Bionano Genomics Announces Inversion Detection Algorithms With Unmatched Sensitivity

Saphyr now calls 98% of inversions genome wide

SAN DIEGO, Sept. 05, 2017 (GLOBE NEWSWIRE) -- Bionano Genomics®, Inc., a company focused on genome structure analysis, today released updated algorithms, analysis tools and visualization software enabling industry leading performance for the genome wide detection of inversions.

Previous releases have focused on the automated calling of large insertions, deletions and translocations. Bionano's genome mapping platform Saphyr™ detects 90% of heterozygous insertions and deletions larger than 1 kilobase pairs (kbp), and 98% of all translocations. Today's release focuses on mapping the order and orientation of the genome, by greatly improving the ability to identify inversions of 30 kbp up to megabases in size.

Inversions are the invisible variants, and have traditionally been the hardest to detect structural events. They are balanced, without gain or loss of sequence, and unlike translocations they don't create major changes in genomic context. Inversions often escape detection by traditional cytogenetic techniques. Chromosomal Microarray can not detect balanced events, and metaphase chromosome spreads can only visualize some megabase size inversions. Next Generation Sequencing approaches often miss inversions because reads from inside the inversion map back to the reference without any indication that the orientation has changed. Detection of the breakpoints often fails, especially if the inversion is flanked by segmental duplications, repeat arrays or other non-unique sequences.

Bionano's Saphyr images megabase length molecules that span most repeats and anchor hundreds of kilobase pairs of sequence flanking inversion breakpoints. The new structural variant detection algorithms analyzing these extremely long molecules now have 98% sensitivity to pick up inversions larger than 50 kbp throughout the genome. All of Bionano's structural variants (SVs) are reported in dbVar-compliant VCF format to ease the integration with smaller variants found by sequencing.

As part of this release, Bionano algorithms now also identify tandem duplications. The improved Variant Annotation Pipeline uses single molecule analysis and Bionano's growing database of common SVs to filter those variants that are rare, *de novo*, or likely to affect genes of interest. This is especially important in genetic disease and cancer studies. Updates to the genome visualization software Bionano Access™ improve functionality, visualization and ease of use, and add compliance with security policies in HIPAA-compliant facilities.

Bionano Solve 3.1, Bionano Access 1.1 and Bionano Tools 1.1 are available now as free downloads from bionanogenomics.com.

Mark Borodkin, Bionano's Vice President of Systems Development comments: "These recent improvements in calling inversions now allow Bionano to provide an automated, highly sensitive platform for the discovery of all major SV types. SVs such as balanced translocations and inversions are some of the most difficult to elucidate, and as such, provide a largely untapped area of disease research. These algorithmic improvements, together with the speed of discovery provided by Bionano's Saphyr system, add a powerful complement to sequencing to advance the understanding of disease."

About Bionano Genomics®

Bionano Genomics, Inc. provides the Irys® and Saphyr™ systems for next-generation mapping (NGM), which is the leading solution in physical genome mapping. NGM offers 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.

Bionano Genomics Media Contact
The Ruth Group
Kirsten Thomas
kthomas@theruthgroup.com
(508)280-6592