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)

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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