

De Novo Genome Mapping with Irys: A New Technology for Facilitating NGS Assembly and Translational Structural Variation Discovery

Presented By: Rachel Pugh, BioNano Genomics

Hosted by: Institute for Genomic, Biocomputing, and Biotechnology

Thursday, February 12th

3:30-4:30pm

Institute for Genomics, Biocomputing, and Biotechnology

Pace Seed Lab

Large Conference Room (#119)

Mississippi State, MS

Abstract:

De novo genome assemblies using only short read data are generally incomplete and highly fragmented due to the intractable complexity found in most genomes. This complexity, consisting mainly of large duplications and repetitive regions, hinders sequence assembly and subsequent comparative analyses. As a result of the remaining limitations of DNA sequencing and analysis technologies, it is not economical to create multiple high quality assemblies of individuals to detect and interpret the many types of structural variation that are refractory to high throughput or short-read technologies. The result is that researchers are limited to observing SNP's and small indels as the genetic basis of phenotypic variation, while mapping studies would allow the previously impossible potential of whole genome, large-scale, structural variant analysis.

We present a single-molecule genome analysis system (Irys) based on NanoChannel Array technology that linearizes extremely long DNA molecules for direct observation. This high-throughput platform automates the imaging of single molecules of genomic DNA hundreds of kilobases in size to measure sufficient sequence uniqueness for unambiguous assembly of complex genomes. High-resolution genome maps assembled *de novo* preserve long-range structural information necessary for structural variation detection and assembly applications

