RD-CODE

GENERAL PUBLIC REPORT

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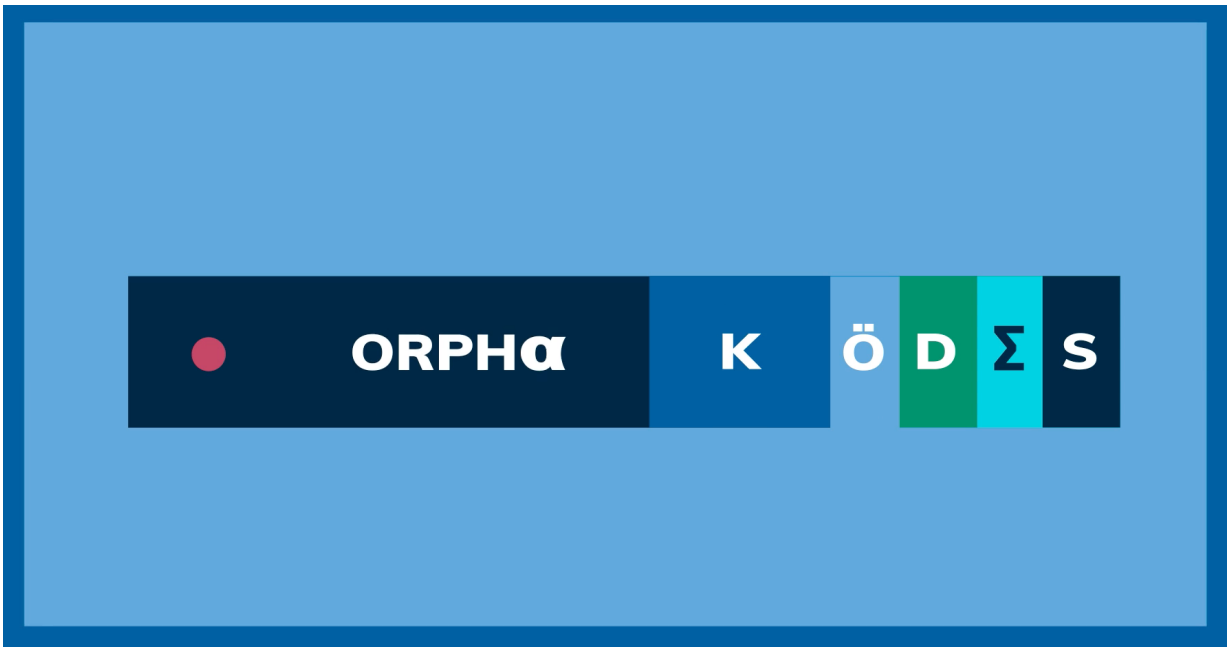
Project Objectives

The Codification for Rare Diseases RD-CODE project, co-funded by the European Union's Third Health Programme, started in January 2019 and ended in December 2021. The objective of this project is to support the Member States in improving gathering information on rare diseases by the implementation of ORPHAcodes, the rare diseases specific codification system. The four countries that were actively committed to developing rare disease codification Czech Republic, Malta, Spain, and Romania successfully implemented ORPHAcodes for the first time. The project has provided sufficient real-world implementation experience for other countries to build on and implement ORPHAcodes in the future. This project has enabled a standardised and consistent level of information on rare diseases to be shared at the European level.

You can watch our project video here.



What are ORPHAcodes?

Rare diseases are numerous and diverse and they require specialised management and treatment.
The commonly used health information terminologies do not include all rare diseases and have difficulty grouping them. The use of only common terminologies for rare diseases in Health Information Systems results in inefficient health system planning and unmet patient needs.

What is a Health Information Terminology?

Health Information Terminology controls the vocabulary used by medical professionals in health systems to describe the diseases the patients are affected by.

Why are these terminologies used?

This terminologies ensure that each disease is referred to with the same name/code. This ensures that all the systems in different settings and countries can understand each other and it allows to collection of robust data for clinic, research, and administrative purposes in order to improve health planning and patients' health pathways.

It is extremely important to know how many people are affected by a given disease and this is even more true for RD. For example, at a national level, governments can negotiate improved pricing structures with manufacturers of extremely expensive rare disease drugs when they know the exact number of people needing treatment. Also, as the global pandemic has shown, governments need exact data on which patients are most vulnerable in order to prioritise vaccination.

This is the reason why it is important that an accurate terminology that defines rare disease diagnosis in medical records is in place. ORPHAcodes is indeed the only terminology that recognises the more than 6, 000 rare diseases and gives each of them a unique, traceable code.

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ORPHAcodes terminology is maintained and curated by ORPHANET at France's Institute of Health and Medical Research. Its production and maintenance have been co-funded by the EC since the early 2000s.

The implementation of ORPHAcodes in information systems is already recognised as best practice by the RARE 2030 foresight study, the European common semantic strategy, and the Europe's Steering Group on Promotion and Prevention of non-communicable diseases. It is also a required data element for the European Patient Summary, patient registries of the European Reference Networks, and European registries as recommended by the Joint Research Centre.

By using ORPHAcodes in national health records and patient registries we will ensure that rare disease data is collected correctly and uniformly. Health data annotated with ORPHAcodes will make it easier for governments to collect and use precise data about rare disease patients, doctors could diagnose and treat patients more accurately and patients can have the correct description of their condition.

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ORPHAcodes also allow countries using different languages and terminologies to communicate with each other.

If every country in Europe used ORPHAcodes in their national health record and patient registries, it would be easy to gather data on patients with the same rare condition in different countries.

This could facilitate the development of new drugs and treatments and create a European space to share and use accurate information about rare diseases. A slide show explaining the benefits of using ORPHAcodes is available here.

What has been done in the 4 implementing countries?

CZECH REPUBLIC

The National Register of Congenital Malformations has implemented ORPHAcodes in its data model. By the end of 2021, two major Czech hospitals were reporting their data to the registry using ORPHAcodes.

874 patients registered with ORPHAcode diagnoses. A comparison between the period 2016-2017 and the Project implementation period has shown a16 fold increase in the number of RD cases identified and diagnosed (from 57 cases to 874 cases).

3 Training sessions held and a Moodle available. In total, 44 clinicians trained.

Mid-term Report and Outcome report available here.

MALTA

Introduction of automatic ORPHAcoding into the data model of three existing registries: congenital malformations registry, cancer registry and the Treatment abroad data.

5,600 patients registered with 1,007 different ORPHAcode diagnoses. A comparison between the period 2016-2017 and the Project implementation period has shown a 1.75 fold increase in the number of RD cases identified and diagnosed (from 800 cases to 1,400 cases/year).

An ORPHAcodes Workshop with the 16 Mater Dei Clinical departments, government representatives, the Directorate for Health Information & Research Registries managers, Hospital Information Systems specialists and consultants was held with 36 participants in total.

A Workshop report is available as well as an outcome report here.

ROMANIA

4 centres are ORPHAcoding: Iasi Regional Medical Genetics Centre , C. I. Parhon Hospital Iasi – Nephrology Department, Timis Regional Medical Genetics Centre and Prader Willi Association - NoRo Expert Centre for Rare Diseases.

1,137 individual diagnoses registered with 193 different ORPHAcodes. A comparison between the period 2016-2017 and the Project implementation period has shown a 5-fold increase in the number of RD cases identified and diagnosed.

4 online training sessions and 30 clinicians trained.

A mid-term report and an outcome report are available here.

SPAIN

6 Regional registries are ORPHAcoding (Comunitat Valenciana, País Vasco, Cataluña, Castilla y León, Madrid and Murcia) and the Sant Joan de Deu Hospital

3,271 ORPHAcodes in use by Spanish registries. A comparison between the period 2016-2017 and the Project implementation period has shown an increase in the number of RD cases identified and diagnosed with 100% of the cases communicated to the national registry with an ORPHAcode; and 95% of the ORPHAcodes from the Master file have an ICD-10-ES (version 2020) equivalent.

95% of the Master file aligned to ICD10-ES

A mid term report and an outcome report are available here.

What is available for all the countries wishing to implement ORPHAcodes into their health information systems?

Orphanet Tools

Orphanet maintains the www.rd-code.eu website where all the RD-CODE related information is available. On the website's ORPHAcoding page you can find information on the Orphanet nomenclature and its related products. Information on guidelines, Orphanet server for nomenclature, Orphanet procedures, and other topics are presented with supporting links to documents and websites of interest providing more detailed information. From this page you can also access the Orphanet tools developed during the project to facilitate flexible implementation into the various IT systems in use in the different countries and/or settings. Find our toolbox below.

since July 2019, Orphanet also releases the Nomenclature Pack annually. These files provide the computable information necessary to achieve the implementation of ORPHAcodes in Health Information Systems, and ensure an easier and more accurate coding. The Pack has been upgraded in July 2021 according to the users' feedback from the multi-stakeholder workshop held in Prague in 2021. In order to help users in the implementation process, a detailed description is produced to provide information on the usage of the nomenclature according to the end-users and implementation settings. This document gives a detailed description of the content of Orphanet coding-dedicated nomenclature files (including classifications files and Orphanet-ICD10 alignments) and the ways to explore them. This document is downloadable for free and supplied with the Nomenclature Pack.

Update of coding guidelines

Following from the RD-ACTION project, a specific working group in the project led by Veneto Region and with the participation of APHP and DIMDI/BfARM was responsible for developing additional necessary rules and guidelines for rare diseases codification using ORPHAcodes across Member States (MS).

Surveys and two RD-CODE Multi-stakeholder workshops have exploring the context of the adoption of ORPHAcodes in implementing partners' countries and obtained their feedback. The working group have used this feedback to refine and update the guidance documents for ORPHAcodes implementation and exploitation to develop up-to-date, effective guidelines.

RD-CODE project outputs are available for further reading here.

- The Background Report on the "coding environment" of implementing countries has been prepared by the WP5 participants, lead by Veneto region, and with the contribution of WP4 participants. It was released in July 2019.
- The document « Specification and implementation manual of the Master file for statistical reporting with ORPHAcodes » has been updated, together with the new release of the Master-file.
- A Report on implementing countries ORPHAcodes adoption: limits and opportunities was released at the end of the project.
- Refined versions of the "Standard procedure and guide for the coding with ORPHAcodes" was also delivered at the end of the project.

Delivery of guideline for undiagnosed patients

To tackle the issue of coding undiagnosed patients' the RD-CODE project set up a multi-stakeholder working group (including SOLVE-RD, X-eHealth and ERN representatives) to issue.

- A collection of existing experiences of coding of undiagnosed or suspected RD patients
- A guidelines proposal
- A consensus document on codification of suspected/undiagnosed rare diseases

You can find the documents here

Beyond RD-CODE

Through implementation of ORPHAcodes, RD-CODE partners have gained valuable experience that can be of benefit to others. The partners and other stakeholders present at the final Multi-Stakeholder workshop have agreed to explore the possibility of creating a virtual **Community of Practice** to build the capacity of other organisations interested in implementing ORPHAcodes. This Community will also allow the partners to continue sharing coding experiences, training materials and tools, as well as review and update the guidance documents as needed.

The success of such a Community is highly dependent on the commitment of RD -CODE partners and others to dedicate time and resources to animate the group, facilitate information sharing and build the Community over time.

In addition, RD-CODE results will benefit other projects and initiatives around health data, making sure that rare diseases are not left behind. These groups include:

- EU X-eHealth project, establishing standards in the European electronic health record exchange format for eHealth, including those for rare diseases.
- European University Hospitals Alliance (EUHA) and the European Children's Hospitals Organisation (ECHO), who are an integral foundation for;
- European Reference Networks for Rare Diseases registries and the ERICA project.
- The European Health Data Space.

WWW.RD-CODE.EU

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