Takeaways from ASHG 2019 in Houston: Users of Bionano's Saphyr System Presented Validation Results for FSHD, Repeat Expansion Disorders and Digital Cytogenetics Among a Growing List of Applications of Saphyr in Human Genetics and Cancer Research

Saphyr provides a platform that streamlines traditional workflows and provides a path to solving previously intractable scientific questions connected to the mechanisms of disease

SAN DIEGO, Thursday, October 24, 2019 -- Bionano Genomics, Inc. (NASDAQ: BNGO), a life sciences instrumentation company that develops and markets Saphyr®, a platform for ultra-sensitive and ultra-specific structural variation detection in genome analysis, today announced the key takeaways from presentations given between October 15-19 by scientists from top institutes and diagnostic companies at the American Society of Human Genetics (ASHG) Annual Meeting and at Bionano's satellite educational event in Houston, TX.

At ASHG and at the satellite education event held prior to the conference and organized by Bionano, clinicians and researchers using the Saphyr system to analyze samples from patients with genetic diseases and cancer presented a series of new findings and validation results that support the adoption of Saphyr as a complement to next-generation sequencing for variant discovery and as a replacement for traditional cytogenetic methods in variant detection for clinical applications.

Alka Chaubey, Ph.D., Head of Cytogenomics at PerkinElmer Genomics summarized the validation of Saphyr technology by PerkinElmer that enabled the development of PerkinElmer's assay for Facioscapulohumeral Dystrophy (FSHD), which PerkinElmer and the University of Iowa developed based on the Bionano EnFocus™ FSHD Analysis tool. Dr. Chaubey presented 100% concordance between the assay and known disease state for publicly available cell lines, and 100% reproducibility among all runs and all FSHD patient samples at multiple test sites and with multiple operators.

Joe Devaney, Ph.D., Associate Director of R&D of diagnostic company GeneDx presented a Saphyr validation study for the detection of the disease-causing variants in FSHD and repeat expansion disorders such as Mytonic Dystrophy 1 and 2. For the 40 samples genotyped for the contraction causing FSHD and the 36 samples genotyped for CNBP gene expansion that causes Mytonic Dystrophy 2, the results generated with Saphyr had a sensitivity, specificity and Positive Predictive Value of 100%.

Dr. Gokce Toruner from the MD Anderson Cancer Center demonstrated 100% concordance between data generated with Saphyr and structural variant data generated with "gold standard" cytogenetic methods in five bone marrow specimens with hematological malignancies. All previously identified cytogenetic abnormalities detected by karyotyping, FISH or microarray analysis were detected by Saphyr. In addition, several novel structural and copy number changes were detected.

Professor Mark Ebbert from the Mayo Clinic used Saphyr to resolve challenging genomic regions implicated in neurodegenerative diseases on 31 brain samples collected post-mortem from patients with ALS, Parkinson's disease, and Alzheimer's disease. One of the causes of ALS is an expansion of a repeat in a gene called C9orf72. While no technology commonly used today for genome analysis has been capable of spanning and measuring the larger repeat expansions of this gene, Dr. Ebbert was able to use the Saphyr system to size a range of expansions from a single brain biopsy, demonstrating a full continuum of mosaicism. In patients with Parkinson's disease, Saphyr resolved the structure of an inverted triplication of an associated gene. In patients with Alzheimer's disease, Saphyr detected structural variants in CR1, an

important gene with a repeat structure that cannot be fully analyzed with next-generation sequencing, and detected inversions in the Tau gene that protect against the disease.

Frances High, MD., Ph.D., from Massachusetts General Hospital for Children at Harvard University presented results of a study on 19 samples from patients with Congenital Diaphragmatic Hernia (CDH), a common and severe structural birth defect that leads to malformation for the developing lungs. Using data from the Saphyr system, Dr. High's team confirmed all high-confidence structural variants that were detected by cytogenetic methods, provided additional higher-resolution detail and elucidated the structure of several, and identified novel likely causative variants. Dr. High announced an upcoming larger study of 50 patient-parent trios, or 150 samples total, to identify novel structural variants that are causative for this disease.

Professor Jennifer Mulle from Emory University School of Medicine used the Saphyr system to study a genetic syndrome characterized by intellectual disability, autism and a 40-fold increased risk for schizophrenia. She identified a high degree of previously undocumented structural variation in the disease region, identified a new gene previously not known to be involved in the disease, and was able to develop a new hypothesis about the disease mechanism from the data generated by Saphyr.

Professors Eric Vilain and Hayk Barseghyan from the George Washington University and Children's National Medical Center presented a number of cases from the Undiagnosed Diseases Network, patients with Disorders of Sex Development, Gitelman's Syndrome, FSHD and Beckwith-Wiedeman syndrome where Bionano was able to provide a molecular diagnosis, identify new likely causative genes, or identify structural variants affecting known or expected disease genes.

Other talks included those by Professor Claudia Carvalho of Baylor College of Medicine, who used data from the Saphyr system to analyze repeat-mediated inversions, which are complex genomic structures that are hard or impossible to resolve with other molecular methods but can predispose to genetic disease; Tina Graves-Lindsay from the McDonnell Genome Institute at Washington University, who corrected structural errors in the official human reference genomes with data generated with Saphyr; and Amir Trabelsi, CEO of Genoox, who announced the new release of a software pipeline that now automatically validates, annotates and classifies Bionano translocation calls from whole genome sequence data.

Erik Holmlin, Ph.D., CEO of Bionano, commented, "The quality of the studies presented and wide variety of applications for Saphyr in genetic disease and cancer research is continuing to increase. Data generated by the Saphyr system are answering difficult questions in complex genetic diseases that have been historically very challenging according to each of the presenters who described their experiences with Saphyr. The validation studies for FSHD, repeat expansion disorders and hematological malignancies are setting the stage for Saphyr to become a routine tool in research and clinical settings. We are grateful to all our customers and collaborators for their inspiration and for their work in showing what Saphyr can do."

More information about Bionano Genomics is available at www.bionanogenomics.com.

About Bionano Genomics

Bionano is a life sciences instrumentation company in the genome analysis space. Bionano develops and markets the Saphyr system, 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 drive the adoption of digital cytogenetics, which is designed to be a more systematic, streamlined and industrialized form of traditional cytogenetics. The Saphyr system comprises an instrument, chip consumables, reagents and a suite of data analysis tools.

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, including among other things, adoption of Saphyr as a routine tool in research and clinical settings and the effectiveness and utility of the Saphyr system in such settings. 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 that our sales, revenue, expense and other financial guidance may not be as expected, as well as risks and uncertainties associated with 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 key clinical studies to demonstrate the effectiveness of our products; 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, 2018 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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