Table S1. Well-characterized inborn errors of metabolism.

Group/Name of IEM	Alternative names	Inheritance	Gene	OMIM gene number
A. DISORDERS OF NITROGEN-CONTAINING COMPOUNDS				
1. Disorders of pyrimidine metabolism				
CAD trifunctional protein deficiency		AR	*CAD*	114010
Dihydroorotate dehydrogenase deficiency	Postaxial acrofacial dysostosis; Miller syndrome; Genée–Wiedemann syndrome	AR	*DHODH*	126064
Uridine monophosphate synthase deficiency	Hereditary orotic aciduria	AR	*UMPS*	613891
Cytosolic pyrimidine 5'-nucleotidase deficiency	Uridine 5'-monophosphate hydrolase 1 deficiency	AR	*NT5C3A*	606224
Activation-induced cytidine deaminase deficiency	Hyper-IgM syndrome type 2	AR	*AICDA*	605257
Uracil-DNA glycosylase deficiency	Hyper-IgM syndrome type 5	AR	*UNG*	191525
See also: Dihydropyrimidine dehydrogenase, dihydropyrimidinase, and β-ureidopropionase deficiencies in group 17.				
2. Disorders of purine metabolism				
Phosphoribosylpyrophosphate synthetase superactivity		XLR	*PRPS1*	311850
Phosphoribosylpyrophosphate synthetase deficiency	Arts syndrome (severe); X-linked Charcot-Marie-Tooth disease type 5 (intermediate); X-linked deafness type 1 (milder)	XLR	*PRPS1*	311850
Adenylosuccinate lyase deficiency		AR	*ADSL*	608222
Muscle adenosine monophosphate deaminase 1 deficiency	Myoadenylate deaminase deficiency	AR	*AMPD1*	102770
Adenosine monophosphate deaminase 2 deficiency	Pontocerebellar hypoplasia type 9 (severe); autosomal recessive spastic paraplegia type 63 (milder)	AR	*AMPD2*	102771
Erythrocyte adenosine monophosphate deaminase 3 deficiency		AR	*AMPD3*	102772
Adenosine deaminase 1 deficiency		AR	*ADA*	608958
Adenosine deaminase 2 deficiency		AR	*ADA2*	607575
Purine nucleoside phosphorylase deficiency		AR	*PNP*	164050
Xanthine oxidase deficiency	Xanthinuria type 1	AR	*XDH*	607633

Hypoxanthine guanine phosphoribosyltransferase deficiency	Lesch-Nyhan syndrome (severe); Kelley-Seegmiller syndrome (milder)	XLR	*HPRT1*	308000
Adenine phosphoribosyltransferase deficiency		AR	*APRT*	102600
Adenylate kinase 1 deficiency		AR	*AK1*	103000
Adenylate kinase 2 deficiency	Reticular dysgenesis	AR	*AK2*	103020
Inosine-5'-monophosphate dehydrogenase deficiency	Retinitis pigmentosa type 10; Leber congenital amaurosis type 11	AD	*IMPDH1*	146690
Thiopurine methyltransferase deficiency		AR	*TPMT*	187680
Inosine triphosphatase deficiency	Early infantile epileptic encephalopathy type 35	AR	*ITPA*	147520
Urate transporter 1 deficiency	Hereditary renal hypouricemia type 1	AR	*SLC22A12*	607096
Urate voltage-driven efflux transporter 1 deficiency	Hereditary renal hypouricemia type 2	AR	*SLC2A9*	606142
3. Disorders of nucleotide metabolism				
3’ repair exonuclease 1 deficiency	Aicardi-Goutières syndrome type 1; familial chilblain lupus; retinal vasculopathy with cerebral leukodystrophy	AD, AR	*TREX1*	606609
Ribonuclease H2 subunit B deficiency	Aicardi-Goutières syndrome type 2	AR	*RNASEH2B*	610326
Ribonuclease H2 subunit C deficiency	Aicardi-Goutières syndrome type 3	AR	*RNASEH2C*	610330
Ribonuclease H2 subunit A deficiency	Aicardi-Goutières syndrome type 1	AR	*RNASEH2A*	606034
Ribonuclease T2 deficiency	Cystic leukoencephalopathy without megalencephaly	AR	*RNASET2*	612944
SAMHD1 deficiency	Aicardi-Goutières syndrome type 5; stenosis, aneurysm, moyamoya and stroke (SAMS association)	AR	*SAMHD1*	606754
RNA-specific adenosine deaminase deficiency	Aicardi-Goutières syndrome type 6 (recessive); dyschromatosis symmetrica hereditaria (dominant)	AD, AR	*ADAR*	146920
MDA5 superactivity	Aicardi-Goutières syndrome type 7; Singleton-Merten syndrome type 1	AD	*IFIH1*	606951
STING superactivity	STING-associated vasculopathy with onset in infancy (SAVI)	AD	*TMEM173*	612374
2’,5’-oligoadenylate synthetase 1 deficiency	Infantile-onset pulmonary alveolar proteinosis with hypogammaglobulinemia	AD	*OAS1*	164350
ABCC6 deficiency	Generalized arterial calcification of infancy type 2 (severe); pseudoxanthoma elasticum (milder)	AR	*ABCC6*	603234

Ectonucleotide pyrophosphatase/phosphodiesterase 1 deficiency	Generalized arterial calcification of infancy type 1; autosomal recessive hypophosphatemic rickets type 2	AR	*ENPP1*	173335
Ectonucleotide pyrophosphatase/phosphodiesterase 1 dimerization deficiency	Cole disease	AD, AR	*ENPP1*	173335
Ecto-5'-nucleotidase deficiency	Arterial calcification due to deficiency of CD73 (ACDC)	AR	*NT5E*	129190
Equilibrative nucleoside transporter 1 deficiency		AR	*SLC29A1*	602193
Equilibrative nucleoside transporter 3 deficiency	H syndrome; familial Rosai-Dorman disease; Faisalabad histiocytosis	AR	*SLC29A3*	612373
4. Disorders of creatine metabolism				
Arginine:glycine amidinotransferase deficiency	Cerebral creatine deficiency syndrome type 3	AR	*GATM*	602360
Guanidinoacetate methyltransferase deficiency	Cerebral creatine deficiency syndrome type 2	AR	*GAMT*	601240
Creatine transporter deficiency	Cerebral creatine deficiency syndrome type 1	XL	*SLC6A8*	300036
5. Disorders of choline metabolism				
Dimethylglycine dehydrogenase deficiency	Dimethylglycinuria	AR	*DMGDH*	605849
Sarcosine dehydrogenase deficiency	Sarcosinemia	AR	*SARDH*	604455
Flavin monooxygenase 3 deficiency	Primary trimethylaminuria	AR	*FMO3*	136132
6. Disorders of glutathione metabolism				
γ-glutamylcysteine synthetase deficiency	Glutamate-cysteine ligase deficiency	AR	*GCLC*	606857
Glutathione synthetase deficiency		AR	*GSS*	601002
5-Oxoprolinase deficiency		AR	*OPLAH*	614243
Glutathione reductase deficiency		AR	*GSR*	138300
Glutathione peroxidase 4 deficiency	Spondylometaphyseal dysplasia, Sedaghatian type	AR	*GPX4*	138322
NRF2 superactivity	Immunodeficiency, developmental delay, and hypohomocysteinemia (IEMDHH)	AD	*NFE2L2*	600492
7. Disorders of ammonia detoxification				
N-acetylglutamate synthase deficiency		AR	*NAGS*	608300
Carbamoylphosphate synthetase I deficiency		AR	*CPS1*	608307

Ornithine transcarbamylase deficiency		XL	*OTC*	300461
Argininosuccinate synthetase deficiency	Citrullinemia	AR	*ASS1*	603470
Argininosuccinate lyase deficiency	Argininosuccinase deficiency; argininosuccinic aciduria	AR	*ASL*	608310
Arginase deficiency	Argininemia	AR	*ARG1*	608313
Mitochondrial ornithine transporter deficiency	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome; ornithine translocase deficiency	AR	*SLC25A15*	603861
Citrin deficiency	Mitochondrial aspartate-glutamate carrier isoform 2 deficiency	AR	*SLC25A13*	603859
Carbonic anhydrase VA deficiency		AR	*CA5A*	114761
See also: Lysinuric protein intolerance in group 8, δ-1-pyrroline-5-carboxylate synthase and ornithine aminotransferase deficiencies in group 16, glutamate dehydrogenase superactivity in group 20, glutamine synthetase deficiency in group 21, pyruvate carboxylase deficiency in group 52, and transmembrane protein 70 deficiency in group 67.				
8. Disorders of amino acid transport				
Hartnup disorder		AR	*SLC6A19*	608893
Iminoglycinuria		AR (or digenic)	*SLC36A2* (± *SLC6A20*)	608331 (± 605616)
Hyperglycinuria		AD	*SLC36A2*	608331
Cystinuria type A		AR	*SLC3A1*	104614
Cystinuria type B		AD, AR	*SLC7A9*	604144
Lysinuric protein intolerance	Dibasic aminoaciduria type 2	AR	*SLC7A7*	603593
Dicarboxylic aminoaciduria		AR	*SLC1A1*	133550
Large neutral amino acid transporter deficiency		AR	*SLC7A5*	600182
Neuronal system A amino acid transporter deficiency	Foveal hypoplasia type 2 with or without optic nerve misrouting and/or anterior segment dysgenesis	AR	*SLC38A8*	615585
See also: Mitochondrial ornithine transporter and citrin deficiencies in group 7, GABA transporter deficiency in group 17, glutamate aspartate transporter, astroglial glutamate aspartate transporter, and mitochondrial glutamate transporter deficiencies in group 20, ASCT1 transporter deficiency in group 23, glycine transporter 1, glycine transporter 2, and mitochondrial glycine transporter deficiencies in group 24, mitochondrial aspartate-glutamate carrier isoform 1 deficiency in group 57, and cystinosin deficiency in group 107.				
9. Aminoacylase deficiencies				
Aspartoacylase deficiency	Canavan disease; aminoacylase 2 deficiency	AR	*ASPA*	608034
Aminoacylase 1 deficiency		AR	*ACY1*	104620
10. Disorders of monoamine metabolism				

Tyrosine hydroxylase deficiency		AR	*TH*	191290
Aromatic L-amino acid decarboxylase deficiency	DOPA decarboxylase deficiency	AR	*DDC*	107930
Dopamine β-hydroxylase deficiency		AR	*DBH*	609312
Monoamine oxidase A deficiency	Brunner syndrome	XLR	*MAOA*	309850
Dopamine transporter deficiency	Infantile Parkinsonism-dystonia	AR	*SLC6A3*	126455
Vesicular monoamine transporter 2 deficiency		AR	*SLC18A2*	193001
11. Disorders of phenylalanine and tetrahydrobiopterin metabolism				
Phenylalanine hydroxylase deficiency	Phenylketonuria	AR	*PAH*	612349
Autosomal recessive GTP cyclohydrolase 1 deficiency		AR	*GCH1*	600225
Autosomal dominant GTP cyclohydrolase 1 deficiency	Segawa syndrome; dystonia type 5A	AD	*GCH1*	600225
6-pyruvoyl-tetrahydropterin synthase deficiency		AR	*PTS*	612719
Sepiapterin reductase deficiency		AD, AR	*SPR*	182125
Dihydropteridine reductase deficiency		AR	*QDPR*	612676
Pterin-4-α-carbinolamine dehydratase deficiency	Primapterinuria; maturity-onset diabetes of the young (MODY) with hypomagnesemia and renal magnesium loss	AR	*PCBD1*	126090
DNAJC12 deficiency	Non-tetrahydrobiopterin-deficient hyperphenylalaninemia	AR	*DNAJC12*	606060
12. Disorders of tyrosine metabolism				
Tyrosinase deficiency	Oculocutaneous albinism type 1	AR	*TYR*	606933
Tyrosine aminotransferase deficiency	Tyrosinemia type 2; Richner-Hanhart syndrome	AR	*TAT*	613018
4-hydroxyphenylpyruvate dioxygenase deficiency	Tyrosinemia type 3	AR	*HPD*	609695
Hawkinsinuria		AD	*HPD*	609695
Homogentisic acid oxidase deficiency	Alkaptonuria	AR	*HGD*	607474
Maleylacetoacetate isomerase deficiency	Benign hypersuccinylacetonemia	AR	*GSTZ1*	603758
Fumarylacetoacetase deficiency	Tyrosinemia type 1	AR	*FAH*	613871
13. Disorders of sulfur amino acid and sulfide metabolism				
Methionine adenosyltransferase I/III deficiency	Mudd's disease	AR	*MAT1A*	610550

Autosomal dominant hypermethioninemia		AD	*MAT1A*	610550
Glycine N-methyltransferase deficiency		AR	*GNMT*	606628
S-adenosylhomocysteine hydrolase deficiency		AR	*AHCY*	180960
Adenosine kinase deficiency		AR	*ADK*	102750
Cystathionine β-synthase deficiency	Classical homocystinuria	AR	*CBS*	613381
Cystathionine γ-lyase deficiency	Cystathioninuria	AR	*CTH*	607657
Methanethiol oxidase deficiency	Extraoral halitosis	AR	*SELENBP1*	604188
Isolated sulfite oxidase deficiency	Sulfocysteinuria	AR	*SUOX*	606887
Mitochondrial sulfur dioxygenase deficiency	Ethylmalonic encephalopathy	AR	*ETHE1*	608451
14. Disorders of branched-chain amino acid metabolism				
Branched-chain aminotransferase 2 deficiency	Hypervalinemia and hyperleucine-isoleucinemia	AR	*BCAT2*	113530
Branched-chain ketoacid dehydrogenase E1α deficiency	Maple syrup urine disease type 1a	AR	*BCKDHA*	608348
Branched-chain ketoacid dehydrogenase E1β deficiency	Maple syrup urine disease type 1b	AR	*BCKDHB*	248611
Dihydrolipoyl transacylase deficiency	Maple syrup urine disease type 2; branched-chain ketoacid dehydrogenase E2 deficiency	AR	*DBT*	248610
Dihydrolipoamide dehydrogenase deficiency	E3 deficiency	AR	*DLD*	238331
Branched-chain ketoacid dehydrogenase kinase deficiency		AR	*BCKDK*	614901
Isovaleryl-CoA dehydrogenase deficiency	Isovaleric acidemia	AR	*IVD*	607036
Isobutyryl-CoA dehydrogenase deficiency		AR	*ACAD8*	604773
2-Methylbutyryl-CoA dehydrogenase deficiency	Short/branched-chain acyl-CoA dehydrogenase deficiency; 2-methylbutyrylglycinuria	AR	*ACADSB*	600301
3-Methylcrotonyl-CoA carboxylase 1 deficiency	3-methylcrotonylglycinuria type 1	AR	*MCCC1*	609010
3-Methylcrotonyl-CoA carboxylase 2 deficiency	3-methylcrotonylglycinuria type 2	AR	*MCCC2*	609014
3-methylglutaconyl-CoA hydratase deficiency	3-methylglutaconic aciduria type 1	AR	*AUH*	600529

Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	Crotonase deficiency	AR	*ECHS1*	602292
3-hydroxyisobutyryl-CoA hydrolase deficiency	β-hydroxyisobutyryl-CoA deacylase deficiency	AR	*HIBCH*	610690
HSD10 disease	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency	AR	*HSD17B10*	300256
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	Hydroxymethylglutaric aciduria	AR	*HMGCL*	613898
Methylmalonate semialdehyde dehydrogenase deficiency		AR	*ALDH6A1*	603178
Propionic acidemia due to propionyl-CoA carboxylase α subunit deficiency		AR	*PCCA*	232000
Propionic acidemia due to propionyl-CoA carboxylase β subunit deficiency		AR	*PCCB*	232050
Methylmalonyl-CoA epimerase deficiency		AR	*MCEE*	608419
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency		AR	*MUT*	609058
Acyl-CoA synthetase family member 3 deficiency	Combined malonic and methylmalonic aciduria	AR	*ACSF3*	614245
Malonyl-CoA decarboxylase deficiency	Malonic aciduria	AR	*MLYCD*	606761
See also: Large neutral amino acid transporter deficiency in group 8, and mitochondrial acetoacetyl-CoA thiolase deficiency in group 84.				
15. Disorders of lysine metabolism				
α-aminoadipic semialdehyde synthase deficiency	Familial hyperlysinemia; saccharopinuria	AR	*AASS*	605113
α-aminoadipic semialdehyde dehydrogenase deficiency	Pyridoxine-dependent epilepsy	AR	*ALDH7A1*	107323
2-aminoadipic 2-oxoadipic aciduria		AR	*DHTKD1*	614984
Charcot-Marie-Tooth disease type 2Q		AD	*DHTKD1*	614984
Glutaryl-CoA dehydrogenase deficiency	Glutaric acidemia type 1	AR	*GCDH*	608801
Succinate-hydroxymethylglutarate-CoA transferase deficiency	Glutaric acidemia type 3	AR	*SUGCT*	609187
5-phosphohydroxylysine phospholyase	Phosphohydroxylysinuria	AR	*PHYKPL*	614683
16. Disorders of proline and ornithine metabolism				
δ-1-pyrroline-5-carboxylate synthase deficiency, cutis laxa phenotype	Autosomal recessive cutis laxa type 3A; autosomal dominant cutis laxa type 3	AD, AR	*ALDH18A1*	138250

δ-1-pyrroline-5-carboxylate synthase deficiency, spastic paraplegia phenotype	Autosomal dominant spastic paraplegia type 9A; autosomal recessive spastic paraplegia type 9B	AD, AR	*ALDH18A1*	138250
Pyrroline-5-carboxylate reductase 1 deficiency	Autosomal recessive cutis laxa type 2B; autosomal recessive cutis laxa type 3B	AR	*PYCR1*	179035
Pyrroline-5-carboxylate reductase 2 deficiency	Hypomyelinating leukodystrophy type 10	AR	*PYCR2*	616406
Proline dehydrogenase deficiency	Proline oxidase deficiency; hyperprolinemia type 1	AR	*PRODH*	606810
Pyrroline-5-carboxylate dehydrogenase deficiency	Hyperprolinemia type 2	AR	*ALDH4A1*	606811
Hydroxyproline dehydrogenase deficiency	Hydroxyprolinemia	AR	*PRODH2*	616377
Prolidase deficiency		AR	*PEPD*	613230
X-prolyl aminopeptidase 3 deficiency	Nephronophthisis-like nephropathy type 1	AR	*XPNPEP3*	613553
Ornithine aminotransferase deficiency	Gyrate atrophy of choroid and retina	AR	*OAT*	613349
Spermine synthase deficiency	Snyder-Robinson syndrome	AR	*SMS*	300105
See also: 4-hydroxy-2-oxoglutarate aldolase 1 deficiency in group 114.				
17. Disorders of β- and γ-amino acids				
Dihydropyrimidine dehydrogenase deficiency		AR	*DPYD*	612779
Dihydropyrimidinase deficiency	Dihydropyrimidinuria	AR	*DPYS*	613326
β-ureidopropionase deficiency	β-alanine synthase deficiency	AR	*UPB1*	606673
Hyper-β-aminoisobutyric aciduria		AR	*AGXT2*	612471
GABA transaminase deficiency		AR	*ABAT*	137150
Succinic semialdehyde dehydrogenase deficiency	4-hydroxybutyric aciduria	AR	*ALDH5A1*	610045
GABA transporter deficiency	Myoclonic-atonic epilepsy	AD	*SLC6A1*	137165
GABA type A receptor α1 subunit deficiency	Early infantile epileptic encephalopathy type 19	AD	*GABRA1*	137160
GABA type A receptor β1 subunit deficiency	Early infantile epileptic encephalopathy type 45	AD	*GABRB1*	137190
GABA type A receptor β2 subunit deficiency	Infantile or early childhood epileptic encephalopathy type 2	AD	*GABRB2*	600232
GABA type A receptor β3 subunit deficiency	Early infantile epileptic encephalopathy type 43	AD	*GABRB3*	137192

GABA type A receptor γ2 subunit deficiency		AD	*GABRG2*	137164
GABA type B receptor subunit 2 deficiency		AD	*GABBR2*	607340
See also: Methylmalonate semialdehyde dehydrogenase deficiency in group 14.				
18. Disorders of histidine metabolism				
Histidine ammonia-lyase deficiency	Histidase deficiency; histidinemia	AR	*HAL*	609457
19. Disorders of tryptophan metabolism				
Kynureninase deficiency	Xanthurenic aciduria; vertebral, cardiac, renal, and limb defects syndrome type 2	AR	*KYNU*	605197
3-hydroxyanthranilic acid 3,4-dioxygenase deficiency	Vertebral, cardiac, renal, and limb defects syndrome type 1	AR	*HAAO*	604521
Tryptophan 2,3-dioxygenase deficiency	Hypertryptophanemia	AR	*TDO2*	191070
See also: Hartnup disorder in group 8.				
20. Disorders of glutamate metabolism				
Glutamate dehydrogenase superactivity	Hyperinsulinism-hyperammonemia syndrome; familial hyperinsulinemic hypoglycemia type 6	AD	*GLUD1*	138130
Mitochondrial glutamate transporter deficiency	Early infantile epileptic encephalopathy type 3	AR	*SLC25A22*	609302
Glutamate aspartate transporter deficiency	EAAT1 deficiency; episodic ataxia type 6	AD	*SLC1A3*	600111
Astroglial glutamate aspartate transporter deficiency	EAAT2 deficiency; early infantile epileptic encephalopathy type 41	AD	*SLC1A2*	600300
Ionotropic glutamate receptor NMDA type subunit 1 dysregulation	Autosomal dominant mental retardation type 8; neurodevelopmental disorder with or without hyperkinetic movements and seizures	AD, AR	*GRIN1*	138249
Ionotropic glutamate receptor NMDA type subunit 2A dysregulation		AD	*GRIN2A*	138253
Ionotropic glutamate receptor NMDA type subunit 2B dysregulation	Early infantile epileptic encephalopathy type 27; autosomal dominant mental retardation type 6	AD	*GRIN2B*	138252
Ionotropic glutamate receptor NMDA type subunit 2D superactivity	Early infantile epileptic encephalopathy type 46	AD	*GRIN2D*	602717
Ionotropic glutamate receptor AMPA type subunit 3 deficiency	Syndromic X-linked mental retardation, Wu type	XLR	*GRIA3*	305915

Ionotropic glutamate receptor AMPA type subunit 4 dysregulation	Neurodevelopmental disorder with or without seizures and gait abnormalities (NEDSGA)	AD	*GRIA4*	138246
Metabotropic glutamate receptor 1 deficiency	Autosomal recessive spinocerebellar ataxia type 13	AR	*GRM1*	604473
Metabotropic glutamate receptor 1 superactivity	Spinocerebellar ataxia type 44	AD	*GRM1*	604473
Metabotropic glutamate receptor 6 deficiency	Congenital stationary night blindness type 1B	AR	*GRM6*	604096
21. Disorder of glutamine metabolism				
Glutamine synthetase deficiency		AR	*GLUL*	138290
22. Disorder of asparagine metabolism				
Asparagine synthetase deficiency		AR	*ASNS*	108370
23. Disorders of serine metabolism				
3-phosphoglycerate dehydrogenase deficiency		AR	*PHGDH*	606879
Phosphoserine aminotransferase deficiency		AR	*PSAT1*	610936
Phosphoserine phosphatase deficiency		AR	*PSPH*	172480
ASCT1 transporter deficiency	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	AR	*SLC1A4*	600229
24. Disorders of glycine metabolism				
Glycine encephalopathy due to glycine decarboxylase deficiency	Nonketotic hyperglycinemia	AR	*GLDC*	238300
Glycine encephalopathy due to aminomethyltransferase deficiency	Nonketotic hyperglycinemia	AR	*AMT*	238310
Glycine transporter 1 deficiency	Glycine encephalopathy with normal serum glycine	AR	*SLC6A9*	601019
Glycine transporter 2 deficiency	Hereditary hyperekplexia type 3	AD, AR	*SLC6A5*	604159
Glycine receptor α1 subunit deficiency	Hereditary hyperekplexia type 1	AD, AR	*GLRA1*	138491
Glycine receptor β subunit deficiency	Hereditary hyperekplexia type 2	AR	*GLRB*	138492
Mitochondrial glycine transporter deficiency	Congenital sideroblastic anemia type 2	AR	*SLC25A38*	610819
See also: Lipoyltransferase 2, lipoic acid synthase, NFU1, BOLA3, glutaredoxin 5, IBA57, and ISCA2 deficiencies in group 25.				

B. DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS				
25. Disorders of lipoic acid and iron-sulfur metabolism				
Lipoyltransferase 2 deficiency	Neonatal severe encephalopathy with lactic acidosis and brain abnormalities (NELABA)	AR	*LIPT2*	617659
Lipoic acid synthase deficiency	Hyperglycinemia, lactic acidosis, and seizures	AR	*LIAS*	607031
Lipoyltransferase 1 deficiency		AR	*LIPT1*	610284
NFU1 deficiency	Multiple mitochondrial dysfunctions syndrome type 1	AR	*NFU1*	608100
BOLA3 deficiency	Multiple mitochondrial dysfunctions syndrome type 2 with hyperglycinemia	AR	*BOLA3*	613183
Glutaredoxin 5 deficiency		AR	*GLRX5*	609588
IBA57 deficiency		AR	*IBA57*	615316
ISCA1 deficiency	Multiple mitochondrial dysfunctions syndrome type 5	AR	*ISCA1*	611006
ISCA2 deficiency	Multiple mitochondrial dysfunctions syndrome type 4	AR	*ISCA2*	615317
ISCU deficiency	Hereditary myopathy with lactic acidosis, Swedish type myopathy with exercise intolerance	AD, AR	*ISCU*	611911
ABCB7 deficiency	Sideroblastic anemia and spinocerebellar ataxia	XLR	*ABCB7*	300135
Ferredoxin reductase deficiency	Auditory neuropathy and optic atrophy	AR	*FDXR*	103270
Frataxin deficiency	Friedreich ataxia	AR	*FXN*	606829
See also: HSPA9 deficiency in group 79.				
26. Disorders of cobalamin metabolism				
Hereditary intrinsic factor deficiency		AR	*GIF*	609342
Cubilin deficiency	Imerslund-Gräsbeck disease, Finnish type	AR	*CUBN*	602997
Amnionless deficiency	Imerslund-Gräsbeck disease, Norwegian type	AR	*AMN*	605799
Haptocorrin deficiency	Transcobalamin I deficiency	AD, AR	*TCN1*	189905
Transcobalamin II deficiency		AR	*TCN2*	613441
Transcobalamin receptor deficiency		AR	*CD320*	606475
Methylmalonic aciduria and homocystinuria, cblF type		AR	*LMBRD1*	612625
Methylmalonic aciduria and homocystinuria, cblJ type		AR	*ABCD4*	603214

Methylmalonic aciduria and homocystinuria, cblC type		AR	*MMACHC*	609831
Epi-cblC		AR	*MMACHC + PRDX1*	609831 + 176763
cblD disease		AR	*MMADHC*	611935
Methionine synthase reductase deficiency	Homocystinuria-megaloblastic anemia, cblE type	AR	*MTRR*	602568
Methionine synthase deficiency	Homocystinuria-megaloblastic anemia, cblG type	AR	*MTR*	156570
Methylmalonic aciduria, cblA type		AR	*MMAA*	607481
Methylmalonic aciduria, cblB type		AR	*MMAB*	607568
Methylmalonic aciduria and homocystinuria, cblX type		XLR	*HCFC1*	300019
27. Disorders of folate metabolism				
Proton-coupled folate transporter deficiency	Hereditary folate malabsorption	AR	*SLC46A1*	611672
Folate receptor α deficiency	Neurodegeneration due to cerebral folate transport deficiency	AR	*FOLR1*	136430
5,10-methylenetetrahydrofolate reductase deficiency		AR	*MTHFR*	607093
Methylenetetrahydrofolate dehydrogenase 1 deficiency	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia (CIMAH)	AR	*MTHFD1*	172460
Dihydrofolate reductase deficiency		AR	*DHFR*	126060
Glutamate formiminotransferase deficiency	Formiminoglutamic aciduria	AR	*FTCD*	606806
28. Disorders of biotin metabolism				
Biotinidase deficiency		AR	*BTD*	609019
Holocarboxylase synthetase deficiency		AR	*HLCS*	609018
29. Disorders of thiamine metabolism				
Thiamine transporter 1 deficiency	Thiamine-responsive megaloblastic anemia; Rogers syndrome; thiamine metabolism dysfunction syndrome type 1	AR	*SLC19A2*	603941
Thiamine transporter 2 deficiency	Biotin-thiamine-responsive basal ganglia disease; thiamine metabolism dysfunction syndrome type 2	AR	*SLC19A3*	606152
Thiamine pyrophosphokinase deficiency	Thiamine metabolism dysfunction syndrome type 5	AR	*TPK1*	606370

Mitochondrial thiamine pyrophosphate transporter deficiency	Amish lethal microcephaly, thiamine metabolism dysfunction syndrome type 3 (severe); bilateral striatal necrosis and progressive polyneuropathy, thiamine metabolism dysfunction syndrome type 4 (milder)	AR	*SLC25A19*	606521
30. Disorders of riboflavin metabolism				
Riboflavin transporter 1 deficiency	Transient riboflavin deficiency	AD	*SLC52A1*	607883
Riboflavin transporter 2 deficiency	Brown-Vialetto-van Laere syndrome type 1	AR	*SLC52A3*	613350
Riboflavin transporter 3 deficiency	Brown-Vialetto-van Laere syndrome type 2	AR	*SLC52A2*	607882
Flavin adenine dinucleotide synthetase deficiency		AR	*FLAD1*	610595
Mitochondrial flavin adenine dinucleotide transporter deficiency	Riboflavin-responsive exercise intolerance	AR	*SLC25A32*	610815
Electron transfer flavoprotein α subunit deficiency	Glutaric acidemia type 2A; multiple acyl-CoA dehydrogenase deficiency type 2A	AR	*ETFA*	608053
Electron transfer flavoprotein β subunit deficiency	Glutaric acidemia type 2B; multiple acyl-CoA dehydrogenase deficiency type 2B	AR	*ETFB*	130410
Electron transfer flavoprotein dehydrogenase deficiency	Glutaric acidemia type 2C; multiple acyl-CoA dehydrogenase deficiency type 2C	AR	*ETFDH*	231675
31. Disorders of niacin and NAD metabolism				
Nicotinamide mononucleotide adenylyl transferase 1 deficiency	Leber congenital amaurosis 9	AR	*NMNAT1*	608700
Mitochondrial NAD kinase 2 deficiency	2,4-dienoyl-CoA reductase deficiency with hyperlysinemia	AR	*NADK2*	615787
NAD(P)HX epimerase deficiency	Apolipoprotein A-I binding protein deficiency	AR	*NAXE*	608862
NAD(P)HX dehydratase deficiency	CARKD deficiency	AR	*NAXD*	615910
Nicotinamide nucleotide transhydrogenase deficiency	Glucocorticoid deficiency type 4	AR	*NNT*	607878
See also: Hartnup disorder in group 8, and kynureninase and 3-hydroxyanthranilic acid 3,4-dioxygenase deficiencies in group 19.				
32. Disorders of pantothenate metabolism				
Pantothenate kinase 2 deficiency	Pantothenate kinase-associated neurodegeneration (PKAN); neurodegeneration with brain iron accumulation type 1	AR	*PANK2*	606157

Coenzyme A synthase deficiency	Coenzyme A synthase protein-associated neurodegeneration (CoPAN); neurodegeneration with brain iron accumulation type 6	AR	*COASY*	609855
33. Disorders of pyridoxine metabolism				
Pyridoxamine 5'-phosphate oxidase deficiency		AR	*PNPO*	603287
Pyridoxal 5'-phosphate binding protein deficiency	PROSC deficiency	AR	*PLPBP*	604436
Tissue-nonspecific alkaline phosphatase deficiency	Hypophosphatasia	AD, AR	*ALPL*	171760
See also: α-aminoadipic semialdehyde dehydrogenase deficiency in group 15, and pyrroline-5-carboxylate dehydrogenase deficiency in group 16.				
34. Disorder of vitamin C metabolism				
L-dehydroascorbate transporter deficiency	GLUT10 deficiency; arterial tortuosity syndrome	AR	*SLC2A10*	606145
35. Disorders of vitamin A metabolism				
β-carotene 15,15'-dioxygenase deficiency	Hypercarotenemia and vitamin A deficiency	AD	*BCO1*	605748
Plasma retinol-binding protein deficiency	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome (recessive); isolated microphthalmia and/or coloboma type 10 (dominant)	AD, AR	*RBP4*	180250
Vitamin A receptor deficiency	Matthew-Wood syndrome; pulmonary hypoplasia-diaphragmatic hernia-anophthalmia-cardiac defect (PDAC) syndrome	AR	*STRA6*	610745
Lecithin retinol acyltransferase deficiency	Leber congenital amaurosis type 14	AR	*LRAT*	604863
Retinal isomerase deficiency	Leber congenital amaurosis type 2	AR	*RPE65*	180069
Retinol dehydrogenase 5 deficiency	Fundus albipunctatus	AD, AR	*RDH5*	601617
Retinol dehydrogenase 12 deficiency	Leber congenital amaurosis type 13	AR	*RDH12*	608830
Interphotoreceptor retinol-binding protein deficiency	Retinitis pigmentosa type 66	AR	*RBP3*	180290
Retinaldehyde dehydrogenase 3 deficiency	Isolated microphthalmia type 8	AR	*ALDH1A3*	600463
Cellular retinaldehyde-binding protein deficiency		AD, AR	*RLBP1*	180090
36. Disorders of vitamin D metabolism				
1-α-hydroxylase deficiency	Vitamin D-dependent rickets type 1A	AR	*CYP27B1*	609506
Vitamin D 25-hydroxylase deficiency	Vitamin D-dependent rickets type 1B	AR	*CYP2R1*	608713

Vitamin D receptor deficiency	Vitamin D-dependent rickets type 2A	AR	*VDR*	601769
Vitamin D 24-hydroxylase deficiency	Infantile hypercalcemia type 1	AR	*CYP24A1*	126065
37. Disorder of vitamin E metabolism				
α-tocopherol transfer protein deficiency	Ataxia with isolated vitamin E deficiency	AR	*TTPA*	600415
38. Disorders of vitamin K metabolism				
γ-glutamyl carboxylase deficiency	Combined deficiency of vitamin K-dependent coagulation factors type 1	AR	*GGCX*	137167
Vitamin K epoxide reductase deficiency	Combined deficiency of vitamin K-dependent coagulation factors type 2	AR	*VKORC1*	608547
Microsomal epoxide hydrolase deficiency	Familial hypercholanemia	AR	*EPHX1*	132810
Menaquinone-4 synthetase deficiency	Schnyder corneal dystrophy	AD	*UBIAD1*	611632
39. Disorders of molybdenum metabolism				
Cyclic pyranopterin monophosphate synthase deficiency	Molybdenum cofactor deficiency type A	AR	*MOCS1*	603707
Molybdopterin synthase deficiency	Molybdenum cofactor deficiency type B	AR	*MOCS2*	603708
Gephyrin deficiency	Molybdenum cofactor deficiency type C	AR	*GPHN*	603930
Molybdenum cofactor sulfurase deficiency	Xanthinuria type 2	AR	*MOCOS*	613274
40. Disorders of copper metabolism				
Copper-transporting ATPase β subunit deficiency	Wilson disease; hepatolenticular degeneration	AR	*ATP7B*	606882
Copper-transporting ATPase α subunit deficiency	Menkes disease (severe); occipital horn syndrome (milder)	XLR	*ATP7A*	300011
ATP7A-related distal motor neuropathy	X-linked distal spinal muscular atrophy type 3	XLR	*ATP7A*	300011
MEDNIK syndrome		AR	*AP1S1*	603531
Acetyl-CoA transporter deficiency	Huppke-Brendel syndrome; congenital cataracts, hearing loss, and neurodegeneration	AR	*SLC33A1*	603690
41. Disorders of iron metabolism				
Hereditary hemochromatosis type 1		AR	*HFE*	613609
Hemojuvelin deficiency	Hereditary hemochromatosis type 2A	AR	*HFE2*	608374
Hepcidin deficiency	Hereditary hemochromatosis type 2B	AR	*HAMP*	606464
Transferrin receptor 2 deficiency	Hereditary hemochromatosis type 3	AR	*TFR2*	604720
Ferroportin deficiency	Hereditary hemochromatosis type 4	AD	*SLC40A1*	604653
Ferritin light chain deficiency	Hereditary L-ferritin deficiency	AD, AR	*FTL*	134790

Ferritin light chain superactivity	Neuroferritinopathy; neurodegeneration with brain iron accumulation 3	AR	*FTL*	134790
Ferritin light chain dysregulation	Hyperferritinemia-cataract syndrome	AR	*FTL*	134790
Hereditary ceruloplasmin deficiency	Aceruloplasminemia	AR	*CP*	117700
Matriptrase 2 deficiency	Iron-refractory iron deficiency anemia	AR	*TMPRSS6*	609862
Hereditary transferrin deficiency	Atransferrinemia	AR	*TF*	190000
Transferrin receptor deficiency	Immunodeficiency type 46	AR	*TFRC*	190010
Divalent metal transporter 1 deficiency	Hypochromic microcytic anemia with iron overload type 1	AR	*SLC11A2*	600523
42. Disorders of manganese metabolism				
Hypermanganesemia with dystonia type 1		AR	*SLC30A10*	611146
Hypermanganesemia with dystonia type 2		AR	*SLC39A14*	608736
SLC39A8 deficiency		AR	*SLC39A8*	608732
See also: ATP13A2 deficiency in group 101.				
43. Disorders of zinc metabolism				
Acrodermatitis enteropathica		AR	*SLC39A4*	607059
Transient neonatal zinc deficiency		AD	*SLC30A2*	609617
Spondylocheirodysplastic Ehlers-Danlos syndrome		AR	*SLC39A13*	608735
Birk-Landau-Perez syndrome		AR	*SLC30A9*	604604
44. Disorders of selenium metabolism				
Selenocysteine insertion sequence-binding protein 2 deficiency		AR	*SECISBP2*	607693
O-phosphoseryl-tRNA(Sec) selenium transferase deficiency	Selenocysteinyl-tRNA(Sec) synthase deficiency; progressive cerebellocerebral atrophy; pontocerebellar hypoplasia type 2D	AR	*SEPSECS*	613009
45. Disorders of magnesium metabolism				
Epithelial magnesium transporter deficiency	Hypomagnesemia with secondary hypocalcemia	AR	*TRPM6*	607009
Sodium-potassium ATPase γ subunit deficiency	Autosomal dominant hypomagnesemia with hypocalciuria; renal hypomagnesemia type 2	AD	*FXYD2*	601814
Claudin 10 deficiency	Hypohydrosis, electrolyte imbalance, lacrimal gland dysfunction, ichthyosis and xerostomia (HELIX) syndrome	AR	*CLDN10*	617579

Claudin 16 deficiency	Familial hypomagnesemia with hypercalciuria and nephrocalcinosis type 1; renal hypomagnesemia type 3	AR	*CLDN16*	603959
Claudin 19 deficiency	Familial hypomagnesemia with hypercalciuria and nephrocalcinosis type 2; renal hypomagnesemia type 5 with ocular involvement	AR	*CLDN19*	610036
Cyclin M2 deficiency	Renal hypomagnesemia type 6	AD, AR	*CNNM2*	607803
Sodium-chloride cotransporter deficiency	Gitelman syndrome	AR	*SLC12A3*	600968
KCNJ10 deficiency	Epilepsy, ataxia, sensorineural deafness, tubulopathy (EAST) syndrome; seizures, sensorineural deafness, ataxia, mental retardation, electrolyte imbalance (SeSAME) syndrome	AR	*KCNJ10*	612780
See also: Pterin-4-α-carbinolamine dehydratase deficiency in group 11, hepatocyte nuclear factor-1β deficiency in group 50, mitochondrial tRNA(Ile) deficiency in group 73, and mitochondrial seryl-tRNA synthetase deficiency in group 74.				
C. DISORDERS OF CARBOHYDRATES				
46. Disorders of carbohydrate transport and absorption				
Blood-brain barrier glucose transporter 1 deficiency	GLUT1 deficiency	AD, AR	*SLC2A1*	138140
Neuronal glucose transporter deficiency	Intellectual developmental disorder with neuropsychiatric features	AR	*SLC45A1*	605763
Glucose transporter 2 deficiency	Fanconi-Bickel syndrome	AR	*SLC2A2*	138160
Intestinal sodium-glucose cotransporter 1 deficiency	Glucose-galactose malabsorption	AR	*SLC5A1*	182380
Congenital sucrase-isomaltase deficiency		AR	*SI*	609845
Trehalase deficiency			*TREH*	275360
Congenital lactase deficiency	Congenital alactasia	AR	*LCT*	603202
Renal sodium-glucose cotransporter 2 deficiency	Familial renal glucosuria type 1	AD, AR	*SLC5A2*	182381
47. Disorders of galactose metabolism				
Galactose-1-phosphate uridylyltransferase deficiency	Classic galactosemia; galactosemia type 1	AR	*GALT*	606999
Galactose epimerase deficiency	Galactosemia type 3	AR	*GALE*	606953
Galactokinase deficiency	Galactosemia type 2	AR	*GALK1*	604313
48. Disorders of fructose metabolism				
Hepatic fructokinase deficiency	Essential fructosuria	AR	*KHK*	614058

Aldolase B deficiency	Hereditary fructose intolerance	AR	*ALDOB*	612724
See also: Fructose-1,6-bisphosphatase deficiency in group 52.				
49. Disorders of the pentose phosphate pathway and polyol metabolism				
Glucose-6-phosphate dehydrogenase deficiency		XLR	*G6PD*	305900
Ribose 5-phosphate isomerase deficiency		AR	*RPIA*	180430
Transaldolase deficiency		AR	*TALDO1*	602063
Transketolase deficiency	Short stature, developmental delay, and congenital heart defects	AR	*TKT*	606781
Sedoheptulose kinase deficiency		AR	*SHPK*	605060
L-xylulose reductase deficiency	Pentosuria; xylitol dehydrogenase deficiency	AR	*DCXR*	608347
50. Disorders of insulin secretion and signaling				
ATP-sensitive potassium channel regulatory subunit deficiency	Familial hyperinsulinemic hypoglycemia type 1	AD, AR	*ABCC8*	600509
ATP-sensitive potassium channel regulatory subunit superactivity	Developmental delay, epilepsy and neonatal diabetes (DEND), permanent or transient neonatal diabetes without neurologic features (severe); maturity-onset diabetes of the young type 12 (milder)	AD, AR	*ABCC8*	600509
ATP-sensitive potassium channel pore-forming subunit deficiency	Familial hyperinsulinemic hypoglycemia type 2	AR	*KCNJ11*	600937
ATP-sensitive potassium channel pore-forming subunit superactivity	Developmental delay, epilepsy and neonatal diabetes (DEND), permanent or transient neonatal diabetes without neurologic features (severe); maturity-onset diabetes of the young type 13 (milder)	AD, AR	*KCNJ11*	600937
Hepatocyte nuclear factor-4α deficiency		AD	*HNF4A*	600281
Hepatocyte nuclear factor-1α deficiency		AD	*HNF1A*	142410
Hepatocyte nuclear factor-1β deficiency		AD	*HNF1B*	189907
Uncoupling protein 2 deficiency		AD	*UCP2*	601693
Insulin deficiency	Permanent neonatal diabetes mellitus (severe); maturity-onset diabetes of the young type 10 (milder)	AD	*INS*	176730
Proinsulin cleavage deficiency	Hyperproinsulinemia	AD	*INS*	176730
Insulin receptor dysregulation	Familial hyperinsulinemic hypoglycemia type 5	AD	*INSR*	147670

Insulin promoter factor 1 deficiency	Maturity-onset diabetes of the young type 4 (dominant); pancreatic agenesis (recessive)	AD, AR	*PDX1*	600733
Neurogenic differentiation factor 1 deficiency	Maturity-onset diabetes of the young type 6 (dominant); permanent neonatal diabetes and neurologic anomalies (recessive)	AD, AR	*NEUROD1*	601724
Krüppel-like factor 11 deficiency	Maturity-onset diabetes of the young type 7	AD	*KLF11*	603301
PAX4 deficiency	Maturity-onset diabetes of the young type 9	AD	*PAX4*	167413
BLK deficiency	Maturity-onset diabetes of the young type 11	AD	*BLK*	191305
APPL1 deficiency	Maturity-onset diabetes of the young type 14	AD	*APPL1*	604299
AKT2 superactivity	Hypoinsulinemic hypoglycemia with hemihypertrophy	AD	*AKT2*	164731
RFX6 deficiency	Mitchell-Riley syndrome (recessive); maturity-onset diabetes of the young (dominant)	AD, AR	*RFX6*	612659
See also: Adenosine kinase deficiency in group 13, glutamate dehydrogenase superactivity in group 20, glucokinase superactivity in group 53, short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency in group 83, catalytic phosphatidylinositol 3-kinase α subunit superactivity in group 93, phosphomannomutase 2, phosphomannose isomerase, ALG3 α-1,3-mannosyltransferase, and ALG6 α-1,3-glucosyltransferase deficiencies in group 115, and phosphoglucomutase 1 deficiency in group 125.				
51. Glycogen storage diseases				
Muscle glycogenin 1 deficiency	Glycogen storage disease type 15; polyglucosan body myopathy type 2	AR	*GYG1*	603942
Muscle glycogen synthase deficiency	Glycogen storage disease type 0b	AR	*GYS1*	138570
Hepatic glycogen synthase deficiency	Glycogen storage disease type 0a	AR	*GYS2*	138571
Glucose-6-phosphate transporter deficiency	Glycogen storage disease type 1b	AR	*SLC37A4*	602671
α-glucosidase deficiency	Glycogen storage disease type 2; Pompe disease	AR	*GAA*	606800
Glycogen debranching enzyme deficiency	Glycogen storage disease type 3; Cori-Forbes disease; limit dextrinosis	AR	*AGL*	610860
Glycogen branching enzyme deficiency	Glycogen storage disease type 4; Andersen disease; adult polyglucosan body disease	AR	*GBE1*	607839
Muscle glycogen phosphorylase deficiency	Glycogen storage disease type 5; McArdle disease	AR	*PYGM*	608455
Liver glycogen phosphorylase deficiency	Glycogen storage disease type 6; Hers disease	AR	*PYGL*	613741
Hepatic phosphorylase kinase α2 subunit deficiency	Glycogen storage disease type 9a	XLR	*PHKA2*	300798

Phosphorylase kinase β subunit deficiency	Glycogen storage disease type 9b	AR	*PHKB*	172490
Hepatic phosphorylase kinase γ2 subunit deficiency	Glycogen storage disease type 9c	AR	*PHKG2*	172471
Muscle phosphorylase kinase α1 subunit deficiency	Glycogen storage disease type 9d	AR	*PHKA1*	311870
HOIL1 deficiency	Polyglucosan body myopathy type 1; HOIL deficiency	AR	*RBCK1*	610924
Cardiac phosphorylase kinase deficiency		AD	*PRKAG2*	602743
Lysosome-associated membrane protein 2 deficiency	Danon disease	XL	*LAMP2*	309060
Laforin deficiency	Progressive myoclonic epilepsy type 2A	AR	*EPM2A*	607566
Malin deficiency	Progressive myoclonic epilepsy type 2B	AR	*NHLRC1*	608072
See also: α-glucosidase deficiency in group 51, glucose-6-phosphatase deficiency in group 52, muscle phosphofructokinase, aldolase A, enolase β, muscle phosphoglycerate mutase, and lactate dehydrogenase A deficiencies in group 53, and phosphoglucomutase 1 deficiency in group 125.				
52. Disorders of gluconeogenesis				
Glucose-6-phosphatase deficiency	Glycogen storage disease type 1a	AR	*G6PC*	613742
Fructose-1,6-bisphosphatase deficiency		AR	*FBP1*	611570
Pyruvate carboxylase deficiency		AR	*PC*	608786
Cytosolic phosphoenolpyruvate carboxykinase deficiency		AR	*PCK1*	614168
53. Disorders of glycolysis				
Hemolytic anemia due to hexokinase deficiency		AR	*HK1*	142600
Hereditary motor and sensory neuropathy, Russe type	Charcot-Marie-Tooth disease type 4G	AR	*HK1*	142600
Retinitis pigmentosa type 79		AD	*HK1*	142600
Glucokinase deficiency	Permanent neonatal diabetes mellitus; MODY type 2	AD	*GCK*	138079
Glucokinase superactivity	Familial hyperinsulinemic hypoglycemia type 3	AD	*GCK*	138079
Glucose-6-phosphate isomerase deficiency		AR	*GPI*	172400
Muscle phosphofructokinase deficiency	Glycogen storage disease type 7; Tarui disease	AR	*PFKM*	610681
Aldolase A deficiency	Glycogen storage disease type 12	AR	*ALDOA*	103850
Triose phosphate isomerase deficiency		AR	*TPI1*	190450
Phosphoglycerate kinase deficiency		XLR	*PGK1*	311800

Muscle phosphoglycerate mutase deficiency	Glycogen storage disease type 10; DiMauro disease	AR	*PGAM2*	612931
Enolase β deficiency	Glycogen storage disease type 13	AR	*ENO3*	131370
Pyruvate kinase deficiency		AR	*PKLR*	609712
Lactate dehydrogenase A deficiency	Glycogen storage disease type 11	AR	*LDHA*	150000
Lactate dehydrogenase B deficiency		AD, AR	*LDHB*	150100
D. MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM				
54. Disorders of pyruvate metabolism				
Pyruvate dehydrogenase E1-α deficiency		XL	*PDHA1*	300502
Pyruvate dehydrogenase E1-β deficiency		AR	*PDHB*	179060
Dihydrolipoamide acetyltransferase deficiency	Pyruvate dehydrogenase E2 deficiency	AR	*DLAT*	608770
Pyruvate dehydrogenase E3-binding protein deficiency	Pyruvate dehydrogenase component X deficiency	AR	*PDHX*	608769
Pyruvate dehydrogenase phosphatase deficiency		AR	*PDP1*	605993
Mitochondrial pyruvate carrier deficiency		AR	*MPC1*	614738
Pyruvate dehydrogenase kinase isoenzyme 3 superactivity		XLD	*PDK3*	300906
See also: Dihydrolipoamide dehydrogenase deficiency in group 14, and pyruvate carboxylase deficiency in group 52.				
55. Disorders of the Krebs cycle				
Mitochondrial aconitase deficiency	Infantile cerebellar-retinal degeneration	AR	*ACO2*	100850
Mitochondrial NADH-dependent isocitrate dehydrogenase 2 superactivity	D-2-hydroxyglutaric aciduria type 2	AD	*IDH2*	147650
Mitochondrial NADPH-dependent isocitrate dehydrogenase 3 β subunit deficiency		AR	*IDH3B*	604526
ATP-specific succinyl-CoA ligase β subunit deficiency	Mitochondrial DNA depletion syndrome type 5	AR	*SUCLA2*	603921
GTP-specific succinyl-CoA ligase α subunit deficiency	Mitochondrial DNA depletion syndrome type 9	AR	*SUCLG1*	611224
Fumarate hydratase deficiency	Fumarase deficiency	AR	*FH*	136850
Fumarate hydratase deficiency, tumoral phenotype	Reed syndrome	AD	*FH*	136850

Mitochondrial malate dehydrogenase deficiency	Early infantile epileptic encephalopathy type 51	AR	*MDH2*	154100
See also: Dihydrolipoamide dehydrogenase deficiency in group 14, succinate dehydrogenase subunits A, B, C and D deficiencies in group 60, and succinate dehydrogenase assembly factors 1 and 2 deficiencies in group 61.				
56. Disorders of metabolite repair				
D-2-hydroxyglutarate dehydrogenase deficiency	D-2-hydroxyglutaric aciduria type 1	AR	*D2HGDH*	609186
L-2-hydroxyglutarate dehydrogenase deficiency	L-2-hydroxyglutaric aciduria	AR	*L2HGDH*	609584
See also: NAD(P)HX epimerase deficiency and NAD(P)HX dehydratase deficiency in group 31.				
57. Disorders of mitochondrial carriers				
Adenine nucleotide translocator deficiency	Mitochondrial DNA depletion syndrome type 12 (cardiomyopathic type); adPEO with mitochondrial DNA deletions type 2	AD, AR	*SLC25A4*	103220
Mitochondrial phosphate carrier deficiency		AR	*SLC25A3*	600370
Mitochondrial aspartate-glutamate carrier isoform 1 deficiency	Early infantile epileptic encephalopathy type 39; aralar 1 deficiency	AR	*SLC25A12*	603667
Cytosolic glycerol-3-phosphate dehydrogenase deficiency	Transient infantile hypertriglyceridemia	AR	*GPD1*	138420
S-adenosylmethionine carrier deficiency	Combined oxidative phosphorylation deficiency type 28	AR	*SLC25A26*	611037
Mitochondrial citrate carrier deficiency	Combined D-2- and L-2-hydroxyglutaric aciduria	AR	*SLC25A1*	190315
Mitochondrial ATP-Mg/phosphate transporter deficiency	Gorlin-Chaudhry-Moss syndrome; Fontaine syndrome	AD	*SLC25A24*	608744
See also: Mitochondrial ornithine transporter and citrin deficiencies in group 7, mitochondrial glutamate transporter deficiency in group 20, mitochondrial glycine transporter deficiency in group 24, mitochondrial thiamine pyrophosphate transporter deficiency in group 29, mitochondrial flavin adenine dinucleotide transporter deficiency in group 30, mitochondrial pyruvate carrier deficiency in group 54, and carnitine-acylcarnitine translocase deficiency in group 82.				
58. Disorders of complex I subunits				
NADH dehydrogenase flavoprotein 1 deficiency		AR	*NDUFV1*	161015
NADH dehydrogenase flavoprotein 2 deficiency		AR	*NDUFV2*	600532

NADH dehydrogenase iron-sulfur protein 1 deficiency		AR	*NDUFS1*	157655
NADH dehydrogenase iron-sulfur protein 2 deficiency		AR	*NDUFS2*	602985
NADH dehydrogenase iron-sulfur protein 3 deficiency		AR	*NDUFS3*	603846
NADH dehydrogenase iron-sulfur protein 7 deficiency		AR	*NDUFS7*	601825
NADH dehydrogenase iron-sulfur protein 8 deficiency		AR	*NDUFS8*	602141
NADH dehydrogenase iron-sulfur protein 4 deficiency		AR	*NDUFS4*	602694
NADH dehydrogenase iron-sulfur protein 6 deficiency		AR	*NDUFS6*	603848
NADH dehydrogenase α subcomplex subunit 1 deficiency		XLR	*NDUFA1*	300078
NADH dehydrogenase α subcomplex subunit 2 deficiency		AR	*NDUFA2*	602137
NADH dehydrogenase α subcomplex subunit 9 deficiency		AR	*NDUFA9*	603834
NADH dehydrogenase α subcomplex subunit 10 deficiency		AR	*NDUFA10*	603835
NADH dehydrogenase α subcomplex subunit 12 deficiency		AR	*NDUFA12*	614530
NADH dehydrogenase β subcomplex subunit 3 deficiency		AR	*NDUFB3*	603839
NADH dehydrogenase β subcomplex subunit 8 deficiency		AR	*NDUFB8*	602140
NADH dehydrogenase β subcomplex subunit 11 deficiency	Linear skin defects with multiple congenital anomalies type 3	XL	*NDUFB11*	300403
NADH dehydrogenase core subunit 1 deficiency		Mit	*MT-ND1*	516000
NADH dehydrogenase core subunit 2 deficiency		Mit	*MT-ND2*	516001
NADH dehydrogenase core subunit 3 deficiency		Mit	*MT-ND3*	516002

NADH dehydrogenase core subunit 4 deficiency		Mit	*MT-ND4*	516003
NADH dehydrogenase core subunit 4L deficiency		Mit	*MT-ND4L*	516004
NADH dehydrogenase core subunit 5 deficiency		Mit	*MT-ND5*	516005
NADH dehydrogenase core subunit 6 deficiency		Mit	*MT-ND6*	516006
59. Disorders of complex I assembly				
NADH dehydrogenase α subcomplex assembly factor 1 deficiency		AR	*NDUFAF1*	606934
NADH dehydrogenase α subcomplex assembly factor 2 deficiency		AR	*NDUFAF2*	609653
NADH dehydrogenase α subcomplex assembly factor 3 deficiency		AR	*NDUFAF3*	612911
NADH dehydrogenase α subcomplex assembly factor 4 deficiency		AR	*NDUFAF4*	611776
NADH dehydrogenase α subcomplex assembly factor 5 deficiency		AR	*NDUFAF5*	612360
NADH dehydrogenase α subcomplex assembly factor 6 deficiency		AR	*NDUFAF6*	612392
FOXRED1 deficiency		AR	*FOXRED1*	613622
NUBPL deficiency		AR	*NUBPL*	613621
ACAD9 deficiency		AR	*ACAD9*	611103
Transmembrane protein 126B deficiency		AR	*TMEM126B*	615533
60. Disorders of complex II subunits				
Succinate dehydrogenase subunit A deficiency		AR	*SDHA*	600857
Succinate dehydrogenase subunit A deficiency, tumoral phenotype	Hereditary paraganglioma syndrome type 5	AD	*SDHA*	600857
Succinate dehydrogenase subunit B deficiency		AR	*SDHB*	185470
Succinate dehydrogenase subunit B deficiency, tumoral phenotype	Hereditary paraganglioma syndrome type 4; Cowden syndrome type 2	AD	*SDHB*	185470

Succinate dehydrogenase subunit C deficiency, tumoral phenotype	Hereditary paraganglioma syndrome type 3	AD	*SDHC*	602413
Succinate dehydrogenase subunit D deficiency		AR	*SDHD*	602690
Succinate dehydrogenase subunit D deficiency, tumoral phenotype	Hereditary paraganglioma syndrome type 1; Cowden syndrome type 3	AD	*SDHD*	602690
61. Disorders of complex II assembly				
Succinate dehydrogenase complex assembly factor 1 deficiency		AR	*SDHAF1*	612848
Succinate dehydrogenase complex assembly factor 2 deficiency, tumoral phenotype	Hereditary paraganglioma syndrome type 2	AD	*SDHAF2*	613019
62. Disorders of complex III subunits				
UQCRB deficiency		AR	*UQCRB*	191330
UQCRC2 deficiency		AR	*UQCRC2*	191329
See also: Mitochondrial cytochrome c1 deficiency in group 68.				
63. Disorders of complex III assembly				
BCS1L deficiency	GRACILE syndrome; Björnstad syndrome	AR	*BCS1L*	603647
TTC19 deficiency		AR	*TTC19*	613814
UQCC2 deficiency		AR	*UQCC2*	614461
LYRM7 deficiency		AR	*LYRM7*	615831
64. Disorders of complex IV subunits				
Cytochrome c oxidase subunit 1 deficiency		Mit	*MT-CO1*	516030
Cytochrome c oxidase subunit 2 deficiency		Mit	*MT-CO2*	516040
Cytochrome c oxidase subunit 3 deficiency		Mit	*MT-CO3*	516050
Cytochrome c oxidase subunit 4I2 deficiency	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	AR	*COX4I2*	607976
Cytochrome c oxidase subunit 6A1 deficiency	Recessive intermediate Charcot-Marie-Tooth disease type D	AR	*COX6A1*	602072
Cytochrome c oxidase subunit 6B1 deficiency		AR	*COX6B1*	124089

Cytochrome c oxidase subunit 7B deficiency	Linear skin defects with multiple congenital anomalies type 2	XL	*COX7B*	300885
65. Disorders of complex IV assembly and ancillary proteins				
Cytochrome c oxidase assembly factor 6 deficiency		AR	*COA6*	614772
COX10 deficiency		AR	*COX10*	602125
COX15 deficiency		AR	*COX15*	603646
COX20 deficiency		AR	*COX20*	614698
SCO1 deficiency		AR	*SCO1*	603644
SCO2 deficiency		AR	*SCO2*	604272
SURF1 deficiency		AR	*SURF1*	185620
LRPPRC deficiency		AR	*LRPPRC*	607544
TACO1 deficiency		AR	*TACO1*	612958
PET100 deficiency		AR	*PET100*	614770
FASTKD2 deficiency		AR	*FASTKD2*	612322
APOPT1 deficiency		AR	*APOPT1*	616003
See also: Mitochondrial phosphate carrier deficiency in group 57.				
66. Disorders of complex V subunits				
Mitochondrial ATP synthase F1 subunit α deficiency		AR	*ATP5F1A*	164360
Mitochondrial ATP synthase F1 subunit δ deficiency		AR	*ATP5F1D*	603150
Mitochondrial ATP synthase F1 subunit ε deficiency		AR	*ATP5F1E*	606153
Mitochondrial ATP synthase F0 subunit 6 deficiency		Mit	*MT-ATP6*	516060
Mitochondrial ATP synthase F0 subunit 8 deficiency		Mit	*MT-ATP8*	516070
67. Disorders of complex V assembly				
Transmembrane protein 70 deficiency		AR	*TMEM70*	612418
68. Disorders of mitochondrial cytochrome synthesis and incorporation				
Mitochondrial cytochrome b deficiency		Mit	*MT-CYB*	516020
Mitochondrial cytochrome c1 deficiency		AR	*CYC1*	123980
Mitochondrial cytochrome c deficiency	Thrombocytopenia type 4	AD	*CYCS*	123970

Holocytochrome c synthase deficiency	Linear skin defects with multiple congenital anomalies type 1	XLD	*HCCS*	300056
69. Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication				
Mitochondrial DNA polymerase γ catalytic subunit deficiency	Mitochondrial DNA depletion syndrome type 1; Alpers-Huttenlocher syndrome; mitochondrial recessive ataxia syndrome (MIRAS); arPEO type 1; adPEO type 1	AD, AR	*POLG*	174763
Mitochondrial DNA polymerase γ accessory subunit deficiency	adPEO with mitochondrial DNA deletions type 4	AD	*POLG2*	604983
Mitochondrial deoxyguanosine kinase deficiency	Mitochondrial DNA depletion syndrome type 3; arPEO with mitochondrial DNA deletions type 4; noncirrhotic portal hypertension	AR	*DGUOK*	601465
MPV17 deficiency	Mitochondrial DNA depletion syndrome type 6	AR	*MPV17*	137960
TWINKLE mitochondrial DNA helicase deficiency	Mitochondrial DNA depletion syndrome type 7; Perrault syndrome type 5; arPEO with mitochondrial DNA deletions type 5	AR	*TWNK*	606075
Mitochondrial thymidine kinase deficiency	Mitochondrial DNA depletion syndrome type 2	AR	*TK2*	188250
Mitochondrial ribonucleotide reductase small subunit deficiency	Mitochondrial DNA depletion syndrome type 8; adPEO with mitochondrial DNA deletions type 5	AD, AR	*RRM2B*	604712
Thymidine phosphorylase deficiency	Mitochondrial neurogastrointestinal encephalopathy syndrome	AR	*TYMP*	131222
DNA2 helicase deficiency	adPEO with mitochondrial DNA deletions type 6	AD	*DNA2*	601810
Mitochondrial ribonuclease H1 deficiency	arPEO with mitochondrial DNA deletions type 2	AR	*RNASEH1*	604123
Mitochondrial genome maintenance exonuclease 1 deficiency	Mitochondrial DNA depletion syndrome type 11	AR	*MGME1*	615076
FBXL4 deficiency	Mitochondrial DNA depletion syndrome type 13	AR	*FBXL4*	605654
See also: ATP-specific succinyl-CoA ligase β subunit and GTP-specific succinyl-CoA ligase α subunit deficiencies in group 55, and adenine nucleotide translocator deficiency in group 57.				
70. Disorders of mitochondrial transcription and RNA transcript processing				
Mitochondrial RNA import protein deficiency	Combined oxidative phosphorylation deficiency type 13	AR	*PNPT1*	610316
Ribonuclease P 5' tRNA processing enzyme deficiency	Combined oxidative phosphorylation deficiency 30	AR	*TRMT10C*	615423

Ribonuclease Z 3' tRNA processing enzyme deficiency	Combined oxidative phosphorylation deficiency 17	AR	*ELAC2*	605367
Mitochondrial poly(A) polymerase deficiency		AR	*MTPAP*	613669
CCA-adding tRNA-nucleotidyltransferase deficiency	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (severe); retinitis pigmentosa and erythrocytic microcytosis (milder)	AR	*TRNT1*	612907
Mitochondrial methionyl-tRNA formyltransferase deficiency	Combined oxidative phosphorylation deficiency type 15	AR	*MTFMT*	611766
tRNA 5-taurinomethyluridine modifier deficiency	Combined oxidative phosphorylation deficiency type 23	AR	*GTPBP3*	608536
tRNA 5-carboxymethylaminomethyl transferase deficiency	Combined oxidative phosphorylation deficiency type 10	AR	*MTO1*	614667
Pseudouridine synthase 1 deficiency	Myopathy, lactic acidosis, and sideroblastic anemia type 1	AR	*PUS1*	608109
tRNA isopentenyl transferase deficiency		AR	*TRIT1*	617840
tRNA methyltransferase 5 deficiency	Combined oxidative phosphorylation deficiency type 26	AR	*TRMT5*	611023
tRNA 5-methylaminomethyl-2-thiouridylate-methyltransferase deficiency	Transient infantile liver failure	AR	*TRMU*	610230
Mitochondrial RNA-processing endoribonuclease deficiency	Cartilage-hair hypoplasia; metaphyseal dysplasia without hypotrichosis; anauxetic dysplasia type 1	AR	*RMRP*	157660
See also: HSD10 disease in group 14.				
71. Mitochondrial ribosomopathies				
Mitochondrial ribosomal large subunit 3 deficiency	Combined oxidative phosphorylation deficiency type 9	AR	*MRPL3*	607118
Mitochondrial ribosomal large subunit 44 deficiency	Combined oxidative phosphorylation deficiency type 16	AR	*MRPL44*	611849
Mitochondrial ribosomal small subunit 16 deficiency	Combined oxidative phosphorylation deficiency type 2	AR	*MRPS16*	609204
Mitochondrial ribosomal small subunit 22 deficiency	Combined oxidative phosphorylation deficiency type 5	AR	*MRPS22*	605810
Mitochondrial ribosomal small subunit 34 deficiency	Combined oxidative phosphorylation deficiency type 32	AR	*MRPS34*	611994

Mitochondrial ribosomal RNA 12S deficiency		Mit	*MT-RNR1*	561000
Mitochondrial ribosomal RNA 16S deficiency		Mit	*MT-RNR2*	561010
72. Disorders of mitochondrial translation factors				
RMND1 deficiency	Combined oxidative phosphorylation deficiency type 11	AR	*RMND1*	614917
Mitochondrial elongation factor G1 deficiency	Combined oxidative phosphorylation deficiency type 1	AR	*GFM1*	606639
Mitochondrial elongation factor G2 deficiency		AR	*GFM2*	606544
Mitochondrial elongation factor Ts deficiency	Combined oxidative phosphorylation deficiency type 3	AR	*TSFM*	604723
Mitochondrial elongation factor Tu deficiency	Combined oxidative phosphorylation deficiency type 4	AR	*TUFM*	602389
C12orf65 release factor deficiency	Combined oxidative phosphorylation deficiency type 7; autosomal recessive spastic paraplegia type 55	AR	*C12orf65*	613541
73. Disorders of mitochondrial tRNA				
Mitochondrial tRNA(Ala) deficiency		Mit	*MT-TA*	590000
Mitochondrial tRNA(Arg) deficiency		Mit	*MT-TR*	590005
Mitochondrial tRNA(Asn) deficiency		Mit	*MT-TN*	590010
Mitochondrial tRNA(Asp) deficiency		Mit	*MT-TD*	590015
Mitochondrial tRNA(Cys) deficiency		Mit	*MT-TC*	590020
Mitochondrial tRNA(Glu) deficiency		Mit	*MT-TE*	590025
Mitochondrial tRNA(Gln) deficiency		Mit	*MT-TQ*	590030
Mitochondrial tRNA(Gly) deficiency		Mit	*MT-TG*	590035
Mitochondrial tRNA(His) deficiency		Mit	*MT-TH*	590040
Mitochondrial tRNA(Ile) deficiency		Mit	*MT-TI*	590045
Mitochondrial tRNA(Leu) 1 deficiency		Mit	*MT-TL1*	590050
Mitochondrial tRNA(Le) 2 deficiency		Mit	*MT-TL2*	590055
Mitochondrial tRNA(Lys) deficiency		Mit	*MT-TK*	590060
Mitochondrial tRNA(Met) deficiency		Mit	*MT-TM*	590065
Mitochondrial tRNA(Phe) deficiency		Mit	*MT-TF*	590070
Mitochondrial tRNA(Pro) deficiency		Mit	*MT-TP*	590075
Mitochondrial tRNA(Ser) 1 deficiency		Mit	*MT-TS1*	590080

Mitochondrial tRNA(Ser) 2 deficiency		Mit	*MT-TS2*	590085
Mitochondrial tRNA(Thr) deficiency		Mit	*MT-TT*	590090
Mitochondrial tRNA(Trp) deficiency		Mit	*MT-TW*	590095
Mitochondrial tRNA(Tyr) deficiency		Mit	*MT-TY*	590100
Mitochondrial tRNA(Val) deficiency		Mit	*MT-TV*	590105
74. Disorders of mitochondrial tRNA incorporation and recycling				
Mitochondrial alanyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 8; progressive leukoencephalopathy with ovarian failure	AR	*AARS2*	612035
Mitochondrial arginine-tRNA synthetase deficiency	Pontocerebellar hypoplasia type 6	AR	*RARS2*	611524
Mitochondrial asparaginyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 24	AR	*NARS2*	612803
Mitochondrial aspartyl-tRNA synthetase deficiency	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	AR	*DARS2*	610956
Mitochondrial cysteinyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 27	AR	*CARS2*	612800
Mitochondrial glutamyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 12	AR	*EARS2*	612799
Mitochondrial histidyl-tRNA synthetase deficiency	Perrault syndrome type 2	AR	*HARS2*	600783
Mitochondrial isoleucyl-tRNA synthetase deficiency	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia (CAGSSS)	AR	*IARS2*	612801
Mitochondrial leucyl-tRNA synthetase deficiency	Perrault syndrome type 4	AR	*LARS2*	604544
Mitochondrial methionyl-tRNA synthetase deficiency	Autosomal recessive spastic ataxia type 3	AR	*MARS2*	609728
Mitochondrial phenylalanyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 14; autosomal recessive spastic paraplegia type 77	AR	*FARS2*	611592
Mitochondrial seryl-tRNA synthetase deficiency	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis (HUPRA)	AR	*SARS2*	612804
Mitochondrial tyrosyl-tRNA synthetase deficiency	Myopathy, lactic acidosis, and sideroblastic anemia type 2	AR	*YARS2*	610957

Mitochondrial valyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 20	AR	*VARS2*	612802
Mitochondrial tryptophanyl-tRNA synthetase deficiency	Mitochondrial neurodevelopmental disorder with abnormal movements and lactic acidosis, with or without seizures	AR	*WARS2*	604733
Mitochondrial and cytoplasmic glycil-tRNA synthetase deficiency	Charcot-Marie-Tooth disease type 2D; distal hereditary motor neuronopathy type 5A	AD	*GARS*	600287
Mitochondrial and cytoplasmic lysyl-tRNA synthetase deficiency		AR	*KARS*	601421
Peptidyl-tRNA hydrolase 2 deficiency	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	AR	*PTRH2*	608625
75. Disorders of mitochondrial fission				
Dynamin-like protein 1 deficiency	Optic atrophy type 5; encephalopathy due to defective mitochondrial and peroxisomal fission type 1	AD, AR	*DNM1L*	603850
Mitochondrial fission factor deficiency	Encephalopathy due to defective mitochondrial and peroxisomal fission type 2	AR	*MFF*	614785
GDAP1 deficiency	Axonal Charcot-Marie-Tooth type 2K; demyelinating Charcot-Marie-Tooth disease type 4A	AD, AR	*GDAP1*	606598
STAT2 deficiency	Immunodeficiency type 44	AR	*STAT2*	600556
UGO-1 like protein deficiency	Hereditary motor and sensory neuropathy type 6B	AR	*SLC25A46*	610826
76. Disorders of mitochondrial fusion				
OPA1 deficiency	Optic atrophy type 1 (dominant); Behr syndrome (recessive)	AD, AR	*OPA1*	605290
OPA3 deficiency	Optic atrophy type 3 (dominant); 3-methylglutaconic aciduria type 3, Costeff syndrome (recessive)	AD, AR	*OPA3*	606580
Mitofusin 2 deficiency	Axonal Charcot-Marie-Tooth type 2A2	AD, AR	*MFN2*	608507
MSTO1 deficiency	Mitochondrial myopathy and ataxia	AD, AR	*MSTO1*	617619
77. Disorders of mitochondrial phospholipid metabolism				
Acylglycerol kinase deficiency	Sengers syndrome	AR	*AGK*	610345
SERAC1 deficiency	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL)	AR	*SERAC1*	614725
Taffazin deficiency	Barth syndrome	XLR	*TAZ*	300394
PNPLA8 deficiency	Mitochondrial myopathy with lactic acidosis	AR	*PNPLA8*	612123

78. Disorders of mitochondrial protein import				
DNAJC19 deficiency	Dilated cardiomyopathy with ataxia (DCMA syndrome); 3-methylglutaconic aciduria type 5	AR	*DNAJC19*	608977
TIMM8A deficiency	Mohr-Tranebjaerg syndrome	XLR	*TIMM8A*	300356
TIMMDC1 deficiency		AR	*TIMMDC1*	615534
TIMM50 deficiency	3-methylglutaconic aciduria type 9	AR	*TIMM50*	607381
GFER deficiency		AR	*GFER*	600924
MAGMAS deficiency	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type	AR	*PAM16*	614336
See also: Acylglycerol kinase deficiency in group 77.				
79. Disorders of mitochondrial protein quality control				
Mitochondrial processing peptidase alpha deficiency	Autosomal recessive spinocerebellar ataxia type 2	AR	*PMPCA*	613036
Mitochondrial intermediate peptidase deficiency	Combined oxidative phosphorylation deficiency type 31	AR	*MIPEP*	602241
CLPB deficiency	3-methylglutaconic aciduria type 7, with cataracts, neurologic involvement and neutropenia	AR	*CLPB*	616254
CLPP deficiency	Perrault syndrome type 3	AR	*CLPP*	601119
LONP1 deficiency	Cerebral, ocular, dental, auricular, and skeletal (CODAS) syndrome	AR	*LONP1*	605490
HSPA9 deficiency	Sideroblastic anemia type 4; epiphyseal, vertebral, ear, nose, plus associated malformations (EVEN-plus) syndrome	AR	*HSPA9*	600548
HSP60 deficiency	Hypomyelinating leukodystrophy type 4 (recessive); autosomal dominant spastic paraplegia type 13	AD, AR	*HSPD1*	118190
Sacsin deficiency	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	*SACS*	604490
m-AAA protease AFG3L2 subunit deficiency	Autosomal recessive spastic ataxia type 5; spinocerebellar ataxia type 28	AR	*AFG3L2*	604581
Paraplegin deficiency	Spastic paraplegia type 7	AD, AR	*SPG7*	602783
HTRA2 deficiency	3-methylglutaconic aciduria type 8	AR	*HTRA2*	606441
Parkin deficiency	Early-onset Parkinson disease type 2	AR	*PRKN*	602544
PINK1 deficiency	Early-onset Parkinson disease type 6	AR	*PINK1*	608309
USP9X deficiency	X-linked mental retardation type 99	XL	*USP9X*	300072

Valosin-containing protein superactivity	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia type 1	AD	*VCP*	601023
See also: X-prolyl aminopeptidase 3 deficiency in group 16.				
80. Other disorders of mitochondrial homeostasis				
Mitochondrial inorganic pyrophosphatase 2 deficiency		AR	*PPA2*	609988
Sideroflexin 4 deficiency	Combined oxidative phosphorylation deficiency type 18	AR	*SFXN4*	615564
AIFM1 deficiency	Combined oxidative phosphorylation deficiency type 6; Cowchock syndrome	XLR	*AIFM1*	300169
ATAD3A deficiency	Harel-Yoon syndrome	AD, AR	*ATAD3A*	612316
Transmembrane protein 126A deficiency	Optic atrophy type 7	AR	*TMEM126A*	612988
C1q binding protein deficiency	Combined oxidative phosphorylation deficiency type 33	AR	*C1QBP*	601269
Trafficking kinesin-binding protein 1 deficiency		AR	*TRAK1*	608112
Mitochondrial calcium uniporter deficiency	Myopathy with extrapyramidal signs	AR	*MICU1*	605084
Nogo-interacting mitochondrial protein deficiency	Optic atrophy type 10	AR	*RTN4IP1*	610502
81. Primary CoQ10 deficiencies				
Prenyl diphosphate synthase subunit 1 deficiency	Primary coenzyme Q10 deficiency type 2	AR	*PDSS1*	607429
Prenyl diphosphate synthase subunit 2 deficiency	Primary coenzyme Q10 deficiency type 3	AR	*PDSS2*	610564
COQ2 deficiency	Primary coenzyme Q10 deficiency type 1	AR	*COQ2*	609825
COQ4 deficiency	Primary coenzyme Q10 deficiency type 7	AR	*COQ4*	612898
COQ6 deficiency	Primary coenzyme Q10 deficiency type 6	AR	*COQ6*	614647
COQ7 deficiency	Primary coenzyme Q10 deficiency type 8	AR	*COQ7*	601683
COQ8A deficiency	Primary coenzyme Q10 deficiency type 4; ADCK3 deficiency	AR	*COQ8A*	606980
COQ8B deficiency	Nephrotic syndrome type 9; ADCK4 deficiency	AR	*COQ8B*	615567
COQ9 deficiency	Primary coenzyme Q10 deficiency type 5	AR	*COQ9*	612837

E. DISORDERS OF LIPIDS				
82. Disorders of carnitine metabolism				
Primary carnitine deficiency		AR	*SLC22A5*	603377
Carnitine palmitoyltransferase 1A deficiency		AR	*CPT1A*	600528
Carnitine palmitoyltransferase 2 deficiency		AR	*CPT2*	600650
Carnitine-acylcarnitine translocase deficiency		AR	*SLC25A20*	613698
ε-N-trimethyllysine hydroxylase deficiency		XLR	*TMHLE*	300777
83. Disorders of fatty acid oxidation and transport				
Short-chain acyl-CoA dehydrogenase deficiency		AR	*ACADS*	606885
Medium-chain acyl-CoA dehydrogenase deficiency		AR	*ACADM*	607008
Very long-chain acyl-CoA dehydrogenase deficiency		AR	*ACADVL*	609575
Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	Familial hyperinsulinemic hypoglycemia type 4	AR	*HADH*	601609
Trifunctional protein α subunit deficiency	Long-chain hydroxyacyl-CoA dehydrogenase or complete mitochondrial trifunctional protein deficiency	AR	*HADHA*	600890
Trifunctional protein β subunit deficiency	Complete mitochondrial trifunctional protein deficiency	AR	*HADHB*	143450
Fatty acid transport protein 4 deficiency	Ichthyosis prematurity syndrome	AR	*SLC27A4*	604194
Docosahexanoic acid transporter deficiency	Autosomal recessive primary microcephaly type 15	AR	*MFSD2A*	614397
TANGO2 deficiency	Metabolic encephalomyopathic crises associated with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration (MECRCN)	AR	*TANGO2*	616830
See also: ACAD9 deficiency in group 59.				
84. Disorders of ketone body metabolism				
Mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency		AR	*HMGCS2*	600234

Succinyl-CoA:3-oxoacid-CoA transferase deficiency		AD, AR	*OXCT1*	601424
Mitochondrial acetoacetyl-CoA thiolase deficiency	β-ketothiolase deficiency; α-methylacetoacetic aciduria	AR	*ACAT1*	607809
Monocarboxylate transporter 1 deficiency		AD, AR	*SLC16A1*	600682
Monocarboxylate transporter 1 superactivity	Familial hyperinsulinemic hypoglycemia type 7	AD	*SLC16A1*	600682
See also: 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency in group 14.				
85. Disorders of fatty acid synthesis and elongation				
Mitochondrial enoyl-CoA reductase deficiency	Mitochondrial enoyl-CoA reductase protein-associated neurodegeneration (MEPAN)	AR	*MECR*	608205
Very long-chain fatty acid elongase 1 deficiency	Ichthyotic keratoderma, spasticity, hypomyelination and dysmorphic features (IKSHD)	AD	*ELOVL1*	611813
Very long-chain fatty acid elongase 4 deficiency, neurologic phenotype	Pseudo-Sjögren-Larsson syndrome (recessive); spinocerebellar ataxia type 34 (dominant)	AD, AR	*ELOVL4*	605512
Very long-chain fatty acid elongase 4 deficiency, retinal phenotype	Stargardt disease type 3	AD	*ELOVL4*	605512
Very long-chain fatty acid elongase 5 deficiency	Spinocerebellar ataxia type 38	AD	*ELOVL5*	611805
86. Disorder of fatty aldehyde metabolism				
Fatty aldehyde dehydrogenase deficiency	Sjögren-Larsson syndrome	AR	*ALDH3A2*	609523
87. Disorders of glycerol metabolism				
Glycerol kinase deficiency		XLR	*GK*	300474
Glycerate kinase deficiency	D-glyceric aciduria	AR	*GLYCTK*	610516
Aquaporin 7 deficiency	Hyperglyceroluria with mild platelet secretion defect	AR	*AQP7*	602974
See also: Cytosolic glycerol-3-phosphate dehydrogenase deficiency in group 57.				
88. Disorders of cytoplasmic triglyceride metabolism				
Long-chain fatty acid-CoA ligase 4 deficiency	X-linked mental retardation 63	XL	*ACSL4*	300157
Lysophosphatidic acid acyltransferase deficiency	Congenital generalized lipodystrophy type 1, Berardinelli-Seip syndrome	AR	*AGPAT2*	603100
Lipin 1 deficiency		AR	*LPIN1*	605518
Lipin 2 deficiency	Majeed syndrome	AR	*LPIN2*	605519
Diacylglycerol acyltransferase deficiency	Congenital diarrhea type 7	AR	*DGAT1*	604900

CGI-58 deficiency	Chanarin-Dorfman syndrome; neutral lipid storage disease with ichthyosis	AR	*ABHD5*	604780
Adipose triglyceride lipase deficiency	Neutral lipid storage disease with myopathy	AR	*PNPLA2*	609059
Perilipin 1 deficiency	Familial partial lipodystrophy type 4	AD	*PLIN1*	170290
Hormone-sensitive lipase deficiency	Familial partial lipodystrophy type 6	AR	*LIPE*	151750
89. Disorders of non-mitochondrial phospholipid metabolism				
Choline kinase β deficiency	Congenital muscular dystrophy, megaconial type	AR	*CHKB*	612395
Phosphocholine cytidylyltransferase 1α deficiency, retinoskeletal phenotype		AR	*PCYT1A*	123695
Phosphocholine cytidylyltransferase 1α deficiency, lipodystrophy phenotype		AR	*PCYT1A*	123695
Phosphatidylserine synthase 1 superactivity	Lenz-Majewski syndrome	AD	*PTDSS1*	612792
Phospholipase A2 group 6 deficiency	Infantile neuroaxonal dystrophy; Seitelberger disease; neurodegeneration with brain iron accumulation type 2B	AR	*PLA2G6*	603604
DDHD1 deficiency	Autosomal recessive spastic paraplegia type 28	AR	*DDHD1*	614603
DDHD2 deficiency	Autosomal recessive spastic paraplegia type 54	AR	*DDHD2*	615003
PNPLA6 deficiency	Autosomal recessive spastic paraplegia type 39; Oliver-McFarlane syndrome; Boucher-Neuhauser syndrome; Laurence-Moon syndrome	AR	*PNPLA6*	603197
CYP2U1 deficiency	Autosomal recessive spastic paraplegia type 56	AR	*CYP2U1*	610670
ABHD12 deficiency	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract (PHARC) syndrome,	AR	*ABHD12*	613599
Diacylglycerol kinase ε deficiency	Nephrotic syndrome type 7; atypical hemolytic uremic syndrome type 7	AR	*DGKE*	601440
See also: Lipin 1 and lipin 2 deficiencies in group 88.				
90. Disorders of non-lysosomal sphingolipid metabolism				
Serine palmitoyltransferase subunit 1 deficiency	Hereditary sensory and autonomic neuropathy type 1A	AD	*SPTLC1*	605712
Serine palmitoyltransferase subunit 2 deficiency	Hereditary sensory and autonomic neuropathy type 1C	AD	*SPTLC2*	605713
Ceramide synthase 3 deficiency	Autosomal recessive congenital ichthyosis type 9	AR	*CERS3*	615276
CYP4F22 omega hydroxylase deficiency	Autosomal recessive congenital ichthyosis type 5	AR	*CYP4F22*	611495

Nonlysosomal glucosylceramidase deficiency	Autosomal recessive spastic paraplegia type 46	AR	*GBA2*	609471
Fatty acid 2-hydroxylase deficiency	Autosomal recessive spastic paraplegia type 35; fatty acid hydroxylase-associated neurodegeneration (FAHN)	AR	*FA2H*	611026
Sphingosine-1-phosphate lyase deficiency		AR	*SGPL1*	603729
Ceramide transfer protein superactivity	Autosomal dominant mental retardation type 34	AD	*COL4A3BP*	604677
See also: GM3 synthase, GM2/GD2 synthase, and GB3 synthase deficiencies in group 123.				
91. Disorders of eicosanoid metabolism				
Thromboxane synthase deficiency	Ghosal hematodiaphyseal syndrome	AR	*TBXAS1*	274180
15-hydroxy prostaglandin dehydrogenase deficiency	Primary hypertrophic osteoarthropathy type 1	AR	*HPGD*	601688
Prostaglandin transporter deficiency	Primary hypertrophic osteoarthropathy type 2	AR	*SLCO2A1*	601460
92. Disorders of palmitoylation				
ZDHHC9 palmitoyltransferase deficiency	X-linked mental retardation, Raymond type	XLR	*ZDHHC9*	300646
Porcupine palmitoyltransferase deficiency	Goltz syndrome, focal dermal hypoplasia	XLD	*PORCN*	300651
Palmitoyl-protein thioesterase 1 deficiency	Neuronal ceroid lipofuscinosis type 1; Santavuori-Haltia disease	AR	*PPT1*	600722
93. Disorders of phosphoinositide metabolism				
Phosphatidylinositol 3,5-bisphosphate-5-phosphatase deficiency, neurologic phenotype	Amyotrophic lateral sclerosis type 11 (dominant); Charcot-Marie-Tooth disease type 4J (recessive)	AD, AR	*FIG4*	609390
Phosphatidylinositol 3,5-bisphosphate-5-phosphatase deficiency, neuroskeletal phenotype	Yunis-Varon syndrome	AR	*FIG4*	609390
Phosphatidylinositol 4,5-bisphosphate-5-phosphatase deficiency	Lowe syndrome, Dent disease type 2	XLR	*OCRL*	300535
Synaptojanin 1 deficiency	Early infantile epileptic encephalopathy type 53; early-onset Parkinson disease type 20	AR	*SYNJ1*	604297
Myotubularin 1 deficiency	X-linked myotubular myopathy	XLR	*MTM1*	300415
Myotubularin-related protein 2 deficiency	Charcot-Marie-Tooth disease type 4B1	AR	*MTMR2*	603557
Myotubularin-related protein 2 regulatory protein deficiency	Charcot-Marie-Tooth disease type 4B2	AR	*SBF2*	607697

Myotubularin-related protein 2 activator deficiency	Charcot-Marie-Tooth disease type 4B3	AR	*SBF1*	603560
Catalytic phosphatidylinositol 3-kinase α subunit superactivity		AD, somatic	*PIK3CA*	171834
Catalytic phosphatidylinositol 3-kinase δ subunit superactivity	Immunodeficiency type 14	AD	*PIK3CD*	602839
Phosphatidylinositol 3-kinase regulatory subunit 1 deficiency	SHORT syndrome, immunodeficiency type 36	AD	*PIK3R1*	171833
Phosphatidylinositol 3-kinase regulatory subunit 2 superactivity	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome type 1	AD	*PIK3R2*	603157
Phosphatidylinositol-3-phosphate 5-kinase deficiency	Corneal fleck dystrophy	AD	*PIKFYVE*	609414
Phosphatidylinositol 4-phosphate 5-kinase deficiency	Lethal congenital contractural syndrome type 3	AR	*PIP5K1C*	606102
Phosphatidylinositol 3,4,5-trisphosphate 3-phosphatase deficiency	PTEN hamartoma tumor syndrome	AD	*PTEN*	601728
Phosphatidylinositol 3,4,5-trisphosphate 5-phosphatase deficiency	Opsismodysplasia	AR	*INPP1L*	600829
Inositol polyphosphate 5-phosphatase deficiency	Joubert syndrome type 1	AR	*INPP5E*	613037
Phosphatidylinositol 4,5-bisphosphate phospholipase C β1 deficiency	Early infantile epileptic encephalopathy type 12	AR	*PLCB1*	607120
Phosphatidylinositol 4,5-bisphosphate phospholipase C β4 deficiency	Auriculocondylar syndrome type 2	AD, AR	*PLCB4*	600810
Phosphatidylinositol 4,5-bisphosphate phospholipase C γ2 deficiency	Autoinflammation and PLCG2-associated antibody deficiency and immune dysregulation (APLAID); familial cold autoinflammatory syndrome type 3	AD	*PLCG2*	600220
Phosphatidylinositol 4,5-bisphosphate phospholipase C δ1 deficiency	Nonsyndromic congenital nail disorder type 3; leukonychia totalis and/or partialis	AD, AR	*PLCD1*	602142
Phosphatidylinositol 4,5-bisphosphate phospholipase C ε1 deficiency	Nephrotic syndrome type 3	AR	*PLCE1*	608414
Inositol 1,4,5-triphosphate receptor type 1 deficiency	Gillespie syndrome; congenital non-progressive cerebellar ataxia; spinocerebellar ataxia type 15	AD, AR	*ITPR1*	147265
94. Disorders of lipoprotein metabolism				
LDL receptor deficiency	Familial hypercholesterolemia	AD, AR	*LDLR*	606945

LDL receptor adaptor protein 1 deficiency		AR	*LDLRAP1*	605747
Hypercholesterolemia due to ligand-defective apo B		AD	*APOB*	107730
Apolipoprotein B deficiency	Familial hypobetalipoproteinemia type 1; normotriglyceridemic hypobetalipoproteinemia	AR	*APOB*	107730
PCSK9 superactivity	Familial hypercholesterolemia type 3	AD	*PCSK9*	607786
PCSK9 deficiency		AD	*PCSK9*	607786
STAP1 deficiency	Familial hypercholesterolemia type 4	AD	*STAP1*	604298
Sitosterolemia due to ABCG5 deficiency		AR	*ABCG5*	605459
Sitosterolemia due to ABCG8 deficiency		AR	*ABCG8*	605460
Angiopoietin-like 3 deficiency	Familial hypobetalipoproteinemia type 2; combined familial hypolipidemia	AR	*ANGPTL3*	604774
Microsomal triglyceride transfer protein deficiency	Abetalipoproteinemia	AR	*MTTP*	157147
Chylomicron retention disease	Anderson disease	AR	*SAR1B*	607690
Apolipoprotein E deficiency	Dysbetalipoproteinemia; hyperlipoproteinemia type 3	AR	*APOE*	107741
Apolipoprotein E superactivity	Inherited lipemic splenomegaly; sea-blue histiocytosis	AD	*APOE*	107741
Lipoprotein glomerulopathy		AD	*APOE*	107741
Lipoprotein lipase deficiency	Hyperlipoproteinemia type 1A	AR	*LPL*	609708
Apolipoprotein C2 deficiency	Hyperlipoproteinemia type 1B	AR	*APOC2*	608083
GPIHBP1 deficiency	Hyperlipoproteinemia type 1D	AR	*GPIHBP1*	612757
Hepatic lipase deficiency		AR	*LIPC*	151670
Lipase maturation factor 1 deficiency	Combined lipase deficiency	AR	*LMF1*	611761
Apolipoprotein A5 deficiency	Late-onset hyperchylomicronemia; hyperlipoproteinemia type 5	AD	*APOA5*	606368
Lecithin:cholesterol acyltransferase deficiency	Norum disease (severe); fish-eye disease (milder)	AR	*LCAT*	606967
Tangier disease	Analaphalipoproteinemia	AR	*ABCA1*	600046
Apolipoprotein A1 deficiency	Hypoalphalipoproteinemia	AD	*APOA1*	107680
Hereditary apolipoprotein A1-related amyloidosis		AD	*APOA1*	107680
Cholesteryl ester transfer protein deficiency	Hyperalphalipoproteinemia type 1	AD	*CETP*	118470

Apolipoprotein C3 deficiency	Hyperalphalipoproteinemia type 2	AD	*APOC3*	107720
95. Disorders of cholesterol biosynthesis				
Mevalonate kinase deficiency	Mevalonic aciduria (severe); hyper-IgD syndrome (milder)	AR	*MVK*	251170
Mevalonate kinase deficiency, porokeratosis phenotype	Porokeratosis type 3	AD	*MVK*	251170
Phosphomevalonate kinase deficiency	Porokeratosis type 1	AD	*PMVK*	607622
Mevalonate pyrophosphate decarboxylase deficiency	Porokeratosis type 7	AD	*MVD*	603236
Farnesyl diphosphate synthase deficiency	Porokeratosis type 9	AD	*FDPS*	134629
Sterol C14 reductase deficiency	Hydrops-ectopic calcification-moth-eaten (HEM) dysplasia, Greenberg dysplasia (recessive); Pelger-Huët anomaly (dominant)	AD, AR	*LBR*	600024
Sterol-C4-methyl oxidase deficiency	Microcephaly, congenital cataract, and psoriasiform dermatitis	AR	*MSMO1*	607545
X-linked dominant sterol-4-alpha-carboxylate 3-dehydrogenase deficiency	Congenital hemidysplasia with ichythyosiform erythroderma and limb defects (CHILD) syndrome	XLD	*NSDHL*	300275
X-linked recessive sterol-4-alpha-carboxylate 3-dehydrogenase deficiency	CK syndrome	XLR	*NSDHL*	300275
X-linked dominant sterol Δ8-Δ7 isomerase deficiency	X-linked dominant chondrodysplasia punctata type 2; Conradi-Hünermann-Happle syndrome	XLD	*EBP*	300205
X-linked recessive sterol Δ8-Δ7 isomerase deficiency	Male EBP disorder with neurologic defects (MEND syndrome)	XLR	*EBP*	300205
Sterol Δ5-desaturase deficiency	Lathosterolosis	AR	*SC5D*	602286
24-dehydrocholesterol reductase deficiency	Desmosterolosis	AR	*DHCR24*	606418
7-dehydrocholesterol reductase deficiency	Smith-Lemli-Opitz syndrome; RSH syndrome	AR	*DHCR7*	602858
96. Disorders of steroid metabolism				
21-hydroxylase deficiency		AR	*CYP21A2*	613815
11-β-hydroxylase deficiency		AR	*CYP11B1*	610613
11-β-hydroxylase superactivity	Glucocorticoid remediable aldosteronism	AD	*CYP11B1*	610613
3-β-hydroxysteroid dehydrogenase deficiency		AR	*HSD3B2*	613890
17-hydroxylase/17,20-lyase deficiency		AR	*CYP17A1*	609300

Steroidogenic acute regulatory protein deficiency	Lipoid adrenal hyperplasia	AR	*STAR*	600617
Cytochrome P450 oxidoreductase deficiency		AR	*POR*	124015
Steroid 18-hydroxylase deficiency	Aldosterone synthase deficiency; corticosterone methyloxidase type 1 deficiency	AR	*CYP11B2*	124080
Steroid 18-oxidase deficiency	Corticosterone methyloxidase type 2 deficiency	AR	*CYP11B2*	124080
Hexose-6-phosphate dehydrogenase deficiency	Cortisone reductase deficiency type 1	AR	*H6PD*	138090
11-β-hydroxysteroid dehydrogenase deficiency	Cortisone reductase deficiency type 2	AD	*HSD11B1*	600713
Glucocorticoid receptor deficiency	Glucocorticoid resistance	AD	*NR3C1*	138040
ACTH receptor deficiency	Melanocortin-2 receptor deficiency; hereditary glucocorticoid deficiency type 1	AR	*MC2R*	607397
Melanocortin-2 receptor accessory protein deficiency	Hereditary glucocorticoid deficiency type 2	AR	*MRAP*	609196
Aromatase deficiency		AR	*CYP19A1*	107910
Aromatase superactivity		AD	*CYP19A1*	107910
Estrogen receptor deficiency	Estrogen resistance	AR	*ESR1*	133430
Side-chain cleavage enzyme deficiency	Desmolase deficiency	AR	*CYP11A1*	118485
17-β-hydroxysteroid dehydrogenase deficiency	17-ketosteroid reductase deficiency; male pseudohermaphroditism with gynecomastia	AR	*HSD17B3*	605573
3-α-hydroxysteroid dehydrogenase type 3 deficiency		AR	*AKR1C2*	600450
11-β-hydroxysteroid dehydrogenase type 2 deficiency	Apparent mineralocorticoid excess	AR	*HSD11B2*	614232
Steroid 5-α-reductase 2 deficiency		AR	*SRD5A2*	607306
Androgen receptor deficiency	Androgen insensitivity syndrome	XLR	*AR*	313700
X-linked spinal and bulbar muscular atrophy	Kennedy disease	XLR	*AR*	313700
Steroid sulfatase deficiency	X-linked ichthyosis	XLR	*STS*	300747
See also: Cytochrome b5 deficiency in group 98.				
97. Disorders of bile acid synthesis				
3β-Hydroxy-Δ5-C27-steroid oxidoreductase deficiency	Congenital bile acid synthesis defect type 1	AR	*HSD3B7*	607764

Δ4-3-oxosteroid 5β-reductase deficiency	Congenital bile acid synthesis defect type 2	AR	*AKR1D1*	604741
Oxysterol 7α-hydroxylase deficiency	Congenital bile acid synthesis defect type 3; autosomal recessive spastic paraplegia type 5A	AR	*CYP7B1*	603711
Sterol 27-hydroxylase deficiency	Cerebrotendinous xanthomatosis	AR	*CYP27A1*	606530
α-methylacyl-CoA racemase deficiency	Congenital bile acid synthesis defect type 4	AR	*AMACR*	604489
Peroxisomal branched-chain acyl-CoA oxidase deficiency	Congenital bile acid synthesis defect type 6	AR	*ACOX2*	601641
Bile acid-CoA:amino acid N-acyltransferase deficiency		AR	*BAAT*	602938
F. DISORDERS OF TETRAPYRROLES				
98. Disorders of heme metabolism				
δ-aminolevulinic acid synthase deficiency	X-linked recessive sideroblastic anemia type 1	XLR	*ALAS2*	301300
δ-aminolevulinic acid synthase superactivity	X-linked protoporphyria	XLD	*ALAS2*	301300
δ-aminolevulinic acid dehydratase deficiency		AR	*ALAD*	125270
Porphobilinogen deaminase deficiency	Acute intermittent porphyria	AD, AR	*HMBS*	609806
Uroporphyrinogen III synthase deficiency	Congenital erythropoietic porphyria; Gunther disease	AR	*UROS*	606938
Uroporphyrinogen decarboxylase deficiency	Porphyria cutanea tarda type 2 (dominant); hepatoerythropoietic porphyria (recessive)	AD, AR	*UROD*	613521
Coproporphyrinogen oxidase deficiency	Hereditary coproporphyria	AD	*CPOX*	612732
Harderoporphyria		AR	*CPOX*	612732
Protoporphyrinogen oxidase deficiency	Variegate porphyria	AD, AR	*PPOX*	600923
Ferrochelatase deficiency	Erythropoietic protoporhyria	AR	*FECH*	612386
GATA1 deficiency		XLR	*GATA1*	305371
Mitochondrial porphyrin transporter deficiency	Familial pseudohyperkalemia type 2; dyschromatosis universalis hereditaria type 3	AD	*ABCB6*	605452
NADH-cytochrome b5 reductase deficiency	NADH diaphorase deficiency	AR	*CYB5R3*	613213
Cytochrome b5 deficiency		AR	*CYB5A*	613218
Heme oxygenase 1 deficiency		AR	*HMOX1*	141250
Biliverdin reductase α deficiency	Hyperbiliverdinemia	AD, AR	*BLVRA*	109750
99. Disorders of bilirubin metabolism and biliary transport				
UDP-glucuronosyltransferase A1 deficiency	Crigler-Najjar syndrome (severe); Gilbert syndrome (milder)	AR	*UGT1A1*	191740

Canalicular bilirubin glucuronide transporter deficiency	Dubin-Johnson syndrome	AR	*ABCC2*	601107
Rotor syndrome		Digenic	*SLCO1B1* + *SLCO1B3*	604843 + 605495
Phosphatidylserine translocator deficiency	Progressive familial intrahepatic cholestasis type 1 (severe); benign recurrent intrahepatic cholestasis type 1 (milder)	AR	*ATP8B1*	602397
Bile salt export pump deficiency	Progressive familial intrahepatic cholestasis type 2 (severe); benign recurrent intrahepatic cholestasis type 2 (milder)	AR	*ABCB11*	603201
Phosphatidylcholine translocator deficiency	Progressive familial intrahepatic cholestasis type 3 (severe); low phospholipid-associated cholelithiasis (milder)	AD, AR	*ABCB4*	171060
Bile acid receptor deficiency	Progressive familial intrahepatic cholestasis type 5	AR	*NR1H4*	603826
Apical bile salt transporter deficiency	Primary bile acid malabsorption	AR	*SLC10A2*	601295
See also: Disorders of cobalamin metabolism (group 26).				
G. STORAGE DISORDERS				
100. Disorders of autophagy				
EPG5 deficiency	Vici syndrome	AR	*EPG5*	615068
WDR45 deficiency	Neurodegeneration with brain iron accumulation type 5; static encephalopathy of childhood with neurodegeneration in adulthood (SENDA); β-propeller protein-associated neurodegeneration (BPAN)	XLD	*WDR45*	300526
SNX14 deficiency	Autosomal recessive spinocerebellar ataxia type 20	AR	*SNX14*	616105
Spatacsin deficiency	Autosomal recessive spastic paraplegia type 11; axonal Charcot-Marie-Tooth disease type 2X; juvenile amyotrophic lateral sclerosis type 5	AR	*SPG11*	610844
Spastizin deficiency	Autosomal recessive spastic paraplegia type 15; Kjellin syndrome	AR	*ZFYVE26*	612012
Adaptor-related protein complex 5 ζ-1 subunit deficiency	Autosomal recessive spastic paraplegia type 48	AR	*AP5Z1*	613653
TECPR2 deficiency	Autosomal recessive spastic paraplegia type 49	AR	*TECPR2*	615000
TBK1 deficiency	Frontotemporal dementia and/or amyotrophic lateral sclerosis type 4	AD	*TBK1*	604834

RAB7 deficiency	Charcot-Marie-Tooth disease type 2B	AD	*RAB7A*	602298
101. Neuronal ceroid lipofuscinosis				
Tripeptidyl-peptidase 1 deficiency	Neuronal ceroid lipofuscinosis type 2, Jansky-Bielchowsky disease (severe), autosomal recessive spinocerebellar ataxia type 7 (milder)	AR	*TPP1*	607998
Neuronal ceroid lipofuscinosis type 3	Vogt-Spielmeyer disease	AR	*CLN3*	607042
Neuronal ceroid lipofuscinosis type 4 (Parry type)	Autosomal dominant Kufs disease	AD	*DNAJC5*	611203
Neuronal ceroid lipofuscinosis type 5		AR	*CLN5*	608102
Neuronal ceroid lipofuscinosis type 6	Autosomal recessive Kufs disease type A	AR	*CLN6*	606725
Neuronal ceroid lipofuscinosis type 7	Macular dystrophy with central cone involvement (milder)	AR	*MFSD8*	611124
Neuronal ceroid lipofuscinosis type 8		AR	*CLN8*	607837
Cathepsin D deficiency	Neuronal ceroid lipofuscinosis type 10	AR	*CTSD*	116840
Progranulin deficiency	Frontotemporal lobar degeneration with TDP-43 inclusions (dominant); neuronal ceroid lipofuscinosis type 11 (recessive)	AD, AR	*GRN*	138945
ATP13A2 deficiency	Neuronal ceroid lipofuscinosis type 12; Kufor-Rakeb syndrome; Parkinson disease type 9; autosomal recessive spastic paraplegia type 78	AR	*ATP13A2*	610513
Cathepsin F deficiency	Autosomal recessive Kufs disease type B	AR	*CTSF*	603539
Neuronal ceroid lipofuscinosis type 14	Progressive myoclonic epilepsy type 3	AR	*KCTD7*	611725
See also: Palmitoyl-protein thioesterase 1 deficiency in group 92.				
102. Sphingolipidoses				
Glucocerebrosidase deficiency	Gaucher disease	AR	*GBA*	606463
Atypical Gaucher disease due to saposin C deficiency		AR	*PSAP*	176801
Acid sphingomyelinase deficiency	Niemann-Pick type A (severe); Niemann-Pick type B (milder)	AR	*SMPD1*	607608
β-galactosidase deficiency, GM1 gangliosidosis phenotype		AR	*GLB1*	611458
β-hexosaminidase α-subunit deficiency	GM2 gangliosidosis, B variant (Tay-Sachs disease)	AR	*HEXA*	606869
β-hexosaminidase β-subunit deficiency	GM2 gangliosidosis, O variant (Sandhoff disease)	AR	*HEXB*	606873
GM2 activator protein deficiency	GM2 gangliosidosis, AB variant	AR	*GM2A*	613109
β-galactosylceramidase deficiency	Globoid cell leukodystrophy; Krabbe disease	AR	*GALC*	245200

Atypical Krabbe disease due to saposin A deficiency		AR	*PSAP*	176801
Arylsulfatase A deficiency	Metachromatic leukodystrophy	AR	*ARSA*	607574
Metachromatic leukodystrophy due to saposin B deficiency		AR	*PSAP*	176801
Formyl-glycine generating enzyme deficiency	Multiple sulfatase deficiency	AR	*SUMF1*	607939
α-Galactosidase A deficiency	Fabry disease	XL	*GLA*	300644
Acid ceramidase deficiency, inflammatory phenotype	Farber disease	AR	*ASAH1*	613468
Acid ceramidase deficiency, primary neurologic phenotype	Spinal muscular atrophy with progressive myoclonic epilepsy	AR	*ASAH1*	613468
Combined saposin deficiency	Prosaposin deficiency	AR	*PSAP*	176801
103. Oligosaccharidoses				
α-neuraminidase deficiency	Sialidosis	AR	*NEU1*	608272
Cathepsin A deficiency	Galactosialidosis	AR	*CTSA*	613111
α-mannosidase deficiency	α-mannosidosis	AR	*MAN2B1*	609458
β-mannosidase deficiency	β-mannosidosis	AR	*MANBA*	609489
α-N-acetylgalactosaminidase deficiency	Schindler disease; Kanzaki disease (milder)	AR	*NAGA*	104170
α-fucosidase deficiency	α-fucosidosis	AR	*FUCA1*	612280
Aspartylglucosaminidase deficiency	Aspartylglucosaminuria	AR	*AGA*	613228
104. Mucolipidoses				
UDP-N-acetylglucosamine-1-phosphotransferase α/β subunit deficiency	Mucolipidosis type 2, I-cell disease (severe); mucolipidosis type 3 α/β, pseudo-Hurler polydystrophy (milder)	AR	*GNPTAB*	607840
UDP-N-acetylglucosamine-1-phosphotransferase γ subunit deficiency	Mucolipidosis type 3 γ	AR	*GNPTG*	607838
Mucolipin 1 deficiency	Mucolipidosis type 4	AR	*MCOLN1*	605248
105. Mucopolysaccharidoses				
α-iduronidase deficiency	Mucopolysaccharidosis type 1H, Hurler syndrome (severe); mucopolysaccharidosis type 1S, Scheie syndrome (milder)	AR	*IDUA*	252800
Iduronate sulfatase deficiency	Mucopolysaccharidosis type 2; Hunter syndrome	XLR	*IDS*	300823
Heparan N-sulfatase deficiency	Mucopolysaccharidosis type 3A; Sanfilippo syndrome type A	AR	*SGSH*	605270

N-acetylglucosaminidase deficiency	Mucopolysaccharidosis type 3B; Sanfilippo syndrome type B	AR	*NAGLU*	609701
Heparan-α-glucosaminide N-acetyltransferase deficiency	Mucopolysaccharidosis type 3C, Sanfilippo syndrome type C (severe), retinitis pigmentosa type 73 (milder)	AR	*HGSNAT*	610453
N-acetylglucosamine 6-sulfatase deficiency	Mucopolysaccharidosis type 3D; Sanfilippo syndrome type D	AR	*GNS*	607664
N-acetylgalactosamine 6-sulfatase deficiency	Mucopolysaccharidosis type 4A; Morquio syndrome type A	AR	*GALNS*	612222
β-galactosidase deficiency, Morquio syndrome phenotype	Mucopolysaccharidosis type 4B	AR	*GLB1*	611458
N-acetylgalactosamine 4-sulfatase deficiency	Mucopolysaccharidosis type 6; Maroteaux-Lamy syndrome; arylsulfatase B deficiency	AR	*ARSB*	611542
β-glucuronidase deficiency	Mucopolysaccharidosis type 7; Sly syndrome	AR	*GUSB*	611499
Hyaluronidase deficiency	Mucopolysaccharidosis type 9; Natowicz syndrome	AR	*HYAL1*	607071
Mucopolysaccharidosis-plus syndrome		AR	*VPS33A*	610034
106. Disorders of lysosomal cholesterol metabolism				
Niemann-Pick disease type C1		AR	*NPC1*	607623
Niemann-Pick disease type C2		AR	*NPC2*	601015
Lysosomal acid lipase deficiency	Wolman disease (severe); cholesteryl ester storage disease (milder)	AR	*LIPA*	613497
107. Disorders of lysosomal transport or sorting				
Cystinosin deficiency	Cystinosis	AR	*CTNS*	606272
Sialin deficiency	Infantile sialic acid storage disease (severe); Salla disease (milder)	AR	*SLC17A5*	604322
Glucocerebrosidase receptor deficiency	Progressive myoclonic epilepsy type 4; action myoclonus-renal failure syndrome	AR	*SCARB2*	602257
See also: Methylmalonic aciduria and homocystinuria, cblF and cblJ types in group 26, and mucolipidosis type 4 in group 104.				
108. Disorders of lysosomal protein degradation				
Cathepsin K deficiency	Pycnodysostosis	AR	*CTSK*	601105
Cathepsin C deficiency	Papillon-Lefevre syndrome; Haim-Munk syndrome	AR	*CTSC*	602365
See also: Cathepsin D and F deficiencies in group 101, and cathepsin A deficiency in group 103.				
H. DISORDERS OF PEROXISOMES AND OXALATE				
109. Disorders of plasmalogen synthesis				
PTS2 receptor deficiency	Rhizomelic chondrodysplasia punctata type 1 (severe); classic Refsum disease type 2 (milder)	AR	*PEX7*	601757

Glycerone 3-phosphate acyltransferase deficiency	Rhizomelic chondrodysplasia punctata type 2	AR	*GNPAT*	602744
Alkylglycerone 3-phosphate synthase deficiency	Rhizomelic chondrodysplasia punctata type 3	AR	*AGPS*	603051
Fatty Acyl-CoA reductase 1 (FAR1) deficiency		AR	*FAR1*	616107
Peroxin 5 long isoform deficiency	Rhizomelic chondrodysplasia punctata type 5	AR	*PEX5*	600414
110. Disorders of peroxisomal β-oxidation				
X-linked adrenoleukodystrophy		XLR	*ABCD1*	300371
Peroxisomal straight-chain acyl-CoA oxidase deficiency	Pseudo-neonatal adrenoleukodystrophy	AR	*ACOX1*	609751
D-bifunctional protein deficiency	Pseudo-Zellweger syndrome (severe); Perrault syndrome type 1 (milder)	AR	*HSD17B4*	601860
L-bifunctional protein deficiency	Fanconi renotubular syndrome type 3	AR	*EHHADH*	607037
Sterol carrier protein-2 deficiency	Leukoencephalopathy with dystonia and motor neuropathy	AR	*SCP2*	184755
See also: α-methylacyl-CoA racemase deficiency in group 97.				
111. Disorder of peroxisomal α-oxidation				
Phytanoyl-CoA hydroxylase deficiency	Classic Refsum disease	AR	*PHYH*	602026
See also: PTS2 receptor deficiency in group 109.				
112. Disorders of peroxisomal biogenesis				
Peroxin 1 deficiency	Peroxisome biogenesis disorder 1A (Zellweger syndrome); peroxisome biogenesis disorder 1B (neonatal adrenoleukodystrophy/infantile Refsum disease); Heimler syndrome type 1	AR	*PEX1*	602136
Peroxin 2 deficiency	Peroxisome biogenesis disorder 5A (severe); peroxisome biogenesis disorder 5B (milder)	AR	*PEX2*	170993
Peroxin 3 deficiency	Peroxisome biogenesis disorder 10A (severe); peroxisome biogenesis disorder 10B (milder)	AR	*PEX3*	603164
Peroxin 5 deficiency	Peroxisome biogenesis disorder 2A (severe); peroxisome biogenesis disorder 2B (milder)	AR	*PEX5*	600414
Peroxin 6 deficiency	Peroxisome biogenesis disorder 5A (severe); peroxisome biogenesis disorder 5B (intermediate); Heimler syndrome type 2 (milder)	AD, AR	*PEX6*	601498

Peroxin 10 deficiency	Peroxisome biogenesis disorder 6A (severe); peroxisome biogenesis disorder 6B (milder)	AR	*PEX10*	602859
Peroxin 11B deficiency	Peroxisome biogenesis disorder 14B	AR	*PEX11B*	603867
Peroxin 12 deficiency	Peroxisome biogenesis disorder 3A (severe); peroxisome biogenesis disorder 3B (milder)	AR	*PEX12*	601758
Peroxin 13 deficiency	Peroxisome biogenesis disorder 11A (severe); peroxisome biogenesis disorder 11B (milder)	AR	*PEX13*	601789
Peroxin 14 deficiency	Peroxisome biogenesis disorder 13A	AR	*PEX14*	601791
Peroxin 16 deficiency	Peroxisome biogenesis disorder 8A (severe); peroxisome biogenesis disorder 8B (milder)	AR	*PEX16*	603360
Peroxin 19 deficiency	Peroxisome biogenesis disorder 12A	AR	*PEX19*	600279
Peroxin 26 deficiency	Peroxisome biogenesis disorder 7A (severe); peroxisome biogenesis disorder 7B (milder)	AR	*PEX26*	608666
113. Peroxisomal disorders not involving lipid metabolism				
Catalase deficiency	Acatalasemia	AR	*CAT*	115500
Alanine-glyoxylate aminotransferase deficiency	Primary hyperoxaluria type 1	AR	*AGXT*	604285
Hydroxyacid oxidase 1 deficiency	Glycolate oxidase deficiency; isolated glycolic aciduria	AR	*HAO1*	605023
114. Disorders of oxalate metabolism				
Glyoxylate reductase/hydroxypyruvate reductase deficiency	Primary hyperoxaluria type 2	AR	*GRHPR*	604296
4-hydroxy-2-oxoglutarate aldolase 1 deficiency	Primary hyperoxaluria type 3	AR	*HOGA1*	613597
Oxalate transporter deficiency		AR	*SLC26A1*	610130
See also: Alanine-glyoxylate aminotransferase and hydroxyacid oxidase 1 deficiencies in group 113.				
I. CONGENITAL DISORDERS OF GLYCOSYLATION				
115. Disorders of N-linked glycosylation				
Phosphomannomutase 2 deficiency	PMM2-CDG	AR	*PMM2*	601785
Phosphomannose isomerase deficiency	MPI-CDG	AR	*MPI*	154550
N-acetylglucosamine-1-phosphate transferase deficiency	DPAGT1-CDG	AR	*DPAGT1*	191350
X-linked recessive UDP-N-acetylglucosamine transferase catalytic subunit deficiency	ALG13-CDG	XLR	*ALG13*	300776

X-linked dominant UDP-N-acetylglucosamine transferase catalytic subunit deficiency	Early infantile epileptic encephalopathy type 36	XLD	*ALG13*	300776
UDP-N-acetylglucosamine transferase anchoring subunit deficiency	ALG14-CDG	AR	*ALG14*	612866
ALG1 β-1,4-mannosyltransferase deficiency	ALG1-CDG	AR	*ALG1*	605907
ALG2 α-1,3/1,6-mannosyltransferase deficiency	ALG2-CDG	AR	*ALG2*	607905
ALG11 α-1,2-mannosyltransferase deficiency	ALG11-CDG	AR	*ALG11*	613666
Lipid-linked oligosaccharide flippase deficiency	RFT1-CDG	AR	*RFT1*	611908
ALG3 α-1,3-mannosyltransferase deficiency	ALG3-CDG	AR	*ALG3*	608750
ALG9 α-1,2-mannosyltransferase deficiency	ALG9-CDG	AR	*ALG9*	606941
ALG12 α-1, 6-mannosyltransferase deficiency	ALG12-CDG	AR	*ALG12*	607144
ALG6 α-1,3-glucosyltransferase deficiency	ALG6-CDG	AR	*ALG6*	604566
ALG8 α-1,3-glucosyltransferase deficiency	ALG8-CDG (recessive); polycystic liver disease type 3 (dominant)	AD, AR	*ALG8*	608103
Oligosaccharyltransferase TUSC3 subunit deficiency	TUSC3-CDG	AR	*TUSC3*	601385
Oligosaccharyltransferase STT3A subunit deficiency	STT3A-CDG	AR	*STT3A*	601134
Oligosaccharyltransferase MAGT1 subunit deficiency	MAGT1-CDG	XLR	*MAGT1*	300715
Translocon-associated protein δ subunit deficiency	SSR4-CDG	XLR	*SSR4*	300090
Mannosyl-oligosaccharide α-1,2-glucosidase deficiency	MOGS-CDG	AR	*MOGS*	601336
α-1,3-glucosidase II subunit α deficiency	Polycystic kidney disease type 3	AD	*GANAB*	104160
α-1,3-glucosidase II subunit β deficiency	Polycystic liver disease type 1	AD	*PRKCSH*	177060
α-1,2-mannosidase I deficiency	MAN1B1-CDG	AR	*MAN1B1*	604346

β-1,2-N-acetylglucosaminyltransferase II deficiency	MGAT2-CDG	AR	*MGAT2*	602616
α-1,6-fucosyltransferase deficiency	FUT8-CDG	AR	*FUT8*	602589
116. Disorders of O-mannosylation				
Protein O-mannosyltransferase 1 deficiency	MDDGA1 (severe); MDDGB1 (intermediate); MDDGC1 (milder)	AR	*POMT1*	607423
Protein O-mannosyltransferase 2 deficiency	MDDGA2; MDDGB2; MDDGC2	AR	*POMT2*	607439
Protein O-mannose β-1,2-N-acetylglucosaminyltransferase deficiency	MDDGA3; MDDGB3; MDDGC3; retinitis pigmentosa type 76	AR	*POMGNT1*	606822
Protein O-mannose β-1,4-N-acetylglucosaminyltransferase deficiency	MDDGA8	AR	*POMGNT2*	614828
β-1,3-galactosaminyltransferase 2 deficiency	MDDGA11	AR	*B3GALNT2*	610194
Protein O-mannose kinase deficiency	MDDGA12; MDDGC12	AR	*POMK*	615247
Methylerythritol 4-phosphate cytidylyltransferase deficiency	MDDGA7; MDDGC7	AR	*ISPD*	614631
Fukutin deficiency	MDDGA4; MDDGB4; MDDGC4	AR	*FKTN*	607440
Fukutin-related protein deficiency	MDDGA5; MDDGB5; MDDGC5	AR	*FKRP*	606596
Ribitol β-1,4-xylosyltransferase deficiency	MDDGA10	AR	*RXYLT1*	605862
β-1,4-glucuronyltransferase 1 deficiency	MDDGA13	AR	*B4GAT1*	605517
β-1,3-glucuronyltransferase/α-1,3-xylosytransferase deficiency	MDDGA6; MDDGB6	AR	*LARGE1*	603590
117. Disorders of O-xylosylation and glycosaminoglycan synthesis				
Xylosyltransferase 1 deficiency	Desbuquois dysplasia type 2	AR	*XYLT1*	608124
Xylosyltransferase 2 deficiency	Spondyloocular syndrome	AR	*XYLT2*	608125
β-1,4-galactosyltransferase 7 deficiency	Progeroid Ehlers-Danlos syndrome type 1; Larsen of Reunion Island syndrome	AR	*B4GALT7*	604327
β-1,3-galactosyltransferase 6 deficiency	Spondyloepimetaphyseal dysplasia with joint laxity type 1; progeroid Ehlers-Danlos syndrome type 2	AR	*B3GALT6*	615291
β-1,3-glucuronyltransferase 3 deficiency	Larsen-like syndrome	AR	*B3GAT3*	606374
Exostosin glycosyltransferase 1 deficiency	Multiple hereditary exostoses type 1	AD	*EXT1*	608177
Exostosin glycosyltransferase 2 deficiency	Multiple hereditary exostoses type 2	AD	*EXT2*	608210
Exostosin-like glycosyltransferase 3 deficiency	Immunoskeletal dysplasia with neurodevelopmental abnormalities	AR	*EXTL3*	605744

Chondroitin sulfate synthase 1 deficiency	Temtamy preaxial brachydactyly syndrome	AR	*CHSY1*	608183
Chondroitin 6-sulfotransferase deficiency	Autosomal recessive Larsen syndrome; spondyloepiphyseal dysplasia Omani type; humerospinal dysostosis	AR	*CHST3*	603799
Dermatan 4-sulfotransferase 1 deficiency	Ehlers-Danlos syndrome musculocontractural type 1	AR	*CHST14*	608429
Dermatan sulfate epimerase deficiency	Ehlers-Danlos syndrome musculocontractural type 2	AR	*DSE*	605942
Corneal N-acetylglucosamine 6-O-sulfotransferase deficiency	Macular corneal dystrophy	AR	*CHST6*	605294
UDP-galactose nucleotidase deficiency	Desbuquois dysplasia type 1; multiple epiphyseal dysplasia type 7	AR	*CANT1*	613165
Sulfate transporter deficiency	Achondrogenesis type 1; atelosteogenesis type 2; diastrophic dysplasia; multiple epiphyseal dysplasia type 4	AR	*SLC26A2*	606718
Phosphoadenosine 5'-phosphosulfate synthetase 2 deficiency	Spondyloepimetaphyseal dysplasia, Pakistani type	AR	*PAPSS2*	603005
Golgi-resident phosphoadenosine phosphate phosphatase deficiency	Chondrodysplasia with joint dislocations, gPAPP type	AR	*IMPAD1*	614010
See also: UDP-glucuronic acid/UDP-N-acetylgalactosamine dual transporter deficiency in group 127.				
118. Disorders of O-GalNAcylation				
Polypeptide N-acetylgalactosaminyltransferase 3 deficiency	Hyperphosphatemic familial tumoral calcinosis type 1; hyperostosis-hyperphosphatemia syndrome	AR	*GALNT3*	601756
Core 1 β-1,3-galactosyltransferase chaperone deficiency	Tn polyagglutination syndrome	Somatic	*C1GALT1C1*	300611
119. Disorders of O-GlcNAcylation				
O-linked N-acetylglucosamine transferase deficiency	X-linked mental retardation type 106	XLR	*OGT*	300255
EGF domain-specific O-linked N-acetylglucosamine transferase deficiency	Adams-Oliver syndrome type 4	AR	*EOGT*	614789
120. Disorder of O-glucosylation				
Protein O-glucosyltransferase deficiency	Dowling-Degos disease type 4	AD	*POGLUT1*	615618
121. Disorders of O-fucosylation				
Protein O-fucosyltransferase deficiency	Dowling-Degos disease type 2	AD	*POFUT1*	607491

Fucose-specific β-1,3-N-acetylglucosaminyltransferase deficiency	Spondylocostal dysostosis type 3	AR	*LFNG*	602576
Fucose-specific β-1,3-glucosyltransferase deficiency	Peters-Plus syndrome	AR	*B3GLCT*	610308
122. Disorders of glycosylphosphatidylinositol biosynthesis				
PIGA-CDG	Multiple congenital anomalies-hypotonia-seizures syndrome type 2; GPI biosynthesis defect type 4; early infantile epileptic encephalopathy type 20	XLR	*PIGA*	311770
PIGC-CDG	GPI biosynthesis defect type 16; autosomal recessive mental retardation type 62	AR	*PIGC*	601730
PIGQ-CDG		AR	*PIGQ*	605754
PIGL-CDG	CHIME syndrome; GPI biosynthesis defect type 5	AR	*PIGL*	605947
PIGW-CDG	Hyperphosphatasia with mental retardation type 5; GPI biosynthesis defect type 11	AR	*PIGW*	610275
PIGM-CDG	GPI biosynthesis defect type 1	AR	*PIGM*	610273
PIGV-CDG	Hyperphosphatasia with mental retardation type 1; GPI biosynthesis defect type 2	AR	*PIGV*	610274
PIGN-CDG	Multiple congenital anomalies-hypotonia-seizures syndrome type 1; GPI biosynthesis defect type 3	AR	*PIGN*	606097
PIGO-CDG	Hyperphosphatasia with mental retardation type 2; GPI biosynthesis defect type 6	AR	*PIGO*	614730
PIGG-CDG	Autosomal recessive mental retardation type 53; GPI biosynthesis defect type 13	AR	*PIGG*	616918
PIGT-CDG	Multiple congenital anomalies-hypotonia-seizures syndrome type 3, GPI biosynthesis defect type 7	AR	*PIGT*	610272
GPAA1-CDG	GPI biosynthesis defect type 15	AR	*GPAA1*	603048
PGAP1-CDG	Autosomal recessive mental retardation type 42; GPI biosynthesis defect type 9	AR	*PGAP1*	611655
PGAP3-CDG	Hyperphosphatasia with mental retardation type 4; GPI biosynthesis defect type 10	AR	*PGAP3*	611801
PGAP2-CDG	Hyperphosphatasia with mental retardation type 3; GPI biosynthesis defect type 8	AR	*PGAP2*	615187
123. Disorders of glycolipid glycosylation				
GM3 synthase deficiency	Amish infantile epilepsy syndrome**;** salt and pepper developmental regression syndrome	AR	*ST3GAL5*	604402

GM2/GD2 synthase deficiency	Autosomal recessive spastic paraplegia type 26	AR	*B4GALNT1*	601873
GD1a/GT1b synthase deficiency	ST3GAL3-CDG	AR	*ST3GAL3*	606494
GB3 synthase deficiency	NOR polyagglutination syndrome	AD	*A4GALT*	607922
124. Disorders of dolichol metabolism				
Cis-isoprenyl transferase deficiency		AD, AR	*DHDDS*	608172
Nogo-B receptor deficiency		AD, AR	*NUS1*	610463
Polyprenol reductase deficiency	SRD5A3-CDG	AR	*SRD5A3*	611715
Dolichol kinase deficiency	DOLK-CDG	AR	*DOLK*	610746
Dolichol-phosphate mannose synthase subunit 1 deficiency	DPM1-CDG	AR	*DPM1*	603503
Dolichol-phosphate mannose synthase subunit 2 deficiency	DPM2-CDG	AR	*DPM2*	603564
Dolichol-phosphate mannose synthase subunit 3 deficiency	DPM3-CDG	AR	*DPM3*	605951
MPDU1-CDG		AR	*MPDU1*	604041
125. Disorders of monosaccharide synthesis and interconversion				
UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase deficiency	GNE myopathy; Nonaka myopathy	AR	*GNE*	603824
UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase superactivity	Sialuria	AD	*GNE*	603824
N-acetylneuraminic acid-9-phosphate synthase deficiency	NANS-CDG; spondyloepimetaphyseal dysplasia Camera-Genevieve type	AR	*NANS*	605202
Glutamine:fructose-6-phosphate transaminase deficiency	Congenital myasthenic syndrome type 12	AR	*GFPT1*	138292
Phosphoglucomutase 1 deficiency	PGM1-CDG; glycogen storage disease type 14	AR	*PGM1*	171900
N-Acetylglucosamine-phosphate mutase deficiency	PGM3-CDG; immunodeficiency type 23	AR	*PGM3*	172100
Glucose-6-phosphatase catalytic subunit 3 deficiency	Severe congenital neutropenia type 4; Dursun syndrome	AR	*G6PC3*	611045
See also: Phosphomannomutase 2 and phosphomannose isomerase deficiencies in group 115.				
126. Disorders of nucleotide-sugar synthesis				
GDP-mannose pyrophosphorylase α subunit deficiency	Alacrima, achalasia, and mental retardation syndrome	AR	*GMPPA*	615495

GDP-mannose pyrophosphorylase β subunit deficiency	MDDGA14; MDDGB14; MDDGC14	AR	*GMPPB*	615320
See also: CAD trifunctional protein deficiency in group 1.				
127. Disorders of Golgi transport				
CMP-sialic acid transporter deficiency	SLC35A1-CDG	AR	*SLC35A1*	605634
UDP-galactose transporter deficiency	SLC35A2-CDG; early infantile epileptic encephalopathy 22	XLD	*SLC35A2*	314375
UDP-N-acetylglucosamine transporter deficiency	SLC35A3-CDG	AR	*SLC35A3*	605632
GDP-fucose transporter deficiency	SLC35C1-CDG	AR	*SLC35C1*	605881
UDP-glucuronic acid/UDP-N-acetylgalactosamine dual transporter deficiency	Schneckenbecken dysplasia; SLC35D1-CDG	AR	*SLC35D1*	610804
See also: SLC39A8 deficiency in group 42.				
128. Glycosylation disorders of vesicular trafficking				
Conserved oligomeric Golgi complex subunit 1 deficiency	COG1-CDG	AR	*COG1*	606973
Conserved oligomeric Golgi complex subunit 4 deficiency	COG4-CDG	AR	*COG4*	606976
Conserved oligomeric Golgi complex subunit 5 deficiency	COG5-CDG	AR	*COG5*	606821
Conserved oligomeric Golgi complex subunit 6 deficiency	COG6-CDG	AR	*COG6*	606977
Conserved oligomeric Golgi complex subunit 7 deficiency	COG7-CDG	AR	*COG7*	606978
Conserved oligomeric Golgi complex subunit 8 deficiency	COG8-CDG	AR	*COG8*	606979
Jagunal 1 deficiency	Severe congenital neutropenia type 6	AR	*JAGN1*	616012
Congenital dyserythropoietic anemia type 2	SEC23B-CDG	AR	*SEC23B*	610512
Achondrogenesis type 1A		AR	*TRIP11*	604505
TRAPPC11-CDG	Limb-girdle muscular dystrophy type 2S	AR	*TRAPPC11*	614138
Cohen syndrome		AR	*VPS13B*	607817

129. Disorders of Golgi homeostasis				
ATP6V0A2-CDG	Autosomal recessive cutis laxa type 2A (wrinkly skin syndrome)	AR	*ATP6V0A2*	611716
ATP6V1A-CDG	Autosomal recessive cutis laxa type 2D	AR	*ATP6V1A*	607027
ATP6V1E1-CDG	Autosomal recessive cutis laxa type 2C	AR	*ATP6V1E1*	108746
ATP6AP1-CDG	Immunodeficiency type 47	XLR	*ATP6AP1*	300197
ATP6AP2-CDG	X-linked mental retardation, Hedera type	XLR	*ATP6AP2*	300556
Transmembrane protein 199 deficiency	TMEM199-CDG	AR	*TMEM199*	616815
CCDC115-CDG		AR	*CCDC115*	613734
Transmembrane protein 165 deficiency	TMEM165-CDG	AR	*TMEM165*	614726
130. Disorder of deglycosylation				
N-glycanase 1 deficiency	NGLY1-CDDG	AR	*NGLY1*	610661

Abbreviations: AD, autosomal dominant; adPEO, autosomal dominant progressive external ophthalmoplegia; AR, autosomal recessive; arPEO, autosomal recessive progressive external ophthalmoplegia; GPI, glycosylphosphatidylinositol; Mit, mitochondrial; MDDGA, muscular dystrophy-dystroglycanopathy with brain and eye anomalies (type A, includes Walker-Warburg syndrome and muscle-eye-brain disease); MDDGB, muscular dystrophy-dystroglycanopathy with mental retardation (type B); MDDGC, limb-girdle muscular dystrophy-dystroglycanopathy (type C); XL, X-linked; XLD, X-linked dominant; XLR, X-linked recessive.