



Classification of Rare Diseases

A worldwide effort to input on the
**International Classification of
Diseases**

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Major problems with ICD10

- Only 240 rare diseases with a specific code
- No visibility of rare diseases in health information systems
- Lack of systematic approach
 - Classification according to major symptom
 - Classification according to aetiology / mechanism
- Confusion between anatomy / organs and Systems
- Confusion between « malformation » and « congenital » and « genetic »

ICD11 : Topic Advisory Group for Rare Diseases (**RD-TAG**),
chair: Ségolène Aymé (**Orphanet**)

ORPHANET previous work

Direct online access to a comprehensive nomenclature of rare diseases, including genetic diseases in a relational database

A unique identifier stable in time

Epidemiological data

The screenshot shows the Orphanet website interface. At the top, it says 'The portal for rare diseases and orphan drugs'. Below that is a navigation menu with tabs for 'Home', 'Search by sign', 'Classification', 'Genes', 'New therapies for patients', 'Epidemiology for professionals', and 'Orphanet website'. The main content area is titled 'Joubert syndrome' and includes a 'SIMPLE SEARCH' section with a search box and radio buttons for 'Exact term', 'Matched in word', 'All words', and 'All in code'. To the right, there is a 'SIMPLE SEARCH OPTIONS' section with a 'Refinement' button. Below the search options, there is a table with columns for 'MESH terms', 'ICD codes', and 'OMIM numbers'. The table lists 'Joubert syndrome' with corresponding codes. A 'SUMMARY' section is also visible, providing a brief description of the syndrome.

Mode of inheritance

Signs and symptoms

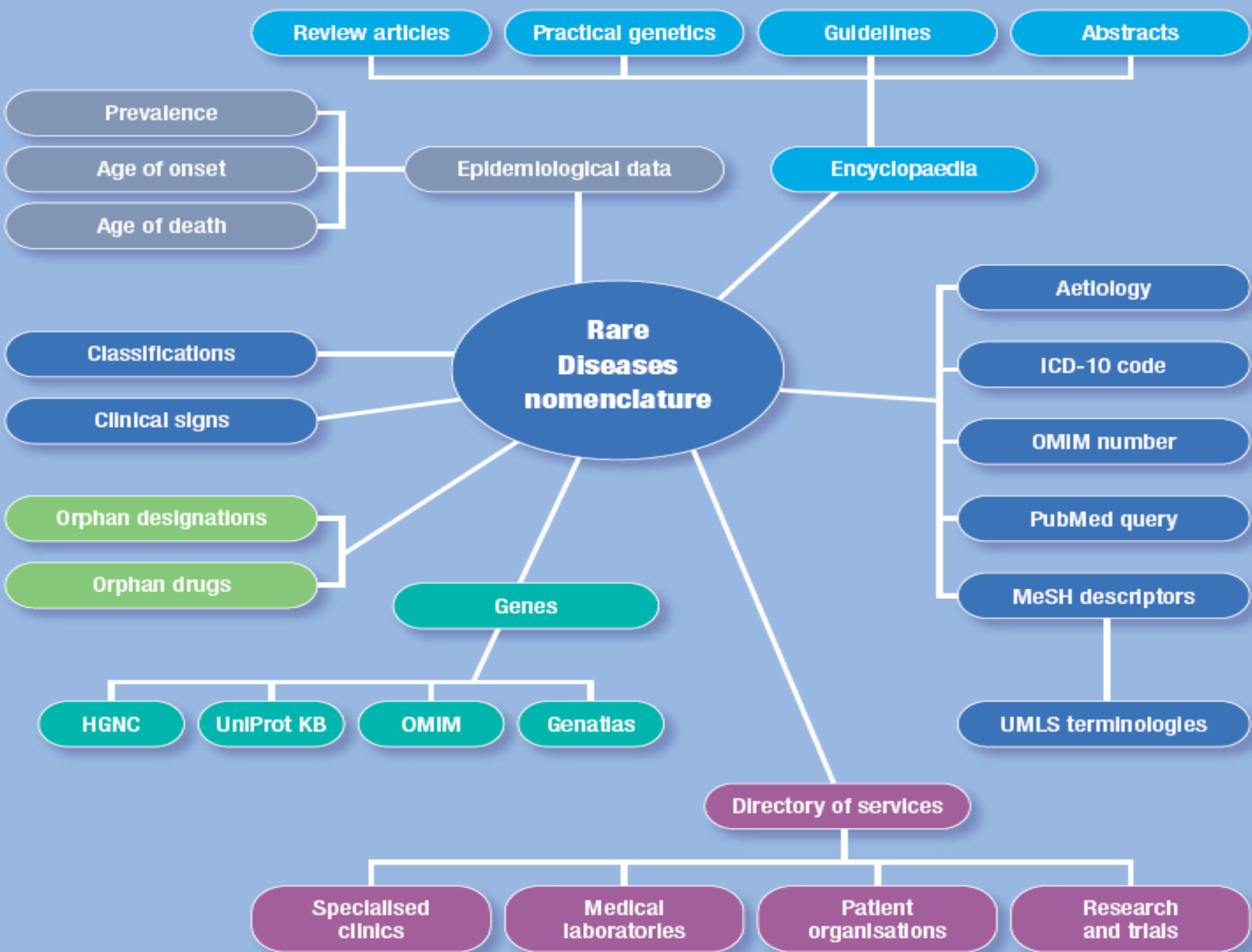
Genes involved in

Classified in a multi-hierarchical, multi-axial classification system

- ✓ Clinical axis
- ✓ Aetiological axis
- ✓ ...

Indexed with

- ✓ MeSH terms
- ✓ ICD codes
- ✓ OMIM numbers
- ✓ Other medical terminologies ongoing



Orphanet directory of diseases

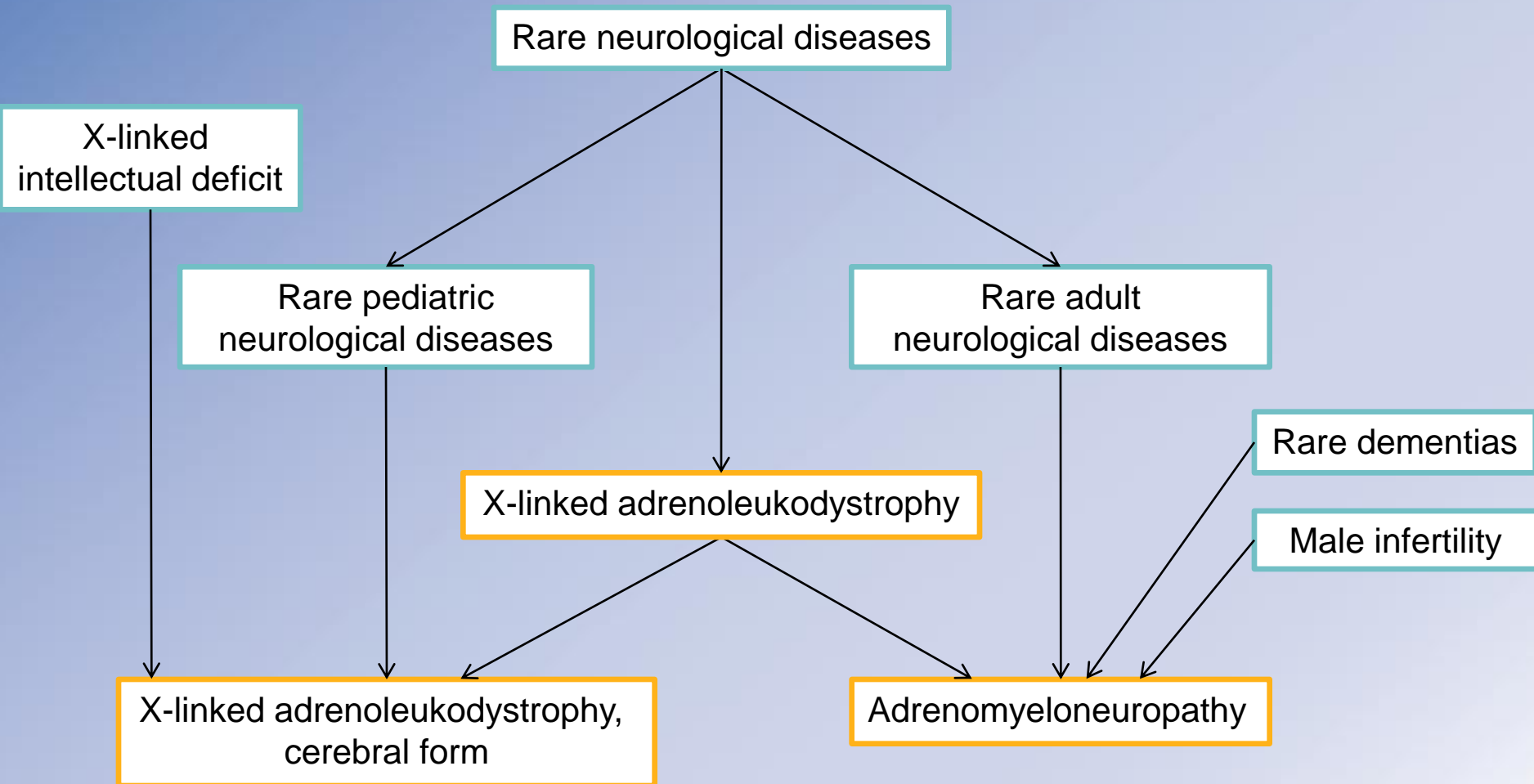
- **Comprehensive list of rare diseases: over 6,000**

- Identity card + genes
- Unique Orpha number
 - ✓ Stable despite the evolution of knowledge
 - ✓ Linked to parent and child diseases in every classification
- **Files available on request**
- Suitable to code clinical activity / lab activity in information systems

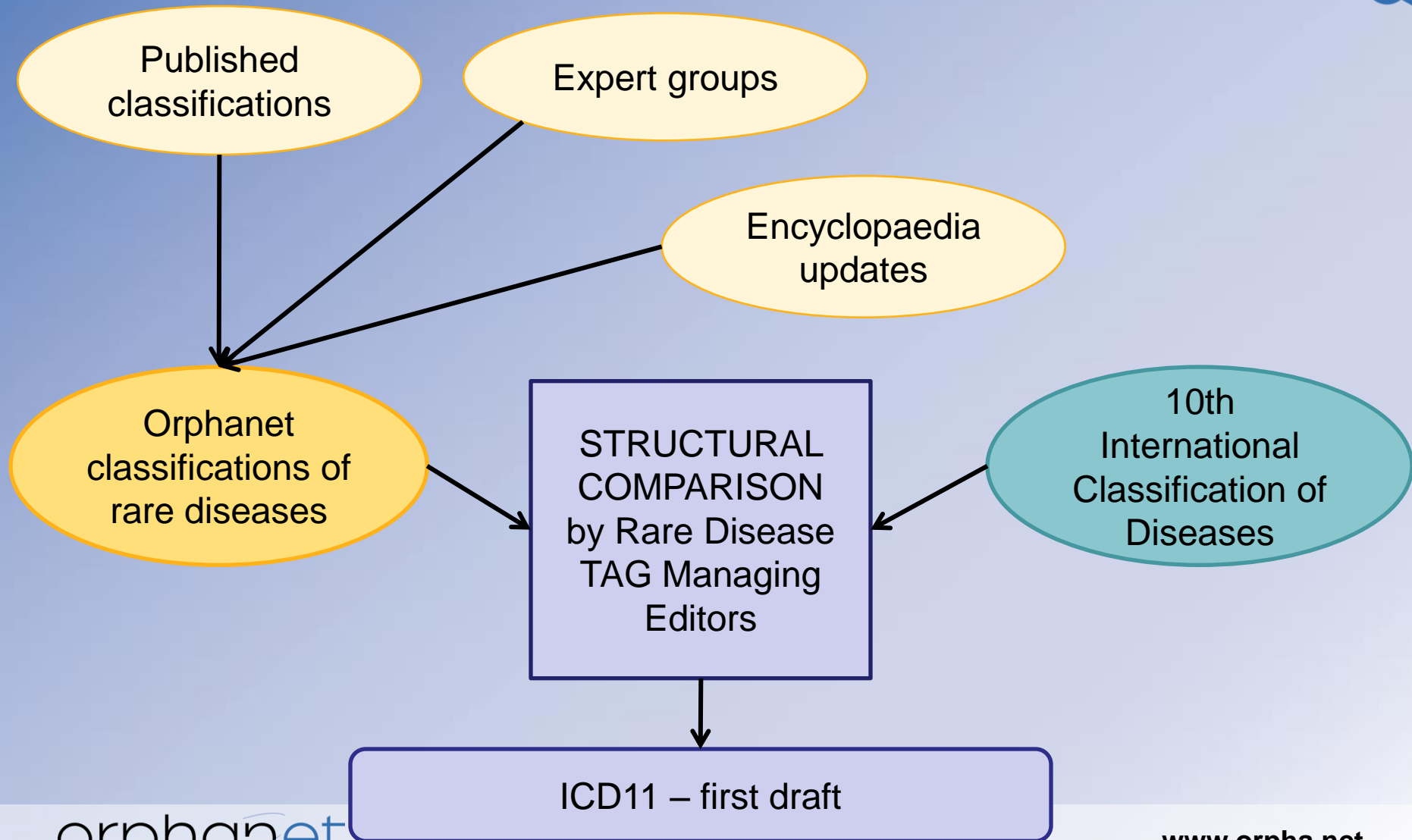
- **Classifications of rare diseases**

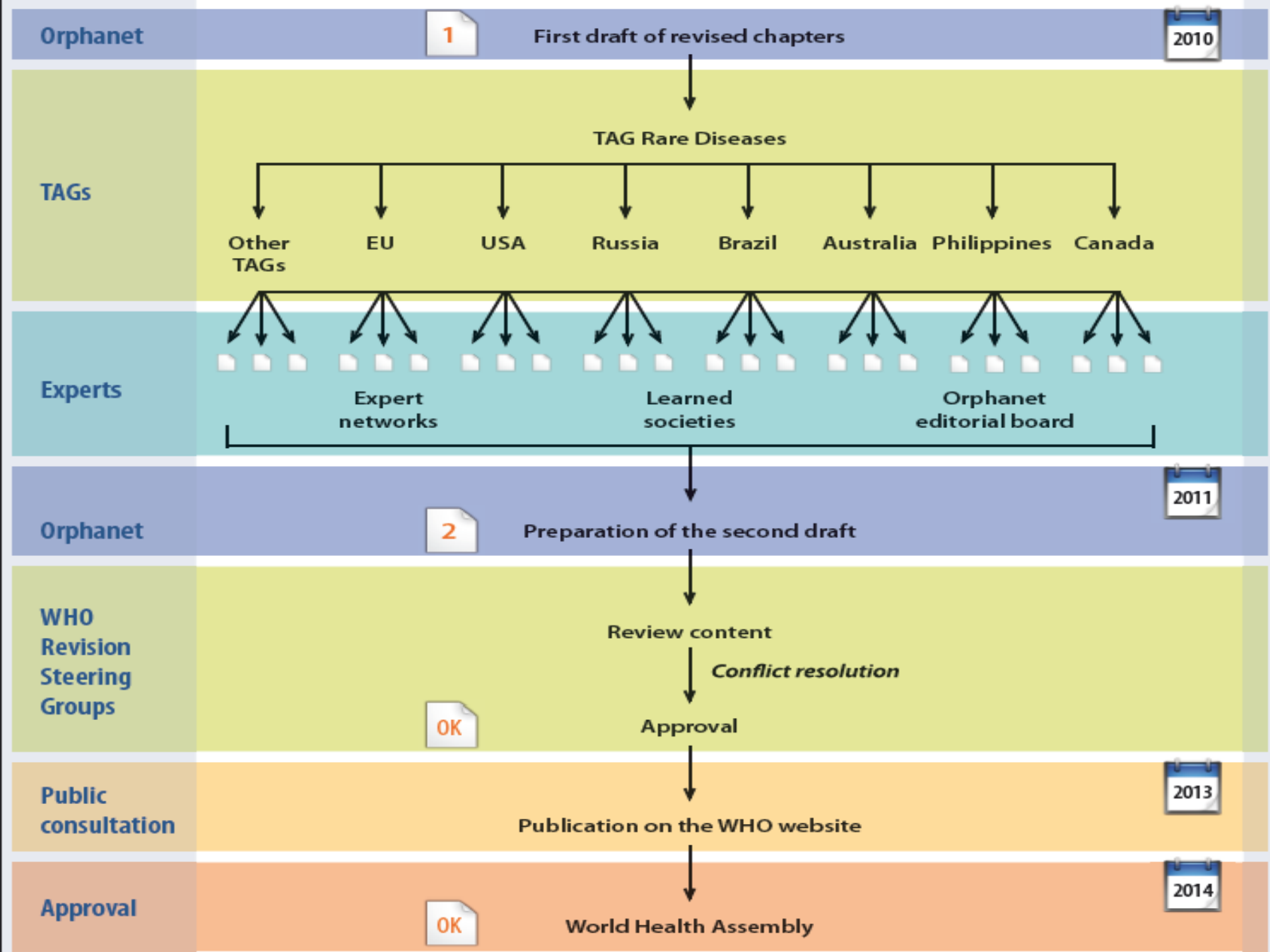
- List of classifications of rare diseases
- Visualisation of each classification
- Possibility to click at any level to obtain detailed information

Orphanet classification of rare diseases is a multihierarchical system that is able to reflect all levels of granularity, from the most general (categories) to the most particular (sub-types), and to follow the evolution of knowledge.



Contribution of Orphanet to the revision process of the ICD





Basis for a revision in the field of rare diseases

- All rare diseases should be finally listed in the Index of ICD11
- Rare Diseases serve as a model to shape the future structure of ICD11 as they are lower nodes in any hierarchy and in all fields in Medicine
- Criterion: medical/surgical specialties involved in the management of the different manifestations of a disease
- Stick to medical specialties organised by system
- Etiological (i.e. genes)/physiopathological criteria further applied, when relevant for diagnosis and/or management

Proposed organisation of chapters

- By system
 - Based on physiology
 - Etiology/mechanism being the final level
 - From the « upper level » to the « lower level »
- Addition of a chapter for mutisystemic diseases
 - Ex: Marfan syndrome is a multisystemic disease
- Chapter for developmental defects of antenatal origin (not only malformations) as in utero development is a process- a « system »)
- Separation of « constitutional » and « acquired »

Contribution to ICD revision process

- Haematological diseases V2 sent
- Endocrinological diseases V2 sent
- Nutritional diseases V1 under revision
- Metabolic diseases V1 under revision
- Immunological diseases V1 under revision
- Neurological diseases V1 under revision
- Malformations/syndromes V1 in progress
- Multisystemic diseases
- Dermatology
- Nephrology
- Ophthalmology

Revised chapters are available in
www.eucerd.eu

Conclusion

- Possibility to propose a profound evolution of the organisation of chapters II to XVIII
 - With a possible migration of almost all existing codes
 - With a common logics applied to all chapters
 - Putting rare/genetic diseases where they should be
 - Everywhere as a lower node
- Provide Orphanet with names of experts willing to review the chapter of their specialty
- Send to Orphanet established classifications to consider
- Promote the Orphanet nomenclature of rare diseases to be used by Information systems to better trace rare diseases

Thank you for your attention !