Largest Clinical Research Study to Date Evaluating Optical Genome Mapping for Analysis of Facioscapulohumeral Muscular Dystrophy Showed Concordance with Southern Blot and Reduced Turnaround Time by 50% as Compared to the Traditional Workflow

September 29, 2021

SAN DIEGO, Sept. 29, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO) today announced the publication of the largest clinical research study to date evaluating optical genome mapping (OGM) for facioscapulohumeral muscular dystrophy (FSHD), which concluded that OGM can be performed more quickly, accurately, and reproducibly than the current gold standard method of Southern blot analysis. This study, now available online before print in *The Journal of Molecular Diagnostics*, describes how OGM can be adopted as an alternative to Southern blot analysis for the identification of chromosomal abnormalities associated with FSHD, a neuromuscular disorder resulting in progressive weakness and atrophy of muscles.

FSHD is typically caused by a contraction of a D4Z4 repeat array near the telomere of chromosome 4 (chr 4) at 4q35, which activates a dormant toxic gene, DUX4, to be overexpressed in skeletal muscle. Because of its complex genotype, FSHD can often be challenging to detect using next-generation sequencing (NGS) and Southern blot analysis, which can be slow, laborious and require radioactive labeling. In this robust study of 351 participants, the authors Stence *et al*., validated the use of OGM with Bionano's Saphyr ® system to determine the size and haplotype of D4Z4 alleles to confirm a diagnosis of FSHD. The study's authors found that OGM had much higher sizing accuracy and was >99% concordant with Southern blot analysis.

"We are pleased to be using optical genome mapping for FSHD mutation analysis because it more reliably identifies the patient's mutation and simplifies the initial analysis for the laboratory," said Aaron D. Bossler, MD, PhD, FCAP, Clinical Professor and Director, Molecular Pathology Laboratory at University of Iowa Hospitals and Clinics. "The extensive validation that we conducted with optical genome mapping demonstrates the power of this new technology to decrease time to result, improve standardization, and help tease out complex cases."

OGM is well-suited for analyzing inherited genetic disorders like FSHD that require the measurement of long, intact DNA molecules for accurate sizing. The higher resolution of OGM relative to traditional techniques can enable improved detection and discrimination ability for clinically relevant variants, as was observed in this study.

"The study completed by Dr. Stence and team demonstrated the superior performance of OGM over traditional molecular genetic testing for FSHD," commented Erik Holmlin, PhD, CEO of Bionano Genomics. "As the largest OGM clinical study to date, the study demonstrated that using the Saphyr system to perform FSHD analysis resulted in a significantly faster turnaround time, as the OGM turnaround time was only 5 days as compared to Southern blot at 11 days. The ability to obtain accurate, reliable data more quickly allowed for the timely application of this information for the management of study participants. We believe that studies like this one could pave the way for broad adoption of OGM for a wide variety of applications in genetic disorders and cancer."

This publication is available at https://pubmed.ncbi.nlm.nih.gov/34384893/

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 www.bionanogenomics.com or www.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 impact of the study results discussed in this press release, including our expectations regarding the study's potential to increase adoption of OGM for analysis of FSHD or other genetic disorders; OGM's superiority in genomic analysis as compared to traditional techniques; the potential benefits resulting from a combination of OGM and NGS technologies; and the advancement of our business 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; incompatibility between our and NGS technologies; future study results that may contradict the results of the study identified in this press release; 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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Source: Bionano Genomics