BioNano Releases Updated Irys® Data Solutions for Improved Structural Variation Analysis in Human Samples

Irys® currently the only platform enabling translational research through large SV discovery

Company to discuss new solution at Festival of Genomics in Boston this week

SAN DIEGO, CA – June 27, 2016 – BioNano Genomics, the leader in physical genome mapping, today announced the release of an update to its Irys® Data Solutions to improve Structural Variation (SV) analysis in human samples.

The Irys Data Solutions is a powerful suite of software and hardware solutions: IrysSolve® hosts the data analysis pipeline; IrysView® visualizes genome map information; and IrysSolve® Compute™ is a server solution optimized to run the IrysSolve software.

In this new version, IrysSolve v2.1 enables better understanding of and improves sensitivity to the diploid nature of human genomes, enabling insights into complex diseases, including Mendelian diseases such as sickle-cell anemia and cystic fibrosis.

IrysSolve® v2.1 includes the following features:

- A new automated haplotype-aware assembler that separates allelic differences in the generated maps;
- A new SV calling algorithm able to quickly and accurately determine individual instances of both homozygous and heterozygous structural variation within a human sample, whether through *de novo* or reference genome mapping.

BioNano will be discussing this update at the Festival of Genomics 2016, being held June 27-29 in Boston, MA. Visit **BioNano Booth #234** on the exhibit floor to learn more. The Company has also been invited to participate in the Festival's **Live Lab**, hosted by Microsoft, to demonstrate the role of the Irys® System within a full genomics lab workflow.

The advantages of this IrysSolve update give researchers the ability to identify insertions and deletions of large sizes in genomic material better than any other solution in the market, including karyotype testing, microarrays and FISH. Compared to previous methods which required more manual curation, this update provides a faster, more efficient, accurate, and cost-effective approach for detecting genomic markers of disease. This ultimately, allows for the acceleration of genomic discovery and translational medicine application.

Recently, samples from a 1000 Genome Project study, which initially demonstrated a seven-fold improvement in SV detection using next-generation mapping (NGM) compared to next-generation sequencing (NGS) and were published in the journal *Genetics*, were re-analyzed using the new IrysSolve update.

Dr. Pui Kwok, M.D., Ph.D., University of California in San Francisco's Cardiovascular Research Institute/Institute for Human Genetics, commented, "The new ability to call for insertions and deletions on human diploid samples will make the adoption of the Irys System much easier than before. Following up on the previous work that my group published in the January edition of the journal *Genetics*, I was very impressed to see how those findings, which took over six months to generate, can now be generated in a

matter of a few hours. Over 90% of the published calls from this extensive study were called in a fully automated way. In addition, the new pipeline was able to detect four times more SV's that were not detected previously. Validation for those is on-going and we will be sharing those findings shortly. I am also especially impressed by the ability of the software to identify heterozygous insertions and deletions with a sensitivity of over 80%. Typically, NGS barely identifies 40-50% of heterozygous insertions and deletions. As with the previous study, Irys NGM's ability to detect more SV's than NGS makes this platform invaluable for human genetics research. This new software opens the potential for this platform to play a major role in accelerating population studies across the globe."

Erik Holmlin, Ph.D., President and Chief Executive Officer of BioNano Genomics, added, "Our newest NGM advancement to the Irys® System provides translational researchers a stand-alone method, that is both rapid and cost-effective, to call large heterozygous SV's with unrivaled heterozygous sensitivity for insertions and deletions from 1kbp in size and larger. This accuracy generated by IrysSolve® v2.1 is unparalleled compared to other solutions, and is a major step forward in being able to identify existing and discover new SV biomarkers that cause human genetic disorders. BioNano is committed to equipping researchers in the human genetics space with powerful NGM tools, such as our ability to export data into the standard VCF industry format, to generate discoveries in the lab and apply them for further understanding of complex genetic disorders in humans. We look forward to presenting our latest advancements at the Festival of Genomics and further partnering with labs to offer this essential SV detection technology."

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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