cWES BGI-XOME

Introduction

Exon genomes of **more than 20,000 genes** in the human genome can be detected at one time, divided into individual and family Trio of the proband (proband and their parents).

The **OMIM** database was mainly used to interpret nearly **4,000 genes** and **more than 5,000 genetic and pathogenic** diseases (ACMG recommendations unrelated to the chief complaint could be made by informed choice) (59 reported genes), which can quickly find pathogenic mutations of single-gene genetic diseases and microdeletions and microduplications of more than 1M on chromosomes (note: *Y chromosome exception;** CNV still belongs to High-throughput sequencing technology is limited in scope, so analysis results that have not been verified by QPCR are presented in additional reports), and to understand the carrying status of some disease genes.

The Power of Knowing

BGI XOME Clinical Whole Exome Sequencing (cWES)

Why Choose BGI XOME cWES?

BGI has vast experience in exome sequencing and rare disease diagnosis, including exome capturing & sequencing, bioinformatics, variants annotation, and interpretation capabilities.

239,000+
WES samples tested

26,000+
Clinical rare disease samples

80+
Genes discovered by BGI

200+
Papers on rare disease diagnosis

BGI's Experience in Clinical Rare Disease Diagnoseis (based on BGI Internal data as of 2020).

Technical limitations

Whole Exome Sequencing Testing is suitable for clinical laboratories. This detection may not cover all possible pathogenic variants in the given genes. The above results are for clinical reference purposes only. For all suspected pathogenic mutations, please validate the results with SANGER sequencing. If you have any queries, please contact your genetic consultant.

Who is suitable for BGI XOME cWES?

WES is intended for use in conjunction with the clinical presentation and other markers of disease progression for the management of patients with rare genetic disorders and especially heterogeneous phenotypes.

Advantages

Thorough analysis

The exons of 20,000 genes were detected at one time, and more than 5,000 single-gene diseases were analyzed and interpreted, as well as 68 types of micro-deletion and micro duplication syndrome (supplementary analysis content). The samples of the proband and the parents of the proband in the all-foreign Trio project were all subjected to all-foreign sequencing and association analysis

Rich experience

Has completed the sequencing of more than 20,000 cases of whole exons, published 205 papers related to a single disease in international journals, accumulated rich experience

Process specification

Sequencing, interpretation, reporting, and other links, with highly standardized operational procedures and management specifications

Team professional

Powerful information analysis, interpretation, and genetic consulting team, to provide you professional services

Sample Requirements

Saliva, Peripheral blood, Blood card, Genomic DNA

Technology

Next Generation Sequencing (NGS)

TAT

25 days

Workflow

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

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Published January 2022.