

ICD revision process – Rare Diseases TAG fact sheet

Composition and organization of Rare Diseases TAG

- As Rare Diseases are disseminated across ICD chapters, the way the TAG was composed and the way it works is somehow different from other TAG.
- The TAG Rare Diseases was composed as to represent large regions of the world, each representative of a region being in charge of identifying the relevant experts and groups of experts to involve for revising a given chapter.
- The TAG members are the following:
 - Europe: Ségolène Aymé (Chair) and Ana Rath, INSERM/Orphanet
 - USA: Stephen Groft, Office of Rare Diseases, NIH
 - Brazil: Eduardo Castilla, ECLAMC/FIOCRUZ
 - Russia : Evgeny Ginter, Russian Academy of Medical Sciences, Moscow
 - Philippines : Carmencita Padilla, University of the Philippines – Manila
 - Australia: Julie McGaughran, Genetic Health Queensland, Brisbane
- The secretariat of the TAG is ensured by Bertrand Bellet, INSERM/Orphanet, Paris

General principles followed in ICD revision proposals for rare diseases

- Preparatory work is performed at Orphanet (INSERM SC11, Paris, France). A grant from the European Commission (DG Sanco) permitted to collect published classifications, to establish an inventory of rare diseases (accessible at www.orpha.net), to index rare diseases with ICD10 codes, OMIM codes and genes, and to develop fact sheets on over 2,500 diseases.
- ICD10 is systematically compared with published classifications and with the Orphanet classification system. Both necessary changes in the classification structure and necessary adds of new rare diseases are identified and organised in a document considered as the first draft of a revised chapter.
- Revised chapters follow a **primarily clinical approach**, only secondarily an aetiological one.
- When several possible names are possible for a disease, **descriptive names formed in accordance with a clinical approach** are preferred.
- Every entity is assigned a **unique identifying number**.
- Rare diseases affecting several body systems are included in every relevant chapter, as ICD11 will be **polyaxial**; but a **main code** is selected (to allow for linearisations) according to the most severe involvement and/or the specialist most likely to be relied on for the management of the disease. In some cases, the choice is open for debate.
- ICD10-style **exclusion notes** are kept when used to clarify the range of content of particular codes, or remove possible ambiguities.
- ICD10-style "default codes" sections (current .8 and .9) are kept in proposals. **Specific default codes are created for rare diseases**, to allow them to be identified as such in order to improve statistics about rare diseases.
- A new dedicated chapter in ICD11 for **multisystemic diseases** will be proposed
- When ready, revised chapters are sent by Orphanet to other TAG members. Each of them sends them around to appropriate experts and expert groups.
- The revised chapters are also sent to other TAG as a contribution to the revision of their own chapter.

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Current and provisional workflow

White areas indicate ongoing or finished tasks. Shaded areas indicate tasks still to be completed.

Chapters	1st draft		2nd draft		Implementation in WHO platform
	Preparation	Expert feedback	Preparation	Expert validation	
Haematology	03-2009	received	12-2009	03-2010	03-2010
Endocrinology	05-2009	received	01-2010	04-2010	04-2010
Nutrition	07-2009	received	02-2010	05-2010	05-2010
Metabolism	09-2009	received / delayed	03-2010	06-2010	06-2010
Immunology	09-2009	received	04-2010	07-2010	07-2010
Neurology	01-2010	04-2010	05-2010	09-2010	09-2010
Dysmorphology	02-2010	05-2010	06-2010	09-2010	09-2010
Multisystemic diseases *	03-2010	06-2010	07-2010	10-2010	10-2010
Eye and ear	04-2010	07-2010	09-2010	12-2010	12-2010
Cardiology- angiology	05-2010	09-2010	10-2010	01-2011	01-2011
Pneumology	06-2010	10-2010	11-2010	02-2011	02-2011
Gastroenterology- Hepatology	07-2010	11-2010	12-2010	03-2011	03-2011
Dermatology	09-2010	12-2010	01-2011	04-2011	04-2011
Urology- Gynaecology	10-2010	01-2011	02-2011	05-2011	05-2011

* Expanded from the current rheumatology chapter (ICD10 ch. 13).