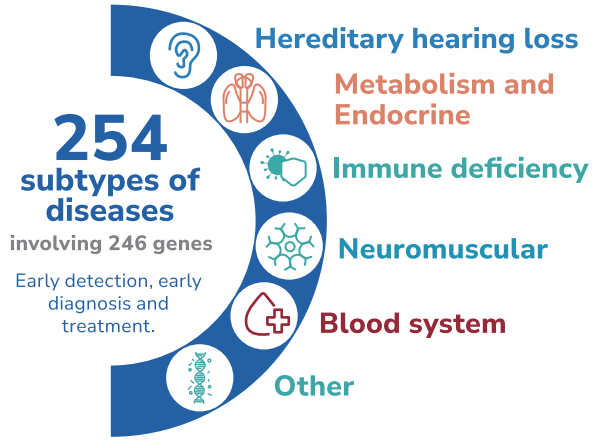
# NOVA™ Newborn Genetic Screening

An efficient genetic disease screening panel for your baby. **One-stop** screening for 254 common diseases such as genetic metabolic diseases, deafness and thalassemia **is provided** for all newborns and children.

## Who should consider the NOVA™ Newborn Genetic Test?

- All newborns (All newborns can be tested immediately after birth, without waiting for 72h/6 lactation)




**Sample types:** 3 Dried Blood Spots (8mm in diameter) or Blood (≥1mL)

**Testing scope:** There are more than 10,000 pathogenic/suspected pathogenic mutation sites in the database of clinical gene detection of neonatal diseases, including SNP, CNV and InDel (≤ 20 bp), and deletion/repetition of some gene exons.

**Testing method:** Chip capture high-throughput sequencing

**Technical Specifications:** The effective sequencing depth of genome is ≥100X, the effective sequencing depth of mitochondria is ≥300X, and the coverage of the target area 20X is >95%

### Scientificness
Screening and inclusion of diseases according to authoritative global guidelines.

### One-stop testing
It can screen the risks of a variety of common genetic diseases, including genetic metabolic diseases, hearing loss, thalassemia, Duchenne muscular dystrophy, spinal muscular atrophy, etc.

### Efficient
The coding region of the target gene was completely sequenced, and various variants including SNP and CNV could be analyzed by a single detection method.

### Accurate interpretation
According to ACMG guidelines, more than 10,000 pathogenic and suspected pathogenic mutation sites have been accurately interpreted.

## Testing Workflow:

Informed consent → Sample collection → Sequencing and analysis → Report → Genetic counseling

**Benefits:**

**Assistant diagnosis:** Screening and diagnosis of potential patients before symptoms appear or in the early stage of the disease, and timely intervention to avoid or reduce the impact of the disease on sick children and their families.

**Fertility guidance:** Genetic screening combined with genetic counseling can evaluate the risk of disease recurrence, and provide scientific guidance for couples to reproduce.

## Disease List:

- Hyperphenylalaninemia (6 Genes)
- Carbamoylphosphate Synthetase Deficiency (1 Gene)
- Maple Syrup Urine Disease (4 Genes)
- Glycine Encephalopathy (3 Genes)
- Hypermethioninemia (4 Genes)
- Hyperornithinemia-hyperammonemia-homocitrullinuria Syndrome (1 Gene)
- Hyperprolinemia (2 Genes)
- Homocystinuria (4 Genes)
- Citrullinemia (2 Genes)
- Argininosuccinic Aciduria (1 Gene)
- Argininemia (1 Gene)
- Tyrosinemia Type (3 Genes)
- Ornithine Transcarbamylase Deficiency (1 Gene)
- N-acetylglutamate Synthase Deficiency (1 Gene)
- Hypervalinemia (2 Genes)
- Histidinemia (1 Gene)
- 2-methylbutyryl Glycinuria (1 Gene)
- 3-methylcrotonyl-coa Carboxylase Deficiency (2 Genes)
- 3-methylglutaconic Aciduria (6 Genes)
- 3-hydroxy-3-methylglutaryl-coa Lyase Deficiency (1 Gene)
- 3-hydroxy-3-methylglutaryl-coa Synthase 2 Deficiency (1 Gene)
- Beta-ketothiolase Deficiency (1 Gene)
- Malonyl-coa Decarboxylase Deficiency (1 Gene)
- Propionicacidemia (2 Genes)
- Succinic Semialdehyde Dehydrogenase Deficiency (1 Gene)
- Methylmalonic Acidemia (14 Genes)
- Multiple Carboxylase Deficiency (1 Gene)
- Biotinidase Deficiency (1 Gene)
- Glutaric Acidemia I (1 Gene)
- 2-methyl-3-hydroxybutyryl-coa Dehydrogenase Deficiency (1 Gene)
- Isobutyryl-coa Dehydrogenase Deficiency (1 Gene)
- Isovaleric Acidemia (1 Gene)
- 2,4-dienoyl-coa Reductase Deficiency (1 Gene)
- Short Chain Acyl-coa Dehydrogenase Deficiency (1 Gene)
- Glutaric Acidemia Ii (3 Genes)
- Acyl-coa Dehydrogenase Deficiency,very Long-chain (1 Gene)
- Carnitine-acylcarnitine Translocase Deficiency (1 Gene)
- Carnitine Palmitoyltransferase Deficiency (2 Genes)
- Trifunctional Protein Deficiency (2 Genes)
- Primary Carnitine Deficiency (1 Gene)
- Long-chain 3-hydroxyacyl-coa Dehydrogenase Deficiency (1 Gene)
- Medium-chain Acyl-coenzyme A Dehydrogenase Deficiency (1 Gene)
- Medium Chain 3-ketoacyl-coa Thiolase Deficiency (2 Genes)
- 3-hydroxyacyl-coa Dehydrogenase Deficiency (1 Gene)
- Ethylmalonic Encephalopathy (1 Gene)
- Krabbe Disease (1 Gene)
- Fabry Disease (1 Gene)
- Niemann-pick Disease (3 Genes)
- Mucopolysaccharidisis (10 Genes)
- Metachromatic Leukodystrophy (1 Gene)
- Gangliosidosis (6 Genes)
- Galactosemia (3 Genes)
- Glycogen Storage Disease (12 Genes)
- Hereditary Fructose Intolerance (1 Gene)
- Peroxisome Biogenesis Disorder (14 Genes)
- Primary Hyperoxaluria Type (3 Genes)
- Glutaric Aciduria Iii (1 Gene)
- Sitosterolemia (2 Genes)
- Hypercholesterolemia, Familial (1 Gene)
- Cerebrotendinous Xanthomatosis (1 Gene)
- Menkes Disease (1 Gene)
- Hypophosphatemia (1 Gene)
- Wilson Disease (1 Gene)
- Progressive Familial Intrahepatic Cholestasis (3 Genes)
- Glucose-6-phosphate Dehydrogenase Deficiency (1 Gene)
- Congenital Bile Acid Synthesis Defect (1 Gene)
- Hypophosphatasia (1 Gene)
- Dihydrolipoamide Dehydrogenase Deficiency (1 Gene)
- Cerebral Creatine Deficiency Syndrome (1 Gene)
- Alagille Syndrome (2 Genes)
- Crigler-najjar Syndrome (2 Genes)
- Pendred Syndrome (1 Gene)
- Usher Syndrome (5 Genes)
- Aminoglycoside-induced Deafness (1 Gene)
- Autosomal Recessive Deafness (14 Genes)
- Autosomal Dominant Deafness (2 Genes)
- Waardenburg Syndrome (3 Genes)
- Wiskott-aldrich Syndrome (1 Gene)
- X-linked Lymphoproliferative Syndrome (2 Genes)
- X-linked Agammaglobulinemia (1 Gene)
- Chronic Granulomatous Disease (1 Gene)
- Severe Combined Immunodeficiency (6 Genes)
- Familial Mediterranean Fever (1 Gene)
- Immunodeficiency With Hyper-igm (1 Gene)
- Severe Congenital Neutropenia (1 Gene)
- Congenital Adrenal Hyperplasia (4 Genes)
- Kallmann Syndrome (4 Genes)
- Adrenal Hypoplasia Congenita (1 Gene)
- Hypothyroidism Congenital (7 Genes)
- Diabetes Mellitus, Permanent Neonatal (2 Genes)
- Familial Hyperinsulinemic Hypoglycemia (4 Genes)
- Pyridoxine-dependent Epilepsy (1 Gene)
- Hereditary Spastic Paraplegia (4 Genes)
- Autosomal Dominate Myotonia Congenita (1 Gene)
- Progressive Muscular Dystrophy (1 Gene)
- Spinal Muscular Atrophy, Type I (1 Gene)
- Dopa-responsive Dystonia (2 Genes)
- Glucose Transporter Type 1 Deficiency Syndrome (1 Gene)
- Early Infantile Epileptic Encephalopathy (2 Genes)
- Brown-vialetto-van Laere Syndrome (2 Genes)
- Diamond-blackfan Anemia (3 Genes)
- Thalassemia (3 Genes)
- Fanconi Anemia (1 Gene)
- Familial Hemophagocytic Lymphohistiocytosis (2 Genes)
- Transcobalamin Ii Deficiency (1 Gene)
- Gitelman Syndrome (1 Gene)
- Leber Hereditary Optic Neuropathy (1 Gene)
- Alport Syndrome (3 Genes)
- Tuberous Sclerosis (2 Genes)
- Cystic Fibrosis (1 Gene)
- Retinoblastoma (1 Gene)

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