ICD10 revision process and rare diseases

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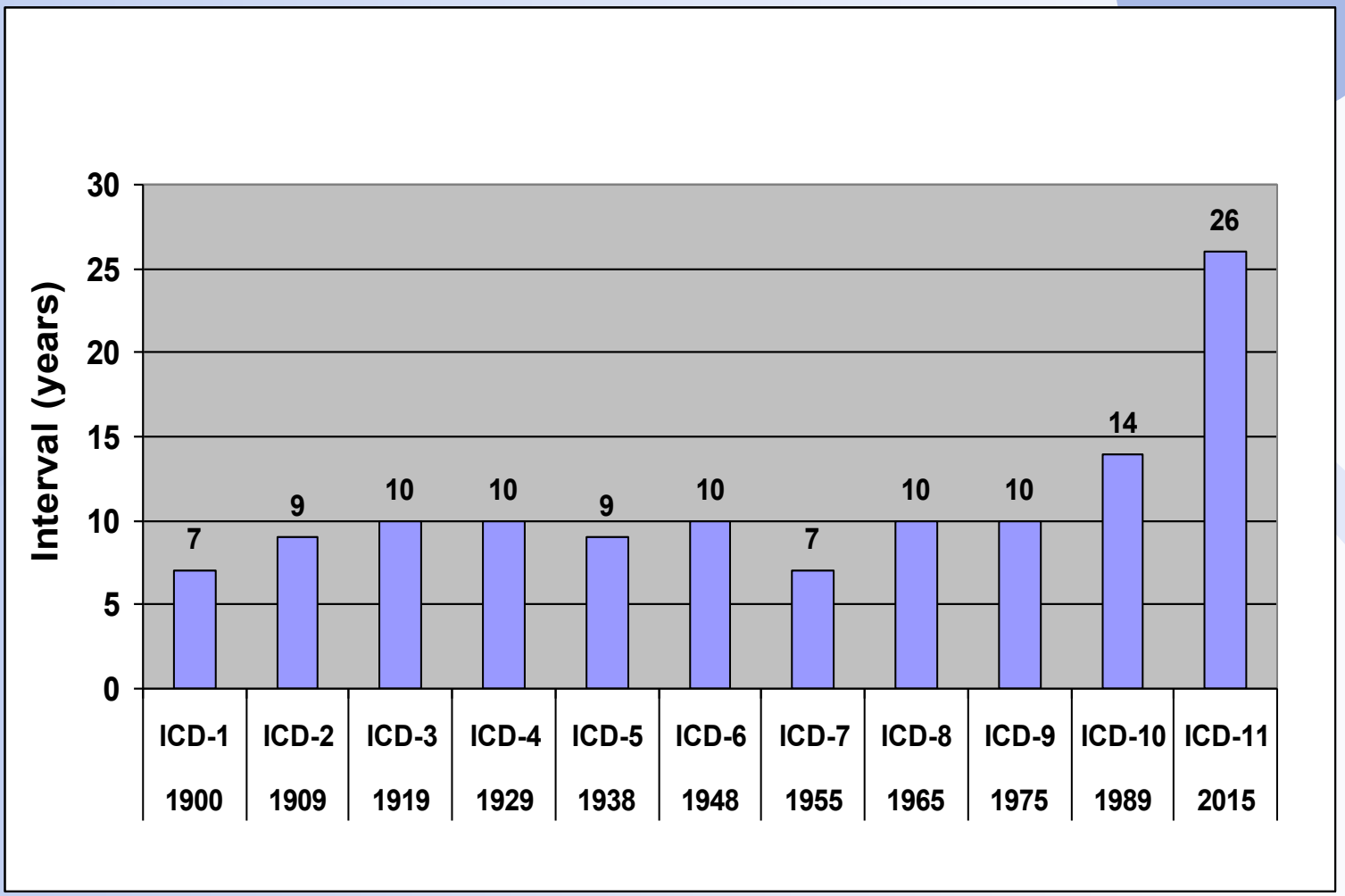
on Rare Diseases

ICD Revisions



ICD Revision Process

- Drafting
  - Taxonomic Guidelines
  - Definition, Diagnosis and Indexing / mapping guidelines
- Overall Structure
- Individual Chapters
- Overseeing the **TOTAL ICD**
  - ALPHA Draft – structured comments
  - BETA Draft – field testing
- Final Draft

➢**WHA Approval**

ICD Revision Work Streams

1. **Scientific Stream**
   - Evidence Based Reviews, Meta analyses
   - Surveys,Validation Studies
   - Add-on protocols for existing studies

2. **Clinical Stream**
   - Clinical utility – linkage to patient reports
   - Treatment Response
   - Phenotypes: gene to behaviour specs

3. **Public Health Stream**
   - Impact on Health Systems –society –service delivery
   - Resource management -reimbursement - accounting
   - IT applications - terminology

Core Classification issues

1. **Definition of the classification entity:**
   - *medical disease, disorder (syndrome), injury, sign, symptom, …*
2. **Clustering of signs, symptoms, & operational features**
3. **Link to underlying pathophysiology & genetic markers**
4. **Clinical utility of the classification entity**
5. **Reliability of the classification entity**
6. **Validity of the classification entity**
7. **Separation of disease and disability elements**
8. **Cultural elements that need to be attended**
9. **Threshold considerations**
10. **Other nosological issues relevant to this disorder**

ICD Revision Applications

As a part of **ICD Knowledge Portal** three main applications:

1. **ICD-10 +** Application
2. **ICD-11 Draft** Creation
3. *(ICD – Terminology/Ontology Tools)**

* *Possibly for display – not directly for WEB entry*

ICD Revision Applications

1. **ICD-10 +** Application

- Designated **Scientific Group Review**
  - Systematic reviews
    - Scientific, Clinical, Public Health Streams
  - Taxonomic rules & definitions

- Open Comments and suggestions
  - **Periodic Continuous Structured peer review**
    - » requested by WHO
  - Open to whole world – all users

ICD Revision Applications

2. ICD-11 Draft

- Codes
  - Inclusions *(all historical links, index terms)*
  - Exclusions
- Definition of the entity
  - Disease, disorder, injury, syndrome, sign, symptom
  - Level of use (Primary Care, Clinical Care, Research)
  - Glossary description
  - Taxonomic ontology status
- Diagnostic Criteria for the entity
  - Clinical and/or research rules for diagnosis

Composition of TAG

- Europe
  - Ségolène Aymé (TAG chair), Ana Rath (Orphanet)
- North America
  - Stephen Groft (Office of RD-NIH)
  - Roberta Pagon (GeneClinics, University of Seatle)
- South America
  - Eduardo Castilla (Clearinghouse of birth defects, Brazil)
- Australia
  - Agnes Bankier (Possum, Murdoch Institute)
- Asia
  - Hyun-Young Park (NIH, Genetic and rare diseases center, Seoul)

What is ongoing

- Chapter by chapter comparison between
  - ICD-10
  - Orphanet classification
  - Published classifications (when available)
- List of proposals for ICD-10+
- Proposal for ICD-11 for the chapter
  - An information scientist was recruited to assist submitting proposals (contract RDTF secretariat 2009-2011)

Orphanet analysis

- Lack of systematic approach
  - Classification according to major symptom
  - Classification according to aetiology / mechanism
- Confusion between anatomy / organs and Systems

  Respiratory system, cardiovascular system, immunological system……
- Confusion between « malformation » and « congenital » and « genetic »

Proposal for general principles

Based on published classifications
and on past experience in coding
at Orphanet

orphanet

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 prenatal developmental defects (not only malformation) as in utero development is a process- a « system »)

ICD-11 proposals

- A dossier with the rationale for proposals is established
- The definitional items chart (WHO) is fullfilled for each disease
- The dossier is submitted
  - to identified best experts by Orphanet and by other TAG members
- A final proposal will be sent to WHO, chapter by chapter, one every month

ICD10+ proposals

- Each proposal is
  - qualified following the WHO revision tool
  - justified (literature)
- Orphanet input on the ICD10 revision
  - based on already validated subclassifications
- Experts for the specialty (Official networks / Societies / Associations)
  - informed in order to add their contributions to the revision process

Networks of experts in Europe

- EUROCAT (congenital malformations)
- ENERCA (congenital anemias)
- SCN (severe congenital neutropenias)
- EUROMUSCLENET (myopathies)
- CAUSE (CHARGE et Usher)
- EINPRDP (rheumatic paediatric diseases)
- IDR (immunodeficiencies)
- TEAM (adult metabolic diseases)
- European Autism Information System (Autistic disorders)
- RARECARE (rare cancers)
- TREAT-NMD (neuromuscular diseases)
- EUROGLYCANET (glycosilation disorders)
- GENESKIN (skin genetic diseases)
- SKINTHERAPY (epidermolysis bullosa)
- CONTICANET (connective tissue cancers)
- HISTIONET
- …….

Conclusions

- 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 diseases where they should be
    - Everywhere as a lower node
- Chapter on Haematology is already available
  - Draft proposal by Orphanet to be sent to TAG members for dissemination to experts
  - Please look at our proposals on the WHO website
- Production of one chapter per month