

GENE	ASSOCIATED CONDITIONS
<b>ABCD1</b>	Adrenoleukodystrophy
<b>ABCD4</b>	cbIJ Type Methylmalonic aciduria and homocystinuria
<b>ACAD8</b>	Isobutyryl-CoA dehydrogenase deficiency
<b>ACADM</b>	MCAD DEFICIENCY
<b>ACADS</b>	SCAD Deficiency
<b>ACADSB</b>	2-methylbutyryl-CoA Dehydrogenase Deficiency
<b>ACADVL</b>	VLCAD deficiency
<b>ACAT1</b>	Alpha-methylacetoacetic aciduria (3-ketothialase deficiency)
<b>ACSF3</b>	Combined malonic and methylmalonic aciduria
<b>ADA</b>	Partial adenosine deaminase deficiency Severe combined immunodeficiency (SCID) due to adenosine deaminase deficiency (ADAD)
<b>AHCY</b>	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
<b>ARG1</b>	Argininemia (Arginase Deficiency)
<b>ASL</b>	Arginosuccinic Aciduria
<b>ASPA</b>	Canavan disease
<b>ASS1</b>	Citrullinemia
<b>AUH</b>	3-methylglutaconic aciduria type I (MCGA1)
<b>BCKDHA</b>	Maple syrup urine disease, type Ia
<b>BCKDHB</b>	Maple syrup urine disease, type Ib
<b>BLM</b>	Bloom syndrome
<b>BTD</b>	Biotinidase deficiency
<b>CBS</b>	Homocystinuria, B6-responsive and nonresponsive types Hyperhomocysteinemic thrombosis
<b>CD320</b>	Methylmalonic aciduria due to transcobalamin receptor defect
<b>CFTR</b>	Congenital bilateral absence of the vas deferens (CVAD) Cystic fibrosis Neonatal hypertrypsinemia
<b>CPT1A</b>	Hepatic carnitine palmitoyl transferase deficiency Type I
<b>CPT2</b>	CPT2 deficiency associated myopathy Hepatic carnitine palmitoyl transferase deficiency Type II Lethal neonatal CPT2 deficiency Susceptibility to acute-infection induced encephalopathy
<b>CYP21A2</b>	Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency Non-classic hyperandrogenism due to 21-hydroxylase deficiency
<b>DBT</b>	Maple syrup urine disease type II
<b>DLD</b>	Dihydrolipoamide dehydrogenase deficiency
<b>DNAJC19</b>	3-methylglutaconic aciduria, type V
<b>DUOX2</b>	Thyroid dysmorphogenesis 6
<b>EFTA</b>	Glutaric acidemia IIA
<b>ETFB</b>	Glutaric acidemia IIB

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<b>EFTDH</b>	Glutaric acidemia IIC
<b>FAH</b>	Tyrosinemia, type I
<b>FANCC</b>	Fanconi anemia
<b>G6PC</b>	Glycogen storage disease Ia
<b>G6PD</b>	Favism Hemolytic anemia due to G6PD deficiency
<b>GAA</b>	Glycogen storage disease II
<b>GALC</b>	Krabbe disease
<b>GALE</b>	Galactose epimerase deficiency
<b>GALK1</b>	Galactokinase deficiency with cataracts
<b>GALT</b>	Galactosemia
<b>GBA</b>	Gaucher disease Type I Gaucher disease Type II Gaucher disease Type III Gaucher disease Type IIIC Perinatal lethal Gaucher disease
<b>GCDH</b>	Glutaric aciduria Type I
<b>GCH1</b>	BH4-deficient Hyperphenylalaninemia B DOPA-responsive dystonia (with or without hyperphenylalaninemia)
<b>GJB2</b>	Autosomal dominant deafness Type 3A Autosomal recessive deafness Type 1A Bart-Pumphrey Syndrome Hystrix-like ichthyosis with deafness Keratitis ichthyosis deafness syndrome Palmoplantar keratoderma with deafness Vohwinkel syndrome
<b>GJB3</b>	Autosomal dominant deafness Type IIB Autosomal recessive deafness Digenic deafness GJB2/GJB3 Erythrokeratoderma variabilis et progressiva
<b>GJB6</b>	Autosomal dominant deafness Type IIIB Autosomal recessive deafness Type IB Clouston type ectodermal dysplasia Type II Digenic GJB2/GJB6 deafness
<b>GLA</b>	Fabry Disease
<b>GNMT</b>	Glycine N-methyltransferase deficiency
<b>HADH</b>	3-hydroxyacyl-CoA dehydrogenase deficiency Familial hyperinsulinemic hypoglycemia type 4
<b>HADHA</b>	LCHAD deficiency Trifunctional protein deficiency
<b>HADHB</b>	Trifunctional protein deficiency
<b>HBA1</b>	Alpha thalassemia Hb Barts Hb H (3 gene deletion) Hb H/Constant Spring disease
<b>HBA2</b>	Alpha thalassemia Hb Barts Hb H (3 gene deletion) Hb H/Constant Spring disease

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<b>HBB</b>	Beta thalassemia major Hb C disease (Hb CC) Hb D disease (Hb DD) Hb E/Beta+ thalassemia Hb C/Beta0 thalassemia Hb C/Beta+ thalassemia Hb D/Beta0 thalassemia Hb D/Beta+ thalassemia Hb E/Beta0 thalassemia Hb EE Hb Variant/ Beta0 thalassemia Hb Variant/Beta+ thalassemia Hb Variants Hb S/Beta0 thalassemia Hb S/Beta + thalassemia Hereditary persistence of fetal hemoglobin Sickle cell anemia (S/S) Sickle cell disease variants Sickle hemoglobin C disease Sickle hemoglobin D disease Sickle hemoglobin E disease
<b>HCFC1</b>	X-linked mental retardation with methylmalonic acidemia and homocysteinemia
<b>HEXA</b>	GM2-gangliosidosis Hex A pseudodeficiency Tay-Sachs disease
<b>HLCS</b>	Holocarboxylase synthetase deficiency
<b>HMGCL</b>	HMG-CoA Lyase Deficiency
<b>HPD</b>	Hawkinsinuria Tyrosinemia, type III
<b>HSD17B10</b>	17-beta-hydroxysteroid dehydrogenase X deficiency Mental retardation X-linked syndromic 10 (MRXS10)
<b>IDUA</b>	Mucopolysaccharidosis Ih Mucopolysaccharidosis Ih/s Mucopolysaccharidosis Is
<b>IKBKAP</b>	Familial dysautonomia
<b>IL2RG</b>	X-linked severe combined immunodeficiency (SCID)
<b>IVD</b>	Isovaleric acidemia
<b>LMBRD1</b>	Methylmalonic acidemia with homocystinuria (Gobbledegook)
<b>MAT1A</b>	Autosomal dominant persistent hypermethioninemia due to methionine adenosyltransferase I/III deficiency
<b>MCCC1</b>	3-Methylcrotonyl-CoA carboxylase 1 deficiency (MCC1D)
<b>MCCC2</b>	3-Methylcrotonyl-CoA carboxylase 2 deficiency (MCC2D)
<b>MCEE</b>	Methylmalonyl-CoA epimerase deficiency
<b>MCOLN1</b>	Mucopolipidosis IV
<b>MLYCD</b>	Malonyl-CoA decarboxylase deficiency
<b>MMAA</b>	Vitamin B-12 responsive methylmalonic aciduria MMAA
<b>MMAB</b>	cbIB complement type Vitamin B-12 responsive methylmalonic aciduria (due to defect in synthesis of adenosylcobalamin)
<b>MMADHC</b>	cbID complement type homocystinuria (Variant 1) cbID complement type homocystinuria (Variant 2) cbID complement type Methylmalonic aciduria and homocystinuria (Variant 1)
<b>MMACHC</b>	Methylmalonic aciduria and homocystinuria, cbIC type

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<b>MTHFR</b>	Homocystinuria due to MTHFR deficiency
<b>MTR</b>	cbI G complementation type homocystinuria-megaloblastic anemia
<b>MTRR</b>	cbI E complementation type homocystinuria-megaloblastic anemia
<b>MUT</b>	Methylmalonic Aciduria due to Methylmalonyl-CoA Mutase deficiency
<b>NPC1</b>	Niemann-Pick disease, type C1 Niemann-Pick disease, type D
<b>NPC2</b>	Niemann-pick disease, type C2
<b>OPA3</b>	C3-methylglutaconic aciduria, type III Optic atrophy 3 with cataract
<b>OTC</b>	Ornithine transcarbamylase deficiency
<b>PAH</b>	Non-PKU hyperphenylalanemia Phenylketonuria
<b>PAX8</b>	Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia
<b>PCBD1</b>	BH4-deficient Hyperphenylalaninemia D
<b>PCCA</b>	Propionicacidemia
<b>PCCB</b>	Propionicacidemia
<b>PTS</b>	BH4-deficient Hyperphenylalaninemia A
<b>QDPR</b>	BH4-deficient Hyperphenylalaninemia C
<b>RECQL3</b>	Bloom Syndrome
<b>SLC5A5</b>	Thyroid dysmorphogenesis 1
<b>SLC22A5</b>	Systemic primary carnitine deficiency
<b>SLC25A13</b>	Adult-onset citrullinemia Type II Neonatal onset citrullinemia Type II
<b>SLC25A20</b>	Carnitine-acylcarnitine translocase (CACT) deficiency
<b>SLC26A4</b>	Autosomal recessive deafness type IV Pendred syndrome
<b>SMN1</b>	Spinal muscular atrophy
<b>SMPD1</b>	Niemann-Pick disease, type A Niemann-Pick disease, type B
<b>TAT</b>	Tyrosinemia, type II
<b>TAZ</b>	Barth Syndrome Familial dilated cardiomyopathy
<b>TCN2</b>	Transcobalamin II deficiency
<b>TG</b>	Thyroid dysmorphogenesis 3 Susceptibility to autoimmune thyroid disease Type III
<b>THRA</b>	Congenital nongoitrous hypothyroidism 6
<b>THRB</b>	Thyroid hormone resistance
<b>TPO</b>	Thyroid dysmorphogenesis 2A
<b>TSHB</b>	Congenital nongoitrous hypothyroidism 4
<b>TSHR</b>	Congenital nongoitrous hypothyroidism 1 Familial gestational hyperthyroidism Nonautoimmune hyperthyroidism
<b>TXNB</b>	Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency