

# Codification for Rare Diseases: the RD-CODE project support to Member States and beyond

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## Introduction

To take public health action on rare diseases (RD) and transform care for RD, it is necessary to:

- > Know how many people are affected
- > The impact on those affected
- > To monitor the medical and societal impact

The commonly used coding systems, including ICD-10, are far from exhaustive in terms of RD coverage and do not allow statistical analysis for all RDs. The ORPHAcodes is the only coding system that makes ALL rare diseases visible in health information systems.

**The RD-CODE project (2019-2021), co-funded by the European Union's Third Health Programme, supports 4 Member States in improving gathering of information on rare diseases by implementation of ORPHAcodes into routine coding systems.**

## Methodology

### ★ Background:

Recommendations produced by the previous European Joint Action, RD-ACTION

### ★ Who:

Czech Republic, Malta, Romania and Spain

### ★ Goal:

implement the ORPHAcodes in country-specific settings: regional codification systems, national registries or main hospitals. Develop userfriendly tools and educational material for the implementation.

### ★ How:

Support is provided by Orphanet (tools and helpdesk) and by AP-HP (France), Veneto region (Italy) and DIMDI (Germany) for guidelines.

## Results

- ★ **Real-life Implementation in 4 Member States since the beginning of the project early 2019**
- ★ User-friendly practical coding resources and coder training have been developed in the 4 countries.
- ★ Effective coding of RD patients has started overall in 2 pilot Hospitals in Czech Republic, 1 in Romania, 1 in Malta and in 5 regional registries in Spain.

### ORPHANET NOMENCLATURE FILES FOR CODING TOOLSET

- > Provide computable information necessary for implementation of ORPHAcodes in Health Information Systems, to ensure easier and accurate coding.
- > Files produced according to the [Recommendation for routine maintenance of codification resources for rare diseases](#) (RD-Action Joint Action 2018).
- > Files underwent review by RD-CODE project partners and key stakeholders.

The toolset includes:

1. the Orphanet nomenclature file,
2. the Orphanet – ICD-10 cross-referencing file,
3. a directory of Orphanet classifications files.

**Download the toolset and supporting documents from ORPHADATA**  
(<http://www.orphadata.org/cgi-bin/ORPHAnomenclature.html>)

- > Available in 9 different languages: Czech, Dutch, English, French, German, Italian, Polish, Portuguese and Spanish.
- > Delivered annually (July), with descriptive documentation.
- > Previous versions are stored, and a log of changes are made available to ensure traceability.

### GUIDANCE FOR CODING UNDIAGNOSED PATIENTS

Work is ongoing collecting experiences of coding undiagnosed or suspected RD patients. A consensual guidance document on how to better code them will follow later in 2020.

### TECHNOLOGICAL SOLUTIONS

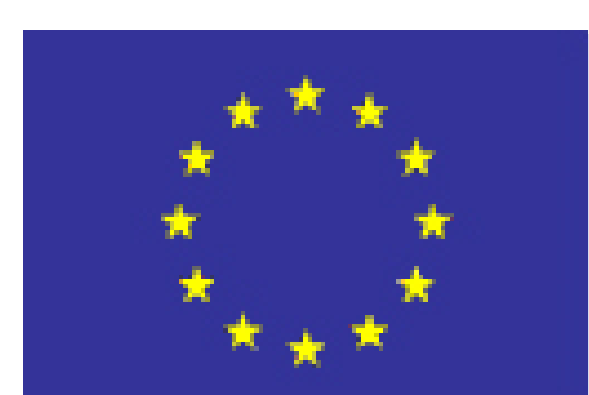
- > An **ORPHAcodes API** is available to add into healthcoders' systems. This free module allows coders to query the API to obtain the specific ORPHAcodes for their situation.
- > **RD-CODE Helpdesk**: live and dedicated to answering questions related to the Orphanet nomenclature content and the implementation of ORPHAcodes in Health Information Systems. An online ticketing system allows requests to be stored, tracked and made visible to others.
- > **RD-CODE Frequently Asked Questions web-page**: regularly updated with the most frequently asked questions sent to the Orphanet team

Updated documentation on ORPHAcodes:

- > Refinement and update of the existing RD-ACTION resources: such as ['Recommendation for routine maintenance of codification resources for rare diseases'](#)
- > New reports: [Background Report on the "coding environment" of implementing countries](#)
- > All technological solutions and documents available on:

★ [www.rd-code.eu](http://www.rd-code.eu)

**Conclusion:** Stakeholder involvement, including the experiences of expertise of ERNs, as well as countries developing ORPHAcodes use, such as Austria and Norway, is key to RD-Code. This collaboration ensures that all the resources developed during the project can be useful to any country wishing to implement ORPHAcodes in the future and ensures transfer of best practice across Member States and beyond. Results of implementation will be also published at the end of the project in June 2021.



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