

Accessibility meets possibility[™]

Resolve whole-genome structural variation with one compact platform



Beyond small variants

Structural variants (SVs), those > 50 bp, are the largest source of genetic variation affecting 3-5x more DNA per genome than single-nucleotide variants (SNVs). 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 polymorphisms (SNPs) and short indels, broadening your understanding of chromosomal abnormalities and expanding your researching in new directions.

	Cytogenetics			Molecular Genetics	
	Karyotype	Microarray	FISH	NGS	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 <49 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 makes SV detection difficult Repetitive regions are difficult 	 Very large SVs, translocation and copy neutral are challenging Expensive

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).

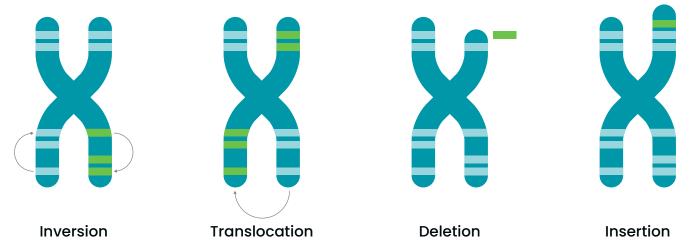


Figure 1: Types of structural variants (SVs). SVs come in several different forms, including inversions, translocations, deletions, insertions, and other, more complex chromosomal rearrangements.



Accessibility meets possibility

Resolve a broader range of variants with one compact platform

Electrify your insights with an end-to-end solution that takes you from sample prep to analysis with precision. With only a 16-hour run time, and an output of up to 130 genomes per year, the OhmX Platform detects a broad range of SVs that will change the way you research.



High Resolution

Genome wide detection of SVs from 300 bp to large and complex Mbp-sized chromosomal aberrations



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



Orthogonal Validation

Reliably confirm other technologies like NGS, long-read sequencing, chromosomal microarray, FISH, and Karyotyping



Integrated Software

Cloud-optimized primary and secondary analysis with robust support

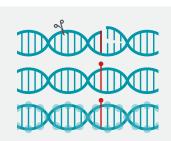
From electronic signals to structural variant detection

The OhmX Platform is based on an interdisciplinary approach combining 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.

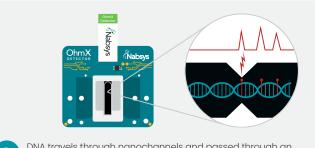


DNA extracted

from sample.



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



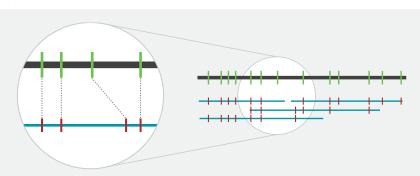
DNA travels through nanochannels and passed through an electronic detector. As tags pass through, they create a signal.



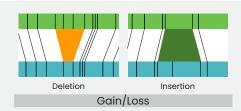
Trace files are generated as tagged DNA passes through the detector. The trace file above represents a single molecule of DNA >100Kb in length.

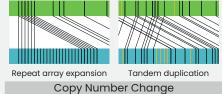


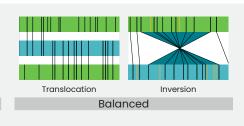
Using time to distance information, Trace files are converted to sequence specific molecules or maps.



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







7 All forms of SVs can be detected by EGM.

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.



Genetic Disease Research

Unravel complexities and gain critical insights with the OhmX Platform, designed to help you push the boundaries of genetic disease research



Cancer Research

Gain deeper insights and drive breakthroughs in cancer research with the OhmX Platform, powered by EGM and expertly engineered to unravel the complexities of SVs in cancer genomes



Cell and Gene Therapy

CGT treatments require a thourough understanding of structural variation, as well as characterisation of genomic impact

Real-time results, powerful insights

Cutting-edge cloud-based pipelines for SV analysis

After data acquisition on the instrument, the signal-processed data can either be remapped and aligned to the human reference genome (e.g., hg38) and then assembled into consensus contigs in HCE for genome-wide SV discovery, or passed directly to SV-Verify, which performs its own competitive remapping against hg38 and candidate SV constructs for targeted variant confirmation.

Human Chromosome Explorer™

Comprehensive, de novo assemblybased, genome-wide SV calling powered by Hitachi High-Tech®

Specialty Pipelines

Expanding suite of new tools for specialty analysis including repeat expansions, copy number variations, digital FISH and more

SV Verify™

Highly accurate alignmentbased variant calling based on a putative SV list to confirm the presence or absene of SVs



OhmX Platform features

Nabsys

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Flexible run times

Cartridges contain preformatted, readyto-use reagents for cleaning, washing, and running experiments on the solidstate 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

introduction into the detector to support longer experimental runs

Easy sample injection

Sample loading port allows for easy injections and walkaway sample

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

Make big discoveries in a small footprint Perform cost-effective, high-quality SV analysis while saving lab space

OhmX Platform Performance Metrics

System

Metric	SV Discovery (Germline)	SV Confirmation
Analysis Pipeline	HCE	SV-Verify
Method	de novo assembly	targeted alignment
Mode of Detection	electronic	electronic
Coverage	100-200x	50-100x
# Genomes/Year	130	240
# Detectors	2	1
Resolution	300 bp	300 bp
Average Tags per 100 kb	25	25
DNA Molecules Measured/Time	> 200k DNA molecules/hour	> 100k DNA molecules/hour

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 Sample Prep Kit	10 samples	10 samples
# Loads per Sample 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



OhmX ordering guide

Category	Part Number	Description	Description	
	900-00002-256	OhmX Analyzer	The Nabsys OhmX Analyzer is a benchtop EGM analyzer featuring precision fluidics and integrated electronics for real-time DNA analysis.	
Instrument	900-00010-002	OhmX Controller	Desktop computer with OhmX software and HDM- Analysis data processing software configured and pre-installed. Includes keyboard and mouse.	
	900-00038-001	OhmX Monitor	A 24-inch color monitor with an LCD display.	
Detectors	900-00058-001	OhmX Detector	Detection chip with integrated gasket featuring an array of nanodetectors for high-sensitivity electronic detection of tagged HMW DNA on the OhmX Analyzer.	
	900-00058-005	OhmX Detector	OhmX Detector bundle - 5 count.	
	900-00058-010	OhmX Detector	OhmX Detector bundle - 10 count.	
	900-00063-001	OhmX Simultaneous Nicking and Labeling Sample Preparation Kit	All sample preparation consumables required to prepare sequence-specific tagged HMW DNA samples for electronic detection in the OhmX Analyzer, 10 samples.	
Kits & Cartridges	900-00056-001	OhmX System Reagent Cartridge	Prefilled cartridge with the necessary reagents to flush, clean, and run detectors for OhmX experiments.	
	900-00057-001	OhmX System Flush Cartridge	Prefilled cartridge containing only DI water for flushing and maintaining analyzer fluidics.	
Accessories	900-00013-001	Waste Tube Replacements	Tubes for liquid waste generated by the OhmX Analyzer (Qty 4).	

Learn more at nabsys.com

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