| | OMIM # | FULL NAME |
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| **A. disorders of amino acid metabolism** | | |
| 1 | 261600 | classical phenylketonuria and hyperphenylala-ninemia |
| 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, homocitrulli-nuria (HHH) |
| 25 | 222700 | lysinuric protein intolerance |
| 26 | 258870 | gyrate atrophy, B6 responsive or non responsive |
| 27 | 238700 | hyperlysinemia (alpha-aminoadipic 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 |

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| 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 |

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| 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 |

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| **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-alphalipoproteinemia |
| 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 PRKAG2) |
| 131 | 230800-230900-231000 | Gaucher disease, type I, II and III |
| 132 | 301500 | Fabry disease |
| 133 | 607014-607016 | Hurler-Scheie disease (MPS I) |

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| 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 mucolipidosis II/III |
| 145 | 269920-604369 | Free sialic storage diseases |
| 146 | 252650 | Mucolipidosis 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 superac-tivity |
| 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 dis-orders 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- |

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| | | 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 prophyria |
| 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 |