

## Study Finds the Combination of Optical Genome Mapping and Short-Read Sequencing Provides a Comprehensive Genome Analysis for Lung Cancer Samples and Enables the Discovery of New Biomarkers

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SAN DIEGO, Sept. 16, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) today announced a study finding the combination of optical genome mapping (OGM) and short-read next-generation sequencing (NGS) provides a comprehensive genome analysis for lung cancer samples and enables the discovery of new biomarkers. This study, appearing in the September 7<sup>th</sup> peer-reviewed issue of *Cell Reports* describes how the combination of OGM and NGS aids in the detection of structural variants (SVs) in non-small cell lung carcinoma (NSCLC), the most common form of lung cancer.

Non-small cell lung carcinoma (NSCLC) is the leading cause of cancer-related death worldwide. Genomic structural variations (SVs), including large indels, inversions, duplications, and translocations, are essential causes of alterations in gene expression and are recognized as hallmarks in tumorigenesis. In this study, recurrently SV-disrupted genes were significantly enriched in cancer-related pathways highlighting the importance of OGM for detecting all classes of SVs. This study utilized short-read NGS and Bionano's OGM to obtain high-technical-confidence somatic SVs to investigate candidate oncogenes in cancer patients. An essential role of high-technical-confidence somatic SVs guided the identification of two new oncogenes (TRIO and SESTD1) in NSCLC.

The authors of this study, Xia *et al.*, claim that NGS and OGM are complementary techniques in oncogene discovery and translational research applications since all somatic variants from 1bp and above can be identified. In addition, the identification of SVs via NGS alone remains challenging because of the nature of the short reads generated by this method. The authors expect to be able to identify more high-technical-confidence SVs and determine more candidate oncogenes of NSCLC or other tumors by using this approach of combining NGS and OGM. The utilization of these two techniques would make them able to more comprehensively quantify the contribution of somatic SVs in cancer as compared to short-read NGS alone.

In addition, the authors confirmed that deletions and inversions have bidirectional influences on gene expression. The analysis by Xia *et al.*, suggests that this bidirectional influence might be the result of independent effects of SVs and open peaks on gene expression. This finding reinforces the importance in being able to accurately detect inversions, which are difficult to detect with NGS but a strength of Bionano's OGM technique on the Saphyr<sup>®</sup> system.

"We believe the combination of NGS and OGM is currently the most comprehensive and cost-effective analysis of the cancer genome," commented Erik Holmlin, PhD, CEO of Bionano Genomics. "This study demonstrates the benefits of combining NGS and OGM data for discovery and translational research in cancer, with the ability to detect from 1bp to >1kb for measuring all classes of structural variants in an integrated approach. We believe this trend will continue across all clinical research applications where the combination of NGS and OGM can provide greater insights for elevating human health and wellness."

This publication is available at https://www.sciencedirect.com/science/article/pii/S2211124721011049

## **About Bionano Genomics**

Bionano is a genome analysis company providing tools and services based on its Saphyr<sup>®</sup> 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. Forwardlooking statements include statements regarding our intentions, beliefs, projections, outlook, analyses or current expectations concerning, among other things: the potential benefits resulting from a combination of OGM and NGS technologies, including comprehensive genome analysis in oncology, and our expectations regarding future trends in clinical research applications of the combined technologies. Each of these forward-looking statements involves risks and uncertainties. Actual results or developments may differ materially from those projected or implied in these forwardlooking 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.

CONTACTS Company Contact: Erik Holmlin, CEO Bionano Genomics, Inc. +1 (858) 888-7610 eholmlin@bionanogenomics.com

Investor Relations: Amy Conrad Juniper Point +1 (858) 366-3243 amy@juniper-point.com

Media Relations: Michael Sullivan Seismic +1 (503) 799-7520 michael@teamseismic.com



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