Informed Consent – Exon Panel

preparent®
CARRIER TEST

About this test

This test looks at your genes to find out if you are a carrier for certain genetic diseases.

A **gene** is made up of DNA, and genes play an important role in our health. People typically have two working copies of most genes. But some people have only one working copy of a gene. The other copy has a change and does not work. This is called being a **carrier** of a genetic disease. Carriers are usually healthy, but their children have a higher risk for disease. **Being a carrier is common.** However, most carriers have not had carrier testing and do not know they are at risk of having a child with a genetic disease. The **genetic diseases** on this test were chosen because they have harmful health effects that often start at a young age and do not have a cure. A full list of diseases tested using the Preparent Exon Panel is located on the back of this form.

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 whether you are a carrier for a genetic disease will help you understand your risk to have a child affected with the diseases tested.
- Negative results are reassuring. Positive results let you and your provider determine the next steps for the identified risk(s).

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 test will be performed or reported on your sample, unless ordered by your provider. Samples received from New York State will be destroyed within 60 days after testing.
- We may contact your provider to obtain follow-up information after your test is complete. This is a standard lab practice, and is required in several states.

Limitations

- Negative results do not guarantee a healthy pregnancy or baby. This test looks for specific changes to your genes. Genetic changes not targeted by this test will not be detected.
- False positive, false negative, and failed results are rare, but possible.

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:

○ Yes, I want to receive the **Preparent® Carrier Test.**

○ No, I do not want to receive the **Preparent® Carrier Test.**

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

PATIENT SIGNATURE _____ DATE _____

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The Preparent® Exon Panel tests for 150+ diseases.

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3-hydroxy-3-methylglutaryl CoA lyase deficiency
3-phosphoglycerate dehydrogenase deficiency
6-pyruvoyl-tetrahydropterin synthase (PTS) deficiency
Abetalipoproteinemia
Adenosine deaminase deficiency
Adrenoleukodystrophy (X-linked)*
Agammaglobulinemia (X-linked)*
Alpha-1 antitrypsin deficiency
Alpha-mannosidosis
Alpha-thalassemia
Alport syndrome, COL4A3-related
Andermann syndrome
Angelman syndrome
Argininosuccinic aciduria
Arthrogryposis, mental retardation and seizures
Aspartylglycosaminuria
Ataxia with vitamin E deficiency
Ataxia-telangiectasia
Autosomal recessive polycystic kidney disease
Bardet-Biedl syndrome, BBS1-related
Bardet-Biedl syndrome, BBS10-related
Bardet-Biedl syndrome, BBS2-related
Beta-hemoglobinopathies
Biotinidase deficiency
Bloom syndrome
Canavan disease
Carnitine palmitoyltransferase IA deficiency
Carnitine palmitoyltransferase II deficiency
Cartilage-hair hypoplasia
Cerebrotendinous xanthomatosis
Chronic granulomatous disease (X-linked)*
Citrin deficiency
Citrullinemia, type I
Combined pituitary hormone deficiency, PROP1-related
Congenital amegakaryocytic thrombocytopenia
Congenital disorder of glycosylation, type IA
Congenital disorder of glycosylation, type IB
Congenital myasthenic syndrome, CHAT-related
Congenital myasthenic syndrome, CHRNE-related
Congenital myasthenic syndrome, RAPSN-related
Crigler-Najjar syndrome
Cystic fibrosis
Cystinosis
D-bifunctional protein deficiency
Dihydrolipoamide dehydrogenase deficiency
Dihydropyrimidine dehydrogenase deficiency
Duchenne/Becker muscular dystrophy (X-linked)*
Dyskeratosis congenita, RTEL1-related
Ehlers Danlos syndrome, type VIIC
Ethlymalonic encephalopathy
Familial dysautonomia
Familial hyperinsulinism, ABCC8-related
Fanconi anemia, type C
Fetal akinesia deformation sequence, DOK7-related
Fragile X syndrome (X-linked)*
Fumarase deficiency
Galactosemia
Gaucher disease
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
GM1-gangliosidosis
GRACILE syndrome
Hereditary fructose intolerance
Hermansky-Pudlak syndrome, type III
Homocystinuria, CBS-related
Hypophosphatasia, autosomal recessive
Inclusion body myopathy, type II
Infantile neuroaxonal dystrophy, type 1
Isovaleric acidemia
Joubert syndrome 2
Junctional epidermolysis bullosa, LAMA3-related
Junctional epidermolysis bullosa, LAMB3-related
Junctional epidermolysis bullosa, LAMC2-related
Juvenile nephronophthisis, type 1
Krabbe disease
Lamellar ichthylosis, type I
Leigh syndrome, French Canadian
Leukoencephalopathy with vanishing white matter
Limb-girdle muscular dystrophy, type 2A
Limb-girdle muscular dystrophy, type 2C
Limb-girdle muscular dystrophy, type 2D
Limb-girdle muscular dystrophy, type 2E
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
Lowe syndrome (X-linked)*
Lysinuric protein intolerance
Maple syrup urine disease, type IA
Maple syrup urine disease, type IB
Maple syrup urine disease, type II
MECP2 duplication syndrome (X-linked)*
Medium-chain acyl-CoA dehydrogenase deficiency
Megalencephalic leukoencephalopathy with subcortical cysts, type I
Metachromatic leukodystrophy
Methylmalonic aciduria, cblC type
Mucolipidosis type IV
Mucolipidosis, type II/III alpha/beta
Mucopolysaccharidosis type IIIA (Sanfilippo A)
Mucopolysachharidosis, type I-Hurler syndrome
Multiple sulfatase deficiency
Muscle-eye-brain disease
Nemaline myopathy 2
Nephrotic syndrome, type I
Neuronal ceroid lipofuscinosis, CLN3-related
Neuronal ceroid lipofuscinosis, CLN5-related
Neuronal ceroid lipofuscinosis, CLN6-related
Neuronal ceroid lipofuscinosis, CLN8-related
Neuronal ceroid lipofuscinosis, PPT1-related
Neuronal ceroid lipofuscinosis, TPP1-related
Niemann-Pick disease, type A and B
Niemann-Pick disease, type C1
Nijmegen breakage syndrome
Nonsyndromic hearing loss, GJB2-related
Nonsyndromic hearing loss, GJB6-related
Omenn syndrome
Ornithine transcarbamylase deficiency (X-linked)*
Ornithine translocase deficiency
Pendred syndrome
Phenylalanine hydroxylase deficiency
POLG-related disorders
Primary carnitine deficiency
Primary congenital glaucoma
Primary hyperoxaluria, type I
Primary hyperoxaluria, type II
Propionic acidemia, PCCA-related
Propionic acidemia, PCCB-related
Pycnodysostosis
Pyruvate carboxylase deficiency
Retinitis pigmentosa 59
Rhizomelic chondrodysplasia punctata, type I
Salla disease
Sandhoff disease
Severe combined immunodeficiency, IL2RG-related (X-linked)*
Sjögren-Larsson syndrome
Smith-Lemli-Opitz syndrome
Spastic ataxia of Charlevoix-Saguenay (ARSACS)
Spinal muscular atrophy
Steroid-resistant nephrotic syndrome
Sulfate transporter-related osteochondrodysplasia
Tay-Sachs disease
Tyrosine hydroxylase deficiency
Tyrosinemia, type I
Usher syndrome, type IB
Usher syndrome, type IC
Usher syndrome, type ID
Usher syndrome, type IF
Usher syndrome, type IIA
Usher syndrome, type III
Very long-chain acyl-CoA dehydrogenase deficiency
Walker-Warburg syndrome
Wilson disease
Wiskott-Aldrich syndrome (X-linked)*
Zellweger spectrum disorders, PEX1-related
Zellweger spectrum disorders, PEX2-related

*X-linked diseases are not tested in males.