

Accessibility meets possibility™

Resolve whole-genome structural variation with one compact platform

Nabsys

Beyond small variants

Structural variants (SVs), defined as variants greater than 50 bp in size, are the largest source of genetic variation affecting 3-5x more DNA per genome than single-nucleotide variants (SNVs).[1] As a major contributor to genomic function and disease, detection of these chromosomal abnormalities is critical.

Unfortunately, traditional methods have left many SVs undetected, even after decades of innovation. Until now. The introduction of genome mapping finally offers a comprehensive view beyond single nucleotide variants and short indels, broadening your understanding of chromosomal abnormalities and expanding your research in new directions.

	Cytogenetics			Molecular Genetics	
	Karyotype	Microarray	FISH	Short-read sequencing	Long-read sequencing
Pros	Large chromosomal abnormalities, Aneuploidies, translocations	Copy number variations (CNVs) across the genome with higher resolution than karyotyping	Detecting and confirming specific known genetic abnormalities, such as gene rearrangements or amplifications	Excels at small variants <500 bp	Excellent detection of small variants and large SVs
Cons	Low resolution and cannot detect small structural variants or subtle genomic changes	Cannot detect balanced structural variants, such as translocations or inversions	Only detects abnormalities in regions specifically probed, missing genome-wide or unexpected changes	• 150 bp reads make SV detection difficult • Repetitive regions are difficult	• Very large SVs, translocation and copy neutral are challenging • Expensive for low VAF apps (e.g. subclonal events)

Discover the new standard in SV analysis

Electronic genome mapping

Nabsys, the pioneer of electronic genome mapping (EGM), uses solid-state nanodetectors to survey long DNA molecules—the reads are used to construct high-density maps with long-range information to detect SVs. Optical DNA mapping technologies that rely on the use of expensive optical imaging are limited in the resolution of neighboring tags by the diffraction limit of light. EGM uses electronic detection to identify tags and does not have the same limitations as optical methods, providing superior resolution of small intervals. This high-density information makes it possible to identify both balanced and unbalanced SVs, in addition to larger chromosomal aberrations and genetic variation missed by next generation sequencing (NGS).

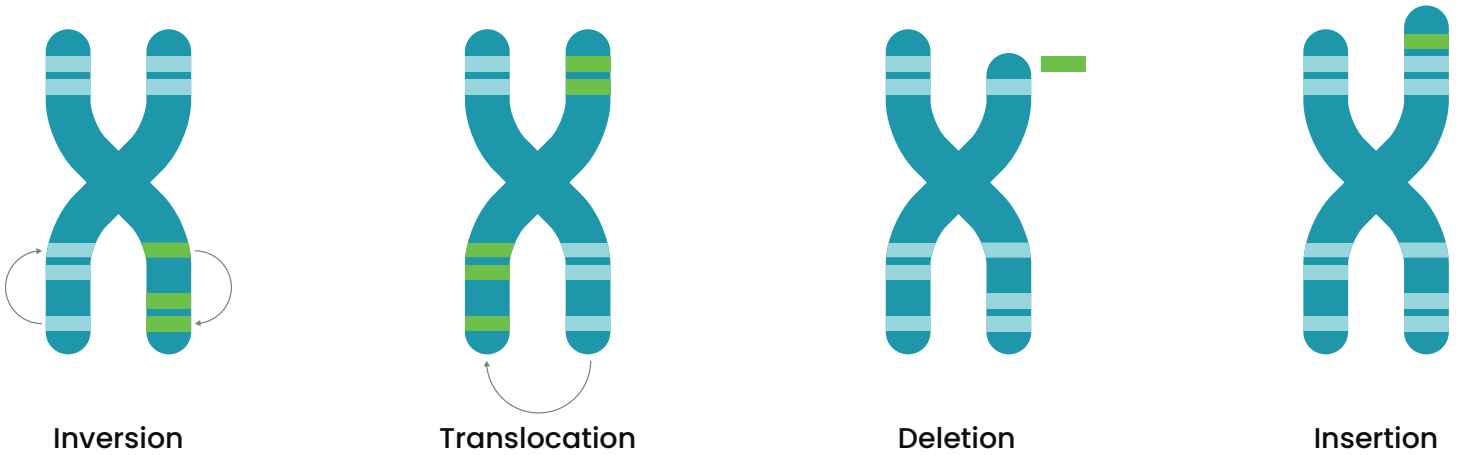

Figure 1: Types of structural variants (SVs). SVs come in several different forms, including inversions, translocations, deletions, insertions, and other complex chromosomal rearrangements.

[1] Chaisson, M.J., Sanders, A.D., Zhao, X. et al. Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nat Commun 10, 1784 (2019).

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Accessibility meets possibility

Resolve a broader range of variants with one compact platform

Electronic genome mapping (EGM) powers the OhmX Platform with a fundamentally simpler approach to structural variant detection. By eliminating expensive lasers, optics, and cameras, OhmX delivers high-resolution genome analysis in a compact, scalable, and cost-effective platform—expanding access to single-molecule insights to all laboratories regardless of budget.

High Resolution

>60% greater labeling density than competing platforms enable genome-wide SV detection down to 300 bp in size

Cost-Effective

3-4x less expensive vs competing tech with no sample batching required to achieve low pricing

Accessible

Small footprint with entire platform fitting within 5"x 17" x 17" saving valuable benchtop space

Single-Molecule Insights

Capture true molecule-to-molecule variation by analyzing individual DNA molecules directly, preserving genomic complexity and revealing heterogeneous and rare structural variants that can be obscured by consensus-based approaches

Integrated Software

Cloud-optimized primary and secondary analysis with robust support

Nabsys

From electronic signals to structural variant detection

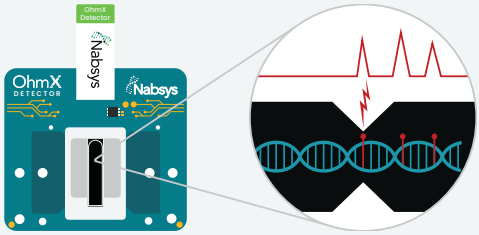
The OhmX Platform combines high-sensitivity electronics, nanofluidics, and computational biology. Central to our EGM technology are novel, solid-state nanodetectors that house 256 parallel nanochannels, each with its own pair of electronic sensors.



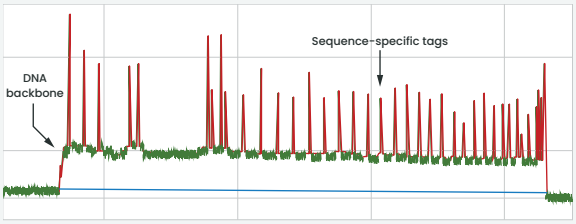
1 High molecular weight (HMW) DNA extracted from sample.



2 Sequence-specific nicking and labeling of HMW DNA at known recognition sites. Labels are added ~4 kb apart genome wide. DNA is coated with binding protein to enhance electronic signal detection.



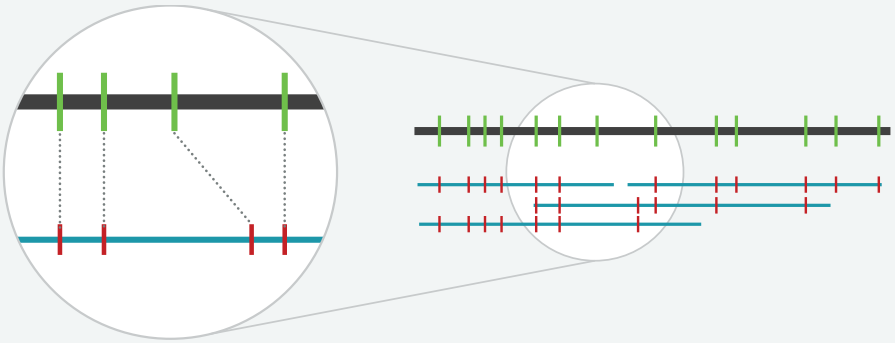
3 DNA travels through nanochannels and passes through an electronic detector. As tags pass through, they create a signal.



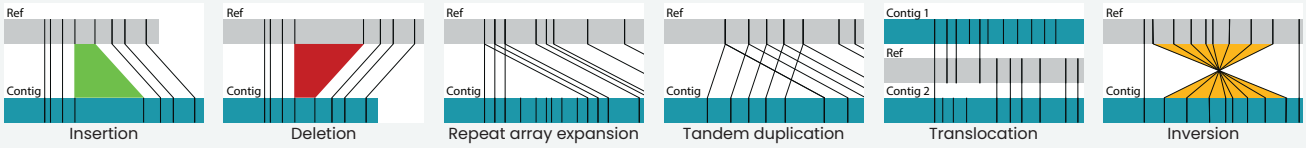
4 Trace files are generated as tagged DNA passes through the detector. The trace file above represents a single molecule of DNA >100 kb in length.



5 Using time to distance information, Trace files are converted to sequence specific molecules or maps.



6 Read maps are aligned to a reference genome by tag pattern. Changes in tag pattern relative to the reference indicate a structural variant.



7 EGM can detect SVs and copy number variations (CNVs).

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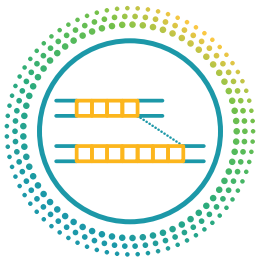
Supercharge your research with EGM

OhmX enables a variety of different SV analyses—from cytogenomics to molecular to long-read validation—based on your research needs ensuring a more complete view of the human genome.



Hematological Malignancy

Unravel the complexities of SVs in cancer genomes and gain deeper insights to drive breakthroughs in oncology research.



Repeat Expansion Disorders

Detect and confirm repeat expansions, delivering advantages in resolution, size detection, and reproducibility across diverse loci and cohorts.



Orthogonal Confirmation

Resolve uncertain SV calls from short- and long-read sequencing, providing clear and confident results.



Cell & Gene Therapy

Gain critical insights and clarity into genomic integrity that other tools can't provide.

Real-time results, powerful insights

Advanced cloud-based SV analysis pipelines

Nabsys Navigator™ powers whole-genome variant discovery and variant verification through our suite of advanced informatics pipelines. With minimal bioinformatics expertise required and no infrastructure to maintain, you'll get the analysis you need to map the genome—without the overhead.

Nabsys Navigator

A secure, cloud-based platform hosting the EGM analytics pipelines with comprehensive run and sample QC tools - your starting point for analysis.

SV-Discover™

Comprehensive genome-wide *de novo* assembly and SV-calling

RepX™

Specialized pipeline delivering estimated repeat sizing for up to 23 repeat expansion loci and molecule-level waterfall plots for visualization of repeat instability

SV-Verify™

Highly accurate alignment-based variant calling based on competitive mapping to putative SVs or reference constructs to confirm presence or absence of SVs

OhmX Platform features

Easy sample injection

Sample loading port allows for easy injections and walkaway sample introduction into the detector to support longer experimental runs

Fluidics automation

Detector carriage precisely aligns the detector with the microfluidic paths, allowing for completely automated experimental runs and data acquisition.

Simple waste collection

Waste is automatically collected and software alerts and instructs when to remove



Flexible run times

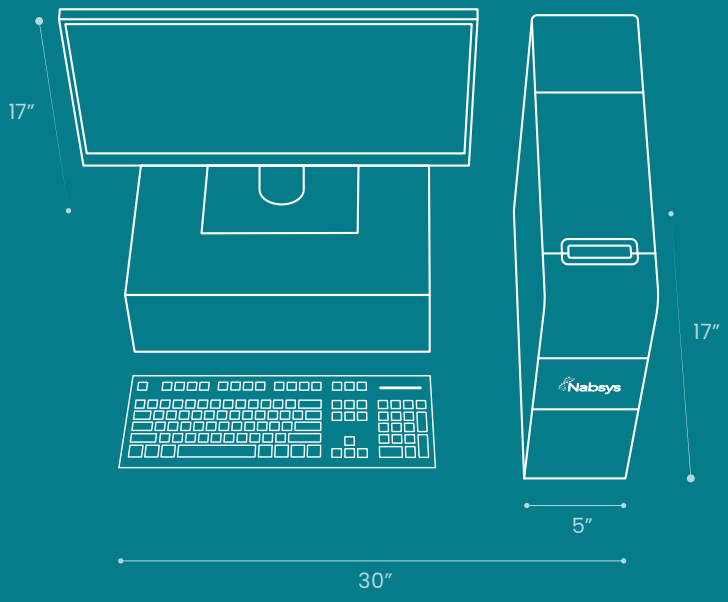
Cartridges contain preformatted, ready-to-use reagents for cleaning, washing, and running experiments on the solid-state nanodetectors, with volumes automatically managed by software

High accuracy and sensitivity

Precisely tuned electronics eliminate sources of noise, increasing detection throughput of tagged HMW DNA molecules

Make big discoveries in a small footprint

Perform cost-effective, high-quality SV analysis while saving lab space



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OhmX Platform Performance Metrics

System

Metric	SV Discovery (Germline)	SV Confirmation
Analysis Pipeline	SV-Discover	SV-Verify
Method	*de novo* assembly	targeted alignment
Mode of Detection	electronic	electronic
Coverage	>200x	50–150x
# Genomes/Year	96	96
# Detectors	2	1
SV Resolution Limit	>300 bp	>300 bp
Average Tags per 100 kb	Up to 25	Up to 25

Sample

Metric	SV Discovery (Germline)	SV Confirmation
# Cells, Blood Input Material	1,000,000 cells; 200–500 µL	1,000,000 cells; 200–500 µL
DNA Output from 1 Extraction	~ 5 µg	~ 5 µg
# Reactions per Genome Prep Kit	10 reactions	10 reactions
# Loads per Genome Prep Reaction	≥ 4 loads	≥ 4 loads
DNA Loaded per Sample	270 ng	270 ng

Data

Metric	SV Discovery (Germline)	SV Confirmation
Data collected (gigabytes) at 150x coverage	360 GB	application-dependent
Variant Allele Frequency	25%	variable
Minimum Tags Per Molecule	6	6
20th Percentile Remapped Length	> 140 kb	> 140 kb
Average Contig Length (N50)	600–1,000 kb	N/A
Reference Coverage of Assembly	> 90%	N/A
Data Export Format	CSV, JSON	CSV, JSON
Resolution	> 300 bp	> 300 bp

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