Reveal More Genomic Variation That Matters With Optical Genome Mapping

Find Variants Other Technologies Just Can't See

The Bionano Saphyr® system detects structural variations in an unbiased manner at much higher sensitivities than sequencing-based technologies, down to 500 bp 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 traditional 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.

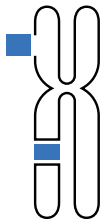
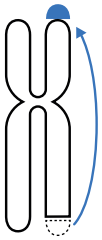
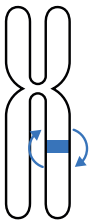
Solid Tumor Research

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

Pair OGM with Sequencing

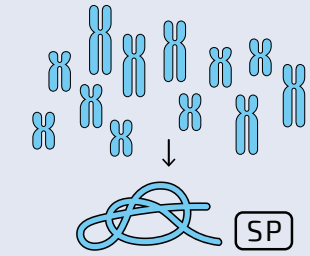
Achieve more comprehensive variant calls by combining sequencing for single-nucleotide variants and indels with OGM for whole genome SV detection.

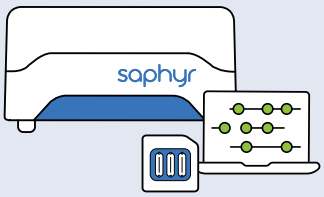
Highly Sensitive Detection Across Structural Variant Types

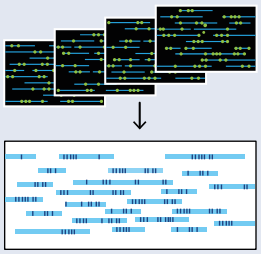
			
Insertions/deletions 500 bp for diploid genomes 5 kbp for mosaic sample	**Balanced and unbalanced translocations** larger than 50 kbp	**Inversions** larger than 30 kbp	**Duplications** larger than 30 kbp

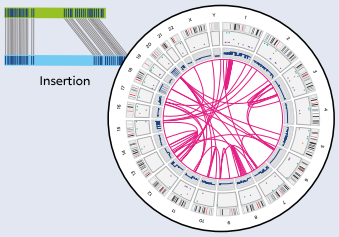
OGM Makes Detecting Structural Variants Easy and Efficient

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 (>500 bp). All major types of large structural variants can be detected at variant allele fractions of 5%.

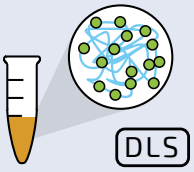


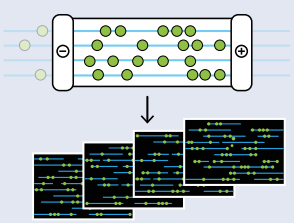


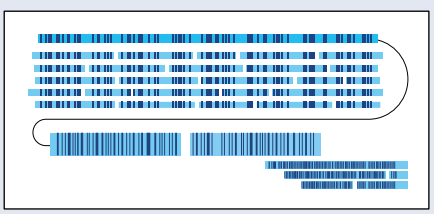




1. Isolate ultra-high molecular weight (UHMW) DNA
2. Label and stain specific sequences across the entire genome
3. Transfer labeled DNA into cartridge for scanning
4. Load, linearize, & image labeled and stained DNA in repeated cycles to scan copies of the whole genome
5. Algorithims convert images 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*

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

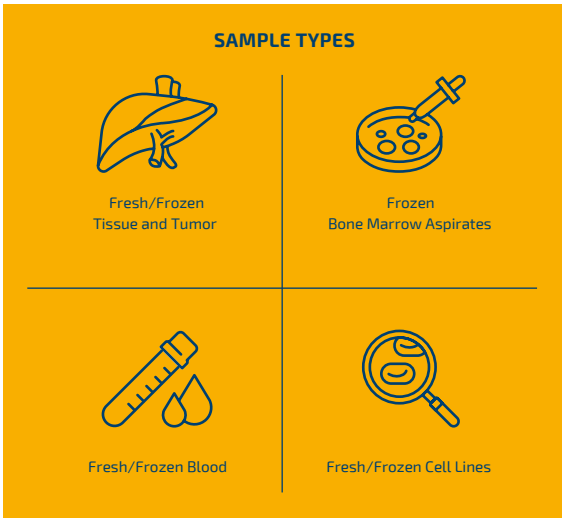
Streamline Your Workflow with Optimized Sample Prep and Labeling Kits

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.



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

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 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
- **De novo Assembly Pipeline** calls heterozygous structural variants with unmatched sensitivity and precision
- **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** calculates 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
- **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

How to Access Bionano Data

Get the Consumables

Reagent Rental Agreement
Run samples in-house on an 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

Order Optical Genome Mapping service

Ready to start a project? We'll collaborate closely with you to ensure a seamless execution of the service. Reach out to us and we will guide you through the whole process. We're here to help!

We accept various sample types

- Whole blood
- Live/frozen cell lines
- Tissue and tumor
- Bone marrow
- Agarose plug

Our laboratory is located in Olomouc, Czech republic. We are ready to pick your samples up at your facility and manage the logistics but you are also welcome to join our staff and take part in processing your samples as a hands-on workshop.

When your project is complete, your data will be delivered to you via the Bionano Access® software.

Order service

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At the Forefront
of Digital Genomics

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