



BioNano Next-Generation Mapping Yields Superior Genomic Assemblies with Greater Contiguity

Study Published in Nature Methods Describes Use of BioNano Irys® with Illumina-Based NGS Methods

SAN DIEGO, CA – May 10, 2016 – BioNano Genomics, the leader in physical genome mapping, today announced publication of another study describing application of its next-generation mapping (NGM) technology as the critical component in achieving high-quality *de novo* human genome assemblies with unprecedented contiguity. In the current study, researchers combined NGM with Illumina-based approaches for next-generation sequencing (NGS) and demonstrated that when the GemCode™ Technology from 10X Genomics is used for sequencing library preparation, the resulting hybrid assembly has comparable or slightly better contiguity to that of NGM-NGS hybrid assemblies achieved with PacBio in previous studies such as that of Pendleton and colleagues at Mt. Sinai published in a June 2015 issue of *Nature Methods*. The results of the new study were published online today in the article "A hybrid approach for *de novo* human genome sequence assembly and phasing" in the May 2016 issue of *Nature Methods*.

Pui-Yan Kwok, M.D., Ph.D., senior author of the article and the Henry Bachrach Distinguished Professor at the University of California, San Francisco, stated, "Genomic information derived from long-range technologies allows us to take discovery several steps beyond that based solely on single nucleotide polymorphisms (SNPs). Using BioNano's Next-Generation Mapping solution in combination with medium-length contiguity information provided by newer sequencing methods, any researcher can produce highly contiguous *de novo* genome assembly of complex genomes. Our approach makes it possible to detect structural variations with base pair resolution across the genome in one set of experiments and open up the field for many new applications."

Dr. Pui Kwok and colleagues at University of California in San Francisco's Cardiovascular Research Institute/Institute for Human Genetics and researchers at the University of Cape Town's Department of Molecular and Cell Biology used BioNano's Irys® System to build physical maps of the genome in which long-range information spanning complex regions such as repeats and other structural variations is intact. They integrated the maps with sequence assemblies based on Illumina short-read sequencing and library preparation from 10X Genomics for linked-reads. This combination of NGM, linked-reads and Illumina-based sequencing, similar to the combination of PacBio sequencing and NGM, overcomes limitations of NGS alone. Here, the hybrid approach with BioNano led to a 57-fold increase in N50 scaffold length over Illumina generated N50 scaffolds and a 4.8-fold increase over the N50 of hybrids with only 10X Genomics' linked-reads and Illumina NGS. The authors pointed out how some 14.3 Mbp of the NA12878 genome, not represented in the hg38 reference genome, was identified and correctly located in the assembly. These findings underscore the limitations of a single reference genome and the importance of using NGM-NGS hybrid assemblies to build highly contiguous reference quality genomes on a population basis to minimize the findings of pseudo-variations that are associated with errors or limits of a given reference.

Erik Holmlin, Ph.D., President and Chief Executive Officer of BioNano Genomics, commented, "I'm pleased to see the growing use of NGM as a key tool in obtaining such high quality genome assemblies. The 10X approach makes it easier to integrate Illumina's NGS together with NGM and expands the opportunity to create much better reference genomes. Improving reference genomes will help applications of BioNano's NGM as a stand-alone tool for discovery of novel, clinically and biologically relevant structural variations (SVs) as well as make NGM a more efficient approach to detecting those SVs already known for their clinical and translational applications."

The *Nature Methods* publication is available at:

<http://www.nature.com/nmeth/journal/vaop/ncurrent/full/nmeth.3865.html>

About BioNano Genomics

BioNano Genomics, Inc., the leader in next-generation mapping (NGM), provides customers with genome analysis tools that advance human, plant, and animal genomics and accelerate the development of clinical diagnostics. The Company's Irys® System uses NanoChannel arrays integrated within the IrysChip® to image genomes at the single-molecule level with average single-molecule lengths of about 350,000 base pairs, which leads the industry. The long-range genomic information obtained with the Irys System helps decipher large, complex DNA repeats, which are the primary cause of inaccurate and incomplete genome assembly.

On its own, next-generation mapping with the Irys System enables detection of structural variants, many of which have been shown to be associated with human disease as well as complex traits in plants and animals. As a companion to next-generation sequencing, next-generation mapping with the Irys System integrates with sequence assemblies to create contiguous hybrid scaffolds that reveal the highly-informative native structure of the chromosome.

Only BioNano Genomics provides long-range genomic information with the cost-efficiency and throughput to keep up with advances in next-generation sequencing.

The Irys System has been adopted by a growing number of leading institutions around the world, including: National Cancer Institute (NCI), National Institutes of Health (NIH), Wellcome Trust Sanger Institute, , BGI, Garvan Institute, Salk Institute, McDonnell Genome Institute of Washington University, and University of California, Davis. Investors in the Company include Domain Associates, Legend Capital, Novartis Venture Fund and Monashee Investment Management.

For more information, please visit us at www.BioNanoGenomics.com.

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