Research Documents Rapid Detection of Structural Variation in a Human Genome Using BioNano's Irys System

SAN DIEGO and SHENZHEN, China — February 9, 2015—**BioNano Genomics**, Inc., the leader in genome mapping, and **BGI,** the world's largest genomics organization, highlight the publication of a peer-reviewed research article and its accompanying data* in *GigaScience*. This article describes the rapid detection of structural variation in a human genome using the high-throughput, cost-effective genome mapping technology of the Irys® System. Structural variations are known to play an important role in human genetic diversity and disease susceptibility. However, comprehensive, efficient and unbiased discovery of structural variations has previously not been possible through next generation sequencing (NGS) and DNA arrays with their inherent technology limitations.

This study showed that the Irys System was able to detect more than 600 structural variations larger than 1kb in a single human genome. Approximately 30 percent of detected structural variations affected coding regions, responsible for making proteins. Proteins participate in virtually every process within cells, suggesting that these structural variations may have a deep impact on human health. The Irys System also accurately mapped the sequence of a virus that had integrated into the genome. The ability to provide this type of information may help inform how virus sequence integration can lead to diseases such as cancer.

"We found that BioNano's Irys System helps overcome the technological issues that have severely limited our understanding of the human genome," said Xun Xu, deputy director at BGI. "In a matter of days and with fewer than three IrysChip®, we were able to collect enough data for *de novo* assembly of a human genome and perform comprehensive structural variation detection without additional technologies or multiple library preparations. BioNano has since improved throughput of the Irys system enabling enough data for human genome *de novo* assembly to be collected in one day on a single IrysChip."

Genome maps built using the Irys System reveal biologically and clinically significant order and orientation of functionally relevant components in complex genomes. This includes genes, promoters, regulatory elements, the length and location of long areas of repeats, as well as viral integration sites.

"The Irys System provides a single, cost-effective technology platform solution to assemble a comprehensive view of a genome and discover and investigate structural variations," said Han Cao, Ph.D., founder and chief scientific officer of BioNano Genomics. "The Irys System enables *de novo* assembly of genomes containing complex, highly variable regions and accurate detection of all types of structural variation, both balanced and imbalanced, within complex heterogeneous samples."

The Irys System has previously been used to map the 4.7-Mb highly variable human major histocompatibility complex (MHC) region and to enable a *de novo* assembly of a 2.1-Mb region in the highly complex genome of *Aegilops tauschii*, one of three progenitor genomes that make up today's wheat.

BGI acquired the Irys System in 2014 to enable comprehensive exploration of structural variation in the human genome and to provide vastly improved assemblies for various organisms that have very complex genomic structure, including those organisms where no reference exists. Together with other

available platforms, BGI aims to provide researchers with the most comprehensive information and comprehensive interpretation.

The article is one of the first articles that are part of *GigaScience's* series *Optical Mapping: New Applications, Advances, and Challenges* (http://www.gigasciencejournal.com/series/OpticalMapping), and is available through this link: http://www.gigasciencejournal.com/content/3/1/34.

*The data for this study, as part of the journal's mission of making published research reproducible and data reusable, are available in the Journal's linked database, *Giga*DB, at http://dx.doi.org/10.5524/100097

About Irys

Irys makes it possible to routinely and accurately detect genomic structural variation and to finish genome assemblies. The fully automated Irys benchtop instrument uses the IrysChip to uncoil and confine long DNA molecules in proprietary Nanochannel Arrays™ where they are uniformly linearized in a highly parallel display for high-resolution, single-molecule imaging. Irys does not employ DNA fragmentation or amplification, which are typical with next-generation sequencing. The result is sequence information over extremely long "reads" ranging from hundreds of kilobases to a megabase, where the sample's valuable structural information is preserved. Irys makes it possible for researchers to directly observe structural variants including replications, deletions, translocations and inversions.

About BGI

BGI was founded in 1999 with the mission to use omics technologies to benefit the human society. The goal of BGI is to make leading-edge genomic science highly accessible through its investment in infrastructure that leverages the best available technology, economies of scale, and expert bioinformatics resources. BGI, which includes both private non-profit genomic research institutes and sequencing application commercial units have established partnerships and collaborations with leading academic and government research institutions as well as global biotechnology and pharmaceutical companies, supporting a variety of disease, agricultural, environmental, and related applications. BGI has achieved a proven track record of excellence, delivering results with high efficiency and accuracy for innovative, high-profile research which has generated over 900 publications in top-tier scientific journals. For more information about BGI, please visit www.genomics.cn.

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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