Bionano Genomics Reports Industry-Leading Detection of Structural Variants with More than 90% Sensitivity

New Bionano Solve 3.0 assembly and analysis software to be released this quarter

SAN DIEGO, CA – February 6, 2017 – Bionano Genomics, the leader in physical genome mapping, today released the first comprehensive dataset outlining the performance of large structural variant (SV) detection in human genomes. In a pre-print publication appearing online, Bionano describes its new assembly and SV discovery algorithms that far outperform array or sequencing-based technologies. The study, "Rapid Automated Large Structural Variation Detection in a Diploid Genome by NanoChannel Based Next-Generation Mapping," demonstrates that Bionano's next-generation mapping (NGM) technology comprehensively detects large SVs, including both heterozygous and homozygous variants, with unrivaled sensitivity and precision in the industry.

Precision medicine initiatives require accurate analyses of human genomes. While improvements in sequencing technology have allowed for spectacular progress in the detection of single nucleotide changes, the analysis of larger SVs has remained ineffective. Standard methodologies for detecting SVs have significant limitations. Chromosomal microarray is insensitive to novel insertions, mobile element insertions, many low copy repeats, and all balanced translocations and inversions. Short-read sequencing methods have low sensitivity to most large variants and often fail in repetitive regions or those with high GC-content. Long-read sequencing has better sensitivity for heterozygous SVs, but is unable to span larger repetitive regions.

The new Bionano Solve 3.0 assembly pipeline within the Bionano Access analysis and visualization software introduces a number of improvements, including the capability to integrate SV calls collected from two separate experiments using orthogonal labeling enzymes. The improvements allow for unprecedented sensitivity and precision to detect large SVs: results showed that Bionano detects all insertions and deletions larger than 1 kilobasepair (kbp) with more than 90% sensitivity. More specifically, Bionano detects homozygous insertions and deletions 1.5 kbp to megabasepairs in size with more than 99% sensitivity while detecting the typically much harder to detect heterozygous insertions and deletions larger than 1.5 kbp with 87% sensitivity. These high sensitivity rates far outperform sequencing-based SV calling. Best-reported rates for PacBio, arguably the next-best solution for SV detection, show 78% sensitivity for homozygous and 54% sensitivity for heterozygous SVs of the same size when tested on the exact same samples, and almost twice the false positive rate as Bionano.

The study also showed that Bionano's NGM detects translocations, whether balanced or unbalanced, with 98% sensitivity, and the chromosomal breakpoint can be determined to a median distance of 2.9 kbp – which is approximately 1,000 times more precise than karyotyping and FISH. Bionano's automated SV calls have a false positive rate of less than 3% for each reported type of structural event, giving researchers high confidence of those SV calls. Unlike array or sequencing technologies, Bionano's NGM also detects inversions, repeat array expansion or contractions, and visualizes complex genomic rearrangements.

Erik Holmlin, Ph.D., CEO of Bionano Genomics, commented, "Geneticists increasingly understand the clinical importance of correct order and orientation in the genome. As demonstrated by this study, highly accurate discovery and analysis of large structural variants is now available. These comprehensive results demonstrate that Bionano's structural variation analysis is by far the most cost-effective, fast, and

accurate technology able to detect a wide spectrum of SV types – balanced and unbalanced, simple and complex, spanning a wide size range."

Studies of patients with cancer and genetic disorders that used Bionano Access analysis will be announced at the upcoming Advances in Genome Biology and Technology (AGBT) General Meeting. Bionano Access will be released this quarter. The bioRxiv publication is available online at http://biorxiv.org/content/early/2017/02/01/102764

About Bionano Genomics

Bionano Genomics, Inc. provides next-generation mapping (NGM), which is the leading solution in physical genome mapping, offering customers whole genome analysis tools that reveal true genome structure and enabling researchers to capture what's missing in their data to advance human, plant and animal genomic research. NGM uses NanoChannel arrays 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 NGM detects and deciphers structural variations (SVs), which are large, complex DNA segments involving repeats that are often missed by sequencing technologies and which are a leading cause of inaccurate and incomplete genome assembly.

As a stand-alone tool, NGM enables the accurate detection of SVs, many of which have been shown to be associated with human disease as well as complex traits in plants and animals. As a complementary tool to next-generation sequencing (NGS), NGM integrates with sequence assemblies to create contiguous hybrid scaffolds for reference-quality genome assemblies that reveal the highly informative native structure of the chromosome. NGM also provides the additional ability to verify, correct and improve a NGS-generated genome assembly.

Only Bionano provides long-range genomic information with the cost-efficiency and high throughput to keep up with advances in NGS.

NGM 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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