An accurate and affordable pre-pregnancy screening test for 150+ genetic disorders

About

Most people do not know they are a carrier for an inherited genetic disease until they have a child with the disease. The BGI VISTA™ Carrier Screen detects over **9,000** mutations across 148 genes, for more than 150 genetic disorders and offers one of the most comprehensive, accurate and affordable pre-pregnancy screening tests on the market.

Why Choose BGI VISTA™ Carrier Screening?

BGI VISTA™ Carrier Screening can be ordered before or during pregnancy and is ideally suited for couples who want to learn about their genetic status, so that they can make more informed reproductive decisions.

Conditions Screened

Option 1: BGI VISTA™ Carrier Screening Targeted Panel 2.0 (screening of 156 conditions)

Option 2: BGI VISTA™ Carrier Screening Targeted Panel (screening of 100 conditions)

Option 3: BGI VISTA™ Carrier Screening mini Panel (screening of 11 conditions)

BGI VISTA™ Carrier Screening covers most common disease such as:

• Duchenne Muscular Dystrophy – the most common form of muscular dystrophy affecting children.

• Wilson disease – Worldwide 1 in 30,000 people have Wilson disease.

• Cystic fibrosis – one of the most common deadly inherited disorder among Caucasians.

• Spinal Muscular Atrophy – 1 in 50 people found to be a carrier of this disease.

• Glycogen Storage Disease– The most comprehensive coverage for all types of pomp disease in the market.

Who should consider BGI VISTA™ Carrier Screening?

- Individuals or couples who want to know more about their genetic status in order to make more informed reproductive decisions
- Individuals or couples receiving donor sperm or eggs and who want to select a donor that doesn't carry the same mutation as the member of the couple who will provide the gametes
- People with a family history of a genetic disease or from an ethnic background known to be at risk for certain genetic diseases and who are therefore at higher risk of being carriers for those diseases
- Couples who are already pregnant and who wish to know more about the genetic health of their pregnancy

The Power of Knowing
BGI VISTA™ Carrier Screening

Sample Requirements

SAMPLE TYPE	QUALITY	REQUIREMENT	SHIPMENT
Saliva	>2mL	Refer to the specific Saliva instructor	Shipped at room temperature in 7 days
Peripheral blood	5mL	Genetic invert the EDTA tube to avoid hemolysis	Stored at -20°C for short term, -80°C for long term; Shipped with dry ice. Please avoid vibrations or shock
DNA	≥3μg	Concentration≥30ng/μL OD260/280(1.8~2.0)	

Suitable for: individuals or couples planning a pregnancy
TA Time: 21 days
Sample: Saliva or peripheral blood
Technology: NGS sequencing
Advantages:
- Safe: test from just peripheral blood or saliva
- Cost effective: screen for multiple diseases with high accuracy
- Flexible: comprehensive or targeted panel options available

Methodology
Next Generation Sequencing (NGS) technology is used to analyze exons in different genes, as well as selected intergenic and intronic regions. These regions are sequenced with high coverage and compared to the normal variation standards and reference database. Vista detects more than 9,000 genetic variants covering more than 150 diseases of clinical importance selected from the HGMD database and ClinVar database.
Detected variations include: single point mutations, small Indel (within 20bp). Large duplications and deletions, balanced translocations, inversions, ploidy changes, uniparental disomy and methylation alterations cannot be detected. Detected mutations are validated with Sanger sequencing. Results are then returned to the ordering healthcare provider who will help interpret the results and provide any follow up genetic counseling or risk management plans.
Only variants classified as "pathogenic" or "likely pathogenic" will be reported, Variants of benign, likely benign or variants of uncertain significance are not reported. Variants are interpreted according to ACMG guidelines.

BGI VISTA™ Carrier Screening Validation
Using next-generation sequencing (NGS), BGI sequenced 379 samples for 148 recessive and X-linked genes involved in severe childhood phenotypes. Known variants identified by the NGS is validated by gold standard methods such as Sanger, MLPA, QPCR and gap-PCR resulting in a sensitivities and specificity more than 99.9%.

	Gold standard methods (Sanger/MLPA/QPCR/gap-PCR)	
NGS test	Positive	Negative
Positive	320	0
Negative	0	59
Sensitivity	>99.9%	
Specificity	>99.9%	

Workflow

1. Conduct pre-test genetic counseling with patient and sign consent form
2. Take sample from patient and send it to BGI
3. Sequencing takes place at BGI laboratory
4. Receive test results 21 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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Testing services not currently available in the United States of America. Please contact a representative for regional availability.