

MULTISTAKEHOLDER WORKSHOP JUNE 2020

FIRST MULTISTAKEHOLDER WORKSHOP

The first RD-CODE multistakeholders workshop was held in Prague on the 22-23 June 2020. It was an « hybrid event », with participants attending from Czech Republic meeting « *de visu* » at the Prague Congress Centre and participants from other countries attending remotely.

The objectives of this workshop were multiple such as learning about the RD-CODE project and [Implementing countries specific activities](#), and to be able to have a forum to discuss country specific experiences and challenges with advice from WP5 team, collaborating partners and external experts. It was also an occasion to disseminate the projects results such as the availability of the [Nomenclature Pack and of the Orphanet API](#) to a wider audience and to collect feedback on how to adapt all Orphanet Codification tools to their real-life use. This workshop provided also an important forum to obtain feedback from implementing countries to [allow refinement and update of the already existing guidance documents for implementation and exploitation:« Standard procedure and guide for the coding with Orphacodes » and the « Specification and implementation manual of the Master file »](#) both developed in the frame of the previous Joint Action on rare Diseases RD-ACTION (2015-2018)). Finally a final session dedicated to liaising with other relevant projects provided an opportunity to exchange with SOLVE-RD experts on the topic of "undiagnosed patients" as well as with eHealth experts. It was also decided that a dedicated workshop needed to be held to address the topic of codification of undiagnosed patients. Full Agenda available [here](#). Slides available [here](#). Report will be available soon!

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.

As decided at the Steering Committee in September, the Coordinating team launched a vote/poll between beneficiaries in order to decide whether to launch an Amendment request for 3 or 6 months. Indeed, following the outbreak of COVID-19 coronavirus in the EU and elsewhere, our grant beneficiaries have experienced difficulties in carrying out implementation of their projects.

Milestones update:

MIs2: *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.

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

News regarding the release of the API have been published on www.rd-code.eu; as well as regarding the Training course held in Czech republic and the Multistakeholder workshop.

Milestones update:

MIs4: *preparation of Multistakeholder Workshop 1* **REACHED**

WORKPACKAGE 3

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

The [RD-CODE project mid-term evaluation report available](#) since April 2020.

Satisfaction survey of Multistakeholder Workshop participants also available [here](#): 95% of the respondents thought that the Workshop either met their expectations (59%) or exceed it (36%).

WP leader: EURORDIS

Milestones update:

*MIs6: mid term participant survey **REACHED***

WORKPACKAGE 4

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** are being developed and **coders trainings** are being 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:

A progress report was delivered (**Czech republic Progress report D4.6**) and is available [here](#).

For the easier implementation of ORPHAcodes and the successful coding process, the [courses for coders](#) (mIs 4.8) have been prepared and the first one was held in February 2020. Due to the sanitary crisis new trainings will be held remotely. All material is available on our website <http://www.uzis.cz/projekt-rd-code>.

Malta update:

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

The **Training course session** scheduled in March 2020 had to be cancelled because of the pandemic. The session was post-poned in October 2020 as a remote session but unfortunately it had to be cancelled as the RD-CODE

Maltese team had to go back to 100% clinical duties in the context of the sanitary crisis in the country. The Steering Committee is currently proposing mitigating actions in order to ensure that the projects progresses.

Romania update:

The progress report was delivered at the end of December (**Romanian Progress Report D4.1**, available [here](#)) together with an update at the Multistakeholder Workshop in June.

After developing the IT tool, young physicians have been trained to introduce cases and they started uploading cases that have been evaluated in 2019 –precise diagnosis of rare disease –more than 200 cases recorded as of July 2020; an evaluation of the section of reports and statistics was also carried out.

Unfortunately due to COVID 19 pandemics all the activity was paralysed for several months.

Now training sessions are planned at distance.

Spanish Update:

Delivery of the "Preliminary results analysis and "Adaptation of procedure for ORPHAcodes use in Spain", D4.4 ([here](#)). This analysis main findings show that the goal to have 75% of the cases with an ORPHAcodes was fulfilled because all the RDs submitted to the national registry have now a matching ORPHAcodes. It also shows that adaptation to ORPHAcodes although possible, is limited by the current codification tools. Efforts towards the establishment of equivalences allow not only to approach the target of systematically report RDs but also help realize which are the assets of currently employed systems and, moreover, their flaws and lacks.

Extension of the project to other countries (mIs 19). The preliminary phase consisting in piloting the use of ORPHAcodes by six regional RD registries in Spain, concluded in the first trimester of 2020. Some other regions in Spain expressed their interest in participating and they were partly involved during the first year of the project. During the first months of 2020, our colleagues in the project from FISABIO contacted other regional registries to identify which ones would be interested in joining the second implementation phase. 3 new regions were onboard: Madrid, La Rioja and Galicia.

A face-to-face meeting was being organized with all the participating regions (including the newly incorporated ones) in June 2020. However, for the time being, the COVID-19 pandemic outbreak makes impossible to

plan such a meeting. In the meantime, we are exploring the way to have the three new regions initiated in the work.

Orphanet update:

An API <https://api.orphacode.org/>, was released as a beta version in December 2019 (**API for visualisation of Orphanet Nomenclature Deliverable 4.2**) and then an upgrade was made in July 2020 (**Facility for ORPHA nomenclature correspondence, Deliverable 4.5**). This newly available version allows for the identification of the correspondence between ORPHAcodes (aggregation level and parent-of-preference for every ORPHAcod e within the hierarchical ORPHA nomenclature). But also preferred terms; synonyms; definition; typology; Orphanet IRL; Status; target ORPHAcod e if inactive; Classification level; Classification; ICD10 and OMIM.

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

Milestones update since last issue:

*MIs 18: Preliminary results Analysis (Fisabio) **REACHED***

*MIs 19: Extension to other regions (CIBER) **REACHED***

*MIs 20: Specifications for API V3 **REACHED***

WORKPACKAGE 5

Regarding the delivery of the 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 (milestone 5.2): results do not implicate any necessary changes to 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. During the workshop in June it was agreed that, in

order to have more countries contributing to this document in the following months and to have a fully complete deliverable 5.2 , a dedicated one day workshop should be scheduled to discuss this Topic. The workshop was held on the 24 September, 15 participants from 6 countries attended. The delivery of the D5.2 Guidelines for undiagnosed diseases codification was delayed to December 2020.

WP Leader: Regione Veneto Participants: APHP; DIMDI

Milestones update:

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 calls were held on the 04/02/2020; 21/04/2020; 16/06/2020; 22/09/2020. 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

**IF YOU HAVE ANY
QUESTIONS:**

**DO NOT HESITATE
TO CONTACT US**

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