Bionano's Saphyr Data Prove Essential in Creating the Most Comprehensive, Ethnically Diverse Collection of Genome Variation Ever Completed

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Article published in Science reveals that optical genome mapping uniquely makes thousands of large structural variant calls

SAN DIEGO, Feb. 25, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) announced the publication in *Science* of a study by the Human Genome Structural Variation Consortium (HGSVC) that used a combination of advanced sequencing and optical genome mapping (OGM) with Saphyr to assemble with high resolution and structural accuracy 64 haplotype-resolved human genomes from 32 individuals representing 25 different populations. The peer-reviewed publication, which previously appeared on the pre-print server bioRxiv, builds on the original Human Genome Project to better capture genetic diversity by cataloging both single nucleotide variations (SNVs) and structural variations (SVs) from each individual reference-quality genome assembly compared to the standard human genome reference. The resulting assemblies and catalog of SNVs and SVs can serve as a diversity panel that researchers and clinicians can use to accelerate the discovery of new therapeutic targets and pathogenic genetic variations by making it easier and faster to filter out variation that is representative of population diversity. They further enable population-specific studies on genetic predispositions to human diseases as well as the potential discovery of more complex forms of genetic variation.

Regarding OGM's contribution to the variants that were identified, OGM uniquely made 5,590 large SV calls missed by long-read based genome assemblies, corresponding to 1,175 unique SV loci. Many of these large SVs consisted of more complex rearrangements or overlap with large repetitive areas called segmental duplications which are associated with developmental delay and adult neuropsychiatric disease, highlighting the importance of OGM in genome structure analysis.

"Bionano is proud that OGM has made such a significant contribution to this impressive work," commented Erik Holmlin, PhD, CEO of Bionano Genomics. "OGM complements even the most advanced sequencing technology and plays an essential role in identifying large structural variants, including those in the most complex parts of the genome, as the authors of this study pointed out. We believe the study by the HGSCV underscores the point that if you are performing a genome analysis without getting an accurate structural picture, you are not doing a deep enough analysis. It further illustrates that when it comes to looking for large SVs, OGM with Saphyr has unmatched sensitivity."

The work by the HGSVC is an extension of the human genome project. In 2001, the International Human Genome Sequencing Consortium announced the first draft of the human genome reference sequence. This reference does not represent a single individual but instead is a composite of humans that could not accurately capture the complexity of human genetic variation. Building that reference, scientists have carried out many sequencing projects over the last 20 years to identify and catalog genetic differences between an individual and the reference genome. Those projects usually focused on small single base changes and missed SVs, which are widely viewed to be more likely than smaller genetic differences to interfere with gene function. This article in *Science* has been published by an international research team and represent a significant advance in the tools available for scientists and clinicians who want to discover new biology en route to new medicines and diagnostics that can improve outcomes for patients.

The publication is available at https://science.sciencemag.org/lookup/doi/10.1126/science.abf7117

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, and providing 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 researchers 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 provides 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 over nine years and has performed over 65,000 tests for those with neurodevelopmental concerns. For more information, visit www.bionanogenomics.com or www.lineagen.com.

Forward-Looking Statements

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 potential for OGM with Saphyr to improve outcomes for patients by enabling the discovery of new medicines and diagnostics; our beliefs regarding the potential benefits of Bionano's Saphyr technology; the significance of large SVs in genetic research; and the execution of Bionano's strategy. 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; 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, 2019 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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