The power of Knowing

BGI XOME™ Targeted Monogenic Disease Testing

About

Monogenic disease is also called Mendelian disease, which results from the modifications in a single gene. They are inherited according to Mendel's Laws; the mutation can be inherited from parents or spontaneous where there is no previous family history and in both cases can be passed to the next generation. there are more than 7000 different types of disease found to be monogenic disease and overall morbidity shows 10/1000 worldwide according to the WHO. BGI's Targeted Monogenic Disease Testing offers a wide range of gene panels covering more than 2000 genes and 1500 monogenic diseases

Who should consider the BGI XOME™ Targeted Monogenic Disease Test?

- Intended for use in conjunction with the clinical presentation and other markers of disease progression for the management of patients with rare genetic disorders.
- Patient who has family history of monogenic disease

TAT: 15-35 working days

Technology: Target region capture based NGS

Sample: Peripheral blood, DNA

Highlights

- Targeted sequencing allows the researcher to focus on one or more **specific regions** and to sequence that target to a high level of coverage without generating significant quantities of non-target data. This increases the chance of finding biologically relevant variants.
- BGI provides panels organized by **10 different body systems** and also a wide range of **smaller gene panels** selected for specific monogenic diseases.
- As of 2017, BGI has tested around **10,000** samples for thalassemia, **1,100,000** samples for hearing loss, as well as a large volume of samples for Hereditary Muscular Disease, Cystic Fibrosis, Neurofibromatosis, Hypertrophic Cardiomyopathy, Inherited Metabolic disorders, Retinitis Pigmentosa, Polycystic Kidney Disease and Marfan Syndrome.

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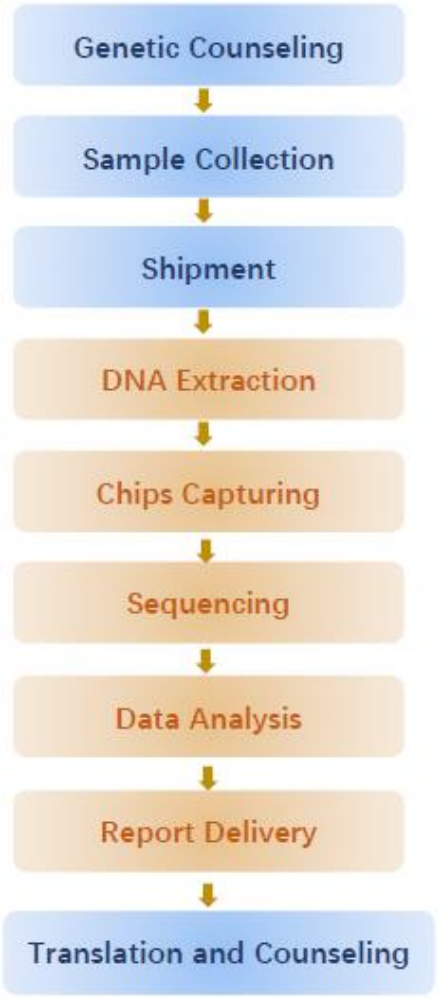
Condition Screened

Over 400 kinds of diseases in 10 human body system and over 1600 specific diseases in panel of 1445 project. Click xome.bgi.com for disease and panel searching.

System	Products
Neuromuscular system	Skeletal neuromuscular genetic disease, heredopathia of nervous, system(older), dysgnosia autism spectrum disorder, inherited oculopathy
Skeletal system	skeletal neuromuscular genetic inherited osteopathy;
Respiratory system	Respiratory diseases(1445 genetic testing)
Digestive system	Monogenic diseases in digestive system
Endocrine system	Inherited metabolic diseases, diabetes(monogenic), sexual development disorders
Urinary system	inherited nephritis
Immune system	Monogenic diseases in immune system
Circulatory system	Hematopathy,inherited arrhythmias,angiocardiopathy(in research)
Skin	Monogenic diseases on skin
Multi-system and other	Mitochondrial diseases,**1445 genetic testing**

Workflow



Sample Requirements

Type	Shipment/delivery
Peripheral blood	Amount for adult is ≧5ml, ≧2ml for Neonate/Child. EDTA or anticoagulation to avoid blood coagulation, proceed shipping directly after sampling with dry ice/ice bag.
DNA	Direct shipping after DNA extraction with dry ice; DNA has to meet the quality of Concentration≧30ng/μl; Quantity ≧ 3μg; OD 260/280 value should be between 1.8-2.0, without degradation, protein or RNA
Saliva	**2ml, sample as instructed**

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