

WHO international classification of diseases (ICD) revision process: incorporating rare diseases into the classification scheme: state of the art

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The problem:

- Most rare genetic diseases are absent from the current International Classification of Diseases (ICD-10)
- Those with a specific code (< 240) are often misclassified
- This results in a lack of visibility and traceability of rare diseases in medical information systems

The opportunity:

- The WHO has started the revision process for the ICD to establish a future version, ICD-11
- A Topic Advisory Group for Rare Diseases (TAG-RD) is led by Orphanet
- Other TAGs deal with each specific chapter
- The Alpha-draft should be ready in 2010

This will increase the visibility of rare diseases in medical information systems

ORPHANET preparatory work

The revision process for rare diseases

- Step 1:** Comparison of ICD-10 with Orphanet classification
- Step 2:** Preparation of a new version including rare diseases using existing ICD-10 chapters
- Step 3:** Submission of new chapters to expert groups and other TAGs
- Step 4:** Incorporation of received comments into a proposal
- Step 5:** Transmission to the WHO of the alpha-draft of each chapter

The revision process is open to everyone:
<https://extranet.who.int/icdrevision>

Example of a revised chapter

ICD-10 → ICD-11 draft

Constitutional megaloblastic anaemias due to vitamin B12 metabolism disorder
Vitamin B12 deficiency anaemia due to congenital intrinsic factor deficiency
 (Congenital pernicious anaemia)
Vitamin B12 deficiency anaemia due to selective vitamin B12 malabsorption with proteinuria
 (Imerslund(-Gräsbeck) syndrome)
Congenital megaloblastic anaemia due to transcobalamin II deficiency
Methylcobalamin deficiency type cbl E
Methylcobalamin deficiency type cbl G
Methylmalonic acidaemia - homocystinuria
 - Methylmalonicacidaemia - homocystinuria, type cbl C
 - Methylmalonicacidaemia - homocystinuria, type cbl D
 - Methylmalonicacidaemia - homocystinuria, type cbl F
Other specified rare constitutional megaloblastic anaemia due to vitamin B12 metabolism disorder

Acquired megaloblastic anaemias due to vitamin B12 deficiency
Acquired megaloblastic anaemia due to vitamin B12 deficiency secondary to intrinsic factor deficiency
 (Biermer disease, Pernicious anaemia)
Excludes: Vitamin B12 deficiency anaemia due to congenital intrinsic factor deficiency
Acquired megaloblastic anaemia due to nutritional vitamin B12 deficiency
 (Acquired megaloblastic anaemia due to dietary vitamin B12 deficiency)
 - Vegan anaemia
 - Nutritional vitamin B12 deficiency due to maternal vitamin B12 deficiency
 • Congenital nutritional vitamin B12 deficiency due to maternal vitamin B12 deficiency
 • Nutritional vitamin B12 deficiency due to maternal vitamin B12 deficiency in breast-fed infants
Other specified acquired anaemias due to vitamin B12 deficiency
 - Other specified rare acquired anaemias due to vitamin B12 deficiency

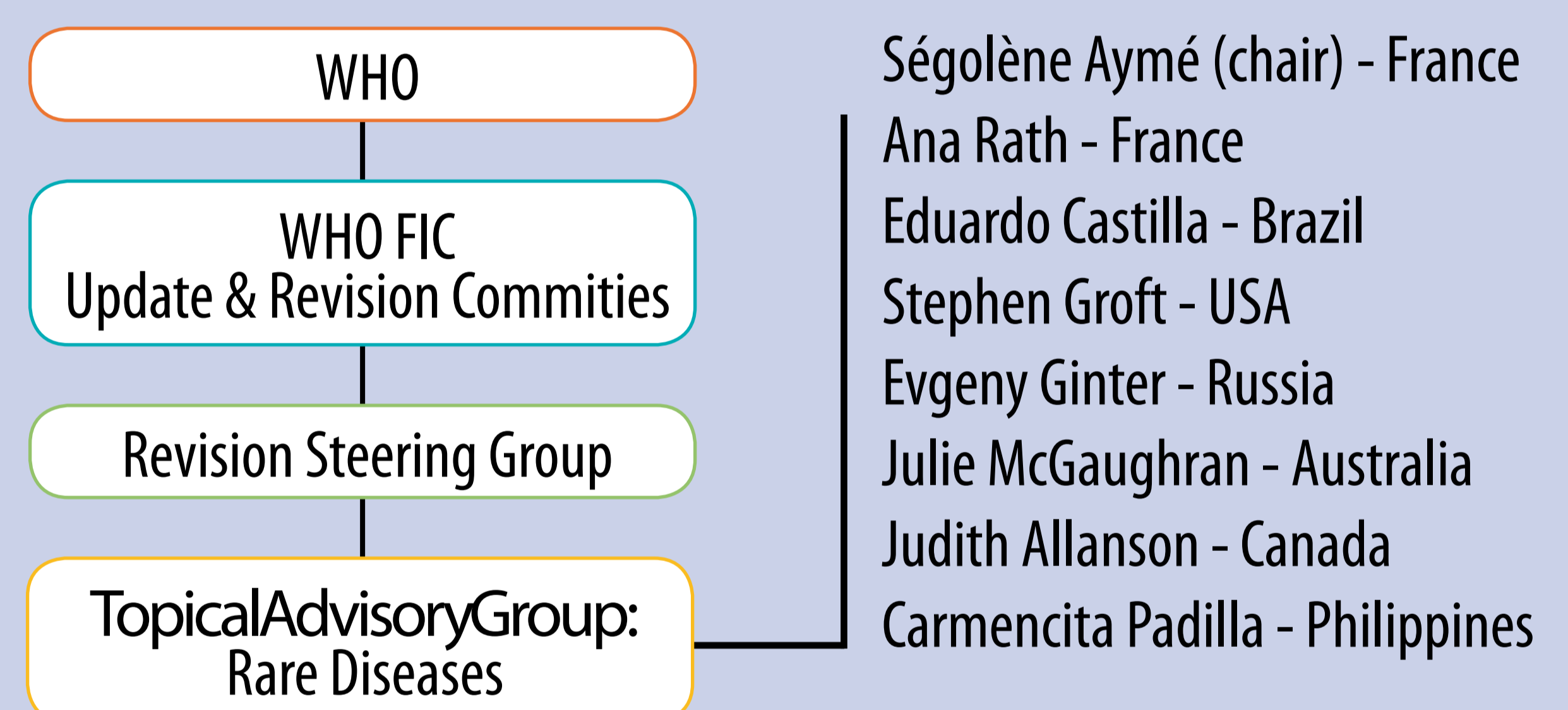
D51 Vitamin B12 deficiency anaemia
Excludes: Vitamin B12 deficiency (E53.8)
 D51.0 Vitamin B12 deficiency anaemia due to intrinsic factor deficiency
 - Anaemia:
 - Addison
 - Biermer
 - pernicious (congenital)
 - Congenital intrinsic factor deficiency
 D51.1 Vitamin B12 deficiency anaemia due to selective vitamin B12 malabsorption with proteinuria
 - Imerslund(-Gräsbeck) syndrome
 - Megaloblastic hereditary anaemia
 D51.2 Transcobalamin II deficiency
 D51.3 Other dietary vitamin B12 deficiency anaemia
 - Vegan anaemia
 D51.8 Other vitamin B12 deficiency anaemias
 D51.9 Vitamin B12 deficiency anaemia, unspecified

WHO ICD over time

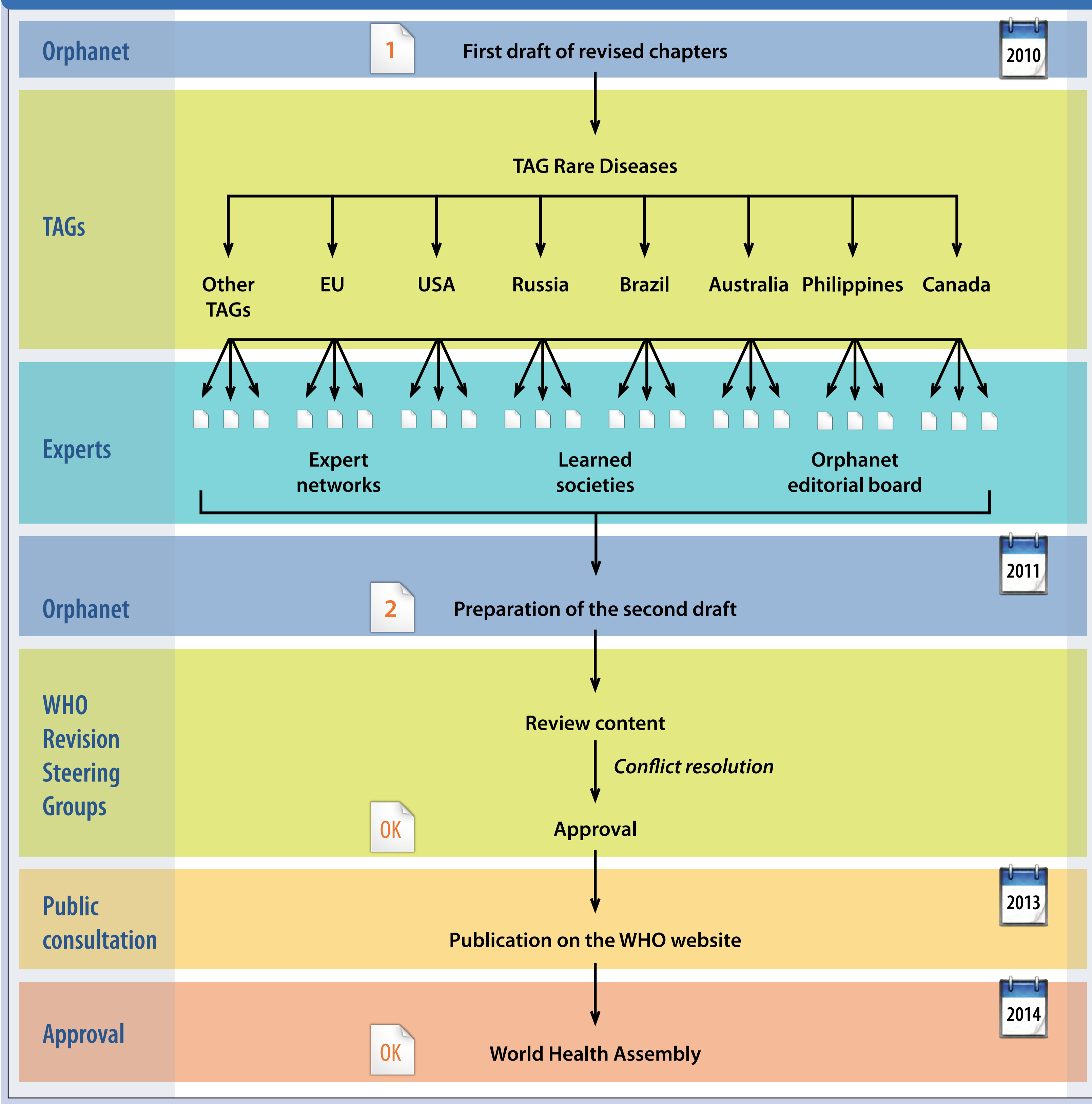


WHO ICD, a worldwide effort

ICD-10 Revision organisation structure



The revision process for rare diseases: from ICD-10 to ICD-11



Take part in the ICD revision process for the chapters that concern your speciality:
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