Introduction Brochure

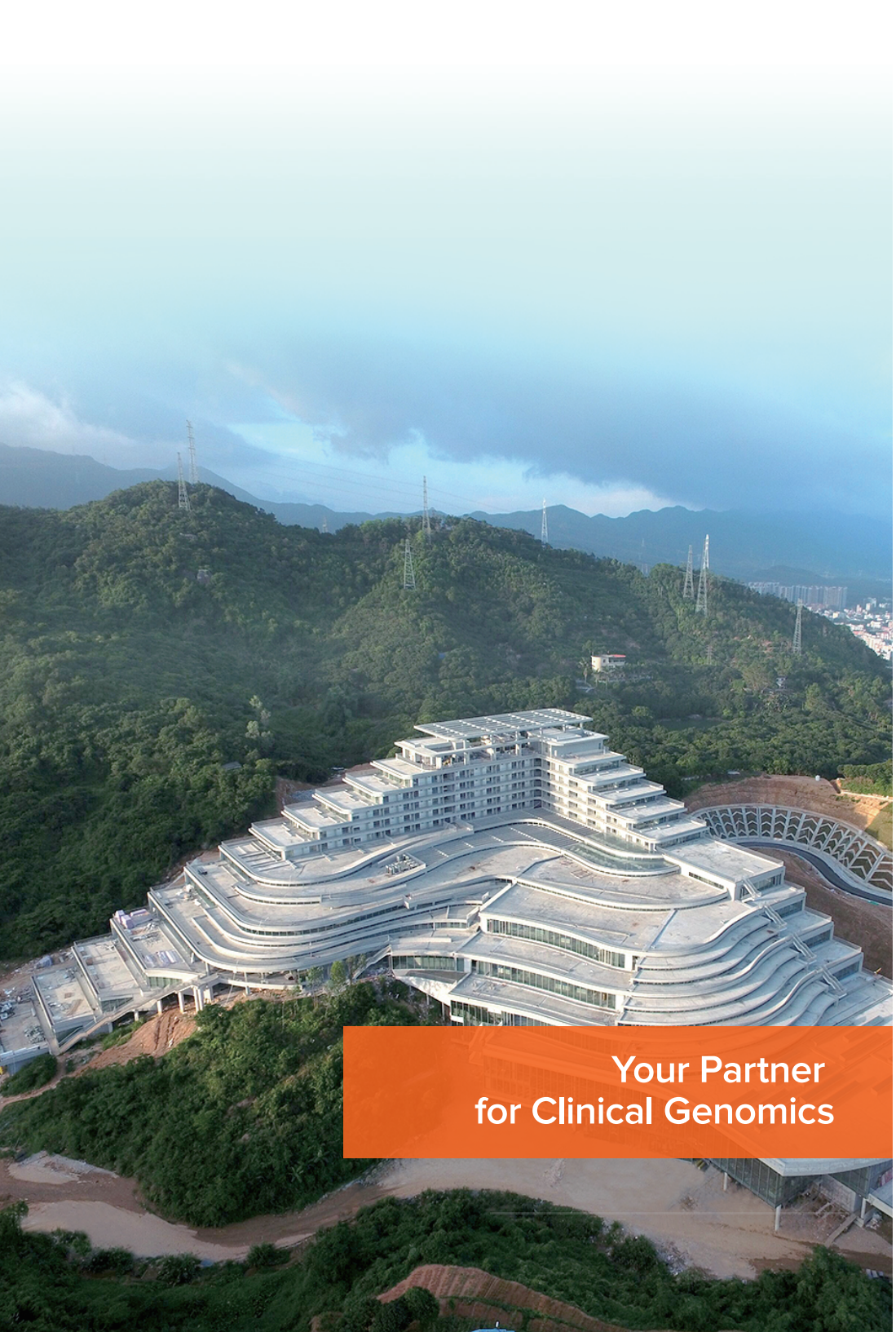
BGI-XOME

CLINICAL WHOLE EXOME SEQUENCING

CLIA ID 99D2135851

Contact us

- TEL: 086-755-2235 4767
- Web: www.bgi.com
- Email: info@bgi.com
- HQ: Building NO.11, Beishan Industry, Futian District, Shenzhen, Gunagzhou, China. 518083



Your Partner
for Clinical Genomics

BGI

Clinical Whole Exome Sequencing

Exon is a region of genomic DNA that directly participates protein translation. The gather of these DNA regions is called exome. Most mutations in DNA level of known diseases(around 85%) are found in exon regions. Conducting genetic test for analyzing human DNA is the most efficient way to reveal the mechanism of hereditary diseases. Whole exome sequencing cannot only assist for determining the pathogenesis but also detect for potential health risk, providing reasonable life guidance. Also, it can conduct carrier screening test for susceptive crow, giving trusty birth guidance.

BGI Clinical whole exome sequencing (WES), utilizes the leading detection technique with the thoughts of state of art among the international genetic area. Oligonucleotide probe is firstly used to capture the exon targeted region, combining high-throughput sequencing and bioinformatic analysis to conduct interpretation for gene loci of hereditary cancer, neuromuscular disease, metabolic disorders, eye-ear-nose disorders, dysplasia, cognitive function and intellectual disability, etc. Personalized healthy life guidance will then be designed according to the test result, so that physicians can know more about individual characteristics, preventing disease generation and providing with advice for overall service of personalized medicine.

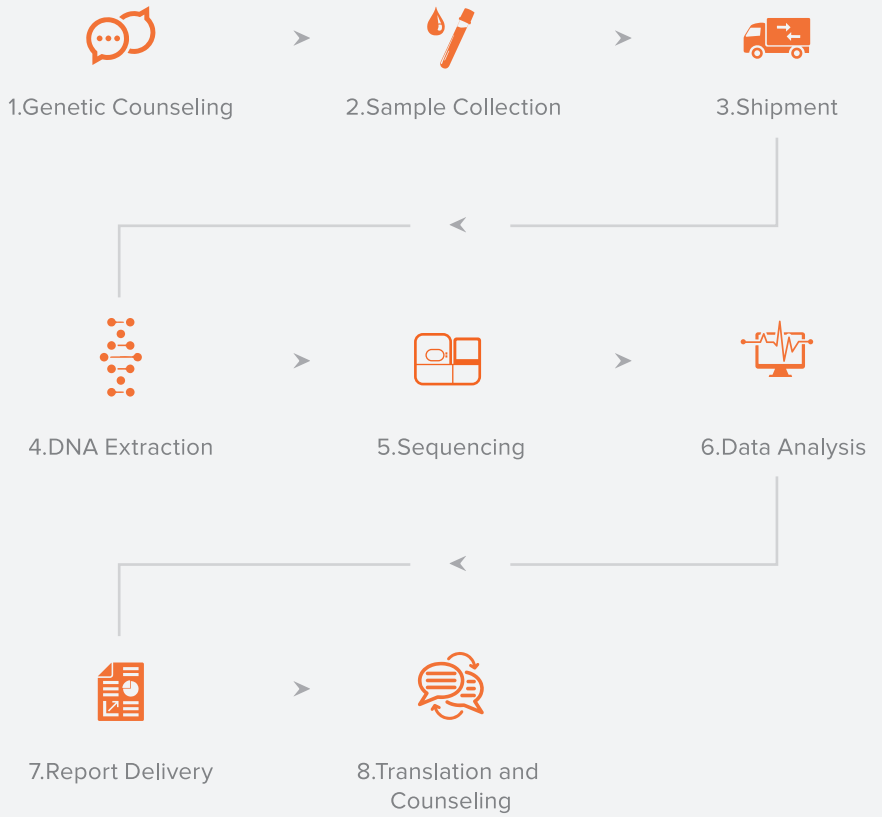
Who is suitable for WES?

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.

- Neuromuscular 853
- Skin 251
- Intellectually Disabilities 158
- Reproductive system 70
- Metabolic function 490
- Digestive System 120
- Caner and tumor 118
- Hearing impairment 122
- Diabetes 90
- Eyes 354
- Immune and infectious diseases 9
- Respiratory System 490
- Circulatory system 427
- Endocrine system 161
- Genetic Skeletal disorder 306
- Urinary system 122

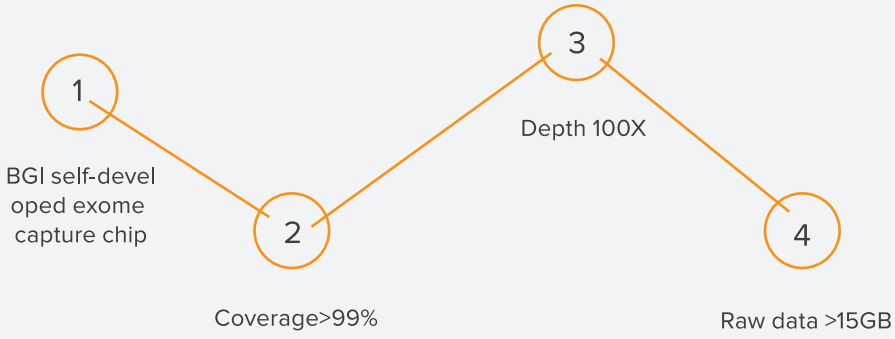
How does BGI WES process

BGI-XOME Whole Exome Sequencing provides you with a comprehensive clinical report, which your complex DNA sequencing data is transferred into.



Whole exome sequencing in BGI

- Turnaround time of less than 40 workdays
- Sample type as peripheral blood, DNA, saliva
- Statistic Data



What does our report involve:

- Sample information
- Test information
- Test result with clear variant information according to ACMG includes gene, reference sequence, nucleic acid alteration and amino acid alteration, mutation location, chromosomal location, reference, zygosity and mutation type.
- Comprehensive clinical Interpretation with specific diagnostic recommendation and explanation of the suspected disease.
- References to publications supporting the medical and scientific results
- Detailed method information and parameters
- Figure(s) related to the result, (if applicable), and list of variant(s) with low frequencies identified on OMIM known disease genes

Also Service for your family:

BGI Clinical Whole Exome Sequencing trio, to fully know about the genetic information of both you and your family.

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