

## RELEASE OF THE NOMENCLATURE PACK

JULY 2019

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The Orphanet nomenclature is used to code the diagnosis of a rare disease with a unique and time-stable identifier, the ORPHANumber (also named ORPHAcode) in order to facilitate data collection, research and analysis. These files are available [here: http://www.orphadata.org/cgi-bin/ORPHAnomenclature.html](http://www.orphadata.org/cgi-bin/ORPHAnomenclature.html)

They provide the computable information necessary to achieve the implementation of ORPHANumbers in Health Information Systems, and ensure easier and accurate coding. Orphanet provides a set of files in XML format, including: the Orphanet nomenclature file, the Orphanet ICD-10 mapping file and a directory containing the Orphanet classifications files. These files are generated and made available once a year, in 9 different languages: Czech, Dutch, English, French, German, Italian, Polish, Portuguese and Spanish. The Polish version should be available very soon. The previous versions and their related change logs remain accessible and downloadable in the dedicated RD-CODE Github repository.

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*« Release of the Nomenclature pack: these files provide the computable information necessary to achieve the implementation of ORPHANumbers in Health Information Systems, and ensure easier and accurate coding. »*

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## TO DO LIST

✓ Remember to record the time spent on the project by the staff listed in the Grant Agreement. For personnel costs declared as actual costs the beneficiaries must keep time records for the number of hours declared. A time record template

is available [here](#). Alternative evidence supporting the number of hours declared can be accepted by CHAFEA if it considers that it offers an adequate level of assurance. Please contact us if you have a doubt concerning your time record system or use the template provided.

## WORKPACKAGE1

The US14-Inserm is coordinating the project, and the objective of this Workpackage\* is to facilitate and ensure the efficient implementation of actions foreseen, based on effective cross-talk between the 5 work packages within the project.

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**"Deliverable1.1  
completed."**

*Milestones update:*

Mls1: organisation of the KOM is completed. **REACHED**

Mls2: Steering committee meetings **REACHED**

## WORKPACKAGE 2

Within this Workpackage (WP leader: Inserm, Participants: CIBERER and IHIS) are the actions undertaken to ensure that the results and deliverables of the project will be made available to the target groups.

Engagement of the stakeholders in early consultation exercise to assess their needs will be carried out, in order for the material developed to be effective and useful.

- Orphanet has launched a survey in order to better tackle the needs of the users in regards of the development of a new Orphanet API

Moreover satisfaction on material produced and suggestion for improvements will be sought.

**Please save the date for the first RD-CODE Multi-stakeholder Workshop to be held in Prague on the 29-30 April 2020**

The Achievements leaflet of the project is available [here](#)

News regarding the release of the Nomenclature PAck have been published in Orphanews and on [www.rd-code.eu](http://www.rd-code.eu)

*Milestones update:*

Mls3: Website layout and content validated by the Steering committee **REACHED**

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**"SAVE THE DATE  
29-30 April RD-  
CODE  
Multistakeholder  
workshop."**

## WORKPACKAGE 3

This Workpackage comprises actions undertaken to verify if the project is being implemented as planned and reaches the objectives . Evaluation will be carried out with quantity (according to the defined indicators and targets) and quality approaches (to assess the needs, and the improvement suggestions) .

A partners'survey is being prepared.

WP leader: EURORDIS

*Milestones update:*

*MIs5: KOM participant survey **REACHED***

## WORKPACKAGE 4

The objective of the implementing countries (**Czech Republic, Malta, Romania and Spain**) is to implement ORPHA codes in Health Information Systems . To achieve this, **user-friendly technical resources** will be developed and **coders trainings** will be organised in order to ensure an **easier and more accurate coding**. The heterogeneity of contexts and settings within the countries will **ease the implementation in other Member States**.

### **Czech Republic update:**

In the pilot phase of this project, field research was carried out at individual workplaces. During the interviews deficiencies were identified that slowed down or even prevented the entering rare disease cases to the NRVV web interface. We are currently working on eliminating these deficiencies (e.g. better formulation of some items in the registry, removal of login problems, creation of a clear manual for entering into the registry, translation of missing terms, updating of codebooks). At the same time, the first cases were entered into the registry to verify the flow of information about the given cases. Currently data collection is also continuing successfully. So far **120 rare disease cases were entered into the National Register of Congenital Malformations through the web interface from Czech coding centers in The General University Hospital in Prague (VFN) and Motol University Hospital**. Feedback meeting are held to discuss difficulties and other discrepancies with the coders to make sure that the data flow will be fluent after involving a higher number of coders into the data entering.

For the easier implementation of ORPHA codes and the successful coding process, the **courses for coders** have been prepared and will be held in the first half of the year 2020. Powerpoint presentations and detailed manual for coders for entering rare disease cases into the web interface

of the National Register of Congenital Malformations were finalized. This manual was also placed on our website <http://www.uzis.cz/projekt-rd-code>. The schedule of the course was prepared with four lecturers in charge of these courses.

#### **Malta update:**

The Review of the current data collection organization for rare diseases will be organised in 2020.

The **Development of a technical solution** is in progress and should be completed towards the end of 2020.

A **Training course session** is being organised and will be delivered by the end of the first trimester of 2020

We were hoping that this activity would be a follow-on from the expert's review in 4.2.1

As a result of this no formal contact has yet been made regarding training material .

#### **Romania update:**

The IT system is working and case registration is ongoing. A report will be delivered by the end of December.

#### **Spanish Update:**

At the beginning of April a meeting was held , procedures were discussed and agreed by all regions. 2 Working groups were set up for codification and informatics.

All regions, FISABIO and CIBERER agreed to work with **Spanish version of the Master file**. Mapping of ORPHA codes included in the MF to ICD-10-ES (Spanish version of ICD-10-CM). **To date, equivalences for around 5300 RD have been proposed (and agreed with the participant regions). This represents close to 90% coverage.**

The unsolved equivalences between ORPHA codes and ICD-10-ES (500 approx.) remain under study and a strategies to solve the mapping of this last 10% is being discussed.

**The MF with the mapped equivalences is in use by the regional registries in order to test the feasibility of its implementation in their information systems.** Reports from the regional registries about the results obtained in the pilot implementation phase are expected at the end of 2019.

The participant regions have initiated the adaptations to the IT system necessary in each case and their compromise is to have them ready by the end of the year.

In parallel, Spanish RDCODE participants are in touch with the Ministry of Health to explore the possibility of coordinating the mapping strategies with another initiative that is being carried out by the Spanish National Reference Center to map ICD-10-ES / ORPHA / SNOMED-CT.

### **Orphanet update:**

**Orphanet Nomenclature Pack has been released in July 2019** while new services and tools to help decision making and visualise the nomenclature/classification will be released soon.

**Orphanet nomenclature help desk** was made available in July 2019: A page with general information on the Orphanet Nomenclature production and maintenance and links for further reading is available: <http://www.rd-code.eu/helpdesk/>

Moreover a **page dedicated to answering questions related to the Orphanet nomenclature content and the implementation of ORPHA codes in Health Information Systems is now available here:** <http://www.rd-code.eu/github/>

For this purpose, an online ticketing system that allows requests to be stored, tracked and made available to others has been set up. This system facilitates an agile and interactive workflow.

Finally a page where can be found **tools which will provide the computable information necessary to achieve implementation of ORPHANumbers in Health Information Systems**, and ensure easier and accurate coding is available here: <http://www.rd-code.eu/tools/>

A **Frequently Asked Questions** will be prepared according to the questions received.

Finally Specifications for the API allowing **to automatise the access to the Orpha nomenclature and classifications** have been delivered and the API will be made available by the end of December as planned.

*WP leader: CIBER Participants: IHIS, MFH Malta, DSP IASI, Orphanet*

*Milestones update since last issue:*

*MIs 9 IT tool development (RO) **REACHED***

*MIs 10 Preparation of implementation in Pilot centre **REACHED***

*MIs 12: Report on survey of Orphanet awareness and needs from the 2 departments most likely to submit to the registry. **REACHED***

*MIs 13: Orphanet nomenclature contact point operational **REACHED***

*MIs 14: Draft general documentation available **REACHED***

MIs 15: Specifications for Facility for ORPHA nomenclature correspondence available **REACHED**

MIs 16: toolkit available **REACHED**

## WORKPACKAGE 5

**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 and it is available [here](#). It is organized into 4 parts describing the coding starting situation of three RD-CODE participant countries (Czech Republic, Malta and Spain) and of Austria (RD-CODE observer). For each country, an overview of the following themes is provided: national RD policy framework, coding framework, RD registration, Centres of Expertise/ERNs, undiagnosed RD patients. Contents are based on the answers provided by country respondents to the on-line survey, which constituted MIs 5.1. Additional material consulted to prepare the Report is listed in the Bibliography section.

Furthermore regarding the delivery of the milestone 5.2 First Report of necessary changes to the Master file and Standard procedures and guide for the coding with ORPHAcodes according to the results of the first workshop: these do not implicate any necessary changes to the Master file and the Standard procedures and guide for the coding with ORPHAcodes document. A discussion about a possible necessary change of the format of the “Master file for statistical reporting with ORPHAcodes” is under consideration. We will report on the changes when a decision is made and technical and content details are finalized.

**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** will be issued as well as a **Consensus document on codification of suspected/undiagnosed rare diseases** will be produced and disseminated. A general document presenting the concepts and the main issues for coding undiagnosed patients in health information systems has been written. It focuses on the coding experiences recommended by the JRC, and the one implemented in France and in Italy. More countries will be contributing to this document in the following months to have a fully complete deliverable 5.2.

*WP Leader: Regione Veneto Participants: APHP; DIMDI*

*Milestones update:*

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**" Deliverable 5.1  
Report explore  
the “coding  
environment” of  
implementing  
countries  
available"**

## RD-CODE News

**IF YOU HAVE ANY  
QUESTIONS:**

**DO NOT HESITATE  
TO CONTACT US**

[pmt.orphanet@inserm.fr](mailto:pmt.orphanet@inserm.fr)

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

*Mls23: First Report of necessary changes to the Master file and Standard procedures and guide for the coding with Orphacodes according to the results of the first workshop **REACHED***

*Mls24: Collection of existing experiences about the coding of undiagnosed rare diseases **REACHED***

## STEERING COMMITTEE NEWS

In order to guarantee an integrated and coherent approach within the project and promptly address any challenges or emerging risks, a steering group composed of all the WP leaders will meet every two months at distance. WP leaders should liaise with the other participants in their WP in order to collect their feedback (Ideally this should be done through regular distant meetings scheduled before the SC meeting and as often as necessary).

The executive committee bi-monthly conference call was held on the 13/05 ; the 01/10 and the 03/12. Read the full reports [here](#).

## OTHER INFO

**NTR**

## WHAT'S NEW ON THE INTERNAL WEBSITE

All material can be found [here](#).

[RD-CODE logo](#)

[RD-CODE presentation template](#)

[RD-CODE KOM report](#)

[RD-CODE Dashboard and tracking table of indicators](#)

[RD-CODE steering Committee reports](#)

[RD\\_CODE deliverable template](#)

[Nb. RD-CODE Grant agreement available on ECAS](#)

[RD-CODE consortium agreement: send an email to \[sylvie.maiella@inserm.fr\]\(mailto:sylvie.maiella@inserm.fr\)](#)