Informed Consent - Exon Global

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

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

PATIENT SIGNATURE _____ DATE _____

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Preparent Exon Global (280+ genes)

Disease / *Gene*
3-Beta-Hydroxysteroid Dehydrogenase Deficiency *HSD3B2*
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency *HMGCL*†
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency *MCCC1*
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency *MCCC2*
3-Methylglutaconic Aciduria *OPA3*
3-Phosphoglycerate Dehydrogenase Deficiency *PHGDH*†
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency *PTS*†
Abetalipoproteinemia *MTTP*†
Achromatopsia *CNGB3*
Acrodermatitis Enteropathica *SLC39A4*
Acute Infantile Liver Failure *TRMU*
Acyl-CoA Oxidase I Deficiency *ACOX1*
Adenosine Deaminase Deficiency *ADA*†
Adrenoleukodystrophy *ABCD1*†*
Agammaglobulinemia *BTK*†*
Aicardi-Goutières Syndrome *SAMHD1*
Alpha-Mannosidosis *MAN2B1*†
Alpha-Thalassemia *HBA1*†, *HBA2*†
Alpha-Thalassemia Intellectual Disability Syndrome *ATRX**
Alport Syndrome *COL4A5**, *COL4A3*†, *COL4A4*
Alstrom Syndrome *ALMS1*
Andermann Syndrome *SLC12A6*†
Argininosuccinate Aciduria *ASL*†
Aromatase Deficiency *CYP19A1*
Arthrogryposis, Mental Retardation, and Seizures *SLC35A3*†
Asparagine Synthetase Deficiency *ASNS*
Aspartylglycosaminuria *AGA*†
Ataxia with Vitamin E Deficiency *TTPA*†
Ataxia-Telangiectasia *ATM*†
Autoimmune Polyglandular Syndrome *AIRE*
Autosomal Recessive Polycystic Kidney Disease *PKHD1*†
Bardet-Biedl Syndrome *BBS1*†, *BBS2*†, *BBS10*†, *BBS12*
Bare Lymphocyte Syndrome *CIITA*
Bartter Syndrome *BSND*
Beta-Hemoglobinopathies, including Sickle Cell Anemia *HBB*†
Beta-ketothiolase Deficiency *ACAT1*
Bilateral Frontoparietal Polymicrogyria *ADGRG1*
Biotinidase Deficiency *BTD*†
Bloom Syndrome *BLM*†
Canavan Disease *ASPA*†
Carbamoyl Phosphate Synthetase I Deficiency *CPS1*
Carnitine Deficiency, Systemic Primary *SLC22A5*†
Carnitine Palmitoyltransferase I Deficiency *CPT1A*†
Carnitine Palmitoyltransferase II Deficiency *CPT2*†
Carpenter Syndrome *RAB23*
Cartilage-Hair Hypoplasia *RMRP*†
Cerebrotendinous Xanthomatosis *CYP27A1*†
Charcot-Marie-Tooth Disease *GJB1**, *NDRG1*
Cholesteryl Ester Storage Disease *LIPA*
Choreoacanthocytosis *VPS13A*
Choroideremia *CHM**
Chronic Granulomatous Disease *CYBB†**
Chronic Granulomatous Disease, Cytochrome b-negative *CYBA*
Ciliopathies, RPGRIP1L-Related *RPGRIP1L*
Citrin Deficiency *SLC25A13*†
Citrullinemia *ASS1*†
Cohen Syndrome *VPS13B*
Combined Malonic and Methylmalonic Aciduria *ACSF3*
Combined Oxidative Phosphorylation Deficiency *TSFM, GFM1*
Combined Pituitary Hormone Deficiency 3 *LHX3*
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficiency *CYP17A1*
Congenital Amegakaryocytic Thrombocytopenia *MPL*†
Congenital Disorder of Glycosylation, Type IA *PMM2*†
Congenital Disorder of Glycosylation, Type IB *MPI*†
Congenital Disorder of Glycosylation, Type IC *ALG6*
Congenital Insensitivity to Pain with Anhidrosis *NTRK1*
Congenital Lipoid Adrenal Hyperplasia *STAR*
Congenital Myasthenic Syndrome *CHAT*†, *CHRNE*†, *DOK7*†, *RAPSN*†
Congenital Neutropenia *HAX1, VPS45*
Corneal Dystrophy and Perceptive Deafness Syndrome *SLC4A11*
Corticosterone Methyloxidase Deficiency *CYP11B2*
CRB1-Related Retinal Dystrophies *CRB1*
Creatine Transporter Defect *SLC6A8**
Cystic Fibrosis *CFTR*†
Cystinosis *CTNS*†
D-Bifunctional Protein Deficiency *HSD17B4*†
Dihydrolipoamide Dehydrogenase Deficiency *DLD*†
Dihydropyrimidine Dehydrogenase Deficiency *DPYD*†
Duchenne/Becker Muscular Dystrophy *DMD*†*

Disease / *Gene*
Dyskeratosis Congenita *RTEL1*†
Dystrophic Epidermolysis Bullosa *COL7A1*
Ehlers-Danlos Syndrome *ADAMTS2*†
Ellis-van Creveld Syndrome *EVC*
Emery-Dreifuss Muscular Dystrophy *EMD**
Enhanced S-Cone Syndrome *NR2E3*
Ethylmalonic Encephalopathy *ETHE1*†
Fabry Disease *GLA**
Factor XI Deficiency *F11*
Familial Dysautonomia *ELP1*†
Familial Hypercholesterolemia *LDLRAP1*
Familial Hyperinsulinism *ABCC8*†, *KCNJ11*
Familial Mediterranean Fever *MEFV*
Fanconi Anemia *FANCA, FANCC*†, *FANCG*
Fragile X Syndrome *FMR1*†*
Fumarase Deficiency *FH*†
Galactokinase Deficiency *GALK1*
Galactosemia *GALT*†
Gaucher Disease *GBA*†
Gitelman Syndrome *SLC12A3*
Glucose-6-Phosphate Dehydrogenase Deficiency *G6PD*†*
Glutaric Acidemia *GCDH*†, *ETFDH, ETFA*
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*†
Glycogen Storage Disease, Type IV *GBE1*
Glycogen Storage Disease, Type V *PYGM*
Glycogen Storage Disease, Type VII *PFKM*
GM1 Gangliosidosis *GLB1*†
GRACILE Syndrome *BCS1L*†
Guanidinoacetate Methyltransferase Deficiency *GAMT*
Hemophilia B *F9**
Hepatocerebral Mitochondrial DNA Depletion Syndrome *MPV17*
Hereditary Fructose Intolerance *ALDOB*†
Hereditary Hemochromatosis *HJV*, *TFR2*
Hereditary Spastic Paraparesis *TECPR2*
Hermansky-Pudlak Syndrome *HPS3*†, *HPS1*
Holocarboxylase Synthetase Deficiency *HLCS*
Homocystinuria *CBS*†, *MTRR*
Hydrolethalus Syndrome *HYLS1*
Hypohidrotic Ectodermal Dysplasia *EDA**
Hypophosphatasia *ALPL*†
Inclusion Body Myopathy *GNE*†
Infantile Cerebral and Cerebellar Atrophy *MED17*
Infantile Neuroaxonal Dystrophy *PLA2G6*†
Isovaleric Acidemia *IVD*†
Joubert Syndrome *TMEM216*†
Junctional Epidermolysis Bullosa *LAMA3*†, *LAMB3*†, *LAMC2*†
Juvenile Nephronophthisis *NPHP1*†
Juvenile Retinoschisis *RS1**
Krabbe Disease *GALC*†
Lamellar Ichthyosis *TGM1*†
Leber Congenital Amaurosis *CEP290, LCA5, RDH12, RPE65*
Leigh Syndrome, French-Canadian *LRPPRC*†
Lethal Congenital Contracture Syndrome *GLE1*
Leukoencephalopathy with Vanishing White Matter *EIF2B5*†
Limb-Girdle Muscular Dystrophy *CAPN3*†, *DYSF, FKRP, SGCA*†, *SGCB*†, *SGCG*†
Lipoprotein Lipase Deficiency *LPL*
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency *HADHA*†
Lowe Syndrome *OCRL*†*
Lysinuric Protein Intolerance *SLC7A7*†
Maple Syrup Urine Disease *BCKDHA*†, *BCKDHB*†, *DBT*†
Medium Chain Acyl-CoA Dehydrogenase Deficiency *ACADM*†
Megalencephalic Leukoencephalopathy with Subcortical Cysts *MLC1*†
Metachromatic Leukodystrophy *ARSA*†, *PSAP*
Methylmalonic Aciduria *MUT, MMAA, MMAB, MMACHC*†, *MMADHC*
Microphthalmia/Anopthalmia *VSX2*
Mitochondrial Complex 1 Deficiency *NDUFAF5, NDUFS6*
Mitochondrial Myopathy and Sideroblastic Anemia *PUS1*
MKS1-Related Disorders *MKS1*
Mucolipidosis III gamma *GNPTG*
Mucolipidosis, Type II/III Alpha/Beta *GNPTAB*†
Mucolipidosis, Type IV *MCOLN1*†
Mucopolysaccharidosis, Type I; Hurler Syndrome *IDUA*†
Mucopolysaccharidosis, Type II; Hunter Syndrome *IDS**
Mucopolysaccharidosis, Type IIIA; Sanfilippo A *SGSH*†
Mucopolysaccharidosis, Type IIIB; Sanfilippo B *NAGLU*

Disease / *Gene*
Mucopolysaccharidosis, Type IIIC; Sanfilippo C *HGSNAT*
Mucopolysaccharidosis, Type IIID *GNS*
Mucopolysaccharidosis, Type IX *HYAL1*
Mucopolysaccharidosis, Type VI; Maroteaux-Lamy *ARSB*
Multiple Sulfatase Deficiency *SUMF1*†
Muscle-Eye-Brain Disease, POMGNT1-Related *POMGNT1*†
Myoneurogastrointestinal Encephalopathy *TYMP*
Myotubular Myopathy *MTM1**
N-acetylglutamate Synthase Deficiency *NAGS*
Nemaline Myopathy *NEB*†
Nephrogenic Diabetes Insipidus *AQP2*
Nephrotic Syndrome *NPHS1*†, *NPHS2*†
Neuronal Ceroid Lipofuscinosis *CLN3*†, *CLN5*†, *CLN6*†, *CLN8*†, *MFSD8, PPT1*†, *TPP1*†
Niemann-Pick Disease, Type A/B *SMPD1*†
Niemann-Pick Disease, Type CI/D *NPC1*†
Niemann-Pick Disease, Type CII *NPC2*
Nijmegen Breakage Syndrome *NBN*†
Nonsyndromic Hearing Loss *GJB2*†, *LOXHD1*
Occipital Horn Syndrome *ATP7A**
Odonto-Onycho-Dermal Dysplasia *WNT10A*
Omenn Syndrome *DCLRE1C*†, *RAG2*
Ornithine Aminotransferase Deficiency *OAT*
Ornithine Transcarbamylase Deficiency *OTC*†*
Ornithine Translocase Deficiency/HHH syndrome *SLC25A15*†
Osteopetrosis, Infantile Malignant *TCIRG1*
Pendred Syndrome *SLC26A4*†
Phenylalanine Hydroxylase Deficiency *PAH*†
POLG-Related Disorders *POLG*†
Pontocerebellar Hypoplasia *RARS2, VRK1*
Primary Ciliary Dyskinesia *DNAH5, DNAI1, DNAI2*
Primary Congenital Glaucoma *CYP1B1*†
Primary Hyperoxaluria *AGXT*†, *GRHPR*†, *HOGA1*
Progressive Cerebello-Cerebral Atrophy *SEPSECS*
Progressive Familial Intrahepatic Cholestasis *ABCB11*
PROP1-Related Combined Pituitary Hormone Deficiency *PROP1*†
Propionic Acidemia *PCCA*†, *PCCB*†
Pycnodysostosis *CTSK*†
Pyruvate Dehydrogenase Deficiency *PDHA1**, *PDHB*
Renal Tubular Acidosis and Deafness *ATP6V1B1*
Retinitis Pigmentosa *CERKL, DHDDS*†, *EYS, FAM161A*
Rhizomelic Chondrodysplasia Punctata *PEX7*†, *AGPS*
Riboflavin Responsive Complex 1 Deficiency *ACAD9*
Roberts Syndrome *ESCO2*
Salla Disease *SLC17A5*†
Sandhoff Disease *HEXB*†
Schimke Immunoosseous Dysplasia *SMARCAL1*
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*†
Spondylothoracic Dysostosis *MESP2*
Stuve-Wiedemann Syndrome *LIFR*
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*†, *PEX6, PEX10*

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

† These genes are included on the Preparent Exon Carrier Test of 150+ genes. Genes without this indicator are unique to the Global 280+ gene test.