Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
DISORDERS OF NITROGEN-CONTAINING COMPOUNDS (n=60)													
Disorders of pyrimidine metabolism													
Dihydroorotate dehydrogenase deficiency	*DHODH*	AR		X	X					N-Carbamyl asparate (U), Purines and pyrimidines (U)		263750;126064	IEM0002
Uridine monophosphate synthase deficiency	*UMPS*	AR			X					Purines and pyrimidines (U)		258900	IEM0003
Deoxythymidylate kinase deficiency	*DTYMK*	AR	X							Amino acids (P), Lactate (P, CSF)		188345	IEM1271
Disorders of purine metabolism													
Myoadenylate deaminase deficiency	*AMPD1*	AR						X	Nausea	CK (P)		102770	IEM0010
Inosine triphosphatase deficiency	*ITPA*	AR					X			ITP (RBC), DNA		147520	IEM0023
Disorders of creatine metabolism													
Creatine transporter deficiency	*SLC6A8*	XL			X					Creatinine (P, U), Guanidino compounds (P, U)	Creatine, arginine and glycine supplementation	300352	IEM0045
Disorders of glutathione metabolism													
5-Oxoprolinase deficiency	*OPLAH*	AR			X					5-Oxoproline (U)		260005	IEM0052
Disorders of ammonia detoxification													
N-Acetylglutamate synthase deficiency	*NAGS*	AR					X	X		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)		237310	IEM0056
Carbamoyl phosphate synthetase I deficiency	*CPS1*	AR					X	X		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline, liver transplant	237300	IEM0057
Ornithine transcarbamylase deficiency	*OTC*	XL					X	X		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline, liver transplant	311250	IEM0058
Argininosuccinate synthetase deficiency Citrullinemia type I	*ASS1*	AR					X	X		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline, liver transplant	215700	IEM0059
Argininosuccinate lyase deficiency	*ASL*	AR					X	X		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline, liver transplant	207900	IEM0060
Arginase 1 deficiency	*ARG1*	AR					X	X		Ammonia (P), Amino acids (P), Purines and pyrimidines (U)	Dietary protein restriction, essential amino acid supplementation; Na phenylbutyrate <20 kg: ≤250mg/kg/dc >20 kg: 5g/m2/d maximum: 12g/day; Na benzoate 250mg/kg/d maximum: 12g/d	207800	IEM0061
Mitochondrial ornithine transporter deficiency	*SLC25A15*	AR					X	X		Orotic acid (U), Ammonia (B), Homocitrulline (U), Amino acids (P)	Dietary protein restriction, essential amino acid supplementation; Na benzoate 250mg/kg/d maximum: 12g/d; Na phenylbutyrate 250mg/kg/d maximum: 12g/d; L-Arginine <20 kg: 100-200mg/kg/d >20 kg: 2.5-6g/m2/d maximum: 6g/d; L-Citrulline 100-200mg/kg/d	238970	IEM0062
Citrin deficiency	*SLC25A13*	AR				X				Ammonia (P), Amino acids (P)	Low carbohydrate diet, MCT oil, ammonia scavengers, liver transplant	605814;603471	IEM0063
Carbonic anhydrase VA deficiency	*CA5A*	AR					X	X		Amino acids (P), Ammonia (P), Glucose (P), Lactate (P)		615751	IEM0064
Disorders of amino acid transport													
Lysinuric protein intolerance	*SLC7A7*	AR			X			X	Protein intolerance	Amino acids (P, U), Ammonia (P), Ferritin (S), Lipid panel (S), Orotic acid (U)	Protein restriction, ammonia scavengers, citrulline	222700	IEM0070
Disorders of monoamine metabolism													
Tyrosine hydroxylase deficiency	*TH*	AR					X		Drooling	Prolactin (P), Biogenic amines (CSF)	L-dopa/Carbidopa	191290	IEM0076
Aromatic L-amino acid decarboxylase deficiency	*DDC*	AR					X		Drooling	3-O-Methylodopa (DBS,P), Organic acids (U), Biogenic amines (CSF)	Pyridoxine, dopa agonist, MAO inhibitor, central anticholinergic	608643	IEM0077
Dopamine beta-hydroxylase deficiency	*DBH*	AR						X	Nausea	Norepinephrine (P, U), Dopamine (P), HVA (CSF), 5HIAA (CSF), L-dopa (CSF)	L-dihydroxyphenylserine (L-DOPS) 100-500 mg po BID or TID	223360	IEM0078

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Dopamine transporter deficiency	*SLC6A3*	AR					X		Drooling	Organic acids (U), Biogenic amines (CSF)	Dopamine agonists	613135;126455	IEM0080
Disorders of phenylalanine and tetrahydrobiopterin metabolism													
Phenylalanine hydroxylase deficiency	*PAH*	AR						X		Amino acids (P)	Phe-restricted diet, large neutral amino acids, glycomacropeptide, sapropterin, pegvaliase	261600	IEM0082
GTP cyclohydrolase 1 deficiency	*GCH1*	AR	X				X		Drooling	Amino acids (P), Pterins (DBS, U, CSF), Biogenic amines (CSF)	L-dopa/Carbidopa	233910	IEM0083
6-Pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	AR							Drooling	Amino acids (P), Pterins (DBS, U, CSF), Biogenic amines (CSF)	Tetrahydrobiopterin, L-dopa/dopa carboxylase inhibitor; 5-hydroxytrypptophan; +/- folinic acid	261640	IEM0085
Sepiapterin reductase deficiency	*SPR*	AR			X				Dysmotility	Pterins (U, CSF), Biogenic amines (CSF)	L-dopa/dopa carboxylase inhibitor; 5-hydroxytrypptophan; MAO inhibitor, serotonin reuptake inhibitor, dopamine agonist, anticholinergics, melatonin	182125	IEM0086
Dihydropteridine reductase deficiency	*QDPR*	AR							Drooling	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
Disorders of sulfur amino acid and sulfide metabolism													
Isolated sulfite oxidase deficiency	*SUOX*	AR					X			α-Aminosemialdehyde (CSF), PLP (CSF), Sulfite (U), Amino acids (P)	Trials of dietary therapy (no proven benefit) low-protein diet restricted in cysteine and methionine; experimental (with minimal or no beneftis): betaine, thiamine, cysteamine, penicillamin	272300	IEM0105
Mitochondrial sulfur dioxygenase deficiency Ethylmalonic encephalopathy	*ETHE1*	AR			X					Organic acid (U), acylcarnitines (P), thiosulphate (P), lactate (B)	Trials of antioxidants (CoQ10, Riboflavin), experimental therapy (no proven benefit) with N-acetylcystein, Metronidazole; Orthotopic liver transplant	602473;608451	IEM0106
Disorders of branched-chain amino acid metabolism													
Branched-chain aminotransferase 2 deficiency	*BCAT2*	AR					X	X		Amino acids (P)		238340;113530	IEM0107
Branched-chain ketoacid dehydrogenase E1 alpha deficiency	*BCKDHA*	AR				X	X	X		Amino acids (P), Organic acids (U)		248600	IEM0108
Branched-chain ketoacid dehydrogenase E1 beta deficiency	*BCKDHB*	AR				X	X	X		Amino acids (P), Organic acids (U)		248600	IEM0109
Dihydrolipoyl transacylase deficiency	*DBT*	AR				X	X	X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Urinalysis	Dietary leucine restriction, BCAA-free medical foods, judicious supplementation with isoleucine and valine, hemodialysis/hemofiltration, trial of enteral thiamine 50-100 mg/day, divided 2x/day 4 week trial, Transplantation of allogeneic liver tissue	248600	IEM0110
Isovaleryl-CoA dehydrogenase deficiency	*IVD*	AR				X	X	X		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Urinalysis	Protein restricted diet; Carnitine 50-100 mg/kg/day; Glycine 150-259 mg/kg/day	243500	IEM0113
Isobutyryl-CoA dehydrogenase deficiency	*ACAD8*	AR						X		Acylcarnitines (DBS, P), Acylglycines (U), Carnitine (P)	Levocarnitine	611283	IEM0114
3-hydroxyisobutyryl-CoA hydrolase deficiency	*HIBCH*	AR					X			Organic acids (U), S-(2-carboxypropyl)-cysteine (U), S-2-carboxypropyl-cysteamine (U)	Moderate protein restricted diet	250620	IEM0120
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*	AR				X				Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), Lactate (P), Ammonia (P)	Low protein diet, carnitine	246450	IEM0122
Methylmalonate semialdehyde dehydrogenase deficiency	*ALDH6A1*	AR						X		Amino acids (P), Organic acids (U)		603178	IEM0123
Propionic acidemia due to propionyl-CoA carboxylase subunit alpha deficiency	*PCCA*	AR				X	X	X		Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, carnitine	232000	IEM0124
Propionic acidemia due to propionyl-CoA carboxylase subunit beta deficiency	*PCCB*	AR				X	X	X		Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, carnitine	232000	IEM0125
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	*MMUT*	AR				X	X	X		Amino acids (P), Organic acids (U), Acylcarnitines (U, P, DBS), Anion gap	Low protein diet, L-Carnitine 100-200 mg/kg/day, vitamin B12, acute management of hyerammonemic crises, orthotopic liver transplant	251000	IEM0127
Acety-CoA-synthase 3 deficiency	*ACSF3*	AR					X	X		Organic acids (U), Acylcarnitines (DBS, P)	mild protein restriction, cobalamin, low dose carnitine	614245	IEM0128
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR						X		Organic acids (U), Acylcarnitines (DBS, P), Lactate (P), Glucose (S), Lipid panel (S)	low fat, high carbohydrate diet, MCT oil, low dose L-carnitine	248360	IEM0129
Disorders of lysine metabolism													
Alpha-amino adipic semialdehyde (AASA) dehydrogenase deficiency	*ALDH7A1*	AR			X			X		Pyridoxal 5'-phosphate, PLP (CSF), B6 vitaminers (CSF, P, U), Pipecolic acid (CSF)		266100	IEM0131
Glutaryl-CoA dehydrogenase deficiency Glutaric acidemia type 1	*GCDH*	AR					X	X		ASAT/ALAT (P), Organic acids (U), Acylcarnitines (U, P, DBS)	Low lysine and tryptophan restricted diet, carnitine, riboflavin	231670	IEM0134

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Disorders of proline and ornithine metabolism													
Delta-1-pyrroline-5-carboxylate synthase deficiency, cutis laxa phenotype	*ALDH18A1*	AD, AR					X			Amino acids (P), Ammonia (P)	Arginine 150 mg/kg/day	219150	IEM0137
Pyrroline-5-carboxylate reductase 1 deficiency	*PYCR1*	AR		X						DNA		612940;614438	IEM0139
Pyrroline-5-carboxylate synthetase deficiency, spastic paraplegia type 9A	*ALDH18A1*	AD		X				X		Amino acids (P), Ammonia (P)	Arginine 150 mg/kg/day	138250;219150	IEM1485
Disorders of β- and γ-amino acids													
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	AR					X			Purines and pyrimidines (U, P)	No treatment in pediatrics, in adults discontinue fluorouracil treatment	274270;612779	IEM0148
GABA transaminase deficiency	*ABAT*	AR					X			GABA free (CSF), β-Alanine (CSF), Homocarnosine (CSF)		137150;613163	IEM0152
Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*	AR					X			Organic acids (U)	Vigabatrin (no proven benefit)	271980;610045	IEM0153
Disorders of tryptophan metabolism													
3-Hydroxykynureninase deficiency	*KYNU*	AR			X					3-Hydroxykynurenine (P, U), NAD+ (P), Kynurenine (U), Xanthurenic acid (U)		605197	IEM0162
Disorders of glutamate metabolism													
Glutamate aspartate transporter deficiency	*SLC1A3*	AD						X	Nausea	DNA		612656	IEM0167
Astroglial glutamate aspartate transporter deficiency	*SLC1A2*	AD					X			DNA		617105	IEM0168
Ionotropic glutamate receptor NMDA type subunit 1 dysregulation	*GRIN1*	AD, AR			X		X			DNA		614254;617820	IEM0169
Ionotropic glutamate receptor NMDA type subunit 2D superactivity	*GRIN2D*	AD	X							DNA		617162	IEM0172
Metabotropic glutamate receptor 1 superactivity	*GRM1*	AD	X							DNA		617691	IEM0177
Disorder of asparagine metabolism													
Asparagine synthetase deficiency	*ASNS*	AR					X			Amino acids (P)		615574	IEM0180
Disorders of glycine metabolism													
Nonketotic hyperglycinemia due to glycine decarboxylase deficiency	*GLDC*	AR					X			Amino acids (P, CSF)	Na benzoate 200-550 mg/kg/day to max 750 mg/kg/day or 5.5 g/m2 BSA in adults up to 16.5 g/m2/day in severe cases; dextrometorphan 3 to 15 mg/kg/day or ketamine	238300	IEM0185
Nonketotic hyperglycinemia due to aminomethyltransferase deficiency	*AMT*	AR					X			Amino acids (P, CSF)	Na benzoate 200-550 mg/kg/day to max 750 mg/kg/day or 5.5 g/m2 BSA in adults up to 16.5 g/m2/day in severe cases; dextrometorphan 3 to 15 mg/kg/day or ketamine	605899	IEM0186
Glycine transporter 1 deficiency	*SLC6A9*	AR					X			Glycine (CSF), Glycine (CSF) / Glycine (P) ratio		617301	IEM0187
DISORDERS OF VITAMINS, COFACTORS, METALS AND MINERALS (n=45)													
Disorders of lipoic acid and iron-sulfur metabolism													
Lipoic acid synthase deficiency	*LIAS*	AR					X			Amino acids (CSF, P), Lactate (P), Protein bound lipoic acid (FB)		614462	IEM0193
Lipoyltransferase 1 deficiency	*LIPT1*	AR						X		Lactate (P), Organic acids (U)		616299	IEM0194
NFU1 deficiency	*NFU1*	AR					X			Amino acids (P), Organic acids (U), Lactate (P, U, CSF), Protein bound lipoic acid (FB)		605711	IEM0195
BOLA3 deficiency	*BOLA3*	AR					X			Amino acids (CSF, P), Lactate (P)		614299	IEM0196

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IBA57 deficiency	*IBA57*	AR					X			Lactate (P), Amino acids (P)		615330	IEM0198
ISCA1 deficiency	*ISCA1*	AR					X			Lactate (P), Amino acids (P)		617613	IEM0199
ISCA2 deficiency	*ISCA2*	AR		X						5-Methyl-THF (CSF), Lactate (P), Amino acids (P)		616370	IEM0200
ISCU deficiency	*ISCU*	AR						X	Nausea	Lactate (P), Myoglobin (U), Complexes I - III activity (muscle)		255125	IEM0201
Ferredoxin reductase deficiency	*FDXR*	AR					X			Complexes I - IV activity (muscle), DNA		617717	IEM0203
Frataxin deficiency (Friedreich ataxia)	*FXN*	AR							Sphincter control problems	Glucose (S)		229300	IEM0204
NFS1 deficiency	*NFS1*	AR				X		X		Lactate (P)		603485	IEM1123
ISD11 deficiency	*LYRM4*	AR		X						Lactate (P)		615595	IEM1122
Ferredoxin 2 deficiency	*FDX2*	AR						X	Nausea	Myoglobin (U), Lactate (P), Organic acids (U)		614585	IEM1121
Disorders of cobalamin metabolism													
Transcobalamin 2 deficiency	*TCN2*	AR			X					Amino acids (P), Organic acids (U), Holotranscobalamin (P)		275350	IEM0209
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR					X			Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P), Blood count	Hydroxycobalamin 1 mg IM daily	277380	IEM0211
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR					X			Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P)	Hydroxycobalamin	614857	IEM0212
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR					X			Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)	Hydroxycobalamin 1 mg IM daily, betaine 250 mg/kg/day; folinic acid 5-30 mg/day; L-carnitine 50-200 mg/kg/day; no protein restriction; maintain normal methionine levels (+/- supplementation)	277400	IEM0213
Epi-cblC	*MMACHC; PRDX1*	AR					X			SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin 1 mg IM daily	609831;176763	IEM0214
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR					X			SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin, betaine	277410	IEM0215
Methylmalonic aciduria, cblDv2 type	*MMADHC*	AR				X		X		SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin, betaine	277410	IEM0215
Methionine synthase deficiency	*MTR*	AR						X		SAM (P,CSF), Amino acids (P), Organic acids (U)	Hydroxycobalamin 1 mg IM daily, betaine 250 mg/kg/day; folinic acid 5-30 mg/day	250940	IEM0217
Methylmalonic aciduria, cblA type	*MMAA*	AR				X		X		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Hydroxycobalamin injections	251100	IEM0218
Methylmalonic aciduria, cblB type	*MMAB*	AR				X		X		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	Low protein diet, carnitine, hydroxycobalamin, ammonia scavengers	251110	IEM0219
Sodium-dependent multivitamin transporter deficiency	*SLC5A6*	AR		X						ALP (P), IgG(S), 1,25-Dihydroxy vitamin D (S)		604024	IEM1124
Disorders of folate metabolism													
Proton-coupled folate transporter deficiency Hereditary folate malabsorption	*SLC46A1*	AR			X				Stomatitis	Folate (S), 5-Methyl-THF (CSF), Blood count (B), Immunoglobulins (S)		229050	IEM0221
5,10-methylenetetrahydrofolate reductase deficiency	*MTHFR*	AR					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 acid (100 mg/day).	236250	IEM0223
5,10-Methenyltetrahydrofolate synthetase deficiency	*MTHFS*	AR					X			5-Methyl-THF (CSF)		604197	IEM1035
Disorders of biotin metabolism													
Biotinidase deficiency	*BTD*	AR							Glossitis Stomatitis	Acyglycines (U), Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)	Biotin 5-10 mg/day	253260	IEM0227
Disorders of riboflavin metabolism													

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Mitochondrial flavin adenine dinucleotide transporter deficiency	*SLC25A32*	AR						X	Nausea	Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)		616839	IEM0237
Myopathic form of CoQ10 deficiency (ETFDH)	*ETFDH*	AR						X		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS), Acyglycines (U)		231675	IEM0240
Disorders of pyridoxine metabolism													
Pyridox(am)ine 5´- phosphate oxidase deficiency	*PNPO*	AR						X		B6 vitaminers (CSF, P)	Pyridoxal phosphate 30 mg/kg/day	610090	IEM0250
Disorders of vitamin D metabolism													
Vitamin D 24-hydroxylase deficiency	*CYP24A1*	AR						X		Calcium (U)		143880	IEM0269
Disorders of vitamin K metabolism													
Microsomal epoxide hydrolase deficiency	*EPHX1*	AR							Malabsorption	Bile acids (S)		607748	IEM0273
Disorders of molybdenum metabolism													
Cyclic pyranopterin monophosphate synthase deficiency	*MOCS1*	AR					X			Purines (U), Uric acid (U, P), Homocysteine, total (P), Sulfite (U), Pipecolic acid (CSF)	cPMP (fosdenopterin)	603707	IEM0275
Molybdopterin synthase deficiency	*MOCS2*	AR					X			Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)		603708	IEM0276
Gephyrin deficiency	*GPHN*	AR					X			Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)		603930	IEM0277
Disorders of copper metabolism													
Copper-transporting ATPase subunit beta deficiency Wilson disease	*ATP7B*	AR							Abdominal pain Drooling	ASAT/ALAT (P), Copper (S, U), Ceruloplasmin (S)	Penicillamine 750-1500 mg/day in adults, (10 mg/kg/day in children) divided in 2-3 doses together with 25 mg pyridoxine ; trientine 900-2500 mg/day in adults divided in 2-3 doses; zinc sulphate 600 mg/day divided in 3 doses in adults	277900	IEM0279
Copper-transporting ATPase subunit alpha deficiency Occipital horn syndrome	*ATP7A*	XL			X		X			Copper (S, U), Ceruloplasmin (S)	Copper chloride or L-histidine 350-500 ug/day IV or SC	304150	IEM0280
MEDNIK syndrome	*AP1S1*	AR			X					ASAT/ALAT (P), Copper (S, U), Ceruloplasmin (S), VLCFA (P)		609313	IEM0282
Disorders of iron metabolism													
Hereditary hemochromatosis (type 1)	*HFE*	AR							Abdominal pain	Iron (S), Ferritin (S), Transferin saturation (S)	Phlebotomy	235200	IEM0284
Transferrin receptor 2 deficiency	*TFR2*	AR							Abdominal pain	Iron (S), Ferritin (S), Transferrin (S)	Phlebotomy	604250	IEM0287
Disorders of zinc metabolism													
Acrodermatitis enteropathica	*SLC39A4*	AR			X					Zinc (S), ALP (P)	Zinc po 150-400 Zn sulfate/day (35-90 mg elemental Zn)	201100	IEM0297
Spondylocheirodysplastic Ehlers-Danlos syndrome	*SLC39A13*	AR							Bifid uvula	Lysyl pyridinoline (U), Hydroxylysyl pyridinoline (U)		612350	IEM0299
Disorders of magnesium metabolism													
Claudin 16 deficiency	*CLDN16*	AR					X		Abdominal pain	Magnesium (P,U), Calcium (P,U), Uric acid (P), Organic acids (U)	Magnesium replacement	248250	IEM0309
Sodium-chloride cotransporter deficiency	*SLC12A3*	AR							Abdominal pain	Calcium (P), Magnesium (S), Potassium (P)		263800	IEM0312
DISORDERS OF CARBOHYDRATES (n=17)													
Disorders of carbohydrate transport and absorption													
Glucose transporter 2 deficiency Fanconi-Bickel syndrome	*SLC2A2*	AR			X				Malabsorption	Chemistry (P,U), Oligosaccharides (U), Amino acids (P)	Corn starch, electrolyte replacement	227810	IEM0316

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Intestinal sodium-glucose cotransporter 1 deficiency	*SLC5A1*	AR			X					Glucose (U), Reducing sugars (stool)		606824	IEM0317
Sucrase-isomaltase deficiency	*SI*	AR			X				Malabsorption	DNA		222900	IEM0318
Trehalase deficiency	*TREH*	AR			X				Abdominal pain	DNA		612119	IEM0319
Lactase deficiency	*LCT*	AR			X					Reducing sugars (stool)		223000	IEM0320
Disorders of galactose metabolism													
Galactose-1-phosphate uridyltransferase deficiency Classic galactosemia	*GALT*	AR						X		Chemistry (P,U), Oligosaccharides (U), Amino acids (P)	Galactose restriction	230400	IEM0322
Galactose epimerase deficiency	*GALE*	AR						X		Chemistry (P,U), Oligosaccharides (U), Amino acids (P)		230350	IEM0323
Triokinase/FMN cyclase deficiency	*TKFC*	AR			X			X		Lactate (P)		618805	IEM1523
Disorders of fructose metabolism													
Aldolase B deficiency Hereditary fructose intolerance	*ALDOB*	AR			X			X	Abdominal pain Feeding habits, abnormal	Glycerol (U), Glucose (P), Coagulation factors (P)		229600	IEM0326
Disorders of the pentose phosphate pathway and polyol metabolism													
Transketolase deficiency	*TKT*	AR			X					Polyols (U, P, CSF), Sugar phosphates (U)	Investigational: thiamine, benfotiamine	617044	IEM0330
Disorders of insulin secretion and signaling													
Insulin promoter factor 1 deficiency	*PDX1*	AR				X				Glucose (P), C-Peptide (S), Glucagon (S)		606392;260370	IEM0344
RFX6 deficiency	*RFX6*	AD, AR			X				Malabsorption	Glucose (P), C-Peptide (S), Glucagon (S), Bilirubin (P), Insulin (P)		615710	IEM0351
Kabuki syndrome	*KMT2D;KDM6A*	Digenic			X		X		Malabsorption	DNA		300867	IEM1491
Beckwith-Wiedemann syndrome	*IGF2;H19;CDKN1C;KCNQ1*	Unknown							Macroglossia	DNA		130650	IEM1513
Glycogen storage diseases													
Glucose-6-phosphate transporter deficiency	*SLC37A4*	AR			X	X			Oral ulcerations	ASAT/ALAT (P), CK (P), Lactate (P), Glucose (P), Biotinidase (P)	Frequent meals, uncooked cornstarch, filgrastim	232220	IEM0355
Alpha-glucosidase deficiency Pompe disease	*GAA*	AR							Macroglossia	ASAT/ALAT (P), CK (P), Glycogen (M)	Alglucosidase alpha	232300	IEM0356
Disorders of gluconeogenesis													
Glucose-6-phosphatase deficiency von Gierke disease	*G6PC*	AR			X	X				ASAT/ALAT (P), Lactate (P), Glucose (S), Glycogen (L), Biotinidase (P)	Frequent meals, uncooked cornstarch	232200	IEM0370
MITOCHONDRIAL DISORDERS OF ENERGY METABOLISM (n=60)													
Disorders of the Krebs cycle													
ATP-specific succinyl-CoA ligase β subunit deficiency	*SUCLA2*	AR					X			Organic acids (U), Acylcarnitines (U, Lactate (P))		612073	IEM0399
GTP-specific succinyl-CoA ligase α subunit deficiency	*SUCLG1*	AR					X			Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), Lactate (P)		245400	IEM0400
Fumarate hydratase deficiency	*FH*	AR		X			X			Lactate (P), Organic acids (U), Catecholamines (P, U)	Alpha-adrenergic receptor blocker	606812	IEM0401
Mitochondrial malate dehydrogenase deficiency	*MDH2*	AR			X		X			Lactate (P), Organic acids (U), Catecholamines (P, U)	Alpha-adrenergic receptor blocker	617339	IEM0403

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Cytosolic NADP+-dependent isocitrate dehydrogenase 1 superactivity	*IDH1*	AR					X			Organic acids (U)		147700	IEM1090
Disorders of mitochondrial carriers													
Mitochondrial citrate carrier deficiency	*SLC25A1*	AR					X			Organic acids (U, P, CSF)		615182	IEM0411
Mitochondrial ATP-Mg-phosphate transporter deficiency	*SLC25A24*	AD					X			DNA		612289	IEM0412
Mitochondrial ATP-Mg-phosphate transporter deficiency	*SLC25A24*	AD		X						DNA		612289	IEM0412
Mitochondrial aspartate aminotransferase deficiency	*GOT2*	AR							Abdominal pain	Serine (CSF)		138150	IEM1138
Disorders of complex I subunits													
NADH dehydrogenase iron-sulfur protein 7 deficiency	*NDUFS7*	AR					X			Lactate (P)		618224	IEM0418
NADH dehydrogenase alpha subcomplex subunit 9 deficiency	*NDUFA9*	AR	X							Lactate (P)		256000	IEM0424
NADH dehydrogenase alpha subcomplex subunit 13 deficiency	*NDUFA13*	AR					X			Lactate (P)			IEM1141
Disorders of complex I assembly													
Complex I assembly factor 8 deficiency	*NDUFAF8*	AR	X							Lactate (P)		618776	IEM1530
Disorders of complex IV subunits													
Cytochrome c oxidase subunit 4I2 deficiency	*COX4I2*	AR				X				DNA		612714	IEM0465
Cytochrome c oxidase subunit 5A deficiency	*COX5A*	AR						X		DNA		603773	IEM1218
Disorders of complex IV assembly and ancillary proteins													
SCO1 deficiency	*SCO1*	AR					X			Lactate (P)		220110	IEM0473
SURF1 deficiency	*SURF1*	AR					X	X		Lactate (P)		256000;616684	IEM0475
LRPPRC deficiency	*LRPPRC*	AR					X			Lactate (P)		220111	IEM0476
PET100 deficiency	*PET100*	AR					X			Lactate (P)		220110	IEM0478
Disorders of complex V subunits													
Mitochondrial ATP synthase F0 subunit 6 deficiency	*MT-ATP6*	MT					X			Lactate (P)		516060	IEM0484
Disorders of complex V assembly													
Transmembrane protein 70 deficiency	*TMEM70*	AR			X				Dysmotility	CK (P), Lactate (P), Ammonia (P), 3-Methylglutaconic acid (U)		614052	IEM0486
Mitochondrial ATP synthase F1 assembly factor 2 deficiency	*ATPAF2*	AR					X			Lactate (P), Organic acids (U)		604273	IEM1151
Disorders of mitochondrial cytochrome synthesis and incorporation													
Mitochondrial cytochrome b deficiency	*MT-CYB*	MT			X				Dysmotility	Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)		516020	IEM0487
Disorders of single large-scale mtDNA deletions													

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Pearson Syndrome						X				Lactate (P, U, Lactate/Pyruvate ratio)		557000	IEM1501
Leigh Syndrome		Unknown						X		Lactate (P)		256000	IEM1505
Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication													
Mitochondrial DNA polymerase gamma catalytic subunit deficiency	*POLG*	AD			X			X	Dysmotility Pseudo-obstruction	Lactate (P), Organic acids (U), ASAT/ALAT (P)		613662	IEM0491
MPV17 deficiency	*MPV17*	AR			X				Dysmotility	Glucose (P), GGT (P), Activity respiratory chain complexes (I, III, IV, and V)		256810	IEM0494
TWINKLE mitochondrial DNA helicase deficiency	*TWNK*	AR						X		DNA		271245;616138	IEM0495
Mitochondrial ribonucelotide reductase subunit 2 deficiency	*RRM2B*	AD, AR			X				Dysmotility	Lactate (P), Amino acids (P)		604712	IEM0497
Thymidine phosphorylase deficiency	*TYMP*	AR		X	X			X	Malabsorption Abdominal pain Dysmotility	Purines and pyrimidines (U), Lactate (P)		131222;603041	IEM0498
Mitochondrial ribonuclease H1 deficiency	*RNASEH1*	AR	X							Lactate (P)		615156	IEM0500
FBXL4 deficiency	*FBXL4*	AR			X				Dysmotility	Lactate (P)		615471	IEM0502
Mitochondrial UMP-CMP kinase 2 deficiency	*CMPK2*	AR			X				Abdominal pain Dysmotility	DNA		611787	IEM1152
Disorders of mitochondrial transcription and RNA transcript processing													
Ribonuclease P 5' tRNA processing enzyme deficiency	*TRMT10C*	AR					X			Lactate (P)		616974	IEM0504
tRNA 5-carboxymethylaminomethyl transferase deficiency	*MTO1*	AR					X			Lactate (P)		614702	IEM0510
Pseudouridine synthase 1 deficiency	*PUS1*	AR						X	Nausea	Lactate (P)		600462	IEM0511
tRNA methyltransferase 5 deficiency	*TRMT5*	AR						X	Nausea	Lactate (P)		616539	IEM0513
tRNA 5-methylaminomethyl-2-thiouridylate-methyltransferase deficiency	*TRMU*	AR				X		X		Lactate (P)		613070	IEM0514
Mitochondrial ribosomopathies													
Mitochondrial ribosomal small subunit 34 deficiency	*MRPS34*	AR	X							Lactate (P)		617664	IEM0521
Disorders of mitochondrial tRNA													
Mitochondrial tRNA(Asp) deficiency	*MT-TD*	MT						X	Nausea	Lactate (P)		590015	IEM0533
Mitochondrial tRNA(Glu) deficiency	*MT-TE*	MT							Macroglossia	Lactate (P)		500009	IEM0535
Mitochondrial tRNA(Gly) deficiency	*MT-TG*	MT						X	Nausea	Lactate (P)		590035	IEM0537
Mitochondrial tRNA(Leu) 1 deficiency	*MT-TL1*	MT						X	Nausea	Lactate (P)		590050	IEM0540
Mitochondrial tRNA(Met) deficiency	*MT-TM*	MT						X	Nausea	Lactate (P)		590065	IEM0543
Mitochondrial tRNA(Phe) deficiency	*MT-TF*	MT						X		Lactate (P)		590070	IEM0544
Mitochondrial tRNA(Trp) deficiency	*MT-TW*	MT						X		Lactate (P)		590095	IEM0549
Disorders of mitochondrial tRNA incorporation and recycling													

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Mitochondrial tyrosyl-tRNA synthetase deficiency	*YARS2*	AR						X	Nausea	Lactate (P), Blood count		613561	IEM0564
Peptidyl-tRNA hydrolase 2 deficiency	*PTRH2*	AR				X				DNA		616263	IEM0571
Mitochondrial prolyl-tRNA synthetase deficiency	*PARS2*	AR					X			Lactate (P)		612036	IEM1078
Disorders of mitochondrial fusion													
OPA1 deficiency	*OPA1*	AD, AR							Dysmotility	Lactate (P)		125250	IEM0577
Disorders of mitochondrial phospholipid metabolism													
SERAC1 deficiency MEGDEL Syndrome	*SERAC1*	AR					X			Lactate (P), Organic acids (U), Filipin staining		614739	IEM0582
Tafazzin deficiency Barth syndrome	*TAZ*	XL					X	X	Nausea	Organic acids (U), Lipid panel (S), Urinalysis, 3-Methylglutaconic acid (U), 3-Methylglutaric acid (U), Carnitine, free (P), Blood count		302060	IEM0583
Disorders of mitochondrial protein import													
TIMM22 deficiency	*TIMM22*	AR		X			X			CK (P), Lactate (P)		607251	IEM1262
Disorders of mitochondrial protein quality control													
Mitochondrial intermediate peptidase deficiency	*MIPEP*	AR			X				Dysmotility	Lactate (P), Organic acids (U)		617228	IEM0593
CLPB deficiency	*CLPB*	AR							Oral ulcerations	DNA		616271	IEM0594
HSP60 deficiency (recessive)	*HSPD1*	AR					X			Lactate (P), Organic acids (U)		612233;605280	IEM0598
Sacsin deficiency	*SACS*	AR	X							DNA		270550	IEM0599
Other disorders of mitochondrial homeostasis													
CHCHD10 deficiency	*CHCHD10*	AD	X							CK (P)		615911;615048	IEM1081
Primary CoQ10 deficiencies													
Coenzyme Q7 hydroxylase deficiency	*COQ7*	AR					X			Lactate (P), Organic acids (U)		616733	IEM0623
Coenzyme 9 deficiency	*COQ9*	AR					X			Lactate (P), CoQ10 (M, P, WBC)		614654	IEM0626
DISORDERS OF LIPIDS (n=35)													
Disorders of fatty acid oxidation and transport													
Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	AR							Protein sensitivity	Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Dicarboxylic acids (U)		231530	IEM0635
Disorders of ketone body metabolism													
3-Hydroxy-3-methylglutaryl-CoA synthase deficiency	*HMGCS2*	AR			X			X		Acyglycines (U), Organic acids (U), Acylcarnitines (P), Fatty acids and ketones (P)		605911	IEM0641
Disorders of fatty acid synthesis and elongation													
3-Hydroxyacyl-CoA dehydratase 1 deficiency	*HACD1*	AR					X			DNA		610467	IEM1172
Disorders of cytoplasmic triglyceride metabolism													

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Diacylglycerol acyltransferase deficicncy	*DGAT1*	AR			X			X		ASAT/ALAT (P), Albumin (S), IgG(S)		615863	IEM0659
Disorders of non-mitochondrial phospholipid metabolism													
Phosphatidylserine synthase 1 superactivity	*PTDSS1*	AD			X					DNA		151050	IEM0667
Phosphatidylserine flippase deficiency	*ATP8A2*	AR					X			DNA		615268	IEM0668
Ethanolaminephosphotransferase 1 deficiency	*SELENOI*	AR							Bifid uvula	DNA		607915	IEM1174
Disorders of eicosanoid metabolism													
Prostaglandin transporter deficiency	*SLCO2A1*	AR		X					Peptic ulcer	DNA		259100;119900 ;259100	IEM0686
Cytosolic phospholipase A2α deficiency	*PLA2G4A*	AR		X	X		2		Abdominal pain Peptic ulcer	DNA		600522	IEM1072
Disorders of phosphoinositide metabolism													
Phosphatidylinositol 3,5-bisphosphate-5-phosphatase deficiency, neuroskeletal phenotype	*FIG4*	AR		X	X					DNA		216340	IEM0691
Phosphatidylinositol 4,5-bisphosphate-5-phosphatase deficiency Lowe syndrome	*OCRL*	XL			X					Amino acids (U), Cholesterol (S), Phosphate (U)		309000	IEM0692
Synaptojanin 1 deficiency	*SYNJ1*	AR					X			Lactate (P)		617389	IEM0693
Myotubularin 1 deficiency	*MTM1*	XL		X						DNA		310400	IEM0694
Phosphatidylinositol 3-kinase regulatory subunit 1 deficiency SHORT syndrome	*PIK3R1*	AD			X					IgG, IgA, IgM (S)		269880;616005	IEM0700
VAC14 deficiency	*VAC14*	AR	X				X		Drooling	DNA		617054	IEM1250
Phosphatidylinositol 4,5-bisphosphate phospholipase C γ2 deficiency	*PLCG2*	AD			X					IgA, IgM, IgG (S)		614878	IEM0709
Disorders of lipoprotein metabolism													
Microsomal triglyceride transfer protein deficiency	*MTTP*	AR							Malabsorption	LDL/HDL cholesterol (P), Apo B (P), Vitamins A/E (P)		200100;157147	IEM0723
Chylomicron retention disease Anderson disease	*SAR1B*	AR							Abdominal distension Malabsorption	Lipid panel (S), Apolipoprotein B (P)		246700	IEM0724
Lipoprotein lipase deficiency (LPL)	*LPL*	AR				X			Abdominal pain	LDL/HDL cholesterol (P), Triglycerides (P)		609708;238600	IEM0728
Apolipoprotein C-II deficiency (APOC2)	*APOC2*	AR				X			Abdominal pain	LDL/HDL cholesterol (P), Triglycerides (P)		608083;207750	IEM0729
GPIHBP1 deficiency	*GPIHBP1*	AR				X				Triglyceriders (S)		615947	IEM0730
Disorders of cholesterol biosynthesis													
Mevalonate kinase deficiency (mild)	*MVK*	AR			X				Malabsorption	Leucotriens (P), Organic acids (U)	Inflammatory control (non-steroidal anti-inflammatory drugs, corticosteroids, IL-1 targeting biologic agents, TNF-alpha blockade, allogenic stem cell transplantation)	260920	IEM0740
Sterol C14 reductase deficiency	*LBR*	AD, AR			X					Sterols (FB)		215140	IEM0745
Smith-Lemli-Opitz syndrome	*DHCR7*	AR		X	X		X		Dysmotility	ASAT/ALAT (P), Lipid panel (S), 7/8-Dehydrocholesterol (P)	Dietary supplementation of cholesterol 25-300 mg/kg/day, +/- bile acids	270400	IEM0753
Disorders of steroid metabolism													
11-beta-hydroxylase type 1 deficiency	*CYP11B1*	AR						X		Steroids (P), Potassium (P), Sodium (P)		202010	IEM0755

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
17-alpha-hydroxylase deficiency	*CYP17A1*	AR						X		Steroids (P), Potassium (P), Sodium (P)		202110	IEM0758
X-linked spinal and bulbar muscular atrophy Kennedy disease	*AR*	XL	X							Steroids (P)		313200	IEM0777
MIRAGE syndrome	*SAMD9*	AD			X					Steroids (P), Corticotropin (P)		617053	IEM1497
Disorders of bile acid synthesis													
3β-Hydroxy-Δ5-C27-steroid dehydrogenase-isomerase deficiency	*HSD3B7*	AR			X					Bile acids (P,U), Bilirubin, conjugated (P), Vitamins D/E		607764	IEM0779
Δ4-3-Oxosteroid-5β-reductase deficiency	*AKR1D1*	AR			X					Bile acids (P,U), Bilirubin, conjugated (P)		604741	IEM0780
Sterol 27-hydroxylase deficiency Cerebrotendinous Xanthomatosis	*CYP27A1*	AR			X					Lipid panel (S), Sterols (P), Cholestane pentol glucuronide (U), 25-Hydroxy-Vitamin D (P)	Chenodeoxycholic acid 750 mg/day (adults), HMG-CoA reductase inhibitors, low density lipoprotein apheresis	213700	IEM0782
Congenital bile acid synthesis defect ACOX2	*ACOX2*	AR			X					Bile acids (P,U)		617308	IEM0784
Bile acid-CoA:aminoacid N-acyl transferase deficiency	*BAAT*	AR			X					Bile acids (P,U), Bilirubin, conjugated (P), Vitamins A, D, E (S)		602938	IEM0785
SLC51A deficiency	*SLC51A*	AR			X					DNA		619484	IEM1681
SLC51B deficiency	*SLC51B*	AR			X					DNA		619481	IEM1682
STORAGE DISORDERS (n=24)													
Disorders of autophagy													
Spatacsin deficiency	*SPG11*	AR	X						Sphincter control problems	DNA		616668	IEM0814
TECPR2 deficiency	*TECPR2*	AR		X						DNA		615031	IEM0817
TBK1 deficiency	*TBK1*	AD	X							DNA		616439	IEM0818
SQSTM1 deficiency	*SQSTM1*	AD	X							DNA		616437	IEM1231
TBCK deficiency	*TBCK*	AR					X		Macroglossia	DNA		616899	IEM1236
ALS2 deficiency	*ALS2*	AR	X							DNA		606353	IEM1237
CHMP2B deficiency	*CHMP2B*	AD	X							DNA		614696	IEM1243
Sphingolipidoses													
Acid sphingomyelinase deficiency Niemann-Pick disease	*SMPD1*	AR					X			Enzyme activity (WBC)		257200;607616	IEM0834
Beta-galactosidase-1 deficiency Morquio syndrome type B	*GLB1*	AR							Gingival hypertrophy Macroglossia	Oligosaccharides (U), Lysosomal enzymes (DBS)	In trial gene therapy (NCT03952637)	230500	IEM0835
Beta-galactosylceramidase deficiency Krabbe disease	*GALC*	AR					X			DNA		245200	IEM0839
Krabbe disease-like disorder due to saposin A deficiency	*PSAP*	AR					X			Sulfatides (U), Protein (CSF), Lysosomal enzymes (DBS)		611722	IEM0840
Alpha-galactosidase A deficiency Fabry disease	*GLA*	XL							Abdominal pain Macroglossia	Globotriaosylsphingosine, Globotriaosylceramide, Proteins (U)	Enzyme replacement therapy	301500	IEM0844
Mucolipidoses													
UDP-N-acetylglucosamine-1-phosphotransferase subunit alpha/beta deficiency	*GNPTAB*	AR							Gingival hypertrophy	Oligosaccharide (U), Glycosaminoglycans, Enzyme activity (S)		252600	IEM0855

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Mucolipin 1 deficiency	*MCOLN1*	AR		X						Gastrin (S), Phospholipids (U)		252650	IEM0857
Mucopolysaccharidoses													
Alpha-iduronidase deficiency	*IDUA*	AR			X					Total GAGs (U), Dermatan sulfate (U), Heparan sulfate (U), Enzyme activity (WBC)	Hematopoietic cell transplantation (HCT), enzyme replacement therapy (laronidase)	607014;607015;607016	IEM0858
Iduronate 2-sulfatase deficiency Hunter disease	*IDS*	XL			X					Total GAGs (U), Dermatan sulfate (U), Heparan sulfate (U), Enzyme activity (WBC)	Enzyme replacement therapy (idursulfase)	309900	IEM0859
Heparan N-sulfatase deficiency Sanfilippo A disease	*SGSH*	AR			X					Mucopolysaccharides (U), Enzyme assay (DBS, L, F)	Clinical trial with intracerebroventricular infusion of chimeric fusion of recombinant enzyme +IGF2	252900	IEM0860
N-acetylglucosaminidase deficiency Sanfilippo B disease	*NAGLU*	AR			X					Mucopolysaccharides (U), Enzyme assay (DBS, S, F)	Clinical trial with intracerebral adenovirus associated viral vector containing human NAGLU cDNA	252920	IEM0861
Heparan-alpha-glucosaminide N-acetyltransferase deficiency	*HGSNAT*	AR			X					Mucopolysaccharides (U), Enzyme assay (DBS, L, F)		252930	IEM0862
N-acetylglucosamine 6-sulfatase deficiency Sanfilippo D disease	*GNS*	AR			X					Mucopolysaccharides (U), Enzyme assay (DBS, L, F)		252940	IEM0863
Mucopolysaccharidosis-plus syndrome	*VPS33A*	AR							Macroglossia	Mucopolysaccharides (U), Oligosaccharides (U)		617303	IEM0869
Disorders of lysosomal cholesterol metabolism													
Lysosomal acid lipase deficiency Wolman disease	*LIPA*	AR			X				Abdominal distension	Lipid panel (S), , Enzyme activity (S)	Enzyme replacement therapy	278000	IEM0872
Disorders of lysosomal protein degradation													
Cathepsin K deficiency	*CTSK*	AR							Periodontitis	DNA		265800	IEM0876
Cathepsin C deficiency	*CTSC*	AR							Periodontitis	DNA		245000	IEM0877
DISORDERS OF PEROXISOMES AND OXALATE (n=18)													
Disorders of plasmalogen synthesis													
Fatty Acyl-CoA reductase superactivity	*FAR1*	AD			X					Plasmalogens (RBC)		616107	IEM1515
Disorders of peroxisomal β-oxidation													
X-linked adrenoleukodystrophy and adrenomyeloneuropathy	*ABCD1*	XL							Sphincter control problems	VLCFA (P)	HCT at early stages of cerebral X-ALD; HSC gene therapy with lentiviral vector	300100	IEM0883
Peroxisomal straight-chain acyl-CoA oxidase deficiency	*ACOX1*	AR			X					VLCFA (P), Plasmalogens (P)		264470	IEM0884
D-bifunctional protein deficiency	*HSD17B4*	AR			X					VLCFA (P), Plasmalogens (P), Organic acids (U)		261515	IEM0885
Disorders of peroxisomal biogenesis													
Peroxin 1 deficiency Zellweger	*PEX1*	AR			X					VLCFA (P), Pipecolic acid (P)		234580;214100;601539	IEM0889
Peroxin 2 deficiency Zellweger	*PEX2*	AR			X					VLCFA (P), Pipecolic acid (P)		614866;614867	IEM0890
Peroxin 3 deficiency Zellweger	*PEX3*	AR			X					VLCFA (P), Pipecolic acid (P)		617370;614882	IEM0891
Peroxin 5 deficiency Zellweger	*PEX5*	AR			X					ASAT/ALAT (P), VLCFA (P), Pipecolic acid (P, U)		214110	IEM0892
Peroxin 6 deficiency Zellweger	*PEX6*	AR			X					VLCFA (P), Pipecolic acid (P)		614862;614863;616617	IEM0893
Peroxin 10 deficiency Zellweger	*PEX10*	AR			X					VLCFA (P), Pipecolic acid (P)		614870;614871	IEM0894

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Peroxin 14B deficiency Zellweger	*PEX11B*	AR			X					VLCFA (P), Pipecolic acid (P)		614920	IEM0895
Peroxin 12 deficiency Zellweger	*PEX12*	AR			X					VLCFA (P), Pipecolic acid (P)		614859;266510	IEM0896
Peroxin 13 deficiency Zellweger	*PEX13*	AR			X					VLCFA (P), Pipecolic acid (P)		614883;614885	IEM0897
Peroxin 14 deficiency Zellweger	*PEX14*	AR			X					VLCFA (P), Pipecolic acid (P)		614887	IEM0898
Peroxin 16 deficiency Zellweger	*PEX16*	AR			X					VLCFA (P), Pipecolic acid (P)		614876;614877	IEM0899
Peroxin 19 deficiency Zellweger	*PEX19*	AR			X					VLCFA (P), Pipecolic acid (P)		614886	IEM0900
Peroxin 26 deficiency Zellweger	*PEX26*	AR			X					VLCFA (P), Pipecolic acid (P)		614872;614873	IEM0901
Disorders of oxalate metabolism													
Hydroxyacid oxidase 1 deficiency	*HAO1*	AR	X							Oxalic acid (P,U), Glycolic acid (P,U)		605023	IEM0904
CONGENITAL DISORDERS OF GLYCOSYLATION (n=45)													
Disorders of N-linked glycosylation													
PMM2-CDG	*PMM2*	AR			X			X		ASAT/ALAT (P), Lipid panel (S), Sialotransferrins (S), Albumin (S), Factor XI (B),		601785	IEM0908
MPI-CDG	*MPI*	AR			X					Sialotransferins (S), Coagulation factors (P), Albumin (S)		602579	IEM0909
DPAGT1-CDG	*DPAGT1*	AR					X			Sialotrasferrins (S)		608093	IEM0910
ALG13-CDG	*ALG13*	XL					X			Sialotrasferrins (S)		300884	IEM0911
ALG13-CDG	*ALG13*	XL					X			Sialotrasferrins (S)		300884	IEM0912
ALG1-CDG	*ALG1*	AR			X		X			Sialotransferrins (S), IGG (P), B cells, circulating (blood)		608540	IEM0914
ALG11-CDG	*ALG11*	AR					X	X		Factor XI (B), Sialotrasferrins (S)		613661	IEM0916
RFT1-CDG	*RFT1*	AR					X			Sialotransferins (S), ASAT/ALAT (P), coagulation factors (P)		612015	IEM0917
ALG3-CDG	*ALG3*	AR					X			Antithrombin III (P), Protein S (S), Sialotrasferrins (S)		601110	IEM0918
ALG12-CDG	*ALG12*	AR		X	X		X		Dysmotility	ASAT/ALAT (P), Glucose (S), Lipid panel (S), Sialotransferrins (S), IGF BP3, .		607143	IEM0920
ALG8-CDG	*ALG8*	AD, AR			X		X			CK (P), Sialotransferrins (S), Albumin (S), Factor IX and XII (B), Antithrombin III (P)		608104	IEM0922
STT3A-CDG	*STT3A*	AR			X				Dysmotility	Sialotrasferrins (S)		615596;601134	IEM0924
GCS1-CDG	*MOGS*	AR		X						ASAT/ALAT (P), Oligosaccharide (U), Sialotransferins (S)		606056	IEM0927
MGAT2-CDG	*MGAT2*	AR		X	X		X			ASAT/ALAT (P), CK (P), Sialotransferrins (S), Factor IX and XII (B), Antithrombin III (P)		212066	IEM0931
FUT8-CDG	*FUT8*	AR					X			Glucose (P), Blood count		618005	IEM0932
STT3B-CDG	*STT3B*	AR			X				Dysmotility	Sialotrasferrins (S)		615597	IEM1193
DDOST-CDG	*DDOST*	AR		X	X					Factor XI (B), Proteins C/S (S), Sialotrasferrins (S)		614507;602202	IEM1192

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
B4GALT1-CDG	*B4GALT1*	AR			X					Antithrombin (B), Factor XI (B), Sialotrasferrins (S)		607091	IEM1194
GFUS-CDG	*GFUS*	AR					X			DNA		137020	IEM1516
SSR3-CDG	*SSR3*	AR						X		DNA		606213	IEM1548
MAN2B2-CDG	*MAN2B2*	AR			X					Sialotransferrins (S), C-reaktive protein, CRP (P), IgE (S)		618899	IEM1549
Disorders of O-mannosylation													
FKRP-CDG B	*FKRP*	AR					X			CK (P), DNA		606612	IEM0941
Disorders of O-GalNAcylation													
GALTNT3-CDG	*GALNT3*	AR							Visceral calcifications	Phosphate (P)		211900	IEM0963
Disorders of O-fucosylation													
B3GALTL-CDG	*B3GLCT*	AR		X	X					DNA		261540	IEM0970
Disorders of glycosylphosphatidylinositol biosynthesis													
PIGA-CDG	*PIGA*	XL					X			ALP (P), GPI-anchored proteins (WBC, F)		300868;300818	IEM0971
PIGV-CDG	*PIGV*	AR			X		X			ALP (P), GPI-anchored proteins (WBC, F)		239300	IEM0978
Disorders of dolichol metabolism													
DPM1-CDG	*DPM1*	AR					X			Factor XI (B), Sialotrasferrins (S)		608799	IEM0995
MPDU1-CDG	*MPDU1*	AR					X			Sialotrasferrins (S)		609180	IEM0998
Disorders of monosaccharide synthesis and interconversion													
PGM1-CDG	*PGM1*	AR							Bifid uvula	ASAT/ALAT (P), CK (P), Ammonia (P), Sialotransferrins (S)		614921	IEM1003
Ubiquitous glucose-6-phosphatase deficiency Dursun syndrome	*G6PC3*	AR			X					Blood count, DNA		612541	IEM1005
Disorders of nucleotide-sugar synthesis													
GMPPA-CDG	*GMPPA*	AR	X	X						DNA		615510	IEM1006
UGDH-CDG	*UGDH*	AR					X			DNA		618792	IEM1556
Disorders of Golgi transport													
SLC35A2-CDG	*SLC35A2*	XL					X			Sialotrasferrins (S)		314375	IEM1009
SLC35C1-CDG	*SLC35C1*	AR							Periodontitis	Neutrophil motility/rolling (B)		266265	IEM1011
SLC35D1-CDG	*SLC35D1*	AR							Abdominal distension	Sialotransferrins (S)		269250	IEM1012
Disorders of vesicular trafficking													
Conserved oligomeric Golgi complex subunit 1 deficiency	*COG1*	AR					X			Sialotransferrins (S)		611209	IEM1013

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Conserved oligomeric Golgi complex subunit 7 deficiency	*COG7*	AR					X			ASAT/ALAT (P), CK (P), Glucose (S), Total/direct bilirubin (S), Sialotransferins (S)		608779	IEM1017
Craniolenticulosutural dysplasia	*SEC23A*	AR							Bifid uvula	DNA		607812	IEM1406
TRAPPC12 deficiency	*TRAPPC12*	AR	X							DNA		617669	IEM1412
NBAS deficiency	*NBAS*	AR						X		ASAT/ALAT (P), DNA		608025	IEM1415
AP3B2 deficiency	*AP3B2*	AR					X			DNA		617276	IEM1425
AP4E1deficiency	*AP4E1*	AR							Drooling	DNA		607244	IEM1235
YIF1B deficiency	*YIF1B*	AR	X							DNA		619125	IEM1517
Disorders of Golgi homeostasis													
ATP6AP1-CDG	*ATP6AP1*	XL				X				Sialotransferrins (S), Ceruloplasmin (S), Copper (S), IgG (S)		300972	IEM1027
Disorder of deglycosylation													
N-glycanase 1 deficiency	*NGLY1*	AR					X			Oligosaccharides (U)		615273	IEM1032
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=26)													
Disorders of nucleotide metabolism													
3' Repair exonuclease 1 deficiency Aicardi-Goutières syndrome type 1	*TREX1*	AR		X			X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 lysophosphatidylcholine	JAK inhibitors (baricitinib, ruxolitinib)	225750	IEM0026
Ribonuclease H2 subunit B deficiency Aicardi-Goutières syndrome type 2	*RNASEH2B*	AR					X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	610181	IEM0027
Ribonuclease H2 subunit C deficiency Aicardi-Goutières syndrome type 3	*RNASEH2C*	AR					X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	610329	IEM0028
Ribonuclease H2 subunit A deficiency Aicardi-Goutières syndrome type 4	*RNASEH2A*	AR					X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	610333	IEM0029
SAMHD1 deficiency	*SAMHD1*	AR					X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	612952	IEM0031
RNA-specific adenosine deaminase deficiency Aicardi-Goutières syndrome type 6	*ADAR*	AR					X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF)	JAK inhibitors	615010	IEM0032
MDA5 superactivity Aicardi-Goutières syndrome type 7	*IFIH1*	AD					X			ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	JAK inhibitors	615846	IEM0033
ABCC6 deficiency	*ABCC6*	AR		X						Pyrophosphate (S), Lactate (P), DNA		264800	IEM0036
RNA-specific adenosine deaminase 2 deficiency	*ADARB1*	AR					X			DNA		618862	IEM1278
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases													
Elongator complex protein 1 deficiency	*ELP1*	AR		X	X			X	Dysmotility	DNA		223900	IEM1291
Glutamyl-prolyl-tRNA synthetase 1 deficiency	*EPRS1*	AR	X							DNA		617951	IEM1307
Phenylalanyl-tRNA synthetase subunit alpha deficiency	*FARSA*	AR					X			DNA		619013	IEM1313
Valyl-tRNA synthetase 1 deficiency	*VARS1*	AR					X			DNA		617802	IEM1319
Disorders of ribosomal biogenesis													

Name (n=339)	Gene	Inheritance	Esophagus	Stomach	Intestines	Pancreas	Feeding difficulties	Vomiting	Other	Laboratory investigations	Speciific treatment	Disorder OMIM No.	IEMbase (hyperlinked)
Childhood-onset motor and cognitive regression syndrome with extrapyramidal movement disorder	*UBTF*	AD	X							DNA		617672	IEM1333
Diamond-Blackfan anemia type 19	*RPL35*	AD			X					Reticulocytes (B)		618312	IEM1360
Shwachman-Diamond syndrome type 1	*SBDS*	AR				X				DNA		260400	IEM1369
Shwachman-Diamond syndrome type 2	*EFL1*	AR				X				DNA		617941	IEM1370
Shwachman-Diamond syndrome, EIF6 type	*EIF6*	AR			X	X				DNA		602912	IEM1372
POLR3K deficiency	*POLR3K*	AR					X			DNA		619310	IEM1658
Disorders of heme metabolism													
Delta-aminolevulinate dehydratase deficiency	*ALAD*	AR			X			X	Abdominal pain Nausea	Porphyrines (U), Delta-ALA (U)		125270	IEM0788
Porphobilinogen deaminase deficiency	*HMBS*	AD, AR			X			X	Abdominal pain Nausea	Porphyrins (U), Porphobilinogen (U), Delta-ALA (U), DNA	Intravenous hemin (haem arginate or haematin) 3-4 mg/kg OD x 4days; carbohydrate infusion Dextrose 10%; liver transplantation	176000	IEM0789
Coproporphyrinogen oxidase deficiency Hereditary coproporphyria	*CPOX*	AD			X			X	Abdominal pain Nausea	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	121300	IEM0792
Protoporphyrinogen oxidase deficiency	*PPOX*	AD			X			X	Abdominal pain Nausea	PBG (U), Porphyrins (U, stools), Fluorescence scanning (P)	Intravenous hemin (haem arginate or haematin) 3-4 mg/kg OD x 4days; carbohydrate infusion Dextrose 10%; liver transplantation	176200	IEM0794
Disorders of bilirubin metabolism and biliary transport													
ATP8B1 deficiency Byler Disease	*ATP8B1*	AR			X	X				Bile acids (P,U), Chloride (sweat)		211600	IEM0805
Apical bile salt transporter deficiency	*SLC10A2*	AR			X					LDL cholesterol (P)		613291	IEM0810
OTHER (n=9)													
Disorders of choline neurotransmission													
Choline transporter deficiency	*SLC5A7*	AR	X				X			DNA		617143	IEM1449
Choline acetyltransferase deficiency	*CHAT*	AR					X			DNA		254210	IEM1450
Disorders of the synaptic vesicle cycle													
Synaptic vesicle glycoprotein 2A deficiency	*SV2A*			X						N-acetylaspartate (MRS), Lactate (MRS)		185860	IEM1466
Synaptobrevin 1 deficiency	*VAMP1*	AD, AR					X			DNA		618323	IEM1467
Torsin 1A deficiency	*TOR1A*	AR					X			DNA		618947	IEM1476
Clathrin heavy chain deficiency	*CLTC*	AD	X	X						DNA		617854	IEM1479
Disorders of organelle interplay													
Hermansky-Pudlak syndrome type 5	*HPS5*	AR			X					DNA		614074	IEM1387