preparent®
exon
CARRIER TEST

global
280+ genes

diseases tested

WHY TEST FOR THESE DISEASES?

- This test includes 280+ 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.

DISEASES TESTED

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*
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*

Disease / *Gene*

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*
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*

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