



# Beijing Grandomics Announces Intent to Use the Saphyr System for Molecular Diagnosis of Facioscapulohumeral Muscular Dystrophy (FSHD) in China

Beijing Grandomics using Bionano Saphyr to find Complex Genome Variants in Clinical Applications

BEIJING and ORLANDO, Fla., Oct. 19, 2017 (GLOBE NEWSWIRE) -- At the 2017 annual meeting of the American Society of Human Genetics in Orlando, Depeng Wang, the Chief Executive Officer of Beijing Grandomics announced the company's intent to develop novel assays for genetic diseases in China using Bionano's Saphyr System for genome mapping. Grandomics acquired the first Saphyr earlier this year as they begin to build a large capacity for generating Bionano data.

One of their first indications is facioscapulohumeral muscular dystrophy (FSHD), a progressive and potentially debilitating neuromuscular disease. FSHD, one of the most prevalent hereditary muscle diseases, is tied to variation in the size of the D4Z4 arrays, in which a 3.3 kbp unit on chromosomes 4 and 10 is repeated multiple times, but only permissive alleles with low copy numbers in chromosome 4 are pathogenic. Southern blot is used to characterize array sizes above and below a threshold level and to identify permissive alleles today, but more precise information such as the specific number of D4Z4 repeats and the mosaic level of variation hold diagnostic value that is not captured by current methods, including second or third generation sequencing technologies.

Wang commented that "FSHD is a serious medical condition with a significant unmet need in clinical testing of a large patient population. The Bionano Saphyr System is uniquely suited for detecting the complex structural variations (SVs) associated with FSHD in a way that will improve patient management. We are very pleased to have Bionano's support for this project."

Dr. Cao Han, the Chief Scientific Officer of Bionano Genomics said, "Bionano is very excited about the efforts of Beijing Grandomics to work on genetic diseases starting with FSHD. They are proven as a leading innovator, providing advanced genomics solutions in China that bring long-range genomic information. Their recognition that Saphyr solves the technical unmet need for FSHD is part of a broader global recognition of Bionano's utility for detecting SVs in translational research and clinical applications as well as in *de novo* genome assembly applications for countless plants and animals."

## About Beijing Grandomics Biosciences Ltd

Beijing Grandomics Biosciences Ltd, China's first third-generation sequencing service provider and the first third-generation sequencing based precision medicine company, has adopted the latest Bionano Genomics Saphyr platform, in addition to six PacBio Sequel and six Oxford Nanopore GridION platforms. The company has many years of experience in research and diagnostic development on third generation sequencing technology. In 2015, the company completed the first Chinese reference genome project using third generation sequencing technology. In 2016, the company completed the technology development on diagnosis of repeat expansion disorders, and is currently working on the construction of 10K Chinese structural variability database.

## About Bionano Genomics

Bionano Genomics, Inc. provides the Irys® and Saphyr™ systems for Bionano Genome Mapping, which is the leading solution in physical genome mapping. Bionano mapping offers customers whole genome analysis tools that reveal true genome structure and enable researchers to capture what's missing in their data to advance human, plant and animal genomic research. Bionano genome mapping uses NanoChannel arrays to image DNA at the single molecule level with average single molecule lengths of about 350,000 base pairs, which leads the genomics industry. The long range genomic information obtained with Bionano mapping detects and deciphers structural variations (SVs), which are large, complex DNA segments involving repeats that are often missed by sequencing technologies and which are a leading cause of inaccurate and incomplete genome assembly. As a stand alone tool, Bionano mapping enables the accurate detection of SVs, many of which have been shown to be associated with human disease as well as complex traits in plants and animals. As a complementary tool to next generation sequencing (NGS), Bionano mapping integrates with sequence assemblies to create contiguous hybrid scaffolds for reference quality genome assemblies that reveal the highly informative native structure of the chromosome. Bionano mapping also provides the additional ability to verify, correct and improve a NGS generated genome assembly. Only Bionano provides long range genomic information with the cost efficiency and high throughput to keep up with advances in NGS.

Bionano mapping has been adopted by a growing number of leading institutions around the world, including: National Cancer Institute (NCI), National Institutes of Health (NIH), Wellcome Trust Sanger Institute, BGI, Garvan Institute, Salk Institute, Mount Sinai and Washington University. Investors in the Company include Legend Capital, Domain Associates, Novartis Venture Fund and Monashee Investment Management.

For more information, please visit [www.BionanoGenomics.com](http://www.BionanoGenomics.com)

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