preparent®

CARRIER TEST

global panels diseases tested

WHY TEST FOR THESE DISEASES?

- This test includes 200+ hereditary diseases that can cause serious health problems, beginning in infancy or childhood.
- Children born with these diseases often benefit from early diagnosis and treatment.
- Most children born with a genetic disease have no affected family members.
- Being a carrier is common. About **1 in 3 people** are found to be a carrier when tested for these diseases.

progenity®

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GLOBAL PANELS DISEASES TESTED

17-alpha-hydroxylase deficiency
17-beta-hydroxysteroid dehydrogenase deficiency, type III
3-beta-hydroxysteroid dehydrogenase deficiency, type II
3-hydroxy-3-methylglutaryl CoA lyase deficiency
3-methylglutaconic aciduria, type III
3-phosphoglycerate dehydrogenase deficiency
Abetalipoproteinemia
Achalasia-addisonianism-alacrima syndrome
Achromatopsia, *CNGA3*-related
Achromatopsia, *CNGB3*-related
Acrodermatitis enteropathica
Adenosine deaminase deficiency
Adrenoleukodystrophy **(X-linked)***
Aicardi-Goutieres syndrome, *RNASEH2C*-related
Aicardi-Goutieres syndrome, *SAMHD1*-related
Aicardi-Goutieres syndrome, *TREX1*-related
Alkaptonuria
Alpha-mannosidosis
Alpha-thalassemia
Alport syndrome, *COL4A3*-related
Alport syndrome, *COL4A5*-related **(X-linked)***
Amish infantile epilepsy syndrome
Andermann syndrome
Argininosuccinic aciduria
Aromatase deficiency
Arthrogryposis, mental retardation and seizures
Arts syndrome **(X-linked)***
Aspartylglycosaminuria
Ataxia-telangiectasia
Ataxia-telangiectasia-like disorder
Ataxia with vitamin E deficiency
Autoimmune polyglandular syndrome, type I
Autosomal recessive polycystic kidney disease
Bardet-Biedl syndrome, *BBS1*-related
Bardet-Biedl syndrome, *BBS10*-related
Bardet-Biedl syndrome, *BBS12*-related
Bartter syndrome, type IV
Beta-ketothiolase deficiency
Beta-thalassemia
Bilateral frontoparietal polymicrogyria
Biotinidase deficiency
Bloom syndrome
Canavan disease
Carnitine palmitoyltransferase I deficiency
Carnitine palmitoyltransferase II deficiency
Carpenter syndrome
Cartilage-hair hypoplasia
Charcot-Marie-Tooth disease, *GJB1*-related **(X-linked)***
Charcot-Marie-Tooth disease, *PRPS1*-related **(X-linked)***
Chediak-Higashi syndrome
Cholesteryl ester storage disease
Choroideremia **(X-linked)***
Citrullinemia, type I
Congenital amegakaryocytic thrombocytopenia
Congenital disorder of glycosylation, type IA
Congenital disorder of glycosylation, type IB
Congenital lipoid adrenal hyperplasia
Congenital neutropenia, *HAX1*-related
Corneal dystrophy and perceptive deafness syndrome
Corticosterone methyloxidase deficiency
Creatine transporter defect, *SLC6A8*-related **(X-linked)***
Crigler-Najjar syndrome
Cystic fibrosis (600 mutations)
Cystinosis
D-bifunctional protein deficiency
Desbuquois dysplasia, type I
Dihydrolipoamide dehydrogenase deficiency
Dihydropyrimidine dehydrogenase deficiency
Du Pan syndrome
Dyskeratosis congenita, *RTEL1*-related
Dyskeratosis congenita, *DKC1*- related **(X-linked)***
Dystrophic epidermolysis bullosa, *COL7A1*-related
Early onset myopathy with fatal cardiomyopathy
Ehlers-Danlos syndrome, type VIIC
Emery-Dreifuss muscular dystrophy **(X-linked)***
Enhanced S-cone syndrome
Ethylmalonic encephalopathy
Fabry disease **(X-linked)***
Familial dysautonomia
Familial hyperinsulinism, *ABCC8*-related
Familial hyperinsulinism, *KCNJ11*-related
Familial Mediterranean fever
Fanconi anemia, type A
Fanconi anemia, type C
Fetal akinesia deformation sequence, *DOK7*-related
Fragile X syndrome **(X-linked)***
Fumarase deficiency
Galactokinase deficiency
Galactosemia
Gaucher disease
Geroderma osteodysplastica
Gitelman syndrome
Glucose-6-phosphate dehydrogenase deficiency **(X-linked)***
Glutaric acidemia, type I
Glycine encephalopathy, *AMT*-related
Glycine encephalopathy, *GLDC*-related
Glycogen storage disease, type IA
Glycogen storage disease, type IB
Glycogen storage disease, type II
Glycogen storage disease, type III
Glycogen storage disease, type IV
Glycogen storage disease, type V
Glycogen storage disease, type VII
GM1-gangliosidosis
GRACILE syndrome
Guanidinoacetate methyltransferase deficiency
Hemoglobinopathy, Hb C
Hemoglobinopathy, Hb D
Hemoglobinopathy, Hb E
Hemoglobinopathy, Hb O
Hemophilia A **(X-linked)***
Hemophilia B **(X-linked)***
Hepatocerebral mitochondrial DNA depletion syndrome, *MPV17*-related
Hermansky-Pudlak syndrome, type III
Holocarboxylase synthetase deficiency
Homocystinuria, *CBS*-related
Hyperphosphatemic familial tumoral calcinosis
Hypohidrotic ectodermal dysplasia
Hypophosphatasia
Isovaleric acidemia
Joubert syndrome 2
Junctional epidermolysis bullosa, *LAMB3*-related
Juvenile retinoschisis **(X-linked)***
Krabbe disease
Leber congenital amaurosis, *LCA5*-related
Leber congenital amaurosis, *RDH12*-related
Leigh syndrome, French Canadian
Limb-girdle muscular dystrophy, type 2A
Limb-girdle muscular dystrophy, type 2D
Limb-girdle muscular dystrophy, type 2E
Limb-girdle muscular dystrophy, type 2I
Lipoprotein lipase deficiency
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
Luteinizing hormone resistance
Maple syrup urine disease, type IA
Maple syrup urine disease, type IB
Medium-chain acyl-CoA dehydrogenase deficiency
MEDNIK syndrome
Metachromatic leukodystrophy
Methylmalonic aciduria, cblA type
Methylmalonic aciduria, cblB type
Methylmalonic aciduria, cblC type
Methylmalonic aciduria, *MUT*-related
Mitochondrial complex IV deficiency
Mitochondrial myopathy and sideroblastic anemia
Mucolipidosis, type II/III alpha/beta
Mucolipidosis, type IV
Mucopolysaccharidosis, type I-Hurler syndrome
Mucopolysaccharidosis, type II-Hunter syndrome **(X-linked)***
Mucopolysaccharidosis, type IIIC
Mucopolysaccharidosis, type VI
Mulibrey nanism
Multiple sulfatase deficiency
Muscle-eye-brain disease
Myotubular myopathy, *MTM1*-related **(X-linked)***
Nemaline myopathy 2
Nephrotic syndrome, type I
Neuronal ceroid lipofuscinosis, *CLN5*-related
Neuronal ceroid lipofuscinosis, *CLN6*-related
Neuronal ceroid lipofuscinosis, *CLN8*-related
Neuronal ceroid lipofuscinosis, *MFSD8*-related
Neuronal ceroid lipofuscinosis, *PPT1*-related
Neuronal ceroid lipofuscinosis, *TPP1*-related
Niemann-Pick disease, type A and B
Niemann-Pick disease, type CI
Niemann-Pick disease, type CII
Nijmegen breakage syndrome
Nonsyndromic hearing loss, *GJB2*-related
Omenn syndrome
Ornithine transcarbamylase deficiency **(X-linked)***
Ornithine translocase deficiency
Pendred syndrome
Phenylalanine hydroxylase deficiency
Pontocerebellar hypoplasia, type IA
Primary congenital glaucoma
Primary hyperoxaluria, type I
Primary hyperoxaluria, type II
Progressive familial intrahepatic cholestasis, type II
Prolidase deficiency
Propionic acidemia, *PCCA*-related
Propionic acidemia, *PCCB*-related
Pseudoxanthoma elasticum
Pycnodysostosis
Pyruvate dehydrogenase deficiency, *PDHB*-related
Pyruvate dehydrogenase deficiency, *PDHA1*-related **(X-linked)***
Retinitis pigmentosa 59
Rhizomelic chondrodysplasia punctata, type I
Salla disease
Sandhoff disease
Severe combined immunodeficiency, *RAG1*-related
Severe combined immunodeficiency, *IL2RG*-related **(X-linked)***
Shwachman-Diamond syndrome
Sickle cell anemia (Hb S)
Sjögren-Larsson syndrome
Smith-Lemli-Opitz syndrome
Spastic ataxia of Charlevoix-Saguenay (ARSACS)
Spinal muscular atrophy
Steroid-resistant nephrotic syndrome
Stuve-Wiedemann syndrome
Sulfate transporter-related osteochondrodysplasia
Tay-Sachs disease
Tumoral calcinosis, normophosphatemic
Tyrosinemia, type I
Usher syndrome, type IB
Usher syndrome, type IC
Usher syndrome, type ID
Usher syndrome, type IF
Usher syndrome, type III
Very long-chain acyl-CoA dehydrogenase deficiency
Vitamin D-dependent rickets, type I
Walker-Warburg syndrome
Wilson disease
Zellweger syndrome spectrum, *PEX6*-related

GLOBAL+ ONLY

Alpha-1 antitrypsin deficiency
Autosomal recessive woolly hair/hypotrichosis
Hereditary fructose intolerance
Hereditary hemochromatosis, *HFE*-related
Hereditary hemochromatosis, *HFE2*-related
Hereditary hemochromatosis, *TFR2*-related
Inclusion body myopathy, type II
Lamellar ichthyosis, type I
Limb-girdle muscular dystrophy, type 2B
Pseudocholinesterase deficiency
Spastic paraplegia, *ZFYVE26*-related
Xeroderma pigmentosum

*X-linked diseases are not tested in males.