

## BIONANO GENOMICS SEMINAR

You're Invited!

### **Part 1. Next-Generation Cytogenomics: High-throughput Mapping of Structural Variation in Cancer and Genetic Disease**

The diagnostic yield in genetic disease has seen very little improvement over the last few decades, despite the introduction of whole genome sequencing.

Bionano Genomics' platform for whole genome mapping offers an extremely long-read technology, providing unmatched sensitivity to detect structural variation, genome wide, at low cost. Our de novo maps can resolve complex repetitive regions, identify Copy Number Variations, and elucidate genome-wide structural variation like balanced/unbalanced translocations, inversions, and indels with much higher sensitivity and precision than sequencing-based methods.

For mosaic samples, Bionano's high coverage depth allows for the detection of any type of structural variant with more than 90% sensitivity, present in as little as 10% of the cells, genome wide, and completely unbiased.

Examples will be presented of how Bionano's platform is helping provide a molecular diagnosis for patients with a variety of genetic disorders by detecting genomic rearrangements and structural variants missed by NGS and cytogenetic methods.

### **Part 2. Bionano Genome Mapping Builds the Most Contiguous and Correct Genome Assemblies**

High throughput sequencing technologies have made the generation of genomic sequences increasingly affordable, but organizing these reads of hundreds to thousands of basepairs into entire genomes remains challenging.

The Bionano Genomics solution for whole genome optical mapping offers an extremely long-read technology, providing the ability to assemble chromosome-arm length maps used to scaffold sequence contigs and correct assembly errors. Additionally, Bionano has unmatched sensitivity to identify all major types of structural variation.

Examples will be presented of how the Bionano Saphyr platform is helping to build the most complete and accurate genome assemblies, and how it compares to other scaffolding methods like Hi-C.

### Event Info

**DATE:**

Tuesday, February 11th, 2020

**TIME:**

Part 1. 10:00 AM – 10:45 AM

Part 2. 10:45 AM – 11:30 AM

**LOCATION:**

Seminar Room 4050a/4050b

Level 4, Adelaide Health and Medical Sciences (AHM) Building

4, North Terrace, Adelaide

**SPEAKER:**

Yingying Wu | Scientific Affairs Manager | BioNano Genomics

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**THANKS TO:**

Mark Corbett | The University of Adelaide



**For more information about the Saphyr® System, please visit [www.bionanogenomics.com](http://www.bionanogenomics.com)**