

## saphyr<sup>™</sup>

Reveal what's missing in your  
genome research with Saphyr  
optical genome maps.



## INTRODUCING SAPHYR™

Saphyr, Bionano's high-speed, high-throughput genome mapping solution, detects and analyzes large structural variations with exceptional sensitivity and specificity to reveal the true structure of any genome. Saphyr's breakthrough speed and throughput combined with unmatched structural variation sensitivity make it the ideal solution for human and translational research applications.



Saphyr is based on Bionano's next-generation mapping (NGM), which is the combination of proprietary NanoChannel arrays with optical genome mapping to image extremely long, high molecular weight DNA in its native state. This technology allows for unparalleled structural variation sensitivity, genome assembly contiguity up to 100 times that of short-read sequencing alone and the accuracy to correct sequencing-based assembly errors.

### STRUCTURAL VARIATION DISCOVERY PLATFORM

#### Resolve large structural variations missed by next-generation sequencing (NGS) systems

Large structural variations are responsible for many diseases and conditions, including cancers and developmental disorders. Saphyr detects structural variations ranging from 1,000 bp to megabase pairs in length and offers assembly and discovery algorithms that far outperform sequencing-based technologies in sensitivity.

- **99% sensitivity** for large homozygous insertions and deletions
- **87% sensitivity** for large heterozygous insertions and deletions
- **98% sensitivity** for translocations

Saphyr provides this performance with a false positive rate of less than 3%. Saphyr also calls inversions, repeats, copy number variants and complex rearrangements.

## ENHANCED SPEED AND THROUGHPUT

### Rapid optical mapping ideal for human research applications

Saphyr features enhanced optics with adaptive loading of DNA utilizing machine learning. The Saphyr Instrument and high-capacity Saphyr Chip™ combine to deliver genome maps at the speed and scale your research demands.

- Long molecules from 100,000 bp to megabase pairs
- Guaranteed 640 Gbp throughput per Saphyr Chip per day for human samples for deep structural variant discovery (320 Gbp per flowcell of molecules larger than 150 kbp)
- Sample to structural variation calls or genome scaffolding in as little as 5 days



## SIMPLIFIED WORKFLOW

### Automation features and intelligent sample preparation simplify the process

Saphyr offers automated features that minimize hands-on time while upgradable components ensure Saphyr delivers value over the long term.

- Requires less than 3 minutes hands-on instrument time per chip
- Automatic optimization of run conditions based on sample characteristics to maximize throughput
- Saphyr Chip Clip protects sample integrity and eliminates the need for instrument wash cycles between runs
- Near-hermetically-sealed carousel auto-loads up to 12 Saphyr Chips (coming late 2017)

## WIDE RANGE OF APPLICATIONS

### Explore a variety of experiment design options

Apply high-resolution physical genome maps to understand genome structure in many areas of research.

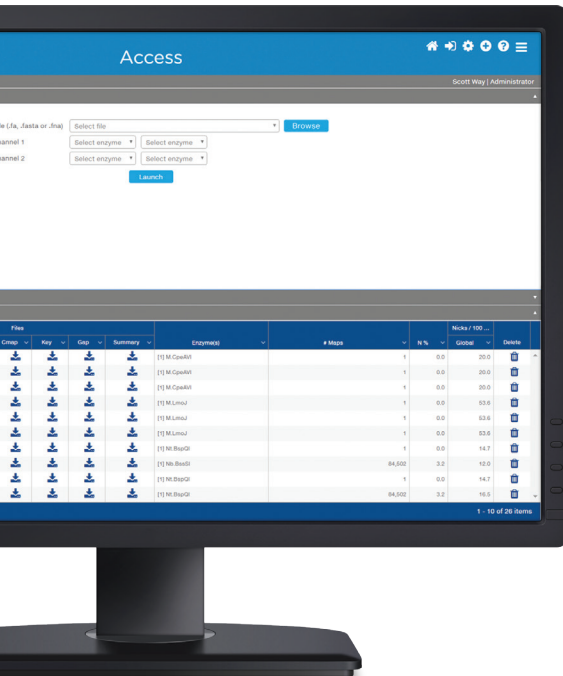
- **Undiagnosed genetic disorders** – close the diagnosis gap by detecting large structural events missed by NGS
- **Gene discovery and therapy development** – identify genes of interest, their locations and how structural variations impact them to inform effective therapy development
- **Cancer** – detect and visualize large rearrangements occurring in cancer genomes
- **Cell line studies** – monitor genomic integrity of cell lines and off-target effects of genetic engineering
- **Selective breeding** – identify areas of biological interest for achieving desirable traits in livestock or crops
- **Evolutionary biology** – see the complete picture of how genomes have evolved and been reorganized
- **Reference genome assembly** – perform *de novo* assembly and correct assemblies generated by sequencing-based systems

## INTELLIGENT DATA SOLUTIONS

### Automate data analysis, monitor and manage the Saphyr remotely

Bionano Access™, your web-based hub for Saphyr™ operations, provides all the software you need for experiment management and Bionano genome mapping in one place.

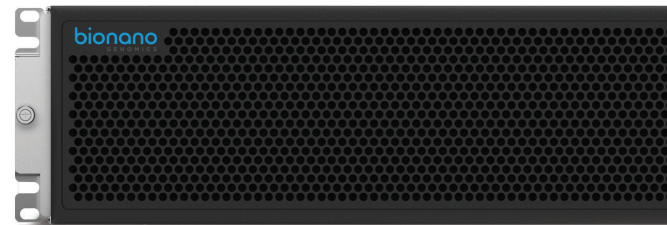
- Set up runs and monitor real-time data quality metrics remotely to flag potential sample quality issues early
- Automatically start *de novo* assemblies and structural variation analysis when the desired amount of data has been collected
- Visualize and manipulate maps and structural variants
- Generate two independent maps from one sample with two enzymes for high sensitivity structural variation calling, improving contiguity of sequence assemblies and automatic correction of assembly errors
- Analyze trios by filtering through uncommon variants to identify inherited and *de novo* variants, and export in a dbVar-compliant VCF file



### Unparalleled processing power

The Saphyr and Bionano Compute Servers offer cluster-like performance in an affordable, compact solution.

- Perform *de novo* assembly of a human genome in approximately 28 hours
- Simple web-based interface enables integration into virtually any network setup
- Cloud-based solutions available



## ORDERING INFORMATION

- **Saphyr Instrument:** Catalog # IN-012-01
- **Saphyr Chip:** Catalog # FC-030-01
- **Saphyr Chip Clip:** Catalog # FC-031-01
- **Saphyr Compute Server:** Catalog # CR-002-01
- **Bionano Compute Server:** Catalog # CR-003-01

**bionano**  
GENOMICS

**Contact us to learn more  
about Saphyr.**

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