Bionano Genome Mapping Found to Be Superior to Existing Clinical Tests for the Identification of Pathogenic Structural Variants in Duchenne Muscular Dystrophy

Work published in Genome Medicine demonstrates Bionano's ability to accurately detect structural variants in a patient cohort

SAN DIEGO, Nov. 01, 2017 (GLOBE NEWSWIRE) -- In a publication in Genome Medicine, a team of researchers from Children's National Health System (CNHS) and the University of California, Los Angeles (UCLA) led by Dr. Eric Vilain report the first molecular diagnoses of patients with genetic disease using Bionano genome mapping.

The scientists used Bionano Irys and Saphyr systems to identify pathogenic structural variants in a series of patients diagnosed with Duchenne muscular dystrophy (DMD), caused by large deletions, insertions, and inversions disrupting the X-linked dystrophin gene. DMD is a severe degenerative muscle disorder mostly affecting boys, for which there is currently no cure.

In this study, Dr. Vilain's team identified both single and multiple exon deletions up to 250 Kbp in size, a 13 Kbp duplication, and a 5.1 Mbp inversion all disrupting the dystrophin gene of study patients. Large structural variants of this size are typically detected with poor sensitivity using NGS or long-read sequencing. They also successfully identified the carrier status in mothers of the patients. The patient with the large inversion had been diagnosed with DMD through an invasive muscle biopsy, after chromosomal microarray, MLPA, PCR sequencing of all 79 exons, and exome sequencing all failed to detect the pathogenic inversion.

The study authors state in the publication that Bionano mapping "has the capacity to replace both MLPA and chromosomal microarrays in the clinical setting." MLPA is a probe-based assay to detect the deletion or duplication of specific loci in the genome. For this study, the authors cite as key advantages over MLPA, Bionano's ability to also provide order and orientation of structural variants and the ability to detect these variants genome wide. Compared to chromosomal microarray, Dr. Vilain's team discusses Bionano's capability to detect balanced events such as inversions and balanced translocations, as well as much smaller variations completely missed by microarray. Relative to NGS, the authors state that Bionano provides higher sensitivity for large structural variants with better false-positive and false-negative rates. Turnaround time and cost are comparable to the aforementioned tests.

The study concludes that Bionano mapping "is poised to become a new tool in the clinical genetic diagnostic strategy and research due to its ability to sensitively identify large genomic variations."

Dr. Erik Holmlin, CEO of Bionano Genomics, comments, "We are excited by Dr. Vilain's team demonstrating Bionano's ability to correctly identify structural variants in patients with genetic disease. The current diagnostic process proves to be a true odyssey for many of the children and parents hoping to find answers or treatment. Dr. Vilain shows that in some cases a single Bionano test could replace or outperform up to four different tests, which would not only simplify testing, but would also dramatically shorten the search for answers. We are also happy with the illustration in this work of how Bionano provides translational researchers with a tool that sees genome variations that other technologies miss. We can't wait to see how other scientists and physicians, inspired by these incredible results, use Bionano

mapping in their genome studies and analyses.”

The study is published in issue (2017) 9:90 of Genome Medicine,

https://doi.org/10.1186/s13073-017-0479-0

About Bionano Genomics

Bionano Genomics, Inc. offers whole genome analysis tools to better understand the genome and its structure. Its high-throughput system Saphyr builds *de novo* maps of the genome by massively parallel imaging of the longest single DNA molecules in the industry. Bionano genome mapping provides comprehensive structural variation (SV) calls, identifying all types of SVs with sensitivities that far exceed those based on next-generation sequencing. When combined with orthogonal sequencing data, Bionano maps can provide the correct structure, order, and orientation to assemble reference-quality genomes.

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