# Preimplantation genetic testing for Aneuploidies(PGT-A)

ДИА•М
современная лаборатория

www.dia-m.ru
заказ on-line

## High Accuracy

CYGNUS S100 Gene Sequencer: Q40＞80%, High sensitivity for positive detection

Sensitivity and Specificity Up to 99%.

## Comprehensive

Detect Aneuploidy and Deletions

Duplications >4Mb of All 46 ChromosomesReliable High standard.

## Cost-effective

Good Performance with Lower Price.

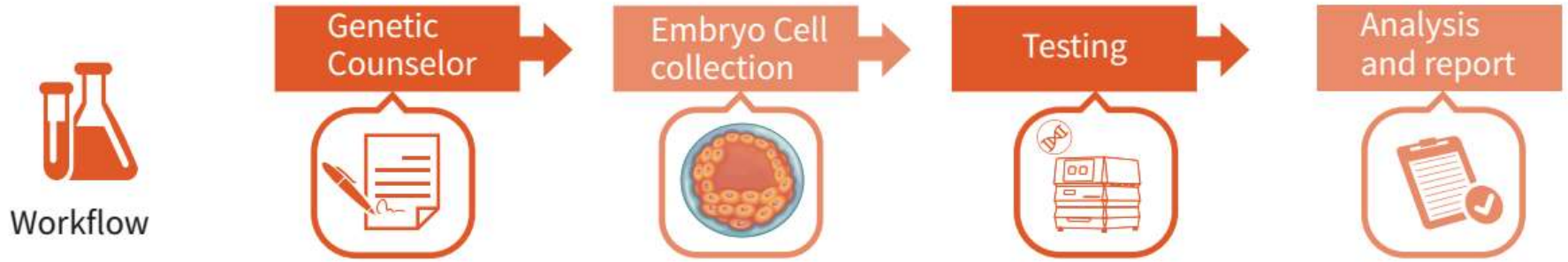
## Multiple Services

Provide both Offshore and Tech Transfer Services .

# Preimplantation genetic testing for Aneuploidies(PGT-A)

**Preimplantation genetic testing for Aneuploidies(PGT-A) uses biopsied embryos on Day3 or Day5/6 by removing a single cell or a few cells. The cells are then amplified and tested by using Next-Generation Sequencing technologies. lt combines low coverage whole genome sequencing of cell WGA products with self-developed bioinformatics software to analyze both embryo aneuploidy and > 4Mb duplication and deletion of 23 chromosomes. The test will detect chromosomal aneuploidy, and chromosome deletion/duplication with larger than or equal to 4Mb reads.**

# Whole solutions




Conduct pre-test counseling and ensure patient provides informed consent for test

Discuss and fill in the Consent Form/Test Request Form with the patient

Embryo biopsy for laboratory operations

Cell lysis and WGA&QC

Library preparation & QC (8h)

Sequencing (20h)

Data analysis & QC (4-7h)

Result interpretion & report

# Single-cell Genome Amplification Process

**3-8cells**

↓

**Cell lysis(12μL)**

50℃ 50 min,
75℃ 15 min

↓

**Whole genome amplification(25μL)**

~2h40min

↓

**WGA product**

> 500ng

# Library Construction Process

**WGA product**
50ng

↓

**Fragmentation, End Preparation & dA-tailing(60μL)**
4℃ 1 min; 37℃ 17 min; 72℃ 20 min

↓

**Adapter Ligation(100μL)**
20℃ 15 min

↓ Purification of Ligation Product

**Index PCR Amplification**
~25min

↓ Purification of PCR Product

**Purified Libary**

# CYGNUS Data Analysis System

# Data Performance

**WGA is performed using the Preimplantation genetic testing for Aneuploidies(PGT-A) WGA Kit, and the library of WGA products is constructed using Preimplantation genetic testing for Aneuploidies(PGT-A) Library Kit.**

**The obtained library is sequenced and analyzed using CYGNUS-S100 high-throughput sequencing Platform and the CYGNUS 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）.**

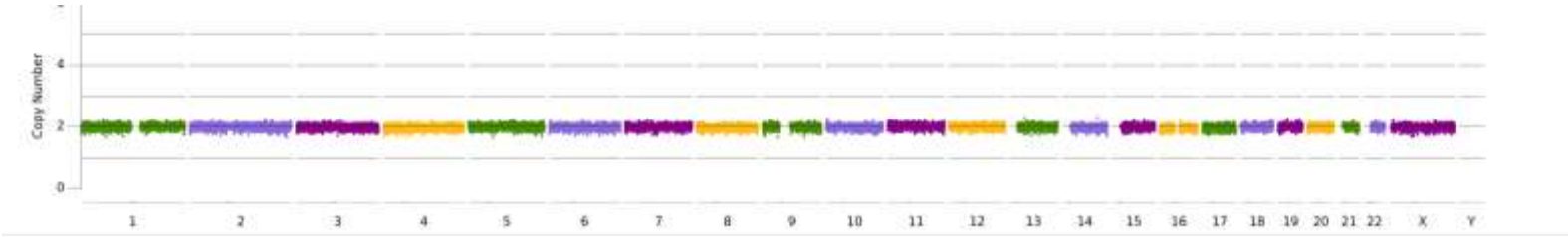



Figure 1

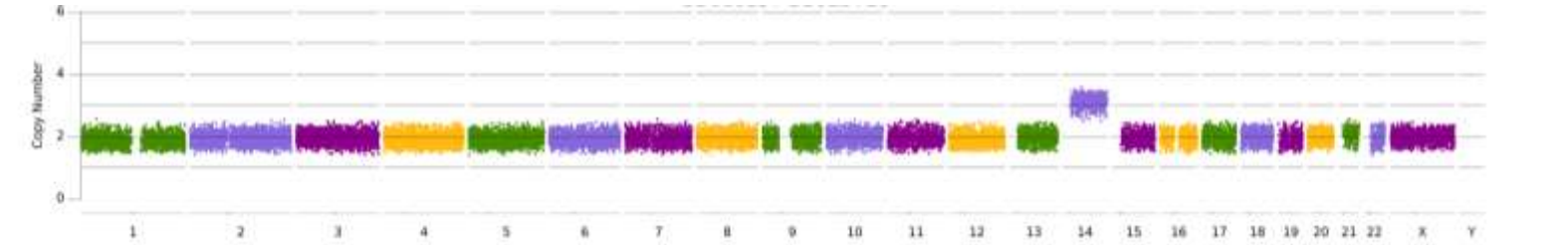



Figure 2

# Data Performance

**The results show that Preimplantation genetic testing for Aneuploidies(PGT-A) WGA Kit has good uniformity and could be used for PGT-Analysis when combined with Preimplantation genetic testing for Aneuploidies(PGT-A) Library Kit. It is easy to operate and superior in performance.**

**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/microduplications from databases such as OMIM, Decipher, ClinVar, ClinGen, dIbVar, and MedGene. The CNV results will be ranked automatically according to ACMG guidelines, evaluates its clinical significance, and provides interpretation reports.**

# Preimplantation genetic testing for Aneuploidies(PGT-A)

| Parameter | Preimplantation genetic testing for Aneuploidies(PGT-A) |
|---|---|
| Specimen | Embryo cells (3-5days) |
| Technology | LOW coverage whole genome sequencing (WGS) |
| Platform | Cygnus S100 Sequencing Platform |
| Read length | SE150 |
| No. of samples per run | 40 |
| Chip Spec | 80-100M |
| Average Unique Reads / sample | 2M |
| Turn-around time<br>Report generation | Local analysis and report system |
| Turn-around time | 7 working days |

# Sample Requirement

| Sample Type | Quantity | Requirements | Shipment |
|---|---|---|---|
| Embryo cells | 5-8 cells | 0.2 or 0.5mL centrifuge tubes are used to send cell samples | Stored at -80 °C, shipped with dry ice |