

Statgen Seminar, Autumn 2011

Large-scale chromosomal anomalies

The GENEVA group has submitted a manuscript (Laurie et al.) to Nature Genetics on large-scale chromosomal anomalies. A parallel submission has been made by the National Cancer Institute (Jacobs et al.) We will finish the quarter by reading these papers, but first we need to read some background.

October 11: Caitlin McHugh, Lisa Brown. (Bruce Weir)

T. Hassold and P. Hunt. 2009. To err (meiotically) is human: the genesis of human aneuploidy. *Nature Reviews Genetics* 2:280-291.

T. Hassold et al. 2007. The origin of human aneuploidy: where we have been, where we are going. 2007. *Human Molecular Genetics* 16:R203-R208.

October 18:

No Seminar. Attend Genome Sciences Symposium.

October 25: Steven Smith, Jan Irvahn. (Tim Thornton)

D.A. Peiffer et al. 2006. High-resolution profiling of chromosomal aberrations using Infinium whole-genome genotyping. *Genome Research* 16:1136-1148.

(background: N.P. Carter. 2007. Methods and strategies for analyzing copy number variation using DNA microarrays. *Nature Genetics* 39:S16-S21.)

November 1: Serge Svedlov, Charles Cheung. (Bruce Weir)

K. Wang et al. 2007. PennCNV: An integrated hidden markov model for high-resolution copy number variation detection in whole-genome SNP genotyping data. *Genome Research* 17:1665-1674.

(background: S. Colella et al. 2007. QuantiSNP: an objective Bayes Hidden-Markov Model ... *Nucleic Acids Research* 35:2015-2025.)

November 8: Kevin Rubenstein, Jerry Kim. (Bruce Weir)

L.K. Conlin et al. 2010. Mechanisms of mosaicism, chimerism and uniparental disomies identified by single nucleotide polymorphism array analysis. *Human Molecular Genetics* 19:1263-1275.

(possible extra: W.P. Robinson. 2000. Mechanisms leading to uniparental disomy and their clinical consequences. *BioEssays* 22:452-459.)

November 15: Matthew Conomos. (Sharon Browning)

B. Rodriguez-Santiago et al. 2010. Mosaic uniparental disomies and aneuploidies as large structural variants of the human genome. *Am J Human Genetics* 87:129-128.

(possible extra: A. Itsara et al. 2009. Population analysis of large copy-number variants and hotspots of human genetic disease. *Am J Human Genetics* 84:148-161.)

November 22: Xiuwen Zheng, Chaozhi Zheng. (Elizabeth Thompson)

A.B. Olshen et al. 2004. Circular binary segmentation for the analysis of array-based DNA copy number data. *Biostatistics* 5:557-572.

(possible extra: Venkatraman and Olshen. 2007. A faster circular binary segmentation for the analysis of array CGH data. *Bioinformatics* 23:657-663.)

November 29: Rui Zhang. (Bruce Weir)

Jacobs et al. 2011. The aging genome: genetic mosaicism and its relationship to cancer. (submitted)

December 6: Jean Morrison. (Ellen Wijsman)

Laurie et al. 2011. Somatic mosaicism for large chromosomal anomalies from birth to old age and its relationship to cancer. (submitted)