



Codification and Classification of Rare Diseases

Activities of the RDTF
working group

RDTF objectives



- To wide access to high quality information
- To assist in the diffusion of good and best practice
- To promote the exchange of ideas and information regarding quality of life issues, and patients preferences and choices
- To promote the availability of high quality epidemiological data
- **To promote the development of a classification and of a coding system to supplement the ICD**
- To promote effective surveillance, early warning...
- To promote the creation of reference centres
- To facilitate the consideration of different models of cross-border health care

WG Coding and Classification



- Issues to be tackled
 - State of art of existing coding systems regarding rare diseases: ICD, Snomed, MeSH, MedDRA
 - Plans for contributing to improve these systems, especially to contribute to the revision of ICD10 in collaboration with WHO
 - Establishment of a database of expert classifications of rare diseases

WG Coding and Classification



- **Workshops:**
 - **First meeting on 11 October 2006**
 - **Second meeting on 2 May 2007**
- **Participation to WHO revision committee:**
 - **15-18 April 2007 in Tokyo**
- **Background activity:**
 - **Part of Orphanet mission**

Orphanet platform as a tool



- Dedicated team of 30 professionals
- Relational database of 5,200 rare diseases
 - Encyclopaedia
 - Genes + proteins + ICD10 + MIM + MeSH
 - Epidemiology, mode of inheritance, age at onset + textual information
- Shared tools between partners
 - Access to files
 - Protected website with all data

Principles guiding action



- Rare Diseases should be traceable in mortality and morbidity information systems
- There are two categories of RD
 - The recurrent RD (?1,500 to 2,000)
 - should have a specific code in ICD11
 - The ultra rare (around 4,000)
 - should be coded as «other specific RD » within relevant subcategory but indexed

Proposal for action



- Step 1: Establish the priority list which deserves a specific code in ICD11
- Step 2: Analyse ICD10 to identify mistakes and gaps
- Step 3: Start contribute to ICD10+
- Step 4: Collect other classification systems

1- Establish the priority list



- Agree on the criteria
 - Any disease coded in a registry of patients or an information system
 - Any disease covered by a support group
 - Any disease with a clinical test
- Establish the list
 - Tests or patients groups or registries....
 - 2 500 in Orphanet
- Validate the list
 - Public consultation
 - Expert review process

2- Analyse ICD10



- Collect all lists of RD with ICD10 code
 - Orphanet, Cineas, UKGTN, Italian registry so far
 - Others to be approached
- Cross match these lists
 - Identify differences: external quality control
- List mistakes, problems and gaps
 - Reach an agreement between experts
 - Document rational for a change

3- Contribute to ICD10+



- Specific ICD10 code exists already
 - Classification is correct: end of action
 - Classification is incorrect: proposal reclassification
- Non specific ICD10 code
 - If in priority list: propose a specific code
 - If not: propose an « other specific RD »
- No ICD10 code
 - If in priority list: propose a specific code
 - If not: propose an « other specific RD »

Composition of TAG

Thus far.....

- Europe:
 - Ségolène Aymé, Ana Rath (Orphanet)
 - Representative of Cineas (Genetics-NL)
 - Representatives of NHS-UK
 - Representatives of Italian registries
 - Representative of Eurocat
- USA:
 - Stephen Groft, Roberta Pagon (Office of RD-NIH)
- Australia:
 - Agnes Bankier (Possum, Murdoch Institute)
- Korea:
 - GH Lee (CDC-Information Center, Seoul)

Indexation of RD in Orphanet

An On-going Process

- ICD-10
 - 324 diseases have a specific code
 - 1,586 have a generic code
- MeSH
 - MeSH terms attributed to 1,149 diseases
- PubMed automatic search tool
 - Available so far for 1,407 diseases

Outcome typologies



- ICD-10 codes do not match
 - Mistake in one of data sets
 - Different interpretations are possible: needs further examination
- ICD-10 codes match
 - RD is correctly coded in ICD-10 (specific)
 - ICD-10 code is not specific: needs for further examination
 - RD is coded in a wrong ICD-10 category: needs further examination

Mismatch due to mistakes

- Multiple endocrine neoplasia
(OMIM 1431100)

- UKGTN D44.8

- Pluriglandular involvement .../...Multiple endocrine adenomatosis

- Orphanet C25.4

- Malignant neoplasm of endocrine pancreas

C75.0

- Malignant neoplasm of ...parathyroid gland

C75.1

- Malignant neoplasm of ... pituitary gland

Mismatch due to mistakes

- Hyperparathyroidism, neonatal severe primary (OMIM 239200)
 - UKGTN E83.5
 - Disorders of calcium metabolism (excludes hyperparathyroidism)
 - Orphanet E21.0
 - Primary hyperparathyroidism

Mismatch due to different interpretations

- Barth syndrome (OMIM 302060)
 - UKGTN E88.8
 - Other specified metabolic disorders
 - Orphanet 142.0
 - Dilated cardiomyopathy

Mismatch due to different interpretations

- Cystinosis, nephropatic (OMIM 219800)
 - UKGTN E72.0
 - Disorders of amino-acid transport... Cystinosis
 - N16.3**
 - Renal tubulo-interstitial disorders in metabolic diseases... *Renal tubulo-interstitial disorders in cystinosis*
 - Orphanet E72.0
 - Disorders of amino-acid transport... Cystinosis

Mismatch due to different interpretations

- CADASIL (OMIM 125310)
 - UKGTN 177.8
 - Other specified disorders of arteries and arterioles
 - Orphanet F01.1
 - Multi-infarct dementia (In: vascular dementia)

Mismatch due to different interpretations

- Norrie disease (OMIM 310600)
 - UKGTN H44.8
 - Other disorders of globe
 - Orphanet Q15.8
 - Other specified congenital malformations of eye

Codes match but...

- They are nonspecific
 - Pulmonary lymphangiectasia, congenital
 - CINEAS Q34.8
 - Orphanet Q34.8
 - Other specified congenital malformations of respiratory system
 - There is no code for congenital lung malformations of vascular origin

Codes match but...

- They are specific, but wrong
 - Ehlers-Danlos syndrome type 1
 - CINEAS Q79.6
 - Orphanet Q79.6
 - Ehlers-Danlos syndrome (In: Congenital malformations of the musculoskeletal system, not elsewhere classified)
 - Should be better classified in M00-M99 (Diseases of the musculoskeletal system and

In conclusion



- Cross-mapping data sets allows
 - To identify mistakes and improve coding
 - To identify ICD-10 problems, i.e.
 - Need for categories rearrangement
 - Need for more specific categories, better reflecting homogeneous groups of rare diseases

Next steps



- Matching of lists of codes is on-going
- Next workshop: 13 November in Luxembourg
- Release of the new version of Orphanet with the classifications Nov 2007