

WOMEN'S HEALTH
genpath
an **OPKO** Health Company



InheriGen Plus Pan-Ethnic Carrier Screen Disease List

CATEGORY LEGEND	
Shortened Life Expectancy	
Cognitive Delay	
InheriGenTx Panel Component	
American College of Medical Genetics (ACMG) Guidelines	
American College of Obstetrics & Gynecology (ACOG) Guidelines	

DISEASE NAME	CATEGORY
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	
Abetalipoproteinemia	
Achalasia-Addisonianism-Alacrima Syndrome	
Achromatopsia, CNGB3 Associated	
Adenosine Deaminase Deficiency	
Adrenal Hyperplasia, Congenital, due to 17-Alpha-Hydroxylase Deficiency	
Agnesis of the Corpus Callosum with Peripheral Neuropathy (Andermann Syndrome)	
Albinism, Oculocutaneous, Type I	
Albinism, Oculocutaneous, Type IV	
Alport Syndrome, Autosomal Recessive	
Antley-Bixler Syndrome (ABS) (Cytochrome P450 Oxidoreductase Deficiency)	
Argininosuccinic Aciduria	
Arthrogryposis, Mental Retardation and Seizures	
Aspartylglucosaminuria (AGU)	
Ataxia Neuropathy Spectrum (ANS)	
Ataxia with Vitamin E Deficiency (AVED)	
Ataxia-Telangiectasia	
Autoimmune Polyglandular Syndrome, Type 1	
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)	
Bardet-Biedl Syndrome 1	
Bardet-Biedl Syndrome 2	
Bardet-Biedl Syndrome 10	
Bardet-Biedl Syndrome 12	
Bernard-Soulier Syndrome, Type A1	
Bernard-Soulier Syndrome, Type C	
Beta Hemoglobinopathies	
Bilateral Frontoparietal Polymicrogyria (BFPP)	
Bloom Syndrome	
Bruck Syndrome-1	
Canavan Disease	
Carnitine Palmitoyltransferase Deficiency, Type 1A	
Carnitine Palmitoyltransferase Deficiency, Type 2	
Carpenter Syndrome	
Cerebrotendinous Xanthomatosis (CTX)	
Ceroid Lipofuscinosis, Neuronal, 1	
Ceroid Lipofuscinosis, Neuronal, 2	
Ceroid Lipofuscinosis, Neuronal, 3	
Ceroid Lipofuscinosis, Neuronal, 5	
Ceroid Lipofuscinosis, Neuronal, 6	
Ceroid Lipofuscinosis, Neuronal, 8	

DISEASE NAME	CATEGORY
Charcot-Marie-Tooth Disease, Type 4D (CMT4D)	
Choroideremia (CHM)	
Ciliary Dyskinesia, Primary 1	
Ciliary Dyskinesia, Primary 3	
Citrin Deficiency	
Coenzyme Q10 Deficiency, Primary, 7	
Cohen Syndrome	
Congenital Amegakaryocytic Thrombocytopenia (CAMT)	
Congenital Disorder of Glycosylation, Type IA	
Congenital Disorder of Glycosylation, Type IB	
Congenital Myasthenic Syndrome, CHRNE-Associated	
Congenital Myasthenic Syndrome, RAPSN-Associated	
CRB1-Associated Retinal Dystrophies	
Crigler-Najjar Syndrome	
Cystic Fibrosis (CF) Expanded	
Cystinosis, Nephropathic	
Deafness, Autosomal Recessive 1A (GJB2)	
Dihydropolipamide Dehydrogenase Deficiency	
Dihydropyrimidine Dehydrogenase Deficiency	
Dyskeratosis Congenita, Autosomal Recessive 5	
Ehlers-Danlos Syndrome, Type VIIC	
Ethylmalonic Encephalopathy	
Factor XI Deficiency (Hemophilia C)	
Familial Dysautonomia	
Familial Hypercholesterolemia, Homozygous, LDLR-Associated	
Familial Hypercholesterolemia, LDLRAP1 Associated	
Familial Hyperinsulinism	
Familial Mediterranean Fever	
Familial Neurohypophyseal Diabetes Insipidus (FNDI), Autosomal Recessive	
Fanconi Anemia, Type C	
Fanconi Anemia, Type G	
Fragile X	
Fructose Intolerance, Hereditary	
Galactosemia	
Gaucher Disease	
Glaucoma 3 Primary Congenital	
Glutaric Acidemia, Type I	
Glutaric Acidemia, Type IIA	
Glutaric Acidemia, Type IIC	
Glycogen Storage Disease, Type IA (GSDIA)	
Glycogen Storage Disease, Type IB (GSDIB)	
Glycogen Storage Disease, Type III (GSDIII)	
Glycogen Storage Disease, Type V (GSDV)	
GRACILE Syndrome	
Hermansky-Pudlak Syndrome 3	
Holocarboxylase Synthetase Deficiency	
Homocystinuria (CBS Deficiency)	
Homocystinuria, cblE Type	

DISEASE NAME	CATEGORY
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome	
Hypophosphatasia	
Inclusion Body Myopathy 2	
Joubert Syndrome 2	
Junctional Epidermolysis Bullosa, LAMA3-Associated	
Junctional Epidermolysis Bullosa, LAMB3-Associated	
Junctional Epidermolysis Bullosa, LAMC2-Associated	
Krabbe Disease	
Lamellar Ichthyosis, Type 1	
Leber Congenital Amaurosis, CEP290-Associated	
Leber Congenital Amaurosis, RDH12-Associated	
Leigh Syndrome, French Canadian Type	
Leukoencephalopathy with Vanishing White Matter	
Limb-Girdle Muscular Dystrophy, Type 2A	
Limb-Girdle Muscular Dystrophy, Type 2C	
Limb-Girdle Muscular Dystrophy, Type 2D	
Limb-Girdle Muscular Dystrophy, Type 2E	
Lipoprotein Lipase Deficiency	
Long-Chain 3-Hydroxyacyl-Coenzyme A Dehydrogenase (LCHAD) Deficiency	
Lysinuric Protein Intolerance	
Maple Syrup Urine Disease, Type 1A	
Maple Syrup Urine Disease, Type 1B	
Meckel-Gruber Syndrome, Type 1	
Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency	
Megalencephalic Leukoencephalopathy with Subcortical Cysts	
Metachromatic Leukodystrophy (MLD)	
Methylmalonic Aciduria and Homocystinuria, Cobalamin C (cblC3)	
Methylmalonic Aciduria, MMAA-Associated (cblA Type)	
Methylmalonic Aciduria, MUT-Associated	
Mitochondrial Complex I Deficiency	
Mitochondrial DNA Depletion Syndrome 6	
Mucopolidosis, Type IV	
Mucopolysaccharidosis Type I (MPS I)	
Multiple Sulfatase Deficiency	
Nemaline Myopathy 2	
Nephrotic Syndrome, Congenital Finnish	
Nephrotic Syndrome, Steroid-Resistant Type 2	
Niemann-Pick Disease, Type A/B	
Niemann-Pick Disease, Type C	
Nijmegen Breakage Syndrome	
Odonto-Onycho-Dermal Dysplasia/Schopf-Schulz-Passarge Syndrome	
Omenn Syndrome	
Osteopetrosis, Autosomal Recessive 1	
Pendred Syndrome	
Peroxisome Biogenesis Disorder 1A (Zellweger)	
Peroxisome Biogenesis Disorder 5A (Zellweger)	
Phenylketonuria (PKU)	

DISEASE NAME	CATEGORY
Phosphoglycerate Dehydrogenase Deficiency	
Polycystic Kidney Disease, Autosomal Recessive (ARPKD)	
Pompe Disease	
Primary Hyperoxaluria, Type I	
Primary Hyperoxaluria, Type II	
Progressive Pseudorheumatoid Dysplasia (PPD)	
Prolidase Deficiency	
Propionic Acidemia, PCCA-Associated	
Propionic Acidemia, PCCB-Associated	
Pseudoxanthoma Elasticum	
Pycnodysostosis	
Pyridoxine-Dependent Epilepsy	
Pyruvate Carboxylase Deficiency	
Retinal Dystrophies, RLBP1-Associated	
Retinitis Pigmentosa 25	
Retinitis Pigmentosa 59	
Retinoschisis 1, X-Linked, Juvenile	
Rhizomelic Chondrodysplasia Punctata, Type 1	
Salla Disease	
Sandhoff Disease	
Sanfilippo, Type A	
Sanfilippo, Type B	
Sanfilippo, Type C	
Segawa Syndrome	
Short/Branched Chain Acyl-CoA Dehydrogenase (SBCAD) Deficiency	
Sialidosis, Type II	
Sjogren-Larsson Syndrome (SLS)	
Smith-Lemli-Opitz Syndrome	
Spastic Tetraplegia, Thin Corpus Callosum & Progressive Microcephaly	
Spinal Muscular Atrophy (SMA)	
Stargardt Disease, Type 1	
Stuve-Wiedemann Syndrome (Schwartz-Jampel Syndrome Type 2)	
Sulfate Transporter-Related Osteochondrodysplasia	
Tay-Sachs Disease	
Tyrosinemia, Type I	
Usher Syndrome, Type 1B	
Usher Syndrome, Type 1C	
Usher Syndrome, Type 1D	
Usher Syndrome, Type 1F	
Usher Syndrome, Type 2A	
Usher Syndrome, Type 3	
Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	
Vitamin D-dependent Rickets, Type I	
Walker-Warburg Syndrome	
Werner Syndrome	
Wilson Disease	
X-Linked Severe Combined Immunodeficiency	