how ecological interactions evolve within microbial populations adapting to environmental stress. Using Pseudomonas aeruginosa, an opportunistic pathogen in cystic fibrosis lung, I evolved replicate populations for 250 generations under four environmental regimes varying in nutrient availability and osmotic stress, including conditions designed to mimic the cystic fibrosis airway. I systematically map pairwise interactions among evolved isolates using flow cytometry-based assays with fluorescent markers, quantifying bidirectional interaction effects comparing strain-specific yields in monoculture and coculture.

Interaction data will be integrated with whole-population genome sequencing to identify parallel evolutionary trajectories and recurrent mutations associated with stress adaptation and altered interaction patterns. I will track temporal dynamics of beneficial mutations showing parallel evolution across replicate populations. Candidate genes include osmo-protective genes (algU, mucA) and quorum-sensing genes (lasR), which may underlie divergent interaction strategies across stress treatments. Together, this work links molecular evolution to emergent ecological interactions, providing experimental evidence for when facilitation evolves under stress and mechanistic insight into how genomic change shapes interaction dynamics.

[A010] Small, Isolated, and Inbred: Genomic Evidence of Strong Population Structure in the Australian humpback dolphin along the Northwestern Australian Coastline

Dr. Adrien Tran Lu Y[1], Dr. Delphine B. H. Chabanne[1,2], PhD Simon J Allen[1,3], PhD Tim Hunt[4], Prof. Tomas Marques Bonet[5,6,7,8], Dr. Guido J. Parra[4], PhD Holly C. Raudino[9], PhD Kelly Waples[9], PhD Erik P. Willems[1], Prof. Michael Krützen[1]

[1]Evolutionary Genetics Group, Department of Evolutionary Anthropology, University of Zurich, Zurich, Switzerland, [2]Centre for Sustainable Aquatic Ecosystems, Harry Butler Institute, Murdoch University, Murdoch, Australia , [3] Ocean Institute, School of Biological Sciences, University of Western Australia, Perth, Australia , [4]Cetacean Ecology, Behaviour and Evolution Lab (CEBEL), College of Sciences and Engineering, Flinders University, Bedford Park, Australia , [5]Department of Medicine and Life Sciences, Institute of Evolutionary Biology (CSIC-UPF), Pompeu Fabra University, Barcelona, Spain, [6]Institut Català de Paleontologia Miquel Crusafont (ICP-CERCA), Universitat Autònoma de Barcelona, Edifici ICTA-ICP, Cerdanyola del Vallès, , Spain, [7]Catalan Institution of Research and Advanced Studies (ICREA), Barcelona, Spain, [8]Centro Nacional de Análisis Genómico (CNAG), Barcelona, Spain, [9]Marine Science Program, Department of Biodiversity, Conservation and Attractions, Perth, Australia

The Australian humpback dolphin (Sousa sahulensis) is endemic to the tropical coastal waters of northern Australia and southern Papua New Guinea, yet their population genetic structure remains poorly understood despite its listing as Vulnerable under the IUCN Red List. To improve understanding of its population biology, we sampled 46 individuals from five localities across the northwestern Australia coastline, spanning more than 1,000 km from Exmouth to Cone Bay. Using low-coverage whole-genome sequencing, we investigated population genetic structure, relatedness, and inbreeding in this elusive species. Our analyses revealed two highly differentiated populations separated by a clear genetic break between Port Hedland and Cygnet Bay, with additional subtle genetic structure among localities. Patterns of relatedness showed pronounced family structure within localities. A Generalized Linear models (GLMM) indicated no effect of sampling date but coastline distance significantly predicted relatedness patterns. Additionally, individuals of different sexes were less related than individuals of the same sex. To explore the consequences of this high relatedness among localities, we also estimated inbreeding coefficients and runs of homozygosity (ROH). Both metrics were high suggesting small and isolated populations, with long term and recent inbreeding. Overall, our findings provide the first genomic evidence of strong population subdivision, pronounced relatedness and high inbreeding patterns in S. sahulensis, consistent with previous studies with microsatellite and mitochondrial markers. These results corroborate that Australian humpback dolphin populations are demographically small and isolated, underscoring the need for locality-specific management, improved connectivity assessment, and targeted monitoring of genetic diversity in future conservation and research efforts.

[A011] Specific mutation bias predicts the emergence of antibiotic resistance

Adrita Chakraborty[1], Dr Deepa Agashe[1]
[1]National Centre For Biological Sciences, Bengaluru, India

Rapid, high-level antibiotic resistance in bacteria often involves specific mutations in target genes, like streptomycin resistance via A→C mutations in rpsL. Therefore, bacteria that sample more A→C mutations should evolve streptomycin resistance faster. Although rapid sampling of such mutations is relatively rare in wild-type bacteria, mutator strains with defective DNA repair genes may have the requisite mutation spectrum, and evolve resistance faster. However, one question remains: can we disentangle the role of mutation bias and rate in the context of antibiotic resistance, and determine which one has a greater impact? We hypothesized that under strong antibiotic selection, a high mutation rate should facilitate adaptation by shortening the time to sample a beneficial resistance mutation. Furthermore, strains whose mutation spectra align with high-benefit resistance mutations in target genes will be more likely to evolve higher resistance. We exposed Escherichia coli mutator strains with varying mutation rates and spectra to high concentrations of antibiotics targeting different cellular functions over a single growth cycle, and measured survival. For most antibiotics, strains with the prerequisite mutation bias for the resistance mutation acquired highest resistance, and sequencing revealed mutations in target and efflux pump genes match the mutation spectra of the respective mutators. However, the highest mutation-rate mutators did not always evolve the highest antibiotic resistance, suggesting that mutation bias has a stronger impact in emergence of antibiotic resistance mutations than mutation rate. Given that mutators often arise spontaneously in laboratory, natural, and clinical settings, we suggest that their mutation spectra deserve more attention.

[A012] Examining Microbial Differences in a Cultural Context: A preliminary study of Medieval Nicaea (İznik)

Ahmet Berkay Demirsecen[1], Baran Acar[2], Dr. Duygu Deniz Kazanci[2], Dr. Melda Meral Ocal[1], Dr. Gulsah Merve Kilinc[3], Dr. Fusun Ozer[4], Prof. Dr. Yilmaz Selim Erdal[4], Dr. Emrah Kirdok[1]
[1]Department of Biotechnology, Faculty of Science, Mersin University, Mersin, Turkey, [2]Department of Biological Sciences, Faculty of Arts and Sciences, Middle East Technical University, Ankara, Turkey, [3]Department of Bioinformatics, Institute of Health Sciences, Hacettepe University, Ankara, Turkey, [4]Department of Anthropology, Faculty of Literature, Hacettepe University, Ankara, Turkey

The human oral microbiota is a dynamic archive that is shaped by the interaction of host lifestyle and environmental exposure. Ancient dental calculus serves as a resilient reservoir for microbial and eukaryotic DNA, enabling high resolution reconstruction of historical oral health, diet, and palaeoenvironment. Here, we investigate medieval Nicaea (İznik,Türkiye), a unique diverse setting where distinct cultural groups coexisted. This diversity could allow us to disentangle how specific cultural practices and social traditions shaped the oral ecosystem within a shared environment.

In this study we sampled dental calculus material from Crusader (N=13), local Muslim (N=10), and Christian (N=12) individuals excavated from three distinct burial sites in Nicaea.

Our preliminary KrakenUniq classification results (all individuals, N=35) reveal a typical oral microbial profile that contains red and orange complex periodontal opportunistic pathogens. Moreover we found a high abundance of Streptococcus species including S. pneumonia, S. sanguinis and S. mutans. While the former two are responsible for respiratory infections and systemic diseases, the latter one is the predominant cause for dental caries. We are currently performing alignment-based analysis to generate robust taxonomic classifications and authenticate ancient DNA specific deamination patterns.

By integrating this genomic data with archaeological context, we aim to investigate microbial differences that could be associated with lifestyle and cultural patterns.

[A013] Nonhuman Primate Models for Hominin Introgression and Genetic Admixture

Alaina Brenner[1], Dr. Brenna Henn[1], Dr. Timothy Weaver[1]
[1]UC Davis, Davis, United States

Hybridization and genetic admixture occur across many mammalian taxa past and present, including isolated lineages of late Pleistocene hominins. However, the dynamics of these genetic exchanges in the first few generations of admixture could result in positive or purifying selection. Therefore, this project investigates the mechanisms of hybridization and admixture in the genomes of contemporary nonhuman primates as a model to understand hominin introgression events in human evolution. Here, we use whole genome sequencing (WGS) to assess local and global ancestry of an admixed colony of 5 generations of 140 macaques once housed at the California National Primate Research Center (CNPRC). Indian and Chinese rhesus macaques are especially relevant due to their similar divergence time (in generations) to humans and Neandertals. These macaque subspecies have accumulated substantial genetic variation since the divergence and subsequent isolation of the two lineages ~160,000 years ago. We identify over 7.5 million SNPs in this dataset, over half of which are segregating at >5% MAF. Inbreeding coefficients in the colony were surprisingly low, with a mean fROH of 0.02. After using local ancestry assignment to discriminate between Indian and Chinese macaque haplotypes, we applied a genome-wide post-admixture scan to evaluate deviations in local ancestry tract distributions. Using forward simulations with SLiM, we can then test whether this pattern is consistent with purifying selection against hybrids in these and extended generations. Simulating this macaque model in comparison to human and Neandertal admixture will further understanding of hybridization signatures within the early generations.

[A014] Inference of the distribution of fitness effects using local genealogies

Alan Izarraras-Gomez[1], Christian Huber[2], Charleston Chiang[3], Diego Ortega-Del Vecchyo[1]
[1]International Laboratory For Human Genome Research, National Autonomous University of Mexico (LIIGH-UNAM), Juriquilla, Mexico, [2]Department of Biology, Pennsylvania State University, State College, USA, [3]Center for Genetic Epidemiology, Department of Population and Public Health Sciences, Keck School of Medicine, University of Southern California, Los Angeles, USA
The distribution of fitness effects (DFE) measures the probability of new mutations having a certain selection coefficient, and as such is central to understanding how natural selection shapes genetic diversity. The DFE has previously been inferred using different summary statistics, such as the Site Frequency Spectra (SFS). A drawback of these approaches is that they focus on summary statistics that might not carry the complete evolutionary features available in genetic data. Notably, all patterns of genetic variation can be derived from the Ancestral Recombination Graph (ARG), a data structure that represents the evolutionary history of a population as a set of local genealogies across the genome. Accordingly, both natural selection and demographic changes are contained within the ARG and cause alterations in coalescent times. In this study, we develop a Monte Carlo approach that first infers a demographic model from synonymous sites to then derive the parameters of a DFE using non-synonymous sites. This is done by computing a likelihood function that uses information from the number of derived allele lineages at different discrete time points from a sample of local trees. We show through simulations that this approach can accurately infer the parameters of a DFE and we apply our method to infer the DFE of new non-synonymous mutations of a hundred unrelated Yorubans from the 1,000 genomes project. We find that the most common type of mutations are highly deleterious (|s| > 10^-2) followed by neutral mutations (0 ≤|s|<10^-5).

[A015] Complex and Messy Prebiotic Chemistry: Obstacles and Opportunities for an RNA World

Alberto Vázquez-Salazar[1]
[1]Cinvestav, Mexico City, Mexico

Traditional prebiotic chemistry experiments often isolated single reactions under clean, controlled conditions, yet early Earth was chemically diverse and physically dynamic. Such primordial complexity likely imposed obstacles, including side reactions, low yields, and unstable intermediates, but it also generated opportunities, including redundant routes, parallel pathways, and environmental filters that could bias mixtures toward subsets of persistent and chemically productive compounds. This work examines how heterogeneous prebiotic settings could generate RNA precursors, including nucleobases, ribose, and phosphate-containing species, through multiple concurrent pathways. Although side reactions can sequester carbon in inert tars and reduce yields of specific targets, networked chemistry can also enhance robustness when different routes converge on shared intermediates, or when apparent byproducts reenter productive cycles. En-vironmental factors such as ultraviolet irradiation, mineral surfaces, wet-dry cycling, and thermal gradients can act as constraints that enrich certain products by differential sta-bility, reactivity, and compartmentalization. In this context, the RNA world hypothesis remains compelling because RNA can store heritable sequence information and catalyze reactions through sequence dependent folding, thereby linking heredity and chemistry within a single polymer. At the same time, the emergence of functional sequence in-formation and of control architectures that couple sequence to reproducible function remains a central open problem, and it sets clear limits on what chemistry alone can explain. Rather than dismissing messy mixtures as irrelevant noise, it is more accurate to treat them as the native context in which concentration mechanisms, environmental cy-cling, and selective persistence could enable the accumulation and survival of RNA re-lated molecules.

[A016] Ancient infectious pressures shaped the evolution of LRRK2 and PRKN

Alessandra Mozzi[1], Rachele Cagliani[1], Diego Forni[1], Uberto Pozzoli[1], Manuela Sironi[2]
[1]Scientific Institute IRCCS E. Medea, Bosisio Parini (LC), Italy, [2]School of Medicine and Surgery, University of Milano-Bicocca, Monza (MB), Italy

LRRK2 (leucine-rich repeat kinase 2) and Parkin (encoded by PRKN) participate in shared molecular pathways and are implicated in the pathogenesis of Parkinson's disease (PD), leprosy, and other diseases. While leprosy likely imposed strong evolutionary pressure, PD's late onset renders it largely invisible to natural selection. Here, we investigated the evolutionary history of LRRK2 and PRKN across primates and human populations. We identified both genes as targets of positive selection in primates, but also detected strong constraints on LRRK2 disease-associated positions. In the case of PRKN, we report that gibbons carry a disease mutation (R33Q) that causes protein instability. Using ancient DNA, we reconstructed the temporal dynamics of LRRK2 and PRKN variants in European populations. The most common PD risk allele, LRRK2 G2019S—which also confers protection against salmonellosis—was absent in Paleolithic individuals but increased rapidly in frequency during the Early Neolithic. We propose that this rise was driven by selective pressure associated with the emergence of human-adapted Salmonella lineages. Analysis of leprosy susceptibility variants in LRRK2, PRKN, and other major loci showed that the frequencies of several risk alleles increased prior to the Middle Ages, when leprosy peaked in Europe. In contrast, we found little evidence for strong selection acting on these variants during the medieval leprosy epidemic itself. Together, our results provide a temporal framework for the emergence and possible positive selection of a major PD-associated mutation and suggest that selective forces—potentially unrelated to Mycobacterium leprae—shaped genetic susceptibility to leprosy before its medieval peak.

[A017] Investigation of temporal changes to gene expression across the reproductive cycle of the zebra finch (Taeniopygia guttata)

Alexander Lawrence[1], Dr Rachel Steward[1], Petronella Wessman[1], Dr Kalle Tunström[1], Dr Kristaps Sokolovskis[2], Anna Runemark[1]
[1]Lund University, Lund, Sweden, [2]Max Planck Institute of Animal Behavior, Radolfzell, Germany
During the avian reproductive cycle individuals undergo vast morphological and physiological changes, especially in the gonads. We aimed to elucidate how these changes are mediated via changes in gene expression. We assessed gene expression in females across the reproductive cycle, and in males in response to the reproductive stage of the partner by exploring differences in expression at the levels of individual genes, gene functions, and of modules of co-expressed genes. We used the zebra finch (Taeniopygia guttata) as a study system. Zebra finches are opportunistic breeders and will enter reproductive mode when light regime and food conditions are met and a partner is available. Our experimental setup paired male and female zebra finches in suitable breeding conditions. We analyzed RNA from 8 individuals per sex from three timepoints. 1. Pre-reproduction, when a nest had been built. Pre-reproductive mode was confirmed during dissection by the presence of an egg in the oviduct. 2. During reproduction, when two eggs had been laid on consecutive days. This timepoint was selected to confirm that the eggs were fertilised as a single egg can be unfertilised. 3. Post-reproduction/incubation, three days after the final egg of a clutch had been laid. This research will inform future work on gene expression in avian species by uncovering which genes, gene functions, and gene co-expression networks are involved in reproduction and differentially expressed across reproductive stages. Additionally, it will uncover the extent to which male gene expression is influenced by the social context of the partner's reproduction.

[A018] Maladaptive aphid-induced transgenerational plasticity is overcome in nature

Alexandra Chávez[1], Amanda de Santana Lopes[1], Martin Lohr[1], Lisa-Marie Laick[1], Martin Schäfer[1], Veit Grabe[2], Shuqing Xu[1], Martin Kaltenpoth[2], Meret Huber[1]
[1]Institute Of Organismic And Molecular Evolution, Johannes Gutenberg-Universität Mainz, Mainz, Germany, [2]Max Planck Institute for Chemical Ecology, , Germany

Whether environment-induced phenotypes are transgenerationally inherited is controversial, as the prevalence, fitness impact, and underlying molecular mechanisms are largely unclear. We assessed these aspects by growing monoclonal, single descendant lineages of the giant duckweed Spirodela polyrhiza under transgenerationally recurring herbivory of the waterlily aphid Rhopalosiphum nymphaeae, both indoors and outdoors. Surprisingly, aphid herbivory experienced five generations ago boosted aphid population growth rates and decreased fitness of the eight duckweed genotypes. Plant and aphid fitness were likely affected by transgenerationally plastic levels of tyramine and jasmonates, albeit metabolite plasticity occurred in different ways: tyramine levels were induced by aphids and these elevated levels were transgenerationally retained under control conditions and further primed under recurring aphid herbivory. This priming likely resulted from the overexpression of two L-tyrosine decarboxylases, the sole biosynthetic enzymes in tyramine production. When overexpressed and artificially supplemented to aphids, tyramine reduced aphid reproduction, suggesting that transgenerationally plastic tyramine levels protect plants across generations. In contrast, the defence jasmonate phytohormones were induced only under transgenerationally recurring aphid herbivory. Unexpectedly, externally applied jasmonates at mild concentrations boosted aphid fitness, implying that aphids manipulate plant defences for their own benefit. Moreover, when grown in large populations outdoors, only tyramine, but not jasmonates, was transgenerationally plastic upon recurring aphid herbivory. Consequently, previous aphid herbivory ultimately benefited rather than reduced plant fitness under recurring herbivory. Taken together, these data show that aphids can manipulate plant defences for their own benefits, possibly via epigenetic inheritance, although plants may overcome such manipulations in natural environments.

[A019] Evaluating the Role of Epistatic Selection in Maintaining Extended Haplotypes in the MHC

Alfredo Marin[1], MSc André Maróstica[1], Professor Montgomery Slatkin[2], Professor Diogo Meyer[1]
[1]Universidade De São Paulo, São Paulo, Brazil, [2]University of California, Berkeley, Berkeley, United States

The conserved extended haplotypes (CEHs) of the MHC region span genes separated by over 3Mb. Even though the MHC displays the highest diversity in the human genome, particularly at the HLA loci, and recombination is expected to break down the haplotypes, the CEHs still reach high frequencies in human populations (>1%). In this work, we tested if this observation can be explained only by the already well established selective pressures acting on the individual HLA loci or if some process that favors combinations of alleles at different loci, such as epistatic selection, is needed. We simulated two loci under a heterozygote advantage regime to model balancing selection maintaining MHC polymorphism. After a period of burn-in, epistasis was introduced by giving a bonus fitness to haplotypes that contain specific combinations of alleles. Our results show that, for the range of parameters we explored (2Ns=0 to 500 for symmetric heterozygote advantage and 2Ns=0 to 200 for epistasis), the overall diversity is much more sensitive to the heterozygote advantage than to epistasis. Only when increasing the intensity of epistasis to very high levels do we see haplotypes rising to frequencies appreciably higher than the balancing selection equilibrium. These results suggest that, although epistatic haplotypes have been reported in the MHC, their fitness effects in natural populations are unlikely to be sufficient to explain the high frequencies observed for CEHs.

[A020] Tandem Repeats in Adaptive Archaic Introgression Across the Human Genome

Alicia Bierly[1], Melody Chang, Hannah Siegel, Mary Crowell, Dr. Carolina Adam, Rori Rolfhs
[1]School of Computer and Data Sciences, University of Oregon, Eugene, United States

Tandem repeats (TRs) are highly variable DNA elements characterized by consecutive motif repetitions, influencing gene expression and protein structure. Their high mutation rates make them compelling sources of adaptive variation in human evolution. An evolutionary process that can be studied is adaptive introgression, defined as the transfer of favorable alleles aiding adaptation, using archaic tracts. These regions of the genome inherited from archaic humans sometimes carry segments of adaptive alleles that were maintained through positive selection. While the role of single-nucleotide polymorphisms (SNPs) in adaptive introgression has been studied, the role of TRs remains unexplored. Scientists studying the MUC19 gene using SNPs identified a positively selected introgressed tract which contains a TR, highlighting the potential in this research. To date, MUC19 represents one example, but no genome-wide study has examined this before. Recent advances in long-read sequencing and introgression mapping enable the investigation of TRs within introgressed regions and offer insights into their joint relationship in human adaptation. Our goal is to investigate the broad role of TRs across the genome in adaptive archaic introgression. Using 1000 Genomes dataset, we constructed a per-individual-haploid table of TR loci and introgressed tracts across 663 individuals. We used LongTR to genotype TRs and hmmix to identify specific genomic tracts of archaic origin. These datasets were merged to determine which TRs overlapped introgressed tracts. Our analysis investigates patterns of divergence in TRs and their relation to archaic introgression. These results could implicate TRs as a source of importance in exploring variation in evolution.

[A021] Comparison of cloacal and fecal microbiomes between two populations of sand lizards (Lacerta agilis)

Alina Fomin[1], Charlotte Enkvist[1], Seraina E. Bracamonte[1], María Cortázar-Chinarro[1], Elin Videvall[1], Mette Lillie[1]
[1]Department of Ecology and Genetics; Animal Ecology, Uppsala Universitet, Uppsala, Sweden
The gut microbiome is involved in many functions of the body, including nutrient absorption, development, behavior and immunity. The gut microbiome is also dynamic; a finely balanced symposium of components that can facilitate fast adaptation to changing environments. Individuals within a population often share broadly similar gut microbiomes, and in small populations, high relatedness, shared lifestyles, and limited geographic ranges further promote this similarity. Microbiome composition is thus strongly shaped by host species and genetic structure but also by external factors such as the diet and environment of the host. When these factors fail to support a healthy gut microbiome, hosts may become more susceptible to disease and experience significantly lower fitness. Therefore, the microbiome may give valuable insights into host health and fitness. This study investigates the sand lizard microbiome using 16S rRNA gene sequencing of cloacal and fecal samples as proxies for the gut microbiome. We are comparing the 16S microbiome composition and diversity across two focal populations of Swedish sand lizard (Lacerta agilis), which have contrasting evolutionary and demographic histories. One population is native, with reduced genetic diversity, and the other is a recently admixed population which was established approximately 20 years ago. The differences in microbiome composition and diversity between these two populations will be investigated in relation to population genetic diversity to gain insights into how the gut microbiome is shaped in sand lizards and build ground work for future conservation efforts.

[A022] Leveraging variation of matri- and patrilinear genomes reveals contrasting patterns of sex-bias dispersal in northern sea lions

Aliya Yakupova[1,2], Camille Kessler[1], Lara Baggio[1], Dr. Claire Peart[1], Dr. Stefanie Grosser[1], Dr. Fidel Botero-Castro[1], Professor Amy Bickham Baird[3], Professor Emeritus John Bickham[4], Professor Joe Hoffman[5], Professor Jochen Wolf[1,2]

[1]Devision of Evolutionary Biology, University of Munich (LMU), Munich, Germany, [2]Department of Microevolution and Biodiversity, Max Planck Institute for Biological Intelligence, Seewiesen, Germany, [3]Department of Natural Sciences University of Houston, Houston, USA, [4]Department of Ecology and Conservation Biology, Texas A&M University, College Station, USA, [5]Department of Evolutionary Population Genetics, Bielefeld University, Bielefeld, Germany

Dispersal is a fundamental driver of species evolution, as it mediates gene flow between populations and has consequences for population structure. In particular, sex-biased dispersal creates predictable genomic patterns characterised as stronger population structure in markers inherited through the philopatric sex, and lower structuring in those inherited through the dispersing sex. It is therefore possible to leverage the variation in independent genomic compartments to quantify dispersal asymmetry. The northern sea lions provide a classic example of A polygynous mating system with limited observational evidence for male-biased dispersal. Here, we investigated sex-biased dispersal in Galápagos (Zalophus wollebaeki; GSL) and Steller sea lions (Eumetopias jubatus; SSL) using whole-genome resequencing data across four genomic compartments, quantifying female-only (mtDNA), mixed (autosome, X chromosome), and male-only (Y chromosome) genetic variation. We found a clear pattern of male-biased dispersal in SSL, as well as strong isolation by distance driven by female philopatry. In contrast, for GSL, our results suggest that females disperse more than previously thought, and that isolation by environment is a main factor explaining population structure for both sexes. Our results emphasise the importance of integrating multiple genomic compartments when interpreting dispersal and connectivity in marine mammals.

[A023] Close together, far apart: does the localization of QTL impact plant domesticability?

Anastasia Kolesnikova[1], Prof Abdullah Kahraman[2], Dr Adrian Brennan[3], Dr Yann Bourgeois[4], Prof Mark Chapman[1]
[1]University Of Southampton, , United Kingdom, [2]Harran University, , Turkey, [3]University of Durham, , United Kingdom, [4]Institut de Recherche pour le Développement, , France

Before the advent of agriculture, humans collected and cultivated a large variety of plants. Out of the thousands of managed species, only 250 are considered domesticated, with the rest having been abandoned. The reason why certain wild plants were successfully domesticated, and not others, remains unknown.

We use the term domesticability to describe an organism's ability to be domesticated. Genetic architecture, or the localization of QTL associated with traits of interest may play a role in domesticability, as recombination may separate or bring together QTL associated with traits selected for during domestication. In addition, QTL effect size may play a role in domesticability, as a large-effect QTL may be easier to select compared to several small-effect QTL.

Co-localization between QTL for traits of agricultural interest may increase domesticability, as beneficial trait pairings will be consistently inherited by offspring. Conversely, large distances between QTL of interest may decrease domesticability, as recombination could separate beneficial trait combinations. We test the hypothesis that QTL of agricultural relevance in progenitor species are more frequently co-located compared to QTLs in never-domesticated wild (NDW) species.

We grew and phenotyped two F7 populations of progenitor (C. reticulatum) x domesticated (C. arietinum) and NDW (C. echinospermum) x domesticated chickpea crosses. Using ddRAD-seq, 96 individuals from each population were genotyped and QTL maps for agriculturally important phenotypes were generated with rqtl2. We test for differences in the localisation of QTL and discuss whether genomic architecture could have contributed to the domesticability of progenitor species.

[A024] Insights into the genomic diversity and domestication of white lupin using pangenome graphs

Matteo Tommaso Ungaro[5], Enrico Bortoletto[2], Magdalena Kroc[3], Karolina Susek[3], Umberto Rosani[2], Elena Bitocchi[4], Dr Andrea Benazzo[1]
[1]University of Ferrara, Ferrara, Italy, [2]University of Padova, Padova, Italy, [3]Polish Academy of Sciences, Poznan, Poland, [4]Polytechnic University of the Marches, Ancona, Italy, [5]University of Rome "La Sapienza", Rome, Italy

Domestication is a human-mediated process promoted by the transition from a hunter-gatherer subsistence economy to a farmer-oriented one. In the white lupin (Lupinus albus), such a process was potentially initiated in the Old World by ancient Greeks; from then onwards, different varieties of this species have been gradually and locally selected across the Mediterranean basin. However, origins and whether domestication involved a single or multiple events remain unclear. Furthermore, genomic structural variants (SV) are recognized for their role in domestication, contributing to local adaptations to diverse environmental conditions in many species. Established bioinformatics approaches combining short reads and linear reference genomes are effective in SNP and INDEL discovery, but they systematically fail to capture the full spectrum of SV, and, consequently, the impact of SV on domestication is often understudied.

To overcome these limitations and investigate the genomic diversity underlying white lupin domestication, we combined PacBio HiFi reads and Hi-C data to construct a chromosome-scale pangenome graph incorporating 16 phased genome assemblies of both wild and landrace accessions from across the Mediterranean basin. The pangenome graph was then integrated with over 300 short-read whole-genome resequenced accessions to characterize genomic variation, reconstruct the domestication history of white lupin, and identify key genomic regions targeted by artificial selection. Our pangenome-based approach offers novel insights into the complex genomic changes associated with white lupin domestication and highlights the contribution of SVs in this process.

[A025] The role of taxonomically restricted genes in brown algal multicellularity

Andrea Celeste López-Díaz[1], PhD Susana M. Coelho[2], PhD Josué Barrera-Redondo[1]
[1]Center For Research And Advanced Studies Of The National Polytechnic Institute (CINVESTAV) Irapuato Unit, Irapuato, Mexico, [2] Max Planck Institute for Biology Tübingen, Tübingen, Germany

Brown algae (Phaeophyceae) constitute the third most complex group of multicellular eukaryotes on the planet. This group separated over a billion years ago from the lineages that gave rise to plants and animals, evolving multicellularity independently from a unicellular ancestor ~450 million years ago. Furthermore, brown algae have heteromorphic life cycles, with morphologically distinct haploid and diploid phases, making them an independent “evolutionary experiment” to study the origin of biological complexity. We use Ectocarpus as a model brown alga to explore the structural and functional origins of proteins linked to important evolutionary innovations. We found that the origin of multicellularity and heteromorphic life cycles are linked to the emergence of multiple families of taxonomically restricted genes (TRGs). Many of these TRGs encode transmembrane proteins or lack known structural domains, suggesting a de novo origin from previously non-genic sequences, considered a key mechanism for genetic innovation. We are currently using gene coexpression network analyses to assess how these TRGs were integrated into pre-existing gene regulatory networks and infer their putative function through their association with functionally annotated genes. Based on the putative function of the gene network and the centrality of the TRGs, the most promising candidates will be targeted for CRISPR-based genome editing to evaluate the phenotypic effect of knocked-out strains and the link of these TRGs to the evolution of multicellularity. This work aims to expand our biological understanding of these ecologically relevant organisms, how their multicellularity evolved, and how TRGs become functional.

[A026] Paralog-specific patterns of predicted pathogenicity inform functional divergence

Angela Dawson[1], Raquel Meyer Nunes[1], Valentina Perez[1,2,3], Dr. Jessica Siltberg-Liberles[1,2]
[1]Department of Biological Sciences, Florida International University, Miami, USA, [2]Biomolecular Sciences Institute, Florida International University, Miami, USA, [3]Center for Genome Research, Florida International University, Miami, USA

Gene duplication is a major force of evolutionary change that allows paralogs to diverge from the ancestral gene and evolve novel functions under relaxed selective pressures. Detecting functional changes from sequence alone is challenging. However, as paralogs diversify to obtain different biological roles, they may develop distinct patterns in their tolerance to missense mutations that reflect changing functional constraints. Comparing such patterns can provide insight into their functional divergence. Missense variant predictors that integrate protein sequence and structure offer a way of assessing mutational tolerance across proteins. This study applies AlphaMissense, a deep learning predictor that predicts missense variant effects through the integration of protein sequence and structural context, to elucidate how patterns of predicted pathogenicity correspond to the evolutionary trajectories of paralogs following duplication events. Focusing on protein families with multiple human paralogs with known functions, we analyzed evolutionary rates, AlphaFold2 confidence, and secondary structure to investigate how conservation and structural properties relate and compare the patterns of these predictions in an evolutionary context. Our preliminary results indicate that pathogenicity scores tend to be highest in conserved residues and their surrounding linkers. Moreover, there are paralog, domain, and function-specific patterns in AlphaMissense pathogenicity scores that show statistically significant differences. We will also examine how structural properties relate to pathogenicity and functional change.

[A027] Building a Multipartite Bacterial Genome Database

Angelica Mariana Jara Servin[1], Alberto Domingo[1,2], Macarena Toll-Riera[1]
[1]Institut de Biologia Evolutiva, CSIC, Barcelona, Spain, [2]Universitat Pompeu Fabra, Barcelona, Spain

Multipartite genomes—defined by the distribution of essential and adaptive functions across a primary chromosome and secondary replicons such as chromids, megaplasmids, and plasmids—are widespread among bacteria. Current estimates indicate that approximately 10% of sequenced bacterial genomes exhibit a multipartite architecture (Harrison et al., 2010). Previous studies show that genes involved in environmental adaptation, pathogenicity, defense mechanisms, and host interactions are frequently located on secondary replicons and, in some cases, are enriched there (Kamp et al., 2013; López-Leal et al., 2014; Liu et al., 2025). Despite this growing body of evidence, the evolutionary forces shaping multipartite genome organization remain poorly resolved, and their ecological distribution across natural environments has not been systematically characterized.

In this work, we reassemble and analyze publicly available bacterial genomic data from ENA and NCBI (SRA), focusing on three major data types: (1) long-read and hybrid long- and short-read genome datasets, (2) shotgun metagenomes derived from both host-associated and non-host-associated environments, and (3) complete bacterial genomes. These datasets are processed using a unified analytical framework to identify and characterize multipartite genome architectures across diverse taxa and environments. Based on these analyses, we curate a comprehensive database that integrates isolate-derived multipartite genomes with environmental profiles of replicon presence and relative abundance.

This integrative approach enables a systematic examination of multipartite genome organization, providing a consistent resource for comparative and ecological analyses of bacterial genome architecture.

[A028] Implication of dioxygen reductase diversity on the growth physiology of Escherichia coli

Anjali Vijay Patil[1], Mr. Akshay M. Shirsath[1], Amitesh Anand[1]
[1]Department of Biological Sciences, Tata Institute of Fundamental Research-Mumbai, Mumbai, India

The rise of oxygen on early Earth prompted molecular adaptations to counter oxygen toxicity and exploit its high reduction potential for energy production. Dioxygen reductases (O_2REDs) were central to this transition. These enzymes reduce oxygen to water and enable aerobic respiration. O_2REDs fall into two major families: heme-copper oxidases and cytochrome bd oxidases. Many bacteria encode members of both families, suggesting functional tuning to fluctuating environments.

Escherichia coli, a facultative anaerobe, encodes one heme-copper oxidase (cytochrome bo_3) and two cytochrome bd oxidases (bd-I and bd-II). Cytochrome bo_3 predominates under aerobic conditions, whereas bd oxidases are typically associated with microaerobic growth. However, additional roles of bd oxidases in redox homeostasis, pathogenicity, and antimicrobial resistance raises a functional paradox: whether these enzymes are physiologically relevant outside the conditions in which they are primarily expressed.

To address this, we engineered E. coli strains lacking either cytochrome bo_3 or both bd oxidases and characterized their physiology under aerobic and hypoxic conditions. Under aerobic conditions, deletion of cytochrome bo_3 did not cause an apparent growth defect, whereas loss of both bd oxidases resulted in severe growth impairment, revealing its unappreciated role in aerobic physiology. Adaptive evolution restored the growth of bd oxidase deficient strain primarily via dysregulation of the ArcAB two-component system. But this compensation came at the cost of impaired stress response pathways, exposing trade-offs in regulatory plasticity.

Ongoing work is examining how O_2RED composition shapes evolutionary trajectories under hypoxic conditions, where respiratory diversity becomes critical.

[A029] Assessing the potential of evolution-informed conservation in Danish butterfly populations

Anna Maria Langmueller[1,2], Mrs. Nathalie Puggaard Elkær Andersen Ibsen[3], Jeppe Bayer Pedersen[3], Line Brun-Witt[3], Dr. Jesper Bechsgaard[3], Trine Bilde[3]
[1]Aarhus Institute of Advanced Studies, Aarhus, Denmark, [2]Center for Quantitative Genetics and Genomics, Aarhus, Denmark, [3]Center for Ecological Genetics, Department of Biology – Genetics, Ecology & Evolution, Aarhus, Denmark

Habitat loss and fragmentation cause declines in insect populations worldwide. Small and isolated populations are at risk of entering a self-reinforcing cycle of decline driven by inbreeding, genetic load, and environmental stress: the so-called extinction vortex. Butterflies can be vulnerable, due to limited dispersal ability and larval host plants specialization, making them sensitive to changes in habitat amount and connectivity. This raises an important question: can targeted habitat restoration and increased connectivity slow or even reverse genomic decline in fragmented butterfly populations?

To address this question, we use spatially explicit, individual-based simulation models implemented in SLiM that can jointly model demographic and genetic processes across dynamic landscapes. These models track dispersal, inbreeding, and the accumulation of deleterious mutations, which allows us to assess how genetic load interacts with habitat structure over time. Additionally, we compare how differences in life-history traits such as dispersal ability and host-plant requirement affect the potential for population recovery under various conservation management scenarios.

The simulations are informed by a unique long-term dataset of whole-genome sequences from a dozen different Danish butterfly species, sampled repeatedly between 1910 and 2023. These data provide empirical estimates of effective population size, genetic diversity, and inbreeding through time. By comparing simulated and observed genomic trajectories, we can assess whether our evolution-informed models can capture real-world population histories and help identify conditions under which habitat restoration is likely to support sustainable population recovery.

[A030] Will biofilm mutants of different Pseudomonas evolve in similar ways in similar environments?

Aparna Biswas[1], Jennifer Pentz[1], Dr. Peter Lind[1]
[1]Umeå University, Umeå, Sweden

Experimental evolution under static conditions with Pseudomonas fluorescens SBW25 repeatedly results in mutants with several distinct colony phenotypes that have increased ability to colonize the air-liquid interface by forming biofilms. The phenotypic and genetic bases of these adaptive mutants are well characterized, which allows evolutionary forecasts to be extended to related Pseudomonads and tested. The opportunistic pathogen Pseudomonas aeruginosa is of particular interest because it commonly evolves mutants with increased biofilm formation during infections, complicating treatment.

To test which evolution could be predicted, we evolved four diverse P. aeruginosa strains under static conditions and isolated mutants with divergent colony morphologies. As predicted, similar “wrinkly spreader” types as found on other species evolved repeatedly in three of the four strains, all with mutations disrupting negative regulation of c-di-GMP production, a signal for biofilm formation. The fourth strain, however, most frequently evolved “fuzzy spreader” types with mutations in a single gene involved in lipopolysaccharide synthesis. Surprisingly, for two strains, mutations were also repeatedly found in genes encoding chaperones with no known link to biofilm formation.

Despite the apparent limited phenotypic solutions to the selective problem, the genetic changes were diverse. While the same genes were repeatedly mutated in each strain, often with identical mutations, there were major differences across strains and other Pseudomonas species previously tested. This can be explained by differences in the relative fitness of mutants and in mutation rates to different phenotypes, often influenced by non-conserved mutational hotspots, limiting the ability to extend evolutionary forecasts across species.

[A031] Recovering genomes from museum blow flies (Diptera: Calliphoridae): a methodological comparison of low-input NGS library preparations.

Dr. Arianna Thomas-cabianca[1], Dr. Claudia Paetzold[1], Anna K. Hundsdoerfer[1]
[1]Senckenberg Natural History Collections Dresden, Dresden, Germany

Museum collections preserve invaluable genomic material from past populations, but DNA from historical specimens is typically fragmented, damaged, and present at low endogenous concentrations. Recent advances in next-generation sequencing (NGS) and historical DNA now enable genome-scale data recovery from such material, provided that laboratory and bioinformatic workflows are carefully optimized.

We present a reproducible methodological framework for recovering genome-wide data from dry-mounted historical insect specimens and a direct comparison of two low-input library-preparation strategies using commercial kits applied to the same individuals. Using non-destructive DNA extraction protocols, we processed ~century-old pinned specimens of Calliphora vicina and constructed sequencing libraries using (i) a double-stranded (DS) library preparation with enzymatic DNA repair and (ii) a single-stranded (SS), low-input approach. Both rely on standardized manufacturer protocols. All libraries were sequenced to the same amount of raw data (13.5 Gb per sample) and evaluated using a standardized pipeline that included quality control, duplication assessment, reference genome mapping, and mapping-based statistics.

Despite identical sequencing effort, the two library preparation approaches differed consistently. DS libraries produced higher effective DNA yield and more unique reads, but also higher duplication rates. At 3x coverage, breadth was higher for DS libraries in all but one sample, recovering ~10% more reference bases. At 10x coverage, SS libraries outperformed DS libraries in all but one sample, covering ~5% more bases.

These results show that differences reflect library preparation strategy rather than sequencing depth, highlighting trade-offs in maximizing usable genomic data. This framework provides practical guidance for applications of museum genomics studies.

[A032] A within-host model of the effective population of a malaria infection

Ariella Gladstein[1], Amy Goldberg[1]
[1]Department of Human Genetics, University Of California, Los Angeles, United States

Plasmodium parasites, the causative agent of malaria, undergo dramatic demographic variation, going from ~10–100 sporozoites during transmission to 10^8–10^{12} parasites at peak infection. This extreme lifecycle violates core assumptions of classical population genetic theory that is commonly used to model parasite evolutionary history and predict drug resistance. We present a theoretical framework for expected neutral genetic variation of a malaria parasite. We use a time-weighted harmonic mean to get life-stage specific effective population sizes (N_e) over four discrete stages of a parasite generation: vector-to-human transmission bottleneck, within-human expansion, human-to-vector transmission bottleneck, and within-vector sexual reproduction. We demonstrate that N_e is determined primarily by transmission bottlenecks, with per-host estimates of hundreds of parasites (N_e ≈ 100-1000) despite census sizes exceeding 10^{10}. Elasticity analysis reveals that the host-to-vector bottleneck has the strongest influence on N_e (ε = 0.58), while peak parasitemia has minimal impact (ε = 0.01). Under the model, we consider stage-specific selection efficacy under nearly neutral theory, suggesting that mutation expressed during blood stage will experience the strongest constraint. Our theoretical predictions are consistent with recent empirical data on differences in measures of genetic diversity at genes with strong stage-specific expression (Perkins et al. 2025, ProcB). Our framework also suggests that different N_e estimation methods may yield conflicting results: heterozygosity-based methods overweight expansion phases, coalescent methods are dominated by bottlenecks, and LD-based methods measure the sexual phase. The model can be extended to understand broad patterns of neutral and adaptive genetic variation in organisms with complex lifecycles.

[A033] Delineating unconventional respiro-fermentative metabolic routes for redox homeostasis

Arpita Biswas[1], Nikita Goel[1], Arjun Patel[2], Snehal Khairnar[1], Saideep Singh[1], Vivek Polshettiwar[1], Amitesh Anand[1]
[1]Tata Institute of Fundamental Research, Mumbai, , India, [2]Department of Bioengineering,University of California,San Diego, , United States of America
Electron transfer between molecules through oxidation-reduction (redox) reactions enables cells to capture, transfer, and utilize energy for various biological processes. Maintaining cellular redox homeostasis is therefore essential for life. The redox state of a cell is strongly determined by the balance between the oxidized and reduced forms of nicotinamide adenine dinucleotide (NAD), a universal cofactor present across all living systems. This balance is intricately maintained by NAD(P)-dependent enzymes, including NADH dehydrogenases, which regenerate NAD^+ to support redox homeostasis and catabolic metabolism. The absence of NADH dehydrogenases perturbs redox homeostasis and impairs bacterial growth, with consequent effects on pathogenicity in some species, thereby making these enzymes attractive targets for novel antimicrobial development. While the physiological effects of perturbing bacterial redox balance are well studied, the alternate compensatory mechanisms that maintain redox homeostasis in the absence of NADH dehydrogenases remain elusive. This study investigates the adaptive redox-balance strategies in NADH dehydrogenase-deficient Escherichia coli. The absence of these enzymes causes a significant growth defect in E. coli and induces fermentative lactate secretion to mitigate redox imbalance via substrate-level NAD^+ regeneration. Adaptive Laboratory Evolution uncovered specific respiro-fermentative metabolic reorganization, including a bypass respiratory chain, which enables growth restoration of the mutant strain. This reorganization provides fitness advantage to the strain via a significant aerotype shift, enabling rewiring of metabolic state from fermentative to respiratory. This study provides fundamental insight into alternative redox maintenance strategies, with potential implications for understanding antimicrobial resistance mechanisms of emerging antimicrobial therapies that target bacterial bioenergetics and redox-balancing enzymes.

[A034] Alterations in hypoxia-related gene expression and physiological responses between resurrected and inhabitant obligate parthenogenetic Daphnia clones from an Arctic lake

Athina Karapli Petritsopoulou[1], Dr. Dörthe Becker[2], Dagmar Frisch[1]
[1]Leibniz Institute of Freshwater Ecology And Inland Fisheries (IGB), Berlin, Germany, [2]NABU-Naturschutzstation Niederrhein, Kleve, Germany

Freshwater ecosystems in the Arctic, facing rapid environmental change, can serve as important model systems for studying the rapid adaptation potential of their inhabitants. Key members of these Arctic ecosystems often reproduce parthenogenetically, raising questions about their capacity to adapt over short time scales and what the possible mechanisms are. Here, we studied an obligate parthenogenetic Daphnia population from a meromictic lake in South-West Greenland using resurrected animals dated to ca. 2011 CE and modern descendants from 2022, forming two temporal subpopulations. The anoxic layer of the lake was previously inhabited by anoxygenic photoautotroph purple sulfur bacteria, a possible food resource for Daphnia. This microbial population, however, seems now to be destabilised, which could be reducing the presence of Daphnia in hypoxic or anoxic waters. In previous work, we found that the two Daphnia temporal subpopulations differ in their genetic structure and physiological traits linked to hypoxia and thermal tolerance. Daphnia clones form three closely related genetic clusters, with only one of them shared between subpopulations. To investigate their transcriptomic and physiological responses to hypoxia in more detail, we exposed two clones from each genetic cluster to medium and severe hypoxia. We performed RNA sequencing and quantified three physiological biomarkers (haemoglobin, glutathione-S-transferase activity and TBARS, a proxy for lipid peroxidation). Diverging gene expression patterns and associated physiological responses between genetic clusters and temporal subpopulations will be discussed in the light of rapid evolutionary change in response to ecosystem changes in a parthenogenetic population.

[A035] Investigating the pre- and post-colonial population structure of Khoe-San descendant populations

Dr. Austin Reynolds[1], Stacy Edington[2], Dr Dana Al-Hindi[3], Neus Font-Porterias[4], Alexandra Surowiec[4], William Palmer[4], Justin Myrick[4], Dr. Paul Norman[5], Dr Caitlin Uren[6], Marlo Moller[6], Dr Brenna Henn[4]
[1]University Of North Texas Health Science Center, Fort Worth, United States, [2]Baylor University, , , [3]University of California San Francisco, , , [4]University of California Davis, , , [5]University of Colorado School of Medicine, , , [6]South African Medical Research Council Centre for Tuberculosis Research, Division of Molecular Biology and Human Genetics, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa, Cape Town, South Africa

The genetic diversity of present-day southern African populations has been significantly shaped by European colonization and the Dutch East India slave trade. San and Khoekhoe populations from Botswana and Namibia retain exceptional linguistic, cultural and genetic diversity, but few Khoisan-speaking groups remain south of the Kalahari Desert. Historically though, far southern Africa was home to many San and Khoekhoe groups. Popular opinion often implies that such populations do not contribute to the ancestry of contemporary South Africans, however previous research has shown a gradient in Khoe-San ancestry, with individuals further from Cape Town having a higher proportion of Khoe-San ancestry. Here we advance this work by characterizing the genetic ancestry of 620 newly sampled individuals from across western South Africa to reconstruct pre-colonial and colonial population structure among these populations. These groups retain majority Khoe-San genetic ancestry (>48%), suggesting the persistence of Khoe-San ancestry to the present day. By isolating the Khoe-San ancestry component, we show that it is intermediate between the ≠Khomani San and Nama, and distinct from Kalahari Khoe-San populations. We also find that signatures of the Indian Ocean slave trade can be traced to Indonesian islands such as Sulawesi, Java, and Flores, while the South Asian ancestry is regionally non-specific. Finally, we constructed spatial models of identity-by-descent segment sharing among the study populations to uncover environmental variables influencing historic migration rates. We find mountain ranges as well as differences in summer/winter rainfall patterns to play a major role in patterns of gene flow in our study region.

[A036] SLiM 5: Expanding the horizons of forward genetic simulation to full genomes

Benjamin Haller[1], Peter Ralph[2], Philipp Messer[1]
[1]Cornell University, , United States, [2]University of Oregon – Eugene, , United States

SLiM, a free open-source software framework for forward genetic simulation, is widely used because of its power, flexibility, and speed. With over 1700 citations, it has become a key tool in population genetics, and is increasingly being used in evolutionary ecology, epidemiology, and conservation genetics. We will provide a brief overview of the major existing features of SLiM prior to version 5, including scriptability with Eidos, model visualization with SLiMgui, non-Wright–Fisher models, continuous-space models, tree-sequence recording, and multispecies modeling. We will then focus on the new features introduced in SLiM 5. The key innovation of version 5 is the ability to simulate multiple chromosomes, including chromosomes of different types (e.g., sex chromosomes, mitochondrial and chloroplast DNA), up to the scale of full genomes. We will illustrate these new capabilities by showing a full-genome model of Drosophila melanogaster.

[A037] UK Biobank data shows local adaptation at TLR1 in response to solar radiation

Chaw Thwe[1], Nicholas Fountain-Jones[1], Phillip Melton[1], Bennet McComish[1]
[1]University of Tasmania, Hobart, Australia

It is increasingly recognized that local adaption to environmental factors underlies differences in autoimmune disease prevalence across the globe. The prevalence of many autoimmune diseases increases with latitude, possibly because of vitamin D insufficiency. We hypothesise that availability of vitamin D exerts selective pressure on immune-system genes whose effects are modulated by the vitamin D receptor.

Using a landscape genomics approach, we conducted a genotype–environment analysis using data from the UK Biobank on a genome-wide scale, focusing on the association with solar radiation levels to detect genomic regions that show signals of selection influenced by UV exposure. Genotypes and associated metadata, including birthplace latitude and longitude were obtained from UK Biobank. Solar radiation levels were obtained from WorldClim. Logistic regression models were run genome-wide using Samβada for three samples of 20,000 White British individuals, followed by meta-analysis, to identify variants undergoing positive selection.

A genome-wide significant signal of local adaptation in response to solar radiation levels was detected at TLR1 on chromosome 4. The strongest signal of selection, with p = 1.78×10^-18, is at rs5743618, a missense SNP in TLR1 that has previously been shown to be under selection in European populations.

TLR1 is known to be involved in regulatory interactions with the vitamin D receptor, providing a likely basis for selection driven by low UV levels.

[A038] A systematic benchmark reveals DNA polymerase combinations for high-quality genome assembly

Bernhard Bein[1,2], Charlotte Gerheim[1], Nicola Gscheidel[3,4], Lucas Canesin[1], Alexander Ben Hamadou[1], Clement Schneider[5], Tilman Schell[1], Xueling Yi[1,2], Carles Galia-Camps[6], Julio Hechavarria-Cueria[2,7,8], Sylke Winkler[3,4], Carola Greve[1], Michael Hiller[1,2]

[1]Senckenberg – Leibniz Institution for Biodiversity and Earth System Research, Frankfurt, Germany, [2]Institute of Cell Biology and Neuroscience, Faculty of Biosciences, Goethe University Frankfurt, Frankfurt, Germany, [3]Max Planck Institute of Molecular Cell Biology and Genetics, Dresden, Germany, [4]DRESDEN Concept Genome Center, Technische Universität Dresden, Dresden, Germany, [5]Senckenberg Research Institute, Goerlitz, Germany, [6]Departamento de Biodiversidad y Biología Evolutiva, CSIC, Museo Nacional de Ciencias Naturales (MNCN), Madrid, Spain, [7]Ernst Strüngmann Institute (ESI) for Neuroscience in Cooperation with Max Planck Society, Frankfurt, Germany, [8]Freie Universität Berlin, Berlin, Germany

PacBio High-Fidelity (HiFi) long-read sequencing has become one of the preferred methods to obtain reads that can be assembled into highly contiguous and complete genome assemblies. Still, many potential samples remain inaccessible to HiFi sequencing because of very small amounts of long DNA fragments available or secondary metabolites inhibiting sequencing performance. Examples include tissue biopsies and small-bodied organisms. We recently showed that amplifying DNA prior to library preparation by long-range Polymerase Chain Reaction (PCR) can unlock aforementioned samples for HiFi sequencing by increasing the number of sufficiently long DNA fragments in the final library. However, amplification can introduce systematic biases caused by uneven fragment sampling and GC-content related polymerase dropouts. So far, systematic tests of different polymerase enzymes on an array of samples are lacking. Here, we test 10 enzymes on three different organisms, a collembolan, a sea slug and a bat and assess polymerase bias by comparing resulting assembly completeness, contiguity and polymerase drop-outs in relation to genome-wide GC and non-B-DNA content. We find that single polymerases perform comparably across samples, indicating that each polymerase has inherent amplification biases. Furthermore, we explicitly characterize polymerase drop-outs on the nucleotide level and show that combining various polymerase read sets can increase assembly contiguity, emphasizing that polymerase-specific biases can thus be complemented. We expect that our insights will inform future approaches linking PCR-amplification and long-read sequencing for the assembly of high-quality reference genomes from limited material.

[A039] Comparative genomics of the weevil species of the genus Rhynchophorus in Southeast Asia

Bisha Shahid[1], Kripa Shrestha[1], Neelu Begum[1], Dr Mariana Lyra[1], Dr Gabriel Riano[1], Theo Busschau[1], Wahizatul Afzan Azmi[2], Purnama Hidayat[3], Nguyen Danh[4], Stephane Boissinot[1]
[1]The Evolutionary Genomics Lab, Center for Genomics and Systems Biology, New York University Abu Dhabi, Abu Dhabi, United Arab Emirates, [2]Research Management Office, University Malaysia Terengganu, Terengganu, Malaysia, [3]Department of Plant protection, Bogor Agricultural University, Bogor, Indonesia, [4]Department of Technology for Pharmaceutical Chemistry,Institute of Chemical Technology, Vietnam Academy of Science and Technology, Chi Minh City, Vietnam
Rhynchophorus ferrugineus, R. bilineatus, and R. vulneratus are closely related species of insects (Coleoptera). They are major palm pests and are morphologically similar yet they differ in diagnostic traits, particularly body coloration, and in geographic distribution across Southeast Asia and Melanesia. Taxonomic identification among these species has historically relied on morphological characters such as color patterning; however, these traits vary continuously across geographic gradients, with intermediate forms commonly observed, indicating that morphology alone is insufficient to reliably delimit species. This reliance on variable morphological traits has contributed to repeated species descriptions and synonymizations, leaving species boundaries unresolved. Here, we use whole-genome sequencing of individuals sampled across Thailand, Malaysia, Vietnam, and multiple islands of Indonesia to conduct a comparative analysis of population structure and genomic differentiation among species in their native ranges. We focus particularly on Indonesia, where distinct color morphs and nominal species occur across neighboring islands, providing a framework to assess how island biogeography, geographic isolation, and genomic divergence relate to species boundaries. By comparing patterns of connectivity and divergence within and between species, this study clarifies relationships among Rhynchophorus species and demonstrates the necessity of genomic approaches for resolving species limits in morphologically variable pest taxa.

[A040] Ancient shared polymorphism investigation on X and autosomes of 250 primates

Bjarke Pedersen[1], Vasili Pankratov, Juraj Bergman, Mikkel Heide Schierup
[1]Birc Aarhus University, , Denmark

Previous investigations into ancient shared polymorphisms (ASP), single nucleotide variation shared between distinct species, have mainly targeted ape lineages and potential sites under strong balancing selection in humans. Primate diversity data provides a rich resource to examine ASP across multiple species and understand fundamental evolutionary mechanisms throughout the primate phylogeny. Coalescent theory predicts the number of ASP based on divergence time, sample sizes, and population sizes. However, directional selection, balancing selection, population structure and ancient admixture also shape ASP patterns. Examining genomic distributions of ASP will reveal how common these processes are among primates.

Additionally, since the X chromosome is disproportionally important for reproductive barriers it might have less ASPs than expected from each relative effective population size due to less impact by introgression. This predicts significantly fewer ASP on the X relative to autosomes.

To investigate whether selection and ancient admixture is common across the primate phylogeny, we intersected SNPs from the autosomes and X chromosomes of ~1500 samples from 259 species. By stratifying the SNPs into mutational classes and CpG/Non-CpG sites we get the number of ASPs for the entire primate phylogeny. We find that the number of ASP are clustered along the genome unless species are very divergent. There are far fewer ASPs on the the X than on the autosomes, indicating that the X chromosome is less affected by ancient admixture

[A041] CAAPA2: A diverse reference panel for improved genotype imputation in underrepresented populations

Caitlin Uren[1,2,3,4], Dayna Croock[1], Victor Borda[5], Simon Gravel[6], Brenna M. Henn[7], Consortium on behalf of all the members of the CAAPA2

[1]South African Medical Research Council (SAMRC) Centre for Tuberculosis Research (CTR), Division of Molecular Biology and Human Genetics (MBHG), Department of Biomedical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University , Cape Town, South Africa, [2]Centre for Bioinformatics and Computational Biology, Stellenbosch University, Cape Town, South Africa, [3]Genomics for Health in Africa (GHA), Africa-Europe Cluster of Research Excellence (CoRE), , South Africa, [4]National Institute for Theoretical and Computational Sciences (NITheCS), , South Africa, [5]University of Maryland School of Medicine, Baltimore, USA, [6]Department of Human Genetics, McGill University, Montreal, Canada, McGill University and Genome Quebec Innovation Centre, Montreal, Canada, [7]Department of Anthropology, UC Davis Genome Centre and Center for Population Biology, University of California Davis, Davis, USA

Genotype imputation is a critical tool in genomic research, yet its accuracy depends heavily on the diversity and representativeness of reference panels. Existing widely-used panels such as the 1000 Genomes Project (1000G), African Genome Resources (AGR), and TOPMed have made significant contributions but remain limited in their representation of global populations, particularly those of African ancestry. The Consortium on Asthma in African Ancestry Populations in the Americas (CAAPA) has developed CAAPA2, comprising approximately 1,000 new high-coverage whole genome sequences spanning major regions of sub-Saharan Africa, analysed in conjunction with approximately 1,100 individuals of African and Indigenous descent from the United States, Caribbean and South America. Here, we evaluate the performance of CAAPA2 as a reference panel for genotype imputation compared to commonly used resources. Genotyping array data from multiple African and American populations was imputed and accuracy was assessed by comparing imputed genotypes to whole genome sequencing data from the same individuals. In addition, imputation quality scores were evaluated to investigate overall performance. Our results demonstrate that CAAPA2 substantially improves imputation performance and accuracy in underrepresented populations compared to existing reference panels. This improved performance highlights the critical importance of diverse, representative reference resources for genomic research. The CAAPA2 reference panel provides a valuable new resource that enhances the power and precision of genetic studies in historically underrepresented populations.

[A042] Microenvironment-regulated plasticity and genetic selection drive a two-stage evolutionary trajectory in cancer

Can Liu[1,3], Dr. Xuemei Lu[1,2,3]
[1]Key Laboratory of Genetic Evolution & Animal Models, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming, China, [2]Yunnan Key Laboratory of Biodiversity Information, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming, China, [3]College of Life Sciences, University of Chinese Academy of Sciences, Beijing, China

Understanding how phenotypic plasticity and genetic selection jointly shape evolutionary trajectories remains a central challenge in evolutionary biology. While cancer provides a tractable system to observe rapid evolution, the interactive contribution of genetics and plasticity remains elusive. By re-analyzing multiple public datasets, we uncovered a two-stage cancer evolution pattern driven by immune-regulated plasticity and clonal sweep.

Specifically, we found that early tumor growth is characterized by high plasticity and weak heritability, with cancer cells occupying diverse, hybrid transcriptional states. These highly plastic cells are consistently enriched for immune-related programs, such as antigen processing, and cytokine communication, suggesting strong regulation by the tumor microenvironment (TME). In contrast, later stages of tumor evolution show increased heritability, reduced plasticity, and clonal or subclonal sweeps, frequently associated with epithelial–mesenchymal transition, and metabolic activity. Model simulations and spatial co-localization analysis further validated this pattern, with highly plastic cells dominating at early stage and co-localizing with immune or stromal cells.

Strikingly, the dominance of plasticity versus genetic selection depends on ecological context. In immune-infiltrated tumors (e.g., LUAD, TNBC), plasticity predominates early and transitions predictably toward heritable states with progression and metastasis. In immune-desert tumors or in vitro systems, this transition is attenuated or absent, and alternative fitness strategies emerge.

Together, these findings support a general evolutionary model in which TME-regulated phenotypic plasticity enables early adaptive exploration, followed by genetic canalization and clonal expansion at later stages. This eco-evolutionary transition provides a unifying framework for context-dependent cancer evolution.

[A043] Sickle-Cell Disease as a condition of African genetic origin but increasingly embedded within non-African genomes

Carlos Passos[1], Dr. Kelly Nunes[1], Dr. Carla Dinardo[2], Professor Ester Sabino[1], Dr. Daniela Werneck[3], Dr. Dahra Teles[4], Dr. Luiz Amorim[5], Professor Brian Custer[6,7], Dr. Grier Page[8], Dr. Shannon Kelly[9], Professor Diogo Meyer[1]

[1]Universidade De São Paulo, São Paulo, Brazil, [2]Hemocentro de São Paulo, São Paulo, Brazil, [3]Fundação Hemominas, , Brazil, [4]Fundação de Hematologia e Hemoterapia de Pernambuco, Recife, Brazil, [5]Instituto de Hematologia Arthur de Siqueira Cavalcanti, Rio de Janeiro, Brazil, [6]Vitalant Research Institute, San Francisco, United States, [7]University of California San Francisco, San Francisco, United States, [8]RTI International Research, Durham, United States, [9] University of California San Francisco Benioff Children's Hospital Oakland, Oakland, United States

Sickle-cell disease (SCD) comprises a group of inherited blood disorders caused by mutations in the HBB gene on chromosome 11. Despite being one of the most common monogenic disorders worldwide, SCD remains among the most neglected in healthcare, highlighting the need for improved early diagnosis, equitable access to care, and strategies to address socioeconomic disparities and stigma. In populations where admixture involving different continental origins started ~20 generations ago, the genetic landscape of SCD is particularly complex, as admixture and recombination between ancestries lead to a progressive decoupling between SCD-associated mutations and their original African genetic background. We investigated this decoupling process using the REDS-III cohort, which includes 2746 Brazilian individuals diagnosed with SCD. We found that SCD patients in Brazil comprise a wide range of continental ancestries, with 53.5% of individuals having genomes with greater European than African ancestry, a strong indicator of how extensive decoupling has progressed. This is in striking contrast to SCD in the USA, where only 3.9% of patients have greater European than African ancestry in three cohorts (OMG, PharmHU and PUSH). For the REDS-III, the correlation between African allele dosage at the HBB locus and the remainder of chromosome 11 gradually declines with increasing distance, reaching a plateau of approximately 10%. This suggests decoupling has been substantial, but remains incomplete. However, despite the substantial decoupling, simulation tests show that this correlation would reach zero in ~10 generations, suggesting that ongoing decoupling may have been slowed by processes such as assortative mating.

[A044] A fine-grained assessment of migration into Ireland from the Early Bronze Age onwards

Catherine Butt[1], Dr Valeria Mattiangeli[1], Dr Daniel Bradley[1], Dr. Lara Cassidy[1]
[1]Trinity College Dublin, Dublin , Ireland

The end of the Neolithic in western Europe was marked by major demographic change and the introduction of Indo-European languages. In the Rhine region, the emergence of Bell Beaker culture is associated with a large influx of steppe-related ancestry, likely derived from Corded Ware populations further east. The spread of Bell Beaker culture into Britain is estimated to have led to a long-term 90% population replacement, with further migratory influxes to the island identified across the Bronze and Iron Age, providing candidate windows for Celtic language entry. However, little is known of how the neighbouring island of Ireland absorbed steppe-related ancestry and Indo-European languages through time, with only three Bronze Age genomes from the northeast published thus far. Here we present over 50 new shotgun sequenced genomes from the Early Bronze Age onwards. Through haplotype-based admixture modelling, we characterise ancestry contributions from Britain and the continent through time, as well as the earlier Neolithic population. This reveals Ireland to have its own unique trajectory after the initiation of the Bronze Age, including long-term survival of local Neolithic ancestry. Using IBD segments, we further uncover fine-grained mobility networks between archaeological sites in Ireland, Britain and the continent.

[A045] Adzuki bean: the genetic basis of domestication and high-latitude cultivation

Chih-Cheng Chien[2], Kai Chun Lee[1], Ken Naito[2], Cheng-Ruei Lee[1]
[1]National Taiwan University, Taipei, Taiwan, [2]National Agriculture and Food Research Organization, Tsukuba, Japan

The adzuki bean is an important legume in East Asian culinary culture. While archaeological traces have been observed in both Japan and China, previous genetic works identified the highest cultivar genetic variation in central China. Using hundreds of wild and cultivar accessions across Asia, we identified unequivocal evidence of Japanese domestication origin. The cultivars had a range expansion into China, followed by a wild-cultivar hybridization in central China, creating the apparent patterns of high cultivar variation in central China. While high-latitude regions such as Hokkaido of Japan and northeastern China are not the native range of this species, currently, these regions are the largest adzuki production zone in these countries. Genetic mapping was performed for flowering time, and we showed that mutations enabling the modern high-latitude cultivation of this crop were ancient variations from the wild progenitor populations.

[A046] Human demography shapes patterns of language diversity across the world

Chiara Barbieri[1,2], Anna Graff[2], Balthasar Bickel[2], Kentaro K. Shimizu[2]
[1]University of Cagliari, Cagliari, Italy, [2]University of Zurich, Zurich, Switzerland

Genes and languages both evolve through transmission, divergence, and contact, but the extent to which their paths align remains unresolved. While both systems accumulate variation through descent with modification, languages are additionally shaped by horizontal transfer and sociocultural boundaries. Local case studies described tight gene-language associations as well as striking mismatches, yet a global, systematic assessment has been lacking. We address this gap using GeLaTo (Genes and Languages Together), a curated database linking genome-wide data to linguistic affiliation for over 500 human populations. Leveraging measures of genetic distance, admixture, and excess homozygosity, we ask: do genetically related populations speak related languages (belonging to the same language families)? Are language isolates associated with genetic isolation? How do demographic processes such as admixture and isolation shape distributions of structural properties of languages?

We find cross continental patterns of genetic cohesiveness behind large language families, with relevant exceptions showing the power of cultural dynamics alone in spreading languages through language shifts. Comparisons between expansive language families and isolates reveal a tendency for genetically isolated populations to speak isolated languages. Integrating quantitative linguistic databases, we further show that genetic contact via admixture is predominantly associated with structural convergence, whereas genetic isolation correlates with increased linguistic diversification. Our work suggests that demography constitutes a neutral baseline model driving dynamics of language and cultural diversification. By studying deviations from this model we can gain insights into non-neutral processes in language evolution based on selection and adaptation.

[A047] Highly repeatable experimental evolution of drug resistance in yeast

Chi-fa Huang[1], Dr. Chih-Yao Chang[1], Dr. Mei-Yeh Lu[1], Dr. Ya-Wen Chang[1], Wen-Hsiung Li[2]
[1]Academia Sinica, Taipei, Taiwan, [2]University of Chicago, Chicago, USA

In nature the chance for an evolutionary event to repeat is very small, mainly because the living environment of an organism changes with time. Thus, a well-known question is “what chance does evolution repeat itself, if the living environment is tightly controlled?” This question is still not well explored because controlling living environments is challenging. To address this question, we studied the evolution of Saccharomyces cerevisiae’s resistance to fluconazole, a very well-studied drug in yeast. At T=0,131 identical populations were subjected to 16 μg/ml fluconazole. Media transfer was then conducted every 48 hours. The concentration of fluconazole was raised to 32 μg/ml at T=16 cycles and to 48 μg/ml at T=48 cycles (equivalent to ~270 generations). Samples from T=15, 30 and 45 were subjected to whole genome sequencing to determine the mutations and their frequencies in each population at T=15, 30 and 45. In total, 20 (single-amino-acid) mutations reached the frequency of f =1, 17 reached f =0.9 and 29 reached 0.5≤ f<0.9. Among these, 18 mutations with f =1, 15 with f = 0.9 and 27 with 0.5≤ f<0.9 occurred in PDR5 (Pleiotropic Drug Response 5). In particular, 1 PDR5 mutation had f =1 in 11, f =0.9 in 8 and 0.5≤ f<0.9 in 7 populations. Moreover, there were 7 other repeated mutations with f ≥0.9. These findings indicate that the experimental evolution of fluconazole resistance in yeast is highly repeatable, driven primarily by recurrent mutations in a key multidrug transporter gene.

[A048] Population genomics of Hyaloperonospora arabidopsidis, a natural Arabidopsis thaliana pathogen

Ching-Yi Huang[1], Miriam Lucke[1], Detlef Weigel[1], Rebecca Schwab[1]
[1]Max Planck Institute For Biology Tübingen, Tübingen, Germany

Hyaloperonospora arabidopsidis (Hpa)–Arabidopsis thaliana (Ath) is a well-established model system for studying plant–microbe interactions and has contributed significantly to our understanding of the arms races between plant immune receptors and pathogen virulence factors, known as effectors. However, most existing studies are based on a small set of well-characterized Hpa isolates, primarily from the UK and the Netherlands. This limited sampling raises the question of whether these data capture the full picture of Hpa genetic variation and the consequent Ath-Hpa interaction. Here, we sequenced 93 Hpa isolates collected across Europe to obtain a broader and deeper view of Hpa diversity in the wild. Phylogenetic analysis of these isolates reveals a clear north–south structure, suggesting postglacial expansion likely along with host migration. By defining populations based on population structure analyses, we observe copy number variation and allelic diversity of effectors among populations, highlighting the local divergence of virulence factors within these Hpa isolates. Overall, our dataset provides a new perspective on Hpa population genomics in Europe, showcases Hpa lineages that differ from well-characterized isolates, and establishes a structured foundation for future functional studies.

[A049] RNA warfare in asexual animals: pathogen-induced ribosome damage signatures correlate with upregulation of horizontally acquired RNA ligases in bdelloid rotifers

Chris Wilson[1], Tim Barraclough[1], Reuben Nowell
[1]University Of Oxford, , United Kingdom

Coevolutionary conflict can favour genetic exchange, from the spread of toxin and resistance genes among microbes to the maintenance of sex in eukaryotes. Bdelloid rotifers are an extreme case: despite lacking males, these microscopic animals have acquired thousands of genes by horizontal transfer from other kingdoms of life. We recently showed that bdelloids attacked by a natural fungal pathogen deploy horizontally acquired biosynthesis genes predicted to make antifungal compounds. Here, we explore a second signal from the same system that points to defence against RNA-targeting virulence factors. Across two bdelloid species challenged with a fungal pathogen, rotifer genes annotated with RNA ligase activity were disproportionately and rapidly upregulated, ranking among the strongest transcriptional responses. This enrichment was specific to pathogen exposure and absent under abiotic stress. Independently, analysis of host ribosomal RNA revealed a pathogen-associated signal of noncanonical base insertion or modification exclusive to the sarcin-ricin loop, a canonical cleavage site for fungal ribotoxins. The magnitude of this signal correlated with host resistance. RNA ligases and repair systems are widely disseminated by lateral transfer among microbes engaged in ribotoxin-mediated conflict. By analogy, bdelloids may have acquired and retained similar machinery to counter ribosome-directed attacks by their own pathogens. This presentation explores the potential link between the transcriptomic and rRNA evidence and invites discussion on how RNA damage, repair and defence intersect in host-pathogen interactions shaped by horizontal gene transfer."

[A050] A deep learning approach to detecting negative frequency-dependent selection

Cindy Santander[1], Andre Luiz Campelo dos Santos[1], Sandipan Paul Arnab[1], Matteo Fumagalli[2,3], Mike DeGiorgio[1]
[1]Department of Electrical Engineering and Computer Science,Florida Atlantic University, Boca Raton, United States, [2]School of Biological and Behavioural Sciences, Queen Mary University of London, London, United Kingdom, [3]The Alan Turing Institute, London, United Kingdom
Balancing selection is a mode of natural selection that maintains genetic diversity through various mechanisms, including negative frequency-dependent selection (NFDS). However, distinguishing the genomic signature of NFDS from those of other balancing selection modes, such as overdominance, remains a significant challenge. Here we outline strategies to improve the modeling of genomic patterns expected under NFDS, with the goal of better differentiating them from signals of neutrality and alternative selection processes. We demonstrate how resource-efficient deep transfer learning, combined with novel data preprocessing and the modeling of genomic autocovariation, can effectively detect and characterize NFDS using either phased or unphased genotypes, and with or without temporal data from ancient DNA. Finally, we offer practical recommendations for both empiricists and method developers on advancing the detection of NFDS in genomic data.

[A051] Genetic insights into convergent acquisitions of placental phenotypes and menstruation in eutherians

Claire Lavergne[1,2], Eulalie Liorzou[1,2], Camille Berthelot[1]
[1]Institut Pasteur, Université Paris Cité, CNRS UMR 3525, INSERM UA12, Comparative Functional Genomics G5 group, Paris, France, [2]Université Paris Cité, BioSPC, Paris, France

Uterine morphology and function have changed rapidly during mammalian evolution, with many traits appearing convergently, such as menstruation, placentation morphology, trophoblast implantation depth, and maternal-fetal interdigitation. The genetic architecture of these convergent acquisitions has not yet been extensively researched. Here, we propose to study the molecular evolution of over 17,000 one-to-one orthologous genes across 117 eutherians to identify genes involved in phenotypic variations in uterine development, pregnancy, and menstruation. Using RERconverge and HyPhy, we investigate genes whose relative evolutionary rates significantly differ in species which have convergently acquired a trait of interest compared to other species. We further characterize which molecular and cellular pathways are likely implicated in these uterine phenotypes using functional enrichment analyses and uterine transcriptomic data across different mammalian species, either publicly available or generated by our lab. In particular, we are interested in uncovering genes and pathways which have contributed to the acquisition of menstruation, a rare trait present in less than 2% of eutherians and which has evolved four independent times. Ultimately, this study will contribute to our understanding of the uterus and reproductive functions across eutherian evolution, by providing novel candidate genes and pathways to investigate.

[A052] Inferring demographic histories and non-random mating dynamics in complex admixture processes related to the Trans-Atlantic Slave Trade with MetHis2.0

Coailinn Egan[1], Dr Romain Laurent[1], Paul Verdu[1]
[1]Eco-Anthropology, MNHN, Paris, France

Admixture -defined as a gene flow between populations that were previously reproductively isolated- is a core mechanism of genetic evolution. Admixture events can be embedded in complex demographic processes, including migrations from several source populations, over multigenerational periods of time, potentially involving non-random mating dynamics.
In the context of the Atlantic Slave Trade in the XVth to XIXth centuries, numerous human populations underwent socio-demographically complex processes of admixture. Forced and voluntary mass migrations led to gene flows between populations from the African, European and American continents. Furthermore, legal and cultural norms regarding socio-economic status, perceived race, language, and place of residence strongly influenced mate choice, possibly resulting in non-random mating dynamics. Non-random mating patterns may also have been spatially and temporally heterogeneous depending on geo-political contexts.
Altogether, understanding the intricacies of human populations' genomic evolution during this historical period is extremely challenging.
We developed MetHis2.0, a bio-informatics pipeline relying on SLiM, allowing the forward-in-time simulation of autosomal, sex-specific and mitochondrial genomic data under complex admixture scenarios, including up to four source populations with parameterizable allele frequencies and migration dynamics with parameterizable timing and intensity. MetHis2.0 also comprises mechanisms of non-random mating based on biological and/or socio-cultural factors, that can be transmitted maternally, paternally or biparentally. It operates within an Approximate Bayesian Computation framework, to formally reconstruct the evolutionary history underlying extant genomic patterns.
We deployed the MetHis2.0-ABC framework to reconstruct complex demographic and socio-cultural dynamics during admixture processes in several extant human populations related to the Trans-Atlantic Slave Trade.

[A053] Recovering Nuclear Genomes from Extinct Xenarthran Megafauna to Reassess Evolution, Ecology, and Physiology

Cole Nickason[1,2], Melanie Kuch[1,3], Dr Frédéric Delsuc[4], Dr. Hendrik Poinar[1,2,3,5]
[1]McMaster Ancient DNA Centre, Hamilton, Canada, [2]McMaster Department of Biochemistry & Biomedical Science, Hamilton, Canada, [3]McMaster Department of Anthropology, Hamilton, Canada, [4]Institut des Sciences de l'Evolution de Montpellier, Université de Montpellier, Montpellier, France, [5] Michael G. DeGroote Institute for Infectious Disease Research, Hamilton, Canada

Extant xenarthrans, sloths, armadillos and anteaters, represent only a small amount of the group's diversity, with a plethora of now extinct megafaunal varieties having thrived prior to the Holocene. Ground sloths, glyptodonts and pampatheriids, were once widespread across the Americas, occupying a range of ecological niches, making them keystone species in Pleistocene ecosystems, and a valuable model for anthropogenic extinction. Despite remaining as iconic fixtures of ice age ecology, a great deal regarding the phenotype and evolution of organisms like Megatherium and Glyptodon, remains contentious. Previous work investigating these questions focused on mitochondrial genomes, as well as more traditional morphological analyses. To better answer phylogenetic questions and gain a more holistic understanding of these organisms and their population dynamics, we aim to reconstruct the nuclear genomes for a diverse set of species. This will include the profiling of a large collection of bones, teeth and other material for endogenous aDNA with shotgun sequencing and targeted enrichment. Additionally, paleofeces will provide an additional reservoir for both endogenous xenarthran genomic material, as well as metagenomic aDNA to answer long standing dietary questions regarding ground sloth digestion. Modern references genomes will allow for genome assembly and inform bait design to enhance capture enrichment of target aDNA molecules. Assembled genomes will enable a comparison to previous phylogenies and allow characterization of markers for key phenotypes of interest, such as enamel production or hair growth. This work also demonstrates how aDNA techniques have progressed, given the often-poor preserving tropical conditions for many species of xenarthrans.

[A054] Rapid genomic and phenotypic change in an asexual Daphnia population

Dagmar Frisch[1], Athina Karapli-Petritsopoulou, Prof. Dr. John Anderson, Dr. Dörthe Becker
[1]Leibniz Institute of Freshwater Ecology and Inland Fisheries (IGB), Berlin, Germany

Obligate parthenogenesis is a widespread phenomenon in extreme environments both in high latitudes and altitudes. This has also been observed in the keystone zooplankton grazer Daphnia, particularly of species belonging to the pulex/pulicaria species complex which have a circumpolar distribution. Arctic lakes are generally nutrient-poor and have low productivity, but in some meromictic lakes, dense photoautotroph bacterial populations in the anoxic zone of the lake can be an important food source for Daphnia. Associated with the recent shift in climatic conditions in the Arctic, this food source may now have diminished considerably, thus relaxing selection pressure on hypoxia tolerance that would have allowed Daphnia to feed in hypoxic or anoxic conditions.
For a deeper understanding of their capacity for rapid adaptation to such changes, we studied members of an obligately parthenogenetic Daphnia population resident in a lake in Southwest Greenland at two time points separated by a decade. We compared genotypic and phenotypic properties of the historical population represented by resurrected lineages dating to 2012 CE, to a those of a modern lake population sampled in 2022 CE. We applied whole genome sequencing to study genomic differences at the temporal subpopulation level and combined this data with physiological trait measurements including respiration rate, critical oxygen limit and thermal tolerance. We will discuss differences in functional traits and their potential genomic underpinnings, providing evidence for rapid phenotypic change with potential adaptive value.

[A055] De novo Transcriptome Assembly of the Coral-associated Gastropod Phenacovolva rosea (Mollusca: Gastropoda: Ovulidae)

Damin Lee[1], Yukyung Kim[1], Jin-Ho Park[1]
[1]Jeonbuk National University, Jeonju, South Korea

Ovulid gastropods, commonly known as allied cowries, are marine mollusks renowned for their vivid coloration and elaborate mantle patterns. They exhibit striking morphological and color mimicry that closely resembles their host corals, representing an exceptional example of host-associated adaptive evolution. Phenacovolva rosea, a coral-associated ovulid gastropod, displays pronounced intraspecific variation in both shell morphology—ranging from narrow and elongated to short and stout forms—and coloration, including red, yellow, and white phenotypes. Such extensive phenotypic diversity within a single species may reflect a strong role of ecological selection driven by host coral specialization and provides an ideal system for investigating the genomic basis of adaptive diversification. Here, we present the de novo transcriptome assembly of P. rosea. This transcriptome provides foundational resources for elucidating the genetic mechanisms underlying intraspecific variation, mimicry, and host-associated adaptation in coral-associated gastropods.

Keywords: Ovulid, symbiosis, intraspecific variation, mimicry, adaptive evolution

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[A056] Phylogenomics relationships of mesquite trees based on complete chloroplast sequencing

Dana Lucia Aguilar[1], Dr Mariana Lyra[1], MC Acosta[2], A CosaCov[2], Dineshkumar Krisnamoorthy[1], Stephane Boissinot[1]

[1]Evolutionary Genomics Lab, Center for Genomics & Systems Biology, New York University Abu Dhabi, , United Arab Emirates, [2]Instituto Multidisciplinario de Biología Vegetal (IMBIV-UNC-CONICET), Córdoba, Argentina

The mesquite trees (genus Prosopis, Neltuma and Strombocarpa) are keystone woody genera in arid and semiarid ecosystems across the world, where they play a central role in ecosystem functioning, soil stabilization, and the provision of resources for wildlife and human populations. These genera also hold substantial economic and cultural importance, particularly in dryland regions. Despite their importance, the phylogenetic relationships and taxonomic boundaries within mesquite trees remain poorly understood. This difficulty is largely driven by low interspecific genetic differentiation and frequent hybridization among sympatric species, patterns that suggest a recent and rapid diversification. In this study, we reconstruct phylogenetic relationships among species of mesquite trees using complete chloroplast genome sequencing. We analyzed approximately 600 individuals, representing more than 30 species, sampled throughout their native geographic distributions. Our analyses reveal contrasting evolutionary histories among clades, ranging from well-structured lineages, with clearly differentiated species, to poorly resolved species complexes characterized by shallow divergence. These findings highlight heterogeneous diversification dynamics within mesquite trees and underscore the challenges of species delimitation in recently radiated dryland taxa. Overall, these results provide new insights into the evolutionary dynamics of these keystone dryland trees and establish a robust framework for future taxonomic, biogeographic, and conservation studies in arid and semiarid regions worldwide.

[A057] De novo genome assembly of a lemur-infecting malaria parasite from long-read metagenomic sequencing

Dashiell Massey[1], Elise Paietta[2], Chris Hennelly[3], Jacob Sadler[3], Jonathan Juliano[3], Anne Yoder[2], Amy Goldberg[1]
[1]University Of California, Los Angeles, Los Angeles, United States, [2]Duke University, Durham, United States, [3]University of North Carolina at Chapel Hill, Chapel Hill, United States
Host-parasite interactions provide an important interface on which selection can act to promote co-evolution. In contrast, host-switching events, in which parasites adapt to invade new hosts, are believed to be relatively rare. However, introduction of parasites to naïve hosts may have outsized evolutionary impact. For instance, sampling of Plasmodium species across apes suggests that P. falciparum, which causes severe malaria in humans, likely diverged from the gorilla parasite P. praefalciparum only 10,000 to 50,000 years ago. Evidence for a zoonotic origin of P. vivax, while less definitive, underscores the value of broadly surveying primate malarias in natural populations. Here, we focus on a gap in existing malaria data: the parasites of lemurs, a species-rich superfamily of primates endemic to Madagascar. Using blood from free-living Avahi and Varecia individuals in the Manombo Special Reserve of southeastern Madagascar, we performed long-read Nanopore sequencing. The resulting metagenomic datasets contain a mixture of lemur (~99.9%) and parasite (~0.1%) sequence. We first enriched for parasite reads using the KrakenUniq metagenomic classifier, then generated two partial genomes (19.2Mb, 87% of conserved genes; 6.6Mb, 12% of genes) from distinct host genera. Consistent with previous mtDNA analysis of other samples, we find that these lemur-infecting parasites form a monophyletic group nested within the primate-infecting Plasmodium. We also examine the evidence for host switching among lemur hosts and for host-specific adaptation. These results contribute to our broader understanding of the evolutionary history and relationships of primate-infecting Plasmodium species and the significance of host-specificity in this large and diverse genus.

[A058] Sex-Specific Ethylene Responses Drive Floral Sexual Plasticity in Cannabis

Adrian S. Monthony[1], Davoud Torkamaneh[1]
[1]Université Laval, Quebec City, Canada

Cannabis sativa L. demonstrates exceptional sexual plasticity, enabling both XX and XY plants to achieve full phenotypic sex reversal via ethylene modulation. Though this trait is widely recognized, its molecular underpinnings are largely unexplored. This study introduces the first multi-omic investigation of hormone-triggered sex change in XX and XY Cannabis plants, merging transcriptomic analysis, ethylene pathway metabolite measurement, and whole-genome sequencing in three diverse genotypes. Applications of silver thiosulfate (STS) and ethephon yielded over 80% phenotypic transformation, yet transcriptomic shifts differed markedly by chromosomal sex. Profiling of 47 ethylene-related genes (ERGs) revealed 14 strong candidates—including CsACS1, CsACO5, CsERF1, and CsMTN—exhibiting sex-specific, temporally dynamic expression profiles that align with a two-phase plasticity framework: initial transcriptional reconfiguration succeeded by reinforcement of novel floral identities. Multiple candidate ERGs were located in non-recombining X chromosome segments or absent on the Y, with generally low nucleotide diversity implying evolutionary pressure. These insights deliver a precise depiction of ethylene-driven sex plasticity, illustrating how equivalent floral phenotypes emerge from divergent regulatory mechanisms in XX and XY individuals. The research propels forward the molecular comprehension of sexual plasticity in dioecious species and pinpoints genes for engineering sex-stable strains in Cannabis and analogous crops.

[A059] Mitochondrial Heteroplasmy Reflects Maternal Lineage and Demographic History in Southern African Populations

Dayna Croock[1], Professor Marlo Möller[1,2,3,4], Dr Caitlin Uren[1,2,3,4], Professor Desiree Petersen[1,2,3,4]
[1]South African Medical Research Council (SAMRC) Centre for Tuberculosis Research (CTR), Division of Molecular Biology and Human Genetics (MBHG), Department of Biomedical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa, [2]Centre for Bioinformatics and Computational Biology, Stellenbosch University, Cape Town, South Africa, [3]Genomics for Health in Africa (GHA), Africa-Europe Cluster of Research Excellence (CoRE), , , [4]National Institute for Theoretical and Computational Sciences (NITheCS), , South Africa
Mitochondrial heteroplasmy has been associated with aging and disease in several populations. However, little is known about the distribution and determinants of heteroplasmy in southern African populations, which harbor extensive mitochondrial diversity. We aimed to investigate the factors influencing heteroplasmic burden across southern African cohorts, with particular focus on mitochondrial ancestry. We analysed three cohorts comprising individuals with active tuberculosis (TB), latent TB infection, and healthy controls, using a combination of targeted mitochondrial sequencing and whole-genome sequencing. Heteroplasmic variants were identified and annotated, followed by statistical models being applied to identify associations with ancestry, age, sex, sequencing depth, and phenotype. Mitochondrial ancestry emerged as a consistent determinant of heteroplasmic burden across all three cohorts. Non-KhoeSan African haplogroups exhibited higher heteroplasmic burden compared to KhoeSan haplogroups, independent of age, sex, or TB phenotype. We identified lineage-specific heteroplasmic variants (m.955 poly-C insertions) that were restricted to the L0a1 haplogroup. These variants were absent in KhoeSan lineages and appear to persist at low-to-intermediate variant allele frequencies (VAF) without reaching fixation, consistent with genetic drift. The restriction of m.955 to L0a1 is plausibly explained by demographic processes associated with the Bantu Expansion. Our study provides the first evidence of lineage-specific heteroplasmy in humans and suggests that the population history of Bantu-speaking Africans and their genetic heritage, rather than disease status, shapes contemporary mitochondrial variation in southern African TB cohorts. These findings highlight the importance of considering ancestry and demographic history in studies of heteroplasmy and provide a foundation for future mitochondrial evolutionary studies.

[A060] Fast and alignment-free flavivirus classification from low-coverage genomes

Denise Kühnert[1], Afsheen Shahid, Jens-Uwe Ulrich
[1]Robert Koch Institute, Berlin, Germany

To address the inherent challenges in viral diagnostics, we developed DiCNN-UniK to overcome the computational bottlenecks and error sensitivity typical of alignment-based virus sequence classification. While traditional methods rely heavily on Multiple Sequence Alignment (MSA), the high genomic variability among viral species often renders these approaches computationally prohibitive and vulnerable to fluctuations in data quality. DiCNN-UniK provides a more efficient alternative by employing a Dual-Input Convolutional Neural Network that generates novel embeddings derived from unique k-mer signatures and universal k-mer libraries. By prioritizing direct k-mer embedding information over simple frequency-based patterns, the model captures a more sophisticated representation of the local genomic context. Notably, this architecture is engineered to analyze full-length virus genomes, bypassing the 512-token limit that frequently constrains existing genomic foundation models.

Empirical evaluation on Flavivirus datasets demonstrates that the model is exceptionally sensitive and robust, achieving 99% accuracy on an independent test set. Despite being trained on complete genomic data, DiCNN-UniK classifies partial sequences without requiring manual preprocessing, maintaining high performance even at genomic coverage as low as 20%. Consequently, DiCNN-UniK establishes a new benchmark for Flavivirus classification, offering a reliable and scalable solution for sequence analysis in real-world scenarios characterized by fragmented data and varying quality.

[A061] Evolving waters: Contrasting the population genetics of brown trout (Salmo trutta) across the island of Ireland

Diarmaid Duffy[1,2,3], Dr Richard Kennedy[2], Dr Karen Delanty[3], Prof Paulo Prodöhl
[1]School of Biological Sciences, Queen's University Belfast, Belfast, United Kingdom, [2]Agri-Food and Biosciences Institute, Belfast, United Kingdom, [3]Inland Fisheries Ireland, Dublin, Ireland

Brown trout (Salmo trutta) are among one of the most ecologically and genetically diverse vertebrates. They exhibit a range of life histories and adaptations to various freshwater and marine environments. In Ireland, S. trutta populations are shaped by their evolutionary legacy and anthropogenic impacts, including habitat modification, stocking and the introduction of invasive species. These pressures, combined with their natural ecological plasticity, have produced complex patterns of population structure and unique genetic diversity across the Ireland. Despite being such an important Irish species, S. trutta populations remain poorly characterised at high genetic resolution, particularly in systems subject to extensive stocking histories and environmental pressures.

To address this knowledge gap, an ongoing study is generating high-resolution population genetic and phylogeographic data from four contrasting freshwater systems across Ireland: Lough Erne in the northwest, Lough Mask in the west, the Carnlough lakes in the northeast uplands, and the Camcor River in the midlands. Lough Erne is influenced by historical stocking and environmental disturbances. Lough Mask supports important ferox trout (Salmo ferox) populations and faces pressures from invasive species. The remote Carnlough lakes are small upland systems with an uncertain stocking history. The Camcor River is home to the distinctive Croneen trout, a population of particular conservation concern.

Using a combination of microsatellites, mitochondrial DNA, and nuclear SNPs, the study explores how contrasting life histories, evolutionary trajectories, and anthropogenic impacts are shaping genetic diversity and population structure across these systems. The findings will provide invaluable insights to inform effective conservation and management strategies.

[A062] Evolutionary Dynamics of Intrinsically Disordered Regions in Eukaryotic Transposable Element Repressors

Diego Forni[1], Rachele Cagliani[1], Alessandra Mozzi[1], Roudin Sarama[2], Uberto Pozzoli[1], Matteo Fumagalli[2], Manuela Sironi[3]
[1]Scientific Institute IRCCS E. Medea, , Italy, [2]School of Biological and Behavioural Sciences, Queen Mary University of London, London, United Kingdom, [3]School of Medicine and Surgery, University of Milano-Bicocca, Monza,

Transposable element (TE) mobilization poses a significant fitness challenge to host genomes. Consequently, a variety of systems have emerged to silence TE activity. Just like TEs, such systems are widespread across the tree of life and their evolution is expected to be shaped by intra-genomic conflicts. To test this hypothesis, we performed an evolutionary analysis of TE control systems across different timescales. We show that a substantial fraction of TE control genes were targets of positive selection during primate evolution, with several proteins of the piRNA-pathway showing a considerable number of positively selected sites. In these proteins, selection was strongest in intrinsically disordered regions (IDRs), particularly those with low conformational entropy; we suggest that positive selection modulates IDR properties by introducing changes in sequence patterns and ensemble features properties, which are in turn related to function.
We further conducted extensive searches of Argonaute (Ago) proteins across eukaryotes. We uncover wide variability in the genomic content of Agos and an unprecedented diversity of domain occurrence, with some domain architectures reminiscent of prokaryotic defense systems. We also show that most Agos harbor IDRs at their termini and we report unusually deep conservation of primary sequence and ensemble features across huge evolutionary distances. We thus suggest that the IDRs of Ago proteins evolved under stabilizing selection to maintain a shared function.
Our data provide insight into the evolution and diversity of TE control systems in eukaryotes and contribute information to the rapidly growing evo-immuno discipline.

[A063] Reconstructing the Demographic History of a Freeze-Tolerant Frog: The Interplay Between Selection and Ne

Dylan Padilla[1], Yara Alshwairikh, Jody Taft, Julie Lee-Yaw, David Skelly[1]
[1]Yale University, , United States

The evolutionary trajectory of natural populations is fundamentally governed by the interplay between selection, which drives local adaptation, and effective population size (Ne), which determines the relative strength of genetic drift and the efficiency of selection. We conducted comprehensive genomic analyses of the wood frog (Lithobates sylvatica) to investigate this dynamic and understand how demographic history influences its adaptive potential. Our data indicate that the modern range of the wood frog is the result of a massive post-glacial connectivity between lineages that were separated by thousands of kilometers of ice for millennia. Fluctuations in Ne and varying rates of contemporary gene flow significantly modulate the efficacy of selection across populations. In populations where Ne is notably low, the influence of stochasticity may impede the fixation of beneficial alleles and accelerate the accumulation of deleterious ones, potentially exacerbating the adaptive potential of populations under certain climate scenarios. Collectively, we suggest that the current genetic architecture of the wood frog is potentially the product of both deep-time Pleistocene isolation and contemporary landscape-level dispersal dynamics. By integrating demographic modeling with functional genomics, this study elucidates how the balance between neutral and selective forces drives the evolution of temperate amphibians. Our findings provide a critical genomic foundation for predicting the resilience of the wood frog as it navigates the pressures of rapid environmental change.

[A064] Genomic consequences of recent isolation in the Marsican brown bear

Elisa Desiato[1], Giulia Fabbri[1], Silvia Fuselli[1], Roberto Biello[1], Paolo Ciucci[2], Emiliano Trucchi[3], Dr Andrea Benazzo[1], Prof Giorgio Bertorelle[1]
[1]Department of Life Sciences and Biotechnologies, University of Ferrara, Ferrara, Italy, [2]Department of Biology and Biotechnologies, Sapienza University of Rome, Roma, Italy, [3]Department of Life and Environmental Sciences, Marche Polytechnic University, Ancona, ltaly

The Marsican brown bear (Ursus arctos marsicanus) is an endemic subspecies of the central Italian Apennines, counting 50–60 individuals. We present a population genomic analysis based on 32 high-coverage whole genomes, substantially expanding available genomic resources. By integrating these data with genomes from European and North American brown bear populations, we assess population structure, genetic diversity, kinship and demographic history.
The Marsican population shows strong genetic differentiation consistent with recent isolation (~1,500 years) and absence of gene flow. Genome-wide relatedness estimates enabled the reconstruction of pedigrees, revealing a highly interconnected population structure and vulnerability to genetic drift and inbreeding.
Given the elevated risk of extinction and the ongoing debate on genetic rescue, we tested whether prolonged isolation has promoted lineage-specific genomic coadaptation that could reduce hybrid fitness following admixture. We focused on mitonuclear interactions within mitochondrial respiratory complexes and compared branch-specific evolutionary rates (ω=dN/dS) among lineages close to the Marsican brown bear using codon-based models in PAML/codeml.
The results are consistent with conserved mitonuclear coevolution along the Marsican lineage, with no evidence of emerging incompatibilities relative to other brown bear populations. Considered alongside previous genomic studies and forward simulations reporting high genetic load, deleterious mutations, distinctive behavioral traits and maintenance of variation at key functional loci, these findings confirm that the population remains at high risk, while suggesting that immediate reintroductions may not be advisable. However, should clear signs of fitness decline arise in the future, mitonuclear incompatibilities are unlikely to represent a major constraint to potential genetic rescue interventions.

[A066] Evolution of reproductive coordination in Azolla

Ellen Sigourney Lorberg[1], Craig Allen Dorsch[1], PhD Sophie de Vries[1]
[1]Georg-August University, Goettingen, Germany

The relationship between Azolla and its N_2-fixing cyanobacterial symbiont (cyanobiont) Trichormus azollae is unique among land plants. The cyanobiont is vertically inherited to facilitate a perpetual, co-dependent symbiosis. The cyanobacteria are maintained in an extracellular pocket in the leaves alongside other facultatively associated bacteria. Azolla predominantly reproduces asexually, via abscission of leaves, and rarely switches to sexual reproduction through spores, in which case the cyanobiont is transmitted maternally by the megasporocarp. We seek to identify the molecular network responsible for cyanobiont transmission in both pathways.

Sexual reproduction has been induced with far-red light. Tissue from various developmental stages of megasporocarps, those involved in cyanobiont transmission during vegetative growth, and those no longer transmitting have been sampled to perform dual-RNA sequencing of A. filiculoides and its cyanobiont. Differentially expressed genes between reproductive modes and developmental stages will be explored and analyses performed to identify the reproductive network. We will conduct population genomics of A. filiculoides populations across Europe to identify selective pressures affecting the reproductive signalling network and transmission of the cyanobiont. Leaf pockets will be isolated through enzymatic digestion of the surrounding plant to characterise the species composition of the microbiome in the pockets across populations.

The aim of this project is to identify how the reproductive network of Azolla was changed and co-opted to co-develop with cyanobacterial transmission and allow for the only truly permanent symbiosis observed in land plants other than the chloroplast.

[A067] Molecular mechanisms of thermal adaptation in urban and forest populations of frogs

Emese Balogh[1], Nikolett Ujhegyi[2,5], Zsanett Mikó[2,3], Andrea Kásler[2,3,4], Zoltán Örkényi[1], Jácint Tökölyi[6], Veronika Bókony[2]

[1]Molecular Ecology Research Group, Department of Zoology, University of Veterinary Medicine Budapest, Budapest, Hungary , , , [2]Department of Evolutionary Ecology, Plant Protection Institute, HUN-REN Centre for Agricultural Research, Budapest, Hungary , , , [3]Department of Systematic Zoology and Ecology, ELTE Eötvös Loránd University, Budapest, Hungary , , , [4]Doctoral School of Biology, Institute of Biology, ELTE Eötvös Loránd University, Budapest, Hungary, , , [5]Department of Wildlife Biology and Management, Institute for Wildlife Management and Nature Conservation, Hungarian University of Agriculture and Life Sciences, Gödöllő, Hungary, , , [6]MTA-DE "Momentum" Ecology, Evolution & Developmental Biology Research Group, Department of Evolutionary Zoology, University of Debrecen, Debrecen, Hungary, ,

Urban habitats are prone to heat events due to the heat-island effect, and wildlife populations may adapt to such environments by increased heat tolerance. We investigated the underlying proximate mechanisms in a mixed sex-determination system, where early-life heat overrides genotypic sex. We hypothesized that sex reversal may enhance thermal tolerance if the early thermal environment serves as a predictor of life-long conditions. We predicted that 1) phenotypic sexes differ in heat tolerance, 2) individuals respond to early heat exposure by developmental plasticity to elevate heat tolerance via altered gene expression, 3) sex reversal results in the sex with higher heat tolerance. Furthermore, we expected these effects to be more likely in urban populations. We tested these predictions in agile frogs (Rana dalmatina), where larval heat stress can masculinizes genotypic females, especially in urban populations. We raised field-collected embryos in a common-garden experiment and exposed half of the larvae to a heat wave in the sex-determination period. Two months after metamorphosis, we measured their Critical Thermal Maximum (CTmax), the upper temperature at which locomotor ability is lost, and performed transcriptomic analyses. Froglets from urban populations had significantly higher CTmax and altered transcriptomic profile. However, neither larval heat treatment nor phenotypic sex influenced CTmax, and sex-reversed individuals did not show elevated CTmax. These findings suggest that, if sex reversal has adaptive value, it lies elsewhere than in matching heat tolerance to the environment. However, our results suggest transgenerational adaptation of heat tolerance to urban habitats by microevolutionally diverged gene-expression responses.

[A068] Archaeogenomic reconstruction of ancient elephant ivory trade routes

Emily Johana Ruiz Puerta[1,2,3], Dr Patricia Pecnerova[1,4]
[1]Department of Biology, University of Lund, , Sweden, [2]Arctic centre, University of Groningen, , Netherlands, [3]Globe Insitute, University of Copenhagen, , Denmark, [4]Department of Biology, University of Copenhagen, , Denmark
Since antiquity, elephant ivory has been valued as a reflection of luxury, status and power. The long-term exploitation has pushed elephants towards extinction, and led to local extirpation of the North African elephant in the Late Roman times. Despite the cultural and economic significance, little is known about the ivory trade and the impacts of overhunting in past cultures, as well as the biological consequences of local extinctions. Here, we start to reconstruct the history of the elephant ivory trade using archaeogenomics, which offers the unique resolution necessary to identify the geographic origin of ivory to population level. We put shipwreck and archaeological ivory from Iberia and North Africa into the context of present-day elephant diversity, using a reference panel of more than 200 elephant genomes spanning the African continent. We show that 8 ancient specimens originating from six different sites and spanning almost two millennia in age point to the same geographic origin in northern Africa. This suggests a remarkable dominance of sourcing ivory from this region. In collaboration with historians and archaeologists, this approach will continue to uncover ancient trade routes and identify lost elephant lineages, enriching our understanding of the evolutionary history of elephants and their importance in human history.

[A069] New strategies for exploring phylogenetic tree space in Bayesian inference

Emma Gomez[1,2], John Huelsenbeck[2]
[1]California State University, Fullerton, Fullerton, United States, [2]University of California, Berkeley, Berkeley, United States

This study investigates several improvements to standard Bayesian MCMC methods for phylogenetic inference, with the goal of enhancing exploration of tree space. In conventional Bayesian phylogenetics, evolutionary relationships are estimated by sampling tree topologies, but commonly used proposal mechanisms often cause Markov chains to become trapped in local optima. To address this, I implement an empirical Bayes approach in which branch lengths are fixed at their maximum likelihood values to avoid confounding between topology updates and branch length variation. I further introduce biased tree proposals that prioritize moves toward neighboring topologies with higher maximum likelihood. Finally, I develop and test larger and more distant topology update moves that extend beyond immediate neighbors, enabling proposals to reach neighbors of neighbors and more far-reaching regions of tree space. These methods are implemented in C++ and evaluated by constructing neighborhoods of varying size, optimizing branch lengths via Quasi-Newton methods, and selecting proposals based on neighborhood-normalized likelihood. Supporting tools, including efficient topology identification through bit-vector mappings and multiple sequence alignment compression, were also developed to reduce redundant likelihood computations and focus inference on the most informative sites.

[A070] Multidisciplinary approach to study population dynamics of the Polar cod in a changing environment

Erica Difronzo[1], PhD Luca Schiavon[2], PhD Sarah Kempf[3], PhD Ilaria A. M. Marino[2], PhD Chiara Papetti[2], PhD Magnus Lucassen[3]
[1]Ca' Foscari University Of Venice, Venice, Italy, [2]University of Padova, Padova, Italy, [3]Alfred Wegener Institut (AWI), Bremerhaven, Germany

The Polar cod (Boreogadus saida Lepechin, 1774) is a key forage species in the Arctic food web, transforming the energy from low to high trophic levels. Its life cycle depends on the Arctic environment, its seasonal ice coverage and low water temperatures. Because the Arctic will experience massive changes as the ice melts due to climate changes, large impacts on the Polar cod distribution and connectivity are expected. In a context of fragile ecological balance, we explore connectivity among populations of Polar cod across the Svalbard and Iceland regions. This investigation requires a multidisciplinary approach that would disclose how adaptation and plasticity contribute to the species coping with climate change.

Preliminary analyses of a SNPs dataset generated with ddRAD-seq revealed limited genetic differentiation between Svalbard and Iceland individuals, suggesting high connectivity across the study area. Nonetheless, a small set of loci appears to be under divergent selection, indicating potential signals of local adaptation despite overall genomic homogeneity.

Besides genetic data, we expect that epigenetic variation, obtained by profiling DNA methylation patterns between regions, might disclose plastic responses to different environmental parameters. These ongoing analyses may detect subtle and short-term differentiation signals that remain undetected through classical genomic analyses, and we anticipate having initial findings to report at the conference.

By integrating genomic and epigenomic approaches, this study seeks to improve our understanding of Polar cod resilience and hence to safeguard the stability of Arctic marine ecosystems under rapid climate change.

[A071] The effect of age and condition on transcript errors

Erik Widén[1], Dr Zeeshan Syed[1], Dr Malin Larsson[2], Dr Martin Brengdahl[1], Dr Claudio Mirabello[2], Dr Martin Johnsson[3], Dr David Berger[4], Dr Urban Friberg[1]
[1]IFM Biology, Linköping University, Sweden, 581 83 Linköping, Sweden, [2]Department of Physics, Chemistry and Biology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Linköping University, 581 83 Linköping, Sweden, [3]3Department of Animal Biosciences, Swedish University of Agricultural Sciences (SLU), Box 7023, 750 07, Uppsala, Sweden, [4]Department of Ecology and Genetics, Uppsala University, Uppsala, Sweden

Organismal function depends on the faithful decoding and expression of genetic information encoded in the genome. A key step in this process is the transcription of DNA into RNA. Errors introduced into RNA molecules during or after transcription can lead to misfolded proteins, with potential consequences for fitness, aging, and disease. Identifying the conditions that elevate transcript error rates is therefore of broad biological interest.

Here, we test whether advanced age and reduced genetic quality, induced by the presence of a major genomic deletion, affect transcript error rates in Drosophila melanogaster. We focus on head tissue, which is enriched for neural cells and is particularly susceptible to proteotoxic stress and age-associated protein aggregation. We find that transcript error rates are elevated in old flies, with an increase of approximately six errors per million bases, consistent with previous studies. In contrast, we detect no effect of reduced genetic quality.

Together, these results suggest that transcript errors may contribute to the aging process, but that their age-related increase is not driven by the decline in physiological condition typically associated with aging.

[A072] Conservation and collapse of ancestral linkage groups across Nematoda

Erna King[1], Lewis Stevens[1], Martha Mulongo[1], Dr Witek Morek[1], Dr Hend Abu-Elmakarem[1], Dr Christopher Laumer[2], Mark Blaxter[1]
[1]Wellcome Sanger Institute, Hinxton, United Kingdom, [2]Natural History Museum, London, United Kingdom

Chromosomes are the fundamental organisational units of eukaryotic genomes. Groups of genes that make up the chromosomes of a common ancestor, termed ancestral linkage groups (ALG), can be inferred for groups where chromosomal-level genomes are available, allowing tracing of long-term chromosomal stability and lineage-specific reorganisation. Seven ALGs, termed
Nigon elements, were previously inferred for the last common ancestor for the nematode order Rhabditida (Class Chromadorea) using genomes sequenced from culturable species and large parasites, missing the unculturable majority too small for standard approaches. We have recently developed ultra-low long-read sequencing and low-input Hi-C approaches which allow us to generate chromosome-level genomes for previously inaccessible parts of the nematode phylogeny. Together with over 100 chromosomal genomes, these support the Nigon model deeper into Chromadorea, where most lineages have undergone relatively few rearrangements but some show dramatic reorganisation of the six ancestral autosomes and one sex-linked Nigon. Beyond Chromadorea, lineages in Dorylaimea exhibit signs of older rearrangements and Nigon partitioning. Together with genomes sequenced for the sister phylum, Nematomorpha, the first chromosomal genomes for the basal class Enoplea do not support extension of the Nigon model beyond the divide from the rest of Nematoda. Genes orthologous with those defined in Nigon X were not associated with the sex chromosome for the basal clade of nematodes or nematomorphs. These chromosomal-scale wild-caught nematode genomes suggest that ALGs of basal Enoplea diverge from those predicted by the Nigon model and demonstrate the power of these methods to uncover the evolutionary history of tiny, unculturable species.

[A073] Seapill Origami: Applying RNA Secondary Structure to Improve the Sphaeromatidae Phylogeny

Ethan Kahn[1], Will Farris[1], Dr. Regina Wetzer[1]
[1]Natural History Museum of Los Angeles County, Los Angeles, United States

Sphaeromatidae is a family of nearly 700 species of small, mostly shallow marine isopods. Its members demonstrate high morphological variability, from smooth or flattened body forms to strong sculpting and protuberances, with or without sexual dimorphism, which has contributed to a long and often conflicted taxonomic history. Previous attempts to construct molecular phylogenies have confirmed monophyly of the family and identified distinct supra-generic groupings, but were unable to resolve many deeper relationships due to extremely variable expansion regions in the otherwise slowly evolving 18S rDNA gene (sequence length 1967–2865 bp). Those studies experimented with eliminating such regions where poor alignment did not appear to represent homologous positions. However, doing so removed important phylogenetic information, collapsing deeper tree nodes. Because hydrogen-bonded stem regions of the ribosomal RNA are more strongly conserved than the unbonded loop regions, sections of these variable insertions can be aligned with high confidence and preserve phylogenetic signal. We produce more accurate alignments for 16S and 18S rDNA, by accounting for the evolutionary constraints of RNA secondary structure, and combine them with COI sequences for better resolution of a multi-locus sphaeromatid phylogeny. Hundreds of sequences are already published for these three loci, including for difficult to sample species, so re-analysis which leverages existing data is a practical way to resolve phylogenetic relationships. We also add representatives of 12 previously unsequenced genera, improving taxonomic coverage. Our phylogeny sets the stage for exploring the biogeography, morphological innovations, reproductive systems, and diversification of this abundant, widespread isopod family.

[A074] Evolution of gene expression and regulation in a haplodiploid pollinator

Eva Adina Baumgarten[1], Jannik Sven Möllmann[1], Dr. Thomas Joseph Colgan[1]
[1]Institute Of Organismic And Molecular Evolution (iomE), Johannes Gutenberg University Mainz, Mainz, Germany

Distinct sexes are found in many organisms and can exhibit differences in a range of traits, including morphological, behavioural, physiological, as well as patterns of gene expression. These differences are especially pronounced in haplodiploid organisms, in which females and males differ in ploidy, creating fundamental contrasts in genome architecture and gene expression landscapes. However, our understanding of whether ploidy leads to differences in gene expression and regulation, as well as the relative strength of evolutionary processes, remains fragmented in haplodiploids. First, using mason bees (Osmia species), we are employing long- and short-read transcriptomics to characterise gene expression differences in amplitude and splicing between the sexes. Second, by combining transcriptomic data with whole-genome sequencing, we will assess whether splice variants are the products of evolutionary processes, such as selection or drift, and how they contribute to sex-biased expression. Collectively, our study will shed light on the evolution and expression of transcript isoform diversity in an ecologically important group of haplodiploid pollinators. Furthermore, it will improve our understanding of how gene expression patterns diverge between sexes with differing ploidy, as well as show how conserved these patterns possibly are across species. These findings will highlight the role of gene expression and regulation as a potential substrate of evolutionary change and provide insight into how sex-specific pressures, selection, and drift interact to shape haplodiploid systems.

[A075] DNA repeats, sources of extremes in DNA composition in orchids?

Eva Hribova[1], Zuzana Chumova[2], Simona Martikanova[1], Jana Cizkova[1], Karol Krak[2], Premysl Balaz[2], Pavel Travnicek[2]

[1]Centre of Plant Structural and Functional Genomics, Institute of Experimental Botany, CAS, Olomouc, Czech Republic, [2]Department of Evolutionary Plant Biology, Institute of Botany, CAS, Pruhonice, Czech Republic

Representatives of Orchidaceae family exhibit extreme variation in genome size, from 1C = 0.21 pg in Lepanthes larvina to 56.1 pg in Liparis purpureoviridis, with the presence of common and partial endoreplication, a unique feature of orchids. Another interesting phenomenon, which is probably related to the phenomenon of partial endoreplication is high variability in GC content in DNA, ranging from 22.5 – 50.5%. Species with PE have on average larger genome sizes than species with conventional endoreplication, due to the accumulation of repetitive elements in non-replicated parts of the genome. It is possible that selective accumulation or suppression of certain repetitive elements in orchid genomes can lead to the enormous variation in their nucleotide composition. In our study, we have analyzed genome size, GC content and composition of repeatomes of selected orchids representing species with conventional and partial endoreplication, including closely related species with extreme variation in nucleotide composition. With the aim to test the hypothesis that the accumulation or loss of specific repetitive elements is responsible for the huge variation in GC content, we used combination of flow cytometry to measure genome size and to estimate the AT/GC composition of the orchids genomes and whole genome skim-sequencing of more than 50 selected orchids to analyse repeatomes. We find out different dynamics of specific repetitive elements within closely related group of species that resulted into the enormous variation of genome size and GC content in the orchids.

[A076] Characterisation of major histocompatibility complex diversity in the sand lizard (Lacerta agilis)

Felicia Axelsson[1]
[1]Uppsala University, Uppsala, Sweden

The Swedish sand lizard (Lacerta agilis) is a red-listed species with small and fragmented populations. This increases the risk of genome-wide loss of genetic diversity, due to genetic drift and population bottlenecks. This loss of diversity may also extend into the highly variable immune gene region: the Major Histocompatibility Complex (MHC). The antigen-presenting genes of the MHC contribute to pathogen-recognition and immune system function in vertebrates. These genes evolving extreme polymorphism in response pathogen-mediate selection. The high degree of polymorphism of the MHC region therefore plays a vital role in disease resistance. This project is investigating the diversity of the MHC (both class I and class II) across Swedish sand lizard populations using amplicon sequencing. MHC variation across populations will be compared to genome-wide genetic variation from whole genome sequencing data. In a world where climate change will likely affect the distribution of disease vectors, such as expanding the distribution of parasites, it is crucial that wild populations have the ability to adapt to new pathogens. This project will contribute to a greater understanding of the adaptive genetic variation in the sand lizard which will help the conservation of the species and other wildlife, and lead to a better understanding of the consequences of loss of genetic diversity and immunocompetence in wild populations.

[A077] Investigating signatures of local adaptation in the ubiquitous forest ectomycorrhizal fungus Cenococcum geophilum

Felix Zimmermann[1,2], Dr. Benjamin Dauphin[1], Dr. Martina Peter[1], Daniel Croll[2], Prof. Mika Tarkka[3,4], Prof. Lars Opgenoorth[5]
[1]Swiss Federal Research Institute WSL, Birmensdorf, Switzerland, [2]University of Neuchâtel, Neuchâtel, Switzerland, [3]Helmholtz Centre for Environmental Research - UFZ, Halle an der Saale, Germany, [4]German Centre for Integrative Biodiversity Research (iDiv) Halle-Jena-Leipzig, Leipzig, Germany, [5]Philipps University Marburg, Marburg, Germany

Temperature and drought stress on European forests has increased tree mortality, yet little is known about mechanisms of climate adaptation in associated ectomycorrhizal fungal partners and the role of shared evolutionary history. Here, we focus on the symbiotic interaction between Pedunculate oak (Quercus robur), an ecologically and economically important tree species, and Cenococcum geophilum, a cosmopolitan, often dominant ectomycorrhizal fungus known for its drought tolerance. Across 21 sampling sites spanning a climatic gradient, we measured soil parameters in situ and extracted modelled atmospheric data. We isolated 400 fungal strains and conducted whole genome sequencing to identify genotype-environment associations and genomic regions underlying local adaptation. Additionally, we will genotype 16 oaks per site to investigate population structure and the shared demographic history of this host-fungus pair. Finally, we will identify co-occurring host and symbiont genotypes associated with specific environmental conditions.
We expect to find various C. geophilum lineages within and across sampling sites, with specific gene variants associated with soil chemistry and water availability. We expect Q. robur populations to be structured mainly along a latitudinal gradient, by topography, and other persistent barriers to gene flow. Finally, we expect to identify genotype-genotype combinations associated with certain climatic and soil conditions and to detect concordant changes in effective population size (Ne) between Q. robur and its dominant symbiotic partner C. geophilum.
Understanding the impact of extreme climatic events on both partners of a symbiotic relationship is essential for assessing processes of environmental adaptation and identifying the gene variants involved in natural selection.

[A078] Genomic Architecture of Adaptation at the Range Limit: X-linked and Autosomal Divergence in Ischnura elegans

Fereshteh Ghadamgahi[1], Sofie Nilén[1], Professor Erik I. Svensson[1]
[1]Lund University, Lund, Sweden

Understanding how species adapt at their geographic range limits is a central question in evolutionary genomics. At these boundaries, populations face intense environmental pressures, including physiological constraints and novel photoperiodic regimes. In this study, we utilized high-depth sequencing data from the damselfly Ischnura elegans—representing a latitudinal transect from Scandinavia to the Mediterranean—to identify the loci underpinning local adaptation. A genome-wide FST scan revealed a highly heterogeneous landscape of differentiation with several prominent "genomic islands" of divergence. Our most significant findings include a major selective sweep on the X-chromosome, where a sharp differentiation peak (FST = 0.73) was identified at the 2.4 Mb locus. Functional annotation reveals this hotspot harbors a cluster of candidate genes, including an axonemal dynein-like locus (LOC124154608), associated with reproductive maturation. Notably, we also identified a novel autosomal selection signal on Chromosome 13 (FST ≈ 0.30). This distal locus contains a protein-coding candidate, LOC124170117 (gastrula zinc finger protein xLCGF3.1-like), suggesting that transcriptional regulation is a key driver of environmental response. Additionally, independent signals on Chromosome 2 point toward a polygenic architecture underlying the physiological transition between northern and southern lineages. These results demonstrate that both the X-chromosome and specific autosomal loci serve as critical genomic targets for rapid divergence, facilitating adaptive responses to contrasting thermal and photoperiodic conditions across the species' range.

Keywords: Genomic islands of divergence, Latitudinal clines, X-linked adaptation, Ischnura elegans, Zinc-finger protein, FST profiling.

[A079] Genetic Signatures Of Mitonuclear Coevolution In The Mexican Biobank.

Finley Grover-Thomas[1], Ram Gonzalez Buenfil[2], Santiago Medina-Muñoz[2], Dr. Andres Moreno-Estrada[2], Professor Aida Andres[1], Dr Florencia Camus[1]
[1]University College London, , United Kingdom, [2]CINVESTAV, , Mexico

Mitochondria play key roles in metabolism and biosynthesis, and have a fundamental impact on cell and individual fitness in eukaryotes. Efficient mitochondrial function is contingent on over 1,000 protein coding genes, the vast majority of which are encoded in the nuclear genome. Differences in mutation rate, recombination and inheritance mean that the mitochondrial and nuclear genomes have somewhat distinct modes of evolution. Nonetheless, mitonuclear theory of coevolution predicts that genes important to mitochondrial function in both genomes will evolve to work together efficiently. A building body of work has found evidence of such coevolution, both in natural and experimental populations, although the underlying mechanisms of this process remain poorly understood.

Here we test the hypothesis of mitonuclear coevolution in Latin American human populations from Mexico which, within the last 500 years, have experienced intense pulses of male-biased admixture from European and African populations. Using sequence data from 6,000 individuals in the Mexican Biobank, we investigate the patterns of local ancestry in nuclear-encoded genes associated with mitochondrial function, as defined by the MitoCarta3.0 database. We detect small, but significant differences in the evolutionary patterns of mitochondrially-associated genes when compared with a background set of similarly sized non-interacting genes, consistent with predictions from mitonuclear coevolutionary theory. We find that the signature differs across mitochondrial pathways and between geographical regions, and identify the genes which are the strongest candidates for mitonuclear-driven positive selection.

[A080] Molecular characterization of temperature tolerance in the methylotrophic yeast Komagataella phaffii (Pichia)

Finley Pitcock[1], Trenton Winters[1], Regina Edgington[1], Damien Wilburn[1,2]
[1]Department of Chemistry and Biochemistry, The Ohio State University, Columbus, United States, [2]Center for RNA Biology, The Ohio State University, Columbus, United States

Thermal tolerance is a highly plastic trait among organisms, with many common laboratory microbial species (e.g., Escherichia coli, Saccharomyces cerevisiae) being highly adapted to a narrow optimum. The yeast Komagataella phaffii (Pichia) is a popular heterologous protein expression system that can produce high yields of recombinant protein with eukaryotic post-translational modifications. Pichia grow to high cell densities on minimal media with many simple carbon sources (dextrose, glycerol, methanol), and the use of carbon source-specific promoters provides an easy induction mechanism for heterologous overexpression. Additionally, we have found that Pichia grow efficiently across a broad temperature range (~4-37 °C), and we hypothesize Pichia could serve as a reliable recombinant expression system for temperature-sensitive proteins. However, it is unknown how gene expression may be affected across this broad temperature range. Here, we investigated the cell doubling times, molecular phenotypes, and metabolic rates of Pichia cultured under various temperatures and carbon sources. We found that Pichia doubling time monotonically decreased as a function of temperature in all carbon sources, but at different non-linear rates. Despite differences in growth rate, we observed that maximum culture cell density was dependent only on the carbon source and not on temperature. Carbon source depletion from the media as a function of temperature was measured by nuclear magnetic resonance (NMR) spectroscopy. Quantitative mass spectrometry-based proteomics and long-read transcriptomics were applied to Pichia grown under temperatures spanning 5-35 °C in different carbon sources. These methods were utilized to identify temperature and carbon-dependent changes in cellular protein expression.

[A081] Evolution of domesticated plasmid replication control in Pantoea.

Florence Muccino[1], Dr. Devani Romero Picazo[1], Dr. Nils Hülter[1], Pr. Dr. Tal Dagan[1]
[1]Institute Of General Microbiology, CAU Kiel, Kiel, Germany

Recent studies estimated that about 10% of the bacterial taxa harbour an accessory chromosome. Such replicons can arise through plasmid domestication – when a plasmid evolves into a vertically inherited chromosome. The mechanisms underlying the coordination of accessory chromosome replication with the primary chromosome are best described in Vibrio cholerae, but remain poorly understood in other taxa. In the plant-associated genus Pantoea, two prevalent plasmids – LPP-1 and LPP-2 – have been identified as domesticated based on phylogenetic evidence, displaying features such as vertical inheritance, presence of core genes, and GC content similar to the chromosome. Here, we investigate if and how the replication of both plasmids is coordinated with the primary chromosome in Pantoea agglomerans R1. Replication dynamics assessed by Marker Frequency Analysis show that LPP-1 replicates in synchrony with the chromosome, whereas replication timing of LPP-2 could not be reliably resolved due to its smaller size. Using droplet digital PCR, we show that both replicons are maintained at a low copy number of approximately one per cell, consistent with chromosome-like replication control. Further investigation of inter-replicon interactions using high-throughput chromosome conformation capture indicates spatial coupling of the chromosomal origin of replication and that of LPP-1. Together, our results provide evidence that LPP-1 is an accessory chromosome in Pantoea and demonstrate that its replication is synchronised with that of the primary chromosome, potentially via physical inter-replicon interactions.

[A082] HoloTrait: A trait-based evolutionary framework for microbiome stability, dysbiosis, and recovery

Franceen Eshun-Wilson[1]
[1]University Of Copenhagen, Copenhagen, Denmark

The stability of host-associated microbiomes is fundamental to host fitness, yet the evolutionary mechanisms governing resilience, collapse, and persistent dysbiosis remain poorly understood. Many existing models treat microbial communities as static taxonomic assemblages, limiting insights into how functional trait interactions shape long-term system dynamics. Here, we introduce HoloTrait, a trait-based simulation framework for modeling holobiont evolution under ecological and selective constraints.

HoloTrait simulates microbial community dynamics across discrete generations using three stages: (i) microbial pool mixing, (ii) within-host proliferation, and (iii) host-level selection implemented via a multivariate Gaussian fitness kernel centered on ecological targets. Microbial communities are represented in trait space using CSO strategies—Competitors, Stress-tolerators, and Opportunists—parameterized through Dirichlet distributions to capture compositional inheritance and variation. By systematically varying selection pressure and environmental targets, we examine how evolutionary pressures shape convergence, resilience, and extinction risk.

Our simulations reveal that intermediate selection strengths promote convergence toward stable equilibria, whereas strong selection induces evolutionary collapse or persistent dysbiosis. Following abrupt shifts in ecological optima, communities with higher trait diversity recover more rapidly and exhibit lower extinction rates. Fitness–distance trajectories reveal nonlinear adaptive dynamics and critical thresholds beyond which recovery becomes unlikely.

HoloTrait provides a general modeling framework linking trait-based community ecology with host–microbiome evolution. By explicitly modeling selection on functional composition through its effect on host fitness, this approach offers a testbed for exploring microbiome resilience, evolutionary tipping points, and holobiont adaptation. Ongoing work integrates empirical gut microbiome data to evaluate predicted transitions between stable and dysbiotic state.

[A083] Population genomics of South American Mennonites.

Maria Castaña Muñoz Arriaga[1], Fernanda Valenga[2], Angelica B. W. Boldt[2,3], Elena Bosch[1], Francesc Calafell[1]

[1]Institut de Biologia Evolutiva (CSIC-UPF), Universitat Pompeu Fabra, Departament de Medicina i Ciències de la Vida, Barcelona, Spain, [2]Laboratory of Human Molecular Genetics, Department of Genetics, Federal University of Paraná, Curitiba, Brazil, [3]. Postgraduate Program in Internal Medicine, Federal University of Paraná (UFPR, Curitiba, Brazil

The Mennonites are an Anabaptist group of Frisian/Flemish origin that originated after the Protestant Reform in the 16th century (other Anabaptist branches are the Amish and the Hutterites). Advocating for a more radical reformation, including pacifism and the doctrine of adult baptism, Mennonites were religiously and politically persecuted and because of that ended up becoming an isolated population. Persecution drove complex migrations from settlements in Danzig (1561) to the Ukraine (1788), and subsequently to North and South America (1874-1946). To ascertain how this complex history is reflected in the genomic diversity of South American Mennonites, we sequenced the full genomes of volunteers from several Mennonite communities in Bolivia and Brazil (N=50 from each country). We have compared individual SNP genotypes with reference populations from Europe, East Asia and the Americas, by means of PCA and ADMIXTURE, to reflect their complex migration patterns and detect possible gene flow from these populations. By measuring the number and length of runs of homozygosity as well as estimating FIS, we have weighed the contribution of isolation and reduced effective population size vs. consanguinity in the inbreeding patterns observed, as well as linking those to the social isolation or openness practices of each community. We want to emphasize that this project was instigated by the Mennonite community itself, which will obtain a return not only in the form of a better appreciation of their history, but also in terms of a grounded awareness of the health implications of that history.

[A084] A genomic snapshot of the Ottoman army in Southern Italy

Mr Giulio Castellano[1], Dr Lehti Saag[2], Claudia Decarolis[2], Mait Metspalu[2], Prof Kristiina Tambets[2], Dr Sandro Sublimi Saponetti[1], Francesco Montinaro[1,2]
[1]Dept. of Biosciences, Biotechnology and Environment, University of Bari, Bari, Bari,Italy, BARI, Italy, [2]Institute of Genomics, University of Tartu, Tartu,Italy, Tartu, Estonia

The Ottoman Empire, also known as the Turkish Empire, originated as an Anatolian principality and expanded into a transcontinental power by integrating vast territories across Southeast Europe, West Asia, and North Africa from the 14th century to the early 20th century, conquering Constantinople in 1453. In July 1480, the Ottoman Empire laid siege to Otranto, located at the southernmost tip of Italy in Apulia, and eventually conquered it on the 11th of August. Despite the conquest lasting just thirteen months, the army led by Commander Ahmed Pascha has been estimated at approximately 18.000 soldiers, who, according to written records, were characterised by diverse geographic origins. In this work, we present the first paleogenomic investigation of eight individuals excavated at Otranto, identified as members of the Ottoman occupying force through archaeological and burial context.

Our analysis shows that, despite the low number of analysed samples and the low sequence coverage, the genomic variation in these individuals is remarkably high, with ancestry traceable to many different areas, including the Balkans and the Mediterranean, reflecting the Empire's complex recruitment strategies and the integration of conquered populations. Notably, one individual shows strong evidence of a Central Asia origin, confirming the high genomic diversity of the Ottoman army.

[A085] Apulia at the crossroads of the Mediterranean: fine-scale population sub-structure and admixture inferred from allele-frequency and haplotype-based analyses

Francesco Perrone[1], Apulia at the crossroads of the Mediterranean: fine-scale population sub-structure and admixture inferred from allele-frequency and haplotype-based analyses Alessia Daponte[1], Apulia at the crossroads of the Mediterranean: fine-scale population sub-structure and admixture inferred from allele-frequency and haplotype-based analyses Luciana De Gennaro[1], Apulia at the crossroads of the Mediterranean: fine-scale population sub-structure and admixture inferred from allele-frequency and haplotype-based analyses Sarah Putignano[1], Aurora Lorusso[1], Apulia at the crossroads of the Mediterranean: fine-scale population sub-structure and admixture inferred from allele-frequency and haplotype-based analyses Mario Ventura[1,2], Francesco Montinaro[1,3]
[1]Department of Biosciences, Biotechnology and Environment, University of Bari, Bari, 70124, Italy, Bari, Italy, [2]Department of Genome Sciences, University of Washington School of Medicine, Seattle, WA, USA, Seattle, USA, [3]Institute of Genomics, University of Tartu. Institute of Genomics. Riia 23b, 51010. Tartu, Estonia, Tartu, Estonia

The Italian Peninsula displays high genetic differentiation compared to other European populations, as a result of long-term migrations and cultural exchanges due to its central position in the Mediterranean Sea. In particular, Southern Italy played a key role in these interconnections, with the Apulian region representing a notable case of study.
In this project, we performed allele-frequency and haplotype-based high-resolution methods to analyze a genome-wide SNP dataset comprising approximately 1,600 Apulian individuals and about 11,500 individuals from West Eurasia and North Africa, sourced from the literature.
We characterized the genetic structure of the Apulian population, investigating admixture events and evolutionary features that have contributed to its genetic variability.
Furthermore, we examined the composition of the Apulian ancestry, its clustering patterns with other European populations, and the impact of demographic and historical events on population structure through the time.
Our results reveal fine-scale genetic substructure within Apulia and multiple admixture signals consistent with historical gene flow. Overall, the outcomes highlight the combined effects of migrations, evolutionary forces, local adaptation, natural selection, and population-specific demographic history that shape present genetic patterns.

[A086] How intracellular mutualism emerges: developmental and evolutionary perspectives through an integrative study of an aphid symbiosis

François Gilbert[1], François Renoz[1]
[1]UCLouvain, Louvain-la-Neuve, Belgium

A central challenge in evolutionary biology is understanding how stable intracellular mutualistic associations between eukaryotes and prokaryotes emerge from free-living or facultative ancestors. Insects provide powerful models to address this question, as many lineages harbor organelle-like bacterial symbionts. While comparative genomics has revealed extensive genome reduction in long-term obligate symbionts, the mechanisms driving the evolutionary transition from a free-living to a host-dependent intracellular lifestyle remain poorly resolved.

Here, we propose an integrative evolutionary framework using the aphid Periphyllus lyropictus and its obligate nutritional symbiont Serratia symbiotica as a model system. Uniquely, this symbiont retains an invasive phenotype, exhibiting both intracellular localization within bacteriocyte cells and extracellular presence across multiple host tissues. This dual lifestyle positions the system along an evolutionary gradient between parasitism and mutualism, offering a rare opportunity to study how these relations shift along the symbiotic continuum.

Addressing such lifestyle transition requires integrative approaches that bridge multiple biological levels, from genome architecture and gene regulation to cellular localization and ecological context. The production of high-quality reference genomes at chromosomal scale appears necessary for understanding these complex systems, which are subject to significant evolutive pressures implicating genomic restructuring, gene loss, and symbiont-mediated horizontal gene transfer.

By combining transcriptomics, tissue-level imaging, and experimental manipulation, we aim to investigate how functional and cellular integration of the symbiont varies across tissues and environmental conditions. This framework highlights the importance of viewing symbioses as dynamic evolutionary trajectories shaped by interactions at multiple levels.

[A087] Genomic insights into the convergent evolution of ant-eating mammals

Rémi Allio[1,2], Christopher A. Emerling[3], Prof. Celine Scornavacca[1], Dr Frédéric Delsuc[1]
[1]ISEM, Univ. Montpellier, CNRS, IRD, Montpellier, France, [2]CBGP, INRAE, CIRAD, IRD, Montpellier SupAgro, Univ. Montpellier, Montpellier, France, [3]Biology Department, Reedley College, Reedley, USA

Understanding the genomic mechanisms underlying convergent evolution is a central question in evolutionary biology. Although convergence is widespread across the tree of life, many aspects of how similar phenotypes arise from distinct evolutionary histories remain unresolved. Ant-eating mammals provide a powerful model system, as myrmecophagy has evolved independently at least seven times in mammals and is associated with striking morphological and physiological similarities. Here, we investigate the molecular basis of convergent adaptation to myrmecophagy within a comparative genomic framework. Using a hybrid sequencing and assembly strategy, we generated 10 new high-quality genomes from myrmecophagous species and close relatives. These genomic resources enabled us to apply state-of-the-art statistical approaches to detect convergent amino acid evolution in protein-coding genes, characterize patterns of gene loss and pseudogenization, and infer gene family expansions and contractions in convergent myrmecophagous lineages across a dataset of more than 200 mammalian genomes. The results show that historical contingency played a major role in the convergent evolution of ant-eating mammals. The two most ancient myrmecophagous lineages (pangolins and anteaters) notably took different evolutionary paths to adapt to the myrmecophagous diet. Our findings illustrate both the influence of historical contingency and the power of natural selection in shaping the genomes of convergent myrmecophagous phenotypes in mammals.

[A088] Genomic adaptation to aridity in the Arabian sand boa

Gabriel Mochales-Riaño[1], Dr. John Neddermeyer[2], Sebastian Kirchhof[3], Theo Busschau[1], Vahid Nikoonejad[2], Marc Tollis[2], Simone Gable[2,4], Stéphane Boissinot[1]
[1]New York University Abu Dhabi, , United Arab Emirates, [2]School of Informatics, Computing, and Cyber Systems, Northern Arizona University, Flagstaff, AZ, USA, , USA, [3]Natural History Museum, Abu Dhabi., , United Arab Emirates, [4]Center for Evolution and Medicine, Arizona State University, Tempe, AZ, , USA

Desert environments impose extreme thermal, hydric, and energetic constraints that require specialized adaptations, yet the genomic basis of arid adaptation in reptiles remains poorly understood. We present a chromosome-level reference genome for the Arabian sand boa, Eryx jayakari, a fossorial snake restricted to hyper arid regions of the Arabian Peninsula. The 1.5 Gb assembly shows high completeness and strong chromosomal synteny with other snakes, providing a robust framework for comparative analyses. Using phylogenomic data from eight additional squamate genomes, we investigated lineage specific shifts in selective pressure across 14,966 single copy orthologs. Gene-wide tests for episodic positive selection identified a small but biologically coherent set of candidate genes after multiple testing correction (n=26).
Notably, olfactory receptor genes were overrepresented among positively selected loci, consistent with enhanced chemosensory reliance in subterranean and visually reduced environments. Gene ontology enrichment suggested functional themes related to vascular development, epithelial integrity, and sensory perception, processes plausibly linked to thermoregulation, water balance, and life underground.
Demographic reconstruction indicated population expansions coinciding with Pleistocene glacial intervals, periods that in Arabia were associated with increased aridity, supporting a historical association between increasing desertification and habitat suitability. Together, these results provide one of the first genome scale views of desert adaptation in a reptile and highlight sensory and physiological pathways as key components of evolutionary responses to extreme aridity.

[A089] Using Demographic Models and Ancient European Genomes to Identify Signals of Selection due to Admixture

Gauri Bhide[1], Kelsey Witt[1]
[1]Department of Genetics and Biochemistry and Institute for Human Genetics, Clemson University, Clemson, United States

Admixture – the exchange of genetic material from previously diverged populations, is an important source of genetic variation. Many modern human populations are admixed, but it is difficult to disentangle these multiple ancestral sources which limits our understanding of how admixture shapes genetic variation. To know how selection affects allele frequencies, we need to disentangle its effects from effects of demographic events such as genetic drifts and bottlenecks. This study uses published ancient European genomes and demographic simulations to provide a better understanding of how admixture affects allele frequencies through positive and negative selection. We achieved this by simulating chromosomes using msprime – a population genetics simulator, and matching them with published genomes, by accounting for various demographic parameters such as population size, gene flow. We modelled the admixture event between Anatolian Neolithic Farmers and Italian Hunter-Gatherers, in Neolithic Europe, to compare the allele frequencies at three different time points (one before admixture event and two after). Since msprime does not account for selection, the comparison enables us to identify the alleles in the genome under positive and negative selection. Our preliminary results show signals of selection in multiple genomic regions that have previously been identified, including OCA2 (decreased melanin production) and genes involved in immune function. We also detected possible selection signals in genes involved in innate immunity and skeletal development. Our work suggests that pairing ancient admixed genomes with demographic simulations is a powerful tool for examining human demographic history.

[A090] Guiding experimental design for efficient sampling of fitness landscapes

Georgios Kalogiannis[1], Prof.Dr. Andreas Wagner[1,2,3]
[1]University Of Zurich, Zurich, Switzerland, [2]Swiss Institute of Bioinformatics, Lausanne, Switzerland, [3]Santa Fe Institute, Santa Fe, United States

Fitness landscapes are high-dimensional topographical maps, where each location on the landscape corresponds to a genotype and its elevation corresponds to fitness. The study of a fitness landscape is central to understanding adaptation, drug resistance, and protein engineering. However, it is challenging due to the non-additive interactions of base pairs along a genotype, and their expansive size that grows exponentially with sequence length and rapidly exceeds feasible experimental coverage. Here I propose a framework that uses combinatorially complete fitness landscapes to derive decision rules for efficient experimental sampling, targeting three key design elements: (1) how many genotypes to assay, (2) how deeply to sequence each genotype, and (3) how much of each variant to read. Deep learning models trained on sparse subsets of only 1-5% of the landscape recover a large portion of top-percentile genotypes, with performance degrading sharply below a landscape-dependent minimum coverage. Contrary to expectations, random genotype selection outperforms diversity-maximising designs, indicating that maximising amino acid coverage does not improve identification of adaptive peaks. Additionally, incorporating protein structural features derived from AlphaFold predictions improves fitness prediction and functional/nonfunctional classification, reducing uncertainty in imputation of unmeasured genotypes. Simulating downsampling reveals a minimum read-depth at which peak recovery is possible, while truncating genotypes shows predictable losses in performance, especially in epistatically dense regions. Together, these results provide practical guidance to the development of protocols and allocating experimental resources, enabling efficient landscape mapping and high-fitness discovery.

[A091] A genomic snapshot of the Chalcolithic-Bronze Age transition from the pile-dwelling site of Ledro, Italy

Nicola Rambaldi Migliore[1], Giacomo Villani[1], Nicola Nannini[2], Valeria Nicolini[1], Anna Tommasi[1], Elisabetta Moroni[1], Anna White[3], Ilaria Piloni[1], Simone Floresta[2], Manuela Pernter[2], Sabrina Buscè[2], Marta Gobbi[2], Amedeo Luigi Zanetti[2], Valentina Miretti[2], Erika Vitali[2], Hannes Schroeder[3], Antonio Salas[4], Anna Olivieri[1], Nicola Carrara[5], Donato Riccadonna[2], Luca Pagani[6], Alessandro Fedrigotti[2], Alessandro Achilli[1]

[1]Department of Biology and Biotechnology, University of Pavia, Pavia, Italy, [2]MUSE - Museo delle Scienze, Trento, Italy , [3]Globe Institute, Faculty of Health and Medical Sciences, Øster Farimagsgade 5, Copenhagen, Denmark, [4]Unidade de Xenética, Instituto de Ciencias Forenses (INCIFOR), Facultade de Medicina, Universidade de Santiago de Compostela, Santiago de Compostela, Spain, [5]Museo della Natura e dell'Uomo, University of Padova, Padova, Italy, [6]Department of Biology, University of Padova, Padova, Italy

The Alpine region has long acted as a semi-permeable barrier within Europe, shaping human mobility, cultural interactions, and genetic structure across deep time. In particular, pile-dwelling settlements in the Alps represent an important archaeological record of prehistoric Europe. However, no genomic data from Alpine lake-dwelling communities have been generated and analyzed so far. In this study, we present a genomic snapshot from the pile-dwelling site near Ledro Lake in Trentino-Alto Adige/Südtirol, northern Italy). This key settlement, located above Lake Garda, at the crossroads between the Po Plain and the Alpine corridors, has been inhabited since at least the transition from the Copper Age and throughout the Bronze Age (2200-1350 BCE).

We analyzed human remains from five individuals recovered from this site that were radiocarbon dated from the Copper Age to the Roman period. We obtained deep genomic data from two of them (at 1.8x and 5x coverage, respectively) and contextualized their genomes within a comparative framework of modern and ancient populations from western Eurasia, as well as newly generated genome-wide data (Human Origins array) for 48 present-day individuals living in Ledro Valley for at least the past two generations. Using allele-frequency analyses and haplotype-based techniques, we investigate how this community related to other contemporary groups in northern Italy and the rest of central and southern Europe and reconstructed their demographic history and admixture processes (e.g., the arrival of the Steppe-related ancestry), providing a first snapshot of a pile-dwelling community in northern Italy.

[A092] ProteomeLM: A proteome-scale language model enables accurate and rapid prediction of protein-protein interactions and gene essentiality across taxa

Cyril Malbranke[1,2], Gionata Paolo Zalaffi[1,2], Anne-Florence Bitbol[1,2]
[1]Institute of Bioengineering, School of Life Sciences, EPFL, Lausanne, Switzerland, [2]SIB Swiss Institute of Bioinformatics, Lausanne, Switzerland

Language models trained on biological sequences are advancing inference tasks from the scale of single proteins to that of genomic neighborhoods. Here, we introduce ProteomeLM, a transformer-based language model that uniquely operates on entire proteomes from species spanning the tree of life. ProteomeLM is trained to reconstruct masked protein embeddings using the whole proteomic context, yielding contextualized protein representations that reflect proteome-scale functional constraints. ProteomeLM spontaneously captures protein-protein interactions (PPI) in its attention coefficients. Furthermore, it enables interactome-wide PPI screening that is substantially more accurate, and orders of magnitude faster, than amino-acid coevolution-based methods. We further develop ProteomeLM-PPI, a supervised model that combines ProteomeLM embeddings and attention coefficients to achieve state-of-the-art PPI prediction across benchmarks and species. Finally, we introduce ProteomeLM-Ess, a supervised gene essentiality predictor that generalizes across diverse taxa. Our results demonstrate the potential of proteome-scale language models for addressing function and interactions at the organism level.

[A093] Microbiome Atlas of Portuguese Aquaculture Fish

Mr. Felipe Schoninger[1], Dr. Maria João Peixoto[2], Gonçalo Themudo[1]
[1]Ciimar / University of Porto, Matosinhos, Portugal, [2]Tallin Zoo, Tallin, Estonia

A critical aspect of sustainable aquaculture is reducing disease burden in farmed fish. This issue is particularly important for larvae, when health assessment is difficult and problems are often detected too late to prevent losses. Non-invasive or minimally invasive techniques for disease prediction could revolutionise the sector by enabling prophylactic interventions, reducing financial loss and enhancing sustainability, animal health, and welfare.

Imbalances in microbial flora are linked to numerous pathologies, yet knowledge of fish microbiome lags behind that of humans and other mammals. Advances in sequencing technologies now enable rapid and affordable characterization of microbial populations. This has the potential to inform on taxonomic diversity, which can serve as the basis of prophylactic measures. Such progress offers an opportunity to accelerate fish microbiome research, providing insights into taxonomic diversity and informing strategies such as dietary supplements to improve inflammatory responses. By comparing microbiomes of animals reared in optimal versus suboptimal conditions, researchers can identify profiles associated with greater disease resilience.

We will create an open repository of metagenomic data for key fish species in Portuguese aquaculture, including trout, seabass and seabream. MAPAfish will characterize the metagenomes of multiple tissues in fish reared under optimal conditions and those exposed to environmental and biological stressors. By integrating publicly available data, MAPAfish aims to leverage metagenomics as a tool for early disease diagnosis and improve aquaculture management.

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[A094] Are chromosomal inversions promoting speciation in the Tyrrhenian wall lizard?

Guannan Wen[1], Daniele Salvi[2], Tobias Uller[3], Nathalie Feiner[1]
[1]Max Planck Institute for Evolutionary Biology, Plön, Germany, [2]University of L'Aquila, L'Aquila, Italy, [3]Lund University, Lund, Sweden

Chromosomal rearrangements are critical drivers of reproductive isolation and local adaptation. We investigated the genomic architecture underlying population divergence in the Tyrrhenian wall lizard Podarcis tiliguerta across Corsica and Sardinia. By integrating RAD-seq data from 47 localities with whole-genome sequencing, we uncovered pronounced population structure both between and within islands, with strong geographic patterns. Genome-wide scans revealed that regions of genetic differentiation are strongly concentrated at one end of each acrocentric chromosome. Using linkage disequilibrium network analysis, PCA-based clustering, and heterozygosity contrasts, we identified 24 candidate inversions co-localizing with these terminal regions, many of which were identified in both islands. Clinal analyses across multiple transects further showed that several inversions exhibit steep, concordant clines and reduced introgression, indicating localized barriers to gene flow despite low genome-wide differentiation. Given the species' inferred ancient hybrid origin, these inversions may have captured and stabilized divergent genomic blocks, serving as a persistent source of variation for early-stages of speciation. Our results highlight how chromosomal inversions act as "islands of differentiation", maintained by endogenous barriers to recombination, thereby facilitating the progression towards reproductive isolation.

[A095] Genomic and phenological variation across the geographic range of sugar maple (Acer saccharum)

Hanna Makowski[1], Alan Bergland[1], Ana Carnaval[2], Meghan Blumstein[1]
[1]University Of Virginia, , United States, [2]City College New York, , United States

Long-lived, wind-pollinated tree species provide a powerful system for examining a central question in evolutionary biology: how does adaptive variation persist despite extensive gene flow? High dispersal capacity can homogenize allele frequencies across populations, yet many widespread species exhibit pronounced genetic and phenotypic structure, suggesting that selection can counteract connectivity. Here, we use genomic data from over 800 individuals spanning the geographic range of sugar maple (Acer saccharum) to characterize patterns of genetic structure and connectivity and evaluate how geographic distance, environmental gradients, and landscape features shape intraspecific diversity. We further assess the genetic basis of intrapopulation variation in leaf-out timing,a fitness-related trait tightly linked to growing season dynamics,across two populations to determine how phenological variation is distributed within and among populations. Together, these results illuminate how local adaptation can emerge and persist in long-lived species with substantial dispersal.

[A096] A pleiotropic hitchhiking model recapitulates alignments between fly wing divergence and variation

Haoran Cai[1]
[1]University of California, Los Angeles, Los Angeles, United States

The alignment between mutational variance (M), standing genetic variance (G), and macroevolutionary divergence (R) in Drosophila wing shape presents a rate paradox: evolution follows genetic lines of least resistance, yet proceeds orders of magnitude slower than quantitative genetic theory predicts. Two explanations have been proposed: ``deleterious pleiotropy,'' where stabilizing selection on unmeasured traits outside the wing morphology complex constrains evolution, and ``correlational selection,'' where selection acts directly on trait covariances. However, recent empirical work has found little evidence of fitness costs associated with wing shape variation beyond effects on flight performance, undermining the deleterious pleiotropy hypothesis. Here, I propose and evaluate a pleiotropic hitchhiking model, in which natural selection targets a single functional trait (wing size) while the complex geometry of wing veins evolves as a correlated response shaped by the structure of M. Using individual-based simulations parameterized with empirical mutational covariances, I demonstrate that univariate selection on a primary trait can recapitulate the observed alignment among M, G, and R. Importantly, the pleiotropic hitchhiking model can also generate slower-than-neutral divergence rates, offering a partial resolution to the rate paradox without invoking hidden pleiotropic costs on unmeasured traits. These findings suggest that the long-term evolution of fly wing geometry may be a largely neutral byproduct of selection on size, constrained by mutational pleiotropy within the measured trait complex.

[A097] Global Pathogen Analysis Platform

Harrison Bo Hua Zhu[1,2,3], Professor Christian Brinch[3], Professor David Alejandro Duchêne Garzon[1], Professor Henrik Wegener[3], Professor Frank Møller Aarestrup Aarestrup[3], Prof Samir Bhatt[1,2]
[1]University of Copenhagen, Copenhagen, Denmark, [2]Imperial College London, London, United Kingdom, [3]Technical University of Denmark, Lyngby, Denmark

The COVID-19 pandemic yielded unprecedented genomic data, yet the capacity to translate these sequences into actionable evolutionary intelligence remains unequally distributed. Sophisticated genomic data preparation and phylogenetic inference often requires bioinformatic expertise inaccessible to many health professionals. Here, we present the vision of the Global Pathogen Analysis Platform (GPAP), a digital public infrastructure designed for rapid and high-performance genomic epidemiology.

Moving beyond static pipelines, we introduce a novel "Agentic AI" framework as the platform's analytical core. Unlike passive Large Language Models, these autonomous bioinformatic agents are designed to reason, plan, and execute multi-step workflows. We present a proof-of-concept (PoC) where an AI agent interprets natural language queries (e.g., "Estimate the time-varying reproduction number from this viral dataset") and autonomously orchestrates the necessary tools - retrieving metadata, performing genomic data processing, and executing phylogenetic inference - while maintaining "human-in-the-loop" verification.

We discuss the challenges of deploying agentic systems in evolutionary biology, particularly regarding hallucination mitigation and reproducibility. By shifting technical execution to an intelligent co-pilot, GPAP aims to accelerate the global detection of evolutionary anomalies, ensuring advanced surveillance tools are available to the entire scientific community.

[A098] The role of lncRNAs in Dryophytes arenicolor (Canyon Treefrog) metamorphosis, an evolutionary perspective

Héctor Herrera-Orozco[1,2], Doctor Hibraim Adán Pérez-Mendoza[2,3]
[1]Posgrado en Ciencias Biológicas, UNAM, Ciudad Universitaria, Coyoacán, México, [2]Laboratorio de Ecología Evolutiva y Conservación de Anfibios y Reptiles, Facultad de Estudios Superiores Iztacala, UNAM, Tlalnepantla de Baz, México , [3]Laboratorio Nacional de Biología del Cambio Climático, SECIHTI, CDMX, México

Anuran metamorphosis is a post-embryonic developmental program that transforms a tadpole into a froglet through extensive morphological, anatomical, and physiological changes regulated by conserved endocrine and molecular signalling pathways. Despite strong conservation of these pathways across vertebrates, the contribution of non–protein-coding transcripts to metamorphosis remains poorly understood. Although only ~5% of the genome encodes proteins, the majority is transcriptionally active, suggesting an important regulatory role for non-coding RNAs. Among these, long non-coding RNAs (lncRNAs) have emerged as key regulators of gene expression at multiple levels.

Here, we investigated the role of lncRNAs during metamorphosis in the Canyon Treefrog by sequencing transcriptomes across multiple life-cycle stages. We annotate both previously described and de novo lncRNAs and identify stage-specific expression patterns through differential expression and functional enrichment analyses. To explore regulatory roles, we construct gene co-expression networks and predict interactions between lncRNAs and other molecular players. Finally, we assess the evolutionary conservation of metamorphosis-associated lncRNAs across vertebrate taxa using structure-aware multispecies alignments and RNA sequence covariance analyses.

Together, this work provides a comprehensive view of lncRNA dynamics during amphibian metamorphosis and offers insights into the evolutionary conservation of non-coding regulatory mechanisms underlying vertebrate development.

[A099] Towards Reconstructing the Genetic History of East Asian Goats

Heeseung Yeum[1], Christina Warinner[2,3], Choongwon Jeong[1,4]
[1]School of Biological Sciences, Seoul National University, Seoul, Republic of Korea, [2]Department of Anthropology, Harvard University, Cambridge, USA, [3]Department of Archaeogenetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany, [4]Institute for Data Innovation in Science, Seoul National University, Seoul, Republic of Korea

One of the earliest species to be domesticated, goats originated from the Anti-Taurus and Zagros mountains in the Near East approximately 11,000 years ago. While East Asian goats are believed to originate from ancient Iranian populations, their dispersal history remains poorly explored. In this study, we developed genome-wide panels of single nucleotide polymorphisms (SNPs) for high-resolution demographic analysis of goats based on published whole genome sequences of 297 modern world-wide Capra individuals, resulting in ~12 million high-quality transversion SNPs. Among them, we chose 974,000 SNPs and designed a set of oligonucleotide probes for targeted capture. We also produced genotype calls of published ancient goat individuals from the Near East, Europe, and China on this panel. Principal Component Analysis (PCA) focused on East Asian domestic goats reveals that modern Chinese goats are genetically distinct from West and South Asian lineages. Interestingly, the earliest domestic goats in China so far sequenced, dated to ~4,000 years ago and from the Shimao archaeological site, displays a strong affinity to modern Chinese goats, while contemporaneous goats from modern-day Uzbekistan exhibits a profile closer to West Asian goats. Considering the extensive human migration in Eurasia between the Early Bronze Age and Iron Age, Eurasian goats likely played a significant role in shaping the genetic profile of Chinese goats, including the Shimao population. We plan to generate genome sequencing data for ancient individuals from the Early-Middle Bronze Age to the modern era across the Eurasian Steppe, Inner Asian Mountain Corridor, and China.

[A100] Regulation of a "tripartite" Toxin Antitoxin system from the honeybee intestinal bacteria Apilactobacillus kunkeei.

Héléna Vassilieff[1], Kristina Näslund[1], Erik Hagström[1], Professor Siv Andersson[1]
[1]Department of Molecular Evolution, Cell and Molecular Biology, Biomedical Centre, Scientific for Life Laboratory, Uppsala Universitet, Uppasla, Sweden

Toxin Antitoxin (TA) systems are ubiquitous among bacteria. The most studied TA systems belong to the type II, which consist of two proteins: one antitoxin that binds and inhibits its cognate toxin. The resulting complex is the regulatory factor of its own bicistronic operon. This regulation and the instability of the antitoxin ensure the maintenance of plasmids carrying these TA systems within bacterial populations through a mechanism known as post-segregational killing (PSK). Interestingly, some type II TA systems present a non-canonical tripartite organization. The supplementary gene can encode a second antitoxin, a transcription regulator, or a chaperone-like protein. However, the prevalence of this tripartite organization remains unknown. Apilactobacillus kunkeei is a fructophilic lactic acid bacterium, which is abundant in the honeybee crop and helps honeybees in their defense against pathogens. In 2022, Dyrhage et al. characterized 102 plasmids from newly sampled strains of A. kunkeei. One type of the plasmids code for antimicrobial peptides which have been shown to inhibit the honeybee pathogen Melissococcus plutonius in vitro. However, 74 of these plasmids had unknown functions. Using the Toxin-Antitoxin database, phylogenetic, structural and clustering analyses, the 74 plasmids were examined to unravel the presence of TA systems. We identified 20 plasmids carrying type II TA systems specific to A. kunkeei and A. apinorum. Remarkably, these systems display an atypical regulatory organization. Our findings challenge the traditional view of the bicistronic type II TA and raise new questions about their regulatory mechanisms and potential roles in PSK.

[A101] A Hierarchical Foundation Model for Microbial Taxonomic Prediction and Downstream Applications

Herui Liao[1], Prof. Tami Lieberman[1]
[1]Mit, Cambridge, United States

Foundation models for microbial compositional data could enable critical downstream applications including disease prediction, microbiome engineering, and biomarker discovery. However, the massive taxonomic vocabulary (>180,000 taxa) and inherent data sparsity pose significant challenges. Here we explore a hierarchical token prediction framework that leverages the natural structure of microbial taxonomy for foundation model development.

Our approach treats each taxon as a token and employs hierarchical masked token prediction across seven taxonomic levels (Domain through Species), where predictions at each level are conditioned on parent taxonomic assignments. We trained the model on over 700,000 metagenomic community samples and evaluated its ability to correctly predict masked taxonomic tokens on the validation data. Preliminary results show 64.26% top-1 token prediction accuracy (the correct taxon ranked first by the model) at species level compared to 17.04% for frequency-based and 2.68% for random baselines, with top-10 accuracy (correct taxon within top 10 predictions) reaching 94.20% versus 38.22% and 16.34% respectively. These initial results suggest the model learns biological patterns beyond simple taxonomic frequency distributions. By capturing hierarchical relationships within community contexts, this framework could enable species-level inference from genus-resolved data (e.g., 16S rRNA gene sequencing) and generate microbial community embeddings for downstream applications including disease prediction and microbial interaction inference.

[A102] A practical comparison of genomic prediction methods in ecology and evolution

Heung Ying Janet Chik[1,2], Prof Stefanie Muff[1,2]
[1]Department of Mathematical Sciences, Norwegian University of Science and Technology, Trondheim, Norway, [2]Gjærevoll Centre, Norwegian University of Science and Technology, Trondheim, Norway

In recent years, genomic data from wild populations have become increasingly available, offering exciting potential for genomic prediction (GP) – the use of genomic data to statistically estimate, for example, the additive contribution (or breeding value) of one's genes to its phenotype. However, while a multitude of GP methods have been developed to predict breeding values, and subsequently additive genetic variance in a population, the application of those methods on wild datasets is still at infancy, and there is currently little examination of their applicability under different trait and population scenarios. Here, we systematically simulated phenotypic and genomic datasets, varying the underlying genetic structure, trait heritability, population size, and SNP marker density, covering realistic ranges found in the wild. We then applied a conventional pedigree animal model, and three GP models – Genomic-BLUP, BayesR, and a recently developed Bayesian principal component ridge regression (BPCRR) model – to these datasets. We estimated respectively the individual breeding values, additive genetic variance, trait heritability, as well as computation time under each scenario. These metrics were then compared with the true values and across methods to assess the precision, accuracy and efficiency of each GP method. Through this systematic investigation, we aimed to not only better understand the strengths and weaknesses of current GP methods, but also to provide a practical guide on GP method selection for evolutionary biologists based on the characteristics of their study population and trait, and hence promote the use of GP in the wild.

[A103] Karyotypic analysis of established primate iPSCs by cytogenetic and M-FISH techniques

Hideyuki Tanabe[1], Risako Nakai[2,3], Ayako Nagahashi[2,4], Yusuke Hamazaki[5], Satoshi Hirata[6], Haruhisa Inoue[2,3,4], Masanori Imamura[5,7]
[1]Research Center for Integrative Evolutionary Science (RCIES), The Graduate University for Advanced Studies, SOKENDAI, Shonan Village, Hayama,, Japan, [2]Laboratory of Stem Cell Medicine, Department of Cell Growth and Differentiation, Center for iPS Cell Research and Application (CiRA), Kyoto University, Kyoto,, Japan, [3]iPSC-based Drug Discovery and Development Team, RIKEN Bioresource Research Center (BRC), Kyoto,, Japan, [4]Medical-risk Avoidance based on iPS Cells Team, RIKEN Center for Advanced Intelligence Project (AIP), Wako,, Japan, [5]Center for the Evolutionary Origins of Human Behavior (EHUB), Kyoto University, Inuyama,, Japan, [6]Wildlife Research Center, Kyoto University, Kyoto,, Japan, [7]Graduate School of Medicine, Kanazawa University, Kanazawa,, Japan
The advancement of methods for establishing the induced pluripotent stem cells (iPSCs) is remarkable and its application for various species is important for the conservation of rare species in the light of biodiversity and for comparative studies with humans from evolutionary viewpoint. To date Imamura et al. have been internationally promoting the project “Turning whole zoo organisms into iPS cells” and have continued to establish iPSCs from rare species (https://stemcells.or.jp/animail/). So far, primate iPSCs derived from the great apes, namely, chimpanzees (including one iPSC from an individual showing symptoms of Down syndrome), bonobos, gorillas, and orangutans, as well as Japanese monkeys, have been established and various studies have been conducted (Nakai et al., 2022, 2018; Imamura et al., 2020, 2018, 2014; Hamazaki et al., 2025). It was difficult to establish the lesser apes’ iPSCs probably due to their cytological and genomic characteristics. To overcome this problem Imamura’s research group has made a lot of technical improvements and finally succeeded in establishment of the white-handed gibbon (Hylobates lar) and siamang (Symphalangus syndactylus) iPSCs (Hamazaki et al., 2025). In this study conventional cytogenetic banding and M-FISH (24XCyte; MetaSystems) techniques were applied onto all primate iPSCs to examine the chromosomal compositions and karyotypes. As a result, it was found that approximately 90% of all iPSCs showed normal karyotypes and remained ones were almost numerically abnormal karyotypes and structural rearrangements were quite rarely detected.

[A104] Global Population Genomics of Marchantia polymorpha: Patterns of Gene Flow Across Subspecies and Continents

Hiromu Nakao[1,2], Johannes Rötzer[1], Svetlana Akimcheva[1], Katharina Jandrasits[1], Takashi Tsuchimatsu[2], Liam Dolan[1], Frédéric Berger[1]
[1]Gregor Mendel Institute of Molecular Plant Biology, Vienna, Austria, [2]University of Tokyo, Graduate school of Science , Tokyo, Japan

Land plants develop multicellular bodies in both phases of the life cycle. In bryophytes – one of the two monophyletic clades of land plants – the multicellular haploid phase (gametophyte) is free-living and produces gametes. Upon fertilisation of an egg by a motile spermatozoid a relatively small spore-producing diploid phase (sporophyte) develops. In addition to sexual reproduction, assexual reproduction occurs in many species, in which genetically, identical clonal propagules are produced. Populations may therefore derive from individuals produced bu sexual or asexual reproduction or both.

Despite their evolutionary importance, the population dynamics and speciation processes of bryophytes remain understudied.

Marchantia polymorpha, a model liverwort distributed across the Northern Hemisphere, comprises three subspecies occupying distinct ecological niches that sometimes occur sympatrically. This system provides a powerful opportunity to investigate early stages of subspecies differentiation in a cosmopolitan bryophyte.

We analyzed approximately 250 natural accessions from East Asia, Europe, and North America. Our results suggest that the three subspecies are diverging nearly simultaneously while remaining incompletely differentiated. Patterns of population structure vary among subspecies, with one showing strong signatures of long-distance dispersal likely driven by human activity. We further examined adaptive evolution within each subspecies, with particular attention to convergent evolution, balancing selection, and the role of gene flow.

Finally, we compared evolutionary trajectories across genomic regions, focusing on differences between autosomes and sex chromosomes, providing insight into genome evolution under the UV sex determination system characteristic of bryophytes. Overall, our results highlight key processes shaping the evolution of bryophyte species.

[A105] Tracing the spread of Celtic languages using ancient genomics

Hugh McColl[1], Guus Kroonen[2], Thomaz Pinotti[1], William Barrie[3], John Koch[4], Johan Ling[5], Jean-Paul Demoule[6], Kristian Kristiansen[1], Martin Sikora[1], Eske Willerslev[1]
[1]Section For GeoGenetics, Globe Institute, University of Copenhagen, Copenhagen, Denmark, [2]Leiden University Centre for Linguistics, Leiden University, Leiden, The Netherlands, [3]Department of Genetics, University of Cambridge, Cambridge, United Kingdom, [4]University of Wales Centre for Advanced Welsh and Celtic Studies, Aberystwyth, United Kingdom, [5]Department of Historical Studies, University of Gothenburg, Gothenburg, Sweden, [6]University of Paris I - Panthéon-Sorbonne, Paris, France

Celtic languages, including Irish, Scottish Gaelic, Welsh and Breton, are today restricted to the Northern European Atlantic seaboard. However, between 3000 and 2000 BP (years before present), Celtic was widely spoken across most of Europe before being largely replaced by Germanic, Latin or Slavic. Despite this rich history, how Celtic became spread across the European continent remains contentious. The debate is currently focused around three main models based on historical linguistics and archaeology: (a) the Celtic-from-the-West model; b) the Celtic-from-the-Centre model; and c) the Celtic-from-the-East model. Here, we utilise new genomic data from Bronze and Iron Age Europe to test how the population histories align with the three models of prehistoric spread of the Celtic languages. We find transmission of Steppe ancestry through the Bell Beaker region from Northwest Europe 4400–4000 BP, followed by population movements from Southwest Europe to the British Isles 3800-3200 BP. Finally, we see a spread of Knovíz-related ancestry from Eastern Central Europe 3200–2800 BP to France, the British Isles and Iberia. While our results support core elements of all three models, we find the Celtic-from-the-East model to be most parsimonious as it fits both the population genetic events discovered and the majority of date estimates for the formation of Proto-Celtic. While we note human history does not always follow the most parsimonious path, any further development of models on the spread of the Celtic languages must reconcile with the new population genomic findings presented.

[A106] Assessing Parental Contribution to Genetic Inheritance: Age-dependent Mutational Landscapes in Macaca fascicularis Families

Hye-Ri Park[1,2], Hyeon-Mu Cho[1], Se-Hee Choe[1], Min-Gyeong Ko[1,2], Yun-Jung Lee[1,2], Sang-Je Park[1], Young-Hyun Kim[1], Jaewon Huh[1,2]

[1]National Primate Research Center, Korea Research Institute of Bioscience and Biotechnology (KRIBB), Cheongju-si, Republic of Korea, [2]Department of Functional Genomics, KRIBB School of Bioscience, Korea University of Science and Technology (UST), Cheongju-si, Republic of Korea

Mutations are genetic alterations that typically arise from spontaneous errors during DNA replication or are induced by environmental factors such as radiation and chemicals. These changes can occur in somatic as well as germ cells and tend to accumulate incrementally over the course of an individual's life, thereby contributing to the biological processes associated with aging. Of particular significance are germline mutations, which arise in reproductive cells and can be transmitted to offspring, and de novo mutations, which emerge spontaneously in the subsequent generation. The analysis of such mutations provides critical insight into hereditary disorders and facilitates the investigation of genetic abnormalities within familial lineages. In this study, we performed whole genome sequencing (WGS) on whole blood (30x coverage) and sperm (50x coverage) to analyze mutation rates and patterns in crab-eating monkeys (Macaca fascicularis). We selected four parent-offspring trios (a total of 16 monkeys; 10 males, 6 females) representing various life stages to collect genomics data across different age points. Over a five-year period, we established a longitudinal genomic dataset of macaque whole blood, buccal and sperm cells. Using this data, we developed comprehensive bioinformatics pipelines to quantify annual mutation accumulation. We performed a longitudinal quantitative analysis to assess the accumulation of mutations in the genome over a five-year interval, specifically evaluating de novo mutations in offspring and mutational profiles in male monkey spermatozoa. Furthermore, we conducted a quantitative pattern analysis to evaluate variations associated with aging and pedigree.

[A107] Ancient genomes from the Shiveet Khairkhan archaeological site reveal the genetic connectivity between Early Iron Age pastoralists

Hyoungmin Moon[1], Mijin Park[1], Juhyeon Lee[1,2], Donghee Kim[1,2], Eun Min Cho[3], Suyeon Kim[3], Soyeong Kang[3], Takashi Gakuhari[4], Dr Choongwon Jeong[1,2]
[1]School of Biological Sciences, Seoul National University, Seoul, 08826, South Korea, [2]Institute of Data Innovation in Science, Seoul National University, Seoul, South Korea, [3]National Research Institute of Cultural Heritage, Daejeon, 34122, South Korea, [4]Institute for the Study of Ancient Civilizations and Cultural Resources, Kanazawa University, Ishikawa, Japan

The Pazyryk culture, well known for its burial mounds in the Altai Mountains, represents a key component of the Scytho-Siberian world-a cultural horizon consisting of Early Iron Age nomadic pastoralists across the Eurasian steppe. However, the genetic relationships between the Pazyryk and other contemporary nomadic populations remain unclear. In this study, we report genome-wide shotgun sequencing data from 5 Pazyryk (BC 400-190) and 6 Xianbei period (BC 80 – AD 340) individuals excavated from the Shiveet Khairkhan archaeological site in Bayan-Ulgii Aimag, Mongolia. We find that the ancient Shiveet Khairkhan individuals exhibit characteristic ancestral profiles of Early Iron Age Central Asian Steppe nomads, best modeled as a three-way admixture of Western steppe herders, “Khovsgol-like” Eastern Asians, and Southern Central Asian “BMAC-like” groups. The Shiveet Khairkhan ancients show no evidence of recent inbreeding and carry two Y-chromosome haplogroups: Q1b, commonly found in the Altai region, and J2a, associated with Western Scythians. To observe the fine-scale affinities between the Shiveet Khairkhan and other Scytho-Siberian populations, we estimated identity-by-descent (IBD) segment sharing among Early Iron Age nomads, using genotypes imputed with the 1000 Genome Project phase 3 reference panel. Our analysis provides an in-depth view into the genetic structure and connectivity within the Scytho-Siberian world by leveraging state-of-the-art population genomics methodologies.

[A108] Past population structure and indicators of gene flow in a malaria parasite, Plasmodium falciparum

Håvard Bjerke[1], Valentin Duda-Jouan, PhD Hugh Mccoll, Frederik Seersholm, Fernando Racimo, Martin Sikora
[1]University Of Copenhagen, , Denmark

Understanding the long-term dynamics of Plasmodium falciparum is essential for contextualizing modern malaria epidemiology and its current population structure. Historical genomic data from pre-eradication Western Eurasia remain extremely limited, leaving its genetic history largely unknown. Here, we present ancient Plasmodium infections in a 230-year-old individual from Aknashen, Armenia. We show that the individual carried a co-infection of three of the major human-infecting Plasmodium parasites, P. falciparum, P. vivax and P. malariae, demonstrating their presence in the southern Caucasus regions before their eradication.

Using in-solution hybridization capture combined with shotgun metagenomic sequencing, we recovered genome-wide P. falciparum nuclear data at 0.13X average read depth, as well as a 3.5X mitochondrial genome. Joint analyses with published ancient genomes and modern population data shows that the ancient Armenian P falciparum has close genetic affinities with pre-eradication strains from southern Europe represented by a previously published sample from Spain, suggesting connectivity and a distinct Western Eurasian genetic cluster. Building on this, we construct admixture graphs and test alternative demographic scenarios for P. falciparum, providing new insights into its historical population structure and dispersal in Europe.

[A109] Evolution of the prokaryotic transcription factor repertoire

Inder Raj Singh[1], Dr. Aswin Sai Narain Seshasayee[1]
[1]National Centre For Biological Sciences, Bengaluru, India

Transcription factors (TFs) regulate transcription initiation. Though they help determine phenotype across the tree of life, they are nonessential for a minimal cellular life and are often absent or nearly so in endosymbiotic and parasitic organisms. In light of this fact and the idea that it is a certain level of organism complexity that calls for transcription regulation, we traced the evolutionary history of TF repertoire on a bacterio-archaeal tree of life using a dataset of >400,000 TFs, grouped into ~1,700 orthologous groups (OGs) across ~3,000 species. The number of TFs, through evolutionary history, matches expectations against genome size derived from extant bacteria. The most ancestral prokaryotes encoded tens of TFs, representing 11 HTH superfamilies. Going by extant functions, these TFs possibly regulated sugar-fermentation metabolism, sensed overall metabolic state and redox, responded to DNA damage or bound metals; many of which are consistent with some reconstructions of the ancestral gene pools. Emergence of new TFs largely follows a smooth cumulative distribution curve, suggesting steady innovation, particularly early in evolution, in contrast to eukaryotes, in which a majority of TF families emerged at the ancestors of multicellular lineages. Over half of all TF OGs had emerged by the time 34% of all nodes in the tree evolved, again in contrast to the late elaboration of these proteins in the animalia ancestors. Gains of TFs late in prokaryotic evolution predominantly feature recycling of protein families discovered elsewhere, consistent with the dominance of horizontal gene transfer in prokaryotic evolution.

[A110] Merging evolutionary timescales to quantify adaptation

Ioanna Kotari[1,2], Carolin Kosiol[3], Rui Borges[4]
[1]Vienna Graduate School Of Population Genetics, , Austria, [2]Institute of Population Genetics, University of Veterinary Medicine, 1210, Vienna, Austria, [3]Centre for Biological Diversity, School of Biology, Fife, KY16 9TH, St Andrews, UK, [4]School of Mathematics and Statistics, University of St Andrews, North Haugh, KY16 9SS, St Andrews, UK

Positive diversifying selection promotes recurrent changes in amino acid sequences, driving adaptive evolution. In phylogenetics, it is commonly inferred using the ratio of non-synonymous to synonymous substitution rates (dN/dS). However, standard codon models typically rely on a single representative genome per species and neglect population-level processes such as selection on synonymous codons and GC-biased gene conversion (gBGC), both of which can bias dN/dS estimates and generate false positives, especially in the presence of within-species variation.

The growing availability of sequences in the genomics era makes it possible to study adaptive evolution with greater resolution by combining interspecific and intraspecific variation. In this study, we introduce a Polymorphism-aware Phylogenetic Model for Codon Evolution (PoMoCod), which models adaptive evolution at a population-genetics scale, while leveraging both fixed and polymorphic differences between species. We compare our framework with standard codon models to assess how population-level forces bias signals of positive selection.

We observed that in the absence of diversifying selection, but in the presence of confounding population forces, dN/dS estimates struggled to remain close to the neutral expectations and frequently produced inflated values. When explicitly simulating selection on non-synonymous sites, the dN/dS recovered the expected signal and correlated well with the simulated values, but consistently overestimated the magnitude of selection. In conclusion, while the dN/dS-based inference remains a valid and useful tool, it can overestimate adaptive signals when population-level processes influence sequence evolution beyond idealised assumptions. This study highlights current limitations and how population-aware approaches can improve the accuracy of evolutionary inference.

[A111] Long-read genome assemblies of museum samples

Ioannis Chrysostomakis[1], Astrid Böhne[1]
[1]Leibniz Institute For The Analysis Of Biodiversity Change, Bonn, Germany

Museum collections worldwide contain thousands of specimens, many from species that are extinct, endangered, or otherwise inaccessible, making the collection of fresh material for genomic analyses difficult or impossible. In this project, we developed wet- and dry-lab techniques to analyze whole genome sequences from museum samples with unique traits, showcasing how museums can be leveraged to improve our understanding of the genetics of vulnerable and rare species using high-quality, long-read data. As part of broader efforts to sequence all life on earth, we explored the genomic potential of particularly interesting vertebrates from wet museum collections that cannot be sampled in the field.

We produced genome assemblies from PacBio long-read HiFi sequencing and performed comparative gene annotation to understand the genomic underpinnings of phenotypic diversity. As a case study, we sequenced a pair of recently diverged catfish species exhibiting a marine-to-freshwater transition. This analysis revealed how transposable elements and genome architecture can diverge in closely related species and accompany rapid adaptations in these systems.

Our findings underscore both the potential and limitations of using museum samples compared to fresh specimens. This research not only delivers well-annotated reference genomes from museum collections but also offers a roadmap for unlocking their genomic potential, demonstrating that historical specimens can yield insights into evolutionary processes and biodiversity that would otherwise remain inaccessible.

[A112] Geolocating Neanderthal Introgression using spatial simulations

Ioannis Patramanis[1], Dr Laurits Skov[1]
[1]Globe Institute, Copenhagen Univeristy, , Denmark

The last two decades have seen significant progress in the study of archaic human groups, such as Neanderthals and Denisovans, and their interactions with the ancestors of present day humans. Questions of when, where and how many times these groups interbred (introgression), have stood at the forefront of the field. Genetic simulations have played an important role in studying these interactions, by allowing us to model and test competing hypotheses, but spatiality; a crucial aspect of population theory, is still largely omitted. We use Slim and its non Wright-Fisher spatial simulation capabilities to investigate different introgression scenarios. We model an expanding 'modern human' population and a smaller and more static 'archaic' population, controlling for the exact location of their interbreeding. We test different scenarios of admixture, such as a short vs a prolonged coexistence of the two populations and a single vs multiple encounters scenario. We generate hundreds of iterations of each scenario and for each one of them, record a number of summary statistics pertaining to introgression, exploring which can best illuminate the origin of an introgression event in deep time. We then compare the summary statistics of our simulations to those generated from published genomes of present day and ancient humans, to assess possible locations of past Neanderthal introgressions into modern humans.

[A113] Comparative organoid systems for functional genomics research in primate skeletal cells

Irina Milicevic[1], Bayaz Suleymanli[1], Dr. Genevieve Housman[1]
[1]Department of Primate Behavior and Evolution, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

Comparative genomic studies using induced pluripotent stem cells (iPSCs) from primates are an important tool for advancing our understanding of primate evolution and development. These cell culture models are especially valuable for studying tissues that are difficult to obtain in a well-preserved state, such as connective tissues. However, most existing models use monolayer cultures, which may not accurately replicate native tissue architecture and function. In this study, we developed spheroid-based culture systems using primate iPSCs to model early osteogenic, chondrogenic, and adipogenic differentiation and to examine gene expression variation within these systems. Specifically, up to four human and four chimpanzee iPSC lines were differentiated into mesenchymal stem cells, aggregated into three-dimensional spheroids, and differentiated into chondrogenic, osteogenic, or adipogenic lineages, and single-cell RNA sequencing data were collected at multiple time points throughout differentiation. Gene expression profiles were obtained from approximately 78,000 cells, revealing differential expression across cell types and between humans and chimpanzees. To identify native early developmental cell types most similar to those in the spheroid cultures, spheroid gene expression profiles were compared to reference datasets of early connective tissue development, including the human embryonic limb cell atlas. Altogether, these comparative primate organoid models provide a novel approach for examining the functional genomics of primate bone, cartilage, and adipose tissue and are helping to clarify the molecular mechanisms underlying evolutionary differences in connective tissue development and maintenance.

[A114] Recombination landscape evolution in Podarcis wall lizards

Iris Liesbeth Ruesink Bueno[1], Julien Dutheil[1], Nathalie Feiner[1]
[1]Max Planck Institute for Evolutionary Biology, Plön, Germany

Wall lizards (genus Podarcis) are extremely diverse in their colour patterns, both between and within species. Throughout the evolutionary history of Podarcis, there have been repeated occurrences of hybridisation between distant lineages, which may have enabled the diversification of the group by bringing together divergent genetic variants and sorting them into novel combinations. The distribution of recombination rates across the genome is bound to have played an important role in the reshuffling of variation in the descendant hybrid lineages. However, little is known about recombination landscapes in Podarcis. To fill this knowledge gap, we are constructing recombination maps for a number of wall lizard populations, both from the same and different species, using whole-genome resequencing data obtained from wild individuals. We apply two different methods to infer historical recombination rates: 1) Pyrho, which makes use of the patterns of linkage disequilibrium within a population together with the likelihood of observing those patterns given a coalescent model that incorporates the population's demographic history, and 2) iSMC, a purely coalescent-based method that can be applied to a single diploid genome. This allows us to establish how the distribution of recombination rates has evolved across the genus, determine whether the degree of recombination landscape divergence mirrors phylogenetic distance and identify any genomic regions where recombination rates are either exceptionally conserved or exceptionally labile in Podarcis.

[A115] Somatic mutation signatures as markers of immune and environmental stress in Gentoo Penguins

Isabel Gamache[1], Dr. Marie-Julie Favé[1]
[1]Biology Department, Concordia University, Montréal, Canada

Somatic mutations accumulate during an organism's lifetime, and can further clonally expand within regenerating tissues, generating somatic mosaicism with implications for immune function, aging, and health. In vertebrates, hematopoietic stem cell-derived lineages are central to immune defense, and somatic evolution within these cells may reflect both endogenous processes and environmental stressors, and may be used for biomonitoring of wildlife health status. Although clonal hematopoiesis (CH) is well documented in humans, its prevalence and evolutionary significance in wildlife remain largely unexplored.

Here, we present the first population-scale analysis of somatic point mutations in hematopoietic cells from a wild avian species, the Gentoo penguin (Pygoscelis papua). Using whole-genome sequencing data from 129 individuals available across genetically differentiated colonies, we adapted a somatic mutation detection pipeline originally developed for human cancer genomics to a non-model seabird. Somatic variants were identified using population-level filtering strategies and cross-species genomic alignment to improve specificity.

We report genome-wide evidence of somatic mutations in hematopoietic cells, demonstrating that CH is not restricted to mammals and can be identified in birds. We further characterize somatic mutational spectra to infer underlying mutagenic processes, enabling distinction between aging-related mechanisms and exogenous influences such as ultraviolet radiation, pollution, or infection-associated inflammation. The strong population structure of Gentoo penguins also enables investigation of population-specific somatic mutation dynamics across ecological contexts. Together, this work proposes to leverage somatic mutation profiling as a novel approach for monitoring immune health and environmental stress in wild populations.

[A116] Sex-specific contemporary selection on educational attainment–associated genetic variation in Estonia

Ivan Kuznetsov[1], Dr Alexandros Giannelis[2], Dr Cornelius A. Rietveld[2], Dr Uku Vainik[1,3,4], dr. Vasili Pankratov[1,5]

[1]Institute of Genomics, University of Tartu, Tartu, Estonia, [2]Erasmus School of Economics, Erasmus University Rotterdam, Rotterdam, the Netherlands, [3]Institute of Psychology, University of Tartu, Tartu, Estonia, [4]Montreal Neurological Institute, McGill University, Montreal, Canada, [5]Bioinformatics Research Centre, Aarhus University, Aarhus, Denmark

In human populations, educational attainment (EA) has been repeatedly shown to be negatively associated with completed fertility, a commonly used proxy for fitness. Associations between the polygenic score for EA (EA PGS) and completed fertility are therefore consistent with ongoing selection acting on education-related genetic variation, although recent evidence suggests that these associations vary across social and demographic environments.

Using data from the Estonian Biobank on more than 45,000 individuals of both sexes born between 1920 and 1975, we examine sex- and environment-specific patterns of contemporary selection in Estonia as reflected in the association between the EA PGS and number of children ever born. We extend previous work by adjusting these relationships for another key fertility indicator, age at first pregnancy (AFP).

In Estonia, both EA and the EA PGS are negatively associated with fertility in women, whereas among men, EA is positively associated with fertility, and the EA PGS shows no significant association. Adjustment for EA reduces the association between the EA PGS and fertility in women by 24%, while adjustment for AFP eliminates the association entirely. Among women, the EA PGS shows significant interactions with EA and AFP. Family-based analyses are consistent with direct genetic effects of the EA PGS on fertility in women.

Overall, in contrast to some previous studies, we observe a negative association between the EA PGS and fertility only in women. This association is partly mediated and moderated by EA and AFP and appears to be largely attributable to direct genetic effects.

[A117] Selection on telomere homeostasis drives the adaptive evolution of TERT in Mimulus

Naseem Samo[1], Linh Nguyen[1], Surbhi Kumawat[1], Jae Young Choi[1]
[1]University Of Kansas, , United States

Cellular replication shortens chromosome ends and without intervention will result in catastrophic genomic instability. Hence, the telomerase maintains chromosome ends through Telomerase Reverse Transcriptase (TERT) protein that uses Telomerase RNA (TR) as a template. Recently we discovered a monkeyflower species (Mimulus lewisii) with duplicated TR genes and had a non-canonical sequence heterogeneous telomere consisting of 6 bp and 7 bp repeat DNA. Here, we investigated the molecular co-evolution between TERT and TR in M. lewisii. We used Nanopore sequencing to assemble new Mimulus species genomes and a clade-specific PAML analysis detected significant positive selection for TERT but only in lineages sharing the TR duplication. Functional basis of the positive selection was examined using yeast-three-hybrid assay testing physical binding between TERT and TR. Results showed M. lewisii TERT binds ancestral TR (TR1) and recently duplicated TR (TR2), but in sister species without the duplicated TR its TERT did not bind TR2. Inter-species domain swap experiments showed the casual region of binding both TR paralogs to two amino acids. Alphafold modeling indicated the residues were located within an intrinsically disorganized domain. Catalytic activity of the M. lewisii telomerase was examined by combining next-generation sequencing with the Telomeric-Repeat-Amplification-Protocol. Results showed M. lewisii TERT synthesized more nucleotide products using TR2 but each product had lower telomere repeat counts, suggesting selection on M. lewisii TERT involved TR paralog preference and telomerase processivity. Finally, we discovered TR2 paralog was associated with significantly longer telomeres, indicating the selection on TERT is ultimately related to telomere homeostasis.

[A118] Ancestral linkage group inference and centromere annotation elucidate rapid chromosomal evolution in the oligocentric sedges and rushes

James McCulloch[1], Dr Marcela Uliano-Silva[1], Dr Charlotte Wright[1], Professor Ian Henderson[2], Sam Ebdon[1], The Darwin Tree of Life Consortium[1], Dr Kamil Jaron[1], Mark Blaxter[1]
[1]Tree of Life, Wellcome Sanger Institute, Hinxton, United Kingdom, [2]Department of Plant Sciences, University of Cambridge, Cambridge, United Kingdom

Holocentric organisms – species which lack a single discrete monocentromere – are hypothesised to have higher rates of chromosomal fusions and fissions. This is because inter-chromosomal rearrangement in monocentrics typically results in chromosomes with two centromeres or none, interfering with meiosis. Holocentric species should not face this problem, but evidence that this facilitates rearrangement has been lacking. The plant families Cyperaceae (sedges) and Juncaceae (rushes) provide a new perspective; unlike “true” holocentrics, most have multiple discrete centromeres (“oligocentromeres”) on each chromosome. Using 44 chromosome-level genomes, syngraph, and CAP, we reconstructed the ancestral linkage groups of sedges and rushes, quantified their extraordinary rates of chromosomal fusion and fission, and annotated and investigated their centromeric repeats. Comparing rearrangement rates to oligocentromere annotations reveals that centromere organisation does correlate with inter-chromosomal rearrangement. We also found surprising patterns in the evolution of centromeric satellite sequences across the clade, including recognisable sequence conservation across tens of millions of years. There has been especially little centromere divergence between 13 species of Carex sedges, in stark contrast to the also-oligocentric Rhynchospora sedges. Furthermore, we identified two possible transitions in centromere organisation: first, putative monocentromeres in one species of the otherwise oligocentric Carex, which would be the first evidence for reversion to monocentricity in any eukaryote, and second, one case where no oligocentromeres could be identified, possibly a transition to “true” holocentricity. Together, this study demonstrates that sedges and rushes are a powerful system for linking centromere organisation to chromosomal rearrangement and genome evolution.

[A119] Streptophyte evolution: the evolutionary origin of the plant terrestrialization toolkit

Prof. Dr. Jan de Vries[1]
[1]University of Göttingen, Göttingen, Germany

Plant terrestrialization was one of the most transformative events in Earth's history. To understand what transpired during this event, my team and I use a comparative approach that integrates molecular data from land plants and their closest algal relatives. Phylogenomic and functional genomic advances in streptophyte algae have revealed that the genes underpinning an array of key traits that we ascribe to land plants were present in their algal progenitors. This applies to both the molecular networks for responding to environmental cues and multicellular morphogenetic processes. Using large-scale phylogenomic datasets, we reconstructed robust frameworks for major streptophyte algal lineages. These analyses reveal deep genetic divergences exceeding 830 million years, ancient origins of multicellularity, and repeated transitions between growth forms as well as aquatic and terrestrial habitats. Further, by performing integrative transcriptomic, genomic, physiological, and metabolomic analyses, we uncovered ancient molecular toolkits underlying stress perception, signaling, and growth regulation. Across multiple streptophyte lineages, stress responses to fluctuating light, temperature, and osmotic conditions are mediated by conserved hubs involving kinases, lipid signaling, cell wall remodeling enzymes, and phytohormone-related pathways. Co-expression network analyses demonstrate that environmental sensing and multicellular developmental programs have been tightly coupled for over 600 million years. Notably, complex morphogenetic processes appear to have been independently actualized from an ancestral toolkit rather than evolving once. Ancient genetic networks for stress resilience, signaling integration, and body plan complexity were repeatedly rewired and redeployed, ultimately enabling the conquest of land by embryophytes.

[A120] Nigritella or Gymnadenia? Resolving the phylogeny of a controversial clade

Jan Klink[1], Professor Mikael Hedrén[1], Professor Niklas Wahlberg[1]
[1]Lund University, Lund, Sweden

For a long time, Nigritella was treated as the sister genus to Gymnadenia (Orchidaceae). With its dense, almost globous inflorescence and non-resupinate flowers, Nigritella's morphology contrasts strongly with Gymnadenia's. However, since the rise of molecular methods, multiple studies have questioned the validity of Nigritella as a genus and rather positioned it within Gymnadenia. Predominantly ITS-data has been used as an argument to merge Nigritella with Gymnadenia. However, studies supporting the sister relation between the two genera also exist. Many orchids are being threatened by altered land use and the changing climate, and a clear morphological but primarily molecular delimitation is essential for conservation efforts. Here we used the Angiosperms353 probe kit to reconstruct the phylogeny of this orchid clade, aiming to resolve Nigritella's taxonomic position once and for all. Our study also includes a wide sampling of the polyploid Nigritellas that occur in the mountain regions of Europe. This sampling allows us to further investigate the, to this day largely unknown, origin of these taxa, ultimately giving us an insight into Nigritella's history on the European continent.

[A121] Functional characterization of casual variants and hitchhikers in recent pathogen-driven sweeps across diverse human groups

Jared Akers[1], Tian Xia[1], Madeline Pitre[1], Grace Oualline[1], Erin Gilbertson[1], Zhenting Wang[1], Stephen Rong[1], Kim Hie Lim[2], John Ray[3], Steven Reilly[1]
[1]Department of Genetics, Yale, New Haven, United States, [2]Singapore Centre for Environmental Life Sciences Engineering, Nanyang Technological University, Singapore, Singapore, [3]Center for Systems Immunology, Benaroya Research Institute, Seattle, United States

Evidence of past selective sweeps can be detected in the genomes of modern human groups, though the molecular mechanisms and phenotypic consequences of these prior adaptations remain largely unknown. While high linkage disequilibrium (LD) has enabled detection of sweep loci, the thousands of tightly linked non-selected variants (hitchhikers) within sweep loci hinder causal variant identification and reliable phenotypic associations. Moreover, most human genetic variation resides in non-coding regions, where interpreting the function of adaptive variants and hitchhikers remains difficult at scale.

To identify recent selective sweeps, we applied LD-based genomic scans to the 1000 Genomes Project (1KG) and the Genome Asia 100k pilot (GA100k) datasets. Using the integrated haplotype score and the number of segregating sites by length statistics, we identified 927 sweep loci across 20 non-recently admixed 1KG populations and 5 GA100k meta-populations. Tissue enrichment analyses revealed immune system regulatory elements were most enriched across 1KG continental groups sweeps, with lymphoid lineages being most enriched (BH p-value 0.0313).

To investigate the molecular mechanisms of these variants, we applied Massively Parallel Reporter Assays in primary T-cells with and without immune stimulus, as well as in T-cell, B-cell, and myelogenous immune cell lines. In total, we probed 11,907 putatively adaptive non-coding variants for functional consequence with 45,193 unique oligos. To uncover the biological processes impacted by adaptation, we linked these functional variants to their gene targets via published Activity-by-Contact models. Our approach reveals the molecular consequences of likely-causal variants and functional hitchhikers which resulted from recent pathogen-driven immune sweeps.

[A122] Breaking the yeast species barrier allows selection on recombinant hybrids

Jasmine Ono[1,2], Duncan Greig[2], G Ozan Bozdag[3]
[1]University of Nottingham, , United Kingdom, [2]University College London, , United Kingdom, [3]Georgia Institute of Technology, , United States of America

The yeast Saccharomyces cerevisiae and its closest relative S. paradoxus are reproductively isolated by hybrid sterility. The two species can readily form diploid hybrids but, during hybrid meiosis, recombination between their chromosomes is strongly inhibited and most of the resulting gametes are aneuploid and inviable. We removed this reproductive barrier by genetic manipulation and recovered recombinant, euploid, haploid hybrid gametes. This allowed us to investigate traits in the haploid hybrids that are masked in their diploid parent. We first focussed on speciation genes, new genetic combinations formed in hybrid genetic backgrounds that do not work well together. We found evidence for these intrinsic genetic incompatibilities between the nuclear genomes of the two species, supporting a role for speciation genes in hybrid sterility. We then investigated temperature adaptation in these hybrids, as temperature tolerance is the main trait known to phenotypically separate different Saccharomyces species. We find that we can detect selection for one species' alleles over the other depending on the temperature environment. These experiments demonstrate that genetically manipulated hybrids can be used to uncover the genetic basis of inter-specific traits.

[A123] Paragastric canal morphogenesis as a model for gut development in the ctenophore Mnemiopsis leidyi

Jason Presnell[1], Nicasio Jurani[1], Allie Graham[1]
[1]University Of Kansas, Lawrence, United States

Digestive systems display striking diversity in their size, shape, and function across animals, yet most of the developmental and evolutionary understanding comes mostly from a small number of bilaterian models. Ctenophores occupy a key phylogenetic position for insights into early animal evolution, and they possess a through-gut with an extensive gastrovascular canal system that differs markedly from digestive systems seen in other non-bilaterians. Thus, the ctenophore gut has the potential to be a valuable model for understanding the early evolution of animal digestive systems. Here, we propose that paragastric canal formation in the ctenophore Mnemiopsis leidyi is a tractable system to investigate gut morphogenesis outside of bilaterians. Through developmental staging we describe the post-embryonic morphogenetic events that shape the paragastric canals from their initial emergence from the embryonic gastrovascular cavity to their connection with the radial canals to form the mature continuous canal system. In addition, through bulk RNA sequencing we identify transcriptional changes associated with these processes and contextualize these data using pre-existing single-cell RNA-sequencing datasets. This has enabled us to identify potential gene candidates that define cell types within the paragastric canals. We are characterizing cellular features underlying paragastric canal growth using fluorescence-based labeling approaches. Together, this work provides a descriptive foundation for paragastric canal morphogenesis and further our overall understanding of ctenophore gut development, highlighting the potential of ctenophores as a comparative system for studying the evolution and development of animal digestive systems.

[A124] Maternal effects and the expression of DNA methyltransferases in Daphnia pulex

Trenton Agrelius[2], Jeff Dudycha[1]
[1]University Of South Carolina, Columbia, United States, [2]University of Notre Dame, South Bend, United States

Maternal environments can strongly affect offspring phenotypes, altering morphology, gene expression, and life history. Epigenetic mechanisms, particularly DNA methylation, provide potential pathways for transferring maternal environmental information to offspring. However, few studies have examined variation in epigenetic mechanisms' responses to maternal effects. Using the crustacean Daphnia, we examined how maternal resource environments influenced offspring DNA methyltransferase (DNMT) expression.

We found striking clone-specific differences in how resource limitation affected DNMT mRNA expression. In one clone, maternal resource limitation elevated expression of three DNMT classes; in the other, only DNMT1 increased. In both clones, DNMT expression of offspring was sensitive to maternal environment but responded in opposite directions. For both life history traits and DNMT expression, maternal environmental effects were stronger when offspring experienced limited resources compared to abundant resources.

To facilitate future investigations, we developed RNAi constructs targeting three DNMT loci. In one clone, RNAi attempts were lethal. In the other, RNAi reduced DNMT expression relative to GFP controls but produced strong cross-reactivity; targeting one DNMT reduced expression of the other two. This cross-reactivity may reflect known immune pathways involving signal transduction stimulated by viral or bacterial signals, or it may reveal previously unknown aspects of DNMT biology.

Our results demonstrate genotype-specific epigenetic responses to maternal resource environments and unexpected regulatory interactions among DNMT genes in Daphnia, highlighting the complexity of epigenetic maternal effects and the need for further mechanistic investigation.

[A125] (What) Can isopod genomes tell us about their ecological transitions?

Jessica A. Thomas Thorpe[1]
[1]Wellcome Sanger Institute, Wellcome Genome Campus, Hinxton, Cambridge, United Kingdom
Ecological transitions – evolutionary shifts between ecosystems or ecological niches – have played a key role in shaping Earth's biodiversity. The crustacean order Isopoda provides an excellent study-system for ecological transition, with species occupying almost every environment on Earth. These include terrestrial woodlice, littoral sea-slaters, freshwater pond-lice and many marine species, including the giant deep-sea isopod and notorious 'tongue-biter' fish parasite. Notably, isopods have undergone multiple independent transitions (and reversals) between marine, freshwater, terrestrial and host-associated habitats, as well as shifts in feeding ecology, throughout their evolutionary history. Studying isopod genomes therefore offers a unique opportunity to uncover the genomic basis of ecological adaptation across many different ecosystems. However, until recently, this opportunity has remained out of reach. As for many crustaceans, generating chromosome-level isopod genomes has proven challenging, with factors such as large genome size, high repeat content and contamination complicating efforts. The Tree of Life Programme in the Wellcome Sanger Institute, UK, has recently overcome these obstacles, producing the first 10 high-quality isopod reference genomes assembled to chromosome-level. These include species from isopod suborders Asellota (freshwater pond-lice and marine isopods) and Cymothooidea (fish parasites and scavengers) making it possible to undertake the first comparative genomic analyses of ecological transition to freshwater and parasitism in isopods. Initial results reveal dynamic genome evolution across Isopoda, with considerable variation in genome size, chromosome number and repeat content between closely related species, as well as extensive genome rearrangement.

[A126] Single-nucleus multi-omics analyses reveal cellular and molecular innovations in the anterior cingulate cortex during primate evolution

Jiamiao Yuan

The anterior cingulate cortex (ACC) of the brain involves higher-level cognitive functions such as emotion and self-awareness, and is closely related to human mental disorders. The ACC of Old World anthropoids contains von Economo neurons (VENs), one of the noticeable cellular innovations during primate brain evolution. Here, we present the profiles of human and macaque ACC gene expression and chromatin accessibility at single-nucleus resolution. We characterized the conserved gene expression, chromatin accessibility, and transcription factor binding patterns in different cell types and neuronal subtypes. Importantly, by combining published mouse data, we discovered the molecular identities and the possible cell lineage origin of primate VENs. In particular, with both in vitro and in vivo experimental validations, we identified a group of human–macaque shared and human-specific VEN marker genes, such as PCSK6, ADAMTSL3, and CDHR3, which may contribute to the morphogenesis and function of VENs. Furthermore, we established connections between genome-wide human-specific mutations and specialized gene regulation in the human ACC, delineating the genetic basis of cellular and functional innovations in the ACC during primate evolution and human origin. These findings provide new insights into the cellular composition and molecular regulation of the ACC and its evolutionary role in shaping higher cognitive functions in humans.

[A127] Admixture History and Tropical Adaptation of the Cambodian Indigenous Peoples

Jianxin Guo[1], Bing Su
[1]National Key Laboratory of Genetic Evolution and Animal model, Kunming Institute of Zoology, Chinese Academy of Sciences, , China
Abstract: Cambodia, located at the center of mainland Southeast Asia, is a key region for understanding early human settlement, language spread, and environmental adaptation. We analyzed genome-wide data from 14 Cambodian indigenous groups, including 12 Austroasiatic, 1 Kra-Dai, and 1 Austronesian. Overall, Cambodian populations show low genetic diversity, slow LD decay, enriched long runs of homozygosity, and persistently small effective population sizes after the Out-of-Africa bottleneck. While lowland Khmer and Lao groups experienced moderate expansion, highland groups remained more isolated and demographically constrained. Genetic differentiation is low among Western Bahnaric highland groups but clear between highland and lowland populations. Importantly, genetic structure does not match linguistic classification: Cambodian groups form a continuous genetic gradient rather than clustering by language family, supporting a model of cultural diffusion instead of large-scale migration. Western Bahnaric groups retain high levels of Andamanese-related ancestry, preserving some of the oldest genetic components in Southeast Asia. Admixture and allele-sharing analyses show that Cambodians carry more Hoabinhian-related and early Southeast Asian ancestry and experienced weaker East Asian gene flow than neighboring populations. In particular, Highland groups show closer affinity to ancient Southeast Asian individuals, indicating deep regional continuity. Additionally, we identified 56 positively selected regions involving genes related to growth, metabolism, immunity, skin function, and reproduction, reflecting adaptation to tropical rainforest environments with high heat, humidity, and pathogen burden.

[A128] Inferences of haplotype sharing and population history across time

Jiaqi Yang[1], Benjamin Peter[1]
[1]Max Planck Institute for Evolutionary Anthropology, , Germany
During the dispersals across Eurasia, modern humans experienced numerous population admixture events. The most well-known ones are probably interactions with Neanderthals and Denisovans, two now-extinct homo lineages that diverged from modern humans as early as 800,000 years ago. Multiple modern human lineages also diverged from one another but later admixed, shaping the genetic landscape of present-day populations. While the summary statistics (e.g., f3/f4) can indicate general relationships between groups, recent method advances have enabled local ancestry inferences (LAIs), which predict the source of a haplotype in an admixed genome. Those inferences could reveal more aspects of admixture, such as selection, timing, and pulses.

In this work, we perform LAIs using both present-day high-coverage genomes and low-coverage ancient genomes, and for ancient genomes, we also generate genotypes by imputation. Using downsampling tests, we demonstrate both the promise and limits of LAIs. We then focus on haplotypes from archaic humans and from deeply diverged modern human groups. We investigate the sharing of those haplotypes across diverse populations. We show that those haplotypes are informative of the common ancestral populations, thus providing insights into past population admixture history.

[A129] Conserved Myosin Anchoring, Divergent C Termini: Structural Bases of Flightin/Paraflightin Functional Evolution in Pancrustacea

Jim Vigoreaux[1], Emily Price[1]
[1]University Of Vermont, Burlington, United States

Pancrustacea is the most species-rich animal clade having successfully colonized aquatic and terrestrial environments. This ecological breadth is enabled in part by the diversification of muscle-driven appendages for locomotion, feeding, defense, and reproductive behaviors. While core sarcomeric proteins are highly conserved, clade-restricted myofibrillar proteins influence myofilament architecture and mechanics to meet specific performance demands. We examined the pancrustacean myosin-binding protein flightin, which in Drosophila is expressed in indirect flight muscle and is required for normal thick-filament length and flexural rigidity, properties critical for high-frequency, high-power contraction. Flightin is defined by a novel WYR domain that anchors to the myosin rod. In crustaceans, we identified a distinct paralog, paraflightin, in which the WYR-like region (paraWYR) differs in sequence and is followed by an extended C-terminal domain. Molecular modeling indicates that paraWYR retains key amino acids predicted to contact the myosin rod, supporting a conserved anchoring function. However, increased domain flexibility and the divergent, lengthened C terminal domain suggest additional, lineage-specific roles in regulating filament length and bending propensity, parameters previously implicated for the flightin C-terminal domain in insects. Together, these results support a model in which WYR-mediated myosin anchoring is conserved across Pancrustacea, while paralog- and domain-level innovations expand the mechanical design space of thick filaments, contributing to muscle-type diversification and the evolution of muscle-driven behaviors.

[A130] Population Genomics of the Asian Corn Borer (Ostrinia furnacalis) in Korea

Jiyeong Shin[1], Hajoung Lee[1], Dr Choongwon Jeong[1,2]
[1]Institute for Data Innovation in Science (IDIS), Seoul National University, Seoul, South Korea, [2]School of Biological Sciences, Seoul National University, Seoul, South Korea

The Asian corn borer (ACB), Ostrinia furnacalis, is a major lepidopteran pest of maize across Asia and Oceania and remains a persistent threat to crop productivity and regional food security. In Korea, ACB outbreaks can cause substantial yield losses and increase reliance on chemical control, yet its population genetic structure and patterns of genomic differentiation within the country are still insufficiently characterized. Establishing a population genomic baseline is critical for interpreting future changes in pest dynamics, including shifts in distribution and management outcomes under ongoing agricultural intensification and environmental change.

Here, we conducted a population genomic analysis of Korean O. furnacalis using a single-nucleotide polymorphism (SNP) dataset. We focused on resolving population structure and genetic differentiation among sampling localities. Principal component analysis (PCA) based on the SNP set revealed clear genetic structuring among Korean populations, including a distinct cluster separated along PC1 and additional differentiation captured by PC2. Comparative inclusion of closely related Ostrinia taxa further supported species-level separation and provided phylogenetic context for interpreting within-species variation.

Overall, our results demonstrate that Korean ACB populations are not genetically homogeneous and instead exhibit measurable regional structure. These findings provide foundational genomic resources for future work, including monitoring gene flow, detecting potential introductions, and establishing reference baselines for investigating adaptive responses (e.g., to host use or control pressures) as additional ecological and phenotypic data become available.

[A131] Exploring mitonuclear coadaptation in a Carpathian contact zone of cryptic alpine newt subspecies

Joanna Jakóbik[1], Zuzanna Bujak[1], Wiesław Babik[2], Katarzyna Dudek[2], Dan Cogălniceanu[3], Maciej Pabijan[1]

[1]Department of Comparative Anatomy, Institute of Zoology and Biomedical Research, Faculty of Biology, Jagiellonian University, Kraków, Poland, [2]Institute of Environmental Sciences, Faculty of Biology, Jagiellonian University, Kraków, Poland, [3]Faculty of Natural Sciences and Agricultural Sciences, Ovidius University of Constanţa, Constanţa, Romania

Nuclear genes involved in oxidative phosphorylation are known to co-evolve with mitochondrial genes to compensate for rapid changes in mtDNA and thus constitute an arena of potential genetic conflict between divergent lineages. We studied two cryptic alpine newt lineages, Mesotriton alpestris alpestris and M. a. reiseri, that admix at a contact zone between the eastern and southern Carpathians. Analysis of nucleotide divergence of transcriptomic data for 13 mitochondrial genes and 453 nuclear genes showed a significantly higher value for mtDNA-encoded genes (mean = 0.08, $p < 0.001$), while for nuclear markers it was lower by an order of magnitude. We observed a higher dN/dS ratio for nuclear genes interacting with mtDNA (mean = 0.38, $p = 0.003$) in comparison to non-interacting genes (mean = 0.29), which supports predictions of mitonuclear coadaptation. We genotyped and mitotyped 709 individuals and found a steep and narrow transition in mtDNA (~20 km width) located in the southern Carpathians. In contrast, we found a broad transition (~150 km) at 42 diagnostic nuclear genes suggestive of a wide hybrid zone. Extensive nuclear gene flow in contrast to the steep mitochondrial cline suggest that overall, mitonuclear mismatch in admixed individuals has a limited effect on reproductive isolation. Only genes involved in mitochondrial transcription show the hallmarks of high differentiation and reduced gene flow between the two lineages, concordant with the pattern for mtDNA. Our results refute the distinct status of the southern Romanian alpine newt lineage, suggesting fusion after secondary contact with the widespread nominate lineage.

[A132] Cell culture-based approach uncovers the role of environment, evolutionary history and introgression in phenotypic plasticity in fish

João Moreno[1,2], Catarina Santos[1], Mariana Moço[1], Romana Santos[2], Gabriela Rodrigues[1], Vítor C Sousa[1]

[1]cE3c – Centre for Ecology, Evolution and Environmental Changes & CHANGE – Global Change and Sustainability Institute, Faculdade de Ciências, Universidade de Lisboa, Lisboa, Portugal, [2]MARE – Centro de Ciências do Mar e do Ambiente (MARE) & ARNET—Aquatic Research Network, Faculdade de Ciências, Universidade de Lisboa, Lisboa, Portugal

Climate change is intensifying thermal stress in freshwater ecosystems, threatening ectothermic organisms with limited dispersal capacity. Phenotypic plasticity, particularly at the molecular level, is critical for short-term responses. Yet, the influence of past evolutionary history, environmental background, and introgression on molecular phenotypic plasticity (MPP) remains poorly understood. The Iberian chubs from the genus Squalius, distributed along a latitudinal gradient with increasing temperature from North to South with evidence of both local adaptation and introgression, provide an ideal system to explore these effects. Using a novel fibroblast cell culture approach, we exposed cells derived from the same individual (i.e., same genotype) to different temperature, quantifying MPP in gene expression at both individual and population levels. Overall, we observed patterns of inter-individual heterogeneity in all populations, with most genes displaying modest changes between conditions. Northern populations [Pego (S. carolitertii)] from colder environments revealed pronounced downregulation, while southern populations [Alcobertas (S. pyrenaicus)] from warmer environments showed predominantly upregulation, revealing the influence of the environmental background of the populations on MPP. Individuals from the introgressed population [Ceira (S. carolitertii)] exhibited the highest number of differentially expressed genes, but also the highest levels of inter-individual variation in plasticity, consistent with possible regulatory disruption due to the admixture of different genomes. These results demonstrate that MPP varies within and among populations and that environmental history, genomic background, and introgression can reshape transcriptional responses. Moreover, the cell culture approach provides a valuable framework to assess mechanisms behind MPP in non-model and endangered species.

[A133] Genomic signatures of virulence and resistance to psittacine beak and feather disease virus (BFDV) in the echo parakeet (Alexandrinus eques)

Dr Johanna Winder[1], Dr Lucy Pecasse[1], Dr Rangsinee Sankhom[1], Simon Tollington[3], Dr Hernán Morales[4], Cock Van Oosterhout[2], Prof Jim Groombridge[1]
[1]Durrell Institute of Conservation and Ecology, University of Kent, Canterbury, United Kingdom, [2]School of Environmental Sciences, University of East Anglia, Norwich, United Kingdom, [3]Durrell Wildlife Conservation Trust, Trinity, Channel Islands, [4]Globe Institute, University of Copenhagen, Copenhagen, Denmark

Circoviruses such as psittacine beak and feather disease virus (BFDV) affect more than 100 species of birds including pigeons, chickens and parrots. Infection poses a significant risk to vulnerable species and threatens spillover to non-avian species. The echo parakeet (Alexandrinus eques) is a species endemic to Mauritius, which has experienced numerous BFDV outbreaks since 2005. An extreme population bottleneck in the 1980s followed by intensive conservation management and continuous monitoring has led to a unique study system with observations and samples for almost every individual in the population, including whole genome sequencing, disease screening (PCR), viral genome sequencing, and extensive phenotypic data. Here, we examine the viral (BFDV) and host (echo parakeet) genomes of infected individuals to identify how infection interacts with host inbreeding and reproductive recruitment, and examine potential genomic features of host immunity. Viral haplotypes showed temporal changes associated with BFDV outbreaks, with increased virulence in more recent outbreaks and subsequently lower recruitment and higher inbreeding. Infection with BFDV reduces recruitment of many (susceptible) individuals, however some infected birds recover and re-enter the breeding population (tolerant), and others avoid infection altogether (resistant). Phenotypic observations and PCR screening were used to classify individuals into these three (R/T/S) categories. Genomes were compared between R/T/S groups to identify candidate resistance loci. Our results highlight the vulnerability of post-bottleneck populations to emerging infection, and identify candidates for genomics-informed conservation management of echo parakeets and other psittacine species.

[A134] Early genomic signatures of domestication in the common ostrich

Johanne Gottenborg[1]
[1]Aarhus University, Aarhus, Denmark

The domestication of animals has profoundly impacted the biology of species, the ecosystems they inhabit, and has led to turning points in human history. Examining the genomic process of early domestication is important to understand: 1) How human activity interacts with species evolution, with or without intend, and 2) which evolutionary forces drive the divergence between domesticated taxa and their wild relatives. However, little is known about the adaptations that occur during the initial phase of domestication, as most animals were domesticated thousands of years ago. The common ostrich (Struthio camelus) has been captured across Africa and the Middle East for centuries, but has only been farmed for the last 150 years, mainly within South Africa. During the time that ostriches have been farmed they have been selected for feather, meat and leather traits. As a results, they provide an example of the early phase of the domestication process, yet the genomic signatures hereof remain unexplored. Here, we compare genomic data of 400 captive ostrich individuals representing the farmed populations from South Africa to wild individuals from all four extant ostrich species and subspecies. Through analyses of demographic history, population structure, and selection scans, we determine the genetic origin of farmed ostriches and detect genomic signatures of the evolutionary forces, both neutral and deterministic, driving the process of domestication.

[A135] Gene expression evolution across primate cardiomyocyte differentiation

John Hurley[1], Alyssa Bogar[2], Jose Gutierrez[2], Dr. Michelle Ward[2]
[1]Biochemistry, Cellular, and Molecular Biology Graduate Program, UTMB, Galveston, United States, [2]Department of Biochemistry and Molecular Biology, UTMB, Galveston, United States

The hourglass model of development posits a phylotypic stage in embryonic development where conservation of morphology and gene expression is observed across diverse vertebrate species. This model has not been investigated across primates for ethical and technical reasons. With the advent of induced pluripotent stem cell (iPSC) technology, developmental processes can now be modelled across humans and chimpanzees. To determine the level of conservation of gene expression across primate development, we differentiated iPSCs from seven humans and seven chimpanzees into cardiomyocytes. We collected cells at five differentiation stages: iPSCs (Day 0), mesoderm (Day 2), cardiac mesoderm (Day 5), early cardiomyocytes (Day 15) and late cardiomyocytes (Day 30) to profile gene expression. Stage-specific protein marker expression is highest in the corresponding stage for both species, and there is no difference in the proportion of cells representing each stage across species. Global gene expression data across 70 samples identifies differentiation stage as the largest contributor to expression variation, followed by species. Jointly modeling the data reveals 11 gene expression trajectories that are largely conserved across species. When considering each differentiation day independently, we identify thousands of differentially expressed genes between species with the fewest observed at the mesoderm and cardiac mesoderm stages. These stages also show the lowest absolute species effect sizes. Pairwise correlations of effect sizes between temporally-adjacent days show highest inter-species correlations between days 2 and 5 and days 5 and 15. Our primate data are consistent with the hourglass model of development with most inter-species conservation at mid-developmental stages.

[A136] Automation-driven Micro-Scale Spatial Metagenomics

Jonas Lauritsen[1], Carlotta Pietroni[1], Bryan Wang[2], Zoé Horisberger[2], Nanna Gaun[1], Jorge Langa[1,3], Iñaki Odriozola[1], Urvish Trivedi[2,4], Antton Alberdi[1], Amalia Bogri[1]
[1]Centre for Evolutionary Hologenomics, Globe Institute, University of Copenhagen, , Denmark, [2]Section of Microbiology, Department of Biology, Faculty of Science, University of Copenhagen, , Denmark, [3]Department of Genetics, Animal Physiology & Physical Anthropology, University of the Basque Country, , Spain, [4]COPSAC, Copenhagen Prospective Studies on Asthma in Childhood, Copenhagen University Hospital, Herlev-Gentofte Hospital, Gentofte, Denmark

Hologenomic research is rapidly reshaping our understanding of microbiome–host interactions across environmental, animal, and human systems by integrating host and microbial genomic analysis. Current hologenomic approaches predominantly rely on metagenomic sequencing of bulk samples, without accounting for spatial resolution, which potentially obscures critical microscale interactions among microbial community members.

To address this, we developed a novel methodology, Micro-Scale Spatial Metagenomics (MSSM), which enables spatially resolved metagenomic analysis of the animal intestine while preserving coordinate metadata on tissue architecture. To achieve high spatial resolution, our method requires a high number of microsamples per intestinal section. To enable such high-throughput data generation, we integrated laboratory automation. We programmed a Tecan DreamPrep 780 liquid handler for DNA library preparation of low-input metagenomic microsamples (pg of DNA), working with volumes as low as 2.5 µL.

The automated workflow improved time efficiency, reduced cost, and increased sustainability, while maintaining a high level of quality to ensure high-calibre research. As such, it provides an important infrastructure for MSSM to uncover fine-scale biological patterns instrumental in understanding complex hologenomic systems. We have implemented MSSM to investigate multiple livestock animal model systems and their gut microbiome dynamics under pathogenic disturbance and dietary modulation, to support sustainable production.

[A137] On the limits of introgression: the early out of Africa conundrum

Jose Miguel Serradell Noguera[1], Alba Nieto Heredia[2], Jaume Bertranpetit i Busquets[1], Oscar Lao Grueso[1]
[1]Institut Biologia Evolutiva - UPF-CSIC, Barcelona, Spain, [2]iTHEMS RIKEN, Wako, Japan

The global expansion of Homo sapiens out of Africa approximately 50,000–60,000 years ago (kya) represents a defining event in human evolutionary history. However, fossil evidence from the Levant, East Asia, and Southeast Asia dated between ~70 and 210 kya raise questions about the potential genetic contribution of an early Out-of-Africa (eOOA) expansion to present-day populations. Current genomic models largely support a near-complete replacement of these early groups by the later expansion, yet they rely on the detectability of introgressed ancestry—either ancient DNA or strong signals of admixture, both of which are absent for eOOA events.

Here, we evaluate whether genetic traces of an eOOA remain detectable in present-day Asian genomes by comparing alternative demographic scenarios using an Approximate Bayesian Computation framework coupled with Deep Learning. Our results indicate that models without eOOA introgression provide the best fit to the data. To explore the limits of detectability, we explored the distribution of fragments in the genome under coalescent events. While long fragments associated with deep Time of the Most Recent Common Ancestors are theoretically expected under introgression, we find that as population divergence times approach ~100 kya, introgression signals rapidly decay and become indistinguishable from background coalescent variation.

These findings suggest that the absence of detectable eOOA introgression may reflect methodological limitations rather than true absence of ancestry, potentially explaining why Neanderthal and Denisovan introgression is detectable while eOOA signals are not, highlighting the need for novel summary statistics specifically tailored to detect introgression in this temporal regime.

[A138] Mode and tempo of de novo gene birth in the brown alga Ectocarpus

PhD Josué Barrera-Redondo[1,2], Jia Xuan Leong[1], Dr. Chris Papadopoulos[3], Dr. Catia Igreja[4], Piero Foncerrada-Elizondo[2], Dr. Rémy Luthringer[1], Dr. Elena Avdievich[1], Agnes Henschen[1], Dr. Fabian B. Haas[1], Dr. M. Mar Albà[3], PhD Susana M. Coelho[1]

[1]Department of Algal Development and Evolution, Max Planck Institute for Biology, Tübingen, Germany, [2]Departamento de Biotecnología y Bioquímica, Centro de Investigación y de Estudios Avanzados del IPN, Irapuato, Mexico, [3]Evolutionary Genomics Group, Hospital del Mar Medical Research Institute, Barcelona, Spain, [4]Department of Integrative Evolutionary Biology, Max Planck Institute for Biology, Tübingen, Germany

Brown algae (Phaeophyceae) are a group of complex multicellular eukaryotes that belong to the supergroup Stramenopila, which diverged from plants, animals and fungi over a billion years ago. Thus, brown algae serve as an independent “evolutionary experiment” to understand the emergence of morphological and molecular novelty. Comparative genomics across the tree of life shows that one quarter of the protein-coding genes in brown algae are found exclusively within Phaeophyceae, with no traceable homologs in other known organisms. We focused on the emergent model species Ectocarpus sp. 7 to trace back the evolutionary origin of recently-evolved brown algal orphan genes. We analyzed the genomes and transcriptomes of eleven species within the Ectocarpus genus and included five species within the order Ectocarpales as outgroups. A combination of genomic phylostratigraphy, synteny-based methods, RNA-seq and Ribo-seq evidence allowed us to detect 734 genes within the Ectocarpus sp. 7 genome with putative signatures of de novo birth from noncoding DNA over the last 20 million years of evolution. We estimated the rate of de novo gene conservation at each node in the phylogeny and evaluated the chromatin context around these genes using histone mark ChIP-seq and Hi-C data. Our study shows a significant enrichment of de novo genes in the sex chromosomes of Ectocarpus, suggesting that they act as cradles for new genes to evolve from scratch. We performed population-level Illumina and ONT genome sequencing within Ectocarpus sp. 7 to uncover the single-nucleotide and structural variants that govern the intra-species dynamics of de novo genes.

[A139] Host associations of Borrelia afzelii ospC and ospE variants in two wild rodent reservoir species

Józefina Wasilewska[1], Przemysław Gawrysiak[1], Jacek Radwan[1]
[1]Adam Mickiewicz University , Poznań, Poland

In the Borrelia burgdorferi sensu lato complex, the ospC and ospE genes encode Outer Surface Proteins that play critical roles in evading host immunity. Both loci are highly polymorphic, yet the mechanisms maintaining this diversity in natural systems remain unresolved. One of the leading hypotheses is multiple niche polymorphism, whereby different reservoir host species represent distinct selective environments favoring specific variants. Empirical support for this model is mixed regarding ospC, while there is currently no direct evidence that the multiple niche polymorphism hypothesis has been tested for ospE.

To test the multiple niche polymorphism hypothesis for B. afzelii, the dominant rodent-associated Lyme borreliosis agent in Europe, we sampled its two key reservoir hosts, the yellow-necked mouse (Apodemus flavicollis) and the bank vole (Myodes glareolus), within a 10-ha field site during the main tick season (April-September). B. afzelii ospC and ospE sequence variants were identified using amplicon sequencing (Illumina MiSeq) to test whether variant occurrence and overall composition differed between host species at either locus. Using this dataset, we evaluate the extent to which host species contribute to the maintenance of outer surface protein polymorphism in B. afzelii while explicitly addressing the current evidence gap regarding ospE.

[A140] Evolutionary history and functional divergence of hydroxycarboxylic acid receptors in primates

Juan C. Opazo[1], Felipe Barros[1], Kattina Zavala[1], Rodrigo Maldonado, Gonzalo Mardones
[1]Universidad San Sebastian, Valdivia, Chile

Hydroxycarboxylic acid receptors are class A G-protein-coupled receptors that act as metabolic sensors linking cellular metabolic status to physiological and immune responses. This family comprises three members (HCAR1, HCAR2, and HCAR3), expressed in metabolically active tissues and immune cells. Here, we investigate the evolutionary history of the most recently derived members, HCAR2 and HCAR3, in primates. Phylogenetic analyses reveal a complex duplicative history involving multiple independent duplication events during ape evolution. As a result, most ape lineages harbor independently originated duplicated copies, whereas non-ape primates retain the ancestral single-copy condition (HCAR2/3). Functional inference indicates that this single-copy gene is equivalent to HCAR2, suggesting that the major functional innovation in apes is associated with the emergence of HCAR3. Gene expression analyses further show that major divergence in tissue expression occurred early, following the duplication that generated HCAR1 and the HCAR2/3 lineage, while HCAR2 and HCAR3 retain largely overlapping expression profiles. Together, these results indicate that refined, context-dependent lipid sensing mediated by HCAR3 evolved repeatedly in apes.

[A141] Differentiating Migration Rates Using Selective Sites Instead of Neutral Sites

Jui-hsuan Chien[1], Professor Hsiao-Han Chang[1]
[1]Institute Of Bioinformatics And Structural Biology, National Tsing Hua University, Hsinchu, Taiwan
Differentiating levels of pathogen migration rates between locations is crucial for identifying infection sources and targets for disease control. Traditionally, researchers have used neutral genetic markers to infer demographic history and migration rates among populations. However, at the within-country level, crucial for implementing disease control strategies, high migration rates lead to very low genetic differentiation, making it difficult to distinguish between various levels of migration. In previous research, we observed that drug resistance sites, under strong positive selection, offered better differentiation of migration rates than neutral sites, which showed no significant signal. To systematically explore the power of selective sites in differentiating migration rates, we utilized the evolution simulator SLiM to construct a model comprising multiple populations interconnected by varying migration rates. We introduced both neutral and selective mutations into these populations and analyzed three statistics: F_ST, sequence similarity between populations, and the proportion of nearly identical sequences between populations. The results showed that, except for F_ST, statistics based on selective sites more accurately identified the highest migration rates than neutral sites, with the extent of improvement depending on the specific migration scale and allele frequencies. Similarly, selective sites yielded better clustering of samples based on pairwise sequence similarity. Our study demonstrates that selective sites have the potential to provide valuable insights into demographic history and suggests that they should not be routinely excluded from the demographic analyses.

[A142] Transcriptomic dynamics of eastern oysters under climate change stress

Julia McDonough[1], Sophia Montague[2], Dr. Thomas Miller[3], Dr. Sarah Donelan[2], Sarah Gignoux-Wolfsohn[1]
[1]University Of Massachusetts Lowell, Lowell, United States, [2]University of Massachusetts Dartmouth, New Bedford, United States, [3]University of Maryland Center for Environmental Science, Solomons, United States

Anthropogenic change is rapidly altering environments, exposing organisms to greater variability than they have historically experienced. Oysters are especially challenged by climate change due to their life history (sessile nature and relatively long lifespan), making it likely they will experience numerous environmental changes within their lifetime. It is therefore critical to understand how early-life experiences will affect oysters' response in the future (referred to as carryover effects) to better prepare for a changing environment. Phenotypic carryover effects have been observed in the tissue and shell growth of oysters; however, the mechanisms underlying these changes remain unknown. Transcriptomics enables assessment of these effects at a molecular level, including responses that may not be detectable in the phenotype. In a fully factorial experiment, we exposed juvenile eastern oysters (Crassostrea virginica) to two consecutive 2 week phases of water treatments with manipulated dissolved oxygen (control or hypoxia, <2 mg/L) and temperature (ambient or warming, +3°C) to simulate climate change predictions under the IPCC 2019 RCP8.5 scenario. Oysters were sacrificed after each exposure and gill tissues were sampled and processed for RNA-seq. We identified differentially expressed genes associated with both general and stressor-specific responses, as well as temporal patterns of transcription. Additionally, we identified transcriptionally frontloaded genes that may serve as targets for priming-induced resilience. These results reveal how early-life stress exposure shapes transcriptomic responses in later environments, potentially promoting adaptive responses and phenotypic carryover effects.

[A143] A Curated Genome-Scale Nucleotide Diversity Panel of Non-Human Primates

Juraj Bergman[1], dr. Vasili Pankratov, Mr Bjarke Meyer Pedersen, Erik Fogh Sørensen, Kasper Munch, Pr Thomas Bataillon, Professor Mikkel Heide Schierup
[1]Aarhus University, , Denmark

Primates constitute one of the most phylogenetically and ecologically diverse Eutherian mammalian orders, with a central role in advancing our knowledge of human evolution, speciation processes and conservation biology. While thousands of whole-genome sequences have been generated across a multitude of primate taxa, discrepancies in data processing - particularly the lack of ploidy-aware variant calling in sex-linked regions - have limited the utility of existing datasets for large-scale comparative analyses. We utilized publicly available short-read sequencing data of non-human primates, recently published primate genome assemblies and a ploidy-aware variant calling procedure to generate a genome-scale nucleotide diversity panel comprising 3,240 individuals from 269 species and 71 genera. To further facilitate cross-species comparisons, we generated a multiple-genome alignment of the 48 primate assemblies used for variant calling. This curated resource of non-human primate diversity provides a foundation for future research in primate evolutionary biology, speciation, and sex chromosome evolution.

[A144] The molecular regulation of floral ultraviolet patterns in sunflower

Justin Chan[1,3], Ms. Ara Jamaldin[1,2], Dr. Marco Todesco[1,2,3]
[1]Micheal Smith Laboratories, The University of British Columbia, Vancouver, CA, [2]Department of Botany and Biodiversity Research Centre, The University of British Columbia, Vancouver, CA, [3]Department of Biology, The University of British Columbia, Vancouver, CA

Understanding the molecular mechanisms controlling plant adaptation is a fundamental goal in evolutionary biology. Recent work has shown that ultraviolet (UV)-absorbing patterns in sunflower ligules —the “petals” of sunflower inflorescences— have two adaptive functions, acting as conspicuous visual signals to attract pollinators and affecting ligule desiccation rates. Sequence and expression variation of the sunflower transcription factor HaMYB111 is associated with the accumulation of UV-absorbing flavonoids, suggesting that it controls UV-pattern variation by regulating genes involved in flavonoid biosynthesis. However, we currently lack a detailed understanding of the regulatory networks that control, and are affected by, HaMYB111 expression in sunflower. For example, the Arabidopsis MYB111 homolog is post-transcriptionally regulated by microRNA858 (miR858). While miR858, and its putative target site, are conserved in sunflower, it is not known whether HaMYB111 is regulated in a similar manner.

To characterize the regulatory networks controlling UV patterning in sunflower ligules, we developed introgression sunflower lines with intermediate UV-patterns and sequenced mRNAs and small-RNAs from the basal UV-absorbing and distal UV-reflecting regions of ligules during early developmental stages, when HaMYB111 is highly expressed. We then compared the expression of genes and miRNAs between these regions to identify key genes and pathways that are differentially regulated. This research contributes to our understanding of the transcriptional networks controlling a widespread adaptive trait, floral UV patterning.

[A145] Rapid ecological diversification of Arabidopsis thaliana populations in urban environments

Justine Floret[1], Prof. Dr. Anja Linstädter[2], Prof. Dr. Stanislav Kopriva[1], Prof Juliette De Meaux[1]
[1]University of Cologne, Cologne, Germany, [2]University of Postdam, Postdam, Germany

With the human population expected to exceed 10 billion by 2050, urban expansion is placing increasing pressure on natural ecosystems and biodiversity. Yet cities are designed as replicated mosaics of green and grey spaces, offering a unique opportunity to test whether ecological heterogeneity can promote the maintenance of genetic diversity.

Indeed, our understanding of the ecological processes driving genetic adaptation remains limited, constraining our ability to predict and mitigate the impacts of climate change. To address this gap, we focus on observing adaptation in action within natural urban habitats.

We designed an experiment to dissect the processes driving these patterns by crossing two well-characterized A. thaliana ecotypes from ecologically distinct spontaneous populations in Cologne (Germany). The offspring population harbors genetic diversity in flowering time, seed dormancy, and fruit production. We seeded 500 habitat patches across the city, each characterized by variation in disturbance, light, and soil composition. We subsequently monitored establishment, and assessed the genetic and genomic diversity of established populations.

Established populations differed in life-history traits, displaying genetic diversity in flowering time, life-cycle length, and dormancy requirement. The experiment demonstrates that a complex millefeuille of interacting ecological filters shapes both the limits of population establishment and the distribution of genomic variation. Several abiotic factors consistently limited establishment, but these limits were contained both by facilitating effects in harsh environments and local selection of adaptive variants in the genome.

Together, these results showcase how urban areas can function as open-air laboratories for elucidating genotype–environment associations.

[A146] Linker histone H1.0 evolution and ferroptosis reveal new perspectives on Greenland shark longevity

Kaiqiao Yang[1], Kazuya Nishiwaki[1], Hideaki Mizobata[1], Jiancheng Liang[1], Shuichi Asakawa[1], Kazutoshi Yoshitake[2], Yuuki Y. Watanabe[3], Nigel E. Hussey[4], Kit M. Kovacs[5], Christian Lydersen[5], Mitsutaka Kadota[6], Shigehiro Kuraku[7,8], Shigeharu Kinoshita[1]
[1]The University Of Tokyo, , Japan, [2]Kitasato University, , Japan, [3]SOKENDAI, , Japan, [4]University of Windsor, , Canada, [5]Norwegian Polar Institute, , Norway, [6]RIKEN, , Japan, [7]National Institute of Genetics, , Japan, [8]Graduate University for Advanced Studies, , Japan

The Greenland shark (Somniosus microcephalus), belonging to the order Squaliformes, is one of the largest extant shark species. In 2016, radiocarbon dating of eye lens tissue estimated an individual to be 392 ± 120 years old, establishing this species as the longest-lived vertebrate known and attracting widespread scientific interest. However, high-quality genomic resources suitable for comprehensive evolutionary and functional analyses have remained limited. Here, we report a chromosome-level genome assembly of the Greenland shark, with a total size of 5.9 Gb, an N50 of 233 Mb, and an overall completeness score of 96.7%. Based on this assembly, we annotated 21,094 protein-coding genes, achieving a high completeness score of 96.4%. Comparative genomic analyses were performed to investigate genetic features associated with the extreme lifespan of this species. We identified strong positive selection acting on the linker histone H1.0 gene, with a single positively selected site involving a substitution from a conserved lysine to arginine. This substitution is predicted to enhance histone–DNA binding, potentially increasing chromatin compaction and protection against molecular damage. In addition, we observed a pronounced expansion of the FTH1b gene family, with 59 copies, suggesting an enhanced capacity to rapidly regulate intracellular iron levels and modulate ferroptosis, a process closely linked to cancer and other age-related diseases. These results provide genomic insights into potential molecular mechanisms associated with the exceptional longevity of the Greenland shark and establish a genomic resource for future studies of cartilaginous fishes and lifespan evolution.

[A147] Investigating phylogenetic incongruence in wild Oryza species through genome-wide phylogenomic analysis

Kanako Koyanagi[1], Yuta Kotoku[1], Yuji Kishima[1]
[1]Hokkaido University, Sapporo, Japan

Phylogenetic incongruence is a hallmark of complex evolutionary histories, which enriches our understanding of how species diversify. In this study, we explored the genomic landscape of the Oryza AA-genome group—a traditional model for plant speciation and domestication—to investigate historical interactions between currently geographically distant species. Using a whole-genome alignment of 11 Oryza species, we detected pervasive signals that contradict the consensus bifurcating species tree. Our analysis revealed evidence of ancient genetic exchange between wild lineages currently isolated across Africa, Asia, Australia, and America. To evaluate these findings, we employed Patterson's D-statistics to distinguish introgression from incomplete lineage sorting, and used molecular dating to estimate the timing of these events. Furthermore, we performed rigorous assessments to rule out technical artifacts, such as those arising from compositional bias and substitution saturation. We also validated the signals against multiple high-quality genome assemblies, including telomere-to-telomere sequences. The results indicated that these signals were consistent with ancient introgression. Our results provide insights into the potential influence of gene flow on the evolutionary history and dispersal of these lineages.

[A148] Beyond the Gradient: Dynamic Repression as the Architect of Developmental Precision

Kasım Taha Karagöz[1,2], Jinshuai Chao[3], Berfin Bahar Şener[1], Dr. Jia Ling[4], Dr. Pınar Önal[1,2]
[1]Department of Molecular Biology and Genetics, Ihsan Dogramaci Bilkent University, Ankara, Turkiye, [2]National Nanotechnology Research Center (UNAM), Ihsan Dogramaci Bilkent University, Ankara, Turkiye, [3]Department of Biology, New York University, New York City, United States of America, [4]Department of Molecular Biology, Princeton University, Princeton, United States of America

How does a morphogen concentration gradient yield absolute precision in a developing embryo? While the Drosophila Bicoid (Bcd) gradient is the paradigm of concentration-dependent patterning, the gradient model faces a paradox. Concentration differences alone fail to explain the precise positioning of target gene expression patterns. Target boundaries are unexpectedly insensitive to Bcd dosage changes, uncorrelated with binding affinity, and formed under saturating morphogen conditions. We propose that high-fidelity patterning requires a "regulatory toolkit" of antagonistic repression. Through motif analysis of 66 Bcd-dependent enhancers, we identified a critical enrichment for transcriptional repressors representing distinct tiers of the segmentation hierarchy: Capicua (maternal), Krüppel (gap), and Runt (pair-rule). We hypothesize that these factors function not merely as binary switches, but as dynamic modulators of the Bcd signal. Supporting this view, our reporter assays indicated that Run and Cic motifs induce repression with distinct temporal dynamics. Furthermore, modification of the endogenous hunchback P2 enhancer to include ectopic repressor motifs resulted in severe, unique segmentation defects and changes in expression patterns, suggesting that distinct repressor dynamics drive specific developmental consequences. We are currently investigating whether these repressors operate via direct steric interference or higher-order chromatin remodeling. Ultimately, this study elucidates how the integration of distinct repressor dynamics modulates a single morphogen gradient to achieve robust developmental patterning, transforming a static signal into a versatile substrate for evolutionary diversity.

[A149] Inversions facilitate parallel adaptive divergence in the wildflower Senecio lautus

Kathleen Mclay[1], Dr Maddie James, Associate Professor Jan Engelstaedter, Professor Daniel Ortiz-Barrientos
[1]University Of Queensland, Brisbane, Australia

Population divergence with gene flow can occur when locally adaptive alleles are contained within regions of reduced recombination such as chromosomal inversions. Instances of parallel evolution provide a valuable opportunity to understand if the contribution of inversions is consistent across replicate instances of adaptation, or if different inversions can arise and yet achieve similar adaptive outcomes. Here we show how chromosomal inversions have contributed to the parallel evolution of distinct ecotypes in the Australian native wildflower Senecio lautus. We identified 47 large (1.1-43.3Mb) polymorphic inversions in five replicate parapatric population pairs of headland and dune ecotypes. We used genotype-environment association analysis of soil variables to identify alleles associated with adaptation to parallel contrasting habitats in replicate pairs. We show inversions are commonly enriched for environmentally associated alleles. Likewise, gene functional enrichment analysis revealed inversions are enriched for genes putatively associated with the adaptive trait of gravitropism that distinguishes the contrasting growth habits of the dune and headland ecotypes. However, only nine of the 47 inversions were shared across all five replicate population pairs. Further, even when inversions were shared by multiple population pairs, they did not consistently segregate by ecotype. While inversions contribute to parallel ecotype divergence in S. lautus, sets of inversions underlying adaptation differ between replicate population pairs, revealing population-specific genomic responses to parallel selective pressures. This work furthers our understanding of the genomic basis of parallel evolution, highlighting that similar adaptive outcomes can arise from variable genomic architectures.

[A150] Phased Assemblies Reveal Hybrid Genomes and Wine-Associated Hanseniaspora Yeasts Population Structure

Katja Doberšek[1], assoc. prof. dr. Neža Čadež[1]
[1]University of Ljubljana, Biotechnical Faculty, Department of Food Science and Technology, , Slovenia

Hanseniaspora yeasts are characterised by accelerated evolution, reduced genomes with low GC content, and extensive gene loss compared to other Saccharomycotina yeasts. Their genomes are highly dynamic, displaying strong variation in heterozygosity, structure, and ploidy both between species and among isolates, representing a rare example of long-term hypermutator lineages in fungi.

We investigated two closely related species with different ecological histories: H. uvarum, long associated with wine environments in Europe, and H. opuntiae, originally isolated from cacti in Hawaii but now increasingly found in European vineyards. Many of there European H. opuntiae isolates show hybrid ancestry with H. pseudoguilliermondii, suggesting hybridization facilitated their spread into wine-associated habitats.

To explore the genomic basis of adaptation to vineyard environments, we generated haplotype-resolved genome assemblies for representative Hanseniaspora isolates and sequenced and phenotyped a collection of 200 isolates comprising H. uvarum, H. opuntiae, and putative hybrids originating from diverse environments.

Population structure analyses revealed distinct lineages within each species that largely correlate with geographic origin, indicating ongoing local adaptation and region-specific evolutionary histories. Phenotypic assays uncovered predominantly species-specific profiles, where hybrid isolates consistently outperformed both, H. uvarum and H. opuntiae isolates under vineyard related stress conditions. Ploidy estimation revealed that most hybrids are triploid, supporting a 2:1 genomic contribution from H. opuntiae and H. pseudoguilliermondii.

Overall, our results indicate that hybridization and changes in genomic architecture are likely important contributors to fitness variation and adaptation in Hanseniaspora.

Keywords: non-model yeasts, genome evolution, population genomics, hybrid genomes

[A152] Ancient DNA reveals long-distance population contact and dynamics in northern East Asia

Ke Wang[1], Yimeng Qi[1], Yetao Zou[1], Xue Zhao[2], Yinqiu Cui[2], Hui Li[1], Li Jin[1]
[1]Fudan University, Shanghai, China, [2]Jilin University, Changchun, China

Inland East Asia, a pivotal geographic region connecting the Eurasian Steppe to the north and west, has fostered a continuous succession of prominent civilizations from antiquity to the present. Despite the region's current high population density, its prehistoric demographic dynamics remain largely undercharacterized.

In this study, we present new genome-wide data for 33 ancient individuals across three archaeological sites in Northern China, spanning 5,000 to 2,500 years before present (BP). Integrating these with modern genomic datasets and employing state-of-the-art analytical tools, we elucidate the mechanisms of genomic formation and interaction in this region. Specifically, our results reveal a dominant Yangshao-related ancestry in the Yellow River Bend, alongside unexpected long-distance genetic exchanges between the Mongolian Steppe and Southern China during the 4th–2nd millennia BP. Concurrently, in Northwest China bordering the Altai-Sayan region, we identified multiple pulses of admixture between autochthonous East Asian lineages and various Bronze Age Eurasian Steppe groups. Specifically, the integration of Altai hunter-gatherer, Eurasian Steppe, and Northern East Asian ancestries drove the prehistoric genetic turnovers and shaped the diversity of this critical inland crossroads in the archaeological site found in northwestern China.

Overall, this study offers critical paleogenomic insights into the prehistoric population dynamics of the Yellow River Bend and Northwest China (5,000–2,500 BP). We characterize a sophisticated landscape of long-distance genetic interactions that integrated inland East Asia into the Eurasian genetic landscape across vast latitudinal and longitudinal gradients.

[A153] A Myosin Heavy Chain Gene Cluster Duplication in an Ancient Lobe-Finned Fish as a Key Event for Terrestrial Life

Kei Tamura[1], Mr. Takumi Miyasaka[1], Mr. Yuto Kato[1], Prof. Akimasa Fukui[2], Dr. Michihiko Ito[1]
[1]Kitasato University, Sagamihara, Japan, [2]Chuo University, Bunkyo, Japan

During vertebrate evolution, skeletal muscles diversified to support efficient locomotion in both aquatic and terrestrial environments, largely through functional specialization of myosin heavy chain (MYH) proteins encoded by multigene myh families. To clarify how myh repertoires changed during the water-to-land transition, we performed comparative phylogenomic analyses across key lineages and integrated these results with developmental expression profiles. We identified a previously unrecognized group of fast-type myh genes that emerged in the common ancestor of lobe-finned fishes after divergence from coelacanths. This fast-type expansion was retained in lungfish and amphibians but was subsequently lost in the amniote lineage, indicating a transient diversification associated with early tetrapod evolution. In amphibians, transcriptome analyses show that these fast-type paralogs are predominantly expressed in embryonic and larval muscles. To explore potential functional consequences, we conducted molecular dynamics analyses of MYH motor domains, which predict increased motor activity in terrestrial fast MYH. This pattern is consistent with accelerated functional divergence following duplication, potentially facilitated by relaxed selective constraint. Together, these findings suggest that duplication of a fast-type myh cluster and its subsequent lineage-dependent fate contributed to skeletal muscle diversification during the emergence of vertebrates onto land. We further propose that this cluster was maintained because it supported larval muscle function, but became unnecessary and was lost in the amniote lineage alongside the evolutionary reduction of the larval stage. More broadly, this transient cluster may have provided a window for developmental and functional exploration, helping to shape lineage-specific innovations during early tetrapod evolution.

[A154] Amylase-binding proteins as indicators of dietary adaptation in the human oral microbiome

Keri Burge[1], Irina Velsko[2], Christina Warinner[1,2]
[1]Harvard University, Cambridge, United States, [2]Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

Bacterial populations evolve and fix genetic changes much more rapidly than humans, and consequently the evolutionary patterns of bacteria within the human microbiome have the potential to serve as a proxy for tracing human behavioral and dietary changes over time. One such shift, the adoption of starch-rich diets, is reflected in humans by copy number expansion of the salivary amylase gene (AMY1). In parallel, certain Streptococcus species in the human oral microbiome produce amylase-binding proteins (ABPs) that interact with human salivary amylase, suggesting co-evolution of these microbes along with human host genetic variation. This project investigates the evolution of ABP genes to better understand how host-associated microbial genes adapt and diversify in response to dietary niche shifts. We examine the distribution and phylogenetic relationships of ABP genes across a wide range of ancient and modern comparative datasets, including industrial and non-industrial human dental plaque, non-human primate and mammalian dental calculus, soil metagenomes, and human gut microbiomes. By establishing a comprehensive baseline of modern ABP diversity and host specificity and contrasting it with ancient human oral diversity, this study provides a framework for testing hypotheses about adaptive evolution and host–microbe coevolution in response to dietary shifts toward increased starch consumption. These findings will inform future analyses of ancient microbiomes and contribute to understanding the coevolutionary relationship between human diet, host genetics, and commensal microbial adaptation.

[A155] Diversity of sex chromosomes in seahorses and pipefish

Kevin Hsiung[1], Prof. Olivia Roth, Astrid Böhne
[1]Leibniz-Institut zur Analyse des Biodiversitätswandels, Bonn, Germany
Sexual reproduction in teleost fishes is remarkably diverse, with frequent transitions of sex chromosomes and shifts in the underlying sex-determining mechanisms. The family Syngnathidae (seahorses, sea dragons, pipehorses, and pipefish) is especially intriguing because the evolution of male pregnancy and sex-role reversal offers a rare opportunity to examine how sexually antagonistic selection influences sex chromosome evolution. Multiple syngnathid sex chromosomes have been described to date, all of which have been reported as male-heterogametic (XY). Here, we analyzed Pool-seq data, individual DNA libraries, and RNA-seq data to investigate sex chromosomes in seven syngnathid species, including the seahorses Hippocampus barbouri and Hippocampus guttulatus, and the pipefishes Corythoichtys haematopterus, Nerophis ophidion, Doryrhamphus bicarinatus, Syngnathus typhle, and Syngnathus rostellatus. Across these species, we detect both XY and ZW non-homologous sex chromosomes in some lineages, while confirming a lack of differentiated sex chromosomes in others. These results add to the growing evidence that Syngnathidae represent a highly dynamic system for studying sex chromosomes and sex determination.

[A156] Effect of past climate change events on demographic history of bats

Kirnalee Patel[1], Effect of past climate change events on demographic history of bats Durbadal Chatterjee[2], Effect of past climate change events on demographic history of bats Ratnesh Karjee[1], Effect of past climate change events on demographic history of bats Kritika Garg[2], Effect of past climate change events on demographic history of bats Balaji Chattopadhyay[1]
[1]Ashoka University, Sonipat, India, [2]IISER Mohali, Mohali, India

Demographic history of a species is crucial for understanding how global climatic fluctuations have shaped its current diversity and distribution. Recent advancements in genomics and the coalescent framework have facilitated the exploration of the demographic history of species using genomic data. We used the Pairwise Sequentially Markovian Coalescent (PSMC) method to reconstruct the demographic history of multiple bat species. Here, we used single whole-genome sequence data for 38 bat species to investigate their demographic fluctuations during the Late Pleistocene. This period is known for its repetitive glacial cycles, the evolution and spread of early humans, and its effects on populations, resulting in the expansion or contraction of several mammalian species. We also employed species distribution modelling to identify how the habitat availability has fluctuated for these bat species during the Late Pliestocene. The reconstructed effective population size (Ne) increased for 8 species and decreased for 30 species from the Last Interglacial Period (LIG) to the Last Glacial Maximum (LGM). We observed an increase in suitable habitat from LIG to LGM for 25 species out of 38 species corresponding to Quaternary climate shifts. This study uses two complementary methods and provides significant insights into understanding how bats respond to climate change events during the Late Pleistocene Glacial cycles.

[A157] Dynamics of adaptation to slowly fluctuating environments

Salvatore Banno[1], Liam Murphy[1], Neha Pandey[1], Katri Ketola[1], Jonna Kulmuni[2], Ilkka Kronholm[1]
[1]Department of Biological and Environmental Science, University of Jyväskylä, Jyväskylä, Finland, [2]Institute for Biodiversity and Ecosystem Dynamics, University of Amsterdam, Amsterdam, Netherlands

How do populations evolve when the environment fluctuates slowly over hundreds of generations? Theoretical models suggest that populations primarily track the environmental optimum through genetic adaptation. Furthermore, is adaptation reversible, or does it involve the fixation of compensatory mutations when the environment returns to its original state? Previous evolutionary experiments have focused on adaptation to drastic, gradual, or rapidly fluctuating changes, whereas adaptation to slow fluctuations remains poorly characterized. We conducted a long-term evolutionary experiment under this regime using populations of fission yeast founded from two isogenic clones differing in their mating type. We gradually introduced and intensified environmental stress to a peak, then removed it at the same rate to return to the original conditions. We chose two environmental stressors, osmotic stress, increased by adding potassium chloride to the growth medium and ethanol stress. Additionally, there was a control treatment where the environment remained constant. Regular rounds of sexual reproduction were also induced, allowing genetic recombination within each population throughout the experiment. We measured population growth rates at peak stress levels to test for adaptation. Preliminary results showed that populations evolved in KCl and EtOH environments are able to grow at their peak stress levels, while ancestors show little to no growth. Furthermore, by following allele frequency changes using population sequencing, we aim to determine whether adaptation occurred and whether populations fixed compensatory mutations when the environment returned to the original state.

[A158] Drivers and constraints to the recurrent evolution of a color pattern in frogs and toads

Sandra Goutte[1]
[1]NTNU, Trondheim, Norway

Certain colors and color patterns are found across numerous distantly related lineages around the world, indicating shared evolutionary constraints at the ecological, physiological or molecular level. This is the case, for example, of the anuran vertebral stripe pattern, an ecologically-relevant color trait thought to help avoid predators' detection under certain circumstances. Here we combine histological analyses with genome- and transcriptome-wide data to pinpoint the genomic variations linked to this trait in taxa spanning a wide range of divergence times. By comparing these lineages, we assess whether this convergent phenotypes arise through shared or distinct molecular mechanisms.

[A159] A multi-scale genotype-phenotype map of developmental regulatory networks

Santiago Herrera-Alvarez[1], Justin Crocker[1], Michael Dorrity
[1]EMBL, Heidelberg, Germany

Phenotypic evolution depends on the generation of variation by mutation, followed by its sorting through selection and drift. Explaining patterns of biological diversity, therefore requires understanding how biological systems produce phenotypes. Yet we still lack a mechanistic framework of phenotype production that links genetic variation to biochemical interactions, cellular behaviors, and organismal form.

In animals, phenotypes are produced through embryonic development, orchestrated by gene regulatory networks (GRNs) that control the timing, location, and magnitude of gene expression. At the biochemical level, interactions between transcription factors (TFs) and DNA are core organizing elements of GRNs: TF–DNA binding initiates and propagates developmental signals in specific cellular contexts. These biochemical interactions are translated at the cellular level into gene-expression programs that define cell identity and behavior. Understanding phenotypic evolution, therefore requires integrating biochemical and cellular phenotypes across developmental scales.

Here, I propose to combine synthetic developmental biology with whole-organism single-cell genomics to ask: how does the biophysical architecture of developmental systems shape the production and evolution of phenotypes? I will focus on two well-characterized systems: the Drosophila Bicoid TF, which controls anterior–posterior patterning, and the zebrafish Brachyury TF, which regulates mesoderm specification. Together, these GRNs provide tractable models to address three questions: (1) how TF–DNA affinity varies across cells in a developing embryo; (2) how this variation influences cell identity and composition; and (3) how perturbations to TF-DNA affinity propagate through the GRNs to produce morphological variation.

[A160] Climate Change, Range Shift and Hybridization: A Genetic Investigation of P. maniculatus and leucopus

Sargon Yousefian Dezag Tekkie[1], Jenna LeBlanc[1], Amanda V. Meuser[2], Vincent Careau[3], Eryn McFarlane[1]

[1]York University, , , [2]University of Stockholm, , , [3]University of Ottawa, ,

Hybridization occurs when genetically distinct species interbreed, producing offspring that can alter evolutionary trajectories. The outcomes of hybridization range from the formation of novel lineages to the erosion of local adaptation, making case-specific study essential. Peromyscus maniculatus (white footed mouse) and P. leucopus (deer mouse) have historically overlapped across parts of eastern North America, yet exhibit little evidence of hybridization in long-term contact zones, suggesting the evolution of strong reproductive barriers. However, climate-driven range shifts are now increasing secondary contact between newly sympatric populations of these species, creating new opportunities for hybridization. In this research, we have investigated whether hybridization occurs under these contemporary conditions and how hybridization rates vary across time. Using genomic data from thousands of SNPs, and long-term research from the Queen's University Biological Station, I will present my results of individual ancestry, quantify putative hybrids, and assess ecological and geographic factors associated with hybridization. By integrating population genomics with environmental context, this study clarifies how rapid, human-driven environmental change reshapes species boundaries.

[A161] Dispersal history of the brown rat inferred from Asian population genomics

Satsuki Hirasawa[1,2], Toshinori Endo[1], Hitoshi Suzuki[1], Naoki Osada[1]
[1]Hokkaido University, Sapporo, Japan, [2]RIKEN, Wako, Japan

The brown rat (Rattus norvegicus) is a commensal species that has substantial impacts on human settlements, public health, and urban environments. Despite its importance, the origin, dispersal patterns, and genetic diversity of wild populations remain incompletely understood.
Fossil records and recent genetic studies have suggested an Asian origin of the brown rat; however, exact location remains debated. In this study, we conducted a comprehensive population genomic analysis using newly sequenced Japanese wild rat genomes together with published datasets. Consistent with previous studies, our results show high phylogenetic diversity in Asia. In addition, the Obihiro population, located in eastern Hokkaido (the northernmost main island of Japan), diverged from other populations approximately 6,500-13,000 years ago, indicating that the earliest population divergence likely occurred in northern Asia. Notably, populations from the Japanese main islands excluding Hokkaido do not show such a pattern, indicating that at least two distinct lineages have existed in the Japanese archipelago.
We further examined admixture with its sister species, Rattus nitidus, and detected strong signals in southern China, as suggested by previous studies. These signals were also detected in European populations, supporting a dispersal scenario in which European populations derived from southern Chinese populations.
Overall, our results clarify the genetic relationships among Asian wild brown rat populations and provide important insights into the origin and dispersal patterns of this species.

[A162] Population-specific signals of positive selection in the human CYP1A2 gene.

Sayaka Chiku[1], Jun Gojobori[1,2], Yoko Satta[1,2]
[1]Department of Evolutionary Studies of Biosystems, SOKENDAI, Shonan Village, Hayama, Japan, [2]RCIES, SOKENDAI, Shonan Village, Hayama, Japan

Human CYP1A2 is one of detoxification enzymes, that metabolizes xenobiotics such as pharmaceuticals, and is highly expressed in human liver. Although substantial inter-individual variation in CYP1A2 expression and activity has been reported, its evolutionary background remains unclear. In this study, I investigated whether specific CYP1A2 SNPs have been subject to positive selection in human populations.

Based on the estimated genomic FST, 11 out of 232 SNPs in the CYP1A2 region (GRCh37) were found in the top 2.5% of the distribution. Among them, rs762551 and rs57295890 showed exceptionally high FST values in the Peruvian population, driven by population-specific derived allele frequencies. In European populations, rs2472304 and rs2470890 also exhibited elevated derived allele frequencies relative to most non-European populations. These SNPs showed strong linkage disequilibrium with neighboring variants in CYP1A2 and CYP1A1 regions, consistent with the effects of European-related introgression observed in admixed American populations. To further test recent positive selection (<10,000 years), population branch statistics were calculated using Peruvian, African, and East Asian populations. Both rs762551 and rs57295890 ranked within the top 2.5% of PBS values on chromosome 15, supporting population-specific selection signals.

To evaluate the significance of these signals, PBS distributions were generated under neutral simulations based on a previously published demographic model for Peru, and compared with the observed values. In addition, linkage disequilibrium between CYP1A1 and CYP1A2 was examined, revealing strong LD among selected SNPs in European populations, likely reflecting historical introgression. Recent results from these analyses will be presented.

[A163] Longitudinal Plasma Proteomic Profiling Connects Genomic Changes to Systemic Aging Phenotypes

Sehee Choe[1], Hyeon-Mu Cho[1], Hye-Ri Park[1,2], Min-Kyeong Ko[1,2], Yun-Jung Lee[1,2], Sang-Je Park[1], Jaewon Huh[1,2]

[1]Korea Research institute of bioscience & Biotechnology, NPRC, Cheongju, South Korea, [2]Department of Functional Genomics, KRIBB School of Bioscience, UST, Cheongju, South Korea

Aging is the strongest risk factor for most chronic diseases and a major determinant of healthspan. Despite its broad impact, aging remains poorly understood due to its complex and multifactorial nature. While the genome is largely stable across the lifespan, age-associated somatic alterations and systemic physiological changes suggest that downstream molecular phenotypes may provide critical insights into aging processes. Age-related immune dysregulation has been repeatedly reported across species, and we have previously observed such alterations at the genomic and transcriptomic levels in primates. In this context, we designed this study to validate whether age-related genomic and transcriptomic changes translate into functional proteomic remodeling, and whether these changes reflect immune dysregulation and shifts in physiological energy allocation, using an integrated multi-omics sampling strategy. Plasma proteomic profiling was performed using the Olink Reveal platform on longitudinal samples from two independent aging-related projects. The dataset included heterogeneous temporal structures: one individual with three consecutive annual measurements, six individuals with continuous second-to-third year follow-up, and eight individuals with non-consecutive first-to-third year comparisons. Analysis revealed age-associated remodeling of the plasma proteome, with changes in proteins involved in immune regulation, inflammation, metabolism, and tissue homeostasis. Longitudinally sampled individuals demonstrated heterogeneity in proteomic aging trajectories, indicating substantial inter-individual variation. Together, these findings support that aging-related proteomic changes reflect systemic physiological reorganization and represent functional consequences of previously observed genomic and transcriptomic alterations. This study demonstrates the potential of plasma proteomics as a platform linking genome-transcriptome-proteome in aging research.

[A164] Peculiar Pigment Pattern and Population Profile of a Poisonous Pufferfish

Seita Miyazawa[1], Hiroyuki Doi[2], Hiroshi Takahashi[3], Tomoaki Nishiyama[4], Shuji Shigenobu[5], Kazuharu Misawa[6,7], Kiyoshi Kikuchi[8], Harumi Sakai[3]
[1]D3 Center, The University of Osaka, Suita, Japan, [2]Osaka Aquarium NIFREL, Suita, Japan, [3]Department of Applied Aquabiology, National Fisheries University, Shimonoseki, Japan, [4]School of Science, Academic Assembly, University of Toyama, Toyama, Japan, [5]National Institute for Basic Biology, Okazaki, Japan, [6]Graduate School of Data Science, Yokohama City University, Yokohama, Japan, [7]RIKEN Center for Advanced Intelligence Project, Chuo-ku, Japan, [8]Fisheries Laboratory, The University of Tokyo, Hamamatsu, Japan

Pufferfish are well-known for their toxicity, yet they also exhibit a remarkable diversity of pigment patterns. Mushifugu (Takifugu exascurus) is a pufferfish endemic to Japan's coastal waters and is characterized by conspicuous labyrinthine patterns. Despite being recorded along both the Sea of Japan and the Pacific coasts, it has a limited distribution and is infrequently observed. Aside from its unique body pattern, mushifugu shows little to no morphological differences from other Takifugu species, often leading to speculation that it may be an interspecific hybrid. In addition, previous theoretical and empirical studies have shown that complex camouflage-like labyrinthine patterns can emerge through the 'pattern blending' caused by hybridization between spotted species, providing support for this possibility. Here, we investigate the phylogenetic origin of mushifugu and its distinctive pattern through population structure analysis and demographic inference in comparison with its closest spotted relative, komonfugu (T. flavipterus).

[A165] Mitochondrial support of cardiac plasticity in the high-elevation deer mouse Peromyscus maniculatus

Shady Kuster[1], Jada Joseph[1], Daniel Sloan[1], Adam Chicco[1], Kathryn Wilsterman[1]
[1]Colorado State University, Fort Collins, United States

Mitochondria are central hubs for important metabolic processes supporting cellular and organismal tasks, but they are dynamic organelles with different roles dependent on their tissue context and the organism's physiological condition. High-elevation hypoxia (low oxygen) causes a cascade of physiological responses in non-adapted animals. One effect of these responses is hypertrophy (overgrowth) of the right ventricle of the heart. Approximately 75,000 years of adaptive evolution of highland-inhabiting deer mice (Peromyscus maniculatus) has resulted in attenuation of this maladaptive plasticity, among others. We hypothesized that the mitochondria from right and left ventricles of the heart differed in their response to simulated high-elevation hypoxia based on highland ancestry of nuclear and mitochondrial genomes. Using highland and lowland derived deer mice and their F1 reciprocal crosses, we tested this hypothesis by acclimating mice to 8 weeks of either cold (5°C) normoxia or cold hypoxia (12kPa) to simulate high-elevation conditions. We measured mitochondrial oxygen consumption in response to mitochondrial substrates using high-resolution mitochondrial respirometry for samples from both ventricles. Mitochondria from both ventricles show a decrease in pyruvate oxidation metrics under cold hypoxia acclimation. The two ventricles differ in a genotype-specific manner in pyruvate-driven leak respiration, but not in an expected pattern. These differences were no longer significant in mitochondria isolated from the left ventricle, indicating that cellular support of mitochondrial function differs between highland and lowland genotypes. Therefore, we observe no mitochondrial differences in hypoxia responses between the two ventricles of the heart.

[A166] The temporal, molecular, and life-history constraints on the evolution of phenotypic plasticity

Shannon Snyder[1], William A. Cresko[1,2]
[1]Institute of Ecology & Evolution, University of Oregon, Eugene, , United States, [2]Department of Bioengineering, Knight Campus for Accelerating Scientific Impact,, Eugene, United States
Transgenerational phenotypic plasticity (TGP), the transmission of environmentally induced phenotypes across generations without changes to DNA sequence, has emerged as a potentially adaptive mechanism by which organisms respond to variable selective pressures. Yet fundamental questions remain: how long do transgenerational effects persist when the environment changes, what fitness costs accompany them, and do the mechanisms underlying inherited phenotypes mirror those producing the original response? Here we address these questions using asexual Daphnia lumholtzi exposed to fish kairomones as a model system for predator-induced TGP.
First, we examined the generational persistence of morphological TGP, demonstrating that predator-induced defensive morphologies are transmitted across multiple generations, including those definitively removed from germline exposure. Next, we investigated life-history trade-offs associated with this inheritance, revealing that transgenerationally expressed phenotypes incur substantial fitness costs manifesting generations after environmental exposure, suggesting TGP is not a costless form of anticipatory plasticity. We further investigated the cellular mechanisms underlying TGP, identifying species-specific proliferative dynamics indicating diverging mechanisms produce conserved predator responses. Finally, we employed differential gene expression analyses to compare transcriptomic profiles of directly exposed (G0) and unexposed but phenotypically expressing (G3) individuals, revealing that inherited and direct TGP are mechanistically distinct despite producing convergent phenotypic outcomes.
Together, these findings indicate that predator-induced TGP in D. lumholtzi is a persistent but costly phenomenon, and that transgenerational inheritance operates through molecular pathways that diverge from those mediating the original plastic response. These results have broader implications for understanding the evolutionary significance and mechanistic basis of nongenetic inheritance.

[A167] Relaxation of selective constraint on the sweet-taste receptor gene TAS1R2 in lorisiform primates

Shoji Kawamura[1], Dr. Qinyuan Ji[1], Dr. Min Hou[1], Dr. Muhammad Shoaib Akhtar[1], Dr. Takashi Hayakawa[2], Dr. Yasuka Toda[3], Dr. Akihiro Itoigawa[4], Dr. Hiroo Imai[5], Dr. Takafumi Ishida[6], Prof. Amanda Melin[7]
[1]Department of Integrated Biosciences, Graduate School of Frontier Sciences, The University of Tokyo, Kashiwa, Japan, [2]Faculty of Environmental Earth Science, Hokkaido University, Sapporo, Japan, [3]School of Life Science and Technology, Institute of Science Tokyo, Yokohama, Japan, [4]Institute for Protein Research, The University of Osaka, Suita, Japan, [5]Molecular Biology Section, Center for the Evolutionary Origins of Human Behavior, Kyoto University, Inuyama, Japan, [6]Department of Bioscience, Graduate School of Science, The University of Tokyo, Hongo, Japan, [7]Department of Anthropology and Archaeology, University of Calgary, Calgary, Canada
Recent research has revealed considerable evolutionary diversity in umami-taste (amino acids and nucleotides) and sweet-taste receptor TAS1R genes across vertebrate species. To contribute to a growing understanding of how diet shapes taste evolution, we studied TAS1R genes in non-anthropoid primates with highly diverse diets, including members of the Strepsirrhini, comprised of Lorisiformes (lorises) and Lemuriformes (lemurs), as well as members of the Tarsiiformes (tarsiers). We employed a targeted capture (TC) approach specifically probing all the three mammalian TAS1R genes, i.e., TAS1R1 (for sensing umami), TAS1R2 (sweet) and TAS1R3 (required for forming heterodimers), followed by short-read massive-parallel sequencing for three lorisiform, four lemuriform, and one tarsiiform species. Analyzing together with publicly available whole-genome assemblies (WGAs) of non-anthropoids, we found that TAS1R1 and TAS1R2 of some lorisiform species were disrupted. The relative evolutionary rates in introns and synonymous sites of all the three TAS1R genes, as well as non-genic genome regions, of lorisiforms were higher than those of lemuriforms, a finding consistent with the higher genome-wide mutation rate of the lorisiforms. We found a similar pattern in the amino acid sequences and nonsynonymous sites of the sweet receptor TAS1R2 in lorisiforms. Evolutionary rates of amino acid sequences and nonsynonymous sites in TAS1R1 and TAS1R3 of lorisiforms were as slow as those of lemuriforms. This suggests that functional constraint on sweet sensing has been relaxed in lorisiform primates since their common ancestor. These results shed a new light on understanding evolutionary diversification of umami and sweet sensing in a diverse group of mammals.

[A168] Evaluating data processing approaches for comparative primate functional genomics research

Shuanghui Chen[1], Irina Milicevic[1], Mary Kelaita[1], Dr. Genevieve Housman[1]
[1]Department of Primate Behavior and Evolution, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

Comparing gene expression variation among species is central to evolutionary and functional genomics research and has substantially advanced our understanding of conserved and divergent regulatory landscapes across taxa in recent years. A major challenge in such studies is deciding how best to process sequencing data to ensure cross-species comparability of sequencing data, as differences in reference genome and annotation quality can bias read mapping and feature calling. For transcriptomic data, several methods have been developed to enable the unbiased comparison of gene count matrices across species, but their relative strengths and limitations have not been systematically characterized across a broad range of comparative research designs. In this study, we evaluated a recently developed reciprocal mapping method (Crossfilt) alongside a commonly used approach that maps reads to a cross-species consensus genome, using RNA sequencing data from six primate species (human, chimpanzee, bonobo, gorilla, orangutan, and macaque) organized into several pairwise (e.g., human vs. gorilla) and multi-species (e.g., human vs. chimpanzee vs. macaque) comparative designs. Read mapping and feature calling metrics across different processing methods and comparative designs revealed variation in the number and genome-wide distribution of sequencing reads and called features that could be reliably used for downstream analyses. These findings, in combination with practical considerations for each method (e.g., generating orthologous annotation), demonstrate the trade-offs inherent in current cross-species transcriptomic data processing methods and highlight the importance of evaluating analytical choices in relation to evolutionary scale and study design.

[A170] Optimization of wet lab protocols for reference genome production of colonial ascidians

Simona Buonanno[1], PhD Grazia Visci[1], PhD Ahana Maitra[1], Melania Spinelli[1], PhD Ilenia Urso[1], PhD Giuseppe Defazio[1], Professor Graziano Pesole[1,2], Prof Carmela Gissi[1,2,3]
[1]Dept of Biosciences, Biotechnologies and Environment, University of Bari "Aldo Moro", Bari, Italy, [2]IBIOM, Institute of Biomembranes, Bioenergetics and Molecular Biotechnologies, CNR, Bari, Italy, [3]Consorzio Nazionale Interuniversitario per le Scienze del Mare, Roma, Italy

Ascidians are solitary or colonial marine chordates that play a central role in the ecology of benthic communities. They are widely studied for their invasive potential, to unravel the chordate evolution, and as model species in evolutionary developmental biology and for several biological processes. However, ascidian biodiversity is currently underestimated due to the frequent occurrence of species complexes revealed by integrative taxonomy, as well as intrinsic challenges in genomic characterization (i.e., small body size, presence of unknown pigments or metabolites, etc).
Within the framework of the Biodiversity Genomics Europe project, we aim to advance ascidian biodiversity research by expanding the production of high-quality reference genomes compliant with Earth BioGenome Project standards.
Here we report the optimization of the wet lab protocols for the extraction of High Molecular Weight DNA (HMW-DNA), RNA, as well as for the generation of Oxford Nanopore Technologies long reads and Arima HiC data from selected species. The analysed taxa were colonial ascidians, mainly belonging to the Styelidae family, including a species-complex case, for which specimens were processed as entire colony, without tunic removal. Protocol optimization was tested on samples preserved in 99% ethanol, RNA later, or flash frozen, to assess whether putatively sub-optimal preservation methods could still be suitable to produce high-quality reference genomes.
Our results demonstrate that even non–flash-frozen samples are suitable for generating high-quality reference genomes and indicate that our optimized protocols can be successfully extended to other challenging colonial ascidian groups including members of the order Aplousobranchia.

[A171] Decoding the Plantain Genome: A Complete Assembly for Structural and Evolutionary Studies

Simona Martikánová[1], Gabriela Majzlíková[1], Zuzana Tulpová[1], Jana Čížková[1], Delphine Amah[2], Moses Nyine[2], Rony Swennen[2,3], Eva Hřibová[1]
[1]Centre of Plant Structural and Functional Genomics, Institute Of Experimental Botany, CAS, Olomouc, Czech Republic, [2]International Institute of Tropical Agriculture, Ibadan, Nigeria, [3]International Institute of Tropical Agriculture, Kampala, Uganda

Plantain is an essential food crop across the tropics, yet genomic resources for many cultivated clones remain incomplete due to polyploidy, high heterozygosity and abundant repeats. Here, we present a chromosome-scale, haplotype-resolved genome assembly and gene annotation of the cultivar '3 Hands Planty' to enable robust evolutionary and applied genomic analyses. Flow cytometry estimated a nuclear DNA content of 1.786 pg (2C), corresponding to ~582 Mbp per 1Cx, consistent with a triploid (AAB genome) genome structure.

High-fidelity PacBio reads and long nanopore reads, Hi-C data and Bionano optical maps enabled resolution of complex repetitive regions, contiguity and haplotype-aware reconstruction. The resulting assembly provides near-complete representation of all chromosomes with substantially reduced gap content. Gene prediction integrated evidence from transcriptomic data and protein homology to produce a high-confidence gene set, supported by standard completeness (~97% BUSCO) and quality assessments.

To place plantain in a broader Musaceae context, we performed comparative analyses across Musa genomes, including orthology inference and phylogenomic reconstruction using single-copy orthologues. These analyses highlighted the conserved syntenic blocks as well as lineage- and cultivar-specific variation in gene content, and structural organization, offering insights into plantain evolution and domestication-related traits.

This genome resource will facilitate genetic and evolutionary studies in banana, support marker development and functional genomics, and accelerate breeding efforts aimed at improving yield stability and resilience.

[A172] Detecting and characterising hybridisation between native and introduced species of Maylandia cichlid fish in Lake Malawi

Sing Wei Lim[1], Moritz Blumer[1], George Turner[2], Bettina Fischer[1], Professor Richard Durbin[1]
[1]University of Cambridge, Department of Genetics, Cambridge, United Kingdom, [2]Bangor University, School of Environmental & Natural Sciences, Bangor, United Kingdom

The haplochromine cichlids of the East African Rift Valley Lakes represent one of the most rapid recent diversifications of vertebrates, with Lake Malawi in particular being home to over 700 species of cichlids. Within this larger radiation, we focus on the Maylandia subradiation, which comprises the genera Maylandia and Cynotilapia. This subradiation is interesting for its diverse phenotypes, including variation in barring, colouration, and dentition.

Using a callset of 952 individuals collected from 24 sublocations across the lake, assigned to 15 species, we characterised the population structure and phylogenetic relationships within this radiation with a combination of SNP and TE-based approaches. Through this, we observed a potential hybrid zone between native Maylandia zebra and anthropogenically introduced Maylandia callainos populations at Mumbo Island, but not at other locations that we sampled. This comprises the first genetic evidence for hybridisation in this species pair, and provides a unique opportunity to study the effects of recent hybridisation caused by human interference, especially potential genomic shock caused by incompatible transposon compositions in differing parental haplotypes. Despite the two species also co-occurring at other locations, this hybrid pair is specific to Mumbo Island, potentially allowing us to explore ecological contexts and population histories favouring hybridisation.

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[A173] Quinone chemistry shapes NADH dehydrogenase utilization in bacterial respiratory chain evolution

Snehal Khairnar[1]
[1]Tata Institute Of Fundamental Research ,mumbai, Mumbai, India, [2]Anjali patil, Mumbai, India, [3]L. Karvannan, Mumbai, India, [4] Dr. Amitesh Anand, Mumbai, India

Aerobic respiration oxidizes NADH via proton-pumping NDH-1 or non-pumping NDH-2, transferring electrons to oxygen through quinones: ubiquinone (UQ) or naphthoquinone (NQ).
Quinones vary in redox potential and history: ancestral low-potential NQ preceded the Great Oxidation Event, followed by high-potential UQ.

Many bacteria, including E.coli, synthesize both quinones and dynamically switch between them in response to environmental cues. Yet UQ-deficient E. coli exhibit impaired aerobic growth, implying that NQ alone may impose metabolic constraints under oxic conditions, whereas several obligate NQ-dependent bacteria perform efficient aerobic respiration. This paradox raises a fundamental evolutionary question: if NQ can support oxygen respiration, why is UQ retained so broadly among aerobic bacteria?

To address this,we evolved NQ-only E. coli via adaptive laboratory evolution (ALE) for aerobic fitness.WGS of independently evolved populations revealed recurrent mutations in genes governing respiratory metabolism and redox homeostasis that consistently increased expression of NDH-2. Functional assays demonstrated that evolved strains achieved enhanced aerobic growth by shifting dependence from NDH-1 to NDH-2, indicating that quinone composition can modulate NADH dehydrogenase utilization at the physiological level.

To assess the evolutionary significance of this physiological coupling, we applied a Hidden Markov Model –based comparative genomics framework to survey genome-wide distributions of quinones and NDHs across 9,000 phylogenetically diverse prokaryotic species. We found robust, widespread enrichment of NDH-2 in NQ-utilizing lineages, whereas UQ-rich lineages favor NDH-1.

These findings demonstrate a tight coupling between quinone chemistry and the architectural organization of bacterial respiratory chains. Future work will investigate whether quinone-dependent specificity extends to other respiratory complexes.

[A174] A joint Bayesian framework for identification of genotype-phenotype associations in birds

Sofia Stroustrup[1], Sze Chai Helen Wong[1], Jacky Kaiyuan Li[2,4], Dr. Nicolas Alexandre[5], John Huelsenbeck[2], Rasmus Nielsen[1,2,3]

[1]Section for GeoGenetics, University of Copenhagen, , Denmark, [2]Department of Integrative Biology, University of California Berkeley, , United States, [3]Department of Statistics, University of California, , United States, [4]Biostatistics Division, University of California, , United States, [5]Department of Ecology and Evolutionary Biology, University of California, , United States

A fundamental goal in evolutionary biology is to understand the molecular drivers of morphological evolution. To facilitate this, we develop a comparative framework for joint evolutionary analysis of morphology, i.e. biological shapes outlined using landmarks, and non-coding genomic regions. Specifically, we combine two Bayesian models; one which estimates evolutionary rates based on novel stochastic diffeomorphic models of shape change and one which estimates DNA substitution rates using state-of-the-art methods. By combining these models we are able to jointly analyze whole-shape geometric morphometrics data and genomic data and pinpoint genotype-phenotype associations using rate correlations along the edges of large phylogenies. We apply our joint Bayesian framework on upstream regulatory regions for approximately 48 candidate genes extracted from the genome-wide alignment generated as a part of the Bird 10,000 Genomes Project and geometric morphometrics data from the Mark My Bird project. These publicly available data sets allow us to analyze up to 348 species per gene.

[A175] Questioning genetic supremacy: investigating the transmission of phenotypic traits and DNA methylation patterns across three generations in quails.

Stacy Rousse[1], Remi Seraphin[1], Sophie Leroux[1], David Gourichon[2], Marta Godia[3], Ole Madsen[3], Frederique Pitel[1], Sonia E Eynard[1]

[1]GenPhySE, Université de Toulouse, INRAE, ENVT, 31326, Castanet-Tolosan, France, , France, [2]PEAT, UE1295 - INRAE Centre Val de Loire, 37380, Nouzilly, France, , France, [3]Animal Breeding and Genomics, Wageningen University & Research, 6700AH Wageningen, The Netherlands, , The Netherlands

Part of the environmental story of individuals lies within their DNA methylation (DNAm). Variations in the environment can trigger changes at the DNAm level and translate to phenotypic variability, even in the unexposed progeny of the individuals directly impacted. However, the amount of environmental information transmitted between generations via epigenetic phenomena is hardly quantifiable. The present study analyses DNAm from Reduced Representation Bisulfite Sequencing (RRBS) data for 1267 quails (Coturnix japonica) within three successive generations following an initial phytoestrogen ingestion. Among 112,561 CpG sites observed, we estimated an average heritability of DNAm of 0.24. After identification of differentially methylated regions (DMR) between control and treated groups in three consecutive generations, we conducted methylation quantitative trait loci (metQTL) analysis to identify regions that seem to escape genetic control. Using phenotypic data available on the same individuals, one can assess the portion of genetic and epigenetic determinism responsible for trait variability, and how it evolves across generations.

[A176] Delineating potential influence of gut microbiota on the pathophysiological outcomes of early life stress

Stuti Srivastava[1], Dr Pratik Chaudhary[1], Ms Nishtha Pange[1], Prof Vidita Vaidya[1], Amitesh Anand[1]
[1]Dept. of Biological Sciences, Tata Institute Of Fundamental Research, , India

Early life adversities profoundly impact host physiology in adulthood. Maternal separation (MS), a well-established rodent model of early life stress (ELS), induces neurocognitive defects, gut dysbiosis, impaired intestinal barrier, and inflammatory pathologies that emerge or worsen with age. Our recent work shows that MS-associated outcomes, including anxiety, depression, peripheral inflammation and blood-brain-barrier dysfunction, are partially rescued by oral supplementation with nicotinamide, a metabolite tightly linked to microbial energy metabolism. Many of these pathologies have been associated with the gut microbiota. However, there lacks an understanding of age-dependent changes in the gut microbial community and how these impact ELS outcomes. We conducted a longitudinal metagenomic analysis of gut microbial communities across the lifespan of MS-exposed rats, with and without nicotinamide supplementation, to characterize age-dependent shifts in the microbiome. Neonatal MS leads to sustained alterations in gut microbial composition and function, with dysbiosis becoming more pronounced with aging. Notably, post-MS nicotinamide treatment largely restores the microbiota disrupted by ELS. Nicotinamide supplementation selectively modulates the abundance of bacterial taxa implicated in its metabolism, including Ligilactobacillus, and significantly alters metabolic pathways related to energy homeostasis. Our findings highlight how ELS can alter metabolic interactions between host and microbiota, potentially accelerating molecular aging and inflammation. Modulation of nicotinamide metabolism represents a promising strategy to rebalance host-microbe molecular interactions and mitigate the long-term consequences of ELS.

[A177] Coding sequence evolution associated with avian beak shape diversification

Sze Chai Helen Wong[1], Sofia Stroustrup[1], Jacky Kaiyuan Li[2], Dr. Nicolas Alexandre[3], John Huelsenbeck[2], Rasmus Nielsen[1,2]
[1]University of Copenhagen, , Denmark, [2]University of California, Berkeley, , USA, [3]University of California, Santa Cruz, , USA

Bird beaks are a classic model for adaptive diversification, yet the genetic changes that contributed to major shifts in beak morphology across evolutionary history remain difficult to pinpoint. Here, we survey genotype–phenotype relationships for approximately 90 candidate loci implicated in beak development, focusing on patterns of molecular evolution in their protein-coding regions. This work uses 363 bird genomes and the reconstructed phylogeny from the Bird 10,000 Genomes Project Phase II, which provides dense taxon sampling for comparative inference. Branch-specific rates of beak shape evolution are re-estimated from landmark-based morphometric data under a Bayesian framework that approximates the Large Deformation Diffeomorphic Metric Mapping process and is implemented with an efficient local-update Markov Chain Monte Carlo algorithm. Posterior estimates of morphological rates are then integrated into codon-based evolutionary analyses to test whether branches with accelerated morphological evolution also show evidence for positive selection at specific codon sites. By combining dense taxon sampling with branch-resolved morphological and molecular analyses, this framework aims to identify coding-sequence changes most plausibly associated with historical shifts in beak morphology, and ultimately to inform how microevolutionary processes translate into macroevolutionary patterns of beak diversification.

[A178] Structure-guided secretome analysis in a fungal plant pathogen provides insights into effector evolution and functional diversification

Thais Dal'Sasso[1,2], Prof. Dr. Eva Stukenbrock[1,2]
[1]Environmental Genomics, Christian-Albrechts University of Kiel, Kiel, Germany, [2]Max Planck Institute for Evolutionary Biology, Plön, Germany

Fungal plant pathogens secrete effector-like proteins that alter host cell physiology and may target plant-associated microbes. However, rapid evolution and low sequence conservation hinder the characterization of these proteins. The phytopathogenic fungus Zymoseptoria passerinii infects Hordeum spp. and its lineages are adapted to wild or domesticated barley. Understanding adaptive evolution in these secreted proteins may shed light on ecological interactions that drive host specialization. We combined structure-based network analysis to unravel the secretome of Z. passerinii. We identified 72 structural clusters, revealing fold-level relationships across divergent sequences. By integrating functional annotations, we showed that effector-like proteins with host immune-interfering functions evolved from a limited set of protein folds, whereas proteins with predicted antimicrobial properties have evolved from diverse fold groups. In several cases, immune-interfering effectors and antimicrobial proteins shared similar protein folds. Physicochemical comparisons indicate that antimicrobial effectors predominantly emerged through amino acid replacements on common effector-enriched scaffolds in Z. passerinii, reconfiguring surface charge and electrostatics. To contextualize these patterns, we analyzed intra- and interspecific structural variation in selected effector-enriched families by comparing Z. passerinii proteins to homologs in the sister species Z. tritici. Homologs retained highly similar core folds but differed locally in loops and surface-exposed regions, consistent with conserved scaffolds supporting diversification of surface properties. Overall, we demonstrate how the secretome of Z. passerinii is built around common structural folds that support diverse biological roles, and we propose that conserved scaffolds combined with physicochemical variation contribute to effector evolution in this species.

[A179] Ancient genomics support deep divergence between Eastern and Western Mediterranean Indo-European languages

Fulya Eylem Yediay[1,2], Tharsika Vimala[1,3], Guus Kroonen[4,5], Serena Sabatini[2], Martin Sikora[1], Rasmus Nielsen[1,3], Kristian Kristiansen[1,2], Eske Willerslev[1,6]
[1]Lundbeck Foundation GeoGenetics Center, Globe Institute, University of Copenhagen, Copenhagen, Denmark, [2]Department of Historical Studies, University of Gothenburg, Gothenburg, Sweden, [3]Departments of Integrative Biology and Statistics, UC Berkeley, Berkeley, USA, [4]Leiden University Centre for Linguistics, Leiden University, Leiden, The Netherlands, [5]Department of Nordic Studies and Linguistics, University of Copenhagen, Copenhagen, Denmark, [6]GeoGenetics Group, Department of Zoology, University of Cambridge, Cambridge, UK

Genomic studies have established that the expansion of Steppe pastoralists during the Bronze Age profoundly shaped European populations, both genetically and linguistically. Current evidence links the spread of Indo-European languages to migrations of Steppe pastoralists starting around the fifth millennium before present (BP). However, the question of how these different Steppe populations relate to the branching of the Indo-European languages spoken along the northern border of the Mediterranean remains contested. Central to this debate is whether Greek clusters with Indo-Iranian or Armenian, and whether Italic and Celtic languages form a single branch. To address these hypotheses, we generated whole-genome data from 379 ancient individuals from across Southern and Central-Eastern Europe and the Eastern Mediterranean (5200–2100 BP). By applying an identity-by-descent-based mixture modelling framework to a dataset composed of ~9,000 ancient genomes, we trace the expansion of Steppe pastoralists into Southern Europe and the Mediterranean. We find that Bell Beaker populations carried Steppe ancestry into Italy, France, and Spain, likely giving rise to the Italic and Celtic languages. In the eastern Mediterranean, Greek and Armenian populations trace their Steppe ancestry to Yamnaya groups, pointing to a common Steppe source for Greek and Armenian speakers. Steppe ancestry in present-day Cyprus, meanwhile, traces to mainland Greece, possibly through Greek settlers who arrived on the island following the Bronze Age collapse. Our results thus offer genomic support for two long-debated models of Indo-European diversification.

[A180] Detecting polygenic selection signatures using tree sequences statistics

Thémis Lemarchand[1], Maud Tenaillon[1], Dr Fanny Pouyet[2], Maud Fagny[1]
[1]GQE-Le Moulon - Université Paris-Saclay, INRAE, CNRS, AgroParisTech, Gif-sur-Yvette, France,
[2]Université Paris-Saclay, CNRS, Laboratoire Interdisciplinaire des Sciences du Numérique, Gif-sur-Yvette, France

Polygenic selection is an important mechanism of the adaptation of organisms to their environment. Nevertheless, it leaves such weak signatures in the genome that most classical statistics only have limited power. Furthermore, most existing approaches try to mitigate this lack of power by either requiring prior knowledge of environmental constraints or by grouping loci based on prior biological knowledge, thus limiting their scopes to known molecular bases of quantitative traits, and/or biological pathways. Recently, estimation of coalescent time based on ancestry recombination graph inferences proved efficient to detect weaker selection signals. This suggests that, among the wide range of tree-based statistics, some might detect yet untapped molecular signatures of polygenic selection. However, their power has never been properly evaluated in this context. In this study, we thus aimed to develop an efficient method to detect signatures of polygenic selection without providing a priori information.

To this end, we simulated different polygenic selection scenarios using the forward-in-time SLiM software. We then compared the power of several summary statistics measuring tree imbalance with those of statistics measuring population differentiation, allele frequencies spectrum changes, or increased haplotype homozygosity.

We showed that tree imbalance is a surprisingly robust index for detecting polygenic selection, even under weak selective pressures, and is twice as powerful as FST, which best detects signatures of relatively high selective pressures. We then proposed an integrated score, that we applied on human genomic data from the 1000 Genomes Project and uncovered known and new selection signals.

[A181] Repeat evolution of cephalic horns in vipers

Theo Busschau[1], Stephane Boissinot[1]
[1]New York University Abu Dhabi, Abu Dhabi, United Arab Emirates

Horn-like cephalic appendages evolved under various selective pressures among squamate reptiles. Yet, among snakes, where horns are most abundant in vipers, the functional and evolutionary significance of horns are poorly understood. Using a comparative phylogenetic approach, we showed that horns evolved multiple times independently with frequent fluctuations in the extent of character states as lineages diversified. To understand the genomic basis of horns, we use the desert vipers (Cerastes Sp.) as a model system. The genus comprises three species, Cerastes cerastes and C. gasperettii and C. vipera, in which supraocular horns are polymorphic - horned and hornless phenotypes are present across all species. Genome-wide association show that horn polymorphism across species is significantly associated with a single 18kb genomic region among genes associated with head and eye phenotypes in other organisms. Using short- and long-read data we identified species- and phenotype-specific structural variants in this region. In Cerastes the hornless phenotype evolved once with a genus-specific tandem duplication in a conserved non-coding element, followed by at least two secondary gains of the horned phenotype associated with independent losses of the tandem duplication. Divergence and diversity statistics around the polymorphic tandem duplication suggest horn polymorphisms across species were maintained by long-term balancing selection, although in the absence of trans-species polymorphism.

[A182] Phenotypic and transcriptomic plasticity in the medaka gut in response to food availability

Tokiho Akiyama[1], Mizuki Hayashi[1], Motoyuki Ogawa[1], Takafumi Katsumura[1,2]
[1]Kitasato University School of Medicine, Sagamihara, Japan, [2]Kyushu University, Fukuoka, Japan

Phenotypic plasticity is considered to facilitate adaptive evolution, a process known as plasticity-led evolution. Previously, we reported that the medaka fish (Oryzias latipes) shows seasonal plasticity in gut length, which has undergone plasticity-led evolution. Medaka gut exhibits longer in summer but shorter in winter. However, the environmental cues triggering this plasticity remain unclear. In this study, we tested whether variation in food availability, which fluctuates seasonally, induces phenotypic plasticity in gut of medaka. We first compared three experimental groups using the laboratory strain d-rR/TOKYO adult medaka aged 3.5–4 months: a control group fed twice daily, and two treatment groups fed either once every two days or four times daily. Gut length and content volume were measured weekly from two to four weeks after the experiment began. Although feeding conditions did not clearly influence gut length, gut content volume varied significantly among treatment groups. Notably, the high-feeding group (four times daily) showed lower gut content, whereas the low-feeding group (once every two days) exhibited higher gut content, implying that food availability induces plasticity in gut storage capacity, which may represent a rapid physiological response preceding the slower morphological plasticity in gut length. Therefore, we next performed comprehensive gene expression analysis (bulk RNA-seq) on the gut and whole brain to identify the molecular mechanisms underlying this plasticity. We will discuss how phenotypic plasticity in response to feeding regime involves molecular changes in multiple tissues and consider its evolutionary significance in the context of plasticity-led evolution in natural populations.

[A183] Genetic basis of innate vocalization patterns in chickens

Tsuyoshi Shimmura[1], Yuki Matsuda[1], Miku Shimamori[1], Shingo Miyakawa[2], Mei Matsuzaki[3]
[1]Tokyo University Of Agriculture And Technology, , Japan, [2]Gifu University, , Japan, [3]Hiroshima University, , Japan

Animals communicate using species-specific sounds, such as barking in dogs, meowing in cats, and croaking in frogs. Many of these vocal patterns are innate, even in chimpanzee, and do not require learning, indicating that vocalization is under genetic control across the animal kingdom. However, molecular mechanisms underlying innate vocal patterns remain largely unknown. Here, we explored the genetic basis of innate vocalization using an indigenous chicken population bred on Okinawa Island, Japan, which exhibits a unique crowing pattern. An integrated approach combining quantitative trait locus (QTL) mapping in a backcross population (n=240, all male) derived from normal and unique crowers, together with a genome-wide population branch statistic (PBS) screen using whole-genome resequencing data (n=110), identified four QTL regions containing three candidate genes. The two of these genes are functionally related to axonal guidance, and one gene harbored nonsynonymous SNPs. Transcriptomic analyses of tissues involved in vocalization, including the nucleus of the brain (midbrain and diencephalon) and the syrinx, revealed no differential expression of the candidate genes between normal and unique crower. This suggests that these genes may exert their effects during earlier developmental stages rather than during vocal production itself. To further elucidate the functional roles of these candidate genes, we are conducting in vitro axonal guidance assay, immunohistochemistry screening, in vivo gene overexpression and knockdown experiments, and phenotypic analysis using genome-edited mice carrying the chicken-derived SNP alleles. The results will be presented at the congress.

[A184] Phylogenomic analyses of transposable elements in simulated populations to disentangle past demographic and transpositional events

Valentin Grenet[1], Yann Bourgeois[1]
[1]Institut De Recherche Pour Le Développement - Délégation Occitanie, Montpellier, France

Polymorphisms and the abundance of transposable elements (TEs) within a genome are influenced by numerous mechanisms involving the TEs themselves and their transposition mechanism, as well as the dynamics of the host population. Demography, through genetic drift, affect the fixation of TEs within a population, thereby contributing to the maintenance of TE diversity. Thus, demographic events have often been considered the cause of large variations in transposition dynamics, leading to bursts or declines in transposition. However, it is unclear how host demography mechanistically influence TE transposition dynamics, if at all. It is therefore difficult to determine whether past demographic events that caused changes in TE diversity really involve variations in transposition dynamics, or whether stochastic mechanisms such as genetic drift are the main processes influencing TE diversity in these cases.

This study aims to determine whether it is possible to identify and differentiate past demographic events from events related to variations in transposition dynamics. It is based on phylogenomic analyses of TEs in simulated populations of genetically simplified individuals using the SLiM software package. The populations consist of a thousand individuals with a specified number of fixed transposable elements in the initial generation, and the simulations go through demographic events (bottlenecks, expansions) and/or transposition events (explosions, declines). Tree sequences are obtained at the end of the simulations, helping estimate the frequency of transposable elements and the effective population size in order to recover past events.

[A185] Evolution of transposable element activity, structural variants and SNPs upon hybridisation

Valentina Peona[1]
[1]Swedish Natural History Museum, , Sweden

Hybridisation can be a facilitator of adaptive evolution. However, hybridisation can also unmask genetic conflicts and cause hybrid incompatibilities and hybrid inviability. Transposable elements (TEs) are one of the main sources of genetic conflicts and hybrid incompatibilities since they can reactivate if they are not being properly silenced. Using an extensive phased genomic dataset of ~300 wheatears (Oenanthe, Aves), I investigate the evolution of TE activity, other structural variants and of mutation rate spectra across a hybrid zone characterised by a clear ancestry continuum. Despite the parental species having evolved a different landscape of active TEs, the hybrids do not suffer from an uncontrolled TE activity or a transgressive methylation patterns. However, the comparison of mutation rate spectra of SNPs shows hybrids and parental species to have diverging spectra that evolved in only 700,000 years. Therefore, in genomes of bird species that recently split like wheatears, mutation rate of SNPs resulted to be the fastest evolving type of mutation.

[A186] Assessing the fitness of HIV-1 drug target protein variants through dynamical properties

Valeria Zoni[1,2], David Ferreiro[1,2], Sara Cabaleiro-Martínez[1,2], Dr Miguel Arenas[1,2]
[1]University Of Vigo, Vigo, Spain, [2]CINBIO, Vigo, Spain

The high mutation rate of HIV-1 has so far hindered the development of effective vaccines and promotes the emergence of drug-resistance mutations against antiretroviral therapies. Understanding how resistance mutations arise and become fixed in viral populations is essential for the design of effective treatments. Although resistance mutations can confer a fitness advantage under drug pressure, they may also compromise protein folding stability or alter binding to natural substrates, thereby hindering their ability to become fixed in the viral population. Characterizing this balance is crucial for predicting which variants are more likely to be maintained in the population. In this study, we investigate the impact of common resistance mutations in clinically observed variants on the folding stability and binding affinity of HIV-1 drug target proteins using molecular dynamics simulations. By accounting for protein conformational ensembles rather than relying on single static structures, we aim to obtain a more realistic description of mutation induced effects on protein function. Overall, this work aims to contribute to the construction of protein fitness landscapes that incorporate protein dynamics, improving our understanding of the emergence and establishment of drug-resistant HIV-1 variants and supporting the development of more effective antiviral strategies.
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Keywords: Protein evolution, protein folding stability, molecular dynamics simulations, protein interactions, protein fitness

[A187] National bioinformatics support for Evolution and Biodiversity research projects at Swedish universities

Verena E. Kutschera[1], Allison M. Churcher[2], André E. R. Soares[3], Cormac M. Kinsella[4], Elisavet Torstensson[4], Estelle Proux-Wéra[1], Guilherme Dias[4], Jason Hill[4], Magnus Alm Rosenblad[5], Mahesh Binzer-Panchal[3], Martin Pippel[4], Miguel A. Redondo[4], Dr. Nikolay Oskolkov[6], Per Unneberg[4], Stephan Nylinder[4], Tomas Larsson[4], Yvonne Kallberg[1], Muhammad Bilal Sharif[7], Qiaoling Deng[8,9], Andreas Wallberg[10], Leif Andersson[8,11], Love Dalén[7,12], Henrik Lantz[3], Lucile Soler[3]

[1]Department of Biochemistry and Biophysics, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Stockholm University, Solna, Sweden, [2]Department of Plant Physiology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Umeå University, Umeå, Sweden, [3]Department of Medical Biochemistry and Microbiology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Uppsala University, Uppsala, Sweden, [4]Department of Cell and Molecular Biology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Uppsala University, Uppsala, Sweden, [5]Department of Chemistry and Molecular Biology, National Bioinformatics Infrastructure Sweden, Lundberg laboratory, University of Gothenburg, Gothenburg, Sweden, [6]Department of Biology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Lund University, Lund, Sweden, [7]Centre for Palaeogenetics, Department of Zoology, Stockholm University, Stockholm, Sweden, [8]Department of Medical Biochemistry and Microbiology, Science for Life Laboratory, Uppsala University, Uppsala, Sweden, [9]The Key Laboratory of Aquatic Biodiversity and Conservation of Chinese Academy of Sciences, Institute of Hydrobiology, Chinese Academy of Sciences, Wuhan, China, [10]Department of Medical Biochemistry and Microbiology, Uppsala University, Uppsala, Sweden, [11]Department of Veterinary Integrative Biosciences, College of Veterinary Medicine and Biomedical Sciences, Texas A&M University, College Station, USA, [12]Department of Bioinformatics and Genetics, Swedish Museum of Natural History, Stockholm, Sweden

The National Bioinformatics Infrastructure Sweden (NBIS) comprises about 120 experts from different research fields, providing bioinformatics and data science support and training to the Swedish life science community. About twenty bioinformaticians and data stewards are supporting research projects from the fields of evolution and biodiversity at Swedish universities. In 2025, the Evolution and Biodiversity team supported 50 active research projects and co-authored 18 peer-reviewed publications. Here we present three of these projects. 1) The genome assembly and annotation of the European cisco (Coregonus albula), a polyploid salmonid fish adapted to fresh and brackish water, was based on PacBio HiFi long read sequencing and Hi-C scaffolding. 2) One of the largest genomes assembled and analyzed by our team was the 19 Gb genome of the Northern krill (Meganyctiphanes norvegica), which revealed insights into ecological adaptation of this large and abundant zooplankton species. 3) The DNAharvester pipeline was developed in Nextflow to process and analyze highly degraded ancient and historical DNA samples, and combines multiple filtering and data-processing strategies within a modular framework. The Evolution and Biodiversity team is not only supporting research projects with hands-on bioinformatics analyses, but is also providing knowledge transfer to researchers, thereby empowering them to address research questions with implications for the biodiversity and climate crises with data science and bioinformatics approaches. NBIS employs their staff permanently, and provides thereby not only continuous bioinformatics support to the Swedish life science community, but also an alternative to the traditional academic career path.

[A188] Comparative investigation of style length and female self-incompatibility reaction mediated via the brassinosteroid pathway in *Linum*

Viktoria Bednarski[1], Aleksandra Losvik[1], Tanja Slotte[1]
[1]Stockholm University, Stockholm, Sweden

Distyly is a classic example of a balanced polymorphism governed by a supergene. In distylous species, reciprocal herkogamy and a heteromorphic self-incompatibility (SI) system promote disassortative mating between floral morphs. While genomic studies have shed light on the genetic architecture of distyly supergenes, in most systems the genetic and physiological basis of floral morph differences and SI remains incompletely understood. Here, we characterize the genetic and physiological underpinnings of style length and female SI reaction in the genus *Linum*, a system where Darwin described distyly. In *Linum*, two candidate genes for distyly are present and conserved at its distyly supergene: *TSS*1 and *WDR*-44. *TSS*1 has been hypothesized to regulate both style length and female SI reaction via the brassinosteroid pathway, but experimental support for a role in SI is lacking. Here, we test this hypothesis experimentally, through a comprehensive set of experiments involving treatment of flower buds with brassinosteroid hormone and inhibitor, followed by morphological measurements and assessment of SI. We additionally test whether the effect of *TSS*1 on style length and female SI is conserved across divergent *Linum* species. Our results provide comparative insight into the physiological pathways underlying distyly, and will shed light on whether the same gene governs both style length differences and female SI reaction, a scenario with major implications for the evolution and loss of distyly.

[A189] Using Hyb-Seq (target capture sequencing) to construct a phylogenetic framework for a tropical plant family, the Calophyllaceae

Wei Wei Seah[1,2,3], Prof.dr. Peter van Welzen[1,2], Dr. Gillian Khew[3], Dr. Rita Pucci[1,2,4], Dr. Rafaela Trad[5]
[1]Naturalis Biodiversity Center, Leiden, Netherlands, [2]Leiden University, Leiden, Netherlands, [3]Singapore Botanic Gardens, National Parks Board, Singapore, Singapore, [4]Leiden Institute of Advanced Computer Science, Leiden, Netherlands, [5]Institute of Biosciences, São Paulo State University (UNESP), Câmpus de Botucatu, Botucatu, Brazil

The pantropical Calophyllaceae is a member of the extremely diverse plant order Malpighiales. Past studies of the family did not include all 14 genera and used only a few genes. They provided evidence for the distinctness of two Indo-Pacific genera, Mesua and Kayea, previously thought to be congeneric owing to perceived overlapping locule number and intergrading stigma shape, as the type species of Mesua was consistently found in a different clade from various species of Kayea. However, they did not clarify the relationships between all genera as some genera were recovered in different clades with varying levels of support. Hence, this study adopted a broad sampling approach based mainly on herbarium specimens and used the Hyb-Seq (target capture sequencing) method to construct a phylogenetic framework that can be used to further examine the evolutionary history and relationships within the family. We sampled a total of 146 Calophyllaceae taxa, representing all genera in the family for the first time. Additional emphasis was made to sample most of the species from Mesua and Kayea to determine species circumscriptions for taxonomic revision. Cratoxylum arborescens (Hypericaceae) was chosen as an outgroup. The phylogenetic tree will provide the foundation to think about the evolution and biogeography of the family. Additionally, it will be used together with specimen images of some species to explore if existing Artificial Intelligence (AI) models are able to extract features that effectively differentiate the species and whether these features can be understood and applied in taxonomic revision to improve the efficiency.

[A190] Continent Surfing: The First Molecular Phylogeny of A Gondwanan Isopod Suborder

William Farris[1], Ethan Kahn[1], Dr. Regina Wetzer[1]
[1]Natural History Musuem Of Los Angeles County, Los Angeles, United States

Phreatoicidea is a suborder of laterally compressed isopods (Crustacea, Peracarida) that first appear in the fossil record and become widely distributed in marine habitats during the Carboniferous period 300-325 MYA. Today, there are 114 described extant species in 6 families that are restricted to freshwater systems, including subterranean aquifers, groundwater-fed streams, springs, seepages, and even muddy road ruts. These extant members are restricted to geographic regions that comprised Gondwanaland in the early Jurassic (200 MYA). Although morphological phylogenies supported a basal relationship to all other isopods, some recent molecular phylogenies instead nest Phreatoicidea with other Isopoda, proving the value of molecular-based analyses. Despite their importance in understanding deeper phylogenies of Isopoda, relationships within the suborder Phreatoicidea have not been addressed with molecular data. For instance, one morphological phylogeny proposed that Pilbarophreatoicus in northwestern Australia is more closely related to two Indian genera than to many other Australian Phreatoicidea. Currently, no molecular data is available to test this claim. This project combines publicly available sequences with new 18S rRNA, 16S rRNA, and Cytochrome Oxidase 1 (COI) data from extant species to construct the most robust molecular phylogeny to date, shedding light on the origins of the current distribution. This effort provides a molecular backbone for the suborder, the first step toward understanding the isolation and radiation of a once ubiquitous taxon.

[A191] Comparative genomics of phylum Tardigrada, based on single-specimen genomes

Witold Morek[1], Lewis Stevens[1], Łukasz Michalczyk[2], Mark Blaxter[1]
[1]Wellcome Sanger Institute, Hinxton, Cambridge, United Kingdom, [2]Jagiellonian University, Kraków, Poland

Tardigrades, commonly known as water bears are a phylum of microscopic animals renowned for their exceptional cryptobiotic abilities. Despite that, the genomic data for these animals remain scarce due to technical challenges associated with their minute size and the lack of stable laboratory cultures. Newly developed methods, such as Picogramme input Multi-modal Sequencing (PiMmS), represent a breakthrough, enabling the sequencing of previously 'unsequenceable' single-specimen meiofauna.

In this study, we used the PiMmS method and highly accurate long-read PacBio HiFi sequencing to generate genomic data for 13 tardigrade species, representing three orders. To facilitate accurate gene prediction, we additionally sequenced transcriptomes (mRNA) from the same individuals. Our analyses focus on comparative genomics and differences in the protein-coding gene repertoires among major tardigrade lineages.

We generated highly contiguous and biologically complete genome assemblies for all species, with contig N50 values ranging from 66 kb to 8.6 Mb (mean 1.7 Mb). We identified between 13,500 and 25,000 protein-coding genes per genome. Orthogroup inference and functional annotation revealed clear phylogenetic differentiation among the three orders, including lineage-specific and tardigrade-specific proteins associated with anhydrobiosis.

Our study demonstrates that PiMmS enables the generation of high-quality reference genomes for tardigrades from a single specimen, with quality comparable to or exceeding genomes derived from bulk laboratory cultures. This offers an opportunity to revolutionise the field of meiofauna genomics.

[A192] What can we learn about speciation from high levels of allele sharing at loci subject to strong balancing selection?

Xavier Vekemans[1], Celine Poux[1], Vincent Castric[1]
[1]University of Lille, , France

Loci subject to strong multiallelic balancing selection often show high levels of trans-species polymorphisms. As suggested by Naoyuki Takahata, knowledge about the number of shared alleles between closely related species may provide quantitative predictions about the minimum number of individuals contributing to a nascent lineage, providing a posteriori information about the demographical context of speciation. In plants, loci hosting the molecular determinants of self-incompatibility systems (S-loci) are characterized by high levels of allelic diversity, due to strong negative-frequency dependent selection acting primarily on the male fitness. We developed bioinformatic approaches to genotype the S-locus from short reads whole-genome resequencing data. Here we present results from almost exhaustive estimates of allelic diversity specieswide and of allelic sharing among related species of Brassicaceae. Our results suggest long-term maintenance of high numbers of ancestral allelic lineages, which would not be compatible with scenarios of speciation with founder effects. One exception, however, corresponds to speciation events associated with allopolyploidy, which systematically break down the self-incompatibility system in Brassicaceae, leading to strong reduction in allelic diversity. In contrast to plant self-incompatibility, the Complementary Sex Determinaing (CSD) gene in honeybees does not show evidence for trans-species allelic sharing, despite very high within-species allelic diversity.

[A193] Why are most of the methods detecting natural selection wrong? And how to detect natural selection properly?

Xianfa Xie[1]
[1]Virginia State University, Petersburg, United States
For the last four decades, detecting natural selection from molecular and genomic data has been a major pursuit in the field of evolutionary biology. A variety of methods have been developed and an extraordinary number of cases claiming the detection of natural selection have been published. However, careful analyses of these methods and those published cases reveal fundamental problems with the underlying approaches and the claims. This presentation will analyze the strengths and weaknesses of the existing methods to detect natural selection and present a clear guideline and integrated approach on how to detect natural selection from molecular and genomic data by taking into consideration of a variety of evolutionary processes affecting the genetic polymorphism and genomic diversity.

[A194] Uncovering the selection on non-homologous end joining and homologous recombination for genome downsizing in polyploid plants

Xiaotong Wang[1,2,3], P Cao[3], Kong Ci[3], Lv Xiangguang[3], IJ Leitch[1], Leitch AR[2]
[1]Royal Botanic Gardens, , United Kingdom, [2]Queen Mary University of London, , United Kingdom, [3]Beijing Life Science Academy, , China

Genome size (GS) variation across angiosperms is shaped by recurrent polyploidy, yet most species exhibit smaller genomes than expected given their polyploid ancestry, suggesting widespread genome downsizing. As the rate of DNA loss following polyploidy is relatively slow, the mechanisms underlying this process remain unclear. Here, we investigate the potential role of double-strand break (DSB) repair pathways in driving genome downsizing. We combined large-scale GS reconstruction across >10,000 plant species with comparative genomic and transcriptomic analyses. First, we reconstructed GS evolution to estimate rates of genome size change following the most recent polyploidy event. Second, we identified orthologous genes involved in DNA repair and recombination pathways and applied linear modelling to assess how gene copy number and functional characteristics relate to GS variation. Third, we analysed gene expression patterns in diploid–polyploid complexes of Gossypium to examine how pathway activity changes with polyploid age. Our results show that genome downsizing is widespread following polyploidy, with GS decreasing over time and more rapidly at higher ploidy levels. Gene copy numbers in DNA repair pathways decline with increasing polyploid age, with the Ku70–Ku80 heterodimer showing a significant association with GS reduction. Expression analyses further indicate that homologous recombination (HR) pathways are upregulated in young polyploids and likely contribute to genome downsizing, potentially through the removal of LTR retrotransposons. In contrast, classical non-homologous end joining (c-NHEJ) activity is associated with indel accumulation rather than genome contraction.

[A195] Brain Evolution in Lungfish: Uncovering Tetrapod Adaptations During the Water-to-Land Transition

Shunping He, Chuanjiang Ma, Xiaoyu Meng, Haixu Wu, Yongjie Chen, Bing Su

The transition from water to land was a pivotal evolutionary step that enabled vertebrate radiation and tetrapod diversification, with the brain playing a central role in environmental adaptation. To investigate the origins of vertebrate brain organization, we studied the African lungfish (Protopterus annectens), the closest extant relative of tetrapods. We generated a high-quality brain annotation based on long-read transcriptomic data and profiled over 60,000 nuclei to construct a comprehensive cell-type atlas using single-nucleus RNA sequencing (snRNA-seq) and single-nucleus ATAC sequencing (snATAC-seq). Integrative analyses with existing datasets revealed conserved cytoarchitectural and molecular features across sarcopterygians. Notably, we identified a conserved pallial organization in lungfish comparable to that of tetrapods. We further uncovered evidence of incipient telencephalic cortical layering, characterized by spatially segregated neuronal populations and graded gene expression patterns, suggesting that key features of cortical organization predate tetrapod divergence. Together, these findings provide new insights into the evolution of vertebrate brain structure during terrestrialization.

[A196] Independent origin of genomic imprinting in placental fish

Xu Wang[1], Ying Zhang[1], Polly Campbell[2], Alec Kingsley[3], Yuridia Reynoso[2], Yuan Kang[1], Chen-Che Jeff Huang[1], John H. Werren[3], David Reznick[2]
[1]Auburn University College Of Veterinary Medicine, Auburn, United States, [2]University of California, Riverside, Riverside, United States, [3]University of Rochester, Rochester, United States
Parent-of-origin genomic imprinting results in monoallelic, often tissue-specific, gene expression. This non-Mendelian expression is characteristic of angiosperms and placental mammals, where a paternally derived genome is active in specialized tissues involved in maternal provisioning (e.g., endosperm and placenta). Because imprinting forfeits the benefits of diploidy, an evolutionary mystery is what selective advantages compensate for this enforced haploidy. The most widely accepted hypothesis is that imprinting results from parental conflict between maternal control of resource allocation and paternal control of resource acquisition. If true, then imprinting should evolve in organisms that independently evolved placentas. Here we show genomic imprinting in a placental poeciliid fish, Poeciliopsis prolifica. We performed reciprocal crosses between two inbred lines and performed transcriptomic analysis on embryonic and placental tissues. Single nucleotide polymorphisms with fixed differences between the inbred lines enable us to identify parent-of-origin dependent biases in allelic expression. Among more than 700 transcripts with consistent parent-of-origin allelic bias in reciprocal crosses, we identified 84 protein-coding genes with robust evidence of imprinting in the P. prolifica placenta, majority of which are not imprinted in embryotic somatic tissues. The newly discovered fish imprinted genes include the teleost ortholog of mammalian imprinted gene, grb10, and a key regulator of glucose metabolism in human placenta, dpp4. Our discovery of imprinting in a placental teleost is the first demonstration of genomic imprinting in a non-mammalian vertebrate and supports an association between imprinting and the evolution of specialized tissues for maternal embryonic provisioning.

[A197] A Comparative TE Annotation Framework for Bees

Xuejing Hu[1], Dr Eckart Stolle[1], Manwen Su[1]
[1]Leibniz Institute for the Analysis of Biodiversity Change(LIB) , Museum Koenig Bonn， , , Germany

Transposable elements (TEs) represent a major component of insect genomes and play important roles in shaping genome structure and evolution. However, comparative studies across bees remain limited by inconsistent TE annotation and lack of consistent and standardized approaches for comparative TE analysis across species.

To address this, we developed a unified comparative TE annotation framework for bees (Apoidea) based on de novo TE discovery across the genomes of 250 bee species. Using Earlgrey and MChelper, we performed genome-wide TE identification, clustering, and consensus sequence construction, generating a non-redundant and standardized TE annotation library that enables cross-species comparability of TE content and genome annotation.

We applied this framework to quantify per-genome normalized TE burden across bee species and further integrated these data with gene loss estimates inferred using TOGA2. Our analyses reveal substantial variation in both TE burden and gene loss across bee lineages. We further explore their distribution in a phylogenetic context to assess potential associations across the bee tree of life.

This study establishes a scalable comparative framework for TE annotation in bees and provides a foundation for integrative analyses of genome evolution and repeat dynamics in insects.

[A198] Trimmiy: Removing erroneously aligned columns from multiple sequence alignments for improved phylogenetic inference.

Yi Liu[1], Dr Laurence Belcher[1], Dr Jonathan Holmes[1], Prof Steven Kelly[1]
[1]University Of Oxford, Oxford, United Kingdom

Multiple sequence alignments are a key component of comparative genomics, supporting both homology assessment and phylogenetic inference. However, unaligned regions and non-phylogenetically informative sites can hinder accurate sequence comparison and affect downstream analyses. Here, we describe Trimmiy, a trimming method for multiple sequence alignments. Trimmiy implements a general trimming framework in which alignment columns are scored using information derived from the structure of the alignment, yielding a flexible and computationally efficient filtering strategy suitable for large-scale phylogenomic analyses. In analyses of single-copy orthologs, trimming with Trimmiy is associated with reduced phylogenetic tree inference error. Future work will extend the framework to additional trimming criteria and benchmarking datasets, and examine its effects on substitution model estimation and downstream evolutionary analyses.

[A199] Genomic diversity and local adaptation in the native range of the global pest Drosophila suzukii

Ying Zhen[1]
[1]Westlake University, Hangzhou, China

Understanding how invasive species adapt despite severe bottlenecks requires characterization of genetic variation in their native ranges. The global spread of Drosophila suzukii from East Asia has been rapid and economically devastating, yet the genomic architecture of its native populations remains poorly resolved. Here, we present whole-genome sequences from 178 D. suzukii isofemale lines sampled across China—the most comprehensive native-range dataset to date. We reveal moderate longitudinal population structure and identify large polymorphic inversion, which is strongly associated with altitude, longitude, and isothermality. This inversion also contributes significantly to population differentiation and shows signatures of local adaptation in subpopulations, despite ongoing gene flow. Integrating native and invasive genomes, we show that adaptive loci in invasive populations are enriched for alleles from standing genetic variation, with divergent selection acting on different invasive populations. Demographic inference reveals strong bottlenecks in invasive populations, while native populations have remained stable following ancient expansion. Admixture analyses indicate multiple admixture events have reshaped invasive genomes, but recent intercontinental gene flow is limited.

[A200] Molecular basis of aggressive behavior in chickens

Yohei Yamada[1], Yuki Matsuda[1], Hiroko Tozawa[2], Masaoki Tsuduki[3], Tsuyoshi Shimmura[1]
[1]United Graduate School of Agricultural Science, Tokyo University of Agriculture and Technology, Fuchu, Japan, [2]Faculty of Agriculture, Tokyo University of Agriculture and Technology, Fuchu, Japan, [3]Graduate School of Integrated Sciences for Life, Hiroshima University, Higashi-Hiroshima, Japan

The earliest stages of animal domestication are thought to have been primarily driven by the selection on behavior. However, the molecular mechanisms underlying aggressive behavior and their modification during domestication remain poorly understood. Chickens represent one of the most intensively domesticated animal species, and modern chicken breeds can be behaviorally classified into two groups: populations that remain at high levels of aggressiveness, reflecting cultural preferences and ancestral traits of the red junglefowl, and populations in which aggressiveness has been reduced through selection for productivity. Using chicken populations with distinct levels of aggressiveness, we aimed to elucidate the molecular mechanisms underlying aggressive behavior by applying an integrative approach combining genomics, transcriptomics, and spatial gene expression analyses. Population Branch Statistics (PBS) based on whole-genome resequencing of 241 individuals identified 20 candidate genes located in highly differentiated genomic regions, including a gene encoding a key enzyme in the serotonin metabolic pathway. This gene harbored a single missense mutation that showed strong population differentiation and was predicted to induce a substantial structural alternation of the enzyme. Transcriptomic analysis of the cerebrum, using RNA-sequencing of 20 individuals, also identified 20 differentially expressed genes, four of which were involved in the tryptophan metabolism pathway. Together, these genomic and transcriptomic findings indicate that serotonergic signaling plays a central role in modulating aggressive behavior in chickens. Our results further suggest that serotonergic regulation of aggression is evolutionarily conserved and has been modified during the domestication process.

[A202] A genome-wide association study (GWAS) reveals the molecular basis of body coloration and morphological traits in ornamental common carp (Nishikigoi)

Yoshihiro Omori[1], PhD Soma Tomihara, MD, PhD Tetsuo Kon, MD, PhD Koto Kon-Nanjo, PhD Hideki Noguchi, PhD Kohji Mabuchi, Atsushi Toyoda
[1]Hiroshima University, Higashi-hiroshima, Japan

Ornamental common carp, Nishikigoi (Cyprinus carpio) have a history of more than 250 years in Japan. They exhibit remarkable phenotypic diversity, including a wide range of body colors and complex color patterns such as red, white, and yellow. In addition to body color diversity, several morphological variations are observed in scales and fins. However, the genetic and molecular mechanisms underlying these phenotypic traits remain largely unexplored.
In this study, we performed whole-genome resequencing of 50 individuals to obtain a comprehensive catalog of genomic variation. We further conducted genome-wide association studies (GWAS) using approximately 117 individuals representing about 40 strains to identify loci associated with ornamental traits. GWAS analyses were carried out for 21 phenotypes, including the Hikari phenotype related to iridophore development and reflective scale formation. These analyses identified multiple significant candidate loci potentially responsible for pigmentation patterns and skeletal morphology.
In addition, transcriptome analyses of 22 individuals provided insights into gene expression profiles and gene networks involved in body color determination and skeletal formation. Together, our results provide a genomic framework for understanding the genetic basis of phenotypic diversification in Nishikigoi. We also discuss how these findings compare with previous genomic studies of other ornamental fish species, such as medaka and goldfish to highlight both shared and lineage-specific mechanisms of trait evolution.

[A203] Early-onset colorectal cancer-associated driver oncogenes revealed by molecular evolutionary analysis.

Yosuke Seto[1], Satoshi Nagayama[2], Ryohei Katayama[1]
[1]Division of Experimental Chemotherapy, Japanese Foundation for Cancer Research, , Japan, [2]Uji-Tokusyukai Medical Center, , Japan

Colorectal cancer (CRC) is the second leading cause of cancer death in the world. The developmental process of CRC is known as multistep carcinogenesis, explained by tumor initiation, tumor promotion, malignant conversion, and tumor progression, caused by mutations occurring on tumor suppressor genes and driver oncogenes over decades. Recently, the increasing incidence rate of colorectal cancer in younger people has become a growing concern. However, the molecular characteristics of these early-onset colorectal cancer (EOCRC) remain poorly understood. In this study, we investigated the mutational landscapes of patients diagnosed with sporadic CRC from 23 to 90 years old and found that the EOCRC had more frequent mutations in NF1, RAF1, and SMAD4. Furthermore, we developed a new method based on evolutionary theory to identify EOCRC-specific driver oncogenes, named SPEC (Selection Pressure Estimation for Cancer). SPEC detected positive selection on NF1, RAF1, and SMAD4 and on specific genic regions in PIK3CA, PTEN, FBXW7, and SMAD4. Our findings indicate that these genetic alterations contribute to the development of EOCRC.

[A204] Antimicrobial and metal resistance genes in bacteria from contaminated Moroccan soils: metal-driven co-selection revealed by metagenomics

Yousra Benghait, Mohamed Blaghen
[1]Department of Biology, University Hassan II - Faculty of Science Ain Chock, Casablanca, Morocco

Heavy metal pollution imposes persistent selective pressures that drive microbial genome evolution and resistome dynamics. In chronically contaminated soils, metal resistance genes (MRGs) and antibiotic resistance genes (ARGs) become tightly coupled through co-selection and horizontal gene transfer (HGT) mediated by shared mobile genetic elements.

Using shotgun metagenomics combined with metagenomic binning, we characterized bacterial communities from long-term contaminated Moroccan soils dominated by Proteobacteria (including Pseudomonas putida, P. aeruginosa, Acinetobacter baumannii, and Enterobacter cloacae) and Firmicutes. Concentrations of Cu, Cd, Zn, Pb, and As were quantified, and resistome profiles were systematically assessed. The arsenic resistance gene arsC, primarily associated with Pseudomonas, was the most abundant MRG, while ereA, a macrolide resistance gene prevalent in Enterobacteriaceae, dominated the ARG landscape. Total MRG abundance showed a strong positive correlation with cadmium concentrations ($r = 0.86$, $p < 0.01$), indicating intense metal-driven selection.

Co-occurrence network analyses revealed signatures of HGT, with the sulfonamide resistance gene sulII (linked to Acinetobacter) co-localizing with multiple MRGs on mobile genetic elements. Similarly, the copper resistance gene copB (associated with Pseudomonas) displayed broad associations with diverse ARGs across taxonomic groups. Cadmium exhibited a high selection intensity, promoting the persistence and spread of ARGs within Proteobacteria and Firmicutes through metal-mediated co-selection.

Overall, our findings demonstrate that chronic heavy metal contamination stabilizes and amplifies antibiotic resistance in soil bacterial communities, highlighting heavy metals as major drivers of resistome evolution in polluted environments.

[A205] Reconstructing phylogenetic trees with compression: Based on nested and hierarchical repetitive substructures

Yu Liu[1], Dr. Jieyu Wang[2], Dr. Zhuoxin Zhang[3]
[1]Beijing Normal University, Zhuhai, China, [2]BGI Genomics, Shenzhen, China, [3]South China Agricultural University, Guangzhou, China

Phylogenetic inference traditionally relies on site-based evolutionary models, such as maximum likelihood, Bayesian inference, and parsimony, which primarily describe substitutions at individual nucleotide or amino-acid positions. While powerful, these frameworks often struggle to capture higher-order evolutionary events, including sequence duplication, segmental insertion–deletion, recombination, and genome-scale structural rearrangements. Such events are especially prevalent in polyploid genomes, repeat-rich regions, and collinearity-dominated genomic data.

Here we introduce a novel, alignment-free phylogenetic framework based on the Ladderpath approach from Algorithmic Information Theory. Ladderpath represents sequences through the minimal generative reuse of nested and hierarchical repetitive substructures ("ladderons"), motivated by the concept of evolution as tinkering. From the resulting hierarchical "laddergraph " representation, we define distance measures that quantify evolutionary divergence in terms of shared and unique structural reuse, without relying on explicit substitution models or sequence alignment.

We evaluate the Ladderpath approach across simulated datasets, experimentally validated phylogenies, standard molecular markers, and genomic collinearity data. The results show that Ladderpath yields phylogenetic trees broadly consistent with established methods under strong phylogenetic signal, while providing distinct advantages in duplication-dominated and repeat-rich scenarios. In particular, the method naturally accommodates unaligned sequences and syntenic blocks, and improves the interpretability of evolutionary relationships by explicitly encoding hierarchical structural information.

Our study demonstrates that compression-based, structure-aware representations offer a complementary perspective to conventional phylogenetic models, extending phylogenetic analysis toward complex and non-tree-like evolutionary processes.

[A206] Experimental validation of phenotypic diversity associated with Jomon ancestry using blood cells and iPSCs

Yuka Nakamura[1], Yusuke Watanabe[1], Hideyuki Tanabe[2], Koichi Higashi[3], Ken Kurokawa[3], Atsushi Toyoda[3], Tomoaki Kato[4], Masayoshi Tsukahara[4], Kae Koganebuchi[1], Masanori Imamura[5], Takafumi Ishida[1], Jun Ohashi[1], Hiroki Oota[1]
[1]The University of Tokyo, Bunkyo-ku, Japan, [2]The Graduate University for Advanced Studies, Miura-gun, Japan, [3]National Institute of Genetics, Mishima-shi, Japan, [4]CiRA Foundation, Osaka-shi, Japan, [5]Kanazawa University, Kanazawa-shi, Japan

Phenotypic diversity within a species arises from genetic polymorphisms. In humans, such diversity is reflected not only in disease risk variants but also in inter-individual differences in physiological, metabolic, and immune traits among healthy individuals. Recent advances in population genomics have elucidated the associations between phenotypic variation and allele frequencies. However, most of these findings are derived from in silico analyses and remain largely unvalidated experimentally. To overcome these limitations, we focused on the admixture history of the Japanese population to establish an experimental framework for validating ancestry-derived phenotypic diversity at the cellular level.

Modern Japanese are admixed descendants of the Jomon people, who already inhabited the Japanese archipelago as hunter-gatherers, and continental migrants who subsequently introduced farming. As a result, individuals vary in the proportion of Jomon-derived ancestry, which accounts for approximately 10–20% of the modern Japanese genome. We aim to investigate how variation in Jomon ancestry contributes to phenotypic diversity in present-day Japanese populations.

Using genome-linked biospecimens from the Tohoku Medical Megabank Organization, we identified individuals with extremely high or low proportions of Jomon-derived alleles. We obtained blood test data (20 individuals per group) and peripheral blood cells (5 individuals per group), from which induced pluripotent stem cells (iPSCs) were established. Comparative analyses of blood test results revealed significantly higher serum uric acid levels in the low-Jomon group compared to the high-Jomon group. In addition, transcriptome analyses indicated downregulation of immune response-related pathways in the low-Jomon group. This framework demonstrates the functional impact of Jomon ancestry.

[A207] Genomic basis of extremely long crowing in chickens

Yuki Matsuda[1], Masaoki Tsuduki[2], Tsuyoshi Shimmura[1,3]
[1]United Graduate School of Agricultural Science, Tokyo University of Agriculture and Technology, Fuchu, Tokyo, Japan, [2]Graduate School of Integrated Sciences for Life, Hiroshima University, Higashi-Hiroshima, Hiroshima, Japan, [3]Research Area of Informatics for Human-Animal Interaction, Advanced Research Center for One Welfare, Tokyo University of Agriculture and Technology, Fuchu, Tokyo, Japan

Vocalization is an essential behavior for animal communication. Most animal species produce innate vocalizations in which the patterns are genetically programmed. The species-specific pattern of vocalization is a crucial component of animal diversity. However, the genetic basis underlying variation in vocalization patterns remains largely unknown. The remarkable diversity of chicken crowing that emerged during domestication provides an excellent model for studying the molecular mechanism of innate vocalization. In this study, we focused on Japanese extremely long-crowing chicken breeds to elucidate the genomic architecture underlying the intraspecific variation of chicken crowing. We used three Japanese long-crowing chicken breeds, whose crowing exceeds ten seconds, together with seven closely related Japanese native breeds. We conducted whole-genome sequencing of 104 individuals to examine population structure and to detect genomic signatures of selection and introgression. Our population structure analyses revealed that the long-crowing chickens have independent origins, while genetic introgression occurred between these breeds. Subsequently, we screened genomic signals of selection shared among three long-crowing breeds by investigating genomic differentiation and reduced nucleotide diversity. Notably, genomic signals of selection at several genes associated with neural functions, including known vocalization-related genes, were shared among long-crowing chickens. These results suggest that mutations associated with the crowing length were shared by introgression, playing a central role in shaping the diversity of chicken crowing patterns. Our findings provide insight into how neurogenomic variation contributes to the diversification of innate vocalization.

[A208] The Most Lethal Marine Toxin: Unraveling Echotoxin's Ecological role via Transcriptomics

Yukyung Kim[1], Damin Lee[1], Jinho Park[1]
[1]Jeonbuk National University, Jeonju-si, South Korea

Marine organisms across diverse phyla harbor chemically diverse toxins through multiple pathways for predation and defense. Among them, Monoplex parthenopeus (Mollusca: Gastropoda) possesses echotoxin, an exceptionally potent hemolytic toxin reported to exhibit the highest lethality among known marine toxins. Despite its extreme toxicity, both the ecological function and evolutionary origin of echotoxin remain poorly understood. Specifically, it remains unclear whether this toxin serves as an adaptive defense mechanism to compensate for the species' large body and slow mobility, or if it fulfills alternative ecological roles.

Addressing these questions requires integrative multi-omics evidence. In this study, we present the transcriptome as a first approach comparing expression levels of toxin-associated tissue (salivary gland) and non-toxin tissues of M. parthenopeus. Our study identifies unreported echotoxin peptides, and proposes a comprehensive scenario for the functional role of echotoxin in this species.

[A209] Benchmarking D-statistics with forward population genetic simulations

Yulin Sun[1]
[1]The University Of Queensland, Brisbane, Australia

Gene flow—the exchange of genetic material between populations or closely related species—is a widespread evolutionary force critical to speciation and adaptation. Accurately detecting and quantifying gene flow from genomic data remains challenging because its signatures can be confounded by incomplete lineage sorting (ILS) and natural selection. D-statistics, originally developed to detect introgression between modern humans and Neanderthals, have become one of the most widely used methods for identifying gene flow. Despite its popularity, its performance across diverse evolutionary scenarios remains poorly characterised. We conducted a systematic assessment of D-statistics using the individual-based forward genetic simulator SLiM. Our simulation design encompasses 163 distinct evolutionary scenarios and 40 negative controls organised into five question blocks testing the effects of divergence time, gene flow characteristics, genetic drift, sampling strategy, natural selection, and gene flow duration. Our results reveal that D-statistics show poor performance across most demographic conditions, with mean sensitivity ranging from 11% to 28% across question blocks. While detection power improves with high gene flow and high recombination rates (reaching 83% in the optimal scenario), performance is severely compromised at low gene flow levels and under short divergence times where ILS is prevalent. Notably, D-statistics maintain high specificity (>95%) across most conditions, confirming their reliability when signals are detected. These findings highlight critical limitations of D-statistics and provide practical guidance for its application in empirical studies of adaptation and speciation.

Keywords: gene flow, introgression, forward simulation

[A210] A Comparative Analysis of Computel and Telomerecat for Estimating Telomere Length in Macaca fascicularis using NGS Data

YunJung Lee[1,2], SeHee Choe[1], HyeonMu Cho[1], HyeRi Park[1,2], MinGyeong Ko[1,2], SangJe Park[1], YoungHyun Kim[1], Jaewon Huh[1,2]
[1]KRIBB, Cheongju , South Korea, [2]UST, Cheongju , South Korea

Telomeres, consisting of TTAGGG repeat sequences at chromosomal termini, are essential for maintaining genomic stability and preventing degradation. Due to the "end replication problem" during lagging strand synthesis, telomeres progressively shorten with each cell division, making telomere length (TL) a recognized hallmark of biological aging. While TL was traditionally measured through experimental assays, advancements in whole-genome next-generation sequencing (NGS) and bioinformatics have enabled TL estimation via various computational tools. In this study, we utilized Macaca fascicularis, an evolutionary proxy for human aging, to evaluate and compare the performance of two prominent computational tools: Computel and Telomerecat. Using paired-end sequencing data collected from 16 individuals over a six-year follow-up period, we processed the data according to each tool's requirements—utilizing FASTQ files for the R-based Computel and BAM files for the Python-based Telomerecat. Our analysis revealed that TL estimates from Telomerecat exhibited a characteristic decreasing trend with age. Conversely, Computel showed an unexpected upward trend. Although neither tool reached statistical significance in its correlation with age—likely due to the limited cohort size—these findings highlight the importance of tool selection in NGS-based telomere analysis. This comparative study provides preliminary insights into the consistency of computational TL estimation in non-human primate models.

[A211] Comparing signals of positive selection inferred by dN/dS models and mutation–selection models across mammalian genomes

Yuqing Peng[1]
[1]Queen Mary University of London, , United Kingdom
Identifying positive selection in protein-coding genes is a core aim of evolutionary genomics. Codon-based dN/dS models implemented in CODEML are widely applied to detect positive selection, but these models can conflate effects of mutation bias, genetic code structure, and amino-acid preferences. Mutation–selection models, offer an alternative framework by explicitly modelling site-specific amino-acid fitness while accounting for underlying mutation processes. A direct comparison between these two approaches is therefore necessary to clarify how model assumptions shape inference of adaptive evolution. Candidate positively selected genes and sites were first identified using CODEML based on likelihood ratio tests and site-specific dN/dS estimates. The same genes were then analysed under MutSel to estimate amino-acid fitness landscapes and to characterise selection patterns at individual sites. Concordance and discrepancies between the two methods were evaluated at both gene and site levels.

[A212] Fruit evolution is key to ecological niches and distribution range divergence in tribe Ligustrinae (Oleaceae)

Yushuang Wang[1,2], Mr Enze Li[1], Mr Xingyong Cui[1], Ms Yuxin Ren[1], Mr Gangmin Zhang[1], Prof. Loren Rieseberg[2], Mr Wenpan Dong[1]

[1]Beijing Forestry University, School of Ecology and Nature Conservation, Beijing, China, [2]University of British Columbia, Department of botany, Vancouver, Canada

Fruits are a key innovation of plants; they are crucial for adaptation to environmental heterogeneity and have led to species diversification and variation in species richness within clades. To evaluate the influence of fruit evolution on species diversification, we assessed the relationship between fruit evolution and climate from phylogenetic and ecological perspectives within the Ligustrinae. This group includes two genera, Syringa and Ligustrum, which have different fruit types (capsules and drupes, respectively). We reconstructed highly supported phylogenetic relationships and divergence times of Ligustrinae species. Distribution patterns showed that Ligustrum and Syringa had significant ecological niches and distribution ranges divergence. Biogeographic results indicated that Ligustrinae originated from the northern temperate zone and gradually spread to the subtropical and tropical zones along with the fruit evolution from capsules to drupes. Ancestral ecological niches reconstruction and current ecological niches analysis indicated that Syringa species are suited to humid and cold areas, whereas Ligustrum species are suited to warm and humid habitats, with Ligustrum having a more extensive ecological niche than Syringa. To further validate at the molecular level how fruit evolution promotes geographic adaptation in Ligustrinae, we integrated SNP-based genomic and transcriptomic analyses to identify molecular signatures underlying fruit type–associated ecological differentiation.

[A213] Uncovering the 3FTx Repertoire in Elapidae Through Reanalysis of Public Transcriptome Data

Yu-Wen Huang[1], Dr. Yeong-Shin Lin[1], Dr. Cheng- Tsung Pan[2]
[1]Institute of Bioinformatics and Systems Biology, National Yang Ming Chiao Tung University, Hsinchu City, Taiwan, [2]Graduate School of Integrated Sciences for Life, Hiroshima University, Higashi-Hiroshima, Japan
Uncover 3FTx repertoire in Elapidae by reanalyzing publicly available transcriptome data

Snakebite envenoming is a neglected tropical disease that causes over 100,000 deaths annually. Characterizing toxin repertoires and their diversity can accelerate the development of universal antivenoms. Three-finger toxins (3FTxs), a superfamily of non-enzymatic proteins with diverse toxicities, are dominant components of Elapidae venoms. According to the first three amino acids of signal peptide, 3FTx can be classified into MKT-3FTx and MKA-3FTx. Although the MKT-3FTx subfamily accounts for most protein variants, the MKA-3FTx subfamily remains largely neglected. A previous study reported four MKA-3FTx copies in the Bungarus multicinctus genome, showing structural divergence and distinct expression in gland and kidney. Here, we designed a bioinformatics pipeline to comprehensively reconstruct the 3FTx catalog from publicly available transcriptome data in Elapidae, comprising 162 samples across 43 species. In addition to the previously reported MKA-3FTx, we identified a novel MKA-3FTx that is ubiquitously expressed across species and tissues, suggesting an unrecognized physiological role in non-venom tissues. We also identified tissue-specific MKA-3FTx expressed in the testis and pancreas of Naja naja, as well as distinct gland-specific MKA-3FTx variants expressed across multiple species, indicating both repertoire expansion and diversification within MKA-3FTx subfamily. Future work will build on these findings by revealing the within-species tissue-specific regulation and reconstructing the evolutionary history of MKA-3FTx subfamily.

[A214] Detecting Positive Selection Using Spatial Autocorrelation and Haplotype Structure in Populations with Limited Dispersal

Amatur Rahman[1], Zachary Szpiech[1]
[1]Penn State University, University Park, United States

Most population genetic analyses assume discrete, well-mixed populations, yet natural populations are often spatially structured, with limited dispersal and geographic constraints on mating. These deviations from panmixia can bias demographic inference, generate spurious associations with environmental variables, and complicate the detection of selective sweeps. A selective sweep occurs when a beneficial mutation rapidly increases in frequency, carrying along linked neutral variants. Genomic regions under positive selection are therefore expected to show characteristic signatures such as long haplotypes at elevated frequency. Haplotype-based approaches generally improve power over allele-frequency-only methods by leveraging this expected pattern, but limited dispersal dynamics modify these signatures. Under limited dispersal, the slow spatial spread of adaptive alleles weakens extended haplotype homozygosity, increases intermediate-frequency variants, and promotes sweeps on multiple haplotype backgrounds, thus reducing the power of standard statistics. Accounting for spatial structure is therefore crucial for detection of selective sweeps.

Recent advances have demonstrated that incorporating geographic information can distinguish positively selected variants from neutrally evolving ones. Here, we extend this insight to haplotype-based approaches by integrating haplotype structure with spatial autocorrelation to construct a joint statistic for detecting positive selection in spatially structured populations with limited dispersal. Using individual-based simulations under a limited-dispersal model, we show that naive application of classical haplotype-based methods misses signals that are recovered by our statistic, while controlling the false positive rate at 1%. Our method also identifies geographic regions where the adaptive allele is likely segregating. We incorporate this new method into the software selscan for easy application.

[A215] Decoding the interactome of biomolecular condensates

Alessandro Bevilacqua[1,2], Swantje Lenz[1,2], Jonas Poehls[1,2], Doris Richter[1,2], Barbara Szewczyk[1,2], Anna Shevchenko[1], Andrej Shevchenko[1], Agnes Toth-Petroczy[1,2,3]
[1]Max Planck Institute of Molecular Cell Biology and Genetics, Dresden, Germany, [2]Center for Systems Biology Dresden, , Germany, [3]Cluster of Excellence Physics of Life, TU Dresden, Dresden, Germany
Biomolecular condensates are dynamic, membraneless organelles organize cellular components by selectively concentrating proteins and nucleic acids in response to internal and external cues. Despite their central role in cell biology, two key aspects remain largely unknown: their comprehensive molecular composition and the network of interactions ("interactome") that governs their assembly and function. Existing proteomic approaches are low throughput and fail to capture the transient, weak, and condition-dependent interactions that define condensates, leaving gaps in our understanding of the function of these dynamic organelles.
This project establishes an in vivo cross-linking mass spectrometry (CL-MS) pipeline to map the interactome of biomolecular condensates with high sensitivity and structural resolution. In order to validate the approach, we investigate stress granules in HEK293 cells, using the well-characterized protein G3BP1 tagged with ALFAtag. After live-cell cross-linking G3BP1-associated proteins are identified by the cross-linked peptides through mass spectrometry. Structural modelling by AlphaLink, reveals the interaction sites, guiding models of the weak, multivalent interactions that drive condensate formation and interactome network.
By delivering a high-throughput, in vivo CL-MS method tailored to study biomolecular condensates, this work elucidates the molecular, structural and network organization of these assemblies. This tool can be applied to map the interactome of uncharacterized condensate-forming proteins, to advance our understanding of how condensates contribute to cellular physiology and stress adaptation, and how their dysregulation is linked to neurodegeneration, cancer, and other pathologies involving aberrant phase separation.

[A216] Evolutionary loss of umami taste receptor gene TAS1R1 in tamarins among TAS1R gene family of ecologically diverse platyrrhine primates

Zhixin Wu[1], Min Hou[1], Dr. Muhammad Shoaib Akhtar[1], Masahiro Hayashi[1], Takashi Hayakawa[2], Amanda D. Melin[3], Shoji Kawamura[1]
[1]Department of Integrated Biosciences, Graduate School of Frontier Sciences, The University of Tokyo, , Japan, [2]Faculty of Environmental Earth Science, Hokkaido University, , Japan, [3]Department of Anthropology and Archaeology, University of Calgary, , Canada
Sensing taste helps animals make decisions about ingesting beneficial foods. The umami and sweet tastes are sensed by TAS1R1-TAS1R3 and TAS1R2-TAS1R3 heterodimers, respectively, in mammals. Studies have revealed evolutionary diversity of genes encoding these receptors, TAS1R1, TAS1R2 and TAS1R3. However, much information has largely relied on whole-genome-assembly data which are often incomplete. Platyrrhine primates (previously called as “New World monkeys”) are suitable for understanding the evolutionary diversity of umami-sweet taste receptor gene family because of their remarkable diversity in diets as well as in color vision which has often been discussed in relation to ecological diversification. In this study, we applied targeted capture followed by short-read high-depth massive parallel sequencing for the three genes from 18 species of platyrrhines from all three platyrrhine Families. While the three genes were overall conservative, we noted that TAS1R1 was disrupted in three genera of Subfamily Callitrichinae, tamarins (Saguinus), Saddle-back tamarins (Leontocebus) and lion tamarins (Leontopithecus), implying three independent disruption events in the three genera or at least two independent disruptions in Leontopithecus and the common ancestor of Saguinus and Leontocebus. This implies less importance of sensing umami in these genera. Their variation pattern of color vision and dietary dependence on tree-sap are also observed in other callitrichine species of which TAS1R1 was intact. Thus, it is currently not certain why their TAS1R1s were disrupted. Further studies on other taste and chemical sensor genes should complement this study and facilitate our understanding on the sensory diversity of platyrrhines and primates in general.

[A217] Does the rediploidization process impact observed duplicate outcomes following the Saccharomyces whole genome duplication?

Zoe Vance[1], Tom Williams[1], James Clark[1]
[1]University Of Bath, , United Kingdom

Whole genome duplication (WGD) is implicated in the emergence of key innovations in multiple lineages, likely playing an important role in evolution. Despite much work investigating WGDs, it was not appreciated until recently that rediploidization, the process by which organisms regain a diploid genome structure following WGD, can play a crucial role in shaping observed outcomes. This process was assumed to be near-instantaneous, however recent developments in fish demonstrate the possibility of a prolonged state of tetraploidy. In these cases, continued meiotic pairing across the multiple genome copies present is hypothesised to delay divergence of duplicate copies, impacting the phylogenetic signal of the duplication so that it appears delayed, or even as multiple signals. Determining whether this finding is generalisable requires evidence from diverse lineages, particularly including cases of hybridisation, where rapid rediploidization is more likely than in simple genome doubling. The yeast WGD, a hybridisation giving rise to the lineage containing brewer's yeast (Saccharomyces cerevisiae), provides an excellent starting point with evidence for widespread duplicate homogenisation and well-defined datasets. Here, we present work aiming to characterise the apparent patterns of duplication following the Saccharomyces WGD and establish whether delayed rediploidization is a plausible model for this event. We confirm extensive disagreement in duplication timing for WGD duplicates, potentially reflecting similar processes to those seen in fish, or more recent ongoing gene conversion unrelated to prolonged tetraploidy. Assessing the relevance of rediploidization across the tree of life will assist in forming more accurate assumptions surrounding the outcomes of WGD events.

[A218] Population genomics of a recently expanded annual weed across its geographic range

Aaditya Narasimhan[1], Marjolaine Rousselle[1], Shengman Lyu[1], John Pannell[1]
[1]University Of Lausanne, , Switzerland

Populations vary genetically across their geographic ranges, which can depend on their expansion history. The sources that lead to the geographic structure of genetic variation can determine the capacity of populations to adapt beyond their ancestral ranges. Here, we studied populations of Mercurialis annua, a dioecious annual weed with a broad distribution in Europe and the Mediterranean resulting from a range expansion from the eastern Mediterranean across Europe. We compiled a genomic dataset of 360 individuals sampled from 26 locations across the range of M. annua to understand its expansion history and the selective consequences associated with expansion, especially for populations established at its geographic range edge. We first affirm the hypothesis that M. annua expanded from a refugium in the eastern Mediterranean, based on patterns of genetic diversity and population structure. We back up this hypothesis by performing demographic simulations to date the time of expansion, along with testing the hypothesis of a second possible source of expansion. We show that M. annua forms two distinct geographic clusters characterised by high migration in northwestern Europe and low migration among populations in the rest of its range. We found that population sizes expanded sequentially from the refugium to northern Europe, with little long-range admixture, consistent with a stepping-stone expansion out of the Levant. Strong declines of genetic diversity towards the Northern edge combined with an accumulation of deleterious load at the northern range edges likely maintain the current range edges of M. annua.

[A219] Bacterial mutators reveal a deterministic role of standing variation in streamlining evolutionary outcomes

Aakanksha Madhwal[1], Dr Deepa Agashe[1]
[1]National Centre For Biological Sciences, Bengaluru, India

Most natural populations harbour substantial genetic diversity, which facilitates adaptation to environmental change by allowing selection to sample from pre-adapted beneficial variants. It also promotes exploration of fitness landscapes by increasing the likelihood of generating new beneficial variants via recombination or epistasis with new mutations. However, microbial evolution experiments typically ignore standing variation, limiting the understanding of the relative roles of standing versus de novo variation, their consequences for evolutionary outcomes, and underlying mechanisms. We measured the effects of standing variation using bacterial hypermutators with increased mutation supply. Populations of two Escherichia coli mutator strains were founded with a gradient of standing variation (1, 4, or 12 genotypes) and evolved for 16 days. Final genotype frequencies were determined using whole-genome sequencing, and adaptation was quantified as changes in population growth rate across genotype mixes and biological replicates of the same mix. Populations with higher standing variation adapted faster, but only in one strain. Intermediate- and high-diversity populations were typically dominated by a single ancestral genotype, although its identity varied across genotype mixes and was not predicted by genotype performance in monoculture. Final population growth rate was strongly positively correlated with the dominant genotype frequency, indicating that winning genotypes drove adaptation. Finally, variation in final growth rates was greater across genotype mixes than across replicates of the same mix. Together, these results show that despite the diversifying potential of high mutation supply and standing genetic variation, evolutionary outcomes are strongly streamlined, with important roles for both standing and de novo variation.

[A220] Ancient Mitogenomes from Belize Provide Insights into the Mobility and Migration of Ancient Communities in Mesoamerica and the Caribbean.

Celia Cleary[1], Angelina Locker[2], Lauren Springs[3,4], Diane Chase[5], Arlen Chase[6], Adrian Chase[7], Melissa Badillo[8], Adella Vallejos[9], Genara Cano[9], Jada Benn Torres[2], Rick Smith[3,10], Dr. Austin Reynolds[11,1]
[1]Department of Anthropology, Baylor University, Waco, United States, [2]Department of Anthropology, Vanderbilt University, Nashville, United States, [3]Department of Sociology and Anthropology, George Mason University, Fairfax, United States, [4]Department of Integrative Biology, University of Texas at Austin, Austin, United States, [5]Office of the Provost, University of Houston, Houston, United States, [6]Department of Comparative Cultural Studies, University of Houston, Houston, United States, [7]Middle American Research Institute and Stone Center for Latin American Studies, Tulane University, New Orleans, United States, [8]Institute of Archaeology, NICH, Benque Viejo del Carmen, Belize, [9]To'one Masehualoon NGO, Corozal, Belize, [10]Department of Women and Gender Studies, George Mason University, Fairfax, United States, [11]Department of Microbiology, Immunology, and Genetics, University of North Texas Health Science Center at Fort Worth, Fort Worth, United States

Archaeological and stable isotope evidence from across the Maya lowlands paints a picture of a demographically mobile population with complex and dynamic relationships between some of the largest urban polities in antiquity. Maya populations have undergone several largescale demographic transformations including the formation and collapse of states, climatic events, and multiple colonial regimes. The nature of the genomic relationship between ancient Maya urban centers, as well as their relationship with present-day Maya communities, remains under-characterized. We present mitogenomic data of sixteen ancient Maya Ancestors recovered from interments in the ancient Maya cities of Caracol and Chactemal (Santa Rita Corozal) located in Western and Northern Belize. These ancient data were combined with 107 newly collected contemporary individuals from Northern Belize, along with over 600 previously published ancient and contemporary comparative mitogenomes recovered from sites spanning Mesoamerica and the Caribbean to reconstruct paleodemographic and mobility patterns of maternal lineages through time. This work was conducted with authorization from the Belize Institute of Archaeology and in collaboration with Maya partners in Corozal. Haplogroup frequencies and median joining haplogroup networks were generated to understand the sharing of haplotypes across time and space in this region. Bayesian fixed molecular clock models were used to estimate matrilineal phylogenetic relationships between community members included in this study. Bayesian Skyline Plots were generated to infer dynamics in matrilineal effective population size. Resulting models were used to reconstruct matrilineal paleodemographic events temporally and geospatially, revealing varied matrilineal links between Maya communities from antiquity to today.

[A221] Revisiting the Classification of Macro- and Microchromosomes in Vertebrates: Insights into Their Origins

Yuichiro Hara[1]
[1]Kitasato University, , Japan

Vertebrate genomes often exhibit a bimodal structure comprising large macrochromosomes and small microchromosomes, a feature increasingly recognized as ancestral. These chromosome types differ not only in size but also in genomic features, such as GC content and gene density, and epigenomic characteristics, including transcriptional regulation activity and DNA replication timing. However, conventional classification of these chromosome types remains tethered to arbitrary size definitions (e.g., nucleotide lengths in chromosomes). Such metrics may fail to account for the vast diversity of genome sizes across taxa, potentially obscuring the functional identity of homologous chromosomes. To address this, we developed a size-independent classification of macro- and microchromosomes based on oligonucleotide composition. We applied this approach to a variety of vertebrate genomes, which strongly suggests that macro- and microchromosomes originated from distinct subgenomes resulting from the second-round whole genome duplication, also known as allopolyploidisation, in early gnathostomes. Furthermore, comparative analysis reveals that while lineages such as elasmobranchs and some birds retain this bimodal architecture, their specific chromosomal compositions have undergone significant reorganization. By redefining the "basic design" of the vertebrate karyotype through intrinsic signatures rather than size, we elucidate the relationship between gene function and chromosomal location—an analysis often masked in mammalian genomes with diverged chromosome structures. This study provides a novel framework for understanding the structural evolution of vertebrate genomes as well as their gene repertoires.

[A222] The evolutionary origin of the Columbian mammoths from the Basin of Mexico and its implications for the Mammuthus genus

Rigoberto Padilla-Bustos[1], Lic. Ángeles Tavares-Guzmán[1], MSc. Alejandra Castillo-Carbajal[1], Eduardo Alejandro Arrieta-Donato[1], Dra. Miriam Bravo-López[1,4], Graduate Ernesto Garfias Morales[1], Viridiana Villa Islas[1,5], Dra. Marcela Sandoval-Velasco[2], Dr. Alejandro López-Jímenez[3], Dr. Joaquín Arroyo-Cabrales[3], Dr Maria C. Ávila-Arcos[1], Dr. Federico Sánchez-Quinto[1]

[1]International Laboratory for Human Genome Research, Universidad Nacional Autónoma de México (UNAM), Queretaro, Mexico, [2]Centro de Ciencias Genómicas, Universidad Nacional Autónoma de Méxicoentro de Ciencias Genómicas, Universidad Nacional Autónoma de México, Cuernavaca, Mexico, [3]Laboratorio de Arqueozoología, Subdirección de Laboratorios y Apoyo Académico, Instituto Nacional de Antropología e Historia, Mexico City, Mexico, [4] Center for Evolution and Medicine, Arizona State University, , USA, [5]Globe Institute, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

Paleogenomic studies suggest that Columbian mammoth (Mammuthus columbi) originated from an ancient hybridization between woolly mammoths (Mammuthus primigenius) and steppe mammoths (Mammuthus trogontherii). While M. columbi's habitat ranged from North to Central America, available genetic data are limited to boreal and temperate regions of North America. In fact, the only M. columbi nuclear genome available to date derives from an individual from Wyoming (USA). The discovery of Columbian mammoth remains in the Basin of Mexico provides a unique opportunity to further understand its evolutionary trajectory by adding information from tropical latitudes. We generated three low-coverage nuclear genomes, one of them >1x, and jointly analyzed these data with other Columbian, woolly and steppe mammoth genomes from America and Eurasia. We performed phylogenetic and demographic analyses, as well as contrast genomic regions of putative M. trogontherii-like origin in Columbian mammoths. We observed the USA M. columbi sample harbors a higher genetic affinity to M. primigenius, while the Mexican Columbian mammoth genome displays a higher proportion of trogontherii-like alleles. We propose an evolutionary model that reconciles these observations. Our study revealed a complex evolutionary origin of the Columbian mammoths, and underscores the importance of wider spatial sampling to better comprehend its evolutionary history.

[A223] Probe efficacy and an initial phylogenomic reconstruction of freshwater crayfish using anchored hybrid enrichment

Meng-Chen Yu[1], Brooke Grubb[1], Katelyn Power[1], Aintzane Santaquiteria[2], Marcos Pérez-Losada[1], Carlos Pedraza[3], Keith Crandall[1,2]

[1]Computational Biology Institute, Department of Biostatistics and Bioinformatics, George Washington University, Washington, United States, [2]Department of Biological Sciences, George Washington University, Washington, United States, [3]Instituto de Ciencias del Mar y Limnología, Universidad Nacional Autónoma de México, Mexico City, Mexico

Freshwater crayfishes represent a diverse lineage of ~750 species and play key ecological roles as ecosystems engineers in freshwater systems. Despite their importance, crayfish systematics remains challenging due to limited diagnostic morphology, high intraspecific plasticity, and poor phylogenetic resolution from legacy genes, constraining species boundaries and conservation action. Improving phylogenetic resolution is therefore essential for understanding crayfish evolutionary history and ecological diversity. We evaluated the efficacy of anchored hybrid enrichment (AHE) probes designed for broad decapod phylogenetics in resolving crayfish relationships across multiple evolutionary depths. We targeted 114 nuclear loci across 300 individuals representing 51 species spanning the crayfish tree of life. Sequence capture and assembly were conducted using HybPiper, and phylogenetic inference was performed using maximum-likelihood analyses in IQ-TREE. To assess the impact of locus inclusion, we compared phylogenies generated using strict (100%) and relaxed (75%) locus recovery thresholds and contrasted these with a phylogeny inferred from legacy genes. Both AHE thresholds produced highly resolved and largely congruent phylogenies with strong support across multiple taxonomic levels. The 75% threshold increased locus representation while maintaining overall topological stability, whereas the 100% threshold yielded a more conservative dataset with reduced taxon coverage. Minor topological differences between thresholds were largely confined to shallow nodes. Comparisons with legacy genes phylogenies revealed several conflicting relationships, indicating widespread mito-nuclear discordance across crayfishes. Overall, our results demonstrate that AHE probes perform effectively across crayfishes, providing robust phylogenetic signal and a framework for future studies of ecological strategies, functional traits, and evolutionary history.

[A224] The Population History of Hybridisation and Local Adaptation in Previously Unknown Admixed Rice Population

Meng-ting Hsieh[1,2], Dr Rafal Gutaker[1], Jose De Vega[2], Rachel Rusholme-Pilcher[2], Anthony Hall[2]
[1]Royal Botanic Garden, Kew, London, United Kingdom, [2]Earlham Institute, Norwich, United Kingdom

Our food security is threatened by global challenges, such as accelerated climate change. Assessing crop genetic diversity is crucial for preserving global genetic diversity as a resource for crop improvement. Here, we characterise a unique indica subpopulation, I5, with an admix-like population ancestry. I5 is a population of traditional low-yielding lowland landraces preserved in the low-altitude area around the Red River Delta with assumed adaptive or culinary value, and thus has been selected and preserved in the area. This project aims to understand how local selection on beneficial traits shapes the crop population structure, its effects on global crop genetic diversity over time, and identify unique genetic resources in I5 that could be used in sustainable crop breeding programs. We constructed and annotated a reference genome for I5. While an annotation-based pan-genome didn't show any megabase-scale structural variation, a SNP-based mapping method revealed multiple short non-indica regions. These regions are suggested to result from one or more hybridisation events between different gene pools, leading to I5 divergence from other indica subpopulations, possibly under selection. To navigate the further details of I5, we reconstructed the relationship between I5 and other rice subpopulations using population-genetics-based tools.

[A225] Inferring shared sources of admixture among populations worldwide using a new technique

Mengzi Chen[1], Professor Garrett Hellenthal[1]
[1]University College London, London, United Kingdom
Several techniques exist to infer whether populations descend from admixture events when two more ancestral sources intermixed, as well as infer admixture proportions and sources. However, many available approaches are limited by only being able to analyse one admixed population at a time. This implicitly assumes that all individuals within the population are genetically homogeneous, with entirely overlapping ancestral sources and the same or similar admixture fractions, and possibly the same admixture dates.

Here we introduce an unpublished technique that jointly analyses multiple individuals/populations and infers the number of unique, genetically-homogeneous admixing sources required to describe their genetic variation patterns sufficiently. In particular it simultaneously estimates (i) the number of ancestral sources, (ii) which individuals (or populations) share one or more ancestral sources, (iii) each individual's admixture proportions, and (iv) the genetic make-up of each admixing ancestral source. In doing so, it can infer whether a particular source of admixture is shared across multiple populations, which is often of archaeological or historical interest, and borrow strength across all such populations to better infer that source's genetic make-up. Our method, which uses input from the haplotype-based programs CHROMOPAINTER and GLOBETROTTER, can implement prior knowledge regarding which sampled surrogate populations best reflect each (unknown) admixing source. Alternatively, it can automatically assign surrogates to source groups using admixture linkage disequilibrium patterns.

We assess power and accuracy via simulations. Analysing real data, we highlight likely shared ancestral sources among populations spanning the former Mongol Empire, and likely shared Asian-related sources among northern European groups.

[A226] Inferring South American camelids history through the lens of whole-genome data in context of their domestication

Michael Orellana[1], William Taylor, Fernando Villanea
[1]University Of Colorado Boulder, Boulder, United States

Domestication is a coevolutionary process in which one species manages the survival and reproduction of another species to obtain services and/or resources. In the Americas, South American Camelids (SACs) were the only large ungulates domesticated during pre-Hispanic times. Given their economic and biological importance, genetic studies have focused on elucidating the biological relationships among SAC species, primarily through analyses of specific genomic regions and uniparental markers. However, the limited genomic data has restricted our ability to accurately infer their demographic history and the genetic impacts of domestication on domesticated species. Here, we analyzed whole-genome data from published studies of the wild species, Lama guanicoe and Vicugna vicugna, and the domesticated species Lama glama and Vicugna pacos. We estimated changes in effective population size in each species based on mitogenome data and whole-genome data. Our results revealed a decline in populations of Vicugna vicugna starting approximately 500 years ago, consistent with the timing of European colonization of the Americas. We performed population structure analyses based on autosomal markers and identified four main ancestry components traced to the wild species. The domesticated Lama glama derives its ancestry from a subset of the ancestries in the wild Lama guanicoe, which is consistent previous domestication literature. Conversely, we find that the domesticated Vicugna pacos draws its ancestry from two lineages of the wild Vicugna vicugna and presents evidence of admixture from the Lama lineage. These results illustrate a more complex evolutionary and domestication history of South American camelids than previously observed through genetic data.

[A227] Comparative Genomic Analysis of Tandem Repeat Variations and Transcriptome Diversity across primate evolution

Min-gyeong Ko[1], Se-Hee Choe[1], Hyeon-Mu Cho[1], Hye-Ri Park[1,2], Yun-Jung Lee[1,2], Sang-Je Park[1], Young-Hyun Kim[1], Jaewon Huh[1,2]
[1]Kribb, Cheongju-si, South Korea, [2]Korea University of Science & Technology (UST), Cheongju-si, South Korea

Comparative genomics reveals that the 1% genetic divergence between humans and chimpanzees significantly contributes to lineage-specific biological traits. This study extends beyond the human-chimpanzee dyad to incorporate rhesus and cynomolgus macaque genomes, aiming to elucidate the broader landscape of primate genomic diversity. We specifically focus on tandem repeat (TR) variations, which exhibit substantial interspecies polymorphism. Having confirmed significant sequence variations among species, our research now aims to characterize their differential expression profiles specifically within reproductive organs and the brain.
Based on high-throughput sequencing data, we identified four candidate genes—THEM5, EIF5B, C14orf159, and DUSP4—containing prominent TR sequences. To validate these findings, we are analyzing tissue-specific expression patterns across a diverse primate panel, including chimpanzees, gorillas, orangutans, and Old World monkeys (OWMs), using cDNA-based assays. Furthermore, we utilize Next-Generation Sequencing (NGS) to systematically analyze transcriptome diversity and understand the evolutionary mechanisms driving phenotypic differences. Our research addresses this by performing multi-species comparative analyses to capture a broad-spectrum view of evolutionary transcriptome shifts. This integrative approach will provide deeper insights into the evolutionary trajectories of human and non-human primate genomes, facilitating a more robust understanding of primate diversity within a condensed timeframe.

[A228] Analysis of a Possible Relaxation of Natural Selection in Human Populations

Miriam Alejandra Jimenez Marmolejo[1], Diego Ortega-Del Vecchyo[1]
[1]International Laboratory for Human Genome Research, National Autonomous University of Mexico, Querétaro, Mexico

Purifying selection is an evolutionary force that removes deleterious genetic variants from populations. The efficacy of purifying selection in humans has been hypothesized to have changed recently on certain genes coinciding with environmental and cultural transitions including the development of agriculture and modern medicine. These transitions are proposed to have modified selective pressures by changing the fitness effects of deleterious variants resulting in a relaxation of purifying selection, whereby mutations that were deleterious in the past are less deleterious in the present. Here, we investigate how the relaxation of purifying selection changes genetic variation patterns. We used forward-time simulations varying the relaxation of purifying selection acting on deleterious mutations. The resulting genetic variation patterns were analyzed using the site frequency spectrum (SFS) by comparing variant counts across distinct frequency classes. The relaxation of purifying selection resulted in a consistent enrichment of rare variants within a derived allele frequency window of 0.75–1.50%, while very low (<0.5%), intermediate, and high-frequency classes remained largely unchanged. We show plans to extend this analysis by integrating genealogical information and analyzing different demographic scenarios consistent with human history with the goal of evaluating evidence for the relaxation of purifying selection in humans.

[A229] Leveraging stochastic mutational mappings to estimate time-resolved, significant changes in selection along a global SARS-CoV-2 phylogeny

Monica Arniella[1], Rasmus Nielsen[1,2]
[1]University of California Berkeley, Berkeley, United States, [2]University of Copenhagen, Copenhagen, Denmark

Viruses evolve drastically faster than their hosts due to several factors, including high mutation rates that affect their evolutionary potential. A common measure of the strength of selection is dN/dS, the ratio of nonsynonymous (dN) to synonymous (dS) changes, which helps identify adaptive substitutions that may be associated with lineage-specific characteristics like enhanced transmission or virulence that often characterize variants of concern (VoC). However, viral lineages sampled in recent timepoints are not fully purged of deleterious mutations, inflating dN closer to the tips of the phylogeny. Thus, understanding changes in selection regime requires separating dN and dS, and the timing of these mutations along the lineages in question. Simulation-based approaches for calculating dN/dS from mutational mappings are especially powerful as they retain counts and timing for substitutions, allowing for the propagation of uncertainty, analysis over time, and tracking the fate of particular alleles. Stochastic mutational mappings also leverage existing ML tree estimates without jointly re-estimating trees and parameters, making this approach well-suited for working with large, publicly accessible phylogenies. Here, we implement previously described frameworks for simulation-based mutational mapping, then apply this method to a curated, global SARS-CoV-2 phylogeny. Using these mappings, we further implement a method to calculate the expectations of dN and dS under different selection scenarios using a Wright-Fisher diffusion model and over time windows in the phylogeny. Our aim in developing and validating this method is to apply it in order to understand the selection trajectories leading to pre- and post-Omicron VoC.

[A230] A Diachronic Paleogenomic Analysis of Consanguinity from the Iron Age to the Early Middle Ages in the Roman World

Asst. Prof. N. Ezgi Altınışık[1,2]
[1]Department of Anthropology, Hacettepe University, Ankara, Türkiye, [2]Department of Health Informatics, Institute of Informatics, Hacettepe University, Ankara, Türkiye

This study investigates the impact of Roman socio-political integration on mating choices using a diachronic dataset of ancient genomes (n≈300) from Southern Europe, the Balkans, and Anatolia (1200 BCE–1000 CE). Using Runs of Homozygosity (ROH), I distinguished background relatedness from recent parental consanguinity, employing segments >20 cM as diagnostic markers. Macro-scale analysis reveals that Southern Europe was the only region exhibiting significant temporal fluctuations, characterized by a sharp "Roman Drop" in consanguinity during the Imperial peak. Conversely, Anatolia and the Balkans maintained stable mating patterns, suggesting regional resilience of marriage networks despite imperial administration. Cross-regional heterogeneity peaked during the Roman period before converging in Late Antiquity as Southern European ROH levels rebounded to match the persistent Anatolian baseline.

To refine micro-scale reconstructions and to classify the parental kinship types and degrees, I developed rohClassifier, an R package utilizing a random forest algorithm on ROH distributions, achieving 93% accuracy in mating simulations. I identified a 5th-century infant from Nicaea and a 2nd-century Eastern migrant in Viminacium as offspring of full-sibling unions (>85% probability). According to the archaeological context, while the Nicaea case suggests institutional care for a socially proscribed union, the Viminacium case reflects enclave endogamy. With additional individuals showing second- or third-degree relatedness, the results demonstrate that while the Roman core underwent a transition toward exogamy, diverse endogamous traditions persisted across the provinces, driven by varying social and cultural motivations.

[A231] Persistence and Context of Human Sedimentary DNA in Modern Burial Soils: Implications for Forensic and Evolutionary Interpretation

Nadescha Viviane Hänggi[1], Nadezda Vilgerte Jochumsen[1], Ph.D. Marie-Louise Kampmann[1], Ph.D. Elena I. Zavala[1,2]
[1]Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark, [2]Section for GeoGenetics, The Globe Institute, University of Copenhagen, Copenhagen, Denmark

Sedimentary ancient DNA (sedaDNA) has become instrumental in evolutionary genomics and ecology, enabling reconstruction of past ecosystems, tracing the presence and movement of species, and exploring population histories. However, persistence, degradation, and interpretability of sedaDNA remain poorly understood. Modern sediment DNA (sediDNA) from known depositional contexts offers the potential to fill this gap. Such insights are relevant to both evolutionary research and forensic investigations, where sed(i/a)DNA could complement or replace skeletal DNA analysis when remains are highly degraded, absent, or cannot be sampled. However, limited understanding of sediDNA taphonomy prevents the development of optimal sampling and authentication strategies and interpretation of downstream results.

Here, we evaluate laboratory and computational workflows for detecting human sediDNA in modern burial soils, using samples collected from controlled human decomposition experiments at two taphonomic research facilities. The known depositional context, donor identity, and spatial sampling design allows for the systematic assessment of how these factors and laboratory workflow translate to sediDNA recovery.

We evaluated DNA extraction, and library preparation protocols with complementary strengths, alongside sequencing approaches, determining a robust framework for the investigation of modern burial soils. We examined human DNA yield, library complexity, and cytosine deamination profiles, illuminating sediDNA taphonomy. These results indicate how reliably detected and authenticated trace human DNA recovered from sediments may support the reconstruction of burial context and prior human presence in forensic applications. By clarifying how context and sampling shape sediDNA signals, this work also informs interpretations of results from modern and ancient sedimentary DNA studies.

[A232] Impact of a jigsaw activity on undergraduate students' conceptual understanding: narrowing the gap between low and high performers

Nadia Aubin-Horth[1], Carla Bautista Rodriguez, Émilie Cousineau
[1]Université Laval, Québec, Canada

A goal of undergraduate education is to teach key theoretical concepts, as well as skills, such as critical thinking and the ability to analyse a new problem. Active learning has been proposed to have a positive impact on the understanding of disciplinary concepts and skills by using existing knowledge, collaborative learning, and teamwork. This type of approach has been shown to have a significant positive impact on achievement of underrepresented persons in science, especially when combined with an inclusive pedagogy approach. A Jigsaw activity, in which students combine their individual knowledge to create an expert opinion as a team to then teach their peers, has been applied at the undergraduate level. However, its effect on the learning of key concepts in undergraduate biology is not documented. We measured the understanding of key evolution and science skills using multiple-choice and true/false items - before and after the Jigsaw activity. We found a negative correlation between the pre-jigsaw score and the learning change ($r = -0.64$, $p < 0.001$), suggesting that students with less prior knowledge showed greater progress. When divided into low and high performers based on the first test, 73% of low performers improved their score after the Jigsaw, compared to 53% of high performers. This result suggests that the Jigsaw activity is effective in narrowing the gap between low and high performers. This could be due to the use of an authentic situation, the collaborative learning happening during the group discussion to reach a consensus expert opinion, and peer-teaching.

[A233] Agmatoploidy and conserved synteny shape karyotype evolution in holocentric Cyperids

Natalia Castro[1], Dr. André Marques[1]
[1]Max Planck Institute for Plant Breeding Research, Cologne, Germany

Holocentromeres represent an alternative centromere organization in which centromeric activity is distributed along the entire chromosome length. Holocentricity facilitates chromosomal rearrangements, which can result in chromosome number increases by fissions without whole-genome duplication, a process known as agmatoploidy. In plants, the largest group with predominantly holocentric chromosomes is the Cyperid clade (families Cyperaceae, Juncaceae, and Thurniaceae). However, recent cytogenetic and genomic studies have identified monocentric species within Juncaceae and Thurniaceae, revealing a complex scenario of karyotype evolution within this group.

Here, we investigated karyotype evolution across Cyperids using comparative synteny analyses based on genomic data. Synteny analyses were conducted at clade-wide and family-level scales to assess patterns of genomic conservation, chromosomal rearrangement dynamics, and signatures of ancestral karyotype organization.

We identified conserved syntenic blocks shared across families, highlighting deep conservation of ancestral genomic regions even after dysploidy events. Within Cyperaceae, we observed chromosome number conservation (n = 5) and high synteny among different genera, suggesting that an ancestral base chromosome number (x = 5) may have been retained and subsequently increased through agmatoploidy. Likewise, Juncaceae exhibited chromosome number variation associated with chromosome fusions as the main driver of karyotypic differences between Juncus and Luzula. Within Luzula, most species shared n = 6 and displayed high collinearity. Interestingly, L. denticulata exhibits a doubled chromosome number also driven by agmatoploidy. Together, our results suggest that karyotype evolution in Cyperids is dominated by dysploidy, while conserved synteny provides clues about ancestral karyotype organization of the Group.

[A234] Collateral fitness effects of mutation are not commonly caused by protein misfolding

Natalie Quan[1], Yuichi Eguchi[1], Ashley Brown[1], Dr. Kerry Geiler-Samerotte[1]
[1]Arizona State University, , United States

Proteins misfold frequently in all cells, and this process has long been viewed as a universal source of mutational harm. Here we provide a broad, quantitative test of how strongly protein stability predicts cellular fitness. By integrating deep mutational scans spanning eight proteins from both bacteria and yeast, expressed under conditions where their function is either essential or gratuitous, we find that destabilizing mutations predict fitness only when the protein's activity is required for growth. When expression is gratuitous, predicted misfolding fails to explain the collateral fitness costs of mutation, even for variants known to misfold biochemically. These findings suggest that misfolding is not a universal, or even a dominant, driver of mutational harm and that other cellular processes may instead underlie the hidden costs of mutation.

[A235] Genetic variation in adrenarche-associated genes suggests shorter childhoods in Neanderthals

Nicholas Hartman[1], Fernando A. Villanea[1]
[1]University Of Colorado Boulder, Boulder, United States

Neanderthals and anatomically modern humans share a close evolutionary relationship, yet they evolved in markedly different environments, subject to distinct selective pressures. These varied selective pressures gave rise to notable differences between the two groups; existing literature suggests that Neanderthals reached adulthood earlier than modern humans. Among living primate species, early growth periods can be truncated or extended as an adaptation to environmental pressures. Humans exhibit a unique pre-puberty stage of development called adrenarche, which extends middle childhood. However, it remains untested if Neanderthals followed this pattern. In this study, we conducted a comparative analysis of the genetic variation of the CYB5A, SULT2A1, HSD3B1, HSD3B2, and CYP17 genes in both Neanderthals and modern humans. We identified ten unique Neanderthal genetic variants across the genes CYB5A (six variants), CYP17A1 (two variants), and SULT2A1 (two variants) not found in non-introgressed human populations. Of these variants, the SULT2A1 and CYB5A variants were also found at varying frequencies in introgressed human populations. In reports of clinical significance, one CYP17A1 variant could cause a deficiency of the CYP17A1 enzyme, and one SULT2A1 variant is associated with a decrease in DHEAS levels (a key hormone in adrenarche). Our future goals are to link these genetic variants with their functional significance on the physiological level. Understanding the genetic factors that impact the adrenarche stage of development in Neanderthals can provide valuable insights into their life histories beyond what the fossil record can show us.

[A236] Genetic Basis of Skeletal Morphology in Archaic Humans

Nicholas Holly[1], Kelsey Witt[1]
[1]Institute of Human Genetics and Department of Genetics and Biochemistry, Clemson University, Clemson, United States

Neanderthals and Denisovans diverged from Anatomically Modern Humans (AMH) ~500kya, and diverged from each other ~400kya. The different lineages have had several admixture events since, with most modern human populations having a small level of Neanderthal and Denisovan DNA, and ancient archaic genomes even showing signs of admixture with each other. However, there are several striking morphological differences in skeletal structure between AMH and archaic humans. Neanderthals and Denisovans share many unique features, such as a generally shorter, broader stature, as well as a more prominent brow and elongated skull, likely owing to their more recent shared ancestry. We examined the differences in the skeletal morphogenesis genes across AMH and archaic humans to uncover the possible genetic basis of Neanderthals and Denisovans' differences in skeletal structure from AMH. We screened the three high coverage Neanderthal genomes for fixed differences relative to AMH and uncovered a number of variants fixed in Neanderthal populations with possible functional significance, including missense variants in the HOXD and HOXA genes, regions often associated with limb anomalies in clinical cases in modern populations. Many of the variants of interest are also shared with the Denisovan genome, tracking with their similar morphology. This points to a high likelihood that the differences in skeletal morphology can be tracked to the genetic level, uncovering new insights about the evolution of the human skeleton.

[A237] Beyond static reference trees: a dynamic framework for human mtDNA phylogeny

Nicole Huber[1], ao. Univ.-Prof. i. R. Mag. Dr. Arne Dür[2], Ao.Univ.Prof. Walther Parson[1,3]
[1]Medical University Of Innsbruck, Innsbruck, Austria, [2]University of Innsbruck, Innsbruck, Austria, [3]Forensic Science Program, The Pennsylvania State University, , USA

Across disciplines, human mitochondrial DNA (mtDNA) phylogeny has traditionally been represented by curated reference trees that provide a stable and interpretable basis for classification and comparison. While such resources are foundational, they are not inherently designed for the systematic and continuous integration of newly generated sequence data. In parallel, the field of pathogen evolution has developed mature infrastructures for real-time phylogenetic analysis, versioning, and interactive exploration.

Here, we present an approach that builds on the curated mitoLEAF mtDNA backbone and applies concepts developed in pathogen phylogenetics to human mtDNA evolution. By embedding the workflow within the Nextstrain ecosystem, we enable phylogeny-guided, versioned expansion of mtDNA lineages while maintaining backbone stability and controlled nomenclature. Updates are handled in a transparent and reproducible manner, allowing phylogenetic changes to be evaluated and tracked over time.

By integrating constrained phylogenetic inference with mutation and metadata context, and by leveraging interactive visualization through Auspice, the framework facilitates systematic inspection of lineage structure, emerging subclades, and gaps in current sampling. Transferring methodological advances from pathogen genomics to human mtDNA offers a pathway to modernize mitochondrial phylogenetic analysis. This approach complements traditional static reference trees with a dynamic, update-friendly framework that benefits evolutionary biology, population genetics, and applications reliant on accurate haplogroup resolution.

[A238] Assembly of megabasepair-long ribosomal DNA arrays using advanced Nanopore basecalling

Nikol Poláková[1], Tamara Potapova[3], Ondřej Hanák[1], Haoyu Cheng[4], Jennifer Gerton[3], Karen H. Miga[2], Monika Cechova[1,2]

[1]Faculty of Informatics, Masaryk University, Brno, Czech Republic, [2]UC Santa Cruz Genomics Institute, University of California, Santa Cruz, USA, [3]Stowers Institute for Medical Research, Kansas City, USA, [4]Department of Biomedical Informatics and Data Science, Yale School of Medicine, New Haven, USA

Ribosomal DNA (rDNA), encoding RNA machinery required for protein synthesis, remains the last unresolved region of human telomere-to-telomere assemblies. Although long-read sequencing has enabled accurate assembly of most repetitive and complex genomic regions, standard methods fail for rDNA arrays due to insufficient sequencing coverage and large repeating units. To overcome this challenge, we applied a highly accurate experimental ONT basecalling model, named hyperbasecalling, to rDNA reads derived from whole-genome sequencing and adaptive sampling datasets (~56x and ~29x coverage, respectively). This model substantially improved read-level accuracy, increasing high-quality reads (>Q25) from 31.4% to 46.5% compared to the super-accuracy model. Using Hifiasm with these high-quality datasets, we tested multiple assembly strategies and successfully reconstructed complete rDNA arrays on chromosome 14 for both haplotypes. We validated the paternal haplotype using both uniquely-mapping long reads, as well as experimental estimates, including those from the parental data. The maternal array displayed periodic hypo- and hypermethylation, whereas the paternal array showed uniformly hypermethylated rDNA units across its entire length. Overall, ~41% and ~59% of the reads exhibited active and inactive signatures. Multiple sequence alignment of individual rDNA units revealed high sequence conservation within each haplotype (86.7% in paternal, 92.2% in maternal). Most differences corresponded to indels of repeated motifs rich in C and T nucleotides, with the paternal array showing extra LR repeats and the maternal TR repeats. These results demonstrate that accurate basecalling combined with strategic read selection improves rDNA assembly, providing a foundation for studying sequence variation and genomic function.

[A239] A ballad of Songbird and Snake: Towards T2T reference genomes for non-model species

Nivesh Jain[1], Giulio Formenti[1], Jack A Medico[1], Marco Sollitto[1,2], Jennifer Balacco[1], Dr Erich Jarvis[1]
[1]The Vertebrate Genome Lab, The Rockefeller University, New York, United States, [2]Department of Biology, University of Florence, Florence, Italy

At the Vertebrate Genome Lab (VGL), we have generated high-quality reference genomes for hundreds of non-model species. A successful genome assembly relies on a complementary data "recipe". We have now successfully developed a novel approach that combines PacBio HiFi sequencing for highly accurate contigs, Oxford Nanopore Technologies (ONT) ultra-long (UL, >100 kb) reads to span repetitive and structurally complex regions, and Arima Hi-C data to provide phasing information and chromosome-scale validation. ONT UL requires the extraction of ultra-high molecular weight (UHMW) DNA, and we have been optimizing protocols to allow the generation of UL data across diverse taxa. This strategy allows the generation of Telomere-to-Telomere (T2T) genome assemblies.

Using this approach, we generated the first fully phased, diploid telomere-to-telomere reference genome for the Zebra Finch (Taeniopygia guttata; preprint available), closing nearly all remaining gaps and adding approximately 90 Mbp of previously missing sequence. This assembly includes complete T2T sequences for all microchromosomes, including dot chromosomes, as well as chromosome 16, which was previously almost entirely absent from the reference genome. The resulting genome is comprehensively annotated for genes, repetitive elements, structural variants, and DNA methylation derived from long-read sequencing. We now plan to routinely extend this strategy to selected non-model species, such as the Olive Python (Liasis olivaceus).

[A240] Evolution of human bipedalism via genetic regulation

Noam Priel[1], Gal Bodek[1], Amit Philosoph[1], Adi Rozenblatt[1], Guy Hirsh[1], Sergey Viukov[1], Yaqub Hanna[1], Dr. David Gokhman[1]

[1]Weizmann Institute Of Science, Rehovot, Israel

Human bipedalism is a defining feature of our lineage. It is supported by a suite of anatomical adaptations that accumulated gradually over the past ~7 million years. Increasing evidence suggests that morphological innovations arise primarily through spatial and temporal changes to gene expression levels mediated by cis-regulatory elements such as promoters and enhancers. To investigate the regulatory basis of bipedal locomotion, we generated hybrid human–chimp and human–gorilla induced pluripotent stem cells (iPSCs). In these hybrid cells, both genomes share a common cellular environment, thereby eliminating trans-acting regulatory differences and enabling the isolation of cis-regulatory effects. We differentiate these hybrid cells into limb bud-derived cartilage organoids, which model developmental programs central to endochondral ossification and the final morphology of long bones. We quantified gene expression across various stages of chondrogenesis to identify human-specific regulatory changes that shape cartilage properties. Our human-ape hybrid cartilage organoids shed light on the developmental mechanisms through which cis-regulatory evolution contributed to human bipedalism.

[A241] A Neural Network Framework Reveals Evidence of Ghost Introgression in Denisovan Introgressed Sequence in Modern Humans

Noel Mcallister[1], Dr. Xinjun Zhang[1], Dr. Sebastian Zöllner[1]
[1]University Of Michigan, Ann Arbor, Ann Arbor, United States

It has long been speculated that modern humans received genetic material indirectly from deeply diverged hominin species through archaic intermediates such as Denisovans, a process known as ghost introgression. Such events are hard to infer because the source populations lack reference genomes. Previous works relied on summaries of allele frequency data such as the site frequency spectrum (SFS), but strong data compression from the SFS reduces power to detect deeply diverged lineages. Here, we develop a Neural network framework for detecting ghost introgression that integrates summary statistics of pairwise sequence differences within and between individuals. Because ghost introgression is expected to generate unusually deep genealogies, pairwise sequence differences serve as proxies for local tree depth and time to the most recent common ancestor, improving sensitivity to introgressed lineages older than the Neanderthal-Denisovan common ancestor without explicit genealogical reconstruction. Detection of ghost introgression in Denisovans is further complicated by the limited availability of high-quality Denisovan genomes. To address this limitation, we train our model using joint information from Denisovan genotypes and modern human haplotypes with possible Denisovan ancestry across fixed-size genomic windows. We simulated 50kb windows and trained the network using over 1.25 million windows balanced between introgressed and non-introgressed classes. We applied our model to 7 Oceanian individuals and the high-coverage Altai Denisovan genome and discovered approximately 2mb of genome sequence harboring evidence of ghost introgression. Our work demonstrates the practicability of machine learning approaches to investigate complex demographic histories under severe data limitations.

[A242] Time-Series Ancient Genomes Reveal Persistence and Connectivity in Patagonia

Patricio Pezo[1], PhD Flávio De Angelis[2], André Strauss[3], Julieta Gómez Otero[4], Silvia Dahinten[4], Rosa Fregel[5], Eduardo G Amorim[2], Maria Laura Parolin[4], Tábita Hünemeier[1]
[1]Instituto De Biología Evolutiva (CSIC-UPF), Barcelona, Spain, [2]Biology Department, California State University, Northridge, United States, [3]Museu de Arqueologia e Etnologia, Universidade de Sao Paulo, Sao Paulo, Brazil, [4]Instituto de Diversidad y Evolución Austral, CONICET, Puerto Madryn, Argentina, [5]Departamento de Genética, Universidad de La Laguna, La Laguna, España

Ancient DNA provides a powerful framework to disentangle long-term genetic continuity from recent demographic connectivity, particularly in regions that remain poorly represented in genomic studies. Here, we investigate this distinction in northeastern Atlantic Patagonia—one of the most understudied regions of southern South America—by integrating a time series of medium- to high-quality ancient genomes (~2,000–300 years BP; mean coverage ~4–7×) with whole-genome data from present-day individuals who self-identify as Mapuche–Tehuelche from the same region.

We show that Ancient Patagonians individuals harbor a distinct Atlantic Patagonian ancestry component that persists in present-day Mapuche–Tehuelche populations, providing evidence of partial genetic continuity across nearly two millennia. This persistence is primarily supported by population structure analyses (ADMIXTURE and Treemix), consistent with the survival of an ancient regional gene pool rather than complete population replacement. Simultaneously, multiple lines of evidence reveal a substantial contribution from Mapuche–Huilliche populations from south-central Chile, accounting for ~39% of the ancestry in present-day individuals. This signal is consistently detected across ADMIXTURE, Treemix, and f-statistics. Recent connectivity is further supported by identity-by-descent (IBD) sharing, indicating relatively recent gene flow, while runs of homozygosity (ROH) distinguish contrasting demographic histories. Ancient Patagonians individuals show extensive long ROH segments, reflecting small effective population sizes and strong genetic drift, whereas present-day Mapuche–Tehuelche exhibit a reduced ROH burden, consistent with recent admixture and increased genetic diversity.

Together, our results demonstrate how time-series ancient genomics from understudied regions can disentangle genetic persistence from recent connectivity, refining evolutionary histories of Indigenous populations in southern South America.

[A243] Insights from and limits to a mosaic molecular clock model for prokaryotes genomes

Chloé-Agathe Azencott[1], Paul Etheimer[1], Michael Sheinman[2], Peter Arndt[3], Tommaso Stentella[3], Florian Massip[1]
[1]CBIO, Mines Paris PSL, Institut Curie, INSERM U1331, Paris, France, [2]Department of Physics of Complex Systems, Weizmann Institute of Science, Rehovot, Israel, [3]Max Planck Institute for Molecular Genetics, Berlin, Germany

The mosaic method is a new pairwise distance method that can be used to build phylogenetic trees. It uses a distinctive way to summarize whole genome alignments of bacterial genomes, the distribution of the distances between consecutive substitutions - that we call the match length distribution (MLD). It was shown that those distributions follow two regimes, containing information on vertical and horizontal inheritance respectively, in the case of the well-studied Enterobacteriaceae family (Sheinman et al, 2024). From the MLD it is possible to estimate evolutionary parameters, such as the time divergence and horizontal gene transfer rates with comparatively few assumptions. This co-estimation aims to infer pairwise distances to reconstruct phylogenies while controlling for horizontal transfer, very common in Bacteria. In this study, we extend this previous work by applying to another clade of Bacteria, the Bacillales order, and to the Methanothermobacter clade of Archaea, thus gauging the procedure's ability to generalize to other clades, deeper evolutionary relationships, and another life domain. We also systematically evaluate it using simulations. We show that this model also applies in other parts of the prokaryotes' tree of life. However some model hypotheses break down for distant species. In addition, simulating genomes along tree branches according to the model proved more challenging than expected. It revealed the need for a more complete model, incorporating rate evolution, but also showed that the simple initial model is reliable, if misspecified. Together with estimating pairwise distances, this model is informative for prokaryotic genome evolution as a whole.

[A244] Historic Overexploitation, Genetic Erosion and Local extinction: Palaeogenomic Insights into the Decline of Eubalaena glacialis in Northeast Atlantic

Dr Goncalo Themudo[1], DR Alba Rey-Iglesia[2], Ines M. dos Santos[3], MSc Rebecca Netels[1], Dr Alfredo López[4], Jose Martínez-Cedeira[4], Dr Rute R. da Fonseca[5], Paula F. Campos[1]
[1]CIIMAR, University of Porto, Matosinhos, Portugal, [2]The Globe Institute, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark, Copenhagen, Denmark, [3]University of Minho, Braga, Portugal, [4]CEMMA, Coordinadora para o Estudio dos Mamiferos Mariños, Apdo 165, 36380 Gondomar, Spain, Nigran, Spain, [5]Center for Global Mountain Biodiversity, GLOBE Institute, University of Copenhagen, Universitetsparken 15, 2100 Copenhagen, Denmark, Copenhagen, Denmark

The North Atlantic right whale (NARW, Eubalaena glacialis) was the primary target for early industrial whalers, particularly the Basques. Once abundant on both sides of the Atlantic, inhabiting temperate and subpolar waters, these whales are now functionally extinct in the Northeast Atlantic and critically endangered. Despite their fragmentary remains, bones are abundant in the archaeological and historical records and reveal a long history and extensive tradition connected to the exploitation of these marine resources.

Using state of the art ancient DNA techniques we characterize genetically the functionally extinct population of NARW from the Northeast Atlantic. We sequenced 17 full genomes from historical specimens from the Cantabrian Sea, dating from the 13-18th century. These data were compared with modern individuals from the Northwest Atlantic. Our results highlight differences in genetic diversity over time, shedding light on the impact of whaling on this species.

Our results suggest that NARW was panmictic, and that there was a significant decrease in diversity since the Middle Ages, with the extinction of most of the lineages present at the time. This work clearly points to the devastating effects commercial whaling had on North Atlantic Right Whales in the Cantabrian Sea and the NARW population.

[A245] Mutational bias in GRINS: How coding sequences accommodate intense nucleotide skews

Paula Thiel Pizarro[1], Prof.Dr. Andreas Wagner[1,2], Dr. Aleksandra Nivina[1]
[1]University of Zurich, , Switzerland, [2]The Sante Fe Institute, , United States of America

GRINS are genetic repeats of intense nucleotide skews. These elements span long stretches of DNA with highly biased distributions of G versus C and T versus A between the two strands. The atypical nucleotide biases in GRINS are also associated with gene conversion, a unidirectional recombination process between homologous regions. In bacteria, GRINS are commonly found in natural product biosynthetic clusters, for instance, in the genes encoding modular polyketide synthases (PKSs). The extent to which GRINS change the nucleotide composition of modular PKSs genes is remarkable: these regions often span thousands of base pairs, with long stretches of DNA predominantly composed of G+T or C+A.

Our goal is to understand how modular PKS genes accommodate such extreme nucleotide skews while remaining active. To address this, we analyze codon usage patterns, map the nucleotide biases along protein sequences, evaluate their effects on DNA and RNA structure, and study the selection pressures acting on GRINS. By comparing genes with and without GRINS, we aim to describe how these nucleotide compositions influence protein divergence. We hypothesize that specific sites tolerate nucleotide skews better and that shifts in mutational bias associated with GRINS enable the exploration of novel mutations that were previously inaccessible. Overall, we aim to determine how intense nucleotide skews shape and constrain the encoding of PKSs. In doing so, we aim to uncover how the atypical nucleotide patterns in GRINS drive the diversity of bacterial natural product biosynthetic clusters.

[A246] Investigating Gene Expression Noise in Embryogenesis – Evolutionary Implications for Duplicate Gene Retention and Compensation

Pavol Kramár[1], Richard J White[1], Ian M Sealy[1], Elisabeth M Busch-Nentwich[1]
[1]School of Biological and Behavioural Sciences, Queen Mary University of London, E1 4NS, , United Kingdom

Gene expression noise (GEN) is defined as the variation in mRNA and/or protein copy numbers of a given gene which stems from the stochastic nature of the biochemical reactions comprising transcription, translation and post-translational modification (Chapal et al., 2019).

In this project, I test the hypothesis that GEN is an evolutionarily conserved factor implicated in paralog retention and compensation following whole-genome duplication. GEN is associated with regulatory complexity and transcriptional plasticity, allowing genes to explore novel phenotypic spaces (Chapal et al., 2019). Such novelty is conducive to neo- and sub-functionalization, i.e., the adoption of novel functional roles by one member of a paralog pair and the partitioning of the ancestral function between both members of a paralog pair, respectively. Both neo- and sub-functionalization are widely accepted models of duplicate gene retention, which is a pre-requisite to paralog compensation (Cai and Marais, 2025; Mattenberger et al., 2017).

Previous investigations have found that paralogs exhibit increased GEN is some model systems (Dong et al., 2011). However, there is no systematic, cross-species, spatiotemporally-resolved comparison of the GEN of paralogs and singletons spanning the entirety of embryonic development.

I employed developmental time-course scRNA-seq data sets to characterize the transcriptional variance landscapes of seven multicellular organisms, varying in their phylogenetic position. I show that in some species, paralogs are more likely to exhibit an evolutionarily conserved and biologically meaningful noise signal compared to singleton genes.

Historically, GEN has been overlooked as an unfavorable feature of complex systems. Here, I provide evidence to a shifting perspective which views GEN as an evolvable trait which is shaped by natural selection and associated with biological function (Raser and O'Shea, 2004; Wolf et al., 2023).

[A247] Identifying selection in Plasmodium populations through IBD troughs rather than peaks

Peng-Yin Ng[1], Professor Hsiao-Han Chang[1]
[1]Institute of Bioinformatics and Structural Biology, National Tsing Hua University, Hsinchu, Taiwan
Identical-by-descent (IBD) analysis has been widely used to identify selection events in Plasmodium parasite populations, with most studies focusing on genomic regions showing exceptionally high IBD as likely targets of positive selection. In our previous study of population dynamics in São Tomé and Príncipe, however, we observed that certain genomic regions consistently exhibited low levels of IBD even within genetic clusters defined by very high overall relatedness (IBD > 0.9). This pattern was shared across clusters, suggesting that selective forces may instead be maintaining or increasing diversity in these regions. To assess how universal this pattern is and to evaluate the utility of this approach for identifying genes under selection, we analyzed worldwide Plasmodium parasite sequences from the open Pf7 dataset using a similar IBD-based framework. Consistent with our earlier findings, clear IBD troughs were detected in most large genetic clusters across ten countries. We hypothesized that these troughs would be enriched for immune-related genes due to diversifying selection. Surprisingly, IBD troughs were instead enriched for genes annotated in the diversity category, while immune-related genes showed a contrasting pattern. The absence of strong antigenic variation signals in intra-cluster analyses suggests that immune-related genes may contribute more prominently to differentiation between clusters rather than to variation within clusters. Ongoing work will further characterize genes in the diversity category and extend the analysis to inter-country comparisons to better understand patterns of selection across geographic regions.

[A248] Tracing the Evolution of Eye Pigmentation Genes in Arthropods and Identification of Such Genes in Ostracod Crustacean Skogsbergia sp.

Phoebe Hall[1], Dr. Todd Oakley
[1]University Of California, Santa Barbara, Santa Barbara, United States
Arthropod eyes exhibit morphological diversity while generally being shaped by a conserved developmental genetic toolkit, supporting investigations into how a shared toolkit can give rise to a variety of phenotypes. Among the conserved genes, white, brown, and scarlet encode ABC transporters required for eye pigmentation and are characterized across model arthropods, yet remain unannotated in many non-model organisms. Ostracod crustaceans are an interesting system for studies of eye evolution and development, particularly because all bioluminescent courtship signaling ostracods share a conserved eye structure which is not present in other ostracods. However, progress developing ostracods as a study system is limited by a lack of annotated genes and functional tools. In this study, I discover and phylogenetically validate orthologs of white, brown, and scarlet in the ostracod Skogsbergia sp. Candidate sequences were identified through BLAST searches against a transcriptome, filtered and translated into protein sequences, and confirmed through phylogenetic analyses alongside known arthropod orthologs. I use these analyses to expand and update existing published arthropod eye pigmentation gene trees from over ten years ago to deepen the understanding of the evolution of such genes. Eye pigmentation genes have a record of use as reporter gene knockdown targets due to the simple phenotypic readout of successful knockdown, thus identification of these genes enables the development of genetic tools in ostracods. This work advances both comparative studies of arthropod eye evolution and the experimental tractability of ostracods as a model system.

[A249] Comparative Genomic Analysis of Independently Miniaturised Annelids

Pia Merete Eriksen[1]
[1]University Of Copenhagen, Copenhagen, Denmark

Aquatic biodiversity is dominated by meiofauna – ecologically vital invertebrates less than a millimetre long. Multiple meiofaunal lineages have evolved independently through miniaturisation within at least twelve of the macroscopic animal phyla, making miniaturisation an extremely common phenomenon. These transitions from macroscopic to microscopic body plans involve dramatic morphological changes, which can include reduction of appendages, simplification of circulatory and sensory systems, and loss of adult-associated morphology. But how are these changes reflected in the genome?

In Annelida (containing bristleworms and earthworms), one well-studied miniaturised species, Dimorphilus gyrociliatus, displays a simplified annelid segmental body plan of only six segments, reduced body cavity, and no appendages. On a genomic level, D. gyrociliatus has a greatly reduced genome, with lower TE repeats and evidence of intron loss. Crucially, it is not known whether these genomic signatures are shared by other, independently miniaturised lineages.

By leveraging recent advances in genomic-techniques, we are now able to generate full, chromosome-scale genomes for these ultra low-input specimens. We generated a chromosome-level genome for the miniaturised annelid Diurodrilus subterraneus, and explored its genomic landscape, assessing genome size, Hox gene repertoire, differential gene expression, and structural elements. We then compared this to Dimorphilus gyrociliatus in order to determine genomic similarities between these two independently miniaturised annelids.

This comparison marks a new foray into the previously unexplorable realm of meiofaunal comparative genomics, and establishes the groundwork for future explorations into the genomic mechanisms underpinning miniaturisation – one of the most widespread evolutionary transitions across the animal tree of life.

[A250] Making the step to multiple hosts: the genomic and transcriptomic drivers of diet plasticity in Tephritis conura flies

Dr Rachel Steward[1], Yajuan Huang[1,2], Dr. Jesús Ortega[1,3], Dr. Kalle Nilsson[1], Anna Runemark[1]
[1]Biology Department, Lund University, Lund, Sweden, [2]Department of Medical Biochemistry and Cell biology, University of Gothenburg, Gothenburg, Sweden, [3]Cavanilles Institute of Biodiversity and Evolutionary Biology, Universidad de Valencia, Paterna, Spain

Phenotypic plasticity is a critical mechanism for colonizing new habitats. By buffering against environmental shifts, plasticity may facilitate adaptive divergence at both micro- and macroevolutionary scales. However, the evolution of these plastic phenotypes remains a significant knowledge gap in the context of environmental change. We investigated the evolution of diet plasticity in the Peacock fly, Tephritis conura, an extreme specialist that lays eggs and develops in thistle buds. While continental populations are restricted to a single host species, Scottish populations have expanded their diet to include multiple hosts. By integrating genomic and transcriptomic data, we aimed to identify the molecular basis of this expanded host use. We found no genomic differentiation among Scottish flies across different hosts, confirming that these represent plastic phenotypes within a panmictic population. Furthermore, while comparisons between Scottish and continental populations revealed potential mechanisms for interacting with and adapting to different host plants, genomic comparisons between plastic and specialist lineages suggest a lack of parallelism between plastic host use and host-associated differentiation. Ultimately, this work advances our understanding of the interplay between genomics and plasticity, providing a foundation for future research into the genomic features underlying multiple host use and ecological speciation.

[A251] Urea transporter evolution: deep conservation, recent adaptation in mammals, and maintenance of the Jka/Jkb polymorphism in the past ~110,000 years

Rachele Cagliani[1], Diego Forni, Uberto Pozzoli, Alessandra Mozzi, Manuela Sironi
[1]Scientific Institute IRCCS E. Medea, Bosisio Parini, Italy

Urea is a central metabolite and many organisms encode membrane integral urea transporters (UTs). Here, we combined motif homology searches, phylogenetics, and molecular evolution with paleogenomics to investigate the evolutionary dynamics of UTs from prokaryotes to human populations. We discovered previously unknown combinations of UT domains with other functional modules in metazoa, protists, and bacteria, suggesting a wider range of functions and regulatory mechanisms for UTs than previously understood. The early origin of UT domains allowed the identification of specific residues that have remained conserved across billions of years. Contestually, we found evidence that UT-B was a target of positive selection in mammals. In particular, selection targeted UT-B in bats, primates and rodents, possibly as the result of a pressure exerted by blood pathogens. In human populations, a single variant (Asp280Asn) in the UT-B protein is responsible for the common Kidd blood group antigens. Here we provide direct evidence for the temporal stability of the Asp280Asn polymorphism over ~110,000 years in Europe and Asia. While we previously proposed this variant to be under balancing selection, this study is the first to use ancient DNA to track its allele frequency through deep time. This novel application of paleogenomics confirms that the polymorphism was maintained at stable frequencies over time, in different European sub-regions, and across continents. Our data show how paleogenomics can provide information on the selective processes in humans, not only limited to directional selection, but also to balancing selection.

[A252] The genomic tug-of-war: disentangling the biological drivers of cytonuclear discordance across angiosperm.

Radhika Dutt, Praveen Karanth

Phylogenetic trees remain our primary tool for tracing evolutionary histories, yet a growing body of evidence reveals that different regions of the genome do not always agree on the same history. This phenomenon — phylogenetic discordance — has emerged as one of the central challenges in modern systematics. Nowhere is this challenge more tractable than in plants, where every cell harbour three independently evolving genomes: a biparentally inherited, recombining nuclear genome, and two maternally inherited, non-recombining organellar genomes (plastid and mitochondrial). These contrasting inheritance modes make angiosperms an ideal system for studying discordance, because each genome independently records a different dimension of evolutionary history.

We constructed independent phylogenies of angiosperms from nuclear, plastid, and mitochondrial genomes and asked three questions. First, how much do the three genomic histories disagree? Substantially — 46 to 62% of branches differ between compartment pairs, and the AU test confirms this conflict is genuine. Second, where is this conflict concentrated? The two organellar genomes agree most with each other, consistent with shared maternal inheritance, while the nuclear genome diverges most — particularly at deep backbone nodes corresponding to ancient rapid radiations. Third, and most critically, what biological processes drive this discordance? We tested two competing hypotheses — incomplete lineage sorting (ILS) during rapid speciation versus introgression through hybridisation — using complementary approaches that exploit different properties of gene trees. our study predicts that ILS produces symmetric discordance while introgression produces asymmetric discordance; branch length distribution analysis predicts that ILS produces a single rate of coalescence while introgression produces a mixture. Both tests converge: ILS is the dominant process (59–73% of branches), with introgression detectable but confined to 10–27% of branches. The most conflicted lineages evolve significantly faster and occupy the angiosperm backbone — the signature of explosive Cretaceous diversification, not rampant gene flow.

These results shift the narrative around cytonuclear discordance: rather than signalling a web of hybridisation, the conflicting genomic histories of flowering plants are largely the predictable consequence of rapid ancient radiations.

[A253] Discovery and Evaluation of Molecular Mimicry between Epstein–Barr Nuclear Antigen 1 and Unrelated Human Proteins Relevant to Multiple Sclerosis

Raquel Battifora[1], Dr. Jessica Siltberg-Liberles[1]
[1]FLORIDA INTERNATIONAL UNIVERSITY, , United States

Antibody cross-reactivity due to molecular mimicry between Epstein–Barr virus (EBV) and human proteins may contribute to the increased risk of Multiple Sclerosis (MS) following EBV infection. MS patients commonly exhibit antibodies that strongly bind Epstein–Barr Nuclear Antigen 1 (EBNA1). Here, we used a computational approach to identify EBNA1 regions with structural similarity to human protein epitopes. EBNA1 structures were analyzed using Epitopedia, a pipeline that detects three-dimensional similarity between pathogen peptides and known immune epitopes using experimental PDB structures and human proteome AlphaFold models. Candidate hits were filtered by Homo sapiens, Rattus norvegicus and Mus musculus matches, recurrence across multiple EBNA1 structures, structural similarity scores, and relevance to MS. Myelin Associated Glycoprotein (MAG) and Coiled-Coil Alpha-Helical Rod Protein 1 (CCHCR1) emerged as proteins of interest. MAG and CCHCR1 are not evolutionarily related and have distinct biological roles, yet both contain epitope regions that resemble EBNA1, suggesting unexpected similarity that may promote immune misrecognition. Phylogenetic analyses showed conservation of the epitope motifs across placental mammals. Structural comparisons using TM-Align indicated high similarity between EBNA1 and CCHCR1 and moderate similarity between EBNA1 and MAG. To assess whether these epitope-level similarities persist under structural flexibility, we generated conformational ensembles (1000 conformations per protein) using BioEmu and performed TM-Score comparisons of the potential epitope regions. This analysis will help determine the distribution of mimicry signals across the conformational ensemble. These findings support further investigation of EBNA1-associated molecular mimicry involving MAG and CCHCR1 as a potential mechanism contributing to MS.

[A254] Protein Evolution Occurs at an Uneven Tempo: Patterns of Rate Variation Across the Tree of Life

Rasit Durak[1], Julien Dutheil[1]
[1]Max Planck Institute For Evolutionary Biology, Plön, Germany

Proteins evolve under a complex interplay of functional constraints and lineage-specific pressures, resulting in heterogeneous patterns of change across sites and through time. One striking but still poorly characterized feature of this process is called heterotachy: the phenomenon whereby individual amino-acid sites undergo shifts in evolutionary rate along different branches of the tree of life. Although heterotachy has long been recognized as a potential source of phylogenetic bias, its prevalence, distribution, and biological determinants across deep evolutionary timescales remain largely unknown. Here, we present a large-scale, phylogenetically comprehensive analysis of heterotachy across the tree of life. Using orthologous protein groups from the OMA (Orthologous MAtrix) database, spanning over 2900 species and all major domains of life, we constructed multiple sequence alignments and applied model-based inference to systematically detect and quantify site-specific rate shifts across phylogenetic branches. Across 4125 orthologous protein families, representing more than 1.8 million aligned amino-acid sites, we find that heterotachy is widespread, with over one quarter of sites showing strong evidence of lineage-specific rate shifts. Beyond documenting its global distribution, we investigate how site-specific properties shape patterns of rate variation, testing whether positional features within proteins influence the likelihood of rate shifts. By integrating large-scale phylogenomic data with explicit analyses of temporal rate heterogeneity, our study provides new insight into the evolutionary processes that generate within-site rate variation. A deeper understanding of heterotachy has important implications for evolutionary modeling, phylogenetic inference, and molecular dating, and sheds light on how proteins diversify across deep evolutionary history.

[A255] Variation in transposable element landscapes across socially parasitic and host ants

Raziyeh Abdilzadeh, Prof. Erich Bornberg-Bauer
[1]Institute for Evolution and Biodiversity, University of Münster, , Germany, [2]Institute for Evolution and Biodiversity, University of Münster, , Germany

Transposable elements (TEs) are major contributors to insect genome evolution, yet closely related species can differ markedly in TE composition. Here, we compare TE landscapes in two Temnothorax longispinosus and Temnothorax americanus ants with contrasting social strategies: a host and a socially parasitic, slave-making lineage. Phylogenetic evidence consistent with Emery's rule supports T. americanus as a derived parasitic lineage relative to its host.

We identify consistent differences in TE landscapes between species. The host genome exhibits higher overall TE abundance and greater TE family diversity, whereas the slave-making ant shows a comparatively reduced overall TE repertoire. Several TE families abundant in T. longispinosus are reduced or underrepresented in RepeatMasker annotations in T. americanus. Divergence in TE content is uneven across classes, with Long Terminal Repeat (LTR) retrotransposons contributing disproportionately to interspecific differences.

TE reduction in the socially parasitic lineage is not uniform. While many TE groups show lower representation, Maverick (Polinton) elements are relatively enriched in T. americanus, indicating class-specific retention rather than genome-wide TE loss. This pattern is consistent with differential persistence of TE classes with distinct structural and evolutionary properties.

Overall, these results demonstrate that closely related ants with contrasting social strategies can exhibit markedly divergent TE landscapes shaped primarily by differential accumulation and retention of existing TE classes, rather than by the emergence of novel TE types. These differences may be consistent with genomic contexts associated with contrasting life histories, including social parasitism, and motivate future work on potential ecological consequences of TE variation.

[A256] A haplotype-based neutrality test, 2D-SFS, uncovers positive selection on an intergenic haplotype in the Japanese population

Risa L. Iwasaki[1], Yoko Satta[2,3], Professor Atsushi Tajima[1]
[1]Department of Bioinformatics and Genomics, Faculty of Medicine, Kanazawa University, 13-1, Takara-machi, Kanazawa, Japan, [2]Research Center for Integrative Evolutionary Science, The Graduate University for Advanced studies (SOKENDAI), Hayama, Japan, [3]Department of Evolutionary Studies of Biosystems, SOKENDAI, Hayama, Japan

Widely used tests for positive selection primarily assess deviations from neutrality at the single-SNP level, often relying on an excess of derived alleles. By treating individual SNPs as the fundamental units of selection, however, these approaches have limited power to detect selection acting on combinations of alleles, particularly haplotypes composed of both derived and ancestral alleles. Here, we apply a haplotype-based neutrality test, the two-dimensional site frequency spectrum (2D-SFS), which detects selection acting on a focal haplotype by comparing intra-allelic variability among chromosomes. This framework enables direct evaluation of selection on specific haplotypes and improves sensitivity to selective processes that may be overlooked by conventional SNP-based approaches.

Using this method, we identified a previously unreported signal of positive selection acting on a specific haplotype in an intergenic region. Based on patterns of linkage disequilibrium, we defined a focal haplotype composed of the derived allele at rs79180241 and two neighboring ancestral alleles. Phylogenetic analysis revealed a star-like pattern, suggesting a rapid recent expansion of this haplotype. Notably, evidence of positive selection was detected exclusively for this haplotype among chromosomes carrying the derived allele.

Although this haplotype is frequent across East Asian populations, a signature of recent positive selection was detected only in the Japanese population, beginning approximately 18 kya. This timing coincides with the post-Last Glacial Maximum and suggests recent local adaptation driven by environmental changes. This haplotype-centered perspective complements existing SNP-based scans and highlights the value of modeling allelic combinations when reconstructing recent, population-specific adaptation in human genomes.

[A257] Investigating the utility of genome-environment association for identifying climate-adapted variation in Aegilops tauschii

Risha Na[1], Dr Adrian Brennan[2], Dr Therése Bengtsson[3], Professor Peter Hambäck[1], Dr Marco Fracassetti[1], Tanja Slotte[1]
[1]Department of Ecology, Environment and Plant Sciences, Stockholm University, Stockholm, Sweden, [2]Department of Biosciences, Durham University, Durham, United Kingdom , [3]Department of Plant Breeding, Swedish University of Agricultural Sciences, Uppsala, Sweden

There is a pressing need to develop more resilient crop cultivars considering the projected effects of climate change on crop yields. Crop wild relatives are promising sources of genetic variation for climate adaptation but remain underutilized. This project aims to test the utility of sequence-based genome-environment association (GEA) for identifying genetic variation relevant to drought adaptation in Aegilops tauschii, the wild progenitor of the bread wheat (Triticum aestivum) D-genome. At the current stage, we have analyzed whole-genome resequencing data of 229 Ae. tauschii originating from across its natural geographical range. Integrating genomic data with high resolution climatic data from the CHELSA database, we apply GEA methods, including redundancy analysis and latent factor mixed models, to identify genetic markers associated with climatic variables that may contribute to local adaptation. We will then validate the performance of GEA by comparing its results with genome-wide association analysis of experimentally measured drought responses. This study will help validate GEA methods for identifying genetic variants involved in climate adaptation and explore their potential to facilitate adaptation characterization of large crop genebank collections.

[A258] Paleogenomic analysis of Mongolian aurochs and cattle reveal connections to both ancient and modern bovids

Robin Singleton[1], Dr Susan Monge[2], Kelsey Witt[1], Dr Katherine Brunson[3], Sloan Williams[2], Dr Lisa Janz[4]

[1]Clemson University, Clemson, United States, [2]University of Illinois Chicago, Chicago, United States, [3]Wesleyan University, Middletown, United States, [4]University of Toronto Scarborough, Toronto, Canada

Aurochs (Bos primigenius) once ranged across Europe and Asia, and populations in the Near East and South Asia gave rise to domestic cattle (Bos taurus and Bos indicus respectively). With the spread of domestic cattle, auroch populations dwindled, leading to their eventual extinction. Before extinction, aurochs were managed alongside and admixed with domestic cattle for millennia. Thus far, most zooarchaeological and paleogenomic research on aurochs has focused on Western Eurasia and Europe, with Eastern Asia remaining understudied. We present novel low coverage genomic sequences from 27 Mongolian aurochs, dating from 1500-8300 YBP and compare them to existing auroch and cattle data—both modern and ancient—to address this gap. Mongolia has a long history of interactions between humans and aurochs and of cattle pastoralism, making it an ideal region to study the roles of domestic cattle and aurochs through time. We find that the Mongolian aurochs resemble Pleistocene North Asian aurochs. Following the introduction of domesticated cattle, Mongolian individuals appear similar to ancient cattle from contemporary Chinese archaeological sites such as Shimao, suggesting a single source population for cattle pastoralism across East Asia. Finally, we find genetic signatures from the ancient Mongolian aurochs in modern East Asian cattle breeds, illustrating the longstanding influence of East Asian aurochs on modern domestic cattle.

[A259] Defensive symbiosis evolves rapidly: experimental evolution reveals viral selection drives Wolbachia spread

Cássia Cesar[1], André Pimentel[1], Rodrigo Cogni[1]
[1]Universidade de Sao Paulo, Sao Paulo, Brazil

Wolbachia is a widespread maternally inherited symbiont infecting approximately 50% of terrestrial arthropod species. Although best known as a reproductive parasite, many Wolbachia strains do not manipulate host reproduction, indicating that alternative strategies—such as mutualism—may promote their spread. Some strains protect Drosophila against viral infection, yet hosts also possess genetically encoded antiviral defenses, raising the question of how host genotype and pathogen pressure jointly shape Wolbachia dynamics. Here, we performed an experimental evolution study over ten generations to test whether viral selective pressure and host genotype (virus-resistant vs. virus-susceptible flies) affect the spread of Wolbachia in Drosophila melanogaster populations. We also asked whether changes in Wolbachia frequency translate into increased host fitness under viral infection. Under virus selection, Wolbachia spread rapidly, increasing from 35% to near fixation in most populations within ten generations. Following selection, Wolbachia presence significantly increased host survival after viral challenge. In contrast, host genotypes had no detectable effect on Wolbachia spread or on post-infection survival. Together, our results provide experimental evidence that pathogen pressure alone can drive the rapid spread of defensive symbionts, supporting natural enemies as a key selective force shaping host–symbiont associations in nature.

[A260] Salvaging genetic data from decades-old degraded giant kelp tissue (Macrocystis pyrifera)

Roy Roberts[1], Dr. Meaghan Clark[1], Dr. Dan Reed[2], Dr. Malin Pinsky[1]
[1]University Of California Santa Cruz, Santa Cruz, United States, [2]University Of California Santa Barbara, Santa Barbara, United States

Giant kelp (Macrocystis pyrifera) supports some of the most productive ecosystems in the world and is central to North American biodiversity and the ocean economy. However, dramatic declines—primarily due to climate change and changing species interactions—have made the protection and restoration of kelp forests a top priority. Understanding the underlying mechanisms that allow or constrain adaptation to climate change is imperative for informing management and restoration efforts. By utilizing historical genomic data, we can directly compare how species were prior to large-scale ecosystem disruptions. The Santa Barbara Long Term Ecological Research site has collected kelp samples monthly for decades. These samples were not collected with the intention of DNA sequencing and thus were not stored optimally to maintain DNA strand integrity. Here, we test methods for salvaging genetic material from 45 samples collected in 2001 at Arroyo Quemada Beach. We employed a modified ancient DNA extraction method designed to target smaller DNA fragments. We successfully retrieved DNA from 42 of 45 historical samples with concentrations ranging from 0.06 ng/ul to 21 ng/ul (mean = 4.2 ng/ul). Initial sequencing results (Illumina 2x61) of 5 individuals show that we have successfully extracted and sequenced Macrocystis pyrifera at low coverage. With increased sequencing effort and optimizations to extractions and library preps we hope to get a much larger portion of the genome covered. Our findings highlight the potential of historical genomic data to enhance our understanding of kelp adaptation and resilience, providing a critical foundation for informing future conservation and management.

[A261] Genetic signature reveals the impact of regional and colonial histories on Mycobacterium tuberculosis evolution in Taiwan

Ruo-Ya Bai[1], Ying-An Chen[1], Chieh-Yin Wu[2], Ju-Hsuan Wang[3], Hsien-Ho Lin[2], Hsiao-Han Chang[1]
[1]Institute of Bioinformatics and Structural Biology, National Tsing Hua University, Hsinchu, Taiwan, [2]College of Public Health, National Taiwan University, Taipei, Taiwan, [3]Interdisciplinary Program of Life Sciences and Medicine, National Tsing Hua University, Hsinchu, Taiwan

Taiwan has experienced multiple waves of colonial events over the past several centuries. Given the restricted cross-country mobility during these periods, colonial episodes may have played a significant role in facilitating pathogen transmission between nations, including Mycobacterium tuberculosis (Mtb). Previous studies have suggested a potential link between Mtb transmission and historical events, as supported by the distinct prevalence of Mtb lineages among specific ethnic populations in different regions of Taiwan. To investigate this connection, we analyzed whole-genome sequencing data from 2,655 Mtb samples collected in Taiwan between 2012 and 2021. We estimated changes in the effective population size (N_e) of Mtb over time and observed a sharp increase in N_e for lineage L2 after 1875, during the Qing dynasty—a period marked by large-scale Han immigration following the lifting of an immigration ban. Similarly, other lineages exhibited increases in N_e following colonization by various nations. Furthermore, coalescent events in phylogenetic analyses and directionality indices supported the timing and direction of transmission, respectively. These findings illustrate how colonial history shaped the evolution of Mtb in Taiwan, highlighting an interesting connection between historical events and pathogen dynamics.

[A262] Dormant immunity in bumblebees

Safira Moog[1], Thomas Joseph Colgan[1]
[1]Institute of Organismic and Molecular Evolution (iomE), Johannes Gutenberg University Mainz, Mainz, Germany

This research project investigates the immunity of bumblebee queens during diapause. Diapause is a dormant state which allows organisms to avoid longer periods of unfavourable climatic conditions. It is characterised by developmental arrest as well as reduced metabolism, making it a relevant strategy for many insect species, including bumblebees. Diapause represents a critical period harbouring challenges such as limited resources and increased risk of infection, especially as reduced metabolic activity may affect immunity. For bumblebees and other insects, the immune system is an important defence against parasite infection and colonisation. However, maintenance and activation of the immune system is also associated with high metabolic costs, which raises the question to which extent insects are able to generate or regulate an immune response during diapause. Furthermore, colder temperatures lead to reduced metabolism in insects, making temperature a crucial factor for immune response during diapause. To improve our understanding of insect immune regulation during diapause and the effect of temperature, we use proteomics to examine the expression of immune proteins such as antimicrobial peptides in the haemolymph of immune challenged bumblebee queens to assess the capacity for immune response during diapause at different temperatures. Additionally, survival and post-diapause colony productivity provide insights into long-term effects of an immune challenge during diapause.

[A263] Ribosome profiling for minimal (gut) consortia for novel gene finding in vivo and in vitro

Klaus Neuhaus[1], Korbinian Pürckhauer[1], Marc Follmer[1]
[1]Technische Universität München, Freising, Germany

Studies on the microbiome often focus on the potential rather than the realized functional activity of microbial communities. Meta transcriptomics and meta proteomics address this gap but face limitations, such as the inclusion of non coding RNA in RNA seq and the restricted resolution of mass spectrometry based proteomics. Ribosome profiling (Ribo seq) offers a powerful complementary approach by sequencing ribosome protected mRNA fragments, thereby providing a direct proxy for protein synthesis with high positional resolution. Ribo seq enables precise proteome inference, discovery of novel and small genes, annotation of open reading frames, and analysis of translational dynamics. However, its application has been largely restricted to single bacterial strains, with only one prior study targeting complex microbiomes. A major challenge is the accurate mapping of short ribosomal footprints within diverse communities containing homologous genes. Defined minimal consortia provide an attractive compromise by reducing complexity while retaining ecological relevance. We established Ribo seq for minimal consortia, demonstrating the method's ability to reveal fine scale regulatory differences and active protein production in mixed cultures. Weakly expressed regions and overlapping genes are suggested to provide the raw material for the evolution of novel genes.

[A264] Retroviral OVEX genes: Evolutionary conservation and possible contribution to avian-specific gonadal functions

Koichi Kitao[1], Kirill Kryukov[2], So Nakagawa[3], Yuya Okuzaki[4], Ken-ichi Nishijima[4], Kenji Ichiyanagi[4]
[1]Riken, Yokohama, Japan, [2]National Institute of Genetics, Mishima, Japan, [3]Tokai University, Isehara, Japan, [4]Nagoya University, Nagoya, Japan

Endogenous retroviruses (ERVs) are genomic remnants of retrovirus integration into host germ cell DNA. The retroviral genes encoded in ERVs are sometimes co-opted for host functions. Most of the co-opted ERV genes have been identified in mammals and are linked to the evolution of highly developed organs in mammals, such as brain and placenta. In contrast, the involvement of ERV genes in birds remains largely unknown. In this study, we identified evolutionarily conserved, avian-specific ERV genes through comparative genomic analyses. We first constructed a list of open reading frames encoding ERV-like proteins (ERV-ORFs) from the genome assemblies of seven Galliformes species, including chicken. Using reciprocal similarity search and phylogenetic analyses, we identified conserved orthologous genes: the previously characterized OVEX1 and a novel gene, OVEX2, which was phylogenetically related to OVEX1. OVEX1 contained open reading frames corresponding to the three major retroviral genes (gag, pol, and env), whereas OVEX2 comprised a pol sequence. OVEX1 was expressed in the ovary, while OVEX2 was expressed in both the testis and ovary. Single-cell RNA-seq data analyses further showed that OVEX1 expression was restricted to the ovarian cortex and granulosa cells, whereas OVEX2 was broadly expressed in male and female gonads. We are currently analyzing OVEX1 and OVEX2 sequences from hundreds of avian genomes to investigate natural selection and lineage-specific presence or absence. These analyses are expected to provide insights into the evolutionary and biological roles of ERV-derived genes in avians.

[A265] Genomic Analysis of Color Pattern Polymorphism in Red Palm Weevils

Kripa Shrestha[1], Neelu Begum[1], Dr Mariana Lyra[1], Theo Busschau[1], Stéphane Boissinot[1]
[1]The Evolutionary Genomics Lab, Center for Genomics and Systems Biology, New York University Abu Dhabi, Abu Dhabi, United Arab Emirates

Color pattern polymorphism is one of the most striking and evolutionarily significant forms of phenotypic diversity in nature. As a highly visible trait, color pattern variation influences a wide range of ecological and behavioral processes, providing a powerful framework for investigating the mechanisms underlying phenotypic evolution. In the Red Palm Weevil (Rhynchophorus ferrugineus), a globally invasive pest with severe economic and environmental impacts, the pronotum exhibits an extensive variation in the size, shape, and distribution of melanic spots, resulting in diverse color patterns. Despite its prominence, the genomic basis of this phenotypic variation remains poorly understood. Here, we integrate image-based morphometric analyses with genome-wide association studies (GWAS) in a globally sampled cohort to characterize color pattern variation, resolve its geographic structuring, and identify its genomic basis. Morphometric analyses reveal multiple recurrent color pattern morphs that exhibit pronounced geographic structure consistent with the species' invasion history. We then identify the genomic region responsible for color pattern polymorphism in this species, and examine the evolutionary processes contributing to these phenotypes. This study highlights the significance of color pattern polymorphism in R. ferrugineus and provides a framework for understanding phenotypic diversification in invasive species.

[A266] HLA–KIR Interactions and Active Tuberculosis Risk in Khoe-San Descent Populations

Kristin Hardy[1], Dr. Neus Font-Ponterias[1,2], Oshiomah Oyageshio[1], Gillian Meeks[1], Dr. Katherine Kichula[2], Dr. Stacey Collister[2], Noah Cline[2], Lena van der Westhuizen, Sabiena Andres, Justin Myrick[1], Caitlin Uren[3], Professor Marlo Möller[3], Dr. Paul Norman[2], Dr. Brenna Henn[1]
[1]University Of California, Davis, , United States, [2]University of Colorado Anschutz, Aurora, United States, [3]Stellenbosch University, Stellenbosch, South Africa

The Northern Cape of South Africa has among the highest incidence of tuberculosis (TB) in the world, with 80% of the population having a latent TB infection. This region is home to individuals of Khoe-San descent, who represent the most genetically diverse population in the world due to their deep ancestral lineages and recent admixture from European, Indonesian, equatorial African, and South Asian populations. Most TB infected individuals are latently infected; however 5-10% of these will progress to active infections. Given the high prevalence of latent TB and genetic diversity in this population, variation in immune loci regulating host defense may be a key determinant of progression to active disease. The human leukocyte antigen (HLA) locus contains genes that are integral to the innate immune system recognizing pathogens. The HLA encodes ligands on the surface of nucleated cells that interact with killer cell immunoglobulin-like receptors (KIR) on the surface of natural killer cells. We identify extreme allelic variation across the HLA in the Northern Cape TB cohort. We tested HLA gene association with active TB using logistic regression models incorporating known risk covariates in n=1000. HLA associations are challenging to model given the extreme diversity at many of the loci. We also model canonical HLA–KIR inhibitory systems (A3/A11–KIR3DL2, Bw4–KIR3DL1, and HLA-C C1/C2–KIR2DL1/2DL2/2DL3), KIR gene haplotypes, and HLA–KIR interaction scores to assess their associations with active TB infection. These analyses will determine whether functional receptor–ligand interactions influence active TB risk in South African Khoe-San descent populations.

[A267] Mitospecies: a novel concept for describing molecular diversity

Krisztián Szabó[1], Tamás Szűts[1]
[1]University Of Veterinary Medicine Budapest, Budapest, Hungary

Mitochondrial markers are frequently used tools in molecular (meta)barcoding and species delimitation of animals. Despite the many advantages, their biggest limitation is that they can only reconstruct maternal lineages due to the uniparental inheritance. That is, in analyses and evaluations based solely on mitochondrial data, we only see one side of the coin, and important events, such as introgressions, hybridizations or male-mediated gene flow, remain invisible to us. This may be especially true in species where males and females contribute very differently to gene flow due to their different lifestyles. Even so, species-level differences in the maternal lineage caused by limited gene flow or local adaptations can represent actual units of the biodiversity (we propose the name „mitospecies"), which can be easily mapped and identified using the barcode approach. We illustrate this concept through a case study on Eresus ladybird spiders, a group with highly male-biased gene flow.

[A268] Population genomic insights into donor site suitability for eelgrass (Zostera marina) restoration

Lauren Sgambelluri[1], Clara Winguth[1], Prof. Dr. Thorsten Reusch[1]
[1]GEOMAR Helmholtz Centre for Ocean Research, Kiel, Germany

Biological conservation is increasingly relying on genomic information. Population genomic approaches utilizing genome-wide single nucleotide polymorphisms (SNPs) can significantly aid conservation efforts by identifying (1) well-connected populations with high gene flow acting as "hubs", (2) genetically diverse populations, and (3) populations harboring putatively adaptive loci that are beneficial for current or future climate change scenarios. Here, we apply such a conservation genomics framework to populations of seagrasses (marine angiosperms), which are among the most threatened ecosystems globally. Widespread declines have been documented since the late 19th century, prompting active conservation efforts. Our model species eelgrass (Zostera marina) is a foundation species forming intertidal and subtidal meadows across temperate regions of the Atlantic and Pacific oceans and the Mediterranean Sea, where losses of genetic diversity and connectivity can compromise both meadow resilience and restoration success. Identifying suitable donor sites is a critical component of effective seagrass restoration, yet genomic criteria for donor selection remain poorly explored. Here, we sampled and whole-genome sequenced 226 Z. marina individuals from 16 meadows across the western Baltic Sea. Using genome-wide SNP data, we characterize population structure, genetic diversity, and connectivity among meadows, and assess gene–environment associations to detect putatively adaptive variation. We highlight how a conservation genomic framework helps in evaluating donor site suitability and informs restoration strategies under current and future environmental change.

[A269] Skin-secreted antimicrobial peptides represent a fast-acting ancestral antipredator defense system in frogs

Lavrans Venken[1], Constantijn Raaymakers[1,2], Charlotte Martin[3], Tom Hellebuyck[2], Steven Ballet[3], Bram Vanschoenwinkel[1], Frank Pasmans[2], An Martel[2], Kim Roelants[1]
[1]Amphibian Evolution Lab, Biology Department, Vrije Universiteit Brussel, Pleinlaan 2, 1050 Elsene, Belgium, [2]Department of Pathology, Bacteriology, and Avian Diseases, Faculty of Veterinary Medicine, Ghent University, Salisburylaan 133, 9820 Merelbeke, Belgium, [3]Organic Chemistry Research Group, Chemistry Department, Vrije Universiteit Brussel, Pleinlaan 2, 1050 Elsene, Belgium

Poisons are a common defense weapon in the animal kingdom, and the faster they ward off a predator, the smaller the risk of injury and death. In anurans (frogs and toads), the skin secretion of many species constitutes a cocktail of toxic peptides that require oral absorption and subsequent spreading throughout the predator's body. This process takes at least a few minutes, a dangerously long time to avoid serious harm and ingestion by the predator. To investigate whether a peptide-based poison is capable of deterring predators through a faster mechanism, we conducted behavioral experiments in which Bombina orientalis frogs were exposed to snake predators. The frogs' poison caused nearly 60% of snakes to release their prey within 30 seconds, indicating the presence of a fast-acting component. Transcriptome and peptidome analyses show that antimicrobial peptides (AMPs), traditionally considered part of the immune system, are by far the highest expressed component of the B. orientalis poison. Experiments in which snakes were offered fish coated with a representative AMP revealed that this peptide alone significantly increased the incidence of prey release, while bradykinin, a common toxin in the poison of many frogs, did not. Comparative analyses of gene architecture and synteny imply that the AMPs of B. orientalis, together with those of other anuran lineages, represent a single evolutionary origin in a common ancestor of all living frogs. This early-evolved antipredator defense system may have acted as a key evolutionary innovation promoting the radiation of this ecologically successful vertebrate clade.

[A270] Benchmarking Genotype Likelihoods in Ancient DNA Studies

Weijia Xue[1], Zhou Zhou[2], Mingbo Yin[1], Lei Zhao[2]
[1]Fudan University, Shanghai, China, [2]East China Normal University, Shanghai, China

Ancient DNA (aDNA) is essential for reconstructing evolutionary history, yet its analysis is often challenged by post-mortem damage (PMD) and fragmentation. Genotype likelihood (GL) models provide critical frameworks for extracting information from degraded samples, but we noticed a lack of systematic benchmarking regarding estimation accuracy and computational efficiency. We evaluated seven state of-the-art GL models, including two different versions of the aDNA-optimized suite Atlas, across 0.5–20× depth and 10^6–10^8 bp lengths. Using simulations with realistic PMD, sequencing errors, and alignment biases, we assessed each model's accuracy in estimating genetic distance and the number of heterozygous sites, alongside computational resource usage. We found that Atlas generally provides superior genetic distance estimation across scenarios. Regarding the number of heterozygous sites, Atlas appears relatively reliable at sequencing depths ≥ 5×, yet it still encounters inaccuracies within the 0.5–5× coverage range. However, this relatively higher precision comes at the expense of significant computational costs; Atlas typically requires longer runtimes and more memory, scaling with genome size. Notably, we observed a "V-shaped" runtime efficiency that becomes more pronounced as genome lengths scale to 10^8 bp, better reflecting its performance in real-world genomic scenarios. While reducing sequencing depth initially speeds up processing by lowering I/O volume, this trend tends to reverse at extremely low depths, where the computational burden of PMD correction begins to outweigh the I/O savings. This study offers a practical guide for selecting paleogenomic tools by highlighting the trade-offs between analytical accuracy and resource requirements.

[A271] Beyond Recombination: Evaluating Mutation Clock approaches to Infer Archaic Admixture Times

Leonardo Nicola Martin Iasi[1], Dr Laurits Skov[2]
[1]Max-Planck-Institute for Evolutionary Anthropology, Leipzig, Germany, [2]Globe Institute, Section for Molecular Ecology and Evolution, University of Copenhagen, Copenhagen, Denmark
The sequencing of Neandertal and Denisovan genomes revealed that non-African modern humans carry introgressed DNA from these archaic populations. This ancestry can be found typically in short genomic segments.
The timing of these admixture events has primarily been estimated using a recombination clock, which infers the time since admixture from the length distribution of introgressed segments. This approach relies on fine-scale recombination maps to estimate recombination probabilities along the genome. However, recombination landscapes differ between human populations, particularly at fine scales, which can introduce bias in admixture dates.
Here we want to test if a mutation-clock could be used to date archaic admixture times. In principle, the ancestral recombination graphs (ARGs) could provide a direct framework for estimating admixture times by reconstructing local genealogies of introgressed segments and identifying post-admixture mutations. However, high-accuracy ARG inference remains computationally prohibitive at the sample sizes required for sufficient statistical power, and available archaic genomes represent diverged lineages rather than the true introgressing populations.
We therefore develop a scalable heuristic that approximates local genealogical structure using overlapping introgressed segments. Using coalescent simulations we show that with smaller sample sizes, both methods based on the true ARG and heuristic mutation-clock methods are underpowered, however when approaching large sample sizes true admixture times can be recovered. We replicate the Neandertal admixture date from recombination-clock based approaches using real data. Our results demonstrate that mutation-based dating of archaic introgression is feasible but only in large-scale genomic datasets.

[A272] Resistance selection allows identification of druggable targets in transmission stages of the human malaria parasite P. falciparum

Leonie Seefeldt[1], Olivia Carril[3], Jerôme Eberhardt[2], Armin Passecker[1], Dr. Leila Tamara Alexander[2], Prof. Dr. Markus Lill[2], Dr. Christof Grüring[1], Dr. Matthias Rottmann[1], Dr. Sebastian Baumgarten[3], Prof. Dr. Till Steffen Voss[1], Prof. Dr. Nicolas Michel Beat Brancucci[1]
[1]Swiss TPH, Basel, Switzerland, [2]Universität Basel, Basel, Switzerland, [3]Institute Pasteur, Paris, France

Most anti-malaria drugs fail at targeting sexual transmission stages, the gametocytes, of malaria parasites, endangering current and future efforts to eliminate or eradicate the disease. Developing such compounds is impeded by our limited understanding of male and female gametocytes. Compared to the symptoms-causing asexual parasites, mature gametocytes adopt a quiescent state that enables them to circulate in the human blood stream for several weeks prior to being ingested by an Anopheles mosquito vector.
Through the screening of diversity-oriented and mode-of-action libraries, we identified molecules that effectively eliminate mature gametocytes in the low nanomolar range. Focusing on three highly potent and chemically tractable compounds, we employed resistance selection, whole-genome sequencing (WGS) and drug-target interaction modeling to uncover their molecular targets. Parasite populations resistant to two of the drugs carried single nucleotide polymorphisms (SNPs) in genes encoding proteins previously reported as targets, supporting the validity of our approach. Notably, WGS of a population highly resistant to the third compound revealed a SNP in a gene encoding a putative ribosomal RNA (rRNA) methyltransferase. Nanopore sequencing combined with methylation profiling uncovered an altered rRNA methylation pattern in drug resistant parasites. We believe this is the first description of drug resistance caused by altered rRNA methyltransferase specificity.
Our findings reveal three distinct malaria transmission-blocking drugs and their targets, as well as a novel resistance mechanism and highlight the vulnerability of malaria transmission stages to proteostasis-targeting drugs.

[A273] Evolutionary transcriptomic analyses identify new potential gamete recognition proteins in lungless salamanders

Liam Johnson[1], Paul Nicolosi[1], Karen Kiemnec-Tyburczy[2], Damien Wilburn[1]
[1]Ohio State University, Columbus, United States, [2]California State Polytechnic University, Humboldt, Arcata, United States

Gamete recognition proteins often evolve at extraordinary rates, driven in part by differing fitness optima of sperm and egg. In animals, many sperm typically race to fertilize relatively few oocytes, and this asymmetry risks polyspermy. This sexual arms race has facilitated repeated evolutionary innovation of new gamete recognition proteins across vertebrates, yet little is known about the composition and molecular interactions of amphibian gametes. Lungless salamanders (family Plethodontidae) of the Appalachian Mountains in the eastern United States are an excellent system to study the evolution of fertilization proteins where many closely related species are found in sympatry with variable rates of hybridization. In this study, we prepared long-read testis and ovary transcriptomes using Oxford Nanopore Technologies-based sequencing for six plethodontid species (Plethodon shermani, P. metcalfi, Desmognathus ocoee, D. wrighti, D. amphileucus, Eurycea guttolineata) and one outgroup (Rhyacotriton variegatus). Candidate gamete recognition proteins were identified based on homology to known fertilization proteins or sequence features consistent with extracellular localization (signal peptide, transmembrane domain, GPI anchor). Paralogs of lungless salamander courtship pheromones were found in all testis transcriptomes, with sperm PRF being a plethodontid-specific protein while SPF was found in all species including Rhyacotriton. Ortholog groups for putative interacting from both sperm and egg proteins were constructed from the data, with evolutionary rates inferred using both branch- and site-based models. Potential molecular interactions were inferred through evolutionary rate covariation analysis and AlphaFold-based multimer modeling. This work establishes a foundation for understanding the molecular basis of species-specific fertilization in salamanders and other amphibians.

[A274] Recombination hotspot co-regulation by Prdm9 and X-linked regulatory microRNAs during meiosis

Lisa-Marie Hofacker[1], Jasmin Cichy, Dr. Elena Damm, Marvin Suhr, Rajalekshmi Narayana Sarma, Anne Anglet, Nicole Thomsen, Dr. Linda Odenthal-Hesse
[1]Max-Planck Institute for Evolutionary Biology, Plön, Germany

In most mammals, the genomic location of meiotic recombination hotspots is determined by the PR domain zinc finger protein PRDM9, whose highly versatile zinc finger array binds specific DNA motifs and thereby specifies the positions of double-stranded DNA breaks and recombination hotspots across the genome.
Using hybrid mouse sperm, we investigated how different Prdm9 alleles interact to shape recombination hotspot usage. By applying repulsion-phase allele-specific PCR, we estimated recombination frequencies and isolated thousands of crossover events at one symmetric hotspot, one asymmetric hotspot, and one hotspot activated by a humanised Prdm9 transgene. These analyses revealed epistatic interactions between Prdm9 heterozygosity, including the humanised Prdm9 allele, and the transgressive recombination modifier Hstx2, affecting recombination rates at specific hotspots.
High-resolution crossover breakpoint analyses further uncovered subtle and distinct effects of these regulatory factors on crossover distribution patterns, highlighting hotspot-specific differences in the fine-scale regulation of meiotic recombination.

[A275] Ancient and recent nuclear introgression between Fennoscandian voles

Lison Zunino[1], Silvia Marková[1], Hayley C. Lanier[1,2,3], Heikki Henttonen[4], Jeremy B. Searle[5], Petr Kotlík[1]
[1]Laboratory of Molecular Ecology, Institute of Animal Physiology and Genetics, Czech Academy of Sciences, Liběchov, Czech Republic, [2]Department of Biology, Program in Ecology & Evolutionary Biology, University of Oklahoma, Norman, USA, [3]Sam Noble Museum, University of Oklahoma, Norman, USA, [4]Natural Resources Institute Finland, Helsinki, Finland, [5]Department of Ecology and Evolutionary Biology, Cornell University, Ithaca, USA

Hybridization is a major evolutionary process in living organisms, occurring within species and less frequently between species. Introgression resulting from hybridization represents an important source of genetic variation in natural populations and can facilitate adaptation to new environments by increasing the frequency of advantageous alleles, although it may also have negative consequences such as outbreeding depression, leading to reduced fitness of hybrids relative to their parental species.

In Fennoscandia (Norway, Sweden, and Finland), the bank vole (Clethrionomys glareolus) and the northern red-backed vole (C. rutilus), which exhibit contrasting climatic distributions, form a contact zone where introgression of mitochondrial DNA (mtDNA) from C. rutilus into C. glareolus has been documented. In these species, previous studies suggest that mtDNA evolution may be linked to habitat and climate. In particular, the introgressed mtDNA has resulted in a distinct mitochondrial lineage in C. glareolus, which may have facilitated local adaptation to colder conditions. No evidence of nuclear introgression was detected in these populations, based on a limited set of markers.

Here, using newly generated whole-genome sequences from multiple populations of both species, we identify signatures of ancient introgression within the hybrid lineage of C. glareolus, as well as evidence of recent introgression in several C. rutilus individuals. These results raise the possibility that adaptive hybridization events may have contributed to the evolutionary history of both species and facilitated local adaptation, and may help to understand how they could response to global change.

[A276] On the Highroad to Reproductive Isolation: Cytonuclear Incompatibilities Between Selfing Populations of Arctic Cochlearia groenlandica (Brassicaceae)

Lovisa Gustafsson[1,2], Dr. Marie Kristine Branderud[2], Dr. Manuel Poretti[3], Dr. Michael Nowak[2], Dr. Desalegn Chala[2], Dr. Abel Gizaw[2], Prof. Anne Brysting[4], Prof. Christian Parisod[3], Prof. Levi Yant[5], Prof. Loren Rieseberg[6], Prof. Christian Brochmann[2], Ass. Prof. Siri Birkeland[2]
[1]The University Centre in Svalbard, Longyearbyen, Svalbard and Jan Mayen, [2]Natural History Museum, University of Oslo, Oslo, Norway, [3]Institute of Plant Sciences, University of Bern, Bern, Switzerland, [4]Centre for Ecological and Evolutionary Synthesis, Department of Biosciences, University of Oslo, Oslo, Norway, [5]Swedish University of Agricultural Sciences, Uppsala, Sweden, [6]Department of Botany, The University of British Columbia, Vancouver, Canada

Our previous research has demonstrated that hybrid sterility barriers have frequently evolved within diploid, circumpolar Arctic plant species. To characterize the genetic mechanisms underlying the rapid evolution of postzygotic reproductive isolation in Arctic plants, we conducted a genetic mapping study on Cochlearia groenlandica (Brassicaceae), using parental populations from coastal areas in Svalbard and Alaska. Through linkage mapping and quantitative trait loci (QTL) analysis, we identified genomic regions associated with gametic and seed production traits in two large reciprocal mapping populations. We detected multiple types of reproductive barriers. Notably, when the mother plant originated from Alaska, approximately 50% of F_2 seeds failed to germinate, whereas reciprocal F_2 seeds had a germination success exceeding 90%. In addition, pronounced trait divergence occurred only in the opposite cross direction: when the mother plant originated from Svalbard, roughly 25% of F_2 hybrids exhibited complete floral developmental failure and stunted growth, resulting in total reproductive breakdown. These traits were absent in the reciprocal population with an Alaskan maternal parent. This asymmetrical pattern points to strong cytonuclear incompatibilities, i.e. incompatibilities between nuclear and organelle genomes, as key drivers of reproductive isolation. Our findings underscore the critical role of cytonuclear interactions in establishing reproductive barriers and highlight their potential contribution to speciation processes in Arctic plant populations.

[A277] Characterizing the patterns of natural selection on fatty acid-related traits

Lu Wang[1], Kaixiong Ye[1]
[1]Department of Genetics, University of Georgia, Athens, United States

The capacity to process and utilize dietary fatty acids (FAs) has been critical to human survival, from early hunter-gatherer diets rich in marine and animal fats to agricultural diets dominated by plant-based foods. These transitions have driven genetic adaptations, most notably at the well-studied FADS (fatty acid desaturase) locus involved in the biosynthesis of long-chain polyunsaturated FAs. However, recent genome-wide association studies (GWAS) have identified many additional variants associated with FA traits whose selection patterns remain largely unexplored.
Here, we integrate GWAS results from the UK Biobank for 20 FA-related traits with multiple selection scans to investigate the evolutionary dynamics of FA metabolism. We assess enrichment of selection signals among GWAS variants using haplotype-based statistics (iHS and nSL) from the 1000 Genomes Project, as well as ancient DNA-informed selection scores. We further apply polygenic adaptation tests, including trait Singleton Density Score (tSDS) and Polygenic Adaptation Likelihood Method (PALM), to characterize polygenic adaptation across timescales.
We find that FA-related metabolic traits exhibit heterogeneous signatures of selection. Thirteen traits show evidence of selection in at least one test, and eleven traits display significant selection enrichment using aDNA-based method. Notably, enrichment of selection signals is found favoring a decreased circulating linoleic acid level from both nSL and ancient DNA-based analyses, whereas saturated FA levels demonstrate polygenic adaptation toward increased circulating levels in PALM. Together, these patterns may reflect shifting dietary practices and lifestyle changes throughout human history, with implications for population differences in metabolic health and nutritional requirements.

[A278] Population genetic statistics tools for the next generation of pool sequencing

Lucas Czech[1], Jeff Spence[2], Moises Exposito-Alonso[3]
[1]University of Copenhagen, , Denmark, [2]University of California San Francisco, , United States, [3]University of California Berkeley, , United States

Evolve-and-Resequence (E&R) experiments have become a popular approach to survey rapid evolution of populations over multiple generations. These experiments allow us to measure shifts in the allele frequencies of a population in response to changing environmental conditions, such as a changing climate.

Pool-sequencing of several individuals at once is a cost-effective and efficient tool to obtain reliable allele frequencies from a population of hundreds to hundreds of thousands of individuals, and is often used in E&R experiments. However, specialized tools to efficiently analyze these data that account for sampling noise were lacking. We developed two software tools to overcome bioinformatic and statistical challenges arising in this context.

First, we present grenepipe, a workflow from raw sequencing data of individuals or pooled populations to genotypes (variant calling) and population allele frequencies. The automated pipeline offers a selection of popular software tools in each step, and produces variant calls and frequency tables. While generally applicable to individual sample data, it offers specialized steps for pool-sequencing, and scales well to large contemporary datasets.

Second, to enable inferences of evolutionary signatures from frequency data, we created grenedalf. It computes unbiased statistics of Fst, Pi, Tajima's D with pool-sequencing data, far outperforming alternative tools, and fixing long-standing statistical issues in the existing tools. It offers a plethora of convenience options, filters, and file formats.

These tools were designed for, and applied in, the GrENE-net project, a large-scale Evolve-and-Resequence experiment in Arabidopsis thaliana, also presented at SMBE by plenary speaker Moises Exposito-Alonso.

[A279] Impact of compositional heterogeneity on divergence time estimation

Lucas Freitas[1], James Clark[2], Prof Philip Donoghue[1]
[1]Bristol Palaeobiology Group, School of Earth Sciences, University of Bristol, Bristol, United Kingdom, [2]Milner Centre for Evolution, Department of Life Sciences, University of Bath, Bath, United Kingdom

Evolutionary timescales are essential to much of evolutionary biology but their estimation is impacted by numerous issues, including compositional heterogeneity, which is widely appreciated as an important factor in phylogeny estimation. However, compositional heterogeneity impact on divergence time estimation remains unclear. Here we used a dataset containing 22 fossil calibrations, 122 orthologous and 29 species, mainly focused in Actinopterygii, to compare the divergence time estimation between site-homogeneous, partitioned and mixture substitution models (MMs). For species tree estimation we used both maximum likelihood and multi-species coalescent approaches, leading to two distinct species trees, which were next used for divergence time estimation. For divergence time estimation, besides the comparison between substitution models, we also compared distinct relaxed clock models and fossil calibration strategies. Strikingly, all substitution model comparisons in the same set (e.g. site-homogeneous, partitioned and MMs under auto-correlated clock model and fossil-rich calibrated tree) generated very similar divergence time estimates, showing that reliable estimates can be produced running the analyses with simple models up to 2,000x faster compared to most complex MMs. We also found that a fossil-rich calibrated tree produced similar divergence time estimates regardless of the clock model used; and decreased the uncertainty of estimates across all comparisons. These results are consistent with a previous study that shows that MMs may have a minor impact in divergence time estimates, but differ from other work, indicating that additional empirical and simulated studies are necessary to clarify the extent of impact of MMs across compositionally heterogeneous and homogeneous datasets.

[A280] Genomic Regulatory Networks Underlying the Evolution of Avian Beak Shape

Lucas Rocha Moreira[1], Dr. Nicolas Alexandre[1], Juan Zanin[1], Olivia Plateau[2], Jeffrey N. Stout[1], Irene M. Kaplow[3], Savannah J. Hoyt[4], Nicole M. Tillquist[4], Rachel J. O'Neill[4], Olivier Fedrigo[1], Richard A. Schneider[5], Anna L. Keyte[1]

[1]Colossal Biosciences, Dallas, United States, [2]University of Bern, Bern, Switzerland, [3]Carnegie Mellon University, Pittsburgh, United States, [4]University of Connecticut, Storrs, United States, [5]University of California at San Francisco, San Francisco, United States

The beaks of birds exhibit remarkable phenotypic diversity, yet the genetic and epigenetic mechanisms underlying this variation remain poorly understood. In this study, we generate and integrate comparative multiomic and morphological datasets to identify regulatory programs that shape the avian beak during development and evolution. We profile the frontonasal, maxillary, and mandibular primordia, which give rise to the beak, across six key developmental stages [Hamburger-Hamilton stages (HH) HH25 to HH40] in four model bird species (chicken, quail, duck, and pigeon), each having distinct beak morphology. Using a multiomics approach including bulk RNA-Seq, ATAC-Seq, Iso-Seq, and scRNA-Seq, we characterize both the transcriptional dynamics and the chromatin accessibility landscapes during the patterning and outgrowth of the beak primordia. In parallel, we assemble a broad comparative anatomy dataset by landmarking high-resolution µCT scans from our study species alongside ~70 additional bird species spanning major avian clades. Integrating available high-quality genomes with this morphological dataset, we identified conserved, convergent, and lineage-specific regulatory sequences associated with changes in beak size and shape. By integrating the regulatory landscape with geometric morphometrics, our work pinpoints candidate genomic features and gene networks that drive evolutionary change. Our results provide new insights into the developmental and regulatory foundations of craniofacial diversity and establish a comprehensive platform for linking genomic architecture to macroevolutionary morphological patterns in birds.

[A281] Warming and Hypoxia as Selective Forces Shaping Marine Bacterioplankton Functional Potential

Indranil Mukherjee[1], Luciana Santoferrara[1]
[1]Hofstra University, , United States

Bacterioplankton (Bacteria and Archaea in marine plankton) experience rapid community changes in eutrophic coastal systems undergoing seasonal warming and deoxygenation. To examine how these pressures shape lineage turnover and functional potential, we combined shotgun metagenomics and bacterioplankton taxonomic profiling during the summer progression in a harbor of the Long Island Sound estuary (New York, USA). Significant changes in bacterioplankton taxonomic composition between early and late summer were mainly driven by temperature and indicated strong seasonal lineage sorting. On the other hand, ongoing metagenomic analyses show that declining dissolved oxygen correlates with enrichment of gene pathways linked to nitrogen and sulfur redox processes, suggesting both a role of oxidative biogeochemical transformations on oxygen consumption and selection for reductive anaerobic or microaerophilic metabolic strategies during hypoxia. Phylogenetic analyses of selected functional genes (e.g., ammonia monooxygenase, nitrate and sulfate reductases) are being conducted under the hypothesis that hypoxia-associated gene variants cluster non-randomly, which would suggest lineage-specific selection for metabolic strategies rather than community-wide changes in functional potential. So far, our analyses indicate that summer warming primarily structures prokaryote populations through broad effects on lineage persistence and replacement, while deoxygenation exerts more targeted selective pressure on functional traits. Overall, our results support that short-term environmental gradients drive rapid reorganization of coastal bacterioplankton, with implications for the future trajectory of microbial functions under intensifying change in coastal ecosystems.

[A282] Plant-Microbial Interactions: Tracing a 2-Million-Year-Old Alliance with Ancient DNA

Luigimaria Borruso[1], Dr. Nikolay Oskolkov[2], Dr. Edoardo Pasolli[3], Dr Federica Villa[4], Dr Tanja Mimmo[1], Eran Elhaik[5], Dr. Luigimaria Borruso[1]
[1]Free University Of Bolzano, Bolzano, Italy, [2]Metabolic Research Group, Latvian Institute of Organic Synthesis, Riga, Latvia, [3]Department of Agricultural Sciences, University of Naples Federico II, Naples, Italy, [4]Department of Food, Environmental and Nutritional Sciences, University of Milan, Milano, Italy, Milano, Italy, [5]Department of Biology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Lund University, Lund, Sweden, Lund,
Plant–microbe interactions in the rhizosphere are central to nutrient cycling and ecosystem functioning. This dynamic soil–root interface is among the most biologically active environments on Earth, and its ecological relevance likely has deep evolutionary origins, as indicated by fossil evidence. However, fossils offer limited information about the taxonomy and functionality of ancient microbial communities, highlighting the need for complementary approaches. Sedimentary ancient DNA (sedaDNA) represents a promising yet underexplored tool to reconstruct past microbial communities and investigate ecological interactions among plants, animals, and microorganisms. Here, we examine ancient plant–microbe associations using 2-million-year-old sedaDNA from the Kap København Formation in Northern Greenland. Building on previous work that reconstructed an ecosystem analogous to a modern boreal forest, we specifically target microbial aDNA to characterise microbial communities, assess co-occurrence patterns with plant taxa, and identify functional traits linked to plant growth promotion. Our results show that this ancient boreal ecosystem hosted a microbial community broadly comparable to modern boreal soils, including several rhizosphere-associated taxa. Many bacteria consistently co-occurred with specific plant taxa, forming a conserved taxonomic core along with a conserved set of plant-growth-promoting traits (PGPT), reflecting those of modern rhizospheres. These findings highlight the potential of sedaDNA to reconstruct ancient ecosystems and infer long-term ecological interactions.

[A283] Phylogenetic correlation between the Type IV Secretion System and HIP1 suggest an adaptation for horizontal gene transfer in cyanobacteria

Luis Delaye[1]
[1]Cinvestav, Irapuato, Mexico

Most cyanobacterial genomes are rich in Highly Iterated Palindromic (HIP) sequences, such as the octamer GCGATCGC, also known as HIP1. Despite being described over 20 years ago, the function of this highly abundant sequence remains a mystery. HIP1 is recognized by two DNA methylases, DmtA and DmtC, and is characterized by its evolutionary conservation and an approximate periodic distribution within genomes. It is currently unknown whether the phylogenetic distribution of HIP1 and other HIPs correlates with the presence of specific genes. Here, we investigated whether there are genes whose phylogenetic distribution among cyanobacterial genomes correlates with these abundant palindromic sequences. Our analysis revealed a strong phylogenetic correlation between several proteins of the Type IV secretion system (T4SS) and the presence of HIP1, an association that persists when considering other HIPs. This finding is highly consistent with recent discoveries showing that HIP1 enhances DNA transformation in a methylation-dependent manner in two distinct cyanobacterial species. We therefore hypothesize that HIP1 function as a conserved adaptation for horizontal gene transfer (HGT) in cyanobacteria, potentially by serving as a DNA recognition sequence in this phylum.

[A284] Correcting for unequal error rates in D-statistics comparisons

Madina Japakhova[1], Janet Kelso[1], Dr. Kay Prüfer[1]
[1]Max Planck Institute For Evolutionary Anthropology, Leipzig, Germany
The D-statistic was developed to test for signals of introgression and has for instance been applied to Neandertals and modern humans (Green et al., 2010). The test counts BABA and ABBA site-patterns in comparisons of four groups, represented by one or several genomes, each. The test is based on the null assumption that neither the first nor the second group should show an excess of allele sharing with the third group. Often, however, analyses include ancient specimen of lower sequence coverage and high error in the sequence data, leading to false signals of introgression using the D-statistics. Here, we explore the possibility to correct D-statistics comparisons that involve low-quality data for one of the groups by leveraging increasingly distant outgroups. This correction is based on the idea that increasingly distant outgroups would increase ABBA sites whenever an error in the first group co-incides with true divergence in the outgroup. By extrapolating the point where the outgroup distance is equal to the distance to the third group, the method aims at equalizing the contribution of the error to ABBA and BABA sites. We explore the performance of the method using both real and simulted data.

[A285] Evolution of gene regulation in the primate endometrium at single-cell resolution

Maëlle Daunesse[1], Anthony Lepelettier[1], Axelle Brulport[1], Leo Zeitler[1], Eulalie Liorzou[1], Camille Berthelot[1]
[1]Institut Pasteur, Paris, France

Menstruation is a rare evolutionary innovation that has arisen independently in several mammalian lineages, yet the gene regulatory mechanisms underlying its emergence remain poorly understood. In primates, the inner lining of the uterus (the endometrium) undergoes cyclical remodelling under hormonal control. In menstruating species, withdrawal of progesterone triggers extensive tissue breakdown and repair, whereas closely related non-menstruating species do not display this response. How such contrasting physiological outcomes in the same tissue evolved, and how they are encoded in gene regulatory landscapes, remains an open evolutionary question.
Here, we use single-nucleus ATAC-seq (snATAC-seq) to compare chromatin accessibility in the endometrium of six primate species spanning menstruating (human, baboon, macaque) and non-menstruating (marmoset, saimiri, mouse lemur) lineages. For each species, we profile endometrial chromatin accessibility at the progesterone peak and following progesterone withdrawal, enabling direct comparison of regulatory responses to a shared hormonal cue. We perform cross-species integration of snATAC-seq data to identify homologous endometrial cell types and orthologous cis-regulatory elements despite extensive regulatory turnover.
This comparative framework is designed to detect regulatory elements that are conserved across primates, as well as elements that have been selectively lost or rewired in menstruating lineages. By focusing on cell-type-specific regulatory responses to progesterone withdrawal, this work aims to reveal how changes in cis-regulatory architecture contributed to the evolution of menstruation and to provide a general framework for studying regulatory innovation using comparative single-cell epigenomics.

[A286] Repeated evolution of pink flowers in alpine Arabidopsis arenosa is driven by PAP2 alleles with pleiotropic effects on UV protection and floral thermogenesis Abstract:

Magdalena Bohutínská[1]
[1]Charles University, , Czech Republic

A central debate in evolutionary biology is whether local adaptation is constrained by the "cost of complexity," favoring mutations with minimal pleiotropy, or driven by large-effect pleiotropic alleles that can overcome migration swamping. While the anthocyanin biosynthetic pathway is a classic textbook example of the "low-pleiotropy" scenario—where selection typically targets specialized, downstream genes—we show that this model does not hold in the context of rapid alpine adaptation. We combined genome-wide association studies (GWAS), petal transcriptomics, and field experiments to investigate the repeated evolution of pink-flowered populations in the mountain rockcress, Arabidopsis arenosa. We identified genetic variation in the Myb transcription factor PAP2 as the primary driver of this transition across multiple independent alpine colonizations. Contrary to the low-pleiotropy paradigm, our transcriptomic analysis reveals that the adaptive PAP2 allele upregulates the entire phenylpropanoid pathway. This coordinated regulation leads to a multifaceted chemical phenotype: the simultaneous increase of visible anthocyanins and "invisible" UV-absorbing flavonols. Through ecophysiological experiments and pollinator observations, we demonstrate that this shift is not driven by pollinator preference. Instead, we find that pink flowers accumulate solar heat significantly faster than ancestral white flowers, accelerating pollen tube growth in cold alpine environments. Our findings suggest that PAP2 acts as a "warming gene," where its high pleiotropy is not a constraint but a key adaptive feature that provides multifaceted advantages—thermal gain and UV protection—essential for survival in extreme environments. This study provides a compelling example of how the reuse of highly pleiotropic alleles facilitates rapid local adaptation despite the presence of gene flow.

[A287] Using ancient DNA to track the phenotypic history of dogs

Mahaut Goor[1], Dr. Lachie Scarsbrook[1,2], Alberto Carmagnini[1], Professor Laurent Frantz[1,2,3]
[1]Institute of Animal Systems Genomics, Faculty of Veterinary Medicine, Ludwig-Maximilians-Universität, Munich, Germany, [2]Palaeogenomics and Bio-Archaeology Research Network, School of Archaeology, University of Oxford, Oxford, United Kingdom, [3]School of Biological and Behavioural Sciences, Queen Mary University of London, London, United Kingdom

Dogs have been domesticated for over 20,000 years through a process that involves strong directional selection. Yet many of the highly derived phenotypes that define modern breeds are widely attributed to positive selection, as well as relaxation of selective constraints, acting primarily within the last ~150 years of intensive breed formation. In fact, although positive selection is presumed to be an important aspect of domestication, we do not yet understand the degree to which it shaped dog biology during the vast majority of their evolutionary history. The genetic architecture of many traits differentiating dog breeds is simple, often underlied by single SNPs or indels. Here, we take advantage of this simple genetic architecture to track the occurrence and frequency of variants underlying known traits in genome-wide data from more than 800 ancient canids spanning ~100,000 years and more than 2,000 modern individuals, to track allele-frequency changes and infer shifts in selective pressure. This approach enables us to quantify the extent to which both purifying and positive selection acted on variants underlying traits that define dog breeds today, across a wide range of archaeological contexts throughout Eurasia and North America, and to address fundamental questions about the nature of selection during the domestication history of dogs.

[A288] Do Living Fossils Evolve Slowly? A Comparative Analysis Across the Tree of Life

Manuela Cascini[1], Professor Simon Ho
[1]University Of Sydney, , Australia

The concept of living fossils has long fascinated scientists, as these taxa appear to persist with little apparent change over deep evolutionary time, seemingly contradicting expectations of continual evolutionary change. Living fossils are often described as species-poor lineages that retain ancestral traits and exhibit relatively slow rates of evolution. However, evidence supporting these claims has been mixed, and the extent to which living fossils are truly exceptional remains unclear.
Here, we investigate patterns of molecular and morphological evolutionary rates in taxa traditionally regarded as living fossils across the tree of life. To assess whether slow evolutionary rates are a distinctive feature of living fossils, we compare them with other species-poor clades that are not typically classified as living fossils. By integrating genomic and morphological data, this study aims to clarify whether living fossils represent a unique evolutionary phenomenon or fall within the broader spectrum of evolutionary rate variation.

[A289] Identifying the ‘roots’: development of markers for within and between species characterization of the Pinus genus.

Margarita Takou[1], Dr Hilke Schroeder[1], Dr Birgit Kersten[1]
[1]Thuenen Institute Of Forest Genetics, , Germany

Species identification is an important part of studying evolutionary ecology, quantifying biodiversity and supporting conservation efforts. Within species, the identification of regional origin contributes to population genetics questions, such as range expansions and hybridization zones. Traditionally, species identification was performed on the field based on phenotypic traits. However, there is an increasing demand for species identification in the wet lab or in silico. In this case, the use of genomic and, especially plastid and mitochondrial genome variation can be of great assistance. Species within the genus Pinus are, in addition to their economic importance, an integral part of the Eurasian ecosystems, which makes their correct identification a priority. Additionally, within species, identifying a population’s origin can stir the management of forest and its products. Here, we present markers for identification of species within the genus Pinus developed for those purposes. We used genome skimming with Illumina sequencing of 59 individuals from 14 species within the Pinus genus’ subgenera to in silico identify unique SNPs for each species. For the species P. pumila, P. cembra, P. sylvestris and P. sibirica we designed and tested in the lab markers based on restriction enzymes and sequencing diversity, while we tested in silico the identified markers across all 14 species using publicly available data. Finally, we repeated the process of identifying markers for specific P. sylvestris European populations. Therefore, we present a vital tool for evolutionary studies and quality control of forest products.

[A290] Resolving population structure and gene flow of two deep sea vestimentiferan worms from the Eastern Pacific

Marharyta Abashkina[1], Josefin Stiller[1]
[1]University of Copenhagen, , Denmark

Deep sea vestimentiferan tube worms represent a unique group of invertebrates adapted to extreme chemosynthetic environments such as methane seeps. The interest in their ecology grows beyond their physiological adaptations, questioning the gene flow and population connectivity, as seep habitats are often separated by tens to hundreds of kilometers across the ocean. Understanding the gene flow between such fragmented systems provides vital insights into the dispersal potential of deep sea fauna. Two species of tube worms: Lamellibrachia barhami and Escarpia spicata have been broadly sampled across the Eastern Pacific (from Alaska to Chile) and Gulf of Mexico within last two decades, yet their population structure remains poorly resolved, because traditional mitochondrial markers (COI and 16S) fail to discriminate through, a phenomenon not widely studied in literature. In this study we investigate the population structure of L.barhami and E.spicata using the whole genome resequencing data and attempt to understand the extent of mito-nuclear discordance in both species. Using genome-wide SNPs, we infer population structure through PCA, Admixture and Fst analyses, and test the stepping stone and isolation by distance dispersal hypothesis. Our results provide the first genome-scale resolution of population structure and gene flow in these related species, finding common patterns of dispersal in deep sea environments, as well as characterise an extent of mito-nuclear phylogenetic discordance and discuss potential reasons behind it.

[A291] Genomic history and ancestral origins of Afro-Mexican populations

Maria Fernanda Garcia Rodriguez[1,2], Dr Shreya Ramachandran[3], Ms María José Palma[4], Mr Juan Esteban Rodríguez-Rodríguez[5,8], Ms Isabella S. Gandara[6,7,14], Dr Marcus Feldman[8], Dr Carlos Bustamante[9], Dr. Andres Moreno-Estrada[10], Dr Cesar A. Fortes-Lima[11], Dr Marcela Sandoval[4], Julie Wesp[12], Dr Maria C. Ávila-Arcos[1]

[1]Laboratorio Internacional de Investigación sobre el Genoma Humano, UNAM, Juriquilla, México, [2]Escuela Nacional de Estudios Superiores Unidad Juriquilla, Universidad Nacional Autónoma de México, Juriquilla, México, [3]Department of Human Genetics University of Chicago Cummings Life Science Center, Chicago, United States, [4]Centro de Ciencias Genómicas, UNAM, Cuernavaca, México, [5]Centro de Investigación y de Estudios Avanzados del Instituto Politécnico Nacional, Alcaldía Gustavo A. Madero, México, [6]Harvard T.H. Chan School of Public Health, Harvard University, Boston Campus, Boston, United States, [7]Massachusetts Institute of Technology, Cambridge, United States, [8]Department of Biology, Stanford University, Stanford, United States, [9]Department of Biomedical Data Science, Stanford University, Stanford, United States, [10]Advanced Genomics Unit, Center for Research and Advanced Studies (Cinvestav), Mexico City, Mexico, [11]Johns Hopkins University Department of Genetic Medicine and McKusick-Nathans Institute, Baltimore, United States, [12]Department of Sociology & Anthropology, North Carolina State University (NCSU), Raleigh, United States, [13]University of Oklahoma, Norman, United States, [14]Stanford University School of Medicine, Stanford, United States

The Mexican population has three principal ancestries: Indigenous American, European, and African. Few studies have explored the genetic contributions of Afro-descendant communities in Mexico, although it holds significant importance in understanding the country's genetic diversity. Our research aims to characterize and analyze the genetic structure of Afro-Mexican populations, tracing their genetic origins at the subcontinental level. Our dataset comprises genotyping data for volunteer participants from Veracruz and the Costa Chica regions of Guerrero and Oaxaca, in addition to whole-genome sequencing data from selected individuals. We conduct genomic analyses including population structure, local ancestry inference and demographic model inference, as well as integrated ancient African individuals from the Colonial Period that can provide insights regarding the historical importance and origin of the Afro-Mexican populations. Results suggest two different ancestry sources within Africa and Indigenous ancestry correlated with geography. Ancient individuals cluster near the modern Afro-Mexican populations, suggesting genetic continuity. The research aims are to highlight the fundamental contribution of Mexico's Afro-descendant populations to our cultural and genetic diversity.

[A292] Impact of Invasion on Gonadal Microbiome Diversity in Red Palm Weevil (Rhynchophorus ferrugineus)

Mariam Alshamsi[1], Dr Mariana Lyra[1], Neelu Begum[1], Stéphane Boissinot[1]
[1]Center for Genomics and Systems Biology, New York University Abu Dhabi, Saadiyat Island,, United Arab Emirates

Heritable endosymbionts are key drivers of insect evolution, impacting host reproduction, physiology, and adaptation. Invasive species represent a unique evolutionary perspective to study the stability of host-microbe interactions undergoing rapid changes. We investigated the gonadal microbiome of the Red Palm Weevil (RPW; Rhynchophorus ferrugineus), a globally invasive agricultural pest, to examine how invasion and adaptation shape endosymbiont diversity.
We characterized the 16S rRNA gene diversity of female gonadal tissues across native (Southeast Asian) and invasive (Middle Eastern/Mediterranean) R. ferrugineus populations and compared them to species of the same genus (R. bilineatus and R. vulneratus).
Our analysis reveals a restructuring of the microbiome associated with invasion, where we observed distinct clustering (beta diversity) separating invasive populations from native ones. Notably, while endosymbionts dominate the microbiome in native populations, we observe additional microbial taxa in the gonads of invasive lineages. Furthermore, analysis between species showed that the R. ferrugineus microbiomes are distinct from those of the other species, R. vulneratus and R. bilineatus, which clustered together, suggesting a conserved microbiome composition that differs significantly from RPW. Thus, we suggest that the microbiome of invasive R. ferrugineus has diverged significantly from its evolutionary origins, creating a distinct profile compared to both native populations and congeneric species.

[A293] Genomic signatures of local adaptation across environmental gradients in the green anole

Dr Mariana Lyra[1], Theo Busschau[1], Stephane Boissinot[1]
[1]New York University Abu Dhabi, Abu Dhabi, United Arab Emirates

Environmental heterogeneity is a major driver of evolutionary processes, including population differentiation, local adaptation, and speciation. We use the green anole (Anolis carolinensis) as a model system to investigate how environmental gradients shape genomic variation and evolutionary diversification. This species spans a broad range of climatic conditions across the southeastern United States, from humid coastal environments to drier and colder inland regions, and exhibits pronounced phenotypic variation across habitats, making it well suited for studies of adaptation across heterogeneous landscapes. Using whole-genome resequencing data from four populations distributed across the species' range, we evaluate the relative roles of natural selection and demographic history in structuring genetic diversity. Population genomic analyses reveal clear genetic differentiation among populations. Genome-wide scans for selection identify candidate genes associated with metabolism, immune function, growth, and exploratory behavior that covary with environmental gradients, highlighting genomic regions potentially involved in adaptive divergence. Together, these results provide insight into the molecular and evolutionary mechanisms underlying population differentiation and local adaptation in a widespread vertebrate.

[A294] Functional and genomic features shape the recombination hotspot landscape of the house sparrow

Marie Raynaud[1], Alex Lawrence[2], Petronella Wessman[2], Dr Mark Ravinet[3], Henrik Jensen[4], Anna Runemark[2], Susan Johnston[1]

[1]Institute of Ecology and Evolution, School of Biological Sciences, University of Edinburgh, Edinburgh, United Kingdom, [2]Department of Biology, Lund University, Lund, Sweden, [3]Centre for Ecological and Evolutionary Synthesis, Department of Biosciences, University of Oslo, Oslo, Norway, [4]Department of Biology and the Gjærevoll Centre, NTNU, Trondheim, Norway

Meiotic recombination is a fundamental evolutionary force. Yet, recombination landscapes vary from species to chromosome level. One striking form of this variation is called heterochiasmy. It describes the difference between male and female recombination rates, but its drivers remain poorly understood. Birds offer a powerful system to investigate the evolution of heterochiasmy because their recombination landscapes are evolutionary stable. Using linkage disequilibrium from whole-genome polymorphism data, we characterized the fine-scale recombination landscape in three wild Eurasian house sparrow populations (Passer domesticus). Contrary to previous findings, we observed that recombination hotspots are not always conserved across chromosomes between the three populations, but they are overall enriched near promoters. To explore the functional basis of these patterns, we performed single-cell RNA- and ATAC-sequencing on female chick ovaries. We identified variation in gene expression and open chromatin regions across the genome in the germ cells, and found that both features showed strong positive associations with recombination hotspot location. With the forthcoming integration of pedigree data, we aim to disentangle between male and female recombination hotspots and to provide first insights into the genetic basis of sex differences in recombination rates and hotspot usage in the house sparrow.

[A295] GEMINI: Miniaturisation, vertebrates, and the genome

Mark D. Scherz[1]
[1]Natural History Museum Denmark, University Of Copenhagen, Copenhagen, Denmark
Miniaturisation (a reduction in body size that alters anatomy and ecology) has played a crucial role in vertebrate evolution, driven by genomic changes. The ERC-funded GEMINI project is investigating the relationship between organismal miniaturisation and genomes. We are generating and aligning high-quality genomes of representatives of the smallest members of each vertebrate order, and their non-miniaturised relatives, enabling us to investigate the potential causes and consequences of miniaturisation from a genomic perspective.

[A296] Niche conservatism during adaptive radiations

Martin Eriksson[1]
[1]Swedish Museum Of Natural History, Stockholm, Sweden

Understanding the formation of biodiversity patterns is a central challenge in evolutionary biology, made urgent by climate change and accelerating extinction rates. A key but poorly resolved pattern is niche conservatism—the tendency of closely related species to retain similar ecological traits. While niche conservatism encapsulates the joint action of ecological, evolutionary, and spatial processes, the mechanistic links among them remain unclear. Adaptive radiations involve a series of extraordinarily rapid speciation and ecological niche diversification events, and an open question is whether such rapid ecological expansions may deviate from typical patterns of niche conservatism and the underlying ecological, evolutionary and spatial processes. To answer these questions, we combine analysis of empirical data from several oceanic island adaptive radiations and perform spatially explicit individual-based simulations. We thus investigate how niche conservatism is influenced by spatial (dispersal along environmental gradients), evolutionary (genetic architecture), and ecological processes during adaptive radiations.

[A297] A new toolbox for simulations and inference in population genetics

Martin Petr[1]
[1]Globe Institute, University of Copenhagen, , Denmark

Computer simulations are of great importance across a wide range of studies, from benchmarking of new methods to inference of model parameters from empirical data. Unfortunately, developing simulations is often a time-consuming and error-prone task even for expert programmers, and the prospect of building custom-tailored simulation code is even more daunting for researchers not comfortable with technicalities of software development.

Here I present a tandem of software frameworks, designed specifically to address these challenges. First, an R package slendr (https://bodkan.net/slendr) unlocks the possibility to simulate data from complex demographic models in just a handful of lines of trivial R code, including standard Wright-Fisher models and models featuring spatial dynamics or natural selection. Second, an R package demografr (https://bodkan.net/demografr), a companion package of slendr, allows researchers to infer parameters of population genetic models using fully automated, paralellized routines for popular inference techniques such as Approximate Bayesian Computation. By utilizing slendr as framework for defining models for inference, demografr eliminates most of the complexities involved in programming traditional simulation-based inference workflows, allowing researchers to focus exclusively on a modeling problem at hand.

In this presentation, I will give an overview of both R packages and demonstrate their unique functionality. By showcasing a series of practical examples relevant for a broad range of modeling contexts, I will show that designing software which is easy to use for both novice researchers and experts alike is not only possible, but is of great importance in facilitating efficiency and reproducibility of computational research.

[A298] Evolutionary role of ZNF492 in human neural progenitor cells

Marula Mathew[1,2], Dr. rer. nat Michael Heide[3], Katja Nowick[1]
[1]Freie University Berlin, Berlin, Germany, [2]Einstein Center for Neurosciences , Berlin, Germany, [3]Deutsches Primaten Zentrum, Göttingen, Germany

Although the human genome shares significant similarity with other primates, certain genetic features are unique and contribute to the evolution of human traits. The human brain is characterised by an expanded neocortex arising from prolonged neurogenesis and an enhanced proliferative capacity of progenitor cells.

Transcription factors are particularly interesting in elucidating evolutionary novelty, as they regulate the expression of numerous target genes. Krüppel associated box domain containing zinc finger genes (KRAB-ZNFs) are the largest family of transcription factors in vertebrates and are amongst the most rapidly evolving genes. Here we investigated one such KRAB-ZNF, ZNF492 that is preferentially expressed in neural progenitor cells (NPCs) of the human cortex. ZNF492 experienced human specific changes in the KRAB-A and zinc finger domains involved in DNA binding and gene regulation, which may contribute to a human specific function of ZNF492. Overexpressing ZNF492 in human NPCs revealed an upregulation of genes involved in maintaining neural stemness. Our analysis of available CHIP-Exo data of ZNF492 on human embryonic kidney cells shows that 30% of target genes have developmental and neuronal functions. To determine its target genes in a neuronal context, we performed CUT&RUN sequencing on iPSC derived NPCs from human and chimpanzee cell lines. Among the validated targets using qPCR are PIK4A and TRIM9, which have a role in neurodevelopment and neurodegenerative disorders. Together, our results suggest an important role of ZNF492 in brain development and disease. The human specific sequence features of ZNF492 might thus have contributed to the evolution of the human brain.

[A299] Longitudinal multi-omics analysis of cynomolgus macaque lifespan uncovers age-related immune patterns

Hyeon-Mu Cho[1], Se-Hee Choe[1], Ja-Rang Lee[1], Hye-Ri Park[1,2], Min-Kyeong Ko[1,2], Yun-Jung Lee[1,2], Sang-Je Park[1], Young-Hyun Kim[1], Jaewon Huh[1]
[1]Korea Research Institute Of Bioscience And Biotechnology (KRIBB), Cheongju, South Korea, [2]Korea University of Science & Technology (UST), Daejoen, South Korea

While cross-sectional approaches have traditionally dominated aging research due to the challenges of long-term sampling, this study utilizes longitudinal analysis to provide a granular view of true intra-individual aging trajectories. To investigate senescence while controlling for environmental confounding factors, eight healthy, specific-pathogen-free (SPF) male cynomolgus macaques (Macaca fascicularis) were studied, divided into four age groups of two individuals each. We integrated multi-omics data—including Whole Genome Sequencing (WGS), RNA-seq, Whole Genome Bisulfite Sequencing (WGBS), and ATAC-seq—to profile age-related molecular phenomena such as mutation accumulation, gene expression shifts, and differential methylation. Our WGS analysis across three tissue types (buccal swab, blood, and sperm) revealed increased mutation accumulation and relatively slightly higher variant allele frequencies (VAF) in older individuals, particularly within buccal swab samples. Furthermore, our synthesized multi-layered datasets suggest a mechanism where increased chromatin accessibility in regulatory regions correlates with mRNA upregulation in older subjects; a subset of these genes also exhibited concurrent CG hypomethylation in the same regions. Consistent with our previous findings, upregulated genes were primarily enriched in inflammatory responses, whereas downregulated genes were associated with lymphocyte activation. As this is an ongoing project, we are currently conducting a comprehensive integrative analysis of these datasets

[A300] Effect of effective population size on the rate of Y chromosome degeneration

Mattéo Todaro[1], Aline Muyle[1]
[1]Institut des sciences de l'évolution, Montpellier, France
The evolution of sex chromosomes is characterized by recombination suppression, which triggers Y chromosome degeneration through the accumulation of deleterious mutations.

While effective population size (Ne) is a fundamental force shaping genomic architecture, its specific influence on the rate of Y degeneration remains unresolved. Theory suggests opposing forces: low Ne accelerates degeneration via genetic drift, whereas high Ne may paradoxically accelerate it through "genetic draft"—where linked deleterious alleles hitchhike with beneficial mutations, particularly in young evolutionary strata where adaptive potential is high. Recombination suppression prevents chromosomes from getting rid of deleterious mutations fixed by “genetic draft”.

To address this paradox, I will use a comparative framework of five closely related Silene species (section Melandrium). These species share a sex chromosome system (originating ~11 Mya) with characterized evolutionary strata but exhibit distinct effective population sizes. Using RNA-seq data and a reference genome (S. latifolia), I will quantify Y degeneration across species and strata. I will estimate selection efficacy and degeneration intensity using dn/ds ratios (mainly for old strata), and FastDFE to estimate the proportion of adaptive substitutions (alpha) reflecting current demographic dynamics, alongside assessments of Y-linked gene loss.

By correlating species-specific autosomal Ne with the proportion of Y-linked gene loss and selection efficacy metrics (dn/ds and alpha) estimated for each evolutionary stratum, this study aims to disentangle whether degeneration is primarily driven by drift or hitchhiking.

[A301] Locating genetic ancestors with ancestral recombination graphs

Matthew Osmond[1]
[1]University Of Toronto, Toronto, Canada

Spatial patterns of genetic relatedness reflect the past movements of genetic ancestors. Our ability to untangle this history has the potential to improve dramatically given that we can now infer the ultimate description of genetic relatedness, the ancestral re-combination graph. We have developed two methods to infer the location of genetic ancestors (and dispersal rates) from ancestral recombination graphs. We have applied one of these methods to Arabidopsis thaliana, humans, crows, and kelp and extended it to include non-contemporary (eg, ancient) samples. This poster will give an overarching view of the methods, the applications, and the extension to ancient DNA.

[A302] Genomic characterization of the island population of Heliconius cydno from Gorgona Island, Colombia

Mayelby Geraldine Rueda Muñoz[1], Alexandra Arias[2], Carolina Pardo-Diaz[2], José Cerca[1], Camilo Salazar[2]
[1]University Of Oslo, Oslo, Norway, [2]Universidad del Rosario, Bogotá, Colombia

Gene flow, demographic change, and geographic isolation interact in complex ways during population divergence. These processes can be difficult to detect and disentangle, especially in insular populations. However, such populations can serve as informative models, as founder events and reduced population size may both accentuate genetic differentiation and obscure historical signals of gene flow.

We used a population genomic approach to investigate the evolutionary history of an insular population of Heliconius cydno from Gorgona Island (Colombia). We analyzed whole-genome data from 53 individuals, including samples from the island population and three nearby continental populations that shared wing patterns with it.

Genome-wide analyses reveal that the Gorgona population is genetically differentiated, consistent with long-term isolation. However, we also find evidence for complex historical relationships. Demographic modelling suggests that past gene flow among the continental populations best explains the genomic and phenotypic patterns, supporting a hybrid origin for the Gorgona population.

Our results underscore the challenges of inferring gene flow in systems characterized by isolation and strong demographic shifts, and highlight the value of integrative analyses for understanding diversification in insular contexts.

[A303] La Quebrada Project: A community-engaged exploration of Afro-descendant heritage in Peru

Maria A Nieves Colón[1], Laura Pott[1], Jaime Zolik[1], Carrie Miller[1], Megan Dorame[1], Zachary Klennert[1], Claire Maass[2], José Luis Santa Cruz Alcalá Santa Cruz Alcalá[3]
[1]University of Minnesota Twin Cities, Minneapolis, United States, [2]Defense POW/MIA Accounting Agency, Joint Base Pearl Harbor Hickam, United States, [3]Apacheta Consultores, San Martin de Porres, Peru

The Trans-Atlantic Slave Trade was the largest forced displacement in history. Although most enslaved peoples were transported to colonial Latin America, African diasporic histories in this region are severely understudied. This project addresses this gap by investigating the genetic diversity of enslaved individuals buried in the historic cemetery of Hacienda La Quebrada; an 18-19th century plantation in the San Luis District of Peru. Our work is conducted in partnership with the modern Afro-Peruvian community of San Luis who have engaged in archaeological and cultural heritage research at this site since 2016. Here we discuss how ongoing engagement with, and responsiveness to, community interests has informed current research questions, methodological approaches, and future project directions. Our team extracted ancient DNA from N=61 individuals from the historic cemetery. Using targeted enrichment and deep sequencing, we recovered mitochondrial genomes at >5X mean variant depth for 95% of the historic individuals (n=58). Preliminary analyses found evidence of diverse maternal ancestries: 77.6% carried African L0-L3 haplogroups, 20.7% carried Indigenous American haplogroups (A/B/C/D), and 1.7% carried European haplogroups (H). Ongoing conversations with community partners revealed that mitochondrial DNA results were highly valued by residents interested in reclaiming the histories of their female ancestors. In response, our ongoing work aims to reconstruct the maternal ancestries and kinship patterns of the cemetery individuals in greater detail. This project demonstrates how community-engaged approaches can advance our understanding of complex demographic histories while also advancing equitable genomics research in the Americas.

[A304] From population genomics to health equity: evolutionary insights from Indigenous populations of Taiwan

Wen-Ya Ko[1,2], Jian-Lian Chen[1,6], Mei-Ling Kang[1], M.D. Valis Tanapima[2,3], Ph.D. Yuan-Feng Lin[4], Ph.D. Chao-Kuang Lin[2,5]

[1]Department of Life Sciences and Institute of Genome Sciences, National Yang Ming Chiao Tung University (NYCU), Taipei, Taiwan, [2]Research Center for Health and Technology Development of Indigenous People of Taiwan, NYCU, Taipei, Taiwan, [3]The Emergency Medicine Department at Cheng Hsin General Hospital, Taipei, Taiwan, [4]Graduate Institute of Clinical Medicine, Taipei Medical University, Taipei, Taiwan, [5]Institute of Science, Technology, and Society, NYCU, Taipei, Taiwan, [6]School of Biological Sciences, Georgia Institute of Technology, Atlanta, USA

Achieving health equity in precision medicine requires evolutionary and population genomic perspectives, yet Indigenous populations remain profoundly underrepresented in human genetic research, leading to inaccurate genomic risk prediction and unequal clinical benefit. Taiwan hosts multiple Austronesian-speaking Indigenous populations central to the Austronesian expansion, but their demographic and evolutionary histories remain insufficiently characterized, constraining ancestry-aware disease risk assessment and equitable clinical implementation. Here, we present an integrative population genomic analysis of Indigenous populations of Taiwan (including Amis, Atayal, Bunun, Paiwan, Puyuma, Rukai, Truku, and Siraya) using genome-wide SNP data, local ancestry inference, and haplotype-based scans for recent positive selection. Analyses of more than 1,300 individuals reveal pronounced genetic differentiation among Indigenous groups, exceeding divergence between Han Taiwanese and other East Asian populations. These results demonstrate deep evolutionary histories among Indigenous Taiwanese peoples and show treating them as a single genetic unit risks inaccurate disease-risk inference and unequal clinical benefit. Focusing on the Pingpu Siraya population, local ancestry tract length distributions support a two-pulse admixture model with Sino-Tibetan–speaking populations, with the earliest event dating to 250 years ago and closely matching historical migration records. Genome-wide iHS scans identify population-specific signals of recent positive selection beyond shared HLA loci, including GYPA/GYPB in Amis and SMYD3 in Atayal, genes involved in erythrocyte biology, host–pathogen interactions, chromatin regulation, and cellular stress responses. Together, our findings demonstrate that incorporating fine-scale population structure, admixture history, and adaptive evolution is essential to prevent further inequities as precision medicine expand.

[A305] Genomic architecture and reproductive trait divergence between Drosophila suzukii and its sibling species

Rahayu Pertiwi[1], Tatsuyuki Kumagai[1], Kyoka Sakai[1], Renta Yamamoto[1], Masatora Tatsuzaki[1], Koutarou Takahashi[1], Dr. Airi Sato[1,2], Dr. Moe Onuma[1], Rie Okada[1], Dr. Yoshitaka Kamimura[3], Dr. Kentaro Tanaka[1], Aya Takahashi[1,4]

[1]Department of Biological Sciences, Tokyo Metropolitan University, Hachioji, Japan, [2]Evolutionary Genetics and Genomics Laboratory, The Rockefeller University, New York, USA, [3]Department of Biology, Keio University, Yokohama, Japan, [4]Research Center for Genomics and Bioinformatics, Tokyo Metropolitan University, Hachioji, Japan

Drosophila suzukii females exhibit a unique oviposition strategy of laying eggs into ripening fruits, in contrast to most Drosophila species that prefer decaying fruits. This niche shift is associated with evolutionary changes in the ovipositor morphology, alterations in the spatial configuration of genital coupling during copulation, and rapid diversification of male genital morphology. To investigate the genetic basis of these interspecific differences, we performed a quantitative trait locus (QTL) analysis using a closely related species, D. subpulchrella. The results showed that loci responsible for the interspecific differences in several reproductive traits, potentially driven by the novel oviposition strategy, mapped to the regions with extensive structural divergence between the two species. We further characterized interspecific differences in genomic structure and transcriptomic divergence within these regions. In addition, analyses of reproductive traits in semi-introgression lines carrying D. subpulchrella chromosomal segments in a D. suzukii genetic background demonstrated that these regions harbor loci contributing to interspecific differences in male genital morphology. Together, our results indicate rapid coevolutionary changes in reproductive traits associated with an ecological niche shift and suggest a link between this phenotypic divergence and underlying genomic architecture of these species.

[A306] Higher frequency of prokaryotic low complexity regions in core and orthologous genes

Fabia Ursula Battistuzzi[1], Vineet Saravanan[2], Alexander Kravetz[3]
[1]Oakland University, Rochester, United States, [2]Cranbrook Schools, Bloomfield Hills, United States, [3]University of Michigan, Ann Arbor, United States

Prokaryotic genome evolution is shaped by mutation, gene duplication, and horizontal gene transfer, yet the interaction of these mechanisms, particularly in relation to low complexity regions (LCRs), remains poorly understood. LCRs are known to be mutation-prone and have been proposed to promote genetic innovation. However, the interaction between LCR-mediated and paralogy-mediated genetic innovation is still unclear. To clarify the interplay between these two evolutionary forces, we analyzed the distribution of LCRs in protein-coding genes from three closely related enterobacteria (Escherichia coli, Salmonella enterica, and Klebsiella pneumoniae) at both species and population levels. Using pangenomic and orthology-based approaches, we categorized genes by duplication history and conservation status and assessed LCR frequencies across these groups. We found that LCRs were consistently enriched in core and orthologous genes rather than in accessory or paralogous ones. This pattern was stable across evolutionary timescales and particularly pronounced in genes involved in cell cycle control and defense. These results suggest that, contrary to prior assumptions, LCRs may serve conserved functional roles rather than acting primarily as agents of evolutionary plasticity even at population-level timescales.

[A307] Adaptation in unstable environments through genome reduction – Extending the May–Wigner theory to genomes

Suhua Shi[1]
[1]School of Life Sciences, Sun Yat-sen University, Guangzhou, China

How mangrove plants adapt to highly fluctuating intertidal environments is a central question in mangrove evolution. Contrary to the expectation that adaptation to multiple stresses requires increased genomic complexity, a comparative survey across major mangrove clades revealed a convergent reduction in both gene number and intergenic regions. Focusing on the largest clade, Rhizophoreae, we investigated the adaptive significance of these genomic reductions. Only a small fraction of gene loss was associated with physiological innovation, whereas most reductions affected genes involved in environmental interactions, likely facilitating adaptation to constant tidal fluctuations. Gene loss accumulated after the colonization of intertidal habitats and largely avoided genes with core woody plant functions. Reduction of intergenic regions was accompanied by decreased regulatory elements and chromatin accessibility. Together, genic and intergenic reductions reshaped gene regulatory networks (GRNs), producing networks that are both smaller and simpler. Here, we extend Robert May's theory (often referred to as the May–Wigner theory or MWT) on food web stability to genomes. MWT predicts that reducing system complexity enhances stability under persistent perturbations. Extending MWT to GRNs, computer simulations showed that genome reduction increases network stability. Consistently, transcriptomic analyses revealed that mangroves evolved more stable gene expression under fluctuating salinity through a shift from stress-responsive to constitutive regulation. Overall, our results demonstrate that genome reduction plays a key adaptive role in mangroves, contributing both to specific phenotypic innovations and to the stabilization of GRNs in unstable environments, as predicted by MWT.

[A308] Hybridization at the range margin: persistence, structure, and introgression

Dr. Toni Gossmann[1], Dr. Sahar Javaheri Tehrani[1], Dr Niloofar Alaei Kakhki[1], Yu-Chi Chen[1], Mansour Aliabadian[1], Martin Päckert[1]
[1]Tu Dortmund University, ,

Hybridization is often viewed as an unstable evolutionary state, leading either to lineage fusion or to narrow hybrid zones maintained by selection against hybrids. Yet at species' range margins, where ecological transitions and demographic constraints intersect, hybridization may generate more complex outcomes. Here, we investigate a songbird lineage at an evolutionary crossroads using whole-genome resequencing of a contact-zone population in the great tit complex (Parus).

Mitochondrial genomes reveal a distinct and coherent mitochondrial lineage associated with intermedius that is not replaced by Parus major haplotypes, alongside occasional mitochondrial introgression from the Central Asian bokharensis lineage. In contrast, nuclear genome-wide analyses place intermedius closest to P. major while clearly differentiating it from both parental lineages and revealing internal substructure associated with variable proximity to bokharensis. Together, these patterns indicate that intermedius represents a distinct evolutionary lineage rather than a transient hybrid population, persisting despite ongoing and spatially structured gene flow.

Genome-wide tests further reveal heterogeneous introgression, including localized allele sharing overlapping a major differentiation outlier containing the pigmentation-related gene, highlighting a candidate case of adaptive introgression. Our results illustrate how hybridization at an evolutionary crossroads can generate lineage persistence, internal structure, and localized introgression, rather than inevitable homogenization, offering a nuanced view of evolutionary outcomes at range margins.

[A309] Spatial Patterns of Heterogeneity in Solid Tumors and Their Evolutionary Origins

Weiwei Zhai[1]
[1]Institute of Zoology, Chinese Academy of Sciences, Beijing, China

Solid tumors exhibit pronounced spatial intra-tumor heterogeneity (sITH), but existing models provide no clear null expectation for spatial heterogeneity in tumor evolution. By sequencing and assembling one of the largest multi-region spatial cohorts to date (MRSC, 1,169 regions from 165 patients), we systematically investigated the spatial patterns of heterogeneity across solid tumors. We found that genetically similar cell populations often cluster into spatially contiguous “blocks”, which we term block-shaped heterogeneity (BSH). Using spatial agent-based models, we investigate how different evolutionary forces including mutation, selection, migration and tissue architecture can influence the spatial distribution of heterogeneity.
In the end, we will explore how BSH might affect the test of selection and contradicts predictions from boundary-growth models. Together, we systematically explored spatial heterogeneity in solid tumors, offering novel insights into tumor evolution, biopsy accuracy and therapeutic resistance.

[A310] Mid-trajectory transcriptome convergence and ancient gene use during plant cell differentiation

Wenfeng Qian[1]
[1]Institute of Genetics and Developmental Biology, Chinese Academy of Sciences, Beijing, China
Developmental hourglass has been widely reported in animals at both morphological and transcriptome levels. While the morphological similarity was not observed in plants the hourglass at the transcriptome level was still observed, leaving a question of why. We hypothesize that the hourglass may be intrinsic as it stays in cell differentiation trajectory. We tested this idea using single-cell transcriptome data in rice seedlings. We found that mid trajectory tends to express genes originated deep in history (phylostratigraphy “old” genes) and housekeeping genes which broadly expressed in various cell types. To further explore this idea, we generated single-cell transcriptomes from five additional cereal seedlings and again observed a mid-trajectory state with maximal cross-species similarity. These findings highlight a cell-differentiation hourglass as an organizing principle of plant cell-type transition from proliferation to differentiation.

[A311] Cross-species comparisons of the chordate digestive tract reveals conservation of pancreatic endocrine cells

Yichen (Serena) Dai[1]
[1]School of Life Sciences, Fudan University, Shanghai, China

Despite their pivotal role, the evolutionary origins of vertebrate digestive systems and key endocrine cell types remain enigmatic. We explored the cellular characteristics of the digestive tract of multiple chordate species using bulk tissue companioned with single-cell RNA sequencing. Our findings reveal segmentation and a rich diversity of cell clusters, and we highlight the presence of epithelial-like, ciliated cells in the amphioxus midgut and describe three types of endocrine-like cells that secrete insulin-like, glucagon-like, and somatostatin-like peptides. Furthermore, Pdx, Ilp1, Ilp2, and Ilpr knockout amphioxus lines revealed that, in amphioxus, Pdx does not influence Ilp expression. We also unravel similarity between different endocrine or endocrine-pathway genes in chordate species in terms of predicted structure, effects on body growth and amino acid metabolism. These findings indicate that the evolutionary alterations involving the regulatory influence of Pdx over insulin gene expression could have been instrumental in the development of the vertebrate digestive system. Furthermore, cross-species comparison between gut cell types suggest early emergence of endocrine cell types secreting glucagon-like and somatostatin-like peptides in the gut of the last chordate ancestor.

[A312] Identification of regulatory elements influencing eye evolution in cave fish

Marcos da Silva[1], Ashlynn Harlow[1], Devin Shennard[1], Alexander Seaver[1], Johanna Kowalko[1], Wynn Meyer[1]
[1]Lehigh University, Bethlehem, United States
Natural variation in regulatory regions is an important driver of phenotype evolution, however establishing genotype-phenotype relationships in such regions poses a challenge due to, for instance, how difficult they are to locate. Here, we took into account the repeated loss of eyes found across cave fishes in order to identify gene regulatory variants that may affect phenotypic evolution, such as eye degeneration. We identified 29 candidate genes that are under positive selection in cavefish and we identified candidate non-coding regions within 1 Mb around of the genes that are highly conserved in an alignment of 65 bony fish. Comparing the sequences of these variants with homologous regions in the cave fish Astyanax mexicanus reference genome, we identified 19 putative regulatory regions comprising at least 20 base pairs and at least one sequence difference between surface and cave reference genomes. We are now in the process of validating these sequence differences in putative regulatory regions by PCR amplification and sequencing in laboratory populations of surface and cave fish. Immediate future work will involve employing reporter gene assays and CRISPR-Cas9 and determine where these regions drive gene expression by cloning the surface versions of these regions into a construct with a reporter gene encoding green fluorescent protein (GFP), enabling visualization of GFP expression in cells where the regulatory region drives expression. We will then inject these constructs into zebrafish embryos harbouring pIGLET landing sites, allowing for reproducible integration into a specific genomic region.

[A313] Constructive neutral evolution explains the emergence of specialised ribosomes in diverse eukaryotes.

Alan Beavan[1], Professor Mary O'Connell[1]
[1]University of Manchester, Manchester, United Kingdom

Throughout eukaryotic evolution, the structure of the ribosome been highly conserved, with a core of 79 protein gene families. However, in many eukaryotes, paralogs of these persist. In the case of specialised ribosomes, the identity of the paralog incorporated into the synthesised ribosomes affects the proteins produced. Specialised ribosomes have been documented across diverse major groups of eukaryotes including fungi, plants, and animals, providing an opportunity to study the convergent evolution of a complex trait. Using phylogenetic gene tree-species tree reconciliation, we ask what drives ribosome specialisation? We observe dynamic generation of ribosomal protein paralogs across eukaryotes, finding that experimentally validated cases of specialisation tend to involve independent duplication of the same genes in different lineages. We outline the ways in which the evolution of specialisation can be generalised. We show that pathways to specialisation are not associated with: (i) any particular part of the 3D ribosome complex, or (ii) shifts in selective pressure on paralogs. We conclude that the emergence and retention of ribosomal protein paralogs is explained by a constructive neutral evolution model rather than adaptive selection, and conclude that specialisation has emerged through drift in lineages of complex eukaryotes with small effective population sizes.

[A314] Epigenetics Meets Osteology: DNA Methylation and Cranial Bone Modeling in an Ontogenetic Sample of Baboons

Alexandra Schuh[1], Elena Essel[2], Joerg Beckmann[3], Lorenzo von Fersen[3], Dr. Genevieve Housman[4], Dr Pr Philipp Gunz[1]
[1]Department of Human Origins, Max Planck for Evolutionary Anthropology, Leipzig, Germany, [2]Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany, [3]Tiergarten der Stadt Nürnberg, Nuremberg, Germany, [4]Department of Primate Behavior and Evolution, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

Cranial shape is highly variable within primates. This variation is widely attributed to differences in gene regulation during ontogeny, underlying bone growth and modeling. DNA methylation provides a proxy for regulatory variation, but methylation landscapes in bone, and how they relate to craniofacial modeling, are poorly documented. We integrate whole-genome bisulfite sequencing with microscopic evidence of bone modeling to link gene regulatory variation to the cellular processes shaping the cranium. In eight Guinea baboon skulls housed at the Natural History Museum of Berlin we quantified osteoblast and osteoclast activity through surface histology, generating detailed maps of facial bone deposition and resorption. In parallel, we generated methylomes from seven fresh-frozen Guinea baboon heads (ages 2 months–15.7 years) from the Nuremberg Zoo, contrasting bone samples from multiple cranial regions (facial and neurocranial), with dentine and muscle. Bone cores were cryo-milled to powder, and DNA was extracted following an ancient DNA protocol.
We document region-specific and individual-level methylation variation across the cranium and relate these patterns to variation in facial bone modeling. Our data reveal spatial variation in gene regulation across different regions of the skull within a single species, filling an important gap in our understanding of how regulatory differences translate into craniofacial morphological diversity. By linking DNA methylation patterns to bone modeling, this study represents an essential first step toward a mechanistic framework for studying craniofacial variation. It lays the groundwork for future integrative approaches aimed at reconstructing the developmental and evolutionary processes underlying craniofacial diversity across extant and extinct primates.

[A315] Leveraging the DNA methylation atlas of purified human cell types to detect putative metastable epialleles in humans

Alice Balard[1], Matt Silver[2], Garrett Hellenthal[1]
[1]University College London (UCL), London, United Kingdom, [2]London School of Hygiene & Tropical Medicine (LSHTM), London, UK

DNA methylation (DNAm) in the mammalian genome is typically highly variable across tissues. However, research into isogenic mice has reported “metastable epialleles” (MEs) -- CpGs that vary between individuals but show high tissue-concordance reflecting DNAm likely established in the early embryo which can be sensitive to environment and lead to lifelong health outcomes.

Identifying human MEs is difficult, requiring samples from all three germ layers per individual. A recent approach demonstrated the potential in leveraging large-scale methylation datasets covering different tissues to identify putative human MEs (Derakhshan 2022). They identified hypervariable CpGs (hvCpGs) that exhibit high interindividual DNAm variation across multiple tissues, with evidence of strong cross-tissue correlation within individuals, characteristic of MEs.

Here we introduce a Bayesian method that increases power to identify hvCpGs, avoiding heuristic thresholds used by Derakhshan and providing a posterior probability of hypervariability per CpG. It corrects for dataset-specific DNAm patterns and accommodates the frequent small sample sizes of whole-genome bisulfite sequencing (WGBS) studies by borrowing strength via hierarchical modelling.

We apply our method to WGBS of 46 purified primary cell types from the Human Cell Atlas, most sequenced in only three individuals (Loyfer 2023). Our approach identifies different classes of hvCpGs, including those highly variable across all three germ layers indicative of putative MEs; and others that are variable mainly in immune-related cell types and enriched for lineage-specific pathways, suggesting later acquisition of variability. Our results expand the catalogue of putative human MEs and provide a scalable framework for mapping early-development epigenetic variation genome-wide.

[A316] Dissecting the molecular mechanisms and evolutionary dynamics of gene regulatory silencers

Allison Wang[1], Jessica Day[2], Dr. Emily Hodges[2,3,4], Dr. John Capra[5,6]
[1]Pharmaceutical Sciences and Pharmacogenomics, University Of California, San Francisco, San Francisco, United States of America, [2]Department of Biochemistry, Vanderbilt University School of Medicine, Nashville, United States of America, [3]Vanderbilt Genetics Institute, Vanderbilt University School of Medicine, Nashville, United States of America, [4]Vanderbilt Ingram Cancer Center, Nashville, United States of America, [5]Bakar Computational Health Sciences Institute, University of California, San Francisco, San Francisco, United States of America, [6]Department of Biotechnology and Therapeutic Sciences, University of California, San Francisco, San Francisco, United States of America

Changes in gene expression contribute to phenotypic differences between individuals and species. Enhancers, non-coding cis regulatory elements (CREs) increasing transcription, are essential for specifying gene regulatory programs, and their role in evolutionary divergence has been studied extensively. Recently, large-scale functional studies have revealed many CREs decrease transcription. These “silencer” elements are common; however, current studies largely ignore their role, since few assays are designed to investigate their activity. Thus, our understanding of their contributions to gene regulatory evolution is limited.

To address this gap, we carried out multiple massively parallel reporter assays (MPRAs) to explore silencer regulatory effects on transcriptional activity and to quantify their divergence between species. We applied ATAC-STARR-seq, an assay testing the transcriptional activity of open chromatin regions, to lymphoblastoid cell lines (LCLs) from human and rhesus macaques. This revealed thousands of silencers active in each species and that silencers have even more rapid divergence in activity between species than enhancers.

To quantify the mechanisms underlying these evolutionary changes, we leveraged the ability of ATAC-STARR-seq to separate the DNA library tested for activity from the cellular context in which the library was constructed. Thus, we were able to test the activity of human silencers in macaque cells and vice versa. This enabled us to dissect activity divergence into its cis and trans components. Trans effects make a substantial contribution to silencer divergence, as seen for enhancers. Understanding silencer regulatory divergence will provide a greater understanding of the gene regulatory landscape’s contribution to both evolution and disease.

[A317] Genome-wide in-silico analysis of conserved miRNAs in aquatic and terrestrial mammals

Beatriz Daros[1], Bastian Fromm[2], Alexandre Paschoal[3], Ana Luiza Lein-Borba[1], Agnello Picorelli[1], Mariana Nery[1]
[1]University of Campinas, Campinas, Brazil, [2]The Arctic University of Norway, Tromsø, Norway, [3]The Rosalind Franklin Institute, Harwell, England

Aquatic mammals evolved adaptations that enable them to thrive in aquatic environments; the molecular mechanisms underlying these transitions remain incompletely understood. From a genomic perspective, most studies have focused on protein-coding regions, leaving gaps in our understanding of non-coding elements. microRNAs (miRNAs), a class of small non-coding RNAs, play key roles in post-transcriptional regulation and have been implicated in adaptive evolution. Here, we characterised the miRNA repertoire of aquatic mammals and phylogenetically related taxa to investigate patterns of conservation, loss, and sequence divergence. We employed MirMachine to annotate conserved miRNAs, and BLAST to assess miRNAs previously reported as exclusive to Bos taurus, Canis lupus, or Equus caballus. For miRNAs classified as absent in aquatic mammals by MirMachine, we conducted analyses that integrated Bowtie-based read-mapping, MUSCLE alignment, and RNAfold-based secondary structure prediction. Across 56 mammalian species, we identified 189 conserved miRNA families and examined loss patterns in 16 of those. Integrated genome and smallRNA-seq analyses demonstrated that only two miRNAs were truly absent in aquatic mammals. The remaining 14 families, initially inferred as lost, were supported by expression data and sequence conservation, despite exhibiting sequence variation. Notably, we identified mature-miRNA variants with fixed nucleotide substitutions in cetaceans for Mir-1842, Mir-330, Mir-339, and Mir-744. Overall, our findings indicate that aquatic mammals possess similar miRNA repertoires to closely related terrestrial species. Rather than miRNA gene loss events, the aquatic transition appears to have driven regulatory diversification through miRNA sequence plasticity. These results highlight the potential role of non-coding regulatory evolution during environmental transitions.

[A318] Evolution of RNA binding in the fungal gene regulatory protein Ssd1

Dr. Benjamin Kleinerman[1], Dr. Benjamin Kleinerman[1], Dr. Benjamin Kleinerman[1]
[1]University Of Edinburgh, Edinburgh, United Kingdom

Proteins gain and lose functions over time, as selective pressures change. Dissecting functional changes in individual protein families reveals molecular evolutionary mechanisms, including in gene regulatory proteins. Ssd1 is a fungal protein that regulates growth and cell wall integrity through RNA-binding, and a "pseudoenzyme", related to the Dis3L2 family of RNA exonucleases. Based on sequence signatures in extant homologs, we hypothesise that first a Dis3L2-like nuclease gained a novel RNA-binding function, resulting in a bifunctional protein. Then, a few bifunctional proteins lost their nuclease function, resulting in the RNA-binding protein Ssd1. We have characterized the Saccharomyces cerevisiae Ssd1 RNA-binding surface, and shown how their interaction with a conserved motif in the 5' UTRs of target mRNAs leads to translational repression and promotes cell wall integrity. To study Ssd1's functional evolution, we developed a plasmid-based system to express Ssd1 homologs in Saccharomyces cerevisiae ssd1Δ strains. We observed conservation of the fungal-specific RNA-binding function in clade Dikarya and a subset of mycorrhizal homologs, suggesting evolution of this function in the common ancestors of these groups, >700 mya. We confirmed that the fungal-specific RNA-binding function is not conserved in Schizosaccharomyces pombe Dis3L2 nuclease, but is conserved in some potentially bifunctional homologs. This data motivates a broader effort to reconstruct the molecular events that led to functional changes in the Ssd1/Dis3L2 family. The project is an exemplary dissection of the molecular evolutionary processes that underly an ancient transition in fungal lifestyle, from unicellularity to hyphal growth and saprotrophy.

[A319] Novel axes of coevolutionary innovation revealed via a cell-level study of butterfly detoxification mechanisms against mustard plants.

Brenda Irene Medina Jiménez[1], Katharina Schneider[1,2], Paula Fernández-Begne[3], Heiko Vogel[4], Prof. Chris Wheat[1]

[1]Stockholm University, Stockholm, Sweden, [2]Swedish University of Agricultural Sciences, Uppsala, Sweden, [3]University of Chicago, Chicago, United States, [4]Max Planck Institute for Chemical Ecology, Jena, Germany

Phytophagous insects such as the butterflies have evolved detoxification genes and gene families in response to the chemical defenses of their host-plants. In particular, the mechanisms pierinae butterflies (Lepidoptera: Pieridae) use to detoxify the glucosinolate (GSL) defenses of their host-plants in the mustard family (Brassicaceae) are well known. Here we explore how larvae respond to variation in GSL defences by studying spatial expression across cell lineages. Using single-nuclei RNA sequencing (snRNA-seq) of midguts from larvae fed on two host-plants differing in GSL composition, we found that the expression of several known detox genes (i.e. nitrile-specifier proteins, NSP) is cell lineage specific. We also reveal sets of co-expressed genes suggesting regulatory networks involved in context-dependent detoxification. We validated our findings using in situ hybridization and CRISPR experiments.

Beyond identifying a plethora of candidate genes for midgut cell-type identification in Lepidoptera, we find that the neofunctionalization of NSP detoxification genes occurred via divergence in gene regulatory networks and cell lineage deployment, adding a novel axis to the ways this detoxification mechanism has escalated in performance. These and other cellular-level insights into the regulatory dynamics of detoxification opens a new era of insect-plant coevolution research.

[A320] Introducing the Baboon Genotype-Tissue Expression Atlas (GTEx), a Community Resource for Comparative Primate Genomics

Brittany Hazzard[1], Dr Laura Cox[2], Dr. Gilad Yoav[1]
[1]University Of Chicago, CHICAGO, US, [2]Wake Forest University School of Medicine, Winston-Salem, US

Animal models are vital to the study of numerous areas of biology, from disease pathogenesis to human evolution. This is particularly true for primate models, which are invaluable for comparative genomic studies that aim to characterize the mechanisms driving human-specific adaptation. In order to extract the full potential of these models, high-quality, population-level genomic resources from multiple primate tissues are needed. While access to primate models has remained scarce, the baboon has proven to be a valuable resource for studies of human health, behavior, and evolution. To enhance the utility of this model, we have begun development of the baboon Genotype-Tissue Expression Project, a database that will utilize single-cell and bulk RNA-sequencing from 57 different tissues collected from 60 genotyped animals to establish a catalog of primate gene regulation for comparative genomic studies. Ultimately, we will use these data to map tissue-specific expression quantitative trait loci (eQTL) in baboons and explore gene regulatory differences between baboons and humans. In the first arm of this project, we performed single-nuclei RNA-sequencing to characterize ~1 million single nuclei from 57 archival tissues sampled from 3-6 individuals, including but not limited to various brain regions, heart, and kidney. We are using these data to deconvolute cell type proportions in bulk tissues, accounting for any individual biases in cell type that may be related to gene expression differences. Our efforts will deliver a large-scale single-cell atlas comprising dozens of baboon tissues, providing the scientific community with a vital resource for comparative genomic studies in primates.

[A321] Leveraging multimodal single-cell data of early primate development to study the evolution of regulatory elements

Dana Lopez-Parra[1], PhD Philipp Janssen[1], PhD Jessica Jocher[1], Antonia Keßler[1], PhD Fiona Edenhofer[1], Prof. Dr. Wolfgang Enard[1], PD Dr. rer. nat. Ines Hellmann[1]
[1]University of Munich, , Germany

Understanding the cis-regulatory code—how enhancers and promoters orchestrate transcriptional output—is central to decoding gene regulation, yet its principles remain elusive. While cis-regulatory elements (CREs) show poor positional conservation between species, gene expression appears remarkably conserved. This makes the comparative analysis of CRE activity and associated changes to regulatory phenotypes a powerful approach to understanding gene regulation, directly linking sequence variation to functional consequences.
We describe the creation of a reference dataset for the comparative study of early primate development through the generation of 10x snMultiome data from embryoid bodies from two human and two macaque individuals.
We address the key challenges in cross-species analysis of multimodal data along a developmental trajectory: identifying comparable cell types and constructing a shared chromatin accessibility feature space. To this end we developed a semi-automated computational pipeline combining classification and marker-based cluster annotation to identify comparable cell types across primates based on the snRNA-seq data. For chromatin accessibility analyses, we will identify orthologous CREs through whole-genome alignments and apply sequence-based activity prediction to establish functional correspondence across species.
This framework will enable quantitative assessment of regulatory network conservation and divergence, directly linking CRE sequence variation to species-specific expression phenotypes and revealing the principles governing transcriptional regulation in early primate development.

[A322] prime-seq2 and zUMIs-prime: an end-to-end framework for scalable cross-species transcriptomics

Felix Pförtner[1], Eva Briem[1], Dr Daniel Richter[1], Prof. Dr. Wolfgang Enard[1], PD Dr. rer. nat. Ines Hellmann[1]
[1]LMU Munich, Anthropology and Human Genomics, , Germany

Comparative transcriptomics is increasingly important to evolutionary biology, but cross-species RNA-seq studies are often limited by scalability and data analysis. Costs limit sample numbers and replicates, constraining statistical power and, thus, the biological insight achievable within a given budget. To make large, well-powered cross-species studies more affordable, we introduce an improved bulk RNA-seq protocol and a dedicated analysis workflow.
prime-seq2 is an optimized, UMI-based 3' early-barcoding bulk RNA-seq protocol that reduces per-sample cost while maintaining sensitive and specific expression quantification. 3' UMI-based protocols are especially suitable for cross-species experiments as they directly provide digital transcript counts. By systematically addressing protocol steps that drive read loss and technical variability, prime-seq2 increases the fraction of usable exonic+intronic UMIs by 60% while maintaining library complexity and technical variance. In practice, this reduces sequencing costs and, combined with low library costs and scalable multiplexing, enables large, well-controlled experimental setups across species.
To translate experimental standardization into computational comparability, we developed zUMIs-prime, a user-friendly processing workflow for UMI-based bulk RNA-seq data. zUMIs-prime automates mapping and UMI-aware counting with STARsolo and generates standardized QC summaries to diagnose potential artifacts. Downstream, modular, user-guided steps for filtering, visualization, differential expression testing, and power simulations support reproducible analyses and study design.
Together, prime-seq2 and zUMIs-prime make large, balanced cross-species transcriptomic designs more feasible, with standardized processing that strengthens reproducibility and interpretation.

[A323] Phylogenetic comparative analysis of single-cell transcriptomes: the evolution of gene expression noise in Saccharomyces

Fernanda Giersdorf[1], David Rogers[1], Julien Y. Dutheil[1]
[1]Max Planck Institute For Evolutionary Biology, , Germany

The role of natural selection in shaping gene expression is well established: higher or lower average expression of particular genes can confer fitness advantages. However, little evidence is available to assess how selection affects cell-to-cell variability in gene expression; can gene expression noise be adaptive or is variation between species attributable to genetic drift? Resolving this debate would require comparison of cell-to-cell variation in genome-wide expression of orthologous genes across a range of related species. Here, we generate a novel, large-scale single-cell transcriptomics dataset of 9 strains from 5 distinct Saccharomyces species and use it to demonstrate unequivocally that gene expression noise is not only profoundly shaped by selection, but the rate of evolution is comparable to that of mean expression itself. From estimates of both gene expression mean and noise for over 4,700 orthologous genes, we show that closely related strains exhibit significantly more similar noise profiles than distant species, allowing us to largely recapitulate the known species phylogeny from expression noise alone. Moreover, mapping gene expression noise as a trait onto a timed evolutionary framework, we demonstrate that different genes evolve noise at different rates.

[A324] S-LiDER: Exploiting Linkage Disequilibrium Geometry to Refine Functional Heritability Estimates

Hannah Snell[1], Dhruv Raghavan[1], Sohini Ramachandran[1], Dr. Ritambhara Singh[1]
[1]Brown University, Providence, United States

Among the complexities of understanding the heritability of diseases in humans, the variation of linkage disequilibrium (LD) across the human genome and between populations remains underexplored. Previous methods, such as LD Score Regression (LDSC, Bulik-Sullivan et al., 2015) and its functionally stratified version (S-LDSC, Finucane et al., 2015), use summary statistics from genome-wide association studies and LD to estimate heritability enrichment in various diseases. Both methods, however, collapse information from the LD matrix so that only the amount of LD is modeled per SNP, thus missing other LD-related patterns in the full LD matrix that may explain variation in disease heritability (such as SNP correlation with many weak independent signals versus one strong signal). As an extension of these methods, LD Eigenvalue Regression (LDER, Song et al., 2022), uses eigenvalue decomposition of the LD matrix to represent LD across spatial genomic regions. Building on these approaches, we present S-LiDER, a conceptual advance to LDER that incorporates functional information to refine heritability estimates. S-LiDER has two modes to represent LD structure: eigenvalue decomposition and a graph-based latent representation derived from a nonlinear model. We validate S-LiDER on quantitative traits and molecular biomarkers represented in the UK Biobank. In the future, we will assess its performance on more diverse genetic backgrounds and developmental insights at the single-cell level.

[A325] Modeling Sex-biased Gene Expression Evolution Across Firefly Phylogeny

Haoqing Du[1,2], Dr. Nicolas Lichilin[3], Dr. Ana Catalán[3], Dr. Sebastian Höhna[1,2]
[1]Department für Geo- und Umweltwissenschaften Paläontologie & Geobiologie,Ludwig-maximilians-universität München, München, Germany, [2]Geo-Bio Center, Ludwig-Maximilians-Universität München, München, Germany, [3]Division of Evolutionary Biology, Faculty of Biology, Ludwig-Maximilians-Universität München, , Germany

Fireflies exhibit various levels of sexual dimorphism, with extreme cases such as female neoteny occurring repeatedly across the whole phylogeny. The sexual dimorphism is often linked to sex-biased gene expression, yet the evolutionary processes driving the divergence of sex-biased gene expression remain poorly understood. Most previous studies have focused on identifying sex-biased genes within individual species, rather than explicitly modeling their evolutionary dynamics. Here, we applied phylogenetic models to directly investigate the evolution of sex-biased gene expression across fireflies. We generated gene expression data from two body parts (heads and bodies) for multiple individuals of both sexes per species, and modelled expression evolution with Ornstein-Uhlenbeck processes that incorporate within-species variances. Our analyses revealed the evolution of female-biased gene expression in the body across the phylogeny, while expression evolution in the head was largely unbiased for most genes. In addition, OU models incorporating branch-specific shifts in optimal expression provided evidence for directional selection acting on gene expression in both sexes within particular clades. Overall, our results provide a phylogenetically explicit view of sex-biased gene expression evolution in fireflies.

[A326] Evolution of alpha-solenoid proteins involved in organellar gene expression

Dr. Céline Cattelin[1,2], Dr. Charles H Robert[2], Dr. Francis-Andre Wollman[1], Dr. Sandrine Bujaldon[1], Ingrid Lafontaine[1]
[1]Photobiology and Physiology of Plastids and Microalgae, P3M, Sorbonne Université, CNRS, Paris, France, [2]Laboratoire de Biochimie Théorique, Université Paris Cité, CNRS, Paris, France

In photosynthetic eukaryotes of the green lineage, the expression of the chloroplast genome is mainly regulated post-transcriptionally, by RNA-binding proteins encoded in the nuclear genome termed organelle trans-acting factors. Most of those identified to date belong to two families of alpha-solenoid proteins- the pentatrico-peptide repeat (PPR) and and octotrico-peptide repeat (OPR)- and interact with specific sequences on their target mRNAs through a domain composed of repeated motifs, allowing their maturation, splicing, editing, stabilization and translation activation. We developed machine learning tools that allowed us to identify 3300 other alpha-solenoid candidates for experimental characterization in Archaeplastida (three of which being currently tested in the lab), which are likely to participate as new regulators of organelle gene expression. As for PPR and OPR, lineage-specific expansions of these new candidates have been observed, but mainly in Chlorophyceae. In particular, we identified new candidates in species in which the regulatory mechanisms of plastid gene expression are still understudied, such as in the glaucophyte Cyanophora paradoxa and the red alga Porphyridium purpureum.

[A327] Single-stranded and non-canonical DNA formation in human and other ape cells with telomere-to-telomere genomes

Dr Jacob Sieg[1], Huiqing Zeng[1], Hana Pálová[1], Linnéa Smeds[1], Saswat Mohanty[1], Angelika Lahnsteiner[2], Francesca Chiaromonte[1,3], Kateryna Makova[1]
[1]Penn State University, University Park, United States, [2] Paris Lodron University, Salzburg, Austria, [3]L'EMbeDS, Sant'Anna School of Advanced Studies , Pisa, Italy
Non-canonical (non-B) DNA secondary structures, such as G-quadruplexes, Z-DNA, cruciforms, and triplex DNA, are mutation hotspots and genome regulators that contribute to evolution and disease. Yet they remain uncharacterized in vivo in complete genomes. Here, we exploited the fact that many non-B DNA structures form single-stranded DNA (ssDNA). Using permanganate/S1 footprinting across 14 cell lines, we generated and analyzed ssDNA profiles for human and six non-human ape telomere-to-telomere (T2T) genomes. Our analyses revealed three major findings. First, Hidden Markov Models applied to our multispecies ssDNA data demonstrated that approximately 11% of ape genomes exist in a state with a high ssDNA level that is conserved across species. These conserved ssDNA loci correspond to genomic domains with specific functions—e.g., replication, transcription, and recombination—each enriched in distinct non-B DNA types. This finding implicates non-B DNA conformations in the regulation of these fundamental genomic processes. Second, in human embryonic and cancer cells, ssDNA was decreased at promoters and enhancers but increased at transposable elements, indicating altered structural regulation during development and cancer. Third, elevated ssDNA levels were found at satellite arrays (ribosomal DNA, centromeres, HSat3, and multiple species-specific satellites), which vary widely among species and were unresolved prior to T2T assemblies. Non-B DNA was enriched at many satellite arrays, and we confirmed its formation using orthogonal biophysical methods for select arrays. Thus, non-B DNA can contribute to satellite expansion and function. Taken together, our ssDNA analyses across ape T2T genomes uncovered conserved and species-specific DNA structural dynamics central to genome regulation.

[A328] uORFs as Evolutionary Drivers of Translational Stability and Gene Expression Regulation

Jian Lu[1], Zhijuan Wang[1]
[1]School Of Life Sciences, Peking University, Beijing, China

Upstream open reading frames (uORFs) are emerging as crucial cis-regulatory elements in the regulation of translation across eukaryotic genomes. While their role in modulating mRNA translation is well recognized, the evolutionary dynamics and contributions of uORFs to stabilizing protein expression levels remain poorly understood. In this study, we explore the evolutionary turnover of uORFs across Drosophila species, focusing on the birth–death dynamics of upstream start codons (uATGs) and their influence on translational variability. Our findings reveal that the retention of uORFs is tightly linked to translational output, with lineage-specific uATGs often associated with reduced translation of downstream coding sequences (CDSs), highlighting a lineage-dependent regulatory effect. Simulations and experimental deletions of uORFs show that these regulatory elements buffer fluctuations in gene translation, particularly during development and across evolutionary time. Notably, we uncover a novel function for uORFs in the regulation of the circadian rhythm, where uORFs in the Clk gene rhythmically attenuate CLK protein translation, influencing both circadian cycling and physiological processes such as sleep patterns. Our results provide new insights into how uORF turnover, natural selection, and translational regulation together shape the evolution of gene expression, offering a novel regulatory mechanism that maintains protein stability across developmental and environmental contexts.

[A329] Siphonophore genomics and mechanisms driving body polymorphism

Namrata Ahuja[1], Jamie Maziarz[2], Dr. Darrin Schultz[3], Dalila Destanovic[1], Dr. Jacob Musser[2], Dr. Casey Dunn[1]
[1]Yale Ecology And Evolutionary Biology, New Haven, United States, [2]Molecular, Cellular, Developmental Biology Department, Yale University, New Haven, United States, [3]Department of Neuroscience and Developmental Biology, University of Vienna , Vienna, Austria
Siphonophores (Cnidaria:Hydrozoa) are gelatinous animals characterized by their coloniality and body polymorphism. They have the largest diversity of functionally specialized bodies (zooids), which include those for feeding, defense and reproduction, and with many species having novel zooid types. Not much is known about how these zooid types develop, differentiate and how new zooids evolve. In order to gain insight into these processes at a finer resolution, we need to understand the mechanisms underlying their development at the genomic level. To do this, we sequenced, assembled and annotated the genome of Nanomia septata (1.5GB, n=8), and built genomic resources for other siphonophores, including a Physalia physalis genome and iso-sequencing references for a few other species. We found that no key cnidarian developmental gene families expanded within siphonophores, though siphonophore genomes are largely rearranged. Using our single cell RNA sequencing from multiple siphonophores, we looked to identify cell types and to better understand zooid specialization and development throughout the clade. We found a cell cluster that is associated with the pneumatophore (a novel feature of the siphonophore body plan), while zooid specific genes are scattered throughout the UMAP. We identified cell clusters primarily marked with marker genes for cnidocytes, gland, and muscle cells. These results show that cells cluster according to cell and tissue type, not specific zooid type. This suggests that differentiated zooids may share a general developmental program and later specialize through other processes.

[A330] A "phylogeny-in-a-dish" approach to study the evolution of context-dependent gene expression in primates

Nelly Helmbrecht[1], Kathrin Köhler[1], Jenny Tung[1,2], Amanda Lea[3], Genevieve Housman[1]
[1]Department of Primate Behavior and Evolution, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany, [2]Departments of Evolutionary Anthropology and Biology, Duke University, Durham, USA, [3]Department of Biological Sciences, Vanderbilt University, Nashville, USA

Genetic variation can facilitate diverse responses to the same environmental stimuli, suggesting that genotype-by-environment interactions (GxE) may play an important role in adaptation in response to environmental selection pressures. However, studying GxE in species that cannot be maintained in controlled environments remains a challenge. In humans, recent developments to study GxE include “village-in-a-dish” cell culture approaches, in which cell lines from multiple individuals are pooled in mixed cultures that can be efficiently tested across multiple experimental perturbations. Employing this approach for eQTL detection increases throughput per experiment while reducing cost and experimental variability. Here, we explore whether this approach can be expanded even further to a “phylogeny-in-a-dish” approach to study GxE evolution in nonhuman primates.

We pooled lymphoblastoid cell lines derived from multiple individuals and species (5 chimpanzees, 2 bonobos, 2 gorillas, and 2 orangutans) and exposed these mixed cultures to up to 45 perturbation and control conditions. We generated single-cell expression (scRNA) profiles for a total of 231 individual-condition combinations. We then compared these data to scRNA and bulk expression (bulkRNA) data generated from unpooled cell cultures in parallel experiments.

Different conditions produced different responses, and we use scATAC data to further explore the role of steady-state chromatin landscapes in conserved and divergent responses among species. Despite this, for both perturbed and control conditions, matched samples showed highly correlated expression patterns between pooled and unpooled culture systems. Overall, this study demonstrates that the phylogeny-in-a-dish approach facilitates research on environmental responses both across many individuals and across evolutionary time scales.

[A331] Disruption of DNA methylation in the hybrid Italian sparrow contributes to transgressive gene expression

Nicholas Planidin[1], Dr Rachel Steward[1], Alex Lawrence[1], Kalle Tunström[1], Petronella Wessman[1], Anna Runemark[1]

[1]Lund University, Lund, Sweden

There is growing evidence that hybridization is an important mechanism for producing novel phenotypes from standing genetic variation. However, the molecular mechanisms underlying the generation of evolutionary novelty in hybrids are poorly understood. For example, hybrid species often exhibit transgressive gene expression, where expression in hybrids is more extreme than in either parental species, which can contribute to novel phenotypic variation. While theory suggests that transgressive expression can arise due to the disruption of regulatory elements in the hybrid genome, there is little empirical evidence testing this hypothesis. To address this open question, we compare changes in gene expression to changes in DNA methylation in the hybrid Italian sparrow (Passer italiae). Specifically, we examine whether transgressively expressed genes exhibit altered DNA methylation at their promoter, gene body, and nearby regulatory elements. Furthermore, we explore whether regions of transposable element proliferation in the P. italiae genome are characterized by reduced DNA methylation. Lastly, by performing these analyses in four independently derived lineages of P. italiae, we examine the repeatability of disruptions to gene regulation and how it contributes to parallelism, or lack thereof, in transgressive expression. By comparing changes in DNA methylation to transgressive gene expression in a hybrid species, we empirically test the role of dysregulation of gene expression in the evolution of novel phenotypes.

[A332] Convergent Gill Architecture, Divergent Cellular Programs: a Multi-omics Approach to Arthropod Breathing

Polychronis Tatsis[1], Klara Elftheriadi[1], Judit Salces[1], Belen Carbonetto[1], David Salamanca-Diaz[2], Jordi Solana[2], Rosa Fernandez[1]
[1]Metazoa Phylogenomics & Genome Evolution Lab, IBE (Instituto de Biologia Evolutiva), Barcelona, Spain, [2]Living Systems Institute, University of Exeter, UK, Exeter, UK
Arthropods mastered the terrestrial biosphere by fundamentally adapting their modes of breathing. While many lineages evolved morphologically dissimilar novel respiratory structures (such as tracheae or lungs), others present a striking evolutionary paradox: the aquatic horseshoe crab (Limulus polyphemus) and the terrestrial crab (Discoplax magna) utilize morphologically similar organs, book gills and gills respectively, to breathe in drastically different physical environments. While these organs exhibit morphological convergence, retaining a shared architecture, the genetic mechanisms that allow this identical form to function in both water and air remain unknown. In order to investigate whether the same gene and cellular programs underlie these morphologically similar organs, we integrated single-nucleus/single-cell RNA-sequencing atlases for these species, together with spatial transcriptomics and Weighted Gene Co-expression Network Analysis (WGCNA), where we further identified regulatory hub genes and spatially localized expression domains. Our scRNA-seq data indicate that the aquatic L. polyphemus book gill exhibits a sensory-active profile, rich in oxygen-sensing (Neuroglobin+) and hormone-responsive cells, reflecting its exposure and dual role in swimming. In contrast, the terrestrial D. magna gill is dominated by structural and contractile cell populations (Titin+, Ankyrin+), suggesting a structurally reinforced organ to resist gravitational collapse within an enclosed chamber in the terrestrial crab. By integrating these three transcriptomic lenses, we provide the first multi-omics approach to these organs, providing key insights into the molecular mechanisms underlying their morphological convergence.

[A333] Gene Coexpression and Microsynteny Networks Uncover Regulatory Constraints and Exaptations for C4 Evolvability in the Brassicales

R. Shawn Abrahams[1]
[1]University of Illinois, Urbana, United States

C4 photosynthesis is among the most convergent traits in eukaryotes, representing a carbon-concentrating mechanism where Rubisco is spatially segregated to bundle sheath cells to reduce photorespiration. This adaptation to hot, arid conditions and low atmospheric CO2 has evolved independently over 60 times across angiosperms. While some clades exhibit numerous independent C4 origins, the Brassicales maintain relatively few despite occupying suitable environments. Here, we integrate gene co-expression networks and gene family microsynteny networks to uncover the genetic architecture underlying C4 evolvability in this clade. Comparative network analysis shows that C3 species' co-expression networks are more similar to each other than to C4 species, yet key topological differences signal early co-option of important pathway enzymes in C4 lineages. Our microsynteny network approach predicts cis-regulatory conservation across evolutionary timescales, providing a framework for understanding pathway assembly constraints. Our analyses reveal syntenic context divergence in the PEPC gene family correlates with tissue-specific expression, limiting the available substrate for PEPC co-option into C4 pathways. We demonstrate that C4-like aminotransferase expression patterns occur in C3 relatives of the Cleomaceae, suggesting pre-existing regulatory architectures. However, co-expression patterns resembling C4 evolution in NAD-ME genes do not appear to represent an exaptation when considering the underlying genetic constraints. Overall, our findings reveal how lineage-specific genetic architecture shapes the probability of C4 evolution, with syntenic organization and regulatory context serving as both enablers and constraints on photosynthetic innovation in the order Brassicales.

[A334] Single-cell multi-omics analysis of maturation age in Atlantic salmon testes

Rachel Carboni[1], PhD Jukka-Pekka Verta[1]
[1]Nord University, Bodø, Norway

Life-history traits are shaped by complex genetic and developmental mechanisms, influencing fitness, adaptive divergence, and evolution. In Atlantic salmon (Salmo salar), allelic variation in the transcription cofactor vgll3 explains nearly 40% of the variation in maturation age through cis-regulatory control of gene expression. Vgll3 is also associated with pubertal timing as well as sex-specific autoimmune diseases in humans, although with smaller effects. Previous research in salmon has found that vgll3 expression was highest in the immature testes and decreased after initiation of puberty. This raises the hypothesis that vgll3 influences cellular differentiation and proliferation through cell-type specific signaling pathways, yet these are not clear based on bulk tissue analyses. To understand the mechanisms mediating genotype-phenotype association in vgll3 at a cellular resolution, we generated the first single-cell atlas of Atlantic salmon testes using the 10x Genomics Multiome ATAC + Gene Expression platform. We sequenced testes from 8 males with different vgll3 genotypes before and after pubertal initiation. We identified 17 distinct clusters of cells based on their transcriptomes, likely representing 10-12 known cell-types. Initial analyses revealed distinct differences in cellular composition between vgll3 genotypes and maturation states. Ongoing analyses of gene expression and chromatin accessibility aim to identify distinct developmental trajectories and gene regulatory networks associated with vgll3 genotype and maturation state for each cell-type. Our study connects genetic differences in a large effect gene to the cell-type specific mechanisms underlying variation in life-history traits that ultimately affect fitness and adaptive divergence.

[A335] Conserved Secretory Programs Across Neuronal and Non-Neuronal Cell Types

Shreejita Chatterjee[1,2], Dr. Judith Bossen[1], Prof. Dr. Thomas Roeder[1]
[1]Christian-albrechts Universitat Zu Kiel, , Germany, [2]Max Planck Institute for Evolutionary Biology , , Germany

Neurons are traditionally characterized by specialized molecular machinery that enables regulated secretion and rapid signal transmission. It remains an open evolutionary question whether this machinery is a neuron-specific innovation or reflects a more ancient, conserved secretory program. In insects, Inka cells are rare epithelial cells embedded in the larval tracheal system of Drosophila melanogaster, traditionally described as non-neuronal endocrine cells that secrete the ecdysis-triggering hormone (ETH). However, recent transcriptomic data challenge this rigid classification, suggesting that neuron-like molecular features may extend beyond the nervous system. Here, we explore the molecular identity and organizational context of Inka cells through integrative transcriptomic analyses combined with genetic approaches.Analysis of larval tracheal cell populations reveals that Inka cells are closely associated with neighboring epithelial cell types, together forming a previously unrecognized epitracheal organ. Within this structure, a distinct cell population exhibits transcriptional enrichment of genes associated with juvenile hormone biosynthesis, suggesting potential coordination among multiple endocrine pathways within a single epithelial system. To place these findings in a broader evolutionary framework, we examined single-cell expression patterns across diverse Drosophila tissues. This comparative perspective indicates that several non-neuronal endocrine and secretory cell types share molecular features commonly associated with neurons, particularly those related to regulated secretion. Together our results support the idea that neuron-like molecular characteristics are distributed across multiple epithelial endocrine systems. The epitracheal organ offers a tractable model to explore how conserved secretory programs are deployed and diversified during the evolution of specialised cell types.

[A336] Breaking the Loop: Cis-Regulatory Disruption of FoxP Autoregulation during Wolf Domestication

Prof. Wiesław Bogdanowicz[1], Msc. Aleksandra G Bilska[1], Dr. Jan Jabłonka[2], Dr. Enikő Kubinyi[3], Dr. Małgorzata Pilot[4], Tadeusz Malewski[1]
[1]Museum and Institute of Zoology, Polish Academy of Sciences, Warsaw, Poland, [2]Institute of Experimental Zoology, University of Warsaw, Warsaw, Poland, [3]Department of Ethology, Eötvös Loránd University (ELTE), Budapest, Hungary, [4]Faculty of Biology, University of Gdańsk, Gdańsk, Poland

The domestication of the gray wolf (Canis lupus) involved extensive behavioral diversification, including changes in vocal communication. While barking is typical of most modern dog breeds, several “ancient” breeds retain wolf-like vocal repertoires, suggesting that derived vocal traits evolved through modification of neural pathways. However, the genomic mechanisms underlying these differences, particularly regulatory variation, remain poorly understood. Members of the FoxP subfamily of forkhead transcription factors (FoxP1, FoxP2, and FoxP4) are key regulators of neural development and vocal communication. In humans, FoxP proteins are essential for speech and language, and in birds they regulate song learning, indicating a deeply conserved role in vertebrate vocal behavior. We hypothesized that changes in cis-regulatory architecture, particularly within potential autoregulatory feedback loops of FoxP genes, contributed to vocal divergence during wolf domestication. To investigate this, we analyzed 5’ upstream regions of FoxP transcripts for transcription factor binding sites (TFBSs) using the JASPAR motif database. Predicted FoxP binding sites occurred in 18 of 23 upstream regions—including all FoxP2 and most FoxP1 transcripts—supporting possible autoregulatory feedback, while FoxP4 showed limited evidence. Comparative genomic analyses revealed a domestication-associated loss of predicted FoxP binding sites in dogs. Across five dog breeds (incl. “ancient” Basenji), these specific FoxP sites were absent from two FoxP1 and two FoxP2 upstream regions that are conserved in wolves. These findings suggest that domestication disrupted FoxP autoregulatory circuits through cis-regulatory changes, potentially altering neural gene expression underlying vocal behavior. This highlights the importance of investigating regulatory evolution in shaping complex behavioral traits.

[A337] Regulatory Inheritance Explains Convergent Expression Patterns in Hybrids and Polyploids

Tomáš Tichopád[1], dr. Jan Eisner[2], dr. Petr Cígler[3], Karel Janko[4]
[1]Faculty of Fisheries and Protection of Waters, University of South Bohemia in České Budějovice, South Bohemian Research Center of Aquaculture and Biodiversity of Hydrocenoses, Zátiší 728/II, 389 25, Vodňany, Czech Republic, [2]Department of Mathematics, Faculty of Science, University of South Bohemia in České Budějovice, Branišovská 1760, 370 05, České Budějovice, Czech Republic, [3]Institute of Organic Chemistry and Biochemistry, Czech Academy of Sciences, 166 10, Prague, Czech Republic, [4]Laboratory of Non-Mendelian Evolution, Institute of Animal Physiology and Genetics of the Czech Academy of Sciences, Rumburská 89, 277 21, Liběchov, Czech Republic

Hybridization and polyploidy are major evolutionary processes with profound impacts on gene expression regulation. Commonly observed patterns arise repeatedly across distantly related hybrid and polyploid taxa including non-additive expression of parental alleles, subgenome dominance, genomic dosage deviations, and transcriptome downsizing. Mechanisms underlying such patterns remain debated and a central unresolved question is whether these outcomes emerge instantaneously as a direct consequence of genome merging and doubling, or whether they evolve subsequently under convergent evolution among unrelated taxa.

We present a unifying framework that disentangles the regulatory effects of hybridization from those of polyploidy, using a thermodynamic model of gene regulation explicitly modeling regulatory divergence between parental species. The model allows us to evaluate how differences in parental gene expression, cell volume, and euchromatic content translate into gene expression patterns in hybrid and polyploid genomes. Across a wide range of biologically realistic scenarios, the model reproduces key empirical patterns observed in natural systems, including expression-level dominance, transcriptome downsizing, and systematic allelic biases.

Our results indicate that many characteristic regulatory features of hybrids and polyploids arise directly from the constrained interaction of inherited, diverged regulatory networks, without requiring subsequent adaptive fine tuning. These theoretical predictions are supported by allele-specific expression data from diploid and polyploid hybrids, which reveal consistent regulatory patterns across unrelated taxa and closely match model expectations.

By integrating modeling with empirical evidence, this work provides a general, predictive framework for understanding gene regulation in hybrid and polyploid organisms, offering insight into the regulatory consequences of genome merging and multiplication.

[A338] Evolution of regulatory chromatin contacts: insights from duplicated genes

Victor Lefebvre[1], PhD Sarah Djebali[2], PhD Sylvain Foissac[3], Anamaria Necsulea[1]
[1]Laboratory of Biometry and Evolutionary Biology (LBBE), Villeurbanne, France, [2]Institut de recherche en santé digestive (IRSD), Toulouse, France, [3]Genetics Physiology and Breeding Systems (GenPhySE), Toulouse, France

In eukaryotes, gene expression is controlled by numerous cis-regulatory elements that can be located far away from their target genes on the linear genome. In these cases, interactions between gene promoters and distant regulatory elements take place through chromatin contacts. These chromatin contacts can now be detected thanks to high-throughput chromosome conformation capture techniques (Hi-C]). While other aspects of gene expression regulation have been well studied, the evolution of regulatory chromatin contacts remains largely unexplored, mainly due to a lack of comparable data across species.

To bypass this problem of cross-species data comparability, we use duplicated genes as a model to study the evolution of regulatory chromatin contacts. This also allows us to study multiple divergence times, within a single species.

We analysed the expression patterns and the chromatin contacts of 1420 pairs of duplicated genes in mouse, using an extensive collection of promoter-centered Hi-C data. We observe that chromatin contacts evolve faster than expression patterns, as previously observed for other aspects of gene regulation. We show that duplicated gene localisation in cis or trans is the main factor associated with chromatin contact divergence. We find a weak but significant correlation between expression divergence and chromatin contact divergence across duplicated gene pairs. This confirms that chromatin contacts play an important role in the evolution of gene expression regulation.

Our results highlight the complex evolutionary dynamics of regulatory chromatin contacts, as well as the relationship with gene expression evolution, across a broad time scale.

[A339] From SNP to function: how combinations of two large-effect locus genotypes shape maturity age through gene regulatory networks

Xin-Di Huang[1], PhD Morgane Frapin[1], PhD Ehsan Pashay Ahi[1], Professor Craig Primmer[1], PhD Jukka-Pekka Verta[2]

[1]Organismal and Evolutionary Biology Research Program, Faculty of Biological and Environmental Sciences, University of Helsinki, Helsinki, Finland, [2]Genomics Division, Faculty of Biosciences and Aquaculture, Nord University, Bodø, Norway

The genotype-phenotype map is fundamental to evolutionary biology, yet GWAS approaches often leave the regulatory mechanisms linking genotype to phenotype unresolved. In Atlantic salmon, variation in a key life-history trait, age-at-maturity, is strongly associated with two large-effect loci linked to the transcriptional regulators vgll3 and six6. This system provides a unique opportunity to investigate how combinations of large-effect locus genotypes shape molecular processes underlying life-history variation. Here, we analyzed gonadal transcriptomes from 90 individuals with homozygous combinations of late (L) and early (E) maturation alleles for vgll3 and six6 during the spawning season. Applying a new transcriptome assembly strategy, we substantially improved genome annotation and identified thousands of previously uncharacterized genes and long non-coding RNAs (lncRNAs), many showing testis-specific expression implicating novel molecular roles in maturation. Gene expression analyses revealed that differentially expressed genes (DEGs) associated with vgll3 and six6 genotypes were significantly enriched on their respective chromosomes (Chr25 and Chr9) across both testis and ovary, indicating chromosome-centered regulatory architectures. Interestingly, undeveloped testis showed a distinct transcriptional profile, with many genes transcribed at higher levels in LL compared to EE genotypes for both loci. 196 DEGs were shared between vgll3*LL and six6*LL genotypes, suggesting joint regulatory effects associated with late maturation. Gene co-expression analyses further identified genotype-specific gene-lncRNA modules. Together, our results demonstrate that large-effect loci shape age-at-maturity through dynamic, stage-specific combined effects as well as stable, chromosome-centered regulatory architectures, providing new insight into how individual and interacting loci contribute to the evolution of complex life-history traits.

[A340] Integrated GWAS and Transcriptome Analyses Identify Candidate Genes for Thick-leg Trait of Vietnamese Dragon Chickens

Xufang Ren[1], XIURONG ZHAO[1], XIAOYU JIANG[1], DENGJING ZUO[1], TAIRAN CHEN[1], GANG WANG[1], XINYE ZHANG[1], Professor LUJAING QU[1]
[1]China Agricultural University, Haidian District, China

Genetic variation drives phenotypic diversity, yet the molecular basis of the distinct thick-leg trait in Vietnamese Dragon Chicken remains unclear. Here, we integrated phenotypic analysis, genome-wide association studies in inter-breed and F_2 populations, and tissue-specific transcriptomics of metatarsal skin, metatarsal cartilage, calf muscle and dorsal skin to decipher this trait's genetic mechanisms. Phenotypic assessment confirmed it is a polygenic trait with non-Mendelian inheritance. Inter-breed genome-wide association studies identified 95 candidate genes including RNF32, ITPRID1 and TOX, while F_2 genome-wide association studies detected 4 genes; only TOX was shared, indicating cross-population stability. Transcriptomic analysis revealed differential expression of genome-wide association studies-identified genes in leg tissues, including TOX and ITPRID1 in metatarsal skin, and ITPRID1 in cartilage. ITPRID1 harbors a 7.8 kb ALVE insertion, but this insertion alone did not directly affect leg circumference. KEGG enrichment highlighted 7 skeletal development-related pathways in metatarsal skin. Collectively, the thick-leg trait is regulated by a polygenic network involving conserved TOX and breed-specific genes like ITPRID1, with tarsometatarsal skin as a key tissue. This study provides comprehensive insights into the genetic mechanisms and molecular basis of the thick-leg trait in Vietnamese Dragon chicken and offers valuable genetic markers for molecular breeding.

[A341] Hypomethylation in Oikopleura dioica genome

Yael Klirs[1], Simona Buonanno[2], Prof Carmela Gissi[2,3,4], Prof Tal Pupko5[5], Prof Dorothee Huchon[1,6]
[1]School of Zoology, George S. Wise Faculty of Life Sciences, Tel-Aviv University, Tel-Aviv, Israel, [2]Department of Biosciences, Biotechnology and Environment, University of Bari Aldo Moro, Bari , Italy , [3]Institute of Biomembranes, Bioenergetics and Molecular Biotechnologies, Consiglio Nazionale delle Ricerche, Bari , Italy , [4]Consorzio Nazionale Interuniversitario per le Scienze del Mare, Roma, Italy , [5]George S. Wise Faculty of Life Sciences, The Shmunis School of Biomedicine and Cancer Research, Tel Aviv University, Tel Aviv , Israel, [6]The Steinhardt Museum of Natural History and National Research Center, Tel Aviv University, Tel Aviv , Israel

Cytosine DNA methylation is a conserved eukaryotic epigenetic modification that plays central roles in gene expression, cellular differentiation, and development. It is widespread across animal phyla, with only a few documented cases of secondary loss. Methylation patterns differ among animals: vertebrates typically exhibit high, genome-wide CpG methylation, whereas invertebrates display sparse or mosaic methylation, with some genes heavily methylated and others nearly unmethylated. Because methylated cytosines are prone to spontaneous deamination to thymine, methylated regions lose CpG sites over evolutionary time, producing CpG observed/expected (O/E) ratios below one; the mosaic methylation characteristic of invertebrates therefore gives rise to bimodal CpG O/E distributions.

We investigated DNA methylation in tunicates and found that the appendicularian Oikopleura dioica exhibits a unimodal CpG O/E distribution centered around one, consistent with absence of cytosine methylation. To test this hypothesis, we searched tunicate proteomes for core enzymes required for cytosine methylation and demethylation. HMMER analyses indicate that O. dioica lacks canonical components of the methylation machinery. In parallel, analysis of Oxford Nanopore reads identified a small set of CpG sites with high apparent methylation frequencies, yet the overall distribution of fractional methylation differed markedly from that seen in other tunicates. In the absence of canonical methylation machinery, and considering the background noise inherent to Nanopore methylation detection, the biological relevance of these signals remains uncertain.

Taken together, our results support loss of genome-wide cytosine methylation in O. dioica, placing this species among the rare animal lineages which underwent secondary loss of DNA methylation.

[A343] A century of decline: tracking genomic erosion in two endangered parrots

Alexandra Brumwell[1], Carolina Pacheco[2], Xuejing Wang[3], Cock Van Oosterhout[4], Carolyn J. Hogg[5], Luke W. Silver[5], Soleille M. Miller[5], Dejan Stojanovic[6], Prof Jim Groombridge[1], Dr Hernán Morales[3]
[1]Durrell Institute of Conservation and Ecology, University Of Kent, Canterbury, United Kingdom, [2]Department of Biology, Science for Life Laboratory, Lund University, Lund, Sweden, [3]Globe Institute, University of Copenhagen, Copenhagen, Denmark, [4]School of Environmental Sciences, University of East Anglia , Norwich, United Kingdom, [5]School of Life and Environmental Sciences, The University of Sydney, Sydney, Australia, [6]Fenner School of Environment and Society, Australian National University, Canberra, Australia

A major challenge in conservation genomics is finding clear, quantifiable ways to assess genomic erosion and its consequences for genetic viability and extinction risk. Traditional approaches have largely relied on genome-wide diversity metrics, but these alone may not fully capture the long-term genomic effects of population decline. Recent advances in whole-genome sequencing (WGS), together with improved methods for extracting and sequencing historical DNA, now make it possible to investigate genomic erosion in greater detail by integrating information on genetic diversity, inbreeding, and genetic load through time.

Using temporal WGS datasets from two critically endangered, phylogenetically divergent parrots—the Orange-bellied Parrot (Neophema chrysogaster) and the Swift Parrot (Lathamus discolor)—that have undergone broadly similar rapid population collapses, we examined genomic change across more than a century of demographic decline. By combining modern genomes with historical museum specimens as early as the 1840s, we reconstructed genetic baselines prior to severe population declines and assessed temporal trends in genome-wide heterozygosity, runs of homozygosity, inbreeding, and genetic load. Using a consistent analytical pipeline, we further evaluated a comparative framework for studying genomic erosion while addressing challenges associated with degraded historical DNA.

By comparing multiple genomic metrics across time, we demonstrate how genomic erosion can be quantified and interpreted using temporal WGS data. Our results highlight the importance of integrating multiple genomic indicators to understand extinction risk and illustrate how temporal genomics can improve conservation assessments and decision-making for declining populations.

[A344] A genomic perspective on the evolution of modern water voles (Arvicola sp.)

Amanda Lindahl[1,2]
[1]Centre for Palaeogenetics, Stockholm, Sweden, [2]Department of Zoology, Stockholm University, Stockholm, Sweden

Water voles (Arvicola sp.) are rodents with a complex evolutionary history. Since their origin in the Pleistocene epoch, they have lived through large-scale climate changes which has shaped the distribution of lineages seen today. Although there is distinct morphological variation between different populations of water voles, little work has been done to investigate the genetic relationships within the genus. Previous work using mitochondrial and nuclear DNA markers has revealed at least five different lineages within the genus: Arvicola amphibius, A. sapidus, A. italicus, A. persicus and A. monticola. However, DNA markers are limited in the amount of information they can reveal regarding a species evolutionary history. Here, we analyze mitochondrial and nuclear whole genome data from ~100 modern and a few ancient specimens from most of the genus current range throughout Eurasia. Our mitochondrial phylogeny is largely concordant with previous findings, but we discover an additional, previously unidentified lineage of water voles. The current taxonomic status of the genus is further challenged by our findings of extensive gene flow between populations currently classified as separate species. Furthermore, we estimate divergence times between different lineages and changes in effective population size over time. Taken together, these results provide us with a comprehensive overview of the genus evolutionary history, which we discuss in the context of large-scale climate change and how it can shape the genetic structure of small mammals.

[A345] Ultrarapid MC1R protein and associated plumage colour evolution in the domestic chicken

Cheng Ma[1], Aenne-Dorothea Liebing[2], Gunnar Kleinau[3], Antje Kamprad[3], Louisa Calabrese[3], Michal Szcepeck[3], Zheng Li[1,4], Mårten Larsson[1], Patrick Scheerer[3], Claudia Stäubert[2], Leif Andersson[1,5]
[1]Department of Medical Biochemistry and Microbiology, Uppsala University, Uppsala, Sweden, [2]Rudolf Schönheimer Institute of Biochemistry, Faculty of Medicine, Leipzig University, Leipzig, Germany, [3]Charité – Universitätsmedizin Berlin, corporate member of Freie Universität Berlin, Humboldt-Universität zu Berlin, Institute of Medical Physics and Biophysics, Group Structural Biology of Cellular Signaling, Berlin, Germany, [4]Ministry of Education Key Laboratory for Biodiversity and Ecological Engineering, College of Life Sciences, Beijing Normal University, Beijing, China, [5]Department of Veterinary Integrative Biosciences, College of Veterinary Medicine and Biomedical Sciences, Texas A&M University, College Station, USA

The domestic chicken exhibits extraordinarily high plumage diversity in sharp contrast to the uniform plumage phenotype in its wild ancestor, the red junglefowl. The melanocortin-1 receptor (MC1R) locus is the most variable pigmentation locus in domestic chicken. Here we have analysed whole-genome sequence data from 10,026 birds, including red junglefowls and 393 populations of domestic chicken. We document ultrarapid evolution of 18 MC1R alleles, due to the accumulation of 9 missense mutations combined with reshuffling of sequence variants within the single MC1R exon. The lack of linkage disequilibrium at the MC1R locus strongly suggests that this combinatorial allele diversity is generated by interallelic gene conversion, possibly promoted by the high guanine-cytosine (GC) content at this locus. Most detected missense mutations have functional consequences for MC1R signalling, as revealed by functional analyses of chicken MC1R variants in transfected cells using second-messenger and bioluminescence resonance energy transfer (BRET) assays, combined with structural information extracted from homology modelling. Both activating and inhibiting mutations were identified. Bioinformatic analysis of whole-genome sequence data also showed that MC1R acts as a modifier locus for the dominant white and recessive white phenotypes caused by mutations in the premelanosome protein (PMEL) and tyrosinase (TYR) genes, respectively. In both cases, MC1R alleles promote a pure white phenotype, either by enhancing eumelanin pigmentation (dominant white) or inhibiting eumelanin pigmentation (recessive white). Finally, the MC1R locus in chicken provides a striking example of allele evolution by the accumulation of multiple mutations with phenotypic effects.

[A346] Estimating Seagrass Meadow Persistence via Clone Aging

Clara E. Winguth[1], Lauren R. Sgambelluri[1], Érika E. Kokubun[1], Dr. Till Bayer[1], Dr. Lei Yu[2], Prof. Dr. Thorsten Reusch[1]
[1]Marine Evolutionary Ecology, GEOMAR Helmholtz Center Of Ocean Research Kiel, Kiel, Germany, [2]Laboratory of Marine Ecology and Environmental Sciences, Institute of Oceanology, Chinese Academy of Sciences, Qingdao, China

Age distributions of many clonal plant species remain elusive, limiting our knowledge on their demography. A recently proposed somatic genetic clock holds great promise for assessing the longevity and persistence of clonal populations. Here, we apply such a clock to populations of a widespread seagrass (eelgrass; Zostera marina), a foundational species that enhances biodiversity, prevents coastal erosion, and stores blue carbon. Clones of eelgrass form via branching and are independent within a few years, resulting in asexual populations of individuals (or ramets) belonging to the same genet (sexually derived individual). The somatic genetic clock measures the somatic genetic variation within a genet that segregates among ramets through a process of somatic genetic drift, with the number of fixed somatic genetic differences being proportional to genet age. Accordingly, we aged 80 genets (249 total ramets) at the seascape scale at 16 eelgrass meadows in the western Baltic Sea. We used microsatellite markers to identify clones, which were then whole-genome resequenced to 50x coverage, and somatic genetic variation was called using Mutect2. Using a previously established standard curve based on clones of known age, we estimated the age of the genets as a proxy for the minimal age of local seagrass meadows. These findings provide a baseline for ecosystem management, as older seagrass meadows have more established ecosystem services, and shed light into eelgrass' current resilience despite clonality.

[A347] Historical and Recent Population Specific Bottlenecks Inflated Differentiation Measures of Louisiana Black Bear

Emily Puckett[1], Heather Clendenin[1]
[1]University of Memphis, Memphis, United States

Significant debate has revolved around the delimitation of subspecific boundaries relative to conservation policy, and specifically how best to maximize limited resources. Frequently, the conservation of subspecies preserves unique genetic diversity, aiding in the long-term preservation of adaptive potential. We evaluate patterns of neutral and adaptive genomic variation across the eastern lineage of the American black bear, and focus on the formally federally protected subpopulations in Louisiana. Despite high values of genetic differentiation (Fst >0.127) of Louisiana populations, we show that serial founder events during range expansion across eastern North America, as well as repeated and population-specific anthropogenic bottlenecks resulted in low genetic diversity. Bottlenecks occurred earlier (1kya) in the northern population of the state and genetic purging of deleterious alleles was observed. Whereas genetic purging has had a weaker impact in the southern population with the more recent bottleneck (0.33kya); this is notable given deleterious phenotypes observed on the landscape in the south. We further show that local adaptation has had a smaller impact than phylogeography on the distribution of genetic variation across the eastern lineage. Our genomic assessment does not support the continued recognition of Ursus americanus luteolus as a subspecies of American black bears. Continued genetic conservation efforts should focus on maintaining or increasing diversity, while supporting ongoing successes of demographic recovery.

[A348] Drought drives inbreeding and local adaptation in a rare endemic of California's Fog Belt

Emma Steigerwald[1], Dr Matthew Guilliams[2], Dr Kristen Lehman[2], Dr Rachel Meyer[1]
[1]University of California, Santa Cruz, , United States, [2]Santa Barbara Botanic Garden, Santa Barbara, United States

California's fog belt harbors one of the world's fog-fed ecosystems, renowned for exceptional plant diversity and endemism. Many species endemic to these coastal systems depend on fog as a critical water source. As fog regimes change rapidly under contemporary climate change, understanding the genomic basis of local adaptation to fog could assist in the conservation management of fog-reliant endemics. We investigated this question in Gaviota tarplant (Deinandra increscens ssp. villosa), an endemic of the Point Conception region—the area experiencing the most dramatic contemporary shifts in fog cover in the state. Using whole-genome resequencing of individuals sampled across the species' fragmented range, we found strong genetic structure among geographically isolated populations, with inbreeding levels predicted by local drought exposure. Genome-wide association analyses revealed a robust signal of local adaptation to fog gradients, with outlier loci showing significant gene ontology (GO) enrichment for biological processes that suggest coordinated responses for optimizing photosynthesis under variable light and mounting antimicrobial defenses. A large, continuous block of outlier SNPs on chromosome six proved to be a massive chromosomal inversion. Comparative synteny analysis across the sunflower tribe (Heliantheae) revealed that this genomic region has been subject to recurrent inversion-based rearrangements, suggesting it represents a long-standing axis of adaptive variation within this alliance. Characterizing the functional and evolutionary dynamics of this inversion polymorphism could ultimately enable managers to identify climate-resilient genotypes for restoration efforts, predict which populations are most vulnerable to fog decline, and prioritize conservation interventions accordingly.

[A349] Effects of fragmented landscapes on the evolution of thermal tolerance in soil arthropods.

Fatima Jlil[1], Daniel Bang Jørgensen[1], Anne Aagaard[1], Jilong Ma[1], Dr. Jesper Bechsgaard[1], Dr Mads Fristrup Schou[1], Trine Bilde[1]
[1]Aarhus University, Aarhus, Denmark

Species' tolerance to environmental conditions relies on a delicate balance between phenotype, genotype, and environment, shaped across space and time by evolutionary forces such as natural selection and genetic drift. Discerning the signature of these processes is challenging, and they are greatly impacted by the species' own dispersal abilities and the levels of habitat fragmentation. Due to their limited mobility, soil-dwelling arthropods like springtails represent an ideal system to investigate the process of adaptation in a low-dispersive taxon and to distinguish the role of selection from drift.

Previous studies have identified small phenotypic differences in heat tolerance between populations but strong latitudinal patterns across large geographical ranges, suggesting local adaptation. However, the degree to which phenotypic variation persists within smaller, less variable geographic regions is poorly understood.

In this study, we evaluated thermal tolerance in 23 populations of Entomobrya nicoleti sampled across Denmark, and detected substantial differences in heat and cold tolerance over a much smaller geographical range. Thermal tolerance showed weak associations with environmental variation and no consistent geographic structure, providing no evidence for phenotypic isolation-by-distance. Phenotypic differentiation was not associated with neutral genetic differentiation (FST), indicating decoupling from neutral population genetic structure. To understand what has shaped these phenotypic differences, we conducted genotype-phenotype-environment association analyses by integrating whole-genome resequencing data and a candidate-gene approach. By combining genomic, phenotypic, and environmental data, this study provides a framework for assessing the drivers of stress tolerance variation and the potential for local adaptation in increasingly fragmented low-dispersal soil arthropods.

[A350] Non-invasive genomic sampling uncovers novel connectivities and origins of confiscated gorillas

Irune Ruiz-Gartzia[1], Harvinder Pawar[1,2], Marina Alvarez-Estape[1], Joseph D. Orkin[1,3,4], Pol Alentorn-Moron[1], Sandra Ruibal-Puertas[1], Harrison J. Ostridge[5], Claudia Fontsere[1,6], Sebastian Cuadros-Espinoza[1], Kirsten Gilardi[7], Julius Nziza[7], Richard Muvunyi[8], Ulrich Maloueki[9,10], Damien Caillaud[11], Neetha Iyer[11], Urban Ngobobo-As-Ibungu[12], Escobar Binyinyi[12], Etienne François Akomo-Okoue[13], Augustin K. Basabose[14], Kamungu Sebulimbwa[14], Alfred Ngomanda[13], Juichi Yamagiwa[15], Miho Inoue-Murayama[16], Yuji Takenoshita[17], Shiho Fujita[18], Barbora Pafco[19], Terence Fuh[20,21], Nikki Tagg[22], Donald Mbohli[22,23], Emmanuel Ayuk Ayimisin[24], Emma Bailey[24], Mattia Bessone[24,25,26], Tobias Deschner[24,27], Paula Dieguez[24,28], Emmanuel Dilambaka[29], Andrew Dunn[29], Anne-Celine Granjon[24,30,31], Josephine Head[24,32], Veerle Hermans[22], Inaoyom Imong[29], Kathryn J. Jeffery[33,34], Kevin Lee[24,35,36,37], Giovanna Maretti[24], David Morgan[38], Emily Neil[24], Christopher Orbell[39,33,13], Martha M. Robbins[40], Crickette Sanz[41,42], Nadege Wange Njomen[43], Jacob Willie[22,44], Barthélémy NGOUBANGOYE[45], Larson Boundenga[46], Franck Prugnolle[47,48], Olivia Cheronet[49,50], Ron Pinhasi[49,50], Ivo Glynne Gut[51,52], Marta Gut[51,52], Karine A. Viaud-Martinez[53], Virginie Rougeron[47,48], Bethan Morgan[54], Christophe Boesch[24,55], Hjalmar S. Kühl[28,56], Mimi Arandjelovic[40,30], Eiji Inoue[57], Katerina Guschanski[58,59], Klara Petrzelkova[19,60], Martine Peeters[61], Magdalena Bermejo[62,63], Germán Illera Basas[63], Torsten Bohm[10], Tierra Smiley Evans[7,64], Javier Prado-Martinez[1], Martin Kuhlwilm[49,50], Esther Lizano[1,65,66], Prof. Tomas Marques Bonet[1,51,65,66,67]

[1]IBE, Institute of Evolutionary Biology (UPF-CSIC), Department of Medicine and Life Sciences, Universitat Pompeu Fabra, Barcelona, Spain, [2]Department of Biology, University of Copenhagen, Copenhagen, Denmark, [3]Département d'anthropologie, Université de Montréal, Montréal, Canada, [4]Département de sciences biologiques, Université de Montréal, Montréal, Canada, [5]UCL Genetics Institute, Department of Genetics, Evolution and Environment, University College London, London, UK, [6]Center for Evolutionary Hologenomics, The Globe Institute, University of Copenhagen, Copenhagen, Denmark, [7]Gorilla Doctors, Karen C. Drayer Wildlife Health Center, One Health Institute, University of California Davis, School of Veterinary Medicine, Davis, USA, [8]Rwanda Development Board, Kigali, Rwanda, [9]Faculty of Sciences and Technologies, Life Sciences, Kinshasa University, Kinshasa, Republic Democratic of the Congo, [10]African Parks, Odzala-Kokoua National Park, Brazzaville, Republic of Congo, [11]Department of Anthropology, University of California, Davis, USA, [12]The Dian Fossey Gorilla Fund International, Kinshasa, DRC, [13]Institut de Recherche en Ecologie Tropicale (IRET/CENAREST), Libreville, Gabon, [14]Centre de Recherche en Sciences Naturelles de Lwiro, Bukavu, RD Congo, [15]Research Institute for Humanity and Nature, , , [16]Wildlife Research Center of Kyoto University, Kyoto , Japan, [17]Department of Zoology, Faculty of Science. Okayama University of Science, Okayama , Japan, [18]Center for General Education, Kagoshima University, Kagoshima, Japan, [19]Institute of Vertebrate Biology, Czech Academy of Sciences, Brno, Czech Republic, [20]WWF DRC country programme, Gombe, DRC, [21]World Wildlife Fund, Avenue des Martyrs, Bangui, Central African Republic, [22]KMDA, Centre for Research and Conservation, Royal Zoological Society of Antwerp, Antwerp, Belgium, [23]Association pour la Protection des Grands Singes (APGS), Nkol-Eton Yaounde , Cameroon, [24]Max Planck Institute for Evolutionary Anthropology, Leizpig, Germany, [25]University of Konstanz, Centre for the Advanced Study of Collective Behaviour, Konstanz, Germany, [26]Department of Animal Societies, Max Planck Institute of Animal Behavior, Konstanz, Germany, [27]Institute of Cognitive Science, University of Osnabrück, Osnabrück, Germany, [28]Senckenberg Museum für Naturkunde Görlitz, Görlitz, Germany, [29]Wildlife Conservation Society (WCS), New York , USA, [30]German Centre for Integrative Biodiversity Research (iDiv), Leipzig, Germany, [31]Leipzig University, Leipzig, Germany, [32]Arcus Foundation, Cambridge, UK, [33]School of Natural Sciences, University of Stirling, Stirling, UK, [34]AMAP, University of Montpellier, Montpellier, France, [35]School of Human Evolution and Social Change, Arizona State University, Tempe, USA, [36]Institute of Human Origins, Tempe, USA, [37]Ngogo Chimpanzee Project, Makerere University Biological Field Station, Fort Portal, Uganda, [38]Lincoln Park Zoo, Lester E Fisher Center for the Study and Conservation of Apes, Chicago, USA, [39]Panthera, New York, USA, [40]Department of Primate Behavior and Evolution, Max Planck Institute for Evolutionary Anthropology, Leizpig, Germany, [41]Washington University in Saint Louis, Department of Anthropology, St. Louis, USA, [42]Congo Program, Wildlife Conservation Society,

Brazzaville, Republic of Congo, [43]WWF Cameroon Country Office, Yaoundé, Cameroon, [44]Terrestrial Ecology Unit (TEREC), Department of Biology, Ghent University (UGent), Ghent, Belgium, [45]Centre de Primatologie, Centre Interdisciplinaire de Recherches Médicales de Franceville (CIRMF), Franceville, Gabon, [46]Unité de Recherche en Ecologie de la Santé (URES), Centre Interdisciplinaire de Recherches Médicales de Franceville (CIRMF), Franceville, Gabon, [47]International Research Laboratory, REHABS, CNRS-Université Lyon 1-NMU, George, South Africa, [48]Sustainability Research Unit, Nelson Mandela University, George, South Africa, [49]Department of Evolutionary Anthropology, University of Vienna, Vienna, Austria, [50]Human Evolution and Archaeological Sciences (HEAS), University of Vienna, Wien, Austria, [51]Centro Nacional de Análisis Genómico (CNAG), Barcelona, Spain, [52]Universitat de Barcelona (UB), Barcelona, Spain, [53]Illumina Laboratory Services, Illumina. Inc, San Diego, USA, [54]San Diego Zoo Wildlife Alliance, San Diego, USA, [55]Wild Chimpanzee Foundation, , , [56]International Institute Zittau, Technische Universität Dresden, Zittau, Germany, [57]Department of Biology, Faculty of Science, Toho University, Chiba, Japan, [58]Institute of Ecology and Evolution, School of Biological Sciences, University of Edinburgh, Edinburgh, UK, [59]Animal Ecology, Department of Ecology and Genetics, Uppsala University, Uppsala, Sweden, [60]Institute of Parasitology, Biology Centre, Czech Academy of Sciences, Branisovska , Ceske Budejovice, [61]TransVIHMI, Université de Montpellier, INSERM, IRD, Montpellier, France, [62]Department of Ecology and Environmental Sciences, University of Barcelona, Barcelona, Spain, [63]Sabina Plattner African Charities (SPAC) NPC, Cape Town, South Africa, [64]Department of Integrative Biology & Infectious Diseases and Vaccinology Division, School of Public Health, University of California Berkeley, Berkeley, USA, [65]Institut Català de Paleontologia Miquel Crusafont (ICP-CERCA), Universitat Autònoma de Barcelona, Cerdanyola del Vallès, Spain, [66]Unidad de Paleobiología, ICP-CERCA, Unidad Asociada al CSIC por el IBE UPF-CSIC, Barcelona, Spain, [67]Catalan Institution of Research and Advanced Studies (ICREA), Barcelona, Spain

Gorillas are a group of African great apes with two species and four subspecies that are currently critically endangered or endangered. Previous studies that analysed the genetics of wild gorillas from non-invasive samples, such as faeces or hair, analysed short mitochondrial or nuclear markers, which may not reflect the wider nuclear genome. Recent technical advances in target capture hybridisation, enrich the endogenous DNA content of non-invasive samples, allowing contiguous genomic regions to be sequenced. Here, we generated georeferenced genetic data from faecal and hair samples of 280 wild gorillas, sampled from three of the four gorilla subspecies, across large parts of their present-day distributions. With this expanded representation of gorilla genetic diversity in the wild, we detected three population clusters in western lowland gorillas, with the Sangha River and its affluents acting as significant barriers to gene flow. We reconstructed patterns of past population connectivity between western lowland gorillas in the north-eastern distribution range and Cross River gorillas, which may have been facilitated by a migration corridor also used by the Central and Nigeria-Cameroon chimpanzee subspecies. Finally, we predicted the geographic origins of wild-born gorillas, achieving a mean prediction error of 65 km, with a population-level resolution for mountain gorillas and some populations of western lowland gorillas. Our work characterises fine-scale population structure in western lowland gorillas, which will be informative for future conservation strategies. This proof of concept in predicting geographic locations of wild gorillas, will be useful for future applications to geolocalise trafficked or rescued gorillas.

[A351] Genomics of Brazilian howler monkeys reveals cryptic population structure and adaptation to folivory

Katherine McVay[1], Bide Chen[1], Erick Figueroa-Ildefonso[1], Jeferson Freitas[2], Luana Portela[2], Dr Bernard Kim[3], Ellie Armstrong[4], Victor Y. Guimarães[5], Ligia S. L. Silveira da Mota[5], Cauê Monticelli[6], João Valsecchi[7], Fernando A. Perini[8], Alcides Pissinatti[9], Patricia Domingues de Freitas[2], Amy Goldberg[10]
[1]Duke University, , United States, [2]Federal University of São Carlos, , Brazil, [3]Princeton University, , United States, [4]University of California, Riverside, , United States, [5]São Paulo State University, , Brazil, [6]São Paulo Wildlife Coordination, , Brazil, [7]Mamirauá Sustainable Development Institute, , Brazil, [8]Federal University of Minas Gerais, , Brazil, [9]Primatology Center of Rio de Janeiro, , Brazil, [10]University of California, Los Angeles, , United States

Genomics of nonhuman primates has expanded, yet most studies consider a single species per genus. Here, we tested for shared and unique evolutionary histories between species within a primate genus to fill this temporal gap. Howler monkeys (genus Alouatta) are the widest-spread neotropical primate radiation. In parts of their range, deforestation and human encroachment threaten these vulnerable species. We assembled 6 de novo long-read reference genomes (n50 ~15Mb), and sequenced 88 howler monkeys (average coverage ~11X) to reconstruct the evolutionary history of four major howler monkey species groups from across Brazil. Genomic population structure contradicts the current IUCN taxonomic classification. We find environmental differences in demography, including an increased proportion of long ROH and decreasing population size estimates in Atlantic Forest howler monkeys, potentially resulting from the increased habitat fragmentation compared to the Amazon rainforest. Finally, we inferred adaptation on multiple timescales by calculating dN/dS on the branch leading to all howler monkeys and compared signatures of recent positive selection between two howler species using G1 statistics. Both timescales were enriched for genes involved in xenobiotic detoxification, likely related to howlers' highly folivorous diet, including a new potentially convergent derived nonsynonymous mutation shared with other folivorous colobine monkeys. Notably, only the species that experienced recent outbreaks of yellow fever shows evidence for adaptation at yellow fever virus entry receptors. The identification of shared and unique demographic and adaptive trajectories demonstrates the utility of intragenus comparative genomics to identify the genetic basis of characteristic phenotypes and species-specific conservation concerns.

[A352] Estimates of long-term effective population size reveal the rapid decline of endangered species

Loveday Lewin[1], Adam Eyre-Walker[1]
[1]University Of Sussex, , United Kingdom

As anthropogenic pressures accelerate species declines and genomic data becomes increasingly accessible, interest has grown in the use of genetic metrics for biodiversity monitoring and population risk assessments. Among these, the effective population size (Ne) is a key parameter that provides information about the genetic condition of a population. While conservation practice typically focuses on contemporary Ne estimates, which capture recent population dynamics, long-term Ne reflects the harmonic mean population size over thousands of years. This longer-term perspective could offer insights into the evolutionary forces that have shaped present-day genetic diversity. We estimated long-term Ne for 60 eukaryotic species using neutral genetic diversity and experimentally derived mutation rates, and compared these values to census population size (Nc) estimates. Ne and Nc were positively correlated, largely following the expected trend of Ne < Nc. Notably, however, this relationship was inverted in several endangered vertebrates, where long-term Ne estimates exceeded current Nc. This pattern likely indicates recent, rapid population declines not yet reflected in the genetic record – a phenomenon known as genetic drift debt. Recognising this signal could provide early warnings of population collapse and inform conservation interventions.

[A353] Between isolation and invasion: the genomic fate of Antillean iguanas

dr. Mirte Bosse[1,2], Michou Weimar[2,3], Merel Hogewind[1], Dr Kat Stewart[4], Dr Catherine Malone[7], Dr Adolphe Debrot[2], Matthijs van den Burgh[5,6]
[1]Vrije Universiteit Amsterdam , Amsterdam, Netherlands, [2]Wageningen University & Research, Wageningen, Netherlands, [3]Royal Rotterdam Zoological & Botanical Gardens, Rotterdam, Netherlands, [4]Leiden University, Leiden, Netherlands, [5]IUCN SSC Iguana Specialist Group, Gland, Switzerland, [6]Burg Biologica, The Hague, Netherlands, [7]Utah Valley University, Orem, United States of America

Loss of genetic diversity poses a major threat to the persistence and adaptive potential of island endemics, where small population sizes and isolation amplify the impacts of environmental change and anthropogenic disturbance. In the Lesser Antilles, multiple iguana lineages are increasingly threatened by habitat loss and inbreeding, with an additional threat from hybridization with other iguana lineages reaching the islands. Together, they provide a powerful system to study the consequences of genetic erosion.

By generating a high-quality de novo assembly for the Saba iguana, we identified unique structural variants. Using whole-genome sequencing from 100 individuals, we investigated population structure, demographic history, genetic diversity, selection, and hybridization across four iguana taxa (Iguana delicatissima, I. iguana, I. insularis, and I. melanoderma), including the critically endangered Saba green iguana.

Population genomic analyses revealed deep divergence among taxa, with endemic island species exhibiting reduced nucleotide diversity, elevated inbreeding, increased genetic load, and strong population structure. In contrast, the widespread I. iguana retained higher genetic diversity and showed evidence of cryptic population subdivision. Genome-wide scans identified signatures of selection across all taxa, consistent with local adaptation.

Hybridization analyses based on SNP ancestry confirmed the presence of first-generation hybrids but found no evidence for widespread cryptic introgression, suggesting that current management actions are limiting genome-wide admixture. However, severe inbreeding in some island populations highlights a conservation dilemma: while hybridization threatens endemic genomic integrity, limited gene flow could mitigate inbreeding depression. Together, these results demonstrate how genomic approaches can inform conservation strategies for genetically eroded populations.

[A354] Do repeat invasions have the same genetic signature: Using population genetics to manage an active invasion.

Neve Kelly[1], Mali Woods[1], Dr Katarina Stuart[1,2], Dr Susan Campbell[3], Prof. Lee Ann Rollins[1]
[1]Evolution & Ecology Research Centre, University of New South Wales, Sydney, Australia, [2]Applied BioSciences, Faculty of Science and Engineering, Macquarie University, Sydney, Australia, [3]Department of Primary Industries and Regional Development, Albany, Australia

Studying evolution in newly formed range-edge populations can inform our understanding of their likelihood of persistence but is challenging because these populations are typically in disequilibrium, with rapidly shifting demographics. Biological invasions provide an excellent model for studying how genetic diversity, relatedness, and sex ratios shift during population formation because multiple invasions of the same species can be compared to identify common patterns. We used temporal genomic data to characterise population dynamics at the expanding range edge of the common starling (Sturnus vulgaris) in Western Australia. This area has experienced multiple, independent starling incursions, observed since the 1970s, some of which have been thought to be extirpated. Using reduced-representation sequencing data (DArT-seq) from 229 individuals sampled over the past two decades from Australia's range core, intermediate, and expanding range edge populations, we reconstructed the genomic signatures of repeated founder events. These data illustrate predictable population genomic patterns following invasion such as short-term elevated relatedness and pronounced sex-ratio shifts. They also demonstrate the efficacy of control operations conducted to manage range-edge incursions.

Our results demonstrate how temporal population genomics can reveal rapid molecular change in newly formed, low-diversity populations, providing a rare window into the evolutionary processes that shape establishment success or failure. This work highlights the value of genomic monitoring to detect early signals of changes to population dynamics and for informing management responses in dynamically changing systems. Although this study utilises a well-studied invasion, such patterns may also be useful for monitoring the health of conserved, diminishing populations.

[A355] Investigating the decline of a threatened Orthopteran combining ecological niche modeling and genomic analyses

Nina Casillas[1,2], Dr. Marianna Simões[2], Dr. Oliver Hawlitschek[3], Dr Eckart Stolle[1], Astrid Böhne[1], Prof. Dr. Martin Husemann[4]
[1]Leibniz Institute for the Analysis of Biodiversity Change (LIB), Bonn, Germany, [2]Senckenberg Research Institute and Natural History Museum Frankfurt, Frankfurt, Germany, [3]University of Zurich, Department of Evolutionary Biology and Environmental Studies, Zurich, Switzerland, [4]Staatliches Museum für Naturkunde Karlsruhe, Karlsruhe, Germany

Climate change and anthropogenic factors heavily contribute to the decline of biodiversity, but nothing beats habitat loss through intensive agricultural practice, urbanization and other anthropogenic land use practices. Although protected areas hold great importance in the conservation of biodiversity, the discrepancy between protected sites and sites which hold a positive conservation status reveals the importance of targeted, systematic analyses of local species which can then be conveyed to policy makers, revitalizing the full spectrum of habitat restoration and biodiversity conservation. Ecological niche modelling and species distribution modelling have emerged as valuable tools in this context to characterize key components within a species' habitat. With the inclusion of genetic and genomic data we can uncover contributors of decline and piece together the process leading to the state of the species observable today. Here, we investigate the decline of the threatened Orthopteran, Gampsocleis glabra, a species known for inhabiting steppe and heathland which has been continuously in decline and regionally extinct across Europe. Through the incorporation of population genomics and ecological niche modelling, we aim to (i) determine key factors influencing the distribution of G. glabra, (ii) estimate the distribution of G. glabra in the paleoclimate comparative to now, and (iii) assess the genomic health of populations along a gradient of ecological suitability while gleaming insight from demographic reconstruction.

[A356] Global genetic structure and diversity in the widespread red fox

Paula Gardner[1], Róisín Ferguson Ferguson[1], Sophie Preckler-Quisquater[2], Dr. Cate B. Quinn[2], Dr Alice Godden[1], Dr. Mark J. Statham[2], Dr. Rafał Kowalczyk[3], Guo-dong Wang[4], Dr. Raquel Godinho[5], Dr. Jan M. Wójcik[3], Dr. Benjamin Sacks[2], Dr Anders Bergström[1]
[1]University Of East Anglia, Norwich, United Kingdom, [2]Mammalian Ecology and Conservation Unit, University of California, Davis, Davis , United States of America, [3]Mammal Research Institute, Polish Academy of Sciences, Białowieża , Poland, [4]State Key Laboratory of Genetic Resources and Evolution, Kunming Institute of Zoolog, Kunming, P.R.China, [5]CIBIO/InBIO University of Porto, Vairão, Portugal

Many species around the world are suffering population decline, but what does population history and structure look like for a species that thrives in the Anthropocene? The red fox (Vulpes vulpes) is likely the land mammal with the largest natural range, living and expanding across diverse environments. To understand the global genetic diversity and history of this successful species, we sequenced 34 red fox genomes across their Northern Hemisphere range to an average coverage of 32x. We find that population structure largely reflects an east-west longitudinal gradient from Europe to North America, with foxes from the Middle East and North Africa clustering separately in PCA and ADMIXTURE analyses. Heterozygosity levels through Eurasia are consistently high, likely in the highest range among mammals. Levels in North America are roughly two-fold lower, likely reflecting population bottlenecks corresponding to migrations into the continents,which MSMC analyses indicate occurred 2-300,000 years ago. We are currently expanding the dataset through additional sequencing, and will perform analyses of divergence times, strength of population differentiation, and possible introgression from other fox species. Understanding the varied evolutionary history of this highly successful carnivore can provide a counterpoint to understand the histories and trajectories of declining species.

[A357] From Inactive Dimers to Functional Tetramers: Evolution of Catalysis in DfrB Enzymes

Samy Faraj[1,2,3], Margaux Raboisson[5], Roxana Tarabuta[2,3,4], Stella Cellier-Goetghebeur[2,3,4], Joelle Pelletier[1,2,3,4]
[1]Chemistry Department, University of Montreal, Montreal, Canada, [2]PROTEO, The Québec Network for Research on Protein, Function, Engineering and Applications, Montreal, Canada, [3]CGCC, Center in Green Chemistry and Catalysis, Canada, Montreal, Canada, [4]Biochemistry Department, University of Montreal, Montreal, Canada, [5]Université Catholique Louvain la Neuve, Louvain, Belgique

Type B dihydrofolate reductases (DfrB) are homotetrameric enzymes that confer high-level resistance to the antibiotic trimethoprim (TMP), that was introduced in the early 1960s. In contrast to canonical FolA dihydrofolate reductases, DfrBs assemble into a distinctive β-barrel architecture: four identical SH3-based protomers form an obligate tetrameric assembly that creates a single functional active site. Indeed, the active site exists only when the tetramer is formed, generating a single, central catalytic pore. This unusual organization underlies the innate insensitivity of DfrBs to TMP; however, the precise structural determinants guiding pore formation and stabilizing the dimer–dimer interfaces remain poorly defined.

To address these gaps, we combined structural modeling, evolutionary analysis and targeted mutagenesis to identify interface residues critical for pore stabilization and modulation of enzymatic activity. Our results reveal how specific inter-protomer contacts govern tetramer assembly and catalytic competence. These findings clarify the mechanistic basis by which tetramerization enables DfrB function.

In addition, we are investigating the evolution of ancestrally inactive dimeric homologs to the active, tetrameric DfrB enzymes. Using bioinformatic analyses and directed evolution, we hypothesize that functional homotetramers can be selected from noncatalytic dimeric precursors by evolving interface residues. This work sheds light on how ancient SH3 domains having binding, but no catalytic, function could be co-opted into a homotetrameric enzyme conferring TMP resistance. More broadly, a deeper understanding of this evolutionary transition may guide the development of novel antimicrobial strategies targeting the dimer–dimer interface of DfrB enzymes.

[A358] Hybridization poses challenges for the conservation of Iberian endemic freshwater fish species

Sofia Mendes[1,2], Estêvão Faustino[2], João Moreno[2,3], Silvia Perea[4], Ignacio Doadrio[4], Carla Sousa Santos[3], Philine G. D. Feulner[1,5], Vitor C. Sousa[2]

[1]Department of Fish Ecology and Evolution, Eawag – Swiss Federal Institute of Aquatic Science and Technology, Kastanienbaum, Switzerland, [2]CE3C – Centre for Ecology, Evolution and Environmental Changes & CHANGE – Global Change and Sustainability Institute, Department of Animal Biology, Faculty of Sciences of the University of Lisbon, Lisbon, Portugal, [3]MARE – Marine and Environmental Sciences Centre & ARNET – Aquatic Research Network. University of Lisbon, Lisbon, Portugal, [4]Museo Nacional de Ciencias Naturales, Madrid, Spain, [5]Institute of Ecology and Evolution, University of Bern, Bern, Switzerland

Freshwater and diadromous fish are one of the most threatened vertebrate groups due to habitat degradation, introduction of invasive species and increasingly strong effects of climate change. Within the Mediterranean Basin Biodiversity Hotspot, the Iberian Peninsula is home to over 50 endemic species of freshwater and diadromous fish. Despite most of them being threatened, their current genomic diversity remains largely unknown. We focused on the Iberian chubs (Squalius spp), a group of primary freshwater fishes with allopatric distributions across Iberian rivers and conservation status ranging from vulnerable to critically endangered. We obtained whole genome resequencing data from 177 individuals covering the distribution of eight Iberian chub species. We first investigated patterns of genetic diversity and differentiation using low coverage whole genome data from 125 individuals. This revealed high levels of intra ($0 \leq F_{ST} \leq 0.61$) and interspecific ($0.44 \leq F_{ST} \leq 0.88$) genetic differentiation. Southern species with smaller ranges showed lowers levels of genetic diversity, highlighting their vulnerability. Despite present-day allopatric distributions, hybridization is pervasive in the group, as we detected three, possibly four, cases of hybridization spanning different geographical and time scales. We then reconstructed the demographic history of these species and admixed populations using a dataset of 60 individuals sequenced at higher coverage. We also performed controlled laboratory experiments to assess their responses to increases in water temperature. Our results support the definition of conservation units below the species level for the Iberian chubs and highlight the challenges associated with preserving admixed populations, as they are often excluded from conservation policy yet represent unique biodiversity.

[A359] The genetic diversity of seagrasses: environment, reproductive system and the nearly neutral theory

Xinyue Teng[1]
[1]Zhejiang University, , China

Seagrasses occupy a vast geographical area and provide important ecosystem services. They are characterized by a large diversity of reproductive systems and environments. Some species are dioecious while others are hermaphrodites or monoecious. Some are fully marine and others live in freshwater environments. Hence, seagrasses are a perfect group of species to assess the relative importance of reproductive system and environment in shaping genetic diversity and test how these factors affect predictions of the Nearly Neutral Theory of molecular evolution (NNT). In the present study we sequenced the transcriptome of 127 individuals from 27 species of the order Alismatales. A well-supported phylogeny confirmed that evolutionary transition from freshwater to marine life occurred multiple times. Species from freshwater environments were, on average, more genetically diverse than those from marine environments. This difference was, in large part, due to the low diversity of dioecious species in marine environments. The latter could be due to pollen limitation and ensuing Allee effect and/or biased sex ratio. Selection was more efficient in freshwater environments than in marine ones, as would be expected under the nearly neutral theory (NNT) and the quantitative fit between data and the NNT prediction was surprisingly good.

[A360] Mutation rate estimate and population genomic analysis reveals decline of koalas prior to human arrival

Toby Kovacs[1], Dr Nicole Foley[2], Dr. Luke Silver[1], Dr Elspeth McLennan[1], Dr William Murphy[2], Prof. Carolyn Hogg[1], Dr Simon Ho[1]
[1]The University Of Sydney, , Australia, [2]Texas A&M University, , USA
The koala (Phascolarctos cinereus), an iconic Australian marsupial, has experienced substantial historical and contemporary population declines. Identifying the drivers of these declines has been hindered by limited genomic data and by uncertainty regarding the koala mutation rate. Here, we provide the first direct estimate of the koala mutation rate, based on four parent-offspring trios, yielding a mean of 6.12×10^{-9} mutations per base pair per generation (95% confidence interval: $5.03–7.45\times10^{-9}$). Using this estimate of the rate, we reconstructed the demographic history of koalas using 458 whole-genome sequences sampled across their entire range. Our results refine the estimated timing of past changes in population size, suggesting a large decline beginning ~100 kya, before the arrival of modern humans in Australia. The koala population then split into five genetic populations 6–30 kya, which are now spread across >3500 km on the east coast of Australia. We also use the koala mutation rate to infer recombination maps for each population, confirming lower recombination rates in marsupials than in eutherian mammals. These findings provide critical insights into the evolutionary history of koalas, while highlighting the impact of using species-specific evolutionary rates in the inference of demographic histories and recombination landscapes. Our estimates of the genome-wide mutation rate and population-specific recombination maps for koalas provide valuable resources for future evolutionary and conservation analyses of marsupials.

[A361] Recent insights into the evolutionary genomics of the critically endangered aye-aye (Daubentonia madagascariensis)

Vivak Soni[1], Susanne Pfeifer[1], Jeffrey Jensen[1]
[1]Arizona State University, Tempe, United States

Endemic to the island of Madagascar, the aye-aye (Daubentonia madagascariensis) is one of the 25 most endangered primate species in the world, maintaining amongst the lowest genetic diversity of any primate measured to date. Rapid deforestation in recent decades in Madagascar has contributed to severe population decline in aye-ayes, with fewer than 1,000-10,000 individuals estimated to remain in the wild according to the IUCN.

Although the characterization of patterns of genetic variation within aye-aye populations is of urgent conservation importance, much remains unknown about these strepsirrhines, in no small part due to their nocturnal and arboreal lifestyle and large geographic ranges. The recent publication of a high-quality, annotated reference genome assembly, together with whole-genome population-level sequencing data has facilitated a raft of recent research elucidating the evolutionary genomics of aye-ayes. These recent studies shed new light on the selective and demographic history of the species, sex-specific pedigree-based and fine-scale divergence-based mutation rates, as well as sex-specific pedigree-based and fine-scale polymorphism-based crossover (CO) and noncrossover (NCO) recombination rates. These new findings are surveyed in this talk, as well as their implications for future conservation management of this critically endangered species.

[A362] Unraveling lost diversity of Arctic marine endemics - investigating ancient belugas and bowhead whales from southern Scandinavia

Wenxi Li[1], Dr. Mikkel Skovrind[2], Paul Szpak[3], Michael Westbury[4], Professor Eline Lorenzen[1]
[1]Globe Institute, University Of Copenhagen, Copenhagen, Denmark, [2]Lund University, Department of Biology, , Sweden, [3]Trent University, Department of Anthropology, , Canada, [4]Technical University of Denmark, Department of Health Technology, , Danmark

The global environment has changed dramatically over the past few decades, especially in the Arctic, and Arctic sea ice is predicted to decline by up to 94% by 2050. By losing their habitats, ice-associated marine mammals are highly vulnerable to changes in sea ice conditions. Historically, the Arctic has undergone dramatic climate shifts across the Pleistocene-Holocene transition, including rapid warming, sea-level rise, and a major decline in sea-ice extent. Yet, how climate change has affected Arctic marine mammals on a long-term scale is still not well understood.

In this study, we focus on southern Scandinavia, a region that experienced rapid deglaciation in the early Holocene and lies far south of the modern distribution of Arctic cetaceans. We employ whole-genome shotgun sequencing and mitogenome capture to recover genomic data from more than 100 ancient beluga whale (Delphinapterus leucas) and bowhead whale (Balaena mysticetus) specimens dating from the Pleistocene-Holocene transition. Both species occupied southern Scandinavian waters during early deglaciation but subsequently disappeared from the region, providing a rare opportunity to investigate eroded and regionally extinct populations.

By integrating the genomic data with isotopic analyses, we aim to investigate how the rapid warming and habitat reorganisation can drive genetic turnover and lineage loss. This work provides one of the few ancient studies of Arctic marine mammals and may provide valuable insights for understanding how global warming and diminishing Arctic sea ice might impact future marine mammal conservation efforts.

[A365] It's A Small World After All: The Unique Mutation Rate and Lifestyle of a Naked RNA Virus

Alyssa Cenzano[1,2], Stephan Baehr[2], Cooper Call[1,2], Methila Pooja[1,2], Samuel Miller[2], Dr. Michael Lynch[1,2]

[1]School of Life Sciences Arizona State University, Tempe, United States, [2]Biodesign Institute Center for Mechanisms of Evolution, Tempe, United States

Mutation rates influence rates of evolution, from adaptation to disease emergence and co-evolution. With the advent of high-throughput sequencing, mutation rates have been precisely measured for most living organisms; and yet, our knowledge of viral mutation rates remains almost entirely based upon reporter constructs, estimates known to vary from modern methods by an order of magnitude or more. Existing mutation rate estimates describe a negative correlation between genome size and mutation rate, and though doubtless this relationship persists in viruses, its magnitude and variance remain poorly characterized. An outlier relative to current examined variance is Narnavirus, a vertically inherited, non-pathogenic replicon encoding a single gene–an RNA polymerase–with a genome size of only 2.5 kilobases. Employing a mutation accumulation experiment, a widespread technique in evolutionary biology that is underutilized in virology studies, we estimate Narnavirus's mutation rate to be 5×10^{-7} substitutions per nucleotide per replication, an unusually low mutation rate estimate for ssRNA viruses with small genome sizes. Furthermore, the dN/dS ratio is calculated to be 0.38, indicating the presence of strong negative selection. These findings suggest that the inferred relationship between genome size or type and mutation rate is by no means absolute, and that viral life cycles may play a role in the evolution of mutation rates.

[A366] Mutation rate evolution in cetaceans: understanding drivers of variability and observation bias in estimates of genomic mutation rate

Amy Van Cise[1], Sophia Garrote, Dr. Phillip Morin
[1]University Of Washington, Seattle, United States

Genomic mutation is the fundamental process driving evolutionary processes that result in contemporary species, subspecies, and locally adapted populations. Thus, genomic studies of divergence, speciation, and evolutionary demographics rely on accurate estimates of mutation rate. Historically, mutation rates have been estimated using a small portion of the genome and/or averaged among multiple species within a family or order, but growing evidence indicates potentially high variability in mutation rate both across the genome and among species. Here, we examine variability in mutation rate within cetaceans based on whole genome sequence data mapped to reference-quality genomes, and from a range of pipeline parameters in order to determine which factors have the greatest effect on variability in substitution rate or its estimation. Our results indicate that mutation rate has varied over evolutionary time and across the genome in cetaceans. Notably, we show that generational mutation rate increases with maximum lifespan while annual mutation rate increases with maximum lifespan and decreases with generation time. Mutation rate also decreases with chromosome size. Similarly, we found significant effects of pipeline parameter choice on mutation rate, which is most affected by the choice of reference genome used in the alignment pipeline and choice of genomic region (i.e. chromosome). These results indicate the importance of thoughtful selection of mutation rate used in cetacean genomic studies, in order to ensure accurate downstream analyses of evolutionary processes

[A367] Mutation and recombination in coppery titi monkeys (Plecturocebus cupreus)

Cyril Versoza[1], Karen Bales[2,3,4], Jeffrey Jensen[1], Susanne Pfeifer[1]
[1]Center for Evolution and Medicine, School of Life Sciences, Arizona State University, Tempe, USA, [2]Department of Psychology, University of California, Davis, Davis, USA, [3]California National Primate Research Center, Neuroscience and Behavior Division, Davis, USA, [4]Department of Neurobiology, Physiology, and Behavior, University of California, Davis, Davis, USA

Mutation and recombination shape population genetic diversity and influence a species' potential adaptive response to environmental changes; yet, despite their importance, our current understanding of these processes is based on studies performed in a relatively small number of organisms. Utilizing novel high-coverage genomic data from parent-offspring trios and multi-sibling families, we provide the first estimates of the rates and patterns of both germline mutation and meiotic recombination in coppery titi monkeys (Plecturocebus cupreus) — a socially monogamous, pair-bonded primate that serves as an important model in behavioral research. Coppery titi monkey males exhibit long reproductive lifespans, afforded by long-term pair bonding in the species' monogamous mating system, and our results demonstrate that, while sex-averaged mutation rates fall within the range of that of other primates, individuals reproducing later in life exhibit both a strong paternal age effect and male mutation bias. Moreover, sex-averaged crossover rates in coppery titi monkeys are comparable with those of other primates; however, no significant difference in recombination rates was observed between the sexes, despite a pronounced maternal age effect in the species. Consistent with haplorrhines, crossover and non-crossover recombination in this platyrrhine are frequently localized at PRDM9-mediated hotspots, characterized by a 15-mer binding motif with substantial similarities to the degenerate motif found in humans. Taken together, these results thus not only further our understanding of how genetic variation is shaped in the coppery titi monkey genome, but they will also be beneficial in future studies of this primate system in biomedical, behavioral, and evolutionary research.

[A368] Variation in mutation and recombination rate within and among species of Drosophilidae

Dr. Yiguan Wang[1,2], Dhobasheni Newman[1], Rashidatu Abdulazeez[1,3], Susan Johnston, Peter Keightley[1], Darren Obbard[1]
[1]University Of Edinburgh, Edinburgh, United Kingdom, [2]Center of Excellence in Molecular Plant Science, Shanghai, China, [3]Ahmadu Bello University, Zaria, Nigeria

Germline mutation and recombination rates are fundamental parameters for population and evolutionary genetics, determining levels of diversity and the efficacy of natural selection. Here, we use parent-offspring sequencing to directly estimate de novo germline mutation and recombination rates for multiple independent unrelated sibships across 20 species in the subfamily Drosophilinae. We use (phylogenetic) mixed models to decompose the variance in rate into within- and among-species components, to test for the role of parental sex, and to identify potential confounding factors, such as the age and origin of flies. We find that mutation rate differed between the sexes and between wild-caught and laboratory-reared flies, and varies among individuals. However, we were unable to detect any among-species variance in mutation rate. As expected, we detected substantial variation in recombination rate within and among species.

[A369] The Human Mutation Spectrum Is Not Neutral

David Castellano[1], Ryan Gutenkunst
[1]University Of Arizona, Tucson, United States

Mutation rates and fitness effects are often treated as independent, but mutation rates are variable and evolve under indirect selection. For example, human European populations experienced a transient increase in the 3-mer mutation rate TCC -> TTC in the past 20,000 years. To quantify indirect selection on mutation spectra, we developed an approach to estimate the distribution of fitness effects (DFE) of nonsynonymous 3-mer mutation types, by analyzing pairs of complementary mutation types to account for GC-biased gene conversion and ancestral state misidentification. We then applied this approach to all 96 possible 3-mer mutation types in humans, using data from the 1000 Genomes Project. We found widely varying DFEs among mutation types and that inferences from a few hundred genomes could predict pN/pS in samples of tens of thousands of genomes. Our DFE estimate for TCC -> TTC mutations is consistent with recent theoretical predictions by Milligan, Amster, and Sella of scenarios under which a moderate number of modifier loci could yield population-specific transient bursts of a specific mutation type.

[A370] Predicting the fate of mutation rate modifiers that shift the mutation bias

Diego Tenoch Morales Lopez[1], Puneeth Deraje[2], Mete Yuksel[2], Dr. Lindi Wahl[1]
[1]University of Western Ontario, London, Canada, [2]University of Toronto, Toronto, Canada
The rate of de novo mutations varies across species and is predicted to be dependent on parameters such as population size, the average generation time, and how far the population is from the fitness peak. Mutations that modify the mutation rate itself often emerge in bacterial evolution experiments, either increasing or decreasing the mutation rate of ancestral lineages. Previous theoretical approaches have delineated the conditions necessary for these modifiers of mutation rate to spread in the population, either by hitchhiking with beneficial mutations or by reducing the load of deleterious mutations. One implicit assumption in previous work is that the fractions of beneficial and deleterious mutations are identical in the modifier and ancestral lineages (which differ only in how often new mutations arise). Recent empirical work, however, has highlighted the fact that modifications of the mutation rate are often accompanied by changes in mutation bias, such that different types of mutations occur at different rates. This can dramatically alter the availability of beneficial and deleterious mutations for the modifier lineage. In this talk, I will present a stochastic model and computer simulations which explore the effect of mutations that modify both the mutation rate and the mutation bias, demonstrating how these two effects simultaneously play a role in the evolution of microbes.

[A371] Cell type and cell division-resolved mutation rates in a hypermutating Nematode

Dustin Mullaney[1,2,3,4], Sam Sgrizzi[1,2], Ian Campbell[2], David Mai[2,3,5], Sneha Ray[2], Aakanksha Singhvi[2], Sanjay Srivatsan[2,4,5,6], Kelley Harris[2,4]

[1]Molecular and Cellular Biology Program, University Of Washington, Seattle, United States, [2]Basic Sciences Division, Fred Hutchinson Cancer Center, Seattle, United States, [3]Computational Biology Program, Fred Hutchinson Cancer Center, Seattle, United States, [4]Department of Genome Sciences, University of Washington, Seattle, United States, [5]Institue for Protein Design, University of Washington, Seattle, United States, [6]Brotman Baty Institue for Precision Medicine, Seattle, United States

Somatic mutations accumulate with each cell division, providing both a natural record of cellular lineage and a window into the mutational processes active in individual cells throughout an organism's lifespan. However, leveraging somatic mutations for lineage tracing or for profiling cell-type-specific mutation rates and spectra is not feasible with existing single-cell sequencing methods. To address this, we applied a novel capsule-based single-cell whole-genome and transcriptome co-sequencing assay (CapGTA) to C. elegans harboring a hypermutator allele of DNA polymerase epsilon (pole-1 P270R), generating a dataset that enables direct comparison of mutation dynamics across cell types in a system with known lineage. We performed CapGTA on thousands of cells from 20 individuals, achieving sufficient sequencing depth for precise variant calling and comprehensive transcriptome profiling. Using transcriptomic profiles for cell type annotation, we compare somatic mutation rate and spectra between cell types, and assess the performance of somatic mutation based lineage tracing utilizing the known C. elegans lineage tree as a benchmark. Preliminary results suggest CapGTA enables lineage reconstruction from somatic mutations, as well examination of cell type and tissue specific mutation rates and spectra. This multimodal dataset opens a new approach to understanding the causes and consequences of somatic mutation heterogeneity in development and disease.

[A372] Evolutionary dynamics of DNA repair genes across primate phylogeny

Emma Diepeveen[1], dr. Vasili Pankratov[1], Prof. Mikkel Heide Schierup[1]
[1]Bioinformatics Research Centre, Aarhus University, Aarhus, Denmark

DNA repair mechanisms are essential for maintaining genomic integrity. When cells experience DNA damage, they must employ appropriate repair strategies, and this pathway choice has critical consequences for mutation accumulation. Base excision repair (BER) corrects small base lesions from oxidation and deamination with minimal helix distortion. For double-strand breaks (DSBs), cells mainly choose between homologous recombination (HR) and non-homologous end joining (NHEJ). HR is precise, template-driven, and theoretically error-free repair, while NHEJ is faster but error-prone due to rejoining without requiring sequence homology. The efficiency and accuracy of pathway selection impacts genomic stability and mutation rates. We leverage high-quality genome alignments spanning over 40 primate species to investigate molecular evolution of genes across BER, HR, and NHEJ pathways. Our comparative genomics approach will reveal whether different repair pathways experience distinct evolutionary pressures across the primate phylogeny. We can integrate evolutionary signatures with functional output by correlating selection patterns to gene expression levels across tissues and observed mutation rates in both germline and somatic contexts. Additionally, we will examine relationships between DNA repair evolution and primate life-history traits such as lifespan, reproductive age, and body size. This multi-layered analysis will show how evolutionary pressures on DNA repair genes in primates relate to their actual repair efficiency and the genomic consequences of pathway choice.

[A373] Re-assessing the correlation between mutation rates and functional elements

Gustavo Valadares Barroso[1], Dr Aaron Ragsdale[1]
[1]University Of Wisconsin-Madison, Madison, USA

Several models of evolutionary biology rest on the premise that de novo mutations occur uniformly at random with respect to their fitness effects. A report of lower mutation rates in Arabidopsis genes drew criticism, but reignited the debate about whether this central tenet holds. Setting aside the Arabidopsis controversy, human somatic cells show evidence of reduced mutation rates in exons, although this result was not replicated in the germline. In contrast, whether mutation rates differ within and outside other constrained elements, such as enhancers, promoters, and phastCons, has not been systematically tested. Here we revisit this topic by taking advantage of three human mutation maps predicted in recent years, each focusing on different aspects of mutation rate variation. Using causal inference, we build a measurement model that represents the (unobserved) true mutation map as a latent variable indicated by each empirical map. This framework allows us to generate a synthetic construct of the mutation landscape that reduces measurement error and integrates the strengths of the individual estimates. We recapitulate previous findings that mutation rates are elevated in exons relative to the genomic background and further suggest that they are also elevated in enhancers and promoter regions. Conversely, we find that mutation rates are lower in phastCons elements and, in fact, negatively correlated with their estimated level of constraint. While this could suggest a systematic bias in the estimation of the mutation rates within functionally constrained regions, the contrasting behavior of phastCons and genes indicates that this topic deserves further consideration.

[A374] Selection for robustness drives the evolution of the per-genome mutation rate

Juliette Luiselli[1], Guillaume Beslon[2], Pieter van den Berg[1]
[1]KU Leuven, , Belgium, [2]INRIA, Lyon, France

Mutation rate variation is recognized as a key factor shaping evolutionary dynamics. Most theoretical and empirical work, however, focuses on variation in per-base mutation rates, independently of the genome structure − the target of the mutations. Selection, in contrast, integrates over replication events and not single mutation events: a replication event can involve zero, one, or multiple mutations. Thus, the mutation load relevant to selection is determined by the per-genome mutation rate, which depends jointly on per-base mutation rate and genome architecture.

Using a model of genome evolution allowing variation in gene copy number and in total genome size, we show that selection on replication-level mutation load can generate trade-offs with mutational robustness. In particular, selection for reduced per-genome mutation load can favor genome architectures with lower redundancy, even when per-base mutation rates are high - contrary to what is expected in a context of selection for mutational robustness where redundancy protects the genome against deleterious mutations. This occurs especially under relatively high rates of structural mutations such as duplications and deletions, which allow for varying the gene copy number but impose strong robustness constrains because of their capacity to greatly disrupt the genome.
Our results show that genome architecture (genome size, gene copy number) is highly relevant to the evolution of robustness, and generates mutation rate variation without changes in per-base mutation processes. More broadly, our findings highlight that variation in per-genome mutation rates can be as important as per-base mutation rate variation in shaping genome evolution and robustness.

[A375] Evolution of Mutational Susceptibility to Disease

Maria Kelly[1], Donate Weghorn[1]
[1]Centre For Genomic Regulation (CRG), , Spain

Cancer arises from somatic mutations shaped by underlying mutational processes. While extensive research has focused on how cancer is controlled once mutations arise in cancer driver genes, it remains unknown whether natural selection has also acted on the root cause: the probability of these mutations occurring in the first place. Here, we quantify evolutionary changes in mutational susceptibility to different processes by reconstructing ancestral genomes across the primate and mammalian phylogenies. Focusing on cancer driver genes, we find a successive decrease in susceptibility to key clocklike COSMIC mutational signatures SBS1 and SBS5 across ~30 million years of primate evolution leading to modern humans. Neutral simulations of the most recent human–chimpanzee evolutionary episode, incorporating local mutation rates and GC bias, reproduce broad trends but fail to account for the magnitude of reduction observed for clocklike and mismatch repair signatures, indicating an enrichment beyond neutral expectation. Extending this framework to germline mutational processes, we find similar decreases in susceptibility to the dominant germline signatures SBS5 and SBS1 when averaging across all protein-coding genes, suggesting that sequence evolution can modulate exposure to both germline and somatic mutation.
These findings suggest that mutational susceptibility is not evolutionarily static. Instead, lineage-specific sequence evolution can reshape exposure to particular mutational processes, linking germline evolutionary dynamics to somatic mutational risk. This work provides an evolutionary framework for understanding how mutation landscapes change across time and may shed new light on Peto's paradox and ultimately contribute to advancements in understanding disease susceptibility and cancer prevention.

[A376] Applications of structurally constrained substitution models of protein evolution to phylogenetic inference

Dr Miguel Arenas[1], Elena Pazos[1], Luis Daniel González-Vázquez[1], Ana Prado-Comesaña[1], David Ferreiro[1], Ugo Bastolla[2]
[1]University of Vigo, Vigo, Spain, [2]Centro de Biología Molecular "Severo Ochoa" – CSIC-UAM, Madrid, Spain

Substitution models describe the rates of evolutionary change among molecular states and are essential for a wide range of evolutionary studies, including the reconstruction of phylogenetic histories and ancestral sequences. At the protein level, the most used substitution models are based on empirical sequence data and rely on assumptions that can lead to unrealistic modeled proteins. Incorporating evolutionary constraints from protein structure has improved the accuracy of protein evolution modeling. Here, we present results from the application of structurally constrained substitution models of protein evolution to diverse phylogenetic inference, including the estimation of substitution and recombination rates and the prediction of evolutionary trajectories of viral proteins. This study was supported by the grants PID2023-151032NB-C21 and PID2023-151032NB-C22 funded by MCIU/AEI/10.13039/501100011033 and by FEDER, UE. LDGV was funded by a fellowship from Xunta de Galicia ED481A-2023/089, programa de axudas á etapa predoutoral da Xunta de Galicia (Consellería de Cultura, Educación, Formación Profesional e Universidades) cofinanciado pola Unión Europea no marco do Programa FSE+ Galicia 2021-2027.

[A377] Accelerated evolutionary rates of Influenza A neuraminidase gene in warm climates

Nina Pust[1], Harrison Zhu[1], Mark P. Khurana[1], Samir Bhatt[1,2], David A. Duchene[1]
[1]Department of Public Health, University Of Copenhagen, Copenhagen, Denmark, [2]MRC Centre for Global Infectious Disease Analysis, Imperial College London, London, United Kingdom

Influenza A virus is characterized by seasonal epidemics and continues to pose a major threat to global public health. Lower temperature and humidity leads to greater survival of the virus in airborne droplets, and this is expected to translate to a general link between local climates and strain emergence. We use a time-stamped global genetic data set to test for the link between the molecular evolutionary rate in surface proteins of two major seasonal strains, H1N1 and H3N2, and the environmental temperature, humidity, and local population density at time of collection. These data reveal evidence for a positive link between temperature and the overall evolutionary rates in the neuraminidase gene, but limited evidence for links between climate and metrics of selection, or with the evolution of hemagglutinin. These data point to a role for warm climates in driving antigenic drift and likely genetic draft in neuraminidase. Thicker respiratory tract mucus in warmer climates is probable to place heavy selection pressures on this gene for enhanced movement of virions through mucus layers. Warm climates globally might play a role in the maintenance of influenza A variants, and seeding of seasonal epidemics, via the emergence of efficient mucus-traversing neuraminidase molecules. We propose that with rising global climates, temperature data will complement finer scale demographic and surveillance data for describing the emergence and spread of novel influenza A strains.

[A378] How precise are mutation rate estimates? Comparison of different approaches to estimate de novo mutation rates

Xi Wang[1], Ms. Chaowei Zhang[1], Mr. Hongbo Wang[1], Dr. Carley Reid[1], Prof. Juha Merilä[1]
[1]Area of Ecology And Biodiversity, School of Biological Sciences, The University of Hong Kong, Hong Kong SAR, China

Availability of de novo mutation rate (μ) estimates based on approaches that rely on bioinformatic validations has increased tremendously during the past few years, but the accuracy and precision of these estimates often remain unclear in lack of Sanger sequencing validation of the mutations. We used both long- and short-read sequencing data and different bioinformatic pipelines to estimate μ, as well as false positive (FPR) and negative (FNR) rates, for family trios of flat-headed loaches (Oreonectes platycephalus). By comparing estimates against PCR-verified mutations, we observed that the top-performing approach (as ranked by F1 score of seven approaches at the same depth) still exhibited a 4% false positive rate (FPR) alongside a 12% false negative rate (FNR). Across the remaining methods, FPR values spanned 4–12%, while FNRs ranged from 8–19%. Irrespective of the bioinformatic approach used, long-read data yielded consistently lower μ estimates than the short-read data because of the larger callable genome sizes. In addition, a higher mapping depth resulted in a lower FNR. These results call for caution on reliance on purely bioinformatic validation of de novo mutations in non-model organisms and raise the possibility that many published μ-estimates, especially if based on low mapping depths, might be biased.

[A379] Bias in Estimation of the Population Scaled Mutation Rate Parameter θ Under High Mutation Regimes

Afarinesh Panahy[1], Dr. Jason A.P. de Koning[1]
[1]University of Calgary, calgary, Canada

The population-scaled mutation rate parameter, θ = 4Nμ, is a fundamental quantity in population genetics. In practice, θ is often estimated using summary statistics such as Watterson's θW and θπ (average pairwise differences). These estimators rely on weak-mutation and infinite-sites approximations, where new mutations are assumed to hit each position only once. When θ is high, recurrent mutation becomes common, these assumptions break down, and θ can be systematically underestimated.

To quantify this effect, I used our program SLiM-Tree, a framework that runs forward population-genetic simulations along phylogenies. Because it tracks polymorphisms through time under finite-sites assumptions, it directly tests how standard estimators behave outside the weak-mutation regime. SLiM-Tree also supports codon-level selection through site-heterogeneous fitness profiles, letting selection act on evolving sequences in a multi-allelic framework.

Across θ values from 0.001 to 0.6, under low and high recombination, and under neutrality and site-heterogeneous purifying selection, θW and θπ performed well at θ = 0.001 but became increasingly downward biased once θ exceeded ~0.05. At θ = 0.6, both estimators severely underestimated the true value, even under neutrality. Likelihood-based approaches were more robust: LAMARC tracked θ closely under neutrality but shifted downward under selection at high θ. Restricting inference to 4-fold degenerate sites did not improve accuracy; estimates were nearly identical to those from full coding alignments, likely due to background selection. These results show that standard summary-statistic estimators can strongly underestimate θ in high-mutation regimes, potentially biasing our understanding of what θ can be in nature.

[B001] From Self-Incompatibility to Mixed Mating: Evolutionary transitions between mating modes in plants

Amit Jangid[1], Prof. Ohad Noy Feldheim[1], Tamar Friedlander[1]
[1]Hebrew University of Jerusalem, Rehovot, Israel

Hermaphroditic plants employ diverse mating strategies, including self- and cross-fertilization or their mixture. Cross-fertilization is often ensured by 'self-incompatibility' (SI), genetic mechanisms rejecting self-pollen and allowing fertilization by nonself-pollen. Self/nonself discrimination relies on molecular recognition between two families of highly diverse proteins, encoded at the plant's S-locus and expressed in its reproductive organs. SI could breakdown following mutations, yielding 'self-compatible' (SC) individuals capable of self-fertilization. Transitions between these mating modes are considered frequent, and several species display SI, SC or their mixture across different populations. Despite the ubiquity of mixed-mating populations, the evolutionary origins and dynamic stability of mixed mating remain poorly understood.

We theoretically study the transitions between SI and SC mating modes, their reversibility and dynamic stability. We construct a model linking the S-locus architecture to the plant mating behavior and use population-level stochastic simulations to study its evolutionary dynamics. We study the effects of two key factors: promiscuity in molecular recognition underlying SI and the inbreeding depression, which penalizes self-compatibility.

We reveal three mating regimes: complete SI, complete SC, and a mixture combining both. The mixed regime, described here for the first time, exhibits long-lasting fluctuations in the abundances of SI and SC individuals, and does not relax to either 'pure' mating strategy. Lastly, we find that transitions between mating modes could be reversible.

The plant mating mode is tightly linked to its adaptive potential under environmental change. Thus, clarifying the factors favoring either mating mode provides insights relevant to conservation in the face of climatic shifts.

[B002] Inferring the demographic history of Caenorhabditis elegans while accounting for its self-fertilising reproductive mode

Chenxi Wang[1], Konrad Lohse[1], Prof. Erik Andersen[2], Dr. Matthew Hartfield[1]
[1]University of Edinburgh, Edinburgh, United Kingdom, [2]Johns Hopkins University, Baltimore, United States of America

Self-fertilisation extensively influences the evolutionary history of hermaphroditic species, including reducing population diversity and hence the effective population size. These changes are usually ignored in population genetic inference methods, which overwhelmingly assume outcrossing. Here, we investigated the demographic history in different populations of Caenorhabditis elegans, a predominantly self-fertilising nematode, using a recent sequential Markov coalescent (SMC)-based method for jointly inferring the population size trajectory and the self-fertilisation rate. Population structure analysis suggests that strains from different Hawaiian islands are more genetically divergent than strains outside Hawaii. Strains from Kauai exhibit the highest level of intrapopulation divergence, while the previously reported “Hawaiian” group consists mostly of strains from the Big Island. The demographic inference for different subpopulations from the Big Island estimated a selfing rate close to 99% and a population decline. Signals of population structure in more recent generations between different subpopulations were also detected, which implies a potential source of confounding effects. We also compared the results with the inference from SMC-based methods not accounting for self-fertilisation plus a rescaling of the population size afterwards. The study investigates the best strategy for demographic inference in predominantly selfing populations and sheds light on the demography of the C. elegans Hawaiian population.

[B003] Evolutionary Origins of Vertebrate Gonad Differentiation in an Agnathan Lineage

Sara Good[1], Ms. Amy Pitzel, Ilya Kisselev
[1]The University Of Winnipeg, Winnnipeg, Canada

Agnathans represent the most basal lineage of extant vertebrates, diverging from gnathostomes after the first round of whole-genome duplication (1R) and prior to the 2R-driven radiation of jawed vertebrates. Although lampreys possess a functional hypothalamus–pituitary–gonadal (HPG) axis, the molecular basis of sex determination and gonadal differentiation in this lineage remains unresolved. We hypothesized that conserved vertebrate gonad differentiation genes exhibit stage-specific and sex-biased expression patterns coincident with key histological transitions during lamprey development.

To test this, we analyzed RNA-seq data from sea lamprey gonads (n = 92) spanning larval, metamorphic, juvenile, and adult stages. Expression counts were analyzed using DESeq2 and weighted gene co-expression network analysis (WGCNA) to identify sex- and stage-associated modules and hub genes. We identified distinct transcriptional shifts during metamorphosis and early juvenile stages, coinciding with gonadal differentiation, including male-biased upregulation of germline-associated genes (n = 826, FDR < 0.05) and meiosis regulators (msh4, sycp1, hormad2), and female-biased expression of oogenesis-related pathways (bmp15, figla). Several hub genes showed orthology to vertebrate gonad differentiation genes predating 2R, while others appear lamprey-specific or expanded within the agnathan lineage. Together, these results delineate a temporally ordered gene regulatory program underlying lamprey gonadal differentiation and suggest that core components of vertebrate sex differentiation networks were already established prior to the emergence of jawed vertebrates.

[B004] Homologous Chromosome Reuse in Sex Chromosome Turnover

Yukako Katsura[1]
[1]Kyoto University, Inuyama, Japan, [2]The Graduate University for Advanced Studies, Hayama, Japan
The Japanese frog Glandirana rugosa, endemic to Japan, exhibits both XY and ZW sex determination systems in different populations, representing a rare example of sex chromosome turnover within a single species. To explore the genetic basis of this phenomenon, we analyzed the X, Y, Z, and W chromosomes using genomic sequencing approaches. All sex chromosomes originated from chromosome 7 and showed substantial sequence homology. Comparative analyses revealed a high degree of sequence similarity between the Y and Z chromosomes, suggesting that they may have originated from the same ancestral chromosome and remained highly homologous at the genomic level. This relationship supports the idea that transitions between XY and ZW systems can occur through the reuse of homologous chromosomes.

[B005] Effect of Polyploidy on Sex Chromosome Dosage Compensation in a Dioecious Plant

Elle Loughran, Aline Muyle[1]
[1]CNRS, Montpellier, France

Dioecy is a breeding system with separate females and males, where sex can be determined by sex chromosomes (for instance, XY males and XX females). Dioecy is generally associated with the evolution of sexual dimorphism. In this study, we tested whether small RNAs (sRNAs) play a role in sexual dimorphism and sex chromosome evolution in Silene latifolia. We generated new data for female and male sRNAs and DNA methylation. We showed that sRNAs are most of the time female-biased in S. latifolia, suggesting that the presence of the Y chromosome globally impacts the epigenome by diluting sRNAs genome-wide in males. We found limited evidence for the regulation of sex-biased genes by sRNAs, but we nonetheless identified a male-biased transcription factor that may potentially be regulated by sex-biased RNA-directed DNA methylation. This transcription factor might contribute to male traits through the regulation of key factors in sex determination and phenotypic sexual dimorphism. Finally, we compared female and male sRNA mapping along the S. latifolia sex chromosomes. We found that X and Y genes are targeted by significantly more sRNAs in males compared to females and pseudoautosomal region genes. Our results suggest that Y genes silencing following Y degeneration leads to the formation of sRNAs that can interact with both X and Y genes in males due to X-Y sequence homology. Our work calls for future investigation of the impact of these sRNAs generated from the Y chromosome on X gene expression in males.

[B006] Sex without crossovers mimics clonal reproduction in a multicellular eukaryote

Meng Zhang[1], Marco Castellani[1], Stefan Steckenborn[1], Maciej Majka[1], Dr. André Marques[1]
[1]Max Planck Institute For Plant Breeding Research, , Germany

Meiotic recombination ensures accurate chromosome segregation and promotes genetic diversity by generating crossovers between homologous chromosomes. While essential in most sexually reproducing organisms, recombination is variably regulated and can be absent in some lineages, a condition known as achiasmy. However, obligate achiasmy in both sexes of a sexual species has not been previously documented. Here, we investigate the beak-sedge Rhynchospora tenuis, a holocentric plant with the lowest known chromosome number among angiosperms (n = 2) and inverted meiosis. Using chromosome-scale genome assemblies from nine accessions, molecular cytogenetics, immunocytochemistry, high-throughput single-gamete sequencing and whole-genome sequencing of controlled crosses, we show that R. tenuis undergoes obligate, genome-wide achiasmatic meiosis in both sexes. Despite normal early meiotic axis formation, synapsis fails, crossovers are not detected cytologically or genetically, and univalents persist at metaphase I. Extensive haplotype-specific accumulation of transposable elements (TEs) generates segregation distortion (e.g. meiotic drive), favouring the transmission of larger, TE-rich chromosomes. Remarkably, sexual reproduction is retained with fertilisation producing viable seeds only when translocation-compatible gametes meet, indicating strong post-meiotic selection that eliminates incompatible homozygous combinations. As a result, all surviving offspring are genetically identical to the maternal genotype, effectively restoring heterozygosity each generation and mimicking clonal reproduction. We propose that the combined effects of recombination loss, low chromosome number, holocentricity, inverted meiosis, and selective transmission of longer chromosomes enable faithful segregation and clonal-like inheritance despite sexual reproduction. These findings challenge the boundary between sex and clonality, revealing a unique evolutionary strategy linking genome architecture, recombination loss, and transmission bias.

[B007] Leaving X: how scale insects repeatedly evolved alternatives to chromosomal sex determination

Andrew Mongue[1]
[1]University of Florida, Gainesville, United States

It is unclear why sex determination systems are faithfully conserved for hundreds of millions of years in some taxa but repeatedly turn over in others. For example, most true bug families employ a conserved X sex chromosomal system, but in scale insects, sex determination systems are more varied. Different families may retain X chromosomes, employ a rare form of haplodiploidy known as paternal genome elimination, or forgo sexual reproduction in favor of simultaneous hermaphroditism. We collected and sequenced representatives of rare scale insects to study genomic changes that coincide with transitions between sex determination systems.We recovered two independent losses of X chromosomal sex determination, once in the family Monophlebidae and a second time at the base of the families that employ paternal genome elimination. In the former, genomic rearrangements begin before the X is lost and any previously X-linked orthologs have been absorbed into one of two massive, shuffled autosomes in the hermaphrodites. In the latter, the X chromosome itself is faithfully preserved in early diverging paternal genome elimination taxa; it has merely lost its sex determination function. We analyze ortholog conservation across transitions and find few to no genes lost, but a number of orthogroups gained, suggesting these large transitions did not require losing previous sex determination genes. Expression data suggests that extreme sexual dimorphism of gene expression predates sex determination turnover and may have helped enable it. Together these new observations illustrate the multiple routes to sex determination turnover and help refine hypotheses for its causes.

[B008] Non-canonical genomic consequences of early sex chromosome degeneration under extreme heterochiasmy in Caribbean robber frogs

Antoine Houtain[1], Prof. Dr. Paris Veltsos[1], Miguel Landestoy[2], Sagar S. Shinde[1], José David Hernández-Martich[2], Roberto Alonso Bosch[3], Kamil Jaron[4], Wen-Juan Ma[1]
[1]Research group of Ecology, Evolution and Genetics, Biology Department, Vrije Universiteit Brussel, Brussels, Belgium, [2]Botanical and Zoological Research Institute, Faculty of Science, Universidad Autónoma de Santo Domingo, Ciudad Universitaria, Santo Domingo 10105, Dominican Republic, [3]Museo de Historia Natural Felipe Poey, Facultad de Biología, Universidad de la Habana, Calle 25 No. 455 entre JyI, Plaza de la Revolución, 10400 La Habana, Cuba, [4]Tree of Life, Wellcome Sanger Institute, Hinxton, United Kingdom

The canonical model of sex chromosome evolution is a one-way trajectory, starting with recombination suppression around a sex-determining locus due to sexually-antagonistic selection. This leads to Y/W chromosome degeneration via gene loss and regulatory disruption. However, in many anurans, recombination suppression may arise through extreme heterochiasmy, in which male recombination is limited to chromosome ends. How degeneration proceeds under this noncanonical route remains poorly understood. We address this gap using Caribbean robber frogs (Eleutherodactylus), a lineage displaying repeated transitions between XY and ZW systems, spanning homomorphic and heteromorphic sex chromosomes and with karyotype variation.
We used RNA-seq and Iso-seq of 16 species to (i) characterize diverse sex chromosomes and turnovers and (ii) test for gene expression dysregulation and dosage compensation. We are now building chromosome-scale phased genome assemblies to (iii) quantify the tempo of Y/W gene loss and (iv) evaluate how turnover and karyotype evolution shape degenerative trajectories. Preliminary analyses indicate frequent XY and ZW systems and suggest non-linear dynamics of gene loss. Sometimes, these patterns are accompanied by signatures of degeneration-mitigating processes, including gene gains and potential dosage compensation. We speculate that Eleutherodactylus heterogeneity in transitions from homomorphic to heteromorphic may be due to their extensive karyotypic variation. Our preliminary results indicate that sex chromosome degeneration is not an inevitable one-way process but a dynamic outcome shaped by recombination landscape, karyotype evolution, and repeated turnover, suggesting that non-canonical routes to recombination suppression can fundamentally reshape both the tempo and genomic consequences of sex chromosome evolution.

[B009] Diversity and Distributions in Egg-Laying vs. Live-Bearing Onychophora

Arianna Lord[1,2]
[1]Eawag, Dübendorf, Switzerland, [2]Harvard University, Cambridge, USA

Sexual reproduction and reproductive mode strongly influence patterns of diversity, dispersal, and speciation, yet many phylogenetically important groups remain poorly studied. Velvet worms (Onychophora), display a variety of reproductive modes ranging from different forms of viviparity to ovoviviparity and oviparity. Despite this, and in contrast to the global dominance and diversity of their highly successful sister group, Arthropoda, velvet worms have relatively low species richness (fewer than 250 species), share a more conserved body plan, and have a global distribution restricted to damp microhabitats in terrestrial realms of the tropics and former Gondwanan continents.
Here, we provide an overview of the evolution of reproductive modes in velvet worms and combine phylogenomic, biogeographic, and species delimitation analyses to investigate patterns of diversity and distribution in egg-laying and live-bearing velvet worms across Australia and New Zealand. By linking reproductive mode to phylogenetic diversity, species ranges, and endemism this work shows how reproductive mode may facilitate or constrain diversification in different environments. Understanding how reproduction relates to diversity, distribution, and persistence has important implications for conservation under environmental change, especially for dispersal-limited and threatened invertebrates.

[B010] Chromosome-level assembly of multiple abalone genomes reveals extensive gene duplication and domain expansion within gamete recognition proteins

Trevor Stepanyan[1], Paul Nicolosi[1], Damien Wilburn[1]
[1]The Ohio State University, Columbus, United States

Sperm-oocyte fusion is critical for the sexual reproduction of most animal species, yet the production of many sperm relative to a few ova risks pathological polyspermy. Multiple reproductive barriers have evolved to ensure only one sperm successfully fertilizes an egg, including the zona pellucida (ZP): a glycoprotein matrix surrounding the oocyte that physically restricts sperm access to the egg plasma membrane. While the ZP is a conserved feature of all animal oocytes, its constituent glycoproteins vary by species in number, domain composition, and intermolecular interactions such that the mechanisms by which sperm penetrate the ZP are poorly understood for most taxa. In the marine mollusk abalone, sperm secrete a small 16 kDa protein called Lysin that disrupts intermolecular interactions between ZP glycoproteins, creating an ~3 μm hole for sperm passage. Lysin-mediated ZP dissolution is species-specific, driven in part by coevolution with its primary receptor VERL. Compared to ~2-6 major ZP proteins per vertebrate species, the abalone ZP is comprised of more than 20 proteins whose functional roles in fertilization are poorly understood. To better characterize ZP protein evolution, we generated chromosome-level assemblies for six abalone species using long read Nanopore sequencing and performed an array of molecular evolutionary analysis. Conserved clusters of ZP genes were found on most abalone chromosomes, with multiple examples of species-specific gene expansions within clusters as well as domain expansions within specific proteins. Expanded phylogenetic analysis to include vertebrate and other invertebrate ZP proteins reveals multiple forms of convergent evolution on independent ZP gene lineages.

[B011] Determining the timing of sex-biased transmission distortion in human trios and its role in mitigating sexually antagonistic conflicts

Janice Planès[1], Dr Romain Laurent[2], Dr Bruno Toupance[2], Pr Thomas Bataillon[3], Elise Lucotte[1]
[1]Ecologie Systematique et Evolution, CNRS, Université Paris-Saclay, AgroParisTech, Gif-sur-Yvette, France, [2]Eco-anthropologie, Muséum national d'Histoire naturelle,CNRS, Université Paris cité, Paris, France, [3]Bioinformatics research center, Aarhus university, Aarhus,

Sexually antagonistic selection drives the emergence of biological differences between the sexes. It occurs when a genetic variant benefits one sex but harms the other, creating genomic conflicts since both sexes share most genes. The resolution of these conflicts leads to sexual dimorphisms. However, unresolved conflicts can persist, and sexually antagonistic polymorphisms have been detected in human populations, despite the high selective cost of maintaining them. In this project, we will detect genomic signatures of sexually antagonistic selection and determine when they arose in the life cycle. We previously detected transmission distortions according to the sex of children: i.e, when parents transmit more often one allele to their sons and another to their daughters, using a genome-scan approach of parents-child trios. Here, I will investigate the timing of sex-biased transmission distortion in humans by looking at transmission distortions according to the sex of the children AND the parents. Indeed, differences in allelic frequencies between the sexes can arise at various stages of the life cycle: 1-during gamete production or competition in males, a given allele may associate preferentially with X or Y chromosomes, if distortion comes from fathers only; 2-at fertilization, ova could select X or Y carrying sperms according to their own genotype, if distortion occurs in mothers only, 3-during embryonic development, the combination sex-genotype could lead to differential embryo survival, or via 4-Selection after birth if distortion occurs in both parents. In this pilot study, we will report results on 600 trios from the 1000 Genomes project.

[B012] Integration of p53 genes into sex determination mechanisms

Elzemiek Geuverink[1], Filippo Guerra[2], Jessie Chen[1], Anna Rensink[1], Eveline Verhulst[2]
[1]University of Groningen, Groningen Institute for Evolutionary Life Sciences, Groningen, Netherlands, [2]Wageningen University, Laboratory of Entomology, Wageningen, Netherlands

Sex in insects is controlled through a cascade of genes, in which a conserved binary switch (transformer) is instructed by an upstream signal of diverse origin. The evolutionary processes underlying the diversity of sex determination genes often involve gene duplication and neofunctionalization. The instructor gene wasp overruler of masculinization (wom) in the haplodiploid wasp Nasonia vitripennis (Hymenoptera; Pteromalidae) is a chimeric gene that includes a paralogous copy of the P53 domain coding sequence, and its expression is required for female development. The Nasonia genome also encodes a conserved copy of p53, orthologous to that found in other invertebrates, which is linked to protection of DNA integrity in germline cells. Most pteromalid wasps contain two homologs of p53: the ortholog (p53) and its paralogous copy (wom or p53p). We tested the function of both homologs in pteromalid wasps to examine their co-option in sex determination. Not only wom itself, but also p53 is involved in feminization of Nasonia diploid embryos. Yet in Muscidifurax wasps, only p53 has been incorporated in the sex determination cascade. Its paralogous p53p copy is expressed in early embryonic development, interacts with P53 itself, but does not impact the sex of the developing embryo when silenced. This suggests that integration of p53 into sex determination preceded the emergence of the novel instructor signal wom. We discuss how radiation of the p53 family in haplodiploid wasps fits in the pivotal role (regulation of transformer expression) of instructor signals in insect sex determination.

[B013] Elucidating molecular mechanisms of sex determination in stick insects

Emelyne Gaudichau[1], Dr. Jelisveta Djordjevic, Prof. Tanja Schwander[1]
[1]Unil, Lausanne, Switzerland

Sex determination is a key process in animal development, triggering the differentiation of primary and secondary sexual traits for reproduction. In animals, sex determination is controlled by conserved genes from the doublesex and mab-3 related transcription factor (DMRT) family. The gene doublesex was first discovered in the fly Drosophila melanogaster, followed by mab-3 in the worm Caenorhabditis elegans and finally the gene DMRT1 in vertebrates. DMRT genes underwent duplication events and evolved sex determination function in multiple animal taxa. In insects, sex determination is controlled by a cascade of genes that evolved around two genes called transformer and the DMRT family gene doublesex. Detailed molecular studies of these two genes in holometabolous insects suggested that insect sex determination is controlled via sex specific splicing. However, studies of a wider range of insect species challenge this assumption. Here we investigated the evolution and sex-specific splicing and expression patterns of the genes doublesex and transformer in stick insects, a hemimetabolous insect order. Surprisingly, we found a duplication of the gene doublesex with both copies featuring similar expression patterns between the sexes and across tissues, and no evidence for sex specific splicing. In contrast, we found evidence for sex-specific splicing of the gene transformer. In combination with new findings in other hemimetabolous insect orders, our data show that sex determination mechanisms in these orders differ strikingly from those in holometabolous insects. Thereupon, the study of non-model organisms will bring insights on the evolution of insect sex determination mechanisms believed to be conserved feature.

[B014] Exploring the Biophysical Consequences of Abalone Lysin Evolution with NMR Spectroscopy and Molecular Dynamics

Eric Todd[1], Damien Wilburn[1,2]
[1]The Ohio State University, Interdisciplinary Biophysics Program, Columbus, United States, [2]The Ohio State University Department of Chemistry and Biochemistry, Columbus, United States
Across the tree of life, fertilization proteins exhibit atypically high evolutionary rates, consistent with coevolutionary chase between female and male proteins. While examples of interacting sperm-egg proteins are limited, a recurring pattern is that high affinity binding is not predictive of functional significance or species specificity. Instead, rapid coevolution can reduce protein structural stability and the regularity of three-dimensional topologies, as high epistasis can restrict the evolvability of binding partners. A model for this phenomenon is abalone Lysin, a 134 amino acid sperm protein that interacts with multiple ZP-N domains within egg VERL (Vitelline Envelope Receptor for Lysin). Despite high sequence divergence between ZP-N domains, Lysin can broadly recognize this structural domain using a single quasi-ordered binding interface that has different affinity for each ZP-N domain, spanning several orders of magnitude. Interspecies residue substitutions have produced subtle changes to this binding interface that are challenging to quantify, but sufficient to enforce species-specific fertilization. Compounding this, rapid evolution produces variation within a population, and conformational shifts in both partners are believed to drive the dissolution; milieux of constant variation that remain elusive to modern biophysical techniques. Utilizing NMR spectroscopy and molecular dynamics, we observe candidate residues and conformations that may be responsible for function and investigate by collecting new spectra with site-specific mutants and cross-species binding partner combinations. Molecular dynamics predicts rapid transient interactions between termini within the quasi-ordered interface likely slowing dynamics to fertilization-relevant, NMR-observable timescales for quantitative evaluations of specific hypotheses, yielding new directions to iteratively hone our understanding.

[B015] Genetic trade offs between reproductive success and disease reveal sex specific pleiotropic pathways

Eva Brigos[1,2], Gerard Muntané[1,3,4], Arcadi Navarro[1,2,5,6,7]
[1]Department of Medicine and Life Sciences, Universitat Pompeu Fabra, Barcelona, Spain, [2]IBE, Institut de Biologia Evolutiva (CSIC-Universitat Pompeu Fabra), Barcelona, Spain, [3]Hospital Universitari Institut Pere Mata (HUIPM), Institut d'Investigació Sanitària Pere Virgili (IISPV-CERCA), Universitat Rovira i Virgili (URV), Reus, Spain, [4]Centro de Investigación Biomédica en Red de Salud Mental (CIBERSAM), Instituto de Salud Carlos III, Madrid, Spain, [5]Centre for Genomic Regulation (CRG), The Barcelona Institute of Science and Technology, Barcelona, Spain, [6]BarcelonaBeta Brain Research Center (BBRC) - Pasqual Maragall Foundation., Barcelona, Spain, [7]Institució Catalana de Recerca i Estudis Avançats (ICREA), Barcelona, Spain

Sex is a fundamental biological variable in human complex-trait genetics. Sex differences in disease risk and life history traits are often attributed to sex-specific selection, but canalization predicts that similar fitness outcomes can arise through different genetic pathways under stabilizing selection. To test sex specificity in fertility–disease trade-offs, we performed sex-stratified GWAS meta-analyses for number of offspring and 62 diseases in females and males. We estimated sex specific genetic correlations with LDSC, compared females and males with LD block jackknife covariance, and mapped pleiotropy with pleioFDR across European LD blocks.

SNP heritability of reproductive success was similar between females (2.94%) and males (3.07%). Genome-wide genetic correlations with disease were largely positive and highly concordant across sexes: ADHD showed the strongest positive correlation and refractive error the strongest negative in both sexes. Only five diseases showed nominal sex differences in genetic correlation, none of which survived multiple testing, indicating a broadly shared genome-wide relationship between fertility and disease risk in females and males.

In contrast, locus-level analyses revealed marked sex specificity. Of 127 pleiotropic loci, only seven were shared between sexes, with pleiotropy substantially more frequent in females than males. This combination of conserved genome-wide correlations and minimal overlap at pleiotropic loci suggests that similar fertility–disease trade-offs are achieved through partly distinct genetic pathways in females and males. These findings are consistent with canalization, whereby stabilizing selection preserves trait-level relationships while allowing sex-specific molecular mechanisms, with implications in disease vulnerability and responses to environmental and demographic change.

[B016] Incipient sex chromosomes in the dioecious alpine plant Silene exscapa.

Fabien Duez[1], Gwyneth Halstead-Nussloch[2], Martin C. Fischer[2], Gabriel Marais[3], Jos Käfer[1], Alex Widmer[2]
[1]ISEM, Montpellier, France, [2]ETH, Zürich, Switzerland, [3]GreenUPorto, Porto, Portugal
Dioecy evolved many times independently in flowering plants, and many dioecious plants have evolved sex chromosomes. While many sex chromosomes are discovered, little is known about the initial steps of recombination suppression. We here focus on Silene exscapa, a species from the sexually diverse Silene acaulis species complex that acquired dioecy rather recently, and independently from other species in the model genus Silene. We use haplotype-based assemblies from a male and a female plant to map whole-genome resequencing data from three different populations, and look for sex specific SNPs, coverage and structural rearrangements between the haplotypes. Amid a large amount of genetic and structural diversity, we detect a small female-specific region on one of the chromosomes common to the three populations. This ZW system has no ZW gametologs and is characterized by W genes only; recombination seems not to have been suppressed outside the sex-determining locus. Only a single gene is fully W-specific, and we discuss its role as a candidate gene for sex determination in the genus Silene.

[B017] The enduring mystery of faster-X and faster-haploid adaptation: reconciling theories with data

Filip Ruzicka[1], Yasmine McDonough, Tim Connallon, Andrea Betancourt, Beatriz Vicoso
[1]Institute Of Science And Technology Austria, , Austria

A widespread pattern in molecular evolution is that X-linked genes adapt faster than autosomal genes. Meanwhile, in evolution experiments, haploid lineages of yeast adapt faster than diploid lineages. These patterns of “faster-X” and “faster-haploid” adaptation have a canonical explanation: new beneficial mutations are recessive, and selection in hemizygotes (haploids) is more efficient than in diploids. Yet this canonical explanation conflicts with theories of dominance itself, which propose that beneficial mutations should be dominant for fitness. According to theories of dominance, “faster-autosome” and “faster-diploid” evolution should be the norm. To reconcile theories of dominance with data, we present new theories and data analyses. First, we present a theory based on Fisher’s geometric model, in which we show that faster-X adaptation can occur even when new beneficial mutations are dominant, provided that the phenotypic effects of mutations are large. Second, we empirically test this theory by exploring correlations between mutational effect sizes and faster-X adaptation in population genomic datasets of fruit flies, house mice, and humans. Finally, we consider a new possibility: that beneficial mutations on the X chromosome, due to the lower background load of segregating deleterious mutations, experience less Hill-Robertson interference on their way to fixation. Overall, our results highlight an enduring mystery in evolutionary genetics, and provide a pathway to its resolution.

[B018] Repeat landscapes and neo-sex chromosome evolution in the Owl Monkey Aotus (Mammalia, Platyrrhini).

Gustavo Toma[1], Dr Edivaldo Oliveira[2,3], M.Sc Klebson Mauricio[4], Dr Liliane Carneiro[4], Dr Marcelo Cioffi[1]

[1]Universidade Federal de Sao Carlos, Sao Carlos, Brazil, [2]Evandro Chagas Institute , Ananindeua, Brazil, [3]Universidade Federal do Para, Belem, Brazil, [4]National Primate Center, Ananindeua, Brazil

Neo-sex chromosomes originate from autosomal-sex chromosome translocations, representing excellent models for studying sex chromosome evolution. However, the role of repetitive DNA in this process remains poorly understood. The repeatome, a genome-wide catalog of repetitive DNA, enables high-throughput screening of sex-biased repeat landscapes, offering insights into repetitive DNA dynamics in complex sex chromosome systems. Owl monkeys (Aotus) exhibit a wide range of diploid numbers and are notable for recurrent emergence of neo-sex chromosomes. Here, we present a comprehensive cytogenomic analysis of the repeatome of the neotropical monkey Aotus infulatus, a species exhibiting an X_1X_2Y sex chromosome system. Using low-coverage genome sequencing from males and females, combined with comparative analyses of three congeneric species (A. nancymaae, A. vociferans, and A. trivirgatus), we characterized the composition, abundance, and evolutionary dynamics of their repeatomes. Our findings indicate that repetitive elements comprise approximately 37% of the A. infulatus genome, with satellite DNAs representing the most variable component among species. Nine satellite DNA families were identified in A. infulatus, which demonstrated strong sequence conservation across the genus, especially from centromeric repeats. This pattern supports an ancestral library of repeats shared among these species, followed by lineage-specific amplification events. Comparative sex-specific analyses revealed a slight enrichment of satellite DNAs in the male genome. Fluorescence in situ hybridization further revealed distinct chromosomal distributions of several satellites, including sex-biased accumulations on the X_1 and Y chromosomes. Together, these results underscore the central role of repetitive DNA, particularly satellite DNAs, in reshaping the architecture of sex chromosomes in A. infulatus.

[B019] Sex-Biased Germline Genome Architecture in the Sea Lamprey (Petromyzon marinus)

Ilya Kisselev[1], Dr. Lara Puetz, Sara Good
[1]The University Of Winnipeg, Winnipeg, Canada

Sex determination in sea lamprey (Petromyzon marinus) remains unresolved, with no known genetic or epigenetic mechanism. Lampreys undergo programmed genome rearrangement (PGR), in which large portions of the germline genome are eliminated from somatic cells, but the functional significance of this process is unclear. Our lab previously identified strong male-biased expression of germline-restricted genes in lamprey gonads, many of which are orthologous to genes involved in spermatogenesis in gnathostomes. These findings led us to hypothesize that PGR may be sex-specific and contribute to sex determination or differentiation.

To test this hypothesis, we generated whole-genome short-read data from egg (n = 6) and milt (n = 6) samples, supplemented by PacBio HiFi sequencing of ovary and egg tissues. We employed a reference-free k-mer framework to identify sex-specific sequences, defining sets based on k-mers unique to one sex and absent in somatic tissues. Additionally, we mapped egg and milt reads to a male-derived reference genome to quantify coverage across germline-restricted scaffolds.

Our analysis revealed sexual dimorphism in germline content. Egg-derived coverage was markedly depleted across putative germline scaffolds, while milt showed extreme enrichment, reaching ~30X of the median genome-wide depth. A de novo female germline assembly recovered only ~25% of male germline content. Furthermore, k-mer profiling identified ~10 million male-specific compared to only ~0.35 million female-specific 51-mers. Overall, the evidence suggests that PGR is highly sex-biased in sea lamprey.

[B020] Differential Amplification of Satellite DNAs in the Sex Chromosomes of the Engystomops freibergi–E. petersi frog Species Complex

Jennifer Pompeo[1], Phd Juan Ferro[2], Dr Domingos Rodrigues[3], Dr Luciana Lourenço[1]
[1]University Of Campinas, Campinas, Brazil, [2]CONICET-UNaM, Posadas, Argentina, [3]Federal University of Mato Grosso, Sinop, Brazil

The Edentulus clade of Engystomops is composed of E.pustulosus, the sister species of the Amazonian E. freibergi–E. petersi species complex. Within this complex, E.freibergi, E.petersi, Engystomops sp. from Amapá (AP)/French Guiana (FG), and Engystomops sp. from Mato Grosso (MT) possess X and Y chromosomes, which exhibit varying degrees of heteromorphism. Previous studies have supported a shared evolutionary origin of the Y chromosome, and distinct compositions of heterochromatic blocks on the E.freibergi and E.petersi X chromosomes. Here, we studied the satDNA content in the genome of Engystomops sp.-MT using the RepeatExplorer pipeline and mapped two satDNAs, EfrSat1 and EfrSat4, onto chromosomes of all species in the complex. EfrSat1 was clustered on the X chromosome of E.freibergi, E.petersi, and Engystomops sp.-MT, but not on the putative X chromosome of Engystomops sp.-AP/FG. Additionally, this satDNA was observed in terminal bands of several chromosomes. In E.“magnus”, which lacks heteromorphic sex chromosomes, EfrSat1 was detected on the long arm of chromosome 11, likely homologous to the X chromosome of the closely related species E.petersi. In E.“selva”, another species without heteromorhic sex chromosomes, EfrSat1 was mapped to terminal heterochromatic bands of multiple chromosomes. EfrSat4 was detected at the centromeres of all chromosomes of E.freibergi, E.“magnus”, E.“selva”, and E.petersi, but not on the sex chromosomes of Engystomops sp.-MT. Neither EfrSat1 nor EfrSat4 was identified in the E.pustulosus genome. These findings suggest that both satDNAs were present in the ancestor of the Amazonian species complex and underwent differential amplification across lineages, potentially contributing to sex chromosome differentiation.
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[B021] Disentangling the autosomal and X-linked response to female-limited selection of X chromosomes in Drosophila melanogaster

Katrine Lund-Hansen[1], Qinyang Li[1], Vatsal Goyal[2], Jessica Abbott[1]
[1]Biology department, Lund University, Lund, Sweden, [2]Birla Institute of Science and Technology, , India

We have previously used female-limited X chromosome (FLX) evolution to study how X chromosomes evolve when released from constraints arising from selection in males. We expected female-limited selection to favour female-beneficial alleles at sexually antagonistic X-linked loci, causing phenotypic feminization. Although we found support for this prediction for some traits, for others we found a puzzling pattern where X-linked evolution appeared to have been cancelled out. Here, we conducted a follow-up experiment to disentangle the effects of the evolved X and autosomes on trait evolution. We found that FLX X chromosomes encoded reduced locomotory activity levels in females, but that this effect seemed to have been counteracted by simultaneous evolution of autosomal loci, consistent with predictions from a simulation model of polygenic adaptation to sex-specific fitness optima. RNAseq data suggests that the response to selection was asymmetric between the sexes, with many more genes being differentially expressed in females compared to males. Female-biased genes were also significantly overrepresented among differentially expressed genes. Interestingly, combining female-evolved autosomes with standard X-chromosomes resulted in altered regulation of many X-linked genes, and vice versa, suggesting extensive coevolutionary effects across the genome. Together with previous work from our group, these results collectively suggest that mismatches between X-chromosomes and autosomes have a much larger effect on expression in females than in males.

[B022] Phylogenomic insights into the genetic basis of repeated parthenogenesis in Timema stick insects

João Souto[1,2], Vincent Mérel[1], Jérôme Goudet[1,2], Tanja Schwander[1]
[1]Department of Ecology and Evolution, University of Lausanne, Lausanne, Switzerland, [2]Swiss Institute of Bioinformatics, University of Lausanne, Lausanne, Switzerland

The repeated evolution of asexual reproduction offers unique insight into the forces shaping transitions between sexual systems. In Timema stick insects, parthenogenesis has evolved at least seven times independently and always through the same cellular mechanism, gamete duplication. This mechanism results in the complete loss of heterozygosity in a single generation. To understand the genetic basis of gamete duplication and convergent evolution of parthenogenesis-associated traits, we analysed whole-genome data from ten Timema species (five sexual, five parthenogenetic). We benchmarked two variant detection strategies, “single reference” and “liftover”, to identify the most suitable approach for interspecific comparisons across highly divergent lineages (~30 MYA). The liftover approach, which maps reads to species-specific genomes and converts coordinates through whole-genome alignments, avoids biases inherent to single reference mapping across deep divergences. Using this approach, we were able to analyse larger genome portions (up to 19% more callable sites) and inferred phylogenetic conflict across lineages using genome-wide D-statistics and branch length analyses. These analyses revealed limited introgression (0.2–0.7% of the genome) but substantial incomplete lineage sorting (ILS), implying that ILS is the main source of phylogenetic conflict among Timema species. We did not identify a single genomic region shared across all parthenogenetic species, suggesting a polygenic or dispersed genetic basis for parthenogenesis. Ongoing analyses using branch length-based approaches and ancestral recombination graphs are focusing on loci with excess shared variation between parthenogenetic lineages to clarify the role of ancestral polymorphisms in the recurrent evolution of parthenogenesis.

[B023] A shared genomic region underlying disparate ecologically and sexually selected traits in two Lake Malawi cichlids

Joel Elkin[1], Aaron Hickey[1], Dr Moritz Blumer[1], Professor George Turner[2], Professor Richard Durbin[1], Dr Emília Santos[1]
[1]University Of Cambridge, , United Kingdom, [2]Bangor University, , United Kingdom
The East African cichlid adaptive radiations have proven an exceptional system for studying the drivers and mechanisms of divergence and speciation, with many studies identifying key ecological innovations as well as sexually selected traits. Thus far, however, comparatively little work has focused on the complex interplay between ecological and sexual selection. A key advantage of the Lake Malawi cichlid radiation is that, in spite of their remarkable phenotypic diversity, many highly diverged species are able to produce fertile hybrid offspring, facilitating linkage mapping as a powerful tool to identify the genetic bases of interspecific divergence. I propose to present the results of an ongoing QTL cross between two cichlid species - a generalist, omnivorous species that inhabits shallow water habitats, and a deep-water species that hunts for buried invertebrates using a specialised sensory system. By examining a range of phenotypes individually, we identified a shared genomic region associated with variation in both this sensory system, and a sexually selected male ornament implicated in courtship and breeding behaviour. To validate this, we conducted a series of follow-up behavioural trials that showed a relationship between these two seemingly disparate traits, as the phenotype of the ornament predicted feeding strategy and success. This raises the hypothesis that selection acting on one trait has influenced the other, and thus broadens our understanding of how ecological and sexual selection interact and jointly contribute to the process of speciation.

[B024] Spatial transcriptomics of the sex-changing fish brain

Kaj Kamstra[1], Chloé van der Burg[1], Jana Longney[2], Sventja von Daake[3], Ava Handley[3], Simon Muncaster[2], Neil Gemmell[1]
[1]University Of Otago, Dunedin, New Zealand, [2]University of Waikato, Tauranga, New Zealand, [3]Malaghan Institute of Medical Research, Wellington, New Zealand

In one of the best examples of phenotypic plasticity know to date, certain fish species can undergo a complete change of sex in adulthood in response to a social cue. Although the brain initiates and drives this process, the central mechanism remains poorly understood. The New Zealand spotty wrasse (spotty; Notolabrus celidotus) is a protogynous (female-to-male sex change) labrid in which sex change can be triggered experimentally by the removal of a dominant male. Here, we apply a spatial transcriptomics approach to investigate the transcriptional profile of the brain before during and after sex change to determine how the brain mediates this astonishing process.

We generated spatially resolved gene expression maps across key neuroendocrine regions during defined stages of social transition. We identify region-specific transcriptional reorganisation associated with sex change, including dynamic regulation of steroidogenic pathways, neuropeptide signalling, and markers of radial glial cells and neural plasticity. Notably, candidate genes previously implicated in sex change show striking spatial heterogeneity, suggesting that localised neurogenomic remodelling underpins the initiation and progression of phenotypic transformation.

Our results demonstrate the power of spatial transcriptomics to bridge molecular, cellular, and behavioural scales in evolutionary biology. By revealing how social information is embedded within the spatial architecture of the brain, this work provides new insights into the mechanisms enabling rapid life-history transitions and highlights spatially resolved genomics as a tool for studying phenotypic plasticity in non-model systems.

[B025] When Auto- and Allopolyploidy Meet: Premeiotic Genome Duplication Stabilizes Meiosis in Asexual Hybrids

Karel Janko[1], dr. Jan Eisner[2], Dr. Stephen Schlebusch[3], Dr. Jan Pačes[4], Dr. Vladimir Trifonov[3]
[1]University of Ostrava, Ostrava 1, Czech Republic, [2]University of South Bohemia, České Budějovice, Czech Republic, [3]Institute of Animal Physiology and Genetics, Liběchov, Czech Republic, [4]Institute of Molecular Genetics, Prague, Czech Republic

Meiosis and sexual reproduction through reduced gametes are vulnerable to disruption when diverged genomes are brought together in hybrids. A convergent outcome of hybridization is the modification of germ cell's cycle leading to premeiotic genome endoreplication (PMER). This enables the formation of bivalents between identical chromosome copies (e.g. AA and BB), ensuring the production of clonal unreduced gametes and the origin of polyploidy.

Yet, PMER creates a paradox: polyploid germ cells contain duplicated sister chromosomes together with diverged allospecific orthologs as well as highly similar non-sister homologs, as exemplified by hexaploid $A_1A_1A_2A_2B_1B_1$ configuration. This should result in multivalent formation and meiotic instability according to standard models of homology search. Yet many hybrid asexuals exhibit evolutionary success and stable meiosis.

We combined cytogenetics, whole-genome sequencing, and polymer-based modeling of chromosome pairing in asexual loaches to test whether avoidance of non-sister pairing reflects adaptive changes in established clones or instead emerges directly from genome duplication and nuclear topology.

Both first-generation and natural clonal hybrids maintain apparently normal meiosis with crossover frequencies similar to their parental strains, yet effectively avoid pairing between orthologous and homologous non-sister chromosome sets. A mechanistic chromosome-pairing model suggests that conventional homology-driven dynamics cannot explain such strict pairing preferences. Instead, premeiotic endoreplication creates spatial constraints that keep duplicated chromosomes in close proximity, enabling rapid zippering between sisters and excluding interference from alternative homologous partners.

Our findings identify premeiotic endoreplication as a key evolutionary innovation that stabilizes polyploids and shapes the relationship between hybridization, recombination, and the evolution of sex.

[B026] Selfish male reproduction in stick insects

Luca Soldini[1], Tanja Schwander[1]
[1]University Of Lausanne, , Switzerland

The transmission of genetic information is fundamental to evolution, yet it is achieved through a striking diversity of reproductive modes. The reasons for, and evolutionary significance of, this diversity remain poorly understood. One possibility is that selfish genetic elements, which promote their own transmission, create or exacerbate genetic conflict, driving the diversification of reproductive modes. Here, we demonstrate that androgenesis—where only the male nuclear genome is transmitted to the next generation—is a selfish male strategy in the stick insect genus Bacillus. Specifically, individuals produced by androgenesis inherit their entire nuclear genome from their father due to the exclusion of the maternal genome during embryogenesis. Using experimental crosses between males from diverse genetic backgrounds and genetically homogeneous females, we show that males differ significantly in their ability to reproduce by androgenesis. Further, using genotyping-by-sequencing on a subset of males, females, and their progeny, we identify cytological mechanisms underlying androgenesis and propose an evolutionary scenario explaining its emergence. Our findings indicate that selfish male strategies can hijack core features of sexual reproduction to establish uniparental inheritance and highlight the central role of selfish genetic elements and genetic conflict in shaping the evolution of reproductive modes.

[B027] Rapid chromosome evolution in an anuran population resulting from secondary contact between lineages with different sex chromosomes

Lucas Henrique Bonfim Souza[1], Juan Martin Ferro[2], Dr Luciana Lourenço[1]
[1]University Of Campinas, Campinas, Brazil, [2]CONICET-UNaM, Posadas, Argentina
The Physalaemus cuvieri – Physalaemus ephippifer species complex shows high chromosomal diversity. A population (CZ Pep-L1B) originating from ancient secondary contact exhibits exclusive heteromorphic Z and W chromosomes, distinct from its parents (Pep and L1B). In contrast to the CZ Pep-L1B population, L1B presents undifferentiated sex chromosomes, whereas Pep displays more heteromorphic Z and W chromosomes. Given the satDNA enrichment on sex chromosomes, we aimed to identify satDNA candidates with distinct chromosomal patterns between the parental lineages Pep and L1B, as well as in the CZ Pep-L1B, to better investigate chromosomal evolution in these frogs. We used the RepeatExplorer (RE) pipeline to analyze short-read WGS data from Pep, L1B, and CZ Pep-L1B. and selected PepSat1–21 for comparison among these clades. PepSat1 was most abundant in Pep and least abundant in L1B, whereas CZ Pep-L1B exhibited an intermediate pattern. Remarkably, chromosome mapping of PepSat1 probes detected pericentromerical signals on the Z chromosome of CZ Pep-L1B, but not on the homeologous chromosome 9 of L1B nor on Z and W chromosomes of Pep. Additionally, PepSat1 clusters were detected in variable numbers (five to seven) on autosome pairs in the three groups. These results reveal the first detected difference between the Z chromosome of CZ Pep-L1B and chromosome 9 of L1B. Together with the NOR on chromosome 7 and a typical W chromosome, this distinctive trait on the Z chromosome supports the hypothesis that the CZ Pep-L1B represents a separate lineage, despite having evolved from secondary contact between Pep and L1B.
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[B028] Human Sex Chromosome Diversity in the Turkana

Margaret (Maggie) Hassler[1,2,3], Dr. Rebecca Siford[1,2,3,4], Dr. Angela Taravella Oill[2,3,5], Dr. Sarah Mathew[1,3], PhD Anne Stone[1,2,3], Dr. Melissa Wilson[4]

[1]School of Human Evolution and Social Change, Arizona State University, Tempe, United States, [2]Center for Evolution and Medicine, Arizona State University, Tempe, United States, [3]Institute of Human Origins, Arizona State University, Tempe, United States, [4]National Human Research Genome Institute, Bethesda, United States, [5]School of Life Science, Arizona State University, Tempe, United States

Comparing nucleotide diversity (π) between the sex chromosomes and autosomes offers insights into sex specific demographic and selective pressures in humans. Yet empirical work on the demographic and selective forces shaping human sex-chromosome evolution has focused largely on non-African populations, leaving a critical gap given Africa's abundance of human genetic diversity. We quantified nucleotide diversity (π) across the autosomes, X chromosome, and Y chromosome to assess sex-chromosome diversity relative to autosomal expectations in 2,586 whole-genome sequences from 27 global populations. Notably, we included 82 Turkana individuals (35 XY, 47 XX), a Nilotic-speaking pastoralist population from northern Kenya that is currently underrepresented in public datasets. In the Turkana, we observe reduced diversity on both sex chromosomes, with an X-to-autosome ratio of 0.70 (neutral expectation 0.75) and a markedly reduced Y-to-autosome ratio of 0.106 (neutral expectation 0.25). By contrast, the Yoruba and Luhya populations consistently fall above the 0.75 expectation; 0.76 and 0.77 for the X, respectively. This result reflects the broad spectrum of sex-specific histories in humans, and the importance of diverse sample cohorts. Currently, we are refining these estimates and performing diversity calculations on the Y chromosome to provide more context for these results. Further research will implement forward and coalescent simulations to evaluate which evolutionary scenarios best explains the observed diversity in the Turkana. By integrating genomic data and evolutionary theory, this study addresses how sex-specific cultural processes and chromosome architecture jointly shape sex-chromosome diversity and informs general principles of sex evolution.

[B029] Male Xist Expression and Atypical X-Chromosome Regulation in the White-Footed Deer Mouse (Peromyscus leucopus)

Maria Andrade Ludena[1], Jonathan Duong[2], Aqsa Motiwala[2], Anthony Long[3], Alan Barbour[2,4], Sha Sun[1]
[1]Department of Developmental and Cell Biology, Charlie Dunlop School of Biological Sciences, University of California, Irvine, Irvine, United States, [2]Department of Microbiology and Molecular Genetics, School of Medicine, University of California, Irvine, Irvine, United States, [3]Department of Ecology and Evolutionary Biology, Charlie Dunlop School of Biological Sciences, University of California, Irvine, Irvine, United States, [4]Department of Medicine, School of Medicine, University of California, Irvine, Irvine, United States

X-chromosome inactivation (XCI) equalizes X-linked gene dosage between XX females and XY males in eutherian mammals and is initiated by the long noncoding RNA Xist, which coats one X chromosome and recruits repressive chromatin marks. Xist expression has therefore been considered female-specific and absent in XY males. Here, we examine Xist expression and X-linked gene regulation in the white-footed deer mouse (Peromyscus leucopus), a rodent with a conventional XY karyotype. Using bulk RNA sequencing, RT-qPCR, and RNA fluorescence in situ hybridization, we detect Xist expression in somatic cells of both female and male P. leucopus. In females, Xist RNA forms a chromosome-wide domain, whereas in males it appears as discrete nuclear puncta. In both sexes, Xist RNA signal colocalizes with the repressive histone modification H3K27me3. Using CUT&Tag profiling of H3K27me3 together with RNA sequencing, we identify X-linked genes that are enriched for H3K27me3 and show reduced expression in males. We further demonstrate that X-linked genes are expressed at lower overall level than autosomal genes in both sexes of P. leucopus, indicating that complete X-to-autosome dosage compensation is not achieved following Xist expression and repressive chromatin marking. These results also demonstrate that Xist expression can occur outside the canonical female-specific X-chromosome inactivation pathway in P. leucopus. Our results challenge the universality of current XCI models and suggest that partial, gene-specific X-chromosome regulation represents a viable and evolutionarily stable dosage compensation strategy in mammals.

[B030] Rare sex, recessive deleterious mutations, and the maintenance of genetic diversity

Mélodie Bastian[1], Dr Fanny Pouyet[1]
[1]LISN, Universite Paris Saclay, Gif sur Yvette, France

Sexual reproduction and recombination are commonly viewed as key mechanisms which limits the accumulation of deleterious mutations and preserve genetic diversity. However, some eukaryotes present facultative sex and extended phases of clonal reproduction. This raises the question of how neutral polymorphism is maintained under reduced recombination.
Here, we investigate how linkage to deleterious mutations shapes genetic diversity in facultatively sexual populations, using Saccharomyces cerevisiae as a model. Genetic diversity can be reduced around a negatively selected site, an effect known as background selection (BGS). However, when deleterious alleles are recessive (dominance $h \leq 0.5$), they can be maintained in a heterozygous state. This can favor haplotype diversity around the selected site and thus increase local genetic diversity, reducing the BGS effect. This effect is known as associative overdominance (AOD).
We simulate populations under various sex frequencies (alpha), selection and dominance coefficients (h), using the forward-in-time SLiM software. We characterize the conditions under which BGS and/or AOD acts. While AOD can emerge in both sexual regimes, we show that facultative sex expands the parameter space in which AOD attenuates BGS diversity reduction. Interestingly, when sex is rare ($alpha \leq 0.001$) and dominance is small ($h \leq 0.25$), AOD increases genome-wide genetic diversity above neutral expectations.
Our results demonstrate how dominance, linked selection, and reproductive mode jointly shape genomic diversity. More broadly, this work provides a theoretical framework for understanding how facultative sex can persist despite limited recombination, contributing to broader insights into the evolution of sex and recombination across eukaryotes.

[B031] Reversible Y chromosome evolution under extreme heterochiasmy in the common frog

Fantin Carpentier[1], Paris Veltsos[1,2], Ezgi Unal[1], Kelsey L. Doucette[3], Ricard Fontserè[1], Alan Brelsford[4], Melissa A. Toups[2,3], Nicolas Perrin[2], Wen-Juan Ma[1,2]
[1]Vrije Universiteit Brussel, Elsene 1050, Belgium, [2]Department of Ecology and Evolution, University of Lausanne, Lausanne, Switzerland, [3]Department of Biology, University of Louisiana at Lafayette, Lafayette, USA, [4]Evolution Ecology and Organismal Biology Department, University of California, Riverside, USA

Sex chromosome evolution is traditionally viewed as a unidirectional process with progressively suppressed recombination around a sex-determining locus, leading to irreversible Y-chromosome degeneration. Many vertebrates deviate from this canonical trajectory, and frogs are a striking example: they retain homomorphic sex chromosomes, exhibit extreme heterochiasmy, and undergo X–Y recombination in sex-reversed females. Here we investigate a Swiss Alpine population of the European common frog (Rana temporaria) with homomorphic XY chromosomes and three coexisting Y haplotypes, using sequencing of pooled samples of distinct Y haplotypes, sequencing of doubled-haploid YY individuals, and RNA-seq. We uncover extreme heterogeneity in X–Y divergence: two fully differentiated Y haplotypes span ~90% of the sex chromosome (~618–625 Mb), forming the largest non-recombining region described in a vertebrate, whereas a semi-differentiated Y harbors a small 4.64-Mb non-recombining region and XX males, also occurring in the population, are indistinguishable from females. Y degeneration is limited to elevated transposable element insertions. There is no gene loss, Y-linked copy decay, faster-X effects, or enrichment of sex-biased genes. All Y haplotypes share a small non-recombining region encompassing the candidate master sex-determining gene Dmrt1, which evolved ~3.47-5.70 Mya. Our results support a dynamic model in which extreme heterochiasmy and recurrent X–Y recombination via sex reversal repeatedly reset X–Y divergence, generating and maintaining multiple Y haplotypes over long timescales. These findings reveal a non-canonical, reversible mode of sex chromosome evolution shaped by sex-specific recombination, challenging the universality of classical models and highlighting non-model vertebrates for understanding sex chromosome diversity.

[B032] Breathing without lungs: integrative omics analyses into the functional complexity of lungless salamander mucus

Paul Nicolosi[1], Liam Johnson[1], Damien Wilburn[1]
[1]The Ohio State University, Columbus, United States

The evolutionary transition in vertebrates from aquatic to terrestrial life required a myriad of evolutionary innovations including many changes to the integument for preventing water loss as well as protecting from UV radiation, damage, and infection. Near the base of the terrestrial vertebrate phylogenetic tree and with life histories including both aquatic and terrestrial phases, amphibian physiology provides a window into this key evolutionary transition. Lungless salamanders (family Plethodontidae) are an especially attractive model for studying these adaptations, being highly speciose, variable in their life histories, reliant on cutaneous respiration, and possess remarkable resistant to chytridiomycosis that has ravaged other amphibian populations. While their skin secretions have been implicated in diverse biological processes, its biochemical composition is uncharacterized, and the molecular basis of its functionality is poorly understood. Here we provide the first molecular investigation of plethodontid mucous using quantitative mass spectrometry-based proteomics paired with long-read transcriptomic sequencing of multiple skin samples. To identify potential evolutionary and functional changes in mucous composition, we examined the secretions of six plethodontid species from three genera including five terrestrial (Plethodon shermani, P. metcalfi, Desmognathus ocoee, D. wrighti, Eurycea guttolineata) and one aquatic species (D. amphileucus). Most identified mucous proteins belonged to multigene families with functions associated with respiration, diffusion, immunity, and chemical signaling, including several novel paralogs of the pheromone sodefrin precursor-like factor (SPF) that we hypothesize may contribute to social communication. Our integrative omics workflow serves as a powerful tool for deeply characterizing molecular systems in non-model systems lacking genomic resources.

[B033] Defining the interaction landscape within the mammalian sperm acrosome through evolutionary and structural analysis

Regina Edgington[1], Hailey Hann[1], Charles Muller[2], Damien Wilburn[1,3]
[1]Department of Chemistry and Biochemistry, The Ohio State University, Columbus, United States,
[2]Department of Urology, University of Washington School of Medicine, Seattle, United States,
[3]Center for RNA Biology, The Ohio State University, Columbus, United States

Gamete recognition proteins often evolve rapidly due to arms race dynamics between sperm and egg, making it challenging to characterize reproductive protein interactions given extensive sequence and compositional divergence. The acrosome is a sperm-specific, membrane-bound organelle that houses many putative gamete recognition proteins organized in a paracrystalline matrix. During fertilization, the acrosome undergoes a regulated release of its protein contents – the “acrosome reaction” – allowing for serial, temporal sets of interactions with the oocyte.

Recent quantitative proteomics work by our laboratory shows that perturbations in acrosomal protein stoichiometry are common in idiopathically infertile human sperm. Despite the well-established importance of the acrosome, the functional roles and molecular interactions of many acrosomal proteins remain poorly understood. Inspired by recent structural prediction tools allowing for the discovery of new reproductive protein complexes such as IZUMO1-SPACA6-TMEM81, we have combined molecular evolutionary analyses with AlphaFold-based structural modeling to identify potential signatures of protein interactions across the acrosomal protein landscape. Using the human acrosomal proteome as a reference, orthologous sequences were identified for 105 mammal species from Ensembl, with evidence of selection inferred using site-based codon models and coevolution with other acrosomal proteins estimated by evolutionary rate covariation. Potential protein interactions were predicted using pairwise AlphaFold3 docking. Supporting this approach, preliminary large-scale AF3 docking analysis recovers the known ACR–ACRBP interaction among the top 2% of predicted complexes. Evolutionarily coupled binding interfaces remain structurally constrained even amid substantial sequence divergence, producing concordant coevolutionary and docking signatures that enable discovery of new reproductive protein interactions.

[B034] Genomic footprints and genetic drivers of rapid sex chromosome turnover in European true frogs

Ricard Fontserè[1], Prof. Dr. Paris Veltsos[1], Kim Roelants[1], Alexander Suh[2], Wen-Juan Ma[1]
[1]Research group of Ecology, Evolution and Genetics, Biology Department, Brussels, Belgium, [2]Leibniz Institute for the Analysis of Biodiversity Change, Museum Koenig Bonn, Bonn, Germany
In sharp contrast to the long-lived, highly degenerated sex chromosomes of mammals and birds, most amphibians, fishes and reptiles undergo frequent sex chromosome turnover. However, the genomic footprints of turnover and the underlying genetic drivers remain poorly understood, limiting rigorous tests of competing hypotheses about their evolution such as mutation load, sexually antagonistic selection, drift, selfish elements. True frogs (Ranidae) have among the fastest documented turnover rates in vertebrates, providing an exceptional comparative framework to identify recurrent drivers and genomic correlates of rapid turnover. We integrate phased chromosome-scale genome assemblies, whole-genome resequencing, and RNA-seq across 6 ranid species to (i) infer sex linkage and quantify sex chromosome differentiation using a unified pipeline (read coverage ratio, SNP density ratio, GWAS, FST, and sex-specific k-mer), (ii) localize sex-determining regions, and (iii) test whether turnover is accompanied by systematic shifts in structural variation (SV) and repeat landscapes. We identify 5 independent sex chromosome transitions across the clade and narrow candidate sex-determining regions in several species. Initial results suggest two possible genetic drivers: duplication/translocation of candidate genes and novel mutations in genes embedded in the sexual development pathway. Strikingly, comparative analyses further reveal that SVs and repeats/TEs insertions track the inferred sex chromosome turnover age, linking it to mutation accumulation. Together, our results reveal the molecular origins of new candidate master sex-determining genes and predictable genomic signatures across repeated transitions, advancing a mechanistic, comparative test of rapid sex chromosome turnover and supporting a key role for mutation accumulation in shaping sex chromosome turnover.

[B035] Evolution of protein interactions underlying fertilization in an early diverging fungus

Sujal Phadke[1], Luiza Borges
[1]California State University San Marcos, CA, San Marcos, USA

Fertilization in sexually reproducing species involves fusion of two cells, which is driven by interaction between cell surface proteins. For instance, fertilization in sexually dimorphic species requires interactions between proteins on egg and sperm surfaces whereas fertilization in protists and fungi that are not usually sexually dimorphic, requires interactions between surface proteins on the cells of 'opposite' mating types. We are studying candidate gamete interaction proteins in an early diverging fungus, Allomyces macrogynus (Am), which shows dimorphic gametes (anisogamy) that are otherwise rarely seen in fungi. Am shows anisogamy in size, pigment, motility and pheromone production, reminiscent of dimorphism seen in metazoa. It is unclear whether anisogamy in Am evolved independently of that in animals or represents ancestral state at the root of Ophisthokonts. We identified homologs of mammalian gamete fusion proteins in the transcriptomics dataset in Am. The present study investigates interactions of these homologs using yeast two-hybrid assays. Our analysis provides insights into the evolution of protein-protein interactions underlying gamete fusion in Am that represents an early diverging lineage following the split of animals and fungi.

[B036] The effects of chromosome number and recombination landscapes on the fitness consequences of different reproductive modes.

Sylvain Glémin[1]
[1]CNRS, Université Rennes, Rennes, France, [2]Uppsala University, Uppsala, Sweden
It is increasingly recognized that transitions between sexual and asexual reproduction involve a wide diversity of mechanisms, including various modifications of meiosis and mating patterns (e.g., selfing, automixis, pseudogamy). To understand these transitions, it is necessary to better quantify the fitness consequences of these atypical reproductive modes beyond the simple—and often oversimplified—two-fold cost of sex. A key question is how segregation and recombination are altered in these modes and what their fitness consequences are compared to regular meiosis and syngamy.
Using a theoretical approach, I will present how the number of chromosomes and the recombination landscape along chromosomes affect the fitness cost of different reproductive modes by modulating the distribution of deleterious mutations transmitted to offspring. In particular, I will show that the deleterious effects of different forms of selfing and automixis are reduced in species with few chromosomes, few crossovers per chromosome, and a heterogeneous recombination landscape. Leveraging publicly available recombination maps, I will quantitatively assess these predictions in species with contrasting karyotypes and recombination patterns.

[B037] Mating system shifts and polyploidy shape evolutionary dynamics in Rorippa

Uliana Kolesnikova[1], Ting-Shen Han[1], Dr. Polina Novikova[1,2]
[1]MPI For Plant Breeding Research, Cologne, Germany, [2]VIB-UGent Center for Plant Systems Biology, Ghent, Belgium

Mating system shifts such as the transition from outcrossing to selfing are common in plants and can affect genetic diversity and gene flow, thereby influencing adaptation and speciation. Transition to selfing typically leads to decrease of genetic diversity, reduced gene flow and reproductive isolation. In contrast, self-compatibility can facilitate the formation and establishment of polyploid hybrids (allopolyploids), leading to complex reticulate evolutionary histories.

To describe the interplay between mating system shifts and speciation patterns, we focus on the Brassicaceae genus Rorippa. This globally distributed genus comprises more than 70 species, of which at least 14 are allopolyploids. Despite obligate outcrossing being ancestral in Rorippa, all allopolyploid species are self-compatible. We sequenced more than 1100 individuals across the Rorippa genus to reconstruct the genus-wide phylogeny and describe the natural variation in mating systems.

We genotyped the S-locus controlling self-incompatibility in Rorippa and showed that more than 40 S-alleles out of 60 are shared with Arabidopsis due to 15 million years of balancing selection. Furthermore, we show the correlation in S-allele frequencies between Arabidopsis and Rorippa, suggesting the conserved dominance hierarchy among S-alleles. We confirm that all allopolyploid Rorippa hybrids are self-compatible and argue that transitions to self-compatibility in allopolyploids are often explained by dominant loss-of-function mutation in one of the S-alleles.

Our results demonstrate that recurrent transitions to self-compatibility, driven by changes at the S-locus, have facilitated allopolyploid formation and shaped the reticulate evolutionary history of Rorippa.

[B038] Sperm competition intensifies purifying selection on testis-specific genes in primates

Vasili Pankratov[1], Mr Bjarke Meyer Pedersen[1], Mengjun Wu[1], Juraj Bergman[1], Prof Mikkel Heide Schierup[1]
[1]Aarhus University, , Denmark
Mating systems with one female mating with different males lead to sperm competition. In turn, sperm competition is thought to impose strong selective pressures on male reproductive biology traits, including relative testis size and efficiency of spermatogenesis. Recent studies provide evidence for sperm competition intensifying purifying selection on spermatogenesis-relevant genes in mammals; however, this evidence remains controversial.
Here, we test the hypothesis that more intense sperm results in stronger purifying selection acting on testis-specific genes in primates.
To address this, we use the ratio of nucleotide diversity at nonsynonymous and synonymous sites in protein-coding genes ($\pi N / \pi S$) as a measure of purifying selection intensity. We estimate this value in 821 genes with testis-specific expression in the great apes and the rhesus macaque, and compare it across primate species with varying levels of sperm competition. In this comparison, we control for exome-wide $\pi N / \pi S$ differences between species due to differences in effective population sizes.
We show that $\pi N / \pi S$ in testis-specific genes is lower in species with higher levels of sperm competition. This result holds both when the reported mating system or the relative testes size is used as a proxy for levels of sperm competition (p-values for the association are 5.5x10-3 and 9.8x10-5).
Our results show that a) evolutionary changes in behaviour can lead to shifts in selective pressures, and b) species with low sperm competition are prone to accumulating (slightly) deleterious mutations affecting spermatogenesis. These findings are also important for understanding genetics of human male fertility.

[B039] Genetic basis of complex traits in closely related black widow spiders

Vladislav Ivanov[1], Kardelen Özgün Uludağ[2], Professor Dr Jutta Schneider[2], Professor Dr Henrik Krehenwinkel[3]
[1]University Of Rostock, Rostock, Germany, [2]University of Hamburg, Hamburg, Germany, [3]University of Trier, Trier, Germany

Recently diverged species provide a great resource to study genetic basis of traits involved in speciation, particularly traits under sexual selection, that are thought to evolve rapidly. Here we present a genome-wide association study of two closely related black widow spiders, Latrodectus hasselti and L.katipo. The endemic New Zealand katipo is estimated to have split from the invasive Australian hasselti only ~100,000 years ago, yet the two species exhibit striking phenotypic divergence, especially in the traits relevant for mating.
Notably, L.hasselti males perform an iconic “suicidal somersault” during copulation, while L.katipo males do not. A specialized abdominal constriction that prolongs male survival during mating is absent in L.katipo but present in L.hasseli. Finally, extreme sexual size dimorphism is more pronounced in the Australian species. To investigate the genetic basis if these differences, we generated F1 hybrids and backcrossed hybrid females to the males of each species. Backcross males were phenotyped for size, abdominal constriction, and mating behavior, and genotyped using low-coverage whole-genome sequencing.
Our analysis reveals a strong association between body size and loci on the sex chromosomes, involving genes with developmental and regulatory functions (e.g. secreting family GPCRs, DAC). In contrast, abdominal constriction is associated with an autosome with genes responsible for muscle contraction and ion transport (e.g. UNC 93, RyRs). Mating behavior was not associated with any genomic region suggesting complex polygenic nature of the trait. Together, these results highlight how rapidly sex-linked and autosomal traits can diverge and provide a framework to study speciation in non-model species.

[B040] The role of genomic imprinting in the evolution of separate sexes

Yangfan Feng[1,2]
[1]Centre National De La Recherche Scientifique (CNRS), Montpellier, France, [2]Institut des Sciences de l'Évolution de Montpellier (ISEM), Montpellier, France

My current research project focuses on the evolution of genomic imprinting and sex chromosomes in plants. The overall goal of my research is to test the hypothesis that sex antagonism can drive the evolution of sex-specific imprinting, and to investigate the role of epigenetics in sex chromosome evolution, particularly sex-biased gene expression, X-linked imprinting, and dosage compensation. To test the sexual antagonism hypothesis, I perform reciprocal crosses for two pairs of species with different sexual systems (dioecious, monoecious, hermaphroditic). I infer imprinted genes using SNPs and parental haplotypes, and compare patterns of genomic imprinting between dioecious species and their co-sexual relatives. To further investigate the association between epigenetic modifications, sex-biased expression, and sex-linked imprinting, as well as the underlying regulatory mechanisms, I integrate DNA methylation data, transcriptomic, and histone modification data generated through Oxford nanopore sequencing, RNA-seq, and CUT & RUN technologies. Ultimately, I aim to elucidate the role of epigenetics in sex chromosome evolution, using Silene species as a model system.

[B041] Structurally complex loci as engines of adaptation: insights from amylase evolution

Dr. Omer Gokcumen[1]
[1]University At Buffalo, Buffalo, United States
Structurally complex loci are among the most dynamic yet poorly understood regions of mammalian genomes. The amylase locus is a canonical example, long associated with starch digestion, metabolism, and dietary adaptation, yet the mechanisms driving its recurrent duplication, extreme structural diversity, and adaptive significance remain unresolved. Here, we integrate long-read sequencing, optical mapping, comparative genomics, and functional assays to dissect amylase evolution across mammals. Using thousands of human genomes and 53 high-quality nonhuman primate assemblies, we resolve 30 structurally distinct human amylase haplotypes and identify more than 15 lineage-specific expansions and contractions across primates. Integration with salivary gland RNA sequencing from multiple Old World monkeys reveals repeated, independent gains of salivary expression mediated by gene duplication. To directly test functional consequences, we generated a mouse model lacking salivary amylase and uncovered diet-dependent circadian and metabolic effects, including altered insulin secretion. Together, these data allow us to (i) resolve the mutational mechanisms underlying rapid structural evolution at the amylase locus, (ii) demonstrate convergent evolution of salivary expression across primates, (iii) detect signatures of positive selection among paralogs and within human populations correlated with dietary starch consumption, and (iv) expand the functional framework through which salivary amylase contributes to adaptation. Our results indicate that persistent structural variation at the amylase locus arises from the interaction of elevated mutational input, likely driven by mobile elements, and recurrent selection acting on the metabolic consequences of salivary expression, highlighting structurally complex genomic regions as versatile engines of evolutionary innovation.

[B042] A toxic gene duplication ignites a sulfide-powered metabolic engine in horses

Kaitlin Taylor[1], Joshua Eis[1], Gianni Castiglione[1]
[1]Vanderbilt University, , United States

The elite performance of the horse (Equus caballus) pushes mammalian physiology to its limits, creating a biophysical paradox: the pressures required for maximal oxygen transport frequently rupture the pulmonary capillaries, while the metabolic demands of continuous grazing risk gastric autolysis. Here, we show that horses resolve aerobic bottlenecks by weaponizing an anaerobic metabolic strategy—deriving power from a poisonous gas (hydrogen sulfide; H2S). We identify an Equus-specific duplication of the cysteinyl-tRNA synthetase (Cars1) locus that mirrors the functional architecture of sulfur-reducing bacteria and is coincident with the explosive adaptive radiation of Equidae. While a chimeric fusion transcript preserves canonical tRNA charging, a subfunctionalized paralog (CARS1-b) has lost this function yet serves as a high-flux "fuel injector" for hydrogen sulfide. By providing volatile sulfide species for the mitochondrion, CARS1-b creates a “biological gunpowder”, priming the cellular engine for explosive maximal energy production. Although this output is toxic in isolation—resembling the metabolic asphyxiation of human Down syndrome—horses harness it through a massively upregulated sulfide "combustion" chamber, mitochondrial sulfide: quinone oxidoreductase (SQOR). We recapitulated this bipartite engine in human cells, finding it sufficient to increase mitochondrial respiration. In horses, this system is strategically deployed to energetically demanding tissues, including the gastric mucosa and the pulmonary blood-gas barrier. Thus, the equine lineage has not merely tolerated a toxic gene duplication but domesticated it, co-opting a volatile anaerobic precursor to engineer the ultimate aerobic athlete.

[B043] Host plant sharing correlates with increased Horizontal Gene Transfer in Lepidoptera

Audrey Portal[1], Dr. Sara Oukkal[1], Prof. Daniel Janzen[2], Prof. Paul Hebert[3], Dr. Julien Varaldi[1], Dr. Sylvain Charlat[4]
[1]LBBE, Université Claude Bernard Lyon 1, Lyon, France, [2]University of Pennsylvania, Philadelphia, United States of America, [3]University of Guelph, Guelph, Canada, [4]PIMIT, Université de La Réunion, Saint-Denis, France

Evolution is traditionally represented as trees, emphasizing the vertical transmission of genetic material from parents to offspring. However, genetic material can also be exchanged across species boundaries, through a phenomenon called Horizontal Transfer (HT).
Although known to predominantly occur among unicellular procaryotic organisms, HT is increasingly well-documented in multicellular eukaryotes, thanks to the generalization of comparative genomics to non-model species. Still, molecular mechanisms, ecological drivers, and adaptative implications of HT remain poorly understood, highlighting the need for large-scale, quantitative analyses.

Lepidoptera have been identified as hotspots of HT, making them a promising system for investigating its drivers. Baculoviruses, highly prevalent viruses in Lepidoptera, may act as vectors of HT in this group, as they frequently harbour fragments of host DNA (particularly Transposable Elements). This leads to the testable prediction that species sharing host plants at larval stage, thus likely exposed to similar viral strains, should be connected by particularly high rates of HT.

Here, we combine extensive ecological and genomic data of 130 Lepidoptera species, derived from a long-term biodiversity survey on Costa Rican insect communities.
Leveraging clustering and phylogenetic approaches, we documented HT events between independent pairs of Lepidoptera insects sharing or not sharing host plants at larval stage.
Our preliminary results suggest a positive association of host plant sharing and HT frequency. Remarkably, this trend shows when considering transfers of Transposable Elements, and is absent when looking at transfers of non-mobile protein-coding genes. These findings are consistent with the hypothesis of baculoviruses as vectors of Horizontal Transfers.

[B044] Pangenomic analysis reveals extent and mutagenic impact of rare transposable elements in Arabidopsis

Aurélien Petit[1], Louna de Oliveira[1], Erwann Caillieux[1], Abdou Khadre Diouf[1], Vincent Colot[1], Pierre Baduel[1]
[1]Institut de Biologie de l'École Normale Supérieure, Université Paris Sciences et Lettres (PSL), UMR 8197 CNRS, U1024 INS, , France

Transposable elements (TEs), because of their ability to move across the genome, are major endogenous mutagens. However, mobile TEs represent only a small fraction of total TE content, and current knowledge of the mobilome largely relies on TE annotations from a single reference genome. While mobilome composition is expected to be conserved across individuals in panmictic species such as Drosophila, its divergence in species with strong population structure remains poorly understood. This is particularly relevant for the predominantly selfing plant Arabidopsis thaliana. To address this question, we leveraged 89 A. thaliana genome assemblies that we produced with Oxford Nanopore Technology (ONT) long-read sequencing and performed a pangenomic graph-based analysis of structural variants (SVs).

This approach identified more than 200 previously uncharacterized TE families showing recent mobilization at the species level. Among them, 122 have full-length copies in the reference genome but were either misannotated or completely missed. Although most of these families are ancestral, they are often absent as full-length copies in many strains and typically remain low copy when present. Nevertheless, collectively they contribute substantially to TE insertion polymorphisms at the species level, with 20% of these insertions undetected using the reference TE library alone. We confirmed the mobilization potential of some novel elements in epigenetic mutant backgrounds and found that their insertions frequently affect functionally relevant genomic regions. These results demonstrate that rare, previously undocumented TE families are a major source of large-effect mutations and a significant driver of genomic diversity in natural populations.

[B045] Ancestral architecture and evolutionary specialization of the vertebrate brain revealed by 3D single-cell transcriptomics in lamprey

Bing Su[1], Haixu Wu[1], Duoyuan Chen[2], Jun Li[3], Tao Zhou[2], Zhenkun Zhuang[2], Zhiwei Niu[2], Zeyu Du[3], Yongjie Chen[1], ShunQin Chuan[3], Chunyan Xu[2], Xun Liao[2], Xiaoyu Meng[1], Jiali Lu[3], Wenxue Cui[3], Youning Lin[2], Fubaoqian Huang[2], Kun Liao[2], Yan Liu[1], Tao Yang[2], Jing Chen[2], Hui Wang[2], Zhiqiang Dong[2], Longqi Liu[2], Xiaodong Fang[2], Xun Xu[2], Qingwei Li[3], Yue Pang[3], Shiping Liu[2]
[1]National Key Laboratory of Genetic Evolution and Animal Model, Kunming Institute Of Zoology, Chinese Academy Of Sciences, Kunming, China, [2]BGI Research, , China, [3]College of Life Science, Liaoning Normal University, Dalian, China

The lamprey, a jawless vertebrate with a primitive yet complex brain, occupies a crucial position for understanding vertebrate brain evolution. Using spatial transcriptomics and single-nucleus RNA sequencing, we generated a 3D molecular atlas of the entire lamprey brain. We identified 209 transcriptionally and spatially distinct cell clusters spanning 14 anatomically defined brain regions, including diverse neuronal, glial, and ependymal populations. The spatial organization of these regions, such as pallial subdivisions and the laminated olfactory bulb, mirrors patterns observed in jawed vertebrates, indicating that regional brain partitioning emerged in the last common vertebrate ancestor. Cross-species analyses revealed a continuum of neuronal specialization across vertebrate evolution. Lamprey neurons exhibit mixed molecular signatures, co-expressing excitatory and inhibitory markers, and likely represent ancestral prototypes of the specialized glutamatergic and GABAergic neurons found in jawed vertebrates. We further identified homologs of Purkinje and granule cells, suggesting a cerebellar-like architecture predates the emergence of the jawed vertebrate cerebellum. These findings illuminate the ancestral cellular architecture of the vertebrate brain and reveal early innovations that underpin its later diversification.

[B046] Genomic context mediate non-neutrality of synonymous mutations in Pseudomonas fluorescens under carbon limitation

Caio Gomes Tavares Rosa[1], Professor Rees Kassen[1]
[1]Mcgill University, Montreal, Canada

Synonymous mutations (SN) do not result in any changes to the protein sequence, therefore, they are often considered neutral. However, there is a growing body of evidence describing scenarios where SNs can change the quantity and/or quality of proteins, causing an improvement in fitness. One of those examples is the two independent SNs that arose in an evolution experiment propagating Pseudomonas fluorescens SBW25 under carbon limitation. These mutations were found in the gene gtsB, which is part of the gtsABCD operon encoding a putative glucose transporter. We found evidence that these mutations were under selection, and a later mutagenesis screening revealed that in that region, SN and non-synonymous mutations were equally capable of being adaptive. Further mutagenesis on the downstream genes gtsC and gtsD indicated that the non-neutrality of SNs was location-dependent. Our current hypothesis suggests that some mutations may affect transcription by generating illicit promoters and influencing the regulation of the gtsABCD operon. Synthetic upregulation of the different fragments of the gtsABCD operon had similar effects on fitness as the SN, but the effect was constrained by which genes were included, suggesting that increased protein levels are relevant only if they happen upstream of a certain region. Manipulation of the levels of the operon only known regulator, gltR, revealed a synergistic effect with some SNs. Altogether, by using this model case, we gained a better understanding of the underlying factors that determine the non-neutrality of SNs, the impact of regulatory networks on adaptation and their evolution over time.

[B047] Genome plasticity driven by transposable elements in ant-farmed fungi

Caio Leal-Dutra[1,2], Pepjin Kooij[3], Jonathan Shik[2], Gareth Griffith[4]
[1]Universidade Federal de Minas Gerais (UFMG), Belo Horizonte, Brazil, [2]University of Copenhagen, Copenhagen, Denmark, [3]Universidade Estadual Paulista - UNESP, Rio Claro, Brazil, [4]Aberystwyth University, Aberystwyth, Wales

Fungi domesticated by fungus-farming ants evolve under unusual population-genetic constraints, including clonal propagation, vertical transmission, and limited opportunities for recombination. These conditions are expected to weaken the efficiency of purifying selection and permit genomic changes that rarely persist in free-living lineages. One potential consequence of this regime is increased Transposable Element accumulation and persistence within the genome. In ant-fungal cultivars, TE enrichment has been proposed to interact with repeat-induced point mutation processes, potentially leading to pronounced CpG depletion—a mutational signature associated with cytosine deamination, biased mutation pressures, and TE-rich genomes. Here, we evaluate patterns of CpG depletion in ant-cultivated basidiomycetes by comparing genomes of fungal cultivars with those of their closest free-living relatives. Using non-overlapping 200 bp windows, we quantify genome-wide CpG Observed/Expected ratios and dinucleotide distributions. In one ant-cultivated genome, TEs comprise >70% of the assembly, and CpG depletion is pervasive, with almost half of windows lacking CpG entirely and exhibiting strongly reduced CpG O/E values. Analyses restricted to TE sequences in this genome reveal an even stronger depletion signal, with TE-derived regions showing an extreme skew toward zero CpG and very low CpG O/E values. In contrast, a free-living relative with substantially lower TE content (12.6%) exhibits near-neutral CpG composition across the genome. Together, these results are consistent with a model in which long-term TE accumulation under clonal propagation has contributed to pervasive CpG erosion and increased genome plasticity in ant-farmed fungi, shaping genome architecture during the establishment and stabilization of the mutualism.

[B048] Searching for shared genomic routes to termitophily in rove beetles (Aleocharinae)

Caroline Høegh-Guldberg[1,2], Eugene Myers[3], Taisuke Kanao[4], Thomas Bourguignon[3], Aleš Buček[1,3]
[1]Institute of Entomology, Biology Centre CAS, České Budějovice, Czech Republic, [2]Depertment of Zoology, Faculty of Science, University of South Bohemia, České Budějovice, Czech Republiic, [3]Okinawa Institute of Science and Technology OIST, Onna-son, Japan, [4]Yamagata University, Yamagata-shi, Japan

The ability to live in symbiosis with termites (termitophily) has evolved independently at least 40 times across Insecta, and is particularly prevalent within rove beetles (Aleocharine), where it has evolved tens of times convergently. Most termitophilous species exhibit one of two characteristic morphological adaptations to termitophily - physogastry or limuloidy - used to evade an aggressive response from their hosts. This project aims to use Aleocharinae as a model system to explore the genetic signatures of the convergent evolution of termitophily by means of comparative genomics. The dataset includes 9 free-living and 26 termitophilous Aleocharinae species covering six independent origins of termitophily. As most Aleocharinae are a few mm in size and thus contain little DNA, PacBio HiFi long-reads have been sequenced from 1-6 individuals per species. The HiFi reads have been assembled for each specimen and scaffolded using OMNI-C chromatin capture sequencing data. Of the 35 species, 18 have an N50 above 10Mbp. Preliminary assemblies suggest that Aleocharinae have relatively small genomes (150-720 Mbp) compared to other Staphylinidae (350-1400Mbp), with no significant difference in genome size between free-living and termitophiles ($p=0.973$). The heterozygosity appears to be significantly lower in termitophilous than in free-living Aleocharinae ($p=0.012$). In addition, latest analyses results on how gene family evolution and gene copy number variation are associated with the convergent evolution of termitophily will be presented.

[B049] Riding the heat wave: transcriptomic insights from yeast

Chloé Haberkorn[1], Dr Jennifer Molinet[1], Pr Rike Stelkens[1]
[1]Stockholm University, , Sweden

Extreme climatic events such as heat waves pose major challenges to species survival and have profound impacts on evolutionary processes. Plasticity is thought to buffer organismal stress, yet the molecular mechanisms underlying plastic responses remain poorly understood. In particular, the role of transcriptional plasticity and stress memory in responding to repeated stress events remains unresolved. Here, we experimentally exposed clonal populations of eight divergent Saccharomyces yeast species with different thermal tolerances to repeated heat waves. We compared their phenotypic and transcriptomic profiles after a few generations of mitotic growth, reflecting transcriptional changes. Warm-adapted species maintained higher growth than cold-adapted species across heat wave exposures. Thermo-generalist species showed intermediate outcomes with one species improving growth across repeated heat waves. To interpret transcriptomic results, we used a conceptual framework separating no-memory (gene expression independent from prior exposure) from memory-associated responses (expression modulated by prior exposure). No-memory responses showed conserved transcriptomic signatures of proteostasis induction, and reduced expression of ribosome biogenesis and translation upon repeated heat waves. Memory-associated responses were more rare and highly species-specific, showing opposite patterns of (de)sensitization in ribosomal and translational pathways in species at the two extremes of thermal tolerance. Together, our results show that thermal resilience can arise through alternative transcriptional changes and suggests that warm- and cold-temperature specialists adopt divergent gene regulatory strategies upon repeated heat waves. With climate change projections indicating more frequent and intense heat waves, understanding plastic responses across species with ecologically and genetically different backgrounds is crucial.

[B050] Transposons as telomeres: conflict and innovation in essential chromosome components

Jae Hak Son[1], Matthew Lawlor[1], Mahek Virani[1], Weihuan Cao[1], Mia Levine[2], Chris Ellison[1]
[1]Rutgers University, New Brunswick, United States, [2]University of Pennsylvania , Philadelphia , United States

The Drosophila telomere is one of the best-studied examples of active transposable elements (TEs) benefitting, rather than harming, a host genome, a phenomenon recently termed “TE addiction”. All Drosophila species lack telomerase, relying instead on head-to-tail arrays of specialized retrotransposons to maintain their chromosome ends. Though this relationship was initially described as mutualistic, recent findings suggest that telomere-specialized TEs evolve antagonistically with their host, despite their essential function. To better understand the coevolution of these TEs with their host genome, and “TE addiction” more generally, we have surveyed long-read assemblies of over 100 species of Drosophila, identifying nearly 400 telomeric TE families. Through phylogenetic and comparative genomic analyses, we find that these telomere-specialized elements evolve with remarkable speed and exhibit striking convergent evolution. We observe repeated losses of telomeric TE lineages and their protein-coding genes across the genus, alongside frequent horizontal transfer events between distantly related lineages. Furthermore, we identify a novel mechanism of TE evolutionary innovation: these elements have repeatedly captured host gene fragments to selfishly suppress the host piRNA pathway. Our results provide insight into the paradoxical nature of TE addiction and its role in genome plasticity while also revealing similarities between this phenomenon and facultative endosymbionts such as Wolbachia, Rickettsiella, and Spiroplasma.

[B051] Opsin phylogeny illuminates the origins of animal vision

Christopher Kay[1], Dr Filippo Nocolini[2], Prof Philip Donoghue[1], Professor Roberto Feuda[2], Prof Davide Pisani[1]
[1]University of Bristol, , United Kingdom, [2]University of Leicester, , United Kingdom

Opsins mediate light detection in most animals, and understanding their evolution is key to the origin of vision. Visual opsins emerged from the neofunctionalisation of a chemoreceptor to detect light. The evolutionary placement of this innovation among the early animal lineages, and the GPCR families they evolved from can shed light on the circumstances around the origin of vision, but the evolutionary distance between different GPCR families has to date obfuscated the precise nature of the opsin outgroups.

The GPCR superfamily containing opsins is one of the largest and most functionally diverse paralogous gene families in eukaryotes. We developed a method to produce and leverage family specific HMM profiles to specifically recover target GPCR families, and using this method combined with quartet analysis and AU tests were able to narrow down on the closest outgroups.

Our results reveal a complex GPCR landscape in early animals and suggest that previously established relationships between opsins and other GPCR families (and linked functions) need re-examination. Previous studies placed Placopsins (of unknown function) as the closest outgroup to opsins, followed by Melatonin Receptors. Instead, our results suggest that these two groups, together with GPRs 24 and 45, constitute the opsin outgroups, and the next closest comprises the Prostaglandin receptors. While most of these families are functionally uncharacterised, the distribution of known ligands among opsin-like proteins suggests that visual opsins likely evolved from receptors binding large lipid-like molecules, not compact heterocyclic ligands.

[B052] Pervasive translation of short open reading frames and de novo gene emergence in Arabidopsis

Claire Patiou[1,2], Christelle Blassiau[2], Isabelle Hatin[4], Dr. Lars A. Eicholt[1], Enora Corler[4], Chloé Ponitzki[2], Laurence Debacker[2], Dr. Pr. Erich Bornberg-Bauer[1,3], Dr. Olivier Namy[4], Dr. Sylvain Legrand[2], Dr. Vincent Castric[2], Dr. Eléonore Durand[2]
[1]Universität Münster, , Germany, [2]Université de Lille, , France, [3]Max-Planck Institute for Biology, , Germany, [4]Université Paris-Saclay, CEA, CNRS, , France

Ancestrally non-genic sequences are now widely recognized as potential reservoirs for the de novo emergence of new genes. Across clades, some de novo genes were proven to have substantial phenotypic effects, and to contribute to the emergence of novel biological functions. Yet, still very little is known about the starting material from which de novo genes emerge, especially in plants. To fill this gap, we generated Ribosome Profiling data from the closely related species Arabidopsis halleri, A. lyrata and A. thaliana and characterized genome-wide patterns of translation across them. Synteny analysis revealed 211 Open Reading Frames (ORFs) that have emerged de novo within the Arabidopsis genus and already exhibit signs of active translation. Most of these de novo translated ORFs were species- and even accession-specific, indicating their transient nature, with patterns of polymorphism consistent with neutral evolution in natural populations. They were also significantly shorter and less expressed than conserved Coding DNA Sequences (CDS), and their GC content increased with phylogenetic conservation. While most of them were located in intergenic regions and are thus newly discovered, 34 were previously annotated as CDS in at least one genome, and are promising putative genes. Our results demonstrate the abundance of translation events outside of conserved CDS, and their role as starting material for the emergence of novel genes in plants.

[B053] Mechanisms of evolution in the open ocean: clues from two populations of North Atlantic Ocean Physalia physalis

Dalila Destanović[1], Raymond Fedrick[1,2], Namrata Ahuja[1], Dr. Inma Herrera[3], Dr. Samuel Church[1,4], Dr. Casey Dunn[1]

[1]Department of Ecology and Evolutionary Biology, Yale University, New Haven, United States, [2]Okinawa Institute of Science and Technology, Okinawa, Japan, [3]Research Group in Biodiversity & Conservation (BIOCON), University Institute ECOAQUA, University of Las Palmas de Gran Canaria (ULPGC), Telde, Spain, [4]Department of Biology, New York University, New York, United States

Scientists have long assumed that open-ocean marine organisms show little population structure and species diversity at a global scale, due to large population sizes, high dispersal rates, and a lack of physical barriers. However, increasing access to genomic data has revealed more population structure in the open ocean than was previously thought. Work from our lab has shown that the hypermobile and cosmopolitan cnidarian Physalia physalis is more diverse than was previously expected, comprising at least four distinct species. It is unclear whether this biodiversity arose through isolation by distance, oceanic current patterns, adaptive divergence, reproductive barriers, or combinations of these processes. To study this, we analyzed populations of the North Atlantic P. physalis. We used WGS Illumina sequences of 61 specimens across this range to identify population structure, uncovering two genetically distinct eastern (Azores to Canary Islands) and western (Gulf of Mexico to Bermuda) populations. To understand the genomic basis of this differentiation, we looked into patterns of divergence across chromosomes. The variation was not uniformly distributed, with clear signals in chromosomes of lower and uniform Fst values, while others had extended regions with clear peaks of divergence. These patterns suggest that the separation between the two populations cannot be explained solely by isolation by distance or oceanic currents. Instead, additional evolutionary processes, potentially involving structural variation and localized selective pressures, likely contribute to population divergence. This study sheds light on mechanisms of diversification and speciation in non-model open ocean species without clear barriers to dispersal.

[B054] How much is enough for genomic surveillance of Escherichia and Klebsiella bloodstream infection-associated lineages, plasmids and AMR genes?

Dorottya Nagy[1,2,3], Valentina Pennetta[1,2], Katie Hopkins[1,5], Aysha Roohi[1,2], Dr Russell Hope[2,3], Dr David Williams[2,3], Dr Alice Ledda[3], Professor Julie Robotham[2,3], Dr Brown CS[2,3], Dr Walker AS[1,2,5], Dr Hopkins KL[2,6], The NEKSUS Consortium[2,3], Dr Samuel Lipworth[1,2,4], Dr Nicole Stoesser[1,2,4,5]
[1]Nuffield Department of Medicine, University of Oxford, Oxford, United Kingdom, [2]NIHR Health Protection Research Unit in Healthcare Associated Infections and Antimicrobial Resistance, University of Oxford in partnership with UKHSA, Oxford, United Kingdom, [3]Antimicrobial resistance (AMR) & Healthcare-associated infection (HCAI) Division, Chief Medical Advisor's Group, UK Health Security Agency, London, United Kingdom, [4]Oxford University Hospitals NHS Foundation Trust, Oxford, United Kingdom, [5]NIHR Oxford Biomedical Research Centre, Oxford, United Kingdom, [6]Antimicrobial Resistance and Healthcare Associated Infections (AMRHAI) Reference Unit, Public Health Microbiology - Reference Microbiology Division, Chief Scientific Officer's Group, UK Health Security Agency, London, United Kingdom

Escherichia and Klebsiella are major causes of bloodstream infections (BSIs) and subject to national surveillance in England(1). Optimal genomic surveillance sampling strategies are unclear. Using data from systematically sequenced E. coli/Klebsiella BSIs (NEKSUS study), we estimated sample sizes to capture representative genetic diversity at multi-locus sequence type (MLST), plasmids and AMR gene (ARG)-levels.

Ten regional NHS microbiology laboratories submitted all E. coli/Klebsiella BSIs, October 2023-March 2024. Isolates were sequenced (Oxford Nanopore Technologies), basecalled (Dorado v5.0.0 super-high accuracy), assembled(2), annotated (MLSTv2.23.0(3), Kraken2v2.1.3(4)), and plasmids clustered (PLINGv2.0.0(5)). Bayesian bootstrapping(6) with power calculation(7) was used to estimate the number of sequences required to achieve 80% sample coverage (i.e. the proportion of the population belonging to MLSTs/plasmid subcommunities/alleles observed in the sample).

2128/2584 (82%) isolates were collected and sequenced/assembled (1471 E. coli and 468 Klebsiella). To capture 80% of MLSTs diversity with 95% certainty, 1,425 (95% credible intervals [CIs]: 1,300-1,575) E. coli and 1,246 (1,150-1300) Klebsiella isolates should be sequenced. Similarly, 1,497 (1,301-1,761) E. coli, but fewer Klebsiella, 998 (809-1,197) are needed to capture 80% of plasmid diversity, while few samples are sufficient to capture 80% of ARG diversity (24 [21-27] E. coli and 67 [56-80] Klebsiella; Fig. 1).

Understanding how much genomic diversity is captured by sampling/sequencing strategies is key to designing affordable surveillance. In England, around 1,500 E. coli and 1,200 Klebsiella BSI isolates should be sequenced to capture ≥80% diversity at MLST/plasmid/ARG-level over 6-months. The unit of genetic diversity influences estimates, therefore calculations should be tailored to public health questions.

[B055] MTALTCO1 is a novel mitochondrial alternative protein that challenges conventional understandings of mitochondrial genomics

Francis Robitaille[1], Amy Campbell[1], Sarah Cornet[1], Liam Nyffeler, Xavier Roucou[2], Francis Robitaille[1]
[1]University Of Montreal, Montreal, Canada, [2]University of Sherbrooke, Sherbrooke, Canada

Mitochondrial alternative Open Reading Frames (ORFs) substantially broaden the functional scope traditionally attributed to mitochondrial DNA, encoding peptides and proteins that participate in diverse cellular processes. These newly identified ORFs are embedded within annotated sequences, both coding and non-coding, and reveal layers of overlapping genetic information. We here report the discovery of MTALTCO1, a 259 amino-acid protein, the longest mitochondrial alternative protein identified to date, encoded by an ORF located within the human cytochrome oxidase 1 gene, in the +3 reading frame. We confirm the expression and mitochondrial origin of MTALTCO1 through multiple independent lines of evidence, including a custom-designed antibody, mass spectrometry-derived peptides, sequence analysis, and inhibitors of mitochondrial expression. Despite encoding AGR codons as arginine, contrary to the prevailing view that these function invariably as stop codons in the vertebrate mitochondrial genetic code, MTALTCO1 shows strong evidence of mitochondrial translation, challenging established models of mitochondrial codon usage and gene expression.

Alternative ORFs embedded in mitochondrial coding sequences have until now prohibited, for conceptual and methodological reasons, any conservation analysis beyond pseudogenization assays. We present the first in-depth analysis of conservation at various phylogenetic scales and across a range of sequence features for this particular kind of ORF. Although the conservation patterns are atypical even within the de novo gene literature, there is nonetheless compelling evidence of selection, which argues against strictly ahistorical or purely causal-role-based interpretations and instead points to the need for a mitochondria-specific framework for de novo gene emergence and alternative protein function.

[B056] The Trehalose Trail- Evolutionary insights into trehalose metabolic pathways and tradeoffs

Ganesh Muthu Gangadharan[1], Dr Sunil Laxman[1], Dr. Aswin Sai Narain Seshasayee[2]
[1]Institute For Stem Cell Science And Regenerative Medicine (bric-instem), Bangalore, India, Bangalore, India, [2]National Centre for Biological Sciences, Bangalore, India

Trehalose is a non-reducing disaccharide widely present in natural environments and known to be synthesized and utilized by organisms across all kingdoms of life, where it functions both as a stress-protective molecule and a mobilizable carbon reserve. Despite its broad ecological presence and functional relevance, the evolutionary history of trehalose synthesis and utilization remains poorly understood. Recently, we reconstructed the evolutionary landscape of trehalose breakdown across ~4,000 prokaryotic and eukaryotic genomes, revealing near-universal conservation of trehalase across eukaryotes, supporting its presence in the Last Eukaryotic Common Ancestor, but for a complete loss in birds. In prokaryotes, these pathways show sporadic distribution, largely shaped by horizontal gene transfer. Building on this framework, we now investigate the evolution of trehalose synthesis to ask how and why organisms invest in producing this sugar. By examining six known trehalose biosynthetic pathways, we show that trehalose synthesis is broadly conserved across eukaryotes up to invertebrates but is entirely absent in vertebrates. Within invertebrates, we observe losses in several early-branching taxa, contrasted by consistent retention across insects which possess haemolymph-based systemic sugar circulation. Trehalose synthesis is present approximately in 70% of prokaryotes with a lineage-specific distribution marked by depletion in Firmicutes and enrichment in Actinobacteria. We aim to quantify the metabolic cost of trehalose synthesis and to determine the ecological and physiological contexts in which investment in stress-protective sugars becomes selectively advantageous. Together, these findings provide an evolutionary framework for understanding investment in stress-protective sugars.

[B057] Comparative Genomics of Domestication Across Laboratory and Semi-Wild Zebrafish Strains

Sitaram Rajaraman, James Ord, Andrew Maalouf, Fredrik Jutfelt, Irene Adrian-Kalchhauser[1]
[1]University of Bern, Bern, Switzerland

Zebrafish (Danio rerio) are a central vertebrate model in biological research, with laboratory strains originating from heterogeneous and often poorly documented domestication histories. This diversity provides an opportunity to investigate the genomic consequences of directed domestication and artificial selection in a vertebrate system.

We present a comparative population genomic analysis of five zebrafish strains spanning a gradient of domestication and genetic diversity: two widely used laboratory strains (AB, TU), two diversity-preserving strains derived from commercial or wild sources (MA, T5D), and a newly established semi-wild population (JU). Using shallow whole-genome resequencing of 95 individuals, including newly generated data for AB, TU, and JU and public datasets for MA and T5D, we quantified standing variation, population structure, and signatures of selection.

Measures of nucleotide diversity, allele frequencies, Tajima's D, and runs of homozygosity reveal genetic bottlenecks and elevated homozygosity in long-domesticated laboratory strains relative to recently derived populations. Selection scans identify candidate loci differentiating domesticated strains from JU, indicating both shared and strain-specific evolutionary responses. We further assess functional consequences of coding variation using protein structure prediction, focusing on epigenetic regulators and key developmental and physiological networks, including neural crest development, endocrine signaling, reproduction, and G-protein–coupled receptor pathways.

Together, our findings establish laboratory zebrafish as a tractable system for studying repeatability and contingency in molecular evolution under directed domestication.

[B058] Evolutionary strategies of bacteria adaptation to leaf surface

Jean-Baptiste Leducq[1]
[1]Université Laval, Québec City, Canada

The phyllosphere, the aerial part of plant, is a vast but nutrient-poor habitat that hosts a diverse community of bacteria, yet the evolutionary strategies enabling adaptation to leaf surfaces remain poorly understood. Hyphomicrobiales are among the most prevalent plant-associated bacteria and provide an ideal system to investigate genome evolution underpinning adaptation and diversification.

Using comparative and reverse ecology approaches, we show that different Hyphomicrobiales lineages rely on contrasting functional solutions to persist in leaf-associated environments. In Lichenihabitantaceae genomes and metagenomes, we identified conserved metabolic pathways involved in the utilization of two specific carbon sources found on leaves and in lichen, respectively, pointing to the ecological versatility of this group and its adaptation to nutrient-limited habitats. In contrast, Methylobacterium lineages exhibit highly dynamic genomes associated with its rapid diversification on leaves. Their success in the phyllosphere is underpinned by methylotrophy—using methanol released from plant cell wall demethylation as a primary carbon and energy source— and anoxygenic photosynthesis. We show that genes underlying methylotrophy and photosynthesis were frequently exchanged via horizontal gene transfer during Methylobacterium radiation and among phyllosphere-associated Hyphomicrobiales, reshaping genome architecture and obscuring phylogenetic signals.

Together, these findings highlight how bacteria adaptation to nutrient-limited habitats can arise through both the retention of specialized metabolic functions and flexible genome evolution, and emphasize leaf surfaces as key arenas for bacterial diversification.

[B059] Interplay between the role of DNA methylation in regulating gene expression and TE-silencing in a reptilian methylome

Joshua Hufton[1], Mats Olsson[2], Alexander Hansson[2], Malavi Sengupta[1], Arild Husby[3], Mette Lillie[1]
[1]Animal ecology, Department of Ecology and Genetics, Uppsala universitet, Uppsala, Sweden, [2]Department of Biological and Environmental Sciences, University of Gothenburg, Gothenburg, Sweden, [3]Evolutionary Biology, Department of Ecology and Genetics, Uppsala University, Uppsala, Sweden

DNA methylation is a major component of eukaryotic genomes, with a important role in the regulation of gene expression and repression of transposable elements (TEs). In vertebrate systems, TE transposition is silenced via hyper-methylation, which contributes to maintaining genome stability. Gene expression regulation is partially mediated by DNA methylation, with hypo-methylation in and around the transcriptional start sites (TSS) leading to higher expression. Mapping the methylome outlines the baseline landscape of DNA methylation across the genome, revealing where regulatory potential is encoded and identifying regions of epigenetic constraint. Here, we characterize the methylome of the squamate reptile, the sand lizard, by integrating short-read and long-read based methylation data and RNAseq data. We show that CpG islands in the TSS are often associated with hypo-methylation in the genome and higher gene expression. Within these regions, there was lower variance in methylation and lower explained variance in gene expression. Repetitive regions in the TSS were often hypo-methylated, and methylation was even lower in the presence of CpG islands in a TSS. Generally, TEs were highly methylated across the genomes. TEs in the TSS often leads to hyper-methylation and lower overall gene expression, independent of presence or absence of CpG islands. Overall occurrences of TEs in TSS was low but more frequent in genes without CpG islands present in their TSS. Patterns in DNA methylation and gene expression are heavily mediated by genomic features, and we see evidence of TE transposition causing higher methylation within the TSS and lower overall gene expression.

[B060] Towards an evolutionary understanding of the allotetraploid shamrock (lesser trefoil; Trifolium dubium)

Katie E. Herron[1], Dr Ann Mc Cartney[1,2], Dr Graham M. Hughes[1]
[1]University College Dublin, Dublin, Ireland, [2]University of California, Irvine, Irvine, USA
Polyploidy has played a central role in the evolutionary history of flowering plants, reshaping genome structure, gene content and repetitive DNA landscapes. The genus Trifolium (clover) provides a valuable framework for investigating these processes, encompassing multiple diploid and polyploid species of ecological, agricultural and cultural importance - including the shamrock, Trifolium dubium. Here, we take a comparative genomic approach across Trifolium, using publicly-available chromosome-scale assemblies to better understand how genome structure has evolved following divergence and hybridisation, with a particular focus on allotetraploid T. dubium. We integrate analyses of repetitive DNA, gene families and large-scale genome organisation to characterise patterns of conservation and divergence across species and between subgenomes. Comparative repeat analyses reveal variation in repeat composition, abundance and structural integrity among lineages, consistent with turnover and lineage-specific evolutionary trajectories. In parallel, gene-based comparisons highlight both shared ancestry and asymmetric patterns of retention and divergence following polyploid formation. Together, these results suggest that genome evolution in T. dubium reflects the combined effects of inherited genomic differences, post-polyploid restructuring and ongoing lineage-specific processes. This work establishes a comparative framework for understanding genome evolution in Trifolium, linking repeat dynamics, gene evolution and genome structure to the evolutionary history of this genus.

[B061] Agricultural Transition in Europe Is Associated with PGA Copy-Number decrease and a Functionally Distinct Haplotype

Kendra Scheer[1], Dr. Jaeyong Choi[2], Dr. Alber Aqil[3], Dr. Jong-Il Kim[4], Dr. Omer Gokcumen[1]
[1]University At Buffalo, Buffalo, United States, [2]Kangwon National University, Chuncheon, Republic of Korea, [3]Univeristy of Michigan, Ann Arbor, United States, [4]Seoul National University, Seoul, Republic of Korea

The Pepsinogen A (PGA) genes, which encode the primary digestive enzyme in the stomach, reside in a complex region of the human genome. Despite exhibiting copy-number variation comparable to the amylase locus, the origins and evolutionary dynamics of the PGA locus remain poorly understood. Using 540 long-read genomes and 98 patient samples, we resolve at least 42 distinct haplotypes, with PGA copy number ranging from 1-7 per haplotype. Transcriptomic data from two cohorts reveals a significant positive association between PGA copy number and gene expression ($p = 0.0359$). Analysis of 2,504 modern short-read genomes reveals continental differences, including a lower median copy number in West Eurasian populations and genome-wide significant population differentiation. Extending this analysis to 185 ancient genomes, we detect a decrease in PGA copy numbers coincident with the agricultural transition in West Eurasia (Wilcoxon $p = 3.8 \times 10^{-7}$). Despite extensive structural diversity, flanking regions exhibit a depletion of SNP variation, except for a haplotype in strong linkage disequilibrium with low copy numbers in non-East Asian populations. The frequency of this lineage increased from ~14% in European hunter-gatherers to ~33% in modern agricultural populations. These variants tag a haplotype lacking PGA3, a paralog that converts to active pepsin more slowly than other PGA genes, and rank among the strongest GWAS associations for other stomach-expressed genes, including CBLIF and LIPF. Together, these findings establish structural variation at the PGA locus as a functionally and evolutionarily relevant region in humans, likely shaped by long-term dietary transitions.

[B062] Comparative Transcriptomic Analysis of Chemosensory Recognition in Frankliniella occidentalis and Thrips palmi Karny

Kunhyang Park[1], MS Nakyoung Kim[2], Dr. Sugi Lee[1], Dae-Soo Kim[1,2]
[1]Korea Research Institute Of Bioscience And Biotechnology (kribb), Daejeon, South Korea, [2]KRIBB School of Bioscience, Korea University of Science and Technology (UST), Daejeon, South Korea
The western flower thrips (Frankliniella occidentalis) and the melon thrips (Thrips palmi) exhibit distinct host plant preferences, suggesting potential differences in their olfactory systems. To investigate whether variation in odor perception contributes to these ecological differences, we performed transcriptome sequencing and analysis of T. palmi. Using RNA-seq and de novo assembly, we identified 44,751 genes in T. palmi. For F. occidentalis, 48,889 genes were retrieved from publicly available RNA-seq data in NCBI, and a comparative transcriptomic analysis was conducted between the two species. Focusing on odorant-binding proteins (OBPs), which play a central role in insect olfaction, we identified and compared OBP gene families across both species. To further characterize the chemosensory system in thrips, we additionally analyzed antennal transcriptomes of F. occidentalis and whole-body RNA-seq data of Frankliniella intonsa. Comparative analyses revealed nine orthologous OBPs and four chemosensory proteins (CSPs) shared among the species. Among the four CSPs, three exhibited high sequence similarity, whereas one showed substantial divergence, particularly within the insect pheromone-binding protein domain. Likewise, of the nine orthologous OBPs, four displayed high sequence conservation, while the remaining five showed marked sequence divergence between species. These patterns suggest that diversification of OBPs and CSPs may underlie species-specific olfactory adaptations. Such molecular differences are likely shaped by host plant specialization and interspecific feeding competition, contributing to ecological divergence among thrips species.

[B063] The molecular origins and implications of segmental duplications across vertebrate genomes

Landen Gozashti[1], Runyang Nicolas Lou[1], Peter Sudmant[1]
[1]Department of Integrative Biology, University Of California Berkeley, Berkeley, United States
Segmental duplications (SDs) are large, low-copy genomic repeats that serve as important drivers of genome evolution and frequently harbor both adaptive and disease-causing loci. Nonetheless, SDs remain almost entirely unexplored outside of a handful of species primarily due to technical challenges associated with their assembly and annotation. Here, we leverage over 500 high-quality vertebrate genomes to explore the molecular origins and functional implications of SDs across diverse vertebrates. We find that waves of transposable element (TE) invasions are associated with SD expansions. Analysis of SD breakpoints suggests that TE-mediated mechanisms drive SD formation across species. Different clades show divergent patterns of SD localization. Some clades primarily exhibit tandemly repeated SDs, whereas others primarily display SDs that are interspersed across or between chromosomes. Nonetheless, genes within SDs are broadly enriched for multicopy families with adaptive functions and show signatures of diversifying selection. Together, our results suggest that TEs dictate the origin and evolution of SD formation, driving rapid changes in genome architecture which shape the distribution of unstable regions and hotspots of adaptive novelty.

[B064] Getting Something from Nothing in Archaea: De Novo Gene Birth Across the Tree of Life

Lars Eicholt[1,2], Tobias Warnecke[2,3]
[1]University of Muenster, Münster, Germany, [2]Department of Biochemistry, University of Oxford, Oxford, United Kingdom, [3]Trinity College, University of Oxford, Oxford, United Kingdom
De novo gene emergence is the evolution of new genes from previously non-coding DNA. As a result, de novo genes — and the natural de novo proteins they encode — typically lack detectable homology to known sequences. Once viewed as highly unlikely, de novo genes are now known to contribute to physiological functions across eukaryotes, including roles in spermatogenesis in flies, cell division in yeast, and development in plants. Despite this progress, evidence from bacteria remains comparatively sparse and suggests that genuinely novel genes arise less frequently and are less often associated with clear function. For archaea, systematic studies of de novo gene emergence are essentially absent, even though archaeal genomes contain many orphan genes without recognizable homologs. Identifying bona fide de novo genes in archaea would test whether de novo gene birth is a conserved evolutionary mechanism across the tree of life, expanding our understanding of how organisms explore sequence space and generate adaptive novelty. Beyond evolutionary insight, archaeal de novo proteins may offer practical advantages for experimental work, as archaeal proteins are often more stable and resilient than their eukaryotic counterparts. By integrating sequence- and structure-based homology searches, synteny analyses, and evolutionary modeling, we identified putative de novo genes across diverse archaeal genomes, supporting de novo gene emergence as a tree-of-life-wide mechanism for generating evolutionary novelty.

[B065] Ecological r-Selection Promotes a Proliferation-Plasticity Synergy Underpinning Cancer Drug Tolerance

Lu Yang[1], Dr. Xuemei Lu[1]
[1]Kunming Institute Of Zoology, Chinese Academy Of Sciences, Kunming, China

Phenotypic plasticity, the ability of genetically identical cells to adopt distinct phenotypic states, is a fundamental mechanism of adaptation across biological systems. By allowing populations to reversibly adjust to environmental fluctuations without genetic change, plasticity shapes evolutionary potential at both cellular and organismal scales. In cancer, this property enables tumor cells to survive therapy and re-establish growth, yet how ecological selection shapes such adaptive flexibility remains to be clarified.

We investigated this question using an experimental evolution framework, propagating isogenic HeLa populations for ~40 passages under contrasting selective regimes: r-selection (low-density, fluctuating environments) and K-selection (high-density, competitive environments). Integrating single-cell transcriptomics, chromatin accessibility, and analyses of published cancer datasets, we found that r-selected cells, traditionally viewed as fast-growing and fragile, exhibit higher transcriptional plasticity and broader drug tolerance than K-selected cells.

By modeling gene-expression landscapes as multidimensional energy surfaces, we show that r-selected populations maintain flexible, non-genetically constrained regulatory architectures that allow rapid state transitions. Comparative analyses across cancer types reveal that this proliferative–plastic phenotype recurs broadly, suggesting a conserved eco-evolutionary strategy that balances growth and adaptability.

Our results demonstrate that proliferation and plasticity can coevolve as complementary traits, linking classical r/K selection theory to cellular adaptation and providing a general ecological framework for understanding how populations persist under fluctuating environmental pressures.

[B066] The cost of altered translation accuracy shapes adaptation to antibiotics in E. coli

Laasya Samhita[1,2], Ms Sharvari Tamhankar[2], Mr Joshua Miranda[2], Dr Aabeer Basu[1], Dr Deepa Agashe[2]
[1]Ashoka University, Sonipat, India, [2]National Centre for Biological Sciences, Bengaluru, India

Protein synthesis, while central to cellular function, is error-prone. The resulting mistranslation is generally costly, but we do not know how these costs compare or interact with the costs imposed by external selection pressures such as antibiotics. We also do not know whether and how these costs are compensated during evolution. It is important to answer these questions, since mistranslation is ubiquitous and antibiotic exposure widespread. We quantified the growth cost of genetically increasing and decreasing mistranslation rates, and exposure to low antibiotic concentrations, in Escherichia coli. Mistranslation costs were generally lower than the cost imposed by antibiotics, and were exacerbated in a strain-specific manner under antibiotic exposure. All strains quickly compensated the antibiotic cost during experimental evolution, via antibiotic- and genotype- specific mutations. In contrast, mistranslation costs were significantly reduced only in some cases, without clear causal mutations. Control populations that evolved without antibiotics consistently compensated the cost of accuracy, and evolved increased antibiotic resistance as a by-product. Our work demonstrates that even when the cost of mistranslation is weak, altered translation accuracy can shape adaptive outcomes and underlying genetic mechanisms, with strong collateral fitness effects for apparently unrelated phenotypes such as antibiotic resistance.

[B067] Hybridization-derived gene copy number variation shapes genome stabilization and evolutionary novelty in a hybrid sparrow species

Qiujie Zhou[1], Kalle Tunström[1], Alex Lawrence[1], Dr Rachel Steward[1], Petronella Wessman[1], Anna Runemark[1]

[1]Department of Biology, Lund University, Lund, Sweden

Interspecific hybridization is a major driver of evolutionary novelty and biodiversity. Among the genetic changes associated with hybridization, gene copy number variations (gCNVs) are expected to be common in hybrid populations. Such gCNVs may arise through segregation of parental alleles that differ in gene copy number, genome instability following hybridization, and heterogeneous gene duplications that might contribute to the maintenance of hybrid fitness by permanently linking overdominant alleles. Hybridization-derived gCNVs may have substantial effects on hybrid fitness and potentially provide genetic substrate for novel phenotypes and adaptation. Despite their potential importance, the role of gCNVs in hybrid genome evolution remains poorly understood. Here, we use the Italian sparrow (Passer italiae), a naturally formed hybrid species with isolated island populations exhibiting divergent yet stabilized genomes, as a model system to address this knowledge gap. By integrating long-read and short-read sequencing data from over 200 individuals representing four hybrid lineages and their two parent species, we will comprehensively identify hybridization-derived gCNVs across the genome. Using this dataset, we aim to (1) characterize the distribution and genomic context of hybridization-derived gCNVs, (2) examine their contribution to hybrid genome stabilization, focusing on heterogeneous and parent-of-origin biased gene duplications, parallel gene copy number changes across lineages, and signatures of selection, and (3) assess their impacts on gene expression and evolutionary novelty. This study will advance our understanding of how hybridization-derived gCNVs contribute to genome evolution in hybrid species.

[B068] Going Retro: Mapping Invasive Retrotransposon Impacts on the Worm Genome

Sara Wighard[1], Dr Alejandro Burga[1]
[1]IMBA (institute Of Molecular Biotechnology), Vienna, Austria

Transposable elements (TEs) constitute a major component of eukaryotic genomes and can strongly alter genomic landscapes. While often detrimental, TEs can also confer adaptive benefits to their hosts. Notably, TEs are frequently horizontally transferred between species and are the biggest correlate of genome size variation between organisms. This ability to cross species barriers and reshape genomes positions them as powerful evolutionary drivers. However, much of our understanding on the effects of TEs remain correlative. I aim to develop an experimental paradigm to study these invasions using a vector in which TEs are heterologously expressed in the C. elegans genome. I will then examine how the TE propogates and the genomic consequences of this over many generations. I aim to quantify the impact on genome expansion as well as the effect on gene regulation. The small genome size and short 3-day generation time of C. elegans makes it an ideal system to examine these changes. To mimic natural horizontal gene transfer events, retrotransposons from invasive species will be targeted. For instance, active and recently expanded LINEs from the largest Caenorhabditis genome, C. japonica, have already been inserted and expressed in C. elegans. Ultimately, candidate TEs for insertion would be extended to more distantly related species. This system would allow us to examine the role of TEs on host genome expansion and gene regulation, as well as their propensity to be expressed in distantly related species. This comprehensive approach should deepen understanding of how biological novelty can arise from these invasive elements.

[B069] Dynamic evolution of retroviral envelope-derived sequences in primates

Koichi Kitao[1], Kirill Kryukov[2], Lihua Jin[3], Yuta Shintaku[4], Dr. Takashi Hayakawa[5], So Nakagawa[3]
[1]RIKEN Center for Integrative Medical Sciences, Yokohama, Japan, [2]Research Organization of Information and Systems, Mishima, Japan, [3]Tokai University School of Medicine, Isehara, Japan, [4]Japan Monkey Centre, Inuyama, Japan, [5]Hokkaido University, Sapporo, Japan

Several endogenous retroviruses (ERVs), remnants of ancient retroviral integrations, are known to retain envelope (env) genes encoding fusogenic proteins in primate genomes. Although most env genes have been degraded by various mutations, a small subset has been co-opted as protein-coding genes in hosts. However, the overall evolutionary dynamics of env sequences across primates remain poorly understood. To address this, we systematically screened and compared env open reading frames (ORFs) from 247 primate genomes. In total, we identified 8,683 nearly intact env-ORFs encoding proteins longer than 400 amino acids, with copy numbers ranging from 3 to 429 among primate species. Sequence similarity–based clustering revealed that previously reported functional env-derived genes generally exhibit low copy numbers and are conserved across multiple primate lineages. Notably, we identified an evolutionarily conserved env ortholog in tarsiers that shows low sequence similarity to other env-ORFs. This tarsier Env protein exhibited cell fusion activity in vitro, suggesting a potential fusogenic function in this lineage. In addition, we found that some co-opted env-derived genes appear to have lost their functions due to nonsense or insertion/deletion mutations in specific primate lineages. Given that many env genes are maintained at low copy numbers and have been reported to be subject to negative selection, the dynamic evolutionary turnover of env-derived genes may be driven by host–virus arms races, particularly because viruses and env-derived genes could share cell-surface receptors.

[B070] Predicting co-option of transposable elements from multi-omics signatures

Sofia Irene Garcia Bautista[1], Dr. Atlanta G. Cook[2], Reader in Bioinformatics Cei Abreu-Goodger[1]
[1]Institute of Ecology and Evolution, University of Edinburgh, Edinburgh, United Kingdom, [2]Centre for Cell Biology, University of Edinburgh, Edinburgh, United Kingdom

Transposable elements (TEs) have shaped mammalian genome evolution, contributing regulatory sequences, novel exons, and protein-coding material. Yet distinguishing which of the millions of TE copies are co-opted—versus neutral or silenced—remains an open problem. Previous studies have typically relied on individual features such as sequence conservation or expression, but these provide incomplete pictures: young elements may be silenced regardless of functional potential, while ancient elements vary widely in their contributions to host biology.

Here, I ask whether co-option can be inferred by integrating molecular signatures across multiple data layers. Using long-read RNA sequencing across human tissues—which enables accurate quantification of repetitive regions—I characterize TE-derived sequences with respect to splicing patterns, tissue-specificity, sequence conservation, epigenetic state, and evolutionary age estimated from divergence to subfamily consensus.

Because no single feature reliably distinguishes co-opted elements, I will develop a statistical framework that combines these signals to probabilistically classify individual TE instances. This approach aims to identify the molecular "fingerprints" that distinguish co-opted sequences from neutral or silenced ones. Ultimately, this work may clarify how repetitive elements transition from genomic parasites to host contributors across evolutionary time.

[B071] Population-scale variation and coordinated dynamics of transposable elements in Mediterranean fig (Ficus carica L.)

Tommaso Giordani[1], Marco Castellacci[1], Gabriele Usai[1], Flavia Mascagni[1], Alberto Vangelisti[1], Samuel Simoni[1], Lucia Natali[1], Andrea Cavallini[1]
[1]Department of Agriculture, Food and Environment, Pisa, Italy

Transposable elements (TEs) are major components of plant genomes and key drivers of genome evolution, however the determinants underlying intraspecific variation in TE abundance remain poorly understood, particularly at TE class or lineage level. Here, we present a population-scale analysis of the TE landscape in the Mediterranean fig (Ficus carica L.), one of the earliest domesticated fruit trees. Using whole-genome Illumina resequencing data from 286 Mediterranean fig genotypes, we quantified genome-wide TE abundance and variability and investigated their co-variation patterns.

On average, TEs accounted for 31.6% of the genome, with moderate variation among genotypes. Retrotransposons were the predominant component, with Gypsy long-terminal-repeat retrotransposons, especially Chromovirus-related families such as Tekay, representing the largest TE fraction, followed by Copia retrotransposons. DNA transposons, especially Helitrons, also contributed substantially to genome composition. Highly abundant lineages (e.g. Tekay, Helitron, CRM, Athila, Tat/Retand, Ale, Angela) showed relatively stable abundance across genotypes, whereas low-abundance lineages (e.g. Galadriel, Reina and Alesia) displayed higher relative variability. Correlation analyses revealed structured co-variation patterns, with strong positive correlations among Copia lineages and among Gypsy Chromovirus-related lineages, indicating that variation in these lineages tended to occur in a coordinated manner across genotypes. Positive correlations, although less pronounced, were also observed between major superfamilies, suggesting the occurrence of common constraints on the proliferation among TE classes.

Overall, this study provides the first comprehensive population-level analysis of TE abundance variation in fig and establishes a framework for investigating genetic control of TE dynamics in a perennial, clonally propagated crop.

[B072] Turnover and introgression shape the dynamic evolution of a sexual mimicry polymorphism shared across the genus Xiphophorus

Tristram Dodge[1], Molly Schumer[1,2]
[1]Stanford University, Stanford, United States, [2]Howard Hughes Medical Institute, Chevy Chase, United States

How functional variation is maintained within species remains a central mystery of evolutionary biology. Although directional selection is expected to rapidly drive alleles to fixation or loss, similar phenotypes sometimes segregate across multiple related species—a pattern unlikely to result from neutral processes. However, it often remains unclear if these phenotypic trans-species polymorphisms reflect a single ancestral origin and maintenance by long-term balancing selection, introgressive hybridization, or independent evolution. To distinguish between these hypotheses, we study a sexual mimicry color polymorphism shared among diverged swordtail fish (Xiphophorus) species. In most lineages, the sexual mimicry phenotype maps to small structural variants that drive cis-regulatory increases in the expression of kit ligand a (kitlga), a known pigmentation gene. In contrast, in other species, the same phenotype associates with a distinct 2 Mb repetitive region on a different chromosome. Using kmer-based approaches and long-read RNA sequencing, we narrow the candidate region to a unique zinc-finger protein, which is predicted to bind near kitlga and may regulate its expression in trans. These distinct genetic architectures likely represent turnover of genetic control via the evolution of new epistatic interactions. Comparisons of near telomere-to-telomere assemblies across the genus reveal substantial structural complexity at these loci, as well as patterns of allele sharing and divergence that are consistent with repeated introgression events and extensive structural evolution. Here, despite stability of a polymorphic phenotype across a genus, there has been unexpectedly dynamic molecular evolution, which may partly reconcile historically opposing perspectives on the prevalence of balancing selection.

[B073] Impact of Helitrons on evolution of multigene families

Vladimir Kapitonov[1], Monica Gould[2], Irina Arkhipova[1,2]
[1]Marine Biological Laboratory, Woods Hole, United States, [2]University of Chicago, Chicago, United States

Helitrons form a distinctive group of eukaryotic mobile elements (DNA transposons) that transpose via a rolling-circle or peel-and-paste mechanisms and are widespread in eukaryotes. Although Helitrons are prone to capturing host gene fragments, previously reported captures were short and nonfunctional. Using computational analyses of Helitron families in the bdelloid rotifers Adineta vaga and Adineta ricciae, we found that each genome contains four families of >10-kb nonautonomous Helitrons encoding two members from two different multigene families. One multigene family comprises >100 genes encoding homologous proteins with F-box and leucine-rich repeat (LRR) domains, a common eukaryotic architecture; in bdelloids these proteins are encoded by >20 short exons. The second multigene family is formed by >200 genes coding for homologous proteins composed of the VIP2 and tetratricopeptide repeat domains (TPR). The VIP2 domain is striking because it is present only in bacteria and bdelloid rotifers; in bacteria, VIP2 disrupts the insect cytoskeleton and can kill insect cells. All four Helitron families have evolved independently in A. vaga and A. ricciae very recently, given the ~98% pairwise identity of copies belonging to the same family. The current activity of the Helitron-encoded F-box-LLR gene in A. vaga and VIP2-TPR gene in A. ricciae is supported by complete RNA-seq read coverage. In old members of multigene families, links to Helitrons are virtually impossible to find. However, recent multicopy genes can be potentially linked to Helitrons. Our findings might be viewed as a “tip of the iceberg” suggesting deep involvement of Helitrons in evolution of multigene families.

[B074] Interlocus gene conversion causes mosaic divergence in tandem paralogues - modeling HMA4 evolution in Arabidopsis halleri

Yannick Schäfer[1], Dr. Yichen Zheng[1], Dr. Piyal Karunarathne[2], Akash Parida[3], Dr. Björn Pietzenuk[3], Dr. Vinod Kumar[3], Prof. Laura Rose[2], Prof. Ute Krämer[3], Prof. Thomas Wiehe[1]
[1]University Of Cologne, Cologne, Germany, [2]Heinrich-Heine University, Düsseldorf, Germany, [3]Ruhr University Bochum, Bochum, Germany

Following gene duplication, the evolutionary trajectories of gene copies can be shaped by various processes including mutation, selection, recombination and interlocus gene conversion (IGC). To explore dynamics and consequences of these processes, we developed a mathematical model that simulates the early evolution of recently duplicated, tandemly arrayed paralogous gene copies with positive selection on the new gene copy. We compared the results of our model to sequence variation in the three tandemly arrayed Arabidopsis halleri HEAVY METAL ATPase 4 (HMA4) gene copies, which are known to undergo IGC. We find that, the effective rates of IGC vary within genes, resulting in a mosaic pattern of divergence. Informed by empirical data, our model captures the impact of IGC on the diversity within each gene copy and the divergence among them. By tailoring the model to the AhHMA4 gene copies, we demonstrate the model's flexibility and its potential to provide insights into the evolutionary dynamics driving the evolution of tandemly arrayed paralogues. This study enhances our understanding of the balance between concerted evolution and divergence in the early evolution of gene families following gene duplication.

[B075] Evolutionary Dynamics of Adherence-Mediating Fimbriae in Pathogenic Bacteria

Gleb Ebert[1], Dr. Astrid von Mentzer[1]
[1]University of Gothenburg, Gothenburg, Sweden

Adherence factors play key roles in determining host and tissue specificity of bacteria. In gram-negative bacteria, fimbriae, in particular those assembled by the chaperone–usher (CU) pathway, often act as adherence factors. They are hair-like extra-cellular structures made of protein building blocks and are often important virulence factors in bacterial pathogens. Operons encoding CU fimbriae are highly diverse and are often located on mobile genetic elements, enabling spread to other bacterial strains through horizontal gene transfer (HGT). Despite the importance of CU fimbriae for virulence and as key vaccine targets, our understanding of their evolution and transmission dynamics is limited as research efforts focus on individual CU operons. This knowledge gap has immediate clinical implications, as demonstrated by the recent emergence of a new variant of CU fimbriae that rapidly increased in prevalence during an outbreak of diarrhoeal disease in Bangladesh, highlighting how CU evolution directly impacts disease epidemiology. The availability of millions of bacterial genomes (> 2.6 million bacterial genomes found in GenBank) and advances in machine-learning make comprehensive discovery feasible. We will map chaperone-usher operons across gram-negative bacteria using a machine learning approach for the first time. Previously unknown CU operons will enable us to reconstruct the evolutionary history of CU fimbriae in unprecedented detail. Furthermore, focusing on CU fimbriae found in the Escherichia/Shigella genera will allow us to characterise their transmission dynamics and model their interaction with molecules found on epithelia of clinically and agriculturally relevant hosts.

[B076] Structural variants and chromatin state: how does genetic architecture shape adaptation to climate?

Claire Mérot[1]
[1]CNRS - Université de Rennes, , France

Climate change is pressing species to their thermal limits and requires a comprehensive understanding of the full breadth of genomic diversity contributing to, or hampering, adaptive responses. A significant fraction of genetic diversity lies in structural variation (SV), such as chromosomal rearrangements or copy-number variants. Recent technological advances provide unprecedented insights into SV prevalence, functional consequences on epigenetic marks, gene expression and traits. To characterize and predict the contribution of SVs to climatic (mal)adaptation, requires that the scale at which SVs are studied increases from a few individuals to the population level, and the integration of comprehensive genetic analyses with the ecological and phenotypic context.

Here, I will reflect on the properties of SVs making them key actors of the response to rapid environmental stresses such as climate warming, using insights from experimental and ecological studies collected through a literature review. Then, I will present empirical results demonstrating how a combination of long-reads, linked-reads and short-reads facilitates the study of SVs at population-scale and along large climatic gradients in the seaweed fly Coelopa frigida. Our results highlight the relevance of SVs in parallel adaptation to climatic variation. In particular, large chromosomal inversions had a significant impact on gene expression and chromatin state, suggesting that their role in adaptation is not only mediated by recombination suppression but also by epigenetic factors. Altogether, we will discuss how improving insights in genome structure is opening new prospects in the study of adaptation and transforming our understanding of species response to environmental change.

[B077] Evolutionary Stasis Traps the Emperor Penguin in a Blue-Shifted Anthropocene Ocean

Nadir Dbouk[1], Isabella Bautista[1], Alexander Van Nynatten[2], Alice DuVivier[3,4], Kristen Krumhardt[3,4], Gianni Castiglione[1]
[1]Vanderbilt University, Nashville, United States, [2]University of Victoria, , Canada, [3]U.S. National Science Foundation National Center for Atmospheric Research, , , [4]Woods Hole Oceanographic Institution, ,

Climate change is rapidly altering the sensory landscapes of the Southern Ocean, yet the physiological capacity of species to track these shifts remains unknown. Here, we identify a "Sensory Trap" in the Emperor penguin (Aptenodytes forsteri) driven by the interaction between evolutionary stasis and sea ice loss. Ancestral sequence reconstruction reveals that the penguin ancestor evolved a "green-tuned" rhodopsin (λmax= 500nm) adapted to turbid coastal waters. Surprisingly, the deep-diving Emperor retained this ancestral phenotype (λmax= 499nm) rather than evolving the blue-shifted vision typical of deep-sea predators, as we observed in the sympatric Adélie penguin (Pygoscelis adeliae; λmax= 493nm). We propose this stasis in the Emperor was maintained by the optical properties of fast ice, which effectively creates a "green" spectral refuge at depth, in contrast to the blue-shifted, open-water niche exploited by the Adélie. Integrating oceanographic modeling with quantum catch simulations, we show that the projected loss of sea ice (SSP3-7.0) strips away this optical canopy, exposing penguins to a blue-shifted spectrum for which they are maladapted. This mismatch causes a 2.6% reduction in photon capture, a loss exacerbated by a critical "Warm-Blooded Penalty." The Emperor's high body temperature (39C) generates thermal noise ~650-fold higher than sympatric ectotherms, forcing their visual system to operate at a detector-limited threshold. In this regime, the 2.6% signal loss is non-linear, triggering a disproportionate ~3.4% collapse in prey detection probability. These results define a cryptic physiological tax on the Emperor penguin, suggesting that optical habitat loss will accelerate population declines beyond current demographic predictions.

[B078] When do genomic proxies reflect fitness? A mutation-based, fitness-explicit view of genetic load and adaptive potential

Alberto Garcia Jimenez[1], Cock Van Oosterhout[2], Dr Hernán Morales[3], Prof. Michael V. Westbury[1]
[1]Department of Health Technology, Section for Bioinformatics, Technical University of Denmark, Kongens Lyngby, Denmark, [2]School of Environmental Sciences, University of East Anglia, Norwich Research Park, Norwich, UK, [3]Globe Institute, University of Copenhagen, Copenhagen, Denmark
Despite well-established evolutionary theory and increasingly available genomic data, the relationship between genetic load, adaptive potential, and extinction risk remains unresolved. Conservation and evolutionary genomics commonly rely on genomic proxies such as heterozygosity, runs of homozygosity, and mutational burden, assuming they reflect realised fitness. Yet populations deviate from mutation–selection–drift equilibrium, and proxy–fitness relationships vary with dominance, epistasis, recombination, environment, and demographic history. This raises a fundamental question: to what extent do commonly used genomic metrics capture the fitness consequences of ongoing evolutionary change?

We address this question using a SLiM-based non-WF model that computes population fitness from per-mutation effects under demographic and environmental change, allowing alternative genotype–fitness mappings to be evaluated within a unified framework. The model separates intrinsic mutation effects from demographic processes, providing a controlled setting to assess genomic change and its fitness consequences.

To quantify genomic contributions to fitness, we introduce Fitness Equivalents (FE), a generalisation of the lethal equivalents concept that expresses genetic effects in a common fitness currency. This formulation considers deleterious and beneficial effects and enables mechanistic decomposition of net fitness change into contributions from segregating versus fixed load, masked versus purged deleterious variation, and adaptive responses from standing or new variation, facilitating comparisons across populations and models.

This framework offers a way to evaluate the limits of genomic proxies, reconcile paradigms emphasising genome-wide diversity versus strongly deleterious load, and clarify the demographic and evolutionary contexts in which genomic metrics may or may not relate to extinction risk, providing interpretive context for conservation genomics.

[B079] Genome Evolution in Coast Redwood (Sequoia sempervirens): First Insights from a New Haplotype-Resolved Assembly

Alexandra Sasha Nikolaeva[1]
[1]UC Berkeley, , United States

Genome evolution in one of the most ancient living lineages on Earth – the coast redwood (Sequoia sempervirens) – remains enigmatic due to the extreme size and complexity of its genome. Coast redwood is a rare polyploid conifer, with a genome exceeding 50 Gb and a clonal mode of dispersal – a reproductive situation, under which the genome of many individual plants may not have gone through a meiosis for thousands of years. Recent analyses of mutation accumulation patterns suggest that at least two of the six genomic copies have begun to diverge, suggesting that the genome might be in the early stages of diploidization.

Early versions of the redwood genome assembly were highly fragmented and collapsed, producing technical limitations for biological interpretation. At the time of the first assemblies, available phasing tools were developed primarily for diploid organisms, making polyploid genome phasing technologically challenging. Over the recent years, however, new approaches to polyploid genome phasing have emerged, combining long-read sequencing with chromatin-conformation (Hi-C) data to resolve complex structure.

In this talk, I present first insights from a new haplotype-resolved assembly of coast redwood genome and discuss genome evolution within the redwood lineage. I will also touch on phylogenetic relationships within the redwood family, which includes another ancient tree, giant sequoia (Sequoiadendron giganteum), using new data. These new results offer a framework for understanding why conifer genomes are typically highly conserved over deep evolutionary time and why redwoods may represent an exception to this rule.

[B080] Genetic diversity and genomic inbreeding signatures correlate with IUCN status across vertebrates

Amanda Gardiner[1], Vertebrate Genomes Consortium[2], Professor Richard Durbin[1,3]
[1]University Of Cambridge, Cambridge, United Kingdom, [2]Vertebrate Genomes Project, , , [3]Wellcome Sanger Institute, Hinxton, Cambridge

Anthropogenic activities are threatening global biodiversity. Genetics can help address this crisis by informing on changes in the demographics of species through space and time. Advances in whole-genome sequencing create new opportunities for demographic analysis, even based just on the two copies of a genome found in a single diploid individual. The Vertebrate Genomes Project (VGP) has the goal of generating high-quality, chromosome-level, haplotype-phased, and annotated reference genomes for all ~70,000 extant vertebrate species. In 2025 its first phase delivered 581, mostly diploid, assemblies spanning approximately 85% of vertebrate orders. Using these data, we quantified inter- and intra-species heterozygosity, runs of homozygosity (ROH), and inferred past effective population sizes (Ne) through the Pairwise Sequentially Markovian Coalescent (PSMC). Results showed a positive correlation between how threatened a species is, as indicated by their IUCN status, and the number of ROH, and a negative correlation between IUCN status and heterozygosity. Threatened species are more likely to exhibit long ROHs reflecting recent inbreeding, though striking ROH patterns are also seen in a range of other species. PSMC suggests frequent recent declines in Ne, with IUCN status negatively correlating with effective population size. We further quantified ROH patterns to see if IUCN status or other genetic and life history traits are correlated with ROH size and frequency, which are indicative of demographic history. Although further work is required, this work suggests that genomic signatures of inbreeding and low population size can be linked with the current IUCN status for a species.

[B081] Evaluating Genetic Diversity Indicators Across Wild and Domesticated Cotton Populations

PhD Ana Wegier[1,2], Valeria Alavez Gómez[1], Roberta Gargiulo[2], Dr. Alicia Mastretta-Yanes[2]
[1]Biology Institute, UNAM, Ciudad de México, Mexico, [2]Royal Botanic Gardens, KEW, Richmond,, United Kingdom

Current genetic indicators, including those adopted under the Kunming–Montreal Global Biodiversity Framework, require careful evaluation to ensure their effectiveness in monitoring and conserving in situ genetic diversity in species that demand integrated approaches, such as wild–domesticated complexes within a single territory. Partitioning these complexes into separate monitoring systems can conflict with local conservation planning, as illustrated by cotton (Gossypium hirsutum) in Mexico. Treating wild and cultivated components independently hampers accurate assessment of evolutionary threats and constrains conservation strategies, facilitating transgenic gene flow into wild populations within centers of origin and diversity.

Cotton exemplifies this complexity: wild, feral, traditionally managed, and domesticated populations form a continuum shaped by divergence times, geographic structure, and long-term human interactions. We present an integrated analysis of SNP data from 587 G. hirsutum individuals representing all continuum components and estimate effective population size (Ne). We combine Ne derived from molecular data with Ne inferred from census size (Nc) to evaluate how explicit consideration of each component improves the precision and reliability of genetic diversity indicators at local and regional scales.

Our results show that threats vary along the continuum. Gene flow from cultivated cotton can introduce domestication-selected traits into wild populations, while rapid turnover of commercial varieties erodes intra- and inter-varietal diversity. By integrating evolutionary and biocultural perspectives, we propose a framework for genetic indicators that conserves populations and the processes generating and maintaining diversity, strengthening conservation of cotton and safeguarding adaptive potential under environmental and socio-economic change.

[B082] A translocation associated with fertility problem and colour phenotype in cattle have been maintained at high frequency by balancing selection

Anna M Johansson[1], Martin Johnsson[1]
[1]Swedish University of Agricultural Sciences, Uppsala, Sweden

A translocation from chromosome 6 to chromosome 29 (called Cs29) in cattle is associated with the colour sidedness phenotype in several cattle breeds and have previously been reported to be associated with a fertility problem called gonadal hypoplasia in a Finnish cattle breed. We have showed that Cs29 is significantly associated with gonadal hypoplasia also in local cattle breeds from northern Sweden. In these breeds the colour sidedness is a desired phenotype that has been selected by humans and thus the both positive and negative effect of Cs29 is an example of balancing selection. We have also showed that Cs29 is still present at high frequencies in these breeds despite breeding efforts to reduce the fertility problem. There is also another translocation (Cs6) that gives the same desired colour phenotype but is not associated with gonadal hypoplasia. With simulations we have investigated how marker-assisted selection against Cs29 can affect the population. Our simulations showed that it is possible to reduce the fertility problem while maintaining the effective population size. This however comes with a cost of requiring many genetic tests. To further investigate Cs29, we have sequenced the breakpoint of the translocation in samples from different cattle breeds, and performed whole genome sequencing of Swedish mountain cattle who carry Cs29.

[B083] From bottlenecks to monitoring: SLiM5 simulations of genomic signals in managed Mauritian bird populations

Anna Stuart[1], Xuejing Wang[2], Cock Van Oosterhout[3], Prof Jim Groombridge[1], Dr Hernán Morales[2]
[1]Durrell Institute of Conservation and Ecology, University Of Kent, Canterbury,, United Kingdom, [2]Globe Institute, University of Copenhagen,, Denmark, [3]School of Environmental Sciences, University of East Anglia, Norwich Research Park, United Kingdom

The loss of genetic diversity is a core component of the biodiversity crisis with lasting consequences for evolutionary potential and population persistence. However, genomic erosion is often difficult to interpret from present-day data alone because of delayed genomic responses to demographic change. In addition, different historical trajectories could produce similar contemporary outcomes. To improve genome-based monitoring and management, we are developing forward-in-time 'digital twin' models using known demographic and management histories to generate quantitative expectations for genome-wide signatures through time.

We first focused on the Mauritius kestrel (Falco punctatus), which underwent an extreme bottleneck and intensive conservation management. Using SLiM5, we built an individual-based model that simulated a historically informed demographic trajectory and explicitly incorporated major management processes. The model was parameterised from species life-history data and empirical demographic records and was validated by its ability to reproduce observed population trajectories across replicated stochastic runs and to match empirical genome-wide patterns. Simulated heterozygosity and ROH-based inbreeding metrics showed close agreement with observed data, including the elevated long-ROH burden characteristic of heavily bottlenecked populations. We then used this digital twin model to explore counterfactual and prospective management histories (e.g. continued versus reduced intervention) and to quantify expected genomic trajectories under each scenario.

Embedding real conservation histories into calibrated 'digital twin' simulations provides a practical way to forecast demographic and genome-wide trajectories under alternative management strategies. It also highlights when apparent population rebounds might still mask slow genomic recovery.

[B084] Harnessing hybridization genomics to guide assisted gene flow in Central European white oak populations

Clara Groot-Crego[1], Sona Pisova[1], Adelin-Ionut Nicorescu[1], Gyula Kovacs[1], Alexandru Lucian Curtu[2], Petar Zhelev[3], Andrea Piotti[4], Camilla Avanzi[4], Sandro Kolbaia[5], Sandor Bordacs[6], Funda Özdemir Değirmenci[7], Zeki Kaya[8], Nikolaos Tourvas[9], Filippos Aravanopoulos[9], Heino Konrad[1], Christian Rellstab[10], Aglaia Szukala[1]

[1]Austrian Research Centre For Forests (BFW), Vienna, Austria, [2]Transilvania University of Brașov, Brașov, Romania, [3]University of Forestry, Sofia, Bulgaria, [4]Institute of Bioscience and Bioresources (IBBR), National Research Council (CNR), Firenze, Italy, [5]National Botanical Garden of Georgia, Tbilisi, Georgia, [6]Hungarian Institute of Agriculture and Life Sciences (MATE), , Hungary, [7]National Botanical Garden of Türkiye (NBGT) / TAGEM, , Türkiye, [8]Middle East Technical University (METU), , Türkiye, [9]Aristotle University of Thessaloniki, Thessaloniki, Greece, [10]Swiss Federal Research Institute WSL, Birmensdorf, Switzerland

Hybridization can foster ecological transitions and adaptation through the exchange of genetic variation between species. Assisted gene flow is increasingly considered in forest management to bolster genetic diversity and introduce climate-adaptive variation into threatened populations. The white oak species complex faces growing risk of maladaptation under future climates. Southeastern Europe harbors untapped white oak diversity adapted to warmer and drier conditions, yet lineage diversity, their evolutionary history, and the role of introgression in shaping climate adaptation remain poorly understood. Characterizing genetic diversity and introgression patterns is therefore key to informing seed selection and assisted gene flow strategies for Central European forests. Here, we integrate whole-genome re-sequencing and leaf-morphology data of 1,205 trees from the white oak species complex across pure and mixed populations spanning from Central Europe to Georgia. We quantify gene flow among lineages and identify candidate loci for climate adaptation through genotype–environment associations. We also examine adaptive introgression's role in shaping diversity and climate resilience. Our genomic data recovers clearly differentiated lineages corresponding to main European white oak species, yet reveals substantial introgression stemming from both ancient and ongoing hybridization. We detect climate-specific introgression patterns between a southern subspecies of Q. robur and Q. pubescens, as well as shared trichome phenotypes. These results suggest that subspecies may exhibit ecologically relevant differentiation partly shaped by introgressed genetic material. This study provides a framework for understanding lineage evolution and adaptive introgression, with direct implications for managing forest reproductive material under climate change.

[B085] Genomic signatures of load accumulation during parallel colonization of the Baltic Sea

Daniel Hancock[1], Manuel Arcieri[2], Dr Jakob Hemmer-Hansen[1]
[1]Technical University of Denmark, Silkeborg, Denmark, [2]Sapienza University of Rome, Rome, Italy
Founder events during colonization reduce effective population size (N_e), potentially increasing genomic load through weakened purifying selection. Conversely, increased inbreeding may expose recessive deleterious alleles, facilitating purging of strongly deleterious variants. The balance between these opposing forces determines whether colonization events lead to long-term fitness loss or resilience. The Baltic Sea, formed ~8,000 years ago and independently colonized by multiple marine fish species, provides a natural experiment to test these dynamics. We investigated patterns of demographic history, inbreeding, and genomic load across five species (Atlantic cod, Atlantic herring, European flounder, Lumpfish, Turbot) spanning the North Sea–Baltic Sea transition zone. Using whole-genome resequencing data, we reconstructed historical N_e, quantified runs of homozygosity, and estimated genomic load within genes inferred to be under purifying selection, classifying variants by functional impact (LOW, MODERATE, HIGH). Demographic reconstructions indicated founder events in most Baltic populations, accompanied by elevated signals of inbreeding. LOW and MODERATE impact mutations also tended to be elevated in these populations, consistent with drift load accumulation. In contrast, HIGH impact mutations showed less consistent patterns among populations, suggesting that purging of strongly deleterious alleles may partially offset the effects of reduced N_e in some cases. Populations lacking clear bottleneck signatures showed no evidence of load elevation at any functional impact. Finally, functional enrichment analyses revealed shared Gene Ontology categories associated with load accumulation across species. Together, these results suggest that parallel colonization can generate predictable signatures of drift load, while outcomes for strongly deleterious variation remain population-specific and context dependent.

[B086] A cross-species population genomic analysis of solitary bees

Dean Hodapp[1], Jannik Sven Möllmann[1], Dr. Thomas Joseph Colgan[1]
[1]Institute of Organismic and Molecular Evolution (iomE), Johannes Gutenberg University Mainz, Mainz, Germany

Understanding the capacity of organisms to respond to ongoing and future environmental challenges is a central goal of evolutionary and conservation biology. Solitary bees are ecologically and economically important pollinators, yet experience widespread declines: though they comprise over 75% of global bee diversity, they may be particularly vulnerable due to higher rates of floral specialisation, prolonged dormancy periods, and the absence of colony-level buffering mechanisms that can mitigate environmental stressors. Despite their importance, solitary bees remain understudied, with information currently lacking on their genetic variation, demographic history, and adaptive potential. To address this gap, we are applying whole-genome resequencing and population genomic-based analyses to determine the adaptive potential of solitary bees using representative species from three of the seven bee families. To provide insights into how these species adapt to changing local environments, we are investigating population structure and genetic variation harboured in contemporary populations to understand the extent of gene flow. In addition, we are analysing the extent and targets of recent selection, as conserved targets of selection may provide insight into genetic adaptation to common pressures in the local environment. Collectively, this study will represent one of the first cross-species population genomic analyses of solitary bees and thus greatly improve our understanding of their biology. Ultimately, these efforts will help aid the development of mitigation strategies and conservation schemes to protect this valuable, but vulnerable group of wild pollinators.

[B087] Epigenomic potential as an evolutionary buffer under extreme genomic erosion in the Apennine brown bear

Diana Lobo[1], Paolo Colangelo[2,3], Laura Gramolini[2], Paolo Ciucci[4], Ian Marc Bonapace[1]
[1]Department of Biotechnology and Life Sciences, University of Insubria, Busto Arsizio, Italy, [2]Research Institute on Terrestrial Ecosystems, Rome, Italy, [3]National Biodiversity Future Center , Palermo, Italy, [4]Department of Biology and Biotechnologies “Charles Darwin”, University of Rome La Sapienza, Rome, Italy

Small and isolated populations experience reduced adaptive potential due to pervasive genetic drift and weakened selection. Epigenetic processes may partly mitigate these constraints by facilitating rapid and reversible phenotypic responses to environmental change. The Apennine brown bear (Ursus arctos marsicanus), a critically endangered population restricted to the central Apennines in Italy, represents an extreme example of long-term isolation and low effective population size in Europe. Using 44 medium-high coverage genomes spanning all European brown bear populations, we evaluated how genomic erosion has shaped both genetic variation and epigenetic potential in this relict lineage. The Apennine bear genome exhibited exceptionally low heterozygosity and extensive fixation, including 21,748 SNPs uniquely fixed in this population and absent from ancestral species (polar and black bears). These variants overlapped 3,420 genes and were predominantly intronic (69.3%) or intergenic (15.8%), consistent with strong genetic drift. Nonetheless, relative to the genomic background, uniquely fixed SNPs were enriched for missense variants, affecting 123 genes with high- or moderate-impact variants and involved in neurodevelopmental, cognitive, and morphological functions. These patterns may underlie distinctive Apennine bear traits, such as smaller body size and reduced aggressiveness. To evaluate consequences for epigenetic regulation, we quantified SNPs that create or disrupt CpG dinucleotides. Apennine bears showed the lowest proportion of CpG-disruptive (17.5%) and the highest of CpG-creative (16.4%) variants among European populations, although fixed variants were disproportionately CpG-disruptive (23.9%). Overall, our results indicate that genomic erosion may progressively constrain epigenetic potential, while elevated CpG content may still support phenotypic plasticity under prolonged isolation.

[B088] Population Dynamics in North Asia since the Last Glacial Maximum

Elena Gusareva[1,2], PhD Tanghua Li[1], PhD Amit Gourav Ghosh[1,2], Ms Namrata Kalsi[1,2], Dr. Vladimir N. Kharkov[2,3], Prof. Tatiana V. Tatarinova[2,4], Prof. Vadim A. Stepanov[2,3], Pfor. Stephan C. Schuster[1,2], Prof. Hie Lim Kim[1,2]

[1]Nanyang Technological University, Singapore, Singapore, [2]GenomeAsia 100K consortium, Singapore, Singapore, [3]Research Institute of Medical Genetics, Tomsk National Medical Research Center, Russian Academy of Science, Tomsk, Russia, [4]University of La Verne, La Verne, USA

Since the Last Glacial Maximum (LGM), humans have been confronting changing environments, exploring new frontiers, and adapting to life across North Asia. The story of its prehistoric population dynamics, previously described as the “Siberian genetic vortex”, is among the most complex, shaped by repeated episodes of admixture and isolation that left enduring traces in the genetic makeup of contemporary population groups. Here, we present whole-genome sequencing data from 1,140 unrelated individuals representing 122 ethnic groups across North Asia and adjacent regions to reconstruct prehistoric population dynamics in the context of environmental change since the LGM. Demographic inference using MSMC and MSMC-IM analyses revealed multiple population divergence events occurred during the last deglaciation period, coinciding with rising temperatures between 20,000, and 6,000 years ago (ya). The latest population split ~6,000 ya shaped the contemporary Siberian population structure. Effective population sizes of several Siberian populations increased by the Holocene Climatic Optimum ~8,000 ya but declined sharply after ~6,500 ya, coinciding with increasing environmental stress and the expansion of early pastoralist groups from West Eurasia and East Asia. West Beringian populations remained genetically isolated until ~3,000 ya, maintaining consistently low effective population size and genetic diversity. Their unique demographic history makes them one of the most distinct populations in North Asia. Our study provides a broad retrospective on the population dynamics of North Asian since LGM, demonstrating the central role of climate change in shaping genetic structure of Indigenous communities in the region.

[B089] Dramatic changes in European Ash forests as result of a large-scale replacement of a native endophyte with its Asian relative

Erik Dahl Kjaer[1], Albin Lobo[1], Chatchai Kosawang[1], James Donnan[1], Jon Kehlet Hansen, Iben Thomsen[1], Lene Nielsen[1]
[1]Københavns Universitet, Frederiksberg C, Denmark

The Asian ascomycete Hymenoscyphus fraxineus was long known as a saprophyte on dead ash leaves with a life cycle similarly to its European relative Hymenoscyphus albidus that feed on European Ash (Fraxinus excelsior) leaves. However, once introduced to Europe, the alien ascomycete took over the ecological niche of H. albidus, where it - beside being a leaf saprophyte - became a severe pathogen killing millions of ash trees. The dramatic Ash Dieback made headlines across Europe, but a small fraction of the European ash trees revealed high levels of tolerance towards the new pathogen, suggesting potential recovery after natural selection in favor of tolerance among the naïve European ash trees. Based on studies since the Ash Dieback symptoms was first observed in Denmark 20 yrs ago, we report what we have learned about the host-fungus interactions, the genetics behind tolerance, and how observed tolerance translates into fitness advantages with corresponding evolution in ash forests. We reflect on general lessons to be learned in relation to management of forests for resilience towards future emerging infectious diseases.

[B090] Delineating drivers of genetic diversity and population structure in data-deficient deep-diving marine mammals

Fang Fei Tham[1], Dr Richard O'Rorke[1], Dr Rochelle Constantine[1], Dr Emma Carroll[1]
[1]School of Biological Sciences, University Of Auckland - Waipapa Taumata Rau, Auckland, 1010, Aotearoa New Zealand

The deep-sea is the most extensive habitat on the globe, harbouring high levels of biodiversity. Yet, little is understood of the drivers of evolutionary processes shaping the genetic diversity down below. Here we focus on two of the most commonly beach-cast marine mammals in Aotearoa New Zealand (NZ): pygmy sperm whales (Kogia breviceps) and goose-beaked whales (Ziphius cavirostris). Both species are poorly understood, deep-diving marine mammals with global distributions, however, are listed as “At-risk – Uncommon” in NZ based on the New Zealand Threat Classification System. Our samples are taonga (treasures) derived from a citizen science reporting system in collaboration with national government and local mana whenua (indigenous people of NZ). Stranding patterns of these data-deficient species offer insights into potential management units or populations. Pygmy sperm whales strand in distinct “hotspots” around Te Ika-a-Māui/North Island NZ, often stranding in cow-calf pairs, whilst goose-beaked whales strand intermittently throughout the entire NZ. We investigate and compare contemporary population structure and genetic differentiation using a genomic single nucleotide polymorphism dataset generated from double-digest restriction-site associated DNA sequencing (ddRAD-seq) and mitochondrial haplotypes. Our results indicate high levels of gene flow, with low levels of population structuring. Historical demography was inferred to examine effective population size trends over time and to investigate correlations to climate perturbations. Overall, we elucidate the drivers of genetic diversity and evolutionary processes to fill knowledge gaps of cryptic deep-diving species, where inadequate information prevents effective assessment for management and conservation strategies.

[B091] Whole-genome sequencing from mammalian snowprints

Harry Gordon[1], Paula Gardner[1], Róisín Ferguson[1], Luca Fumagalli[2], Dr Marta De Barba[3,4], Dr Anders Bergström[1]

[1]University of East Anglia, Norwich, United Kingdom, [2]Laboratory for Conservation Biology, Department of Ecology and Evolution, Biophore, University of Lausanne, Lausanne, Switzerland, [3]Department of Biology, Biotechnical Faculty, University of Ljubljana , Ljubljana, Slovenia, [4]DivjaLabs Ltd., , Slovenia

Effective genetic monitoring of populations is limited by the availability of high-quality samples, especially in elusive species. Snowprints represent an underexplored source of DNA that in areas with a snowy season is more frequently encountered than other commonly used sources. Previous studies have used mitochondrial DNA analysis and microsatellite genotyping on snowprints for species and individual level identification. Here we explore the efficacy of recovering whole genomes from snowprint samples, analysing DNA from 5 wolf and 4 bear snowprints. We find that endogenous DNA levels are generally very low, on the order of 1%, except in one wolf sample where it is ~60%. We recovered one high coverage wolf genome (27x) and 7 low (0.1-0.5x) and ultra-low (<0.1x) coverage wolf and bear genomes. Even for the low and ultra-low coverage genomes, population genetics analyses revealed useful information about individual ancestry. In addition to endogenous genomes, we recovered one high coverage canid pathogen genome from a wolf sample, and one low coverage cow genome from a bear sample. These results highlight the potential, but also limitations, of snowprints as a non-invasive source for genomic monitoring of wildlife and their ecosystems.

[B092] Fine-scale population structure and genetic diversity of the sugar kelp, Saccharina latissima, in Ireland

Jack Burton[1,2,3], Dr. Pamela Walsh[1,2], Dr. Heather Moore[4], Prof. Christine Maggs[3], Prof Paulo Prodöhl[3]
[1]Queen's Marine Laboratory, , United Kingdom, [2]QUB, School of Mechanical and Aerospace Engineering, , United Kingdom, [3]QUB, School of Biological Sciences, , United Kingdom, [4]Agri-Food and Biosciences Institute, , United Kingdom

Assessing genetic resources is essential for the conservation of species with both economic and ecological importance. Genetic assessments provide insights into a population's adaptive potential and, ultimately, its ability to persist in changing environments. Higher genetic diversity increases the likelihood of survival by providing the variability needed to tolerate a broader range of abiotic conditions.

Kelps are foundational species in temperate and cold-water coastal ecosystems. In Europe, kelp forests are dominated by Saccharina and Laminaria species, which deliver key ecosystem services that support coastal communities and the wider marine environment. These services include nutrient removal in eutrophic areas, the absorption of pCO2 that mitigates ocean acidification and warming, and the provision of nursery grounds for fish stocks.

Although global kelp decline is moderate, significant regional losses highlight the importance of local factors in determining kelp resilience to global change. Abiotic factors, including temperature, salinity, wave exposure, nutrient availability, pH, and pCO2, strongly influence kelp distribution and survival.

This study builds on previous research investigating the genetic diversity of sugar kelp (Saccharina latissima) populations along the coastlines of Northern Ireland and the Donegal region. By adopting a fine scale sampling approach and developing species-specific microsatellite markers, this work provides a more detailed understanding of population structure and connectivity in the area. These results will contribute to establishing a genetic baseline from future monitoring and will help inform biosecurity policy aimed at reducing the risk of genetic translocation along the coastline.

[B093] Plasticity across generations: DNA methylation correlates with intergenerational response to parasite infection in an endangered species

James Bazely[1], Dr Eugenie Yen[1], Alice Balard[1,3], Dr James Gilbert[1,3], Ms Kirsten Fairweather[2], Mr Artur Lopes[2], Mr Albert Taxonera[2], Professor Stephen Rossiter[1], Professor Christophe Eizaguirre[1]
[1]Queen Mary University Of London, London, United Kingdom, [2]Project Biodiversity , Santa Maria, Cabo Verde, [3]University College London, London, United Kingdom

Parasite infection represents a pervasive and often cryptic stressor in wild populations, yet our ability to detect, quantify, and compare host responses across individuals and generations is limited by a lack of reliable biomarkers. This constraint is particularly acute in long-lived and threatened species, where experimental approaches and repeated sampling are rarely feasible. Phenotypic plasticity is a key component of an effective host response to infection, and if such responses can be transmitted to offspring, this may confer an adaptive advantage. Epigenetic mechanisms such as DNA methylation are well suited to this role, as they integrate environmental exposure with stable and potentially heritable molecular signatures. Here, we investigate whether DNA methylation captures host responses to parasite infection and serves as a biomarker of infection status across generations in a wild population of loggerhead sea turtles infected with the parasitic worm O. margoi. We generated whole-genome DNA methylation profiles from blood samples of 29 nesting females and 116 of their hatchlings. We identified differential methylation between infected and uninfected females, including loci associated with immune-related genes. Hatchlings also exhibited patterns of differential methylation associated with maternal infection status, despite no direct exposure to the parasite. Loci identified as differentially methylated functioned as predictors of maternal infection status in both mothers and offspring, demonstrating the potential of DNA methylation as a biomarker of infection and maternally mediated immune priming. Together, these results highlight the potential of epigenetics to inform host–parasite interactions and act as a biomarker for cryptic infection in wildlife.

[B094] Using genomics to inform the conservation of Lake Tanganyika's top predator fishes (Lates spp)

Jessica Rick[1], Catherine Wagner[2]
[1]The University Of Arizona, Tucson, United States, [2]University of Wyoming, Laramie, United States
Top predators have oversized influence on food webs and ecosystem dynamics, and predator introductions or declines can have dramatic consequences for biodiversity. The four-species radiation of Lates fishes in Lake Tanganyika are important to the lake's incredibly biodiverse ecosystem and its fishery, which supports over 1 million people who live in its basin. The combined pressures of overfishing, habitat degradation, and climate change have reduced population sizes and body sizes in Lates species; however, it remains unclear the relative contributions of each of these forces and the best management actions to ensure the species' persistence into the future. Combining reduced-representation and whole genome data, and pairing these with population genomic, phylogenomic, and demographic modeling approaches, we characterize the evolutionary history and current population structure of these four species to better inform their management at a lake-wide scale. We find that the colonization of Lake Tanganyika by Lates fishes occurred 1-2 Mya, which is more recent than other major and diverse clades within the lake. Diversification of the clade into four endemic species occurred within the lake during a time of dramatic changes in lake levels driven by glacial-interglacial cycles, and multiple admixture events among Lates species during the mid- to late-Pleistocene indicate that hybridization was common historically (although not currently) among the four species. Finally, a lack of population structure in three of the four species indicates that management needs to occur on a lake-wide scale. Together, our findings suggest a dynamic and environmentally-linked evolutionary history of this predator radiation.

[B095] Integrating Genetics and Ecology to Assess the Extinction Risk of European Birds

Jiaqi Xu[1], Guojie Zhang[1], Shaohong Feng[1]
[1]Center for Evolutionary & Organismal Biology, Zhejiang University, Hangzhou, China

Understanding species extinction risk is a core goal of evolutionary biology and a pressing societal challenge. Traditional conservation assessments prioritize contemporary threats but overlook historical genetic legacies, underestimating risks exacerbated by climate warming's intertwined genetic-ecological impacts. In this study, we bridge this gap by integrating genomic, ecological, and climatic modeling to reconstruct evolutionary trajectories of 489 European bird species during the Last Glacial-Interglacial Cycle (LGIC). Our analysis revealed a continent-wide late-Pleistocene population decline, with 66.6% of species experiencing demographic contraction or severe bottlenecks. The severity of historical decline strongly predicted contemporary genomic erosion, and surprisingly, IUCN Least Concern species exhibited higher genetic load than threatened taxa, underscoring the prevalence of cryptic vulnerabilities. These vulnerabilities were shaped by an interplay of intrinsic functional traits and extrinsic interspecific competition, which together mediated species' responses to past climate shifts. Building on these insights, we developed a three-dimensional risk assessment framework incorporating genetic diversity, interspecific competition pressure, and trait-mediated climate vulnerability. This approach identified high-risk lineages, particularly within Passeriformes, and offers a transformative perspective for biodiversity conservation by bridging deep-time evolutionary history with contemporary ecological mechanisms.

[B096] Effect of landscape heterogeneity on the conservation of genetic diversity of Zea mays ssp. mexicana

José Carlos Moreno Juárez[1], Dr. Alicia Mastretta-Yanes[2], PhD Ana Wegier[3], Dr. Mariana Benítez Keinrad[4], Dr. Marcela Sandoval-Velasco[5], Dr Diana María Rivera Rodriíguez[6], Dr. Lino de la Cruz Larios[7], Dr. José de Jesús Sánchez González[7]

[1]Universidad Nacional Autónoma De México, Posgrado en Ciencias Biológicas , Ciudad de México, Mexico, [2]Royal Botanic Gardens, Kew. Nature-based solutions Team, Kew, London, United Kingdom, [3]Universidad Nacional Autónoma de México. Botanic Garden, Biology Institute, Ciudad de México, México, [4]Universidad Nacional Autónoma de México. Ecology Institute, Ciudad de México, México, [5]Universidad Nacional Autónoma de México. Center of Genomic Sciences, Cuernavaca, México, [6]Departamento de Ciencias Básicas, Tecnológico Nacional de México, Campus Tlajomulco, Tlajomulco de Zuñiga, México, [7]Universidad de Guadalajara. Centro Universitario de Ciencias Biológicas y Agropecuarias, Zapopan, México

Zea mays ssp. mexicana (teosinte) is a crop wild relative distributed across Mexico. This subspecies played a crucial role in maize adaptation to high altitudes and maintains admixture with maize to this day. Teosinte frequently co-occurs with maize in landscapes with intermediate perturbation; however, land use conversion and agricultural intensification have driven population declines. Using public GBS genomic data generated by the National Commission of Biodiversity (CONABIO) spanning the entire known distribution of Z. mays ssp. mexicana, we analyzed its genetic diversity and structure. We identified four populations consistent with previous descriptions: Chalco, Central Plateau, Durango, and Nobogame. The continuous, broadly distributed Chalco and Central Plateau populations exhibit higher genetic diversity and effective population sizes than the geographically restricted and isolated Durango and Nobogame populations. Consequently, we analyzed the scale of landscape effect on genetic diversity. We observed that higher genetic diversity is associated with high land-use heterogeneity. This suggests that heterogeneous landscapes can sustain larger populations of generalist species like teosinte and facilitate connectivity. This landscape genetics approach provides critical information on the scale at which public policy tools should be designed for the conservation of Z. mays ssp. mexicana and crop wild relatives.

[B097] Using historical butterfly specimens to track genomic erosion in clouded apollo populations in Sweden

Lars Littmann[1,2], Dr. David Díez del Molino[1,2,3,4], Prof. Chris Wheat[2,4]
[1]Centre for Palaeogenetics, Stockholm University, Stockholm, Sweden, [2]Department of Zoology, Stockholm University, Stockholm, Sweden, [3]Science for Life Laboratory, Stockholm University, Stockholm, Sweden, [4]Department of Genetics and Bioinformatics, Swedish Museum of Natural History, Stockholm, Sweden

The clouded apollo (Parnassius mnemosyne) experienced strong population decline linked to changes in agricultural practices throughout Europe in the last century. This has led to local extinctions, especially in Fennoscandia. In Sweden, the clouded apollo persists in the wild in two geographically isolated and differentiated populations that display low genetic diversity. We generate whole-genome sequences of historical specimens from museum collections, including samples both from within and from outside of Sweden, to shed light on the processes that account for the Swedish population's current genomic status. We compare previously published modern Swedish genomic sequences against their historical counterparts to describe the impact of recent population decline on the genetic diversity encountered in Swedish clouded apollo. In addition, we compare the historical Swedish population against historical populations that existed closer to the species' central range and closer to presumed glacial refugia in the Alps. This additional comparison provides more information about what degree of genetic diversity can be considered typical, thus allowing for a more accurate estimation of the impact of rapid population decline. Furthermore, by comparing across both space and time, we aim to disentangle the effects of recent population decline on the genetic diversity displayed by Swedish clouded apollo from factors such as the Swedish position at the periphery of the species' distribution and the species' post-glacial recolonization of Sweden.

[B098] Genome-wide signatures of drift in African wild dogs overcome within 10 generations following reintroduction

Laura Tensen[1], Xuejing Wang[2], Jessica Watermeyer[3], Cole du Plessis[4]
[1]University of Greifswald, Greifswald, Germany, [2]Globe Institute, Copenhagen, Denmark, [3]African Wildlife Conservation Fund, Chishakwe, Zimbabwe, [4]Endangered Wildlife Trust, Johannesburg, South Africa

Conservation translocations have become important assets in saving African wild dogs (Lycaon pictus) from local extinction, which have declined drastically due to anthropogenic pressures. In South Africa, wild dogs were eradicated except for a small, isolated population remaining in Kruger National Park. Due to reintroductions into private reserves, the country now holds a viable metapopulation of over 150 individuals that is used as a donor to repopulate other countries in southern Africa. This Range Expansion Project has become one of the most extensive efforts worldwide to recover population numbers of an endangered carnivore, allowing a unique opportunity to quantify the genomic effect of founder effects, population isolation, and conservation translocations. For this purpose, we resequenced 30 genomes of wild dogs from Kruger, private reserves, and outside protected areas. Demographic reconstructions indicate that populations were historically large (10,000-40,000 individuals) but began declining gradually ~500-100 kya, coinciding with Mid-Pleistocene climate shifts, followed by a sharp collapse ~0.8 kya. The negative correlation between nucleotide diversity and runs of homozygosity (ROH) suggests that low genetic variation is primarily driven by inbreeding rather than by demographic history. Reintroduced populations in private reserves exhibited the highest variation and lowest deleterious load. The short ROHs and close genetic affinity to an individual from Kenya further highlight the effectiveness of ongoing translocations and population admixture. The absence of drift and relaxed selection on trait variance in translocated groups imply that genetic resilience can be restored within 10 generations, providing valuable guidance for managing genetic variation in threatened species.

[B099] Immunogenetic Signatures Underlying Pathogen Tolerance Following a Severe Population Bottleneck

Leiting Yang[1], Cock Van Oosterhout[2], Prof Jim Groombridge[3], Dr Hernán Morales[1], Carolina Pacheco[4]
[1]Globe Institute, University Of Copenhagen, Copenhagen, Denmark, [2]School of Environmental Sciences, University of East Anglia, Norwich, United Kingdom, [3]Durrell Institute of Conservation and Ecology (DICE), University of Kent, Canterbury, United Kingdom, [4]Department of Biology, Lund University, Lund, Sweden

The Pink Pigeon (Nesoenas mayeri), an endangered species endemic to Mauritius, experienced a catastrophic population bottleneck in the 1990s, dwindling to approximately ten wild individuals. While conservation interventions have successfully increased population numbers, the species remains threatened by genomic erosion and high susceptibility to pathogens such as Trichomonas gallinae. Previous genomic studies have demonstrated an association between reduced genome-wide diversity and disease susceptibility, motivating a deeper investigation into the immunogenetic basis of host resilience.

Here, we combine whole-genome sequencing with a high-resolution annotation of immune-related genes to characterize patterns of immune diversity and their evolutionary dynamics. Using a multi-evidence immunogenomic annotation of the Pink Pigeon reference genome, we delineate immune loci at the level of genes, pathways, and functional modules, enabling detailed genomic-scale analyses of inmunogenetic diversity. We analyze a contemporary population from Île aux Aigrettes, integrating genomic data with fitness records —including lifespan and infection history— to identify immune loci and pathways associated with pathogen resistance.

By linking immunogenomic annotation with population genomic variation and individual-level disease data, this study provides a framework for understanding how immune diversity is structured and maintained in a severely bottlenecked species facing persistent disease pressure.

[B100] Genomic erosion and purging in populations of the Swedish sand lizards

Lucia Caroli[1], Tobias Uller[1], Nathalie Feiner[2]
[1]Lund University, Lund, Sweden, [2]Max Plank Institute for Evolutionary Biology, Plön, Germany
The sand lizard (Lacerta agilis) is a threatened species in Sweden, with an estimated 20-40% population decline over the past decades, primarily due to changes in land use leading to the loss of open sandy habitats. We use high-coverage whole-genome sequences from 45 populations across Sweden to identify the history of population contraction and expansion and quantify contemporary levels of inbreeding. We further test whether purifying selection can slow down the accumulation of deleterious variation in isolated and recently fragmented populations. The results reveal that populations at the northern range margin have very low genetic diversity, high inbreeding and show no evidence of purging, suggesting high extinction risk. Our results provide key demographic and population genomic information that can be implemented in conservation management strategies, as recent examples of habitat restoration have shown that successful recoveries are still possible.

[B101] Genomic connectivity, adaptation and conservation management of Australian little penguins

Lucy Ockert[1], Dr Joanna Day[2], Emeritus Professor William Sherwin[1], Professor Lee Rollins[1]
[1]University of New South Wales, Kensington, Australia, [2]Taronga Conservation Society Australia, Mosman, Australia

Little penguins (Eudyptula minor) are an iconic species with specialised adaptations to the marine environment, yet are in decline throughout Australia. Some geographically marginal populations have declined by up to 98% in recent decades, despite strongholds that remain in the centre of the species' range. Escalating climatic extremes, emerging disease threats such as highly pathogenic avian influenza, and penguins' exceptionally low evolutionary rate could jeopardise the species' capacity to adapt rapidly enough to mitigate further declines.

Using the most geographically comprehensive genomic dataset for Australian little penguins to date, we applied a genomic approach to address key conservation challenges among in-situ and ex-situ populations. We characterised population structure, connectivity, and genetic diversity of in-situ populations to identify biologically meaningful management units, assess gene flow across Australia, and examine the impact of drastic declines on standing genetic variation. We identified genomic signatures of selection associated with environmental gradients known to impact breeding success, such as extreme heat and wind events. Additionally, we developed the first seabird pangenome to comprehensively assess genome-wide variation and examine immune gene diversity in little penguins, relevant to emerging disease threats. In parallel, we evaluated the conservation value of the ex-situ metapopulation while clarifying the relatedness and diversity of these individuals, and established guidelines for integrating genomic data into existing studbooks.

Collectively, these approaches provide critical insights into the evolutionary processes and adaptive capacity of little penguins, delivering genomics-informed recommendations to support conservation strategies for little penguins in a rapidly changing climate.

[B102] The genetic basis behind highly admixed ecotypes: the case of boreal and eastern migratory caribou

M. Lisette Delgado[1], Micheline Manseau[2], Cory Fournier[2], Daria Martchenko[1], Rebecca S. Taylor[2], Kirsten Solmundson[4], Arthur R. Rodgers[3], Janet Jackson[3], Vicki Trim[4], Joanie Kennah[4], Dr. Paul J. Wilson[1]

[1]Trent University, Peterborough, Canada, [2]Environment and Climate Change Canada, Ottawa, Canada, [3]Ontario Ministry of Natural Resources, Thunder Bay, Canada, [4]Government of Manitoba, , Canada

Ecotypes can be defined as phenotypes within a species distinguished by differences in behavior and/or life history. These differences are assumed to hold important adaptive variation, but for many species, the genetic basis behind ecotypes is not completely understood. Identifying these genetic differences can be key for the management, monitoring, and conservation of ecotypes, particularly for ecotypes which differences are rooted mainly in their behavior, exhibit high gene flow, and which habitats are threatened by anthropogenic activities. Here, we focused on eastern migratory and boreal caribou (Rangifer tarandus) from Ontario and Quebec (Canada). These ecotypes with overlapping distributions are differentiated by their calving strategies and conservation statuses. Whole genome analysis using 109 high coverage and 222 low coverage genomes revealed that caribou are more genetically similar to geographically close individuals than to caribou with the same ecotype. Despite high interbreeding levels between ecotypes, admixture analyses detected two ancestral components in each province, indicating a genetic basis of the ecotypes. Association analysis identified multiple small regions that significantly distinguished the ecotypes, but these regions were different in each province. Despite the absence of large structural variants (SVs) or supergenes, results showed that an SNP-only approach is not sufficient to distinguish highly admixed ecotypes. Other SVs, particularly deletions, differentiated between the two ecotypes. Our findings reveal a larger overlapping zone in Ontario, a recent and independent evolution of eastern migratory in both provinces, and suggest tailored management strategies in each province.

[B103] From deserts to forests: sequencing of the giant forest hog completes the genomic history of the African Suids

Marta Maria Ciucani[1], Sabhrina Gita Aninta[1], Nuno Filipe Gomes Martins[1], David A. Duchêne[2], Xi Wang[1], Ignacio A. Lazagabaster[3,4], Xiaodong Liu[1], Frederik Filip Stæger[1], Mikkel Schubert[5], Thomas Bøggild[1], Shixu He[1], Zilong Li[1], Rafael Reyna[6], Erik Meijaard[7], Thomas M. Butynski[8], Vincent B. Muwanika[9], Charles Masembe[9], Sylvain Dufour[10], Philippe Gaubert[11], Hans R. Siegismund[1], Ida Moltke[1], Anders Albrechtsen[1], Rasmus Heller[1]

[1]Department of Biology, University of Copenhagen, Ole Maaløes Vej 5, 2200 Copenhagen N, Denmark, , Denmark, [2]Department of Public Health, University of Copenhagen, Øster Farimagsgade 5, 1353 Copenhagen, Denmark, , Denmark, [3]Centro Nacional de Investigación sobre Evolución Humana CENIEH, Burgos 09002, Spain, , Spain, [4]Department of Evolution, Ecology and Behaviour, University of Liverpool, Liverpool L69 3BX, United Kingdom, , United Kingdom, [5]Novo Nordisk Foundation Center for Basic Metabolic Research, University of Copenhagen, Copenhagen, Denmark, , Denmark, [6]El Colegio de la Frontera Sur, Lerma, Campeche, México, 24500, , México, [7]Borneo Futures, Bandar Seri Begawan, Brunei Darussalam, , Brunei Darussalam, [8]Eastern Africa Primate Diversity and Conservation Program, Nanyuki 10400, Kenya, , Kenya, [9]College of Agricultural and Environmental Sciences, Makerere University, Kampala, Uganda, , Uganda, [10]SYLVATROP Consulting, Conches-sur-gondoire, France, , France, [11]Université de Toulouse, Toulouse INP, CNRS, IRD, Centre de Recherche sur la Biodiversité et l'Environnement (CRBE), Bâtiment 4R1, 118 route de Narbonne, 31062 Toulouse Cedex 9, France, , France

The giant forest hog (GFH, Hylochoerus meinertzhageni), the largest and most elusive wild pig, is found in a disjunct range across equatorial Africa, inhabiting forests and woodlands. The GFH is morphologically classified into three subspecies but currently understudied. Here, we present the first whole-genome data of nine GFHs, from Guinea and Uganda, representing the two geographically most distant subspecies. We resolve the placement of the GFH among African suids as sister to the warthogs, with divergence around 4.6 Mya, and the two GFH populations splitting at least 0.5 Mya before following distinct demographic trajectories. The deep evolutionary split is confirmed by species level genetic differentiation value and the lowest heterozygosity level in the Guinean population compared to all the African suids, probably as a result of the long-term low Ne and isolation.

Our findings have implications for understanding the consequences of habitat change in African rainforests and reveal a very deep, previously underappreciated evolutionary divergence within the GFH that may affect its conservation.

[B104] Genetic diversity, census size, conservation status and the efficacy of purifying selection across plant species

Tongzhou Tao[1,2], Martin Lascoux[2], Professor Jun Chen[1]
[1]Zhejiang University, Hangzhou, China, [2]Uppsala University, Uppsala, Sweden
In the present study we leverage genomic data from a unique set of 21 species with well documented small population size and conservation status that belong to the Chinese Plant Species with Extremely Small Populations (PSESP) program. The latter is an initiative aimed at monitoring the status of threatened plant species in Southwest China. To this core genomic dataset we added genomic data from 80 plant species, for which indirect estimates of census number were obtained. We used this dataset to assess the relationship between genetic diversity, conservation status, census size, purifying selection efficacy and different effective population size estimates. While some of the effective population sizes are informative on conservation status, demographic parameters may be favoured for species in danger of extinction.

[B105] Immune Signatures of Gastrointestinal Helminth Infection in Transcriptomes of European Bison

Mateusz Konczal[1], Marta Kołodziej-Sobocińska[2], Rafał Kowalczyk[2], Prof Jacek Radwan[1]
[1]Adam Mickiewicz University in Poznan, , Poland, [2]Mammal Research Institute, Polish Academy of Sciences, , Poland

Understanding the genes and molecular pathways that shape host responses to infection is essential for advancing our knowledge of host–parasite interactions and their ecological and evolutionary implications. Such insights are especially valuable for conserving endangered species that may be vulnerable to emerging or novel pathogens. Here, we investigated the impact of the recently introduced gastrointestinal nematode Ashworthius sidemi on gene expression in its novel host, the European bison (Bison bonasus). We analyzed abomasal transcriptomes (n = 45) from individuals characterized by a wide range of infection intensities (0–8,620 parasites per host). Despite substantial variation in parasite burden, differential expression analyses detected no individual genes significantly associated with infection intensity. However, gene set enrichment analyses based on p-value distributions revealed multiple immune-related gene ontology categories, including B and T cell activation, neutrophil chemotaxis, inflammatory responses, and regulation of IL-6 production. These findings indicate that European bison mount a clear yet subtle transcriptional response to A. sidemi infection and highlight molecular pathways potentially involved in mediating host defense against this emerging parasite. Ongoing work will assess whether genomic erosion in these functionally important genes has occurred by comparing contemporary and ancient bison populations.

[B106] Large scale, low coverage population genomics assessing genetic variation and population structure across vulnerable Swedish sand lizard populations

Mette Lillie[1], Patrik Rödin Mörch, Erik Wapstra, Sven-Åke Berglind, Jacob Höglund, Mats Olsson
[1]Uppsala University, , Sweden

Genomic data is revolutionizing our understanding of how evolutionary processes shape genetic diversity, local adaptation, and extinction risk in threatened species. Here, we apply a large-scale, low-coverage whole-genome sequencing approach to investigate the evolutionary and demographic history of the Swedish sand lizard (Lacerta agilis), a habitat specialist persisting in fragmented populations across northern Europe. Using data from over 100 individuals sampled across Sweden, we infer patterns of population structure, diversity, and genetic load. We reveal strong genomic differentiation among regional populations and a pronounced latitudinal decline in genetic diversity, consistent with postglacial range expansion followed by drift and habitat-driven isolation. Several peripheral populations show signatures of elevated realized genetic load, reflecting historical bottlenecks and limited gene flow. The relict Sörmon population shows deep genomic divergence even from its nearest neighbors, suggesting a distinct evolutionary trajectory and potentially high conservation value. Moreover, northern populations display high load and small effective sizes, indicating higher vulnerability to ongoing environmental change. Our results illustrate the power of population genomic data in uncovering the combined effects of demographic history and recent fragmentation on genome-wide variation and mutational load. Our findings underscore the value of integrating evolutionary and conservation genomics to guide management actions for species in changing landscapes.

[B107] Joint inference of demography and selection from genomic time series

Paul Bunel[1], Mathieu Uhl, Miguel De Navascues[1], Simon Boitard[1], Bertrand Servin[1]
[1]Inrae, Montpellier, France

Genomic samples collected from a single population over several generations offer direct access to the genetic diversity changes occurring within a specific time period. This provides information about both demographic and adaptive processes acting on the population during that period, making genomic time series particularly valuable for the monitoring of vulnerable or declining populations.
A common approach to analyze such data is to model observed allele counts in finite samples using a Hidden Markov Model (HMM) where hidden states are true allele frequencies over time.
While several methods following this approach have been developped, they generally focus on estimating either the effective population size N (neglecting selection) or selection coefficients (s) along the genome (considering N as know). Thus, these two questions are considered sequentially, if not independently, of each other.
Here I will show that estimations of N obtained from genomic time series may be biased in the presence of selection. In order to solve this issue, I will then present several possible strategies for the joint inference of N and s, and show their performance on simulated data.

[B108] Comparative population genomics of oil-collecting bees in the highly fragmented Brazilian Cerrado biodiversity hotspot

Paulo Sousa[1], Dr. Belinda Kahnt[1]
[1]Martin-Luther-University Halle-Wittenberg, , Germany

Understanding how life-history traits shape population genetic pattern is crucial for species conservation in a rapidly changing world. The Brazilian Cerrado is a global hotspot for bee diversity, including oil-collecting bees, but is experiencing some of the highest rates of habitat loss and fragmentation worldwide. Oil bees of the genus Centris and Epicharis provide an ideal system to study how life-history traits such as body-size and diet breadth shape their population genomic response to such habitat alterations. Using whole-genome sequencing (WGS) and a novel approach to handle mixed-ploidy datasets, we investigated the population genomics of five Centris and two Epicharis species, ranging from extreme oil specialists to generalists and small/medium (ITD: $\bar{x}$= 4.14 mm) to large-bodied species (ITD: $\bar{x}$= 9.49 mm), across twelve locations with varying levels of fragmentation in the Brazilian Cerrado. We hypothesised that smaller species and/or oil specialists would show a lower genetic diversity (Nei's H), higher population genetic differentiation (FST) and more structure than larger species and/or oil generalists, rendering them more vulnerable to habitat loss and fragmentation. Interestingly, although genetic diversity varied among species, populations of all species showed little genetic differentiation and a general lack of population genetic structure. These results suggest that high dispersal ability may currently buffer these oil bees against the genetic effects of fragmentation. Nevertheless, continued habitat loss may erode this resilience over time, highlighting the importance of conserving and restoring habitat connectivity to ensure the long-term persistence of these enigmatic oil bees.

[B109] Harbour porpoise whole genomes document a rapid radiation across the North Atlantic and guides management units

Morgan McCarthy[1,2], Jonas Teilmann[3], Rune Dietz[3], Signe Sveegaard[3], Magie Aiken[1], Iben Stockholm[4], Elena Gladilina[5], Karina Vishnyakova[6], Nicolas Dussex[1], Pavel Gol'din[7], Morten Tange Olsen[2,3]
[1]Swedish Museum Of Natural History, Stockholm, Sweden, [2]Globe Institute, University of Copenhagen, Copenhagen, Denmark, [3]Aarhus University, Roskilde, Denmark, [4]University of Copenhagen, Copenhagen, Denmark, [5]BioEcoLinks NGO, Odesa, Ukraine, [6]Ukrainian Scientific Centre of Ecology of the Sea, Odesa, Ukraine, [7]I.I. Schmalhausen Institute of Zoology of National Academy of Sciences of Ukraine, Kyiv, Ukraine

The harbour porpoise (Phocoena phocoena) is a coastal toothed whale, occurring across the northern hemisphere from subarctic to temperate regions. Curiously, across this range, multiple distinct subspecies and populations have been described, indicating barriers to gene flow and adaptations to local environmental conditions. Here, we sequenced 41 genomes from harbour porpoises in the North Atlantic, Belt Sea and Black Sea to assess the demographic history, adaptations and guide management units of Atlantic harbour porpoises. We find the divergence between the North Atlantic and Black Sea lineages to have occurred 200-100 kya, broadly corresponding to the onset of the Penultimate Glacial Period, suggesting global cooling as a potential driver of their divergence. In contrast, the Belt Sea porpoise appears to be of more recent post-LGM origin. Selection analysis of Belt Sea porpoises relative to the North Atlantic identified multiple genomic regions associated with cold tolerance and vision, which we hypothesize plays a role in the Belt Sea porpoises' adaptation to a cooler and higher turbidity environment. Strikingly, we found that the Black Sea porpoises were characterized by substantially lower diversity than other porpoises and cetaceans in general, by excessive short-length runs of homozygosity (ROHs), and elevated genetic load. The accumulation of these loss-of-function mutations in the Black Sea indicates that it has not yet purged deleterious mutations, which could pose a threat to this vulnerable subspecies' overall fitness in the long run if its abundance drops to unsustainable levels.

[B110] Genomic analysis of field-collected feathers: An optimized ddRAD protocol for noninvasive monitoring of dispersal in the endangered capercaillie

Román Sapino[1], Irene Figueroa[2], Jose Castresana[1]
[1]Institute Of Evolutionary Biology (CSIC-UPF), Barcelona, Spain, [2]Paisatges Vius, L'Esquirol, Spain
Noninvasive sampling enables the genomic monitoring of wildlife, particularly endangered species, for which minimizing disturbance is essential. Although field-collected feathers have been widely used for traditional genetic monitoring, their application for high-throughput sequencing remains limited due to the degradation and low yield of recovered DNA. Consequently, the potential of feathers for generating high resolution genomic data remains largely uncharacterized. In this context, reduced representation sequencing methods, such as ddRAD, can facilitate the generation of SNPs distributed throughout the genome. In this study, we optimized a laboratory protocol that included the use of different types and parts of feathers for DNA extraction, molecular species identification, endogenous DNA quantification by qPCR, and ddRAD genomic library construction. We then used this approach with Western Capercaillie (Tetrao urogallus) feathers collected in the field from a population in the eastern Pyrenees to characterize genetic connectivity. The protocol generated a SNP panel with sufficient resolution for performing individualization and relatedness analysis. Individual identification of the feathers allowed us to track the movements of capercaillies. In addition, kinship analysis revealed sibling relationships between spatially separated subpopulations, demonstrating functional connectivity between them. Overall, this protocol provides a framework for assessing contemporary dispersal in avian species using noninvasively collected feathers and offers a valuable tool for developing informed management strategies for habitats and species in fragmented landscapes.

[B111] Applying genomics to the management of Wallacean flagship taxa: evaluating the evolutionary potential of insurance populations

Dr Anik Budhi Dharmayanthi[2], Dr Sabhrina Gita Aninta[3], Dr Alberto Carmagnini[1], Yonathan[4], Yohana Tri Hastuti[5], Glen Kartiko[6], Professor Laurent Frantz[1], Prof Gono Semiadi[2], Rosie Drinkwater[1]
[1]Ludwig-maximilians-universität München, Munich, Germany, [2]Research Centre for Biosystematics and Evolution, National Research and Innovation Agency, Cibinong, Indonesia, [3]University of Copenhagen, Copenhagen, Denmark, [4]Global Species Management Plan Consortium, Indonesian Zoos and Aquariums Association, Jakarta, Indonesia, [5]Taman Safari Indonesia, Anoa studbook, Bogor, Indonesia, [6]Surabaya Zoo, Babirusa studbook, Surabaya, Indonesia

Anoa (Bubalus sp.) and babirusa (Babyrousa sp.) are iconic flagship taxa endemic to biodiversity hotspot of Wallacea, Indonesia. Facing severe threats from anthropogenic activities such as habitat fragmentation and poaching, these species are now managed under a Global Species Management plan (GSMP). The GSMP bridges in-situ protection with ex-situ insurance populations and to safeguard their future, the GSMP prioritises genomically informed practices to ensure population viability.

We sequenced 53 anoa and babirusa from Indonesian zoos and analysed their ancestry, genomic diversity and relatedness. We also evaluated metrics of genomic erosion, including heterozygosity, inbreeding, and mutational load, and compared this to wild populations. Ancestry analysis revealed relatively pure lineages in ex-situ anoa, whereas babirusa populations were highly admixed across zoos. While we identified strong kinship within each zoo, overall levels of inbreeding and mutational load in the ex-situ populations remain comparable to wild counterparts, highlighting their potential to maintain the long-term survival of wild populations.

Using these metrics, we can provide genomic-based breeding recommendations for the GSMP. This work includes active consultation with the studbook keepers and local stakeholders to familiarise conservation practitioners with a localized, evidence-based framework for breeding recommendations. This collaborative approach ensures long-term evolutionary resilience for these vulnerable species in a rapidly changing world.

[B112] Demographic changes in a declining narwhal population in East Greenland using a 15-year time series

Sam Fenton[1], Dr Marie Louis[1,2], Professor Mads Peter Heide-Jørgensen[2], Professor Eline Lorenzen[1]
[1]University Of Copenhagen, , Denmark, [2]Greenland Institute of Natural Resources, , Greenland

The continued disappearance of Arctic sea ice represents a potential tipping point that will have cascading effects on top predators in the region both by changing prey availability and by reducing habitats. The narwhal (Monodon monoceros) is considered to be the most sensitive Arctic marine mammal to climate change, due to its narrow niche and specialised feeding habits. Narwhals are already showing the effect of habitat loss at the species level with evidence of an alarming decline (approximately 2636 individuals in 2008 to 365 in 2022) in Southeast Greenland, over the last two decades. Given the wider impact of the extirpation of a top predator from the ecosystem and the importance of the species to Inuit communities and their cultural heritage, understanding the demographic consequences of this rapid population decline is crucial. However, using samples from just a single time point may not give an accurate reflection of how population dynamics have changed over time. We therefore used a time series of narwhal samples (N=348) collected over 15 years in the Scoresby Sound region of East Greenland. I will present the latest results from this study where we generated whole-genome dataset to: 1) estimate changes in abundance of narwhals in Scoresby since 2005; 2) identify individuals and their relatedness through time; 3) infer changes in population composition and social structure using estimates of sex and relatedness; 4) assess the level of differentiation and changes in connectivity with neighbouring sub-populations along the East Greenland coastline.

[B113] Tracking the Recovery of Bowhead Whales in Qeqertarsuup tunua, West Greenland, through a 25-Year Genomic Time Series

Santiago Jose Avila Quintero[1], Professor Eline Lorenzen[1], Dr Mads Peter Heide-Jørgensen[2], Shyam Gopalakrishnan[2]
[1]Globe Institute, Section for Molecular Ecology and Evolution, , Denmark, [2]Greenland Institute of Natural Resources, Department of Birds and Mammals, , Greenland, [3]Globe Institute, Section for Hologenomics, , Denmark

The Arctic is warming four times faster than the global average, causing rapid sea ice loss and warmer water intrusion. These rapid changes threaten the recovery of bowhead whales (Balaena mysticetus), a top Arctic predator severely depleted during commercial whaling. While bowheads became the first species under formal protection nearly a century ago, recovery trends vary across the four recognised stocks. For the Eastern Canada–West Greenland (ECWG) stock, abundance in Qeqertarsuup tunua (Disko Bay), a historic stronghold, has notably increased since the early 21st century, following near disappearance due to commercial whaling. High productivity here traditionally supports high site fidelity during the spring and early summer, especially for females. However, less nutritious prey arriving with warmer southern waters now creates an energy deficit that may alter both whale abundance and staying time in the bay.

Here, we use a 25-year biopsy time series of over 1,100 samples from Disko bay to study population-level responses to changes in the environmental conditions. We use genome-wide data to identify individuals and establish kinship across the study period. These genomic data provide preliminary results for annual abundance, visiting frequency, and staying time. We aim to establish a baseline for population-level responses, using the bowhead whale as a sentinel to evaluate how shifting Arctic conditions impact long-term species stability.

[B114] Genomic consequences of admixture in a Swedish sand lizard population

Seraina Emilia Bracamonte[1], Mats Olsson[2], Erik Wapstra[3], Willow R. Lindsay[2], Mette Lillie[1]
[1]Uppsala University, , Sweden, [2]Gothenburg University, , Sweden, [3]University of Tasmania, , Australia
Small and fragmented populations are vulnerable to genetic diversity loss, inbreeding depression and the effects of deleterious mutations. Translocation of individuals for genetic rescue can mitigate these effects by increasing diversity and population size, which has the potential to increase population persistence. Here, we studied the genomic consequences of genetic rescue using a sand lizard population that was established via experimental admixture of individuals from a highly inbred Swedish sand lizard population with individuals from other low-diversity populations in southern Sweden. Offspring of these crosses were released on a small island approx. 20 years prior to sampling (5-6 generations), and the low hatching success and hatchling viability observed in the main founder population have since disappeared. Using low-coverage whole-genome resequencing, we found increased genetic diversity and reduced realized genetic load in the admixed populations compared to its main founder population. We showed that contributions of southern Swedish populations persist in the admixed population without replacing the ancestry of the main founder. Our study highlights the benefits of admixture for genetic rescue which may provide valuable adaptive variation for population recovery and persistence.

[B115] Museum samples track declining genetic diversity of the Eastern honey bee (Apis cerana)

Shanlin Liu[1], Xin Zhou[2], Tom Gilbert[3]
[1]Institute Of Zoology, , China, [2]China Agricultural University, , China, [3]University of Copenhagen, , Denmark

Apis cerana has the broadest native range of all Asiatic honey bees and is a crucial pollinator; nevertheless, it has seen a dramatic population shrinkage over the last hundred years. Despite the profound impact of modern agriculture on bee health, the degree to which anthropogenic activities have steered the evolutionary trajectory of A. cerana remains poorly understood. Utilizing museum specimens, we reconstructed a genomic time-series spanning over a century—including samples from approximately 120, 70, and 40 years ago—to capture the populations before and during the intensification of modern agriculture. Our results demonstrate a widespread loss of genetic diversity resulting from population decline and increased fragmentation. Notably, we identified strong selective signatures in genes related to the nervous system, specifically nicotinic acetylcholine receptors. These findings offer direct genomic evidence of how long-term insecticide exposure has shaped the adaptive landscape of a key pollinator.

[B116] Ecotypic diversity as a pathway to persistence under environmental change in a Californian butterfly

Veronika Mrazek[1], Gabriela Montejo-Kovacevich[1], Jana Olivia Weigel[1]
[1]Department of Ecology and Genetics, EBC, Uppsala University, Uppsala, Sweden

Rapid anthropogenic environmental change forces populations to adapt quickly or follow suitable conditions through range-shifts, often exposing them to novel ecological challenges. For phytophagous insects, these may require rapid adaptation to new hostplants, imposing strong selection on traits underlying survival, reproduction, and behavior.

The Edith's checkerspot butterfly (Euphydryas editha) was the first species to be recorded shifting northwards due to climate change (Parmesan, 1996) and exhibits striking host-associated heritable ecotypes that differ in phenology, oviposition behavior, climate sensitivity and conservation status. While a large body of works has described the ecological adaptations of these ecotypes (Singer et al.), less is known about the underlying molecular basis.

We use more than 300 re-sequenced whole genomes from 15 E. editha populations spanning Pedicularis, Collinsia and other hostplants to investigate the evolutionary genomics of hostplant adaptation. We ask: (i) how genomic differentiation is structured among ecotypes relative to geography and gene flow; (ii) whether adaptation to alternative hostplants is associated with specific genomic regions across populations; and (iii) how specific host adaptive alleles may be able to spread and hinder or aid conservation of vulnerable populations.

We aim to provide an understanding of the genomic basis of ecotypic diversity in the Edith's checkerspot butterfly, and how such diversity may provide evolutionary pathways for persistence under climate-driven habitat change. Understanding ecotypic adaptation at the genomic level can inform conservation strategies - such as assisted gene flow or ecotype-informed translocations - to enhance the resilience of vulnerable populations in a rapidly changing world.

[B117] Molecular-aided management of genetic resources: a case study in traditional horse populations from France

Yumi Shakya[1], Clémentine Bonnin[2], Sophie Danvy[3], Bernard Dumont - Saint Priest[3], Eric Rousseaux[2], Margot Sabbagh[3], Etienne Verrier[1], Bertrand Servin[4], Gwendal Restoux[1]
[1]GABI, INRAE, Jouy-en-Josas, France, [2]GenPhyse, INRAE, Toulouse, France, [3]SFET, , France, [4]IFCE, , France

Many populations of domestic animal species are endangered by small census sizes, leading to strong genetic drift, causing diversity loss and inbreeding depression. In-situ conservation programs are usually based on partial, error-prone pedigree information. To address these limitations, we investigated the utility of molecular markers to better detect the extent of genetic diversity loss induced by management practices.

Our study is based on a sample of 976 animals from 18 traditional French horse breeds (≈50 per breed), across two time periods separated by a major change in their genetic management practices. The samples were genotyped using a 10K SNP array dedicated to the characterization of genetic resources. The genetic structure of the breeds clearly distinguishes light “territorial” breeds from heavier draft breeds, as well as potential gene flow between the two groups. These genetic relationships could be linked to historical records, demonstrating the usefulness of genomic data in tracing back the creation and evolution of breeds.

In addition, the evolution over the two time periods of molecular-based kinship and inbreeding coefficients exhibited distinct patterns, reflecting a slightly different effect of management practices on each estimate. The results also differed between breeds. This led us to propose a new metric that combines both kinship and inbreeding estimates, tested by simulations, to better inform conservation programs on population management.

Our study demonstrates the usefulness of genomic markers for monitoring genetic diversity and evaluating the effectiveness of management programs, while underscoring the need for cautious interpretation of molecular metrics in conservation programs.

[B118] Beyond Misfolding: Mapping How Cellular Context Converts Mutations into Fitness Costs

Ashley Brown[1,2], Natalie Quan[1,2], Dr. Kerry Geiler-Samerotte[1,2]
[1]Center for Mechanisms of Evolution, Tempe, United States, [2]School of Life Sciences, Tempe, United States

Mutations can reduce fitness even when a protein's native function is irrelevant. Such costs are referred to as "collateral" and arise purely from how a mutant protein behaves in the cell, through toxic aggregation, aberrant interactions, or the disruption of energetic balance. In a deep mutational scan of yellow fluorescent protein (YFP) expressed in Saccharomyces cerevisiae, many variants show fitness defects that are not explained by the toxicity of misfolded proteins alone. If not misfolding, then what cellular limitation explains these fitness defects?

We address this using a deep mutational scan of mutant YFP variants, where fitness differences reflect collateral effects by design. We measure variant fitness in pooled liquid-culture competitions across stresses that probe distinct proteostasis routes and cellular demands. These include varying expression load and temperature to alter protein burden, and chemically targeting specific pathways, including the proteasome, vacuolar proteases, Hsp90, and membrane composition, with matched controls.

By comparing responses across stresses, we aim to identify classes of mutants whose collateral costs are uniquely buffered or exacerbated in specific conditions. This work will map how cellular context determines which mutations impose fitness costs, implicating mechanisms that may convert molecular damage into evolutionary burden.

[B119] Understanding RNase P regulation: How E. coli evolves to compensate for defects in an essential and universally conserved enzyme

Emily Tonge[1], Professor Leif Kirsebom[2], Dr Arianne Babina[1]
[1]University Of Glasgow, , United Kingdom, [2]Uppsala University , , Sweden

The regulation and maintenance of essential RNA–protein complexes place significant constraints on evolution across all domains of life, as these complexes frequently coordinate core cellular processes and regulate adaptive responses to environmental changes. RNase P is an ancient and universally conserved ribonucleoprotein enzyme required for tRNA maturation, yet the molecular pathways governing its expression, assembly, and stability remain poorly characterised in vivo. Since its discovery in the 1970s, several conditionally lethal bacterial mutants have been used as tractable model systems to understand RNase P function. One widely studied example is the temperature-sensitive rnpA49 allele in Escherichia coli, which destabilises the RNase P holoenzyme by disrupting interactions between its RNA and protein subunits. Using this mutant strain, we recently identified several classes of spontaneous suppressor mutations that partially restore growth at non-permissive temperature, including copy number increases of RNase P subunit genes and loss-of-function mutations in Lon protease and RNase R, suggesting novel links between these regulatory pathways and RNase P. In the present study, we serially passaged representative suppressor lineages under sublethal temperature stress to investigate how these initial compensatory mutations shape long-term evolutionary recovery of the E. coli rnpA49 strain and to define, at a systems level, the pathways governing RNase P regulation, degradation, and turnover. Together, this work elucidates the in vivo regulation of an essential ribonucleoprotein enzyme in a key model organism and more broadly reveals principles underlying the maintenance, robustness, and evolutionary plasticity of universally conserved RNA–protein complexes.

[B120] Core elements play distinct roles in promoter birth and transcriptional regulation

Hsin-Hung Chou[1]
[1]National Taiwan University, Taipei, Taiwan

Gene expression shapes phenotypes and evolution. However, studies of gene regulation focus on transcription factors, overlooking core promoters. To investigate how promoters emerge and regulate transcription, we determined the sequence–function landscapes of core elements, -35 and -10, in constitutive and transcription factor-regulated promoters in Escherichia coli. Characterization of in vivo transcriptional landscapes and in vitro RNA polymerase–promoter interactions showed the -10 element as essential for promoter evolution from random sequences. In contrast, the -35 element, though broadly conserved, is dispensable for promoter birth. Instead, it exerts greater impact on gene regulation via coordinated interactions with transcription activators and RNA polymerase. We further showed that evolution fine-tunes the -35 and -10 sequences of transcription factor-regulated promoters to achieve near-maximal fold changes by lowering basal while elevating induced expression. A notable exception is PluxI, whose leaky expression provides a crucial baseline for initiating quorum sensing. These findings elucidate promoter design principles and underscore the interdependence and coevolution of core elements, RNA polymerase, and transcription factors.

[B121] Clonal interference and the fate of synonymous mutations in P. fluorescens

Jacob Van Oorschot[1], Dr. Aaron Hinz[1], Dr. Patrick Lypaczewski[1], Dr. Rees Kassen[1]
[1]Mcgill University, Montreal, Canada

Synonymous mutations are often assumed to have no noticeable effect on phenotype. However, mounting evidence suggests synonymous mutations that can affect fitness may arise often. Putative mechanisms to explain these effects include codon bias and gene expression altered at the level of transcription or translation. Nonetheless, beneficial synonymous mutations are recovered less frequently in evolution experiments than expected from site-directed mutagenesis experiments for reasons that remain unclear. Here, we test the hypothesis that the depletion of adaptive synonymous mutations in microbial experimental evolution is the result of high mutation supply rates generating clonal interference, resulting in the enrichment of the more abundant class of large effect nonsynonymous mutations. To test this idea we evolved Pseudomonas fluorescens for 600 generations in low glucose, a condition where beneficial synonymous mutations that create internal promoter sites in a glucose uptake operon had previously been observed, across varying mutation supply rates. Sequencing evolved populations reveals the prevalence of nonsynonymous and synonymous mutations to evaluate the prediction that synonymous mutations will be more prevalent when mutation supply rates, and so clonal interference, are low. Our results emphasize that synonymous mutations are not always neutral and shed light on the conditions under which synonymous mutations contribute to adaptation.

[B122] Genome editing reveals: distinct and independent effects on life-history traits of three SNPs in one intron in Drosophila melanogaster

Julia Beets[1], Janine Mariën[1], Prof. Jacintha Ellers[1], dr. Mirte Bosse[1,2], Katja Hoedjes[1]
[1]Vrije Universiteit Amsterdam, Amsterdam, Netherlands, [2]Wageningen University & Research, Wageningen, Netherlands

Genome-wide approaches such as GWAS and experimental evolution have associated many candidate SNPs with complex traits, yet the functional impact of individual variants remains difficult to resolve due to linkage, non-independence, and the lack of causal validation. Addressing these challenges requires an integrative approach including direct functional tests. Here, we investigate three intronic life-history–associated SNPs in Eip75B identified by experimental evolution in Drosophila melanogaster, combining long-read Nanopore sequencing to characterize local haplotype structure with precise CRISPR–Cas9 editing to test the effects of individual SNPs.
Long-read sequencing of the experimentally evolved populations revealed a rapid decay of haplotype homozygosity surrounding the focal SNPs, with allelic asymmetry. These patterns of allele-specific selection per SNP, dependent on selection regimes of experimental evolution, are consistent with a contribution of the variants to fitness-related traits. Using CRISPR–Cas9 genome editing, we independently introduced each of the three intronic SNPs by allelic replacement to directly test their functional and causal effects on life-history traits. Phenotyping of life-history traits revealed distinct and independent effects of each SNP, on fecundity, longevity, starvation resistance and egg volume. By integrating identification of fitness-associated SNPs from experimental evolution, haplotype-based selection analysis, and targeted single-nucleotide genome editing, our study provides a fine-scale assessment of the genetic basis of life-history evolution. Together, our results demonstrate that individual nucleotide variants within an intron can independently shape complex life-history trade-offs, underscoring how individual SNPs can shape complex life-history trade-offs and the importance of testing their functional effects for understanding this genetic architecture.

[B124] Using saturation mutagenesis to map the evolutionary forces that shaped human regulatory elements

Omer Ronen[1], Simon Fishilevich[1], Fumitaka Inoue[2], Prof. Tomas Marques Bonet[3], Yoav Navon[1], Georgii Bazykin[4], Dr. David Gokhman[1]

[1]Department of Molecular Genetics, Weizmann Institute of Science, Rehovot, Israel, [2]Institute for the Advanced Study of Human Biology (WPI-ASHBi), Kyoto University, Kyoto, Japan, [3]Institute of Evolutionary Biology (UPF-CSIC), PRBB, Barcelona, Spain, [4]Department of Biology, Emory University, Atlanta, USA

Adaptation via natural selection is the process by which the incredible fit between species and their environment has evolved. However, despite the good understanding of evolutionary adaptation at the phenotypic level, we have a limited understanding of the genetic changes that made us human. Particularly, we are still far from understanding how adaptation presents itself at the molecular level. This task is even more difficult in the noncoding parts of the genome, where most adaptations occur. Saturation mutagenesis is an approach that allows researchers to compare true evolutionary changes to the full spectrum of possible mutations. We apply it using massively parallel reporter assays (MPRAs), allowing us to generate the largest saturation mutagenesis library of regulatory regions in the human genome, consisting of 270 diverse regulatory elements. To identify signatures of selection, we develop evolutionary tests for selection on regulatory elements, and apply them to individual noncoding regulatory elements, as well as classes of elements, such as human accelerated regions, ultra-conserved regions, and more. We identified signals of positive and negative selection in various elements and discovered specific variants that have likely driven human-specific phenotypes, e,g., in IRF4, a key transcription factor that affects pigmentation and skin diseases. This work sheds light on the selective forces that shaped human noncoding regulatory elements and identifies key substitutions in human adaptations.

[B125] Ribosomal frameshifting drives natural selection against codon repeats

Ruoxi Wang[1], Jianzhi Zhang[1]
[1]Department of Ecology and Evolutionary Biology, University Of Michigan, Ann Arbor, United States
Ribosomal frameshifting refers to the phenomenon in which the ribosome slips into the +1 or −1 reading frame during translation, producing frameshifted or truncated proteins. Although ribosomal frameshifting is commonly considered harmful, it is widespread, programmed, and indispensable in some viruses. A recent study demonstrated that short codon repeats can induce ribosomal frameshifting, inferred codon-repeat-associated frameshifting (CRFS) from the human proteome, and identified a CRFS event that produces an altered human protein functionally antagonistic to the canonical protein. These observations prompted the suggestion that human CRFS represents an important, regulated mechanism for increasing gene product diversity. Here we propose and test a competing hypothesis that CRFS largely reflects a molecular error and is generally deleterious. This error hypothesis makes distinct predictions about the frequency, distribution, and conservation of codon repeats that induce frameshifting, which are strongly supported by our genomic analyses. Codon repeats with a high probability of inducing frameshifting are rarer in protein-coding regions than in comparable non-coding regions, and are especially avoided in the coding regions of highly expressed, highly conserved, and essential genes. Furthermore, codon repeats are depleted from the middle of coding sequences, consistent with selection against the production of large truncated proteins. Together, these results support the error hypothesis of CRFS and suggest that cellular processes are less orderly and precise than is commonly thought.

[B126] Ancestral reconstruction of the emergence of catalytic activity through multimerisation of a SH3 binding domain

Stella Cellier-Goetghebeur[1,2,3], Samy Faraj[1,2,3], Roxana Tarabuta[1,2,3], Margaux Raboisson[1,2,3], Claudèle Lemay-St-Denis[1,2,3], Joelle Pelletier[1,2,3,4]
[1]Department of Biochemistry and Molecular Medicine, Université de Montréal, , Canada, [2]PROTEO, The Québec Network for Research on Protein, Function, Engineering and Applications, , Canada, [3]CGCC, Center in Green Chemistry and Catalysis, , Canada, [4]Department of Chemistry, Université de Montréal, , Canada

The Src Homology 3 domain is a binding domain found in all domains of life but is rarely catalytic. One exception is found in the bacterial dihydrofolate reductase type B (DfrB), a homotetrameric enzyme in which a SH3 domain and a short N-terminus constitute each monomer. DfrB enzymes catalyze the reduction of dihydrofolate into tetrahydrofolate by a proximity-based mechanism of catalysis, and they confer resistance to the synthetic antibiotic trimethoprim (TMP) that selectively inhibits the ubiquitous bacterial dihydrofolate reductase FolA. Although DfrB enzymes were discovered in the context of TMP resistance, our previous work has shown that their presence is not limited to the resistome, and that DfrB enzymes have the potential to provide unique insight into the emergence of catalytic function, namely how a mechanism of proximity-based catalysis can emerge from a binding domain.
We hypothesize that the evolution of the active homotetrameric DfrB enzyme was threefold: a monomer-monomer interface followed by a dimer-dimer interface would have evolved in ancestral SH3 domains in parallel with the capacity to bind dihydrofolate and NADPH to gain a dihydrofolate reductase catalytic activity. To explore this, we have reconstructed ancestral sequences from a phylogeny of DfrB and DfrB-like SH3 domains. Our results are coherent with the stepwise evolution of a dimer followed by an active homotetramer, with the interactions at the two interfaces gradually becoming stronger with evolutionary time. Our work provides insight into the evolution of a catalytic homotetramer from a monomeric binding domain.

[B127] Co-evolution of Olfactory and Taste Receptors in Birds

Xulan Shen[1], Guojie Zhang[1], Shaohong Feng[1]
[1]Zhejiang University, Hangzhou, China

Olfaction and taste are key chemical senses shaping environmental perception and dietary niche adaptation, yet their evolutionary relationship remains debated: do they reflect compensatory trade-offs or ecology-driven specialization? We address this using a systems-level phylogenomic framework across 363 bird species spanning diverse lineages and ecological niches. We identify a genome-wide co-evolution signature: across species diversification, olfactory receptor (OR) copy-number change rates are significantly negatively associated with taste receptor (TR) substitution rates, consistent with compensatory trade-offs between the two systems. TR repertoires are comparatively stable, with TAS1R2 consistently appears absent across birds and recurrent loss-of-function signatures of TAS1R1 enriched in aquatic predatory lineages. In parallel, OR repertoires exhibit pronounced lineage-specific dynamics: core waterbirds show substantial pseudogene expansion, whereas nocturnal birds display functional OR expansion, alongside hotspots of repertoire turnover. We further link OR/TR dynamics to ecological drivers while accounting for shared ancestry. Finally, focusing on key ecological transition branches, trait-transition mapping suggests that accelerated OR copy-number change can coincide with adaptive signals in TR genes, consistent with transition-associated synergy during niche shifts. Together, these results reveal stage-dependent co-evolution of avian OR and TR systems—broad antagonistic trade-offs coupled with cooperative remodeling at key ecological transitions—challenging a simple “trade-off versus specialization” dichotomy and highlighting birds as a powerful model for multisensory evolution.

[B128] A Clostridium tetani genome recovered from 3000-year-old human remains in Papua New Guinea

Konstantina Cheshmedzhieva[1,2], Glenn Summerhayes[3], Martin Kuhlwilm[1,2], Katerina Douka Douka[1,2], Sojung Han[1,2]
[1]University Of Vienna, Vienna, Austria, [2]HEAS (Human Evolution and Archaeological Sciences), Vienna, Austria, [3]University of Otago, Dunedin, New Zealand

Clostridium tetani is the causative agent of the neuroparalytic disease tetanus, which infects humans and other mammals. These bacteria are abundant in the environment such as soil and faeces, and upon entering open wounds and meeting anaerobic conditions, its spores germinates and produces a highly potent neurotoxin (TeNT), leading to paralyzing the hosts. A previous study which compared ancient and modern lineages showed that the strains cluster following geography, rather than time. Human remains were identified in Yuku cave in the Papua New Guinean highlands at 1,280m above sea level, and dated shortly after the Austronesian expansion more than 3,000 years ago. After ancient DNA extraction and sequencing, here we have recovered a Clostridium tetani genome from a human bone fragment from this cave at 9.2-fold average depth and 89% breadth of coverage on the chromosome, and 4.4-fold depth and 52% breadth of coverage on the plasmid. Notably, we found evidence for the presence of Clostridium tetani DNA in multiple bone fragments from the same layer of the cave, which belong to the same male human individual. We see clear postmortem damage patterns on the mapped reads of both human and bacterial origin, demonstrating the authentic ancient origin of the DNA. The genome was furthermore confirmed to belong to a single strain, with a high degree of identity between sequencing reads. We performed phylogenetic analyses in which we explore the clustering of this ancient strain with other known lineages.

[B129] Pangenome graph-guided assembly of ancient pathogen genomes

Daria Evseeva[1,2], Caitlin A. Mitchell[1], Jun. Prof. Dr. Maria Spyrou[1,3]
[1]Microbial Archaeogenomics, Institute for Archaeological Sciences, Department of Geosciences, Eberhard Karl University Of Tübingen, Tübingen, Germany, [2]Max Planck Institute for Biology, Tübingen, Germany, [3]Senckenberg Centre for Human Evolution and Palaeoenvironment at the University of Tübingen, Tübingen, Germany

Sequencing of ancient bacterial pathogens provides key insights into their long-term evolutionary history and reveals the genomic changes underlying historical epidemic outbreaks. Many determinants of bacterial pathogenicity are known to reside in the accessory genome and are shaped by mobile genetic elements, recombination, and genome rearrangements. However, detection of these processes in ancient genomes is strongly constrained by reference bias arising from the use of a single modern reference for genome reconstruction. Pangenome graphs constructed from collections of all available complete modern genomes may offer a powerful alternative to linear references for read mapping, and genome assembly. By representing the full diversity of the modern mobilome, pangenome graphs enable more comprehensive investigation of mobile genetic elements in ancient pathogens. Here, we benchmark the capabilities and limitations of pangenome graph-guided assembly for reconstructing bacterial mobilomes using simulated ancient DNA reads generated from publicly available complete Yersinia pestis genomes. We compare the performance of two alternative pangenome graph-guided assembly approaches: direct mapping of ancient DNA reads to the pangenome graph followed by variant calling versus de novo assembly of ancient DNA reads followed by contig mapping to the pangenome graph. Finally, we apply the pangenome-guided assembly framework to authentic ancient Y. pestis datasets recovered from human skeletal remains from prehistoric archaeological contexts, demonstrating its utility for genome reconstruction in ancient pathogen datasets.

[B130] Characterization of the ancient human oral microbiota recovered from teeth

Vincent Thygesen[1], Dr Florentin Constancias[2], Dr Antonio Fernandez-Guerra[1], Professor Michael Givskov[1], Professor Daniel Belstrøm[1], Martin Sikora[1]
[1]University Of Copenhagen, Copenhagen, Denmark, [2]ETH Zürich, Zürich, Switzerland

Metagenomic sequencing of ancient DNA extracted from dental calculus has yielded important insights
into the composition and functional diversity of the human oral microbiota in the past. Ancient human
teeth have been sampled widely for studies of human host DNA, however their characteristics as potential sources for ancient human oral microbiota have yet to be determined.

Here, we present a comprehensive characterization of the microbiota retrieved from 1,313 ancient humans ranging from the Upper Paleolithic to historic times. Analyzing species abundance profiles derived against a modified version of Genome Taxonomy DataBase (GTDB), we demonstrate that oral microbiota are frequently recovered from ancient human teeth samples. We further characterize the composition of the teeth microbiota of 217 high abundance samples against large-scale reference dataset including publicly available modern and ancient oral microbiomes.

Our study demonstrates that ancient metagenomic data extracted from ancient human teeth can provide
a hitherto under-exploited resource to study human oral microbiome evolution.

[B131] Investigating the evolutionary dynamics underlying DNA virus immune response

Rutvi Rajpara[1], Dr Erik Abner[1], Dr Monika Karmin[1], Dr Irene Gallego Romero[1,2], Michael Dannemann[1]
[1]University of Tartu, Tartu, Estonia, [2]The University of Melbourne, , Australia

DNA viruses such as herpes viruses, and polyomaviruses, can persist in humans and establish infections for the lifetime. Although these infections are usually asymptomatic and the amount of viral DNA present in blood can vary widely between individuals. Higher viral loads have been linked to immune dysfunction and disease risk. Recent large biobank studies using whole-genome sequencing (WGS) have shown that viral load is shaped by demographic factors and host genetics, especially variation in the HLA region. (Kamitaki N et al. 2025). Our group's recent analyses have shown that archaic has played a role in shaping in virus load associated loci, with multiple Neandertal and Denisovan derived haplotypes on chromosome 17 and within chromosome 6 MHC region being associated with DNA virus levels in participants of the UK Biobank. The objective of this project is to apply this viral-load and host-genetics framework to the Estonian Biobank using long-read sequencing data. By integrating viral load quantification with GWAS and archaic haplotype mapping, we aim to test whether population-specific evolutionary histories similarly shape host–virus interactions in Northern Europe.

[B132] Advancing hominin taxonomic identification through an extended enamel palaeoproteomics database

Viridiana Villa Islas[1], Ragnheiður Diljá Ásmundsdóttir, Zandra Fagernäs, Dr Laurits Skov, Frido Welker
[1]University Of Copenhagen, , Denmark

Human evolution dates back millions of years, especially in tropical regions. Thus, obtaining DNA for the accurate identification of fragmented hominin skeletal material remains challenging. However, enamel proteins have been shown to survive for more than 2-3 million years in tropical regions, allowing us to explore the distant past of human evolution using molecular evidence. Commonly, the analysis of the proteins recovered from enamel samples is restricted to the ca.13 enamel core proteins. Still, additional proteins have been reported in dental enamel protein extracts, potentially containing single amino acid variants (SAAVs) that are informative for the taxonomic assignment of unidentified hominin teeth. To explore the potential of these non-core enamel proteins, we recovered (in silico) the protein sequence variation of 86 proteins reported in the dental enamel literature for present-day humans (1000 Genomes Project), three Neanderthals, and one Denisovan. By translating these proteins and subsequently analysing them, we catalogued proteins containing unique SAAVs in Denisovans, Neanderthals, and modern humans, creating an extended database for palaeoproteomic analysis. The common practice in the palaeoproteomics field has relied on the human protein reference as a surrogate for modern humans; here, we show that adding global present-day human variation to the database led to the recognition of specific archaic SAAVs and several SAAVs shared between modern humans and archaic hominins. Taxonomic and phylogenetic analysis of these hominin groups can be refined using the database created when enamel is used as a starting material for the recovery of ancient proteins.

[B133] The Genomic Adaptation of Peromyscus Maniculatus (North American Deer Mouse) in High-Altitude Environments

Larissa Rockenbach[1], Allie Graham[1,2]
[1]University Of Kansas, Lawrence, United States, [2]University of Kansas Department of Molecular Biosciences, Lawrence, United States

High-altitude environments impose many physiological challenges, including reduced oxygen availability, aridity, and increased UV exposure. Understanding how mammals adapt to these conditions is central to evolutionary biology and offers greater insight into the genetic basis of survival in extreme habitats. The North American deer mouse (Peromyscus maniculatus) is a widespread and ecologically adaptive species and offers a powerful model for investigating genomic adaptation across altitudinal gradients. Earlier studies of high-altitude adaptation in mammals have emphasized changes in hemoglobin function and ventilatory control. However, the broader genomic architecture underlying these adaptations is still incompletely characterized. The general question addressed in this study is whether P. maniculatus populations living at high elevation show consistent signatures of selection in genes associated with hypoxia tolerance. In this project, I examined whole-genome sequencing data from 50 individuals from two populations - low (122 m) and high altitude (1585 m). Using the genetic variation from this data, I have performed outlier analyses to find whether there are specific variants in genes that are statistically associated with high-altitude; suggesting they are important in adaptation to this harsh environment. Identifying variation will provide a foundation for understanding how selective pressures act on multiple physiological systems in response to high-altitude stressors. More broadly, this study will highlight the deer mouse as a model for looking at the genetic basis of environmental adaptation. By focusing on candidate genes linked to hypoxia tolerance, the project contributes to a wider perspective on how selective pressures shape genomic diversity and resilience across taxa.

[B134] Diverse Patterns of Transposable Element Expression in Macaca mulatta and Tissue-Specific Regulation of Adjacent Genes

MS Nakyoung Kim[1], MS. KunHyang Park[1], Dr. Sugi Lee[1], Dae-Soo Kim[1]
[1]Korea Research Institute of Bioscience and Biotechnology (KRIBB), Gwahangno, Yuseong-gu, South Korea

Transposable elements account for approximately half of primate genomes and contribute to phenotypic diversity by influencing gene regulation through diverse mechanisms. Increasing evidence suggests that TEs play important roles in both physiological and pathological processes. Because TEs contain regulatory motifs, they can function as cis-regulatory elements that modulate gene expression. In this study, we systematically analyzed TE expression across 13 different tissue types in the rhesus macaque, the most widely used non-human primate model for human biology, using publicly available RNA-seq datasets. Our aim was to characterize tissue-specific TE expression patterns and explore their potential contributions to gene regulation.

We found that TEs are not randomly distributed across the rhesus macaque genome but show distinct enrichment patterns depending on their genomic locations. While most TEs located in intergenic regions were transcriptionally silenced, highly expressed TEs were markedly enriched in the thymus. Clustering analysis identified four major TE expression patterns associated with tissue type and the evolutionary age of the TEs. Notably, 365 short interspersed nuclear elements and 269 long interspersed nuclear elements were significantly enriched at transcription start sites , suggesting potential roles in transcriptional regulation. Furthermore, we identified tissue-specific highly expressed TEs located within coding DNA sequence and upstream regions, whose neighboring genes frequently exhibited elevated expression levels. Overall, this study provides a comprehensive overview of TE expression across rhesus macaque tissues and highlights the role of TEs as tissue-specific regulators of nearby gene expression in primates.

[B136] Diaporthe species present an one-compartment genome architecture

Alicia Sofia Tempesta Brasil Silka[1], Doctor Chirlei Glienke[1], Doctor Desirre Alexia Lourenço Petters-Vandresen[1]
[1]Federal University Of Parana (ufpr), Curitiba, Brazil

Diaporthe (Sordariomycetes) is a cosmopolitan genus comprising over 1300 species of diverse lifestyles, including saprobes, endophytes, and plants and animals pathogens, with a high level of genetic diversity and complex phylogenetic resolution. Another remarkable feature of Diaporthe is their production of Secondary Metabolites (SMs) that are potentially bioactive and are encoded in Biosynthetic Gene Clusters (BGCs). Moreover, many phytopathogenic Diaporthe species pose relevant threats to economically important crops. To further explore the biotechnological potential and gain deeper understanding of the diverse Diaporthe lifestyles, this study evaluates publicly available Diaporthe genome assemblies under a comparative approach, characterizing the genomic architecture of Diaporthe genomes. We employ 79 genome assemblies, 70 of which are publicly available in NCBI and JGI Mycoscosm, and 9 newly generated assemblies from Brazilian Diaporthe strains from Pantanal and Cerrado biomes. Gene predictions were generated using Funannotate, and for functional annotation, BGCs, CAZymes, proteases, lipases and effectors were annotated. Overall, Diaporthe genomes are relatively larger than the average of Sordariomycetes genomes (~57MB for Diaporthe vs. ~42Mb for Sordariomycetes), with a higher number of protein-coding genes (~13399 for Diaporthe vs. 11968 for Sordariomycetes). Interestingly, although several Sordariomycetes species present compartmentalized genomes (such as observed for some Colletotrichum and Fusarium species), Diaporthe genomes present features of one-compartment genome architecture, suggesting some differences in evolutionary processes that shaped the genome organization of Diaporthe species when compared to other Diaporthales and Sordariomycetes genera.

[B137] Massive genomic flux in a young autogamous diploid revealed through new approach to legacy sequence data

Alistair Hockey[1], Dr. Robert Syme[1,2], Dr. Jana Cizkova[3], Dr. Eva Hribova[3], Dr. Janine Croser[4], Prof. Megan Ryan[1], Dr. Judith Lichtenzveig[1]

[1]The UWA School of Agriculture and Environment, University of Western Australia, Crawley, Australia, [2]Seqera Labs, Barcelona, Spain, [3]The Institute of Experimental Botany, Czech Academy of Sciences, Olomouc, Czech Republic, [4]4 South Australian Research and Development Institute, Urrbrae, Australia

Species cohesion depends on gene flow to maintain a unified evolutionary pathway. Recombination suppression within inversion polymorphisms disrupts gene flow and drives genetic differentiation between haplotypes, leaving distinct signatures in population variant data, such as high linkage disequilibrium and locus-specific population structure. Yet, such patterns are often missed when statistics are calculated using randomly selected genome-wide markers. The role of inversion in the divergence and speciation of diploid species is being increasingly recognised. Yet, most studies document ancient inversion polymorphisms (>1 MYA) in allogamous species.

We present evidence of extensive genome differentiation in an autogamous diploid species, identified through a multifactorial re-examination of publicly available sequence data integrated with modern cytogenetics. Cicer echinospermum P.H. Davis (2n = 16) is a young legume species, which shared a common ancestor with chickpea (C. arietinum L.) 95–127 KYA. While genome-wide variation analysis separates C. echinospermum into geographically distinct populations, we discovered large structural variations within and between populations. Two large inversion polymorphisms were validated by assembly comparisons, short-read breakpoint mapping, and karyotyping using fluorescent in situ hybridisation. By analysing local patterns in sequence variation, we detected dozens of similar haplotype blocks across the genome comparable to well-studied older inversion polymorphisms in allogamous species, but with distinct SNP signatures demonstrating their recent occurrence. This is the first evidence of rapid, large-scale genomic differentiation in an autogamous species, and suggests we are documenting a rare case of contemporary evolutionary flux.

[B138] Convergent evolution of immune regulation as a foundation of mammalian high-altitude adaptation

Allie Graham[1], Nathan Clark[2]
[1]University Of Kansas, Lawrence, United States, [2]University of Pittsburgh, Pittsburgh, United States
Adaptation to chronic hypoxia in vertebrates has been studied primarily through the lens of oxygen transport and utilization, with extensive focus on hemoglobin evolution, HIF signaling, and cardiopulmonary physiology. While this work has uncovered striking lineage-specific solutions, it has also revealed limited molecular convergence beyond a small set of canonical pathways. As a result, other cellular systems that may constrain or facilitate persistence at high altitude remain underexplored.
Recent physiological and transcriptomic evidence suggests that hypoxia is not solely an energetic stressor but also a potent inducer of inflammation and innate immune activation. Acute low-oxygen exposure activates NF-κB and inflammasome pathways, responses that can drive tissue damage and cardiopulmonary pathology if chronically sustained. Notably, several high-altitude-adapted populations exhibit attenuated inflammatory responses under hypoxia, raising the possibility that immune regulation itself is a key axis of altitude adaptation. Here, we use a comparative genomic framework to test for shared signatures of selection on immune and inflammatory pathways across high-altitude mammals. Leveraging a 120-mammal whole-genome alignment and phylogenetically informed analyses of evolutionary rate shifts, we examine 15 independent high-altitude lineages. Integrating these genomic results with comparative transcriptomic data from high- and low-altitude taxa reveals a recurring pattern: hypoxia-induced inflammatory responses are systematically dampened in high-altitude lineages. Together, our findings reframe high-altitude adaptation as fundamentally a problem of immune homeostasis. Managing inflammation, rather than optimizing oxygen transport alone, appears to be the primary physiological challenge that vertebrates must overcome to persist in chronically hypoxic environments.

[B139] Genetic trade-offs in the pleiotropic architecture of human cardiometabolic health

Anna Basquet Muniente[1,2], Claudia Vasallo[3], Eva Brigos-Barril[1,2], Fabio Barteri[2,3,4], Gerard Muntané[1,5,6], Hafid Laayouni[1,3], Arcadi Navarro[1,2,3,7,8]

[1]Department of Medicine and Life Sciences, Universitat Pompeu Fabra, Barcelona, Spain, [2]Institut de Biologia Evolutiva (IBE), CSIC - Universitat Pompeu Fabra, Barcelona, Spain, [3]BarcelonaBeta Brain Reasearch Center - Pasqual Maragall Foundation, Barcelona, Spain, [4]Museu de Ciències Naturals de Barcelona, Barcelona, Spain, [5]Hospital Universitari Institut Pere Mata (HUIPM), Institut d'Investigació Sanitària Pere Virgili (IISPV-CERCA), Universitat Rovira i Virgili (URV), Reus, Spain, [6]Centro de Investigación Biomédica en Red de Salud Mental (CIBERSAM), , Instituto de Salud Carlos III, Madrid, Spain, [7]Centre for Genomic Regulation (CRG), The Barcelona Institute of Science and Technology, Barcelona, Spain, [8]Institució Catalana de Recerca i Estudis Avançats (ICREA), Barcelona, Spain

Genome-wide association studies (GWAS) reveal extensive genetic overlap among complex traits and diseases, highlighting pleiotropy as a pervasive feature of human genetic architecture. From an evolutionary perspective, pleiotropy constrains adaptation by coupling phenotypes through shared genetic effects and generating trade-offs that limit independent optimization. We investigate pleiotropic relationships between major cardiometabolic diseases and anthropometric and physiological traits used as markers of cardiovascular health. Using large-scale GWAS summary statistics, we detect widespread variant sharing with substantial heterogeneity in effect size and direction. Distinguishing concordant from discordant effects reveals numerous variants consistent with trade-offs across cardiometabolic phenotypes. Notably, gene WNT9B emerges as a pleiotropic hub influencing multiple cardiometabolic phenotypes, whereas the 10q24.32 locus represents a trade-off cluster with discordant effects across traits and additional associations with other complex diseases. Together, these findings frame cardiometabolic disease architecture as a consequence of evolutionary constraint and underscore the importance of pleiotropy when interpreting disease–trait correlations.

[B140] Comparative genomic insights into rediploidisation after whole genome duplication

Anthony Redmond[1], Daniel J. Macqueen[2], Aoife Mclysaght[3], WGDip Consortium
[1]School of Medicine, University College Dublin, , Ireland, [2]The Roslin Institute and Royal (Dick) School of Veterinary Studies, University of Edinburgh, , UK, [3]Smurfit Institute of Genetics, Trinity College Dublin, , Ireland

Genome sequencing projects over the past 25 years have revealed the importance of ancient whole genome duplication (WGD) events in shaping the evolution of diverse eukaryotic lineages through provision of numerous and important genes, often at critical evolutionary junctures. After WGD, the newly generated multiple homologous chromosomes may pair non-preferentially during meiosis, resulting in genetically tetrasomic inheritance. Rediploidisation, the subsequent return to preferential bivalent pairing and disomic inheritance, marks the 'birth' of duplicated loci (ohnologs) from tetrasomic alleles, and hence the onset of duplicate divergence in sequence and function. Consequently, the evolutionary and functional outcomes of WGD centrally depend on this process, which is considered WGD's greatest mystery. Here, we leverage the increasingly dense sampling of publicly available genomes, synthesising across multiple timescales and (mainly vertebrate) taxa, and pair this with development of novel WGD analysis approaches to provide insights into the mystery of rediploidsation. We show that rediploidisation can be asynchronous, resulting in duplicated genomic segments with divergence onsets tens of millions of years apart, including in different species, despite arising from shared WGD events. Thus, some ohnologs may only resolve from alleles (and begin sequence and function divergence) long after WGD, and if speciation has occurred in the meantime they will do so fully independently in each daughter lineage. The temporal uncoupling of WGD and ohnologue emergence has crucial implications for the differential evolutionary outcomes of WGD across species, but also for the methods we use to detect and interpret WGD events across the tree of life.

[B141] Asynchronous evolution of mammalian antimicrobial peptides balances constraint and diversification

Athulya Girish Kizhakke[1], Dipendra Nath Basu[1], Imroze Khan[1]
[1]Ashoka University, Sonepat, India

Antagonistic interactions with pathogens exert strong selection for immune diversity, yet immunity also faces evolutionary constraints due to its essential role in survival. Asynchronous evolution—where immune components are gained, lost, or repurposed independently—may resolve this paradox. Antimicrobial peptides (AMPs) are ancient, diverse components of innate immunity that provide an ideal system for studying the molecular diversification underlying immune system evolution. Using 255 published mammalian genomes—representing species with diverse life histories and ecological niches, and consequently facing distinct immune challenges—and comparative phylogenomics, we estimated gene evolutionary rates and trajectories of protein sequence evolution across 28 AMP families. We also examined how processes such as convergence and divergence shape this evolution. We found that AMP families evolve at different rates across species, with individual AMPs showing accelerated evolution in some lineages but reduced rates in others. Rates of AMP evolution are correlated with ecological factors such as diet and habitat, driving differential evolution of AMPs with specific functions. For example, several AMPs showed accelerated evolution in marsupials, likely due to their unique reproductive strategy of nurturing underdeveloped neonates in pouches exposed to harsh environments. Protein sequences of ecologically relevant AMPs show both convergence and divergence, suggesting site- or domain-specific evolutionary pressures. For example, Bactericidal permeability-increasing protein, which showed a higher evolutionary rate in aquatic mammals, exhibited both convergent and divergent amino acid sites in these lineages. Our results indicate that asynchronous, modular evolution enables immune systems to balance functional constraint with adaptive diversification.

[B142] SwarmGenomics: Unlocking the Potential of Public Genomic Data

Aure Kylmänen[1], Yu-Chi Chen[1], Dr. Sahar Javaheri Tehrani[1], Dr. Nikolas Vellnow[1], Dr. Justin Wilcox[1], Dr. Toni Gossmann[1]
[1]Tu Dortmund University, Dortmund, Germany

Public genomic repositories such as the SRA, GenBank, and ENA contain petabytes of sequencing data spanning all major branches of life. Although generated for specific studies, these datasets remain an underutilised source of evolutionary and population genomic information. We present SwarmGenomics, a comprehensive, user-friendly pipeline that transforms archived sequencing data into a powerful resource for comparative and population genomic research.

SwarmGenomics streamlines the processing of whole-genome sequencing (WGS) data obtained entirely from public archives, allowing users to assemble raw reads, generate reference genomes, and conduct modular analyses with minimal bioinformatics overhead. Built around open science principles, it supports a broad range of evolutionary applications, including estimates of genetic diversity, detection of runs of homozygosity (ROH), inference of demographic history via PSMC modeling, and exploration of repetitive elements, mitochondrial genomes, and nuclear mitochondrial DNA segments (NUMTs).

By treating public repositories as a research “field”, SwarmGenomics demonstrates how large-scale discovery can emerge from reanalysing existing data. Its modular and collaborative design lowers technical barriers, increases reproducibility, and reduces the environmental and financial costs of new data generation, aligning with the symposium’s vision of an open, archive-driven future for molecular evolution research.

[B143] Slow lorises with active viruses: The use of publicly available genomes in the searching for novel endogenous retroviruses

Charles Michie[1], Hayley Free, Prof. Vincent Nijman, Dr. Ravinder Kanda
[1]Oxford Brookes University, , United Kingdom

Endogenous retroviruses (ERVs) are the result of exogenous infections by retroviruses in germline cells. The ERV can then be passed down through generations as part of the hosts genome. The majority of ERV research has focused on humans and other great apes, where their impact on genome evolution and involvement in disease has been well established. However, little is known if these observations hold across the wider primate order as very little research has been conducted outside of Haplorhini. Using two publicly available genomes we investigated ERVs in strepsirrhine primates, with a focus on the endangered slow lorises (Nycticebus). Using RepeatModeler2 and custom bioinformatic pipelines, we identified 15 novel ERV families within the Nycticebus genomes, representing betaretroviruses and gammaretroviruses. Looking at the genomic distribution and population dynamics of the identified families revealed a high amount of diversity, such as copy number and insertion ages. A considerable number of the novel ERV families identified possessed identical LTRs and near complete open reading frames, indicating recent viral integrations, supported by insertional polymorphism. We further use the publicly available genomes and ERVs found to provide insight on the identification of cryptic species, and highlights the usefulness of these structural variants as a measure of genomic diversity. Additionally, there is a high level of interaction between slow lorises, other wild animals and humans due to their involvement in the illegal pet trade and use as traditional medicine, and the risk of zoonotic transmission of these recent viruses to humans cannot be discounted.

[B144] Horizontal gene transfers underpin ribose heterotrophy and central carbon metabolism remodeling in Gloeobacteraceae (Cyanobacteria)

Edi Sudianto[1], Professor Denis Baurain[1], Luc Cornet[1]
[1]University Of Liege, Liege, Belgium

Public genomic repositories enable large-scale reanalysis of evolutionary processes across deeply branching lineages. Using comparative genomics and phylogenetic analyses of publicly available cyanobacterial genomes, we investigate the evolutionary history of Gloeobacterales, an early-diverging cyanobacterial order distinguished by the absence of thylakoid membranes. We show that horizontal gene transfer has played a major role in shaping the central carbon metabolism of this lineage.

In Gloeobacteraceae, one of the two families within the order, we identify a complete ribose ABC importer and associated metabolic enzymes enabling ribose uptake and assimilation alongside photosynthesis, consistent with a photomixotrophic lifestyle. More broadly, core metabolic pathways—including the Calvin–Benson–Bassham cycle, the pentose phosphate pathway, and the Embden–Meyerhof–Parnas pathway—exhibit a mosaic structure resulting from the integration of foreign genes. Phylogenetic analyses indicate multiple independent transfer events from diverse bacterial donors, including other cyanobacteria and Pseudomonadota, with transferred genes dispersed across the genome.

These results reveal Gloeobacterales as a metabolically dynamic lineage shaped by recurrent gene acquisition and demonstrate how systematic reuse of public genomic data can uncover previously unrecognized patterns of metabolic evolution.

[B145] Investigating the predictability of indels across insect

Frances Swift[1], Dr Simon Martin[1]
[1]The University of Edinburgh, , United Kingdom

Compared to single nucleotide polymorphisms (SNPs), we know relatively little about the impact and evolution of structural variants (SVs). Previous work has shown that SVs such as insertions and deletions (indels) can have significant molecular impacts. However, we lack knowledge about the relative abundance of indel variants compared to other types of genetic variation, and we lack predictive understanding about which characteristics of indels are most important in determining their impacts and therefore evolutionary fates.

I will present two analyses investigating the predictability of indels across insects using publicly available data from the Darwin Tree of Life Project. The first analysis uses a MCMCglmm model to assess how consistently pairwise indel and SNP diversity covary across species and genomic compartments. I will describe how indel abundance is generally highly predictable, with outlier cases that can be attributed to differences in genome composition, as well as some small but significant differences between insect orders that may reflect underlying differences in mutational and/or selective processes.

The second analysis uses an emerging method to quantify molecular impact of non-coding indels via individual level measurement of allelic imbalance between two phased haplotype assemblies using RNA-seq data from the same individual. I will briefly present my novel pipeline to quantify allelic imbalance between two phased haplotypes in a reference-bias free manner. I will then demonstrate how this pipeline can be used to explore cis-associations between indels and allelic imbalance at nearby genes to determine how predictable specific indel characteristics are of allelic imbalance across insects.

[B146] Correcting for Global Synonymous Selection Improves the Accuracy of Episodic Positive Selection Inference

Hannah Verdonk[1,2], Dr. Alyssa Pivirotto[3], Dr. Jody Hey[2], Dr Sergei L Kosakovsky Pond[1,2]
[1]Institute For Genomics And Evolutionary Medicine, Temple University, Philadelphia, United States, [2]Department of Biology, Temple University, Philadelphia, United States, [3]Library & Information Technology Services (LITS), Bryn Mawr College, Philadelphia, United States

The ratio of nonsynonymous to synonymous substitution rates (ω) constitutes a fundamental parameter for inferring adaptive protein evolution, predicated upon the assumption that synonymous substitutions are selectively inert. This premise, however, is increasingly untenable given evidence of selection acting on synonymous substitutions, driven by various biological processes such as translational efficiency and mRNA stability. We demonstrate that unmodelled synonymous selection introduces substantial bias into ω estimation, resulting in elevated false positive rates in tests for positive selection. To rectify this, we present BUSTED+S+MSS, a statistical framework incorporating Multiclass Synonymous Substitution (MSS) models into BUSTED, a method for detecting episodic selection. Our approach accounts for global synonymous constraints by partitioning synonymous codons into empirically-derived rate classes. Application to publicly available data from five diverse clades – Drosophila, Caenorhabditis, Enterobacteria, Saccharomyces, and Primates – reveals that the inclusion of MSS components consistently improves model fit and reduces the proportion of genes inferred to be under positive selection. In Enterobacteria, genes retaining significance under the corrected model exhibit weaker constraint on synonymous substitutions (dSs), consistent with the hypothesis that unmodelled purifying selection drives spurious signals of adaptation. Furthermore, an information-theoretic analysis indicates that while site-specific variation (SRV) provides the primary correction, global synonymous rate variation (MSS) contributes a distinct second-order correction. In highly divergent alignments, these signals act together to improve model fit. The BUSTED+S+MSS framework, especially when coupled with an "error-sink" to absorb alignment artifacts, offers a computationally feasible means to disentangle adaptive nonsynonymous substitution from the confounding effects of synonymous constraint.

[B147] No Longer Frog-otten: publicly available anuran genomes reveal a diversity of vertebrate endogenous retroviruses

Hayley Free[1], Charles Michie[1], Professor Richard Nichols[2], Dr Ravinder Kanda[1]
[1]Oxford Brookes University, Oxford, United Kingdom, [2]Queen Mary University of London, London, United Kingdom

The study of endogenous retroviruses (ERVs), genomic remnants of ancient viral infections, has primarily been focused on mammals. ERVs have shaped mammalian genome evolution and influenced disease – but can more be revealed by looking beyond this group of vertebrates, to both untangle the evolutionary history of retroviruses and understand ERVs in non-mammalian genomes? Anurans (frogs and toads) have comparable genome sizes and number of extant species, providing an ideal comparison to mammals. With increasing efforts to sequence the genomes of non-mammalian species, here we construct a bioinformatic pipeline to screen 46 publicly available anuran genomes for ERVs, identifying 56 novel ERV families. Although there are still challenges regarding the assembly quality of several anuran genomes, we identified an abundance of ERV loci with characteristics of recent activity (intact genes and little divergence between their long terminal repeats). We defined an extensive new clade of epsilonretroviruses, possessing a distinctive envelope gene, and prompting reclassification of previously known amphibian and fish ERVs. We also shed light on the relationship of LTR retrotransposons and ERVs, suggesting multiple independent transitions between LTR retrotransposons and retroviruses. The addition of potentially active and recombined retroviruses warrants acknowledgment regarding emerging infectious disease, where anurans are involved in the human-animal interface with use in food consumption, medicine, and the pet trade. The discovery of 56 novel ERV families (with multiple genomic loci) identified in Anura greatly expands the known diversity of retroviruses outside of mammals, and provides fundamental data on understanding these elements across vertebrates.

[B148] A timetree resource for common mammals in zoological parks

Jack Craig[1], Dr. S. Blair Hedges, Dr. Sudhir Kumar
[1]Temple University, Philadelphia, United States

Zoos provide the first and most common encounters most people have with biodiversity, both locally and globally. They are direct interfaces between the public and the pattern and process of evolution, enabling tangible connections between the animals visitors observe, their phylogenetic relationships, and the timing of their origins. Charismatic mammalian megafauna capture the greatest share of this attention and, therefore, are ideal ambassadors. But this educational goal would require a comprehensive timetree of zoo mammals, which does not yet exist because many mammals in zoos represent distinct local varieties (subspecies), such as the Western Gorilla (Gorilla gorilla gorilla) and the Indian Elephant (Elephas maximus indicus). This facet of biodiversity is underrepresented in most large-scale phylogenetic consensus trees since subspecies are typically only a focus in phylogenies of very closely related animals. We address this shortfall by assembling a molecular timetree of 423 mammals from zoos worldwide, including an unprecedented 201 named subspecies across 50 species. This tree and the evolutionary trends it captures, form the basis of a web-accessible educational resource to be made available to zookeepers and patrons as part of the TimeTree of Life database. Users can intuitively subset our tree to match a zoo's collection, then export a range of custom-tailored educational and display materials. This feature offers an opportunity to extend zoos' missions of biodiversity protection and education into the realm of evolution.

[B149] Denisovan variants in CNTNAP2 likely contributed to human adaptation in the Americas

Kelsey Witt[1], Matt Barker[1]
[1]Clemson University, Clemson, United States

Tens of thousands of years ago, modern humans admixed with archaic humans (Neanderthals and Denisovans), and many of those archaic variants are found in modern genomes today. Many of these archaic variants helped modern humans adapt to new environments. The role of archaic variation in human adaptation in the Americas remains under-explored, despite the wide variety of environments present in North and South America. In this project, we explored the evolutionary history and archaic introgression of CNTNAP2, a gene involved in brain development. Previous work identified Denisovan variants of CNTNAP2 in admixed American populations. We further characterized the presence of the Denisovan CNTNAP2 haplotype in ancient and modern populations worldwide, using published genomic data from the Simons Genome Diversity Project and 1000 Genomes Project and published ancient American genomes. We found that the haplotype is almost entirely restricted to the Americas and was found in multiple ancient Indigenous American populations at high frequency, suggesting that it was under selection before European colonization. The haplotype was not likely under strong positive selection but instead shows signals of balancing selection. We also examined the Denisovan haplotype for functional impacts and identified a SNP in an intronic long non-coding RNA, suggesting a role in gene regulation. Our work shows that archaic variants played an important role in human adaptation in the Americas, and further exploration of the functional role of archaic variants will expand our understanding of human evolutionary history and the legacy of archaic introgression.

[B150] Non-canonical DNA studied in bird telomere-to-telomere genomes can explain sequencing drop-out on dot chromosomes

Linnéa Smeds[1], Dr Jacob Sieg[1], Simona Secomandi[2], Dr Chul Lee[2], Dr Francesca Chiaromonte[1], Dr Erich Jarvis[2], Giulio Formenti[2], Dr Kateryna Makova[1]
[1]Penn State University, University Park, United States, [2]Rockefeller University, New York, United States

Non-canonical (non-B) DNA motifs are genomic sequences capable of folding into three-dimensional structures distinct from the canonical right-handed helix. These structures regulate gene expression but also serve as mutation hotspots and are linked to cancer. Because non-B DNA is difficult to sequence, its annotations have been incomplete in most genome assemblies. Telomere-to-telomere (T2T) assemblies now overcome this limitation. Here, we provide a comprehensive analysis of seven types of non-B DNA motifs (e.g., G-quadruplexes and Z-DNA) in the zebra finch (Taeniopygia guttata) T2T genome. Motif content varied strongly by chromosome categories; gene-rich dot chromosomes showed the highest motif levels (16.1-31.7%), microchromosomes intermediate levels (6.5-18.9%), and macrochromosomes the lowest (6.0-7.1%). Within chromosomes, Z-DNA was enriched at some centromeres, and G-quadruplexes were enriched at promoters and 5'UTRs. Low methylation at G-quadruplexes suggests they can form and contribute to gene regulation in these regions. Comparable patterns of non-B DNA distribution were observed in the near T2T chicken (Gallus gallus) genome as well as in several other species across the bird phylogeny. Circular Dichroism validated the in vitro formation of four among the most common G-quadruplexes in zebra finch. Overall, our findings indicate that the non-B DNA distribution reflects the distinctive architecture of avian genomes, implicating non-canonical DNA in gene expression and centromere organization. The unusually high density on dot chromosomes is negatively correlated with PacBio sequencing depth, and thus helps explain why these chromosomes have posed exceptional challenges for sequencing.

[B151] Beyond consensus genomes: a raw-read four-gamete framework uncovers intra-host recombination

Luis Daniel González Vázquez[1,2], Dr Miguel Arenas[1,2], Dr Darren P Martin[3]
[1]CINBIO, University of Vigo, 36310 , Vigo, Spain, [2]Department of Biochemistry, Genetics and Immunology, University of Vigo, 36310 , Vigo, Spain, [3]Division of Computational Biology, Department of Integrative Biomedical Sciences, Institute of Infectious Diseases and Molecular Medicine, University of Cape Town, 7700, Cape Town, South Africa

Viral intra-host recombination has remained virtually invisible to genomic surveillance because standard analyses rely on consensus genomes –unrealistic pseudosequences that show only predominant variants. Mining around 600 000 SARS-CoV-2 short read sequencing datasets that cover every wave of the pandemic, with a recombination detection framework that we developed based on the four-gamete test directly applied to raw next-generation sequencing reads, we investigated intra-host viral recombination, thus considering the signal of rare viral variants. The results provided a rich catalogue of hidden recombination events and traced high-resolution recombination landscapes across the entire viral genomes. The adaptability of our framework was showcased in a parallel scan of around 6 000 HIV-1 short read sequence datasets, where it retrieved numerous allele combinations concordant with recombination. Indeed, control simulations showed high sensitivity and low error rate in our framework, albeit contingent on the genetic diversity present in each sample. These findings underscore the largely untapped potential of analyses of raw reads in molecular evolution studies. Equipped with an intuitive graphical interface, the framework enables large-scale and near-real-time monitoring of intra-host recombination, providing a new dimension for understanding genome-wide viral evolution. L.D.G.-V. was funded by a fellowship from Xunta de Galicia ED481A-2023/089, programa de axudas á etapa predoutoral da Xunta de Galicia (Consellería de Cultura, Educación, Formación Profesional e Universidades) cofinanciado pola Unión Europea no marco do Programa FSE+ Galicia 2021–2027. MA was supported by the Grant CNS2023-144363 funded by MICIU/AEI/10.13039/501100011033 and by European Union NextGenerationEU/PRTR. This study was supported by the Grant PID2023-151032NB-C22 funded by MICIU/AEI/10.13039/501100011033 and by FEDER, UE.

[B152] Toxins among the branches: Evolution of xenobiotic processing gene families in bats

M. Elise Lauterbur[1], Bat1K 21-families group
[1]University Of Vermont, Burlington, United States

Animals encounter an enormous diversity of xenobiotic compounds from plant secondary metabolites and insect toxins to heavy metals and anthropogenic pesticides. Their ability to metabolize these chemicals depends on large, rapidly evolving detoxification gene families. Cytochrome P450 monooxygenases (CP450s), glutathione S transferases (GSTs), sulfotransferases (SULTs), and ATP binding cassette (ABC) transporters frequently undergo lineage-specific expansions and contractions, yet the tempo, mode, and ecological drivers of this gene family evolution remain poorly resolved across mammals.

Bats provide a powerful comparative system for studying toxin adaptation via gene family evolution because they occupy chemically diverse dietary niches and are represented by a rapidly growing collection of high quality genomes in public repositories. Leveraging more than 100 bat assemblies as part of the Bat1K Project, we examine the evolutionary dynamics of major detoxification gene families across Chiroptera. To address challenges inherent to large scale reuse of heterogeneous genomic data, we compare multiple strategies for gene family identification and gene–species tree reconciliation. This framework reveals repeated and independent shifts in CP450 and GST copy number, including striking variation among closely related Myotis species. Across deeper phylogenetic scales, we identify additional cases of copy number evolution in xenobiotic processing pathways consistent with shared selective pressures imposed by chemically similar diets and environmental contaminants. By integrating diverse genomic resources and systematically mining existing datasets, this work uncovers the molecular signatures of toxin adaptation in bats and highlights the broader potential of public genomic archives for studying the evolution of complex, environmentally responsive gene families.

[B153] Comparative evolution of genes across 581 vertebrate genomes reveals lineage-specific adaptive dynamics

Marco Sollitto[1,2], Terence Murphy[3], Claudio Ciofi[1], Erich D. Jarvis[2], Giulio Formenti[2]
[1]Department of Biology, University of Florence, 50019, Sesto Fiorentino, FI, Italy, [2]Vertebrate Genome Laboratory, The Rockefeller University, New York, USA, [3]National Center for Biotechnology Information, National Library of Medicine, National Institutes of Health, 20894, Bethesda, USA
High-quality reference genomes provide an unprecedented opportunity to investigate the evolution of protein-coding genes across vertebrates. Here, we generated and analyzed 581 chromosome-level assemblies from the Vertebrate Genomes Project (VGP), each annotated with either RefSeq or EGAPx evidence-driven annotation pipelines, to produce a comprehensive view of gene evolution. By integrating orthogroup inference, gene family size modelling, and comparative phylogenomic analyses, we identified lineage-specific enrichments in protein-coding genes and functional categories, revealing both expansions and contractions associated with important phenotypic features. Genome-wide screens for positive selection, coupled with structural and functional domain predictions, uncovered candidate genes driving adaptive change in distinct vertebrate clades. Our results highlight recurrent patterns of gain, loss, and accelerated evolution across major lineages, pointing to convergent functional trajectories in immunity, sensory perception, reproduction, and stress response. Taken together, this study provides the most extensive synthesis to date of protein-coding gene evolution, made possible with high-quality vertebrate genomes, demonstrating how standardized annotations and large-scale comparative genomics can illuminate the molecular basis of lineage diversification.

[B154] Comparative transcriptomics suggests that breast secretory epithelium reuses ancestral molecular modules that predate mammalian evolution.

Marie Saitou[1]
[1]Norwegian University Of Life Sciences, , Norway

The mammary gland is a defining evolutionary innovation of mammals. It represents a specialized epithelial tissue with secretory functions regulated by hormonal and transcriptional mechanisms. Although mammary differentiation and function have been extensively studied, the evolutionary relationship between mammary transcriptional programs and those of other secretory epithelia remains insufficiently characterized.

Here, we used bulk RNA-seq data to analyze gene expression patterns in human breast tissue in comparison with other tissues containing epithelial secretory components, as well as non-secretory tissues. Through differential expression analyses, co-expression network reconstruction, and integration with single-cell expression references, we identified transcriptional programs shared across secretory epithelia and those preferentially enriched in breast tissue.

Our analyses delineate three major classes of expression patterns: genes broadly upregulated across multiple secretory tissues, genes showing preferential expression in breast tissue, and genes exhibiting sex-associated expression differences within breast samples. These classes correspond to distinct functional themes, including epithelial organization, metabolic and hormone-responsive processes, and immune-related functions.

Comparative analyses of evolutionary conservation further reveal that many genes preferentially expressed in breast tissue are deeply conserved across vertebrates, indicating that mammary specialization has largely arisen through the regulatory redeployment of ancestral, pre-mammalian gene repertoires rather than through lineage-specific gene innovation. Together, these results support a model in which mammary gland identity emerges from the regulatory re-weighting and integration of conserved epithelial secretory modules. By placing human breast tissue gene-expression data within a comparative evolutionary framework, this study provides molecular insight into how complex organ-level traits evolve through modular regulatory architectures.

[B155] EVOLUTION AND ADAPTATION OF Dothideomycetes: INFLUENCE OF THE DYNAMICS BETWEEN PROLIFERATION AND CONTROL OF TRANSPOSABLE ELEMENTS

Matheus Sampaio Araujo[1], Dr Chirlei Glienke[1], Dr Desirrê Alexia Lourenço Petters-Vandresen[1]
[1]Federal University Of Parana (ufpr), Curitiba, Brazil

Transposable elements (TEs) are mobile genetic elements widely distributed in eukaryotic genomes, and their mobilization frequently leads to negative effects on organisms, due to the possibility of disruption and inactivation of essential genes, as well as the potential for deleterious rearrangements. To minimize these negative impacts, several eukaryotic genomes present defense mechanisms against repetitive sequences, regulating the proliferation of TEs using methylation, RNA interference, and chromatin modifications. In filamentous fungi, the repeat-induced point mutation (RIP) mechanism acts through the activity of DNA methyltransferases, which can generate a bias in the dinucleotide proportion of the affected sequences. However, the proliferation of TEs can also result in positive impacts: in several phytopathogenic fungi, a physical association is observed between TEs and pathogenicity and virulence genes, and the diversification of many of these genes has been mediated by TEs mobilization. Thus, the existence of a balance between the proliferation activity of TEs and the activity of defense mechanisms could play important roles in the evolutionary conflicts between phytopathogens and their plant hosts. Therefore, the objective of this study is to evaluate the influence of the dynamics between control and proliferation of TEs on the evolution and adaptation of the Dothideomycetes, the largest class of fungi from Ascomycota group, using publicly available genomes to characterize the distribution patterns of TEs and RIP signatures. With this broader approach, we expected to obtain a better understanding of the dynamics of this diverse fungi group, exploring gaps regarding the role and influence of TEs in these processes.

[B156] Transition of tetracycline resistance plasmid pT181 from independent multicopy replicon to predominantly integrated chromosomal element over 65 years

Megan Phillips[1], Robert Petit[2], Daniel Weissman[3], Timothy Read[4]
[1]Population Biology, Ecology, & Evolution Program, Graduate Division of Biological and Biomedical Sciences, Laney Graduate School, Emory University, Atlanta, USA, [2]Wyoming Public Health Laboratory, Cheyenne, USA, [3]Department of Physics, Emory University, Atlanta, USA, [4]Division of Infectious Diseases, Department of Medicine, Emory University, Atlanta, USA

Mobile genetic elements (MGEs) facilitate the spread of genes in a microbial population and are major sources of bacterial genetic diversity. One of these MGEs, the small plasmid pT181, confers tetracycline resistance in bacterial pathogen Staphylococcus aureus. pT181 was one of the earliest sequenced S. aureus plasmids and has been isolated in both clinical and livestock-associated strains for decades. Bacterial genome analysis tools and high-quality sequences with metadata are publicly available, but these resources remain underleveraged for examining historical data, especially when studying the evolution of MGEs. Using publicly available reads and metadata, we explore the evolution of pT181 over nearly seven decades of samples to identify trends in sequence evolution, copy number change, and spread across S. aureus and beyond. pT181 was prevalent across S. aureus (found in 9.5% of 83,366 genomes tested), with a conserved sequence outside of three hypervariable regions. The history of pT181 is characterized by spread across strains, variation in plasmid copy number, and increasing presence of the plasmid as an integrated element in the chromosome, rather than as an independent replicon. We have identified multiple chromosomal integration locations of the plasmid, including outside of the previously characterized staphylococcal cassette chromosome mec (SCCmec). We find that pT181 has been transferred across Staphylococcaceae and into a Gram-negative species. Further analysis of underused dimensions of public sequencing data, such as read depth and sample dates, will help us build a more nuanced understanding of microbial genomes and understand how MGEs evolve in a population.

[B157] Comparative ape and human genomics reveals natural protection mechanisms against human diseases

Nachshon Egyes[1], Simon Fishilevich[1], Katharina Lange[1], Nitzan Haim[1], Mimi Shwartz[1], Ron Moran[1], Georgii Bazykin[2], Martin Kuhlwilm[3], Prof. Tomas Marques Bonet[4], Dr. David Gokhman[1]
[1]Department of Molecular Genetics, Weizmann Institute of Science, Rehovot, Israel, [2]Department of Biology, Emory University, Atlanta, USA, [3]Departmentof Evolutionary Anthropology, University of Vienna, Vienna, Austria, [4]Institut de Biologia Evolutiva, Consejo Superior de InvestigacionesCientíficas, Universitat Pompeu Fabra, Barcelona, Spain

Human evolution gave rise to extraordinary adaptations, including the threefold expansion of brain size, the transition to upright walking, and the loss of body hair. At the same time, these changes have also reshaped the genetic architecture of diseases, contributing to the emergence of severe disorders, including schizophrenia, obstetric complications, myocardial infarction, and carcinoma. Here, we investigate the evolutionary dynamics of disease emergence and disappearance by comparing the pathogenicity of Mendelian disorders in humans vs other great apes. Specifically, we intersected high-confidence disease-causing variants from the public ClinVar archive with the recently-released genomes of 332 great ape individuals spanning diverse populations. Strikingly, we identified 382 variants that cause severe diseases in humans, yet segregate without deleterious effects in apes, suggesting protection mechanisms that either evolved in apes or were lost in humans. This tolerance often extends beyond a single variant to protect entire genes, effectively conferring protection against a Mendelian disorder. Interestingly, genes underlying fertility and hearing disorders are particularly likely to become protected in evolution. We further illustrate this principle by uncovering a protective mechanism in chimpanzees against a pathogenic allele in BMP15, which in humans causes complete ovarian dysgenesis, but is the wildtype state in chimpanzees. Together, our targeted approach sheds light on the evolutionary dynamics of disease resistance and provides a framework for leveraging naturally occurring protection mechanisms to inform therapeutic development.

[B158] Comparative genomics provides insights into genome evolution in Orthoptera

Pinli Hu[1]
[1]University Of St Andrews, , United Kingdom

Orthoptera exhibit pronounced diversity in genome size and organization, yet the evolutionary processes shaping their genomic diversity remain incompletely understood. Here, we perform a comparative genomic analysis across representative Orthoptera species using newly generated and publicly available genome assemblies. We investigate genome composition, gene content evolution, and gene order conservation to elucidate patterns of genome evolution in this group.

Comparative analyses reveal extensive gene gain and loss across Orthoptera lineages, with lineage-specific expansions and contractions of gene families contributing to functional diversification. Analyses of gene order uncover varying degrees of conservation and reorganization among species, indicating dynamic genomic restructuring during Orthopteran evolution. In parallel, differences in repetitive element composition further contribute to genomic divergence among lineages. Phylogenomic inference based on genome-wide orthologs provides an evolutionary framework for interpreting these patterns.

Overall, our study highlights the dynamic evolution of Orthoptera genomes, shaped by coordinated changes in gene content and gene order. These findings provide new insights into genome evolution in Orthoptera and establish a valuable comparative genomic resource for future evolutionary and functional studies.

[B159] Evolutionary patterns and processes become better defined as the timetree of life grows larger

S. Blair Hedges[1], Jack Craig[1], Michael Suleski[1], Kelly Abramowitz, John Allard[1], Ava Beasley[1], Dr. Sudhir Kumar[1]
[1]Temple University, Philadelphia, United States

TimeTree is a tree-of-life public repository that synthesizes times of divergence among species from published genomic and other molecular studies and makes original and synthetic data, including divergence times, available to researchers and the public. This resource continues to be mined by diverse scientists, especially evolutionary biologists. Now, with over 5,000 studies and 200,000 species, it contains approximately 10% of all described species. Here, we make use of the entire database to draw inferences about general evolutionary patterns and processes. The current timetree of life continues to show that species diversity is expanding at a constant rate of diversification, overall and in smaller groups. Also, the modal stem time of species is surprisingly consistent across eukaryotes, as is time-to-speciation. However, higher Linnaean taxonomic ranks, in general, show temporal inconsistencies that limit their comparison across different types of organisms. Looking back at how divergence times have changed over the last 30 years shows that most nodes have remained consistent while some key nodes in the tree of life have lowered in divergence time in recent years, possibly from over-calibration by researchers. The latest version of TimeTree has new tools for researchers, especially those studying ecology, evolution, and biodiversity, including a lineages-through-time graphical presentation and pre-computed metrics for use in evolutionary and biodiversity analyses. These metrics include phylogenetic diversity, evolutionary distinctiveness, equal splits, and pendant edge. Data may be downloaded or retrieved through the TimeTree application programming interface.

[B160] Diversification of highly repetitive silk protein heavy-chain fibroin across butterflies and moths

Samantha Standring[1], Danyon Gedris[1], Gabriela Jijon[1], Ashlyn Powell[1], Naomi Young[1], Natalie Goldston[1], Jacqueline Heckenhauer[2], John Sproul[3], Deborah Matthews-Lott[4], Jahir Rayhan[4], Rachit Singh[4], Bert Foquet[4], Akito Kawahara[4], Charlotte Wright[5], Cheryl Hayashi[6], Richard Baker[6], Paul Frandsen[1]

[1]Department of Plant and Wildlife Sciences, Brigham Young University, Provo, United States, [2]Senckenberg Research Institute and Natural History Museum, Frankfurt am Main, Germany, [3]Department of Biology, Brigham Young University, Provo, United States, [4]McGuire Center for Lepidoptera and Biodiversity, Florida Museum of Natural History, Gainesville, United States, [5]Wellcome Sanger Institute, Hinxton, United Kingdom, [6]American Museum of Natural History, New York City, United States

Silk is a fundamental biomaterial used by hundreds of thousands of species across the tree-of-life. Across Lepidoptera (butterflies and moths), silk serves a wide range of functions: larvae construct mats, tunnels, nests, and cases, while pupae build diverse types of cocoons and cocoon attachments. Despite this remarkable behavioral and structural diversity, the genetic underpinnings of silk use across the group remain poorly understood outside of a few model species. Heavy-chain fibroin and light-chain fibroin form the structural core of silk fibers, providing the crystalline structure that underlies the mechanical strength and elasticity of silk. Here, we identify and annotate light- and heavy-chain fibroin genes from public genome assemblies of more than 300 species of Lepidoptera. We find extensive variation in the repetitive region of heavy-chain fibroin both within and among species, likely derived from recombination-based processes such as unequal exchange. We identify 26 species with duplicated heavy-chain fibroin, including three species with multiple duplications. We characterize the silk natural history of all sampled species and find that the amino acid composition in the heavy-chain fibroin protein is correlated with wandering silk, aquatic case building, and leaf folding. The data set that we compiled is the first step in characterizing silk genetics across the diversity of a charismatic order of insects.

[B161] Recycling published data from wild Drosophila populations to investigate de novo gene retention.

Sarah Lucas[1], Marie Lebherz[1], Amanda Glaser-Schmitt[2], Prof. Dr. John Parsch[2], Dr. Pr. Erich Bornberg-Bauer[1,3,4]

[1]University Of Münster, Münster, Germany, [2]Ludwig Maximilian University of Munich, Munich, Germany, [3]Max-Planck Institute for Biology, Tübingen, Germany, [4]Okinawa Institute of Science and Technology Graduate University, Onna, Japan

Genomes are dynamic and mutable, much like the species they encode. Through genomic changes ancestrally non-genic sequence can give rise to de novo genes which are an important source of novelty for species to adapt and evolve. A majority of de novo genes experience a rapid birth-and-death life cycle. Despite the body of literature describing multiple mechanisms for how de novo genes form, it remains unclear what mechanisms help them escape their rapid life cycle to persist and become retained within a genome. One potential mechanism we explore is if de novo genes persist through indirect selection by hitchhiking and being in linkage with genes under positive selection. Using two Drosophila species with a combined 11 populations as a model, we plan to test how often the de novo genes are found near regions under selection. For this work, we have already screened through 120 published datasets to find the ideal reference population for each of our fly strains to calculate FST. The result of this analysis will indicate how prevalent this mechanism is for de novo gene retention. If it is the primary way de novo genes are retained, future research will focus on the de novo genes located near positively selected regions. Alternatively, hitchhiking could represent only a minor portion of how these genes are retained which would encourage future research to investigate alternative mechanisms to retain de novo genes including direct selection on the de novo genes themselves and genetic drift.

[B162] Genetic Pathway-Level Convergence Underlies the Diversity of Mammalian Life-Histories

Theodoros Danis[1]
[1]Vanderbilt University, Nashville, United States, [2]Uppsala University, Uppsala, Sweeden, [3]Vanderbilt University, Nashville, United States

Life-history traits such as gestation length, brain size, and lifespan vary widely across eutherian mammals. Although comparative studies have identified extensive correlations among these traits, the genetic mechanisms underlying their joint evolution remain poorly understood.

Here, we integrate life-history data from 9 traits with genomic data from ~200 species spanning major eutherian mammal clades to investigate the molecular basis of mammalian life-history variation. To identify gene-trait associations, we used a phylogenomic framework linking gene evolutionary rate shifts to phenotypic variation across species. Analyses were conducted across all eutherian mammals, as well as within major clades (Primates, Rodentia, and Chiroptera), using both raw trait values and allometrically corrected residuals for gestation length, lifespan, and brain mass to identify gene–trait associations.

Across all analyses, we identified 66 unique genes whose evolutionary rates were significantly associated with life-history variation. Most gene-trait associations were both trait- and clade-specific. Only one gene, CRYGB, was significantly associated (after correcting for body mass) with more than two traits, namely brain mass, sexual maturity, and gestation length. Accounting for allometric scaling consistently yielded fewer but stronger gene-trait associations, demonstrating that correcting for body size exposes distinct evolutionary signals rather than obscuring them.

Despite the low overlap of genes associated with different life-history traits, we found that these genes function in the same regulatory programs related to development, cell-cycle and transcriptional regulation across traits and taxonomic orders. These results suggest that mammalian life-history evolution arises through pathway-level convergence via lineage-specific genetic routes, reconciling phenotypic convergence with genomic modularity.

[B163] Structural genetic diversity across the Tree of Life: macro-evolutionary patterns of intra-specific structural variation

Thomas Brazier[1], Claire Lemaitre[2], Claire Mérot[1]
[1]Ecobio, CNRS/university Of Rennes, Rennes, France, [2] INRIA, IRISA, GenScale team, Rennes, France
Understanding why some species have higher levels of genetic diversity than others is central in ecology, and has important implications for the evolution and conservation of species. Current research in evolutionary genomics is largely based on Single Nucleotide Polymorphism (SNP), therefore neglecting the genetic diversity and complexity introduced by Structural Variants (SVs). Leveraging the Darwin Tree of Life project – which routinely generates long reads and high-quality genome assemblies for a broad range of taxa accessible in public genomic repositories – we thoroughly reprocess long read data to characterize SV diversity across the eukaryotic tree of life. In addition to providing a necessary and valuable description of SV distribution and determinants on a large phylogenetic scale, this comprehensive dataset allows us to address key evolutionary questions through the lens of structural variation. For example, we investigate how variation in effective population size between species drives the distribution of structural genetic diversity at a macro-evolutionary scale. Genetic diversity also appears to be predicted by the species' biology and ecology. We thus compare species with different life histories (such as longevity and body size), as well as variations in geographic range and population census size, to test whether these factors also apply to SVs. Overall, our integrative analysis, combining genomic and ecological data from diverse public databases (ENA, NCBI, GoaT, Gbif), should be key to understanding the variability of (structural) genetic diversity across species and examining the role of SVs in evolutionary processes.

[B164] Convergent mechanisms underlying polar adaptation in fishes

Tina Cai[1], Joanna Kelley[1], Samuel Bogan[1]
[1]University of California, Santa Cruz, Santa Cruz, United States

Polar oceans are unfavorable to most species on Earth due to their subfreezing water temperatures and high oxygen saturation. However, fish from different suborders and families have independently colonized polar oceans, exhibiting convergent evolution of physiological adaptations. While genomic signatures of polar adaptation have been characterized within individual lineages of fishes, the extent of convergent molecular evolution shared among polar fishes is poorly understood. We compared 53 publicly available, high-quality, long-read genomes of polar and temperate fishes that collectively experienced at least seven polar invasions. We analyzed 5,490 protein-coding genes to detect convergence in positive selection, relaxed selection, and gene family evolution in polar lineages. We found that convergent positive selection in polar lineages was limited to a small subset of genes involved in blood cell formation, immune response, apoptosis, and signal transduction, which have all potentially facilitated the mitigation of cold-induced stresses. Relaxed selection was detected on genes related to heat shock responses, cranial skeletal system development, and intracellular transport: three pathways for which phenotypic losses-of-function have been described in polar fishes. Some genes that underlie adaptations to extreme cold, such as antifreeze proteins and those related to cellular organization, increased in copy number. In contrast, genes less essential for polar adaptation contracted. Our findings identify several pathways in which convergent molecular evolution reflects known convergent physiological adaptations to polar oceans, further illuminating the evolutionary basis of adaptation to this stressful environment.

[B165] Lungworm infection triggers sex-specific effects in gene expression in their cane toad hosts

Tsering Chan[1], Felipe Floreste[3], Greg Brown[2], Boris Yagound[1], Michael Kasumovic[1], Professor Lee Rollins[1]
[1]University Of New South Wales, Sydney, Australia, [2]Macquarie University, Sydney, Australia, [3]University of São Paulo, São Paulo, Brazil

Biological invasions often disrupt host-parasite dynamics, through changes in host density, presence of competition, and novel pathogens. Additionally, sex is known to affect susceptibility to pathogens through physiological and behavioural factors, such as differences in immune investment or habitat use. Yet, the interplay between invasion history and sex in shaping the arms race between host and parasite is poorly understood. Here, we investigated the transcriptomic response of male and female invasive cane toads (Rhinella marina) to infections by their co-introduced parasitic lungworm (Rhabdias pseudosphaerocephala).

We used captive-bred toads raised in a common garden setting to examine changes in host gene expression over the course of lungworm infection. We ran a time-series experiment, sampling lung and spleen tissues at 3, 14, and 28 days post-infection (DPI). Our RNA-seq results showed that the difference in gene expression between control and infected hosts was greatest at 14 DPI in both male and female toads. However, there was a striking sex-specific response to infection, each sex targeting a different organ. In male toads, we found most differentially expressed genes in the lungs, the site of the parasite infection. By contrast, female toads showed a greater response in the spleen, a key organ of the immune system. Our results suggest that male and female toads use different defensive strategies when infected by lungworms, likely reflecting sex-specific differences in life history and ecology within this host-parasite relationship

[B166] Evolution of early animal immunity

Blaire Padayhag, Dibyaswarupa Mohanty[2], Aoife Mclysaght[3], Mark Pi Daniel Macqueen[4], Helen Dooley[5], Anthony Redmond[1]
[1]University College Dublin, , Ireland, [2]University of Limerick, , Ireland, [3]Trinity College Dublin, , Ireland, [4]University of Edinburgh, , U.K., [5]University of Maryland School of Medicine, Baltimore, U.S.A
The complement system is a core frontline innate immune defense in animals. However, while this system is extensively studied in higher vertebrates, its evolutionary origins remain poorly understood. Previous studies have revealed that complement genes are present in many invertebrate lineages but with reduced, or distinct in the case of some arthropods, components. The presence of C3, MASP, and Bf in cnidarians indicates that an ancestral simplified lectin/alternative-type complement pathway already existed in the common ancestor of bilaterians and cnidarians. Despite efforts to identify complement system components in the remaining animal lineages, placozoans, sponges and comb jellies, it is not clear whether the last common ancestor of animals possessed a complement system, or whether it evolved later in animal evolution. Here, leveraging the increasing availability of genomes from these lineages, as well as from close unicellular holozoan relatives of animals, we reassess the origins of the complement system. Our results confidently reveal the presence of a complement gene repertoire comparable to at least that of cnidarians and in sponges, as well as uncovering unexpected ancient gene duplication events in complement gene families. While evidence for a complement system is less clear in comb jellies and unicellular animal relatives, using a novel gene tree analyses method, we recover significant support for the complement system existing in the ancestor of animals under at least some conditions. We present an updated model for the first complement system, including timing the early steps in its elaboration.

[B167] Phylogenetic comparative analysis of APOBEC3 Z-domain gene family evolution: Implications for bat immunity

Brenda Delamonica[1,2], group Bat1K 21 family, Piotr Mieczkowski[3], Simon Anthony[4], Tanya Lama[5], Mani Larijani[6], Thomas MacCarthy[1,2], Liliana M Davalos[7,8]
[1]Stony Brook University, Stony Brook, United States, [2]Laufer Center for Physical and Quantitative Biology, Stony Brook, United States, [3]Department of Genetics, Lineberger Comprehensive Cancer Center, University of North Carolina, Chapen Hill, United States, [4]Department of Pathology, Microbiology, and Immunology, University of California-Davis School of Veterinary Medicine, Davis, United States, [5]Department of Biological Sciences, Smith College, Northampton, USA, [6]Department of Molecular Biology and Biochemistry, Simon Fraser University, Burnaby, Canada, [7]Department of Ecology and Evolution, Stony Brook University, Stony Brook, United States, [8]Consortium for Inter-Disciplinary Environmental Research, Stony Brook, United States

APOBEC3 genes encode enzymes involved in innate immunity which include 1-3 enzymes (A3Z) in most non-primate mammals, having expanded to 7 family members (A3A-H) in primates. Recent studies have shown that bats have even more diversified APOBEC3-like enzymes which outnumber the repertoire of primates. While this expansion is implicated in unique features of bat immunity, studies have evaluated only eight of 1500 bat species. Broader taxonomic sampling of bat diversity would enable testing hypothesized links between APOBEC3 evolution and bat-virus interactions. To this end, we deployed a novel Z-domain search algorithm, Exon Targeted Retrieval and Classification Toolbox (ExTRaCT) to search for sequences encoding the catalytic region, or Z-domain, of APOBEC3 genes in 102 species representing all 21 bat families. We reconciled species and gene trees to infer the evolution of Z-domains across the bat phylogeny and used Bayesian methods to relate Z-domain evolution to viral diversity with known bat reservoirs. We found evolutionary convergence of Z3 domain loss, high birth-death turnover in the Z1 and Z2 domains that leads to increased diversity of APOBEC3 and identified patterns of Z-domain speciation and duplication that correlated, respectively, negatively and positively, with the number of defined viruses carried by a species. While extensive bat diversity constrains broad generalizations about bat A3 evolution, the results from our sample highlight evolutionary processes, instead of gene counts, as better correlates of known bat-virus interactions.

[B168] Tracing the early evolution of bilaterian antiviral immunity through Xenacoelomorpha

Carmen Fontanes Eguiguren[1], Tracing the early evolution of bilaterian antiviral immunity through Xenacoelomorpha Mark Pickering[2], Anthony Redmond[3]
[1]University College Dublin, School of Medicine, Health Sciences Centre, Dublin, Ireland, [2]University College Dublin, School of Medicine, Health Sciences Centre, Dublin, Ireland, [3]University College Dublin, School of Medicine, Health Sciences Centre, Dublin, Ireland

Xenacoelomorpha are a phylum of small, flat-bodied marine worms. Their phylogenetic position is heavily debated, with conflicting studies favouring their being either the sister group to all other bilaterians or to ambulacrarians. As such, they represent a key reference lineage for understanding the early evolution and diversification of bilaterian traits. Despite this importance, Xenacoelomorpha remain severely understudied compared to other deep branching bilaterian lineages, with little known about various aspects of their biology, including their immune system. Characterising immune traits in this lineage may help to assess the extent of immune diversification across deeply divergent bilaterian lineages, including to uncover which immune system components existed in the bilaterian ancestor. Here, taking careful consideration of the rapid evolutionary rate of both Xenacoelomorpha and immune genes combined with deep phylogenetic divergence from other bilaterians, we investigate the Xenacoelomorph antiviral immune repertoire using comparative genomic and phylogenetic approaches. Among other findings, we characterise RIG-I-like receptors (RLRs), antiviral RNA sensing receptors that remain poorly characterized outside classical bilaterian model organisms, in Xenacoelomorpha for the first time. We re-examine both the evolutionary diversification of RLRs, tracing their evolutionary history across bilaterians and other animals, as well as integrating multiple antiviral gene families to assess how antiviral immunity has been shaped during early bilaterian evolution. Our study contributes to understanding of the conserved and lineage-specific features of antiviral RNA sensing in Xenacoelomorpha and Bilateria.

[B169] Population discontinuity in the Paris Basin linked to evidence of the Neolithic Decline

Frederik Seersholm[1], Abigail Ramsøe, Jialu Cao, Philippe Chambon, Karl-Göran Sjögren, PhD Hugh Mccoll, Fabrice Demeter, Charleen Gaunitz, Lasse Vinner, Jesper Stenderup, Gabriele Scorrano, Ralph Fyfe, T. Douglas Price, Morten Fischer Mortensen, Sascha Krüger, Torben Dehn, Svend Illum Hansen, Kristine Vesterdorf, Thorfinn Sand Korneliussen, Morten Allentoft, Kristian Kristiansen, Laure Salanova, Eske Willerslev, Martin Sikora

[1]University of Copenhagen, , Denmark

At the transition between the third and the fourth millennium BC, there is evidence for a population decline concurrent with the end of megalith building across continental northwestern Europe. In Scandinavia this “Neolithic decline” is followed by a massive population turnover, as farming communities disappeared and were replaced by people with steppe ancestry. In Western Europe, however, ancestry associated with Neolithic farmers persisted beyond the Neolithic decline, and it remains unclear whether a similar demographic replacement occurred. To investigate the population dynamics around the Neolithic decline in present-day France, we sequenced 133 ancient genomes from the allée sépulcrale at Bury. Located in the Paris area, Bury spans two burial phases separated by a hiatus with no burial activity: one phase directly preceding the Neolithic decline in the late fourth millennium BC, and a later phase some time after the Neolithic decline. Our analysis revealed that the two burial phases at Bury represented largely discontinuous genetic groups of a markedly different social organisation as inferred from three large pedigrees. We show that the difference between the two burial phases can be linked to a northwards movement of Neolithic ancestry from the south which only spread into the Paris Basin after the Neolithic decline, at around 2,900 BC. Together with genetic evidence of various infectious diseases in the dataset, such as Yersinia pestis and Borrelia recurrentis, these findings detail a population turnover at the end of the fourth millennium BC, offering a possible explanation for the cessation of megalith building.

[B170] Repeated evolution of viral resistance in Caenorhabditis through mutation of a glycosyltransferase gene

Aurélien Richaud[1,2], Cigdem Alkan[1,2], Daria Martynow[1,2], Tony Bélicard[1,2], Nanako Takeda[1,2], Gaotian Zhang[1,2], Marie-Anne Félix[1,2]
[1]Institut de Biologie de l'Ecole Normale Superieure, , France, [2]Centre national de la recherche scientifique (CNRS), , France

Host-pathogen interactions often evolve rapidly within a species, allowing to identify specific factors involved in the interaction using natural genetic resources. Comparative analyses of closely related host-pathogen systems can further reveal conserved mechanisms of interaction. Natural viral infections in two nematodes, Caenorhabditis elegans and Caenorhabditis briggsae, offer a powerful model to address these questions.

The Orsay virus (ORV) is an RNA virus naturally infecting intestinal cells of C. elegans. Although a previously identified deletion polymorphism in the gene drh-1, a homolog of human RIG-I, explains part of the intraspecific variation in ORV replication, the wild strain MY10 remains highly resistant despite carrying the deletion. Here, we dissect the genetic basis of MY10 resistance and demonstrate a rare non-synonymous allele in the gene, gtnt-1, encoding a putative glycosyltransferase, as the causal variant.

Three ORV-related viruses (Santeuil virus, Le Blanc virus, and Melnik virus) infect wild C. briggsae strains. Among strains showing different responses to SANTV and LEBV, we noticed three C. briggsae strains only harboring limited genetic variation, strikingly, including a 45-bp deletion allele in Cb-gtnt-1, the ortholog of gtnt-1, from the C. briggsae strain JU516. We demonstrate the causality of Cb-gtnt-1 in altering host viral infection rate and relative fitness.

In addition to the C. elegans MY10 and C. briggsae JU516 polymorphisms, other wild strains in both species carrying putative null alleles in gtnt-1 and Cb-gtnt-1 also exhibit viral resistance. Altogether, these results suggest repeated evolution of viral resistance through a conserved glycosylation pathway in two closely related Caenorhabditis species.

[B171] Time series RNA-seq shows temporal expression of host resistance related genes

Luca Pecalli[1], Dieter Ebert[1]
[1]University Of Basel, Basel, Switzerland

Coevolution has been suggested to be a major driver of biodiversity, shaping genetic and phenotypic evolution in natural populations. The crustacean Daphnia magna and the bacteria Pasteuria ramosa have been coevolving for at least 15 million years. D. magna genotypes differ strongly in their susceptibility to given isolates of P. ramosa. Genomic studies have identified genomic regions enriched for glycosyl transferases, but strong linkage prevented to pinpoint the genes that are responsible for these resistance polymorphisms. Parasite attachment is believed to be driven by the interaction between the proteins present on the surface of the parasite and the sugars attached to the glycoproteins on the cuticle of the host. To better characterize the molecular actors underlying this interaction, we conducted a time series transcriptomic study in D. magna across the entire moulting cycle. This approach allowed us to resolve the temporal expression of resistance-associated genes and to link them to genes involved in cuticle synthesis. We identified key glycosyltransferases are expressed during the building of the new cuticle and determined their co-expression with specific cuticular proteins. Moreover, we pinpointed three strong candidate genes for functional validation via knock-out or knock-down approaches in future work. Our works complements previous studies in creating a holistic picture of the evolution and maintenance of genetic diversity at resistance loci.

[B172] Rodent Lung Mycobiomes as a Lens for Pathogen Surveillance and Phylosymbiosis

Luisa Zamora Chavez[1], Dr. Dakota Rowsey[2], Dr. Paris Salazar-Hamm[3], Nathan Upham[1]
[1]Arizona State University, Tempe, United States, [2]Science Museum of Minnesota, St. Paul, United States, [3]New Mexico Institute of Mining and Technology, Socorro, United States

Microbial communities play critical roles in the fitness, evolution, and community structure of wildlife hosts. Moreover, zoonotic pathogens can pose threats to biodiversity, human health, as well as create significant economic losses. While bacteria and viruses in microbiomes are comparatively well studied, few systematic studies have focused on mammalian lung mycobiome (fungal microbiome) composition. Using ITS1 metabarcoding of 65 lung samples from five rodent species across three families (Heteromyidae, Cricetidae, and Sciuridae) collected along an elevational gradient in the southwestern United States, we investigate the relative influences of host phylogeny, habitat, and dominance of a single fungal taxon on lung mycobiome composition. Rodents were sampled across desertscrub, desert grassland, interior chaparral, oak woodland, and conifer forest habitats. We detect diverse fungal assemblages, including the potentially pathogenic Blastomyces parvus. Notably, two expected fungal pathogens common in small mammals of this region, Coccidioides and Pneumocystis, are entirely absent from our samples. Lung mycobiome composition is most strongly structured by host phylogenetic relatedness, with closely related hosts harboring more similar fungal communities. Together, these patterns suggest evidence of phylosymbiosis, in which microbiome composition parallels host evolutionary relationships, while also highlighting the capacity for environmental exposure and dominance by particular fungal taxa to modify lung mycobiomes. These insights have implications for regional wildlife management by improving understanding of how host species, evolutionary history, and habitat jointly shape fungal assemblages and by clarifying the role of small mammals as potential reservoirs for emerging fungal pathogens.

[B173] Gill-associated microbiome as indicators of population stress and condition in Eastern Baltic Cod

Maria Cortazar[1]
[1]Uppsala University, Uppsala, Sweden
The skin microbiome contributes to host health by facilitating pathogen exclusion, priming the immune system modulation and maintaining skin homeostasis. We examined how the composition of the skin microbiota is associated with body size and condition in two populations of eastern Baltic cod (Gadus morhua callarias), in the southern (Bornholm basin) and northern (Åland Sea). These locations are characterized by differences in body size and experience distinct hydrographic conditions, including salinity, oxygen and temperature. We used 16s rRNA gene metabarcoding to compare skin microbiota diversity and composition in these populations. We found significant differences between the populations in both alpha and beta diversity, with higher microbial diversity observed in the Åland cod characterized by larger body size. Moreover, we identified a greater number of significantly enriched Gene Ontology (GO) metabolic pathways in Åland cod as compared to those from the Bornholm Basin, with many pathways absent in Bornholm. However, other GOs, such as those associated with the nervous system, were uniquely detected in the Bornholm cod. Differences in microbial communities can be influenced by external environmental factors, determined by the habitats where individual cod reside. These results indicate a potential relationship between the skin microbiome and host health; however, direct evidence for a causal impact remains limited, highlighting the importance of incorporating host skin microbiota interactions in studies of aquatic populations.

[B174] Divergent and convergent evolution in avian TLR4 receptor system

Michal Vinkler[1], Mrs. Seran Yıldız[1], Dr. Marian Novotný[1], Dr. Martin Těšický[2,3]
[1]Charles University, Faculty of Science, Prague, Czech Republic, [2]Ludwig Maximilian University, Faculty of Veterinary Medicine, Munich, Germany, [3]Institute of Vertebrate Biology, Czech Academy of Sciences, Brno, Czech Republic

During co-evolution with pathogens, host immune systems diversify through continual arms races. This process is evident at the interspecific level as adaptive divergence in immune genes and their protein products. However, in response to similar selective pressures, different species may also evolve similar protein features, resulting in molecular convergence. Birds, owing to their broad ecological diversity and the availability of compact, high-quality genomes, provide an excellent system for exploring how positive selection shapes immune gene variation. In this study, we investigate divergent and convergent molecular patterns in two TLR4-associated immune receptor complexes: TLR4–MD2 and CD180–MD1. First, we identified the distribution of positively selected sites within the proteins. These sites frequently occurred at ligand-binding and dimerization interfaces, suggesting functional adaptations that fine-tune pathogen-recognition mechanisms. Next, using 3D protein modelling, we analysed divergence and convergence in surface electrostatic potential patterns. We established a novel approach that restricts protein surface analysis to functionally important regions, thereby increasing the sensitivity of molecular-convergence detection. Surface-charge phenotypes formed clusters that were independent of phylogenetic relationships, indicating multiple instances of convergent evolution in receptor phenotypes across avian taxa. The resulting patterns of amino-acid variation in immune genes were highly complex. Overall, our findings highlight convergent evolution as an important force shaping immune innovation.

[B175] Population-scale discovery and analysis of non-reference endogenous retrovirus insertions in wild house mice

Tomoki Yano, Toyoyuki Takada, Kazumichi Fujiwara, Dai Watabe, Shutaro Hirose, Hiroshi Masuya, Toshinori Endo, Naoki Osada
[1]Hokkaido University, Sapporo, Japan, [2]RIKEN, BioResource Research Center, Tsukuba, Japan, [3]National Institute of Genetics, Mishima, Japan

Endogenous retroviruses (ERVs) represent a major source of structural variation in mammalian genomes, yet their diversity in wild populations remains poorly understood. Here, we conduct a comprehensive genome-wide survey of non-reference ERV insertions in wild house mice (Mus musculus) to characterize their distribution, evolutionary dynamics, and contribution to genomic variation. Using a newly developed bioinformatics pipeline, we detected and annotated over 100,000 non-reference ERV insertions from short-read sequencing data across 163 wild mouse genomes. Our analyses revealed marked differences in ERV insertion patterns among subspecies and populations, including variation in genomic localization and population-specific polymorphisms. These heterogeneous patterns suggest distinct evolutionary histories and host–retrovirus interactions across populations. For instance, we describe the distribution of the ERV-derived Fv4 locus, which shows subspecies-restricted occurrence and confers resistance to murine leukemia viruses (MLVs), illustrating how ERV polymorphisms can intersect with host evolution.
This study provides the first large-scale population genomic scan of ERV diversity in wild house mice. By cataloguing extensive polymorphism in non-reference ERV insertions, our results highlight the role of ERVs as dynamic genomic elements that contribute to structural variation and adaptive evolution.

[B176] Mammal-microbe evolution from global to local scales

Nathan Upham[1], Luisa Zamora Chavez[1], Jessica Neumaier[1], Ryan Nguyen[1], Damien Rivera[1], Julia Nitschmann[1], Alexander Fenlon[1], Gilma De Leon[1], Jonathan Hughes[1], Morgan Pierce[1]
[1]Arizona State University, , United States

Outbreaks of Ebola, monkeypox, and SARS-related CoVs have all originated in wild mammals, as have many disease-causing fungi that colonize small mammal lungs to reproduce. However, the host ecology of these zoonotic diseases is not well understood, lacking predictive rules of which types of species interactions carry the greatest risks of cross-species pathogen transmission and why. To address gaps in mammal-microbe knowledge, our lab is unifying traditional approaches in field mammalogy with genomics and ecological data science. We are pursuing three related projects: (i) a pipeline for re-inferring global mammal phylogeny from public DNA data; (ii) tools for taxonomically updating public data on host-microbe interactions and testing the sensitivity of species-level spillover risk models; and (iii) investigating which types of rodent-virus and rodent-fungal interactions are derived from long-term co-evolutionary relationships. Here, I present results on the latter project, leveraging collections from our elevational survey of wild rodents from 2021-2023 in the Santa Catalina Mountains near Tucson, Arizona, USA (n=10 localities). Focusing on co-distributed groups of deer mice (Peromyscus), pocket mice (Chaetodipus), and chipmunks (Neotamias), we examined patterns of lung fungal diversity (ITS amplicon sequencing) and liver viral diversity (rolling circle amplification) at sites from 1,100–2,400 meters. We find evidence of community-level phylosymbiosis in both fungi and viruses, with elevation also contributing substantially. Strong co-phylogenetic signals among hosts and several microbial lineages (e.g., Blastomyces, anelloviruses) are now being investigated relative to host RNA from the same tissues to characterize states of immune tolerance versus disease.

[B177] Integrating RNA structure and protein interactions to uncover the mechanisms of viral and cellular ires function

Riccardo Delli Ponti[1]
[1]Italian Istitute Of Technology, ,

RNAs fold into complex structures that critically influence gene expression. A prominent class of regulatory elements resides in the 5’ untranslated region (5’ UTR), where internal ribosome entry sites (IRESs) promote cap-independent translation by directly engaging the ribosome. First discovered in viral genomes, IRESs have been classified into four types according to their structural compactness and factor requirements. While viral IRESs are well studied, cellular IRESs remain poorly understood: they display limited sequence conservation, reduced structural compactness, and variable dependence on auxiliary RNA-binding proteins known as IRES trans-acting factors (ITAFs). Whether their activity relies mainly on RNA structure or protein assistance remains unresolved. Here, we present a computational framework that combines in silico mutagenesis and RNA–protein interaction profiling to investigate IRES mechanisms and guide the design of synthetic elements.

[B178] Uncovering the Global HLA diversity

Sim Chin Yee (Faith)[1], Khor Seik-Soon[1], PhD Amit Gourav Ghosh[2], Kim Hie Lim[1,2]
[1]Singapore Centre for Environmental Life Sciences Engineering, , Singapore, [2]Asian School of the Environment, , Singapore

The Human Leukocyte Antigen (HLA) region is the most polymorphic in the human genome due to its key role in mediating immune responses. Its allelic diversity and distribution are shaped by demographic forces such as natural selection, admixture, and epidemiological history. Here, we identified HLA alleles using whole-genome sequencing datasets for 8 key HLA genes (HLA-A, -B, -C, -DPA1, -DPB1, -DRB1, -DQA1, -DQB1) at 3-field resolution. Our dataset comprises 7,502 individuals from 392 ethnic groups and 72 countries, including underrepresented populations from Asia and South America. Ethnic groups in Africa exhibited the highest levels of HLA diversity, with values approximately twofold greater than those observed in lower-diversity regions such as South America, Oceania, Beringia, and Island Southeast Asia. Despite the high diversity of HLA, the principal coordinates analysis (PCA) displayed strong continental clustering based on HLA class I and II alleles, especially among individuals of African, South American and Island-Southeast Asian descent. Finer intra-continental sub-clusters were observed among class I and II genes, distinguishing populations from island vs. mainland Southeast Asia, Mesoamericans vs. South Americans, and even the Han Chinese of the Southeastern region vs. other ethnic groups in China. Together, these findings suggest how population history, environmental conditions, and pathogen-driven selection have influenced HLA allele distribution and diversity in human populations worldwide.

[B179] Phageome dynamics across infant development in a wild primate gut microbiome

Sophia Kottke[1,2,3], Alice Baniel[4], Abebew A. Haile[5], Jacinta C. Beehner[6,7], Thore J. Bergman[7,8], Amy Lu[9], Noah Snyder-Mackler[1,2,10], Joan B. Silk[1,3], Arvind Varsani[2,10,11], India Schneider-Crease[1,2]
[1]School of Human Evolution and Social Change, Arizona State University, , United States, [2]The Center for Evolution and Medicine, Arizona State University, , United States, [3]The Institute of Human Origins, Arizona State University, , United States, [4]Institut des Sciences de l'Évolution de Montpellier, , Canada, [5]Ethiopian Wildlife Conservation Authority, Debark, Ethiopia, , Ethiopia, [6]Department of Anthropology, University of Michigan, , United States, [7]Department of Psychology, University of Michigan, , United States, [8]Department of Ecology and Evolutionary Biology, University of Michigan, , United States, [9]Department of Anthropology, Stony Brook University, , United States, [10]School of Life Sciences, Arizona State University, , United States, [11]Biodesign Center for Fundamental & Applied Microbiomics, Arizona State University, , United States

The dynamics between bacteria and the viruses that infect them (bacteriophages) can impact mammalian health. In early life, shifts in bacterial communities that are driven by phage activity and life cycle have the potential to shape developmental trajectories. The early-life dynamics of the bacteriophage-dominated virome are poorly characterized in primates, which limits our understanding of host-microbe relationships. Here, we addressed that gap by using fecal metagenomic data (n = 244) from wild geladas (Theropithecus gelada) to characterize shifts in phage composition across developmental stages, parallels between mothers and offspring, and proportions of lysogenic and lytic cycle phages (i.e., integration into bacterial genomes vs lysing bacteria). We found that samples from weaned individuals had 64% higher phage richness (exp(β) = 1.64) and higher Shannon diversity (+7.19 units) than those from unweaned individuals, and that weaning significantly explained community structure (R^2 = 0.02, p = 0.001). During the first year of life, infant phageomes shifted from lysogenic phages toward lytic phages, with a 13% increase in lytic phages around weaning (exp(β)=1.126, p=0.004). Gelada offspring had 68% lower phage richness (exp(β) = 0.32) and lower Shannon diversity (-1.54 units) than their mothers (66 samples from 34 pairs). These results demonstrate that phage diversity increases as infants shift from a mother dependent to an independent diet and that shifts in phage life history strategies accompany dietary and physiological transitions.

[B180] Coordinated evolution of the DTX3L–PARP antiviral complex

Léa Picard[1], Emeline Esnouf[2], Stéphanie Jacquet[2]
[1]Okinawa Institute of Science and Technology, , Japan, [2]UMR 5290 MIVEGEC, CNRS, IRD, , France
Viruses are fundamental drivers of host immune diversification, exerting sustained selective pressure on antiviral defense pathways. Understanding how host–virus interfaces have evolved over deep evolutionary timescales is therefore critical for interpreting present-day antiviral immunity and anticipating future viral threats. Here, we investigate the evolutionary history of DTX3L, an interferon-inducible E3 ubiquitin ligase that forms a functional complex with PARP9 and PARP14 to regulate antiviral responses. Using a comparative genomics framework including synteny analyses, phylogenetic inference, positive selection tests, and protein modeling across 128 metazoan species, we show that DTX3L originated from an ancestral duplication event coincident with early bryozoan diversification, followed by a conserved syntenic rearrangement with PARP9, suggesting early functional coupling. In contrast to other members of the DTX family, DTX3L exhibits strong and pervasive signatures of positive selection across mammals. Notably, positively-selected residues are enriched on the surface of the RING and DTC domains, implicated in protein–protein interactions, indicating recurrent evolutionary remodeling at functional interfaces. Consistent with this, both PARP9 and PARP14 also exhibit signatures of strong positive selection, with selected sites clustering within regions involved in complex assembly. Overlapping patterns of episodic selection across multiple mammalian orders suggest coordinated evolutionary pressure acting on the DTX3L–PARP9–PARP14 complex as a functional module, consistent with recurrent host–virus arms races. Together, these findings identify DTX3L as a previously underappreciated but central target of viral-driven evolution, highlighting its key role in shaping vertebrate interferon-mediated antiviral defenses.

[B181] Context-specific immune gene regulation is highly pleiotropic and actively evolving in human populations

Tauras VILGALYS[1]
[1]University Of Georgia, Athens, United States

Pathogens are among the strongest selective pressures across the tree of life, yet immune loci remain highly variable. How natural selection maintains this variation is unclear, but competing selective pressures imposed by diverse pathogens and non-communicable disease risk may play a key role. Understanding the pleiotropic consequences of genetic variation on the immune response and disease risk is therefore critical to understand immune evolution.

Here, I integrate datasets profiling gene expression in primary human immune cells exposed to diverse immune stimuli, identifying over 2,000 genes harboring at least one expression quantitative trait locus (eQTL). Approximately 30% of these eQTL exhibit context-dependent effects, differing between stimulated and unstimulated conditions in at least one cell type. These immune response eQTL have different properties than baseline regulatory variants: they show increased cell-type specificity and are enriched for genes under stronger selective constraint.

Immune response eQTL are highly pleiotropic. They often affect gene expression in the response to multiple pathogens, and are enriched for variants that are also associated with inflammatory and autoimmune disorders. Moreover, these context-dependent regulatory variants are enriched for signatures of recent positive selection relative to other eQTL. These results highlight immune pleiotropy as a central driver of immune evolution, helping maintain immunogenetic diversity. Immune pleiotropy also suggests trade-offs between infectious and non-communicable diseases which may explain why alleles that increase risk for inflammatory and autoimmune disease have been maintained or even favored in recent human evolution.

[B182] Evolutionary responses of Pseudomonas entomophila to insect-associated thermal stress reveal enhanced fitness and virulence

Triveni Shelke[1], Dr Vanika Gupta[2], Dr Ishaan Gupta[1]
[1]Indian Institute Of Technology Delhi, India, New Delhi, India, [2]University of Delhi, Delhi, India
The widespread incidence of insecticide resistance among pests highlights the need to understand insect–insecticide interactions. Bacterial species possess insecticidal properties, and studying insect–pathogen dynamics can provide critical insights for sustainable insecticides. Experimental evolution provides a framework to elucidate the adaptive strategies that pathogens develop to survive and proliferate under insect-driven environmental selection pressures. Pseudomonas entomophila (Pe), an entomopathogen, survives optimally at 30°C, although insects exist in temperatures around 18°C to 30°C. We found that post-infection with Pe in Drosophila melanogaster has lower mortality at 18°C than at 30°C, as Pe struggles to grow at lower temperatures. Therefore, using the serial transfer method, we subjected Pe to thermal selection, reducing the temperature from 30°C to 18°C for 120 days. The evolved bacteria exhibited improved growth under the lower thermal stress and increased virulence, as measured by higher mortality in infected flies. Physiological assays further revealed that the evolved bacteria had enhanced protease activity when compared to the ancestor at 18 °C. Genomic analysis of the evolved bacteria revealed mutations in genes involved in host interaction, secretion systems, and motility regulation. The limited genetic mutations observed led to deciphering the role of 6mA as adaptive memory. Differential methylation in genes related to the cell periphery, transmembrane and ATP binding was noted in evolved bacteria. These patterns suggest that epigenetic modulation may contribute to adaptive responses affecting cellular interfaces and metabolic processes. The findings suggest insect-associated thermal stress drives the evolution of virulence in the entomopathogen with implications for pest-management strategies.

[B183] Genome-wide coadaptation constrains loss of the cag pathogenicity island in Helicobacter pylori

Xiaoqi Zang[1], Prof Daniel Falush[1]
[1]Shanghai Institute Of Immunity And Infection Chinese Academy Of Science, , China
Most Helicobacter pylori strains encode the cag pathogenicity island (cagPAI), which mediates translocation of the CagA oncoprotein into host cells and promotes colonization. Despite this, cagPAI-deficient strains are globally distributed and predominate in parts of southern Europe. Here, we analyzed 9,188 globally sampled H. pylori genomes to investigate how multilocus gene coadaptation shapes the emergence and persistence of cagPAI loss.
Genome-wide association analyses identified strong differentiation at loci encoding the vacuolating cytotoxin vacA and at outer membrane protein gene clusters, including configurations involving hopQ. Aggregating highly differentiated loci into a polygenic coadaptation score revealed a bimodal distribution across populations, indicating the existence of discrete genomic states associated with the presence or absence of the cagPAI. In most populations, cagPAI-deficient lineages occupy a distinct, coadapted genomic background, whereas recently emerged cagPAI losses show intermediate profiles consistent with ongoing genetic adjustment through recombination.
Striking population-specific patterns were observed. In East Asia, cagPAI loss occurs frequently but rarely progresses toward a stable coadapted state, with the notable exception of Okinawa, where a persistent hpEAsia cagPAI-deficient lineage has arisen following recombination with strains of other ancestries. These findings demonstrate that the fitness consequences of cagPAI loss are strongly dependent on genomic background and that loss of this major virulence determinant is constrained by the evolution of a coadapted multilocus genotype.
More broadly, our study illustrates how large-scale gene coadaptation governs pathogen evolution and identifies genetic configurations that may predict the success or failure of virulence loss, with implications for disease risk and eradication strategies.

[B184] Honey bee pathogen profiling: molecular insights into microbial and parasitic communities

Yeahji Jeong[1]
[1]Queen Mary University Of London, , United Kingdom

Insect pollinators are crucial to natural ecosystems and global food security, as 87% of flowering plants require pollination for reproduction. In the UK alone, pollination services contribute over £430 million annually. Among pollinators, honeybees play a dominant role, yet 15-40% of bee colonies are lost annually. One major driver of these losses is infectious disease, with pathogens being a key factor in colony decline. While well-documented pathogens such as deformed wing virus have been extensively studied, broader trends of pathogen infections in honeybees remain largely understudied. Moreover, many potential pathogens affecting honeybees have yet to be investigated, and global environmental changes may be facilitating the emergence of new pathogen variants, further complicating disease dynamics. Notably, honeybee pathogens are increasingly reported to spill over into wild pollinator species, disrupting local pollinator communities and potentially contributing to broader biodiversity declines.

To address this, I employed Kraken, a bioinformatics tool for taxonomic classification. I analysed published RNA-Seq datasets to examine the feasibility of RNA-Seq for detecting pathogens and to find general trends in pathogen infections across different tissues in multiple countries. Additionally, I applied this technique to honeybee samples collected by beekeepers from the British Beekeepers Association to create a pathogen distribution map in England and Northern Ireland, and to investigate correlations between pathogen infections and genetic traits. By exploring whether certain honeybee genetic factors influence susceptibility or response to infections, this study aims to uncover patterns in pathogen distribution, seasonal fluctuations, and potential long-term impacts on honeybee populations and pollinator communities.

[B185] Recombination rate landscape in Lepidoptera group with rapid karyotype evolution

Alice Groudko[1,5], Shakirah Namuli[1,5], Franz Chandi[2,5], María José Sánchez Carvajal[2,5], Alex Arias[2,5], Kimberly Gavilanes Cordova[2], Dr Patricio Salazar-Carrión[1], Caroline Bacquet[2], Prof Kanchon Dasmahapatra[4], Joana Isabel Meier[1,3], Karin Näsvall[1]
[1]Tree of Life Programme, Wellcome Sanger Institute, Hinxton, United Kingdom, [2] Universidad Regional Amazónica Ikiam, Tena, Ecuador, [3]Department of Zoology, University of Cambridge, Cambridge, United Kingdom, [4]Department of Biology, University of York, York, United Kingdom, [5]These authors contributed equally, ,

Neotropical Ithomiini butterflies are among several Lepidoptera clades showing extreme variation in chromosome number, which ranges from 5 to 120 across species and can differ even within the same population. Chromosomal rearrangements (fusions and fissions) can contribute to the establishment of reproductive barriers due to meiotic chromosome mispairing or accumulation of incompatibilities via suppressed recombination, thereby influencing global and local recombination landscape. At the same time, rearrangements themselves can arise from ectopic homologous recombination. Despite the mutual importance of recombination and chromosomal rearrangements, these processes have rarely been studied together, particularly in species with holocentric chromosomes.

In two Ithomiini butterfly lineages – Mechanitis polymnia and Mechanitis lysimnia – sister species with similar chromosome numbers but highly rearranged karyotype, we estimate recombination rates from population-level whole genome sequencing data (184 and 40 individuals, respectively). To reduce the uncertainty from demography estimates, we calibrate the absolute values with low-resolution recombination map from pedigree data. In addition, to obtain direct estimates of meiotic recombination and chromosomal rearrangement in sperm, we develop a novel protocol for material preparation from flash frozen spermatophores for bulk HiC and single cell DNA sequencing. With inferred chromosomal fusions and fissions, this study allows to investigate rearrangement-induced alteration of recombination rate landscape, its relationship with genomic architecture, and consequences for genetic diversity, purifying selection, and local adaptation.

[B186] Divergence and meiotic chromosome behavior along the polyploid continuum

Alison Scott[1]
[1]Max Planck Institute For Plant Breeding Research, Köln, Germany
Polyploids complicate meiotic recombination due to the existence of multiple potential partners for each homolog: autopolyploids have multiple sets of homologs, while allopolyploids are comprised of two sets of divergent homeologs, which may pair regardless. The relationship between genomic diversity and chromosomal behavior is central to our understanding of polyploid genetics, impacting allelic inheritance patterns and shaping the rediploidization process either by promoting diversity or serving as a homogenizing force.

The genus Arabidopsis is polyploid-rich, with multiple natural independent tetraploid lineages. Ranging from from autopolyploids with two sets of homologous chromosomes, to allopolyploids between divergent taxa, these polyploids exhibit a range of meiotic behaviors. Using comparative genomics, population genetics, and cytogenetic methods, I examine the interplay of genomic divergence and meiotic chromosome behavior and how they impact evolution in natural polyploid lineages.

[B187] Recombination rate and efficiency of linked selection in small and large stickleback populations

Mr. Hongbo Wang[1], Chaowei Charlene Zhang[1], Dr. Carley Reid[1], Prof. Juha Merilä[1]
[1]Area of Ecology & Biodiversity, University Of Hong Kong, Hong Kong, Hong Kong

Population genetic theory predicts that natural selection will be more efficient in large than small populations because genetic drift reduces the efficiency of selection in small populations. Small populations adapting to new environments can also be expected to evolve higher recombination rates to facilitate adaptation as well as to dissociate and purge harmful mutations. We tested these hypotheses (1) by investigating differences in the strength of association between nucleotide diversity (π) and recombination rate across the genomes of nine-spined sticklebacks (Pungitius pungitius) from four small freshwater (mean Ne = 2 578) and four large marine (mean Ne = 86 742) populations, as well as (2) by comparing recombination rates between small and large populations using population specific linkage maps. We found the predicted positive correlation of π with recombination rate from all but the smallest freshwater populations, suggesting prevalent linked selection even after accounting for variation in GC/CpG content, and gene density. Mean recombination rates did not differ between freshwater and marine populations, except that the smallest Ne freshwater population exhibited significantly elevated recombination rate. GWAS analyses suggested a polygenic basis for recombination rates. These results suggest an important role for linked selection in reducing π in low recombination regions of both small and large populations. Moreover, as predicted by theory, at least one of the small freshwater populations appears to have evolved a higher recombination rate than its marine ancestors.

[B188] The Pattern of Polymorphism in Arabidopsis thaliana Reflects Non-Random Genome Organization

Elizaveta Grigoreva[1], Dr Thomas J. Ellis[1], Dr Anna Igolkina[2], Dr Polina Yu. Novikova[3], Dr Magnus Nordborg[1]
[1]Gregor Mendel Institute, Vienna, Austria, [2]Ludwig Maximilian University of Munich, Munich, Germany, [3]VIB-UGent Center for Medical Biotechnology, Gent, Belgium

The pattern of sequence polymorphism within and between species has long been of interest because it reflects the interplay of different evolutionary processes. It has frequently been reported that nucleotide diversity varies along the genome in many species. In some, diversity is highest in regions proximal to the centromeres; in others, it is higher in the chromosome arms.

Such variation has been observed across diverse taxa, including fruit flies, Arabidopsis, rice, barley, and humans.

In A. thaliana, diversity shows a striking pattern, with approximately a two-fold increase in pericentromeric regions compared to chromosome arms. From a population genetics perspective, diversity is determined by two forces: the mutation rate (μ) and the effective population size (Ne). Therefore, variation in genomic diversity should reflect variation in one or both of these components.

In this study, we revisit patterns of polymorphism in A. thaliana using long-read genome assemblies, confirming this long-standing observation. The mutation rate difference hypothesis was not supported by divergence data of A. lyrata, and a simple background selection model based on recombination rate is insufficient to explain the observed pattern. However, we find that genes that transposed during the speciation process tend to change their levels of polymorphism according to their genomic neighborhood which might reflect deeper evolutionary process of the genome organization.

[B189] Comparing fine-scale mutation and recombination landscapes in two rhesus macaque populations as assessed from both short- and long-read sequencing

Gabriella Spatola[1], Cyril Versoza[1], Vivak Soni[1], Erangi Heenkenda[1], Jeffrey Jensen[1], Susanne Pfeifer[1]
[1]Center for Evolution and Medicine, School of Life Sciences, Arizona State University, Tempe, United States

Genomic diversity amongst primates is fundamentally shaped by species- and population-specific rates of mutation and recombination. In this study, we infer fine-scale mutation and recombination rate maps for the rhesus macaque (Macaca mulatta) — the most widely used non-human primate model in biomedical research — leveraging both short- and long-read sequencing data from two distinct populations of Chinese and Indian descent. In so doing, we draw comparisons between the rates estimated from each dataset, highlighting both biologically meaningful differences between the populations as well as artefactual differences likely arising from systematic biases and differences in the utilized sequencing technologies. Consistent with previous observations in humans, we find evidence for a high rate of turnover in recombination hotspots over a short evolutionary time span, resulting in population-specific recombination maps. Given that these rates are necessary components for the interpretation of other diversity-shaping processes and events, including those characterizing both the underlying demographic and selective histories, the incorporation of these population-specific maps into future evolutionary models will thus aid both our understanding of primate evolution in general as well as genome-wide association and fine-mapping studies of disease traits studied in these populations specifically. Moreover, as linkage disequilibrium (LD)-based recombination rate estimates from primates have not previously been generated using long-read sequencing data, these results also establish a foundation for future population genomic analyses and inform assessments of their impact on empirical measurements.

[B190] Do similars attract? Extensive association of repetitive sequences characterizes the NORs in an anuran species complex

Helena Milanez[1], Msc Bruno Silva[1], Dr Luciana Lourenço[1]
[1]University of Campinas, Campinas, Brazil

Nucleolar organizer regions (NORs) represent important chromosome markers in the species complex Physalaemus cuvieri – Physalaemus ephippifer, which harbors extensive cryptic diversity. Genetic lineages exhibit variable patterns in NOR number and chromosomal location, features that have been useful even for studying secondary contact zones. Furthermore, the heteromorphic sex chromosomes observed in P. ephippifer (Pep) carry NORs, as do the sex chromosomes of a population derived from secondary contact between Pep and its sister lineage, L1. Preliminary studies revealed different satellite DNAs (satDNAs; i.e. PepBS, PepSat16-147 and PepSat25-282) associated with the nucleolar rDNA within the Pep-L1 clade. Given the molecular dynamics of different repetitive elements, this association is potentially related to rDNA mobility. Therefore, in order to gather insights into this matter, the present study aimed to compare NOR composition across this group through analysis of short-read WGS data and in situ chromosome mapping. The findings revealed higher complexity of the NORs, as additional repetitive sequences (i.e., CL37, CL193, PepSat18-1073) were identified in their intergenic spacers in genomic data. Diagnostic features of transposable elements were suggested in some sequences, whose characterization remains to be further studied. Although most lineages showed similar sequences associated with the nucleolar rDNA, chromosome mapping revealed intra-karyotipic diversity. Moreover, nucleotide divergence analysis of PepSat16-147 suggested that its variants are shared across different clades, and meiotic analysis evidenced recombination between ectopic NORs in L1. Therefore, a complex arrangement of repetitive sequences characterizes these chromosome markers, whose evolutionary dynamic is suggested to be influenced by mechanisms counteracting sequence homogenization.

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[B191] The Evolutionary Dynamics of Extended Recombination Suppression at a Distyly Supergene

Huiqin Yi[1], Aleksandra Losvik[1], Dr Marco Fracassetti[1], Tanja Slotte[1]
[1]Department of Ecology, Environment and Plant Sciences, Science for Life Laboratory, Stockholm University, 106 91, Stockholm, Sweden, , Sweden

Supergenes maintain adaptive multi-trait polymorphisms by suppressing recombination between sets of co-adapted alleles. Although expansion of recombination suppression and associated genomic decay are well-characterized for sex chromosomes, it remains unknown whether similar evolutionary processes operate at other supergenes. We address this fundamental question using the distyly supergene (S-locus) in Linum perenne as a model system. Unlike other Linum S-loci, the S-locus supergene of L. perenne exhibits extensive recombination suppression across a multi-megabase genomic region, providing an ideal natural experiment to test the theory of extended recombination suppression. To characterize the evolutionary dynamics and genomic consequences of this expansion, we have generated a chromosome-scale, haplotype-phased genome assembly of L. perenne and reconstructed the history of recombination suppression through analysis of synonymous divergence. By conducting systematic tests for signatures of genic decay, including elevated dN/dS, gene loss, and transcriptional silencing, we test whether such signatures are more prevalent in older non-recombining regions of the dominant haplotype. This work not only advances our understanding of supergene evolution but also provides a critical empirical case study on how recombination rate variation shapes long-term genome architecture.

[B192] Meiosis (re)invented? Prdm9-independent meiotic recombination in dogs

Jasmin Cichy[1], Linda Odenthal-Hesse
[1]Max-Planck-Institute For Evolutionary Biology, Plön, Germany

Successful recombination is a driving force of evolution, being crucial for sexually reproducing organisms to generate functional germ cells and ensure genetic variation in the offspring. In this fine-tuned, yet enigmatic process, the highly conserved zinc-finger protein PRDM9 in particular was found indispensable in many metazoans: Lack of functional Prdm9 in mammals typically leads to early meiotic arrest and infertility, including in humans.

However, Canidae evolved to lack functional Prdm9 millions of years ago without suffering cytotoxic consequences.

What mechanism saves canine germ cells from meiotic arrest?

Utilizing cutting-edge Nanopore sequencing tools, I so far identified the haplotypes of thirty dogs for seven hotspots with some of the highest recombination frequencies proposed by LD analyses. For the first time, I successfully adapted high-resolution sperm-typing techniques to unravel the canine Prdm9-independent recombination landscape at the fine-scale.

For a hotspot on chromosome 27 overlapping a CpG island and a promoter, my results show significantly different recombination frequencies between individuals – contradicting the suggested uniformity of dog hotspots in the absence of highly polymorphic Prdm9.

Through staining of individual germ cells, we further provide first insights into how stage-specific gene expression is affected by natural Prdm9-loss, and how active transcription and recombination can successfully coexist in dogs.

With these results we can start understanding what canine mechanism is able to overcome any cytotoxic consequences of lacking Prdm9 – and whether it is an evolutionary reactivation of a primordial recombination pathway, or a step forward to a newly evolved DNA repair mechanism.

[B193] PRDM9 in Cnidarians: Structure and evolution of a major driver of recombination maps

Amélie Rudler[1], Laurent Duret[1], Jérémie Vidal-Dupiol[3], Olivier Rey[1], Pauline Buso[3], Riccardo Rodolfo-Metalpa[4], Victor Bonito[5], Océane Romatif[3], Juliette Pouzadoux[3], Jean-François Allienne[1], Catriona Munro[6], Sophie Arnaud-Haond[7], Christoph Grunau[1], Eve Toulza[1], Julie Clement[1]
[1]IHPE, Univ. Perpignan Via Domitia, Univ. Montpellier, CNRS, Ifremer, Perpignan, France, [2]Laboratoire de Biométrie et Biologie Évolutive, Université Claude Bernard Lyon 1, CNRS UMR 5558, Villeurbanne, France, [3]IHPE, Univ Montpellier, CNRS, IFREMER, Univ Perpignan Via Domitia, Montpellier, France, [4]ENTROPIE, IRD, Université de la Réunion, IFREMER, Université de Nouvelle-Calédonie, Nouméa, Nouvelle Calédonie, [5]Coral Coast Conservation Center, Votua Village, Baravi, Nadroga, Fiji, [6]Sorbonne Université, CNRS, Laboratoire de Biologie du Développement de Villefranche-sur-mer (LBDV), Villefranche-sur-Mer, France, [7]MARBEC, Université de Montpellier, Ifremer, CNRS, IRD, Sète, France

Cnidarians, particularly reef-building corals, are subject to rapid environmental changes. This makes their ability to generate and maintain genetic diversity through meiotic recombination essential for their long-term survival. Where and how recombination occurs has important and direct consequences for species adaptation. However, the factors and molecular mechanisms that determine the location of recombination sites in cnidarians are largely unknown.
In many vertebrates, PRDM9, a histone methyltransferase, specifies the position of recombination sites via its DNA-binding zinc finger (ZnF) domain. This ZnF domain evolves quickly and diversifies within species, yielding dynamic and hypervariable recombination landscapes. PRDM9 originated in the metazoan ancestor, yet it has been lost multiple times. Its presence and diversity in early-diverging clades, such as cnidarians, have not been well studied.
Here, we examined 28 cnidarian genomes and searched for the four essential functional domains of PRDM9. We identified full-length PRDM9 in 26 of the 28 species and multiple copies in ten of them, suggesting paralogous functions in recombination. Truncated PRDM9s were also identified in most species, indicating dynamic duplication and loss events.
ZnF diversity was assessed directly from the 28 genome assemblies and by sequencing hundreds of wild individuals across five genus. Substantial variation was observed at amino acid positions that interact with DNA, as expected for PRDM9-driven recombination sites. Taken together, these findings suggest that PRDM9 may be functional in most cnidarians, shedding light on the mechanisms that generate genetic diversity in reef organisms and potentially contributing to their adaptive potential to global changes.

[B194] Fitness consequences of recombination hotspot positioning in dogs and wolf-dog hybrids

Julien Joseph[1], Diana Lobo[2,3], Isabel Salado[4], Daniele Battilani[5], Logan Kistler[6], Audrey T. Lin[6,7], Romolo Caniglia[8], Elena Fabbri[8], Paolo Ciucci[5], Sara Ravagni[5], Carlès Vila[4], Jennifer Leonard[4], Tomaz Skrbinsek[9,10], Astrid Vik Stronen[9,10], Raquel Godinho[2,3], Cristiano Vernesi[11]
[1]Université Claude Bernard Lyon 1, LEHNA UMR 5023, CNRS, ENTPE, F-69622, Villeurbanne, France, [2]CIBIO/InBIO, University of Porto, 4485-661, Vairão, Portugal, [3]BIOPOLIS, Program in Genomics, Biodiversity and Land Planning, CIBIO, 4485-661, Vairão, Portugal, [4]Doñana Biological Station-CSIC, Avd. Americo Vespucio 26, 41092, Seville, Spain, [5]Department of Biology and Biotechnologies "Charles Darwin", Università di Roma La Sapienza, Roma, Italy, [6]Department of Anthropology, National Museum of Natural History, Smithsonian Institution, Washington, USA, [7]Richard Gilder Graduate School, American Museum of Natural History, New York, USA, [8]Unit for Conservation Genetics (BIO-CGE), Istituto Superiore per la Protezione e la Ricerca Ambientale, Via Ca' Fornacetta n. 9, 40064 Ozzano Emilia, Bologna, Italy, [9]Biotechnical Faculty, University of Ljubljana, Jamnikarjeva ulica 101, 1000, Ljubljana, Slovenia, [10]DivjaLabs d.o.o., Aljaževa ulica 35a, 1000, Ljubljana, Slovenia, [11]Research and Innovation Centre-Fondazione Edmund Mach, S. Michele all'Adige, Italy

In many eukaryotes, meiotic recombination events are concentrated in hotspots located in promoter-like features. In other species, the use of these hotspots is limited either because hotspots are directed elsewhere or because these species lack hotspots altogether. The reason for this variability remains unknown, mostly because the fitness consequences of recombining inside or outside promoter-like features are unclear. Recombining in promoters could increase selection efficacy by concentrating recombination close to functional sequences. It could also limit deleterious ectopic recombination in repeated elements, which are mostly located outside promoters. However, since recombination can be mutagenic, recombining in functional sequences could increase the mutation load. Additionally, recombination in promoters could disrupt combinations of co-adapted promoter and enhancer alleles. In this study, we introduce a method for inferring selection on recombination events using whole genomes from admixed individuals resulting from ancient and recent introgression. We then apply this method to ~300 wolf-dog admixed individuals. For both ancient and recent admixture, we demonstrate that breakpoints between wolf and dog ancestry reflect the dog recombination landscape with a strong concentration in promoter-like features. However, breakpoints resulting from ancient admixture are significantly depleted in the three most chromosomal rearrangement-prone repeat families, which are predominantly found outside promoter-like features. Conversely, we found no evidence of selection on recombination events located in selectively constrained promoters. Overall, our results suggest that the main fitness effect of recombining in promoter-like features is to limit deleterious chromosomal rearrangements, with weaker effects on the load of small mutations and linkage dissipation.

[B195] Directional selection on quantitative traits can drive evolution of recombination rate in natural populations

LeAnn Williams[1], Dr. Amy Dapper[1], Dr. Bret Payseur[2]
[1]Mississippi State University, Starkville, United States, [2]University of Wisconsin–Madison, Madison, United States

Meiotic recombination is a core evolutionary process that ensures faithful chromosomal segregation and alters haplotype frequencies. Despite biological constraints on recombination, the rate at which it occurs varies substantially across organisms, and the role of natural selection in shaping that variation remains uncertain. Population genetics models clearly predict that directional selection with negative epistasis indirectly favors the evolution of higher recombination rates. Still, the expected outcome in natural populations is not straightforward, and support from empirical studies is inconsistent. To narrow the gap between theoretical predictions and empirical results, we pursued a novel approach that leverages existing theory but applies it to a more realistic genomic architecture. Using forward simulations of quantitative traits, we measured the capacity for recombination rate to evolve in genomes harboring both recombination rate loci and loci targeted by directional selection on an unrelated trait. Our results demonstrate that existing theory extends to complex genomes. At the same time, the ability to detect evolved differences in recombination rate between populations is reduced by variable outcomes, potentially explaining inconsistencies between previous empirical studies. We also applied our simulation framework to interpret findings from a population of mice that evolved both extreme body size and higher recombination rates on an isolated island. We discovered that when the genetic architecture of body size is complex, strong directional selection is capable of generating the observed amount of evolution in the recombination rate.

[B196] Multi-omics reveal drivers of divergent sex-specific recombination in two passerines

Lisa Ammer[1], Marie Raynaud[1], Dr John McAuley[1], Dr Diana Robledo-Ruiz[2], Petronella Wessman[3], Alexander Lawrence[3], Bertrand Servin[4], Alexandra Pavlova[2], Anna Runemark[3], Henrik Jensen[5], Susan Johnston[1]

[1]University of Edinburgh, Edinburgh , United Kingdom, [2]Monash University, Melbourne, Australia, [3]Lund University, Lund, Sweden, [4]INRAE, Castanet-Tolosan, France, [5]NTNU, Trondheim, Norway

Heterochiasmy, the sex-specific variation in recombination rates and landscapes, is a widespread phenomenon that shapes genetic diversity and evolutionary dynamics. Many passerine species exhibit highly conserved karyotypes, yet show substantial, divergent heterochiasmy at both genome-wide and regional levels. This study employs a comparative approach across two species with opposing heterochiasmic profiles, the house sparrow (Passer domesticus) and the helmeted honeyeater (Lichenostomus melanops cassidix), to investigate the underlying genetic architecture and functional drivers of these patterns.

By mapping over 240,000 crossovers across 13,000 house sparrow meioses, we demonstrate a strong, heritable, female-biased recombination rate. We integrate these landscapes with single-nucleus RNA sequencing (snRNA-seq) of meiotic female germ cells to identify molecular drivers of crossover positioning. Through spatial modelling approaches, we demonstrate how oocyte-specific transcriptional activity likely facilitates protein access to open chromatin, physically shaping the recombination landscape. In contrast, the helmeted honeyeater represents a rare case of genome-wide male-biased recombination among passerines. We apply quantitative genetic frameworks to a pedigree of 257 birds and 53,000 SNPs to characterise the recombination landscape and estimate trait heritability in this species for the first time.

Comparing these divergent systems allows us to move beyond species-specific descriptions and provides a critical baseline for evaluating whether heterochiasmy is driven by conserved genomic constraints or distinct sex-specific pressures. Our multi-omic approach offers new insights into how genetic architecture and sex-specific cellular environments contribute to the evolution of recombination within the avian lineage.

[B197] Genetic differentiation is constrained to regions of suppressed recombination in locally adapted populations with higher gene flow

Dr. Maria Akopyan[1,2], Anna Tigano[2,3], Arne Jacobs[2,4], Aryn Wilder[2,5], Nina Therkildsen[2]
[1]University Of California, Riverside, Riverside, United States, [2]Cornell University, Ithaca, United States, [3]Fisheries and Oceans Canada, Nanaimo, Canada, [4]University of Glasgow, Glasgow, United Kingdom, [5]San Diego Zoo Wildlife Alliance, San Diego, United States

The impact of genome structure on adaptation is a growing focus in evolutionary biology, revealing an important role for recombination landscapes in shaping genetic diversity across genomes and among populations. This is particularly relevant when local adaptation occurs despite gene flow, where clustering of differentiated loci can maintain locally adapted variants by reducing recombination between them. However, the limited genomic resources for non-model species, including reference genomes and recombination maps, have constrained our understanding of these patterns. In this study, we leverage the Atlantic silverside—a non-model fish with extensive local adaptation across a steep latitudinal gradient—as an ideal system to explore how genome structure influences adaptation under varying levels of gene flow, using a newly available reference genome and multiple recombination maps. Analyzing low-coverage whole-genome data from multiple populations, we found a continuum of genome-wide differentiation increasing from south to north, reflecting higher connectivity among southern populations and reduced gene flow at northern latitudes. With increasing gene flow, the number and clustering of FST outliers also increased, with differentiated loci found exclusively within regions of suppressed recombination: chromosomal inversions and putative centromeric regions. Notably, sequence divergence was only evident in inversions, supporting their role in adaptive divergence with gene flow, whereas centromeric regions appeared differentiated because of low recombination and diversity, with no indication of elevated divergence. Our results support the hypothesis that clustered genomic architectures evolve with high gene flow and enhance our understanding of how different types of recombination modifiers may be linked to different evolutionary processes.

[B198] The effect of linkage on polygenic adaptation

Fanny Laugier[1], Matthew Hartfield[1]
[1]University Of Edinburgh, Edinburgh, United Kingdom

Recombination rates strongly influence selection outcomes, causing selected to loci to act independently if rates are high or, conversely, co-segregate together if rates are low. Recombination rates are especially important in influencing the evolution of polygenic traits, which are shaped by multiple loci. Advances in sequencing, theoretical and statistical methods now allow us to better investigate how these traits adapt to their environment. However, many population genetic models and analyses assume that polygenic loci act independently due to free recombination. We have little knowledge of how linkage affects polygenic adaptation, and how it could influence statistical analysis of this phenomena. Here, we aim to quantify how much recombination is required to transition between these two evolutionary regimes. Using a two-locus analytical model, we first investigate how recombination and selection strength jointly determine the dynamics of selected variants. Specifically, we aim to predict the recombination rate at which alleles act independently, to when they jointly fix as a single haplotype. We subsequently test the generality of these predictions using multilocus simulations in SLIM. Specifically, we evaluate whether recombination thresholds inferred from two-locus theory scale with the number and effect sizes of selected loci. Our results will inform when to expect formation of linked adaptive regions in genome data, improving inference and interpretation of analyses. They also shed light on the impact that recombination plays in affecting responses to polygenic selection.

[B199] Genetic determinants of intraspecific variation in crossover frequencies in the honeybee, Apis mellifera

Turid Everitt[1], Luong Hieu Trinh[1], Demetris Taliadoros[1], Tilman Rönneburg[1], Anna Olsson[1], Joachim de Miranda[2], Bertrand Servin[3], Matthew Webster[1]
[1]Uppsala University, Uppsala, Sweden, [2]Swedish University of Agricultural Sciences, Uppsala, Sweden, [3]INRAE, Toulouse, France

Meiotic recombination facilitates natural selection and is necessary for correct chromosomal segregation in most sexually reproducing species. Crossover rates vary greatly both within and among species, but the determinants of this variation are not fully understood. The honeybee Apis mellifera has extremely high recombination rates. Honeybee males (drones) are haploid, which enables the distribution of crossovers to be directly estimated from the progeny of a single reproductive female (queen). Here we map crossover events in the honeybee using whole genome sequencing of 1509 drone progeny of 184 queens. This allows us to assay intra-specific variation in recombination rate and its genetic and non-genetic determinants. We estimate the average crossover rate as 23 cM/Mb, with between 22 and 88 crossovers events detected in individual offspring. We estimate 28% of this variation is additive heritable variation among queens. There is no effect of queen age or genetic background on crossover rate. A genome-wide association study identifies variation in the gene mlh1 as associated with mean crossover rate. We estimate that variation in the gene is associated with a 10% difference in crossover rate between the two homozygous genotypes at the most significant SNP. This gene has a well-established role in recombination and variation in the gene could affect crossover rates by affecting resolution of Holliday junctions as crossovers. This is the first gene found to be associated with recombination rate variation in an insect. Adaptive evolution of this gene could potentially underlie the extremely high recombination rates in honeybees.

[B200] A null model for the evolution of the recombination landscape

Mete Yuksel[1], Puneeth Deraje[1], Dr. Nicole Mideo[1], Matthew Osmond[1]
[1]Dept. Ecology & Evolutionary Biology, University of Toronto, Toronto, Canada

Meiotic recombination is a key event in the life cycles of sexually reproducing species and is thought to confer the primary evolutionary advantage of sex: shuffling of genetic variation. Interestingly, the landscape (i.e., probability distribution) of recombination along chromosomes appears to be rapidly-evolving in many systems. Excitingly, there are now several (gamete-, pedigree-, and LD-based) methods to infer recombination landscapes and test hypotheses about the causes and consequences of their evolution within and between species. However, there are few mathematical models that describe how variation in recombination along the genome manifests and evolves and, thus, a dearth of methods to quantify the tempo and mode of landscape evolution. To this end, we develop a null model for how the recombination landscape evolves under neutrality. In this model, cis modifiers of recombination are laid down, compete for probability, and drift in frequency. The dynamics are approximated by a Gaussian process in space (the genome) and time. The modification rate, effects of modifiers, and distribution of locations where modifiers arise determine the timescale of landscape turnover and the patterns of covariance in historical and present-day recombination events/maps. We show how this naturally gives rise to two methods for inferring parameters of the landscape evolution process — the expected number, location, and effect of modifiers separating individuals, populations, or species — over micro- and macro-evolutionary timescales, respectively.

[B201] The genetic architecture, variation, and evolution of X-chromosomal SpermiR clusters in Mus musculus subspecies affecting recombination rates and hybrid sterility

Rajalekshmi Narayana Sarma[1], Jasmin Cichy[1], Nicole Thomsen[1], Linda Odenthal-Hesse[1]
[1]Max Planck Institute For Evolutionary Biology, Plön, Germany

Meiotic recombination ensures accurate homologous chromosome segregation during gametogenesis, maintaining fertility and preventing aneuploidy, while also generating genetic diversity, a hallmark of sexual reproduction. Recombination rates vary widely across species, sexes, chromosomes, and genomic regions. In mice and humans, PRDM9 directs recombination at hotspots and is the only known hybrid sterility gene, until recently. While hybrid sterility remains oligogenic, the miR465 cluster containing a family of microRNAs on the X chromosome has emerged as a key player, regulating hybrid sterility and the meiotic pachytene checkpoint. However, the specific microRNA(s) responsible were unknown.

Here, for the first time, we resolved the complex, repetitive architecture of the miR465 locus and surrounding microRNAs in the SpermiR region on the second hybrid sterility locus on the X chromosome (HstX2), across 16 mouse strains, from three Mus musculus subspecies, using hybrid assemblies and custom mappings. We found strong synteny with no major structural variations. Reannotation revealed copy number variation in the mir465 cluster, with mature miRNA regions more conserved than loop sequences. Notably, SNVs in the seed region altered secondary structure and target specificity. SNVs in Mmc and CNVs in Mmm correlated with recombination rate and hybrid sterility phenotypes, respectively. We identified validated SpermiR targets that overlapped with meiosis-expressed genes, implicating them in meiotic regulation. The evolutionary trajectory of the SpermiR region mirrors that of the species, underscoring its role in hybrid sterility and reproductive isolation. Our findings reveal how genetic elements at Hstx2 evolve to modulate hybrid sterility and recombination rate.

[B202] Consequences of low effective recombination on deleterious mutations near regions under balancing selection in wild populations of C. elegans

Roman Stetsenko[1], Prof. Erik Andersen[2], Matthew Hartfield[1]
[1]Institute of Ecology an Evolution, University of Edinburgh, Edinburgh, United Kingdom, [2]Department of Biology, Johns Hopkins University, Baltimore, USA

Due to its very high selfing rate, the nematode Caenorhabditis elegans exhibits low effective recombination which is expected to exacerbate the effect of linked selection. This effect of linked selection is particularly visible in hyperdivergent regions which exhibit higher genetic diversity compared to the overall diversity in the genome. Indeed, it was argued that these regions are maintained by balancing selection over long evolutionary timescales. In addition to its effect on neutral diversity, balancing selection can also cause an excess of linked deleterious mutations (some evidence in humans and Brassicaceae). We were interested to know if hyperdivergent regions were enriched for deleterious mutations and if the degree of enrichment depended on the strength of these deleterious mutations. We used whole-genome short-read sequences from 41 Hawaiian isotypes of C. elegans and generated a SIFT database for the N2 reference genome (WS283) of C. elegans. We obtained variants in coding sequences annotated with their SIFT score. The position of hyperdivergent regions for each accession is taken from Lee et al. (2021). We found that hyperdivergent regions harbour a higher proportion of deleterious variants relative to neutral SNPs than the rest of the genome (non-hyperdivergent regions). We also found that this difference between hyperdivergent regions and the rest of the genome is greatly reduced for mildly deleterious variants. These results suggest stronger selective interference in hyperdivergent regions leading to an increased mutation load, highlighting the potential cost of balancing selection in a species with low effective recombination.

[B203] Intra- and interspecific population genomics uncover the emergence of a supergene underlying a morpho-social polymorphism in the ant genus Myrmecina

Romuald Laso-Jadart[1,2], Elise J. Gay[1,2], Joshua Ducancel[1,2], Pascaline Chifflet-Belle[1,2], Stefano Mona[1,2], Claudie Doums[1,2]
[1]Institut de Systématique, Evolution, Biodiversité (ISYEB), Paris, France, [2]EPHE, PSL Research University, Paris, France

Supergenes, through recombination suppression, contribute to the evolution of complex phenotypes by locking functional elements. The ant species Myrmecina graminicola is an ideal system to study their origin, combining social (monogyny versus polygyny) and wing (apterous versus winged queens) polymorphisms controlled by a supergene with three haplotypes: Mo, Ma, Pa. The first two are associated with monogyny and the derived Pa with polygyny. Apterism is caused by an insertion containing the awing gene, within both Ma and Pa.
We assembled the genomes of haploid males from Mo and Pa, using long-read sequencing and Hi-C data, revealing two large inversions. We further investigated the origin and maintenance of both polymorphisms using population genomics, phylogeography, and phylogenomics. We whole-genome sequenced 231 individuals from M. graminicola European populations, together with individuals from sympatric (M. sicula) and allopatric congeneric species (M. nipponica and M. americana). Our results suggest that the insertion harbors higher diversity in Sicily where it most likely integrated the Ma background, through exchanges with M. sicula, before spreading northward. In contrast, Pa is present only in M. graminicola and is almost absent in Italy, and likely originated in Northern Europe ~1 Mya BP, where it captured the awing insertion from Ma, thereby linking polygyny with apterism. This scenario highlights an alternative route to phenotypic divergence, driven by interspecific exchange of genes and their integration within non-recombining regions. We advocate the integration of both intra- and interspecific data to reconstruct the evolutionary history in genomic regions where selection and recombination vary spatio-temporally.

[B204] Recombination dynamics and patterns of molecular evolution in a widespread species with holocentromeres

Dr. André Marques[1], Ruiyi Qiu[1], Dr. André Vanzela[2], Dr. Steven Dreissig[3], Andrés Gatica[4]
[1]Max Planck Institute For Plant Breeding Research, Cologne, Germany, [2]State University of Londrina, Londrina, Brazil, [3]Leibniz Institute of Plant Genetics and Crop Plant Research, Gatersleben, Germany, [4]University of Costa Rica, San Pedro de Montes de Oca, Costa Rica

Recombination shapes genome evolution by modulating linkage disequilibrium and the efficacy of natural selection. In monocentric species, recombination is typically suppressed in (peri-)centromeric regions, which often coincide with low gene density, reduced genetic variation within population, and elevated population differentiation. In contrast, holocentric chromosomes lack a single centromeric region and provide an alternative chromosomal architecture to examine how recombination landscapes relate to molecular evolution.

In the holocentric genus Rhynchospora, recent studies have revealed substantial variation in recombination landscapes among species, ranging from distal-biased to more random distributions along chromosomes, despite broadly even gene distributions. However, these insights are largely based on individual-level data, and how recombination landscape variation relates to sequence diversity, selection, and population differentiation at the population level remains unknown.

Here, we address this gap using Rhynchospora nervosa, a widely distributed holocentric species across the Americas. Preliminary analyses indicate genomic differentiation among populations from distinct geographic regions. Building on this, we are generating population genomic data from more than 350 individuals sampled across diverse habitats, including both contemporary populations and historical herbarium specimens. This extensive spatial and temporal sampling enables us to investigate how recombination landscapes interact with demographic history and local adaptation to shape genome-wide patterns of diversity and selection.

By integrating population genomic analyses with recombination landscape inference, this study provides new insights into how chromosome architecture modulates the relationship between recombination and molecular evolution in holocentric genomes.

[B205] The harmonic mean recombination rate: properties, measurement, and implications

Tyler Kent[1], Carl Veller[1]
[1]University Of Chicago, , United States

Metrics of genome-wide recombination should measure what recombination does. In evolutionary genetics, metrics of genome-wide recombination should therefore measure the degree to which recombination shuffles allelic combinations genome-wide, with reference to the evolutionary processes that this shuffling influences. Here, we explore properties of the harmonic mean recombination rate, rhar, which is known to mediate several important evolutionary-genetic processes, such as the genome-wide reduction in genetic diversity due to linked selection and the reduction in genetic variance of traits under stabilizing selection. We show that, relative to the arithmetic mean recombination rate rbar, rhar is extremely sensitive to both fine- and broad-scale variation in the recombination landscape, and to the density along chromosomes of the loci between which it is measured. Consequently, the effect of recombination on the evolution of a complex trait is sensitive to details of the trait's genetic architecture. We further demonstrate that the number of chromosomes and the distribution of selected sites along chromosomes, via their influence on rhar, likely play a major role in shaping differences between species in the loss of genetic diversity due to linked selection.

[B206] Mutation and recombination rates in free-ranging deer: implications for speciation and demographic inference

Aaron Shafer[1]
[1]Trent University, , Canada

Recombination and mutation generate the diversity required for evolutio. Their rates (rho and mu) are key parameters in population genetic models, yet for most wild populations, empirical estimates remain elusive. Here, I present data from free-ranging white-tailed deer. We identified germline substitutions from whole genome sequencing in known parent-offspring trios allowing for us to estimate the denovo mutation rate. Using population re-sequencing, and both with long-read (HiFi) and Hi-C sequencing of deer semen, we generated a recombination rate and produced comparative recombination landscapes between sister species of deer. We applied these novel rates to temporal estimates of demography to demonstrate the impact on population genetic inferences. Further, we present recombination maps, identifying areas of suppressed recombination, and discuss the implications for speciation in deer.

[B207] South American genetic diversity and adaptive strategies shaped by climatic dynamics since the late Pleistocene

PhD Amit Gourav Ghosh[1,2], Elena S. Gusareva[1,2], Prof. Hie Lim Kim[1,2]
[1]Nanyang Technological University, , Singapore, [2]Genome Asia 100K, ,

South America, the last continent settled by humans, harbours diverse Indigenous populations across extreme and contrasting climates. Ancient genome studies have provided key insights into the peopling of the continent, and others have reported adaptations to extreme environments, particularly in the high-altitude Andes. However, relatively few whole-genome studies of contemporary South American populations exist, limiting our understanding of how past climatic conditions shaped regional demographic histories and adaptive responses.

Here, we integrate paleoclimatic reconstructions with 2,677 whole-genome data, including 55 Indigenous American populations, to investigate demographic history and adaptation across Andes, Amazonia, Gran Chaco, and Patagonia. We identify the formation of deep Amazonian lineages during early-Holocene forest expansion, followed by widespread mid-Holocene population splits associated with regional rainfall reorganisation. In Andes, Chipaya diverged from other highland groups during the Central Andean Pluvial Event (~13 kya), whereas additional Andean divergences coincide with mid-Holocene Lake Titicaca lowstands. Gran Chaco populations exhibit later regional differentiation consistent with prolonged aridity around ~4 kya, while Patagonian demographic histories reflect the impact of Holocene volcanic activity.

We further detect population-specific adaptive trajectories through time. Selection on cardiopulmonary and lung immunity pathways intensifies in the Andes from the late-Holocene, while Amazonians show strengthened antiviral and metabolic stress responses from the mid- to late-Holocene. In the Gran Chaco, genes related to water reabsorption and xenobiotic metabolism display contrasting selection histories, whereas Patagonians exhibit sustained selection on thermogenesis-related pathways since the mid-Holocene. Together, our results demonstrate how climatic heterogeneity has shaped demographic structure and adaptive responses across South America.

[B208] Detecting Human-Specific DNA Methylation Changes through Comparative Analysis of Great Apes and Ancient Hominins

Chen Leibson[1], Anat Marom[1], Malka Nissim-Rafinia[1], Alaa Darwish[1], Sara Issac[1], Amir Eden[1], Benjamin Yakir[2], Prof. Eran Meshorer[1,3], Prof. Liran Carmel[1]
[1]Department of Genetics, Alexander Silberman Institute of Life Sciences, Hebrew University of Jerusalem, Jerusalem, Israel, [2]Department of Statistics, The Faculty of Social Science, Hebrew University of Jerusalem, Jerusalem, Israel, [3]Edmond and Lily Safra Center for Brain Sciences, Hebrew University of Jerusalem, Jerusalem, Israel

Human lineages, including anatomically modern and archaic humans such as Neanderthals and Denisovans, exhibit profound morphological and behavioral differences from nonhuman African apes. Because regulatory divergence is thought to be a primary driver of phenotypic evolution, we sought to identify epigenetic shifts occurring since the human-chimpanzee split.
Utilizing DNA methylation as a proxy for gene expression in ancient samples, we compared methylation maps across modern humans, archaic humans, and nonhuman African great apes. We experimentally generated whole-genome methylation maps from the bones of a gorilla, two chimpanzees, and six modern humans. These were integrated with reconstructed profiles from a Neanderthal, a Denisovan, and 28 high-coverage ancient modern humans, supplemented by methylation array data.
By applying stringent filtering criteria, we identified over 350 differentially methylated regions (DMRs) that distinguish humans from nonhuman African apes. These DMRs are most significantly enriched in pathways related to bone remodeling and heart formation and function. Furthermore, by using the gorilla as an outgroup, we identified dozens of DMRs specific to the chimpanzee lineage. Finally, we investigate the underlying mechanisms of these epigenetic changes, including their association with specific genomic features and the evolutionary gain or loss of CpG sites. These results provide insights into the regulatory landscape of human evolution.

[B209] Ancient DNA reveals the northeastern Tibetan Plateau as a hub of regional populations during the Bronze Age

Chengcheng Zhang[1], Ke Wang[1]
[1]Fudan University, Shanghai, China

The northeastern Tibetan Plateau served as a critical corridor connecting the Yellow River Basin with highland populations, yet the genetic dynamics underlying Bronze Age cultural transitions in this region remain incompletely understood. Here, we present genome-wide data from 28 Bronze Age individuals from the Shangsunjia cemetery in Qinghai, associated with the Kayue and Tangwang cultures. Our analyses reveal that the Shangsunjia population falls within the East Asian genetic continuum and exhibits close affinities with Neolithic groups from the northeastern Tibetan Plateau, the upper Yellow River, and the West Liao River Basin. We observe genetic continuity across the Kayue-Tangwang cultural transition, with no evidence for large-scale population replacement, although rare Tangwang outliers show limited external gene flow from Mongolian Plateau-related sources. Admixture modeling indicates that both groups derive primarily from a mixture of Zongri-related plateau ancestry and Late Neolithic Yellow River farmer ancestry, reflecting sustained interaction between highland and lowland populations. Additionally, Qinghai-related ancestry contributed to the formation of several populations in southwestern China, consistent with dispersals along the Tibeto-Burman corridor. These findings establish the upper Yellow River and northeastern Tibetan Plateau as a genetic hub linking the Central Plains, the West Liao River region, and southwestern China throughout the Bronze Age.

[B210] The genomics of the first identified Irish Mesolithic community

Corey Alwell[1], Dr. Lara Cassidy[1]
[1]Department of Genetics, Trinity College Dublin, Dublin, Ireland

Continuous human occupation of Ireland began around ten thousand years ago, when hunter-gatherer (HG) populations from Europe made their way to Irish shores. Only two genomes of individuals from this Irish Mesolithic have been sequenced, both from near the end of this era. Here, we present new whole genome data for Irish Mesolithic individuals from Killuragh Cave in the south of the island. The individuals recovered from Killuragh share highly inflated levels of identical-by-decent (IBD) segments with one another, making this the first Mesolithic community characterised in Ireland or Britain. No evidence of strict patrilocality or matrilocality is observed, as both Y chromosome and mtDNA diversity are high. We further find little evidence of sex-biased demography across HG Europe, in keeping with ethnographic datasets. In the European context, IBD data show greatest connection of Irish HGs with Western Hunter-Gatherer individuals of North-Western and Northern Europe, rather than Atlantic coastal regions, suggesting a continental, and not Atlantic, initial colonisation of Ireland. Also leveraging IBD, we present a holistic appraisal of population size trajectories for many major European HG groups, spanning the period ~35-6kya, identifying periods of growth, stability and decline. We show that the island population size trajectory of Irish HGs is unique amongst these European groups, with Ireland experiencing a severe population bottleneck coinciding with the 8.2kBP climatic cooling event. What follows is a period of extreme island isolation, resulting in some of the greatest degrees of Runs-of-Homozygosity (RoH) identified in any pre-farming population.

[B211] Fault-tolerant pedigree reconstruction from pairwise kinship relations

Edward C. Huang[1,2], Kevin A. Li[3], Vagheesh M. Narasimhan[3]
[1]Stanford University, Stanford, United States of America, [2]Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany, [3]The University of Texas at Austin, Austin, United States of America
Pedigrees reconstructed from biologically related ancient genomes have revealed many insights into (pre)history. To our knowledge, all reported ancient pedigrees have been manually reconstructed, as existing pedigree reconstruction methods are ill-suited for the quality and nature of ancient DNA data. Here, we introduce repare, an open-source software method to automatically reconstruct pedigrees from inferred pairwise kinship relations, which are readily obtainable from ancient genomes. This method reconstructs pedigrees by iteratively incorporating pairwise kinship relations into a set of candidate pedigrees, with pruning and sampling to reduce its search space. It optionally considers supporting information such as haplogroups and skeletal age-at-death estimates. We evaluate this method on a variety of simulated pedigrees with varying error rates and missingness. We also use this method to reconstruct several published pedigrees that were originally manually reconstructed; for one, we present a potential alternative topology. repare optionally incorporates user-inferred pedigree constraints, enabling "human-in-the-loop" reconstruction workflows. Especially when used with these user-inferred constraints, we find that repare represents a powerful and flexible tool for ancient pedigree reconstruction.

[B212] Establishing biological links on a small and large scale: palaeogenetic study of individuals from the Saint-Memmie hypogeum (Marne, 3000 BCE)

Félicie Beck[1], Maël Lefeuvre[1], Isabelle Richard[2], Marie-Claude Marsolier[3], Céline Bon[1]
[1]UMR 7206 Eco-anthropologie, Muséum national d'Histoire naturelle, CNRS, Université Paris-Cité, Paris, France, [2]Institut National de Recherches Archéologiques Préventives (Inrap), Paris, France, [3]Institute for Integrative Biology of the Cell (I2BC), Institut des Sciences du Vivant Frédéric Joliot, CNRS UMR 9198, CEA Saclay, Gif-sur-Yvette, France

Hypogea are large collective burials that were particularly common in the Paris Basin during the Late Neolithic period.
The hypogea of Saint-Memmie and Les Mournouards II (Marne), dating to 3000 and 3300 BCE respectively, are two notable examples, each having been used for the burial of around fifty individuals. Paleogenetic analyses of the human remains found in these hypogea raise questions about the origin of these populations and the existence of long-distance mobility during the Late Neolithic period.

These studies first revealed that individuals from Saint-Memmie and Les Mournouards II are closely related, and that these hypogea seem to have been used as familial burials. Secondly, individuals from Saint-Memmie and Les Mournouards II had a greater Western Hunter-Gatherer (WHG) Mesolithic ancestry than individuals from the Middle Neolithic Paris Basin. Such an increase in WHG ancestry has also been observed in contemporary individuals associated with the Wartberg culture (Rhineland-Westphalia) in northwestern Germany. Beyond their genetic similarities, cultural links in funerary practices have been observed between these two communities.

To better understand the relationships between these communities (Late Neolithic Paris Basin populations and Wartberg culture groups), we conducted analyses of identical-by-descent (IBD) segments.

These analyses revealed distant biological relationships between individuals from the two groups, supporting the hypothesis of a common origin. They also allowed us to identify biological links and to highlight patterns of community mobility between northeastern France and western Germany during the Late Neolithic period.

[B213] Reconstructing the Bantu expansion through an individual-based simulation integrating genomic and linguistic evolution

Francesco Giannelli[1,2], Marco Gargano[3], Harald Hammarström[4], Carina Schlebusch[1,2,5]
[1]Human Evolution Program, Department of Organismal Biology, Evolutionary Biology Centre, Uppsala University, Uppsala, Sweden, [2]Center for the Human Past, Department of Organismal Biology, Uppsala University, Uppsala, Sweden, [3]Department of Life and Environmental Sciences, Marche Polytechnic University, Ancona, Italy, [4]Department of Linguistics and Philology, Uppsala University, Uppsala, Sweden, [5]Palaeo-Research Institute, University of Johannesburg, Johannesburg, South Africa
The Bantu-speaking population's expansion across Sub-Saharan Africa is one of the most significant demographic and cultural events in the history of the African continent. Previous studies have combined linguistic, archaeological and genomic data to infer possible migration routes, but despite the efforts, the dynamics of this expansion remain debated. Here we present an individual-based, forward simulation framework in a dynamic landscape setting to reconstruct Bantu-speaking people's expansion over the last ~5,000 years. Our model integrates both genomic and linguistic evolution, comparing our results with modern and ancient genome-wide SNP data and Bantu language data, to explicitly test scenarios of eastward, westward, and bidirectional expansions, investigating which expansion model best reproduces the observed genetic and linguistic diversity distributions. This framework is the first to integrate genomic and linguistic evolution in a two-dimensional, landscape-aware simulation that accounts for environmental change over millennia. This work offers new insights into how human populations spread, adapt to changing environments, and co-evolve genetically and linguistically across large spatial and temporal scales. Preliminary results show that the model performs as intended: genetic differences increase with geographic distance, and linguistic differences broadly follow genetic patterns. Overall, this supports the idea that language evolution largely reflects population history, while still allowing us to identify a few meaningful exceptions.

[B214] Understanding sedaDNA taphonomy through time

Frederikke M. Sønderborg[1], Pablo Nicolás Arellano Caicedo[2], Magnus August Ravn Harding[2], Taru Verma[2], Karina Krarup Sand[2], Ph.D. Elena I. Zavala[1,2]
[1]Department of Forensic Medicine, University Of Copenhagen, Copenhagen, Denmark, [2]Globe Institute, University of Copenhagen, Copenhagen, Danmark

Sedimentary ancient DNA (sedaDNA) is increasingly used in palaeoenvironmental and archaeological research for exploring past environments and human occupations. It has even been shown that hominin DNA recovered from sediments over 100,000 years old can be used to identify population turnovers. However, limited understanding of how sediment mineralogy affects DNA preservation and recovery constraints both sedaDNA recovery and interpretation.
Minerals exhibit distinct DNA adsorption and preservation properties that depend on environmental conditions, and some minerals, such as clay, can effectively preserve DNA through adsorption while hindering extraction. In addition, increasing our understanding of sedaDNA taphonomy has the potential to provide information about how recovered DNA was deposited.

In this study, we investigate these dynamics using sediment from Bronze Age sites in south-west England that have known archaeological contexts as case studies.
Peat sediments from a log coffin and clay-rich sediments from two settlements were characterized for their mineral composition and pH, and analyzed for the presence of ancient environmental DNA. We observe clear mineralogical differences within and between sites (i.e. clay types) which relate to variation in the capacity for DNA adsorption, and highlight the utility of sample-specific extraction and processing protocols. We also explore how DNA adsorption to the identified minerals is influenced by the body fluid from which the DNA originates.
These findings allow us to examine the interplay among mineralogy, DNA adsorption, and taphonomic processes, and how they influence sedaDNA preservation over time, providing key insights into how we can use sedaDNA to trace human evolution.

[B215] Centromere-Spanning Haplotypes Provide Insight into Human Evolution

Hailey Loucks[1], Sasha Langley[2,3], Fedor Ryabov[1,4,5], Julian K. Lucas[1], Julian Menendez[1], Prajna Hebbar[1], Maxamillian G. Marin[6,7], Andrew P. Blair[1], Taylorlyn Stephen[1], Gary H. Karpen[3], Viviane Slon[8], Richard E. Green[1], Karen H. Miga[1], Ivan A. Alexandrov[8], Charles H. Langely[2]
[1]UC Santa Cruz Genomics Institute, University of California, Santa Cruz, United States, [2]Department of Evolution and Ecology, University of California Davis, Davis, United States, [3]Department of Molecular and Cell Biology, University of California, Berkeley, Berkeley, United States, [4]Centre for Biomedical Research and Technology, HSE University, Moscow, Russia, [5]Center for Molecular and Cellular Biology, Moscow, Russia, [6]Department of Data Science, Dana-Farber Cancer Institute, Boston, United States, [7]Department of Biomedical Informatics, Harvard Medical School, Boston, United States, [8]Department of Anatomy and Anthropology & Department of Human Molecular Genetics and Biochemistry, Faculty of Medical and Health Sciences, Tel Aviv University, Tel Aviv, Isreal

Human centromeric regions undergo limited meiotic recombination, generating multi-megabase regions of linkage disequilibrium that preserve long-range haplotypic structure across populations. These centromere-spanning haplotypes (cenhaps) therefore retain deep evolutionary signals, yet their population dynamics and evolutionary history remain poorly characterized. Here, we investigate the origin, structure, and evolutionary trajectories of cenhaps across global human populations. We identified cenhap lineages across all chromosomes and estimated the ages of deeply divergent lineages by integrating population variation data from the 1000 Genomes Project with hundreds of complete genome assemblies from Human Pangenome Reference Consortium (HPRC) Release 2. Our analyses revealed multiple cenhap lineages with rapid frequency increases consistent with positive selection, including a chromosome 12 lineage that arose >0.5 million years ago and expanded to >30% frequency in out-of-Africa populations within the past 70,000 years ($p<0.001$). We also detected centromeric haplotypes introgressed from Neanderthals, showing high sequence homology across both repetitive and non-repetitive regions, whereas Denisovan contributions were limited. Neanderthal ancestry was further associated with copy number variation in centromere-proximal genes, including ALG10 and olfactory receptor clusters. Slight sequence variants of centromeric higher-order repeats were lineage-specific and provide informative markers for assigning cenhap identity in low-coverage ancient genomes. Together, these findings demonstrate that cenhaps encode rich evolutionary and population-genetic information and establish a scalable framework for analyzing centromeric diversity, archaic introgression, and selection across human history.

[B216] Genomic mosaic structure in mainland Southeast Asia through haplotype-based analyses of ethnic minority groups

Koki Yoshida[1], Kazunari Matsudaira[2], Nao Kiso[1], Wannapa Settheetham-Ishida[3], Danai Tiwawech[4], Surin Pookajorn[5], Phaiboon Duangchan[6], Nancy Bird[7], Yusuke Watanabe[1], Nao Nishida[8], Katsushi Tokunaga[9], Ryosuke Kimura[2], Paul Verdu[10], Garrett Hellenthal[7], Takafumi Ishida[1], Hiroki Oota[1]
[1]Department of Biological Sciences, Graduate School of Science, The University of Tokyo, Tokyo, Japan, [2]Department of Human Biology and Anatomy, Graduate School of Medicine, University of the Ryukyus, Okinawa, Japan, [3]Faculty of Medicine, Khon Kaen University, Khon Kaen, Thailand, [4]National Cancer Institute, Bangkok, Thailand, [5]Faculty of Archaeology, Silpakorn University, Bangkok, Thailand, [6]Srinakharinwirot University, Bangkok, Thailand, [7]UCL Genetics Institute, Department of Genetics, Evolution and Environment, University College London, London, UK, [8]Department of Genomic Function and Diversity, Institute of Integrated Research, Institute of Science Tokyo, Tokyo, Japan, [9]Genome Medical Science Project, National Institute of Global Health and Medicine, Japan Institute for Health Security, Tokyo, Japan, [10]MR7206 Eco-anthropology, CNRS-MNHN-Université Paris Cité, Paris, France

Mainland Southeast Asia (MSEA) is home to diverse ethnic minority groups, as well as urban populations. The population history of MSEA has often been explained by the Two-layer model, which posits that present-day hunter-gatherers are direct descendants of ancient hunter-gatherers associated with the Hoabinhian lithic culture that persisted from more than 40,000 years ago until around 4,000 years ago, whereas agriculturalists descend from farmers who migrated southward from East Asia around 4,000 years ago. Although this model has gained support from recent physical anthropological studies, increasing evidence suggests complex admixture between hunter-gatherers and agriculturalists, leaving the fine-scale population history unresolved.

In this study, we analyzed genomic data from ethnic minority groups in Thailand, including hunter-gatherers, collected several decades ago and therefore considered relatively less affected by recent admixture. By combining SNP array data from these populations with ancient genomes, we applied a haplotype-based approach—less sensitive to the genetic drift in small populations—to reassess population structure in MSEA within the Two-layer model framework.

Using Neolithic agriculturalist individuals excavated in MSEA (MSEA_LN) and a Hoabinhian-associated hunter-gatherer individual as source populations, we estimated ancestry composition across ethnic minorities. All examined ethnic minorities harbored both ancestral sources. Agriculturalist minorities were predominantly MSEA_LN-related, whereas the hunter-gatherer group, Mani (Maniq), retained nearly 50% Hoabinhian-related ancestry. Additionally, other non-agricultural minority groups showed an ancestry profile similar to that of agriculturalist groups, suggesting a recent subsistence shift.

These results provide new support for the Two-layer model and insight into the population history of ethnic minority groups.

[B217] Tracing the demographic shifts of a Roman city on the Danubian Frontier during the Migration Period through paleogenetics

Leonardo Vallini[1], Jens Blöcher[1], Laura Winkelbach[1], Regina Molitor[2], Ivan Bogdanović[3], Francesca Gentilin[1], Dieter Quast[2], Ilija Mikić[3], Joachim Burger[1]
[1]University Of Mainz, , Germany, [2]Leibniz-Zentrum für Archäologie, Mainz, , Germany, [3]Institute of Archaeology, Belgrade, , Serbia

During Late Antiquity, the Roman city of Viminacium – located in present-day Serbia on the Danubian frontier at the crossroads of the Balkan Peninsula, the Black Sea and the western part of the Roman Empire - acted both as a commercial hub and a strategic military base. It eventually developed into one of the largest cities in the region becoming the capital of the province of Upper-Moesia. With the transition from Late Antiquity to the Migration Period, this region of the Roman Empire underwent profound political and social transformations. In Southeastern Europe, these changes were accompanied by increased migration into the empire. For Viminacium, these developments had devastating consequences, with a reduction of the city size after relocation of the military garrison, and culminating with the city's destruction at the hands of the Huns in 441. The city was then rebuilt by Emperor Justinian, only to be sacked again by the Avars in 582. To study the history of the city beyond the available historical records, and to investigate whether the aforementioned changes had an effect on its demography, we generated 155 new whole genomes from the Viminacium necropoleis, focusing on the 4th, 5th and 6th centuries. We document a significant shift in the ancestry of Viminacium in the 5th century, marked by a decline in genetic diversity with respect to the previous period and a further shift in the 6th, potentially reflecting the mutated policies of the Late Roman Empire or the effect destructions and restoration of the city.

[B218] Optimising imputation and IBD segments retrieval in ancient genomes

Linda Ongaro[1], Dr. Emily M Breslin[1], Dr. Marco Rosario Capodiferro[1], Emilia Huerta-Sánchez[1,2], Dr. Lara M Cassidy[1]
[1]Trinity College Dublin, Dublin, Ireland, [2]Ecology and Evolutionary Biology and Center for Computational and Molecular Biology, Brown University, Providence, United States of America
Ancient DNA has revolutionised the study of genetic variation and population movements through time. However, significant gaps in ancient genomic data, together with uneven geographic and temporal sampling, limit fine-scale characterisation of genomic structure and relatedness among individuals and archaeological sites.
Here, we evaluate a two-step imputation pipeline for low-coverage ancient genomes. First, genotype imputation is performed on individual samples using GLIMPSE with the 1000 Genomes Project reference panel. Second, we construct a multi-individual dataset of high-confidence genotype calls from GLIMPSE and use it as input for Beagle5, enabling an additional round of imputation and phasing to improve genotype accuracy and reduce missing data. To benchmark the pipeline, we downsampled high-coverage ancient genomes, including both shotgun sequences and 1240k SNP capture data. We systematically assessed each step of the pipeline under different filtering thresholds, including genotype posterior probability following imputation. Because heterozygous sites are particularly challenging to impute in ancient DNA, we prioritised heterozygote sensitivity as our primary metric of imputation performance, alongside overall missingness.
Finally, we assess the downstream impact of the second imputation by identifying Identity-by-Descent (IBD) segments using refinedIBD, including long shared fragments, and validating performance in known parent–offspring pairs.
Overall, this study demonstrates how multi-step imputation strategies can enhance ancient genomic analyses, enabling more accurate inference of population structure, relatedness, and evolutionary dynamics.

[B219] From population branch statistics to human traits: phenotypic insights into recent human adaptation

Manuel Ferrando-Bernal[1]
[1]Ucsf, San Francisco, United States

Understanding how recent natural selection has shaped human phenotypic diversity remains a central challenge in evolutionary genomics. While population-based statistics can robustly detect lineage-specific selection, linking these signals to biological traits and their timing is less straightforward.

Here, we integrate a normalized Population Branch Statistic (PBSn) with phenotype enrichment analyses based on the Human Phenotype Ontology (HPO) to investigate the phenotypic targets of recent human adaptation. We identify genomic regions showing population-specific evidence of selection and map these signals to genes and associated human phenotypes. By combining these results with information on the age of selected variants, we explore how different classes of traits have been preferentially targeted across distinct evolutionary time periods during the last ~50,000 years.

This framework enables the systematic interpretation of population-specific selection signals in terms of human phenotypes and provides temporal context for adaptive events. Our results highlight how integrating selection scans with phenotype ontologies can reveal biologically meaningful patterns of recent human evolution and generate testable hypotheses about the environmental and functional drivers of adaptation

[B220] Archaic introgression in pre-European contact Americas

Martina Gulì[1], Linda Ongaro[1], Dr Léo Planche[2], Dr Maria C. Ávila-Arcos[3], Flora Jay[2], Emilia Huerta-Sánchez[1,4], Marco Rosario Capodiferro[1]
[1]Smurfit Institute of Genetics, Trinity College Dublin, Dublin, D02 CX56 Dublin 2, Ireland, [2]Interdisciplinary Laboratory of Digital Sciences, Université Paris-Saclay, Orsay, 91400, France, [3]International Laboratory for Human Genome Research, Universidad Nacional Autónoma de México, Querétaro, Mexico, [4]Ecology and Evolutionary Biology and Center for Computational and Molecular Biology, Brown University, Providence,, USA

When the North American ice sheets melted around 17 ka, small groups of humans emerging from Beringia dispersed rapidly across the Americas, reaching South America in a remarkably short time. These founding populations, adapted to the extreme Arctic environment for thousands of years, were suddenly exposed to a wide range of novel environmental conditions. How they adapted to these new environments in such a short time remains unclear.

One potential source of genetic variation that may have facilitated their rapid adaptation is archaic introgression, the genomic regions inherited from Neanderthals and Denisovans during earlier admixture events outside Africa. Understanding how much archaic ancestry was carried into the Americas, and whether these regions contributed to adaptation during the peopling of the continents, remains an open question.

To address these questions, we reconstructed archaic introgression patterns by directly analysing ancient genomes belonging to pre-European-contact individuals. We built a dataset of available shotgun genomes and imputed their genotypes. After assessing imputation accuracy on high-coverage ancient genomes, we inferred global and local archaic ancestry across time and space in both North and South America. Our results reveal patterns of archaic ancestry in the Americas before European colonisation and lay the groundwork for future investigations.

[B221] Tracking the demography in the Ryukyu Archipelago using time-series ancient genomes

Masatoshi Matsunami[1], Yosuke Kawai[2], Leo Speidel[3], Tsuneo Kakuda[4], Mai Takigami[5], Noboru Adachi[4], Yasushi Tamashiro[6], Chiaki Katagiri[7], Masami Takenaka[8], Minako Imamura[1], Ryosuke Kimura[1], Ken-ichi Shinoda[5], Hideaki Kanzawa-Kiriyama[5]
[1]University of the Ryukyus, Ginowan, Japan, [2]National Center for Global Health and Medicine, , Japan, [3]RIKEN, Wako, Japan, [4]University of Yamanashi, Chuo, Japan, [5]National Museum of Nature and Science, Tsukuba, Japan, [6]Nakijin Village History and Culture Center, Nakijin, Japan, [7]Okinawa Prefectural Archaeological Center, Nishihara, Japan, [8]Kagoshima Women's College, , Japan

The Ryukyu Archipelago is southernmost islands of Japan. From this region, human bones at the Paleolithic Period were excavated. However, there are weak genetic continuity between Paleolithic and modern Ryukyu people, and migrations in the Neolithic Period or later are important to understand to the origin of modern Ryukyu population. In Japan, indigenous Jomon hunter-gatherers lived during the prehistoric period. Although Jomon culture began 16,000 before present (BP) in Japanese mainland (Hondo), archeological evidence suggests that the start of Ryukyu Jomon culture was at around 8,000 BP. We previously sequenced 25 ancient genomes derived from the Ryukyu Jomon period (6,700–900 BP), and found genetic differentiation between Hondo and Ryukyu Jomon is dating back to 7,500 BP. After divergence from Hondo Jomon, Ryukyu Jomon experienced severe genetic bottlenecks. Although both Hondo and Ryukyu Jomon populations eventually received admixture from continental East Asia, we observe a 600-year time lag. These different demographic histories of Japanese Hondo and Ryukyu population cause different rates of Jomon ancestry of modern Japanese. The timing of this admixture in Ryukyu corresponds to the start of the Ryukyu medieval period (i.e. Gusuku period; 900-500 BP). However, it is still unknown that how these admixtures contribute to the modern Ryukyu population. In this study, we newly sequenced 14 ancient genomes derived from the Gusuku period. These Gusuku population received more genetic influence from Hondo population than Ryukyu Jomon. Our study highlights the detailed demography of the Ryukyu population.

[B222] Unlocking secrets of the epigenome: towards high-resolution bisulfite sequencing of ancient DNA (aDNA)

Natassja Brien[1], Alex Denning[3], Dr. Katherine van Schaik[2,6,7], Dr. Hendrik Poinar[4,5,1]
[1]Department of Biology, McMaster University, Hamilton, Canada, [2]Radiology and Radiological Sciences, Musculoskeletal Imaging, Vanderbilt University, Nashville, USA, [3]University of Pennsylvania, Philadelphia, USA, [4]Department of Anthropology, McMaster University, Hamilton, Canada, [5]Department of Biochemistry, McMaster University, Hamilton, Canada, [6]Classical and Mediterranean Studies, Vanderbilt University, Nashville, USA, [7]Electrical and Computer Engineering, Vanderbilt University, Nashville, USA

In the quest to understand our shared human past more deeply, researchers have sought to expand the informative power of ancient DNA, while working within the taphonomic limits of ancient samples. One such development is the study of the epigenome which has generated powerful insights, such as methylation levels from a Neanderthal individual and estimation of methylation maps of ancient human populations. Detailing that information across past populations who have lived through infectious disease, climate upheaval, and famine gives us new insights in what it means to be human. In time, methylation studies could lead to precise age estimation and refinement of gene expression studies in functional genomics. However, methodological rigour is needed to study DNA methylation at single base pair resolution in aDNA datasets. A few studies have attempted to apply modern bisulfite conversion methods to ancient DNA, but none have directly compared different conversion methods until recently. Here we provide a side-by-side comparison of multiple bisulfite conversion workflows to determine a) the most effective conversion of unmethylated cytosines for ancient DNA while balancing b) the least damaging treatment which provides the most informative read data, via loci-specific target capture enrichment. Although sodium bisulfite conversion is common in modern workflows, this study shows that an ammonium bisulfite conversion is more likely to produce accurate data without sacrificing overall DNA recovery from important, ancient samples. A rigorously validated bisulfite conversion method would enable aDNA researchers to begin to conduct single-loci methylation analyses and expand the possibilities of ancient epigenomics.

[B223] Genomic insights into population connectivity among Classic Maya centres

Niall Moore[1], Dr. Seiichi Nakamura[2], Grace Johnston[1], Dr. Alexander Urrizar[3], Dr. Nobuyuki Ito[4], Dr. Masahiro Ogawa[2], Prof. Hie Lim Kim[5], Dr. Takashi Gakuhari[6], Dr. Shigeki Nakagome[1,6]
[1]Trinity College Dublin, , Ireland, [2]Komatsu University, , Japan, [3]Universidad de San Carlos de Guatemala, , Guatemala, [4]Nagoya University, Nagoya, Japan, [5]Nanyang Technological University, , Singapore, [6]Kanazawa University, , Japan

The Classic Maya was among the most complex societies of pre-Columbian America; however, key questions remain regarding its formation and internal population structure. Although long-distance interaction is widely considered a hallmark of Classic Maya sociopolitical networks, the extent of population mobility among major centres has yet to be clearly defined. Here, we present whole-genome sequences from nine individuals recovered at Tikal, an influential Classic Maya capital, and co-analyse these data with previously reported genomes from two other Maya centres, Copán and Chichén Itzá, alongside other ancient and present-day American populations. Our demographic analyses indicate that the Classic Maya populations form a relatively tight genetic cluster within a broader continental context, while also revealing signals consistent with differential genetic connectivity among these centres. In particular, we observe little to no identity-by-descent sharing between individuals from Tikal and Copán, based on a limited sample size, as well as greater diversity of links to populations outside the Maya region in Chichén Itzá. These results illustrate how ancient genomics can provide new insights into patterns of interaction and mobility within and beyond Classic Maya centres.

[B224] Regulatory remodeling and phenotypic relevance of lineage-specific sites across human lineages

Qiong Zhu[1], Yaoxi He[2], Yini Xiao[1], Zonglin Cai[1], Kai Liu[2], Bing Su[2], Lu Chen[1]
[1]State Key Laboratory of Genetic Engineering, Center for Evolutionary Biology, School of Life Science, Fudan University, Shanghai, China, [2]State Key Laboratory of Genetic Resources and Evolution, National Key Laboratory of Genetic Evolution and Animal Model, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming, China

The functional characterization of lineage-specific sites (LSS) has mainly focused on selected coding changes, leaving their regulatory roles and phenotypic relevance insufficiently understood. Here, using six high-coverage archaic genomes and ALFA modern human allele-frequency data, we constructed five LSS catalogs for modern-human-, Neanderthal-, Denisovan-, archaic-human-, and hominin-specific sites. Across the five catalogs, all LSS sets were enriched in enhancer annotations, and genes linked to these sites showed enrichment for olfactory receptor-related functions. Regulatory analyses further showed that transcription factor (TF) genes associated with LSS were more highly expressed across tissues than non-LSS TF genes, and modern human-specific TF-gene-related LSS were enriched for eQTLs in multiple brain regions and the uterus. TFBS-related LSS were enriched in open chromatin and showed higher predicted functional impact, supporting their potential regulatory effects. Among high-impact motif-disrupting sites, LSS preferentially affected specific TF motif modules rather than being evenly distributed across binding motifs. These motif-related TF-target genes were enriched for developmental and tissue-remodeling processes, hormone and metabolic responses, inflammatory signaling, apoptosis, neural/glial development, and tissue homeostasis. HPO enrichment further highlighted endocrine, skeletal, neuroendocrine, reproductive, and eye-development phenotypes, supporting the phenotypic relevance of LSS-associated regulatory changes. Together, these results suggest that LSS may have contributed to human lineage differentiation by reshaping transcription factor binding landscapes and downstream regulatory networks.

[B225] Using ancient DNA methylation to Predict Exposure to Smoke in Ancient Societies

Raphael Sirat[1,2], Prof. Liran Carmel[1], Benjamin Yakir[2], Professor Eran Meshorer[1,3]
[1]Department of Genetics, The Alexander Silberman Institute of Life Sciences, The Hebrew University of Jerusalem, Jerusalem, Israel, [2]Department of Statistics and Data Science, the Hebrew University of Jerusalem, Jerusalem, Israel, [3]Edmond and Lily Safra Center for Brain Sciences (ELSC), the Hebrew University of Jerusalem, Jerusalem, Israel

DNA methylation in blood of present-day individuals has been shown to provide reliable prediction of exposure to smoke. Using this predictive model for ancient societies might provide insights on their patterns of fire use. However, reconstructed levels of DNA methylation in ancient genomes is characterized by increased standard errors and lower resolution compared to modern methods. Moreover, modern predictive models were developed for blood, whereas ancient DNA methylation maps typically represent skeletal tissues.

To overcome these limitations, we developed a multi-step framework. First, we used DNA methylation data across multiple tissues to develop a predictive pan-tissue model. Second, we adapted the predictive model to the distinctive features of ancient DNA methylation. We validated the framework using simulations.

We applied the model to 53 ancient genomes and predicted levels of smoke exposure. Together with temporal and spatial information, as well as cultural context, we identified patterns of smoke exposure changes across time, space and culture.

[B226] The Royal Warship Kronan: Revealing the Crew through Archaeogenomics

Reyhan Yaka[1,2], Prof Anders Götherström[1,2]
[1]Centre for Palaeogenetics, Stockholm University, Stockholm, Sweden, [2]Department of Archaeology and Classical Studies, Stockholm University, Stockholm, Sweden
The Swedish Royal Ship Kronan, one of the largest warships of its time, tragically exploded and sank near Öland Island in the Baltic Sea in 1676, claiming the lives of over 800 crew members. Designed for both warfare and the projection of royal power, Kronan carried a crew of sailors and soldiers, including mercenaries bound for the continent. The Kronan crew thus provides a unique snapshot of 17th-century Sweden. Archaeological excavations have recovered extensive human remains from the wreck. Osteological analyses further identified several individuals as children, raising questions about recruitment practices, military regulations, and the possible roles of children aboard the ship, including potential familial connections to adult crew members.

Building upon these archaeological findings, we conducted archaeogenomic analyses of the recovered human remains. We generated ancient genomes from 61 individuals (53 adults and 8 children) and applied state-of-the-art methods, including Identity-by-Descent (IBD) analysis. We investigated the demographic composition of the crew, identified kinship patterns, and assessed genetic affinities relative to present-day populations from Sweden and Europe. Our results reveal that the crew comprised individuals from diverse regions across Sweden and neighbouring areas, including broader Scandinavia. Together with the identified kinship ties among crew members, these findings shed light on recruitment strategies and the social dynamics of the 17th-century Swedish navy. This study complements existing archaeological research and provides valuable genomic data for future interdisciplinary work in archaeogenomics and archaeology.

[B227] Reconstructing the genetic landscape and evolutionary history of Pakistan using ancient and modern genomes

Rodelmar Ocampo Luna[1], Luis Enrique Ramirez Serrano[1], MSc Maria Jose Palma Martinez[1], Undergraduate student Iqui Balam Perez Martinez[1], Zara Paulina Martinez Sanchez[1], Dra. Miriam Bravo-López[4,5], Dr Maria C. Ávila-Arcos[2], Archeologist Asma Ibrahim[3], Dr Cesar Fortes-Lima[6,7], David Reich[8], Mashaal Sohail[1]

[1]Centro de Ciencias Genómicas, Universidad Nacional Autonoma de Mexico, Cuernavaca, Mexico, [2]Laboratorio Internacional del Genoma Humano, Universidad Nacional Autonoma de Mexico, Juriquilla, Mexico, [3]State Bank of Pakistan Museum, Archives & Art Gallery department, Karachi, Pakistan, [4]School of Human Evolution and Social Change, Arizona State University,, Tempe, USA, [5]Institute of Human Origins, Arizona State University, Center for Evolution and Medicine, Arizona State University,, Tempe, USA, [6]Department of Genetic Medicine and McKusick-Nathans Institute, Johns Hopkins University School of Medicine, Baltimore, USA, [7]Human Evolution Program, Department of Organismal Biology, Evolutionary Biology Centre, Uppsala University,, Uppsala, Sweden, [8]Harvard Medical School, Boston, USA

We characterize the genetic ancestral landscape and evolutionary history of Pakistan over ~3,700 years by integrating whole genome sequencing data from nine present-day cultural groups (n=279, joint 1000 Genomes–HGDP dataset) with genome-wide data from 152 ancient Pakistani individuals including 20 unpublished genomes. Comparisons between autosomal, mitochondrial and Y-chromosome variation reveal distinct structure patterns, reflecting sex-specific demographic processes.

Despite high levels of consanguinity, Y-chromosome haplogroups are broadly shared across Pakistani groups and globally, consistent with Pakistan's role as a historical crossroads linking the Middle East, Asia, and Europe. Significant sex-biased admixture is detected in Kalash, with greater female contributions from South and Central Asia and male-biased contributions from Middle East.

We observe shared ancestries across time from Iron Age individuals to present-day Burusho, Hazara, Pathan, Punjabi, and Sindhi individuals. In contrast, Brahui, Balochi, Makrani, and Kalash show limited shared ancestries with these ancient individuals, consistent with varying demographic histories. African ancestries are detected in Makrani, Sindhi, and Balochi individuals, concordant with historical accounts of forced migrations.

Local ancestry Tracts analyses of 48 Makranis (HGDP, Laso-Jadart(2017)) identify founding admixture events involving African ancestors between 12 and 14 generations ago, followed by additional migratory pulses from African and Eurasian sources, aligning with historical records from the Swahili coast slave trade by the Omani empire and Europeans into the 20th century. Finally, population branch statistics identify multiple loci under selection, with Kalash showing the highest number of group-specific signals. Across groups, SLC24A5, associated with skin pigmentation, emerges as a prominent candidate.

[B228] Genomic Analysis and Environmental Adaptation in the Prehistoric Western Mediterranean

Samantha Rossini[1], Cecilia Paradiso[1], Paolo Abondio[1], Emma Buckingham[2], Francesca Cortese[3], Lucilla D'Alessandro[4], Fabrice Demeter[5,6], Francesco Fabbri P.[7], Luca Grassigli G.[8], Sarah Harvey[9], Nunzia Libianchi[4], Laura Maniscalco[10], E. McConnell B.[10], Bernardette Mercieca-Spiteri[11], Fernando Molina[12], Fabrizio Nicoletti[13], Francesco Pacelli[14], Victoria Peña[15], Mauro Rubini[16], Cynthianne Spiteri[11,17], Stefano Spiganti[14], Maria Turco[13], Giorgia Vincenti[7], Paola Zaio[18,19], Alfredo Mederos Martín[20], Federico Rolfo M.[3], Serena Sabatini[21], Gabriele Scorrano[1]
[1]Department of Biology, University of Tor Vergata, Rome, , [2]University of Missouri, Columbia, , [3]Department of History, Cultural Heritage, Education, and Society University of Rome Tor Vergata, Rome, , [4]Istituto autonomo Villa Adriana e Villa d'Este, Rome, , [5]Laboratoire d'Eco-Anthropologie UMR7206, CNRS, MNHN, Université de Paris, Paris, , [6]Geogenetics Centre, Globe Institute, University of Copenhagen, Copenhagen, , [7]Museo e Istituto Fiorentino di Preistoria, Firenze, , [8]University of Perugia - Department of Humanities - Ancient and Modern Languages, Literatures, and Civilizations, Perugia, , [9]Department of Modern and Classical Language Studies, Kent State University , Ohio, , [10]Florida Atlantic University, Boca Raton, , [11]Superintendence Of Cultural Heritage, Valletta, , [12]University of Granada, Department of Prehistory and Archaeology, Granada, , [13]Superintendency for Cultural and Environmental Heritage of Catania, Catania, , [14]Archeologo, Associazione Acqua, Orvieto, , [15]Universidad Complutense, Centro de Estudios Fenicios y Púnicos, Madrid, , [16]University of Foggia, Ministry of Cultural Heritage, Environment, and Tourism, Foggia, , [17]Department of Life Sciences and Systems Biology, University of Turin, Turin, , [18]Superintendence of Archeology, Fine Arts and Landscape for the Provinces of Frosinone and Latina, Rome, , [19]Istituto Italiano di Paleontologia Umana, Piazza Ruggero Bonghi 2, 03012 , Anagni, , [20]Department of Prehistory and Archaeology, Faculty of Philosophy and Letters, Autonomous University of Madrid. Cantoblanco Campus, Madrid, , [21]Department of Historical Studies, University of Gothenburg, Gothenburg,

The transition from the Neolithic to the Bronze Age (ca. 8000–3000 BP) marks one of the most profound transformations in European prehistory. In central and northern Europe, this period is associated with large-scale population movements, substantial genetic turnover, and major social reorganisation. In contrast, archaeological evidence from the western Mediterranean: Italy, Iberia, southern France, and Malta, suggests markedly different trajectories, characterised by cultural transmission, heterogeneous mobility patterns, and regionally specific responses to climatic stress, particularly during the 4.2 ka event.

This project aims to genetically characterize populations from the western Mediterranean spanning 5,000 years from the Neolithic to the Metal Ages, with a focus on the Chalcolithic, and to test whether the population decline observed around 4.2 ka reflect large-scale migration or social restructuring within largely continuous populations under climate stress.

The project will analyse over 400 skeletal samples from Spain, France, Italy, and Malta. Ancient DNA will be extracted from teeth and petrous bones, followed by purification, double-stranded library preparation, and sequencing on an Illumina platform.

Preliminary analyses yielded usable data for most of the individuals analysed, with endogenous DNA content ranging from 6% to 73%, and autosomal coverage > 0.1X. Deamination rates of 27–42% confirmed the ancient origin of the DNA, and mitochondrial contamination estimates are below 5%. In conclusion, initial analyses show good DNA preservation despite a western Mediterranean temperate climate. Further analyses will enable a comprehensive genetic characterization of the sampled populations.

[B229] Genomic Echoes of the Past: The Evolutionary History of Sicily

Rajiv Boscolo Agostini[1], Margherita Vanni[2], Maria Teresa Vizzari[1], Federico De Pizzol[1], Patricia Santos[1], Alessandra Modi[2], Giulio Catalano[3], Chiara Barbieri[4], Stefania Sarno[5], Carla Calò[4], David Caramelli[2], Paolo Francalacci[4], Marco Sazzini[5], Luca Sineo[3], Stefania Vai[2], Silvia Ghirotto[1]
[1]Department of Life Sciences and Biotechnology, University of Ferrara, Ferrara, Italy, [2]Department of Biology, University of Florence, Florence, Italy, [3]Department of Biological, Chemical and Pharmaceutical Sciences and Technologies, University of Palermo, Palermo, Italy, [4]Department of Life and Environmental Sciences, University of Cagliari, Cagliari, Italy, [5]Department of Biological, Geological, and Environmental Sciences, University of Bologna, Bologna, Italy

Sicily is located at the center of the Mediterranean Sea and has long served as a crossroads for population movements and cultural interactions since its first colonization approximately 20,000 years ago, playing a crucial role in the peopling of Europe. Despite a rich archaeological record and the growing availability of ancient genomic data, key evolutionary and demographic processes remain poorly understood, thus preventing a comprehensive reconstruction of human population dynamics across the whole Mediterranean basin. Here, we present 18 newly generated ancient genomes (average coverage ranging from 0,1 to 4) from 11 Sicilian necropolises, spanning from the Paleolithic (~14,000 BCE) to the Middle Ages. These data, combined with 23 previously published ancient Sicilian genomes and approximately 500 ancient individuals from the Mediterranean basin and Europe, provide high-resolution insights into demographic dynamics, ancestry patterns, and genetic contributions within the island. We detect evidence of Steppe pastoralist ancestry from approximately 2,200 BCE, along with elevated proportions of Iranian-related ancestry compared to the Italian mainland, consisted with increased connectivity with the Aegean, including groups such as the Minoans and Mycenaeans. From the Iron Age onward, a North African genetic component emerges, reaching its maximum during the Middle Ages. We also identified genealogical affinities among necropolises and local patterns of genetic continuity through time. Overall, our analyses provide direct genomic evidence of the long-term genetic consequences of cultural interactions that shaped Sicily’s complex genetic heritage over the past fourteen millennia.

[B230] Reframing the Neolithic Y-chromosome bottleneck in Europeans under purifying selection and structure

Stefan Strütt[1,2], Dr. Adamandia Kapopoulou[1,3,4], Dr. Sandra Oliveira[1,5], Dr. Stephan Peischl[1,2], Prof. Dr. Laurent Excoffier[1,3]

[1]Computational and Molecular Population Genetics Lab, Institute of Ecology and Evolution, University of Bern, Baltzerstrasse 6, 3012 Bern, Switzerland, Bern, Switzerland, [2]Interfaculty Bioinformatics Unit, University of Bern, Bern 2012, Switzerland, Bern, Switzerland, [3]Swiss Institute of Bioinformatics, Lausanne 1015, Switzerland, Bern, Switzerland, [4]Theoretical Ecology and Evolution, University of Bern, Bern 2012, Switzerland, Bern, Switzerland, [5]Human Genetic and Cultural Evolution, Department of Evolutionary Biology and Environmental Studies, University of Zurich, Switzerland, Zürich, Switzerland

A pronounced asymmetry between mitochondrial and Y-chromosome diversity characterizes human genetic variation. Intriguingly, Y-chromosomal data indicate a significant reduction in diversity approximately 4,000–8,000 years ago, coinciding with the onset of farming. This pattern has often been attributed to cultural shifts toward patrilocality, exogamy, and increased variance in male reproductive success. However, because uniparental loci lack recombination, they are sensitive to purifying selection and Hill–Robertson interference, processes that can mimic signatures of a demographic contraction. Here, we re-evaluate the cultural interpretation of reduced male-specific diversity by analyzing ancient and modern European genomes using theoretical models that explicitly incorporate the confounding effects of weak purifying selection. Accounting for these effects, we find no evidence for a pronounced male-specific Neolithic bottleneck. Instead, the disparity between mitochondrial and Y-chromosomal diversity may reflect genomic architecture rather than sex-biased demography. In conclusion, our findings caution against treating non-recombining regions as neutral loci and highlight the need to integrate selection into demographic inference when recombination is absent.

[B231] A new method for inferring demographic history and structure from the distribution of heterozygous sites

Tommaso Stentella[1], Paul Etheimer[2,3], Florian Massip[2,3,4], Michael Sheinman[5], Peter Arndt[1]
[1]Max Planck Institute For Molecular Genetics, Berlin, Germany, [2]CBIO Mines ParisTech, Paris, France, [3]Institute Curie, Paris, France, [4]INSERM, Paris, France, [5]Weizmann Institute of Science, Rehovot, Israel

In the last two decades, several methods to infer the demographic history of a population from whole genome sequence data have been developed. Despite these efforts, current methods remain computationally demanding, limiting researchers' ability to efficiently explore parameter space and to estimate confidence intervals of model parameters. As a consequence, it is difficult to quantitatively study natural populations, where the true history is unknown, and to decide whether inferred demographic events are accurate. This is especially true if data are limited, for instance when only one genome is available.

Here, we consider the distances between heterozygous sites in single diploid samples, deriving novel and simple analytical results for their distribution under varying population histories. These theoretical expectations allow us to efficiently fit and compare a large number of parametric models of varying complexity. The most probable model output by our method automatically partitions the demographic history into an optimal number of epochs having arbitrary duration, with parameters and confidence intervals estimated jointly. Moreover, we provide a distribution of histories that are compatible with the data.

We show that our method can quickly infer thousands of unique demographic histories, for instance from each of the single genomes in the 1000 Genomes Project. Such analyses reveal fine-scale variation at the individual level within and across populations, additionally allowing us to explore population structure through time. Our new method and analysis enhance our understanding of the extent to which demographic history can be reconstructed from whole genome sequence data in any species.

[B232] Archaeogenomic Evidence for Social Organization and Connectivity in Teotihuacan

Xavier Roca-Rada[1,2,3,13], María Fernanda Elías Navarro[4,13], Linda R. Manzanilla-Naim[5,13], Evelyn Collen[6], Kalina Kassadjikova[7], Andrés de Jesús López-Tamayo[4], Daniela Robles Orozco[4], Yassine Souilmi[3], Leonard Taufik[3,8], João C. Teixeira[3,9,10], Lars Fehren-Schmitz[7], Emilia Huerta-Sánchez[1,2,11,14], Bastien Llamas[3,14], Rafael Montiel[12,14], Dr. Andres Moreno-Estrada[4,14]
[1]Department of Ecology, Evolution, and Organismal Biology, Brown University, Providence, USA, [2]Center for Computational Molecular Biology, Brown University, Providence, USA, [3]Australian Centre for Ancient DNA, School of Biological Sciences, Adelaide University, Adelaide, Australia, [4]Aging Research Center, Cinvestav Sede Sur, Mexico City, Mexico, [5]Instituto de Investigaciones Antropológicas, Universidad Nacional Autónoma de México, Mexico City, Mexico, [6]Technology Advancement Unit, Genetics and Molecular Pathology, SA Pathology, Adelaide, Australia, [7]UCSC Paleogenomics, University of California Santa Cruz, Santa Cruz, USA, [8]Mochtar Riady Institute for Nanotechnology, Tangerang, Indonesia, [9]CIBIO, Centro de Investigação em Biodiversidade e Recursos Genéticos, InBIO Laboratório Associado, Campus de Vairão, Universidade do Porto, Porto, Portugal, [10]Evolution of Cultural Diversity Initiative, School of Culture, History and Language, The Australian National University, Canberra, Australia, [11]Smurfit Institute of Genetics, Trinity College Dublin, Dublin, Ireland, [12]Unitat d'Antropologia Biològica, Departament de Biologia Animal, de Biologia Vegetal i d'Ecologia, Universitat Autònoma de Barcelona, Bellaterra, Barcelona, Spain, [13]These authors contributed equally. , , , [14]These authors jointly supervised this work., ,

Teotihuacan, located in the Basin of Mexico, was the largest city of the Classic period (~100–650 CE) in Mesoamerica, a historical region spanning present-day central Mexico to western Honduras. As a center of political, economic, and cultural influence, it played a crucial role in pre-Hispanic American history. However, the absence of human archaeogenomic data from the city has precluded insight into genetic aspects of social organization, demography, and interregional connectivity.
We generated genome-wide data from two contrasting contexts within Teotihuacan: Teopancazco and the Palace of Xalla, the former a multi-ethnic neighborhood associated with specialized craft production and the latter an elite political and administrative hub. We analyzed 10 burials from Teopancazco and 49 bone fragments scattered through ritual practices in Xalla, recovering 13 individual genome-wide datasets.
In Teopancazco, individuals were unrelated and exhibited high heterozygosity, consistent with a diverse, non-kin-based community. Conversely, Xalla showed close biological relationships, including multiple bone fragments from the same individual and distant relatives, consistent with a male-oriented social organization linked to gendered ritual practices. We also inferred large effective population sizes, consistent with Teotihuacan's scale and agricultural lifestyle.
Genetic structure broadly reflected geography, placing most Teotihuacan individuals within central Mesoamerican variation, while revealing a single Mayan outlier from southern Mesoamerica. Additional signals indicated interactions with the Maya region and weaker affinities with Aridoamerica to the north.
Our study reveals contrasting social structures and large-scale connectivity in Teotihuacan, underscoring the value of integrating genetic, archaeological, and historical evidence to reconstruct the evolutionary past of pre-Hispanic civilizations.

[B233] Understanding brain evolution and psychiatric disease risk through the lens of Neanderthal divergence

Yaen Chen[1], Dr. Tony Capra[1]
[1]University of California, San Francisco (UCSF), San Francisco, United States
Many neuropsychiatric disorders are thought to be human-specific, as cortical morphology and white-matter tracts involved in higher cognitive ability underlie many psychiatric disorders. How this evolutionary trade-off for increased brain complexity may influence susceptibility to psychiatric disorders is unclear.

Here, we explore structural brain evolution and associated psychiatric risk in modern humans through comparison to Neanderthal genomes. Modern humans and Neanderthals exchanged genetic information ~47,000 years ago, and due to this introgression, DNA of Neanderthal ancestry is segregating in more than half of the human genome. However, the modern human genome contains many regions lacking Neanderthal introgression, or “Neanderthal-free loci,” that likely reflect selection against regions with functional divergence.

To investigate evidence for functional divergence between humans and Neanderthals in specific brain regions, we estimated heritability for 259 cortical and subcortical neuroimaging phenotypes encoded in Neanderthal-free regions using Gene-ENviroment Interaction Estimator (GENIE) across 42,886 individuals in the UK Biobank. We observed significant enrichment in the volume of the right hippocampus (h2 enrichment=1.72, p=0.00712). We also observed substantial depletion of heritability in Neanderthal-free loci for amygdalar and thalamic subregions (h2 enrichment=0.20-0.27).

For the same Neanderthal-free loci, we observed significant heritability enrichment using Linkage Disequilibrium (LD) Score Regression across six psychiatric phenotypes: schizophrenia, attention-deficit hyperactivity disorder, major depressive disorder, cigarette and alcohol use, and anorexia nervosa (h2 enrichment=1.7-2.3). This suggests that genomic regions without evidence of Neanderthal introgression are enriched for psychiatric risk. Overall, this work expands our knowledge of modern human brain evolution and elucidates links between neurobiology and psychiatric diseases.

[B234] Studying archaic human structural variation with high-coverage genomes

Yaniv Swiel[1], Diyendo Massilani[2], Hannes Tausend[1], Alexander Hübner[1], Stéphane Peyrégne[1], Janet Kelso[1]
[1]Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany, [2]Yale School of Medicine, New Haven, USA

Structural variants (SVs), defined as genomic alterations greater than 50 base pairs, affect substantially more of the genome than smaller variants and contribute significantly to human disease and adaptation. However, the study of SVs in archaic hominins has been limited by the characteristics of ancient DNA (short read lengths, DNA damage) and because SVs tend to occur in repetitive regions of the genome which are inaccessible to mapping-based approaches.

While de novo assembly-based approaches offer potential insights into these repetitive regions, the fragmented and damaged nature of ancient DNA poses significant challenges and has largely prevented their application. Using high-coverage archaic human genomes, we show that de novo assembly can reliably identify novel SVs that cannot be detected by mapping-based approaches. We then characterise the SV landscape in archaic hominins and highlight SVs that have contributed to hominin evolution.

[B236] Tracing the evolutionary history of Saccharimonadia (TM7) bacteria through paleogenomics

I-Ting Huang[1], Irina Velsko[2,3], Alexander Hübner[2,3], Batbileg Bor[4], Colleen Cavanaugh[5], Christina Warinner[1,2,3]

[1]Department of Anthropology, Harvard University, Cambridge, United States, [2]Department of Archaeogenetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany, [3]Associated Research Group of Archaeogenetics, Leibniz Institute for Natural Product Research and Infection Biology – Hans Knöll Institute, Jena, Germany, [4]Department of Microbiology, ADA Forsyth Institute, Somerville, United States, [5]Department of Organismic and Evolutionary Biology, Harvard University, Cambridge, United States

Saccharimonadia, members of the enigmatic Candidate Phyla Radiation (CPR) bacteria, are core members of the human oral microbiome and have been implicated in disease, but are difficult to cultivate, leaving their species diversity, phylogeny, and evolutionary history largely unresolved. We analyzed over four thousand Saccharimonadia genomes reconstructed from environmental, modern host-associated and archaeological samples, including present-day and ancient humans up to 30,000 years old and Neanderthals up to 100,000 years old. Phylogenetic analyses suggest the presence of 78 well-defined species clusters and revealed a revised evolutionary history with at least seven major habitat transitions from the environment into mammalian microbiota, followed by subsequent dental biofilm adaptation and oral or gut niche specialization. We identified three new well-supported species found among ancient humans and present-day foragers and subsistence horticulturalists but not among industrialized populations, suggesting a potential recent loss of these clades due to lifestyle changes. Metagenomic and pangenomic analyses revealed features of genomic plasticity underlying habitat transitions, including lineage-specific acquisition of prophages and phage defense systems, and patterns of GC-content variation, with important implications for models of bacterial evolution and host-microbial interactions. By tracing the evolutionary trajectory of Saccharimonadia through both ancient and modern samples, this study represents one of the most comprehensive temporal evolutionary datasets for any CPR lineage, providing empirical insight into the evolution of small genomes and host-microbe interactions across deep time.

[B237] The evolution of gene regulatory networks during the transition from unicellular to multicellular life

Jan Kreider[1], Tobias Uller[1], Professor Charlie Kinahan Cornwallis[1]
[1]Lund University, , Sweden

The evolution of multicellularity has fueled the complexity and diversity of life on Earth. While multicellularity has evolved repeatedly in some clades, such as green algae, other clades consist exclusively of unicellular organisms. Why has multicellularity evolved so many times in some lineages, but not others? An untested idea is that multicellularity readily evolves from unicellular organisms that prolong their replicative (multicellular) life-stages under specific environmental conditions ("plastic multicellularity"). Here, we investigate if plasticity in multicellularity has preceded the evolution of obligately multicellular life using a theoretical model of evolving gene regulatory networks. If a gene regulatory network can plastically produce multicellular groups in response to environmental change, then this should bias the effect of genetic mutation to produce similar multicellular phenotypes more often than expected by chance ("developmental bias"). Consequently, transitions from plastic multicellularity to obligate multicellularity should be facilitated. To test this, we compare how likely obligate multicellularity evolves from gene regulatory networks that have been selected for plastic multicellularity vs. networks that have been selected for obligate unicellularity. We also derive predictions for how regulatory interactions between genes change during the evolutionary transition to multicellularity. In the future, we intend to test these predictions using gene expression data across multiple origins of obligate multicellularity in green algae.

[B238] Unicellular and Multicellular Selection Impose Fundamentally Distinct Constraints on Cellular Phenotype Evolution

Mark Kim[1], Matt Pennell[1]
[1]Cornell University, Ithaca, United States

Single-cell sequencing data from various organisms has revealed that cellular phenotypes (e.g., gene expression) are often remarkably low-dimensional, suggesting the existence of semi-discrete gene expression programs. A number of researchers have remarked that a distinction between unicellular and multicellular life is that unicellular organisms that can display multiple gene-expression programs must do so at different life stages or in different environments (i.e., they are sequentially realized) whereas multicellular organisms can display multiple gene-expression programs at the same time (i.e., they are concurrently realized). However, the consequences of this distinction for the evolutionary dynamics of cellular phenotypes are poorly understood.

Here, we address this question using a geometric model of cellular phenotype evolution. We simulated evolutionary dynamics under alternative regimes in which multiple task demands are realized either sequentially or concurrently, directly analyzing how temporal realization structure alone shapes evolutionary constraints.

We find that when task demands are weakly divergent, sequential and concurrent realization lead to similar evolutionary outcomes. As task divergence increases, however, the results diverge sharply. Sequential realization produces broad dispersion of evolved phenotypes, including specialization toward individual tasks, but is associated with reduced performance on both tasks and a lower capacity to express distinct phenotypes for distinct tasks. Concurrent realization, in contrast, maintains higher performance on both tasks and a greater capacity to express distinct phenotypes for distinct tasks. These results show that the distinction between unicellular and multicellular modes of phenotype realization can qualitatively reshape evolutionary dynamics and constraints, even when the genotype–phenotype mapping is unchanged.

[B239] Proteins that emerged after the Great Oxygenation Event used less now-scarce manganese and iron-sulfur clusters, but not less heme

Nandini Manepalli[1], Sawsan Wehbi[2], Joanna Masel[3]
[1]University Of Arizona, Department of Molecular and Cellular Biology, Tucson, United States,
[2]University of Arizona, Genetics Graduate Interdisciplinary Program, Tucson, United States,
[3]University of Arizona, Department of Ecology and Evolutionary Biology, Tucson, United States

Many proteins bind transition metals, which catalyze biological processes by facilitating electron exchange reactions. The advent of oxygenic photosynthesis triggered the Great Oxidation Event (GOE), a major transition in which oxygen-depleted, reducing oceans transformed to oxidizing conditions. The GOE made some transition metals (e.g. iron and manganese) less soluble and hence less bioavailable, and other metals (e.g. zinc) more soluble. Here, we examine which transition metals were used more often by ancient, pre-GOE protein domains (annotated as Pfams), compared to Pfams that emerged more recently. We use age classifications derived by Wehbi et al. (2024) from Pfam trees reconciled with species trees. Pfams were assigned to pre-LUCA (having multiple diversified copies by the time of LUCA), LUCA, LACA, LBCA, and supergroup-specific phylostrata. Patterns of transition metal adoption could be driven either by supply (changing abundance with the GOE) or functional demand. We find that younger Pfams are less likely to bind manganese, supporting the supply hypothesis. However, younger Pfams use more iron, despite its scarcity following the GOE, supporting the functional demand hypothesis. Older Pfams use iron more often via iron-sulfur clusters, and less often via heme, consistent with a sulfur-rich ancient world, and with heme's greater resistance to oxidative stress. Preliminary results on zinc show a temporal trend supporting the supply hypothesis, while high zinc usage by even ancient Pfams suggests that pre-GOE zinc might have been more bioavailable than is suggested by geochemical models.

[B240] Are interphylum spiralian relationships resolvable?

Ana Serra Silva[1], Prof Maximilian Telford[1]
[1]University College London, , United Kingdom

The phyla making up the major animal clade of Spiralia have been clear since the advent of molecular phylogenetics; the relationships between these spiralian phyla have not. The lack of consensus over the relationships between these important animal phyla might be a clue implying their emergence in an explosive radiation. Focusing on the five largest spiralian phyla (Annelida, Brachiopoda, Mollusca, Nemertea and Platyhelminthes) and using two phylogenomic datasets, we have applied site-bootstrapping and taxon-jackknifing to explore this example of taxonomic instability. Analyses on the 105 possible rooted trees relating them showed that interphylum branches are very short. Preference for rooting Spiralia on Platyhelminthes is a long-branch artefact. Most analyses on the 15 unrooted trees showed a preference for the same topology but the support over other solutions was non significant. We conclude that the spiralian phyla emerged in rapid succession resulting in a difficult to resolve radiation. The deep history we infer for Spiralia has wide ranging implications for our interpretation of Cambrian fossils and for the evolution of traits such as biomineralization, segmentation and larvae.

[B241] Ancient metagenomics reveals subglacial microbiomes driven by oxygen availability

Bianca De Sanctis[1], Nicholas Dragone[2], Ciara Wanket[3], Cliff Bueno de Mesquita[2], Sarah Crump[3], Gavin Piccione[4], Berkhashni Nirula[3], Halle Bender[3], Russell Corbett-Detig[3], E. Troy Rasbury[5], Graham Edwards[6], Abigale Hawthorn[3], John Welch[1], Alexandra Rouillard[7], Dr. Beth Shapiro[3], Jill Mikucki[8], Terrence Blackburn[3]

[1]University Of Cambridge, , United Kingdom, [2]University of Boulder Colorado, , USA, [3]University of California Santa Cruz, , USA, [4]Brown University, , USA, [5]Stony Brook University, , USA, [6]Trinity University, , USA, [7]Umea University, , Sweden, [8]University of Tennessee, , USA

Beneath Earth's glaciers and ice sheets lies an aquatic realm where ice, water, rock, and microbial life interact, driving chemical reactions that can collectively influence the global carbon cycle, polar oceans, and climate. Efforts to describe subglacial microbiomes have been limited by the challenge of cleanly drilling through hundreds of meters of ice, such that only a few sites have ever been directly sampled. Here we use ancient metagenomics to present the first spatiotemporal characterization of subglacial bacteria and archaea. We extracted DNA from 25 subglacial precipitate samples, sedimentary accumulations of minerals that form in subglacial waters prior to exposure on the surface. The precipitates studied here formed between 16,000 and 570,000 years ago beneath the Antarctic and Laurentide Ice Sheets. We show that postmortem DNA damage patterns can reliably distinguish between ancient subglacial and modern surface taxa, and that this approach can enable reconstruction of subglacial microbiomes across poles and ice ages. Our analysis suggests that subglacial microbiomes are dominated by chemolithoautotrophs, ultra-small microbes, and taxa closely related to those found in deep subsurface or extreme cold and hypersaline environments. These microbiomes split into two distinct clusters distinguished by oxygen availability and redox conditions, irrespective of geography or age. Geochemical measurements of subglacial redox state, measured either indirectly via precipitate calcite Fe and Mn concentrations or directly via water reduction potential, reproduce these same two clusters exactly. Our findings describe how subglacial water redox states are held in balance by microbes, hydrology, and oxygen input from fresh subglacial meltwater..

[B242] Developing profile mixture models for large-scale structural phylogenetics

Charley G. P. McCarthy[1], Dr. Paul Roginski[1], Romain B. Leroy[1], Prof. Purificación López-García[1], Dr Laura Eme[1,2], Dr David Moreira[1]
[1]Université Paris-Saclay & CNRS, AgroParisTech, France, [2]University of Rhode Island, Kingston, United States of America

Protein structure is more conserved than protein sequence. Protein structures may therefore possess useful phylogenetic signal at evolutionary depths beyond the reach of protein sequences.

The advent of high-quality protein structure prediction methods such as AlphaFold enables generation of protein structure data across many organisms and proteins, regardless of culturability or solubility. Combined with the development of the 3Di structural alphabet and structure search and alignment software like FoldSeek and FoldMason, it is now possible to perform structural phylogenetics using the same approaches as conventional sequence-based phylogenetics.

Sequence phylogenetics typically incorporates a substitution matrix which describes the substitution process at a single site, and a profile mixture model which accounts for differing site-specific processes across many sites. These models can be estimated empirically or from user-defined data. Equivalent models for structural phylogenetics have not been readily available.

A study by Garg and Hochberg (2025) presented a structural substitution matrix estimated from the Swiss-Prot AlphaFold database. Structural phylogenetics using this matrix showed promising results in reconstructing the phylogenies of some universal paralogs, outperforming sequence phylogenetics for the same paralogs. For larger-scale analysis involving many phylogenetic markers, structural profile mixture models would be desirable.

We have estimated structural profile mixture models, following the approaches used to estimate the C-series and UDM models for sequence phylogenetics. Our models are estimated from large clusters of AlphaFold-predicted protein structures sampled from prokaryotes and eukaryotes. We will show examples of their application to structural phylogenetic analysis of deep relationships within the tree of life.

[B243] The emergence of metabolisms through Earth history and implications for biospheric evolution

Edmund Moody[1], Tom Williams[2], Sandra Álvarez-Carretero[3], Gergely Szöllősi[4], Prof Davide Pisani[5], Timothy Lenton[6], Prof Philip Donoghue[5]
[1]University Of Barcelona, , Spain, [2]University of Bath, , United Kingdom, [3]University College London, , United Kingdom, [4]Okinawa Institute of Science and Technology Graduate University, , Japan, [5]University of Bristol, , United Kingdom, [6]University of Exeter, , United Kingdom

We investigate the evolution of microbial metabolisms from the last universal common ancestor to the extant biota through comparative phylogenomics, reconciling the evolution of the genes that underpin metabolic pathways with a time-calibrated tree of life. We find that the majority of metabolic pathways were established within the first 2 billion years of Earth history, with pathways accreting at different rates. Methanogenesis and acetogenesis are recovered to be among the earliest energy metabolisms, whereas photosynthetic pathways achieved completeness by 2 Ga, much later than most previous studies have envisaged. Horizontal exchange of metabolic genes is widespread, but it has occurred largely among closely related lineages and for some pathways there is a strong signal of vertical inheritance. We also find that the rate of horizontal gene transfer has been higher in Bacteria than in Archaea through evolutionary history. Finally, we evaluate how our reconstructed history of metabolism can help to constrain hypotheses of biospheric evolution, considering the entropic and Darwinized Gaia hypotheses as well as a simple neutral model for the assembly of biogeochemical cycles.

[B244] The Dayhoff Exchange Score - a new metric to understand entropic saturation in phylogenomic datasets

James Fleming[1,2], Prof Torsten Struck[2]
[1]University Of Barcelona, Barcelona, Spain, [2]University of Oslo Natural History Museum, Oslo, Norway

Entropic saturation - where large amounts of independent changes along branches within a dataset obscure its phylogenetic signal - is a significant issue in deep time phylogenetic datasets. To combat this, at deep time scales, amino acid alignments are used in contrast to nucleotides, as the combination of a larger alphabet and the wobble effect makes the average amino acid more conserved over time. However, even here, entropic saturation in phylogenetic datasets may drive some key controversies in the evolution of early Metazoa and beyond.

The Dayhoff Exchange score is a novel metric to rapidly calculate entropic saturation in amino acid datasets at the whole dataset and sequence level. In the same way that exchanges within nucleotide categories should be conserved with respect to between category changes (transitions and transversions), this should be true of the six Dayhoff categories - functional categories that can determine an amino acid's structure and secondary properties. The DE-Score measures this ratio, then normalises it for dataset size, to output a value that can be easily compared to a critical saturation point, and to the DE-Scores of other alignments. The DE-Score is a biologically-informed, easy to comprehend amino acid saturation metric that requires no prior phylogenetic analysis, is unbiased by dataset size and can be directly compared between alignments. This metric is significantly correlated with more direct measures of entropy, and outperforms many current existing metrics.

[B245] GLADE: Accurate Reconstruction of Orthogroup Histories and Gene Gain, Loss, and Duplication

Dr Laurence Belcher[1], Prof Steven Kelly[1]
[1]University Of Oxford, , United Kingdom

Changes in gene content through gain and loss play a key role in the adaptation and diversification of species. Accordingly, our ability to detect and accurately document the history of these changes is important for our understanding the evolutionary trajectories of life on Earth. Here we present GLADE, a tool that accurately reconstructs gene gains, losses, duplications for a set of species under consideration and uses this information to infer ancestral gene contents for every speciation event in the species tree. GLADE requires as input only a standard OrthoFinder results directory, and outputs the full evolutionary history of every orthogroup, including branch-specific changes and reconstructed ancestral genomes. We benchmark GLADE using both real and simulated data and and show that GLADE accurately identifies orthogroup gains, losses, and duplications, and reconstructs ancestral orthogroup sizes with higher precision and overall accuracy than any competitor method. To illustrate the utility of the method, we apply GLADE to a dataset of 78 mammalian genomes and uncover repeated contractions in orthogroups associated with tooth formation on branches leading to ant- and termite- eating mammals - revealing convergent genomic signatures underlying this dietary specialization. GLADE and accompanying documentation and tutorials are freely available at https://github.com/lauriebelch/GLADE/.

[B246] A Comprehensive Phylogenomic Framework for Brachiopod Evolution among Lophotrochozoans

Mingzhu Yang[1], Aodhán Butler[2,3], Natalia López Carranza[4], Jakob Vinther[1], Rowan Whittle[5], Daniela Henkel[6], Hana Jurikova[7], Gert Wörheide[8], Dr Mattia Giacomelli[9], Prof Philip Donoghue[1], Prof Davide Pisani[1], Sandra J. Carlson[10], Erik A. Sperling[2]
[1]University of Bristol, Bristol, UK, [2]Stanford University, Stanford, USA, [3]Uppsala University, Uppsala, Sweden, [4]University of Kansas, Lawrence, USA, [5]British Antarctic Survey, Cambridge, UK, [6]GEOMAR Helmholtz Centre for Ocean Research Kiel, Munich, Germany, [7]University of St Andrews, St Andrews, UK, [8]Ludwig-Maximilians-Universität, Munich, Germany, [9]University of Barcelona, Barcelona, Spain, [10]University of California, Davis, Davis, USA

Brachiopods were among the first metazoans to appear during the Cambrian radiation. They are one of the most informative clades for the study of topics requiring the integration of knowledge from biology and palaeontology, such as the assembly of animal body plans, the evolution of biomineralized structures, and rates of molecular and morphological evolution. Answering such questions requires a robust phylogeny of the living representatives, yet persistent questions remain about lophophorate monophyly, brachiopod monophyly, and the relationships of phosphatic and calcitic brachiopods. We present the first phylogenomic analysis of Brachiopoda and close lophotrochozoan relatives, inferred using state-of-the-art across-site compositional heterogeneous models. We find that brachiopods are monophyletic, with the clade of inarticulated phosphatic lingulids and discinids (Linguliformea) sister to the inarticulated calcitic Craniiformea, and both together as sister to the clade of articulated calcitic Rhynchonellata (Rhynchonellida, Thecideida, and Terebratulida). These phylogenomic results are largely consistent with earlier morphological analyses of extant Brachiopoda, but Thecideida is unexpectedly nested within Terebratulida. We recover a monophyletic Lophophorata, with strong support for Brachiopoda as the sister group of Bryozoa plus Phoronida, in agreement with recent phylogenomic analyses of the Spiralia. Our brachiopod phylogeny implies at least one evolutionary transition in primary mineralogy, which is unknown in any other metazoan group.

[B247] Database-centred curation of BUSCO derived phylogenomic datasets

Morgan Jones[1], Ms Mingzhu Yang[1], Dr Mattia Giacomelli[2], Dr Chrstopher Laumer[3], Prof Philip Donoghue[1], Prof Davide Pisani[1]
[1]University Of Bristol, , United Kingdom, [2]University of Barcelona, , Spain, [3]Natural History Museum, London, United Kingdom

The rapid expansion of genomic data in public repositories such as NCBI Genome offers greater opportunities for large-scale phylogenomic analyses. However, the continuously growing volume of data presents major challenges for building robust phylogenomic datasets. Genome-wide superalignments are often computationally inefficient and sensitive to paralogy, while reduced datasets based on single-copy orthologs can provide a more tractable and accurate approach for species tree inference (Pisani et al., 2022). The BUSCO pipeline (Simão et al., 2015) is widely used to identify such markers and to assess genome completeness. However, there can be substantial disagreement between BUSCO-defined single-copy orthologs and orthogroups inferred using de novo methods such as OrthoFinder (Emms and Kelly, 2015; Liu et al., 2024), raising concerns about hidden paralogy in BUSCO-derived datasets.

Here, we present a database driven command-line program designed to systematically curate and clean BUSCO-based phylogenomic datasets. Rather than treating BUSCO output as definitive, our framework explicitly evaluates BUSCO single-copy candidates against orthogroups inferred from comparative genomic analyses, retaining only those genes that show consistent single-copy ortholog identity across high-quality reference proteomes. Ortholog candidates, efforts to reduce hidden paralogy, and intermediate results are persistently recorded in a database, enabling reproducible dataset construction and iterative refinement as new genomes are published.

We then demonstrate this workflow using an ecdysozoan case study, constructing and comparing BUSCO-derived datasets before and after paralogy-aware curation. We aim to understand how these changes to composition can impact the inferred relationships among the moulting animals.

[B248] Revisiting deep archaeal relationships with an expanded phylogenomic dataset

Pedro Romero-Tena[1], Romain Leroy[1], Ana Gutiérrez-Preciado[1], Dr Laura Eme[1,2], Dr Purificación López-García[1], Dr David Moreira[1]
[1]Unité d'Ecologie Systématique et Evolution, CNRS, Université Paris-Saclay, AgroParisTech, Site IDEEV 12, Route 128, 91190, Gif-sur-Yvette, France, [2]Department of Cell and Molecular Biology, Center for Biotechnology and Life Sciences, University of Rhode Island, 120 Flagg Road 02881, Kingston, USA
Archaeal diversity is still being uncovered, and newly sampled lineages continue to reshape the phylogeny of the third Domain of life. Although the monophyly of several major clades is now well supported, the placement of many lineages, especially near the deepest splits, remains unresolved. This uncertainty likely reflects a combination of limited taxon sampling, heterogeneous evolutionary rates, lineage-specific adaptations (including compositional biases associated with extreme environments or to parasitism), and horizontal gene transfer, all of which can mislead phylogenomic inference.
To address these challenges, we conducted comprehensive phylogenomic analyses of Archaea using an expanded and rigorously curated dataset. We assembled an extensive set of conserved proteins spanning diverse cellular functions, integrating markers commonly used in recent studies while substantially expanding taxon sampling to include underrepresented clades and deep-branching taxa. We inferred archaeal phylogenies from concatenated protein alignments using state-of-the-art maximum likelihood approaches and evaluated robustness across alternative marker and taxon subsamplings. Finally, we mapped key evolutionary traits onto the updated phylogeny, including archaeal-specific metabolisms such as methanogenesis and adaptations such as hyperhalophily and epibiotic parasitism, to infer their relative timing and order of emergence within Archaea.

[B249] IQ2MC: A New Framework to Infer Phylogenetic Time Trees Using IQ-TREE 3 and MCMCtree with Mixture Models

Piyumal Demotte[1], Muthukumaran Panchaksaram[2], Hashara Kumarasinghe[1], Nhan Ly-Trong[1], Mario dos Reis[2], Dr Bui Quang Minh[1]
[1]School of Computing, College of Systems and Society, Australian National University, Canberra, Australia, [2]School of Biological and Behavioural Sciences, Queen Mary University of London, London E1 4NS, United Kingdom
IQ-TREE and MCMCtree are widely used tools to infer phylogenies and estimate divergence times, respectively. As MCMCtree performs fast Markov Chain Monte Carlo sampling to infer the times along a fixed tree topology, it would be natural to use IQ-TREE to estimate the tree. However, it is currently not possible to integrate these tools seamlessly as MCMCtree requires pre-calculation of the gradient and Hessian for fast approximation of the likelihood, which is unavailable in IQ-TREE. Furthermore, MCMCtree only implements a small subset of substitution models, and complex models such as the mixture models are not available. This is an important limitation because complex substitution models are required for reliable estimates of divergence times in deep phylogenies. Here, we introduce the new IQ2MC pipeline, which facilitates the integration of IQ-TREE 3 and MCMCtree, substantially speeds up the pre-calculation steps, and allows the use of a wide range of IQ-TREE's complex substitution and partition models in divergence time inference. In the IQ2MC pipeline, IQ-TREE 3 is used to calculate the gradient and Hessian of the substitution model likelihood, and then MCMCtree is used for MCMC sampling of divergence times. We showcase the practical benefits of IQ2MC by quantifying the effects of DNA and amino acid mixture models on divergence time estimation using simulated data and four empirical datasets representing placental mammals, plants, eukaryotes/prokaryotes, and metazoans. Our new pipeline provides speedups of up to two-fold compared with MCMC-based timetree inference in other existing software.

[B250] Parallel and scalable balanced minimum evolution algorithm for large-scale phylogenetic inference

Qiyun Zhu[1,2], Chris Tapo[1]
[1]Biodesign Center for Fundamental and Applied Microbiomics, Arizona State University, Tempe, United States, [2]School of Life Sciences, Arizona State University, Tempe, United States
The rapid growth of biological sequence data demands phylogenetic methods that are both scalable and accurate. Distance-based approaches, most notably neighbor joining (NJ), are widely adopted not only in molecular evolution but also in emerging applications involving alternative distance measures. Although NJ has benefited from decades of optimization, it is fundamentally a greedy heuristic for the balanced minimum evolution (BME) problem. A mathematically robust BME algorithm has been developed and implemented in the FastME package, producing tree topologies more accurate than NJ, and competitive with more computationally intensive methods. However, beyond the original FastME implementation, there has been little development aimed at scaling BME to the size of modern phylogenomic datasets. We present a redesigned implementation of the BME algorithm that substantially improves performance while preserving optimality with respect to the BME objective. The key algorithmic improvement is the replacement of recursive tree traversal with a flattened, incrementally growing tree array. This design not only reduces traversal overhead, improves memory locality, but also for the first time enables parallelization of the BME algorithm, as it permits direct indexing of subtree nodes. Benchmarks show that our implementation achieves more than 20-fold speedup and uses roughly half the memory of FastME, while producing identical or equally optimal trees under the BME criterion. It can effectively analyze datasets with over 100,000 taxa, which are otherwise prohibitive in FastME. Our algorithm is released as part of the open source Python library scikit-bio. It substantially broadens access to robust BME-based phylogenetic inference at scale.

[B251] The effect of gene tree dependence on summary methods for species tree inference

Wanting He[1], Prof. Celine Scornavacca[2], Dr Yao-ban Chan[1]
[1]The University Of Melbourne, , Australia, [2]Université Montpellier, , France
Gene trees often differ from each other and from the underlying species tree due to processes such as incomplete lineage sorting. Species trees are generally inferred by combining gene trees reconstructed from sequence data using summary methods, among which ASTRAL is widely used. While prior simulation studies suggest that ASTRAL is highly accurate, they generally assume input gene tree are independent, which corresponds to infinite recombination between loci. This assumption is unrealistic, as nearby or co-evolving genes have dependent phylogenies.

We develop a method for simulating a gene tree conditional on another dependent gene tree, allowing explicit control over the level of dependence between loci. Using simulated gene trees under our model, we reassess the accuracy of ASTRAL when its independence assumption is violated, considering both true gene trees and gene trees estimated from sequence alignments. Our results show that a fixed level of dependence effectively reduces the number of independent loci by a constant factor, thereby decreasing inference accuracy in a predictable manner.

We further explore the implications of gene tree dependence for contemporary phylogenomic datasets. Loci are often sampled far apart to achieve near independence, which limits the number of genes available for inference. However, full independence is not required for accurate species tree estimation, and excluding loci may reduce accuracy. This introduces a trade-off between the number of genes included and their dependence. To address this, we propose a method to identify the minimum genomic separation required to maintain satisfactory inference accuracy.

[B252] Implementing a general SMC approximation to enable whole chromosome simulations beyond humans.

Hossameldin Ali[1], Gertjan Bisschop[1], Abdelmajid OMARJEE, Derek Setter, Konrad Lohse, Jerome Kelleher[1]
[1]University of Oxford, , United Kingdom

Msprime is a state-of-the-art population genetics simulator that has become an essential tool for evolutionary analysis. Msprime primarily uses the standard coalescent with recombination model (CwR model), i.e., Hudson's model, to simulate Ancestral Recombination Graphs (ARGs) backwards in time. While the efficient implementation of the CwR model in msprime enables large-scale simulations in a short time, species with large recombination rates and large population sizes, like Drosophila, remain challenging to simulate. Here we present a new implementation of a general Sequentially Markovian Coalescent (SMC) model in msprime 1.4 that addresses this gap and makes whole-chromosome simulations of species with Drosophila-like parameters feasible. More significantly, the newly implemented SMC model is a flexible model where the SMC approximations relative to the CwR can be adjusted by the user from a full SMC approximation model, to the SMC' model, and all the way to the full CwR model.

The SMC' model, in particular, is considered a good approximation of the CwR model. This is supported by the similar distribution of pairwise coalescence times between ARGs simulated under either model. Using the new implementation of the SMC model, we show the differences between ARGs simulated under the CwR model and SMC'. Fundamentally, the SMC' model approximates the CwR model by ignoring common ancestor events between disconnected genomic segments. Our findings suggest that while these approximations might not affect an ARG's coalescence time distribution, the unmodelled events connect the different trees making up the ARG, and hence affect an ARG's structure.

[B253] The Interplay of Kinship and History: Why Close Relatives Improve Demographic Inference from IBD

Cole Williams[1], Sohini Ramachandran[1]
[1]Brown University, Providence, United States

Demographic history shapes patterns of identity-by-descent (IBD) sharing, yet the interplay between genealogical relatedness and population history is often overlooked. While studies usually remove close relatives from population-genetic analyses to reduce bias in parameter inference, modern datasets—where, for example, 30-74% of biobank participants may have close relatives sampled—make this approach increasingly impractical and potentially detrimental.

Using msprime, we simulated IBD sharing across diverse scenarios, including a constant population size, a European out-of-Africa model, and the Quebec founder population. We demonstrate that IBD segment age depends on segment length, genealogical kinship, and population history. For example, 2 cM segments in the Quebec model have a median age of 16 generations, compared to 90 generations in the out-of-Africa model.

Furthermore, we reassess the tool IBDNe, which leverages coalescent expectations for IBD segment lengths to infer recent effective population size (Ne). We find that including close relatives in randomly ascertained samples produces substantially more accurate estimates of recent Ne (RMSE 265 vs. 7,817 for the most recent 5 generations in simulations with constant Ne=10,000). In general, filtering relatives induces a 2–3-fold upward bias and creates oscillatory trajectories lasting 10-25 generations, meaning removing relatives discards the primary signal required for recent Ne inference. For instance, removing second-degree relatives discards 65% of segments estimated to be one generation old. Our findings suggest that close relatives represent essential genealogical structure. ARG-based approaches—which explicitly model the full genealogical relationships among samples—provide a natural framework for incorporating rather than discarding close relatives in demographic inference.

[B254] Reconstructing hybrid speciation via the ancestral recombination graph

Alexander Mackintosh[1], Oliver White[2], Mark Carine[2], Prof Mark Chapman[3], Jairo Patiño[4], José Cerca[1,5,6]

[1]Swedish Museum of Natural History, Stockholm, Sweden, [2]The Natural History Museum, London, UK, [3]University of Southampton, Southampton, UK, [4]Instituto de Productos Naturales y Agrobiología (IPNA-CSIC), La Laguna, Tenerife, Spain, [5]SciLifeLab, Stockholm, Sweden, [6]University of Oslo, Oslo, Norway

Homoploid hybrid speciation (HHS) is the formation of a reproductively isolated hybrid species without a change in ploidy. Recent investigations of HHS have focused on confirming the hybrid origin of species using genome sequence data. Obtaining insight into the HHS process itself is more challenging and requires approaches that leverage much of the information contained in patterns of sequence polymorphism. Within the genus Argyranthemum (Asteraceae), two species on the island of Tenerife have been suggested to have evolved independently from hybridisation between the same parental species. Here we study the history of HHS in these species using an ancestral recombination graph (ARG) framework. We perform MCMC sampling of ARGs and use the resulting marginal genealogies to reconstruct a five-taxon demographic history with multiple hybrid-speciation events and variation in continuous gene flow through time. We identify regions of the genome where local genealogies are better explained by a demographic history with reduced post-speciation gene flow. This approach therefore provides information about both the temporal dynamics and genetic basis of HHS in nature.

[B255] Unraveling the dynamics of selection at HLA loci: the impact of balancing selection at different levels of biological organization

André Maróstica[1], Alfredo Marin[1], Professor Montgomery Slatkin[2], Professor Diogo Meyer[1]
[1]Universidade De São Paulo, São Paulo, Brazil, [2]UC Berkeley, Berkeley, United States of America
Balancing selection increases genetic variability and the average coalescence time at selected sites relative to neutral expectations. The Human Leukocyte Antigen (HLA) loci are known to be under such selection, as evidenced by the existence of Trans-Species Polymorphism (TSP) and extreme nucleotide diversity. However, their putative targets of selection remain unresolved, as it is plausible that individual SNPs, alleles defined by SNP strings, or multi-locus haplotypes characterized by alleles at different HLA loci define the molecular phenotype that is selected. Another key challenge is elucidating the mode of selection at the HLA, which may be explained by selective regimes other than balancing selection, such as partial selective sweeps favoring advantageous alleles. We approached these questions using publicly available datasets, evolutionary simulations, and Ancestral Recombination Graphs (ARGs). We found extremely low nucleotide diversity and shallow coalescence time among gene copies or chromosomes sharing an HLA allele or haplotype, although copies from different HLA lineages had deep coalescence times. Additionally, we found that high-frequency alleles and haplotypes had lower within-lineage diversity and shorter coalescence times than infrequent ones, suggesting that selective events recurrently homogenize them. Using simulations, we showed that this pattern of deep coalescence times between alleles, and shallow within, is consistent with symmetric balancing selection, which retains lineages over long spans of time but also promotes allelic turnover. These results clearly indicate that balancing selection shaped genetic diversity patterns across multiple levels of biological organization at the HLA, contributing to both deep and recent phylogenetic relationships.

[B256] The dawn of phylogenetic comparative graph neural networks (PCGNNs): Applications to quantitative trait imputation and ancestral state reconstruction

Austin Patton[1], Ryan York[1]
[1]Arcadia Science, Emeryville, United States

Phylogenetic comparative methods (PCMs) are foundational tools in evolutionary biology and beyond, enabling researchers to account for evolutionary non-independence in comparative studies across species. Chief among these are methods to study trait evolution, infer ancestral character states, and impute missing species traits. However, conventional approaches rely on fitting explicit evolutionary models, often via maximum likelihood optimization, which scale poorly to many traits or large phylogenies and can suffer from model inadequacy when applied to real biological data. Here, we present a phylogenetic comparative graph neural network (PCGNN) architecture designed to jointly perform ancestral state reconstruction and trait imputation from partially missing species trait data and a phylogenetic tree. Our model is based on a GPS Transformer architecture with relative positional encodings that capture phylogenetic distances between nodes. We trained the model on 500,000 simulated phylogenies spanning five tree diversification models, with ten traits per tree simulated under five trait diversification models. We compared PCGNN's performance to mvMORPH, a widely used maximum likelihood implementation of multivariate trait evolution models. For trait imputation, performance was largely comparable between methods, with modest differences limited to specific tree and trait model combinations. In contrast, PCGNN consistently outperformed likelihood-based methods for ancestral state reconstruction, regardless of prediction depth. Importantly, inference with PCGNN was orders of magnitude faster than likelihood-based approaches and scaled to large phylogenies for which likelihood methods were computationally intractable. Together, these results highlight the potential of graph neural networks as fast, flexible, and scalable alternatives to classical PCMs in phylogenetic comparative biology.

[B257] Towards scalable ARG-based inference of speciation

Francisco De Borja Campuzano Jiménez[1], Dr. Arthur Zwaenepoel[1], Hannes Svardal[1,2]
[1]Evolutionary Ecology Group, University of Antwerp, Antwerp, Belgium, [2]Naturalis Biodiversity Center, Leiden, Netherlands

The possibility of reconstructing ancestral recombination graphs (ARGs) from empirical data has revolutionised the field of population genetics. In the past few years, an increasing number of methods that treat reconstructed ARGs as data have been created. However, despite being an extension of phylogenetic trees, ARGs remain less known in the field of speciation genomics.

In this work, we explore a set of highly scalable approximations of the likelihood of ARGs in the structured coalescence regime relevant for speciation. This novel approach serves a double purpose. On the one hand, we aim to quantify the bias introduced by state-of-the-art ARG reconstruction software (SINGER) and decrease the bias in downstream analysis by conditioning on a known phylogenetic tree using a technique known as importance sampling. On the other hand, we propose an approximate scheme for doing joint inference of demography and selection against gene flow. We found that fitting models in which effective migration rate varies along the genome directly to the reconstructed ARG is an effective approach to identify barriers to gene flow.

This work highlights the promising potential of ARG-based approaches in speciation research while also addressing some key methodological challenges.

[B258] Methods for inferring coalescent tree topologies from genomic data: a comparison based on the transmission of reproductive success

Frederic Austerlitz[1], Mr Ferdinand Petit[1,2], Dr Jérémy Guez[1,2], Dr Léo Planche[2], Dr Evelyne Heyer[1], Dr Bruno Toupance[1], Flora Jay[2], Dr Fanny Pouyet[2]
[1]Laboratoire d'Eco-anthropologie, Paris, France, [2]Laboatoire Interdisciplinaire des Sciences du Numérique, Gif sur Yvette, France

Inferring coalescent trees from genomic data has become a major subject in population genomics, particularly with the recent advances in ancestral recombination graph reconstruction methods. Yet, how well these methods perform under imbalanced genealogies remains unclear. Such imbalances can arise from processes such as cultural transmission of reproductive success (CTRS) or positive selection. Using simulated genomic data, we benchmarked three major software packages, Relate, tsinfer and SINGER, by comparing the imbalance of reconstructed trees by these methods with that of the true simulated trees, for three indices that quantify this imbalance. All methods performed well under scenarios yielding balanced trees. However, their accuracy declined as imbalance increased. Performances also varied with mutation rate, recombination rate, and sample size. This study opens possibilities for applying these methods to infer CTRS or positive selection in large-scale genomic datasets, using simulation-based inference such as approximate Bayesian computation.

[B259] Identifying modern human-specific adaptations using Ancestral Recombination Graphs

Luiz Carlos Machado[1], Leyan Wang[2], Prof. Adam Siepel[1]
[1]Simons Center for Quantitative Biology, Cold Spring Harbor Laboratory, Cold Spring Harbor, United States, [2]Integrative Biology and Data Science, University of California, Berkeley, United States
Identifying loci under positive selection is essential for understanding the evolutionary history of modern humans. Particularly informative are alleles fixed or nearly fixed in contemporary populations but absent in Neanderthal and Denisovan genomes. These modern human-specific variants likely represent adaptive innovations, yet their detection remains challenging as classic selection scans have limited power to detect ancient, high-frequency selective sweeps.

To overcome this limitation, we employed an Ancestral Recombination Graph (ARG) framework that detects selection through distortions in local tree topologies and coalescence times. We first applied this approach to previously modern human-specific candidates, detecting strong and consistent selection signatures at loci including ADSL and NOVA1.

We then extended this analysis genome-wide across protein-coding regions, identifying dozens of non-synonymous variants that rose to high frequency (derived allele frequency > 0.95) in modern humans while remaining absent from archaic genomes. These variants exhibit characteristic genealogical signatures of selective sweeps, including reduced coalescence times around the selected sites.

Our results demonstrate that ARG-based methods provide greater power to detect modern human-specific adaptations than traditional summary statistics, particularly for ancient selective events that have reached or approached fixation. Beyond methodological advances, this work provides a curated set of high-confidence candidate coding variants for functional validation experiments, such as humanized models, enabling future investigation of the phenotypic consequences underlying these modern human-specific adaptations.

[B260] Folio: a stable encoding of phylogenies

Mark Tanaka[1], Eleanor Chadwick[1], Ruiting Lan[1], Andrew Francis[2]
[1]School of Biotechnology and Biomolecular Sciences, University Of New South Wales, Sydney, Australia, [2]School of Mathematics and Statistics, University Of New South Wales, Sydney, Australia

A phylogeny can be effectively stored or manipulated by encoding it suitably. For example, the Newick format conveniently stores trees in a machine- and human-readable form. Phylogenies can be enlarged as new sequences are added to databases, and such tree updating is useful where new taxonomic groups appear in real time, for instance, as pathogen isolates are sequenced. Although Newick has been extended to accommodate newly placed leaves, a further challenge is to preserve existing information in an encoding while expanding a tree by adding sequences iteratively. Here, we present a tree encoding - the folio - which is a set of leaf "addresses"; each address records the path from a reference vertex (e.g. the root) to a leaf. We have devised a simple set of rules to assign new addresses to placed leaves. The encoding is stable in that existing addresses do not change as further leaf addresses are added to the folio. The tree can be uniquely recovered from a folio of addresses. Due to the properties of the encoding, it has the potential to be used in a variety of phylogenetic analyses. For instance, it can help establish a stable classification scheme where pathogen trees grow during an epidemic. Because the folio encoding readily lends itself to extracting subtrees, it can also facilitate the analysis of local regions of a tree to identify notable clades. Finally, folios can be extended to encode phylogenetic networks.

[B261] Multiple Merger Detection with Polymorphism Data

Miles Anderson[1]
[1]Tum, Freising, Germany

The prevailing model used to describe the ancestry of samples is the Kingman coalescent, which assumes a tree consisting only of binary mergers. However, coalescent models which assume a multiple merger process often produce a better fit to observed data. Many methods for parameter inference on genomic data rely on statistical and mathematical properties of the Kingman coalescent. However, assumption of a Kingman coalescent process when a multiple merger process better reflects the biological reality, can result in parameter inferences that are wildly inaccurate. As such, the ability to discern whether an organism's reproduction more closely resembles a Kingman or multiple merger process, could reduce bias and inaccuracy from inference methods. Given the assumed prevalence of multiple merger in many organisms of interest, it is worth questioning how much of the current analysis of many economically and ecologically important species is biased by this model misspecification. With this in mind, we propose an ABC method that can leverage signals from allele frequencies and linkage disequilibrium in polymorphism data to detect the presence of multiple merger coalescence and reject a Kingman coalescence model. The novelty of this method is in its applicability to recombining genomes with arbitrary demographic histories, whereas hitherto existing methods primarily focus on non-recombining genomes and differentiating multiple merger from population expanding Kingman processes.

[B262] Museomics of the Unwelcomed. A Genomic Pipeline to Explore Historical Samples

Alice Piai[1,2], Dr. Toni De-Dios[3], Fabio Barteri[1,2], Dr. Carles Lalueza-Fox[1]
[1]Museu de Ciències Naturals de Barcelona, Barcelona, Spain, [2]Institute of Evolutionary Biology (IBE, UPF–CSIC), Department of Medicine and Life Sciences, Universitat Pompeu Fabra, Parc de Recerca Biomèdica de Barcelona (PRBB), Barcelona, Spain, [3]Estonian Biocentre, Institute of Genomics, University of Tartu, Tartu, Estonia

Natural history collections represent an exceptional and unexplored resource for temporal genomics and evolutionary biology. By comparing genetic variations across time points, historical specimens reveal molecular, environmental, and anthropogenic signatures of selection while offering the possibility to identify pathogens and study disease evolution. However, maintaining these collections requires a collaborative dialogue between researchers and curators to improve extraction and preservation methodologies, as well as continued genomic advances to maximize data recovery from highly heterogeneous DNA sources.

Here we present a bioinformatic pipeline for high-throughput analysis of low-DNA museum specimens, enabling ancient pathogen screening and generation of outputs suitable for population genomic analyses. Our framework integrates non-destructive DNA extraction with established bioinformatic tools, combining the nf-core/eager pipeline (https://nf-co.re/eager/2.5.1/) with KrakenUniq (https://github.com/fbreitwieser/krakenuniq) for pathogen detection, and downstream genotyping workflows optimized for low-coverage data.

To validate this approach, we are analyzing museum specimens of mosquitoes from three genera (Aedes, Culex, Anopheles), spanning multiple decades and geographic locations. Mosquitoes have shaped human history through dispersal via colonization and environmental changes. By integrating historical samples with modern data, we aim to reconstruct mosquito evolution, detect adaptation to urban contexts, identify signatures of pesticide resistance, and retrieve ancient pathogens to understand disease epidemiology across time.

Preliminary validation on museum specimens demonstrates successful DNA retrieval, establishing the feasibility of genomic exploration of these collections while preserving sample integrity. This pipeline, designed for mosquitoes but broadly applicable to diverse taxa, provides a preservation-conscious workflow for investigating evolution, adaptation and pathogen dynamics across temporal scales in diverse museum taxa.

[B263] Disentangling ancient admixture and incomplete lineage sorting using DNA from museum specimens, a case study in Sphingidae (Lepidoptera)

Dr. Victor Moreno-Gonzalez[1], Dr. Tomáš Melichar[2], Dr. Michael San Jose[3], Prof. Dr. Daniel Rubinoff[3], Anna K. Hundsdoerfer[1]

[1]Senckenberg Naturhistorische Sammlungen Dresden, Dresden, Germany, [2]Sphingidae Museum, Pribram , Czech Republic, [3]University of Hawaii, Honolulu, U.S.A.

Over the last decades, historical DNA (hDNA) has become an important source of genetic information for studies in systematics and evolutionary biology. Despite that, its use in organisms that have diverged recently and rapidly is still challenging. Hawkmoths (Lepidoptera: Sphingidae) are one of those groups that represent this type of taxa well. Among them, Hyles and Rhodafra present a long-standing taxonomic puzzle, with molecular data questioning their monophyly and validity. To shed light on this issue, a phylogenomic analysis was carried out using genomic data from numerous Hyles species and historical DNA from Rhodafra samples obtained in a dedicated ancient DNA laboratory. Phylogenies reconstructed using both nuclear SNPs and mitochondrial genomes support that both genera are monophyletic sister clades. In addition, introgression analyses reveal that ancient admixture and incomplete lineage sorting have shaped the evolution of the clade. Moreover, we found evidence of gene flow between Rhodafra and a basal Hyles lineage. This study demonstrates the power of integrating museum genomics to solve taxonomic problems and disentangle the effects of hybridization and lineage sorting in rapid radiations.

[B264] Climatic influences on the genetic connectivity of sky islands rodents

Damien Rivera[1], Nathan Upham[1]
[1]Arizona State University, Tempe, United States

The Madrean Sky Islands are a system of 56 mountains with mesic woodland habitat isolated by desert, located primarily in the southwestern United States and northern Mexico. These habitats have been repeatedly isolated and re-connected during Pleistocene climatic cycles, but with unknown impacts on the genetic connectivity of animals across the region. Here, we seek to understand how climatic cycles have affected genome evolution in populations of the Brush Mouse (Peromyscus boylii), a common rodent on these sky islands. We expect that sky island rodent dispersal is limited by historical habitat connectivity, resulting in higher genetic distinctiveness, smaller effective population sizes, and higher mutation rates in more isolated habitats, consistent with the drift-barrier hypothesis. To test this, we generated whole-genome sequences from individuals across 14 mountains using both historical (n=93; 1921-2001) and recently collected specimens (n=18; 2021-2023). Then, we calculated FST and compared genetic distances to environmental distances across sky islands. We find that P. boylii from the most environmentally isolated mountain are indeed the most genetically distinct (FST = 0.4861). However, the FST values for the other sampled sky islands did not follow the expectation, perhaps because P. boylii continues to occasionally disperse between mountains in close proximity. Future work will focus on identifying trios of mother-father-offspring from recently collected specimens (n=147; 2021-2025) to detect region-wide variation in germline mutation rates, increasing our understanding of how biogeography affects the drift-selection balance in wild populations.

[B265] Beyond bones: Pleistocene to Holocene transition in Central Europe through sedimentary ancient DNA

Dheeraj Chaudhary[1,2], Taylor Grandfield[3], Magdalena Krajcarz[4], Dr Maciej Tomasz Krajcarz[5], Dr Magdalena Sudoł-Procyk[4], Dr Marine Frouin[3], Dr Viviane Slon[1,2]
[1]The Gray Faculty of Medical and Health Sciences, Tel Aviv University, Tel Aviv , Israel, [2]The Dan David Center for Human Evolution and Biohistory Research, Tel Aviv University, Tel Aviv, Israel, [3]Department of Geosciences, Stony Brook University, Stony Brook, USA, [4]Department of Environmental Archaeology and Human Paleoecology, Institute of Archaeology, Faculty of History, Nicolaus Copernicus University in Torun, Torun, Poland, [5]Institute of Geological Sciences, Polish Academy of Sciences, Warszawa, Poland

The Pleistocene to Holocene transition in Europe was marked by a fundamental reorganization of terrestrial ecosystems, including the decline of cold-adapted fauna and the establishment of temperate biomes. Although this interval is central to understanding long-term ecological change, faunal reconstructions from archaeological sites are often fragmentary, partly due to uneven skeletal preservation and challenges in establishing robust chronological frameworks. Consequently, the timing and structure of faunal turnover at many sites remain poorly resolved.
Here, we use sedimentary ancient DNA (sedaDNA) to investigate this transition at two sites in southern Poland: Perspektywiczna Cave and Poręba Dzierżna. We extracted DNA from 76 sediment samples, converted the DNA fragments to single-stranded DNA libraries, and enriched them for mammalian mitochondrial DNA. Sequenced fragments were taxonomically assigned, and authenticity was assessed based on post-mortem damage patterns.
Twelve samples from Perspektywiczna Cave showed good ancient DNA preservation, yielding sedaDNA from ten mammalian families. These taxa include species consistent with the zooarchaeological record, and taxa not previously documented at the site. Several families exhibit layer-by-layer changes in species composition, including taxa with contrasting ecological preferences consistent with climatic shifts. Notably, some samples contain mixtures of Pleistocene and Holocene taxa, indicating post-depositional mixing, highlighting the importance of accounting for site formation processes in sedaDNA research.
Integrated with luminescence dating and archaeological, geological, and zoological analyses, these sedaDNA results enable the reconstruction of local faunal dynamics across a major prehistoric transition.

[B266] Ancient genomes illuminate the origins and dynamic history of East Asian cattle

Donghee Kim[1,2], Dr Naifan Zhang[3], Dr Xingcheng Wang[4], Ms Tianshu Li[4], Mr Jian Li[4], Ms Chang Li[4], Ms Shengnan Lian[4], Ms Xinyue Shao[5], Dr Songmei Hu[6], Ms Miaomiao Yang[7], Mr Jie Zhang[8], Mr Yongqiang Wang[8], Mr Qiurong Ruan[8], Mr Idilisi Abuduresule[8], Mr Linheng Mo[9], Ms Wenyan Li[10], Mr Xiaoning Guo[7], Ms Wenying Li[8], Mr Jing Shao[7], Dr Zhouyong Sun[7], Mr Yaqi Tian[7], Dr Hui Wang[11], Mr Ruilin Mao[12], Mr Cunshi Zhu[13], Mr Xiaoyang Wang[13], Ms Xiaoyan Ren[14], Dr Weilin Wang[15], Mr Yan Ding[7], Mr Pengcheng Zhang[16], Mr Liping Yang[7], Dr Jianen Cao[17], Ms Yu Dang[18], Mr Da Ha[19], Mr Wei Zhang[20], Ms Linshan He[21], Dr Chunxue Wang[4,22], Dr Lixin Wang[4,23], Dr Quanchao Zhang[4,22], Dr Jing Yuan[24], Dr Xiaohong Wu[25], Dr Chao Ning[25], Dr Dawei Cai[4,22,23], Dr Choongwon Jeong[1,2]
[1]School of Biological Sciences, Seoul National University, Seoul, Republic of Korea, [2]Institute for Data Innovation in Science, Seoul National University, Seoul, Republic of Korea, [3]National Centre for Archaeology, Beijing, China, [4]School of Archaeology, Jilin University, Changchun, China, [5]Department of Archaeology, University of Southampton, Southampton, United Kingdom, [6]Institute of Cultural Heritage, Shandong University, Qingdao, China, [7]Shaanxi Provincial Institute of Archaeology, Xi'an, China, [8]Xinjiang Institute of Culture Relics and Archaeology, Urumqi, China, [9]Hunan Provincial Institute of Cultural Relics and Archaeology, Changsha, China, [10]Hebei Provincial Institute of Cultural Relics and Archaeology, Shijiazhuang, China, [11]Institute of Archaeological Science, Fudan University, Shanghai, China, [12]Gansu Provincial Institute of Cultural Relics and Archaeology, Lanzhou, China, [13]Ningxia Institute of Culture Relics and Archaeology, Yinchuan, China, [14]Qinghai Provincial Institute of Cultural Relics and Archaeology, Xining, China, [15]School of Archaeology and Museology, Shanxi University, Taiyuan, China, [16]Shaanxi Yulin City Cultural Relics Archaeological Exploration Task Force, Yulin, China, [17]Institute for the History of Science and Technology, Inner Mongolia Normal University, Hohhot, China, [18]Institute of Cultural Relics and Archaeology of Inner Mongolia Autonomous Region, Hohhot, China, [19]Hulunbuir Museum, Hulunbuir, China, [20]Heilongjiang Provincial Institute of Cultural Relics and Archaeology, Harbin, China, [21]Yunnan Provincial Institute of Cultural Relics and Archaeology, Kunming, China, [22]Bioarchaeology Laboratory, Jilin University, Changchun, China, [23]Research Center for Chinese Frontier Archaeology, Jilin University, Changchun, China, [24]Institute of Archaeology, Chinese Academy of Social Sciences, Beijing, China, [25]School of Archaeology and Museology, Peking University, Beijing, China

The evolutionary history of domesticated cattle in East Asia for the last 5,000 years remains largely obscure. Here, we investigate the origins and evolution of cattle genetic diversity in China by analyzing shotgun genome sequences of 166 ancient bovines spanning a 10,000-year period, encompassing now-extinct East Asian aurochs and domesticated cattle from key archaeological cultures. East and North Asian aurochs were distinct from western aurochs, although East Asian aurochs harbored ~15% western ancestry. The first domesticated cattle in the Yellow River region derived ~10% of their ancestry from local aurochs with an uneven genome-wide distribution. Early cattle from Xinjiang were genetically distinct and partially contributed to the later northern Chinese cattle. Indicine admixture became widespread only in the Medieval period in northern China.

[B267] Genomic Continuity and Subtle Change in Central Asian Sheep from the Bronze to the Iron Age

Germán Hernández Alonso[1], André Elias Rodrigues Soares[2], Nikolaus Boroffka[3], Kai Kaniuth[4], Oskar Schröder[5], Leonid Sverchkov[6], Norbert Benecke[7], Torsten Günther[1]
[1]Human Evolution Program, Department of Organismal Biology, Uppsala University, Uppsala, Sweden, [2]National Bioinformatics Infrastructure Sweden, Department of Medical Biochemistry and Microbiology, Uppsala University, Uppsala, Sweden, [3]Eurasia Department, German Archaeological Institute, Berlin, Germany, [4]Department of Ancient and Modern Cultures, Institute of Near Eastern Archaeology, Ludwig-Maximilians-Universität München, Munich, Germany, [5]Department of River Ecology, Helmholtz Center for Environmental Research, Magdeburg, Germany, [6]Institute of Fine Arts, Academy of Sciences of the Republic of Uzbekistan, Tashkent, Uzbekistan, [7]Department of Natural Sciences, German Archaeological Institute, Berlin, Germany

The domestication of sheep around 10,000 years ago in the Fertile Crescent was a key development in the rise of agropastoral societies. Although the genetic history of European sheep has been extensively studied, Central Asia—a crucial Eurasian crossroads—remains underexplored. Here, we investigate the genetic diversity and evolutionary history of Central Asian domestic sheep using whole-genome data from 53 ancient bone remains dating to the Bronze and Iron Ages (4,000–2,000 years ago) from southern Uzbekistan.

Admixture analyses indicate that all Asian ancient sheep genomes show additional genetic introgression from wild Asiatic Mouflon relative to Neolithic sheep, likely happening during the initial eastward expansion of domestic sheep. These admixture signals remain stable over time, suggesting minimal later recruitment from wild stock.

Our temporal dataset reveals a largely homogeneous regional gene pool, suggesting long-term genetic continuity, yet subtle shifts reflecting gene flow from other regions are detectable over time. Late Bronze Age sheep show greater genetic affinity to contemporaneous populations from the Eurasian steppe than to both earlier Middle Bronze Age and later Iron Age genomes. This signal appears diluted in Iron Age sheep, possibly due to gene flow from western regions, as indicated by closer affinity with Iranian Iron Age sheep. These dynamics may reflect broader temporal influences, including Steppe pastoralist expansions in the Bronze Age and the subsequent dominance of the Achaemenid Empire.

This study demonstrates the value of temporal genomic data for detecting local demographic processes and emphasises the deep entanglement of human and livestock evolution.

[B268] Extending the Shelf Life: Optimizing Ancient DNA Recovery from Highly Degraded Material in Museum Collections

Hannah Moots[1,2,5], Juliana Larsdotter[1,2], Florentine Tübbesing[1,2], Dr. Jarosław Wilczyński[3], Dr. Marc Händel[4], Love Dalén[1,2], Dr. David Díez-del-Molino[1,2]
[1]Centre For Palaeogenetics, Stockholm, Sweden, [2]Stockholm University, Stockholm, Sweden, [3]Polish Academy of Sciences , Kraków, Poland, [4]Austrian Academy of Sciences, Vienna , Austria, [5]Swedish Natural History Museum, Stockholm, Sweden

Here, we present an optimized protocol that significantly improved DNA recovery from a set of ~30,000-year-old mammoth sub-fossils, some of which have been curated in museum collections for over 100 years. We conducted two key comparisons to evaluate the impact of pre-extraction and damage-repair treatments. First, we compared a traditional drilling-based approach without bleach or predigestion to a crushing method using a mortar and pestle, followed by a 4-minute bleach treatment. Second, we assessed the effects of three USER enzyme damage-repair strategies: 3-hour, 30-minute, and no treatment. The protocol optimization was facilitated by the use of a plate-based library preparation method (Gilardet et al 2025), which allowed us to prepare 84 libraries (six per individual) in a single batch, minimizing batch effects and enabling robust cross-treatment comparisons.

Our findings demonstrate the benefits of a bleach pre-treatment in recovering DNA from these materials, as it significantly reduces DNA from non-target organisms. The USER enzyme treatment conditions had less of an impact on DNA recovery, indicating that partial and full USER treatments are feasible for material of this antiquity. We emphasize the importance of flexible, context-sensitive methods when working with highly degraded material. These optimizations have broad relevance for improving aDNA yields from museum-held collections, expanding the scope of specimens that can be included in genetic analyses of the past.

[B269] Yak X: an archaic bovine from Denisova Cave

Jonas Oppenheimer[1], Alexandre Gilardet[1], Maxim Kozlikin[2], Katerina Douka Douka[3], Peter Heintzman[1], Beth Shapiro[4], Love Dalén[1]
[1]Centre for Palaeogenetics, Stockholm, Sweden, [2]Institute of Archaeology and Ethnography, Siberian Branch, Russian Academy of Sciences, Novosibirsk, Russia, [3]Department of Evolutionary Anthropology, Faculty of Life Sciences, University of Vienna, Vienna, Austria, [4]Department of Ecology and Evolutionary Biology, University of California Santa Cruz, Santa Cruz, USA
Ancient DNA offers a direct lens into the past, enabling the discovery of diversity which no longer exists today. An extinct mitochondrial lineage deeply divergent from yaks, termed yak X, was recently recovered from Denisova Cave and its surroundings, although the genome-wide relationship of this lineage to yaks and other bovines remained unclear. Here, we generate nuclear genomes from 10 yak X specimens within the broader Altai region that date from ~200,000 to 27,000 years ago. We show that they belong to a highly distinct bovine group which diverged from yaks more than 500,000 years ago. This population may correspond to the Baikal yak, which has been morphologically described from the fossil record. We also document potential turnover in yak X around the Eemian interglacial, which may have been accompanied by a decline in genetic diversity. Finally, unlike divergent hominin lineages with the Altai, we find little evidence for gene flow between yak X and other bovines found in the region, including bison and aurochs, despite their co-occurrence. Overall, these results suggest that yak X corresponds to a newly discovered bovine species and reveals the power of deep-time paleogenomics to still uncover new megafaunal lineages and document population transitions.

[B270] A multi-species study of domestic animals in Neolithic Sweden

Katia Bougiouri[1], Lasse Eskildsen[1], Malou Blank[2], Torbjörn Ahlström[3], Tony Axelsson[2], Karl-Göran Sjögren[2], Frederik Seersholm[1], Martin Sikora[1]
[1]Globe Institute, University of Copenhagen, Copenhagen, Denmark, [2]Department of Historical Studies, University of Gothenburg, Gothenburg, Sweden, [3]Department of Archaeology and Ancient History, Lund University, Lund, Sweden

The relationship between humans and domestic species has been central to the development of human cultures, societies and migrations. Advances in ancient genomic approaches have led to an increasing number of ancient animal genomes from time periods going as far back as the Paleolithic. However, most studies have focused on continental level questions related to domestication origins and large scale population patterns. More local scale questions focusing on sociocultural aspects within sites and regions, such as kinship and relatedness, can offer unique cases to study mobility and breeding patterns of domestic animals through time, while also providing further insights of disease prevalence and pathogen evolution. In this study, we delve into local genetic patterns of domestic species from Neolithic sites found in the area of Falbygden in western Sweden, where previous studies on humans have reconstructed pedigrees of up to six-generations and found evidence of plague[1]. By using >200 genomes from domestic cattle, pigs and dogs, we explore patterns of relatedness and inbreeding within and between sites from Sweden. Our results highlight the importance of local scale studies to provide new insights into domestic management strategies, livestock trade networks and husbandry in Neolithic Scandinavia.

1. Seersholm, F. V. et al. Repeated plague infections across six generations of Neolithic Farmers. Nature 632, 114–121 (2024).

[B271] Dogs were widely distributed across Western Eurasia during the Palaeolithic

Lachie Scarsbrook[1,2], William A Marsh[3], Eren Yüncü[4], Lizzie Hodgson[5], Audrey T Lin[6,7], Maria De Iorio[8], Olaf Thalmann[1], Mark G Thomas[9], Dr Anders Bergström[10], Angela Noseda[11,12], Sarieh Amiri[13,14], Fereidoun Biglari[15], Dušan Borić[16], Katia Bougiouri[17], Alberto Carmagnini[1], Maddalena Giannì[18], Tom Higham[18,19], Ophelie Lebrasseur[20,2], Anna Linderholm[21,22], Marcello A Mannino[23], Caroline Middleton[24], Gökhan Mustafaoğlu[25], Angela Perri[26], Joris Peters[27,28], Mike Richards[29], Özlem Sarıtaş[30], Pontus Skoglund[31], Rhiannon E Stevens[32], Chris Stringer[3], Kristina Tabbada[2], Helen M Talbot[5], Laura G Van der Sluis[18,19], Silvia M Bello[3], Vesna Dimitrijevic[33], Louise Martin[32], Marjan Mashkour[11,13], Simon A Parfitt[32,3], Sonja Vukovic[33], Selina Brace[3], Oliver E Craig[5], Prof. Dr. Douglas Baird[24], Sophy Charlton[5], Greger Larson[2], Ian Barnes[3], Laurent AF Frantz[1,34]

[1]Palaeogenomics Group, Institute of Palaeoanatomy, Domestication Research and the History of Veterinary Medicine, Ludwig Maximilian University of Munich, München, Germany, , , [2]Palaeogenomics and Bio-Archaeology Research Network, School of Archaeology, University of Oxford, Oxford, UK, , , [3]Centre for Human Evolution Research, Natural History Museum, London, UK, , , [4]Department of Biological Sciences, Middle East Technical University, Ankara, Türkiye, , , [5]BioArCh, Department of Archaeology, University of York, York, UK, , , [6]Richard Gilder Graduate School, American Museum of Natural History, New York, USA, , , [7]Department of Anthropology, National Museum of Natural History, Smithsonian Institution, Washington DC, USA, , , [8]Yong Loo Lin School of Medicine, National University of Singapore, Singapore & Institute for Human Development and Potential, A*STAR, , , [9]Department of Genetics, Evolution and Environment, University College London, London, UK, , , [10]School of Biological Sciences, University of East Anglia, UK, , , [11]BioArchéologie, Interactions Sociétés Environnements, Muséum National d'Histoire Naturelle, Paris, France, , , [12]Histoire Naturelle des Humanités Préhistoriques, Muséum National d'Histoire Naturelle, Paris, France, , , [13]Bioarchaeology Laboratory, Central Laboratory, University of Tehran, Iran, , , [14]University of Georgia, Athens, Georgia, USA, , , [15]Paleolithic Department, National Museum of Iran, Tehran, Iran, , , [16]Dipartimento di Biologia Ambientale, SAPIENZA Università di Roma, Rome, Italy, , , [17]Section for Molecular Ecology and Evolution, Globe Institute, University of Copenhagen, Copenhagen, Denmark, , , [18]Department of Evolutionary Anthropology, Faculty of Life Sciences, University of Vienna, Vienna, Austria , , , [19]Human Evolution and Archaeological Sciences (HEAS) Network, University of Vienna, Vienna, Austria , , , [20]Centre de Recherche sur la Biodiversité et l'Environnement, Université de Toulouse, Toulouse INP, CNRS, IRD, CRBE, Toulouse, France, , , [21]Centre for Palaeogenetics, Stockholm University, Stockholm, Sweden, , , [22]Department of Geological Sciences, Stockholm University, Stockholm, Sweden, , , [23]Department of Archaeology and Heritage Studies, Aarhus University, Aarhus, Denmark, , , [24]Department of Archaeology, Classics and Egyptology, University of Liverpool, UK, , , [25]Haci Bayram Veli University, Ankara, Türkiye, , , [26]Chronicle Heritage, Phoenix, Arizona, USA, , , [27]Institute of Palaeoanatomy, Domestication Research and the History of Veterinary Medicine, Ludwig-Maximilan-University Munich, München, Germany, , , [28]SNSB-State Collection of Palaeoanatomy Munich, München, Germany, , , [29]Simon Fraser University, Burnaby, Canada, , , [30]Department of Prehistory, Division of Archaeology, Faculty of Arts and Sciences, Hitit University, Çorum, Türkiye, , , [31]Ancient Genomics Laboratory, The Francis Crick Institute, London, UK, , , [32]Institute of Archaeology, University College London, London, UK, , , [33]Laboratory for Bioarchaeology, Department of Archaeology, University of Belgrade - Faculty of Philosophy, Serbia, , , [34]School of Biological and Behavioural Sciences, Queen Mary University of London, London, UK, ,

Archaeological evidence suggests that dogs diverged from wolves during the Palaeolithic, over 15,000 years ago. The earliest unequivocal genetic evidence, however, is associated with dog remains from Mesolithic archaeological contexts (~10,900 years ago). We generated both nuclear and mitochondrial genomes from canid remains at Pınarbaşı in Türkiye (~15,800 years ago) and Gough's Cave in the British Isles (~14,300 years ago), as well as from dogs excavated from two Mesolithic sites in Serbia (Padina 11,500-7,900 years ago, and Vlasac ~8,900 years ago). Our analyses indicate that a genetically homogeneous dog population was already widely distributed across Europe and Anatolia during the Late Upper Palaeolithic (by at-least 14,300 years ago). This finding suggests that dogs were

exchanged amongst genetically and culturally distinct Western Eurasian Late Palaeolithic human populations, namely the Magdalenian, Epigravettian, and Anatolian Hunter-Gatherers. Lastly, we identify a major influx of Eastern Eurasian dog ancestry during the Mesolithic, concomitant with the movement of Eastern Hunter-Gatherer populations into Europe, which led to the establishment of the primary ancestry characteristics that define European dog populations today.

[B272] Metagenomics studies of Stone Age anthropogenic birch bark tar from Estonia

Madeleine Thirolle[1], Kadri Irdt[1], Theis Jensen[4], Dr. Toni De-Dios[1], Rémi Barbieri[1], Irina Khrustaleva[1,2], Aivar Kriiska[2], Helja Kabral[1], Hannes Schroeder[4], Ester Oras[2,3], Mait Metspalu[1]
[1]Institute of Genomics, University Of Tartu, Tartu, Estonia, [2]Institute of History and Archaeology, University of Tartu, Tartu, Estonia, [3]Institute of Chemistry, University of Tartu, Tartu, Estonia, [4]Globe Institute, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark
Birch bark tar has emerged as a unique and valuable source of ancient biomolecules. It was produced through the dry distillation of birch bark (Betula sp.). During the Stone, Bronze and Early Iron Age, this material was widely used as an adhesive for tools and pottery and was often processed by chewing. Remarkably, birch tar chunks surviving from thousands of years ago have been shown to preserve ancient human and oral microbiome DNA (aDNA), addressing the scarcity of human remains in Baltic regions for the Prehistoric period.

Our purpose is to investigate the oral microbiome, the immune responses and diet to deepen our understanding of the health conditions and shed light on the lifestyle of hunter-gatherer communities.
Using metagenomics, we aim to explore the diversity and prevalence of the human oral microbiome (HOM) and understand if the commensal species found are associated with pathogenic state or diseases. As well as investigating eukaryote DNA to learn more about the environment and food consumption.
Our analyses were conducted on twenty-one birch tar lumps from ten settlement sites of Estonia, dated from 9th to the 1st Millennium BCE.

We revealed an insight of the bacterial sources and the microbial diversity preserved in the samples. The investigation done on the eukaryote DNA showed the presence of several mitochondrial animal DNA. We verified two of them, a pike and a bear, whose mitochondrial DNA was better preserved, by phylogenetic analysis.

[B273] Reducing read loss in competitive mapping of ancient DNA

Mads V Hartmann[1], Kirstine Tersbøl Melsen[1], Deon de Jager[2], Michael V. Westbury[1]
[1]Technical University Of Denmark, , Denmark, [2]University of Copenhagen, ,

Ancient DNA datasets are typically characterised by low endogenous content and substantial contamination from environmental and modern sources. Competitive mapping against multiple reference genomes is widely used to reduce contamination, but standard implementations discard reads that map to more than one locus or reference. As a result, genuine endogenous reads originating from conserved or homologous regions are often lost, reducing usable data and introducing coverage biases.

Using simulated ancient DNA datasets with realistic fragmentation and damage patterns, we quantify read loss during competitive mapping and show that a substantial fraction of endogenous reads are removed due to sequence conservation rather than true contamination. These observations motivate an alternative to standard competitive mapping strategies that treat ambiguous alignments as uninformative.

To address this limitation, we present a computational approach that retains multi-mapping reads during competitive mapping and assigns their most likely origin based on relative alignment similarity rather than mapping quality thresholds alone. By resolving ambiguous mappings instead of discarding them, the method preserves endogenous signal while still excluding contaminant reads. Applied to simulated data, this approach substantially increases recovered read counts and improves coverage uniformity without inflating false positives. The framework is broadly applicable to ancient DNA analyses where competitive mapping is required and maximising retention of authentic endogenous data is critical.

[B274] Describing early sheep domestic genetic diversity using ancient genomes from Asikli Höyuk, in Central Anatolia.

Pedro Morell Miranda[1,2], Dr. Edson Sandoval-Castellanos[3], Sheila Geiger[3], Dr. Stefan Krebs[5], Professor Laurent Frantz[2], Professor Joris Peters[3,6], Professor Ivica Medugorac[4]
[1]Center For Paleogenetics - Stockholm University, Stockholm, Sweden, [2]Paleogenomics group - Ludwig-Maximilians University of Munich, Munich, Germany, [3]Paleoanatomy group - Ludwig-Maximilians University of Munich, Munich, Germany, [4]Population Genetics group - Ludwig-Maximilians University of Munich , Munich, Germany, [5]Gene Center, Laboratory for Functional Genome Analysis, Ludwig-Maximilians University of Munich, Munich, Germany, [6]Bavarian State Collection for Palaeoanatomy, Munich, Germany

Sheep was amongst the earliest livestock species to be domesticated in the Fertile Crescent around 12.000-10.000 years ago. However, the region, process and demographic changes that led to this domestication are poorly understood. Recent genetic studies suggested that this domestication may have started somewhere in Anatolia from a wild population significantly different from the extant Anatolian mouflon.

To disentangle the history of early domestic sheep, we sequenced 72 high quality ancient genomes from Aşıklı Höyük, a site reflecting the earliest stages of sheep management and domestication, in addition to other Early Pre-Pottery Neolithic (PPN) sites in southwest Asia, including Göbekli Tepe and Nevali Cori, both early megalithic sites in Anatolia built by communities of foragers. The pooling of these ancient genomes along publicly available (modern and ancient) genomes enables us to elucidate the genetic structure of pre-domestic and early domestic sheep populations in Neolithic southwest Asia. By then focusing on a vertical-sampling of an Early Neolithic site with over a millennium of occupation we were able to describe the transition from hunting to herding this species, and we were then able to describe the relationship of these early domestic sheep with later Early Neolithic sheep from other regions.

[B275] Predicting fitness effects of genetic variants by protein language models: experimental validation and calibration in plant species

Guillaume Ramstein[1], Natasha Johansen[1], Anko Blaakmeer[2], Yannick El Khoury[3], Camous Moslemi[1]
[1]Aarhus University, , Denmark, [2]Carlsberg Research Laboratory, , Denmark, [3]University of Copenhagen, , Denmark

Protein language models (pLMs) are currently among the most established AI models for predicting variant effects. However, few studies have compared observed versus predicted effects, especially in plants. A critical step for real-world implementation (e.g., in plant breeding) is calibrating pLM scores against the fitness parameters used in evolutionary genetics, specifically the (scaled) selection coefficient of point mutations.

In this presentation, I explore the relationship between scores from ESM (a foundational pLM) and fitness effects in model grass species. I will discuss three key findings: (1) the association between ESM scores and allele purging in Brachypodium mutant populations; (2) the distribution of fitness effects in natural sorghum populations; and (3) the direct validation of candidate variants via targeted mutagenesis in sorghum.

Our results suggest that ESM predicts the fitness effects of missense variants more accurately than standard tools based on multiple sequence alignment (SIFT) or genomic language modeling (PlantCaduceus). Furthermore, we find that ESM scores are well-calibrated to selection coefficients, showing a significant enrichment for deleterious effects among variants with negative ESM scores. Finally, we identify a log-linear mapping between ESM scores and variant fitness.

Our findings highlight both the promise and the current limitations of pLMs in plant genetics. Additionally, our work provides new datasets for validating variant effect prediction tools, offering a much-needed benchmarking resource for the plant science community.

[B276] Paying for expression: the adaptive landscape of gene regulation in the arabinose operon

Anuradha Mukherjee[1], Gopinath Chattopadhyay[1], Prof.Dr. Andreas Wagner[1]
[1]University Of Zurich, Zurich, Switzerland

Gene regulation evolves under competing selective pressures of the metabolic cost of gene expression and the fitness benefit it provides in permissive environments. Although this trade-off is central to evolutionary theory, empirical adaptive landscapes of regulatory sequences remain poorly characterized in native chromosomal contexts, where regulatory variation directly couples to organismal fitness.

Herein, we investigate the adaptive landscape of prokaryotic gene regulation using the chromosomally encoded arabinose operon of Escherichia coli. Focusing on the AraC-AraI2 regulatory interaction, we generate large CRISPR-mediated libraries of araI2 binding-site variants at the native chromosomal locus and quantify their evolutionary dynamics under controlled ecological conditions that modulate operon activation. By tracking variant frequencies during population growth using deep sequencing, we infer relative fitness effects arising from differences in AraC binding strength, capturing the combined consequences of expression cost and metabolic benefit.

We further examine how regulatory context, specifically negative regulation and operon level crosstalk with the xylose system, shapes fitness landscapes generated by cis-regulatory variation at araI2. This approach reveals how the same regulatory variants can experience distinct selective pressures depending on network context and environmental conditions.

Together, this work provides a minimally invasive, population-level map of how cis-regulatory variation and regulatory context jointly determine fitness in bacteria. More broadly, it establishes a general strategy for linking molecular variation in gene regulation to evolutionary outcomes under natural selection.

[B277] Phylogenetic variation in intrinsic antimicrobial drug susceptibility constrains resistance evolution

Camille Bédard[1,2,3,4,5,6], Isabelle Gagnon-Arsenault[1,2,3,4,5,6], Mégane Bernier[1,2,3,4,5,6], Dr. Christian Landry[1,2,3,4,5,6]

[1]Département de Biochimie, de Microbiologie et de Bio-informatique, Université Laval, Québec, Canada, [2]Institut de Biologie Intégrative et des Systèmes (IBIS), Université Laval, Québec, Canada, [3]Le regroupement québécois de recherche sur la fonction, l'ingénierie et les applications des protéines (PROTEO), Québec, Canada, [4]Centre de Recherche sur les Données Massives (CRDM), Québec, Canada, [5]Centre de Recherche en Infectiologie (CRI), Québec, Canada, [6]Institut Intelligence et Données (IID), Québec, Canada

The evolution of antimicrobial resistance threatens treatment efficiency and disease control. Azole antifungals inhibit Erg11, an essential and highly conserved enzyme in fungi. Amino acid substitutions in Erg11 are a common mechanism of resistance. Comparative analyses from the literature indicate that some Erg11 mutations confer azole resistance across several distantly related fungi. Yet, many additional resistance mutations appear to be species specific, suggesting constraints on resistance evolution that remain uncharacterized.

To address this, we first investigated how intrinsic susceptibility to azoles varies among yeasts. We quantified the intrinsic susceptibility of Erg11 from 30 Saccharomycotina. Despite strong sequence conservation, substantial interspecific differences with a strong phylogenetic signal were observed. To help infer the evolutionary origins of these differences, we also reconstructed and experimentally phenotyped eight ancestral Erg11 sequences.

We then examined whether intrinsic susceptibility influences the evolution of acquired resistance. We systematically mutated Erg11 of both the human pathogen Candida albicans and S. cerevisiae, which diverged over 200 million years ago and differ in intrinsic susceptibility to azoles. While we identified ~1,000 resistance-conferring variants per azole tested for C. albicans, S. cerevisiae exhibited approximately 50% fewer resistance mutations. However, ~80% of the smaller set of resistance-conferring mutations in S. cerevisiae are transferable to C. albicans.

Together, these findings show that the mutational landscape of azole resistance is constrained by intrinsic drug susceptibility. Ongoing work aims to identify the structural changes responsible for reduced susceptibility, define how they shape resistance evolution, and build predictive models of resistance mutations in Erg11 across fungi.

[B278] Exploring Antifungal Resistance Through a Heterologous Expression System to Study Convergent Evolution and Fitness Landscapes

David F. Jordan[1,2,3,4,5,6], Mathilde Roland-Caverivière[1,2,3,4,5,6], Camille Bédard[1,2,3,4,5,6], Alexandre K. Dubé[1], Isabelle Gagnon-Arsenault[1,2,3,4,5,6], Dr. Christian Landry[1,2,3,4,5,6]
[1]Département de Biochimie, microbiologie et bio-informatique, Université Laval, Québec, Canada, [2]Institut de Biologie Intégrative et des Systèmes (IBIS), Québec, Canada, [3]PROTEO-Regroupement Québécois de Recherche sur la Fonction, l'Ingénierie et les Applications des Protéines, Québec, Canada, [4]Centre de Recherche en Données Massives de l'Université Laval, Université Laval, Québec, Canada, [5]Centre de recherche en infectiologie, Université Laval, Québec, Canada, [6]Institut intelligence et données, Université Laval, Québec, Canada

Antifungal resistance often emerges through mutations in ergosterol synthesis genes. In particular, mutations in ERG11 in various species confer resistance to the widely-used azoles. Mutations in ERG3 and ERG6, though less common, have been found in laboratory experiments to confer resistance to multiple classes of antifungal drugs across many species.

To examine whether fungal pathogens converge on the same resistance mutations, we built a heterologous expression system that allows resistance testing of various orthologs of ERG3, ERG6 and ERG11 in budding yeast. From this, convergent evolution of resistance mechanisms was observed, as mutations in conserved residues led to resistance phenotypes, except in highly diverged mold ERG11 orthologs.

This system is currently being used to study the Candida auris ortholog of ERG3. We are constructing a mutant library of every possible amino acid substitution in the protein. Using competitive growth assays in multiple conditions, we can trace a portrait of antifungal resistance emergence through ERG3 mutations. We hope to better understand why certain evolutionary paths leading to resistance are followed or not. One important question is why resistance more often appears through ERG11 mutations, while ERG3 mutations seem to be disfavored. By exploring the genotype-phenotype maps of both resistance to multiple antifungals and fitness cost, we hope to elucidate this question, along with providing explanatory mechanisms at the protein level. By better understanding the costs and benefits of different mutations, we can potentially better guide future antifungal drug use, as well as inform the future design of ERG3-targeting drugs.

[B279] Fitness landscape estimation in the wild: genotypes and phenotypes underlying reproductive success in an Antirrhinum majus flower colour hybrid zone

Hilde Schneemann[1], Sean Stankowski[2], Prof. Nicholas H Barton[1], David Field[3]
[1]Institute Of Science And Technology Austria, Klosterneuburg, Austria, [2]University College London, London, United Kingdom, [3]Macquarie University, Sydney, Australia

Fitness landscape estimation requires fitness estimates, which are challenging to obtain in wild populations, especially for plants producing thousands of widely dispersed seeds. I present such a fitness landscape – connecting genotypes to flower colour phenotypes and fitness – based on the long-term study of a natural flower colour hybrid zone between Antirrhinum majus var. striatum and A. majus var. pseudomajus in the Spanish Pyrenees. Fitness varies across space, time, flower colour, and multilocus genotypes, generating a dynamic genotype-phenotype-fitness map mediated by pollinator visitation. I compare the empirical fitness landscape estimates to verify assumptions on ruggedness and dimensionality that differentiate theoretical fitness landscape models.

[B280] Landscape or seascape? Mapping an empirical fitness landscape in different genetic and environmental backgrounds

Ilya Carey[1], Prof.Dr. Andreas Wagner[1]
[1]Department of Evolutionary Biology and Environmental Studies, University Of Zurich, , Switzerland
Fitness landscapes provide a powerful framework for understanding adaptive evolution, yet empirical studies remain limited in scale and scope. In particular, few studies have mapped combinatorically complete protein fitness landscapes in vivo, and little is known about how landscape topography changes across genetic and environmental contexts. Such changes could profoundly affect peak accessibility, ruggedness, and the predictability of evolutionary trajectories, with important implications for antimicrobial resistance evolution.

Here, I map a combinatorially complete fitness landscape for a 9-bp region of the chromosomal gyrA gene in Escherichia coli, which encodes the GyrA subunit of DNA gyrase, the target of the widely used fluoroquinolone antibiotics. Using CRISPR-Cas9 gene editing, I am generating a variant library of 262,144 unique genotypes and quantifying their fitness under sublethal antibiotic exposure via bulk competition experiments and deep sequencing.

To determine how landscape topography depends on context, I am systematically varying (i) environment—by altering Mg^{2+} concentration and antibiotic identity (ciprofloxacin versus gatifloxacin)—and (ii) genetic background—by introducing resistance-associated mutations in parC. In total, I will construct eight fitness landscapes spanning combinations of genetic and environmental conditions.

By analysing peak accessibility, epistasis, ruggedness, and simulated adaptive walks, I will test whether fitness landscapes are stable across contexts or instead shift substantially, reshaping evolutionary trajectories. These results will help clarify how genetic and environmental variables interact to influence adaptive evolution and will improve our understanding of the dynamics and predictability of antibiotic resistance evolution. Ultimately, this research could help determine whether fitness landscapes are rather fitness seascapes.

[B281] Pleiotropic stabilizing selection shapes genetic architecture of complex traits

Joshua Schraiber[1], Matt Pennell[2], Michael Edge[1]
[1]University Of Southern California, Los Angeles, United States, [2]Cornell University, Ithaca, United States

Decades of GWAS have revealed that many complex traits in humans are highly polygenic, shaped by many loci of very small effect. The genetic architecture of these traits, that is, the genome-wide distribution of causal variants, their effect sizes, and their allele frequencies, is shaped by the combined action of evolution forces, including mutation, natural selection, and genetic drift. An understanding of how these forces shape genetic architectures crucial to the application of standard tools in statistical genetics. However, the canonical methods rely on a model of evolution that implicitly assumes no drift and very strong selection. Moreover, there is increasing evidence of a substantial role of pleiotropy in shaping the genetic architecture of traits. Here, we present theoretical and empirical results demonstrating the importance of pleiotropic stabilizing selection in determining genetic architecture. First, we find that, compared to the single trait case, the genetic variance is distributed among rarer alleles in the pleiotropic case. Nonetheless, among causal variants, effect sizes are unimpacted by pleiotropy. However, when causal variants are sparse and not precisely identified, as is common in statistical genetics, the impact of pleiotropy is visible. In that case, high pleiotropy limits exist which can be exploited to easily model effect sizes that span orders of magnitude. By applying this model in a framework analogous to LD-score regression, we find substantial evidence for pleiotropic stabilizing selection shaping the genetic architecture of human traits.

[B282] Molecular evolution on an adaptive seascape in fluctuating environments

Laetitia Britschgi[1], Prof.Dr. Andreas Wagner[1,2,3]
[1]University of Zurich, Zurich, Switzerland, [2]The Santa Fe Institute, Santa Fe, USA, [3]Swiss Institute of Bioinformatics, Lausanne, Switzerland

Fitness landscapes are mappings from genotypes to fixed fitness values. Fitness seascapes are an extension of this framework. They incorporate fluctuating environments in which changing selective pressures modulate the fitness of genotypes. Such environmental change can transform a landscape and impose novel evolutionary opportunities and constraints. Recent advances in biotechnology now permit the empirical mapping of hundreds of thousands of genotypes connected by single mutational steps, enabling the study of evolutionary processes at unprecedented molecular resolution. We leverage empirical fitness measurements from a combinatorially complete fitness seascape spanning all genotypic variants of a nine–base-pair region of the E. coli folA gene, encoding the active site of dihydrofolate reductase (DHFR), under both an antibiotic-treated and untreated condition. Using evolutionary simulations, we examine the effects of fluctuating environments on the evolutionary dynamics of antibiotic susceptibility and resistance in DHFR. We will summarize our findings on how environmental change influences genotype fitness and population demography, with respect to both the initial population and the environmental history experienced by the population. To this end, we systematically vary environmental change rates—from rapid, generation-to-generation fluctuations to slow transitions allowing fixation of fitness peaks—and examine stochastic environmental variation with differing noise spectra. We will investigate whether populations evolving under fluctuating environments attain higher fitness than those evolving under static conditions, as predicted by theoretical studies. Further, we will characterize how population composition varies with environmental fluctuations, ranging from fixation on environment-specific fitness peaks to occupation of general peaks that persist across environments.

[B283] Probability and Distribution of Epistatic Interactions

Luka Biočanin[1], Dr. Snøfrid April Kleppe[1], Dr. Ilkka Kronholm[1]
[1]University Of Jyväskylä, , Finland

Different individuals can experience varied effects from the same mutation. An essential factor contributing to this phenomenon lies in the dependence of a mutation's outcome on the genetic context within which it appears. This phenomenon is known as epistasis. Epistatic interactions have proven to be significant in certain facets of evolution, such as reproductive isolation as well as recombination and speciation. However, its role in adaptation remains disputed and there is a widespread consensus that epistatic variance lacks evolutionary influence. Despite this, increasing evidence suggests that epistasis at the level of gene interactions contributes to additive variance, but how important this is depends on the distribution of epistatic effects. In this research we used mutation-accumulation (MA) lines of the filamentous fungus Neurospora crassa generated in a previous experiment, and performed half-dialelic cross between them. We measured their growth rate as the key fitness component and, genotyped the known mutations that occurred during the MA experiment in the offspring to assess epistatic interactions. Our goal is to calculate the probability that two mutations exhibit epistatic interactions (P_ε), as well as the mean distribution of epistatic effects ($\bar{\varepsilon}$).

[B284] A Mutation–Selection Codon Model with Stochastic Tunneling for Multi-Nucleotide Substitutions

Oliver Lyon[1], Bianca DeSanctis[2], Dr. Jason A.P. de Koning[1]
[1]University Of Calgary, Calgary, Canada, [2]University of California Santa Cruz, Santa Cruz, United States of America

Models of molecular evolution are often mis-specified, limiting our ability to correctly interpret the causes of observed sequence differences. Several lines of evidence indicate that multi-nucleotide substitutions: a) are poorly explained by standard codon models; b) are common enough that codon models that include them ("empirical" models) fit real data better than models restricted to serial single-nucleotide substitutions; and c) have been proposed as a pervasive source of false positives in scans for positive selection. To disentangle these observations, we develop a novel mutation-selection codon substitution model that accounts for population-level processes that allow multi-nucleotide substitutions without requiring multi-nucleotide mutations. In particular, through the analysis of transient dynamics of extinction sojourns for an individually deleterious variant, we model the rate at which rescue mutations may occur. In this scenario, both mutations can then go to fixation together-a process known as stochastic tunneling. This formulation retains tractable phylogenetic likelihood computations by numerically integrating diffusion approximations for the time-integrated mutational target size for rescue mutations, without additional parameters beyond a standard mutation–selection model. Using this framework, we show that intracodon stochastic tunneling increases the expected rate of fitness-valley crossing, and that ignoring tunneling can bias inference of evolutionary parameters relevant to rugged, multi-peaked fitness landscapes. We apply this framework to explore the relative contributions of complex mutational mechanisms and population-level processes to multi-nucleotide substitutions observed in phylogenetic data, with the goal of clarifying when such substitutions constitute evidence of positive Darwinian selection and when they do not.

[B285] Employing Pichia display to characterize rapidly evolving reproductive proteins in the marine mollusk abalone

Trenton Winters[1], Damien Wilburn[1]
[1]The Ohio State University Department of Chemistry and Biochemistry, Columbus, United States
The extraordinarily rapid evolution of animal reproductive proteins confounds our understanding of the molecular mechanisms that mediate fertilization. Animal oocytes are typically surrounded by a specialized matrix, the zona pellucida (ZP), that restricts access to the egg plasma membrane and regulates the rate of fertilization. The ZP is a meshwork of glycoproteins containing one or more ZP-N domains that evolve at different rates. In the marine mollusk abalone, sperm dissolve the ZP by secreting Lysin, a small, 134-residue protein that recognizes ZP-N domains with varying affinities and species-specificities using a common, quasi-ordered binding interface. We hypothesize that Lysin has evolved to maintain affinity for conspecific egg proteins at the cost of reduced affinity for heterospecific proteins from closely related species. To evaluate this hypothesis, we developed a yeast display system to express Lysin from monophyletic Pacific abalone (8 species) on the surface of yeast (Pichia) and functionally characterize their affinity for recombinantly expressed ZP-N domains from the same taxa. Fluorescence Assisted Cell Sorting can bin yeast by Lysin expression levels and by ZP-N binding affinity via tagging the ZP-N domains and displayed Lysin with orthogonal fluorophores, with Nanopore long-read sequencing identifying and counting Lysin variants within bins for the estimation of variance coefficients. Deep mutational scans on the ancestral state of Lysin with multiple ZP-N variants further characterize the Lysin sequence space beyond natural variation. Mapping the fitness landscape of Lysin will deepen our understanding of the rapid, adaptive evolution between gamete recognition proteins and mechanisms of fertilization.

[B286] A simple test demonstrates that many prokaryotic accessory genes are adaptive.

Adam Eyre-Walker[1], Dr Ying Eyre-Walker[1], Dr Cara Conradsen[2], Dr Michiel Vos[2]
[1]University Of Sussex, , United Kingdom, [2]University of Exeter, , United Kingdon
Bacterial pangenomes often contain thousands of genes that are not present in all strains, the so-called accessory genes. Whether these are adaptive, neutral or deleterious remains contentious. Here we introduce a simple test to differentiate between these possibilities and apply the test to the accessory genes from the pangenomes of 500 Escherichia coli and Staphylococcus aureus strains. These bacteria are highly divergent and have very different pangenomes in terms of their size. We find evidence that at least 70% of all the accessory genes are adaptive in both species in the sample we have analysed. This is a substantial increase on previous estimates.

[B287] Bringing clownfishes into the pangenome era: what more can we learn about their diversification ?

Anna Marcionetti[1], Pr. Nicolas Salamin[1]
[1]University of Lausanne, , Switzerland

Clownfishes (genus Amphiprion) comprise 28 species that rapidly diversified following the evolution of mutualism with sea anemones. Host specialization is considered the primary driver of this diversification, but community composition and interspecific interactions also appear to shape patterns of divergence among clownfishes. Moreover, the genus exhibits highly variable geographic distributions, suggesting the existence of additional cryptic species, particularly within widespread taxa such as Amphiprion clarkii.

Recent comparative and population genomic studies have started to uncover the genomic mechanisms underlying this diversification. Genomic structural variations (SVs) have also been observed within clownfishes, though the extent to which these SVs are fixed within species remains unclear. Growing evidence indicates that some SVs segregate within species - for example, a large inversion on chromosome 18 appears to be polymorphic in A. clarkii. However, the prevalence and functional significance of such SVs in clownfish diversification remain poorly understood, partly because traditional linear-reference approaches fail to capture the full complexity of these genomic rearrangements.

Here, we aim to characterize SV diversity within and among clownfish species using a pangenomic framework. Leveraging the extensive genomic resources now available - including high-quality reference genomes and population-level whole-genome sequencing data - we seek to construct a comprehensive pangenome for the group. This approach will enable a more complete reconstruction of clownfish evolutionary history and provide new insights into the roles of structural variation in adaptation, diversification, and population connectivity.

[B288] Pangenome-based genotyping of structural variants in medical cohorts

Chiara Paleni[1], Davide Bolognini[1], Andrea Guarracino[2,3], Thomas Sumner Dudley[1], Alessandro Raveane[1], Federica Santonastaso[1], Edoardo Giacopuzzi[1], Licia Iacoviello[4,5], Peter H. Sudmant[6], Erik Garrison[2], Nicole Soranzo[1,7,8]

[1]Human Technopole, Milan, Italy, [2]Department of Genetics, Genomics and Informatics, University of Tennessee Health Science Center, Memphis, United Stated of America, [3]Bioinnovation and Genome Sciences, The Translational Genomics Research Institute (TGen), Phoenix, United Stated of America, [4]Research Unit of Epidemiology and Prevention, IRCCS Neuromed, Pozzilli, Italy, [5]Department of Medicine and Surgery, LUM University, Casamassima, Bari, Italy, [6]Department of Integrative Biology, University of California Berkeley, Berkeley, United Stated of America, [7]University of Cambridge, Cambridge, United Kingdom, [8]Wellcome Sanger Institute, Genome Campus, Hinxton, United Kingdom

Pangenome graphs built from long-read assemblies enable comprehensive variant discovery, particularly at highly complex and structurally variable loci such as the HLA. However, most large-scale biobanks rely on short-read sequencing. Moreover, translating pangenome-based genotyping to population and medical genomics applications, such as linking genotypes to molecular traits, remains challenging. We present an application of pangenomes for structural variant (SV) genotyping with short-read data in a Southern-Italian medical cohort, representative of an understudied Mediterranean population.

Our method, COSIGT, assigns haplotype sequences from pangenome graphs to short-read samples by comparing read coverage and haplotype copy-number vectors. We extended COSIGT to convert haplotype assignments into structural variant genotypes by decomposing the pangenome graph at variable regions, enabling downstream analysis with standard variant analysis tools. We applied this approach to the Moli-sani cohort, comprised of >6,000 short-read samples. Using the HPRCy2 pangenome reference, we benchmarked COSIGT on the hyper-variable HLA genes. We then genotyped structural variants across challenging, medically relevant genes (CMRGs).

For HLA typing, COSIGT shows comparable accuracy to specifically designed tools despite being a general-purpose approach. Over 47 CMRGs, we can genotype hundreds of structural variants in the Moli-sani cohort, demonstrating scalability to population-level datasets. Additionally, only 38% of SVs are strongly tagged by neighbouring SNPs, demonstrating that this approach can potentially discover new associations and help explain known ones.

Pangenome-based genotyping with COSIGT bridges the gap between pangenome resources and large-scale medical cohorts, enabling structural variant analysis and association studies at complex loci using widely available short-read sequencing data.

[B289] Reference-free pangenomes illuminate genomic diversity in African Anopheles malaria vectors

Clothilde Chenal[1], Francis Isidore Garcia Totanes[1], Diego Ayala[2], Mara Lawniczak[1]
[1]Wellcome Sanger Institute, Cambridge, United Kingdom, [2]MIVEGEC, IRD, CNRS, Univ. Montpellier, Montpellier, France

Understanding local adaptation and speciation in major malaria vectors requires accurate representation of genomic variation across populations and species. In Anopheles mosquitoes, analyses relying on single linear reference genomes fail to fully capture structural variation, presence–absence gene polymorphisms, and deeply divergent haplotypes, and thus introduce reference bias into population-genetic inference. Here, we describe the development of a reference-free pangenome framework for four major African malaria vectors—Anopheles gambiae, Anopheles coluzzii, Anopheles arabiensis, and Anopheles funestus—based on individuals collected across Africa. Using a combination of graph-based and assembly-driven pangenome approaches, we aim to capture both shared and species-specific genomic diversity, with a focus on variation potentially shaped by ecological gradients and vector control pressures. Ongoing comparative analyses across species are being used to assess how alternative genomic representations influence estimates of genetic diversity, population differentiation, and gene flow, particularly in genomic regions associated with insecticide resistance, ecological adaptation, and reproductive isolation. We further evaluate the extent to which reference-free pangenomes may improve read-mapping across populations and reduce biases in allele-frequency estimation, thereby facilitating the detection of locally adaptive and structurally complex loci. This work explores how evolution-oriented pangenome designs, coupled with broad continental sampling, can support more robust population-genetic inference. More broadly, it highlights the potential of pangenome approaches to advance studies of adaptation and speciation in major public-health relevance.

[B290] Structural variants in Malawi cichlids radiation.

Ilia Artiushin[1], Hannes Svardal
[1]UAntwerpen, Antwerpen, Belgium

Cichlid species flock is one of the most prominent model systems in microevolution. A trove of genomic data is collected and analyzed in the last decade leading to discovery of genomic mechanisms of speciation and local adaptation. At the same time most of the published studies are focused on SNP datasets and ignore structural variants (SV). Recent publications discovered the presence of extensive structural variation in cichlid genomes (spanning up to 30% of genome length). Some of SV classes (large-scale inversions, TEs) have gained attention of researchers previously, but majority of SV variation is still understudied.
We constructed whole-genome alignments and variation graph from 15 ONT-based genome assemblies (5 recently published, 10 newly sequenced) and used short read sequencing data on another 1800 samples (400 newly sequenced) to investigate patterns of structural variation in Malawi cichlids species flock.

[B291] Constructing pangenome gene graphs with hundreds of thousands of bacterial genomes

Jackie Toussaint[1,2], Dr. Samuel Horsfield[1], Dr. Zamin Iqbal[3], Dr. Neil MacAlasdair[1], Leonie Lorenz[1,2], Dr. Gerry Tonkin-Hill[4], Dr. John Lees[1]
[1]EMBL-EBI, Cambridge, United Kingdom, [2]University of Cambridge, Cambridge, United Kingdom, [3]University of Bath, Bath, United Kingdom, [4]University of Melbourne, Melbourne, Australia

The availability of hundreds of thousands of genomes for individual bacterial species provides an unprecedented opportunity to obtain a comprehensive view of population-wide diversity, represented by pangenomes. Pangenome gene graphs, a network-based representation of the presence and order of orthologous genes in a population, enable a number of unique functions not available from conventional sequence clustering methods such as MMseqs2 or CDHit, such as distinguishing paralogs, removal of erroneous genes, and detection of recombination or horizontal gene transfer events. However, existing bacterial pan-genome gene graph generation tools are not designed for large-scale analyses and become infeasible to run with thousands of isolates.

Here, we present “Pangenomerge”, a method for fast, accurate and reproducible bacterial pan-genome gene graph construction, with which we generate species-wide pan-genome gene graphs containing hundreds of thousands of genomes. Pangenomerge identifies strains within large datasets using PopPUNK, before generating individual strain-level pangenome gene graphs using ggCaller and Panaroo. Pangenomerge then iteratively merges these strain-level pangenomes using MMSeqs2, whereby genes are rapidly mapped between graphs and then scored by synteny to select and merge orthologous genes into a single complete pangenome graph.

Using species-wide pangenome gene graphs generated by Pangenomerge, we conduct population-level analyses, including identification of core and accessory genes, recombination and horizontal gene transfer events, operon synteny within a diverse population, and gene-level selection, as well as inference of species- and gene-level phylogenetic history, demonstrating the broad utility of this method for population genomics analyses.

[B292] A pangenome approach for studying rapid adaptation in a species complex

Karim El Akoum[1], Vera van Santvoort[2], Professor Steven AJ Declerck[3], Professor Kathrin A Otte[4]
[1]University Of Hamburg, Hamburg, Germany, [2]Vera van Santvoort, Wageningen, Netherlands, [3]Steven AJ Declerck, Wageningen, Netherlands, [4]Kathrin A Otte, Hamburg, Germany

Salinization poses an ongoing threat to freshwater biodiversity, requiring rapid evolutionary responses from keystone species. This project investigates the capacity for rapid adaptation to increased salinity in three Daphnia taxa: the D. pulex complex, the D. galeata complex, and Simocephalus vetulus. Using outdoor mesocosm experiments, we assess adaptive potential both in single-species treatments and community settings to account for the influence of intra and inter-specific competition on evolutionary trajectories. A central focus of this project is the role of hybridization within the D. pulex and D. galeata complexes each as a facilitator of rapid adaptation. To address this, we employ a pangenome-based approach to detect signatures of selection. By representing the entirety of genetic material which includes the core genome shared by all individuals and the accessory genome unique to specific genotypes or hybrid crosses, a pangenome effectively eliminates reference bias, ensuring that divergent alleles or structural variations acquired through introgression are considered. This is particularly interesting in species complexes like D. pulex and D. galeata which are characterized by frequent gene flow within the complex. By utilizing a pangenome, we can account for genetic variations present among the competing genotypes therefore capturing a more complete picture of the genetic architecture underlying salt tolerance. Our findings will highlight the importance of a pangenome-based approach and clarify how standing genetic variation, hybridization, and multi-species interactions collectively shape the adaptive responses of a key functional group to increasing levels of salinity in freshwater ecosystems.

[B293] Simulating population pangenomes with complex structural variants under the coalescent using MSpangenome.

Lucien Piat[1], Sukanya Denni[1], Siegfried Dubois[2], Ludovic Duvaux[1]
[1]INRAE - Biogeco, Bordeaux, France, [2]INRIA - IRISA, Rennes, France

Structural variants (SVs), such as insertions, deletions and inversions, contribute substantially to genetic and phenotypic variation but remain understudied. The recent emergence of variation graphs provides an efficient framework to represent such variants in population genomics and pangenome analyses, capturing genomic diversity beyond single-reference approaches. However, these methods are still emerging, and genome simulations are required for benchmarking and testing evolutionary hypotheses. Existing genome-wide SV simulators generate only simple variants and rely on oversimplified evolutionary scenarios, resulting in unrealistic star-like genealogies. Conversely, coalescent-based simulators such as msprime support complex demographic histories but do not allow the simulation of complex SVs.

Here, we introduce MSpangenome, a Python-based pangenome simulation workflow. Given a demographic model and a reference sequence, msprime is used to simulate the coalescent history of recombination blocks across the genome. Each coalescent tree is then traversed lineage by lineage to chronologically introduce SVs drawn from a broad panel of variant classes allowing nested and overlapping events. This process incrementally constructs a coalescent-aware variation graph, which can be exported in Graphical Fragment Assembly (GFA) format. Genome sequences can also be generated as FASTA files.

We illustrate the utility of MSpangenome through benchmarking experiments with PGGB and Minigraph-Cactus, revealing contrasting responses to increasing genetic diversity: PGGB better preserves complex graph topologies under high diversity, whereas Minigraph-Cactus shows reduced structural resolution.

By linking coalescent data structures with bi-oriented variation graphs, MSpangenome provides a flexible framework for simulating population pangenomes under complex demographic scenarios, enabling benchmarking, method development and simulation-based inference.

[B294] Towards an unbiased characterization of genetic polymorphism

Dr Magnus Nordborg[1]
[1]Gregor Mendel Institute, Vienna, Austria
Making sense of whole-genome polymorphism data is challenging, but it is essential for achieving a complete picture of genetic variation and overcoming the biases in SNP data. We have analyzed nearly 500 long-read genomes of Arabidopsis thaliana to illustrate these issues. Genome size variation in A. thaliana is mostly due to tandem repeat regions that are difficult to assemble. However, while the rest of the genome varies little in length, it is full of structural variants, mostly due to rare transposon insertions so that the pangenome coordinate system grows rapidly with sample size. Our data provides an unprecedented view of the mobilome, massively improving our understanding for transposon activity. Finally, we show how short-read data are biased by read mapping. SNP calling is biased by the choice of reference genome, and both transcriptome and methylome profiling results are affected by mapping reads to a reference genome rather than to the genome of the assayed individual. In conclusion, whole-genome polymorphism data will greatly improve our understanding of genome evolution, and will in particular revolutionize the analysis of transposable elements.

[B295] Haplotype-resolved panNLRome in cultivated and wild bananas (Musa spp.) seen through the pangenomic lens

Marek Glombik[1], Camilla Ryan[1], Carolina Olave-Achury[1], Alex Durrant[1], Leah Catchpole[1], Naomi Irish[1], Kendall Baker[1], Karim Gharbi[1], Jose De Vega[1]
[1]Earlham Institute, Norwich, United Kingdom

Bananas (Musa spp.) represent 15% of global fruit production, with over 139 million metric tons produced in 2023. However, the majority of global banana production relies on germplasm susceptible to Fusarium wilt, caused by Fusarium oxysporum f. sp. cubense Tropical Race 4 (FocTR4). Due to the clonal nature of banana reproduction, the introduction of genetic diversity is limited to the somatic accumulation of mutations, somaclonal variation introduced via tissue culture, breeding crosses between sexual accessions, or genome editing. Variation in resistance gene sequences and copy number is therefore key to combating current and future biotic threats to banana production. Here, we sequenced and assembled the genome of ten banana lines spanning modern cultivars of diverse subgenome compositions, as well as wild accessions, displaying either susceptibility or resistance to FocTR4. We combined these data with publicly available genomes and identified orthologous nucleotide-binding leucine-rich repeat (NLR) genes across all accessions. While a large number of NLR orthogroups were shared among most lines, many line-specific NLRs were also detected. We subsequently constructed a pangenome graph and focused on NLR-containing regions to characterize their variation at the haplotype level. In total, we identified over 180 NLR node clusters encompassing nearly 4,000 NLRs across all genomes. These clusters exhibit haplotype-specific variation in NLR copy number, reflecting rapid and ongoing evolution driven by structural variation. Together, our results reveal a dynamic NLR landscape in Musa and provide a genomic framework for identifying resistance-associated haplotypes, informing future breeding strategies for durable Fusarium wilt resistance.

[B296] Independent hybridization events, followed by genomic rearrangements, shape genome evolution of polyploid A. kamchatica

Maria Vasilarou[1], Uliana Kolesnikova[1], Charles Addo Nyarko[1], Terezie Mandakova[2], Alison Scott[1], Dr. Polina Novikova[1,3]
[1]Max Planck Institute For Plant Breeding Research, Cologne, Germany, [2]CEITEC, , Czech Republic, [3]VIB-UGent Center for Plant Systems Biology, Ghent, Belgium

Allopolyploidy, a combination of hybridization and whole-genome duplication, results in instant speciation from diploid progenitors due to karyotypic differences. The fusion of two divergent genomes in a single nucleus often leads to genomic instability, with various outcomes ranging from extinction to the successful establishment of a neopolyploid. Arabidopsis kamchatica is an established allotetraploid with a complex evolutionary history, involving multiple hybridization events between A. lyrata and A. halleri. Here, we investigate the genome structure within the species, using chromosome-level assemblies from multiple populations. Extracting information from A. kamchatica's pangenome, we aim to link the genomic and population structure of the different lineages to the independent historical origins of the species. Unique genomic rearrangements were identified in different A. kamchatica genomes, involving not only homeologous exchanges between the subgenomes, but also a large non-homologous reciprocal translocation in the genome of one accession. We further validated these genomic rearrangements using cytological techniques. Moreover, experimental evaluation of hybridization between the different accessions indicates limited compatibility, as the progeny are partially sterile. Genomic instability in allopolyploids can contribute to reproductive isolation and, in some cases, promote further diversification of the species. Overall, the evolutionary history of A. kamchatica and its genomic structure appear intertwined: historical founder events have shaped the genome, which in turn can influence evolutionary trajectories by establishing barriers between lineages. Pangenome approaches provide a powerful framework to capture these dynamics.

[B297] Graph Variant Explorer: Pangenomic analysis of ancient and modern DNA

Marianne Dehasque[1], Cormac Kinsella[2,3], Torsten Günther[2]
[1]UC Santa Cruz, Santa Cruz, United States, [2]Uppsala University, Uppsala, Sweden, [3]NBIS, Uppsala , Sweden

Ancient DNA (aDNA) is a powerful tool for studying evolutionary processes across species. However, due to DNA degradation and fragmentation over time, most analyses must be restricted to single-nucleotide polymorphisms (SNPs), even though genomic structural variants (SVs) represent a much larger source of genetic diversity. Methods to detect SVs in aDNA would therefore be highly valuable. A key methodological challenge is the failure to map short, damaged reads to a reference assembly (i.e., reference bias), especially when those reads contain alternative alleles. While linear assemblies lack variant information, pangenome graphs capture the genomic variation of constituent assemblies within an efficient data structure, such that mapping sequence reads to graphs significantly improves mapping success for alternative alleles. For ultra-short aDNA reads, graph references therefore offer the possibility to detect types of historical genetic variation that are undetectable using linear references. Here, we present GRAVE (graph variant explorer), a reproducible Nextflow workflow for aligning aDNA reads to pangenome graphs, authenticating aDNA data and identifying both SNPs and SVs. The workflow produces output files compatible with standard population genomics analyses. We expect that by incorporating diverse haplotypic backgrounds during mapping, GRAVE will enhance the recovery of authentic endogenous DNA while also reducing reference bias and unlocking complex SVs for aDNA research. Although our initial application focuses on sheep domestication, leveraging ancestral haplotypes from wild and feral sheep, the pipeline is broadly applicable to other species and study systems.

[B298] K-mer counts in pooled sequencing data rapidly estimate frequencies of known alleles in a pangenome graph

Miles Roberts[1]
[1]UC Berkeley, Berkeley, United States

Pangenomic references, as opposed to single-genome references, are the new standard for population genetic analyses. However, genotyping new sequencing samples against a reference pangenome can become computationally costly as either the reference pangenome scales in size or samples scale in coverage. Genotyping known variants can be massively sped up by looking for short exact matches between sequencing reads and known variants (i.e., k-mers) using tools like BayesTyper and Pangenie. However, these and similar tools are restricted to estimating only diploid genotypes whereas many population genetic and experimental evolution datasets rely on pooling DNA from many genotypes into one sequencing library (i.e., pool-seq) to cut labor costs. Instead of diploid genotypes, pool-seq data estimates allele frequencies. We present a method for counting allele-specific k-mers in pool-seq data that can estimate frequencies of known pangenomic alleles in much less time than vg giraffe. We first tested the method on simulated population genetic data and found that allele-specific k-mer counts give similarly accurate allele frequency estimates to vg giraffe but in about 1/10th the time. Next, we investigated empirical sequencing datasets and observed that allele-specific k-mer counts in sequencing reads recapitulate allele frequencies in previously published variant sets. Overall, counts of allele-specific k-mers can quickly estimate frequencies of known alleles in a pangenome from pool-seq data, either providing preliminary results before committing to pangenome alignment or allow scaling to large pool-seq datasets.

[B299] Identifying unique variation and genetic load in Dutch cattle breeds through a pangenome

Rensco Hogers-Bijlaart[1], dr. Mirte Bosse[1,2], Dr. Chris Barratt[1,3], Dr. Annemieke Rattink[1,3], prof. Martien Groenen[1], Dr. Richard Crooijmans[1], Dr. Aniek Bouwman[1]
[1]Wageningen University & Research, Wageningen, the Netherlands, [2]Amsterdam Institute of Life and Environment (A-Life), Amsterdam, the Netherlands, [3]Centrum voor Genetische Bronnen Nederland, Wageningen, the Netherlands

Genetic variation within cattle has historically been its strength, but has turned into one of its main challenges. The focus on higher productivity in farming systems has resulted in a dwindling population of traditional breeds, which has accelerated in the last few years. This trend is particularly evident in the Netherlands, where most Dutch cattle breeds are marked as endangered, and in the last four years populations have shrunken by as much as 90 %. As such, our aim was to capture and analyse the currently still present genetic diversity in Dutch cattle breeds before it is too late. For this we used a pangenome with de novo assembled genomes of traditional Dutch cattle breeds. Genetic variations were used to investigate diversity, while genetic load was calculated using both GERP and bovine CADD scores.

Using a pangenome we were able to assess the number and type of variants that are unique and fixed within each breed, showing what genetic diversity could be lost in the near future. The genetic variants found in these breeds were used to estimate the genetic load per breed using GERP and bovine CADD scores, showing an increase in the number of deleterious mutations in the Dutch breeds compared to Holstein-Friesian populations. With these results we showcase the use of pangenomes to assess the genetic diversity and load of populations, enhancing the community's capacity to deploy results in conservation and management.

[B300] Pangenomics for De-Extinction: Towards the Great Passenger Pigeon Comeback

Simona Secomandi[1], Suziane Barcellos[1], Ben J. Novak[2], Erich D. Jarvis[1]
[1]The Rockefeller University, New York, United States, [2]Revive & Restore, Sausalito, United States

The passenger pigeon (Ectopistes migratorius) was the most abundant bird species in North America, numbering in the billions before being driven to extinction in the early 1900s. Thousands of museum specimens exist, and its closest relative, the band-tailed pigeon (Patagioenas fasciata), is still extant. Thus, the passenger pigeon is a strong candidate for de-extinction through ecological replacement, an approach that adapts a living organism to fulfill lost ecological roles using targeted phenotypic changes, thereby supporting ecosystem resilience and biodiversity. Here, we generated ancient DNA (aDNA) datasets from multiple passenger pigeon specimens and two sets of Vertebrate Genomes Project-quality, haplotype-resolved genomes. The first includes multiple band-tailed pigeons sampled across the species' geographic range, which we used to construct a pangenome graph to aid aDNA mapping. The pangenome effectively mitigated reference bias, particularly in indels, and improved variant calling relative to a single-reference approach, enabling the reconstruction of more complete pseudo-haplotype consensus genomes for the extinct species and expanding the catalogue of passenger-pigeon-fixed variants. The second set includes genomes from multiple Columbidae species that share ecological or phenotypic traits with the extinct species, providing a comparative framework to assess evolutionary signatures associated with those traits. Leveraging the completeness of the multi-species set of genomes we will be able to detect signatures of selection to generate a prioritization schema for gene editing of candidate loci. Together, these efforts represent a first step towards the restoration of the ecological role of the passenger pigeon, highlighting the potential of genomic intervention for de-extinction and conservation.

[B301] The diversity and evolutionary history of structural variation in European cattle genomes

Tobias van Elst[1], Alexander Leonard[1], Benjamin D. Rosen[2], Timothy P. L. Smith[3], Wai Yee Low[4], Hubert Pausch[1]

[1]ETH Zurich, Zurich, Switzerland, [2]U.S. Department of Agriculture, Agricultural Research Service, Animal Genomics and Improvement Laboratory, Beltsville, USA, [3]U.S. Department of Agriculture, Agricultural Research Service, U.S. Meat Animal Research Center, Clay Center, USA, [4]Davies Livestock Research Centre, School of Animal and Veterinary Sciences, University of Adelaide, Roseworthy, Australia

The diversification of cattle (Bos taurus) has been shaped by extensive admixture among breeds and with other Bos species. Although gene flow within the Bos species complex has been characterized at the level of single nucleotide variants (SNV), its impact on structural variant (SV) diversity remains poorly understood. SVs are likely to have played an important role in domestication and adaptation given their substantial impacts on genome evolution and gene expression. In this study, we used a pangenomics approach to explore the diversity and evolutionary history of SVs across European cattle breeds. We integrated 53 high-quality cattle assemblies of 27 diverse European breeds into a pangenome with Minigraph-Cactus. By mapping publicly available short read whole genome sequencing data of 1,075 samples, we genotyped ~ 120,000 SVs with vg call across 80 taurine breeds, wisent (Bison bonasus) and the extinct aurochs (Bos primigenius). The resulting SV genotypes reveal substantial diversity and breed (group) specific clustering, mirroring previous findings with SNVs. We also explored how previously hypothesized introgression events among domestic taurine cattle (European and North African), wisent and aurochs shaped the landscape of structural variation in current breeds alongside selection. We put a particular emphasis on known quantitative trait loci of economic importance. Our work provides the most comprehensive catalog of SVs in European cattle to date. It also elucidates the evolutionary history of SVs in European cattle, highlighting the roles of introgression and selection, and demonstrates how high-quality pangenome references enable leveraging short read data to analyze structural variation.

[B302] From bacteria to humans, pangenome graphs provide unified coordinates for diversity, evolution, and somatic variation

Yuxin Ning[1], Professor Franz Baumdicker[2], Professor Sven Nahnsen[1]
[1]University Of Tübingen, Quantitative Biology Center, Tübingen, Germany, [2]Justus-Liebig-Universität Gießen, Gießen, Germany

Pangenome graphs offer many benefits compared to linear reference and reduce reference bias by incorporating multiple haplotypes and genome structures. With graph-based methods, complex variations like structural variants (SVs) and rearrangements can be more accurately and comprehensively represented. The resulting pangenome graph also provides further insights for downstream analysis. In this work, we explore how the pangenome graph can be leveraged for evolutionary analysis and to assess the underlying diversity and selective forces, focusing on how genomic regions, sequence segments, and variants cluster within the graph.

We construct bacteria pangenome graphs to investigate how the paths corresponding to the different individual genomes traverse the sequence segments within the graph. This offers a framework to identify shared structural patterns and lineage specific grouping. Extending this idea to the human genome, we incorporate clinical long-read data with graph references from the Human Pangenome Reference Consortium (HPRC). Beyond the enhanced detection of complex somatic SVs that are often missed or misrepresented by linear references, the path-based perspective on reference pangenome graphs also offers new approaches to characterize recurrent structural patterns and clonal relationships across tumors.

Overall, we highlight the potential of pangenome graphs to uncover new layers of genomic information by analyzing the clustering and paths within it, to explore variant co-occurrence across samples, genomic relationships, and local diversity measures for SVs using the coordinate system implied by the pangenome graph. This approach contributes to ongoing efforts to standardize graph-based methods for evolutionary genomics with important implications for both somatic and population-level applications.

[B303] Using ancient DNA to study historical plant communities surrounding lakes in Victoria, Australia through the Holocene

Callum Swift[1], Dr Vilma Pérez[2], Dr Rafal Gutaker[4], Dr Faidra Katsi[1], Professor Barry Lomax[1], Victoria Miller[3], Associate Professor Janelle Stevenson[3], Alastair Wills[1], Associate Professor Michela Mariani[1], Assistant Professor Andrew Clarke[1]
[1]University of Nottingham, Nottingham, United Kingdom, [2]Adelaide University, Adelaide, Australia, [3]The Australian National University, Canberra, Australia, [4]Royal Botanic Gardens, Kew, London, United Kingdom

Eastern Maar Country within Victoria, Australia is home to many volcanic lakes, which provide a rich archive of past landscapes. Traditional Knowledges and archaeological evidence suggest that this region was rich in plant diversity and included many Australian native food plants used by Indigenous Australians prior to British colonisation. We aim to generate novel empirical evidence of historical plant communities and landscape management in Eastern Maar Country by examining plant ancient DNA from Holocene lake sediments. By studying multiple samples taken from a sediment core it is possible to observe changes in the local plant community through time.

Previous sedimentary ancient DNA (sedaDNA) studies examining historical plant communities highlight the lack of plant genetic reference data as a common issue. This challenge is particularly prevalent in Australia-based research, due to the lower representation of Australian plant species in global databases. Angiosperms-353 genetic data are readily accessible within the Kew Tree of Life for plants across a broad range of families and these data can provide improved taxonomic coverage for historical plant communities within sedaDNA. To help resolve the lack of representation, we explore the potential of using Angiosperms-353 genetic data to rapidly increase the size of the plant reference library, which will – in turn – improve reconstructions of historical plant communities from sedaDNA.

[B304] Tracing almond domestication through ancient DNA and metabolomics

Jazmín Ramos-Madrigal, Nathan Wales, Michael D. Martin, Renato Daniel La Torre Ramírez, Aintzane Rueda, Jesús Guillamón, Neta Manella, Ehud Weiss, Guillem Pérez-Jordá, Alyson Mitchell, Tzion Fahima, Marijke v.d. Veen, Laurent Bouby, Riccardo A. Cigliano, Prof M. Thomas P. Gilbert, Raquel Sánchez-Pérez[1]
[1]PLANT BREEDING DEPARTMENT. CEBAS-CSIC, ESPINARDO, MURCIA, Spain
The domestication of almond (Prunus dulcis, syn. Amygdalus communis) transformed a bitter, amygdalin-rich seed into a globally cultivated nut, yet its origins and early evolutionary dynamics remain poorly understood. We sequenced genomes from 23 archaeological almonds spanning the Levant, North Africa, Italy, Greece, and Iberia (ca. first millennium BC to medieval period), together with modern cultivars and wild relatives, and integrated these data with metabolomic profiling. Ancient genomes reveal early geographic structure, clonal propagation, and a mosaic pattern of domestication.

At the Sweet kernel (Sk) locus, which controls bitterness through a bHLH transcription factor regulating the amygdalin biosynthetic pathway, the sweet allele was not fully fixed in antiquity. Both alleles were present, and amygdalin was chemically detected in Iberian kernels. Metabolomic profiles place ancient almonds in an intermediate state between modern bitter and sweet forms. Together, these results reveal a prolonged, heterogeneous domestication process shaped by cultural exchange, incomplete selection, and later wild introgression.

[B305] The lost tree of Rapa Nui: Genome assembly of the endemic, extinct-in-the-wild Sophora toromiro

Signe Klemm[1], Søren B. Hansen[1], Sebastián Yancovic Pakarati[2], Asal Forouzandeh[3], Jazmín Ramos-Madrigal[1], Jens Soelberg[4], Tina Jørgensen[4], Tue Kjærgaard Nielsen[3], J. Víctor Moreno-Mayar[1]
[1]Globe Institute, University of Copenhagen, , Denmark, [2]Manu Project, Rapa Nui, Chile, [3]Department of Plant and Environmental Sciences, University of Copenhagen, , Denmark, [4]Natural History Museum, University of Copenhagen, , Denmark

Rapa Nui is famous for its enigmatic monoliths, moai, spread across a barren landscape. Another mystery is the extensive deforestation of the island's native flora that happened across a few centuries following Polynesian settlement of the island around 1200 AD. Because of Rapa Nui's extreme isolation and volcanic origin, its native flora consists of only ~50 species, nine of which are endemic. One of these is Sophora toromiro — endemic to Rapa Nui and extinct in the wild since the 1960s. Why the tree disappeared and the magnitude of its use by the Rapanui people remains unclear. Toromiro holds a strong cultural importance for Rapanui, and according to tradition it was used for carving spiritual figures, moai kavakava, and rongorongo scripture tablets.

We sampled fresh leaves from a toromiro specimen that is a direct descendant of plausibly the last tree on the island. We extracted the material with a protocol optimised for plant DNA and sequenced it extensively using long (Nanopore) and short (Illumina) reads. We assembled the genome de novo into 833 contigs (N50=1Mb) with a total size of 586Mb and an average depth of 69x, covering 97.6% of Fabales genes.

Demographic reconstructions using this high-quality toromiro genome have a major bearing on understanding potential anthropogenic effects on Rapa Nui. Furthermore, this genome serves as crucial reference data for metagenomic studies requiring accurate species identification, especially on Rapa Nui, where native flora is understudied. Thus, the toromiro genome represents vital infrastructure for unravelling the history of Rapa Nui.

[B306] Patterns of aDNA Damage Through Time and Environments – lessons from herbarium specimens

Stefano Porrelli[1], Alice Fornasiero[2], Hong Phuong Le[1], Wenzhe Yin[3,4], Maria Navarrete Rodriguez[2], Nahed Mohammed[2], Axel Himmelbach[5], Andrew C. Clarke[3], Nils Stein[5,6], Paul J. Kersey[1], Rod A. Wing[2,7], Rafal M. Gutaker[1]

[1]Royal Botanic Gardens, Kew, London, United Kingdom, [2]King Abdullah University of Science and Technology, Thuwal, Saudi Arabia, [3]University of Nottingham, Sutton Bonington, United Kingdom, [4]The University of Hong Kong, Hong Kong, SAR, China, [5]Leibniz Institute of Plant Genetics and Crop Plant Research, Seeland, Germany, [6]Martin Luther University of Halle-Wittenberg, Halle (Saale), Germany, [7]University of Arizona, Tucson, USA

Herbarium collections are a vast but underutilized resource for ancient DNA research, containing over 400 million specimens with detailed metadata and spanning centuries of global biodiversity. Understanding patterns of DNA preservation in natural collections is crucial for optimizing ancient DNA studies and informing future curation practices. We analysed genomic data for 573 herbarium specimens from six plant species from the genera Hordeum and Oryza collected from the Americas and Eurasia over 220 years. Using standardized laboratory protocols and shotgun sequencing, we quantified DNA degradation and elucidated factors that accelerate it. We find significant age-dependent DNA fragmentation rates, indicating temporal degradation processes not detected in prehistoric samples. In our analysis, DNA decay rates in herbarium specimens were almost eight times faster than in moa bones, reflecting fundamental differences in tissue composition and preservation environments. Environmental conditions at the time of specimen collection emerged as the major determinants of post-mortem damage rates, with the interaction term between temperature and genus being the dominant driver of cytosine deamination. We find no effect of sample storage on DNA damage and degradation. These findings provide insights into how climatic origin, preservation environment, taxonomic identity and age influence DNA preservation while highlighting opportunities for improving institutional preservation practices. Due to standardized preservation conditions, museum collections can provide better insights into DNA damage and degradation over time than archaeological and paleontological samples.

[B307] ClonalRef: A novel palaeogenomic workflow for vegetatively propagated crops

Surabhi Ranavat[1], Amy Holguin[1], Jazmín Ramos-Madrigal[2], Dr Nathan Wales[1]
[1]BioArCh, Department of Archaeology, University of York, , United Kingdom, [2]Center for Evolutionary Hologenomics, Globe Institute, University of Copenhagen, , Denmark

Bioinformatic analyses of ancient DNA sequencing data from vegetatively propagated crops present unique challenges. As these crops do not reproduce sexually, their inheritance patterns challenge assumptions of kinship and are prone to database biases due to shared ancestry. These biological features hinder estimation of genetic diversity, relatedness among lineages and domestication processes through time in such crops. To address these challenges, we present a Nextflow pipeline named ClonalRef for constructing reference panels from whole genome DNA sequencing data and integrating ancient DNA datasets. This pipeline provides a reproducible workflow for mapping, variant calling, kinship estimation, and domestication trait inference tailored to clonally propagated crops. As a case study, we applied this approach to grapevine (Vitis vinifera) to create a reference panel from publicly available whole genome DNA sequencing data of more than 2000 modern samples including both V. vinifera ssp. vinifera and V. vinifera ssp. sylvestris across original grape cultivation regions. We use ClonalRef to perform genotype likelihood inference of ancient DNA samples from published and unpublished datasets. We estimated the relatedness among the samples using the package READ v2 and performed genome-wide association analyses for domestication traits such as berry colour and muscat flavour. We demonstrate how ClonalRef provides an adaptable strategy for palaeogenomic analyses of other vegetatively propagating crops by integrating modern datasets with ancient genomes while accounting for their specific biological features, in a reproducible pipeline, deployable on other high performance compute clusters, to evaluate kinship patterns and cultivation practices across time.

[B308] Polygenic Differentiation Reveals Selection on Autoimmune Disease Variants in the Finnish Population

Somnath Chindhe[1], Päivi Onkamo[1], Elina Salmela[1,2], Miguel Baltazar-Soares[1,3]
[1]Department of Biology, University Of Turku, Finland, Turku, Finland, [2]Department of Archaeology, University of Turku, 20014 Turku, Finland, Turku, Finland, [3]University of Jyvaskyla, Jyvaskyla, Finland

Natural selection is expected to eliminate genetic variation with negative impact on organismal fitness, such as deleterious alleles associated with increased disease risk. Genome-wide association studies (GWAS) identify genetic variants associated with disease risk, have revealed a) deleterious alleles are largely present in the human population and b) most diseases are polygenic. Across evolutionary time-scales, individual loci provide a limited perspective on how selection has modulated them, whether deleterious disease alleles once conferred advantages in past. Hence, polygenic tests are required to capture subtle, genome-wide shifts in allele frequencies linked to disease risk. Here, we test whether local adaptation has shaped polygenic disease risk by quantifying differences in genetic value (allele-frequency weighted by GWAS effect sizes) between Finnish and pan-European (UKBB) populations for 11 diseases. For each, we applied Polygenic Differentiation test to assess departures from neutrality, comparing the Finnish and pan-European populations. Our results show that autoimmune diseases and hypothyroidism exhibited elevated genetic value differences, particularly among SNPs with higher disease risk. To explore how natural selection could have acted on these SNPs, we analysed genotype data from Finnish individuals in the 1000 Genomes Project using ancestral recombination graph methods to estimate allele-frequency changes and selection coefficients. Analyses identified 15 candidate SNPs exhibiting signatures of selection. Functional analysis using public eQTL data showed that genes linked to these SNPs were enriched for processes related to immune response. These findings suggest that these SNPs may have conferred an advantage during past infections but would increase disease risk among modern Finns.

[B309] Invasive African sacred ibis in Taiwan: insight from comparative genomics and population genetics

Chen Siang Ng[1]
[1]National Tsing Hua University, Hsinchu, Taiwan

The African sacred ibis (Threskiornis aethiopicus) has successfully established invasive populations in Taiwan, raising concerns about its ecological impact and prompting questions about the genetic basis of its adaptability. In this study, we generated the first chromosome-level genome assembly for the species and conducted comparative and population genomic analyses to uncover the molecular foundations of its invasion success. Resequencing of 34 individuals, including zoo and wild populations across Taiwan, revealed a single-source introduction from zoo escapees, a significant genetic bottleneck 7–11 generations ago, and a current wild population size of ~4,000 individuals. Despite reduced heterozygosity, wild effective populations exhibit signs of local adaptation, particularly in genes related to immunity, cognition, reproduction, and responses to environmental stress. We identified expanded gene families associated with immune defense, neural function, and metabolism, indicating a multifaceted genomic architecture that enables behavioral flexibility and physiological resilience. Furthermore, genes under positive selection in the Taiwanese wild population include those involved in pathogen recognition, synaptic plasticity, and bone development, potentially contributing to the ibis's characteristic morphology and ecological versatility. Functional enrichment analyses highlighted adaptive pathways, including cAMP signaling, endocannabinoid regulation, and progesterone-mediated reproduction. Together, these results demonstrate how genome-wide adaptations—spanning immunity, cognition, reproduction, and skeletal traits—have contributed to the African sacred ibis's ability to thrive in novel environments. This study offers a comprehensive genomic perspective on biological invasions, providing valuable insights into the evolutionary processes that shape invasive potential.

[B310] A likelihood-based method to infer the dispersal and the impact of natural selection using spatiotemporal data

Alessandro Lopez-Hernandez[1], Diego Ortega-Del Vecchyo[1]
[1]Computational Population Genetics Group. LIIGH-UNAM, Querétaro, Mexico
Natural selection is a process that drives spatial and temporal changes in allele frequencies. Here, we propose a method to estimate the impact of natural selection acting on deleterious and advantageous alleles while incorporating spatial and temporal information. The method estimates the dispersal coefficient of individuals and the impact of natural selection acting on a set of alleles using a maximum likelihood approach. The approach uses spatial data from the geographical location of a sample, temporal information from the age of the sample, as well as genotypic data from the sample taken. The likelihood-based method employs partial differential equations to model spatiotemporal allele frequency changes due to the joint action of the dispersal of individuals and the impact of natural selection acting on a set of alleles. To assess the robustness of the method, we performed forward in time simulations under a continuous and a discrete space. We collected allele frequency trajectories from the simulated individuals and used the resulting data to evaluate the accuracy of the method to estimate the dispersal coefficient of individuals and the impact of natural selection acting on a set of alleles. We apply the method to data from the Allen Ancient DNA Resource (AADR) from ancient humans that lived in Eurasia during the holocene. Furthermore, we discuss possible applications of our method to other species.

[B311] Cryptic polygenic adaptation of body mass in a wild meerkat population

Alexander Downie[1], Elizabeth Mittell[2], Kasha Strickland[2], Harry Ames[2], Marta Manser[3,4], Tim Clutton-Brock[4,5], Loeske Kruuk[2], Josephine Pemberton[2], Jenny Tung[1,6,7,8,9]
[1]Max Planck Institute For Evolutionary Anthropology, Leipzig, Germany, [2]Institute of Ecology and Evolution, University of Edinburgh, Edinburgh, United Kingdom, [3]Department of Evolutionary Biology and Environmental Studies, University of Zurich, Zurich, Switzerland, [4]Kalahari Research Centre, Kuruman River Reserve, Van Zylsrus, South Africa, [5]Department of Zoology, University of Cambridge, Cambridge, United Kingdom, [6]Department of Biology, Duke University, Durham, United States, [7]Department of Evolutionary Anthropology, Duke University, Durham, United States, [8]Canadian Institute for Advanced Research, Toronto, Canada, [9]Faculty of Life Sciences, Institute of Biology, Leipzig University, Leipzig, Germany

Evolution by natural selection requires heritable variation in fitness-associated traits and results in an increase in the frequency of advantageous, trait-associated genetic variants over time. Case studies demonstrating both properties remain rare in natural populations, especially for complex traits. Here, we combine phenotypic, life history, and genome resequencing data from 3,124 wild meerkats (Suricata suricatta) in the Kalahari Desert in South Africa to investigate the genomic response to selection on adult body mass. We find that adult mass is heritable and strongly predicts lifetime reproductive success. We show that adult mass is, as expected, a highly polygenic trait, although inbreeding allowed identification of a rare, large-effect allele. We use gene dropping simulations through the 11-generation population pedigree to establish null expectations for allele frequency change in the absence of selection. Over the course of the study, putative mass-increasing alleles have tended to rise in frequency relative to background expectation, consistent with adaptation in response to selection for larger size. We therefore estimate that the genetic component of adult mass has increased by ~0.28 standard deviations in 33 years. Surprisingly, realized adult mass has decreased over the same period. Our results suggest that the response to selection on body mass may be a form of genetic compensation for co-occurring environmental deterioration, including increased rates of drought and extreme temperature. Together, our results demonstrate how genomic analyses in long-term field studies can capture polygenic responses to selection in nature, even when these responses are incompletely mirrored by changes in the traits themselves.

[B312] Genomic determinants of reproductive success in a critically bottlenecked parrot

Audald Lloret-Villas[1], Jim J. Groombridge[2], Rangsinee Sankhom[2], Cock Van Oosterhout[3], Carolina Pacheco[4], Vikash Tatayah[5], Sion Henshaw[5], Simon Tollington[6], Dr Hernán Morales[1]
[1]University of Copenhagen, , Denmark, [2]University of Kent, , UK, [3]University of East Anglia, , UK, [4]University of Lund, , Sweden, [5]Mauritian Wildlife Foundation, , Mauritius, [6]Nottingham Trent University, , UK

The threatened Mauritius parakeet (Alexandrinus eques) is the only surviving parrot species in Mauritius and the Mascarenes. The species underwent an extreme population bottleneck driven by habitat loss and invasive species, reaching only ~20 individuals in 1986. Although population size has since recovered through intensive genetic rescue and managed breeding programs, the downlisting from Critically Endangered to Vulnerable on the IUCN Red List in 2019 has not been accompanied by a corresponding increase in genetic diversity or heterozygosity, raising concerns about long-term extinction risk.

To investigate how genomic erosion affects fitness and to assess the genetic basis of reproductive success in this critically bottlenecked population, we combined a comprehensive temporal genomic dataset - including 70 genomes from founding individuals immediately following the bottleneck and over 200 genomes from modern individuals - with detailed records of breeding events and reproductive success spanning the last 35 years. High coverage sequencing and pedigree data from both highly successful and unsuccessful parents, as well as from the surviving offspring, provide a unique opportunity to identify deleterious and beneficial allelic combinations influencing fitness. In addition, summarized lifetime reproductive success for 150 individuals with both genomic and reproductive data enables genome-wide association analyses to identify genomic regions enriched for fitness-related variation.

Together, these results inform conservation strategies aimed at mitigating genetic load and improving the effectiveness of managed breeding programs.

[B313] Improved inference on the architecture of complex traits in populations under strong polygenic selection by combining population and quantitative genetics

Elise Tourrette[1], Hélène Gilbert[1], Bertrand Servin[1]
[1]INRAE - Génétique Physiologie Système d'Elevage, Toulouse, France
Many traits affecting fitness are complex, quantitative phenotypes that are genetically determined by multiple loci. The evolution of populations is therefore governed by the response of a large fraction of the genome to phenotypic selection, i.e. populations are under *polygenic selection*. But beyond its effect at causal loci, selection also imposes constraints on the transmission of neutral alleles by genetic drift as it drives successful matings. This effect is particularly important in populations experiencing rapid genetic changes through the combined effects of drift and selection.

In this work we show how this combination of effects biases inference of polygenic selection on fitness in a high drift, high selection context. As current GWAS methods do not fully correct for this bias, we developped a new method that effectively disentangles drift from selection by combining selective effects estimated from genetic time series and quantitative effects estimated using classical GWAS models. This new approach offers better estimates of quantitative effects at QTLs for fitness traits and should help to better predict their evolution in response to new adaptive challenges and/or in other populations of shared ancestry.

[B314] Studying protective alleles of Sub-Saharan African Ancestry in Iberia: A genetic study of the Morenos from Huelva

Carlos Sarabia[1], Juan Luis Vázquez-Alemán[2], Francesc Calafell[1], Elena Bosch[1], Prof. David Comas[1]
[1]Institute Of Evolutionary Biology (IBE-UPF CSIC), Barcelona, Spain, [2]Department of History, Geography and Anthropology, Faculty of Humanities, University of Huelva, Huelva, Spain

Adaptation in the real world normally appears in populations that violate classical assumptions of equilibrium, experiencing bottlenecks, recent admixture, and strong environmental change. In such contexts, selection may act rapidly and efficiently, yet its genomic signatures are difficult to disentangle from drift. The Iberian Peninsula, which holds the highest genetic diversity and the highest levels of North African and Sub-Saharan African (SSA) ancestry in Europe, offers an opportunity to explore these dynamics. We focus on the Moreno population from Gibraleón (Huelva, Spain), a community documented since the 1540s and historically linked to the African diaspora. Using genome-wide data, we reconstruct population structure, quantify West African ancestry and test for genomic regions showing excess SSA local ancestry beyond neutral expectations. Malaria - historically persistent in southwestern Iberia- is a showcase example of a disease imposing strong selective pressures, but we adopt a general framework centered on infectious disease and immune-related variation. Our study asks whether adaptive admixture has contributed to present-day susceptibility or resistance profiles, and whether such signals can be robustly detected in highly admixed genomes. We discuss key technical challenges, including reduced statistical power and ancestry misassignment, and demonstrate how ancestry-aware methods can solve these problems. Together, our findings emphasize that admixture is not merely a demographic record, but a potential engine of rapid adaptation in populations.

[B315] A wavelet based multiscale alternative to sliding window analyses in population genetics

Eduardo Martin González Orozco[1], Leonardo Iasi[1], Benjamin Peter[1]
[1]Max Planck Institute for Evolutionary Anthropology, , Germany

A key problem in population genetics is to reconstruct evolutionary processes from genomic data. Under the coalescent model, there are two sources of variability. First, genetic drift and recombination yield an ancestral recombination graph (ARG), We refer to the randomness in this process as branch noise. Second,because in most cases the ARG is unobserved inference must rely on polymorphism patterns, which introduce mutational noise

A large class of summary statistics such as nucleotide diversity and Tajima's D are often used to study local patterns of variations, for example to detect loci under selection. These statistics can be defined in terms of the ARG and polymorphisms, and are typically estimated using sliding windows One issue with this approach is that the window size is usually chosen arbitrarily, which can lead to issues: If windows are too small, they are dominated by mutational noise; if they are too large, we may lose localized signals.

Here, we propose an alternative framework based on the Haar wavelet transform. We show that, as a baseline, it can be interpreted as a multiscale generalization of sliding windows; equivalent to calculating sliding windows of different sizes. We show that mutational noise and branch noise impact genetic diversity at different scales, and thus their effects can be separated using the wavelet transform. Consequently we can use wavelet-based denoising techniques to obtain a clearer, data-driven representation of the underlying evolutionary signal, providing a robust alternative to sliding-window analyses

[B316] Genomic Consequences of Extreme Isolation in the Raute

Xavier Martí-Pérez[1], Francisco Ceballos[1], Inez Derkx[2], Sudarhan Subedi[3], Prajwal Rajbhandari[4], Anita Gyawali[5], Jaume Bertranpetit[1], Lucio Vinicius[2], Andrea B Migliano[2], Elena Bosch[1]
[1]Universitat Pompeu Fabra, Barcelona, Spain, [2]University of Zurich, Zurich, Switzerland, [3]Tribhuvan University, Kathmandu, Nepal , [4]Research Institute for Bioscience and Biotechnology (RIBB), Lalitpur, Nepal, [5]Committee to study the social, cultural, economic and geographical and habitat of Raute community, Surkhet, Nepal

The Raute (~140 individuals) are Nepal's last nomadic hunter-gatherers. To quantify the genomic consequences of long-term isolation, we generated 15× whole-genome sequences for 96 Raute and 16 neighboring Dailekh farmers and analyzed them in global and Himalayan reference panels. PCA and ancestry deconvolution reveal predominant Himalayan-related ancestry, with closest affinity to the Tharu (a formerly foraging group). Despite this shared regional background, the Raute display extreme autozygosity: nearly all individuals have >20% of the genome in ROH >1.5 Mb. ROH profiles show very high tract counts (~150–200) and extraordinary cumulative lengths (~0.5–0.8 Gb), dominated by intermediate-to-long segments (2–8 Mb and >8 Mb). Comparing the number of ROH (NROH) to the sum of ROH lengths (SROH) indicates drift-dominated inbreeding (many ROH of moderate length) rather than systematic consanguinity (few extremely long ROH), consistent with high FROH (~0.14–0.25) and near-zero/negative FIS. Demographic inference with GONE2 further supports severe recent isolation, yielding an effective population size of ~30 individuals over the last ~23 generations. Building on this framework, we are testing how extreme drift and bottlenecks shape mutational load and signatures of local adaptation. These results provide a rare genomic view of human resilience under sustained demographic collapse.

[B317] How does inbreeding affect local adaptation in the mountain plant Arabis alpina?

Freya Way[1], Cara Duffy[1], Konrad Lohse[1], Nicky Lustenhouwer[2], Matthew Hartfield[1]
[1]Institute of Ecology and Evolution, University of Edinburgh, Edinburgh, United Kingdom, [2]School of Biological Sciences, University of Aberdeen, Aberdeen, United Kingdom

The process of adaptation to the environment can be impacted by many factors, one of which is the species' mating system. Self-fertilisation occurs in 42% of angiosperms, including rare species and crops, yet its impact on adaptation in natural populations is unresolved. On one hand, selfing causes extreme inbreeding and reduces the efficacy of selection by decreasing the effective population size and effective recombination. However, theory also indicates that selfing can increase the fixation of recessive beneficial mutations, and promote local adaptation by limiting pollinator-mediated gene flow. To better understand how inbreeding affects adaptation in nature, this project investigates local adaption to altitude in the wild mountain plant Arabis alpina, which exhibits regional differences in its selfing rate. We sampled leaf tissue, phenotype and site data at high and low altitude from self-compatible populations in France and self-incompatible populations in Italy. We obtained whole genome sequences, combined our new dataset with existing data, and are comparing how the intraspecific variation in the mating system affects the population structure and genomic signals of local environmental adaptation. This information will eventually help us understand how selfers may differ from outcrossers in their responses and resilience to environmental change. In addition, we are interested in how non-equilibrium conditions affect data analysis as selfing species tend to have strong population structure and defy classic population genetic assumptions such as hardy Weinberg equilibrium.

[B318] Looking for the ones that never made it out: purifying selection on de novo mutations during spermatogenesis

Genís Garcia-Erill[1], Peter Porsborg[1], Vinod Kumar Singh[2], Anders Charmouh Poulsen[1], Sofia Winge[3], Lasse Hansen[4], Mengjun Wu[1], Marta Pelizzola[4], Oliver Hansen[2], Juliana Berner[5], Dr Christina Hvilsom[6], Carmen Oroperv[2], Hank Charlton[1], Asger Hobolth[4], Pr Thomas Bataillon[1], Søren Besenbacher[2], Dr Kristian Almstrup[3], Mikkel Schierup[1]
[1]Bioinformatics Research Centre, Aarhus University, Aarhus, Denmark, Aarhus, Denmark, [2]Department of Molecular Medicine (MOMA), Aarhus University Hospital, Aarhus, Denmark, [3]Department of Growth and Reproduction, Copenhagen University Hospital, Copenhagen, Denmark, [4]Department of Mathematics, Aarhus University, Aarhus, Denmark, [5]Copenhagen Zoo, Frederiksberg, Denmark, [6]Globe Institute, University of Copenhagen, Copenhagen, Denmark

De novo mutations in the germline are the source of novel genetic variation that fuels evolution. Selection can act on these mutations depending on their functional impact. By characterizing depletion of de novo mutations in functional categories, it is possible to infer purifying selection when sufficient numbers of mutations are available. However, such analyses cannot determine the developmental stage at which selection acts. Recent advances in high-fidelity sequencing such as PacBio HiFi sequencing allow de novo mutations to be accurately identified from bulk sequencing data, even when supported by a single read. When applied to sperm or testis tissue, these approaches enable the detection of mutations arising in the male germline. In principle, this makes it possible to compare the functional consequences of de novo mutations across different stages of spermatogenesis and thereby characterize the strength of purifying selection acting during this process. To assess the feasibility of this approach, we perform power analyses to determine the amount of de novo mutation events needed to detect selection signals under different selection strengths. We then compare functional annotations of de novo mutations from different spermatogenesis stages in human and other primates using PacBio HiFi sequencing of testes and sperm. We observe a lower proportion of deleterious de novo mutations in sperm relative to testes samples, suggesting there is indeed a detectable effect of purifying selection acting on de novo mutations during spermatogenesis.

[B319] Local ancestry inference identifies adaptive admixture in Neolithic Europe

Georgia Mies[1], Iain Mathieson[1]
[1]Department of Genetics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, United States

During the Neolithic transition, migrating Anatolian farmers admixed with local European hunter-gatherers. Simultaneous shifts in ancestry, diet, and environment likely imposed strong selective pressures. Recent selection in admixed populations can be detected using local ancestry inference; however, existing methods were developed and validated primarily in present-day populations, and their performance in ancient DNA, where reference panels are smaller, data are sparser, and admixture is more ancient, remains unresolved.

We evaluate six local ancestry inference methods on 176 imputed genomes from admixed European Neolithic farmers, comparing inferred ancestry proportions, tract lengths, and signals of selection. All methods overestimate hunter-gatherer ancestry when source sample sizes are unbalanced (0.30–0.36 vs. qpAdm: ~0.20), although individual-level estimates are highly correlated (R>0.85). Filtering by posterior probability and using optimized source sample sizes for each method reduces this bias (0.20–0.30). Tract length distributions vary widely, producing admixture time estimates differing by more than an order of magnitude. Only RFMix produces a distribution consistent with archaeological expectations (~10 generations).

Meta-analysis accounting for correlations across methods identifies four genomic regions with consistent ancestry deviations, replicated in an independent dataset. These included increased hunter-gatherer ancestry at the HLA region (immune adaptation), increased farmer ancestry at SLC24A5 (skin pigmentation), and two novel candidates: FADS1 (fatty acid metabolism) and PER3 (circadian regulation).

Our results demonstrate that local ancestry inference can recover biologically meaningful signals of adaptation in recently admixed, ancient populations, but methodological choices strongly shape inferences, highlighting the need for careful validation when applied to ancient admixture.

[B320] Testing for local adaptation in rice landraces from Southeast Asia using gene expression

Isaura Rosas Reinhold[1], Dr Michael Purugganan[1]
[1]New York University, New York City, United States

Natural selection is the dominant force driving adaptive differentiation between populations and species. Thus, it is critical to characterize and quantify selection to understand how populations and ecotypes adapt to local conditions. To address this, we collected rice leaves and roots from 36 landraces cultivated in a common garden experiment at the International Rice Research Institute in the Philippines. Samples were collected during the dry season 40 days after sowing from three different treatments: irrigated, rainfed lowland, and upland fields. We performed RNA extraction on two biological replicates across these conditions and tissues (roots and leaves), resulting in 1,296 samples.

Sequencing was conducted using the 3' prime-end method, allowing us to analyze > 22,000 genes. Our analysis showed that gene expression in leaves is less differentiated between treatments compared to roots. Interestingly, the ecotypes to which landraces are locally adapted do not appear to significantly affect gene expression; instead, treatment is the primary observed effect. Differentially Expressed Gene analysis showed a very similar genetic landscape in leaves in all treatments, while roots exhibited higher differentiation across treatments. These results correlate with fitness data from the same experiment, which indicated no significant effects from treatment or ecotype. Further analyses are required, including selection analysis on gene expression using both transcriptomic and phenotypic data. Additionally, we aim to evaluate selection on gene regulatory networks (GRNs) to test for differences in selection patterns across specific networks and ecotypes.

[B321] Adaptive Divergence in a Sea of Gene Flow: Selection, Hybridization, and Structure in Baltic Herring

Jake Goodall[1,2], Mats Pettersson[1,2], Anastasia Andersson[3], Iris Dahlin[4], Nils Ryman[3], Gunnar Ståhl[3,5], Lovisa Wennerström[4], Leif Andersson[1,2], Linda Laikre[3]
[1]Uppsala University, Uppsala, Sweden, [2]SciLifeLab, Uppsala, Sweden, [3]Stockholm University, Stockholm, Sweden, [4]Swedish University of Agricultural Sciences, Uppsala, Sweden, [5]Biolab, Skärholmen, Sweden

Understanding how natural selection can maintain adaptive divergence amid high homogenizing gene flow is challenging, particularly in marine species, which often exhibit high connectivity. Atlantic herring (Clupea harengus) offers a unique study system in this respect, setting sharp islands of adaptive divergence against species-wide high effective population sizes, wide dispersal, and minimal differentiation at selectively neutral loci. Using >4,500 spawning individuals genotyped at >4,500 SNPs, and sampled from 150 Baltic Sea locations, we performed a high-resolution genetic study to assess adaptive variation in natural Baltic herring populations.

Initially, we explored the major spawning ecotypes in the Baltic Sea (spring versus autumn spawners), reconfirming strong differentiation at spawning- and photoperiod-associated loci. However, we also document a subset of putative hybrid individuals with offset spawn timings, intermediate allele frequencies, and high heterozygosity at said loci. These individuals highlight ongoing gene flow even among seasonal-spawning ecotypes.

At a finer spatial scale, the spring-spawning ecotype exhibits population structure and clinal patterns of divergence across both environmental and temporal gradients, which involve large structural inversion regions. Patterns of linkage disequilibrium partitioning (using Ohta's statistics) were consistent with divergence maintained by local selection despite ongoing gene flow, and we observed multiple local populations with distinct SNP profiles relative to the regional genetic background.

Together, these results highlight how local selection can shape genomic landscapes in natural populations, and highlight Atlantic herring as a powerful system for understanding adaptive divergence under homogenizing gene flow.

[B322] Age reshapes selection on melanoma driver mutations

Jeffrey Townsend[1], Ms Yihan Liu[1], Dr Jeffrey Mandell[1], Dr Moein Rajaei[1], Prof Jorge Alfaro-Murillo[1]
[1]Yale University, New Haven, United States

A central goal of molecular evolutionary analysis is to distinguish the contributions of mutation and selection to genetic change. Aging is a dominant risk factor for cancer, yet it remains unclear whether age reshapes tumor evolution primarily by altering mutation supply, the strength of selection on driver variants, or both. Here, we analyze age as a continuous measure and test how mutation and selection vary. We integrated whole-exome sequencing data from The Cancer Genome Atlas with targeted sequencing from AACR Project GENIE, analyzing 5,472 melanomas to estimate variant-specific selective advantages as functions of age.

Neutral mutation rates were inferred from synonymous substitutions and mutational covariates, with site-specific rates calibrated using COSMIC mutational signatures. Selection coefficients were estimated across age strata and modeled continuously within a unified Poisson-likelihood framework. Neutral mutation rates increased only modestly with age, and UV-associated mutational processes dominated across all age groups, although endogenous signatures became more prominent in older tumors. In contrast, selection exhibited strong, variant-specific age dependence. The selective advantage of BRAF V600E declined steeply with age, whereas NRAS Q61 substitutions showed increasing selection in older individuals. TERT promoter mutations became more frequent with age but conferred progressively weaker selective advantages, and gene-level analyses of TP53, PTEN, and CDKN2A revealed monotonic declines in selection.

Together, these results demonstrate that age reshapes somatic evolutionary trajectories primarily through systematic changes in selection rather than mutation supply, illustrating how life-history variables can be treated as evolutionary environments for age-aware inference of fitness effects in structured populations.

[B323] Signatures of differential local adaptation indicate climate matching with specific native-range habitats in invasive raccoons (Procyon lotor)

Joanna Kołodziejczyk[1], Maciej Konopiński[1], Eric Hoffman[2], Aleksandra Biedrzycka[1]
[1]Institute Of Nature Conservation Polish Academy Of Sciences, Kraków, Poland, [2]Department of Biology, University of Central Florida, Orlando, USA

Invasive species pose a major threat to ecosystems through habitat destruction, disease transmission, and competition for resources. Invasion success depends on the adaptive potential (i.e., the ability to respond to selection pressures that is dependent on the levels of genetic variation) of the invading species. In the presented study, we used reduced representation sequencing of native and European invasive raccoon populations to identify putatively adaptive changes across geographic regions. Our findings indicate that invasive raccoon populations experienced a genome-wide reduction in genetic diversity; however, we detected signatures of selection that were putatively related to local adaptation within the invasive range. Analysis of loci under selection associated with either native or invasive populations revealed parallel selection pressures between the invasive range and one of the native populations, all inhabiting similar climatic niches. In contrast, the native population from a different climatic region experienced distinct selective pressures. These results highlight the importance of habitat similarity in invasion processes. They demonstrate that, when predicting the potential range of an invasive species, it is not only the geographic distribution of the native range that should be considered, but also the specific characteristics of the habitat. Genomic regions under selection were linked to traits that promote rapid expansion, including reproduction, cognitive function, stress response, immunity, and energy production. Overall, this study illuminates how contemporary selection in raccoons may accelerate the invasion process.

[B324] Numerical challenges when inferring selection and population size history from the site frequency spectrum

Jonathan Wiese[1], Matthias Steinrücken[1]
[1]Department of Ecology and Evolution, University of Chicago, Chicago, United States
Inferring both non-equilibrium demography and selection from observed genetic data is a central goal in population genetics. The site frequency spectrum (SFS) is a widely used summary of observed genetic variation, and in order to utilize it for inference, we need to model how the shape of the expected SFS depends on the parameters of interest. Here we implement and analyze several numerical approaches, including the exact Wright-Fisher (WF) model, diffusion-based approximations, and moment-based approaches, to compute the expected SFS for non-equilibrium demographic models with selection. We test each approach on simulated data using a maximum likelihood framework for parameter inference. For positive selection, we find that all methods produce comparable results in the weak selection, small population size regime. However, diffusion-based methods diverge from the exact WF model when selection is moderate to strong, even when standard approximations assuming weak selection are relaxed. Furthermore, moments-based methods are only numerically stable for weak selection, while exact WF-based methods are computationally intractable for large population sizes. We conclude that the optimal method to compute an expected SFS depends on both the strength of selection and the population size history. For likelihood-based inference of demographic and selection parameters, we recommend a blended approach, in which different methods are used in their appropriate regimes in the parameter space.

[B325] Can a natural Daphnia magna population (rapidly) adapt to a new predator?

Kathrin Otte[1], Jost Borcherding[2], Lisa Hermann[2], Eric von Elert[2]
[1]Hamburg University, Hamburg, Germany, [2]Cologne University, , Germany

Predation is a key factor shaping ecosystems, and the invasion of a fish predator can severely impact freshwater habitats by altering zooplankton communities. Daphnia are famous for showing several defence strategies when facing fish, including morphological, life-history and behavioral changes. The selection pressure by predation on Daphnia populations is typically strong and phenotypic changes in response to changes in predator regime rapid and often complex. The underlying genetic architecture has been described as polygenic and several hundreds of single nucleotide polymorphisms (SNPs) are involved in adaptation to fish predation.
Here, we study a recent fish invasion to a previously fish-free gravel pit lake in Northern Germany and its consequences for the local Daphnia magna population. After its establishment in 2006, the lake harbored a rich zooplankton community including Daphnia magna and Daphnia pulex. In 2019, first individuals of the fish species common sunbleak (Leucaspius delineatus) were detected and the fish population increased massively over the following years. We analyzed the consequences for the zooplankton community in general and, more specifically, the genomic and phenotypic characteristics and changes in the local D. magna population. We further compare our results to predictions made from population genetic simulations mimicking the scenario of fish invasion as well as to data from other European D. magna populations experiencing selective pressure by fish predation.

[B326] Genomic conflicts between heat and cold tolerance during development in a desert bird

Lea Huber[1], Professor Charlie Kinahan Cornwallis[2], Dr Molatelo Kekana[3], Neleke Lotz[3], Dr Zanell Brand[3], Prof Schalk Cloete[3,4], Dr Anel Engelbrecht[3], Dr Mads Fristrup Schou[1]
[1]Department of Biology, Aarhus University, Aarhus, Denmark, [2]Department of Biology, Lund University, Lund, Sweden, [3]Directorate of Animal Sciences, Western Cape Department of Agriculture, Elsenburg, South Africa, [4]Department of Animal Sciences, Stellenbosch University, Stellenbosch, South Africa

A key challenge for endotherms is maintaining a constant body temperature despite exposure to temperature fluctuations throughout life. During development changes in body size can alter vulnerability to both heat and cold. This may mean that thermoregulatory responses that are adaptive in young and small individuals are maladaptive when individuals are older and larger, potentially constraining thermal adaptation. We tested this idea by examining the molecular responses to heat (42°C) and cold (12°C) exposure of 1-week and 8-week old ostrich chicks (Struthio camelus), a species native to highly fluctuating thermal environments and characterised by rapid early growth. We found that 34% of genes responding to temperature treatments were involved in both heat and cold responses, implying that these responses partly share a genetic basis that is modulated by gene expression. The thermal stress response of many of these genes (38%) was further affected by age. For example, genes upregulated early in life were downregulated later in development, and vice versa. These results suggest that heat and cold responses change throughout development, driven by shifts in thermoregulatory demands, potentially resulting in balancing selection due to opposing directional selection across ages. We tested this hypothesis by contrasting the genomic variation in differentially expressed genes across two ostrich subspecies adapted to different thermal environments. Our results show that, while thermoregulatory responses appear adaptively modulated across age, this modulation also creates conflicts that may constrain thermal adaptation across life.

[B327] A quantitative approach of polymorphism in bacteria beyond synonymous non-synonymous classification : impact of lifestyle in E. coli/Shigella

Manolo Mischler[1], Doctor Lucile Vigué[1], Doctor Giancarlo Croce[2], Professor Martin Weigt[3], Professor Olivier Tenaillon[1]

[1]Université Paris Cité, Inserm, Institut Cochin, Paris , France, [2]Department of Oncology UNIL CHUV, Ludwig Institute for Cancer Research, University of Lausanne, Lausanne, Switzerland, [3]Sorbonne Université, CNRS, Institut de Biologie Paris Seine, Computational and Quantitative Biology LCQB, Paris, France

Determining the distribution of fitness effects (DFE) of new mutations has been a long-standing goal of population genetics since the advent of genomic sequencing. Current methods are based on the comparison of the non-synonymous and synonymous site frequency spectra (SFS) and infer the global shape of the DFE by examining the progressive depletion of non-synonymous mutations with increasing mutation frequencies in a genome sample.

In parallel, a plethora of bioinformatics methods aiming to directly infer the impact of individual amino-acid on protein function have been developed. These tools are validated by measuring their accuracy of predicting the outcomes of large experimental mutational assays, which are time- and resource-expensive. Taking advantage of the broad availability of bacterial genomes, we use naturally segregating polymorphism as a ground truth to test whether mutational effects predicted in silico are consistent with the predictions of population genetics.

We extracted the alleles of 2,358 core genes from a set of 81,440 E. coli genome and computed a fitness score for all possible mutations in these genes using Direct Coupling Analysis (DCA), a protein model that infers site-specific preferences for amino-acids and intra-protein epistatic interactions. We show that combining large scale population genomics with mutation effect inference tools can help us investigate the intensity of selection quantitatively from synonymous to non-synonymous mutations as well as across different E. coli lineages experimenting diverse ecological lifestyles. Overall we show that DCA offers a new layer of analysis for understanding how polymorphism segregate in bacterial populations across times and lifestyles.

[B328] Identification of positive selection in the ancestral source populations of East Asians using ancient genomes

Masixuan Zhao[1,2], Dr. Dan Ju[1], Qiaomei Fu[1]
[1]Key Laboratory of Vertebrate Evolution and Human Origins, Institute of Vertebrate Paleontology and Paleoanthropology, Chinese Academy of Sciences, Beijing, China, [2]University of Chinese Academy of Sciences, Beijing, China

Numerous studies have identified signals of natural selection in East Asians based on present-day genomes, but population admixture events throughout East Asia may have obscured historical selective events. Ancient genomes that represent the source populations for present-day individuals can aid in identifying past selective sweeps and pinpointing the ancestral populations in which these events occurred. Thus, we assembled a large dataset of published ancient genomes from East Asia (N = 900). With the Ohana framework, we performed unsupervised learning of population structure on this dataset, identifying components related to ancestries such as Jomon and Tibetan (K=8). We conducted admixture-aware genome-wide selection scans, testing each population individually for loci with allele frequencies deviating from expectations based on genome-wide covariances. We detected selection at EPAS1 (P = 2.98 ×10^-9) in the ancestry components associated with Qinghai-Tibet Plateau populations, a known adaptive locus implicated in adaptation to low-oxygen environments. We also identified selection on rs1229984 (P = 4.79×10^-11) a missense variant in ADH1B, that influences alcohol metabolism occurring on the component related to southern Chinese and Southeast Asian populations, which strengthens the connection of this variant to rice farming. Finally, we detected a novel selection signal at IFNAR2 (P = 1.41×10^-20), a gene involved in antiviral immune response, on a component related to Jomon ancestry. Together, our results demonstrate that ancestry-resolved analyses with the aid of ancient genomes can uncover adaptive events that are otherwise difficult to resolve in modern populations, offering new insights into the population context of selection in East Asia.

[B329] Limited evidence for adaptive evolution of metabolic risk in Polynesian populations

Dang Liu[1,19], Iotefa Teiti[2], Gaston Rijo de León[1], Mathilde André[1], Elise Kerdoncuff[1], Maguelonne Roux[1], Leo Speidel[3,4], Maite Aubry[2], Jérémy Choin[5], Anita Teissier[2], Vaea Richard[2], Zhi Li[1], Christine Harmant[1], Aurélie Bisiaux[1], Anthony Jaquaniello[1], Tuterarii Paoaafaite[2], Lisa Dian[2], Kiyojiken Chung[2], Annie Forster[6], Adrian V. S. Hill[6], George Aho[7], Ratu Vereniki Raiwalui[8], Alexander Ioannidis[9], Dr. Andres Moreno-Estrada[10], Alexander J. Mentzer[6], Phillip Endicott[11], Mark Stoneking[12], Frédérique Valentin[13], Jacques Vernaudon[14], Guillaume Molle[15], Eric Conte[15], Mary Walworth[16], Aymeric Hermann[15,17], Van-Mai Cao-Lormeau[2], Etienne Patin[1], Lluis Quintana-Murci[1,17]
[1]Human Evolutionary Genetics Unit, Institut Pasteur, Université Paris Cité, CNRS UMR2000, Paris, France, [2]Laboratory of Research on Emerging Viral Diseases, Institut Louis Malardé, Papeete, French Polynesia, [3]Ancient Genomics Laboratory, Francis Crick Institute, , UK, [4]Genetics Institute, University College London, , UK, [5]Department of Human Evolutionary Biology, Harvard University, Cambridge, USA, [6]Centre for Human Genetics, University of Oxford, , UK, [7]Department of Paediatrics, Vaiola Hospital, Nuku'alofa, Tonga, [8]Oceania Hospital Private Limited, , Fiji, [9]Department of Biomolecular Engineering, University of California Santa Cruz, , USA , [10]National Laboratory of Genomics for Biodiversity, Advanced Genomics Unit, CINVESTAV, Irapuato, Mexico, [11]Evolutionary Biology Department, University of Tartu, , Estonia, [12]Laboratoire de Biométrie et Biologie Evolutive, CNRS UMR5558, Université Lyon 1, Villeurbanne, France, [13]CNRS UMR8068, MSH Mondes, Nantes, France, [14]Maison des Sciences de l'Homme du Pacifique, CNRS UAR2503, Université de la Polynésie Française, , French Polynesia, [15]Centre International de Recherche Archéologique sur la Polynésie, UR4242, Université de la Polynésie Française, , French Polynesia, [16]Centre for Linguistic Research on East Asian (CRLAO), École des Hautes Études en Sciences Sociales (EHESS), CNRS UMR8563, , France, [17]CNRS UMR8068 Temps, , France, [18]Chair Human Genomics and Evolution, Collège de France, , France, [19]Current affiliation: Department of Evolutionary Biology and Environmental Studies, University of Zurich, , Switzerland

Polynesia exhibits some of the highest prevalence rates of metabolic disorders. This elevated risk has been hypothesized to arise either from natural selection favoring disease-associated alleles during periods of food scarcity or from founder effects during Polynesian settlement. Although a few high-frequency variants associated with obesity and type 2 diabetes have been identified in Polynesians, the evolutionary origins of their increased metabolic risk remain elusive. To address this question, we generated whole-genome sequences and collected phenotypic information from 1,761 individuals across 24 Polynesian islands. We first searched for genomic signatures of ongoing selective sweeps and identified candidate loci, including TARBP1, involved in amino acid metabolism, which carries a non-synonymous variant present at a frequency of 30-50% in Polynesians but rare or absent elsewhere. In contrast, variants associated with lipid metabolism showed no evidence of strong selection, including the CREBRF variant and a new variant in ANGPTL8, suggesting a predominant role of genetic drift in shaping their frequencies. We further evaluated polygenic adaptation across metabolic traits, using tests for excess population variance in polygenic scores and for the relationship between GWAS effect sizes and selection coefficients, estimated from ancestry-masked genealogies with PALM. Across all analyses, we found no evidence for local polygenic adaptation, supporting the view that metabolic traits, including those underlying obesity and type 2 diabetes, have largely evolved under neutrality in Polynesians. Our results underscore how studying understudied populations can provide key insights into the evolutionary sources of phenotypic variation in health and disease.

[B330] FKBP5 Genetic Variants Under Selection Are Associated with Immune and Metabolic Phenotypes in the Taiwanese Han Population

Mei-Ling Kang[1], I-Hui Lee[2,3], Yoko Satta[4], Wen-Ya Ko[1,5]
[1]National Yang Ming Chiao Tung University, Taipei, Taiwan, [2]Institute of Brain Science, School of Medicine, National Yang Ming Chiao Tung University, Taipei, Taiwan, [3]Department of Neurology, Neurological Institute, Taipei Veterans General Hospital, Taipei, Taiwan, [4]Department of Evolutionary Studies of Biosystems, SOKENDAI (The Graduate University for Advanced Studies), , Hayama, Japan, [5]Research Center for Health and Technology Development of Indigenous People of Taiwan, National Yang Ming Chiao Tung University, Taipei, Taiwan

FKBP5 regulates key components of the stress response that influence survival, reproduction, immune function, and behavior, core determinants of evolutionary fitness, yet the evolutionary forces shaping its genetic diversity in human populations remain poorly understood. Identifying variants under positive or purifying selection can reveal functionally important alleles contributing to variation in FKBP5-related diseases. Here, we integrate evolutionary genetic analyses with functional association approaches to investigate how natural selection has shaped genetic variation at FKBP5 in the Han Taiwanese population. We analyzed 2,274 variants within the FKBP5 locus from whole-genome sequencing data of 1,485 individuals in the Taiwan Biobank. Comparisons of site frequency spectra (SFS) across SNP classes stratified by CADD score (a proxy for predicted deleteriousness/functional importance) identified 217 variants (CADD ≥ 10) enriched for rare alleles relative to synonymous variants, consistent with purifying selection. To test for recent positive selection, we applied Singleton Density Score (SDS) analysis and detected a strong signal at the intronic variant rs75552415 (SDS = 4.31, $p = 8.34 \times 10^{-6}$). Phenome-wide association analysis in 99,688 individuals across 34 traits revealed that rs75552415 is significantly associated with serum glutamic oxaloacetic transaminase (SGOT; $p = 1.34 \times 10^{-6}$), implicating liver-related metabolic function. Integration with public functional datasets further indicated enrichment for lymphocyte-specific expression quantitative trait locus (eQTL) signals among candidate variants under purifying selection. Linkage disequilibrium analyses confirmed independence from the neighboring major histocompatibility complex. Together, these results suggest that natural selection on FKBP5 contributes to immune and metabolic trait variation.

[B331] Showcasing evolutionary patterns in marine phytoplankton with unresolved dispersal patterns through whole genome based molecular phylogenies of polar Chlamydomonas sp.

Michael Dills[1,2], Dr. Shripathi Bhat[1,2], Dr. Kim Præbel[1,2]
[1]Uit-the Arctic University Of Norway, Tromsø, Norway, [2]The Norwegian College of Fishery Science, Tromsø, Norway

Chlamydomonadaleans are a globally distributed clade of green algae populating nearly all photic habitats from sea ice to temperate soils. Along with their key role as primary producers in biogeochemical processes, these organisms offer valuable insights into evolutionary processes, dispersal patterns, and inter-species gene flow within globally distributed phytoplankton species. Despite being isolated from every major ocean basin, little is known about marine Chlamydomonas strains – especially those found in the Arctic. This project used whole genome sequencing and comparative genomics to characterize adaptive signatures across a selection of polar adapted marine Chlamydomonadaleans; providing a resource for taxonomy, as well as insights into the ecology of marine clades and their adaptations to the extreme latitudes of Earth's oceans.

[B332] Inferring the history of gene copy number evolution

Moritz Otto[1]
[1]University Of Cologne, , Germany

Gene duplication plays a crucial role in the adaptive evolution and diversification of organisms by creating extra copies of genes that can evolve new functions while preserving the original. Duplicated genes can become fixed in populations or appear as copy number variants. Selection can act on both, gene copy number as well as sequence conservation due to functional constraints.
Here, we present a modeling framework that infers selective pressure (s) and recombination rate (r) directly from gene copy number distributions, using a fitness function favoring genetic diversity combined with an unequal recombination model.
Assuming a pure duplication process, we further apply a random-walk approach to estimate duplication rates (d) and to date duplication events, allowing us to distinguish between few ancient and many recent duplications that yield identical copy number distributions.
Finally, we implement backward-in-time structured coalescent simulations, reinterpreting population structure as genealogical distance defined by copy number differences. We apply this model to the NB-ARC domain counts of NLR genes in A. thaliana to infer the number and times of duplication events that have led to the present day copy number distribution.

[B333] Joint effects of balancing selection and population bottlenecks on the evolution of a regulatory region of human anti-viral APOBEC3

Naoko Fujito[1,2], Sundaramoorthy Revathidevi[2,4], Yoko Satta[3], Ituro Inoue[2]
[1]Brain Research Institute, Niigata University, Niigata, Japan, [2]National Institute of Genetics, Mishima, Japan, [3]SOKENDAI , Hayama, Japan, [4]SRM Institute of Science and Technology, Kattankulathur, India
Human anti-viral APOBEC3s are crucial components of the innate immune response, playing a key role in inhibiting viral replication and proliferation by inducing mutations in viral genomes. In this study, in a regulatory region of ~16 kb in the APOBEC3 gene cluster on human chromosome 22, we identified three distinct haplogroups that are maintained at high frequencies in both African and non-African populations. Despite the long persistence of these haplogroups, one of which is shared by archaic hominins, the nucleotide diversity within each haplogroup was surprisingly low in non-Africans. Genome-wide empirical distribution and coalescence-based simulation tests showed that the simultaneous observations of low within-haplogroup diversity and high between-haplogroup divergence were incompatible with neutrality. We also identified an ancestral sequence group exclusively in African populations. To explain these features and elucidate the underlying evolutionary mechanisms, we performed forward simulations to model the joint effects of balancing selection and population bottlenecks. The results demonstrated the operation of balancing selection over the past ~1 million years, as well as a hitherto unexplored reduction in within-haplogroup diversity. The only unexplained observation was the low diversity within African haplogroup I, which is unaffected by the non-African-specific bottleneck. We hypothesized that an extra haplotype turnover within haplogroup I occurred prior to the Out-of-Africa dispersal of modern humans. This study highlights the functional importance of the APOBEC3 regulatory region in terms of evolutionary conservation in Hominidae, a target of natural selection in modern humans, and its relevance in GTEx eQTL.

[B334] Population Genomics Analysis of Rhynchophorus ferrugineus across its native and invasive range

Neelu Begum[1], Dr Mariana Lyra[1], Theo Busschau[1], Dr Gabriel Riano[1], Stephane Boissinot[1]
[1]New York University Abu Dhabi, Centre of Genomics and Systems Biology, United Arab Emirates

The red palm weevil (RPW; Rhynchophorus ferrugineus), is a pest species that bores into the soft tissue of palms, eventually resulting in the death of the host plant. The RPW is known to feed on 46 different palm species. It is native to tropical Asia but has spread to many parts of the world with a wide range of climates. Here, we present a comprehensive analysis of genomic variation in populations of RPW from more than 15 countries, covering most of the range of this species and including populations from the native range as well as invasive populations. We compare whole-genome variation to determine the demographic history of the range expansion and to identify the genetic basis of adaptation to novel habitats during the expansion.

We characterized population structure and generated phylogenetic trees for the mitogenome and Y chromosome. Our analysis reveals distinct genetic clusters, each with different demographic signatures, providing evidence of founding events from two major source regions. Our demographic inference shows that a stepwise invasion scenario is too simplistic. Instead, more complex histories involving serial introductions into the Middle East, followed by secondary dispersal into surrounding regions, are better supported by the data. By integrating population genomics with invasion history, this study reconstructs the spread of the RPW and generates concrete hypotheses that can be integrated into targeted surveillance of this invasive pest and the design of improved pest management and control strategies.

[B335] Linkage disequilibrium scaling improves robustness and power to detect genomic regions under selection in genetically structured species

Petri Kemppainen[1], Professor Frédéric Guillaume[1]
[1]Department of Organismal and Evolutionary Biology, University of Helsinki, Helsinki, Finland
Understanding the genetic basis of adaptive evolution is central to predicting evolutionary responses to environmental change. Genome scans and genotype–environment association methods are widely used to detect loci under selection, yet they often ignore correlations among loci (linkage disequilibrium, LD), reducing power with increasing genetic structuring and demographic complexity. This limits their use to species with high connectivity and strong signals of selection. Here, we introduce a novel LD-scaled association statistic, F', which incorporates local LD directly into genotype–environment association tests using a fast permutation-based quantile transformation. Using forward-in-time simulations with known ground truth, we evaluate F' across two widely used methods—latent factor mixed models (LFMM) and EMMAX—and assess performance at the level of outlier regions rather than individual SNPs. We further integrate uncertainty in parameter choice and inference method using a consistency-based framework. Across simulations, LD-scaling and joint inference substantially increased detection power and robustness, yielding up to an order-of-magnitude improvement under strong genetic structuring and nearly doubling performance on average across scenarios. Applying this framework to three- and nine-spined stickleback datasets, we recover well-established genomic regions associated with parallel marine–freshwater adaptation with markedly reduced background noise compared to standard analyses. Together, our results demonstrate that explicitly leveraging LD structure and integrating over analytical uncertainty provides a powerful and computationally efficient extension to genome scans for selection in highly genetically structures species, improving robustness and interpretability in both simulated and empirical settings.

[B336] Using phylogenetics to determine the fitness cost of resistance in viruses and bacteria

Pleuni Pennings
[1]Université De Montpellier, Montpellier, France

It would be great if we knew which drug resistance mutations and which drug resistance genes come with a fitness cost.
This would help us better understand the dynamics of drug resistance in pathogen populations and it would help us plan interventions to reduce the burden of resistance.
However, the question of fitness costs of resistance has been surprisingly hard to answer, except in a few cases. If the cost is fairly small (a few percent per year) and if it mostly impacts a pathogen's ability to do host-to-host transmission, it may be impossible to measure such a cost in the lab.
Instead, I propose to apply a combination of population genetic and phylogenetic methods to large, existing databases of pathogen genomes and phenotypes. This will help us detect selection against resistance genes and mutations in viruses (HIV) and bacteria (E. coli).
Using hand-drawn slides and handouts I will present the problem, a possible solution and some new results on HIV and E. coli.

[B337] Tracing the impact of natural selection in admixed human populations from Mexico.

Ram Gonzalez-Buenfil[1], MSc. FERNANDA MIRÓN-TORUÑO[2], Dr. Austin Reynolds[3], Dr. Andres Moreno-Estrada[1]

[1]Aging Research Center, Cinvestav Sede Sur, Center for Research and Advanced Studies of the National Polytechnic Institute, Mexico City, Mexico, [2]Department of Biology, Queen Mary University of London, London, United Kingdom, [3]Department of Microbiology, Immunology, and Genetics, University of North Texas Health Science Center at Fort Worth, , United States of America

Events of admixture (i.e. gene flow between populations) contribute to shaping human genetic diversity, often exposing populations to bottlenecks, novel environments, and selective pressures. While its role in demographic history is well studied, admixture's contribution to local adaptation remains underexplored. Present-day Mexicans descend primarily from Indigenous Americans and Europeans who began admixing ~500 years ago, following a period of population collapse and reorganization. We hypothesize that adaptation in modern Mexicans may reflect directional selection on ancestry-specific variants that arose either pre- admixture or rapidly increased in frequency afterward. Using genotype data from 6,000 individuals in the Mexican Biobank, sampled across all 32 states and genotyped at 1.8 million SNPs, we applied admixture-aware methods based on local ancestry patterns and allele frequency shifts to detect selection signatures across three distinct geographic regions. We find that European ancestry in the North and Central regions is enriched in immune-related pathways, whereas Indigenous American ancestry in the North is enriched in keratinization pathways, involved in skin physiology. In the Mayan region, European ancestry is enriched in pathways involved in vitamin K–dependent coagulation and gamma-carboxylation, both sensitive to dietary intake. Finally, association analyses with Mexican Biobank phenotypes link candidate regions to lipid and glucose-related traits, inflammation, and blood pressure. These findings reveal that natural selection in Mexico has acted in an ancestry- and region-specific manner, shaping distinct biological processes linked to present-day health, and illustrate how selection can operate in the context of recent admixture through a dynamic relationship with gene flow.

[B338] Fine-scale genetic structure and selection signatures underlying disease susceptibility in South Asia

Rubén Muñoz Miranda[1]
[1]Centro de Ciencias Genómicas, Cuernavaca, Mexico, [2]Wellcome Sanger Institute, Hinxton, United Kingdom, [3]Centro de Ciencias Genómicas, Cuernavaca, Mexico, [4]Laboratorio Internacional de Investigación sobre el Genoma Humano , Querétaro, Mexico, [5]Wellcome Sanger Institute, Hinxton, United Kingdom, [6]Centro de Ciencias Genómicas, Cuernavaca, Mexico

Despite the high genetic diversity of South Asians, their representation in genetic studies remains limited. This genetic diversity reflects multiple cultural groups, historical migrations, and recent endogamous practices, offering a key framework for exploring local adaptation in structured communities. Using whole-exome sequencing data (n = 25,049) and Global Screening Array data (n = 28,898) from the East London Genes and Health cohort, we analyzed the genetic structure of British Pakistani and Bangladeshi individuals using Louvain clustering on Identity-by-descent networks to identify genetic communities with biological relevance, without assuming predefined groups. Our findings show that although Pakistanis and Bangladeshis share similar ancestry components, they exhibit distinct genetic structures, with Pakistani genetic communities showing clearer substructures. This substructure partially overlaps cultural structure at some resolutions but lacks strict correspondence due to complex demographic histories. We then applied the Population Branch Statistic to these genetic communities to identify natural selection signals at the gene and SNP level, revealing several candidate genomic regions associated with complex traits, including skin pigmentation (SLC24A5/rs1426654), triglyceride and cholesterol levels (ANGPTL8), hemolytic disorders such as jaundice (SCLO1B1), and vitamin B12 levels (CD320/rs2232780). Furthermore, oligosaccharide glucosidase activity was enriched among genes and SNPs at extreme PBS values (> 99.5 percentile) implicated in glucose metabolic pathways, and intimately related to type 2 diabetes disease burden. These findings provide novel insights into the significance of evolutionary pressures, for example through the thrifty gene hypothesis, in shaping variations in metabolic and other traits associated with common diseases today.

[B339] Signatures of selection in admixed Indonesian cattle breeds

Sabhrina Gita Aninta[1], Xi Wang[1], Rasmus Heller[1]
[1]University of Copenhagen, Kobenhavn N, Denmark

Adaptive introgression is known among the domestic livestock breeds in Asia as introgression from wild relatives which assist local adaptation to their environment alongside many other traits desired by humans. In this study, we characterise selection signals across the genomes of admixed Indonesian cattles (Bos indicus x javanicus). As a cattle breed with history of introgression for highly divergent species (FST ~0.6), detecting positive selection signals on these breeds require approaches that account for the intermediate allele frequency often found in admixed species. In inferring selection on such regions, common practice is to look for outlier regions of extreme ancestry from the minor parent ancestry. However, in doing so, we missed signals of selection in other regions. To investigate the full pattern of selection in the admixed Indonesian cattles, we used population branch statistics (PBS) to scan 90 whole genome sequences of four admixed Indonesian cattle breeds against their parent ancestry, B. indicus (zebu cattle) and B. javanicus (bali cattle). From these four, two breeds come from one island (Aceh and Pesisir from Sumatra Island) and the other two from another Madura and Pasundan from Java Island), presenting an opportunity to evaluate how different selection signals repeated within similar ancestry mix. Corroborating the genes found in previous studies with PBS and other traditional selection scans including admixture-aware ones (i.e. Q95 and AdaptMix), we compare signals found in other breeds of domestic species and establish a comprehensive genomic resource for a group of admixed cattle breeds from the tropics.

[B340] Evolutionary consequences of distyly loss in Linum: A shift toward higher selfing rates?

Zoe Postel[1], Panagiotis-Ioannis Zervakis[1], Dr Marco Fracassetti[1], Aleksandra Lovsik[1], Matias Wanntorp[1,2], Lucile Soler[3], Allison Churcher[4], Aelys M. Humphrey[1], Tanja Slotte[1]
[1]Department of Ecology, Environment and Plant Sciences, Science for Life Laboratory, Stockholm University, Stockholm, Sweden, [2]Department of Medical Biochemistry and Microbiology, Uppsala University, Uppsala, Sweden, [3]SciLifeLab, National Bioinformatics Infrastructure Sweden (NBIS), Department of Medical Biochemistry and Microbiology, Uppsala University, , Sweden, [4]Department of Plant Physiology, National Bioinformatics Infrastructure Sweden, Science for Life Laboratory, Umeå University, , Sweden

Flowering plants exhibit diverse mating systems that shape genetic diversity, adaptability, and fitness. Distyly is a floral polymorphism promoting disassortative pollination between two morphs with reciprocal herkogamy. Its breakdown is often associated with the loss of self-incompatibility and floral dimorphism and is frequently assumed to lead to increased self-fertilization in the resulting homostylous lineages. We studied the genomic consequences of distyly breakdown in wild flax (Linum spp.), a genus in which distyly has been lost independently at least six times. We investigated these consequences at two complementary scales. First, we tested whether this loss is associated with relaxed selective pressure as expected with increased selfing, at a broad phylogenetic scale. Using divergence data from nuclear and chloroplast genes of 13 distylous and six homostylous Linum species, we found mixed evidence for reduced efficacy of selection in homostylous taxa, potentially reflecting variation in the timing of mating system shifts or the absence of a systematic increase in selfing rates following loss of distyly. Second, we examined population-level consequences using high-quality genome assemblies of two closely related species: the distylous outcrosser Linum perenne and the homostylous Linum leonii. Population genomic analyses and selective pressure estimates revealed that, contrary to expectations and other results in the genus, L. leonii exhibits low selfing rates, with genomic signatures better explained by demographic processes than by mating system shifts. Together, our comparative approach highlights variable evolutionary pathways following distyly loss and refines selective pressure expectations and genomic signatures for this widespread evolutionary transition in flowering plants.

[B343] EXPLORING BULL KELP MICROBIAL DIVERSITY ACROSS AN ENVIRONMENTAL GRADIENT IDENTIFIES EUKARYOTE MICROBIOTA AND UNRAVELS ASSOCIATED METABOLIC PATHWAYS.

Etienne Emonot[1], Maximiliana Bogan[2], Doctor Coraline Rousseau[2], Pauline Giraud[3], Doctor Catherine Leblanc[1], Doctor Ludovic Delage[1], Professor Catherine Pfister[2]
[1]orbonne Université, CNRS, UMR 8227, Integrative Biology of Marine Models, Station Biologique de Roscoff, 29680 Roscoff, France, Paris, France, [2]University of Chicago, Department of Ecology & Evolution, Chicago IL 5 60637, USA, Chicago, USA, [3]Université de Rennes, CNRS, UMR 6074, Institut de Recherche en Informatique et Systèmes Aléatoires, Rennes 3500 Rennes, France, Rennes, France
Kelps are foundational species forming vast underwater forests on temperate and arctic coasts. These forests provide a unique habitat for marine species. The kelps themselves host microbial communities on their blades. Very little is known, however, of the metabolisms mediating interactions or how and when they evolved. The bull kelp, Nereocystis luetkeana, grows all along the north eastern American Pacific Ocean coast, and several beds have been studied in the Puget Sound, an inland sea connecting Seattle to the ocean. The different kelp beds are on a gradient of nutrients, temperature, salinity and anthropization making them a very interesting population to explore the metabolic adaptation of their microbiota. Their prokaryotic microbiota has been described along the gradient, but eukaryotic microorganisms associated with these kelps remained unknown. Metabarcoding analyses using the Parada 16S/18S specific primers allowed us to identify core microbes as well as opportunist eukaryotes and bacteria on the blades of N. luetkeana over five sites in the Puget Sound, following the environmental gradient up to Seattle. We found that microbial diversity on the blades was driven by the gradients, while tissue age had a smaller effect. Eukaryotic epibionts were found closer to the sea while green algae preferred inner city beds. Then, using the EsMeCaTa metabolism prediction tool, we were able to predict metabolic functions of each microbial community, and which taxon contributed to which pathway. We draw a complex picture of the communities on the kelp's blade as well as formulate hypotheses on the metabolites mediating such interactions.

[B344] Integrated Host-Microbiome Contributions to Terrestrial Adaptations in Mangrove Sesarmid Crabs

Ka Wing Karen Wong[1], Tom Kwok-Lun Hui[1,2], Irene Ching-Nam Lo[1], Prof Ling Ming Tsang[1]
[1]Simon F. S. Li Marine Science Laboratory, School of Life Sciences, The Chinese University of Hong Kong, , Hong Kong, [2]The Hong Kong Branch of the Southern Marine Science and Engineering Guangdong Laboratory (Guangzhou), HKUST, , Hong Kong

Terrestrialization has evolved repeatedly in brachyuran crabs and is traditionally framed as morphological and physiological innovations addressing multiple terrestrial stressors. Recent evidence highlights substantial microbiome contributions, shifting the perspective towards crabs and their microbiota as integrated holobionts. Mangrove sesarmid crabs, which span a wide gradient of terrestriality and dominate as herbivores, provide an ideal system to test this framework.

We investigated into two key aspects of terrestrial adaptation: nutrition through herbivory and ammonia tolerance during aerial exposure, through host transcriptomics and microbiome analyses. For herbivory-related adaptations, transcriptomes of the host hepatopancreas (site of active enzyme production) showed elevated constitutive expression of lignocellulolytic and detoxification genes (e.g. glycosyl hydrolases and UDP-glucuronosyltransferases) in sesarmids relative to non-herbivorous crabs, reflecting enhanced capacity for vascular plant digestion and xenobiotic processing associated with herbivory. Meanwhile gut microbiomes were enriched in lignocellulose-degrading pathways, supporting cooperative digestion as holobiont.

For ammonia management, gill transcriptomes of sesarmid crabs revealed higher expression level of genes involving in ammonia conversion than marine crabs, illustrating the molecular underpinnings for ammonia tolerance during aerial exposure. Functional contribution from gill microbes is reflected by enriched nitrogen-transforming taxa in sesarmids than non-sesarmids. Significant phylosymbiosis signals in both gut and gill microbial communities with host crabs and detection of host-microbe co-diversification signatures further suggested long-term evolutionary coupling between crabs and their symbionts. Overall, these results demonstrated that sesarmid crabs achieve terrestrial herbivory and aerial ammonia tolerance through both host adaptations and microbial functions across digestive and respiratory interfaces, revealing holobiont-level mechanisms underlying terrestrial adaptations.

[B345] Invisible engineers: distribution and diversity of nutrient-cycling prokaryotes associated with marine invertebrates

Maggie Georgieva[1], Mark Blaxter
[1]Wellcome Sanger Institute, Hinxton, United Kingdom

Symbioses with prokaryotes are essential for the survival of many marine organisms: this is especially apparent in deep-sea chemosynthetic ecosystems, where bacteria use chemically-reduced fluids to nourish hosts such as molluscs, sponges and annelids. While it is now known that chemosynthesis is widespread and occurs throughout ocean habitats and depths, the distribution of nutrient-cycling processes more generally, and their occurrence in symbioses with invertebrates across a range of marine environments, is not well resolved: this is key to understanding ocean ecosystem function as well as nutrient flow within the ocean. The Tree of Life Programme, which aims to sequence reference genomes for all eukaryotes on Earth, has generated over 1000 genomes for marine invertebrates so far, with the majority also including accidental sequencing of genetic information for associated prokaryotes termed ‘cobionts’. In this study, this comprehensive dataset is leveraged to reconstruct cobiont genomes, explore their diversity and metabolic potential, and ultimately to uncover the extent of associations between marine invertebrate hosts and bacteria and archaea capable of nutrient-cycling metabolisms such as nitrogen fixation and sulfur oxidation. Preliminary results reveal that bacterial clades that are typical symbionts of hydrothermal vent animals also occur in non-vent sponges, while Nitrosopumilus ammonia-oxidising archaea that are frequent symbionts of sponges also occur in some annelids, bryozoans and cnidarians. Uncovering their roles will provide vital insight into interdependent ocean communities during a critical time in Earth’s history. We thank colleagues in the Aquatic Symbiosis Genomics project and the Darwin Tree of Life project for data generation.

[B346] The effect of host genetic bottlenecks on microbiome variance

Niek Barmentlo[1], Prof. Jacintha Ellers[1], Dr Christina Hvilsom[2], dr. Mirte Bosse[1]
[1]Amsterdam Institute for Life and Environment (A-LIFE), Vrije Universiteit Amsterdam, Amsterdam, Netherlands, [2]Globe Institute, University of Copenhagen, Copenhagen, Denmark

Population reductions typically lead to a reduction in overall population genomic diversity. As such, studying wildlife populations with a history of population reductions allows for assessing the effect of genetic variance on phenotypic variance. Recent experimental evidence suggests that variance in microbiome composition is also dependent on host genetic variance. In particular the microbial composition of the mucosal layer of the mammalian intestines is thought to largely be affected by host genetics, and typically shows high fidelity to host taxonomy. Hosts can affect the gut microbial composition through immune functioning, which itself is affected by host genetics. The current research aims to establish a link between population level genetic diversity, and the number of bacteria supported by the entire population in the gut. We set out to compare two related free-living wild boar populations; one in Southern Germany (n=17) and a historically bottlenecked population in Denmark (n=19). Genomic diversity of the Danish population is known to be still impoverished, in particular at loci related to immune functioning. Wild boars were DNA sequenced for host DNA and bacterial 16s rRNA gene. For the 16s amplicon sequencing, we sequenced both a theorized host controlled microbial community (mucosal ileum) and a more diet based microbial community (caecum fecal matter). Theory suggests that the mucosal ileum microbial community of the bottlenecked Danish population will prove less diverse than the German population. This pattern is not (or less pronounced) expected in the caecum fecal matter due to limited host genetic control.

[B347] Coral holobionts evolve in response to a devastating disease

Sarah Gignoux-Wolfsohn[1], Caroline DeSouza[1], Nikea Ulrich[1], Felicia Aronson[1], Lauren Fuess[2], Brooke Sienkiewicz[1]

[1]University Of Massachusetts Lowell, Lowell, United States, [2]Texas State University, San Marcos, United States

Microbial members of the holobiont provide diverse mechanisms by which their longer-lived hosts can evolve in response to rapid environmental change, quickly imparting fitness benefits to the holobiont as a whole. Corals have long been upheld as a model holobiont due to their obligate associations with endosymbiotic dinoflagellates in the family Symbiodiniaceae, as well as their highly diverse microbial communities. The novel infectious disease stony coral tissue loss disease (SCTLD) is currently acting as a strong selective force sweeping through Caribbean corals. Unlike other coral diseases, SCTLD afflicts over 20 species of coral, with disease severity and mortality varying by species. In some locations, outbreaks of this disease have killed up to 90% of corals, giving it the potential to irreversibly change the composition of Caribbean reefs. In spite of this devastation, some corals continue to survive disease exposure, suggesting long-term resistance or tolerance. We are using a natural experiment tracking coral health statuses and changes in the microbiome before, during, and after an SCTLD outbreak to explore holobiont evolution in response to an introduced disease. We find differences in both the Symbiodiniaceae and bacterial communities on the taxonomic, gene, and SNP level across resistant and susceptible colonies. Some of these differences existed before disease introduction, suggesting selection on standing genetic (microbial) variation, while others are more plastic. This work demonstrates mechanisms of rapid evolution in response to disease in multiple members of a marine holobiont.

[B348] Contaminant Detection for the Study of Host-associated Microbial Signals in Historical Specimens

Siu-kin Ng[1], Dr Rafal Gutaker
[1]Royal Botanic Gardens, Kew, London, United Kingdom

The study of host-associated microbiomes in historical herbarium samples offers a unique opportunity to investigate the anthropogenic influences on plant-microbe interaction over past centuries. However, identifying meaningful microbial signals is complicated by post-mortem contamination introduced during collection, processing, and long-term preservation in museums and herbaria. Consequently, microbial profiles observed in historical specimens are a mixture of taxa originating from multiple sources. DNA damage patterns can aid in identifying reads derived from recent microbial contaminants, but they have limited power for detecting older contaminants. Duplicate specimens preserved in multiple herbaria, as well as multiple specimens curated by the same collector, when analyzed in conjunction with associated metadata (e.g. collection time and place, plant physiological and ecological information), can provide important clues about post-mortem contamination. Without a priori knowledge about the distributions and the underlying structure of microbial taxa (both host-associated microbes and contaminants) in historical samples, a machine-learning approach offers greater flexibility for detecting post-mortem contamination in this situation. Specifically, we devised an abstract probabilistic framework that enumerates plausible scenarios of post-mortem contamination in historical collections. Within this framework, we synthesized simulated metagenomic datasets that recapitulate these scenarios and used them to train and cross-validate a variational autoencoder model for the effective detection of microbial contaminants in real-world datasets. Overall, this approach advances our ability to distinguish microbial interactions that were biologically relevant during the life of host organism from post-mortem contamination, thereby unlocking museum collections as a valuable resources for studying temporal changes in microbial interactions.

[B349] Measuring selection on rare disease using AI phenotype imputation in electronic health records (EHR)

Daniel Jordan[1], Emily Sun[1], Ron Do[1]
[1]Icahn School Of Medicine At Mount Sinai, New York, United States

Measures of evolutionary constraint or conservation are often treated as evidence for variants or genes being causal to disease, especially in the context of rare genetic disease or rare variants, where it may be difficult to collect enough observations to test association with disease directly. We have developed RarePT, an AI phenotype imputation method to detect undiagnosed or misdiagnosed rare disease in large population datasets. Using AI imputed phenotypes, we conducted a burden association test to identify genes where rare (MAF < 1e-5) variants are associated with rare phenotypes in the UK Biobank, without relying on signals of selection as a proxy for function. We identified 301 genes associated with 229 rare phenotypes, including both known and novel associations. To validate the accuracy of these associations, we queried an LLM to compare associations to previously annotated gene-phenotype annotations. We found that significantly more associations corresponded to previously known annotations than expected by chance ($p < 0.01$, permutation test). Next, we assessed the relationship between rare disease associations and evolutionary constraint measured in gnomAD. Counterintuitively, we observed that genes associated with rare disease are not significantly more constrained than other genes. However, we found that constraint varies widely between phenotype categories, with the most constraint observed in pregnancy-related, genitourinary, and musculoskeletal phenotypes and the least in hematological/immune, respiratory, and gastrointestinal phenotypes. This analysis demonstrates the potential of AI-derived phenotypes to identify the targets of natural selection, and points to the potential of future AI approaches to define these targets more precisely.

[B350] Uncovering Non-Identifiable Demographic Histories using Generative AI

Ekaterina Noskova[1], Konrad Lohse
[1]University Of Edinburgh, Edinburgh, United Kingdom
Reconstructing demographic history from genetic data provides key insights into population size changes, splits, and migrations. However, demographic inference often faces the problem of model non-identifiability, where distinct histories generate similar genetic patterns. Generative AI is well suited for uncovering complex structures and subtle patterns in high-dimensional data, making it a promising approach to investigate this problem. I will present how I plan to use generative AI models to explore the space of indistinguishable demographic scenarios and how it can provide new perspectives on demographic inference.

[B351] Accurate and flexible reconciliation with neural networks

Enzo Marsot[1], Laurent Jacob[2,4,5], Damien DE VIENNE[1,3,5], Bastien Boussau[1,5]
[1]LBBE, Lyon, France, [2]LCQB, Paris, France, [3]FORTH, Heraklion, Greece, [4]LCQB, Sorbonne Université, Paris, France, [5]CNRS, , France

Phylogenetic reconciliation consists in inferring a set of duplication, transfer and loss events that can explain the discrepancy between a gene tree and a species tree. Several methods address this problem, including ALE and GeneRAX.

Sadly, the models of evolution in use do not account for the non-independence between several gene families, and models accounting for branch lengths information in the species tree or the gene tree are prohibitively costly.

Therefore, we turn to machine learning methods and simulation-based inference. This field consists in simulating data and training a machine learning model to perform inference on this simulated data. The trained model can then be used on real data to infer any information of interest.

Phyloformer and Phyloformer 2 are two recent examples in phylogenetics : these methods enable inferring gene trees from sequences and are trained on simulated alignments and gene trees.

We are trying to view reconciliation as the problem of mapping nodes of a gene tree onto the species tree, thereby mapping the trajectory of gene copies that have left extant descendants. To do so, we train our model by teacher forcing, which consists in letting the model predict the mapping of a given unmapped gene node onto the species tree, given the other already mapped gene tree nodes. During inference, we can sequentially map the gene tree nodes to obtain the reconciliation.

We obtain results competitive with standard reconciliation methods at a fraction of the computing cost, especially when the simulated data diverges from ALE’s assumptions.

[B352] Uncertainty and Interpretability in Neural Network Architectures for Population Genetic Inference

Professor Franz Baumdicker[1], Andrew D Kern, Lei Lei, Sara Mathieson, Jiseon Min, Yuxin Ning, Nathaniel S Pope, Dylan Ray, Daniel R. Schrider, Huiting Xu, Leon Zong
[1]University Of Giessen, , Germany

Machine learning methods are increasingly used in population genetics to perform inference in complex settings where established likelihood-based approaches are infeasible. At the same time, there remain fundamental questions about uncertainty quantification and interpretability.

We present a machine learning architecture that provides uncertainty estimates and clarify how embedding networks extract evolutionary information from genetic data in comparison to established summary statistics.
First, we introduce neural posterior estimation (NPE), a simulation-based inference framework that combines the benefits of Approximate Bayesian Computation (ABC) and supervised deep learning. NPE directly approximates posterior distributions and enables efficient Bayesian inference from high-dimensional data. We show that NPE can be applied both to classical summary statistics and to feature representations learned by embedding networks, such as convolutional neural networks (CNNs) trained on raw genotypes.
Second, we examine the interpretability of CNNs in population genetics. Using post hoc analyses, we investigate how CNNs internally represent genetic variation when trained for tasks such as detecting selection or distinguishing real from simulated data. We find that some classical summary statistics are readily approximated by embedding networks, while others are poorly captured. Moreover, correlations between learned features and traditional statistics can arise not only from the training objective, but also from architectural inductive biases of the networks themselves. Consequently, some learned features should be interpreted with caution and may reflect model design rather than evolutionary signal.
Finally, we highlight how theoretical summary statistic based results can be used to improve and guide the training of neural networks in population genetics.

[B353] Pixel2Phenotype: Hyperspherical Self-Supervised Latent Spaces for High-Throughput Drosophila Morphology

Hyoungjun Park[1], Laura Campama Val[1], Paco Majic[1], Robert Prevedel[1], Justin Crocker[1], Anna Kreshuk[1]
[1]European Molecular Biology Laboratory, Heidelberg, Germany

Advances in genome engineering and high-throughput imaging have created new opportunities to systematically map genotype–phenotype relationships, yet current phenotyping pipelines remain constrained by manual annotations and limited ability to quantify complex 3D morphological variation. We introduce a self-supervised deep learning framework for large-scale morphological phenotyping in Drosophila development using volumetric Optical Coherence Microscopy (OCM). We develop a representation-learning pipeline that embeds 3D samples into a geometry-aware latent space designed to capture continuous phenotypic diversity across genetically perturbed populations. The approach combines contrastive self-supervision with hyperspherical embedding constraints to learn a structured latent manifold that preserves meaningful morphological relationships. Only a small, user-defined subset of volumes is manually annotated into phenotype categories; these sparse labels are used to fine-tune or distill the learned representation without disrupting its global geometry. This enables accurate phenotype classification, discovery of intermediate morphologies, and quantification of phenotypic penetrance and heterogeneity across thousands of samples.

[B354] Evolving Proteins Under Fitness Constraints: Does Complex Protein Modeling Help?

Kaiyu Qiu[1]
[1]Max Planck Institute for Biology Tübingen, Tübingen, Germany
Sequence evolution simulation is a fundamental tool in phylogenetic inference and evolutionary hypothesis testing. However, widely used empirical substitution models (e.g., LG, JTT) assume site independence and lack explicit structural or functional constraints, limiting their ability to capture realistic protein evolutionary dynamics. Meanwhile, modern protein models developed for structure prediction and design offer rich representations of protein fitness, yet their potential for evolutionary simulation remains underexplored.

Here, we present a flexible sequence evolution simulation framework, SeqSimFit, that integrates diverse fitness landscapes, including classical biophysical models, statistical coevolutionary models, and state-of-the-art deep learning–based protein models. By simulating evolutionary trajectories under these constraints, we systematically assess their ability to reproduce key statistical properties of natural protein families. Fitness-guided simulations consistently outperform traditional empirical models, yielding more realistic amino-acid compositions, improved site-specific rate heterogeneity, stronger coevolutionary signals, and family-specific substitution patterns.

The framework further supports complex evolutionary scenarios through customizable fitness functions. As a demonstration, we simulate the evolution of fold-switching proteins using multi-state fitness landscapes that balance competing structural constraints, illustrating how alternative folds can be maintained over evolutionary time.

Overall, this work provides a quantitative benchmark for protein sequence evolution driven by modern fitness models and introduces a general tool for exploring the interplay between protein biophysics, structure, and long-term sequence divergence.

[B355] Toward a Prospective Framework for AI in Evolutionary Biology

Ryan York[1], Prachee Avasthi[1]
[1]Arcadia Science, Berkeley, United States

The last 25 years have been biology's first era of big data, culminating with biological foundation models (BFMs): large-scale ML models that learn directly from raw biological sequences or structures. The success of BFMs is fundamentally constrained by the quality of their training data. Public databases have grown exponentially and opportunistically since 2000, accumulating pervasive imbalances, biases, and pseudoreplication. As a result, BFMs can achieve impressive performance by exploiting redundancy rather than learning generalizable biological principles, leading to diminishing returns as model scale increases. The next era of biology requires a more prospective, inference-driven approach to data collection and model development. We present a Bayesian framework that treats learning as an iterative process and optimizes for an entropy-based measure of information gained (IG) from new observations. Applying this framework to the AlphaFold2 database, we quantify the IG contributed by individual proteomes and demonstrate how these values can guide data acquisition across biological contexts. We find that IG is highly context-dependent: efforts to maximize overall novelty benefit most from sampling eukaryotic proteomes, particularly plants, whereas prokaryotic taxa provide greater efficiency per protein sampled. Accounting for this context-dependency, we show how to estimate a theoretical minimum amount of data required for a given task. These results demonstrate that an inference-driven approach to BFM development is feasible and may substantially accelerate biological discovery while reducing the resources to learn generalizable evolutionary mechanisms.

[B356] A Computer Vision Framework for Quantifying Rapid Adaptation

Stylianos Mavrianos[1], Vera van Santvoort[2], Sven Teurlincx[2], Steven Declerck[2,3], Kathrin Otte[1]
[1]University Of Hamburg, Hamburg, Germany, [2]Department of Aquatic Ecology, Netherlands Institute of Ecology (NIOO-KNAW),, Wageningen, The Netherlands, [3]Laboratory of Aquatic Ecology, Evolution and Conservation, Department of Biology, KULeuven, Leuven, Belgium

Quantifying changes in species composition and population structure is essential for identifying the underlying mechanisms of rapid adaptation. However, the labor-intensive nature of manual specimen identification, counting and body length measurement remains a primary bottleneck in evolutionary biology studies, severely restricting the scale and temporal resolution of experiments. Current methods fail to address the need for high-throughput community-scale analyses; thereby limiting the advances in the field. To address the demand for analysis, we implement a YOLOv12-based computer vision framework designed for the automated analysis of high-resolution biological images. This approach utilizes state-of-the-art deep learning to generate pixel-precise masks, enabling the automated extraction of morphological features, such as surface area, width, and length, which serve as proxies of demographic shifts. By overcoming the constraints of manual data acquisition, this framework allows researchers to analyze massive datasets at a fraction of time and speed while remaining precise. This capability enables more complex experimental designs and higher sampling frequencies, ultimately facilitating a deeper understanding of the selective pressures and mechanisms that drive adaptive responses in changing environments.

[B357] Revisiting estimation of bacterial mutation rates, fitness effect of mutations and resulting

evolutionary dynamics through simulation-based methods

Aurélien Tauzin[1], Antoine Frenoy[2]
[1]Université Grenoble-alpes, Grenoble, France, [2]Université Grenoble-alpes, Grenoble, France
The mutation rate is a key parameter of evolution as it defines the speed at which genetic diversity is generated de novo.
A traditional approach to estimate mutation rate for bacteria is the fluctuation test, which consists in growing bacteria in a non-selective medium and plating them in a selective environment to count resistant mutants. A mathematical model is then fitted on these experimental data to infer mutation rate.
However, this model makes restrictive assumptions regarding the effect of the mutations on the fitness (no effect or fixed effect) and the growth conditions (no death in the culture, no sampling) which do not always represent reality.
We developped a simulation-based method to infer mutation rate from experimental data, taking into account non-standard demographics and fitness effect of the mutation. This was made possible by the development of a very fast forward-in-time simulator, which can accommodate deviations from the classical hypotheses mentioned above, coupled with an MCMC parameter inference method.
We show that this method can infer mutation rate and other parameters (such as fitness effect) with comparable performance than analytical tools in simple scenarios where these tools work, and also works well in scenarios where these other methods do not (including non-constant death rate, distribution of fitness effects, etc).
We further highlight the consequences of considering several mutations conferring the same resistant phenotype albeit with different fitness effects in absence of stress, a classical situation in fluctuation tests that is usually neglected due to the lack of appropriate methods.

[B358] Learning evolutionary constraints across the tree of life using a codon language model

Claudèle Lemay-St-Denis[1], Rachel Kolodny[1]
[1]University of Haifa, Haifa, Israel

Protein-coding sequences encode overlapping signals, including species-specific constraints related to translation, mRNA stability, and co-translational folding. Consequently, proteins with conserved functions across the tree of life can be encoded by very different coding sequences shaped by host codon usage and tRNA availability. Despite their widespread use to generate a coding sequence adapted to a target host, current codon optimization strategies only partly capture these evolutionary constraints and typically operate by modifying a fixed amino acid sequence, making them only partially successful.

Here, we propose an alternative, evolution-inspired framework for generating coding sequences tailored to a target species, and conditioned on protein sequences. We introduce an encoder–decoder AI model that couples protein semantic representations with species-specific codon grammars. The encoder leverages pretrained protein language models to capture functional and structural constraints from the input protein sequence. The decoder, that is also conditioned on the target species, then generates a codon sequence autoregressively. To disentangle protein semantics from species-specific coding biases, we train the model on clusters of homologous sequences sharing the same protein function but originating from different taxa, ensuring that protein–gene pairs are not trivially identical.

This approach enables the systematic generation of coding sequences for proteins that have not evolved in a given species, while remaining consistent with learned evolutionary constraints. Beyond applications in heterologous expression and protein design, our model provides a new tool to probe how evolutionary constraints affect coding sequences across lineages, offering a data-driven framework to study the relationship between protein function and genome evolution.

[B359] Beyond classification accuracy: using model uncertainty to better understand lncRNA sequences

Daniel García-Ruano[1,2], Mikaël Georges[1,2], Saswat Kumar Mohanti[3,4], Rahma Baaziz[2], Kateryna Makova[3,5], Macha Nikolski[1,2], Domitille Chalopin[1,6]
[1]University of Bordeaux, Bordeaux, France, [2]Institute of Cellular Biochemistry and Genetics, CNRS UMR5095, Bordeaux, France, [3]Molecular, Cellular, and Integrative Biosciences, Huck Institutes of the Life Sciences, Penn State University, University Park, , USA, [4]Department of Biology, Penn State University, University Park, , USA, [5]Center for Medical Genomics, Penn State University, University Park and Hershey, , USA, [6]ImmunoConcEpT, UMR 5164, Bordeaux, France
Long non-coding RNAs (lncRNAs) have gained significant attention in recent years, yet distinguishing them from protein-coding transcripts remains challenging, owing to the lack of clear sequence features that reliably separate the two biotypes. The recent GENCODE annotation efforts—revealing tens of thousands of novel lncRNA sequences—together with frequent biotype reassignment due to misclassification emphasize the need to identify lncRNA-specific sequence characteristics.

In our study, we repurpose existing AI-based coding potential classifiers to move beyond standard benchmarking and ask which specific transcripts are misclassified and why. To this end, we train and test 8 tools, and group transcripts based on classification uncertainty and agreement between the models. Moreover, beyond conventional classification features (e.g., transcript sequence, RNA structure, and protein properties), we incorporate genomic context information by evaluating repetitive elements and non-B DNA motifs within transcript exons. This framework allows us to compare sequence properties in distinct transcript groups, including gold-standard transcripts (correct, unanimous and certain classification) and difficult-to-classify sequences (incorrect but consensual classification or high agreement on high uncertainty), refining our understanding of lncRNA versus coding sequence characteristics.

Altogether, by leveraging model agreement and uncertainty together with underexplored genomic features, we identified sequence characteristics of transcripts that are difficult to classify. This novel approach paves the way for the development of better transcript classifiers, but also improves our understanding of the intriguing nature of lncRNA sequences.

[B360] Pan-Mammalian genomic insights into cancer resistance and therapeutic target discovery

Giada Padovani[1], Leslie Smith[1], Nathan Gilman[1], Dr. James Cahill[1], Dr. Kiley Graim[1]
[1]University of Florida, Gainesville, United States

Translating pre-clinical cancer discoveries into effective human treatments is particularly challenging, especially for rare cancers, which often lack enough patients to conduct clinical trials and build effective models. However, identifying model organisms suitable for studying a particular disease can be difficult: pre-clinical trials using the common mouse model have a 92% failure rate in Phase I trials. Utilizing mammals with higher clinical and genomic similarity to human diseases as non-traditional models, such as dog models of breast cancer and sarcomas, can yield more successful results and highlight the promise of comparative oncology. Suitable model organisms for a cancer will be those who share high genomic similarity with humans across genes associated with the given cancer. One way of identifying this genomic similarity is to analyze evolutionary information across mammals. Here, we apply PhyloP across the Zoonomia 241 species alignment to identify conserved and accelerated regions across mammalian species and compare it to a null model with shuffled genomic information. Within each of the 19 Zoonomia orders, we do pairwise comparisons between species and perform enrichment analysis in human cancer genes by statistically evaluating PhyloP conservation and acceleration scores in all COSMIC cancer genes. This allows us to select the most enriched genes related to cancer in each species across the mammalian trees. We identified pan-mammalian mechanisms of cancer resistance and susceptibility by analyzing the evolutionary history of cancer-related genes and pathways. Our results demonstrate that incorporating evolutionary information into model organism selection is a way to identify promising model organisms.

[B361] Training an AI model to predict population extinction based on genomic features

Jasmyn Gooding[1], Dr Thomas Birley[2], Dr Johanna Winder[2], Prof Jim Groombridge[3], Dr Hernán Morales[4], Cock Van Oosterhout[2], Dr Taoyang Wu[1]
[1]School of Computing Sciences, University Of East Anglia, UK, Norwich, United Kingdom, [2]School of Environmental Sciences, University of East Anglia, Norwich, England, UK, Norwich, United Kingdom, [3]Durrell Institute of Conservation and Ecology, Division of Human and Social Sciences, University of Kent, UK, , United Kingdom, [4]Globe Institute, University of Copenhagen, Copenhagen, Denmark, Copenhagen, Denmark

The current biodiversity crisis is marked by rapid, widespread population declines that places many populations at imminent risk of extinction. Existing assessment frameworks emphasise demographic decline and habitat threats, but do not explicitly evaluate longer-term genomic signatures such as genetic erosion, elevated realised load, and loss of adaptive potential. These processes can continue to undermine population viability even after demographic recovery. The question is whether we can predict the risk of future extinction caused by genomic erosion based on genomics data, and whether AI models can help in such assessments.

We use forward-time simulations in SLiM to generate a large synthetic dataset of bottlenecked populations spanning 256 demographic scenarios (varying ancestral size, bottleneck severity and duration, and fecundity), with replicate populations sampled at multiple timepoints before, during, and after decline. Each simulation implements a diploid genome with defined chromosome length and gene architecture (based on the flycatcher chromosome 23), incorporating recombination and mutation. We extract windowed genomic profiles from individuals alongside population-level summary statistics of diversity, inbreeding, and genetic load.

Preliminary analyses indicate that extinction-associated signatures can be detected before bottleneck onset in lineages that later go extinct. We then evaluate an artificial neural network for predicting the long-term fate of populations at each timepoint, achieving consistent high levels of accuracy. We further assess a set of engineered composite genomic features to improve the models. Together, this work provides a proof-of-principle showing it might be possible to use AI models trained on genomics data to predict future extinction risk.

[B362] Machine learning identifies the evolutionary genomic and statistical features that predict cross-population replication of GWAS signals

Joseph Lachance[1], Ujani Hazra[1]
[1]Georgia Institute Of Technology, Atlanta, United States

GWAS have identified hundreds of thousands of trait-associated variants, yet many of these associations fail to replicate across ancestries. It is not fully known why some of these trait associations “travel well” and others do not. Here, we characterized the SNP-level determinants of GWAS replication using well-powered (~105,000 sample) GWAS findings from European and East Asian ancestry cohorts. These analyses focused on twelve independent blood-related traits, with ten traits used as a training set and two traits used as a validation set. SNPs were annotated with nineteen evolutionary, and functional genomic features, and GWAS replication was modeled as a binary classification task using five different machine learning models. Of these machine learning models, XGBoost was best able to predict replicating and non-replicating SNPs. The most informative feature was the genetic variance contribution in the discovery cohort. Statistical genetic features with large SHAP values included effect size, minor allele frequencies, and LD-scores. Evolutionary genomic features (e.g., iHS, XP-EHH, Fst, and GERP scores) had a modest impact on replication prediction. Somewhat surprisingly, functional genomic features (e.g. chromatin accessibility, TF-binding, and eQTL status) were largely non-informative. Local SHAP values revealed that intermediate replication probabilities were often shaped by competing signals, suggesting that non-replication frequently arises from complex trade-offs of multiple features. The framework described here can inform future efforts to prioritize variants that will improve the portability of polygenic risk scores.

[B363] Sequence-Based Detection of Programmed Ribosomal Frameshifts Using HMMs

Margaux Aubel[1], Amit Alon[3], Oded Rechavi[3], Prof Yitzhak Pilpel[2], Paola Laurino[1]
[1]Okinawa Institute of Science and Technology (OIST), , Japan, [2]Weizman Institute of Science, Rehovot, Israel, [3]Tel Aviv University, Tel Aviv, Israel

Programmed ribosomal frameshifting (PRF) can happen when ribosomes switch the reading frame at a specific position of the mRNA at a defined rate. While PRF is a well-characterized strategy in viruses to regulate protein ratios, evidence for PRF in cellular organisms remains sparse. The best-studied example is the antizyme gene in yeast and mammals, where a conserved +1 frameshift is essential to translate the functional protein. Due to the limited number of known PRF genes, especially in non-viral sequences, no general predictor of PRF currently exists.

Here, we present an evolutionary sequence-based framework to predict PRF by combining codon-wise periodic nucleotide conservation with Hidden Markov Models (HMMs). Coding sequences exhibit a characteristic three-base periodicity in nucleotide conservation, reflecting the different selective constraints across codon positions. We quantify this periodic signal using Fourier transformation and train an HMM that infers the most likely reading frame along a sequence to identify and predict candidate PRF events.

We subsequently evaluate performance on well-characterized coding sequences with experimentally verified reading frames, demonstrating the model's ability to recover known PRF genes. Further, we use the model to predict novel PRF sites across a broad range of species. Our results highlight how modeling evolutionary conservation patterns can enable interpretable detection of translational recoding events from sequence data alone.

[B364] AI-based prediction of variant fitness effects in Arabidopsis lyrata

Valentina Burskaia[1], Dr. Polina Novikova
[1]Vlaams Instituut voor Biotechnologie (PSB UGent), Ghent, Belgium, [2]University of Antwerp, Antwerp, Belgium

Genomic language models are increasingly used for variant effect prediction in protein engineering and medical genomics. Here, we assess their utility for estimating fitness effects of standing genetic variation in natural populations of Arabidopsis lyrata. A. lyrata provides an advantageous study system. Its predominantly outcrossing mating system facilitates high-resolution genotype–phenotype mapping, while close phylogenetic relationship to the model species A. thaliana enables the use of extensive functional genome annotation. We leverage deep evolutionary signal from multiple-sequence alignments of angiosperm genes for model training. Model predictions are evaluated by cross-validation against fitness-related metrics derived from distributions of fitness effects (DFE) and genome-wide association studies (GWAS). We analyze two complementary datasets. First, we use a Palearctic-wide panel of more than 1,000 A. lyrata accessions. Second, we incorporate data from in vivo selection experiments.

[B365] Immune genes on the move: adaptive introgression in the European house mouse hybrid zone

Dr. Jamie Winternitz[1], Dr. Victor Hugo Jarquín[2], Dr. Emanuel Heitlinger[3], Prof. John Baines[4,5], Prof. Guila Ganem[6], Prof. Carole Smadja[6]
[1]Hamburg Universität, Hamburg, Germany, [2]Max-Delbrück-Centrum für Molekulare Medizin (MDC), Berlin, Germany, [3]Head of Medical Microbiology, Rhineland-Palatinate State Investigation Office, Landau, Germany, [4]Evolutionary Medicine, Christian-Albrechts-University of Kiel, Kiel, Germany, [5]Max Planck Institute for Evolutionary Biology, Plön, Germany, [6]The Institute of Evolutionary Science of Montpellier (ISEM), Montpellier, France
Introgression can provide an important source of adaptive genetic variation when populations are far from their fitness optimum, such as during range expansion or rapid environmental change, by introducing novel alleles or restoring diversity lost through founder events. Genes under balancing selection, which maintains genetic variation across time and space, are expected to be particularly prone to adaptive introgression when their diversity is beneficial. Immune-related genes frequently experience balancing selection due to heterogeneous pathogen pressure, making them strong candidates for biased introgression. Natural hybrid zones provide powerful systems for testing these predictions because they represent regions of secondary contact where locus-specific introgression can be evaluated against genome-wide ancestry patterns. We test two hypotheses across the European house mouse hybrid zone, where Mus musculus musculus and M. m. domesticus meet, using published genomes and newly generated whole-exome sequencing data from samples spanning two transects. First, range expansion reduces functional genetic diversity, including at immune genes, thereby promoting adaptive introgression. Second, immune genes exhibit stronger introgression than non-immune functional categories and genome-wide background genes. We quantify introgression using cline analyses and compare patterns among innate and adaptive immune genes, sensory perception genes, and nearly 20,000 background protein-coding genes. We confirm that heterozygosity declines with distance from the origin of range expansion in both subspecies and find that immune genes show biased introgression relative to background loci, with effects varying across immune classes and transects. These results support a role for adaptive introgression in maintaining immune diversity during range expansion.

[B366] Evolutionary repair of a broken mitotic entry switch

Chantelle Endeley, Maria Rosa Domingo Sananes[1]
[1]Nottingham Trent University, Cliffton, United Kingdom

Biological processes are often regulated by molecular networks that create system-level behaviours, such as switches and oscillators. Over the last few decades, we have gained significant insights into the function of these systems from both theoretical and experimental perspectives. However, we lack knowledge of how these systems originated and how they continue to evolve. We have taken an experimental evolution approach to analyse how fission yeast cells adapt to defects in the control of mitotic entry. In this organism, mitosis is triggered by activation of the Cdc2-Cdc13 complex, which occurs when the Cdc25 phosphatase removes Wee1-dependent phosphorylation from Cdc2. Positive feedback loops between Cdc2 and its regulators help to ensure a robust and abrupt entry into mitosis by creating a bistable switch. We evolved cells with defects in the switch in four different strains (loss-of-function mutations in Wee1, Cdc25, and Cdc2, and loss of Cdc25 feedback regulation) along with wild-type controls for >250 generations in minimal medium at 30˚C. Across most populations and strains, we observed increases in fitness and changes in cell size, with cells shifting toward wild-type sizes. The most significant changes in fitness and cell size occurred in cells with Cdc25 defects. Interestingly, whole-genome sequencing revealed that cells with different defects in Cdc25 acquired distinct mutations. Specifically, cells with defective, temperature-sensitive Cdc25 acquired mutations in cell cycle regulators in the same control network (Cdc2 and Wee1), whereas cells with defects in Cdc25 feedback regulation mostly acquired mutations in regulatory subunits of the proteasome.

[B367] How can you survive accumulating loads of toxins? – The hidden roles of insect Na,K-ATPase β-subunits

Marlena Herbertz[1], Prof. Dr. Susanne Dobler[1]
[1]University of Hamburg, Hamburg, Germany

The Na,K-ATPase (NKA) is a key transmembrane carrier essential for nervous function, cellular homeostasis, signaling, and steering morphogenetic processes in animals. Cardiac glycosides (CGs) can fatally inhibit the NKA, but they may also be useful for treating heart issues and cancer. While most insect NKAs can be inhibited by CGs, our model system the large milkweed bug O. fasciatus has a long evolutionary history with CG-containing plants and has developed four paralogs of the α-subunit with different activities and resistance levels. This adaptation enables the bug to feed on plants containing CGs and use the toxins for its own defense.

In addition to the well investigated α-subunit the NKA comprises a β-subunit, but its function is poorly understood. In O. fasciatus, four β-subunits exist, and their tissue-specific distribution suggests that they have roles beyond supporting the α-subunit. These subunits may help the organism adapt to environmental changes, but their specific functions remain largely unknown.

I study the roles of β-subunits in septate junction (SJ) formation and CG compartmentalization. Knockdown of the β-subunits cause molting issues and abnormal tracheal morphology, indicating their role in SJ formation. This is also supported by recent colocalization studies which showed an association of the β-subunits with specific SJ proteins. Data from my current study suggest that one of the four β-subunits is involved in CG compartmentalization. Further, the HPLC analyses show that CGs are modified in the hemolymph to produce stronger toxins against vertebrate NKAs, highlighting the bug's ability to handle and utilize CGs effectively.

[B368] Evolution induced state shifts in a long-term microbial community experiment

Mikko Kivikoski[1,2], Johannes Cairns[1,2,3,4], Shane Hogle[3], Sanna Pausio[3], Lutz Becks[5], Ville Mustonen[1,2], Teppo Hiltunen[3]

[1]Department of Computer Science, University of Helsinki, , Finland, [2]Department of Organismal and Evolutionary Biology, University of Helsinki, , Finland, [3]Department of Biology, University of Turku, , Finland, [4]Turku Collegium for Science, Medicine and Technology (TCSMT), University of Turku, , Finland, [5]Aquatic Ecology and Evolution, Limnological Institute, University of Konstanz, , Germany

Environmental perturbations change the composition of species communities. Species can respond to perturbations through evolutionary changes and undergo evolutionary rescue. Evolutionary rescue of one species could affect the interacting species and potentially lead to recovery of the non-perturbed community composition. Here, we present a four-year evolution experiment involving a 23-species synthetic bacterial community propagated in two environments: a control medium and the same medium supplemented with the antibiotic streptomycin. Through combined analyses of community composition and genome evolution, we quantified the temporal changes in species abundances and the evolutionary trajectories of individual community members. The extended duration of the experiment enabled the detection of adaptive mutations and community state shifts that occur only over long evolutionary timescales. We show that community dynamics are environment dependent and reproducible across replicates, and that evolution of streptomycin resistance in a previously streptomycin-sensitive species on its own can induce abrupt community state shifts. Understanding such community-level outcomes of single species adaptation is important for understanding how evolutionary rescue of one species affects the whole community.

[B369] Transposon evolution under contrasting mating systems in Arabidopsis lyrata populations.

Adrian Contreras Garrido[1], Louna de Oliveira[2], Aurélien Petit[2], Sylvain Legrand[1], Christelle Blassiau[1], Jean-Marc Aury[3], Pierre Baduel[2], Vincent Castric[1]
[1]Univ. Lille, CNRS, UMR 8198, Evo-Eco-Paleo., Lille, France, [2]Institut de Biologie de l'École Normale Supérieure, Université Paris Sciences et Lettres (PSL), UMR 8197 CNRS., Paris, France, [3]Genoscope. Génomique Métabolique., Évry, France

The transition from outcrossing to self-fertilisation, a common shift in flowering plants, influences the patterns and distribution of genetic variation, profoundly altering the dynamics of natural selection. Selfing increases homozygosity, decreases the efficacy of recombination, and reduces effective population size. While the effect of selfing on "classical" mutations has been intensively studied, their effects on other genomic elements has been under-examined. Transposable elements (TEs) are mobile genetic sequences that constitute a substantial portion of eukaryotic genomes, affecting gene function, genome structure, and evolutionary processes such as adaptation and speciation. Their abundance and distribution is influenced by a balance between their propagation, host silencing, and the efficacy of selection in removing detrimental insertions. Hence, in contrast to "classical" deleterious mutations,which arise at a uniform and largely constant rate, TEs have their own evolutionary dynamics. Predicting the effect of selfing on the genomic TE content has so far remained challenging.

Here, we generated a genome graph of geographically close North American populations of Arabidopsis lyrata, a subset of which has undergone a switch to selfing to investigate the rates of TE gain and loss within and between outcrossing and selfing populations. We explore these dynamics in a large collection of manually curated and philogenetically distinct sets of transposons. We inform these TE dynamics by inferring the demographic history of these North American A. lyrata populations. Overall, we aim to further increase our understanding of the intricate co-evolution of TE and host during different host mating systems.

[B370] Do accessory chromosomes impact the phenotype of insect-pathogenic fungi?

Aleksandra Gesiorska[1], Andi Wilson[1], Hanne Griem-Krey[2], Michael Habig[2], Henrik H. De Fine Licht[1]
[1]Department Of Plant And Environmental Sciences, University of Copenhagen, Frederiksberg, Denmark, [2]Fungal Evolutionary Genetics, University of Kiel, Kiel, Germany

Accessory chromosomes (ACs) are semi-autonomous genomic elements whose transmission, gene content, and evolutionary dynamics often resemble those of selfish genetic elements. In the fungal genus Metarhizium, AC presence and composition vary markedly among isolates, suggesting a role in niche adaptation. In M. brunneum, isolates differ in insect virulence and extent of mutualistic plant-root association, implying a genetic basis for these varying phenotypes.

To investigate whether ACs drive this phenotypic divergence, we compared two M. brunneum isolates: one lacking ACs and one harbouring two complete ACs. RNA-seq libraries were generated under four biologically relevant conditions: early and late stages of infection on locust (Locusta migratoria) and during the colonisation of the roots of monocot (Hordeum vulgare) and dicot (Arabidopsis thaliana) plants. Differential expression profiling revealed condition-specific transcriptional signatures that reflect both the nature of the host interaction and the distinct gene repertoires conferred by the accessory elements.

We identified accessory genes that are strongly upregulated during either root colonisation or insect infection. Although many have core-genome homologues, their protein sequences are divergent, suggesting functional innovation. Phenotypic assays across varied carbon and nitrogen sources further link expression patterns to metabolic capabilities, underscoring the functional relevance of AC-encoded genes.

Collectively, our findings demonstrate that ACs have an impact on the trade-off between pathogenicity and mutualism in M. brunneum. By producing phenotypic differences on the host organism, ACs may be biasing transmission toward environments where their gene content confers a selective advantage, thereby influencing fungal ecology and evolution.

[B371] Ploidy variation and mating system shifts shape genome evolution in Arabidopsis lyrata

Anna Glushkevich[1], Dr Alison Scott[1], Dr. Polina Novikova[1,2,3]
[1]The Max Planck Institute for Plant Breeding Research, Cologne, Germany, [2]VIB-UGent Center for Plant Systems Biology, Ghent, Belgium, [3]Ghent University, Ghent, Belgium

Whole genome duplications and changes in mating type highly affect genome evolution. For example, polyploidy can increase mutational load and reduce purifying selection because of additional chromosome copies. Transition to selfing, on the contrary, can increase purifying selection due to increased homozygosity. However, a decrease in effective recombination rate can negate this effect.

Here we use 30 high-quality genomes of Arabidopsis lyrata across the species range to study structural variation and signatures of purifying selection in diverse lineages. A. lyrata has undergone several independent transitions to selfing as well as whole genome duplications, making it a useful model for comparing genome evolution under these transitions.

We used the software Pannagram to characterize the whole pool of structural variants (SVs). We identified almost a million SVs across all genomes. Longer SVs, representing mostly transposable element (TE) insertions, had different frequency patterns from shorter SVs, representing random mutations. Siberian lineages, both diploid and tetraploid, showed higher TE insertion rates. The Siberian population underwent a bottleneck, and may have partially lost the ability to control TE insertions. This resulted in higher TE insertion rates and larger genomes (about 10-20 Mb) than in the European populations. Nevertheless, Siberian selfing populations showed lower TE insertion rates and genome size, suggesting a role of mating system for TE activity regulation.

Tetraploids from different lineages demonstrated enhanced both long and short SV numbers. Also, they had more structural variants inside coding sequences of the genes, suggesting relaxed purifying selection in tetraploids.

[B372] How mobile elements structure the distribution of accessory genes

Bram van Dijk[1]
[1]Utrecht University, Utrecht, Netherlands

Across microbial ecosystems, accessory genes are unevenly distributed among mobile genetic elements (MGEs). For example, temperate phages frequently encode host-derived traits (“morons”), fungi harbor mobile chromosomes carrying pathogenicity factors, and genes for antimicrobial resistance are often mobilized, particularly on plasmids. Other genes, however, remain stably chromosomal. Why do some genes become mobile, while others do not?

In this talk, I will discuss the evolutionary and ecological principles that govern accessory gene mobility across different biological systems. Using computational modeling of various systems (primarily microbial ecosystems and fungal plant pathogens) I will show that accessory gene carriage is not incidental, but instead reflects constraints imposed by MGE life cycles, transmission modes, and interactions with host populations. A key insight from these models is that genes tend to become mobilized when they are weakly selected, and that long-term evolutionary dynamics can favor mobilization as a means to enhance either the population's evolvability or the persistence of such traits. These findings have some counterintuitive implications. For example, they suggest that crop rotations in agriculture - while effective at reducing disease outbreaks and improving soil health - may simultaneously select for fungi with greater evolvability, enabling them to infect multiple hosts and rapidly adapt to changing host environments. Taken together, this work identifies general rules that help explain why accessory genes cluster on specific mobile elements across microbes and fungi.

[B373] Adaptive Divergence of Topoisomerase II Drives Satellite DNA-mediated Hybrid Incompatibility

Cara Brand[1]
[1]Rutgers University, , United States

DNA satellites are expansive arrays of noncoding tandem repeats that comprise a substantial fraction of eukaryotic genomes. As the most rapidly evolving class of DNA, satellites were historically dismissed as “junk DNA” with no functional consequences for the organism. However, growing evidence demonstrates that some satellites are selfish, proliferating across host genomes and biasing inheritance at the expense of host fitness. The deleterious consequences of satellite DNA evolution select for adaptive changes in proteins that either suppress selfish proliferation or mitigate its collateral damage. Under this framework, molecular arms races between satellite DNA and interacting proteins can generate incompatibilities between satellites from one species and diverged proteins from another. Consistent with this model, DNA repeat instability and misregulation underlie numerous post-zygotic reproductive barriers, yet the underlying molecular mechanisms remain poorly understood. To explore the diverged molecular machinery and cellular pathways driving species incompatibilities, we experimentally probed a long-studied incompatibility between Drosophila melanogaster and D. simulans that arises from mis-segregation of the D. melanogaster-specific 359bp satellite in hybrid embryos. We show that the D. simulans allele of the essential enzyme Top2 causes this lethal incompatibility. Using interspecies gene swaps combined with genetics and cell biology, we demonstrate that adaptive divergence of Top2 impairs the resolution of 359bp-induced topological stress, leading to catastrophic chromosome segregation defects in hybrid embryos. Together, these findings reveal how molecular arms races driven by selfish satellite DNA can reshape even essential housekeeping genes, with direct consequences for genome stability and reproductive isolation between closely related species.

[B374] The origin, spread, and demise of a recently active mammalian DNA transposon

Harsha Sen[1], Dr. Landen Gozashti[2], Dr. Peiwei Chen[3], Dr. Charles Feigin[4], Dr. Paul Frazel[1], Dr. Cedric Feschotte[3], Dr. Ricardo Mallarino[1]
[1]Princeton University, Princeton, United States, [2]Stanford University, Stanford, United States, [3]Cornell University, Ithaca, United States, [4]LaTrobe University, Melbourne, Australia

Transposable elements (TEs) make up approximately half of most mammalian genomes. While Class I retrotransposons such as long interspersed nuclear elements (LINEs) and endogenous retroviruses show evidence of recent activity in almost all studied mammalian lineages, Class II DNA transposons are thought to be extinct in mammals outside of bats. By analyzing diverse marsupial reference genomes, we have identified recent Tc1/mariner DNA transposon activity in the marsupial sugar glider, Petaurus breviceps. Interestingly, consensus sequence comparison identifies nearly identical transposons in several distantly related Australian amniotes, suggestive of horizontal gene transfer. Using small RNA sequencing, we find that the male germline tissues of the sugar glider mount a robust piRNA response to this recently amplified DNA transposon. piRNAs are short RNAs that, in complex with Argonaute proteins, enable the host cell to silence TEs via mRNA degradation and DNA methylation. piRNA biogenesis involves both sense and antisense TE transcripts, and the genomic source of the latter is not well understood. A large number of antisense piRNAs for this element originate from an antisense transposon insertion into the intron of a host gene. Long-read RNA sequencing analyses are underway to investigate the molecular mechanisms by which this transposon is expressed in the germline, empirically testing the hypothesis that DNA transposon coding sequences are spliced into host transcripts. Altogether, this work identifies a recently active DNA transposon in a mammalian genome, horizontally transferred across Australian amniotes, and will uncover the molecular mechanisms by which it is both expressed and silenced in the germline.

[B375] Investigating three-tiered interactions of ribosomal DNA, transposable elements, and satellite DNA as mechanisms of rapid genome evolution

John Sproul[1], Paul Frandsen, Rachel Christensen
[1]Brigham Young University, , United States

Despite the fundamental roles played by repetitive elements (REs) in genome composition, structure and function, the finer details of how REs impact evolutionary processes remain understudied. We investigate recurring patterns of rapid genome evolution driven by multi-tiered interactions of three repetitive sequence types: satellite DNA (satDNA), ribosomal DNA (rDNA), and transposable elements (TEs). Our study system of ground beetles (Carabidae) shows striking rapid genome evolution such that overall genome content of recently diverged sister groups may show >50% divergence due to rapid turnover of repetitive sequences. We find strong evidence of a recurring process in which fragments of ribosomal DNA (rDNA) have escaped functional rDNA clusters and proliferated extensively in repeat-rich regions. Thus, rDNA is the parent sequence of new satDNA motifs that account for rapid RE evolution. Our analyses implicate R-elements, a broadly conserved superfamily of TEs, as key drivers in this process of rDNA-derived satDNA proliferation. Therefore, our findings strongly suggest a three-tiered network of interactions among satDNA, rDNA, and R-elements that repeatedly drives rapid genomic divergence among species. We use long-read assemblies to clarify the sequence-level structure of recently proliferated rDNA-derived satellite repeats and find frequent formation of higher-order repeat structures, including distinctive, multi-kilobase palindromic structures. We find preliminary data that suggests proliferating rDNA-derived repeats that carry R-element insertion sequences may trigger a positive feedback loop of rapid genome evolution in some species. Our findings have implications for understanding complex interactions among sequence types in repeat-rich genomic regions and the fundamental mechanisms of genome divergence and speciation.

[B376] Teaching an old protein new tricks: the evolution of novelty through the lens of a selfish element

Julian Ross[1,2], Polina Tikanova[1,2], Andreas Hagmüller[1], Kolin Echano[1], Keyu Xiao[4], Till Balla[3], Peter Duchek[1], Rob Jelier[4], Danny Nedialkova[3], Gang Dong[5], Dr Alejandro Burga[1]
[1]Institute of Molecular Biotechnology (IMBA), Vienna, Austria, [2]Joint Doctoral School of the University of Vienna and the Medical University of Vienna, , , [3]Max-Planck-Institute for Biochemistry, Martinsried, Germany, [4]KU Leuven, Leuven, Belgium, [5]Max Perutz Labs, Vienna, Austria

Gene duplication is a fundamental evolutionary mechanism that drives expansion and diversification of protein families. However, the extent to which duplicated genes acquire entirely novel molecular functions – beyond subtle modifications of existing ones – remains poorly understood. To address this gap we investigated how KLMT-1, a toxin that is part of a selfish toxin-antidote (TA) element, rapidly evolved from FARS-3, an essential subunit of the phenylalanyl-tRNA synthetase complex, in the nematode C. tropicalis.

Surprisingly, we found that despite originating from a tRNA synthetase, KLMT-1 does not kill embryos by disrupting tRNA abundance, charging, or modification. Instead, we discovered that KLMT-1, unlike its ancestor FARS-3, localizes to the centrosome of developing embryos. Remarkably, this localization, as evidenced using centrosomal markers, is not constitutive but specifically triggered during early embryogenesis. We experimentally validated key amino acid differences between KLMT-1 and FARS-3 that are required for both centrosomal localization and toxicity. In-silico screening of centrosomal proteins revealed an unexpected and novel interaction of KLMT-1 with the major mitotic regulator AIR-1 (Aurora kinase A), which we showed is essential for the localization and function of the toxin. In line with these findings, our lineage tracing results suggest that KLMT-1 expression induces cell division abnormalities in developing worm embryos, consistent with a role as a disruptor of centrosome function.

Collectively, our results illustrate how radically new molecular functions can arise through gene duplication and highlight how selfish TAs can serve as powerful platforms for organisms to explore otherwise inaccessible sequence space of essential genes.

[B377] Conjugation promotes the segregation of novel plasmid alleles under non-selective conditions

Lisa Marie Hartmann[1], Dr. Mario Santer[1], Dr. Nils F. Hülter[1], Pr. Dr. Tal Dagan[1]
[1]Institute of General Microbiology, Kiel, Germany

Plasmids are extrachromosomal elements in prokaryotes that are able to transfer between cells. Mobile plasmids are present in their host cell with multiple copies; consequently, they have a higher mutational supply compared with monoploid replicons, thus enabling the rise of intracellular diversity (termed heteroplasmy). At the same time, the establishment of novel plasmid alleles is constrained by segregational drift, where even beneficial alleles can be rapidly lost. Horizontal plasmid transfer has the potential to alter allele composition in the population, however, the effect of plasmid transfer on plasmid allele dynamics remains poorly understood. Here, we show that plasmid transfer via conjugation accelerates the emergence of homoplasmic hosts. Establishing an experimental system for following conjugative plasmid allele dynamics, we quantified the dynamics of a novel resistance allele in Acinetobacter baylyi under non-selective conditions. A novel resistance allele was introduced into the ancestral donor population, generating heteroplasmic hosts that segregated over time. Daily mating of donors with recipients revealed that the recipient population comprised mostly homoplasmic hosts. The recipient plasmid allele composition was similar to the novel allele frequency in the donor population. Rare heteroplasmic recipients likely emerged following the transfer of two plasmid variants to one recipient prior to plasmid establishment. A mathematical model calibrated with the experimental results predicts that plasmid transfer has a small effect on the ratio of homoplasmic novel to ancestral allele hosts over time. Our study demonstrates conjugation as an alternative route for the segregation of novel plasmid alleles.

[B378] Recent expansion of LTR Retrotransposons in the genome of the Swedish Sand Lizard, Lacerta agilis

Malavi Sengupta[1], Joshua Hufton[1], Dr. Mats Olsson[2], Dr. Mette Lillie[1]
[1]Department Of Ecology And Genetics, Animal Ecology, Uppsala University, Uppsala, Sweden, [2]Department of Biological & Environmental Sciences, Gothenburg University, Gothenburg, Sweden

Transposable Elements (TEs) are DNA sequences that can move independently inside eukaryotic genomes and can cause large-scale changes to genomic architecture and gene regulation. Characterisation of the mobilome (TE contect of a genome) can provide valuable insights into genome evolution and historical TE activity, as well as identify recent or ongoing transposition. Here, we characterized the mobilome of the sand lizard, Lacerta agilis. We compare this to the mobilomes of 13 other high-quality squamate references genomes, and the Tuatara, Sphenodon punctatus, which was used as an outgroup. Contrary to earlier studies in reptiles, we observe that genome size is correlated with TE content (Spearman's ρ = 0.836), indicating that TE expansions and contractions have played a role in genome evolution. Furthermore, we observe signatures of recent activity of two LTR retrotransposons families, Gypsy and Copia, which appear to have expanded in the sand lizard genome. Insights into the evolutionary history of these expansions is inferred from lineage-specific phylogenies and LTR-divergence, providing estimates into the timing and origins of these elements. The recent accumulation of these LTR retrotransposons throughout the genome of a Swedish sand lizard may impose a substantial cost through increased mutation load and genomic instability. This pattern could therefore potentially represent a genetic threat to a species already of conservation concern.

[B379] Transposable Elements Facilitate the Origin and Evolution of a Novel Antifreeze Protein Gene in Snailfishes

Nathan Rives[1], Prabodh Bajpai, Xuan Zhuang
[1]University Of Arkansas, Fayetteville, United States

Transposable elements (TEs) are increasingly recognized as raw material for new gene evolution, but we still lack mechanistically detailed examples of how TE-derived fragments are assembled into a novel functional gene in vertebrates. Type I antifreeze proteins (AFPI) in polar and subpolar fishes are ideal for studying gene birth under strong selection, and our recent work (published on MBE) shows that three AFPI-bearing lineages evolved this nearly identical, alanine-rich protein independently from distinct precursors. In contrast, the genomic origin of AFPI in snailfishes has remained insufficiently resolved, despite its clear ecological importance for survival in freezing habitats. Here, we generated a new chromosome-scale long-read genome assembly for the Atlantic snailfish Liparis atlanticus, combined with Iso-Seq, RNA-seq, and comparative genomics across AFPI-positive and AFPI-negative snailfishes and teleost outgroups, to reconstruct the origin of snailfish AFPI and quantify the contribution of TEs. We show that snailfish AFPI arose as a mosaic gene assembled from multiple genomic sources, including TE-derived sequences, previously noncoding DNA, and untranslated fragments of a housekeeping gene. TE insertions flank and interdigitate with AFPI across the locus, with specific TE families contributing candidate promoter and UTR-associated sequence as well as providing substrates for duplication and translocation, leading to lineage-specific expansion and divergence of AFPI copy number. Together, these results reveal a TE-facilitated, mosaic origin of a novel antifreeze protein gene in snailfishes and illustrate how TEs, fragmented gene precursors, and noncoding regions can be stitched together to generate a new, strongly selected gene in a natural vertebrate system.

[B380] Co-evolutionary dynamics of piRNA and KZFP defense systems against transposable elements

Yusuke Nabeka[1], Hideki Innan[1]
[1]The Graduate University for Advanced Studies, SOKENDAI, Hayama, Japan

Transposable elements (TEs) pose a significant threat to genomic stability. To counteract this, host organisms have evolved sophisticated defense mechanisms, including the PIWI-interacting RNA (piRNA) pathways and the more recently evolved Krüppel-associated box zinc finger proteins (KZFPs). While both systems utilize sequence-specific recognition and share common epigenetic machinery to silence TEs, it remains unclear why a seemingly redundant KZFP system evolved and persists alongside the already effective piRNA pathway.

We address this by modeling the co-evolutionary dynamics of TEs and these two defense systems. Our model focuses on a key mechanistic asymmetry: piRNA silencing is "reactive," triggered by transposition into piRNA clusters, whereas KZFPs offer "constitutive" suppression, using their DNA-binding domain. Using a deterministic population genetic model, we derived the conditions required for a KZFP allele to invade a population already at equilibrium with piRNA regulation. We found that KZFPs can invade and stably coexist with piRNAs, specifically when KZFP efficacy is intermediate and fitness costs are low. These analytical results were tested using individual-based simulations incorporating linkage disequilibrium and genetic drift. Our findings suggest that KZFPs and piRNAs do not simply coexist but interact in non-additive ways through their shared TE targets. By defining the conditions for their coexistence, we provide a theoretical basis for understanding why complex, layered defense systems evolve and persist.