GeneMind

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 24 hours

Flexible

Compatible with common used NGS library prep kits and bioinformatics tools

High sensitive

Accurately detect different types of mutations even in a low copies

◆ Introduction of PGT-A

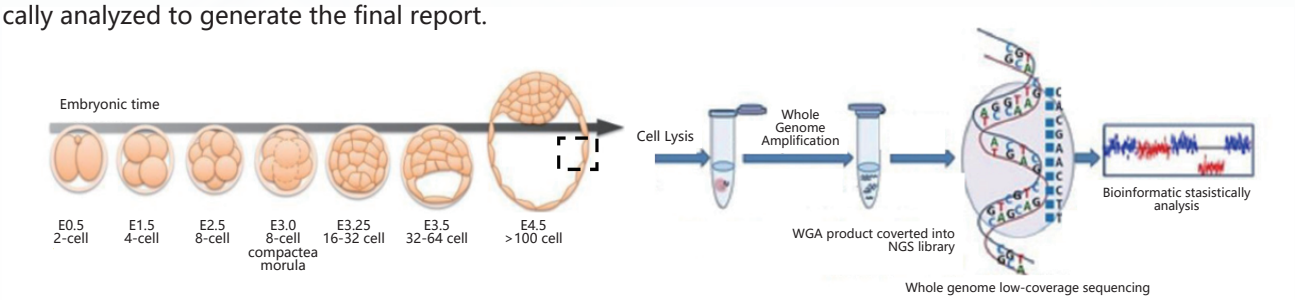
Single-cell whole genome amplification (WGA) and whole genome low-coverage sequencing technologies are used to detect chromosomal copy number variations (CNVs) by sequencing the genomic DNA from 5-10 embryo cells. The differences in the number of respective chromosomes are statistically analyzed using 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 the research of in vitro fertilization (IVF) or intracytoplasmic sperm injection (ICSI). These genetic insights may increase the probability of success for future pregnancies and healthier future generations.

Using GeneMind single-cell amplification kits, PGT-A library preparation sets and together with GenoLab M* / FASTASeq 300*/ SURFSeq 5000* next-generation sequencing system could accurately detect different mutations including 23 pairs of chromosomal aneuploidy abnormalities, microdeletions / microduplications over 4 Mb in the whole region, and microdeletions / microduplications over 1 Mb in the known region.

◆ Detection Process

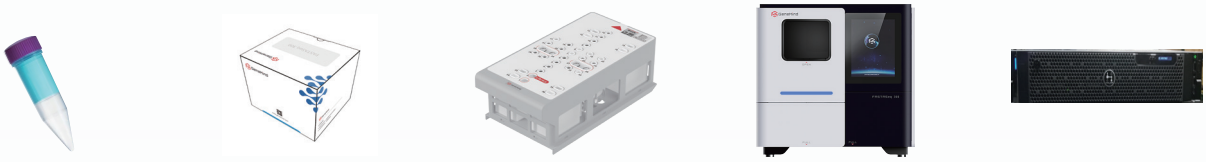
Picogram quantities of genomic DNA is extracted from embryo cells and then subjected to do the WGA. The amplification products can be sequenced by the high-throughput gene sequencing platform and then automatically analyzed to generate the final report.



◆ Workflow on GenoLab M* Sequencing System

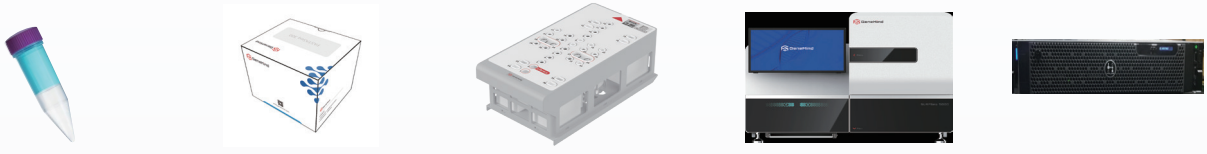
4 h WGA | 2.5 h Library preparation | 0.5 h Library pooling and loading | 15 h Sequencing | 2 h Analysing and reporting

◆ Workflow on FASTASeq 300* Sequencing System



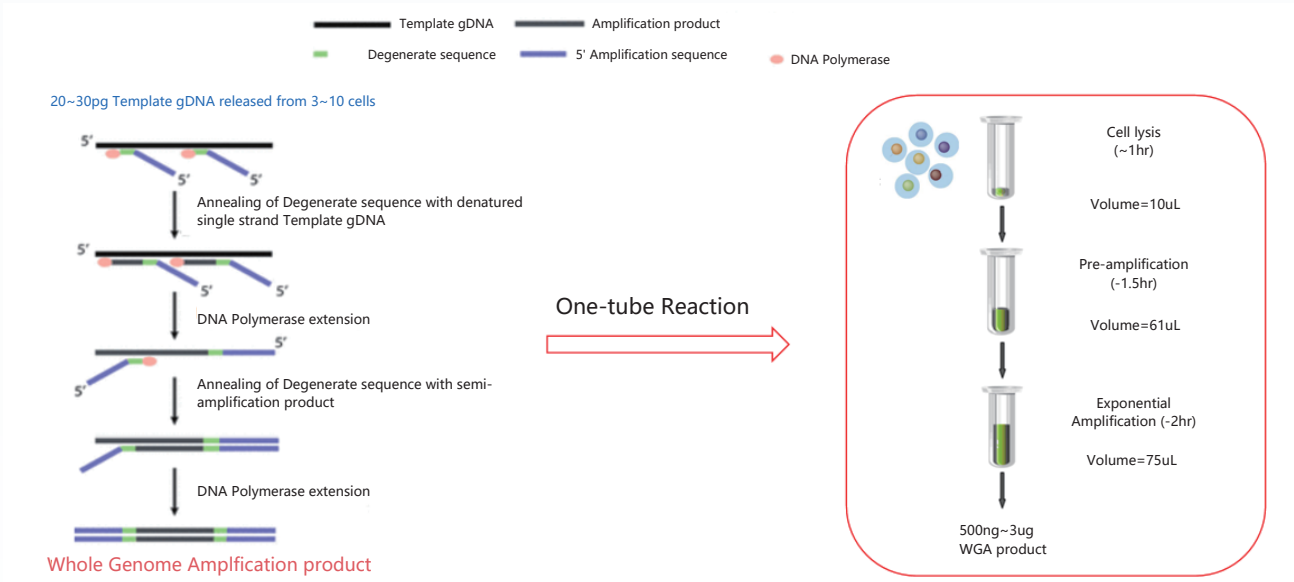
4 h WGA | 2.5 h Library preparation | 0.5 h Library pooling and loading | 7 h Sequencing | 2 h Analysing and reporting

◆ Workflow on SURFSeq 5000* Sequencing System

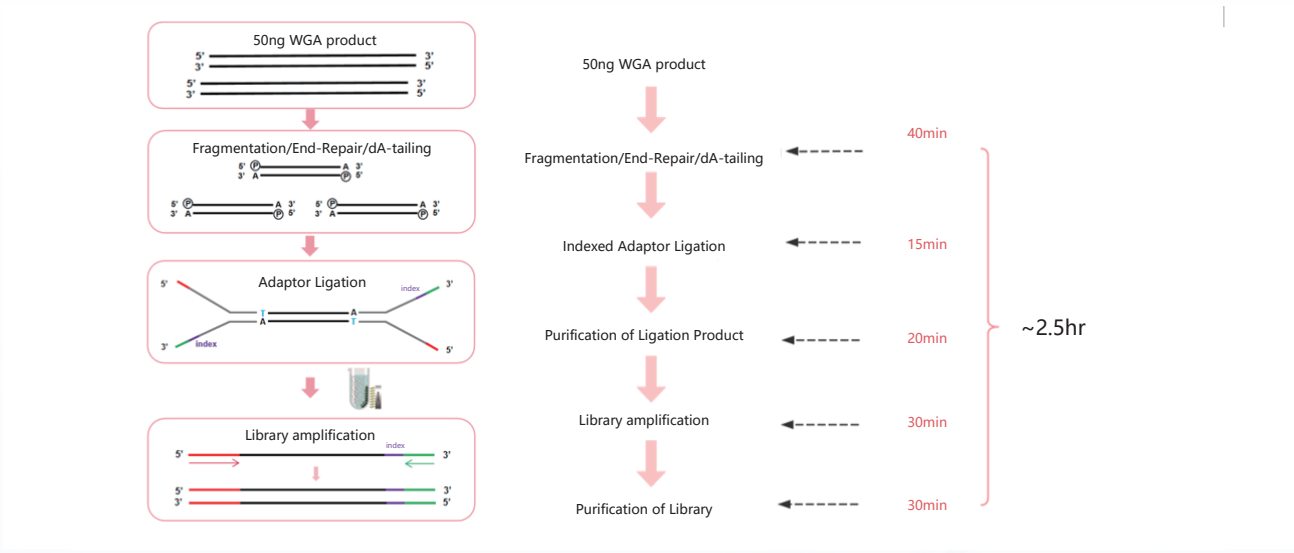


4 h WGA | 2.5 h Library preparation | 0.5 h Library pooling and loading | 14 h Sequencing | 2 h Analysing and reporting

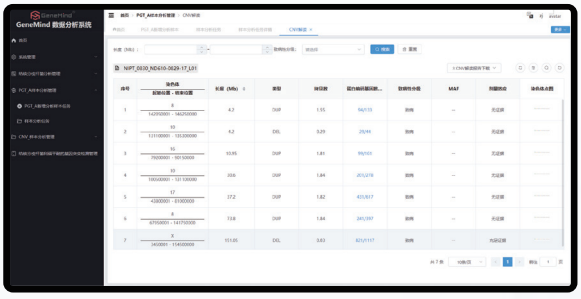
◆ Single-cell Genome Amplification Process



◆ Library Construction Process

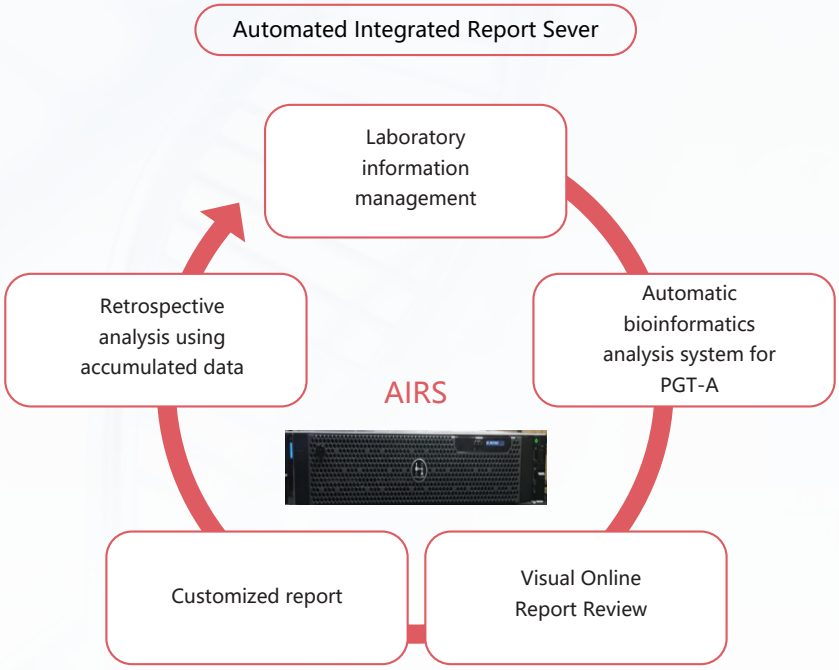


◆ GeneMind Data Analysis System



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 1 Mb in the known region as well as more than 30% of chimeric abnormalities.



◆ Data Performance

WGA is performed using the GenoVerse Single-cell WGA Kit V1.0, and the library of WGA products is constructed using the Preimplantation Genetic Testing For Aneuploidy Detection Library preparation Kit V1.0.

The obtained library is sequenced and analyzed using the GenoLab M* / FASTASeq 300*/ SURFSeq 5000* next-generation sequencing system 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 a 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 V1.0 has good uniformity and could be used for PGT-A analysis when combined with Preimplantation Genetic Testing For Aneuploidy Detection Library preparation Kit

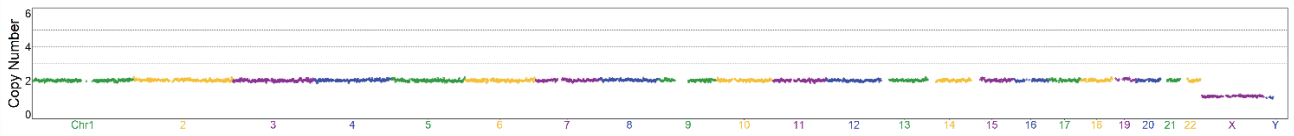


Figure 1. Scatter plot of whole genome copy number — negative samples

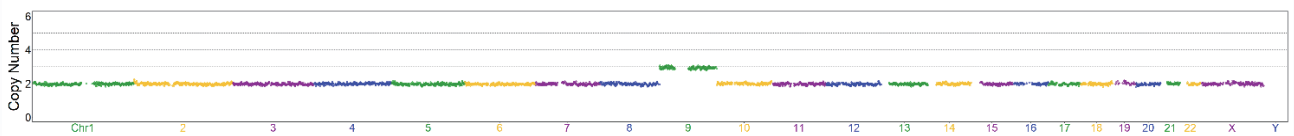


Figure 2. Scatter plot of whole genome copy number — T9 samples

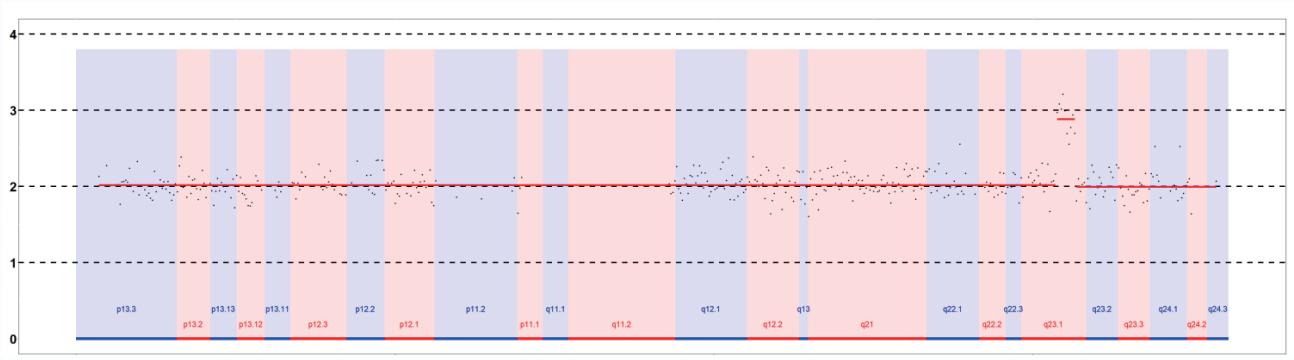


Figure 3. Scatter plot of Chr16 copy number — dup (16) (q23.1) samples

In addition, the chromosomal aneuploidy data analysis software includes the CNV interpretation system, which can automatically annotate CNV results and help end-users further understand the relation between the clinical symptoms and CNVs. The CNV interpretation system provides information about microdeletions/micro-duplications from databases such as OMIM, Decipher, ClinVar, ClinGen, dbVar, and MedGene. The CNV results will be ranked automatically according to ACMG guidelines, evaluates its clinical significance, and provides interpreta-

◆ GeneMind PGT-A Solution

PARAMETER			
Sample volume	5-10 embryo cells		
Library preparation	WGA		
Method	Whole genome sequencing		
Read length	SE75		
Sequencing platform	GenoLab M*	FASTASeq 300*	SURFSeq 5000*
No. of samples/run	1 FCM : 32 2 FCM/1 FCH : 64 1 FCM+1 FCH: 96 2 FCH: 128	1 FCM : 12 1 FCH : 32	1 FCM : 64 2 FCM/1 FCH : 256 1 FCM+1 FCH: 320 2 FCM: 128 2 FCH: 512
Turn-around-time	24 h	16 h	23 h
Reads requirement/sample	Average raw reads per sample ≥ 5M, or unique reads per sample ≥ 3.5M		
Report generation	Local analysis and report system		

◆ 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 SURF-Seq 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.

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*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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