

Mathematical & Computational Biology Seminar

Organizer: Valerie Hower

Thursday, 3:00–4:00pm, 740 Evans

July 16 **Itsik Pe'er**, Columbia University

Genetic segments shared within and between human populations

The availability of cost-effective, high throughput technologies to genotype common alleles has yielded an unprecedented wealth of genome-wide data on human variation, deeply sampled within and across populations. We have developed a rapid method that facilitates extensive evaluation of shared genetic segments across millions of sample-pairs. We report analysis of such sharing to improve understanding of recent genetic history of samples, both genome-wide as well as for specific loci. We show extensive hidden relatedness between individuals that share segments Identical By Descent (IBD) within populations. Such sharing provides estimates of demographic parameters. Specifically, for Ashkenazi Jewish populations we demonstrate a severe bottleneck 20-25 generations before present. We show genetic sharing to be focused at regions that suggest a causal mechanism for ancient sharing rather than recent relatedness, such as the HLA and the commonly polymorphic inversion of 5Mbp on chromosome 8p23.1. Finally, we show how genetic sharing can be used for analysis of genome-wide association studies.