InheriGen Plus

Pan-Ethnic Carrier Screen Disease List

- 17α-hydroxylase/17,20-lyase Deficiency
- 3-Hydroxy-3-Methylglutaryl CoA lyase deficiency
- 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency
- Abetalipoproteinemia
- Achromatopsia, CNGB3-associated
- Adenosine Deaminase Deficiency
- Agenesis of the Corpus Callosum with Peripheral Neuropathy (Andermann Syndrome)
- Antley-Bixler Syndrome
- Argininosuccinic Aciduria
- Aspartylglycosaminuria
- Ataxia Neuropathy Spectrum (ANS)
- Ataxia with Vitamin E Deficiency
- Ataxia-Telangiectasia
- Autoimmune Polyglandular Syndrome, Type 1
- Autosomal Recessive Polycystic Kidney Disease
- Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)
- Bardet-Biedl Syndrome, BBS10-associated
- Bardet-Biedl syndrome, BBS12-associated
- Bardet-Biedl Syndrome, BBS1-associated
- Bernard-Soulier syndrome (BSS), Type A1
- Bernard-Soulier syndrome (BSS), Type C
- Beta-thalassemia
- Bilateral Frontoparietal Polymicrogyria
- Bloom Syndrome
- Canavan Disease
- Carnitine Palmitoyltransferase Deficiency, Type 1A
- Carnitine Palmitoyltransferase Deficiency, Type 2
- Carpenter syndrome
- Cerebrotendinous xanthomatosis
- Charcot-Marie-Tooth Disease, Type 4D
- Choroideremia
- Citrin Deficiency
- Cohen Syndrome
- Congenital Amegakaryocytic Thrombocytopenia (CAMT)
- Congenital Disorder of Glycosylation, Type Ia
- Congenital Disorder of Glycosylation, Type Ib
- Congenital Finnish Nephrosis
- Congenital Myasthenic Syndrome, CHRNE-associated
- Congenital Myasthenic Syndrome, RAPSN-associated
- CRB1-associated retinal dystrophies
- Crigler-Najjar syndrome
- Cystic Fibrosis
- Cystinosis
- Dihydrolipoamide Dehydrogenase Deficiency
- Dihydropyrimidine dehydrogenase deficiency
- Ethylmalonic Encephalopathy
- Factor XI Deficiency (Hemophilia C)
- Familial Dysautonomia
- Familial Hypercholesterolemia, LDLRAP1-associated
- Familial Hypercholesterolemia, LDLR-associated
- Familial Hyperinsulinism
- Familial Mediterranean Fever
- Familial Neurohypophyseal Diabetes Insipidus (FNDI), Autosomal Recessive
- Fanconi Anemia Group C
- Fanconi Anemia Group G
- Fragile X Syndrome
- Galactosemia
- Gaucher Disease
- Glutaric Acidemia, Type I
- Glutaric Acidemia, Type IIA
- Glutaric Acidemia, Type IIC
- Glycogen Storage Disease, Type Ia
- Glycogen Storage Disease, Type Ib
- Glycogen Storage Disease, Type II (Pompe Disease)
- Glycogen Storage Disease, Type III
- Glycogen Storage Disease, Type V (McArdle Disease)
- GRACILE Syndrome
- Hermansky-Pudlak syndrome
- Holocarboxylase Synthetase Deficiency (Multiple Carboxylase Deficiency)
- Homocystinuria (CBS Deficiency)
- Homocystinuria, cblE type
- Hurler Syndrome (mucopolysaccharidosis type I)
- Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome
- Hypophosphatasia
- Inclusion body myopathy 2
- Joubert syndrome 2
- Junctional Epidermolysis Bullosa, Herlitz, LAMA3-associated
- Junctional Epidermolysis Bullosa, Herlitz, LAMB3-associated
- Junctional Epidermolysis Bullosa, Herlitz, 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 (VWM)

InheriGen Plus

Pan-Ethnic Carrier Screen Disease List

- 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
- Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- Megalencephalic Leukoencephalopathy with Subcortical Cysts
- Metachromatic Leukodystrophy
- Methylmalonic Acidemia, MMAA-associated
- Methylmalonic Acidemia, MUT-associated
- Methylmalonic Aciduria and Homocystinuria, Cobalamin C (cblC) Type
- Mucolipidosis, Type IV
- Navajo Neurohepatopathy
- Nemaline Myopathy, NEB-associated
- Nephrotic Syndrome, Steroid-Resistant Type 2
- Neuronal Ceroid Lipofuscinosis, CLN5-associated
- Neuronal Ceroid Lipofuscinosis, CLN6-associated
- Neuronal Ceroid Lipofuscinosis, CLN8-associated
- Neuronal Ceroid Lipofuscinosis, PPT1-associated
- Neuronal Ceroid Lipofuscinosis, TPP1-associated
- Niemann-Pick Disease, Type A/B
- Niemann-Pick Disease, Type C
- Nijmegen Breakage Syndrome
- Oculocutaneous Albinism, Type 1
- Oculocutaneous Albinism, Type 4
- Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge Syndrome
- Pendred Syndrome
- Phenylketonuria (PKU)
- Primary Ciliary Dyskinesia, DNAH5-associated
- Primary Ciliary Dyskinesia, DNAI1-associated
- Primary Congenital Glaucoma
- Primary Hyperoxaluria, Type 1
- Primary Hyperoxaluria, Type 2
- Progressive Pseudorheumatoid Dysplasia
- 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, EYS-associated
- Rhizomelic Chondrodysplasia Punctata, Type 1
- Salla Disease
- Sandhoff Disease
- Sanfilippo, Type A
- Sanfilippo, Type B
- Sanfilippo, Type C
- Segawa Syndrome
- Severe Combined Immunodeficiency, Athabaskan-type (SCIDA)
- Short/Branched Chain Acyl-CoA Dehydrogenase (SBCAD) Deficiency
- Sialidosis, Type 2
- Sjoegren-Larsson syndrome
- Smith-Lemli-Opitz Syndrome
- Spinal Muscular Atrophy (SMA)
- Stargardt Disease
- Stuve-Wiedemann syndrome (Schwartz-Jampel Syndrome Type 2)
- Sulfate Transporter-Related Osteochondrodysplasias
- Tay-Sachs Disease
- Triple-A syndrome (Allgrove syndrome; Achalasia-Addisonianism-Alacrima)
- Tyrosinemia
- Usher Syndrome, Type IB
- Usher Syndrome, Type IC
- Usher Syndrome, Type ID
- Usher Syndrome, Type IF
- Usher Syndrome, Type II
- Usher Syndrome, Type III
- Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
- Vitamin D-dependent Rickets, Type I
- Walker-Warburg Syndrome
- Werner Syndrome
- Wilson Disease
- X-Linked Juvenile Retinoschisis
- X-Linked Severe Combined Immunodeficiency
- Zellweger Syndrome Spectrum