Informed Consent – Ashkenazi Jewish

preparent®
carrier test

About this test

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

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

The Preparent® Ashkenazi Jewish Carrier Test checks for the disorders listed on the back of this form.

- These disorders 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 disorders 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 disorders tested. Your risk to have a child with these disorders 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 disorders is **lower** than most other people.

Benefits

- Finding out these results will help you understand your risk to have a baby affected with the disorders 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 Ashkenazi Jewish Carrier Test

◯ Yes, I want to receive this test.

◯ No, I do not want to receive this test.

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

PATIENT SIGNATURE _____ DATE _____

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Preparent Ashkenazi Jewish Carrier Test (35 genes)

Disorder / *Gene*
3-phosphoglycerate dehydrogenase deficiency, *PHGDH*
Abetalipoproteinemia, *MTTP*
Bloom syndrome, *BLM*
Canavan disease, *ASPA*
Carnitine palmitoyltransferase II deficiency, *CPT2*
Congenital amegakaryocytic thrombocytopenia, *MPL*
Cystic fibrosis, *CFTR*
Dihydrolipoamide dehydrogenase deficiency, *DLD*
Ehlers-Danlos syndrome, type VIIC, *ADAMTS2*
Familial dysautonomia, *IKBKAP*
Familial hyperinsulinism, ABCC8-related, *ABCC8*
Familial Mediterranean fever, *MEFV*
Fanconi anemia, type C, *FANCC*
Fragile X syndrome, *FMR1*
Gaucher disease, *GBA*
Glycogen storage disease, type IA, *G6PC*
Glycogen storage disease, type IV, *GBE1*
Glycogen storage disease, type VII, *PFKM*
Hermansky-Pudlak syndrome, type III, *HPS3*
Joubert syndrome 2, *TMEM216*
Krabbe disease, *GALC*
Maple syrup urine disease, type IB, *BCKDHB*
Mucolipidosis, type IV, *MCOLN1*
Nemaline myopathy 2, *NEB*
Niemann-Pick disease, type A and B, *SMPD1*
Nonsyndromic hearing loss, GJB2-related, *GJB2*
Pontocerebellar hypoplasia, type IA, *VRK1*
Retinitis pigmentosa 59, *DHDDS*
Smith-Lemli-Opitz syndrome, *DHCR7*
Spinal muscular atrophy, *SMN1/SMN2*
Tay-Sachs disease (DNA), *HEXA*
Usher syndrome, type IF, *PCDH15*
Usher syndrome, type III, *CLRN1*
Walker-Warburg syndrome, *FKTN*
Wilson disease, *ATP7B*