

Genoox Collaborates with Bionano to Enhance the Detection of Disease Causing Structural Variants in DNA

The Genoox Integrated Platform is Designed to Offer Solutions for Combining Genoox Technology for Aligning and Calling NGS Short-reads with Bionano Genome Mapping Technology to Better Understand Complexities of DNA mutations

CALIFORNIA, April 10, 2018 - Genoox, the healthcare technology company on a mission to make it easier for doctors, clinicians and researchers to run clinical genetic applications and act on genetic sequencing results, announced a collaboration today with Bionano Genomics, a company that offers optical mapping to better understand the genome and its structure. Together, the two companies intend to develop a genome informatics workflow to integrate and annotate Bionano structural variant calls with sequencing data within the Genoox platform.

Based on initial outcome data and the potential to enhance research in the area of rare diseases, the Center for Genetic Medicine Research at Children's National Health System in Washington D.C., which is ranked #1 in the Nation by U.S News for babies and top 10 children's hospital overall, will be the initial user of the Genoox integrated platform. The new technology offers Children's National a single platform for sensitive, accurate detection of structural variations and genetic mutations for rare disease, not previously possible, helping to speed genetic diagnosis for children and families.

Traditional next-generation sequencing (NGS) technology excels in detecting small variant mutations that can cause disease. However, the human genome also contains structural variants (SVs), which account for the largest number of divergent base pairs in the genome. Most cannot be detected by short-read sequencing technologies, which suffer from low sensitivity and high false positive rates. Bionano's Saphyr system combined with its new DLS labeling chemistry and a suite of analysis tools performs much better at calling structural variants of all types than sequencing based methods. By using Genoox technology for aligning short read sequence data to Bionano's SV calls, the high sensitivity of Bionano calls will be combined with the basepair precision of NGS.

The Genoox integrated platform will: identify SV breakpoints to single base pair resolution if matching NGS reads are available; cross-validate the Bionano and NGS variant calls, thereby increasing the calling confidence; enable the detection of a comprehensive set of both SNV and SV mutations on one common platform; and utilize the Genoox platform for automatic processing, annotation, data integration and reporting of results.

"Genoox and Bionano are solving some of the biggest problems in genomics with our new integrated platform," said Amir Trabelsi, co-founder and CEO of Genoox. "To date, NGS technology has been a powerful tool to detect smaller DNA mutations. The Genoox and Bionano collaboration now allows researchers, such as the team at Children's National, to quickly and easily detect smaller, SNV type mutations, as well as larger, structural variants on one software platform. This will empower researchers and clinicians to gain a deeper understanding of the human genome, more accurately assess impact on biological function, and enable personalized medicine and treatment."

Mark Borodkin, Chief Operations Officer of Bionano Genomics, commented: “We are delighted to collaborate with Genoox to provide a new way to integrate Bionano and NGS data. Bionano’s unprecedented ability to discover structural variants has led to broader adoption in human health research. While Bionano’s SV calls are sufficient for many applications, some more complex indications require a tight integration with the smaller NGS-called variants. We believe the Genoox integrated platform with Bionano data will provide an important tool in disease research.”

Genoox and Bionano intend to co-sell and co-market the integrated platform for research and future clinical applications.

About Genoox

Genoox is a genomic analysis company on a mission to make clinical genetic sequencing more accessible and affordable. Using its cloud based platform, Genoox manages the entire genetic sequencing process from raw data collection to the delivery of clinical actionable insights. By automating data interpretation and providing millions of in-house and public data points through a proprietary search engine, Genoox aims to improve patient outcomes with increased accuracy and efficiency, enabling personalized medicine. Users across research, clinical and medical facilities can then securely share complex clinical research using a set of customizable tools developed to analyze data and generate actionable reports.

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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