Results Presented at ESHG 2019 Showcase the Advantages of Saphyr over Traditional Cytogenetics Methods for the Detection of Structural Variants in Patients with Genetic Disease and Cancer

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Newly Released Bionano Access Software Provides Access to Structural Variants at 5% Allele Fraction

SAN DIEGO, June 17, 2019 (GLOBE NEWSWIRE) -- 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 announces additional presentations at the European Human Genetics Conference, the annual conference of the European Society of Human Genetics (ESHG), Gothenburg, Sweden. Bionano also announces the release of the latest update of its Bionano Access and Bionano Solve data analysis software and pipelines, which have been critical in advancing the use of Saphyr for applications of Bionano mapping in clinical settings.

On Monday, June 17th, researchers will highlight results in oral presentations and poster presentations of studies comparing Bionano optical mapping to technologies used in traditional cytogenetics workflows used for patient testing in genetic diseases and oncology.

In a poster from the team of Dr. Alex Hoischen at Radboud University Medical Center in the Netherlands entitled, “Next generation cytogenetics in medical genetics with high-resolution optical mapping,” Tuomo Mantere and colleagues describe the comparison of Bionano optical mapping on Saphyr with a suite of traditional cytogenetic methods – fluorescence in situ hybridization (FISH), karyotyping and chromosomal microarray (CMA). In Dr. Hoischen’s presentation as part of a workshop held Sunday, June 16th at the same conference, he provided an up to the minute report on the ongoing study, saying that so far roughly 20% of the hematologic malignancy cases in the study had been completed and that about 35% of the human genetic disease cases were complete. The authors concluded that Bionano optical mapping with Saphyr identified all clinically reported variants with variant allele fraction greater than 10% in patients tested for hematologic malignancies and similarly that all known aberrations were identified in patients tested for constitutional genetic disorders. The authors further reported that in many cases, the complex pathogenic variants identified by Saphyr proved to be more complex than had been previously known based on an understanding developed with lower resolution methods such as FISH, karyotyping and CMA. The finding of additional complexity implies the potential for Saphyr to be used to even further stratify patients in the future, leading to better treatment decisions and patient outcomes.

The identification of pathogenic structural variants in complex cancer genomes like the ones presented by the Radboud team is facilitated by the recent release of Bionano’s new and improved analysis software, Bionano Access and Bionano Solve. Specifically, Bionano’s new Rare Variant Pipeline allows for the detection of all major structural variant types with high sensitivity and precision present at an allele fraction of just five percent, meaning that variants present in just a small fraction of cells in a complex, heterogeneous tumor sample can still be detected – a feat that appears to be nearly impossible with sequencing based analysis methods. Bionano scientists are presenting initial results generated with this pipeline, and detailing the recently released fast isolation protocol for ultra-high molecular weight DNA, Bionano Prep SP, in various presentations at ESHG as well.

In a second study, this one entitled “Evaluation of the Bionano optical mapping technology as a replacement of conventional cytogenetics in a diagnostic setting”, Charlotte Keith and team from Western General Hospital in Edinburgh, Scotland, compared Bionano optical mapping to karyotyping and CMA on benchmark samples, including those containing balanced translocations and large deletions. In one case, the higher resolution of optical mapping resolved the breakpoint of a translocation to being within a gene, and demonstrated that the translocation truncated the gene, potentially altering the function of the gene. The authors report that the successful proof of principle study has cleared the way to a larger validation, which upon successful completion, could potentially enable use of the Bionano Saphyr system in clinical workflows.

Finally, in an oral presentation, Lisanne Vervoort from the Katholieke Universiteit Leuven, Belgium is presenting a study on DiGeorge syndrome, a syndrome typically affecting infants with symptoms including heart defects, developmental delay and frequent infections. Using Bionano optical mapping as part of a large collaboration including with sites in the United States, the team from Leuven have been able to decode the complex, repetitive region of the genome that is responsible for the disease. Furthermore, the authors leveraged data from a large population study of 154 human samples from different ethnic groups to determine the ethnic differences in the frequency of these specific genomic structures to potentially explain ethnic variation in the disease.

Erik Holmlin, PhD, CEO of Bionano, commented, “We are now seeing over and over again that Saphyr has the performance and streamlined workflow to potentially replace the traditional methods in cytogenetics with a digital one based on Bionano optical mapping. The progress in all these studies is just tremendous.”

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.

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