Talks

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The Baldwin effect reloaded (BE 2.0)

Since the late 1890s and until today, the history of the Baldwin effect (thereafter BE) is mainly a history of controversies about its conceptual definition and evolutionary scope. Proponents of the BE supported the view that, in altered environmental conditions, phenotypic plasticity is the key factor in allowing a population to avoid extinction. Opponents of the BE pointed out that phenotypic plasticity, by masking genetic variation, slows gene-level evolution. To settle this debate, we combine extinction, selection, and plasticity within the same quantitative framework. We develop a stochastic population model featuring the minimal number of ingredients to account for genetic drift, variable population size, variable environment, competition, mutation, and plasticity. We study evolutionary rescue of the population (arrival and invasion of an adaptive genetic mutant) in the altered environment for different values of phenotypic plasticity, here quantified as the probability that the maladapted genotype develops into the adapted phenotype. Our claim is that the BE is a genuine evolutionary mechanism whereby phenotypic plasticity promotes evolutionary rescue by delaying extinction, but that BE is limited to intermediate values of plasticity. More specifically, we show that the speed of adaptation actually peaks at intermediate levels of phenotypic plasticity: at low levels of plasticity, the population size gets depleted too rapidly, whereas at high levels of plasticity, the selective advantage of adaptive variants is too small.

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Relating the evolutionary fitness costs of loss-of-function mutations to their pathogenic consequences in humans

A number of recent disease studies have reported an enrichment of causal variants in “mutation intolerant genes”. Here, we consider this relationship explicitly, by relating pathogenic consequences at present-day to inferred fitness effects of loss of function mutations. To this end, we first infer posterior distributions for the evolutionary fitness costs of loss-of-function (LOF) mutations for 17,744 autosomal and 679 X-linked genes, given the observed frequencies of LOF mutations in 56,855 individuals. Estimated fitness costs are on the order of 1% on average; they are typically largest for X-linked genes, whether or not they have a Y homolog, followed by autosomal genes, and smallest for genes in the pseudoautosomal region. We then compare the distribution of fitness effects (DFE) for all possible de novo LOF mutations to the distribution for mutations identified in pedigree studies of individuals diagnosed with one of six severe complex diseases. All six cohorts are enriched for mutations with estimated fitness effects in excess of 10%, indicating that causal mutations tend to be highly deleterious, and are likely to be quite recent in origin. We see an effect of disease severity, with a larger enrichment of such highly deleterious variants for early onset developmental disorders than Schizophrenia or Tourette syndrome. We also detect influences of the pedigree study design on the types of mutations detected for a given diagnosis: thus, we find that in autism studies, mutations identified in probands of simplex family studies are more deleterious on average than those in multiplex family studies, and mutations found in female probands are more deleterious on average than those in males. More generally, this approach allows us to characterize the types of mutations that contribute to complex disease risk and how they are transmitted in populations.

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Scarlet tide - the first report of sex chromosomes in red algae

Red algae (Rhodophyta) belong to the oldest evolutionary lineages of photosynthetic eukaryotes and comprise one of the largest phyla of algae on the planet. Most of the red algal species are multicellular and macroscopic, live in marine environment and have a complex life history with alternation of three, rather than two, generations. Particularly interesting are the aspects of the evolution of sex determination during the haploid life stage (UV sex chromosomes), however, the sex chromosomes in Rhodophyta have never been described to date. In this study, we used high-quality genomic, transcriptomic and genetic marker data to provide the first report of the red algal UV sex chromosomes using three species from the order Gracilariales. We report the genomic architecture and gene content of the male (V) sex chromosomes and their nonrecombining regions. The UV system in Gracilariales shows distinct evolutionary history not only from the well-studied XY and ZW systems but also from the other algal UV systems described so far. Nevertheless, some striking similarities exist, indicating remarkable universality of the underlying processes shaping sex chromosome evolution across major eukaryotic supergroups.

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Using site frequency spectrum to detect purifying selection on endemic island ungulates

Population genetic theory states that small populations are prone to losing their genome-wide variation and their ability to adapt to future environmental change. Quantifying deleterious mutations and assessing how it affects populations' survival has hence been a routine of population genomic analysis of small populations. However, many methods disregard the heterozygous sites to avoid having to assume dominance effect in genotypes containing deleterious alleles thus missing information from heterozygous sites. We used site frequency spectrum to enable the inclusion of derived alleles from all genotypes and used a composite likelihood approach to detect shifts in frequency that might indicate purifying selection. We test this method on a meta-population of endemic ungulates, anoa (*Bubalus* spp., 'dwarf buffalo'), and babirusa (*Babyrousa* spp., 'pig-deer') who are endemic to the Wallacea, an archipelago in Indonesia, Southeast Asia, inhabiting islands of different sizes across the region, i.e. the large island of Sulawesi and the smaller islands of Togian and Buton, and reported different extent of spectra shift as across the different subpopulations as a measure of purifying selection.

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Assessing inbreeding in the genus *Antirrhinum* using ROH analysis

Repeated mating between close relatives leads to increased regions of homozygosity in the genomes of offspring, termed runs of homozygosity (ROH). Using genome-wide sequencing data it is possible to assess the history of inbreeding within a population based on the length and number of ROH across individuals. Using a RADseq database containing 24 species from the self-incompatible plant genus *Antirrhinum*, the patterns of inbreeding across the whole genus was assessed, with a clear differentiation in historical inbreeding identified between the two major subsections of the genus. Additionally, ROH was assessed using haplotag short read sequencing from ~1000 individuals from a wild population of the species *Antirrhinum majus*, enabling the characterization of the distribution of inbreeding within this obligately outcrossing plant species population. Furthermore, by comparing inbreeding coefficients calculated from ROH against measured fitness-proxy trait data for the 1000 A. majus individuals, the level of inbreeding depression in the population was also calculated.

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Sweepstakes reproductive success via pervasive and recurrent selective sweeps

The concept of sweepstakes reproductive success, posits enormous variance and skew in individual reproductive output of high fecundity organisms. However, it is unknown whether highly fecund organisms reproduce by sweepstakes and if they do, the relative. roles of neutral and selective sweepstakes. Here we use coalescent-based statistical analysis of genomic population data and show that selective sweepstakes are a strong candidate for explaining recruitment dynamics in the highly fecund Atlantic cod. The sweepstakes result from recurrent and pervasive selective sweeps of new variation generated by mutation. We show that the Kingman coalescent and the Xi-Beta coalescent (modelling random sweepstakes), including complex demography and background selection, are inadequate explanations. Our results show that sweepstakes reproduction processes and multiple-merger coalescent models are relevant and necessary for understanding genetic. diversity in highly fecund natural populations.

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Evolution of a sex-linked supergene determines unusual reproductive strategies in fungus gnats

Fungus gnats (Diptera: Sciaridae) exhibit peculiar reproductive strategies: some species are digenic (producing mixed-sex broods) while others are monogenic (producing single-sex broods), and sex is determined maternally through elimination of paternal chromosomes in the developing zygote. Previous cytological observations revealed that monogenic reproduction in *Bradysia coprophila* is controlled by a large X-linked inversion. By combining multiple sequence data types for variant calling and using unconventional genome assembly techniques to assemble highly homomorphic chromosomes, we assembled and characterised the inversion. We have shown that it is likely a 'supergene' composed of multiple distinct young strata that accumulated recently (<0.4mya) in a stepwise process. We identified the breakpoints of the supergene and revealed the extent of its functional degradation and repetitive landscape. Our findings demonstrate how genomic rearrangements can drive turnover of complex phenotypes such as fundamental changes in reproductive strategy.

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Genome polarisation for detecting barriers to geneflow

Semi-permeable barriers to geneflow in principle allow distantly related organisms to capture and exchange pre-adapted genes potentially speeding adaptation. However, describing barriers to geneflow on a genomic scale is non-trivial. We extend classic diagnostic allele counting measures of geneflow across a barrier to the case of genome-scale data. Diagnostic Index Expectation maximisation (*diem*) polarises the labelling of bistate markers with respect to the sides of a barrier. An initial state of ignorance is enforced by starting with randomly generated marker polarisations. This means there is no prior on population or taxon membership of the genomes concerned. Using a deterministic data labelling, small numbers of classic diagnostic markers can be replaced by large numbers of markers, each with a diagnostic index. Individuals' hybrid indices (genome admixture proportions) are then calculated genome wide conditioned on marker diagnosticity; within diploid, haplodiploid and/or haploid genome compartments; or indeed over any subset of markers, allowing classical cline width/barrier strength comparisons along genomes. Along-genome barrier strength heterogeneity allows for barrier regions to be identified. Further, blocks of genetic material that have introgressed across a barrier are easily identified with high power. diem indicates panmixis among *Myotis myotis* bat genomes, with a barrier separating low data quality outliers. In a *Mus musculus domesticus*, *Mus spretus* analysis diem adds multiple introgressions of olfactory (and vomeronasal) gene clusters in one direction to previous demonstrations of a pesticide resistance gene introgressing in the opposite direction across a strong species barrier. diem is a genomes analysis solution which scales over reduced representation genomics of thousands of markers to treatment of all variant sites in large genomes. While the method lends itself to visualisation, its output of markers with barrier-informative annotation will fuel research in population genetics, phylogenetics and association studies. diem can equip such downstream applications with millions of informative markers.

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Uncovering gene-family founder events during major evolutionary transitions in animals, plants and fungi using GenEra

Background: The emergence of new genes is an important driver of evolutionary novelty. Yet, we lack a conceptual and computational approach that accurately traces gene-family founder events and effectively associates them with trait innovation and major radiation events.
Results: We present GenEra (https://github.com/josuebarrera/GenEra), a DIAMOND-fuelled gene-family founder inference framework that addresses previously raised limitations and biases of founder gene detection in genomic phylostratigraphy by accounting for homology detection failure (HDF), accelerating gene-family founder computations from several months to a few days for any query genome of interest. We analyzed 30 genomes to explore the emergence of new gene families during the major evolutionary transitions in plants, animals, and fungi. The detection of highly conserved protein domains in these gene families indicate that the neofunctionalization of pre-existing protein domains is a richer source of gene-family founder events compared to de novo gene birth. We report vastly different patterns of gene-family founder events in animals and fungi before and after accounting for HDF, while only plants show a consistent pattern of gene-family emergence after accounting for HDF. We show that the transition to multicellularity in streptophytes, the terrestrialization of land plants and the origin of angiosperms are associated with gene-family founder bursts; as well as the evolution of bilateral symmetry in animals.
Conclusions: Our results indicate that the impact of HDF on the inferred patterns of gene emergence is lineage-dependent, suggesting that plants are more likely to evolve novelty through the emergence of new genes compared to animals and fungi.

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Mito-nuclear discordance explains the ambiguous phylogenetic relationship of the red fox (*Vulpes vulpes*) and Rüppell's fox *(V. rueppellii*)

Interspecific hybridization can lead to introgression, but its genomic impact depends on the interplay of selection, drift and gene-flow. The arid-adapted Rüppell's fox (*Vulpes rueppellii*) has previously been suggested to be the sister species of the red fox (*Vulpes vulpes*), albeit presumably nested within its mtDNA diversity. This paraphyly could indicate recent divergence of *V. rueppellii*, questioning its classification as a distinct species. We here analyse high-resolution mitochondrial and genome-wide ddRAD and whole genome resequencing data from both species, with special focus on sympatric areas (North Africa and the Near East). We identified five mitochondrial clades, confirming with high support the paraphyly of *V. vulpes*: all *V. rueppellii* individuals clustered in a 'Palearctic' clade, intermingled but not shared with *V. vulpes*. In contrast, species trees of autosomal loci showed the two species as overall strongly differentiated sister lineages. The whole genome data showed an ancient signal of gene flow from *V. rueppellii* into *V. vulpes*, while ddRAD data from a larger sample size of individuals revealed a signal of recent hybridization (a recent hybrid between the two species, found in Egypt), along with pronounced gene flow among *V. vulpes* populations. Genetic diversity was higher within *V. vulpes* populations than in those of *V. rueppellii*. Our findings are consistent with the broad habitat flexibility and wide geographic range of V. vulpes, allowing this generalist to cope with environmental and food availability changes. On the contrary, the desert inhabitant *V. rueppellii* appears more vulnerable to habitat and environmental changes, with scarcity of resources promoting population fragmentation and increased inbreeding. Furthermore, the mito-nuclear discordance suggests an early divergence and extended time for adaptation in *V. rueppellii,* followed by introgression - supporting its classification as a distinct species.

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Meiotic drive and sperm competition

The X-linked 'Sex Ratio' (SR) meiotic drive system in *Teleopsis dalmanni* stalk-eyed flies causes degeneration of all non-carrier Y-sperm, leading to the production of female-only offspring broods. We tested whether the killing of sperm reduces drive-male fertility during sperm competition. Previous studies suggest that drive-males sire fewer offspring, either because they produce fewer or lower-quality sperm. However, our recent findings contradict this view, as drive-males transfer the same numbers of viable sperm during mating. We investigate this further by performing reciprocal mating trials to measure the success of drive-male sperm in competition with standard male sperm (one mating each). Although the success of individual males varied greatly, there was no difference in the number of offspring sired by drive and standard males, regardless of their mating position. We performed a further experiment, to test the success of drive-males under higher competition - when a female is multiply mated - and report our preliminary findings. Overall, we find drive-males are not at a disadvantage during sperm competition. This suggests that the evolution of larger testes in drive-males entirely mitigates the costs of sperm loss caused by meiotic drive. This situation is unlike that observed with other Dipteran species, such as Drosophila, and helps explain the high ~20% SR frequency in wild populations of *T. dalmanni*.

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The Darwin Tree of Life project: reference genomes for all of biodiversity

The Darwin Tree of Life project is a collaboration between biodiversity, genomics and bioinformatics teams* across Britain and Ireland that aims to generate reference quality genome sequences for all of the species with which we share these islands. After three years of operation and innovation we have built a fully operational process that takes a specimen from the wild through nucleic acid extraction, sequencing, assembly, annotation and public release. Our data now contributes about 2 in every 5 reference genomes submitted to the INSDC public databases, and all data are fully openly available through project portals (see https://portal.darwintreeoflife.org/). The DToL consortium is poised to transform the landscape of genomics by developing methods that can be applied to the very small (and very large) and to all branches of the tree of eukaryotic life - and to deliver at scale. We are making discoveries about patterns of centromeric and telomeric sequence, the structure and evolution of chromosomes, the divergence between sister species and the phylogenetic structure of the diversity of life.* The DToL consortium includes the Royal Botanic Gardens Kew, the Royal Botanic Gardens Edinburgh, the Natural History Museum London, the Marine Biological Association Plymouth, the Wytham Woods field station of the University of Oxford, the Earlham Institute, the University of Oxford, the University of Edinburgh, the University of Cambridge and the European Bioinformatics institute, with collaborators in University College Dublin, natureScot and CABI among (many) others. DToL is funded by the Wellcome Trust and through in-kind and local funding to collaborators.

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Large polymorphic chromosome inversions in the Lake Malawi cichlid radiation

With more than 800 haplochromine cichlid species, Lake Malawi is home to one of the most specious vertebrate radiations. The disparity between the immense phenotypic variation and the very limited genetic differentiation among species makes the radiation an ideal model system to study the genomic mechanisms underlying speciation. Chromosomal inversions can play crucial roles in adaptation, as they supress recombination between the ancestral and the inverted haplotype and can protect advantageous allele combinations. Inversions could therefore contribute to local adaptation and eventually speciation in a sympatric radiation, where ongoing introgressive gene flow would otherwise antagonize divergence. However, there is no evidence for a role of inversions in the largest vertebrate radiations. Here, we report the identification of five large (~20 Mb) chromosomal inversions, which we genotyped across more than 2,000 Malawi cichlids from over 250 species. These inversions are polymorphic across species boundaries but also within individual populations, suggesting potential roles in adaptation, which is supported by genotype frequencies that deviate from Hardy-Weinberg equilibrium. Some inversions act as active sex determining systems in a subset of species. Our current work explores the majority of cases where the inversions are not sex determining, and we plan to specifically target potential roles in speciation and adaptation.

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Traditional phylogenetic models are insensitive to variations in the effective population size

A substitution represents the emergence and fixation of an allele in a population or species and is the fundamental event from which phylogenetic models of sequence evolution are devised. As a result of the increasing availability of genomic sequences, we can currently leverage interspecific variability while reconstructing the tree of life. Substitutions can thus more realistically be modeled as the product of mutation, selection, and genetic drift. However, it remains unclear whether this additional complexity affects our measures of evolutionary times and rates. This study seeks to answer this question by contrasting the traditional substitution model with a population genetic counterpart by using data from 4385 individuals distributed across 179 populations and representing 17 species of animals, plants, and fungi. We found that when the population genetics dynamic is modeled via the substitution rates, the evolutionary times and rates are well correlated, suggesting that the substitution models are able to capture the time and pace of their population counterpart. However, a closer inspection of this result showed that the traditional models largely ignore the effect of population size, even when explicitly accounted for in the substitution rates. Our results indicate that superimposing population-genetics results on the substitution rates may be efficiently used to study mutation and selection biases, while other data sources (e.g., life history traits or polymorphisms) need to be additionally integrated to infer historical demography. We expect our results to help develop mutation-selection phylogenetic models and inspire unified frameworks between large and short evolutionary time scales.

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Meiotic drive adaptive testes enlargement during early development in the stalk-eyed fly

The sex-ratio (SR) X-linked meiotic drive system in stalk-eyed flies destroys all Y-bearing sperm. Unlike other SR systems, drive males do not suffer fertility loss. They have greatly enlarged testes, which compensate for gamete killing. We predicted that enlarged testes arise from extended development with resources re-allocated from the accessory glands, as these tend to be smaller in drive males. To test this, we tracked the growth of the testes and accessory glands of wildtype and drive males over 5-6 weeks post-eclosion before males attained sexual maturity. Neither of the original predictions are supported by this data. Instead, we found that the drive-male testes were enlarged at eclosion, reflecting a greater allocation of resources to the testes during pupation. In addition, there was no evidence that the greater allocation of resources to the testes during adult development retarded accessory gland growth. There was evidence of a general trade-off with eyespan, as males with larger relative eyespan had larger accessory glands but smaller testes. These findings support the idea that enlarged testes in drive males arise as an adaptive allocation of resources to traits that enhance male reproductive success.

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Inferring different modes of natural selection using PoMos in RevBayes

The interplay between mutation, genetic drift, directional, and balancing selection in shaping populations' diversity is highly convoluted and hard to disentangle. This requires sophisticated phylogenetic models that have high degrees of flexibility and can handle multi-individual data. For these purposes, our group has developed a polymorphism-aware phylogenetic set of models called PoMos. These models have recently been proven effective in inferring species trees as well as mutational effects, fixation biases and, in some cases, GC-bias rates of great apes and grasshoppers. For ease of use, the models are now implemented in the open-source Bayesian inference framework RevBayes. In this study, we further developed PoMos to study neutral, directional and, for the first time, balancing selection. The main benefit of our models for studying the balancing selection is that PoMos allow for ancestral polymorphisms that can be maintained, and parameters that can measure frequency-dependent selection. We have applied our new approach to a set of simulated data to test the inferential framework. We also investigate real sequences of African human populations to understand the evolutionary history of regions of the genome that are known to be under balancing selection driven by malarial parasites.
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Geographic variation in gene flow from a genetically distinct migratory ecotype drives population genetic structure of coastal Atlantic cod (*Gadus morhua L.)*

Identifying how physical and biotic factors shape genetic connectivity among populations in time and space is essential to our understanding of the evolutionary trajectory as well as the management of marine species. Atlantic cod is a widespread and commercially important marine species displaying several ecotypes with different life history strategies. Using three sets of SNPs: neutral, informative, and genome-inversion linked, we studied population genetic structure of ~2500 coastal Atlantic cod (CC) from 40 locations along Norway's 2500 km coastline, including nine fjords. We observed: (1) a genetic cline, suggesting a mechanism of isolation by distance, characterized by a declining FST between CC and North East Arctic Cod (NEAC - genetically distinct migratory ecotype) with increasing latitude, (2) that in the north, samples of CC from outer-fjord areas were genetically more similar to NEAC than were samples of CC from their corresponding inner-fjord areas, (3) greater population genetic differentiation among CC sampled from outer-fjord areas along the coast, than among CC sampled from their corresponding inner-fjord areas, (4) genetic differentiation among samples of CC from both within and among fjords. Collectively, these results permit us to draw two main conclusions. First, that differences in the relative presence of the genetically highly distinct, migratory ecotype NEAC, declining from north to south and from outer to inner fjord, plays the major role in driving population genetic structure of the Norwegian CC. Second, that there is limited connectivity between CC from different fjords. These results suggest that the current management units implemented for this species in Norway should be divided into smaller entities. Furthermore, the situation where introgression from one ecotype drives population genetic structure of another, as is the case here, may exist in other species and geographical regions, thus creating additional challenges for sustainable fisheries management.

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Asexuality is not faithfully transmitted by contagion in *Daphnia pulex*

In some taxa, new emergences of asexual lineages are possible through contagious asexuality, where rare males from obligate asexual lineages can transmit asexuality to new lineages by cross-mating with sexual females. With such 'contagious asexuality' scenario, it is often assumed that asexuality can be immediately transmitted intact from the asexual to the new hybrid lineages. In this paper, we investigate in detail whether asexuality is faithfully transmitted in such crosses. We studied the reproductive modes of F1s produced by crossing sexual females to males from an obligate parthenogen lineage in *Daphnia pulex*. While the parental asexual lineage is an obligate parthenogen reproducing clonally, we find that the F1s show a wide diversity of reproductive modes. We do not find discrete classes of sexual vs. asexual F1s. Rather, some F1s appear to be able to reproduce both sexually and asexually. Moreover, when they are able to reproduce asexually (about 20 % of F1s), they do not reproduce clonally, as shown by frequent loss of heterozygosity (LOH) among their parthenogenetic offspring. Such LOH can lead to large fitness reduction by revealing recessive deleterious mutations, which may therefore largely impact the chance of establishment of contagiously-produced asexual lineages. We also found that these F1s are difficult to produce and have strongly reduced fertility rates, particularly for asexual F1s compared to natural ones, indicating that the initial fitness of these contagiously-produced asexual lineages is also often low. Together, our results indicate that asexuality is not transmitted intact with 'contagious' crosses. Such crosses rather result in diverse, non-binary, and non-clonal offspring, on which subsequent selection may act.

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Speciation dynamics in plants versus animals

Speciation describes the process of progressive accumulation in genomes of one type of mutations: species barriers. These barriers reduce the fitness of hybrid individuals relative to parental fitness. If this process is described to be continuous in time, it ultimately produces two discrete entities called species. Despite the continuous nature of speciation, it can be subdivided into three stages according to the degree of permeability to gene flow: 100% permeable (populations), semi-permeable (semi-isolated species) and 100% impermeable (species).In a previous study, using demographic inference in 61 pairs of animals with different values of molecular divergence, we showed that semi-permeable species were common in animals up to levels of net divergence of ~2%.In this study, we perform the same demographic inferences to test gene flow in ~400 pairs of plants. This analysis reveals large differences in speciation dynamics between plants and animals, including the importance of gene flow in natural populations.

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Contribution of mitonuclear genomes to local adaptation

Mitochondrial metabolism is regulated by a series of enzyme complexes within the mitochondrion, with their functioning being highly sensitive to the environment. Indeed, many studies have found that mitochondrial haplotype frequencies tend to associate with latitude and altitude, which has led to the hypothesis that mitochondrial genomes could contribute to local adaptation. Here we investigate this hypothesis by using the model system Drosophila melanogaster from the Australian eastern cline. Previous work has described two major mitochondrial haplotypes, whose frequencies exhibit an opposing pattern of clinal variation - one predominating in the north (tropical), whereas the other more common in the south (temperate). Our work builds on this model, and we have created mitonuclear cybrid populations to test the contribution of both genomes to life-history traits and mitochondrial bioenergetics. I'll present evidence for these inter-genomic interactions being quite important contributors to local adaptation and species persistence.

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Re-evaluating Loricate Choanoflagellate Phylogenetics

Choanoflagellates of the order Acanthoecida possess an extracellular coat in the form of basket constructed from silica strips called the lorica. The taxon has been consistently recovered as monophyletic in molecular phylogenies. Based upon differences in lorica development and morphology, as well as the presence or absence of a motile dispersal stage, species are labelled as either nudiform or tectiform. Whilst Acanthoecida is robustly resolved in molecular phylogenies, the placement of the root of the clade is less certain with two different positions identified in past studies. One recovered root has been placed between the nudiform family Acanthoecidae and the tectiform family Stephanoecidae. An alternative root placement falls within the tectiform species, recovering the monophyletic Acanthoecidae nested within a paraphyletic Stephanoecidae. Presented here is a 14-gene phylogeny, based upon nucleotide and amino acid sequences, which strongly supports tectiform paraphyly. The horizontal transfer of a ribosomal protein gene, from a possible SAR donor, into a subset of acanthoecid species provides further, independent, support for this root placement. Differing patterns of codon usage bias across the choanoflagellates are proposed as the cause of artefactual phylogenetic signals that lead to the recovery of tectiform monophyly.

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Modern human expansion from (southern?) Africa: Origin, non-African ancestry, and linearity

Modern humans are understood to have expanded across the continents from Africa. It has been reasoned that this expansion involved bottlenecks; in previous studies, diversity in certain genetic and skeletal qualities fell whilst geographical distance from Africa progressed. Whilst some diversities have been utilised for estimating where the expansion launched from, it is unsettled which part of Africa witnessed the origin. To try pinpointing this origin, the present research used genetic diversities and cranial attributes (e.g., Y-chromosomal diversity, mitochondrial diversity, cranial shape diversity, cranial size dimorphism). From the present and previous research, pooled together were estimates of the origin. Variables used in this pooling each gave an area of origin specifically within Africa whether in the present research or previously. In some analyses with sub-Saharan Africans, autosomal microsatellite heterozygosity was adjusted for non-African ancestry to counter a possible effect of non-African ancestry on estimating the origin. Africa was not within the area of origin regarding Y-chromosomal diversity; Asia had part, or all, of the area. For other variables, the area of origin was unique to Africa. Southern Africa was the pooled, estimated origin. The autosomal microsatellite heterozygosity of sub-Saharan Africans also favoured southern Africa being the origin (regardless of whether non-African ancestry was accounted for). Yet, this heterozygosity did not seem to decrease prior to the expansion getting to circa. 2,000-3,000 km. Reasons for this non-linear trend are considered. All in all, this research suggests that the south is the region of Africa where the expansion most likely started (this abstract is based on a preprint and research cited in there, and the title regarding this abstract is from that preprint which is: Cenac, Z. (2022). Modern human expansion from (southern?) Africa: Origin, non-African ancestry, and linearity. bioRxiv. https://doi.org/10.1101/2022.07.31.500977).

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Re-re-re-re-re-re-re-re-re defining parallel and convergent evolution

The repeated evolution of phenotypes as a response to similar selective pressures has long fascinated biologists, as it offered important evidence for the role of natural selection in evolution. This discussion focused on the concepts of 'parallel' and 'convergent' evolution, however as the field of evolutionary biology expanded to encompass different disciplines (development, genetics, natural history), the two terms became more nuanced, were used interchangeably and even with conflicting meanings, resulting in confusion. Unintentionally, there has been a loss of the focus on the evolution of sameness, here defined as a similar solution to ecological challenges. In this paper, I highlight the central role of the evolution of sameness in our understanding of biology and propose the 'Sameness conceptual framework' and a set of definitions for parallel, convergent and repeated evolution. This framework separates observed phenotypes from vectorial phenotypic evolution, and integrates genomic sources of evolution. Importantly, the framework brings restates the focus of sameness in the study of parallel and convergent evolution.

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Inferring the distributions of fitness effects and proportions of strongly deleterious mutations

The distribution of fitness effects (DFE) among mutations is central in evolutionary genetics as the shape of this distributions has implications for several evolutionary phenomena such as mating system evolution, the rate of adaptive evolution, and the structure of the genetic load. Despite the DFE being extensively studied, the effects of strongly deleterious mutations are difficult to infer since such mutations are unlikely to be present in a sample of haplotypes, so dataset on which inference is based may contain very little information about strongly deleterious mutations. Recent work has attempted to correct for this by expanding the classical gamma-distributed DFE model to include extra parameters, explicitly accounting for strongly deleterious mutations. Here, we use simulations to investigate one such method, adding a parameter (p_{lth}) to capture the proportion of strongly deleterious mutations. We show that p_{lth} can improve the model fit when applied to individual species, but usually underestimates the true proportion of strongly deleterious mutations. Its inclusion also artificially maximizes the likelihood when used to jointly infer a DFE from multiple species. Given the use of p_{lth} and related methods, our results are highly relevant with respect to avoiding model artifacts and improving future tools for DFE inference.

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Mutation rate estimates in mice using mutation accumulation experiments: the first of its kind

Mutations are the source of genetic variation that drive evolutionary change but determining the rate of new mutations is difficult because new mutations are rare and often deleterious, leading to removal by selection when they arise. One method to overcome this difficulty is by using mutation accumulation (MA) experiments, where multiple inbred or clonal lines are maintained for generations from a single ancestral source followed by observation of the number of genetic differences between the descendants and the ancestors. MA experiments provide estimates of mutation in the near absence of selection by keeping population sizes small, and they have been used to estimate mutation rates in several organisms with short generation times, but never in mammals. We performed Illumina whole-genome sequencing (WGS), at an average depth of ~30X, on the founders and descendants from several MA experiments, which included four inbred strains of mice: C57BL/6JR, BALB/cAnNR, FVB/NR, and C3H/HeN. The resulting whole-genome sequences of 68 descendants, each from its own full-sib inbred line, as well as the 10 ancestral founders of those lines, provided the first estimates of the mutation rate in mammals from an MA experiment. The mutation rates estimates in the four strains ranged from 5.59 x10^-9 mutations per nucleotide per generation in BALB/cAnNR to 6.45 x10^-9 in C3H/HeN. These mutation rate estimates are remarkably similar among strains and similar to previous estimates using other methods. Altogether, these results provide important evidence on the precision of mutation rate estimation using MA experiments in mammals, which was previously unknown.

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Genes controlling mimicry in two morphs of a South American moth (*Chetone histrio*)

Mullerian mimicry is a striking example of convergent evolution by natural selection. Among Lepidoptera, mimicry and defensive coloration are particularly widespread causing convergence in wing patterns between phylogenetically distant species. For example, the south American moth *Chetone histrio* displays wing color patterns shared with different butterfly species (Heliconiini/Ithomiini) from which they diverged over 100 Mya. In this study, we compared two morphs of the *Chetone histrio* moths found in sympatry and dissected the genomics regions responsible for the difference in color patterns. Association mapping and other population genomics approaches identified a large inversion (900kb) associated with the variations in wing color patterns. This inversion contains a gene, cortex, involved in adaptive differences in wing colour patterns among multiple Lepidoptera. Furthermore, this inversion has locked together additional genes, also reported in many co-mimetic species, and thus might form a supergene. Altogether these results suggest that the genetic mechanisms controlling defense coloration are highly conserved among Lepidoptera.

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Genetic basis of mimicry in the Moth *Chetone histrio*

Mullerian mimicry is a striking example of convergent evolution by natural selection. Among Lepidoptera, mimicry and defensive coloration are particularly widespread causing convergence in wing patterns between phylogenetically distant species. For example, the south American moth *Chetone* displays wing color patterns shared with different butterfly species (Heliconis/Ithiominii) from which they diverged over 100 Mya. In this study, we compared two morphs of the *Chetone* moths found in sympatry and dissected the genomics regions responsible for the difference in wing patterns. Association mapping and other population genomics approaches identified a large inversion (900kb) associated with the variations in wing color pattern. This inversion contains a gene, cortex, involved in adaptive differences in wing coloration among multiple Lepidoptera. Furthermore, this inversion has locked together additional genes, also reported in many co-mimetic species, and thus might form a supergene. Altogether these results suggest that the genetic mechanisms controlling defence coloration are highly conserved among Lepidoptera.

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Phylonumtomics uncovers ancient mitogenes in vertebrate genomes

Sporadically genetic material that originates from an organelle genome integrates into the nuclear genome. However, it is unclear what processes facilitate and maintain such an integration of organelle DNA into the nuclear genome over longer evolutionary time. Recently two major hypotheses have put forward to explain the existence of mitochondrial genes in the nuclear genome (NUMTs) that are deeply divergent to the host's mitochondrial genome: recent introgression from another species or adaptive evolution. To address whether these intriguing possibilities do indeed play a role in the evolutionary process we scanned the genomes of more than 1,000 avian and mammalian species for NUMTs. Surprisingly we identified a widespread subclass of NUMTs that showed substantial divergence to the host species' mitochondrial genome. We can show that for these NUMTs signatures of introgression are widespread across mammals but that there is very limited evidence for introgression in birds. We can also show that a substantial fraction of deeply divergent NUMTs are maintained by selection, which raises the question whether they might be functional.

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Wolbachia reduces virus infection in a natural population of *Drosophila*

Wolbachia is a maternally transmitted bacterial symbiont that is estimated to infect approximately half of arthropod species. In the laboratory it can increase the resistance of insects to viral infection, but its effect on viruses in nature is unknown. Here we report that in a natural population of *Drosophila melanogaster*, individuals that are infected with Wolbachia are less likely to be infected by viruses. By characterising the virome by metagenomic sequencing and then testing individual flies for infection, we found the protective effect of Wolbachia was virus-specific, with the prevalence of infection being up to 15% greater in Wolbachia-free flies. The antiviral effects of Wolbachia may contribute to its extraordinary ecological success, and in nature the symbiont may be an important component of the antiviral defences of insects.

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Genomic basis of insularity and ecological divergence in barn owls (*Tyto alba*) of the Canary Islands

Islands, and the particular organisms that populate them, have long fascinated biologists. Due to their isolation, islands offer unique opportunities to study the effect of neutral and adaptive mechanisms in determining genomic and phenotypical divergence. In the Canary Islands, an archipelago rich in endemics, the barn owl (*Tyto alba*), present in all the islands, is thought to have diverged into a subspecies (*T. a. gracilirostris*) on the eastern ones, Fuerteventura and Lanzarote. Taking advantage of 40 whole-genomes and modern population genomics tools, we provide the first look at the origin and genetic makeup of barn owls of this archipelago. We show that the Canaries hold diverse, long-standing and monophyletic populations with a neat distinction of gene pools from the different islands. Using new method, less sensitive to structure than classical FST, to detect regions involved in local adaptation to insular environments, we identified a haplotype-like region likely under selection in all Canaries individuals and genes in this region suggest morphological adaptations to insularity. In the eastern islands, where the subspecies is present, genomic traces of selection pinpoint signs of adapted body proportions and blood pressure, consistent with the smaller size of this population living in a hot arid climate. In turn, genomic regions under selection in the western barn owls from Tenerife showed an enrichment in genes linked to hypoxia, a potential response to inhabiting a small island with a marked altitudinal gradient. Our results illustrate the interplay of neutral and adaptive forces in shaping divergence and early onset speciation.

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The role of microbial symbioses in the evolution of specialised lifestyles in tropical ants

Ants represent one of the most ecologically dominant and diverse groups of organisms on the planet. Contributing to their ecological success has been their ability to inhabit unusual and often extreme environments. It is thought that their capacity to form enduring symbiotic relationships with microbes may have played a role in enabling ants to colonise these niche habitats. However, few ant lineages have been studied for endosymbioses so it is unclear how often these relationships have evolved, and whether they are implicated in the colonisation of specialised niches. Here I test this hypothesis using a wide range of tropical ant species from different genera and subfamilies, as well as vastly different lifestyles and dietary niches. Using deep 16s rDNA sequencing I analyse the microbial community present in >300 ants, to identify key drivers of microbial composition, such as diet, habitat, or taxonomy. Preliminary analyses suggest ants that occupy highly specialized lifestyles, such as mutualistic ant plant association, may have formed strong symbioses with a single dominant microbe. This analysis contributes to the wider understanding of how the major evolutionary transition into symbiotic relationships arose in insects, as well as more specifically how they may have contributed to ants' ecological success.

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Exploring the effects of whole genome duplication on structural variation using pangenomics

Whole genome duplication (WGD) is the most dramatic mutation a genome can undergo, causing enormous disruption to cellular processes. Paradoxically, many polyploids are successful, frequently invading new niches. In order to survive and thrive, young autopolyploids have undergone rapid evolution of DNA management genes, involved in meiosis and mitosis. Instability in these processes could accelerate generation of structural variants (SVs), increasing the pool of large-scale mutations upon which selection acts. Further, theory predicts increased genetic load at higher ploidies, which may manifest as large effect SV. We investigate this in natural populations of the outcrosser *Arabidopsis arenosa*, which has undergone WGD 20-31k generations ago. Here, we use PacBio and Oxford Nanopore long-read sequencing of five diploid and five autotetraploid *A. arenosa* individuals representing all major lineages across the species range to examine how WGD impacts the landscape of SVs within a species. We construct de novo assemblies of each individual and take a pangenomic approach to detect SVs. We can use this robust catalogue to genotype SVs in an extensively sampled dataset of 660 short read sequenced *A. arenosa* individuals, enabling us to explore the impact of SVs in ecotypic differentiation across a large geographic range in a successful autopolyploid. Combining new approaches with traditional population genomics will allow us to consider both the impact of whole genome duplication on the genomic landscape of SVs, and in turn how they might contribute to the occasional spectacular adaptability of polyploid species.

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Genetic structure, host specificity and speciation of small ermine moths (*Yponomeuta*)

Small ermine moths from the genus *Yponomeuta* are host-specific lepidopterans, with 9 species found in Britain, each associated with a single (or small group of) host plant species. For example, a spindle ermine will eat and lay eggs only on spindle plants, but an apple ermine will eat and lay eggs only on apple trees. Previous crossing experiments show that, in general, first generation hybrids between two *Yponomeuta* species will eat both species of plants associated with their parents, but further crosses typically do not eat anything and die without themselves breeding, thus creating a mechanism for ongoing speciation and suggesting a heritable and thus genetic basis of host association. A recent karyotyping study found evidence for several large inversions between two species, supporting a genetic cause for host specificity, but whole genome sequences of these species have not yet been studied. A recent mitochondrial DNA study showed a very low level of genetic divergence within a species complex containing five species common in the UK, as does unpublished reduced-representation genomic sequencing data. Here, I will discuss our initial investigation of new reference genomes assembled by the Darwin Tree of Life project, along with re-sequencing datasets for 30-40 individuals from five British species.

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A mutation-selection model of protein evolution under persistent positive selection

In this talk we discuss the use of first-principles of population genetics to construct a mutation-selection model of codon substitution under persistent positive selection (PPS), and study the relationship between site-specific selection coefficients and the non-synonymous to synonymous rate ratio (dN/dS). We show that in many cases persistent positive selection can be operative even if dN/dS < 1. We show that classical codon models are the special case of the mutation-selection model under PPS when the fitness of amino acids are assumed to be equal at sites.

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Unexpected genomic variation in eastern British Eurasian otters (*Lutra lutra*) revealed by whole genome sequencing

Conservation genetic studies of many endangered species have typically been based on genotyping of microsatellite loci and sequencing of short fragments of mtDNA. With more recently available genomic approaches, it is unclear to what extent increased power and resolution might alter insights from previous genetic approaches. For example, previous genetic work on Eurasian otters in the UK otters identified four genetically differentiated ‘stronghold’ populations in the UK, derived from regional sub-populations that survived the population crash in the 1950-80s. Here we provide a matched comparison of whole genome resequencing versus microsatellite genotyping and control region sequencing for 45 samples from all UK stronghold populations. We found that genomic analyses (autosomal and mitogenome data) confirmed some aspects of population structure shown previously, but also revealed significant shortcomings of previously used genetic markers: (i) identification of two strongly divergent mitochondrial lineages not previously identified using control region fragments, and (ii) that otters in the east of England are genetically distinct and highly variable. Both the origin of this unexpected variation, and the impact on otter conservation in the UK, are currently unclear, but may be related to releases of non-native Eurasian otters in England in the late 20th century. Our work highlights that even reasonably well studied species may harbour genetic surprises, if studied using modern high-throughput sequencing methods.

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Unravelling dynamics of long-term polygenic adaptation in *D. simulans*

Most of the traits in nature have a polygenic architecture: many contributing loci of small effect and phenotypic changes that can be achieved by different combinations of alleles, a phenomenon called genetic redundancy. Therefore, the genomic signature of polygenic adaptation is difficult to distinguish from genetic drift. However, experimental evolution with a moderate number of founder chromosomes results in sufficiently strong linkage disequilibrium such that selected haplotype blocks experience substantial allele frequency shifts in a moderate number of generations. Nevertheless, it is not clear if these allele frequency changes reflect the joint effects of multiple loci with aligned effect sizes or a much smaller number of loci with larger effect sizes. Here, we take advantage of 10 *Drosophila simulans* replicate populations which showed a strong selection response for 99 selected haplotype blocks during 60 generations. Maintaining these populations for up to 200 generations in the same environment enables us to distinguish between many weakly selected alleles and few strongly selected alleles by following the trajectories of the haplotype blocks for additional 140 generations. We show that recombination uncouples the allele frequency trajectories of SNPs in the selected haplotype blocks. We provide evidence that these dynamics are more consistent with many selection targets of weak effects than with a small number of strongly selected alleles.

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Evolution, ecology, and genetics of live-bearing and egg-laying reproduction

Live-bearing (viviparity) has evolved from egg-laying (oviparity) many times in vertebrates. The life history consequences of being one or the other parity mode are well recognised but the genetic basis was previously uncharacterised and therefore its evolutionary generality not known. In my talk I will share some recent results on the genetics and phenotypes of the youngest known example of parity mode switch in an amniote – Europe's common lizard. Over several years of field research at a unique hybrid zone, we detailed female phenotypes in high resolution, including reproductive output and eggshell characteristics. Using admixture mapping, selection analysis, and transcriptomics, we pinpointed functional genes of oviparity or viviparity, signals of selection at recent time scales, and identified a range of candidate genes, some of which are known from mammalian pregnancy. In a quantitative analysis of candidate genes across vertebrates at deep evolutionary time scales, we identified significant levels of sharing of viviparity-related genes across very divergent lineages. Our findings suggest that deeply shared genetic toolkits are involved in the evolution of viviparity. Emerging work suggests that genome structure evolves quickly and is associated with alternative reproductive lineages. Further experimental approaches will be key to identifying causative genetic variation.

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The Trojan Rhino √ê subspecies-level admixture leads to increased mutational load with implications for genetic rescue strategies

In endangered species conservation, genetic rescue by translocation is a common strategy to restore eroded genetic diversity. For the most depleted and at risk populations, the only recourse may be mixing subspecies, in spite of deep evolutionary divergence. However, subspecies-level admixture can have unintended consequences which may compromise conservation success. For the Critically Endangered Eastern black rhino, we show that mixing between divergent subspecies causes increased mutational load in the recipient population. This load undergoes purging over time, but in small populations with inbreeding, deleterious mutations could become fixed, resulting in extinction. We use Slim models to test whether deeply divergent admixture decreases time to extinction in small populations, compared to admixture between more moderately diverged groups, in spite of the benefits of increased heterozygosity. We argue that quantifying mutational load before implementing genetic rescue strategies is important to avoid conservation catastrophe.

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Why does genetic diversity vary across the human genome?

Genetic diversity varies across the human genome. It has recently been demonstrated that the background selection likely explains some of this variation, but what about the remainder? In this talk I will consider the role of variation in the rate of mutation, biased gene conversion and the genealogical process. The mutation rate appears to explain a substantial fraction of the variation, particularly at larger scales, with neither of the other two processes having much influence at all. However, there still seems to be much to understand about variation at smaller spatial scales.

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What can intra-molecular variation tell us about patterns of adaptation at the species level?

The frequency and nature of adaptive mutations are widely variable across species. However, what determines such variation is not fully understood. Several studies have reported that the molecular adaptive rate also varies substantially within genomes, providing evidence for the key role of variables such as recombination, mutation rate, and gene function. More recently, we showed that this rate not only varies between genes but also at the intra-genic level, with protein structure acting as a major determinant of adaptive evolution. This effect, however, varied in strength between species, suggesting that the variation within genes may help explain interspecies patterns of adaptation. Here, we address this hypothesis using a comparative population genomics approach across multiple species with distinct life-history traits and sites with different structural properties. We fit models of distributions of fitness effects to estimate the rate of adaptive substitutions at the amino-acid residue level. Our results suggest an interaction between molecular rates of adaptation acting at the residue and at the species level, thus emphasizing the importance of using intramolecular variation to shed light on the molecular basis of adaptation.

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The scope for sexual antagonistic polymorphism in polygenic traits

Males and females often favour different characteristics, leading to genetic conflicts across the sexes. Theory suggests that these conflicts can maintain genetic variation by generating sexually antagonistic balancing selection, but, so far, most of this theory has neglected the genetic complexity of quantitative traits. Here, we use mathematical modelling and computer simulations to investigate the evolution of a polygenic trait that has different phenotypic optima in males and females. We show that the conditions necessary to maintain variation across multiple loci are significantly more restrictive than at a single-locus, with very strong conflict required. In particular, we show that several sex-specific fitness landscapes known to produce balancing selection in single-locus models by generating dominance reversal, in fact drive disruptive selection across loci in a polygenic trait, due to the presence of negative fitness epistasis between alleles. Together our results indicate that sex-specific selection is not a straightforward source of balancing selection in polygenic traits.

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What selective forces drive the coevolution of sickle-associated haplotypes in the malaria parasite *P.falciparum*?

Malaria parasites impart a strong selective force on the human genome, but whether human resistance mechanisms in turn shape parasite variation has historically been less well understood. However, it was recently discovered that protein-altering single nucleotide polymorphisms in three regions of the malaria parasite genome - termed Pfsa1-3 - are strongly associated with the human sickle haemoglobin (HbS) polymorphism which confers significant resistance to malaria infection. The 'Pfsa+' mutations have unusual population-genetic features, including between-locus linkage disequilibrium and allele frequencies that covary with those of HbS. A natural hypothesis is that the Pfsa+ mutations enable parasites to more easily infect, grow, and cause disease in HbS-carrying individuals. At the same time, the mutations appear not to have fixed in any population raising questions as to the evolutionary forces at play. To investigate this, we used whole-genome sequence data from 5380 community-sampled infections from the MalariaGEN Pf6 resource and 518 severe infections from The Gambia and Kenya. First, we analysed haplotype homozygosity but found little evidence of recent positive selection at the three Pfsa loci. However, the haplotypes carrying Pfsa+ mutations carry multiple mutations in high linkage disequilibrium with the core Pfsa+ SNPs, and are clearly shared between African populations, suggesting there may be a single mutational origin at each locus. Further corroborating this, we developed methods to identify and call genome structural variation at the Pfsa3 locus. This uncovered a set of ancestrally related complex structural rearrangements that are closely linked to the Pfsa+ SNPs and are also shared across populations. These results might suggest stable maintenance of the Pfsa polymorphisms over long evolutionary timescales, but the full set of evolutionary forces and the underlying biological mechanisms are yet to be determined and necessitate further investigation.

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The genomic footprint of social stratification in admixing American populations

Cultural and socioeconomic differences stratify human societies and shape their genetic structure beyond the sole effect of geography. Despite mating being limited by the permeability of sociocultural stratification, most demographic models in population genetics often assume random mating. Taking advantage of the correlation between sociocultural stratification and the proportion of genetic ancestry in admixed populations, we sought to infer the former process in the Americas. To this aim, we define a mating model where the individual proportions of the genome inherited from Native American, European and sub-Saharan African ancestral populations constrain the mating probabilities through ancestry-related assortative mating and sex bias parameters. We simulate a wide range of admixture scenarios under this model. Then, we train a deep neural network and retrieve good performance in predicting mating parameters from genomic data. Our results show how population stratification shaped by racial and gender hierarchies have constrained the admixture processes in the Americas since the European colonisation and the subsequent Atlantic slave trade.

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An Infinite Sites Model for Polygenic Adaptation: _Sweeps or Allele-Frequency Shifts?

While classical population genetics focuses on the dynamics of single loci, quantitative genetics has a more trait-centered view. We combine these two approaches to describe adaptation of quantitative traits in panmictic populations of finite size by studying the evolutionary dynamics at individual loci. We use an infinite sites model assuming freely recombining loci (sites) contributing additively to a trait under weak non-epistatic directional selection. Thus, we examined the effects on polygenic adaptation of selection, random genetic drift, mutation rate and mutation effects drawn from a distribution. We present accurate and computationally tractable approximations for the evolution of the mutant-frequency distributions, of the phenotypic mean and variance and of the number of segregating sites. The mathematical model is based on a combination of branching process theory (for the initial stochastic phase) and deterministic theory. Although diffusion theory leads to simple expressions for many quantities, time-dependent results seem to be out of reach. Our approach yields this time dependence and is especially accurate in the initial phase of adaptation. In addition, our model refines classical results for the stationary phase under long-term selection, where the phenotypic variance has stabilized. Analytic approximations are tested by comprehensive simulations based on a Wright-Fisher model. As an application, we explore the pattern of adaptation, i.e., when the response of the trait is mainly caused by selective sweeps at few loci and when it is due to subtle allele-frequency shifts at many loci. We found that the population-wide mutation rate is the most important parameter to define the pattern, whereas selection strength primarily determines the rate of adaptation. The mutation effect distribution has a minor influence on both.

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The rarer-sex effect

The study of sex allocation - that is, the investment of resources into male versus female reproductive effort - yields among the best quantitative evidence for Darwinian adaptation and has long enjoyed a tight and productive interplay of theoretical and empirical research. The fitness consequences of an individual's sex allocation decisions depend crucially upon the sex allocation behaviour of others and, accordingly, sex allocation is readily conceptualised in terms of an evolutionary game. Here, I investigate the historical development of understanding of a fundamental driver of the evolution of sex allocation - the rarer-sex effect - from its inception in the writing of Charles Darwin in 1871 through to its explicit framing in terms of consanguinity and reproductive value by William D. Hamilton in 1972. I show that step-wise development of theory proceeded through refinements in the conceptualization of the strategy set, the payoff function and the unbeatable strategy.

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Population structure, distribution, and hybridisation of British oak trees

Great Britain has two native oak tree species, *Quercus robur* and *Quercus petraea*. Both are distributed throughout Britain, on the north-western range of both species distribution. It is difficult to distinguish the species morphologically. They are known to hybridise, but hybrids are very difficult to identify from the phenotype. Oaks are wind pollinated, which could lead to little population stratification across large areas, however, it is not known if this is the case in Britain. This study analyses whole genome re-sequenced oaks of both species from across Britain, assesses population structure, species allocation and distribution and the presence and distribution of hybrids.

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Hybridisation in parasites of the *Leishmania Viannia* subgenus

Leishmania parasites are the causative agents of leishmaniases, a group of neglected tropical diseases in poverty stricken areas of the world. The parasites are transmitted by the bites of infected female sandflies, with various animals - typically dogs - acting as reservoirs. Species of the Viannia subgenus cause cutaneous and mucocutaneous leishmaniasis which present as skin lesions and the destruction of mucous membranes, respectively. Treatment for leishmaniases are expensive, toxic and often varying in efficacy. Complicating the treatment options is the tendency for Viannia species to hybridise, the extent to which is currently debated by the Leishmania community. Additionally, this subgenus is a source of taxonomic debate. Here I present a phylogenetic analysis of nine Viannia species, two of which were sequenced de novo here. I provide evidence for species differentiation, solve taxonomic uncertainties and highlight the problems that can only be solved by population-scale genomic sequencing. Based upon the genome of *L. braziliensis*, I show that up to half of the genome displays signatures of recent hybridisation, and quantify the extent to which these species are 'sharing' their genomes. This work will contribute to further genomic analyses of *Leishmania Viannia* species, and aid in future treatment options for the people so badly affected by these parasites.

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Ecologically mediated mutation rate plasticity provides new insights on microbial evolution

Spontaneous point mutations are the ultimate source of genetic variation and, for microbes, can confer resistance to most synthetic antibiotics. Understanding mutation rates is both essential for a fundamental understanding of microbial evolution and suggests sustainable routes to mitigating antimicrobial resistance. Mutation rate is, in part, determined by microbes' ecological environment. Density-associated mutation rate plasticity (DAMP) is a widespread phenomenon resulting in populations at low population density mutate at elevated rates. However, the mechanisms producing DAMP remain elusive. Here we bring together dynamic ODE modelling with experimental measurements of mutation rates and natural mutagens (reactive oxygen species, ROS), to get insights into the DAMP mechanism. ODE modelling predicts that, when individuals' production and/or degradation rates of ROS respond to density, the population shows DAMP. Our experiments in Escherichia coli confirmed that reducing ROS production through anaerobic growth or reducing degradation rates via deletion of katE, katG, ahpC and ahpF (all involved in ROS, specifically hydrogen peroxide, removal) eliminates DAMP. We also explore the ability of ROS degrading cells to modulate the mutation rate of cocultured degradation deficient cells. Further, we show that cells lacking the fur gene have high mutation rates and no DAMP. These cells have an elevated free iron pool, which increases the intracellular concentration of highly mutagenic hydroxyl radicals. Finally, we link DAMP to dynamic fluctuations in the external hydrogen peroxide concentrations generated during the culture cycle. Thus, DAMP mechanisms provide new insights into how ecological factors determine the spontaneous mutation rate and highlight the importance of mutation rate variability in the course of evolution.

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Plastic response in population-specific gene networks to a common stressor: larval crowding in *Drosophila simulans*

Phenotypic plasticity is the phenomenon of genetically identical individuals producing different phenotypes in response to the environment. Previous studies utilize differential responses between evolved and ancestral populations in different environments to understand the evolution of plasticity. Here, we study the plastic response of populations with different evolutionary histories to a common stressor: larval density, which varies with the availability of food and affects many life-history traits, growth rate, body size etc. We used gene expression to study the mechanistic basis of phenotypic plasticity in response to larval crowding in *Drosophila simulans*. As expected, larval density results in highly plastic gene expression patterns in all three populations-more than 1400 genes exhibited significant gene expression differences. Clustering of genes with shared gene expression reaction norms uncovered both conserved gene expression networks that respond similarly in all three populations as well as population-specific networks, with distinct reaction norms among populations. Finally, we also find evidence for population-specific networks, which exhibit very heterogeneous expression changes in response to larval crowding in other populations. We propose that phenotypic plasticity in local populations is the joint effect of conserved gene regulatory networks and population-specific networks. Our results suggest that the comparison of diverged natural populations provides an excellent approach to understand the evolution of regulatory networks of conserved traits in the context of local adaptation.

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Symbiont-driven niche expansions shape the diversification of insects’

For over 300 million years, insects have relied on symbiotic microbes for nutrition and defence. However, it is unclear whether specific ecological conditions have repeatedly favoured the evolution of symbioses, and how this has influenced insect diversification. Using data on 1849 microbe-insect symbioses across 402 insect families, we found that symbionts have allowed insects to specialize on a range of nutrient-imbalanced diets, including phloem, blood and wood. Across diets, the only limiting nutrient consistently associated with the evolution of obligate symbiosis was B vitamins. The shift to new diets, facilitated by symbionts, had mixed consequences for insect diversification. In some cases, such as herbivory, it resulted in spectacular species proliferation. In other niches, such as strict blood feeding, diversification has been severely constrained. Symbioses therefore appear to solve widespread nutrient deficiencies for insects, but the consequences for insect diversification depend on the feeding niche invaded.

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Multiple optima - evidence of balancing selection in the genome of the fire ant

Multiple optima - evidence of balancing selection in the genome of the fire ant. Red fire ants are fascinating superorganisms with diverse phenotypes. Each colony contains individuals from three castes, queens, males, and workers that differ dramatically in morphology, physiology, behaviour, and lifespan. Furthermore, this species has two distinct social forms with single-queen colonies which have higher fitness in new habitats, and multiple-queen colonies that can outcompete single-queen colonies in dense habitats. How can a single genome encode such divergent phenotypes, where the genome seems to be under simultaneous selection for multiple optima? In some cases, the solution lies in phenotype-specific gene expression. However, we also hypothesize that some fire ant genes are under balancing selection, i.e. where selection simultaneously favours multiple divergent alleles. To test whether this type of intragenomic conflict occurs in the ants, we analysed whole genome sequences of 240 haploid male *Solenopsis invicta* fire ants collected from across the native range of this model species. We indeed found dozens of genes with strong signatures of balancing selection. These genes contribute to multiple processes including immunity, neural development, and circulation. Our findings also reveal examples of caste intragenomic conflict that can be resolved. Overall, our results contribute to understanding how different castes, social forms, and immune pressures are encoded in the genome, and the diversity of evolutionary pressures superorganismal evolution must respond to.

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Chromosome-specific inbreeding depression in red deer

Lowered fitness as a result of inbreeding, inbreeding depression, is of significant concern for the conservation of both wild and livestock species. Previous studies have used genome-wide SNP genotypes to gain accurate measures of genomic inbreeding coefficients (Fgrm and FROH) in order to estimate inbreeding depression in a range of traits. Beyond genome-wide effects, it is of interest to determine if particular genomic regions confer excess inbreeding depression, for which a number of different approaches have been used. Here we present a novel approach to estimate chromosome-specific inbreeding depression using >35,000 SNP genotypes in >3,000 individuals in a wild population of red deer. We use multi-membership models to estimate the effect each chromosome's inbreeding coefficient (FROHChr) has on birth weight and survival. We show that using this method is a more conservative measure of chromosome-specific inbreeding depression and that such effects are overestimated using alternative models.

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The human gut microbiome explored at single nucleotide resolution.

The gut microbiome is essential to the wellbeing and health of its human host, yet most studies can only resolve the gut microbial community at genus or species level. Yet we do know that two bacterial strains of the same species can differ by more than half their genome. Furthermore, in clinically relevant microbes, pathogenicity is often a trait encoded at the strain - not species - level. Therefore, my group develops the technologies to track bacterial strain in metagenomic time series, and to investigate evolutionary pressures. I will present some of our studies, where we can show the immense persistence of gut bacteria in healthy human cohorts. A large fraction of microbial species (but not all!) are frequently exchanged among family members, reflected in the selective pressures on their genes. Investigating microbiomes in patients, strain resolved metagenomics becomes even more important, as many difference between healthy and diseased microbiomes can only be detected using strain resolved metagenomics.

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Reproductive value and selection in age-structured populations

As genes flow between different states of the world, this may structure the ancestry of a population, such that different classes of individual contribute differently to future generations - i.e. they have differential reproductive value. Such class structure has important consequences for a range of evolutionary processes, shaping the effects of selection, mutation, and random drift. Yet there has been a longstanding confusion surrounding the relationship between reproductive value and evolutionary change in age-structured populations. Here we provide a synthesis of recent research in order to clarify the fundamental link between reproductive value and the force of selection as a function of an individual's age and use this understanding to resolve a recent debate as to the link between extrinsic mortality and the evolution of senescence. We also discuss how reproductive value naturally emerges in several other problems involving age-structure, including calculations of effective population size, effective mutation rate, and the definition of generation time.

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Does genetic diversity predict fitness in wild arthropod populations?

Does genetic diversity predict fitness in wild arthropod populations? A positive relationship between genetic diversity and population fitness is a fundamental expectation of population genetic theory. We have evidence for this in laboratory or captive populations; however, when population genetic diversity is estimated in a laboratory setting, effective population size is typically reduced artificially by using inbred lines. This makes it difficult to disentangle the effects of inbreeding from diversity in standing genetic variation at population level. Studies on genetic diversity in natural populations show ambiguous effect on population performance, and data from some taxonomic groups are scare. Here I performed a systematic review on the relationship between genetic diversity and performance in natural populations of arthropods. With the criteria that the relationship between estimates of genetic diversity and at least one component of fitness was determined in three or more populations in the wild, it resulted in only 33 studies. The majority studies were on immunity and reproductive performance, mostly demonstrating a positive correlation between genetic diversity and performance. For other performance traits, the data was scanty with mixed results. A strong taxonomic bias was detected with most studies performed on social hymenopteran species. Furthermore, most studies used relatively few microsatellite markers, which may not accurately capture genome-wide genetic diversity. This work reveals important gaps in the field, including the need for studies that include estimates from a higher number of populations, from a wider range of arthropod taxa and performance traits, and using whole-genome methods to obtain more markers to robustly detect relationships between genetic diversity and population performance.

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Investigating the Functional Effects of Genes Involved in Male Genital Divergence between Drosophila Species

The male genitalia are among the most rapidly evolving structures within the animal kingdom. Within the fruit fly genus, Drosophila, males of the Drosophila melanogaster complex display dramatic variation in the size and shape of the substructures of the genitalia. Tartan and sox21b have been previously identified as genes contributing to male genital size differences between two closely related species, *Drosophila simulans* and *Drosophila mauritiana*. My research investigates the evolutionary forces that drove the evolution of these genes by firstly determining if the male genital size differences mediated by these genes contribute to sexual selection within species and reproductive isolation between species. Further, by investigating the pleiotropic effects of these genes in other structures by considering the expression of regulatory elements at various developmental time points within the genitalia, as well as expression at other tissues out-with the male genitalia.

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Local adaptation through time and space: the case of the European barn owl

Climatic variations are known to subject living species to significant evolutionary stress. Responses can be diverse, including different adaptations to distinct environments. In the context of post-glacial recolonisation, a paradigm would suggest that such adaptation occurs in territories out of the glacial refugia. The Western Palearctic barn owl (*Tyto alba*), a raptor species that recolonised Europe from Southern refugia after the Last Glacial Maximum (LGM), offers a great opportunity to test this framework. First, we evaluated how climatic conditions encountered nowadays by the barn owl have changed compared to those from the LGM. Using Species Distribution Modelling, we found that, in Northern Europe, individuals face conditions similar to or slightly different from the ones present in the refugia 20√ï000 years ago. We scanned the whole genomes of 74 European barn owls from 9 populations using an innovative approach of population-specific FST to detect traces of selection. Combined with an RDA approach, we identified which of these regions were associated with temperature and precipitation, revealing traces of local adaptation throughout the genome in every considered population. We also identified a large genomic region indicating signs of selection in the two Southern glacial refugia. Interestingly, the Species Distribution Model showed that it is in the Iberian Peninsula that climatic changes seem to have been the greatest. Altogether, our findings challenge the paradigm described above: they suggest that individuals found nowadays in the geographical locations of the LGM refugia had to adapt to changed conditions, whereas on the other hand, individuals outside of refugia face conditions similar to those in the refugia 20000 years ago, leading to lower selective pressures. With this study, we emphasise the importance of considering the ecological niche as a dynamic entity and show how population genomics coupled with ecological modelling may help to understand where and how species adapt to their changing environment.

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Drift and temporal dynamics of allele frequency with aestivation. How do we interpret temporal Ne estimates?

Malaria vector populations exhibit strong seasonal fluctuations that their sizes can reach very low in the dry season then rebound rapidly with the first rain, often by several orders of magnitude. One popular hypothesis is aestivation, a form of dormancy, to account for their survival in drier months as well as the expansion when conditions improve. This work quantifies its impact on the temporal change in allele frequency and Ne estimation. We show by mathematical derivation that aestivation gives conflicting temporal Ne estimates in two ways: First, the dry season Ne is overestimated by 1/1-α2, where is the proportion of aestivating individuals. Second, the overall Ne can be lower than the harmonic mean of those from shorter intervals. We apply our findings to estimate the breeding sizes and the hidden aestivating size for a dataset collected from Thierola of Mali between 2008-2010.

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Threading new data into reconstructed genealogies

The evolutionary history of a sample of sequences is fully captured by their genealogy in the form of an ancestral recombination graph (ARG), which contains information on shared ancestors and the evolutionary events that have created the observed present-day genetic diversity. The problem of reconstructing plausible genealogies for a given input dataset has seen significant recent progress, with several powerful and scalable tools now available. The related problem of adding a new sequence to a fixed reference ARG has recently drawn significant interest, motivated by the scalability this approach offers when working with very large genealogies, the possibility of adding new data without repeating computation, and as a way of resampling ARGs. We present a new approximate but principled method for accurately threading a sequence into an inferred genealogy (with the method being exact in certain cases). We use the timing of the mutations that are shared by the reference genealogy and the new sequence, to construct a state space of possible branches with which the new sequence can coalesce at each genomic location. Simulation studies with human-like parameters demonstrate that the method maintains excellent accuracy while scaling well (sub-linearly in the sample size) and running in a fraction of the time compared to de novo genealogy reconstruction. Our method naturally handles issues such as the presence of recurrent mutation or low data quality and can be readily applied to incorporate various types of genetic data into reference genealogies, for instance unphased genotype data, or non-contemporary samples (such as ancient DNA).

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How ecological inheritance shapes polymorphism

By consuming resources, polluting, or engineering, organisms continuously modify their environment, often impacting the fitness of current but also future conspecifics. When such ecological inheritance disproportionately affects kin, selection favours environmental modifications that increase the fitness of downstream individuals. This can in turn steer the gradual evolution of ecologically-relevant traits, e.g. lowering the attack rate on a resource or enhancing the maintenance of a niche. How selection shapes variation in such traits within populations, however, remains poorly understood. In particular the conditions under which selection becomes disruptive and leads to ecological polymorphism are still unclear. To fill this gap, we investigate mathematically the coevolution of multiple traits in a group-structured population when these traits affect the group environment, which is then bequeathed to future generations. We examine when such coevolution leads to polymorphism as well as the type of associations among traits that are favoured by selection. We find that due to ecological inheritance towards kin, a positive association among two traits is favoured (i) when these traits have synergistic effects on the environment (which increases fitness); or when one trait improves the environment while the other (ii) has synergistic effects with the environment on fitness; or (iii) increases the likelihood that future kin benefit from this environment. To illustrate this, we model the coevolution of (a) the attack rate on a renewable resource, which deteriorates environmental conditions for future generations, with (b) dispersal between groups, which reduces the likelihood that kin suffers from such deterioration. We find that under a wide spectrum of conditions, selection favours the emergence of two highly-differentiated morphs: one that readily disperses and depletes local resources; and another that maintains these resources and tends to remain philopatric. Remarkably, these two consumer types coexist in spite of relying entirely on the same resource. Beyond this example, our results suggest that ecological inheritance can contribute to variation of functional traits and lead to complex ecological polymorphism.

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The genomics of parasitism loss in European lampreys

Major evolutionary shifts over short timescales provide an excellent window into the processes and mechanisms underlying phenotypic evolution and speciation. However, the genomic basis underlying such major phenotypic shifts often remains unknown, limiting our understanding of the mechanisms underlying the origin of novel phenotypes. Lampreys haven't changed much in their appearance over the last 380 million years, yet they show drastic life-history variation that has repeatedly evolved within and across taxa. In the European river lamprey complex (Lampetra sp.), up to three life-histories with different trophic and migratory phenotypes co-exist: the ancestral anadromous-parasitic species, a lake-migratory parasitic form, and the resident non-parasitic brook lamprey. To investigate the evolutionary history and genomic architecture of life-history evolution in European *Lampetra*, we combine whole-genome resequencing of all three life-history forms from multiple populations across Europe with the first chromosome-scale *Lampetra* reference genome. We found evidence for a single loss of parasitism in the Northern European *Lampetra* lineage, with potential additional losses in Southern Europe. Interestingly, lake-migratory parasitic lamprey were highly divergent from the other two life-history forms, suggesting an early split of parasitic forms prior to the loss of parasitism in Northern Europe. Genomic comparisons between life-history forms revealed a few large genomic regions that were consistently divergent between parasitic and non-parasitic *Lampetra*. By making use of historical admixture between parasitic and non-parasitic lamprey, we showed that these divergent regions show strong signals of selection against introgression in non-parasitic lamprey. This further suggests that these genomic regions are involved in the loss of parasitism and potentially in speciation. Overall, our results reveal a complex evolutionary history of parasitism loss in European *Lampetra* and highlight a major role of a few large-effect loci in shaping lamprey life-history evolution. These results highlight the usefulness of admixture events in mapping the genomic basis of major phenotypic shifts and generally broaden our understanding of life-history evolution in an ancient vertebrate group.

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Conservation genetics of Galápagos mockingbirds: from immune genes to genomes

Limited population size modulates molecular evolution at the population level by increasing the rate of genetic drift and inbreeding. The accumulation and expression of deleterious variation in small populations is a major concern in the field of conservation genetics. Impoverished genetic variation is another problem that small populations face in the longer term due to the increased rate of genetic drift. These inheritance factors may exacerbate the vulnerability of small populations, but it is also possible that other mechanisms such as purging and the evolution of drift robustness outweigh them. Understanding the population genetic consequences of limited population size is still fraught with many puzzles. In my PhD project, I focused on a dozen populations of 4 mockingbird species living on islands of different sizes in Galápagos where migration is limited. We investigated the extent to which genetic drift, determined by island size, affects genetic polymorphism and the resulting variation in molecular phenotypes in multiple immune genes, and whether whole-genome genetic load is a function of island size. First, we have shown that polymorphism in the major histocompatibility complex gene, an important immune receptor, is determined by island size, but the number of functional supertypes was conserved in all populations. Second, we have shown that the three pathogen recognition receptors from the Toll-like receptor family are subject to different constraints by purifying selection. Again, variation in molecular phenotype was robust to population size, except for the smallest population of *Mimus trifasciatus* with an effective population size of less than 20. Third, re-sequencing the whole genome of 8 populations of mockingbirds showed that neutral diversity as well as inbreeding rate scale negatively with island size, but the amount of additive genetic load has the opposite trend, at least in populations of *M. parvulus*, indicating the effect of purging. Overall, our empirical study of molecular variation in natural populations has shown that neutral polymorphism is strongly dependent on island size. However, this effect of genetic drift blurs at the level of phenotypic or functional molecular variation in immune genes, and the accumulation of genetic load across the genome is likely to be outweighed by purging. This suggests that size-limited populations are to some extent resistant to the concept of adverse effects of inheritance in small populations.

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Quantifying the consequences of haploid selection inferred on springtail sex chromosomes

Quantifying the effects of various evolutionary pressures is difficult. That is mostly, because the species we analyse are usually subjected to multiple pressures at times and disentangling those is sometimes rather difficult. One of the long standing discussions is whether the homogametic sex chromosomes (X or Z) are expected to be feminised (for spending majority of their time in females) or masculinised (for experiencing haploid selection in males) as compared to autosomes. Globular springtails (symphypleona) are an order of hexapods with a unique inheritance that allows to test this prediction empirically. Males are initially diploid, but pass only the maternal genome to the next generation. Furthermore, two of the paternal chromosomes are eliminated during early embryogenesis (X chromosomes), while four more chromosomes remain diploid during the lifespan of a male and get eliminated during spermatogenesis (autosomes). We sampled globular springtails across the UK for both whole genome resequencing and RNA-seq experiments to measure the efficacy of selection as well as relative levels of masculinisation and/or feminisation on autosomes and X chromosomes respectively.

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Bottlenecks can constrain and channel evolutionary paths

Population bottlenecks are commonplace in experimental evolution, specifically in serial passaging experiments where microbial populations alternate between growth and dilution. Natural populations also experience such fluctuations caused by seasonality, resource limitation, or host-to-host transmission for pathogens. Yet, how unlimited growth with periodic bottlenecks influence the adaptation of populations is not fully understood. Here we study theoretically the effects of bottlenecks on the accessibility of evolutionary paths and on the rate of evolution. We model an asexual population evolving on a minimal fitness landscape consisting of two types of beneficial mutations with the empirically supported trade-off between mutation rate and fitness advantage, in the regime where multiple beneficial mutations may segregate simultaneously. In the limit of large population sizes and small mutation rates, we show the existence of a unique most likely evolutionary scenario, determined by the size of the wild-type population at the beginning and at the end of each cycle. These two key demographic parameters determine which adaptive paths may be taken by the evolving population by controlling the supply of mutants during growth and the loss of mutants at the bottleneck. We do not only show that bottlenecks act as a deterministic control of evolutionary paths but also that each possible evolutionary scenario can be forced to occur by tuning demographic parameters. This work unveils the effects of demography on adaptation of periodically bottlenecked populations and can guide the design of evolution experiments.

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Investigating the population history of present-day Georgia using diachronic archaeogenomic data

The South Caucasus was a hub of cultural innovations within West Eurasia throughout prehistory. Starting around 1000 BCE, significant societal changes led to language diversification and the formation of the first states of Colchis and Iberia kingdoms in the region of present-day Georgia. Throughout this period, Greek colonies were established on the Black Sea coast to the west, while to the south the degree of suzerainty of the Persian Median empire on Colchis remains unclear. In the 4th century CE the Kingdom of Iberia was one of the first states to convert to Christianity, and later, during the migration period of the Early Middle Ages, the presence of various groups from Inner Asia and East Europe (i.e. Alans, Huns, and Mongols) is attested. It is unclear to what degree these diverse contacts were linked to movement of people. To resolve this question and to understand how various modes of gene flow shaped the population genetics of present-day Georgia, we generated genome-wide aDNA data for a time-transect of more than 200 Georgian individuals from the Early Bronze Age to the Early Middle Ages. This is the first time that aDNA from historical periods in the Caucasus is analyzed at this scale. Our analyses identify a remarkable genetic continuity throughout more than three millennia, with a small portion of non-local genetic outliers. We also found a genetic pattern reflecting the geographical structure between west and east Georgia since Middle Bronze Age. A relatively large portion of high-coverage data enabled us to apply new IBD-segment methods and to study haplotype structure among individuals buried a few kilometers apart. By analyzing background relatedness, we found that in contrast to the smaller-sized Early Middle Ages villages, the contemporaneous capital harbored large populations. Finally, we identified that several Early Medieval individuals with artificial skull elongations genetically originated from Central Asia, Central Europe and North Caucasus, showing that this cultural practice was linked to movement of people.

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Coevolution of species' niche and range in spatially and temporally variable environments

How does a species' range evolve when environment changes both in time and space, and what rate of change can a species still withstand? As the environment varies through time and space, both the species' niche and its geographic range change. Typically, to reduce the computational complexity, the problem has been considered assuming fixed genetic variance. Yet, both spatial and temporal variation influence genetic variation: genetic variance generally increases with spatial and/or temporal variation (until it the change becomes too large). This study uses dimensional analysis and simulations to integrate earlier theories and highlight the effect of the interaction between spatial and temporal variation. Specifically, I show that in large populations, spatial variability enables adaptation to faster temporal change. In contrast, in small populations, the interaction is more complex: adaptation to temporally changing optimum is hindered when spatial heterogeneity is large. As neighbourhood size decreases, the threshold where genetic drift overwhelms adaptation is approached - and the extra lag load due to temporal change (in addition to load due to dispersal across heterogeneous environments) starts to prevent adaptation. Thus, the ability of the population to adapt to temporally varying environment can change abruptly as the population's neighbourhood size (the size of the population within one-generation dispersal range) decreases.

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Sexual dimorphism in recombination rates & landscapes in wild house sparrows.

Meiotic recombination is an essential feature of sex and an important driver of diversity of eukaryotic genomes. It is beneficial as it increases genetic variance for fitness and ensures proper segregation of chromosomes during meiosis. However, it is also costly, as it is mutagenic and can uncouple allele combinations previously built up by selection. This cost/benefit dynamic may vary depending on evolutionary context, leading to variation in recombination rates from the chromosome to species level. Recombination often shows large differences between the sexes. This can vary in direction and strength, but why this happens remains poorly understood. Investigating sex differences in landscapes and genetic architecture of recombination rate variation can help us to better understand this process. We used pedigree and genome-wide SNP data to characterise recombination landscapes and individual recombination rates in a wild population of house sparrows (*Passer domesticus*). We used Poisson log normal mixture models to classify genomic regions into female-, male- and unbiased recombination rates to investigate correlates for sex differences in recombination. Individual recombination rates were heritable in both sexes, with genome-wide association analyses suggesting that rate variation is polygenic i.e. driven by many small-effect loci. Our findings provide a foundation for future investigation of associations with individual fitness and potential microevolution of recombination.

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Quantitative genetic variation from new mutations in mice

TBC

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Weakly deleterious natural genetic variation greatly amplifies probability of resistance in multiplexed gene drive systems

Evolution of resistance is a major barrier to successful deployment of gene drive systems to suppress natural populations, which could greatly reduce the burden of many vector borne diseases. Multiplexed guide RNAs that require resistance mutations in all target cut sites is a promising anti-resistance strategy, since in principle resistance would only arise in unrealistically large populations. Using novel stochastic simulations that accurately model evolution at very large population sizes, we explore the probability of resistance due to three important mechanisms: 1) non-homologous end-joining mutations, 2) single nucleotide mutants arising de novo or, 3) single nucleotide polymorphisms pre-existing as standing variation. Our results explore the relative importance of these mechanisms and highlight a complexity of the mutation-selection-drift balance between haplotypes with complete resistance and those with an incomplete number of resistant alleles. We find this leads to a qualitatively new phenomenon where weakly deleterious naturally occurring variants greatly amplify the probability of multi-site resistance compared to de novo mutation. This key result provides design criterion for anti-resistance multiplexed systems, which in general will need a larger number of gRNAs compared to de novo expectations. This theory may have wider application to the evolution of resistance or evolutionary rescue when multiple changes are required before selection can act.

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Simultaneous Inference of Past Demography and Selection from the Ancestral Recombination Graph under the Beta Coalescent

The reproductive mechanism of a species is a key driver of genome evolution. The standard Wright-Fisher model depict reproduction of individuals in a population and assumes that each individual produces a number of offspring negligible compared to the total population size. Yet many species of plants, invertebrates, prokaryotes or fish exhibit neutrally skewed offspring distribution or strong selection events yielding few individuals to produce a number of offspring of up to the same magnitude as the population size. As a result, the genealogy of a sample is characterized by multiple individuals (more than two) coalescing simultaneously to the same common ancestor. The current methods developed to detect such multiple merger events do not account for complex demographic scenarios or recombination and require large sample sizes. We tackle these limitations by developing two novel and different approaches to infer multiple merger events from the ancestral recombination graph (ARG): a sequentially Markovian coalescent (SM_C) and a graph neural network (GNNcoal). We first give proof of the accuracy of our methods to estimate the multiple merger parameter and past demographic history using simulated data under the _-coalescent model. Secondly, we show that our approaches can also recover the effect of a positive selective sweeps along the genome. Finally, we are able to distinguish skewed offspring distribution from selection while simultaneously inferring the past variation of population size. Our findings stress the aptitude of neural networks to leverage information from the ARG for inference, and the added value of comparing ad hoc SMC and GNN methods.

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The patterns of codon usage between chordates and arthropods are different but co-evolving with mutational biases

Different frequencies amongst codons that encode the same amino acid (i.e. synonymous codons) have been observed in multiple species, and studies have focused on uncovering the dynamics that drive such codon usage bias. Two main narratives have been proposed: on one hand, mutations, and on the other, translational selection, are working to produce different frequencies of synonymous codons, with a combined effect being the most likely explanation. However, not many studies have been able to measure and disentangle these effects that leave similar traces on the genome in multiple species. Here, we have developed a codon model that allows the disentangling of mutation, selection on amino acids and synonymous codons, and GC-biased gene conversion (gBGC). It employs a Bayesian estimator to assess the inter-species variability of codon composition found in multiple species in light of those forces. In this study, we analysed a dataset of 415 species belonging to chordates and 191 species of arthropods. We observed that chordates need more synonymous codon categories to explain the empirical codon frequencies compared to arthropods (29 versus 14). This suggests that chordates have more pronounced patterns of codon usage. Additionally, in both phyla, selection forces act more negatively as the GC content of a codon is increasing, indicating that selection at the genome-wide level acts to balance the mutational processes that promote G/C alleles. Methylation at CpG sites seems to also explain patterns of codon usage bias in chordates but not so much in arthropods. This study shows that while both chordates' and arthropods' codon preferences seem to be dominated by mutational biases, they differ in their extent of codon usage.

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The rate and spectrum of de novo structural mutations in *Mus musculus*

Structural mutations (SM) are known to be common in populations, but segregate at low frequencies and are expected to have large phenotypic effects. Knowledge on the rate at which new SMs arise in the genome is, however, limited. Here, we maintained mutation accumulation (MA) lines of the house mouse (Mus musculus; strain C3H) by full-sib mating, and used PacBio HiFi to sequence the MA founders and four MA lines after 15 generations. Using read, assembly, and pan-genome approaches, we estimated the rate of de novo mutation for different categories of SMs to be _=4.11x10e-10 per site and generation, implying the haploid genomic rate, M, is about one event per generation. This is about one order of magnitude lower than estimates of the single nucleotide mutation rate. Transposable element (TE) insertions represented the most common SM (_=1.10x10e-10, M=0.29), after contractions and expansions of tandem repeats. All TEs were retrotransposons, predominantly Intracisternal A Particles (59%). The presence of active LINEs and SINEs likely explains the insertion of a new gene retrocopy in one of the MA lines, implying a higher rate than previous estimates based on population data. We also found one duplication (_=0.05x10e-10, M=1.30x10e-2), presumably due to non-allelic homologous recombination, and five deletions (_=0.25x10e-10, M=6.54x10e-2), all involving TE annotation, with one of them resulting in the deletion of a previously duplicated sequence. Our results show that SMs occur at substantial rates, and that the spectrum of new SMs is dominated by mutations related to various types of repetitive sequences.

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Nanopore transcriptome skimming: a promising strategy for meiofaunal biodiversity assessment

Molecular tools have revolutionized the study of meiobenthic ecology and systematics, but to date have nearly exclusively focused on DNA templates and amplicon sequencing. The limits of this approach are widely known: single-marker phylogenies are often misleading, and high-resolution coding markers can be difficult to amplify across a diversity of taxa, forcing fallback onto more limited-resolution rDNA loci. The wide availability of nanopore sequencing, which now offers read accuracies exceeding 99% at any insert size, but at a similar marginal cost to short-read sequencing, with little up-front capital expense, motivates consideration of alternative strategies. Here, I explore the value of ONT-based sequencing of large multiplexed pools of full-length cDNAs in biodiversity assessment. The intrinsic skewness of gene expression, with especially mtDNA loci showing conserved high expression in most animal tissues, ensures that even at shallow depth (~50,000 reads per target), hundreds of genes receive sufficiently high coverage to call high-accuracy consensus sequences suitable for evolutionary inferences. This can be achieved at minimal cost (<£10/specimen), so long as specimens are sufficiently fixed to preserve RNA integrity, and specimens treated in this way can also be later used for whole genome assembly or deeper transcriptome sequencing. I will present a proof-of-concept demonstration of this approach using terrestrial microturbellarians, an overlooked taxon whose high diversity we are exploring for the first time in the UK. I further discuss strategies currently in development to increase the throughput of transcriptome skimming to 10,000s-100,000s of uniquely indexed samples, a level of multiplexing compatible with whole-community, individual-resolved transcriptome profiling of meiobenthic samples.

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How directional selection and genetic drift drive contemporary evolution of the kelp Laminaria digitata: a spatio-temporal genomic study

Analyzing genome-wide SNPs across multiple time points, rather than from a single time point allows to understand how genetic drift and directional selection drive contemporary evolution. Here, we address these issues using thousands of SNPs (ddRAD-Seq) across four populations of the cold-water kelp Laminaria digitata. These populations were sampled at two time points, (separated by about ten years) along a latitudinal gradient. Different outlier detection tests, including simulations to analyse neutral trajectories of allele frequencies, were ran at both temporal and spatial scales. These tests led to the identification of 52 outlier SNPs, with ten of them belonging to a family of enzymes involved in metabolic processes. Our results from the spatial scale indicated that a significant number of outliers could be involved in response to sea-surface temperature (SST) variation. This finding was consistent with the inference of two temporal outliers identified twice in two populations exposed to the highest mean SST. On the contrary, estimates of contemporary effective size, and temporal variance in allele frequencies sustained the hypothesis that genetic drift was more pronounced in peripheral than central populations. This would suggest a lower evolutionary potential of peripheral populations that can be questioned by the lack of detection or poor detectability of temporal outliers. Overall, our results point to the importance of combining spatial and temporal genome-wide investigations to give more insights into the contemporary evolution of key species facing environmental changes.

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Quantifying Inbreeding Depression

The measurement of inbreeding and its impact on fitness is essential in many areas of biology, including human genetics and the conservation of endangered species. Individual inbreeding coefficients used to be estimated with pedigrees, but nowadays genomic based estimates have proven to be more robust. However, there is no consensus on how to best estimate inbreeding from genomic markers in order to infer inbreeding depression. To address this question, we simulated traits based on empirical human whole genome sequencing data from populations of various sizes (from the 1000 Genomes Project) and on simulated genomes from a large pedigree of a polygamous species. We investigated how different genomic estimators of individual inbreeding coefficients (based on Identity by descent segments or on individual SNPs) as well as the partitioning of allele frequency and linkage disequilibrium spectrum affect inbreeding depression detection and accuracy. Critically, we show that partitioning the allele frequency spectrum or linkage disequilibrium does not improve the estimation of inbreeding depression, but we found that accounting for the non-independence of observations is essential.

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How Effective is Sexual Selection in Purging Deleterious Mutations in Experimentally Evolved *Drosophila*?

Most new mutations are neutral or deleterious. Sexual selection is hypothesised to be a force for purging deleterious mutations from the genome, as secondary sexual characteristics can act as a signal for the fittest members with the lowest genetic load to mate and spread their genes, while individuals with a high proportion of deleterious mutations fall out of the mating pool. Here we investigate the contribution of sexual selection on purging the genome of deleterious mutations using *D. pseudoobscura* fruit fly populations whereby the extent of sexual selection has been manipulated and strictly controlled. Females from these populations have been kept either in enhanced polyandry (6 males and 1 female), or strict monogamy (1 male and 1 female) for 200 generations. We used polyDFE to calculate the distribution of fitness effects in each treatment at generation 85 and 200 over the course of the experiment and found that strictly monogamous populations contain a significantly smaller proportion of highly deleterious mutations than polyandrous populations across the genome. We also see that this difference between treatments increases greatly between generation 85 and 200 on the 3rd and X chromosomes, which have been previously identified as containing sexual selection gene hotspots. We conclude that sexual selection, rather than purge the genome of highly deleterious mutations, instead may induce a higher genetic load brought about by high sexual conflict and a widening gap between the fitness and sexual selection optima.

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Genome downsizing after polyploidy: mechanisms, rates and selection pressures

The amount of DNA in each cell of an organism, called the 'genome size', varies enormously between species. Indeed, amongst flowering plants there is a known ~2,400 fold range in genome size (from 60,000 -150,000,000 bp per 1C genome). However, most plant species have smaller genomes than would be expected given the incidence of polyploidy in their ancestry, suggestive of selection against large genome sizes. This talk explores potential selection pressures acting on genome size, focussing in particular on nutrient limitation. We show from grassland nutrient experiments in UK, Germany, Inner Mongolia and from the Nutrient Network that species with large genomes are favoured in the presence of nitrogen and phosphate fertilizers, suggestive of selection against large genomes when they are limiting (as in most soils of the world). But available data suggest that there are only small rates of DNA loss per generation (4-70 Mb/million years, <500 bp/generation) which poses a problem as to how selection could act given the size of even the smallest plant genome is three orders of magnitude larger. I propose that a solution to this problem is that genome downsizing is an emergent property of polyploidy, which after time has ecological advantages to plants growing in nutrient poor habitats.

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Sex allocation trade-offs can explain the evolution of male and female heterogamety

Sex chromosomes, defined as chromosomes bearing a sex-determining region (SDR), are a common sex determination system in species with separate sexes. In most cases, the SDR behaves as a diallelic locus with Mendelian inheritance, where one allele causes its bearer to develop as a female while the other causes it to develop as a male. Complete dominance of one allele over the other is found at the SDR, so that one sex is always heterozygous (heterogametic), while the other is always homozygous (homogametic). Most theory developed on sex chromosomes evolution postulates the existence of an SDR, and focuses either on the evolutionary consequences of heterogamety or on the turnover between already established male and female heterogametic systems (XY and ZW systems). However, the evolutionary mechanisms leading to the emergence of an SDR in species that were previously devoid of it, and in particular the mechanisms leading some species to acquire an XY rather than a ZW system, remain elusive. Here, we model the co-evolution of sex allocation and its underlying genetic architecture in diploids, and show that an SDR can readily emerge from this co-evolution. Furthermore, we show that whether male or female heterogamety evolves is dictated by the shape of gain curves.

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The establishment of locally adaptive inversions in structured populations

Inversions have been proposed to facilitate local adaptation, by linking together locally coadapted alleles at different loci. Classic prior work addressing this question theoretically has considered the spread of inversions in “continent-island” models in which there is a unidirectional flow of maladapted migrants into the island population. In this setting, inversions are most likely to establish when selection is weak, because stronger local selection more effectively purges maladaptive alleles, thus lessening the advantage of inversions. Here, we show this finding only holds under the limited conditions studied. We study the establishment of inversions in a “two-deme” model, which explicitly considers the dynamics of allele frequencies in two demes linked by bidirectional migration. For symmetric selection and migration, we find that stronger local selection increases the flow of maladaptive alleles and favours inversions, the opposite of the pattern seen in the asymmetric continent-island model. Furthermore, we show that the strength and symmetry of selection and migration not only alter the establishment probability of adaptive inversions, but also change the likelihood that an inversion captures an adaptive haplotype in the first place. Considering the combined process of capture and invasion shows that inversions are most likely to be found when locally adaptive loci experience strong selection. In addition, inversions that establish in one deme also protect adaptive allele combinations in the other, leading to differentiation between demes. Stronger selection in either deme once again makes differentiation between populations more likely. In contrast, differentiation is less likely when migration rates are high because adaptive haplotypes are rare. Overall, this analysis of evolutionary dynamics across a structured population shows that established inversions are most likely to have captured strongly selected local adaptation alleles.

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The role of epigenomic mechanisms in species adaptation to environmental change.

Fellowship pitch: Climate change and anthropogenic impact are currently the major drivers of species loss. Understanding how species adapt to rapid environmental change is vital if we are to mitigate these losses. The field of evolution is in the midst of a paradigm shift with researchers acknowledging that molecular mechanisms beyond the stable genome contribute to adaptive processes. Epigenetic modifications are one such mechanism, whereby chemical modifications of the DNA can affect how the underlying genes function and produce phenotypic variation. Epigenetic mechanisms are predicted to be a valuable source of selective substrate in clonal and highly inbred species which are particularly vulnerable to rapid environmental change. This fellowship will utilise experimental evolution and population epigenetics in order to identify a potential role for DNA methylation (an epigenetic mark) in species adaptation to climate warming. Transgenerational experiments using three species of arthropod will determine inducible and heritable DNA methylation marks. The relationship between DNA methylation and the underlying genome and any phenotypic responses in terms of fitness changes and changes in gene expression will be assessed. Using three species will also allow the identification of a potentially conserved response across arthropods. Finally, wild populations of these species across a temperature gradient will be sampled, in order to establish if inducible and heritable DNA methylation marks are found in natural populations. This project will deliver a significant development in our understanding of how epigenetic mechanisms could contribute to species adaptation to a major driver of species loss worldwide.

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Introduction to a crop wild relative to investigate plant & pathogen co-evolution

Agriculture is a relatively recent phenomenon. This means that crop pathogens have recently specialised from their wild hosts. Crop domestication has reduced genetic diversity, which are no longer continually co-evolving with pathogens in the same way that wild hosts do. Crop-pathogen systems represent an idealised natural experiment to understand pathogen adaptation. In addition, many pathogens can survive on both crops and their wild relatives. This opens enormous potential to understand the evolution of wild host resistance as well as pathogen emergence, invasion, and adaptation to monocultures. Here we introduce the beet-rust pathosystem. Sugar beet is one of the most recently domesticated crops and suffers yield loss due to the obligate biotrophic rust that lives on both wild and crop hosts. We use this wild-agricultural system to outline methodologies for understanding co-evolution in host and pathogen. In wild sea beets we test the feasibility of GWAS to identify novel resistance to a crop rust. In the rust pathogen we are using measures of genetic differentiation to highlight genes that may be important for adaptation specifically to the beet crop. We are developing this system as a model for surveillance to identify pathogen emergence as well as durable wild resistance.

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Inference of host-parasite interaction matrices using genome-wide polymorphism data

Coevolution is driven by genotype x genotype (G x G) interactions between hosts and their symbionts. Recent developments in joint genome wide association analysis of host and pathogen polymorphism data greatly advanced the identification of the genes underlying these interactions. However, deciphering the specific form and magnitude of these GxG interactions has proven difficult and currently relies on experimental approaches. Based on a theoretical model of host-parasite interactions, we first derive four statistics to complement joint genome wide association studies. We demonstrate the efficiency of these indices in discriminating various GxG interaction matrices in simulated data sets by numerical estimation integrated into an ad hoc Approximate Bayesian Computation method. The indices are based on extracting relevant information from polymorphism data of randomly sampled uninfected hosts and randomly sampled infected host and their respective parasite strains. Second, we apply our method to a SNP data set of 451 European humans and their infecting HCV virus strains supplemented by polymorphism data from the 1000 genomes project. Our model based approach recaptures ca. 150 significant interactions previously found. By applying our inference framework, we show, that these significant interactions between MHC-genes and HCV genes seem not to follow matching-alleles interactions but rather gene-for-gene interactions. We speculate that this relationship is due to an only recent expansion of HCV population in Europe and the low prevalence of HCV in the human population.

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Investigating the drivers of diversity maintenance after a colonisation bottleneck

The factors that contribute to diversity maintenance during a population bottleneck remain poorly understood, particularly the relative importance of balancing selection at a genome-wide scale. I used whole genome sequence data from an isolated population of butterfly, *Danaus chrysippus*, to reconstruct the population's demographic history and investigate the role of neutral and selective processes in maintaining genetic diversity during a recent bottleneck. While sequence data revealed diversity remained relatively high, inbreeding was41.25x greater than the source population, indicating diversity may not correlate simply with population health. I was unable to accurately estimate the demographic history of the population using leading genomic tools, calling into question their suitability in non-model species. I used simulations to create a model that fit the data well, suggesting a bottleneck~363 years ago which reduced the population from 5m to 100 individuals, followed by a 50year recovery to a current Ne of 50,000. Simulations also revealed an association between genome-wide polymorphism and population recovery speed, suggesting that the speed of population recovery was a major factor contributing to maintenance of polymorphism throughout the genome. I found no evidence for selection maintaining genome-wide polymorphism, suggesting neutral processes such as recovery speed were largely sufficient for genome-wide diversity maintenance. However, the level of heterozygosity at genome-wide polymorphic loci was significantly higher than expected, suggesting the presence of balancing selection. Further research is needed to determine if the balancing selection detected was an important driver of diversity maintenance during the bottleneck or is simply acting on the polymorphism maintained by neutral processes.

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Parallel Evolution in the Integration of a Co-obligate Aphid Symbiosis

Insects evolve dependence - often extreme - on microbes for nutrition. This includes cases in which insects harbour multiple endosymbionts that function collectively as a metabolic unit. How do these dependences originate, and is there a predictable sequence of events leading to the integration of new symbionts? While co-obligate symbioses, in which hosts rely on multiple nutrient-provisioning symbionts, have evolved numerous times across sap-feeding insects, there is only one known case in aphids, involving *Buchnera aphidicola* and *Serratia symbiotica* in the Lachninae subfamily. Here, we identify three additional independent transitions to the same co-obligate symbiosis in different aphids. Comparing recent and ancient associations allow us to investigate intermediate stages of metabolic and anatomical integration of Serratia. We find that these uniquely replicated evolutionary events support the idea that co-obligate associations initiate in a predictable manner: through parallel evolutionary processes. Specifically, we show how the repeated losses of the riboflavin and peptidoglycan pathways in *Buchnera* lead to dependence on *Serratia*. We then provide evidence of a stepwise process of symbiont integration, whereby dependence evolves first. Then, essential amino acid pathways are lost, which coincides with the increased anatomical integration of the companion symbiont. Finally, we demonstrate that dependence can evolve ahead of specialized structures, and in one case with no direct nutritional basis. More generally, our results suggest the energetic costs of synthesizing nutrients may provide a unified explanation for the sequence of gene losses that occur during the evolution of co-obligate symbiosis.

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Slower-X: Reduced efficiency of selection in the early stages of X

Differentiated X chromosomes are expected to have higher rates of adaptive divergence than autosomes, if new beneficial mutations are recessive (the 'faster-X effect'), largely because these mutations are immediately exposed to selection in males. The evolution of X chromosomes after they stop recombining in males, but before they become hemizygous, has not been well explored theoretically. We use the diffusion approximation to infer substitution rates of beneficial and deleterious mutations under such a scenario. Our results show that selection is less efficient on diploid X loci than autosomal and hemizygous X loci under a wide range of parameters. This 'Slower-X' effect is stronger for genes affecting primarily (or only) male fitness, and for sexually antagonistic genes. These unusual dynamics suggest that some of the peculiar features of X chromosomes, such as the differential accumulation of genes with sex-specific functions, may start arising earlier than previously appreciated.

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Mutation, recombination, and transposition rates in *Drosophila melanogaster* and *D. simulans*

Mutation, recombination, and transposition rate are core parameters in models of evolution. They impact genetic diversity, responses to ongoing selection, and levels of genetic load. However, even for models such as *Drosophila melanogaster* and *D. simulans*, few estimates are available and we have little idea of how rates vary between individuals, sexes, populations, or species. Here we provide direct estimates of these rates for a West African and a European population of D. melanogaster and a European population of *D. simulans*. Compared to the European D. melanogaster, we find the West African population has a lower mutation rate and transposition rate, but a higher recombination rate. The European *D. simulans* population has a similar mutation rate to European *D. melanogaster*, but a significantly higher recombination rate and a (non-significantly) lower transposition rate. Overall, we find paternal-derived mutations are more frequent than maternal ones in both species

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Local genetic adaptation in chimpanzees

Chimpanzees, along with bonobos, are our closest living relatives and are endangered, with numbers in continuous decline. They inhabit a diversity of habitats in sub-Saharan Africa, from cool wet rainforest to hot dry savannah, which provides the opportunity for local genetic adaptations. Understanding such adaptations would provide fundamental insight into chimpanzee evolution and essential information for planning conservation efforts. However, studies of fine-scale adaptation in chimpanzees have been hindered by the difficulties associated with accessing genetic samples from endangered wild populations. Our work analyses a new dataset of unprecedented scale, consisting of 828 exomes from non-invasive samples of wild chimpanzees, together with newly collected environmental data, to investigate local adaptation in chimpanzees. We find that SNPs with exceptionally large allele frequency differences between populations were enriched for virus-response functions, highlighting the importance of pathogen-mediated adaptation in chimpanzees in the recent past. Together with our previous work investigating ancient adaptation in chimpanzees (Schmidt et al., 2019; Pawar et al., 2022), our results strongly suggest that pathogens in general, and the Simian Immunodeficiency Virus (SIV) in particular, have been major selection pressures throughout chimpanzee evolution. Infectious disease is currently a major cause of mortality in wild chimpanzee populations, therefore understanding patterns of local adaptation to pathogens has important implications for chimpanzee conservation. Adaptation to environmental factors is also important, and critically relevant for conservation given the potential for population-specific adaptations to highly localised habitat variables. To gain insights into adaptation to a range of habitat-related environmental pressures, we integrate a wealth of environmental variables with the exome sequence data using genotype-environment association (GEA) analyses. We identify genome-wide significant signatures of genetic adaptation within subspecies, as well as some evidence of potential convergent adaptation across subspecies, especially along the forest-savannah habitat gradient. Our work demonstrates how non-invasive sampling can be used to investigate fine-scale adaptation in endangered populations of large mammals. We find evidence of genetic adaptation to pathogens and habitat type in chimpanzees which has important implications for the conservation of this species.

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Demography and evolutionary history of grey wolf populations around the Bering Strait

Glacial and interglacial periods throughout the Pleistocene have been substantial drivers of change in species distributions. Earlier analyses suggested that modern grey wolves (*Canis lupus*) trace their origin to a single Late Pleistocene Beringian population that expanded east and westwards, starting ca. 25,000 years ago (ya). Here, we examined the demographic and phylogeographic histories of extant populations around the Bering Strait with wolves from two inland regions of the Russian Far East (RFE) and one coastal and two inland regions of North-western North America (NNA), genotyped for 91,327 single nucleotide polymorphisms. Our results indicated that RFE and NNA wolves had a common ancestry until ca. 34,400 ya, suggesting that these populations started to diverge before the previously proposed expansion out of Beringia. Coastal and inland NNA populations diverged ca. 16,000 ya, concordant with the minimum proposed date for the ecological viability of the migration route along the Pacific Northwest coast. Demographic reconstructions for inland RFE and NNA populations reveal spatial and temporal synchrony, with large historical effective population sizes that declined throughout the Pleistocene, possibly reflecting the influence of broad-scale climatic changes across continents. In contrast, coastal NNA wolves displayed a consistently lower effective population size than the inland populations. Differences between the demographic history of inland and coastal wolves may have been driven by multiple ecological factors, including historical gene flow patterns, natural landscape fragmentation, and more recent anthropogenic disturbance.

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A dynamic epibiont community associated with the bone eating worm Osedax

Osedax, the deep-sea annelid found at sunken whalefalls, is known to host bacterial endosymbionts intracellularly in specialized roots that help it feed exclusively on vertebrate bones. Past studies, however, have also made mention of external bacteria on their trunks. During a 14-year study, we reveal a dynamic, yet persistent, succession of epibionts integrated into the epidermis of Osedax that change over time as the whale carcass degrades on the sea floor. The epibionts associated with seven species of Osedax are initially dominated by the genus Arcobacter (at early time points < 24 months), Sulfurospirillum at intermediate stages (~ 50 months), and Sulfurimonas at later stages (>140 months) of whale carcass decomposition. Metagenome analysis of the epibiont metabolic capabilities suggests a transition from heterotrophy to autotrophy along the successional gradient, and differences in their capacity to metabolize oxygen, carbon, nitrogen, and sulfur. Compared to free living relatives, the epibionts were highly enriched in transposable elements, implicating genetic exchange on the host surface, and contained numerous secretions systems with eukaryotic-like protein domains, suggesting a long evolutionary history with these enigmatic, yet widely distributed deep-sea worms.

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Rapid evolution of the regulatory network formed by miRNAs and their target genes in *Arabidopsis halleri* and *A. lyrata*.

MicroRNAs (miRNAs) are a class of small non-coding RNAs that play important regulatory roles in plant and animal genomes. They are produced from a primary transcript forming a short foldback structure from which a mature miRNA is processed and negatively regulates a series of mRNA targets. Identifying the forces under which the regulatory network formed by miRNAs and their target genes can be modified in the course of evolution is an important question. To address this question, we first de novo assembled a reference genome and used deep small RNA sequencing to perform a detailed annotation of miRNA genes in the closely related species *Arabidopsis halleri* and *A. lyrata*. As many as 43% of the 256 and 232 annotated miRNA genes were specific to either species, and thus emerged recently, while 57% were deeply conserved across diverged land plant lineages. We then compared the functional constraint over these two classes of miRNA genes and their predicted target sites using whole genome sequencing data of 150 *A. halleri* and 100 *A. lyrata* individuals from across the species ranges. We observed lower nucleotide diversity in the mature miRNA sequence than in the other parts of the hairpin (stem, terminal loop), and also lower nucleotide diversity in the miRNA-targeted region as compared to the neighboring regions along the CDS. This suggests that evolution of the mature miRNA sequence and its complementarity target sequence are constrained by purifying selection. This pattern was less pronounced for the recently emerged miRNA genes than for the evolutionarily conserved ones, suggesting that young miRNA genes generally evolve under weaker selective constraints than older ones. Overall, our results suggest a rapid birth and death process of the miRNA repertoire, whereby new miRNA genes can arise quickly, a small number of which are retained, eventually integrating into ‘core’ biological processes.

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Hybrid fitness effects modify fixation probabilities of introgressed alleles

Hybridization is a common occurrence in natural populations, and introgression is a major source of genetic variation. Despite the evolutionary importance of adaptive introgression, classical population genetics theory does not take into account hybrid fitness effects. Specifically, heterosis (i.e. hybrid vigor) and Dobzhansky-Muller incompatibilities influence the fates of introgressed alleles. Here, we explicitly account for polygenic, unlinked hybrid fitness effects when tracking a rare introgressed marker allele. These hybrid fitness effects quickly decay over time due to repeated backcrossing, enabling a separation-of-timescales approach. Using diffusion and branching process theory in combination with computer simulations, we formalize the intuition behind how hybrid fitness effects affect introgressed alleles. We find that hybrid fitness effects can significantly hinder or boost the fixation probability of introgressed alleles, depending on the relative strength of heterosis and Dobzhansky-Muller incompatibilities effects. We show that the inclusion of a correction factor (representing the compounded effects of hybrid fitness effects over time) into classic population genetics theory yields accurate fixation probabilities. Despite having a strong impact on the probability of fixation, hybrid fitness effects only subtly change the distribution of fitness effects of introgressed alleles that reach fixation. Although strong Dobzhansky-Muller incompatibility effects may expedite the loss of introgressed alleles, fixation times are largely unchanged by hybrid fitness effects.

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Selective whole-genome amplification reveals population genetics of *Leishmania braziliensis* from primary patient samples

In Brazil, *Leishmania braziliensis* is the main causative agent of the neglected tropical disease, cutaneous leishmaniasis (CL). CL can present on a spectrum of disease severity and often is refractory to treatment, yet the parasite factors that may contribute to disease presentation and patient treatment outcome are not well understood, in part because successfully isolating and culturing parasites from patient lesions remains a major challenge, and because adaption to culture has been shown to induce widespread genetic changes in *Leishmania*. Here we describe the development of selective whole genome amplification (SWGA) for Leishmania and show that this method enables culture-independent analysis of whole parasite genomes obtained directly from primary patient skin samples, all while avoiding artifacts associated with adaption to culture. We show that SWGA can be applied to multiple Leishmania species residing in different host species, suggesting that this method can be broadly useful in both experimental infection models and clinical studies. Finally, we show that parasite genomes generated by SWGA of skin biopsies collected from patients in Corte de Pedra, Bahia, Brazil exhibit substantial genetic diversity and can be integrated with published whole genome data from parasites isolates to expand our understanding of Leishmania population genetics in Brazil.

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Gene duplication facilitates phenotypic innovation in ant castes

The evolution of superorganismality in ants and subsequent development of phenotypically distinct castes has provided us with one of nature's most complex polyphenisms; a single genome producing the discrete phenotypes of workers, queens and males. Our understanding of the processes required to encode such dramatically distinct types of individuals is limited, making ants an excellent system for studying phenotypic innovation. To understand the molecular mechanisms underpinning this extreme polyphenism we identified gene duplications within the genome of the red fire ant *Solenopsis invicta* dating back to the evolution of superorganismality. We generated tissue-specific whole transcriptome gene expression data from 19 distinct tissues from all three castes, the largest dataset of its kind. We characterised differences in expression of duplicated genes and gene families across the three castes. We find many examples of duplication and caste-specific subfunctionalisation, with individual genes becoming adapted to the phenotypes in which they are expressed. In particular, this process occurred extensively in chemical communication, with high expression in antenna and cuticular tissues. Our results shed light on how gene duplications allow for a single genome to encode a wide array of distinct castes in the ants and are central to phenotypic innovation in this system.

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Awkward asexual allopolyploid assembly

Genome assembly has historically been limited by the length and quality of reads available, the complexity of the target genome, and the interaction between these two factors. Long-read technology has allowed greater accuracy and contiguity in the generation of assemblies, and many chromosome-scale assemblies of non-model organisms continue to be announced. An exception to this is in the case of complex polyploids, especially allopolyploids. Increased repeat content and copy number, in addition to genome-wide ‘heterozygosity’, ensure allopolyploid assembly remains challenging, an issue which can be exacerbated by eccentric, non-model, sexual systems. I have produced a chromosomal assembly of the genome of *Meloidogyne javanica*, a mitotically parthenogenetic allopolyploid root-knot nematode, responsible for billions of dollars of agricultural losses annually. Here I discuss the modification of genome assembly approaches to take into account the difficulties of complex genomes such as from asexual allopolyploids and talk about the evolutionary questions we can address by including some non-model ‘problematic’ genomes.

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Endless forms most beautiful, some not: a novel adaptive phenotype illustrates the large mutational target of a trait under negative selection

Adaptive mutations arise very rarely and are therefore difficult to identify and characterise during their initial spread. In addition, adaptation largely occurs through changes in allele frequencies at many different loci, obscuring signatures of single adaptive de novo mutations. We recently identified a mutant phenotype ('curly-wing') which inhibits the ability of males in Hawaiian populations of field crickets (*Teleogryllus oceanicus*) to produce sexually selected song. Songlessness protects male curly-wing crickets against a fly (*Ormia ochracea*), which deposits deadly parasitic larvae onto singing male crickets. Curly-wing appears to be a newly emerged phenotype, having not been reported in long-term study populations prior to our first identification in 2017, nor in ancestral Polynesian populations of *T. oceanicus*. We assayed fitness costs and heritability of curly-wing, mapped it to a single autosomal region, and characterised transcriptomic variation in developing wing samples. Our results suggest curly-wing is probably associated with a very large genomic inversion, with pervasive effects on gene expression, and negatively impacts fitness-related traits in both sexes. Together with observations of flatwing phenotypes in the current and prior studies, our results challenge widely held notions regarding the opportunity for adaptation mutations to arise and spread, and illustrate the large mutational target presented by a trait under strong negative selection.

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Identifying the Nature of Adaptation to Micronutrients in Modern Humans

Micronutrients are necessary dietary components across many species. These trace metals and vitamins accumulate in the diet via the local plants and animals consumed, which in turn depends on the concentration of micronutrients within the local soil. Hence, species and populations occupying different environments are exposed to varying levels of these essential dietary micronutrients, which may act as a local selective pressure, particularly in environments with extreme concentrations, or lack thereof, of micronutrients. Differential levels of micronutrients in the diet are therefore pertinent for examining local adaptation in modern humans, since our species occupy an extraordinary range of environments. Moreover, micronutrients are especially essential for modern humans since, with the exception of vitamin D, they cannot be synthesised in the body. Micronutrients are optimum at only a narrow range and deficiencies can result in an increased risk of cardiovascular, metabolic and infectious disorders, particularly during periods of development. Thus, dietary levels of micronutrients present a strong candidate for a local selective force across modern human populations. We have applied recently developed tree-recording techniques and the summary statistic Fst to gene sets associated with 13 different micronutrients across 40 global populations and demonstrate widespread evidence for micronutrient levels driving adaptation across human history. Here, we contrast the evolutionary history of genes associated with the metabolism and uptake of Iron, Calcium, Zinc, Selenium and Iodine, which represent the range of a) localisation of selection signatures over geographical area and b) polygenicity of selection. We also explore potential cultural drivers for selection on Iron and Calcium in Eurasian populations, highlighting the complexity of selection history in these essential dietary components.

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Utilising rare variant sharing to identify non-recurrent rare mutations in Anopheles gambiae mosquitoes

While the sharing of mutations can be highly informative by indicating common origins between individuals, recurrent mutations - those mutants that have arisen multiple times in a population's history on different branches of the genealogy - have the potential to drastically alter the reliability of inferences made with population genomic data. While rare mutants, which have the potential to facilitate inference of recent demography, are often assumed to have such a low probability of being recurrent that the possibility is ignored, in species with large, diverse populations this is often non-negligible, and need to be excluded to ensure effective downstream analysis. However, very little work exists to address this problem. Here, we propose a probabilistic method to identifying non-recurrent rare mutations, utilising the presence of other mutually shared variants on surrounding genomic tracts. We then illustrate this approach empirically using population genomic data from the malaria vector Anopheles gambiae, specifically examining doubleton mutations, for which we estimate that up to 33% of have resulted from independent mutation events. We find that our method is well suited to this genetically diverse species, identifying ~80% of non-recurrent doubletons in the dataset at 95% confidence. We then demonstrate the utility of these non-recurrent rare variants for inference on the effects of genomic position, selection, and geography.

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Wright or Fisher? A genomic test of alternative theories of dominance

Famously discovered by Mendel in backyard experiments with garden peas, dominance is a basic component of genetics describing the unequal effects of alternative genetic variants on trait expression. But why does it exist at all? A century ago, Fisher put forward the hypothesis that modifiers evolve to mask the deleterious effects of (initially co-dominant) mutations. Wright countered instead that dominance is attributable to the concave mapping of genotype to fitness. In the intervening years, a consensus has emerged in favour of Wright, yet the evidence supporting Wright is far from definitive. Here, we use genomic data from ~125,000 human exomes to test whether mutational deleteriousness scales negatively with dominance (as predicted by Wright) or not (as predicted by Fisher). By partitioning mutations into categories of deleteriousness using genome annotations and by estimating dominance based on relative genetic loads on the X chromosome and autosomes, we can test their alternative hypotheses. Overall, our data suggest that increasingly deleterious mutations are increasingly recessive. Wright, in other words, appears to have been right.

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Two different adaptive speciation mechanisms operate during adaptation to a novel hot environment

Ecological speciation and mutation-order speciation are two different mechanisms of adaptation-driven speciation. Both mechanisms predict different patterns of reproductive isolation for replicate populations adapting to the same environment. With ecological speciation, barriers to gene flow emerge between populations from different environments, but not among replicate populations from the same environment. Mutation-order speciation predicts reproductive isolation among populations adapted to the same environment. We demonstrate that both speciation processes occurred within about 100 generations when replicate *Drosophila simulans* populations adapted to a novel, hot environment. Gene expression analysis identified the underlying molecular mechanisms. Premating ecological speciation is the byproduct of an altered lipid metabolism, which also changed the cuticular hydrocarbon (CHC) composition in hot-evolved flies. Postmating reproductive isolation supports mutation-order speciation most likely driven by co-evolution of reproduction-associated genes. Adaptation processes can rapidly induce incipient speciation and different speciation mechanisms affect pre- and postmating reproductive isolation. We propose that the definition of mutation-order speciation should be expanded to account for polygenic processes from standing genetic variation.

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Do reticulate relationships between tropical trees drive diversification?

Radiations that generate exceptionally species-rich groups are a fundamental component of diversification. Hybridisation likely underlies many radiations because it allows the interspecific transfer of genetic variants conferring adaptation to contrasting environments, often resulting in phenotypic variation exceeding that of the parental lineages (sensu the 'combinatorial' speciation model). This broadening of phenotypic variation can facilitate adaptation to novel niches and so trigger a radiation. Rapid radiations explain a significant portion of the hyperdiversity of the neotropical flora, where a single hectare of Amazonian Forest may host more tree species than all of Europe (>300 spp.). However, the influence of hybridisation on rainforest tree radiations remains largely unexplored. We address this knowledge gap using Inga, a genus of neotropical rainforest trees that typifies such recent radiations, with >300 species having arisen in the last 6Ma, and within which hybridisation occurs. Specifically, we use phylogenomic approaches to understand whether reticulation preceded rapid diversification in Inga, and hence whether hybridisation may have catalysed this radiation. Moreover, ecological work shows that insect herbivore pressure structures Inga communities, and that sister species of Inga differ significantly in their anti-herbivore defence chemistry, alluding to the role of defence chemistry in divergence. Accordingly, using whole-genome resequencing we will assess whether hybridisation facilitates the transfer of linkage blocks containing chemical defence loci, facilitating the movement of adaptive variation between Inga species. This will allow us to determine whether ecological divergence following the transfer of adaptive variants via hybridisation plays a role in generating the remarkable diversity of rainforest trees.

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Fitness landscapes and the role of ploidy in heterosis.

The genetic basis of heterosis has received much attention, but it remains unclear whether general unifying mechanisms exist that can explain all of the empirical patterns (e.g. dosage effects and progressive heterosis). Fitness landscape models have been successful at generating many patterns of hybrid fitness, but so far have not dealt with factors such as ploidy, which can affect heterosis outcomes. Here I will present analytical results for a generalised version of Fisher's geometric model that applies to hybrids of any ploidy and between any number of parents. Re-analysing published data from genotypically matched diploid and tetraploid hybrid maize of variable genomic composition (Washburn et al. 2019; Yao et al. 2020), I find that our theoretical model closely matches the patterns observed and sheds light on the mode of segregation and allelic dosage effects in tetraploids.

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Equilibrium allele frequency in terms of invasion and fixation conditions

Genetic variation can be maintained by balancing selection. This requires that an allele ‘invades’ (increases in frequency when rare) but does not ‘fix’ (declines near 100% frequency). Balancing selection pushes these alleles towards an intermediate ‘equilibrium’ frequency. Here, we use a general formulation to show that the equilibrium frequency can often be written as a simple ratio of the conditions required for invasion and fixation. This relationship requires that marginal fitness increases linearly with allele frequency, a common assumption in population genetic models. This expression can be used in models with features of biological interest including sex-specific selection, meiotic drive, imprinting, selfing, parthenogenesis, stage-structure, and spatial structure. We therefore connect many models and provide a concise and interpretable expression for the equilibrium allele frequency under balancing selection.

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A P-element invasion in experimental populations sheds light on the silencing of a newly invading transposon

To study how silencing against a newly invading transposable element is established, we introduced the P-element, one of the most widely studied eukaryotic transposons, into the naive lines of *D. erecta*. We monitored the P-element invasion for 50 generations in 3 replicates by sequencing the genomic DNA, the small RNAs and the transcriptome at regular intervals. In two out of three replicates, the spread of the P-element was curbed by the piRNA pathway within 20 generations. siRNAs against the P-element were observed before piRNAs suggesting that siRNAs may be responsible for establishing the initial response against a newly invading TE. The emergence of piRNAs did not result in lower P-element expression levels but prevented the splicing of a P-element intron. Interestingly the spread of the P-element could not be stopped in one replicate, despite the presence of P-element insertions in piRNA clusters and siRNAs against the P-element. Although the piRNA pathway was functional in this replicate, the ping-pong cycle failed to respond to the P-element invasion, resulting in the absence of sense piRNAs that bind to Ago3. Based on our observation, we propose a novel model for how transposons are controlled in *Drosophila*.

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How much power do we have to detect recurrent selective sweeps?

The detection of selective sweeps from population genomic data often relies on the premise that the beneficial mutation has fixed very near the time of sampling. As it has been previously shown that the power to detect a selective sweep is dependent on the time since fixation as well as the strength of selection, it is naturally the case that strong and recent sweeps leave the strongest genomic signatures. However, the biological reality is that beneficial mutations enter populations at a rate, a rate which will thus determine the mean wait time between sweep events. An important question thus remains of whether we have power to detect recurrent selective sweeps when they are modeled as part of a distribution of fitness effects (DFE) - as they of course are - rather than as a single isolated event as is commonly modeled. Here we use simulations to understand how much power there is to detect recurrent selective sweeps using the SweepFinder composite likelihood method and the H12 haplotype statistic. We ran forward-in-time simulations in SLiM using parameterisations from the Drosophila melanogaster literature, simulating under a range of beneficial selection coefficients, with beneficial mutations incorporated into a full DFE. We also simulated under a variety of population size change models, incorporating recombination and mutation rate heterogeneity, in order to explore a biologically relevant parameter space. Our results suggest a need for great caution when interpreting such selection scans.

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Hyper-divergent haplotypes punctuate the genome of the model parasitic nematode *Heligmosomoides bakeri*

Nematodes are the most abundant animal on our planet and many have evolved to parasitise other organisms, including humans. Despite a long history of studying host genetics within the context of host-parasite interactions, parasite genetics are often neglected. Recent studies have shown that the genomes of free-living nematodes contain ancient, hyper-divergent haplotypes containing genes associated with environmental response. Although similar haplotypes likely exist in parasite populations, sequencing their genomes has previously been hindered by their small size and high genetic diversity, which prevents sequencing pools of individuals. Here, we apply a novel low-input sequencing approach to sequence the genomes of several individual males of two parasitic nematode species: *Heligmosomoides bakeri*, a parasite of domestic mice and a widely used laboratory model for gastrointestinal nematode infection, and *Heligmosomoides polygyrus*, a related parasite of wood mice. Despite a long-standing debate over whether these two taxa belong to a single species, we show that their genomes show levels of divergence consistent with millions of years of independent evolution. Consistent with findings in free-living nematodes, we show that the genomes of both species, including the purportedly inbred *H. bakeri*, contain hyper-divergent haplotypes. Importantly, we find that these haplotypes often contain genes that are known to interact with the host immune system, including a homolog of H11, a secreted antigen that has been proposed as a vaccine candidate against gastrointestinal parasites of sheep.

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Limited dispersal speeds up genetic adaptation via non-additive mutations

The probability and the expected time for new beneficial alleles to fix via selective sweeps are two relevant quantities in population genetics. Such sweeps may be hard - where the fixing allele originates from a new mutation - or soft, in which the fixing allele comes from the species' pool of standing variation. The mean time taken by these different sweeps is well understood in well-mixed populations where individuals interact and compete randomly. Many natural populations, however, are subdivided and dispersal-limited. While such dispersal limitation is known to influence the probability that non-additive beneficial alleles will fix, its effect on the time taken by such fixation remains understudied. Here we show how limited dispersal and population subdivision affect the time scale of adaptation via hard and soft sweeps. We find that dispersal limitation always increases the time taken by an additive allele to sweep. In contrast, for sweeps of non-additive alleles, we find non-monotonic effects of dispersal limitation on the rate of adaptation: it decreases the time an allele takes to fix as dispersal is initially limited, but rises the time of sweeps if it is further limited past below a dispersal threshold. Regarding soft sweeps, we find that adaptation of recessive alleles no longer benefits from dispersal limitation. Overall, adaptation is faster under realistic dispersal rates when mutations have non-additive fitness effects. This work puts forward our understanding of the pace of genetic adaptation under the influence of limited dispersal, population structure and genetic dominance - in particular for non-additive alleles.

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Local adaptation to hosts and parasitoids shape *Hamiltonella defensa* genotypes across aphid species

Facultative symbionts are common in insects and can provide their hosts with significant adaptations. Yet we still have a limited understanding of what shapes their distributions, such as why particular symbiont strains are common in some host species yet absent in others. To address this question, we genotyped the defensive symbiont *Hamiltonella defensa* in 26 aphid species that commonly carry this microbe. We found that Hamiltonella strains were strongly associated with specific aphid species and that strains found in one host species rarely occurred in others. To explain these associations, we reciprocally transferred the Hamiltonella strains of three aphid species, *Acyrthosiphon pisum*, *Macrosiphoniella artemisiae* and *Macrosiphum euphorbiae*, and assessed the impact of Hamiltonella strain on: the stability of the symbiosis, aphid fecundity and parasitoid resistance. We demonstrate that the Hamiltonella strains found in nature are locally adapted to specific aphid hosts, and their ecology: aphids tend to carry Hamiltonella strains that are efficiently transmitted to their offspring, non-lethal, and that provide strong protection against their dominant parasitoid species. Our results suggest that facultative symbiont distributions are shaped by selection from natural enemies, and the host itself, resulting in locally adapted symbioses that provide significant benefits against prevailing natural enemies.

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No doubing the Houting: Genomic insights into the evolutionary and demographic history of whitefish (Coregonus spp.) populations in Denmark

Integrating genomic data is frequently highlighted as a way of improving insight into the evolutionary dynamics of threatened populations, yet critics are voicing skepticism and calling for case studies of the application of conservation genomics. We demonstrate how the use of genomic data sheds light on the intricate demographic and evolutionary history of the morphologically diverse European whitefish species complex (*Coregonus lavaretus*, ELW), including the North Sea houting (*Coregonus oxyrhynchus*, NSH), which by some has been considered a separate species due to pronounced morphological divergence and ability to tolerate oceanic salinities. Through analysis of dense ddRAD sequencing and WGS data, we show that the North Sea houting is significantly differentiated from other Danish whitefish populations. Analyses of demographic history support a previous hypothesis of rapid population expansion following whitefish colonization of Denmark after the last ice age but contradicts estimates of a very recent divergence time between NSH and ELW. Using fastsimcoal, assuming a standard mutation rate and a generation time of 3.5 years, NSH was estimated to have diverged from ELW at least ~13.500 years ago. There was no evidence of contemporary gene flow between NSH and ELW. Genome scans reveal signs of local adaptation in NSH and significant enrichment for gene ontology terms related to salinity tolerance, which was not observed in ELW populations. Additionally, analysis of runs of homozygosity (ROH) show that, despite low effective population sizes, Danish populations of whitefish and houting exhibit only minor inbreeding. Our results suggest that the NSH is more evolutionary divergent from ELW than previously thought. While its species status may be questioned, the increased power and resolution of genomic data confirms that the North Sea houting is an evolutionary unique form which merits conservation as an independent unit.

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Phylogenomics suggests a single origin of terrestriality in Isopods

Isopoda are a large and morphologically diverse order within Crustacea, comprising ~10,000 species across 11 suborders, the most speciose of which is Oniscidea, the group containing woodlice and sea-slaters (>4000 species). Adapted to almost every environment on earth, isopods occupy a wide range of habitats; marine Isopoda are found across different depths - from the benthic zone to the deep sea, amphibious littoral isopods occupy the shores and coastal regions, freshwater aquatic isopods can be found in both open and subterranean water sources and while terrestrial isopods predominantly inhabit moist, damp environments, there are some that are adapted to live at high altitudes or even in the most arid deserts. But despite being a well-recognised, globally distributed and widely studied order, the evolutionary relationships within Isopoda are still not yet well resolved. Traditional morphological studies propose very different relationships between the different isopod suborders, and recent molecular studies (based predominantly on nuclear ribosomal data) have called into question the monophyly of some of the most accepted suborders, including the Cymothoidea (fish parasites) and Oniscidea (terrestrial isopods). The latter instead suggests a closer relationship between littoral and marine isopods and implies that isopods may have adapted to life on land multiple times in their evolutionary history. The analysis presented here draws together all available transcriptomic and genomic datasets across the order and, generating a phylogeny based on approx. 1000 arthropod BUSCOs, produces a surprising result - that the terrestrial isopods may actually be monophyletic, and relationships based on nuclear ribosomal data may be the result of bias due to long branch attraction.

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Birds demography inference based on genomic data

The quantification of demographic change is a common, yet tricky question in population genetics. Here, techniques based on genetic information (allele frequency spectra) are experienced on a diverse set of bird species, which present different types of demographic structure, history (decline, steady...) at different time scales... Some progress on species using genetic data will be presented, including based on the genome of the green woodpecker, *Picus viridis*, that we assembled recently.

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Recombination in the barn owl

Recombination and mutation are the fuel of evolution. Because recombination rates modify the efficacy of selection and shape the diversity landscape, understanding what drives variation in recombination is an important step in understanding how evolution proceeds. Broad scale recombination rates are governed by the number and length of chromosomes available, with smaller chromosomes having higher recombination rates per base pair. In turn, fine scale estimates vary in a way that depends on the presence or absence of the PRDM9 gene. Birds are an excellent setting to study recombination because of the presence of chromosomes of different sizes and the absence of PRDM9. However, most studies have focused on members of the Passerine order which boasts a conserved karyotype limiting broad scale comparisons of recombination. Here we present the first recombination landscape of an owl species, using both linkage mapping and linkage disequilibrium methods. Despite having a larger chromosome number and smaller chromosome size than Passerines, barn owls do not exhibit increased recombination rates but rather a more evenly distributed recombination landscape along the sequence. Our results suggest that the interplay of chromosome number and size is not always a good predictor of broad scale recombination rates.

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Selective sweeps in *Drosophila serrata*

Understanding the extent to which microevolutionary adaptation relies on novel beneficial mutations, as opposed to previously neutral standing genetic variation, is an important goal of evolutionary genetics. Progress towards this goal has been enhanced during the genomic era through the study of selective sweeps. Selective sweeps fall into two categories: hard sweeps via new mutations and soft sweeps via pre-existing mutations. However, data are currently lacking on the relative frequency of these two types of selective sweep. In this study, we examined 110 whole genome sequences from *Drosophila serrata* sampled from eastern Australia and searched for hard and soft sweeps using a deep learning algorithm (partialS/HIC). Analyses revealed that approximately 12.2% of the *D. serrata* genome was directly impacted by sweeps (11.2% soft sweeps vs. 1.0% hard sweeps), and completed sweeps were ~6-times more frequent than partial sweeps. Over half of the genome was indirectly impacted by positive selection in being linked to sweeps. Gene ontology enrichment analysis further supported our confidence in the accuracy of sweep detection as several traits expected to be under frequent selection due to evolutionary arms races (e.g. immunity and sperm competition) were detected. Within sweep regions and those flanking them, there was an over-representation of SNPs with predicted deleterious effects, suggesting positive selection drags deleterious variants to higher frequency due to their linkage with beneficial loci. This study provides insight into the direct and indirect contributions of positive selection in shaping genomic variation in natural populations.

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What can we learn about the history of divergence from the fitness of hybrids?

Reproductive isolation can evolve as a by-product of genomic divergence, but it is less clear whether different modes of divergence will lead to different kinds or strengths of isolation. Models based on Dobzhansky-Muller incompatibilities or holely landscapes, imply that hybrid fitness should be largely unaffected by the mode of divergence. By contrast, methods focussing on individual traits, such as the QTL sign test, are premised on different modes of divergence leading to differences in the types of factors fixed. Analyses using Fisher's geometric model can reconcile these two pictures. Results suggest that the fitness of hybrids is determined by simple measures of the similarity of the changes at different loci, which do vary systematically with the divergence history. Results also show how, under directional selection, the dominance and additive effects contain complementary information about the history.

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Repeated genomic adaptation to climate and its drivers across >300 million years of plant evolution

Repeatability in the genetic basis of adaptation to environment is a phenomenon that has been documented across the tree of life. Repeatability is commonly observed as re-use of shared or introgressed pre-adapted alleles (common within species), or through independent modification of orthologous genes (more likely across species). The latter is less well characterised, yet poses interesting questions for what genes may be critically important for adaptation and why. Here, we present a comparative analysis of the repeatability of genotype-environment associations (GEA) from 29 previously-published datasets from 25 species spanning the plant kingdom. We identify groups of orthologous genes that show signatures of repeated association to a panel of climate variables (N = 21), and are most likely due to independent evolution as opposed to allele re-use. Within these orthogroups are a large number of genes that appear to drive adaptation in a large number of species, including known candidate loci involved in phenology, flowering traits, and auxin signaling. We also demonstrate that repeated adaptation to drought and changing temperatures are associated with a higher order of functional repeatability, with significant enrichments of interactions among genes with evidence of repeatability across different species. Finally, we explore the properties of genes exhibiting signatures of repeated adaptation across any climate variable using various Arabidopsis databases. Here we show that repeatability is associated with multiple forms of pleiotropy. Explicitly, genes from repeatable orthogroups tend to be more broadly expressed across tissue types and exhibit evidence of co-expression with more genes within co-expression networks. In conclusion, we demonstrate repeatable genetic adaptation driven by independent evolution across the plant kingdom in response to climatic selection, driven disproportionately by genes with elevated pleiotropy and likely greater effect sizes.

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Autosomal editors for efficient genetic biocontrol

The release of sterile males to control pest populations is the most widely used genetic method for population suppression to date. However, this approach requires repeat releases of high numbers of individuals to achieve significant suppression, which may be impractical for many species. This inefficiency arises because each generation the load-inducing property of the males is lost and therefore new males need to be released to maintain impact. A possible approach to improve efficiency is to release individuals carrying load-inducing genetic elements which persist for more than one generation. Using a deterministic, discrete generation computer simulator we explore one such method: the release of individuals carrying autosomal editors which can induce load by creating mutations at functional loci elsewhere in the genome. We compare the efficiency, localizability and temporal dynamics of a variety of designs which differ in their fitness effects and evaluate them against some previously proposed genetic strategies. We find that some autosomal editor designs can be more efficient than releasing individuals carrying dominant mutations directly and that efficiency can be further improved by increasing the number of target sites and releasing the editor alongside a second construct which allows for temporary drive of the first.

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Dose-, tissue- and pesticide-specific effects of insecticides on gene expression in bumblebees

Social bees are important insect pollinators of wildflowers and crops, yet their populations are declining because of intensified agriculture and the use of insecticides. Multiple studies reported that neurotoxic insecticides in fact have various sub-lethal effects that hinder pollinator survival. Such findings show that methods used in acute toxicity tests are insufficient to determine the whole spectrum of negative effects of insecticides, and have repeatedly led to restrictions in insecticide usage, challenging the workflows of insecticide manufacturers and regulators. We suggest that the sub-lethal effects of insecticides may be easier to understand if their impacts on pollinator health is measured in a high-resolution manner. Here, we exposed *Bombus terrestris* bumblebees to acute and chronic doses of three insecticides: Clothianidin, Acetamiprid, and Sulfoxaflor. We find major differences in the effects of acute and chronic exposure to the same insecticide. We also find major differences in the effects of insecticides in brains, Malpighian tubules, and leg muscles. We discuss the implications of the specific findings on our understanding of how insecticides act and why their effects could have been misevaluated. Our new findings demonstrate the power of high-resolution molecular methods for disentangling the unintended effects of insecticides. Such 'toxicogenomic' approaches, which are widely used in assessments of drugs for humans, are a powerful tool that can accurately assess the effects of insecticides on non-target species, improve insecticide safety assessments, and inform regulatory efforts.

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Storing and analysing ARGs using tskit

Ancestral Recombination Graphs, or ARGs, capture the full genetic genealogy of a set of sampled genomes, and are thus considered fundamental in population genetics. A number of software tools can now be used for simulation or even to infer ARG-like structures from genetic variation data. However varying definitions of the term ‘ARG’ is leading to terminological confusion. We argue that the ‘succinct tree sequence’ data structure, as implemented in the tskit library, can act as a unifying format to store all of these ARG-like structures. In this format, the edges of the graph are annotated with regions of transmitted genome, which allows annotations to be restricted to regions relevant to the samples. Such ‘sample-resolved’ ARGs allow local trees along the genome to be extracted very efficiently, enabling rapid, scalable analysis. Moreover, a focus on storing ancestral *genomes* rather than ancestral *events* allows tskit to store ARGs without needing to specify exactly how and when recombination events took place. This allows precise representation of the knowable features of an ARG while still maintaining tight correlation between local trees: an important feature of recombining genomes. We discuss how tree sequences can be used to represent the different outputs of a selection of modern inference tools, and illustrate the scalability of our approach using genealogical graphs comprised of millions of genomes.

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The hidden Merian Element of Lepidoptera disguised by a conserved karyotype

Lepidoptera, the butterflies and moths, is a diverse order of insects with a largely conserved karyotype of 31 chromosomes, suggesting that the last common ancestor of the group also had 31. Against this background of karyotypic stasis, a subset of species display dramatic variation in patterns of genome and chromosome organisation. Recently, high-quality, chromosomal genome assemblies have been generated for over 200 species of Lepidoptera, enabling us to ask fundamental questions surrounding the processes that shape genome organisation in Lepidoptera. We used phylogenetically-aware analysis of conserved single-copy orthologs on chromosomes from 210 Lepidoptera and 4 Trichoptera (caddisflies; outgroup) to predict ancestral linkage group number and content across Lepidoptera. We find 31 ancestral linkage groups at the base of Ditrysia, as predicted from early comparisons of *Melitaea cinxia* to other lepidopteran genomes. However, we inferred 32 ancestral linkage groups (also known as 'Merian elements') rather than 31 in the last common ancestor of Lepidoptera, represented in the dataset by the splitting of the early-diverging lineage of Micropterigidae from Ditrysia. We identify a specific fusion of two linkage groups that was present in the last common ancestor of sampled Ditrysia. These two linkage groups are also distinct in the Trichoptera genomes. The ditrysian fusion was not suspected previously, as early lepidopteran lineages, such as *Micropterix aruncella* sampled here, have 31 chromosomes. However, *Micropterix aruncella* has a unique sex-autosome fusion. Fusions of autosomes with the Z are common across Lepidoptera. Since the ditrysian fusion, lepidopteran chromosomes have remained remarkably stable with the vast majority of species having undergone none or just one fusion event. However, we identify eight independent lineages that have evaded the typical constraints of genome structure and undergone extensive reorganisation. These independent events allow us to identify two main modes in which genome reorganisation occurs: through numerous fission events; or through complex, fusion and fission events. Together, these analyses demonstrate how chromosomally-contiguous genomes across Lepidoptera offer an unprecedented opportunity to explore the role of selective constraint in the evolution of genome structure.

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Evolution of a young social supergene in fire ants

Evolution How can a new form of social organisation evolve? Many species of *Solenopsis* fire ants have the ancestral single-queen form of social organisation while some species also include a derived form of social organisation with multiple queens in the colony. This dimorphism is determined by two variants of a 'social supergene', where suppressed recombination leads to joint inheritance of alleles of hundreds of genes. To understand how the social chromosome evolved, we collected fire ant colonies from across South America and sequenced the genomes of haploid males carrying alternate versions of the supergene. We applied a coalescent-based approach to build phylogenies from these genomes. We show that the supergene variant responsible for multiple-queen colonies evolved in one species and repeatedly spread to other species through introgressive hybridization. The retention of the supergene variant after introgression suggests that the costs of hybridization are outweighed by the adaptive benefits of the multiple-queen social form and by the green beard behaviour of the variant-carrying workers. Our work highlights how supergene architecture enabled a new social form to evolve in one species and recurrently permeate species boundaries. Our analyses additionally reveal remarkable genome-level characteristics of the young fire ant supergene system.

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Species divergence driven by ecology and mating system in taxonomically complex British *Euphrasia*

Taxonomically complex groups (TCGs) are characterised by populations that are difficult to assign to discrete and unambiguously defined species. A good non-model plant system for investigating the evolutionary factors blurring species boundaries are eyebrights (*Euphrasia*), a genus that has remarkable diversity in ploidy, mating system and ecology. In Great Britain there are 21 species usually found in different ecological niches, which are exceptionally hard to identify on the basis of morphology or DNA barcoding. Here, we test the hypothesis that species boundaries are maintained by narrow regions of the genome likely to underlie adaptive divergence, while the rest of the genome experiences extensive gene flow. To understand geographic genetic structure and the nature of species differences, we applied genotyping-by-sequencing (GBS) and spatially-aware clustering methods to extensive population samples from all British eyebright species. We found: 1) ploidy is not a complete barrier for gene flow, instead, a distinctive genetic cluster which mainly consists of the selfing heathland specialist *E. micrantha* in Northern Scotland, 2) other genetic clusters largely correspond to geographic regions rather than individual species, 3) closely related species showed low species-level differentiation with a few genomic outlier regions. Our results show eyebrights are characterised by extensive homogenising hybridisation, with species differences maintained by few genomic regions. Distinctive taxa, such as *E. micrantha*, might have experienced a different post-glacial colonization history or maintained their distinct identities due to selfing. Overall, our results highlight how ecological factors and mating system may play a crucial role in shaping genomic divergence in this taxonomically complex group.

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Quantifying genome-wide DNA methylation to assess the adaptive capacity of endangered sea turtles in a warming world

Understanding what contributes to the adaptive capacity of endangered species is essential for predicting how they will respond to climate change. For endangered species with long-lived, slowly reproducing life histories, it has been proposed that plasticity underpinned by epigenetic diversity may be particularly relevant for population persistence, as genetic adaptation alone may be too slow to contend with the unprecedented rate of anthropogenic global warming. Sea turtles are such threatened species, due to their ectothermic physiology and temperature-dependent sex determination (TSD) system, with most populations projected to become almost entirely feminised by the century end. Here, we focus on plastic responses of the TSD system in relation to thermal environment, using loggerhead sea turtles (*Caretta caretta*) that nest on the Cabo Verde Archipelago (East Atlantic) as a model system. After producing a chromosomal-scale reference assembly for this species, we set up a split clutch incubation experiment in field conditions with warm and cool treatment groups, then performed whole genome bisulfite sequencing (WGBS) to compare genome-wide DNA methylation patterns between the treatment groups. We will report on regions of interest in relation to their TSD system, which have conservation implications for the management of sea turtle populations in a rapidly warming world.

Posters

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Comparative genomic reveals common diversity and signature of selection in Saudi Arabian indigenous chicken

Indigenous chickens possess unique adaptations to harsh conditions such as high temperatures and cold challenges. The Arabian Peninsula (AP) local chickens are mostly found outdoors, being reared by sheep and camel herders for secondary production purposes. These birds show high resilience to extreme temperatures (hot, cold), typical to the desert environment of the country. Here, we aimed to investigate the genome diversity and to identify candidate genome regions and candidate genes that show strong evidence of positive selection for thermotolerance in fifteen indigenous chicken populations. A total of 156 chickens grouped in 15 populations were investigated: Black AlHufuf KFU (n = 13, Saudi Arabia), Brown AlHufuf KFU (n = 15, Saudi Arabia), Buquyaq village (n = 12, Saudi Arabia), Saudi (n = 5, Saudi Arabia), Fayoumi, (n = 10, Egypt), Omani (n = 21, Oman), Chantecler (n = 10, Canada), Dulug (n = 10, China), Tibetan (n = 5, China), Gafera (n = 10, Ethiopia), Gesses (n = 10, Ethiopia), Hugub (n = 10, Ethiopia), Kido Gesses (n = 10, Ethiopia), Negasi Amba (n = 10, Ethiopia) and Alfa Midir, (n = 10, Ethiopia) were mapped to the chicken reference genome Gallus gallus (GRCg6a). SNPs data were generated following the GATK best practices protocol restricted to bi-allelic sites. After we removed SNPs in linkage disequilibrium (LD) by applying LD pruning on SNPs with PLINK version 1.9, the population structure among all populations was inferred with principal component analysis (PCA) and the estimation of proportion of admixture ancestry. To detect genomic signatures to cold and hot adaptation, we performed FST and Hp anlaysis. ResultsThe results of genetic structure analysis reveal the clustering of all populations with their geographic region of origin. In all populations, 24,906,132 SNPs were detected. The average nucleotide diversity for the 15 populations is about 0.0021. About 24.0% of the bi-allelic SNPs are novel (\ dbSNP release 107 (2022). Among all the studied populations, the highest level of linkage disequilibrium (LD) (~ 0.30) is observed in Chantecler and Fayoumi, at marker pairs distance of 1 kb, while the lowest is recorded in both Omani and Black AlHufuf KFU populations (~ 0.10). The first and second principal components accounted for 18.1% and 15.6% of the variation. PCA 1 clearly separates the Ethiopian populations from the Chinese and Arabian Peninsula populations, while PCA 2 separates Arabian Peninsula populations from the Chantecler, as well as the Ethiopian population from Dulug and Chantecler. Admixture analysis shows evidence of shared ancestry among all Arabian Peninsula indigenous lines.ConclusionAll populations are clearly separated based on their geographic areas of origin. Chicken populations from Ethiopia, China, Canada were included as reference populations for ease of comparison and interpretation of results. Functional analysis of the candidate genes will increase our understanding of the genetic basis of resilience to extreme temperatures.

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Deconvoluting genomes of Plasmodium and Hepatocystis parasites from a co-infected monkey

African monkeys are infected with malaria parasites from two closely related genera, Hepatocystis and Plasmodium. Hepatocystis are transmitted by Culicoides biting midges, while the vectors of Plasmodium are Anopheles mosquitoes; the life cycles of the two parasites also differ. Numerous Hepatocystis species have been found infecting most African monkeys. In contrast, only two Plasmodium species have been found in African monkeys: Plasmodium gonderi and Plasmodium sp. DAJ_2004 (also known as P. mandrilli). P. gonderi has been well characterized, and a genome sequence is available. In contrast, there has been little study of P. sp. DAJ-2004, and only the mitochondrial DNA sequence is known. A dried blood spot was obtained from a crested mona monkey (Cercopithecus pogonias) from Cameroon. PCR amplification using mtDNA (cytb) primers produced a sequence we identified as P. sp. DAJ_2004. To determine the genome sequence of P. sp. DAJ_2004 we first designed primers for selective whole-genome amplification (SWGA), using P. gonderi as the target species, and a genome from Cercopithecus sabaeus as the host, since it represents a close relative to the original host with good genome quality. We then sequenced the whole genome of P. sp. DAJ_2004 using Illumina MiSeq short-reads, and PacBio HiFi reads.Our analyses of the read libraries, initially using BlobTools, revealed that the sequences came from two different species: P. sp. DAJ_2004, as well as a strain of Hepatocystis. We have used a series of bioinformatics analyses to separate the reads of the two species, and performed de novo genome assembly and gene annotation for both, to produce the first draft genome sequence of P. sp. DAJ_2004, as well as a Hepatocystis genome (the second to date). These data can uncover the phylogenetic relationships of these parasites to other Plasmodium and Hepatocystis species, and can be used to address a variety of questions about the evolution of malaria parasites infecting primates.

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Private PAMs for gene drive localization

Gene drives may provide an effective means of malaria intervention through population suppression of malaria vectors in the A. gambiae species complex. CRISPR-based homing drives have proven highly effective in laboratory settings and require a protospacer adjacent motif (PAM) for transfer of the drive between chromosomes. Different Cas endonucleases have different PAM specificities, but the PAM of SpCas9 is NGG, any nucleotide followed by a guanine dinucleotide.Because an NGG PAM site is required for Cas9 cleavage of a locus, private PAM sites have been suggested for drive localization. Previously, using the phase 2 data of Ag1000G,PAMs were identified on the African islands of Mayotte and Bioko, with varying levels of frequency within the population, and resistance outside of the population. This search has been expanded to the phase 3 AG1000G data, including over 3000 whole genome sequences from across Africa, and has been used to isolate the specific PAM sequences that are most private to each A. gambiae sample set. Using these private PAMs identified in this process, in combination with a double-drive system, could allow for localized suppression of malaria vectors.

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The Darwin Tree of Life Consortium

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The Darwin Tree of Life project - Species Prioritisation and Data Access

The Darwin Tree of Life project* (see https://darwintreeoflife.org/) aims to generate reference quality genome sequences for all of the species with which we share these islands. This decadal project needs to work closely with stakeholders in biodiversity assessment, conservation, ecological genetics, evolutionary genomics, biotechnology and wider community to ensure that we sequence the right species first. Our current species selection criteria are based around phylogenetic disparity (we aim to prioritise sequencing of at least one representative from each of the 4177 taxonomic families represented on and around our shared islands) and around the three Is: interesting, important, iconic. Species nominated because of these three Is contribute strongly to demonstration of the utility of the data by engaging with live research and analysis projects. For the PopGroup community we are keen to hear of your priorities: what species would you like a reference quality genome sequence for? We will be collecting species nominations, and are especially interested in hearing from you - whether student, postdoc, or PI. We will be able to review with you (live) the current status (collection, sequencing) of all the species on the current DToL priority and long lists, and show how you can access all our data freely from public portals. If we do not have a sample from your target species, we can explore wether you can supply specimens for sequencing. * The DToL consortium includes the Royal Botanic Gardens Kew, the Royal Botanic Gardens Edinburgh, the Natural History Museum London, the Marine Biological Association Plymouth, the Wytham Woods field station of the University of Oxford, the Earlham Institute, the University of Oxford, the University of Edinburgh, the University of Cambridge and the European Bioinformatics institute, with collaborators in University College Dublin, natureScot and CABI among (many) others. DToL is funded by the Wellcome Trust and through in-kind and local funding to collaborators.

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A complex colour polymorphism is associated with a single gene in wood tiger moths

Colour is often used as an aposematic warning signal, with predator learning expected to lead to a single colour pattern within a population. However, there are many puzzling cases where aposematic signals are also polymorphic. The wood tiger moth, Arctia plantaginis, uses bright hindwing colours as a signal of unpalatability. Males have discrete colour morphs which vary in frequency geographically. In Finland, both white and yellow morphs can be found within the same populations, and these colour morphs also differ in behavioural and life-history traits which contribute to the maintenance of this polymorphism. In similar cases, such complex polymorphisms are controlled by supergenes. In contrast, we show that male colour is linked to a duplicated copy of a yellow family gene that is only present in the white morphs. This white-specific copy, which we name valkea, is highly upregulated during wing development, and we use CRISPR/Cas9 to knockout this gene to validate its function in producing white pigment. The yellow family genes have been linked to melanin synthesis and behavioural traits in other insect species. Our results add to only a few examples of seemingly paradoxical and complex polymorphisms which are associated with single genes.

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Low levels of synonymous polymorphism at the start of bacterial genes

It has been previously show that the rate of synonymous substitution is lower at the start of the gene in enteric bacteria, than in the centre of the gene. It is thought that this due to selection against the formation of secondary structure at the start of the gene preventing the binding of the ribosome. But how common is this pattern? To investigate this we have looked at levels of synonymous polymorphism at the start of bacterial genes in more than 50 species, spanning a wide variety of taxa. We find that levels of synonymous diversity are significantly depressed in almost all species and that the scale of the depression is very similar, both in terms of the proportional effect and the √íhalf-life√ì over which diversity increases.

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Role of chromosomal inversions in adaption and speciation in the flat periwinkle Littorina fabalis

Chromosomal inversions are frequently documented for their involvement in adaptation and speciation across various organisms. Inversions inhibit recombination and allow favourable combinations of adaptive alleles to be inherited together, which facilitates divergence. Conversely, inversions are often subject to balancing selection which leads to the maintenance of polymorphisms within populations. Given these opposing evolutionary forces acting on inversions, it is important to study their relative effects at various scales of divergence from ecotypes to species. Inversions are well-characterised in the genus of marine snails, Littorina, especially in L. saxatilis, where these rearrangements are linked to habitat adaptation. We focus on identifying inversions and their origins in the ecotypes of flat periwinkle, L. fabalis, in exposed and sheltered habitats across North Europe, and comparing their patterns of differentiation and divergence to the sister species L. obtusata and the more distant relative L. saxatilis. Through haplotagging, a long-range linked-read method, and Illumina sequencing, genomic variants can be mapped to contiguous reference genomes for these species. The additional linkage information from haplotagging allows for the accurate reconstruction of inversions that is otherwise challenging in short read datasets. We hypothesise that inversions can be used repeatedly in the L. fabalis ecotypes across their range to adapt to similar habitats, as well as in L. obtusata and L. saxatilis. Whether the inversions pre-date species divergences or are passed between hybridising species will provide insight into the role of inversions in promoting speciation and adaptation.

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Genomic Diffusions: structure of a continuous genome under stabilising selection, mutation and sex.

The infinitesimal model aims at explaining the evolution of complex traits by assuming that a very large number of loci interact additively. This means a given genome is characterized by a sequence of genetic values along its chromosomes. Studies typically focus on the law of the sum of these values, generally assumed Gaussian in quantitative genetics, or consider a large but still finite number of loci. Here, we introduce a mathematical object, which we call genomic diffusion, to keep track of the whole genomic structure, and in particular to simulate the genetic value of a chromosomic block which has been submitted to stabilising selection, sex, and mutation. We justify this model as a first-order approximation when stabilising selection is weaker than mutation.

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Estimation of Bayesian p-values via Approximate Bayesian Computations: posterior predictive checks (PPC) for population genetic models applied to somatic evolution in haematopoietic stem cell populations

The population of haematopoietic stem cells (HSCs) resides primarily within the bone marrow and is responsible for the production of all types of blood cells. HSCs also circulate, along with some of their progenitor cell descendants, as a rare fraction of cells in peripheral blood. As an individual ages, the HSC population undergoes somatic evolution (mutation accumulation, selection and random drift). The somatic evolution of the HSC population is of interest as an accessible model system for the study of somatic evolution, and because of its role in haematological diseases (including myelo-proliferative disorders, myelomas and leukemias). We obtained samples from 10 individuals: 2 neonates (who provided cord blood samples), and 8 adults (aged from 29 to 81 years). None of these individuals showed evidence of haematological disease. We sequenced 3579 genomes from single-cell-derived colonies of haematopoietic stem cell/multipotent progenitors (HSC/MPPs) across these 10 donors. For each individual we constructed a single cell phylogeny (using MPBoot), with the identified mutations assigned to tree branches.These single cell phylogenies revealed a precipitous decline in clonal diversity after the age of about 65, as certain clades which appeared early in life begin to attain higher frequencies. The dN/dS analysis of this (and other) mutation data suggests that positive selection is pervasive within HSCs from both young and old. The simplest family of models we considered to be compatible with the dN/dS results was a multi-type birth-death process with clone-specific positive selection. We used a (simulation-based) Approximate Bayesian Computation (ABC) method to fit such a model to the observed single cell phylogenies, and to estimate the rate at which advantageous (driver) mutations enter the HSC population throughout life, and to estimate the distribution of fitness effects.The marginal posterior density for the rate of driver mutations was broadly comparable with the estimate obtained from the dN/dS analysis. Fitness effects were found to be in the range 5-10 \%, with a heavy tail of rare drivers conferring greater selective advantage (s>10 \%). However, some features of the observed single cell phylogenies exhibited evidence of departure from phylogenies simulated under the simple positive selection model. This observation presents us with a challenging problem, because the space of alternative models is vast, and difficult to circumscribe, from our current knowledge about the somatic evolution of stem cell populations (including HSC populations).Various procedures for \emph{model criticism}, including \emph{posterior predictive model checking} (here abbreviated to PCC), have been developed in order to make decisions about the compatibility of the proposed models with the observed data. Here we used a PCC method which is applicable to the output from the ABC method. In this PPC method, a chi-squared discrepancy variable is computed from a vector of summary statistics.In the case of our single cell phylogeny data, the vector of summary statistics was computed from up to 4 (equally spaced) time points on the phylogeny. When only the first 2 or 3 time points were included, we found little evidence that the observed phylogenies were incompatible with the simple positive selection model. However, when all 4 time points were included, we found that many of the observed phylogenies from 5 of the donors became incompatible with the simple positive selection model.The p-value estimates obtained from our PPC method are subject to Monte Carlo error. Therefore, in order to establish the robustness of our conclusions, it was necessary to determine the magnitude of this Monte Carlo error. We constructed approximate confidence intervals for the Bayesian p-values by using a (stratified) bootstrap resampling method, which supported the robustness of our conclusions from the PPC analysis.

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Comparative annotation of Drosophila genomes

Accurate prediction of structural elements (protein-coding genes, non-coding RNA genes, repeat elements) of a genome assembly is the first step for downstream evolutionary analysis. There are two categories of genome annotation methods: ab initio approach, prediction of gene structures (promoters, exon-intron junctions, coding and non-coding regions) using statistical models (such as the Hidden Markov Model) and sequence alignment based methods, which uses sequence alignment of known RNAs, cDNA and proteins to discover transcripts. Annotation pipelines such as BRAKER2, MAKER2, RefSeq, and AUGUSTUS can combine both sources of transcript prediction to generate final annotation. MAKER2, most commonly used pipeline by individual research groups, can make use of gene prediction tools (such as AUGUSTUS, GeneWise, SNAP and GeneMark) to provide a fully automated annotation pipeline. However, the pipeline is not easy to use as it highly relies on the parallel computing paradigm. Also, it does not attempt to make orthology predictions and it does not annotate UTR sequences.In this study we have used the Comparative Annotation Toolkit pipeline to annotate ~250 Drosophila genomes. CAT can combine up to four parameterizations of AUGUSTUS, including Comparative AUGUSTUS, and transMap projections to produce annotation sets on each genome in a whole genomes alignment. It integrates all common methods of genome annotation, such as transcript projection, transcriptome and proteome alignments, simultaneous gene-finding, and single-genome gene identification based on full-length cDNA reads. CAT leverages high-quality reference annotation to project well-curated reference annotation onto the rest of genomes in the alignment.

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Analysing Current Methods in Local Adaptation

Finding local adaptation is a crucial first step in answering many evolutionary biology problems, from researching species divergence to calculating rates of adaptive evolutionary change and conservation management. A common technique, known as Qst-Fst, for identifying selection in quantitative traits compares the variation seen in neutral markers with that shown in the phenotype of interest. The classical Qst-Fst comparison's broad assumptions have necessitated the development of new methods. One example of these assumptions is the expectation of a uniform migration between subpopulations. Assuming uniform gene flow creates a bias in the neutral expectation, which has been shown to increase type I and type II errors. Ovaskainen et al. (2011) described a method for detecting local adaptation in complex population structures, but this method has yet to be thoroughly tested. Here we evaluate how this method performs in simulated populations distributed in different structures and subject to neutral and/or selective forces.

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Explain human handedness: combining kin selection, sex, parental and parent-of-origin effects

The frequency of left-handedness in human populations is low but substantial (10.6%). Previous twin studies have shown human handedness is heritable, but only 6% of the heritable variance could be explained by known genetic factors, raising a question of √ímissing heritability√ì. The maintenance of left-handedness has been suggested to imply negative frequency dependent selection, for example owing to a √ísurprise√ì advantage of left-handers in combat situations when most opponents are more used to fighting against right-handers. In such social interactions, selection acting upon handedness could be mediated by genetic relatedness between social partners√Ç, i.e. kin selection. Here, we develop a kin-selection model to explore the evolutionary forces shaping handedness. Depending on the context of combat, left-handedness can be selfish (within-group combat) or altruistic (between-group combat), we show that (1) higher relatedness favours a lower/higher frequency of left-handedness when within-/between-group combats are common; (2) differential relatedness of males and females to their social partners may favour a sex difference in handedness; (3) differential relatedness of males and females to their parents√ï social partners may induce a parental effects in handedness; (4) differential relatedness of maternal-origin versus paternal-origin genes to their carrier√Øs social partners may generate an intragenomic conflict over handedness; and (5) this may further drive the evolution of parent-of-origin-specific gene effects (√ígenomic imprinting√ì) and associated clinical disorders. These parent-of-origin effects might hold the key to detecting the missing genes for handedness.

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Endogenous HERV-K retroviral insertions in the chimp genome

Endogenous retroviruses (ERVs) are retroviruses which have integrated into the germline of the host genome through their encoded viral reverse transcriptase. Within the genome ERVs can remain active but usually accumulate mutations overtime, becoming non-functional. They are present in almost all vertebrate species, attributing to 8% of the human genome. Human ERVs (HERVs) are found in humans and primates. The HERV-K(HML-2) group are associated with multiple diseases in humans, but their role is not understood. Although this group integrated into the primate genome ~40 million years ago, activity of HERV-K(HML-2) has been observed in humans and gorillas (Gorilla spp.), but not in chimpanzees (Pan troglodytes) and bonobos (Pan paniscus). Considering Pan is more closely related to humans than gorillas, it would be expected for HERV-K(HML-2) activity to be similar across these species. This study identified HERV-K(HML-2) insertions in reference genome assemblies of Pan species. This study forms a basis for identifying species-specific insertions and for investigating the level of HERV-K(HML-2) activity in Pan compared to other primate genomes. This could aid in further understanding the evolution and extent of HERV-K(HML-2) activity, and possibly serve as a model for HERV-K(HML-2) pathogenicity.

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Detection and Quantification of Retroviruses in the Mountain Chicken Frog (Leptodactylus fallax)

Across three captive collections of the critically endangered mountain chicken frog (Leptodactylus fallax) in Great Britain, intestinal tumours (adenocarcinomas) have been detected as one of the most frequent causes of ill health and death. Through next-generation sequencing of the tumour tissues, viral avian myelocytomatosis oncogenes were identified from two genera of retroviruses, an alpha- and epsilon-retrovirus. As retroviral infections have been associated with the development of intestinal tumours in vertebrates, the aim of this study was to investigate the relationship of retroviral presence with the intestinal tumours in mountain chicken frogs. By screening various tissues with reverse transcriptase (RT) PCR, here we show the ubiquitous detection of alpha- and epsilon-retroviral RNA in 98% (47 of 48) and 95% (39 of 41) mountain chicken frog individuals respectively, of which 15 of 48 had tumours. This was supported by quantitative RT-PCR for alpharetrovirus, which showed no significant difference in viral load between those with tumours and those without, or between different populations (wild-caught and captive-bred). Through further RT-PCR screening, alpharetroviral RNA was also detected in two of five additional amphibian species (Trachycephalus resinifictrix and Bufo bufo), indicating that alpharetrovirus are not restricted to the mountain chicken frog. To date, the majority of endogenous retroviruses (ERVs) identified in amphibian genomes are of the epsilonretrovirus genera, whereas ERVs of the alpharetrovirus genera are primarily found in mammals and birds. Although not conclusive of whether the alpharetrovirus is exogenous or endogenous, this is the first report of alpharetrovirus RNA detection in Anuran tissue samples. There is limited research into amphibian ERVs, including their potential role in disease conditions, therefore further investigation into the alpha- and epsilon-retrovirus will add to the understanding of ERVs in vertebrates. Additionally, given their critically endangered status in the wild, elucidating on tumour aetiology in captive mountain chicken frogs is vital for their ex-situ conservation.

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Genetics projects in the Centre for Forest Protection

This poster will summarise the current genetics focused projects in the work of the Centre for Forest Protection - a joint venture between the Royal Botanical Gardens, Kew and Forest Research

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Coalescent effective population size: between the stepping stone and the island model

The effective population size Ne is often regarded as the size of an idealized population, which undergoes the same effects of drift or inbreeding as the regarded one. In the field of coalescent theory, it is equivalent to the expected coalescence time of a random lineage pair in this population. My focus is on the coalescent effective population size in the continuum between the one-dimensional circular stepping stone model and the island model. I show how the coalescent effective population size changes when a certain fraction of migration goes to the neighbouring demes and the rest of the migrants disperses globally. I explore this concerning the percentage of local/global migration for different migration rates and numbers of demes in the population (while keeping the total population size constant). The formula for the effective population size of a more general migration model is used to describe the behavior of the effective size in the continuum. To represent the expected coalescence time of the stepping stone and island model, I use mathematical formulas as well as the population genetics simulator msprime.

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Comparative genomics of programmed DNA elimination in Oscheius nematodes

Programmed DNA elimination refers to the developmentally-regulated removal of DNA from somatic cells. This process means that the genome of the soma differs from that of the germline. This phenomenon was first observed in a parasitic ascaridid nematode and has since then been found sparsely across distantly related eukaryotes. We previously found programmed DNA elimination in Oscheius tipulae, a free living nematode closely related to Caenorhabditis elegans. In O. tipulae, germline-restricted DNA is eliminated from each end of every chromosome. New telomeres are added at the break sites. To better understand this process, we have assembled the genomes of four additional Oscheius species. All species eliminate DNA. While all species eliminate DNA from the chromosome ends, as in O. tipulae, some also eliminate DNA from chromosome-internal sites. These internal breaks are healed by the addition of neo-telomeres, and result in different chromosome numbers in somatic and germline cells. Some sites of cleavage correlate with chromosomal structural variation between species. Our work sheds light on a widespread yet understudied feature of eukaryotic genome biology and suggests routes to elucidate a mechanism of programmed DNA elimination.

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Can Biased X-Chromosome Inactivation Be Explained By Sexual Antagonism?

During mammalian development, one of the two X chromosomes in each cell of a female is rendered transcriptionally silent through a process called X-chromosome inactivation (XCI). Though this typically results in an even split between cells expressing the maternal or paternal X chromosome, XCI can also be biased, with some X chromosomes showing a heritable tendency to avoid becoming the inactive X. Using a two-gene, two-allele model, we explore a hypothesis that sexual antagonism can drive the evolution of biased XCI. The first gene, a modifier that influences the choice of which X to inactivate, is linked to a second, sexually antagonistic gene, which starts at its polymorphic equilibrium. We show that a modifier allele that causes its chromosome to avoid inactivation can invade the population and become linked to the female-beneficial allele. We also show that after invading and spreading, the modifier allele can, under some conditions, lead to reduced female fitness. In addition, we examine how a direct cost of the modifier allele, which results from an effective reduction in heterozygosity in females, may influence its ability to invade a population. Finally, we explore the success of the modifier allele in a finite population, and show that the √ídrift barrier√ì to its invasion may be too high. Thus, although the modifier can invade under idealized conditions, in actuality it is unlikely to do so in any natural population.

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Title

It is currently unknown why additive genetic variance (VA) varies strongly between different traits. Under mutation-selection balance, VA variation between traits is expected if the mutational input or the strength of (stabilising) selection varies between these traits. However, it is currently unknown whether variation in mutation or selection contributes most to differences in VA among traits. Using RNA-seq data from natural isolates and mutation accumulation lines of Escherichia coli, I will assess the degree to which variation in VA across several thousand gene expression traits can be explained by the strength of selective pressure acting on their expression or by the rate at which their expression accumulates mutational variance. The strength of selection on these traits cannot be measured directly, and so will be inferred by looking at the nucleotide composition of the confirmed promoter regions of all E.coli genes. Promoter regions were chosen as they have been shown to be an important determinant of variation in gene expression and because they are uniquely associated with a particular gene. Therefore, their sequence evolution is primarily dictated by selection acting directly on the expression of that gene. Following the methods by Thorpe et al., I will compare the promoter sequences of closely and distantly related E.coli species. By counting the number of singletons (mutations appearing in only one strain) and SNPs (mutations appearing in multiple strains), selection strength can be estimated. This information will then be included in a double-hierarchical generalised linear model which will assess the relative importance of mutation and selection in shaping additive genetic variation between gene expression traits.

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New Species from Old Genes?

Introgression is the transfer of genetic material between species through hybridisation and has been identified as a potential driver of adaptation to new environments. Species in the plant genus Antirrhinum (Snapdragons) have been split into three morphological sections, relating to their ecology: Antirrhinum, Streptosepalum and Kickxiella. Phylogenetic analyses indicate there has been a second independent evolution of Kickxiella morphology from within section Antirrhinum. One hypothesis for this is that loci responsible for the Kickxiella morphology were introgressed into a section Antirrhinum genetic background. Analyses using SNPs from short read sequence data support the occurrence of introgression, but further investigation did not find any substantial regions in the genome with signal of introgression. One explanation for this may be that these introgression events were ancient and the introgressed regions have been broken down in the genome over time.

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Using biological invasions to understand rapid adaptation to new environments: a genomic reconstruction of the house sparrow global spread

The well-known house sparrow (Passer domesticus) is a small bird renowned for its charismatic nature and close association to humans. House sparrows can be found living and breeding on almost every continent in the world, almost exclusively in near proximity to human habitation. The house sparrow is thought to have spread across Eurasia from the Middle and Near East alongside the spread of agriculture and the establishment of fixed settlements during the Neolithic era. However, due to multiple intentional and accidental introductions from the mid-19th century onwards, the species is also invasive in Australasia, Southern Africa, and the Americas. In modern North America, the house sparrow now exists from Southern Panama all the way to the Northwest territories of Canada, attesting to the species√ï ability to survive in a diverse range of environments. This research aims to understand evolutionary success in biological invasion by reconstructing the history of the house sparrow invasive spread. Using whole genome resequencing, we generate further understanding of the genomic structure within the invasive range. Our results reconstruct the house sparrow invasion history to identify demographic changes between populations. We also studied parallel latitudinal clines within the United States and Australia to demonstrate the parallel evolution of new traits from bottlenecked founder populations, suggesting that certain genome regions and traits may be more beneficial for evolutionary success. Invasive species are an excellent model for understanding complex evolutionary processes, and this research will inform future work on understanding the genomics of successful adaptation to new environments.

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High recombination rate affects demography inference

Reconstructing population history is an essential step to study initial stages of the speciation continuum. Mutation and recombination are important parameters in demography inference. For instance, in methods related to ancestral recombination graphs (ARGs), distribution of mutations is used to infer local trees, and historical recombination events are effectively represented by transition of marginal trees along the sequence. Although ratio between recombination and mutation rates varies across species, as well as along a genome, the effect of this variation on demography inference have not been well studied. Here, using population genomic simulations, we investigate the effect of recombination/mutation ratio on two ARG-related demography inference methods: multiple sequentially Markovian coalescent (MSMC2) and Relate. When average recombination rate is smaller than or equal to mutation rate, historical effective population size and split time were inferred well. However, under higher average recombination rate than mutation rate, inference of both effective size and split time were deviated from the truth. This effect of recombination rate on demography inference can be resolved by masking high-recombining genomic regions. Our results suggest that demography inference using ARG-related methods should be carried out with caution when applied in species whose reference genomes contain high-recombining regions.

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Aneuploidies are an ancestral feature in trypanosomatids, where an ancestral chromosome duplication was maintained across several clades

Aneuploidy, the presence of an aberrant number of chromosomes in a cell, usually results in severe abnormalities in multicellular eukaryotes as humans. However, some unicellular eukaryotes rely on aneuploidy as a mechanism to allow rapid adaptation to changing environments, having a positive fitness in stress conditions and promoting drug resistance. Aneuploidies have been largely described in protozoan parasites as Leishmania and Trypanosoma cruzi, where duplicated chromosomes vary in different hosts and can promote drug resistance. Interestingly, their closely related parasite Trypanosoma brucei, is mainly euploid. Hence, to evaluate if aneuploidies are an ancestral or recent feature in trypanosomatids we estimated the chromosome copy number variation in several Trypanosomatidae species, including Angomonas, Crithidia, Leptomonas and T. vivax, using whole genome sequencing and read depth coverage variations. Aside from the T. brucei, T. evansi and T. vivax, all the remaining species have evidence of aneuploidies, indicating that it is an ancestral character in these parasites. The presence of aneuploidies could be detrimental in T. brucei clade, as their genome is packed in a lower number of larger chromosomes. Next, we evaluated if there were consistent chromosomal duplications in the evaluated species. Leishmania√ïs chromosome 31 is constantly supernumerary, a fact reassured by our analysis of ~200 isolates from L. donovani and L. infantum populations in Africa, Asia and Brazil. This chromosome had an increased nucleotide diversity (¬ππ), which is expected, if the supernumerary chromosomes maintain a larger long term population size. Similarly, redundant copies of genes could allow a rapid adaptation and diversification without loss of function. Regarding the other trypanosomatid species, the chromosomes that have most of its genes orthologous to Leishmania chromosome 31 were also consistently supernumerary, even in the euploid T. brucei clade where regions of this chromosome are observed in two chromosomes, 4 and 8. We evaluated the function of these shared duplicated genes and we found genes involved in housekeeping functions as osmoregulation and response to stress, diverse cytoskeleton mediated processes such as cell morphogenesis, flagellar motility and cell division, energy obtaining pathways, host immune system evasion, infectivity and intracellular trafficking.

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Is the mutation pattern in bacteria affected by the presence of particular DNA repair enzymes?

The pattern of mutation is determined by a broad variety of factors including the chemical stability of the nucleotides, the mutagens that the DNA is exposed to and a constellation of enzymes that replicate and repair DNA. Perhaps surprisingly the suite of genes involved in DNA replication and repair varies amongst bacteria. Does this affect the pattern of mutation? To investigate this question, we have inferred the pattern of mutation from synonymous polymorphisms at 4-fold degenerate sites from over 300 bacterial species for which also have information on the presence or absence of many DNA repair genes. Initial studies indicate that the presence or absence of some DNA repair genes has an effect on the pattern of mutation.

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Host-parasite phylogeography: a tale of glaciation and local adaptations

In the northern hemisphere, the demography and spatial genetic structure of diverse taxa have been shaped by migration patterns after the last glacial maximum. Animals originating from different glacial refugia differ in their genetic mark-up due to demographic processes but they can also display genetic differences caused by adaptation do local conditions. Despite growing use of genome-wide markers in vertebrates to study phylogeography, similar studies of invertebrates are scarce, in particular when it comes to nematode parasites. Parasite population structure is heavily influenced by demography of their hosts but it is also shaped by other processes such as local co-adaptation with their hosts or past host switches. In the present study, using bank vole Myodes glareolus and its parasites Heligmosomum mixtum and H. glareoli we we aim to reconstruct host and parasites phylogeographic patterns in the context of post-glacial migration patterns, and to identify loci differentiating the studied populations.

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Understanding the origin of biodiversity of Oreochromis Alcolapia

Adaptive radiations give us an insight into the processes that generate biodiversity and can be initiated by hybridisation events. Following interspecific breeding, advantageous alleles can be positively selected for and integrated back into conspecific genomes. Alternatively, hybrid speciation can occur in which a hybrid species becomes reproductively isolated from conspecific species, although this is more common following polyploid events. One such adaptive radiation is the Alcolapia species flock native two soda lakes in East Africa and this clade has undergone rapid diversification driven by ecological partitioning and sexual selection. Alcolapia is a relatively young clade, estimated to be less than 10,000 years old and three of its species are known to readily hybridise in nature. It has been observed that at a site on the eastern margins of the lake, fish have unusual phenotypes that are intermediate of both A. latilabris and A. alcalica. These hybrid fish have unusually large lips compared to A. alcalcia and the position of their mouth is less inferior than expected of A. latilabris. Previous work has studied the morphometrics of these hybrids showing that they cluster in between A. latilabris and A. alcalica found at other sites on PCA space. However, the genetic basis of this population at the hybrid site is unknown and remains to be investigated. I would very much like to present the results of analysis from RAD sequencing data about the origin of naturally occurring novel phenotypes via a poster at the PopGroup meeting. I hope to have a PCA figure and a STRUCTURE plot of my findings so far.

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Genomics of hybridisation in British native flowering plants

Hybridisation can have various evolutionary outcomes, from homogenising the differences between populations to generating phenotypic and genotypic novelty. However, the majority of those important phenomena are evaluated in case studies of evolutionary model species chosen for their notable hybridisation outcomes, and whether those findings represent generalities of natural hybridisation in a wider range of taxa remains unknown. As such, this project aims to explore the general patterns of natural hybridisation in the British flora using genomic approaches. The British flora is a well-studied flora with extensive records of hybrids, rendering it the ideal system to understand natural hybridisation. Specifically, we seek to understand the extent of postglacial introgression in British native flowering plants. To address the question, six conspicuous hybridising species pairs from diverse taxonomic groups have been collected across the UK. The extent of introgression between species will be quantitatively evaluated using demographic modelling based on whole genome data.

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Cryptic hybridization between the ancient lineages of Natterer's bat (Myotis nattereri)

Climatic change and adaptation have a major impact on the spatial distribution and demographic history of taxa and are therefore a major driver of biological diversification. Populations may become geographically separated for longer periods leading to independent evolution among them. Secondary contact zones between populations after periods of allopatric evolution offer rare and promising opportunities for studying processes such as population divergence and the evolution of hybridization barriers in a spatiotemporal context. In this study, we describe the spatial extent of a hybrid zone and quantify past population dynamics between the two genetically distinct but morphologically indistinguishable lineages nattereri and crypticus within the Myotis nattereri species complex. We sequenced 62 bats by whole-genome resequencing throughout the contact zone across Central and southeastern Europe. Using a suite of demographic methods, we demonstrate that the degree of admixture varies largely among individuals along a geographic gradient, suggesting a lack of intrinsic reproductive barriers. Although bats are highly mobile, populations are genetically structured on a narrow geographic scale, indicating the lack of long-distance gene flow.Overall, we demonstrate how a whole-genome view on the spatial distribution of evolutionary lineages can improve substantially our understanding on how biogeography and demography contribute to morphologically cryptic diversification in a well-known European mammal.

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Detecting episodic positive selection in phylogenomic dataset using branch-site model and its statistical properties

Over evolutionary time, diverged species undergo variations in substitution rate at the protein level due to selective pressures. Codon-species alignments are used to understand molecular evolution in protein-coding genes using non-synonymous (dN) to synonymous (dS) substitution rate ratio, __=_dN/dS, where __ > 1 indicates positive selection,___ = 1 as neutral evolution, and __< 1 purifying selection. Using branch-site model, we can estimate __ and observe evolution at codon sites and at specific lineages (episodic) of phylogenies. Branch-site test is based on likelihood ratio test (model comparison using maximum log-likelihoods) between a null model assuming neutral evolution or conserved sites and alternate model assuming positive selection. For multiple protein-coding gene hypotheses (phylogenomic dataset), we can apply branch-site model to observe which class of protein-families undergo positive selection. In this study, I construct a series of realistic simulations to study the statistical power of the model (power of positive selection), especially its effects upon correcting for false discovery rate (FDR). My results shows the importance of FDR-correction especially when there is model misspecification due to higher rate of synonymous sites in phylogenomic datasets.

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The genetic basis of convergence in tropical butterflies

Convergent evolution of mimetic colour patterns is widespread in neotropical lepidoptera due to the trait√ïs vital role in predator defence. Coupled with its role in sexual selection, colour pattern has the potential to drive speciation. However, little is known about the genetic basis of colour pattern across lepidoptera, and the contribution of different evolutionary mechanisms to the repeated convergence of colour pattern is unknown. We constructed reference genomes for two species of ithomiine butterfly, Hypothyris anastasia and Hypothyris semifulva, and sequenced the whole genomes of 56 individuals. Using a genome wide association study (GWAS) and a range of genomic techniques, we compared the genomes of closely related subspecies to identify loci involved in controlling colour pattern. Next, these regions will be compared to similarly patterned species across a range of evolutionary timescales, and searched for signs of introgression.

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Unravelling the genetic architecture of a balanced lethal system in newts

In a balanced lethal system, recessive lethal alleles are present on two homologous chromosomes with each chromosome bearing a different lethal allele. Functionality is retained because lethal alleles on one chromosome are compensated by a functional alleles on the other chromosome. These chromosomes are passed on to the next generation randomly, which results in the non-lethal combination of chromosomes in only half of the offspring, the other half dies. Triturus newts possess a balanced lethal system that is also known as √íchromosome 1 syndrome√ì √ë there are two versions of chromosome 1 √ë 1A and 1B and only individuals heterozygous for chromosome 1 (1A 1B) survive. In order to dissect the genomic architecture of this phenomenon we are first assembling the genome of Triturus newts. One of the biggest challenges in this genome assembly is the genome size √ë Triturus genome is >20Gb and highly repetitive, ~70%. For the genome assembly, we are deploying long-read Nanopore sequencing alongside linkage map and HiC sequencing.

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Investigating polymorphic mimicry in the Neotropical Catfish, Corydoras fulleri.

Mimetic interactions have long fascinated biologists and the evolution of such systems provide powerful evidence for natural selection. M¬üllerian mimicry, where co-mimics are unpalatable, leads to the evolution of a shared warning pattern, due to the shared cost of predator education. Therefore, occurrences of colour polymorphism in M¬üllerian mimics are paradoxical. Corydoras fulleri is a polymorphic Neotropical catfish with three morphs. All three morphs are found in sympatry and form a probable M¬üllerian mimicry ring with two additional sympatric Corydoras sp which resemble two of the polymorphic patterns. Using this rare polymorphic mimetic species, we are investigating the genetic basis of colour pattern mimicry. Using low coverage whole genome resequencing we aim to identify portions of the genome associated with colour pattern polymorphism and investigate the nature of any identified polymorphism, e.g. structural variants, transposable element insertions etc.

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Selective whole-genome amplification reveals population genetics of Leishmania braziliensis from primary patient samples

In Brazil, Leishmania braziliensis is the main causative agent of the neglected tropical disease, cutaneous leishmaniasis (CL). CL can present on a spectrum of disease severity and often is refractory to treatment, yet the parasite factors that may contribute to disease presentation and patient treatment outcome are not well understood, in part because successfully isolating and culturing parasites from patient lesions remains a major challenge, and because adaption to culture has been shown to induce widespread genetic changes in Leishmania. Here we describe the development of selective whole genome amplification (SWGA) for Leishmania and show that this method enables culture-independent analysis of whole parasite genomes obtained directly from primary patient skin samples, all while avoiding artifacts associated with adaption to culture. We show that SWGA can be applied to multiple Leishmania species residing in different host species, suggesting that this method can be broadly useful in both experimental infection models and clinical studies. Finally, we show that parasite genomes generated by SWGA of skin biopsies collected from patients in Corte de Pedra, Bahia, Brazil exhibit substantial genetic diversity and can be integrated with published whole genome data from parasites isolates to expand our understanding of Leishmania population genetics in Brazil.

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Reference panel phasing for low-coverage sequencing imputation

Genetic and phenotypic data of multiple individuals of a given species are required to study the genetic architecture, the consequences of inbreeding or the phenotypic and allelic frequencies variations through time and space. However, and despite the reduced cost of next-generation sequencing, whole-genome sequencing (WGS) of large sample sizes remains expensive. Low-coverage WGS (lcWGS) has been proposed as a solution to this problem through proved robustness in model species sequencing (ie. human, mice). For instance, two loci were associated with major depressive disorder in humans through genome-wide association studies (GWAS) in individuals sequenced at 1.7x. The main lcWGS workflow requires a probabilistic representation of genotypes instead of fixed genotype calls. Because of this and high missing rates, imputation is used to refine lcWGS variant genotype likelihoods and fill any missing gaps between lcWGS reads. Imputation has a wide application such as to identify genomic regions associated with lung cancer risk in African Americans or to assess population dynamics of ancient Maltese populations. Specifically, imputation typically uses a reference panel of densely genotyped haplotypes to predict missing genotypes. It has been observed that having an accurate reference panel leads to substantially higher imputation quality. To obtain a high-accuracy reference panel, individual variants can be assigned to haplotype blocks through phasing, which can be done using different information: Read-based, linking variants on the same sequenced fragment; Family based, by studying the transmission of alleles through generation; Population based, by statistically assessing the link between variants. While family-based phasing has led to more accurate results than read-base phasing with WhatsHap software in humans, the results for other organisms and its further implication in imputation are unknown. Our aim is to assess the output of different types of phasing (read-based, family based with single and multiple trio pedigree phasing, population based) in barn owl European populations (Tyto alba). We will then asses how differently phased reference panels perform to impute lcWGS.I will thus present the reference panel composition, the results from comparing different phasing strategies, as well as the results of our pipeline for variant calling & imputation. On a broader sense, this work will help establish the highest-accuracy lcWGS workflow on a non-model species.

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The ecology of palm genomes: repeat-associated genome size expansion is constrained by aridity

Genome size varies 2400-fold across plants, influencing their evolution through changes in cell size and cell division rates which impact plants√ï environmental stress tolerance. Repetitive element expansion explains much genome size diversity, and the processes structuring repeat √îcommunities√ï are analogous to those structuring ecological communities. However, which environmental stressors influence repeat community dynamics has not yet been examined from an ecological perspective. We measured genome size and leveraged climatic data for 91% of genera within the ecologically diverse palm family (Arecaceae). We then generated genomic repeat profiles for 141 palm species, and analysed repeats using phylogenetically informed linear models to explore relationships between repeat dynamics and environmental factors. We show that palm genome size and repeat √îcommunity√ï composition are best explained by aridity. Specifically,Ty3-gypsy and TIR elements were more abundant in palm species from wetter environments, which generally had larger genomes, suggesting amplification. By contrast,Ty1-copia and LINE elements were more abundant in drier environments. Our results suggest that water stress inhibits repeat expansion through selection on upper genome size limits. However, elements that may associate with stress-response genes (e.g.Ty1-copia) have amplified in arid-adapted palm species. Overall, we provide novel evidence of climate influencing the assembly of repeat √îcommunities√ï

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Genomic phylogeography of Rana iberica

During the Pleistocene, refugial species in the Iberian Peninsula had to contend with extreme climatic cycles in addition to vicariance dividing the large continental range. Iberian endemic species therefore provide an excellent opportunity to investigate the importance of geography vs demography in the development of genetic clusters and diversity hotspots.We expand on previous phylogeographic studies of the Iberian endemic refugial frog species Rana iberica, to investigate the genomic phylogeography of the species from a metapopulation stand-point. Previous authors identified 3 three major mitochondrial DNA lineages, including a highly distinct lineage from the Sierra de Guadalupe. We genotyped a large genomic (RADSeq) data set of 308 individuals from 51 sites comprising the total range of the species. We aimed to see if the Sierra de Guadalupe cluster could be recovered using nuclear DNA data, and if this genetic structuring persisted with more continuous sampling of the region. In addition, we looked for recent gene-flow that can now be detected using fine-scale genomic data.We found that nuclear DNA data was largely concordant with mitochondrial DNA and identified a general 'Isolation By Distance' signal in addition to confirming the presence of the distinct Sierra de Guadalupe cluster. The results here contribute to explaining the high variation in phylogeographic responses observed in refugial species: the determinants of hotspots of diversity depend not only on historic refugia acting as stable 'cores' for the metapopulation, but also on the contact of distinct genetic lineages after isolation.

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Gene Association Studies Reveal Distinct Communities that Exist within Microbial Pangenomes

Gene Association Studies Reveal Distinct Communities that Exist within Microbial PangenomesLahiru Thomas Sooriyabandara1, Alan Beavan1, Fiona Whelan1, James McInerney1 1School of Life Sciences, University of Nottingham, Nottingham, United KingdomUnderstanding molecular evolution in the context of bacterial pangenomes is key to addressing biomedical challenges including the emergence of antimicrobial resistance, pathogenesis and nosocomial infections. Several methods have been developed in recent years in pangenome assembly and analysis that aim to identify gene dependencies and key predictors of microbial phenotypes. It is also important to understand the role of regulatory genes and epigenetic modifiers across the pangenome. Campylobacter, Listeria and Neisseria datasets were compiled from the NCBI database. Only complete genomes were used and all strains and modifiers that pertained to the Campylobacter, Listeria and Neisseria genus were included in the analysis. BUSCO was used for genome quality control (QC) and only genomes that exceeded a threshold of 0.90 were included in pangenome assembly and in subsequent analysis. PROKKA was used for genome annotation and Panaroo was used to assemble the pangenome for Campylobacter (553 genomes), Listeria (320 genomes) and Neisseria (228 genomes). Coinfinder and machine learning (ML) were used as comparative approaches in pangenome analysis across all 3 datasets. Coinfinder identified 5 distinct gene communities in the Listeria dataset compared with 6 that were identified using the machine learning approach and random forests. 5 gene communities were identified by both Coinfinder and machine learning in the Campylobacter dataset. These gene communities that reside within the pangenome all have a distinct function and may confer a particular survival advantage. Genes of interest that were identified by both analyses include: accB, ubiG, yddE. accB has an important role in acetyl-CoA carboxylase activity, lipid metabolism and fatty acid biosynthesis. ubiG is involved in methylation, ubiquinone and cofactor biosynthesis. yddE is an uncharacterised protein that has a role in ATP binding and unidirectional conjugation. These 3 genes are involved in cellular survival and metabolism. There was a clear difference between the visualisation of the networks in each pangenome. An overlap of 15-25% for the Campylobacter, Listeria and Neisseria datasets in terms of pairwise gene associations was observed. The initial results indicate that Coinfinder and ML are complementary approaches rather than comparative. Considerable differences in pairs of gene associations between the 2 approaches with relatively little overlap and a notable difference between the visualisations of the networks suggest that these 2 methods of pangenome analysis may examine different aspects of the pangenome and reveal complementary biological insights. It is also possible that these differences could in part be explained by the fact that the ML approach produces a directed network whereas Coinfinder produces an undirected network. Therefore, the ML approach may produce more precise results in terms of the gene associations reported.

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Multilocus barriers and local adaptation in haplodiplontic populations

Theoretical and empirical research on the population genetics of speciation has largely focused on organisms with a life cycle dominated by either the haploid or diploid phase, although many species, in particular cryptogams, have life cycles where both phases are prominent. Depending on the genetic architecture of local adaptation and reproductive isolation, the different dynamics of selection in biphasic compared to monophasic life cycles are expected to have an effect on the accumulation of barriers to gene flow and the maintenance of local adaptation and reproductive isolation in the presence of migration. In the context of a broader project on natural selection and speciation in bryophytes, we study the establishment of barriers to gene flow and locally beneficial variants in theoretical haploid-diploid population models. We quantify the strength of a multilocus barrier to gene flow and its effect on the maintenance of local adaptation in a metapopulation of haplodiplontic organisms using the device of effective migration rates. Our results indicate that the relative amount of locally selected variants whose fitness effects are biased to either phase, can have a marked impact on the strength of a multilocus barrier to gene flow. We discuss further questions and avenues of future research concerning the evolutionary genetics of speciation in haplodiplontic species in general and bryophytes in particular.