



Bionano's Saphyr Plays Essential Role in Identifying Three Previously Unknown Genetic Mutation Types in Cancer in Study from Weill Cornell

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Publication in the peer-reviewed journal *Cell* identifies three new distinct classes of structural variations associated with poor outcomes in breast and ovarian cancer, gastrointestinal cancers, and melanomas, helping provide new insights on aggressive tumor growth that could support future development of targeted therapeutics and personalized treatment

SAN DIEGO, Oct. 08, 2020 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) announced today that its Saphyr system played a key role in a research study by Marcin Imielinski, M.D., Ph.D., Assistant Professor of Pathology and Laboratory Medicine at Weill Cornell Medical School, which was published in the peer-reviewed journal *Cell*. The study identified three new distinct classes of structural variations (SVs) in the DNA of thousands of cancer samples across multiple cancer types, and used Saphyr to characterize the precise structure and genomic location of these variations. These previously unknown types of complex rearrangements help explain different mechanisms that enable cancer cells to expand and grow rapidly while simultaneously evading natural defense mechanisms and treatment. With the addition of Bionano's Saphyr data, the study demonstrated that these structural variants generate a large number of fusion proteins and represent therapeutic targets and/or prognostic biomarkers of disease progression.

While cancer genomes traditionally have been studied with a combination of low-resolution cytogenetic methods or with next-generation sequencing (NGS), Bionano's Saphyr instrument provides long-range data that enables a high-resolution, long-range view into the cancer genome and cancer biology. At a presentation providing an update on the study given at the March 31, 2020 Advances in Genome Biology and Technology Conference, Dr. Imielinski explained how short-read data alone is unable to resolve the structure of these newly discovered structural variants because short reads provide a very local view of the genome, and further explained how integrating long-range data generated by Bionano's optical mapping technology was essential to obtaining a complete understanding of these structures, including their biological significance.

As previously announced, a high-profile paper published last month by UCSD Professor Vineet Bafna in *Nature Communications* similarly used a combination of NGS and Bionano's Saphyr data to resolve the structure of extra-chromosomal circular DNA, another type of complex rearrangement of the cancer genome which similarly allows for tumor cells to increase their growth rate while evading natural defenses. In both studies, only the combination of short-read sequencing technology with optical mapping data generated by Saphyr was able to fully resolve these complex structures and provide a comprehensive picture of the cancer genome.

"We are impressed by this important body of work and the ingenuity from Dr. Imielinski's team," said Erik Holmlin, Ph.D., chief executive officer of Bionano Genomics. "By combining NGS with Bionano's genome imaging data, they were able to discover, analyze and fully characterize these novel variants that were so large and complex that they were hidden in plain sight from traditional sequencing methods. We believe these studies help generate a rapid increase in understanding of the complexity of cancer genomes driven by the long-range data that only Saphyr can provide, with a cost and throughput that makes the analysis of large sets of cancer samples possible. Since many of the most effective anti-cancer drugs target specific fusion proteins, we believe the discovery of these novel structural variants and the many fusions they generate has the potential to guide the future development of new therapeutics, better cancer diagnostics and a more efficient use of existing cancer drugs."

[The publication is available at <https://doi.org/10.1016/j.cell.2020.08.006>.](https://doi.org/10.1016/j.cell.2020.08.006)

About Bionano Genomics

Bionano is a genome analysis company providing tools and services based on its Saphyr system to scientists and clinicians conducting genetic research and patient testing, and providing diagnostic testing for those with autism spectrum disorder (ASD) and other neurodevelopmental disabilities through its Lineagen business. Bionano's Saphyr system is a platform for ultra-sensitive and ultra-specific structural variation detection that enables researchers 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, and 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 over nine years and has performed over 65,000 tests for those with neurodevelopmental concerns. For more information, visit www.bionanogenomics.com or www.lineagen.com.

Forward-Looking Statements

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: Bionano's contribution to understandings of extra-chromosomal circular DNA and structural variations in cancer, including its ability to influence improved use of existing cancer drugs or future development of better diagnostics and new therapeutics; Saphyr's capabilities in comparison to and in conjunction with traditional sequencing methods and NGS-based methods; and Saphyr's potential as an essential tool for personalized medicine and furthering understanding of cancer in the medical community. 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; 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, 2019 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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