

PGT-A

Preimplantation Genetic Testing for Aneuploidy (PGT-A) of GeneMind

Easy-to-use

Automated and simplified sequencing and data analysis workflow

Fast

Quick test from sample preparation to final report within 12 hours

Flexible

Compatible with common used NGS library preparation kits and bioinformatics tools

High sensitive

Accurately detect different types of mutations even in low copies



AAGAGC CcAACT CTAAT
GTGTC ATG AT GATAA
TACG CACGAA
TGGCC CAG CACTT
TATAC CTTCA
GATAGT GAG CCGG
ACCTGGC AGACCTG
ATGGGCA TTTT
TGGGCA TTTT
GATTAACG
TTATCTTCC

◆ Introduction of PGT-A

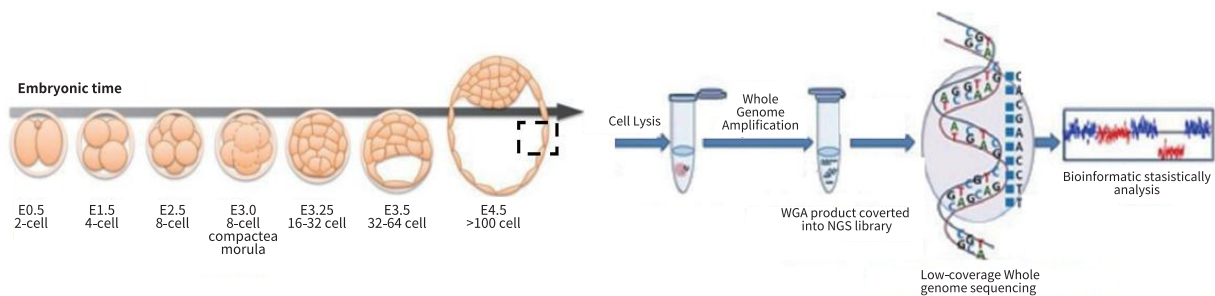
Single-cell whole genome amplification (WGA) and low-coverage whole genome sequencing technologies are used to detect chromosomal copy number variations (CNVs) by sequencing the genomic DNA from 5-10 embryo cells. The differences in the number of respective chromosomes are statistically analyzed using the data analysis software to evaluate whether any CNVs are detected in the whole cell genome.

Preimplantation genetic testing for aneuploidy identifies chromosomal abnormalities in embryo samples for their search of *in vitro* fertilization (IVF) or intracytoplasmic sperm injection (ICSI). These genetic insights may increase the probability of success for future pregnancies and healthier generations.

Using GeneMind single-cell amplification kits, PGT-A library preparation sets and together with GenoLab™ M/ FASTASeq™ 300/SURFSeq™ 5000/ FASTASeq™ S next-generation sequencing (NGS) system could accurately detect different mutations including 23 pairs of chromosomal aneuploidy abnormalities, microdeletions/microduplications over 4 Mb in the whole region.

◆ Detection Process

Picogram quantities of genomic DNA is extracted from embryo cells and then subjected to do the WGA. The amplification products can be sequenced by the NGS platform and then automatically analyzed to generate the final report.



◆ Manual Workflow (48 samples around 24 hr)

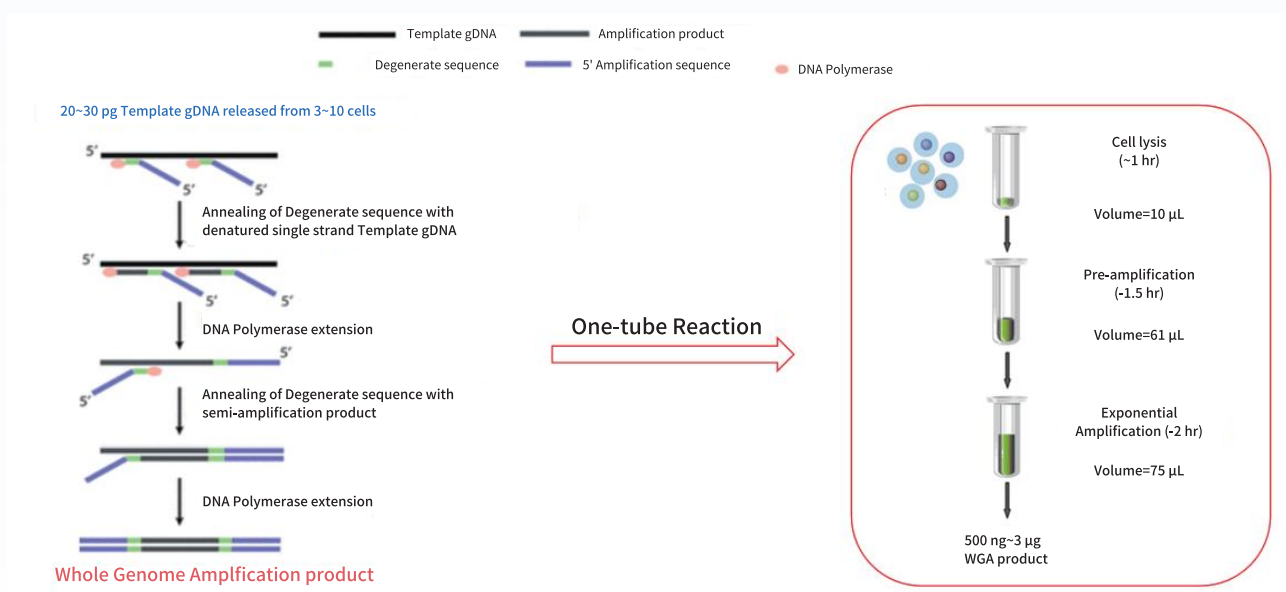


◆ Automated Workflow (48 samples around 24 hr)

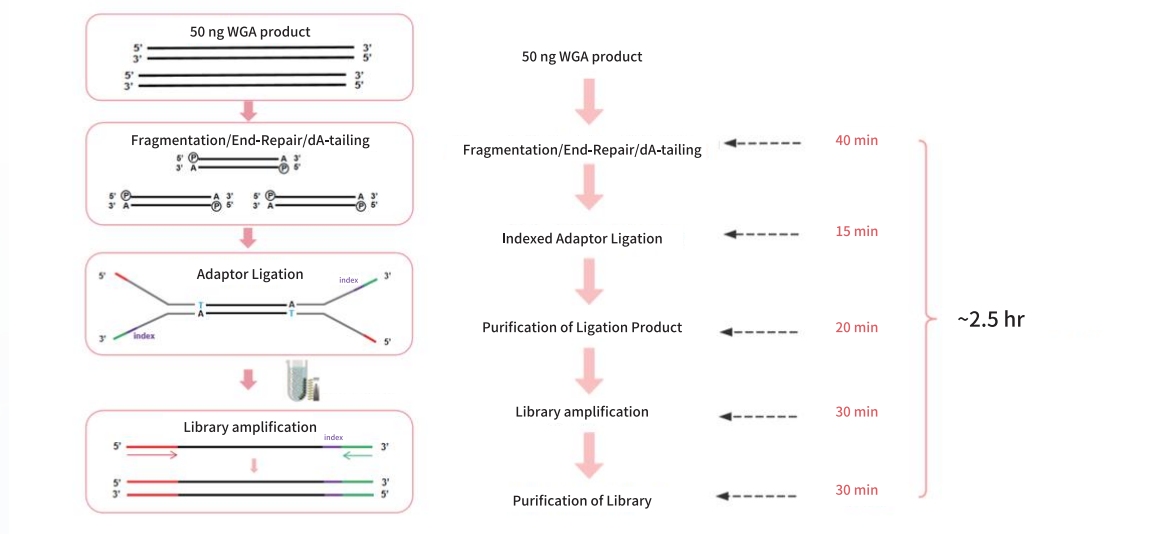


Note: Please note that the time shown in the workflow is validated in GeneMind's lab on October, 2025, it may be different due to the local situation, such as the sample volumes, lab equipment and the proficiency of operator.

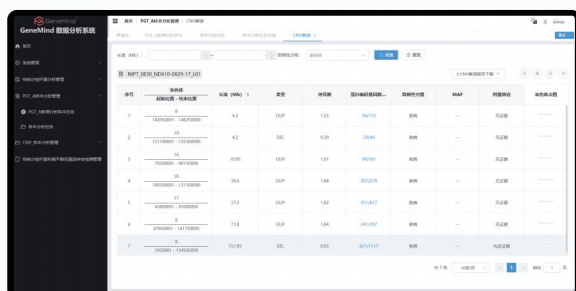
◆ Single-cell Genome Amplification Process



◆ Library Construction Process

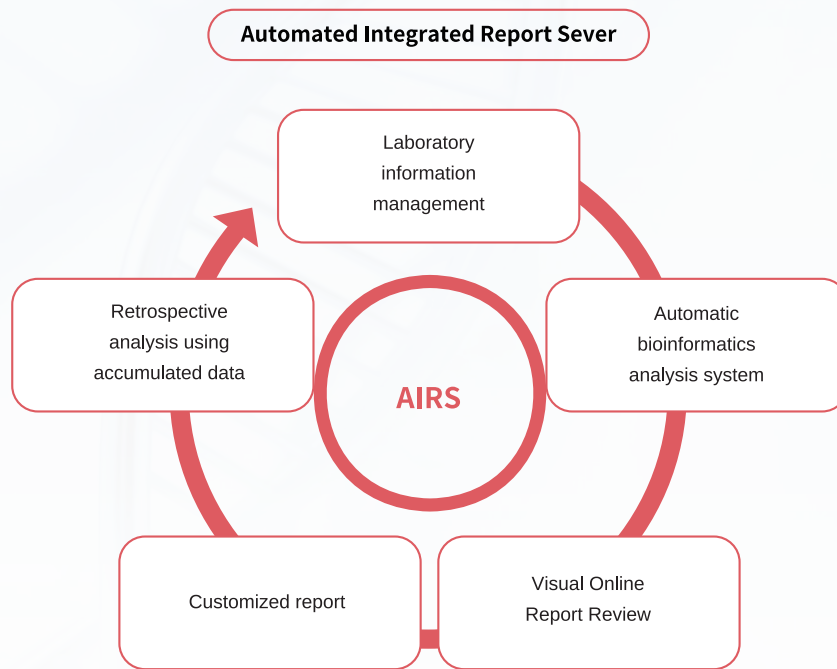


◆ GeneMind Data Analysis System



Chromosomal aneuploidy data analysis and report software installed in on-site server

The chromosomal aneuploidy data analysis software can accurately detect chromosomal aneuploidy, chromosomal deletions or duplications over 4 Mb in the whole region as well as more than 30% of chimeric abnormalities.



◆ Data Performance

WGA is performed using the GenoVerse Single-cell WGA Kit, and the library of WGA products is constructed using the Preimplantation Genetic Testing For Aneuploidy Detection Library preparation Kit.

The obtained library is sequenced and analyzed using the GenoLab™ M/ FASTASeq™ 300/SURFSeq™ 5000/ FASTASeq™ S NGS systems and the GeneMind chromosomal aneuploidy data analysis software, respectively.

The data analysis results show that reads are quite evenly distributed in the whole genome (Figure 1); The chromosomal aneuploidy abnormalities can be accurately identified even in quite low copies (Figure 2); Some microduplications as low as 1 Mb can be successfully detected as well (Figure 3).

The results show that GenoVerse Single-cell WGA Kit has good uniformity and could be used for PGT-A analysis when combined with Preimplantation Genetic Testing For Aneuploidy Detection Library preparation Kit.

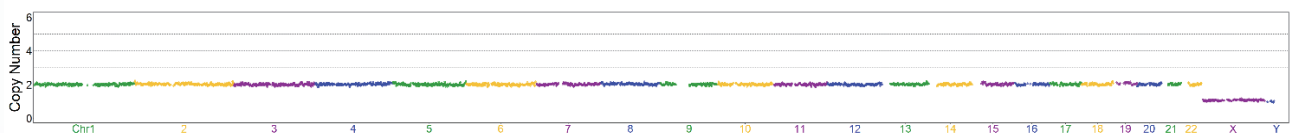


Figure 1. Scatter plot of whole genome copy number — negative sample

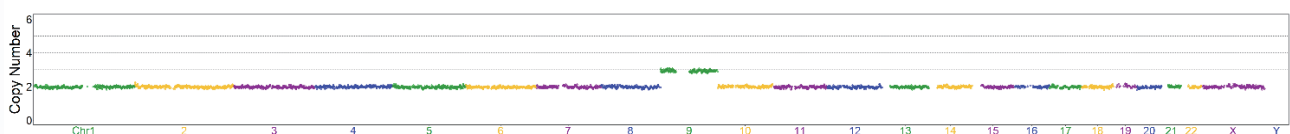


Figure 2. Scatter plot of whole genome copy number — T9 sample

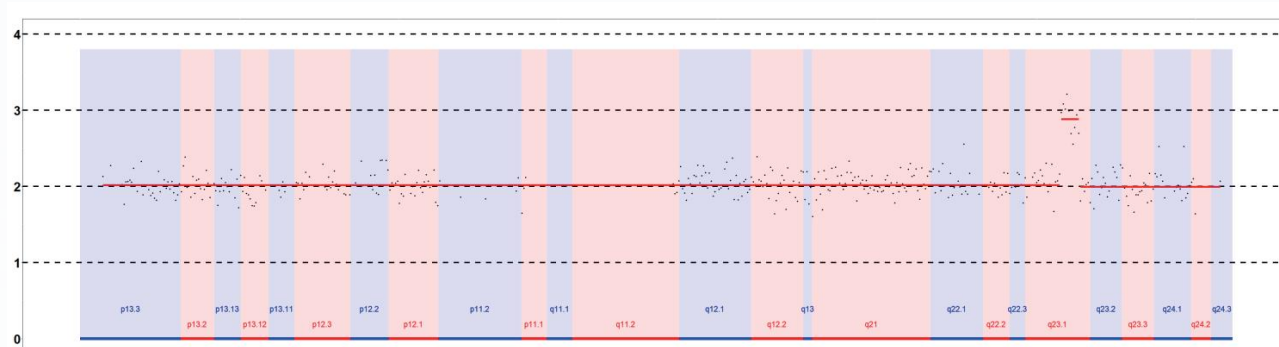


Figure 3. Scatter plot of Chr16 copy number — dup (16) (q23.1) sample

In addition, the chromosomal aneuploidy data analysis software includes the CNV annotation system, which can automatically annotate CNV results and help end-users further understand the relation between diseases and CNVs. The CNV annotation system provides information about microdeletions/microduplications from databases such as OMIM, Decipher, ClinVar, ClinGen, dbVar, and MedGene. Subsequently, it automatically ranks the CNV results according to ACMG guidelines, evaluates their clinical significance and generates interpretation reports.

◆ GeneMind PGT-A Solution

PARAMETER				
Sample volume	5-10 embryo cells			
Library preparation	WGA			
Method	Whole genome sequencing			
Read length	SE75 for GenoLab™ M/FASTASeq™ 300/FASTASeq™ S, SE50 for SURFSeq™ 5000			
Sequencing platform	FASTASeq™ S	FASTASeq™ 300	GenoLab™ M	SURFSeq™ 5000
Number of samples/run	1 FCL: 3 1 FCM: 6	1 FCM: 16 1 FCH: 36 1 FCP: 64	1 FCM: 32 1 FCH: 64	1 FCH: 160 1 FCP: 280
Turnaround time ¹	12-14 h	13.5-18 h	22-25.5 h	23-26.5 h
Reads requirement/sample	GenoLab™ M/FASTASeq™ 300/FASTASeq™ S: Unique reads per sample ≥ 3.5 M, SURFSeq™ 5000: Unique reads per sample ≥ 4 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.

◆ About GeneMind



GeneMind Biosciences Co., Ltd was established in 2012 and is headquartered in Shenzhen, China. Its facility includes more than 10,000 square meters of research and development laboratories and GMP production lines.

Specializing in the independent R&D and manufacturing of molecular diagnosis technology platform centred on DNA sequencing system, GeneMind 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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