



SEE STRUCTURAL VARIATION LIKE  
NEVER BEFORE WITH BIONANO  
OPTICAL GENOME MAPPING

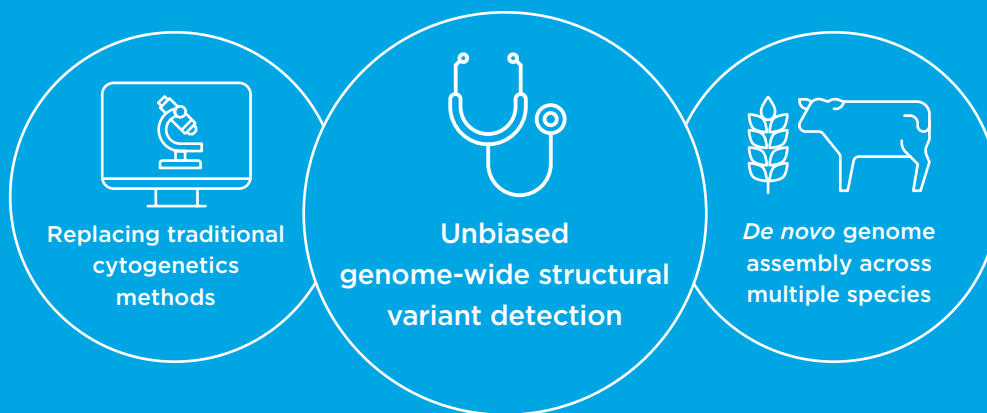


# Picture Your Next Discovery with Bionano Optical Genome Mapping



The Saphyr® System by Bionano Genomics is a genome imaging tool for high-speed, high-throughput structural variant detection and analysis with exceptional sensitivity and specificity.

## THE SAPHYR SYSTEM IS IDEAL FOR



## WIDE RANGE OF APPLICATIONS



### Undiagnosed Genetic Disorders

Complete the diagnostic odyssey by detecting large structural events missed by NGS and cytogenetic methods.



### Hematologic Malignancies

Detect relevant mutations at >1,000x higher resolution over karyotyping with genome imaging.



### Gene Discovery and Therapy

Identify genes of interest, their locations, and how SVs impact them, for effective therapy development.



### Cell Line Stability

Detect genome instability caused by insertions/deletions introduced during DNA replication in cell lines.



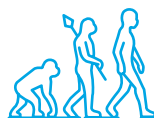
### Solid Tumor Research

Detect somatic rearrangements in heterogeneous tumors down to 1% allele fraction.



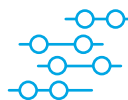
### Genetic Engineering Studies

Identify areas of biological interest for achieving desirable traits in model systems or crops.



### Evolutionary Biology


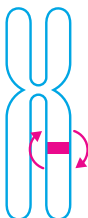

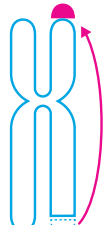
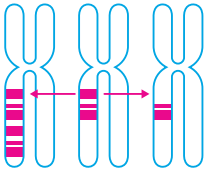
See the complete picture of how genomes have evolved and have been reorganized.



### Reference Genome Assembly

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

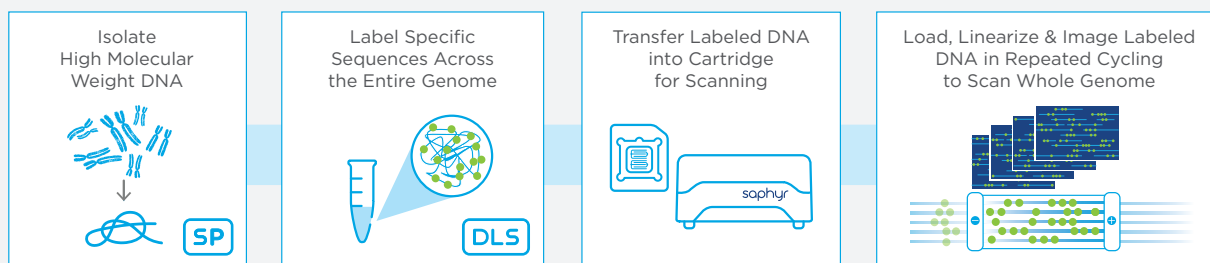
## WITH THE SAPHYR SYSTEM, YOU CAN DETECT STRUCTURAL VARIANTS OTHER TECHNOLOGIES MISS

				
<b>Insertions/deletions</b> larger than 500 bp	<b>Inversions</b> larger than 30 kbp	<b>Duplications</b> larger than 30 kbp	<b>Balanced and unbalanced translocations</b> larger than 50 kbp	<b>Fractional copy number variations</b> larger than 500 kbp
<b>99% sensitivity</b>	<b>99% sensitivity</b>	<b>97% sensitivity</b>	<b>95% sensitivity</b>	<b>97% sensitivity</b>
<b>False positives below 2%</b>				

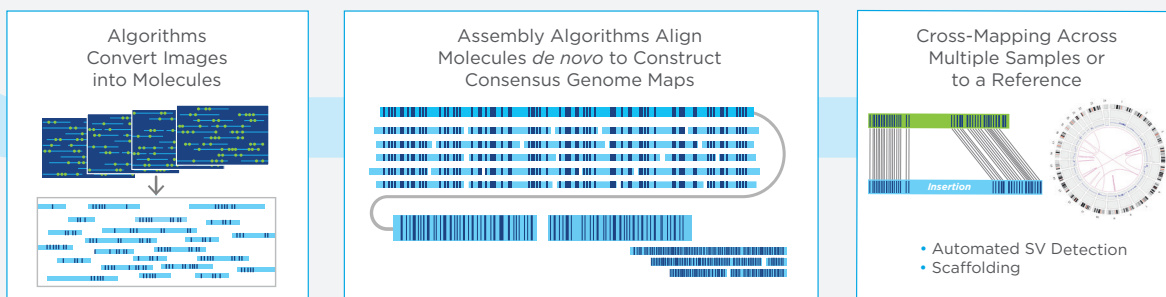
## SAPHYR MAKES DETECTING STRUCTURAL VARIANTS WITH HIGH SPECIFICITY AND SENSITIVITY EASY AND EFFICIENT

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), at sensitivities as high

as 99%, with false positive rates below 2%. All major types of large structural variants can be detected, even at allele fractions as low as 1%, which is not enabled by any other genomics technology.



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



Total processing time: as little as 4 days

# 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.

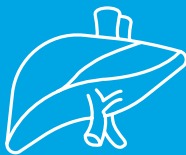


## OPTIMIZED KITS FOR EVERY NEED

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 Bionano systems.

Starting with UHMW DNA purified using the appropriate Bionano Prep Kit, fluorescent labels are attached to a 6 bp sequence motif, occurring 15 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



Plant Tissue



Blood



Cell Lines

	Fresh/frozen Blood	Cell Culture	Bone Marrow Aspirates (BMA)	Tissue	Plant
Bionano Prep Kit (plus lysis)	✓	✓		✓	✓
Bionano Solution Phase (SP) Prep Kit	✓	✓	✓	✓	
Sample Input	1.5 x 10 <sup>6</sup> cells White blood cells (WBCs) for blood and BMA			10 – 30 mg	2g young leaves

## 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



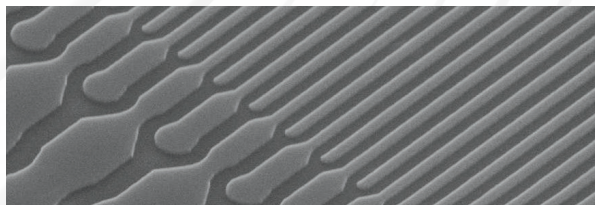
Fast sample loading



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 Bionano 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

Bionano Access® is a web-based hub for Saphyr® operations, provides all the software for experiment management and Bionano applications, including variant annotation and reporting. With Bionano Access, you can:

- Set up and monitor runs remotely to flag potential sample-quality issues
- Perform one-click 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 most out of your Bionano data.

- **Rare Variant Pipeline** has >90% sensitivity to detect SVs down to 1% variant allele fraction, completely unbiased and genome wide, allowing for the analysis of heterogeneous tumor/mosaic samples
- **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 info, 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

## 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 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

- Expect to run servers for >25% of the time
- Execute consistent loads
- Ideal solution when internet access not permitted



#### COMPUTE ON DEMAND

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



# Bionano Technical Support

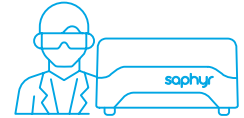
The Bionano Support team is committed to your smooth onboarding and continued success.

## 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](https://bionanogenomics.com/support)



[support@bionanogenomics.com](mailto:support@bionanogenomics.com)

## 3 Ways to Get Bionano Data

### GET THE SERVICE



### BIONANO DATA SERVICES

Submit your samples to Bionano or a certified service provider

- Fresh/frozen human samples accepted: Tissue, Blood, Cultured Cells, Bone Marrow Aspirates
- Non-human samples accepted only through service providers

### GET THE CONSUMABLES

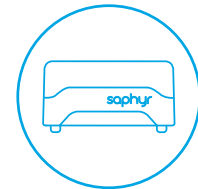


### REAGENT RENTAL AGREEMENT

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

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

### GET THE SAPHYR SYSTEM



### 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)
	80003	Bionano Prep Plant Tissue DNA Isolation Kit (5 Reactions)
	80002	Bionano Prep Animal Tissue DNA Isolation Kit (10 Reactions)
	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)

To order, please contact  
[orders@bionanogenomics.com](mailto:orders@bionanogenomics.com),  
 call 1.858.888.7600 or  
 contact your sales person directly.



[bionanogenomics.com](https://bionanogenomics.com)