

Mathematical & Computational Biology Seminar

Organizer: Valerie Hower

Wednesday, 2:00–3:00pm, 939 Evans

Oct. 28 **Ben Raphael**, Brown University

Structural variation in human and cancer genomes

Genome variation occurs on a continuum of scales ranging from single nucleotide differences to large structural rearrangements. In the past few years, structural variants including deletions, insertions, inversions, and translocations of large blocks of DNA sequence have been shown to be a significant fraction of the genetic differences in human populations. Structural variants also arise as somatic mutations during the lifetime of an individual and are frequently found in cancer genomes. I will describe computational approaches to address several problems arising in structural variation studies of human and cancer genomes. These include: (i) a framework for classifying and comparing structural variants measured using a variety of DNA sequencing technologies; (ii) combinatorial algorithms to find the most parsimonious sequence of rearrangements and duplications that transform one genomic sequence into another; (iii) a technique to identify groups of interacting genes, or pathways, that are mutated at significant frequency in specific cancer types. I will illustrate applications of these approaches to data from two ongoing projects: The Cancer Genome Atlas and the 1000 Genomes Project.