

Developments in Genome Analysis

Research and Clinical Applications

November 7, 2023

Room MS3154, Medical Sciences Building

9:00 – 9:10 Opening Remarks

9:10 – 9:40 **Talk 1: Jordan Lerner-Ellis, PhD, FACMG (Mt. Sinai Hospital Toronto)**

Canadian Genomic Databases & Novel Computing Platforms: Tools for Molecular Diagnostics & Biomedical Research

9:40 – 10:10 **Talk 2: Adam Shlien, PhD, FCCMG (The Hospital for Sick Children Toronto)**

Massive clinical utility of genomics in childhood cancer

10:10 – 10:40 Coffee Break

10:40 – 11:10 **Talk 3: Fritz Roth, PhD (University of Pittsburgh School of Medicine)**

Towards a Contextual Atlas of Variant Effects

11:10 – 11:40 **Talk 4: Tehmina Masud, MBBS PhD (Deep Genomics)**

Predicting UTR Variant Effects with Deep Learning and BigRNA, an RNA Foundation Model

11:40 – 12:35 **Keynote 1: Tomi Pastinen, MD, PhD (University of Missouri-Kansas City School of Medicine, Children's Mercy)**

First Comprehensive Genetic Test: The Impact of HiFi Sequencing on Diagnoses and Variant Discovery

12:35 – 1:30 Lunch Break

1:30 – 1:35 Session Introduction

1:35 – 2:30 **Keynote 2: Laura Arbour, MSc, MD, MSc, FRCPC, FCCMG (UBC Island Medical Program)**

The Silent Genomes Project - Building and implementing a sustainable Indigenous background variant library (IBVL) to improve diagnostic success for Indigenous patients with genetic disease

2:30 – 3:00 **Talk 5: Ashish Deshwar, MD, PhD, FRCPC (The Hospital for Sick Children Toronto)**

Bringing Transcriptomics to the Clinic: RNA sequencing as a diagnostic tool in rare disease

3:00 – 3:30 **Talk 6: Jessica Gu, Msc, MS, CGC (Medcan)**

Elective Whole Genome Sequencing in an Ostensibly Healthy Canadian Population

3:30 – 4:00 Coffee Break

4:00 – 4:45 **Panel Discussion: Opportunities and challenges of genomic medicine**

Rebekah Jobling, MD, FRCPC – molecular geneticist/clinical geneticist at SickKids

Jordan Sam, MHSc Medical Genomics – genomics data scientist at St. Michael's Hospital

Kim Amburgey, MSc – genetic counsellor, program director at Advanced Therapeutics node of PCH initiative at SickKids

Hanna Faghfoury, MD, FRCPC, FCCMG – geneticist at UHN, program director Medical Genetics training program

Ashish Deshwar, MD, PhD, FRCPC - physician scientist at the Division of Clinical and Metabolic Genetics at SickKids

Moderation: **Stacy Hewson, MSc, MSc, CCGC, CGC** - Program Director of MSc Genetic Counselling at University of Toronto and Director of Genetic Counselling at The Hospital for Sick Children

4:45 – 5:00 Concluding statements

