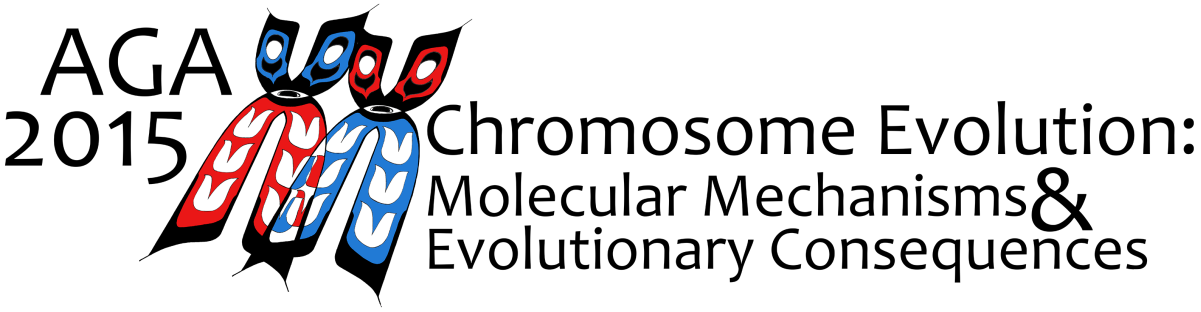
SPEAKER ABSTRACTS



Wilhelmine Key Lecture

The Evolution of Genome Structure by Natural and Sexual Selection

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Evolution of the karyotype was at the heart of evolutionary genetics for 50 years, but fell out of the spotlight with the rise of molecular biology. New sequencing technologies are now revealing that genome structure is even more dynamic than previously imagined. What evolutionary forces drive these changes in chromosome number, structure, and function? This talk will survey what we've discovered about the evolution of autosomal rearrangements and sex chromosomes using theory, data from the field, and comparative analysis. I'll end by discussing our most recent results that show sex chromosomes have surprising effects on population demography.

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Chromosomal hybrid zones and speciation in mice and shrews

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The western house mouse (*Mus musculus domesticus*) and common shrew (*Sorex araneus*) show dramatic chromosomal variation, with around 100 chromosomal races in each species distinguished by different sets of metacentric chromosomes that have arisen by Robertsonian fusion and whole-arm reciprocal translocation. Numerous hybrid zones have been described in the two species at the contact of different races. In some cases the hybrids are characterised by extremely long chain or ring configurations at meiosis I, associated with high levels of gametogenic abnormality and reduced fertility. In accordance with hybrid zone theory, hybrid zone width varies with the complexity of the meiotic chromosomal configuration; where the hybrids are expected to have low fertility the hybrid zone is narrower. Other features of the hybrid zones fit the classic tension zone model, e.g. localization of zones in regions of low population density. There is evidence of behavioral isolation between chromosomal races in some zones, which may be a response to selection against unfit hybrids (i.e. reinforcement). However, selection appears to have favored novel homozygous forms in other hybrid zones, thereby de-escalating rather than escalating the speciation process. While the role of hybrid unfitness in determining hybrid zone structure in terms of chromosomal clines is relatively straightforward, the relationship between hybrid unfitness, recombination suppression and gene flow is more difficult to untangle. I will describe recent data recording recombination using MLH1 foci, and examining gene flow using mapped microsatellites and SNPs. All-in-all the western house mouse with its access to all the cell biological and genomic tools of the laboratory mouse, and the common shrew with its extraordinary diversity of chromomosomal hybrids from numerous hybrid zones, are wonderful models to examine the role of chromosomal rearrangements in speciation.

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Drivers of sex chromosome-autosome fusions and their roles in speciation

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Sex chromosomes often fuse with autosomes. What drives sex chromosome-autosome fusions? What are the roles of sex chromosome fusions in speciation? To adress these questions, we first compiled cases of sex chromosome-autosome fusions in vertebrates. We found that Y chromosomes fuse with autosomes more often than other sex chromosomes in fishes and reptiles, whereas X-autosome fusions also occur at similar frequencies in mammals. Theoretical analysis showed that the prevalence of Y-autosome fusions can be explained by fixation of deleterious fusions by drift under the conditions of male-biased origination rates or male-biased variance in reproductive success. The high prevalence of X-autosome fusions in mammals may be explanined by female meiotic drive for fusions, because we found a significant correlation between autosomal karyotype and the prevalence of X-autosome fusions. Next, in order to investigate the roles of sex chromosome-autosome fusions in speciation, we used two Japanese stickleback species that were brought into secondary contact after a long alloparic period. Our mapping studies, genome sequence analysis and gene expression analysis indicated that the neo-sex chromosome created by Y-autosome fusions tends to accumulate genes important for reproducive isolation and sexually dimorphic genes. Our simulations further indicate that gene flow after secondary contact between two populations under contrasting sexually antagonistic selection can drive the evolution of sex chromosome-autosome fusions. Taken together, our data indicate that the Y-autosome fusion in the Japan Sea sticklebacks may be driven by gene flow after secondary contact and act as sites for the accumulation of nucleotide differeneces important for speciation.

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Neo-sex chromosome evolution and speciation in beetles

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Sex chromosomes play disproportionately large roles in adaptation and speciation but the links between these two processes remain unclear. The tempo of genome evolution is thought to be particularly rapid during transitions to new (neo) sex determining chromosomes yet the impact of neo-sex chromosome evolution on incipient speciation has received little attention. I will discuss our research combining extensive crossing experiments with population and comparative genomic data in the neo-XY mountain pine beetle system. With these data we show a rapid progression of intrinsic reproductive isolation between closely related populations that is geographically coupled with extensive genetic and functional differentiation of the neo sex chromosomes. Our results provide insights into the early stages of sex chromosome evolution and its potential to drive incipient speciation.

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The dark side of the genome: A high-resolution study of human centromeric regions

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Human centromeric regions are predominantly defined by long arrays of near-identical tandem repeats, or satellite DNA. This sequence homogeneity prevents standard assembly, and as a result centromeres and sites typically associated with blocks of constitutive heterochromatin are omitted from even the most well studied reference genomes. To address this challenge, I present a graph-based approach to characterize both satellite and non-satellite sequence organization within each centromere-assigned gap in the human reference genome. Utilizing these sequence maps I am able to apply a k-mer based mapping approach to conduct high-resolution studies within specific chormosome-assigned centromeric arrays.

Intersecting available functional datasets from an extensively studied B-lymphoblastoid cell line (GM12878), I provide an initial epigenomic map human centromere arrays, with a focus on the satellite array (DXZ1) on chromosome X. Despite the accumulation of repressive epigenetic marks, centromeric satellite DNAs are transcribed as non-coding RNAs, with short RNA localizing primarily to the chromatin compartment of the nucleus. This work also identifies sites of transcription factor enrichment and histone modification across repetitive DNAs. These studies are expected to establish a new niche in the genome sciences that integrates the use of both computational and experimental approaches to better understand these specialized regions of the genome.

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Identification and validation of gray mouse lemur (*M. murinus*) centromeres using genome assembly and cytological approaches

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Centromeres are crucial chromosomal loci that are essential for chromosome segregation and genome stability. Although centromeres are largely defined by sequence-independent (epigenetic) mechanisms they are often located in regions containing repetitive DNA. Alpha satellite DNA is the most predominant repetitive DNA in primates. In humans the 171bp basic repeat unit (monomer) is tandemly arranged into higher order repeat structures (HORs). Old and new world monkeys are generally thought to lack centromeric repeats with HOR structure however recent reports have suggested that hierarchically ordered repeats are a general feature of simian primates. At average adult weight of ~60 grams gray mouse lemurs (*Microcebus murinus*) are among the smallest primates in the world and are remarkably long-lived for their size. They share many similarities to humans that afford comparisons of molecular mechanisms underlying metabolism aging and chromosome biology. *M. murinus* has 66 chromosomes all of which are telocentric (similar to mouse) or acrocentric (like human chromosomes 13 14 15 21 and 22) except for the X chromosome that is metacentric (like the human X). We sought to identify the centromeres in this organism. Using the combination of BioNano optical mapping and PacBio long read sequencing technology we searched for sequences that mapped at or near gaps in the genome assembly. Tandem Repeat Finder was used to screen raw PacBio sequence data for all *M. murinus* repeats and a 53-bp sequence motif (AS53) was identified as being overly abundant throughout the genome and repeating for stretches of up to 40 kilobases. A biotinylated oligo representing the 53-bp sequence was hybridized to *M. murinus* metaphase chromosomes using FISH. Fluorescent AS53 probe signal was detected at every centromere region except for M. murinus chromosome X (MIMX). The abundance of AS53 varied widely among chromosomes and did not correlate with chromosome size. Using immunofluorescence-FISH with antibodies to human CENP-A the centromeric histone variant required for centromere assembly and maintenance we observed co-localization of centromere proteins and AS53. These results indicate that *M. murinus* centromeres except for MIMX share the same genomic sequence that serves as the site of CENP-A assembly and kinetochore formation. The absence of AS53 at the centromere of MIMX suggests a unique genomic history for *M. murinus* sex chromosomes that remains to be more deeply explored. Our study underscores the power of using genomic sequence information particularly optical genomic maps to identify a key genomic locus that is important for chromosome function and inheritance.

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Overcoming AbCENs: Recurrent loss of an essential gene in holocentric insects

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Faithful chromosome segregation in all eukaryotes relies on centromeres the chromosomal sites that recruit kinetochore proteins and mediate spindle attachment during cell division. Fundamental to centromere function in most eukaryotes is a histone H3 variant CenH3 that initiates kinetochore assembly on centromeric DNA. CenH3 is conserved throughout most eukaryotes; its deletion is lethal in all organisms tested. These findings established the paradigm that CenH3 is the defining component of all centromeres. Our recent findings undermined this paradigm of CenH3 essentiality. We showed that CenH3 was lost independently in four lineages of insects. These losses are concomittant with dramatic changes in their centromeric architecture in which each independently transitioned from monocentricity (centromeric determinants localized to a single chromosomal region) to holocentricity (centromeric determinants dispersed over entire chromosome). This implies that CenH3 function must have been acquired by another protein that localizes chromosome-wide resulting in the spread of the kinetochore over large parts of the chromosome and ultimatively giving rise to holocentromeres. In our ongoing work we are characterizing these unique CenH3-deficient kinetochores to find the first component that has functionally replaced CenH3 thereby elucidating the transition from monocentric to holocentric chromosomes in insects

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Using the B chromosome of maize to study the remarkable fluidity of centromere function

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The B chromosome of maize is a supernumerary chromosome that is neither necessary nor detrimental at low copy number. Sequencing of the B chromosome has revealed that it is largely composed of repeats of various types several of which are specific to this chromosome and that the genes are predominantly pseudogenized. Homologies to the normal A chromosomes indicate an ancient progenitor as well as a more recent addition raising interesting questions about the origin of the B and its deterioration. The B chromosome exhibits an accumulation mechanism that drives its presence in populations despite that fact that it is basically inert. The centromere of the B chromosome of maize contains a specific repeat that is interspersed along its length. This fact has facilitated studies of this particular centromere because it can therefore be studied against the background of the other centromeres in the cell that have similar repeat composition among themselves. Cases of inactivation of the B centromere led to the realization that centromere activity could be turned off regularly and inactivation can even be directed in a tug of war between large and derived smaller versions of the B centromere. In collaboration with the Fangpu Han lab (Chinese Academy of Sciences Beijing) many examples of de novo centromere formation upon loss of the B centromere or its inactivation have demonstrated that centromeres can form over unique DNA regularly. The experimental evidence suggests that centromere inactivation or de novo formation can occur over the span of a single cell cycle and then is epigenetically inherited for generations.

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Genetic novelty in essential centromeric proteins in Drosophila

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Chromosome segregation is essential for the faithful transmission of genetic information to daughter cells during cell division. In eukaryotes, accurate chromosome segregation relies on specific chromosomal regions called centromeres, which recruit kinetochore proteins that help mediate spindle attachment to chromosomes during meiosis and mitosis. In many eukaryotes, catastrophic defects in chromosome segregation and lethality result from deletions of the specialized histone H3 variant CenH3, which is the foundational protein of the kinetochore; Despite its essential role in eukaryotic chromosome segregation, *CenH3* and other essential porteins evolve rapidly in plants and animals. I will present two case-studies of genetic innovation in centromeric proteins in Drosophila. One involves a gene duplication of a heterochromatin protein, whose daughter gene acquired an essential chromosome segregation function in *Drosophila melanogaster* (Ross et al. Science 2013). Second is gene duplications of the *CenH3* gene in Drosophila species, which may have specialized for germline centromeric function.

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Chromosome gone wild: consequences of centromere-associated female mieotic drive in *Mimulus*

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Centromeres, which mediate faithful chromosomal segregation in mitosis and meiosis, may also act as selfish genetic elements. Specifically, a centromeric variant that can preferentially move to the egg pole during asymmetric female meiosis (found in most plants and animals) will gain a transmission advantage and spread despite any costs to individual fitness. Centromeric drive is posited to play important roles in shaping karyotypic variation, kinetochore protein evolution, and even the development of species barriers, but there has been little direct evidence of either centormeric drive or its effects. In *Mimulus guttatus* (yellow monkeyflower), a centromere-associated driver (*D*) segregates within a natural population, providing a rare opportunity to examine the costs and consequences of this theoretically ubiquitous phenomenon. Genomic data show that *D* (and large associated chromosomal region) has recently swept to intermediate frequency (~35%), and field fitness measures indicate that strong individual costs should favor the evolution of suppressors. Genetic mapping experiments point to one of *Mimulus*'s two copies of the centromere-specific histone (CenH3A), as a potential suppressor, and population genomic analyses reveal a strong signature of recent positive selection on CenH3A in the *M. guttatus* population with centromeric drive. These analyses ground-truth theoretical arguments for antagonistic coevolution between the protein and DNA components of centromeres, and suggest that centromeric drive does indeed have profound effects on both genic and chromosomal evolution.

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Cell biological mechanisms of meiotic drive

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Biased chromosome segregation in female meiosis I in violation of Mendel's First Law is a form of meiotic drive leading to preferential retention of a chromosome in the egg at the expense of its homolog which is lost in the polar body. This phenomenon has significant implications for chromosome evolution but the underlying cell biological mechanisms remain unclear. Using mouse as a model system we previously showed that differences in centromere strength between homologous chromosomes predict the direction of drive. Stronger centromeres manifested by increased kinetochore protein levels and altered interactions with spindle microtubules are preferentially retained in the egg. We now provide correlative evidence that differences in centromere strength are associated with differences in centromere DNA sequence. This finding is consistent with models that invoke meiotic drive to explain centromere expansion. We also show that drive depends on asymmetry in microtubules of the meiotic spindle. Our observations suggest that signals from the oocyte cell cortex regulate microtubule dynamics to create an asymmetric spindle and that homologous chromosomes with centromeres of different strengths orient preferentially on this asymmetric spindle leading to biased chromosome segregation.

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Repetitive DNA dynamics and hybrid incompatibilities

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Repetitive DNAs including transposable elements (TEs) and non-coding satellites colonize eukaryotic genomes. Their ability to rapidly evolve in sequence and copy number makes them a major driver of genome evolution. Genetic studies of hybrid sterility and lethality (hybrid incompatibilities, or HIs) demonstrate that HI-causing genes often localize to repeat-rich heterochromatin and repress repetitive DNA sequences, suggesting that repetitive DNA is an indirect cause of HI. We have also discovered one case where a species-specific satellite DNA directly causes hybrid lethality between Drosophila species.

These findings argue for the importance of a deeper understanding of repetitive DNA variation, but repeat-rich regions of most eukaryotic genome sequences are under-assembled or absent. We have developed new methods to identify and quantitate repetitive DNA from unassembled genome sequences, and are applying them to comprehensively analyze repetitive DNA from hundreds of natural population samples of Drosophila. We have discovered significant levels of polymorphism for different repeat families and are currently assaying samples with divergent repeat structures for effects on chromosome segregation.

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The Y chromosome contributes to sex-specific aging in *Drosophila*

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Heterochromatin suppresses repetitive DNA, and a loss of heterochromatin has been observed in aged cells of several species, including humans and Drosophila. Males often contain substantially more heterochromatic DNA than females, due to the presence of a large, repeat-rich Y chromosome, and male flies generally have shorter average life spans than females. Here we show that repetitive DNA is mobilized more rapidly in old male flies relative to females, and repeats on the Y chromosome are disproportionally derepressed during aging. This is associated with a loss of heterochromatin and an enrichment of active histone marks at repetitive elements during aging in male flies, and a general redistribution of repressive chromatin marks in aged males. By generating female flies that contain a Y chromosome (XXY flies) and male flies lacking a Y (X0) or containing two Y's (XYY), we show that repeat derepression and average lifespans are directly correlated with the number of Y chromosomes in Drosophila. Thus, our data show that sex-specific chromatin differences contribute to sex-specific aging.

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The essential tension: understanding the balance of genome defense and genomic autoimmunity

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Transposable elements (TEs) are key determinants of variation in genome architecture and they can spread rapidly in sexual species. While there are abundant examples of TE mediated adaptation TEs are mostly harmful agents of mutation and chromosome rearrangement. In light of the burden that TEs impose mechanisms of genome defense by RNA silencing have evolved to limit their harm. However as with any system of immunity there is an inherent challenge to distinguishing "self" from "non-self". In the case of genome defense by piRNAs this occcurs when TE insertions nucleate transcriptional silencing of neigboring genes. Through backcrosses in D. virilis we have shown that these effects can be imposed across multiple generations even in the absence of the initiating TE insertion. Therefore "genomic autoimmunity" can be seen as a cost born across generations. Additional genetic analysis of hybrid dysgenesis in D. virilis also shows that protection against TE-induced sterility is likely dependent on the dose of silencing RNAs that match the broader population of resident TEs. These studies therefore reveal a key tension between maintaining a sufficient dose of genome defense and minimizing off-target effects. This tension may explain our observation that a primary response to changes in the TE landscape is increased translational efficiency in the piRNA machinery. We propose that the tension between genome defense and genomic autoimmunity plays a crucial role in shaping the evolution of RNA silencing pathways.

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Evolution of meiosis in response to genomic and habitat challenges

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Meiosis is essential for the fertility of eukaryotes and its core structures and progression are conserved across kingdoms. Nevertheless, its protein components are often less conserved in primary sequence than we might expect, and meiotic proteins sometimes show evidence of having experienced directional selection. Why? What challenges does meiosis face that might cause it to adaptively evolve? How are meiotic functions changed in response? Evidence from a range of studies shows that two important factors that can challenge the stability of meiosis and drive evolutionary responses are genome duplication and environmental factors, especially temperature. To understand how meiosis evolves in response to challenges, we use *Arabidopsis arenosa*, which occurs naturally as an autotetraploid and a diploid, with both cytotypes having colonized a range of habitats. We found that eight interacting meiotic proteins critical for axis formation and synapsis show strong evidence of having been under selection in the tetraploid lineage. This is associated with a reduction in crossover number in tetraploids, and a greater tendency for terminal localization of chiasmata. More recently, we found that distinct alleles of two of the same genes under selection in tetraploids, also show strong evidence of having been under selection in a diploid *A. arenosa* lineage. This lineage colonized a warmer lowland habitat (the ancestral form is found in cooler mountain environments) and we found that this lineage evolved greater temperature tolerance of meiosis. The finding that distinct alleles of the same genes came under selection twice independently suggests that at least within species the solutions available to alter meiosis in response to different challenges may be relatively constrained, or that different challenges stress the same subsets of systems (in this case the axes and synaptonemal complex) in similar ways.

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The evolutionary turnover of recombination hotspots contributes to speciation in mice

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Meiotic recombination is required for segregation of homologous chromosomes and is essential for fertility. Recombination predominantly occurs at recombination hotspots defined by the speciation gene *Prdm9*. Gradual erosion of hotspots by gene conversion is predicted to reduce reproductive fitness, putting selective pressure on *Prdm9* to change its DNA binding specificity to define new sets of hotspots. To understand the dynamics of hotspot turnover and the role of *Prdm9* in speciation we mapped recombination hotspots in mouse subspecies carrying different *Prdm9* alleles and in their hybrids. We found that hotspot erosion indeed reduces recombination potential, and gene conversion in populations with different *Prdm9* alleles increases sequence diversity specifically at hotspots that become active in the hybrids. As such hotspots are depleted for crossover formation, we propose that sequence divergence generated by hotspot turnover creates impediments for recombination in hybrids, leading to meiotic failure, reduction in fertility and eventually, speciation.

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Defying the meiotic paradigm: sex chromosome pairing, recombination, and segregation in mammals

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The production of haploid gametes during meiosis is dependent on the homology-driven processes of pairing synapsis and recombination. On the mammalian heterogametic sex chromosomes these key meiotic activities are restricted to the pseudoautosomal region (PAR) a short segment of near-perfect sequence homology between the X and Y. In many species including humans pairing or recombination failure between the X- and Y-linked PARs is associated with infertility and sex chromosome aneuploidy. Despite its important roles in meiosis the PAR is rapidly evolving both at the level of DNA sequence and structure. These dueling characteristics of PAR biology raise the unresolved question of how faithful sex chromosome segregation is buffered against the rapid accumulation of homology-disrupting mutations. To begin to address this open question I am investigating the interplay of PAR evolution and function in two interfertile house mouse subspecies characterized by structurally divergent PARs *Mus musculus domesticus* and *M. m. castaneus*. Previously intersubspecific *M. m. domesticus* and *M. m. castaneus* F1 hybrids were shown to harbor an increased frequency of pachytene spermatocytes with unpaired sex chromosomes. Building on these observations I show that XY recombination is also reduced in this intersubspecific hybrid genetic background. However contrary to predictions the observed reduction in XY pairing and recombination in hybrids is not associated with reduced fertility or an increased frequency of sex chromosome aneuploidy relative to the parental subspecies. Together these results demonstrate that accurate sex chromosome disjunction is robust to perturbations of the standard meiotic paradigm in house mice. These findings add to mounting evidence that the mechanisms of meiotic sex chromosome pairing and recombination are fundamentally distinct from those governing the autosomes and suggest that the evolution of this important genomic region may not be as rigidly constrained by its meiotic functions as previously hypothesized.

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Avian W and mammalian Y chromosomes convergently retained dosage sensitive regulators

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We sequenced a female-specific chromosome, the W chromosome of the chicken, and identified 28 genes or gene families mapping to three euchromatic blocks or chromomeres. All 28 genes have homologs on the Z chromosome, and they are the remains on the chicken W of 685 genes found on the autosomal ancestors of avian sex chromosomes. Analyzing the 28 ancestral genes of the chicken W chromosome, together with ancestral W-linked genes predicted in 13 other avian species, we found that, like mammalian Y chromosomes, avian W chromosomes were shaped by selection to maintain the ancestral dosage of homologous sex chromosome gene pairs that function as broadly-expressed regulators of key cellular processes. Indeed, the human autosomal orthologs of three chicken Z-W gene pairs are implicated in congenital disorders traced to haploinsufficiency. We propose that, like the Y chromosome in man, the W chromosome is essential for the viability of the heterogametic sex in birds.

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The fragile Y hypothesis – A role for Y-chromosome aneuploidy in sex chromosome evolution

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Y-chromosome loss leading to XO sex chromosome systems are common in many male heterogametic groups. However despite a clear understanding of how Y-chromosomes are expected to decay our understanding of why some lineages frequently lose Y-chromosomes while others do not is limited. The fragile Y hypothesis proposes that in species with chiasmatic meiosis the rate of Y-chromosome aneuploidy and the size of the recombining region have a negative correlation. The fragile Y hypothesis provides a number of novel insights not possible under traditional models of sex chromosome evolution. Specifically, increased rates of Y aneuploidy may impose positive selection for gene movement off the Y; translocations and fusions which expand the recombining region; and alternative meiotic segregation mechanisms (achiasmatic or asynaptic). This hypothesis as well as existing evidence for the frequency of Y-chromosome aneuploidy raise doubts about the the long-term stability of the human Y-chromosome despite recent evidence for stable gene content in older non-recombining regions.

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Multiple sex chromosome transitions in Diptera

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In many species, including many mammals and insects, sex is determined by the presence of sex-chromosomes. Dipteran insects, such as Drosophila melanogaster, generally have XY sex chromosomes and a conserved karyotype, consisting of six chromosomal arms (5 large rods and a small dot). This XY pair of Drosophila was assumed to be ancestral and shared by most Dipteran insects, an assumption supported by the apparent homology of the mosquito and Drosophila X-chromosomes. Here, we analyze the genomes of 37 species belonging to 22 different families of Diptera, and uncover tremendous hidden diversity in sex chromosome karyotypes. The most frequently sex-linked chromosome is the small dot chromosome, presumably reflecting the ancestral karyotype of higher Diptera. However, we also identify species with undifferentiated sex chromosomes, and others where a different chromosome replaced the dot as a sex chromosome, or where up to three chromosomes became incorporated into the sex chromosomes. While surprising in itself, this diversity also allows us to test theories of sex chromosome evolution more systematically than was previously possible. Our multi-species transcriptome analysis shows that dosage compensation has evolved multiple times in flies, consistently through upregulation of the single X in males. However, X chromosomes generally show a deficiency of genes with male-biased expression, possibly reflecting sex-specific selective pressures. These species thus provide a rich resource to study sex chromosome biology in a comparative manner, and show that similar selective forces have shaped the unique evolution of sex chromosomes in diverse fly taxa.

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Sex chromosomes in the Drosophila male germline: speciation and regulation

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Sex chromosomes play a special role in the evolution of postzygotic reproductive isolation. For reasons that remain unclear the X chromosome accumulates genetic factors that cause interspecific hybrid sterility several times faster than the autosomes. The rapid evolution of hybrid incompatibility on the X almost certainly contributes to Haldane's rule, the large X-effect and the relative paucity of X-linked interspecific gene flow between species. I will present findings from our efforts to determine why the X is a hotspot for hybrid incompatibilities in *Drosophila*. Using an ultrahigh-resolution genetic mapping approach we seek to determine the molecular functions and evolutionary histories for a panel of X-linked hybrid male sterility genes. As a complementary approach we also investigate several general explanations for the special role of the X chromosome in hybrid male sterility and make new discoveries concerning the specialized transcriptional regulation of the X in the *Drosophila* male germline.

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Sex and speciation: did sex chromosome degradation and turnover trigger mammalian divergence?

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Here I propose that the evolution of the sex determining *SRY* gene and definition of a novel XY chromosome pair in therian mammals ~166 MYA imposed a reproductive barrier with the ancestral population of mammal-like reptiles and triggered the speciation event that led to the evolution of therian mammals. I also propose that more recently (~145 MYA) Robertsonian fusion of the therian XY pair with an autosome posed a reproductive barrier that promoted divergence of eutherian (placental) mammals and marsupials. Humans and other therian mammals share a sex chromosome pair composed of a highly conserved X and a small Y chromosome which progressively degenerated and specialised. The male-dominant SRY gene on the Y diverged from its X-borne partner SOX3 about 166 MYA the time that therians diverged from prototherian mammals (monotremes such as platypus). In reptiles and even monotremes this XY pair is represented by autosomes. The bizarre X1X2X3X4X5Y1Y2Y3Y4Y5 in monotremes with homology to the bird ZW suggests that the sex chromosomes of reptile-like mammals were terminally degraded. Translocation with autosomes stabilised this system in monotremes wheareas the evolution of SRY led to sex chromosome turnover in therians. Hybrids between animals with the ancient system and the new SRY system would have had high frequencies of sex reversal intersex development and infertility ensuring divergence of prototherian and therian mammals. Several modern rodent lineages have variant sex determining systems (including complete loss of the Y) suggesting that rodents are undergoing a new explosion of speciation driven by Y chromosome degradation and sex chromosome turnover. This theory conflicts with the long-prevailing paradigm that speciation results from accumulation of small mutational differences in isolated populations but receives support from the many groups of fish and reptiles in which closely related species have different sex determination mechanisms and interspecies hybrids are infertile.

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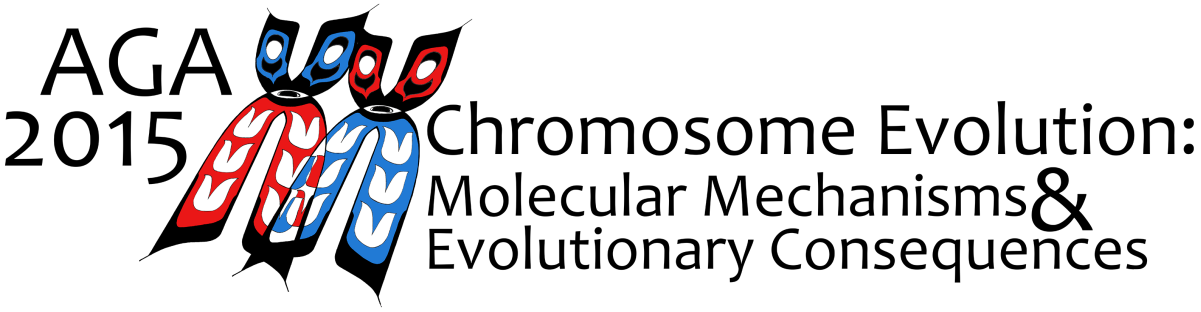
POSTER ABSTRACTS



Speciation, genomics and chromosome rearrangements in cryptic butterfly species

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Understanding the key forces underlying population differentiation and speciation, the forces that ultimately generate biodiversity, is a fundamental quest in evolutionary biology. To get full insight into these processes, identification and characterization of the genetic underpinnings of reproductive isolation is key. Until recently, speciation genetic research was limited to identification of post-zygotic incompatibility genes between deeply divergent lineages of model organisms that easily could be reared in the lab. However, to pinpoint the genetic basis of genes that play a role in speciation in nature, recently diverged taxa from natural populations need to be examined.

We know from previous research that specific regions of the genome may act as drivers of differentiation and these are frequently associated with chromosome inversions. Of particular interest is therefore to investigate the mechanisms behind chromosome rearrangements and how they affect genome evolution and population differentiation processes.

To address these questions we work with three closely related cryptic species of the European wood-white (Leptidea) butterflies. The three species split recently and the traits involved in isolation are most likely related to genital morphology, behavior and chemical signaling. In addition, one of the species has extreme variation in karyotype between populations and show signs of ongoing speciation where crosses between populations with distinct karyotypes result in hybrids with reduced fitness. By using a combination of classical genetic mapping, candidate gene analysis and comparative genomics we try to identify and characterize the genetic regions that might have been involved in the build-up of reproductive isolation between these cryptic species of butterflies and to investigate if any of these loci are associated with chromosome rearrangements.

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Degeneration of the Y chromosome of *Rumex hastatulus*: a search for signs of adaptation

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Genetic sex determination, including X and Y chromosome systems, has evolved many times independently in plants and animals. Furthermore, and of key interest to this project, the chromosomes responsible for sex determination repeatedly become heteromorphic: the Y is genetically degenerate due to its loss of recombination. The degeneration of the Y may be due to the fixation of slightly deleterious alleles because of a reduced effective population size caused by the removal of strongly deleterious mutations (Background selection) (Charlesworth, 1994) or because beneficial alleles cannot fix as fast on the Y as on the X, since beneficial alleles on the nonrecombining Y chromosome are more often in high linkage disequilibrium with deleterious alleles (Ruby-in-the-rubbish) (Rice, 1996; Kim and Orr, 1998). Here, I propose Whole Genome Sequencing of both sexes in both races of the sorrel *Rumex hastatulus* to further establish what proportion of Y-linked genes has fully degenerated as compared to silenced. To investigate the Ruby-in-the-Rubbish hypothesis, we will investigate what proportions of positively selected genes found on the X chromosome show signs of gene silencing and/or gene loss. If differential positive selection on the X chromosome was important in Y gene silencing and/or deletion, we would expect to see an enrichment of gene silencing and loss for homologs of X-linked genes that show evidence for positive selection. Finally, we will generate a linkage map for each sex chromosome race. We hope to use the genetic linkage map to test whether the non-recombining region of the Y-chromosome was established in strata, and investigate the spatial patterns of gene silencing and loss.

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Fine-scale recombination rate variation on the *C. elegans* X chromosome

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Meiotic recombination is a major driver for increasing genetic diversity within species. However, variation in recombination rate exists within and across chromosomes, which potentially affects how different regions of the genome evolve. In the genetic model nematode, *Caenorhabditis elegans*, there are broad-scale differences in recombination rate across the lengths of its six chromosomes, each of which is characterized by a gene-dense central region with a lower recombination rate flanked by the two chromosome arms with higher recombination rates. However, we do not know the extent of variation within these large domains. To address this gap in our understanding, we generated a large collection of near-isogenic lines that were screened for a single recombination event occurring within a 1.5 Mb region on the left arm of the X chromosome. From 814 genotyped recombinant lines, we found significant variation in recombination rate within this region. However, consistent with prior work in *C. elegans*, we do not find evidence for local recombination hotspots. Covariates that explain a significant part of the deviance in a generalized linear model are dependent on the scale of analysis. GC content is significantly negatively associated with recombination rate variation at approximately 25 kb resolution, but not at 5-10 kb resolution. This result is in contrast with what has been found in other organisms, including mammals, *Apis mellifera*, and *Drosophila melanogaster*. We also identified several small (4-6 bp) DNA motifs that are associated with recombination rate at these different resolutions. These covariates may underlie higher-order biological mechanisms, such as nucleosome positioning and chromatin conformation, that are mediating sites of crossover events.

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The evolution of sex-specific specialization on neo-XY chromosomes

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Sex chromosomes play an important role in speciation, and particularly in the evolution of postzygotic isolation. Surprisingly, how sex chromosome turnover may facilitate the evolution of new species has received little attention. The mountain pine beetle has recently formed (neo) sex chromosomes that occurred via a fusion of the ancestral X to the largest autosome. These types of fusion have the potential to drive rapid and dramatic changes in genome architecture as they thrust what was once an autosome into sex biased transmission and differing male/female selective environments. Previous analyses suggest that there is polymorphism for deletions on the neo-Y and that they correlate with reproductive incompatibilities. Here, we describe analyses of gene expression data aimed at determining the level of divergence in expression of the neo sex chromosomes. We generated genome-wide expression data (RNA-seq) from beetle heads, ovaries and testes. We find that the ancestral X chromosome shows female biased gene expression. We also find that the neo-Y is enriched for testis specific genes. Interestingly, neo-X expression shows little sex biased expression. Our results reveal that the neo sex chromosomes appear to be on their way to becoming heteromorphic in gene content and expression and neo-Y expressed genes are present in deletions.

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A non-recombining chromosome is associated with social organization in three major lineages of Formica ants

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Ants have repeatedly evolved a polygynous (= multiple queens per colony) social structure from a monogynous ancestor. We recently identified a non-recombining supergene, or "social chromosome" associated with these alternative social structures in Formica selysi. Here, we test whether the social chromosome is specific to F. selysi, or shared across other Formica species. We find that monogynous and polygynous individuals of other species in different subgenera are much more divergent at the social chromosome than across the rest of the genome. All species we tested harbor two divergent haplogroups at this chromosome, suggesting that this supergene may control social organization in many species throughout the genus. Comparison of genetic maps of the alternative haplotypes shows at least three large-scale structural rearrangements in the non-recombining region. The evolutionary strata on this chromosome will shed light on the evolution of supergenes and the genetic basis of social behavior.

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Sex-specific aging in Drosophila: a role for the Y chromosome?

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Differences in lifespan between males and females have been observed across many taxa, and in most XY species, males have a shorter average lifespan than females. Accumulating evidence suggests that chromatin misregulation, in particular loss of heterochromatin associated with aberrant expression of transposable elements (TEs), contributes to organismal aging. Our previous research has demonstrated that male and female Drosophila differ in their chromatin landscape, especially with regards to the distribution of heterochromatin, and we posited that the large, repeat-rich Y chromosome is responsible for much of these differences. Our current work seeks to address whether sex-specific chromatin is correlated with differences in lifespan between males and females, and whether the presence of the Y chromosome contributes to both observations. We addressed these questions by collecting lifespan data from wildtype *D. melanogaster* males and females, as well as XXY females and XO and XYY males. We also collected RNA-seq and ChIP-seq data from young and old individuals of these various karyotypes. Our results suggest that the Y chromosome does contribute to sex-specific chromatin as well as the shorter male lifespan; individuals with a Y chromosome, regardless of whether they are male (XY) or female (XXY), have a shorter lifespan than individuals without a Y chromosome (XX females or XO males). We find that while both sexes show an increase in TE expression and a loss of heterochromatin during aging, both changes are significantly greater in males than females. Additionally, repetitive elements that are Y-linked show more misregulation in males than other repeats in the genome. We suggest that the Y chromosome contributes to sex-specific chromatin differences and is particularly susceptible to misregulation during aging, likely contributing to the differences in lifespan we see between males and females.

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Flow cytometric detection of holocentric chromosomes in plants

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Holocentric structure of chromosomes is a stable species-specific feature which has evolved independently in various eukaryotic lineages of plants and animals. From historical reasons monocentric chromosomes are considered “normal state” and holocentric chromosomes “peculiarity”. Therefore monocentric chromosomes are automatically assumed in all lineages in which holocentrism has not been proven. Whereas chromosomes were counted in approximately 30% of plant species so far their structure has been analyzed in a much smaller portion of them. Thus the distribution of holocentric chromosomes across the tree of life is very likely an underestimate of their actual incidence. This underestimate is further deepened by the limits of current detection methods of holocentrism which rely on karyological microscopic techniques that are time consuming require a relatively high level of expertise and may easily fail when chromosomes are small and numerous which is often the case in holocentrics.

We introduce a new microscope independent method that has a potential for massive screening for holocentrism in plants. It is based on a flow-cytometric detection of endopolyploid cells in tissues formed after gamma irradiation. We have analyzed 13 monocentric and 8 holocentric species and found that while gamma irradiation induces endopolyploidy in monocentrics it has negligible effects on holocentrics. The application of this method does not require any special skills and can be applied across whole plant phylogeny. The knowledge of phylogenetic distribution of holocentrism could help to solve the fundamental mystery of holocentrism: Why this alternative chromosomal structure has repeatedly evolved in eukaryotic lineages?

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Mechanism of centromere inactivation on a neo-Y chromosome in threespine stickleback fish

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Chromosome number varies greatly among species and can even vary between closely related species. Variation in chromosome number results from chromosomal fission and fusion events. Chromosome fusions can play a beneficial role in evolution; for example, fusions between autosomes and sex chromosomes can link together autosomal genes with different fitness effects in the two sexes with sex-determination genes on sex chromosomes. While such fusions can be beneficial, many chromosome fusions are likely detrimental because they result in a dicentric chromosome. Because dicentric chromosomes are inherently unstable, one centromere must undergo "centromere inactivation" to allow for the proper segregation of the entire chromosome. Centromere inactivation may occur by either a genetic deletion of one centromere, or by epigenetic inactivation via the removal of the centromere specific histone variant (CENP-A). The Japan Sea population of threespine sticklebacks (*Gasterosteus aculeatus*) has a fusion between the ancestral metacentric Y chromosome and an acrocentric autosome. This "neo-Y" chromosome has been fixed in the population, suggesting it only has one active centromere. To begin to test which mechanism of centromere inactivation has occurred on the neo-Y, I needed to first identify the centromeric repeat in stickleback. Using both computational and experimental approaches, I identified a repeat that hybridizes to the centromeric constriction on almost all of the threespine stickleback chromosomes. However, preliminary data suggests that the ancestral Y chromosome centromere is different. Ongoing experiments will identify the Y-specific centromere and then assess whether genetic or epigenetic inactivation has occurred on the neo-Y. Furthermore, we are assessing whether meiotic drive has played a role in the evolution of the neo-Y fusion. The Japan Sea neo-Y chromosome therefore provides an excellent model to understand how and why fusions between Y chromosomes and autosomes have evolved in other systems.

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Introgression of rearranged chromosomes leads to reproductive isolation and species formation

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Chromosomal rearrangements such as translocations and inversions may constitute a rapid means of adaptation to environmental changes. These chromosomal changes may also lead to reproductive isolation through the suppression of recombination between rearranged and non-rearranged sequences or through the perturbation of meiosis and the formation of aneuploid gametes. One major issue with models of chromosomal speciation is that chromosomal rearrangements are viewed as underdominant and thus are unlikely to fix in a population. We recently discovered a new system of natural Saccharomyces populations constituted of two divergent lineages (B and C) that have been isolated and that diverged during the last glaciation in North America. These linages now have partially overlapping geographic distributions and whole genome sequencing analysis shows that they have hybridized in the past. The descendants of these hybrids now appear as what was first thought to be C strains, but further analyses revealed that a small fraction of their genomes was composed of introgressed B-like sequences. These introgressed strains are mainly found within the sympatric area between the parental strains. One of the main features of these introgressed strains is that they show enhanced growth at different temperatures that correspond to phenotypes found in B strains. Introgressed strains also harbor a chromosomal translocation that is present as rare chromosomal variation in the B lineage. We experimentally crossed these strains and found that introgressed strains are partially reproductively isolated from both parental species. Moreover, genome sequencing of the progeny of crosses with the C strains revealed abnormal segregation of the translocated elements, explaining a part of the reproductive isolation. More experiment are needed in order to assess how this chromosomal rearrangement became fixed in this new lineage. This new yeast system illustrates the predominant role of chromosomal changes and hybridization in species formation.

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A powerful tool to delimit and characterize black flies: polytene chromosome rearrangements analysis in Simulium (Psilopelmia) species of Colombia

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Black flies (Diptera:Simuliidae) are a widespread and diverse group of insects that play important roles as vectors, bioindicators and food sources for other species; however, their morphological homogeneity has been a challenge for taxonomists. Our goal in this study was to characterize species of the subgenus Psilopelmia in the larval stage through cytological data, such as inversions, relative positions of 16 chromosome markers in the polytene chromosomes and B chromosome (If present). Samples were collected from 2011 to 2014 at National Natural Parks and localites in the eastern and central mountain range of Colombia.. Larvae were collected in Carnoy´s fixative. Material was stored at 4°C until processed for staining. A total of 112 individuals were analyzed, and their idiograms were established and associated with the respective morphological specimen (e.g., gill histoblast configuration, postgenal cleft shape, and cephalic apotome pattern). Ten idiograms were constructed. Simulium tunja and S. ignescens are very similar morphologically; therefore, the first level of classification was based on the position of the Nucleolar Organizer (NO). Idiograms for which the NO was located in chromosome I were categorized as “like S. tunja” (cytoforms 1 to 6), and idiograms for which the NO was found in chromosome III were categorized as “like S. ignescens” (cytoforms 1, 2 and 3). One additional idiogram was constructed for S. furcillatum. Most of the morphotypes have been assigned with a species name only tentatively because an exact match with the formal names has not yet been found??. In general, a high correspondence between morphological and cytological data was found. However, high diversity was detected in S. ignescens 2, both morphologically and cytologically. Therefore, it is possible that there are some species complexes and phenotypic plasticity. This is the most comprehensive study (morphology-cytogenetics) for Psilopelmia in Colombia, and provides a framework for future research.

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B Chromosome dynamics and sequence evolution in African Cichlid fish

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B chromosomes are non-essential, dispensable, supernumerary chromosomes that are present in addition to the normal ("A") karyotype of an organism. B chromosomes can be regularly found in some, but not all, individuals of a given species. They are estimated to occur in 15% of all eukaryotes covering a wide range of taxa from fungi to plants to animals and even mammals. A smaller number of studies show B chromosomes to play a functional role, including a study that showed B chromosomes were involved in sex-determination of at least one Lake Victorian cichlid species. Here we examine whole genomes of more than 20 populations of Lake Malawi cichlid fish and find B chromosomes present at intermediate frequency in at least 7 species. In each species the B chromosomes are present only in female fish, suggesting a mechanism of drive in females and a probable role in sex-determination. We sequenced the genomes of 12 individual females from the 7 species harboring B chromosomes to characterize the diversity of B chromosomes within Lake Malawi and to identify a set of core sequences present in all Lake Malawi B chromosomes. We characterize the dynamic regions of the B chromosome that are expanding and contracting between different individuals and populations. Comparison of these Lake Malawi B chromosomes to the B chromosome of the Lake Victorian cichlid, *Astatotilapia latifasciata*, reveals a very small amount of sequence overlap between B chromosomes in the two lakes. We find only one intact gene present in the B chromosomes from both lakes suggesting a potential common origin and gene responsible for the B chromosome propagation mechanism. We propose that the remainder of the B chromosome is composed of a certain set of sequences that are free to come and go, occasionally acquiring sequences that provide a function.

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Quantification of repetitive sequence from a sex-associated B Chromosome in Lake Malawi cichlid fish

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While every species has its own typical set of chromosomes referred to as the A chromosomes (As), some species have B chromosomes (Bs) which are supernumery, nonessential chromosomes found in some but not all members of a population. Bs do not follow the typical ploidy rules nor go through meiosis and mitosis in the same manner as A chromosomes. A peculiar case of B chromosomes has been found in several species of Lake Malawi cichlids, where Bs are found solely in females. This study aimed to identify the mechanism by which Bs are maintained in these fishes. Whole genome sequencing of individuals with and without Bs revealed B-specific sequences blocks. SNPs unique to the B chromosome permitted the design of B-specific amplification primers. These primers allowed the rapid identification of B-carriers through PCR. B-carriers are solely female, no males were observed to have the B. Variation in the copy number of B-specific repeats was quantified by qPCR. Cluster analysis of repeat copy number provides evidence that *Metriaclima lombardoi* females carry a single B chromosome, confirming previous karyotyping results. The fact that individuals carry only a single B chromosome is consistent with two potential drive mechanisms: one involving nondisjunction and preferential segregation in a pivotal mitotic division leading to the germline, and the other involving preferential segregation during meiosis I.

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The 21 chromosomes of *Heliconius melpomene*

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Almost all *Heliconius* butterflies have 21 chromosomes but the outgroup genus *Eueides* all have 31. It appears that ten chromosome fusions occurred over a period of about five million years but since then most *Heliconius* species have had no change in chromosome number. I will present a number of major improvements to the *Heliconius melpomene* genome assembly increasing the scaffold N50 to 1.1 Mb and placing 98% of the genome onto chromosomes. Using this improved assembly we are beginning to consider the possible implications of the shift in chromosome number for speciation in *Heliconius*.

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Chromosomal polymorphism and reproductive isolation in natural nascent species pairs

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Salmonids are choice model organisms for speciation cytogenetics: they are pseudo-tetraploids, which, along with small population sizes, is expected to facilitate the evolution and fixation of chromosomal rearrangements. In northeastern North America, two Lake Whitefish (*Coregonus clupeaformis*) lineages have colonized post-glacial lakes ~12,000 YBP, following ~60,000 years of allopatry. A dwarf limnetic form has evolved repeatedly since then from the normal benthic form. Evidence suggests that the accumulation of genetic and possibly chromosomal incompatibilities in allopatry may have facilitated this divergence. We applied conventional and molecular cytogenetic methods to three natural pairs of Lake Whitefish sympatric populations, in order to i) test the null hypothesis of no difference in chromosomal rearrangements between pure lineages and ii) test the hypothesis of chromosomal instability in healthy and malformed hybrid embryos. Karyotypes of parental forms did not show obvious differences at the whole-chromosome level, whereas more detailed cytogenetic analyses revealed an extensive polymorphism partly shared by the three species pairs. Multi-variate analyses refuted the null hypothesis by revealing ongoing genomic rearrangements between these incipient species, consistent with chromosomal divergence in allopatry, followed by gene flow in sympatry. Our results also support the hypothesis of mitotic instability in healthy backcrosses, through an increased intra-individual variance of chromosome numbers. In malformed backcrosses, extensive aneuploidy corresponding to multiples of the haploid number (n=40, 2n=80, 3n=120) was found, indicating meiotic breakdown in their F1 parent. Importantly, these conclusions would have been difficult to reach through sequencing approaches. As such, cytogenetics is highly complementary to genomics, and should not be neglected towards a full understanding of the genomic bases of speciation.

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Bipartite structure of the inactive mouse X chromosome

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We analyzed allele-specific chromatin contacts by a new Hi-C assay that uses DNase I for chromatin fragmentation to evaluate structural changes associated with X inactivation and imprinting in mouse F1 hybrid systems in which alleles can be distinguished based on single nucleotide polymorphisms. Both in vivo (brain) and in vitro (Patski cells) the two X chromosomes have strikingly different 3D configurations. Two superdomains of frequent long-range intrachromosomal contacts separated by a hinge region are specifically observed on the inactive X chromosome. Such a bipartite 3D organization has been also reported in human lymphoblastoid cells. We found that the genomic content of the superdomains is rearranged between species but that part of the hinge region is conserved and located near the Dxz4/DXZ4 locus that binds CTCF on the inactive X. In mouse the hinge region also contains a minisatellite Ds-TR adjacent to a promoter with strong CTCF binding. Both Dxz4 and Ds-TR bind nucleophosmin and are enriched in nucleolus-associated chromatin suggesting anchoring to the nucleolus. Genes that escape X inactivation and regions enriched in CTCF or RNA polymerase are preferentially located on the periphery of the inactive X while LINE1 elements are preferentially on the interior. Genes subject to silencing exhibit fewer detectable short-range intrachromosomal contacts than escape genes. This transcription-coupled pattern is also evident for imprinted genes in which more chromatin contacts are detected on the expressed allele suggesting greater constraint on the organization of expressed genomic regions.

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Drosophila Y chromosomes preserve a history of genetic conflict

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The fundamental cellular processes of meiosis and gametogenesis can be exploited by selfish genetic elements in order to distort transmission in their own favor. Such intragenomic conflict can lead to a prolonged arms race between distorter and suppressor, which creates an evolutionary paradox: Genes with essential roles in highly regulated processes such as chromosome segregation, can also have highly dynamic evolutionary histories, including duplication and a rapid rate of amino acid replacement. Recent work in the house mouse has identified a massive amplification of three meiosis-related genes on the Y chromosome, likely due to a history of segregation distortion and suppression between the mouse X and Y chromosomes. We have recently identified a similar phenomenon in four species of Drosophila, involving genes with functions related to meiosis and gametogenesis. Across the Drosophila genus, these genes are single-copy and highly conserved, however in certain lineages, they have been tandemly duplicated on the X chromosome and highly amplified on the Y chromosome. These results expand the list of candidate sex-ratio distorters in Drosophila and support a growing body of work that suggests genetic conflict plays an important role in the evolution of gene content and genome architecture.

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Structure and decay of a proto-Y region in Tilapia, *Oreochromis niloticus*

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Sex-determination genes drive the evolution of adjacent chromosomal regions. Sexually antagonistic alleles near sex-determination genes favor alterations to the genomic architecture, such as inversions, to reduce recombination. Gradual decay of the clonally inherited alterations soon begins affecting gene expression and functionality, thus applying selective pressures to evolve dosage compensation mechanisms. In this study, we used whole genome sequencing of pooled families to identify and characterize the structure of a young sex chromosome in Tilapia, *Oreochromis niloticus*. We demonstrate an 8.8Mb block of differentiation with an increased density of SNPS which encode for high impact functional changes. We also report gonad transcriptome data that shows a significant female-biased expression pattern within the inversion. This female- biased expression profile indicates the lack of a dosage compensation mechanism within *O. niloticus*. Therefore, this region demonstrates a pattern that would be consistent with an inversion containing a proto-Y.

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Evolutionary history of chromosomal races within the *Cryptocercus punctulatus* complex (Insecta: Blattodea), a dead wood specialist from the Southern Appalachian mountains

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The Southern Appalachian region is topographically complex, was never glaciated, and repeatedly served as a major Pleistocene refuge for numerous forest-dependent species. Flightless terrestrial invertebrates may be excellent models for reconstructing how past environmental changes structured biodiversity in this hotspot because, unlike more mobile vertebrates, they were able to persist in situ within sheltered microrefugia. Wood-feeding insects in particular would have tracked the changing distribution of forest habitats with very high fidelity. Here we investigate the evolutionary history of a wingless wood-feeding cockroach, *Cryptocercus punctulatus*. This species complex is comprised of four chromosomal races (male 2n = 37, 39, 43 or 45), with karyotypic differences due to Robertsonian changes involving the fission or fusion of non-homologous autosomes. These races have parapatric distributions, but range boundaries do not currently coincide with any obvious physiogeographic barriers or ecotones. It is therefore unclear whether historical scenarios in which genetic drift is the primary evolutionary force (e.g., allopatric divergence during isolation in different refugia, followed by re-expansion) could plausibly explain the spatial distribution of genetic diversity in this group. To address this, we conducted phylogeographic analyses based on mitochondrial and nuclear DNA sequence data from ~300 individuals sampled from ~100 sites in the Southern Appalachians. To determine whether functional differences among races exist, which may be indicative of the action of selection (i.e., local adaptation), complementary analyses of geo-spatial data were used to assess niche overlap (and by extension, ecological exchangeability).

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Identifying sex determination genes on the young asparagus Y

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Garden asparagus (*Asparagus officinalis*) is a dioecious species with a recently evolved, homomorphic sex chromosome pair which makes it ideal for studying the earliest events in sex chromosome evolution. One parsimonious evolutionary path from hermaphroditism to dioecy and a sex chromosome pair is that at least two genes, a suppressor of femaleness and a promoter of maleness, must successively occur in perfect linkage on a Y. We have explored this hypothesis in garden asparagus by genetically mapping sex determination to a small (<2Mb) non-recombining region on the proto-Y chromosome that actively differentiates males (XY) from females (XX). We have identified two independent male-to-hermaphrodite mutants that implicate a single gene in this non-recombining region on the Y as responsible for dominantly interrupting pistil development. Anther development is not affected in these mutants. To identify the gene(s) responsible for anther sterility in XX females, we assessed gene expression profiles in male and female spears and identified known anther development genes that exhibit male-biased expression. A small subset of genes exhibiting male-baised expression have also been identified as sex-linked. By identifying a female suppressor gene that does not influence flower development, these results support the hypothesis that at least two genes are necessary in the conversion of an autosome to a sex chromosome pair.

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Molecular evolutionary analyses reveals positive selection in the rapidly-evolving synaptonemal complex in the Drosophila genus

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The synaptonemal complex (SC) is a highly conserved meiotic structure seen across eukaryotes that functions to hold the homologs together during meiosis and facilitate exchange. Five Drosophila exclusive proteins have been identified as the components: C(3)G, C(2)M, Cona, Ord, and the newly identified Corolla. Each protein is necessary for proper meiotic function; mutations lead to reduced crossing over and chromosomal non-disjunction. Despite the conserved nature of the SC and the key role that these five proteins have in meiosis in *D. melanogaster*, they display little apparent sequence conservation outside the genus. We have performed a molecular evolutionary analysis to determine the nature of selection that might explain this lack of apparent conservation. Several species of Drosophila have no recognizable sequences corresponding to these crucial SC components and identity declines rapidly in diverged species. SC gene sequences are changing more rapidly than the genome wide average and this can in part be explained by the action of positive selection in almost every SC component, with the exception of C(2)M. Interestingly, across the phylogeny, we find no evidence that changes in the rate of evolution of one component can drive corresponding changes in other components. Rather, the rate of evolution is primarily determined by physical position within the SC. Finally, there is evidence of positive selection at the population level in *D. melanogaster* and *D. simulans* suggesting that adaptation in the SC is ongoing. The SC may be evolving due to many different factors including relaxed selective constraint, its role in shaping the recombination landscape, and its interactions with the centromere and role in centromere clustering.

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Exploring ecological consequences of a selfish sex ratio distorting chromosome in a booklouse

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Selfish genetic elements are pervasive in nature and are thought to play a role in several fundamental evolutionary processes (such as sex determination). In many systems selfish genetic elements cause sex ratio distortion in the offspring of organisms that carry them in order to gain a transmission advantage to the next generation. Recently, a maternally transmitted selfish sex ratio distorting element has been discovered in a species of booklouse: *Liposcelis* nr*. bostrychophila*. This species is sexual and contains two female morphs, one that carries the selfish element and produces exclusively female offspring (that also carry the element) while the other does not carry the element and produces both male and female offspring. Genetic evidence suggests that the sex ratio distorting element is a selfish chromosome. The presence of this selfish element in *L.* nr. *bostrychophila* populations has important ecological consequences. Populations containing this selfish element are inherently unstable due to the higher reproductive potential of females carrying the element (who avoid the cost of producing males). Without some counterbalancing force females carrying the selfish element will overtake those not carrying the element, which will result in extinction of the population (due to a lack of males). I am investigating how the ecology of this species allows for the persistence of this selfish element and have found that carrying the selfish element is associated with substantial fitness costs: both in longevity and fecundity. Additionally, other ecological factors such as sex allocation in females not carrying the element likely play a role in the persistence of this element. These investigations have shed light on how this selfish element persists in wild booklouse populations.

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Genomic landscape of long terminal repeat retrotransposons (LTR-RTs) and solo LTRs as shaped by ectopic recombination in chicken and zebra finch

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Transposable elements (TEs) are nearly ubiquitous among eukaryotic genomes but TE contents vary dramatically across phylogenetic lineages. Several mechanisms have been proposed as drivers of TE dynamics in genomes including the fixation/loss of a particular TE insertion by selection or drift as well as structural changes in the genome due to mutation (e.g. recombination). In particular recombination can have a significant and directional effect on the genomic TE landscape. For example ectopic recombination removes internal regions of long terminal repeat retrotransposons (LTR-RTs) plus one long terminal repeat (LTR) resulting in a solo LTR. In this study we focus on the intra-species dynamics of LTR-RTs and solo LTRs in bird genomes. The distribution of LTR-RTs and solo LTRs in birds is intriguing because avian recombination rates (RRs) vary widely within a given genome. We used published linkage maps and whole genome assemblies to study the relationship between RRs and LTR-removal events in the chicken and zebra finch. We hypothesized that regions with low RRs would harbor more full-length LTR-RTs (and less solo LTRs) than regions with high RRs. We tested this hypothesis by comparing the ratio of full-length LTR-RTs and solo LTRs across chromosomes across non-overlapping megabase windows and across physical features (i.e. centromeres and telomeres). The chicken data statistically support the hypothesis that RRs are inversely correlated with LTR-RT density at the level of chromosomes (Spearman's Rank Correlation Test rho = -0.52 p = 0.009) and 1-Mb non-overlapping windows (rho = -0.14 p = 0.008). We also found the ratio of full-length to solo LTRs near chicken telomeres was significantly lower than those ratios near centromeres (Mann-Whitney U Test p = 0.02). Our results suggest a potential role of ectopic recombination in shaping the avian LTR-RT genomic landscape.

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Recombination hotspots of *Ficedula* flycatchers

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Both theory and empirical data indicate that the efficacy of selection is strongly dependent on the recombination rate. Characterizing regional variation in recombination rates across a genome is therefore critical for the understanding the forces that shape patterns of variation in genetic diversity within species and genetic differentiation between species. Genetic linkage map analysis is a traditional method to estimate recombination rates by counting the number of meioses per physical distance in a given pedigree. However the resolution of recombination rate variation in pedigree-based studies is often too coarse (Mb scale) to pick up important fine-scale variation (kb scale). Recently genome-wide patterns of linkage disequilibrium (LD) have been used to estimate ultra fine-scale recombination rate variation and to detect recombination hotspots, which are narrow regions in the genome with unusually high recombination rate as compared to the genomic average. Here we use large-scale full genome re-sequencing data to estimate population-scaled recombination rates (rho) in four populations of collared flycatchers (*Ficedula albicollis*) across western Europe. Our data shows that variation in the population based recombination rate was highly correlated with our previous pedigree based recombination rate estimates ($r= 0.708$, $p<0.0001$) suggesting that our rho estimate captured a substantial amount of the underlying variation in recombination rate across the collared flycatcher genome. We detected about 1000 recombination hotspots with significantly elevated rho; these were characterized by high GC content and enrichment of repeat sequences. In addition we estimated rho for four populations of a close relative the pied flycatcher (*F. hypoleuca*) which shows similar patterns to what we found in the collared flycatcher. Population-specific recombination hotspots and sequence motifs associated with hot-spots will be discussed.

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Characterization of a large vertebrate genome using shotgun and laser capture chromosome sequencing

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The salamander *Ambystoma mexicanum* (Mexican axolotl) is an important model system in regeneration, development and genome/chromosome evolution studies. All salamander genomes are greatly expanded in size relative to other extant tetrapods. Despite the size of the axolotl genome, its gene orders are highly conserved with those of reptilian and mammalian genomes and provide valuable information for reconstructing several features of ancestral tetrapod genomes. As such, the axolotl genome provides both a unique perspective on the evolution of genome biology and presents a major challenge toward the development of genomic resources.

To characterize *A. mexicanum* genome structure, we sequenced whole genomic DNA (600 billion bases). Analysis of the shotgun dataset estimates the genome to be ~32Gb with repetitive sequences making up ~40%. As part of a multipronged approach to reduce assembly complexity, we developed strategies to capture, amplify and sequence individual dyads. Initial analyses revealed the libraries provide high sensitivity and specificity to the linkage groups of the Ambystoma linkage map. Our analyses show that chromosome-targeted sequencing presents an efficient strategy for simultaneously reducing assembly complexity and generating broad-scale scaffolding information for large genomes. In addition to refining the existing map and resolving key events in the evolution of salamander and chicken genomes, this technique has been used to target specific chromosomes of interest, including the salamander's recently evolved sex chromosomes. Libraries developed from this chromosome have been used to create a preliminary assembly for the axolotl sex chromosome. Conserved synteny studies with chicken corroborate previous analyses and have implications for understanding early sex chromosome evolution.

Our pilot studies suggest that the laser-capture sequencing approach can be adapted for other purposes and is applicable to a broad range of biological questions, including chromosomal scaffolding of genomes for organisms that are not amenable to laboratory culture and genomic characterization of microscopically identifiable cells (e.g. cancer or germ cells).

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Evidence for balancing selection and chromosomal rearrangements during ecological speciation in *Ostrinia* moths

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Divergence in seasonal timing of mating is often an important reproductive isolating barrier between species. Seasonal temporal isolation occurs between populations of European corn borer (*Ostrinia nubilalis*) due to differences in when diapause is broken in the spring that results from differences in the length of diapause termination time. Differences between long and short termination time have been previously mapped to a single QTL of major effect (*Pdd*) on the sex (Z) chromosome. We developed a fine scale linkage map of the corn borer Z chromosome and identified a 2 Mb interval containing *Pdd*. Along a latitudinal cline in voltinism in the field loci closely linked to *Pdd* show signs of balancing selection suggesting selection for life cycle fitting into season length may drive the evolution of diapause termination time. We also found that between Z- and E-pheromone strains of corn borers a large region around *Pdd* (~10 Mb) shows reduced recombination and elevated genetic divergence (F_{ST}) indicating that a chromosomal rearrangement (such as an inversion) has occurred in one strain capturing *Pdd* and potentially other genes contributing to reproductive isolation between strains.

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Evolution of centromeric proteins in percid fishes

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We had previously isolated the CENP-A gene from the percid species *Etheostoma tallapoosae* and identified exons of neighboring genes to which we could design PCR primers to amplify this gene from other darter species. Based on the analysis of the CENP-A coding sequence from a total of eleven darter species we were able to show that adaptive evolution of the N-terminal tail of CENP-A has occurred in this family of fishes. A statistically supported determination of this adaptive evolution was obtained even if CENP-A sequences of only three of the most distantly related darter species were compared. To determine if any of the other centromeric proteins adaptively co-evolved with CENP-A we sequenced the genomes of the three distantly related percid species *Percina crypta*, *Etheostoma chuckwachattee*, and *Etheostoma tallapoosae*. We identified orthologs of thirty centromeric protein genes in each of these species and subjected the coding sequences of these genes to Ka/Ks tests of positive selection. Of the thirty genes tested seven showed statistically significant results for positive selection. This data would suggest that selective pressure of centromeric drive may be exerted on a subset of proteins that make up the centromere/kinetochore.

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Sex chromosomes, gene expression and speciation

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One emergent pattern in speciation has been the large role of sex chromosomes in divergence and barriers to gene exchange (Large X effect). Sequence divergence on the X evolves rapidly compared to autosomes (Faster-X) particularly in protein-coding sequences of genes with male-biased or testis specific expression (in male XY systems). A common assumption is that there is also faster-X expression divergence which could lead to the disruption of spermatogenesis in hybrids. Faster-X expression has only been evaluated in few systems and in all cases using whole testis transcriptomes that conflate gene X expression at different stages of spermatogenesis. Here we test the link between faster-X evolution and disruption of X gene expression in hybrids by evaluating gene expression in specific cell types at different time-points in spermatogenesis.

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Holocentromeres are defined by genome-wide centromere-specific repeat arrays interspersing the gene-containing chromatin

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Holocentric chromosomes lack a primary constriction and, in contrast to monocentrics, they form kinetochores distributed along almost the entire poleward surface of the chromatids, to which the spindle fibers attach. Although their kinetochores have been molecularly characterized in a few cases, no centromere-specific DNA sequence could be identified for any holocentric organism so far. It has been proposed that centromere-specific sequences, as described for many monocentric species, might not occur in holocentrics probably due to their different chromosome organization. Here we show that the holokinetic centromeres (holocentromeres) of the Cyperaceae plant species Rhynchospora pubera (2n = 10) are highly enriched in a CENH3-interacting centromere-specific satellite family (Tyba) as well centromeric retrotransposons (CRRh). We propose that Tyba satellite repeat-rich regions are preferred sites for the deposition of centromeric nucleosomes and serve as potential kinetochore attachment sites. The discovery of centromere-specific sequences in a holocentric organism gives further support to the idea that such sequences have a key involvement in the long-term maintenance of centromeres. We show that holocentric chromosomes are composed by multiple kinetochore/centromere units rather than possessing a diffuse kinetochore organization. This observation favors the ancient ‘polycentric’ model and agrees with recent findings for C. elegans. Analysis of BAC sequences identified centromere units ranging from 3 to 16 kb intermingled with gene coding sequences and transposable elements. Methylation of histone marks has further revealed that there is no eu- and heterochromatin compartmentalization at large-scale in the genome of this species, suggesting that the centromere structure effects overall genome organization. As a functional adaption, an apparent coordinated cell cycle-dependent shuffling of centromere units results in the formation of functional (poly)centromeres during mitosis, giving rise to the so-called holocentromeres. Thus, different types of holocentromeres exist, namely with and without centromere-specific sequences.

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Satellite DNA in the *Ancistrus* genome revealed through next generation sequencing

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Satellite DNA are non-coding sequences of 100 to 500 bp with hundreds and thousands copies tandemly repeat. They are located mainly in the pericentromeric and telomeric chromosome regions, besides in some cases in sex-chromosomes. In this work we studied the genome of a siluriforme fish group belonging to the *Ancistrus* genus. This group of species is very interesting from the genetic point of view because they have features like large karyotypic diversity, presence of sex chromosomes systems XX/XY and ZZ/ZW types, presence of supranumerary chromosomes and chromosomal polymorphisms. Cytogenetic tools such analysis as diploid number, NOR and C band has been shown to be good markers for the differentiation of species and populations of Ancistrus, but the molecular cytogenetic techniques such as mapping and analysis of repetitive sequences, is still employed in that group. *Ancistrus* sp used here is from the Currupira river (Mato Grosso-Brazil) and show a 2n=44 ZZ/ZW karyotype. The genome of the species was sequenced through Illumina next-generation sequencing (NSG) and identified the most abundant tandem repeats using the RepeatExplorer software. The RepeatExplorer software analysis revealed the occurrence of 175 clusters being five of them putative satDNAs, larger than 100 bp long. It was constructed primers and the agarose gel after the PCR reaction also indicated satellite DNA features, like ladder pattern. In the next step these sequences will be submit like a probe in the FISH experiments with the intention of knowing the localization of these putative satellite and if they are related with heterocromatic regions, such as those that are present in the sex chromosomes. The results integration the chromosomal and NGS data are powerful tools and permitted a deeper analysis of chromosomes mainly the sex chromosomes, contributing to decipher satDNA organization and sex chromosome evolution, issues that are poorly understood in fish.

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Revalidation of *Leptodactylus brevipes* Cope, 1887 (Amphibia, Anura, Leptodactylidae) based in cytogenetic and molecular evidences

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The genus Leptodactylus comprises 74 species of frogs, distributed in the Neotropics, between Argentina and southern United States, including Antilles. *Leptodactylus brevipes* was originally described by Cope in 1887, from individuals collected in Chapada dos Guimarães, Mato Grosso state, in the Cerrado. This species was later placed in the synonymy of *Leptodactylus petersii* Steindachner, 1864, a species that occurs in the Amazon basin, and described from specimens collected in São Gabriel da Cachoeira, in Amazonas state, Brazil. Now, our studies involving cytogenetics and mitochondrial gene sequencing (COI and 16S) of the individuals collected in those type localities suggest that it is necessary a review in the taxonomic status of the specimens from Chapada dos Guimarães, which exhibits notable external morphological similarity with *L. petersii*. The cytogenetic analyzes show that *L. petersii* from type locality have a 2n = 22 karyotype, as almost all of the other Leptodactylus species studied so far, whereas all karyotyped *L. cf. brevipes* presented 2n = 20 chromosomes. This lowest chromosome number was confirmed by meiotic analysis. The NOR sites in both species were in the proximal region of the long arms of the chromosomes 4. Interstitial telomeric sequences were not present after FISH experiments. The molecular data corroborate cytogenetic findings, suggesting that the specimens analyzed from Amazonia and Cerrado could be different species. Additionally, based in our population analyzes; we provide a new distribution range for *L. cf. brevipes*.

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Evidence for gene flow but ineffective selection on a selfish X-chromosome in *Drosophila neotestacea*

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The sex-ratio X-chromosome (SR) is a selfish chromosome that promotes its own transmission to the next generation by destroying Y-bearing sperm in the testes of carrier males. In some wild populations of *D. neotestacea*, up to 30% of the X-chromosomes are SR chromosomes. We investigated the molecular evolutionary consequences of SR and determined the evolutionary relationship between the standard X-chromosome (ST) and SR. Using a sample of *D. neotestacea* males from throughout the species range, we sequenced SR and ST males at 11 randomly chosen X-linked loci that span the ST X-chromosome, as well as seven randomly chosen autosomal loci. We analyzed patterns of genetic differentiation between the ST and SR chromosomes, performed a phylogenetic analysis to infer the evolutionary history of SR, and compared levels of recombination, linkage disequilibrium, and nucleotide polymorphism between ST and SR. Our results indicate that while SR and ST are strongly differentiated and selection may be acting differently on SR, there is evidence of gene flow between ST and SR through recombination and/or gene conversion. We suggest that gene flow with ST could prevent the degradation of SR from mutation accumulation and allow its long-term persistence at relatively high frequencies.

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Evolutionary history of VSAREP satellite DNA family in *Varanus* (Varanidae, Squamata)

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Characterization of VSAREP satellite DNAs, which were isolated from Asian varanid lizard (*Varanus salvator macromaculatus*) in our previous study leads to understand their molecular structure of C-heterochromatin in the pericentromeric region of chromosome 2q, the centromeric region of chromosome 5, and 3 pairs of microchromosomes. To investigate the nucleotide specificity and evolutionary process of VSAREPs in varanid lizard lineage, dot blot hybridization of VSAREPs were performed to genomic DNA of Asian, Australian, and African varanids. VSAREP sequences were cross-hybridized with genomic DNA of Asian and Australian varanids (*V. salvator macromaculatus*, *V. salvator komaini*, *V. salvator sulfer*, *V. salvator ziegleri*, *V. bengalensis*, *V. rudicollis*, *V. dumerilii*, *V. salvadorii*, *V. acanthurus*, *V. gouldii*, and *V. rosenbergi*). However no hybridization was observed in African varanids (*V. exanthematicus*, *V. niloticus*, *V. jobiensis*, and *V. obor*). This suggests that VSAREPs have originated after Asian and Australian varanids had diverged from African varanids approximately 40 million years ago. Semi-quantitative PCR using VSAREPs specific conserved sequences was conducted to elucidate their genomic organization. The polymeric ladder DNA bands were found in the genome of Asian varanids, and the ladder-like DNA bands in Australian varanids. This implies that VSAREPs are arranged in tandem arrays in Asian varanid genomes, but not in its Australian varanid relatives. The ladder DNA bands were then sequenced randomly, and showed the level of sequence divergence about 30% between Asian and Australian varanids. Cross-species FISH mapping using VSAREPs in Australian varanids identified weak hybridization signals at short arm of one pair of microchromosome in *V. acanthurus* and in pericentromeric regions of chromosome 2q in *V. gouldii* and *V. rosenbergi*. This suggests that VSAREPs were located in the centromeric and pericentromeric regions of some chromosome pairs in the common ancestor of Asian and Australian varanids, followed by lineage specific dispersion and amplification in other chromosomes.

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Gekkota retains the highly conserved linkage homology with other squamate reptiles that have many microchromosomes

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A cytogenetic map of *Gekko hokouensis* (GHO) with few or no microchromosomes, which retains the ancestral karyotype of Gekkota, was constructed with 86 functional genes and compared it with those of four Toxicofera species that have many microchromosomes (*Elaphe quadrivirgata*, *Varanus salvator macromaculatus*, *Leiolepis reevesii rubritaeniata*, and *Anolis carolinensis*) and that of a Lacertidae species (*Lacerta agilis*; LAG) with only one pair of autosomal microchromosomes. Ten pairs of GHO chromosomes corresponded to most of all macrochromosomes and/or macrochromosome arms of the Toxicofera species and eight macrochromosomes of *L. agilis*. This suggests that the linkage groups of Toxicofera and Lacertidae are also highly conserved in *G. hokouensis*, although *G. hokouensis* has a diversified karyotype as well as *L. agilis*. The reorganization of macrochromosomes in Toxicofera, Lacertidae, and Gekkota was mainly caused by centric fusion/fission and chromosomal inversion of several chromosome pairs. However, four GHO chromosomes and one LAG chromosome, which were not homologous to each other, were composed of chromosome segments homologous to Toxicofera microchromosomes. This suggests that repeated fusions of microchromosomes might have occurred independently in each lineage of Gekkota and Lacertidae, leading to the disappearance of microchromosomes and the appearance of small-sized macrochromosomes.

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Analysis of meiotic drive in reshaping chromosome structure in the *Drosophila virilis* subgroup

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There are many differences in chromosome size, shape, and number which results in varied karyotypes among species, and in some cases within species. The mechanisms that drive such karyotype diversity are poorly understood. My goal is to investigate the role of biased segregation in female meiosis, i.e., meiotic drive, as a force that contributes to the evolution of chromosomal form and to the reorganization of karyotypes. The closely related species pair, *Drosophila americana* and *Drosophila novamexicana*, are exemplars for understanding mechanisms of karyotype evolution. Since their recent divergence (~half million years), *D. americana* has evolved two different centromeric fusions: one fusion between the 2nd and 3rd chromosomes, and the other fusion between the X and 4th chromosomes. The 2-3 fusion is fixed in *D. americana*; however, the X-4 centromeric fusion remains polymorphic within the species. However, there are three different inversions associated with the X-4 fusion that could be playing a role in chromosome segregation. My focus is to evaluate the hypothesis that meiotic drive, favors the fused metacentric chromosome forms in *D. americana*, and that it is responsible for the rapid change in karyotype structure in the *D. americana* lineage. Using two different methods, my results reveal that the fused X-4 chromosome in heterozygous females is transmitted to the offspring at a biased frequency of 54%-58% regardless the of the inversion state of the chromosomes. These results suggest that meiotic drive plays a role in reshaping chromosomes in *D. americana*, and that the chromosome fusion is the factor in meiotic drive in *D. americana*, whereas the inversions do not play a role in drive. As complement to the transmission bias I have measured, I am currently, applying cytological methods to visualize differences in the centromeres between chromosome forms and the arrangement of those chromosomes in female meiosis.

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Cytogenetic characterization of repetitive elements in Asian swamp eel (*Monopterus albus*)

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Contrary to chromosome number (2n = 42–48) and size of teleost fish, Asian swamp eel (*Monopterus albus*) shows a few number of chromosome with large chromosome size. In this study, to investigate chromosome constitution of swamp eel, karyotyping were performed using Giemsa staining, C-banding, PI- and DAPI-staining, and fluorescence in situ hybridization (FISH) with repetitive elements (18S–28S rRNA genes, fosmid clone comprising undefined repetitive sequences, Rex1 retrotransposon, and telomeric repeats). Diploid chromosome numbers were 24 chromosomes, all of them were acrocentric chromosomes. Intense DAPI stains were observed in centromeric regions of all chromosomes. Surprisingly, C-positive heterochromatin blocks and intense PI-stains were found in centromeric regions of one acrocentric chromosome pair. However, at the same chromosome pair, the presence of C-positive and PI-positive bands were also observed at the telomeric end of one acrocentric chromosome as heteromorphism. This suggests that at the telomeric end, the heteromorphic C-positive/PI positive band have been arisen by the accumulation of repetitive sequences with GC-rich. Hybridization signals of 18S–28S rRNA genes were localized to the centromeric region at the same position with PI positive band. Chromosomal localization of the fosmid clone which contained undefined repetitive sequences was located in the telomeric regions of two acrocentric chromosome pairs, and telomeric repeats were found at telomeric ends of all chromosomes and interstitial site of one acrocentric pair. Rex1 retroelements were preferentially distributed in centromeric regions of all acrocentric chromosomes. Rex3 retroelements were localized to entirely whole chromosome arm of 11 acrocentric pairs and in the pericentromeric region of 1 acrocentric pair. This suggests that the presence of Rex1 and Rex3 retroelements compartmentalization have occurred in Asian swamp eel. The present results provide prerequisite data of repetitive elements in Asian swamp eel, and lead us to understand chromosome structure of swamp eel.

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Interactions of inner kinetochore proteins with fission yeast regional centromeres

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Centromeres of the fission yeast *Schizosaccharomyces pombe* lack the highly repetitive sequences that make most other 'regional' centromeres refractory to analysis. To map fission yeast centromeres, we applied H4S47C-anchored cleavage mapping, and native and cross-linked chromatin immunoprecipitation with paired-end sequencing. H3 nucleosomes are nearly absent from the central domain, which is occupied by centromere-specific H3 (cenH3 or CENP-A) nucleosomes with two H4s per particle that are mostly unpositioned and are more widely spaced than nucleosomes elsewhere. Inner kinetochore proteins CENP-A, CENP-C, CENP-T, CENP-I and Scm3 are highly enriched throughout the central domain except at *tRNA* genes, with no evidence for preferred kinetochore assembly sites. These proteins are weakly enriched and less stably incorporated in H3-rich heterochromatin. CENP-A nucleosomes protect less DNA from nuclease digestion than H3 nucleosomes, while CENP-T protects a range of fragment sizes. Our results suggest that CENP-T particles occupy linkers between CENP-A nucleosomes, and may account for the 'chromatin smear' seen in fission yeast upon electrophoresis of centromeric chromatin treated with micrococcal nuclease. In contrast to the precisely positioned single-wrap CENP-A particles on young human alpha satellite or centromeric nucleosomes of budding yeast (*Saccharomyces cerevisiae*), which contain a single H4 molecule, our results indicate that classical regional centromeres of fission yeast differ from other centromeres by the absence of CENP-A nucleosome positioning.

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A genomic comparison of SINE composition in the tiger and domestic cat

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With the recent exponential increase in available genomic data, non-coding DNA regions including short interspersed nuclear elements (SINEs) have emerged as significant contributors to eukaryotic genetic and phenotypic diversity, especially in the mammalian genome. Previously, using comparative genomics methods, we characterized the Carnivore specific SINE family (Can-SINEs) in the Feliform (cat-like) suborder, inluding two subfamilies specific to the Felidae family. This work revealed multiple Can-SINE expansions, as well as historical introgression and rapid speciation events. Now, we attempt to determine the proliferation model for the four Feliform-specific SINE subfamilies in the domestic cat and tiger using the recently completed *Panthera trigris* whole genome sequence. These four subfamilies are classified as the ancient and extinct IA and IB subfamilies and currently active IIA and IIB subfamilies. We find that while each of the two Felid-specific SINE subfamilies, IIA and IIB, are active in the both the domestic cat and tiger, the level of activity for each subfamiy differs. In addition, we find that the germ-line Can-SINE instances support the more recently proposed 'transposon model'[1] of proliferation in which a significant portion of daughter elements that also have replicative activity over the more commonly accepted 'master copy' hypothesis of SINE proliferation in which only one or a few SINE elements produce numerous non-replicative daughter copies.

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Linkage mapping with paralogs exposes regions of residual tetrasomic inheritance in chum salmon (*Oncorhynchus keta*)

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The common ancestor of salmonids had a whole genome duplication approximately 100 million years ago. Gene sequence similarity due to shared ancestry complicates the assessment of genetic variation, as paralogs can be difficult to distinguish from each other. In some salmonids, including chum salmon (*Oncorhynchus keta*), assessment of genetic variation is further complicated by incomplete rediploidization subsequent to the whole genome duplication and ongoing residual tetrasomic inheritance in parts of the genome. Sequences derived from paralogs are often confounded and removed prior to further analyses, leaving the underlying loci uncharacterized. This exclusion can lead to bias in subsequent analyses if evolutionary forces, such as selection and genetic drift, are different for paralogous loci. We present a novel method to resolve confounded paralogs by observing allelic segregation patterns in gynogenetic haploid offspring and demonstrate its effectiveness by constructing two linkage maps for chum salmon; with and without these newly resolved loci. We find that the resolved paralogous loci are not randomly distributed across the genome. A majority are clustered in expanded subtelomeric regions of 14 linkage groups, suggesting a significant fraction of the chum salmon genome may be missed in analyses excluding paralogs. Transposable elements have been proposed as drivers of genome evolution and, in salmonids, may have a role in the rediploidization process by driving differentiation between homeologous chromosomes. Consistent with that hypothesis, we find a reduced fraction of transposable element annotations among paralogous loci, which predominantly occur in the genomic regions that lag in the rediploidization process.

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Neo-sex chromosome inheritance in *Silene* hybrids

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Neo-sex chromosomes form through the major restructuring of ancestral sex chromosome systems. This restructuring often consists of the fusion of an autosome to an existing sex chromosome, resulting in new sex-chromosome formations (e.g. X1-X2-Y or X-Y1-Y2). If neo-sex chromosomes are improperly inherited, abnormal development and reproductive isolation may occur. In this study, we examined the consequences of dissimilar sex chromosome systems to interspecies hybrids from the flowering plant genus *Silene*. In *Silene diclinis*, sex-chromosome resturcturing is thought to have involved a reciprocal translocation between the Y chromosome and an autosome, yielding *S. diclinis* males with two Y chromosomes (Y1-Y2) and females with a neo-X in addition to the ancestral X. This neo-sex chromosome system is thought to have evolved from an ancestral XY system that is still present in *Silene latifolia*. These two species do not hybridize naturally, and improper inheritance of different sex-chromosome systems could contribute to reproductive isolation. We used sex chromosome-linked, species-specific, polymorphic markers and chromosome squashes to determine whether the major restructuring of sex chromosomes prevented proper inheritance in a variety of *S. diclinis* and *S. latifolia* hybrid crosses created in the greenhouse, including some F2- and later-generation hybrids as well as hybrids with aberrant sex expression (e.g., neuter, hermaphrodite). We found that, for individuals that survived to flowering, proper segregation had occurred in hybrids despite the differences in sex chromosome structure between the two species. Since the sex-chromosomes segregated properly in later-generation hybrids and hybrids with aberrant sex expression, this indicates that incompatibility of sex-chromosome structure alone does not result in complete reproductice isolation between *S. diclinis* and *S. latifolia*. Rather, issues with sexual development in these hybrids are likely caused by intrinsic genetic incompatability rather than improper sex chromosome inheritance.

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Sequencing the threespine stickleback Y chromosome: a vertebrate model system for sex chromosome evolution

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Suppression of recombination between young sex chromosomes leads to rapid sequence divergence between heteromorphic pairs. Although recent progress has been made in characterizing sequence divergence of genes scattered across sex chromosomes in plants vertebrates and invertebrates these studies typically use the X chromosome as a reference. This makes it impossible to study novel regions and structural rearrangements on the Y chromosome. Furthermore the early stages of Y chromosome structural evolution are not well understood as complete Y chromosome assemblies have only been generated for a handful of mammalian X/Y systems. The threespine stickleback fish (*Gasterosteus aculeatus*) is a powerful model system in which to explore early vertebrate sex chromosome evolution as it possesses a relatively young X/Y sex chromosome pair that is less than 16 million years old. In order to generate a complete sequence assembly of the threespine stickleback Y chromosome we have identified 108 Y-chromosome specific clones from a bacterial artificial chromosome (BAC) library. We Sanger sequenced the BAC clones and generated 12.1 Mb of contigs that cover the estimated 17.5 Mb non-recombining region of the Y chromosome. We used optical mapping to bridge gaps and assemble the contigs into scaffolds. Even at this young stage of evolution the threespine stickleback Y chromosome harbors two distinct evolutionary strata that correspond to known structural rearrangements as well as large arrays of repetitive sequence. Genes on the Y chromosome are at various levels of degeneration: many genes have experienced degeneration in coding regions and/or expression levels and some genes have either been completely deleted from the Y or have been silenced. Our ongoing sequencing efforts will result in the first complete sequence of a young vertebrate Y chromosome providing important insights to the early stages of sex chromosome evolution.

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CenH3 evolution reflects meiotic symmetry and monocentric/holokinetic chromosomal structure as predicted by the centromere drive model

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The centromere drive model explaining rapid evolution of eukaryotic centromeres predicts differential evolution of centromeric histone H3 (CenH3) with regard to the type of meiosis and chromosomal structure. However, despite the impression one might get from the literature, this key prediction of the centromere drive model has not only never been confirmed, but it has never been tested. To provide evidence for or against such evolution of CenH3, we have inferred the selective pressures acting on CenH3 in sixteen eukaryotic clades, including plants, animals, fungi and ciliates, using codon-substitution models. We have found that CenH3 has been evolving adaptively much more frequently in clades with asymmetric meiosis and monocentric chromosomes compared with clades displaying only symmetric meiosis or holokinetic chromosomes which provides actual evidence confirming the prediction of centromere drive model. Our findings indicate that the evolution of asymmetric meiosis required CenH3 to evolve adaptively more often to counterbalance the negative consequences of centromere drive. Nevertheless, with the appearance of holokinetic chromosomes, CenH3 was no longer forced to adapt to centromere drive, suggesting that holokinetism itself is enough to suppress it.

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