> “Understand Possible Genetic Causes of Repeated Miscarriage or Confirm Suspected Chromosomal Abnormality”

About

Chromosomal deletion or duplication syndromes result from deletion or duplication of parts of chromosomes. They may cause severe congenital anomalies and significant intellectual and physical disabilities. Vista™ Chromosome Sequencing uses NGS based low coverage whole genome sequencing (WGS) to help detect chromosome abnormalities before or during pregnancy. Vista™ Chromosome Sequencing can be performed on a variety of different sample types and results are available within **12 working days**.

To date, BGI has processed more than **12,000 samples** and detected over **4000 positive cases**.

The Power of Knowing

BGI Vista™ Chromosome Sequencing

Why Choose Vista™ Chromosome Sequencing?

The results obtained from testing can reduce a patient's emotional burden and information from the results can improve the chances of a future successful pregnancy. If the chromosome abnormality in the fetus is identified as causing the pregnancy loss, parents can usually be reassured that the chance of recurrence is low, and avoid a costly medical check-up.

Conditions Screened

Option 1: BGI VISTA™ Chromosome Sequencing-5M: deletions or duplications >5Mb

Option 2: BGI VISTA™ Chromosome Sequencing-5M: deletions or duplications >100Kb

Who should consider Vista™ Chromosome Sequencing?

- Individuals or couples who have suffered from repeated miscarriage or who have had a child who suffers from a genetic condition

- Individuals or couples who have had abnormal ultrasonography results, but with negative CGH, SNP results

- Anyone suspected of chromosome abnormality populations or patients who want to understand if a chromosome abnormality was a factor in a miscarriage

Sample Requirements

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

TA Time: 12 working days (chromosome sequencing)

Sample: Abortion Tissue, Core Blood (Gestation week ≥12 weeks), Peripheral Blood, DNA and Amniotic Fluid

Technology: Low coverage WGS

Advantages: High throughput, High resolution, Accurate

Methodology

Chromosome sequencing combines high throughput sequencing technologies with biological analysis to obtain accurate information about abnormalities across all 24 chromosomes.

Next generation sequencing enables chromosome sequencing to detect aneuploidy, deletions or duplications larger than 100kb.

Technical limitations

1. Cannot detect chromosomes with high rate of duplications or pyknosis (Abnormalities near centromere or telemetric regions: such as Robertsonian translocation)

2. Cannot detect polyploidy, abnormalities smaller than 100Kb or chimeras with low percentages

3. Patients who have had allogeneic transfusions or organ transplants are not eligible for testing

Chromosomal regions that cannot be sequenced

Chromosome	Area	Chromosome	Area
Chr1	p11.2-q12,p35.3,p36.3,q21.1,q21.2,q32.1,q44	Chr13	p-q11,q31.1,q34
Chr2	p11.1-q11.1,p11.2,p25.2,q12.3-q13,q23.1	Chr14	p-q11.1
Chr3	p11.1-q11.1,p14.1	Chr15	p-q11.1,q11.2
Chr4	p11-q11,q35.2	Chr16	p11.1-q11.2,p11.2
Chr5	p11.1-q11.1	Chr17	p11.1-q11.1, p11.2, p13.3
Chr6	p11.1-q11.1,q16.1,q27	Chr18	p11.1-q11.1,q21.2
Chr7	p11.1-q11.1,q32.2,q36.2	Chr19	p11-q11
Chr8	p11.1-q11.1,q21.2, q24.3	Chr20	p11.1-q11.2
Chr9	p11.1-q12,p12,p11.2,q13,q21.1,q22.2,q34.1	Chr21	p-q11.2
Chr10	p11.1-q11.2,q11.2,q26.2	Chr22	p-q11.2
Chr11	p11.1-q11,q21	ChrX	p11.1-q11.1
Chr12	p11.1-q11,q24.3	ChrY	p11.2-q11.1,q12

Workflow

1. 

Conduct pre-test genetic counseling with patient and sign consent form

2. 

Take sample from patient according to their different health condition and send it to BGI

3. 

Sequencing takes place at BGI laboratory

4. 

Receive test results 12 working days later

5. 

Conduct post-test genetic counseling with patient

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

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