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Guest Editorial

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Chromosome Evolution: Molecular Mechanisms and Evolutionary Consequences

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Long before the advent of "next-generation" or even "first-generation" sequencing, biologists could observe the genome of an organism using relatively simple cytogenetic methods. As a result, we now have information about the number and structure of chromosomes in thousands of organisms. Even at this relatively crude level of observation, it is clear that there is extensive variation in chromosome number and structure among species, and that there can even be dramatic differences in karyotype between closely related species. Cytogenetic studies have also revealed that males and females of many species differ in karyotype with visible sex chromosomes. Understanding both the causes and consequences of such changes in chromosome number and structure has been of long-standing interest to evolutionary biologists (White 1973; King 1993).

Evolutionary geneticists like Theodosius Dobzhansky focused on chromosomal inversions, not only as polymorphisms that varied within species, but also as potential drivers of divergence and speciation (Dobzhansky 1970). However, the development of molecular biology shifted the focus of many evolutionary biologists to sequence-based variation, and interest in the role of chromosomal change in evolutionary processes fell mostly out of vogue.

At the same time as interest in chromosomes was waning in evolutionary biology, the molecular biology revolution enabled scientists to investigate the molecular mechanisms that underlie chromosome structure and function. Thus, a large body of work over the last few decades has led to a detailed molecular understanding of chromosomes and their component parts such as chromatin, centromeres, and telomeres, as well as how chromosomes recombine with each other and are transmitted through meiosis.

This molecular understanding of chromosome structure is now becoming more relevant for evolutionary biologists, as advances in genome sequencing have revealed additional variation in chromosome structure that cannot be observed by cytogenetic methods. These findings have reminded evolutionary biologists that variation in chromosome structure is widespread and might have evolutionary consequences. Thus, there has been a resurgence of interest in classic questions: How do chromosomal differences evolve between populations or species? What evolutionary forces drive differences in chromosome structure among populations and species, or between sexes? What are the evolutionary consequences of differences in chromosome structure? And now, we can add new questions about the molecular mechanisms that underlie these differences in chromosome structure and number. Addressing these questions truly requires an interdisciplinary approach that cuts across traditional divides in biology.

Although I would now consider myself an evolutionary geneticist, my training was in molecular and developmental genetics. For most of my career, I have been surrounded by colleagues working on the detailed molecular and genetic mechanisms that underlie different aspects of chromosome structure. My own research spans these fields, and I am interested in both the molecular mechanisms that underlie variation in chromosome structure between sexes, populations, and species as well as the evolutionary consequences of this variation for adaptation and speciation. Thus, I have long dreamed of bringing together people working on both molecular and evolutionary aspects of chromosome evolution in order to stimulate exchange of ideas between scientists with different perspectives. Serving as the American Genetic Association (AGA) President in 2015 provided me with the perfect opportunity to do.

The 2015 AGA Presidential Symposium, entitled "Chromosome Evolution: Molecular Mechanisms and Evolutionary Consequences," was held August 17–20, 2015 at Islandwood on Bainbridge Island near Seattle, Washington, USA. This is an idyllic location for a small and intimate meeting that encourages interaction and discussion among participants. In organizing the symposium, I invited diverse speakers working on a broad range of model systems (multiple species of plants, invertebrates, and vertebrates) and using a variety of approaches (cytogenetics, genomics, genetics, phylogenetics, molecular biology, computation, theory) to investigate the evolution of chromosome rearrangements, chromatin, centromeres, meiosis, recombination, and sex chromosomes. This breadth of research interests extended to the nearly 100 participants who came from 11 countries and 5 continents. In particular, there was an excellent group of student participants, with 18 students from 4 countries generously supported by funds from the AGA. What united all of these

speakers and participants was a willingness to discuss and share ideas outside of their traditional fields.

This special issue of the *Journal of Heredity* captures the breadth of research presented and discussed at the symposium. In his AGA Key Distinguished Lecture and accompanying article (this issue), Mark Kirkpatrick provides a broad overview of both recent theory and data on the evolutionary forces that drive chromosome and genome evolution. He takes us on a whirlwind journey though the field, revisiting classic questions on the maintenance of chromosome inversion polymorphisms in populations, and presenting new data on the effect on inversions on adaptation, the evolutionary forces that drive the turnover and formation of sex chromosomes, and the evolutionary consequences of sex chromosome evolution for sex ratio. This review provides the perfect backdrop to the nine additional articles in this special issue.

As with Dobzhansky's focus on inversion polymorphisms in *Drosophila*, there is still much to be gained by in-depth and long-term studies of particular model systems. Fredy Colorado-Garzón (AGA student travel awardee) and colleagues (this issue) have focused on chromosome diversity within a group of black flies in Colombia. Their study highlights how classical karyotype information is still of fundamental importance in taxonomy and can be used to demonstrate the existence of cryptic species. Jeremy Searle and colleagues (Giménez et al., this issue) have reviewed a half-century of their work on house mice from a small (70 km) region in the Alps, which harbors house mice with a dazzling array of karyotypic diversity. Differentiation between these chromosomal races is mostly due to Robertsonian fusions between acrocentric chromosomes. Their collective studies on this patchwork of chromosomal races and hybrids have provided new insights to the evolutionary consequences of chromosome rearrangements, particularly in hybrid zones. Interestingly, fixation of Robertsonian fusions is suggested to result from meiotic drive. In a beautiful example of collaboration between evolutionary and molecular biologists, Michael Lampson (one of the speakers at the meeting) used some of these same mouse populations to demonstrate that differences in centromere size between fused and unfused chromosomes underlies the process of meiotic drive (Chmátal et al. 2014).

Although classical cytogenetic analyses remain a key tool, computational and genomic analyses are new tools that have provided unprecedented insight to the molecular changes occurring on chromosomes during evolution. For example, whole genome sequencing across *Drosophila* species revealed that species with a small population size do not have a higher load of transposable elements in the genome, which is contrary to the trends observed across a large number of species. To explain this finding, Sam Groth and Justin Blumenstiel (this issue) use simulations to show that increases in horizontal gene transfer and recombination rate might actually lead to higher levels of transposable elements in larger populations. The evolutionary dynamics of transposable and repetitive elements are important to more than than genome size, as they are commonly found in specific regions of the chromosome, such as centromeres and telomeres, and also accumulate in specific regions of the genome, such as B chromosomes and sex chromosomes. By tracking centromeric repetitive elements using sophisticated computational methods, as well as genomic sequences for modern and archaic humans, Karen Miga (this issue) demonstrates that the well-known fusion of two ancestral primate chromosomes that created human chromosome 2 occurred before the divergence between modern and archaic humans. Similarly, Frances Clark, Matthew Conte (AGA student travel awardee) and colleagues (this issue) identified repetitive elements in whole genome sequencing data that were consistent with the presence of B chromosomes in cichlid fish from Lake Malawi. They further used molecular and cytogenetic tools to track these B chromosomes across species and generations, which allowed them to make predictions about the mitotic and meiotic mechanisms that might underlie drive of B chromosomes in these species.

The special issue concludes with a set of papers on sex chromosomes, which are a unique and dynamic region of the genome. Kohta Yoshida, Takashi Makino, and Jun Kitano (this issue) demonstrate that deleterious mutations are already accumulating on one of the youngest known vertebrate sex chromosomes, which is found in the Japan Sea stickleback fish. This experimental article is followed by three comprehensive reviews on the evolution of sex chromosomes across three different groups. Alex Harkess (AGA grant awardee) and Jim Leebens-Mack (this issue) take us on a tour of the diverse mechanisms of sex determination across flowering plants, with a nice historical perspective as well as an overview of the range of exciting current research in the field. Heath Blackmon, Laura Ross, and Doris Bachtrog (this issue) perform a tour-de-force analyses of the diverse sex determination mechanisms present in insects. Finally, Tariq Ezaz, Kornsorn Srikulnath, and Jennifer Graves (this issue) provide a provocative view of the origin of sex chromosomes in amniotes (reptiles and mammals) that leaves us with many new questions to answer in future research.

I hope that the pioneers of the study of chromosome evolution would be as invigorated as I am by the remarkable new insights that have been gained using these diverse approaches and systems. However, the articles in this special issue also remind us that many classic questions remain unanswered and many new questions have emerged. What is clear is that understanding the evolutionary forces and molecular mechanisms that drive changes in chromosome number and structure across species will continue to inspire a new generation of diverse biologists.

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References

Chmátal L, Gabriel SI, Mitsainas GP, Martínez-Vargas J, Ventura J, Searle JB, Schultz RM, Lampson MA. 2014. Centromere strength provides the cell biological basis for meiotic drive and karyotype evolution in mice. *Curr Biol*. 24:2295–2300.

Dobzhansky TG. 1970. *Genetics of the evolutionary process*. New York: Columbia University Press.

King M. 1993. *Species evolution: the role of chromosome change*. Cambridge (UK): Cambridge University Press.

White MJD. 1973. *Animal cytology and evolution*. 3rd ed. Cambridge (UK): Cambridge University Press.