From ICD10 to ICD11
Proposal for a general approach and incorporation of rare diseases

Ségolène Aymé,

COMP meeting -1 April 2009

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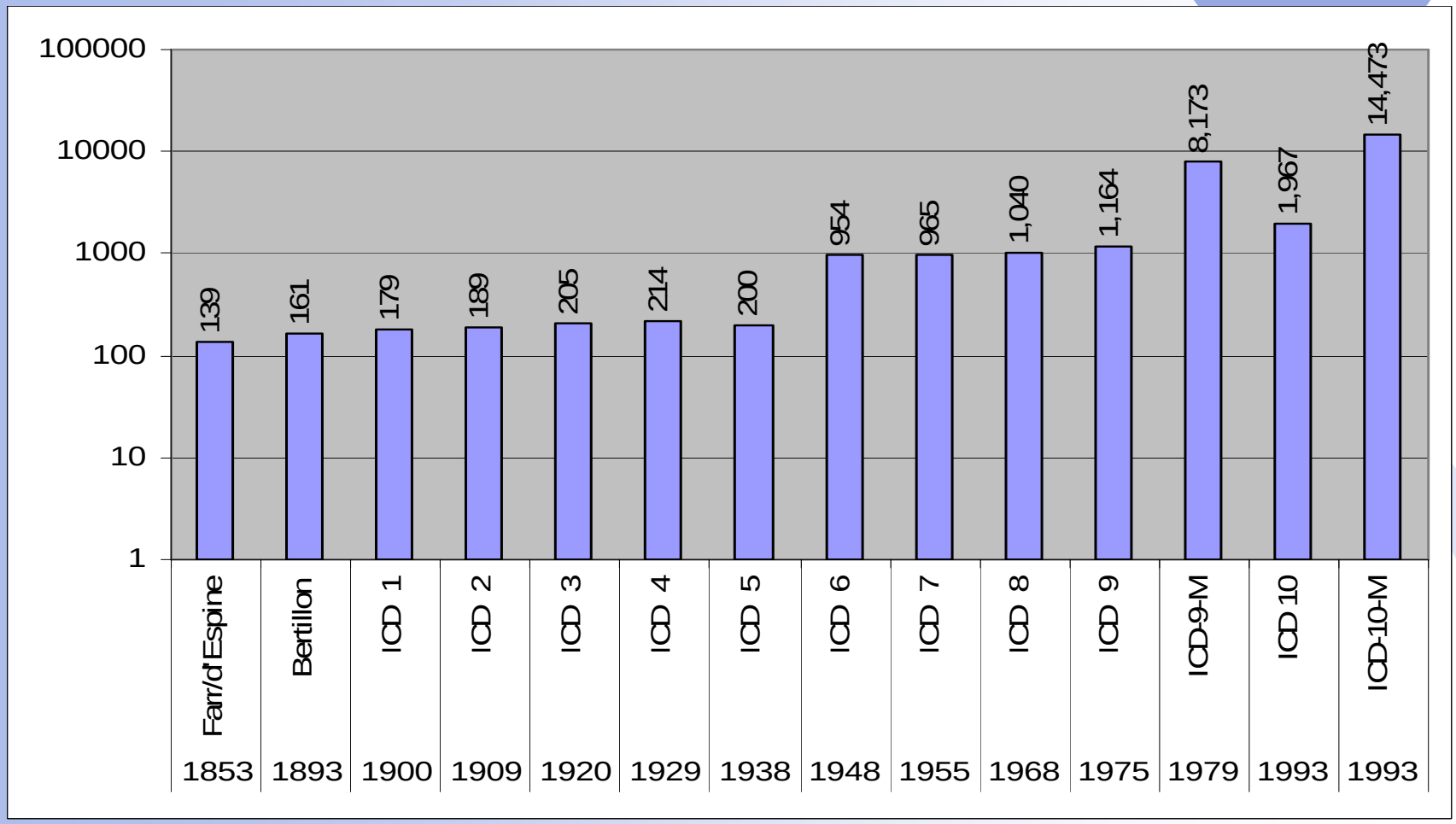
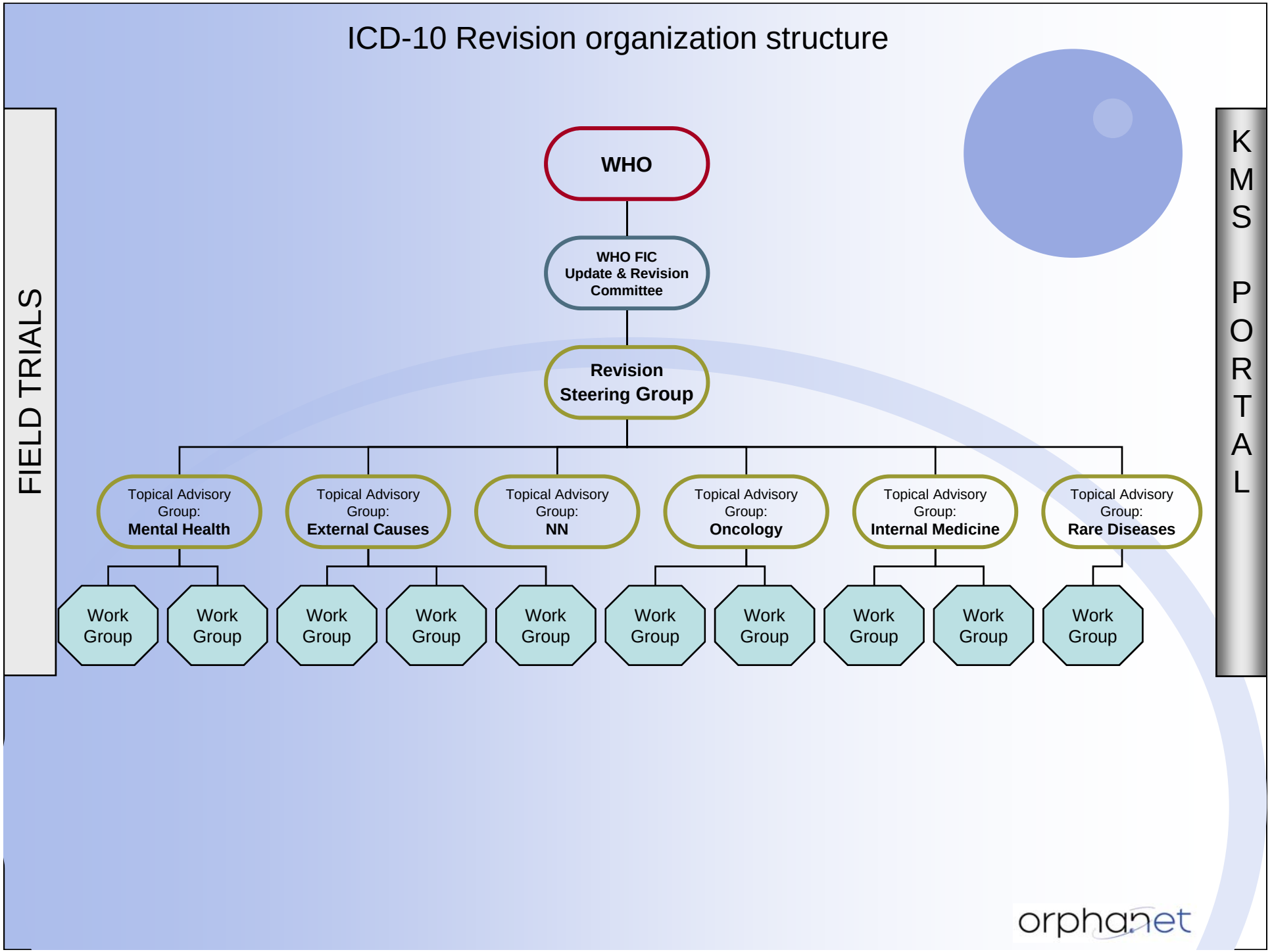
ICD Revisions

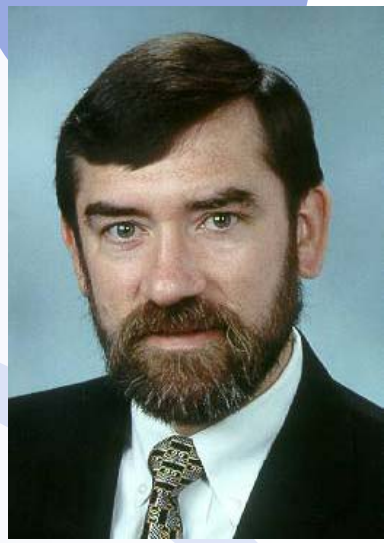


ICD-10 Revision organization structure



The Revision Steering Group

Chair of the RSG



Christopher G Chute

Mayo Clinic

Rochester, USA

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The Revision Steering Group

Rare Diseases

Chair: Ségolène Aymé

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Rare Diseases Platform

Broussais Hospital, Paris, France

Role of TAG

- Advise WHO in all steps leading to the revision
- Establish workgroups and partners to involve
  - Generation of necessary evidence
  - Develop proposals for changes
  - Establish procedures
  - Facilitate cross fertilization of ideas and reducing redundant efforts
- Advise in developing various drafts of topic segments in line with the overall production timeline of ICD11
- Advise in developing protocols for and in implementing field trials

Composition of TAG Rare Diseases

- Europe:
  - Ségolène Aymé, Ana Rath (Orphanet)
- USA:
  - Stephen Groft, Roberta Pagon (Office of RD-NIH)
- Korea:
  - Hyun-Young Park (CDC-Information Center, Seoul)
- Brazil:
  - Eduardo Castilla
- Argentina:
  - Monica Rittler
- Mexico:
  - Osvaldo Mutchinick
- Russia:
  - Evgeny Ginter
- China:
  - Yiming Wang

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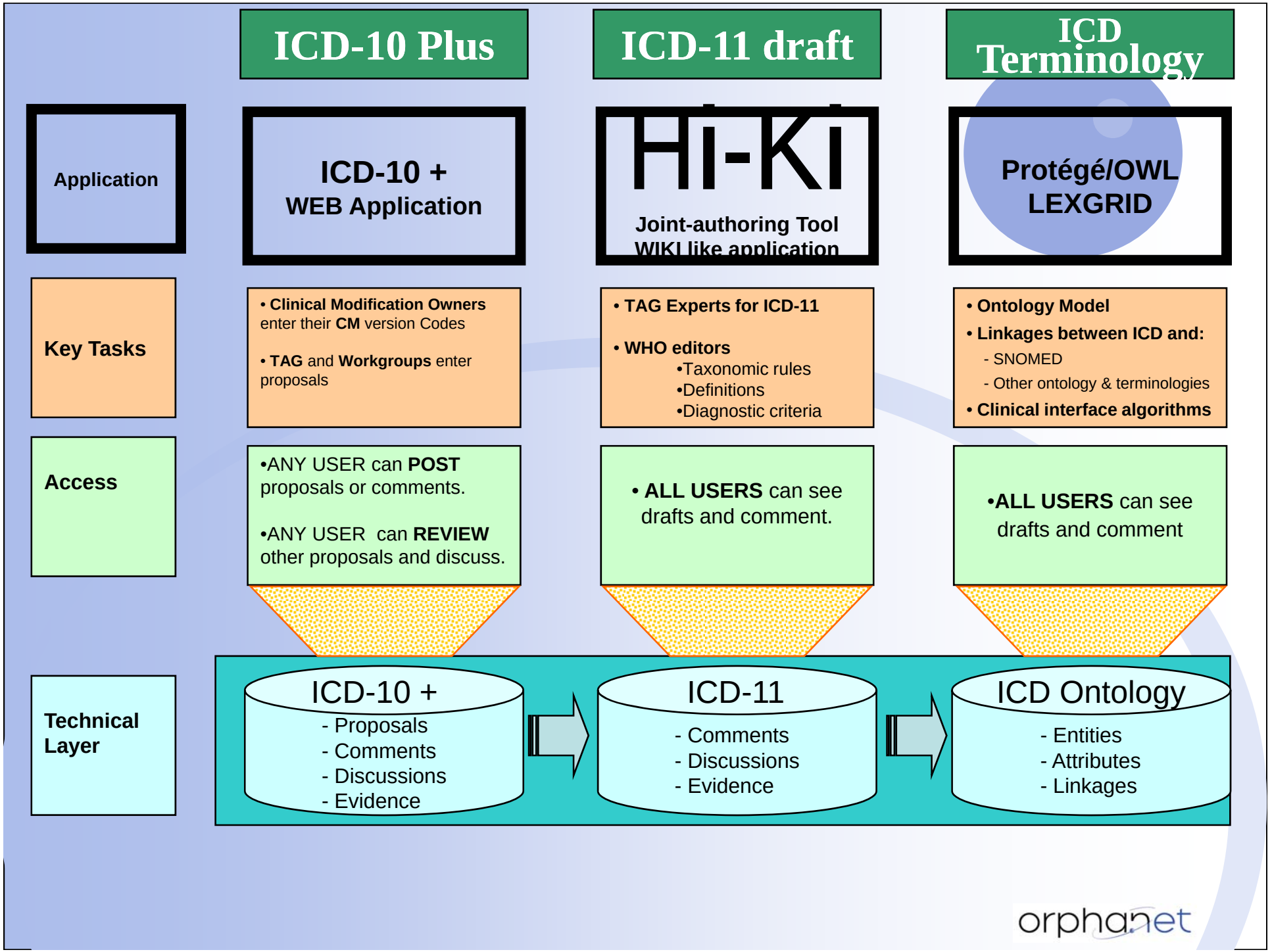
Experts for peer-review

- Experts identified by Orphanet
  - Editorial board members
  - Orphanet scientific advisory board at country level
  - European networks / Learned societies
- Same process in each region
  - Decided by each TAG member


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
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Change Proposal Generation

- Implicit creation of change proposal by changing local view
- Can be extracted into machinable format
- Editors and reviewers can see changes in context
  - What would it look like if adopted
- User can write explicit proposal

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)

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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

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Preparatory work at Orphanet

Step 1: collect published classifications and build a clinical one

Since 2006

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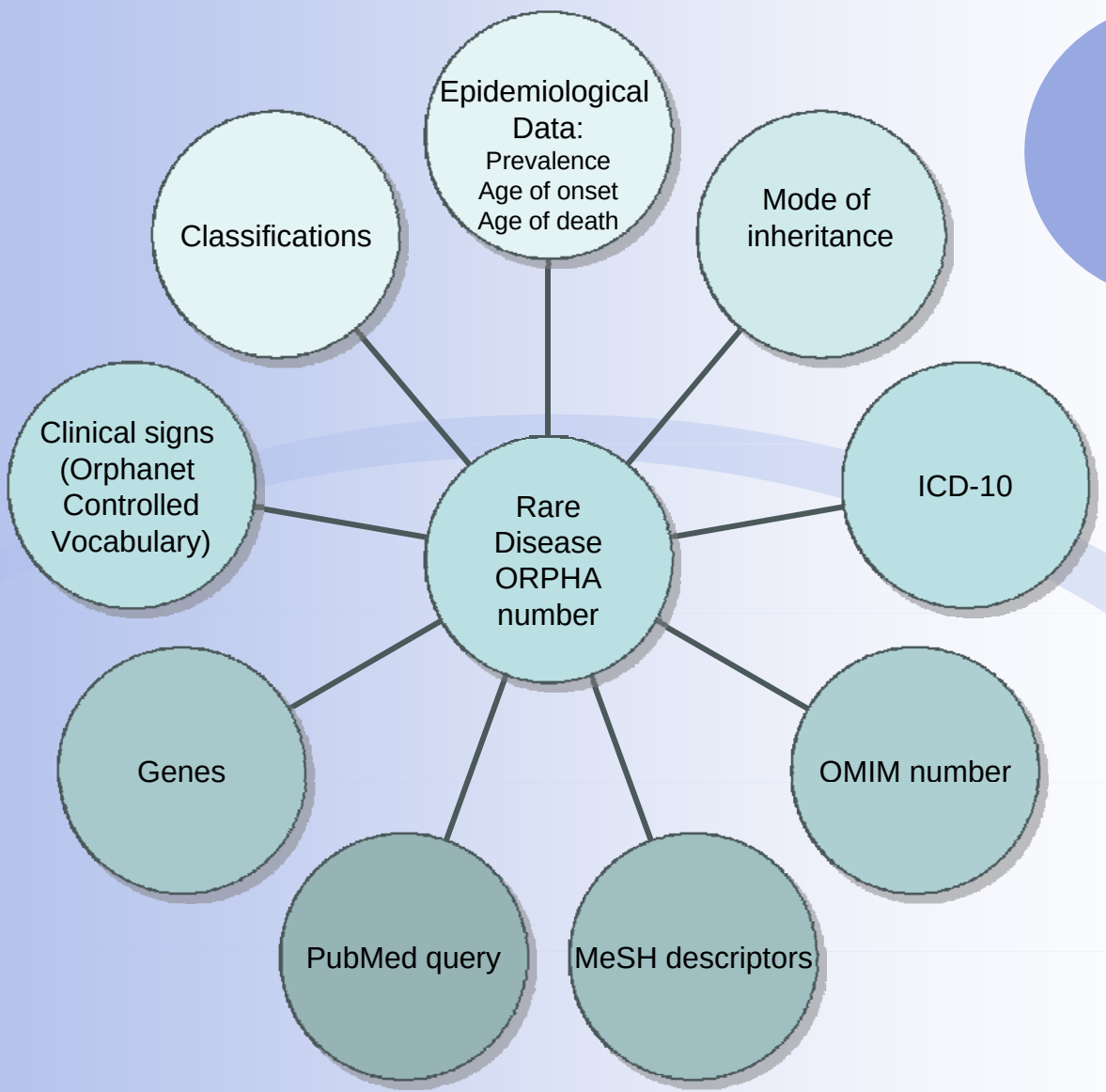
Classification of rare diseases

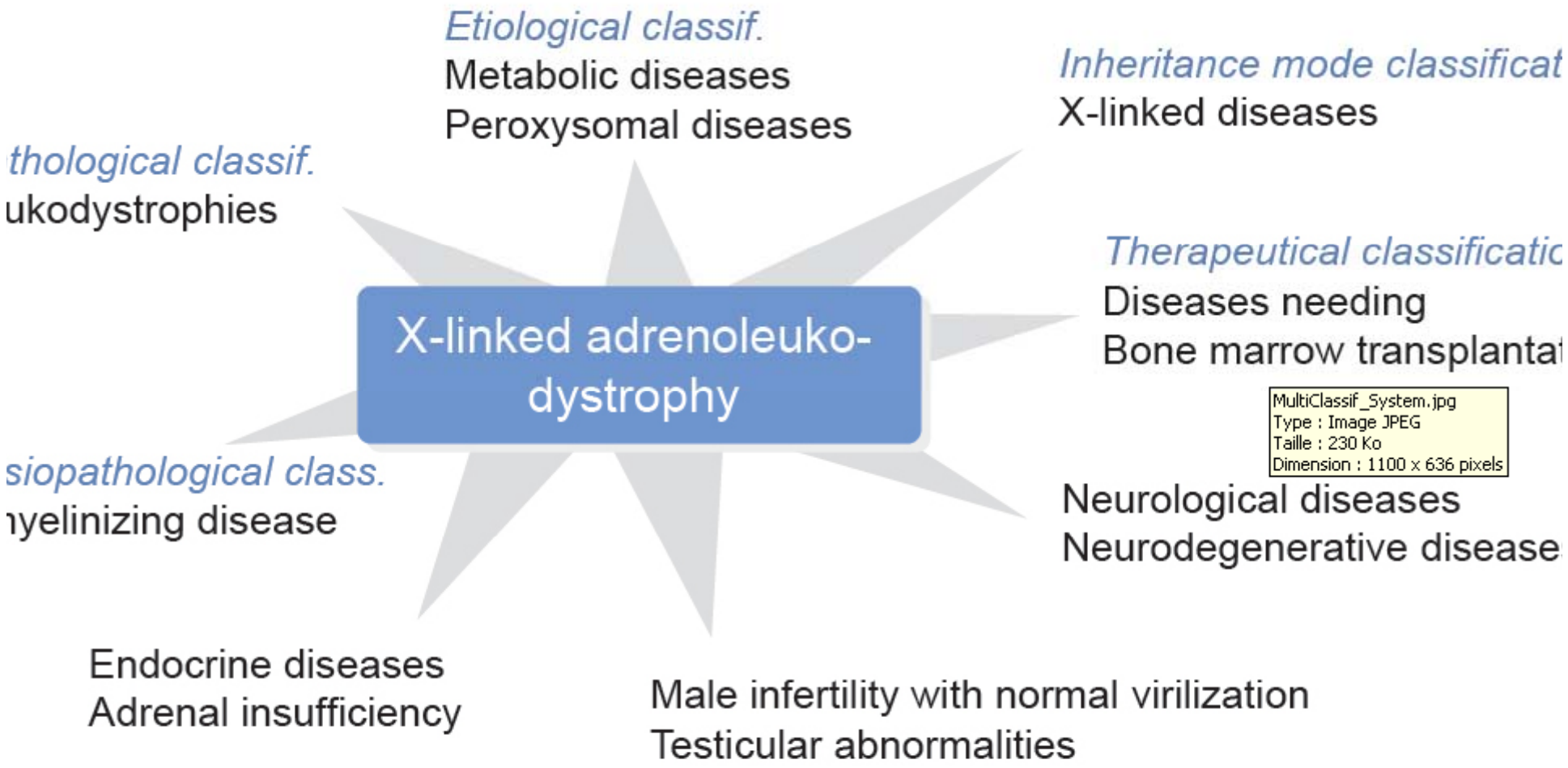
- Methodology:
  - Collection of published classifications of specific groups of rare diseases
    - Clinical classifications
    - By mechanism or by aetiology
  - Building up of a comprehensive classification of Orphanet entries to serve the needs of Health Care information systems: the Orphanet classification of rare diseases
    - Based on
      - Literature (reference texts, scientific publications)
      - Experts working groups
    - Validated by the scientific advisory board of Orphanet
    - Regularly updated

Rationale for a clinical classification

- Criterion: medical/surgical specialties involved in the management of the different manifestations of a disease
- Stick to medical specialties organised by system, then by group of age (pediatrics/adults), then by subspecialty (i.e. epileptology)
- Etiological/physiopathological criteria were futher applied, when relevant for diagnosis/management


Epidemiological Data:
Prevalence
Age of onset
Age of death
Classifications
Mode of inheritance
Clinical signs (Orphanet Controlled Vocabulary)
Rare Disease ORPHA number
ICD-10
Genes
OMIM number
PubMed query
MeSH descriptors

MULTI-CLASSIFICATION SYSTEM



Orpha nomenclature

- Comprehensive list of rare diseases (<5,850)
  - Identity card + genes
  - Unique Orpha number
    - Stable what ever is the evolution of knowledge
    - Linked to parent and child disease in every classification
  - Files available on request
    - for use in information systems
    - For research purpose
    - At running cost for non-profit use + Material Transfer Agreement

- Classifications of rare diseases (by September 08)
  - List of all published classifications
  - Visualisation of each classification
  - Possibility to click at any level to have the detailed information

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SEARCH A DISEASE

Disease name gaucher → OK

OTHER SEARCH OPTION(S)

> Search a group of diseases

- Classification of rare forms of dementia
- Orphanet classification of rare neurological diseases
- Orphanet classification of rare eye diseases
- Classification of sucking/swallowing disorders
- Orphanet classification of inborn errors of metabolism
- Orphanet classification of genetic diseases

SEARCH A DISEASE

Disease name gaucher → OK

OTHER SEARCH OPTION(S)

> Search a group of diseases

return to list of classifications

Orphanet classification of inborn errors of metabolism

Source [↗]

- Metabolic disease, rare
  - Metabolic disease involving complex molecules
    - Lysosomal disease
      - Sphingolipidosis
        - Gaucher disease
          - **Gaucher disease type 1**

return to list of classifications

Additional information

Orphanet Reports series

> Prevalence of rare diseases
> Orphan drugs with MA

Getting involved /informed

> Read the newsletter
> Read OJRD [↗]
> Contact other patients/families
> OrphanXchange [↗]
> Register your activity

: : Orphanet classification of rare neurological diseases

Source [↗]

- Neurological disease, adult, rare
  - Ataxia, adult [+]
  - Central nervous system and retinal vascular disease, rare [+]
  - Complex regional pain syndrome [+]
  - Dementia, rare [+]
  - Encephalitis [+]
  - Epilepsy, adult, rare [+]
  - Headache, rare [+]
  - Idiopathic orthostatic hypotension [+]
  - Komar syndrome [+]
  - Leukodystrophy, adult [+]
  - Locked-in syndrome [+]
  - Medullar disease, adult, rare [+]
  - Movement disease, adult, rare [+]
  - Nervous system tumour, adult [+]
  - Neurodegenerative disease, adult, rare [+]
  - Neuroimmunological disease, adult [+]
  - Neuroleptic malignant syndrome [+]
  - Neurometabolic disease [+]

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: : Orphanet classification of rare neurological diseases

Source [↗]

- Neurological disease, adult, rare [-]
  - Leukodystrophy, adult
    - Adrenomyeloneuropathy [+]
    - CACH syndrome, juvenile or adult form [+]
    - Leukodystrophy, unknown [+]
    - Leukoencephalopathy - dystonia - motor neuropathy [+]
    - Metachromatic leukodystrophy [+]

return to list of classifications

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Preparatory work at Orphanet

Step 2: Analyse ICD10 and identify problems

Since 2007

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Identification of coding difficulties

- Preliminary study on ICD-10 coding for rare diseases
  - Identification of specific codes: 240 ONLY
  - Identification of rare diseases listed under a category
  - Cross-matching with other datasets (CINEAS, UKGTN, Veneto register)
  - Typology of problems for coding rare diseases with ICD-10

Matching incorrect specific codes

- 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

Problematic groups of diseases

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

Coding according to the cause or the consequence?

- Alkaptonuria E70.2
- Tyrosinemia type 1 E70.2
  - E 70.2: Disorders of tyrosine metabolism
    - Alkaptonuria
    - Tyrosinemia
    - …
  - Both diseases are coded to the cause but phenotypes are quite different
    - Alkaptonuria: pigmentation disorders, muscular-skeletal symptoms, arthropathy… or asymptomatic
    - Tyrosinemia: severe hepatocellular necrosis (early onset form), renal tubulopathy (late onset form)

How to code hereditary forms of diseases?

- Melanoma, familial
  - C43 Melanoma, malignant
  - A Z80 code should be added to familial forms of cancers?
- What about familial forms of not rare diseases?
  - Parkinson disease, familial form
    - G20 Parkinson disease

First conclusions

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

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

Preparatory work at Orphanet

Step 3: Contribute to ICD10+
and ICD11
Since January 2009

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Process at Orphanet

- 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)

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

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 NIH
- A final proposal will be sent to WHO, chapter by chapter, one every month

Proposal for general principles

Based on published classifications
and on past experience in using
codes at Orphanet

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Rare diseases and biological systems

- Biological systems are networks
  - Hierarchy of networks
  - Higher nodes are limited in numbers :Minority of nodes account for a large amount of connections:
    - Metabolic, signalling, immunity, repair, stress….
  - Common Diseases: extreme values at one upper node
  - Rare Diseases: extreme values at lower nodes

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 »)

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 and under review by experts
- Production of one chapter per month
  - Endocrinology (April), Immunology (May)….