



RD-CODE project overview



Prague - 22 June 2020



★ <http://rd-code.eu>



This presentation is part of the project 826607 RD-CODE which has received funding from the European Union's Health Programme (2014-2020).

The content of presentation represents the views of the author only and is his/her sole responsibility; it can not be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.



Improved codification for rare diseases is cited as a priority in the Council Recommendation on an action in the field of rare diseases (2009)

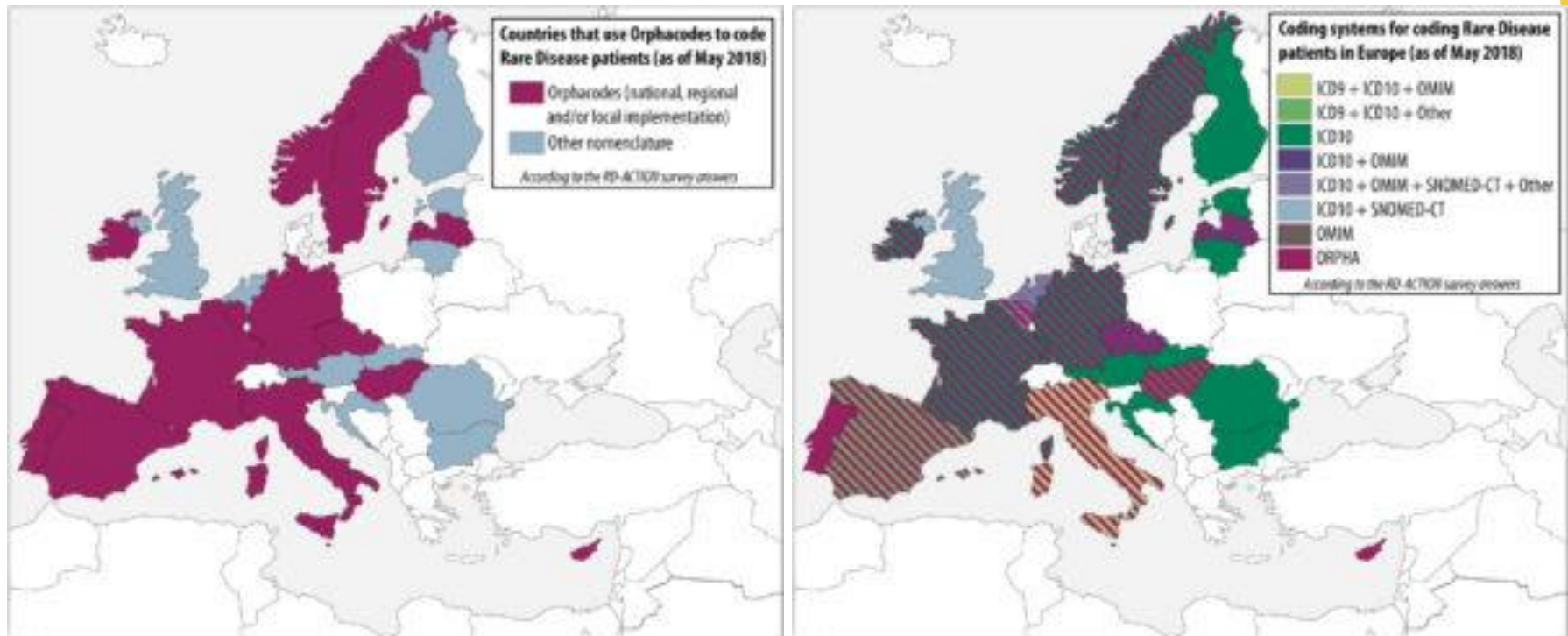
Steps so far in Europe

- > **2009-2012 RDTF Joint Action:** support cross-referencing terminologies & ICD11 revision process for RD
- > **2012 on: EUCERD Joint Action:** continuation of the RDTF work
 - ★ Sept 2012: EUCERD JA workshop on cross-referencing terminologies
 - ★ March 2014: EUCERD JA workshop on Orphacodes in HIS
- > **July 2014: CEGRD:** draft recommendation on codification for rare diseases
- > **October 2014 (JRC-ISPRA):** Workshop on the next steps to promote the implementation of Orphacodes in MS
- > **November 2014:** Adoption of CEGRD recommendation for codification of RD (adoption of ORPHA code by MS)
- > **2015-2018:** RD-Action WP5: Guidance for implementing Orphacodification

Rare disorders in Orphanet

- > Since 1997: Inventory of rare disorders (prev<1/2 000)
 - ★ Mapped to OMIM
- > 2005: Mapping to ICD-10
- > 2007: Classification of rare disorders
- > 2009: RD-TAG in ICD-11 revision: start mapping ORPHA-ICD11
- > 2011: Mappings to UMLS, SNOMED CT, MeSH, MedDRA
- > 2014: ORDO (Orphanet ontology of rare diseases) in collaboration with the EBI.
- > 2015: Collaboration agreement with IHTSDO (SNOMED CT-ORPHA mapping file)
- > 2018: HOOM (HPO-ORDO ontological module)

Europe map of RD coding (2017)



<http://www.rd-action.eu/workpackage/workpackage-5/>

RD-CODE partners

 **Inserm** orphane**net**

 **ciberer**

 **ÚZIS**

 Fundació per al Foment de la
Investigació Sanitària i Biomèdica
de la Comunitat Valenciana

health.gov.mt

 **DSP IASI**
Direcția de Sănătate Publică Iași



 **REGIONE DEL VENETO**

 **DIMDI**
medical knowledge
German Institute of Medical
Documentation and Information
BfArM

 **BNDMR**
Banque Nationale de Données
Maladies Rares

 **EURORDIS**
RARE DISEASES EUROPE

Maastricht University (on behalf of the EMRaDi project)

HSE

HL7 International Foundation

Consejería de Salud. Región de Murcia.

Servei Català de la Salut (CatSalut) (in collaboration with a Rare Diseases Assessor Group)

Departamento de Salud del Gobierno de Navarra (Regional Department of Health of Navarra)

Dirección General de Salud Pública de la Junta de Castilla y León

Departamento de Salud, Gobierno Vasco (Health Department, Basque Government)

Institute of Mother and Child

Dirección General de Salud Pública. Generalitat Valenciana

UMF-IASI



ERKNet

ERNICA

ERN CRANIO



MetabERN

European Reference Network
for Hereditary Metabolic Disorders



ERN BOND

Connective Tissue and Musculoskeletal
Diseases (ERN ReCONNET)

RD-CODE objectives

- > To promote the use of the Orphanet nomenclature for implementation into routine coding systems.
- > To allow for a standardised and consistent data and information sharing at the European level
- > To provide a useful real-world implementation experience to be drawn upon other countries in the future.

Expected outputs

Implementation 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** 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.

Where are we now?

Implementation

- Spain
- Czech Republic
- Roumania
- Malta

Support

- Helpdesk
- Nomenclature pack
- Codification tools

Guidelines

- Guidance documents for codification and exploitation
- Standard procedures and guide for Orphacoding
- Consensus document on codification of undiagnosed patients

1/2019

lessons



learned

6/2021

My wishlist

- > Everyone has a clear picture of what works and what is difficult in implementation experiences:
 - ★ towards operational solutions
- > Closer interactions are in place between implementers, support providers and recommendations teams:
 - ★ towards transferable lessons from the field
- > Operational solutions are found for giving undiagnosed RD patients visibility in HIS
 - ★ towards applicable recommendations

« If we cannot count rare diseases patients,

Rare diseases patients do not count »

Gareth Baynam

RD-CODE: implementing Orpha codification in EU countries

LET'S HAVE FRUITFUL DISCUSSIONS