International Classification of Diseases in the field of rare diseases Revision process

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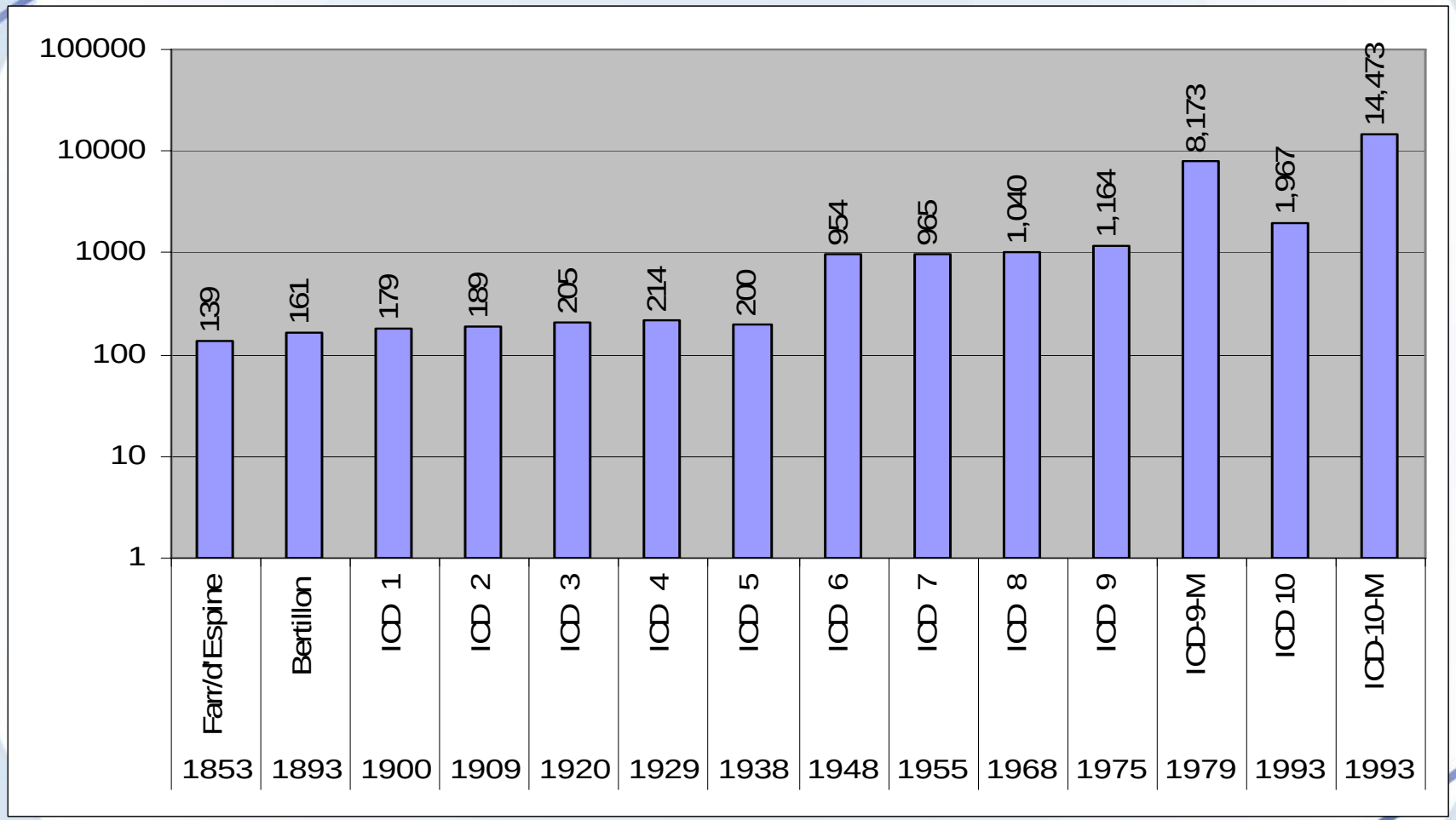
At WHO

ICD Revisions



With the courtesy of WHO

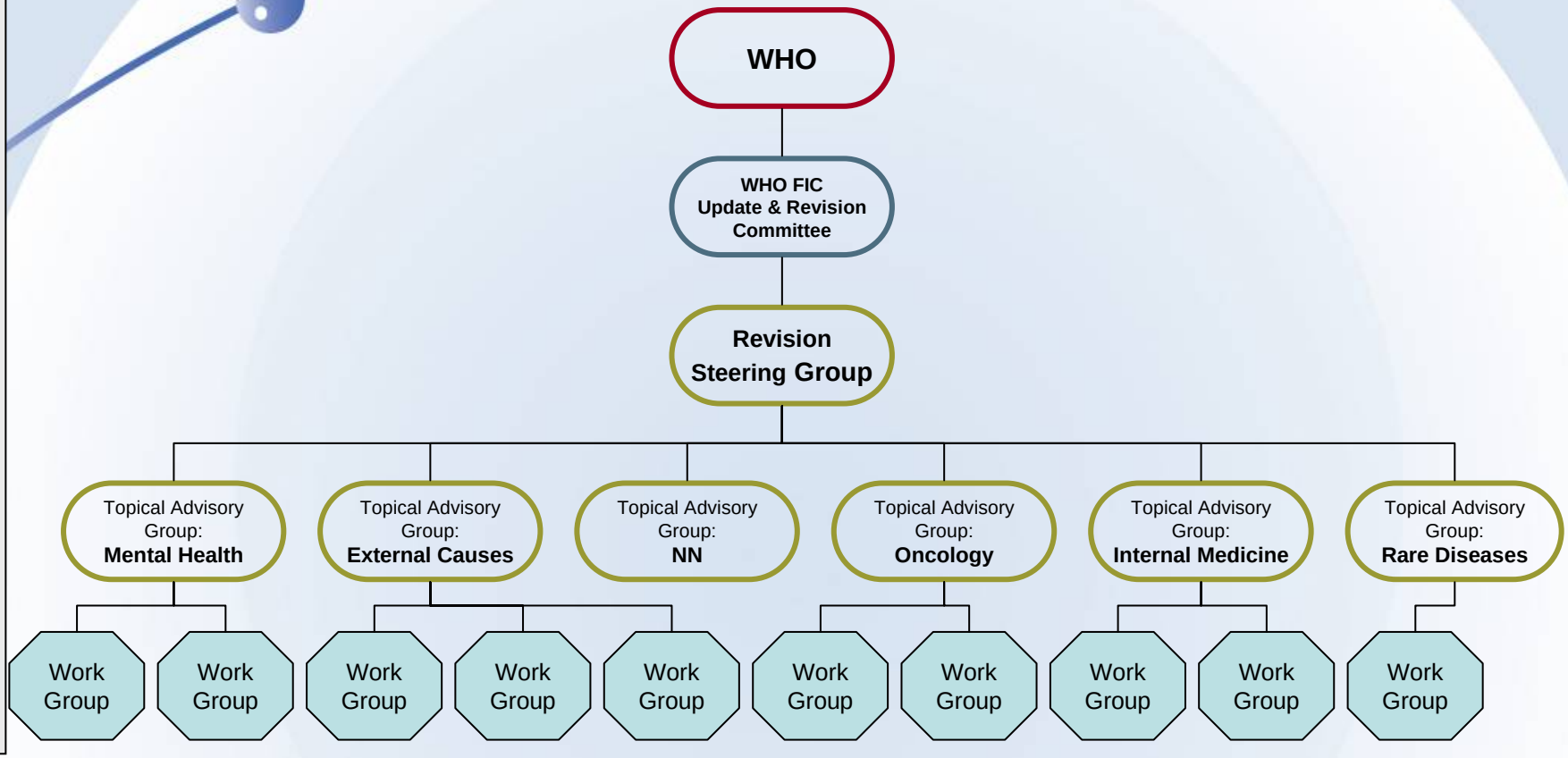
Disease: a concept linked to level of knowledge

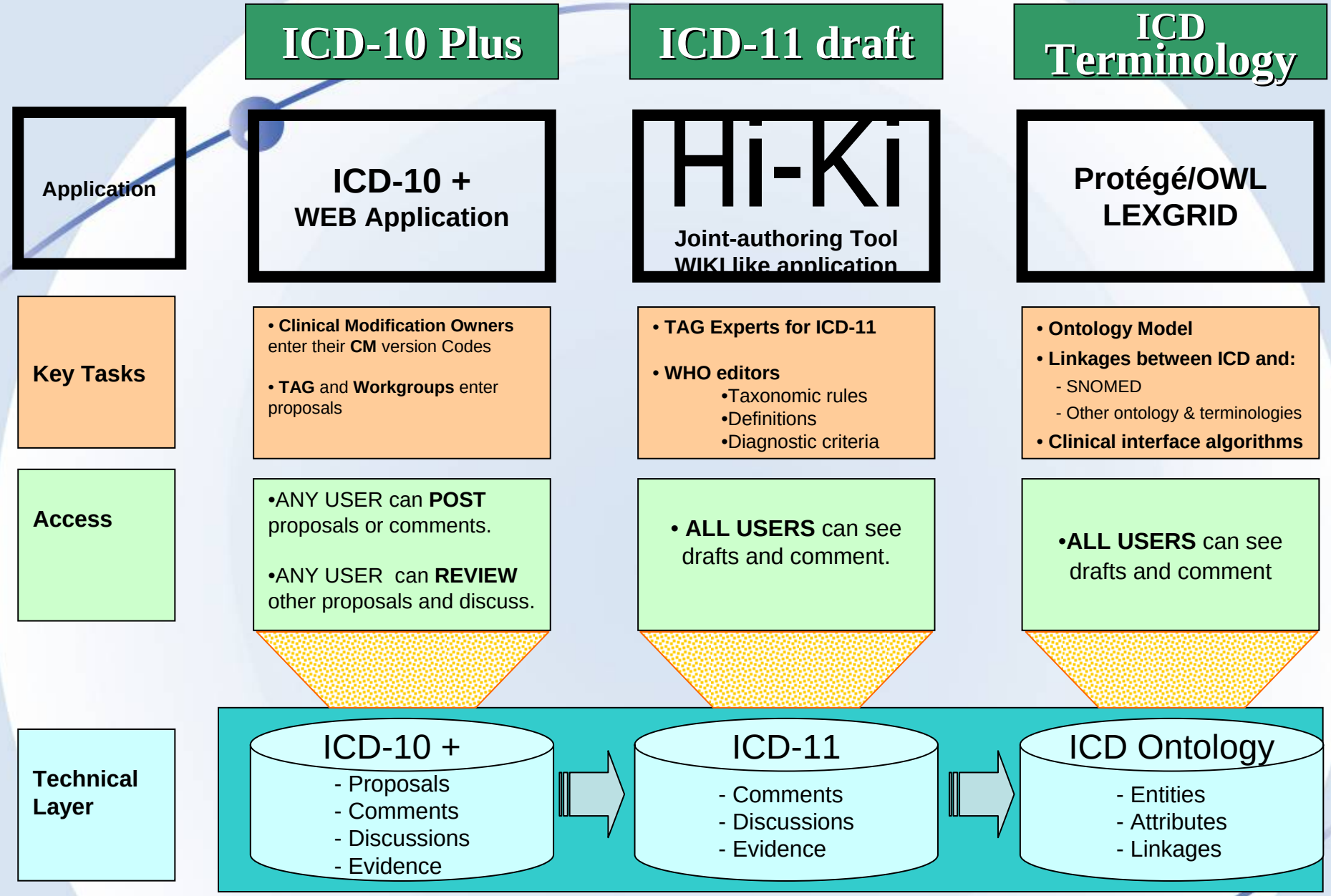
- Recognisable pattern of signs and symptoms
  - Clinical approach
- with a unique mechanism
  - Physiopathological approach
- with unique course and prognosis
  - Clinical evolution approach
- with a unique response to intervention and treatment
- with an established cause
  - Genetic origin

ICD-10 Revision organization structure

FIELD TRIALS

KMS PORTAL




ICD-10 Plus
ICD-11 draft
ICD Terminology
Application
ICD-10 +
WEB Application
Hi-Ki
Joint-authoring Tool
WIKI like application
Protégé/OWL
LEXGRID
Key Tasks
• Clinical Modification Owners enter their CM version Codes
• TAG and Workgroups enter proposals
• TAG Experts for ICD-11
• WHO editors
•Taxonomic rules
•Definitions
•Diagnostic criteria
• Ontology Model
• Linkages between ICD and:
- SNOMED
- Other ontology & terminologies
• Clinical interface algorithms
Access
•ANY USER can POST proposals or comments.
•ANY USER can REVIEW other proposals and discuss.
• ALL USERS can see drafts and comment.
•ALL USERS can see drafts and comment
Technical Layer
ICD-10 +
- Proposals
- Comments
- Discussions
- Evidence
ICD-11
- Comments
- Discussions
- Evidence
ICD Ontology
- Entities
- Attributes
- Linkages

Hierarchy of Hiki Authority
by ICD Domain

0 Revision Steering Committee

1 Revision Domain/Topic Working Groups

2 Accredited Experts

- Designated by Working Group Members

3 Accredited Persons

- Designated by Experts

4 Registered Interested Persons (Public)

Tentative timeline

- 2010 : **Alpha version** (ICD 10+ → ICD 11draft)
  - **+1 YR : Commentaries and consultations**
- 2011 : **Beta version** & Field Trials Version
  - **20+2 YR : Field trials**
- 2013 : **Final version** for public viewing
  - **2014 : WHA Approval**
- 2015+ implementation

RDTF WG Coding and Classification

- RDTF Workshops:
  - 11 October 2006
  - 2 May 2007
  - 13 November 2007
  - 6 February 2008
- Participation to WHO revision committee:
  - 15-18 April 2007 in Tokyo
  - 29-31 October 2007 in Trieste
  - 9-11 April 2008 in Geneva

Principles guiding action

- Rare Diseases should be traceable in mortality and morbidity information systems
- There are two categories of RD
  - The recurrent RD (about 1,500)
    - should have a specific code in ICD11
  - The ultra-rare (over 4,000)
    - should be coded as “other specific RD”
    - within relevant subcategory

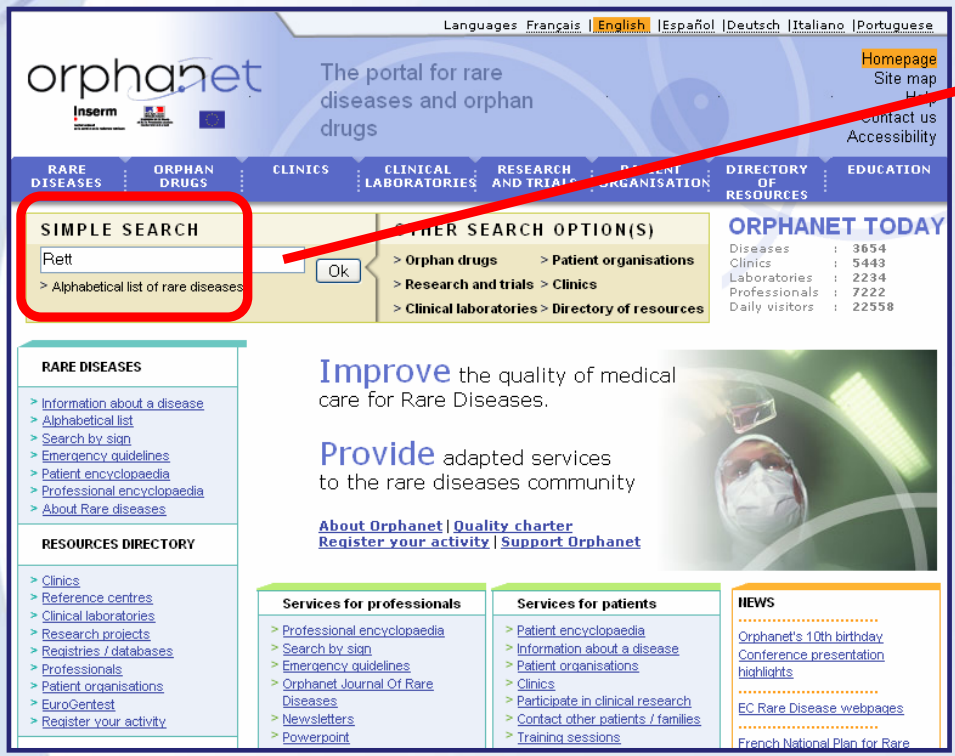
Phase I
Collection of data necessary for classifying diseases

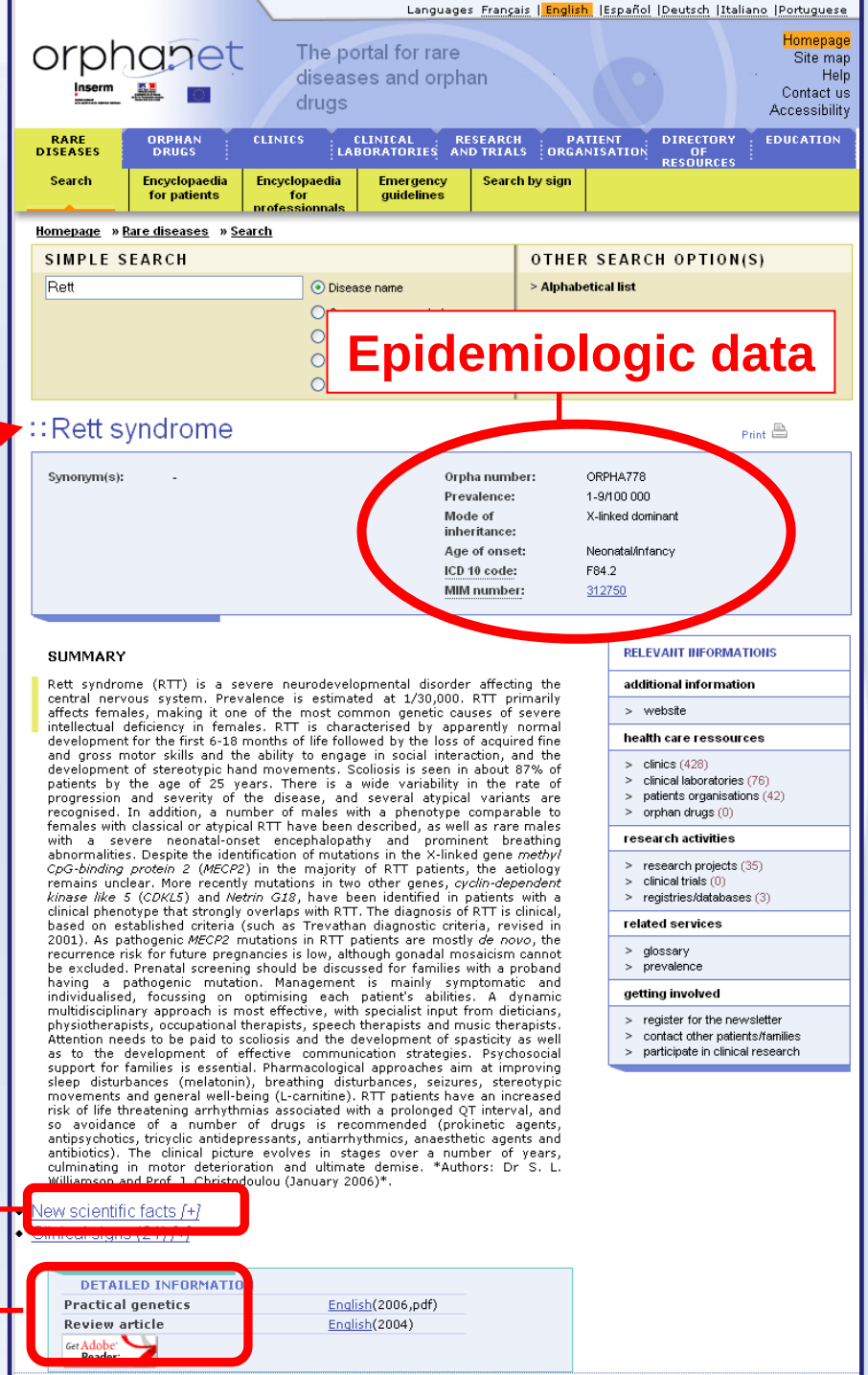
Use of the Orphanet database of diseases to propose ontologies

Orphanet platform as a tool

- Relational database of 5,800 rare diseases
  - Encyclopaedia
  - Genes + proteins + ICD10 + MIM + MeSH
  - Epidemiology, mode of inheritance, age at onset + textual information
  - ontologies
- Shared tools between partners
  - Access to files
  - Protected website with all data

Simple search: by disease name

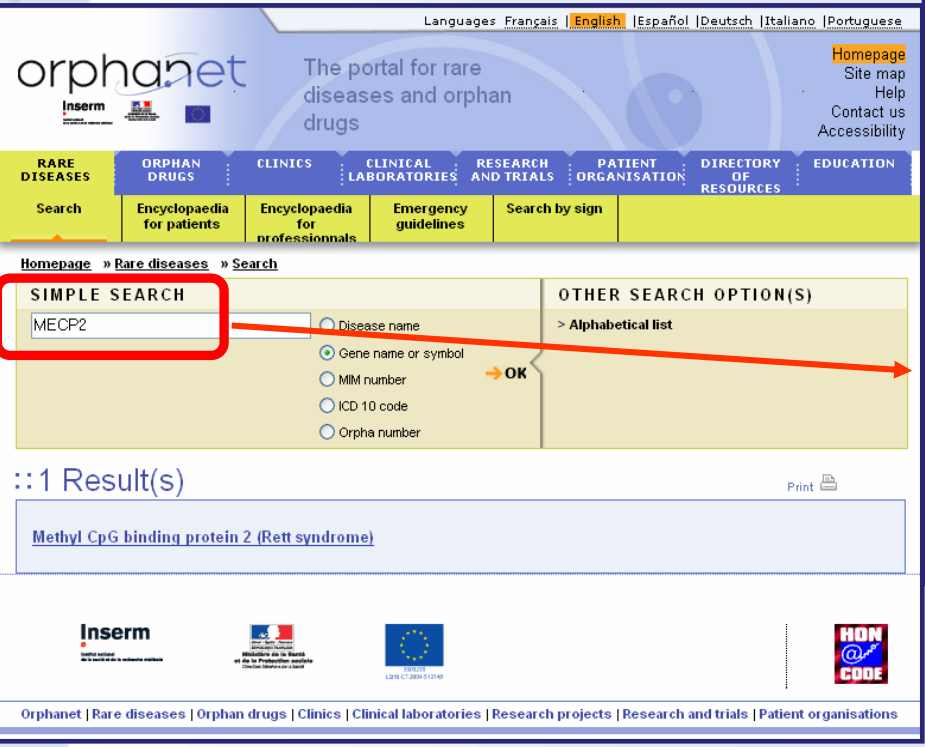


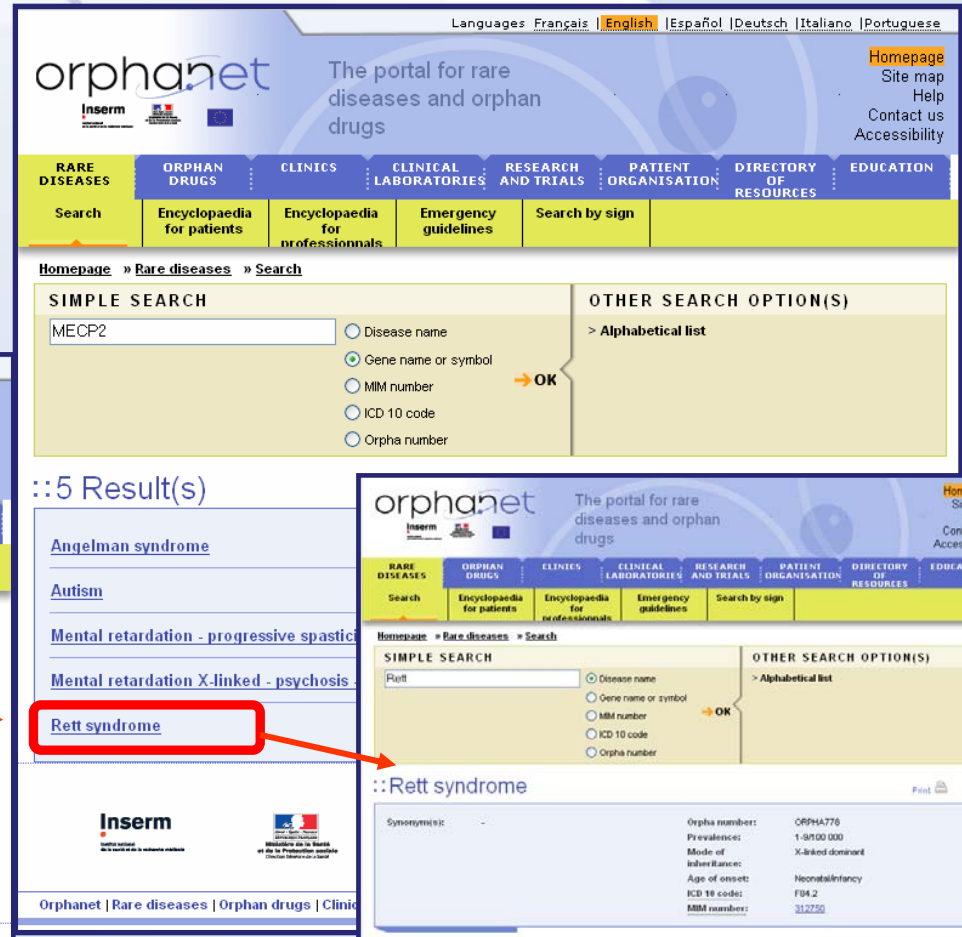


Scientific news

'Review articles/Practical genetics'

Simple search: by gene name





Phase II: analysis of data to define classification approach

- Comparison of ICD10 codes
  - Between databases of rare diseases
  - To identify true mismatches
  - To have a typology of possible approaches
- Collection of classifications
  - already published
  - By medical specialty: establised by Orphanet

Phase III: Document true mistakes in ICD10

- Due to
  - the evolution of knowledge
  - the structure of the current classification
- Propose an evolution to WHO
  - ICD10+
  - Wikipedia-like tool

Ex: possible different interpretations

- CADASIL (OMIM 125310)
  - UKGTN I77.8
    - Other specified disorders of arteries and arterioles
  - Orphanet F01.1
    - Multi-infarct dementia (In: vascular dementia)

Ex: incorrect specific code

- Marfan syndrome
  - Q87.4
    - In: Q87 Other specified congenital malformation syndromes affecting multiple systems

Could have been included in

- M30-M36 Systemic connective tissue disorders

Ex: inappropriate categories

- VATER association
  - Q87.2 Congenital malformation syndromes predominantly involving limbs
    - Holt-Oram
    - Klippel-Trenauney-Weber
    - …
    - VATER

VATER = Vertebral defects, Anal atresia, TracheoEsophageal fistula with esophageal atresia and Radial dysplasia

Problematic groups of diseases

- Systemic diseases (Internal medicine)
  - Marfan disease in Q87.4 (malformations)
  - Amyloidosis in E85 (metabolic disorders)
  - Mastocytosis in:
    - Q82 (Other congenital malformations of skin)
    - C96.2 (Malignant mast cell tumour)
  - Mediterranean fever
    - E85.0 (Non-neuropathic heredofamilial amyloidosis) in Metabolic disorders

Ex: Problematic groups of diseases

- Skin diseases
  - Some skin diseases are coded as malformations
    - Q80 **Congenital ichthyosis**
    - Q81 **Epidermolysis bullosa**
    - Q82.1 **Xeroderma pigmentosum**
    - *etc.*

Problematic groups of diseases

- Mental retardation
  - ICD-10 codes for MR are based on QI
    - Coding can only be done on an individual basis
    - Coding for syndromes with MR it is, therefore, not possible

Phase IV: Propose a new classification

- Proposal from the Topic Advisory Group
  - Criteria: Scientific evidence + clinical utility + health system utility + public health usefulness
  - Draft by Orphanet team: core concepts and structures
  - First review by TAG members
  - Second Review open to all for comments

Thank you for your attention

Register with us if you wish to be involved