CONDITION	OMIM	GENE	
● 17-beta-hydroxysteroid dehydrogenase X deficiency	300438	HSD17B10	XL
● 2-methylbutyrylglycinuria	610006	ACADSB	AR
● 3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200	MCCC1	AR
● 3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210	MCCC2	AR
● Aarskog-Scott syndrome; Mental retardation, X-linked 16	305400	FGD1	XL
● Achondrogenesis Ib	600972	SLC26A2	AR
● Achromatopsia-3	262300	CNGB3	AR
● Acyl-CoA dehydrogenase, medium chain, deficiency of	201450	ACADM	AR
● Acyl-CoA dehydrogenase, short-chain, deficiency of	201470	ACADS	AR
● Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	CYP11B1	AR
● Adrenal hyperplasia, congenital, due to 17-alpha-hydroxylase deficiency	202110	CYP17A1	AR
● Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	201910	CYP21A2	AR
● Adrenoleukodystrophy	300100	ABCD1	XL
● Alkaptonuria	203500	HGD	AR
● Allan-Herndon-Dudley syndrome	300523	SLC16A2	XL
● Alpha-methylacetoacetic aciduria	203750	ACAT1	AR
● Alpha-thalassemia/mental retardation syndrome	301040	ATRX	XL
● Alport syndrome, autosomal recessive	203780	COL4A4	AR
● Anauxetic dysplasia	607095	RMRP	AR
● Androgen insensitivity	300068	AR	XL
● Argininemia	207800	ARG1	AR
● Argininosuccinic aciduria	207900	ASL	AR
● Arts syndrome	301835	PRPS1	XL
● Aspartylglucosaminuria	208400	AGA	AR
● Ataxia with isolated vitamin E deficiency	277460	TTPA	AR
● Ataxia-telangiectasia	208900	ATM	AR
● Auditory neuropathy, autosomal recessive, 1	601071	OTOF	AR
● Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	240300	AIRE	AR
● Bardet-Biedl syndrome 1	209900	BBS1	AR
● Bardet-Biedl syndrome 10	615987	BBS10	AR
● Bardet-Biedl syndrome 14; Joubert syndrome 5; Meckel syndrome 4; Senior-Loken syndrome 6	615991; 610188; 611134; 610189	CEP290	AR
● Bardet-Biedl syndrome 2	615981	BBS2	AR
● Bartter syndrome, type 4a	602522	BSND	AR
● Duchenne muscular dystrophy; Becker muscular dystrophy	310200; 300376	DMD	XL
● Biotinidase deficiency	253260	BTD	AR
● Bjornstad syndrome	262000	BCS1L	AR
● Canavan disease	271900	ASPA	AR
● Carbamoylphosphate synthetase I deficiency	237300	CPS1	AR
● Carnitine deficiency, systemic primary	212140	SLC22A5	AR
● Carnitine-acylcarnitine translocase deficiency	212138	SLC25A20	AR
● Cerebral creatine deficiency syndrome 1	300352	SLC6A8	XL
● Cerebrotendinous xanthomatosis	213700	CYP27A1	AR
● Ceroid lipofuscinosis, neuronal, 1	256730	PPT1	AR
● Ceroid lipofuscinosis, neuronal, 10	610127	CTSD	AR
● Ceroid lipofuscinosis, neuronal, 2	204500	TPP1	AR
● Ceroid lipofuscinosis, neuronal, 3	204200	CLN3	AR
● Ceroid lipofuscinosis, neuronal, 5	256731	CLN5	AR
● Ceroid lipofuscinosis, neuronal, 6	601780	CLN6	AR
● Ceroid lipofuscinosis, neuronal, 7	610951	MFSD8	AR
● Ceroid lipofuscinosis, neuronal, 8	600143	CLN8	AR
● Charcot-Marie-Tooth disease, type 4B1	601382	MTMR2	AR
● Charcot-Marie-Tooth disease, type 4C	601596	SH3TC2	AR
● Charcot-Marie-Tooth disease, type 4D	601455	NDRG1	AR
● Charcot-Marie-Tooth Neuropathy Type 4A	214400	GDAP1	AR
● Cholestasis, benign recurrent intrahepatic, 2	601847	ABCB11	AR
● Chondrodysplasia punctata, X-linked recessive	302950	ARSF	XL
● Chondrodysplasia punctata, X-linked recessive	302950	ARSE	XL
● Citrullinemia	215700	ASS1	AR
● Citrullinemia, adult-onset type II; type II, neonatal-onset	603471; 605814	SLC25A13	AR
● Coffin-Lowry syndrome	303600	RPS6KA3	XL
● Combined malonic and methylmalonic acidemia	614265	ACSF3	AR
● Cone-rod dystrophy 3	604116	ABCA4	AR
● Cone-rod dystrophy, 604393 (Congenital Leber Amaurosis, 4)	604393	AIPL1	AR
● Cone-rod dystrophy, X-linked, 1	304020	RPGR	XL
● Congenital disorder of glycosylation, type Ia	212065	PMM2	AR
● Corneal dystrophy, Fuchs endothelial, 4	613268	SLC4A11	AR
● CPT deficiency, hepatic, type IA	255120	CPT1A	AR
● CPT II deficiency, lethal neonatal	608836	CPT2	AR
● Cystathioninuria	219500	CTH	AR
● Cystic Fibrosis; Congenital bilateral absence of vas deferens	219700; 277180	CFTR	AR
● Cystinosis, atypical nephropathic	219800	CTNS	AR
● Cystinuria	220100	SLC3A1	AR
● Cystinuria	220100	SLC7A9	AR
● Deafness, autosomal recessive 12	601386	CDH23	AR
● Deafness, autosomal recessive 18A	602092	USH1C	AR
● Deafness, autosoma recessive 1A; DFNB1A	220290	GJB2	AR
● Deafness, autosomal recessive 23	609533	PCDH15	AR
● Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	600791	SLC26A4	AR
● Deafness, digenic, GJB2/GJB3	220290	GJB3	AR, DD
● Deafness, X-linked 2	304400	POU3F4	XL
● Dent disease 2	300555	OCRL	XL
O Dihydrolipoamide dehydrogenase deficiency	246900	DLD	AR
● Dysprothrombinemia	613679	F2	AR
● EBD inversa	226600	COL7A1	AR
● Ehlers-Danlos syndrome, type VI	225400	PLOD1	AR
● Ellis-van Creveld syndrome	225500	EVC2	AR
● Emphysema due to AAT deficiency	613490	SERPINA1	AR
● Epidermolysis bullosa, junctional, Herlitz type; non-Herlitz type	226700; 226650	LAMB3	AR
● Epidermolysis bullosa, junctional, Herlitz type; non-Herlitz type	226700; 226650	LAMC2	AR
● Epilepsy, X-linked, with variable learning disabilities and behavior disorders	300491	SYN1	XL
● Epileptic encephalopathy, early infantile, 1	308350	ARX	XL
● Ethylmalonic encephalopathy	602473	ETHE1	AR
● Fabry disease	301500	GLA	XL
● Factor V deficiency	227400	F5	AR

	CONDITION	OMIM	GENE	
●	Factor XI deficiency, autosomal dominant & recessive	612416	F11	AR, AD
●	Familial Mediterranean fever, AR	249100	MEFV	AR
●	Fanconi anemia	227650	FANCA	AR
●	Fanconi anemia, complementation group C	227645	FANCC	AR
●	Favism	134700	G6PD	XL
●	Folate malabsorption, hereditary	229050	SLC46A1	AR
O	Fragile X syndrome	300624	FMR1	XL
O	Friedreich ataxia with retained reflexes	229300	FXN	AR
●	Fructose intolerance	229600	ALDOB	AR
●	Fumarase deficiency	606812	FH	AR
●	Galactokinase deficiency with cataracts	230200	GALK1	AR
●	Galactose epimerase deficiency	230350	GALE	AR
●	Galactosemia	230400	GALT	AR
●	Gaucher disease, perinatal lethal	608013	GBA	AR
●	Glutamate formiminotransferase deficiency	229100	FTCD	AR
●	Glutaric acidemia IIA	231680	ETFA	AR
●	Glutaric acidemia IIB	231680	ETFB	AR
●	Glutaric acidemia IIC	231680	ETFDH	AR
●	Glutaricaciduria, type I	231670	GCDH	AR
●	Glycine encephalopathy	605899	GLDC	AR
●	Glycine encephalopathy	605899	AMT	AR
●	Glycine N-methyltransferase deficiency	606664	GNMT	AR
●	Glycogen storage disease Ia	232200	G6PC	AR
●	Glycogen storage disease Ib	232220	SLC37A4	AR
●	Glycogen storage disease II / Pompe Disease	232300	GAA	AR
●	Glycogen storage disease IIIa	232400	AGL	AR
●	Glycogen storage disease IV	232500	GBE1	AR
●	GM1-gangliosidosis, types I, II and III	230500; 230600; 230650	GLB1	AR
●	Goldmann-Favre syndrome	268100	NR2E3	AR
●	HARP syndrome	607236	PANK2	AR
●	Hartnup disorder	234500	SLC6A19	AR
●	Heimler syndrome, type 2	616617	PEX6	AR
●	Hemochromatosis, type 3	604250	TFR2	AR
●	Hemochromatosis: Type 2A; HFE2 Related	602390	HFE2	AR
●	Hemophilia A	306700	F8	XL
●	Hemophilia B	306900	F9	XL
●	Histidinemia	235800	HAL	AR
●	HMG-CoA lyase deficiency	246450	HMGCL	AR
●	Holocarboxylase synthetase deficiency	253270	HLCS	AR
●	Homocystinuria due to MTHFR deficiency	236250	MTHFR	AR
●	Homocystinuria-megaloblastic anemia, cbl E type	236270	MTRR	AR
●	Homocystinuria, B6-responsive and nonresponsive types	236200	CBS	AR
●	Homocystinuria, cblD type, variant 1 / Methylmalonic aciduria and homocystinuria, cblD type / Methylmalonic aciduria, cblD type, variant 2	277410; 277410; 611935	MMADHC	AR
●	Hyper-IgD syndrome; Mevalonic aciduria	260920; 610377	MVK	AR
●	Hypercholesterolemia, familial	143890	LDLR	AR, CD
●	Hypercholesterolemia, familial, autosomal recessive	603813	LDLRAP1	AR
●	Hyperinsulinemic hypoglycemia, familial, Type 2	601820	KCNJ11	AD, AR
●	Hypermethioninemia due to adenosine kinase deficiency	614300	ADK	AR
●	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	613752	AHCY	AR
●	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	250850	MAT1A	AR
●	Hyperoxaluria, primary, type 1	259900	AGXT	AR
●	Hyperoxaluria, primary, type II	260000	GRHPR	AR
●	Hyperoxaluria, primary, type III	613616	HOGA1	AR
●	Hyperphenylalaninemia, BH4-deficient, A	261640	PTS	AR
●	Hyperphenylalaninemia, BH4-deficient, C	261630	QDPR	AR
●	Hyperphenylalaninemia, BH4-deficient, D	264070	PCBD1	AR
●	Hyperprolinemia, type II	239510	ALDH4A1	AR
●	Hyperthyroidism, familial gestational; Hypothyroidism, congenital, nongoitrous, 1	603373; 275200	TSHR	AD, AR
●	Hypogonadotropic hypogonadism 7 without anosmia	146110	GNRHR	AR
●	Hypothryoidism, congenital, nongoitrous 4	275100	TSHB	AR
●	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	PAX8	AD
●	Ichthyosis, congenital, autosomal recessive 1	242300	TGM1	AR
●	Immunodeficiency, X-linked, with hyper-IgM	308230	CD40LG	XL
●	Isovaleric acidemia	243500	IVD	AR
●	Joubert syndrome 2	608091	TMEM216	AR
●	Joubert syndrome 3	608629	AHI1	AR
●	Joubert syndrome 4	609583	NPHP1	AR
●	Joubert syndrome 8	612291	ARL13B	AR
●	Krabbe disease	245200	GALC	AR
●	LCHAD deficiency	609016	HADHA	AR
●	Leber congenital amaurosis	204000	GUCY2D	AR
●	Leber congenital amaurosis 13	612712	RDH12	AR
●	Leber congenital amaurosis 2	204100	RPE65	AR
●	Leber congenital amaurosis 8	613835	CRB1	AR
●	Leigh syndrome, due to COX deficiency	256000	SURF1	AR, Mit
●	Leigh syndrome, French-Canadian type	220111	LRPPRC	AR
●	limb-girdle muscular dystrophy type 2B	253601	DYSF	AR
●	Lipoid adrenal hyperplasia	201710	STAR	AR
●	Lissencephaly, X-linked	300067	DCX	XL
●	Macular corneal dystrophy	217800	CHST6	AR
●	Malonyl-CoA decarboxylase deficiency	248360	MLYCD	AR
●	Mannosidosis, alpha-, types I and II	248500	MAN2B1	AR
●	Maple syrup urine disease, type Ia	248600	BCKDHA	AR
●	Maple syrup urine disease, type Ib	248600	BCKDHB	AR
●	Maple syrup urine disease, type II	248600	DBT	AR
●	MASA syndrome / CRASH syndrome	303350	L1CAM	XL
●	McArdle disease	232600	PYGM	AR
●	Meckel syndrome 1	249000	MKS1	AR
●	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	300749	CASK	XL
●	Mental retardation syndrome, X-linked, Siderius type	300263	PHF8	XL
●	Mental retardation, X-linked	300486	OPHN1	XL
●	Mental retardation, X-linked 1/78	309530	IQSEC2	XL
●	Mental retardation, X-linked 12/35	300957	THOC2	XL
●	Mental retardation, X-linked 21/34	300143	IL1RAPL1	XL
●	Mental retardation, X-linked 30/47	300558	PAK3	XL
●	Mental retardation, X-linked 41	300849	GDI1	XL
●	Mental retardation, X-linked 58	300210	TSPAN7	XL
●	Mental retardation, X-linked 63	300387	ACSL4	XL
●	Mental retardation, X-linked 9	309549	FTSJ1	XL
●	Mental retardation, X-linked 90	300850	DLG3	XL
●	Mental retardation, X-linked 94	300699	GRIA3	XL
●	Mental retardation, X-linked 97	300803	ZNF711	XL
●	Mental retardation, X-linked 99	300919	USP9X	XL
●	Mental retardation, X-linked syndromic 5	304340	AP1S2	XL
●	Mental retardation, X-linked syndromic, Raymond type	300799	ZDHHC9	XL
●	Mental retardation, X-linked syndromic, Turner type	300706	HUWE1	XL
●	Mental retardation, X-linked, Asperger syndrome susceptibility, X-linked	300427	NLGN4X	XL
●	Mental retardation, X-linked, FRAXE type	309548	AFF2	XL
●	Mental retardation, X-linked, syndromic 13	300055	MECP2	XL
●	Mental retardation, X-linked, syndromic 14	300676	UPF3B	XL
●	Mental retardation, X-linked, syndromic 15	300354	CUL4B	XL
●	Mental retardation, X-linked, syndromic, Claes-Jensen type	300534	KDM5C	XL
●	Metachromatic leukodystrophy	250100	ARSA	AR
●	Methylmalonic aciduria and homocystinuria, cblC type	277400	MMACHC	AR
●	Methylmalonic aciduria and homocystinuria, cblF type	277380	LMBRD1	AR
●	Methylmalonic aciduria and homocystinuria, cblJ type	614857	ABCD4	AR
●	Methylmalonic aciduria, mut(0) type	251000	MUT	AR
●	Methylmalonic aciduria, vitamin B12-responsive	251100	MMAA	AR

	CONDITION	OMIM	GENE	
●	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	251110	MMAB	AR
●	Methylmalonyl-CoA epimerase deficiency	251120	MCEE	AR
●	Microphthalmia, isolated 3	611038	RAX	AR
●	Mucolipidosis II and III, alpha/beta	252500; 252600	GNPTAB	AR
●	Mucolipidosis IV	252650	MCOLN1	AR
●	Mucopolysaccharidisis type IIIA (Sanfilippo A)	252900	SGSH	AR
●	Mucopolysaccharidosis Ih	607014	IDUA	AR
●	Mucopolysaccharidosis II	309900	IDS	XL
●	Mucopolysaccharidosis IVA	253000	GALNS	AR
●	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	NAGLU	AR
●	Mucopolysaccharidosis type IIIC (Sanfilippo C)	252930	HGSNAT	AR
●	Mucopolysaccharidosis type IIID	252940	GNS	AR
●	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200	ARSB	AR
●	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	236670	POMT1	AR
●	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	613150	POMT2	AR
●	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	253280	POMGNT1	AR
●	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	613153	FKRP	AR
●	Muscular dystrophy, limb-girdle, type 2A	253600	CAPN3	AR
●	Muscular dystrophy, limb-girdle, type 2D	608099	SGCA	AR
●	Muscular dystrophy, limb-girdle, type 2E	604286	SGCB	AR
●	Myotonia congenita, dominant; recessive	160800; 255700	CLCN1	AR, AD
O	Nemaline myopathy 2, autosomal recessive	256030	NEB	AR
●	Nephrotic syndrome, type 1	256300	NPHS1	AR
●	Neutropenia, severe congenital 3, autosomal recessive	610738	HAX1	AR
●	Niemann-Pick disease, type A	257200	SMPD1	AR
●	Niemann-Pick disease, type C1	257220	NPC1	AR
●	Niemann-pick disease, type C2	607625	NPC2	AR
●	Nijmegen Breakage Syndrome (Ataxia telangectasia, type 1)	251260	NBN	AR
●	Norrie disease	310600	NDP	XL
●	Nystagmus 6, congenital, X-linked	300814	GPR143	XL
●	Ornithine transcarbamylase deficiency	311250	OTC	XL
●	Osteogenesis imperfecta, type VIII	610915	LEPRE1	AR
●	Pelizaeus-Merzbacher disease	312080	PLP1	XL
●	Peroxisomal acyl-CoA oxidase deficiency	264470	ACOX1	AR
●	Peroxisome biogenesis disorder 1A (Zellweger)	214100	PEX1	AR
●	Peroxisome biogenesis disorder 6A (Zellweger)	614870	PEX10	AR
●	Peroxisome biogenesis disorder 9B; Rhizomelic chondroplasia punctata, type I	614879; 215100	PEX7	AR
●	Phenylketonuria	261600	PAH	AR
●	Phosphoglycerate kinase 1 deficiency	300653	PGK1	XL
●	Pituitary hormone deficiency, combined, 2	262600	PROP1	AR
●	Polycystic kidney and hepatic disease	263200	PKHD1	AR
●	Primary ciliary dyskinesia	608644	DNAH5	AR
●	Propionic acidemia	606054	PCCA	AR
●	Propionic acidemia	606054	PCCB	AR
●	Pyruvate carboxylase deficiency	266150	PC	AR
●	Pyruvate dehydrogenase E1-beta deficiency	614111	PDHB	AR
●	Renpenning syndrome	309500	PQBP1	XL
●	Retinitis pigmentosa 2	312600	RP2	XL
●	Retinitis pigmentosa 25	602772	EYS	AR
●	Retinitis pigmentosa 26	608380	CERKL	AR
●	Retinitis pigmentosa 39	613809	USH2A	AR
●	Retinitis pigmentosa 43	613810	PDE6A	AR
●	Retinitis pigmentosa 45	613767	CNGB1	AR
●	Retinitis pigmentosa 46	612572	IDH3B	AR
●	Retinitis pigmentosa 49	613756	CNGA1	AR
●	Retinitis pigmentosa 59	613861	DHDDS	AR
●	Retinoschisis	312700	RS1	XL
●	Rhizomelic chondrodysplasia punctata, type 3	600121	AGPS	AR
●	Sandhoff disease, infantile, juvenile, and adult forms	268800	HEXB	AR
●	SCID, autosomal recessive, T-negative/B-positive type	600802	JAK3	AR
●	Segawa syndrome, recessive	605407	TH	AR
●	Severe combined immunodeficiency due to ADA deficiency	102700	ADA	AR
●	Severe combined immunodeficiency, X-linked	300400	IL2RG	XL
●	Shwachman-Bodian-Diamond syndrome	260400	SBDS	AR
●	Smith-Lemli-Opitz syndrome	270400	DHCR7	AR
●	Spastic ataxia, Charlevoix-Saguenay type	270550	SACS	AR
●	Spastic paraplegia 11, autosomal recessive	604360	SPG11	AR
●	Spastic paraplegia 7, autosomal recessive	607259	SPG7	AR
●	Spinal muscular atrophy, type I	253300	SMN1	AR
●	Tay-Sachs; GM2-gangliosidosis, several forms	272800	HEXA	AR
●	Thalassemia, beta	613985	HBB	AR
●	Thalassemias, alpha-	604131	HBA1	AR, AD
●	Thalassemias, alpha-	604131	HBA2	AR, AD
●	Thrombocytopenia, congenital amegakaryocytic	604498	MPL	AR
●	Thryoid dyshormonogenesis 6	607200	DUOX2	AR
●	Thyroid dyshormonogenesis 1	274400	SLC5A5	AR
●	Thyroid dyshormonogenesis 2A	274500	TPO	AR
●	Thyroid dyshormonogenesis 3	274700	TG	AR
●	Thyroid dyshormonogenesis 4	274800	IYD	AR
●	Thyroid dyshormonogenesis 5	274900	DUOXA2	AR
●	Thyroid hormone resistance	274700	THRB	AR
●	Treacher Collins syndrome 3	248390	POLR1C	AR
●	Trifunctional protein deficiency	609015	HADHB	AR
●	Tyrosinemia, type I	276700	FAH	AR
●	Tyrosinemia, type II	276600	TAT	AR
●	Usher syndrome, type 1G	606943	USH1G	AR
●	Usher syndrome, type 2D / Deafness, autosomal recessive 31	611383	DFNB31	AR
●	Usher syndrome, type 3A	276902	CLRN1	AR
●	Ventricular tachycardia, catecholaminergic polymorphic, 2	611938	CASQ2	AR
●	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness	615441	TRDN	AR
●	VLCAD deficiency	201475	ACADVL	AR
●	Walker-Warburg syndrome, Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	253800	FKTN	AR
●	Wilson disease	277900	ATP7B	AR
●	Wolman disease (lysosomal acid lipase deficiency)	278000	LIPA	AR
●	X-linked mental retardation (XLMR) associated with macrocephaly	300659	BRWD3	XL