Bionano Genomics Announces Global Launch of its New Direct Labeling Chemistry for Genome Mapping and Structural Variation Analysis

Direct Labeling chemistry provides chromosome arm length maps and improves SV detection resolution down to 500 bp for as little as $500 per genome

To be showcased at the AGBT General Meeting this week

SAN DIEGO, Feb. 12, 2018 (GLOBE NEWSWIRE) -- Bionano Genomics, the genome mapping company, today announced the global commercial launch of new chemistry for sequence motif labeling called Direct Label and Stain (DLS). DLS is a non-destructive labeling chemistry that improves every aspect of Bionano genome mapping. When used in the Bionano workflow, DLS enables unprecedented sensitivity and resolution for structural variant discoveries and the most accurate and contiguous genome assemblies.

Prior to the DLS chemistry, Bionano offered its customers NLRS (nick, label, repair and stain) kits, which use nicking endonucleases to make sequence-specific nicks, where fluorescently labeled nucleotides were subsequently incorporated, followed by a ligation reaction to repair the nicks. The NLRS process is highly robust and specific, but it introduces systematic double-stranded breaks that limited the contiguity of Bionano maps. Overcoming these systematic breaks often required the use of two different nicking enzymes to create two separate sequence motif maps that could be overlaid in analysis. Because DLS has no destructive steps in the workflow, the systematic molecule breaks are eliminated. Therefore, the DLS protocol is substantially streamlined. It involves fewer steps and fewer enzymes and a single sequence motif map is usually sufficient to obtain the same or better results seen with the NLRS chemistry, effectively doubling the throughput of the Saphyr system.

The DLS chemistry has been evaluated and hardened by Saphyr users in an extensive early access program, which revealed the robustness and significant additional value of the new approach. For example, genome maps generated using DLS are 50-fold longer on average than when using the NLRS chemistry. Chromosome arms and full chromosomes are often assembled in single maps, reducing the need for other chromosome-scale scaffolding methods. Sequence assemblies scaffolded with DLS maps reach the highest contiguity and accuracy reported to date. DLS also improves sensitivity for all structural variant calls, yielding robust detection of heterozygous insertions and deletions as small as 500 bp, which is well below the previous limit of 1kb. DLS also improves sensitivity to larger balanced and unbalanced structural variants, which had been impacted by the systematic strand breaks associated with high-density nickase-based labeling.

Dr. Jim Broach, Director of the Penn State Institute for Personalized Medicine, is one of the first DLS users and participants in the DLS early-access program. Dr. Broach is using Bionano mapping to identify structural rearrangements in cancer cell lines, and samples from patients with pediatric or adult leukemia. He believes DLS is a game changer for its ability to assemble entire chromosome arms of leukemia genomes in single maps, improving the visualization of complex and large events.

Among the dozen prominent researchers who have participated in the DLS early-access program were Erich Jarvis, Ph.D., Rockefeller University Professor and Howard Hughes Medical Institute Investigator, and Olivier Fedrigo, Ph.D. at Rockefeller. They have used DLS and Saphyr to develop reference quality genome assemblies in connection with their vertebrate G10K and bird B10K collaborative efforts. Prof. Pui-Yan Kwok at the University of California San Francisco and Academia Sinica in Taipei, Taiwan, is using DLS' improved SV discovery to study the genomes of a large cohort of patients with genetic disease and their parents.

Erik Holmlin, Ph.D., CEO of Bionano, commented, "Last year we introduced Saphyr addressing the need for high-throughput users. With the new Direct labeling chemistry we improve every aspect of Bionano genome mapping. Saphyr has seen incredible adoption, with one instrument sold every ten days since launch. With DLS, Saphyr gets twice the sample throughput for a fraction of the cost per genome. With a streamlined protocol, significantly longer molecules and chromosome arm length maps, DLS helps researchers understand the structure of the genome like never before.

"We are incredibly excited about DLS' ability to detect structural variants down to 500 bp in size. This breakthrough makes Bionano mapping the perfect complement to exome or whole genome sequencing. High throughput users can now afford to add high coverage Bionano data to every genome sequenced at just $500 per human genome mapped."

The primary applications of Saphyr in human genomics include SV discovery for translational and clinical research including cancer, undiagnosed genetic disorders, gene discovery and therapy development; in non-human genomics, applications include selective breeding, trait development, evolutionary biology and reference-quality genome assembly. The DLS chemistry makes mapping with Saphyr more affordable and attainable than ever and it allows high-volume users to map a human genome for $500. DLS kits are available for order now.

DLS at AGBT General Meeting

Data generated using DLS will be showcased in oral and poster presentations at the Advances in Genome Biology and Technology (AGBT) General Meeting this week.

Posters presented will focus on DLS' ability to detect and visualize SVs in cancer and genetic disease, and on *de novo* genome assembly.

More information on DLS is available at www.bionanogenomics.com/DLS

Details on Bionano's presence at AGBT can be found at https://bionanogenomics.com/bionano-university/articles/agbt/

About Bionano Genomics

Bionano Genomics, Inc. offers whole genome analysis tools to better understand the genome and its structure. Its high-throughput system Saphyr builds *de novo* maps of the genome by massively parallel imaging of the longest single DNA molecules in the industry. Bionano genome mapping provides comprehensive structural variation (SV) calls, identifying all types of SVs with sensitivities that far exceed those based on next-generation sequencing. When combined with orthogonal sequencing data, Bionano maps can provide the correct structure, order, and orientation to assemble reference-quality genomes.

For more information, please visit www.BionanoGenomics.com

Bionano Genomics Media Contact
The Ruth Group
Kirsten Thomas
kthomas@theruthgroup.com

(508)280-6592