



1st Workshop

Documents on Orphacode implementation

WP 5-Team

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★ <http://rd-code.eu>

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OBJECTIVES OF WP5 OF RD-ACTION

- > **Main aim:** to develop a toolset to assist Member States (MS) in implementing the Orphacodes in their health system
 - ★ Define common objectives for coding RD
 - ★ Provide guidance and common standards
 - ★ Create consistency across MS

MAIN TOOLS & DOCUMENTS ON ORPHACODING IMPLEMENTATION BY WP5 OF RD ACTION

- Standard procedure and guide for coding with Orphacodes
- Master file for statistical reporting with Orphacodes (MF)
- Specifications and implementation manual for MF
- Recommendation for routine maintenance of codification resources for rare diseases

OBJECTIVES OF WP5 OF RD-CODE

- > Exploring the context of the adoption of ORPHAcodes in implementing partners' countries
- > Obtaining feedback from implementing countries regarding implementation of ORPHAcodes
- > Refinement and update of the existing guidance documents for implementation
 - ★ Standard procedure and guide for coding with Orphacodes
 - ★ Specifications and implementation manual for MF
- > Coding of undiagnosed or suspected RD patients
 - ★ Report on existing experience
 - ★ Guidelines proposal on coding
 - ★ Consensus document

WP5 work achieved so far

- > Background Report exploring the “coding environment” of implementing countries
 - ★ available on WP5 part of RD-Code website
- > Standard procedure and guide for coding with Orphacodes, RD-Code 1st update
- > Specifications and implementation manual for MF, RD-Code 1st update
- > Collection of existing experiences about the coding of undiagnosed or suspected rare diseases patients in Electronic Health Records (draft version)

WP5 work achieved so far

- > Background Report exploring the “coding environment” of implementing countries (Deliverable 5.1)
 - ★ Online questionnaire
 - ★ Detailed information about Health Care System, National Plan/Strategy, Morbidity and Mortality coding frameworks, Patient Identifiers, RD Registrations, ERNS, Undiagnosed RD Patients issues
- > Respondents: Spain, Czech Republic, Malta (WP4) plus Austria and Norway (observer countries)
- > Available at <http://rd-code.eu>

WP5 work achieved so far

- > Standard procedure and guide for coding with Orphacodes, RD-Code 1st update
- > Specifications and implementation manual for MF, RD-Code 1st update
 - ★ no feedback from implementing countries so far that implicate any changes to the documents
 - ★ updates on changes of providing MF data by Orphanet included
- > **VERY IMPORTANT:** to get actual feedback from implementing countries on their experiences

WP5 work achieved so far

- > Collection of existing experiences about the coding of undiagnosed or suspected rare diseases patients in Electronic Health Records (draft version)
 - ★ definition of undiagnosed RD patients
 - ★ reasons to identify undiagnosed RD patients
 - ★ existing experiences in coding of undiagnosed RD patients

Standard procedure and guide for coding with Orphacodes

- > **Guideline 1** - Several tools and strategies could be set at MS level to produce data or statistics for RD, nevertheless each country should set this strategy accordingly to a standard principle of maximizing exhaustiveness as well as possible re-use of existing data collections.
- > **Guideline 2** - Code the data in a way that the reporting can compile to the granularity of the international recommended list of Orphacodes (MF-granularity). If no further national needs for reporting are necessary, use the codes from the MF directly.
- > **Guideline 3** - Whenever possible capture the information of the diagnostic assertion for all RD cases. Use the Options “Suspected rare disease”, “Confirmed rare disease” and “Undetermined diagnosis”. Additional options might be helpful.

Standard procedure and guide for coding with Orphacodes

- > **Guideline 4** – Although rare disease registries (disease, population or patient based) should promote the use of data standards to increase interoperability of their data, they should not be the only instruments upon which the EU strategy to produce health statistics for RD at population level relies.
- > **Guideline 5** - Update your coding resource according to the internationally agreed cycle in order to have the most recent coding file and to ensure comparability.
- > **Guideline 6** - If Orphacodes are used together with another national coding system for morbidity coding, the two systems should be linked in a standardized way to ensure that code combinations are standardized and the coding effort for the user is minimized.

Standard procedure and guide for coding with Orphacodes

- > Has the implementation process of Orphacodes in your country been realized corresponding to the guidelines?**
- > Would you recommend any changes or additions to the guidelines based on your experience?**

MF and Specifications and implementation manual for MF

- MF concept: to set a granularity level for statistical reporting with Orphacodes to achieve international interoperability on Rare Disease data

Orphanet nomenclature 2019:

9.320 active ORPHAcodes

2.176 groups of diseases in different hierarchy levels

6.148 disorder level

996 subtypes of diseases

MF and Specifications and implementation manual for MF

- > **Has the MF been used during the implementation process of Orphacodes?**
- > **Do you think the idea of a standalone MF is helpful for the implementation or statistical reporting in your country?**
- > **The planned Revision of the specification and implementation manual will include descriptions on how to access MF granularity from the nomenclature file from Orphanet.**

Do you see the need for a standalone Manual or would you like to see this information included in the Nomenclature file description?