Helping parents to detect genetic risks, provide reproductive guidance, and prevent birth defects.

Specimen Requirement

2-5 mL peripheral blood

Target Population

All couples who are planning a pregnancy or during early pregnancy, including those:

- With normal phenotypes and no family history of monogenic diseases.

- Who wants to have a healthy baby using assisted reproductive technology (ART).

- Who are close blood relatives.

Detection Time

- Before or during early pregnancy.

- In early pregnancy, it is recommended that both parents be detected.

- Reasonable disease selection
- Large database of mutations following ACMG guidelines
- Extensive experience in genetic disease screening and research
- Complete lab qualifications and quality certifications
- Genetic counseling expert team and online search tools

Technology

Target area capture - high-throughput sequencing technology

Characteristics

Rigorous: Inclusion of **prenatal and early pregnancy diseases with clear pathogenic genes** and **severe and early onset diseases**, following requirements of international guidelines.

Comprehensive: Detection of exon regions, splicing regions, some intron regions, promoter regions and UTR regions containing **1200+ target genes**.

Accurate: Applying strict performance evaluation and quality control standards; pathogenicity of sites determined in strict accordance with **ACMG** guidelines.

Highly efficient: Screening of **1200+ autosomal recessive and X-linked single-gene diseases in 1 test**, improving the comprehensive prevention and control of birth defects.

Testing Content

Carrier Screening screens for more than 1200 genes associated with 1200+ recessive monogenic diseases.

Classification	Diseases
Inherited Metabolic Disorders	Wilson Disease; Primary Carnitine Deficiency; Phenylketonuria; Hyperphenylalaninemia, BH4-deficient, A; MUT-Related Methylmalonic Acidemia; Methylmalonic Aciduria and Homocystinuria cblC type, 2,4-Dienoyl-CoA Reductase Deficiency, 2-Methylbutyryl Glycinuria, 3-beta-Hydroxysteroid Dehydrogenase Deficiency, etc.
Neuromuscular Skeletal Related Disorders	Duchenne Muscular Dystrophy; Spinal Muscular Atrophy; Joubert Syndrome 2; Joubert Syndrome 3, Ullrich congenital muscular dystrophy 1, Spinal muscular atrophy, X-linked 2, infantile, etc.
Cutaneous Disorders	Oculocutaneous Albinism Type 1; Autosomal Recessive Congenital Ichthyosis 1; Autosomal Recessive Congenital Ichthyosis 4A; Autosomal Recessive Congenital Ichthyosis 4B; Sjögren-Larsson syndrome, X-Linked Dyskeratosis Congenita, Epidermolytic hyperkeratosis, etc.
Circulatory System Disorders	Hemophilia B; Alpha-thalassemia; Beta-thalassemia; Sickle Cell Anemia, Methemoglobinemia Due to Deficiency of Methemoglobin Reductase, Thiamine-Responsive Megaloblastic Anemia Syndrome, etc.
Endocrine Issues and Autoimmune Disorders	Hemophagocytic lymphohistiocytosis, familial, Severe combined immunodeficiency, B cell-negative; X-Linked Severe Combined Immunodeficiency, Congenital Adrenal Hyperplasia due to 11-beta-Hydroxylase-Deficiency, DCLRE1C-Related Severe Combined Immunodeficiency, Common Variable Immune Deficiency 8 with Autoimmunity, etc.
Gastrointestinal and Urinary System Disorders	Progressive Familial Intrahepatic Cholestasis 2; Alport syndrome 2, autosomal recessive; Nephrotic syndrome, type 1, Polycystic Kidney Disease 4 with or without Polycystic Liver Disease, etc.
The Respiratory and Otorhinolaryngology System Disorders	Cystic Fibrosis; Autosomal Recessive Deafness 1A; Autosomal Recessive Deafness 4, with Enlarged Vestibular Aqueduct, Congenital Cataracts, Hearing Loss, And Neurodegeneration, etc.
Complex Multisystem Disorders	Meckel Syndrome 2, X-Linked Hypohidrotic Ectodermal Dysplasia; COACH syndrome, etc.