



**Deliverable 4.1**  
**OD4RD2 State of play survey**  
*31.08.2023*

This survey represents deliverable 4.1 of the OD4RD2 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 May, 04th and May, 28th 2023. It has been produced by the leaders of the OD4RD2 - Work Package 4 (WP4). The OD4RD2 project has been launched in April 2023 for a 33 months period.

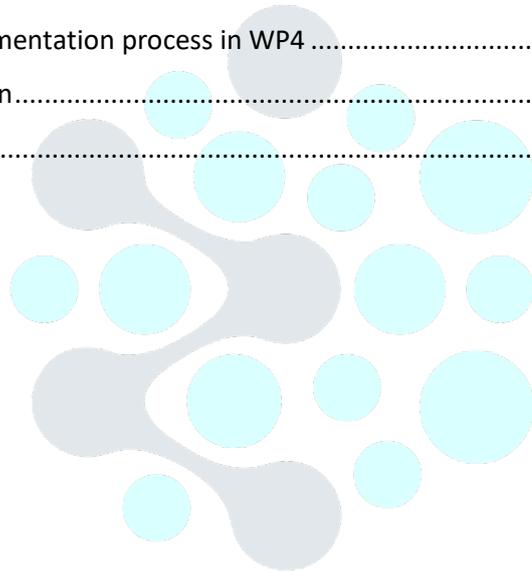
More information on the activities of OD4RD can be found at [www.OD4RD.eu](http://www.OD4RD.eu).

**Disclaimer:**

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OD4RD

Orphanet Data For Rare Diseases

## 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 RD and therefore possesses great significance in its entirety. More than 6,000 different RD are actually known. Since 89% of all RD have a prevalence less than 1:1,000,000 each 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. Still RD are poorly represented in existing medical terminologies in use, and there is no terminology specific to RD, 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 June 8th 2009 on an action in the field of RD, of the improvement in codification of RD. This harmonised and standardised codification was seen as 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 RD field. The nomenclature is aligned with several non-RD terminology resources allowing for semantic interoperability in a context of heterogeneity of coding systems used in different countries and contexts. It is a preferred code system in the Patient Summary Guideline of the eHealth Network<sup>1</sup>, part of the Set of common data elements for Rare Diseases Registration<sup>2</sup> and used in multiple international projects, such as the X-eHealth project on exchanging electronic health records<sup>3</sup>. The OD4RD1 and OD4RD2 projects 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 different settings (health records, registries) and amongst countries, by the dissemination of good coding practices at the data source level.
3. To support evidence-based decision-making in the frame of the European strategy around ERNs, by contributing to the exploitation of reference corpus of data and information on RD.

The project started in January 2022 with a pilot phase ending in March 2023, in which the basics for achieving the project's goals were developed and tested. Thus, the first national action plans have already been developed, which included activities in the areas of training, networking and the establishment of national helpdesks. Fifteen countries participated in this

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<sup>1</sup> [https://health.ec.europa.eu/publications/ehn-guideline-patient-summary\\_en](https://health.ec.europa.eu/publications/ehn-guideline-patient-summary_en)

<sup>2</sup> [https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements\\_en](https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements_en)

<sup>3</sup> <https://www.x-ehealth.eu/>

pilot phase OD4RD1, and six new countries joined the project in the second phase OD4RD2, starting in April 2023.

## Objective

The objective of WP4 is to ensure coordinated local 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.

To fulfil this objective the assessment of the current situation in terms of codification of RD patients in health information systems across the participating countries is necessary. It will allow to adapt the national action plans to the specific situation of each participating country.

## Methodology

The questions of the survey were taken from the state of play survey of the OD4RD1 project. BfArM and INSERM worked together to identify key questions that needed to be answered regarding the coding systems in participating countries and discussed them with the OD4RD2 Executive Committee. The questions were divided into four sections: existing structures and plans, helpdesks and support, training and overall implementation process in WP4. The questions were submitted partly as a multiple choice procedure and partly as open comment fields.

The survey was developed with an online platform tool (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, Bulgaria, Czech Republic, Estonia, Finland, Germany, Ireland, Italy, Latvia, Lithuania, Netherlands, Norway, Poland, Portugal, Slovenia, Spain, Sweden and Switzerland\***

\*Switzerland is participating in WP4 as an observer

Romania as a participant of OD4RD2 will officially join the project in January 2024 and has not completed the survey. Of OD4RD1 participants France did not participate in the actual survey.

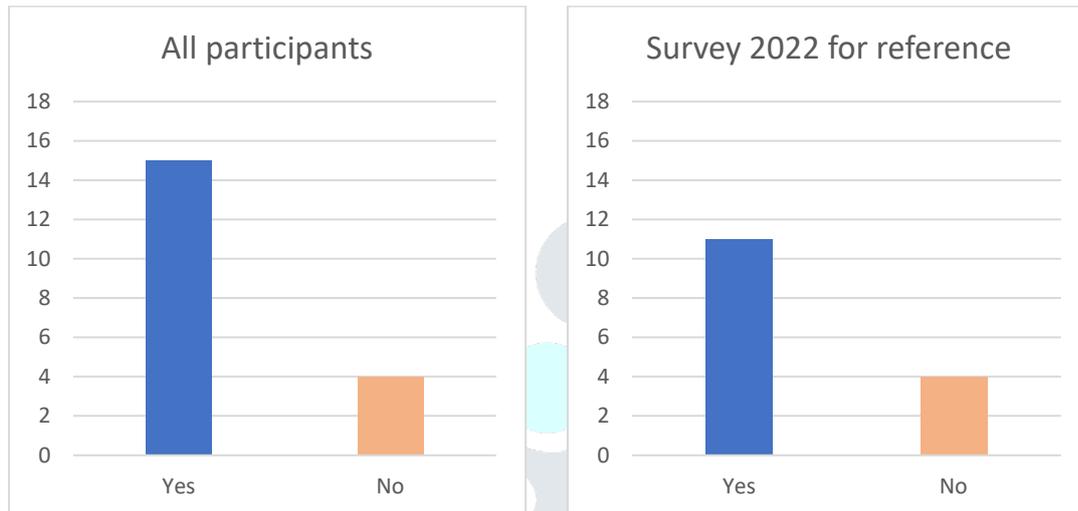
Austria, Belgium, Czech Republic, Finland, Germany, Italy, Netherlands, Norway, Poland, Portugal, Slovenia, Spain, Sweden and Switzerland will be referred to in the following as OD4RD1 participants.

Bulgaria, Estonia, Ireland, Latvia and Lithuania will be referred to in the following as new participants.

## Results

### Section A: Section on existing structures and plans

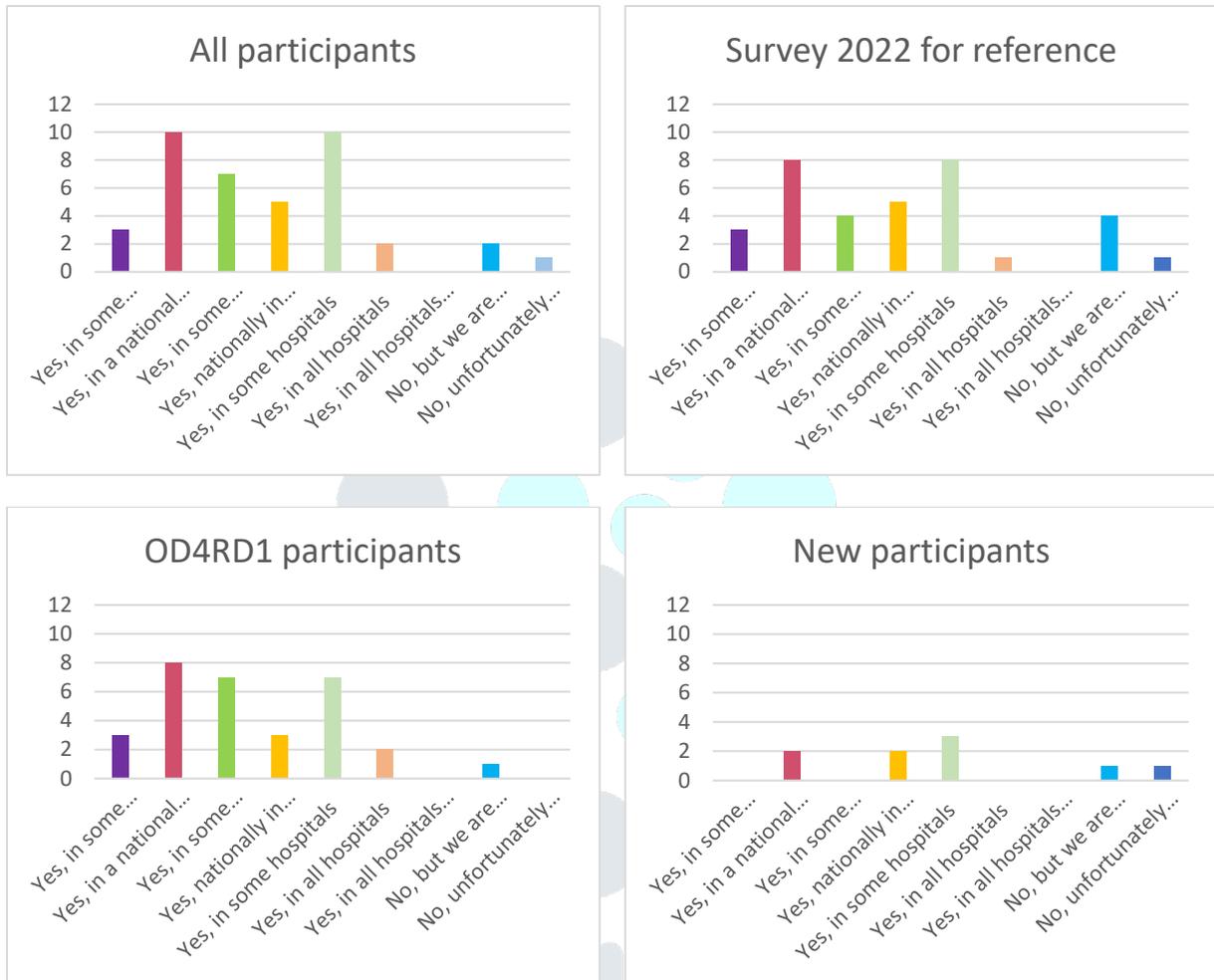
Does your country have a regional or national plan/strategy to code RDs?



**Figure 1: National strategy/plan to code RD**

A majority of the participating countries (15, or 79%) have a regional or national plan or strategy to code RD. In only four of the participating countries (Estonia, Spain, Sweden and Norway) such a plan is not yet in place. With four of the five new OD4RD2 joining countries also having such a plan or strategy, the percentage compared to the 2022 survey increases from 73% to 79%.

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



**Figure 2: Usage of ORPHAcodes at MS and/or regional level**

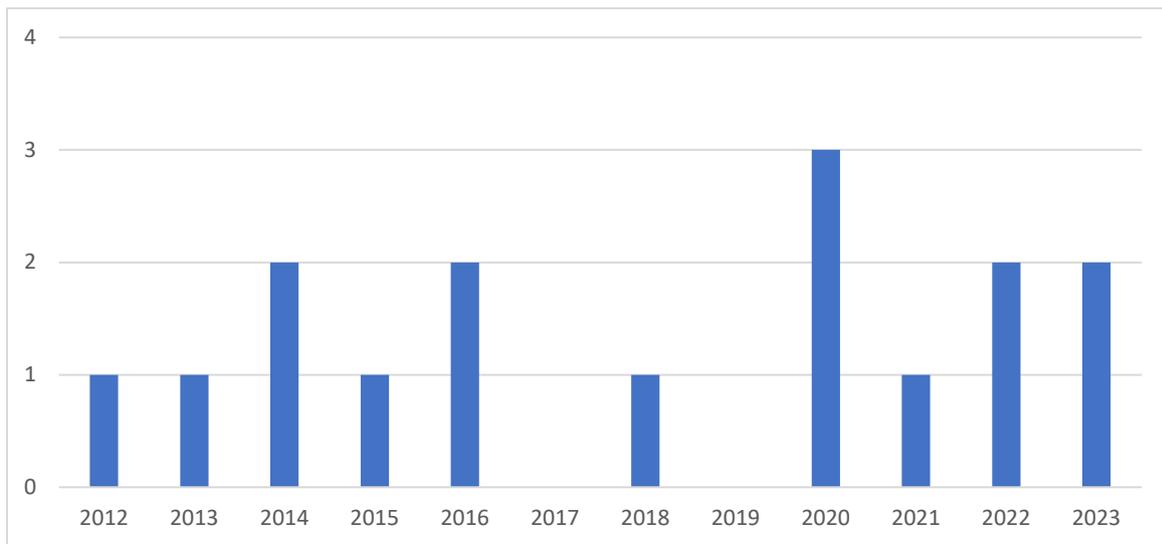
**Explanation:**

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

In two participating countries ORPHAcodes are used to produce data or statistics for RD in all hospitals. In most cases ORPHAcodes are used in registries, either national registries (10) and/or regional registries (3). In most countries ORPHAcodes are used in centers of expertise for RD, either nationally (5) or regionally (7). In three countries ORPHAcodes are not yet used for that purpose, but two of them are preparing for implementation. Among the five countries that participated in 2022 that did not use ORPHAcodes at the time of last year’s survey, only



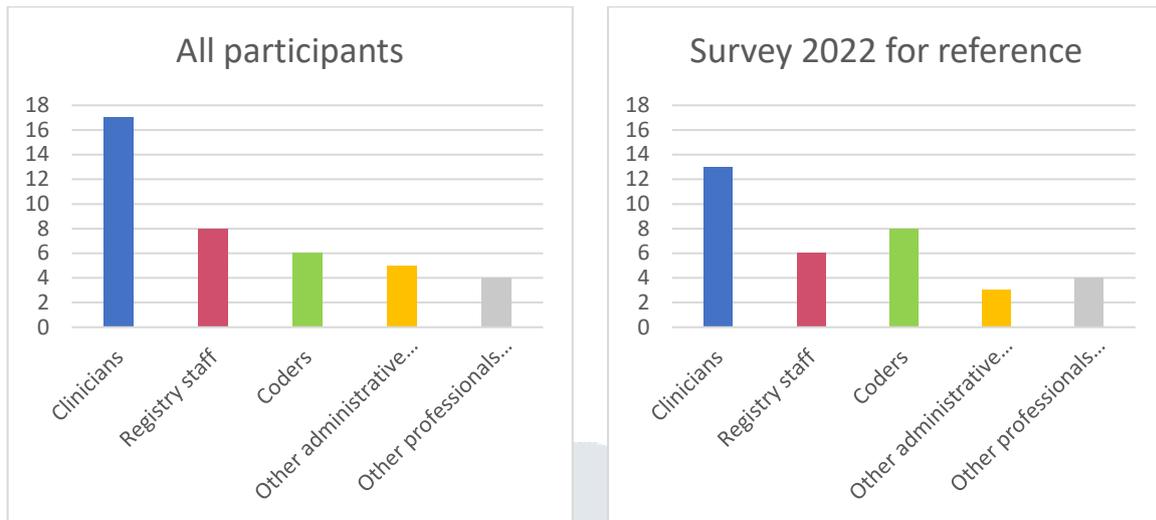
**If yes, when did the recording of RD patients using ORPHAcodes start in your country?**



**Figure 4: Starting year of using ORPHAcodes, number of countries**

In most of the countries using ORPHAcodes the usage started in 2020 (3), in two countries each it started in 2014, 2016, 2022 and 2023 respectively, and in one country each it started in 2012, 2013, 2015, 2018 and 2021. That means that half of the countries using ORPHAcodes have experience of more than four years, four countries are still collecting first experiences. Three of the new countries joining OD4RD can already refer to a usage of ORPHAcodes for more than six years.

**If yes, who is involved in the codification process?**



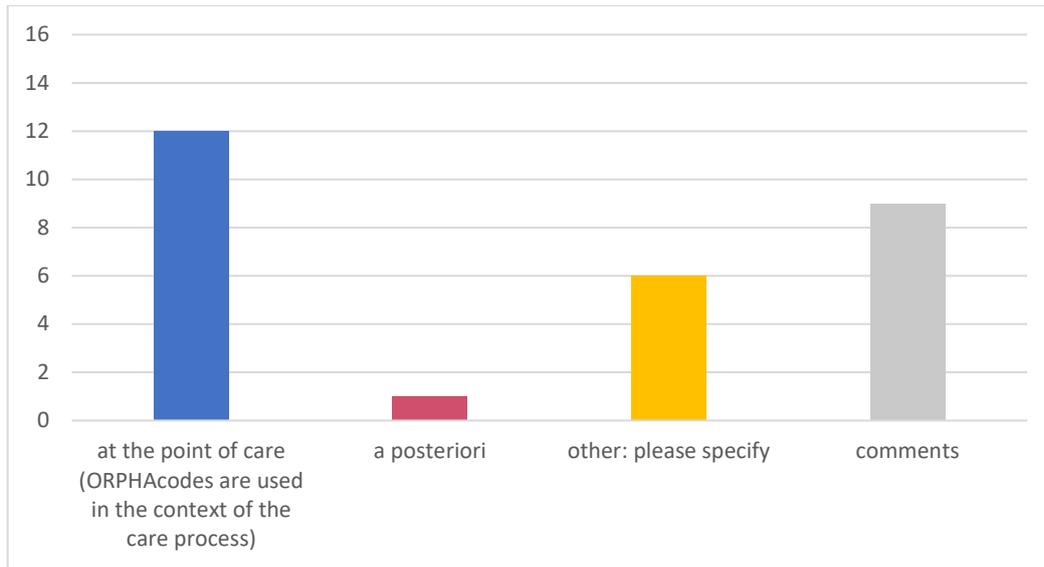
**Figure 5:**  
**Involved personnel in codification process**

**Explanation:**

- 1<sup>st</sup>: Clinicians
- 2<sup>nd</sup>: Registry staff
- 3<sup>rd</sup>: Coders
- 4<sup>th</sup>: Other administrative staff
- 5<sup>th</sup>: Other professionals (please specify)

In 89% of the countries clinicians are involved in the codification process, representing the most important user group in the coding of RD. Furthermore, the competence varies a lot: in 42% of the countries registry staff takes care of the codification. In 32% of the countries coders do the coding work. Coders are not involved in the five new participating countries, which represents the clearest difference to the result of last year's survey. In 47% of the countries coding is done by other administrative staff (26%) or other professionals (21%). Additionally in some countries the nursing staff, study nurses, clinical laboratory geneticists, clinical research assistants, secretaries or external coding specialists are involved in the codification process.

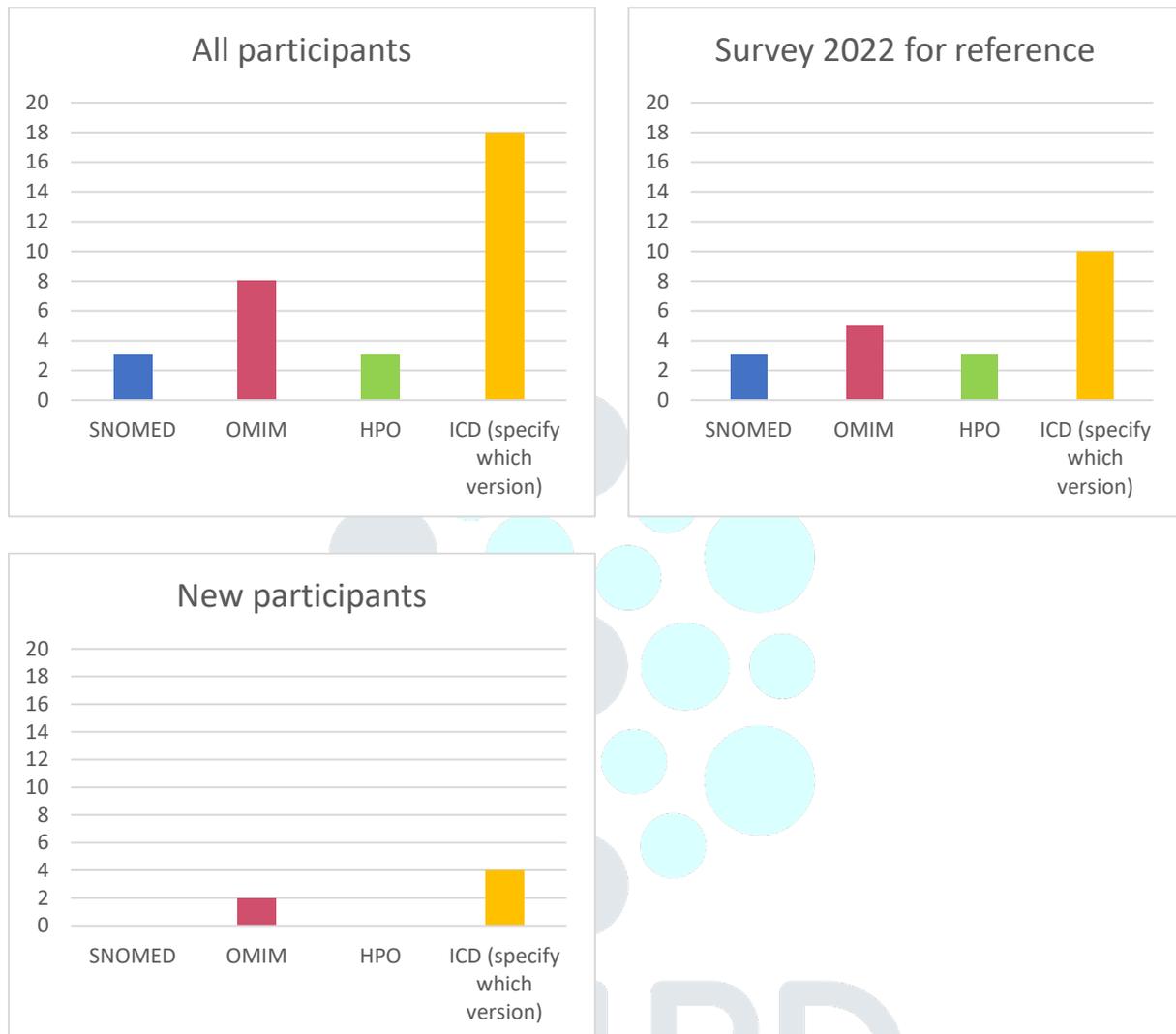
**Please describe at which point of the patient pathway ORPHAcodes are used in your implementation experience:**



**Figure 6: ORPHAcodes usage at point of patient pathway**

In twelve countries ORPHAcodes are used at the point of care and in one country they are used a posteriori. Of the six countries that voted “other”, in two of them ORPHAcodes are used at the point of care and a posteriori. In one case the ORPHAcode is added in genetic reports, in another case the coding will be performed at RD expert centers without more specification. Two countries commented that 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?**



**Figure 7:**  
**Usage of ORPHAcodes together with other nomenclatures/terminologies/coding systems**

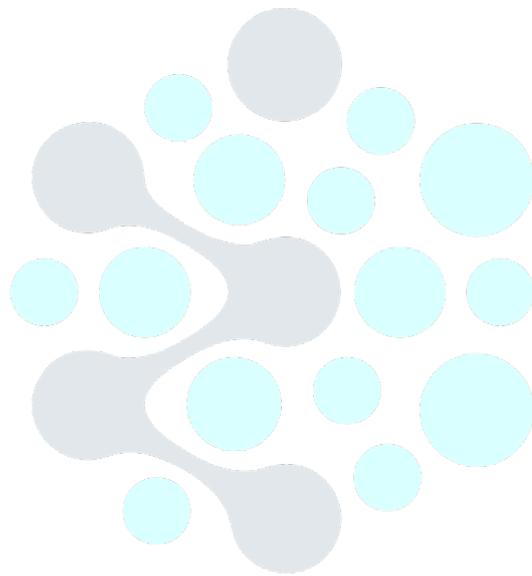
In most of the participating countries (18) ORPHAcodes are used together with ICD for RD coding. ICD-10-WHO or national modifications are used in fifteen countries, one participant stated the usage of both ICD-10 and ICD-9, two participants did not specify the ICD version. SNOMED or HPO are used in three countries respectively. OMIM as an additional terminology is used in eight countries. OMIM is used in two of the new participating countries, while SNOMED and HPO are not used in any of them.

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

Eight participants confirmed an alignment between ORPHAcodes and ICD-10 and/or SNOMED, OMIM and HPO. One participant stated that an alignment is to be implemented possibly in the coming months.

**Could you briefly describe the process put in place?**

The alignment between national modifications of ICD-10 and ORPHAcodes is maintained either annually by the national team, by collaborators of national RD registries, directly via the Orphanet nomenclature files, the Orphanet/SNOMED-CT alignment files, or as a pilot in a few centers of expertise. In one country, a manual cross referencing between ICD10 and ORPHAcodes is practiced. While in another country, due to the ending of the RD-CODE project which enabled the maintainance, alignment is only kept for 24 RDs in a national registry.



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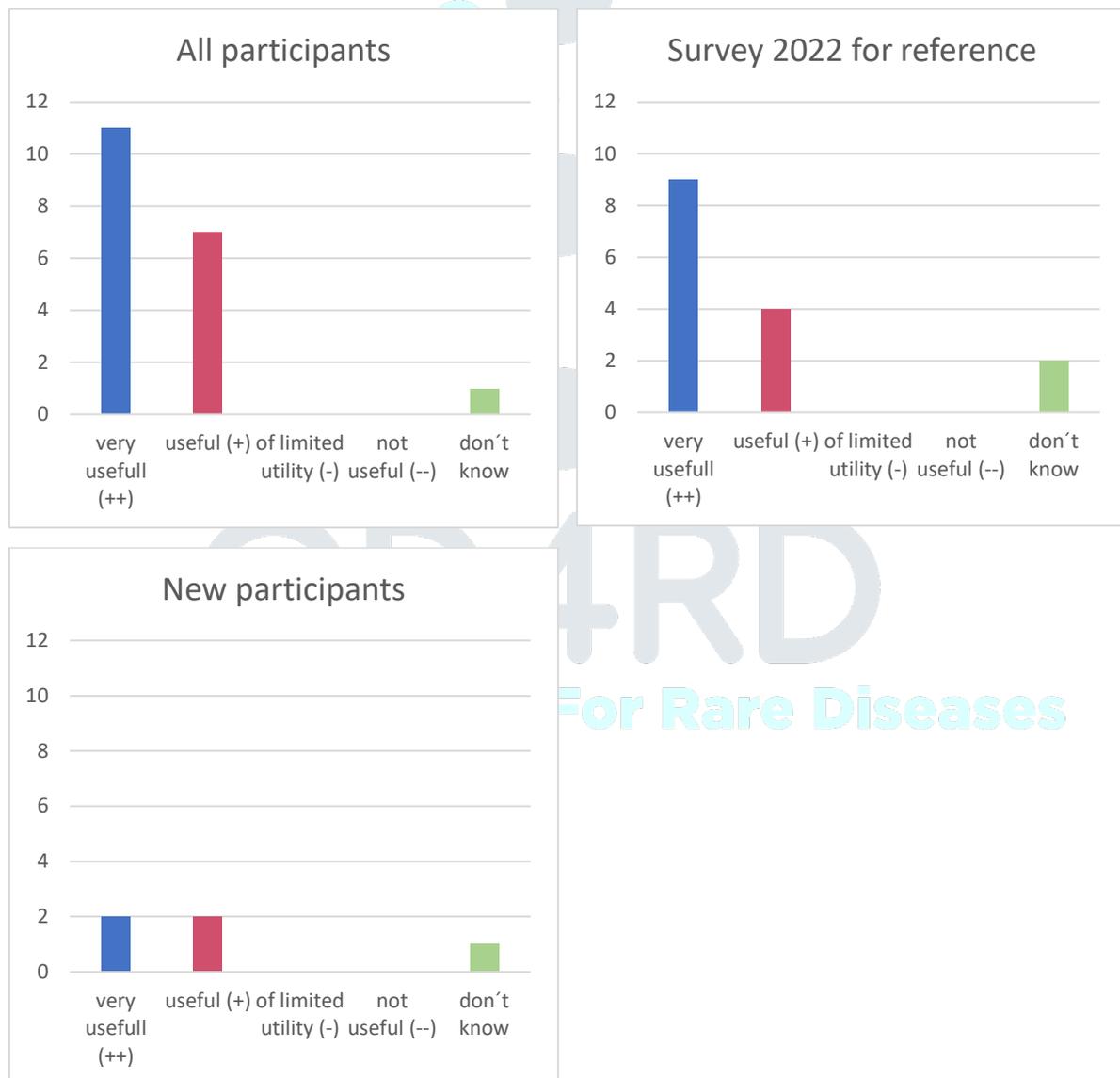
Orphanet Data For Rare Diseases

## 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 eleven countries a process is in place to manage questions coming from ORPHAcodes users via GitHub, by e-mail and/or phone. In one country a coding working group is answering questions at monthly meetings. Three national hubs are still in the implementing process of their helpdesks, while four countries do not have one yet.

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

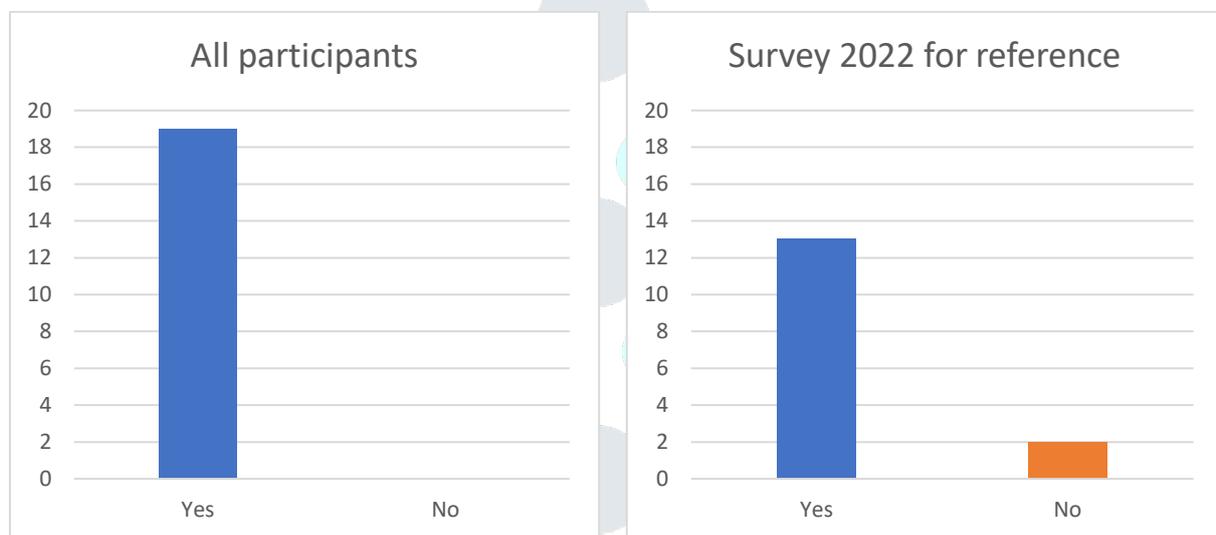


**Figure 8: Estimation of usefulness of helpdesk setup**

The majority of participants consider providing a local helpdesk as very useful (11) or useful (7), only one new participant (Latvia) is actually uncertain about the benefit.

The two countries that chose *don't know* in the 2022 survey evaluate the usefulness of setting up a national helpdesk as *very useful* (1) or *useful* (1). One country that established a national helpdesk changed their estimation from *useful* to *very useful*, while another one that hasn't implemented a helpdesk yet changed their estimation from *very useful* to *useful*. Among the five new countries only one has already implemented a national helpdesk, but four of them estimate the implementation as *very useful* (2) or *useful* (2).

**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?**

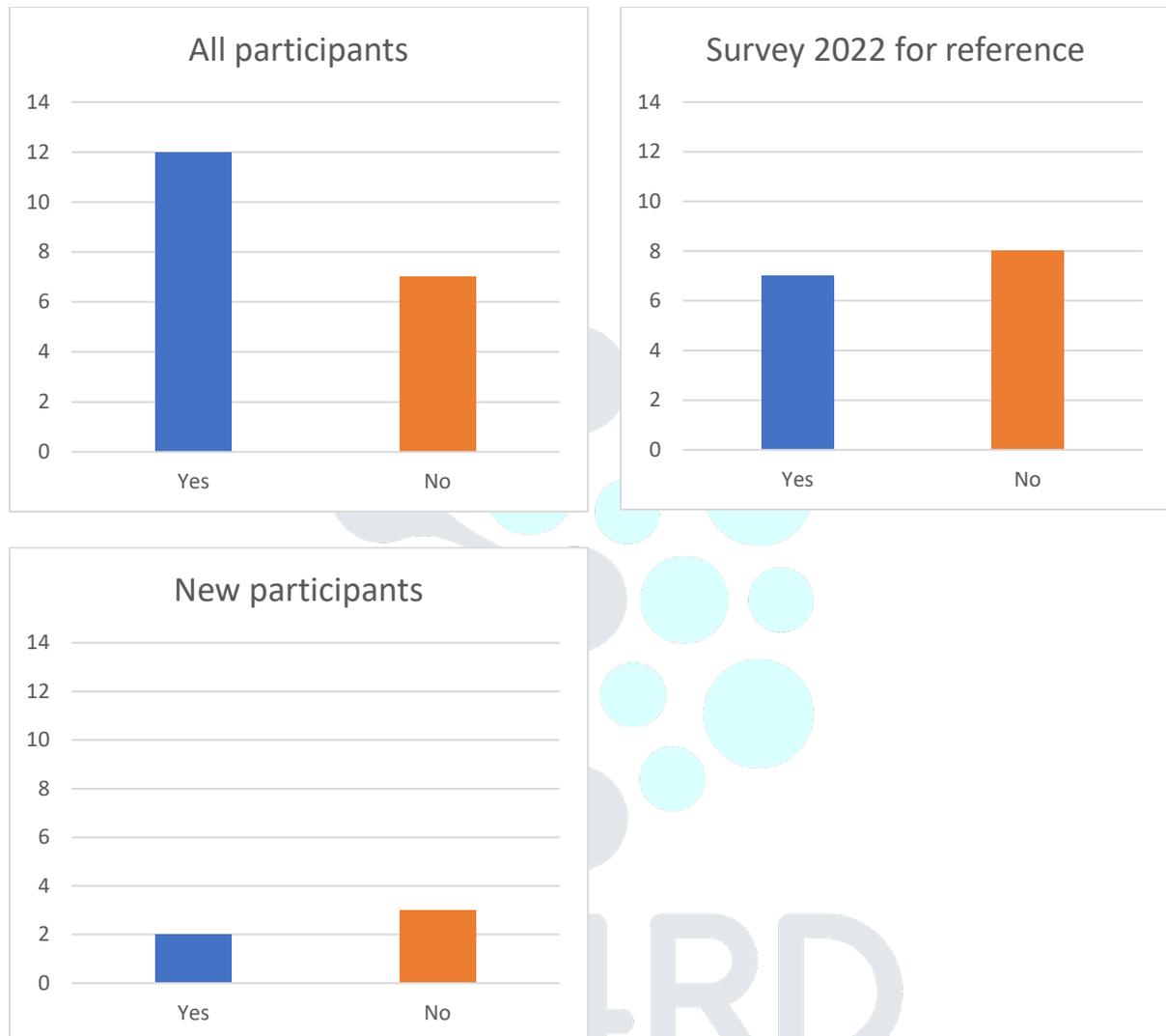


**Figure 9: Estimation of usefulness of national community of practice**

All nineteen participants consider a national community of practice as helpful. In last year's survey, thirteen participants answered this question with yes, while two participants were skeptical.

## Section C: Section on Training

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



**Figure 10: Organized training sessions**

Twelve countries have already organized training sessions for users involved in the codification process, while seven have not yet. Compared to the results of the 2022 survey (7 yes, 8 no) there is a significant increase in the number of countries that organized training sessions. It is worth mentioning that two of the new countries have already organized training sessions too.

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

Of the twelve countries that have already organized training sessions for coders, four developed new training resources like presentations, flyers, videos or quizzes. The other eight countries made use of existing material from Orphanet, RD-CODE and EJP-RD, mostly modifying and translating them into their national language.

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

In eleven of the twelve countries that already have organized training sessions for RD coders, the training sessions have been organized on purpose. One country has embedded them into other trainings. From the seven countries that have not yet organized training sessions, four of them intend to organize future training sessions on purpose, while three would prefer to integrate them into other training sessions.

**Did you develop training material?**

Seven countries already developed training material, mostly presentations in their local language, but also translated videos, flyers, exercises, guidelines on ORPHAcoding, leaflets or quizzes. One country that has not yet organized training sessions is preparing an online course.

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

A lot of comments submitted recommend the use of practical examples for ORPHAcoding of various clinical diagnoses, e.g. through hands-on sessions. It is recommended to concentrate on the area of expertise of the participants (e.g. ORPHAcodes describing neurological RDs for neurologists etc.). Additionally, the need of clear and uniform coding with ORPHAcodes and their benefit for improvement in diagnostics and therapy of RD should be demonstrated. Furthermore, the training sessions should allow to understand the multi-hierarchical Orphanet nomenclature more in-depth, how to use ORPHAcodes correctly, to show the usage of group codes and to clarify how to code patients affected by a multi-systemic syndrome. An interactive presentation of the coding procedure directly on the Orphanet website could be helpful, depending on the solution of ORPHAcoding in the various countries, and could clarify where to find necessary information about RD coding. Also, the training sessions should be of limited length and should be tailored to groups with different levels of prior knowledge. Feedback from advanced countries in terms of ORPHAcoding and experience reports from recognized experts in the field of RD who already use ORPHAcodes could be very helpful to include into trainings. Furthermore, it would be important to show benefits of ORPHAcoding for clinicians, e.g. regarding the recruiting of patients for clinical trials or research projects. Especially the advantages of using ORPHAcodes over classifications/terminologies like SNOMED-CT or ICD-10 for the coding of RDs were regarded as useful in training sessions. Finally providing lists of RDs and related ORPHAcodes for clinicians and utilizing the HPO classification have 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?**

As in last year's survey the lack of legislative framework to use ORPHAcodes and the lack of incentives for using them are considered as main barriers encountered in the implementation process. Other barriers that were mentioned are low motivation for implementation of ORPHAcodes into hospital information systems due to high expenditure, little available time and personnel to dedicate, low RD priority and lack of funding for administrative staff in hospitals. The next barrier is the lack of knowledge about the high benefit of ORPHAcoding of RD among decision-makers and several clinicians. It has been seen that it can be difficult to convince the different societies of medical specialists to support ORPHAcoding. Also, the lack of knowledge about the OD4RD project at institutional level and ERN-affiliated health care providers were mentioned. If ORPHAcoding has started, it might be difficult to reach a consensus decision on regional and/or national level regarding the usage of ORPHAcodes. Technical challenges in the software or health care record application/solution might also cause problems in the implementation process. Finally, the language barrier might be a problem in countries that do not yet provide a translated version of the Orphanet nomenclature.

#### **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?**

The need for support by health authorities like ministry of health and health insurance companies to enforce the (mandatory) usage of ORPHAcodes by hospitals or registries is reported as the main driver. Therefore, to demonstrate the benefits for advances in diagnostics and therapy for RD patients, but also the benefits for hospitals, clinicians and the health care system would be important. The reimbursement should better reflect the treatment costs of RD patients, with ORPHAcoding providing higher level of granularity to payers compared to ICD-10. Even a European guidance and incentives for the implementation of ORPHAcodes into electronic health records have been suggested. A special reimbursement for RD would increase the motivation of the stakeholders to implement and use ORPHAcodes. Also, it will be important to increase the communication with ERN representatives by the national teams supported by coordinated efforts.

Effective training activities, performed by the national teams and Orphanet experts, using sufficient training material, should disseminate the benefits and correct usage of ORPHAcoding. It has been considered important to provide IT expertise or a dedicated tool to facilitate the implementation of ORPHA codes into health information systems. A clear explanation on the support they could receive from the Orphanet IT team at INSERM regarding a technical solution facilitating the automatic entry of ORPHAcodes data would be helpful. Regarding the ORPHAcoding by users it will be important to have a user-friendly solution. ORPHAcodes should be implemented in a way that no additional step is required from the clinician when registering a diagnosis. A convenient tool for ensuring the coding process or

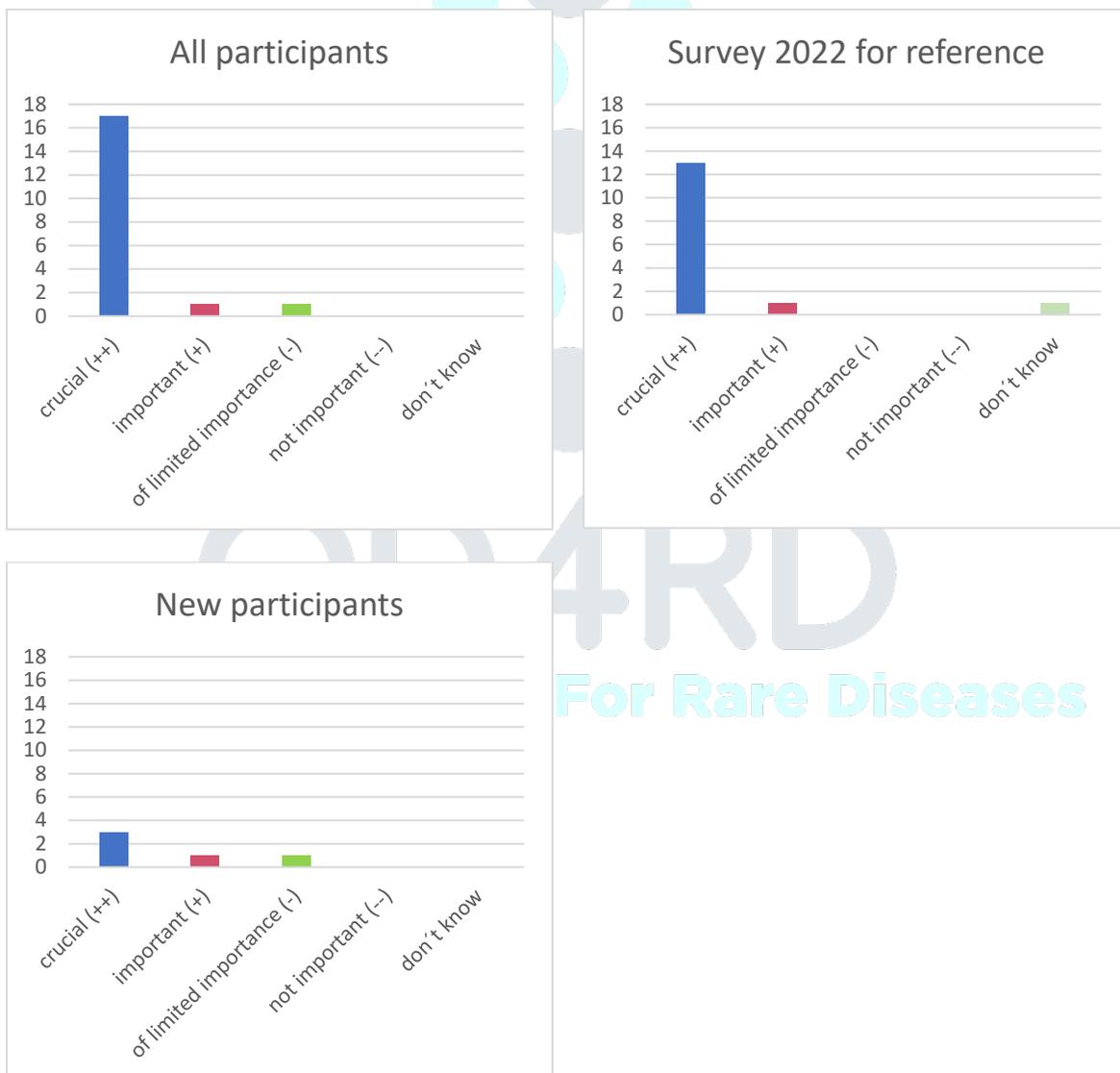
lists of RD and ORPHA codes provided to clinicians to help using codes in clinical practice could be helpful in this regard.

Finally, it has been emphasised that sharing of experience, as well as useful information material and documents between participating national teams will be essential to successfully achieve the project goals.

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

The rated level of the need of involvement of the main stakeholders for a successful ORPHA codes implementation is shown in the following illustrations. Participants had the opportunity to choose between the importance levels *crucial*, *important*, *of limited importance*, *not important* and *don't know*.

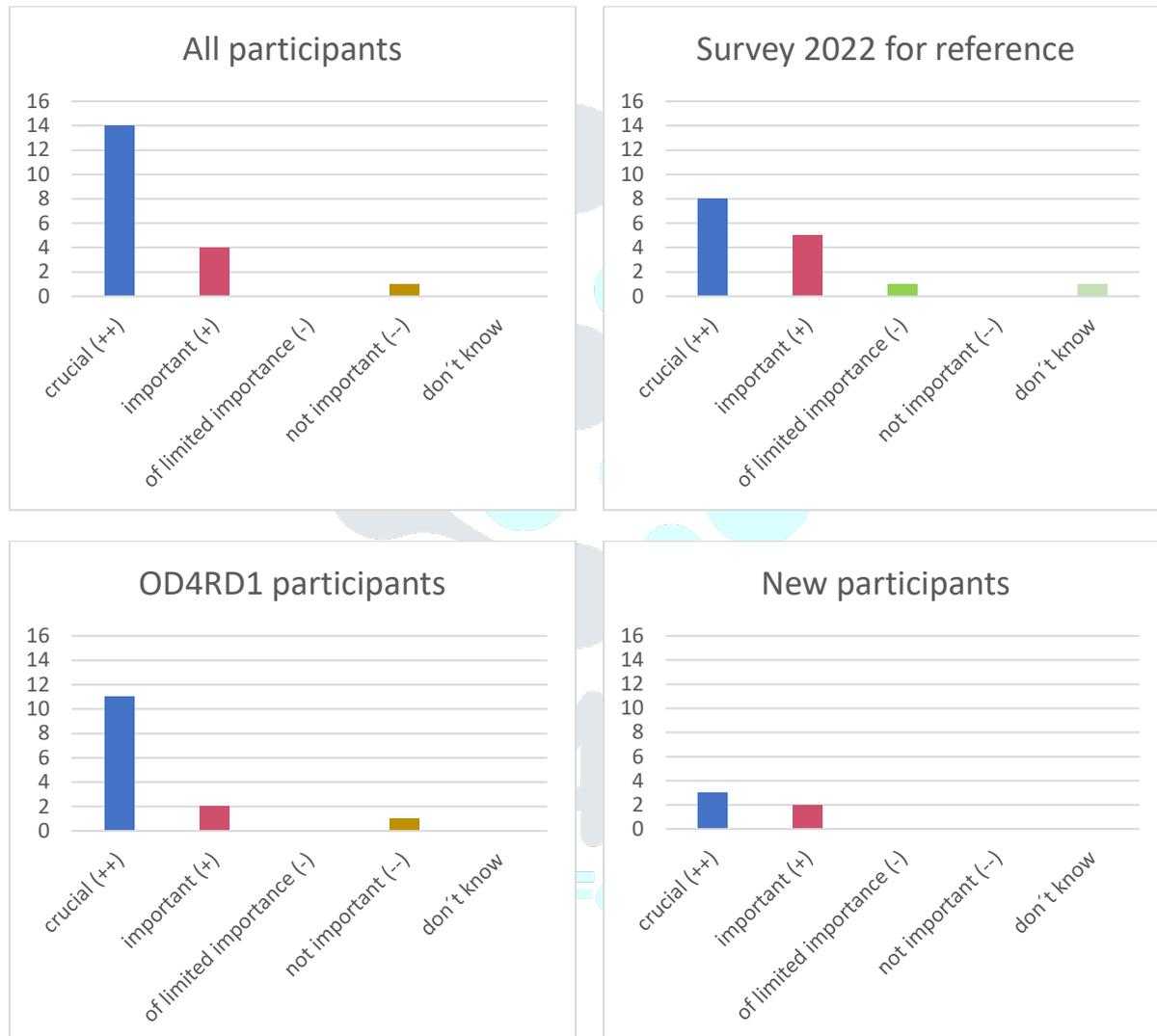
Health authorities representatives:



**Figure 11: Need of involvement of health authorities representatives**

As in the previous year's survey, the involvement of health authorities representatives was again rated as most important with regard to the need of involvement for a successful ORPHAcodes implementation. Seventeen participants rated their involvement as *crucial*, while one of the new participants rated the integration as *important* or *of limited importance* respectively.

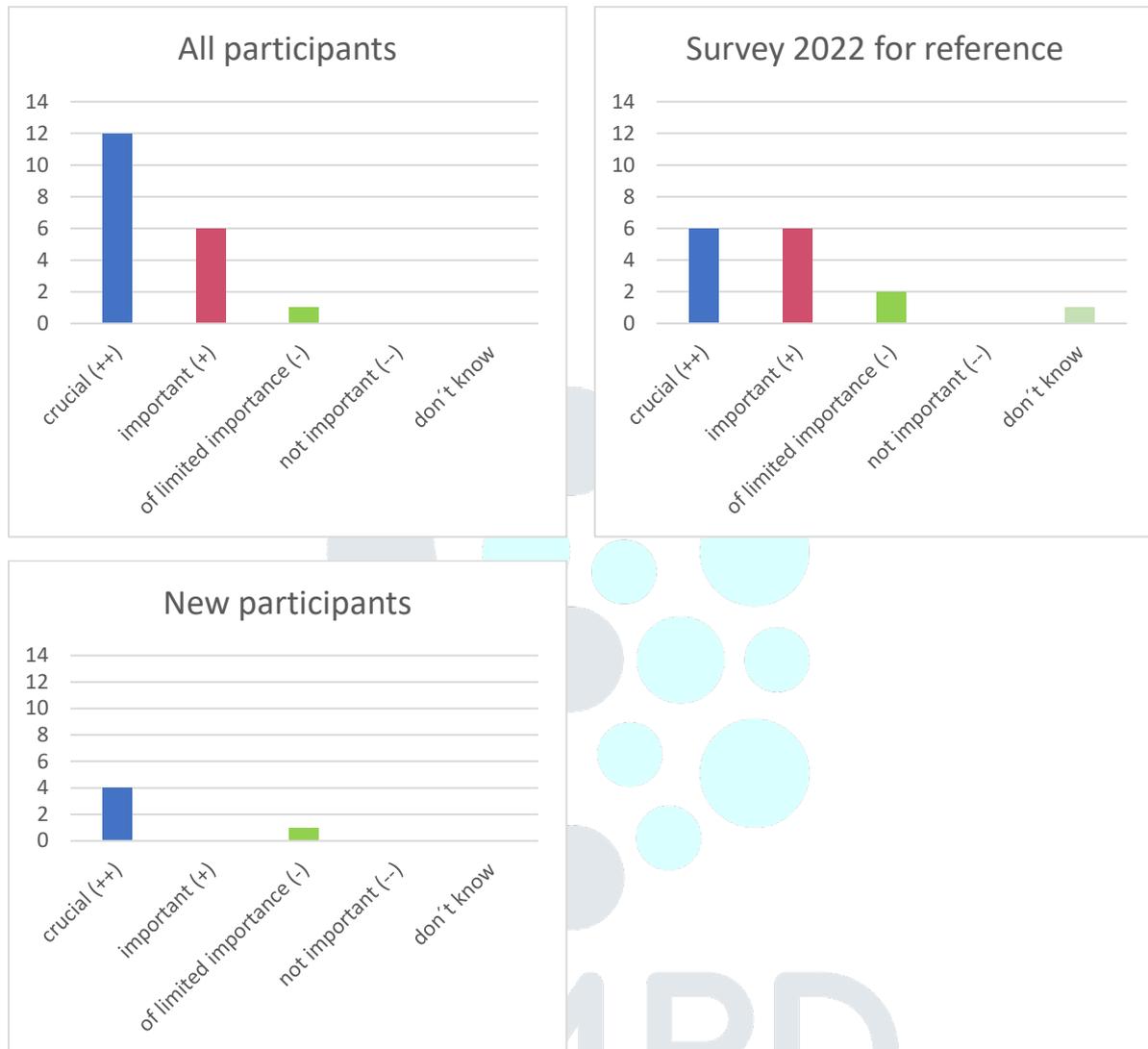
Clinicians involved in HCP members of ERNs:



**Figure 12: Need of involvement of clinicians involved in HCP members of ERNs**

Clinicians involved in HCP members of ERNs were rated as the second most important stakeholders for successful ORPHAcodes implementation, up from third place in the 2022 survey. Comparing the histograms in figure 12 it can be noticed that especially the OD4RD1 participants consider this group to be more important than a year ago. This may result from the experience of the OD4RD1 project.

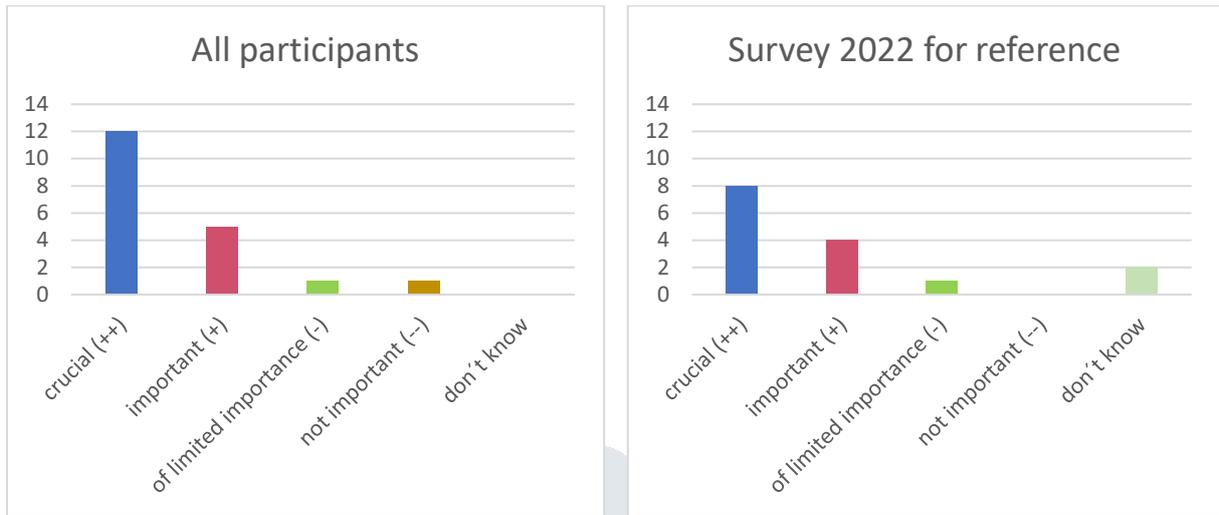
Clinicians working in other RD Centers:



**Figure 13: Need of involvement of clinicians working in other RD centers**

The clinicians working in other RD centers were rated as significantly more important for the successful implementation of ORPHAcodes than in last year’s survey. Twelve participants rated their involvement as *crucial*, compared to six in last year’s survey. The rating of *important* was given by six participants, the same result as in last year’s survey. Four of the five new participants rated the involvement of clinicians working in other RD centers as *crucial*.

Registry managers:

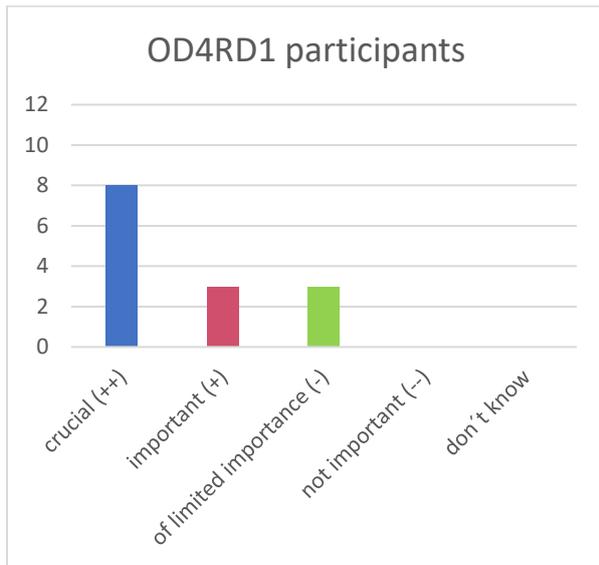


**Figure 14: Need of involvement of registry managers**

The need for involvement of registry managers was rated in fourth place similar to the rating in the 2022 survey. The figures of both surveys look very similar, only one participant did rate the involvement of registry managers as *not important*.

Hospital managers:





**Figure 15: Need of involvement of hospital managers**

The rating of the need of involvement of hospital managers for a successful ORPHAcodes implementation declined from second place in the 2022 survey to fifth place in the 2023 survey. The participants of OD4RD1 are particularly responsible for this assessment. Their rating of *limited importance* gains two approvals at the expense of the ratings *crucial* and *important*, which each lost one rating.

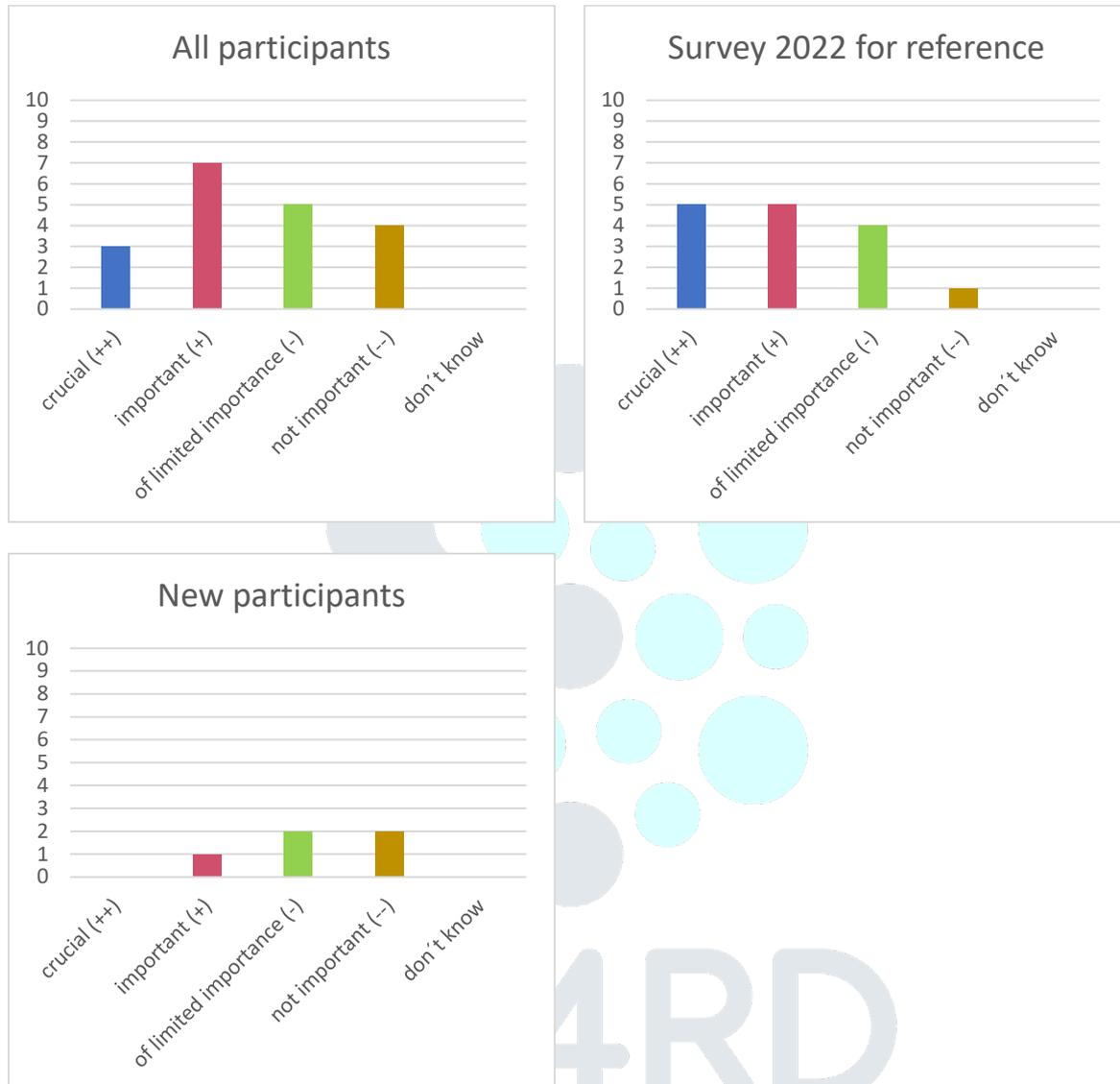
ERNs representatives:



**Figure 16: Need of involvement of ERN representatives**

Ranked sixth in the need of involvement for a successful ORPHAcodes implementation are ERN representatives, the same rank as in the survey of last year. Although the importance is apparently rated higher in the current survey: the rating of *crucial* gained six votes at the expense of two *don't know* ratings.

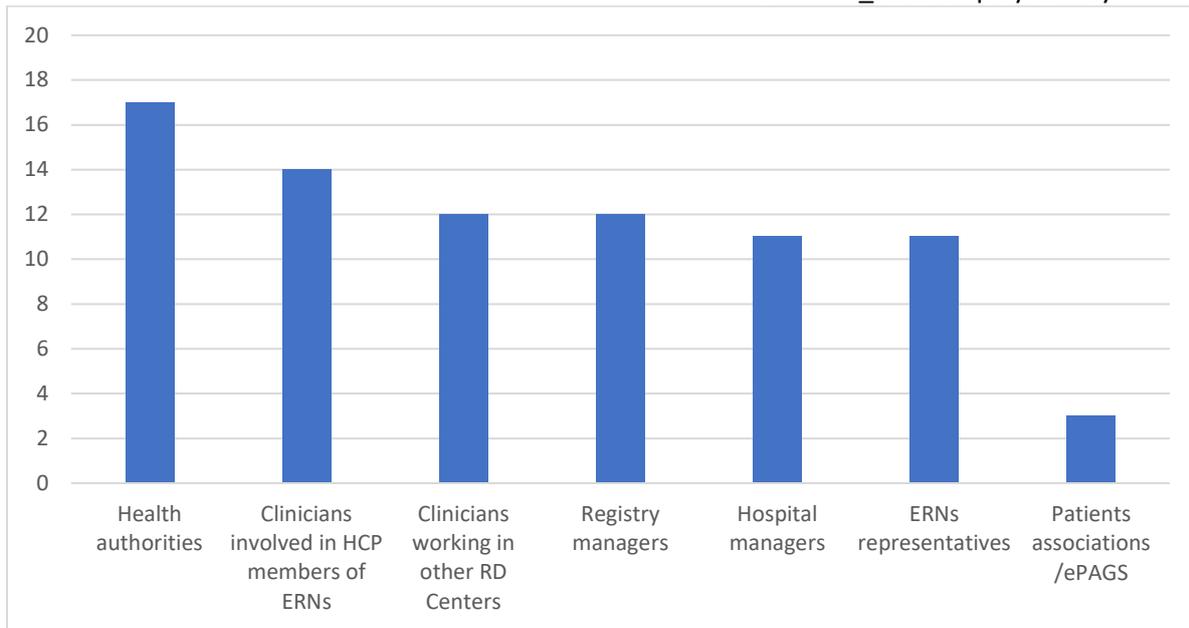
Patients associations /ePAGS:



**Figure 17: Need of involvement of patient associations**

Finally, the need of involvement of patient associations was the lowest rated of the list of stakeholders offered for selection, as it was in the 2022 survey. Especially among the new participants, there was only one rating as *important*, two voted for *of limited importance*, and two valued *no importance* in their involvement.

In summary, the involvement of all the stakeholders offered for selection was considered at least *important* for a successful implementation of ORPHAcodes by most participants. Figure 17 shows the need of involvement ratings as *crucial* for the different stakeholders. The involvement of health authorities is rated as *crucial* in most answers, the involvement of patients associations in the fewest.

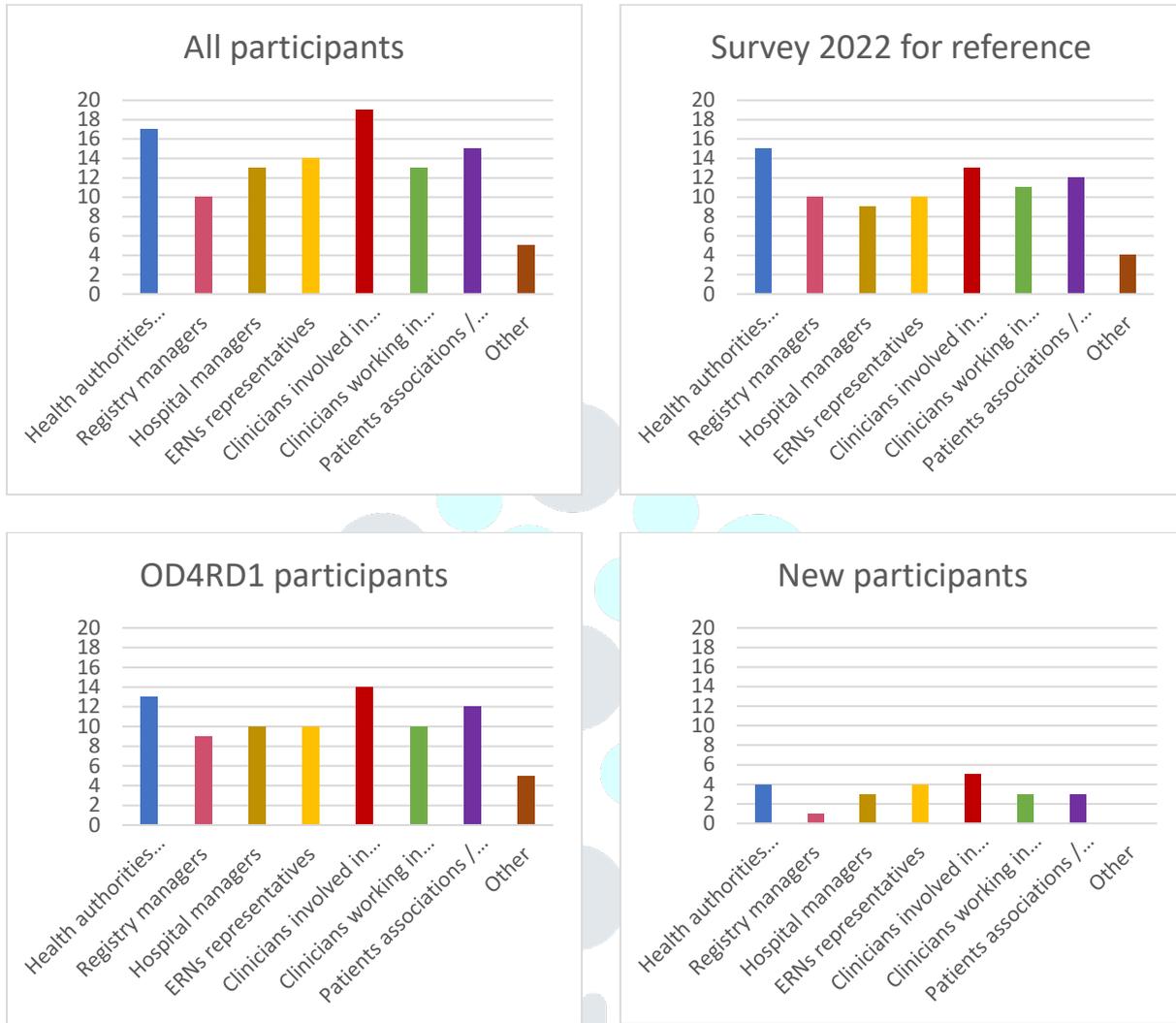


**Figure 18: Crucial need of involvement of the following stakeholders for a successful ORPHAcodes implementation**

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

The following additional stakeholders were listed by the participants in a free comment box: software manufacturers and developers of health information systems; electronic health record suppliers; other professionals involved in codification; primary care physicians; health insurance companies; researchers, data and IT managers and politicians; national authorities responsible for information systems in the national health system, including a national health insurance fund; national digitalisation agency setting the national interoperability framework and eventually distributing the nomenclature; learned societies.

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



**Figure 19: Existing institution contacts**

**Explanation:**

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

Other mentioned institutions, where contacts exist: Health information system developers (2), health care data/IT managers (1), electronic health record suppliers (1) and health insurance companies (1).

Contacts with clinicians involved in HCP members of ERNs exist in all participating countries, while health authorities representatives, with which most existing contacts were named in the

2022 survey, were mentioned by seventeen countries. Consistent with the results from last year's survey existing contacts with patient associations were mentioned the third most frequently (15), followed by existing contacts with ERN representatives (14), hospital managers and clinicians working in other RD centers (13 each). The least contacts exist with registry managers (10). Compared with the 2022 survey contacts with clinicians involved in HCP members of ERNs (6), hospital managers (4) and ERN representatives (4) increased the most.

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

A lot of different expectations have been listed.

One key expectation is to implement ORPHAcodes into practice and to expand the use of ORPHAcodes in ERN members of the European countries, to somehow make ORPHAcoding mandatory in the EU. Therefore, the experience gained and the challenges identified in OD4RD1 should help to refine and extend the strategies in OD4RD2. It is expected to share resources and success stories and benefit from the experiences of each other. OD4RD2 should provide guidance on how to deal with various stakeholders, increase the visibility of the project, including the national hub activity, among target stakeholders.

The creation of an efficient national and international helpdesk framework, a toolkit for implementation that includes guidelines, training videos, FAQs and shared implementation plans from other participants, having access to pathways and solutions between all partners within the project and to participate in training sessions are also key expectations. The setup of national trainings and online courses to increase awareness of the relevance and benefits of ORPHAcoding and to improve the clinicians' knowledge of diagnoses and ORPHAcodes were also mentioned.

Another key expectation is the discussion of practical use cases to develop common coding guidelines with OD4RD GitHub support, and the development and completion of data in the Orphanet nomenclature. Also the alignment of ORPHAcodes with the SNOMED terminology and the ICD-11 classification was mentioned. Furthermore, the implementation of national action plans for the introduction and the development of ORPHAcoding, with technical and training support especially for national hubs that consist of very few team members, are expected. Also, guidance on implementation of ORPHAcoding into local IT systems is considered important.

Finally, it is expected to convince health care professionals to participate in the project, whether there will be support by healthcare authorities or not, and to engage ERNs effectively so that health care providers linked to them would implement ORPHAcodes to initiate showcase projects that can lead to further dissemination of ORPHAcoding.

## Discussion and conclusion

79% of the countries participating in the survey have a regional or national plan or strategy to code RD, while the overall picture of RD coding with ORPHAcodes remains very different in the countries participating in WP4. The range extends from already implemented ORPHAcodes with linkage to ICD-10 in national coding systems used in all hospitals to not yet using ORPHAcodes at all. Half of the countries that use ORPHAcodes can already look back on an experience of more than 4 years. When ORPHAcodes are used it is at the point of care in most cases, and it is mostly performed by clinicians. Also, the use of the different coding systems (ICD-10 WHO and national modifications, SNOMED-CT, OMIM, HCP) is very heterogeneous between the participating countries.

The usefulness of setting up a “local” helpdesk managing requests from users during the implementation process was almost unanimously rated as important or very important, and in eleven countries there is already a process in place to manage questions emerging from ORPHAcodes users via GitHub, e-mail and/or phone. The establishment of helpdesks in the remaining countries should be an important aid in the further implementation of ORPHAcodes. Effective training activities, performed by the national teams and Orphanet experts, using sufficient training material has been identified as one of the main drivers for a successful ORPHAcodes implementation. Twelve countries can already contribute with experience from the trainings and/or workshops already held. The trainings should disseminate the benefits and correct usage of ORPHAcoding, ideally tailored to the needs of the addressed personnel.

The lack of legislative framework to use ORPHAcodes and the lack of incentives for using them were considered as main barriers encountered in the implementation process. Therefore, it will be of particular importance to convince national health authorities of the benefits of ORPHAcoding to introduce or expand it. It should be helpful that in most cases contacts with the national health authorities already exist. The demonstration of resulting benefits for advances in diagnostics and therapy for RD patients, but also the benefits for hospitals, clinicians and the healthcare system would be important to impart. The reimbursement should better reflect the treatment effort of RD patients, with ORPHAcoding providing higher level of granularity to payers compared to ICD-10. Also, the engagement of clinicians involved in HCP members of ERNs was rated as important for a successful ORPHAcodes implementation. Furthermore, it was considered important to provide IT expertise or a dedicated tool to facilitate the implementation of ORPHAcodes into health information systems.

Finally, it was emphasised that the sharing of experience, useful information material and documents between participating national teams will be essential to successfully achieve the project goals. The experience from the pilot project has shown that the regular holding of virtual meetings is of particular importance for this purpose. The experience gained in OD4RD1 should help to refine and extend the strategies in OD4RD2.

# Annex

## Questionnaire

Section A: Section on existing structures and plans	
<p><b>A1. For which national hub (country) are you filling this survey?</b></p> <input type="text"/>	<p>Yes, in all hospitals and all outpatient settings <input type="checkbox"/></p> <p>Comment <input type="text"/></p>
<p><b>A2. Does your country have a regional or national plan/strategy to code RDs?</b></p> <p>Yes <input type="checkbox"/></p> <p>No <input type="checkbox"/></p>	<p>No, but we are preparing for implementation <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>No, unfortunately not yet <input type="checkbox"/></p> <p>Comment <input type="text"/></p>
<p><b>A3. Does your country use ORPHAcodes at MS level and/or regional level to produce data or statistics for RD?</b></p> <p>Yes, in some regions in registries <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Yes in a national registry <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Yes in some regions in centres of expertise for RDs <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Yes, nationally in centres of expertise for RDs <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Yes, in some hospitals <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Yes, in all hospitals <input type="checkbox"/></p> <p>Comment <input type="text"/></p>	<p><b>A4. If yes, when did the recording of RD patients using ORPHAcodes start in your country?</b></p> <input type="text"/> <p><b>A5. If yes, who is involved in the codification process?</b></p> <p>Clinicians <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Registry staff <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Coders <input type="checkbox"/></p> <p>Comment <input type="text"/></p> <p>Other administrative staff <input type="checkbox"/></p> <p>Comment <input type="text"/></p>

Other professionals (please specify)

Comment

**A6. Please describe at which point of the patient pathway ORPHAcodes are used in your implementation experience:**  
 at the point of care (ORPHAcodes are used in the context of the care process)   
 a posteriori   
 other: please specify

**A7. Are ORPHAcodes used in a stand-alone modality or are they used together with other nomenclatures/terminologies/coding systems for RD coding?**

SNOMED

Comment

OMIM

Comment

HPO

Comment

ICD (specify which version)

Comment

**A8. If yes, do you maintain an alignment between ORPHAcodes and these systems?**

**A9. Could you briefly describe the process put in place?**

**Section B: Section on Helpdesks and support:**

**B1. 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?**

**B2. How would you rank the usefulness of setting up a "local" helpdesk managing requests from users during the implementation process?**

Very useful (++)   
 useful (+)   
 of limited utility (-)   
 not useful (-)   
 don't know

**B3. 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?**

Yes   
 No

**Section C: Section on Training**

**C1. Did you organize training sessions for users involved in the codification process?**

Yes   
 No

**C2. Did you use already existing resources or have you created new ones?**

**C3. Have the training sessions been organized on purpose or were they embedded in other training events?**

organized on purpose   
 embedded in other training events (please describe the training event)

**C4. Did you develop training material?**

**C5. Based on your experience, what do you find particularly useful to be proposed in training sessions?**

**Section D: Section on overall implementation process in WP4**

**D1. According to your experience, which were the main barriers encountered in the implementation process that we should address in WP4?**

**D2. 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?**

**D3. How would you rank the need of involvement of the following stakeholders for a successful ORPHAcodes implementation in your country?**

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

	1	2	3	4	5
Health authorities representatives	<input type="checkbox"/>				
Registry managers	<input type="checkbox"/>				
Hospital managers	<input type="checkbox"/>				
ERNs representatives	<input type="checkbox"/>				
Clinicians involved in HCP members of ERNs	<input type="checkbox"/>				
Clinicians working in other RD Centers	<input type="checkbox"/>				
Patients associations /ePAGS	<input type="checkbox"/>				

**D4. Please specify if you think other stakeholders need to be involved:**

**D5. Are you already in contact with the following institutions in your country?**

Health authorities representatives

Comment

Registry managers

Comment

Hospital managers

Comment

ERNs representatives

Comment

Clinicians involved in HCP members of ERNs

Comment

Clinicians working in other RD Centers

Comment

Patients associations /ePAGS

Comment

Other

Comment

**D6. What are your key expectations for WP4 to achieve together in this OD4RD2 project ?**

**Thank you for completing this survey.**