bionano™

Reveal More Genomic Variation That Matters With Optical Genome Mapping
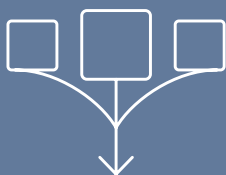
saphyr

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, at high resolution and routinely at 5% variant allele fraction.

Empower Your Lab with the Saphyr System

A workflow alternative to multiple traditional cytogenetic methods

Unbiased genome-wide structural variant detection

Find genetic variation missed by sequencing and cytogenetic methods

Wide Range of Applications

Constitutional Genetic Disorders
Detect genome-wide SVs at >10,000x higher resolution over karyotyping.

Hematologic Malignancies
Detect genome-wide CNVs and fusions, including fusion partners.
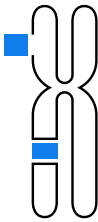
Gene Discovery and Therapy
Identify genes of interest, their locations, and how SVs impact them, for effective therapy development.
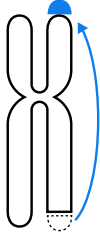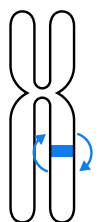
Cell Bioprocessing Quality Control
Detect transgenes and identify unwanted genomic changes introduced in cell culture.

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

Pair OGM with Sequencing
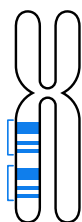Achieve more comprehensive variant calls by combining sequencing for single-nucleotide variants and indels with OGM for whole genome SV detection.
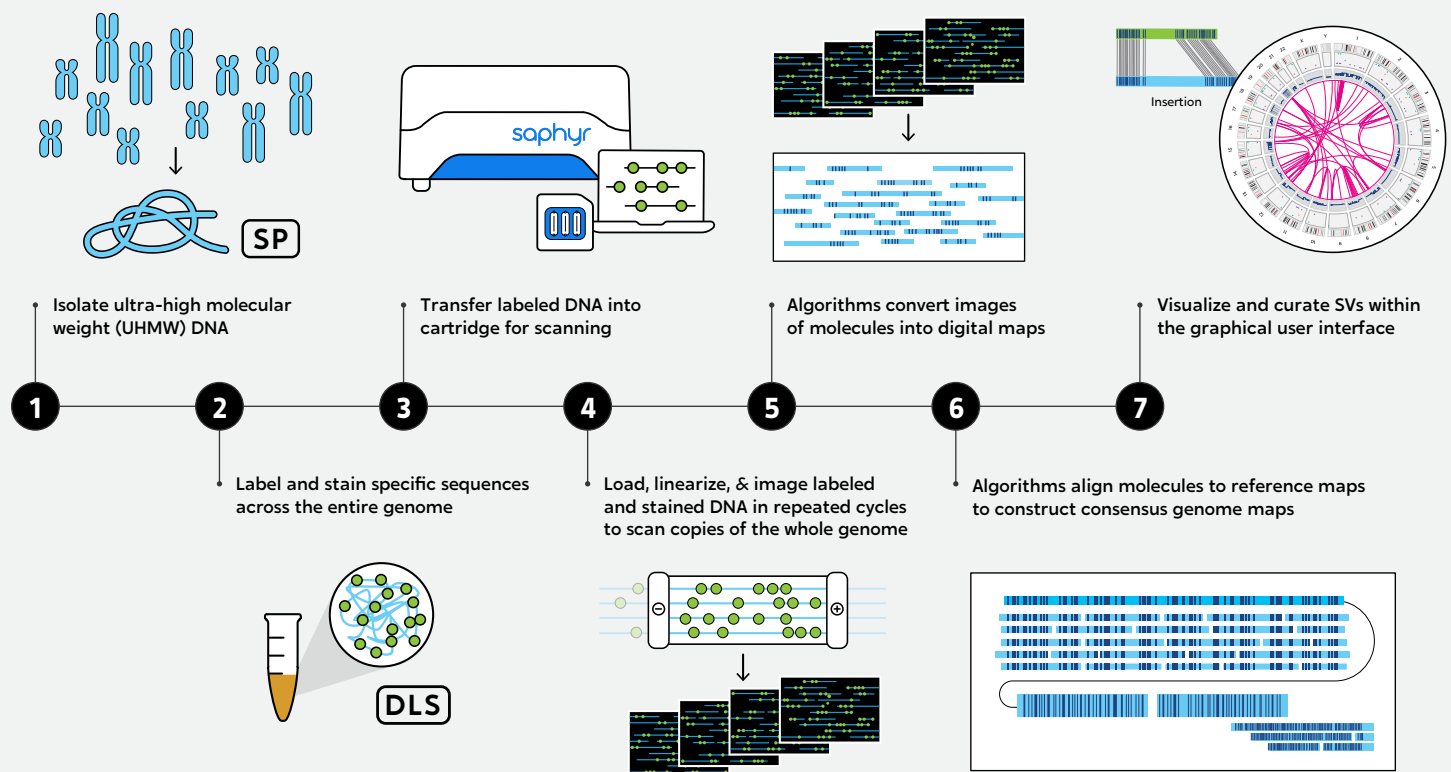
Highly Sensitive Detection Across Structural Variant Types

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

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 at high resolution. All major types of large structural variants can be detected at variant allele fractions of 5%.

1 Isolate ultra-high molecular weight (UHMW) DNA

2 Label and stain specific sequences across the entire genome

3 Transfer labeled DNA into cartridge for scanning

4 Load, linearize, & image labeled and stained DNA in repeated cycles to scan copies of the whole genome

5 Algorithms convert images of molecules into digital maps

6 Algorithms align molecules to reference maps to construct consensus genome maps

7 Visualize and curate SVs within the graphical user interface

Total processing time: as few as 3 days*

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

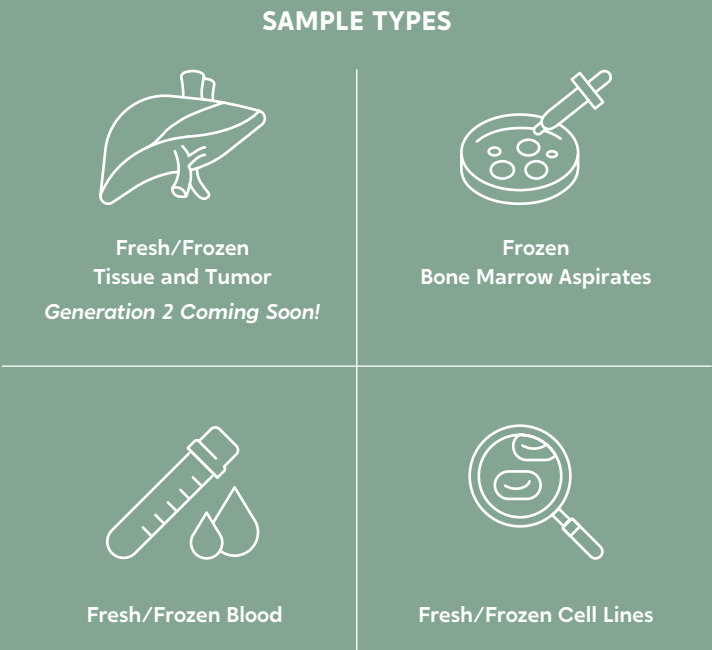# Streamline Your Workflow with Optimized Sample Prep and Labeling Kits

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

Isolate UHMW DNA from Important Sample Types

The Bionano sample prep (SP) kits are capable of purifying UHMW DNA from tissue and tumor, bone marrow aspirate (BMA), blood and cells as well as plant and animal tissue. The latest SP Generation 2 (SP-G2) kits further increase robustness, throughput, and consistency for sample preparation. This sample diversity makes OGM suitable for use in a broad range of studies and applications in oncology, constitutional genetic disease, bioprocessing, and general research.

Bionano SP kits require 1.5 million cells (blood, cell lines and BMA) or 10 – 30 mg of tissue as input. UHMW DNA is isolated in about four hours using a lyse, bind, and wash process, and novel paramagnetic disks.

SAMPLE TYPES

Fresh/Frozen Tissue and Tumor
Generation 2 Coming Soon!

Frozen Bone Marrow Aspirates

Fresh/Frozen Blood

Fresh/Frozen Cell Lines

	Fresh/Frozen Blood	Fresh/Frozen Cell Culture	Frozen Bone Marrow Aspirates (BMA)	Fresh/Frozen Tissue and Tumor
Sample Input	1.5 x 10^6 cells White blood cells (WBCs) for blood and BMA			10 – 30 mg
Sample Preparation Generation 2 Kits	✔	✔	✔	Coming soon!
Previous Sample Preparation Kits (Generation 1)	✔	✔	✔	✔

Experience the Power of Saphyr Chips®

Bionano Saphyr Chip® consumables 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:

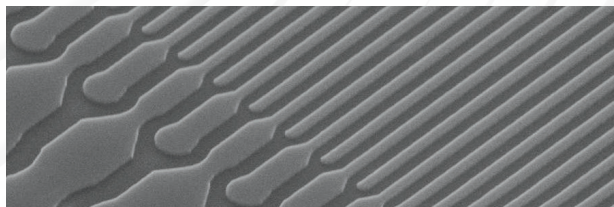Leverages adaptive loading of DNA utilizing machine learning

Fast sample loading

Allows automatic optimization of run conditions to maximize throughput

Saphyr Chip Clip protects and seals sample integrity

Saphyr Chip's Nanochannels Linearize UHMW DNA for Single-molecule Imaging

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. Confined in this nanofluidic environment, DNA molecules linearize across hundreds of thousands of nanochannels where they can be imaged to reveal the underlying genomic structure and structural variation.

"We need research data on structural variants—like OGM provides—to ultimately help patients. OGM 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 system operations, providing all the software needed for experiment management and OGM applications. With Bionano Access, you can:
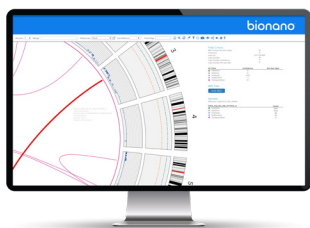
- Set up and monitor runs remotely
- Execute structural variant detection and annotation pipelines
- Visualize and filter SVs to generate variant reports
- Generate *de novo* assemblies for population-specific reference genomes

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 down to an average of 5% variant allele fraction for analysis of heterogeneous or mosaic samples
- *De novo* **Assembly Pipeline** calls heterozygous structural variants with unmatched sensitivity and precision, down to >500bp
- **Guided Assembly Pipeline** provides unbiased detection of SVs genome-wide with high confidence for constitutional and cancer applications
- **Copy Number Variation Pipeline** detects CNVs from 500 kbp down to 10% variant allele fraction with high sensitivity
- **Variant Annotation Pipeline** calculates SV frequency based on control and external databases, provides gene annototations, and performs trio or paired-sample analyses
- **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 in the *FMR1* gene

BIONANO VIA™ SOFTWARE
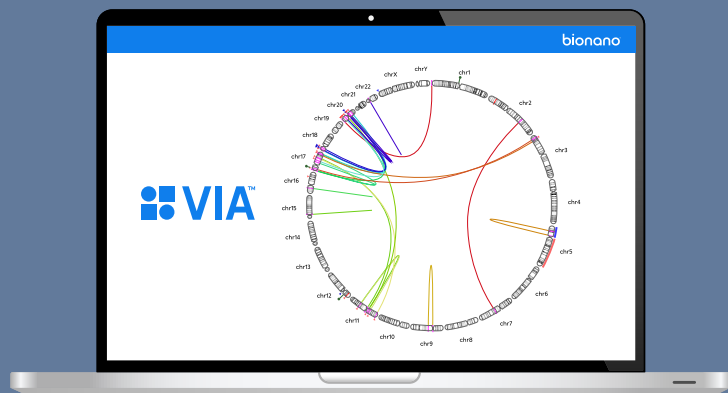
Harness the Full Potential of Your Data.

VISUALIZE
Generate powerful visualizations for enhanced contextualization across multiple variant types

INTERPRET
Accelerate time to results with intelligent automation for filtering, classifying, annotating and interpreting data across technologies

REPORT
Deliver clear, highly visual reports that support informed decision-making

INTEGRATE
View and fully analyze data from OGM, NGS and microarray into one system for a simplified and integrated workflow

BIONANO SOLUTIONS

Bionano provides a complete suite of hardware, software, and cloud-based solutions for end-to-end experiment management and bioinformatics processing of OGM data. The Saphyr Compute Server and Bionano Compute Server offer on-site cluster-like performance in an affordable, compact solution, capable of performing multiple simultaneous analyses and sustaining continuous throughput. Bionano Compute On Demand is a pay-per-use solution accessible through Bionano Access web server for your Bionano Solve operations.

Compute Options

Compute On Demand

- Execute variable workloads
- No upfront server costs required
- Data centers compliant with IPAA, CSA, SOC2, ITAR regulations
- Work on large genomes

Compute Server

- Lower cost per sample for typical instrument owners
- Highly tuned and tested solution
- Best security for data protection
- Internet access not required

Bionano Technical Support

The Bionano Customer Solutions 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 Customer Solutions 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

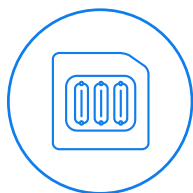**Contact Bionano Customer Solutions Team**

bionano.com/support

support@bionano.com

How to Access Bionano Data
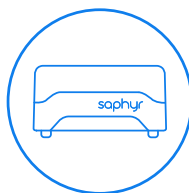
Get the Consumables

Reagent Rental Agreement

Run samples in-house on a Bionano Saphyr® instrument placed at your institution with a fixed reagent commitment

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

Get the Saphyr System

Hardware and Consumables Agreement

Purchase a Saphyr system and consumables for your instation

- Installation and training included
- No mimimum consumable commitment to purchase system

Bionano Genomics Ordering Guide

Category		Part No.	Product
Kit	GENERATION 2	80060	Bionano Prep SP-G2 Blood & Cell DNA Isolation Kit
		90151	Bionano Prep SP-G2 Bone Marrow Aspirate DNA Isolation Kit
		80046	DLS-G2 DNA Labeling Kit
		90145	Bionano Prep SP-G2 Blood & Cell — 12 Genome Bundle, G3.3
		90146	Bionano Prep SP-G2 BMA — 12 Genome Bundle, G3.3
		90147	Bionano Prep SP-G2 Blood & Cell — 12 Genome Bundle, G2.3
		90148	Bionano Prep SP-G2 BMA — 12 Genome Bundle, G2.3
	GENERATION 1	80042	Bionano Prep SP Blood & Cell DNA Isolation Kit v2
		90103	Bionano Prep SP Bone Marrow Aspirate DNA Isolation Kit v2
		80038	Bionano Prep SP Tissue & Tumor DNA Isolation Kit
		80005	Bionano Prep DLS Labeling Kit
		90101	Bionano Prep SP Tissue & Tumor 30 Genome Bundle G2.3*
		90106	Bionano Prep SP Blood & Cell v2 30 Genome Bundle G2.3*
		90107	Bionano Prep SP BMA v2 30 Genome Bundle G2.3*
Chip		20440	Saphyr Chip® G3.3
		20366	Saphyr Chip® G2.3
Instrument		90067	Saphyr® System with Bionano Access Server, 1-color
		90099	Saphyr® Instrument (1-color) Extend Warranty Service Contract (1 year)
Compute Hardware		80083	Saphyr Compute Server, Gen4
		80084	Bionano Compute Server, Gen4
Cloud Computing		90047	Bionano Compute On Demand, US
		90128	Bionano Compute on Demand, Canada
		90060	Bionano Compute On Demand, Europe
		90052	Bionano Compute On Demand, Germany
Service		90088	Bioinformatic Customer Compute Setup
Maintenance		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.

To order, please contact orders@bionano.com or

Call **1.858.888.7600** or contact your sales person directly.

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