Bionano Genomics' Saphyr® System Used for Highly Accurate Detection of the Genetic Disorder FSHD as an Alternative to Southern Blot

Efforts to Validate Saphyr for Digital Cytogenetics are Accelerating

SAN DIEGO, Feb. 06, 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 the release of a key publication by scientists at Wenzhou Medical University, Wenzhou Central Hospital, The First Hospital of Kunming and Berry Genomics, a leading provider of solutions for pre-natal diagnostic testing in China. The authors used the Bionano Saphyr system to analyze patient samples to obtain highly accurate molecular diagnoses of facioscapulohumeral muscular dystrophy (FSHD) in a multi-generation pedigree. They praised Bionano's "moderate sample requirements and short time frame compared to Southern hybridization" together with its "potential to identify structural variants such as deletions, duplications or rearrangements." FSHD, one of the most prevalent hereditary muscle diseases, is tied to variation in the size of D4Z4 arrays, in which a 3.3 kilo base pair unit on chromosome 4 is repeated multiple times. Southern blot is used to characterize array sizes above and below a threshold level today, but these workflows are slow and cumbersome and can generate results that are difficult to interpret.

Scientists at these leading academic medical centers, together with commercial diagnostic laboratory Berry Genomics, used Bionano genome mapping to correctly characterize the molecular structure of the FSHD locus in the affected individuals of a five-generation pedigree. According to their publication, Bionano's Saphyr enabled the study to identify the founder of the disease within the pedigree as well as a variant of the FSHD1 region involving a duplication of one allele. This variant appears to manifest a clinically milder form of FSHD, suggesting the need to test for it in conjunction with other analysis to provide a more definitive prognosis.

Erik Holmlin, PhD, CEO, of Bionano Genomics, said, "This study is one of the most extensive in FSHD since we first began work in this disease with Johns Hopkins in 2017. The comparison of Bionano genome mapping to existing methods such as Southern blot illustrates how Bionano Saphyr offers an improvement in workflow while providing highly accurate results with the potential to increase clinical performance and utility by readily adding new clinical markers, such as the structural variation tied to a potentially milder form of FSHD described in this study, without modifying the assay or workflow." He added, "Based on the innovations in our instrument, software and sample preparation, efforts to demonstrate and validate Saphyr for digital cytogenetics are accelerating. We expect several studies to begin and others to reach completion in the near future."

About Bionano Genomics
Bionano is a life sciences instrumentation company in the genome analysis space. The Company 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 streamline the study of changes in chromosomes, which is known as cytogenetics. The Saphyr system comprises an instrument, chip consumables, reagents and a suite of data analysis tools.

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