Codification for rare diseases

2019-2021

To take public health action on rare diseases, it is necessary to know how many people are affected by these diseases (prevalence), how these conditions impact those affected, and to monitor rare diseases' medical and societal impact. The lack of basic epidemiology for rare diseases across Europe contributes to a lack of recognition and hinders the development of appropriate services and policies. While about 70% of the world's health expenditures (reimbursement and resources) using ICD-10, only 7% of rare diseases have a specific ICD-10 code. This means that rare diseases are under-reported, under-recognised and under-resourced in health care systems using ICD-10 and other coding systems including ICD-11 and SNOMED-CT. There is a need to implement a specific coding system that makes rare diseases patients visible in health statistics. In 2017 the Steering Group on Health Promotion, Disease Prevention and Management of Non-Communicable Diseases selected the codification of rare diseases as a priority area to be implemented as best practice, and thus a 'RD codification' call was included into the following annual health programme.

In 2019, the RD-CODE project starts as a fine example of transferring best practice across Member States. The objective of this project, coordinated by INSERM (Orphanet-US14) is to support four Member States (Czech Republic, Malta, Romania and Spain) in improving gathering information on rare diseases by implementing ORPHAcodes (rare diseases specific codification system) into their routine codification sytsems.

The work and resources developed by the Workpackage 5 of the RD-ACTION supported and harmonised the implementation process and the process of data exploitation at European level. With the knowledge that has been gained during the RD-CODE project, new rules and guidelines for the use of ORPHAcodes and for sharing data at European level have been defined. The coordination and cooperation between other Member States or projects, as well as with European Reference Networks, will ensure the long term success of the project.

The project is supported by a grant in the frame of the Third EU Health Programme.

Expected outcomes and available results

The aim of the RDCODE project is to promote the use of the Orphanet nomenclature for implementation into routine coding systems. This enables a standardised and consistent level of information to be shared at European level. Starting with countries that have no systematic implementation of the ORPHA codification yet, but that are actively committed to doing so, this project provides a useful real-world implementation experience to be drawn upon other countries in the future. Benefits of implementing ORPHAcodes in Health Information systems are shown in the slideshow here and in the video here.

Implemention in 4 countries

The objective of the implementing countries (**Czech Republic, Malta, Romania and Spain**) is to implement ORPHAcodes in Health Information Systems. To achieve this, **user-friendly technical resources** have been developed and **coder training sessions for more than 100 clinicians** have been organised in order to ensure a **easier and more accurate coding**. The heterogeneity of contexts and settings within the countries will **ease the implementation in other Member States**. All info is available here: **http://www.rd-code.eu/workpackage-4-implementation-in-member-states**

Tool-kits and support

Orphanet Nomenclature material specific for coding purposes is available **http://www.orphadata.org/cgi-bin/ORPHAnomenclature.html**

General information on the Orphanet Nomenclature production and maintenance and links for further reading is available**: http://www.rd-code.eu/helpdesk/**

A page dedicated to answering questions related to the Orphanet nomenclature content and the implementation of ORPHA codes in HIS is now available here: **http://www.rd-code.eu/github/**

A ***Frequently Asked Questions and*** **new services and tools to help implementation and coding decision-making (API & nomenclature/classification visualization have been released: http://www.rd-code.eu/tools/**

Enhancements

Exploring the implementing partners' context for the adoption of ORPHAcodes and getting feedback from implementing countries regarding adoption of ORPHAcodes for RD coding (report available here) is an asset for developing representative guidelines.

Refinement and update of the already existing resources (guidance documents for implementation and exploitation) based on the feedback from implementing countries has been delivered (here)

Guidelines for coding undiagnosed RD patients

To tackle the undiagnosed patients' coding issue a **collection of existing experiences** of coding of undiagnosed or suspected RD patients has been released as well as a **consensus recommendation on codification of suspected and of undiagnosed rare diseases,** in particular by using a specific ORPHAcode.

Target Groups:

- Patient groups, Hospital managers as well as policy makers, including European expert groups (ERNs Board of Member States) will benefit as they will be able to compare more reliable data and identify patients better once this standardised coding approach is used.
- Investigators in clinical research will benefit from a reliable identification of RD patients in health information systems and will be able to capture data from the clinical setting consent allowing.
- Additional target stakeholders have been identified that either currently work in parallel in cross-border rare disease identification for healthcare and research; or who will have future projects in rare disease codification.

These stakeholders require the information and expertise generated by this project, but also by their participation to the rare disease codification community, can help ensure the transposition of the generated results.

For more information about our objectives, achievements and partnerships, consult our website at www.rd-code.eu

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