

bionano

Elevate the OGM Sample-to-Report Workflow with the Stratus™ System

- Consolidate multiple cytogenetic workflows into one
- Detect structural variants genome-wide
- Reveal pathogenic variants at scale



bionano.com

STRATYS™

bionano

Ionic® Purification System for FFPE Samples



IONIC™

bionano

Harness the Full Potential of Your Data. VIA™ Analysis Software is the Way.



VIA™

bionano

Power Discovery Across the Genomic Variant Continuum with VIA™ Analysis Software

The latest release of the Bionano VIA software will bring you the only decision support analysis tool that builds critical insight across the variant continuum of single nucleotide variants (SNVs), copy number variants (CNVs), and structural variants (SVs). Get ready for a new era of fully contextualized genomic discovery.



Input data from many widely adopted genomic analysis platforms

- Optical genome mapping (OGM)
- NGS
- Microarray

Find more genomic variation that matters with intelligent decision support

- Leverage automation to accelerate accurate analysis, classification and reporting
- Gain new insight to disease impact with complete, unique visualization of genomic variants
- Lower multiple data types to corroborate genomic variation for better discovery and greater efficiency

Learn more, visit bionano.com/via-software-preview

SOFTWARE

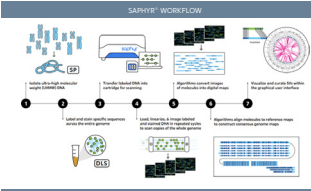
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COMPUTE SOLUTIONS

Bionano Compute Solutions: Flexible and Scalable On-premise and Cloud Computing Options

Bionano compute solutions include 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 On Demand simplifies the way you perform genome assembly, hybrid scaffolding and structural variant analysis, without the need of any additional infrastructure, giving you the flexibility and scalability your experiment deserves.

SAPHYR™ WORKFLOW



OVERVIEW OF COMPUTE OPTIONS

COMPUTE SERVER	COMPUTE ON DEMAND
• Ideal for on-site use with an on-premise solution	• Ideal for on-site use with an on-premise solution
• Scalable compute workflow	• Scalable compute workflow
• Reliable access to compute resources	• Reliable access to compute resources
• Work on target genomes up to 24 Gb genome size	• Work on target genomes up to 24 Gb genome size

COMPUTE SOLUTIONS

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CELL BIOPROCESSING

Redefine Cell Line QC with Optical Genome Mapping

Characterizing cell line genomic integrity and editing events is of critical importance to R&D and QC/QC teams working in cell and gene therapy programs.

Traditional cytogenetic and NGS methods have significant limitations in variant class coverage, resolution, turnaround time and scalability. This necessitates using a constellation of these methods to characterize and monitor genomic integrity and gene-editing events (off- and on-target) which can be complicated, time-consuming, and expensive.

Bionano Saphyr™ Optical Genome Mapping system



The Bionano Saphyr™ Optical Genome Mapping system delivers game-changing high performance and comprehensive variant class coverage with an unbiased, genome-wide approach and digital readout — without revealing proprietary sequence data.

Move Your Program Forward with Optical Genome Mapping

	OGM	Karyotype	qPCR	Microarray	NGS Panel
Genome-wide, Unbiased Detection	✓	✓	✗	✓	✓
High Resolution	✓	✗	✗	✓	✓
Variant Class Coverage	✓	✗	✗	✓	✓
Limit of Detection (LOD)	✓	✓	✓	✗	✓
Time to Result	✓	✗	✓	✓	✓
Sample-to-Answer Workflow	✓	✗	✓	✓	✗
Automated Variant Calling	✓	✗	✓	✓	✓
Cost	✓	✓	✓	✓	✗

CELL BIOPROCESSING

APPLICATIONS

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CANCER RESEARCH

Hematological Malignancies and Solid Tumor Research

Genome variation common in cancer is too complex for low coverage whole genome sequencing. Complete rearrangements as well as highly repetitive regions of the genome present additional challenges for short- and long-read sequencing technologies.

Optical Genome Mapping (OGM) detects structural variants in an unbiased manner, and at much higher resolution than sequencing-based technologies, and routinely at 95% variant allele fraction.

Genome-wide rearrangements are hallmarks of cancer transformation. Deciphering complex genomic structures requires a combination of short-read sequencing as well as long-range information with high coverage of unique rearrangements, with promising coverage of the genome. To date, NGS applications in the clinic are limited to either low coverage across the whole genome, or high coverage across rearranged DNA, and the short reads make deciphering the complex rearrangements more difficult. The breadth of cancer genomic rearrangements is too complex to be fully resolved by NGS. OGM is a powerful tool for cancer diagnosis and discovery, high cost and low throughput of long-read sequencing technologies, along with the high cost of cancer genomics are barriers to these technologies.

OGM uncovers large structural variations beyond what short- and long-read sequencing can see.

- Inversions/deletions: 100 bp to 100 kb
- Balanced and unbalanced: Larger than 10 kb
- Translocations: Larger than 10 kb
- Duplications: Larger than 10 kb

5% Variant allele detection

400X Coverage per sample

bionano.com/cancer

CANCER RESEARCH

bionano

CYTOGENOMICS

Revolutionizing Cytogenomics

Consolidate traditional cytogenetic assays into a single workflow with Optical Genome Mapping (OGM) and the Bionano Saphyr™ System

In spite of the revolutionizing sequencing technologies, karyotyping has been the gold standard for cytogenetic research and diagnosis. It has been modified the way cytogeneticists have used it, but the fundamental principle remains the same: a human karyotype is a visual representation of the human genome. While NGS identifies single-nucleotide variants along with small structural variants, OGM identifies large-scale structural variants, such as inversions and translocations. However, NGS relies on short-read sequencing that can miss large structural variants, which necessitates the use of other technologies. Long-read sequencing is still limited to resolution.

These limitations make direct visualization of the DNA the most reliable approach for the identification of structural variants to date.

Increasingly, the way structural variants are detected in clinical practice has increasingly moved over the past decades, and mainly relies on traditional cytogenetic methods such as karyotyping. Fluorescence in situ hybridization (FISH) and array-based technologies have of these methods, alone or in combination, can detect large-scale structural variants and small-scale copy number variants to provide a complete diagnostic and prognostic assessment of the patient or tumor genome.

OGM transforms traditional cytogenetic techniques by delivering an improved visualization of the genome by going from microscope to a fully digital, high-resolution chromosomal analysis.

Only here we apply OGM's full bandwidth to genome, and the resulting data is a high-resolution, genome-wide, automated analysis in a structured array that Bionano's Saphyr™ system provides. This allows us to identify OGM's greater resolution than by karyotyping and detection at 100 bp, or 4-10 kb by chromosome banding analysis.

Cytogenomics

Visual patterns on short DNA molecules to detect structural variants

Saphyr™

Automates the imaging of 100,000 short DNA molecules to reveal structural variants in a single step

bionano.com/cytogenomics

CYTOGENOMICS

bionano

GENETIC DISEASE RESEARCH

Genome-wide Detection of All Structural Variants

Advances in sequencing technologies have barely changed the way structural variants are detected. NGS relies on short-read sequencing that are required to reference a human genome and fails to identify most large insertions, deletions, and copy number variations in the two-thirds of the genome that is repetitive. In addition, NGS does not reliably detect balanced SVs such as inversions and translocations.

Optical Genome Mapping (OGM) directly visualizes patterns of labels on intact DNA molecules to detect structural variation.

OGM accurately detects all classes of structural variants, which are typically unable to detect mutations or identify them at the scale required. Instead, OGM detects both deletions and insertions starting at 100 bp with high sensitivity. It detects repeat variants down to 100 bp, or 4-10 kb by chromosome banding analysis.

Unlike sequencing-based methods, which are typically unable to detect mutations or identify them at the scale required. Instead, OGM detects both deletions and insertions starting at 100 bp with high sensitivity. It detects repeat variants down to 100 bp, or 4-10 kb by chromosome banding analysis.

OGM by the Numbers

- Up to 99% SV detection sensitivity
- 500 bp Lower limit of resolution for detection of deletions and insertions
- 3-4 days Turnaround time
- 10,000X Resolution compared to karyotype

bionano.com/geneticdisease

GENETIC DISEASE RESEARCH

Elevate the OGM Sample-to-Report Workflow with the Stratys™ System



Consolidate multiple
cytogenetic workflows
into one



Detect structural variants
genome-wide



Reveal pathogenic
variants at scale



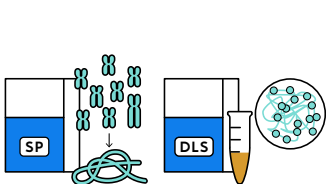
Reach a New Level of Workflow Speed and Simplicity

Reveal all classes of structural variants and leave the limits of traditional cytogenetics and sequencing behind with Optical Genome Mapping (OGM) and the Bionano Stratys™ System.



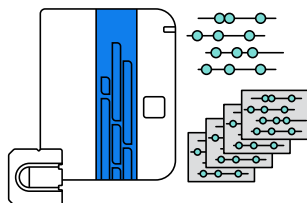
The OGM Workflow

1 PREP → **2 IMAGE** → **3 COMPUTE** → **4 INTERPRET**



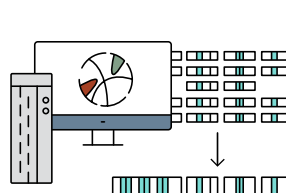
BIONANO PREP KITS

Isolate ultra-high molecular weight (UHMW) DNA. Label and stain specific sequences across the entire genome.



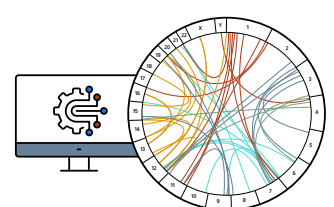
STRATYS™

Load labeled DNA into chip for scanning. Linearize and image labeled and stained DNA in repeated cycles to scan copies of the whole genome.



STRATYS™ Compute

Algorithms align molecules to reference genome to construct consensus maps and call all classes of structural variants genome-wide.

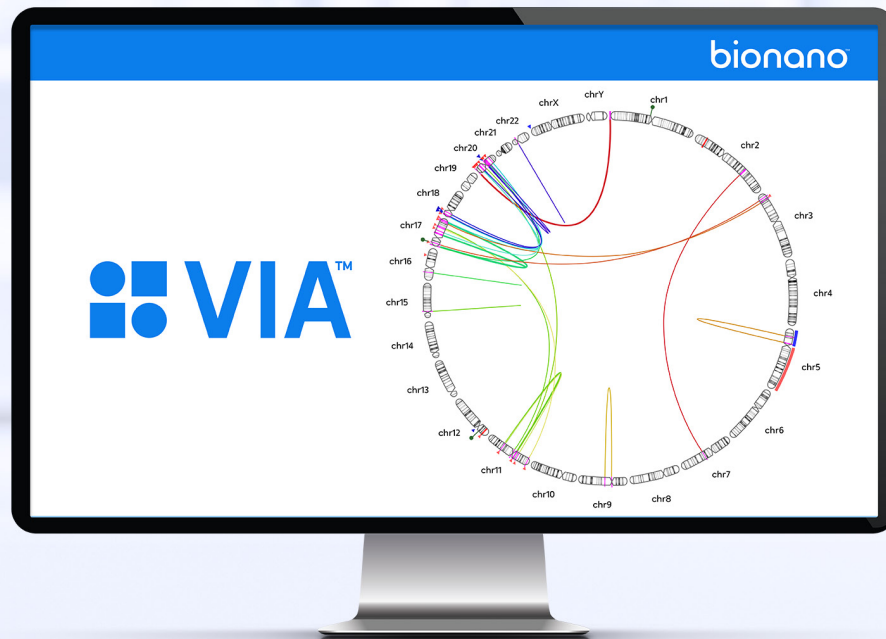


VIA™

Tertiary analysis software to visualize, interpret, and report OGM data.



Stratys™ Compute



Bionano VIA™ Software

Superior Structural Variant Detection

OGM delivers high-resolution detection and automated analysis of all structural variant classes genome-wide.

- | | |
|--------------------------------|--|
| ✓ Aneusomies | ✓ Interchromosomal Fusions |
| ✓ Interstitial Variants | ✓ Translocations |
| • Insertion | ✓ Copy Neutral Loss of Heterozygosity |
| • Deletion | ✓ Complex Rearrangements |
| • Duplications | • Ring Chromosome |
| • Inversion | • Chromothripsis |
| • Copy Number Variants | • Chromoplexy |

Detect SVs
starting at
500 bp

VAF of
5%
or lower*

Automated
**VARIANT
CALLING**

Reveal New Insights Across Applications



ONCOLOGY



GENETIC DISEASE



CELL AND GENE THERAPY

*Lower VAF percentages can be achieved when using the Stratys Plus Chip, which enables up to 1,200x genome coverage.

Rise to the Challenge of Comprehensive SV Detection at Scale

Scalable throughput and unprecedented flexibility empower you to uncover critical insights and make a transformative impact on human health.



High Sample Capacity

The Stratys instrument provides capacity for 12 single-sample, random-access chips. Load and unload chips as they complete—no need to wait for another sample to finish.



Ultimate Flexibility

No batching required. Optimize cost and efficiency in your lab based on level of demand, loading from one to 12 samples in any run.



"Jump the Queue" for Priority Samples

With reserved space for three samples so you can perform on-demand runs without disrupting the analysis of other samples on the system.



Reduced Turnaround Time

The Stratys system's simplified workflow and scalable sample capacity enables you to deliver results faster while managing lab resources more effectively.

Stratys Performance				
Target Effective Coverage ¹	100x	200x	400x	1200x ²
Expected Variant Allele Fraction (VAF) for SV Detection	≥ 50%	≥ 20%	≥ 5%	lower than 5% ⁴
Stratys Throughput Per Week ³ /Samples	24/7 operation: 260 40 hour operation: 120	24/7 operation: 168 40 hour operation: 60	24/7 operation: 96 40 hour operation: 60	24/7 operation: 30 40 hour operation: 24
Imaging Time (One batch of 12 chips)	6 – 10 hours	12 – 16 hours	18 – 22 hours	60 – 70 hours

1. Target Effective Coverage based on 80% map rate.

2. Requires Stratys Plus Chip.

3. Maximum throughput on one Stratys system; assuming that enough samples are available to run at 100% capacity. Typical performance based on various human control samples run at Bionano.

4. Refer to LIT-00017 Redefining Cell Characterization and QC for Genomic Integrity and Off-Target Monitoring and ask your Bionano contact for more details.

Optimized Reagents and Consumables for Your Applications

Choose the most efficient path to results with application-specific sample preparation kits and chips for the Stratys system.



BIONANO SAMPLE PREPARATION KITS



Application-specific kits optimized for Stratys chips.



Compatible with a wide range of sample types:
Fresh and Frozen: Cell Lines, Blood, Bone Marrow Aspirates, Tissue and Tumor

STRATYS CHIPS



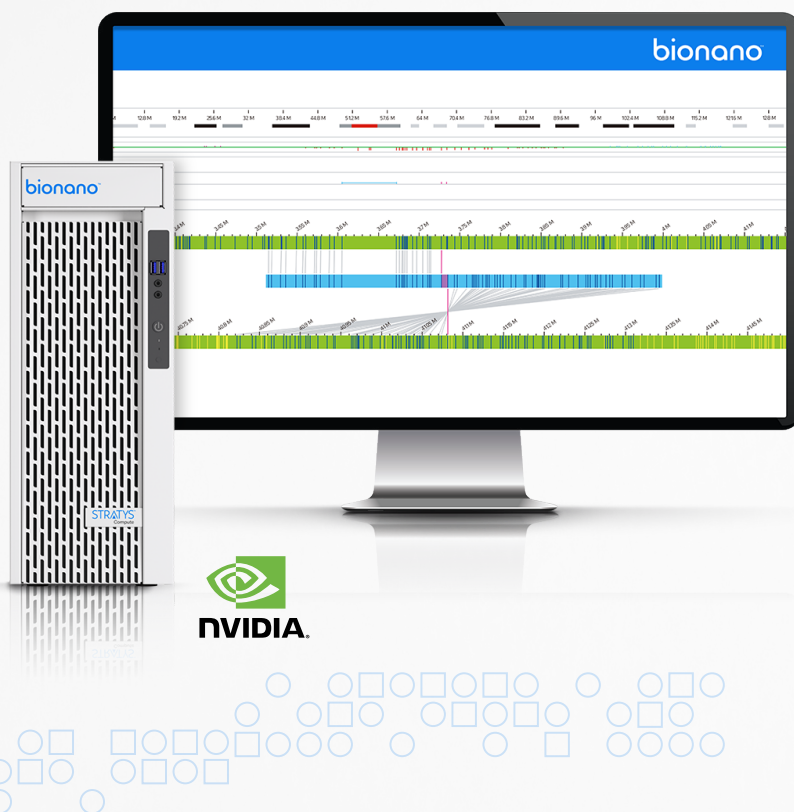
Stratys chips provide a nanofluidic environment in which DNA molecules linearize across hundreds of thousands of nanochannels where they can be imaged to reveal the underlying genomic structure and structural variation.

STRATYS CHIP TYPES AND APPLICATIONS

		Stratys Core Chip			Stratys Plus Chip			
Target Effective Coverage		100x	200x	400x	100x	200x	400x	1200x
Recommended Applications								
FSHD		●			●			
Genetic Disease	Prenatal		●			●		
	Postnatal		●			●		
	RUGD		●			●		
Oncology	Hematological Malignancies			●			●	●
	Solid Tumors			●			●	●
Cell and Gene Therapies	Stem Cells			●			●	
	iPSCs			●			●	
	T-cell and Primary Cell			●			●	●

Stay in the Flow with Streamlined Data Processing

The Stratys Compute high performance workstation and Bionano software provide the throughput to keep your workflow moving at the speed of scale.



STRATYS COMPUTE HIGH PERFORMANCE WORKSTATION



Data Processing Powerhouse

Stratys Compute powered by NVIDIA RTX GPUs delivers significantly faster runtimes and lower compute costs. Cutting-edge technology powers acceleration achieved through software releases.



Supports a Constant Flow of Data

Stratys Compute's advanced technology and intelligent processing allow a continuous flow of data from the Stratys instrument for optimal workflow scalability.



Best Sensitivity for OGM

With the latest informatics pipeline for OGM data, Stratys Compute offers the best sensitivity for genome-wide detection of all classes of SVs using OGM.

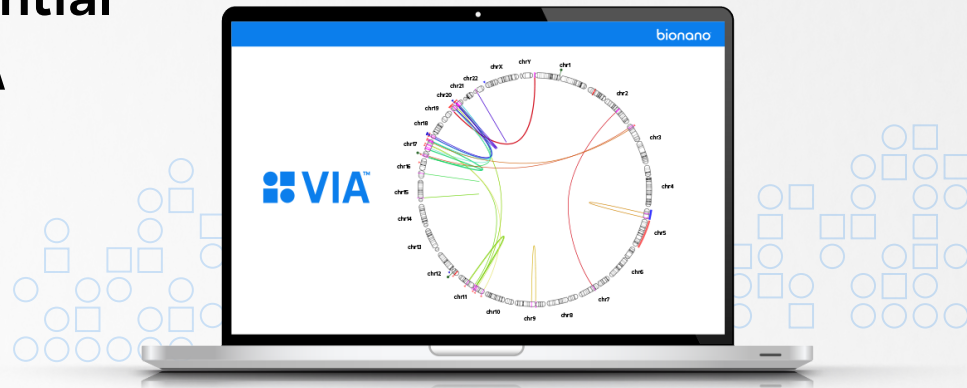


Minimal IT Resources Required

The Stratys Compute workstation is installed alongside the Stratys instrument and does not require installation in a data center, reducing IT burden and implementation time.

Harness the Full Potential of Your Data with VIA

Variant Intelligence Applications™ (VIA) software empowers your lab to deliver meaningful insights from across technologies—no bioinformatician required.



BIONANO VIA™ SOFTWARE



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

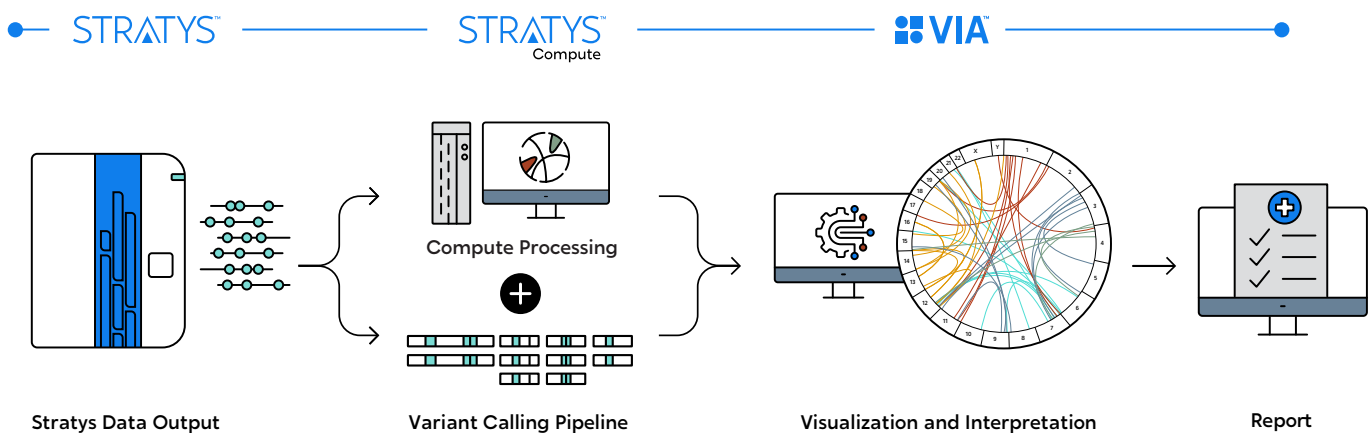
Deliver clear, highly visual reports that support informed decision-making.



Integrate

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

OGM DATA FLOW



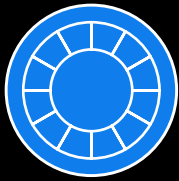
Structural Variation Detection at Scale with Stratys



Detect SVs
starting at
500 bp



VAF of
5%
or lower*



Scalable throughput
with capacity for
12 Samples
simultaneously



Up to
1200x
target effective
coverage*

*Lower VAF percentages can be achieved when using the Stratys Plus Chip, which enables up to 1200x target effective coverage.

Move Forward with Bionano



OPTICAL GENOME
MAPPING



NUCLEIC ACID
PURIFICATION



ANALYSIS
SOFTWARE

 STRATYSPHERE



Easy to Implement.
Minimal Training Required.
Get Started with Stratys Today.



orders@bionano.com



1.858.888.7600



Learn more at bionano.com/Stratys-Systems

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CG-00078_Rev A_Stratys Brochure_Effective Date 1/30/2024



Ionic® Purification System for FFPE Samples

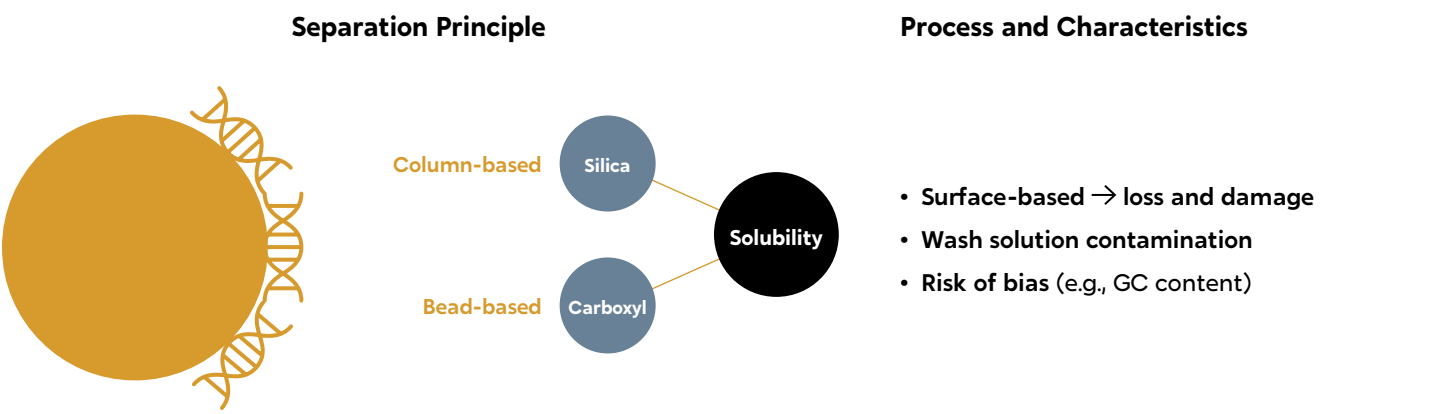


Why We Developed a New Approach to Nucleic Acid Purification

The commonly used bead- and column-based extraction technologies have followed the same fundamental workflow for over 20 years. This workflow uses ethanol, chaotropic salts, and other solutions to bind nucleic acid to a silica membrane or surface-labeled bead, which is then washed prior to the nucleic

acid being stripped off the solid support into an elution buffer. During this typically laborious process, the nucleic acid may be denatured, dehydrated, and fragmented. The eluate is also susceptible to contamination from wash buffers or beads.

Traditional Purification



Disadvantages

Incomplete binding to or removal from the solid support	• Nucleic acid loss compromises data quality when sample input quantity is limited • Recovery can be biased by fragment length or GC content • For researchers, reduced biological insight • For clinicians, less actionable information and false negatives
Contamination from wash buffers and bead coatings	• Low purity leading to false negatives and compromised data
Workflow with multiple hands-on steps	• Throughput bottleneck and potential errors; excessive use of disposable tips and labware

Isotachophoresis, a Superior Approach to Nucleic Acid Separation

Isotachopheresis (ITP) separates and concentrates charged molecules in solution solely based on their electrophoretic mobility. Biological samples are gently lysed and added to the Ionic® Fluidic Chip. An electric field is then applied to the chip and the nucleic acid is isolated in its natural, native form. The nucleic

acid is not denatured or dehydrated, and there's no binding to, or stripping from, fixed surfaces. The result is a higher yield of pure nucleic acid that is less fragmented and free from bead or wash buffer contamination.

Isotachopheresis

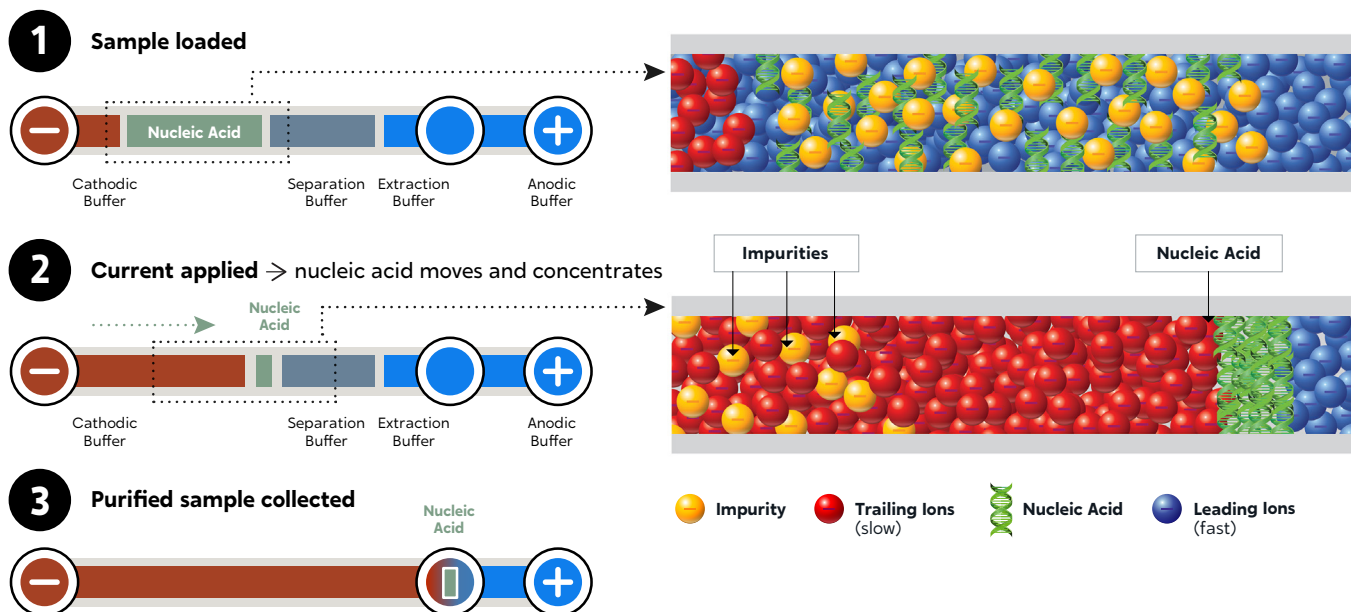
Separation Principle



Process and Characteristics

- Solution-based → high yield and integrity
- High purity
- Best representation of sample
- Result = better data

Simple, Automated Charge-based Sample Prep in Solution

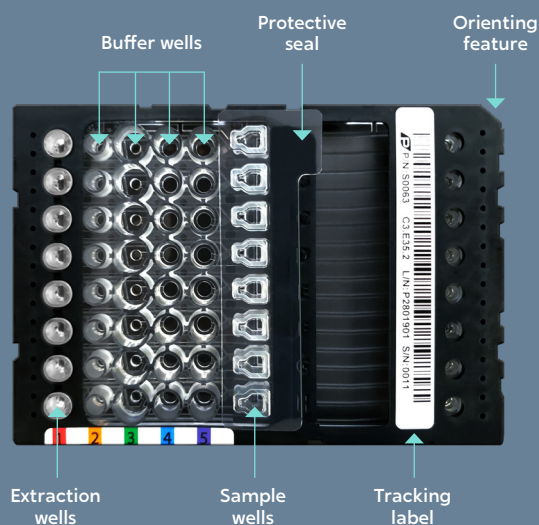
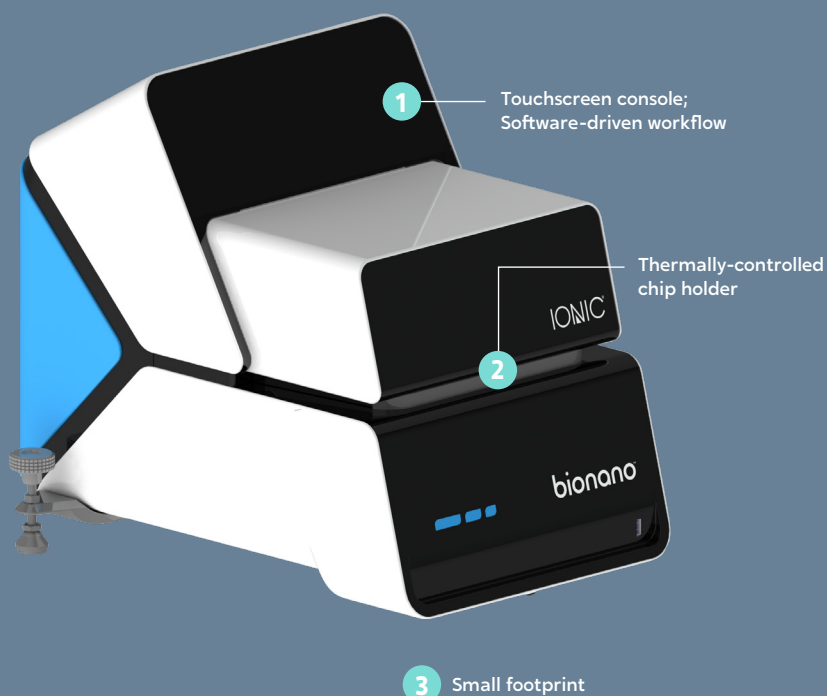


Ionic[®] Purification System

How it Works

The Ionic[®] Fluidic Chip is placed on the Ionic system and separation buffers are loaded. The chip is then primed. Next, biological samples are added into the 8 sample wells and purification using isotachopheresis begins. By applying an electric field across the length of a chip microchannel, the Ionic system separates and

concentrates nucleic acid between buffers with higher and lower mobilities. Impurities fall behind the low mobility buffer and are separated from target nucleic acids. As target nucleic acids pass through the channel, an integrated sensor stops the current once nucleic acids reach the extraction well.



The Next-Gen Sample Preparation System

The revolutionary Ionic Purification System requires no binding, stripping, or washing from fixed surfaces for higher yields, higher quality nucleic acids, and ultimately, better data for your research.

- No organic solvents
- No harsh, high-salt buffers
- No system programming
- No beads or repetitive washing
- No hands-on mixing, separation, sample transfers, or buffer exchanges
- No pumps, valves, or other moving parts

Rapid Purification of Precious Samples in Just ONE Hour

Ionic System Workflow

1

Load Run Buffers



Add buffers from reagent kit –
use touchscreen to start priming

8 mins
(manual)

2

Load Samples



Load 8 samples –
use touchscreen to start the run

65 mins
(automated)

3

Get Purified Nucleic Acid



Collect samples when touchscreen displays
"Run Complete"

2 mins
(manual)



< 5 mins
Total Hands-on Time
per Sample



75 mins
Total Run-time

Simplified Nucleic Acid Preparation

The Ionic system is so different, its advantages are most readily understood in contrast with conventional nucleic acid extraction and purification methods:



Higher nucleic acid yields

No sample loss associated with binding nucleic acids to, or stripping from, fixed surfaces



Simple workflows with fully automated separations

No columns or beads and no repetitive washing



Reduced nucleic acid fragmentation

No harsh high-salt buffers or organic solvents

Simplified FFPE Workflow Saves Time and Money

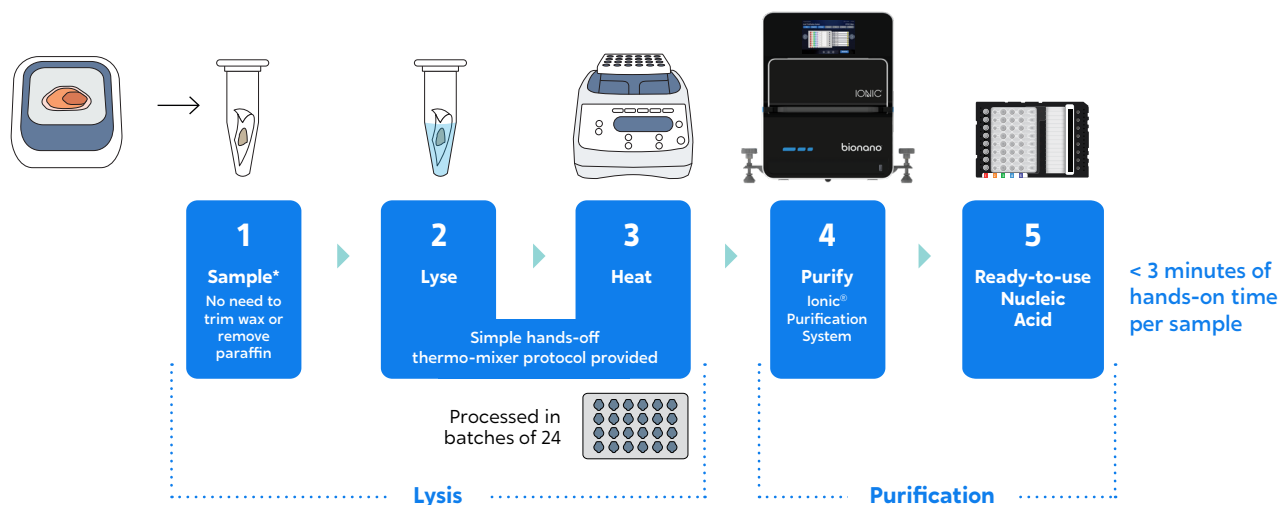
Purigen's FFPE protocols greatly simplify the processing of FFPE samples. For example, the Ionic® FFPE to Pure DNA protocol reduces the hands-on time to less than

3 minutes per sample and enables working directly from scrolls. The protocol also eliminates the need for a separate paraffin removal step.

Column-based Kit Workflow



Isotachopheresis Workflow



*Compatible with scrolls or slides

Flexibility for Working with Scrolls or Slides

The Ionic system produces more DNA and RNA from FFPE samples without requiring the use of slides (slide use and microdissection is optional). The ability to obtain comparable nucleic acid yields when using

scrolls (versus slide mounted FFPE slices) greatly simplifies the workflow when sample micro-dissection is not required. This allows projects to be completed faster and at a lower cost.

Superior Nucleic Acid Recoveries from FFPE Samples

A vast majority of clinical samples used in oncology research are stored as FFPE tissues, which often contain degraded or fragmented nucleic acid. Conventional extraction methods are labor intensive and can

further damage nucleic acid during the extraction and purification process. The Ionic system simplifies and accelerates nucleic acid purification, resulting in higher yields of higher quality DNA.

Higher yields of DNA from FFPE Samples

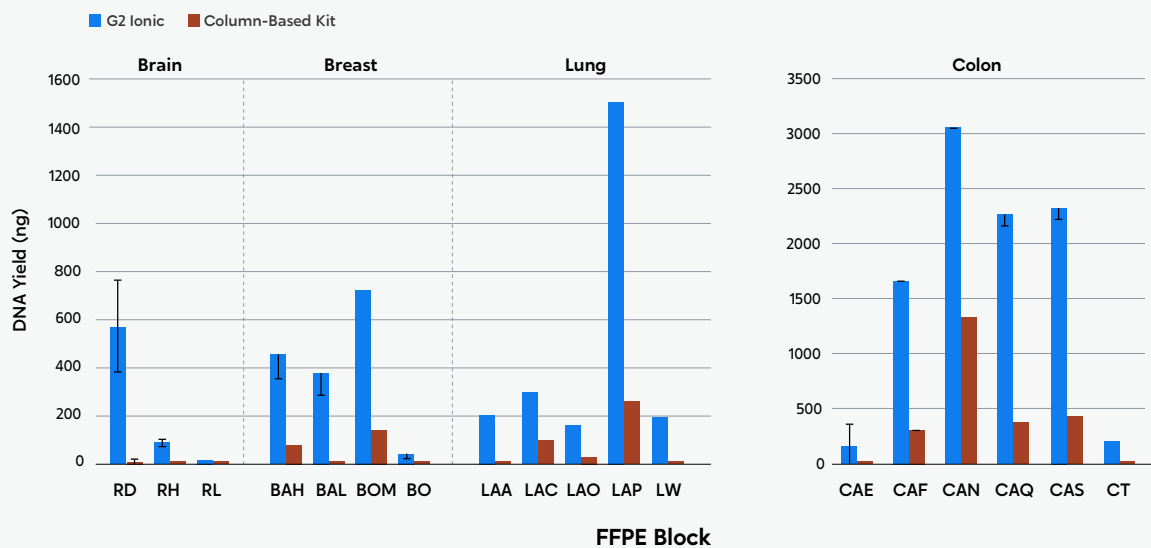


FIGURE 1: Comparison of DNA yields from consecutive sections of 18 FFPE tissue blocks purified on the Ionic Purification System using the G2 FFPE DNA kit or a commercially available column-based kit. The total DNA yield for each sample was determined by multiplying the extract volume by the DNA concentration derived from the Qubit 1x dsDNA High Sensitivity assay. Error bars represent standard deviation from the average DNA yield. More DNA is recovered using the G2 FFPE DNA kit for each of the 18 FFPE blocks with expected increase in DNA yield of 3-5x on average compared to the column-based kit, with greater yield advantages for lower yielding samples.

Nucleic acid yields from scrolls using the Ionic Purification System are on average 3.5x higher when compared to yields from a column-based kit.

Higher Quality DNA vs. Column-based Kits

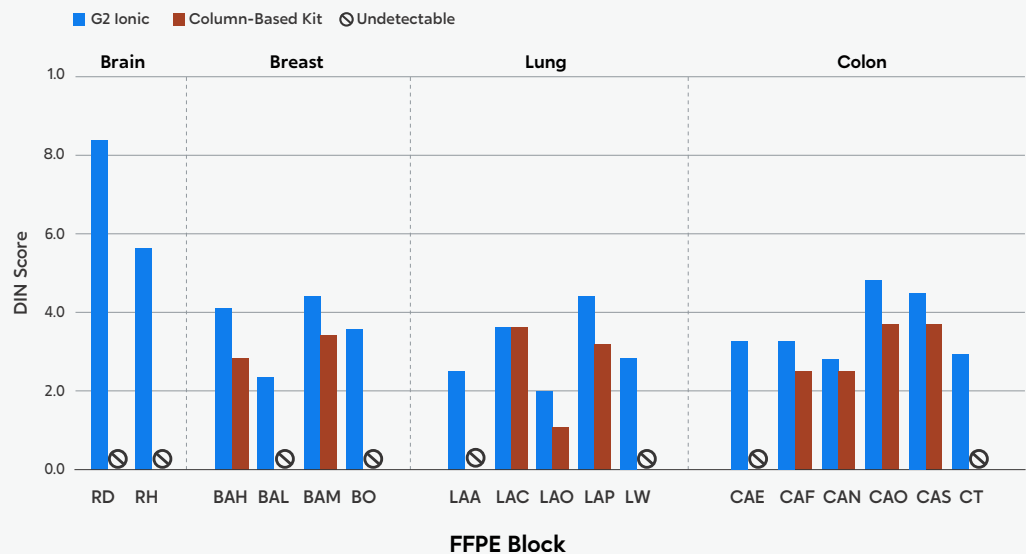


FIGURE 2: Comparison of DNA from consecutive sections of 18 FFPE tissue blocks purified on the Ionic Purification System using the G2 FFPE DNA kit or a commercially available column-based kit. The DNA Integrity Number (DIN) was calculated for each extract using a Genomic DNA ScreenTape on the Agilent TapeStation. The Ionic G2 DNA quality is typically higher than the column-based competitor, and nearly half of the column-based eluates had a concentration that was below that required for DIN score calculation.

Improved NGS Data Quality from FFPE Samples

In addition to increased yields and a greatly simplified workflow, data quality is also improved. Data below shows purified DNA from FFPE samples analyzed using the Agilent SureMASTR Tumor Hotspot sequencing panel which includes 252 amplicons ranging in size from 128–245 bps. To highlight coverage differences related to the sample purification technology, all data was normalized to a reference sample to

remove the effect of coverage differences introduced by differences in target amplicon amplification efficiencies. The reference data set was generated using the average results obtained from the purification of a high-quality sample using both Ionic system and column-based techniques (as such the reference sample is not biased to either technology).

More Uniform Coverage vs. Column-based Kits

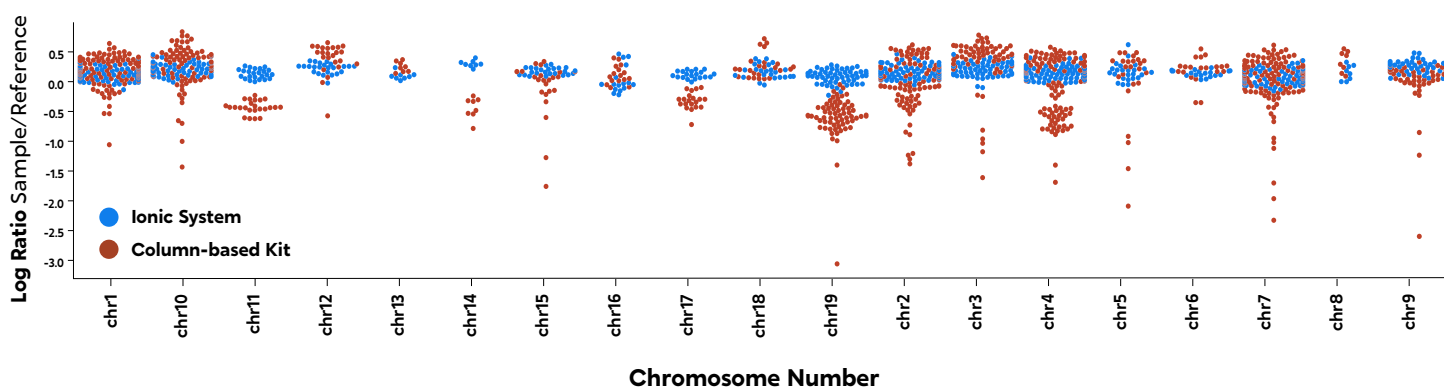


FIGURE 3: Results shown on a chromosome level. Ionic system eluates show tighter clustering more centered around the zero line. This is indicative of more uniform sequencing coverage.

Less Coverage Bias vs. Column-based Kits

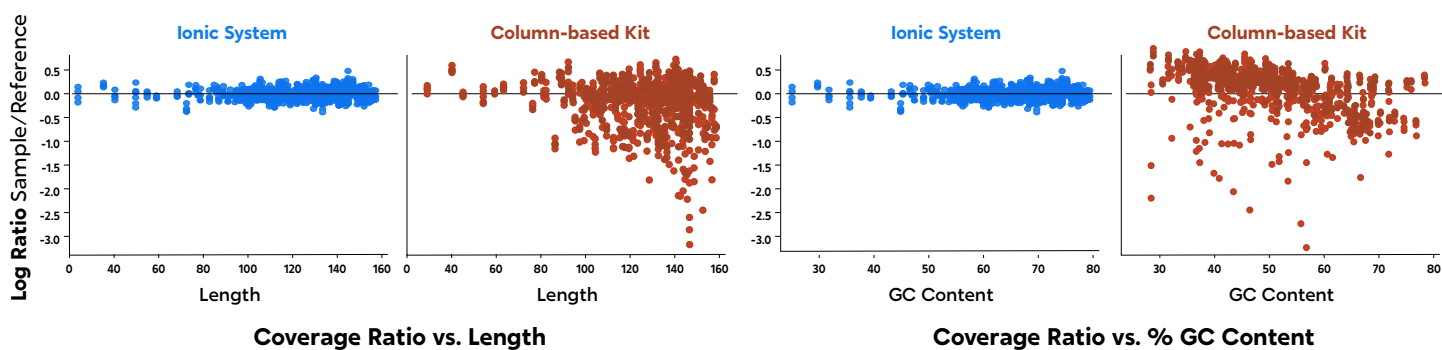


FIGURE 4: Results shown relative to amplicon length and amplicon GC content. Ionic system samples show superior uniformity for both amplicon length and GC content, with very little deviation from the expected coverage. Column-based purification shows a bias towards shorter amplicon lengths and lower GC content amplicons.

- Ionic system purification shows no bias towards amplicon length.
- Ionic system purification shows no bias towards GC content.

A Better Solution for RNA from FFPE Samples

The Ionic Purification System provides for the automated purification of RNA from FFPE tissue samples with less hands-on time than conventional bead and column-based methods. To help scientists overcome the sample preparation bottleneck

commonly associated with FFPE samples, the Ionic system provides a simple workflow that co-purifies both mRNA and miRNA with higher yields versus column-based extraction kits.

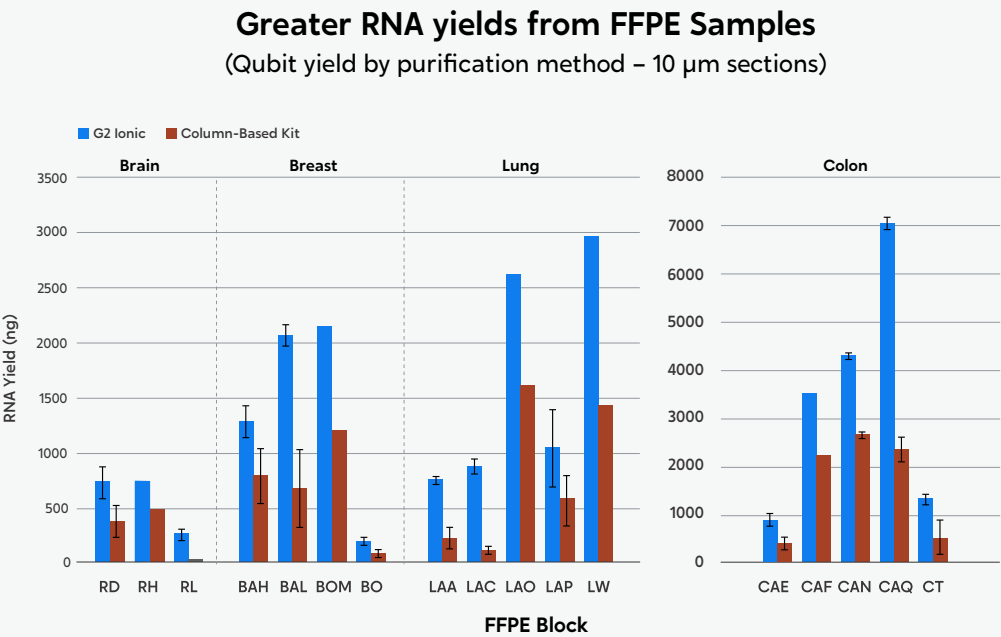


FIGURE 5: Comparison of RNA yields from consecutive sections of 17 FFPE tissue blocks purified on the Ionic Purification System using the G2 FFPE RNA kit or a commercially available column-based kit. The total RNA yield for each sample was determined by multiplying the extract volume by the RNA concentration derived from the Qubit HS RNA assay. Error bars represent standard deviation from the average RNA yield. More RNA is recovered using the G2 FFPE RNA kit for each of the 17 FFPE blocks with expected increase in RNA yield of 2-3x on average compared to the column-based kit

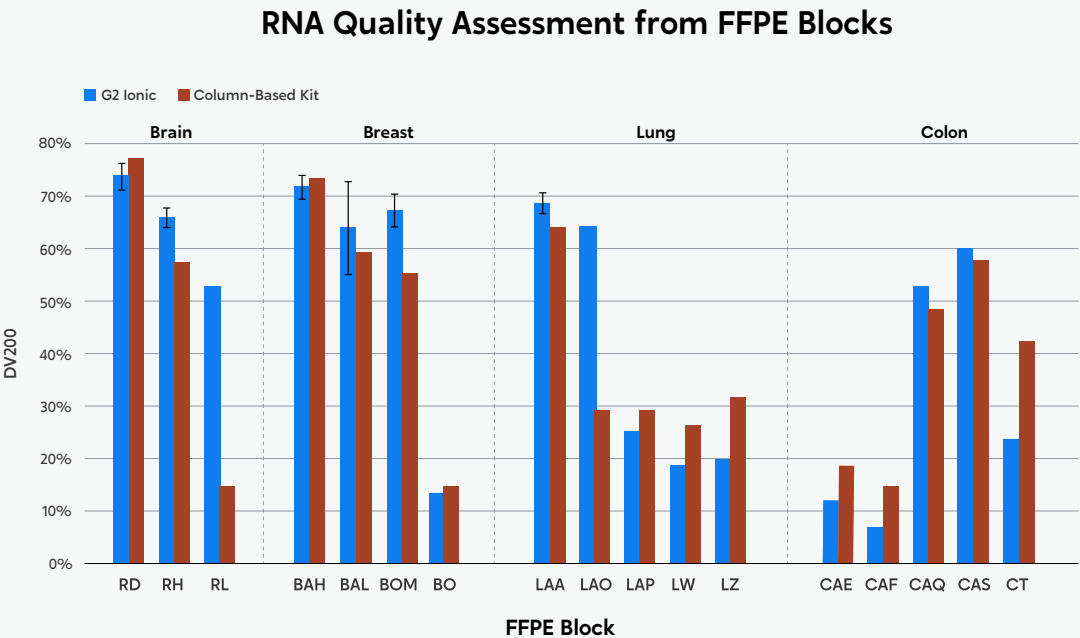


FIGURE 6: Comparison of RNA quality from consecutive sections of 17 FFPE tissue blocks purified on the Ionic Purification System using the G2 FFPE RNA kit or a commercially available column-based kit. All RNA extracts were normalized to 1.5 ng/ul input concentration and the DV200 (percentage of RNA fragments >200 bp) was calculated using a High Sensitivity RNA ScreenTape on the Agilent Tapestation. For higher quality FFPE blocks (DV200 >30%) the G2 Ionic RNA quality is consistent with or higher than the column-based kit. Since the Ionic purification process co-extracts short RNAs, such as miRNA, the G2 Ionic DV200 values may appear lower than the column-based kit for especially poor quality FFPE blocks (DV200 <30%).

Get Higher miRNA Yields from FFPE Samples

For researchers studying either the relationship between gene expression and microRNA expression or focusing purely on microRNA expression in FFPE tissue samples, the Ionic system provides more miRNA than the market-leading column-based miRNA kit. More

impressive, is that no additional steps are required. The Ionic FFPE to Pure RNA kit produces both mRNA and more miRNA from FFPE samples in a single, simple workflow.

Total RNA Purification with Higher Yields of miRNA

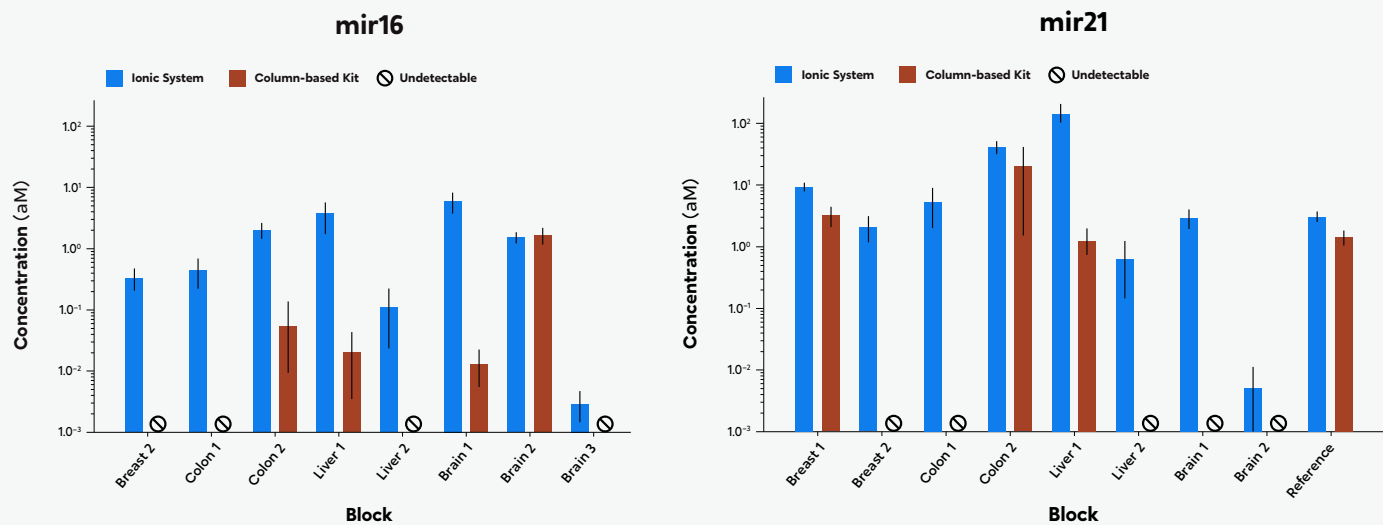


FIGURE 7: RNA from replicate sections of 8 FFPE sample blocks were purified using either the Ionic FFPE to Pure RNA Kit or a column-based miRNA extraction kit. The extracted and purified samples from each kit were analyzed by qPCR and the Applied Biosystems TaqMan Advanced miRNA Assays for miR-16 and miR-21. The concentration of the target miRNA represented in each sample was extrapolated and plotted against the tissue type of the source FFPE sample block. The Ionic system produced samples with a higher concentration of miRNA in all but one of the samples tested. For several samples the column-based miRNA extraction kit did not yield a detectable amount of miRNA.

Reproducible miRNA Expression Profiles

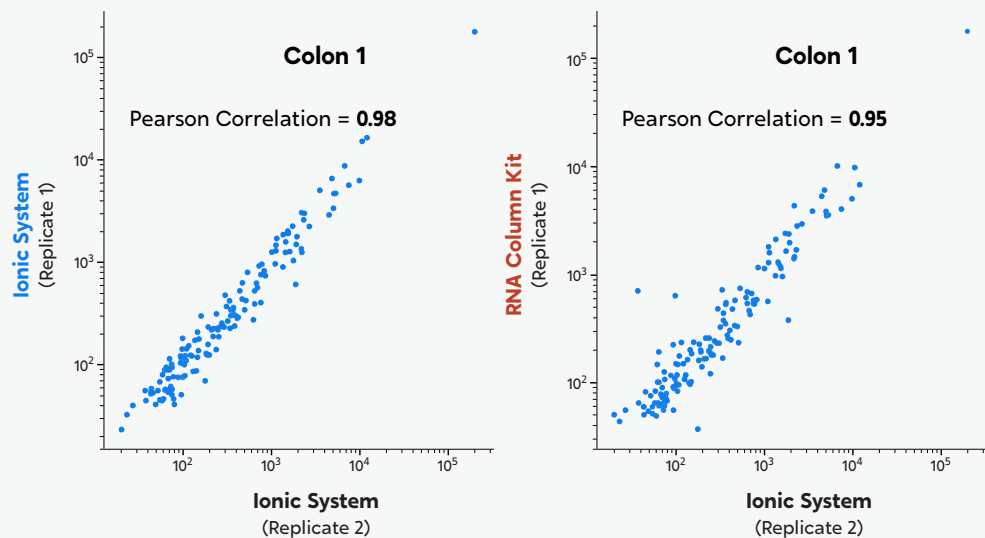


FIGURE 8: Samples from "Colon 1" of FIGURE 9 were analyzed for miRNA expression using the NanoString nCounter Human miRNA panel. The level of miRNA expression between replicate samples purified using the Ionic system has a Pearson correlation of 0.98. The level of miRNA expression between replicate samples purified using the Ionic system and the column-based miRNA kit has a Pearson correlation of 0.95. This analysis indicates a high reproducibility of miRNA expression across replicate samples purified using the Ionic system that is comparable to that of the column-based miRNA extraction kit.

Simultaneous Extraction of RNA and DNA from FFPE

The Ionic® FFPE Complete Purification Kit is used with the Ionic system to enable the automated purification of DNA and RNA, including microRNA from FFPE tissue samples. The kit provides a protocol, Ionic® Fluidic Chips and reagents to enable the Ionic system to automate DNA and RNA purification using an

innovative isotachopheresis technology. Samples are prepared for purification on the Ionic system using a simple lysis procedure that can be automated using a programmable thermomixer without any need for micro-dissection or de-paraffinization using harsh chemicals.

Comparison of Total Hands-on Time vs. Manual Methods

	IONIC	Manual Bead-based	Manual Column-based
Lysis time	1.5 hrs	Overnight	1 hr
RNA isolation	2 hrs	2 hrs	2.5 hrs
Lysis time	1.7 hrs	3 hrs	3.5 hrs
Total time	5.2 hrs	13 hrs	6.5 hrs
Total hands-on time	1.5 hrs	6 hrs	7 hrs

TABLE 1: In a study conducted by a third-party genomic services lab, this table shows 3 extraction methods that were used to compare the hands-on time and total time to extract and purify RNA and DNA from 8 samples. Replicate 10 µm sections of FFPE samples were extracted and purified using either the Ionic system, a market-leading manual bead-based kit, or a manual column-based kit.

Six adjacent sections of a 10 µm thickness were harvested from 6 FFPE tissue blocks containing brain, breast, colon, or lung tissue. DNA and RNA were extracted and purified from 4 of the 6 sections using the published workflow for the Ionic FFPE Complete Purification Kit. DNA and RNA were extracted and purified from the remaining sections using the published workflow for either a market-leading manual column-based kit or a market-leading manual bead-based kit.

The average estimated time to process 8 samples through the Ionic FFPE Complete Purification kit was 5 hours and 12 minutes with a hands-on time of 1 hour and 30 minutes (**TABLE 1**). This results in 11.25 minutes of hands-on time per sample to extract both DNA and RNA. The estimated time to process 8 samples through the column-based kit was 7 hours with most of that time being hands-on. This results in a hands-on time of 52.5 minutes per sample. Using a similar calculation, the hands-on time for the manual bead-based approach was 45 minutes per sample.

Improvement to DNA Yield with Comparable RNA Yield

The simplified workflow of the Ionic FFPE Complete Purification Kit provides simultaneous extraction and purification of FFPE samples without compromising yield.

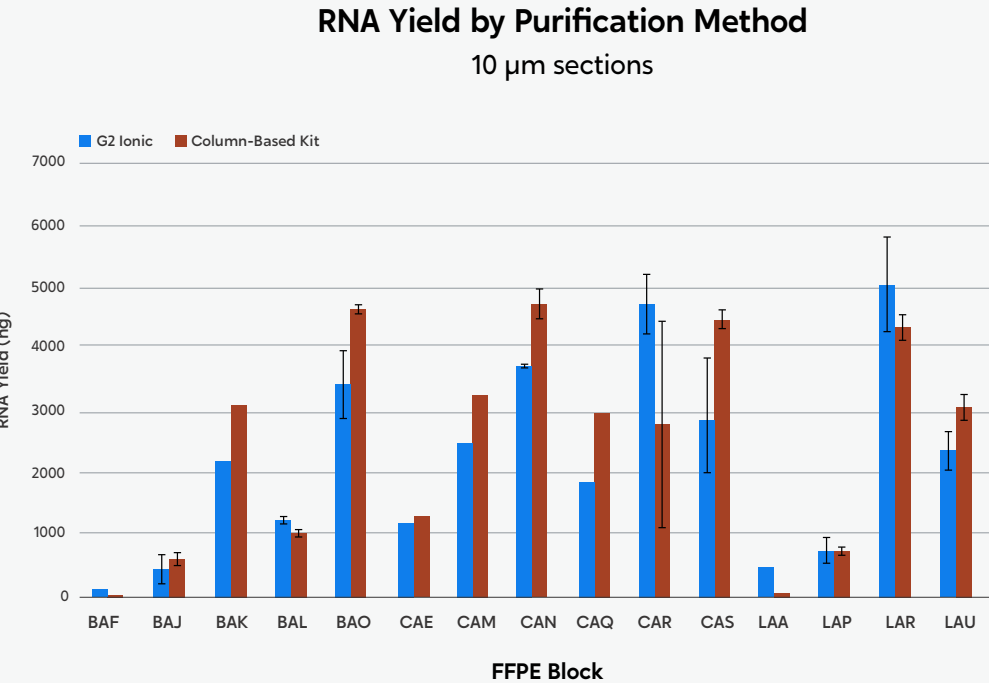


FIGURE 9: Comparison of RNA yields from consecutive sections of 15 FFPE tissue blocks purified on the Ionic Purification System using the G2 FFPE Complete kit or a commercially available column-based kit. The total RNA yield for each sample was determined by multiplying the extract volume by the RNA concentration derived from the Qubit HS RNA assay. Error bars represent standard deviation from the average RNA yield. A similar or higher amount of RNA is recovered using the G2 FFPE Complete kit for 8 of the 15 FFPE blocks with an expected RNA yield increase of ~15% on average compared to the column-based kit

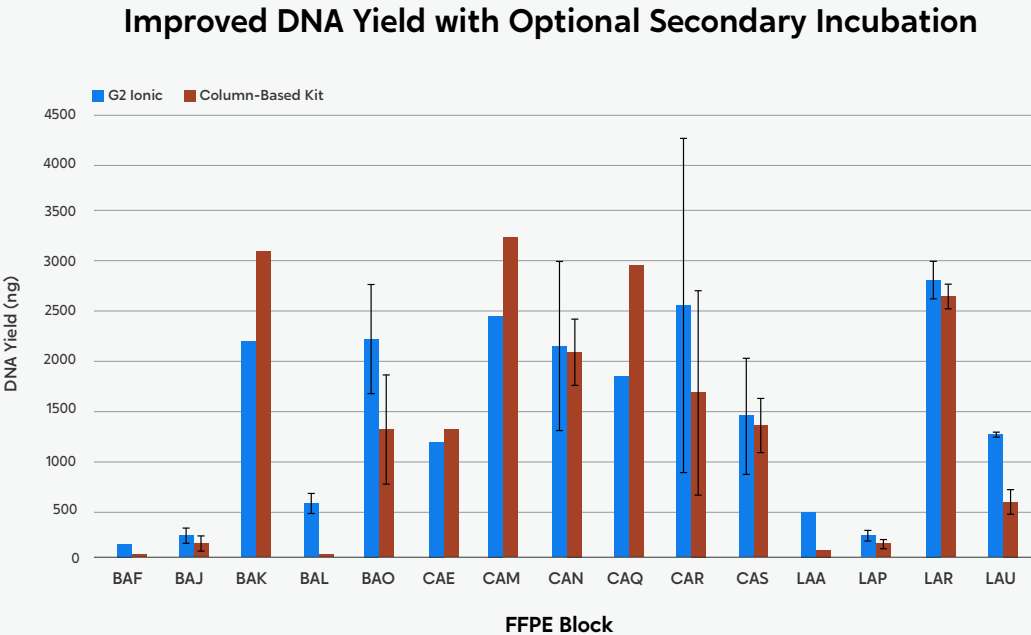


FIGURE 10: Comparison of DNA yields from consecutive sections of 15 FFPE tissue blocks purified on the Ionic Purification System using the G2 FFPE Complete kit or a commercially available column-based kit. The total DNA yield for each sample was determined by multiplying the extract volume by the DNA concentration derived from the Qubit 1x dsDNA High Sensitivity assay. Error bars represent standard deviation from the average DNA yield. More DNA is recovered using the G2 Complete kit for 12 of the 15 FFPE blocks with increased DNA yield of 1.5-2x on average compared to the column-based kit and greater yield advantages for low yielding samples.

A Better Solution for DNA Purification from Tissue Samples

The Ionic® Tissue to Pure DNA Kit provides automated purification of DNA from fresh frozen tissue samples with less hands-on time than conventional bead and column-based methods.

Comparison of Ionic Tissue to Pure DNA Kit to 3 Column-based Methods

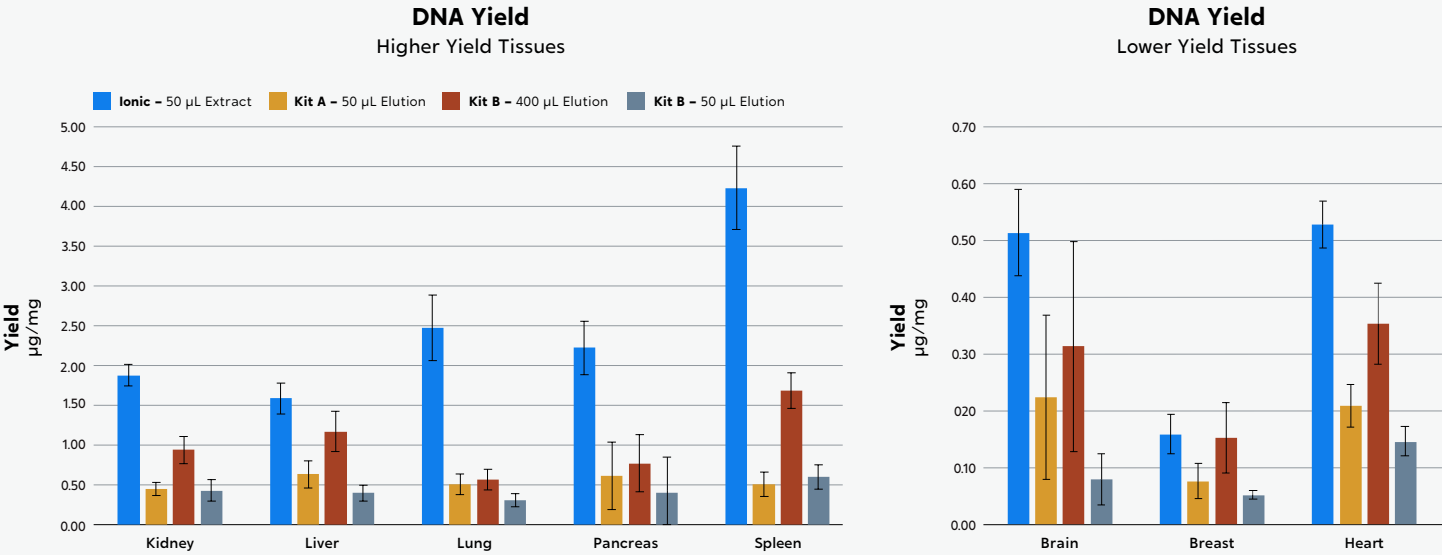


FIGURE 11: DNA extraction was performed on 8 different tissue types with four different extraction methods on a total of 632 specimens. Each tissue type/condition has an n-value ranging from 6 to 47 individual specimens. Pancreas, brain, breast, and heart specimens were derived from one donor and kidney, liver, lung, spleen were from two donors. Error Bars represent the 95% CI on the mean DNA yield (µg) per mg of tissue from replicate specimens (1-10 mg).

Linearity of Yield Across Tissue Input Amounts

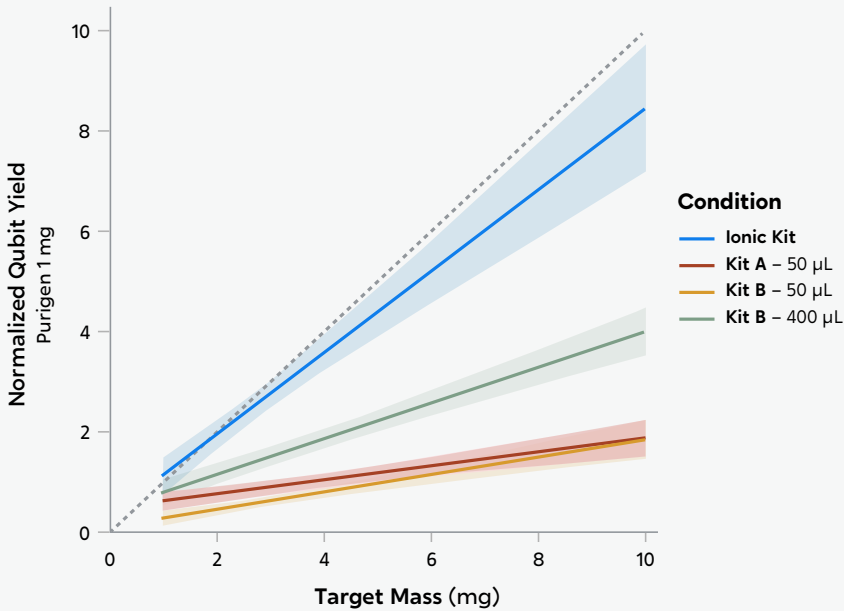


FIGURE 12: DNA yields were normalized to the Ionic kit yield for 1 mg on a by-tissue basis. The dotted line represents a theoretical linear extraction efficiency up to 10 mg tissue (assuming optimal recovery by Ionic kit at 1 mg specimens). The Ionic kit tracked the theoretical linear extraction efficiency more closely than the column-based methods.

Maximize Yields For WBCs, PBMCs, and Cultured/Sorted Cells

The Ionic® Cells to Pure DNA Kit supports a wide range of cell types including white blood cells (WBC) or peripheral blood mononuclear cells (PBMC) isolated from blood as well as cultured or sorted

cells. The standard input range is 50k to 5 million cells. Customized protocols for as few as 10 cells are available upon request.

Higher Yields for Blood-based, Cultured, and Sorted Cells

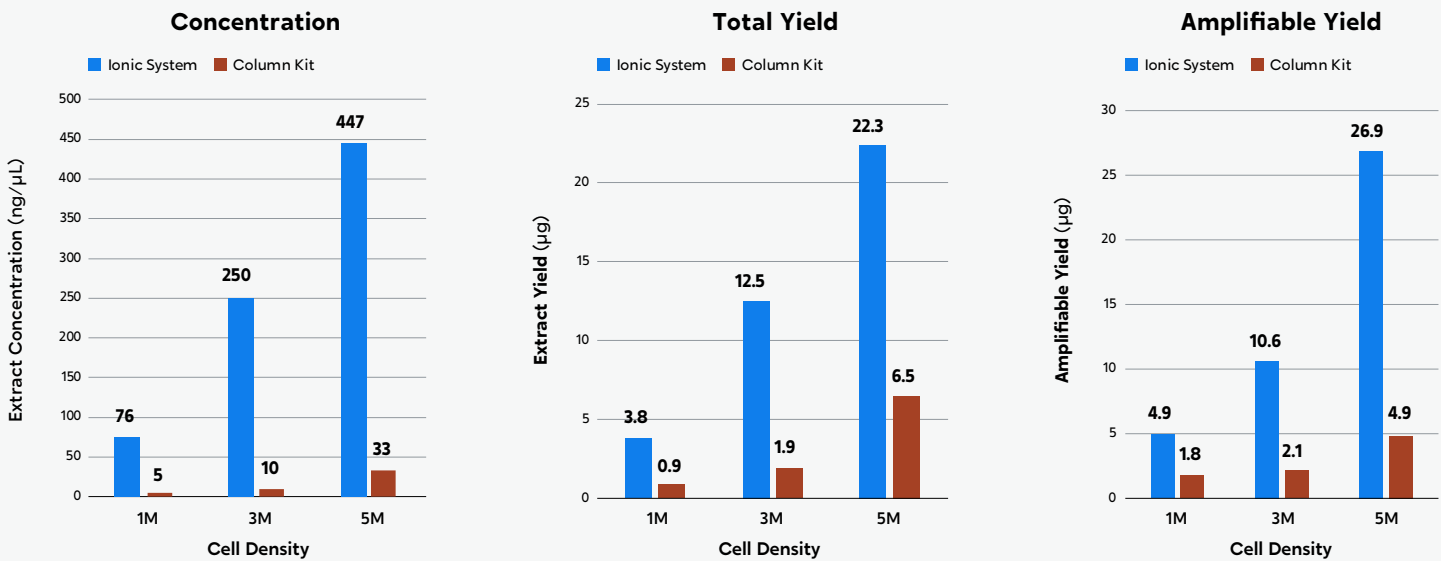


FIGURE 13: Peripheral Blood Mononuclear Cells (PBMCs) were isolated via Ficoll gradient, White Blood Cells (WBCs) were pelleted from lysed whole blood, and GM24385 cells were pelleted from culture media. Extractions were performed for each cell type at amounts ranging from 1-5 million cells then quantified via Qubit assay.

Consistent Length Profiles with Average Length Above 20k bp

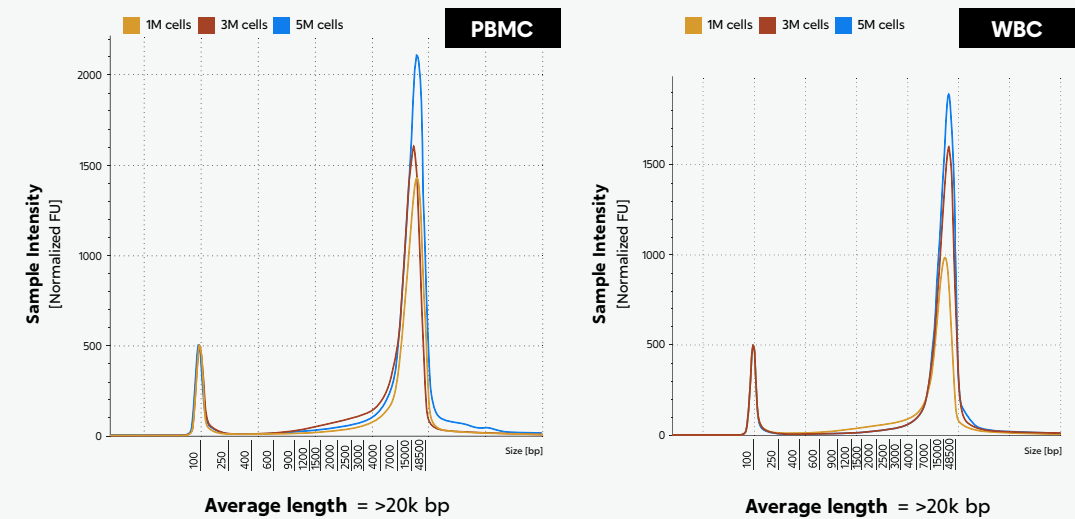


FIGURE 14: DNA Extracts from the Ionic System were evaluated on a TapeStation to determine their size profile. Size profiles were consistent across cell type and quantity.

Products

Instrument		Configuration	Part No.	
	Ionic® Purification System Includes: Power cord, installation, warranty	Standard	44001	
	Warranty Information • 12 months coverage • Initial response within 8 business hours • On-site response within 3 business days • Includes parts and materials • Parts, materials • On-site labor			
Kits		Configuration	Part No.	
	Ionic® G2 FFPE to DNA Kit Includes: Fluidic chips, room temp reagents, -20°C reagents	6-Chip Kit	 90164	
	Ionic® G2 FFPE to RNA Kit Includes: Fluidic chips, room temp reagents, -20°C reagents	6-Chip Kit	 90167	
	Ionic® G2 FFPE Complete Kit Includes: Fluidic chips, room temp reagents, -20°C reagents	6-Chip Kit	 90170	
Service Contracts		Includes	Term	Part No.
Comprehensive Service Plan		• 1 planned maintenance visit • Initial response within 8 business hours • On-site response within 3 business days • Minimum 90-day warranty on replacement parts • Parts, materials • On-site labor, per diem charges • Phone and email support • Remote support sessions • Software upgrades	12 months	44900
Planned Maintenance-only Plan		• 1 planned maintenance visit • Initial response within 12 business hours • On-site response within 5 business days • 90-day warranty on all replacement parts • 10% discount on Parts and Materials • 10% discount on on-site labor, per diem charges • Phone and email support • Remote support sessions • Software upgrades	12 months	44901
Time and Materials Service		• Hourly service work	N/A	44902

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To order, please contact orders@bionano.com or Call **1.858.888.7600** or contact your sales person directly.

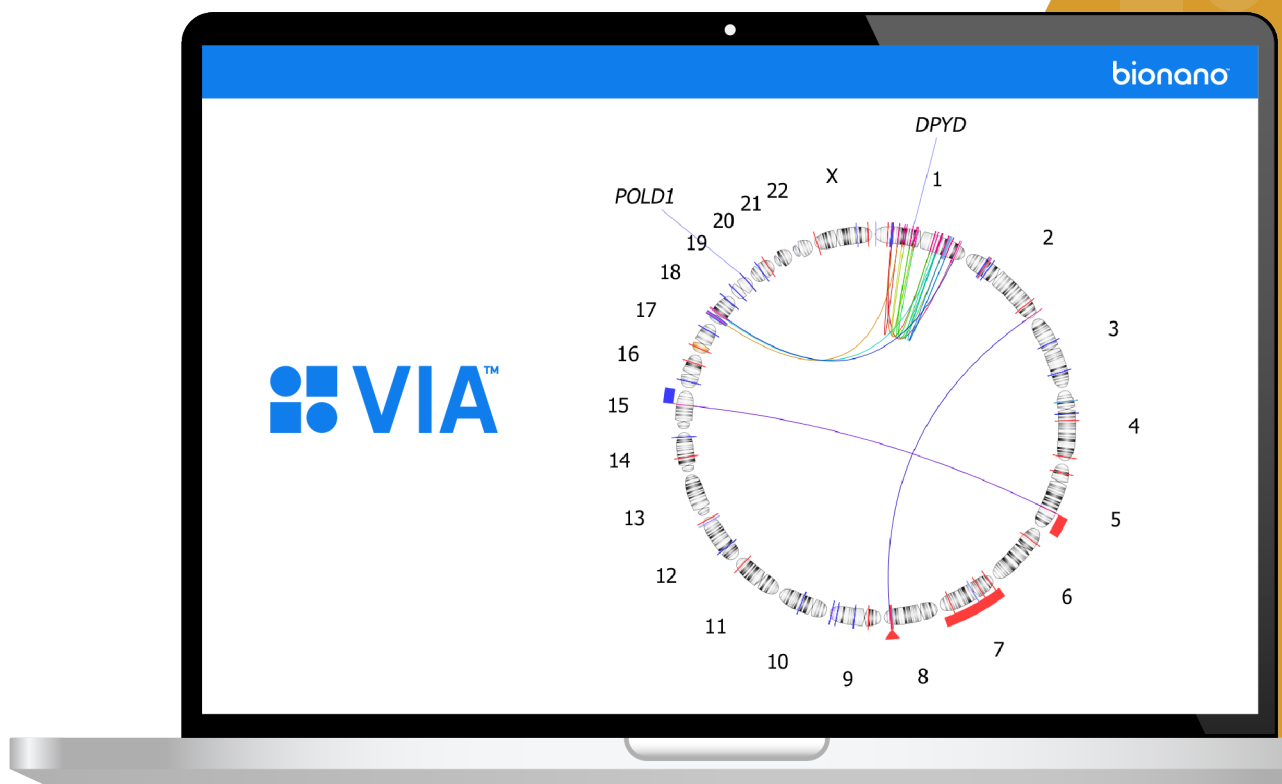
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CG-00022_Rev A_Ionic Brochure_Effective Date 04/18/2023

bionano™

**Harness the Full Potential of Your Data.
VIA™ Analysis Software is the Way.**



Introducing VIA™ Analysis Software

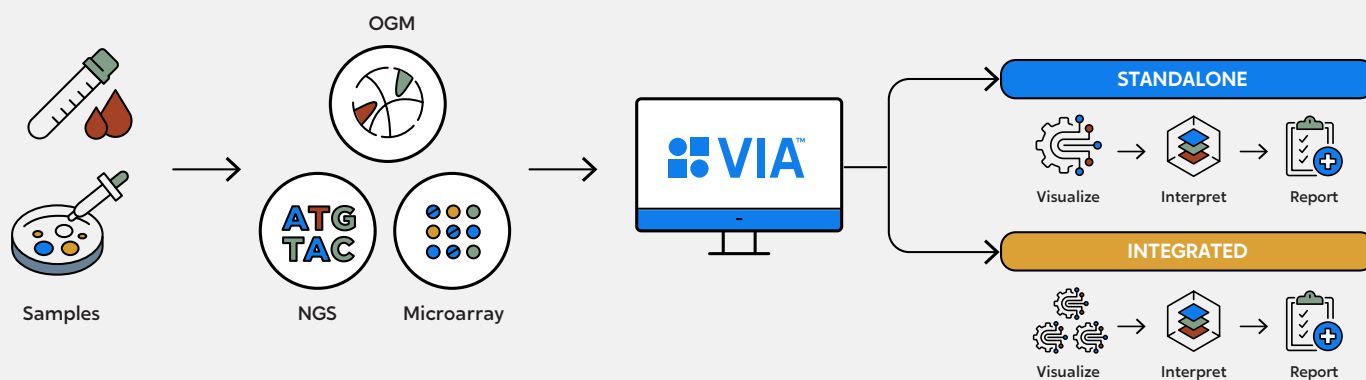
A faster and more complete genomic data analysis platform for clinical research

Existing data analysis solutions for genomics can be laborious, time-consuming and incomplete. You have to manage various systems, navigate complex workflows and spend inordinate amounts of time hunting for annotations and creating data visualizations. Good news: those days are over.

VIA analysis software radically simplifies your sample-to-report workflow and empowers you to deliver more meaningful answers from across technologies faster. VIA brings various data types into one view and automates variant calling, interpretation and annotation, so you can reduce turnaround times and optimize operations at every step.

How it Works

VIA is the only analysis software that combines secondary and tertiary analysis, receiving and interpreting data from Optical Genome Mapping (OGM), Next-Generation Sequencing (NGS) and microarrays to contextualize all classes of genomic variation and drive meaningful insights.



Experience the Benefits of VIA



VISUALIZE

Generate powerful visualizations for enhanced contextualization across multiple variant types



REPORT

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



INTERPRET

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



INTEGRATE

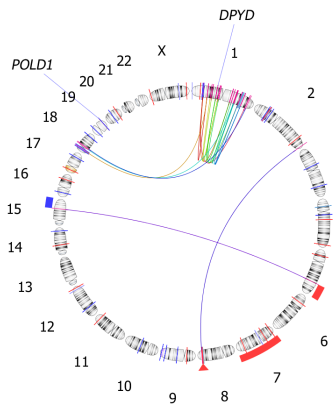
View and fully analyze data from OGM, NGS and microarray into one system for a simplified and integrated workflow

Instant Visualization, Zero Hassle. Automate Data Display for Powerful Insights.

VIA amplifies the story of your data by automatically generating powerful visualizations of different variants independently or simultaneously, so you can see them in the context of other variant types occurring in the same genomic region. VIA allows you to drill down to single chromosome, gene, and nucleotide levels for deeper analysis.

Leverage Multiple Views: Genome Browser, Karyogram, Track View and Circos Plot

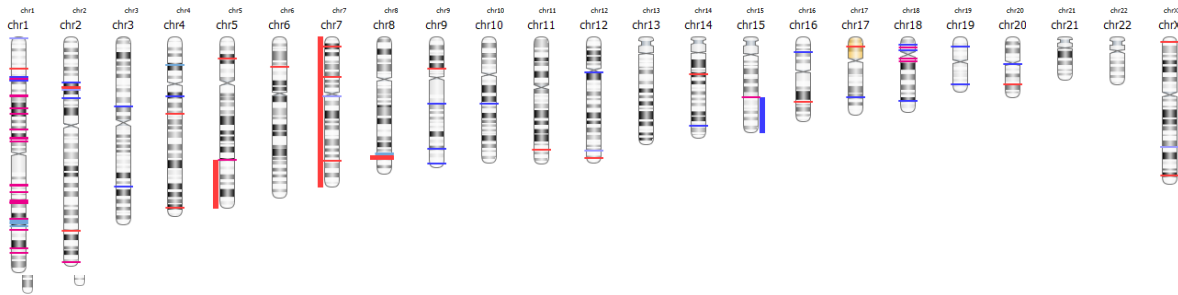
Circos Plot View



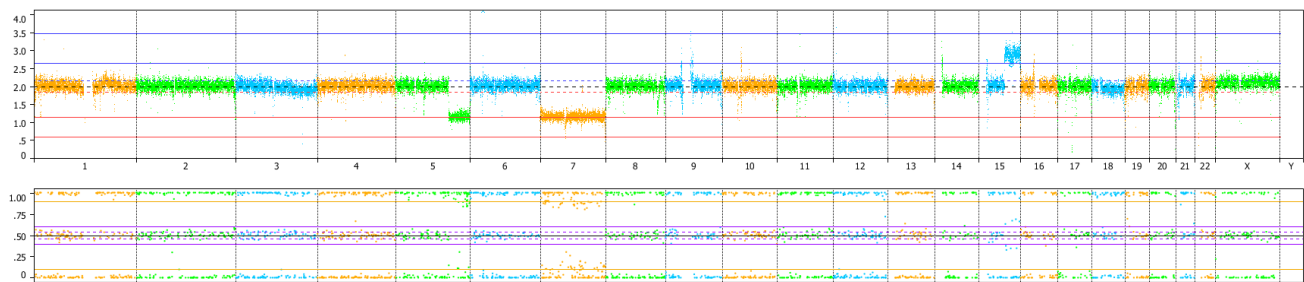
ADDITIONAL VISUALIZATIONS AVAILABLE

- Chromosome browser with multiple tracks displayed
- Variant Information details
- Detailed events table

Ideogram View



Whole Genome Representation



Representation of genomic variants in an ideogram and whole genome plots for logR intensity and B-Allele frequency from OGM data provided in VIA.



Reveal Greater Insights with Powerful Informatics

VIA provides highly sensitive detection of mosaic and non-mosaic CNV and AOH/LOH events by leveraging a new SNP-FASST3 algorithm. For OGM data, VIA offers a novel capability to extract B-Allele Frequency (BAF) for enhanced visualization and variant calling based on the allelic contribution.

Put Your Analysis on Autopilot and Get to What Matters Faster

VIA introduces intelligent automation for filtering, classifying, annotating and interpreting data across technologies, no more hours manually searching through databases. VIA accelerates time to results to drive faster, better classification of variants by pathogenicity while reducing associated costs.

ANNOTATE



Integrate gene/variant annotation from external databases and previously reported variants from historical data.

FILTER



Automatically classify variants using a rules-based engine, dynamically filter against curated datasets and remove irrelevant variants.

INTERPRET



Capture expert interpretation from analysts automatically and implement case signoffs.



External Annotation Databases*

- OMIM
- RefSeq
- DGV
- DECIPHER
- Gene Imprint
- Segmental Duplications
- ClinGen
- ClinVar
- CIVIC
- Heme Guidelines
- ACMG 2019 Guidelines
- Human Phenotype Ontology
- Custom Databases



Internal Annotation Options

- Build your own central database.
- Input your annotations.
- Look back at past samples and compare them to new sample findings.
- Curate local knowledgebase of expert interpretations.
- Link out to Genomenon Mastermind to streamline literature search.

*External resources are curated and updated in the software on a regular cadence.

Flexible Reporting that Simplifies Decision Making

Easily create reports with simplicity and clarity thanks to VIA's comprehensive set of customizable reporting options to clearly communicate results incorporating various medical society guidelines. Bring the most relevant information to the surface along with visuals and data that support decision-making and capture guideline-based evidence.

VIA also provides the option for utilizing the Hematological Malignancy and Postnatal workflows for a streamlined analysis and reporting solution for OGM data. See next page for details.

Easily Customize Your Reports

Different labs have different needs for their report structures. VIA makes it easy to create a report with only the sections you need.



A Single Platform for All Your Variant Data. VIA is the One.

VIA eliminates complex data analysis workflows by bringing data types together in one centralized platform, including your historical data from previous cases. You can seamlessly transition between technologies and all classes of genomic variation within VIA.



View and analyze OGM, NGS, and microarray data without switching between systems.



Perform analysis of structural variants, copy-number variants, allelic imbalances, single nucleotide variants and indels for enhanced discovery of causative findings that may be missed if analyzed independently.



Retain and recall historical case interpretations for faster analyses.

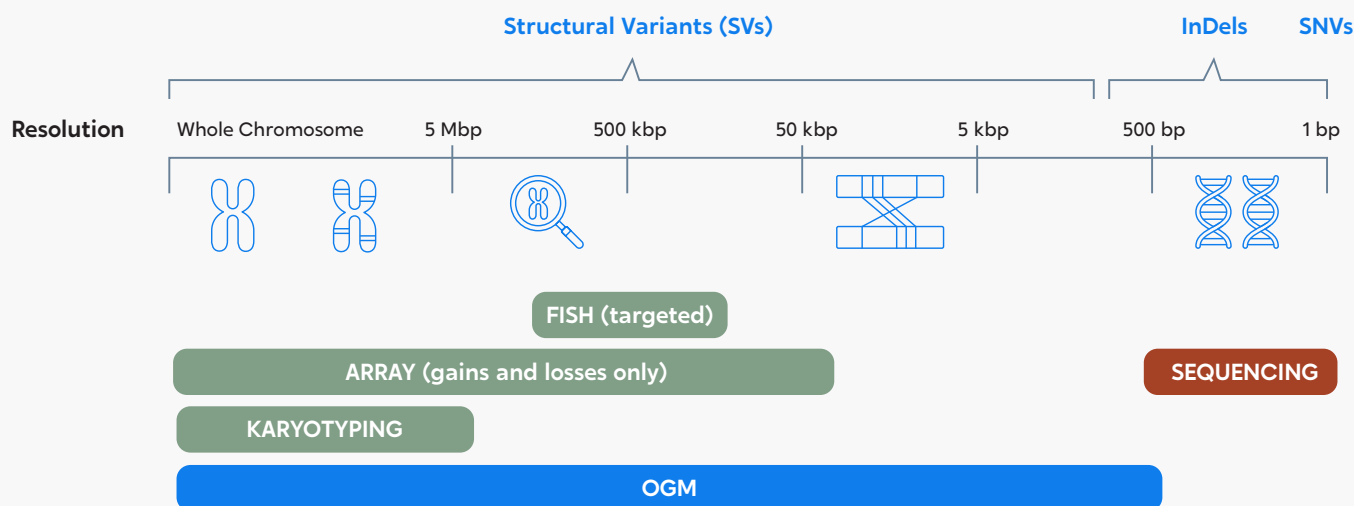


VIA ADAPTS TO YOUR NEEDS

VIA is configurable to your specific analytical needs, including cancer and constitutional applications employing OGM, NGS or array methodologies.

Generate More Meaningful Insights by Combining Genomic Data Types

Combining data from NGS with OGM gives you a very comprehensive view of the genomic variant continuum by detecting SNVs, CNVs and SVs of all classes and sizes. Bringing these two data types is akin to having cytogenetic and molecular genetic lab data in one. VIA software lets you analyze these data in one system for more powerful insights.



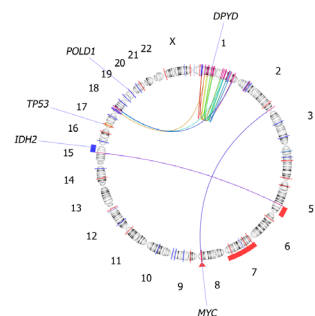
Streamline your Cytogenetic Analysis with OGM and VIA Software

Hematological Malignancies

The latest versions of Bionano™ Solve, Bionano Access™ and VIA™ software combine to deliver a comprehensive and automated workflow for analyzing, interpreting and reporting SVs, CNVs and CN-LOHs detected by OGM associated with hematological malignancies.

This end-to-end workflow uses intelligent software automation to accelerate time-to-result for hematologic cancer samples analyzed by OGM, including curated resources from key databases and guideline-based variants relevant to hematological diseases.

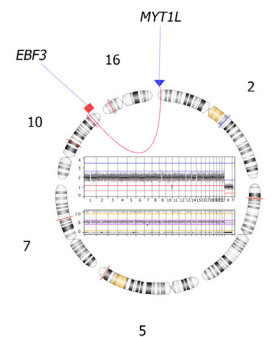
VIA™ automatically pre-classifies OGM-detected variants using a rules-based engine that evaluates pathogenicity and tiering evidence. All oncology workflows are fully configurable to meet your site-specific requirements.



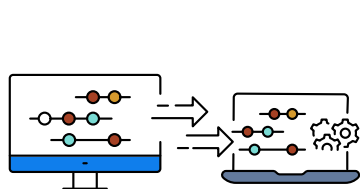
Postnatal Diseases

By unifying the investigation of SVs, CNVs, and LCSH regions at ultra-high cytogenetic resolution on a single platform, OGM data analyzed in VIA™ delivers more relevant insights than conventional microarray or karyotyping for constitutional disorders.

With intelligent automation for variant filtering, rules-based auto-classification aligned with professional guidelines, and streamlined reporting tools, VIA™ enables rapid analysis of singleton or parental duo/trio samples within minutes.

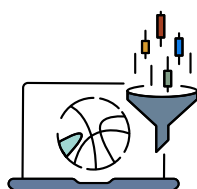


OGM & VIA Analysis Workflow



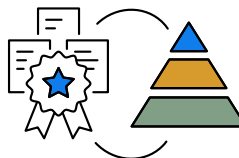
1

Connect OGM Data from Access to VIA to initiate variant pre-classification using intelligent automation



2

Review variants utilizing curated annotations, advanced visuals, and dynamic filters to assign interpretations



3

Associate Tiered variants to guidelines derived from professional communities and societies



4

Complete case sign-off and generate a configurable report to communicate results

Say Goodbye to Multiple Systems and Simplify Your Workflow with VIA™ Analysis Software



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

Move Forward with Bionano



**NUCLEIC ACID
PURIFICATION**



**GENOME MAPPING
SYSTEMS**



**ANALYSIS
SOFTWARE**

To schedule a demo or order VIA, contact
Customer Service, call or email us.



customer-service@bionano.com



1.858.888.7600

Learn more at bionano.com/via-software

Power Discovery Across the Genomic Variant Continuum with VIA™ Analysis Software

The latest release of the Bionano VIA software will bring you the only decision support analysis tool that builds critical insight across the variant continuum of single nucleotide variants (SNVs), copy number variants (CNVs), and structural variants (SVs). Get ready for a new era of fully contextualized genomic discovery.



Import data from many widely adopted genomic analysis platforms

Optical genome mapping (OGM)

Next-gen sequencing (short-read and long-read)

Chromosomal microarrays

Find more genomic variation that matters with intelligent decision support



Leverage automation to accelerate accurate analysis, classification and reporting



Gain new insight to disease impact with complete, unique visualization of genomic variants

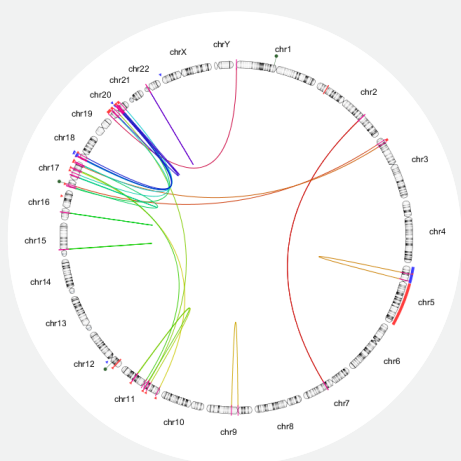


Layer multiple data types to contextualize genomic variation for better discovery and greater efficiency

QR Code

To learn more, visit bionano.com/via-software-preview

Accelerated Interpretation of Genomic Profiles for Hematological Malignancies

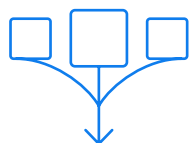


With VIA, you can use intelligent automation to quickly identify the causative variants associated with hematological malignancies leveraging key new software features, including variant autoclassification, dynamic filtering, and comprehensive visualization of all classes of genomic variants along with streamlined report generation for a complete analysis workflow.

VIA circos plot (left) visualizes all classes of variants: Structural variants (lines), copy number variants (bars), sequence variants (pins)



Chromosome browser view overlays OGM and NGS data, identifying a genomic region impacted by a translocation, loss, and sequence mutation.



Enhance discovery of causative findings with integrated analysis of structural variants with sequence variants



Perform disease-specific or pan-cancer analyses by associating variants with guideline-based cytogenomic findings



Achieve faster turnaround times with efficient analysis workflows and flexible reporting capabilities

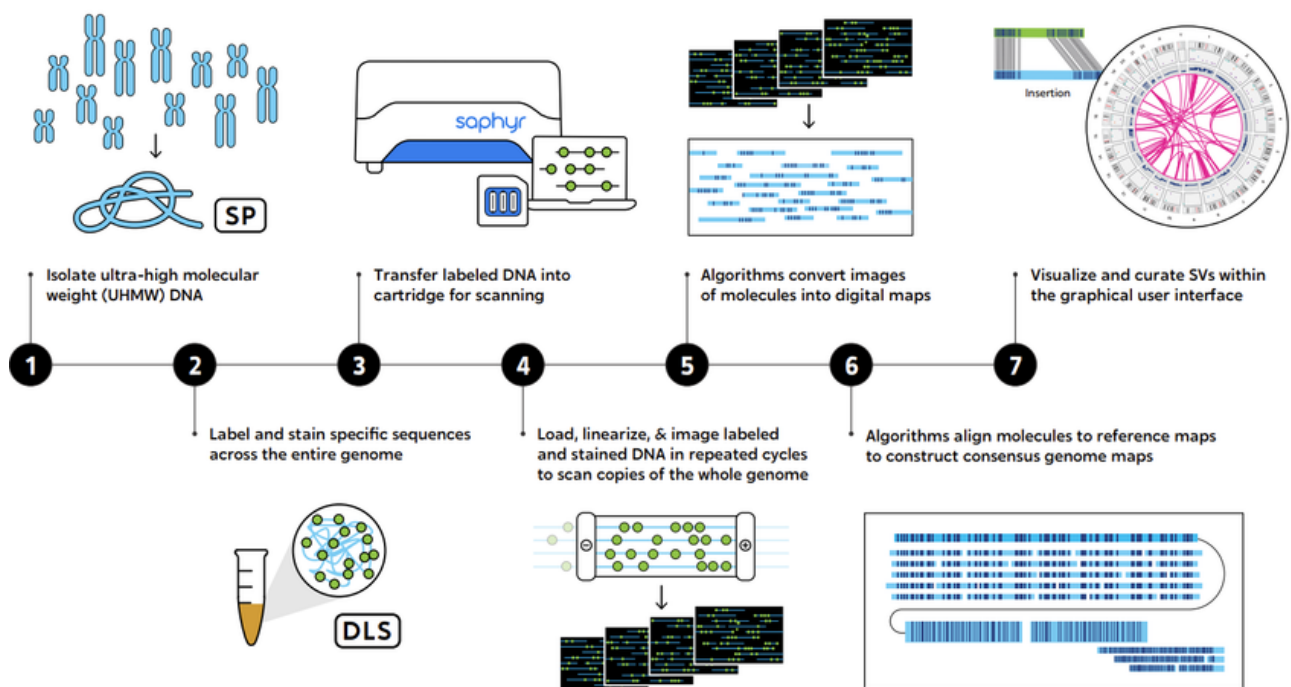
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Bionano Compute Solutions: Flexible and Scalable On-premise and Cloud Computing Options

Bionano compute solutions include 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 On Demand simplifies the way you perform genome assembly, hybrid scaffolding and structural variant analysis, without the need of any additional infrastructure, giving you the flexibility and scalability your experiment deserves.

SAPHYR® WORKFLOW



OVERVIEW OF COMPUTE OPTIONS



COMPUTE SERVER

- Control compute costs with on premise solution
- Execute consistent workloads
- Internet access not required
- Reliable access to compute resources



COMPUTE ON DEMAND

- Execute variable workloads
- No upfront server costs required
- Data centers compliant with IPAA, CSA, SOC2, ITAR regulations
- Work on large genomes up to 24 Gbp genome size

Typical Ranges for Bionano Solve Processes

HUMAN	North America Tokens		Germany Tokens		Europe Tokens		Typical Job Processing Time**
Compute On Demand	typical	min/max	typical	min/max	typical	min/max	
De novo assembly*, 400 Gbp	9	1/22	22	1/38	10	1/29	12-24 hours
De novo assembly*, 800 Gbp	12	1/25	25	1/45	11	1/31	18-50 hours
Rare Variant Analysis, 1500 Gbp	8	3/38	24	3/38	8	3/38	10-12 hours
Rare Variant Analysis, 5Tbp	30	5/75	95	5/125	30	5/75	15-30 hours
EnFocus™ FSHD/Fragile X	4	1/8	7	3/8	4	1/8	2-4 hours
Guided Assembly, Constitutional 800Gbp	10	3/38	17	3/64	10	3/38	8-20 hours
Guided Assembly, Low Allele Frequency 1500Gbp	11	3/38	12	3/64	11	3/38	10-20 hours

Compute Server, Gen4	Typical Time to Complete†	Typical Sample Throughput††	
		ONE SERVER	TWO SERVERS
De novo assembly*, 400 Gbp	10-16 hours	10-16 per week	20-30 per week
De novo assembly*, 800 Gbp	13-20 hours	8-12 per week	20-30 per week
Rare Variant Analysis, 1500 Gbp	5-8 hours	20-30 per week	40-60 per week
EnFocus™ FSHD/Fragile X	1-2 hours	Up to 60 a week	N/A

Token ranges provided here are typical for good quality data, defined as having Map Rate > 80%, molecule N50 (>20kbp) > 180kbp. Actual min/max ranges are calculated for each submitted compute job based on data quantity and quality.

*de novo assembly runs without a reference will require more processing to first generate an automatic rough assembly. The "max" may vary based on quality of input data

**Time to complete hours are estimated based on the users of the current version of Bionano Access and availability of cloud resources. Queue time to initiate processing with cloud resources can vary significantly.

†Typical time to complete hours are estimated based on internal tests processing good quality data using the current version of Bionano Access.

††Estimated sample throughput based on typical processing time of good quality data with network access to compute resources spanning 7 days per week.

	Part No.	Product
Computing Hardware	80083	Saphyr Compute Server, Gen4
	80084	Bionano Compute Server, Gen4
Cloud Computing	90047	Bionano Compute On Demand, US
	90052	Bionano Compute On Demand, Germany
	90060	Bionano Compute On Demand, Europe
	90128	Bionano Compute On Demand, Canada

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Redefine Cell Line QC with Optical Genome Mapping

Characterizing cell line genomic integrity and editing events is of critical importance to R&D and QC/CMC teams working in cell and gene therapy programs.

Traditional cytogenetic and NGS methods have significant limitations in variant class coverage, resolution, turnaround time and scalability. This necessitates using a constellation of these methods to characterize and monitor genomic integrity and gene-editing events (off- and on-target) which can be complicated, time-consuming, and expensive.

Bionano Saphyr® Optical Genome Mapping system



The Bionano Saphyr® Optical Genome Mapping system delivers game-changing high performance and comprehensive variant class coverage with an unbiased, genome-wide approach and digital readout — without revealing proprietary sequence data.

Move Your Program Forward with Optical Genome Mapping

	OGM	Karyotype	ddPCR	Microarray	NGS Panel
Genome-wide, Unbiased Detection	✓	✓	✗	✓	✓
High Resolution	✓	✗	✓	✓	✓
Variant Class Coverage	✓	✓	✗	✓	✓
Limit of Detection (VAF%)	✓	✓	✓	✗	✓
Time-to-Result	✓	✗	✓	✓	✓
Sample-to-Answer Workflow	✓	✗	✓	✓	✗
Automated Variant Calling	✓	✗	✓	✓	✓
Cost	✓	✓	✓	✓	✗

Move Forward with Us

Several early adoption sites, including both academic research foundations and commercial biotechnology organizations, have shared their experiences with OGM across a variety of applications

Organization	Application	Methods	Findings
CiRA Foundation (Japan)¹	Evaluating the effects of CRISPR-Cas9 gene editing	Performed a stringent genomic integrity assessment of CRISPR-Cas9 edited iPSC subclones, using WGS, karyotyping and OGM	OGM uniquely identified unexpected chromosomal translocations and inversions introduced by gene editing
Oklahoma Medical Research Foundation (USA)²	Evaluating the effects of prolonged cell culture on induced pluripotent stem cells (iPSCs)	Measured the effects of cell culturing in two iPSC lines in parallel for 50 passages and examined them at multiple time points using OGM	OGM identified substantial changes in the iPSC line genomes, including deletions, insertions, balanced translocations and inversions
Verve Therapeutics (USA)³	Evaluating genomic integrity after CRISPR-Cas genome engineering in a primary liver cell line used in drug development	Assessed for chromosomal rearrangements and large insertions or deletions in a liver cell line treated with a single course gene editing drug in development	They showed that no additional SVs accumulated after treatment when compared with untreated controls
bit.bio (UK)⁴	Cytogenetic quality control of iPSCs	Assessed the cytogenetic health of iPSC banks at commercial scale	They adopted OGM in-house as a single workflow solution, replacing an outsourced two-assay process, reducing TAT from 5 weeks to 1 week and improving the quality of SV data



See How OGM Can Support Your Cell QC Needs

The Bionano Saphyr® system is available through either a rental agreement or purchase. Installation, training, software, and support are included.

Tell us about your project at bionano.com/contact-us and our team will reach out to you.



“We were immediately impressed by the quality of data produced by Saphyr. **It also reduced costs per sample and turnaround time from 5 weeks to under 1 week. We've gained unprecedented clarity as to the genetic health of our cell lines.**”

Arran Constantine | Scientist MSAT, bit.bio

Commenting on adoption of OGM in-house for quality control of iPSCs

References: 1. Kitano et al. Mol Ther Methods Clin Dev. 2022;26:15-25. doi: <https://doi.org/10.1016/j.omtm.2022.05.010> 2. Dubose et al. Genes. 2022; 13(7):1157. doi: <https://www.mdpi.com/2073-4425/13/7/1157> 3. Verve Therapeutics press release. April 26, 2022. <https://ir.vervetx.com/news-releases/news-release-details/verve-therapeutics-presents-comprehensive-analysis-target-4> 4. Bit.bio website. May 5, 2022. <https://www.bit.bio/blog/how-our-culture-enables-new-technologies>



Hematological Malignancies and Solid Tumor Research

Genome variation common to cancer is too complex for low coverage whole genome sequencing. Complex rearrangements as well as highly repetitive regions of the genome present additional challenges for short- and long-read sequencing technologies.

Optical Genome Mapping (OGM) detects structural variations in an unbiased manner and at much higher sensitivities than sequencing-based technologies, and routinely at 5% variant allele fraction.

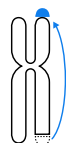
Extreme genomic rearrangements are hallmarks of cancer manifestation. Deciphering complex genomic structures requires a combination of short-read sequencing as well as long-range information with high coverage to unlock heterogeneity while providing a complete picture of the genome. To date, NGS applications in the clinic are limited to either low coverage across the whole genome, or high coverage exome sequencing that disregards 98% of the genome. The repetitive nature of the genome makes two-thirds largely inaccessible by short-read sequencing, and the short reads make elucidating the complex rearrangements seen in cancer impossible. The throughput of current long-read sequencing technologies is too limited to be realistically applied to cancer diagnostics or discovery. High cost and low throughput of long-read sequencing technologies, among other things, make cancer genomics intractable to those technologies.

OGM uncovers large structural variations beyond what short- and long-read sequencing can see.



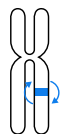
Insertions/deletions

500 bp for diploid genomes
5 kbp for mosaic sample



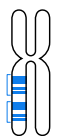
Balanced and unbalanced translocations

Larger than 50 kbp



Inversions

Larger than 30 kbp



Duplications

Larger than 30 kbp



Variant allele
fraction
detection



Coverage
per sample

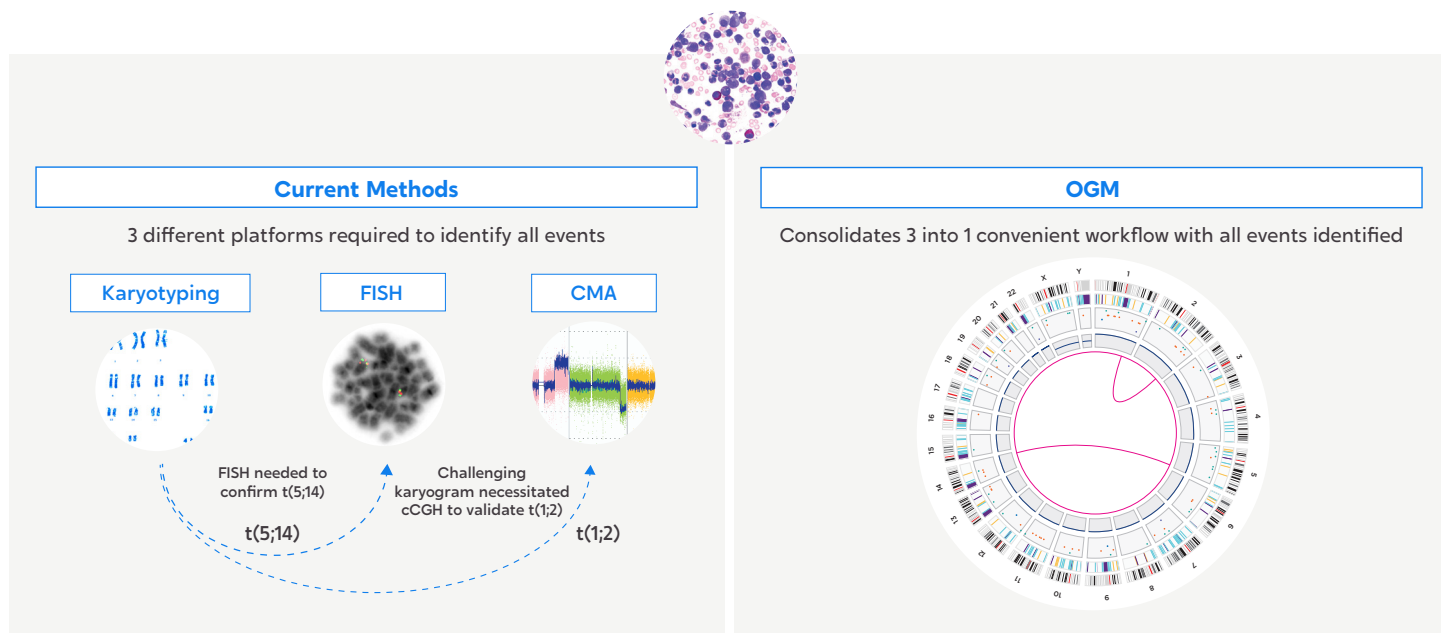
The workflow is simple, and it starts with ultra-high molecular weight DNA isolation from blood, cells, tissue or tumor biopsies. A single enzymatic reaction places 500,000 fluorescent labels all throughout the genome at a specific sequence motif occurring approximately 20 times per 100 kbp in the human genome. The long, labeled DNA molecules are linearized in nanochannel arrays on a Saphyr chip® and imaged in an automated manner by the Saphyr® Instrument. Changes in patterning or spacing of the labels are detected automatically, genome-wide, to call all structural variants.

OGM Consolidates Karyotyping, FISH, and CMA in a Single Workflow

Identification of complex rearrangements in cancer often requires a combination of approaches, which are typically performed sequentially. This sequential approach manages costs, but increases the turnaround time and complicates the understanding of the genetic makeup of the sample. A leukemia sample presented a challenging karyogram, necessitating chromosomal microarray

as a complement to validate t(1;2). Suspicion of a previously missed t(5;14) called for a FISH analysis. Results from karyotyping and FISH were inconclusive since the acceptor chromosome couldn't be clearly identified. The two events were captured by OGM in a single experiment and benign structural variants were automatically filtered out without complex pipelines!

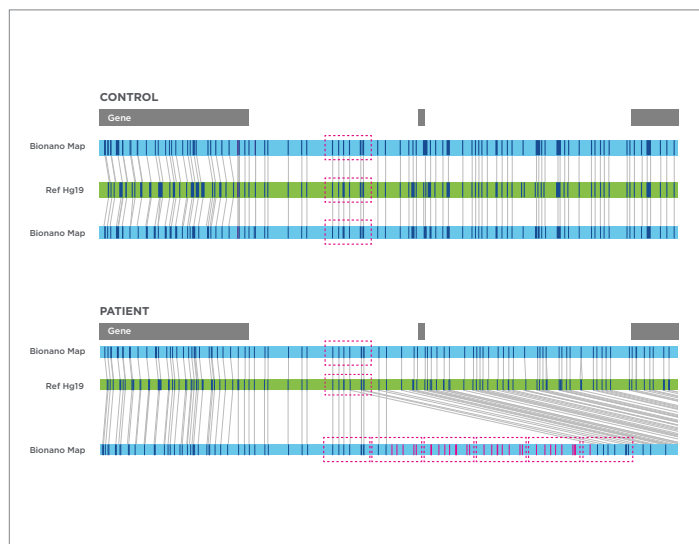
Challenging Leukemia Sample



OGM Identifies Variants in and Around Genes Implicated in Cancer That are Missed by Short- and Long-read Sequencing

Because they are too long:

A case of familial cancer known to be linked to a particular gene was studied for a decade without any molecular evidence of events perturbing that gene. OGM identified a 38 kb cassette that was amplified in six tandem copies just upstream of this gene. Saphyr imaged single molecules spanning all 230 kbp of this tandem repeat, which allowed it to be observed directly, as opposed to inferred algorithmically, as is the case with other molecular methods.²

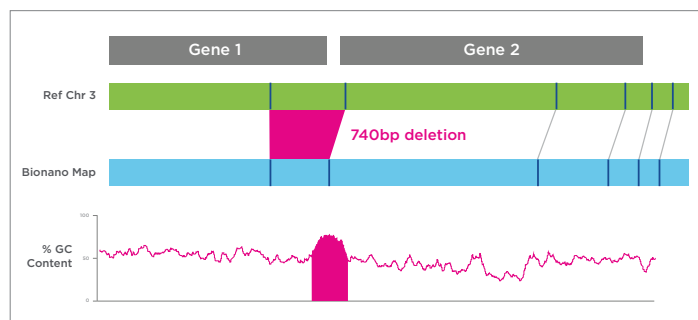


Because they are too rare:

The study of complex rearrangements in heterogenous cancer samples with long-read sequencing is done at the expense of coverage, throughput or cost. Reaching the low allelic fraction necessary to identify SV in biopsies requires deeper coverage than long-read sequencing can offer for a reasonable price. At 400X coverage depth in a human cancer sample, OGM can routinely detect variants at 5% allele fraction.

Because they are inaccessible by sequencing:

In a patient-derived model of uveal melanoma, no pathogenic variant or epigenetic event was found explaining the loss of expression of gene 1 using NGS, which is typically associated with this cancer. OGM identified a 740 bp deletion in the promoter of gene 1. This region was missed by sequencing due to the high GC content of that region.³

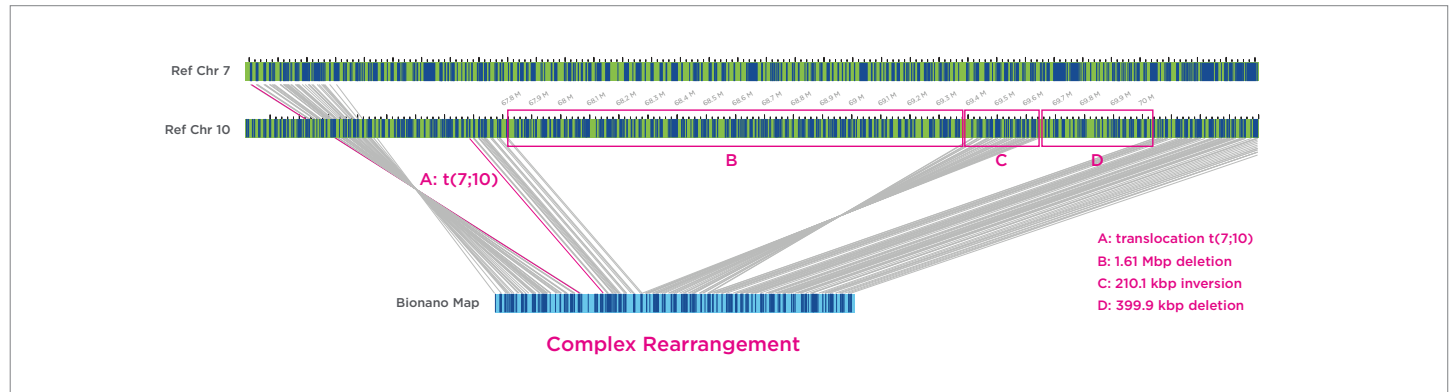


OGM Identifies Mutation Signatures and Brings Order Without Complex Pipelines

In highly rearranged cancer samples:

Cancer samples often display a high number of structural variants, and the limitations of the short-read length of NGS are particularly detrimental for correctly reconstructing these chained fusion events. In this patient-derived breast cancer cell line,

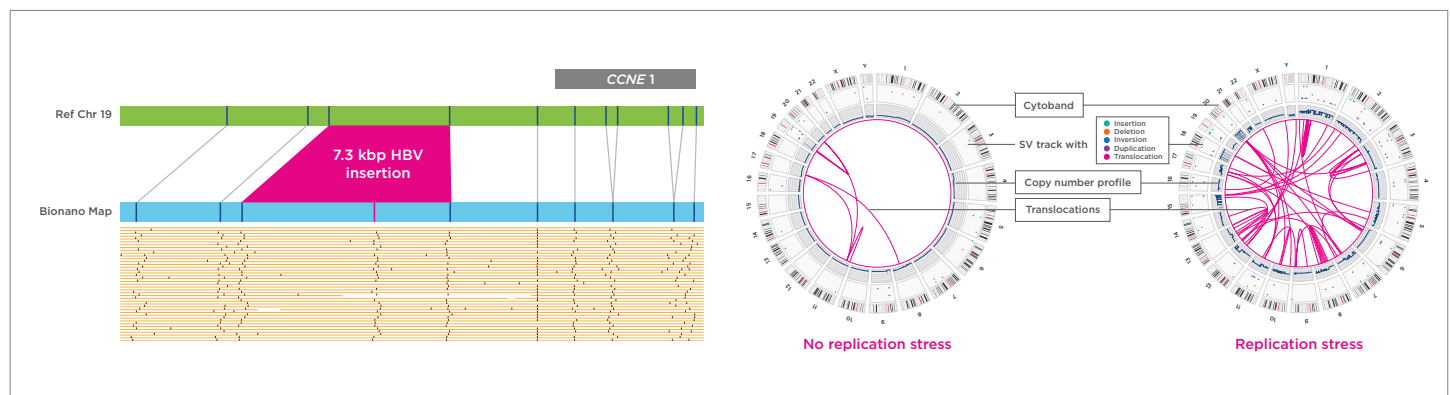
a consensus map resulting from the alignment of dozens of molecules spanning the region allowed OGM to identify a succession of a translocation, deletions and an inversion missed by short- and long-read sequencing.



In samples where stratification is needed:

The ability to stratify patient samples based on mutational profiles usually requires extensive bioinformatic data curation downstream of sequencing. In a hepatocellular carcinoma study, our built-in pipeline automatically provides enough information to distinguish samples with or without a replication stress signature. The circo plot

on the right in the image below shows replication stress resulting from a Hepatitis B virus insertion 9 kb upstream of the Cyclin E gene. Patients could be stratified based on the accumulation of DNA damage to determine if they are candidates for novel innovative therapies such as PARP inhibitors.⁴



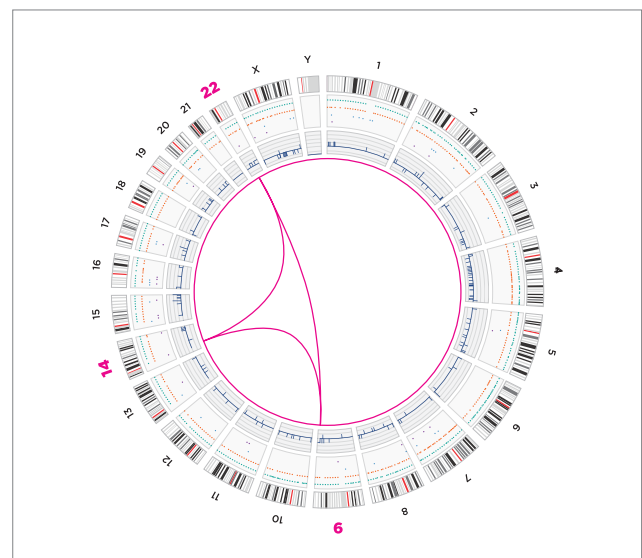
OGM is a Streamlined Tool For Personalized Medicine

Because it automatically identifies classical actionable fusions:

Rapid identification of *BCR-ABL1* translocations in Acute Lymphoblastic Leukemia is urgent for leveraging potential therapeutic options. Imatinib is a specific inhibitor of the fusion protein resulting from that translocation. OGM easily identifies complex and rare events such as this 3-way translocation of the Philadelphia chromosome t(9;22)(q34;q11).⁵

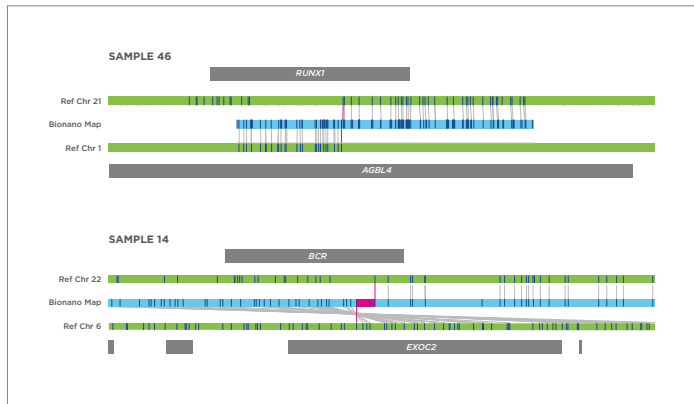
Because it identifies novel fusions:

In a study conducted on 48 leukemia samples, OGM identified 2178 rare structural variants, of which 95 were rare interchromosomal translocations, and 23 were unique calls containing potential gene fusions, leading to new avenues of research for drug development.⁵



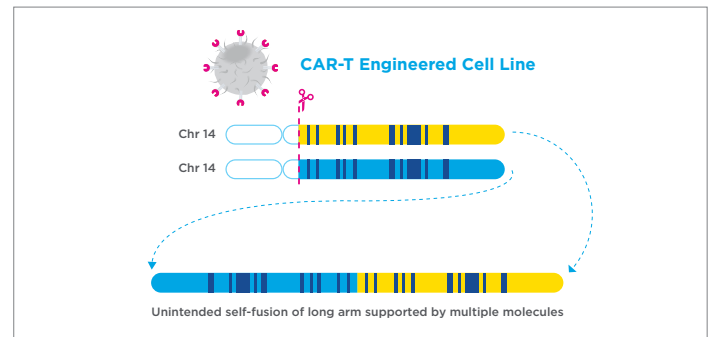
Because it easily identifies rare/new fusions of known genes:

In a large systematic comparative study between OGM and classical cytogenetics, OGM identified novel, non-recurrent fusions never reported before. In both cases shown here, one of the two fusion partners is well-known in leukemia as a gene fusion. These events were missed by classical cytogenetics, either because of their low allelic frequencies or because of the targeted FISH approach classically used in a diagnostics context.⁵



Because the ultrasensitive, automatic, genome-wide detection of rearrangements brings safety in CAR-T cells manufacturing:

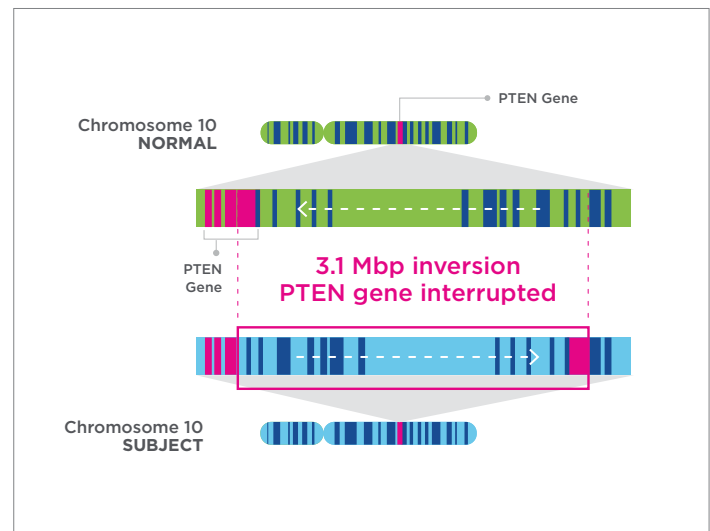
CAR-T cells are a powerful new therapeutic tool, but these engineered immune cells are often poorly characterized or remain unchecked for unwanted alterations. OGM can rapidly collect several thousand folds of coverage of a human genome, allowing for the detection of undesired rearrangements. Here, an unwanted inverted self-fusion of a large part of the long arm of chromosome 14 was detected in an engineered cell line, and several imaged molecules validated this fusion.



Combined with NGS, OGM Provides a Comprehensive Understanding of Cancer Events, From SNVs to Multi Mega Base Pair Structural Variations

To perform complete tumor profiling for cancer discovery:

In 12 leukemia genomes analyzed with OGM and whole-genome sequencing (WGS), dozens of genes were affected by multiple structural and single nucleotide variants. Most of these genes were not previously associated with leukemias or with cancer in general. The potential for novel biomarker discoveries, even in such a small sample size, is unexpectedly large. Many of the SVs were affecting regulatory sequences, absent from whole exome sequencing approaches. Intergenic mutations are a known mechanism for tumor escape from chemotherapy. TCGA data showed that expression differences in several of these novel biomarker genes are associated with outcomes and provide useful information for follow-up regarding response to chemotherapy. In the example shown here, a 3.1 Mbp inversion was detected by OGM breaking the *PTEN* gene, while a SNV was detected by WGS in the other *PTEN* allele. This compound heterozygous variant made this subject a candidate for trials with drugs attempting to rescue *PTEN* variants.⁶



To learn more about Bionano OGM applications for cancer research, visit bionano.com/cancer

References: 1. Hoischen A. Whole genome imaging to streamline cancer cytogenetics and identify novel rearrangements and biomarkers. ESHG Bionano workshop 2019. <https://bionanogenomics.com/videos/eshg-2019-workshop-series/> 2. Bocklandt S. Whole genome imaging to streamline cancer cytogenetics and identify novel rearrangements and biomarkers. AMP Bionano workshop 2019. <https://bionanogenomics.com/videos/amp-2019-series-dr-sven-bocklandt/> 3. Gentien D, Saberi-Ansari E, Servant N, et al. Multi-omics comparison of malignant and normal uveal melanocytes reveals novel molecular features of uveal melanoma. bioRxiv 2022.03.11.483767; doi: <https://doi.org/10.1101/2022.03.11.483767> 4. Péneau C, Imbeaud S, La Bella T, et al. Hepatitis B virus integrations promote local and distant oncogenic driver alterations in hepatocellular carcinoma. Gut. 2022;71(3):616–626. doi: [10.1136/gutjnl-2020-323153](https://doi.org/10.1136/gutjnl-2020-323153) 5. Neveling K, Mantere T, Vermeulen S, et al. Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. Am J Hum Genet. 2021;108(8):1423–1435. doi: [10.1016/j.ajhg.2021.06.001](https://doi.org/10.1016/j.ajhg.2021.06.001) 6. Xu J, Song F, Schleicher E, et al. An integrated framework for genome analysis reveals numerous previously unrecognized structural variants in leukemia patients' samples. bioRxiv 563270; doi: <https://doi.org/10.1101/563270> 7. Luebeck J, Coruh C, Dehkordi SR, et al. AmpliconReconstructor integrates NGS and optical mapping to resolve the complex structures of focal amplifications. Nat Commun. 2020;11(1):4374. Published 2020 Sep 1. doi: [10.1038/s41467-020-18099-z](https://doi.org/10.1038/s41467-020-18099-z)

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Revolutionizing Cytogenomics



Consolidate traditional cytogenetic assays into a single workflow with Optical Genome Mapping (OGM) and the Bionano Saphyr® System

In spite of the revolution sequencing technologies have brought about for genomics research and diagnostics, it has barely modified the way cytogenomic labs look at chromosomal aberrations.

While NGS identifies single- nucleotide variants along with small insertions and deletions (<150 bp), it fails to identify most large insertions, deletions, and copy-number variations in repetitive regions of the genome. NGS does not reliably detect balanced SVs such as inversions and translocations. Moreover, NGS relies on short-read sequences that are mapped to a reference human genome, which introduces bias while calling structural variants. Long-read sequencing is still limited in its resolution.

These limitations make direct visualization of the DNA the most reliable approach for the identification of structural variants to date.

Unsurprisingly, the way structural variants are detected in clinical samples has minimally evolved over the past decades, and mainly



10,000x

**greater resolution
compared to karyotyping**

relies on traditional cytogenetic methods such as karyotyping, Fluorescent in situ Hybridization (FISH) and array-based technologies. None of these methods alone can address complex cases due to technical limitations and need to be combined and complemented with molecular methods such as MLPA, qPCR, RNA-seq to provide a complete therapeutic and prognostic assessment of the patient or tumor genome.

OGM transforms traditional cytogenetics techniques by delivering an improved visualization of the genome by going from microscope to a fully digital, high-resolution chromosomal analysis.

Only here, we apply 500,000 such bands to a genome, and the imaging is performed in an extremely high-throughput, automated manner in nanochannel arrays that linearize megabase-size molecules. This allows us to see events at 10,000x greater resolution than by karyotyping: insertions and deletions at 500 bp, vs ~5 Mbp by chromosome banding analysis.

Cytogeneticists

Visualize patterns on intact DNA molecules to detect structural variation



Saphyr®

Automates the imaging of 1000x more label patterns on intact DNA molecules to detect structural variation in massively parallel nanochannel arrays



OGM Provides Highly Sensitive Detection of All Classes of Structural Variants

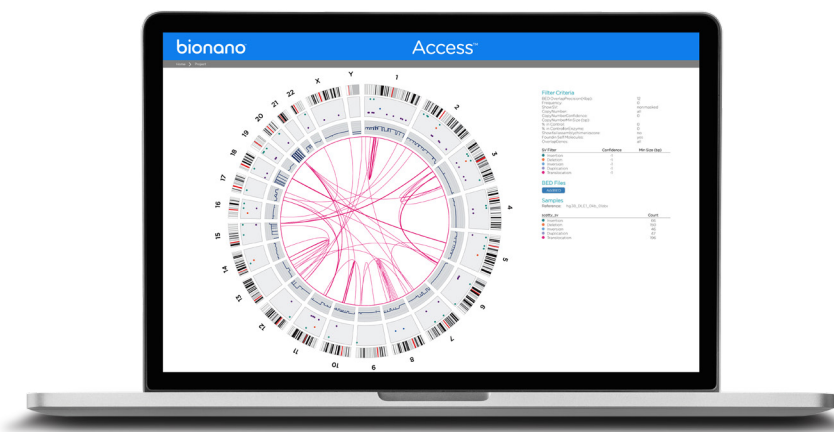
Validation studies around the world are confirming OGM's performance. OGM finds the clinically relevant variants detected by karyotyping, FISH and microarray combined, and reveals many that are missed.

OGM demonstrated 100% concordance with karyotyping, FISH, and chromosomal microarray in constitutional disorders in two studies appearing in the July 2021 issue of the American Journal of Human Genetics (AJHG).

The authors in these back-to-back AJHG publications describe OGM as a better alternative to traditional cytogenetics assays for both

inherited genetic disease and hematologic malignancy applications since it consolidates multiple antiquated methods requiring manual integration for interpretation into a single workflow with higher resolution for detection of all classes of structural variants.¹

Publication of a study of 76 subjects by authors at The University of Texas MD Anderson Cancer Center evaluated the utility of OGM as an alternative to traditional cytogenomic methods for the characterization of structural variation in myelodysplastic syndrome (MDS). The study was published in Blood, the journal of the American Society of Hematology (ASH), and presented at the 2021 ASH annual conference.²



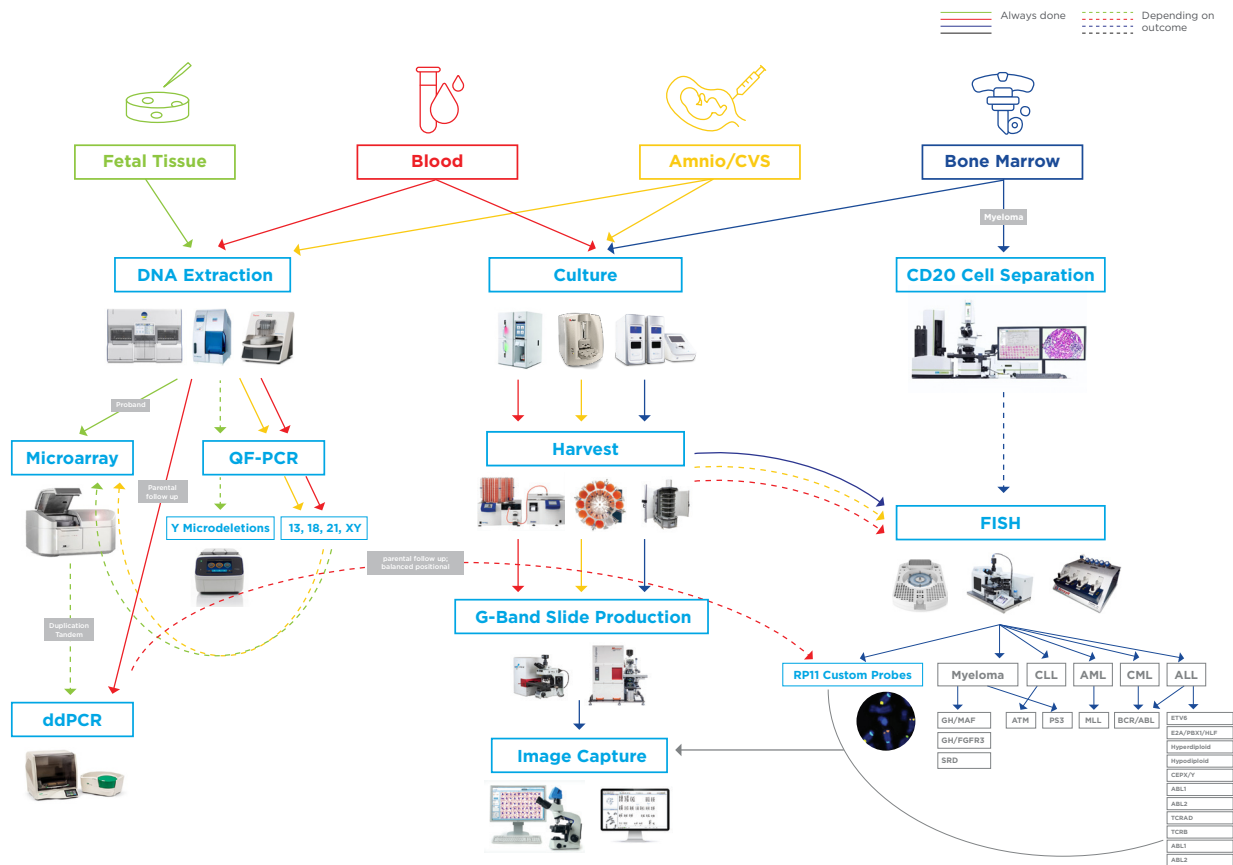
Method	Resolution	
Karyotyping	~5 – 10 Mbp	Extensive training required for interpretation. Slow, labor-intensive data collection and analysis. Cell culture required. ⁴
FISH	~100 kbp	Targeted, extremely limited approach; only shows handful of variants. Slow, labor-intensive data collection. Requires validation of every lot of probes utilized. ⁴
Array-based Techniques	~50 kbp	Cannot detect balanced rearrangements. Cannot resolve nature of a structural aberration. ⁴
OGM	As low as 500 bp	Fully automated detection of: CNVs, repeat expansions, FSHD1-related repeat contractions, unbalanced events from single exon level to aneuploidies, balanced events, inversions, translocations, gene fusions, and AOH.

OGM Consolidates the Traditional Cytogenetic Assays into a Single Workflow

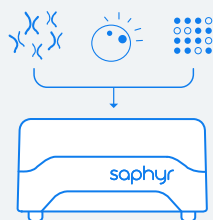
OGM is the **ONLY** technology that allows for the highly sensitive detection of all structural variant types, even those present at low allele fraction in heterogenous cancer samples, in an unbiased genome-wide manner.

By providing a complete and unambiguous picture of the genome structure, OGM identifies prognostic markers not currently monitored, and enables a complete characterization of the cancer or subject's genome, **replacing multiple cytogenetic tests that make up the gold standard.**

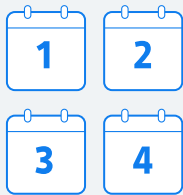
Current Cyto Lab Workflow



Experience the Advantages of OGM and Saphyr



One streamlined workflow



Sample to result in as few as 4 days⁵



Variant allele fraction detection



Coverage per sample

3 Ways to Get Bionano Data

Get The Service



Bionano Data Services

Submit your samples to Bionano Data Services and receive an appropriately filtered set of structural variant calls. SV data is presented using the Bionano Access® visualization software. Files can be exported in the format of your choice.

The Bionano Support team will work with you on experiment design and analysis training. Full analysis is available as an option.

Sample Types Accepted – Frozen, Mammalian Preferred

- Tissue Biopsies
- Cultured Cells
- Blood
- Bone Marrow Aspirates

Pricing available upon request.

- Diploid/Genetic disease samples collected at 120x
- Mosaic/Cancer samples collected at 400x
- Low-frequency variant samples collected up to 1600x

Get The Consumables



Reagent Rental Agreement

Run samples in-house with a Saphyr® Instrument free of charge for the duration of your project. The Bionano Support team will install the Saphyr System and provide training on sample preparation, instrument operation, and data analysis.

Please contact Bionano to request a reagent rental quote.

- Pricing is based on total genome commitment during a 6-month period
- Per-genome pricing includes DNA isolation, labeling, chips, and Bionano Compute On Demand
- Installation and training included

Get The Saphyr® System



System AND Consumables Purchase

Purchase the Saphyr System for your institution without any reagent commitment. The Bionano Support team will install the Saphyr System and provide training on sample preparation, instrument operation, and data analysis.

Saphyr System Components

- Saphyr Instrument
- Saphyr Chips
- Bionano Prep Kits
- Bionano Access Software
- Bionano Compute On Demand (optional)
- Bionano Access Server

Please contact Bionano for a detailed quote. Pricing may vary, depending on region, availability, and secondary hardware needs.

- Saphyr System includes installation and training
- Volume-based, per-genome consumable pricing
- Consumables include DNA isolation, labeling, chips, and Bionano Compute On Demand

To see all genetic disease case studies, presentations, and additional materials, visit bionano.com/cytogenomics

References: 1. Levy B, Baughn LB, Chartrand S, et al. A national multicenter evaluation of the clinical utility of optical genome mapping for assessment of genomic aberrations in acute myeloid leukemia. medRxiv 2020.11.07.20227728. doi.org/10.1101/2020.11.07.20227728 2. Hui Yang, Guillermo Garcia-Manero, Guillermo Montalban-Bravo, et al. High-Throughput Characterization of Cytogenomic Heterogeneity of MDS Using High-Resolution Optical Genome Mapping. Blood 2021; 138 (Supplement 1): 105. doi: <https://doi.org/10.1182/blood-2021-154005> 3. Neveling K, Mantere T, Vermeulen S, et al. Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. Am J Hum Genet. 2021;108(8):1423-1435. doi:10.1016/j.ajhg.2021.06.001 4. Silva M, de Leeuw N, Mann K, et al. European guidelines for constitutional cytogenomic analysis. Eur J Hum Genet. 2019;27(1):1-16. doi:10.1038/s41431-018-0244-x 5. For human samples collected at 100x and analyzed through the de novo assembly pipeline.

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bionano™



Genome-wide Detection of All Structural Variants

Advances in sequencing technologies have barely changed the way structural variants are detected. NGS relies on short-read sequences that are mapped to a reference human genome and fails to identify most large insertions, deletions, and copy-number variations in the two-thirds of the genome that is repetitive. In addition, NGS does not reliably detect balanced SVs such as inversions and translocations.

Optical Genome Mapping (OGM) directly visualizes patterns of labels on intact DNA molecules to detect structural variation.

OGM accurately detects all classes of structural variants, which include balanced translocations, inversions, complex rearrangements, deletions, gains, insertions, and repeat expansions and contractions. All classes of structural variants are detected at high sensitivities.

Unlike sequencing-based methods, which are typically unable to detect insertions or identify where the extra sequence is inserted, OGM detects both deletions and insertions starting at 500 bp with high sensitivity. It detects mosaic variants down to as little as 5% variant allele fraction because it uses single-molecule analysis.

Ultra-high molecular weight DNA is isolated from blood, cells, tissue or tumor biopsies, and a single enzymatic reaction places 500,000 fluorescent labels all throughout the genome at a specific sequence motif. The labeled DNA molecules are linearized in nanochannel arrays on the Saphyr chip and imaged in an automated manner. Changes in the patterning or spacing of the labels are detected automatically, genome-wide, to call all structural variants.

OGM by the Numbers



SV detection sensitivity



Lower limit of resolution for detection of deletions and insertions



Turnaround time

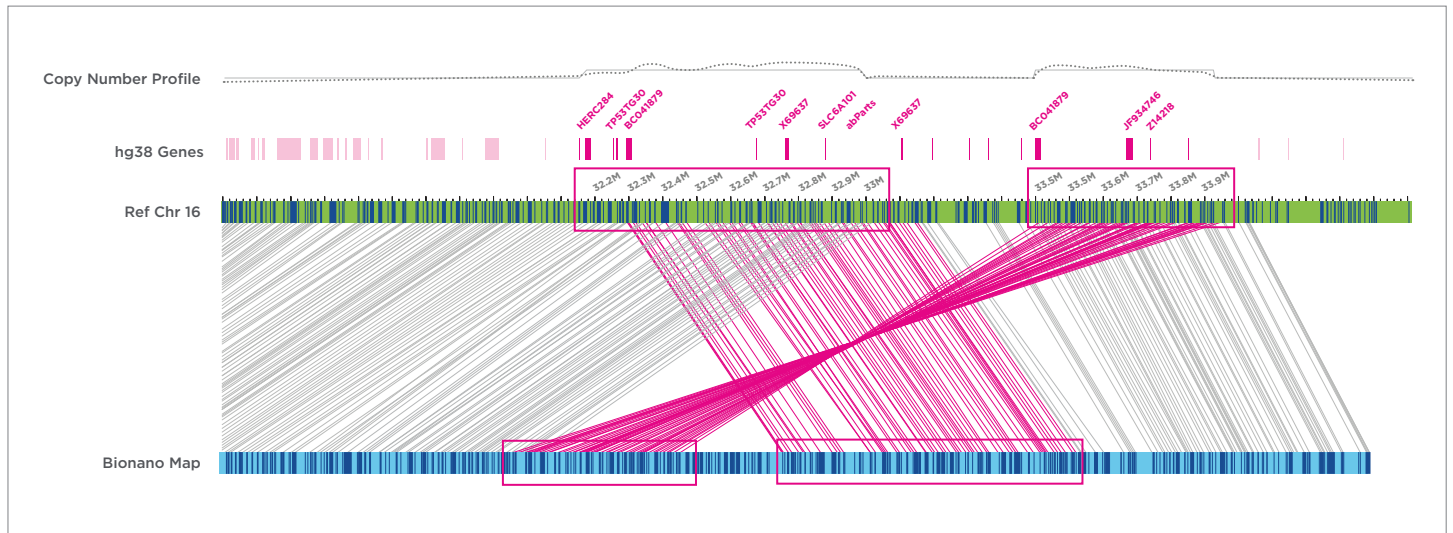


Resolution compared to karyotype

OGM Finds New Candidate Genes

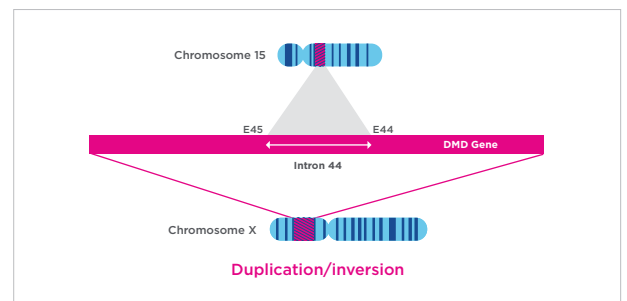
In a newborn with Congenital Diaphragmatic Hernia (CDH), a severe developmental disorder affecting the diaphragm, lungs and sometimes heart, OGM detected two adjacent duplications,

one direct and one inverted. OGM revealed a much more complex architecture than could be inferred from microarray data, and identified several additional candidate genes for CDH.¹



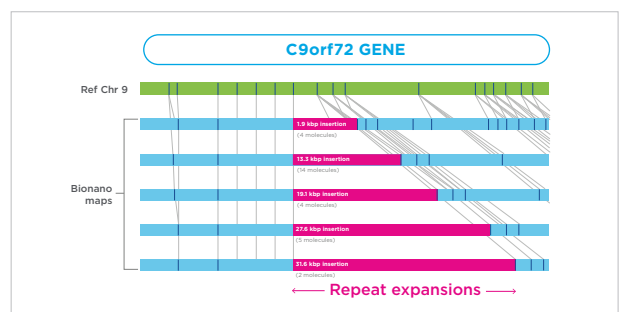
OGM Identifies New Variants in Known Genes

In a subject with Duchenne Muscular Dystrophy (DMD), a 420 kbp segment from chromosome 15 was duplicated in an inverted orientation in intron 44 of the Dystrophin gene. This insertion was not detected by NGS, and while chromosomal microarray can detect the duplication, its location and, therefore, implication in DMD could not be determined.²



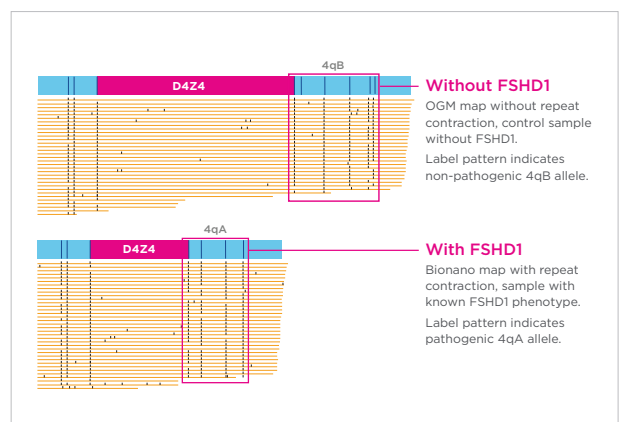
OGM Reveals Repeat Expansions

In a single postmortem brain sample from an ALS subject, OGM detected a highly mosaic range of expansions of the *C9orf72* GGGGCC repeat, ranging from the reference allele (not shown) to a 32 kbp expansion. No modern technology has been capable of spanning and measuring these large *C9orf72* repeat expansions.³



OGM Detects FSHD1

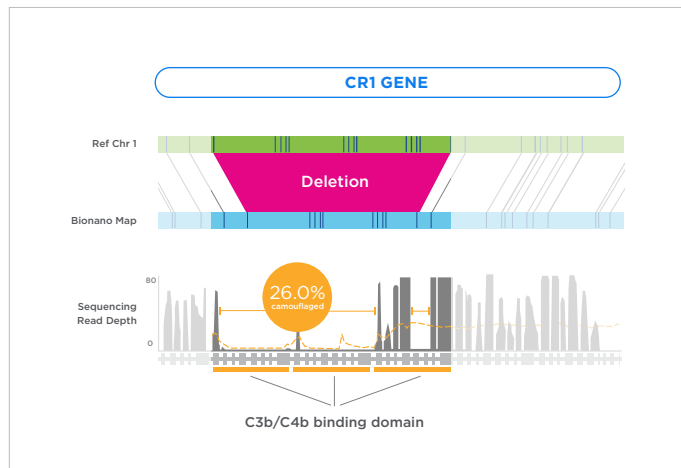
Facioscapulohumeral Muscular Dystrophy (FSHD1) is a common form of muscular dystrophy with an extremely complex genotype. Correct genotyping requires the accurate sizing of a very large repeat region in the subtelomeric region of chromosome 4, a correct determining of the pathogenic vs non-pathogenic allele, and the distinction between the chromosome 4 repeat and an almost identical repeat on chromosome 10 not related to the disease. A team from the University of Iowa published the largest clinical research study to date evaluating OGM for FSHD1. The study, published in the Journal of Molecular Diagnostics, concluded that OGM can be performed more quickly, accurately, and reproducibly than the current gold standard method of Southern blot analysis.⁴



OGM Finds Variants Other Technologies Can't See

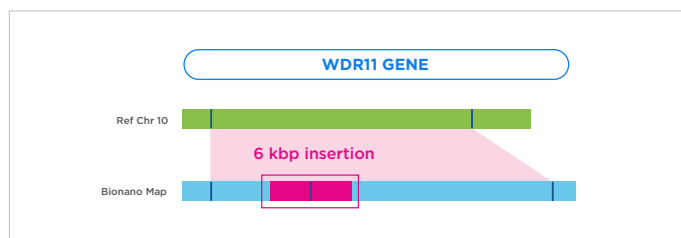
Because they're camouflaged:

Many protein-coding exons are 'camouflaged' in NGS datasets because of variably repeated binding domains—the exons occur in more than one gene or in tandem within the same gene, making correct alignment of short reads impossible. OGM allowed for the direct measurement of the number of C3b/C4b binding domains for each haplotype in *CR1*, an Alzheimer-associated gene, in this subject with Alzheimer's Disease.³



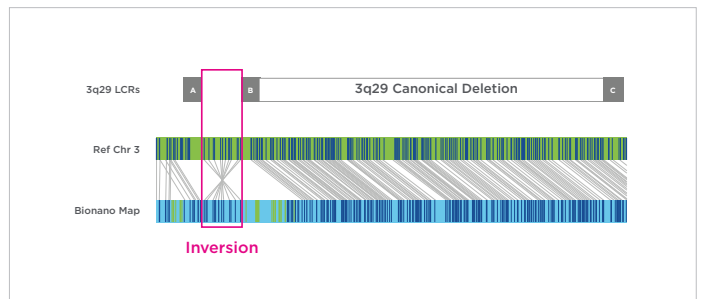
Because they're insertions:

While deletions are somewhat easier to detect by NGS, insertions are rarely picked up from NGS data. In a genetic male subject with gonadal dysgenesis, OGM identified a 6 kbp insertion in the *WDR11* gene, associated with abnormal testes development and cryptorchidism.²



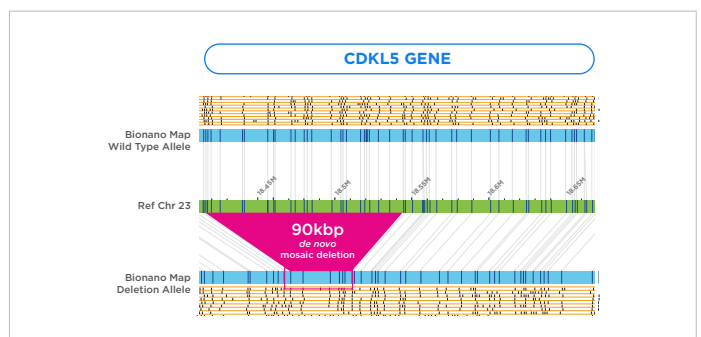
Because they're flanked by segmental duplications or are in other complex regions of the genome:

3q29 Microdeletion syndrome is present when a 1.5 Mbp region between two segmental duplications (also called Low Copy Repeats (LCR)) is deleted. It is thought that inversions in the parents between LCRs in this region predispose to the deletion in the child. Here, OGM detected a 350 kbp inversion between LCR A and B, something that's not been possible with any other genome analysis technology.⁵



Because they're mosaic:

A juvenile subject with epilepsy, hypotonia and developmental delay, extensively studied as part of the Undiagnosed Disease Network, remained undiagnosed. OGM found a 90 kbp mosaic deletion in *CDKL5*, an X-linked gene essential for normal brain development and function, and a phenotype that perfectly matches the subject's. The wild type allele is shown on top, and the deletion allele below.²

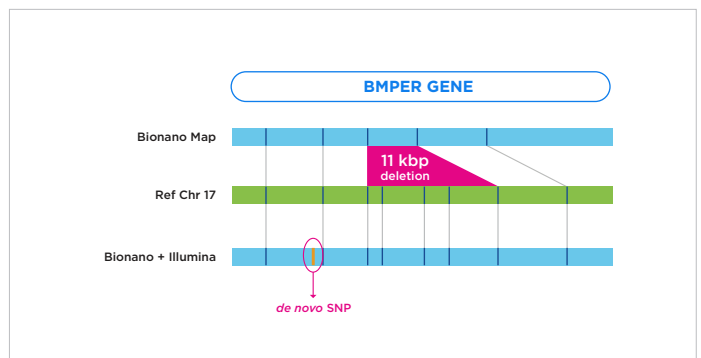


OGM Can be Combined with NGS

To find compound heterozygous variants:

In a subject with a complex phenotype and a variety of growth and developmental defects, OGM detected an 11 kbp deletion in the *BMPER* gene inherited from the mother, while a *de novo* SNP in the same gene was detected on the other allele. This combination creates a compound heterozygous mutation, only detected by a combination of NGS and OGM.²

BMPER is an autosomal recessive gene that regulates the Bone Morphogenetic Protein (BMP) and the phenotype matched a potential *BMPER* disruption.



3 Ways to Get Bionano Data

Get the Service



Bionano Data Services

Submit your samples to Bionano Data Services and receive an appropriately filtered set of structural variant calls. SV data is presented using the Bionano Access® visualization software. Files can be exported in the format of your choice.

The Bionano Support team will work with you on experiment design and analysis training. Full analysis is available as an option.

Sample Types Accepted – Frozen, Mammalian Preferred

- Tissue Biopsies
- Cultured Cells
- Blood
- Bone Marrow Aspirates

Pricing available upon request.

- Diploid/Genetic disease samples collected at 120x
- Mosaic/Cancer samples collected at 400x
- Low-frequency variant samples collected up to 1600x

Get the Consumables



Reagent Rental Agreement

Run samples in-house with a Saphyr® Instrument free of charge for the duration of your project. The Bionano Support team will install the Saphyr System and provide training on sample preparation, instrument operation, and data analysis.

Please contact Bionano to request a reagent rental quote.

- Pricing is based on total genome commitment during a 6-month period
- Per-genome pricing includes DNA isolation, labeling, chips, and Bionano Compute On Demand
- Installation and training included

Get the Saphyr® System



System AND Consumables Purchase

Purchase the Saphyr System for your institution without any reagent commitment. The Bionano Support team will install the Saphyr System and provide training on sample preparation, instrument operation, and data analysis.

Saphyr System Components

- Saphyr Instrument
- Bionano Access Software
- Saphyr Chips
- Bionano Compute On Demand (optional)
- Bionano Prep Kits
- Bionano Access Server

Please contact Bionano for a detailed quote. Pricing may vary, depending on region, availability, and secondary hardware needs.

- Saphyr System includes installation and training
- Volume-based, per-genome consumable pricing
- Consumables include DNA isolation, labeling, chips, and Bionano Compute On Demand

To see all genetic disease case studies, presentations, and additional materials, visit bionano.com/geneticdiseases

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Contact your Bionano Regional Business Manager to get started.

sales@bionano.com | 858.888.7600 | bionano.com

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