

## **ICD-11 revision process for Rare Diseases**

### **Disorders of the immune system**

*ICD-10 chapter III, section D80-D89*

*Draft structure n°2*

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*2<sup>nd</sup> draft for ICD revision for rare diseases - Confidential*  
*Chapter III, codes D80-D89: Disorders of the immune system*

**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 *Developmental anomalies*, which represents Orphanet's proposal for the next ICD version to be released in 2014.

You are invited to:

1. Check the ICD-11 draft structure and the way rare diseases are represented in it. (See the checklist of issues to be addressed on page 5.)
2. Send your feed-back to Orphanet **before the end of November 2011.**

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3. Disseminate this invitation to your colleagues who are experts in this field as we wish to involve as many experts as necessary

## 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. Working groups or individual experts invited by the Topic Advisory Groups (TAG) review the proposals. To learn more about the whole revision process: <http://www.who.int/classifications/icd/revision/en/index.html>

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 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.

Orphanet is a comprehensive peer-reviewed database of information on rare diseases. Over 6,000 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. The Orphanet dataset is available and can be downloaded at [www.orphadata.org](http://www.orphadata.org)

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>

The Alpha version of ICD-11 is expected **by the end of 2011**.

## 2. General principles for ICD revision

**The current ICD-10 classification is mono-hierarchical, meaning that every entity can figure only at one point in the classification.** The rationale for this choice was to avoid double counting, since the ICD is primarily used as a statistic tool. This is a problem however for numerous diseases that can be associated with more than one body system (chapters being broadly organised along them). In such cases, one system must then be given priority, and *exclusion notes* are put in the other relevant chapters to redirect users to the correct code.

**In the future ICD-11, the classification shall become poly-hierarchical, and every entity shall be assigned a unique identifying number:** diseases will be able to figure in all relevant places in the classification (for instance, the several endocrine diseases associated with developmental anomalies will figure both in the endocrine and developmental anomalies chapters). 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

mono-hierarchical system; and for statistics, it is still necessary to avoid double counting. Therefore, **the ICD-11 will also feature linearisations, i.e. versions allowing for a mono-hierarchical approach.** We suggest that the priority specialty should be 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.

**A new dedicated chapter will be created for multisystemic diseases in ICD-11.** In the current ICD-10 they are found scattered in various chapters, sometimes on questionable grounds.

**The Orphanet proposal for a new ICD classification generally follows a clinical rather than aetiological approach.** Groups of diseases are preferentially defined on the basis of shared clinical features.

The WHO style guide recommends to avoid eponyms in disease nomenclature, except when their use is dominant and well established (e.g. Alzheimer disease, Huntington disease, etc.).

### *3. Specific issues for the revision of the chapter on immunological diseases*

The current ICD-10 classification of immunological diseases is mostly made of the block of codes [D80-D89](#) *Certain disorders involving the immune mechanism*. Other relevant diseases are included in the block [D70-D77](#) *Other diseases of blood and blood-forming organs*. Both blocks are part of ICD-10's chapter III *Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism*.

Both should be united in ICD-11 into a separate chapter *Disorders of the immune system*.

However, this does not cover every disease of immune origin. The ICD is mostly organised by body systems, and many diseases of immune origin that are clinically specific to one particular system are therefore coded in the corresponding chapter; this is especially the case for autoimmune and autoinflammatory diseases. For instance, *Myasthenia gravis* is coded in neurology rather than in immunology.

**We propose that autoimmune and autoinflammatory diseases should be classified firstly in the chapter(s) corresponding to the body system(s) they affect** (or in the new chapter for multisystemic diseases if appropriate); this classification should get priority in linearisations. **Secondly, they should be classified in this chapter for immune diseases.**

**Not every autoimmune and autoinflammatory disease is included yet in this 2<sup>nd</sup> draft for immune diseases.** They will be included in a final section that remains to be constructed, and that will be released later. We intend to build it while working on the planned chapter for multisystemic diseases.

The new classification is first presented as a compressed version, featuring only the first three top-levels, so as to provide a view on the general structure. The full classification is then given, including all levels of detail.

**4. Checklist for reviewing this revision proposal**

- Is the revised structure of the chapter relevant and sound considering the expected uses of the ICD-11?
  - Mortality reporting
  - Morbidity reporting
  - Clinical practice
  - Research practice
  - Primary care
  - Public Health Reporting
- Are there entities lacking in this chapter?
- Are there entities of doubtful status in this chapter?
- Are there entities that could be improperly understood by coders because of their denomination or their place in the hierarchy?
- For diseases that can be included in several medical specialties, do you agree with the choice of the priority specialty for the linearised version of the ICD-11?

## **Legend of notations used in tables**

**Top-level** divisions are indicated by **grey shading**.

**Main denomination of entities** are mentioned in **plain writing**. They are systematically associated with reference numbers in Orphanet's database of rare diseases, when possible; a dash is used when no Orpha number precisely matches the entity.

**Current ICD-10 codes** are also indicated when they exactly match an entity.

**Synonyms** are put in **italics** below the main denominations.

**Exclusions** are indicated just below a head entry : they are systematically marked with the notation **Excludes:**

**Inclusion of an entity** is usually left implicit. However, if a head entry has both exclusions and inclusions, the beginning of the list of inclusions is marked with the notation **Includes:**

**Example:**

183663	Hyper-IgM syndrome with susceptibility to opportunistic infections <i>HIGM with susceptibility to opportunistic infections</i>
183666	<b>Excludes:</b> Hyper-IgM syndrome without susceptibility to opportunistic infections
101088	<b>Includes :</b> Hyper-IgM syndrome type 1 <i>Hyper-IgM syndrome due to CD40 ligand deficiency</i> <i>Hyper-IgM syndrome due to CD40L deficiency</i> <i>X-linked hyper-IgM syndrome</i> <i>HIGM1</i>
101090	Hyper-IgM syndrome type 3 <i>Hyper-IgM syndrome due to CD40 deficiency</i> <i>HIGM3</i>

Here:

- *Hyper-IgM syndrome without susceptibility to opportunistic infections* is excluded from the group *Hyper-IgM syndrome with susceptibility to opportunistic infections*.
- *Hyper-IgM type 1* and *Hyper-IgM type 2* are included as subtypes under *Hyper-IgM syndrome with susceptibility to opportunistic infections*.
- *HIGM with susceptibility to opportunistic infections* is a synonym of the main denomination *Hyper-IgM syndrome with susceptibility to opportunistic infections*.
- *Hyper-IgM syndrome due to CD40 ligand deficiency*, *Hyper-IgM syndrome due to CD40L deficiency*, *X-linked hyper-IgM syndrome* and *HIGM1* are synonyms of the main denomination *C syndrome*.
- *Hyper-IgM syndrome due to CD40 deficiency* and *HIGM3* are synonyms of the main denomination *Hyper-IgM syndrome type 3*.

**ICD-11 revised draft structure  
for  
Immunological diseases**

*Top-level classification only*

Orpha

ICD-11 table draft

ICD-10

**101997 Primary immunodeficiencies**

**101988 Primary immunodeficiencies due to disorders of innate immunity**

101987	Constitutional neutropenias	<b>D70</b>
183681	Functional neutrophil defects	<b>D71</b>
101992	Defects in the complement system	<b>D84.1</b>
93665	Autoinflammatory syndromes with immunodeficiency	
183710	Genetic susceptibility to particular pathogens	
75391	Immunodeficiency with natural-killer cell deficiency	

**179006 Primary immunodeficiencies due to disorders of adaptive immunity**

101972	Combined immunodeficiencies	<b>D81</b>
101977	Primary immunodeficiencies with predominantly antibody defects	<b>D80</b>
169346	DNA repair defects other than combined T-cell and B-cell immunodeficiencies	
169355	Immunodeficiency syndromes with autoimmunity	
169361	Immune dysregulation diseases	
—	Other defects in adaptive immunity	

**183716 Complex primary immunodeficiency syndromes**

2314	Autosomal dominant hyperimmunoglobulin E syndrome	<b>D82.4</b>
302	Epidermodysplasia verruciformis	
98813	EDA-ID syndrome	
69088	OL-EDA-ID syndrome	
51636	WHIM syndrome	

**— Acquired immunodeficiencies**

**178996 Acquired neutropenias**

47612	Felty syndrome	
86872	T-cell large granular lymphocyte leukaemia	
2688	Adult idiopathic neutropenia	<b>D70</b>

**— Human immunodeficiency virus [HIV] disease**

—	Human immunodeficiency virus [HIV] disease resulting in infectious and parasitic diseases	<b>B20</b>
—	Human immunodeficiency virus [HIV] disease resulting in malignant neoplasms	<b>B21</b>
—	Human immunodeficiency virus [HIV] disease resulting in other specified diseases	<b>B22</b>
—	Human immunodeficiency virus [HIV] disease resulting in other conditions	<b>B23</b>
—	Unspecified human immunodeficiency virus [HIV] disease	<b>B24</b>

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Orpha	ICD-11 table draft	ICD-10
—	<b>Diseases of thymus</b>	E32
—	<b>Excludes:</b> Thymic aplasia or hypoplasia with immunodeficiency [Monosomy 22q11 and Nezelof syndrome]	D82.1
589	<b>Excludes :</b> Myasthenia gravis	G70.0
—	<b>Persistent hyperplasia of thymus</b>	E32.0
—	<b>Hypertrophy of thymus</b>	
—	<b>Abscess of thymus</b>	E32.1
100100	<b>Thymic tumours</b>	
3398	Thymic epithelial tumour	
97289	Thymic endocrine tumour	
169105	<b>Good syndrome</b>	
—	<b>Other disorders involving the immune system</b>	D89
—	<b>Excludes:</b> Monoclonal gammopathy	D47.2
—	<b>Excludes:</b> Transplant failure and rejection	T86
95431	<b>Excludes:</b> Twin to twin transfusion syndrome	
—	<b>Chronic neutrophilia</b>	
—	Hereditary chronic neutrophilia	
—	Acquired chronic neutrophilia	
—	<b>Benign hypergammaglobulinaemic purpura</b>	D89.0
91139	<b>Simple cryoglobulinaemia</b>	D89.1
91138	<b>Excludes:</b> Mixed cryoglobulinaemia	D89.1
91378	<b>Hereditary angioedema</b>	D84.1
100050	Hereditary angioedema type 1	
100051	Hereditary angioedema type 2	
100054	Hereditary angioedema type 3	
—	<b>Autoimmune lymphoproliferative syndrome-related disorders</b>	
—	Diazani autoimmune lymphoproliferative disease	
—	RAS-associated autoimmune lymphoproliferative disease	
238510	Lymphoproliferative diseases with immunodeficiency	
—	<b>Autoimmune diseases</b>	
	SECTION IN CONSTRUCTION	



# **ICD-11 revised draft structure for Immunological diseases**

*Full classification*

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*Chapter III, codes D80-D89: Disorders of the immune system*

Orpha	ICD-11 table draft	ICD-10	Main code when elsewhere	Comments
101997	<b>Primary immunodeficiencies</b>			
101988	<b>Primary immunodeficiencies due to disorders of innate immunity</b>			
101987	<b>Constitutional neutropenias</b>	<b>D70</b>		
	<i>Congenital neutropenias</i>			
175	<b>Excludes:</b> Cartilage-hair hypoplasia			developmental anomalies
42738	<b>Includes:</b> Severe congenital neutropenia	<b>D70</b>		
486	Autosomal dominant severe congenital neutropenia			
86788	X-linked severe congenital neutropenia			
—	CSF3R-related severe congenital neutropenia			
99749	Kostmann syndrome	<b>D70</b>		
178503	Dursun syndrome			
	<i>Pulmonary arterial hypertension - leukopenia - atrial septal defect</i>			
2686	Cyclic neutropenia	<b>D70</b>		
	Constitutional neutropenia with extra-haematopoietic manifestations			
99749	<b>Excludes:</b> Kostmann syndrome	<b>D70</b>		
183678	<b>Includes:</b> Hermansky-Pudlak syndrome with neutropenia	<b>E70.3</b>		
	<i>Hermansky-Pudlak syndrome type 2</i>			
	<i>HPS2</i>			
193	Cohen syndrome			developmental anomalies
811	Shwachman-Diamond syndrome	<b>D61.0</b>		haematology
	<i>Congenital lipomatosis of pancreas</i>			
79259	Glycogen storage disease type 1B			metabolism
	<i>Type 1B glycogenosis</i>			
111	Barth syndrome			metabolism
	<i>Cardioskeletal myopathy-neutropenia</i>			
	<i>3-methylglutaconic aciduria type 2</i>			
	<i>MGA2</i>			
	<i>BTHS</i>			
2739	Onychotrichodysplasia - neutropenia			dermatology
	<i>Trichothiodystrophy type G</i>			
	<i>ONMR syndrome</i>			
	<i>Itin syndrome</i>			
2690	Neutropenia - monocytopenia - deafness			

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90023	Primary immunodeficiency syndrome due to p14 deficiency <i>Primary immunodeficiency syndrome with short stature</i>			
221046	Poikiloderma with neutropenia <i>Poikiloderma with neutropenia, Clericuzio type</i>		dermatology	
183681	<b>Functional neutrophil defects</b> <i>Severe recurrent infections with present neutrophils</i>	<b>D71</b>		
379	Chronic granulomatous disease <i>Chronic septic granulomatosis</i>	<b>D71</b>		
169142	Recurrent infection due to specific granule deficiency <i>Neutrophil-specific granule deficiency</i>			
183707	Neutrophil immunodeficiency syndrome <i>Rac 2 deficiency</i>			
2968	Leukocyte adhesion deficiency <i>LAD</i>			
99842	Leukocyte adhesion deficiency type I <i>LAD-I</i>			
99843	Leukocyte adhesion deficiency type II <i>Rambam-Hasharon syndrome</i> <i>CDG syndrome type IIc</i> <i>CDG IIc</i> <i>LAD-II</i> <i>GDP-fucose transporter deficiency</i>			
99844	Leukocyte adhesion deficiency type III <i>LAD-III</i> <i>Leukocyte adhesion deficiency-1 variant</i> <i>LAD-I variant</i>			
2587	Myeloperoxidase deficiency <i>MPO deficiency</i>			
678	Papillon-Lefèvre syndrome <i>Palmoplantar keratosis - periodontopathy</i>		dermatology	
362	Glucose-6-phosphate-dehydrogenase deficiency <i>G6PD deficiency</i> <i>Favism</i>	<b>D55.0</b>	haematology	

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
101992	<b>Defects in the complement system</b> <i>Immunodeficiency with a complement cascade protein anomaly</i>	<b>D84.1</b>		
2134	<b>Excludes:</b> Atypical haemolytic-uraemic syndrome		haematology	
447	<b>Excludes:</b> Paroxysmal nocturnal haemoglobinuria	<b>D59.5</b>	haematology	
169147	<b>Excludes:</b> Hereditary angioedema <b>Includes:</b> Immunodeficiency with an early component of complement deficiency <i>Deficiency of complement initial pathway</i> Complement component C1q deficiency Complement component C1r/C1s deficiency		multisystemic diseases	Hereditary angioedema is included in the immunology chapter since patients are seen in immunology clinics and treated with a blood product. However, it is not included here as there is no immunodeficiency, but in the category "Other disorders of the immune system".
1481	Complement component C2 deficiency Complement component C4 deficiency			
169150	Complement component C3 deficiency Immunodeficiency with a late component of complement deficiency <i>Deficiency of complement terminal pathway</i> Complement component C5 deficiency Complement component C6 deficiency Complement component C8 deficiency Complement component C9 deficiency			
169467	Immunodeficiency with factor B deficiency Recurrent Neisseria infections due to factor D deficiency Complement component C4b-binding protein deficiency Immunodeficiency with decay accelerating factor deficiency <i>Immunodeficiency with DAF deficiency</i> <i>Immunodeficiency with CD55 deficiency</i>			
200418	Immunodeficiency with factor I anomaly			
200421	Immunodeficiency with factor H anomaly			
2966	Immunodeficiency with properdin deficiency			
169464	Immunodeficiency with CD59 deficiency Immunodeficiency with MASP-2 deficiency <i>Mannan-binding lectin serine protease 2 deficiency</i> Immunodeficiency with MBL deficiency <i>Mannan-binding lectin deficiency</i> <i>Mannose-binding protein deficiency</i> <i>MBP deficiency</i>			

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93665	<b>Autoinflammatory syndromes with immunodeficiency</b>		multisystemic diseases	
342	Familial mediterranean fever <i>Periodic disease</i>	<b>E85.0</b>	multisystemic diseases	
93563	Paediatric familial mediterranean fever <i>Paediatric periodic disease</i>		multisystemic diseases	
32960	TNF receptor 1 associated periodic syndrome <i>TRAPS syndrome</i> <i>Autosomal dominant periodic fever</i> <i>Familial hibernian fever</i>		multisystemic diseases	
343	Hyperimmunoglobinaemia D with recurrent fever <i>Hyperimmunoglobulinaemia D with periodic fever</i> <i>Partial mevalonate kinase deficiency with recurrent fever, with or without hyperimmunoglobinaemia D</i> <i>Hyper-IgD syndrome</i> <i>HIDS</i> <i>Dutch-type periodic fever</i>		multisystemic diseases	
69126	Pyogenic arthritis - pyoderma gangrenosum - acne <i>Familial recurrent arthritis</i> <i>PAPA syndrome</i> <i>FRA</i>		multisystemic diseases	
77297	Majeed syndrome <i>Chronic recurrent multifocal osteomyelitis - congenital dyserythropoietic anaemia - neutrophilic dermatosis</i>		multisystemic diseases	
90340	Blau syndrome <i>Caspase recruitment domain-containing protein 15 deficiency</i> <i>CARD15 deficiency</i>		multisystemic diseases	
42642	Periodic fever, aphthous stomatitis, pharyngitis and adenopathy <i>PFAPA syndrome</i> <i>Marshall syndrome with periodic fever</i>			There is another entity called Marshall syndrome, characterised by ocular and craniofacial abnormalities, sensorineural hearing loss and anhidrotic extodermal dysplasia. Because of the ambiguity, an alternative name is preferred here as a main term.
208650	Cryopyrin-associated periodic syndrome <i>CAPS</i>		multisystemic diseases	
1451	Chronic infantile neurological, cutaneous and articular syndrome <i>CINCA syndrome</i> <i>Infantile-onset multisystem inflammatory disease</i> <i>Neonatal-onset multisystem inflammatory disease</i> <i>Prieur-Griscelli syndrome</i> <i>NOMID syndrome</i> <i>IOMID syndrome</i>		multisystemic diseases	

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47045	Familial cold autoinflammatory syndrome <i>Familial cold urticaria</i> FCAS FCU	L50.2	multisystemic diseases	
575	Muckle-Wells syndrome <i>Urticaria - deafness - amyloidosis</i>		multisystemic diseases	
210115	Autoinflammatory disease due to interleukin-1 receptor antagonist deficiency <i>DIRA</i>		multisystemic diseases	
183710	<b>Genetic susceptibility to particular pathogens</b>			
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency <i>IRAK4 deficiency</i>			
183713	Pyogenic bacterial infections due to MyD88 deficiency			
748	Mendelian susceptibility to mycobacterial diseases <i>MSMD</i> <i>Idiopathic infection caused by BCG or atypical mycobacteria</i>			Are the various subtypes of this group of diseases relevant for the ICD-11 ?
—	Autosomal recessive mendelian susceptibility to mycobacterial diseases <i>AR MSMD</i>			
—	Complete autosomal recessive interferon-gamma receptor 1 deficiency <i>Complete AR IFNGR1 deficiency</i>			
—	Partial autosomal recessive interferon-gamma receptor 1 deficiency <i>Partial AR IFNGR1 deficiency</i>			
—	Complete autosomal recessive interferon-gamma receptor 2 deficiency <i>Complete AR IFNGR2 deficiency</i>			
—	Partial autosomal recessive interferon-gamma receptor 2 deficiency <i>Partial AR IFNGR2 deficiency</i>			
—	Autosomal recessive interleukin 12 receptor beta-1 chain deficiency <i>AR IL12RB1 deficiency</i>			
—	Autosomal recessive interleukin 12p40 deficiency <i>AR IL12B deficiency</i>			
—	Autosomal recessive tyrosine kinase 2 deficiency <i>Autosomal recessive hyper-IgE syndrome with atypical mycobacteriosis</i>			

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—	Autosomal dominant mendelian susceptibility to mycobacterial diseases <i>AD MSMD</i>			
—	Autosomal dominant interferon-gamma receptor 1 deficiency <i>AD IFNGR1 deficiency</i>			
—	Autosomal dominant Stat1 deficiency <i>AD STAT1 deficiency</i>			
—	X-linked recessive mendelian susceptibility to mycobacterial diseases <i>XR MSMD</i>			
1930	Herpes simplex encephalitis <i>Herpes simplex neuroinvasion</i> <i>Herpetic encephalitis</i> <i>HSV encephalitis</i>	<b>B00.4</b>	neurology	
75391	<b>Immunodeficiency with natural-killer cell deficiency</b> <i>Immunodeficiency with NK-cell deficiency</i>			
179006	<b>Primary immunodeficiencies due to disorders of adaptive immunity</b>			
101972	<b>Combined immunodeficiencies</b> <i>Combined T and B cell immunodeficiency</i>	<b>D81</b>		
268330	Severe combined immunodeficiencies [SCID] and related disorders			
183660	Severe combined immunodeficiencies [SCID]			
—	Severe combined immunodeficiency [SCID] with low T and B cell numbers <i>T- B- severe combined immunodeficiency</i>	<b>D81.1</b>		
—	Severe combined immunodeficiency [SCID] T- B- due to RAG1 deficiency <i>Severe combined immunodeficiency [SCID] T- B- due to recombination activating gene 1 deficiency</i>			
—	Severe combined immunodeficiency [SCID] T- B- due to RAG2 deficiency <i>Severe combined immunodeficiency [SCID] T- B- due to recombination activating gene 2 deficiency</i>			
277	Severe combined immunodeficiency [SCID] T- B- due to adenosine deaminase deficiency	<b>D81.3</b>		
39041	Severe combined immunodeficiency [SCID] with hypereosinophilia <i>Omenn syndrome</i>			

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231053 275	Severe combined immunodeficiency [SCID] with sensitivity to ionizing radiation Severe combined immunodeficiency [SCID], alymphocytotic type <i>Agammaglobulinaemia, alymphocytotic type</i> <i>Severe combined immunodeficiency [SCID], Athabaskan type</i> <i>Severe combined immunodeficiency [SCID] due to DNA cross-link repair protein 1C deficiency</i> <i>Severe combined immunodeficiency [SCID] due to DCLRE1C deficiency</i> <i>Severe combined immunodeficiency [SCID] due to Artemis protein deficiency</i>			
169079	Severe combined immunodeficiency [SCID] - microcephaly - growth retardation - sensitivity to ionizing radiation <i>NHEJ1 syndrome</i> <i>Cernunnos deficiency</i> DNA-ligase IV deficiency <i>LIG4 syndrome</i>			
99812 33355	Severe combined immunodeficiency [SCID] with reticular dysgenesis <i>Severe combined immunodeficiency [SCID] with leukopenia</i> <i>Generalized haematopoietic hypoplasia</i> <i>Congenital aleukocytosis</i> <i>Reticular dysgenesis</i> <i>De Vaal disease</i>	<b>D81.0</b>		
—	Severe combined immunodeficiency with low or normal B cell numbers <i>T- B+ severe combined immunodeficiency</i>	<b>D81.2</b>		
276	Severe combined immunodeficiency [SCID] T- B+ due to gamma chain deficiency <i>Severe combined immunodeficiency [SCID] T- B+, X-linked</i>			
35078	Severe combined immunodeficiency [SCID] T- B+ due to JAK3 deficiency			
169090	Severe combined immunodeficiency [SCID] due to CRAC channel dysfunction <i>Immune dysfunction due to T-cell inactivation due to calcium entry defect</i>			

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169154	Severe combined immunodeficiency [SCID] T- B+ due to IL-7R-alpha deficiency			
169157	Severe combined immunodeficiency [SCID] T- B+ due to CD45 deficiency			
169160	Severe combined immunodeficiency [SCID] T- B+ due to CD3-delta/CD3-epsilon/CD3-zeta			
228003	Severe combined immunodeficiency [SCID] due to CORO1A deficiency <i>Severe combined immunodeficiency [SCID] due to coronin-1A deficiency</i>			
169100	Severe combined immunodeficiency due to CD25 deficiency <i>Severe combined immunodeficiency [SCID] due to interleukin-2 receptor alpha chain deficiency</i>			
911	Severe combined immunodeficiency [SCID] due to ZAP70 deficiency <i>Severe combined immunodeficiency [SCID] due to zeta-associated-protein 70 deficiency</i>			
169095	Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy <i>Severe combined immunodeficiency [SCID] due to winged helix deficiency</i>			
231154	Severe combined immunodeficiency [SCID] T+ B+ due to partial RAG1 deficiency <i>Severe combined immunodeficiency [SCID] T+ B+ due to partial recombination activating gene 1 deficiency</i>			
—	Immunodeficiency due to absence of thymus	<b>D81.4</b>		
83471	Nezelof syndrome <i>Thymic aplasia</i>			
567	Monosomy 22q11 <i>Conotruncal anomalies face syndrome</i> <i>Velocardiofacial syndrome</i> <i>Shprintzen syndorme</i> <i>Sedlackova syndrome</i> <i>Microdeletion 22q11</i> <i>DiGeorge syndrome</i> <i>DiGeorge sequence</i> <i>CATCH 22</i>	<b>D82.1</b>	developmental anomalies	
—	Major histocompatibility complex deficiency <i>Bare lymphocyte syndrome</i>			
34592	Major histocompatibility complex class I deficiency <i>Immunodeficiency by defective expression of HLA class 1</i> <i>Bare lymphocyte syndrome type 1</i>	<b>D81.6</b>		

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
572	Major histocompatibility complex class II deficiency <i>Immunodeficiency by defective expression of HLA class 2</i> <i>HLA class 2-negative Severe combined immunodeficiency [SCID]</i> <i>Bare lymphocyte syndrome type 2</i>	<b>D81.7</b>		
183663	Hyper-IgM syndrome with susceptibility to opportunistic infections <i>HIGM with susceptibility to opportunistic infections</i>	<b>D80.5</b>		The current ICD-10 D80.5 code is described as "Immunodeficiency with increased immunoglobulin M", and includes all hyper-IgM syndromes. It must be split between those with and without opportunistic infections, with reciprocal exclusions to prevent confusion.
183666	<b>Excludes:</b> Hyper-IgM syndrome without susceptibility to opportunistic infections			
101088	<b>Includes:</b> Hyper-IgM syndrome type 1 <i>Hyper-IgM syndrome due to CD40 ligand deficiency</i> <i>Hyper-IgM syndrome due to CD40L deficiency</i> <i>X-linked hyper-IgM syndrome</i> <i>HIGM1</i>			
101090	Hyper-IgM syndrome type 3 <i>Hyper-IgM syndrome due to CD40 deficiency</i> <i>HIGM3</i>			
3322	Hoyeraal-Hreidarsson syndrome <i>Progressive pancytopenia - immunodeficiency - cerebellar hypoplasia</i>		haematology	
760	Immunodeficiency due to purine nucleoside phosphorylase deficiency  <i>Purine nucleoside phosphorylase deficiency</i> <i>PNP deficiency</i> <i>Immunodeficiency due to PNP deficiency</i>	<b>D81.5</b>		Main code here in linearizations.
906	Wiskott-Aldrich syndrome <i>Eczema-thrombocytopenia-immunodeficiency syndrome</i>	<b>D82.0</b>		
169082	Combined immunodeficiency due to CD3 gamma deficiency			
169085	Susceptibility to respiratory infections associated with CD8 alpha chain mutation <i>Familial CD8 deficiency</i>			
169446	Autosomal recessive hyperimmunoglobulin E syndrome <i>Autosomal recessive hyper-IgE syndrome</i> <i>Hyperimmunoglobulin E syndrome type 2</i> <i>Nonskeletal hyper IgE syndrome</i> <i>Autosomal recessive HIES</i> <i>Severe combined immunodeficiency [SCID] due to DOCK8 deficiency</i>  <i>Severe combined immunodeficiency [SCID] due to dedicator of cytokinesis 8 protein deficiency</i>	<b>D82.4</b>		The current ICD-10 D82.4 code is described as "Hyperimmunoglobulin E [IgE] syndrome", and includes all hyper-IgM syndromes. It must be split between autosomal dominant and autosomal recessive forms.
157949	<b>Excludes:</b> Autosomal dominant hyperimmunoglobulin E syndrome Primary immunodeficiency with skin granulomas <i>Combined cellular and humoral deficiencies and multiple granulomas</i>			

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
1493	Corpus callosum agenesis – cataract – immunodeficiency  <i>Dionisi-Vici-Sabetta-Gambarara syndrome</i> <i>Vici syndrome</i>		developmental anomalies	
220465	Laron syndrome with moderate lymphopenia immunodeficiency <i>Laron-like syndrome</i> <i>Short stature due to STAT5b deficiency</i>		endocrinology	
169349	Immuno-osseous dysplasia		bone diseases	
1830	Schimke immuno-osseous dysplasia <i>Spondyloepiphyseal dysplasia - nephrotic syndrome</i> <i>Schimke syndrome</i>		bone diseases	
175	Cartilage-hair hypoplasia <i>Autosomal recessive metaphyseal chondrodysplasia</i> <i>Metaphyseal chondrodysplasia, McKusick type</i>		bone diseases	
2951	Thumb absent - short stature - immune deficiency		developmental anomalies	
221139	Combined immunodeficiency with facio-oculo-skeletal anomalies		developmental anomalies	
101977	<b>Primary immunodeficiencies with predominantly antibody defects</b> <i>Immunodeficiency predominantly affecting antibody production</i>	<b>D80</b>		
183669	Hereditary hypogammaglobulinaemia or agammaglobulinaemia Isolated agammaglobulinaemia	<b>D80.0</b>		
47	X-linked agammaglobulinaemia <i>BTK-deficiency</i> <i>Agammaglobulinaemia, Bruton type</i>	<b>D80.0</b>		
33110	Autosomal recessive agammaglobulinaemia <i>Agammaglobulinaemia, non-Bruton type</i> <i>Autosomal agammaglobulinaemia</i> Syndromic agammaglobulinaemia			
632	Short stature due to growth hormone isolated deficiency with X-linked hypogammaglobulinaemia			
—	Malignant myelodysplasia with hypogammaglobulinaemia			
2268	ICF syndrome <i>Immunodeficiency - centromeric instability - facial anomalies</i> <i>Centromeric instability immunodeficiency syndrome</i>			
83617	Agammaglobulinaemia - microcephaly - craniosynostosis - severe dermatitis			

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
1572	Common variable immunodeficiency disorders CVID <i>Idiopathic immunoglobulin deficiency</i> <i>Primary hypogammaglobulinaemia</i> <i>Primary antibody deficiency</i>	D83		3 possibles causes of CVID : <ul style="list-style-type: none"> <li>• intrinsic B cell defect</li> <li>• intrinsic T cell defect</li> <li>• TNFR deficiency</li> </ul> but this does not define subtypes here : according to the International Union of Immunological Societies states, it is not yet possible to divide CVID by the names of the immune defects, and they are better represented as a group with unknown aetiologies.
183675	Recurrent infections associated with immunoglobulin isotypes deficiency			
69127	Selective deficiency of immunoglobulin A <i>Selective gamma-A-globulin deficiency</i> <i>Immunoglobulin A deficiency</i> <i>IgA deficiency</i> <i>Selective deficiency of IgA</i>	D80.2		
—	Selective deficiency of immunoglobulin G subclasses <i>Selective deficiency of IgG subclasses</i>	D80.3		
—	Selective deficiency of immunoglobulin M <i>Selective deficiency of IgM</i>	D80.4		
2571	X-linked immunoneurologic disorder <i>Woods-Black-Norbury syndrome</i>		neurology	
183666	Hyper-IgM syndrome without susceptibility to opportunistic infections <i>HIGM without susceptibility to opportunistic infections</i>	D80.5		The current ICD-10 D80.5 code is described as "Immunodeficiency with increased immunoglobulin M", and includes all hyper-IgM syndromes. It must be split between those with and without opportunistic infections, with reciprocal exclusions to prevent confusion.
183663	<b>Excludes:</b> Hyper-IgM syndrome with opportunistic infections			
101089	<b>Includes:</b> Hyper-IgM syndrome type 2 <i>Activation-induced cytidine deaminase deficiency</i> <i>Hyper-IgM syndrome due to AID deficiency</i> <i>Hyper-IgM syndrome due to AICDA deficiency</i> <i>HIGM2</i>			
101091	Hyper-IgM syndrome type 4 <i>HIGM4</i>			
101092	Hyper-IgM syndrome type 5 <i>Hyper-IgM syndrome due to uracil N glycosylase</i> <i>Hyper-IgM syndrome due to UNG deficiency</i> <i>HIGM5</i>			

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
169443	Specific antibody deficiency with normal immunoglobulin concentrations and normal number of B cells <i>Antibody deficiency with near-normal immunoglobulins or with hyperimmunoglobulinaemia</i>	<b>D80.6</b>		
70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency			
169139	Transient hypogammaglobulinaemia of infancy	<b>D80.7</b>		
178389	Osteopetrosis - hypogammaglobulinaemia <i>Autosomal recessive osteoclast-poor osteopetrosis with hypogammaglobulinaemia</i> <i>Autosomal recessive osteopetrosis type 7</i>			
3132	Say-Barber-Miller syndrome <i>Microcephaly - hypogammaglobulinaemia - abnormal immunity</i>		developmental anomalies	
2621	Low birth weight - dwarfism - dysgammaglobulinaemia <i>Christian-Rosenberg syndrome</i> Other specified immunodeficiency with predominantly antibody defects	<b>D80.8</b>		
169346	<b>DNA repair defects other than combined T-cell and B-cell immunodeficiencies</b>			
100	Ataxia-telangiectasia <i>Louis-Bar syndrome</i>	<b>G11.3</b>	neurology	
647	Nijmegen breakage syndrome <i>Autosomal recessive nonsyndromal microcephaly with normal intelligence</i> <i>Immunodeficiency - microcephaly - chromosomal instability</i> <i>Microcephaly - immunodeficiency - lymphoreticuloma</i> <i>Ataxia-telangiectasia, variant 1</i> <i>Seemanova syndrome type 2</i> <i>Berlin breakage syndrome</i> <i>AT V1</i> <i>NBS</i>		developmental anomalies	
125	Bloom syndrome <i>BS</i>		developmental anomalies	
1775	Dyskeratosis congenita <i>DKC</i> <i>Zinsser-Engman-Cole syndrome</i>		dermatology	
169355	<b>Immunodeficiency syndromes with autoimmunity</b>			
3261	Autoimmune lymphoproliferative syndrome <i>Canale-Smith syndrome</i> <i>FAS deficiency</i>			
3453	Autoimmune polyendocrinopathy type 1	<b>E31.0</b>	endocrinology	

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Orpha	ICD-11 table draft	ICD-10	Main code when elsewhere	Comments
	<i>Autoimmune polyendocrinopathy - candidiasis - ectodermal dystrophy syndrome</i> <i>Autoimmune hypoparathyroidism - chronic candidiasis - Addison's disease</i> <i>Hypoparathyroidism - Addison's disease - mucocutaneous candidiasis</i> <i>Multiple endocrine deficiency - Addison's disease - candidiasis</i> <i>Autoimmune polyglandular syndrome type 1</i> <i>Autoimmune polyendocrine syndrome type 1</i> <i>APECED syndrome</i> <i>MEDAC syndrome</i> <i>HAM syndrome</i> <i>APS1</i>			
37042	X-linked immune dysregulation – polyendocrinopathy – enteropathy <i>Autoimmune enteropathy type 1</i> <i>IPEX</i>		gastroenterology	
50816	Spondylometaphyseal dysplasia with combined immunodeficiency <i>Roifman-Melamed syndrome</i>		developmental anomalies	
169361	<b>Immune dysregulation diseases with immunodeficiency</b>			
238510	Lymphoproliferative diseases with immunodeficiency			Also classified in autoimmune lymphoproliferative syndrome-related disorders.
—	Autoimmune lymphoproliferative syndrome with recurrent infections			
238505	Autosomal recessive lymphoproliferative disease			
2442	X-linked lymphoproliferative disease	<b>D82.3</b>		
	<i>Duncan syndrome</i> <i>Purtilo syndrome</i> <i>Immunodeficiency following hereditary defective response to Epstein-Barr virus</i>			
158032	Haemophagocytic lymphohistiocytoses <i>Haemophagocytic syndromes</i>			
167	Chediak-Higashi syndrome	<b>E70.3</b>		
79477	GrisCELLI syndrome type 2 <i>Hypopigmentation - immunodeficiency with or without neurologic impairment</i>			
540	Familial haemophagocytic lymphohistiocytosis <i>Hereditary haemophagocytic lymphohistiocytosis</i>			
79430	Hermansky-Pudlak syndrome <i>HPS</i>			
183678	Hermansky-Pudlak syndrome with neutropenia <i>Hermansky-Pudlak syndrome type 2</i> <i>HPS2</i>		constitutional neutropenias (above)	

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
231500	Hermansky-Pudlak syndrome with pulmonary fibrosis <i>HPS with pulmonary fibrosis</i>			
231512	Hermansky-Pudlak syndrome without pulmonary fibrosis <i>HPS without pulmonary fibrosis</i>			
231531	Hermansky-Pudlak syndrome type 7 <i>HPS7</i>			
231537	Hermansky-Pudlak syndrome type 8 <i>HPS8</i>			
—	<b>Other defects in adaptive immunity</b>			
28000	Idiopathic CD4 lymphocytopenia			
228423	Monocytopenia with susceptibility to infections <i>Monocytopenia and mycobacterial infection syndrome</i> <i>MONOMAC</i> <i>Combined immunodeficiency with susceptibility to mycobacterial, viral and fungal infections</i>			
1334	Chronic mucocutaneous candidiasis <i>CMC</i>			
137631	Lung fibrosis - immunodeficiency - gonadal dysgenesis		pneumology	
79124	Hepatic veno-occlusive disease - immunodeficiency <i>VODI syndrome</i>		hepatology	
183716	<b>Complex primary immunodeficiency syndromes</b>			
2314	<b>Autosomal dominant hyperimmunoglobulin E syndrome</b> <i>Autosomal dominant hyper-IgE syndrome</i> <i>Hyperimmunoglobulin E-recurrent infection syndrome</i> <i>Hyperimmunoglobulin E syndrome type 1</i> <i>Autosomal dominant HIES</i> <i>Buckley syndrome</i> <i>Job syndrome</i> <i>AD-HIES</i>	<b>D82.4</b>		The current ICD-10 D82.4 code is described as "Hyperimmunoglobulin E [IgE] syndrome", and includes all hyper-IgE syndromes. It must be split between the autosomal dominant form (among <i>Primary immunodeficiency with predominantly antibody defects</i> ) and the autosomal recessive form (among <i>Combined immunodeficiencies</i> ).
169446	<b>Excludes :</b> Autosomal recessive hyperimmunoglobulin E syndrome			

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<b>Orpha</b>	<b>ICD-11 table draft</b>	<b>ICD-10</b>	<b>Main code when elsewhere</b>	<b>Comments</b>
302	<b>Epidermodysplasia verruciformis</b> <i>Lewandowsky-Lutz syndrome</i>		dermatology	
98813	<b>EDA-ID syndrome</b> <i>Hypohidrotic ectodermal dysplasia with immunodeficiency</i>		dermatology	
69088	<b>OL-EDA-ID syndrome</b> <i>Anhidrotic ectodermal dysplasia - immunodeficiency - osteopetrosis – lymphoedema</i>		bone diseases	
51636	<b>WHIM syndrome</b> <i>Warts-hypogammaglobulinaemia-infections-myelokathexis</i>			

## Acquired immunodeficiencies

178996	<b>Acquired neutropenias</b> <i>Immunologic neutropenias</i>			
47612	<b>Felty syndrome</b>			
86872	<b>T-cell large granular lymphocyte leukaemia</b> <i>T-cell chronic lymphocytic leukaemia</i> <i>T-cell LGL leukaemia</i>			
—	<b>Secondary agranulocytosis</b>		<b>D70</b>	
—	Toxic secondary agranulocytosis			
—	Drug-induced secondary agranulocytosis			
2688	<b>Adult idiopathic neutropenia</b>		<b>D70</b>	

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Orpha	ICD-11 table draft	ICD-10	Main code when elsewhere	Comments
—	<b>Human immunodeficiency virus [HIV] disease</b>	<b>B20-B24</b>	infectiology	<b>This whole section in grey is not revised by Rare Diseases TAG.</b>  Consider a double classification both in infectiology and immunology. Includes the spectrum of human immunodeficiency virus infection that ranges from asymptomatic seropositivity, through AIDS-related complex (ARC), to acquired immunodeficiency syndrome (AIDS).
—	<b>Human immunodeficiency virus [HIV] disease resulting in infectious and parasitic diseases</b>	<b>B20</b>	infectiology	
—	HIV disease resulting in mycobacterial infection	<b>B20.0</b>	infectiology	
—	HIV disease resulting in tuberculosis		infectiology	
—	HIV disease resulting in other bacterial infections	<b>B20.1</b>	infectiology	
—	HIV disease resulting in cytomegaloviral disease	<b>B20.2</b>	infectiology	
—	HIV disease resulting in other viral infections	<b>B20.3</b>	infectiology	
—	HIV disease resulting in candidiasis	<b>B20.4</b>	infectiology	
—	HIV disease resulting in other mycoses	<b>B20.5</b>	infectiology	
—	HIV disease resulting in Pneumocystis jirovecii pneumonia	<b>B20.6</b>	infectiology	
—	HIV disease resulting in multiple infections	<b>B20.7</b>	infectiology	
—	HIV disease resulting in other infectious and parasitic diseases	<b>B20.8</b>	infectiology	
—	HIV disease resulting in unspecified infectious or parasitic disease	<b>B20.9</b>	infectiology	
—	<b>Human immunodeficiency virus [HIV] disease resulting in malignant neoplasms</b>	<b>B21</b>	infectiology	
—	HIV disease resulting in Kaposi sarcoma	<b>B21.0</b>	infectiology	
—	HIV disease resulting in Burkitt lymphoma	<b>B21.1</b>	infectiology	
—	HIV disease resulting in other types of non-Hodgkin lymphoma	<b>B21.2</b>	infectiology	
—	HIV disease resulting in other malignant neoplasms of lymphoid, haematopoietic and related tissue	<b>B21.3</b>	infectiology	
—	HIV disease resulting in multiple malignant neoplasms	<b>B21.7</b>	infectiology	
—	HIV disease resulting in other malignant neoplasms	<b>B21.8</b>	infectiology	
—	HIV disease resulting in unspecified malignant neoplasm	<b>B21.9</b>	infectiology	
—	<b>Human immunodeficiency virus [HIV] disease resulting in other specified diseases</b>	<b>B22</b>	infectiology	
—	HIV disease resulting in encephalopathy	<b>B22.0</b>	infectiology	
—	Dementia in human immunodeficiency virus [HIV] disease	<b>F02.4</b>		
—	HIV disease resulting in lymphoid interstitial pneumonitis	<b>B22.1</b>	infectiology	
90081	HIV disease resulting in wasting syndrome	<b>B22.2</b>	infectiology	
—	<i>Slim disease</i>			
—	<i>HIV disease resulting in failure to thrive</i>			
—	HIV disease resulting in multiple diseases classified elsewhere	<b>B22.7</b>	infectiology	

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Orpha	ICD-11 table draft	ICD-10	Main code when elsewhere	Comments
—	<b>Human immunodeficiency virus [HIV] disease resulting in other conditions</b>	<b>B23</b>	infectiology	
—	Acute HIV infection syndrome	<b>B23.0</b>	infectiology	
—	HIV disease resulting in (persistent) generalized lymphadenopathy	<b>B23.1</b>	infectiology	
—	HIV disease resulting in haematological and immunological abnormalities, not elsewhere classified	<b>B23.2</b>	infectiology	
—	HIV disease resulting in other specified conditions	<b>B23.8</b>	infectiology	
—	<b>Unspecified human immunodeficiency virus [HIV] disease</b>	<b>B24</b>	infectiology	
—	<b>Diseases of thymus</b>	<b>E32</b>		Myasthenia gravis is ultimately to be classified here as well as in neurology.
—	<i>Excludes:</i> Thymic aplasia or hypoplasia with immunodeficiency [Monosomy 22q11 and Nezelof syndrome]	<b>D82.1</b>	primary immunodeficiencies (above)	
589	<i>Excludes:</i> Myasthenia gravis	<b>G70.0</b>	neurology	
—	<b>Persistent hyperplasia of thymus</b>	<b>E32.0</b>		
—	<b>Hypertrophy of thymus</b>			
—	<b>Abscess of thymus</b>	<b>E32.1</b>		
100100	<b>Thymic tumours</b>			The whole group is also to be included in oncology.
3398	<b>Thymic epithelial tumour</b>			
99867	Thymoma			
99868	Thymic carcinoma			
99869	Thymus malignant tumour			
97289	<b>Thymic endocrine tumour</b> <i>Carcinoid tumour of the thymus</i> <i>Thymic neuroendocrine tumour</i>			Also included in endocrinology.
169105	<b>Good syndrome</b> <i>Thymoma-immunodeficiency</i>			

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Orpha	ICD-11 table draft	ICD-10	Main code when elsewhere	Comments
—	<b>Other disorders involving the immune system</b>	<b>D89</b>		
—	<b>Excludes:</b> Monoclonal gammopathy	<b>D47.2</b>	haematology	
—	<b>Excludes:</b> Transplant failure and rejection [including Graft versus host disease and Anti-HLA hyperimmunization]	<b>T86</b>	complications of surgical and medical care	
95431	<b>Excludes:</b> Twin to twin transfusion syndrome		neonatology	
—	<b>Chronic neutrophilia</b>			
—	Hereditary chronic neutrophilia			
—	Acquired chronic neutrophilia			
—	<b>Benign hypergammaglobulinaemic purpura</b>	<b>D89.0</b>		
	<i>Polyclonal hypergammaglobulinaemia</i>			
	<i>Polyclonal gammopathy</i>			
91139	<b>Simple cryoglobulinaemia</b>	<b>D89.1</b>		
	<i>Cryoglobulinaemia type 1</i>			
	<i>Monoclonal cryoglobulinaemia</i>			
91138	<b>Excludes:</b> Mixed cryoglobulinaemia	<b>D89.1</b>	angiology	
91378	<b>Hereditary angioedema</b>	<b>D84.1</b>	multisystemic diseases	Hereditary angioedema is included in this chapter since patients are seen in immunology clinics and treated with a blood product.
	<i>Hereditary non histamine-induced angioedema</i>			
	<i>Hereditary bradykinine-induced angioedema</i>			
	<i>Hereditary angioneurotic oedema</i>			
	<i>Familial angioneurotic oedema</i>			
	<i>HAE</i>			
100050	<b>Hereditary angioedema type 1</b>			
	<i>Hereditary angioneurotic oedema type 1</i>			
	<i>HAE-I</i>			
	<i>C1 esterase inhibitor [C1-INH] quantitative deficiency</i>			
100051	<b>Hereditary angioedema type 2</b>			
	<i>Hereditary angioneurotic oedema type 2</i>			
	<i>HAE-II</i>			
	<i>C1 esterase inhibitor [C1-INH] qualitative deficiency</i>			

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Orpha	ICD-11 table draft	ICD-10	Main code when elsewhere	Comments
100054	<b>Hereditary angioedema type 3</b> <i>Hereditary angioneurotic oedema type 3</i> <i>HAE-III</i> <i>Inherited estrogen-associated angioedema</i> <i>Inherited estrogen-associated angioneurotic oedema</i> <i>Inherited estrogen-dependent angioedema</i> <i>Inherited estrogen-dependent angioneurotic oedema</i>			
—	<b>Autoimmune lymphoproliferative syndrome-related disorders</b>			
—	<b>Diazani autoimmune lymphoproliferative disease</b>			
—	<b>RAS-associated autoimmune lymphoproliferative disease</b>			
238510	<b>Lymphoproliferative diseases with immunodeficiency</b>			Also classified in immune dysregulations disorders with immunodeficiency.
—	Autoimmune lymphoproliferative syndrome with recurrent infections			
2442	X-linked lymphoproliferative disease <i>Duncan syndrome</i> <i>Purtilo syndrome</i>			
	<i>Immunodeficiency following hereditary defective response to Epstein-Barr virus</i>			
238505	Autosomal recessive lymphoproliferative disease			
—	<b>Autoimmune diseases</b>			

SECTION IN CONSTRUCTION