Centre for
Heart Lung Innovation
UBC and St. Paul's Hospital

Centre for Heart Lung Innovation Seminar Series

Understanding human gene regulation through interpretable machine learning

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Friday May 5th 11:30 – 12:30 PM
James Hogg Conference Centre (JHCC) Room 103
(ZOOM Meeting ID: 662 2255 0438; passcode: 623137)

Hosted by Andrew Sandford

"Genome-wide association studies (GWAS) have identified tens of thousands of genetic variants associated with human diseases and traits, but the vast majority of these associations are not backed by a hypothesized mechanism. Understanding such disease-associated variants is hampered by the incompleteness of annotations functional elements in the genome. To improve our annotation of genomic elements, large-scale projects like ENCODE, CEEHRC and IHEC have recently engaged in epigenome mapping. These projects are enabled by high-throughput sequencing techniques for genome-scale measurement of biochemical activity of chromatin in cellular samples. These datasets quantify various facets of gene regulation such as genome-wide measurements of transcription factor binding or histone modifications using ChIP-seq, measurements of open chromatin using DNase-seq or ATAC-seq, RNA transcription using RNA-seq, and others. My group aims to improve our understanding and annotating of the genome using machine learning methods. I will present our recent work performing comprehensive annotation of chromatin states in human cells using segmentation and genome annotation (SAGA), integrating measures of 3D genome conformation from Hi-C, and ensuring that these approaches are robust and reliable."

This event is a Self-Approved Group Learning Activity as defined by the Maintenance Certification Program of the Royal College of Physicians and Surgeons of Canada