BioNano Mapping Technology Achieves Most Contiguous Clone-free Human Genome Assembly to Date

Integrating multiple platforms - the new gold standard?

SAN DIEGO — June 29, 2015—BioNano Genomics, Inc., the leader in genome mapping, with a group of top research institutes, announced today that a collaboration led by the Icahn School of Medicine at Mount Sinai resulted in the creation of the first comprehensive analysis of a diploid human genome using two complementary single DNA molecule methods for sequencing and genome mapping. This study, published today in Nature Methods, provided the comprehensive analysis of the contiguous diploid human genome assembly obtained without using inference-based methods from cloned libraries. Mount Sinai coordinated the activities to sequence, map, and analyze a diploid human genome with the goal to integrate single-molecule sequence data and BioNano genome mapping data. This approach generated a de novo assembled genome that was reference quality and improved upon the contiguity observed from traditional sequencing methods. The combination of BioNano genome mapping and Pacific Biosciences sequencing resulted in an improvement in the contiguity of the initial sequence assembly nearly 30-fold and the initial BioNano genome map assembly nearly 8-fold.

"This is the first study demonstrating that our genome mapping technology and single molecule sequencing technology complement each other to generate a reference quality whole genome assembly with haplotype blocks several hundreds of kilobases long," said Han Cao, Ph.D., founder and chief scientific officer of BioNano Genomics. "This is also the first full de novo assembly of a human genome leveraging intact long native DNA (> 150 kb), without any clone libraries and the artifacts that cloning can introduce."

The researchers' objective was to investigate information sometimes missed with sequencing, such as long range repeats and rearrangements, which are clinically important in complex diseases such as cancer. Since one attribute of BioNano genome mapping is to retain long intact native molecules and accurately detect structural variation, adding this technology to the investigation helped to resolve underlying complex genomic rearrangements, such as chromothripsis, using direct imaging.

"Many large and complex forms of variation are missed by traditional next generation sequencing approaches," said Ali Bashir, Ph.D., Assistant Professor of Genetics and Genomics at the Icahn School of Medicine at Mount Sinai and senior author of the study. "Combining long read sequencing and BioNano genome mapping produces highly contiguous de novo assemblies, enabling unbiased comparison of nearly complete genomes – something we have been trying to do for years."

During the comparison of the generated genome with the current reference genome, the team also found that an underrepresentation of tandem repeats was systematically observed in the human reference genome including an exonic expansion in apoliprotein (LPA). The LPA gene is involved in regulating plasma lipid levels and has been shown to be associated with risk of cardiovascular disease. Quantifying these long 5.6 kb repeats over the span of hundreds of kilobases may enable researchers to assess health risk.

The study, titled, "Assembly and Diploid Architecture of an Individual Human Genome via Single Molecule Technologies," appears in the June 29 issue of Nature Methods and characterizes the genome

NA12878, because it has been sequenced in numerous studies, including the 1000 Genomes Project, and offers multiple opportunities for comparison.

About BioNano Genomics

Headquartered in San Diego, BioNano Genomics is delivering an altogether better way of gaining a fully informed understanding of genomes. The Company's platform provides researchers and clinicians the most comprehensive, organized and actionable picture of a genome with unprecedented insights into how the individual components of genomes are ordered, arranged, and interact with each other. BioNano Genomics works with institutions in life science, translational research, molecular diagnostics and personalized medicine. The Company is supported by private investors and grant funding from genomics programs at federal agencies, including the NIH and NIST-ATP.

www.BioNanoGenomics.com

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BioNano Media Contact
Jessica Yingling Ph.D.
Little Dog Communications Inc.
+1.858.344.8091
jessica@litldog.com