Anhang: Inzidenz von angeborenen Stoffwechselstörungen in Österreich

SSIEM classification of Inborn Errors of Metabolism 2011

Disease group / disease	ICD10	OMIM
1. Disorders of amino acid and peptide metabolism		
1.1. Urea cycle disorders and inherited hyperammonaemias		
1.1.1. Carbamoylphosphate synthetase I deficiency		237300
1.1.2. N-Acetylglutamate synthetase deficiency		237310
1.1.3. Ornithine transcarbamylase deficiency		311250
S Ornithine carbamoyltransferase deficiency		
1.1.4. Citrullinaemia type1		215700
S Argininosuccinate synthetase deficiency		
1.1.5. Argininosuccinic aciduria		207900
S Argininosuccinate lyase deficiency		
1.1.6. Argininaemia		207800
S Arginase I deficiency		
1.1.7. HHH syndrome		238970
S Hyperammonaemia-hyperornithinaemia-homocitrullinuria syndrome		
S Mitochondrial ornithine transporter (ORNT1) deficiency		
1.1.8. Citrullinemia Type 2		603859
S Aspartate glutamate carrier deficiency (SLC25A13)		
S Citrin deficiency		
1.1.9. Hyperinsulinemic hypoglycemia and hyperammonemia caused by activating mutations in the GLUD1 gene		138130
1.1.10. Other disorders of the urea cycle		238970
1.1.11. Unspecified hyperammonaemia		238970
1.2. Organic acidurias		
1.2.1. Glutaric aciduria		
1.2.1.1. Glutaric aciduria type I		231670
S Glutaryl-CoA dehydrogenase deficiency		
1.2.1.2. Glutaric aciduria type III		231690
1.2.2. Propionic aciduria	E711	232000
S Propionyl-CoA-Carboxylase deficiency		
1.2.3. Methylmalonic aciduria	E711	251000
1.2.3.1. Methylmalonyl-CoA mutase deficiency		
1.2.3.2. Methylmalonyl-CoA epimerase deficiency		251120
1.2.3.3. Methylmalonic aciduria, unspecified		
1.2.4. Isovaleric aciduria	E711	243500
S Isovaleryl-CoA dehydrogenase deficiency		
1.2.5. Methylcrotonylglycinuria	E744	210200
S Methylcrotonyl-CoA carboxylase deficiency		
1.2.6. Methylglutaconic aciduria	E712	250950
1.2.6.1. Methylglutaconic aciduria type I	E712	250950
S 3-Methylglutaconyl-CoA hydratase deficiency		
1.2.6.2. Methylglutaconic aciduria type II	E723	302060
S Barth syndrome		
S Taffazin deficiency		
1.2.6.3. Methylglutaconic aciduria type III	E723	258501
S Costeff syndrome		
1.2.6.4. Methylglutaconic aciduria type IV	E723	250951
1.2.6.5. Methylglutaconic aciduria type V		610198

1.2.7. 3-Hydroxy-3-methylglutaric aciduria	E728	246450
S 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency		
1.2.8. 2-Methylbutyric aciduria		610006
S 2-Methylbutyryl-CoA dehydrogenase deficiency		
1.2.9. 2-Methyl-3-hydroxybutyric aciduria		300438
S 17-beta-hydroxysteroid dehydrogenase type 10 deficiency		
S HSD10 deficiency		
S 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency		
1.2.10. Alpha-methylacetoacetic aciduria	E712	203750
S Beta-ketothiolase deficiency		
S Mitochondrial acetoacetyl-CoA thiolase deficiency		
S 3-Oxothiolase deficiency		
1.2.11. Isobutyric aciduria		611283
S Isobutyryl-CoA dehydrogenase deficiency		
1.2.12. Methacrylic aciduria	E711	250620
S 3-Hydroxyisobutyryl-CoA deacylase deficiency		
1.2.13. 3-Hydroxyisobutyric aciduria		236795
S 3-Hydroxyisobutyrate dehydrogenase		
1.2.14. Methylmalonate semialdehyde dehydrogenase deficiency		603178
1.2.15. L-2-hydroxyglutaric aciduria		236792
S L-2-hydroxyglutarate dehydrogenase defect		
1.2.16. D-2-hydroxyglutaric aciduria		600721
1.2.16.1. D-2-hydroxyglutarate dehydrogenase deficiency		609186
1.2.16.2. Mitochondrial isocitrate dehydrogenase deficiency		147650
1.2.17. Aminoacylase deficiency		
1.2.17.1. Aminoacylase 1 deficiency		609924
1.2.17.2. Aminoacylase 2 deficiency		271900
S Aspartoacylase deficiency		
S Canavan disease		
S van Bogaert-Bertrand disease		
1.2.18. Methylmalonate semialdehyde dehydrogenase deficiency		603178
1.2.19. Other organic acidurias		
1.3. Disorders of the metabolism of branched-chain amino acids not classified as organic acidurias		
1.3.1. Branched-chain amino acid transferase		238340
1.3.2. Maple syrup urine disease	E710	248600
S Branched-chain alpha-keto acid dehydrogenase complex deficiency	E710	248600
S BCKD deficiency	E710	248600
1.3.2.1. BCKD E1 alpha subunit of deficiency		
S Maple syrup urine disease type Ia		
1.3.2.2. BCKD E1 beta subunit of deficiency		
S Maple syrup urine disease type Ib		
1.3.2.3. Dihydrolipoamide branched chain transacylase deficiency		248610
S Maple syrup urine disease type II		248610
S E2 deficiency		248610
1.3.2.4. Unspecified BCKD deficiency		248610
1.3.3. Other disorders of branched-chain amino acid metabolism		
1.4. Disorders of phenylalanine or tyrosine metabolism		
1.4.1. Phenylalanine hydroxylase deficiency		261600
S Phenylketonuria		
S Mild Hyperphenylalaninaemia		
1.4.2. Tyrosinaemia type II		276600
S Tyrosine aminotransferase deficiency		
1.4.3. Tyrosinaemia type III		276710

	S 4-hydroxyphenylpyruvate dioxygenase deficiency		
1.4.4.	Hawkinsinuria		140350
	S 4-Hydroxyphenylpyruvate hydroxylase deficiency		
1.4.5.	Alkaptonuria		203500
	S Homogentisate 1,2 - dioxygenase deficiency		
1.4.6.	Tyrosinaemia type I		276700
	S Fumarylacetoacetase deficiency		
1.4.7.	Transient tyrosinaemia of the neonate		
1.4.8.	Other disorders of phenylalanine or tyrosine metabolism		
1.5.	**Disorders of the metabolism of sulphur amino acids**		
1.5.1.	Methionine adenosyltransferase I/III deficiency	E721	250850
	S MAT deficiency		
	S MAT I/III deficiency		
	S Hypermethioninemia, Isolated persistent		
1.5.2.	Glycine N-methyltransferase deficiency	E728	606664
	S GNMT deficiency		
1.5.3.	S-adenosylhomocysteine hydrolase deficiency	E721	180960
	S AHCY		
	S SAHH		
	S Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase		
1.5.4.	Cystathionine beta-synthase deficiency	E721	263200
	S Homocystinuria		
	S CBS deficiency		
1.5.5.	Cystathionase deficiency	E721	219500
	S Cystathione gamma - lyase deficiency		
	S Gamma - cystathionase deficiency		
	S Cystathioninuria		
1.5.6.	Isolated sulfite oxidase deficiency	E721	272300
	S Sulphite oxidase deficiency		
	S Sulphocysteinuria		
1.5.7.	Methionine synthase deficiency-cblG	E721	250940
	S Methylcobalamin deficiency, cblG type		
	S Homocystinuria-megaloblastic anemia due to defect in cobalamin metabolism, cblG complementation type		
1.5.8.	Methionine synthase reductase deficiency-cblE	E721	236270
	S Homocystinuria-megaloblastic anemia due to defect in cobalamin metabolism, cblE complementation type		
	S Vitamin B12-responsive homocystinuria, cblE type		
	S Methylcobalamin deficiency, cblE type		
1.5.9.	Other genetic defect in methionine cycle or sulfur amino acid metabolism	E721	
1.5.10.	Unspecified disorder of homocysteine metabolism	E721	
1.5.11.	Unspecified disorder of methionine metabolism	E721	
1.5.12.	Secondary non-genetic disorders of methionine cycle and other sulfur amino acids	E729	
1.6.	**Disorders of histidine, tryptophan or lysine metabolism**		
1.6.1.	Histidinaemia	E708	235800
	S Histidase deficiency		
1.6.2.	Urocanase deficiency	E708	276880
1.6.3.	Glutamate formiminotransferase deficiency	E728	229100
	S Forminotransferase deficiency		
	S Forminoglutamic aciduria		
	S Figlu-uria	E728	229100
1.6.4.	Tryptophanaemia	E708	
	S Tryptophan-2,3-Dioxygenase deficiency		

1.6.5. Hyperlysinaemia		
S Alpha-aminoadipic semialdehyde synthase deficiency		605113
1.6.5.1. Hyperlysinaemia type I		238700
S Lysine : 2-oxoglutarate reductase deficiency		
1.6.5.2. Hyperlysinaemia type II		268700
S Saccharopine dehydrogenase deficiency		
S Saccharopinuria		
1.6.6. 2-Aminoadipic aciduria		204750
1.6.7. 2-Oxoadipic aciduria		245130
1.6.8. Hydroxykynureninuria		236800
S Kynureninase deficiency		
S Xanthurenic aciduria		
1.6.9. Hydroxylysinuria		236900
1.7. Disorders of serine, glycine or glycerate metabolism		
1.7.1. Phosphoglycerate dehydrogenase deficiency	E728	606879
1.7.2. Phosphoserine phosphatase deficiency		172480
1.7.3. Phosphoserine aminotransferase deficiency		610992
1.7.4. Nonketotic hyperglycinaemia	E725	238300
S Glycine cleavage deficiency		
1.7.4.1. P protein deficiency, *GLDC* gene		238300
1.7.4.2. T protein deficiency, *AMT* gene		238310
1.7.4.3. H protein deficiency, *GCSH* gene		238330
1.7.5. Sarcosinaemia	E725	268900
S Sarcosine dehydrogenase deficiency		
1.7.6. D-glyceric aciduria		220120
S D-glycerate kinase deficiency		
1.8. Disorders of ornithine or proline metabolism		
1.8.1. Ornithine aminotransferase deficiency		
S Gyrate atrophy of retina and choroid		
1.8.2. Hyperprolinaemia type I		
S Proline oxidase deficiency		
1.8.3. Hyperprolinaemia type II		
S Pyrroline-5-carboxylate dehydrogenase deficiency		
S Aldehyde dehydrogenase deficiency		
1.8.4. Hypoprolinaemia		
S Pyrroline-5-carboxylate synthase deficiency		138250
1.8.5. Cutis laxa, autosomal recessive, type IIb		179035
S Pyrroline-5-carboxylate reductase deficiency		
1.9. Disorders of amino acid transport		
1.9.1. Lysinuric protein intolerance	E723	222700
S SLC7A7 carrier deficiency		
1.9.2. Cystinuria	E720	220100
1.9.3. Cystinuria-hypotonia syndrome (contiguous gene defect)		606407
1.9.4. Hartnup disease	E720	234500
1.9.5. Iminoglycinuria		242600
1.9.6. Lowe syndrome	E720	309000
1.9.7. Other disorders of amino acid transport		
1.10. Other disorders of amino acid metabolism		
1.10.1. Glutamine synthetase deficiency		
1.11. Disorders of the gamma-glutamyl cycle		
1.11.1. Glutathionuria		
S Gamma-glutamyl transpeptidase deficiency		231950
1.11.2. Cysteinylglycinase deficiency		
1.11.3. Oxoprolinuria		260005
S Oxoprolinase deficiency		

1.11.4. Gamma-glutamylcysteine synthetase deficiency		230450
1.11.5. Glutathione synthetase deficiency		266130
1.12. Other disorders of peptide metabolism		
1.12.1. Prolidase deficiency		170100
S Iminodipeptiuria		
1.12.2. Carnosinaemia		212200
1.12.3. Homocarnosinosis	E728	236130
1.13. Other disorders of amino acid and protein metabolism		
2. Disorders of carbohydrate metabolism		
2.1. Disorders of galactose metabolism		
2.1.1. Classical galactosaemia		230400
S Galactose-1-phosphate uridyltransferase deficiency		
2.1.2. Galactokinase deficiency		230200
2.1.3. Uridine diphosphate galactose-4-epimerase deficiency		230350
2.2. Disorders of fructose metabolism		
2.2.1. Essential fructosuria		229800
S Fructokinase deficiency		
2.2.2. Hereditary fructose intolerance		229600
S Fructose-1-phosphate aldolase deficiency		
2.3. Disorders of pentose metabolism		
2.3.1. Essential pentosuria		260800
S L-xylulose reductase deficiency		
2.3.2. Ribose-5-phosphate isomerase deficiency		608611
2.3.3. Transaldolase deficiency		606003
2.4. Disorders of glycerol metabolism		
2.4.1. Glycerol kinase deficiency		307030
2.4.2. Complex glycerol kinase deficiency due to contiguous gene deletion		300679
2.5. Disorders of glyoxylate metabolism		
2.5.1. Primary hyperoxaluria type I		260000
S Alanine-glyoxylate aminotransferase deficiency		
2.5.2. Primary hyperoxaluria type II		260000
S Hydroxypyruvate reductase deficiency		
S D-glycerate dehydrogenase deficiency		
2.6. Disorders of glucose transport		
2.6.1. Glucose transporter 1 deficiency (blood-brain barrier)		606777
S GLUT1 deficiency syndrome		
2.6.2. Glucose transporter 2 deficiency		227810
S Glycogen storage disease type XI		
S GLUT2 deficiency syndrome		
S Fanconi-Bickel syndrome		261670
2.6.3. Glucose/galactose malabsorption		606824
S Glucose/galactose cotransporter (SGLT1) deficiency		
2.7. Disorders of gluconeogenesis		
2.7.1. Fructose-1,6-bisphosphatase deficiency		229700
2.7.2. Pyruvate carboxylase deficiency		266150
2.7.3. Phosphoenolpyruvate carboxykinase deficiency	E744	261650
2.8. Glycogen storage disorders		
2.8.1. Glycogen storage disease type 1a		232200
S GSD Ia		
S von Gierke disease		
S Glucose-6-phosphatase deficiency		
2.8.2. Glycogen storage disease type 1b		232220
S GSD Ib		
S Glucose-6-phosphate transport deficiency		

2.8.3. Glycogen storage disease type II		232300
S GSD II		
S Pompe disease		
S Lysosomal alpha-1,4-glucosidase deficiency		
2.8.4. Glycogen storage disease type III		232400
S GSD III		
S Cori disease		
S Amylo-1,6-glucosidase (debrancher) deficiency		
2.8.5. Glycogen storage disease type IV		232500
S GSD IV		
S Andersen disease		
S Glycogen branching enzyme deficiency		
2.8.6. Glycogen storage disease type V		232600
S GSD V		
S McArdle disease		
S Muscle phosphorylase deficiency		
2.8.7. Glycogen storage disease type VI		232700
S GSD VI		
S Hers disease		
S Hepatic glycogen phosphorylase deficiency		
2.8.8. Glycogen storage disease type VII		232800
S GSD VII		
S Tauri disease		
S Muscle phosphofructokinase deficiency		
2.8.9. Glycogen storage disease type IX		306000
S GSD IX		
S Phosphorylase kinase deficiency		261740
2.8.9.1. Hepatic phosphorylase kinase deficiency		306000
S GSD IXa		306000
2.8.9.2. Hepatic and muscle phosphorylase kinase deficiency		261750
S GSD Ixb		261750
2.8.9.3. Muscle phosphorylase kinase deficiency		300559
S GSD IXd		300559
2.8.9.4. Cardiac muscle phosphorylase kinase deficiency		261740
2.8.10. Glycogen storage disease type X		
S GSD X		
S Muscle phosphoglycerate mutase deficiency		261670
2.8.11. Glycogen storage disease type XI		227810
S GSD XI		
S Fanconi-Bickel syndrome		261670
S GLUT2 deficiency syndrome		261670
2.8.12. Glycogen storage disease type XIV		
S Muscle phosphoglucomutase 1 deficiency		612934
S GSD XIV		
2.8.13. Glycogen storage disease type XV		
S Glycogenin deficiency		612934
S GSD XIV		
2.8.14. Glycogen storage disease type 0a		240600
S GSD 0a		
S Liver glycogen synthase deficiency		
2.8.15. Glycogen storage disease type 0b		611556
S GSD 0b		
S Muscle glycogen synthase deficiency		
2.8.16. Other glycogen storage disease		
2.8.16.1. Muscle LDH deficiency		612933
2.8.16.2. Aldolase A deficiency		611881

2.8.16.3. Beta-enolase deficiency		612932
2.8.16.4. Muscle phosphoglycerate kinase deficiency		300653
2.8.17. Unspecified glycogen storage disease		
2.9. Other carbohydrate disorders		
2.9.1. Lactose intolerance		223000
S lactase deficiency		
2.9.2. Disaccharide intolerance 1		222900
S Sucrase-isomaltase deficiency		
2.9.3. Trehalase deficiency		612119
3. Disorders of fatty acid and ketone body metabolism		
3.1. Disorders of lipolysis		
3.2. Disorders of carnitine transport and the carnitine cycle		
3.2.1. Carnitine transporter deficiency	E713	212140
S Systemic primary carnitine deficiency		
S Carnitine uptake deficiency		
3.2.2. Carnitine palmitoyltransferase I (CPTI) deficiency	E713	255120
3.2.3. Carnitine acylcarnitine translocase deficiency	E713	212138
3.2.4. Carnitine palmitoyltransferase II (CPTII) deficiency	E713	255110
3.3. Disorders of mitochondrial fatty acid oxidation		
3.3.1. Very long - chain acyl CoA dehydrogenase deficiency	E713	201475
S VLCAD deficiency		
3.3.2. Mitochondrial trifunctional protein deficiency	E713	143450
3.3.2.1. Isolated deficiency of long-chain 3-hydroxyacyl-CoA dehydrogenase	E713	143450
S LCHAD deficiency		
3.3.2.2. Isolated deficiency of long-chain 3-ketoacyl CoA thiolase	E713	143450
3.3.3. Medium - chain acyl CoA dehydrogenase deficiency	E713	201450
S MCAD deficiency		
3.3.4. Short - chain acyl CoA dehydrogenase deficiency	E713	201470
S SCAD deficiency		
3.3.5. 3-alpha-hydroxyacyl- CoA dehydrogenase deficiency	E713	231530
S HADH deficiency		
S Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency		
S SCHAD deficiency		
3.3.6. Multiple acyl-CoA dehydrogenase deficiency	E713	231680
S Glutaric aciduria type II	E713	231680
3.3.6.1. Electron transfer flavoprotein deficiency, alpha chain	E713	231680
3.3.6.2. Electron transfer flavoprotein deficiency, beta chain	E713	130410
3.3.6.3. ETF-ubiquinone oxidoreductase deficiency	E713	231675
S ETF-QO deficiency		
S Electron transfer flavoprotein dehydrogenase deficiency		
3.4. Disorders of ketone body metabolism		
3.4.1. 3-Hydroxy-3-Methylglutaryl-CoA synthase deficieny		600234
3.4.2. Succinyl-CoA:3-Oxoacid-CoA transferase (SCOT) deficiency	E798	245050
3.4.3. Cytosolic acetoacetyl-CoA thiolase deficiency	E712	100678
3.5. Other disorders of fatty acid and ketone body metabolism		
3.5.1. Long - chain acyl CoA dehydrogenase deficiency	E713	201460
3.5.2. Malonyl CoA decarboxylase deficiency	E798	248360
S Malonic aciduria		
4. Disorders of energy metabolism		
4.1. Disorders of pyruvate metabolism		
4.1.1. Pyruvate dehydrogenase complex deficiency		
4.1.1.1. Pyruvate dehydrogenase E1α subunit deficiency		312170

4.1.1.2. Pyruvate dehydrogenase E1β subunit deficiency		179060
4.1.1.3. Dihydrolipoyl transacetylase deficiency		245348
S PDHC E2 deficiency		608782
4.1.1.4. Dihydrolipoyl dehydrogenase deficiency		248600
S PDHC E3 deficiency		608782
4.1.1.5. Pyruvate dehydrogenase E3 binding protein deficiency		245349
S Protein X deficiency		608782
4.1.1.6. Pyruvate dehydrogenase phosphatase deficiency		608782
4.1.1.7. Pyruvate dehydrogenase deficiency, unspecified		312170
4.1.2. Pyruvate kinase deficiency		266200
4.2. Disorders of the citric acid cycle		
4.2.1. 2-Oxoglutarate dehydrogenase deficiency		203740
S 2-Ketoglutarate dehydrogenase complex deficiency		
4.2.2. Fumarase deficiency		136850
S Fumaric aciduria		
4.3. Mitochondrial respiratory chain disorders		
4.3.1. Respiratory chain disorders caused by mutations of mtDNA		
4.3.1.1. Large-scale single deletion of mtDNA		
4.3.1.1.1. Pearson Syndrome		557000
4.3.1.1.2. Kearns Sayre Syndrome		530000
S Chronic External Ophthalmoplegia (CPEO), pigmentary degeneration of retina, myopathy and cardiomyopathy [onset before 20 yrs]		
4.3.1.1.3. Chronic Progressive External Ophthalmoplegia (CPEO) with Mitochondrial Myopathy [onset after 20 yrs]		n/a
4.3.1.2. Point mutations of mtDNA		
4.3.1.2.1. Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes, MELAS		540000
4.3.1.2.2. Myoclonic epilepsy associated with ragged red fibres, MERRF		545000
4.3.1.2.3. Neuropathy Ataxia and Retinitis Pigmentosa, NARP		551500
4.3.1.2.4. Leber Hereditary Optic Neuropathy, LHON		535000
4.3.1.2.5. Maternally Inherited Leigh Syndrome, MILS		256000
4.3.1.2.6. Sporadic Leigh Syndrome		256000
4.3.1.2.7. Maternally inherited Mitochondrial Dystonia		500001
4.3.1.2.8. Maternally inherited Mitochondrial Cardiomyopathy		n/a
4.3.1.2.9. Maternally inherited Mitochondrial Myopathy		n/a
4.3.1.2.9.1. 'Pure' Mitochondrial Myopathy		n/a
4.3.1.2.9.2. Lethal Infantile Mitochondrial Myopathy		551000
4.3.1.2.9.3. Mitochondrial Myopathy with Diabetes Mellitus		500002
4.3.1.2.9.4. Mitochondrial Myopathy with Reversible cytochrome c oxidase (COX) Deficiency		500009
4.3.1.2.10. Maternally inherited deafness and diabetes, MIDD		520000
4.3.2. Respiratory chain disorders caused by mutations of nuclear DNA		
4.3.2.1. Mitochondrial DNA Depletion Syndromes		
4.3.2.1.1. Alpers-Huttenlocher Syndrome (*POLG*)		203700
4.3.2.1.2. Hepatocerebral (*DGUOK*, *MPV17*, *PEO1*)		251880
4.3.2.1.3. Myopathic (*TK2*)		609560
4.3.2.1.4. Encephalomyopathy with methylmalonic aciduria (*SUCLA2*)		612073
4.3.2.1.5. Fatal Infantile Lactic Acidosis with methylmalonic aciduria (*SUCLG1)*		245400
4.3.2.1.6. Encephalomyopathic with renal tubulopathy (*RRM2B*)		612075
4.3.2.1.7. Childhood-onset autosomal dominant optic atophy		165500

(*OPA1*)		
4.3.2.1.8. Mitochondrial Neurogastrointestinal Encephalopathy, MNGIE (*ECGF1*)		603041
4.3.2.2. Multiple mtDNA Deletion Syndromes		
4.3.2.2.1. Progressive External Ophthalmoplegia Autosomal Dominant (PEOA)		
4.3.2.2.1.1. PEOA1 (*POLG*)		157640
4.3.2.2.1.2. PEOA2 (*ANT1*)		609283
4.3.2.2.1.3. PEOA3 (*PEO1*)		609286
4.3.2.2.1.4. PEOA4 (*POLG2*)		610131
4.3.2.2.1.5. PEOA5 (*RRM2B*)		613077
4.3.2.2.2. Progressive External Ophthalmoplegia Autosomal Recessive (PEOB)		258450
4.3.2.2.3. Sensory Ataxic Neuropathy, Dysarthria and Ophthalmoparesis, SANDO		607459
S Mitochondrial Recessive Ataxic Syndrome, MIRAS (*POLG*)		
S Spinocerebellar Ataxia with Epilepsy, SCAE		
4.3.2.2.4. Optic Atrophy 1 and Deafness (*OPA1*)		125250
S Optic Atrophy, Deafness, Ophthalmoplegia, Myopathy		
4.3.2.3. Leigh Syndrome, LS		256000
S Subacute Necrotizing Encephalopathy		
4.3.2.3.1. LS with leukodystrophy (*SDHA*, *SURF1*)		220110
4.3.2.3.2. LS with cardiomyopathy (*COX10*, *COX15*)		220110
4.3.2.3.3. LS with French-Canadian ethnicity (*LRPPRC*)		220111
4.3.2.3.4. LS with nephrotic syndrome (*PDSS2*)		607426
4.3.2.3.5. LS with nephropathy (*COQ2*)		607426
4.3.2.4. Ubiquinone (CoQ10) deficiency (Non-LS)		607426
4.3.2.4.1. Early-onset ataxia with oculomotor apraxia and hypoalbuminaemia (*APTX*)		607426
4.3.2.4.2. Deafness, encephaloneuropathy, obesity and valvulopathy (*PDSS1*)		607426
4.3.2.4.3. Cerebellar atrophy, ataxia and seizures (*CABC1*)		607426
4.3.2.5. Growth Retardation, Aminoaciduria, Cholestasis, Iron overload, Lactic acidosis and Early death (GRACILE) Syndrome (*BCS1L*)		603358
4.3.2.6. Renal tubulopathy, encephalopathy and liver failure (*BCS1L*)		124000
4.3.2.7. Cardio-encephalopathy with hyperammonaemia (*TMEM70*)		604273
S ATP synthase deficiency, nuclear-encoded		
4.3.2.8. Exercise Intolerance with Lactic Acidosis		
4.3.2.8.1. Complex I deficiency; riboflavin responsive (*ACAD9*)		611126
4.3.2.8.2. Complex I and II deficiency (*ISCU*)		255125
4.3.2.9. Isolated Oxidative Phosphorylation Defects with Variable Phenotype (Not Classified Elsewhere)		
4.3.2.9.1. Complex I structural subunit gene defect (NDUFV1, NDUFV2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFA1, NDUFA2, NDUFA11)		n/a
4.3.2.9.2. Complex I assembly gene defect (C20orf7, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, C80orf38, NUBPL, FOXRED1)		n/a
4.3.2.9.3. Complex II structural subunit gene defect (SDHA, SDHB, SDHC, SDHD)		n/a
4.3.2.9.4. Complex II assembly gene defect (SDHAF1)		n/a
4.3.2.9.5. Complex III structural subunit gene defect (UQCRB, UQCRQ)		n/a

4.3.2.9.6. Complex III assembly gene defect		n/a
4.3.2.9.7. Complex IV structural subunit gene defect (COX6B1)		n/a
4.3.2.9.8. Complex IV assembly gene defect (SCO1, SCO2, SURF1, COX10, COX15, TACO1, FASTKD2)		n/a
4.3.2.9.9. Complex V structural subunit gene defect (ATP5E)		n/a
4.3.2.9.10. Complex V assembly gene defect (ATPAF2, TMEM70)		n/a
4.3.2.10. Mitochondrial Protein Translation Defects		
4.3.2.10.1. Combined Oxidative Phosphorylation Defect 1, COXPD1 (EFG1)		609060
4.3.2.10.2. Combined Oxidative Phosphorylation Defect 2, COXPD2 (MRPS16)		610498
4.3.2.10.3. Combined Oxidative Phosphorylation Defect 3, COXPD3 (TSFM)		610505
4.3.2.10.4. Combined Oxidative Phosphorylation Defect 4, COXPD4 (TUFM)		610678
4.3.2.10.5. Combined Oxidative Phosphorylation Defect 5, COXPD5 (MRPS22)		611719
4.3.2.10.6. Combined Oxidative Phosphorylation Defect 6, COXPD6 (AIFM1)		300816
4.3.2.10.7. Combined Oxidative Phosphorylation Defect 7, COXPD7 (C10ORF65)		613559
4.3.2.10.8. Myopathy, Lactic Acidosis and Sideroblastic Anaemia 1, MLASA1 (PUS1)		600462
4.3.2.10.9. Acute Infantile Liver Failure (TRMU)		613070
4.3.2.10.10. Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation, LBSL (DARS2)		611105
4.3.2.10.11. Pontocerebellar hypoplasia Type 6 (RARS2)		611523
4.3.2.10.12. Myopathy, Lactic Acidosis and Sideroblastic Anaemia 2, MLASA2 (YARS2)		613561
4.3.3. Respiratory chain deficiencies with no known genetic basis		
4.3.3.1. Complex I deficiency		252010
4.3.3.2. Complex II deficiency		252011
4.3.3.3. Complex III deficiency		124000
4.3.3.4. Complex IV deficiency		220110
4.3.3.5. ATP synthase deficiency		604273
4.3.3.6. Combined respiratory chain deficiency		n/a
4.4. Mitochondrial membrane transport disorders		
4.4.1. Mitochondrial substrate carrier disorders		
4.4.1.1. Mitochondrial phosphate carrier deficiency (SLC25A3)		600370
4.4.1.2. Mitochondrial aspartate glutamate carrier 1 deficiency (SLC25A12)		603667
4.4.1.3. Mitochondrial glutamate carrier 1 deficiency (SLC25A22)		609302
4.4.1.4. Mitochondrial carrier SLC25A38, haem biosynthesis, sideroblastic anaemia		610819
4.4.2. Mitochondrial protein import disorders		
4.4.2.1. Mohr-Tranebjaerg syndrome (TIMM8A)		300356
4.5. Unspecified mitochondrial disorders		
4.5.1. Leigh syndrome with no known genetic or respiratory chain deficiency		256000
4.5.2. Ethylmalonic Encephalopathy (*ETHE1*)		602473
4.5.3. Anaemia, sideroblastic, and spinocerebellar ataxia, ASAT (*ABCB7*)		301310
4.6. Disorders of creatine metabolism		
4.6.1. Creatine transporter deficiency		
S X-linked creatine deficiency syndrome		300352
S SLC6A8 deficiency		300352
4.6.2. Guanidinoacetate methyltransferase deficiency		612736

4.6.3. Arginine:glycine amidinotransferase deficiency		612718
4.7. Other disorders of energy metabolism		
5. Disorders in the metabolism of purines, pyrimidines and nucleotides		
5.1. Disorders of purine metabolism		
5.1.1. Primary idiopathic gout		138900
5.1.2. Familial juvenile hyperuricaemic nephropathy		162000
S Familial nephropathy with gout		
5.1.3. Adenylosuccinate lyase deficiency		103050
5.1.4. AICAR transformylase deficiency		601731
S IMP cyclohydrolase deficiency		
5.1.5. Adenosine deaminase deficiency		102700
5.1.6. Deoxyguanosine kinase deficiency		251880
5.1.7. Myoadenylate deaminase deficiency		102770
5.1.8. Lesch-Nyhan syndrome		308000
S Hypoxanthine-guanine phosphoribosyltransferase deficiency		
5.1.9. Adenine phosphoribosyl transferase deficiency		102600
5.1.10. Phosphoribosyl pyrophosphate synthetase 1 defects		311850
5.1.10.1. Phosphoribosyl pyrophosphate synthase superactivity		300661
5.1.10.2. X-linked Charcot-Marie-Tooth disease-5		311070
5.1.10.3. Arts syndrome		301835
5.1.10.4. X-linked sensorineural deafness		304500
5.1.11. Inosine triphosphatase deficiency		147520
5.1.12. Adenosine deaminase superactivity		
5.1.13. Purine nucleoside phosphorylase deficiency		164050
5.1.14. Mitochondrial Ribonucelotide Reductase subunit 2 deficiency		604712
5.1.15. Xanthinuria type I		278300
S Xanthine oxidase deficiency		
5.1.16. Xanthinuria type II		603592
S Combined deficiency of xanthine and aldehyde oxidase		
5.1.17. Thiopurine S-methyltransferase deficiency		610460
5.2. Disorders of pyrimidine metabolism		
5.2.1. Orotic aciduria type I		258900
S Uridine monophosphate synthase deficiency		
5.2.2. Orotic aciduria type II		258920
S Orotidine - 5 -phosphate decarboxylase deficiency		
5.2.3. Pyrimidine - 5 - nucleotidase deficiency		266120
5.2.4. Dihydroorotate dehydrogenase deficiency		263750
5.2.5. Uridine-5'-monophosphate hydrolase superactivity		266120
5.2.6. Thymidine phosphorylase deficiency		131222
5.2.7. Thymidine kinase 2 deficiency		609560
5.2.8. Dihydropyrimidine dehydrogenase deficiency		274270
5.2.9. Dihydropyrimidinase deficiency		222748
5.2.10. Beta-ureidopropionase deficiency		613161
S Beta-alanine synthase deficiency		
5.2.11. Hyper-beta-alaninaemia		237400
S Beta-alanine-2-ketoglutarate transaminase deficiency		
5.2.12. Beta-aminoisobutyrate-pyruvate transaminase deficiency		210100
5.3. Disorders of nucleotide metabolism		
5.3.1. Aicardi-Goutières Syndrome (AGS)		
5.3.1.1. AGS1		225750
S TREX1 deficiency		
S DNase III deficiency		
5.3.1.2. AGS2		610181
S RNASEH2B deficiency		

5.3.1.3. AGS3		610181
S RNASEH2C deficiency		
5.3.1.4. AGS4		610181
S RNASEH2A deficiency		
5.3.1.5. AGS5		612952
S SAMHD1deficiency		
5.3.2. RNASET2-deficient cystic leukoencephalopathy		612951
6. Disorders of the metabolism of sterols		
6.1. Disorders of sterol biosynthesis		
6.1.1. Mevalonate kinase deficiency		610377
S Mevalonic aciduria		610377
S Hyper-IgD syndrome (HIDS)		260920
6.1.2. Smith - Lemli - Opitz syndrome	Q871	270400
S 7-Dehydrocholesterol reductase deficiency	Q871	270400
6.1.3. X-linked dominant chondrodysplasia punctata 2		302960
S Conradi-Hünermann syndrome		302960
S 3β-hydroxysteroid- Δ8, Δ7-isomerase deficiency		302960
6.1.4. Congenital hemidysplasia with ichtyosiform erythroderma and limb defects		308050
S CHILD syndrome		308050
S 3β-hydroxysteroid C-4 dehydrogenase deficiency		308050
6.1.5. Desmosterolosis		602398
S Desmosterol reductase deficiency		602398
S 3β-hydroxysterol- Δ24-reductase deficiency		602398
6.1.6. Lathosterolosis		607330
S 3β-hydroxysterol Δ5-desaturase deficiency		607330
6.1.7. Greenberg skeletal dysplasia		215140
S Hydrops-ectopic calcification-moth-eaten skeletal dysplasia		215140
S 3β-hydroxysterol Δ14-reductase deficiency		215140
6.2. Disorders of bile acid biosynthesis		
6.2.1. 3- β-hydroxysterol Δ5-oxidoreductase/isomerase deficiency		
S Progressive familial intrahepatic cholestasis type 4		
6.2.2. Δ4-3-oxysterol 5β-reductase deficiency		
6.2.3. Oxysterol 7-alpha-hydroxylase		
6.2.4. Cholesterol 7-alpha-hydroxylase		
6.2.5. Cerebrotendinous xanthomatosis		213700
S Sterol 27-hydroxylase deficiency		213700
S Van Bogaert-Scherer-Epstein disease		213700
S Cholestanol storage disease		213700
6.3. Disorders of bile acid metabolism and transport		
6.3.1. Bilirubin UDP-glucuronosyltransferase 1 deficiency		
S Crigler-Najjar disease		
S Gilbert disease		
6.3.2. Byler disease		
S Progressive familial intrahepatic cholestasis type 1		
S ATP8B1 deficiency		
6.3.3. Progressive familial intrahepatic cholestasis type 2		
S ABCB11 deficiency		
6.3.4. Progressive familial intrahepatic cholestasis type 3		
S ABCB4 deficiency		
S Class III multidrug resistance P-glycoprotein deficiency		
6.4. Other disorders in the metabolism of sterols		
6.4.1. X-linked ichthyosis		308100
S Steroid sulphatase deficiency		

S Steroid sulphatase deficiency due to contiguous gene deletion		
7. Disorders of porphyrin and haem metabolism		
7.1.1. Erythropoietic porphyria		177000
S Ferrochelatase deficiency		
7.1.2. X-linked dominant protoporphyria		300752
S Erythroid d 5-aminolevulinate synthase (gain of function)		
7.1.3. Variegate porphyria		176200
S Protoporphyrinogen oxidase deficiency		
7.1.4. X-linked sideroblastic anaemia (XLSA)		300751
S Erythroid 5-aminolevulinate deficiency		
7.1.5. Congenital erythropoietic porphyria		263700
S Uroporphyrinogen III synthase deficiency		
7.1.6. Acute intermittent porphyria		176000
S Porphobilinogen deaminase deficiency		
7.1.7. Hereditary coproporphyria		121300
S Coproporphyrinogen oxidase deficiency		
7.1.8. Porphyria cutanea tarda type I (sporadic)		176090
S Hepatic uroporphyrinogen decarboxylase deficiency		
7.1.9. Porphyria cutanea tarda type II (familial)		176100
S Uroporphyrinogen decarboxylase deficiency		
7.1.10. Acute hepatic porphyria		612740
S Delta aminolevilinic acid dehydratase deficiency		
8. Disorders of lipid and lipoprotein metabolism		
8.1. Inherited hypercholesterolaemias		
8.1.1. Disorder of low density lipoprotein receptor	E780	143890
S Fredrickson type IIa hyperlipoproteinaemia	E780	143890
8.1.1.1. Familial hypercholesterolaemia – homozygous	E780	
8.1.1.2. Familial hypercholesterolaemia – heterozygous	E780	
8.1.2. Sitosterolaemia	E755	210250
S Phytosterolaemia		210250
S Sitosterolaemia with xanthomatosis	E755	210250
8.2. Inherited hypertriglyceridaemias		
8.2.1. Familial chylomicronaemia	E786	238600
S Hyperlipidaemia Type 1	E786	238600
8.2.1.1. Familial lipoprotein lipase deficiency	E786	238600
8.2.1.2. Familial apolipoprotein C - II deficiency	E786	207750
8.2.2. Familial hypertriglyceridaemia	E786	238600
8.3. Inherited mixed hyperlipidaemias		
8.3.1. Familial dysbetalipoproteinaemia	E782	107741
S Apolipoprotein E deficiency	E782	107741
S Fredrickson type III hyperlipoproteinaemia	E782	107741
S Remnant hyperlipidaemia	E782	107741
8.3.1.1. Dysfunctional apo E		
8.3.2. Familial combined hyperlipoproteinaemia		
8.3.3. Hepatic lipase deficiency		
8.4. Disorders of high density lipoprotein metabolism		
8.4.1. Apolipoprotein A-I deficiency	E786	
8.4.2. Tangier disease	E786	205400
S Familial hypoalphalipoproteinaemia	E786	205400
8.4.3. Lecithin cholesterol acyltransferase deficiency		
8.4.3.1. Fish-eye disease	E786	136120
8.4.3.2. Norum disease	E786	245900
8.4.4. Familial hyperalphalipoproteinaemia		

S Cholesterol ester transfer protein deficiency		
8.5. Inherited hypolipidaemias		
8.5.1. Familial abetalipoproteinaemia	E786	200100
S Microsomal triglyceride transfer protein (MTTP) deficiency	E786	157147
8.5.2. Familial hypobetalipoproteinaemia	E786	200100
S Apolipoprotein B deficiency	E786	107730
8.5.3. Anderson disease		
8.6. Other disorders of lipid and lipoprotein metabolism		
8.6.1.1. Sjøgren - Larsson syndrome	Q898	270200
S Fatty alcohol:NAD+ oxidoreductase deficiency	Q898	270200
8.6.1.2. Pancreatic triacylglycerol lipase deficiency	E888	246600
8.6.1.3. Pancreatic colipase deficiency	E755	120105
8.7. Unspecified disorders of lipid and lipoprotein metabolism		
9. Congenital disorders of glycosylation and other disorders of protein modification	E778	
S CDG	E778	
9.1. Disorders of protein N-glycosylation		
9.1.1. Phosphomannomutase 2 deficiency	E744	601785
S PMM2-CDG	E744	601785
S CDG-Ia	E744	601785
9.1.2. Phosphomannose isomerase deficiency	E778	602579
S MPI-CDG		602579
S CDG-Ib	E778	602579
9.1.3. Glucosyltransferase 1 deficiency	E744	603147
S ALG6-CDG		603147
S CDG-Ic	E744	603147
9.1.4. Mannosyltransferase 6 deficiency	E744	601110
S NOT56L-CDG		601110
S CDG-Id	E744	601110
9.1.5. Mannosyltransferase 8 deficiency	E744	607143
S ALG 12-CDG		607143
S CDG-Ig	E744	607143
9.1.6. Glucosyltransferase 2 deficiency	E744	608104
S ALG 8-CDG		608104
S CDG-Ih	E744	608104
9.1.7. Mannosyltransferase 2 deficiency		607906
S ALG 2-CDG		607906
S CDG-Ii		607906
9.1.8. UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency		608093
S DPAGT1-CDG		608093
S CDG-Ij		608093
9.1.9. Mannosyltransferase 1 deficiency		608540
S HMT1-CDG		608540
S CDG-Ik		608540
9.1.10. Mannosyltransferase 7-9 deficiency		608776
S DIBD1-CDG		608776
S CDG-Il		608776
9.1.11. Flippase of Man5GlcNAc2-PP-Dol deficiency		611633
S RFT1-CDG		611633
S CDG-In		611633
9.1.12. N-acetylglucosaminyltransferase deficiency		602616
S MGAT2-CDG		602616
S CDG-IIa		602616
9.1.13. Glucosidase 1 deficiency		606056

S GLS1-CDG		606056
S CDG-IIb		606056
9.1.14. TUSC3-CDG		601385
9.1.15. SRD5A3-CDG		
9.2. Disorders of protein O-glycosylation	E744	
9.2.1. O-xylosylglycan synthesis deficiencies		
9.2.1.1. EXT1 deficiency		608177
9.2.1.2. EXT2 deficiency		608210
9.2.1.3. Beta-1,4-galactosyltransferase 7 deficiency		604327
S B4GALT7 deficiency		604327
9.2.2. O-N-acetylgalactosaminylglycan synthesis deficiencies		
9.2.2.1. Polypeptide N-acetylgalactosaminyl transferase deficiency		601756
S GALTNT3 deficiency		601756
9.2.3. O-xylosyl/N-acetylgalactosaminylglycan synthesis deficiencies		
9.2.3.1. SLC35D1 deficiency		610804
9.2.4. O-mannosylglycan synthesis deficiencies		
9.2.4.1. Protein-O-mannosyltransferase 1 deficiency		607423
S POMT1 deficiency		607423
9.2.4.2. Protein-O-mannosyltransferase 2 deficiency		607423
S POMT1 deficiency		607423
9.2.4.3. Protein-O-mannose beta-1,2-N-acetyglucosaminyltransferase deficiency	E744	606822
S POMGNT1 deficiency	E744	606822
9.2.4.4. Fukutin deficiency	E744	607440
S FKTN deficiency		607440
9.2.4.5. Fukutin-related protein deficiency		606596
S FKRP deficiency	E744	606596
9.2.4.6. N-acetylglucosaminyltransferase-like protein deficiency		603590
S LARGE deficiency		603590
9.2.4.7. O-fucose-specific beta-1,3-N-acetylglucosaminyltransferase deficiency		602576
S SCDO3 deficiency		602576
9.2.4.8. O-fucose-specific beta-1,3-N-glucosyltransferase deficiency		610308
S B3GALTL deficiency		610308
9.3. Disorders of glycosphingolipid and glycosylphosphatidylinositol anchor glycosylation		
9.3.1.1. Lactosylceramide alpha-2,3-sialyltransferase deficiency		609056
S SIAT9 deficiency		609056
9.3.1.2. Phosphatidylinositolglycan, class M deficiency		610273
S PIGM deficiency		610273
9.4. Disorders of multiple glycosylation and other glycosylation pathways		
9.4.1. GDP-Man:Dol-P mannosyltransferase deficiency		603503
S DPM1-CDG		603503
S CDG-Ie		603503
9.4.2. Lec35 deficiency		608799
S MPDU1-CDG		608799
S CDG-If		608799
9.4.3. Beta-1,4-galactosyltransferase 1 deficiency		607091
S B4GALT1-CDG		607091
S CDG-IId		607091
9.4.4. UDP-GlcNAc epimerase/kinase deficiency		600737
S GNE-CDG		600737
9.4.5. CMP-sialic acid transporter deficiency		605634
S SLC35A1-*CDG*		605634

S CDG-IIf		605634
9.4.6. GDP-fucose transporter deficiency		605881
S SLC35C1-CDG		605881
S CDG-IIc		605881
9.4.7. Dolichol pathway deficiencies		
9.4.7.1. Dolichol kinase deficiency		610768
S DK1-CDG		610768
S CDG-Im		610768
9.4.8. Conserved oligomeric Golgi (COG) complex deficiency		
9.4.8.1. Component of COG complex 7 deficiency		606978
S CDG-IIe		606978
9.4.8.2. Component of COG complex 1 deficiency		606973
S CDG-IIg		606973
9.4.8.3. Component of COG complex 8 deficiency		606979
9.4.9. V-ATPase deficiencies		
9.4.9.1. V0 subunit A2 of vesicular H(+)-ATPase deficiency		611716
S ATP6VOA2-*CDG*		611716
S COPII component SEC23B		610512
S SEC23B-CDG(CDAII)		610512
9.5. Disorders of protein ubiquitinylation		
9.6. Other disorders of protein modification		
10. Lysosomal disorders		
10.1. Mucopolysaccharidoses	E76.	
10.1.1. MPS I, Hurler, Scheie disease	E76.0	252800
S Alpha-iduronidase deficiency		
10.1.2. MPS II, Hunter disease	E76.1	309900
S Iduronate 2-sulphatase deficiency		
10.1.3. MPS IIIA, Sanfilippo A disease	E76.2	252900
S Heparan - N - sulphatase deficiency		
10.1.4. MPS IIIB, Sanfilippo B disease	E76.2	252920
S N-acetyl-alpha-D-glucosaminidase deficiency		
10.1.5. MPS IIIC, Sanfilippo C disease	E76.2	252930
S Acetyl-CoA alpha-glucosaminide acetyltransferase deficiency		
10.1.6. MPS IIID, Sanfilippo D disease	E76.2	252940
S N-acetylglucosamine-6-sulphatase deficiency		
10.1.7. MPS IVA, Morquio A disease	E76.2	253000
S N-acetylgalactosamine-6-sulphatase deficiency		
10.1.8. MPS IVB, Morquio B disease	E76.2	253010
S Beta-galactosidase deficiency		
10.1.9. MPS VI, Maroteaux - Lamy disease	E76.2	253200
S N-acetylgalactosamine - 4 - sulphatase deficiency		
S Arylsulphatase B deficiency		
10.1.10. MPS VII, Sly disease	E76.2	253220
S Beta-glucuronidase deficiency		
10.1.11. MPS IX	E76.2	601492
S Hyaluronidase deficiency		
10.2. Oligosaccharidoses	E77.0	
10.2.1. Aspartylglucosaminuria	E77.1	208400
S Aspartylglucosaminidase deficiency		
10.2.2. Fucosidosis	E77.1	230000
S Alpha-fucosidase deficiency		
10.2.3. Alpha - D – mannosidosis	E77.1	248500
S Alpha-mannosidase deficiency		
10.2.4. Beta - D – mannosidosis	E77.1	248510

S Beta-mannosidase deficiency		
10.2.5. Schindler disease	E77.1	104170
S Alpha-N-acetylgalactosaminidase deficiency		
10.2.5.1. Schindler disease type I	E77.1	104170
10.2.5.2. Kanzaki disease	E77.1	104170
S Schindler disease type II		
10.2.6. Sialidosis	E77.1	256550
S Alpha-neuraminidase		
10.3. Sphingolipidoses	E75.0	
10.3.1. GM1-gangliosidosis	E75.1	230500
S Beta-galactosidase deficiency		
10.3.2. GM2-gangliosidosis	E75.0	268800
10.3.2.1. GM2-gangliosidosis 0-variant,	E75.0	268800
S Sandhoff disease		
S Total hexosaminidase deficiency		
10.3.2.2. GM2-gangliosidosis B-variant	E75.0	272800
S Tay-Sachs disease		
S Hexosaminidase A deficiency		
10.3.2.3. GM2-gangliosidosis AB-variant	E75.0	272750
S GM2 activator deficiency		
10.3.3. Gaucher disease	E75.2	230800
S Glucocerebrosidase deficiency		
10.3.4. Krabbe disease	E75.2	245200
S Galactocerebrosidase deficiency		
10.3.5. Metachromatic leukodystrophy		250100
S Arylsulphatase A deficiency	E75.2	
10.3.6. Prosaposin deficiency	E75.2	176801
10.3.6.1. Saposin A deficiency	E75.2	611722
S Krabbe disease due to Saposin A deficiency	E75.2	
10.3.6.2. Saposin B deficiency	E75.2	249900
S Metachromatic leukodystrophy due to Saposin B deficiency	E75.2	
10.3.6.3. Saposin C deficiency	E75.2	610539
S Gaucher disease due to Saposin C deficiency	E75.2	
10.3.6.4. Saposin D deficiency		
10.3.7. Fabry disease	E75.2	301500
S Alpha-galactosidase deficiency		
10.3.8. Farber disease	E75.2	228000
S Ceramidase deficiency		
10.3.9. Niemann-Pick disease type A or B	E75.2	257200
S Sphingomyelinase deficiency		
10.3.10. Niemann-Pick disease type C	E75.2	257220
10.3.10.1. Niemann-Pick disease type C1	E75.2	257220
10.3.10.2. Niemann-Pick disease type C2	E75.2	607625
10.4. Ceroid lipfuscinoses, neuronal (CLN)		
10.4.1. CLN1, Santavuori-Haltia disease	E75.4	256730
S Lysosomal palmitoyl protein thioesterase-1 deficiency	E75.4	
10.4.2. CLN2, Jansky-Bielschowsky disease	E75.4	204500
S Lysosomal tripeptidyl-peptidase-1 deficiency	E75.4	
10.4.3. CLN3, Batten Spielmeyer-Vogt disease	E75.4	204200
S Lysosomal transmembrane CLN3 protein deficiency	E75.4	
10.4.4. CLN4A, Kufs disease recessive type	E75.4	204300
10.4.5. CLN4B Kufs disease dominant type	E75.4	162350
10.4.6. CLN5 Finnish variant	E75.4	256731
S Lysosomal transmembrane CLN5 protein deficiency	E75.4	
10.4.7. CLN6	E75.4	601780

10.4.8. CLN7	E75.4	610950
S CLN Turkish variant	E75.4	
10.4.9. CLN8, Northern epilepsy type	E75.4	600143
10.4.10. CLN9	E75.4	609055
10.4.11. CLN10	E75.4	610127
S Cathepsin D deficiency	E75.4	
10.5. Lysosomal export disorders		
10.5.1. Cystinosis	E72.0	219800
S Cystinosin deficiency		
10.5.2. Salla disease/infantile sialic acid storage disease		269920
S Solute carrier family 17 member 5 (SLC17A5) deficiency		
10.6. Other lysosomal disorders		
10.6.1. Mucolipidosis II, I-cell disease	E77.0	252500
S N-acetylglucosamine-1-phosphotransferase (alpha/beta) deficiency		
10.6.2. Mucolipidosis III, Pseudo-Hurler polydystrophy	E77.0	252605
S N-acetylglucosamine-1-phosphotransferase (gamma) deficiency		
10.6.3. Mucolipidosis IV	E75.1	252650
S Mucolipin-1 deficiency		
10.6.4. Multiple sulphatase deficiency	E76.2	272200
S Sulphatase-modifying factor 1 (SUMF-1) deficiency		
10.6.5. Wolman/cholesterol ester storage disease	E75.5	278000
S Acid lipase deficiency		
10.6.6. Pompe disease, GSD type II	E74.0	232300
S Acid alpha-1,4-glucosidase deficiency		
10.6.7. Sialuria		269921
S UDP-N-acetylglucosamine 2-epimerase deficiency		
10.6.8. Danon disease		300257
S Lysosomal-associated membrane protein-2 (LAMP2) deficiency		
10.6.9. Cathepsin-related disorders		265800
10.6.9.1. Galactosialidosis	E77.1	256540
S Lysosomal protective protein deficiency		
S Cathepsin A deficiency		
10.6.9.2. Papillon-Lefèvre syndrome		245000
S Cathepsin C deficiency		
10.6.9.3. Pycnodysostosis		265800
S Cathepsin K deficiency		
10.6.10. Hermansky-Pudlak Syndrome	E70.3	203300
11. Peroxisomal disorders		
11.1. Disorders of peroxisome biogenesis		
S Zellweger spectrum disorder		214100
S Hyperpipecolic acidaemia		239400
11.1.1. Zellweger spectrum disorder, severe form		214100
S Zellweger syndrome		214100
11.1.2. Zellweger spectrum disorder, attenuated form		214100
11.1.2.1. Neonatal adrenoleukodystrophy		202370
11.1.2.2. Infantile Refsum disease		266510
11.1.3. Zellweger spectrum disorder, unclassified clinical severity		214100
11.1.3.1. PEX1 deficiency		602136
11.1.3.2. PEX2 deficiency		170993
11.1.3.3. PEX3 deficiency		603164
11.1.3.4. PEX5 deficiency		600414
11.1.3.5. PEX6 deficiency		601498

11.1.3.6. PEX10 deficiency		602859
11.1.3.7. PEX12 deficiency		601758
11.1.3.8. PEX13 deficiency		601789
11.1.3.9. PEX14 deficiency		601791
11.1.3.10. PEX16 deficiency		603360
11.1.3.11. PEX19 deficiency		600279
11.1.3.12. PEX26 deficiency		608666
11.2. Rhizomelic chondrodysplasia punctate		
11.2.1. Rhizomelic chondrodysplasia punctata type 1		215100
S PTS2 receptor deficiency		
S PEX7 deficiency		601757
11.2.2. Rhizomelic chondrodysplasia punctata type 2		222765
S Isolated dihydroxyacetone phosphate acyltransferase deficiency		
11.2.3. Rhizomelic chondrodysplasia punctata type 3		600121
S Isolated alkyl-dihydroxyacetone phosphate synthase deficiency		
11.3. Disorders of peroxisomal alpha-, beta and omega-oxidation		
11.3.1. X-linked adrenoleukodystrophy		300100
S Schilder disease		
S ALD		
11.3.2. Peroxisomal acyl-CoA oxidase 1 deficiency		264470
11.3.3. Peroxisomal D-bifunctional protein deficiency		261515
S DBP deficiency		
11.3.4. Sterol carrier protein deficiency		
S SCPx deficiency		
11.3.5. Alpha-methylacyl-CoA racemase deficiency		604489
S AMACR deficiency		
11.3.6. Refsum disease		266500
S Phytanoyl-CoA hydroxylase deficiency		
11.4. Other peroxisomal disorders		
11.4.1. Primary hyperoxaluria type I		259900
S Alanine:glyoxylate aminotransferase deficiency		
11.4.2. Acatalasaemia		115500
S Catalase deficiency		
12. Disorders of neurotransmitter metabolism		
12.1. Disorders in the metabolism of biogenic amines		
12.1.1. Tyrosine hydroxylase deficiency		191290
12.1.2. Aromatic L-amino acid decarboxylase deficiency	E728	608643
12.1.3. Dopamine beta-hydroxylase deficiency	E250	223360
12.2. Disorders in the metabolism of gamma-aminobutyrate		
12.2.1. Succinic semialdehyde dehydrogenase deficiency	E722	271980
S 4-Hydroxybutyric aciduria		
12.2.2. GABA transaminase deficiency	E728	137150
12.3. Other disorders of neurotransmitter metabolism		
13. Disorders in the metabolism of vitamins and (non-protein) cofactors		
13.1. Disorders of folate metabolism and transport		
13.1.1. Hereditary folate malabsorption	E538	229050
S SLC 46A1 deficiency		
S Proton-coupled folate transporter (PCFT) deficiency		
13.1.2. Cerebral folate deficiency due to FOLR1 deficiency	-	613068
S Neurodegeneration due to cerebral folate transport deficiency	-	
13.1.3. Methylenetetrahydrofolate reductase deficiency	E711	236250
S MTHFR deficiency		
S Homocystinuria due to deficiency of N(5,10)-		

methylenetetrahydrofolate reductase activity		
13.1.4. Other genetic disorders in folate transport and metabolism	D528	-
13.1.5. Unspecified disorders of folate transport and metabolism	D528	-
13.1.6. Secondary disorders of folate transport and metabolism	D529	-
13.1.7. Cerebral folate deficiency due to autoantibodies-non-genetic	-	-
13.2. Disorders of cobalamin absorption, transport and metabolism		
13.2.1. Intrinsic factor deficiency	D510	609342
S IFD		
S Transcobalamin III deficiency		
S TCN III, TCN3 deficiency		
13.2.2. Enterocyte intrinsic factor receptor deficiency	D511	261100
S Imerslund Gräsbeck syndrome		
S Selective malabsorption of cyanocobalamin		
S Intrinsic factor-cobalamin receptor deficiency		
S IFCR deficiency		
13.2.2.1. Intrinsic factor receptor deficiency due to *CUBN* mutations	D511	602997
S Cubulin deficiency		
13.2.2.2. Intrinsic factor receptor deficiency due to *AMN* mutations	D512	605799
13.2.3. Haptocorrin deficiency	D512	189905
S Transcobalamin I deficiency		
S TCN 1 deficiency		
S TC I, T1 deficiency		
S Vitamin B12-binding protein 1 deficiency		
S Cobalophilin deficiency		
S B12-binding alpha-globulin deficiency		
13.2.4. Transcobalamin II deficiency	D512	275350
S TCN2 deficiency		
S TC II deficiency		
S Vitamin B12-binding protein 2 deficiency		
13.2.5. Defect in adenosylcobalamin synthesis-cbl A	E711	251100
S Methylmalonic aciduria, cblA type		
S Methylmalonic acidemia, cblA type		
S Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblA type		
13.2.6. Defect in adenosylcobalamin synthesis-cbl B	E711	251110
S Methylmalonic aciduria, cblB type		
S Methylmalonic acidemia, cblB type		
S Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB type		
13.2.7. Defect in adenosylcobalamin synthesis-cblD-MMA	E728	277410
13.2.8. Defect in methylcobalamin synthesis-cblD-HC	E728	277410
13.2.9. Combined defect in adenosylcobalamin and methylcobalamin synthesis-cblC	E728	277400
S Methylmalonic aciduria and homocystinuria, cblC type		
S Methylmalonic acidemia and homocystinuria, cblC type		
S Methylmalonic aciduria and homocystinuria, vitamin B12-responsive		
13.2.10. Combined defect in adenosylcobalamin and methylcobalamin synthesis-cblD	E728	277410
S Methylmalonic aciduria and homocystinuria, cblD type		
13.2.11. Combined defect in adenosylcobalamin and methylcobalamin synthesis-cblF	E728	277380
S Methylmalonic aciduria and homocystinuria, cblF type		
S Methylmalonic acidemia and homocystinuria, cblF type		
S Lysosomal membrane cobalamin transporter deficiency		
13.2.12. Transcobalamin receptor (TCblR/CD320) defect		606475

13.2.13. Other genetic defect in cobalamin transport and metabolism	D518	-
13.2.14. Unspecified disorder of cobalamin absorption, transport and metabolism	D518	-
13.2.15. Secondary non-genetic disorders of cobalamin absorption, transport and metabolism	D518	-
13.3. Disorders of pterin metabolism	E701	
13.3.1. Guanosine 5 triphosphate cyclohydrolase I deficiency	E701	233910
13.3.2. 6-Pyruvoyl-tetrahydropterin synthase deficiency	E744	261640
13.3.3. Sepiapterin reductase deficiency	E701	612716
13.3.4. Quinoid dihydropteridine reductase deficiency	E744	261630
13.3.5. Pterin 4 carbinolamine dehydratase deficiency	E888	125310
S Primapterinuria	E888	125310
13.4. Disorders of vitamin D metabolism and transport		
13.5. Disorders of biotin metabolism		
13.5.1. Biotinidase deficiency	D818	253260
13.5.2. Holocarboxylase synthetase deficiency		253270
13.6. Disorders of pyridoxine metabolism		
13.6.1. Pyridoxine-dependent seizures		266100
S Alpha-amino adipic semialdehyde (AASA) dehydrogenase deficiency		
S Antiquitin (ALDH7A1) gene defect		
13.6.2. Pyridoxamine 5´-oxidase deficiency	E531	610090
S Pyridoxal-phosphate dependent seizures	E531	610090
13.7. Disorders of thiamine metabolism		
13.7.1. Thiamine-responsive megaloblastic anemia syndrome	E519	249270
S THTR1 deficiency (SLC19A2)	E519	249270
13.7.2. Biotin-responsive basal ganglia disease		607483
S THTR2 deficiency (SLC19A3)		607483
13.7.3. Microcephaly, Amish type		607196
S Mitochondrial thiamine pyrophosphate carrier deficiency (SLC25A19)		606521
13.8. Disorders of molybdenum cofactor metabolism		
13.8.1. Molybdenum cofactor deficiency	E798	252150
13.8.1.1. Mo cofactor deficiency, complementation group A	E798	603707
S MOCS1 deficiency	E798	603707
13.8.1.2. Mo cofactor deficiency, complementation group B	E798	603708
S MOCS2 deficiency	E798	603708
13.8.1.3. Mo cofactor deficiency, complementation group C	E798	603930
S GPHN (gephyrin) deficiency	E798	603930
13.9. Other disorders of vitamins and cofactors		
13.9.1. TTP1 deficiency	E560	277460
S Familial isolated Vitamin E deficiency	E560	277460
13.9.2. Vitamin K epoxide reductase deficiency	E561	607473
13.9.3. Retinol binding protein deficiency	E509	180250
13.9.4. Pantothenate kinases deficiency	E568	234200
S Neurodegeneration with brain iron accumulation 1	E568	234200
S Hallervorden-Spatz disease	E568	234200
14. Disorders in the metabolism of trace elements and metals		
14.1. Disorder of copper metabolism	E830	
14.1.1. Menkes syndrome	E830	309400
14.1.2. Occipital horn syndrome	Q796	304150
14.1.3. Wilson disease	E830	277900
14.2. Disorder of iron metabolism	E831	
14.2.1. Hereditary haemochromatosis		

14.2.1.1. Hereditary haemochromatosis Type 1	E831	235200
14.2.1.2. Hereditary haemochromatosis Type 2	E831	235200
14.2.1.3. Hereditary haemochromatosis Type 3	E831	235200
14.2.1.4. Hereditary haemochromatosis Type 4	E831	235200
14.2.2. Neonatal haemochromatosis	E831	
14.2.3. Haemosiderosis, acquired	E831	
14.3. Disorder of zinc metabolism	E832	
14.3.1. Acrodermatitis enteropathica	E832	201100
14.3.2. Hyperzincemia and hypercalprotectinemia	E832	194470
14.4. Disorder of phosphate, calcium and vitamin D metabolism	E835	
14.5. Disorder of magnesium metabolism	E834	
14.5.1. Hypermagnesaemia	E834	
14.5.2. Hypomagnesaemia	E834	
14.5.3. Primary hypomagnesaemia	E834	
14.5.3.1. Isolated familial renal hypomagnesaemia	E834	
14.5.3.2. Familial hypokalaemia – hypomagnesaemia	E876	
14.5.3.3. Familial hypomagnesaemia – hypercalciuria	E888	
14.5.3.4. Isolated familial intestinal hypomagnesaemia	E834	
14.5.4. Secondary hypomagnesaemia	E834	
14.5.4.1. Neonatal hypomagnesaemia	P712	307600
14.5.4.2. Hypomagnesaemic tetany in newborn	P713	
14.5.4.3. Drug induced hypomagnesaemia	E834	
14.5.5. Hypomagnesaemic tetany	E834	
14.6. Disorders in the metabolism of other trace elements and metals		
15. Disorders and variants in the metabolism of xenobiotics		
15.1. Disorders and variants of cytochrome P450-mediated oxidation		
15.2. Disorders and variants of other enzymes that oxidise xenobiotics		
15.2.1. Trimethylaminuria	E888	602079
15.3. Disorders and variants of xenobiotics conjugation		
15.4. Disorders and variants of xenobiotics transport		