The Largest Study Conducted with Bionano's Platform to Date Reveals Never-Before-Seen Human Genomic Structural Variations

Bionano optical mapping uniquely identifies structural variation, a previously inaccessible class of genomic variation associated with disease phenotypes, in a study comprising 154 humans across 26 distinct populations

SAN DIEGO, March 05, 2019 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (NASDAQ: BNGO), a life sciences instrumentation company that develops and markets Saphyr®, a platform for ultra-sensitive and ultra-specific structural variation detection in genome analysis, announced the publication of the largest study of human genomes on its platform to date.

In the study published in the March 4 issue of Nature Communications, a team of scientists from UCSF, Drexel University, and The Chinese University of Hong Kong used the Bionano system to analyze structural variation in a record 154 humans. The genomes of these individuals had been previously sequenced as part of the 1000 Genomes Project.

Bionano optical mapping identified 8.5 times more large insertions in the same samples than previously reported by the 1000 Genomes Project using short-read sequencing, and 35% more large deletions. Not surprisingly, many of the large structural variations were flanked by repetitive elements, which short-read sequencing fails to accurately resolve, rendering the included structural variants undetectable.

Many of the structural variants (SVs) and copy number variants (CNVs) associated with disease phenotypes show significant ethnic variation as well. This finding highlights a severe weakness in the current practice in personalized medicine and population-wide sequencing projects where short-read sequences are aligned to just a single reference genome. Without a thorough characterization of the underlying structural variation, the study authors state, "alignment of short-reads to these ethnically variable disease-associated regions will lead to errors in analysis."

The long-range information provided by megabase-size molecules only Bionano is capable of mapping allowed the authors to resolve variation in many of the most intractable regions of the genome, such as segmental duplications, subtelomeric regions, pericentromeric regions and repetitive regions of the Y-chromosome. In addition, the study identified ~60 Mb of non-redundant genome content not found in the hg38 reference genome.

Sven Bocklandt, PhD, Head of Scientific Affairs at Bionano Genomics, commented, "This study of the largest human population mapped on the Bionano platform demonstrates the power of Bionano's technology to reveal structural variation missed by short-read sequencing. The ethnically diverse study reveals that one reference genome does not fit all, and that it is impossible for a genome analysis based on short-read sequencing alone to correctly characterize all clinically relevant genome variation at the root of human disease in individuals across different populations. The sheer size of this study demonstrates that Bionano genome mapping is a high-speed, cost-effective technology suitable for large population-scale studies. Given recent advancements in the workflow for Saphyr, our most advanced system for optical mapping, studies like this one can now be done on a routine basis for only $500/sample in just 4 weeks of data collection. We expect to see Saphyr in many more discovery studies as well as leading the push into the clinic as the platform that makes digital cytogenetics a reality."

About Bionano Genomics
Bionano is a life sciences instrumentation company in the genome analysis space. The Company develops and markets the Saphyr system, a platform for ultra-sensitive and ultra-specific structural variation detection that enables researchers and clinicians to accelerate the search for new diagnostics and therapeutic targets and to drive the adoption of digital cytogenetics, a more systematic, streamlined and industrialized form of traditional cytogenetics. The Saphyr system comprises an instrument, chip consumables, reagents and a suite of data analysis tools.

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