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PRESENTATION ABSTRACTS

THE 54TH POPULATION GENETICS GROUP MEETING

A VIRTUAL CONFERENCE
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PopGroup 54 Abstracts

Talk abstracts

Jan 4th, 2021

Session 1

1. The genetic basis of male colour polymorphism in the wood tiger moth

Presenter: Melanie Brien, University of Jyväskylä
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Colour is often used as an aposematic warning signal. Predator learning is expected to lead to a single colour pattern within a population, but there are many puzzling cases where aposematic signals are also polymorphic. The wood tiger moth, Arctia plantaginis, uses its bright hindwing colours as a signal of their unpalatability. Males have discrete yellow, red or white colour patterns, and frequency of male colour morphs varies geographically. In Finland, both white and yellow morphs can be found and these colour morphs also differ in behavioural and life history traits, such as immunity, chemical defences and flight patterns. Complex polymorphisms such as these are often explained by supergenes, groups of tightly linked alleles. Conversely, we found a simple genetic basis for the male colour polymorphism. Using a QTL analysis of lab crosses, GWAS of wild populations, and RNAseq analysis, we found a single gene in the 'yellow' family genes controlling the switch between white and yellow colour. These 'yellow' 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 controlled with a simple genetic basis.

2. Rapid genomic adaptation in experimental populations of Trinidadian guppies (Poecilia reticulata)

Presenter: Mijke van der Zee, University of Exeter
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Phenotypic evolution can occur more quickly than previously thought but understanding the genetic basis of rapid evolution is still in its infancy. Until recently it was difficult to detect such rapid adaptation at the genomic level. Four replicated experimental translocations of high-predation (HP)guppies to uncolonized low-predation (LP) habitats in northern Trinidad provide a unique opportunity to study this phenomenon. These transplanted guppies have evolved phenotypes similar to naturally-colonised low-predation populations in 8-10 generations. These adapted phenotypes persist in subsequent generations in lab environments, suggesting a prominent role for genetic adaptation. Here, we present results from whole genome scans of each experimental population, sampled 8-10 generations post introduction, and their high-predation source. Patterns of genome-wide variation indicated only minor changes from the source. However, examining runs of homozygosity revealed a bottleneck in one population, which agrees with population census data. Using a combination of haplotype genome scans and a novel multivariate approach based on allele frequency change vectors, we found signals of convergent evolution in all four populations. Specifically, we found a region on chromosome 15 under strong selection in three of the four populations, and our multivariate approach revealed more subtle parallel changes in allele frequency in all four populations across this region. Investigating patterns of genome-wide selection in this uniquely replicated experiment offers a remarkable insight into the mechanisms underlying rapid adaptation that could potentially be extrapolated to other species and populations experiencing rapidly changing environments.

3. Revisiting Wolbachia diversity between invasive and native Rhagoletis cherry fruit flies in the light of comparative genomics

Presenter: Thomas Wolfe, Institute for Forest Entomology, Forest Pathology and Forest Protection (IFFF), Boku, Vienna, Austria
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Wolbachia is a maternally inherited obligate endosymbiont that can induce a wide spectrum of effects in its host ranging from mutualism to reproductive parasitism. At the genomic level, frequent recombination events within and between strains, transposable element activity, and horizontal acquisition of strains between host species have made Wolbachia one of the most dynamic bacterial systems we know. The invasive cherry fruit fly Rhagoletis cingulata recently arrived in Europe from America it now co-occurs with the native cherry pest Rhagoletis cerasi. This shared distribution facilitated interactions between the two fly species and led to a directional horizontal Wolbachia transmission from the native to the invasive fly. Wolbachia diversity in this model was initially described based on a limited number of molecular markers. Similarities between the invasive strain wCin2 and the native strain wCer2, suggested not only a directional transmission from the native to the invasive species but also bi-directional transfers. Here, comparative genomic analysis reveals striking differences between the wCin2 strain in its native range, the wCin2 strain in its invasive range, and wCer2. Therefore we performed a comparative genome study of wCin2 in natural populations of native and invasive R. cingulata and wCer2 in co-occurring R. cerasi flies. We find major structural rearrangements such as inversions and duplications between these genomes as well as differences in prophage content. Also, cytoplasmic incompatibility inducing genes show many differences between wCin2 and wCer2 that provide insights into the selective conditions shaping these Wolbachia strains. This study lays out new perspectives for the genomics of Wolbachia in the Rhagoletis fruit fly system and illustrates the benefits of using whole-genome sequencing to characterize diversity in natural populations of this highly dynamic endosymbiont.

4. Factors affecting the genomic distribution of runs of homozygosity in red deer populations

Presenter: Anna Hewett, University of Edinburgh
Co-Authors: Josephine Pemberton, University of Edinburgh

Inbreeding results in genomic segments that are identical by descent, which appear as runs of homozygosity (ROH) in genomic data. In addition to ROH indicating inbreeding level in a population, the genomic distribution of ROH can reveal information about other causal mechanisms. Previous studies on livestock show multiple factors can affect ROH distributions including recombination, population demography and selection. Here we use two wild populations of red deer, one from the Scottish island of Rum (3046 individuals) and one from the Scottish mainland (157 individuals) to search for ROH >2.5Mb across 35,132 autosomal SNPs. ROH were searched for using both physical and genetic marker map positions to investigate the effect of recombination on ROH calling. Within-population differences using these methods shows that ROH distribution is more uniform using genetic map position, highlighting the role of recombination. Additionally, when using physical map position, between-population patterns are similar and more correlated, suggesting consistent recombination rates between populations. Following this, using genetic map positions we found 5 ROH hotspots (where ROH were unusually common in the population) in Rum deer, with >15% of the population having a ROH at the same location. As these hotspots persist after accounting for recombination we suggest they may mark regions of positive selection. Consistent with this, haplotype diversity in ROH hotspots was greatly reduced in comparison to the rest of the genome. In conclusion, we show the key role of recombination in the distribution of ROH and accounting for this by using the genetic map enables hotspots to be more clearly identified.

5. Life tables drive genetic diversity in marine fishes

Presenter: Pierre Barry, ISEM, CNRS, University of Montpellier
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Genetic diversity, the amount of variation in the genome sequence between individuals of the same species, varies among species for ecological and evolutionary reasons that are not fully understood. The neutral theory predicts that genetic diversity of a given species scale with its effective population size (Ne). In real populations, multiple factors that modulate the variance in reproductive success among individuals cause Ne to differ from the total number of individuals (N). Among these, age-specific mortality and fecundity rates are known to have a direct impact on the Ne/N ratio. However, the extent to which vital rates account for differences in genetic diversity among species remains unknown. We addressed this question by measuring the genome-wide average genetic diversity in 300 marine teleost fish from 16 different species. Adult lifespan was the best predictor of genetic diversity, long-lived species showing low level of genetic diversity. Retrieving the life tables of each of these species, we show that variance in reproductive success generated only by life tables is positively correlated to observed genetic diversity and can reproduce its variability. We further show that the increase reduction of Ne compared to the census size with adult lifespan may be explained by their particular age-specific survival rate (high juvenile and low adult mortality) and increasing fecundity with age. Our study highlights the importance of taking into account variance in reproductive success and particularly life tables to understand the pattern of genetic diversity. This path the way to understand the taxon-specific determinants of genetic diversity.

6. Balancing selection maintains ancient genetic diversity in C. elegans

Presenter: Lewis Stevens, Wellcome Trust Sanger Institute
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The mating system of a species profoundly influences its evolutionary trajectory. Across the tree of life, selfing species have evolved from outcrossing species thousands of times. The transition from outcrossing to selfing significantly decreases the effective population size, effective recombination rate, and heterozygosity of a species. These changes lead to a reduction in genetic diversity, and therefore adaptive potential, by intensifying the effects of genetic drift and linked selection. Selfing has evolved three times independently in the nematode genus Caenorhabditis, including in the model organism Caenorhabditis elegans, and all three selfing species show substantially reduced genetic diversity relative to outcrossing species. It remains unclear how selfing species with limited genetic diversity can adapt to the same diverse environments inhabited by their outcrossing relatives. Here, we discovered previously uncharacterised levels and patterns of genetic diversity by examining the whole-genome sequences from 609 wild C. elegans strains isolated worldwide. We found that genetic variation is concentrated in punctuated hyper-divergent regions that cover 20% of the C. elegans reference genome. These regions show signatures of long-term balancing selection and are enriched in genes that mediate sensory perception, pathogen response, and xenobiotic stress. Using long-read genome assemblies for 15 wild isolates, we found that hyper-divergent haplotypes contain unique sets of genes and show levels of divergence comparable to that found between Caenorhabditis species that diverged millions of years ago. Our results provide an example for how a species can avoid the evolutionary "dead end" associated with selfing via the long-term maintenance of genetic diversity.

7. The genetic architecture of target-site resistance to pyrethroid insecticides in the African malaria vectors Anopheles gambiae and Anopheles coluzzii

Presenter: Dr Chris Clarkson, Wellcome Sanger Institute
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Resistance to pyrethroid insecticides is a major concern for malaria vector control because these compounds are used in almost all insecticide-treated bed-nets, and are also used for indoor residual spraying. Pyrethroids target the voltage-gated sodium channel (VGSC), an essential component of the mosquito nervous system, but substitutions in the amino acid sequence can disrupt the activity of these insecticides, inducing a resistance phenotype. Here we use Illumina whole-genome sequence data from phase 2 of the Anopheles gambiae 1000 Genomes Project (1,142 wild caught samples) to provide a comprehensive account of genetic variation in the Vgsc gene in mosquito populations from 13 African countries. We describe 20 novel non-synonymous nucleotide substitutions at appreciable population frequency, and map these variants onto a molecular model of the protein to investigate the likelihood of a pyrethroid resistance phenotype. The genetic backgrounds of haplotypes carrying resistance alleles were analysed to determine which alleles have experienced recent positive selection, and to investigate the geographical distribution. We describe ten distinct haplotype groups carrying known resistance alleles; five are localised to a single geographical location, and five include haplotypes from different countries, in one case separated by over 3000 km. Markers are identified that could be used to design high-throughput, low-cost genetic assays for tracking the spread of pyrethroid resistance in the field. Our results demonstrate the molecular basis of target-site pyrethroid resistance in malaria vectors is more complex than previously appreciated, provide a foundation for the development of new genetic tools for insecticide resistance management.

8. Placing ancient DNA sequences into reference phylogenies

Presenter: Rui Martiniano, School of Biological and Environmental Sciences, Liverpool John Moores University
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Placing ancient DNA sequences into reference phylogenies

9. Not that clean: aquaculture mediated translocation of cleaner fish leads to hybridization and introgression on the northern edge of the species range

Presenter: Ellika Faust, University of Gothenburg
Co-Authors: Jansson, Eeva, Institute of Marine Research - Bergen; André, Carl, University of Gothenburg; Tallaksen Halvorsen, Kim, Institute of Marine Research - Austevoll; Dahle, Geir, Institute of Marine Research - Bergen; Knutsen, Halvor 4,5, Institute of Marine Research - Flødevigen & University of Agder; Quintela, María, Institute of Marine Research - Bergen; A. Glover, Kevin, Institute of Marine Research - Bergen & University of Bergen

Every year millions of wild caught wrasse are transported large distances to be used as cleaner fish for parasite control in marine salmon farms. Despite the environmental and genetic differences, translocated cleaner fish are able to escape and reproduce with local wild populations. Here we developed and implemented a panel of 84 SNPs to quantify the proportion of escaped and hybridized non-local corkwing wrasse (Symphodus melops) in western Norway, the world´s largest salmon producer, and an area where translocated cleaner fish are extensively used for parasite control. We found that escapees and hybrids may constitute up to 20 % of the local population at the northern edge of the species range. This introgression of southern genetic material at the edge of the species distribution has thus altered the

local genetic composition, and could obstruct local adaptation and further range expansion. Surprisingly, in other parts of the species distribution where salmon farming is also common, few escapees and hybrids were found. A possible explanation would be that the smaller newly established populations in the leading northern edge of this species range are more prone to introgression. The results obtained in this study, and the high genomic divergence between the main export and import regions, puts the sustainability of mass-translocation of non-local wild wrasse in question, and suggests the need for revision of the current management regime.

10. Overdominance maintains hypervariability at a sex-determination locus
Presenter: ILIK SACCHERI, University of Liverpool
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Primary sex-determination switches are subject to very rapid rates of evolutionary turnover for a variety interesting reasons, such as sabotage by endosymbionts. In Lepidoptera (WZ females / ZZ males), the canonical mechanism is the interaction between W-linked Feminizer and Z-linked Masculinizer. We have discovered an alternative mechanism in the butterfly Bicyclus anynana, that is based purely on the zygosity of Masc. This apparently single-locus mechanism comes with a cost, which is that all Masc homozygotes are lethal (through disrupted dosage compensation). Consequently, selection strongly promotes rarity, and over evolutionary time has resulted in extreme levels of allelic diversity across two exons of Masc. This system constitutes an example of true overdominance and suggests a radically different mechanism for controlling the primary protein signal.

Session 2

11. How good are predictions of the effects of selective sweeps on levels of neutral diversity?
Presenter: Brian Charlesworth, University of Edinburgh
Co-Authors: None

Selective sweeps are thought to play a significant role in shaping patterns of variability across genomes; accurate predictions of their effects are, therefore, important for understanding these patterns. A commonly used model of selective sweeps assumes that alleles sampled at the end of a sweep, and that fail to recombine with wild-type haplotypes during the sweep, coalesce instantaneously, leading to a simple expression for sweep effects on diversity. It is shown here that there can be a significant probability that a pair of alleles sampled at the end of a sweep coalesce during the sweep before a recombination event can occur, reducing their expected coalescent time below that given by the simple approximation. Expressions are derived for the expected reductions in pairwise neutral diversities caused by both single and recurrent sweeps in the presence of such within-sweep coalescence, although the effects of multiple recombination events during a sweep are only treated heuristically. The accuracies of the resulting expressions were checked against the results of simulations. For even moderate ratios of the recombination rate to the selection coefficient, the simple approximation can be substantially inaccurate.

12. Jointly inferring demography and purifying selection
Presenter: Parul Johri, Arizona State University
Co-Authors: Charlesworth, Brian, University of Edinburgh, Jensen, Jeffrey, Arizona State University

The question of the relative evolutionary roles of adaptive and nonadaptive processes has been a central debate in population genetics for nearly a century. While advances have been made in the theoretical

development of the underlying models, and statistical methods for estimating their parameters from large-scale genomic data, a framework for an appropriate null model remains elusive. A model incorporating evolutionary processes known to be in constant operation, genetic drift (as modulated by the demographic history of the population) and purifying selection, is lacking. Here, we investigate how the strength of purifying selection (background selection) affects patterns of variation at "neutral" sites near functional genomic components, and affects demographic inference when complete neutrality is assumed. The results demonstrate that, even after masking functional genomic regions, background selection effects may result in the false inference of population growth under models of both constant population size and decline. We propose a potential solution – a novel statistical framework for jointly inferring the contribution of the relevant selective and demographic parameters, accounting for linkage. Finally, we reanalyze genome-wide population-level data from a Zambian population of Drosophila melanogaster, and find that it has experienced a much slower rate of population growth than was inferred when the effects of purifying selection were neglected. Our approach represents an appropriate null model, against which the effects of positive selection can be assessed.

13. Is there any evidence for changes in natural selection acting on horse coat colours during domestication?

Presenter: Zhangyi He, Imperial College London
Co-Authors: Dai, Xiaoyang, University of Bristol; ; Beaumont, Mark, University of Bristol; ; Yu, Feng, University of Bristol;

Recent advances in ancient DNA (aDNA) preparation and sequencing techniques have made available an increasing amount of high-quality time serial samples of segregating alleles in ancestral populations. Such an additional temporal component allows direct access to the most important dimension in evolution, time, which has the promise of providing improved power for the inference of natural selection. With the increased power to link genotypes to phenotypes in natural populations, it is now possible to infer natural selection acting on the phenotype from genetic time series, which provides important insights into how natural selection shapes the distribution of phenotypic variation in time. However, it becomes intractable with most existing methods to handle the scenario that the phenotype is determined by multiple genes due to their interactions such as genetic linkage (e.g., KIT13 and KIT16 for horse coat patterns) and epistatic interaction (e.g., ASIP and MC1R for horse coat colours). To address this issue, we develop a novel Bayesian framework to infer natural selection acting on the phenotype from genotype frequency time series data while accounting for genetic linkage and epistatic interaction. Our procedure also provides statistical hypothesis testing to determine whether there are any changes in natural selection due to an event of interests like domestication. In addition, we model the flexibility of the sample individuals with uncertain genotypes in our approach, which is common in aDNA due to postmortem DNA damage. We believe that our method is a much needed advancement that will allow accurately utilising aDNA data in future.

14. Using full genome and epigenome data to infer past species history and life-history traits

Presenter: Thibaut Sellinger, Professorship for Population Genetics, Technical University of Munich, Germany
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Many methods based on the Sequentially Markovian Coalescent (SMC) have been and are being developed. These methods can be applied to many different species and make use of full genome sequence data to uncover population demographic history. We have recently developed a SMC method extending the original theoretical framework to estimate simultaneously the demographic history and biological variables, selfing and seed banking, affecting the ratio of population recombination rate by

population mutation rate (Sellinger et al. 2020). Furthermore, although convergence proofs in particular cases have been given using simulated data, a clear outline of the performance limits of SMC methods is lacking. We explore the limits of this methodology and present a tool that can be used to help users quantify what information can be confidently retrieved from given datasets. We then study the consequences for inference accuracy when violating the hypotheses and the assumptions of SMC approaches, such as the presence of transposable elements, variable recombination and mutation rates along the sequence and SNP call errors. We also provide a new interpretation of the SMC using the estimated transition matrix and offer recommendations for the most efficient use of these methods. At last, in this presentation we will present two recent new extensions of our method to 1) infer variation of selfing rates in time, and 2) include neutral methylation polymorphism data in addition to SNPs data. We show that methylome data boost the statistical power of the inference and improve its accuracy.

15. How are chromosomal inversions involved in adaptation to heterogeneous environments? (Population genomics using low-coverage whole-genome sequencing in a seaweed fly)
Presenter: Claire Mérot, IBIS, Université Laval, Québec, Canada
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The distribution of genetic diversity within a species, which includes the totality of geographic patterns of adaptive and non-adaptive differentiation, is highly variable across a genome. Genomic heterogeneity is shaped by the recombination landscape of each chromosome which is heavily impacted by structural genomic rearrangements. Large inversions can form massive co-segregating genomic blocks that evolve local adaptations even the face of gene flow. The seaweed fly Coelopa frigida harbours such chromosomal inversions and it occurs along a bioclimatic gradient and across a range of heterogeneous habitat. Here, we aim at examining the adaptive and evolutionary significance of the inversions relatively to collinear genome. For this, we assembled a high-quality genome to analyse 1,446 shallow whole-genome sequences, and we found that intra-specific genetic diversity is strongly structured by large non-recombining portions of the genome, including chromosomal inversions. Further, we detected only weak population differentiation despite the sampled locations being spread along 1500km and 10° of latitude, suggesting a high level of gene flow and extensive shared polymorphism. Contrasting with the rest of the genome, inversions and low-recombining regions differentiate populations more strongly than collinear regions, either along an ecogeographic cline or at a more fine-grained scale. Inversions and low-recombining regions were disproportionately involved in associations with environmental factors. We pointed out associations with phenotypes and candidate adaptive genes. Altogether, our results highlight the importance of recombination in shaping the selection-migration balance and indicate that inversions represent a reservoir of standing polymorphism facilitating adaptation to environmental heterogeneity at local and large scales. Note: Gene expression associated with a major chromosomal inversion in C. frigida will be presented in a companion talk by Dr. E. Berdan.

16. Using pool-seq data to detect chromosomal inversions contributing to adaptive divergence
Presenter: Anja Marie Westram, IST Austria; Nord University, Norway
Co-Authors: Faria, Rui, University of Porto; Morales, Hernan, University of Copenhagen; Barton, Nick, IST Austria; Johannesson, Kerstin, University of Gothenburg; Butlin, Roger, University of Sheffield

Evidence for the contribution of chromosomal inversions to adaptive divergence between populations is accumulating. However, with short-read sequencing data, the identification of inversions and their population frequencies remains challenging. This is the case especially when pooled sequencing data are used (i.e. no individual genotypes are available). In addition, it is often unclear which selection pressures work on inversions when diverging populations are separated along multiple environmental axes. Here, we use local adaptation in the marine snail Littorina saxatilis to address both points. L. saxatilis contains

two distinct and partially isolated ecotypes, adapted to wave-exposed vs. high-predation habitats. We obtained pool-seq data for each ecotype from 11 European locations. We used a simple visual approach to infer population inversion frequencies from these data, and found large differences in inversion frequencies between ecotypes for multiple inversions, sometimes across all sampled locations. We then used our extensive geographical sampling to narrow down the selection pressures working on these inversions. In some locations, the wave-exposed habitat is associated with high shore levels and the predation habitat is associated with low shore levels, while in other locations this relationship is reversed. We could therefore disentangle the effect of habitat vs. shore level on inversion frequencies. We found that multiple inversions that were assumed to be associated with ecotype adaptation are in fact associated with shore level, suggesting that they may contribute to e.g. temperature adaptation or desiccation tolerance.

17. A chromosomal inversion underlies forest adaptations in deer mice

Presenter: Olivia Harringmeyer, Harvard University
Co-Authors: Hager, Emily, Harvard University; Wooldridge, Brock, Harvard University; Hoekstra, Hopi, Harvard University

When organisms colonize novel environments, often multiple traits are involved in adaptation, raising the question of how these adaptive traits may coevolve and be co-inherited. To examine this question at the genetic level, we focused on two ecotypes of the North American deer mouse, Peromyscus maniculatus, a forest and a prairie form, thought to have recently diverged (~10kya) when the ancestral prairie form colonized new forested habitat following the last glacial retreat. Forest mice have substantially longer tails and darker coat colors than their prairie counterparts—two traits that likely arose as adaptations to the new forest environment. Using quantitative trait locus (QTL) mapping, we found that variation in both tail length and coat color map to the same genomic region on chromosome 15. This 41-Mb region shows suppressed recombination, elevated Fst, and extended linkage disequilibrium, indicating that it is likely a chromosomal inversion, which we confirmed with long-read sequencing. Next, we sampled mice along a 50-km transect between the forest and prairie populations and re-sequenced their genomes. We found that the inversion changes in frequency steeply across the transect, suggesting that the inversion may have been subjected to divergent selection associated with environmental differences. Finally, we found that the inversion was present in many, but not all, forest populations, suggesting different genetic mechanisms contribute to parallel forest phenotypes in deer mice. Together, our findings highlight how large chromosomal rearrangements can facilitate rapid adaptive events through linkage of multiple beneficial alleles affecting multiple phenotypic traits.

18. Evolutionary transitions from oviparity to viviparity are associated with convergent evolution of protein-coding genes and conserved non-coding elements in Cyprinodontiformes.

Presenter: Leeban Yusuf, University of St Andrews
Co-Authors: Lemus, Yolitzi Saldivar, National Autonomous University of Mexico, Thorpe, Peter, University of St Andrews, Garcia, Constantino Macías, National Autonomous University of Mexico, Ritchie, Michael G, University of St Andrews

Viviparity is a reproductive mode in which pregnant females retain developing embryos inside their reproductive tracts and give birth directly. In vertebrates alone, the transition to viviparity has occurred more than 150 times. This transition is associated with physiological and morphological changes, including eggshell reduction and maternal-fetal nutrient transfer. However, relatively little is known about the genetic changes associated with the convergent evolution of viviparity. Here, we use Cyprinodontiformes, an order of ray-finned fish comprising both viviparous and oviparous species, to identify regions of the genome that are associated with viviparity. We searched for signatures of molecular convergence amongst 21 species across the order (16 viviparous and 5 oviparous representing two transitions), in orthologous protein-coding genes (17,572) and conserved non-coding regions (729,185), by comparing substitution patterns between viviparous and oviparous species. We compared

substitution patterns at three levels: (a) at amino acid sites, (b) across entire protein-coding genes and (c) in conserved non-coding regions. We found (a) 2,038 convergent amino acid changes, (b) 532 genes that showed a significant association between relative evolutionary rate and the transition to viviparity and (c) 245 conserved non-coding elements that showed significant acceleration in divergence in branches where transitions to viviparity occurred. We show using gene ontology and RNA-seq, that these genetic changes are significantly enriched in functions related to embryonic development and that these genes show higher-than-average expression levels in mammalian placental tissues. Overall, these results indicate that molecular convergence of genes and non-coding regions underlie transitions from oviparity to viviparity in Cyprinodontiformes.

19. A gene's eye view of sexual antagonism
Presenter: Thomas Hitchcock, University of St Andrews
Co-Authors: Gardner, Andy, University of St Andrews

Females and males may face different selection pressures. Accordingly, alleles that confer a benefit for one sex often incur a cost for the other. Classic evolutionary theory holds that the X chromosome, whose sex-biased transmission sees it spending more time in females, should value females more than males, whereas autosomes, whose transmission is unbiased, should value both sexes equally. However, recent mathematical and empirical studies indicate that male-beneficial alleles may be more favoured by the X chromosome than by autosomes. We develop a gene's-eye-view approach that reconciles the classic view with these recent discordant results, by separating a gene's valuation of female versus male fitness from its ability to induce fitness effects in either sex. We use this framework to generate new comparative predictions for sexually antagonistic evolution in relation to dosage compensation, sex-specific mortality and assortative mating, revealing how molecular mechanisms, ecology and demography may drive variation in masculinization versus feminization across the genome.

20. Mother’s curse is pervasive across a large mito-nuclear Drosophila panel
Presenter: Florencia Camus, University College London
Co-Authors: Carnegie, Lorcan, University College London; Reuter, Max, University College London; Fowler, Kevin, University College London; Lane, Nick, University College London

The maternal inheritance of mitochondrial genomes entails a sex-specific selective sieve, whereby mutations in mitochondrial DNA can only respond to selection acting directly on females. In theory, this enables male-harming mutations to accumulate in mitochondrial genomes if they are neutral, beneficial, or only slightly deleterious to females. Ultimately, this bias could drive the evolution of male-specific mitochondrial mutation loads, an idea known as mother’s curse. Earlier work on this hypothesis has mainly used small Drosophila panels, in which naturally-sourced mitochondrial genomes were coupled to an isogenic nuclear background. However, the lack of nuclear genetic variation has precluded robust generalization. Here we test the predictions of mother’s curse using a large Drosophila mito-nuclear genetic panel, comprising 9 isogenic nuclear genomes coupled to 9 mitochondrial haplotypes, giving a total of 81 different mito-nuclear genotypes. This enables systematic testing of both mito-nuclear interactions and mitochondrial genetic variance. Following a predictive framework, we performed a screen for wing centroid size, as this trait is highly sexually dimorphic and depends on metabolic function. We confirmed that the trait is sexually dimorphic and show high levels of mito-nuclear epistasis. Importantly, we report that mitochondrial genetic variance has a greater impact on male versus female Drosophila, in 8 out of the 9 nuclear genetic backgrounds. These results demonstrate that the maternal inheritance of mitochondrial DNA does indeed modulate male life-history traits in a more generalisable way than previously envisaged.

Session 3

21. The effect of habitat loss and fragmentation on genetic variation: insights using spatio-temporal models

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Recent anthropogenic activities have largely contributed to habitat loss and fragmentation across the world, leading to 70% of worldwide remaining forests to be within 1 km of forest's edges (Haddad et al., 2015). In addition, it is estimated that 17% of the tropical moist forests have disappeared between 1990-2019 (Vancutsem et al., 2020), and predictive models suggest that by 2050 undisturbed forests will disappear entirely in large tropical humid regions. Habitat loss and fragmentation is expected to affect species population genetic variation, by decreasing within-population genetic diversity and increasing genetic differentiation among isolated populations. Most species worldwide have a geographically restricted dispersal, which results in a positive relationship between genetic and geographic distances, known as "isolation-by-distance" (IBD). In this work we used spatially explicit individual-based simulations to investigate i) how spatial genetic diversity changes within habitat fragments due to habitat contraction (local scale), taking into account habitat quality and edge-effect; and ii) how fast IBD pattern are lost following habitat loss and fragmentation (landscape scale). The aim of the present study is to provide a measure that quantify the additive effect of multiple edges in heterogeneous habitats on genetic diversity in order to formally test molecular edge effect in natural populations. Moreover, we provide insights on the rate at which IBD is lost after habitat loss and fragmentation and how past demographic events can influence such patterns.

22. The evolution of sex along an environmental gradient

Presenter: Louise Fouqueau, UMI EBEA 3614, Evolutionary Biology and Ecology of Algae, CNRS, Sorbonne Université, UC, UACH, Station Biologique de Roscoff, CS 90074, Place Georges Teissier, 29688 Roscoff
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While temporally changing environments generally favour sex and recombination, the effects of spatial environmental heterogeneity have been less explored. To remedy this, we used a classical model of adaptation along an environmental gradient to study the selective forces acting on reproductive mode evolution in the central and marginal parts of the distribution range of a species. The model considers a polygenic trait under stabilizing selection (the optimal trait value changing across space) and includes a demographic component imposing range limits. This model allowed us to explore the origin of geographic parthenogenesis, a term applied to describe a large variety of patterns where sexuals and related asexuals differ in their geographic distribution. The results show that in the central part of the range (where populations are well adapted), recombination tends to increase the mean fitness of offspring in regimes where drift is sufficiently strong (generating a benefit for sex), while it has the opposite effect when the effect of drift stays negligible. In agreement with previous results, we found that asexuality is often favoured in marginal populations, as it can preserve adaptation to extreme conditions. In general, conditions leading to important maladaptation at range limits (high effective environmental gradient, strong effect of drift at loci coding for the trait) increase the strength of selection for asexuality.

23. Addressing past demography, local ancestry and selection with genealogies and ancient human DNA

Presenter: Alice Pearson, University of Cambridge
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Recently, two new approaches have transformed our understanding of human population history. Firstly, sequencing of ancient DNA gives us a snapshot in time of the genetic variation present before historical events such as population bottlenecks and natural selection have obscured these signatures in the modern gene pool. Ancient DNA has thus revealed what cannot be determined from modern genomes alone. Secondly, the development of methods to reconstruct population histories and genealogies from genetic variation data has allowed inference of historical and current evolutionary processes in real world populations by understanding how these processes alter genealogies. New methods allow us to combine both approaches to infer genealogies involving both present-day and ancient individuals. I will discuss a method, using machine learning and tree sequences built by Relate from ancient and present-day Europeans and West Asians, that allows me to infer local ancestry along chromosomes and past demographic processes in a structured population, with the potential to explore time-resolved histories of selection and population size.

24. Deciphering Mechanisms of Trans-Specific Polymorphism in Arabidopsis

Presenter: Krzysztof Stankiewicz, University of Vienna/Gregor Mendel Institute
Co-Authors: Nordborg, Magnus, Gregor Mendel Institute. Hermisson, Joachim, University of Vienna.

Over time, polymorphisms segregating within a population reach fixation and are replaced by new mutations. This leads to the divergence of once closely related species, as shared polymorphisms are replaced by private polymorphisms. However, neutral expectations for trans-species polymorphism (TSP) based on molecular divergence timing between species often underestimate the observed amount of TSP. This phenomenon has previously been noted in Arabidopsis. We confirm this by using an expanded dataset with 1,135 accessions of A. thaliana, 85 individuals of A. lyrata, and 64 individuals of A. halleri. We observe that 2-5% of SNPs segregate in multiple species, which is 2 orders of magnitude higher than levels expected due to recurrent mutation, and several orders higher than the levels expected due to incomplete lineage sorting. We observe a positive correlation between TSP and coalescence time across the genome. Pericentromeric regions show fewer substitutions and higher levels of both private polymorphism and TSP. However, recurrent mutations alone do not explain this trend, as we see extensive clustering of TSP. This suggests that point processes, such as balancing selection, play a role in the generation or maintenance of this class of polymorphism. Previous reports have not found any blocks of identity by descent between A. thaliana and other Arabidopsis species, making recent gene flow unlikely. However, we observe considerable trans-specific haplotype structure in multiple regions and elevated LD between TSPs. We are currently using multiple de novo chromosome-level PacBio genome assemblies to understand the evolution of TSP, including possible trans-specific structural polymorphisms.

25. Isolation by Distance in Populations with Long-Range Dispersal

Presenter: Tyler Benjamin Smith, Emory University
Co-Authors: Weissman, Daniel, Emory University

Limited dispersal results in isolation by distance in spatially structured populations, in which individuals found further apart tend to be less related to each other. Models of populations undergoing short-range dispersal predict a close relation between the distance individuals disperse and the length scale over which two sampled individuals are likely to be closely related. In this work, we study the effect of long jumps on patterns of isolation by distance by replacing the typical short-range dispersal kernel with a

long-range, power-law kernel. We find that incorporating long jumps leads to a slower decay of relatedness with distance, and that the quantitative form of this slow decay contains visible signatures of the underlying dispersal process.

26. Local adaptation & speciation: misapprehensions at multiple dimensions
Presenter: Nathan White, University of Sheffield
Co-Authors: Butlin, Roger, University of Sheffield & University of Gothenburg

Divergent selection applied to one or more traits drives local adaptation and may lead to ecological speciation. Divergent selection on many traits might be termed 'multidimensional' divergent selection. There is a commonly held view that multidimensional divergent selection is likely to promote local adaptation and speciation to a greater extent than unidimensional divergent selection. We find that this view does not adequately hold up to scrutiny- in part because key concepts have become confused, but mainly because the theory is under-developed, and no experimental comparisons have been made. Using simulations, we address certain misapprehensions regarding multidimensional selection and disentangle core concepts such as the overall strength of selection and the number of loci involved. Finally, we present data from an experimental evolution study testing the evolution of local adaptation under unidimensional and multidimensional divergent selection regimes.

27. Population genomics, insights of domestic adaptation and dispersal in the kissing bug, Rhodnius ecuadoriensis
Presenter: Luis Enrique Hernandez Castro, University of Edinburgh
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Understanding the genomic basis of disease vectors adaptation to the human habitat and their dispersal capabilities is paramount for effective and sustainable control interventions, yet both mechanisms remain poorly understood in many arthropod-borne diseases. Using the main Chagas disease vector in southern Ecuador and northern Peru as a model, we genotyped 2,552 SNP markers of 272 Rhodnius ecuadoriensis triatomines to investigate gene flow, signatures of local adaptation and drivers of population structure and connectivity among populations. Evidence of high triatomine gene flow (e.g., low FST values, and little ecotype genetic/phylogenetic variation) between domestic and silvatic populations in several sites across the study area suggested insecticide-base control can be hindered. Genomic scans, based on machine learning (e.g., Random Forest), multivariate statistics (e.g., redundancy analysis) and FST-heterozygosity outlier method, revealed putative SNP loci under a strong signal of local adaptation which mapped to some annotated regions in the Rhodnius prolixus genome. Our isolation-by-distance and resistance generalised mixed models with maximum-likelihood population effects

parametrisation showed Rhodnius ecuadoriensis populations connectivity is driven by landscape heterogeneity. Moreover, we were able to detect highly connected and isolated population clusters which need to be target differently by vector control programmes.

28. Genomic convergence in guppies is dictated by limited shared variation, large haplotypes, and subtle allele frequency shifts

Presenter: James Whiting, University of Exeter
Co-Authors: Paris, Josie, University of Exeter; Van der Zee, Mijke, University of Exeter; Parsons, Paul, University of Sheffield; Weigel, Detlef, Max Planck Institute for Developmental Biology; Fraser, Bonnie, University of Exeter

High- (HP) and low-predation (LP) Trinidadian guppies are a textbook example of convergent evolution. LP guppies have repeatedly evolved from HP sources to be more colourful and have longer life histories, but, despite a plethora of phenotypic data, whether this process involves the same genes is unknown. Here, we present results from whole-genome sequencing data from five HP-LP river pairs to address this question, highlighting constraints that are predicted to reduce the likelihood of genetic convergence. We first analyse the distribution and structuring of genetic variation across our rivers, observing strict drainage-structuring, recurrent LP bottlenecks, and some limited between-river introgression. These results imply constraints on shared adaptive variation among LP populations. Selection scans across all river pairs highlight limited overlapping regions under selection, but genes of similar function are under selection in all five rivers, suggesting limited functional redundancy. Finally, we explore a large candidate haplotype fixed in three LP populations from the same drainage. We explore this region by reconstructing its phylogenetic history and using a novel multivariate method involving allele frequency (AF) vectors. Phylogenetics suggest these three LP populations have fixed a common ancestral haplotype, but further analysis of subtle AF changes hints all five rivers may be experiencing parallel and non-parallel selection at a TE-rich region within the haplotype. Combined, these results present the guppy as a compelling system in which convergent phenotypes are derived by convergent and nonconvergent changes at the genome level, and points towards important limitations in governing this phenomenon.

29. Genome-wide admixture patterns in a diverse set of replicated anthropogenic and natural hybrid populations support a polygenic architecture of species barriers in marine mussels

Presenter: Alexis Simon, Institut des sciences de l'évolution, Montpellier
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Human-mediated transport creates secondary contacts between genetically differentiated lineages, bringing new opportunities for genetic admixture and gene exchange. When similar introductions occur in different places, they provide informally replicated experiments for studying hybridisation outcomes. We recently identified two anthropogenically admixed lineages in the Mytilus edulis complex of species. One hybrid lineage results from the admixture of Mediterranean M. galloprovincialis with North Sea M. edulis and is confined in big commercial ports. The other results from the admixture of Atlantic M. galloprovincialis with Scandinavian M. edulis in Norway. Additionally, anthropogenic hybridization can be compared with multiple naturally admixed populations found in a large mosaic hybrid zone across France and the British Isles. We first genotyped 4,279 mussels with 77 ancestry-informative markers. This analysis allowed us to evidence parallelism encompassing natural and anthropogenic admixtures,as well as lab crosses, suggesting the effect of admixture is repeatable. To investigate more precisely the outcome of admixture events at the genome scale, we sequenced 206 reference and admixed genomes at low coverage. We confirmed repeatability but somewhat surprisingly did not observe any strong deviations to the genomic average admixture proportion. This observation does not fit classical post-

zygotic isolation models of large effect speciation genes and hints us towards a more polygenic architecture of species barriers.

30. Population genetics of the meadow spittlebug Philaenus spumarius, the main insect vector of Xylella fastidiosa in Europe

Presenter: Roberto Biello, Department of Crop Genetics, John Innes Centre, Norwich, United Kingdom
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The meadow spittlebug, Philaenus spumarius (Linnaeus, 1758) (Hemiptera: Aphrophoridae) is a xylem-feeding insect that vectors the bacterial pathogen Xylella fastidiosa, which has caused dramatic losses in wide variety of crops and most recently severely damaged the olive production industry in southern Italy. X. fastidiosa is present also in other European countries but not in the UK. P. spumarius is native to the Palearctic region, and was unintentionally introduced in other areas (e.g., the USA and New Zealand). The insect is considered a eurytopic and polyphagous species, tolerating a wide range of environmental factors and feeding on a variety of plant species. To improve our knowledge of the evolution of this species and assess the genetic structure of UK populations, we first assembled its 2.7 Gb genome. In addition, we resequenced whole genomes at 10x coverage of 116 individuals from six localities in the UK (Norfolk, Kent, Sussex, Yorkshire, Wales and Scotland). Across Europe, P. spumarius is split into three divergent mitochondrial lineages of which two are found in the UK – one in the north and one in the south – whereas the third is predominantly found in southern Europe. SNP data from whole-genome sequencing showed a weak genetic structure among the UK populations supported by isolation by geographical distance with evidence of admixture between the two lineages as a result of secondary contact. To investigate this further, we are generating a chromosome-level assembly of P. spumarius and have started to resequence genomes of individuals from populations across Europe, USA and New Zealand.

Session 4

31. Ethiopia's "tree against hunger": an ancient clonal crop with extensive phylogenetic diversity but unexpectedly low heterozygosity.

Presenter: Oliver White, Royal Botanic Gardens Kew
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Enset (Ensete ventricosum) is a close relative of the banana and a staple starch crop for 20 million people in the Ethiopian Highlands. Whilst wild populations of enset are sexually reproducing, domesticated landraces with important agronomic traits are maintained exclusively by clonal propagation. Farmers identify hundreds of distinct landraces based on a range of traits, therefore understanding the genomic basis of enset diversity is an essential step in securing sustainable food security in Ethiopia. We applied reduced representation sequencing to 250 samples across domesticated landraces and wild relatives.

Farmer survey data for landrace traits were also collated and compared with phylogenetic relationships. Phylogenetic analysis found a monophyletic origin of domesticated enset and extensive landrace diversity across Southern Ethiopia. Important agronomic traits were non-randomly distributed in the phylogeny, suggesting potential for future selection. Feral samples and evidence of introgression with wild populations was identified, suggesting sexual reproduction may occur in the absence of indigenous farming practices. Heterozygosity was lower than expected for domesticated populations, converse to the prediction that somatic mutations will accumulate with long term clonal propagation. We provide initial evidence that this may be due to a strong population bottleneck during domestication. Our study highlights the need to understand the mechanisms responsible for maintaining genetic variation in clonal crops such as enset, particularly in the context of climate change.

32. Evolutionary history and genetic connectivity across highly fragmented populations of an endangered daisy

Presenter: Yael Rodger, Monash University, Australia
Co-Authors: Pavlova, Alexandra, Monash University. Sinclair, Steve, Arthur Rylah Institute. Pickup, Melinda, Greening Australia. Sunnucks, Paul, Monash University.

Conservation management can be aided by knowledge of genetic diversity and evolutionary history, so that ecological and evolutionary processes can be preserved. The Button Wrinklewort daisy (Rutidosis leptorrhynchoides) was a common component of grassy ecosystems in south-eastern Australia. It is now endangered due to extensive habitat loss and the impacts of livestock grazing, and is currently restricted to a few small populations in two regions >500 km apart, one in Victoria, the other in the Australian Capital Territory and nearby New South Wales (ACT/NSW). Using a genome-wide SNP dataset, we assessed patterns of genetic structure and genetic differentiation of twelve natural diploid populations. We estimated intrapopulation genetic diversity to scope sources for genetic management. Bayesian clustering and principal components analyses showed strong population genetic differentiation between the two regions, and substantial substructure within ACT/NSW. A coalescent tree-building approach implemented in SNAPP indicated evolutionary divergence between the two distant regions. Among the populations screened, the last two known remaining Victorian populations had the highest genetic diversity and effective population size over time, despite having among the lowest recent census sizes. A maximum likelihood tree method implemented in TREEMIX indicated little or no recent gene flow among populations except potentially close neighbours. Populations that were more genetically distinctive had lower genetic diversity, implying that drift in isolation is likely driving population differentiation, hence re-establishing gene flow among them is desirable. This work illustrates how SNP data can support evidence-based conservation decision-making that embraces the importance of protecting evolutionary processes.

33. Understanding telomere lengthening in a wild vertebrate population

Presenter: Thomas James Brown, University of East Anglia
Co-Authors: Spurgin, Lewis, University of East Anglia; Dugdale, Hannah, University of Groningen; Komdeur, Jan, University of Groningen; Burke, Terry, University of Sheffield; Richardson, David, University of East Anglia

Telomeres have been advocated to be important markers of biological age in evolutionary and ecological studies. Evidence indicates that telomere shortening is associated with environmental and life-history stressors and linked to subsequent mortality risk. In recent years there has been growing acceptance that telomere lengthening – an apparent increase in telomere length between repeated samples from the same individual – also occurs. However, the exact circumstances, and consequences, of telomere lengthening are poorly understood. Using longitudinal data from the Seychelles warbler (Acrocephalus sechellensis), we tested whether telomere lengthening – which occurs in adults of this species – is associated with specific stressors, namely, reproductive effort, food availability, malarial infection and cooperative breeding. Telomere dynamics of males were not associated with stressors. Telomere

shortening was observed in females under greater stress (i.e. low food availability, infected with malaria), while telomere lengthening was observed in females experiencing good conditions (i.e. high food availability, assisted by helpers and without malaria). These results indicate that, at least for females, telomere lengthening occurs in circumstances more conducive to self-maintenance. Importantly, both females and males with lengthened telomeres had improved subsequent survival relative to individuals that experienced no change, or shortening, telomeres – indicating that telomere lengthening is associated with individual fitness. These results indicate that telomere dynamics are bidirectionally responsive to the temporal changes in stress that an individual faces, and thus may poorly reflect the accumulation of stress over the lifetime. As such this study challenges how we think of telomeres as a marker of biological age.

34. Whole-genome analysis of historical samples shows no evidence for a genetic decline of North American monarch butterflies.

Presenter: Venkat Talla, Postdoctoral researcher, Emory University
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Human-caused environmental change and habitat destruction are driving the loss of biodiversity in many species, including insects, birds and mammals. Drastic declines in census sizes of a population should result in concomitant declines in genetic diversity. Genetic drift is a process by which alleles are either removed or fixed randomly in a natural population. As genetic drift is stronger in smaller populations, genetically impoverished populations risk inbreeding, reduced adaptive potential and increased fixation of deleterious mutations. It is thus critical to monitor the genetic diversity of populations to determine the impact of environmental changes and conservation efforts on population health. With the advent of sequencing technologies, researchers can now employ next-generation sequencing of natural history collections to quantify the temporal change in genome-wide genetic diversity. Recent studies have used this approach to demonstrate rapid reductions in genetic diversity in European honeybees, Sunda fruit bats, and the Crested Ibis reflecting the reductions in their census population sizes. North American monarchs are known to make one of the longest seasonal migrations of any insect species. They fly as many as 4,500 km to reach their overwintering sites in central Mexico from their breeding grounds in the United States and Canada. Recent studies have raised concerns over the monarch butterfly, culminating in a petition to the US Fish and Wildlife Service to protect monarchs under the Endangered Species Act. In this study, we examined full-genome sequences from monarchs sampled 120-150 generations apart to quantify the extent of genetic decline in North America.

35. Infection dynamics, dispersal, and adaptation: Understanding the lack of recovery in a remnant frog population following a disease outbreak

Presenter: Donald McKnight, James Cook University
Co-Authors: Carr, Leah, University of New England; Bower, Deborah, University of New England; Schwarzkopf, Lin, James Cook University; Alford, Ross, James Cook University; Zenger, Kyall, James Cook University;

Emerging infectious diseases can cause dramatic declines in wildlife populations. Sometimes these declines are followed by recovery, but many populations do not recover. Studying differential recovery patterns may yield important information for managing disease-afflicted populations and facilitating population recoveries. In the late1980s, a chytridiomycosis outbreak caused multiple frog species in Australia's Wet Tropics to decline. Populations of some species (e.g., Litoria nannotis) subsequently recovered, while others (e.g., Litoria dayi) did not. We examined the population genetics and current infection status of L. dayi, to test several hypotheses regarding the failure of its populations to recover: 1) a lack of individual dispersal abilities has prevented recolonization of previously occupied locations, 2) a loss of genetic variation has resulted in limited adaptive potential, and 3) L. dayi is currently adapting to chytridiomycosis. We found moderate to high levels of gene flow and diversity (Fst range: <0.01–0.15; minor allele frequency: 0.192–0.245), which were similar to previously published levels for recovered L.

nannotis populations. This suggests that dispersal ability and genetic diversity do not limit the ability of L dayi to recolonize upland sites. Further, infection intensity and prevalence increased with elevation, suggesting that chytridiomycosis is still limiting the elevational range of L. dayi. Outlier tests comparing infected and uninfected individuals consistently identified 18 markers as putatively under selection, and several of those markers matched genes that were previously implicated in infection. This suggests that L. dayi has genetic variation for genes that affect infection dynamics and may be undergoing adaptation.

36. Genomic tales: An history of European barn owl.

Presenter: Cumer Tristan, Department of Ecology and Evolution, University of Lausanne, Lausanne, Switzerland
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Current patterns of genetic diversity are largely driven by the past demographical population history. In Europe, during the Last glacial Maximum (LGM), most species were found only in a few isolated refugia around the Mediterranean Sea. At the end of LGM, these species recolonized northern environments via different routes, adapted to new environments along the way, and eventually, lineages from distinct refugia came into contact again. In this study, we investigated the demographic history of barn owl (Tyto alba), a cosmopolitan bird in Europe, by taking advantage of whole genome sequencing of over a hundred individuals, from different locations distributed all around the Mediterranean Sea and in continental Europe. Sampled individuals fall into two distinct linages: one represents the near East while all continental European individuals descend from the second. Haplotype information provides evidence of limited gene flow between these lineages in the Anatolian region. In continental Europe, we identify the Iberian Peninsula as a main refugium for this species, which largely contributes to the diversity of current populations in the North of the Alps. While these data are coherent with previous results based on microsatellite markers, we also identify a more cryptic refugium in a region encompassing Italy, Greece and the Aegean Sea. Our data suggest that a contact zone between these two different lineages is located in the Balkans Peninsula. This highly mobile bird provides a good model to study the genomics of range expansion after glaciation, investigate the genetic basis of local adaptation considering past demography and understand genomic implications of various barriers to gene flow, in both neutral and selective perspectives.

37. Hybridisation and introgression in the Scottish wildcat

Presenter: Jo Howard-McCombe, University of Bristol
Co-Authors: Senn, H., Royal Zoological Society of Scotland; Lawson, D., University of Bristol; Kitchener, A., National Museums Scotland; Beaumont, M., University of Bristol

The Scottish wildcat (Felis silvestris) is one of the UK's most endangered mammal species and is now considered to be functionally extinct in the wild in Scotland. Hybridisation with domestic cats is the most serious threat to wildcats in the UK, counteracting all efforts to prevent decline due to habitat loss and persecution. Using both ddRAD and whole-genome datasets (108 and 45 individuals, respectively), we aim to determine the timescale and mode of introgression in Scottish wildcats and evaluate the effectiveness of current hybrid tests. Using this information, we aim to better inform management of the captive population and generate a robust model for future wildcat conservation. We show that a population of wildcats genetically distant from domestic cats is still present in Scotland, though these individuals are found almost exclusively within the captive breeding program; most wild-living cats sampled were introgressed to some extent. An ABC approach to modelling the ddRAD data supported significant recent introgression in the wildcat population, and whole-genome haplotypic data appears to corroborate these results. We also used the ddRAD data to compare the accuracy of the existing field-

based hybrid tests and show that the current low-cost SNP test is a better predictor of the ddRAD score than one based on morphology.

38. The impact of recurrent origins, genetic drift and gene flow on the genetic structure of allopolyploid marsh orchids (Dactylorhiza, Orchidaceae)

Presenter: Anna-Sophie Hawranek, Department of Botany and Biodiversity Research, University of Vienna, Rennweg 14, A-1030 Vienna, Austria
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Recurrently emerging polyploids are known from various organisms, and are especially frequent in plants. Early generation polyploids must quickly recover from the genetic bottleneck associated with their origin. Their genetic variation can be enriched either via multiple origins out of distinct parental populations, or from subsequent introgression from relatives. Inferences of polyploid evolutionary history are hampered by the complex inheritance and difficult allele dosage assessment. We focus here on the genetic structure of an allotetraploid European complex within Dactylorhiza, which has originated through recurrent unidirectional crosses of the same diploid parental pair. With a RADseq data set comprising hundreds of accessions across their distribution area, we apply a genotype likelihoods-based method to first separate the paternal from maternal homoeologs, taking as reference allelic frequencies assessed across about one hundred representatives of both parental taxa. The consistent signal obtained from each polyploid subgenome uncovers a complex genetic structure shaped by polytopic independent origins, isolation by distance, and opportunistic, asymmetrical and partly intense regional gene flow between allopolyploids. Each primary allopolyploid lineage expanded to occupy large European areas. While segregating further as a result of genetic drift in allopatry, the allopolyploids secondary came into contact in several sympatric areas. Furthermore, our analyses revealed a more frequent introgression from diploids to tetraploids, likely facilitated by the absence of an endosperm in this plant system. Our inferences uncover main phases and contributors during the evolution of allopolyploid marsh orchids, whereas our analytic approach should prove useful for other studies featuring non-model allopolyploids.

39. Conservation genomics of Wallacea's endemic ungulates

Presenter: Sabhrina Gita Aninta, Queen Mary University of London, UK; Natural History Museum, London, UK
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Wallacea is a biodiversity hotspot characterised by several endemic mammals, including the anoa (“dwarf buffalo”, Bubalus spp.) and babirusa (“deer-pig”, Babyrousa spp.). Increasing hunting pressure and local deforestation have led to the decline of around 7% and 28% for anoa and babirusa baseline population respectively, and their persistence will highly depend on their long term survival potential. To assess the adaptive potential of the remaining population, we generated whole genome sequences of 20 individuals from each taxa and determined the extent of inbreeding in these individuals. Using a method that jointly

estimates heterozygosity and runs of homozygosity (ROH) implemented in ROHan, we found that anoa has lower heterozygosity than babirusa and tends to possess longer ROH segments. This suggests that anoa was likely more affected by recent inbreeding than babirusa. Anoa also contain a higher mutation load than babirusa, with different abundance of deleterious mutations across different regions. Given that mutation load likely reflects older demographic processes than long ROH, this raises the possibility that anoa populations which were likely smaller in the past (prior to anthropogenic disturbances) may be more affected by recent inbreeding than babirusa. To further test this hypothesis, we are now generating genomes from animals sampled from the early 20th century, which predate major recent deforestation. Although our findings still need further tests and more samples to more confidently determine the population history of the taxa, these results suggest that different populations possess different adaptive potential that needs to be managed accordingly.

40. Conservation genetics for the management of black rhinoceros (Diceros bicornis michaeli) in Tanzania.

Presenter: Ronald Vincent Mellya, University of Glasgow
Co-Authors: Fyumagwa, Robert-Tanzania Wildlife Research Institute, Arusha, Tanzania; Macha, Emmanuel-Tanzania National Parks, P.O. Box 3134, Arusha, Tanzania.; Mjingo, Ernest-Tanzania Wildlife Research Institute, Arusha, Tanzania.; Hopcraft, Grant- University of Glasgow, Glasgow, G12 8QQ, United Kingdom.; Mable, Barbara-University of Glasgow, Glasgow, G12 8QQ, United Kingdom.;

The genetic structure of a population indicates the potential for local adaptation, and maintenance of genetic diversity through gene flow among populations allows organisms to evolve and contend with continuous environmental changes. The loss of genetic diversity may reduce reproductive fitness, which may increase risks of extinction. Investigation of patterns of genetic variation provides information on the dispersal of species, mating behaviors and population boundaries, which can allow conservation managers to estimate rates of inbreeding and to design specific translocation strategies to increase evolutionary potential. Furthermore, a more comprehensive understanding of the distribution of genetic variants among populations could provide critical forensic evidence in the case of poached animals. However, despite its high conservation profile, still nothing is known about existing levels of genetic variation of the remaining populations of the Eastern black rhinoceros (Diceros bicornis) in Tanzania. Therefore, the aim of this study is to apply conservation genetics techniques to generate information on the genetic status of all extant six Black rhinoceros populations from four protected areas in Tanzania (Serengeti National Park, Ngorongoro Conservation Area Authority, Mkomazi National Park and Grumeti Reserve) and one neighbor population from Kenya (Maasai Mara) for: 1) estimating rates of inbreeding and movement of individuals between populations; 2) establishing population viability models for management. This information will be used to formulate a genetically viable management strategy for managing black rhinoceros populations in Tanzania, including designing specific translocation strategies for maximizing distribution of genetic variation across subpopulations.

Jan 5th, 2021

Session 1

41. Deleterious effects in runs of homozygosity with different map lengths reveal patterns of purging in free-living Soay sheep.

Presenter: Martin A. Stoffel, University of Edinburgh
Co-Authors: Johnston, Susan E., University of Edinburgh, Pilkington, Jill G., University of Edinburgh, Pemberton, Josephine M., University of Edinburgh

Inbreeding and purging shape fitness by affecting patterns of deleterious mutations, but how this works in wild populations remains largely elusive. Here, we explored inbreeding depression and purging by comparing the deleterious effects contained in runs of homozygosity (ROH) with long, medium and short genetic map lengths using simulations and empirical analyses. Simulations across a range of selection and

dominance coefficients consistently showed a decrease in deleterious mutations from long to short ROH, reflecting how purifying selection has had more time to act on older and hence shorter haplotypes. We tested this empirically by estimating the effects of individual inbreeding coefficients FROH based on long, medium and short ROH in a dataset of 4592 wild Soay sheep genotyped at 417K SNP loci. In line with the simulations, inbreeding depression in survival was strongest when FROH was based on long ROH, weaker for medium ROH and absent for short ROH. Our study illustrates how patterns of selection against deleterious mutations can be quantified by comparing young and old identical-by-descent haplotypes forming ROH. Importantly, however, we show that strong inbreeding depression persists in a wild population despite ongoing purging and a historically small population size.

42. Why so white? Barn owls in the British Isles

Presenter: Ana Paula Machado, Department of Ecology and Evolution, University of Lausanne, Switzerland
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Islands and the atypical organisms that inhabit them have greatly contributed to the development of evolutionary theory. With the large-scale data available nowadays, one can attempt to disentangle the roles of demographic history and natural selection in generating intraspecific genetic and phenotypic diversity of island populations. In continental Europe, barn owls (Tyto alba) display a plumage colouration cline: owls in the South are mostly white coloured whereas in the North they are dark-reddish. Unlike the neighbouring populations in North-western Europe, barn owls in the British Isles are predominantly white. The reasons for such disparity in coloration have thus far eluded our understanding. Using whole-genome resequencing data for 61 individuals in western Europe and the British Isles, we show that British Isles barn owl populations are isolated from their mainland counterparts and this is supported by capture-recapture data. Demographic simulations suggest that the British Isles and North-western Europe originated from the same Southern refugium in two waves, but likely split from each other early on after the last Glacial period. Genome scans confirm a lower differentiation between insular and Southern populations and a higher differentiation in Northern mainland populations at regions linked to colour determination. Overall, our results suggest that demography, as opposed to selection, may be the main evolutionary driver of the white colouration of these insular populations.

43. Selection on ancestral genetic variation fuels parallel ecotype formation in bottlenose dolphins

Presenter: Marie Louis, University of St Andrews - University of Copenhagen
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What are the mechanisms that allow species to extend their ranges and adapt to the novel environmental conditions they find in the newly available habitat? The study of parallel adaptation of pairs of populations to similar environments can provide great insights into this question. Here, we test for parallel evolution driven by habitat specialization in a highly social marine mammal, the common

bottlenose dolphin, Tursiops truncatus, and investigate the origins of the genetic variation driving local adaptation. Coastal ecotypes of common bottlenose dolphins have recurrently emerged in multiple regions of the world from pelagic ecotype populations, when novel habitat became available. Analyzing the whole genomes of 57 individuals using comparative population genomics approaches, we found that coastal ecotype evolution was relatively independent between the Atlantic and Pacific, but related between different regions within the Atlantic. We find repeated selection on ancient alleles in the coastal populations, present as standing genetic variation in the pelagic populations. Genes evolving under parallel selection across distant coastal habitats have roles in cognitive abilities and feeding. Therefore, parallel adaptation in long-lived social species may be driven by a combination of ecological opportunities, selection acting on ancient variants, and stable behavioural transmission of ecological specialisations. Tried and tested genetic variation that has been subject to repeated bouts of selection, may promote linked adaptive variants with minimal pleiotropic effects, thereby facilitating their persistence at low frequency in source populations and enabling parallel evolution.

44. Is temperature adaptation between D. simulans and D. melanogaster parallel?

Presenter: Dagný Ásta Rúnarsdóttir, Vienna Graduate School of Population Genetics, Vetmeduni Vienna.
Co-Authors: Schlötterer, Christian, Institut für Populationsgenetik, Vetmeduni Vienna.; Nolte, Viola, Institut für Populationsgenetik, Vetmeduni Vienna.

Parallel phenotypic responses to similar environmental stressors has been described not only for different populations, but also between species. Since many of these traits are highly complex and selected variants may not be shared, it is not clear to what extent this parallel evolution can be also seen on the molecular level. Here, we address this question by studying the transcriptomic response of polymorphic founder populations from two species, Drosophila melanogaster and D. simulans to fluctuating high (18/28°C) and low (10/20°C) temperature regimes. In both species a strong adaptive response in gene expression is seen, but only 0.03% of the genes shared a significant expression change in the same direction. Nevertheless, considering the full set of genes, a clear signal of a shared response to temperature could be seen in both species. We test the impact of genetic redundancy of temperature adaptation by quantifying the degree of parallel evolution of both species for single genes and on the level of biological processes. Our results have broad implications for current attempts to understand parallel evolution on the molecular level.

45. A ddRAD glimpse into the evolutionary origins of an Arctic charr adaptive radiation

Presenter: Marina de la Cámara, University of Iceland
Co-Authors: Kapralova, Kalina H., University of Iceland;; Morrissey, Michael B., University of St Andrews

Arctic charr in lake Thingvallavatn (Iceland) represents one of the most iconic systems of adaptive divergence. Like other freshwater fish involved in sympatric ecological diversifications, Arctic charr has diverged along the benthic-limnetic ecological axis, resulting in four different ecomorphs. Two of these morphs are benthic, foraging on gastropods on the bottom of the lake: a small (SB) and a large benthic (LB). The two others are limnetic: a planktivorous (PL) and a piscivorous (PI), which feed mainly on zooplankton and three-spined sticklebacks, respectively. Phenotypically, they differ in size, trophic morphology, body shape, colouration, behaviour, life history traits and parasitic load. They are also genetically distinct, although the evolutionary history of the PI morph remains unknown. Due to our lack of knowledge about its biology, its potential misidentification and lower representation in the lake, this morph is generally not included in genomic studies on the Thingvallavatn system. Hypotheses about the origin of the PI range from an ontogenetic shift from the PL morph to a case of hybridisation. Here, we use reduced representation genome data (ddRAD-seq) to look at population structure, introgression and signatures of selection to investigate the evolutionary origin of this ecomorph.

46. Genomic signatures of population history and adaptation across island bird populations

Presenter: Claudia Anne Bethany Martin, University of East Anglia (UEA)
Co-Authors: Armstrong, Claire, UEA; Illera, Juan Carlos, Oviedo University; Emerson, Brent C., Institute of Natural Products and Agrobiology (IPNA-CSIC); Richardson, David S., UEA; Spurgin, Lewis G., UEA

Island archipelagos provide excellent models to understand evolutionary processes. Colonisation events and gene flow may interact with selection to shape genetic variation at different spatial scales. Landscape-scale variation may drive selection within islands, while long-term evolutionary processes may drive divergence between distantly related populations. Here, we examine patterns of population history and selection between Berthelot's pipit populations (Anthus berthelotii), a passerine endemic to three North Atlantic archipelagos. We use RAD-seq markers to uncover greater detail on: (1) Population history across this species range; (2) the strength of selection acting between recently diverged island populations (<8,000 yrs) compared to those inhabiting different archipelagos (>2 million yrs); and (3) identify loci under selection between recently diverged populations. We ask whether genomic data suggest similar patterns of ecological adaptation across geographic and temporal scales in this species. Together these analyses provide better understanding of how population history, drift and selection shape ecological diversity.

47. Investigating mitochondrial evolution in UK Lepidoptera over the 20th Century.

Presenter: Greg Hurst, University of Liverpool
Co-Authors: Siozios, Stefanos, University of Liverpool; Whiteford, Sam, University of Liverpool; Yung, Carl, University of Liverpool; Yung, Carl, University of Liverpool; MacGregor, Callum, University of York; Bridle, Jon, University College London; Hill, Jane, University of York; Saccheri, Ilik, University of Liverpool; and the Velocity Project team.

Mitochondria were once 'the' neutral marker used for demographic and phylogeographic inference. More laterally, mtDNA has been found to have richer evolutionary history of adaptive evolution, as well evolutionary changes driven by indirect selection from coinherited heritable microbes. We employed a population genomic approach, comparing museum and contemporary material, to directly determine the degree of change in mtDNA haplotype frequency over time. We present a comparison of mtDNA haplotypes in archival museum material collected in the early 20th century with spatially matching conspecifics collected in the early 21st century for 11 species of UK Lepidoptera, and use these to assess the degree of mtDNA evolution that has occurred in the last 100 years.

48. Nature and variability of de novo mutations in Chlamydomonas

Presenter: Eugenio López-Cortegano, University of Edinburgh (United Kingdom)
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Mutations play a key role in the evolution of populations, since they regenerate genetic variation and fuel adaptation. Here, we examine the nature and variability of mutation in the green algae of the genus Chlamydomonas. First, we used short-read Illumina sequencing of mutation accumulation (MA) lines to compare the mutational properties of C. reinhardtii and its close relative C. incerta. These results show that the mutation rate, the distribution of inter-mutation distances, and the genomic factors associated with the mutation rate are similar in the two species, allowing for the prediction of mutability across-species. However, the spectrum of single nucleotide mutations (SNM) differs between the two species, indicating this factor has more freedom to diverge. In addition, the rates of SNM and insertion and deletion (INDEL) variants rates were highly correlated ($r > 0.97$), and variability in the mutation rate between MA lines was substantial for the two species. Second, we used long-read PacBio sequencing on

two C. reinhardtii MA lines. Preliminary results show that the mutation rate for structural variation is μ = 3.45 x 10e-10, which is about the same order of magnitude as that of INDELs (μ = 4.24 x 10e-10), and most of these variants correspond to transposition events or occur within transposons (~3/4). However, other variants possibly affecting coding regions were observed, with a mean size of the order of several kilobases in length. Our results provide new insight into the source and nature of mutational variation in Chlamydomonas, and the evolution of their genomes.

49. Runaway GC evolution in gerbil genomes

Presenter: Rodrigo Pracana, Deparment of Zoology, University of Oxford
Co-Authors: Dai, Yechen, University of Oxford; Holland, Peter, University of Oxford

Biases in mutation and recombination can cause the fixation of non-adaptive and maladaptive alleles in populations. For instance, deleterious mutations can spread in the population by processes such as GC-biased gene conversion or as a result of low recombination rates. The fat sand rat (Psammommys obesus, a species of gerbil) carries a ~10 Mb region of the genome with approximately 80 genes that have an extremely high GC content. Several of the genes in the region, including the homeobox gene PDX1, have a high nonsynonymous GC-skew, despite considerable sequence conservation across other vertebrates. This nonsynonymous divergence suggests that this species has been affected by non-adaptive GC accumulation. Here, we measure substitution rates for AT-to-GC, GC-to-AT and GC-conservative mutations for orthologous genes of four gerbil species, which we compare to mouse and rat. We show that (1) all genes in the region have an elevated substitution rate for all mutational categories; (2) that the substitution rate for AT-to-GC mutations in the region is higher than for the other categories, consistent with the observed GC skew; and (3) that gerbils have other clusters of GC-skewed genes scattered across their genome. Our results suggest that the gerbil lineage has been affected by the evolution of genomic regions with high recombination rates, probably higher than documented in other mammals. We discuss that gerbils may represent a model with which to study the evolution of recombination, as well as to study the evolution of compensatory mechanisms against the deleterious effects of GC-biased gene conversion.

50. Evolution of phenotypic variance provides insights into the genetic basis of adaption

Presenter: Wei-Yun Lai, Vienna Graduate School of Population Genetics, Vetmeduni Vienna, Vienna, Austria.
Co-Authors: Nolte, Viola, Institut für Populationsgenetik, Vetmeduni Vienna, Vienna, Austria; Jakšić, Ana Marija, École polytechnique fédérale de Lausanne, Lausanne, Switzerland; Schlötterer, Christian, Institut für Populationsgenetik, Vetmeduni Vienna, Vienna, Austria.

Most traits are polygenic and the contributing loci can be identified by GWAS. Their adaptive architecture is, however, poorly characterized. Here, we propose a new approach to study the adaptive architecture, which does not depend on genetic data. Relying on experimental evolution we measure the phenotypic variance in replicated populations during adaptation to a new environment. Extensive computer simulations show that the evolution of phenotypic variance in a replicated experimental evolution setting is a powerful approach to distinguish between oligogenic and polygenic adaptive architectures. We apply this new method to gene expression variance in male Drosophila simulans before and after 100 generations of adaptation to a novel hot environment. The variance change in gene expression was indistinguishable for genes with and without a significant change in mean expression after 100 generations of evolution. We conclude that adaptive gene expression evolution is best explained by a highly polygenic adaptive architecture. We propose that the evolution of phenotypic variance provides a powerful approach to characterize the adaptive architecture, in particular when combined with genomic data.

51. Repeated recruitment is the dominant source of adaptive variation in autopolyploid Arabidopsis arenosa

Presenter: Veronika Konečná, Charles University, Faculty of Science
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Relative contributions of pre-existing vs de novo genomic variation to adaptation are poorly understood, especially in higher ploidy organisms, which maintain increased variation. Here we assess this in high resolution using autotetraploid Arabidopsis arenosa, which repeatedly adapted to toxic environments. Leveraging a fivefold replicated design, we demonstrate parallel adaptation and assess selection on both structural variants (specifically TEs) and SNPs in 78 resequenced individuals. We discovered substantial parallelism in candidate genes involved in ion homeostasis and stress signalling. We further modelled parallel selection in these loci and inferred that selection repeatedly sampled from a shared pool of variants in nearly all cases, supporting theoretical expectations. A single, striking exception is represented by TWO PORE CHANNEL 1, which exhibits convergent evolution from independent de novo mutations at an identical site at the calcium channel selectivity gate. Taken together, this suggests that polyploid populations can rapidly adapt to environmental extremes, calling on both pre-existing variation and novel polymorphisms.

52. Whole-genome resequencing reveals the genetic bases for local adaptation in the wild tomato species Solanum chilense

Presenter: Kai Wei, Professorship for Population Genetics, Department of Life Science Systems, Technical University of Munich Freising, Germany
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When colonizing new habitats, plants need to adapt to new challenges of the environment. Examining adaptive signatures under positive selection, while taking into account the effect of the demographic history, aids our understanding of how plant species evolve to overcome these environmental constraints. The wild tomato species Solanum chilense occurs from mesic regions in Southern Peru to hyper-arid habitats around the Atacama desert in Northern Chile. The species also encompasses habitats from the Pacific coast up to 3500 m of altitude in the Andes mountains. Thus, S. chilense represents an ideal system to study the interplay between demography and natural selection. We used whole-genome re-sequencing data from six Solanum chilense populations (total of 30 individuals) representing the whole species distribution range. First, population structure and demographic history analysis supports two independent colonization events, with mild bottlenecks, starting from the central group of populations toward isolated coastal and highland regions in the south. Second, positive selection analyses revealed that biological regulation genes related to flowering, circadian rhythm, root hair differentiation and vernalization response could promote adaptation events in the different populations. Furthermore, genotype-environment association tests revealed genes of the circadian rhythm and flowering time (AMY3) likely responsible for the adaptation of highland habitats subject to intense solar radiation. Our study provides a new set of candidate pathways underpinning local adaptation to different environmental conditions of the newly colonized habitats in an outcrossing plant species which colonized extremely arid habitats.

Session 2

53. A large and diverse autosomal haplotype is associated with sex-linked colour polymorphism in the Trinidadian guppy

Presenter: Josephine Paris, University of Exeter
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Colour polymorphism provides a tractable trait to explore the evolution of sexual selection and sexual antagonism. In the Trinidadian guppy, male colouration patterns are influenced by both natural and sexual selection, while increased pattern variation is thought to be a result of negative frequency dependent selection. Since colour patterns in guppies are often inherited faithfully from father to sons, it has been presumed that colour genes are physically linked to the sex determining loci, perhaps as a 'supergene'. But to date, the identity and genomic location of the underlying colour pattern genes has remained elusive. We used a pool-sequencing approach on four 'Iso-Y lines' of guppies, where colour was inherited along the patriline, but backcrossed into the stock population every generation for 40 generations. We uncovered a surprising architecture for colour pattern evolution. Iso-Y lines were consistently divergent at a large diverse haplotype (~4.8 Mb) on an autosome, not the sex chromosome. This haplotype was also associated with sex-specific diversity and strong linkage when we examined whole genome sequencing of the natural source population. We hypothesise that colour polymorphism is driven by Y-autosome epistasis, and conclude that predictions of sexual antagonism should focus on incorporating the effects of epistasis in understanding complex adaptive architectures underlying intralocus sexual conflict.

54. Genomic signature of ongoing sexual conflict in red deer
Presenter: Lucy Peters, University of Edinburgh
Co-Authors: Johnston, Susan, University of Edinburgh; Pemberton, Josephine, University of Edinburgh

In species with two sexes, males and females largely share the same genome. Due to differing biology and life histories however the sexes can have divergent fitness optima for the same traits, thus resulting in sexual conflict. The resolution of sexual conflict often leads to the development of dimorphism in phenotypes between the sexes. Here we explore evidence for genomic signatures of sexual conflict in red deer, a species that shows extensive sexual dimorphism. We use evidence for sex specific transmission distortion as an indication of differential selection on parental gametes and/or pre-natal viability. Transmission distortion occurs if a parent is more likely to transmit one allele over the other which can be dependent both on parent and on offspring sex. Our red deer study system benefits both from a multigenerational pedigree to inform the choice of trios used in this analysis as well as extensive genomic marker data from a 50k SNP chip. This analysis was further complemented by determining parental chromosome phase and extensive simulations of random allele inheritance given the pedigree structure. Finally, any regions showing evidence of sex specific allele differences can be linked to reproductive fitness and survival data available for the majority of individuals used in this study. This study gives valuable insight into the management of the genome within a single population of a species with sexes exhibiting very different life history strategies.

55. A single QTL with large effect is associated with female functional virginity in an asexual parasitoid wasp
Presenter: Wen-Juan Ma, University of Kansas
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During the transition from sexual to asexual reproduction, a suite of reproduction-related sexual traits become superfluous, and may be selected against if costly. Female functional virginity refers to asexual

females resisting to mate or not fertilizing eggs after mating. These traits appear to be among the first that evolve during the gradual transition from sexual to asexual reproduction. The genetic basis of female functional virginity remains elusive. Previously, we reported that female functional virginity segregates as a single recessive locus in the asexual parasitoid wasp Asobara japonica. Here, we investigate the genetic basis of this trait by quantitative trait loci (QTL) mapping and candidate gene analyses. Consistent with the segregation of phenotypes, a single QTL of large effect was found spanning over 4.23 Mb and comprising at least 131 protein-coding genes, of which 15 featured sex-biased expression in the related sexual Asobara tabida. We speculate that two of these 15 genes may be of particular interest: CD151 antigen and nuclear pore complex protein Nup50. Overall, our results are consistent with a single gene or a cluster of linked genes underlying rapid evolution of female functional virginity in the transition to asexuality. Once a mutation for rejection to mate has swept through a population, the region comprising the gene(s) does not get smaller due to lack of recombination in asexuals.

56. Sex-specific selection drives the evolution of alternative splicing in birds

Presenter: Thea Rogers, Department of Animal and Plant Sciences, University of Sheffield
Co-Authors: Palmer, Daniela, Department of Animal and Plant Sciences, University of Sheffield, Wright, Alison, Department of Animal and Plant Sciences, University of Sheffield

Males and females in many species are frequently exposed to conflicting selection pressures, yet the sexes share an almost identical set of genes. Gene regulation is widely assumed to resolve these conflicting sex-specific selection pressures, and although there has been considerable focus on elucidating the role of gene expression level in sex-specific adaptation, other regulatory mechanisms have been overlooked. In particular, alternative splicing enables different transcripts to be generated from the same gene, meaning that exons which have sex-specific beneficial effects can in theory be retained in the gene product, while exons with detrimental effects can be skipped. Using male and female transcriptome data, we combined patterns of alternative splicing with population genomic methods to test how sex-specific selection acts on broad patterns of alternative splicing (Rogers et al. in press MBE). We identify hundreds of genes that have sex-specific patterns of splicing, and establish that sex differences in splicing are correlated with phenotypic sex differences. Additionally, we find that alternatively spliced genes have evolved rapidly as a result of sex-specific selection, and suggest that sex differences in splicing offer another route to sex-specific adaptation when gene expression level changes are limited by functional constraints. Overall, our results shed light on how a diverse transcriptional framework can give rise to the evolution of phenotypic sexual dimorphism.

57. Complex relationships and adaptive divergence between Maylandia species of Malawi cichlids

Presenter: Margarita Samborskaia, EMBL EBI, previously Department of Genetics, Cambridge University
Co-Authors: Margarita Samborskaia(1)*, Tyler Linderoth(1)*, Hannes Svardal(2), Martin Genner(3), George Turner(4), Richard Durbin(1,5); Department of Genetics, University of Cambridge, UK; University of Antwerp, Belgium; University of Bristol, Bristol, UK; Bangor University, Bangor, UK; Wellcome Sanger Institute, Cambridge, UK

Using over 300 whole-genome sequences, we characterized the genetic relationship among eight species of the rock-dwelling cichlids in the Maylandia genus from Lake Malawi. We found a clear discrepancy between current species classifications and phylogenetic relationships between M. emmiltos and M. fainzilberi, whereby one population of M. emmiltos is genetically more similar to M. fainzilberi than a neighboring M. emmiltos population. We also discovered that apparent admixture between these species results in males possessing different dorsal fin patterning, a potentially relevant trait for assortative mating. In order to uncover traits and loci playing a prominent role in species formation we scanned the genomes of 102 M. emmiltos and M. fainzilberi to look for regions of aberrant divergence against their similar genomic backgrounds. We discovered 224 SNPs with allele frequency differences between the species that are statistically unlikely to be a consequence of drift alone. Accordingly, 75% of these divergent SNPs fall within regions harboring extensive linkage and/or levels of within and between

species polymorphism consistent with selection. Among 46 genes showing evidence of adaptive divergence, three genes are clearly involved in vision (slc24a2, opn1lw1, opn1sw2) highlighting evolution of the visual system as potentially key to cichlid speciation. We find other developmental genes showing signs of selection that could underlie morphological differences and provide candidates for experimental validation.

58. The limits of evolutionary convergence in sympatry: reproductive interference and developmental constraints leading to local diversity in aposematic signals

Presenter: Maisonneuve Ludovic, Institut de Systématique, Evolution, Biodiversité (ISYEB), Muséum national d'Histoire naturelle, CNRS, Sorbonne Université, EPHE, Université des Antilles CP 50, 57 rue Cuvier, 75005 Paris, France
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Mutualistic interaction between defended species is a striking case of evolutionary convergence in sympatry, driven by protection against predators, brought by mimicry. However, such convergence is often limited: sympatric defended species frequently display different or imperfectly similar warning traits. Convergence in warning trait can involve distantly-related species displaying drastically different traits in allopatry that may affect the level of convergence. Moreover, warning traits are also implied in mate recognition, so that trait convergence might result in heterospecific courtship and mating. Here we investigate the strength and direction of convergence in warning trait in defended species with different ancestral traits, using a mathematical model. We specifically determine the effect of phenotypic distances among ancestral traits of converging species, and costs of heterospecific sexual interactions on imperfect mimicry and trait divergence. Our analytical results confirm that reproductive interference limits the convergence of warning trait, leading to imperfect mimicry or complete divergence. More surprisingly, we show that reproductive interference can change the direction of convergence depending on the relative species density. We also show that reproductive interference can generate imperfect mimicry only between species with different ancestral traits. Our model therefore highlight that convergence triggered by Müllerian mimicry not only depends on relative defense levels, but that relative species densities, heterospecific sexual interactions and ancestral traits interferes in the direction and strength of convergence between species.

59. An investigation of MHC-dependent sexual selection in a free-living population of sheep

Presenter: Wei Huang, Institute of Evolutionary Biology, University of Edinburgh
Co-Authors: Pemberton, Josephine, Institute of Evolutionary Biology, University of Edinburgh

MHC genes are one of the most polymorphic gene clusters in vertebrates and play an essential role in adaptive immunity. There is evidence that sexual selection also plays a role in maintaining MHC diversity, but the specific mechanisms are controversial. In this study, we investigate evidence for non-random MHC-dependent mating patterns in a free-living population of Soay sheep. Using a large number of sheep diplotyped at the MHC class IIa region and genome-wide SNPs, together with field observations of consorts, we found sexual selection against one of eight haplotypes, C, in males at the pre-copulatory stage and sexual selection on female MHC heterozygosity acting in opposition directions at the pre- and post-copulatory stages. We also found disassortative mating at the post-copulatory stage, along with strong evidence of inbreeding avoidance at both stages. However, results from generalized linear mixed models suggest that the pattern of MHC-dependent disassortative mating could be a by-product of inbreeding avoidance. These results suggest that there is selection on the MHC at the pre- and post-

copulatory stages, but that apparent disassortative mating with respect to the MHC may be driven by inbreeding avoidance.

60. A genetic switch for male-limited UV-iridescence in an incipient species pair
Presenter: Joseph Hanly, The George Washington University
Co-Authors: Ficcarotta Vince, GWU; Porter Adam, UMass Amherst; Martin Arnaud, GWU

Sexual dimorphism is widespread in animals, and may be used for mate competition or recognition. Two species of Colias butterflies have come into secondary contact in the eastern United States in the last century due to anthropogenic land use changes. One species, C. eurytheme, exhibits strong male-specific UV iridescence on the ventral wing surface, while its sister species C. philodice does not exhibit any UV reflectance. The presence or absence of UV plays a major role in female mate choice, along with a suite of other phenotypes which were previously hypothesized to be sex linked and which likely act as pre-zygotic barriers to hybridization. We generated F2 and backcross hybrids between the sister taxa, and performed 2bRAD sequencing, allowing us to map the presence or absence of UV to the transcription factor bab, the homolog of which has previously been linked the development and evolution of sexually dimorphic abdominal patterns in Drosophila species. Immunostaining and CRISPR/Cas9 perturbation experiments suggest that bab is an inhibitor of UV, and may have been the target of repeated selection for the presence or absence of this sexually dimorphic trait in the Coliadinae.

61. Origin of a germ-line restricted chromosome via ancient introgression in fungus gnats
Presenter: Kamil S. Jaron, The University of Edinburgh
Co-Authors: Hodson, Christina N., The University of Edinburgh.; Ross, Laura, The University of Edinburgh.

Germline restricted DNA has evolved in diverse animal taxa: in several vertebrate clades, nematodes, and flies. In these lineages, either portions of chromosomes or entire chromosomes are eliminated from somatic cells early in development, restricting portions of the genome to the germline. Little is known about why germline restricted DNA evolves. We investigated the origin of germline restricted chromosomes in the fungus gnat Bradysia tilicola (Diptera; Sciaridae), which carries two large germline restricted chromosomes. We sequenced and assembled the genome of B. tilicola and identified the germline restricted chromosome sequences using coverage and kmer-based techniques. First, we found that the GRC genes are extraordinarily divergent from their within-genome paralogs. Further, phylogenomic analysis with 13 related flies in the same infraorder revealed that more than 70% of GRC genes confidently branch within another Dipteran family (Cecidomyiidae), suggesting that these chromosomes have arisen in Sciaridae through introgression from a related lineage. These results suggest that the germline restricted chromosomes may have evolved through an ancient hybridization event, opening up many questions about how this may have occurred. Using molecular dating using a single amber record for the calibration suggests the hybridization occurred a long time ago (116 - 50 mya) between extremely diverged lineages. The split of the ancestral lineages is estimated to happen 31 - 97 my before hybridization. Furthermore, both these fly families feature paternal genome elimination. The potential link of the peculiar reproduction to the introgression will be discussed.

62. Most transposable element insertions are deleterious and purged from evolving Drosophila populations
Presenter: Anna Maria Langmüller, Institute of Population Genetics, Vetmeduni Vienna
Co-Authors: Nolte, Viola, Institute of Population Genetics, Vetmeduni Vienna. Schlötterer, Christian, Institute of Population Genetics, Vetmeduni Vienna.

Transposable elements (TEs) are selfish genetic elements, which can move autonomously in the host genome. The TE abundance in natural populations is highly biased towards beneficial or neutral TEs, because deleterious insertions are most likely purged from the population, and will not be detected.

Here, we took advantage of experimental evolution to follow TE insertion dynamics in a D. simulans population that is being invaded by the P-element. We used a set of isofemale lines, of which about 35% contained a fresh, naturally invaded P-element. During the maintenance of the lines the P-element copy number increased, reflecting an ongoing P-element activity. Reasoning that the small effective population size of the isofemale lines prevented efficient purging of deleterious P-element insertions, we followed the fate of P-element insertions which occurred in the isofemale lines in large outbred populations. We show that despite the total number of P-element insertions remained constant, the majority of the P-element insertions from the isofemale lines is efficiently purged from the evolving populations and could no longer be detected. Our results are the first empirical demonstration that the majority of P-element insertions were deleterious to the host, but could be efficiently purged in outbred populations. Hence, the majority of the persisting P-element insertions are either beneficial or effectively neutral.

63. Double drives and private alleles for localized population genetic control

Presenter: Katie Willis , Imperial College London
Co-Authors: Burt, Austin, Imperial College London

Synthetic gene drive constructs could provide highly efficient interventions to control disease vectors and other pests. This efficiency derives in part from leveraging natural dispersal and gene flow to spread the construct and its impacts from one population to another. However, sometimes only specific populations are in need of control. Since many gene drive designs use sequence-specific nucleases, one way to restrict their spread would be to exploit sequence differences between target and non-target populations. We propose and model a series of low threshold double drive designs for population suppression, each consisting of two constructs, one imposing a reproductive load on the population and the other inserted into a differentiated locus and controlling the drive of the first. Simple, deterministic, discrete-generation simulations are used to assess the alternative designs. We find that the simplest double drive designs are more robust to pre-existing resistance at the differentiated locus than single drive designs, and that more complex designs incorporating sex ratio distortion can be more efficient still, even allowing for successful control when the differentiated locus is neutral and there is up to 50% pre-existing resistance in the target population. Similar designs can also be used for population replacement. Double drives should be considered when efficient but localized population control is needed and there is some genetic differentiation between target and non-target populations. To assess the potential of double drives for localized control of the malaria transmitting mosquito, Anopheles gambiae, we screened over 1000 whole genome sequences identifying putative differentiated target sites.

64. Quantitative genetics of stress responses vary with the intensity of environmental perturbation

Presenter: Pedram Samani, University College London
Co-Authors: Saintain, Q., University College London; Speed, D., University College London; Jeffares, D., University of York ; Bähler, J., University College London; Reuter, M., University College London

Evolutionary rescue (ER) occurs when populations rely on adaptation to escape decline and extinction due to large, abrupt environmental shifts. Understanding the factors that allow or limit ER is fundamental for our ability to conserve populations of threatened species, as well as to manage responses to antibiotics or pesticides. The capacity of populations to show ER depends critically on the amount of genetic variation in relevant traits that is available for selection to act on. In complex environments, evolutionary responses will further be shaped by genetic correlations between traits that allow organisms to respond to specific environmental stressors. Here, we investigated how the quantitative genetics of stress responses vary with the intensity of environmental perturbation, assaying growth of wild isolates of fission yeast (Schizosaccharomyces pombe) under 8 stressors (NaCl, LiCl, CaCl2, KCl, high temperature, H2O2, TBH, MSB). We used a combination of high throughput phenotyping, quantitative genetics and genome-wide association mapping to infer the genetic architecture of environmental responses. Our results indicate that the genetic architecture of stress responses varies with the intensity

of stress. Genetic covariations of stress response were overwhelmingly positive among highly stressful conditions, but the genetic architecture was more complex, with both positive and negative correlations among stress responses, at medium intensity. We interpret these changes in the genetic architecture in the light of our current understanding of core and specific stress response pathways, as well as genetic mapping of functional polymorphisms in our experimental strains.

Session 3

65. Maintenance of adaptive dynamics and no detectable load in a range-edge out-crossing plant population

Presenter: Margarita Takou, University of Cologne
Co-Authors: de Meaux, Juliette, University of Cologne

During range expansion, edge populations are expected to face increased genetic drift. The increased genetic drift could compromise adaptive dynamics, preventing the removal of deleterious mutations and slowing down adaptation. Here, we contrast populations of the European sub-species Arabidopsis lyrata ssp petraea, which expanded its Northern range after the last glaciation. We detect a strong bottleneck in the range-edge population and document a 4.9% excess of derived non-synonymous variants per individual in the range-edge population, which suggests an increase of the genomic burden of deleterious mutations. However, a small net decrease in per-individual fitness is predicted by the demographic history of the range-edge population. Consistent with this prediction, the range-edge population is not impaired in its growth measured in a common garden experiment. We further observe that the allelic diversity at the self-incompatibility locus has remained unchanged. Thus, strict outcrossing is ensured, which evolves under negative frequency-dependent selection. Furthermore, genomic footprints indicative of selective sweeps are broader in the Northern population but not less frequent than the core population. We conclude that the outcrossing species A. lyrata ssp petraea shows a strong resilience to the effect of range expansion.

66. The load of deleterious mutations sheltered by self-incompatibility alleles in Arabidopsis

Presenter: Audrey LE VEVE, Laboratoire Evolution, Ecologie, Paléontologie, UMR 8198 CNRS-Université de Lille
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Self-incompatibility (SI) is a genetic system preventing selfing in hermaphroditic plant species. In Brassicaceae, the sporophytic SI system is controlled by a genomic region, the S-locus, and is a classical case of a particular type of balancing selection (negative frequency-dependent selection). SI enforces heterozygosity and promotes allelic diversity at the SI genes themselves, but also at the linked genomic region surrounding the S-locus. Because of enforced heterozygosity, SI is expected to result in the effective sheltering of recessive deleterious mutations. However, direct experimental evidence for this load have remained scarce. Here, we test the hypothesis that balancing selection has counteracted purifying selection in genomic regions physically linked to S locus by allowing the maintenance of deleterious variants. We focused on natural populations of two self-incompatible species, A. halleri and A. lyrata. We resequenced the genomic regions linked to the S-locus and a series of random genomic regions to identify segregating mutations and evaluate their potential deleterious effect based on conservation and functional annotation. We find a higher density of deleterious mutations in S-locus flanking than in control genes. However, contrary to theoretical expectations and opposite to results in mammalian MHC, these deleterious mutations segregated at lower population frequencies around the S locus as compared to the genomic background. Interestingly, the signal detected varied across natural

populations and across species, raising the possibility that differences in their demographic history may be an under-appreciated factor of the accumulation of the sheltered load.

67. Dissecting the evolutionary mechanisms driving Alpine whitefish diversification

Presenter: Rishi De-Kayne, University of Edinburgh/University of Bern/Eawag
Co-Authors: Selz, Oliver M. (Eawag), Seehausen, Ole, (Eawag/University of Bern), Feulner, Philine G. D., (Eawag/University of Bern)

The recent origin and repeated diversification of salmonids in post-glacial lakes make them an ideal system in which to study fundamental questions relating to genome evolution underpinning ecological diversification and speciation. Within the Alpine whitefish (Coregonus spp.) species complex, the independent diversification into six or more sympatric whitefish species in multiple lakes across Switzerland are thought to have been facilitated by ecological and reproductive strategy differentiation with sympatric species varying in diet, body length, gill-raker number, and spawning depth and time. Using a large whole-genome resequencing dataset including 96 whitefish individuals spanning 21 species from five independent lake systems, we investigated the evolutionary mechanisms that have driven Alpine whitefish diversification within Switzerland. Our phylogenetic approach indicates that diversification of ecologically distinct species has occurred within lake systems, as sympatric species are largely each other's closest relatives. However, rare pelagic species show evidence of contemporary introgression between lake systems, likely as a result of the anthropogenic movement of fish between lake systems. Across lake systems, the genomes of the most common whitefish ecomorphs are characterised by parallel differentiation in a number of regions along the length of the genome. This suggests that the postglacial whitefish diversification was facilitated by wide-spread standing genetic variation enabling the rapid built up of genome-wide differentiation, additionally fuelled by admixture-derived variation.

68. Overlapping gene expression changes and correlated expression shifts characterise the repeated evolution of zinc-tolerance in Silene uniflora.

Presenter: Daniel P Wood, Bangor University
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Recent anthropogenic changes have prompted rapid local adaptation across many taxa, but the extent to which these are associated with repeatable genetic changes are unclear. The coastal perennial Silene uniflora has recently colonised abandoned mines in British Isles whose soils contain high levels of toxic heavy metals, forming zinc-tolerant ecotypes. Genetic data supports multiple independent colonisation events, with little evidence of gene flow between the zinc-tolerant populations. To investigate the extent to which the mechanisms of zinc tolerance are shared across repeated origins of the trait, we performed RNA-seq on roots of two independently originating zinc-tolerant populations, and their respective non-tolerant sister populations, in both control and zinc-contaminated solutions. We identified a number of shared changes in gene expression in the two tolerant populations in the absence of zinc, as well as a largely nonoverlapping set of genes showing an expression response to zinc in tolerant populations. We also investigated the extent to which these genes were overrepresented in the subset of genes undergoing positive selection in the zinc-tolerant populations. Our results show that zinc-tolerance in independently colonising populations of S. uniflora on mines is associated with highly convergent changes in gene expression, both in the presence and absence of the zinc stressor. This suggests that the mechanistic basis of local adaptation can be highly repeatable, even over very short timescales.

69. Genetic variation for adaptive traits is associated with polymorphic inversions in Littorina saxatilis

Presenter: Eva Koch, University of Sheffield
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Chromosomal inversions have long been recognized for their role in local adaptation. By suppressing recombination in heterozygous individuals, they can maintain co-adapted gene complexes and protect them from homogenizing effects of gene flow. However, to fully understand their importance for local adaptation we need to know their influence on phenotypes under divergent selection. For this, the marine snail Littorina saxatilis provides an ideal study system. Divergent ecotypes adapted to wave action and crab predation occur in close proximity on intertidal shores with gene flow between them. Here, we used F2 individuals obtained from crosses between the two ecotypes to test for associations between genomic regions and traits distinguishing the Crab/Wave ecotypes, including size, shape, shell thickness and behaviour. We show that most of these traits are influenced by previously detected inversion regions that are divergent between ecotypes. We thus gain a better understanding of one important underlying mechanism responsible for the rapid and repeated formation of ecotypes: divergent selection acting on inversions. We also found that some inversions contributed to more than one trait suggesting that they may contain several loci involved in adaptation, consistent with the hypothesis that suppression of recombination within inversions facilitates differentiation in the presence of gene flow.

70. Genomic evidence for inbreeding depression and purging of deleterious genetic variation in Indian tigers
Presenter: Anubhab Khan, University of Glasgow
Co-Authors: None

Increasing anthropogenic habitat fragmentation leads to wild populations of endangered species becoming small, isolated, and threatened by inbreeding depression. However, small populations may be able to purge recessive deleterious alleles as they become expressed in homozygotes, thus reducing inbreeding depression and increasing population viability. We used genome sequencing of 57 tigers to estimate individual inbreeding and mutation loads in a small-isolated, and two large-connected populations in India. As expected, the small-isolated population had substantially higher average genomic inbreeding (FROH=0.57) than the large-connected (FROH=0.35 and FROH=0.46) populations. Surprisingly, the small-isolated population had the lowest loss-of-function mutation load, likely due to high inbreeding leading to purging of highly deleterious recessive mutations. While fewer, the loss-of-function alleles in the small-isolated population tended to have higher frequencies and homozygosity than in the large populations, suggestive of elevated inbreeding depression here despite apparent purging of some highly deleterious loss-of-function mutations. The large populations had lower missense mutation loads than the small-isolated population, but were not identical, possibly due to differential historical founder effects and bottlenecks. Frequency distributions of damaging and neutral alleles uncover genomic evidence that purifying selection has removed part of the mutation load across India. Together, these results provide the strongest genomic evidence for efficient purifying selection in both small and large populations, as well as strong fitness cost to increased inbreeding. Finally, we establish a novel paradigm for genetic rescue and propose source populations be prioritized based on differentiation at loci with frequent deleterious alleles, rather than traditional measures of genome-wide differentiation.

71. History or demography? Determining the drivers of genetic variation in North American plants
Presenter: Julia López Delgado , Leiden University; Institute for Biodiversity and Ecosystem Dynamics (University of Amsterdam)

Co-Authors: Meirmans, Patrick, Institute for Biodiversity and Ecosystem Dynamics (University of Amsterdam)

Understanding the impact of historical and demographic processes on genetic variation is essential for devising conservation strategies and predicting responses to climate change. Recolonization after Pleistocene glaciations is expected to leave distinct genetic signatures, characterised by lower genetic diversity in previously glaciated regions. Populations' positions within species ranges also shape genetic variation, following the central-marginal paradigm dictating that peripheral populations are depauperate, sparse and isolated. However, the general applicability of these patterns and relative importance of historical and demographic factors remains unknown. Here, we analysed the distribution of genetic variation in 91 native species of North American plants by coupling microsatellite data and Species Distribution Modelling. We tested the contributions of historical climatic shifts and the central-marginal hypothesis on genetic diversity and structure. Decreased diversity was found with increased distance from potential glacial refugia, coinciding with the expected make-up of postglacially colonised localities. At the range periphery, lower genetic diversity, higher inbreeding levels and genetic differentiation were reported, following the assumptions of the central-marginal hypothesis. History and demography were found to have approximately equal importance in shaping genetic variation.

72. A shared regulatory allele of Agouti contributes to parallel evolution of cryptically colored beach mice

Presenter: Brock Wooldridge, Harvard University
Co-Authors: Kautt, Andreas; McFadden, Sade; Domingues, Vera; Hoekstra, Hopi

Despite the many examples of parallel evolution1, there are few cases in which both the genetic basis and evolutionary history of these repeated traits are understood. The oldfield mouse, Peromyscus polionotus, is widespread across the southeastern United States, where they exhibit considerable coat-color variation as an adaptation against avian predation2. Their dorsal coats range from dark brown in mainland habitat to a near white appearance that has evolved independently on the white sand beaches of Florida's Gulf and Atlantic coasts3. Here, we focus on a phenotypically variable population of oldfield mice, P. p. albifrons, which, by using a new chromosome-level genome assembly and resequencing data, we are able to show is a sister group to dark-colored mainland mice and a likely source population for the pale Gulf Coast beach mice. By characterizing 15 pigmentation traits in 160 albifrons mice, we find that the extent of pigmentation is associated with a ~2kb noncoding region upstream of the pigmentation gene Agouti. Extended tracts of homozygosity in the Agouti haplotype associated with light pigmentation suggest that light pigmentation has experienced recent and strong positive selection. Finally, this same light haplotype is fixed in both Gulf Coast and Atlantic beach mice although these populations are separated by >3,000km. Given the evolutionary history of this clade, these results suggest that this allele has been maintained in the mainland populations and repeatedly spread to beach mice, thereby facilitating their rapid and parallel evolution.

73. The Fast Mutation Rate of the Nematode Strongyloides ratti

Presenter: Rob Morris, University of Liverpool
Co-Authors: Mark Viney, University of Liverpool

Strongyloides ratti is a parasitic nematode of rats that is common in wild rat populations in the UK. Its life cycle has an obligate parthenogenetic parasitic phase, and a facultative free-living sexual phase. We have calculated the mutation rate in the 42 Mbp S. ratti genome, using long-term, laboratory maintained parasite lines. We find that the mutation rate is in the range of 7 x 10-6 to 7 x 10-7 base substitutions per site per generation, with this range depending on the strictness with which we identify SNPs. This S. ratti rate is approximately two orders of magnitude higher than that of another nematode, Caenorhabditis elegans, whose rate is 2.7 – 9 x 10-9.

74. Ancient and modern stickleback genomes reveal the demographic constraints on adaptation
Presenter: Andrew Foote, Department of Natural History, Norwegian University of Science and Technology (NTNU), University Museum, 7491 Trondheim, Norway
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Adaptation is typically studied by comparing modern populations from contrasting environments. Individuals persisting in the ancestral habitat are typically used to represent the ancestral founding population, however, it has been questioned whether these individuals are good proxies for the actual ancestors. To address this, we applied a paleogenomics approach to directly access the ancestral genepool, partially sequencing the genomes of two 11-13,000-year-old stickleback recovered from the transitionary layer between marine and freshwater sediments of two Norwegian isolation lakes, and comparing them with 30 modern stickleback genomes from the same lakes and adjacent marine fjord, in addition to a global dataset of 20 genomes. The ancient stickleback shared genome-wide ancestry with the modern fjord population, whereas modern lake populations have lost substantial ancestral variation following founder effects, and associated drift and selection. Freshwater-adaptive alleles found in one ancient stickleback genome have not risen to high frequency in the present-day population from the same lake. Comparison to the global dataset suggested incomplete adaptation to freshwater in our modern lake populations. Our findings reveal the impact of population bottlenecks in constraining adaptation due to reduced efficacy of selection on standing variation present in founder populations.

75. A genome wide analysis of the Pleistocene black bear from ancient cave sediment DNA
Presenter: Bianca De Sanctis, Department of Genetics, University of Cambridge, Cambridge UK.
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Ancient genomics have revolutionized our understanding of phylogenetics and population dynamics of past species. The field is criticized, however, for causing permanent damage to precious fossil remains. Here, we show it is possible to obtain genome scale sequence data of past mammalian species directly from ancient sediments. We obtain ancient DNA from the black bear, Ursus americanus, from layers of the Chiquihuite cave in northern Mexico, dating to 16 and 13.5 thousand years before present (ka BP). By comparing these samples to a reference panel of 83 modern black bears throughout North America, we can begin to illuminate the complex demographic history of the black bear. Our findings represent a step towards bringing ancient environmental DNA into the genomic era and hold promise to eventually make ancient genomics independent of fossil remains.

76. Low Spontaneous Mutation Rate and Pleistocene Radiation of Pea Aphids
Presenter: Varvara Fazalova, University of Oxford, Department of Zoology
Co-Authors: Nevado, Bruno, CE3C – Faculdade de Ciências, Universidade de Lisboa

Accurate estimates of divergence times are essential to understand the evolutionary history of species. It allows linking evolutionary histories of the diverging lineages with past geological, climatic, and other changes in environment and shed light on the processes involved in speciation. The pea aphid radiation

includes multiple host races adapted to different legume host plants. It is thought that diversification in this system occurred very recently, over the past 8,000–16,000 years. This young age estimate was used to link diversification in pea aphids to the onset of human agriculture, and led to the establishment of the pea aphid radiation as a model system in the study of speciation with gene flow. We re-examine the age of the pea aphid radiation, by combining a mutation accumulation experiment with a genome-wide estimate of divergence between distantly related pea aphid host races. We estimate the spontaneous mutation rate for pea aphids as 2.7×10^−10 per haploid genome per parthenogenetic generation. Using this estimate of mutation rate and the genome-wide genetic differentiation observed between pea aphid host races, we show that the pea aphid radiation is much more ancient than assumed previously, predating Neolithic agriculture by several hundreds of thousands of years. Our results rule out human agriculture as the driver of diversification of the pea aphid radiation, and call for re-assessment of the role of allopatric isolation during Pleistocene climatic oscillations in divergence of the pea aphid complex.

Session 4

77. It didn't happen yesterday: selective sweeps in genealogical time.
Presenter: Gertjan Bisschop, University of Edinburgh
Co-Authors: Lohse, Konrad, University of Edinburgh; Setter, Derek, University of Edinburgh

Current methods of identifying signatures of hard selective sweeps in the genome are limited in two key ways: i) selection is assumed to have occurred immediately prior to sampling the population and ii) the analytic predictions are generally limited to expectations for very drastic summaries of single-nucleotide polymorphisms. We present a tractable method of describing the effect of a selective sweep on the genealogical histories in the surrounding genome. We explicitly model both the timing and the selection coefficient of the adaptive event. In addition, this framework allows us to go beyond expected coalescence times and the site frequency spectrum and to derive the full distribution of genealogical histories. We use these analytic results to develop a simple composite likelihood inference framework that leverages information contained in the joint distribution of closely linked variants. We use simulations to show that our analytic framework has high power to identify historically adaptive regions of the genome and unlike sweep-scans based on the SFS can correctly co-estimate the strength and timing of selection.

78. Assembly-free quantification of organellar pseudogenes
Presenter: Hannes Becher, University of Edinburgh
Co-Authors: Nichols, Richard A, Queen Mary University of London

Nuclear pseudogenes of organellar origin (NUMTs and NUPTs) are commonly treated as a problem, since they can mislead or obscure our reading of true organellar sequence in many species. We argue for a more positive view – that they are fascinating in their own right and provide valuable information for studies of hybridisation and the evolution of organellar genomes. The assembly-free identification and quantification for such pseudogenes is challenging. Here, we propose a novel statistical method to quantify the nuclear proportion of organellar pseudogenes based on low-coverage population sequencing data. We apply our method to data from the grasshopper Podisma pedestris, which has a hybrid zone in the French Alps. In addition to quantifying mitochondrial pseudogenes, we find mitochondrial divergence between the hybridising sub-populations, but little signs of differentiation between their pseudogenes.

79. A unified genealogy of modern and ancient genomes
Presenter: Anthony Wilder Wohns, University of Oxford. Broad Institute of MIT and Harvard.
Co-Authors: Wong, Yan, University of Oxford. Jeffery, Ben, University of Oxford. Akbari, Ali, Harvard University. Patterson, Nick, Harvard University. Reich, David, Harvard University. Kelleher, Jerome, University of Oxford. McVean, Gil, University of Oxford.

The sequencing of modern and ancient genomes from around the world has revolutionised our understanding of human history and evolution. However, the general problem of how best to characterise the full complexity of ancestral relationships from the totality of human genomic variation remains unsolved. Patterns of variation in each data set are typically analysed independently, and often using parametric models or data reduction techniques that cannot capture the full complexity of human ancestry. Moreover, variation in sequencing technology, data quality and in silico processing, coupled with complexities of data scale, limit the ability to integrate data sources. Here, we introduce a non-parametric approach to inferring human genealogical history that overcomes many of these challenges and enables us to build the largest dated genealogy of both modern and ancient humans yet constructed. The genealogy provides a lossless and compact representation of multiple datasets, addresses the challenges of missing and erroneous data, and benefits from using ancient samples to constrain and date relationships. Using simulations and empirical analyses, we demonstrate the power of the method to recover relationships between individuals and populations, as well as to identify the descendants of ancient samples. Finally, we show how applying a simple nonparametric estimator of ancestor geographical location to the inferred genealogy recapitulates key events in human history. Our results demonstrate that whole-genome genealogies are a powerful means of synthesising genetic data and contain rich insight into human evolution.

80. Unravelling the determinants of the molecular adaptive rate within genomes and between species
Presenter: Ana Filipa Moutinho, University of Sussex
Co-Authors: Galtier, Nicolas, University of Montpellier; Eyre-Walker, Adam, University of Sussex; Dutheil, Julien, Max Planck Institute for Evolutionary Biology

The frequency and nature of adaptive mutations are widely heterogeneous between species. For instance, fruit flies and wild mice exhibit a much higher adaptive substitution rate than primates and plants. What determines this variation is, however, not fully understood. Over the years, 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 have shown that this rate not only varies between genes but also at the intra-genic level, with the relative solvent accessibility acting as a major determinant of protein adaptive evolution. This effect, however, varied in strength between species, suggesting that the variation within genes may help explain inter-species patterns of adaptation. Here, we use a comparative population genomics approach across (1) multiple species with distinct life-history traits, (2) genes with different functional categories and age, and (3) sites with different structural properties. We fit models of distributions of fitness effects to estimate the rate of adaptive substitutions at the protein and amino-acid residue level. This approach allows us to evaluate the relative importance of several drivers of the rate of adaptive evolution at distinct organisational levels and evolutionary scales. Our study further emphasises the importance of using intramolecular variation to shed light on the molecular basis of adaptation.

81. Estimating Nm
Presenter: Daria Shipilina, 1. IST Austria 2. Department of Ecology and Genetics, Uppsala University
Co-Authors: Barton Nicholas, IST Austria

How best to estimate the number of migrants in a subdivided population, Nm ? The fixation index, Fst, traditionally used to estimate Nm, has serious limitations, as it discards most genealogical information. More recent methods for estimating Nm fit models of isolation with migration, using either the site frequency spectrum (SFS) or blockwise likelihoods or use Approximate Bayesian computation approach (ABC). The key problem is that although an abundance of genomic data allows us to distinguish between specified models, we do not know how to explore the vast space of possible population structures. In this work, we compare Fst with more elaborate methods of Nm estimation. To this end, we benchmark the performance of various Nm estimators against data simulated with a simple population structure, interpreting and comparing the performance of more complex models. We focus on a single non-

recombining locus under the infinite sites model, and ask how best to estimate Nm under the island model, assuming very many demes. We introduce a new estimator, based on the haplotype structure within each deme, which captures population structure in the recent past. We show that for the island model, information about Nm is completely contained in the numbers of distinct haplotypes. We further demonstrate that this haplotype-based estimator is a robust estimator of the recent Nm, when fitted to different population structures.

82. Bait-ER: fishing for selection in a pool of drifting alleles

Presenter: Carolina Barata, University of St Andrews
Co-Authors: Borges, Rui, Institute of Population Genetics, Vetmeduni Vienna; Kosiol, Carolin, Centre for Biological Diversity, University of St Andrews

Researchers have been testing evolutionary theory predictions for tens if not hundreds of generations in the lab. However, most laboratory populations are small, such that genetic drift plays a substantial role in determining the fate of polymorphic variation. Consequently, estimating and testing for selection genome-wide proves challenging. Despite numerous efforts to investigate allele frequency changes in lab experiments, methods for detecting selection still suffer from high false positive rates and low statistical power. Some others do not even consider replicate populations. For that, we have developed a fully Bayesian approach to estimate selection coefficients aimed at time series allele frequency datasets. Our method is based on the Moran model of nucleotide evolution which allows for overlapping generations. This is a key feature because many experimental evolution studies do not have discrete generations. One other crucial advantage to our Bayesian approach is that we can consider the variance of each estimate and not only its average. This allows for a full statistical analysis of each candidate locus. Furthermore, Bait-ER accounts for consistency across replicates whilst considering noise caused by pooled sequencing. We tested our method's performance quite extensively using simulated data. It shows high accuracy in difficult demographic scenarios as well as in other more complex scenarios involving recombination rate variation and hitchhiking. Lastly, we tested Bait-ER on a recently published experiment of Drosophila simulans populations in the laboratory. Bait-ER produces a few very significant peaks throughout the genome. These can very well correspond to major effect loci that contribute to adaptation of complex polygenic traits.

83. Modelling the spatiotemporal spread of beneficial alleles using low-coverage ancient genomes

Presenter: Rasa Audange Muktupavela, University of Copenhagen
Co-Authors: Korneliussen Thorfinn, University of Copenhagen; Novembre John, University of Chicago; Racimo Fernando, University of Copenhagen

Ancient genome sequencing technologies now provide the opportunity to study natural selection with unprecedented detail. Rather than relying on indirect footprints left by selection on present-day genomes, we can directly observe whether a given allele was present or absent in a particular region of the world at almost any period of human history within the last 10,000 years. Methods for studying selection, however, often rely on grouping individuals, time stretches or sections of a map into discrete units. These approaches often fail to account for the fact that selection is a spatiotemporal process occurring on a landscape, which limits our ability to perform inference about parameters of interest, like the geographic origin of a mutation. Here, we extend a previously developed framework for inferring the spread of beneficial alleles on present-day data using two-dimensional partial differential equations. Under this framework, we can now handle time-stamped ancient samples, as well as genotype likelihoods from low-coverage genomes. We apply the new method to a panel of published ancient West Eurasian genomes and provide dynamic maps showcasing the spread of candidate beneficial alleles over time and space. We also provide estimates for the geographic origin of the mutation, strength of selection and diffusion rate for each of these alleles.

84. The Demes standard for demographic models: because '-I 3 10 4 1 -ma x 1.0 2.0 3.0 x 4.0 5.06.0 x' hurts!

Presenter: Jerome Kelleher, University of Oxford
Co-Authors: Gower, Graham, Copenhagen; Ragsdale, Aaron, McGill; Thornton, Kevin, UC Irvine

As our data improves, the demographic models that we estimate are becoming increasingly complex, often with tens of populations and hundreds of parameters. Yet there is no standard for how these models should be communicated: simulators and inference methods use different and incompatible methods of defining their input and output. The closest thing to a standard is the syntax used to define demographic models in the classical ms program, which while concise, is not easy for humans to comprehend. We propose the ""Demes"" standard for interchanging demographic models in population genetics. Based on mature technologies such as JSON and YAML, this well-defined model is a good balance between being easily understood by both humans and computers. As part of the work of the PopSim consortium, we hope that this approach can be adopted widely, leading to fewer errors and improved reproducibility.

85. Inferring population history using whole-genome data

Presenter: Rita Rasteiro, School of Biological Sciences, University of Bristol, Bristol, Uk
Co-Authors: Bridle, Jon, Genetics, Evolution & Environment, Div of Biosciences, University College London, UK; Beaumont, Mark, School of Biological Sciences, University of Bristol, Bristol, UK

Several computational approaches have been introduced into population genetics, with the aim of providing improved inference of demographic history from genome-level samples. The use of approximate Bayesian computation (ABC) for whole-genome analysis can be challenging particularly when considering the scale of simulation needed. However, there are potential advantages in using ABC when complex models are considered, including those that model the interplay of natural selection and demographic inference. We introduce a model-based simulation methodology to study evolutionary adaptation and demography from whole-genome data. It incorporates both coalescent and forward simulations, where we jointly model selection and allow effective population size fluctuations through time, and Expectation Propagation ABC (EP-ABC). EP-ABC makes use of advances in machine-learning and a “divide to conquer” approach to enable efficient distributed computation in genomic analysis. Finally, we apply our approach to study the population history of a cichlid population from Tanzania.

86. Statistical models for the genetic characterization of honeybee colonies from whole genome pool sequencing data

Presenter: Eynard Sonia, GenPhySE, INRAE, Université Toulouse, INPT, INP-ENVT, Castanet Tolosan, France; UMT PrADE, INRAE, Avignon, France
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Characterizing the genetic diversity of populations allows to better understand their demographic history and their adaptation to selective pressures. Social insects, such as honeybee, live in colonies which ultimately are the relevant evolutionary and selective units for such species. However, performing large scale genetic analyses of honeybees is a real challenge, due to the specific reproduction mechanism including multi-male insemination, making the genotype of a bee colony a mixture of contribution from the queen and the mating drones. To fully describe a colony it is thus necessary to separate queen genotype from allele frequencies in the mating drones. In this work we propose an approach to characterize the genotype of the honeybee queen from pool sequencing of workers from the colony. We introduce statistical models for the analysis of pool sequence data allowing to reconstruct individual queen genotypes. Using simulations we developed a theoretical reconstruction process suiting a large range of colonies and population genetic compositions. We validated this approach using real data and finally applied our procedure to data coming from a field study. We propose to first describe the colony genetic composition, in terms of major European subspecies. From these genetic compositions it is then possible to group colonies in a homogeneous population making possible queen genotype reconstruction using across-colony information. In addition to a better understanding of the population dynamics of honeybees, our approach promises to facilitate the genetic analysis of complex traits, and can be used for genome wide association studies on phenotypes of interests.

87. Parsimonious reconstruction of genealogies and recombination detection for SARS-CoV-2

Presenter: Anastasia Ignatieva, University of Warwick
Co-Authors: Hein, Jotun, University of Oxford; Jenkins, Paul A., University of Warwick

The processes of genetic mutation and recombination are fundamental drivers of viral evolution. Mutation events induce small changes within the genome during replication, which are often visible in sequencing data. Recombination, on the other hand, occurs when the same cell is co-infected by two different strains, whose genetic material is mixed together before being passed on to an offspring viral particle - now potentially possessing very different pathogenic properties. The detection of recombination events from a sample of genetic data is a very challenging problem. I will discuss the combinatorial, algorithmic and statistical aspects of reconstructing possible genealogical histories of a given dataset, and present an application to detecting recombination within SARS-CoV-2 sequencing data.

88. Avoiding ascertainment bias using haplotype based methods

Presenter: Angeles de Cara, CIBIO, Universidade do Porto
Co-Authors: Perez-Pardal, Lucia (CIBIO); Khalatbari, Leili (CIBIO); Yusefi, Hosein (CIBIO); Beja-Pereira, Albano (CIBIO)

The development of single nucleotide polymorphims arrays has been crucial in our understanding of human and livestock population diversity. SNP arrays are thus a great resource, considerably cheaper than whole genome sequencing. It is however important to carefully describe how the array has been developed, which individuals have been used or which markers are included. These is because these arrays are developed to provide the most information about those markers, which may be related to genes of particular interest. Thus, they are not random markers from the genome, and their site frequency spectrum is not necessarily neutral. In humans, in order to make demographic inferences from many individuals, the Human Origins array was developed using 13 panels of SNPs. This has allowed other researchers to develop corrections by taking into account the design. However, when using other panels like those developed for domestic species into their wild counterpart, we need to take great care. We will show here an example of how to avoid the ascertainment bias when using data from a SNP array developed for domestic animals into their wild ancestor, by combining haplotype-based methods. This will allow us to make demographic inferences and show recent bottlenecks and expansions in some populations.

Introduction to the NEOF facility

www.neof.org.uk

NEOF is a new £13m facility funded by the Natural Environmental Research Council (NERC) to provide expert advice, training and state-of-the-art instrumentation to allow environmental and evolutionary biologists to address new questions using omic methods. Our aim is guide users from all levels through experimental design, sample preparation, data generation and analysis, coupled with training in our visitor facility, workshops and online courses. We also provide pilot funding to test adventurous ideas and to introduce and train new users. We will announce the first pilot round directed at early career researchers. In addition to the latest genomic platforms (Illumina Novaseq, PacbIo Sequel II, 10X, etc) we also wish to broaden the scope of omics methods in the population genetics community through proteomics and metabolomics. Profs Steve Paterson (University of Liverpool) and Terry Burke (University of Sheffield) will outline the opportunities that NEOF can provide to population genetic researchers, and members of the NEOF team will be on hand during poster sessions for potential project discussion.

Jan 6th, 2021

Session 1

89. Reproductive isolation among lineages of Silene nutans: a potential involvement of plastid-nuclear incompatibilities

Presenter: Zoé POSTEL, University of Lille
Co-Authors: POUX, Céline, Univ. Lille; GALLINA, Sophie, Univ Lille; VARRE, Stéphane, Univ Lille; GODE, Cécile, Univ Lille ; SCHMITT, Eric, Univ Lille; VAN ROSSUM, Fabienne, Meise Botanic Garden,; TOUZET, Pascal, Univ Lille

How species arise and maintain is one of the main questions in evolutionary biology. Speciation often occurs through the emergence of pre- and post-zygotic barriers to reproduction, leading to reproductive isolation. The emergence of post-zygotic barriers can be the result of genetics incompatibilities and notably cytonuclear incompatibilities leading to hybrid breakdown, through the combination of mismatched nuclear and cytoplasmic gene products in hybrids. Many plastid protein complexes include subunits encoded by plastid and nuclear genomes leading to coevolution between the plastid and nuclear genes encoding these complexes. This coevolution will be disrupted among individuals combining plastid and nuclear genome coming from different species or divergent populations, leading to incompatibilities between plastid and nuclear genes and ultimately to reduced hybrid fitness. In Silene nutans (Caryophyllaceae), four lineages genetically differentiated have been identified and show strong and asymmetric reproductive isolation (high proportion of seedling mortality and chlorotic hybrids), suggesting the presence of plastid-nuclear incompatibilities between these four lineages. We tried to identify plastid and nuclear genes encoding subunits of plastid proteins complexes, using gene-capture methodology and RNAseq, by searching over most of the plastid genes and their potential nuclear interactors, the non-synonymous substitutions fixed differently between lineages. The search for signatures of positive selection on these variants revealed a potential involvement of genes involved in plastid gene expression but also in the photosynthetic machineries.

90. Human endogenous retroviruses in cancer.
Presenter: Michal Izydorczyk, Oxford Brookes University
Co-Authors: Kanda, Ravinder K, Oxford Brookes University

Human endogenous retroviruses (HERVs) are remnants of past retroviral infections, that have inserted into the human genome and are inherited by the progeny. There are many families of these retroviruses, the youngest of which, HERV-K, is speculated to be still active in modern humans, due to observed insertional polymorphism between individuals. HERV-K has also been implicated in a number of diseases, including various cancers. A number of studies have reported increased HERV-K expression in various cancer tissues and implied a possible role for HERV-K in cancer development, although to date there is no direct evidence of that activity – all known HERV-K insertions are thought to be be non-functional (inactivated by mutations). We queried a number of genomes from different cancer datasets (available from the TCGA database), to look for novel HERV-K insertions. We have identified a number of cancer-specific HERV-K insertions, that have not been previously described. We discuss our results.

91. Aneuploidy prevalence differs among Saccharomyces cerevisiae genetic backgrounds
Presenter: Eduardo Scopel, University of Georgia
Co-Authors: Hose, James, University of Wisconsin-Madison; Bensasson, Douda, University of Georgia; Gasch, Audrey, University of Wisconsin-Madison

Individuals carrying an aberrant number of chromosomes can vary widely in their expression of aneuploidy phenotypes. A major unanswered question is the degree to which an individual's genetic makeup influences its response to karyotypic imbalance. Here we took a population-genetics perspective to investigate the selective forces influencing aneuploidy prevalence in Saccharomyces cerevisiae populations as a model for eukaryotic biology. We analyzed genotypic variation recently published for over 1,000 Saccharomyces cerevisiae strains spanning dozens of genetically defined clades and ecological associations. Our results show that the prevalence of chromosome gain and loss varies by clade and can be better explained by differences in genetic background than ecology. The phylogenetic context of lineages showing high aneuploidy rates suggests that increased aneuploidy frequency arose multiple times in S. cerevisiae evolution. These results have important implications for understanding genetic variation in aneuploidy prevalence in health, disease, and evolution.

92. Variation in the effective population size across the human genome
Presenter: Adam Eyre-Walker, University of Sussex
Co-Authors: Soni, Vivak, University of Sussex

The level of genetic diversity varies across a genome; this is thought to be due to three or four factors – variation in the mutation rate, variation in the effective population size (Ne), the stochastic nature of the evolutionary process and the direct effects of natural selection. Attempts to estimate the distribution of Ne across the human genome have suggested that the variation is modest but a precise quantification has yet to be made. We have developed a method that infers the distribution of Ne from the density of SNPs and de novo mutations (DNMs). Surprisingly we find that there is less variation in the density of SNPs than variation in the mutation rate, as inferred from DNMs. This suggests that regions of the genome with high mutation rates have lower Ne, a conjecture that is supported by the observation that the regression between SNP and DNM density is shallower than anticipated. The results suggest that linked selection is prevalent in the human genome, and contributes to substantial variation in Ne.

93. Correlated admixture landscapes despite reduced shared ancestry in multiple hybrid wood ant populations

Presenter: Pierre Nouhaud, Organismal & Evolutionary Biology Research Programme, University of Helsinki, Helsinki, Finland
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While admixture is widespread across the tree of life, it is still unclear how the interplay between selective and neutral processes impact the sorting of genetic variation in hybrid genomes. Replicated admixture events provide a good opportunity to disentangle between evolutionary forces at play during hybridization. The two wood ant species Formica aquilonia and Formica polyctena naturally hybridize in Southern Finland, where multiple populations have been characterized, all carrying different mitotypes. Here we ask how repeatable is the admixture landscape by resequencing whole genomes from three hybrid populations as well as parental, non-admixed individuals. Reconstructing the hybridization history revealed that admixture events are recent (less than 50 generations ago) and that hybrid populations had very little time of shared ancestry, if any. Despite this, variations in local ancestry along the genome were highly correlated between the three hybrid populations, at levels which exceed expectations under a neutral, shared origin scenario. Different processes could explain this apparent contradiction, both selective (e.g., positive or negative selection, including genetic incompatibilities) and neutral (e.g., GC-biased gene conversion, structural variants). Their possible effects and interplay with recombination rate variation will be discussed.

94. Ongoing and ancient genomic introgression in Oreochromis tilapias

Presenter: Adam Ciezarek, Earlham Institute
Co-Authors: Ford, Antonia, University of Roehampton; Durbin, Richard, University of Cambridge; Etherington, Graham, Earlham Institute; Malinsky, Milan, University of Basel; Mehta, Tarang, Earlham Institute; Nash, Will, Earlham Institute; Bradbeer, Stephanie, University of Bristol; Gracida Juarez, Carlos, University of Bristol; Smith, Alan, University of Hull; Penso-Dolfin, Luca, German Cancer Research Centre; Ngatunga, Benjamin, Tanzania Fisheries Research Institute; Shechonge, Asilatu, Tanzania Fisheries Research Institute; Tamatamah, Rashid, Tanzania Fisheries Research Institute; Haerty, Wilfried, Earlham Institute; Di Palma, Federica, University of East Anglia; Genner, Martin, University of Bristol; Turner, George, Bangor University;

The Oreochromis tilapias are an economically important group of fish for aquaculture, whose production has expanded dramatically in the last two decades. A direct consequence of this success has been the introduction of exotic species in Tanzania, a hotspot for Oreochromis diversity. Introductions have had significant negative ecological effects on indigenous Oreochromis species, including species displacement and loss of population structure through hybridisation, having potential implications for local adaptations. This history of introgression has made untangling the population history of Oreochromis difficult. We address this issue using genome-wide sequencing data across 600 individuals from 29 Oreochromis species from across Tanzania and east Africa, identifying strong signatures of introgression between species at both the population and individual level. We identify a case where a nearly discovered population of O. korogwe is being threatened by ongoing hybridisation with the invasive O. niloticus. We designed a reduced panel of 96 SNPs to further identify cases of introgression between the O. niloticus and O. leucostictus and the native O. urolepis. This SNP panel will allow users to cheaply and reliably identify introgression from these invasive species. In addition to ongoing hybridisation in several species, we find that ancestral introgression has also been prevalent across the Oreochromis radiation,

with D statistics and phylogenetic networks indicating several introgression events. We anticipate that our results will have important implications when managing the translocation of species for food production.

95. Genomic evidence of parasexual reproduction in parasite Leishmania tropica

Presenter: Tim Downing, Dublin City University
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Unicellular kinetoplastid parasite Leishmania tropica infects humans causes a wide range of cutaneous and often life-threatening disease pathologies. Leishmania's digenic life cycle includes meiosis in the vector sandfly and mitotic cell division in the mammalian host. The predominance of mitotic over meiotic cycles coupled with low coinfection rates of genetically distinct parasites in the sandfly can result in patchy heterozygosity patterns. Moreover, chromosome instability during cell division can yield aneuploidy so chromosome copy numbers can be tri-, tetra- or penta-somic. Direct evidence of classical haploid meiotic gametes has not been observed in L. tropica, resulting in questions about potential parasexual reproduction. Here, genome-wide SNP diversity from 22 L. tropica isolates showed extensive chromosome-specific runs of homozygosity and heterozygosity. Three isolates had a chromosome 36 recombination breakpoint at an inferred origin of replication, and other chromosomes had similar changes at strand-switch regions separating polycistronic transcriptional units. Five isolates had a tetra-or penta-somic chromosome 31 whose heterozygosity ratios changed from 1:3 to 2:2 to 3:1 or 4:1 for different regions. One isolate had a split chromosome 10 with a breakpoint at a strand-switch region, yielding a monosomic 5' end and a tetrasomic 3' end. All 22 had a 200 Kb heterozygous deletion on chromosome 14, and 19 had a 266 Kb centromere-spanning heterozygous deletion on chromosome 29. All isolates originated in the Middle East, including one with two genetically distinct leishmaniasis infections three years apart. This illustrates the need for genomic surveillance of tropical parasites to detect emerging hybrids that could spread more widely.

96. Early-life social, seasonal and environmental effects on telomere length in a wild mammal

Presenter: Hannah Dugdale, University of Groningen
Co-Authors: van Lieshout, Sil, U Leeds, Badás, Elisa, U Leeds & Groningen, Bright Ross, Julius, U Oxford, Bretman, Amanda, U Leeds, Newman, Chris, U Oxford, Buesching, Christina, U Oxford, Burke, Terry, U Sheffield, Macdonald, David, U Oxford, Dugdale, Hannah, U Leeds & Groningen

Early-life environmental conditions can provide a source of individual variation in life-history strategies and senescence patterns. Conditions experienced in early-life can be quantified by measuring telomere length, which acts as a biomarker of survival probability. Here, we investigate whether seasonal changes, weather conditions and group sizes are associated with early-life and/or early-adulthood telomere length in a wild population of European badgers (Meles meles). We found shortening of telomere length from spring to winter and lengthening over the winter torpor period, at both the between- and within-individual level in the first three years of life. Additionally, cubs experiencing higher mean daily temperatures and higher mean daily rainfall with low variability in spring, had longer early-life (<1 year old) telomere lengths, coinciding with food availability and foraging success. However, cubs born in groups with more cubs did not have significantly shorter early-life telomeres, providing no evidence of resource competition. After sexual maturity, in early adulthood (i.e. 12–36 months) we found no

association between telomere length and same-sex adult group size (i.e. intra-sexual competition). We demonstrate that early-life and early-adulthood seasonal changes and early-life weather conditions are associated with telomere length, which impact cub survival probability. Thus, telomere length in badgers functions as a biomarker that reflects the physiological consequences of early-life adversity and subsequent effects on cub survival probability.

97. Patterns of gene expression in seaweed flies are strongly shaped by a chromosomal inversion
Presenter: Emma Berdan, Stockholm University
Co-Authors: Claire Mérot University Laval; Maren Wellenreuther, University of Auckland; Roger Butlin, Sheffield University; Henrik Pavia, Gothenburg University; Kerstin Johannesson, Gothenburg University

Chromosomal inversions often facilitate adaptation and speciation but the reduced recombination they provide makes it difficult to separate adaptive variation from neutral variation. Combining gene expression studies with standard population genomic techniques provides a way forward. Here we complement standard population genetic techniques (see talk by Mérot) with gene expression studies to pinpoint putative adaptive variation and examine the role of cis vs. trans regulation. We use the seaweed fly, Coelopa frigida, which has a large inversion on the first chromosome that encompasses 10% of the genome and has far reaching phenotypic effects from body size to development to behavior. We examined the effect of the inversion on global expression patterns in adults and larvae and found that the inversion strongly guided expression patterns in adults but had little effect in larvae. Within adults, the inversion had stronger effects on male expression than female expression matching the magnitude of the inversion related phenotypic differences in two sexes. Across sexes and life stages the inversion mostly had cis-effects although some loci outside of the inverted region were also affected. GO enrichment analysis of our outlier loci suggested key differences in chemical signaling/sensing and digestion/metabolism. We discuss these results in light of previous phenotypic work on C. frigida, the adaptive role of the inversion, and our population genomic results.

98. Low cost of pleiotropy during polygenic adaptation to a novel environment
Presenter: Christian Schlötterer, Institut für Populationsgenetik, Vetmeduni Vienna
Co-Authors: Christodoulaki, Eirini, Institut für Populationsgenetik, Vetmeduni Vienna; Nolte, Viola, Institut für Populationsgenetik, Vetmeduni Vienna

Pleiotropy describes the phenomenon that a gene affects multiple phenotypes, but the extent of pleiotropy is still disputed. Pleiotropy has important consequences on genetic adaptation – response to selection is not only be determined by allele frequency and effect size, but also by costs of pleiotropy. In absence of empirical data, we developed a novel approach to detect and quantify the cost of pleiotropy associated with polygenic adaptation. We measured allele frequency changes in a mixture of replicate populations, that just adapted to a new, shared trait optimum using different sets of genes. Based on the allele frequency changes in the mixed populations we observed measurable, but low costs of pleiotropy. We propose that the costs of pleiotropy were reduced by the modular architecture of gene expression, which facilitated adaptive gene expression changes with low impact on other functions.

99. Busy about the Darwin Tree of Life: Acquisition, sequencing and assembly of the genomes of hundreds of species
Presenter: Mark Blaxter, Wellcome Sanger Institute
Co-Authors: The Darwin Tree of Life Partnership

A high quality reference genome sequence has the ability to transform evolutionary and population genetics of any species. We are engaged in a decadal project to generate high-quality genomes for all the species in Britain and Ireland - marine, freshwater and terrestrial and plant, fungal, animal and “protist”. The Darwin Tree of Life project (DToL) is a partnership between biodiversity organisations and their networks of expert collectors (involving Natural History Museum, London, Royal Botanic Gardens Kew, he

Royal Botanic Garden Edinburgh, Marine Biology Association and Wytham Woods research station of the University of Oxford), sequencing partners (Sanger Institute, Earlham Institute and University of Edinburgh) and analysis teams (University of Cambridge, and EMBL-EBI). Together we have built a system that takes a specimen from the field through identification, processing, sequencing, assembly and annotation to produce a reference genome in ENA annotated in Ensembl. We aim to produce chromosomally-contiguous assemblies, and to capture and release mitochondrial and symbiont genomes. We release all our data openly as they are generated. See https://darwintreeoflife.org for an introduction to the project, https://tolqc.cog.sanger.ac.uk/index.html for our raw and in-process data and https://portal.darwintreeoflife.org/ for representation of our data in ENA. All genomes will be published openly in Wellcome Open Research. I will present on behalf of my many colleagues our successes in the first year of the project, a sneak look at the first hundreds of genomes, and a map for the future of DToL.

Session 2

100. Predictability in Evolution
Presenter: Jasmine Ono, University College London
Co-Authors: None

We know that evolution can generate novelty, but we also expect natural selection to be predictable. If a new genotype arises that is more fit than its parents, it should outcompete its parents with some probability. Over the course of time, we expect the population of genotypes to become increasingly more fit on average. But how well can we hope to predict these evolutionary outcomes? At the level of phenotype? Genotype? And what information would help us to do so? Genetic constraints can limit potential routes to adaptation, creating more repeatable, or predictable, outcomes. Similarly, epistasis between potentially adaptive mutations can direct populations down a certain evolutionary path. Experimental evolution, where we are able to control the evolutionary environment and measure the repeatability of adaptation, seems like the ideal place to look for predictability. I will discuss these questions in light of some of my experimental evolution work in yeast, as well as explore how large-scale datasets might (or might not) inform our expectations.

101. Signatures of relaxed selection on social genes
Presenter: Laurie Belcher, University of Oxford
Co-Authors: None

Theory predicts that social behaviour such a cooperation will leave a distinct signature of selection in genomes. Because the fitness effects of a social gene are felt in a different individual than the one expressing the gene, selection is relaxed. The characteristic signature of this relaxation of selection is elevated sequence polymorphism and divergence. Despite these clear theoretical predictions, there is a lack of empirical work testing this theory. Such studies would be particularly useful in microbes, where they can help address ongoing debates over the extent to which certain traits are social. We address this issue by investigating the signatures of relaxed selection on social genes in Pseudomonas aeruginosa – a microbe with many well-studied social behaviours such as the production of public goods allowing iron-scavenging. By comparing social to non-social genes, we demonstrate that social genes show the characteristic signatures of relaxed selection with significantly elevated polymorphism and divergence. Such signatures have previously been interpreted independently as signatures of positive or balancing selection, but we show that they are most consistent with relaxed selection. Our findings demonstrate that looking for signatures of selection is a strong method to answer debates on microbial sociality.

102. Mutation rate plasticity: effects of nutrient availability, growth rate and population density.
Presenter: Guillaume Gomez, University of Manchester

Co-Authors: Knight, Christopher G., University of Manchester; Mcbain, Andrew, University of Manchester;

Spontaneous mutations are the fuel of evolution. Their occurrence rates can vary depending on the environment. Such plastic control of mutation rate may or may not be adaptive. Two long-standing methods are used to disentangle these rates from other evolutionary effects in microbial populations. With the fluctuation test, Luria and Delbrück in 1943 assayed the appearance of marked clones in batch culture. In continuous culture, Novick and Szilard in 1950 used the drift of marked cells to estimate mutation rate. Recent fluctuation studies have identified density-associated mutation rate plasticity (DAMP) where populations reaching high densities have relatively lower mutation rates, across domains of life. These two effects could hypothetically be the same – DAMP depends on growth rate. Alternatively, DAMP has primarily been tested by altering sugar availability, suggesting a nutrient effect, which is also known to affect mutation rate. Here we test these relationships by performing both continuous culture and fluctuation assays while varying both sugar concentrations, dilution rates and antibiotic markers. Using a bacterial strain of Escherichia coli which exhibits DAMP in batch culture, we find that, in continuous culture, all three effects are detectable and separable – decreases in mutation rate occur independently with increasing growth rate, increasing glucose and increasing population density. This behaviour illuminates the complex ways in which key ecological and physiological factors all contribute and interact to set mutation rates. It provides a basis, both for exploring the molecular mechanisms of these effects and for understanding how mutation rate plasticity affect the course of evolution.

103. Synonymous genetic variation can both build and break mutational hotspots

Presenter: James Horton, University of Bath

Co-Authors: Jackson, Robert W. University of Birmingham. Priest, Nicholas K. University of Bath. Taylor, Tiffany B. University of Bath.

Here we show that synonymous sequence facilitates extreme parallel evolution during the evolutionary rescue of flagellar motility. An immotile variant of the soil microbe, Pseudomonas fluorescens, swiftly recovers flagellum-dependent motility through parallel de novo mutation. This typically manifests within 96 h under strong selection through repeatable mutation within the nitrogen pathway's histidine kinase gene, ntrB. We found that evolution was parallel to nucleotide resolution in over 95% of cases in minimal medium (M9), with lineages repeatedly fixing an identical mutation (ntrB A289C). This repeatable de novo mutation was robust to nutrient condition despite evidence for antagonistic pleiotropy across nutrient regimes. Competition assays against alternative motile alleles revealed some evidence for selection enforcing repeated fixation of ntrB mutants, but there was no evidence for clonal interference driving parallel evolution to nucleotide resolution. Instead, the introduction of 6 synonymous substitutions surrounding the mutational hotspot reduced parallel evolution from >95% to 0% at the site. In a reciprocal experiment, we introduced 31 synonymous substitutions into a homologous strain that did not ancestrally evolve in parallel and observed that parallel evolution at the site rose from 0% to 100% in minimal media. We propose that the mechanism underlying this effect is the formation of single-stranded stem-loop DNA structures that cause extremely localised heterogeneity in de novo mutation. Our results reveal that unique quirks in how DNA is structured at specific loci can strongly bias evolutionary outcomes.

104. Experimental investigation of transcription factor rewiring dynamics in a bacterial model of gene regulatory network evolution

Presenter: Matthew James Shepherd, Milner Centre for Evolution, University of Bath

Co-Authors: Jackson, Robert, University of Birmingham, Hurst, Laurence, University of Bath, Taylor, Tiffany, University of Bath

Gene regulatory networks (GRNs) are fundamental control circuits formed by cis- and trans-regulatory elements that control expression of biological functions. An important mechanism of network architecture evolution is transcription factor rewiring. This process allows a regulator to gain or lose connections to target genes and facilitates the evolution of new transcription factors. Here we use a previously documented experimental model of transcription factor rewiring in the bacterium Pseudomonas fluorescens to investigate the rules governing this mechanism of GRN evolution. An immotile strain of P. fluorescens was engineered by knockout of the flagellum master regulator FleQ, and this strain reliably revolves flagellar regulatory control and the ability to swim by rewiring the FleQ-homologous nitrogen regulator NtrC. In this study, we eliminated the NtrC pathway to explore alternative rewiring routes and discover factors that drive patterns of rewiring. In a ΔntrC mutant, motility repeatedly re-evolved by regulatory rewiring, however this took >10 days longer to occur than for NtrC, only evolved in <10% of repeats and provided a far poorer motility phenotype. Genomic and transcriptomic analyses shed light on the complex regulatory dynamics involved in evolutionarily repurposing these transcription factors. They indicate two contrasting methods that rescue flagellar expression in the absence of ntrC, one targeting regulatory network structure local to a rewired regulator, and another achieving motility through changes to global gene expression control of a significant number of genes. This work provides key empirical data for understanding factors which influence the evolution of GRNs and the phenotypes that they control.

105. Watching the grass grow: Assessing lethal mutagenesis as a strategy for treating acute respiratory viral infection

Presenter: Chris Illingworth, MRC Biostatistics Unit, University of Cambridge
Co-Authors: None

Lethal mutagenesis is a well-established strategy for treating RNA viral infection. Increasing the viral mutation rate increases the load of deleterious mutations upon the virus, negatively impacting viral fitness. The conserved structure of the viral protein targeted by mutagenic drugs makes lethal mutagenesis a promising approach for the treatment of multiple RNA viral species. However, clinical experience of such drugs provides sometimes ambiguous results, with apparently positive effects in patients not being matched by a significant decrease in viral load. We here consider a case of chronic respiratory syncytial virus infection in a severely immunocompromised child in which a novel antiviral strategy, combining ribavirin and favipiravir, led to a positive clinical outcome. We combine deep sequence data describing the evolution of the viral population with population genetic modelling to shed light on the outcome of this case. Our model enables us to estimate changes in the mutation rate, and in viral fitness, induced by drug therapy. We explain why reducing viral load may not be necessary for treatment to be successful. Our work sets out a path for the quantitative investigation of proposed treatments for severe RNA viral infection.

106. Neural networks as optimal estimators of the mutation rate or the effective population size for variable recombination rates

Presenter: Franz Baumdicker, Cluster of Excellence Controlling Microbes to Fight Infections, University of Tübingen
Co-Authors: Burger, Klara, Cluster of Excellence Controlling Microbes to Fight Infections, University of Tübingen.; Pfaffelhuber, Peter, Department of Mathematical Stochastics, University of Freiburg.

Although machine learning methods in population genetics face multiple specific challenges, it is meanwhile clear that they are a promising technique to build more powerful inference tools, especially for problems that are hard to tackle down with classical methods. In contrast, in population genetics, many theoretical results have been developed within the last decades and enabled us to identify the best inference technique for some scenarios. For example, the estimation of mutation rate, or equivalently effective population size, is well understood, at least if recombination is low or high. In these two scenarios the best estimation methods, namely Fu's and Watterson's estimator, are known and well

understood. For intermediate recombination rates, the theoretical development of optimal estimators is much more involved. Machine learning tools could help to develop good estimators in these involved scenarios, but it is difficulat to assess how well suited these tools are for different applications in population genetics if no benchmark is available. Here we investigate simple feed-forward neural networks for the estimation of the mutation rate and compare their performance with the frequently used optimal estimators introduced by Fu and Watterson. We find that neural networks can reproduce the known optimal estimators if provided with the appropriate features, but can otherwise easily underperform. Remarkably, only one hidden layer is necessary to obtain a single estimator that performs almost as well as the optimal estimators for low and high recombination rates and provides a superior estimation method for intermediate recombination rates at the same time.

107. Structure of the global population of cyanobacterium Microcoleus

Presenter: Petr Dvorak, Palacky University Olomouc, Department of Botany
Co-Authors: Stanojkovic, Aleksandar, Palacky University Olomouc, Department of Botany, Skoupy, Svatopluk, Palacky University Olomouc, Department of Botany, Poulickova Aloisie, Palacky University Olomouc, Department of Botany

The diversity of prokaryotic species is immense, but the forces shaping the evolution of this diversity are still only poorly understood. Here, we investigated the role of geographical isolation in prokaryotic speciation. In animals and plants, allopatric speciation is still considered as the most common mode of speciation. But prokaryotes have much higher dispersal capacities, and thus, sympatric speciation is more likely to occur, because distant populations are connected by gene flow. We chose cyanobacterium Microcoleus vaginatus as a model system to study distribution patterns within prokaryotes. We isolated and sequenced 16S rRNA and ITS regions of 500 strains originating from all continents except South America. The phylogenetic reconstruction and ancestral state reconstruction revealed large diversity, which suggests that there are many lineages at species level, although M. vaginatus is considered as one species. We found that most of the lineages are geographically structured and geographical and genetic distance correlate. However, some instances of almost genetically identical strains from distant localities were found. Moreover, each locality was inhabited by one to five distantly related lineages indicating high local genetic diversity. Further, we sequenced 120 Microcoleus genomes from our strain collection. We found similar pattern to 16S rRNA and ITS analysis. Pan-genome analysis of two lineages revealed that each genome has unique gene composition. Moreover, the pan-genome is extremely diverse with tens of thousands of genes for each Microcoleus lineage.

108. Understanding the causes and consequences of gene regulatory network rewiring patterns using experimental evolution

Presenter: Tiffany Taylor, University of Bath
Co-Authors: Flanagan, Louise, University of Bath. Horton, James, University of Bath. Shepherd, Matthew, University of Bath

Previous work has shown that gene regulatory networks (GRNs) are dynamical such that connections are forged and lost frequently across an evolutionary timescale. But, how are opportunities for rewiring events between GRNs revealed to selection? And what are the evolutionary consequences of alternative regulatory pathways in evolving populations? The Taylor lab uses a combination of experimental evolution and molecular tools with Pseudomonas fluorescens, to explore rewiring of the flagellar regulation network as a model system. Our aim is to provide a greater general understanding of the common underlying principles that determine GRN rewiring patterns. I will discuss recent revelations from my lab group that reveal the role of mutation bias in driving rewiring patterns, and the evolutionary consequence of strong biases in evolving populations.

109. Gametic Competition under Different Mating Systems
Presenter: Michael Francis Scott, University of East Anglia
Co-Authors: Immler, Simone, University of East Anglia

Sperm and pollen can experience strong competition for fertilization. However, the competitors are determined by the behaviour of the adults that produce them. Here, we use mathematical modelling to evaluate selection on sperm/pollen under different mating systems: monandry, polyandry, selfing, and outcrossing. We evaluate the spread of adaptive mutations, genetic load, and the maintenance of genetic polymorphism for genes that express their haploid genotype in sperm/pollen to a greater or lesser extent. Despite creating locally intense competition involving heterozygous males, we find that both monandry and selfing reduce the overall efficacy of selection on haploid genotypes. The difference between mating systems is expected to be even more stark when sperm/pollen have diploid expression because no selection occurs in matings that involve a single male. These predictions can therefore be tested using gene sets that have different pollen/sperm expression patterns across populations/species with different mating systems.

Session 3

110. Adaptation to altitude in Heliconius butterflies
Presenter: Gabriela Montejo-Kovacevich, University of Cambridge
Co-Authors: Martin, Simon, University of Edinburgh. Meier, Joana, University of Cambridge. Salazar, Patricio, University of Sheffield. Bacquet, Caroline, Universidad Regional Amazonica Ikiam. Salazar, Camilo, Universidad del Rosario. Nadeau, Nicola, University of Sheffield. Jiggins, Chris, University of Cambridge.

Adaptation to local environmental conditions can determine evolutionary divergence along environmental clines. Altitudinal clines are steep environmental gradients that provide the ideal setting for studying local adaptation while minimising the pervasive effects of geographic isolation and genetic drift on population structure. A key question is how strong and detectable selection is in the wild and how convergent local adaptation to the environment is among closely related species. Here we tackle these by studying continuous populations of Heliconius butterflies ranging 1500m in elevation across both sides of the Andean mountains. We integrate: (i) whole genome scans to find signatures of positive selection in high altitudes, (ii) comparative phenotypic methods to find the traits conferring adaptation, (iii) common-garden rearing experiments to test their heritability and (iv) quantitive association studies to find candidate loci for those traits. We used population branch statistics with 600 whole-genome sequenced wild Heliconius from four replicated altitudinal clines and found strong signatures of selection to high altitude, as well as high levels of convergence between clines on either side of the Andes. With a wild collection of over 3000 individuals and common-garden rearing, we show that wings are rounder at high elevations, both within and between species, and that wing shape is heritable. Finally, we found candidate genomic regions underlying wing shape variation through association mapping, which were also diverging across elevations. Our work succeeds in linking signatures of selection across genomes to local environmental adaptation, while revealing tractable potentially adaptive traits.

111. Combining population genomics with genome editing: Towards an integrative view of cold tolerance in Drosophila
Presenter: Sonja Grath, LMU Munich
Co-Authors: Königer, Annabella, LMU Munich; Ramnarine, Timothy J. S., LMU Munich; Mußgnug, Selina, Genecenter Munich; Yilmaz, Vera M., LMU Munich

Temperature is one of the major factors influencing the geographical distribution and abundance of many animals. As the body temperature of insects mainly follows the external environment, tolerance towards thermal extremes is essential for adaptation to new environments such those arising through range expansion or climate change. In our laboratory, we established Drosophila ananassae as model to elucidate the genetic basis of cold tolerance. Here, I present two stages of our project. First, we identified candidate genes for cold tolerance by means of comparative transcriptomics and a genome-wide QTL mapping experiment with cold-tolerant and cold-sensitive fly strains. We further established molecular genetic tools for genome editing by CRISPR/Cas in this species. Second, as we still lack information on the function of these candidate genes in tissues particularly relevant for cold tolerance, we will use the tools developed in stage 1 to perform tissue-specific genome editing. We further aim to identify polymorphisms related to cold tolerance based on genome sequencing of strains originating from cold-tolerant and cold-sensitive populations. The results of these experiments will shed light on thermal adaptation of natural populations. In addition, the combination of state-of-the-art genomic and genome editing methods will provide new information on the function of different genes, both in the whole organism and individual tissues. These data will not only elucidate the function of genes and polymorphisms associated with cold tolerance, but will also allow the analysis of additional phenotypes and comparison with other species to identify general patterns of environmental adaptation.

112. Species assignment in the Anopheles genus using targeted amplicon sequencing
Presenter: Marilou Boddé, University of Cambridge
Co-Authors: Makunin, Alex, Wellcome Sanger Institute; Korlevic, Petra, Wellcome Sanger Institute & the European Bioinformatics Institute; Durbin, Richard, University of Cambridge; Lawniczak, Mara, Wellcome Sanger Institute

The Anopheles genus contains about 500 formally recognised species, approximately 100 of which are capable of transmitting human malaria. Species indistinguishable by morphology may differ in behaviour, phenotype and genomic content, all of which can impact efficacy of malaria control efforts. Over the last year our group developed a genus-wide targeted amplicon sequencing panel to facilitate large-scale monitoring of Anopheles populations. Combining information from these amplicons allows for a more nuanced species assignment than single gene (e.g. COX1) barcoding, which is desirable in the light of Anopheles' permeable species boundaries. I will present a hierarchical species assignment method working on these amplicon sequences. The first layer assigns a sample to a clade based on a nearest neighbour approach; the second layer uses a variational auto encoder to predict the species. I will show a concordance study of the amplicon data with the Ag1000G [1,2] whole genome data for the Anopheles gambiae complex. This panel will be used to survey Anopheles species diversity and plasmodium transmission patterns through space and time on a large scale; we aim to analyse half a million mosquitoes in the next five year. References: 1. Ag1000G Consortium, Nature (2017) 2. Ag1000G Consortium, Genome Research (2020)

113. The story behind the strains: Examining the phylogeography of wild yeast from woodlands
Presenter: Jacqueline Peña, University of Georgia
Co-Authors: Peña, Jacqueline, University of Georgia and Bensasson, Douda, University of Georgia

The Saccharomyces genus includes the iconic model system, Saccharomyces cerevisiae, that is the workhorse for scientific research and biotechnology because of its tractability and short generation time. With advancements in sequencing technology, genomes are paving the way to resolve the natural history of S. cerevisiae and other Saccharomyces species. The biogeography of wild S. cerevisiae is less clear because it has been heavily influenced by domestication, which increases dispersal and reduces geographic structure. To determine if wild S. cerevisiae isolates are distinct from domesticated isolates, I examined whether biogeographic patterns of wild S. cerevisiae isolates from woodlands show genetic structure. I asked: Do woodland populations show genetic substructure that is consistent with isolation

by distance? I used publicly available whole-genome data to determine the population structure and phylogenetic relationships. I was able to recapitulate known phylogenetic relationships of domesticated S. cerevisiae lineages and found that woodland populations from around the world are genetically distinct. This suggest that natural S. cerevisiae isolates have not been drowned out by the effects of domestication and that strains from a single woodland can then be used to assess local adaptation.

114. Polymorphism-Aware Phylogenetic Models and their Applications

Presenter: Carolin Kosiol, University of St Andrews
Co-Authors: 1) Barata, Carolina, University of St Andrews; 2) Borges, Rui, Vetmeduni Vienna

The increased availability of sequenced genomes both from closely related species and from individuals of the same species, offers a great opportunity to study the speciation and evolutionary history of populations, provided we can properly model the process of sequence evolution using inter and intraspecific data together. In my group, we have developed a new method called POlymorphisms-aware phylogenetic MOdel (PoMo). It extends any DNA substitution model and additionally accounts for polymorphisms in the present and in the ancestral population by expanding the state space to include polymorphic states in a continuous Markov process. It is a selection-mutation model which separates the mutation process from the fixation process. Thereby, a Moran process is used to model genetic drift. PoMo naturally accounts for incomplete lineage sorting because ancestral populations can be in a polymorphic state. Our method can accurately and time-efficiently estimate the parameters describing evolutionary patterns for phylogenetic trees of any shape (species trees, population trees, or any combination of those). We have implemented the approach into Maximum Likelihood software package and recently developed a Bayesian approach for molecular dating. I will present what can be learned by applying these methods to genome-wide data sites of great ape populations about ancestral population history of this species. Finally, I will also discuss how the new methods could be applied to populations of fruit flies that have recently been subject to an experimental evolution study for sexual mating system.

115. Genetic barriers to historical gene flow between cryptic species of alpine bumblebees revealed by comparative population genomics

Presenter: Matthew Webster, Uppsala University
Co-Authors: Christmas, Matthew, Uppsala University; Jones, Julia, University College Dublin; Olsson, Anna, Uppsala University; Wallerman, Ola, Uppsala University; Bunikis, Ignas, Uppsala University; Kierczak, Marcin, Uppsala University; Peona, Valentina, Uppsala University; Suh, Alexander, Uppsala University ; Miller-Struttmann, Nicole, Webster University ; Geib, Jennifer, Appalachian State University

Evidence is accumulating that gene flow commonly occurs between recently-diverged species, despite the existence of barriers to gene flow in their genomes. However, we still know little about what regions of the genome become barriers to gene flow and how such barriers form. Here we compare genetic differentiation across the genomes of bumblebee species living in sympatry and allopatry to reveal the potential impact of gene flow on genome divergence and uncover the nature of genetic barrier loci. We first compared the genomes of the alpine bumblebee Bombus sylvicola and a previously unidentified sister species living in sympatry in the Rocky Mountains, revealing prominent peaks of elevated genetic divergence that co-localize with centromeres and regions of low recombination. This same pattern is observed between the genomes of another pair of closely-related species living in allopatry (B. bifarius and B. vancouverensis). Strikingly however, statistical analysis of genetic variation in these two comparisons indicates that the peaks of divergence contain loci that have acted as barriers to historical gene flow in the sympatric, but not the allopatric comparison. Our results suggest that intrinsic barriers to gene flow between species may often accumulate in regions of low recombination and near centromeres through processes such as genetic hitchhiking, and that divergence in these regions is accentuated in the presence of gene flow.

116. The processes underlying supergene evolution

Presenter: Mathieu Joron, CEFE CNRS, Montpellier, France
Co-Authors: Jay, Paul, CEFE CNRS Univ Montpellier France; Chouteau, Mathieu, LLEISA CNRS Univ Guyane French Guiana; Aubier, Thomas, University of Zurich Switzerland; Whibley, Annabel, University of Auckland New Zealand; Llaurens, Violaine, ISYEB CNRS Sorbonne Univ Paris France; Joron, Mathieu, CEFE CNRS Univ Montpellier France

Chromosomal inversions are often involved in coordinating complex phenotypes, such as major covariation of behavior and morphology in birds, fishes, insects or mammals. Despite featuring prominently in the early history of genetics, why and how inversions become associated with polymorphic traits remains obscure. Here we show that the polymorphic inversions underlying the supergene inheritance of wing-pattern mimicry polymorphism in a butterfly are maintained by the accumulation of deleterious variation. Using a combination of genomics and in vivo fitness analyses, we reveal that 3 inversions associated with ecologically advantageous morphs (mimicry) have built up a heavy recessive mutational load from the sequential accumulation of mutations and transposable elements. This load is expressed in homozygotes as a strong viability cost, preventing inversions from fully replacing ancestral chromosome arrangements. So, despite a strong selective advantage when they form, inversions generate selection against their own fixation and promote their maintenance at intermediate frequency. But this does not explain how differentiated forms arise. By exploring the fate of overdominant inversions in a two-population model, we show that admixture between differentiated, locally-adapted populations may readily lead to the evolution of supergenes and stable polymorphisms of differentiated forms. Our results allows us revisiting the idea that supergenes form in response to the existence of alternative ecological optima corresponding to the different phenotypic forms found segregating stably in populations.

117. Relative timing of admixture during range expansion of Zaprionus indianus

Presenter: Aaron A. Comeault, Bangor University
Co-Authors: Kautt, Andreas, Harvard University; Matute, Daniel, University of North Carolina

Introduced species have become a common component of biological communities around the world. A central goal in invasion biology is therefore to identify the demographic and evolutionary factors that underlie successful introductions. Here we use whole genome sequences, collected from populations in the native and introduced ranges of the African fig fly, Zaprionus indianus, to quantify genetic relationships among them, identify potential sources of the introductions, and test for selection at different spatial scales. We find that geographically widespread populations in the western hemisphere are genetically more similar to each other than to lineages sampled across Africa, and that these populations share a mixture of alleles derived from differentiated African lineages. Using patterns of allele-sharing and demographic modelling we show that Z. indinaus have undergone a single expansion across the western hemisphere with admixture between African lineages predating this expansion. We also find support for selection that is shared across populations in the western hemisphere, and in some cases, with a subset of African populations. This suggests either that parallel selection has acted across a large part of Z. indianus's introduced range; or, more parsimoniously, that Z. indianus has experienced selection early on during (or prior-to) its expansion into the western hemisphere. We suggest that Z. indianus's range expansion has been facilitated by admixture and selection, and that management of the invasion in the western hemisphere should focus on movement of individuals within this region rather than between the western and eastern hemisphere.

118. The genetic basis and evolutionary processes shaping adaptive trait divergence under pervasive gene flow in a marine fish

Presenter: Arne Jacobs, University of Glasgow

Co-Authors: Akopyan, Maria, Cornell University; ; Wilder, Aryn, San Diego Zoo Institute for Conservation Research; ; Tigano, Anna, University of New Hampshire; ; Baumann, Hannes, University of Connecticut; ; Therkildsen, Nina. O, Cornell University

Determining how species locally adapt to different temperature regimes in marine environments despite strong gene flow is a major question in eco-evolutionary research. Genomic rearrangements that suppress recombination and limit the dissociation of adaptive variants (e.g. inversions) are hypothesized to play a major role in local adaptation in marine species. Yet, inversions have not been directly linked to adaptive trait variation in most cases. Using the Atlantic silverside (Menidia menidia), a marine fish distributed along a steep thermal cline at the US Atlantic Coast, we test the role of polymorphic inversions and spatially-varying selection in local adaptation in this ecological model species. Combining quantitative trait locus (QTL) mapping with low-coverage whole genome resequencing of 576 Atlantic silversides from the entire species' range, we i) map the genetic basis of adaptive traits and ii) investigate the role of spatially-varying selection in shaping the underlying adaptive genetic variation. We identify multiple large-effect QTL associated with highly divergent adaptive traits, e.g. growth rate, of which several co-localise with large polymorphic inversions on multiple chromosomes. QTL-linked inversions show varying latitudinal clines and are associated with different environmental variables, indicative of spatially-variable selection pressures. Furthermore, signatures of selection vary across the species' range, not only between inversions but also within inversions. Overall, our study represents a major step forward for our understanding of local adaptation in a marine fish by directly implicating polymorphic inversions in adaptive trait divergence under gene flow and elucidating the evolutionary processes shaping the underlying genetic variation.

119. Can we use plant pathogen genetic variation in the wild to understand adaptation in crop pathogens?
Presenter: Mark McMullan, Earlham Institute
Co-Authors: Percival-Alwyn, Lawrence, NIAB.; Kaithakottil, Gemy, Earlham Institute; Swarbreck, David, Earlham Institute; Hall, Neil, Earlham Institute

Here we look for the signal of adaptation to an agricultural environment between populations of fungi living on wild sea beet and agricultural sugar beet. This work has implications for understanding how pathogens evolve and evade host defence as well as our ability to better defend our crops. Sugar beet (Beta vulgaris) is one of the most recently domesticated crop species and here we explore genetic diversity of its rust pathogen Uromyces beticola, which is found on both wild and agricultural beets. We have assembled and annotated a 600Mb genome in order identify population genetic measures associated with adaptation to agriculture. Until recently all crop pathogens lived exclusively on wild hosts. The introduction of farming has produced hosts that are genetically very similar, and their resistance (chemical and genetic) is controlled by humans. The pathogens of crops that were domesticated longer ago, may well have speciated from their wild counterparts but more recently domesticated crops may still harbour pathogens that are capable of infecting both wild and agricultural hosts. Our hypothesis is that the selection pressures of an agricultural environment will be visible in the genes responsible for pathogen success in agriculture.

Session 4

120. Pervasive selection biases inferences of the species tree
Presenter: Rui Borges, Vetmeduni Vienna
Co-Authors: Boussau, Bastien, Université de Lyon; Szöllősi, Gergely, Eötvös University; Kosiol, Carolin, University of St Andrews

Despite the importance of natural selection in species' evolutionary history, phylogenetic methods that take into account population-level processes ignore selection. Assuming neutrality is often based on the idea that selection occurs at a minority of loci in the genome and is unlikely to significantly compromise phylogenetic inferences. However, selection might behave more pervasively, as it the case of nearly neutral evolving mutations. Genome-wide processes like GC-bias and some of the variation segregating at the coding regions are known to evolve in the nearly neutral range. As we are now using genome-wide data to estimate species tree, it is just natural to ask whether weak, but pervasive, selection is likely to blur species tree inferences. We developed a polymorphism-aware phylogenetic model, specially tailored for measuring signatures of nucleotide usage biases, to test the impact of selection in the species tree. Our analyses indicate that while the inferred relationships among species are not significantly compromised, the genetic distances are systematically underestimated in a node-height dependent manner: i.e., the deeper nodes tend to be more underestimated than the shallow ones. Such biases have implications for molecular dating. We dated the evolutionary history of 31 worldwide fruit fly populations, and we found signatures of GC-bias considerably affecting the estimated divergence times (up to 21%) in the neutral model. Our findings call for the need to account for selection when quantifying divergence or dating species evolution.

121. The effect of epistasis on local adaptation with gene flow
Presenter: Martin Pontz, Universität Wien
Co-Authors: Bürger, Reinhard, Universität Wien

There is little consensus on the importance of epistasis for local adaptation. Here, we investigate the possibility of establishment of weakly locally adaptive de-novo mutations in the face of gene flow, linkage and epistatis. This process is studied by applying a two-type branching process to the underlying two-locus two-allele model with continent-island migration. The new mutation appears on the island and its offspring distribution depends on the background allele. We use the theory of two-type branching processes to determine the dependence of the establishment probability on the parameters, i.e., selective coefficients of the alleles, migration and recombination rate and epistasis. In general, this establishment probability can not be expressed in closed form, however, we derive an explicit expression that is a good approximation. We use this approximation to predict the average establishment probability over all possible linked mutations.

122. The collapse of genetic incompatibilities in a hybridizing population
Presenter: Tianzhu Xiong, Harvard University, Organismic and Evolutionary Biology
Co-Authors: Mallet, James, Harvard University

Genetic incompatibilities have long been considered as one of the hallmarks of speciation due to their role in reproductive isolation. Previous analyses of the stability of epistatic incompatibility show that it is subject to collapse upon hybridization. In the present work, we derive explicitly the distribution of the lifespan of two-locus incompatibilities, and show that genetic drift, along with recombination, is critical in determining the time scale of collapse. Classical incompatibilities survive longer in smaller populations when incompatible alleles are (co)dominant, but collapse more quickly otherwise. Metastable incompatibilities involving mismatched regulatory elements have a collapse time scale determined by the exponential of the absolute number of recombination events per unit time, and thus are even more sensitive to population size and genomic structures. Together, the theory suggests that the load of genetic incompatibilities between hybridizing species is a dynamic process sensitive to the noise of reproduction, especially when hybrids are rare. Interpreting patterns of post-zygotic barriers in naturally hybridizing species should not always assume deterministic equilibrium on barrier loci.

123. The effects of population size contraction on the rate of adaptive evolution in humans
Presenter: Vivak Soni, University of Sussex

Co-Authors: Eyre-Walker, Adam, University of Sussex

Expanding population size tends to generate artefactual evidence of adaptive evolution because slightly deleterious mutations that were fixed in the ancestral population no longer segregate in the population. Bergman and Eyre-Walker (MBE, 2019) observed strong correlations between the rate of adaptive evolution and amino acid dissimilarity (as measured by the difference in polarity, volume or Pn/Ps between amino acids) in Drosophila melanogaster. We find that these correlations are much weaker in humans. This can be explained by the population contraction in humans since the human-chimpanzee split which tends to reduce genuine correlations between the rate of adaptive evolution and amino acid dissimilarity. Here we show that population size increases can artifactually generate negative correlations between an estimate of the rate of adaptive evolution and the mean strength of selection against deleterious mutations, even if there is no adaptive evolution, and that the reverse is true in cases of population contraction.

124. Ancestry-related assortative mating and sex bias driven by social stratification in admixing American populations

Presenter: Mas-Sandoval, Alex, Department of Life Sciences, Silwood Park campus, Imperial College London, Ascot, UK
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The cultural and socioeconomic stratification of human societies has led to non-random mating and shaped the structure of populations beyond the sole effect of geography. In fact, in recently admixed populations, their structure is mainly modulated by the effect of assortative mating related to the ancestral components of the admixing populations, which is frequently asymmetric between women and men. Nevertheless, the effect of assortative mating is currently largely underestimated or disregarded in most population genetics studies. In this study we analyze the footprint of the mating patterns on the structure of the admixing populations across the Americas, driven by cultural and socioeconomic factors. To shed light on these questions, we derive a mechanistic model of ancestry-related assortative mating and sex bias and generate simulations of various complex admixture scenarios. We then implement and train an artificial neural network to infer the mating parameters of admixing American populations from genomic data. We show how ancestry-related assortative mating and sex bias has constrained the admixture process in the Americas between Native American, European and sub-Saharan African genetic components since the European colonization and the subsequent Atlantic slave trade. Furthermore, we highlight the need of a fine analysis of complex admixture events to avoid the misinterpretation of admixture dates and ratios. Finally, we stress the importance of the cultural and socioeconomical context for understanding the evolutionary processes of the genetic history of human populations.

125. Divergent selection and linked deleterious mutations with gene flow: diploid vs hemizygous chromosomes

Presenter: Vítor C Sousa, CE3C University of Lisbon
Co-Authors: João C. Frazão, cE3c – University of Lisbon; ; Emily E. Bendall, University of Kentucky;; Gonçalo Caneira, cE3c – University of Lisbon;; Catherine R. Linnen, University of Kentucky;; Vitor C. Sousa, cE3c – University of Lisbon

Genomic scans of differentiation are widely used to detect outliers potentially under divergent selection. However, the effect of removing deleterious mutations can confound these analyses. To quantify this effect we model migration, divergent selection and deleterious mutations, using deterministic and SLiM simulations. Furthermore, we compare genetic differentiation patterns in sex-limited hemizygous chromosomes (e.g. X/Z sex-chromosome or haplodiploid autosomes) with diploid autosomes. For models

with divergent selection, we found higher peaks of differentiation in hemizygous than diploid chromosomes, consistent with more efficient selection against maladaptive immigrant alleles due to hemizygosity. Our results predict that background selection (BGS) increases differentiation, but this effect vanishes with increasing migration rates. For recessive and slightly deleterious mutations, we predict diploid chromosomes to be affected by associative overdominance (AOD) at low recombining regions, creating valleys of differentiation. Interestingly, such valleys of differentiation due to AOD are also seen for hemizygous chromosomes, but at a much narrower range of the parameter space. We compare these predictions with genome-wide data from hybridizing haplodiploids (Neodiprion sawflies). Our results indicate that AOD and BGS create heterogeneous genomic patterns, potentially biasing genomic scans to detect divergent selection.

126. A simple approximation for the strength of selection for recombination caused by interference among mutations

Presenter: Denis Roze, CNRS
Co-Authors: None

One of the most widely cited hypothesis to explain the evolutionary maintenance of genetic recombination states that the reshuffling of genotypes at meiosis increases the efficiency of natural selection by reducing interference among selected loci. However, most models on the evolution of recombination consider only a few selected loci, and the possible strength of indirect selection for recombination under more realistic settings remains difficult to quantify. Using models in which deleterious mutations occur along a whole chromosome, I will show that the strength of selection for recombination can often be approximated by a simple expression of NeU and NeR (where Ne is the effective population size, and U, R the deleterious mutation rate per chromosome and chromosome map length), and is not affected much by details of the genetic architecture of fitness (selection and dominance coefficients of deleterious alleles, epistasis), nor on the genetic architecture of recombination rate variation. Indirect selection generated by interference among mutations may compensate for substantial costs associated with recombination when linkage is tight; however, its effect generally stays weak in large, highly recombining populations.

127. Hitch-hiking revisited

Presenter: Nicholas Hamilton Barton, IST Austria
Co-Authors: None

Maynard Smith and Haigh (1974) showed how the fixation of a favourable mutation would reduce diversity in the surrounding genome. This classic model of a selective sweep has been highly influential, and has inspired methods for inferring the action of selection from sequence data. Yet, several aspects of the theory are under-appreciated, and inference methods remain limited. I review the effects of selective sweeps on linked genealogies, and show how this allows inference of selection from haplotype structure.

128. Polygenic adaptation to a new environment under self–fertilisation

Presenter: Matthew Hartfield, Institute of Evolutionary Biology, The University of Edinburgh
Co-Authors: Glémin, Sylvain, Université de Rennes, CNRS, ECOBIO UMR 6553, F-35000 Rennes, France.

Many quantitative traits are likely subject to polygenic selection, where many genetic variants influence the trait. Elucidating the dynamics of polygenic selection is currently a major focus of both theoretical and empirical evolutionary genetics research. The role of the mating system is often overlooked in these studies; it is important to consider it as many natural populations (especially plants) often reproduce uniparentally to some degree. It can fundamentally change the dynamics of polygenic selection by increasing homozygosity within a population, which also weakens the efficacy of recombination. We present results arising from a computational model of a population experiencing different levels of self–

fertilisation, as it adapts to an environmental change. Mutations either affect a single trait, or pleiotropically affect multiple traits. We investigate how self–fertilisation either helps or hinders adaptation to a new environment. We show that a combination of self–fertilisation and pleiotropy causes a transition from selection acting on individual alleles, to the formation of clonal genotypes consisting of co–adapted gene complexes.

129. Expectation and properties of an allele sharing, moment-based estimator of population specific FST

Presenter: Jerome Goudet, Dept Ecology & Evolution, University of Lausanne, Switzeland
Co-Authors: Bruce Weir, Deaprtment of Biostatistics, University of Washington, Seattle, USA

Being able to properly quantify genetic differentiation is key to understand a species evolutionary potential. One central statistic in this context is FST, the relative mean coancestry within populations relative to the total. Researchers have been estimating FST globally or between pairs of populations for a long time. More recently, it has been proposed to estimate population specific FSTs. Here, we review the several definitions and estimation of FSTs, and insist that they are measured relative to a reference population. We show the good statistical properties of a allele sharing, method of moments based estimator of FST (global, population specific and pairwise) under a very general model population structure, and point to the limit of Likelihood and Bayesian estimators when the populations are not independent. Last, we show that recent attempts to estimate absolute, rather than relative, mean coancestry fail to do so.

130. Do we need to estimate the probability of identity by descent?

Presenter: Bruce Weir, University of Washington
Co-Authors: Jerome Goudet, University of Lausanne

Probabilities of identity by descent (ibd) are widely used in population and quantitative genetics. Other than by methods based on runs of homozygosity or on inferred ibd segments in phased genotype data that require tuning of search parameters, ibd probabilities are not estimable from SNP data. The classic Li-Horvitz estimator, based on observed and expected heterozygosity, estimates the within-population inbreeding coefficient f (Wright's Fis) rather than the ibd probability F (analogous to Wright's Fit). For a large number of SNPs it is unbiased with large sample sizes and it holds for individuals and populations. The individual-specific estimates F1 and F3 in the GCTA package, also for large sample sizes, are unbiased for (f-4g) and (f-2g), where g is the average within-population coancestry coefficient for an individual with all other individuals in the sample providing the necessary sample allele frequencies. Allele-sharing estimates are unbiased for f for all sample sizes and give consistent rankings across all samples. We suggest these estimates are sufficient for many applications. For example, the predicted genetic variance for an additive quantitative trait is (1+F)A, where A is the additive variance component, but the trait variance for a sample of individuals has an expected value of (1+f)A. Forensic match probabilities, shown by Balding and Nichols to depend on the population structure parameter theta (Wright's Fst), can be estimated with database allele frequencies and allele-sharing estimates of within-population versions of theta. Within-population measures, as estimated by allele-sharing methods, can be negative as they are not ibd probabilities but they are relevant for empirical studies.

Poster abstracts

Poster Room 1

1. Demography of island populations of barn owls in the Eastern Mediterranean

Presenter: Alexandros Topaloudis, University of Lausanne
Co-Authors: Machado, Ana Paula, University of Lausanne; Goudet, Jerome, University of Lausanne

Island populations are characterized by distinct demographic histories due to their isolated nature. Population size bottlenecks during colonization and reduced gene flow will accelerate the divergence of insular populations and leave a distinct mark on the DNA of individuals. Using present-day genomic data, we can attempt to infer the demography of such populations and shed light on the forces that shaped their polymorphism. The barn owl (Tyto alba) has colonized almost every continent but little is known about insular populations of the species. In this study we make use of whole genome sequencing of 67 owls from islands and the mainland around the Eastern Mediterranean and employ a suite of cutting-edge population genomics tools to characterize present-day diversity, quantify divergence and elucidate past demography. We show that populations from the islands of Crete, Cyprus and the Aegean archipelago have very distinct demographic histories and diversity, reflecting different origins and connectivity. We argue that such differences are non-trivial on such a small geographic scale for a cosmopolitan species and probably stem from impeded over-water dispersal. We highlight that islands in proximity and with similar geological histories may harbor populations of the same species each with unique genetic compositions, necessitating discrete conservation management.

2. In-silico cross-contamination and its effects on whole genome phylogenies in Saccharomyces cerevisiae

Presenter: Audrey K. Ward, University of Georgia
Co-Authors: Scopel, Eduardo, University of Georgia. Shuman, Brent, University of Georgia. Momany, Michelle, University of Georgia. Bensasson, Douda, University of Georgia.

Population genetic analysis depends on the quality of whole genome sequences. Contamination of sequence data may occur prior to sequencing, or during multiplex sequencing as a result of cross-contamination or barcoding issues. While it is common to check for interspecies contamination, identifying contamination from the same species is less prevalent, even though this may have a major impact on the accuracy of downstream analyses. In order to study the effects of contamination on genome analyses, we have taken multiple lineages of Saccharomyces cerevisiae with varying ploidies and contaminated them along a range from 0-50%. Using a common population genetic pipeline, we produced phylogenies for each contamination level in order to assess how contamination affects phylogenetic analysis. Results suggest that even low levels of contamination among sample sequences can produce significant changes that could cause misunderstanding of evolutionary relationships between closely related lineages.

3. Which data for estimating Runs of Homozygosity?

Presenter: Eléonore Lavanchy, Department of Ecology & Evolution, University of Lausanne, Switzerland
Co-Authors: Goudet Jérôme, Department of Ecology & Evolution, University of Lausanne, Switzerland

Quantifying inbreeding and its deleterious consequences are central in many areas of Biology, from human genetics to conservation biology. Until recently, a standard measure of inbreeding was the average excess homozygosity. However, because parents transmit large chunks of their chromosomes to their offspring, strong inbreeding usually occurs in blocks. Such segments, also called Runs of Homozygosity (ROHs) have been linked to a plethora of deleterious phenotypes, notably in humans. Identifying ROHs is straightforward with Whole Genome Sequencing (WGS) data. However, in the literature, ROHs analyses have been performed on reduced representations of the genome (such as RAD-sequencing, Whole-exome data, SNPs arrays, etc.) and whether ROHs can be reliably estimated with these types of data is still controversial. To address this problem, we used simulations to test which types of data are suitable for ROHs estimation and we aim to propose technical guidelines, such as the minimum SNP density, to reliably call ROHs. We simulated WGS-like data for different populations with varying characteristics such as mating system and population size. Then, we subsampled SNPs to mimic sequencing techniques leading to reduced representation of the genome. ROHs analyses performed with these methods (i.e. RAD-sequencing, SNP arrays data, etc.) are then compared to results obtained with WGS data. The objective of the study is to propose case-specific guidelines based on populations characteristics and sequencing data, to help researchers aiming to perform population genetics analyses based on ROHs.

4. The genomics of adaptation to climate in European great tit (Parus major) populations
Presenter: Joanne Stonehouse, The University of Sheffield
Co-Authors: Slate, Jon The University of Sheffield

Wild great tit (Parus major) populations represent an attractive ecological model species to understand the genomics of climate adaptation. The species is distributed across most of Eurasia and is non-migratory, meaning different study populations have adapted to a wide range of local climatic conditions since the last glacial period. Genomic tools for this species include a high quality annotated reference genome and a 500K high density SNP chip that has been used to genotype many wild populations as part of the Great Tit HapMap Project. By combining local climate data with SNP genotype data from 20 different European populations, this study assessed the genomics of adaptation to different climatic conditions. Multivariate climate data for each study site were extracted from the WorldClim v2 database and principal components analysis was used to identify four principal components that explain most of the climatic variation. A Bayesian genome-environment analysis using the genomic and climate data identified putative climate adaptation genes, with 19 and 34 candidates 'decisively' associated with principal components 1 and 2 respectively. An enrichment analysis of biological function Gene Ontology (GO) terms identified over-represented terms and pathways among the genes under selection for climate. The observation that numerous genes and genetic pathways are associated with climate variables suggests that climate adaptation has occurred, at least partially, through microevolutionary change and that adaptation is polygenic and genetically complex. It may also indicate that there is substantial genetic variation that can help further adaptation to future changes.

5. Population genomics, insights of domestic adaptation and dispersal in the kissing bug, Rhodnius ecuadoriensis

Presenter: Luis Enrique Hernandez Castro, University of Edinburgh
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Understanding the genomic basis of disease vectors adaptation to the human habitat and their dispersal capabilities is paramount for effective and sustainable control interventions, yet both mechanisms remain poorly understood in many arthropod-borne diseases. Using the main Chagas disease vector in southern Ecuador and northern Peru as a model, we genotyped 2,552 SNP markers of 272 Rhodnius ecuadoriensis triatomines to investigate gene flow, signatures of local adaptation and drivers of population structure and connectivity among populations. Evidence of high triatomine gene flow (e.g., low FST values, and little ecotype genetic/phylogenetic variation) between domestic and silvatic populations in several sites across the study area suggested insecticide-base control can be hindered. Genomic scans, based on machine learning (e.g., Random Forest), multivariate statistics (e.g., redundancy analysis) and FST-heterozygosity outlier method, revealed putative SNP loci under a strong signal of local adaptation which mapped to some annotated regions in the Rhodnius prolixus genome. Our isolation-by-distance and resistance generalised mixed models with maximum-likelihood population effects parametrisation showed Rhodnius ecuadoriensis populations connectivity is driven by

landscape heterogeneity. Moreover, we were able to detect highly connected and isolated population clusters which need to be target differently by vector control programmes.

6. Combining population genomics with genome editing: Towards an integrative view of cold tolerance in Drosophila

Presenter: Sonja Grath, LMU Munich
Co-Authors: Königer, Annabella, LMU Munich
Ramnarine, Timothy J. S., LMU Munich
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Temperature is one of the major factors influencing the geographical distribution and abundance of many animals. As the body temperature of insects mainly follows the external environment, tolerance towards thermal extremes is essential for adaptation to new environments such those arising through range expansion or climate change. In our laboratory, we established Drosophila ananassae as model to elucidate the genetic basis of cold tolerance.
Here, I present two stages of our project. First, we identified candidate genes for cold tolerance by means of comparative transcriptomics and a genome-wide QTL mapping experiment with cold-tolerant and cold-sensitive fly strains. We further established molecular genetic tools for genome editing by CRISPR/Cas in this species. Second, as we still lack information on the function of these candidate genes in tissues particularly relevant for cold tolerance, we will use the tools developed in stage 1 to perform tissue-specific genome editing. We further aim to identify polymorphisms related to cold tolerance based on genome sequencing of strains originating from cold-tolerant and cold-sensitive populations. The results of these experiments will shed light on thermal adaptation of natural populations. In addition, the combination of state-of-the-art genomic and genome editing methods will provide new information on the function of different genes, both in the whole organism and individual tissues. These data will not only elucidate the function of genes and polymorphisms associated with cold tolerance, but will also allow the analysis of additional phenotypes and comparison with other species to identify general patterns of environmental adaptation.

Poster Room 2

7. Characterising and visualising gene families within Galaxy using GeneSeqToFamily and Aequatus

Presenter: Anil S. Thanki, Earlham Institute, Norwich - UK
Co-Authors: Soranzo, Nicola, Earlham Institute.
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The phylogenetic information inferred from the study of homologous genes helps us to understand the evolution of gene families and plays a vital role in finding ancestral gene duplication events as well as identifying genes that are under positive selection within species. We developed GeneSeqToFamily (Thanki et al. GigaScience 2018), a Galaxy

workflow based on the Ensembl GeneTrees pipeline which generates gene families, providing sequence alignments, associated phylogenetic trees, as well as details about exon conservation. This workflow helps users to run large-scale gene family analyses on a friendly web user interface, while still allowing flexibility in parameter configuration and tools choice. Results of the GeneSeqToFamily workflow can be visualised within Galaxy using the Aequatus plugin, which is part of a standalone Aequatus tool, providing an in-depth view at multiple scales from gene synteny preservation and to structure conservation through protein domains projection. Since publication, GeneSeqToFamily has been used to analyse gene families across plants, vertebrates and fish genomes. We are scaling up the GeneSeqToFamily workflow to analyse large numbers of species (>200) as well as species with large and complex genomes (> 100,000 genes). All tools used in the GeneSeqToFamily workflow and the full workflow itself are available to install from the Galaxy ToolShed. GeneSeqToFamily is also available for use from the UseGalaxy.eu public instance. Source code for GeneSeqToFamily and Aequatus is available from GitHub.

8. Is the guppy Y chromosome recently evolved from an X chromosome?
Presenter: Deborah Charlesworth, University of Edinburgh

The guppy Y chromosome carries the vast majority of genes present on its X counterpart, which can be explained by occasional recombination with the X. Sex determination in the guppy might therefore be controlled by a single gene or a physically small sex-linked region. Such sex-determining loci are known in other fish, as well as in other organisms, and can be explained by so-called “turnover” events, in which a sex-determining region evolves in a new genomic location, often on a different chromosome. Turnovers that cause the sex-determining region to become autosomal allow homozygotes to be generated. If the region is extensive and highly degenerated (a Y or W chromosome-like genome region), this will often prevent the spread of the new sex-determining factor. Loss of an established degenerated sex chromosome is therefore unlikely, and indeed turnovers appear to be commonest in organisms with homomorphic sex chromosomes whose sex-determining regions are physically small. However, loss of a Y can potentially occur if an active maleness factor appears on an ancestral X chromosome. I will describe evidence suggesting that such a non-canonical origin could account for the properties of the guppy Y chromosome.

9. Individual variation in reaction norms but no directional selection in reproductive plasticity of a wild passerine population
Presenter: Heung Ying Janet Chik, University of Groningen, Imperial College London
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In the plant—insect—insectivorous bird food chain, directional changes in climate can result in mismatched phenology, potentially affecting selection pressures. Phenotypic plasticity in the timing of breeding, characterized by reaction norm slopes, can help maximize fitness when faced with earlier prey emergence. In the blue tit, the timing of tree budburst influences food availability for chicks through caterpillar phenology and the resulting food abundance patterns. Thus, the timing of tree budburst might serve as a better proxy for cue

for egg-laying. Here we tested for the laying date—budburst and the clutch size—laying date reaction norms, and examined 1) the among-individual variance in reaction norm intercepts and slopes; and 2) the selection differentials and gradients on these intercepts and slopes. Using long-term data of oak (genus Quercus) budburst and blue tit (Cyanistes caeruleus) reproduction, we applied within-subject centering to detect reaction norms, followed by bivariate random regression to quantify among-individual variance in reaction norm properties and their covariance with fitness. Individuals significantly differed in intercepts and slopes of both laying date—budburst and clutch size—laying date reaction norms, and directional selection was present for an earlier laying date and a larger clutch size (intercepts), but not on plasticity (slopes). Results suggested that individuals have their own regimes for adjusting egg-laying and clutch size, and that heritability and stabilizing selection of reaction norms could be tested as a next step. This study provides further support of individual variation of phenotypic plasticity in birds.

10. Which is the Effect of Domestication on the Genome?

Presenter: IOANNA -THEONI VOURLAKI, Centre for Research in Agricultural Genomics (CRAG), Autonomous University of Barcelona (UAB), Vall Moronta s/n, Cerdanyola del Valles, Barcelona, 08193, Spain
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The civilization of the human's society has been relying on the domestication of plants and animals, establishing the crucial necessity to study it as an evolutionary and demographic model. As we know, domestication is a recent process, in comparison to the history of species, making the study of standing variants of significant importance since it is expected that the new mutations would not have the time to reach fixation under stationary models. Domestic environment can alter the selective effects of variants provoking different outcomes. Here, we are interested in studying the contribution of shared variants and comparing to the exclusive ones between two recent diverged populations differentiated by an environmental change that modified a proportion of the ancestral fitness effects in one of the populations (mimicking the domestication process). In this study, simulated data were generated under several evolutionary and demographic conditions considering weak and strong selective effects, mild or strong bottlenecks, or small versus large modifications of selective effects. The comparative analysis of these simulated data allows us to understand the impact of different scenarios on empirical observations. Our results indicate a high contribution of the shared variants to the global divergence of these populations, the strong effect of demographic events, and the capacity to detect weak positive selection in scenarios where an important number of functional variants switched their selective effect.

11. Using phased diploid genomes to test for natural mating in Candida albicans yeast from human and oak trees

Presenter: Sydney McCall, University of Georgia, Institute of Bioinformatics
Co-Authors: Bensasson, Douda, University of Georgia

The yeast, Candida albicans, is an opportunistic pathogen that inhabits most humans and is occasionally found on plants. It has also been considered a model asexual diploid in theoretical population genetics . Predictions about its molecular evolution have been difficult to test because of a lack of phased diploid genome sequence. Here we use recently-generated phased genome sequence for four strains of C. albicans from oak trees and human to test for recombination. More specifically, we used chromosome painting and compared orthologous sequences from the right and left arms of each chromosome. We discuss the prevalence of mitotic recombination and the evidence for possible mating in natural populations.

12. How variable is the Drosophila microbiome?
Presenter: Xiaomeng Tian, Institute of Population Genetics, Vetmeduni Vienna
Co-Authors: Stefanie Gerstbauer, Institute of Population Genetics, Vetmeduni Vienna
Viola Nolte, Institute of Population Genetics, Vetmeduni Vienna
Christian Schlötterer, Institute of Population Genetics, Vetmeduni Vienna

The strong influence of the microbiome on its host is well-documented. Drosophila provides an excellent system to study how the microbiome affects host phenotypes-a rather short generation time of the host in combination with a simple microbiome, which can be cultured. However, previous study focused on the composition changes of microbiome in Drosophila, the intraspecific genomic variation of different components of the Drosophila microbiome have been largely neglected. Here we studied the the variability of Acetobacter, one of the major components of the Drosophila microbiome. Using whole genome sequencing of individual strains, we address the following questions: How much sequence divergence can be found among Acetobacter strains from the same Drosophila population? How variable is the gene content of Acetobacter. Do Acetobacter strains from Drosophila kept in different environments differ functionally? This work will provide a detailed insight into microbiome variability in Drosophila.

Poster Room 3

13. Comparing the effect of SNP density on quantitative analyses of a wild population
Presenter: Caelinn James, University of Edinburgh
Co-Authors: Knott, Sara, University of Edinburgh, Narravo, Pau, University of Edinburgh, Pemberton, Josephine, University of Edinburgh

The Soay sheep (Ovis aries) of the St. Kilda archipelago are an unmanaged population from the primitive domestic sheep breed introduced to the island over three thousand years ago. Those sheep residing in the Village Bay area of Hirta (one of the islands in the archipelago) have been subjects of an individual based study since 1985, which has been recording phenotypic data of the sheep (e.g. birth weight or leg length), their pedigree, census location, as well as environmental factors such as vegetation density. In addition, the sheep are also genotyped; 7,700 Soay sheep have been genotyped on the Illumina OvineSNP50 Genotyping BeadChip, which contains 50K SNPs. Using these data, we will estimate SNP-

based trait heritabilities and identify variants associated with observed phenotypic differences.

Recently, 188 Soay sheep have been genotyped using the Illumina Ovine Infinium HD SNP BeadChip containing 450K SNPs, allowing for imputation from the low-density SNP panel to the higher density panel for the rest of the sheep. We repeated the heritability estimation and mapping analyses performed with the lower-density genotypes with the high-density imputed genotype data, and compared the results to answer the following questions: (I) do heritability estimates differ between the two SNP densities?, (II) can we recapitulate the associations found with the low-density genotype panel with the high-density one?, and (III) do we find novel associations with the high-density SNP panel? Overall, we aim to elucidate the importance of SNP density when investigating quantitative genetic traits in this sheep population.

14. X-linked meiotic drive can boost population size and persistence
Presenter: Carl Mackintosh, UCL
Co-Authors: Pomiankowski, Andrew, UCL, Scott, Michael, UEA

X-linked meiotic drivers cause X-bearing sperm to be produced in excess by male carriers, leading to female-biased sex ratios. Here, we find general conditions for the spread and fixation of X-linked alleles. Our conditions show that the spread of X-linked alleles depends on sex-specific selection and the way they are transmitted rather than the time spent in each sex. Applying this logic to meiotic drive, we show that polymorphism is heavily dependent on sperm competition induced both by female and male mating behaviour and the degree of compensation to gamete loss in the ejaculate size of drive males. We extend these evolutionary models to investigate the demographic consequences of biased sex ratios. Our results suggest driving X-alleles that invade and reach polymorphism (or fix and do not bias segregation excessively) will boost population size and persistence time by increasing population productivity, demonstrating the potential for selfish genetic elements to move sex ratios closer to the population-level optimum. However, when the spread of drive causes strong sex ratio bias, it can lead to populations with so few males that females remain unmated, cannot produce offspring and go extinct. This outcome is exacerbated when the male mating rate is low. We suggest that researchers should consider the potential for ecologically beneficial side effects of selfish genetic elements, especially in light of proposals to use meiotic drive for biological control.

15. Muller's ratchet-like effect in a highly recombining bacterial species
Presenter: Elise Tourrette, None
Co-Authors: Falush, Daniel, Institut Pasteur of Shanghai

Helicobacter pylori can be subdivided into four main subpopulations: African, Asian, European and Middle Eastern ones (Falush et al. 2003, Montano et al. 2015). The Asian population has been formed after an out-of-Africa bottleneck and the European and Middle Eastern ones are the result of a hybridization between the African and Asian populations. The African and Asian populations present similar levels of synonymous diversity while the

level of non-synonymous mutations is higher in the Asian population (higher dN/dS). The European and Middle Eastern populations present a higher level of synonymous diversity while their dN/dS lie between the values of the African and Asian populations. Moreover, the non-synonymous mutations show a deficit in Asian ancestry in the European and Middle Eastern subpopulations. This excess of non-synonymous mutations after a bottleneck is similar to a Muller's ratchet effect, which is the accumulation of deleterious mutations in asexual populations. However, H. pylori is a highly recombining species which should remove this effect. In this poster, we present simulations of the evolution of the African and Asian populations after the bottleneck and then during their admixture, aimed at finding the set of parameters to reproduce these observations and then explain them.

16. The effect of habitat loss and fragmentation on genetic variation: insights using spatio-temporal models

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Recent anthropogenic activities have largely contributed to habitat loss and fragmentation across the world, leading to 70% of worldwide remaining forests to be within 1 km of forest's edges (Haddad et al., 2015). In addition, it is estimated that 17% of the tropical moist forests have disappeared between 1990-2019 (Vancutsem et al., 2020), and predictive models suggest that by 2050 undisturbed forests will disappear entirely in large tropical humid regions. Habitat loss and fragmentation is expected to affect species population genetic variation, by decreasing within-population genetic diversity and increasing genetic differentiation among isolated populations. Most species worldwide have a geographically restricted dispersal, which results in a positive relationship between genetic and geographic distances, known as "isolation-by-distance" (IBD). In this work we used spatially explicit individual-based simulations to investigate i) how spatial genetic diversity changes within habitat fragments due to habitat contraction (local scale), taking into account habitat quality and edge-effect; and ii) how fast IBD pattern are lost following habitat loss and fragmentation (landscape scale). The aim of the present study is to provide a measure that quantify the additive effect of multiple edges in heterogeneous habitats on genetic diversity in order to formally test molecular edge effect in natural populations. Moreover, we provide insights on the rate at which IBD is lost after habitat loss and fragmentation and how past demographic events can influence such patterns.

17. Estimating effective population size via linkage disequilibrium with overlapping generations

Presenter: Luis Alberto García Cortés, Departamento de Mejora Genética Animal. Instituto Nacional de Investigación Agraria (INIA), Madrid (Spain)
Co-Authors: M. Ángeles R. de Cara, CIBIO (Centro de Investigação em Biodiversidade e Recursos Genéticos), Universidade do Porto (Portugal)

In order to maintain genetic diversity, knowledge of the effective population size (Ne) is crucial. This allows us to establish the adaptive potential of the population. There are several ways to measure it, using ecological or genetic data. Recently, given the abundance of marker data, it has become commonplace to estimate it via linkage disequilibrium. This estimate has the advantage of only requiring one sample in time, but it is very sensitive to other genetic parameters. We will start by addressing the issues of using linkage disequilibrium to estimate Ne and how to better estimate it for simple populations without age-structure. We will show results using three similar but different estimates available in the literature, and their dependence on map distance and population size. We will then show how to address the issue of overlapping generations, by deriving sets of equations. In both cases, we compare analytical with simulation results. Lastly, we will show an example of how to implement our method into real data, using publicly available data of the white-crowned sparrow, whose life-history traits have been well characterised.

18. Conservation genetics for the management of black rhinoceros (Diceros bicornis michaeli) in Tanzania.

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The genetic structure of a population indicates the potential for local adaptation, and maintenance of genetic diversity through gene flow among populations allows organisms to evolve and contend with continuous environmental changes. The loss of genetic diversity may reduce reproductive fitness, which may increase risks of extinction. Investigation of patterns of genetic variation provides information on the dispersal of species, mating behaviors and population boundaries, which can allow conservation managers to estimate rates of inbreeding and to design specific translocation strategies to increase evolutionary potential. Furthermore, a more comprehensive understanding of the distribution of genetic variants among populations could provide critical forensic evidence in the case of poached animals. However, despite its high conservation profile, still nothing is known about existing levels of genetic variation of the remaining populations of the Eastern black rhinoceros (Diceros bicornis) in Tanzania. Therefore, the aim of this study is to apply conservation genetics techniques to generate information on the genetic status of all extant six Black rhinoceros populations from four protected areas in Tanzania (Serengeti National Park, Ngorongoro Conservation Area Authority, Mkomazi National Park and Grumeti Reserve) and one neighbor population from Kenya (Maasai Mara) for: 1) estimating rates of inbreeding and movement of individuals between populations; 2) establishing population viability models for management. This information will be used to formulate a genetically viable

management strategy for managing black rhinoceros populations in Tanzania, including designing specific translocation strategies for maximizing distribution of genetic variation across subpopulations.

Poster Room 4

19. A needle in a stack of genomic data: a few outlier loci reveals subtle genetic structure in a highly dispersive deep-sea species

Presenter: Adrien Tran Lu Y , Institut des Sciences de l'Evolution (ISEM) ,CNRS
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Understanding population connectivity is a central question to evolutionary ecology and its applications in terms of conservation and management. The use of indirect genetic approaches has been especially useful in cases where direct observation of migration is not possible, like in most marine environments including the deep sea. Hydrothermal vent species have drawn a lot of attention due to the oasis-like distribution of their unique habitat. Nevertheless, previous studies have met limited success to infer fine-grained structure because many approaches have used few loci and generally provide a global but noisy picture of the demo-genetic events affecting them. By offering access to thousands of markers across the genome, reduced-representation sequencing now makes it possible to identify loci with greater information on ancestry and fine-scale population structure compared to the genome-wide average.
In this study, we explored range-wide population connectivity of the deep-sea hydrothermal vent snail Ifremeria nautilei with restriction-site-associated DNA (RAD) sequencing. The global picture unraveled with ~10,000 markers is two panmictic genetic clusters, isolated in two distinct clusters of oceanic basins separated by 2000 km (Fst = 0.41). The application of several outlier detection methods allowed us to pinpoint two phenomena that would have remained undetected otherwise, namely local introgression from one population into the other and reveal limitation to demographic connectiviy at a fine-scale. These results illustrate how a few outlier loci in a large genomic stack of neutral markers can carry useful information about cryptic barriers to dispersal in high gene flow marine species.

20. NEOF: NERC Environmental Omics Facility

Presenter: Chris Owen, Liverpool - CGR

NEOF is a new £13m facility funded by the Natural Environmental Research Council (NERC) to provide expert advice, training and state-of-the-art instrumentation that will allow environmental and evolutionary biologists to address new questions using omic methods. Members of our team will be available to discuss potential projects at the poster session. Our aim is guide users from all levels through experimental design, sample preparation, data generation and analysis, coupled with training in our visitor facility, workshops and online courses. We also provide pilot funding to test adventurous ideas and to introduce and train new users. We will announce the first pilot round directed at early career researchers. In

addition to the latest genomic platforms (Illumina Novaseq, Pacbio Sequel II, 10X, etc) we also wish to broaden the scope of omics methods in the population genetics community through proteomics and metabolomics.

21. Population genomics shows last European stand of Artemisia laciniata is diverse despite population size

Presenter: Christina Hedderich, Vienna Graduate School of Population Genetics, University of Vienna Department of Botany and Biodiversity Research
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The advances in sequencing and bioinformatics approaches are opening new avenues for the use of historical samples in genomic studies. Herbarium specimens are affected by preservation history or sample age often leading to high DNA fragmentation. Conservation efforts of critically endangered species can gain important insights from the use of genomic approaches, and the information obtained can be significantly enhanced by incorporating museomics data from already extinct populations. Artemisia laciniata (Asteraceae) is a critically endangered species in Europe, with a single relict population surviving in Eastern Austria; additional populations are present in the cold-dry steppes of South Siberia and Eastern Asia. Using RADseq of accessions from extinct populations in Central Europe (> 100 years), together with representatives of extant populations in Europe and Asia, generated around 40,000 loci. Despite DNA fragmentation, analyses of DNA damage indicated that the reads from the historical samples suffered only little degradation, but in general resulted in lower yield as compared to modern accessions. Clustering analyses revealed a clear genetic separation between the extant European and Asian samples, whereas the extinct European populations rather shared genetic ancestry with the Asian populations. Despite the low population size in the relict European population, inbreeding was estimated to be rather low in comparison to that of the extinct populations. These results provide solid argumentation for further conservation efforts, both ex-situ and in-situ, and importantly they demonstrate a major enhancement of conservation genomic studies by using museomics data.

22. Dosage compensation is associated with female consequences of an X-linked mutation

Presenter: Jack Rayner, University of St Andrews
Co-Authors: Thomas Hitchcock, Nathan Bailey

Recent theory indicates that dosage compensation mediates sexual antagonism over X-linked genes. This expectation is based on the assumption that dosage compensation scales phenotypic effects between the sexes, which is largely untested. We evaluated this by quantifying transcriptome variation associated with a recently arisen, male-beneficial, X-linked mutation across tissues of the field cricket Teleogryllus oceanicus, and testing the relationship between the completeness of dosage compensation and female gene expression effects. Dosage compensation in T. oceanicus was variable across tissues but generally incomplete, such that relative expression of X-linked genes was typically greater in females. Relative expression of X-linked genes was also unexpectedly high in adult somatic tissues. Seeming to support the assumption that dosage compensation scales phenotypic effects between sexes, we found tissues with less complete dosage compensation showed female-skewed effects of the X-linked allele. In gonads, where expression of X-linked genes was most strongly female-biased, ovaries-limited genes were much more likely to be X-linked than were testes-limited genes, supporting the view that incomplete dosage compensation favours feminisation of the X. Our results are consistent with the expectation that sex chromosome dosage compensation scales phenotypic effects of X-linked genes between sexes, and thus empirically substantiate this assumption underlying the theoretical role of dosage compensation in determining the dynamics of sexual antagonism on the X.

23. Inversion detection across a species range: a case study in Littorina saxatilis
Presenter: James Reeve, University of Gothenburg
Co-Authors: Faria, Rui, CiBio University of Porto
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Inversions are thought to play a major role in responses to divergent selection, acting as large effect loci that can store multiple genes involved in local adaptation to different environments. Recently, 17 putative inversions in the sea snail (Littorina saxatilis) have been detected along a beach in Sweden, some of which are thought to contribute to divergent adaptation between a predation resistant “crab” ecotype and a dislodgement resistant “wave” ecotype. Phenotypic responses to this environmental contrast have been observed at many sites across Europe, but the role of these inversions in differentiation is unknown at a broader scale. This project uses heterozygosity estimates from whole genome data collected from across the North Atlantic coastline to put the inversions detected in Swedish snails into a broader geographical context.

24. HMMploidy: inference of ploidy levels from short-read sequencing data
Presenter: Matteo Fumagalli, Imperial College London
Co-Authors: Soraggi, Samuele, University of Aarhus

The inference of ploidy levels from genomic data is important to understand molecular mechanisms underpinning genome evolution.
However, current methods based on allele frequency and sequencing depth variation do not have power to infer ploidy levels at low- and mid-depth sequencing data, as they do not account for data uncertainty.

Here we introduce HMMploidy, a novel tool that leverages the information from multiple samples and combines the information from sequencing depth and genotype likelihoods. We demonstrate that HMMploidy outperforms existing methods in most tested scenarios, especially at low-depth with large sample size. HMMploidy further allows for local inferences of ploidy change to detect within-chromosome variations.
We apply HMMploidy to sequencing data from the pathogenic fungus Cryptococcus neoformans and retrieve pervasive patterns of polyploidy and aneuploidy, even when artificially downsampling the data.
We envisage that HMMploidy will have wide applicability to low-depth sequencing data from polyploid and aneuploid species.

Poster Room 5

25. Ortholog misidentification and the animal tree of life.
Presenter: Charley McCarthy, University of Nottingham
Co-Authors: Mulhair, Peter, University of Leeds; Siu-Ting, Karen, Queen's University Belfast; Creevy, Chris, Queen's University Belfast; Pisani, Davide, University of Bristol; O'Connell, Mary, University of Nottingham

Our understanding of how complex tissues and systems have evolved within animals is contingent upon a clear reconstruction of animal evolutionary history. However, there remain conflicting hypotheses regarding important nodes within the animal tree of life (ATOL), such as the root of the tree and relationships amongst the Eumetazoa (“true animals”). Some of these conflicts have arisen due to various analytic errors, such as mismodelling of sequence evolution or sampling biases. We are currently examining the effect of ortholog misidentification on conflicting hypotheses of animal evolution, using an approach which was previously demonstrated to resolve conflicting hypotheses within Lissamphibia. We applied this approach to examine whether gene family data in ATOL datasets can recapitulate uncontentious relationships within the ATOL, e.g. the monophyly of Bilateria. First, we re-analysed 4 previously-published ATOL datasets which supported rooting the ATOL at either Ctenophora (comb jellies) or Porifera (sponges). We find that these datasets - regardless of their eventual tree topology - contain gene families which strongly recapitulate the monophyly of Ctenophora but insufficiently recapitulate other animal groups. We then constructed a reduced 20-taxa ATOL dataset which was enriched for orthologous signal across the ATOL, using multiple ortholog detection methods and filtering for truly-orthologous gene families. Preliminary analyses suggest that this approach retains gene families with greater recapitulation of major animal relationships, and we are currently investigating the impact this has on downstream phylogenetic inference.

26. Prevalence and polymorphism of a mussel transmissible cancer in Europe
Presenter: Maurine Hammel, ISEM, Univ Montpellier, CNRS, EPHE, IRD, Montpellier, France and IHPE, Univ Montpellier, CNRS, Ifremer, Univ Perpignan, Via Domitia, France
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Cancer cells are egoist entities that derived from and escaped the immune system controls of a multicellular organism. Cancerous cell lines usually become extinct with the death of their host; however some cell lines -called transmissible cancers- acquired the potential to infect a new host from the same or even another species. We studied the population genetics of one of those newly discovered transmissible cancers affecting Mytilus mussels. Two independent Bivalves Transmissible Neoplasia (BTN) lineages originating from M. trossulus have been described. One lineage, MtrBTN1, observed in East Pacific M. trossulus populations and the other, MtrBTN2, in European M. edulis and South American M. chilensis populations. Here, we performed an extensive screening of genetic chimerism, a hallmark of transmissible cancer, by genotyping SNPs of blood and mantle samples of European mussels (M. edulis, M. galloprovincialis and their hybrids). We detected MtrBTN2 at low prevalence in M. edulis, and also in M. galloprovincialis and hybrids although at a much lower prevalence. We then analyzed the genotypes of 6 tumors from M. edulis hosts and observed a diversity of genotypes that appeared more introgressed or more ancestral than MtrBTN1 and reference M. trossulus individuals. The observed polymorphism is most likely due to somatic null alleles caused by structural variations or point mutations in primer-binding sites leading to enhanced detection of the host allele. Also, despite low prevalence, multiple divergent sub-lineages (confirmed by mtCOI sequences) are co-spreading in the same geographic area, suggesting a quite long diversification of MtrBTN2 since its emergence.

27. A selfish chromosome creates interpopulation incompatibilities
Presenter: Rudi Verspoor, University of Liverpool
Co-Authors: Price, Tom, University of Liverpool
Hurst, Greg, University of Liverpool
Paterson, Steve, University of Liverpool
Betancourt, Andrea, University of Liverpool

Selfish Genetic Elements (SGEs) can rapidly spread in and between populations, influencing the ecology and evolution of species. In Drosophila subobscura, inter-population hybrid males suffer severe fitness costs that could prevent a sperm killing selfish X-chromosome from spreading. Genetic conflict causing co-evolution seems to be creating incompatibilities between populations- potentially driving the early stages of speciation.

28. Is there a zinc/salt tolerance trade-off in Silene uniflora adapting to mine spoils.
Presenter: Sarah Coates, MEFGL lab, School of Natural Sciences, Bangor University
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There are an increasing number of threats to plants across the globe through anthropogenic activities such as air pollution, mining, urbanisation and deforestation. Rapid adaptation can enable the survival of plant species in response to anthropogenic disturbances, however, the predictability of this process and its underlying genetic basis is not well characterised. Trade-offs can occur between certain traits during adaptation, such that adapting to novel environments causes a fitness reduction in the parental environment. Being able to understand the basis of these trade-offs and predict them will help broaden our understanding of the consequences of adaptation. Silene uniflora (Sea Campion) is a small perennial wildflower found mainly on the coasts of the UK. It is an ideal candidate for the study of rapid adaptation, as multiple populations have independently colonised heavy metal contaminated mine sites in the last 250 years, providing replicates of the same adaptive process. We found that two zinc-resistant populations share expression changes in response to zinc associated with tolerance, suggesting that these mine-adapted populations have evolved convergent mechanisms to cope with zinc toxicity. To test whether there is a trade-off between salt and zinc defences, we will investigate the extent to which zinc resistant mine populations are less resistant to salt than nearby coastal populations. We will also characterise the underlying expression changes associated with salt tolerance in the species.

29. The effect of population density on gene expression in adult Drosophila simulans
Presenter: Tejashwini Hegde, Institute of Population Genetics, Vetmeduni Vienna
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Density is an important environmental factor as it affects the life-history traits, growth rate, disease transmission etc. within a population. Studying density-dependent effects in holometabolous insects such as Drosophila simulans, is particularly interesting as the larval density also affects many adult phenotypes. The main aim of this study is to understand the plasticity in gene expression of adult males in response to larval density. The time of eclosion is substantially more variable for high larval density than for low larval density. We accounted for this by measuring gene expression in adult flies collected at one day intervals, resulting in one sample for low density and three samples for high density experiments. A large number of genes was differentially expressed between high and low density samples. Significantly fewer differences were observed between high density samples collected at different time points, which is surprising as early eclosing flies are exposed to fewer waste products (e.g. urea) than late eclosing flies. We will link gene expression changes to previously observed high-level phenotypic differences between high and low density conditions. In particular the hypothesis of insufficient protein availability at high larval densities will be tested.