Informed Consent - Exon

preparent® exon
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 Exon 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.
- Negative results are reassuring. Positive results let you and your provider determine the next steps for the identified risk(s).

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

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

PATIENT SIGNATURE _____ DATE _____

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Preparent Exon (150+ genes)

Disease / *Gene*
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency *HMGCL*
3-Phosphoglycerate Dehydrogenase Deficiency *PHGDH*
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency *PTS*
Abetalipoproteinemia *MTTP*
Adenosine Deaminase Deficiency *ADA*
Adrenoleukodystrophy *ABCD1**
Agammaglobulinemia *BTK**
Alpha-1 Antitrypsin Deficiency *SERPINA1*
Alpha-Mannosidosis *MAN2B1*
Alpha-Thalassemia *HBA1, HBA2*
Alport Syndrome *COL4A3*
Andermann Syndrome *SLC12A6*
Angelman Syndrome *UBE3A*
Argininosuccinate Aciduria *ASL*
Arthrogryposis, Mental Retardation, and Seizures *SLC35A3*
Aspartylglycosaminuria *AGA*
Ataxia with Vitamin E Deficiency *TTPA*
Ataxia-Telangiectasia *ATM*
Autosomal Recessive Polycystic Kidney Disease *PKHD1*
Bardet-Biedl Syndrome *BBS1, BBS2, BBS10*
Beta-Hemoglobinopathies, including Sickle Cell Anemia *HBB*
Biotinidase Deficiency *BTD*
Bloom Syndrome *BLM*
Canavan Disease *ASPA*
Carnitine Deficiency, Systemic Primary *SLC22A5*
Carnitine Palmitoyltransferase I Deficiency *CPT1A*
Carnitine Palmitoyltransferase II Deficiency *CPT2*
Cartilage-Hair Hypoplasia *RMRP*
Cerebrotendinous Xanthomatosis *CYP27A1*
Chronic Granulomatous Disease *CYBB**
Citrin Deficiency *SLC25A13*
Citrullinemia *ASS1*
Congenital Amegakaryocytic Thrombocytopenia *MPL*
Congenital Disorder of Glycosylation, Type IA *PMM2*
Congenital Disorder of Glycosylation, Type IB *MPI*
Congenital Myasthenic Syndrome *CHAT, CHRNE, DOK7, RAPSN*
Crigler-Najjar syndrome *UGT1A1*
Cystic Fibrosis *CFTR*
Cystinosis *CTNS*
D-Bifunctional Protein Deficiency *HSD17B4*
Dihydrolipoamide Dehydrogenase Deficiency *DLD*
Dihydropyrimidine Dehydrogenase Deficiency *DPYD*
Duchenne/Becker Muscular Dystrophy *DMD**
Dyskeratosis Congenita *RTEL1*
Ehlers-Danlos Syndrome *ADAMTS2*
Ethylmalonic Encephalopathy *ETHE1*
Familial Dysautonomia *ELP1*
Familial Hyperinsulinism *ABCC8*
Fanconi Anemia *FANCC*
Fragile X Syndrome *FMR1**
Fumarase Deficiency *FH*
Galactosemia *GALT*
Gaucher Disease *GBA*
Glucose-6-Phosphate Dehydrogenase Deficiency *G6PD**
Glutaric Acidemia *GCDH*
Glycine Encephalopathy *AMT, GLDC*
Glycogen Storage Disease, Type Ia *G6PC*
Glycogen Storage Disease, Type Ib *SLC37A4*
Glycogen Storage Disease, Type II *GAA*
Glycogen Storage Disease, Type III *AGL*
GM1 Gangliosidosis *GLB1*
GRACILE Syndrome *BCS1L*
Hereditary Fructose Intolerance *ALDOB*
Hermansky-Pudlak Syndrome *HPS3*
Homocystinuria *CBS*

Disease / *Gene*
Hypophosphatasia *ALPL*
Inclusion Body Myopathy *GNE*
Infantile Neuroaxonal Dystrophy *PLA2G6*
Isovaleric Acidemia *IVD*
Joubert Syndrome *TMEM216*
Junctional Epidermolysis Bullosa *LAMA3, LAMB3, LAMC2*
Juvenile Nephronophthisis *NPHP1*
Krabbe Disease *GALC*
Lamellar Ichthyosis *TGM1*
Leigh Syndrome, French-Canadian *LRPPRC*
Leukoencephalopathy with Vanishing White Matter *EIF2B5*
Limb-Girdle Muscular Dystrophy *CAPN3, SGCA, SGCB, SGCG*
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency *HADHA*
Lowe Syndrome *OCRL**
Lysinuric Protein Intolerance *SLC7A7*
Maple Syrup Urine Disease *BCKDHA, BCKDHB, DBT*
MECP2 Duplication Syndrome *MECP2**
Medium-Chain Acyl-CoA Dehydrogenase Deficiency *ACADM*
Megalencephalic Leukoencephalopathy with Subcortical Cysts *MLC1*
Metachromatic Leukodystrophy *ARSA*
Methylmalonic Aciduria *MMACHC*
Mucolipidosis, Type II/III Alpha/Beta *GNPTAB*
Mucolipidosis, Type IV *MCOLN1*
Mucopolysaccharidosis, Type I; Hurler Syndrome *IDUA*
Mucopolysaccharidosis, Type IIIA; Sanfilippo A *SGSH*
Multiple Sulfatase Deficiency *SUMF1*
Muscle-Eye-Brain Disease, *POMGNT1*-Related *POMGNT1*
Nemaline Myopathy *NEB*
Nephrotic Syndrome *NPHS1, NPHS2*
Neuronal Ceroid Lipofuscinosis *CLN3, CLN5, CLN6, CLN8, PPT1, TPP1*
Niemann-Pick Disease, Type A/B *SMPD1*
Niemann-Pick Disease, Type CI/D *NPC1*
Nijmegen Breakage Syndrome *NBN*
Nonsyndromic Hearing Loss *GJB2, GJB6*
Omenn Syndrome *DCLRE1C*
Ornithine Transcarbamylase Deficiency *OTC**
Ornithine Translocase Deficiency/HHH syndrome *SLC25A15*
Pendred Syndrome *SLC26A4*
Phenylalanine Hydroxylase Deficiency *PAH*
POLG-Related Disorders *POLG*
Primary Congenital Glaucoma *CYP1B1*
Primary Hyperoxaluria *AGXT, GRHPR*
PROP1-Related Combined Pituitary Hormone Deficiency *PROP1*
Propionic Acidemia *PCCA, PCCB*
Pycnodysostosis *CTSK*
Pyruvate Carboxylase Deficiency *PC*
Retinitis Pigmentosa *DHDDS*
Rhizomelic Chondrodysplasia Punctata *PEX7*
Salla Disease *SLC17A5*
Sandhoff Disease *HEXB*
Severe Combined Immunodeficiency *IL2RG**
Sjögren-Larsson Syndrome *ALDH3A2*
SLC26A2-Related Skeletal Dysplasias *SLC26A2*
Smith-Lemli-Opitz Syndrome *DHCR7*
Spastic Ataxia of Charlevoix-Saguenay *SACS*
Spinal Muscular Atrophy *SMN1*
Tay-Sachs Disease *HEXA*
Tyrosine Hydroxylase Deficiency *TH*
Tyrosinemia *FAH*
Usher Syndrome *CDH23, CLRN1, MYO7A, PCDH15, USH1C, USH2A*
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency *ACADVL*
Walker-Warburg Syndrome and Other *FKTN*-Related Diseases *FKTN*
Wilson Disease *ATP7B*
Wiskott-Aldrich Syndrome *WAS**
Zellweger Spectrum Disorders *PEX1, PEX2*

*These genes cause X-linked disease and are not tested in males. The remaining genes cause autosomal recessive diseases.