

ICD-11 revision process for Rare Diseases
Chapter IV
E70-E90

Metabolic diseases

Draft structure n° 1

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Chapter IV: Metabolic diseases

Introduction and table of contents

You are kindly invited to participate to the World Health Organisation's *International Classification of Diseases* (ICD) revision process. The following document will help you in making your comments. You will find:

1. The rationale and the general methodology of the ICD revision for rare diseases
2. The ICD-11 draft structure for *Metabolic diseases*, which represents Orphanet's proposal for a new ICD.
3. Groups of diseases classified as metabolic in ICD10 that need to be moved in ICD11.

You are invited to:

1. Comment the ICD-11 draft structure indicating whether you agree or disagree with the new global structure and the way rare diseases are represented in it.

2. Send your feed-back to Orphanet.

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3. Disseminate this invitation to your colleagues who are experts in this field.

1. Rationale and general methodology

WHO has established various Topic Advisory Groups to serve as planning and coordinating advisory bodies in the update and revision process for specific areas. A Revision Steering Group oversees the overall revision process. An internet-based workspace documents revision proposals that obtain evidence from analysis of available data. WHO collaborates through this platform with all interested parties. Working groups organized by the Topic Advisory Groups (TAG) review the proposals. To learn more about the whole revision process:

<https://extranet.who.int/icdrevision/help/docs/ICDRevision.pdf>

A TAG for rare diseases was established in April 2007 as rare diseases should now be traceable in mortality and morbidity information systems. The production of the basic information to establish a first draft of the classification of rare diseases has been assigned to Orphanet and will contribute to the whole revision process, as rare diseases involve all areas of medicine.

The workplan is as follows:

1. Proposals from TAG for a new ICD structure are expected before the end of 2009.
2. A decision about it, after compilation of all proposals, should be taken in April 2010. This will define the category layers based on consensus hierarchies, called the Alpha version.
3. Work on the Beta version will then start at TAG level to populate the model and finalise the proposals. The Beta version will be due by the end of 2010. In 2011 field testing will start.

In order to prepare the ICD revision process, Orphanet has collected a series of rare diseases classifications mainly based on scientific grounds (aetiology and mechanism). To complement these classifications, Orphanet has developed a strictly clinical in-house classification to meet the needs of the clinicians. All the classifications can be viewed on the Orphanet website. They now serve as a basis to build the ICD-11 proposals. For an overview on the general methodology of Orphanet classification:

<http://www.orpha.net/data/patho/Pro/en/OrphanetClassificationRareDiseases.pdf>

Orphanet is a comprehensive peer-reviewed database of information on rare diseases. Over 5,800 are inventoried, and the database of diseases is updated monthly according to the evolution of knowledge. Each Orphanet entry is indexed with MeSH terms, Orphanet thesaurus of clinical signs and symptoms, ICD-10 codes, and linked to the OMIM database, to an in-house genes database and to PubMed as well as to other websites of interest. For each Orphanet entry there is an identity card with epidemiological data (prevalence rank, mode of inheritance, age of onset) and a set of synonyms. Orphanet produces a peer-reviewed encyclopaedia covering more than 2,600 entries and updated continuously.

2. Rationale for metabolic diseases revision

The current ICD10 classification of metabolic diseases is currently made of the following block of codes, which are a subset of current ICD chapter IV *Endocrine, nutritional and metabolic diseases*:

[E70-E90](#) Metabolic disorders

The current ICD classification is monoaxial, meaning that every entity can figure only at one point in the classification; however, many diseases are associated with more than one medical specialty. In the future ICD, every entity shall be assigned a unique identifying number, which will allow the classification to become *polyaxial*: diseases will be able to figure in all relevant places in the classification (for instance, McArdle disease will figure among both metabolic and muscular diseases). This system will be fully operational in the electronic version of the future ICD. However, in the paper version, it will still be necessary for space reasons to keep the current monoaxial system: one medical specialty must then be given priority, and *exclusion notes* are put in the other relevant chapters to redirect users to the correct code. The priority specialty is related to the body system most severely affected by the disease and/or the specialist most likely to be relied on for the management of the disease. In a number of cases however, the choice is questionable and ultimately quite arbitrary. For multisystemic diseases, Orphanet supports the creation of a new dedicated chapter in ICD11.

The Orphanet proposal for a new ICD classification follows a clinical rather than aetiological approach. Groups of diseases are preferentially defined on the basis of shared clinical features. In the monoaxial ICD paper version, metabolic diseases primarily associated with one particular system will be coded in the chapter for the relevant system, rather than in the chapter for metabolic diseases.

Disorders of neurotransmitter metabolism are not dealt with in this document : they will be dealt with in Orphanet's revision proposal for the neurology chapter.

Some disease groups classified as "metabolic" in ICD10 are not usually reckoned as such (for instance, ICD10 includes cystic fibrosis among metabolic diseases). We suggest to move them out of this chapter; they are given for reference at the end of this document.

ICD-11 draft structure for Metabolic diseases

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Chapter IV: Nutritional diseases

ICD-11 draft structure for metabolic diseases

NB: Main sections are highlighted in grey.

NB: Synonyms are in italics

NB: Appropriate classification for excluded entries will be revised in the proper chapters. The corresponding ICD10 codes are indicated when existent.

<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Metabolic diseases by endogen intoxication		
Disorders of amino-acid or proteins metabolism		
Disorders of phenylalanine or tyrosine metabolism		
Phenylketonuria		Intermediate levels are biochemically defined from aminoacid metabolic pathways. They are kept so as to avoid lumping together all disorders of protidic metabolism and thus increase clarity in understanding the group as a whole, but may be not clinically relevant for coding. Deleting them can be envisioned.
Classical phenylketonuria	E70.0	
<i>Phenylalanine hydroxylase total deficiency</i>		
<i>Typical phenylketonuria</i>		
Nonclassical phenylketonuria		
<i>Phenylalanine hydroxylase partial deficiency</i>		
<i>Atypical phenylketonuria</i>		
Mild hyperphenylalaninaemia	E70.1	
Excludes: 6-pyruvoyl-tetrahydropterin synthase deficiency		
Maternal hyperphenylalaninaemia	E70.1	
Tyrosinaemia, type 1	E70.2	The various types of tyrosinaemia do not form a clinically distinct group of diseases, and should be distinguished systematically. Unqualified "tyrosinaemia" should either be removed or redirected to the form 1.
<i>Hepatorenal tyrosinaemia</i>		
<i>Tyrosinaemia</i>		
<i>Hypertyrosinaemia</i>		
<i>Tyrosinosis</i>		
Tyrosinaemia, type 2	E70.2	
<i>Oculocutaneous tyrosinaemia</i>		
Tyrosinaemia, type 3	E70.2	
Alkaptonuria	E70.2	
<i>Ochronosis</i>		
Hawkinsinuria		Include in dermatology.
Tyrosine oxidase temporary deficiency		
Excludes: Albinism	E70.3	
Disorders of histidine metabolism	E70.8	
Histidinaemia		
Formiminoglutamic aciduria		
Disorders of tryptophan or lysine metabolism	E70.8	
Hypertryptophanaemia		
Hyperlysinaemia		
Glutaryl-CoA dehydrogenase deficiency		

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ICD11 table draft	Corresponding ICD10 code	Comments
2-aminoadipic aciduria		
Encephalopathy due to hydroxykynureninuria		
Seizures - intellectual deficit due to hydroxylysinuria		
Saccharopinuria		
Pellagra-like skin rash – neurological manifestations		
Disorders of leucine, isoleucine or valine metabolism		
Leucinosi	E71.0	
<i>Maple-syrup-urine disease</i>		
3-hydroxy-3-methylglutaric aciduria		
Disorders of cystein or methionin metabolism	E72.1	
Cystathioninuria		
Classic homocystinuria		
Methioninaemia		
Sulfite oxidase deficiency		
Isolated sulfite oxidase deficiency		
Sulfite oxidase deficiency due to molybdenum cofactor deficiency		
N5-methylhomocysteine transferase deficiency		
Excludes: Transcobalamin II deficiency	D51.2	
Disorders of ornithine or proline metabolism	E72.4	
Hyperornithinaemia		
<i>Ornithinaemia</i>		
Hyperprolinaemia type I		
Hyperprolinaemia type II		
Disorders of serine or glycine metabolism	E72.5	
Non-ketotic hyperglycinaemia		
Isolated nonketotic hyperglycinaemia type 1		
Isolated nonketotic hyperglycinaemia type 2		
Sarcosinaemia		
Neurometabolic disorder due to serine deficiency		
3-Phosphoserine phosphatase deficiency		
3-Phosphoglycerate dehydrogenase deficiency		
Disorders of amino-acid transport	E72.0	
Hartnup's disease		
Hyperdibasic aminoaciduria type 1		
Lysinuric protein intolerance		
<i>Hyperdibasic aminoaciduria type 2</i>		
Iminoglycinuria		
Excludes: Cystinuria		Include in nephrology.

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Organic acidurias		Disorders of intermediary metabolism with characteristic accumulation of carboxylic acids in urine. Classical forms are systemic, while cerebral form are dominated by cerebral symptoms with less prominent laboratory abnormalities.
Classical organic acidurias		
Methylmalonic aciduria - homocystinuria		
Methylmalonicacidaemia - homocystinuria, type cbl C		
Methylmalonicacidaemia - homocystinuria, type cbl D		
Methylmalonicacidaemia - homocystinuria, type cbl F		
Methylmalonic aciduria, vitamin B12 unresponsive		
Methylmalonic aciduria, vitamin B12 responsive		
Vitamin B12 responsive methylmalonic acidaemia, type cbl A		
Vitamin B12 responsive methylmalonic acidaemia, type cbl B		
Vitamin B12 responsive methylmalonic acidaemia, type mut-		
Propionic aciduria		
Isovaleric aciduria		
Multiple carboxylase deficiency		
Multiple carboxylase deficiency, due to biotinidase deficiency		
Multiple carboxylase deficiency, due to holocarboxylase synthetase deficiency		
Ketoacidosis due to betaketothiolase deficiency		
3 hydroxyisobutyric aciduria		
3-hydroxy-3-methylglutaric aciduria		
3-methylcrotonylglycinuria		
3-methylglutaconic aciduria		
MGA		
3-methylglutaconic aciduria, type 1		
3-methylglutaconic aciduria, type 3		
<i>Costeff syndrome</i>		
3-methylglutaconic aciduria, type 4		
Excludes: Barth syndrome (= 3-methylglutaconic aciduria, type 2)		Include below in mitochondrial diseases.
2-methylbutyryl-CoA dehydrogenase deficiency		
Isobutyryl-CoA dehydrogenase deficiency		
Cerebral organic aciduria		
Glutaryl-CoA dehydrogenase deficiency		
2-hydroxyglutaricaciduria		
L-2-hydroxyglutaricaciduria		
D-2-hydroxyglutaricaciduria		
Canavan disease		
Malonic aciduria		
Ethylmalonic aciduria encephalopathy		
Excludes: 4-Hydroxybutyric aciduria		Include in neurology.

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Disorders of urea cycle metabolism	E72.2	
Argininaemia		
Hyperargininaemia		
Arginase deficiency		
Argininosuccinic aciduria		
Carbamoylphosphate synthetase deficiency		
Citrullinaemia		
Triple H (HHH) syndrome		
Hyperammonaemia due to N-acetylglutamate synthetase deficiency		
Progressive neurodegeneration - joint laxity - cataract		
<i>Excludes:</i> Disorders of ornithine metabolism	E72.4	Include in disorders of ornithine and proline metabolism (above).
<i>Excludes:</i> Lysinuric protein intolerance		Include in disorders of aminoacid transport (above).
Gamma-glutamyl cycle disorder		
Glutathione synthetase deficiency		
5-oxoprolinase deficiency		
Gamma-glutamyl transpeptidase deficiency		
Gamma-glutamylcysteine synthetase deficiency		
Peptide metabolism disorder		
Carnosinaemia		
Prolidase deficiency		
Homocarnosinosis		
Porphyria		
Porphyria, congenital erythropoietic	E80.0	
Protoporphyria, erythropoietic		
Porphyria, acute hepatic		
Coproporphyria, hereditary		
Porphyria, acute intermittent	E80.2	
Porphyria variegata		
Porphyria of Doss		
Porphyria, chronic hepatic		
Hepatoerythropoietic porphyria		
Porphyria cutanea tarda	E80.1	
Other specified metabolic diseases by endogen intoxication		
French type sialuria		
Trimethylaminuria		

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
<p>Excludes: Lafora disease Excludes: Hyperoxaluria</p>		<p>Include in neurology as epilepsy. Include in nephrology.</p>
Energy metabolism disorder		
<p>Disorders of fatty-acid metabolism</p> <p>Excludes: Schilder's disease</p> <p>Muscle carnitine palmitoyltransferase deficiency Carnitine palmitoyl transferase 1 deficiency Carnitine palmitoyl transferase 2 deficiency</p> <p>Mitochondrial trifunctional protein deficiency</p> <p>Carnitine uptake deficiency</p> <p>Carnitine-acylcarnitine translocase deficiency</p> <p>Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency</p> <p>Medium chain acyl-CoA dehydrogenase deficiency</p> <p>Multiple FAD dehydrogenase deficiency</p> <p>SCAD deficiency</p> <p>Acyl-CoA dehydrogenase, very long chain, deficiency of Acyl-CoA dehydrogenase, long chain, deficiency of Acyl-CoA dehydrogenase 9 deficiency</p> <p>3-hydroxyacyl-CoA dehydrogenase, short chain, deficiency of</p> <p>3-hydroxy 3-methylglutaryl-CoA (HMG) synthase deficiency</p>	<p>E71.3 G37.0</p>	<p>Include in neurology.</p>
<p>Mitochondrial respiratory chain disorders</p> <p>Kearns-Sayre syndrome</p> <p>MELAS syndrome</p> <p>MERRF syndrome</p> <p>NARP/MILS syndrome</p> <p>Pearson syndrome</p> <p>Mitochondrial disease of nuclear origin</p> <p>NADH-CoQ reductase deficiency</p> <p>Cataract cardiomyopathy</p> <p>Leigh syndrome Leigh syndrome due to cytochrome c oxidase deficiency</p> <p>Proximal tubulopathy - diabetes mellitus - cerebellar ataxia</p> <p>Lactic acidosis, congenital</p> <p>Barth syndrome</p> <p>Oxoglutaricaciduria</p> <p>Fumaric aciduria</p>	<p>H49.8</p>	

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ICD11 table draft	Corresponding ICD10 code	Comments
Pyruvate dehydrogenase deficiency E1 deficiency E2 deficiency E3 (lipoamide oxydoreductase) deficiency PDH phosphatase deficiency Pyruvate decarboxylase deficiency	E74.4	
Succinate CoQ reductase deficiency Coenzyme Q cytochrome c reductase deficiency ATP synthetase deficiency Myoneurogastrointestinal encephalopathy syndrome Coenzyme Q 10 (CoQ10), deficiency Mitochondrial ADN deletions and duplications Mitochondrial DNA depletion syndrome GRACILE syndrome Saguenay-Lac-St. Jean cytochrome oxidase deficiency Ataxia - leukodystrophy - tubulopathy, due to cytochrome c oxidase deficiency Cardiomyopathy - hypotonia, due to cytochrome c oxidase deficiency Cardiomyopathy - hypotonia - lactic acidosis Leukoencephalopathy with brain stem, spinal cord involvement - lactate elevation Hypotonia with lactic acidaemia and hyperammonaemia Deletion 2p21 (Hypotonia-cystinuria syndrome) Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3 Zellweger-like syndrome without peroxisomal anomalies Alpers syndrome <i>Excludes:</i> Leber hereditary optic neuropathy <i>Excludes:</i> Leber 'plus' disease <i>Excludes:</i> Wolfram syndrome <i>Excludes:</i> Progressive external ophthalmoplegia		Include in ophthalmology. Include in ophthalmology. Include in endocrinology. Include in ophthalmology.
Creatine biosynthesis disorder Guanidinoacetate methyltransferase deficiency Arginine:glycine amidinotransferase deficiency Intellectual deficit, X-linked, with seizures, short stature and midface hypoplasia		
Carbohydrate metabolism disorder Glycogen storage disease Glycogen storage disease due to glycogen synthase deficiency (type 0) Glycogen storage disease due to glycogen synthase deficiency, hepatic form Glycogen storage disease due to glycogen synthase deficiency, muscular form	E74 E74.0	The naming of glycogenosis either uses numbers or eponyms. The numbering is confusing in a few cases (notably glycogen storage disease type 6) and not all entities have an eponym in use. Therefore we chose to name entities after the enzymatic defect responsible for the disease. Numbers and eponyms are recalled in parentheses.

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ICD11 table draft	Corresponding ICD10 code	Comments
Glycogen storage disease due to glucose-6-phosphate system deficiency (type 1, Von Gierke disease)		
Glycogen storage disease due to glucose-6-phosphatase deficiency (type 1A)		
Glycogen storage disease due to glucose-6-phosphate translocase deficiency		
Glycogen storage disease type 1B		
Glycogen storage disease type 1C		
Glycogen storage disease type 1D		
Glycogen storage disease due to acid maltase deficiency (type 2 - Pompe disease)		
Glycogen storage disease due to LAMP-2 deficiency (type 2B - Danon disease)		
Glycogen storage disease due to glycogen debranching enzyme deficiency (type 3 - Cori-Forbes disease)		
Glycogen storage disease due to glycogen branching enzyme deficiency (type 4 - Andersen disease - amylopectinosis)		
Glycogen storage disease due to muscle phosphorylase deficiency (type 5 - McArdle disease)		
Glycogen storage disease due to liver phosphorylase deficiency (type 6 or 6B - Hers disease)		
Glycogen storage disease due to muscle phosphofructokinase deficiency (type 7 - Tarui disease)		
Glycogen storage disease due to phosphorylase kinase deficiency (type 6A or 8 or 9)		
Glycogen storage disease due to phosphoglycerate mutase deficiency (type 10 -Di Mauro disease)		
Glycogen storage disease due to GLUT2 deficiency (type 11 - Bickel-Fanconi)		
Glycogen storage disease due to aldolase A deficiency (type 12)		
Glycogen storage disease due to enolase beta deficiency (type 13)		
Glycogen storage disease due to phosphoglucomutase 1 deficiency (type 14)		
Disorders of fructose metabolism	E74.1	
Essential fructosuria		
Hereditary fructose intolerance		
Disorders of galactose metabolism	E74.2	
Galactosaemia		
Galactokinase deficiency		
UDP-galactose-4-epimerase deficiency		
Galactose-1-phosphate uridylyltransferase deficiency		
Gluconeogenesis disorder	E74.4	
Fructose-1,6-bisphosphatase deficiency		
Phosphoenolpyruvate carboxykinase (PEPCK) deficiency		
Phosphoenolpyruvate carboxykinase 1 (Pepck1) deficiency		
Phosphoenolpyruvate carboxykinase 2 (Pepck2) deficiency		
Pyruvate carboxylase deficiency		
Ketolysis disorder		
Ketoacidosis due to betaketothiolase deficiency		
Succinyl-CoA acetoacetate transferase deficiency		

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Other specified energy metabolism disorders		
Triose phosphate-isomerase deficiency		
Multiple carboxylase deficiency		
Multiple carboxylase deficiency, due to biotinidase deficiency		
Multiple carboxylase deficiency, due to holocarboxylase synthetase deficiency		
Phosphoglycerate kinase 1 deficiency		
Succinic acidaemia		
Glucosephosphate isomerase deficiency		
Diphosphoglycerate mutase deficiency of erythrocyte		
Dicarboxylicaminoaciduria		
D-glycericacidaemia		
Fatal infantile cytochrome C oxidase deficiency		
Enolase deficiency		
Enolase deficiency, type 1		
Enolase deficiency, type 2		
Enolase deficiency, type 3		
Enolase deficiency, type 4		
Lactate dehydrogenase deficiency		
Phosphoglucomutase deficiency		
Phosphoglucomutase deficiency type 1		
Phosphoglucomutase deficiency type 2		
Phosphoglucomutase deficiency type 3		
Phosphoglucomutase deficiency type 4		
Butyrylcholinesterase deficiency		
Lipoamide dehydrogenase deficiency		
Myoneurogastrointestinal encephalopathy syndrome		
Glutaryl-CoA oxidase deficiency		
Hyperinsulinism-hyperammonaemia syndrome		
3-hydroxylacyl-CoA dehydrogenase deficiency		
3-hydroxyacyl-CoA dehydrogenase, short chain, deficiency of		
Excludes: Chondrodysplasia punctata lethal, neonatal		Include in skeletal diseases.
Excludes: Anaemia due to adenosine triphosphatase deficiency		Include in haematology.
Excludes: Haemolytic anaemia due to red cell pyruvate kinase deficiency		Include in haematology.
Excludes: Anaemia due to adenosine triphosphatase deficiency		Include in haematology.

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Defects in synthesis or catabolism of complex molecules		
Peroxisomal disease		
Refsum disease		
Refsum disease, infantile form		
Zellweger syndrome		
Adrenoleukodystrophy, neonatal		
Pseudoadrenoleukodystrophy		
Pseudo-Zellweger syndrome		
Bifunctional enzyme deficiency		
Acatalasaemia		
Pipecolic acidaemia		
<i>Excludes:</i> Chondrodysplasia punctata, rhizomelic type		Include in skeletal diseases.
<i>Excludes:</i> Bile acid synthesis defect, congenital, type 4		Include in hepatology.
Lysosomal disease		
<i>Excludes:</i> Glycogen storage disease due to LAMP-2 deficiency		Include above among glycogen storage diseases.
<i>Excludes:</i> Neuronal ceroid lipofuscinosis		Include in neurology.
Progressive epilepsy-intellectual deficit, Finnish type		
Adult neuronal ceroid lipofuscinosis		
Infantile neuronal ceroid lipofuscinosis		
<i>ICD11 table draft</i>		
Juvenile neuronal ceroid lipofuscinosis		
Congenital neuronal ceroid lipofuscinosis		
Late infantile neuronal ceroid lipofuscinosis		
<i>Corresponding ICD10 code</i>		
Lipid storage disease	E75.5	<i>Comments</i>
Niemann-Pick disease, type C		
Niemann-Pick C1 disease		
Niemann-Pick C2 disease		
Niemann-Pick disease, Nova Scotia type		
Niemann-Pick D disease		
Wolman disease		
Cholesteryl ester storage disease		
Lysosomal transport defect		
Free sialic acid storage disease		
<i>Excludes:</i> Cystinosis		Include in nephrology.

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Mucopolysaccharidosis	E76	
Mucopolysaccharidosis type 1	E76.0	
Hurler syndrome		
Scheie syndrome		
Hurler-Scheie syndrome		
Mucopolysaccharidosis type 2 (Hunter syndrome)	E76.1	
Mucopolysaccharidosis type 3 (Sanfilippo syndrome)	E76.2	
Mucopolysaccharidosis, type 3A (Sanfilippo syndrome, type A)		
Mucopolysaccharidosis, type 3B (Sanfilippo syndrome, type B)		
Mucopolysaccharidosis, type 3C (Sanfilippo syndrome, type C)		
Mucopolysaccharidosis, type 3D (Sanfilippo syndrome, type D)		
Mucopolysaccharidosis type 4 (Morquio syndrome)	E76.2	
Mucopolysaccharidosis type 6 (Maroteaux-Lamy syndrome)	E76.2	
Mucopolysaccharidosis type 7 (Sly syndrome)	E76.2	
Mucopolysaccharidosis type 9 (Natowicz syndrome)		
Oligosaccharidosis	E77.1	
Alpha-mannosidosis		
Aspartylglucosaminuria		
Beta-mannosidosis		
Fucosidosis		
Sialidosis		
<i>Lipomucopolysaccharidosis</i>		
<i>Mucopolipidosis type 1</i>		
Sialidosis type 1		
Sialidosis, type 2		
N-acetyl-alpha-D-galactosaminidase deficiency		
Alpha-N-acetylgalactosaminidase deficiency, type 1		
Alpha-N-acetylgalactosaminidase deficiency, type 2		
Alpha-N-acetylgalactosaminidase deficiency, type 3		
Neuroaxonal dystrophy, Schindler type		
Mucopolipidosis	E77.0	
Mucopolipidosis type 2		
Mucopolipidosis type 3		
Mucopolipidosis type 4		
Excludes: Sialidosis (mucopolipidosis type 1)		Mucopolipidosis type 1 is a former name for sialidosis.
Sphingolipidosis	E75	
Mucosulfatidosis		
Farber lipogranulomatosis		
Krabbe disease		

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ICD11 table draft	Corresponding ICD10 code	Comments
Sandhoff disease		
Fabry disease		
Metachromatic leukodystrophy		
Galactosialidosis		
GM1 gangliosidosis		
Gangliosidosis GM1, type 1		
Gangliosidosis GM1, type 2		
Gangliosidosis GM1, type 3		
Gaucher disease		
Gaucher disease - ophthalmoplegia - cardiovascular calcification		
Gaucher disease, type 1		
Gaucher disease, type 2		
Gaucher disease, type 3		
Perinatal-lethal Gaucher disease		
Tay-Sachs disease		
Niemann-Pick disease, type A		
Niemann-Pick disease, type B		
Niemann-Pick disease, type E		
Encephalopathy due to prosaposin deficiency		
Acid phosphatase deficiency		
Protein glycosylation disorder		
CDG syndrome		
CDG syndrome, type Ia		
CDG syndrome, type Ib		
CDG syndrome, type Ic		
CDG syndrome, type Id		
CDG syndrome, type Ie		
CDG syndrome, type If		
CDG syndrome, type Ig		
CDG syndrome, type Ih		
CDG syndrome, type Ii		
CDG syndrome, type Ik		
CDG syndrome, type IL		
CDG syndrome, type lia		
CDG syndrome, type lib		
CDG syndrome, type lid		
CDG syndrome, type lie		
CDG syndrome, type Ij		

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ICD11 table draft	Corresponding ICD10 code	Comments
<p>CDG syndrome, type Im CDG syndrome, type lih Excludes : CDG syndrome, type Ilc (Leukocyte adhesion deficiency type 2)</p>		Include in immunology.
Sterol metabolism disorder		
Anomaly of bile acid synthesis		
Xanthomatosis cerebrotendinous		Include in hepatology.
Excludes : Bile acid synthesis defect with cholestasis and malabsorption		Include in hepatology.
Bile acid synthesis defect, congenital, type 1		Include in hepatology.
Bile acid synthesis defect, congenital, type 3		Include in hepatology.
Bile acid synthesis defect, congenital, type 2		Include in hepatology.
Sterol biosynthesis disorder		
Mevalonicaciduria		
Desmosterolosis		
Lathosterolosis		
CHILD syndrome		
Excludes : Antley-Bixler syndrome		Include in skeletal diseases.
Excludes : Greenberg dysplasia		Include in skeletal diseases.
Excludes : Smith-Lemli-Opitz syndrome		Include in developmental abnormalities.
Excludes : X-linked dominant chondrodysplasia punctata		Include in skeletal diseases.
Excludes : Partial mevalonate kinase deficiency with recurrent fever +/- hyperIgD		Include in internal medicine.
Excludes : Hyperimmunoglobinaemia D with recurrent fever		Include in internal medicine.
Purine or pyrimidine metabolism disorder		
Purine metabolism disorder		
Lesch-Nyhan syndrome	E79	
Kelley-Seegmiller syndrome		
Adenosine monophosphate deaminase deficiency		
Purine nucleoside phosphorylase deficiency		
Adenylosuccinate lyase deficiency		
Phosphoribosylpyrophosphate synthetase superactivity		
Excludes : Xanthinuria		Include in nephrology.
Xanthinuria, type I		Include in nephrology.
Xanthinuria, type II		Include in nephrology.
Xanthinuria, hereditary isolated		Include in nephrology.
Xanthine oxidoreductase (XOR), isolated deficiency		Include in nephrology.
Excludes : 2,8 dihydroxyadenine urolithiasis		Include in nephrology.

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Pyrimidine metabolism disorder Orotic aciduria hereditary Dihydropyrimidine dehydrogenase deficiency Myoneurogastrointestinal encephalopathy syndrome Dihydropyrimidinuria Beta-ureidopropionase deficiency Excludes: Anaemia due to pyrimidine 5' nucleotidase deficiency		Include in haematology.
Disorders of mineral metabolism	E83	
Excludes: Dietary mineral deficiency	E58-E61	Include in nutrition.
Excludes: Parathyroid disorders	E20-E21	Include in endocrinology.
Excludes: Vitamin D deficiency	E55	Include in nutrition.
Excludes: Disorders of iron metabolism, including:		Most disorders due to iron metabolism are best classified as haematologic or hepatologic, sometimes neurologic. They are nevertheless to be mentioned here to provide coders with appropriate redirections.
Iron overload disease		
Haemochromatosis	E83.1	Include in hepatology.
Ferroportin disease		Include in hepatology.
Neonatal haemochromatosis		Include in hepatology.
African iron overload		Include in hepatology.
Friedreich ataxia		Include in neurology
Neurodegeneration with brain iron accumulation		Include in neurology
Aceruloplasminaemia		Include in neurology
Atransferrinaemia		Include in haematology.
Microcytic anaemia with liver iron overload		Include in haematology.
Iron deficiency anaemias	D50	Include in haematology.
Sideroblastic anaemias	D64.0-D64.3	Include in haematology.
Excludes: Non-endocrine disorders of phosphocalcic metabolism		Disorders of phosphocalcic metabolism are difficult to classify as a group. Many are related to parathyroid disorders; non-endocrine entities are to be classified in skeletal or renal disorders.
Non-endocrine disorders of phosphorus metabolism, including:	E83.3	
Familial hypophosphataemia		Include with skeletal abnormalities.
Hypophosphatasia		Include with skeletal abnormalities.
Vitamin-D-resistant osteomalacia		Include with skeletal abnormalities.
Vitamin-D-resistant rickets		Include with skeletal abnormalities.
adult osteomalacia	M83	Include with skeletal abnormalities.
osteoporosis	M80-M81	Include with skeletal abnormalities.

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ICD11 table draft	Corresponding ICD10 code	Comments
Non-endocrine disorders of calcium metabolism	E83.5	
Familial hypocalciuric hypercalcaemia		Include in nephrology.
Idiopathic hypercalciuria		Include in nephrology.
chondrocalcinosis	M11.1-M11.2	Include with skeletal abnormalities.
adult osteomalacia	M83	Include with skeletal abnormalities.
osteoporosis	M80-M81	Include with skeletal abnormalities.
Disorders of copper metabolism	E83.0	
Menkes disease		
<i>Kinky hair disease</i>		
<i>Steely hair disease</i>		
Occipital horn syndrome		
Wilson disease		
Benign familial copper deficiency		
Disorders of zinc metabolism	E83.2	
Acrodermatitis enteropathica		
Disorders of magnesium metabolism	E83.4	
Hypermagnesaemia		
Hypomagnesaemia		
Other specified disorders of mineral metabolism	E83.8	
Disorders of lipoprotein metabolism and other lipidaemias		
	E78	
Hyperlipidaemia		
Pure hypercholesterolaemia	E78.0	
Hypercholesterolaemia, familial		
Defective apolipoprotein B-100, familial		
Pure hyperglyceridaemia	E78.1	
Major hypertriglyceridaemia		
Hyperlipoproteinaemia type 1	E78.2	
Hyperlipoproteinaemia type 4		
Hyperlipoproteinaemia type 5		
Mixed hyperlipidaemia		
Hyperlipidaemia type 3		
Hyperlipidaemia familial combined		

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Hyperalphalipoproteinaemia		
Hyperalphalipoproteinaemia, familial		
Hyperlipidaemia due to hepatic triglyceride lipase deficiency		
Hyperlipidaemia due to cubilin deficiency		
Other specified hyperlipidaemia	E78.4	
Hypolipidaemia	E78.6	
<i>Lipoprotein deficiency</i>		
Hypoalphalipoproteinaemia		
LCAT deficiency		
Tangier disease		
Hypoalphalipoproteinaemia, familial		
Hypobetalipoproteinaemia		
Hypobetalipoproteinaemia, familial form		
Benign familial hypobetalipoproteinaemia		
Abetalipoproteinaemia / Homozygous familial hypobetalipoproteinaemia		
Chylomicron retention disease		
Other specified disorders of lipoprotein metabolism	E78.8	
Phytosterolaemia		
Excludes: Xanthomatosis cerebrotendinous		
Disorders of fluid, electrolyte and acid-base balance		
Volume depletion	E86	This whole group of codes is not made of diseases, but of various pathological conditions. Nevertheless, it may be useful to keep it so as to code various causes of mortality without previously identified illness.
Dehydration		
Depletion of volume of plasma or extracellular fluid		
Hypovolaemia		
Excludes: dehydration of newborn	P74.1	
Excludes: hypovolaemic shock:		
· NOS	R57.1	
· postoperative	T81.1	
· traumatic	T79.4	
Fluid overload	E87.7	
Hyperosmolality and hypernatraemia	E87.0	
<i>Sodium [Na] excess</i>		
<i>Sodium [Na] overload</i>		

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<i>ICD11 table draft</i>	<i>Corresponding ICD10 code</i>	<i>Comments</i>
Hypo-osmolality and hyponatraemia <i>Sodium [Na] deficiency</i>	E87.1	
Excludes: Syndrome of inappropriate secretion of antidiuretic hormone	E22.2	
Hyperkalaemia <i>Potassium [K] excess</i> <i>Potassium [K] overload</i>	E87.5	
Hypokalaemia <i>Potassium [K] deficiency</i>	E87.6	
Other specified disorders of electrolyte and fluid balance, not elsewhere classified Hyperchloraemia Hypochloraemia	E87.8	
Acidosis	E87.2	
Lactic acidosis Excludes: Lactic acidosis due to inborn error of metabolism	various	
Metabolic acidosis Respiratory acidosis Excludes: diabetic acidosis	E10-E14 with common fourth character .1	
Alkalosis Metabolic acidosis Respiratory acidosis	E87.3	
Mixed disorder of acid-base balance	E87.4	
Other specified rare metabolic disorders	E88.8	

**Groups included in the metabolic diseases
section of ICD10, to be reclassified in ICD11**

Groups included in the metabolic diseases section of ICD10, to be reclassified in ICD11

Albinism	E70.3	Include in dermatology.
Ocular albinism		
Oculocutaneous albinism		
Chediak(-Steinbrinck)-Higashi syndrome		
Hermansky-Pudlak syndrome		
Lactose intolerance	E73	Include in gastroenterology.
Congenital lactase deficiency	E73.0	
Secondary lactase deficiency	E73.1	
Other lactose intolerance	E73.8	
Lactose intolerance, unspecified	E73.9	
Other disorders of intestinal carbohydrate absorption	E74.3	Include in gastroenterology.
Glucose-galactose malabsorption		
Sucrase deficiency		
Excludes: lactose intolerance	E73.-	
Other specified disorders of carbohydrate metabolism	E74.8	Include in nephrology.
Essential pentosuria		
Oxalosis		
<i>Oxaluria</i>		
Renal glycosuria		
Gilbert's syndrome	E80.4	Include in hepatology.
Crigler-Najjar syndrome	E80.5	Include in hepatology.
Other disorders of bilirubin metabolism	E80.6	Include in hepatology.
Dubin-Johnson syndrome		
Rotor's syndrome		
Disorder of bilirubin metabolism, unspecified	E80.7	Include in hepatology.
Cystic fibrosis	E84	This is not a metabolic disease. The appropriate chapter is debatable.
Includes: mucoviscidosis		
Cystic fibrosis with pulmonary manifestations	E84.0	
Cystic fibrosis with intestinal manifestations	E84.1	
Meconium ileus	P75*	
meconium obstruction in cases where cystic fibrosis is		
Excludes: known not to be present	P76.0	

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Cystic fibrosis with other manifestations	E84.8	
Cystic fibrosis with combined manifestations		
Cystic fibrosis, unspecified	E84.9	
Amyloidosis	E85	Include in new chapter for multisystemic diseases.
Excludes: Alzheimer's disease	G30	
Non-neuropathic hereditary amyloidosis	E85.0	
Familial Mediterranean fever		
Hereditary amyloid nephropathy		
Neuropathic hereditary amyloidosis	E85.1	
Amyloid polyneuropathy (Portuguese)		
Hereditary amyloidosis, unspecified	E85.2	
Secondary systemic amyloidosis	E85.3	
Haemodialysis-associated amyloidosis		
Organ-limited amyloidosis	E85.4	
Localized amyloidosis		
Other amyloidosis	E85.8	
Amyloidosis, unspecified	E85.9	
Lipodystrophy, not elsewhere classified	E88.1	Include in endocrinology.
Excludes: Whipple's disease	K90.8	
Lipomatosis, not elsewhere classified	E88.2	Include in endocrinology
Lipomatosis dolorosa [Dercum]		