

# REVEAL MORE GENOMIC VARIATION THAT MATTERS WITH OPTICAL GENOME MAPPING

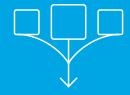


## Find Variants Other Technologies Can't See with Optical Genome Mapping



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.

#### **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.



#### **Cell Bioprocessing Quality Control**

Detect transgenes and identify unwanted genomic changes introduced in cell culture.



### **Hematologic Malignancies**

Detect all genome-wide CNVs and fusions, including fusion partners.



#### **Solid Tumor Research**

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



### **Gene Discovery and Therapy**

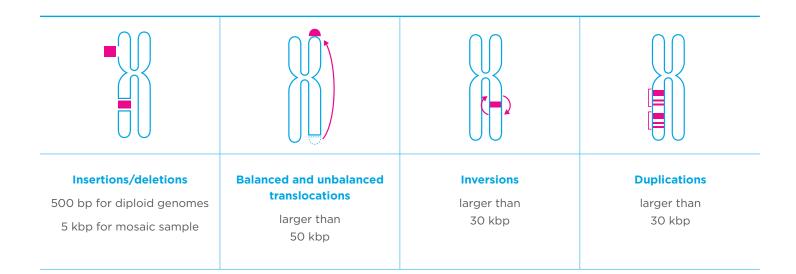
Identify genes of interest, their locations, and how SVs impact them, for effective therapy development.



#### 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.

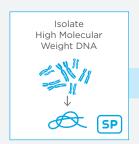
#### HIGHLY SENSITIVE DETECTION ACROSS STRUCTURAL VARIANT TYPES



#### **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%.

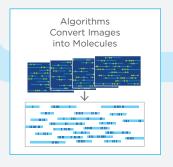


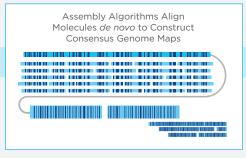


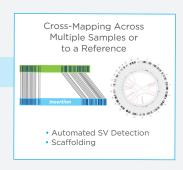




#### High-throughput, High-resolution Imaging of Megabase Length Molecules







Total processing time: as few 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.



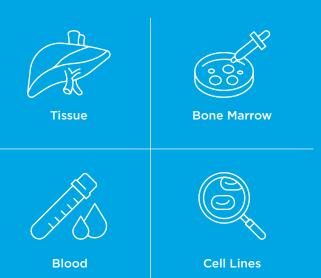
#### **OPTIMIZED KITS FOR YOUR NEEDS**

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, on average, 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**



	Fresh/frozen Blood	Cell Culture	Bone Marrow Aspirates (BMA)	Tissue
Bionano Solution Phase (SP) Prep Kit	<b>\</b>	<b>\</b>	<b>/</b>	<b>✓</b>
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:**





Leverages adaptive loading of DNA utilizing machine learning



Fast sample loading



Allows automatic optimization of run conditions to maximize throughput



Saphyr Chip Clip protects 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® 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 populationspecific 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

#### **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 Customer Solutions team is committed to your smooth onboarding and continued success.

#### **ONBOARDING**



**TRAINING** Be it reviewing your applications, identifying the right Every Saphyr system is accompanied by exceptional

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 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**





#### SAPHYR ASSURE





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 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**



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 vour 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 (10 Reactions)		
	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 v2 30 Genome Bundle G2.3*		
	90107	Bionano Prep SP BMA v2 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		

<sup>\*</sup>Compatible only with Saphyr\* System #90023 and #90067 (Saphyr\* Instrument #60325 and #60396)

**Contact your Bionano Regional Business Manager to get started.** 



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bionanogenomics.com