Day Four of Bionano's Next-Generation Cytogenomics Symposium: Saphyr Solves Genetic Mysteries, Enables Study of Complex Genetic Diseases, Simplifies Muscular Dystrophy Testing

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1. *Saphyr can measure large expansions of disease-causing genomic repeats, impossible with other modern techniques, enabling study of a broad range of currently inaccessible genetic diseases*

2. *Genetic disease cases undiagnosed using existing methods were solved by optical genome mapping*

3. *Assays developed on Saphyr by University of Iowa and KU Leuven for FSHD muscular dystrophy testing provide unambiguous results in half the time, for half the cost of current standard*

4. *Bambino Gesù Hospital in Rome, the largest children's hospital in Italy, is implementing a Saphyr-based assay they developed for routine diagnostic use in children with genetic disorders*

SAN DIEGO, Jan. 15, 2021 (GLOBE NEWSWIRE) -- Bionano Genomics, Inc. (Nasdaq: BNGO) announced that day four of its five-day Next-Generation Cytogenomics Symposium featured seven Saphyr users presenting their results and experiences using the Saphyr® system for optical genome mapping (OGM) to analyze the genomes of patients with genetic diseases largely caused by the expansion or contraction of genomic repeats. Repetitive parts of the genome can expand to tens of thousands of copies and alter the function of genes they are in or near. Long repeats are typically inaccessible by sequencing, while OGM's imaging of long molecules allows for the repeats to be spanned and accurately sized. The presentations by scientists and clinicians from leading hospitals and medical research institutions in Europe and the US showed that Saphyr allows for the study of the many regions of the genome that contain repeats and couldn't be accurately assessed with sequencing technologies or other modern methods.

Dr. Viola Alesi, Bambino Gesù Children's Hospital, Rome, presented a clinical validation study of Saphyr for genetic disease testing. They are validating OGM with Saphyr on 30 samples of children that carry chromosomal rearrangements, copy number variants, repeat expansions or have undiagnosed genetic disorders. She presented multiple cases where Saphyr identified several types of structural variants in the same patient, such as a repeat expansion and a deletion that together caused disease, something they had not been able to do with any other single platform. Dr. Alesi announced that the hospital is introducing the assay they developed on Saphyr for routine diagnostic use in the hospital, specifically for patients where standard cytogenetic testing or gene sequencing was unable to make a diagnosis and are currently validating Saphyr for the analysis of pediatric brain tumors.

Dr. Alex Hoischen, Radboud University, the Netherlands, presented on his research use of OGM with Saphyr to solve undiagnosed genetic disease cases, reporting a preliminary 25% additional diagnostic yield for Saphyr. He discussed the case of a child with severe intellectual disability where sequencing with Illumina and PacBio technologies was unable to find pathogenic variants. Analysis of the family with Saphyr identified a deletion affecting the important brain gene *NSF*. Because the gene is near a duplicated area, the deletion had been invisible to the analysis by sequencing. In a child with a rare and aggressive brain tumor which is typically caused by a mutation in the *SMARCB1* gene, three different sequencing technologies found no causative variants. Analysis with Saphyr identified a large insertion in the gene, which was missed by sequencing. Dr. Hoischen has solved at least 4 other previously unsolved genetic disease cases to be presented in the future. He concluded that Saphyr enables a cytogenetic revolution, especially for leukemia where current testing requires many parallel tests and where deep coverage of the genome with OGM provides a sensitivity that's unmatched by sequencing technologies.

Dr. Birgitt Schuele, Stanford University School of Medicine, used OGM to measure a large repeat expansion in a family with spinocerebellar ataxia type 10, a disease characterized by uncoordinated movements, dementia and/or seizures, as a result of brain atrophy mainly in the cerebellum. With Saphyr she was able to size the very large repeat and detect somatic mosaicism, neither of which has been possible with next-generation sequencing or PCR.

Dr. Mark Corbett, University of Adelaide, Australia and Dr. Christel Depienne, University of Essen, Germany, both study different genes involved in familial adult myoclonic epilepsy (FAME), which is a disease characterized by hand tremors and epilepsy. Dr. Corbett showed how a single Saphyr experiment would have identified the gene that causes this disease, which took him over a decade with other technologies. Dr. Depienne identified the very long pathogenic expansions and concluded that Saphyr allows for the study of the many of parts of the genome that contain repeats and couldn't be assessed until now.

Dr. Aaron Bossler, Director of the Molecular Pathology Lab at the University of Iowa, the largest testing site in the US for Facioscapulohumeral Muscular Dystrophy (FSHD), presented on an assay they developed for FSHD testing on Saphyr that they have implemented in their clinic, replacing a test based southern blot technology. Dr. Bossler summarized that in comparison with southern blot, the Saphyr-based assay they developed provides results for half the cost, in half the time, requires much less sample and much less technician hands-on time, and has better quality control while removing the use of radioactive labeling.

Dr. Jeroen Depreeuw, KU Leuven Hospital, Belgium, discussed their work validating the assay they are developing for FSHD testing. Preliminary results show that OGM with Saphyr was concordant with the gold standard southern blot, and for samples where the standard test was uninformative,

Saphyr data provided unambiguous results. The team hopes to finish developing their assay for FSHD testing on Saphyr and implement it for use in their clinic this spring.

The symposium concludes today with a session starting at 10 am EST dedicated to the analysis of the genomes of COVID-19 patients. The full lineup of speakers and registration access is available at http://bit.ly/3pLPT28

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 research use only 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: the timing and content of the presentations identified in this press release; the effectiveness and utility of Bionano's technology in basic genetic research and clinical settings, and in the contexts and applications contemplated by the presentations identified in this press release; adoption of Saphyr as a standard platform in research and pathology settings; and the execution of Bionano's strategy. 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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