Study Shows the Combination of OGM and NGS with Linked-Reads Detects Significantly More Clinically Relevant Variants Resulting in Higher Success Rates for Resolving Previously Unclassified Subjects in Genetic Disease Research

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SAN DIEGO, Oct. 14, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO), developer of the Saphyr® system that uses optical genome mapping (OGM) for the detection and analysis of structural variants (SVs), today announced the publication of a study in which OGM and next-generation sequencing (NGS) with linked-reads were used together to resolve genetic diseases that were previously unclassified after evaluation by whole-exome sequencing (WES) alone. This study, from the University of California, San Francisco (UCSF) and Children's Hospital Oakland (now UCSF Benioff Children's Hospital Oakland) and appearing in the September 23, 2021 peer-reviewed issue of *Nature Publishing Journal of Genomic Medicine*, shows the benefit of combining OGM with short-read sequencing for improved detection of clinically relevant variants in genetic disease research.

The study's authors, Shieh, *et al*., describe their Full-Genome Analysis (FGA) approach with automated analysis using NGS linked-read sequencing and OGM to evaluate a full spectrum of genetic variants found in inherited genetic diseases. FGA identified structural variants and small variants with an increase in detection capability of 40% (20 of 50 cases). The number of resolved cases attributable to SVs was notable in the study, as 50% of exome-negative cases (four of eight cases) were solved by identifying an SV or rearrangement. The authors also identified candidate variants in another 60% (18 of 30 cases) for future follow-up.

In one specific case, the authors found a rare 32 kb heterozygous de novo intronic duplication within the *NHEJ1* gene that was not detected by standard microarray analysis because it was small and intronic. It had also escaped detection in copy number variants called from short-read, whole genome sequencing (WGS) data but was easily identified with their FGA method using OGM.

The findings reinforce that the combination of OGM and NGS used in the FGA method detects and localizes SVs such as duplications missed by WGS, and can quickly identify translocations and phase variants across long distances. For individuals with undiagnosed conditions, these two technologies encompass what is currently provided by the combination of chromosome analysis – karyotyping, microarray testing and short-read WGS. FGA provides information beyond current assays and results in higher resolution genome maps that can be used for future studies.

As observed in the study, Bionano's Saphyr ® system, which can detect all classes of SVs, such as insertions, deletions, inversions, duplications, translocations and copy number variations, complemented NGS. Accordingly, when used together, they can provide the clinical research community with the ability to see the entire genome and identify a more comprehensive set of genetic variants to diagnose rare diseases accurately.

Erik Holmlin, PhD, CEO of Bionano Genomics, commented, "This study's results go beyond just reinforcing the power of OGM – it shows us how we can significantly impact people's lives by combining NGS and OGM in genome analysis to find answers. Fifty percent of the participants in this study had clinically relevant variants hidden in their genomes that WES alone had not uncovered. OGM, together with a linked-read assay using NGS, revealed key answers for these participants. We believe there is tremendous potential in using OGM together with NGS to understand disease from the very beginning of any study. Congratulations to Dr. Shieh and his team on this tremendous progress."

This publication is available at https://www.nature.com/articles/s41525-021-00241-5

About Bionano Genomics

Bionano is a genome analysis company providing tools and services based on its Saphyr® system to scientists and clinicians conducting genetic research and patient testing; it also provides diagnostic testing for those with autism spectrum disorder (ASD) and other neurodevelopmental disabilities through its Lineagen business. Bionano's Saphyr system is a research use only platform for ultra-sensitive and ultra-specific structural variation detection that enables scientists and clinicians to accelerate the search for new diagnostics and therapeutic targets and to streamline the study of changes in chromosomes, which is known as cytogenetics. The Saphyr system is comprised of an instrument, chip consumables, reagents and a suite of data analysis tools. Bionano offers genome analysis services to provide access to data generated by the Saphyr system for researchers who prefer not to adopt the Saphyr system in their labs. Lineagen has been providing genetic testing services to families and their healthcare providers for more than nine years and has performed more than 65,000 tests for those with neurodevelopmental concerns. For more information, visit bionanogenomics.com or lineagen.com.

Forward-Looking Statements of Bionano Genomics

This press release contains forward-looking statements within the meaning of the Private Securities Litigation Reform Act of 1995. Words such as "may," "will," "expect," "plan," "anticipate," "estimate," "intend" and similar expressions (as well as other words or expressions referencing future events, conditions or circumstances) convey uncertainty of future events or outcomes and are intended to identify these forward-looking statements. Forward-looking statements include statements regarding our intentions, beliefs, projections, outlook, analyses or current expectations concerning, among other things: the utility of the combination of OGM and NGS, including with respect to enabling the accurate diagnosis of rare diseases, and its potential benefits to the research community. Each of these forward-looking statements involves risks and uncertainties. Actual results or developments may differ materially from those projected or implied in these forward-looking statements. Factors that may cause such a difference include the risks and uncertainties associated with: the impact of the COVID-19 pandemic on our business and the global economy; general market conditions; changes in the competitive landscape and the introduction of competitive products; observations from studies may not be replicated or have the anticipated benefits once implemented by the clinical research community; changes in our strategic and commercial plans; our ability to obtain sufficient financing to fund our strategic plans and commercialization efforts; the ability of medical and research institutions to obtain funding to support adoption or continued use of our technologies; the loss of key members of management and our commercial team; and the risks and

uncertainties associated with our business and financial condition in general, including the risks and uncertainties described in our filings with the Securities and Exchange Commission, including, without limitation, our Annual Report on Form 10-K for the year ended December 31, 2020 and in other filings subsequently made by us with the Securities and Exchange Commission. All forward-looking statements contained in this press release speak only as of the date on which they were made and are based on management's assumptions and estimates as of such date. We do not undertake any obligation to publicly update any forward-looking statements, whether as a result of the receipt of new information, the occurrence of future events or otherwise.

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