Bionano and Genoox Launch Integrated Platform for Identification of Structural Variants in DNA

SAN DIEGO, Oct. 17, 2018 (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, and 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 today the launch of the Genoox Integrated Platform for identification of structural variants.

While Next-Generation Sequencing (NGS) made the detection of single nucleotide variants (SNV) affordable and straightforward, identifying structural variants (SVs) from NGS data has proven much harder. Despite large scale efforts, algorithmically inferring SVs has limited sensitivity and typically generates large numbers of false positives. 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 with Bionano's SV calls, the high sensitivity of Bionano calls will be combined with the basepair precision of NGS. The Genoox Integrated Platform further automatically validates and confirms SV calls and integrates and annotates SVs with smaller sequence variants in the same genome. It provides sensitive, accurate detection of structural variation and genetic mutations, not previously possible, helping to speed genetic diagnosis.

The new platform combines raw NGS read data with Bionano-based SV calls from a single patient. Bionano's SV data are used to guide the alignment of NGS reads. Evidence from both technologies is used simultaneously to provide increased breakpoint accuracy and confidence. Detected SVs, copy number variants, indels and single nucleotide variants are then annotated by aggregating data from multiple clinical and population frequency databases, and automated AI-based classification according to ACMG guidelines. A customizable clinical report on all variants is generated, including Bionano-only and NGS-only calls, reducing the complexity of determining and reporting the pathogenicity of genomic variants.

Mark Borodkin, COO of Bionano, commented: "Bionano genome mapping is the gold standard for comprehensive SV detection, but clinical analysis of patient genomes often requires a complete view of the genome. The Genoox Integrated Platform with Bionano data allows researchers and clinicians to detect, annotate and report on all variants; from SNPs to SVs. We believe this platform will provide an important tool in disease research."

"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. "The product launch now allows researchers 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."

Genoox and Bionano will begin to co-sell and co-market the integrated platform immediately.

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 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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