The Genome Institute at Washington University Receives Irys System from BioNano Genomics to Advance Research for Genome Reference Consortium

Irys System to be employed in research to improve the quality of the human reference genome

SAN DIEGO and ST. LOUIS — Oct. 14, 2014—BioNano Genomics announced today the purchase of an Irys™ System by The Genome Institute at Washington University in St. Louis to assist, among other projects, in improving the quality of the human reference genome for the Genome Reference Consortium (GRC). The goal of the GRC is to ensure that the human, mouse and zebrafish reference assemblies are biologically relevant by closing gaps, fixing errors and representing complex variation. The team of researchers at The Genome Institute, led by Dr. Richard Wilson, will use the Irys System in their workflow as they determine the sequence, location and orientation of genes and transposable elements in the human genome.

The Genome Institute originated to play a key role in the Human Genome Project, an international effort to decode the human genetic blueprint, and ultimately contributed 25 percent of the finished sequence. Although the Human Genome Project was completed in 2003, research continues to not only finish sections of the sequence that contain gaps, missing information and misaligned or misrepresented regions but also to understand how genomic variations account for diversity and disease.

“We are focused on many of the issues associated with sequencing human genomes, which include resolving complex repetitive or highly variable regions, improperly assembled areas, and regions for which no sequence exists in the current reference resource,” said Richard K. Wilson, Ph.D., Director of The Genome Institute and Professor of Genetics at Washington University in St. Louis. “We will use BioNano’s Irys System to produce high-quality human genome sequences and better understand regions of the genome that are associated with human disease."

The GRC was created to improve the sequence quality and accuracy of the human assembly as well as improve the reference genomes of other model organisms, including the mouse and zebrafish, which are often used as models for human disease. The members of the Genome Reference Consortium are The Genome Institute at Washington University, National Center for Biotechnology Information (NCBI), European Bioinformatics Institute (EBI), and Wellcome Trust Sanger Institute.

“The Irys System will be used to order and orient the sequences from next-generation sequencing technologies as well as penetrate difficult repeat regions to inform genome assembly, annotation, and structural variation analysis,” said Bob Fulton, Director of Project Development and Management at The Genome Institute. “With a clearer knowledge of structural variations we can begin to accurately assemble the human genome and use that information to discover biologically significant patterns in structural variations that are relevant to genetic diversity and human diseases.”

Genome maps built with the Irys System reveal the biologically and clinically significant order and orientation of functionally relevant components of complex genomes, including genes, promoters, regulatory elements as well as the length and location of long areas of repeats across the entire genome. Assembling such a comprehensive view of a genome has previously been a major hurdle for

genomics researchers because information about repetitive elements and structural variations are lost with next generation sequencing methods.

“We are excited to help The Genome Institute in their mission to make the best possible human reference available to scientists around the world,” said Todd Dickinson, Ph.D., vice president, Global Commercial Operations of BioNano Genomics. “Comprehensive detection of all genome variation will lead to a better understanding of human health and disease and will be essential to advance genomic research into the clinic.”

About Irys System

Irys makes it possible to routinely and accurately detect genomic structural variation and to finish genome assemblies. The fully automated Irys benchtop instrument uses the IrysChip to uncoil and confine long DNA molecules in proprietary Nanochannel Arrays™ where they are uniformly linearized in a highly parallel display for high-resolution, single-molecule imaging. Irys does not employ DNA fragmentation or amplification, which are typical with next-generation sequencing. The result is sequence information over extremely long “reads” ranging from hundreds of kilobases to a megabase, where the sample’s valuable structural information is preserved. Irys makes it possible for researchers to directly observe structural variants including replications, deletions, translocations and inversions.

About BioNano Genomics

Headquartered in San Diego, BioNano Genomics is delivering an altogether better way of gaining a fully informed understanding of genomes. The Company’s platform provides researchers and clinicians the most comprehensive, organized and actionable picture of a genome with unprecedented insights into how the individual components of genomes are ordered, arranged, and interact with each other. BioNano Genomics works with institutions in life science, translational research, molecular diagnostics and personalized medicine. The Company is supported by private investors and grant funding from genomics programs at federal agencies, including the NIH and NIST-ATP.

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