SickKids RESEARCH INSTITUTE
Genetics & Genome Biology

Genetics & Genome Biology Program
Invited Speaker Series

The anatomy of transcription regulation

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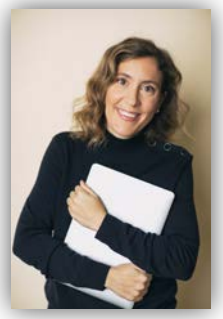

Date: Friday, May 28, 2021
Time: 12PM

I am interested in understanding how the information encoded in our genome determines gene expression variation across individuals and tissues. To address this fundamental question, we use a combination of cutting-edge computational analyses, next generation sequencing, and high-throughput functional assays.
In my talk, I will first present a project aimed at understanding how the human transcriptome changes with individual traits, such as age, ethnicity or Type II diabetes across tissues. To do this, we take advantage of the GTEx project dataset, for which we have transcriptome information from hundreds of human individuals and tissues spanning up to 15,000 samples. We apply a novel statistical framework to study how gene expression, splicing, and cell type composition simultaneously changes across tissues and individuals with different phenotypes. Second, I will present our work to characterize non-coding regions of the genome. Indeed, genome wide association studies have identified hundreds of genomic loci harboring common genetic variants associated with disease susceptibility. However, most of these loci are located in non-coding regions which compared to coding regions are much harder to characterize. Our lab combines cutting-edge computational analyses and high-throughput functional assays to systematically study non-coding regulatory elements such as enhancers and promoters. We take advantage of the recent development of massively parallel reporter assays (MPRA) to analyze tens of thousands of individual DNA oligonucleotides simultaneously in different cellular conditions. The ultimate goal is to understand how disruption of these non-coding regulatory elements by specific genetic variants mediates differences in disease susceptibility between individuals.

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Host: Philipp Maass, Phd