Bionano Genomics Announces Peer-Reviewed Publication from Johns Hopkins University Outlining a Stepwise Approach to the Adoption of Optical Genome Mapping for Cancer Analysis

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SAN DIEGO, Nov. 22, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (BNGO), provider of optical genome mapping (OGM) solutions on the Saphyr® system and the leading software for genomic data visualization, interpretation and reporting, today announced the publication of a study by Johns Hopkins University in the *Journal of Clinical & Anatomic Pathology* outlining a stepwise approach to adoption of OGM for cancer analysis in the cytogenetics lab.

"This publication is by an outstanding team at Johns Hopkins University and we believe it represents the type of foundational work needed to establish where OGM fits in the cancer analysis lab and the types of subjects and samples that should be analyzed with OGM," commented Dr. Alka Chaubey, chief medical officer of Bionano Genomics. "Knowing how different samples perform with OGM and the variants it detects can allow us to build a paradigm for working with OGM alongside other powerful tools in molecular pathology and cytogenomics as we push forward in our mission to transform the way the world sees the genome."

Conducted as a blinded comparison to a comprehensive collection of tools, this study compared results from OGM to those from whole-genome chromosomal microarrays (CMA) from Illumina, fluorescence *in-situ* hybridization (FISH) probes from Abbott, a targeted panel by next-generation sequencing (NGS) from Illumina, a gene fusion panel by gene expression on the nCounter from NanoString and traditional g-banding by karyotyping. The cohort comprised five different cancer subjects and multiple sample types: four leukemia/lymphoma subjects and one solid tumor subject across three bone marrow samples, one peripheral blood sample and one solid tumor sample (kidney tissue from a Wilm's tumor subject).

The findings by OGM were concordant with those obtained by CMA and NGS for copy number variants (CNVs) and FISH and karyotyping for balanced structural variations (SVs) such as inversions and translocations. Sensitivity compared to CMA was 96% (22/23 CNVs detected) excluding copy neutral loss of heterozygosity calls. Sensitivity compared to karyotyping and FISH was 100% (98/98 loci detected). OGM also revealed substantially more SVs than the traditional methods, including an additional 51 CNVs and 20 SVs. Of the variants revealed by OGM that were not detected by the standard methods, 52% involved genes and 7.7% of them involved known cancer genes. The other 48% were classified as variants of unknown significance (VOUSs). The authors point out that these VOUSs have the potential to play a role in further refining patient diagnosis and identifying novel proteins that could be therapeutic targets.

OGM was also used in the study to provide high resolution analysis of subjects with complex karyotypes exhibiting chromothripsis. Chromothripsis, or chromosome shattering, results in highly complex chromosomal structures that are typically very challenging to unravel by CMA, FISH and karyotyping. OGM provides a more comprehensive view across the genome that targeted methods like FISH cannot give and it has been shown to have a higher resolution than traditional methods as well. Compared to karyotyping, which has a resolution of 5 Mbp, OGM's resolution is 10,000 times higher and compared to CMA, OGM's resolution is 20-100 times higher, depending on the probe density used on the array. The authors used OGM to reveal and characterize chromothripsis (complex genome structures) in leukemia subjects with unprecedented scope and resolution, which they said can be extremely helpful in determining if there are druggable variants present, markers consistent with aggressive disease or disease that's treatment refractory.

The principal conclusions of this publication is that OGM provides an alternative workflow that provides valuable genomic information often with higher resolution than traditional methods without sacrificing sensitivity. OGM is complementary to methods like NGS, which reveal sequence variants, and provides an opportunity to simplify and consolidate workflows for SV analysis by using OGM as an alternative to CMA, FISH and karyotyping.

This publication is available at http://www.clinpathology.com/wp-content/uploads/2021/05/JCAP-6-117.pdf.

About Bionano Genomics

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 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.lineagen.com or www.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 ability to build a paradigm for working with OGM alongside other tools in molecular pathology and cytogenomics; the potential role of VOUSs, including those detected by OGM, in refining patient diagnosis and identifying possible therapeutic targets; and OGM's ability to simplify workflows for SV analysis as compared to CMA, FISH and karyoptyping and be complementary to NGS. 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 or improvements on existing methods, such as CMA, FISH, karyotyping and NGS; failure of future study results to support those demonstrated in the study referenced in this press release; changes in our strategic and commercial plans; inability 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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