

	OMIM #	FULL NAME
A. disorders of amino acid metabolism		
1	261600	classical phenylketonuria and hyperphenylalaninemia
2	261640	phenylketonuria due to PTPS deficiency
3	261630	phenylketonuria due to DHPR deficiency
4	264070	phenylketonuria due to PCD deficiency
5	128230	DOPA-responsive dystonia (TH, SPR, GCH1)
6	248600	leucinose, maple syrup urine disease (MSUD)
7	276700	tyrosinemia type 1
8	276600	tyrosinemia type 2
9	276710	tyrosinemia type 3
10	203500	alkaptonuria
11	236200	homocystinuria, B6 responsive and non responsive
12	236250	homocystinuria due to MTHFR deficiency
13	236270-250940	homocystinuria-megaloblastic anemia Cbl E & G type
14	250850	methionine S-adenosyltransferase deficiency
15	606664	glycine N-methyltransferase deficiency
16	180960	S-adenosylhomocystine hydrolase deficiency
17	237300	hyperammonemia due to CPS deficiency
18	311250	hyperammonemia due to OTC deficiency
19	215700	citrullinemia type I
20	605814-603471	citrullinemia type II
21	207900	argininosuccinic aciduria (ASL deficiency)
22	207800	argininemia (arginase deficiency)
23	237310	hyperammonemia due to NAGS deficiency
24	238970	hyperornithinemia, hyperammonemia, homocitrullinuria (HHH)
25	222700	lysinuric protein intolerance
26	258870	gyrate atrophy, B6 responsive or non responsive
27	238700	hyperlysinemia (alpha-amino adipic semialdehyde synthase deficiency)
28	238300-330-310	non ketotic hyperglycinaemia
29	234500	hartnup disorder
30	601815-172480	Disorders of serine metabolism
31	239500-239510-237000-612652-179035	Disorders of proline/hydroxyproline/pyrroline 5-carboxylate metabolism
32	606407	hypotonia-cystinuria
33	309000	Lowe oculocerebral syndrome
B. organic acidemias		
34	251100-110 277400-410-380	methylmalonic aciduria (CblA,B,C,D,F)

35	251000	methylmalonic aciduria mutase deficiency type
36	275350	transcobalamin 2 deficiency
37	232000	propionic acidemia
38	243500	isovaleric aciduria
39	210200	3-methylcrotonylglycinuria
40	250950-302060-258501-610198	3 methylglutaconic aciduria type 1, 2, 3, 4, 5
41	246450	HMG CoA lyase deficiency
42	231670	glutaric aciduria type 1
43	305950	glutaric aciduria type 2 (MADD)
44	600721-236792	L-2 and D-2-OH glutaric aciduria
45	271980	4-hydroxybutyric aciduria (SSADH deficiency)
46	611283	isobutyryl CoA dehydrogenase deficiency (ACAD8)
47	600301	short/branched chain acylCoA dehydrogenase deficiency (SBCADD)
48	248360	malonic aciduria
C. biotin-responsive disorders		
49	253270	holocarboxylase synthase deficiency
50	253260	biotinidase deficiency
51	607483	biotin responsive basal ganglia disease
D. disorders of carbohydrate metabolism and glycogen storage diseases		
52	222900	sucrase isomaltase deficiency
53	606824	congenital glucose/galactose malabsorption
54	229600	hereditary fructose intolerance
55	230400	galactosemia (uridylyltransferase deficiency)
56	230350	galactosemia (epimerase deficiency)
57	230200	galactosemia (galactokinase deficiency)
58	606003	transaldolase deficiency
59	266150	pyruvate carboxylase deficiency
60	261680	phosphoenolpyruvate carboxykinase (PEPCK) deficiency
61	229700	fructose 1,6-bisphosphatase deficiency
62	240600	glycogenose 0 (glycogen synthase deficiency)
63	232200	glycogenose Ia and Ib (G6Pase)
64	232400	glycogenose 3 Cori
65	232500	glycogenose 4 Andersen
66	232600	glycogenose 5 Mc Ardle
67	232700-750-740	glycogenose 6 Hers
68	232800	glycogenose 7 Tarui
69	306000-261750-613027-300559	glycogenose IX phosphorylase kinase
70	261670	glycogenosis X (phosphoglycerate mutase deficiency)

71	300653	phosphoglycerate kinase deficiency
72	612933	lactate dehydrogenase deficiency (type XI)
73	611881	aldolase A deficiency (type XII)
74	612932	β -enolase deficiency (type XIII)
75	606777	glucose transporter defect De Vivo syndrome (GLUT-1)
76	227810	fanconi-bickel syndrome (GLUT-2)
E. hyperinsulinism		
77	606762	hyperinsulinism-hyperammonemia syndrome (HiHa)
78	256450-602485-609968 and other	familial hyperinsulinemic hypoglycemia, types 1-7
F. disorders of mitochondrial energy metabolism		
79	312170	pyruvate dehydrogenase deficiency
80	252010	mitochondrial complex I deficiency
81	252011	mitochondrial complex II deficiency
82	124000	mitochondrial complex III deficiency
83	220110	mitochondrial complex IV deficiency
84	604273	mitochondrial complex V deficiency
85	540000-551500-545000 and other	mitochondrial DNA mutation/deletion/elongation
86	603041-174763 and other	nuclear DNA mutation affecting mitochondrial structure and/or function
87	251880 and other	mitochondrial DNA depletion syndrome
88	530000-557000-256000-203700-545000 and other	clinical syndrome associated with mitochondrial disorder (report+board advise)
89	606812 and other	krebs cycle enzyme deficiencies
90	607426	coQ10 synthesis defects
G. mitochondrial beta oxidation defects		
91	212140	camitine transporter deficiency
92	255120	camitine palmitoyltransferase 1 deficiency
93	212138	camitine translocase
94	255110	camitine palmitoyltransferase 2 deficiency
95	201470	short chain acyl coa dehydrogenase deficiency (SCADD)
96	601609	short chain 3 oh acyl coa dehydrogenase deficiency (SCHADD)
97	201450	medium chain acyl coa dehydrogenase deficiency (MCADD)
98	201460	long chain acyl coa dehydrogenase deficiency (LCADD)
99	143450	long chain 3 oh acyl coa dehydrogenase deficiency (LCHADD)
100	201475	very long chain acyl coa dehydrogenase deficiency (VLCADD)
101	600890	mitochondrial trifunctional protein

H. disorders of ketone body metabolism		
102	605911	3-hydroxy-3-methylglutaryl-coA synthase deficiency
103	246450	3-hydroxy-3methylglutaryl-coA lyase deficiency
104	245050	succinyl-coA 3-oxoacid coA transferase deficiency (scot)
105	203750	β-ketothiolase deficiency
I. disorders of sterol, bile acid, lipid and lipoprotein metabolism		
106	270400	smith-lemli-opitz syndrome
107	251170	mevalonic aciduria
108	607765	3β-hydroxy δ5 c27 hydroxysteroid dehydrogenase deficiency
109	214950	α methylacyl-CoA racemase deficiency
110	604741	δ4-3-oxosteroid 5β reductase deficiency
111	603711	oxysterol 7α hydroxylase deficiency
112	213700	cerebrotendinous xanthomatosis
113	238600	familial lipoprotein lipase deficiency & apo c2 deficiency
114	200100	Abetalipoproteinemia
115	205400	Tangier disease
116	151660-604367-608600 and other	Inborn hypertriglyceridemia and lipodystrophy syndromes
117	245900-136120	LCAT-deficiency (Norum disease and Fish-eye disease)
118	107680	Hypo-alpha lipoproteinemia
119	268200	LPIN1 lipid myopathy
120	278000	Wolman & cholesteryl ester storage disease
J. peroxisomal disorders		
121	214110	zellweger spectrum (peroxisome biogenesis defects)
122	264470	acyl-CoA oxidase deficiency
123	261515	D-bifunctional protein deficiency
124	214950	2-methyl-CoA racemase deficiency
125	300100	X-linked adrenoleukodystrophy
126	215100	Rhizomelic chondrodysplasia punctata
127	266500	Refsum disease
128	266510	Refsum disease infantile form
K. lysosomal disorders		
129	232300	glycogen storage disease type 2 (Pompe)
130	300257-602743	glycogen storage disease type 2b (Danon and PRK-AG2)
131	230800-230900-231000	Gaucher disease, type I, II and III
132	301500	Fabry disease
133	607014-607016	Hurler-Scheie disease (MPS I)

134	309900	Hunter disease (MPS II)
135	252900-252920-252930-252940	Sanfilippo A; B; C; D (MPS III)
136	253000-253010-253200-253220-253230	other MPS types (IV, V, VI, VII, VIII)
137	257200-607616	Niemann-Pick type A and B
138	257220-607625	Niemann-Pick type C
139	230500-272800 and other	GM1 and GM2 gangliosidosis
140	250100	Metachromatic Leukodystrophy
141	254200	Krabbe leukodystrophy
142	248510	Mannosidosis
143	256550-256540	Sialidosis and galactosialidosis
144	252500-252600-252605	I-cell disease and mucopolipidosis II/III
145	269920-604369	Free sialic storage diseases
146	252650	Mucopolipidosis IV
147	230000	Fucosidosis
148	272200	multiple sulfatase deficiency
149	228000	Farber lipogranulomatosis
150	256730-204500-601780-204200 and other	ceroid lipofuscinosis type 1-10
L. disorders of purine and pyrimidine metabolism		
151	311850-860	phosphoribosyl pyrophosphate synthetase superactivity
152	103050	adenylosuccinate lyase deficiency
153	278300	xanthinuria
154	308000	Lesch-Nyhan syndrome
M. disorders of creatine metabolism		
155	601240	guanidinoacetate methyltransferase deficiency (GAMT)
156	602360	arginine:glycine amidinotransferase deficiency (AGAT)
157	300352	creatine transporter deficiency
N. disorders of neurotransmitter and small peptide metabolism		
158	230450-231900-260005 and other	γ-glutamyl transpeptidase deficiency and other disorders of glutathion metabolism
159	613068	folate transporter defects
160	602079	trimethylaminuria & dimethylglycine dehydrogenase deficiency
161	608643	aromatic amino-acid decarboxylase (AADC)
162	613163	gaba transaminase deficiency
163	300615	monoamine oxidase deficiency
164	223360	dopamine beta-hydroxylase deficiency
165	266100	pyridoxine dependent seizures
166	610090	pyridoxamine 5-phosphate oxidase deficiency (pyri-

		doxal-P responsive seizures)
167	252150-272300	molybdenum cofactor deficiency, sulfite oxidase deficiency
168	271900	acetylaspartic aciduria (Canavan's disease)
O. Congenital defects of glycosylation (CDG)		
169	212065-602579-611209-611182 and other	Congenital disorders of N- and/or O-glycosylation
P. porphyrias		
170	301300	5-aminolevulinic acid synthase deficiency
171	125270	5-aminolevulinic acid dehydratase porphyria
172	176000	acute intermittent porphyria
173	121300	hereditary coproporphyria
174	176200	variegate porphyria
175	263700	congenital erythropoietic porphyria
176	177000	erythropoietic protoporphyria
Q. disorders of copper metabolism		
177	309400	menkes disease
178	227900	Wilson disease
R. other progressive neurodegenerative diseases		
179	203450	Alexander disease (GFAP)
180	256600	Infantile neuroaxonal dystrophy (PLA2G6)
181	603896	Vanishing white matter disease
182	312080	Pelizaeus-Merzbacher disease
S. connective tissue disorders		
183	166200-166210-259420 and other	osteogenesis imperfecta