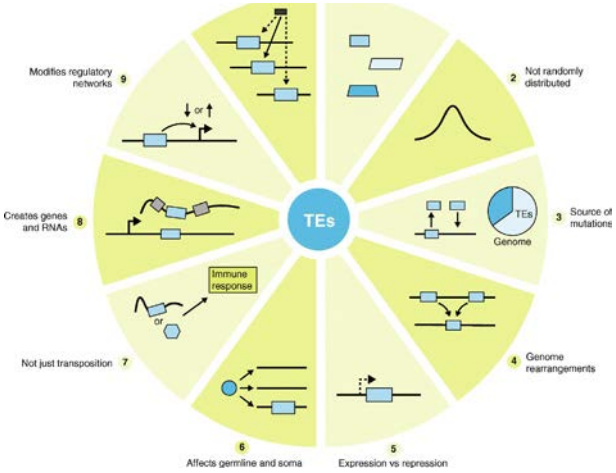
GUEST SPEAKER SEMINAR

Molecular Genetics
UNIVERSITY OF TORONTO

Unmasking transposable elements



A substantial proportion of the genome of many species is derived from transposable elements (TEs). TEs have contributed numerous regulatory, transcript and protein innovations and have also been linked to disease. We will describe two stories that explore the role and impact of TEs. In the first story, copy number variants (CNVs) are known to affect a large portion of the human genome and have been implicated in many diseases. Although whole-genome sequencing (WGS) can help identify CNVs, most analytical methods suffer from limited sensitivity and specificity, especially in regions of low mappability. To address this, we developed PopSV, a CNV caller that relies on multiple samples to control for technical variation. We demonstrate that our calls are stable across different types of repeat-rich regions and validate the accuracy of our predictions using orthogonal approaches. Applying PopSV to 640 human genomes, we find that low-mappability regions are approximately 5 times more likely to harbor germline CNVs, in stark contrast to the nearly uniform distribution observed for somatic CNVs in 95 cancer genomes. In the second story, we will look at the impact of TEs on gene regulatory networks. We will show that TEs have been a major contributor to open chromatin regions in the human genome, especially in primate-specific regions. We will present data from different from non-human primate species and show how they can be used to identify putatively functional TE-derived sequences.

Dr. Guillaume Bourque
McGill University

Host: Dr. Quaid Morris

Date: Monday December 10, 2018
Time: 4PM
Place: MSB 2172