Deliverable 4.1
OD4RD State of play survey
31.03.2022

This survey represents deliverable 4.1 of the OD4RD project, which has received funding from the European Union. This survey aims at analyzing the level of implementation of rare disease patient coding across member states. The survey was conducted through an online questionnaire sent to the members of the Work package 4 contact list. This document presents the results collected between March, 10 and March, 23 2022. It has been produced by the leaders of the OD4RD - Work Package 4. The OD4RD project has been launched in January 2022 for a 12 months period.

More information on the activities of the OD4RD can be found at www.OD4RD.eu

Disclaimer:

The findings and conclusions in this report are those of the contributors, who are responsible for the contents; the findings and conclusions do not necessarily represent the views of the European Commission or national health authorities in Europe. Therefore, no statement in this report should be construed as an official position of the European Commission or a national health authority.

Content summary

Context

The field of rare diseases (RD) is one in which the value of trans-national data collection, sharing and exploitation for evidence-based policy, healthcare and research has been demonstrated and targeted by several European initiatives. An estimated 30 M European citizen's population suffer from a rare disease and therefore possesses great significance in its entirety. More than 6,000 different rare diseases are actually known. Since only few patients suffer from a certain rare disease the critical amount of data necessary to improve knowledge and action-taking can only be achieved by cumulative data collection at different countries in a standardised way. Rare diseases are poorly represented in existing medical terminologies in use, and there is no terminology specific to rare diseases, except the domain-specific Orphanet nomenclature of rare diseases (ORPHAcodes). This nomenclature and classification system was developed and maintained thanks to European support after the recognition as a priority, in the Council Recommendation of 8 June 2009 on an action in the field of rare diseases, of the improvement in codification of rare diseases. This harmonised and standardised codification is instrumental to implement the 2011 Cross Border Health-care Directive as far as RD are concerned. Since then, a comprehensive nomenclature system has been developed in line with the continuous evolution of knowledge in the constantly evolving rare disease field. The nomenclature is aligned with several non-RD specific terminology resources allowing for semantic interoperability in a context of heterogeneity of coding systems used in different countries and contexts. The OD4RD project will build on the specific Orphanet expertise, and on its organisation as a long-lasting, well-established network, to fulfil the following general objectives:

1. To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, by the maintenance and the support to the implementation of the Orphanet nomenclature of RD.

2. To contribute to the harmonisation of data collection amongst settings (health records, registries) and amongst countries, by the dissemination of good coding practices at the data source level.

3. To contribute at supporting evidence-based decision-making in the frame of the European strategy around ERNs, by supporting the exploitation of reference corpus of data and information on RD.

Objective

The objective of Work package 4 is to ensure support for the local implementation of ORPHAcodes in national HCPs hosting ERNs and national HCPs linked to ERNs by establishing Orphanet national nomenclature support hubs.

The aim of the questionnaire is to assess the situation and needs of end users in regards to ORPHAcodes implementation in the WP4 participating countries.

Methodology

BfArM and INSERM teams worked together to identify key questions that needed to be answered regarding the coding systems in participating countries and discussed them with the Excom. The questions were divided into four sections: existing structures and plans, helpdesks and support, training and overall implementation process in WP4. The questions could be submitted partly in a multiple choice procedure and partly in open comment fields.

The survey was created on an online platform (Lime Survey). The link to this online survey was sent to all participating countries of WP4. Results were extracted from the online tool and analysed by the leading team of WP4.

Participating countries:

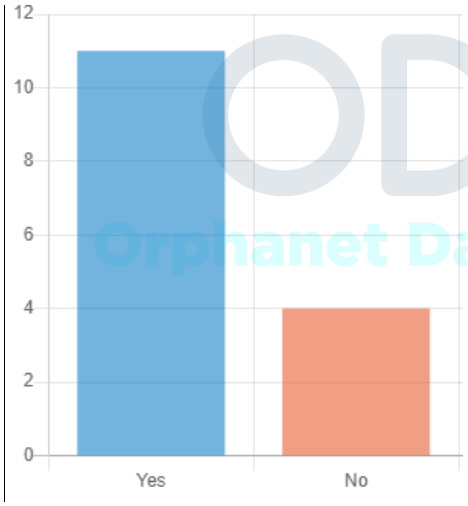
Austria, Belgium, Czech Republic, Finland, France, Germany, Italy, Netherlands, Norway, Poland, Portugal, Slovenia, Spain, Sweden and Switzerland*

*Switzerland is participating in WP4 as observer

Results

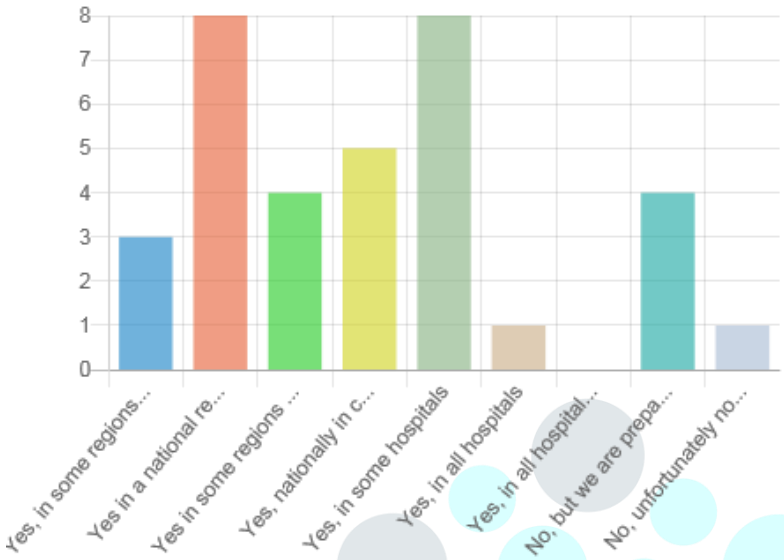
Section A: Section on existing structures and plans

Does your country have a regional or national plan/strategy to code RDs?



Most of the participating countries (11, or 73%) have a regional or national plan or strategy to code rare diseases. In only four of the participating countries such a plan is not yet in place.

Does your country use ORPHAcodes at MS level and/or regional level to produce data or statistics for RD?



Explanation:

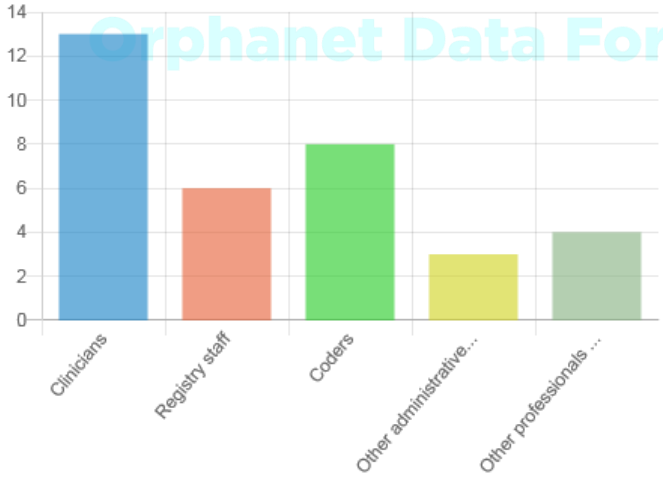
1st: Yes, in some regions in registries
2nd: Yes, in a national registry
3rd: Yes, in some regions in centers of expertise for RDs
4th: Yes, nationally in centers of expertise for RDs
5th: Yes, in some hospitals
6th: Yes, in all hospitals
7th: Yes, in all hospitals and all outpatient settings
8th: No, but we are preparing for implementation
9 th: No, unfortunately not yet

In only one of the countries ORPHAcodes are used to produce data or statistics for rare diseases in all hospitals. In 5 countries ORPHAcodes are not yet used for that purpose, but 4 of them are preparing for implementation. In most cases ORPHAcodes are used in registries, either national registries (8) and/or regional registries (3). In most countries ORPHAcodes are used in centers of expertise for RD, either nationally (5) or regional (4).

If yes, when did the recording of RD patients using ORPHAcodes start in your country?

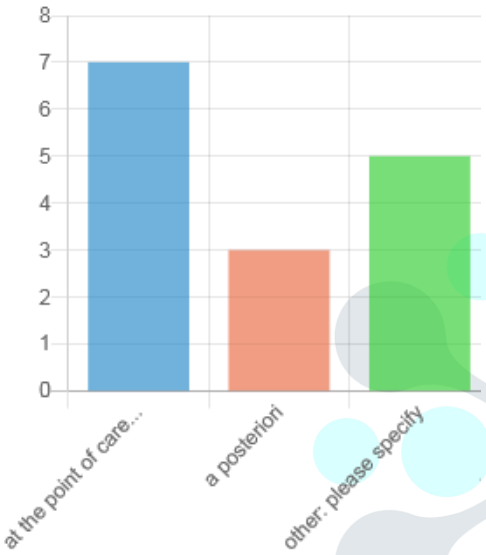
8 of the countries that are using ORPHAcodes already submitted information about the starting year. Most of them started in 2020 (3), one each in 2012, 2013, 2016, 2018 and 2021. That means that at least half of the countries using ORPHAcodes are still collecting first experiences, only a few countries can already look back on a somewhat longer experience.

If yes, who is involved in the codification process?



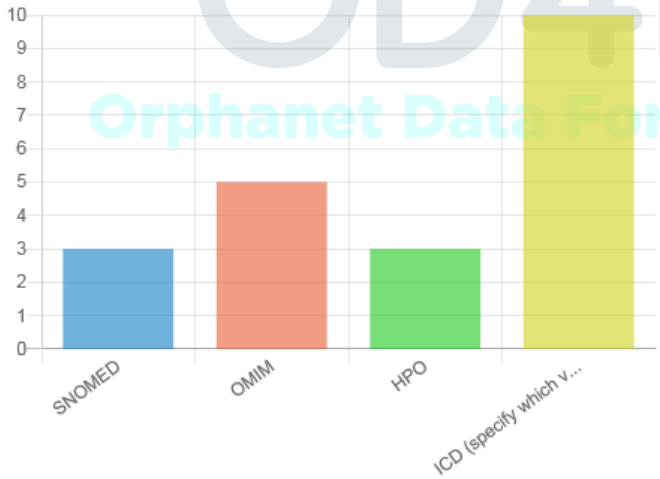
In 86% of the countries clinicians are involved in the codification process. In 40% of the countries registry staff takes care of the codification. 53% of the countries do their coding work by coders and in 46% of the countries it is done by other administrative staff (20%) or other professionals (26%). Additionally in some countries the nursing staff, clinical laboratory geneticists or clinical research assistants are involved in the codification process.

Please describe at which point of the patient pathway ORPHAcodes are used in your implementation experience:



In 7 countries ORPHAcodes are used at the point of care and in 3 countries they are used a posteriori. Of the 5 countries that voted “other”, in 3 of them ORPHAcodes are used at the point of care and a posteriori. In one country ORPHAcodes are not used yet.

Are ORPHAcodes used in a stand-alone modality or are they used together with other nomenclatures/terminologies/coding systems for RD coding?



In most of the participating countries (10) ORPHAcodes are used together with ICD for RD coding. ICD-10-WHO or national modification are used in 9 countries, 2 participants stated the usage of both ICD-10 and ICD-9, 1 participant the usage of both ICD-10 and ICD-11. In 3 countries SNOMED is used in addition to ORPHAcodes, OMIM as an additional terminology is used in 5 countries and HPO in 3 countries.

If yes, do you maintain an alignment between ORPHAcodes and these systems?

Seven participants confirmed an alignment between ORPHAcodes and ICD-10 and/or SNOMED, OMIM and HPO.

Could you briefly describe the process put in place?

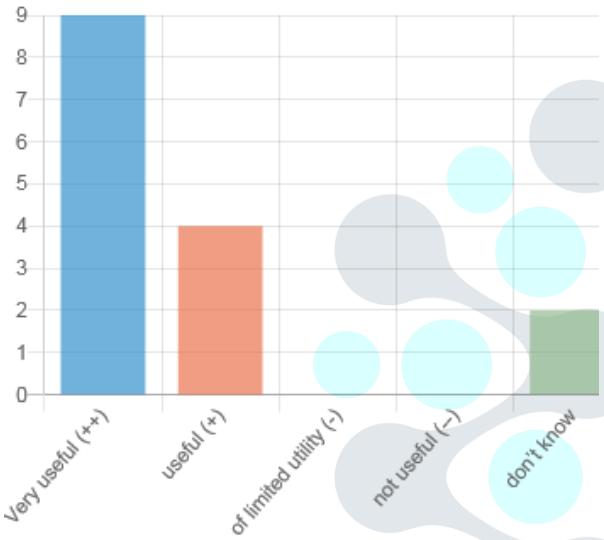
The alignment between national modification of ICD-10 and ORPHAcodes is maintained by 3 countries, whereby this was partly developed within the framework of the RD-CODE project. An alignment of SNOMED and ORPHAcodes is maintained in one country. A manual cross referencing between ORPHAcodes and different coding systems is used in 4 countries, a preparation in form of a piloting project will be started in one country.

Section B: Section on Helpdesks and support:

In case of problems in the use of the ORPHAcodes during the codification process, is there a process in place to manage questions emerging from users? i.e. Helpdesk?

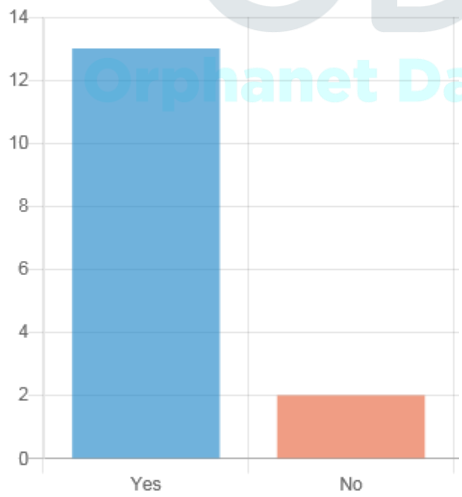
In 7 countries there is a process in place to manage questions emerging from ORPHAcode users. In 4 cases the Orphanet national team will be available for answering questions, in 3 countries these are members of the national RD registries.

How would you rank the usefulness of setting up a "local" helpdesk managing requests from users during the implementation process?



The overwhelming number of participants (13) consider the setting up of a local helpdesk as very useful or useful, only two are actually uncertain about the benefit.

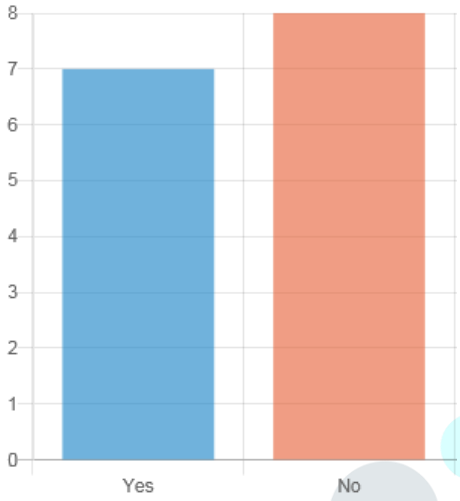
Do you think a national community of practice could be useful to share problems and best practices regarding the ORPHAcodes' use to code RD patients?



Again, an overwhelming number of participants see a national community of practice as helpful, only two participants answered no to this question.

Section C: Section on Training

Did you organize training sessions for users involved in the codification process?



7 countries already have organized training sessions for RD coders. In 8 countries this has not yet been offered.

Did you use already existing resources or have you created new ones?

From the 7 countries that have already organized training sessions for coders, 4 of them used existing training material from EJP-RD project (2), from EJP-RD and RD-CODE projects (1, in addition to self-created material) or from different Orphanet resources (1). 3 countries mainly used self-created resources.

Have the training sessions been organized on purpose or were they embedded in other training events?

In all 7 countries that already have organized training sessions for RD coders, the training sessions have been organized on purpose.

Did you develop training material?

5 countries already developed training material, mostly presentations on coding with ORPHAcodes (4), one as part of the EJP-RD project. One country is preparing material for upcoming trainings.

Based on your experience, what do you find particularly useful to be proposed in training sessions?

Almost all comments submitted recommend the use of practical examples for ORPHAcoding of various clinical diagnoses. Also the need of clear and uniform coding with ORPHAcodes and their benefit for improvement of diagnostics and therapy of RD should be explained. The training sessions should allow to understand the multi-hierarchical Orphanet nomenclature more in-depth. Furthermore the training sessions should be of limited length and should be tailored to groups with different levels of prior knowledge. Also “take home leaflets” with concise information how to approach ORPHAcoding has been suggested.

Section D: Section on overall implementation process in WP4

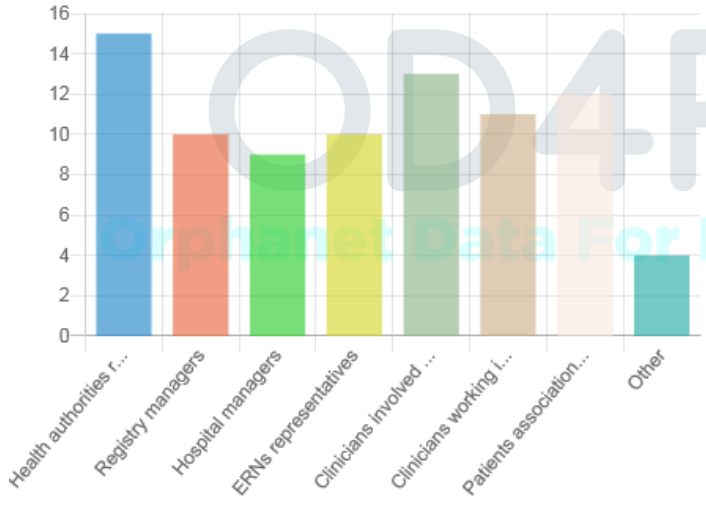
According to your experience, which were the main barriers encountered in the implementation process that we should address in WP4?

Considered as a main barrier is the lack of legislative framework to use ORPHAcodes and the lack of incentives for using them. Also the low motivation for implementation of ORPHAcodes into hospital information systems (high expenditure of time and personnel, lack of funding for H.I.S. software companies) is mentioned. Another barrier is that the high benefit of ORPHAcoding of RD is not yet clear to all clinicians, who will find it difficult to add new routines to their already very busy schedule. The shortage of medical personnel in a lot of countries requires a simple and not time consuming solution for coding with ORPHAcodes.

According to your experience, which are/could be the main drivers for a successful ORPHAcodes implementation in your country that we should address in WP4?

As main driver has been cited the need of support by health authorities like ministry of health and health insurance companies to force the usage of ORPHAcodes. Demonstration of resulting benefits for ERNs and university hospitals (producing indicators) might help. Highlighting the usefulness of using ORPHAcodes for diagnostics and therapy of RD patients would be very important. Easy to understand and not time consuming training activities should disseminate these benefits. It also might be necessary to have a technical solution that facilitates ORPHAcodes automatic data capture to RD registries. Furthermore a European guidance for implementation of ORPHAcodes into electronic health records has been recommended.

How would you rank the need of involvement of the following stakeholders for a successful ORPHAcodes implementation in your country?



Explanation:

1st: Health authorities representatives
2nd: Registry managers
3rd: Hospital managers
4th: ERNs representatives
5th: Clinicians involved in HCP members of ERNs
6th: Clinicians working in other RD centers
7th: Patients associations / ePAGS
8th: Other

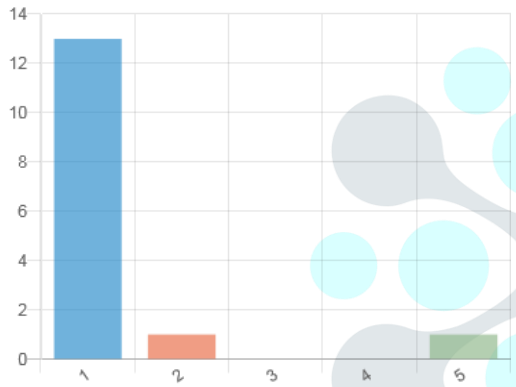
Please specify if you think other stakeholders need to be involved:

The following additional stakeholders were listed: national authorities responsible for information systems in the national health system, authorities that fund clinical research in the field of rare diseases, developers and suppliers of health information systems, health insurance companies, learned societies, national digitalisation agency setting the national interoperability framework and eventually distributing the nomenclature.

The rated level of the need of involvement of the main stakeholders for a successful ORPHAcodes implementation is shown in the following illustrations:

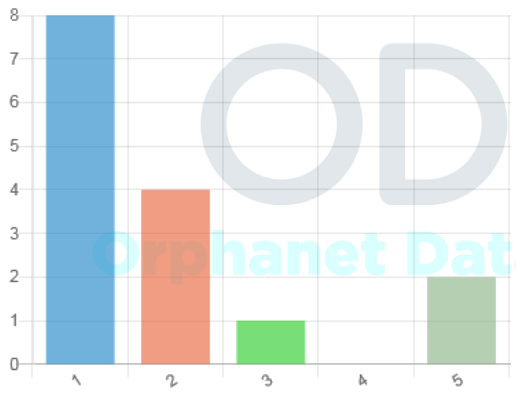
Health authorities representatives:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don't know



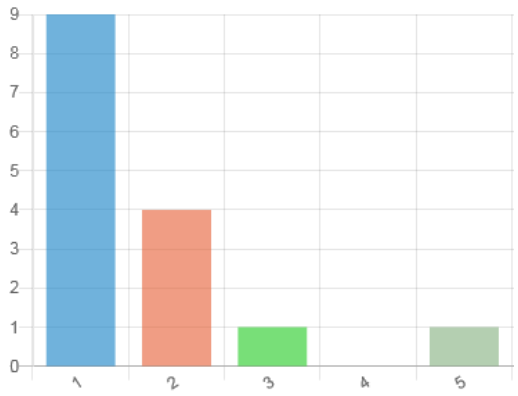
Registry managers:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don't know



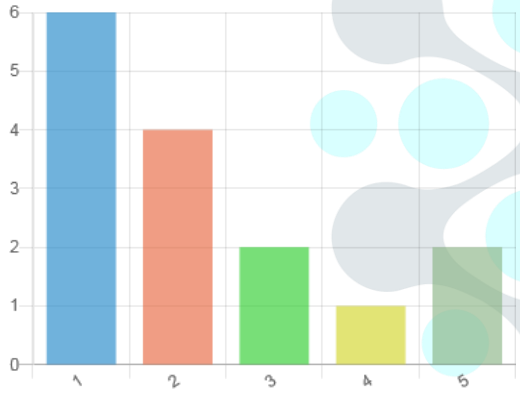
Hospital managers:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don't know



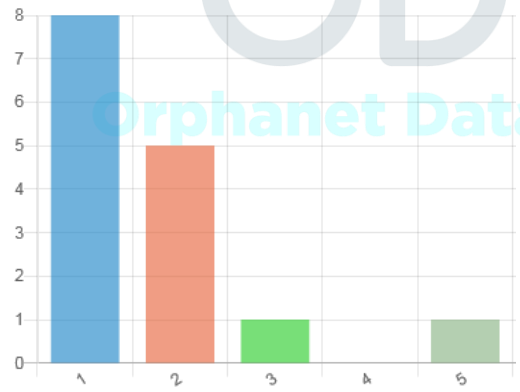
ERNs representatives:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don't know



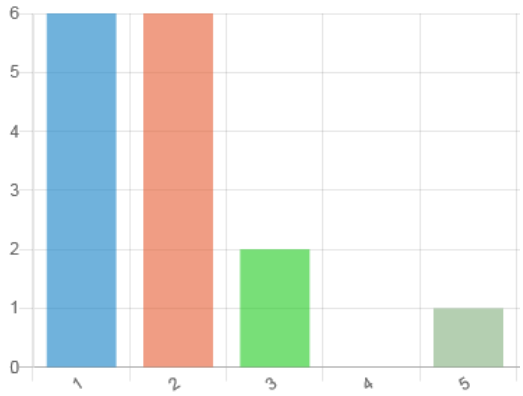
Clinicians involved in HCP members of ERNs:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don´t know



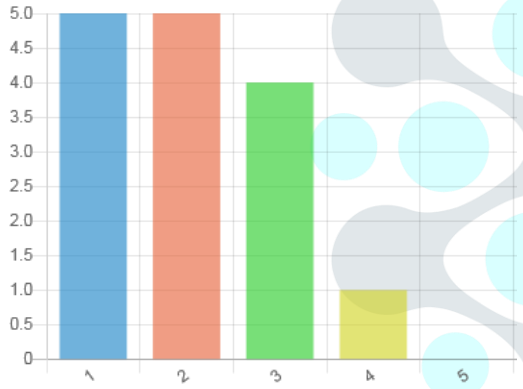
Clinicians working in other RD Centers:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don't know



Patients associations /ePAGS:

1: Crucial (++) 2: important (+) 3: of limited importance (-) 4: not important (--) 5: don't know



The involvement of health authorities representatives is considered most important, followed by clinicians involved in HCP members of ERNs, hospital managers, registry managers, cinicians working in other RD centers, ERN representatives and patient associations.

Are you already in contact with the following institutions in your country?

Contacts were indicated as follows:

Health authorities representatives: 15

Clinicians involved in HCP members of ERNs: 13

Patients associations /ePAGS: 12

Clinicians working in other RD Centers: 11

Registry managers: 10

Hospital managers: 10

ERNs representatives: 10

Other: Health information system developers (2), health insurance companies (1)

What are your key expectations for WP4 to achieve together in this project year?

A lot of different expectations have been listed:

Increase awareness of the relevance of ORPHAcodes and push forward the implementation in national coding systems or at regional level and to electronic patient records. Establish national plans for the introduction or further development of coding with ORPHAcodes as a basis for the work in the coming years.

Having a strategy reaching the key stakeholders in each participating country and actually having started conversations with them. Increase access to information related to rare diseases and facilitate the transfer of knowledge to clinical practice. Establish a relationship with ERN representatives and identify a methodology for the implementation of the European registry of rare diseases. Be able to convince the professionals to participate in this project and to set up a system to support clinicians in the ongoing implementation and steadily increasing use of the ORPHAcodes.

To constitute national codification support hubs and develop common guidelines concerning challenging coding issues and answers to FAQ in cooperation with other national hubs. Share success stories and benefit from the experiences of each other. Create an efficient national and international helpdesk infrastructure.

To get strong support regarding properly training on team members.

Discussion and conclusion

The overall picture of RD coding with ORPHAcodes turns out to be very different between work package 4 participating countries. The range extends from already implemented ORPHAcodes with linkeage to ICD-10 in national coding systems used in all hospitals to not yet using ORPHAcodes at all. Only a few countries can already look back on a somewhat longer experience with ORPHAcoding. When ORPHAcodes are used it is at the point of care in most cases, and is mostly performed by clinicians. Also the use of the different coding systems (ICD-9, ICD-10 WHO and national modifications, ICD-11, SNOMED, OMIM, HCP) is very heterogeneous between the participating countries.

The need to develop a process for answering questions on coding with ORPHAcodes represents a desire expressed by most participants. The establishment of national and international helpdesks should be able to implement this wish. Also the implementation of training programs is desired by most participants. On these topics, the exchange of experiences with already experienced participants during the project year should be very helpful. Training material from the EJP-RD project is already partly used and should be expanded during the project year, as well as guidelines and recommendations from the RD-CODE project. Training tailored to the implementation of ORPHAcodes into hospital information systems could be helpful in overcoming further hurdles. Very helpful regarding the acceptance of usage of ORPHAcodes should be the highlighting of their fundamental importance in achieving progress in diagnostics and therapy of RD patients.

As a main barrier encountered in the implementation process of ORPHAcodes is considered to be the lack of legislative framework. Consequently the involvement of health authorities representatives is considered as crucial by almost all participants. It should be very helpful that in all cases contacts already exist. Also the involvement of the ERNs is considered to be important by the participants. Contacts do not yet exist in every country; these should be built up and expanded. Considering the implementation of ORPHAcodes to national coding systems the experience from the RD-CODE project should be very useful.