

Bionano Genomics sees encouraging results from study of Saphyr

By Liz Hollis, Staff Writer

The results from a study comparing San Diego-based Bionano Genomics Inc.'s Saphyr to traditional cytogenetic methods for the clinical analysis of leukemia genomes revealed that the system was 100% concordant with the standard of care for the detection of somatic chromosomal abnormalities.

In addition, a team from the Radboud University Medical Center in the Netherlands highlighted that while the focus of the study was on determining the concordance for diagnostically reported variants, the Bionano technology also found novel variants. Specifically, they identified 23 potential gene fusions of which only four were previously observed.

All of this matters because gene fusions are important prognostic markers in cancer and are routinely targeted for drug development. The Bionano study provided several potential biomarkers.

The study

A total of 48 patient samples with a combination of myeloid and lymphoid leukemias were analyzed using standard cytogenetic analysis. All samples had an allele fraction of the pathogenic variants of at least 10%. Thirty-seven samples were considered simple and 11 were labeled as complex, based on the number of large structural abnormalities. When Bionano analyzed all the patient samples, the team identified all previously reported aberrations. In addition, Bionano allowed for a better resolution and a more complete picture of complex aberrations.



Erik Holmlin,
CEO of Bionano
Genomics Inc.

In a diagnostic set-up, the comprehensive analysis of all cytogenetic aberrations requires a combination of techniques, such as karyotyping, fluorescence in situ hybridization and copy-number variation-microarrays. This paper, according to Erik Holmlin, CEO of Bionano, aims to help illustrate how Saphyr can replace four existing tools that give the information needed for patient testing in leukemias and lymphomas. He added that the system could do it in a much more efficient workflow.

“This is a platform that addresses a significant unmet need across genome analysis, which is the ability to, with a degree of



Saphyr system. Credit: Bionano Genomics Inc.

high sensitivity and high specificity, reveal large chromosomal rearrangements ... in the genome,” Holmlin told BioWorld. Holmlin explained that 90% of leukemias and lymphomas are caused by some type of structural rearrangement. “Sequencers, which focus on detecting single nucleotide changes in the genome are unable to detect ... large rearrangements.” The Saphyr was designed for researchers to detect novel chromosomal rearrangements that could be driving a specific disease. It also aims to help cytogenetic labs that are using traditional methods.

“This is a very new tool. It reveals all sorts of information that hasn’t been seen before,” he added. “And this is something that clinicians and researchers are very excited about because they know that it’s possible to further stratify patients who are coming into their clinics. They may say, ‘well, this is AML,’ but now, with the Bionano system, because they’re able to see the standard markers that they’re trained to look for and a whole bunch of markers that may have novel significance, then they are able to really dramatically enhance the ability to manage patients.”

Looking ahead

When asked what’s next for the company, Holmlin noted that there are other studies to validate the system. He added that there is a study in France, as well as one in North America

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involving multiple sites. In terms of next steps, the company hopes to kick off a series of validation studies “that go above and beyond the one that was published.” These would aim to expand the scope of validation. “And then we would expect to see commercial adoption of the platform by labs throughout the U.S. and Europe as a replacement for their traditional tools.”

And it appears to have attracted a lot of interest. The company reported in January that Gaithersburg, Md.-based Genedx Inc. had adopted Saphyr to develop assays that will be the next step toward finding more answers for patients and families with muscular dystrophies, developmental and reproductive disorders.

For his part, Alexander Hoischen, associate professor, genomic

technologies and immuno-genomics at the department of human genetics of RUMC, praised the smooth, quick implementation of the Saphyr system. “Less than a year after training we now have successfully run more than 150 samples, including the 48 leukemia samples which we now present in this publication. Other samples include novel research findings for unsolved rare disease cases and known cytogenetic abnormalities [that] may validate the Saphyr system’s use for constitutional aberrations. We are excited about the very high concordance rate between Bionano technology and current standard of care methods and believe that this technology has the potential to revolutionize cytogenetics in the near future.” The company priced its \$20.6 million IPO Aug. 21, 2018.