>RD·CODE NEWS

UNDIAGNOSED PATIENTS CODING EXPERIENCES AND GUIDELINES DOCUMENT AVAILABLE

UNDIAGNOSED PATIENTS EXPERIENCES AND GUIDELIENS FOR CODING AVAILABLE

To tackle the issue of coding undiagnosed patients' a collection of existing experiences of coding of undiagnosed or suspected RD patients has been produced, a guidelines proposal has been issued as well as a consensus document on codification of suspected/undiagnosed rare diseases.

Importantly definitions of the terms used in the document are provided. These definitions are to be considered in the frame of the RD-CODE project only. They serve the purpose of making trans-national statistics possible, based on the same indicators. Thus, within the RD-CODE project the definition of an undiagnosed patient as per RD-CODE project is as follows: undiagnosed patients are patients for whom no clinically known disorder could be confirmed by a RD expert center after up-to-date state-of-the-art diagnostic workup.

The document is available **here** since the end of December 2020.

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. At the last SC meeting held on the 17 November 2020 It was decided to submit an amendment request in order to ask for a 6 months extension to the grant agreement due to the delay in delivery of the work by the teams because of the pandemic. Also, we will ask for the termination of DIMDI activities and inclusion of the new partner Bfarm which has replaced DIMDI.

Deliverables that will be delayed are listed below:

Del Rel. No	Title	Lead Beneficiary	Est. Del. Date (annex I)	Rev. Due Date
D5.2	Undiagnosed Diseases Document	APHP	30 Sep 2020	31 Dec 2020
D4.3	Orphacode Training report (MT	MFH	31 Mar 2020	30 Mar 2021
D5.3	Implementing countries Report adoption report	RV	31 Dec 2020	31 Dec 2021
D1.2	Internal newsletter	INSERM	30 Jun 2021	31 Dec 2021
D2.2	Multistakeholders Workshop reports	CIBER	30 Jun 2021	31 Dec 2021
D2.3	Dissemination report	INSERM	30 Jun 2021	31 Dec 2021
D2.5	Leaflet 2-MD2	INSERM	30 Jun 2021	31 Dec 2021
D2.6	Layman version of the executive summary of the final report_MD3	INSERM	30 Jun 2021	31 Dec 2021
D3.2	Final evaluation report	EURORDIS	30 Jun 2021	31 Dec 2021
D4.8	Outcome report (CZ)	IRIS CR	30 Jun 2021	31 Dec 2021
D4.9	Outcome Report (MT)	MFH	30 Jun 2021	31 Dec 2021
D4.10	Final report (RO)	DSP IASI	30 Jun 2021	30 Dec 2021
D4.11	Final report (ES)	CIBER	30 Jun 2021	31 Dec 2021
D 5.4	Update to coding resources and guidelines	DIMDI	30 Jun 2021	31 Dec 2021

Milestones update:
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.

The Achievements leaflet of the project is available here

News regarding the release of the new visualization facility for the Orpha Nomenclature have been published on www.rd-code.eu; as well as regarding the Training course held in Malta and the undiagnosed Patients coding experiences and guidelines.

The Spanish teams have **presented at the following meetings:**

- Virtual Congress of the Spanish Society of Epidemiology (SEE), and the Portuguese Association of Epidemiology (APE). (2020) - Oral presentation (October 2020) "Improving the identification of rare diseases: from ICD-10 to ORPHA"
- 16th World Congress on Public Health 2020 – Poster presented (October 2020)

The Czech team has attended Klinikon Meeting which had a special focus on RD (see Czech Republic update section for more info).

Furthermore, a manuscript describing the mapping process between ORPHAcodes and ICD-10-ES, and its outcomes has been submitted to the Orphanet Journal of Rare Diseases (see Spain update section for further info).

Milestones update: NTR

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) .

The final evaluation report production has been delayed because of the extension request

WP leader: EURORDIS

Milestones update: NTR

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** 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:

Training sessions for clinicians will take place online starting January 2021 via Moodle.

The **registration of cases with ORPHAcodes** continues and currently there are 500 registered cases from 3 workplaces, they are planning to involve additional cooperating hospitals in the Czech Republic.

KlasifiKon conference: this conference was held on December 2nd, 2020. It featured rare diseases as the main topic. Materials from the conference are available online (in Czech) at https://www.uzis.cz/index.php?pg=aktuality&aid=8428

ORPHAcodes are available since January 1st, 2021 for reporting to insurance companies in the Czech Republic. Hospital information systems are now in the process of adaptation (the team provides consultations to SW developers).

Malta update:

The **Development of a technical solution** is in progress and should be completed thanks to the fruitful discussions held during the **Workshop held in Malta on the 24 and 27 November.**

This event "Introduction of ORPHAcoding in Malta" was organized into 3 sessions. An introduction on the Maltese RD registry and on the additional resources available: Registry for Treatment Abroad, Registry for Congenital Anomalies, Hospital Activity Analysis, and ERNs registries. Followed by a presentation of the Spanish experience and How-to best support ERNs. Then a session on ORPHAcodes and how-to code, as well as a presentation of the French RD registry. Finally, a third session on the Veneto region experience and on the available guidelines and procedures made available by Workpackage 5 followed up by a round table to discuss the next steps vis à vis of the project and of the Maltese delivery of the tool for registration of RDs.

Audience present was: Clinical chairs, Heads of Registers from DHIR, coders, Treatment Abroad Manager, Marigold Foundation and other representatives.

Romania update:

Unfortunately, due to COVID 19 pandemics all the activity was paralyzed for several months. Registration of cases re-started.

Now training sessions are planned at distance.

They continue registering cases.

Spanish Update:

An **Updated version of the Master file in Spanish with 98% RD coverage was distributed to the regional registries in October**.

Non-solved equivalences between Orphacodes and ICD-10-ES (about 100) are being investigated together with Orphanet officer.

As regards the 3 new additional regions participating in the 2nd phase of the project (Madrid, La Rioja and Galicia): the collaboration terms have already been agreed between Madrid. FISABIO is working on similar agreements with the other two regions so that collaborations run smoothly. However progresses have been slowed down because of the COVID19 pandemic.

In addition, **the Navarre region is advancing towards the implementation of ORPHAcodes** in order to catch up with the rest of the registries initially enrolled in the project.

A manuscript describing the mapping process between ORPHAcodes and ICD-10-ES, and its outcomes has been submitted to the Orphanet Journal of Rare Diseases and is currently "under review". A preprint version of the publication is accessible through https://www.researchsquare.com/article/rs-122664/v1

Orphanet update:

Web-based applications development by Orphanet was foreseen during this project to facilitate the informatic access to nomenclature data and allow flexible implementation into the various IT systems in use in the different countries and/or settings.

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 the official release and upgrade was made in June 2020 (Facility for ORPHA nomenclature correspondence, Deliverable 4.5). The **Orphanet Data visualisation tool** https://dataviz.orphacode.org/ is

delivered in December 2020. All interrogations exploit the above-mentioned Orphanet API also released within this project, showing at the same time the nice exploitation potential of the Orphanet API. This tool allows to search for the clinical entities (groups of disorders, disorders, sub-types) that are present in the nomenclature pack (here) in all the API languages. Find out more info about the tool here.

Also, the Frequently Asked Question Page has been updated, do not hesitate to consult it!

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

Milestones update since last issue:NTR

WORKPACKAGE 5

In addition to the important delivery mentioned in the Editorial of this issue "UNDIAGNOSED PATIENTS EXPERIENCES AND GUIDELIENS FOR CODING", the milestone 5.2 First Report of necessary changes to the Master file was also delivered together with a new updated version of the Master File (all info available here).

WP Leader: Regione Veneto Participants: APHP; DIMDI

Milestones update since last issue :

*Mls 5.2 First Report of necessary changes to the Master file according to the results of the first workshop **REACHED***

*Mls 5.5 Undiagnosed Diseases Document **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 a 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; 17/11/2020. Read the full reports here.

Next sessions will be held: 19/01/2021; 16/03/2021; 04/05/2021 and 01/06/2021.

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
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