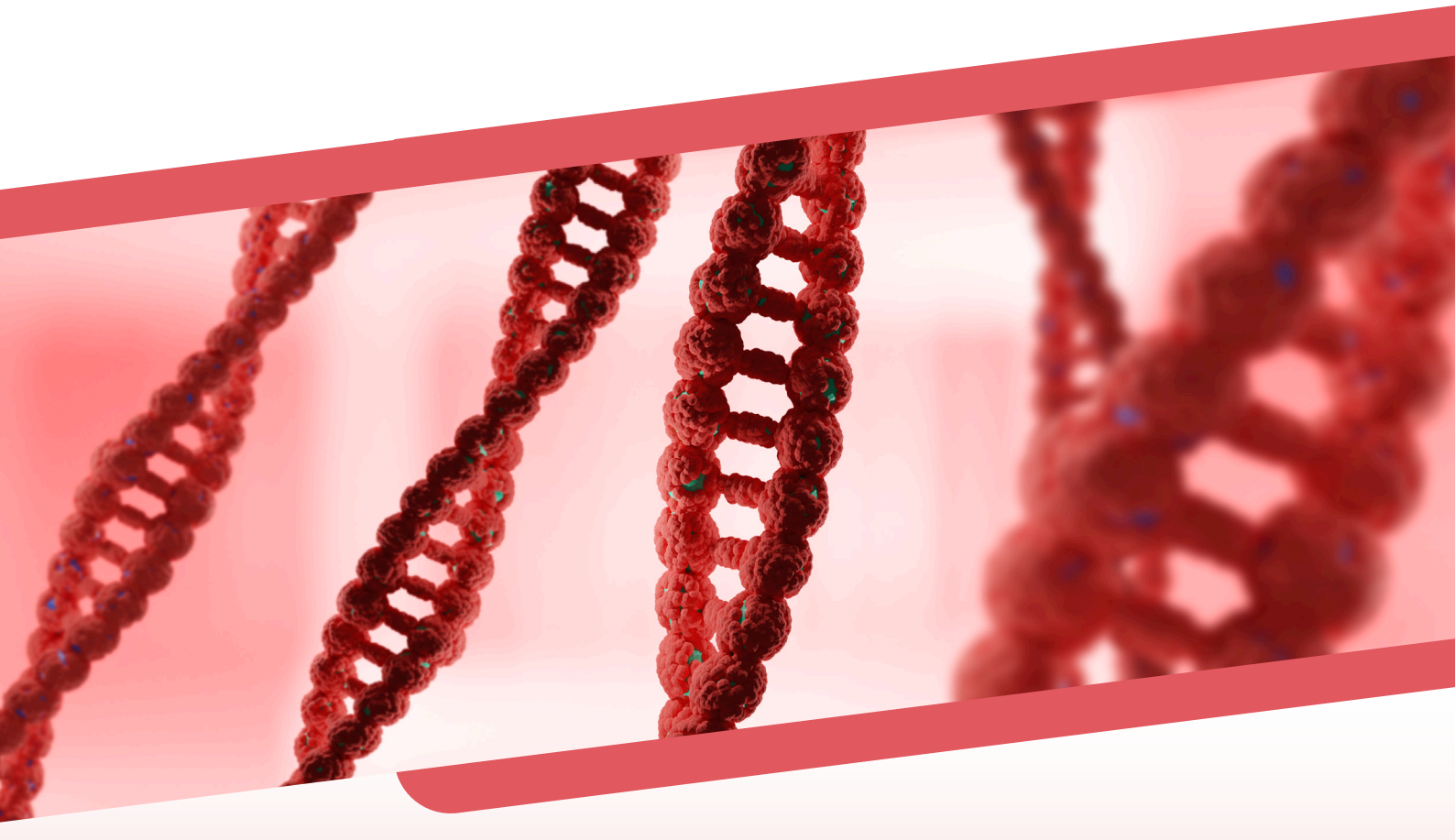
GeneMind

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 in one time
- **Fast and easy**
  Provide manual and automated library preparation workflow, sequencing data is automatically imported into the bioinformatics analysis system
- **Economical and flexible vendor**
  Optional to select single or dual flowcell, 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 worldwide,and their incidence varies between different countries. Birth defects usually lead to fetal death, perinatal death, infant death or child disabilities.

Chromosome aberrations, including aneuploid, triploid and deletion or duplication of large chromosome segments, are a major cause of birth defects. Additionally, copy number variant syndromes (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 solution based on the high throughput sequencing GenoLab M* platform, capable of detecting genetic aberrations, including chromosoma aneuploidy and CNVs larger than 100 kb. This solution is of great importance to assist the analysis of miscarriage genetic factors and to help doctors well guide couples to get pregnant again more scientifically.

Technology	Karyotyping	FISH	CMA	CNV-Seq
Cell culture	Yes	No	No	No
Detection range	All chromosome duplications/deletions/structural abnormalities	Chromosome deletion/duplication in target region	All chromosome duplications/deletions	All chromosome duplications/deletions
Precision	Abnormal chromosome number, structural abnormalities of more than 5Mb	NA	Chromosome number and fragment number are abnormal	Chromosome number and fragment number are abnormal
Turnround Time	10-15d	2-3d	5d	2-3d
Advantage	Gold standard, equilibrium translocations and inversion can be detected	No restrictions on material, easy to operate	No restrictions on material, higher resolution to detect micro-deletion/mirco-duplication	No restrictions on material, higher resolution to detect micro-deletion/mirco-duplication
Disadvantage	Cell culture required, time consuming	Low throughput, high cost	Low throughput, high cost	Heteroploidy and heterozygosity loss could not be detected

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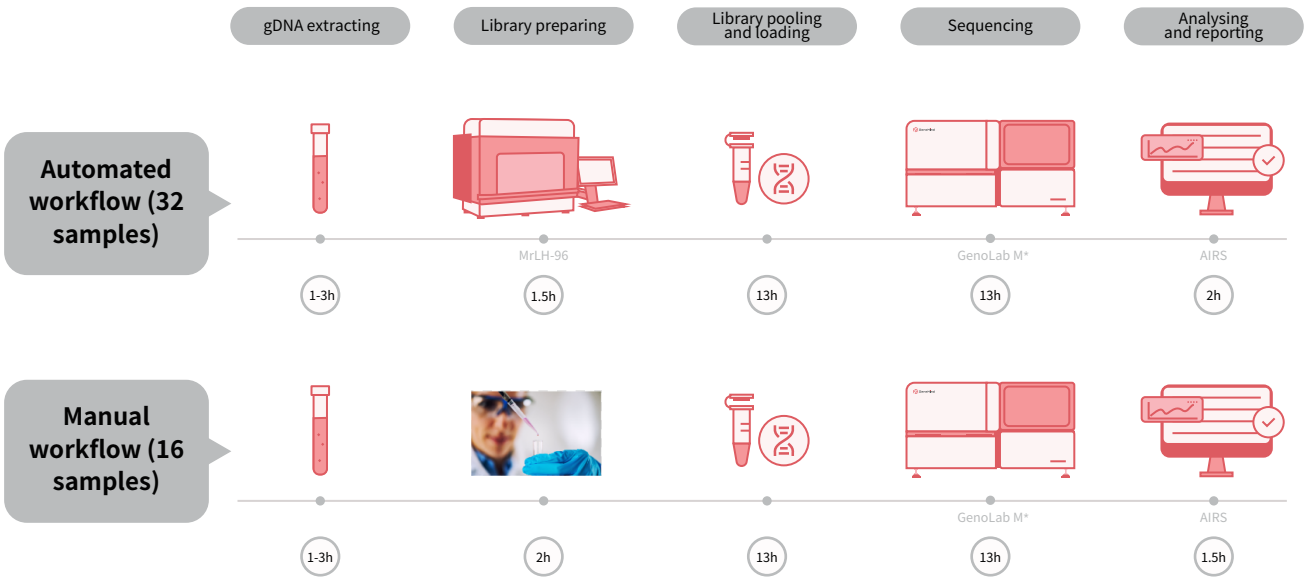
Product Introduction:

Based on GeneMind GenoScout CNV library preparation reagent and GenoLab M* high throughput sequencing platform that can directly sequence abortion/induced labor tissue, amniotic fluid, blood of patients quickly and accurately, without cell culture process. It can comprehensively evaluate aneuploidy of 23 pairs of chromosomes and microdeletions/microduplicates larger than 100 kb.

Parameter		CNV-seq
Sample volume		gDNA⩾10ng
Sample type		Blood, amniotic fluid, abortion tissue, fetal villus tissue, urine etc
Method		NGS
No. of samples per run(1 PC +1 NC included)	GenoLab M*	1 FCM :16 2 FCM/1 FCH :32 1 FCM+1 FCH: 48 2 FCH: 64
Read length		SE75
Average unique reads/ sample		⩾15 M
TAT		2-3 d
Report generation		Local analysis and report system

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.

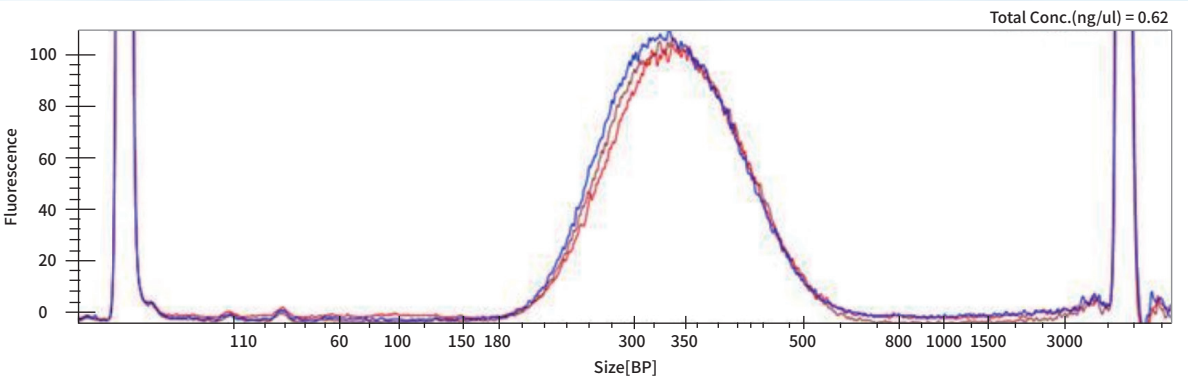


Fig 1. Library size distribution map

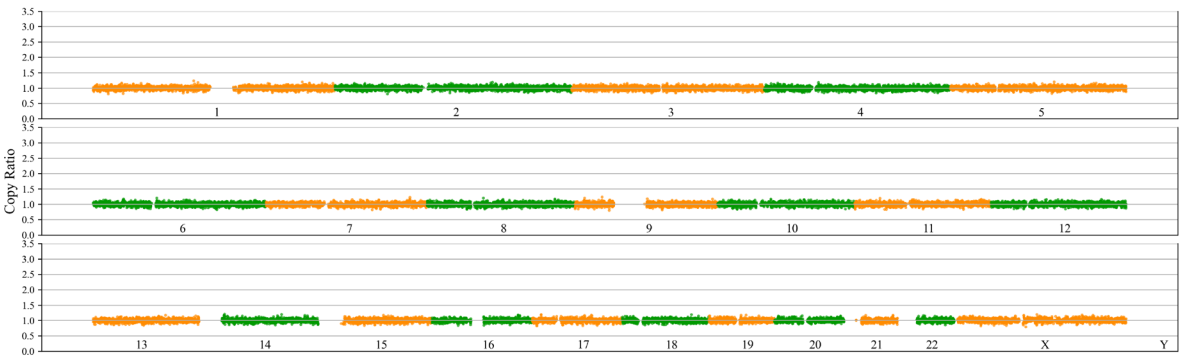


Fig 2. Genome-wide copy number scatter map - negative sample

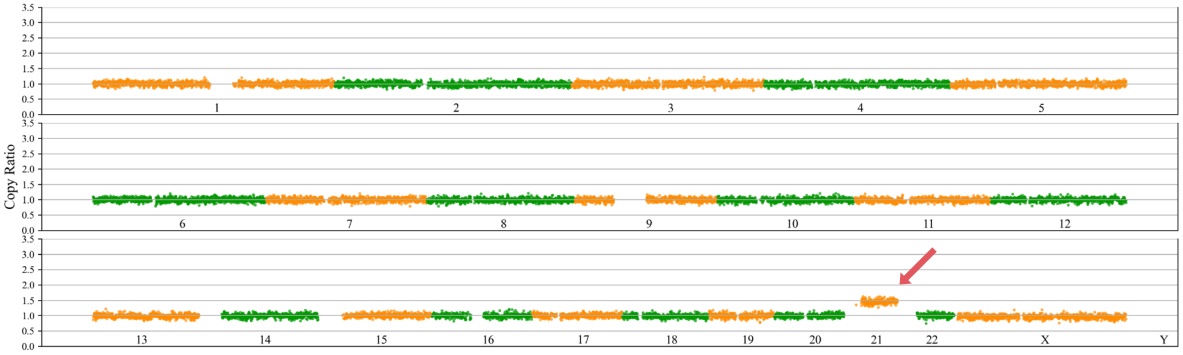


Fig 3. Genome-wide copy number scatter map - T21 sample

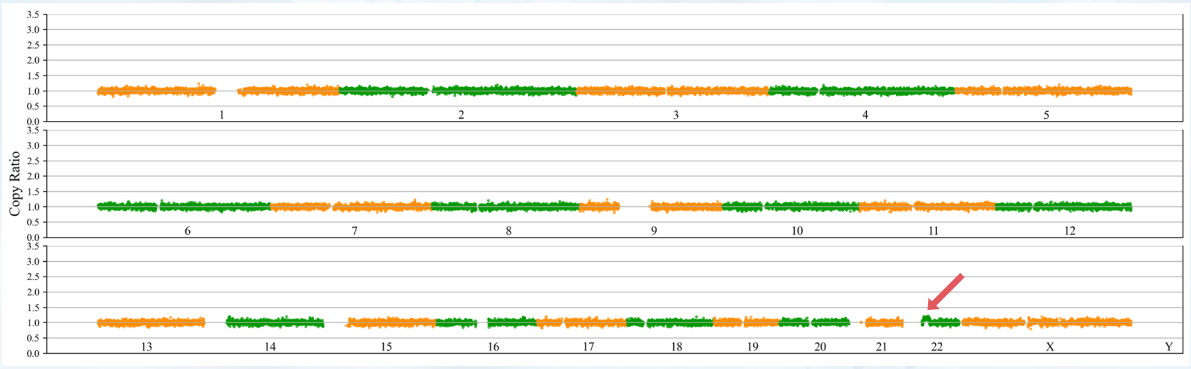


Fig 4. Genome-wide copy number scatter map - mosaics sample

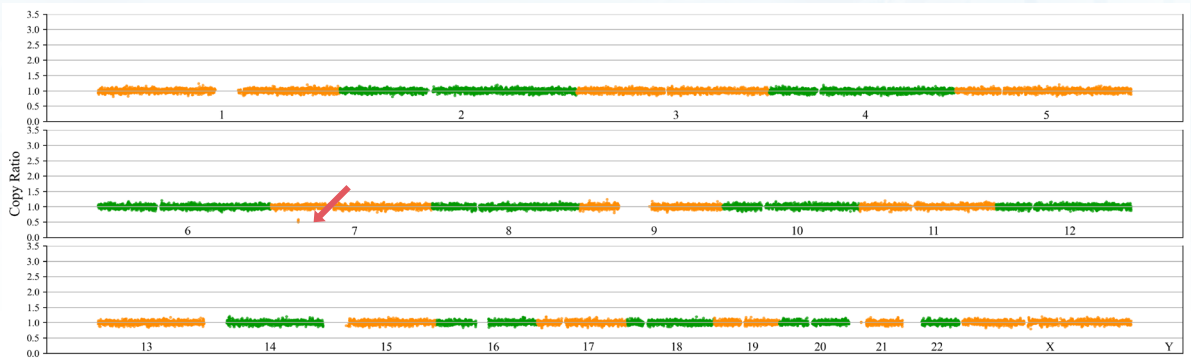


Fig 5. Genome-wide copy number scatter map - 100 kb of microdeletions sample

The data analysis results show that:

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

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Summary:

High sensitive

The sample input volume requirement as low as 10ng, microdeletion/microduplications as low as 100Kb can be successfully detected as well

High speed

1-3h for library prep;

< 1 day from sample to sequencing;

Effectively shorten the sample turnround time

Cost effective

Core materials are development by GeneMind, reduce the reagent consumption

Good stability

High detection accuracy and good data performance, solution has been backed by several 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* and SURFSeq 5000* 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.

*Unless otherwise informed, GeneMind sequencing platform and related sequencing reagents are not available in the USA, Canada, Australia, Japan, Singapore, Western Europe, Southern Europe and Nordic countries yet.

GeneMind Biosciences Co., Ltd.

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