Bionano Genomics Announces Global Launch of Saphyr™, its Newest and Most Advanced System for Genome Mapping and Structural Variation Analysis

Saphyr brings comprehensive structural variation detection with industry-leading sensitivity, specificity at high speed and high throughput

To be showcased at the AGBT General Meeting this week

SAN DIEGO, CA – February 13, 2017 – Bionano Genomics, a company focused on genome structure analysis, today announced the global commercial launch of Saphyr™, its newest and most advanced system for genome mapping. Saphyr is a high-speed, high-throughput platform that delivers unparalleled sensitivity and specificity in structural variation (SV) detection, genome assembly contiguities up to 100-fold longer than those of short-read sequence assemblies and the structural accuracy to correct sequencing-based errors. Saphyr's primary applications in human genomics include SV discovery for translational and clinical research including undiagnosed genetic disorders, gene discovery and therapy development, and cancer; in non-human genomics, applications include selective breeding, evolutionary biology and reference-quality genome assembly.

Saphyr is based on Bionano's next-generation mapping (NGM), which is the combination of its proprietary NanoChannel arrays with optical genome mapping. Saphyr performs NGM at speeds and throughputs that make it feasible to study large populations and highly complex genomes in a fraction of the time and with substantially fewer consumables than were required with earlier generations, making it a very efficient and cost-effective solution for NGM. Saphyr also improves on the data quality of prior systems. Finally, Saphyr is significantly easier to use than earlier generations, with substantial reductions in the hands-on time required to run a chip and with adaptive loading, an automated routine that optimizes run conditions for each sample to maximize throughput.

Erik Holmlin, Ph.D., CEO of Bionano, commented, "We believe Saphyr offers a quantum leap forward in the ability of researchers to make genome structure analysis a cornerstone of their studies. The value and utility of our NGM for SV detection and genome finishing is increasingly recognized, but many potential users were seeing speed and throughput as particularly onerous bottlenecks when they sought to apply NGM to population-scale studies or to analysis of highly complex genomes such as cancer or plants. Saphyr shatters these bottlenecks as a tool that reliably and efficiently uncovers genomic information that lies in the blind spots of other technologies. Saphyr opens the door to uncovering the genetic basis of many traits, especially drivers at the root of serious human health conditions."

Eric Vilain, M.D., Ph.D., human geneticist and physician, became the first Saphyr user. He is seeking to determine the anomalies of study participants who have undiagnosed genetic disorders.

Dr. Vilain said, "Saphyr offers the possibility to identify disease-causing structural variants in parts of the genome that are inaccessible by sequencing. We used Irys®, Bionano's previous generation, in the past; however, the throughput increase provided by Saphyr allows us to map structural variants in one patient per day, enabling the potential to diagnose many more diseases in a high-throughput manner. We are excited to present the first results using Saphyr from a study of patients with undiagnosed genetic disorders at AGBT tomorrow evening."

Additional prominent researchers are adopting Saphyr:

- **Erich Jarvis, Ph.D., Rockefeller University Professor and Howard Hughes Medical Institute Investigator,** and **Olivier Fedrigo, Ph.D. at Rockefeller**, plan to use Saphyr to develop reference quality genome assemblies in connection with their vertebrate G10K and bird B10K collaborative efforts;
- **James Broach, Ph.D., Director of the Penn State Institute for Personalized Medicine at the Penn State College of Medicine,** will apply Saphyr to studies of thyroid cancer and other human clinical studies;
- **Berry Genomics in Beijing, China**, a leader in non-invasive prenatal testing (NIPT), is using two Saphyr systems for applications in human translational and clinical research in the effort to develop molecular diagnostic tools.

Saphyr Features

The Saphyr System features a suite of tools for fast and comprehensive analysis of human and other species complex genome structure. The Saphyr instrument images the genome using fast, single-fluor sensitivity and high resolution scanning. It applies state-of-the-art, machine learning based DNA loading of the new, high-throughput Saphyr™ Chip. The suite allows researchers to perform deep heterozygous SV discovery in a single run, using only one Saphyr Chip. The Saphyr instrument requires less than 3 minutes hands-on instrument time per chip, and automatically guides and monitors the loading of the DNA sample, ensuring seamless image processing and reducing manual intervention.

To simplify the SV discovery workflow, Saphyr comes with the new Bionano Access™ analysis and visualization software. Bionano Access is a web-enabled solution that researchers can use to easily set-up experiments, start runs, monitor data quality metrics in real-time and automatically start *de novo* assemblies and SV discovery analysis when enough data are collected. Visualization of the results through Bionano Access is especially fast and feature-rich and allows researchers to visualize and interact with complex SVs. Access also comes with a powerful trio analysis application that sifts through a filtered set of uncommon SVs in a family to identify inherited and *de novo* SVs and allows these to be visualized and exported in a dbVar-compliant VCF file for downstream analysis.

The Bionano Solve 3.0 assembly pipeline within Access allows users to run SV analysis or hybrid scaffolding. Bionano Solve 3.0 allows for unprecedented sensitivity and precision to detect large SVs, as described in the recent bioRxiv paper demonstrating that the pipeline automatically detects all insertions and deletions larger than 1 kilobasepair (kbp) with more than 90% sensitivity and detects translocations with 98% sensitivity. Irys customers will also benefit from the Bionano Access software through a free upgrade available this quarter.

Saphyr at AGBT General Meeting

Bionano will showcase Saphyr in the **Saphyr Room (#213, 2nd Floor)** at the Advances in Genome Biology and Technology (AGBT) General Meeting this week.

Research focusing on NGM's ability to improve genome assembly and SV detection will be presented, with key opinion leaders available in the Saphyr Room to discuss their experience and research using NGM in prostate cancer, undiagnosed genetic disorders and muscular dystrophy.

More information on Saphyr is available at www.bionanogenomics.com/AGBT2017 and a full list of Bionano activities at AGBT is available here.

About Bionano Genomics®

Bionano Genomics, Inc. provides the Irys and Saphyr systems for next-generation mapping (NGM), which is the leading solution in physical genome mapping. NGM offers customers whole genome analysis tools that reveal true genome structure and enabling researchers to capture what's missing in their data to advance human, plant and animal genomic research. NGM uses NanoChannel arrays to image DNA at the single-molecule level with average single-molecule lengths of about 350,000 base pairs, which leads the genomics industry. The long-range genomic information obtained with NGM detects and deciphers structural variations (SVs), which are large, complex DNA segments involving repeats that are often missed by sequencing technologies and which are a leading cause of inaccurate and incomplete genome assembly.

As a stand-alone tool, NGM enables the accurate detection of SVs, many of which have been shown to be associated with human disease as well as complex traits in plants and animals. As a complementary tool to next-generation sequencing (NGS), NGM integrates with sequence assemblies to create contiguous hybrid scaffolds for reference-quality genome assemblies that reveal the highly informative native structure of the chromosome. NGM also provides the additional ability to verify, correct and improve a NGS-generated genome assembly.

Only Bionano provides long-range genomic information with the cost-efficiency and high throughput to keep up with advances in NGS.

NGM 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, Mount Sinai and Washington University. Investors in the Company include Domain Associates, Legend Capital, Novartis Venture Fund and Monashee Investment Management.

For more information, please visit www.BionanoGenomics.com.

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