PRECONCEPTION CARRIER SCREENING CONDITIONS LIST

DISEASE	GENE SYMBOL
3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	*HMGCS2*
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*
3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*
Acyl-CoA Dehydrogenase Deficiency,Medium-Chain	*ACADM*
Acyl-CoA Dehydrogenase Deficiency,Short-Chain	*ACADS*
Acyl-CoA Dehydrogenase Deficiency,Very Long-Chain	*ACADVL*
Adenosine Deaminase Deficiency	*ADA*
Alpha-Mannosidosis	*MAN2B1*
Alpha-thalassemia	*HBA1, HBA2*
Alport syndrome 2, autosomal recessive	*COL4A3, COL4A4*
Argininosuccinic aciduria	*ASL*
Autosomal Recessive Congenital Ichthyosis 1	*TGM1*
Autosomal Recessive Congenital Ichthyosis 4A	*ABCA12*
Autosomal Recessive Congenital Ichthyosis 4B	*ABCA12*
Autosomal Recessive Deafness 1A	*GJB2*
Autosomal Recessive Deafness 4, with Enlarged Vestibular Aqueduct	*SLC26A4*
Autosomal Recessive Osteopetrosis 1	*TCIRG1*
Beta-Ketothiolase Deficiency	*ACAT1*
Beta-thalassemia	*HBB*
Biotinidase Deficiency	*BTD*
Canavan Disease	*ASPA*

Carbamoylphosphate Synthetase I Deficiency	*CPS1*
Citrullinemia	*ASS1*
COACH syndrome	*TMEM67, CC2D2A*
Congenital Disorders of Glycosylation Ia	*PMM2*
Cystic Fibrosis	*CFTR*
Duchenne Muscular Dystrophy	*DMD*
Ellis-van Creveld Syndrome	*EVC2*
Fabry Disease	*GLA*
Familial Hyperinsulinemic Hypoglycemia 2	*KCNJ11*
Familial Hyperinsulinemic Hypoglycemia 4	*HADH*
Fanconi anemia, complementation group A	*FANCA*
Fanconi anemia, complementation group C	*FANCC*
Fanconi anemia, complementation group D2	*FANCD2*
Fanconi anemia, complementation group G	*FANCG*
Fanconi anemia, complementation group I	*FANCI*
Galactosemia	*GALT*
Glutaric Acidemia I	*GCDH*
Glutaric acidemia IIA	*ETFA*
Glutaric acidemia IIB	*ETFB*
Glutaric acidemia IIC	*ETFDH*
Glycine encephalopathy	*AMT, GLDC*
Glycogen Storage Disease Type Ia	*G6PC*
Glycogen Storage Disease Type Ib	*SLC37A4*
Glycogen Storage Disease Type Ic	*SLC37A4*

Glycogen Storage Disease Type II	*GAA*
Glycogen Storage Disease type IV	*GBE1*
Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*
Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*
Hemophagocytic lymphohistiocytosis, familial, 4	*STX11*
Hemophagocytic lymphohistiocytosis, familial, 5	*STXBP2*
Hemophilia B	*F9*
Hermansky-Pudlak Syndrome 1	*HPS1*
Hermansky-Pudlak Syndrome 3	*HPS3*
Holocarboxylase synthetase deficiency	*HLCS*
Homocystinuria Due to Cystathionine Beta-Synthase Deficiency	*CBS*
Homocystinuria-megaloblastic anemia cblE type	*MTRR*
Homocystinuria-Megaloblastic Anemia cblG type	*MTR*
Hurler Syndrome	*IDUA*
Hurler-Scheie Syndrome	*IDUA*
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	*SLC25A15*
Hyperphenylalaninemia, BH4-deficient, A	*PTS*
Hypophosphatasia, childhood	*ALPL*
Hypophosphatasia, infantile	*ALPL*
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*
Isovaleric Acidemia	*IVD*

Joubert Syndrome 17	*C5orf42*
Joubert Syndrome 2	*TMEM216*
Joubert Syndrome 3	*AHI1*
Joubert Syndrome 5	*CEP290*
Joubert Syndrome 6	*TMEM67*
Joubert Syndrome 9	*CC2D2A*
Krabbe Disease	*GALC*
LAMA3-Related Junctional Epidermolysis Bullosa	*LAMA3*
LAMB3-Related Junctional Epidermolysis Bullosa	*LAMB3*
LAMC2-Related Junctional Epidermolysis Bullosa	*LAMC2*
Limb-Girdle Muscular Dystrophy type 2A	*CAPN3*
Limb-Girdle Muscular Dystrophy type 2B	*DYSF*
Limb-Girdle Muscular Dystrophy type 2C	*SGCG*
Limb-Girdle Muscular Dystrophy type 2D	*SGCA*
Maple Syrup Urine Disease Type 1A	*BCKDHA*
Maple Syrup Urine Disease Type 1B	*BCKDHB*
MCEE-Related Methylmalonic Acidemia	*MCEE*
Meckel Syndrome 2	*TMEM216*
Meckel Syndrome 3	*TMEM67*
Meckel Syndrome 4	*CEP290*
Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	*MLC1*
Metachromatic Leukodystrophy due to Arylsulfatase A	*ARSA*
Methylmalonic Aciduria and Homocystinuria cblC type	*MMACHC*
Methylmalonic Aciduria and Homocystinuria cblD type	*MMADHC*

MMAA-Related Methylmalonic Acidemia	*MMAA*
MMAB-Related Methylmalonic Acidemia	*MMAB*
Mucopolysaccharidosis II	*IDS*
Mucopolysaccharidosis Type IIIA	*SGSH*
Mucopolysaccharidosis Type IIIB	*NAGLU*
Mucopolysaccharidosis type IIIC	*HGSNAT*
Mucopolysaccharidosis type IIID	*GNS*
Mucopolysaccharidosis type IVA	*GALNS*
Mucopolysaccharidosis type IVB	*GLB1*
Mucopolysaccharidosis type V	*IDUA*
Mucopolysaccharidosis type VI	*ARSB*
MUT-Related Methylmalonic Acidemia	*MUT*
Nephronophthisis 11	*TMEM67*
Nephronophthisis 3	*NPHP3*
Nephropathic Cystinosis	*CTNS*
Nephrotic syndrome, type 1	*NPHS1*
Netherton syndrome	*SPINK5*
Neuronal Ceroid-Lipofuscinoses 1	*PPT1*
Neuronal Ceroid-Lipofuscinoses 2	*TPP1*
Neuronal Ceroid-Lipofuscinoses 3	*CLN3*
Neuronal Ceroid-Lipofuscinoses 4A	*CLN6*
Neuronal Ceroid-Lipofuscinoses 5	*CLN5*
Neuronal Ceroid-Lipofuscinoses 6	*CLN6*
Neuronal Ceroid-Lipofuscinoses 7	*MFSD8*
Niemann-Pick Disease Type A	*SMPD1*

Niemann-Pick Disease Type B	*SMPD1*
Niemann-Pick Disease Type C1	*NPC1*
Niemann-Pick Disease Type C2	*NPC2*
Oculocutaneous Albinism Type 1	*TYR*
Oculocutaneous Albinism Type 2	*OCA2*
Oculocutaneous Albinism Type 3	*TYRP1*
Oculocutaneous Albinism Type 4	*SLC45A2*
Oculocutaneous Albinism Type 6	*SLC24A5*
Oculocutaneous Albinism Type 7	*C10orf11*
Omenn syndrome	*RAG1, RAG2*
Ornithine Transcarbamylase Deficiency	*OTC*
Osteoporosis-pseudoglioma syndrome	*LRP5*
Peroxisome biogenesis disorder 1A(Zellweger)	*PEX1*
Phenylketonuria	*PAH*
Primary Carnitine Deficiency	*SLC22A5*
Progressive Familial Intrahepatic Cholestasis 2	*ABCB11*
Progressive Familial Intrahepatic Cholestasis 3	*ABCB4*
Progressive Familial Intrahepatic Cholestasis 4	*TJP2*
Propionicacidemia	*PCCA, PCCB*
Severe combined immunodeficiency, B cell-negative	*RAG1, RAG2*
Sickle Cell Anemia	*HBB*
Sitosterolemia	*ABCG5, ABCG8*
Sjögren-Larsson syndrome	*ALDH3A2*
Smith-Lemli-Opitz syndrome	*DHCR7*
Spinal Muscular Atrophy	*SMN1*

Tay-Sachs Disease	*HEXA*
Tyrosinemia Type 1	*FAH*
Wilson Disease	*ATP7B*
Wolfram Syndrome 1	*WFS1*
X-Linked Adrenal Hypoplasia Congenita	*NR0B1*
X-Linked Centronuclear Myopathy	*MTM1*
X-Linked Hypohidrotic Ectodermal Dysplasia	*EDA*
X-Linked Ocular Albinism	*GPR143*
X-Linked Severe Combined Immunodeficiency	*IL2RG*