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 identity 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 exits 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 I42.0
    - 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 disoders in cystinosis*
  - Orphanet E72.0
    - Disorders of amino-acid transport… Cystinosis

Mismatch due to different interpretations

- CADASIL (OMIM 125310)
  - UKGTN I77.8
    - 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