

How to make rare diseases visible in European healthcare systems

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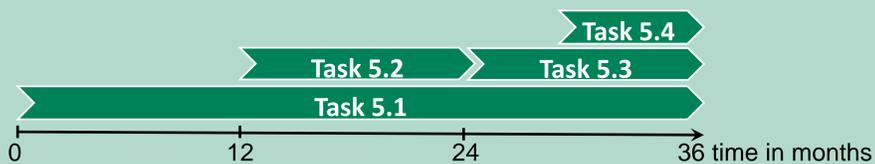
Work Package 5

OBJECTIVES

The approach of the Work package 5 (WP) of the Rare Disease Joint Action, "Steering, maintaining and promoting the adoption of Orphacodes across MS", in order to make rare diseases visible in European healthcare systems is based on the following objectives:

- to identify already existing implementations for coding of rare diseases (RD) in member states (MS)
- to develop standard procedures and a guide for coding with the Orpha nomenclature
- to create a European integrated master file
- to define recommendations for routine coding

TASKS TIMELINE



METHOD

The WP 5 consists of 4 tasks :

- define and set a strategy and tools to implement the Orphacodes in European countries (Task 5.1)
- specify the required resources for coding rare diseases (RD) consistently across Europe (Task 5.2)
- promoting of the Orphacodes across MS by sharing coding tools and testing the master resource (Task 5.3)
- plan for the next steps to address long-term maintenance and sustainability of the resources and guidelines (Task 5.4)

Deliverables are set to ensure the achievement of the objectives:

- Review document of existing technical implementations for RD coding of MS (D5.1)
- Standard procedures and guide for the coding with Orphacodes (D5.2)
- An European integrated master file (D5.3)
- A set of coding helping tools for RD (D5.4)
- Draft recommendation for routine maintenance (D5.5)

RESULTS

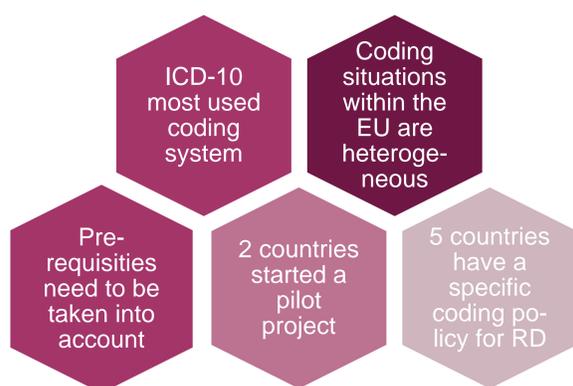
Survey

In the framework of Task 5.1 a survey was performed in September and the results have been published in December 2015.

PARTICIPATION

- 20 countries (5 non-EU countries)
- 26 datasets could be used (54 answers in total)
- All survey participants are also participating in the RD-JA

RESULTS AND CONCLUSION



The implementation of RD coding in health information systems or registries in the framework of the RD-Action is a necessary step to:

- enable wide data analysis
- overcome the actual limit of obtaining sound epidemiological data about RD patients
- avoid biases in epidemiological data analysis

➤ To ensure sustainability and maintenance, the next steps, such as the master file, will be developed with consideration of the survey results.

Master ID	ICD-10-Code Version 2016	ICD-10-Additional (Asterisk)-Code 2 Version 2016	ICD-10-Additional (Asterisk)-Code 3 Version 2016	National morbidity code 1	National morbidity code 2	National morbidity code 3	Orpha Number (03.2016)	Name en	Name de-DE
	Q41.1						1201	Apple peel syndrome	Apple-peel-Syndrom
	Q41.1						1201	%	Apfelschalen-Syndrom
	Q41.9						1201	Atresia of small intestine	Dünndarmatresie
	Q41.8						1201	Jejunoileal atresia	%
	Q41.1						1201	Christmas tree syndrome	%

Figure 1 European integrated master file for standardized coding (please note that this is a draft under discussion)

Deliverables	2015		2016				2017				2018	
	3	4	1	2	3	4	1	2	3	4	1	2
Review												
Coding rules and guidelines					D5.1							
Master file							D5.2					
Coding helping tools												D5.4
Draft recommendations												D5.3
												D5.5

European integrated master file

CONCEPT

A first draft of the European integrated master file has been developed (see figure 1). In the draft the fields Master ID, ICD-10-Code, Orpha Number and the English name of the disease are pre-filled in by the WP 5 team. This should ensure the standardization and central management of the data. Every country then fills in the missing data, such as the national morbidity codes (e.g. SNOMED-CT, national extensions) and matches the name of the disease in their respective language. At the end, with the help of the Master ID the datasets from all countries could be merged centrally to become one master file.

OUTLOOK

After the draft is verified a first version of the master file will be forwarded to participants of the WP 5 for testing and feedback in summer 2016. According to the feedback, the master file will be adapted and further developed for routine coding.