

ICD-11 revision process for Rare Diseases
Chapter IV
E50-E68

Nutritional diseases

Draft structure n° 1

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Draft for ICD revision for rare diseases - Confidential
Chapter IV: Nutritional 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 Nutritional diseases which represents the proposal for a **new ICD**.
3. The ICD-10+ draft structure for Nutritional diseases, which allows the introduction of some changes in the present ICD-10 version in order to prepare the ICD-11.
4. A series of proposals for each individual modification submitted in the ICD-10+ WHO web platform.

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. You can send your feed-back to Orphanet.

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2. 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 systematic reviews 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.

While the structure of ICD-11 is being prepared, low-level modifications can immediately be proposed for ICD-10+ on the ICD Update and Revision Platform available on the Internet (<https://extranet.who.int/icdrevision/>).

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



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(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 ICD-10+ revision

Diseases can be mentioned in the ICD-10 in three possible ways:

- **Inclusion terms** are included in the ICD tabular list, which is accessible from the WHO website, and published in the Volume I of the ICD-10 printed version. The tabular list is not exhaustive.
- **Index terms** are synonyms or names of lesser entities not mentioned in the tabular list, only mentioned in the alphabetic index. The alphabetic index tends to be exhaustive. Each index term refers to a code in the tabular list. They are published in the Volume III of the ICD-10 printed version, but are not visible via the WHO website (where they only redirect to the appropriate code).
- **Exclusion terms** are used to indicate the main code of entries that could have been correctly classified in some chapter, but that are actually included elsewhere in the classification.

A preliminary study on how rare diseases should be represented in ICD-10+ and ICD-11 was made, in order to identify those diseases that need a specific code in order to be traceable in the Information systems for statistics, and those which can be included as a group under a specific category (as inclusion terms or index terms). New specific codes are requested for rare diseases in ICD-10+ that meet at least two of the following criteria:

- they have a dedicated patient support group
- there is a diagnostic test available
- there is a specific treatment available
- prevalence figures exist. For prevalence figures:
http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_decreasing_prevalence_or_cases.pdf

Rare diseases that do not meet these criteria are proposed in ICD-10+ as inclusion terms under a category for the more prevalent of them, and as index terms for those with a few cases described in the literature.

3. Rationale for nutritional diseases revision

Nutritional diseases should be understood as related to inappropriate dietary intake of some or several nutrients. *Metabolic diseases*, related to the internal processing of nutrients, are not included in this revision proposal.

The current ICD10 classification of nutritional diseases is currently made of three blocks of codes :

[E40-E46](#) Malnutrition

[E50-E64](#) Other nutritional deficiencies

The logo for Orphanet, featuring the word "orphanet" in a lowercase, sans-serif font. The "o" is blue, and the "rphanet" is black. A blue arc is positioned above the "n" and "e".

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[E65-E68](#) Obesity and other hyperalimantation

Our revision deals with the last two blocks only: no change was suggested in the malnutrition block, as no rare disease is to be added in it.

The classification follows a clinical rather than aetiological approach. Many nutritional deficiencies are associated with disorders clinically specific of one particular system (e.g. iodine deficiency with hypothyroidism, iron deficiency with anaemias, etc.) and are thus coded in the corresponding chapter, rather than in the nutrition chapter. In the latter, exclusion notes are used to refer to such diseases all the same; we suggest to use them quite systematically.

A general problem is the coding of iatrogenic diseases. In the current ICD10, iatrogenic disorders in every chapter are regularly included separately under a series of specific codes (labelled “post-procedural”). However, since the number of post-procedural codes is quite small, it is impossible to code many post-procedural entities with precision. How iatrogenic disorders will be represented in ICD11 is not settled, but the current system needs revision.

The problem is especially acute with nutritional disorders, because there is no code at all for iatrogenic nutritional diseases. We suggest for ICD10+ that the primary code should be used to identify the nutritional deficiency, completed by an additional code for external causes (chapter XX), as is often done already in the current ICD10 in such situations. Codes for medical and surgical care as external causes are Y40 to Y84.

**ICD-11 draft structure
for
Nutritional diseases**

ICD-11 draft structure for nutritional diseases

NB: New terms are highlighted in grey.

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

NB: Synonyms are in italics

Comments

ICD11 table draft

Correspondance in
 ICD-10
 E40-E46

Malnutrition

THIS SECTION HAS NOT BEEN REVISED, and no rare disease is to be added there

Identification of iatrogenic vitamin deficiencies is to be settled in ICD11. **Vitamin deficiencies**

This section has not been modified save for adding an exclusion note.

Vitamin A deficiency **E50**

Hypovitaminosis A

Vitamin A deficiency with conjunctival xerosis **E50.0**

Vitamin A deficiency with Bitot's spot and conjunctival xerosis **E50.1**

Bitot's spot in the young child

Vitamin A deficiency with corneal xerosis **E50.2**

Vitamin A deficiency with corneal ulceration and xerosis **E50.3**

Vitamin A deficiency with keratomalacia **E50.4**

Vitamin A deficiency with night blindness **E50.5**

Vitamin A deficiency with xerophthalmic scars of cornea **E50.6**

Other ocular manifestations of vitamin A deficiency **E50.7**

Xerophthalmia NOS

Other manifestations of vitamin A deficiency **E50.8**

*Follicular keratosis due to vitamin A deficiency+ (L86 *)*

Xeroderma

Vitamin A deficiency, unspecified **E50.9**

Excludes: sequelae of vitamin A deficiency **E64.1**

Excludes: Hypercarotenaemia and vitamin A deficiency

Thiamine deficiency **E51**

Vitamin B1 deficiency

Beriberi **E51.1**

Dry beriberi

Wet beriberi

Other manifestations of thiamine deficiency **E51.8**

Thiamine deficiency, unspecified **E51.9**

Excludes: sequelae of thiamine deficiency **E64.8**

Excludes: Thiamine-responsive megaloblastic anemia

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Comments	ICD11 table draft	Correspondance in ICD-10
	Cyanocobalamin deficiency	E53.8
	<i>Cobalamin deficiency</i>	
	<i>Vitamin B12 deficiency</i>	
	Excludes: <u>vitamin B 12 deficiency anaemia</u>	D51
	Vitamin B deficiency, unspecified	E53.9
	Excludes: <u>sequelae of vitamin B deficiency</u>	E64.8
	Ascorbic acid deficiency	E54
	<i>Deficiency of vitamin C</i>	
	Scurvy	
	Excludes: <u>scorbutic anaemia</u>	D53.2
	<u>sequelae of vitamin C deficiency</u>	E64.2
	Vitamin D deficiency	E55
	<i>Avitaminosis D</i>	
	Excludes: <u>adult osteomalacia</u>	M83
	Excludes: <u>osteoporosis</u>	M80-M81
	Excludes: <u>sequelae of rickets</u>	E64.3
	Excludes: <u>juvenile (idiopathic) osteoporosis</u>	
The code should reflect that rickets is often secondary to another disease.	Rickets	E55.0
	<i>Infantile or juvenile osteomalacia due to vitamine D deficiency</i>	
	Note: Rickets as a consequence of chronic disease should be coded with the latter's code.	
	Excludes: <u>vitamin-D-resistant rickets</u>	E83.3
	Vitamin D deficiency, unspecified	E55.9
	Deficiency of vitamin E	E56.0
	Excludes: Ataxia, Friedreich-like, with selective vitamin E deficiency	
	Deficiency of vitamin K	E56.1
	Excludes: <u>deficiency of coagulation factor due to vitamin K deficiency</u>	D68.4
	<u>vitamin K deficiency of newborn</u>	P53
	Deficiency of other vitamins	E56.8
	Excludes: <u>sequelae of other vitamin deficiencies</u>	E64.8
	Vitamin deficiency, unspecified	E56.9

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Comments	ICD11 table draft	Correspondance in ICD-10
Identification of iatrogenic mineral deficiencies is to be settled in ICD11.	Dietary mineral deficiencies	E61
	Use additional external cause code (Chapter XX), if desired, to identify drug, if drug-induced.	
	Excludes: <u>disorders of mineral metabolism</u>	E83
	Excludes: <u>iodine-deficiency-related thyroid disorders</u>	E00-E02
	Excludes: <u>sequelae of malnutrition and other nutritional deficiencies</u>	E64
	Dietary calcium deficiency	E58
	Excludes: <u>disorder of calcium metabolism</u>	E83.5
	<u>sequelae of calcium deficiency</u>	E64.8
	Dietary selenium deficiency	E59
	Keshan disease	
	Excludes: <u>sequelae of selenium deficiency</u>	E64.8
	Dietary zinc deficiency	E60
	Dietary copper deficiency	E61.0
	Dietary iron deficiency	E61.1
	Excludes: <u>iron deficiency anaemia</u>	D50
	Dietary magnesium deficiency	E61.2
Literature is scarce, the clinical significance of this condition is unclear.	Dietary manganese deficiency	E61.3
Literature is scarce, the clinical significance of this condition is unclear.	Dietary chromium deficiency	E61.4
Literature is scarce, the clinical significance of this condition is unclear.	Dietary molybdenum deficiency	E61.5
Literature is scarce, the clinical significance of this condition is unclear.	Dietary vanadium deficiency	E61.6

Comments	ICD11 table draft	Correspondance in ICD-10
No revision has been made in this section.	<p>Other nutritional deficiencies</p> <p><i>Excludes:</i> <u>dehydration</u></p> <p><i>Excludes:</i> <u>failure to thrive</u></p> <p><i>Excludes:</i> <u>feeding problems in newborn</u></p> <p>Essential fatty acid [EFA] deficiency</p> <p>Imbalance of constituents of food intake</p> <p>Deficiency of multiple nutrient elements</p> <p>Other specified nutritional deficiencies <u>Nutritional cardiomyopathy NOS+ (I43.2*)</u></p> <p>Unspecified nutritional deficiency</p> <p>Sequelae of malnutrition and other nutritional deficiencies</p> <p>Sequelae of protein-energy malnutrition <i>Excludes:</i> <u>retarded development following protein-energy malnutrition</u></p> <p>Sequelae of vitamin A deficiency</p> <p>Sequelae of vitamin C deficiency</p> <p>Sequelae of rickets Use additional code (M40.-), if desired, to identify kyphosis</p> <p>Sequelae of other nutritional deficiencies</p> <p>Sequelae of unspecified nutritional deficiency</p>	<p>E63</p> <p>E86</p> <p>R62.8</p> <p>P92</p> <p>E63.0</p> <p>E63.1</p> <p>E61.7</p> <p>E61.8 + E63.8 E61.8 + E63.8 (†), I43.2 (*)</p> <p>E61.9 + E63.9</p> <p>E64 E64.0 <u>E45</u> E64.1 E64.2 E64.3</p> <p>E64.8 E64.9</p>
	<p>Obesity and other nutritional excess</p> <p>Obesity <i>Excludes:</i> <u>adiposogenital dystrophy</u> <u>lipomatosis NOS</u> <u>lipomatosis dolorosa [Dercum disease]</u> <u>Syndromic obesities (e.g. Prader-Willi syndrome)</u></p> <p>Obesity due to excess calories</p> <p>Drug-induced obesity Use additional external cause code (Chapter XX), if desired, to identify drug.</p>	<p>E65-E68</p> <p>E66 E23.6 E88.2 E88.2</p> <p>E66.0 E66.1</p>

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Comments	ICD11 table draft	Correspondance in ICD-10
	Extreme obesity with alveolar hypoventilation Pickwickian syndrome	E66.2
	Leptin-related genetic obesity Obesity due to congenital leptin deficiency Obesity due to congenital leptin resistance Obesity due to leptin receptor gene deficiency Obesity due to pro-opiomelanocortin deficiency Obesity due to prohormone convertase-I deficiency Obesity due to melanocortin-4 receptor deficiency	
	Other obesity Morbid obesity	E66.8
	Obesity, unspecified	E66.9
	Localized adiposity Fat pad	E65
	Familial symmetric lipomatosis Launois-Bensaude adenolipomatosis	E88.8
	Other nutritional excess	E67
	Excludes: <u>nutritional excess NOS</u>	R63.2
	<u>sequelae of nutritional excess</u>	E68
	Hypervitaminosis A	E67.0
	Hypercarotenaemia	E67.1
	Hypercarotenaemia and vitamin A deficiency	
	Megavitamin-B₆ syndrome	E67.2
	Hypervitaminosis D	E67.3
	Copper excess Indian childhood cirrhosis Tyrolean infantile cirrhosis Idiopathic copper toxicosis	
	Other specified nutritional excess	E67.8
	Sequelae of nutritional excess	E68

Revision proposals to the current ICD-10
Chapter IV
E50-E68

Nutritional diseases

(ICD-10 +)

Modifications proposed to ICD-10 (ICD-10 +)

NB: New inclusion terms are highlighted in grey.

NB: Modifications are highlighted in red

NB: Terms to be deleted are crossed out; terms to be renamed are crossed out and followed by the new suggested denomination.

ICD10 code

ICD10+ modifications

Malnutrition (E40-E46)

Note: The degree of malnutrition is usually measured in terms of weight, expressed in standard deviations from the mean of the relevant reference population. When one or more previous measurements are available, lack of weight gain in children, or evidence of weight loss in children or adults, is usually indicative of malnutrition. When only one measurement is available, the diagnosis is based on probabilities and is not definitive without other clinical or laboratory tests. In the exceptional circumstances that no measurement of weight is available, reliance should be placed on clinical evidence.

If an observed weight is below the mean value of the reference population, there is a high probability of severe malnutrition if there is an observed value situated 3 or more standard deviations below the mean value of the reference population; a high probability of moderate malnutrition for an observed value located between 2 and less than 3 standard deviations below this mean; and a high probability of mild malnutrition for an observed value located between 1 and less than 2 standard deviations below this mean.

Excludes: [intestinal malabsorption \(K90.- \)](#)
[nutritional anaemias \(D50-D53 \)](#)
[sequelae of protein-energy malnutrition \(E64.0 \)](#)
[slim disease \(B22.2 \)](#)
[starvation \(T73.0 \)](#)

E40 Kwashiorkor
Severe malnutrition with nutritional oedema with dyspigmentation of skin and hair.
Excludes: [marasmic kwashiorkor \(E42 \)](#)

E41 Nutritional marasmus
Severe malnutrition with marasmus
Excludes: [marasmic kwashiorkor \(E42 \)](#)

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ICD10 code	ICD10+ modifications
E42	Marasmic kwashiorkor Severe protein-energy malnutrition [as in E43]: <ul style="list-style-type: none">· intermediate form· with signs of both kwashiorkor and marasmus
E43	Unspecified severe protein-energy malnutrition Severe loss of weight [wasting] in children or adults, or lack of weight gain in children leading to an observed weight that is at least 3 standard deviations below the mean value for the reference population (or a similar loss expressed through other statistical approaches). When only one measurement is available, there is a high probability of severe wasting when the observed weight is 3 or more standard deviations below the mean of the reference population. Starvation oedema
E44	Protein-energy malnutrition of moderate and mild degree
E44.0	Moderate protein-energy malnutrition Weight loss in children or adults, or lack of weight gain in children leading to an observed weight that is 2 or more but less than 3 standard deviations below the mean value for the reference population (or a similar loss expressed through other statistical approaches). When only one measurement is available, there is a high probability of moderate protein-energy malnutrition when the observed weight is 2 or more but less than 3 standard deviations below the mean of the reference population.
E44.1	Mild protein-energy malnutrition Weight loss in children or adults, or lack of weight gain in children leading to an observed weight that is 1 or more but less than 2 standard deviations below the mean value for the reference population (or a similar loss expressed through other statistical approaches). When only one measurement is available, there is a high probability of mild protein-energy malnutrition when the observed weight is 1 or more but less than 2 standard deviations below the mean of the reference population.
E45	Retarded development following protein-energy malnutrition Nutritional: <ul style="list-style-type: none">· short stature· stunting Physical retardation due to malnutrition
E46	Unspecified protein-energy malnutrition Malnutrition NOS Protein-energy imbalance NOS

ICD10 code

ICD10+ modifications

Other nutritional deficiencies (E50-E64)

Excludes: [nutritional anaemias \(D50-D53 \)](#)

E50 Vitamin A deficiency

Excludes:

[sequelae of vitamin A deficiency \(E64.1 \)](#)

Excludes:

Hypercarotenaemia and vitamin A deficiency

E50.0 Vitamin A deficiency with conjunctival xerosis

E50.1 Vitamin A deficiency with Bitot's spot and conjunctival xerosis

Bitot's spot in the young child

E50.2 Vitamin A deficiency with corneal xerosis

E50.3 Vitamin A deficiency with corneal ulceration and xerosis

E50.4 Vitamin A deficiency with keratomalacia

E50.5 Vitamin A deficiency with night blindness

E50.6 Vitamin A deficiency with xerophthalmic scars of cornea

E50.7 Other ocular manifestations of vitamin A deficiency

Xerophthalmia NOS

E50.8 Other manifestations of vitamin A deficiency

[Follicular keratosis due to vitamin A deficiency + \(L86 *\)](#)

[Xeroderma due to vitamin A deficiency + \(L86 *\)](#)

E50.9 Vitamin A deficiency, unspecified

Hypovitaminosis A NOS

E51 Thiamine deficiency

Excludes:

[sequelae of thiamine deficiency \(E64.8 \)](#)

Excludes:

Thiamine-responsive megaloblastic anemia

E51.1 Beriberi

Beriberi:

· dry

· [wet+ \(I98.8* \)](#)

E51.2 Wernicke's encephalopathy

Excludes : Korsakov's psychosis or syndrome

· non alcoholic Korsakov syndrome F04

· alcohol induced Korsakov syndrome F10.6

· Korsakov syndrome induced by other psychoactive substances (F11-F19 with common fourth character)

E51.8 Other manifestations of thiamine deficiency

E51.9 Thiamine deficiency, unspecified

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ICD10 code		ICD10+ modifications
E52	Niacin deficiency [pellagra] Deficiency: · niacin(-tryptophan) · nicotinamide Pellagra (alcoholic) Excludes:	sequelae of niacin deficiency (E64.8)
E53	Deficiency of other B group vitamins Excludes:	sequelae of vitamin B deficiency (E64.8) vitamin B 12 deficiency anaemia (D51.-)
E53.0	Riboflavin deficiency Ariboflavinosis	
E53.1	Pyridoxine deficiency Vitamin B ₆ deficiency Excludes:	pyridoxine-responsive sideroblastic anaemia (D64.3)
E53.8	Deficiency of other specified B group vitamins Deficiency: · biotin · cyanocobalamin · folate · folic acid · pantothenic acid · vitamin B ₁₂	
E53.9	Vitamin B deficiency, unspecified	
E54	Ascorbic acid deficiency Deficiency of vitamin C Scurvy Excludes:	scorbutic anaemia (D53.2) sequelae of vitamin C deficiency (E64.2)
E55	Vitamin D deficiency Avitaminosis D Excludes : Excludes : Excludes : Excludes:	adult osteomalacia (M83.-) osteoporosis (M80-M81) sequelae of rickets (E64.3) juvenile (idiopathic) osteoporosis

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ICD10 code		ICD10+ modifications
E55.0	Rickets, active Osteomalacia due to vitamine D deficiency: · infantile · juvenile Excludes-:	rickets: -coeliac (K90.0) -Crohn's (K50.-) -inactive (E64.3) -renal (N25.0) · vitamin-D-resistant rickets (E83.3) Rickets caused by a chronic disease should be coded with the latter's code.
E55.9	Vitamin D deficiency, unspecified Avitaminosis-D	
E56	Other vitamin deficiencies Excludes:	sequelae of other vitamin deficiencies (E64.8)
E56.0	Deficiency of vitamin E Excludes:	Ataxia, Friedreich-like, with selective vitamin E deficiency
E56.1	Deficiency of vitamin K Excludes:	deficiency of coagulation factor due to vitamin K deficiency (D68.4) vitamin K deficiency of newborn (P53)
E56.8	Deficiency of other vitamins	
E56.9	Vitamin deficiency, unspecified	
E58	Dietary calcium deficiency Excludes:	disorder of calcium metabolism (E83.5) sequelae of calcium deficiency (E64.8)
E59	Dietary selenium deficiency Keshan disease Excludes:	sequelae of selenium deficiency (E64.8)
E60	Dietary zinc deficiency	

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ICD10 code	ICD10+ modifications
E61	Deficiency of other nutrient elements Use additional external cause code (Chapter XX), if desired, to identify drug, if drug-induced. Excludes:
	disorders of mineral metabolism (E83.-) iodine-deficiency-related thyroid disorders (E00-E02) sequelae of malnutrition and other nutritional deficiencies (E64.-)
E61.0	Copper deficiency
E61.1	Iron deficiency Excludes:
	iron deficiency anaemia (D50.-)
E61.2	Magnesium deficiency
E61.3	Manganese deficiency
E61.4	Chromium deficiency
E61.5	Molybdenum deficiency Excludes:
	Encephalopathy due to sulphite oxidase deficiency (main code E72.1 disorders of sulfur-bearing amino-acid metabolism)
E61.6	Vanadium deficiency
E61.7	Deficiency of multiple nutrient elements
E61.8	Deficiency of other specified nutrient elements
E61.9	Deficiency of nutrient element, unspecified
E63	Other nutritional deficiencies Excludes :
	dehydration (E86) failure to thrive (R62.8) feeding problems in newborn (P92.-) sequelae of malnutrition and other nutritional deficiencies (E64.-)
E63.0	Essential fatty acid [EFA] deficiency
E63.1	Imbalance of constituents of food intake
E63.8	Other specified nutritional deficiencies
E63.9	Nutritional deficiency, unspecified Nutritional cardiomyopathy NOS+ (I43.2*)
E64	Sequelae of malnutrition and other nutritional deficiencies
E64.0	Sequelae of protein-energy malnutrition Excludes:
	retarded development following protein-energy malnutrition (E45)
E64.1	Sequelae of vitamin A deficiency
E64.2	Sequelae of vitamin C deficiency
E64.3	Sequelae of rickets Use additional code (M40.-), if desired, to identify kyphosis
E64.8	Sequelae of other nutritional deficiencies
E64.9	Sequelae of unspecified nutritional deficiency

ICD10 code

ICD10+ modifications

Obesity and other **hyperalimentation** > **nutritional excess** (E65-E68)

E65 Localized adiposity
Fat pad

E88.8 > new code Familial symmetric lipomatosis

Launois-Bensaude adenolipomatosis

Index only: Madelung's disease

E66 Obesity

Excludes: [adiposogenital dystrophy \(E23.6 \)](#)

Excludes: [lipomatosis NOS \(E88.2 \)](#)

Excludes: [lipomatosis dolorosa \[Dercum\] \(E88.2 \)](#)

Excludes: [Syndromic obesities \(e.g. Prader-Willi syndrome\)](#)

E66.0 Obesity due to excess calories

E66.1 Drug-induced obesity

Use additional external cause code (Chapter XX), if desired, to identify drug.

E66.2 Extreme obesity with alveolar hypoventilation

Pickwickian syndrome

new code Leptin related genetic obesity

Obesity due to congenital leptin deficiency

Obesity due to congenital leptin resistance

Index only: Obesity due to leptin receptor gene deficiency

Index only: Obesity due to pro-opiomelanocortin deficiency

Index only: Obesity due to prohormone convertase-I deficiency

Index only: Obesity due to melanocortin-4 receptor deficiency

E67 **Other hyperalimentation > Other nutritional excess**

Excludes: [hyperalimentation NOS \(R63.2 \)](#)

[sequelae of hyperalimentation \(E68 \)](#)

E67.0 Hypervitaminosis A

E67.1 Hypercarotenaemia

Hypercarotenaemia and vitamin A deficiency

E67.2 Megavitamin-B₆ syndrome

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ICD10 code	ICD10+ modifications
E67.3	Hypervitaminosis D
new code	Copper excess
	<i>Index only:</i> Indian childhood cirrhosis
	<i>Index only:</i> Tyrolean infantile cirrhosis
	<i>Index only:</i> Idiopathic copper toxicosis
E67.8	Other specified nutritional excess
E68	Sequelae of nutritional excess

Argumentation for ICD revision proposals for ICD-10 Chapter IV

Nutritional diseases

These proposals are available at the WHO ICD revision platform

<https://extranet.who.int/icdrevision>

Reminder:

Would you wish to participate in the discussion in the revision platform, you need to create an account in order to obtain your login/password.

Otherwise, your comments are welcome at Orphanet
(see page 2 for contacts)

ICD revision proposal n° 1664

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1664>

Title : **New code for thiamine-responsive megaloblastic anaemia with diabetes mellitus and sensorineural deafness**

Primary Code Affected : D53

Secondary Codes Affected : E13, E51, H90

Detailed Description

[new code: D53.x] Thiamine-responsive megaloblastic anaemia with diabetes mellitus and sensorineural deafness
Rogers syndrome

E13 Other specified diabetes mellitus [See before E10 for subdivisions]

Excludes: diabetes mellitus (in):

· ...

· **Add: Excludes:** Thiamine-responsive megaloblastic anaemia with diabetes mellitus and sensorineural deafness (D53.x)

· ...

E51 Thiamine deficiency

Excludes: sequelae of thiamine deficiency (E64.8)

Add: Excludes: Thiamine-responsive megaloblastic anaemia with diabetes mellitus and sensorineural deafness (D53.x)

H90.3 Sensorineural hearing loss, bilateral

Add: Excludes: Thiamine-responsive megaloblastic anaemia with diabetes mellitus and sensorineural deafness (D53.x)

Additions are to be reflected in the alphabetic index.

Rationale

There is no explicit indication for coding this syndrome and because it involves several systems there are many interpretations and codes possible. Since ICD is monoaxial one manifestation has to be favoured, and exclusions are to be made to avoid improper coding at other possible places.

The choice of a code under the D53 group is thus somewhat arbitrary; it mainly follows the emphasis of the denomination of the megaloblastic anaemia present in this entity.

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- they have a dedicated patient support group
- there is a diagnostic test available
- there is a specific treatment available
- prevalence figures exist, other than a mere number of cases.

For diseases that do not meet these criteria, a mere index term is requested for the rarest ones (those with only a few cases described), an inclusion term for the less rare ones. Epidemiological data are regularly updated on Orphanet's website (www.orpha.net).

Description of the disease is available for instance in the following article :

Ricketts CJ, Minton JA, Samuel J, Ariyawansa I, Wales JK, Lo IF, Barrett TG, *Thiamine-responsive megaloblastic anaemia syndrome: long-term follow-up and mutation analysis of seven families*, Acta Paediatr. 2006 Jan;95(1):99-104.

Pubmed abstract:

AIM: Thiamine-responsive megaloblastic anaemia syndrome (TRMA) is the association of diabetes mellitus, anaemia and deafness, due to mutations in SLC19A2, encoding a thiamine transporter protein. This is a unique monogenic form of vitamin-dependent diabetes for which there is limited long-term data. We aimed to study genotype-phenotype relationships and long-term follow-up in our cohort. METHODS: We have studied 13 patients from seven families and have follow-up data for a median of 9 y (2-30 y). RESULTS: All patients originated from Kashmir or Punjab, and presented with non-immune, insulin-deficient diabetes mellitus, sensorineural deafness and a variable anaemia in the first 5 y of life, the anaemia progressing to megaloblastic and sideroblastic changes in the bone marrow. The anaemia and diabetes mellitus responded to oral thiamine hydrochloride 25 mg/d, but during puberty thiamine supplements became ineffective, and almost all patients require insulin therapy and regular blood transfusions in adulthood. All patients are homozygous for mutations in the SLC19A2 gene. We have identified a novel missense mutation (T158R) that was excluded in 100 control alleles. CONCLUSION: Diabetes in this syndrome is due to an insulin insufficiency that initially responds to thiamine supplements; however, most patients become fully insulin dependent after puberty. A mutation screening strategy is feasible and likely to identify mutations in almost all cases.

Supporting Publication Web Links

Title **Orphanet record for Thiamine-responsive megaloblastic anaemia with diabetes mellitus and sensorineural deafness**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=49827

Title **Pubmed reference for Ricketts et al. 2009**

Web address (URL) <http://www.ncbi.nlm.nih.gov/pubmed/16373304?dopt=Abstract>

ICD revision proposal n° 1665

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1665>

Title : Friedreich-like ataxia with selective vitamin E deficiency

Primary Code Affected : G11.1

Secondary Codes Affected : E56.0

Detailed Description

E56.0 Deficiency of vitamin E

Add: **Excludes:** Ataxia, Friedreich-like, with selective vitamin E deficiency

G11.1 Early-onset cerebellar ataxia*

Note: Onset usually before the age of 20

Early-onset cerebellar ataxia with:

- essential tremor
- myoclonus [Hunt's ataxia]
- retained tendon reflexes

Friedreich's ataxia (autosomal recessive)

X-linked recessive spinocerebellar ataxia

Friedreich-like ataxia with selective vitamin E deficiency

Additions are to be reflected in the alphabetic index.

Rationale

Friedreich-like ataxia with selective vitamin E deficiency is an autosomal recessive condition genetically distinct from Friedreich ataxia but clinically quite similar: difficulties to coordinate movements and dysarthria, associated with other neurological signs (loss of reflexes, decrease of deep sensation, pes cavus and scoliosis). Plasma levels of vitamin E are very low. When left untreated, patients' neurological condition progressively worsens, leading to autonomy loss. Substitutive treatment with high doses of vitamin E may slow or even stop progression of the disease.

See Orphanet's record (link below) for basic information about this disease.

We suggest to represent this disease with the same code than for true Friedreich's ataxia, on account of the clinical similarity, but an exclusion note should be made in vitamin E deficiencies.

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- there is a specific treatment available
- prevalence figures exist, other than a mere number of cases.

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Supporting Publication Web Links

Title **Orphanet record for Friedreich-like ataxia with selective vitamin E deficiency**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=96

ICD revision proposal n° 1666

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1666>

Title : **Nutritional excess vs. hyperalimentation**

Primary Code Affected : E65-E68

Secondary Codes Affected :

Detailed Description

E65-E68 ~~Obesity and other hyperalimentation~~ > Obesity and nutritional excess

...

E67 ~~Other hyperalimentation~~ > Other nutritional excess

Excludes: [hyperalimentation NOS \(R63.2 \)](#)
[sequelae of hyperalimentation \(E68 \)](#)

E67.0 Hypervitaminosis A

E67.1 Hypercarotenaemia

E67.2 Megavitamin-B₆ syndrome

E67.3 Hypervitaminosis D

E67.8 ~~Other specified hyperalimentation~~ > Other specified nutritional excess

Rationale

The word “hyperalimentation” is prone to be narrowly understood as “hyperphagia”, which is too restrictive for this set of codes which potentially covers excessive nutrient intakes as well as excessive eating. We suggest to change the wording of the code description to avoid such a misinterpretation.

Supporting Publication Web Links

Title

Web address (URL)

ICD revision proposal n° 1667

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1667>

Title : New code for leptin-related genetic obesities

Primary Code Affected : E66

Secondary Codes Affected :

Detailed Description

[new code : E66.x]Genetic obesity, leptin-related

Obesity due to congenital leptin deficiency

Obesity due to congenital leptin resistance

Matching additions are to be made in the alphabetic index. Moreover, the following specific forms could be ascribed to the new code as index terms only :

Obesity due to leptin receptor gene deficiency

Obesity due to pro-opiomelanocortin deficiency

Obesity due to prohormone convertase-I deficiency

Obesity due to melanocortin-4 receptor deficiency

Rationale

There is no way to code some genetic obesities explicitly in the current scheme : they have to be subsumed in code *E66.8 Other obesity*. *Orphanet's classification of rare endocrine diseases* features the following entities as genetic obesities (see the appended note about how *Orphanet's classifications of rare diseases* are elaborated) :

Rare endocrine disease

Genetic obesity

Fragile X syndrome

Prader-Willi syndrome

Prader-Willi syndrome due to chromosome 15 uniparental disomy, maternal origin

Prader-Willi syndrome due to paternal deletion of 15q11.13

Prader-Willi syndrome due to paternal deletion of 15q11.13, type 1

Prader-Willi syndrome due to paternal deletion of 15q11.13, type 2

Prader-Willi syndrome due to translocation

Prader-Willi syndrome due to imprinting mutation

WAGR syndrome

Cohen syndrome

Albright hereditary osteodystrophy

Pseudohypoparathyroidism, type 1A

Pseudohypoparathyroidism, type 1C

Pseudopseudohypoparathyroidism

Alström syndrome

Borjeson-Forssman-Lehmann syndrome

Choroideremia - deafness - obesity

Schinzler syndrome

Wilson-Turner syndrome

Hydrocephalus - obesity - hypogonadism

Bardet-Biedl syndrome

Obesity due to congenital leptin deficiency

Obesity due to congenital leptin resistance

Obesity due to pro-opiomelanocortin deficiency

Obesity due to prohormone convertase-I deficiency

Obesity due to melanocortin-4 receptor deficiency

Obesity due to leptin receptor gene deficiency

MEHMO syndrome

MOMO syndrome

Uniparental disomy of maternal origin, chromosome 14

Vasquez-Hurst-Sotos syndrome

While a high number of these entities are actually complex syndromes for which other codes could be more appropriate, leptin-related obesities should be mentioned in this chapter. We suggest a code to be created for them as a whole, and

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specific forms to be created as index terms. Basic facts about the individual forms can be found in Orphanet's records linked below.

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- there is a specific treatment available
- prevalence figures exist, other than a mere number of cases.

For diseases that do not meet these criteria, a mere index term is requested for the rarest ones (those with only a few cases described), an inclusion term for the less rare ones. Epidemiological data are regularly updated on Orphanet's website (www.orpha.net).

Supporting Publication Web Links

Title **Orphanet classification of rare diseases – Methodological note**

Web address (URL) <http://www.orpha.net/data/patho/Pro/en/OrphanetClassificationRareDiseases.pdf>

Title **Orphanet record for Obesity due to congenital leptin deficiency**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=66628

Title **Orphanet record for Obesity due to congenital leptin resistance**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=179490

Title **Orphanet record for Obesity due to leptin receptor gene deficiency**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=179494

Title **Orphanet record for Obesity due to pro-opiomelanocortin deficiency**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=71526

Title **Orphanet record for Obesity due to prohormone convertase-I deficiency**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=71528

Title **Orphanet record for Obesity due to melanocortin-4 receptor deficiency**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=71529

ICD revision proposal n° 1668

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1668>

Title : Precisions to vitamin D deficiency

Primary Code Affected : E55

Secondary Codes Affected : M81.5

Detailed Description

E55 Vitamin D deficiency

Add: **Avitaminosis D**

Excludes: [adult osteomalacia \(M83.- \)](#)
[osteoporosis \(M80-M81 \)](#)
[sequelae of rickets \(E64.3 \)](#)
[juvenile \(idiopathic\) osteoporosis \(M81.5\)](#)

E55.0 Rickets, active

Osteomalacia Add: **due to vitamine D deficiency:**

- infantile
- juvenile

Excludes: rickets

~~[coeliac \(K90.0 \)](#)~~

~~[Crohn's \(K50.- \)](#)~~

~~[inactive \(E64.3 \)](#)~~

~~[renal \(N25.0 \)](#)~~

~~[vitamin-D-resistant \(E83.3 \)](#)~~

Add: **Note:** Rickets caused by a chronic disease should be coded with the latter's code.

E55.9 Vitamin D deficiency, unspecified

Avitaminosis D

M81.5 Idiopathic osteoporosis

Add: **juvenile (idiopathic) osteoporosis**

Additions are to be reflected in the alphabetic index.

Rationale

Avitaminosis D is a synonym of *vitamin D deficiency* and as such should be coded E55; E55.9 should only be used for unspecified forms.

Juvenile osteoporosis is a rare condition with decreased bone mass which is however correctly mineralised, in contrast with rickets where primary mineralisation is defective. The distinction follows the adult distinction between osteoporosis, with decrease in bone mass and deterioration of bone microarchitecture vs. osteomalacia, with defective primary mineralisation of bone.,). A review (in French) can be found in:

Delalande D, Jung C, Labedan I, Lechevalier P, Madre C, Roche S, Koné-Paut I, *Les ostéoporoses juvéniles*, Archives de pédiatrie, 2008, 15(4):420-30.

English abstract:

Osteoporosis is induced by a disorder of the bone turnover that generates an accelerated destruction process and leads to the rarefaction of the protein matrix. The RANK-L/RANK/OPG system is the main actor of the bone remodelling regulation. Juvenile osteoporoses may have primary or secondary aetiologies. The main causes include constitutional bone fragilities, and osteoporoses, which are secondary to chronic inflammatory diseases and sustained steroid treatment. Etiologic diagnosis relies on a clinical basis, and is often made too lately when complications occur. Osteodensitometry is a sensitive and noninvasive tool for measuring mineral bone density in children. The reliability of results is limited by the variations due to patients' age, gender, pubertal stage, and by the length of bone pieces. The optimal treatment of osteoporosis is preventive, and includes accurate nutritional diet, D vitamin-calcium supplementation and regular physical

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activity. Biphosphonates are used for treatment of symptomatic osteoporoses. Careful utilization is required in childhood because their late potential secondary effects are still unknown. New antiresorptive drugs and other that stimulate osteoformation are successfully used in adults. Their effects have not been studied in the paediatric population.

Inactive rickets seems to be an old-fashioned term for sequelae of rickets. Does it need to be kept as an inclusion term?

The list of chronic diseases associated with rickets currently given is not exhaustive and could be replaced by a general remark notifying that in such cases the chronic disease code should be used. Alternatively, if rickets is important in the clinical picture, it could be mentioned using double coding : the chronic disease would then receive the primary code with dagger, and rickets coded secondarily with asterisk. ICD10 rules would then require all chronic conditions with important association with rickets to be listed explicitly so as to allow for this coding.

Supporting Publication Web Links

Title **Orphanet record for Juvenile osteoporosis**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=85193

Title **Pubmed reference for Delalande et al. 2008**

Web address (URL) <http://www.ncbi.nlm.nih.gov/pubmed/18329256?dopt=Abstract>

ICD revision proposal n° 1669

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1669>

Title : **New code for familial symmetric lipomatosis**

Primary Code Affected : E65-E68

Secondary Codes Affected : E88.8

Detailed Description

[new code in group E65-E68] Familial symmetric lipomatosis

Launois-Bensaude adenolipomatosis

Lipomatosis central non-encapsulated

Lipomatosis familial benign cervical

E88 Other metabolic disorders

Use additional external cause code (Chapter XX), if desired, to identify drug, if drug-induced. **Excludes:** histiocytosis X (chronic) ([D76.0](#))

(...)

E88.8 Other specified metabolic disorders

~~Launois-Bensaude adenolipomatosis~~

Trimethylaminuria

Changes and additions are to be reflected in the alphabetic index. We also suggest to add the following term as index term only (see discussion below) :

Madelung's disease [new code]

Rationale

Familial symmetric lipomatosis is a form of obesity characterised by a large and symmetrical accumulation of adipose mass at the level of the head, neck and upper trunk. It is currently lumped together with other metabolic disorders than cannot be put under better defined group of codes. We suggest to move it in the group of codes used for obesity: *E65-E68 Obesity and other hyperalimentation*.

Basic facts about this disease (including its various denominations) can be found in Orphanet's record linked below. For a more developed description of the disease, see for instance the following article :

Verna G, Kefalas N, Boriani F, Carlucci S, Choc I, Bocchiotti MA, *Launois-Bensaude Syndrome: an unusual localization of obesity disease*, Obesity Surgery, 2008, 18(10):1313-7

Pubmed abstract:

BACKGROUND: Launois-Bensaude syndrome is a rare pathology consisting of adipose masses symmetrically distributed mainly in the superior part of the body. Men are especially affected between age of 30 and 60 as well as chronic alcohol abusers. Etiopathogenesis is attributable to mutations or deletions of mitochondrial DNA, and alcohol is a possible cofactor. METHODS: The current treatment of the disease is described based on the authors' experience. Four cases treated in our department are retrospectively reviewed regarding comorbidities and type of surgery performed. RESULTS: A relevant and long-lasting reduction of fat bulges has been obtained in all cases with no major complications except for a mild anemia. CONCLUSION: Launois-Bensaude syndrome causes a functional rather than esthetic concern due to the peculiar localization of fat bulges. Currently, the only effective therapy is surgery, through lipectomy or liposuction of adipose bulges.

We suggest that the denomination Madelung's disease should not be used as inclusion term, but as index term only, because of possible confusion with Madelung's deformity, which is totally different. The presence of both side by side in the alphabetic index will warn there that these are indeed two conditions.

Supporting Publication Web Links

Title **Orphanet record for Familial symmetric lipomatosis**

Web address (URL) http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=2398

Title **Pubmed reference for Verna et al. 2008**

Web address (URL) <http://www.ncbi.nlm.nih.gov/pubmed/18408978?dopt=Abstract>

ICD revision proposal n° 1670

URL : <https://extranet.who.int/icdrevision/PropD.aspx?prop=1670>

Title : New code for dietary copper excess

Primary Code Affected : E67

Secondary Codes Affected : T56.4

Detailed Description

Hierarchic list

E67 Other hyperalimantation

(...)

[new code: E67.x] Copper excess

Alphabetic index

Copper excess E67.x

Indian childhood cirrhosis E67.x

Tyrolean infantile cirrhosis E67.x

Idiopathic copper toxicosis E67.x

Rationale

Excessive copper intake has been shown to cause disorders with mostly hepatic manifestations - distinct from Wilson disease (or hepatolenticular degeneration) which is a metabolic disease of genetic origin. An assessment of the morbid risk of copper excess can be found in:

Uauy R, Maass A, Araya M, *Estimating risk from copper excess in human populations*, American Journal of Clinical Nutrition, 2008, 88(3):867S-71S.

Pubmed abstract :

Risk assessment for nutrients assumes a single population with a normal distribution of indexes of requirements and excess. Toxic levels are by definition intakes above the upper level; for copper, however, because we lack noninvasive, sensitive biomarkers of storage or early damage from excess, excess is based on the infrequent occurrence of clinical disease, such as unexplained liver cirrhosis. We examine the limitations of this approach for copper given the very low prevalence of clinical and subclinical disease and suggest that the population risk for copper excess be based on hepatic copper loading as a potentially quantifiable measurement. The challenge ahead is to develop biomarkers that predict the population risk of elevated hepatic copper stores and thus the possibility of disease in a population.

Examples of copper excess disorders given in the article are *Indian childhood cirrhosis*, *Tyrolean childhood cirrhosis*, *Idiopathic copper toxicosis*, related to high exposure to dietary copper with a possible genetic predisposition.

The only way currently to code these diseases is to use a toxicology code T56.4, which is quite inappropriate for situations such as described in this article, with chronic excess rather than acute poisoning. We thus suggest a new code to be created under E67 group, which already contains excessive intake of nutrient such as hypervitaminoses or hypercarotenaemia.

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- prevalence figures exist, other than a mere number of cases.

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For diseases that do not meet these criteria, a mere index term is requested for the rarest ones (those with only a few cases described), an inclusion term for the less rare ones. Epidemiological data are regularly updated on Orphanet's website (www.orpha.net).

Supporting Publication Web Links

Title **Pubmed reference for Uauy et al. 2008**

Web address (URL) <http://www.ncbi.nlm.nih.gov/pubmed/18779311?dopt=Abstract>

ICD revision proposal n° 1671

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1671>

Title : **Post-procedural nutritional deficiencies**

Primary Code Affected : E50-E64

Secondary Codes Affected : Y40-Y84

Detailed Description

Other nutritional deficiencies (E50-E64)

Add: Use external causes codes Y40-Y84 when secondary to medical or surgical care.

Excludes: nutritional anaemias ([D50-D53](#))

Rationale

How should post-procedural nutritional disorders be coded - for instance, impaired nutrient absorption following bariatric surgery ?

In the current ICD10, iatrogenic disorders in every chapter are regularly included separately under a series of specific codes (labelled “post-procedural”) : but chapter IV lumps together endocrine, nutritional and metabolic diseases, and detailed post-procedural codes are available for some common endocrine disorders only. This makes it impossible to identify post-procedural nutritional deficiencies with any precision ; the only code available is **E89.8 Other postprocedural endocrine and metabolic disorders** without other precision.

Alternatively, the nutritional deficiency can be coded with the appropriate code while keeping entirely silent about its being iatrogenic.

We suggest for ICD10+ that the primary code should be used to identify the nutritional deficiency, completed by an additional code for external causes (chapter XX), as is often done already in the current ICD10 in such situations. Codes for medical and surgical care as external causes are Y40 to Y84.

Supporting Publication Web Links

Title

File

ICD revision proposal n° 1672

URL : <https://extranet.who.int/icdrevison/PropD.aspx?prop=1672>

Title : **Exclude Korsakov syndrome from Wernicke encephalopathy**

Primary Code Affected : E51.2

Secondary Codes Affected : F04, F10.6, F11-F19

Detailed Description

E51.2 Wernicke's encephalopathy

Add: **Excludes** : Korsakov's psychosis or syndrome

· non alcoholic F04

· alcohol induced F10.6

· induced by other psychoactive substances (F11-F19 with common fourth character .6)

...

Rationale

Wernicke encephalopathy and Korsakov syndrome are related neuropsychological disorders related to thiamine deficiency, often caused by alcohol abuse. The relationship between the two entities is recognized in the use of the cover name Wernicke-Korsakov encephalopathy.

Wernicke encephalopathy typically features mental confusion, ataxia and nystagmus; Korsakov syndrome features with persistent learning and memory deficiencies, and frequently follows Wernicke encephalopathy. Thiamine administration is use to relieve Wernicke encephalopathy but often fails to do so with developed Korsakov syndrome.

For a review about these conditions, see for instance

Sechi G, Serra A., Wernicke's encephalopathy: *new clinical settings and recent advances in diagnosis and management*, Lancet Neurology, 2007 May;6(5):442-55.

Abstract:

Wernicke's encephalopathy is an acute neuropsychiatric syndrome resulting from thiamine deficiency, which is associated with significant morbidity and mortality. According to autopsy-based studies, the disorder is still greatly underdiagnosed in both adults and children. In this review, we provide an update on the factors and clinical settings that predispose to Wernicke's encephalopathy, and discuss the most recent insights into epidemiology, pathophysiology, genetics, diagnosis, and treatment. To facilitate the diagnosis, we classify the common and rare symptoms at presentation and the late-stage symptoms. We emphasise the optimum dose of parenteral thiamine required for prophylaxis and treatment of Wernicke's encephalopathy and prevention of Korsakoff's syndrome associated with alcohol misuse. A systematic approach helps to ensure that patients receive a prompt diagnosis and adequate treatment.

The clinical distinction between the two conditions justifies that several codes exist, but an exclusion note for Korsakov syndrome under Wernicke encephalopathy would be welcome to avoid improper coding.

Note that the place of Wernicke encephalopathy is based on aetiological considerations, while Korsakov syndrome is classified on clinical grounds; the current ICD10 lacks consistency there. Such diseases could also be coded in the neurology chapter; this has to be taken into account for ICD11.

The combined denomination Wernicke-Korsakov encephalopathy was evoked in proposal n° 1135 and ascribed to code F10.6.

Supporting Publication Web Links

Title **Pubmed reference for Sechi and Serra 2007**

The logo for Orphanet, featuring the word "orphanet" in a lowercase, sans-serif font. The "o" is stylized with a blue arc above it, and the "n" has a blue arc above it as well. The "e" is also stylized with a blue arc above it.

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Web address (URL) <http://www.ncbi.nlm.nih.gov/pubmed/17434099?dopt=Abstract>