List of IMDs presenting with myopathies (n=358)

Name	Gene	Weakness	Hypotonia	Exercise intolerance	Rhabdomyolysis	Abnormal pathology	Other	Laboratory investigations	Specific treatment	OMIM dierase no.	IEMbase ID (hyperlinke
DISORDERS OF NITROGEN-CONTAINING COMPOUNDS (n=42)											
Disorders of pyrimidine metabolism											
Pyrimidine-5'-nucleotidase I deficiency	*NT5C3A*				X			Uric acid (U)	Uridine, possibly ribose	266120;606224;191720	IEM0004
Disorders of purine metabolism											
Myoadenylate deaminase deficiency	*AMPD1*			X			Muscle cramps	CK (P)		102770	IEM0010
Xanthine oxidase deficiency	*XDH*	X						Purines and pyrimidines (U, P), Uric acid (U)		278300;607633	IEM0016
Adenylosuccinate synthase-like 1 deficiency	*ADSS1*	X						CK (P)		617030	IEM1276
Disorders of creatine metabolism											
Arginine:glycine amidinotransferase deficiency	*GATM*	X						Creatinine (P, U), Guanidino compounds (P, U)	Creatine supplementation	612718	IEM0042
Arginine:glycine amidinotransferase aggregation syndrome	*GATM*	X						Creatinine (P, U), Guanidino compounds (P, U)	Creatine supplementation	612718	IEM0043
Creatine transporter deficiency	*SLC6A8*						Muscle mass, low	Creatinine (P, U), Guanidino compounds (P, U)	Creatine, arginine and glycine supplementation	300352	IEM0045
Disorders of glutathione metabolism											
Gamma-glutamylcysteine synthetase deficiency	*GCLC*	X						Hemoglobin (B), Reticulocytes (B), Glutathione (RBC)	Avoid drugs that precipitate hemolytic crisis in G6PD deficiency (phenobarbital, acetylsalyciclic acid, sulfonamides)	230450	IEM0049
Glutathione synthetase deficiency, sever	*GSS*	X						5-Oxoproline (U), Hemoglobin (B), Reticulocytes (B), Glutathione (RBC), Lactate (P)	Na bicarbonate to treat metabolic acidosis, antioxidants (vitamin C, E), avoid drugs like acetylsalycylic acid, phenopbarbital, sulfonamides)	266130	IEM0050
Methylthioadenosine phosphorylase deficiency	*MTAP*	X						DNA		156540	IEM1245
Disorders of amino acid transport											
Lysinuric protein intolerance	*SLC7A7*	X						Amino acids (P, U), Ammonia (P), Ferritin (S), Lipid panel (S), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline	222700	IEM0070
Aminoacylase deficiencies											
Aspartoacylase deficiency (Canavan disease)	*ASPA*	X	X					N-Acetylaspartic acid (U, P, CSF)		271900	IEM0074
Aminoacylase 1 deficiency	*ACY1*	X						Organic acids (U)		609924	IEM0075
Disorders of monoamine metabolism											
Tyrosine hydroxylase deficiency	*TH*		X					Prolactin (P), Biogenic amines (CSF)	L-dopa/Carbidopa	191290	IEM0076
Dopamine beta-hydroxylase deficiency	*DBH*		X					Catecholamines (U, P), Biogenic amines (CSF)	Droxidopa	223360	IEM0078
Dopamine transporter deficiency	*SLC6A3*		X					Biogenic amines (CSF, U)	Dopamine agonists	613135;126455	IEM0080
Disorders of phenylalanine and tetrahydrobiopterin metabolism											
Phenylalanine hydroxylase deficiency	*PAH*	X	X					Amino acids (P)	Phe-restricted diet, large neutral amino acids, glycomacropeptide, sapropterin, pegvaliase	261600	IEM0082
Autosomal recessive GTP cyclohydrolase 1 deficiency	*GCH1*	X	X					Amino acids (P), Pterins (DBS,U), Biogenic amines (CSF)	L-dopa/Carbidopa	233910	IEM0083
Autosomal dominant GTP cyclohydrolase 1 deficiency (Segawa disease)	*GCH1*		X					Amino acids (P), Pterins (DBS,U), Biogenic amines (CSF)	L-dopa/Carbidopa	600225	IEM0084
6-Pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	X	X					Amino acids (P), Pterins (DBS, U), Biogenic amines (CSF)	Tetrahydrobiopterin, L-dopa/dopa carboxylase inhibitor; 5-hydroxytrypptophan; +/- folinic acid	261640	IEM0085
Sepiapterin reductase deficiency	*SPR*	X						Pterins (DBS, U), Biogenic amines (CSF)	Tetrahydrobiopterin, L-dopa/dopa carboxylase inhibitor; 5-hydroxytrypptophan; MAO inhibitor, serotonin reuptake inhibitor,	182125	IEM0086
Dihydropteridine reductase deficiency	*QDPR*	X	X					Amino acids (P), Pterins (DBS,U), DHPR activity (DBS), Biogenic amines (CSF), 5-methyl-THF (CSF)	Low Phe, L-dopa, 5-hydroxytryptophan, folinic acid	261630	IEM0087
4-hydroxyphenylpyruvate dioxygenase-like protein deficiency	*HPDL*		X					DNA		619026	IEM1667
Disorders of sulfur amino acid and sulfide metabolism											
S-adenosylhomocysteine hydrolase deficiency	*AHCY*	X						Amino acids (P), Homocysteine (P), S-Adenosylcysteine (P)	Methionine or protein restricted diet, phosphatidylcholine and creatine supplements, liver transplant	613752	IEM0100
Adenosine kinase deficiency	*ADK*	X						SAM/SAH (P), ASAT/ALAT (P), Glucose (S), Amino acids (P), Purines (U), Total/direct bilirubin (S)		614300	IEM0101
Disorders of branched-chain amino acid metabolism											

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Branched-chain ketoacid dehydrogenase E1 alpha deficiency (MSUD)	*BCKDHA*	X	X					Amino acids (P), Organic acids (U)		248600	IEM0108
Branched-chain ketoacid dehydrogenase E1 beta deficiency (MSUD)	*BCKDHB*	X	X					Amino acids (P), Organic acids (U)		248600	IEM0109
Dihydrolipoyl transacylase deficiency	*DBT*	X	X					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P),	Dietary leucine restriction, BCAA-free medical foods, judicious supplementation with isoleucine and valine, hemodialysis/hemofiltration,	248600	IEM0110
Dihydrolipoyl dehydrogenase deficiency	*DLD*	X			X			Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P),	Protein/BCAA restriction; trial of ketogenic/high-fat diet; DCA supplementation (50-75 mg/kg/day); thiamine, CoQ10, Riboflavin, Lipoic	248600	IEM0111
Isovaleryl-CoA dehydrogenase deficiency	*IVD*	X	X					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P),	Protein restricted diet; Carnitine 50-100 mg/kg/day; Glycine 150-259 mg/kg/day	243500	IEM0113
2-Methylbutyryl-CoA dehydrogenase deficiency	*ACADSB*	X	X					Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Glucose (S)		610006	IEM0115
3-Methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	X					Muscle pain	Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P),	In symptomatic patients +/- protein restriction; +/- carnitine	210200	IEM0116
3-Methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	X					Muscle pain	Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P),	In symptomatic patients +/- protein restriction; +/- biotin	210200	IEM0117
3-hydroxyisobutyryl-CoA hydrolase deficiency	*HIBCH*	X	X					Organic acids (U), S-(2-carboxypropyl)-cysteine (U), S-2-carboxypropyl-cysteamine (U)	Moderate protein restricted diet	250620	IEM0120
Disorders of lysine metabolism											
DHTKD1 deficiency	*DHTKD1*	X					Muscular atrophy	Amino acids (P), Organic acids (U)		204750	IEM0132
Disorders of proline and ornithine metabolism											
Pyrroline-5-carboxylate reductase 2 deficiency	*PYCR2*	X						DNA		616420	IEM0140
Ornithine aminotransferase deficiency	*OAT*	X				X		Guanidino compounds (U,P,CSF), Amino acids (P)	Low protein diet	258870	IEM0146
Pyrroline-5-carboxylate synthetase deficiency, spastic paraplegia type 9A	*ALDH18A1*	X					Muscle wasting	Amino acids (P), Ammonia (P)	Arginine 150 mg/kg/day	138250;219150	IEM1485
Disorders of glutamate metabolism											
Ionotropic glutamate receptor NMDA type subunit 1 dysregulation	*GRIN1*	X						DNA		614254;617820	IEM0169
Thorase deficiency	*ATAD1*		X					GABA free (CSF)		618011	IEM0175
Disorder of glutamine metabolism											
Glutaminase deficiency	*GLS*	X						Amino acids (U, CSF)		618328	IEM1083
Disorder of asparagine metabolism											
Asparagine synthetase deficiency	*ASNS*	X						Amino acids (P)		615574	IEM0180
DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS (n=30)											
Disorders of lipoic acid and iron-sulfur metabolism											
NFU1 deficiency	*NFU1*	X						Amino acids (P), Organic acids (U), Lactate (P, U, CSF), Protein bound lipoic acid (FB)		605711	IEM0195
IBA57 deficiency	*IBA57*	X						Lactate (P), Amino acids (P)		615330	IEM0198
ISCU deficiency	*ISCU*	X		X	X			Lactate (P), Myoglobin (U), Complexes I - III activity (muscle)		255125	IEM0201
Ferredoxin reductase deficiency	*FDXR*	X						Complexes I - IV activity (muscle), DNA		617717	IEM0203
Ferredoxin 2 deficiency	*FDX2*	X		X	X		Muscle cramps	Myoglobin (U), Lactate (P), Organic acids (U)		614585	IEM1121
Disorders of folate metabolism											
5,10-methylenetetrahydrofolate reductase deficiency	*MTHFR*	X						Amino acids (P), Homocysteine (P), 5-Methyltetrahydrofolate (CSF), Folate (S)	Betaine (9000 mg/day), folinic acid (45 mg/day), vitamin B12 (1000 mg/week), vitamin B6 (300 mg/day) and profilactic acetylsalicylic	236250	IEM0223
Disorders of riboflavin metabolism											
Riboflavin transporter 2 deficiency	*SLC52A3*	X						Flavins (B), Organic acids (U), Acylcarnitines (P,DBS)		211530	IEM0234
Riboflavin transporter 3 deficiency	*SLC52A2*	X						Flavins (B), Organic acids (U), Acylcarnitines (P,DBS)		614707	IEM0235
Flavin adenine dinucleotide synthetase deficiency	*FLAD1*	X						Flavins (B), Acyglycines (U), Organic acids (U), Acylcarnitines (P,DBS)		255100	IEM0236
Mitochondrial flavin adenine dinucleotide transporter deficiency	*SLC25A32*	X		X				Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)		616839	IEM0237

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Electron transfer flavoprotein α subunit deficiency (GA2A)	*ETFA*	X	X		X		Skeletal myopathy	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)		231680	IEM0238
Electron transfer flavoprotein β subunit deficiency (GA2B)	*ETFB*	X	X		X		Skeletal myopathy	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)		231680	IEM0239
Multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)	*ETFDH*	X	X		X		Skeletal myopathy	Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)		231675	IEM0240
Disorders of pantothenate metabolism											
Pantothenate kinase 2 deficiency (Hallervorden-Spatz disease)	*PANK2*		X					Iron (brain), DNA	Possible iron chelation, possible panthotenate	234200	IEM0246
Mitochondrial coenzyme A transporter deficiency	*SLC25A42*	X						Lactate P)		610823	IEM0249
Disorders of pyridoxine metabolism											
Pyridoxal kinase deficiency	*PDXK*	X					Muscle atrophy	DNA		618511	IEM1560
Disorders of vitamin A metabolism											
Vitamin A receptor deficiency (Matthew-Wood syndrome)	*STRA6*	X						DNA		601186	IEM0256
Disorders of vitamin D metabolism											
Vitamin D 1-α-hydroxylase deficiency	*CYP27B1*	X						Amino acids (U), ALP (P), Calcium (P), Phosphate (P), PTH (S), 1,25-Dihydroxy vitamin D (S)		264700	IEM0266
Vitamin D 25-hydroxylase deficiency	*CYP2R1*	X						Amino acids (U), ALP (P), Calcium (P), Phosphate (P), PTH (S), 1,25-Dihydroxy vitamin D (S)		600081	IEM0267
Vitamin D receptor deficiency	*VDR*	X						ALP (P), Calcium (P), Phosphate (P)	Calcitriol, calcium, cinacalcet	277440	IEM0268
Disorders of molybdenum metabolism											
Cyclic pyranopterin monophosphate synthase deficiency (MoCo deficiency A)	*MOCS1*	X	X					Purines (U), Uric acid (U, P), Homocysteine, total (P), Sulfite (U), Pipecolic acid (CSF)	cPMP (fosdenopterin)	603707	IEM0275
Molybdopterin synthase deficiency (MoCo deficiency B)	*MOCS2*	X	X					Purines (U), Uric acid (U, P), Homocysteine, total (P), Sulfite (U), Pipecolic acid (CSF)		603708	IEM0276
Gephyrin deficiency (MoCo deficiency C)	*GPHN*	X	X					Purines (U), Uric acid (U, P), Homocysteine, total (P), Sulfite (U), Pipecolic acid (CSF)		603930	IEM0277
Molybdenum cofactor sulfurase deficiency	*MOCOS*	X						Purines and pyrimidines (U, P), Uric acid (U)		603592	IEM0278
Disorders of copper metabolism											
Copper-transporting ATPase subunit alpha deficiency	*ATP7A*	X						Copper (S, U), Ceruloplasmin (S)	Copper chloride or L-histidine 350-500 ug/day IV or SC	300489	IEM0281
Acetyl-CoA transporter deficiency (Huppke-Brendel syndrome)	*SLC33A1*	X	X					Copper (S, U), Ceruloplasmin (S)		614482	IEM0283
Disorders of zinc metabolism											
Spondylocheirodysplastic Ehlers-Danlos syndrome	*SLC39A13*						Muscle atrophy	Lysyl pyridinoline (U), Hydroxylysyl pyridinoline (U)		612350	IEM0299
Disorders of magnesium metabolism											
Epithelial magnesium transporter deficiency	*TRPM6*						Muscle cramps	Calcium (P), Magnesium (S)		602014	IEM0306
Sodium-potassium ATPase γ subunit deficiency	*FXYD2*	X					Muscle cramps	Calcium (P, U, Magnesium (S, U)		154020	IEM0307
Sodium-chloride cotransporter deficiency (Gitelman syndrome)	*SLC12A3*	X					Muscle cramps	Calcium (P), Magnesium (S), Potassium (P)		263800	IEM0312
DISORDERS OF CARBOHYDRATES (n=23)											
Disorders of carbohydrate transport and absorption											
Glucose transporter 1 deficiency	*SLC2A1*	X	X					Glucose (P, CSF)	Ketogenic diet, clinical trials with triheptanoin	606777;612126;601042;614847	IEM0314
Glycogen storage diseases											
Muscle glycogenin 1 deficiency	*GYG1*	X						Glycogen (M)		613507	IEM0352
Muscle glycogen synthase deficiency	*GYS1*	X						Glycogen (M)		611556	IEM0353
Amylo-1,6-glucosidase (debrancher) deficiency (Cori-Forbes)	*AGL*	X						ASAT/ALAT (P), CK (P), Lactate (P), Glucose (S), Glycogen (L), Biotinidase (P)	D,L-3-hydrobutyrate, ketogenic and high-protein diet	232400	IEM0357
Glycogen branching enzyme deficiency (Andersen)	*GBE1*	X	X				Muscular atrophy	ASAT/ALAT (P), Bilirubin (P), Coagulation factors (P), Glycogen (L)		232500	IEM0358

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Muscle glycogen phosphorylase deficiency (McArdle disease)	*PYGM*	X		X			Muscle cramps; Muscle pain	CK (P), Glycogen (M), Uric acid (P)		232600	IEM0359
Muscle phosphorylase kinase deficiency	*PHKA1*	X		X			Muscle cramps; Muscle pain	CK (P), Glycogen (M), Uric acid (P)		300559	IEM0364
HOIL1 deficiency	*RBCK1*	X					Myalgia	CK (P), DNA		615895	IEM0365
Cardiac phosphorylase kinase deficiency	*PRKAG2*	X						CK (P), Glucose (S)		600858;261740;194200	IEM0366
Lysosome-associated membrane protein 2 deficiency (Danon disease)	*LAMP2*	X	X				Muscle cramps	ASAT/ALAT (P), CK (P), Glycogen (M)		300257	IEM0367
Laforin deficiency	*EPM2A*	X						DNA		254780	IEM0368
Liver glycogenin 2 deficiency	*GYG2*		X					DNA		300198	IEM1524
Disorders of gluconeogenesis											
Pyruvate carboxylase deficiency	*PC*		X					Amino acids (P), Lactate (P), Glucose (P), 3-OH-Butyrate/Acetoacetate (P)		266150	IEM0372
Disorders of glycolysis											
Hereditary motor and sensory neuropathy, Russe type	*HK1*	X						Bilirubin (P), Blood count (B), DNA		605285	IEM0375
Glucose-6-phosphate isomerase deficiency	*GPI*	X						Bilirubin, unconjugated (P), Hemoglobin (B), Reticulocytes (B), Enzyme activity		615802	IEM0379
Muscle phosphofructokinase deficiency (Tarui disease)	*PFKM*	X		X			Muscle cramps; Muscle pain	CK (P), Glycogen (M), Uric acid (P)		232800	IEM0380
Aldolase A deficiency	*ALDOA*	X			X			Bilirubin (P), Reticulocytes (B), Aldolase A (Ec)		611881	IEM0381
Triosephosphate isomerase deficiency	*TPI1*	X						Dihydroxyacetone phosphate (Ec)		615512	IEM0382
Phosphoglycerate kinase deficiency	*PGK1*	X					Muscle cramps; Muscle pain	Reticulocytes (B)		300653	IEM0383
Muscle phosphoglycerate mutase deficiency (DiMauro disease)	*PGAM2*	X					Muscle cramps; Muscle pain	CK (P), Myoglobin (U)		261670	IEM0384
Enolase beta deficiency	*ENO3*	X					Muscle cramps; Muscle pain	CK (P), Glycogen (M)		612932	IEM0385
Lactate dehydrogenase A deficiency	*LDHA*	X					Muscle cramps; Muscle pain	CK (P), Myoglobin (U), Lactate (P)		612933	IEM0387
Lactate dehydrogenase B deficiency	*LDHB*				X			LDH (P), Myoglobin (U)		614128	IEM0388
MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM (n=107)											
Disorders of pyruvate metabolism											
Pyruvate dehydrogenase kinase isoenzyme 3 superactivity	*PDK3*	X						DNA		300905	IEM0395
Disorders of the Krebs cycle											
Mitochondrial malate dehydrogenase deficiency	*MDH2*	X						Lactate (P), Organic acids (U)		617339	IEM0403
Disorders of mitochondrial carriers											
Adenine nucleotide translocator deficiency	*SLC25A4*	X		X			Muscle atrophy	Lactate (P)		609283	IEM0406
Mitochondrial phosphate carrier deficiency	*SLC25A3*	X		X				Lactate (P)		610773	IEM0407
Mitochondrial aspartate-glutamate carrier isoform 1 deficiency (Aralar deficiency)	*SLC25A12*	X	X					Lactate (P)		612949	IEM0408
S-adenosylmethionine carrier deficiency	*SLC25A26*	X						Pyruvate (P)		616794	IEM0410
Mitochondrial citrate carrier deficiency	*SLC25A1*	X						Organic acids (U, P, CSF)		615182	IEM0411
Mitochondrial ATP-Mg-phosphate transporter deficiency (Fontaine syndrome)	*SLC25A24*	X						DNA		612289	IEM0412
Disorders of complex I subunits											
NADH dehydrogenase flavoprotein 2 deficiency	*NDUFV2*	X						Lactate (P)		618229	IEM0414
NADH dehydrogenase iron-sulfur protein 1 deficiency	*NDUFS1*	X						Lactate (P)		618229	IEM0415
NADH dehydrogenase iron-sulfur protein 2 deficiency	*NDUFS2*	X						Lactate (P)		618228	IEM0416

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NADH dehydrogenase iron-sulfur protein 3 deficiency	*NDUFS3*	X						Lactate (P)		256000;252010	IEM0417
NADH dehydrogenase iron-sulfur protein 7 deficiency	*NDUFS7*	X						Lactate (P)		618224	IEM0418
NADH dehydrogenase iron-sulfur protein 8 deficiency	*NDUFS8*	X						Lactate (P)		618222	IEM0419
NADH dehydrogenase beta subcomplex subunit 3 deficiency	*NDUFB3*	X						Lactate (P)		252010	IEM0427
NADH dehydrogenase core subunit 1 deficiency	*MT-ND1*	X		X				Lactate (P)		252010	IEM0430
NADH dehydrogenase core subunit 2 deficiency	*MT-ND2*			X				Lactate (P)		252010	IEM0431
NADH dehydrogenase core subunit 3 deficiency	*MT-ND3*	X						Lactate (P)		252010	IEM0432
NADH dehydrogenase core subunit 5 deficiency	*MT-ND5*	X						Lactate (P)		252010	IEM0435
NADH dehydrogenase alpha subcomplex subunit 6 deficiency	*NDUFA6*	X						Lactate (P)		252010	IEM0436
NADH dehydrogenase alpha subcomplex subunit 8 deficiency	*NDUFA8*		X					Lactate (P)		603359	IEM1528
NADH dehydrogenase subunit C2 deficiency	*NDUFC2*	X						Lactate (P)		619170	IEM1529
Complex I assembly factor 2 deficiency	*NDUFAF2*	X						Lactate (P)		618233	IEM0438
NUBPL deficiency	*NUBPL*	X						Lactate (P)		618242	IEM0444
Transmembrane protein 126B deficiency	*TMEM126B*	X		X				Lactate (P)		618250	IEM0446
VAC14 deficiency type 2 (Lenk-Ploski syndrome)	*VAC14*	X						DNA		617054	IEM1249
Disorders of complex II subunits											
Succinate dehydrogenase subunit A deficiency	*SDHA*	X						Lactate (P)		252011	IEM0447
Succinate dehydrogenase subunit D deficiency	*SDHD*	X		X		X		Lactate (P), Organic acids (U)		252011	IEM0452
Succinate dehydrogenase complex assembly factor 1 deficiency	*SDHAF1*	X						Lactate (P), Organic acids (U)		252011	IEM0454
Disorders of complex III subunits											
UQCRB deficiency	*UQCRB*	X		X				Lactate (P)		615158	IEM0456
UQCRC1 deficiency	*UQCRC1*		X					DNA		619279	IEM1654
Disorders of complex IV subunits											
Cytochrome c oxidase subunit 1 deficiency	*MT-CO1*	X			X			Lactate (P)		516030	IEM0462
Cytochrome c oxidase subunit 2 deficiency	*MT-CO2*	X						Lactate (P)		516040	IEM0463
Cytochrome c oxidase subunit 3 deficiency	*MT-CO3*	X						Lactate (P)		516050	IEM0464
Cytochrome c oxidase subunit 6B1 deficiency	*COX6B1*	X						Lactate (P)		220110	IEM0467
Disorders of complex IV assembly and ancillary proteins											
SCO1 deficiency	*SCO1*	X						Lactate (P)		220110	IEM0473
SCO2 deficiency	*SCO2*	X					Muscular atrophy	Lactate (P)		604377;608908	IEM0474
APOPT1 deficiency	*APOPT1*	X						DNA		220110	IEM0480
CEP89 deficiency	*CEP89*	X						Lactate (P), Amino acids (P)		615470	IEM1150
Cytochrome c oxidase assembly factor 3 deficiency	*COA3*			X				Lactate (P)		619058	IEM1146
Disorders of complex V subunits											
Mitochondrial ATP synthase F1 subunit δ deficiency	*ATP5F1D*	X		X	X			Ammonia (P), Lactate (P), 3-Methylglutaconic acid (U)		603150	IEM0482
Mitochondrial ATP synthase F0 subunit 8 deficiency	*MT-ATP8*	X		X				Lactate (P)		516070	IEM0485

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Disorders of mitochondrial cytochrome synthesis and incorporation											
Mitochondrial cytochrome b deficiency	*MT-CYB*	X		X			Muscle cramps	Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)		516020	IEM0487
Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication											
Mitochondrial DNA polymerase gamma catalytic subunit deficiency	*POLG*	X		X				Lactate (P), Organic acids (U)		607459	IEM0491
Mitochondrial DNA polymerase gamma accessory subunit deficiency	*POLG2*	X						Lactate (P)		610131	IEM0492
Mitochondrial deoxyguanosine kinase deficiency	*DGUOK*	X						a-fetoprotein (S), Lactate (P), Amino acids (P), Sialotrasferrins (S)		251880;601465	IEM0493
MPV17 deficiency	*MPV17*	X						Lactate (P)		256810	IEM0494
Mitochondrial thymidine kinase 2 deficiency	*TK2*	X					Muscular atrophy	CK (P), Lactate (P)		609560;188250	IEM0496
Thymidine phosphorylase deficiency	*TYMP*	X						Purines and pyrimidines (U), Lactate (P)		131222;603041	IEM0498
DNA2 helicase deficiency	*DNA2*	X						DNA		615156	IEM0499
Mitochondrial ribonuclease H1 deficiency	*RNASEH1*	X						Lactate (P)		615156	IEM0500
Mitochondrial genome maintenance exonuclease 1 deficiency	*MGME1*	X						DNA		615084	IEM0501
Topoisomerase 3α deficiency	*TOP3A*	X		X				DNA		618098	IEM1073
Mitochondrial UMP-CMP kinase 2 deficiency	*CMPK2*	X						DNA		611787	IEM1152
Disorders of mitochondrial transcription and RNA transcript processing											
Pseudouridine synthase 1 deficiency	*PUS1*	X		X				Lactate (P)		600462	IEM0511
tRNA methyltransferase 5 deficiency	*TRMT5*	X		X				Lactate (P)		616539	IEM0513
Mitochondrial methionyl-tRNA methyltransferase deficiency	*NSUN3*	X						Lactate (P)		617491	IEM1257
Mitochondrial RNA polymerase deficiency	*POLRMT*	X						Lactate (P), 5-Methyl-THF (CSF)		601778	IEM1534
Mitochondrial ribosomopathies											
Mitochondrial ribosomal large subunit 12 deficiency	*MRPL12*	X					Hypotrophy	Lactate (P)		602375	IEM1155
Mitochondrial oxodicarboxylate carrier deficiency	*SLC25A21*	X					Muscle atrophy	Organic acids (U), Quinolinic acid (U)		607571	IEM1209
Mitochondrial ribosomal small subunit 25 deficiency	*MRPS25*	X						Lactate (P, CSF)		619025	IEM1539
Mitochondrial ribosomal small subunit 39 deficiency	*PTCD3*		X					Lactate (P)		619057	IEM1540
Disorders of mitochondrial tRNA											
Mitochondrial tRNA(Ala) deficiency	*MT-TA*	X					Myalgia	Lactate (P)		590000	IEM0530
Mitochondrial tRNA(Asn) deficiency	*MT-TN*	X						Lactate (P)		590010	IEM0532
Mitochondrial tRNA(Asp) deficiency	*MT-TD*			X			Muscle pain	Lactate (P)		590015	IEM0533
Mitochondrial tRNA(Glu) deficiency (with COX Deficiency or Diabetes Mellitus)	*MT-TE*	X						Lactate (P)		500009	IEM0535
Mitochondrial tRNA(Gln) deficiency	*MT-TQ*	X						Lactate (P)		590030	IEM0536
Mitochondrial tRNA(Gly) deficiency	*MT-TG*	X		X			Myalgia	Lactate (P)		590035	IEM0537
Mitochondrial tRNA(His) deficiency	*MT-TH*	X						Lactate (P)		590040	IEM0538
Mitochondrial tRNA(Leu) 1 deficiency	*MT-TL1*	X		X	X			Lactate (P)		590050	IEM0540
Mitochondrial tRNA(Leu) 2 deficiency	*MT-TL2*	X						Lactate (P)		590055	IEM0541
Mitochondrial tRNA(Lys) deficiency	*MT-TK*	X						Lactate (P)		590060	IEM0542
Mitochondrial tRNA(Met) deficiency	*MT-TM*	X		X				Lactate (P)		590065	IEM0543

Name	Gene	Weakness	Hypotonia	Exercise intolerance	Rhabdomyolysis	Abnormal pathology	Other	Laboratory investigations	Specific treatment	OMIM diserase no.	IEMbase ID (hyperlinke
Mitochondrial tRNA(Pro) deficiency	*MT-TP*	X						Lactate (P)		590075	IEM0545
Mitochondrial tRNA(Val) deficiency	*MT-TV*	X						Lactate (P)		590105	IEM0551
Disorders of mitochondrial tRNA incorporation and recycling											
Mitochondrial asparaginyl-tRNA synthetase deficiency	*NARS2*	X						Lactate (P)		616239	IEM0554
Mitochondrial tyrosyl-tRNA synthetase deficiency	*YARS2*	X		X				Lactate (P), Blood count		613561	IEM0564
Mitochondrial valyl-tRNA synthetase deficiency	*VARS2*	X						Lactate (P)		615917	IEM0565
Mitochondrial tryptophanyl-tRNA synthetase deficiency	*WARS2*	X						Lactate (P)		617710	IEM0566
Peptidyl-tRNA hydrolase 2 deficiency	*PTRH2*	X						DNA		616263	IEM0571
Mitochondrial and cytoplasmic glycyl-tRNA synthetase deficiency	*GARS1*	X					Muscle atrophy	DNA		601472;619042	IEM1537
Disorders of mitochondrial fission											
Dynamin-like protein 1 deficiency	*DNM1L*	X						VLCFA (P), Lactate (P)		614388	IEM0572
GDAP1 deficiency	*GDAP1*	X					Muscular atrophy	DNA		607831;214400	IEM0574
UGO-1 like protein deficiency	*SLC25A46*						Lower limb muscle hypotrophy	Lactate (P), Organic acids (U)		616505	IEM0576
OPA1 deficiency (Behr syndrome)	*OPA1*	X						Lactate (P)		125250	IEM0577
Mitofusin 2 deficiency	*MFN2*	X					Muscle atrophy	DNA		609260;617087	IEM0579
MSTO1 deficiency	*MSTO1*	X						DNA		617675	IEM0580
MIEF2 deficiency	*MIEF2*	X					Muscle pain	CK (P), DNA		619024	IEM1378
Disorders of mitochondrial phospholipid metabolism											
Acylglycerol kinase deficiency (Sengers syndrome)	*AGK*	X		X			Skeletal myopathy	Lactate (P), Organic acids (U)		212350;614691	IEM0581
Tafazzin deficiency (Barth syndrome)	*TAZ*	X	X	X				Organic acids (U), Lipid panel (S), Urinalysis, 3-Methylglutaconic acid (U), 3-Methylglutaric acid (U), Carnitine, free (P), Blood count	Elamipretide	302060	IEM0583
PNPLA8 deficiency	*PNPLA8*	X						Lactate (P), Pyruvate (P)		251950	IEM0584
MICOS complex subunit MIC13 deficiency	*MICOS13*	X	X					Lactate (P), Organic acids (U)		616658	IEM1375
MICOS complex subunit MIC26 deficiency	*APOO*	X						Lactate (P), Pyruvate (P), Acylcarnitines (P, DBS)		300753	IEM1376
CHCHD2 deficiency	*CHCHD2*		X					DNA		616710	IEM1377
Disorders of mitochondrial protein import											
TIMM22 deficiency	*TIMM22*	X						CK (P), Lactate (P)		607251	IEM1262
Disorders of mitochondrial protein quality control											
Mitochondrial processing peptidase alpha deficiency	*PMPCA*	X						DNA		213200	IEM0591
CLPB deficiency	*CLPB*		X					DNA		616271	IEM0594
Parkin deficiency	*PRKN*		X					DNA		600116	IEM0603
PINK1 deficiency	*PINK1*		X					DNA		605909	IEM0604
Valosin-containing protein superactivity	*VCP*	X						DNA		167320	IEM0606
Other disorders of mitochondrial homeostasis											
C1q binding protein deficiency	*C1QBP*	X						Lactate (P)		617713	IEM0613
Mitochondrial calcium uniporter 1 deficiency	*MICU1*	X						CK (P)		615673	IEM0615
CHCHD10 deficiency	*CHCHD10*	X						CK (P)		615911;615048	IEM1081

Name	Gene	Weakness	Hypotonia	Exercise intolerance	Rhabdomyolysis	Abnormal pathology	Other	Laboratory investigations	Specific treatment	OMIM dierase no.	IEMbase ID (hyperlinke
Primary CoQ10 deficiencies											
Prenyl diphosphate synthase subunit 2 deficiency	*PDSS2*	X						Lactate (P), CoQ10 (M, P, WBC)		614652	IEM0619
Coenzyme Q2 polyprenyltranferase deficiency	*COQ2*	X						Lactate (P), CoQ10 (M, P, WBC)		609825;607426	IEM0620
Coenzyme Q8A (ADCK3) deficiency	*COQ8A*	X						Lactate (P), CoQ10 (M, P, WBC)		612016	IEM0624
ADCK2 deficiency	*ADCK2*	X						DNA			IEM1664
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=22)											
Disorders of heme metabolism											
Delta-aminolevulinate dehydratase deficiency	*ALAD*						Muscle pain	Porphyrines (U), Delta-ALA (U)		125270	IEM0788
Porphobilinogen deaminase deficiency	*HMBS*						Muscle pain	Urobilinogen (U), Porphyrines (U), Delta-ALA (U)	Intravenous hemin (haem arginate or haematin) 3-4 mg/kg OD x 4days; carbohydrate infusion Dextrose 10%; liver transplantation	176000	IEM0789
Coproporphyrinogen oxidase deficiency	*CPOX*						Muscle pain	PBG (U), Porphyrins (U, stools), Porphobilinogen (U)	Intravenous hemin (haem arginate or haematin) 3-4 mg/kg OD x 4days; carbohydrate infusion Dextrose 10%; liver transplantation	121300	IEM0792
Protoporphyrinogen oxidase deficiency	*PPOX*						Muscle pain	PBG (U), Porphyrins (U, stools), Fluorescence scanning (P)	Hemin, givosiran	176200	IEM0794
Disorders of nucleotide metabolism											
RNA-specific adenosine deaminase 2 deficiency	*ADARB1*	X						DNA		618862	IEM1278
Ectonucleoside triphosphate diphosphohydrolase 1 deficiency	*ENTPD1*	X						DNA		615683	IEM1279
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases											
Methionyl-tRNA synthetase 1 deficiency	*MARS1*	X						DNA		156560	IEM1312
Seryl-tRNA synthetase 1 deficiency	*SARS1*	X						DNA		617709	IEM1315
Tryptophanyl-tRNA synthetase 1 deficiency	*WARS1*	X					Muscle atrophy	DNA		617721	IEM1317
Tyrosyl-tRNA synthetase 1 deficiency	*YARS1*	X					Muscle atrophy	DNA		608323	IEM1318
AIMP1/p43 deficiency	*AIMP1*		X					DNA		260600	IEM1320
AIMP2/p38 deficiency	*AIMP2*	X						DNA		618006	IEM1321
Disorders of ribosomal biogenesis											
Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	*UBTF*	X						DNA		617672	IEM1333
RPL3L deficiency	*RPL3L*	X						DNA		619371	IEM1672
Disorders of organelle interplay											
Griscelli syndrome type 2	*RAB27A*		X					DNA		607624	IEM1395
Disorders of choline neurotransmission											
VAPB deficiency	*VAPB*						Muscular atrophy	DNA		608627	IEM1399
Choreoacanthocytosis	*VPS13A*	X						DNA		200150	IEM1400
VPS13C deficiency	*VPS13C*		X					DNA		616840	IEM1401
Choline transporter deficiency	*SLC5A7*	X						DNA		617143	IEM1449
Choline acetyltransferase deficiency	*CHAT*	X						DNA		254210	IEM1450
RIC3 acetylcholine receptor chaperone deficiency	*RIC3*		X					DNA		610509	IEM1451
Acetylcholine receptor subunit epsilon deficiency	*CHRNE*	X						DNA		605809;616324;608931	IEM1452
DISORDERS OF LIPIDS (n=36)											
Disorders of carnitine metabolism											

Name	Gene	Weakness	Hypotonia	Exercise intolerance	Rhabdomyolysis	Abnormal pathology	Other	Laboratory investigations	Specific treatment	OMIM dierase no.	IEMbase ID (hyperlinke
Primary carnitine deficiency	*SLC22A5*	X	X		X		Skeletal myopathy	Total/free carnitine (DBS, P)	Carnitine	212140	IEM0627
Carnitine palmitoyltransferase 2 deficiency	*CPT2*	X	X		X		Skeletal myopathy	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P)	Frequent feeds, low-fat diet, MCT supplementation	255110	IEM0629
Carnitine acylcarnitine translocase deficiency	*SLC25A20*	X	X				Skeletal myopathy	Ammonia (P), Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Glucose (S)	Frequent feeds, low-fat diet, MCT supplementation	212138	IEM0630
Carnitine palmitoyl-transferase 1C deficiency	*CPT1C*	X					Muscle atrophy	DNA		616282	IEM1165
Disorders of fatty acid oxidation and transport											
Short-chain acyl CoA dehydrogenase deficiency (Ethylmalonic aciduria)	*ACADS*			X				Organic acids (U), Acylcarnitines (DBS, P)		201470	IEM0115
Very long-chain acyl CoA dehydrogenase deficiency	*ACADVL*	X			X		Skeletal myopathy	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Glucose (S), Dicarboxylic acids (U)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	201475	IEM0634
Isolated deficiency of long-chain 3-hydroxyacyl-CoA dehydrogenase	*HADHA*	X	X		X		Skeletal myopathy	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Dicarboxylic acids (U)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	600890	IEM0636
Trifunctional protein subunit α deficiency	*HADHA*	X					Skeletal myopathy	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Dicarboxylic acids (U)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	609015	IEM0636
Isolated deficiency of long-chain 3-ketoacyl CoA thiolase (1 patient)	*HADHB*	X	X				Skeletal myopathy	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Dicarboxylic acids (U)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	143450	IEM1247
Trifunctional protein subunit β deficiency	*HADHB*	X					Skeletal myopathy	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Dicarboxylic acids (U)	Frequent feeds, low-fat diet, MCT supplementation; triheptanoin	609015	IEM0637
TANGO2 deficiency	*TANGO2*				X			Organic acids (U), Acylcarnitines (DBS, P), CK (P), Lactate (P), Glucose (S), Dicarboxylic acids (U)		616878	IEM0640
Disorders of fatty acid synthesis and elongation											
3-Hydroxyacyl-CoA dehydratase 1 deficiency	*HACD1*	X						DNA		610467	IEM1172
Cytosolic acetyl-CoA carboxylase 1 deficiency	*ACACA*	X						Organic acids (U)		613933	IEM1170
Disorders of glycerol metabolism											
D-glycerate kinase deficiency	*GLYCTK*	X	X					D-glycerate (P, U, CSF)		610516	IEM0653
Disorders of cytoplasmic triglyceride metabolism											
Lipin 1 deficiency	*LPIN1*	X			X		Muscle cramps	CK (P), Myoglobin (U)		268200	IEM0657
Adipose triglyceride lipase deficiency	*PNPLA2*	X				X	Muscle atrophy	CK (P)		610717	IEM0661
Perilipin 1 deficiency	*PLIN1*						Lipodystrophy	Cholesterol (S), Triglyceride (S)		613877	IEM0662
Hormone-sensitive lipase deficiency	*LIPE*						Lipodystrophy	CK (P), Cholesterol (S), Triglyceride (S)		615980	IEM0663
Seipin superactivity	*BSCL2*	X						AST/ALT (P), Triglyceride (S), Insulin (S)		600794;270685	IEM1055
Disorders of non-mitochondrial phospholipid metabolism											
Choline kinase β deficiency	*CHKB*	X				X		CK (P)		602541	IEM0664
Phosphocholine cytidylyltransferase 1α deficiency, lipodystrophy phenotype	*PCYT1A*						Lipodystrophy	Acetylcholine (P), Phosphocholine (P)		608940	IEM0666
Phosphatidylserine flippase deficiency	*ATP8A2*		X					DNA		615268	IEM0668
Disorders of palmitoylation											
Palmitoyl-protein thioesterase 1 deficiency	*PPT1*						Muscular atrophy	Lysosomal enzymes (DBS)		256730	IEM0689
Disorders of phosphoinositide metabolism											
Synaptojanin 1 deficiency	*SYNJ1*		X					Lactate (P)		615530	IEM0693
Myotubularin 1 deficiency	*MTM1*	X						DNA		310400	IEM0694
Myotubularin-related protein 2 deficiency	*MTMR2*	X						DNA		601382	IEM0695
Myotubularin-related protein 2 regulatory protein deficiency	*SBF2*	X						DNA		604563	IEM0696
Myotubularin-related protein 2 activator deficiency	*SBF1*						Muscle atrophy	DNA		615284	IEM0697
Phosphatidylinositol 3-kinase regulatory subunit 2 superactivity	*PIK3R2*						Muscle atrophy	DNA		603387	IEM0701

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Phosphatidylinositol 4-phosphate 5-kinase deficiency	*PIP5K1C*	X					Muscle atrophy	DNA		611369	IEM0703
Inositol polyphosphate 5-phosphatase K deficiency	*INPP5K*	X						DNA		617404	IEM1037
Disorders of lipoprotein metabolism											
Hereditary apolipoprotein A1-related amyloidosis	*APOA1*	X						Lipid panel (S), Apolipoprotein A-I		105200	IEM0736
Mevalonate kinase deficiency	*MVK*		X					Leucotriens (P), Organic acids (U)	Inflammatory control (non-steroidal anti-inflammatory drugs, corticosteroids, IL-1 targeting biologic agents, TNF-alpha blockade,	610377	IEM0740
Geranylgeranyl pyrophosphate synthase deficiency	*GGPS1*	X						CK (P)		606982	IEM1185
Disorders of steroid metabolism											
X-linked spinal and bulbar muscular atrophy (Kennedy disease)	*AR*						Muscular atrophy	Steroids (P)		313200	IEM0777
Disorders of bile acid synthesis											
α-Methylacyl-CoA racemase deficiency	*AMACR*				X			Bile acids (U), VLCFA (P), Vitamins D/E (P)	Cholic acid	604489	IEM0783
STORAGE DISORDERS (n=30)											
Disorders of autophagy											
Spatacsin deficiency	*SPG11*	X					Muscle atrophy	DNA		616668	IEM0814
TBK1 deficiency	*TBK1*	X						DNA		616439	IEM0818
RAB7 deficiency	*RAB7A*	X						DNA		600882	IEM0819
SQSTM1 deficiency	*SQSTM1*	X					Muscle atrophy	DNA		617158	IEM1228
TBCK deficiency	*TBCK*	X						DNA		616899	IEM1236
ALS2 deficiency	*ALS2*	X						DNA		607225	IEM1239
DCTN1 deficiency	*DCTN1*	X					Muscle atrophy	DNA		607641	IEM1240
MTMR14 deficiency	*MTMR14*	X						DNA		611089	IEM1242
CHMP2B deficiency	*CHMP2B*	X	X				Muscle atrophy	DNA		614696	IEM1243
ATG7 deficiency	*ATG7*	X						DNA		619422	IEM1677
Neuronal ceroid lipofuscinosis											
Tripeptidyl-peptidase 1 deficiency (Jansky-Bielschowsky disease)	*TPP1*						Muscular atrophy	Lysosomal enzymes (DBS)		204500	IEM0820
CLN3 disease	*CLN3*	X	X				Muscular atrophy	Peripheral smear, DNA	In trial gene therapy (NCT03770572)	204200	IEM0821
CLN5 disease	*CLN5*						Muscular atrophy	DNA		256731	IEM0823
CLN6 disease	*CLN6*						Muscular atrophy	DNA		601780	IEM0824
CLN7 disease	*MFSD8*						Muscular atrophy	DNA	Antisense oligonucleotide (N=1)	610951	IEM0825
CLN8 disease	*CLN8*						Muscular atrophy	DNA		600143	IEM0826
Cathepsin D deficiency	*CTSD*						Muscular atrophy	Lysosomal enzymes (DBS)		610127	IEM0827
Progranulin deficiency	*GRN*						Muscular atrophy	DNA		614706	IEM0828
ATP13A2 deficiency (Kufor-Rakeb syndrome;)	*ATP13A2*		X					DNA		606693	IEM0829
Cathepsin F deficiency (Kufs disease)	*CTSF*						Muscular atrophy	DNA		603539	IEM0830
CLN14 disease	*KCTD7*						Muscular atrophy	DNA		611726	IEM0831
Sphingolipidoses											
Beta-hexosaminidase subunit alpha deficiency (Tay-Sachs disease)	*HEXA*	X						Oligosaccharides (U), Lysosomal enzymes (DBS)		272800	IEM0836

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Beta-hexosaminidase subunit beta deficiency (Sandhoff disease)	*HEXB*	X						Oligosaccharides (U), Lysosomal enzymes (DBS)		268800	IEM0837
GM2 activator protein deficiency	*GM2A*		X					Oligosaccharides (U), Lysosomal enzymes (DBS)		272750	IEM0838
Arylsulfatase A deficiency (Metachromatic leukodystrophy)	*ARSA*	X						Enzyme (DBS, L, F), Protein (CSF), Sulfatides (U)	Hematopoietic stem cell transplantation (HSCT)	250100	IEM0841
Metachromatic leukodystrophy-like disorder due to saposin B deficiency	*PSAP*	X						Sulfatides (U), Protein (CSF), Lysosomal enzymes (DBS)		249900	IEM0842
Formyl-glycine generating enzyme deficiency	*SUMF1*	X	X					Sulfatide (U), Glycosaminoglycans (U)		272200	IEM0843
Acid ceramidase deficiency, primary neurologic phenotype (Farber disease)	*ASAH1*	X					Muscular atrophy	Lysosomal enzymes (DBS)		228000	IEM0846
Mucolipidoses											
Mucolipin 1 deficiency	*MCOLN1*	X						Gastrin (S)		252650	IEM0857
Disorders of lysosomal transport or sorting											
Nephropathic cystinosis	*CTNS*	X						Cystine (WBC, PMN)	Cysteamine	219800;219900;219750	IEM0873
DISORDERS OF PEROXISOMES AND OXALATE (n=17)											
Disorders of peroxisomal β-oxidation											
Peroxisomal straight-chain acyl-CoA oxidase deficiency	*ACOX1*	X	X					VLCFA (P), Plasmalogens (P)		264470	IEM0884
D-bifunctional protein deficiency (Pseudo-Zellweger syndrome)	*HSD17B4*	X	X					VLCFA (P), Plasmalogens (P), Organic acids (U)		261515	IEM0885
Phytanoyl-CoA hydroxylase deficiency (Classic Refsum disease)	*PHYH*						Muscular atrophy	Pipecolic acid (P, U), Phytanic acid (S, U), Protein (CSF)	Phytanic acid restriction	266500	IEM0888
Acyl-CoA-binding domain-containing protein 5 deficiency	*ACBD5*	X						VLCFA (P), Pipecolic acid (P)		616618	IEM1191
Peroxin 1 deficiency (Zellweger)	*PEX1*	X	X					VLCFA (P), Pipecolic acid (P)		234580;214100;601539	IEM0889
Peroxin 2 deficiency (Zellweger)	*PEX2*	X	X					VLCFA (P), Pipecolic acid (P)		614866;614867	IEM0890
Peroxin 3 deficiency (Zellweger)	*PEX3*	X	X					VLCFA (P), Pipecolic acid (P)		617370;614882	IEM0891
Peroxin 5 deficiency (Zellweger)	*PEX5*	X	X					ASAT/ALAT (P), VLCFA (P), Pipecolic acid (P, U)		214110	IEM0892
Peroxin 6 deficiency (Zellweger)	*PEX6*	X	X					VLCFA (P), Pipecolic acid (P)		614862;614863;616617	IEM0893
Peroxin 10 deficiency (Zellweger)	*PEX10*	X	X					VLCFA (P), Pipecolic acid (P)		614870;614871	IEM0894
Peroxin 14B deficiency (Zellweger)	*PEX11B*	X	X					VLCFA (P), Pipecolic acid (P)		614920	IEM0895
Peroxin 12 deficiency (Zellweger)	*PEX12*	X	X					VLCFA (P), Pipecolic acid (P)		614859;266510	IEM0896
Peroxin 13 deficiency (Zellweger)	*PEX13*	X	X					VLCFA (P), Pipecolic acid (P)		614883;614885	IEM0897
Peroxin 14 deficiency (Zellweger)	*PEX14*	X	X					VLCFA (P), Pipecolic acid (P)		614887	IEM0898
Peroxin 16 deficiency (Zellweger)	*PEX16*	X	X					VLCFA (P), Pipecolic acid (P)		614876;614877	IEM0899
Peroxin 19 deficiency (Zellweger)	*PEX19*	X	X					VLCFA (P), Pipecolic acid (P)		614886	IEM0900
Peroxin 26 deficiency (Zellweger)	*PEX26*	X	X					VLCFA (P), Pipecolic acid (P)		614872;614873	IEM0901
CONGENITAL DISORDERS OF GLYCOSYLATION (n=39)											
Disorders of N-linked glycosylation											
DPAGT1-CDG	*DPAGT1*					X		Sialotrasferrins (S)		608093	IEM0910
MGAT2-CDG	*MGAT2*	X						ASAT/ALAT (P), CK (P), Sialotransferrins (S), Factor IX and XII (B), Antithrombin III (P)		212066	IEM0931
Disorders of O-mannosylation											
POMT1-CDG	*POMT1*	X						CK (P), Sialotrasferrins (S)		236670;613555;609308	IEM0933
POMT2-CDG	*POMT2*	X						CK (P), DNA		613150;613156;613158	IEM0934

Name	Gene	Weakness	Hypotonia	Exercise intolerance	Rhabdomyolysis	Abnormal pathology	Other	Laboratory investigations	Specific treatment	OMIM dierase no.	IEMbase ID (hyperlinke
POMGNT1-CDG	*POMGNT1*	X					Muscle-eye-brain disease	CK (P), DNA		253280;613151;613157	IEM0935
POMGNT2-CDG	*POMGNT2*	X						CK (P), DNA		614830;618135	IEM0936
B3GALNT2-CDG	*B3GALNT2*	X					Muscle-eye-brain disease	CK (P), DNA		615181	IEM0937
POMK-CDG	*POMK*	X					Muscle-eye-brain disease; Muscle	CK (P), DNA		616094;615249	IEM0938
CRPPA-CDG	*CRPPA*	X					Muscle-eye-brain disease	CK (P), DNA		614643;616052	IEM0939
FKTN-CDG A	*FKTN*	X					Muscle-eye-brain disease; Muscle	CK (P), DNA		253800	IEM0940
FKTN-CDG B	*FKTN*	X						CK (P), DNA		613152	IEM0940
FKTN-CDG C	*FKTN*	X						CK (P), DNA		611588	IEM0940
FKRP-CDG A	*FKRP*	X					Muscle-eye-brain disease	CK (P), DNA		613153	IEM0941
FKRP-CDG B	*FKRP*	X						CK (P), DNA		606612	IEM0941
FKRP-CDG C	*FKRP*	X			X		Muscle hypertrophy;	CK (P), DNA		606596	IEM0941
RXYLT1-CDG	*RXYLT1*	X						CK (P), DNA		615041	IEM0942
B4GAT1-CDG (Walker-Warburg syndrome)	*B4GAT1*	X						CK (P), DNA		615287	IEM0943
LARGE1-CDG	*LARGE1*	X					Muscle hypertrophy	CK (P), DNA		613154;608840	IEM0944
Disorders of O-xylosylation and glycosaminoglycan synthesis											
CHST14-CDG	*CHST14*	X						DNA		601776	IEM0956
Sulfate transporter deficiency	*SLC26A2*	X						DNA		226900;222600;256050;600972	IEM0960
Disorder of O-glucosylation											
POGLUT1-CDG LGMDR21	*POGLUT1*	X						DNA		615696	IEM0967
Disorders of glycosylphosphatidylinositol biosynthesis											
PIGS-CDG	*PIGS*	X						DNA		618143	IEM1059
Disorders of dolichol metabolism											
SRD5A3-CDG	*SRD5A3*	X	X					Antithrombin III (P), Protein S (S), Sialotrasferrins (S)		612379	IEM0993
DPM2-CDG	*DPM2*	X						ASAT/ALAT (P), CK (P), Sialotransferrins (S)		615042	IEM0996
DPM3-CDG	*DPM3*	X						ASAT/ALAT (P), CK (P), Sialotransferrins (S), Dolichol-P-mannose (S)		612937	IEM0997
Disorders of monosaccharide synthesis and interconversion											
UDP-GlcNAc epimerase-kinase deficiency (Nonaka myopathy)	*GNE*	X				X	Muscle wasting	CK (P), Sialotransferrins (S)		600737;605820	IEM0999
PGM1-CDG	*PGM1*	X			X			ASAT/ALAT (P), CK (P), Ammonia (P), Sialotransferrins (S)		614921	IEM1003
N-acetylneuraminate pyruvate lyase deficiency	*NPL*	X						Sialic acid, free (U)		611412	IEM1212
Disorders of nucleotide-sugar synthesis											
GMPPB-CDG	*GMPPB*	X			X			CK (P), Sialotrasferrins (S)		615350;615351;615352	IEM1007
UGDH-CDG	*UGDH*	X						DNA		618792	IEM1556
Disorders of vesicular trafficking											
TRAPPC11-CDG	*TRAPPC11*	X					Muscle pain	CK (P)		615356	IEM1022
TRAPPC2L deficiency	*TRAPPC2L*	X			X			CK (P)		618331	IEM1408
TRAPPC4 deficiency	*TRAPPC4*	X						DNA		618741	IEM1409

Name	Gene	Weakness	Hypotonia	Exercise intolerance	Rhabdomyolysis	Abnormal pathology	Other	Laboratory investigations	Specific treatment	OMIM dierase no.	IEMbase ID (hyperlinke
SCYL1 deficiency	*SCYL1*	X					Muscle atrophy	ASAT/ALAT (P), GGT (P)		616719	IEM1416
AP4S1 deficiency	*AP4S1*	X						DNA		607243	IEM1232
Rubicon deficiency	*RUBCN*	X						DNA		615705	IEM1431
Disorders of Golgi homeostasis											
TMEM165-CDG	*TMEM165*	X						ASAT/ALAT (P), CK (P), Sialotransferrins (S)		614727;614726	IEM1031
SLC9A7-CDG	*SLC9A7*	X						DNA		300368	IEM1063
VMA21-CDG	*VMA21*	X					Muscle atrophy	ASAT/ALAT (P), CK (P)		310440	IEM1555
OTHER (n=12)											
Disorders of the synaptic vesicle cycle											
DYNC1H1 deficiency	*DYNC1H1*	X						DNA		614228;614563;158600	IEM1457
Dynamin 1 deficiency	*DNM1*	X						DNA		616346	IEM1458
Dynamin 2 deficiency	*DNM2*	X						DNA		606482;602378;160150	IEM1459
SNAP25 deficiency	*SNAP25*	X						DNA		616330	IEM1463
Synaptobrevin 1 deficiency	*VAMP1*	X						DNA		618323	IEM1467
Synaptotagmin 2 deficiency	*SYT2*	X						DNA		616040	IEM1474
Clathrin heavy chain deficiency	*CLTC*	X						DNA		617854	IEM1479
Disorders of lysosome-related organelle biogenesis											
X-Linked hypophosphatemia	*PHEX*	X						ALP (P), Phosphate (P)		307800	IEM1495
Kearns Sayre Syndrome		X						Protein (CSF), 5-Methyl-THF (CSF)		530000	IEM1502
Maternally Inherited Deafness and Diabetes (Ballinger-Wallace Syndrome)	*MT-TL1, MT-TE, MT-*	X						Glucose (P)		520000	IEM1504
Leigh Syndrome		X						Lactate (P)		256000	IEM1505
Miscalenous											
C2orf69 deficiency	*C2ORF69*	X	X					DNA		619423	IEM1678