Quantitative genetics with structured populations.

Matthew Ackerman

Quantitative genetic parameters, such as additive or dominance variation, are often difficult to estimate with classical methods; experimental crosses may be difficult or impossible and pedigrees may contain errors, be of limited depth, or be unavailable. The advent of inexpensive whole genome sequencing has made it possible to estimate these parameters by sequencing individuals from population samples, yet this approach is hampered by both cryptic population structure within samples and inaccurate genotype calling in low coverage sequence. We created a program, mapgd, which accurately recovers maximum likelihood estimates of genotypic probabilities and several population genetic parameters from low coverage sequence sampled from populations with non-random mating, most importantly recovering the correct estimators of additive and dominance variation. However, hidden population structure remains problematic for our method. Several techniques for removing the effects of this hidden structure are investigated through simulations, focusing on the influence of population structure on the prior probability that an individual possesses an allele. Interestingly, in simulated panmictic populations of realistic size, techniques which adjust these priors significantly improve parameter estimation. Finally we show that our program can be applied to human population genomic data, although this analysis remains in its early stages.

The Strength of Selection Against Neanderthal Introgression

Ivan Jurić, **Simon Aeschbacher**, and Graham Coop

Hybridisation between humans and Neanderthals has resulted in a low level of Neanderthal ancestry scattered across the genomes of many modern-day humans. After hybridisation, on average, selection appears to have removed Neanderthal alleles from the human population. Quantifying the strength and causes of this selection against Neanderthal ancestry is key to understanding our relationship to Neanderthals and how populations remain distinct after secondary contact. We developed a novel method for estimating the genome-wide average strength of selection and the density of selected sites using estimates of Neanderthal allele frequency along the human genome. We confirm that East Asians had somewhat higher initial levels of Neanderthal ancestry than Europeans even after accounting for selection. We find that most of the purifying selection against Neanderthal ancestry is best understood as acting on many weakly deleterious alleles. The majority of these alleles were likely effectively neutral — and segregating at high frequency — in Neanderthals, but became selected against after entering human populations of larger effective size. These alleles potentially imposed a heavy genetic load on the early-generation human–Neanderthal hybrids. Our work suggests that differences in effective population size may play a more important role in shaping levels of introgression than previously thought.

Exomes of historical and modern rabbit populations reveal parallel adaptation to myxoma virus across two continents

Joel M. Alves
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In 1950, the myxoma virus was used as a biological weapon to control invasive populations of European rabbit in Australia. Initially, the virus killed 99% of the infected animals and decimated entire populations. Two years later, the virus reached Europe causing a similar outcome. In both continents, a rapid evolution of rabbit resistance was observed. We investigated the genetic basis of resistance in Australia, France and the UK by sequencing the exomes of rabbits collected before the pandemic and modern samples. We found six genes where the same genetic variants had changed in frequency over the last 60 years. All of these genes are linked with the poxvirus replication or immunity. This suggests that the evolution of resistance to myxoma virus involved parallel genetic changes in the two continents.

Codweb: whole-genome sequencing uncovers extensive reticulations among Atlantic, Arctic, and Pacific cod-fish

Einar Árnason and Katrín Halldórsdóttir
University of Iceland

We have performed whole-genome sequencing of Pacific, Arctic and Atlantic codfish including ecotypes of Atlantic cod. Phylogenetic network analysis showed extensive introgression of cod-fish. Their evolution appears more a web than a tree. Whole-genome sequencing furthers our understanding of extensive reticulations, selection, and adaptation among cod-fish, which are among the most important fish stocks in the world and has implications for management of marine resources in times of rapid climate change.

Quantifying the mutation and selection pressures on insertions and deletions in the great tit (*Parus major*) using a novel maximum likelihood approach.

Henry Barton and Kai Zeng

Insertions and deletions (INDELs) remain understudied, despite being the most common form of genetic variation after SNPs. This partly stems from the challenge of correctly polarising INDELs into insertions and deletions, and the problem that polarisation error confounds signals of selection. Consequently the role of INDELs in genome evolution is poorly resolved, especially in non-model organisms. The recent publication of a high-quality great tit genome provides the unique opportunity to study INDELs in a wild avian population. This is pertinent, with INDELs implicated in genome contraction in highly recombining areas such as avian microchromosomes. Such findings acredit this to neutral evolution, but little is known about the selective pressures on INDELs in wild populations. Thus, we extend the model of Glémin et al., (2015) in a maximum likelihood approach to estimate the mutation and selective parameters for deletions and insertions. Our approach accounts for polarisation error, overcoming issues plaguing previous interpretations of INDEL data. We applied the model to high coverage (~44X) whole genomes from 10 European great tits. We demonstrate stronger negative selection on deletions than insertions and see regional variation in mutational and selective pressures on INDELs. The success of this approach demonstrates its potential in other organisms.

How many traits can be optimised by selection?

Nick Barton
IST Austria

How many traits can be kept close to their optima by stabilising selection? This number is apparently limited to ~4Ne by the "drift load", yet this is hard to reconcile with the apparent complexity of many organisms. A more robust limit is set by the variance in reproductive success; this suggests that selection accumulates information most efficiently in the infinitesimal regime, when selection on individual alleles is comparable with random drift. While most variance in fitness may be due to alleles with large Ns, substantial amounts of adaptation may be due to weakly selected alleles.

Inference of distribution of fitness effects and proportion of adaptive substitutions from polymorphism data

Paula Tataru, Maéva Mollion, Sylvain Glémin and **Thomas Bataillon**

Inference of DFE and rate of adaptive molecular evolution -alpha -from patterns of polymorphism (SFS) and divergence data has been a longstanding goal of evolutionary genetics. An assumption shared by numerous methods inferring DFE and alpha from such data is that beneficial mutations contribute only negligibly to the polymorphism data. Accordingly, DFEs comprising only deleterious mutations tends to be estimated from SFS, and alpha is inferred using divergence data from an outgroup. Here, we develop a hierarchical probabilistic framework that extends on previous methods and also can infer DFE and a from polymorphism data alone. We use extensive simulations to examine the performance of our method. While an outgroup is needed to obtain an unfolded SFS, we show that both a full DFE, comprising both deleterious and beneficial mutations, and alpha can be inferred without resorting to divergence data. Inference of DFE from polymorphism data alone can in fact provide more reliable estimates, as it does not rely on strong assumptions about a shared DFE between the outgroup and ingroup species. Not accounting for the contribution of beneficial mutations to polymorphism data substantially biases estimates of the DFE and alpha. We illustrate these points using simulated data, and published chimpanzee exome data set.

The history of introgression across a hybrid zone inferred from two transcriptomes

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The grasshopper, *Podisma pedestris*, has two parapatric races which differ by a sex-chromosome fusion. The races form a hybrid zone in the Southern French Alps which formed after the end of the last glacial maximum.

From lab crosses (Barton 1980) we know that there is strong selection against hybrids and crosses of hybrids of hybrids. But there is only limited information on the nature genomic introgression happening. The fused chromosome cline is about 800 m wide, much narrower than the selection against hybrids would suggest, but much wider than expected for a neutral marker (5500 m approx). Using an approach developed by Lohse et al. (2016) we fit different parameter sets of the structured coalescent using polymorphism data from two transcriptomes. We are particularly interested in how the history of gene flow between the races corresponds to hypotheses of their colonisation history.

Barton, N. H. 1980. "The Fitness of Hybrids between Two Chromosomal Races of the Grasshopper *Podisma pedestris*." *Heredity* 45 (1): 47–59. doi:10.1038/hdy.1980.49

Lohse, K., M. Chmelik, S. H. Martin, and N. H. Barton. 2016. "Efficient Strategies for Calculating Blockwise Likelihoods Under the Coalescent." *Genetics* 202 (2): 775–86. doi:10.1534/genetics.115.183814

Can the infinitesimal model explain artificial selection?

Stefanie Belohlavy, Gemma Puixeu Sala, Frank Chan, Nick Barton

Much progress has been made in identifying loci of large effect that influence complex traits. However, typically much of the trait variance is not accounted for by identifiable loci. Instead, genetic variance is generally consistent with the infinitesimal model, which is based on very many genes of small effect. Thus, the number and distribution of effects of the loci responsible for the bulk of the variation remain unclear. We investigate whether complete sequence data from populations of mice artificially selected for tibia length allows us to reject the infinitesimal model, and hence to estimate the number of genes and the distribution of their effects.

Transcription-associated compositional skews in Drosophila genes

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Local deviations from Chargaff's second parity rule are commonly observed around replication origins, transcription start sites and splice sites. Here, we use a whole-genome dataset of nearly 200 haplotypes of fruitflies (*Drosophila melanogaster*) to investigate compositional skews in introns. After excluding intronic regions close to replication origins and splice sites, a mild excess of T over A and a stronger excess of C over G nucleotides is observed on the coding strand, indicating a transcription-associated mechanism as an underlying cause. We furthermore find a positive correlation between compositional skew and gene expression, consistent with transcription-associated mutation bias (TAMB). However, mutation rates inferred from singleton frequencies show no strand-specific bias for C *vs*. G and inconsistent bias for A *vs*. T nucleotides, suggesting directional selection for strand- specific nucleotide composition.

Human population genetics of Papua New Guinea

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Human populations in Sahul, the ancient continent encompassing Australia, Tasmania and New Guinea, appears to have been independent from the rest of the world for ~50ky. Yet little is known about the genetic history of these populations and how it compares to histories elsewhere. We genotyped 382 individuals from 85 different language groups across Papua New Guinea (PNG) on 1.7 million genome-wide sites. We find very strong population structure, with F_{ST} values between groups much higher than within areas of similar size in Eurasia, mirroring the enormous cultural and linguistic diversity of PNG. There is a strikingly sharp genetic divide between the highlands and the lowlands, with population structure in the highlands being largely independent of that in the lowlands. Combined with a major increase in effective population size in the highlands in the last 10ky and split times between populations within the same timeframe (inferred from relative cross coalescence rates between high-coverage genome sequences), our results suggest a Holocene population expansion in the highlands, most likely linked to the spread of agriculture and the Trans-New Guinea language family. The study thus reveals both similarities and differences between the genetic histories of populations in PNG and Eurasia.

The hidden history of *Wolbachia* coevolution with nematodes

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Symbiotic interactions shape the diversity of the natural world. *Wolbachia* are common alphaproteobacterial endosymbionts of terrestrial arthropods and nematodes. In most arthropods, *Wolbachia* behave as reproductive parasites, while in nematodes they may be essential, mutualist symbionts. The main group of nematode hosts, vertebrate-parasitic Onchocercidae ("filarial nematodes") are important human and veterinary parasites, and antibiotic treatment (killing the nematodes *via* killing of their symbionts) is already used in clinical practice. The filarial nematodes harbour a range of different strains of *Wolbachia* that are classified into different supergroups. Using genomic data we have tracked the evolution of the association between *Wolbachia* and their nematode hosts. *Wolbachia* DNA is frequently laterally transferred into the host genome. We have used these insertions to extend our study into the phylogenetic past to reveal the palaeobiology of extinct symbionts and the dynamics of symbiont acquisition and loss. Our analyses reveal at least two invasions of filarial nematodes by Wolbachia in filarial nematodes, and show that the supergroup D symbionts of *Brugia* and related species are latecomers to this important symbiosis. Several species have lost their symbionts, as evidenced by the presence of *Wolbachia* DNA in their nuclear genomes in the absence of a live *Wolbachia* infection.

Combining markers with different mutation rates to infer the population genetic consequences of fragmentation in dwarf birch

James S. Borrell, Nian Wang, Richard A. Nichols & Richard J. A. Buggs

Genetic analyses of fragmented tree populations seldom find the loss of genetic diversity predicted by simple population genetic models. Here, we investigate this paradox in populations of dwarf birch (*Betula nana*), which has declined substantially over recent decades and now persists in a handful of relict sites across the UK. We compare these populations with large unfragmented populations in Scandinavia, using genetic markers with differing mutation rates. We combine PCR-generated microsatellites (PCR-SSRs), RADseq generated transition and transversion SNPs, and novel sequence derived microsatellites generated from RADseq reads (RAD-SSRs). The UK populations show higher differentiation, but appear resilient to the extreme effects of genetic drift until surprisingly small effective population size. Genetic structure is best explained by recent population bottlenecks coinciding with historic shifts in land use. We find that small microsatellite datasets contain as much information as thousands of genome wide SNPs for many applications. Furthermore, combining information from multiple independent marker sets improved confidence in our conclusions; firstly because they are affected differently by genetic drift and gene flow, and secondly because they have different ascertainment biases. Finally, we describe how RAD-SSRs may overcome many of the limitations of PCR-SSRs, providing a method to generate a high mutation-rate marker set from RADseq data.

The effects of sex-biased gene expression and X-linkage on rates of adaptive sequence evolution in *Drosophila*

José L. Campos, Keira Johnston and Brian Charlesworth

A faster rate of adaptive evolution of X-linked genes compared with autosomal genes can be caused by the fixation of new recessive or partially recessive advantageous mutations (the Faster-X effect). This effect is expected to be largest for mutations that affect only male fitness, and absent for mutations that affect only female fitness. We tested these predictions in *Drosophila melanogaster* by using coding sequences of genes with different levels of sex-biased expression, and estimating the extent of adaptive evolution of nonsynonymous mutations from polymorphism and divergence data. We detected both a Faster-X effect and an effect of male-biased gene expression: male-biased genes on both the X chromosome and the autosomes evolve the fastest. However, contrary to predictions, a Faster-X effect occurred also for female-biased genes on the X. These genes showed, however, the highest recombination rates, which are associated with higher rates of adaptive evolution. When we correct for recombination, female-biased genes on the X do not show a Faster X-effect anymore. Results were similar either using germline or somatic gene expression data. The same analyses of non-coding UTR regions showed a Faster-X effect for all the groups of genes considered. UTRs showed also higher levels of positive selection than coding sequence regions.

Longshanks Selection Experiment I: Genomics of Selection for a Gain Trait

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Understanding how the genome evolves through recombination in response to selection is a central challenge in evolutionary genetics, in particular for "gain" traits with a polygenic basis. I will introduce a unique selection experiment dubbed "Longshanks" created by Campbell Rolian in Calgary. By selecting mice with increased tibia length relative to body mass for 20 generations, he has generated mice with as much as 20% longer tibiae than in a random-bred population on the same genetic background. This major increase in mean tibia length has evolved rapidly despite a small constant breeding population of 32 mice per replicate line. We have sequenced pooled Longshanks and control genomes to identify candidate loci, showing both parallel and independent selection response. By intersecting SNPs undergoing significant frequency changes with functional genomics data, we identified three candidate limb enhancers at two major developmental regulators, *Bapx1* and *Gli3.* Using transgenic reporter assays we showed that a maximum of 13 SNPs in these three enhancers could account for both increased enhancer activity (at *Gli3*) and decreased activity (at *Bapx1*, a bone repressor), both of which are consistent with increased tibia length. Our results demonstrate the power of a systems genetics approach to dissect organismal traits to the level of individual basepairs.

Inferring parameters of positive selection from the relation between synonymous diversity and rate of protein sequence evolution in *Drosophila*

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An analysis of sequence data on whole genomes from a Rwandan population of *Drosophila melanogaster* showed a negative relation between the nonsynonymous (NS) site divergence and synonymous site diversity for a gene. By applying estimates from DFE-alpha of the distribution of fitness effects of deleterious mutations in protein sequences and UTRs to models of background selection (BGS), we found that BGS explains only part of this relation. Using estimates of the rates of substitution of positively selected NS and UTR mutations, we fitted joint models of BGS and selective sweeps to the data, and obtained estimates of the mean scaled selection coefficients for both NS and UTR mutations, as well as the proportion of mutations at NS and UTR sites that are positively selected. The mean scaled selection coefficients (Ne x s) were in the hundreds, with larger values for UTRs than NS sites; the proportions of positively selected mutations were approximately 0.0001. There was evidence for a negative relation between the level of selective constraint on a protein sequence and its mean positive selection coefficient. We found that the rate of gene conversion has a large effect on the parameter estimates.

Evolution of sex-biased gene expression in a dioecious plant

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Separate sexes and sex-biased gene expression have repeatedly evolved in animals and plants, but the underlying changes in gene expression remain unknown. We studied two species in the plant genus Silene; in *S. latifolia* separate sexes (dioecy) and sex chromosomes evolved within the past 10 MY, while *S. vulgaris* is gynodioecious, with a polymorphism such that, although females are present in populations, many individuals are hermaphrodites. Using *S. vulgaris* hermaphrodites as an outgroup, one can examine expression changes in flower buds and in rosette leaves during the evolution of dioecy. We excluded genes with expression limited to one or the other sex, which may reflect presence/absence of sex-specific structures. Sex-biases of genes expressed in both sexes have evolved in the dioecious species. Expression changes were commoner in genes located on the sex chromosomes than the autosomes. Most changes occurred in females rather than males, with higher and lower expression in females leading to female-biased and male-biased expression, respectively. This supports a scenario in which sex-biased gene expression evolved during the evolution of dioecy to resolve intralocus sexual conflicts over the allocation of resources.

Do bacterial pathogens have smaller genomes than their non-pathogenic relatives?

Michael Casey, **Jane Charlesworth**, John Welch, Lucy Weinart

A major outstanding question in prokaryote genetics and evolution is how bacteria evolve to become pathogenic. Much work has focused on identifying genetic variants involved in the phenotype of pathogenesis, and for searching for known 'virulence factors' in newly sequenced genomes in order to predict the pathogenicity of novel isolates, with variable success. Here we take a different approach to this problem. It has been observed many times, across all major groups of bacteria, that pathogens tend to have smaller genomes than their non-pathogenic relatives, but this has never been rigorously tested. Here, we use a comparative phylogenetic analysis of pairs of pathogen and non-pathogen species to test this hypothesis, and to try to disentangle some potential confounding factors, such as ability to persist inside host cells, or whether the bacteria are obligate pathogens that require a host in order to replicate, and discuss some hypotheses that aim to distinguish whether this is a by-product or cause of pathogenicity, and which population genetic processes could explain the observed result.

The Anopheles gambiae 1000 Genomes Project: population genomics and vector control

Clarkson, C.S., Miles, A., Harding, N.J., Bottà, G. and Kwiatkowski, D.P. - on behalf of the *Anopheles gambiae* 1000 Genomes Project.

The *Anopheles gambiae* 1000 Genomes Project (Ag1000G) is conducting whole-genome deep sequencing of wild-caught malaria vectors from populations across Africa with three core objectives: discover genetic variation, describe population structure/history and to connect these with malaria epidemiology and vector ecology. Phase 1 of the project includes sequence data from 765 specimens collected in eight countries. These data have been used to discover over 52 million SNPs, on average one SNP every two accessible bases, providing the first genome-wide view of the spectacular diversity within natural populations. Data have been publicly released and constitute the largest open access genomic resource available for any vector species. Here we provide an overview of the Ag1000G phase 1 data resource and initial results of population genetic analyses, focusing on detecting selection and gene flow. Analyses of population structure reveal a complex mosaic, with incomplete speciation, geography, demography and selection all influencing gene flow across the species' range. We illustrate this with the *Vgsc* gene, a target site for insecticides, where we show multiple independent haplotypes were involved in selective sweeps, shared between species and between populations separated by thousands of kilometres, indicating an extraordinary potential for mutations to spread.

Genetic regulators of life cycle transitions in the brown alga *Ectocarpus*

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The brown algae are members of the supergroup chromalveolata, and as such are very distantly related both to animals and to green plants. This group of seaweeds evolved complex multicellularity independently of animals and green plants and is one of only a small number of eukaryotic groups that has acquired this level of developmental complexity. The life cycle of *Ectocarpus* involves an alternation between two independent multicellular organisms, the sporophyte and the gametophyte. We have shown that the identities of the two generations are not determined by ploidy, but rather are determined genetically. Several life cycle mutants are currently being studied, including the *ouroboros* and *samsara* mutants, which both exhibit complete conversion of the sporophyte generation into a gametophyte. The *ouroboros* and *samsara* mutations not only correspond to key developmental regulators but also represent a new class of homeotic mutant in which there is a switch between developmental programs at the level of the whole organism rather than at the organ or tissue level. Characterisation of *Ectocarpus* life cycle mutants at the molecular level is providing insights into how multicellular development programs may have been built on to pre-existing regulatory networks controlling life cycle progression.

UV sex chromosome evolution in seaweeds

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Although the core mechanisms of sexual reproduction (meiosis and syngamy) are highly conserved across eukaryotes, the mechanisms that determine whether an individual is male or female are remarkably labile. In genetically controlled sexual systems, gender is determined by sex chromosomes, which have emerged independently and repeatedly during evolution. We are using the brown algae, a group that diverged from plants and animals more than a billion years ago, to gain insights into the evolution of haploid (UV) sex chromosomes. We investigated the conservation of sex-linked gene content among six brown algal species, belonging to two lineages, the kelps and the Ectocarpales, focusing on gene movement in/out of the U and V-specific regions. We analysed the consequences of sex linkage for the evolution of genes that have been residing in the sex-specific region in the different lineages at different evolutionary times. Our results highlight the dynamic nature of the U and V gene content across the different species compared with autosomes. We discuss how U and V gene content evolution may be related with the specific life history traits of each lineage, focusing on the level of sexual dimorphism, life cycle and sexual system.

A rapid shift from dioecy to hermaphroditism in response to severe mate limitation in an annual plant

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Most flowering plants are hermaphroditic, but separate sexes have evolved independently in many plant lineages. Unlike most animal species, males and females of many dioecious plants retain the ability occasionally to produce flowers of the opposite gender, i.e. they show 'inconstant' sex expression. It has long been thought that selection for reproductive assurance under conditions where mates are limited would provide an advantage to inconstant individuals, eventually allowing a shift from dioecy back to hermaphroditism, but this hypothesis has never been tested. Here we report results from an experimental evolution study in which we removed males from populations of the dioecious annual plant, *Mercurialis annua*, thus subjecting the remaining females to strong mate limitation. We observed a rapid shift in the level of male-flower production by females, with mate-limited females evolving over four generations of natural selection to produce 52 times more pollen than those maintained in a control treatment with a 1:1 sex ratio. Despite this dramatic and rapid shift towards functional hermaphroditism in our experiment, we found that variation for male-flower production remained high, with many individuals still completely female in their sex expression. We discuss the significance of these results for our understanding of transitions among sexual systems in flowering plants.

The genetic basis of convergent iridescent colouration in *Heliconius* butterflies

Emma Curran, Nicola Nadeau

Iridescence, produced by the scattering of light from nano-scale structures, is responsible for some of the brightest and most impressive colours in nature. It plays an important role in visual communication in both animals and plants. Despite this, research into the genetic basis of colour has focused entirely on colour produced by pigments, rather than structures. The *Heliconius* butterflies of South and Central America are well-known for their brightly coloured mimetic warning patterns. On the Western slopes of the Andes in Colombia and Ecuador, co-mimics *Heliconius erato* and *Heliconius melpomene* display an iridescent blue which is absent in the rest of these species' extensive range. The iridescent races of these co-mimics form a hybrid zone with non-iridescent races of the same species. We sampled individuals of each species across this hybrid zone, and quantified iridescence using photography and measurements of scale structures. Here, we use genome-wide association mapping to scan whole genome sequence data for SNPs associated with variation in iridescence. This will allow us to close in on candidate genes responsible for iridescence in each species, and ask whether this convergent phenotype is also convergent on the genetic level.

No evidence for a role for inversions in maintaining species barriers between *Heliconius melpomene* and *Heliconius cydno*

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Chromosomal inversions are known to maintain species barriers by restricting recombination between loci that distinguish species pairs. We hypothesized that barriers between the hybridizing sympatric species *Heliconius melpomene* and *H. cydno* may have been maintained by inversions. We constructed fine-scale recombination maps and generated long sequence reads for populations of these species from Panama to measure the occurrence of inversions between these species. We find no substantial inversions that would be sufficient to generate species barriers, although some small inversions may be present. I will present these results and discuss their implications in the light of existing speciation theories.

Evolution of chiral shells from chiral cells

Angus Davison, University of Nottingham

While components of the pathway that establishes left-right asymmetry have been identified in diverse animals, from vertebrates to flies, it is striking that the genes involved in the first symmetry-breaking step have remained wholly unknown in the most obviously chiral animals, the gastropod snails. Why are almost all organisms invariant in their left-right asymmetry, except for snails? In this talk, I will discuss our search to find the symmetry-breaking gene in the pond snail, and the implications that this has had for understanding how mirror image snails evolve, and why chirality is normally invariant in other organisms.

A hidden Markov model approach to the analysis of ancient introgression

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Whole genome sequence analysis has indicated that gene flow or introgression between deeply divergent populations is more common than previously expected. For example, there is evidence of archaic introgression into modern humans from Neanderthals, Denisovans, and possibly from other archaic hominins both before and after leaving Africa. Where we have a genome sequence from the potential source population, as with Neanderthals, we can use that in an admixture model to analyse the pattern of introgression. However even without a direct source sequence we can look for a signal of local clusters of variants which derive from a deeply diverged lineage, as with the S* statistic (Plagnol and Wall, 2006, and later updates), which looks for divergent haplotypes in windows. Here we introduce a related hidden Markov model approach which explicitly models both divergence and introgression times, and supports model comparison between alternative serial introgression hypotheses. We demonstrate the application of this method to recently published indigenous Australian and Papuan genome sequences (Malaspinas et al., 2016).

Co-variation in levels of nucleotide diversity in homologous regions of the avian genome long after completion of lineage sorting

Ludovic Dutoit, Nagarjun Vijay, Carina F. Mugal, Christen M. Bossu, Reto Burri, Jochen Wolf & Hans Ellegren

Closely related species may show similar levels of genetic diversity in homologous regions of the genome due to shared ancestral variation still segregating in the extant species. However, after completion of lineage sorting such co-variation is not necessarily expected. On the other hand, if the processes that govern genetic diversity are conserved, diversity may potentially co-vary even among distantly related species. We aligned the genomes of two divergent bird species, collared flycatcher and hooded crow, and identified >600 Mb of regions of conserved synteny. From analyses of whole-genome re-sequencing data in large population samples of both species we found nucleotide diversity in 200 kb windows to be well correlated (Spearman's $\rho = 0.401$). The correlation remained highly similar after excluding coding sequences. To explain this co-variation, we suggest that a stable avian karyotype and a conserved landscape of recombination rate variation render the diversity-reducing effects of linked selection similar in divergent bird lineages. Our observations imply that genetic diversity is to some extent predictable.

Rapid adaptation in highly fecund populations

Bjarki Eldon and Wolfgang Stephan

Evolution can occur at a high pace, much higher than originally envisioned by Darwin. Examples abound. We consider the evolution of a single- or multi-dimensional trait in highly fecund populations with sweepstakes reproduction (HFSR). Many marine populations, forest trees, and pathogens such as viruses, may possibly be characterised by HFSR. The value of the trait may be influenced by one or many (in diploids) unlinked loci through different dominance mechanisms. If many loci affect the trait in a diploid population, the effects may be unequal. We allow for mutation, and random environmental effects. Using simulations we compare the time to reach optimum trait value between populations with versus without HFSR. The trait optimum may be fixed, or shift, as in response to environmental change. Our objective is to understand if and how the number of loci, dominance mechanism, and HFSR interact to drive adaptation. Recent genomic studies show that this polygenic view of adaptation is highly relevant. Our main results are that populations with HFSR can reach the trait optimum in considerably shorter time than populations without HFSR.

From ancient DNA to ancient cities in 5 seconds

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Recent studies have demonstrated that geographical origin can be accurately inferred from genomic data and prompted us to embark on the unresolved question of inferring the geographical origin of skeletal finds, thus far assumed to be synonymous with their burial site. Whereas geographical inference based on anatomical or morphological information is highly complex and error-prone, particularly when the remains are physically damaged or fragmented, using ancient DNA for localization entails different challenges due to the lack of intermediate samples over space or time, the small number of SNPs, and their spurious nature. We developed the ancient Geographic Population Structure (aGPS), an admixture-based method, which implements a genetic clustering approach that uses the relationship between admixture and geography to predict geographical locations. Applied to a genomic dataset of 94 ancient Eurasians (Pleistocene – Iron Age), aGPS localized ~55% of the samples within 0-200km from their burial site, ~30% within 200-1,000km, and the remaining within 1,000-3,250km, with an overall average accuracy of 553km. Applied to a new dataset that was not used for calibration, aGPS obtained a mean accuracy of 799km for 191 ancient samples. Biodiversity centers were localized mainly in central and eastern Europe. Biodiversity showed a positive correlation with time.

The genetic basis of erythrocyte sickling in deer

Alexander Esin, L. Therese Bergendahl, Joseph Marsh, Tobias Warnecke

Sickle cell anaemia is caused by a single point mutation in the β-globin gene. A small change with large consequences: mutant hemoglobin (HbS) molecules polymerize into tubules upon deoxygenation, coercing red blood cells (RBCs) into the characteristic sickle shape that first came to the attention of the medical community in 1910 when Herrick described "peculiar elongated and sickle-shaped blood corpuscles" in a patient with severe anaemia. Remarkably, however, sickled erythrocytes had already been observed 70 years earlier when Gulliver noticed a range of oddly shaped cells, many in the now familiar sickle form, in the blood of white-tailed deer (*Odocoileus virginianus)*. In this talk, I will describe our efforts to unravel the genetic basis of sickling in deer, to re-trace critical steps in beta-globin evolution across 14 deer species, to elucidate whether sickling is adaptive, and to explore similarities and differences to the human disease state.

Is there coadaptation between the nuclear and mitochondrial genomes?

Adam Eyre-Walker

The mitochondrial and nuclear genomes are expected to coevolve because most of the genes involved in mitochondrial function are encoded in the nucleus. But is the genetic variation we observe within a species coadapted such that beneficial mitochondrial and nuclear alleles are typically found together in the same individual? This question has been extensively studied in model organisms but the question has taken on a new significance because of mitochondrial replacement therapy (MRT) in humans. If beneficial combinations of alleles are typically found together then we might find that MRT leads to incompatibilities between the nuclear and mitochondrial genomes leading to a loss of fitness. I have performed a meta-analysis of the results from animal studies in which the mtDNA from one strain has been introgressed onto the nuclear background of another strain. Sometimes the effects are negative, suggesting coadaption, but often they are positive. Overall I find little evidence that introgressing mtDNA onto a new nuclear background is on average deleterious. This is consistent with population genetic theory. I conclude that mito-nuclear incompatibilities are unlikely to be a problem for MRT.

Genetic architecture of social organisation in the ant *Pheidole pallidula*

Emeline Favreau, Claude Lebas, Rodrigo Pracana, Eckart Stolle, Max Reuter, Yannick Wurm

Species that live in societies are immensely successful, and exhibit tremendous variation in how their societies are organised. While theory explains the costs and benefits of different forms of social organisation, we have little empirical knowledge of how changes in social organisation evolve.
To examine this, we focus on a fundamental social trait that varies across ants: the number of reproductive queens in each colony. In the Mediterranean ant *Pheidole pallidula*, colonies within the same population contain either one or multiple queens. Each social form is known to express specific phenotypic traits, such as worker aggressivity. Similar dimorphic social organisation convergently evolved in two phylogenetically distant ant species. In those species, social dimorphism is determined by "supergene" regions containing hundreds of genes.
We are performing a genome-wide association study to identify loci that differ between single- and multiple-queen *P. pallidula* colonies. This will clarify whether a supergene also determines social dimorphism in this species - and may thus be required for intra-specific variation in social organisation, or whether alternate genetic architectures are possible. The results will inform us about general molecular patterns underlying convergent evolution of phenotypes.

Predictable allele frequency changes due to habitat fragmentation in the Glanville fritillary butterfly

Toby Fountain, Marko Nieminen, Jukka Sir n, Swee Chong Wong, Rainer Lehtonen and Ilkka Hanski
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Describing the evolutionary dynamics of extinct populations is challenging as their genetic composition prior to extinction is generally unknown. The Glanville fritillary butterfly has a large extant metapopulation in the Åland Islands, but declined to extinction in the nearby fragmented SW Finnish archipelago during the 20th century. We genotyped historical samples from both the extinct and extant populations for 222 SNPs across the genome, including SNPs from candidate genes and putatively neutral regions. We found significantly reduced genetic diversity prior to the SW Finnish populations extinction. We also identified 14 outlier loci among candidate gene SNPs in which allele frequencies have changed more than expected based on changes in neutral SNPs. A comparison of these outlier allele frequencies in three independent contemporary datasets, where detailed demographic and ecological information was available, showed consistent allele frequency shifts in extinct populations with populations with high turnover and populations occupying fragmented landscapes. This indicates that extinct populations had an increase in alleles associated with good colonisation capacity before their extinction. These results highlight a potentially common situation in changing environments: evolutionary changes are not strong enough to fully compensate for the direct adverse effects of environmental change and thereby rescue populations from extinction.

Deep branch gene flow in an hybridization rich speciation process

Susana Freitas, Miguel Carretero, James Harris, Roger Butlin

Hybridization and gene flow during speciation processes has been given more attention in recent years with the appearance of more informative tools and data from NGS approaches. The study of how hybridization has shaped speciation is of crucial importance to the understanding of how adaptation and speciation work. Species in the model genus studied here are known to have hybridized recurrently in the past, giving rise to hybrid parthenogenetic species. Moreover, hybridization is still found to happen in the present between individuals of sexual species when in sympatry with parthenogenetic individuals.
Given this evidence frequent past, and ongoing hybridization, we asked if there was hybridization that lead to new hybrid sexual species or at least consistent and significant gene flow between different sexual clades. To test this, we used capture sequence data with markers across the genome so that we could identify the different contributions that past lineages might have made to current species.
We found strong evidence that hybridization happened more frequently than previously considered, and that not only parthenogenetic hybrid species but also sexual species reflect past hybridization events. The strong evidence of deep branch gene flow shows speciation is not self-regulating, and that diverging clades may still hybridize and exchange genetic variability.

Y-chromosome introgression among species and cytotypes of a dioecious herb

Paris Veltsos, **Jörn F. Gerchen**, John R. Pannell

Annual species in the genus *Mercurialis* show a remarkable diversity of cytotypes and sexual systems. *M. annua* and *M. huetii* in central Europe are diploid and dioecious, but tetraploid and hexaploid populations of *M. annua* on the Iberian Peninsula and in North Africa are either hermaphroditic or androdioecious, a rare mating system consisting of males and monoecious plants. Using a newly developed Y-linked genetic marker, we show that males in androdioecious populations of *M. annua* share a common Y-haplotype with two perennial species of *Mercurialis*, *M. reverchonii* and *M. elliptica*, while the Y-haplotypes of other annual *Mercurialis* species are more differentiated. These results are in conflict with phylogenies based on autosomal and chloroplast markers, which suggest that all annual *Mercurialis* species form a monophyletic clade that is clearly differentiated from perennial *Mercurialis* species. We interpret this pattern as a result of Y-chromosome introgression from perennial into annual *Mercurialis* species, possibly explained by a strong outcrossing advantage of the male phenotype, allowing it to quickly invade hermaphroditic populations.

Investigating the relationship between the substitution rate and genome size of bacteria.

Beth Gibson and Adam Eyre-Walker

Substitution rates are known to vary between species across all kingdoms of life. Here we explore variation in substitution rates across 25 species of bacteria. Intriguingly, we have found a correlation between the substitution rate per site per year and genome size, a relationship that has been noted previously over a broader range of microbes including viruses. Here, we investigate the factors that may influence this association, including the rate of mutation per generation, GC content, generation time and the efficiency of natural selection, i.e. the effective population size, and whether these can explain the observed relationship between genome size and substitution rate.

Can applied conservation genomics help to save the critically endangered Burmese roofed turtle (*Batagur trivittata*) in the wild?

F. Gözde Çilingir, Frank E. Rheind, Kritika M. Garg, Kalyar Platt, Steven G. Platt, David P. Bickford

One of the rarest turtle species, *Batagur trivittata*, is a Myanmar-endemic freshwater species and comprises < 10 breeding individuals in the wild. For the last 15 years eggs have been collected from the wild and reared in captivity (n>700). In this study, we describe the population genetic and pedigree structure of the turtles to enable re-introduction programs for re-establishing self-sustaining populations in the wild. We sampled almost half of the known World population and identified 1134 neutral SNPs throughout the turtles' genome. We revealed 4 distinct genetic clusters in the wild population and a total of 60 individuals with the lowest homozygosity by loci were released into the wild. Our pedigree analysis revealed that there are only 2 females and 4 males contributing to the current gene pool of the species. Genomic tools utilized in this study are directly shaping the species' management strategies and will help to contribute to other chelonian conservation projects.

The Facultative Sex Coalescent with Recombination and Gene Conversion

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Many diploid organisms are facultative sexuals, alternating between sexual and asexual reproduction. However, little is currently known concerning the distribution of neutral genetic variation among facultative sexual organisms except in very simple cases. Understanding this distribution is important when making inferences about rates of sexual reproduction, effective population size, and demographic history. Here we extend coalescent theory in diploids with facultative sex. A key result for non-recombining genomes is that when sex is rare, gene conversion becomes a significant force in reducing diversity within individuals. This can reduce genomic signatures of infrequent sex (i.e., elevated within-individual allelic sequence divergence) or entirely reverse the predicted patterns. We also create a new coalescent simulation algorithm that accommodates the complexities arising from partial sex, which also includes meiotic recombination and multi-site gene conversion. We use this algorithm to explore how linkage disequilibrium decays with high yet facultative sex rates. These models offer improved methods for assessing null patterns of molecular variation in facultative sexual organisms.

Discovery and microevolution of a new bacterial species

[1]Falk Hildebrand, [1]Jaime Huerta-Cepas, [2]Toni I Gossmann, [1]Sebastian Waszak, [1]Anna Pryszlak,[1]Ana Zhu, [1]Sonja Blasche, [1]Anita Voigt, [1]Peer Bork

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Recent advances in metagenomics have revealed considerable genetic variation among the microbes that populate the human gut. It has been shown that multiple strains of a bacterial species can coexist in a microbial community. However, accurately differentiating between strains in metagenomic samples is mostly not possible, even though pathogenicity usually is strain specific.
In this work we exploit single nucleotide variants (SNVs) in the human gut microbiome to identify and delineate distinct bacterial strains. We were able to reconstruct the genome of a highly abundant yet previously uncharacterized bacterial species found in an antibiotics-treated patient. From the assembled genomes of particular strains each highly abundant in specific gut samples, SNV profiles were created which span multiple time points, enabling us to explore rare SNVs arising and propagating within a bacterial population in its natural niche. Following up with culturing approaches, experimental validation of antibiotic resistance genes, we characterize a novel bacterial genus and report its unprecedented discovery under circumstances where the host environment undergoes unusual perturbations as a consequence of medical treatment.

Transposable element divergence in the *Drosophila pseudoobscura* group

Tom Hill and Andrea J. Betancourt

The evolutionary dynamics of transposable elements (TEs) has been shown to differ across the tree of life, even between closely related species that share a close evolutionary history. The *Drosophila pseudoobscura* group is known to exist sympatrically and occasionally hybridize, with increased divergence occurring in fixed inversions between species. Here, using sequence data from the *Drosophila pseudoobscura* species group, we assess the TE content of each species and find divergence in TE content, including an accumulation of TEs in *D. persimilis* relative to other species, despite evidence of gene flow and recurrent TE transmission between species. We also find rapid accumulation of TEs upon initial acquisition, followed by differing rates of extinction of TEs between species. Together, this suggests that in under 10 million years, the dynamics of the TEs within the genome can change rapidly, resulting in the rapid differentiation in TE dynamics seen between species and potentially even inversion types.

Using DSPS-HIV Simulations and Phylogenetic Analysis to Investigate Under-sampled Hosts in the UK Heterosexual HIV Epidemic

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The UK HIV Drug-Resistance-Database [UK-RDB] holds one-hundred thousand viral sequences for HIV+ individuals, who are sampled at diagnosis. From these sequences, we can reconstruct viral phylogenies of the UK HIV epidemic, informing us about how it is spreading and how it has changed over time. In particular, we look at 'clusters' of closely-related sequences to learn about recent and closely-linked transmissions.
In the UK heterosexual epidemic, there are six times as many clusters containing only women as only men. As sexual transmission of HIV between women is rare, this implies we are 'missing men' from the sampled transmission chain. This may be because men are diagnosed later, are not retained in treatment, or transmit more.
Here, we use a simulation of the UK HIV epidemic (using the DSPS-HIV, an individual-based S-I-R epidemic simulator which produces realistic viral sequences) alongside network analysis of HIV-RDB sequences to investigate the factors that influence viral transmission, and use these to replicate the observed tree structure with an excess of female-only clusters. We found that increased male transmission and delayed diagnosis play an important role in excess female-only clusters, illustrating the utility of simulations to help investigate factors leading to particular phylogenetic observations.

Demographic history of Northeast Africa revealed by genome wide population genetic data.

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Due to their geographic location, history, and linguistic diversity, Sudan and South Sudan cover an important region for human demographic history. This region has not yet been explored extensively using high-resolution genome-wide population genetic data. We investigate 18 Sudanese and South Sudanese populations by genotyping 221 individuals for approximately 5 million SNPs. We compare this novel dataset to available SNP data of surrounding geographic regions, as well as the 1000 Genomes and HGDP datasets. This dense marker set allows us to address questions relating to the demographic history of various populations from the area and reveals detailed population structure correlating with geography and linguistic affiliation.
In particular, we find a divide between the Northeastern populations versus the Western and Southern populations, which is mainly driven by non-African back-admixture. There have likely been several admixture and migration events from Eurasian sources into Northeast Africa, but the Semitic expansion into the area has had a strong demographic impact on the Sudanese populations, which was followed by cultural and linguistic transitions. In contrast to the Northeastern populations, populations of Western and South Sudan remained relatively isolated and seem to have a deep history in this part of Africa.

Genetic diversity and the efficiency of selection

Jenny James, David Castellano, Adam Eyre-Walker

Selection is expected to be more efficient in species that are more diverse because both the efficiency of natural selection and DNA sequence diversity are expected to depend upon the effective population size. We explore this relationship across a dataset of 751 mammal species for which we have mitochondrial polymorphism data. We introduce a method by which we can examine the relationship between our measure of the efficiency of natural selection, the non-synonymous relative to the synonymous nucleotide site diversity (π_N/π_S), and synonymous nucleotide diversity (π_S), avoiding the statistical non-independence between the two quantities. We show that these two variables are strongly negatively and linearly correlated on a log scale. The slope is such that as π_S doubles π_N/π_S is reduced by 34%. We also show that the slope of this relationship differs between the two phylogenetic groups for which we have the most data, rodents and bats.

Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast.

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Large structural variations (SVs) in the genome are harder to identify than smaller genetic variants but may substantially contribute to phenotypic diversity and evolution. Here we analyze the effects of SVs on gene expression, quantitative traits, and intrinsic reproductive isolation in the yeast *Schizosaccharomyces pombe*. We establish a high-quality curated catalog of SVs in the genomes of a worldwide library of *S. pombe* strains, including duplications, deletions, inversions and translocations. We show that copy number variants (CNVs) frequently segregate within closely related clonal populations, are weakly linked to single nucleotide polymorphisms (SNPs), and show other genetic signals consistent with rapid turnover. These transient CNVs produce stoichiometric effects on gene expression both within and outside the duplicated regions. CNVs make substantial contributions to quantitative traits such as cell shape, cell growth under diverse conditions, sugar utilization in winemaking, whereas rearrangements are strongly associated with reproductive isolation. Collectively, these findings have broad implications for evolution and for our understanding of quantitative traits including complex human diseases. See: http://biorxiv.org/content/early/2016/10/03/047266

What explains rare and conspicuous colours in a snail? A test of time-series data against models of drift, migration or selection

Kerstin Johannesson and Roger K Butlin

Conspicuous colour morphs of a prey species may be maintained at low frequencies alongside cryptic morphs. Negative frequency-dependent selection by predators using search images is often suggested without rejecting alternative explanations. Using a maximum likelihood approach we fitted predictions from models of genetic drift, migration, constant selection, heterozygote advantage or negative frequency-dependent selection to time-series data of colour frequencies in isolated populations of a marine snail, re-established with perturbed colour morph frequencies and followed for >20 generations. Snails of conspicuous colours (white, red, banded) are naturally rare in the study area (usually <10%) but frequencies were manipulated to levels of ~50% (one colour per population) in 8 populations at the start of the experiment in 1992. In 2013, frequencies had declined to ~ 15–45%. Drift alone could not explain these changes. Migration could not be rejected in any population, but required rates much higher than those recorded. Directional selection was rejected in three populations in favour of balancing selection. Heterozygote advantage and negative frequency-dependent selection could not be distinguished statistically, although overall the results favoured the latter. Populations varied idiosyncratically as mild or variable colour selection interacted with demographic stochasticity, and multiple mechanisms may contribute to maintaining the polymorphisms.

The genomics of polymorphism in a sexually selected trait

Isabel Santos Magalhaes †, Christoph Hahn †, Andrea Robson, Alan Smith, Marcel Haesler, Kyle Young, Martin Genner, **Domino Joyce***

Whether populations can divergence solely as a result of sexual selection in the face of gene flow, is contentious. Theory predicts that it may be possible if female preference and male signals diverge, but evidence is minimal and often inferred retrospectively, after speciation. In this study we address this issue by analysing three lekking populations of a bower-building fish which build bowers to attract females. We found that the 3 populations are indistinguishable genetically, in male nuptial colour, and morphologically, but have distinct bower shapes. We use behavioural observations and selection gradient analysis to demonstrate significant non-linear selection on bower shape across populations. Using RAD-seq data we were also able to identify regions of the genome which differ between these populations, and correlated specific bower traits with genomic regions important in differentiation. Our data demonstrate a role for differential selection on an extended phenotype in population differentiation, and indicate a genomic basis for the behavioural changes associated with this divergence.

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momi2: A new method to compute the joint sample frequency spectrum of multiple populations

John A. Kamm, Jonathan Terhorst, and Yun S. Song

The multipopulation sample frequency spectrum (SFS) is a popular statistic for inferring demographic parameters such as population sizes, split times, and migration rates. Despite methodological progress in recent years, existing methods suffer from numerical instability and high computational complexity when multiple populations are involved and the sample size is large. Here we present "momi2", a new method for exact computation of the SFS. momi2 improves on the numerical stability and runtime of previous methods by employing several computational and mathematical innovations, including techniques from Bayesian graphical models, and a novel application of the "lookdown construction" of the Moran model. Additionally, momi2 is highly flexible, using automatic differentiation to perform gradient-based optimization for a wide range of demographic models. In particular, momi2 automatically and transparently computes derivatives for user-defined Python functions specifying an arbitrary mapping from parameter values to demographic histories, and uses these derivatives to search for the maximum likelihood demography. Using both simulated data and the SGDP dataset, we demonstrate the ability of momi2 to infer complex multipopulation models with dozens of parameters, including bottlenecks, exponential growth, divergence times, and admixture proportions.

Patterns of linkage disequilibrium in flycatcher genomes clarify the causes and consequences of fine-scale recombination rate variation in birds

Takeshi Kawakami, Carina F. Mugal, Alexander Suh, Alexander Nater, Reto Burri, Linnéa Smeds, & Hans Ellegren

Recombination rate is highly heterogeneous across the genome of many animals and plants, but we still lack a clear picture of the underlying mechanism particularly in non-mammalian species. Here we estimate fine-scale population recombination rate based on the patterns of linkage disequilibrium across the genomes of two closely related flycatcher species (*Ficedula albicollis* and *F. hypoleuca*). We found 400-3,085 recombination hotspots in populations of these species. Location of hotspots was conserved between species, but intensity of hotspot activity was different between species as represented by differences in the strength of GC-based gene conversion. Recombination hotspots were significantly associated with CpG islands (CGIs) regardless of whether CGIs were at promoter regions or away from genes. Moreover, hotspots were associated with transposable elements (TEs), and, consequently, recombination rate was higher in regions containing TEs. The association between TEs and recombination may be indirect due to shared preferences of the transposition machinery and the recombination machinery, both of which opportunistically target accessible open chromatin regions. This association suggests that both distribution of TEs and fine-scale variation in recombination rate may be tightly associated with the evolution of epigenetic landscape.

Processing variant data for millions of samples

Jerome Kelleher and Gil McVean October 11, 2016

Encoding the history of a sample of individuals subject to recombination as coalescence records leads to very efficient simulation algorithms. A key property of this representation is that it is extremely compact, allowing very large simulations to be stored and processed. In this talk we discuss the possibility of using this approach to represent observed variation data. To illustrate the potential advantages, we simulated the ancestry of 1 million samples under the coalescent with recombination for a 100Mb chromosome with human-like parameters using msprime. The simulation required about 16 minutes to run and only 160MB to store the complete genealogical history and information on all 1.1 million variants. In contrast, encoding this information as a variant matrix in VCF format required 2.1TB of storage. We demonstrate that many queries are also far more efficient when using coalescence records than is possible using current state-of-the-art methods.

Estimating a set of coalescence records consistent with arbitrary input data is of course a challenging and computationally expensive problem. However, we believe that the advantages of substantially reduced storage and data processing times outweigh this cost. We present some initial results on estimating coalescence records from data.

Sperm Mediated Epigenetic Effects in Zebrafish (*Danio rerio*).

Kiehl, B; Anava, S; Rechavi O; Immler, S

A recent study in zebrafish (*D. rerio)* showed that changes in social environment, specifically the intensity of sperm competition, do not only lead to modifications in sperm motility and velocity, the paternally experienced environment of sperm competition also translates into differences in hatching timing and larval survival. I aim to assess where in the sperm the trigger for this mechanism lies and how this is transferred into the offspring. I investigate the role of sperm in parental effects and aim to identify potential mechanisms causing paternal effects (i.e. sperm-mediated epigenetic mechanisms). First, I isolate RNA from sperm and create a transcriptome for the zebrafish sperm (project reference), then perform experiments to manipulate the paternal effects and look into the underlying molecular mechanisms. I assess the causes for variation in epigenetic effects in males by manipulating eg sperm competition levels among males and comparing the epigenetic patterns in their sperm using 3 approaches: (A) comparing the RNA pro le (B) investigating chromatin structures and (C) determining methylation patterns. Experimental tools are combined with transcriptomics and epigenomics to address these questions using a model organism.

Selection acting on toll-like receptors in bank voles

Agnieszka Kloch
University of Warsaw

Parasite-driven selection is a key factor influencing the evolution of vertebrate immune genes. For many years, the best studied example had been genes of the major histocompatibility complex (MHC) constituting part of the acquired branch of the immune response. Recent studies of innate immunity genes suggest that the mechanisms of selection acting on MHC are not a rule for immunity genes. Here, I will present results of an ongoing project aimed at characterizing the genetic polymorphism of bank vole TLR genes in the context of parasite-driven selection. Generally, the polymorphism of TLRs presenting viral motifs was much lower than in TLRs presenting bacterial motifs. The most variable was TLR2, and SNPs within this gene was significantly associated with blood parasite infections.

Dynamics of a natural P-element invasion in experimentally evolving populations of D. simulans

Robert Kofler

The P-element, one of the best understood eukaryotic transposable elements (TEs) recently invaded natural D. simulans populations. We captured a natural *D. simulans* population from Florida at an early stage of the invasion and set up a replicated experimental evolution study in hot and cold environments. This opens the unprecedented opportunity to study a natural invasion of a TE with the aid of high throughput sequencing technologies in replicated populations evolving in different environments. We show that in all replicate populations of a given environment the P-element rapidly spreads with a remarkable consistency. In the hot environment P-element copy numbers increased 16-fold up to generation 20 and attained a stable copy number of about 30 per haploid genome. No further increase could be noted during the next 40 generations of experimental evolution. By contrast, at cold conditions the speed of the invasion is much slower, the P-element multiplied 4-fold by generation 30. Interestingly, the P-element invasion only results in a modest reactivation of resident TE families in D. simulans. The levelling out of the P-element invasion at hot conditions could be due to i) a balance between transposition events and negative selection against TE insertions ii) non-autonomous truncated copies of the P-element, which have been shown to down-regulate transposition activity and iii) piRNAs. RNA-seq and small RNA-seq analysis argues that the dominant factor containing the spread of the P-element is the emergence of piRNAs complementary to the P-element.

The evolution of arthropod small RNA pathways

Samuel H. Lewis, Melanie Tanguy, Lise Frezal, Peter Sarkies, Eric Miska, Francis M. Jiggins

Small RNA pathways are evolutionarily ancient and present in the vast majority of eukaryotic species. However, the study of small RNA pathways has largely been limited to a few model organisms, establishing functional paradigms that are often assumed to apply to distantly related species. In the arthropods, *Drosophila melanogaster* has set a paradigm in which the siRNA pathway suppresses viruses and transposons in the soma and germline, and the piRNA pathway is restricted to an anti-transposon role in the germline. However, novel functions have recently been discovered in the small RNA pathways of other arthropods, suggesting that the *D. melanogaster* model may not be representative. We therefore sought to characterize the biogenesis and function of small RNAs across the arthropods. We chose a diverse set of 12 species from across the arthropod phylogeny, dissected germline and somatic tissues from males and females of each species, and sequenced the small RNA and total RNA content of each tissue. While the role of siRNAs in antiviral immunity appears to be broadly conserved across the arthropods, piRNAs have frequently evolved to defend against transposons in the soma. Our results suggest that an unprecedented diversity of small RNA pathways has evolved within the arthropods.

Comparative genomics of sexual and asexual nematode s- origins, ploidy, and molecular evolution

Amir Szitenberg, Laura Salazar, Vivian Blok, Soumi Joseph, Dominik Laetsch, Valerie Williamson, Mark L. Blaxter, **Dave Lunt**

Root Knot Nematodes are extraordinarily polyphagous, and amongst the world's most successful crop pests despite many being apomicts without any of the advantages of sexual recombination. These species are also a fascinating biological system for the study of interspecific hybridisation and speciation, ploidy evolution, genome structure, and molecular evolution processes in genomes with and without meiosis. We have sequenced Root Knot Nematodes using PacBio and Illumina and present a comparative analysis of 22 genomes to discuss the remarkable genome structure found. We compare the evolutionary processes shaping the genomes of both mitotic and meiotically reproducing species, in terms of overall structure and content, and molecular evolutionary patterns. We present molecular evolutionary analyses of the rate and pattern of meiosis-specific loci in apomicts since the loss of sexual reproduction. Finally we ask whether our views of the 'queen of problems' aren't overly simplistic given modern genomic perspectives.

A novel reproductive trait conferring high siring success is associated with postzygotic isolation in an annual plant

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The evolution of novel sexual dimorphic traits is thought to facilitate the speciation process in many lineages, particularly animals. In this study, we reveal the evolutionary origin of a novel male-like inflorescence structure that confers high siring success on otherwise hermaphroditic individuals of the annual plant *Mercurialis annua*, which is associated with reproductive isolation. The male-like inflorescence of *M. annua* is usually only found on males, and hermaphrodites typically possess female-like inflorescences with male flowers that are poorly positioned for outcross siring success. In contrast, the novel trait combines the male-like structure with female flowers in hermaphrodites and, as in males, confers high siring success. It is thus a good candidate for a trait under sexually antagonistic selection. We will present data from common garden experiments that characterise the distinctive morphology of the typical and novel hermaphrodite morphs and their relative siring abilities. Crosses reveal that the two morphs are in fact reproductively isolated from one another. The two morphs are genetically distinct and form two phylogenetic clusters, despite sympatry. Flow cytometry confirms that both lineages are hexaploid, but that they differ somewhat in genome size. Our results uncover a new species in the *M. annua* species complex. They help to explain why the sexually antagonistic inflorescence trait has not spread more widely and throw new light on the origins of the remarkable sexual-system diversity in the clade of annual *Mercurialis* species.

Genetic parallelism in multiple adaptive radiations of three-spined stickleback.

Isabel S. Magalhaes, James Whiting*, Daniele D'Agostino, Paul Hohenlohe and **Andrew D.C. MacColl**
(*joint first authors)

The extent to which evolution is repeatable is an important open question. Here we examine this in multiple adaptive radiations of three-spined stickleback, by quantifying the the extent to which patterns of association between environmental and genetic variation are repeated across radiations. We correlated the allele frequency distribution of a large sample of SNPs from 77 distinct stickleback populations from 4 independent adaptive radiations to a set of 6 environmental factors that describe local environmental variation such as water chemistry and parasite loads. Our major goals were to determine the relative contributions of different environmental variables in driving local genetic adaptation, and to determine the extent of parallelism in loci involved in adaption across radiations. We hypothesised that there is repeatability in the loci involved in the adaptation of individuals to similar environments across radiations. We detected a significant number of genes associated with one or more environmental variables and found that some genes are correlated with the same environmental variable in two or more radiations suggesting parallelism and therefore repeatability.

Using RAD-sequencing to investigate the Genetic Control of Colour-pattern in the 2-spot and 10-spot ladybirds.

Tamsin M. O. Majerus, University of Nottingham

Understanding the genetic basis of adaptation is a fundamental aim in evolutionary biology. Ladybirds are colourful and popular insects, as well as economically important biological control agents. Ladybird colour-patterns influence their survival and reproduction. Bright, contrasting colours serve as warning colours that the ladybirds may be toxic or distasteful to predators. Colour-patterns can also be important in determining ladybird mate choice.
Breeding experiments in the 2- and 10-spot ladybirds, *Adalia bipunctata* and *A*. *decempunctata*, show that the variation in their colour-patterns is controlled by one gene. It is not known if it is the same gene in both species, or how the gene creates patterns.
Crosses with different colour-patterns have been bred. DNA extracted from parents and 94 offspring per cross, has been analysed via RAD (Restriction-site Associated DNA)-sequencing. Markers have been identified which are linked to colour-pattern. This has allowed construction of a linkage map around the gene controlling colour-pattern in both species. In addition breeding data of the crosses involved has provided information about other genetic and evolutionary factors important in ladybird ecology.
The findings will provide insight into factors controlling ladybird colour-pattern, survival and reproduction. This impacts on ladybird use for biocontrol and has broad ecological significance.

The house spider genome reveals a whole genome duplication during arachnid evolution

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Gene duplication plays an important role in the evolutionary diversification of organisms. There is increasing evidence for the large-scale duplication of genes in some chelicerate lineages including two rounds of whole genome duplication (WGD) in horseshoe crabs. To investigate this further we sequenced and analysed the genome of the common house spider *Parasteatoda tepidariorum*. We found pervasive duplication of both coding and non-coding genes in this spider, including two clusters of Hox genes and duplicates in approximately 40% of microRNA families. It appears that while some of these duplicated genes have been produced by tandem duplication, analysis of the synteny of the *P. tepidariorum* genome suggests that there has been a WGD during the evolution of spiders. To investigate when this event occurred we compared the *P. tepidariorum* genome with the genomes of other chelicerates. Our findings suggest that this putative WGD occurred in the common ancestor of spiders and scorpions and is independent of the WGDs in horseshoe crabs. Furthermore, characterisation of the sequence and expression of paralogs in *P. tepidariorum* suggests many of these genes underwent neofunctionalisation and/or subfunctionalisation after duplication, and therefore may have contributed to the diversification of spiders and other arachnids.

The genetic basis of genital evolution among *Drosophila* species

Cláudia C. Mendes, Joanna F. Hagen, Kentaro M. Tanaka, Maria D. S. Nunes and Alistair P. McGregor

External male genitalia can exhibit remarkable morphological diversity to the extent that even closely related *Drosophila* species show striking variation in genital structures. Here, we focused on two species of the *D. simulans* clade, *D. mauritiana* and *D. simulans*, which show considerable differences in the morphology of two external genital structures: the claspers and the posterior lobes. Using QTL and fine-scale introgression mapping, we identified several regions on chromosome arm 3L and 3R that contribute to interspecific variation in clasper size. We also found that the regions involved in clasper size differences are mutually exclusive from those with effects on posterior lobe morphology, suggesting that different genes underlie the evolution of these genital traits. We then performed an RNAi screening in *D. melanogaster* to identify positional candidate genes that regulate male genital development. This approach allowed us to find compelling candidate genes, including *Grunge* and *male-specific lethal* 3, and we are now using CRISPR/Cas9 in *D. simulans* and *D. mauritiana* to investigate if these candidate genes have contributed to genital evolution between these species. Therefore, our study has provided new insights into the individual genes and the genetic interactions that underlie genital development and evolution.

Selection and gene flow at major colour pattern loci in mimetic Heliconius butterflies

Markus Moest, Simon H. Martin, Camilo Salazar, Steven Van Belleghem, Chris D. Jiggins

The significance of hybridization and introgression as sources of adaptive variation and their role in speciation have recently received considerable attention. However, the frequency of these processes, their importance for adaptation and speciation, and the genomic consequences of adaptive introgression are still not well-understood.
Heliconius butterflies are unpalatable, a fact they signal to their potential predators via conspicuous colour patterns. Variation among locally different wing colour patterns is explained by just four distinct genomic regions and differences in regulatory elements therein. Several species are known to hybridize and exchange genetic material, including colour pattern alleles.
Using a combination of a targeted sequence-capture of 10 Mb, comprising all four colour pattern and random genomic regions, and whole-genome re-sequencing, we have compiled a dataset for 500 individuals from several populations across the *H. melpomene/timareta/cydno* and 'silvaniform' clades. The dataset covers a multitude of different colour patterns as well as natural replicates of populations and species sharing the same colour patterns. We analyse this powerful dataset to elucidate the evolutionary history of the various colour pattern alleles, characterise the extent and role of selection at these loci, and test scenarios of adaptive introgression and hybrid speciation. I am going to present the first results from this project with a focus on selective processes acting on the genomic regions underlying colour pattern variation.

Passenger pigeon genomes reveal the cost of natural selection for a large population

Gemma G. R. Murray
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Passenger pigeons were once the most abundant bird in North America, numbering several billion individuals. In the late 19th century hunting drove a sudden decline in their population, and by 1914 they were extinct. While larger populations are predicted to experience a greater efficacy of natural selection, this may be counteracted by linkage between loci under selection. Through comparisons of patterns of molecular variation across the genomes of four passenger pigeons and two band-tailed pigeons, the closest living relative of the passenger pigeon, we reveal the joint impact of population size and recombination rate on neutral diversity and the efficacy of selection. We find that both neutral diversity and the efficacy of selection vary substantially across the passenger pigeon genome, and correlate with expected rates of recombination: high-recombination regions have higher neutral diversity, higher rates of adaptive substitution and lower rates of deleterious change than low-recombination regions. This is not found in the band-tailed pigeon genome, which has a much smaller population size. This demonstrates that the variable recombination landscape observed across bird species can influence the efficacy of natural selection, and sheds some light on the factors that may have influenced the rapid extinction of passenger pigeons.

Inbreeding depression in lifetime reproductive success of house sparrows

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Inbreeding threatens the survival of small populations of naturally outbreeding species. However, studies taking advantage of modern genomic tools to study inbreeding depression in natural populations are rare.
We used data from a long-term study of eight insular house sparrow poulations in Northern Norway to examine inbreeding depression in 3150 adult individuals over their lifetime. The study islands face relatively similar environmental conditions but they differ in population histories. This study setting allows investigation of the effects that variance in inbreeding, population size and population growth rates have on the magnitude of inbreeding depression.
The individuals were genotyped for ca. 185,000 SNPs distributed across the house sparrow genome. We found that inbreeding coefficients estimated from multi-generational pedigrees correlated well with genome wide heterozygosity and with inbreeding coefficients estimated from SNPs (F_{GRM}). As expected, the correlation was stronger for the populations with deeper pedigrees. Using the genomic F_{GRM} estimate, we found that inbred individuals had lower lifetime reproductive success, measured as the number of recruiting offspring produced during their lifetime. The strength of inbreeding depression did however vary between islands, and appeared to be related to population size and inter-individual variance in the level of inbreedig.

Pervasive adaptive plasticity limits evolutionary potential to environmental change

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Phenotypic plasticity, the ability to express multiple phenotypes from the same genome, is a key adaptation to variability in habitat quality, in particular seasonal variation. However, there is little empirical knowledge on the role of plasticity in facilitating evolutionary adaptation to new environments. Here, we use a seasonal Afrotropical butterfly to quantify the extent of adaptive phenotypic plasticity across the transcriptome, and to reveal the potential for evolutionary change in such plasticity. We uncover high amounts of seasonal plasticity in the transcriptome, reflecting a broad, genome-wide plasticity program, consistent with the broad suite of traits involved in the seasonal adaptation. Strikingly, intrapopulation genetic variation for this plastic response is highly depleted. Our study species inhabits a stable savannah environment, where temperature reliably predicts seasonal transitions. This environment has likely selected for a single environmental response, optimally tuned to capture the link between environmental cue and seasonal progression, depleting standing variation for alternative reaction norms. Under climate change, existing seasonal cues will likely break down, and dominant reaction norms become maladaptive. Depleted variation for plasticity may crucially limit evolutionary potential and population persistence when conditions change. Thus, phenotypically plastic species may in fact be surprisingly vulnerable to climate change.

Hallmarks of early sex-chromosome evolution in the dioecious plant Mercurialis annua

Kate E. Ridout, Dmitry A. Filatov, Paris Veltsos, Aline Muyle, Gabriel A.B. Marais, Olivier Emery, Pasi Rastas and **John R. Pannell**

Separate sexes have evolved independently numerous times in flowering plants. Because these transitions have often been quite recent, they provide an opportunity to study the early stages in the evolution of sex chromosomes. Here, we report evidence, from a combination of short- and long-read sequencing, gene expression and transcript-based genetic mapping, of relatively mild degeneration of a Y chromosome in the dioecious annual plant *Mercurialis annua*. Our analysis has identified hundreds of new potentially sex-linked genes additional to the unique marker published to date; many of these have transcripts that map to a single linkage group in two mapping families. Some of the sex-linked genes are likely to be in the pseudoautosomal region of the sex chromosomes, but divergence between the X- and Y-linked gametologs implies complete sex-linkage for many of them. Twelve X-linked and 192 Y-linked loci contained stop codons, with five of the X- linked genes also pseudogenized on the Y. Approximately 5% of *M. annua* genes show sex-biased gene expression. While those genes with female-biased expression did not map preferentially to the sex-linked contigs, male-biased genes were significantly underrepresented on the X. We discuss our results in terms of the remarkable diversity of sexual systems in *M. annua* and possibility of recent sex-chromosome turnover.

What happens to sex-biased gene expression following a transition to asexuality?

Darren J. Parker, Jens Bast, Kirsten Jalvingh, Tanja Schwander

Males and females feature strikingly different phenotypes, despite sharing most of their genome. The resolution of this apparent paradox is through differential gene expression, whereby genes are expressed at different levels in each sex. This resolution however is likely to be incomplete, leading to conflict between males and females over the optimal expression of genes. We test the hypothesis that gene expression in females is constrained from evolving to its optimum level due to sexually antagonistic selection on males, by examining gene expression changes in 5 obligate asexual species of stick insect, which do not produce males. Contrary to our prediction we find that asexual females decrease expression of female-biased genes, and hypothesise that this is due to the decay of sexual traits in asexual females.

Inbreeding depression by environment interactions in a free-living mammal population.

Josephine Pemberton & Camillo Berenos

Experimental studies often find that inbreeding depression is more severe in harsh environments, but the few studies of *in situ* wild populations available to date rarely find strong support for this effect. We investigated evidence for inbreeding depression by environment interactions in nine traits in the individually-monitored Soay sheep population of St Kilda, using genomic inbreeding coefficients based on 37,037 SNP loci, and population density as an axis of environmental variation. All traits showed variation with population density and all traits showed some evidence for depression due to either an individual's own inbreeding or maternal inbreeding. However, only six traits showed evidence for an interaction in the expected direction, and only two interactions were statistically significant. We discuss reasons why wild population studies may generally fail to find strong support for interactions between inbreeding depression and environmental variation compared with experimental studies.

Do novel immune alleles give hosts an advantage over local parasites?

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In host-parasite coevolution, hosts that carry novel anti-parasite alleles are predicted to be at a selective advantage. However, this prediction is difficult to test in wild populations, and has yet to be demonstrated experimentally. In the present study, we use semi-wild populations of the Trinidadian guppy (*Poecilia reticulata*), monogenean *Gyrodactylus* parasites ("gyros"), and major histocompatibility complex immune genes (MHC) to test this 'novel alleles' hypothesis. Fish in our experiment were F2 descendents of controlled crosses between guppy populations with distinct MHC profiles. These fish were subjected to experimental infections with gyros from one of their founding populations, and were MHC-genotyped at the end of the trials. We then tested fish genotype class ("novel", "local", "mixed") as a predictor of fish gyro load. In general, fish with "novel" MHC class II genotypes (i.e. only carrying alleles from a *different* stream than that of the parasites) had the lowest gyro loads. The precise picture was more complicated, with "mixed" genotypes performing differently pre- and post-peak infection, and with one controlled cross showing the reverse of the main general effect (i.e. "local" genotypes had the lowest gyro loads). Overall, our results support the 'novel alleles' hypothesis, but with several nuances and caveats.

Genetic structure within and between deep-ocean trenches in the hadal amphipod *Bathycallisoma*.

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The depth differentiation hypothesis posits that within-species, among-population genetic differentiation will be reduced in the deep ocean because of reduced environmental variation and limited barriers to gene flow. Such ideas have gained some support for abyssal species, but make little recourse to the genetic structure of species associated with the obvious geographic discontinuities in the oceans such as seamounts, vents, cold seeps and, most notably, hadal trenches.
Here we describe patterns of genetic structure within and between populations of the cosmopolitan deep-sea amphipod *Bathycallisoma schellenbergi*, taken from various depths below 6000m in the Mariana and Kermadec trenches and intervening abyssal plain. Genetic differentiation is gauged using RAD-genotyping, providing scope for the identification of outlier loci and depth related trends in allele frequency. The overall pattern that emerges is that hadal trenches promote divergence and limit dispersal, counter to the depth differentiation concept but in line with high species endemism characteristic of the hadal zone.

A universal mechanism generating clusters of differentiated loci during divergence-with-migration

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Genome-wide patterns of divergence provide a basis for understanding the mechanisms driving adaptive divergence of populations in the face of gene flow. An intriguing empirical finding is that in some cases differentiated loci are randomly scattered across the genome while in other cases they are concentrated to narrow genomic regions forming 'islands' or 'clusters of divergence'. Thus a key question to understand the genetic mechanisms of local adaptation is: what are the factors that determine the observed more or less heterogeneous genomic patterns of divergence [1-4]? It has been suggested that clustering of diverged adaptive loci could occur if mutations close to an already diverged locus are more likely to establish than mutations at a distance [2-3]. Recently, new arguments have been put forward suggesting that this effect is too weak to be important, and genomic rearrangements or other means of suppressed recombination were suggested as more likely mechanisms [4]. However, we show that it is not necessary to appeal to mechanisms that suppress recombination for clusters of differentiated loci to form during divergence-with-migration [1]. Using multi-locus simulations of divergence under a model with migration, selection and recombination, we demonstrate that there is a universal mechanism leading to concentrated patterns of divergence early on in the divergence process…

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A Flexible Inference of Complex Population Histories and Recombination from Multiple Genomes

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Analyzing whole genome sequences provides an unprecedented resolution of the historical demography of populations. In the process, most inferential methods either ignore or simplify the confounding effects of recombination and population history on the observed polymorphism. Going further, we build upon an existing analytic approach that partitions the genome into blocks of equal (and arbitrary) size and summarizes the polymorphism and linkage information as blockwise counts of SFS types (bSFS). We introduce a novel composite likelihood framework, using the bSFS, that jointly models demography and recombination and is explicitly designed to scale up to multiple whole genome sequences. The flexible nature of our method further allows for arbitrarily complex population histories using unphased and unpolarized whole genome sequences (https://github.com/champost/ABLE). We review the demographic history of the two known Orangutan species for the first time using multiple genome sequences (over 160 Mbp in length) from each population. Our results indicate that the orangutan species diverged approximately 650-950 thousand years ago. After speciation, secondary contact modelled as pulse admixture (~300,000 years ago) is shown to have a better support than continuous gene flow which corresponds to dispersal opportunity coupled with the periodic sea-level changes in South East Asia.

Telomeres reveal the (hidden) effects of inbreeding and environmental variation in a passerine bird.

Richardson, D. S., Bebbington, K., Spurgin, L. G., Fairfield, E. A., Dugdale, H. L., Komdeur, J., Burke, T.

Telomere loss is greatly affected by oxidative stress – an individuals' inability to fully buffer the free radicals produced by internal processes during life – and relative telomere length has been found to predict survival and lifespan in various species. Consequently telomere length can be used as a biomarker of an individual's condition and ageing process. As such they also provide an opportunity to measure the (potentially hidden) costs of genetic and environmental factors. For example, quantifying the cost on inbreeding in natural populations can be difficult. First, inbreeding depression may vary with ecological stress and only be detectable over long time periods. Second, parental homozygosity may indirectly affect offspring fitness, thus confounding analyses. Finally, measurement of inbreeding coefficients, survival and reproductive success may often be too crude to detect costs in wild populations. We studied relative telomere length in a population of Seychelles warblers (*Acrocephalus sechellensis*) to assess the lifelong impact of Inbreeding depression. In juveniles, individual homozygosity was negatively associated with telomere length but only in poor seasons. In adults, individual homozygosity was consistently negatively related to telomere length, suggesting the accumulation of inbreeding depression during life. Maternal homozygosity also negatively predicted offspring telomere length. Our results show that inbreeding costs are environmentally dependent at certain life stages but may accumulate throughout life. Further analysis is now assessing the full extent and impact of environmental variation on Telomere length and survival in this population.

Estimating barriers to gene flow from distorted isolation by distance patterns

Harald Ringbauer, Alexander Kolesnikov, Nick Barton

In geographically extended populations and in case of limited migration, nearby pairs of individuals are more similar to each other than geographically well separated pairs - a classical isolation by distance pattern. Here we study how a barrier to gene flow distorts these patterns. We use the resulting theoretical insights to detect and subsequently estimate the strength of a barrier to gene flow. To this end, we fit the underlying covariance structure of genotypes with latent Gaussian models. This gives rise to a novel inference method that can detect barriers to gene flow in continuous populations from individual genotype and position data. We show that this method works well on simulated data, and apply it to an *Antirrhinum majus*

Effects of interference between selected loci in partially selfing populations

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Mating systems have important population consequences in terms of mean fitness and inbreeding depression, due to increased homozygosity caused by inbreeding. However, inbreeding also reduces the effect of recombination between loci, causing stronger interference effects among those loci. Using different theoretical models (representing directional selection against deleterious alleles without epistasis, or stabilizing selection acting on quantitative phenotypic traits), I will show that different forms of between-locus interactions affect the mutation load and inbreeding depression in partially selfing populations: correlations in homozygosity among loci (identity disequilibrium), and linkage disequilibrium caused by epistatic interactions. In general, interactions between loci tend to reduce purging of deleterious alleles caused by partial selfing. If time allows, I will also present some results on the effects of selfing on the evolution of recombination rates and mutation rates.

Evolution of Assortative Mating in a Polygenic Trait Model of Speciation with Gene Flow

Himani Sachdeva, Nick Barton

Assortative mating is an important driver of speciation in populations with gene flow and is predicted to evolve under certain conditions in few-locus models. However, the evolution of assortment is less understood for mating based on highly variable quantitative traits. We explore this scenario for a two-deme model with migration, by considering a single polygenic trait subject to divergent viability selection across demes, as well as assortative mating and sexual selection within demes, and investigate how trait divergence is shaped by various evolutionary forces. We also study the evolution of assortment via invasion of modifiers of mate discrimination and show that the evolutionarily stable assortment strength has an intermediate value under a range of migration-selection parameters, due to subtle effects such as selection for moderate hybridization between diverged populations, or for optimal variance. We further clarify how strong linkage disequilibrium between loci and the high genetic variation that characterizes polygenic traits impact assortment evolution and divergence.

Gene transfers onto the Drosophila Y chromosome are not rare, but their rates differ among species

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The Y chromosome is a unique genomic environment defined by a lack of recombination and male-limited inheritance. With all 13 known Y-linked genes in *Drosophila melanogaster* arising from autosomal gene duplications over the past 63 million years, these events are assumed to be rare (<1 novel gene/million years). Using a novel NGS-powered genomic scan we show that gene transfers to the Y-chromosome are much more common than previously thought: 45 unique Y-linked transfers in three *Drosophila* species were identified. Accounting for retrotranspositions, which were only detected in the *D. simulans* clade, we detected significant differences in the rate of gene transfers among species. With least 10% of the genes transferred to the Y-chromosome being functional (ω ~0.1), we propose that the high transfer rates in the *D. melanogaster* clade are a recent phenomenon.

Genetic diversity in *Copaifera langsdorffii* Desf: Results from RADseq data for a tropical tree species at different biomes.

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Copaifera langsdorffii is a Neotropical tree with wide distribution in the Brazilian Atlantic rain forest and savanna. Genetic analyses can identify the scale at which tree species are impacted by human activities, and provide useful demographic information for management. In recent years, it has become feasible to obtain whole genomic variation for non-model organisms since next generation sequencing (NGS) approaches have been developed and refined. Here, we evaluated the feasibility of RADseq in 95 individuals distributed in fragmented forest of the São Paulo Midwest to simultaneously identify and genotype single nucleotide polymorphisms (SNPs). We identified and genotype 2797 high-confidence SNPs across six sites distributed along different vegetation structure demonstrating the value of RADseq for generating polymorphic loci for population genetic analyses in these trees. Genetic diversity measures revealed slight differences for all populations, however the population on Botanic Garden showed moderate high values. Ho values were lower than He in all populations, indicating an excess of homozygotes. Differentiation of F_{ST} was low (0.023), but significant (0.007 - 0.044, c.i 95%) among populations. Was observed a clear correlation between geographic distance versus genetic distance. The analysis of spatial genetic structure (SGS) in populations of *C. langsdorffii* suggested isolation by distance…

Barriers to arid zone gene flow create genetic divergence in parapatric subspecies

Amy L. Slender, Marina Louter, Steven A. Myers, Tessa Bradford, Michael G. Gardner, Sonia Kleindorfer

Understanding the evolutionary consequence of contemporary sand dune barriers in an increasingly arid Australia is important to predict gene flow and migration. Two Australian parapatric songbird subspecies of the Thick-billed Grasswren (*Amytornis modestus*) have different arid-zone related ecotypes and mitochondrial haplotypes. At the parapatric border between the subspecies there is a dune field where grasswrens are known to be absent. The morphotype of one of the subspecies extends past the dunes so it is not clear whether divergence between the subspecies is related to this potential geographic barrier. Using 1975 SNPs, we identified two genetic clusters separated by the dunes. The presence of one admixed individual and mitochondrial introgression to the east of the dunes suggest the dunes are a porous (low gene flow) barrier and the subspecies are maintaining incomplete geographical separation. We found six loci that are potentially under diversifying selection, indicating a possible role for selection in the divergence of subspecies. The selection pressure driving this divergence appears to be independent of traits used to define ecotypes, as the ecotypes did not align with genetic populations. Our study shows that dunes may facilitate divergence within species highlighting the importance of geographic isolation in the divergence of subspecies.

The footprint of natural selection on genetics, morphology and behavior in a wild bird

Lewis G. Spurgin, Mirte Bosse, Veronika N. Laine, Ella F. Cole, Josh A. Firth, Phillip Gienapp, Andrew G. Gosler, Keith McMahon, Jocelyn Poissant, Irene Verhagen, Martien A. M. Groenen, Kees van Oers, Ben C. Sheldon, Marcel E. Visser, Jon Slate

Unbiased population genomics analyses are commonly used to identify genes under selection, but few studies using these approaches have been able to bridge the gap from signatures of selection to phenotypic trait variation, covariance with fitness, and the agents driving selection. Using extensive genomic and ecological data from long-term study populations of the great tit (Parus major) in the UK and the Netherlands, we unify genotypic, trait, and fitness variation with ecological data. We found that genes under differential selection between populations were associated with craniofacial features in humans, or with bill morphology in Darwin's finches, suggesting bill morphology as a candidate trait under selection. Genetic analyses suggest ongoing selection for longer bills in the UK population; a finding supported by bill length measurements from museum specimens and a 30-year time series dataset. Finally, genetic variation at a bill-length locus was associated with usage of bird feeders in the UK, suggesting that the evolution of longer bills may have been driven by the increase in supplementary bird feeding across the UK over the last century. Together, our results provide an integrated analysis of adaptive evolution, from genomes to phenotypes and behaviour.

Large-scale structural rearrangements in the fire ant genome.

Eckart Stolle, Rodrigo Pracana, Yannick Wurm

The fire ant *Solenopsis invicta* is an important global pest and an important model for social evolution. Its genome was assembled from short read sequences in 2010, and thus remains highly fragmented. This fragmentation has handicapped much molecular-genetic research.
We performed deep sequencing of a haploid fire ant male using the BioNano Genomics Irys nanochannel-based optical mapper to obtain nicking-enzyme recognition-site patterns for genomic fragments from 150,000 to 1,900,000 nucleotides long. Combining the resulting optical map with genetic maps and the old genome data we assembled a high-contiguity "optical" reference genome.
By comparing these data with similar data we generated from other males, we detected large structural variation among fire ants. We further characterise these structural variants using population scale genomics data, revealing genomic regions that may be involved in adaptation.

Hybridisation and gene flow in the Lake Malawi cichlid adaptive radiation – Locating introgression and non-tree-like ancestry on a large phylogeny

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With its more than 600 evolutionarily young and ecologically diverse species, the Lake Malawi cichlid adaptive radiation provides an outstanding system to study fundamental questions about the evolution of biodiversity and species formation. Intriguingly, all species are in principle capable to hybridise and produce fertile offspring. Using whole genome sequencing data of 130 individuals from 71 species, we test for gene flow and genomic introgression across Lake Malawi cichlid species. F4-statistic (a form of ABBA-BABA test) reveals massive signatures of genetic exchange both within the radiation and with riverine outgroups. We present a method to disentangle the correlated signals of multiple F4-tests to obtain independent estimates of introgression for both internal and external branches of a phylogeny. More generally, these "tree distortion scores" can be seen as branch-specific measures of how well the genetic ancestry is described by a species tree. In Malawi cichlids, genetic exchange is commonly between genetically distant species with similar ecology, consistent with adaptive introgression. To test this, we map the genomic distribution of introgressed segments and correlate the (mostly short) segments with gene function and signatures of selection. Finally, we report a few large regions (10s of megabases) of introgression between major clades of the radiation, some still segregating within species today.

Speciation genomics in cryptic European wood-white butterflies.

Venkat Talla

Wood-white (*Leptidea*) butterflies are similar in external appearance but can be separated based on genital morphology, behaviour, host-plant preference and karyotype structure. In addition, populations within species sometimes show significant variation in chromosome numbers (e.g. 2n = 56–106 in *L. sinapis*). We obtained a high quality genome assembly by sequencing a Swedish *L. sinapis* larva obtained from four generations of full-sib inbreeding and used population samples from six different populations representing three closely related species of the cryptic complex to study patterns of genomic differentiation between these species and model their demographic histories. We found that genomic differentiation was highly heterogeneous in all comparisons with some evidence of parallelism and the genomic data indicated deeper divergence times than previous estimates based on mitochondrial DNA. There was no indication of post-divergence gene-flow between any species pair. We also detected significant intraspecific differentiation between specific populations of both *L. sinapis* and *L. juvernica*. In *L. sinapis* the highest level of differentiation was observed between populations with distinct karyotypes. In addition, *L. sinapis* populations seems to have undergone a considerable expansion recently while the two other species showed little to moderate variation in historical population size. The results provide information on the speciation history in general and how differentiation processes affect genome evolution in diverging lineages in particular.

Quantifying selection with Pool-seq time series data

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Because allele frequency trajectories capture the essential information about the underlying evolutionary forces, the analysis of such time series is becoming increasingly popular. Of particular interest are evolve and resequence studies, since they do not only provide time series data, but also allow for replication. Nevertheless, the available methods to estimate selection coefficients (*s*) from time series data are designed for the analysis of single replicates only.
To fill this gap we developed a statistical framework to estimate both *s* and the dominance parameter (*h*) by combining the information of multiple replicates. We show that the significance of $\hat{s}$ can be assessed and heterogeneity of *s* (or optionally *h*) across replicates can be tested.
Applying our novel approach to a comprehensive set of computer simulations suggests that our method is accurate and unbiased. Furthermore, we show that replicated time series provide higher accuracy than a single replicate, when we assume the same overall total sequencing coverage. We propose that many time series experiments will benefit from our new approach.

Influence of male-male competition on the evolution of sexual dimorphism in a wind-pollinated herb

Jeanne Tonnabel, Patrice David, Etienne Klein, John R. Pannell

Sexual selection in animals is generally recognized as a force promoting the divergence of phenotypic characters between males and females. In particular, female choice and male-male competition have been shown to trigger extreme cases of sexual dimorphism in many animal species. But to what extant could such mechanisms also explain the evolution of sexual size dimorphism in dioecious plants? In herbs, males are often smaller than females, but in wind- pollinated species, plant height is expected to enhance male competitive abilities by enhancing the dispersal of pollen from greater heights. Here, we present first results from an experimental evolution study with the wind-pollinated species *Mercurialis annua* that aims to determine whether, and how, male-male competition affects selection on plant architecture, and thus how it contributes to the evolution of sexual dimorphism. We present data on female fecundity and male siring success, based on marker-assisted paternity analysis, to estimate sex-specific selection gradients in experimental arrays at two contrasting plant densities. Our study has implications for the way sexual selection influences sexual size dimorphism in plants, where size has implications for both siring success and access to resources.

Surfing on the seascape: when frequent environmental change prevents extinction

Barbora Trubenová, Martin Krejca, Per Kristian Lehre, Timo Kötzing

Environmental fluctuations are expected to increase in frequency and severity within the near future. If these environmental changes are too widespread and severe for individuals to be able to migrate to more suitable areas or acclimatise physiologically, they can prove fatal. Whether these species succeed in adapting and escape extinction is a major evolutionary question receiving growing interest from a range of biological disciplines. Using a multilocus evolutionary model, we investigate the effects of the frequency of environmental fluctuations on the adaptability of living organisms. We use mathematical methods from theoretical evolutionary computation to investigate the interplay between fluctuation frequency, shape of the fitness landscape and the number of loci contributing to adaptation. We show that populations may survive either by adapting rapidly to the current environment, or by becoming generalists that survive, though not thrive, in a wide range of conditions. The evolutionary strategy that evolves depends on the frequency and severity of the environmental change, as well as the number of loci under selection. Furthermore, we prove that under some circumstances frequent environmental change, if not too severe, can inhibit over-specialisation to given conditions and thus prevent extinction.

Do hybrid sterility loci show distinctive signatures in the genomic landscape of differentiation between house mouse subspecies?

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A central goal in evolutionary biology is to understand the genetic basis of speciation. One strategy to identify loci involved in reproductive isolation is to compare the genomes of closely related species and identify highly differentiated candidate regions. Recently, this population-genomics approach has become widely adopted in diverse taxa. An alternative strategy is genetic mapping of reproductive isolation traits. Because mapping studies in natural populations are rare, it is not clear whether these two approaches will converge on the same candidates. We previously identified loci associated with male sterility in a genome-wide association study of mice from a hybrid zone. Here, we estimated sequence diversity and divergence using genomes of eight individuals each from three *Mus musculus domesticus* populations and three *M. m. musculus* populations. We compared genetic differentiation between subspecies in sterility loci to genome-wide averages to determine if they show distinctive signatures in the genomic landscape. We find that differentiation is higher, on average, in sterility loci, but these regions would not stand out in outlier scans. Hence, we cannot propose signatures that might be used to identify new candidate regions for reproductive isolation. However, measures of differentiation will be useful for prioritizing candidate genes within sterility loci.

Genetic differentiation of primitive cattle breeds illustrate aurochs (*Bos primigenius)* admixture

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The domestication of taurine cattle initiated ~10 000 years ago in the Near East from a wild aurochs (*Bos primigenius*) population followed by their dispersal through migration of agriculturalists to Europe. Although gene flow from wild aurochs still present at the time of this early dispersion is still debated, some of the extant primitive cattle populations are believed to possess the aurochs-like primitive features. In this study, we use genome-wide single nucleotide polymorphisms to assess relationship, admixture patterns and demographic history of an ancient aurochs sample and European cattle populations. Population admixture analysis indicates a zebu gene flow in the Balkan and Italian Podolic cattle populations. Our analysis supports the previous report of gene flow between British and Irish primitive cattle populations and local aurochs. In addition, we show evidence of aurochs gene flow in the Iberian cattle populations indicating wide geographical distribution of the aurochs. Runs of homozygosity reveal that demographic processes like genetic isolation and breed formation have contributed to genomic variations of European cattle populations. We conclude that in addition to factors such as ancient human migrations, isolation by distance and cross-breeding, gene flow between domestic and wild-cattle populations also has shaped genomic composition of European cattle populations.

Selecting for spots: phenotypic plasticity in the Harlequin ladybird

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In recent years the Harlequin ladybird (*Harmonia axyridis*) has seen much attention as a result of its invasive nature outside its native range in Asia. One of its colour morphs, *f. succinea*, shows a striking case of phenotypic plasticity, dramatically increasing dorsal melanin pigmentation when reared in a cold environment. The direction of this response is in line with the thermal melanism hypothesis and it is thought that this plastic ability may have aided this species in becoming such a successful invasive organism. *Harmonia axyridis f. succinea* provides an excellent and very relevant opportunity in which to investigate the molecular mechanisms underlying phenotypic plasticity. Understanding the characteristics of invasive species is crucial for the management and prevention of their ecological impact. A year-long selection experiment in which lines underwent selection based on their degree of plasticity has just been completed. The design features six lines (High, Low and Unselected), full-sibling groups reared at two extreme temperatures, and automated image analysis. Results reveal significant differences between control lines and selection lines. This work demonstrates the genetic variance for this trait, and the selection lines will be a valuable resource for future investigations of the molecular mechanisms involved.

Comparison of gene expression between XX and XY males in the common frog

Paris Veltsos, Melissa Toups, Nicolas Rodrigues, Alan Brelsford, Nicolas Perrin

The common frog, Rana temporaria, possesses homomorphic sex chromosomes. We have recently discovered striking variation in sex chromosome di erentiation in the species. Male haplotypes range from indistinguishable from female haplotypes to male-specific haplotypes spanning the majority of the sex chromosome length. No phenotypic distinction between these XX and XY males has been detected in morphology, hormone production, or reproductive success even in a population where they naturally mix. We investigate the genetic consequences of having a Y chromosome by comparing the transcriptome of both types of male to that of females. The results are discussed in the context of masculinisation of the male transcriptome by a Y chromosome, dosage compensation and genetic di erentiation across the Y chromosome.

Can pollen quantity and quality explain differential siring success between self-compatible and –incompatible individuals of a perennial plant?

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Evolution toward selfing is one of the most frequent transitions to have occurred in flowering plants, and its causes, mechanisms and consequences continue to pose puzzles. For instance, it is not well understood under what conditions newly self-compatible (SC) plants can invade self-incompatible (SI) populations; such invasion and spread might be hampered by the fact that the production of pollen is expected to be reduced in SC plants, which can reduce outcrossing opportunities. Moreover, theory suggests that SC plants might enjoy lower siring success that SC plants because of differences in patterns of parent-offspring conflict. In contrast to these expectations, SC individuals of the perennial plant *Linaria cavanillesii* enjoy substantially better outcross siring success than their SI counterparts. Here, we tested the hypothesis that differences in pollen competition after pollination could help to explain the higher siring success of SC plants. Paternity analysis using microsatellites on seeds produced following experimental crosses among SC and SI individuals of *L. cavanillesii* indeed revealed a higher siring success for pollen from SC individuals when competing with that from SI individuals on equal terms. We consider why SC has so far failed to spread in natural populations of this perennial plant.

Genetic recapture: a general framework for understanding wild population demography

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Capture-mark-recapture (CMR) methods have been widely used to study wild populations. Less known and less used are two genetic recapture (GR) methods, which can be employed to estimate population demography without the capturing and marking procedures. The first method resembles the camera trap method and uses randomly collected noninvasive DNA samples (e.g. faeces) to analyse an individual's "recapture" rate, and thus population density. The 2nd method uses DNA samples to recapture a family rather than an individual, and thus to estimate genetic population parameters such as effective population size and migration rate. The two genetic methods have several advantages over CMR. They do not require capturing and marking animals. They use the natural, universal, high resolution markers of DNA and the same marker analysis methods (PCR, sequencing). They have a higher recapture rate because the recapture unit, family, can contain many individuals and thus are easier targets than a single individual. The genetic methods greatly widen the application scopes (species) of CMR, and can be used to study historical population demography from ancient DNA samples. I briefly show how to infer effective population size and migration rate from GR methods.

Demographic inference from the site frequency spectrum: a comparison of RADseq *vs* whole genome sequencing in pied flycatchers (*Ficedula hypoleuca*)

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The site frequency spectrum (SFS) plays a central role in population genetics: many commonly used statistics, such as theta and F_{ST}, can be directly calculated from the SFS. Because its shape reflects underlying population genetic processes, it can be used for population genetic inference of demography, natural selection and population structure.
Compared to whole genome re-sequencing, restriction-site associated DNA sequencing (RADseq) may introduce additional bias and error; however, it is unclear to what extent the effect of such RAD-specific errors on the distribution of allele frequencies will lead to different conclusions compared to those based on the analysis of whole genome re-sequencing data.
We generated RADseq data for pied flycatcher individuals that were part of a previously published re-sequencing study. Comparing the results of SFS-based population genetics analyses between the two sequencing strategies, we find that diversity as estimated from RADseq data is consistently lower than when estimated from re-sequencing data; however, SFS-based demographic inference from RADseq data yielded similar results to those from re-sequencing data, both with regards to model choice and parameter estimation.

Evolutionary persistence and physiological costs of self-splicing introns

Marina Rudan, Jelena Repar, Anita Krisko, **Tobias Warnecke**

Self-splicing introns are mobile genetic elements that populate several highly conserved protein-coding genes in fungal and plant mitochondria. They have persisted in yeast populations over long periods of evolutionary time despite making no adaptive contribution to their host (as far as we know). In part, their persistence in the host genome has been attributed to their capacity to continuosly re-invade the population. In addition, it has been suggested that they have a minimal impact on host fitness given their ability to splice themselves out of their host mRNAs and thereby reconstitute functional reading frames. In this talk, I will challenge this notion by exposing both mutational and physiological costs of self-splicing introns in *Saccharomyces cerevisiae.* In particular, I will show that, unexpectedly, removing self-splicing introns is deleterious to the host. Based on a series of experiments to unravel the nature of costly intron removal, I suggest that the persistence of self-splicing introns can in part be explained by compensatory evolution in the host: Initial invasion is costly and mutations arise in the host genome to ameliorate the deleterious effect of self-splicing introns, but the genomic changes turn bad when the initial offenders are removed, thus rendering intron loss deleterious.

Going the long (molecule) way: Detecting structural features influencing recombination rate via SMRT-seq and optical mapping in the crow genome

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Accurate and contiguous genome assembly is key to a understanding of the processes shaping genomic diversity and evolution. Yet, it is frequently constrained by constitutive heterochromatin, usually characterized by highly repetitive DNA. As a key feature of genome architecture it is associated with centromeric and telomeric regions and influences meiotic recombination. Here we assess the impact of large tandem repeat arrays on the recombination rate landscape in the Eurasian crow. We assembled two genome references using single-molecule real-time (SMRT) sequencing and single-molecule optical maps. A three-way comparison including the published short-read assembly constructed for the same individual allowed assessing assembly properties and pinpointing mis-assemblies. We characterized 36 previously unidentified large repetitive regions at sequence assembly breakpoints, the majority of which contained complex arrays of a 14-kb satellite repeat or its 1.2-kb subunit. We estimated the population-scaled recombination rate (ρ) and found it to be significantly reduced in these regions. Our findings are consistent with an effect of low recombination in regions adjacent to centromeric or sub-telomeric heterochromatin, and add to our understanding of the processes generating genetic diversity. By combining three independent technologies, our results highlight the importance of adding a layer of information on genome structure inaccessible to each approach independently.

Towards identifying the genomic basis of pre-zygotic reproductive isolation in a sympatric species complex of intertidal isopods (*Jaera albifrons*)

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A fundamental question in speciation research is how genomic divergence establishes initial barriers to gene flow among sympatric populations. Many examples of genomic differentiation or structural chromosomal polymorphism among nascent species have been described, but there are few examples where such polymorphism has been explicitly linked to phenotypic characters that confer reproductive isolation. The *Jaera albifrons* species complex comprises four sympatric species of intertidal isopods in the UK and is a prime example of prezygotic reproductive isolation. Species are diagnosed morphologically by extent and location of setation of legs in males, which provide courtship stimuli to conspecific females during mating and thus present an elegant mechanism to prevent heterospecific gene flow. Here we characterise the genomic landscape of reproductive isolation in this species complex by sequencing and assembling the genome of *J. ischiosetosa* and ascertaining sequence divergence among all species using RADseq. These results pave the way for examining how genomic islands of elevated divergence co-locate with genes differentially expressed during juvenile development, QTL for leg phenotype, and loci displaying segregation distortion.

Studying the genomic basis of ecotype divergence in hybrid zones of marine snails

Anja M. Westram, Tomas Larsson, Pragya Chaube, Kerstin Johannesson, Roger Butlin

The marine snail *Littorina saxatilis* contains two morphologically and behaviourally distinct ecotypes, which are adapted to wave exposure and crab predation, respectively. These ecotypes have evolved repeatedly in multiple geographical locations across Europe and show partial reproductive isolation. This system is therefore ideally suited to study adaptive divergence and early stages of speciation. In previous work, we have used genome scans to identify outlier loci between ecotypes, potentially reflecting genomic regions under divergent selection. While genome scans represent a first step for identifying candidate loci, the extent of divergent selection acting on these loci is often unclear, and functional roles and distribution in the genome are unknown. To gain more insight into the genomic basis of ecotype divergence, we have sequenced 40,000 markers in hundreds of *L. saxatilis* sampled across a hybrid zone on the Swedish west coast, where ecotypes interbreed. We have identified loci with steep allele frequency clines across the habitat transition, and show that these loci often correspond to outliers from earlier work, supporting the role of divergent selection. I will also present results about the genomic distribution of potentially selected loci, obtained from combining hybrid zone data with a genome assembly and a genetic map.

Molecular basis of phenotypic divergence between two orchid species (*Dactylorhiza*) with distinct ecological optima)

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The orchid family is the largest in the angiosperms, but little is known about the molecular basis of the significant varia- tion they exhibit. We investigate here the transcriptomic divergence between two European terrestrial orchids, *Dactylorhiza incarnata* and *D. fuchsii*, and integrate these results in the context of their distinct ecologies that we also document. Clear signals of lineage-specific adaptive evolution of protein-coding sequences are identified, notably targeting elements of biotic defence, including both physical and chemical adaptations in the context of divergent pools of pathogens and herbivores. In turn, a substantial regulatory divergence between the two species grown in a common garden appears mainly driven by adaptation/acclimation to abiotic conditions. Finally, *Dactylorhiza incarnata* appears to suffer from insufficient small RNA control over the activity of RNA-dependent DNA polymerase, resulting in increased activity of class I transposable elements and, over time, in larger genome size than that of *D. fuchsii*. Altogether, biological response to selection, accumulated during the history of these orchids, appears governed by their microenvironmental context, in which biotic and abiotic pressures act synergistically to shape transcriptome structure, expression and regulation.

Role of sexual selection in guppy sex chromosome formation

Alison E. Wright, Iulia Darolti, Natasha I. Bloch, Vicencio Oostra, Ben Sandkam, Severine D. Buechel, Niclas Kolm, Felix Breden, Beatriz Vicoso & Judith E. Mank

Sex chromosomes evolve once recombination is halted between a homologous pair of chromosomes. This process has occurred independently throughout many eukaryotic clades, however we still lack an empirical understanding of the forces catalyzing sex chromosome formation. The dominant model of sex chromosome evolution predicts that recombination will be suppressed between emerging sex chromosomes in order to resolve sexual conflict. Here, we test this model using whole genome and transcriptome resequencing data in the guppy, a classic model for sexual selection where it has long been known that many male color traits are Y- linked. We show that the sex chromosomes in guppies are remarkably young with no perceptible degradation of Y chromosome gene content or activity. Using replicate wild populations with differing levels of sexual conflict over color, we show that sexual selection has lead to convergent expansion of the non-recombining region and increased Y chromosome divergence. These results provide the first empirical support for longstanding models of sex chromosome formation, and suggest an important role for sexual selection and sexual conflict in genome evolution.

Estimating variation in both the effective population size and the mutation rate across the genome in a non-equilibrium population

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It is well known that both the effective population size (N_e) and the mutation rate (u) vary across the genome. Understanding this variability is fundamental for making sense of the process of evolution. On the other hand, failing to account for this heterogeneity could bias estimates of past demographic events. In this study, I show that it is possible to simultaneously estimate variation in both N_e and u using polymorphism data alone, provided that the population has experienced recent changes in population size. This is achieved by resolving a numerical issue that limits the sample size to which a general result for predicting the site-frequency spectrum in non-equilibrium populations can be applied. Simulations suggest that the method is accurate, flexible, and scalable to whole-genome data. This method should be useful for researchers interested in estimating past demographic changes, quantifying intra- chromosomal variation in N_e and u, and making comparisons between autosomes and the X chromosome (e.g., detecting sex-biased demographic processes and male- driven evolution).