Bionano Announces Publication that Demonstrates OGM's Utility for Resolving Complex Genetic Variation in Inherited Retinal Disease

April 6, 2023

SAN DIEGO, April 06, 2023 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) today announced the publication of a study from researchers at Radboud University Medical Center showing the use of optical genome mapping (OGM) to identify gene disruptive structural variants (SVs) that might contribute to inherited retinal diseases (IRDs). OGM detected a large inversion (173 megabases) impacting the USH2A gene that was initially ignored as a false positive in short-read next-generation sequencing (NGS) data. The authors sought to determine whether OGM could improve SV detection in genetically unexplained IRD cases. Their results identified several pathogenic SVs had been overlooked in their initial genome analyses by NGS. The publication illustrates the potential impact of combining NGS and OGM for more comprehensive variant analysis.

Inherited retinal diseases (IRDs) are a clinically and genetically heterogeneous group of disorders that can cause severe vision loss or even blindness and are often degenerative. Genetic research in this area may help to improve the accuracy of diagnosis, prognostication, and treatment prospects of targeted therapeutics. Approximately 30% of individuals suspected of IRD lack a conclusive genetic diagnosis after genome sequencing has been performed.

Study authors used OGM to search for pathogenic SVs in an IRD sample and resolved a subject that had been genetically unexplained for decades.

- Researchers used OGM to analyze Usher syndrome type-II (OMIM 276901), a recessively inherited disorder characterized by retinitis pigmentosa and congenital hearing loss and associated with variants in several genes, an important one being USH2A
- OGM detected a 173 megabase USH2A-disruptive pericentric inversion on chromosome 1 that had been overlooked and misinterpreted during previous analysis by NGS. The finding by OGM provided a genetic explanation for this subject after decades of research. Reanalysis of NGS data for 427 IRD cases subsequently yielded 30 likely pathogenic SVs in 29 IRD probands. Notably, 8 of the identified pathogenic variants (>25%) were overlooked during the initial analysis

"We are pleased to see the study authors note OGM's utility for the detection of structural variants that may be relevant to the understanding of IRD. With new gene therapy and clinical trials related to IRD, research into the underlying genetic cause of IRDs is critical. We believe this study underscores the importance of OGM as an integral part of genome analysis, especially in complex disorders where it can aid in discovering new variants and validating the findings of other methods," commented Erik Holmlin, PhD, president and chief executive officer of Bionano.

This publication can be found here.

About Bionano Genomics

Bionano Genomics 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. Through its BioDiscovery business, 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. For more information, visit www.bionanogenomics.com, www.bionanolaboratories.com or www.biodiscovery.com

Bionano's OGM products are for research use only and not for use in diagnostic procedures.

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 "believe," "can," "could," "may," "might," "potential," 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, OGM's utility for genetic disease research including research directed to identifying gene disruptive structural variants (SVs) that might contribute to inherited retinal diseases (IRDs) , the ability and utility of OGM to detect SVs compared to next-generation sequencing (NGS), and the ability and utility of combining NGS and OGM data for more comprehensive variant analysis. 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: global and macroeconomic events, such as the impact of the COVID-19 pandemic and the ongoing Ukraine-Russian conflict and related sanctions, on our business and the global economy; general market conditions; changes in the competitive landscape and the introduction of competitive technologies or improvements to existing technologies; failure of OGM to prove useful for research in areas including rare genetic diseases, including IRDs; the ability of OGM better detect SVs that were missed or ignored when using NGS data; the ability to combine NGS and OGM data to achieve more comprehensive variant analysis; future study results contradicting the results reported in the paper referenced 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; 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, 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 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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