

GeneMind Copy Number Variation Sequencing (CNV-seq) Solution

Explore Life's Mysteries for Better Healthcare

Highlight:

➤ Wide detection range

Aneuploidy, large fragment deletion/duplication, and CNVs over 100 kb can be detected simultaneously in a single assay.

➤ Fast and easy

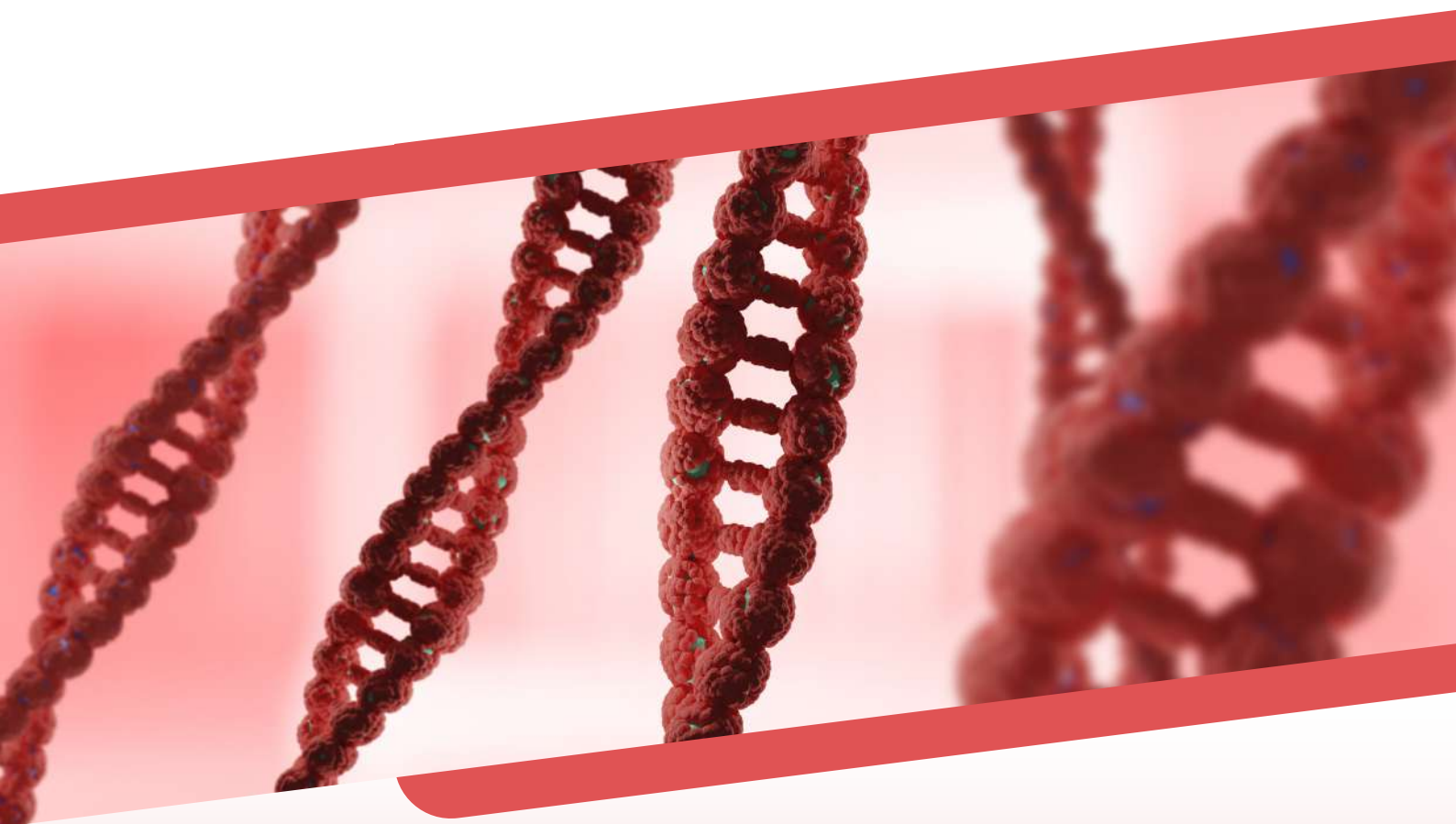
Supports both manual and automated library preparation; seamless integration with bioinformatics analysis platform.

➤ Economical and flexible solution

Single or dual flowcell is optional, NIPT and CNV-seq can be sequenced at the same run.

➤ Accurate data performance

High detection accuracy with good data stability.



Background:

According to the World Health Organization, birth defects affect 4–8% of births globally, with incidence rates varying across regions. These conditions are significant contributors to fetal, perinatal, and infant mortality, as well as childhood disabilities.

Chromosome aberrations, including aneuploidy, triploidy, and large segment deletions or duplications, are major genetic causes of birth defects. Additionally, CNV syndromes, which are caused by pathogenic copy number variations (pCNVs), lead to intellectual disability, multiple congenital anomalies, autistic spectrum disorders and other diseases. GeneMind CNV-seq is a robust solution utilizing GenoLab™ M/FASTASeq™ 300/SURFSeq™ 5000 next-generation sequencing (NGS) systems. It enables accurate detection of chromosomal aneuploidy and CNVs larger than 100 kb.

Which technique to choose?

Technology	Karyotyping	FISH	CMA	CNV-Seq
Cell culture required	Yes	No	No	No
Detection range	Chromosome duplications/deletions/structural abnormalities	Chromosome deletions/duplications in target region	Chromosome duplications/deletions	Chromosome duplications/deletions
Precision	Abnormal chromosome number, structural abnormalities of more than 5Mb	NA	Chromosome number and chromosomal microdeletions/micro-duplications more than 100kb	Chromosome number and chromosomal microdeletions/micro-duplications more than 100kb
Turnround Time (d)	10-15	2-3	10-14	2-3
Advantage	Gold standard for aneuploidy, equilibrium translocations and inversion can be detected	The gold standard for detecting chromosomal mosaicism more than 15%	No restrictions on material, higher resolution to detect micro-deletion/micro-duplication	No restrictions on material, higher resolution to detect micro-deletion/micro-duplication
Disadvantage	Cell culture required, time consuming	Low throughput, high cost, fresh cell required	Low throughput, high cost	Heteroploidy and loss of heterozygosity (LOH) could not be detected

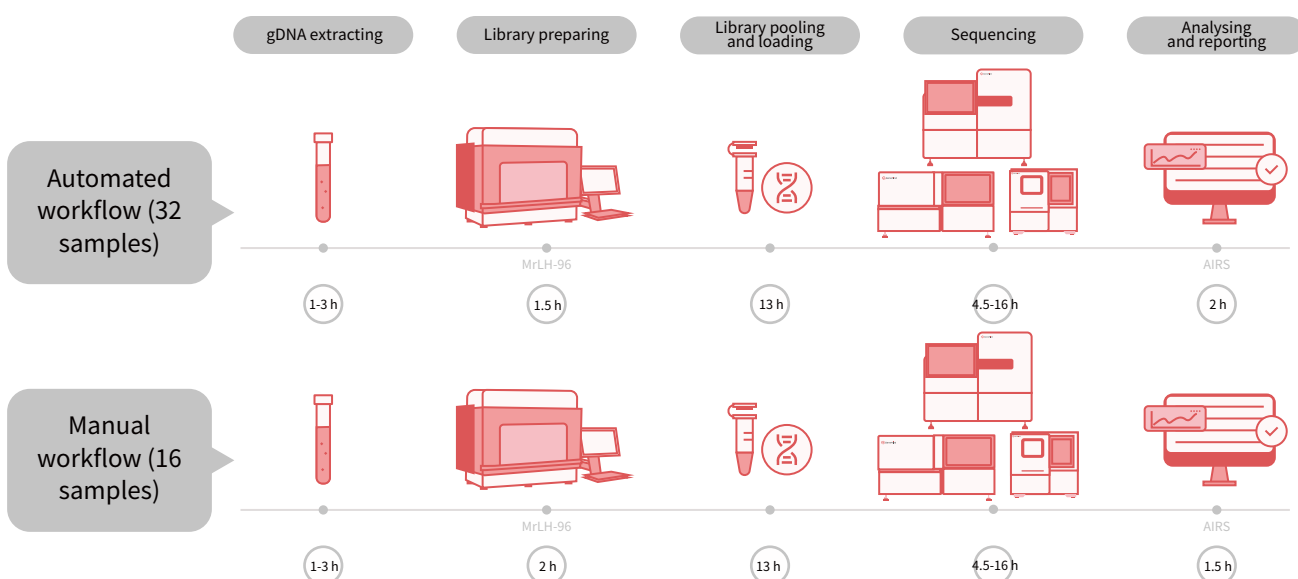
Product Introduction:

Based on the GeneMind GenoScout CNV library preparation reagent, low - pass whole genome sequencing can directly and quickly sequence the abortion tissue and blood of patients accurately, without the cell culture process. It can comprehensively evaluate the aneuploidy of 23 pairs of chromosomes as well as microdeletions/microduplications larger than 100 kb.

Parameter	CNV-seq		
Sample volume	gDNA $\geq$ 10ng		
Sample type	Abortion tissue, Blood		
Sequencing platform	FASTASeq™ 300	GenoLab™ M	SURFSeq™ 5000
Number of samples per run (1 Positive Control +1 Negative)	1 FCM: 4 1 FCH: 12 1 FCP: 24	1 FCM: 12 1 FCH: 24	1 FCH: 96 1 FCP: 160
Turnaround time ¹	22-27 h	30.5-34.5 h	31.5-35.5 h
Read length	SE75 for GenoLab™ M, FASTASeq™ 300, SE50 for SURFSeq™ 5000		
Average unique reads/sample	GenoLab™ M/FASTASeq™ 300 $\geq$ 15 M, SURFSeq™ 5000 $\geq$ 20 M		
Report generation	Local analysis and report system		

1. Please note that the turn-around-time shown in the table is validated in GeneMind's lab, just for reference. It may be different due to the local situation, such as the sample volumes, lab equipment and the proficiency of operator.

Testing Workflow:



Data Performance:

GenoScout Chromosome Copy Number Variation Library Preparation Kit V1.0 was used to construct the library, and the distribution of library fragments was stable, smooth and concentrated.

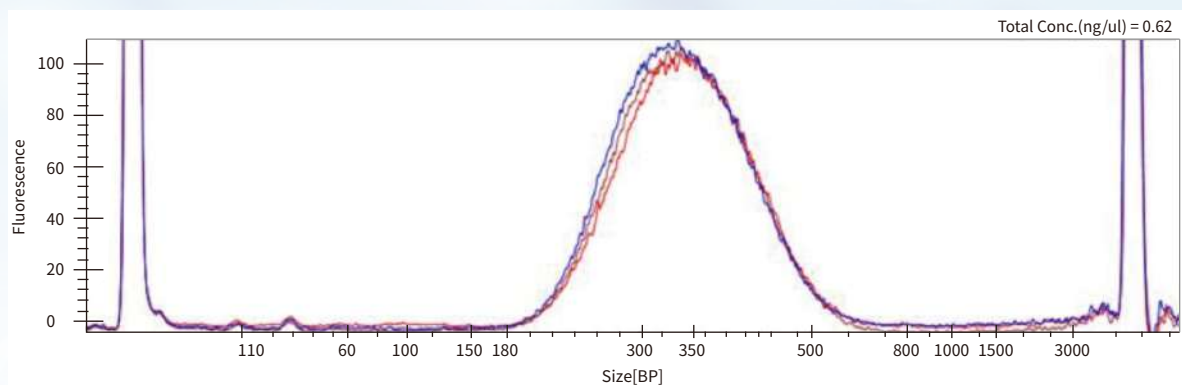


Fig 1. Library size distribution map

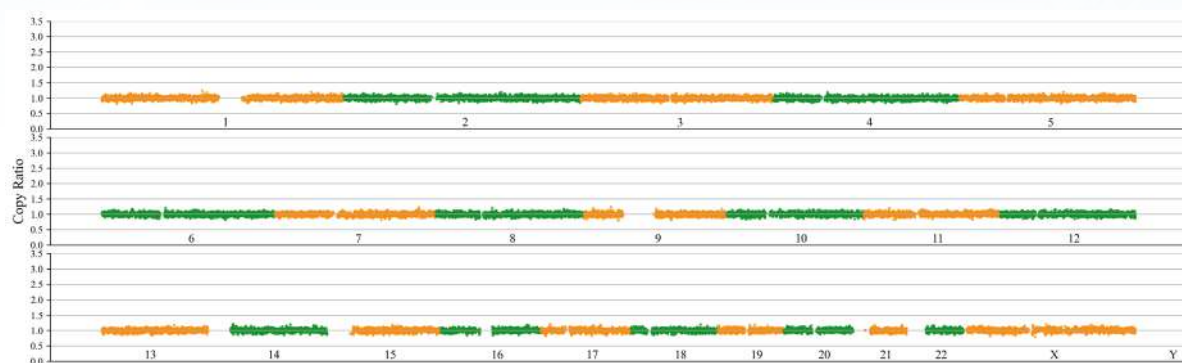


Fig 2. Genome-wide copy number scatter map - negative sample

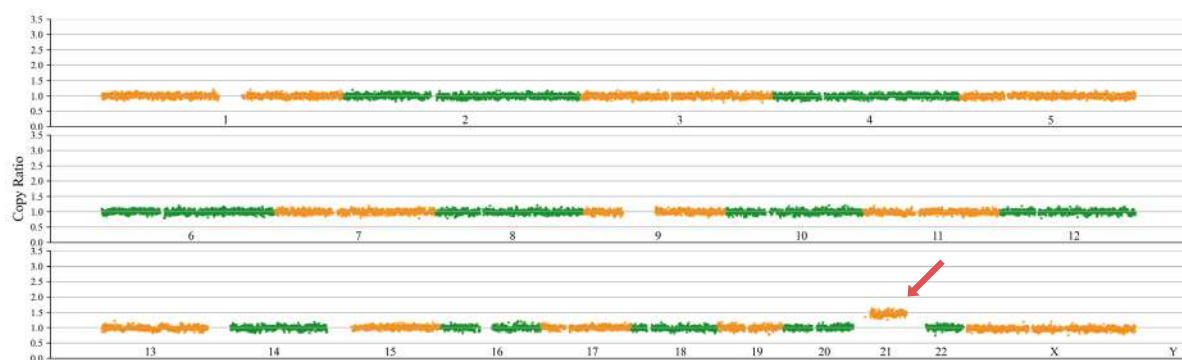


Fig 3. Genome-wide copy number scatter map - T21 sample

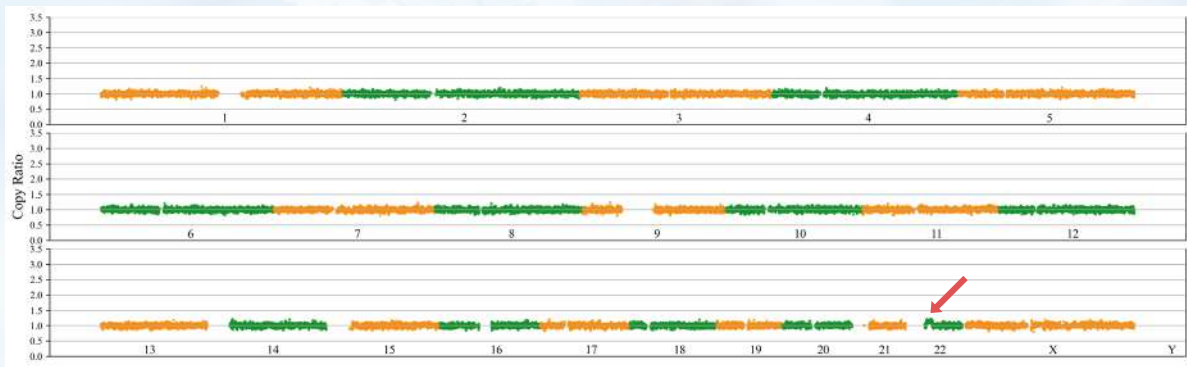


Fig 4. Genome-wide copy number scatter map - mosaics sample

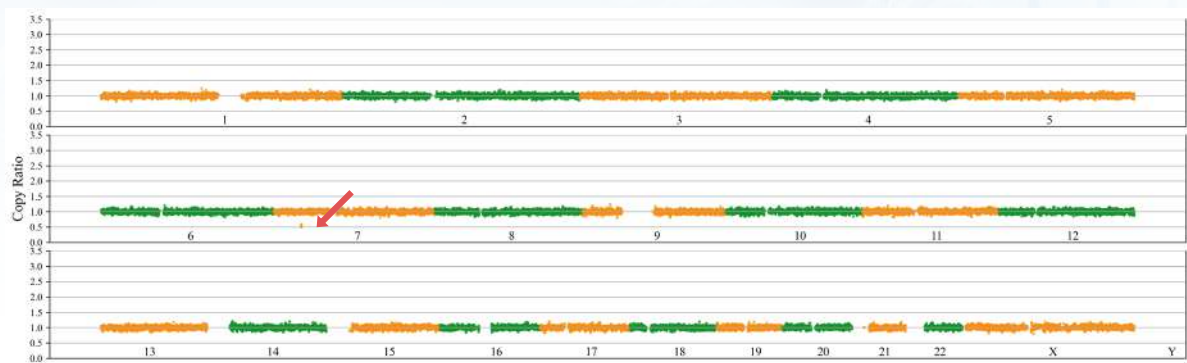


Fig 5. Genome-wide copy number scatter map - 100 kb of microdeletions sample

The data analysis results show that:

- ✓ Reads were evenly distributed in all locations of the genome with good data uniformity (Figure 2)
- ✓ Have relatively high sensitivity for detecting chromosomal aneuploidy abnormalities (Figure 3)
- ✓ Low ratio chimeras as low as 10% has been detected (Figure 4)
- ✓ 100 kb of microdeletions/duplications is accurately detected (Figure 5)

Summary:



High sensitivity

Requires as little as 10 ng of sample input; detects microdeletions and microduplications down to 100 kb.



High speed

1-3 h for library preparation;

< 1 day from sample to sequencing;

Effectively shorten the sample turnaround time



Cost effective

Core materials are developed by GeneMind, reduce the reagent consumption



Good stability

High detection accuracy and superior data quality, with performance validated by multiple clinical partners

About GeneMind:

GeneMind Biosciences Co., Ltd. was founded in 2012. The headquarters is located in Luohu, Shenzhen. Its facility includes more than 10,000 square meters of research and development labs and GMP production line.

Specializing in the independent R&D and manufacturing of molecular diagnosis technology platform centred on DNA sequencing system, GeneMind Biosciences is devoted to constructing a precision medicine ecosystem to serve human life and health through collaboration with genetic testing service providers and medical institutions.

GeneMind has launched the high-throughput sequencing platform GenoLab™ M, FASTASeq™ 300, SURFSeq™ 5000, SURFSeq™ Q, and FASTASeq™ S through independent research and innovation, and offers the “equipment-reagent-flowcell-software” total platform solution. GeneMind is one of the few technology companies with core upstream technology and independent brand of gene sequencing in the world.

※ Disclaimer

This document is based on company data of October, 2025, and is provided for reference only.

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