Recessive Carrier Full Gene Screen

Recessive Carrier Genetic Screening Test is a comprehensive preconception and prenatal carrier screening test. It provides physicians with information about the risks of inherited diseases of their patients' future children. The Recessive Carrier Genetic Screening Test follows the American College of Obstetricians and Gynecologists (ACOG) and Amercican College of Medical Genetics (ACMG) recommendations, and using the most advance Next Generation Sequencing (NGS) technology to conduct full screening for more than 173 recessive genetic diseases.

This screening is test is for those who wish to understand their risks of passing the diseases to their children, Those with a family history of recessive disease may also benefit from the screening. It is also advisable for couples to do it together.

UNDERSTANDING CARRIER STATUS

- Each person has two copies of the genetic materials, one copy inherited from each parent.
- Many genetic diseases are recessive, meaning the disease is caused by inheriting a mutation at the same DNA location from both parents.
- If a parent carries a mutation in one of the two copies of the DNA, he/she is a carrier of the genetic disease.
- A disease carrier is unlikely to have any symptoms, however if the both parents are carriers, the child will have a 25% chance of inheriting both copies of the mutation, thus leading to the development of the genetic disease.

Common Recessive diseases at a glance

Fragile X Syndrome

Carrier rate: 1: 151 in females (general population)

Fragile X syndrome is a genetic condition that causes a range of developmental problems including learning disabilities and cognitive impairment. A condition is considered X-linked if the mutated gene that causes the disorder is located on the X chromosome. In males (who have only one X chromosome), a mutation in the only copy of a gene in each cell causes the disorder. In most cases, males experience more severe symptoms of the disorder than females.

Affected individuals usually have delayed development of speech and language by age 2. Most males with fragile X syndrome have mild to moderate intellectual disability, while about one-third of affected females are intellectually disabled. Children with fragile X syndrome may also have anxiety and hyperactive behavior such as fidgeting or impulsive actions. They may have attention deficit disorder (ADD), which includes an impaired ability to maintain attention and difficulty focusing on specific tasks. About one-third of individuals with fragile X syndrome have features of autism spectrum disorders that affect communication and social interaction. Fragile X syndrome is inherited in an X-linked dominant pattern.

Spinal Muscular Atropy

Carrier rate 1:47 – 72

Spinal muscular atrophy (SMA) is a genetic disease that results in progressive muscle weakness and paralysis. The condition occurs in 1 in 10,000 live births and affects both males and females.

There are three types of SMA. The most severe type is usually diagnosed within the first few months of life. Affected children have severe muscle weakness and typically do not survive past the age of 2.

The other two types of SMA, which are less common than the severe type, involve a lesser degree of muscle weakness. Most affected individuals need to use wheelchairs or need assistance with walking. Life expectancy for the less severe types ranges from the teenage years to adulthood. Those with the mildest form of SMA are expected to have a normal lifespan.

Pendred syndrome

Carrier rate: 1:50 (Chinese)

It is a disorder typically associated with hearing loss and a thyroid condition called a goiter. If a goiter develops in a person with Pendred syndrome, it usually forms between late childhood and early adulthood. In most people with Pendred syndrome, severe to profound hearing loss caused by changes in the inner ear (sensorineural hearing loss) is evident at birth.

Phenylketonuria (commonly known as PKU)

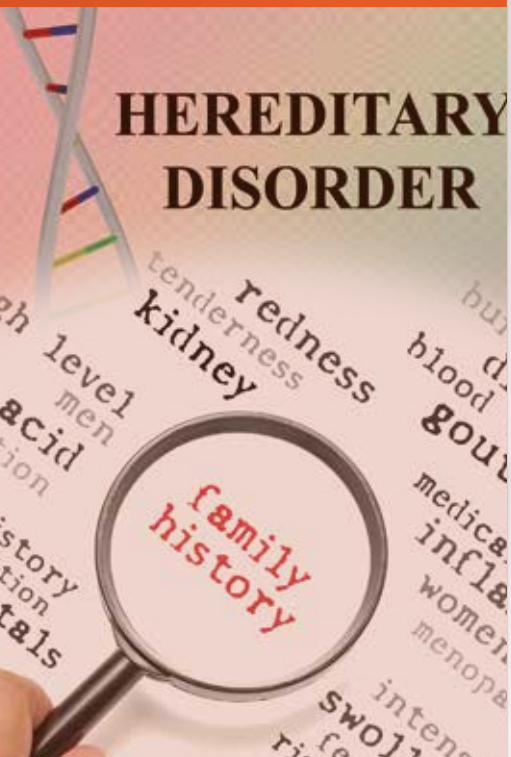
Carrier rate: 1:53 (Chinese)

It is an inherited disorder that increases the levels of a substance called phenylalanine in the blood. The signs and symptoms of PKU vary from mild to severe. The most severe form of this disorder is known as classic PKU. Infants with classic PKU appear normal until they are a few months old. Without treatment, these children develop permanent intellectual disability. Seizures, delayed development, behavioral problems, and psychiatric disorders are also common.

Smith-Lemli-Opitz syndrome

Carrier rate: 1: 68 (General population)

It is a developmental disorder that affects many parts of the body. This condition is characterized by distinctive facial features, small head size (microcephaly), intellectual disability or learning problems, and behavioral problems. Many affected children have the characteristic features of autism, a developmental condition that affects communication and social interaction. The signs and symptoms of Smith-Lemli-Opitz syndrome vary widely. Mildly affected individuals may have only minor physical abnormalities with learning and behavioral problems. Severe cases can be life-threatening and involve profound intellectual disability and major physical abnormalities.



Wilson's disease

Carrier rate: 1:90 (Asian)

Wilson disease is an inherited disorder in which excessive amounts of copper accumulate in the body, particularly in the liver, brain, and eyes. Symptoms are typically related to the brain and liver. Liver related symptoms include vomiting, weakness, fluid build up in the abdomen, swelling of the legs, yellowish skin, and itchiness. Brain related symptoms include tremors, muscle stiffness, trouble speaking, personality changes, anxiety, and seeing or hearing things that others do not. Most people with Wilson's disease are diagnosed between the ages of 5 and 35, but it can affect younger and older people, as well.

Beta -Thalassemia

Carrier rate: 1: 100 (Chinese)

Thalassemia encompasses a varied group of inherited blood disorders, including some that are relatively mild and others that may cause severe anemia and other serious problems. The signs and symptoms of thalassemia major appear within the first 2 years of life. Children develop life-threatening anemia. They do not gain weight and grow at the expected rate (failure to thrive) and may develop yellowing of the skin and whites of the eyes (jaundice).

隱性遺傳病帶病者全基因檢測項目 (>173 疾病)
4C Recessive Disease Carrier Status Full gene screening (>171 diseases)

SCAD Deficiency
17-beta-hydroxysteroid dehydrogenase X deficiency
2-methylbutyryl-CoA Dehydrogenase Deficiency
3-hydroxyacyl-CoA dehydrogenase deficiency
3-Methylcrotonyl-CoA carboxylase 1 deficiency (MCC1D)
3-Methylcrotonyl-CoA carboxylase 2 deficiency (MCC2D)
3-methylglutaconic aciduria type I (MCGA1)
3-methylglutaconic aciduria, type III
3-methylglutaconic aciduria, type V
Adrenoleukodystrophy Hb EE

Alpha-methylacetoacetic aciduria (3-ketothialase deficiency)
Argininemia (Arginase Deficiency)
Arginosuccinic Aciduria
Ault-onset citrullinemia Type II
Autosomal dominant deafness Type 3A
Autosomal dominant deafness Type IIB
Autosomal dominant deafness Type IIIB Autosomal dominant persistent hypermethioninemia due to methionine
adenosyltransferase I/III deficiency
Autosomal recessive deafness
Autosomal recessive deafness Type 1A

Autosomal recessive deafness Type IB
Autosomal recessive deafness type IV
Autosomal recessive Methionine adenosyltransferase deficiency
Barth Syndrome
Bart-Pumphrey Syndrome
Beta thalassemia major
BH4-deficient Hyperphenylalaninemia A
BH4-deficient Hyperphenylalaninemia B BH4-deficient Hyperphenylal-aninemia C
BH4-deficient Hyperphenylalaninemia D

Biotinidase deficiency
Bloom Syndrome
Canavan disease
Carnitine-acylcarnitine translocase (CACT) deficiency
cbl E complementation type homocystinuria-megaloblastic anemia
cbl G complementation type homocystinuria-megaloblastic anemia
cblB complement type Vitamin B-12 responsive methylmalonic aciduria (due to defect in synthesis of adenosylcobalamin)
cblD complement type homocystinuria (Variant 1)
cblD complement type homocystinuria (Variant 2)
cblD complement type Methylmalonic aciduria and homocystinuria

cblJ Type Methylmalonic aciduria and homocystinuria
Citrullinemia
Clouston type ectodermal dysplasia Type II Cogenital bilateral absence of the vas deferens (CVAD)
Combined malonic and methylmalonic aciduria
Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency
Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia
Congenital nongoitrous hypothyroidism 1
Congenital nongoitrous hypothyroidism 4
Congenital nongoitrous hypothyroidism 6

Hb Barts
Hb C disease (Hb CC)
Hb C/ Beta° thalassemia
Hb C/Beta^{+} thalassemia
Hb D disease (Hb DD)
Hb D/ Beta° thalassemia
Hb D/Beta^{+} thalassemia
Hb E/ Beta° thalassemia
Hb E/Beta^{+} thalassemia

Hb H (3 gene deletion)
Hb H/Constant Spring disease
Hb S/ Beta° thalassemia
Hb S/Beta^{+} thalassemia
Hb Variant/ Beta° thalassemia
Hb Variant/Beta^{+} thalassemia
Hb variants/Alpha thalassemia
Hemolytic anemia due to G6PD deficiency
Hepatic carnitine palmitoyl transferase deficiency Type I
Hepatic carnitine palmitoyl transferase deficiency Type II

Hereditary persistence of fetal hemoglobin
Hex A pseudodeficiency
HMG-CoA Lyase Deficiency
Holocarboxylase synthetase deficiency
Homocystinuria due to MTHFR deficiency
Homocystinuria, B6-responsive and nonresponsive types
Hyperhomocysteinemic thrombosis
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
Hystrix-like ichthyosis with deafness
Isobutyryl-CoA dehydrogenase deficiency

Isovaleric acidemia
Keratitis ichthyosis deafness syndrome
Krabbe disease
LCHAD deficiency
Lethal neonatal CPT2 deficiency
Malonyl-CoA decarboxylase deficiency
Maple syrup urine disease type II
Maple syrup urine disease, type Ia
Maple syrup urine disease, type Ib
MCAD Deficiency

Mental retardation X-linked syndromic 10 (MRXS10)
Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic aciduria due to Methylmalonyl-CoA Mutase deficiency
Methylmalonic aciduria due to transcobalamin receptor defect
Methylmalonyl-CoA epimerase deficiency
Mucolipidosis IV
Mucopolysaccharidosis Ih
Mucopolysaccharidosis Ih/s
Mucopolysaccharidosis Is
Neonatal hypertrypsinemia

隱性遺傳病帶病者全基因檢測項目 (>171疾病) (續...)
Appendix 4C Recessive Disease Carrier Status Full gene screening (>171 diseases) (con't)

cblJ Type Methylmalonic aciduria and homocystinuria	Mental retardation X-linked syndromic 10 (MRXS10)
Citrullinemia	Methylmalonic aciduria and homocystinuria, cblC type
Clouston type ectodermal dysplasia Type II Cogenital bilateral absence of the vas deferens (CVAD)	Methylmalonic aciduria due to Methylmalonyl-CoA Mutase deficiency
Combined malonic and methylmalonic aciduria	Methylmalonic aciduria due to transcobalamin receptor defect
Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency	Methylmalonyl-CoA epimerase deficiency
Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia	Mucolipidosis IV
Congenital nongoitrous hypothyroidism 1	Mucopolysaccharidosis Ih
Congenital nongoitrous hypothyroidism 4	Mucopolysaccharidosis Ih/s
Congenital nongoitrous hypothyroidism 6	Mucopolysaccharidosis Is
	Neonatal hypertrypsinemia
CPT2 deficiency associated myopathy	Neonatal onset citrullinemia Type II
Cystic Fibrosis	Niemann-Pick disease, type A
Digenic deafness GJB2/GJB3	Niemann-Pick disease, type B
Digenic GJB2/GJB6 deafness Dihydrolipoamide dehydrogenase deficiency DOPA-responsive dystonia (with or without hyperphenylalaninemia)	Niemann-Pick disease, type C1
	Niemann-pick disease, type C2
Erythrokeratodermia variabilis et progressiva	Niemann-Pick disease, type D
Fabry disease	Nonautoimmune hyperthyroidism
Familial dilated cardiomyopathy Familial dysautonomia	Non-classic hyperandrogenism due to 21-hydroxylase deficiency
	Non-PKU hyperphenylalanemia
	Optic atrophy 3 with cataract
Familial gestational hyperthyroidism	Ornithine transcarbamylase deficiency
Familial hyperinsulinemic hypoglycemia type 4	Palmoplantar keratoderma with deafness
Favism	Partial adenosine deaminase deficiency
Galactokinase deficiency with cataracts	Pendred syndrome
Galactose epimerase deficiency Galactosemia	Perinatal lethal Gaucher disease
Gaucher disease Type I	Phenylketonuria
Gaucher disease Type II	Propionicacidemia
Gaucher disease Type III	Severe combined immunodeficiency (SCID) due to adenosine deaminase deficiency (ADAD)
Gaucher disease Type IIIC	Sickle cell anemia (S/S)
	Sickle cell disease variants
Glutaric acidemia IIA	Sickle hemoglobin C disease
Glutaric acidemia IIB	Sickle hemoglobin D disease
Glutaric acidemia IIC	Sickle hemoglobin E disease
Glutaric aciduria Type I	Susceptibility to acute-infection induced encephalopathy
Glycine N-methyltransferase deficiency	Susceptibility to autoimmune thyroid disease Type III
Glycogen storage disease Ia Glycogen storage disease II	Systemic primary carnitine deficiency
GM2-gangliosidosis	Tay-Sachs disease
Hawkinsinuria	Thryoid dyshormonogenesis 6
	Thyroid dyshormonogenesis 1
Fragile X Syndrome	Thyroid dyshormonogenesis 2A
Spinal Muscular Atrophy	Thyroid dyshormonogenesis 3
Vitamin B-12 responsive methylmalonic aciduria	Thyroid hormone resistance
VLCAD deficiency	Transcobalamin II deficiency
Vohwinkel syndrome	Trifunctional protein deficiency
X-linked mental retardation with methylmalonic acidemia and homocysteinemia	Tyrosinemia, type I
	Tyrosinemia, type II
X-linked severe combined immunodeficiency (SCID)	Tyrosinemia, type III

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Clinical Laboratory Improvement Amendments

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