

**Recommendation
on Ways to Improve
Codification for Rare Diseases
in Health Information Systems**

**Adopted at the 3rd meeting of the
Commission Expert Group on Rare Diseases
12-13 November 2014**

Statement about the rare disease coding issue

- Improved codification for rare diseases is cited as a priority in the *Council Recommendation on an action in the field of rare diseases* (2009).
- Currently, only a small fraction of rare diseases have codes in international nomenclatures, making it a challenge to trace patients with rare diseases in health information systems on a national and international level.
- Having codes for each rare disease would help European and national health authorities obtain a better knowledge of healthcare pathways and of their impact on specialised health care services (centres of expertise for instance) as well as on a country's budget planning for health and social service.
- The current versions of the International Classification of Diseases (ICD10) and SNOMED include only a limited number of codes for specific rare diseases. Some countries have developed a national extension of these codes to better code, in particular but not exclusively, rare diseases.
- Effective coding would also provide data for clinical research which is needed in this field.
- To ensure that most rare diseases will have a code in the next edition of ICD (ICD11), the European Commission has supported the revision process through the Joint Action to support the Secretariat of the RDTF (N°20082291)/EUCERD and the EUCERD Joint Action (N°20112201). Currently over 5000 rare diseases are listed in the beta version of ICD11. The release of ICD11 is not expected before 2017, although the actual implementation in MS will happen later. As Orpha codes will be linked with ICD11 the switch from ICD10 to ICD11 will be made easier.
- Orphanet gives free access, available under [Creative Commons Attribution-NoDerivs](#) Licence, to a nomenclature of rare diseases, fully aligned with ICD10; ICD11 (beta version), and other nomenclatures including SNOMED-CT, MeSH, MedDRA and OMIM. It is provided in several IT formats to ease integration in different IT systems (xml, OWL, obo) at www.orphadata.org.
- Orpha codes have been already adopted as a complement to ICD10 by two Member States, France and Germany, and in some Italian regions (covering half of the Italian population). France has developed a tool to support the coding process, called LORD. Germany has aligned the Orpha codes with its ICD national extension (ICD10-GM) and plans to add the missing codes in the next two years. DIMDI is developing a file for implementation in existing IT systems which makes coding easy for coders and standardises coding results, if coded in both systems.

Recommendations

In the context of the improvement of codification for rare diseases being cited as a priority in the Council Recommendation on an action in the field of rare diseases, the Commission Expert Group on Rare Diseases recommends the following:

1. The lack of data about rare diseases, due to the absence of codes for most rare diseases, deserves a special effort in epidemiology to make rare diseases more visible in the healthcare systems, in parallel with the ongoing process to incorporate codes for rare diseases in ICD and SNOMED-CT, as this process will not provide full results before several years.
2. An analysis of the number of rare diseases currently coded by each ICD national extension system should be conducted to have a documented picture of the coding situation.
3. The Orpha codes should be further promoted within the development process of WHO's ICD11, in order to allow a seamless transition of RD classification from Orpha codes to ICD-11 when the latter is finally released.
4. MSs interested in using the Orpha codes should set up a working party to define the optimal strategy and the next steps at country and at EU level. This effort should involve representatives of the national and regional coding agencies to explore the details and reach a consensus on solutions to be proposed. It should also involve key stakeholders of the eHealth community, of relevant services at the EC (including the Joint Research Centre Registration Platform), rare disease patient registries, current organisations involved in the development of solutions (Orphanet, DIMDI, BNDMR), and patient organisations.
5. On this basis, MSs should consider adding Orpha codes to their country's health information system and explore the feasibility and resources needed to do so. Rare diseases codification should be included in MS National Plan for Rare Diseases.
6. Interested MSs together with the Commission should seek possibilities to support, at EU level, the implementation process of the identified solutions.