BioNano Genomics Highlights Study Using Next-Generation Mapping for 7-fold Improvement in Structural Variation Detection versus NGS

Results published today in peer-reviewed journal, Genetics

SAN DIEGO, CA – January 5, 2016 – BioNano Genomics, the leader in physical genome mapping, today announced results from a study demonstrating the enhanced ability of single-molecule next-generation mapping (NGM) using BioNano's Irys® System to detect and reveal genome-wide large structural variations, including many previously unpublished variants that may have the potential to disrupt gene function or regulation.

Results from the study, entitled "Genome-wide structural variation detection by genome mapping on nanochannel arrays,"[1] were published early online in the peer-reviewed journal *Genetics* and have been published today in the January 5, 2016 issue, which is the first issue of the journal's Centennial year.

Led by researchers from four institutions using multiple independent algorithmic analysis methods, the study confirmed that single-molecule and *de novo* genome maps generated using BioNano's Irys System for the well-studied CEU trio from the 1000 Genomes Project enabled efficient identification and validation of structural variants – including insertions, deletions, and inversions – greater than 5 kilobase pairs (kbp) in size. The study detected and manually confirmed seven times more large insertions and deletions than previously found in the 1000 Genome Consortium, which is likely a result of the inability of next-generation sequencing (NGS) to accurately detect large structural variations.

Pui-Yan Kwok, M.D., Ph.D., the Henry Bachrach Distinguished Professor at the University of California, San Francisco, commented, "Despite the rapid reduction in cost and increase in throughput of NGS, comprehensive large structural variation information in complex genomes remain elusive from routine NGS study. Using BioNano's Irys System, we identified 909 insertions and 661 deletions, including 800 novel insertions that were unidentified in the 1000 Genomes Project – for a remarkable seven-fold improvement in sensitivity in detecting structural variations compared to NGS. Based on our study, we were able to determine the novel insertions and deletions with maps assembled from the native long DNA molecules in one experiment. The detection of numerous novel insertions and deletions in this study demonstrates that long-range genome analysis on the Irys platform is a very powerful approach."

Han Cao, Ph.D., Chief Scientific Officer of BioNano Genomics, added, "This study demonstrates the ability of our Irys System to provide high-quality genomic architecture information for the specific detection of structural variations with higher sensitivity than NGS in the human genome. This improvement is important because large structural variants are known to be associated with diseases, especially complex traits. Using NGM, we identified at least four homozygous deletions that disrupt genes known to affect disease susceptibility, confirming that NGM reveals gene disruptions that may have clinical implications. In our view, the addition of NGM as a genomics tool will enable researchers to look at the full scope of genome variations including large structural variations. NGM together with NGS will increase our understanding of the human genome's relationship with phenotypes and diseases. We believe that this data will further accelerate the wide adoption of next-generation mapping technology among genetic and biomedical researchers."

References

1. Mak, A. et al. "Genome-wide structural variation detection by genome mapping on nanochannel arrays." *GENETICS*, January, 5 2016. Vol. 202 no. 1, 351-362. Available online at: http://www.genetics.org/content/202/1/351.full

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, Broad Institute of MIT and Harvard, BGI, Garvan Institute, Salk Institute, and McDonnell Genome Institute of Washington University. Investors in the Company include Battelle Ventures, Domain Associates, Legend Capital, Novartis Venture Fund, Federated Kaufmann, Monashee Investment Management, and Gund Investment Corporation.

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

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