WOMEN'S HEALTH
genpath
an OPKO Health Company

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InheriGen

InheriGen Plus Disease List

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	
Agenesis 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	
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)	
Dihydrolipoamide 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	
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	
Mucolipidosis, 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)	
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	