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SPEAKER SERIES

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Human genome 2.0 : why a pangenome graph is better for genetic and epigenetic analyses

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Host: Shyam PRABHAKAR

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3.00pm – 4.00pm

GIS Seminar Room (Level 2)
60 Biopolis Street, Genome, Singapore 138672

About The Speaker

Dr. Guillaume Bourque is a Professor in the Department of Human Genetics at McGill University, a Canada Research Chair in Computational Genomics and Medicine and the Director of Bioinformatics at the McGill Genome Center. He leads the Canadian Center for Computational Genomics and the Epigenomics Mapping Center at McGill. He is also a Principal Investigator at the Institute for the Advanced Study of Human Biology (ASHBi) of Kyoto University. Dr. Bourque is on the Scientific Steering Committee of the International Human Epigenome Consortium (IHEC) and on the Steering Committee of the Global Alliance for Genomics and Health (GA4GH). He leads the CFI-funded SecureData4Health computational platform and a new CIHR-funded project called the Pan-Canadian Genome Library, which will allow for easier analysis and sharing of genomic data across the country. His research interests are in comparative and functional genomics with a special emphasis on applications of next-generation sequencing technologies and transposable elements.

About The Seminar

Genomic analyses often start by mapping reads to a reference genome. But, in every individual, there are DNA variants and sequences that are unique to that individual and reads coming from those regions will often be ignored. Thankfully, progress in long-read technologies and assembly can now efficiently deliver telomere-to-telomere genomes. Applying such approaches to a diverse panel of individuals combined with the development of graph-based genomic tools, the Human Pangenome Reference Consortium has just released the first human pangenome reference graph. This new resource is meant to alleviate the limitations of relying on a single linear human genome as the first step of most genetic and epigenetic analyses. In this talk, I will summarize some of the benefits of using the pangenome reference. In particular, I will show how this new reference can be used to extract missing signal when looking for genetic variants in a rare disease cohort called Genomic Answers for Kids. I will also describe the results of a new study using a genome-graph looking at epigenetic changes before and after influenza infection in monocyte-derived macrophages extracted from more than 30 individuals of different ancestry. Finally, considering the importance of data sharing in genomics, I will introduce a project called the Pan-Canadian Genome Library, which will establish the framework for Canada's management and sharing of human genomic data.