Informed Consent – Global

preparent®
carrier test

About this test

This test tells you if you're a carrier of a genetic disease.

Being a carrier means one of your genes has a change, and it doesn't work. Carriers of a disease are often healthy but can have a child with the disease.

Preparent® Global tests for the diseases listed on the back of this form.

- These diseases were chosen because they have harmful health effects.
- These effects often start at a young age and do not have a cure.
- Having a child with one of these diseases can happen to anyone, no matter your health, age, ethnicity, or family history.

What test results mean

Positive (abnormal) results
mean that you are a carrier for one (or more) of the genetic diseases tested. Your risk to have a child with these diseases is **higher** than most other people. Follow-up testing may be recommended.

Negative (normal) results
mean that you are not a carrier for any of the gene changes tested. Your risk to have a child with these diseases is **lower** than most other people.

Benefits

- Finding out these results will help you understand your risk to have a baby affected with the diseases tested.

Limitations

- Negative results do not guarantee a healthy pregnancy or baby. These tests look for specific changes to your genes. Changes not targeted by these tests will not be detected.
- False positive, false negative, and failed results are rare, but can happen.

Your privacy is protected

- We keep your results and information private. We only send results to the ordering provider, unless you give us permission to send elsewhere. You can contact us for a copy of your results.
- No other clinical test is performed or reported on a sample, unless ordered by a provider. We may contact a provider to obtain follow-up information. This is a normal lab practice and required in several states.
- Only anonymous samples and data are used for lab quality and data sharing programs. These are normal lab practices. We destroy samples received from New York State within 60 days after testing.

Before signing this form, I had the chance to talk about this test with my healthcare provider or someone he/she chose, and genetic counseling has been recommended before and after testing. My questions have been answered and I have all of the information that I need to decide. I understand that this test is voluntary. I have decided that:

- Preparent® Carrier Test
  - ◯ Yes, I want to receive the **Preparent® Carrier Test - Global**
  - ◯ No, I do not want to receive the **Preparent® Carrier Test - Global**

PATIENT NAME *(please print)* _____ DATE OF BIRTH _____

PATIENT SIGNATURE _____ DATE _____

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Preparent Global (200+ diseases)

Disease / Gene
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

Disease / Gene
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

Disease / Gene
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.