

GENETIC CONDITIONS



ENDOCRINE

DISEASE <i>Gene</i>	AR	AD	XL
Congenital Adrenal Hyperplasia <i>CYP11B1</i>	●	●	
<i>CYP17A1, HSD3B2, POR, STAR</i>	●		
Congenital Hypothyroidism <i>PAX8, THRA</i>		●	
<i>SLC5A5, TG, TPO, TSHB</i>	●		
<i>TSHR</i>	●	●	
Pendred Syndrome <i>SLC26A4</i>	●		

HEMOGLOBIN

DISEASE <i>Gene</i>	AR	AD	XL
Beta-Thalassemia <i>HBB</i>	●		
S, Beta-Thalassemia (Sickle Cell Beta-Thalassemia) <i>HBB</i>	●		
S,C Disease (Sickle Cell Disease) <i>HBB</i>	●		
S,S Disease (Sickle Cell Disease, Sickle Cell Anemia) <i>HBB</i>	●		

METABOLIC

DISEASE <i>Gene</i>	AR	AD	XL
2-Methyl-3-Hydroxybutyric Aciduria <i>HSD17B10</i>		●	●
2,4 Dienoyl-CoA Reductase Deficiency (NADKD1) <i>NADK2</i>	●		
3-Methylglutaconic Aciduria Type I <i>AUH</i>	●		
β-Ketothiolase Deficiency <i>ACAT1</i>	●		
Argininemia <i>ARG1</i>	●		
Benign Hyperphenylalaninemia <i>PAH</i>	●		
Biotinidase Deficiency <i>BTD</i>	●		
Carnitine Palmitoyltransferase Type I Deficiency <i>CPT1A</i>	●		
Carnitine Uptake Defect/Carnitine Transport Defect <i>SLC22A5</i>	●		
Cerebrotendinous Xanthomatosis <i>CYP27A1, LHX3</i>	●		
Citrullinemia, Type II <i>SLC25A13</i>	●		
Classic Phenylketonuria <i>PAH</i>	●		
Congenital Disorder of Glycosylation 1b <i>MPI</i>	●		
Crigler-Najjar Syndrome <i>UGT1A1</i>	●		
Fabry Disease <i>GLA</i>			●
Galactokinase Deficiency <i>GALK1</i>	●		
Glutaric Acidemia Type I <i>GCDH</i>	●		

HEARING LOSS

DISEASE <i>Gene</i>	AR	AD	XL
Non-Syndromic Hearing Loss			
<i>CDH23, MYO15A, OTOF, TMIE, TMPRSS3, TPRN, TRIOBP</i>	●		
<i>GJB2, GJB6, TECTA</i>	●	●	
Syndromic Hearing Loss			
Jervell and Lange-Nielsen Syndrome <i>KCNE1, KCNQ1</i>	●		
Pendred Syndrome <i>SLC26A4</i>	●		
Shah-Waardenburg Syndrome <i>SOX10</i>		●	
Usher Syndrome Type 1C <i>USH1C</i>	●		
Usher Syndrome 1G <i>USH1G</i>	●		
Usher Syndrome Type 2A <i>USH2A</i>	●		
Usher Syndrome IID <i>DFNB31</i>	●		
Waardenburg Syndrome <i>PAX3</i>	●	●	

DISEASE <i>Gene</i>	AR	AD	XL
2-Methylbutyrylglucosuria <i>ACADSB</i>	●		
3-Methylcrotonyl-CoA Carboxylase Deficiency <i>MCCCI, MCCC2</i>	●		
3-Phosphoglycerate Dehydrogenase Deficiency <i>PHGDH</i>	●		
Abetalipoproteinemia <i>MTTP</i>	●		
Argininosuccinic Aciduria <i>ASL</i>	●		
Biopterin Defect In Cofactor Biosynthesis <i>GCHI</i>	●	●	
Carnitine Acylcarnitine Translocase Deficiency <i>SLC25A20</i>	●		
Carnitine Palmitoyltransferase Type II Deficiency <i>CPT2</i>	●		
Cerebral Creatine Deficiency Syndrome <i>GAMT, GATM</i>	●		
Citrullinemia, Type I <i>ASS1</i>	●		
Classic Galactosemia <i>GALT</i>	●		
Combined Pituitary Hormone Deficiency <i>PROPI</i>	●		
Corticosterone Methyloxidase Deficiency <i>CYP11B2</i>	●		
Cystinosis <i>CTNS</i>	●		
Galactoepimerase Deficiency <i>GALE</i>	●		
Glucose-6-Phosphate Dehydrogenase Deficiency <i>G6PD</i>			●
Glutaric Acidemia Type II <i>ETFA, ETFB, ETFDH</i>	●		

Glycogen Storage Disease Type O <i>GYS2</i>	●		
Glycogen Storage Disease Type Ib <i>SLC37A4</i>	●		
Glycogen Storage Disease IIIa <i>AGL</i>	●		
Hereditary Fructose Intolerance <i>ALDOB</i>	●		
Holocarboxylase Synthase Deficiency <i>HLCS</i>	●		
Hypercholesterolemia <i>LDLR</i>	●	●	
Hypophosphatasia <i>ALPL</i>	●	●	
Isovaleric Acidemia <i>IVD</i>	●		
Lipoprotein Lipase Deficiency (LPL) <i>LPL</i>	●		
Lysinuric Protein Intolerance <i>SLC7A7</i>	●		
Malonic Acidemia <i>MLYCD</i>	●		
Maple Syrup Urine Disease Type III <i>DLD</i>	●		
Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency <i>HADH</i>	●		
Methylmalonic Acidemia with Homocystinuria <i>ABCD4, LMBRD1, MMACHC, MMADHC</i> <i>HCFC1</i>	●		●
Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) <i>MUT</i>	●		
Methylmalonyl-CoA Epimerase Deficiency <i>MCEE</i>	●		
Mucopolysaccharidosis Type II (Hunter Syndrome) <i>IDS</i>			●
Nephrogenic Diabetes Insipidus Type II <i>AQP2</i>	●	●	
Niemann-Pick Disease Type C1 <i>NPC1</i>	●		
Ornithine Translocase Deficiency; Triple H Syndrome <i>SLC25A15</i>	●		
Primary Hyperoxaluria Type II <i>GRHPR</i>	●		
Propionic Acidemia <i>PCCA, PCCB</i>	●		
Transient Infantile Liver Failure <i>TRMU</i>	●		
Tyrosine Hydroxylase Deficiency <i>TH</i>	●		
Tyrosinemia, Type II <i>TAT</i>	●		
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) <i>ACADVL</i>	●		
X-Linked Adrenoleukodystrophy <i>ABCD1</i>			●

Glycogen Storage Disease Ia <i>G6PC</i>	●		
Glycogen Storage Disease Type II (Pompe) <i>GAA</i>	●		
Glycogen Storage Disease VI <i>PYGL</i>	●		
HMG-CoA Lyase Deficiency <i>HMGCL</i>	●		
Homocystinuria <i>CBS</i>	●		
Hypermethioninemia <i>AHCY, GNMT</i> <i>MAT1A</i>	●	●	
Isobutyrylglycinuria <i>ACAD8</i>	●		
Krabbe Disease <i>GALC</i>	●		
Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD Deficiency) <i>HADHA</i>	●		
Lysosomal Acid Lipase Deficiency <i>LIPA</i>	●		
Maple Syrup Urine Disease <i>BCKDHA, BCKDHB, DBT</i>	●		
Medium-Chain Acyl-CoA Dehydrogenase Deficiency <i>ACADM</i>	●		
Metachromatic Leukodystrophy <i>ARSA</i>	●		
Methylmalonic Acidemia (Cobalamin Disorders) <i>MMAA, MMAB</i>	●		
Methylmalonic Aciduria and Homocystinuria <i>MTR, MTRR</i>	●		
Mucopolysaccharidosis Type I <i>IDUA</i>	●		
N-Acetylglutamate Synthase Deficiency <i>NAGS</i>	●		
Niemann-Pick Disease Type A/B <i>SMPD1</i>	●		
Ornithine Transcarbamylase Deficiency <i>OTC</i>			●
Primary Hyperoxaluria Type I <i>AGXT</i>	●		
Primary Hyperoxaluria Type III <i>HOGA1</i>	●	●	
Short-Chain Acyl-CoA Dehydrogenase Deficiency <i>ACADS</i>	●		
Trifunctional Protein Deficiency <i>HADHA, HADHB</i>	●		
Tyrosinemia, Type I <i>FAH</i>	●		
Tyrosinemia, Type III <i>HPD</i>	●	●	
Wilson Disease <i>ATP7B</i>	●		

OTHER – Genetic, Immunodeficiency, Pulmonary, Musculoskeletal

DISEASE <i>Gene</i>	AR	AD	XL
Cystic Fibrosis <i>CFTR</i>	●		
Spinal Muscular Atrophy due to homozygous deletion of exon 7 & 8 in SMN1 <i>SMN1, SMN2</i>	●		

DISEASE <i>Gene</i>	AR	AD	XL
Severe Combined Immunodeficiencies <i>ADA, IL7R, JAK3</i> <i>IL2RG</i>	●		●
T-cell Related Lymphocyte Deficiencies <i>PIK3CD</i>		●	

● AR: AUTOSOMAL RECESSIVE

● AD: AUTOSOMAL DOMINANT

● XL: X-LINKED