UNIVERSITÉ LAVAL

INSTITUT DE BIOLOGIE INTÉGRATIVE ET DES SYSTÈMES IBIS

C O N F É R E N C E

Guillaume Bourque, Ph.D.

Associate Professor, Department of Human Genetics, McGill University
Director of Bioinformatics, McGill University & Genome Quebec Innovation Center (MUGQIC)
Montréal

Unmasking transposable elements

LE JEUDI 21 FÉVRIER 2019 À 12 H 30

Pavillon Charles-Eugène-Marchand, salle Hydro-Québec (1210)

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.

Lunch et breuvages seront offerts.
SVP confirmer votre présence sur : https://doodle.com/poll/r392ew3w83nvci27
avant le mercredi 20 février, 10 h

Hôtes : Christian Landry et Mathieu Hénault

Responsable : Dr Robert M. TANGUAY
robert.tanguay@ibis.ulaval.ca