Group Details Structure of Centromeric Transition Regions of Human Genome

A group of more than dozen researchers, including three at the UW, has completed a six-year project examining the structure and evolution of centromeric transition regions on the human genome. Their results appear in the Aug. 19 issue of the journal Nature.

The study provides a detailed structure of the human genome near the centromere, one of the most difficult areas to study. The centromere is the junction of the pair of chromatids that make up a chromosome. It ensures the proper segregation of chromosomes during cell division.

The research group was led by Evan Eichler, associate professor of genome sciences at the UW, and his colleagues. Much of the research was done at Case Western Reserve University School of Medicine, where Eichler was on the faculty prior to joining the UW this year. The group also included researchers from the University of California, Santa Cruz; Washington University in St. Louis; Simon Fraser University, in British Columbia; University of Bari, in Italy; and Pennsylvania State University.

Eichler and his colleagues discovered that centromeric transition regions are made up of gene sequences copied from the area between the euchromatin and heterochromatin. They found that genetic turnover has moved these sequences into the centromeric transition regions, suggesting the genome is very dynamic over time.

The process has led to the formation of at least 28 new genetic transcripts, the researchers found, with most of those expressed in the testis. That suggests the centromeric transition region may be an incubator for new genes.