



BGI-XOME Clinical Whole Genome Sequencing (cWGS)

Background Introduction

The human genome contains 3 billion base pairs and over 20,000 genes. Whole genome sequencing (WGS) technology enables sequencing of the entire DNA sequence of the genome, providing comprehensive genomic information with advantages such as broad coverage and high uniformity. WGS can detect both coding and non-coding regions of nuclear genes (including exons, introns, promoter regions, and intergenic areas), as well as mitochondrial gene variations. It identifies various types of mutations, including point mutations, small insertions and deletions, copy number variations, and structural variations. Compared to whole exome sequencing, WGS offers higher diagnostic accuracy for clinical genetic disorder testing and is currently an efficient solution for uncovering the genetic causes of diseases.

Product Description

The product utilizes genomic DNA from the test subject's blood or other tissue, which is fragmented and prepared for whole genome sequencing and variation detection via a high-throughput sequencing platform. It allows for the simultaneous analysis of over 20,000 human genes, including more than 4,800 known disease-causing genes from the OMIM database and over 6,000 single-gene disorders, with a focus on genes relevant to the proband's phenotype. Detectable variations include point mutations (SNVs), small insertions/deletions (InDels), exon-level copy number variations (ExonCNVs), large copy number variations (Large CNVs), mitochondrial gene variations, as well as structural variations (SVs) such as loss of heterozygosity (LOH), translocations, inversions, and certain dynamic mutations like short tandem repeats (STRs).

Product Advantages



Comprehensive Detection

The BGI-XOME cWGS product can detect point mutations and small insertions/deletions in both coding and non-coding regions of nuclear genes (including exons, introns, promoter regions, and intergenic regions), exon-level copy number variations, large-scale copy number variations in the genome, and mitochondrial variations. It can also identify loss of heterozygosity (LOH), translocations, inversions, and some dynamic mutations.



Time-efficient and effective

A single clinical WGS test provides results nearly equivalent to a combination of clinical whole exome sequencing (WES), whole mitochondrial genome sequencing, and CNVSeq, reducing the time for disease diagnosis and lowering overall medical costs.



Standardized process

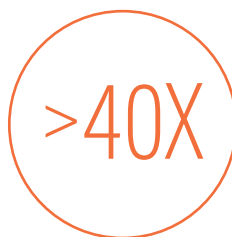
The sequencing, interpretation, and reporting steps follow highly standardized operating procedures and management protocols.



Professional team

Our professional team have published over 200 papers on monogenic diseases in international journals and offer expert data analysis, interpretation and genetic counseling services.

Sequencing Parameters



Deduplicated
Sequencing Depth



20X
Coverage



200X Mitochondrial
coverage

with mitochondrial
copy number ≥ 50



Test Includes

- SNV / Indel
- Exon CNV
- Chromosome level CNV
- Mitochondrial genome
- Deep Intron
- Structure variation
- Loss of heterozygosity
- Dynamic mutation

Testing Options

BGI Clinical Whole Genome Testing offers two options:



Individual Whole Genome Testing:

Whole genome sequencing is performed on the proband alone. Based on the clinical information and phenotype provided by the proband, high-throughput whole genome analysis and interpretation are conducted, and an individual whole genome testing report is generated.



Family Whole Genome Testing (Trio):

Whole genome sequencing is conducted on the proband and his/her parents. Data analysis and interpretation are based on the clinical information and phenotype of the proband, and a family whole genome testing report is issued, indicating the presence of the proband's positive variants in the family members.

Note: For Whole Genome Trio testing, samples from both the proband and their parents must be submitted simultaneously.

Purpose of Testing

Diagnostic Support:

The test helps identify genetic causes for patients suspected of having a genetic disorder, especially in cases of undiagnosed complex diseases with suspected genetic origins, providing valuable support for clinical diagnosis.

Reproductive Guidance:

Once the genetic cause of a disorder is identified, the test can offer couples informed reproductive advice.

Sample Requirements

- 5 mL of peripheral blood from an adult
- $\geq 3\mu\text{g}$ (30ng/ μL) of genomic DNA

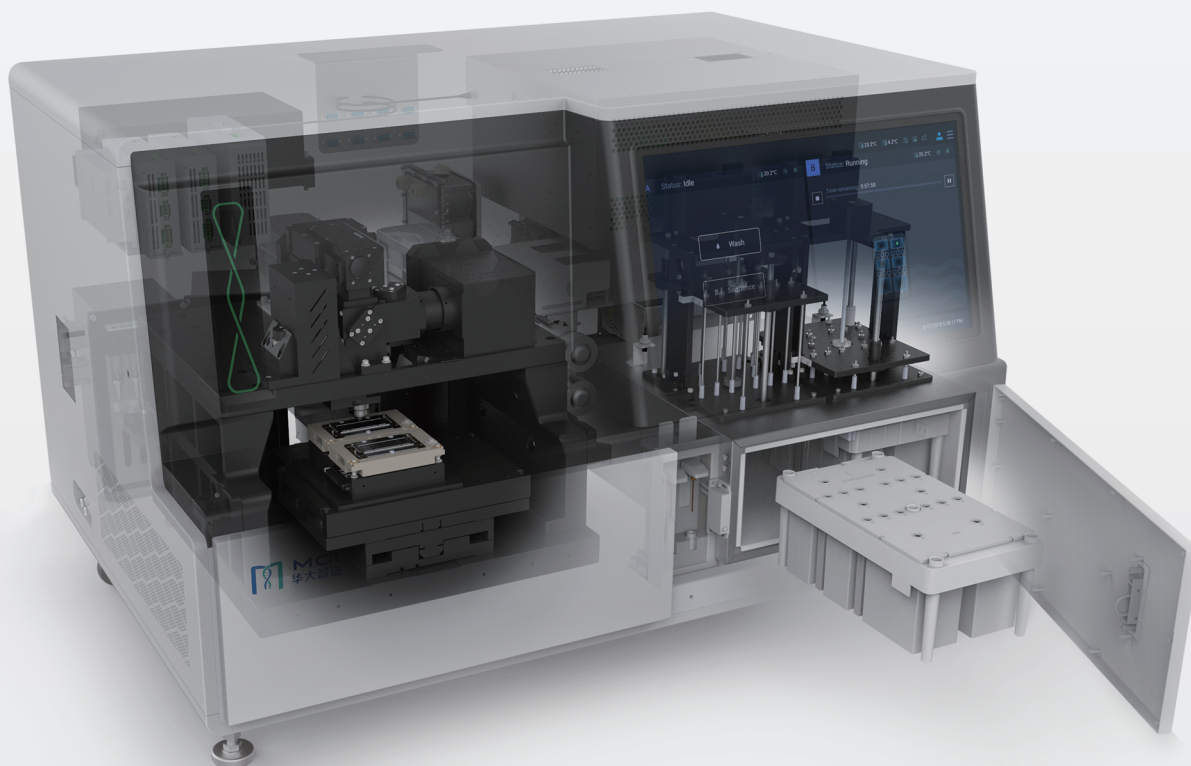
Note: For cWGS Trio testing, samples from both the proband and their parents must be submitted simultaneously.

Testing Turnaround Time (TAT)



≤ 30 days

TAT is calculated from the Hong Kong center sample acceptance to the Shenzhen laboratory report release.



DNBSEQ-G400 Sequencer

Suitable Population

Patients with Monogenic Genetic Disorders:

- Individuals diagnosed with a monogenic genetic disorder (Proband only or Trio),
- People with a family history of monogenic genetic disorders (Trio testing is recommended).

Clinically Diagnosed Fetuses with Malformations:

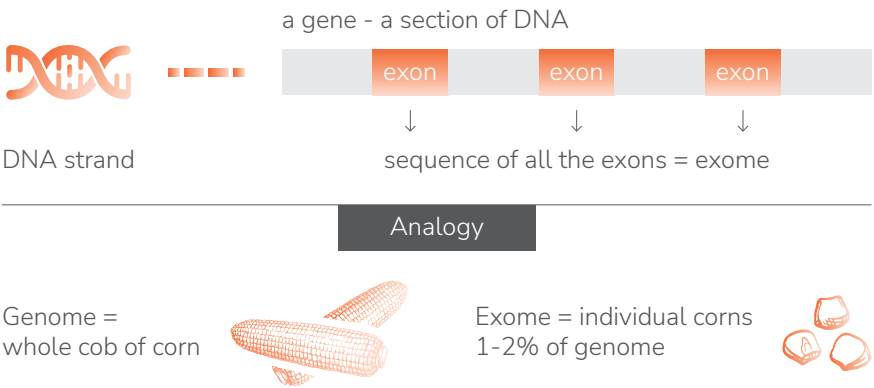
- Families with clinically diagnosed malformed fetuses (with negative chromosomal test results or no chromosomal testing) or those with a history of having a malformed fetus and similar phenotypes in a current pregnancy.

Recommended Conditions for Testing:

- Congenital multi-system abnormalities
- Autism spectrum disorders
- Neurodevelopmental disorders
- Growth and developmental delays
- Intellectual disability
- Epilepsy
- Critically ill newborns
- Complex diseases requiring differential diagnosis

Technical Limitations

This detection may not cover all the possible pathogenic mutations in a given gene.



Workflow

- 1 Genetic Counselling
- 2 Sample Collection
- 3 Shipment
- 4 DNA Extraction and QC
- 5 Sequencing
- 6 Data Analysis
- 7 Report Delivery
- 8 Translation and Counselling

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