TCAG New Technologies Seminar

Rapid Structural Variation Detection and *De Novo* Assembly in Human and Complex Genomes Using Extremely Long Single-Molecule Imaging

Date: Thursday May 23
Time: 10:30 – 11:30 am
Location: Room 14-203, MaRS TMDT
101 College St.

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Despite continued cost reduction of raw base generation, improvement in base-calling accuracy, and recent advances in read length, complete *de novo* assembly and genome wide structural variant analysis of individual large complex genome remain expensive and challenging.

We present a rapid genome-wide analysis method based on the new NanoChannel Array technology (***Irys***™) that dynamically streams and linearizes extremely long DNA molecules for direct image analysis of tens of gigabases per run. This high throughput platform automates the imaging of genomic DNA hundreds to thousands of kilobases in length at the single-molecule level, for unambiguous assembly of complex genomes. High-resolution genome maps assembled *de novo* via unique sequence motif labeling, preserving long-range structural information that is intractable by current short read NGS platforms. This information is independent of current sequencing biochemistry and algorithms with built-in long range haplotyping and is critical to validate past and future genomic sequencing assembly data and discover new structural variants. It is simple and straightforward to set up and operate, amenable to whole genome structural variation comparative studies of large populations.

Here we present the first human *de novo* genome map assembly by the single molecule ***Irys*** system and analysis of complex regions of a variety of organisms using this approach. Unlike paired end sequencing approaches that are cumbersome and biased towards detecting more deletions than insertions, hundreds of large structural variants were uncovered with this direct view approach. We have corrected errors in previous assemblies, spanned many of the remaining gaps, identified known and novel structural variants and phased haplotype blocks—including in the highly variable regions involved with important immune system function. This technology and method will allow new discoveries and change our view toward understanding genome architecture and functions.

Hosted by The Centre for Applied Genomics and the Ontario Genomics Institute

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