Classification of rare diseases: a worldwide effort to contribute to the International Classification of Diseases

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ORPHANET JOURNAL OF RARE DISEASES

ORAL PRESENTATION Open Access

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From 5th European Conference on Rare Diseases (ECRD 2010)
Krakow, Poland. 13-15 May 2010

Most rare diseases are absent in the International Classification of Diseases (ICD10) and those with a specific code are often misclassified. As a consequence, morbidity and mortality due to rare diseases is invisible in health information systems. To overcome this difficulty, Orphanet (http://www.orpha.net) has established a partnership with WHO to ensure a fair representation of rare diseases in general. Orphanet has collected all published expert classifications and established a database of phenotypes indexed with ICD10 codes, MIM codes, genes, mode of inheritance, age of onset and class of prevalence. Phenotypes are assigned to as many classification systems as necessary to represent them. The Orphanet nomenclature of rare diseases is a stable one, directly exploitable by information systems and available on request. A WHO Topic Advisory Group on rare diseases has been established to manage the revision process. The first revised chapters currently circulating among experts and expert groups for review are Haematology, Endocrinology, Nutrition, Metabolism, Immunology, Neurology and Malformations. Revised chapters follow a primarily clinical approach, only secondarily an aetiological one up to the gene level. When several possible names are available for a disease, descriptive names formed in accordance with a clinical approach are preferred. Every entity is assigned a unique identification number. Rare diseases affecting several body systems are included in every relevant chapter, as ICD11 will be poly-axial, but a main code is proposed to allow for linearisation, according to the most severe involvement and/or the specialist most likely to be relied on for the management of the disease. The rare disease community is invited to take an active part as the results will condition the visibility of all activities in the field. All the revised chapters open for comments are available on http://www.eucerd.eu

Published: 19 October 2010

doi:10.1186/1750-1172-5-S1-O21
Cite this article as: Rath *et al.*: Classification of rare diseases: a worldwide effort to contribute to the International Classification of Diseases. *Orphanet Journal of Rare Diseases* 2010 **5**(Suppl 1):O21.

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