



CANSSI ONTARIO STATISTICS SEMINARS

LISA STRUG

Director, Data Sciences Institute
Director, CANSSI Ontario
Professor, University of Toronto
Senior Scientist, The Hospital for Sick Children

Genome Data Science: Towards understanding GWAS loci

Abstract: Genome Data Science includes the collection, manipulation, storage, visualization and extraction of information from increasingly larger, more heterogeneous and complex genomic data, in reproducible, fair and ethical ways. In the webinar I will describe our use of genome data science to identify genetic contributors to disease, using the example of the Canadian Cystic Fibrosis Gene Modifier Study. I will discuss data collection, manipulation, visualization, genome-wide association study (GWAS) analysis, and the development of novel tools to integrate heterogeneous data. These tools are necessary because GWAS identifies loci rather than the responsible gene(s), relevant tissue(s) of origin or mechanism of action for a studied trait. To understand the mechanism by which a locus contributes to disease requires functional investigation, and this needs to be hypothesis-driven, in a relevant cellular model of the contributing gene. To bridge this gap and guide the design of functional studies, we have developed data visualization and statistical integration tools implemented in our software LocusFocus, which integrates two sets of summary statistics, for example from a GWAS and gene expression analysis to pinpoint the most likely responsible gene and tissue of origin. Implementing LocusFocus at a genetic locus for Cystic Fibrosis (CF) lung disease variability identified through GWAS, I will demonstrate how it can guide our understanding of the mechanism by which the locus contributes to disease.



DATE & TIME

21 NOV 2024

11:30 AM-12:30 PM ET



HYBRID

Live on Zoom and

In Person at the University of Guelph, Room SSC1303, Summerlea Science Complex, 50 Stone Road East, Guelph, ON



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