

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® Standard 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.

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 - Standard**
- No, I do not want to receive the **Preparent® Carrier Test - Standard**

PATIENT NAME (please print)

DATE OF BIRTH

PATIENT SIGNATURE

DATE

5230 S. State Road, Ann Arbor, MI 48108 USA • Tel +1 855-293-2639 • progenity.com

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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.

Preparent Standard (29 diseases)

Disease / Gene
Alpha-thalassemia <i>HBA1, HBA2</i>
Beta-thalassemia <i>HBB</i>
Bloom syndrome <i>BLM</i>
Canavan disease <i>ASPA</i>
Congenital amegakaryocytic thrombocytopenia <i>MPL</i>
Cystic fibrosis <i>CFTR</i>
Dihydroipoamide dehydrogenase Deficiency <i>DLD</i>
Familial dysautonomia <i>IKBKAP</i>
Familial hyperinsulinism, ABCC8-related <i>ABCC8</i>
Fanconi anemia, type C <i>FANCC</i>
Fragile X syndrome (X-linked)* <i>FMR1</i>
Gaucher disease <i>GBA</i>
Glycogen storage disease, type IA <i>G6PC</i>
Hemoglobinopathy, Hb C <i>HBB</i>
Hemoglobinopathy, Hb D <i>HBB</i>
Hemoglobinopathy, Hb E <i>HBB</i>
Hemoglobinopathy, Hb O <i>HBB</i>
Joubert syndrome 2 <i>TMEM216</i>
Maple syrup urine disease, type IA <i>BCKDHA</i>
Maple syrup urine disease, type IB <i>BCKDHB</i>
Mucopolipidosis, type IV <i>MCOLN1</i>
Nemaline myopathy 2 <i>NEB</i>
Niemann-Pick disease, type A and B <i>SMPD1</i>
Sickle cell anemia (Hb S) <i>HBB</i>
Spinal muscular atrophy <i>SMN1</i>
Tay-Sachs disease <i>HEXA</i>
Usher syndrome, type IF <i>PCDH15</i>
Usher syndrome, type III <i>CLRN1</i>
Walker-Warburg syndrome <i>FKTN</i>

*X-linked diseases are not tested in males.