BioNano's Next-Generation Mapping (NGM) Used to Reveal the Genome Structure of the Korean Genome

NGM plays important role in revealing full range of structural variations across human genomes
Highlights importance of population-specific references to advance clinically-relevant precision medicine

SAN DIEGO, CA – October 14, 2016 – BioNano Genomics, Inc., the leader in physical genome mapping, today announced that the *de novo* assembly of the genome of a Korean individual (AK1) has been just published in the October print issue of the journal, *Nature*. The article is titled, "*De novo* assembly and phasing of a Korean human genome."[1]

Researchers used next-generation mapping (NGM) using the Irys® System from BioNano Genomics and SMRT sequencing from Pacific Biosciences as the primary tools for *de novo* genome assembly, they added 10x Genomics to assist with haplotype phasing and Illumina sequencing for sequence error correction. The result is a high quality, diploid reference genome with unprecedented completeness, as measured by the hybrid scaffold N50 of approximately 45 Mbp. Remarkably, eight chromosomal arms are resolved into single scaffolds. The observed quality metrics make this Korean genome assembly the most contiguous diploid human genome assembly to date.

In addition, findings from the AK1 assembly revealed more than 18,000 previously unreported structural variations (SVs) when compared to GRCh38, a commonly used human reference. In a time of precision medicine, population-specific reference genomes are becoming essential to address population specific variations. This study provides extensive investigation of novel, previously unreported and Asian-specific SVs, and reports clinically-relevant alleles. In total, 47% of the insertions and 76% of the deletions represent new discoveries.

Jeong-Sun Seo, M.D., Ph.D., from the Seoul National University College of Medicine and lead author of the study, commented, "Through this research we demonstrate the power of de novo genome assembly and phasing by integrating multiple genomic technologies, including NGM, SMRT sequencing, and others for the generation of a high-quality contiguous genome assembly and detection of the full range of SVs. In particular, NGM played a critical role as it provides longer range information compared to other long read commercial solutions, so we relied on NGM when there were conflicts in the data. Importantly, the findings demonstrate the important genomic differences of the Asian ancestral group from other groups, and how ethnic diversity limits the utility of a single human genome reference. Moreover, it supports the argument that more representative, population-specific, reference-quality genome assemblies must be incorporated into the population-based initiatives underway around the world to uncover ethnic diversity underlying the human genome."

Erik Holmlin, Ph.D., CEO of BioNano Genomics, commented, "NGM is an essential tool for achieving the contiguities described in this study and for revealing the true structure of the genome. Long-range genomic information attained from optical mapping with BioNano's Irys® System provides the foundation on which complete and accurate reference-quality genome assemblies can be built, which will prove crucial in realizing precision medicine."

Reference

1. Seo, J.S. *Nature*. 13 October 2016. doi:10.1038/nature20098

About The Irys® System

The Irys® System provides a comprehensive view of the whole genome via single molecule imaging, facilitating high resolution de novo mapping without the guidance of a reference genome and generates valuable insights about the biology of the genome based on information about the order, orientation, arrangement, and interaction of genomic components. The Irys System uses IrysPrep Reagents to extract and label long DNA molecules and the IrysView and IrysSolve software to provide powerful *de novo* assemblies and analysis of the genome.

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 DNA at the single-molecule level with average single-molecule lengths of about 350,000 base pairs, which leads the genomics industry. The long-range genomic information obtained with the Irys System helps decipher complex DNA involving 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 (NGS), 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, Mount Sinai and Washington University. Investors in the Company include Domain Associates, Legend Capital, Novartis Venture Fund and Monashee Investment Management.

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

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