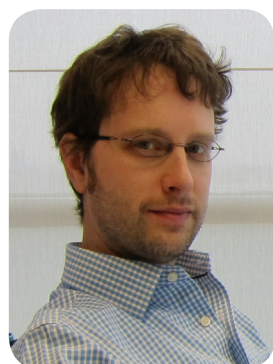




The **McLaughlin Centre** at The University of Toronto is a proud partner of **CIHR STAGE** and jointly presents:



## Dr. Benjamin Voight

Assistant Professor  
Department of Systems Pharmacology  
and Translational Therapeutics  
Department of Genetics  
**Perelman School of Medicine**  
**University of Pennsylvania**

### Genome-wide mutation rates viewed through a seven-nucleotide window

**Friday**  
**March 4, 2016**  
**12:00 – 1:00 pm**

The Hospital for Sick Children  
Daniels Hollywood Theatre  
Room 1246, 1st Floor, Black Wing  
555 University Avenue, Toronto, ON

**Abstract:** The rate of single-nucleotide polymorphism varies substantially across the human genome and fundamentally influences evolution and incidence of genetic disease. Previous studies have only considered the immediately flanking nucleotides around a polymorphic site—the site's trinucleotide sequence context—to study polymorphism levels across the genome. Moreover, the impact of larger sequence contexts has not been fully clarified, even though context substantially influences rates of polymorphism. I will describe a new statistical framework we apply to data from the 1000 Genomes Project to demonstrate that a heptanucleotide (seven nucleotide) context window explains a large fraction of variability in substitution probabilities in the human genome along. We identify new, mutation promoting sequence motifs as well as additional rate variation at sequence contexts known to be highly mutable (CpG sites). I will describe application of our model and computational tools we are developing to interpret human disease genetic studies of de novo mutational burden.

**CIHR STAGE** is a training program in genetic epidemiology and statistical genetics, housed at the University of Toronto Dalla Lana School of Public Health, and funded by the Canadian Institutes of Health Research through the Strategic Training Initiative in Health Research Program. Seminars are sponsored by The Hospital for Sick Children; the Lunenfeld-Tanenbaum Research Institute of Mount Sinai Hospital; the Ontario Institute for Cancer Research; the Department of Statistics of the University of Toronto; the Ontario Cancer Institute of the University Health Network; and the CIHR Institute of Genetics.

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