



BioNano Announces Adoption of the Popular VCF Format

Irys Data Integrated by Industry Leading Visualization Tools for Genomic Discovery

SAN DIEGO, CA – June 7, 2016 – BioNano Genomics, the leader in physical genome mapping, today announced two major advances in the integration of data from the Irys® System into the industry’s standard formats and visualization tools. The first advance is the adoption of the Variant Call Format (VCF v4.3) to represent Structural Variation (SV) calls from the IrysSolve™ pipeline. The second is that data generated by Irys are now supported by the industry’s leading genomics visualization software applications. The compatibility of Irys data with the VCF format and with commonly used visual software applications empowers researchers to fully take advantage of next-generation mapping (NGM) data using familiar tools and data formats.

Originally developed for the 1000 Genomes Project, the VCF format offers researchers a standard for storing the most prevalent types of sequence variations together with rich annotations.

“As we continue to refine our stand-alone SV-calling capabilities, the adoption of the VCF standard has allowed us to share BioNano’s novel SVs with researchers looking to incorporate the Irys datatype into their workflows. BioNano has already provided SV calls in the VCF format to the 1000 Genomes Project and we will be releasing a tool to the community to convert SVs into the VCF format shortly,” said Mark Borodkin, Vice President of Systems Development at BioNano Genomics.

Additionally, third-party genomics visualization developers have taken the initiative to support the visualization of data generated by Irys due to the growing number of leading genomic researchers that use and have adopted NGM. The list of third-party genomics visualization tools supporting NGM data now includes:

- Anno-J Annotation Browsing 2.0 by The Salk institute
- gEVAL Genome Evaluation Browser by Wellcome Trust Sanger Institute
- IGV Integrative Genomics Viewer by The Broad Institute
- JBrowse Genome Browser by University of California Berkeley – [BioNano Plugin Available](#)

William Chow, Lead Developer of gEVAL and Senior Bioinformatician at Wellcome Trust Sanger Institute said, “BioNano genome maps have become an important dataset to add to our repertoire of analyses in gEVAL for identifying and improving complex genomic regions as part of our genome assembly curation efforts. We have and are still using the data to aid in resolving issues ranging from gap closure to capturing misassemblies or variations in human, mouse and zebrafish reference genomes.”

About BioNano Genomics

BioNano Genomics, Inc., the leader in next-generation mapping (NGM), provides customers with genome analysis tools that advance human, plant, and animal genomics and accelerate the development of clinical diagnostics. The Company’s Irys® System uses NanoChannel arrays integrated within the IrysChip® to image genomes at the single-molecule level with average single-molecule lengths of about 350,000 base pairs, which leads the industry. The long-range genomic information obtained with the Irys System helps decipher large, complex DNA repeats, which are the primary cause of inaccurate and incomplete genome assembly.

On its own, next-generation mapping with the Irys System enables detection of structural variants, many of which have been shown to be associated with human disease as well as complex traits in plants and animals. As a companion to next-generation sequencing, next-generation mapping with the Irys System

integrates with sequence assemblies to create contiguous hybrid scaffolds that reveal the highly-informative native structure of the chromosome.

Only BioNano Genomics provides long-range genomic information with the cost-efficiency and throughput to keep up with advances in next-generation sequencing.

The Irys System has been adopted by a growing number of leading institutions around the world, including: National Cancer Institute (NCI), National Institutes of Health (NIH), Wellcome Trust Sanger Institute, BGI, Garvan Institute, Salk Institute, McDonnell Genome Institute of Washington University, and University of California, Davis. Investors in the Company include Domain Associates, Legend Capital, Novartis Venture Fund and Monashee Investment Management.

For more information, please visit us at www.BioNanoGenomics.com.

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