preparent® exon
CARRIER TEST

exon
150+ genes

diseases tested

WHY TEST FOR THESE DISEASES?

- This test includes 150+ genes associated with hereditary diseases that can cause serious health problems, beginning in infancy or childhood.
- Children born with these diseases often benefit from early diagnosis and treatment.
- Most children born with a genetic disease have no affected family members.

progenity®

WH-48017-01 REV 022020

DISEASES TESTED

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*
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*