



## **BIONANO GENOMICS INTRODUCES NEW PRODUCT TO FULLY DETECT HUMAN GENOMIC STRUCTURAL VARIATION AND FINISH GENOME ASSEMBLIES**

*First look of the new, high-throughput V2 chip for the Irys System will be at ASHG2013*

SAN DIEGO AND BOSTON—October 22, 2013—[BioNano Genomics](#) announced today the launch of the IrysChip™ V2, a new, high-throughput chip for the Irys™ System that now supports human genome analysis. The product introduction occurs at the American Society for Human Genetics (ASHG) Annual Meeting taking place October 22 to 26 in Boston. With a 15-fold increase in throughput, the new chip now makes possible the full detection of structural variation present in each human genome and significantly advances the finishing of genome assemblies. Structural variation can involve millions of nucleotides of heterogeneity within every genome and may be a fundamental key to understanding human diversity and disease.

“We know of a handful of clinically significant, structural variations, like the Philadelphia chromosome associated with chronic myelogenous leukemia,” said Erik Holmlin, Ph.D., president and CEO of BioNano Genomics. “However, before this new chip for the Irys System, we have not had a way to comprehensively study structural variation. Today, researchers can use the Irys System to discover biologically significant patterns in structural variations, which can be relevant for developing new drugs and diagnostics as well as for creating new agriculture and biofuel products.”

Structural variation occurs in all genomes and includes genomic changes, such as deletions, duplications, copy-number variants, insertions, inversions and translocations. Recent research suggests that five percent of the human genome is defined as structurally variant, involving more than 800 independent genes as well as unknown effects on non-coding areas, which often contain regulatory elements that control gene expression. By analyzing extremely long stretches of DNA, up to 1 million base pairs, the Irys System is a long-read technology that allows the direct observation of the natural DNA structure to detect the structural variations present in each genome.

“Movement or changes in genomes are common. Large segments of the genome are being moved around, rearranged, and inverted, but these variations are not able to be quickly, accurately and reproducibly detected by traditional methods,” said Pui-Yan Kwok M.D., Ph.D., Henry Bachrach Distinguished Professor at UCSF School of Medicine.

Dr. Kwok, a long-time Irys user and whose lab is a beta test site for the new IrysChip V2, continued, “Using our Irys System, we have recently identified thousands of copy-number variations (CNVs) across a family of individuals. The significantly increased throughput of the new V2 chip now makes human analysis routine. Further, by leveraging the long single-molecule detection on Irys, we can exhaustively characterize structural variation inherited as specific parental haplotypes across entire chromosomes.”

Short-read next-generation sequencing (NGS) methods, where the DNA is cut into smaller pieces, has left many genomes largely incomplete. Short-read methods lose the information about structural variations, including the underlying arrangement of genes and the length and location of long areas of repeats, which often occur in large genomes of plants and animals. Because of this information loss, genome assembly has been a very difficult, inaccurate and labor-intensive task, and comparative genome analysis of structural variation has been hindered.



“By significantly improving the process to finish a genome assembly, we are not only freeing time and resources for deeper analysis and research, but we are also unlocking the information contained in a complete genome,” said Dr. Holmlin. “Launching this new chip means our partners across the world, who are using the Irys System to look at structural variation and finish genome assemblies in everything from bacteria and viruses to insects and plants, can now expand to studying human and can do it all faster.”

BioNano is holding a workshop at ASHG entitled, *“The Missing Genome: Revealing the True Extent of Structural Variation in Humans with Single-Molecule De Novo Assembly,”* which will take place October 24 from 12:30 pm to 2:00 pm in Room 260 of the Boston Convention and Exposition Center.

### **About Irys**

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 NIH and NIST-ATP.

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