REVEAL MORE GENOMIC VARIATION THAT MATTERS WITH OPTICAL GENOME MAPPING

Find Variants Other Technologies Can't See with Optical Genome Mapping

The Bionano Saphyr® System detects structural variations in an unbiased manner at much higher sensitivities than sequencing-based technologies, and routinely at 5% variant allele fraction.

THE SAPHYR SYSTEM IS IDEAL FOR

Consolidating traditional cytogenetic methods into a single workflow

Unbiased genome-wide structural variant detection

Finding variation missed by sequencing-based methods

WIDE RANGE OF APPLICATIONS

Constitutional Genetic Disorders
Find more answers by detecting large structural events missed by NGS and cytogenetic methods.

Hematologic Malignancies
Detect relevant mutations at > 1,000x higher resolution over karyotyping.

Gene Discovery and Therapy
Identify genes of interest, their locations, and how SVs impact them, for effective therapy development.
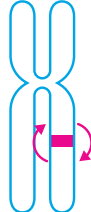
Cell Bioprocessing Quality Control
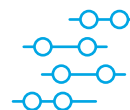Detect transgenes and identify unwanted genomic changes introduced in cell culture.

Solid Tumor Research
Detect somatic rearrangements in heterogeneous tumors at 5% variant allele fraction.

Reference Genome Assembly
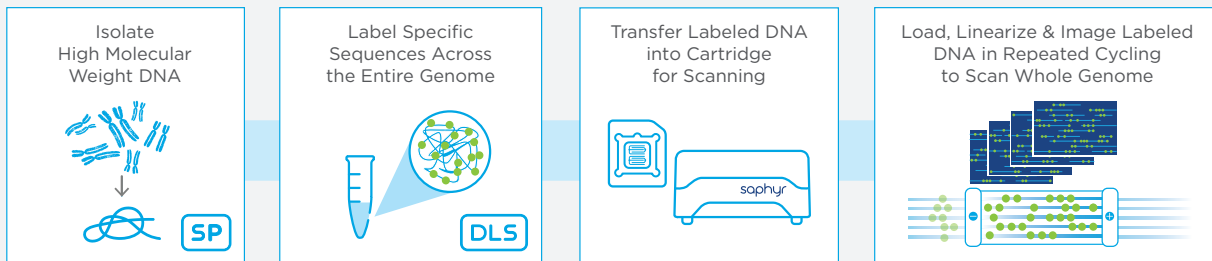Achieve highest quality, error-free *de novo* assemblies by correcting order and orientation of sequencing contigs.

HIGHLY SENSITIVE DETECTION ACROSS STRUCTURAL VARIANT TYPES

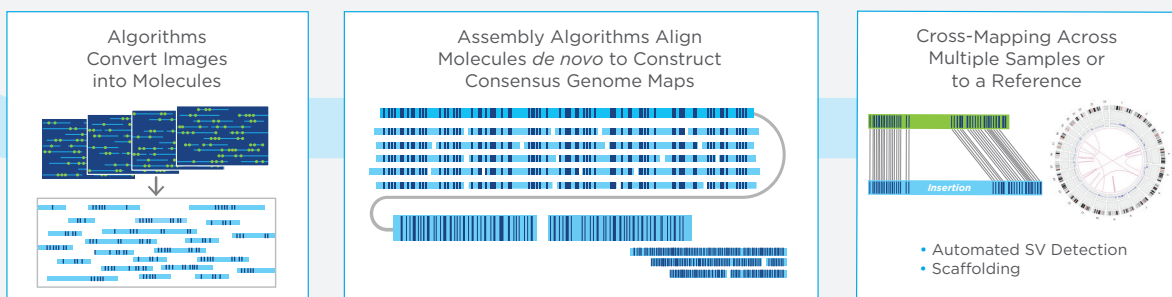| **Insertions/deletions**	**Balanced and unbalanced translocations**	**Inversions**	**Duplications**
500 bp for diploid genomes	larger than 50 kbp	larger than 30 kbp	larger than 30 kbp
5 kbp for mosaic sample			

OGM MAKES DETECTING STRUCTURAL VARIANTS EASY AND EFFICIENT

The Saphyr System images ultra-long, linearized DNA molecules labeled at specific sequence motifs. Comparative analysis of the label patterns over long contiguous reads across the whole genome reveals structural variants (>500 bp). All major types of large structural variants can be detected at variant allele fractions of 5%.

Isolate High Molecular Weight DNA — SP

Label Specific Sequences Across the Entire Genome — DLS

Transfer Labeled DNA into Cartridge for Scanning

Load, Linearize & Image Labeled DNA in Repeated Cycling to Scan Whole Genome

High-throughput, High-resolution Imaging of Megabase Length Molecules

Algorithms Convert Images into Molecules

Assembly Algorithms Align Molecules *de novo* to Construct Consensus Genome Maps

Cross-Mapping Across Multiple Samples or to a Reference

insertion

- Automated SV Detection
- Scaffolding

Total processing time: as little as 4 days*

* For human samples collected at 100x and analyzed through the *de novo* assembly pipeline.

Streamline Your Workflow with Optimized Sample Prep and Labeling Kits

Bionano Prep Kits™ provide the critical reagents necessary to extract and label ultra-high molecular weight (UHMW) DNA that is compatible with Saphyr.

OPTIMIZED KITS FOR EVERY NEED

The latest Bionano SP DNA prep kits are capable of purifying UHMW DNA in as little as 4 hours using a lyse, bind and wash process and novel paramagnetic disks. The resulting purified DNA is several megabases in length and is optimal for downstream use with Saphyr Systems.

Starting with UHMW DNA purified using the appropriate Bionano Prep Kit, fluorescent labels are attached to a 6 bp sequence motif, occurring 20 times per 100 kbp. Fluorescent labels are attached via the direct label and stain technology (DLS) which is nondestructive and leaves DNA samples intact. The result is uniquely identifiable genome-specific label patterns that enable *de novo* map assembly, anchoring sequencing contigs, and discovery of structural variations starting at 500 bp.

SAMPLE TYPES
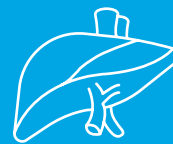
Tissue

Bone Marrow

Blood

Cell Lines

	Fresh/frozen Blood	Cell Culture	Bone Marrow Aspirates (BMA)	Tissue
Bionano Solution Phase (SP) Prep Kit	✓	✓	✓	✓
Sample Input	1.5 x 10^6 cells White blood cells (WBCs) for blood and BMA			10 – 30 mg

Experience the Power of Saphyr Chips®

Bionano Saphyr chips utilize hundreds of thousands of massively parallel nanochannels that linearize long, labeled DNA molecules, allowing the Saphyr Instrument to directly image your samples.

SAPHYR CHIP FEATURES:

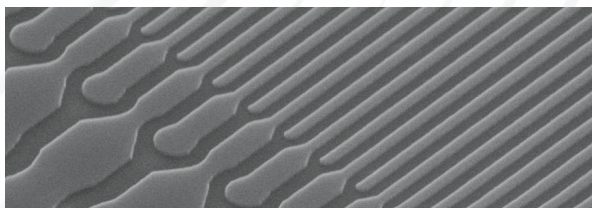Enhanced optics with adaptive loading of DNA utilizing machine learning

Fast sample loading

Automatic optimization of run conditions to maximize throughput

Saphyr Chip Clip protects sample integrity

Saphyr Chip's Nanochannel Technology Keeps DNA Intact

Saphyr Chip's nanochannels allow only a single linearized DNA molecule to travel through while preventing the molecule from tangling or folding back on itself. This nanofluidic environment allows molecules to move swiftly through hundreds of thousands of parallel nanochannels simultaneously, enabling high-throughput processing to build an accurate Bionano genome map.

"We need research data on structural variants like OGM provides, to ultimately help patients. It has the potential to change the way we diagnose diseases and eventually how we manage them."

Dr. Rashmi Kanagal-Shamanna
The University of Texas, M.D. Anderson Cancer Center

Intelligent Data Solutions

Manage and monitor data generation on Saphyr and generate genome assemblies and variation reports in one place.

BIONANO ACCESS SOFTWARE

Bionano Access® Software is a web-based hub for Saphyr® operations, providing all the software needed for experiment management and OGM applications. With Bionano Access, you can:

- Set up and monitor runs remotely to flag potential sample-quality issues
- Perform structural variation calling and annotation
- Filter and generate variant reports
- Automate *de novo* and somatic variant detection by comparing multiple samples and export in a dbVar-compliant VCF file
- Generate *de novo* assemblies for population-specific reference genomes

ANALYSIS PIPELINES

Bionano Access has several analysis pipelines to get the most out of your OGM data.

- **Rare Variant Analysis Pipeline** detects SVs genome-wide without bias, including analysis of heterogeneous tumor/mosaic samples down to an average level of detection of 5% variant allele fraction
- **Copy Number Variation Pipeline** detects copy number changes from 500 kbp up to aneuploidies, down to 10% variant allele fraction with high sensitivity
- **Variant Annotation Pipeline** filters all SV calls based on the frequency of variants in a built-in control database, and external databases. It annotates calls by providing overlapping gene information, and performs trio-analysis and tumor-normal comparison
- ***De novo* Assembly Pipeline** builds consensus maps for genome assembly projects to scaffold sequence contigs into chromosome arm-length assemblies, or to call heterozygous structural variants with unmatched sensitivity and precision
- **Bionano EnFocus™ FSHD Analysis Pipeline** for targeted measurement of the D4Z4 repeat array on chromosome 4
- **Bionano EnFocus™ Fragile X Analysis Pipeline** for targeted measurement of the CGG trinucleotide repeat array in the *FMR1* gene

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COMPUTE SOLUTIONS

BIONANO COMPUTE ON DEMAND

Bionano Compute On Demand is a pay-per-use solution accessible through Bionano Access web application for your Bionano Solve operations. Compute On Demand simplifies the way you perform analyses, without the need of any additional infrastructure, giving you the flexibility and scalability your experiment deserves.

Advantages include:

- Analyze large genomes and a high number of samples simultaneously
- Perform pipeline analysis operations without worrying about server capacity
- Data encrypted for secure operations
- Data centers compliant with IPAA, CSA, SOC2, ITAR regulations
- Genomic data accessible only to end-users and deleted post-processing

BIONANO COMPUTE SERVERS

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 20 hours
- Simple web application interface enables integration into virtually any network setup

COMPUTE OPTIONS

COMPUTE SERVER
- Lower cost per sample for typical instrument owners
- Highly tuned and tested solution
- Best security for data protection

COMPUTE ON DEMAND
- Execute variable workloads
- No upfront server costs required
- Receive data from service providers
- Work on large genomes

Bionano Technical Support

The Bionano Support team is committed to your smooth onboarding and continued success.

ONBOARDING

Be it reviewing your applications, identifying the right solution based on your experimental goals, or discussing your computational needs, our Field Applications and Field Service teams are with you, every step of the way.

TRAINING

Every Saphyr system is accompanied by exceptional training to get you started on the right foot. This onsite training of users covers the entire workflow, from sample preparation to data review and assessment using Bionano Access.

CONTINUED SUPPORT

The Bionano Customer Solutions team is available anytime to help with ongoing and future projects, troubleshooting and resolving issues, and ensuring you get the most value possible from your Saphyr System.

SAPHYR ASSURE

Saphyr Assure is the optional automated system health monitoring service that continuously inspects data quality and instrument performance. Performance issues are diagnosed early and validated updates are automatically ready for installation.

YOUR BIONANO SUPPORT TEAM

Field Application Scientists
Regular calls to provide product and administrative updates and review project status and pipeline

Technical Support
Available via e-mail and phone

Field Service Engineers
Performance of annual preventive maintenance and onsite and remote system troubleshooting

Contact Bionano Customer Solutions team

bionanogenomics.com/support

support@bionanogenomics.com

3 Ways to Get Bionano Data

GET THE SERVICE	GET THE CONSUMABLES	GET THE SAPHYR SYSTEM

BIONANA DATA SERVICES

Submit your samples to Bionano or a certified service provider

- Fresh/frozen human samples accepted: Tissue, Blood, Cultured Cells, Bone Marrow Aspirates
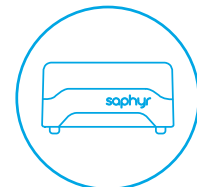
REAGENT RENTAL AGREEMENT

Run samples in-house with a free Bionano Saphyr® instrument

- Flexible reagent commitment terms
- Installation, Compute On Demand analysis, and training included

SYSTEM AND CONSUMABLES PURCHASE

Purchase a Saphyr System for your institution

- Installation and training included

Bionano Genomics Ordering Guide

CATEGORY	Part No.	Product
KITS	80042	Bionano Prep SP Blood and Cell DNA Isolation Kit v2 (10 Reactions)
	90103	Bionano Prep SP Bone Marrow Aspirate (BMA) DNA Isolation Kit v2 (10 reactions)
	80038	Bionano Prep SP Tissue and Tumor DNA Isolation Kit
	80005	Bionano Prep DLS Labeling Kit (10 Reactions)
	90101	Bionano Prep SP Tissue and Tumor 30 Genome Bundle G2.3*
	90106	Bionano Prep SP Blood and Cell DNA Isolation Kit, DLS Labeling Kit v2 and Saphyr G2.3 Chips – 30 Genome Bundle G2.3*
	90107	Bionano Prep SP BMA DNA Isolation Kit, DLS Labeling Kit v2 and Saphyr G2.3 Chips – 30 Genome Bundle G2.3*
CHIP	20366	Saphyr Chip® G2.3 (formerly 3x1300*)
INSTRUMENT	90067	Saphyr® System with Bionano Access Server, 1 color
	90099	Saphyr® Extended Warranty Service Contract (1y)
COMPUTING	80013	Saphyr Compute Server
	80014	Bionano Compute Server
	90088	Bionano Bioinformatic Customer Compute Setup
	90047	Bionano Compute On Demand, US
	90060	Bionano Compute On Demand, Europe
	90098	Bionano Compute On Demand, Germany
DATA SERVICES	90072	100x Human Genome Sample Analysis
	90073	400x Human Genome Sample Analysis
	90074	Advanced Genome Sample Analysis

*Compatible only with Saphyr® System #90023 and #90067 (Saphyr® Instrument #60325 and #60396)

Contact your Bionano Regional Business Manager to get started.

✉ **sales@bionanogenomics.com**

📞 **1.858.888.7600**

bionano®
GENOMICS

bionanogenomics.com