

# About

Chromosomal abnormalities result from the deletion or duplication of parts of chromosomes. Chromosomal abnormalities can cause miscarriage, fetal malformation, severe congenital anomalies, significant intellectual and physical disabilities.

BGI VISTA™ Chromosome Sequencing uses low-coverage whole-genome sequencing to detect chromosome abnormalities before or during pregnancy.

To date, BGI has processed more than 50,000 samples and detected over 15,000 positive cases.

## **Conditions Screened**

#### Option 1: BGI VISTA™ Chromosome Sequencing-1M

- Triploid
- Aneuploidy for all chromosomes
- deletions or duplications >1Mb

### Option 2: BGI VISTA™ Chromosome Sequencing-100K

- Aneuploidy for all chromosomes
- deletions or duplications >100Kb

# BGI VISTA™ Chromosome Sequencing can be provided for people who

- want to confirm the high-risk screening test results
- have experienced a pregnancy loss
- with a pregnancy associated with fetal abnormalities by ultrasound
- Is suspected of chromosome abnormality
- Maternal age is more than 35 years of age

# Why Choose BGI VISTA™ Chromosome Sequencing?

Prenatal diagnostic testing is used to confirm screening test high-risk results, including a family history of chromosome abnormality, and to ensure that the fetus does not carry significant chromosomal changes. This test can also be used to investigate the causes of infertility and pregnancy loss.

- Detection of Triploid, all whole chromosome abnormalities as well as deletions/duplications
- High resolution of 100K
- High sensitivity and specificity
- A small amount of sample is required

# Sample Requirements

SAMPLE TYPE		QUALITY	REQUIREMENT	SHIPMENT
Embryonic sample	Tissue from abortion	>100mg	Wash with NS or PBS till colorclarified	Stored at -20°Cfor short term, -80°C for long term; Shipped with dry ice. Please
	Amniotic fluid	≥10ml		
	Cord blood	≥1ml	Use EDTA tube for sampling	
Peripheral blood		Adult ≥5ml Newborn >1ml	Genetic invert the EDTA tube to avoid hemolysis	avoid vibrations or shock
DNA		>1ug	Sample Concentration> 30ng/ul OD260/280(1.8~2.0)	

# Methodology

BGI VISTA™ Chromosome Sequencing combines high throughput sequencing technologies and short tandem repeat (STR) with biological analysis to obtain accurate information about abnormalities across all 24 chromosomes.

Next-generation sequencing enables chromosome sequencing chromosome abnormalities, including aneuploidy for all chromosomes, triploid, deletions and duplications

Product Name	For individuals with	
BGI VISTA™ Chromosome Sequencing-1M	Early abortion (less than 12 weeks with spontaneous abortion)	
	A high-risk result of a screening test (NIPS)	
DCI //CTA™ Chromosomo Coguencia da 100/	More than 12 weeks with spontaneous abortion, Stillbirth	
BGI VISTA™ Chromosome Sequencing-100K	Abnormal ultrasound, such as fetal structural abnormalities	
	Chromosome disorder, suspected for chromosomal disorders	

## **Technical limitations**

- Cannot detect chromosomes with a high rate of duplications or pyknosis (Abnormalities near centromere or telemetric regions: such as Robertsonian translocation)
- Ocannot detect polyploidy, abnormalities smaller than 100Kb or chimeras with low percentages
- Patients who have had allogeneic transfusions or organ transplants are not eligible for testing

Contact your local BGI representative for more information or email **info@bgi.com**. More information can also be found on our website. **www.bgi.com** 









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Testing services not currently available in the United States of America. Please contact a representative for regional availability.

# **TA Time:**21 days **Sample:**

- Abortion Tissue
- Core Blood (Gestation week ≥12 weeks)
- Peripheral Blood
- DNA
- Amniotic Fluid

#### Technology:

Low-coverage WGS +STR

## Workflow











