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 8% 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. 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).

The work and resources developed by the Workpackage 5 of the RD-ACTION will support and harmonise the implementation process and the process of data exploitation at European level. With the knowledge that will be gained in the implementation process, new rules and guidelines for the use of ORPHAcodes and for sharing data at European level can be defined. The coordination and cooperation between other Member States or projects, as well as with European Reference Networks, will assure the long term success of the project.

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

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Expected outcomes

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 will provide a useful real-world implementation experience to be drawn upon other countries in the future.

Implemention in four 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** will be developed and **coder training sessions** will be 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**.

Tool-kits and support

Orphanet Nomenclature material specific for coding purposes will be made available as well as **new services and tools to help decision making and visualise the nomenclature/classification.**

A new Orphanet nomenclature help desk will be made available in 2019: a unique contact point for codification issues and for data, model and technical issues. A ***Frequently Asked Questions*** will be prepared according to the questions received.

Enhancements

Exploring the implementing partners' context for the adoption of ORPHAcodes and getting feedback from implementing countries regarding adoption of ORPHAcodes for RD coding 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 will be delivered.

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 will be produced, a **Guidelines proposal** as well as a **consensus document on codification of suspected/undiagnosed rare diseases** will be produced and disseminated.

Target Groups: Patient groups, decision makers as well as policy makers, including European expert groups (ERNs Board of Member States and the Steering Group on Promotion and Prevention, or SGPP) 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 classification; 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, 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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