Table S2: Disorders with individual families described or poorly characterized (enzyme not known, no enzymatic or molecular confirmation).

Group/Name of IMD	Alternative names	Inheritance	Gene	OMIM gene number	Reference
2. Disorders of purine metabolism					
AICAR transformylase/IMP cyclohydrolase deficiency	AICA-ribosiduria	AR	*ATIC*	601731	1
Adenosine deaminase superactivity		AD	*?*	?	2–4
Adenylate kinase 7 deficiency	Primary male infertility with multiple morphological anomalies of the flagella	AR	*AK7*	615364	5
3. Disorders of nucleotide metabolism					
Ecto-5'-nucleotidase superactivity	Nucleotidase-associated pervasive developmental disorder	?	*? NT5E*	? 129190	6–8
6. Disorders of glutathione metabolism					
γ-glutamyl transpeptidase deficiency	Glutathioninuria; γ-glutamyl transferase deficiency	AR	? GGT1	? 612346	9–13
Dipeptidase deficiency		AR	?	?	14
8. Disorders of amino acid transport					
Lysine malabsorption syndrome		?	?	?	15
Dibasic aminoaciduria type 1		AD	?	?	16
Methionine malabsorption syndrome	Oasthouse disease; Smith-Strang disease; methioninuria	?	?	?	17–20
Cationic amino acid transporter 2 deficiency		AR	*SLC7A2*	601872	21
13. Disorders of sulfur amino acid and sulfide metabolism					
Methionine adenosyltransferase II deficiency		AD	*MAT2A*	601468	22
14. Disorders of branched-chain amino acid metabolism					
Branched-chain ketoacid dehydrogenase phosphatase deficiency		AR	*PPM1K*	611065	23

3-hydroxyisobutyrate dehydrogenase deficiency		AR	*HIBADH*	608475	24
15. Disorders of lysine metabolism					
Diaminopentanuria	Cystine-lysinuria	?	?	?	25,26
Hydroxylysinuria		AR	*? AGPHD1*	? 614681	27–29
17. Disorders of β- and γ-amino acids					
GABA type A receptor α6 subunit deficiency		AD	*GABRA6*	137143	30,31
GABA type A receptor δ subunit deficiency		AR	*GABRD*	137163	32
Serum carnosinase deficiency	Carnosinemia; homocarnosinosis	AR	? *CNDP1*	? 609064	33–38
18. Disorders of histidine metabolism					
Urocanase deficiency	Urocanic aciduria	AR	*UROC1*	613012	39–41
Histidinuria		AR	?	?	42–45
19. Disorders of tryptophan metabolism					
Kynurenine-3-hydroxylase deficiency		?	*KMO*	603538	46
Aminocarboxymuconate semialdehyde decarboxylase superactivity	Picolinate carboxylase superactivity	AR	*ACMSD*	608889	47
Blue diaper syndrome		?	?	?	48
20. Disorders of glutamate metabolism					
Glutamate decarboxylase 1 deficiency	Spastic cerebral palsy type 1	AR	*GAD1*	605363	49
21. Disorders of glutamine metabolism					
Glutaminase 1 superactivity		AD	*GLS1*	138280	50
24. Disorders of glycine metabolism					
Glycine encephalopathy due to H protein deficiency		AR	*GCSH*	238330	51,52
25. Disorders of lipoic acid and iron-sulfur metabolism					
Ferredoxin 2 deficiency		AR	*FDXL1*	614585	53
ISD11 deficiency	Combined oxidative phosphorylation deficiency 19	AR	*LYRM4*	613311	54
NFS1 deficiency	Infantile mitochondrial complex II/III deficiency (IMC23D)	AR	*NFS1*	603485	55

26. Disorders of cobalamin metabolism					
Methylmalonic aciduria and homocystinuria due to ZNF143 deficiency		AR	*ZNF143*	603433	56
Methylmalonic aciduria and homocystinuria due to Ronin deficiency		AR	*THAP11*	609119	57
28. Disorders of biotin metabolism					
Sodium-dependent multivitamin transporter deficiency		AR	*SLC5A6*	604024	58
32. Disorders of pantothenate metabolism					
Mitochondrial coenzyme A transporter deficiency		AR	*SLC25A42*	610823	59
33. Disorders of pyridoxine metabolism					
Intestinal alkaline phosphatase anchoring deficiency		AD	*ALPI*	171740	60
35. Disorders of vitamin A metabolism					
Retinol dehydrogenase 11 deficiency	Retinal dystrophy, juvenile cataracts, and short stature syndrome	AR	*RDH11*	607849	61
41. Disorders of iron metabolism					
Ferritin heavy chain dysregulation	Hereditary hemochromatosis type 5	AD	*FTH1*	134770	62
43. Disorders of zinc metabolism					
Asymptomatic familial hyperzincemia		AD	*?*	?	63,64
Hyperzincemia with hypercalprotectinemia		?	*?*	?	65–70
45. Disorders of magnesium metabolism					
Epidermal growth factor deficiency	Isolated autosomal recessive hypomagnesemia; renal hypomagnesemia type 4	AR	*EGF*	131530	71
KCNA1 deficiency		AD	*KCNA1*	176260	72,73
46. Disorders of carbohydrate transport and absorption					
MAP17 deficiency	Familial renal glucosuria type 2	AR	*PDZK1IP1*	607178	74

49. Disorders of the pentose phosphate pathway and polyol metabolism					
L-arabinosuria		?	*?*	?	75
Sorbitol dehydrogenase deficiency		AR	*SORD*	182500	76,77
50. Disorders of insulin secretion and signaling					
AKT2 deficiency		AD	*AKT2*	164731	78
51. Glycogen storage diseases					
HOIL1 interacting protein deficiency		AR	*RNF31*	612487	79
52. Disorders of gluconeogenesis					
Mitochondrial phosphoenolpyruvate carboxykinase deficiency		AR	*PCK2*	614095	80,81
55. Disorders of the Krebs cycle					
α-ketoglutarate dehydrogenase deficiency		AR	*OGDH*	613022	82–86
Mitochondrial malate dehydrogenase deficiency, tumoral phenotype	Familial paraganglioma	AR	*MDH2*	154100	87
57. Disorders of mitochondrial carriers					
Mitochondrial aspartate aminotransferase deficiency		AR	*GOT2*	138150	88
Mitochondrial dicarboxylate transporter deficiency		AR	*SLC25A10*	606794	89
58. Disorders of complex I subunits					
NADH dehydrogenase α subcomplex subunit 11 deficiency		AR	*NDUFA11*	612638	90
NADH dehydrogenase α subcomplex subunit 13 deficiency		AR	*NDUFA13*	609435	91
NADH dehydrogenase β subcomplex subunit 9 deficiency		AR	*NDUFB9*	601445	92
62. Disorders of complex III subunits					
UQCRQ deficiency		AR	*UQCRQ*	612080	93
63. Disorders of complex III assembly					
UQCC3 deficiency		AR	*UQCC3*	616097	94
64. Disorders of complex IV subunits					
Cytochrome c oxidase subunit 8A deficiency		AR	*COX8A*	123870	95
65. Disorders of complex IV assembly and ancillary proteins					

Cytochrome c oxidase assembly factor 3 deficiency		AR	*COA3*	614775	96
Cytochrome c oxidase assembly factor 5 deficiency		AR	*COA5*	613920	97
Cytochrome c oxidase assembly factor 7 deficiency		AR	*COA7*	615623	98
Cytochrome c oxidase assembly factor 14 deficiency		AR	*COX14*	614478	99
NADH dehydrogenase α subcomplex subunit 4 deficiency		AR	*NDUFA4*	603833	100
CEP89 deficiency		AR	*CEP89*	615470	101
67. Disorders of complex V assembly					
Mitochondrial ATP synthase F1 assembly factor 2 deficiency		AR	*ATPAF2*	608918	102
69. Disorders of mitochondrial DNA depletion, multiple deletion, or intergenomic communication					
Mitochondrial UMP-CMP kinase 2 deficiency		AR	*CMPK2*	611787	
70. Disorders of mitochondrial transcription and RNA transcript processing					
Mitochondrial transcription factor A deficiency		AR	*TFAM*	600438	103
Mitochondrial poly(A) exoribonuclease deficiency		AR	*PDE12*	616519	104
71. Mitochondrial ribosomopathies					
Mitochondrial ribosomal large subunit 12 deficiency		AR	*MRPL12*	602375	105
Mitochondrial ribosomal small subunit 7 deficiency		AR	*MRPS7*	611974	106
Mitochondrial ribosomal small subunit 23 deficiency		AR	*MRPS23*	611985	107
74. Disorders of mitochondrial tRNA incorporation and recycling					
Mitochondrial threonyl-tRNA synthetase deficiency	Combined oxidative phosphorylation deficiency type 21	AR	*TARS2*	612805	108,109
Mitochondrial glutamyl-tRNA(Gln) amidotransferase subunit A deficiency		AR	*QRSL1*	617209	107

77. Disorders of mitochondrial phospholipid metabolism					
PNPLA4 deficiency		XLR	*PNPLA4*	300102	107
79. Disorders of mitochondrial protein quality control					
Pitrilysin metallopeptidase 1 deficiency		AR	*PITRM1*	-	110
YME1L1 deficiency	Optic atrophy type 11	AR	*YME1l1*	607472	111
80. Other disorders of mitochondrial homeostasis					
Mitochondrial thioredoxin 2 deficiency	Combined oxidative phosphorylation deficiency type 29	AR	*TXN2*	609063	112
Mitochondrial thioredoxin reductase 2 deficiency	Selenoprotein Z deficiency; glucocorticoid deficiency type 5	AR	*TXNRD2*	606448	113
81. Primary CoQ10 deficiencies					
COQ5 deficiency		AR	*COQ5*	616359	114
82. Disorders of carnitine metabolism					
Carnitine palmitoyltransferase 1C deficiency	Autosomal dominant spastic paraplegia type 73	AR	*CPT1C*	608846	115
γ-butyrobetaine hydroxylase deficiency		AR	*BBOX1*	603312	116
Carnitine acetyltransferase deficiency		AR	*CRAT*	600184	117,118
83. Disorders of fatty acid oxidation and transport					
Long-chain acyl-CoA dehydrogenase deficiency		AR	*ACADL*	609576	119
Medium-chain 3-ketoacyl-CoA thiolase (MCKAT) deficiency		?	*?*	?	120
Isolated mitochondrial long-chain ketoacyl-CoA thiolase deficiency		AR	*HADHB*	143450	121
Long-chain fatty acid plasma membrane transporter deficiency		?	*?*	?	122
84. Disorders of ketone body metabolism					
Cytosolic acetoacetyl-CoA thiolase deficiency		?	*ACAT2*	100678	123,124
85. Disorders of fatty acid synthesis and elongation					

Cytosolic acetyl-CoA carboxylase 1 deficiency		?	*ACACA*	200350	125
Mitochondrial acetyl-CoA carboxylase 2 deficiency		AR	*ACACB*	601557	88
3-Hydroxyacyl-CoA dehydratase 1 deficiency		AR	*HACD1*	610467	126
Trans-2-enoyl-CoA reductase deficiency	Autosomal recessive mental retardation type 14	AR	*TECR*	610057	127
90. Disorders of non-lysosomal sphingolipid metabolism					
Ceramide synthase 1 deficiency	Progressive myoclonic epilepsy type 8	AR	*CERS1*	606919	128,129
Ceramide synthase 2 deficiency		AD	*CERS2*	606920	130
Alkaline ceramidase 3 deficiency	Early childhood-onset progressive leukodystrophy	AR	*ACER3*	617036	131
91. Disorders of eicosanoid metabolism					
Leukotriene C4 synthase deficiency		AR	*?LTC4S*	246530	132,133
92. Disorders of palmitoylation					
ZDHHC15 palmitoyltransferase deficiency	X-linked mental retardation type 91	XLD	*ZDHHC15*	300576	134
Hedgehog acyltransferase deficiency		AR	*HHAT*	605743	135
93. Disorders of phosphoinositide metabolism					
Phosphatidylinositol 4,5-bisphosphate 3-kinase regulatory subunit deficiency	Ataxia-oculomotor apraxia type 3	AD	*PIK3R5*	611317	136
Phosphatidylinositol 4,5-bisphosphate phospholipase C β3 deficiency	Spondylometaphyseal dysplasia with corneal dystrophy and developmental delay	AR	*PLCB3*	600230	137
Inositol 1,4,5-triphosphate receptor type 2 deficiency	Isolated anhidrosis with normal sweat glands	AR	*ITPR2*	600144	138
95. Disorders of cholesterol biosynthesis					
Geranylgeranyl pyrophosphate synthase deficiency	Atypical femoral fractures with bisphosphonates	AD	*GGPS1*	606982	139
97. Disorders of bile acid synthesis					
Cholesterol 7α-hydroxylase deficiency		AR	*CYP7A1*	118455	140
PMP70 deficiency	Congenital bile acid synthesis defect type 5	AR	*ABCD3*	170995	141

Bile acid CoA ligase deficiency		AR	*SLC27A5*	603314	142
100. Disorders of autophagy					
ATG5 deficiency	Autosomal recessive spinocerebellar ataxia type 25	AR	*ATG5*	604261	143
115. Disorders of N-linked glycosylation					
Oligosaccharyltransferase DDOST subunit deficiency	DDOST-CDG	AR	*DDOST*	602202	144
Oligosaccharyltransferase STT3B subunit deficiency	STT3B-CDG	AR	*STT3B*	608605	145
β-1,4-galactosyltransferase deficiency	B4GALT1-CDG	AR	*B4GALT1*	137060	146
117. Disorders of O-xylosylation and glycosaminoglycan synthesis					
Chondroitin sulfate N-acetylgalactosaminyltransferase 1 deficiency		AR	*CSGALNACT1*	616615	147
122. Disorders of glycosylphosphatidylinositol biosynthesis					
PIGP-CDG	GPI biosynthesis defect type 14; early infantile epileptic encephalopathy type 55	AR	*PIGP*	605938	148
PIGY-CDG	Hyperphosphatasia with mental retardation type 6; GPI biosynthesis defect type 12	AR	*PIGY*	610662	149
128. Disorders of vesicular trafficking					
Conserved oligomeric Golgi complex subunit 2 deficiency	COG2-CDG	AR	*COG2*	606974	150
Disorder of ADP-ribosylation					
Glutamyl ribose 5-phosphate storage disease		? XLR	?	?	151–154

Abbreviations: AD, autosomal dominant; AR, autosomal recessive; XL, X-linked; XLD, X-linked dominant; XLR, X-linked recessive

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