The differences between cis- and trans-gene inactivation caused by heterochromatin in Drosophila, pp. 93–106
Yury A. Abramov, Aleksei S. Shatkshik, Oksana G. Maksimenko, Silvia Bonaocorri, Vladimir A. Gvozdev, and Sergey A. Lavrov

The heterochromatin position-effect—in which a gene’s expression is disrupted by insertion into heterochromatin—remains enigmatic. The authors characterized position-effect variegation in a new Drosophila system using a high throughput approach. They found opposing effects on genes expression that varied at different developmental stages. Cis-repression near the new eu-heterochromatin border was independent of trans-inactivation of reporter genes on the homologous, non-rearranged chromosome, with distinct molecular features.

Spindle assembly and chromosome segregation requires central spindle proteins in Drosophila oocytes, pp. 61–75
Arunika Das, Shital J. Shah, Bensen Fan, Daniel Pak, Daniel J. DiSanto, Anna Maria Himman, Jeffry M. Cesario, Rachel A. Battaglia, Nicole Demos, and Kim S. McKin

Oocytes segregate chromosomes in the absence of centrosomes, and the central spindle plays a conserved and critical role in this acentrosomal process. Das et al. investigate central spindle protein function in acentrosomal spindle assembly and chromosome segregation, identifying genes that interact with the Kinesin 6 Subito and the Chromosome passenger complex component Incenp. They also identify cytokinesis proteins and downstream effectors that are required for homologous chromosome bi-orientation at metaphase I. These results suggest a novel meiotic function for proteins normally involved in mitotic anaphase and cytokinesis.

Deep history of East Asian populations revealed through genetic analysis of the Ainu, pp. 261–272
Chongwon Jeong, Shigeki Nakagome, and Anna Di Rienzo

The Ainu descend from prehistoric Japanese hunter-gatherers. Jeong et al. use genome-wide variation data to show that they represent one of the deepest branches of human diversity in East Asia, basal to all present-day East Asian farmers. Interestingly, they do not detect signatures of a connection between the Ainu and Tibetans, rejecting a long-held hypothesis based on Y chromosome data. Unlike other East Asians, the Ainu are more closely related to northeast than central Siberians, suggesting a potential connection between the Ainu ancestors and Beringian populations, whose descendants became the first people of the Americas.

Genetical genomics of behavior: a novel chicken genomic model for anxiety behavior, pp. 327–340
Martin Johnsson, Michael J. Williams, Per Jensen, and Dominic Wright

The identification of genes responsible for behavioral variation has ramifications ranging from medical research to evolutionary theory on personality syndromes. To identify the genes underlying anxiety behaviors the authors used a combination of conventional and expression QTL analysis, and an advanced intercross between wild and domestic chickens. This approach yielded 10 potentially causal genes for anxiety that were also correlated with mouse anxiety behavior and human psychiatric disorders, indicating the potentially translatable nature of genes underlying anxiety.

Genome-wide structural variation detection by genome mapping on nanochannel arrays, pp. 351–362

Comprehensive whole genome structural variation detection is challenging using current approaches. Mak et al. show that genome mapping with long, fluorescently labeled DNA molecules imaged on nanochannel arrays can be used for locally phased whole genome structural variation detection without sequencing. The authors generated genome maps from a trio from the 1000 Genomes Project and identified structural variations >5 kb in size. The individuals have many more structural variants than previously published, including some with the potential to disrupt gene function.

Identifying regulators of morphogenesis common to vertebrate neural tube closure and Caenorhabditis elegans gastrulation, pp. 123–139

Neural tube defects (NTDs) are common and severe birth defects, yet the functions of most NTD genes are unknown. Sullivan-Brown et al. used Caenorhabditis elegans to study NTD gene homologs, finding spf-3 and members of the WAVE complex were required for Caenorhabditis elegans gastrulation, a process that shares characteristics with vertebrate neural tube closure. Nckap1, a WAVE complex member, was also required for neural tube closure in Xenopus embryos. This work provides a new platform for studying the in vivo roles of NTD genes.

Rapid short-read sequencing and aneuploidy detection using MinION Nanopore technology, pp. 37–44
Shan Wei and Zev Williams

MinION is a portable nanopore-based sequencer primarily designed for single-molecule sequencing of long DNA fragments (>6 kb). Wei and Williams describe a library preparation and data analysis method to enable rapid sequencing of short fragments (<1 kb) and demonstrated its clinical applicability by performing aneuploidy detection in prenatal and miscarriage samples, with sequencing in less than 4 hours. This broadens the application of MinION technology, making it a promising tool for applications that involve rapid short-read sequencing.

This Month in the American Journal of Human Genetics

Mathieu Deschamps, Guillaume Laval, Maud Pagny, Yuval Ivan, Laurent Abel, Jean-Laurent Casanova, Etienne Pato, and Luis Quintana-Murci

Michael Dannemann, Aida M. Andres, and Janet Kelso

Throughout history, human evolution has been shaped by interactions with pathogens. Alleles conferring fitness most often arise from random mutation, but introgression of genomic material from archaic humans can provide a separate reservoir of protective variation. Now, using complementary approaches, Deschamps et al. and Dannemann et al. uncover signatures of selection and evidence of introgression from the Neanderthal and Denisovan genomes in human innate immunity genes. These findings suggest that admixture with archaic humans played a key role in shaping the modern human immune system. Of course, pathogens evolve quickly, and so it will be interesting to determine the modern-day function of these introgressed alleles.