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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 on January 2019 and ends in December 2021.

The objective of this project is to support Member States in improving gathering information on rare diseases by the implementation of ORPHAcodes, the rare diseases specific codification system. Four countries that had no systematic 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 in the future. This enables a standardised and consistent level of information on rare diseases to be shared at European level.

You can watch our project video [here](#).

WHAT ARE ORPHAcodes?

Rare diseases are numerous and diverse, and require specialized management and treatment.

The commonly used health information terminologies do not include all rare diseases, and have difficulty grouping them. Use of only common terminologies for rare diseases in Health Information Systems result in inefficient health system planning and unmet patient needs.

WHAT IS A HEALTH INFORMATION TERMINOLOGY ? a controlled vocabulary used by medical professionals in health systems to describe diseases the patients are affected by.

WHY HEALTH INFORMATION TERMINOLOGIES ARE USED ? Their purposes are multiple : they ensure that each disease is referred to with the same name/code. This ensure that all the systems in different settings and countries can understand each other and it allows to collect robust data for clinic purposes, research purposes 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 prioritize 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 recognizes the more than 6, 000 rare diseases and gives each of them a unique, traceable code.

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.

ORPHAcodes also allows countries using different languages and terminologies to communicate.

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 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?

<p>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.</p>	<p>3 Training sessions held and a Moodle available. In total, 44 clinicians trained.</p>
<p>Czech Republic UZIS</p>	
<p>By the end of 2021, 874 new cases from these hospitals had been coded using ORPHAcodes, approximately 290 per year. Compared to the period 2016-2017, the number of cases with a rare disease diagnose inserted in the national registry has increased more than 16 times (2016-2017: 57 rare disease cases inserted in the registry compared to 965 rare disease cases inserted in the registry in the period 2019-2021).</p>	<p>Mid-term Report and Outcome report available here: http://www.rd-code.eu/workpackage-4-implementation-in-member-states/</p>

<p>Introduction of automatic ORPHAcoding into the data model of three existing registries: congenital malformations registry, cancer registry and the Treatment abroad data</p>	<p>A ORPHAcoding Workshop was held representatives from the 16 Mater Dei Clinical departments, government representatives, all Directorate for Health Information & Research Registries managers, Hospital Information Systems specialists and consultants . 36 participants in total.</p>
<p>Malta Ministry of Health- Mater Dei Hospital</p>	
<p>5,600 are patients registered with 1,007 different ORPHAcodes 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).</p>	<p>A Workshop report is available as well as an outcome report here: http://www.rd-code.eu/workpackage-4-implementation-in-member-states/</p>



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.

4 online training sessions and 30 clinicians trained

**Romania
DSP Iasi**

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.

A mid-term report and an outcome report are available here:<http://www.rd-code.eu/workpackage-4-implementation-in-member-states/>

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

95% of the Master file aligned to ICD10-ES

**Spain
CIBER-FISABIO**

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 with an ICD-10-ES (version 2020) equivalent.

A mid term report and an outcome report are available here:<http://www.rd-code.eu/workpackage-4-implementation-in-member-states/>

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 provide 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:

- [the Nomenclature Pack](#) (more info below)
- [the Orphanet API](#)
- [the DataViz tool](#)

Orphanet has also released the [Nomenclature Pack](#) annually since July 2019. These files provide the computable information necessary to achieve the implementation of ORPHAcodes in Health Information Systems, and to ensure easier and accurate coding. The Pack has been upgraded in July 2021 according to the users' feedback from the [Multistakeholder 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 (<http://www.orphadata.org/cgi-bin/img/PDF/ORPHAnomenclaturexmlcontent.pdf>) 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 [Multistakeholder](#) workshops have exploring the context of the adoption of ORPHAcodes in implementing partners' countries and obtained their ing 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: <http://www.rd-code.eu/workpackage-5-enhancements/>

- 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' a three works have been produced:

- 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 at: <http://www.rd-code.eu/workpackage-5-enhancements/>.

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](#)