



Bionano Genomics' Saphyr System Shown to be Indispensable for the Analysis of Certain Genetic Disease Causing Variants

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Recent publication shows Saphyr's unique capability in resolving extremely complex structural variants that can cause common genetic disorders called microdeletion syndromes

SAN DIEGO, Oct. 15, 2020 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) announced that a study led by scientists and clinicians from the Institute for Human Genetics at the UCSF School of Medicine and the Department of Pediatrics at the University of Colorado School of Medicine and published in bioRxiv used Bionano's proprietary genome imaging technology to identify novel disease causing variants in patients with three different genetic diseases and in a diverse control dataset of 154 individuals. The study found that Bionano's Saphyr System was able to comprehensively analyze complex genome structures called segmental duplications and helped identify several novel structural variations associated with each disease causing locus increasing the understanding of these diseases.

Segmental duplications are large segments of repetitive sequences tens to hundreds of thousands of base pairs in size. Short-read and long-read sequencing technologies cannot span these large segments of the genome. Only Bionano's optical mapping technology can image single molecules that are so long that they span the segmental duplications. These repetitive sequences can interact with each other when sperm or eggs are created and their rearrangement can cause severe genetic disease. Some of the most common of such diseases are microdeletions at 7q11.23, also known as Williams-Beuren syndrome (WBS), 15q13.3 microdeletion syndrome, 16p12.2 microdeletion syndrome and 22q11.2 deletion syndrome, also known as DiGeorge syndrome.

This study, published in bioRxiv, provides a population-level analysis of segmental duplications in 154 people and in patients with WBS, 15q13.3, and 16p12.2 microdeletion syndromes. Several novel SVs were detected for each locus, and the exact disease causing rearrangement was determined with much higher accuracy than was formerly possible without Saphyr. As previously announced, a recent publication in the journal Nature published on July 22, 2020 also discussed the unique contribution of Bionano's optical mapping technology to understanding the genetic causes of DiGeorge syndrome.

Erik Holmlin, Ph.D., CEO of Bionano Genomics commented, "The microdeletion and microduplication syndromes are common genetic disorders, yet the exact genomic structures that cause them have been difficult or impossible to characterize with current sequencing-based methods. Even though microdeletion syndromes are commonly represented by hallmark features, in many cases a wide variability in clinical features is observed. Being able to understand and measure the subtle structural differences in microdeletions among different patients could allow for better clinical or therapeutic management. An increasing number of studies have relied on Bionano's Saphyr system to characterize disease-causing structural variants that could not be correctly analyzed with other molecular techniques. We will continue to make our technology available to researchers everywhere who want to greatly expand the capabilities of their genomic analysis."

The publication is available at <https://www.biorxiv.org/content/10.1101/2020.04.30.071449v1.full>

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 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, and 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 contribution of Bionano's technology to the analysis or understandings of microdeletion syndromes and future development of better clinical or therapeutic management for such diseases; the effectiveness and utility of Bionano's technology in clinical settings; Saphyr's capabilities in comparison to other genome analysis technologies; the benefits of Bionano's optical mapping technology and its ability to facilitate genomic analysis in future studies; and Bionano's strategic plans. 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.

CONTACTS

Company Contact:

Erik Holmlin, CEO
Bionano Genomics, Inc.
+1 (858) 888-7610
eholmlin@bionanogenomics.com

Investor Relations Contact:

Ashley R. Robinson
LifeSci Advisors, LLC
+1 (617) 430-7577
arr@lifesciadvisors.com

Media Contact:

Darren Opland, PhD
LifeSci Communications
+1 (617) 733-7668
darren@lifescicomms.com



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