

## Presentation at ASHG Showcases New Capabilities for Optical Genome Mapping with Detection of Allelic Imbalance and Absence of Heterozygosity Further Expanding Its Utility in Revealing More Clinically Relevant Variants

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SAN DIEGO, Oct. 20, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO), developer of the Saphyr<sup>®</sup> system that uses optical genome mapping (OGM) for the detection and analysis of structural variants (SVs), today announced that Bionano scientists presented a poster at the American Society of Human Genetics (ASHG) conference that showcased new capabilities for OGM on the Saphyr<sup>®</sup> system with detection of allelic imbalance and absence of heterozygosity (AOH), which further expands its utility in revealing more clinically relevant variants. These new OGM capabilities are expected to be released to Bionano's customers in upcoming versions of our Access and Solve<sup>™</sup> software.

Regions with AOH, also referred to as loss of heterozygosity, regions/runs of homozygosity, or long continuous stretches of homozygosity are routinely used by researchers to gain genomic insights into the progression of various cancers and determine susceptibility for recessive disorders. For example, some regions with AOH may be indicative of uniparental isodisomy (UPD) or regions of the genome identical by descent (IBD).

In the poster presented at the ASHG conference titled, "Optical genome mapping capability expanded to enable detection of absence of heterozygosity," the study's authors, Rao, *et al.*, describe a method for AOH detection based on OGM results from the Saphyr<sup>®</sup> system. Measurement and representation of allelic imbalance enables OGM to detect triploidy and other chromosomal imbalances and may shed light on mosaic SVs. This capability could further expand the utility of OGM in constitutional genetic disease research.

Erik Holmlin, PhD, CEO of Bionano Genomics, commented, "This new capability for OGM helps strengthen our ability to support comprehensive genome analysis for cytogenomics and molecular pathology laboratories. We believe the detection of triploidy, regions associated with imprinted chromosomal disorders and IBD substantially improves the utility of OGM for clinical research applications. This increased utility could make the adoption of the Saphyr<sup>®</sup> system more compelling for labs seeking to compliment SNP-based microarrays and next-generation sequencing."

### About Bionano Genomics

Bionano Genomics' mission is to transform the way the world sees the genome through OGM solutions, diagnostic services and software. Bionano's genome analysis solutions can enable researchers and clinicians to reveal answers to challenging questions in biology and medicine. Bionano pioneered OGM, which is a workflow for ultra-sensitive and ultra-specific detection of SVs. OGM is enabled on the Saphyr<sup>®</sup> system, a single-molecule imaging instrument with reagents for isolation and sequence-specific labeling of ultra-high molecular weight DNA and software for SV detection and visualization. Bionano offers OGM solutions for applications across basic, translational and clinical research. Through its Lineagen business, Bionano also provides diagnostic testing for patients with clinical presentations consistent with autism spectrum disorder and other neurodevelopmental disabilities. Through its BioDiscovery business, Bionano also offers an industry-leading, platform-agnostic software solution, which integrates next-generation sequencing (NGS) and microarray data designed to provide analysis, visualization, interpretation and reporting of copy number variants, single-nucleotide variants and AOH across the genome in one consolidated view. In addition, this software is expected to serve as the foundation of Bionano's ongoing efforts to develop data interpretation solutions tailored for cytogenomics and molecular pathology labs where the combination of NGS and OGM can potentially reveal more answers in genetic disease and cancer research than NGS alone. For more information, visit [bionanogenomics.com](http://bionanogenomics.com), [lineagen.com](http://lineagen.com) or [biodiscovery.com](http://biodiscovery.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: our beliefs regarding the improved utility of OGM for clinical research applications, including in constitutional genetic disease research, as a result of the new capabilities discussed in this press release; our expectations regarding increased adoption of Saphyr as a result of such improved utility; and our 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; the integration of BioDiscovery into our business; 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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