# Largest Study of Mendelian Disease Highlights how OGM, alongside other Techniques, May Enhance Success Rates in Identifying Molecular Causes of Genetic Disease

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SAN DIEGO, Sept. 14, 2023 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO), today announced the publication of a study detailing the challenges associated with the detection and interpretation of causal variants in Mendelian disease, using a cohort of 4,577 families. The study authors conducted a comprehensive analysis of samples from the cohort in which non-sequencing-based challenges were encountered, and then reanalyzed samples using exome sequencing (ES), whole-genome sequencing (WGS), chromosomal microarray (CMA) and optical genome mapping (OGM), finding that addressing these challenges alone could boost diagnostic yield of causal variants by approximately 71% and underscoring the need for a thorough approach to undiagnosed diseases.

The study authors reanalyzed and reinterpreted 314 samples with negative clinical exome or genome sequencing using a combination of traditional and novel techniques and were able to identify the likely causal variant in 54.5% of samples. OGM was used to analyze samples that remained negative after ES and was particularly relevant in the detection of large variants that were below the limit of detection of CMA.

OGM's utility was illustrated in a pedigree structure case where researchers reviewed challenges associated with imprinting disorders appearing to be autosomal recessive phenotypes. OGM was used to analyze three pediatric cases of split hand/foot malformation syndrome after karyotyping, ES and RNA sequencing all failed to identify a causal variant. OGM revealed a tandem duplication impacting multiple genes in all three cases, indicating paternal gonadal mosaicism. In addition to this finding, the authors reported additional instances where OGM was used to resolve cases involving structural variants coming from multiple classes that were highly complex.

"This case study, published in *Nature*, is the largest study of its kind on Mendelian disorders and it highlights the fact that definitive molecular diagnosis of disease is heavily encumbered by a number of complexities related to the actual data generation itself. The authors demonstrate that the combination of multiple techniques may reveal additional information that can help improve the success rate of molecular diagnosis in Mendelian disease. We believe they also underscore OGM's ability to resolve highly complex cases involving structural variations and show that OGM may play a significant role in covering the gap in variant detection left by other methods. The findings of this combined approach involving multiple orthogonal methods give researchers a rich data set that may result in increased number of diagnoses, which can inform treatment decisions and reproductive choices," commented Erik Holmlin, president and chief executive officer of Bionano.

The publication can be viewed here: https://www.nature.com/articles/s41467-023-40909-3#Sec1.

**About Bionano**

Bionano is a provider of genome analysis solutions that can enable researchers and clinicians to reveal answers to challenging questions in biology and medicine. The Company's mission is to transform the way the world sees the genome through OGM solutions, diagnostic services and software. The Company offers OGM solutions for applications across basic, translational and clinical research. Through its Lineagen, Inc. d/b/a Bionano Laboratories business, the Company also provides diagnostic testing for patients with clinical presentations consistent with autism spectrum disorder and other neurodevelopmental disabilities. The Company also offers an industry-leading, platform-agnostic software solution, which integrates next-generation sequencing and microarray data designed to provide analysis, visualization, interpretation and reporting of copy number variants, single-nucleotide variants and absence of heterozygosity across the genome in one consolidated view. The Company additionally offers nucleic acid extraction and purification solutions using proprietary isotachophoresis technology. For more information, visit www.bionano.com, www.bionanolaboratories.com or www.purigenbio.com.

*Unless specifically noted otherwise, Bionano's OGM products are for research use only and not for use in diagnostic procedures.*

**Forward-Looking Statements of Bionano**

This press release contains forward-looking statements within the meaning of the Private Securities Litigation Reform Act of 1995. Words such as "believe," "can," "could," "may" and similar expressions (as well as other words or expressions referencing future events, conditions or circumstances and the negatives thereof) 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 of OGM, alongside other techniques, to reveal additional information and enhance success rates in identifying molecular causes of genetic disease, boost diagnostic yield of causal variants in Mendelian disease and play a significant role in covering the gap in variant detection left by other methods; the possibility that involving multiple orthogonal methods may result in increased number of diagnoses, which can inform treatment decisions and reproductive choices; and other statements that are not historical facts.

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 geopolitical and macroeconomic developments, such as recent and future bank failures, the ongoing Ukraine-Russia conflict, related sanctions, and any global pandemics, on our business and the global economy; challenges inherent in developing, manufacturing and commercializing products; our ability to further deploy new products and applications and expand the markets for our technology platforms; failure of our OGM solutions to be adopted for the detection and interpretation of causal variants in Mendelian disease; the failure of OGM in combination with other techniques to enhance success rates in identifying molecular causes of Mendelian disease in a manner consistent with the study results described in this press release; future study results that contradict or do not support the study results described in this press release; our expectations and beliefs regarding future growth of the business and the markets in which we operate; changes in our strategic and commercial plans; our ability to obtain sufficient financing to fund our strategic plans and commercialization efforts; and 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, 2022 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 are under no duty to update

any of these forward-looking statements after the date they are made to conform these statements to actual results or revised expectations, except as required by law. You should, therefore, not rely on these forward-looking statements as representing our views as of any date subsequent to the date the statements are made. Moreover, except as required by law, neither we nor any other person assumes responsibility for the accuracy and completeness of the forward-looking statements contained in this press release.

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