Integrated GENOMICS Cores

The Saphyr

Bionano's Third Generation Optical Mapping Solution

The HTSF is proud to announce the addition of the Saphyr, an investment in the future of genome research. The Saphyr is BioNano's third generation optical mapping solution and delivers rapid, high throughput, long range genome mapping with structural variation detection capabilities critical for genome research.

Overview

The Saphyr is BioNano's latest high-speed, high-throughput genome mapping solution, with the capability of analyzing structural variations efficiently and accurately. This new system uses a unique high-throughput optical mapping technology to detect structural variants missed by next-generation sequencing (NGS) systems. The speed and flexibility prove that the Saphyr is the answer for both human and translational research projects.

Through powerful cutting-edge technology, the Saphyr offers new opportunities in genome research at prices that can fit individual project's research goals. Although best suited for human genome research, the Saphyr offers opportunities in both animal and plant genome research, thus allowing researchers from diverse scientific backgrounds to reap the benefits of faster turnaround time, flexible performance and improved yield.

Solutions to Current Problems

Structural Variation

The Saphyr fills a gap in genome research by providing rapid, high-throughput, long-range genome mapping with unrivaled abilities in structural variation detection. The Saphyr provides extremely long images while detecting large-scale structural variations overlooked by previous NGS systems. The Saphyr identifies structural variations ranging from 500 bp to megabase pairs, offers assembly and discovery algorithms, and calls inversions, repeats, and copy number variant at the below sensitivities.

- 99% sensitivity for homozygous insertions/deletions larger than 500 base pairs
- 95% sensitivity for heterozygous insertions/deletions larger than 500 base pairs
- 95% sensitivity for balanced and unbalanced translocations larger than 50,000 base pairs
- 99% sensitivity for inversions larger than 30,000 base pairs
- 97% sensitivity for duplications larger than 30,000 base pairs
- 97% sensitivity for copy number variants larger than 500,000 base pairs

The Saphyr provides these services with a false positive rate of less than 2% (BioNano Website).

Flexible Mapping Options

The Saphyr can generate high-resolution physical genome maps with greater speed than previous systems and can run most genomes in one day. Due to the Saphyr's dual-flowcell design, two independent maps can be created from one sample, with two enzymes, and the data can be combined. The generation of two maps provides the opportunity to correct assembly errors with greater accuracy. Additionally, the dual-flowcell design can accommodate unrelated samples, which allows researchers to map two separate genomes in 24 hours.

Implications for Human Genome Research

While improvements in sequencing technology have allowed for remarkable advancement in the detection of single nucleotide changes, standard methodologies have been ineffective in analyzing larger structural variations and typically fail in repetitive regions.

The Saphyr is best suited for researchers who want to create de novo maps of genome structure. The Saphyr can also detect large structural variants, such as deletions, duplications, inversions, and translocations, found in cancers and genetic disorders. Published studies report that, the Saphyr correctly identified genomic rearrangements in prostate cancer and was used to diagnose patients with Duchenne Muscular Dystrophy. Although the Saphyr can be used for both animal and plant focused research, it is best suited for human research.

Summary

The Saphyr offers a more accurate alternative to traditional NGS systems and a rapid turnaround time, in addition to more flexible mapping options that can fit individual project's research goals. This new technology provides new abilities to explore a diverse variety of experimental design options, ranging from diagnosing genetic disorders to examining the genomic integrity of cell lines.

To learn more about the Saphyr, including sample suitability, preparation, and pricing, please contact:

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