Combination of Bionano Genomics and 10x Genomics data produces high quality mammalian genome at low cost

Annotated assembly of the Hawaiian monk seal genome released by NCBI

July 25, 2017 – SAN DIEGO, CA and BETHESDA, MD – NCBI, the National Center for Biotechnology Information, has released the annotated genome assembly of the monk seal, *Neomonachus schauinslandi*. Bionano maps have been used to scaffold the majority of recent reference quality genomes. While these reference genomes involved costly sequencing technologies, the monk seal assembly was produced with a cost effective combination of Bionano, Illumina and 10x Genomics.

The final assembly produced a scaffold N50 of 29.65 Mbp, representing a 215x improvement over an assembly produced with Illumina short-read sequencing alone. The longest scaffold of the combined Bionano and 10x Genomics assembly is 84.77 Mbp, approaching full-chromosome scale. Bionano maps improved the contiguity ensuring correct order and orientation, sized gaps and corrected gap sizes in the 10X Genomics *de novo* assembly. As an orthogonal, non-sequencing based assembly method, Bionano maps validated the assembly which is not possible with other sequence-based scaffolding technologies.

The quality of the Bionano scaffolding was tested by translating the seal gene sequences and aligning the positions of matching proteins to both human and dog databases. Extremely high alignments genome wide showed conserved synteny with human and other mammals. 98% of the total estimated genome length was accounted for by the 170 Bionano hybrid scaffolds, and 99% of the BUSCO conserved gene set were within the 170 scaffolds, indicating that the hybrid scaffolds were of high quality.

In the related bioRxiv publication, the authors describe that despite significant reduction in sequencing costs, the assembly of high quality genomes remains difficult and expensive. With a material cost of less than $15,000 and a total project time of a few weeks, this monk seal assembly is of the same high quality as other recently released genome at a small fraction of the time and cost. Combining the two orthogonal methods of Chromium Linked-Reads with Bionano optical maps can make the assembly of high quality genomes routine and significantly improve our understanding of comparative genome biology while reducing costs.

Erik Holmlin, Ph.D., CEO of Bionano, commented, “This genome assembly demonstrates the power of combining Bionano genome mapping with 10x Genomics’ Linked-Read sequencing to create a reference-quality assembly. Bionano optical maps have been used to improve the quality of genome assemblies obtained using technology provided by Illumina, PacBio, Oxford Nanopore, Dovetail, NRGene and 10x Genomics. Our technology is sequencing platform agnostic. No matter how you generate your sequence, we improve contiguity, order and orientation while correcting assembly errors. The particular combination of Bionano with linked-read sequencing is an extremely cost-effective and time-saving combination, which can become the standard for large scale reference genome projects.”

The publication describing the assembly is available on the pre-print server bioRxiv (http://www.biorxiv.org/content/early/2017/04/19/128348). The annotated assembly is available from NCBI at https://www.ncbi.nlm.nih.gov/genome/annotation_euk/Neomonachus_schauinslandi/100/

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.

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