Informed Consent – Standard 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 only have 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 Standard 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® Standard Panel tests for 29 diseases recommended for carrier testing by medical guidelines.

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

*X-linked diseases are not tested in males.