IMD Scotland agreed classification of Inherited Metabolic Disorders for the Clinical Audit System (based on SSIEM classification)

Condition	Subcondition
1.1. Urea cycle disorders and inherited hyperammonaemias	
	1.1.1. Carbamoylphosphate synthetase I deficiency
	1.1.2. N-Acetylglutamate synthetase deficiency
	1.1.3. Ornithine transcarbamylase deficiency
	S Ornithine carbamoyltransferase deficiency
	1.1.4. Citrullinaemia type1
	S Argininosuccinate synthetase deficiency
	1.1.5. Argininosuccinic aciduria
	S Argininosuccinate lyase deficiency
	1.1.6. Argininaemia
	S Arginase I deficiency
	1.1.7. HHH syndrome
	S Hyperammonaemia-hyperornithinaemia-homocitrullinuria syndrome
	S Mitochondrial ornithine transporter (ORNT1) deficiency
	1.1.8. Citrullinemia Type 2
	S Aspartate glutamate carrier deficiency (SLC25A13)
	S Citrin deficiency
	1.1.9. Hyperinsulinemic hypoglycemia and hyperammonemia caused by activating mutations in the GLUD1 gene
	1.1.10. Other disorders of the urea cycle
	1.1.11. Unspecified hyperammonaemia
1.2. Organic acidurias	
	1.2.1. Glutaric aciduria - unspecified
	1.2.1.1. Glutaric aciduria type I
	S Glutaryl-CoA dehydrogenase deficiency
	1.2.1.2. Glutaric aciduria type III
	1.2.2. Propionic aciduria
	S Propionyl-CoA-Carboxylase deficiency
	1.2.3. Methylmalonic aciduria
	1.2.3.1. Methylmalonyl-CoA mutase deficiency
	1.2.3.2. Methylmalonyl-CoA epimerase deficiency
	1.2.3.3. Methylmalonic aciduria, unspecified
	1.2.4. Isovaleric aciduria
	S Isovaleryl-CoA dehydrogenase deficiency
	1.2.5. Methylcrotonylglycinuria
	S Methylcrotonyl-CoA carboxylase deficiency
	1.2.6. Methylglutaconic aciduria - unspecified
	1.2.6.1. Methylglutaconic aciduria type I
	S 3-Methylglutaconyl-CoA hydratase deficiency
	1.2.6.2. Methylglutaconic aciduria type II
	S Barth syndrome
	S Taffazin deficiency
	1.2.6.3. Methylglutaconic aciduria type III
	S Costeff syndrome
	1.2.6.4. Methylglutaconic aciduria type IV
	1.2.6.5. Methylglutaconic aciduria type V
	1.2.7. 3-Hydroxy-3-methylglutaric aciduria
	S 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency
	1.2.8. 2-Methylbutyric aciduria
	S 2-Methylbutyryl-CoA dehydrogenase deficiency
	1.2.9. 2-Methyl-3-hydroxybutyric aciduria
	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
	S Beta-ketothiolase deficiency
	S Mitochondrial acetoacetyl-CoA thiolase deficiency
	S 3-Oxothiolase deficiency
	1.2.11. Isobutyric aciduria
	S Isobutyryl-CoA dehydrogenase deficiency
	1.2.12. Methacrylic aciduria
	S 3-Hydroxyisobutyryl-CoA deacylase deficiency

1.2.13. 3-Hydroxyisobutyric aciduria
S 3-Hydroxyisobutyrate dehydrogenase
1.2.14. Methylmalonate semialdehyde dehydrogenase deficiency
1.2.15. L-2-hydroxyglutaric aciduria
S L-2-hydroxyglutarate dehydrogenase defect
1.2.16. D-2-hydroxyglutaric aciduria - unspecified
1.2.16.1. D-2-hydroxyglutarate dehydrogenase deficiency
1.2.16.2. Mitochondrial isocitrate dehydrogenase deficiency
1.2.17. Aminoacylase deficiency - unspecified
1.2.17.1. Aminoacylase 1 deficiency
1.2.17.2. Aminoacylase 2 deficiency
S Aspartoacylase deficiency
S Canavan disease
S van Bogaert-Bertrand disease
1.2.18. Methylmalonate semialdehyde dehydrogenase deficiency
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
1.3.2. Maple syrup urine disease
S Branched-chain alpha-keto acid dehydrogenase complex deficiency
S BCKD deficiency
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
S Maple syrup urine disease type II
S E2 deficiency
1.3.2.4. Unspecified BCKD deficiency
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
S Phenylketonuria
S Mild Hyperphenylalaninaemia
1.4.2. Tyrosinaemia type II
S Tyrosine aminotransferase deficiency
1.4.3. Tyrosinaemia type III
S 4-hydroxyphenylpyruvate dioxygenase deficiency
1.4.4. Hawkinsinuria
S 4-Hydroxyphenylpyruvate hydroxylase deficiency
1.4.5. Alkaptonuria
S Homogentisate 1,2 - dioxygenase deficiency
1.4.6. Tyrosinaemia type I
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
S MAT deficiency
S MAT I/III deficiency
S Hypermethioninemia, Isolated persistent
1.5.2. Glycine N-methyltransferase deficiency
S GNMT deficiency
1.5.3. S-adenosylhomocysteine hydrolase deficiency
S AHCY
S SAHH
S Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
1.5.4. Cystathionine beta-synthase deficiency
S Homocystinuria
S CBS deficiency
1.5.5. Cystathionase deficiency
S Cystathione gamma - lyase deficiency
S Gamma - cystathionase deficiency
S Cystathioninuria

1.5.6. Isolated sulfite oxidase deficiency
S Sulphite oxidase deficiency
S Sulphocysteinuria
1.5.7. Methionine synthase deficiency-cblG
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
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
1.5.10. Unspecified disorder of homocysteine metabolism
1.5.11. Unspecified disorder of methionine metabolism
1.5.12. Secondary non-genetic disorders of methionine cycle and other sulfur amino acids

1.6. Disorders of histidine, tryptophan or lysine metabolism

1.6.1. Histidinaemia
S Histidase deficiency
1.6.2. Urocanase deficiency
1.6.3. Glutamate formiminotransferase deficiency
S Forminotransferase deficiency
S Forminoglutamic aciduria
S Figlu-uria
1.6.4. Tryptophanaemia
S Tryptophan-2,3-Dioxygenase deficiency
1.6.5. Hyperlysinaemia - unspecified
S Alpha-aminoadipic semialdehyde synthase deficiency
1.6.5.1. Hyperlysinaemia type I
S Lysine : 2-oxoglutarate reductase deficiency
1.6.5.2. Hyperlysinaemia type II
S Saccharopine dehydrogenase deficiency
S Saccharopinuria
1.6.6. 2-Aminoadipic aciduria
1.6.7. 2-Oxoadipic aciduria
1.6.8. Hydroxykynureninuria
S Kynureninase deficiency
S Xanthurenic aciduria
1.6.9. Hydroxylysinuria

1.7. Disorders of serine, glycine or glycerate metabolism

1.7.1. Phosphoglycerate dehydrogenase deficiency
1.7.2. Phosphoserine phosphatase deficiency
1.7.3. Phosphoserine aminotransferase deficiency
1.7.4. Nonketotic hyperglycinaemia - unspecified
S Glycine cleavage deficiency
1.7.4.1. P protein deficiency, GLDC gene
1.7.4.2. T protein deficiency, AMT gene
1.7.4.3. H protein deficiency, GCSH gene
1.7.5. Sarcosinaemia
S Sarcosine dehydrogenase deficiency
1.7.6. D-glyceric aciduria
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
1.8.5. Cutis laxa, autosomal recessive, type IIb
S Pyrroline-5-carboxylate reductase deficiency

1.9. Disorders of amino acid transport

1.9.1. Lysinuric protein intolerance

S SLC7A7 carrier deficiency
1.9.2. Cystinuria
1.9.3. Cystinuria-hypotonia syndrome (contiguous gene defect)
1.9.4. Hartnup disease
1.9.5. Iminoglycinuria
1.9.6. Lowe syndrome
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
1.11.2. Cysteinylglycinase deficiency
1.11.3. Oxoprolinuria
S Oxoprolinase deficiency
1.11.4. Gamma-glutamylcysteine synthetase deficiency
1.11.5. Glutathione synthetase deficiency

1.12. Other disorders of peptide metabolism

1.12.1. Prolidase deficiency
S Iminodipeptiuria
1.12.2. Carnosinaemia
1.12.3. Homocarnosinosis

1.13. Other disorders of amino acid and protein metabolism

2.1. Disorders of galactose metabolism

2.1.1. Classical galactosaemia
S Galactose-1-phosphate uridyltransferase deficiency
2.1.2. Galactokinase deficiency
2.1.3. Uridine diphosphate galactose-4-epimerase deficiency

2.2. Disorders of fructose metabolism

2.2.1. Essential fructosuria
S Fructokinase deficiency
2.2.2. Hereditary fructose intolerance
S Fructose-1-phosphate aldolase deficiency

2.3. Disorders of pentose metabolism

2.3.1. Essential pentosuria
S L-xylulose reductase deficiency
2.3.2. Ribose-5-phosphate isomerase deficiency
2.3.3. Transaldolase deficiency

2.4. Disorders of glycerol metabolism

2.4.1. Glycerol kinase deficiency
2.4.2. Complex glycerol kinase deficiency due to contiguous gene deletion

2.5. Disorders of glyoxylate metabolism

2.5.1. Primary hyperoxaluria type I
S Alanine-glyoxylate aminotransferase deficiency
2.5.2. Primary hyperoxaluria type II
S Hydroxypyruvate reductase deficiency
S D-glycerate dehydrogenase deficency

2.6. Disorders of glucose transport

2.6.1. Glucose transporter 1 deficiency (blood-brain barrier)
S GLUT1 deficiency syndrome
2.6.2. Glucose transporter 2 deficiency
S Glycogen storage disease type XI
S GLUT2 deficiency syndrome
S Fanconi-Bickel syndrome
2.6.3. Glucose/galactose malabsorption
S Glucose/galactose cotransporter (SGLT1) deficiency

2.7. Disorders of gluconeogenesis

2.7.1. Fructose-1,6-bisphosphatase deficiency
2.7.2. Pyruvate carboxylase deficiency
2.7.3. Phosphoenolpyruvate carboxykinase deficiency

2.8. Glycogen storage disorders

2.8.1. Glycogen storage disease type 1a
S GSD Ia

S von Gierke disease
S Glucose-6-phosphatase deficiency
2.8.2. Glycogen storage disease type 1b
S GSD Ib
S Glucose-6-phosphate transport deficiency
2.8.3. Glycogen storage disease type II
S GSD II
S Pompe disease
S Lysosomal alpha-1,4-glucosidase deficiency
2.8.4. Glycogen storage disease type III
S GSD III
S Cori disease
S Amylo-1,6-glucosidase (debrancher) deficiency
2.8.5. Glycogen storage disease type IV
S GSD IV
S Andersen disease
S Glycogen branching enzyme deficiency
2.8.6. Glycogen storage disease type V
S GSD V
S McArdle disease
S Muscle phosphorylase deficiency
2.8.7. Glycogen storage disease type VI
S GSD VI
S Hers disease
S Hepatic glycogen phosphorylase deficiency
2.8.8. Glycogen storage disease type VII
S GSD VII
S Tauri disease
S Muscle phosphofructokinase deficiency
2.8.9. Glycogen storage disease type IX - unspecified
S GSD IX
S Phosphorylase kinase deficiency
2.8.9.1. Hepatic phosphorylase kinase deficiency
S GSD IXa
2.8.9.2. Hepatic and muscle phosphorylase kinase deficiency
S GSD Ixb
2.8.9.3. Muscle phosphorylase kinase deficiency
S GSD IXd
2.8.9.4. Cardiac muscle phosphorylase kinase deficiency
2.8.10. Glycogen storage disease type X
S GSD X
S Muscle phosphoglycerate mutase deficiency
2.8.11. Glycogen storage disease type XI
S GSD XI
S Fanconi-Bickel syndrome
S GLUT2 deficiency syndrome
2.8.12. Glycogen storage disease type XIV
S Muscle phosphoglucomutase 1 deficiency
S GSD XIV
2.8.13. Glycogen storage disease type XV
S Glycogenin deficiency
S GSD XIV
2.8.14. Glycogen storage disease type 0a
S GSD 0a
S Liver glycogen synthase deficiency
2.8.15. Glycogen storage disease type 0b
S GSD 0b
S Muscle glycogen synthase deficiency
2.8.16. Other glycogen storage disease - unspecified
2.8.16.1. Muscle LDH deficiency
2.8.16.2. Aldolase A deficiency
2.8.16.3. Beta-enolase deficiency
2.8.16.4. Muscle phosphoglycerate kinase deficiency
2.8.17. Unspecified glycogen storage disease

2.9. Other carbohydrate disorders

2.9.1. Lactose intolerance
S lactase deficiency
2.9.2. Disaccharide intolerance 1
S Sucrase-isomaltase deficiency
2.9.3. Trehalase deficiency

3.1. Disorders of lipolysis

3.2. Disorders of carnitine transport and the carnitine cycle

3.2.1. Carnitine transporter deficiency
3.2.2. Carnitine palmitoyltransferase I (CPTI) deficiency
S Carnitine uptake deficiency
3.2.3. Carnitine acylcarnitine translocase deficiency
3.2.3. Carnitine acylcarnitine translocase deficiency
3.2.4. Carnitine palmitoyltransferase II (CPTII) deficiency

3.3. Disorders of mitochondrial fatty acid oxidation

3.3.1. Very long - chain acyl CoA dehydrogenase deficiency
S VLCAD deficiency
3.3.2. Mitochondrial trifunctional protein deficiency - unspecified
3.3.2.1. Isolated deficiency of long-chain 3-hydroxyacyl-CoA dehydrogenase
S LCHAD deficiency
3.3.2.2. Isolated deficiency of long-chain 3-ketoacyl CoA thiolase
3.3.3. Medium - chain acyl CoA dehydrogenase deficiency
S MCAD deficiency
3.3.4. Short - chain acyl CoA dehydrogenase deficiency
S SCAD deficiency
3.3.5. 3-alpha-hydroxyacyl- CoA dehydrogenase deficiency
S HADH deficiency
S Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
S SCHAD deficiency
3.3.6. Multiple acyl-CoA dehydrogenase deficiency - unspecified
S Glutaric aciduria type II
3.3.6.1. Electron transfer flavoprotein deficiency, alpha chain
3.3.6.2. Electron transfer flavoprotein deficiency, beta chain
3.3.6.3. ETF-ubiquinone oxidoreductase deficiency
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
3.4.2. Succinyl-CoA:3-Oxoacid-CoA transferase (SCOT) deficiency
3.4.3. Cytosolic acetoacetyl-CoA thiolase deficiency

3.5. Other disorders of fatty acid and ketone body metabolism

3.5.1. Long - chain acyl CoA dehydrogenase deficiency
3.5.2. Malonyl CoA decarboxylase deficiency
S Malonic aciduria

4.1. Disorders of pyruvate metabolism

4.1.1.1. Pyruvate dehydrogenase E1α subunit deficiency
4.1.1.2. Pyruvate dehydrogenase E1Beta subunit deficiency
4.1.1.3. Dihydrolipoyl transacetylase deficiency
S PDHC E2 deficiency
4.1.1.4. Dihydrolipoyl dehydrogenase deficiency
S PDHC E3 deficiency
4.1.1.5. Pyruvate dehydrogenase E3 binding protein deficiency
S Protein X deficiency
4.1.1.6. Pyruvate dehydrogenase phosphatase deficiency
4.1.1.7. Pyruvate dehydrogenase deficiency, unspecified
4.1.2. Pyruvate kinase deficiency

4.2. Disorders of the citric acid cycle

4.2.1. 2-Oxoglutarate dehydrogenase deficiency
S 2-Ketoglutarate dehydrogenase complex deficiency
4.2.2. Fumarase deficiency
S Fumaric aciduria

4.3. Mitochondrial respiratory chain disorders

4.3.1. Respiratory chain disorders caused by mutations of mtDNA - unspecified
4.3.1.1. Large-scale single deletion of mtDNA
4.3.1.1.1. Pearson Syndrome
4.3.1.1.2. Kearns Sayre Syndrome
S Chronic External Ophthalmoplegia (CPEO), pigmentary degeneration of retina, myopathy and cardiomyopathy [onset before
4.3.1.1.3. Chronic Progressive External Ophthalmoplegia (CPEO) with Mitochondrial Myopathy [onset after 20 yrs]
4.3.1.2. Point mutations of mtDNA - unspecified
4.3.1.2.1. Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes, MELAS
4.3.1.2.2. Myoclonic epilepsy associated with ragged red fibres, MERRF
4.3.1.2.3. Neuropathy Ataxia and Retinitis Pigmentosa, NARP
4.3.1.2.4. Leber Hereditary Optic Neuropathy, LHON
4.3.1.2.5. Maternally Inherited Leigh Syndrome, MILS
4.3.1.2.6. Sporadic Leigh Syndrome
4.3.1.2.7. Maternally inherited Mitochondrial Dystonia
4.3.1.2.8. Maternally inherited Mitochondrial Cardiomyopathy
4.3.1.2.9. Maternally inherited Mitochondrial Myopathy - unspecified
4.3.1.2.9.1. Pure Mitochondrial Myopathy
4.3.1.2.9.2. Lethal Infantile Mitochondrial Myopathy
4.3.1.2.9.3. Mitochondrial Myopathy with Diabetes Mellitus
4.3.1.2.9.4. Mitochondrial Myopathy with Reversible cytochrome c oxidase (COX) Deficiency
4.3.1.2.10. Maternally inherited deafness and diabetes, MIDD
4.3.2. Respiratory chain disorder caused by mutations of nuclear DNA - unspecified
4.3.2.1. Mitochondrial DNA Depletion Syndrome - unspecified
4.3.2.1.1. Alpers-Huttenlocher Syndrome (POLG)
4.3.2.1.2. Hepatocerebral (DGUOK, MPV17, PEO1)
4.3.2.1.3. Myopathic (TK2)
4.3.2.1.4. Encephalomyopathy with methylmalonic aciduria (SUCLA2)
4.3.2.1.5. Fatal Infantile Lactic Acidosis with methylmalonic aciduria (SUCLG1)
4.3.2.1.6. Encephalomyopathic with renal tubulopathy (RRM2B)
4.3.2.1.7. Childhood-onset autosomal dominant optic atophy (OPA1)
4.3.2.1.8. Mitochondrial Neurogastrointestinal Encephalopathy, MNGIE (ECGF1)
4.3.2.2. Multiple mtDNA Deletion Syndrome - unspecified
4.3.2.2.1. Progressive External Ophthalmoplegia Autosomal Dominant (PEOA)
4.3.2.2.1.1. PEOA1 (POLG)
4.3.2.2.1.2. PEOA2 (ANT1)
4.3.2.2.1.3. PEOA3 (PEO1)
4.3.2.2.1.4. PEOA4 (POLG2)
4.3.2.2.1.5. PEOA5 (RRM2B)
4.3.2.2.2. Progressive External Ophthalmoplegia Autosomal Recessive (PEOB)
4.3.2.2.3. Sensory Ataxic Neuropathy, Dysarthria and Ophthalmoparesis, SANDO
S Mitochondrial Recessive Ataxic Syndrome, MIRAS (POLG)
S Spinocerebellar Ataxia with Epilepsy, SCAE
4.3.2.2.4. Optic Atrophy 1 and Deafness (OPA1)
S Optic Atrophy, Deafness, Ophthalmoplegia, Myopathy
4.3.2.3. Leigh Syndrome, LS - unspecified
S Subacute Necrotizing Encephalopathy
4.3.2.3.1. LS with leukodystrophy (SDHA, SURF1)
4.3.2.3.2. LS with cardiomyopathy (COX10, COX15)
4.3.2.3.3. LS with French-Canadian ethnicity (LRPPRC)
4.3.2.3.4. LS with nephrotic syndrome (PDSS2)
4.3.2.3.5. LS with nephropathy (COQ2)
4.3.2.4. Ubiquinone (CoQ10) deficiency (Non-LS) - unspecified
4.3.2.4.1. Early-onset ataxia with oculomotor apraxia and hypoalbuminaemia (APTX)
4.3.2.4.2. Deafness, encephaloneuropathy, obesity and valvulopathy (PDSS1)
4.3.2.4.3. Cerebellar atrophy, ataxia and seizures (CABC1)
4.3.2.5. Growth Retardation, Aminoaciduria, Cholestasis, Iron overload, Lactic acidosis and Early death (GRACILE) Syndrome (B
4.3.2.6. Renal tubulopathy, encephalopathy and liver failure (BCS1L)
4.3.2.7. Cardio-encephalopathy with hyperammonaemia (TMEM70)
S ATP synthase deficiency, nuclear-encoded
4.3.2.8. Exercise Intolerance with Lactic Acidosis - unspecified
4.3.2.8.1. Complex I deficiency; riboflavin responsive (ACAD9)
4.3.2.8.2. Complex I and II deficiency (ISCU)
4.3.2.9. Isolated Oxidative Phosphorylation Defects with Variable Phenotype (Not Classified Elsewhere) - unspecified

4.3.2.9.1. Complex I structural subunit gene defect (NDUFV1, NDUFV2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7,
4.3.2.9.2. Complex I assembly gene defect (C20orf7, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, C80orf38, NUBPL, FOXRED1)
4.3.2.9.3. Complex II structural subunit gene defect (SDHA, SDHB, SDHC,SDHD)
4.3.2.9.4. Complex II assembly gene defect (SDHAF1)
4.3.2.9.5. Complex III structural subunit gene defect (UQCRB, UQCRQ)
4.3.2.9.6. Complex III assembly gene defect
4.3.2.9.7. Complex IV structural subunit gene defect (COX6B1)
4.3.2.9.8. Complex IV assembly gene defect (SCO1, SCO2, SURF1, COX10, COX15, TACO1, FASTKD2)
4.3.2.9.9. Complex V structural subunit gene defect (ATP5E)
4.3.2.9.10. Complex V assembly gene defect (ATPAF2, TMEM70)
4.3.2.10. Mitochondrial Protein Translation Defect - unspecified
4.3.2.10.1. Combined Oxidative Phosphorylation Defect 1, COXPD1 (EFG1)
4.3.2.10.2. Combined Oxidative Phosphorylation Defect 2, COXPD2 (MRPS16)
4.3.2.10.3. Combined Oxidative Phosphorylation Defect 3, COXPD3 (TSFM)
4.3.2.10.4. Combined Oxidative Phosphorylation Defect 4, COXPD4 (TUFM)
4.3.2.10.5. Combined Oxidative Phosphorylation Defect 5, COXPD5 (MRPS22)
4.3.2.10.6. Combined Oxidative Phosphorylation Defect 6, COXPD6 (AIFM1)
4.3.2.10.7. Combined Oxidative Phosphorylation Defect 7, COXPD7 (C10ORF65)
4.3.2.10.8. Myopathy, Lactic Acidosis and Sideroblastic Anaemia 1, MLASA1 (PUS1)
4.3.2.10.9. Acute Infantile Liver Failure (TRMU)
4.3.2.10.10. Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation, LBSL (DARS2)
4.3.2.10.11. Pontocerebellar hypoplasia Type 6 (RARS2)
4.3.2.10.12. Myopathy, Lactic Acidosis and Sideroblastic Anaemia 2, MLASA2 (YARS2)
4.3.3. Respiratory chain deficiencies with no known genetic basis
4.3.3.1. Complex I deficiency
4.3.3.2. Complex II deficiency
4.3.3.3. Complex III deficiency
4.3.3.4. Complex IV deficiency
4.3.3.5. ATP synthase deficiency
4.3.3.6. Combined respiratory chain deficiency

4.4. Mitochondrial membrane transport disorders

4.4.1. Mitochondrial substrate carrier disorder - unspecified
4.4.1.1. Mitochondrial phosphate carrier deficiency (SLC25A3)
4.4.1.2. Mitochondrial aspartate glutamate carrier 1 deficiency (SLC25A12)
4.4.1.3. Mitochondrial glutamate carrier 1 deficiency (SLC25A22)
4.4.1.4. Mitochondrial carrier SLC25A38, haem biosynthesis, sideroblastic anaemia
4.4.2. Mitochondrial protein import disorder - unspecified
4.4.2.1. Mohr-Tranebjaerg syndrome (TIMM8A)

4.5. Unspecified mitochondrial disorders

4.5.1. Leigh syndrome with no known genetic or respiratory chain deficiency
4.5.2. Ethylmalonic Encephalopathy (ETHE1)
4.5.3. Anaemia, sideroblastic, and spinocerebellar ataxia, ASAT (ABCB7)

4.6. Disorders of creatine metabolism

4.6.1. Creatine transporter deficiency
S X-linked creatine deficiency syndrome
S SLC6A8 deficiency
4.6.2. Guanidinoacetate methyltransferase deficiency
4.6.3. Arginine:glycine amidinotransferase deficiency

4.7. Other disorders of energy metabolism

5.1. Disorders of purine metabolism

5.1.1. Primary idiopathic gout
5.1.2. Familial juvenile hyperuricaemic nephropathy
S Familial nephropathy with gout
5.1.3. Adenylosuccinate lyase deficiency
5.1.4. AICAR transformylase deficiency
S IMP cyclohydrolase deficiency
5.1.5. Adenosine deaminase deficiency
5.1.6. Deoxyguanosine kinase deficiency
5.1.7. Myoadenylate deaminase deficiency
5.1.8. Lesch-Nyhan syndrome
S Hypoxanthine-guanine phosphoribosyltransferase deficiency
5.1.9. Adenine phosphoribosyl transferase deficiency
5.1.10. Phosphoribosyl pyrophosphate synthetase 1 defect - unspecified

5.1.10.1. Phosphoribosyl pyrophosphate synthase superactivity
5.1.10.2. X-linked Charcot-Marie-Tooth disease-5
5.1.10.3. Arts syndrome
5.1.10.4. X-linked sensorineural deafness
5.1.11. Inosine triphosphatase deficiency
5.1.12. Adenosine deaminase superactivity
5.1.13. Purine nucleoside phosphorylase deficiency
5.1.14. Mitochondrial Ribonucelotide Reductase subunit 2 deficiency
5.1.15. Xanthinuria type I
S Xanthine oxidase deficiency
5.1.16. Xanthinuria type II
S Combined deficiency of xanthine and aldehyde oxidase
5.1.17. Thiopurine S-methyltransferase deficiency

5.2. Disorders of pyrimidine metabolism

5.2.1. Orotic aciduria type I
S Uridine monophosphate synthase deficiency
5.2.2. Orotic aciduria type II
S Orotidine - 5 -phosphate decarboxylase deficiency
5.2.3. Pyrimidine - 5 - nucleotidase deficiency
5.2.4. Dihydroorotate dehydrogenase deficiency
5.2.8. Dihydropyrimidine dehydrogenase deficiency
5.2.6. Thymidine phosphorylase deficiency
5.2.7. Thymidine kinase 2 deficiency
5.2.8. Dihydropyrimidine dehydrogenase deficiency
5.2.9. Dihydropyrimidinase deficiency
5.2.10. Beta-ureidopropionase deficiency
S Beta-alanine synthase deficiency
5.2.11. Hyper-beta-alaninaemia
S Beta-alanine-2-ketoglutarate transaminase deficiency
5.2.12. Beta-aminoisobutyrate-pyruvate transaminase deficiency

5.3. Disorders of nucleotide metabolism

5.3.1. Aicardi-Goutieres Syndrome AGS - unspecified
5.3.1.1. Aicardi-Goutieres Syndrome AGS1
S TREX1 deficiency
S DNase III deficiency
5.3.1.2. Aicardi-Goutieres Syndrome AGS2
S RNASEH2B deficiency
5.3.1.3. Aicardi-Goutieres Syndrome AGS3
S RNASEH2C deficiency
5.3.1.4. Aicardi-Goutieres Syndrome AGS4
S RNASEH2A deficiency
5.3.1.5. Aicardi-Goutieres Syndrome AGS5
S SAMHD1deficiency
5.3.2. RNASET2-deficient cystic leukoencephalopathy

6.1. Disorders of sterol biosynthesis

6.1.1. Mevalonate kinase deficiency
S Mevalonic aciduria
S Hyper-IgD syndrome (HIDS)
6.1.2. Smith - Lemli - Opitz syndrome
S 7-Dehydrocholesterol reductase deficiency
6.1.3. X-linked dominant chondrodysplasia punctata 2 (Conradi-Huenermann)
S Conradi-Hünermann syndrome
S 3β-hydroxysteroid- Δ8, Δ7-isomerase deficiency
6.1.4. Congenital hemidysplasia with ichtyosiform erythroderma and limb defects
S CHILD syndrome
S 3β-hydroxysteroid C-4 dehydrogenase deficiency
6.1.5. Desmosterolosis
S Desmosterol reductase deficiency
S 3β-hydroxysterol- Δ24-reductase deficiency
6.1.6. Lathosterolosis
S 3β-hydroxysterol Δ5-desaturase deficiency
6.1.7. Greenberg skeletal dysplasia
S Hydrops-ectopic calcification-moth-eaten skeletal dysplasia

S 3β-hydroxysterol Δ14-reductase deficiency

6.2. Disorders of bile acid biosynthesis

6.2.1. 3-Beta-hydroxysterol Delta 5-oxidoreductase isomerase deficiency
S Progressive familial intrahepatic cholestasis type 4
6.2.2. Delta 4-3-oxysterol 5Beta-reductase deficiency
6.2.3. Oxysterol 7-alpha-hydroxylase
6.2.4. Cholesterol 7-alpha-hydroxylase
6.2.5. Cerebrotendinous xanthomatosis
S Sterol 27-hydroxylase deficiency
S Van Bogaert-Scherer-Epstein disease
S Cholestanol storage disease

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
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
S Ferrochelatase deficiency
7.1.2. X-linked dominant protoporphyria
S Erythroid d 5-aminolevulinate synthase (gain of function)
7.1.3. Variegate porphyria
S Protoporphyrinogen oxidase deficiency
7.1.4. X-linked sideroblastic anaemia (XLSA)
S Erythroid 5-aminolevulinate deficiency
7.1.5. Congenital erythropoietic porphyria
S Uroporphyrinogen III synthase deficiency
7.1.6. Acute intermittent porphyria
S Porphobilinogen deaminase deficiency
7.1.7. Hereditary coproporphyria
S Coproporphyrinogen oxidase deficiency
7.1.8. Porphyria cutanea tarda type I (sporadic)
S Hepatic uroporphyrinogen decarboxylase deficiency
7.1.9. Porphyria cutanea tarda type II (familial)
S Uroporphyrinogen decarboxylase deficiency
7.1.10. Acute hepatic porphyria
S Delta aminolevilinic acid dehydratase deficiency

8.1. Inherited hypercholesterolaemias

8.1.1. Disorder of low density lipoprotein receptor - unspecified
S Fredrickson type IIa hyperlipoproteinaemia
8.1.1.1. Familial hypercholesterolaemia - homozygous
8.1.1.2. Familial hypercholesterolaemia - heterozygous
8.1.2. Sitosterolaemia
S Phytosterolaemia
S Sitosterolaemia with xanthomatosis

8.2. Inherited hypertriglyceridaemias

8.2.1. Familial chylomicronaemia - unspecified
S Hyperlipidaemia Type 1
8.2.1.1. Familial lipoprotein lipase deficiency
8.2.1.2. Familial apolipoprotein C - II deficiency
8.2.2. Familial hypertriglyceridaemia

8.3. Inherited mixed hyperlipidaemias

8.3.1. Familial dysbetalipoproteinaemia - unspecified
S Apolipoprotein E deficiency
S Fredrickson type III hyperlipoproteinaemia
S Remnant hyperlipidaemia
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
8.4.2. Tangier disease
S Familial hypoalphalipoproteinaemia
8.4.3. Lecithin cholesterol acyltransferase deficiency
8.4.3.1. Fish-eye disease
8.4.3.2. Norum disease
8.4.4. Familial hyperalphalipoproteinaemia
S Cholesterol ester transfer protein deficiency

8.5. Inherited hypolipidaemias

8.5.1. Familial abetalipoproteinaemia
S Microsomal triglyceride transfer protein (MTTP) deficiency
8.5.2. Familial hypobetalipoproteinaemia
S Apolipoprotein B deficiency
8.5.3. Anderson disease

8.6. Other disorders of lipid and lipoprotein metabolism

8.6.1.1. Sjogren - Larsson syndrome
S Fatty alcohol:NAD+ oxidoreductase deficiency
8.6.1.2. Pancreatic triacylglycerol lipase deficiency
8.6.1.3. Pancreatic colipase deficiency

8.7. Unspecified disorders of lipid and lipoprotein metabolism

9.1. Disorders of protein N-glycosylation

S CDG
9.1.1. Phosphomannomutase 2 deficiency
S PMM2-CDG
S CDG-Ia
9.1.2. Phosphomannose isomerase deficiency
S MPI-CDG
S CDG-Ib
9.1.3. Glucosyltransferase 1 deficiency
S ALG6-CDG
S CDG-Ic
9.1.4. Mannosyltransferase 6 deficiency
S NOT56L-CDG
S CDG-Id
9.1.5. Mannosyltransferase 8 deficiency
S ALG 12-CDG
S CDG-Ig
9.1.6. Glucosyltransferase 2 deficiency
S ALG 8-CDG
S CDG-Ih
9.1.7. Mannosyltransferase 2 deficiency
S ALG 2-CDG
S CDG-Ii
9.1.8. UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency
S DPAGT1-CDG
S CDG-Ij
9.1.9. Mannosyltransferase 1 deficiency
S HMT1-CDG
S CDG-Ik
9.1.10. Mannosyltransferase 7-9 deficiency
S DIBD1-CDG
S CDG-Il
9.1.11. Flippase of Man5GlcNAc2-PP-Dol deficiency
S RFT1-CDG
S CDG-In

9.1.12. N-acetylglucosaminyltransferase deficiency
S MGAT2-CDG
S CDG-IIa
9.1.13. Glucosidase 1 deficiency
S GLS1-CDG
S CDG-IIb
9.1.14. TUSC3-CDG
9.1.15. SRD5A3-CDG

9.2. Disorders of protein O-glycosylation

9.2.1. O-xylosylglycan synthesis deficiency - unspecified
9.2.1.1. EXT1 deficiency
9.2.1.2. EXT2 deficiency
9.2.1.3. Beta-1,4-galactosyltransferase 7 deficiency
S B4GALT7 deficiency
9.2.2. O-N-acetylgalactosaminylglycan synthesis deficiency - unspecified
9.2.2.1. Polypeptide N-acetylgalactosaminyl transferase deficiency
S GALTNT3 deficiency
9.2.3. O-xylosyl/N-acetylgalactosaminylglycan synthesis deficiency - unspecified
9.2.3.1. SLC35D1 deficiency
9.2.4. O-mannosylglycan synthesis deficiency - unspecified
9.2.4.1. Protein-O-mannosyltransferase 1 deficiency
S POMT1 deficiency
9.2.4.2. Protein-O-mannosyltransferase 2 deficiency
S POMT1 deficiency
9.2.4.3. Protein-O-mannose beta-1,2-N-acetyglucosaminyltransferase deficiency
S POMGNT1 deficiency
9.2.4.4. Fukutin deficiency
S FKTN deficiency
9.2.4.5. Fukutin-related protein deficiency
S FKRP deficiency
9.2.4.6. N-acetylglucosaminyltransferase-like protein deficiency
S LARGE deficiency
9.2.4.7. O-fucose-specific beta-1,3-N-acetylglucosaminyltransferase deficiency
S SCDO3 deficiency
9.2.4.8. O-fucose-specific beta-1,3-N-glucosyltransferase deficiency
S B3GALTL deficiency

9.3. Disorders of glycosphingolipid and glycosylphosphatidylinositol anchor glycosylation

9.3.1.1. Lactosylceramide alpha-2,3-sialyltransferase deficiency
S SIAT9 deficiency
9.3.1.2. Phosphatidylinositolglycan, class M deficiency
S PIGM deficiency

9.4. Disorders of multiple glycosylation and other glycosylation pathways

9.4.1. GDP-Man:Dol-P mannosyltransferase deficiency
S DPM1-CDG
S CDG-Ie
9.4.2. Lec35 deficiency
S MPDU1-CDG
S CDG-If
9.4.3. Beta-1,4-galactosyltransferase 1 deficiency
S B4GALT1-CDG
S CDG-IId
9.4.4. UDP-GlcNAc epimerase/kinase deficiency
S GNE-CDG
9.4.5. CMP-sialic acid transporter deficiency
S SLC35A1-CDG
S CDG-IIf
9.4.6. GDP-fucose transporter deficiency
S SLC35C1-CDG
S CDG-IIc
9.4.7. Dolichol pathway deficiency - unspecified
9.4.7.1. Dolichol kinase deficiency
S DK1-CDG
S CDG-Im
9.4.8. Conserved oligomeric Golgi (COG) complex deficiency - unspecified

9.4.8.1. Component of COG complex 7 deficiency
S CDG-IIe
9.4.8.2. Component of COG complex 1 deficiency
S CDG-IIg
9.4.8.3. Component of COG complex 8 deficiency
9.4.9. V-ATPase deficiency - unspecified
9.4.9.1. V0 subunit A2 of vesicular H(+)-ATPase deficiency
S ATP6VOA2-CDG
S COPII component SEC23B
S SEC23B-CDG(CDAII)

9.5. Disorders of protein ubiquitinylation

9.6. Other disorders of protein modification

10.1. Mucopolysaccharidoses

10.1.1. MPS I, Hurler, Scheie disease
S Alpha-iduronidase deficiency
10.1.2. MPS II, Hunter disease
S Iduronate 2-sulphatase deficiency
10.1.3. MPS IIIA, Sanfilippo A disease
S Heparan - N - sulphatase deficiency
10.1.4. MPS IIIB, Sanfilippo B disease
S N-acetyl-alpha-D-glucosaminidase deficiency
10.1.5. MPS IIIC, Sanfilippo C disease
S Acetyl-CoA alpha-glucosaminide acetyltransferase deficiency
10.1.6. MPS IIID, Sanfilippo D disease
S N-acetylglucosamine-6-sulphatase deficiency
10.1.7. MPS IVA, Morquio A disease
S N-acetylgalactosamine-6-sulphatase deficiency
10.1.8. MPS IVB, Morquio B disease
S Beta-galactosidase deficiency
10.1.9. MPS VI, Maroteaux - Lamy disease
S N-acetylgalactosamine - 4 - sulphatase deficiency
S Arylsulphatase B deficiency
10.1.10. MPS VII, Sly disease
S Beta-glucuronidase deficiency
10.1.11. MPS IX
S Hyaluronidase deficiency

10.2. Oligosaccharidoses

10.2.1. Aspartylglucosaminuria
S Aspartylglucosaminidase deficiency
10.2.2. Fucosidosis
S Alpha-fucosidase deficiency
10.2.3. Alpha D mannosidosis
S Alpha-mannosidase deficiency
10.2.4. Beta D mannosidosis
S Beta-mannosidase deficiency
10.2.5. Schindler disease - unspecified
S Alpha-N-acetylgalactosaminidase deficiency
10.2.5.1. Schindler disease type I
10.2.5.2. Kanzaki disease
S Schindler disease type II
10.2.6. Sialidosis
S Alpha-neuraminidase

10.3. Sphingolipidoses

10.3.1. GM1-gangliosidosis
S Beta-galactosidase deficiency
10.3.2. GM2-gangliosidosis - unspecified
10.3.2.1. GM2-gangliosidosis 0-variant,
S Sandhoff disease
S Total hexosaminidase deficiency
10.3.2.2. GM2-gangliosidosis B-variant
S Tay-Sachs disease
S Hexosaminidase A deficiency
10.3.2.3. GM2-gangliosidosis AB-variant

S GM2 activator deficiency
10.3.3. Gaucher disease
S Glucocerebrosidase deficiency
10.3.4. Krabbe disease
S Galactocerebrosidase deficiency
10.3.5. Metachromatic leukodystrophy
S Arylsulphatase A deficiency
10.3.6. Prosaposin deficiency
10.3.6.1. Saposin A deficiency
S Krabbe disease due to Saposin A deficiency
10.3.6.2. Saposin B deficiency
S Metachromatic leukodystrophy due to Saposin B deficiency
10.3.6.3. Saposin C deficiency
S Gaucher disease due to Saposin C deficiency
10.3.6.4. Saposin D deficiency
10.3.7. Fabry disease
S Alpha-galactosidase deficiency
10.3.8. Farber disease
S Ceramidase deficiency
10.3.9. Niemann-Pick disease type A or B
S Sphingomyelinase deficiency
10.3.10. Niemann-Pick disease type C
10.3.10.1. Niemann-Pick disease type C1
10.3.10.2. Niemann-Pick disease type C2

10.4. Ceroid lipfuscinoses, neuronal (CLN)

10.4.1. CLN1, Santavuori-Haltia disease
S Lysosomal palmitoyl protein thioesterase-1 deficiency
10.4.2. CLN2, Jansky-Bielschowsky disease
S Lysosomal tripeptidyl-peptidase-1 deficiency
10.4.3. CLN3, Batten Spielmeyer-Vogt disease
S Lysosomal transmembrane CLN3 protein deficiency
10.4.4. CLN4A, Kufs disease recessive type
10.4.5. CLN4B Kufs disease dominant type
10.4.6. CLN5 Finnish variant
S Lysosomal transmembrane CLN5 protein deficiency
10.4.7. CLN6
10.4.8. CLN7
S CLN Turkish variant
10.4.9. CLN8, Northern epilepsy type
10.4.10. CLN9
10.4.11. CLN10
S Cathepsin D deficiency

10.5. Lysosomal export disorders

10.5.1. Cystinosis
S Cystinosin deficiency
10.5.2. Salla disease/infantile sialic acid storage disease
S Solute carrier family 17 member 5 (SLC17A5) deficiency

10.6. Other lysosomal disorders

10.6.1. Mucolipidosis II, I-cell disease
S N-acetylglucosamine-1-phosphotransferase (alpha/beta) deficiency
10.6.2. Mucolipidosis III, Pseudo-Hurler polydystrophy
S N-acetylglucosamine-1-phosphotransferase (gamma) deficiency
10.6.3. Mucolipidosis IV
S Mucolipin-1 deficiency
10.6.4. Multiple sulphatase deficiency
S Sulphatase-modifying factor 1 (SUMF-1) deficiency
10.6.5. Wolman/cholesterol ester storage disease
S Acid lipase deficiency
10.6.6. Pompe disease, GSD type II
S Acid alpha-1,4-glucosidase deficiency
10.6.7. Sialuria
S UDP-N-acetylglucosamine 2-epimerase deficiency
10.6.8. Danon disease
S Lysosomal-associated membrane protein-2 (LAMP2) deficiency

10.6.9. Cathepsin-related disorder - unspecified
10.6.9.1. Galactosialidosis
S Lysosomal protective protein deficiency
S Cathepsin A deficiency
10.6.9.2. Cathepsin-related disorder - Papillon-Lefevre syndrome
S Cathepsin C deficiency
10.6.9.3. Pycnodysostosis
S Cathepsin K deficiency
10.6.10. Hermansky-Pudlak Syndrome

11.1. Disorders of peroxisome biogenesis

S Zellweger spectrum disorder
S Hyperpipecolic acidaemia
11.1.1. Zellweger spectrum disorder, severe form
S Zellweger syndrome
11.1.2. Zellweger spectrum disorder, attenuated form
11.1.2.1. Neonatal adrenoleukodystrophy
11.1.2.2. Infantile Refsum disease
11.1.3. Zellweger spectrum disorder, unclassified clinical severity
11.1.3.1. PEX1 deficiency
11.1.3.2. PEX2 deficiency
11.1.3.3. PEX3 deficiency
11.1.3.4. PEX5 deficiency
11.1.3.5. PEX6 deficiency
11.1.3.6. PEX10 deficiency
11.1.3.7. PEX12 deficiency
11.1.3.8. PEX13 deficiency
11.1.3.9. PEX14 deficiency
11.1.3.10. PEX16 deficiency
11.1.3.11. PEX19 deficiency
11.1.3.12. PEX26 deficiency

11.2. Rhizomelic chondrodysplasia punctata

11.2.1. Rhizomelic chondrodysplasia punctata type 1
S PTS2 receptor deficiency
S PEX7 deficiency
11.2.2. Rhizomelic chondrodysplasia punctata type 2
S Isolated dihydroxyacetone phosphate acyltransferase deficiency
11.2.3. Rhizomelic chondrodysplasia punctata type 3
S Isolated alkyl-dihydroxyacetone phosphate synthase deficiency

11.3. Disorders of peroxisomal alpha-, beta and omega-oxidation

11.3.1. X-linked adrenoleukodystrophy
S Schilder disease
S ALD
11.3.2. Peroxisomal acyl-CoA oxidase 1 deficiency
11.3.3. Peroxisomal D-bifunctional protein deficiency
S DBP deficiency
11.3.4. Sterol carrier protein deficiency
S SCPx deficiency
11.3.5. Alpha-methylacyl-CoA racemase deficiency
S AMACR deficiency
11.3.6. Refsum disease
S Phytanoyl-CoA hydroxylase deficiency

11.4. Other peroxisomal disorders

11.4.1. Primary hyperoxaluria type I
S Alanine:glyoxylate aminotransferase deficiency
11.4.2. Acatalasaemia
S Catalase deficiency

12.1. Disorders in the metabolism of biogenic amines

12.1.1. Tyrosine hydroxylase deficiency
12.1.2. Aromatic L-amino acid decarboxylase deficiency
12.1.3. Dopamine beta-hydroxylase deficiency

12.2. Disorders in the metabolism of gamma-aminobutyrate

12.2.1. Succinic semialdehyde dehydrogenase deficiency

S 4-Hydroxybutyric aciduria
12.2.2. GABA transaminase deficiency

12.3. Other disorders of neurotransmitter metabolism

13.1. Disorders of folate metabolism and transport

13.1.1. Hereditary folate malabsorption
S SLC 46A1 deficiency
S Proton-coupled folate transporter (PCFT) deficiency
13.1.2. Cerebral folate deficiency due to FOLR1 deficiency
S Neurodegeneration due to cerebral folate transport deficiency
13.1.3. Methylenetetrahydrofolate reductase deficiency
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
13.1.5. Unspecified disorders of folate transport and metabolism
13.1.6. Secondary disorders of folate transport and metabolism
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
S IFD
S Transcobalamin III deficiency
S TCN III, TCN3 deficiency
13.2.2. Enterocyte intrinsic factor receptor deficiency Imerslund Grasbeck
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
13.2.2.2. Intrinsic factor receptor deficiency due to AMN mutations
13.2.2.2. Intrinsic factor receptor deficiency due to AMN mutations
13.2.3. Haptocorrin deficiency
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
S TCN2 deficiency
S TC II deficiency
S Vitamin B12-binding protein 2 deficiency
13.2.5. Defect in adenosylcobalamin synthesis-cbl A
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
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
13.2.8. Defect in methylcobalamin synthesis-cblD-HC
13.2.9. Combined defect in adenosylcobalamin and methylcobalamin synthesis-cblC
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
S Methylmalonic aciduria and homocystinuria, cblD type
13.2.11. Combined defect in adenosylcobalamin and methylcobalamin synthesis-cblF
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
13.2.13. Other genetic defect in cobalamin transport and metabolism
13.2.14. Unspecified disorder of cobalamin absorption, transport and metabolism

13.2.15. Secondary non-genetic disorders of cobalamin absorption, transport and metabolism

13.3. Disorders of pterin metabolism

13.3.1. Guanosine 5 triphosphate cyclohydrolase I deficiency
13.3.2. 6-Pyruvoyl-tetrahydropterin synthase deficiency
13.3.3. Sepiapterin reductase deficiency
13.3.4. Quinoid dihydropteridine reductase deficiency
13.3.5. Pterin 4 carbinolamine dehydratase deficiency
S Primapterinuria

13.4. Disorders of vitamin D metabolism and transport

13.5. Disorders of biotin metabolism

13.5.1. Biotinidase deficiency
13.5.2. Holocarboxylase synthetase deficiency

13.6. Disorders of pyridoxine metabolism

13.6.1. Pyridoxine-dependent seizures
S Alpha-amino adipic semialdehyde (AASA) dehydrogenase deficiency
S Antiquitin (ALDH7A1) gene defect
13.6.2. Pyridoxamine 5-oxidase deficiency
S Pyridoxal-phosphate dependent seizures

13.7. Disorders of thiamine metabolism

13.7.1. Thiamine-responsive megaloblastic anemia syndrome
S THTR1 deficiency (SLC19A2)
13.7.2. Biotin-responsive basal ganglia disease
S THTR2 deficiency (SLC19A3)
13.7.3. Microcephaly, Amish type
S Mitochondrial thiamine pyrophosphate carrier deficiency (SLC25A19)

13.8. Disorders of molybdenum cofactor metabolism

13.8.1. Molybdenum cofactor deficiency - unspecified
13.8.1.1. Mo cofactor deficiency, complementation group A
S MOCS1 deficiency
13.8.1.2. Mo cofactor deficiency, complementation group B
S MOCS2 deficiency
13.8.1.3. Mo cofactor deficiency, complementation group C
S GPHN (gephyrin) deficiency

13.9. Other disorders of vitamins and cofactors

13.9.1. TTP1 deficiency
S Familial isolated Vitamin E deficiency
13.9.2. Vitamin K epoxide reductase deficiency
13.9.3. Retinol binding protein deficiency
13.9.4. Pantothenate kinases deficiency
S Neurodegeneration with brain iron accumulation 1
S Hallervorden-Spatz disease

14.1. Disorder of copper metabolism

14.1.1. Menkes syndrome
14.1.2. Occipital horn syndrome
14.1.3. Wilson disease

14.2. Disorder of iron metabolism

14.2.1. Hereditary haemochromatosis - unspecified
14.2.1.1. Hereditary haemochromatosis Type 1
14.2.1.2. Hereditary haemochromatosis Type 2
14.2.1.3. Hereditary haemochromatosis Type 3
14.2.1.4. Hereditary haemochromatosis Type 4
14.2.2. Neonatal haemochromatosis
14.2.3. Haemosiderosis, acquired

14.3. Disorder of zinc metabolism

14.3.1. Acrodermatitis enteropathica
14.3.2. Hyperzincemia and hypercalprotectinemia

14.4. Disorder of phosphate, calcium and vitamin D metabolism

14.5. Disorder of magnesium metabolism

14.5.1. Hypermagnesaemia
14.5.2. Hypomagnesaemia
14.5.3. Primary hypomagnesaemia - unspecified
14.5.3.1. Isolated familial renal hypomagnesaemia
14.5.3.2. Familial hypokalaemia - hypomagnesaemia

14.5.3.3. Familial hypomagnesaemia - hypercalciuria
14.5.3.4. Isolated familial intestinal hypomagnesaemia
14.5.4. Secondary hypomagnesaemia - unspecified
14.5.4.1. Neonatal hypomagnesaemia
14.5.4.2. Hypomagnesaemic tetany in newborn
14.5.4.3. Drug induced hypomagnesaemia
14.5.5. Hypomagnesaemic tetany

14.6. Disorders in the metabolism of other trace elements and metals

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
15.3. Disorders and variants of xenobiotics conjugation
15.4. Disorders and variants of xenobiotics transport
16.0 Inborn Errors otherwise unspecified