



***RD-CODE Mid-Term Evaluation
Report***
REPORT

30 March 2020

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

The content of the [insert type of document] represents the views of the authors only and is their sole responsibility; it cannot 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.

TABLE OF CONTENTS

EXECUTIVE SUMMARY OF PROJECT ACHIEVEMENTS	3
<i>OBJECTIVE 1. ESTABLISH AN EFFECTIVE AND EFFICIENT GOVERNANCE IN THE PROJECT</i>	<i>3</i>
<i>OBJECTIVE 2. TO DISSEMINATE PROJECT INFORMATION AND OUTPUTS TO INCREASE AWARENESS AND UNDERSTANDING</i>	<i>3</i>
<i>OBJECTIVE 3. MEASURE THE KPI OF THE PROJECT AND INDICATORS PER SPECIFIC OBJECTIVE AND ADMINISTER SATISFACTION SURVEYS</i>	<i>3</i>
<i>OBJECTIVE 4. IMPLEMENTATION IN FOUR COUNTRIES</i>	<i>3</i>
<i>OBJECTIVE 4.5 CENTRALISE AND DEVELOP INFORMATION ON THE ORPHANET NOMENCLATURE FOR CODIFICATION PURPOSES</i>	<i>5</i>
<i>OBJECTIVE 5. PROVIDE BACKGROUND ANALYSIS OF THE “CODING ENVIRONMENT” IN IMPLEMENTING COUNTRIES</i>	<i>5</i>
ANNEX 1. INDICATORS PER SPECIFIC OBJECTIVE (STATUS MARCH 2020)	6

EXECUTIVE SUMMARY OF PROJECT ACHIEVEMENTS

Overall, the RD Code project is progressing at a good pace and the partners are delivering good quality results and on time. We are confident that at the end of the project we would have been able to achieve our project goals.

Following CHAFEA recommendations regarding the Covid19- Coronavirus outbreak the project coordinating team has decided to postpone:

- The RDCODE Training workshop to be organised in Malta on the 30 March
- RD-CODE Multistakeholder Workshop planned originally to 29th and 30th April.

OBJECTIVE 1. ESTABLISH AN EFFECTIVE AND EFFICIENT GOVERNANCE IN THE PROJECT

The Steering Committee calls are organised on a regular basis; reports have been submitted in due time and project partners are satisfied with Orphanet's management and support.

OBJECTIVE 2. TO DISSEMINATE PROJECT INFORMATION AND OUTPUTS TO INCREASE AWARENESS AND UNDERSTANDING

Dissemination activities are regularly reported in the Steering Committee calls. Three articles on the project have been published in Orphanews and several abstracts have been submitted for the ECRD Congress in 2020. The website was launched in March 2019 and the majority of the project partners consider that the dissemination materials help to understand the project goals.

OBJECTIVE 3. MEASURE THE KPI OF THE PROJECT AND INDICATORS PER SPECIFIC OBJECTIVE AND ADMINISTER SATISFACTION SURVEYS

Evaluation aspects are discussed in each Steering Committee call. Three surveys have been administered from the beginning of the project until March 2020:

- Participants satisfaction survey of Kick-off meeting (Milestone 3.1)
- Mid-term partners satisfaction survey (Milestone 3.2)
- Survey on toolset (INSERM)

OBJECTIVE 4. IMPLEMENTATION IN FOUR COUNTRIES

Objectives 4.1-4.4. Implementation in the four countries (Czech Republic, Malta, Romania and Spain)

Czech Republic. To enforce the use of ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation

manual of the Master file" in major rare diseases centers reporting to the National Register of Congenital Malformations.

Two hospitals have accepted to participate in the pilot. Number of hospitals accepting to participate in the piloting. At the end of 2019, 126 new cases from these hospitals had been coded using ORPHAcodes, well above the target of the project (50). The National Register of Congenital Malformations has managed to implement ORPHAcodes in its data model. By the end of 2019, the two hospitals were reporting their data to the registry using ORPHAcodes.

A training session for 17 clinicians has been organised in March and two others are planned. A satisfaction survey and knowledge test will be administered after the 3 workshops.

Malta. To introduce ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" in the daily practice of registries including rare diseases.

Malta has introduced automatic Orphacoding into the data model of three existing registries: congenital malformations registry, cancer registry and the Treatment abroad data. More than 200 cases were reported using ORPHAcodes in 2019.

Romania. To introduce ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" in the daily practice of a major centre dealing with rare diseases diagnosis

Romania has achieved the target of coding 30 rare diseases using ORPHAcodes. The number of cases reported per year using ORPHAcodes is 200 and the health authorities have used the information to inform the decision on the allocation of funds for 2 National Health Programmes for the diagnosis and treatment of rare diseases. They have organised one training session with the participation of 12 clinicians.

Spain. To pilot the implementation of ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" at the rare diseases registries in selected regions in Spain.

Six Spanish regions have made adaptations in their processes towards the implementation of ORPHAcodes (Comunitat Valenciana, País Vasco, Cataluña, Castilla y León, Murcia y Navarra) and five of them have actually implemented (Comunitat Valenciana, País Vasco, Cataluña, Castilla y León, Murcia).

The target regarding the percentage of cases coded with an ORPHAcodes (75%) has been reached with 100% of the cases communicated to the national registry with an

Orphacode; and 90% of the ORPHAcodes from the Master file with an ICD-10-ES equivalent.

OBJECTIVE 4.5 CENTRALISE AND DEVELOP INFORMATION ON THE ORPHANET NOMENCLATURE FOR CODIFICATION PURPOSES

Orphanet has organised 12 codification project internal meetings. The mid-term partners satisfaction survey revealed that the representatives of two countries participating in the project are satisfied with the tools and documents provided by Orphanet (Malta and Romania representatives did not respond to the survey).

OBJECTIVE 5. PROVIDE BACKGROUND ANALYSIS OF THE “CODING ENVIRONMENT” IN IMPLEMENTING COUNTRIES

A report with the background analysis of the “coding environment” in implementing countries was published in June 2019. The report covers 3 out of 4 implementing countries answered the questionnaire (Spain, Czech Republic and Malta, Romania did not answer). The background Report accessible in the project’s website ([here](#))

Detailed information on the project’s KPI per objective is provided in Annex 1.



ANNEX 1. INDICATORS PER SPECIFIC OBJECTIVE (STATUS MARCH 2020)

		Status March 2020	Comments
Specific Objective Number	1.1 to 1.4		
Specific Objective	1.1. Establish an effective and efficient governance in the project 1.2. Ensure smooth communication and information exchange amongst project participants & stakeholders 1.3 Provide day to day administrative support to the partners 1.4. Ensure all communication with the CHAFEA and the DG SANTE: including timely presentation of all deliverables, technical and financial reports. 1.5. Ensure risk management		
Process Indicator(s)	Target		
Steering committee meetings	8	6	On track
Output Indicator(s)	Target		
Report availability	8	6	On track All reports are made available to partners at the latest one week after the conference call
Outcome/Impact Indicator(s)	Target		
Satisfaction level of partners (coordination activities)	>90%	100% of project partners agree or strongly agree that the Coordination team is effectively supporting the partners to deliver on a timely manner	On track (Ref. Mid-term Partners satisfaction survey).
Specific Objective Number	2		
Specific Objective	To disseminate project information and outputs to increase awareness and understanding		
Process Indicator(s)	Target		
Dissemination item included in the agenda of each steering committee meeting	8	6	On track A dedicated space for WP2 updates and



		Status March 2020	Comments
			discussion is included in each SC agenda
Output Indicator(s)	Target		
Article in Orphanews	4	3	On track Implementing ORPHA codes in European countries: the RDCODE project approved
Results presentation in congress	1		On track Several abstracts have been submitted to the ECRD 2020
Webpage availability	1	Achieved March 2019	www.rd-code.eu
Outcome/Impact Indicator(s)	Target		
Downloads of documents increase Y1-Y2-Y3	increase (baseline Y1)	Downloads form Nov 2019 to 16 March 2020 Documents available on Rd-CODE were downloaded approx. 200 times Nomenclature pack available on orphadata was downloaded more than 1500 times in all languages (more than 700 in English only)	To be compared to April 2020-June 2021 Downloads
Satisfaction on available documentation Y1-Y3	increase (baseline Y1)	80% (This value will represent the baseline year 1)	Result of the Mid-term partners satisfaction survey (Milestone 3.2) To be compared with the results from final survey
Specific Objective Number	3.1 and 3.2		
Specific Objective	3.1. Measure the KPI of the project and Indicators per specific objective 3.2. Elaborate Satisfaction surveys for project partners (regarding coordination and dissemination activities),		



		Status March 2020	Comments
Process Indicator(s)	Target		
Evaluation item included in the agenda of each steering committee meeting	8	6	On track A dedicated space to discuss WP3 is foreseen in every SC meeting
Output Indicator(s)	Target		
Reports production	2	1	On track
Surveys production	4	3	On track Participants satisfaction survey of Kick-off meeting 1 (Milestone 3.1) Mid-term partners satisfaction survey (Milestone 3.2) Survey on toolset (INSERM)
Outcome/Impact Indicator(s)	Target		
Improvement of KPI from mid-term to final term Satisfaction of end users (regarding the project overall) increased from mid-term to final term	Increase	NA	Final partners satisfaction survey scheduled for M29
Specific Objective Number	4.1.1 (CZECH REPUBLIC)		
Specific Objective	To enforce the use of ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" in major rare diseases centers reporting to the National Register of Congenital Malformations.		
Process Indicator(s)	Target		
Number of hospitals accepting to participate in the piloting	2	2	Achieved
Output Indicator(s)	Target		
Number of newly reported cases from these hospitals coded using ORPHAcodes per year	50	126	The target was already outreached at the end of 2019



		Status March 2020	Comments
Outcome/Impact Indicator(s)		Target	
National Register of Congenital Malformations manages to implement ORPHAcodes in its recording system		1	NRCM/NRVV has Orpha codes in the data model
Specific Objective Number	4.1.2 (CZECH REPUBLIC)		
Specific Objective	To train coding clinical experts in the use of ORPHAcodes focused on hospitals participating to ERNs.		
Process Indicator(s)		Target	
Number of training sessions performed		3	1 At the end of March 2020, 2 other are already planned
Output Indicator(s)		Target	
Number of clinicians trained		15	19 The target was already outreached with the first training session organised
Outcome/Impact Indicator(s)		Target	
Satisfaction and knowledge of trainees		90%	NA Satisfaction and knowledge validation will be performed after 3 training sessions
Specific Objective Number	4.1.3 (CZECH REPUBLIC)		
Specific Objective	To increase the number of hospitals reporting rare diseases cases into the National Register of Congenital Malformations using ORPHAcodes.		
Process Indicator(s)		Target	
Number of hospitals reporting to the register using ORPHAcodes per year		7	2 On track
Output Indicator(s)		Target	
Number of cases reported using ORPHAcodes by these hospitals per year		200	126 The number of cases will be evaluated after two completed years
Outcome/Impact Indicator(s)		Target	
A comparison between the period 2016-2017 and the Project implementation period will render		at least a 50% increase in the number of cases identified	NA The increase will be evaluated after two completed years



		Status March 2020	Comments
significant results as regards overall RD cases identified, accuracy of reporting and variety of diagnoses reported. A report will be issued.			
Specific Objective Number	4.2 (MALTA)		
Specific Objective	To introduce ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" in the daily practice of registries including rare diseases.		
Process Indicator(s)	Target		
To introduce automatic Orphacoding into the data model of three existing registries: congenital malformations registry, cancer registry and the Treatment abroad data	3	Achieved for the 3 registries	They will be uploaded once the IT system overhaul is completed.
Output Indicator(s)	Target		
Number of cases reported using ORPHAcodes per year	200	Achieved, more than 200 cases reported in year 1	
Outcome/Impact Indicator(s)	Target		
A comparison between the period 2016-2017 and the Project implementation period will render significant results as regards overall RD cases identified, accuracy of reporting and variety of diagnoses reported with accurate prevalence data per RD registered in the general Maltese population reported.	50% increase	Work in progress	
Specific Objective Number	4.3.1 (ROMANIA)		
Specific Objective	To introduce ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" in the daily practice of a major centre dealing with rare diseases diagnosis.		
Process Indicator(s)	Target		



		Status March 2020	Comments
Number of rare diseases for which the ORPHAcodes has been implemented by M12	30	30	Achieved
Output Indicator(s)	Target		
Number of cases reported using ORPHAcodes per year	200	200	Achieved
Outcome/Impact Indicator(s)	Target		
Health authorities will have correct numbers that will enable them to allocate adequate funds for National Health Programmes (for diagnosis and therapy of rare diseases)	2 Health Programmes (for rare diseases and for the prevention and diagnosis of congenital defects and genetic disorders)	2	Achieved
Specific Objective Number	4.3.2 (ROMANIA)		
Specific Objective	To train coding clinical experts in the use of ORPHAcodes focused on hospitals included in the ERNs.		
Process Indicator(s)	Target		
Number of training sessions	4	1	More training sessions will be organised during the last period of the project to meet the objective
Output Indicator(s)	Target		
Number of clinicians trained	30	12	See above
Outcome/Impact Indicator(s)	Target		
Clinicians involved in rare diseases diagnosis will know better how to report their results and will do it in an unitary way. Based on Y1 results other centres will decide to introduce ORPHAcodes.	3 additional centres	100% of the clinicians surveyed agree or strongly agree that the Romanian team is supporting the other rare diseases centres to use ORPHAcodes	
Specific Objective Number	4.3.3 (ROMANIA)		
Specific Objective	To introduce ORPHAcodes in other centres involved in ERNs. M16-30		
Process Indicator(s)	Target		
Number centres that will join the Orphacoding	3	NA	To be reported in M30



		Status March 2020	Comments
Output Indicator(s)	Target		
Number of patients with rare diseases reported using ORPHAcodes	100	NA	To be reported in M30
Outcome/Impact Indicator(s)	Target		
Other National Health Programmes will benefit of the results concerning the frequency of rare diseases. Based on the proofs provided, Health authorities will be able to decide if they want to generalize Orphacoding for Romania	2 other National Health Programmes	NA	To be reported in M30
Specific Objective Number	4.4.1 (SPAIN)		
Specific Objective	To pilot the implementation of ORPHAcodes according to the "Standard procedure and guide for the coding with ORPHAcodes" and the "Specification and implementation manual of the Master file" at the rare diseases registries in selected regions in Spain.		
Process Indicator(s)	Target		
Number of regions that have made adaptations in their processes towards the implementation of ORPHAcodes at M12	6	6	Achieved Comunitat Valenciana, País Vasco, Cataluña, Castilla y León, Murcia y Navarra
Output Indicator(s)	Target		
Nº of regions having implemented ORPHAcodes by M12	4	5	Achieved Comunitat Valenciana, País Vasco, Cataluña, Castilla y León, Murcia
Outcome/Impact Indicator(s)	Target		
Percentage of cases coded with an Orphacode at M12	75%	100% of the cases communicated to the national registry with an ORPHAcodes; and 90% of the ORPHAcodes from the Master file with an ICD-10-ES equivalent	Achieved
Specific Objective Number	4.4.2 (SPAIN)		



		Status March 2020	Comments
Specific Objective	Following the pilot evaluation phase, to disseminate the results in order to involve other regions in the Project.		
Process Indicator(s)	Target		
Nº of new regions working towards the implementation of ORPHAcodes after M15	2	NA	Applicable after M15
Output Indicator(s)	Target		
Nº of new regions having implemented ORPHAcodes by M27	2	NA	To be reported after M27
Outcome/Impact Indicator(s)	Target		
Percentage of cases coded with an Orphacode at M27	75%	NA	To be reported after M27
Specific Objective Number	4.5 (Orphanet)		
Specific Objective	Centralise and develop information on the Orphanet nomenclature for codification purposes		
Process Indicator(s)	Target		
Codification project Orphanet internal meetings	12	12	Achieved
Output Indicator(s)	Target		
Number of new documents available per year	1	1	Achieved FAQ, Xml content description, RD-code website (http://www.rd-code.eu/introduction/)
Outcome/Impact Indicator(s)	Target		
Satisfaction on access to the information and quality per annum. (Survey of the 4 implementing countries satisfaction before/after the release of the webpage and documents)	Increase in the satisfaction level at the end of project (M30) compared to baseline (M3).	Ongoing The survey has a goal to evaluate the needs of each country member and understand their expectations in terms of providing resources. In the end, we will base on this report to develop user-friendly technical resources, and to ensure an easier	The preliminary results of the survey, show that more than half of participants are satisfied with the codification documents made available online. So far, requests for other formats and some additional datas are notified. We are waiting for more responses in order to take into consideration the users needs for the new version of these files.



		Status March 2020	Comments
		and more accurate rare disease coding.	
Survey of the 4 implementing countries satisfaction before/after the release of the webpage and documents		Ongoing	On track Mid-term partners satisfaction survey (Milestone 3.2) Representatives of two (Spain and Czech Republic) countries responded to the mid-term partners satisfaction survey Romania and Malta did not respond.
Specific Objective Number 5.	5.1		
Specific Objective	Provide background analysis of the “coding environment”_in implementing countries		
Process Indicator(s)	Target		
Questionnaire definition exploring background analysis of the “coding environment” in implementing countries.	Analysis of information coming from all the implementing countries	Achieved March 2019	3 out of 4 implementing countries answered the questionnaire (Spain, Czech Republic and Malta). Romania did not answer, despite several reminders. 2 additional countries (observers) answered: Austria and Norway.
Output Indicator(s)	Target		
Document on existing “coding environments” regarding RD coding in implementing countries	1	Achieved June 2019	The document describes the coding environment of the countries which completed the questionnaire before the agreed deadline: Spain, Czech Republic, Malta
Outcome/Impact Indicator(s)	Target		
Results accessible through the project website	Results dissemination at WS: 1 multi-stakeholder workshop 1 international conference	Background Report accessible in the project website	For each country, an overview of the following themes is provided: national RD



		Status March 2020	Comments
	1 national event/implementing country		policy framework, coding framework, RD registration, Centres of Expertise/ERNs, undiagnosed RD patients. We were supposed to present results at the Multistakeholder WS, which was planned in Prague, but has been postponed due to Covid19.
Specific Objective Number	5.2		
Specific Objective	Analysis of the “coding environment” in implementing countries following ORPHAcodes adoption		
Process Indicator(s)	Target		
Definition of a questionnaire exploring changes in the “coding environments” in implementing countries following ORPHAcodes adoption.	Analysis of information coming from all the implementing countries	NA	Due for 31/12/2020
Output Indicator(s)	Target		
Document on lessons learned from implementing countries following ORPHAcodes adoption.	1	NA	Due for 31/12/2020
Outcome/Impact Indicator(s)	Target		
Results accessible through the project website.	Results dissemination at WS: 1 multi-stakeholder workshop; 1 international conference; 1 national event/implementing country	NA	Results will be disseminated after 31/12/2020
Specific Objective Number	5.3		



		Status March 2020	Comments
Specific Objective	Refinement of the "Standard procedure and guide for the coding with ORPHAcodes"(17) and the "Specification and implementation manual of the Master file" (19)		
Process Indicator(s)	Target		
Analysis of need for change of the "Standard procedure and guide for the coding with ORPHAcodes"[17] and the "Specification and implementation manual of the Master file" [19] was performed following Objective 5.1 and 5.2 as well as Workshops in WP 2	Analysis of information specific on the two documents	NA	Due for 30/09/2020
Output Indicator(s)	Target		
Document on lessons learned and need for updating	1	NA	Due for 30/06/2021
Outcome/Impact Indicator(s)	Target		
Results accessible through the project website	Results dissemination at WS: 1 multi-stakeholder workshop; 1 international conference;	NA	Results will be disseminated after 30/06/2020