

Deliverable D4.5
OD4RD2 Lessons Learned final report
March 2026

OD4RD
Orphanet Data For Rare Diseases



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Context

This document represents deliverable D4.5 of the OD4RD2 project, which has received funding from the European Union. It contains the lessons learned from the activities in work package 4 by the members of OD4RD2 for the duration of the project. The document has been produced by the leaders of the OD4RD2 work package 4. The OD4RD2 project was launched April 1st, 2023 and is planned to continue until 31st of March 2026. The OD4RD2 project represents a continuation and renewal of the OD4RD project which was launched in January 2022 and was active until March 31st, 2023. More information on the activities and deliverables of the OD4RD can be found at:

<http://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.

Background

There are more than 6,000 recognized rare diseases ([The Orphanet Nomenclature and Classification of rare diseases: a standard terminology for improved patient recognition and data interoperability](#))(Lucano C, 2026), a number that continuously increases as scientific knowledge advances and new conditions are identified. In Europe RD is defined as a disease that does not affect more than 1 person per 2000, or 0.05%, in the European Population. Although each individual diagnosis is rare an estimated 30 million people in the European Union suffer from a rare disease (Nguengang Wakap S, 2020). A substantial proportion of these diseases are chronic conditions which manifest in childhood and are often associated with lifelong impairment. Many are hard to diagnose, sometimes due to the rarity, and as a result often diagnosed extremely late.

Due to the severity and substantial impact of RD, advances in diagnostics and therapy in the field are particularly important, but also particularly challenging due to the small number of identified patients of each diagnosis. In addition, the amount of data needed for research may only be obtainable by compiling data from several countries. To this end, data must be structured in such a way that it can be combined into a cohesive data collection.

In order to permit tracking of rare diseases in health care systems, they must be designated a unique code, as a means to separate them from each other as well as from more frequently occurring diseases. In many disease coding systems, such as ICD-10, which is in common use, a large number of RD lack a unique code. This complicates the identification of and follow-up of RD patients. It was stated as early as 8.6.2009, by the Council of the European Union for action in the field of RD, that: "An appropriate classification and codification of all RD is necessary in order to give them the necessary visibility and recognition in national health systems." ([Council Recommendation 2009/C 151/02 of 8 June 2009 on an action in the field of rare diseases](#)). To improve the coding of RD the use of ORPHAcodes was recommended by the Commission Expert Group on Rare Diseases in 2014 and later selected a best practice for coding RD by the European Commission (EC) in 2017 ([Implementations of best practices European Commission](#)), as the exploitation of ORPHAcodes' annotated health data increases the visibility of people living with a RD (Mazzucato M, 2023), (Gunne E, 2020), (Chiu ATG, 2018).

Several projects have been previously launched and completed in the quest to improve the health and life situation for RD patients in Europe. OD4RD2 continues to build on the foundation established through several previously launched and completed projects aiming to improve health and life situation for RD patients in Europe, including RD codification, such as RD-Action, RD-CODE and OD4RD. Coding of rare disease using ORPHAcodes has steadily progressed toward greater implementation, coordination, and sustainability. What began as exploratory projects has matured into a structured framework supporting the integration of ORPHAcodes across the member countries. The SoA (State of the Art of Rare Disease activities in Europe) was an annual report that aimed to coordinate, map, and evaluate national and EU-level policies to reduce fragmentation, improve diagnosis, and accelerate access to treatments for rare disease patients. The 2018 report can be found here ([Overview Report – RD-ACTION : DATA AND POLICIES for rare diseases](#)). The overview report and previous reports can be found here ([State of the art reports](#)). The iterative approach between projects has allowed continuous improvement and adaptation to identified needs. Early initiatives (RD-Action and RD-CODE) focused on developing standards, producing guidelines and recommendations, and establishing a framework which enables aggregating European data into a reliable and useful data collection as well as beginning the implementation of ORPHAcodes in four selected countries. OD4RD and OD4RD2 continues to build on the lessons learned from previous projects and has had their emphasis on supporting the practical implementation of ORPHAcodes, specifically in health care providers (HCP) hosting or participating in European Reference Networks (ERN), as well as improving the Orphanet nomenclature by using the expertise within the ERNs. Collaboration with the European Reference Networks (ERNs) has also deepened, positioning them as central partners in validating and applying ORPHAcodes in specialized care. This partnership strengthens the link between clinical expertise, coding practice, and research. Orphanet expertise and status as a long-lasting, well-established consortium is a great asset in the quest to improve the knowledge and care available about RD and to RD patients. The evolution of coordinated implementation efforts across Europe marks a crucial step forward in European RD data management. Through the RD-Action, RD-CODE and OD4RD projects, Orphanet have made it possible for European countries to produce homogenous and reliable data, which enables earlier diagnosis, a higher quality of research and the ability to advance RD treatment.

Project Objectives

The OD4RD2 project is organized in 5 separate work packages (WP), each with designated goals and objectives. The project is finalized upon completion of all WP objectives. This report is a deliverable of work package 4, with the objective 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.” To some extent, establishment of national hubs was completed in some member states (MS) before OD4RD2 commenced, therefore the support provided from the hubs is viewed as the main activity during this phase of the project. This report aims to summarize the lessons learned during the course of OD4RD2 through the establishment and activities of the national hubs established during the project and tasked with supporting the implementation and use of

ORPHACodes. The lessons learned during the entirety of the project is summarized in this document.

-National Action Plans were devised by each national hub, outlining a path to support the national implementation of ORPHACodes. Each plan was constructed based on the unique state of play in each country (see [deliverable D4.1](#) for survey results and [deliverable D4.2](#) for content of National Action Plans) and the described activities tailored to support ORPHACode implementation in the current RD landscape of the country.

Several measures have been taken to facilitate and support the ORPHACode implementation process in the member countries. These measures include providing trainings for the national hubs, information material directed towards decision makers, institutions, and the public, and providing a national helpdesk for support. Each activity represents a major step on the path to achieving the wp4 project goals.

Method

Data collection

The lessons learned (LL) have been collected on several separate occasions during the OD4RD2 project, initially using a free text format. The key success factors and obstacles mentioned in the initial collections were compiled by the WP4 coordinators and used to create a survey on the identified topics which was distributed to the National Hubs to accumulate data on the relevant topics from all participating countries. In addition, statistics on issues were extracted from GitHub and analysed regarding the number of tickets, countries concerned, and flags used.

The survey was organized in sections mirroring main activities performed in the scope of the OD4RD2 project: State of Play, Activities, Trainings, Communication, Networking and Action Plans. A majority of the survey questions were close ended and aimed to collect an assessment of performed activities from each country. Data were collected and measured by a range of different metrics such as answering yes/no, contributing/not contributing to success, estimated effort/impact, agree/do not agree or multiple-choice questions. In addition, open ended questions were added for each section to ensure collection of any data which was missed by the close ended questions.

On survey questions related to the State of play in each country the national hubs were asked to answer both if a statement applied to the country (yes/no/unknown) and to estimate the impact (positive/neutral/negative/unknown) it has had on the implementation process in the country. For activities performed by the National Hubs the respondents were instead asked to estimate both the effort required and the impact achieved for selected activities. The activities selected for inclusion in the survey were those which had been previously reported as contributing to success/constituting an obstacle by one or more National Hubs and performed in the scope of the OD4RD2 project.

Trainings are considered a core activity of the OD4RD2 project and have been performed by all participating countries, and the topic was allocated considerable attention in the final LL

survey. Responses for in-person and on-line trainings were collected separately to allow for comparisons between the training methods while more general aspects did not make a distinction between the delivery method. For networking and communication, the National Hubs were asked to report which methods had been successfully employed during the duration of the project as well as for which target audience it was successful (Health care professionals, Patients, Policy makers, Health care decision makers).

Responses were obtained from 19 of the participating countries of WP4. Orphanet Sweden, in the role of Co-lead of WP4 in OD4RD2 performed the task of compiling the collected data. The compilation and analysis were performed using excel, as well as diagrams and charts produced in Tableau web application.

Participating countries

Austria, Belgium, Bulgaria, Czech Republic, Estonia, Finland, Germany, Ireland, Italy, Latvia, Lithuania, Norway, Poland, Portugal, Romania, Slovenia, Spain, Sweden, the Netherlands.

The coordinating team of the project and of the Orphanet Network is located at Inserm, France. On this account, France has not developed an Orphanet Nomenclature National Hub in the frame of the OD4RD project but has continuously provided supporting activities for the implementation of ORPHAcodes since implementation of the French 2 National Plan on RD, predating the beginning of OD4RD in 2022. Annual trainings are organised with the French RD National Reference Networks and an active helpdesk exist since 2019.

Result

State of play

Regulatory instruments

In previous LL data collections several state of play factors have been mentioned as either contributing to success or as an obstacle, depending on the state of play in the country. For instance, in relation to whether: ORPHAcoding of RD patients is mandatory, if ORPHAcodes use is incentivized by reimbursement, if ORPHAcodes are used in registries, if ORPHAcodes are used by ERNs with members in the country and if the country participates in other European projects.

A total of seven countries reported that ORPHAcodes have been made mandatory in their respective countries, with three countries indicating that making ORPHAcodes mandated contributed to success (Lithuania, Czech Republic, Poland). Among the twelve countries that indicated ORPHAcodes not being currently mandatory, five reported that this had a negative impact, (Spain, Bulgaria, Sweden, Belgium, Romania), and one country (The Netherlands) that reported this having had no effect. In total ten countries reported that the impact could not be estimated, four in which mandatory ORPHAcoding was in place (Austria, Germany, Slovenia, Latvia) and six where it was not (Estonia, Ireland, Norway, Italy, Finland, Portugal). (See figure 1 below).

That the use of ORPHAcodes is incentivized by affecting how reimbursement for health care services is calculated was reported in four countries, and two of these (Germany, Portugal) indicated that the impact of this incentive could not be estimated, while the other two

(Poland, Czech Republic) indicated that it had contributed to success. Two countries reported it to be unknown if ORPHAcodes did affect reimbursement (Lithuania, Latvia). In the remaining twelve countries, in which ORPHAcodes use is not incentivized by reimbursement, a majority (Austria, Estonia, Bulgaria, Ireland, Sweden, Norway, Slovenia, Finland) reported that the impact of an incentive could not be estimated, three countries (Spain, Belgium, Romania) reported that the lack of reimbursement incentives had had a negative effect, while two countries (The Netherlands, Italy) reported that it had no effect. (See figure 1 below.)

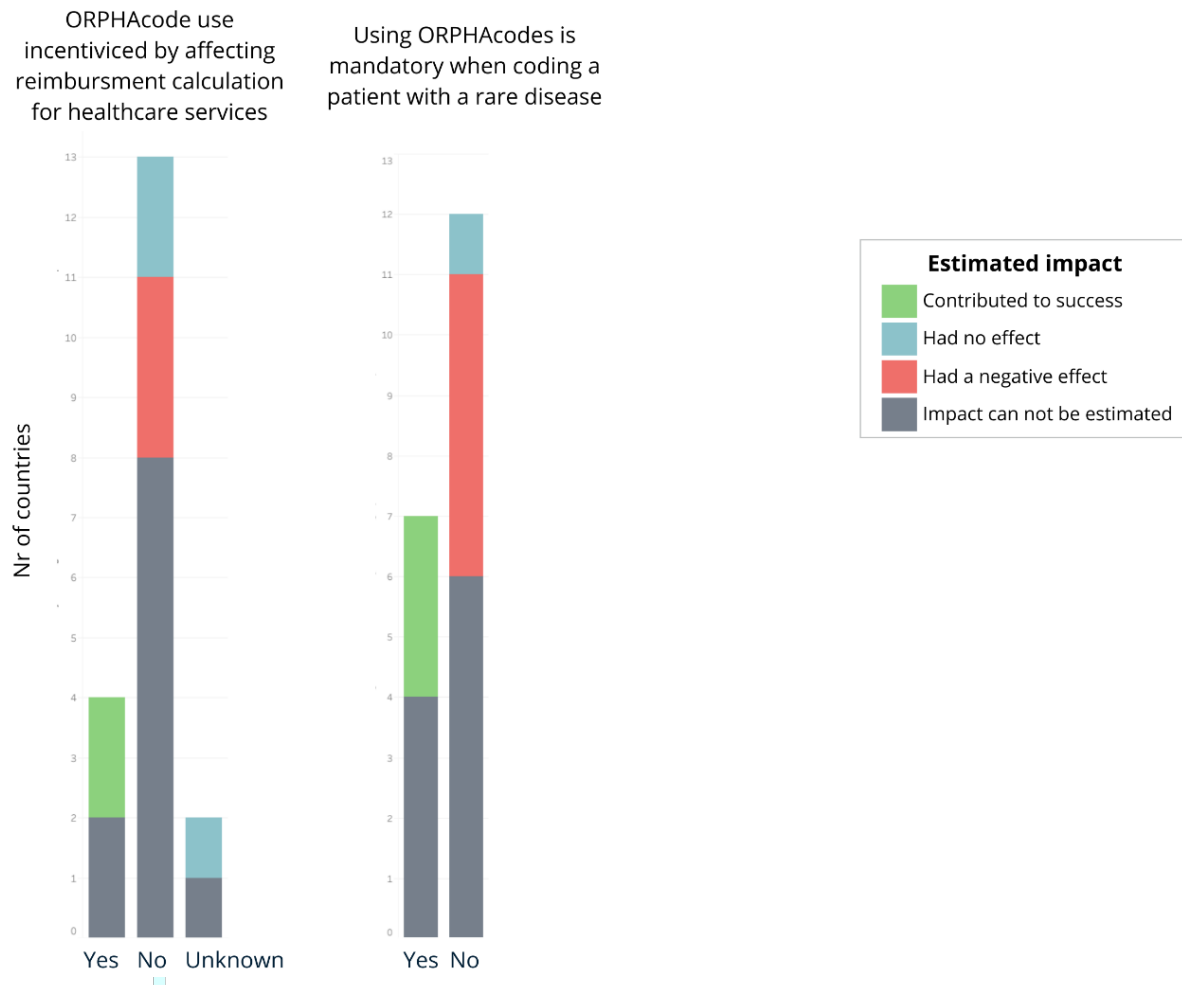


Figure 1) Regulatory instruments, perceived effect on ORPHAcodes implementation
 Two bar charts showing the distribution of responses on whether ORPHAcodes use is 1) incentivized by affecting reimbursement for health care services (left) or 2) mandatory for coding RD patients (right) (N=19).

Registries

Two questions addressed the use of ORPHAcodes in registries, one regarding national registries and the other regarding registries within the European Reference Networks (ERNs).

A majority of the countries (Austria, Estonia, Germany, Spain, Bulgaria, The Netherlands, Poland, Ireland, Belgium, Norway, Czech Republic, Lithuania, Italy, Latvia) participating in OD4RD2 contributes to ERN-registries in which ORPHAcodes are used. Half of these countries (Spain, Bulgaria, The Netherlands, Poland, Belgium, Czech Republic, Lithuania) estimated this had had a positive effect on ORPHAcode implementation, while the remaining were unable to estimate the impact. Similarly, fourteen countries (Estonia, Germany, Spain, Bulgaria, Poland, Sweden, Belgium, Norway, Czech Republic, Lithuania, Slovenia, Italy, Latvia, Finland) report that ORPHAcodes are used in national RD/ other registries, and eight of these (Spain, Bulgaria, Sweden, Belgium, Norway, Czech Republic, Lithuania, Finland) report this to have had a positive impact on ORPHAcode implementation. One country (Slovenia) reports this to have had no effect while the remaining countries thought an effect couldnot be estimated.

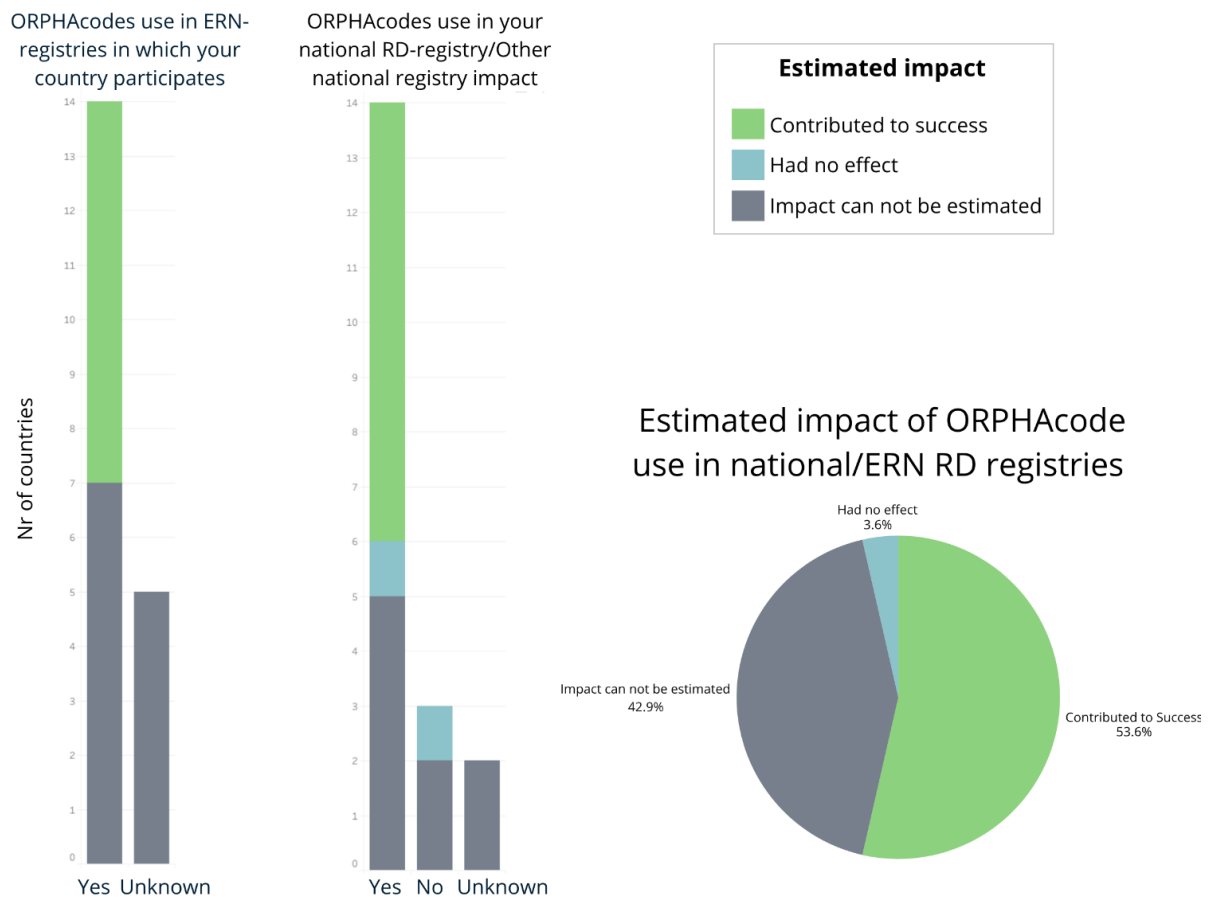


Figure 2) ORPHAcode use in registries, perceived effect on ORPHAcode implementation

The two bar charts show the distribution of responses on whether ORPHAcodes are used in 1) ERN-registries in which the country participates (left) or 2) national RD registry or other national registry (middle) (N=19). Circle diagram (right) visualizes the combined results from both “yes”-bar of the bar charts, concluding that countries in which ORPHAcodes are used in a registry a slight majority of the National Hubs perceived this to have a positive impact (N=28).

Participation in European endeavours

Data was also collected on participation in other European projects, for instance JARDIN, ERICA, et cetera. The results indicate that all countries participating in OD4RD2 are also engaged in other

international EU-funded initiatives and all are members in an ERN which uses ORPHAcodes. Fourteen of the responding countries (Austria, Germany, Spain, Bulgaria, The Netherlands, Poland, Ireland, Sweden, Belgium, Norway, Czech Republic, Lithuania, Romania, Finland) indicated that such participation has had a positive influence on the implementation of ORPHAcodes, while the remaining five reported the impact could not be estimated. Similarly, all countries reported the presence of members in an ERN actively using ORPHAcodes in the country. Twelve of these (Spain, Bulgaria, The Netherlands, Poland, Ireland, Sweden, Belgium, Czech Republic, Lithuania, Slovenia, Romania, Finland) reported this to have had a positive effect on ORPHAcodes implementation while the remaining countries could not estimate the impact.

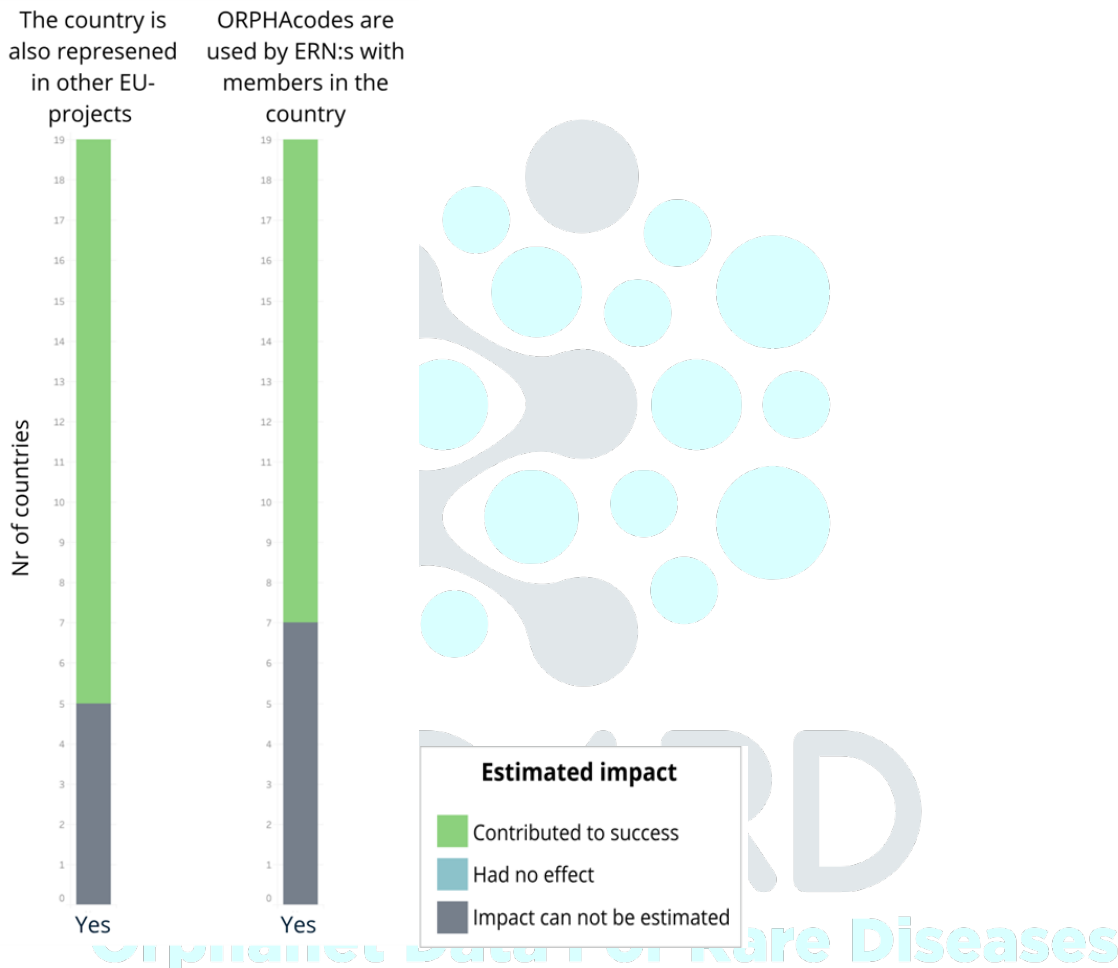


Figure 3) Participation in ERN:s and European projects, perceived effect on ORPHAcodes implementation Bar charts showing the distribution of responses on whether countries participating in the OD4RD2 project are 1) also contributing to other European projects in the field of RD (left) or 2) the country has ERN-members in an ERN which actively is using ORPHAcodes (right) (N=19).

Activities

Effort and impact

In the activities section of the Lessons Learned data collection the National Hubs were asked to assess both the level of effort required and the impact achieved for a range of activities conducted within the scope of the OD4RD2 project. These included activities spanning multiple facets of

ORPHAcodes implementation and included six questions on specific tasks related to trainings, communication, networking, and on-demand data services.

Data was collected on three aspects related to trainings: the impact and effort required to translate training materials, adapting training materials to new formats (such as recordings, e-courses, quizzes, et cetera), and performing previously prepared trainings. For performing previously prepared trainings, two countries (Estonia, Finland) reported them having a low impact, while also requiring a low effort to perform. Five countries estimated previously prepared trainings to have an intermediate impact, out of which three estimated the effort required to also be intermediate (Austria, Slovenia, Italy) while one country estimated the effort to be high (Belgium), and one estimated the effort to be low (Czech Republic). Eight countries reported the impact for this activity to be high of which half (Spain, Bulgaria, Norway, Portugal) estimated the effort to be intermediate, while two estimated the effort to be high (Germany, Poland) and two low (Ireland, Sweden). Only a small minority of the countries reported any of the training related activities to have had a low impact. Romania did not perform the activity and did not estimate the impact.

Adapting training material to new formats was reported as having low impact and requiring low effort by one country (Finland) and low impact and intermediate effort by one (Belgium). The latter possibly reflecting the multilingual situation in the nation. Nine countries estimated this activity to have an intermediate impact, out of which five (Poland, Czech Republic, Romania, Finland, Portugal) estimated the effort to also be intermediate while three (Austria, The Netherlands, Norway) estimated a high effort. No countries reported the activity to be low effort.

The remaining five countries estimated adaption of trainings to be an activity resulting in high impact, and two of those (Spain, Ireland) estimated the effort to be intermediate while three estimated the effort to be low (Germany, Bulgaria, Sweden). Two countries (Romania, Estonia) did not perform the activity and did not report the impact.

Translating training materials was performed by twelve participating countries, of which five (Austria, Bulgaria, Norway, Italy, Portugal) considered both the effort required and the impact achieved to be intermediate. One country (Belgium) estimated the effort required to be high and one (Czech Republic) the effort required to be low. Two countries estimated the effort to be intermediate but the impact to be high (Spain, Sweden) while one (Poland) estimated both impact and effort to be high.

Two countries estimated the impact of producing communication materials to be high, while one of these estimated the effort required to also be high (Germany) and one (Ireland) considered the effort required to be intermediate. Three countries (Austria, Poland, Czech Republic) estimated both effort and impact to be low for communication activities.

Ten countries estimated the impact of producing communication materials to be intermediate, and six of those estimated it to also require an intermediate effort to produce (Estonia, Bulgaria, Italy, Romania, Finland, Portugal) while two countries (Spain, Sweden) thought the effort was low, and two countries (Ireland, Belgium) considered the effort to be high. One country (Slovenia) did not perform the activity and did not estimate the effort while one (The Netherlands) did not perform the activity but still considered the effort to be intermediate had it been done. Two countries (Lithuania, Latvia) did not estimate the impact but considered the effort required to be intermediate and high, respectively.

Participating in networking activities was considered a high effort and high impact activity by eight countries (Austria, Estonia, Germany, Bulgaria, Poland, Ireland, Norway, Portugal). Two countries (Lithuania, Latvia) did not estimate the impact but considered the effort required to also be high. Six countries (Spain, The Netherlands, Sweden, Slovenia, Italy, Romania) estimated both the impact and effort required to be intermediate, while three countries (Belgium, Czech Republic, Finland) estimated the impact intermediate but the effort to be high. Compiled fourteen countries considered the impact and effort required to be balanced.

Providing tailored materials on request (such as lists of ORPHA codes, mappings, etc) was considered a high impact activity by seven countries, of which three (Austria, Poland, Ireland) estimated the effort required to be high, and the same number of countries (Bulgaria, Belgium, Portugal) estimated the effort to be intermediate. One country (Sweden) estimated the effort required to be low while impact was considered high.

Seven countries considered the activity to have an intermediate impact and five of those (Estonia, Spain, Norway, Czech Republic, Finland) considered the effort required to be intermediate as well, while two (The Netherlands, Romania) considered the effort to be low. Two countries (Latvia, Lithuania) estimated the impact to be high but did not estimate the effort required and one country (Italy) considered both impact and effort to be low.

The countries were evenly divided between the activity having a high and intermediate impact with seven respondents for each. The aggregated data on effort and impact required for selected activities is presented in the bubble chart below, see figure 4.

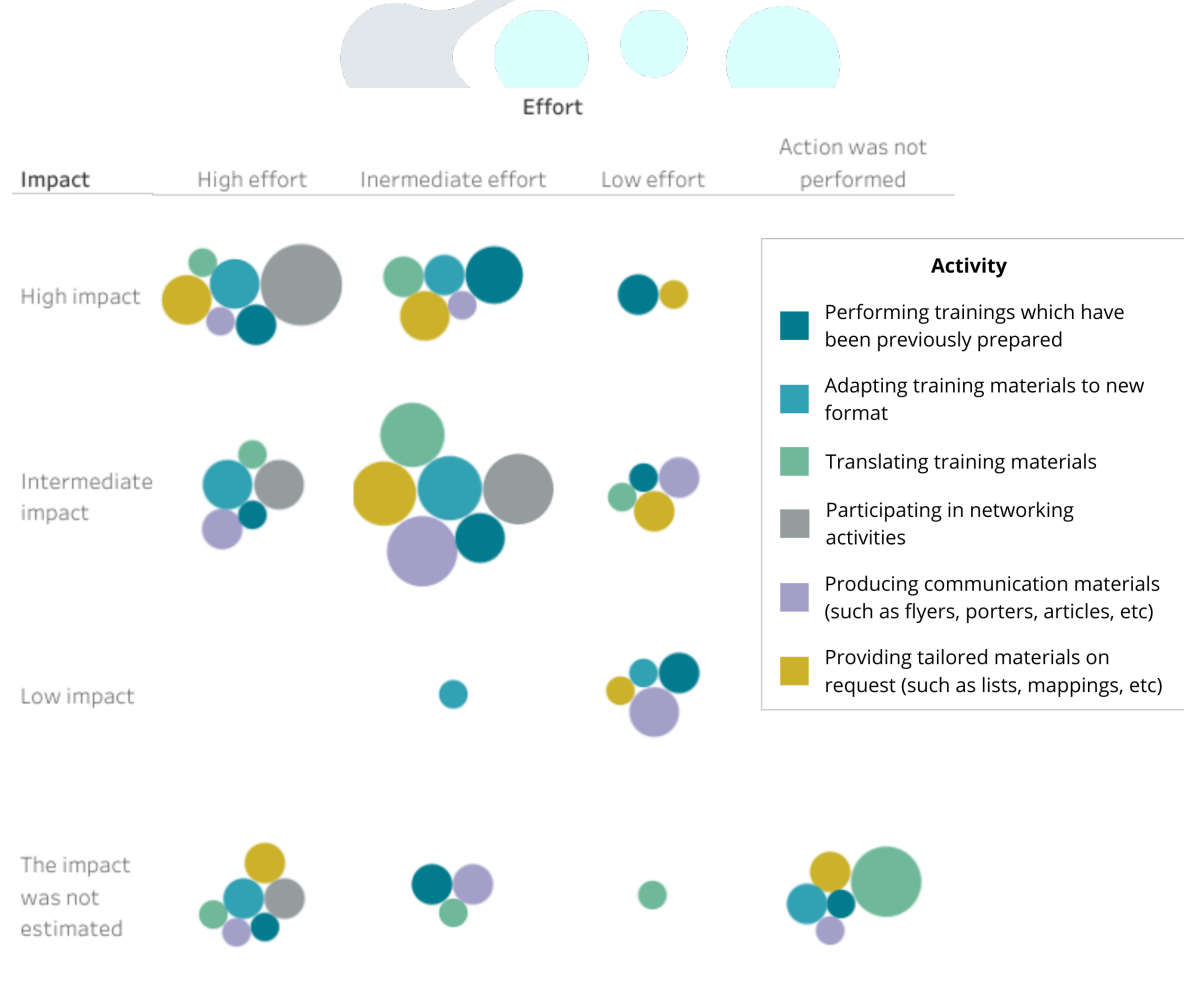


Figure 4) Estimation of impact and effort required for selected OD4RD2 activities.

Bubble chart showing the combined answers by all countries on the effort required and impact achieved for a selection of performed OD4RD2 activities. Each bubble represents a specific activity, with its colour indicating the category of activity performed and its size the number of responses.

Trainings

Training methods

In addition to the training-related questions included in the activities section, the LL data collection survey included further specific questions on training formats. Specifically, these questions were included to allow for comparison between in-person and on-line trainings and to identify key factors to success as well as obstacles to successful trainings performed by the National Hubs. The aggregated responses are presented in the bubble chart below (see figure 5.)

The results indicate that a majority of countries considered group size to have some influence on training effectiveness for both online and in-person trainings. For in-person trainings, fifteen countries agreed or somewhat agreed that group size affects the impact of training, while only twelve agreed or somewhat agreed when considering on-line trainings. The impact of group size was somewhat lower in online settings than in face-to-face sessions.

A majority of National Hubs agreed that targeting the training content by adapting materials and sessions to the specific needs of participants positively affected training outcomes, regardless of delivery method. For online trainings, thirteen countries completely agreed, whereas for in-person trainings, eleven countries shared this viewpoint.

Both online and in-person as delivery methods were considered to have added benefit by providing an opportunity to network with professionals. Even if this effect was reported to be more pronounced for in-person trainings (ten countries completely agreed, four somewhat agreed) than for on-line trainings (four countries completely agreed, nine countries somewhat agreed).

Seventeen of the National Hubs completely or somewhat agree that the resources required to organize an on-line training is less than for an in-person training, of the remaining countries one considers in-person events to be less resource intense while one did not answer. The results are mirrored in the answers received for in-person trainings.

The countries were also asked to estimate whether each type of training was a cost-effective measure, given the circumstances. A large majority of the National Hubs (eleven countries) completely agreed, while seven somewhat agreed that online trainings are a cost-effective measure for supporting ORPHAcodes implementation. In contrast, two countries completely agreed and six somewhat agreed that in-person trainings was a cost-effective activity.

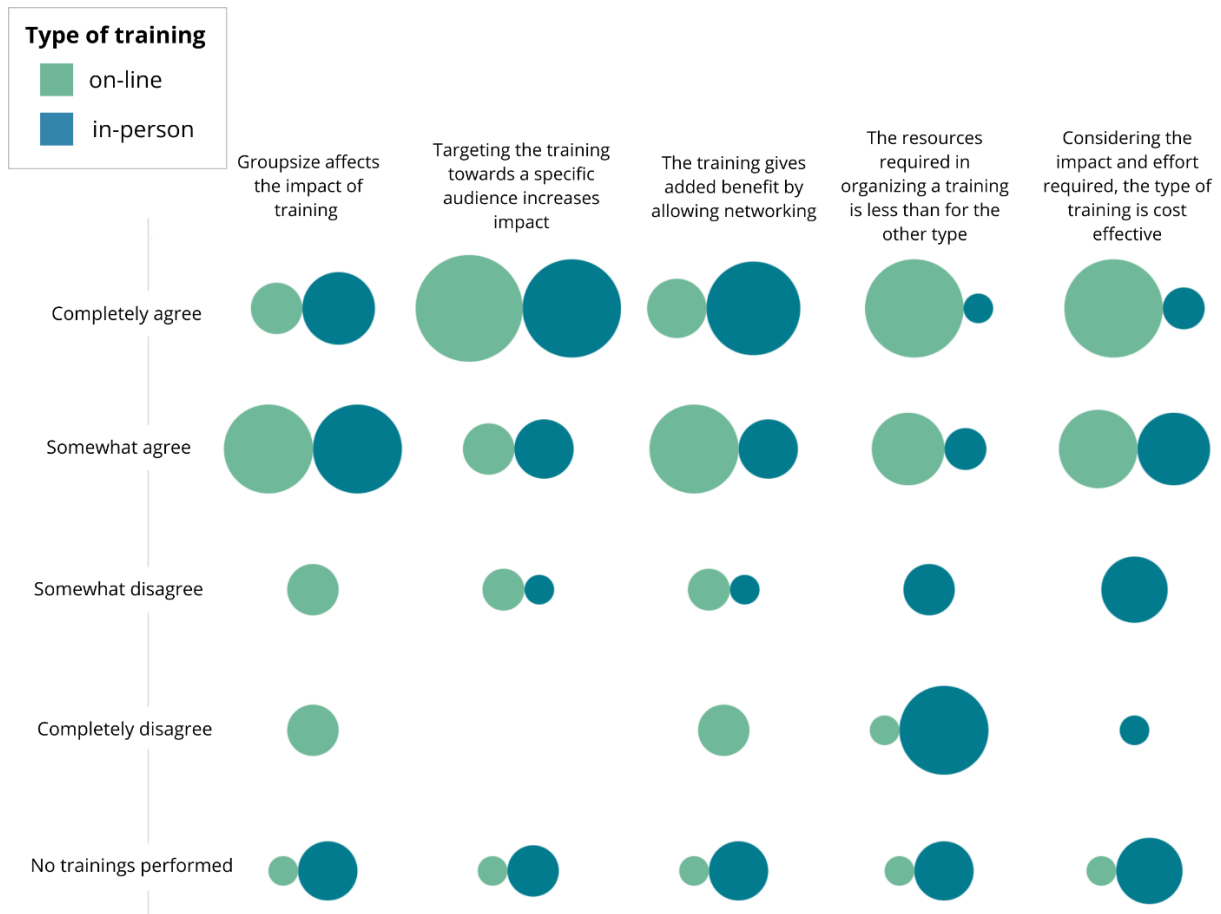


Figure 5) Evaluation of factors impacting training outcomes

Bubble chart showing the combined answers on the stance on selected statements regarding trainings and delivery methods from all National Hubs within the OD4RD2 project. In the chart the colour corresponds to a training format (light green for on-line and blue for in-person) while the size of the bubble reflects the number of countries providing a particular response. The x-axis displays statements concerning various aspects of training (e.g., accessibility, engagement, applicability), while the y-axis reflects the level of agreement expressed by the National Hubs.

Time required for training

The duration of the training sessions has been a topic of debate during the entirety of the OD4RD2 project. The time required for attending trainings has been reported as an obstacle to participation from clinicians while a too brief time limit would not allow for inclusion of all the needed information. The question was posed separately for in-person and on-line events, offering higher specificity than when the questions have been explored previously. In order to provide precise detail on the optimal duration of a training session, the countries were asked to select a time limit between 30 minutes and > 120 minutes or provide a time of their own. The compiled results including both in-person and on-line events are found in figure 6.

It can be concluded that a duration of one hour is the most popular choice for a training session, regardless of delivery method, twelve countries prefer a duration of 60 minutes for on-line events and nine countries also for events in-person. However, a trend of preferring a longer duration for in-

person events can be seen, as results indicate seventeen countries prefer a duration of 60 minutes (*or more*) while eleven countries prefer 60 minutes (*or less*) for in-person events.

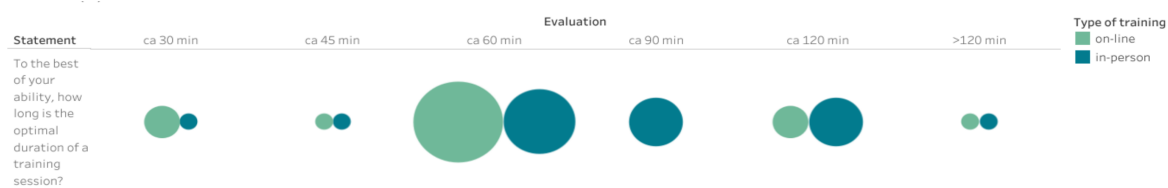


Figure 6) Estimation of optimal time required for trainings.

Bubble chart showing the combined answers from all countries on which is the optimal duration of a training session. In the chart the colour corresponds to a training format (light green for on-line and blue for in-person) while the size of the bubble reflects the number of countries providing a particular response. The x-axis displays suggested training durations between 30 min and more than 120 min, while the y-axis contains the posed question answered by the National Hubs.

The National Hubs were asked if the time required to participate in trainings was assessed to be an obstacle to professionals attending trainings, which had been reported in previous LL collections. All responding countries except one (Portugal) agreed that this was a barrier limiting professional attendance, confirming previous results. In addition, the National Hubs were presented a list containing suggestions on novel training materials and methods and asked to check those which they considered to contribute to mitigating this obstacle.

The aggregated responses are presented in the bubble chart below (*figure 7*). In this visualization, the colour of each bubble corresponds to the type of additional training material suggested, while the size of the bubble reflects the number of countries assessing the strategy to be viable. The method most anticipated to be successful was “developing on-line self-train materials (such as e-courses) available on demand” which was supported by fourteen countries (Spain, Bulgaria, The Netherlands, Poland, Ireland, Sweden, Belgium, Norway, Czech Republic, Slovenia, Italy, Romania, Finland, Portugal), closely followed by “Recording training materials and make them available on demand” supported by thirteen countries (Germany, Spain, Bulgaria, Poland, Ireland, Sweden, Belgium, Czech Republic, Lithuania, Italy, Romania, Finland, Portugal). In contrast, only a small minority of three countries (Estonia, Latvia, Poland) estimated developing classical paper-based training materials would be helpful. Additionally, one country (Austria) emphasized the value of a compiled collection of good practices, highlighting that access to curated examples and guidance could further support professional learning by allowing self-studies.



Figure 7) Viable training materials to mitigate the obstacle of required time for participation
 Circle Bubble chart visualizing National Hubs responses on training materials which would help mitigate the obstacle of time required to participate in trainings for professionals. One colour corresponds to one type of material and the size of the bubble to the number of countries which consider the material a viable strategy. The most popular strategy was Developing on-line self-train materials (such as e-courses) available on demand (N=14) followed by Recording training materials and make them available on demand (N=13), Developing classical (paper based) training materials for distribution (N=5) and having the collection of good practices (N=1).

Most successful training

All countries were asked which training method they considered to have been the most successful in their country. The responses were evenly distributed, with eight countries preferring classical in-person trainings (Estonia, Bulgaria, Ireland, Belgium, Norway, Czech Republic, Slovenia, Latvia) and nine preferring on-line trainings (Germany, Spain, Poland, Sweden, Lithuania, Italy, Romania, Finland, Portugal). One country (Austria) was not able to select the most successful method. (See figure 8)

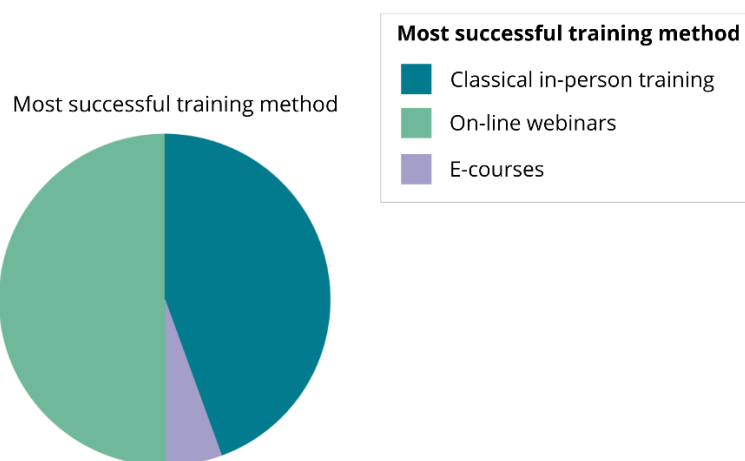


Figure 8) Most successfully employed training delivery method.

Circle diagram visualizing country responses on the question “Which training method do you think have been the most successful in your country?”. Nine countries considered on-line webinars to have been most successful while eight favoured classical in-person training and one developing E-course. (N=18)





Networking

Networking methods

The survey also examined which networking methods were perceived as successful for different target audiences. The results indicate that networking with professionals was the most consistently successful activity. Regardless of the method used, a large majority consisting of at least sixteen countries, reported having success when networking with professionals. In addition, each networking method included in the study was reported to have been successfully applied to all target populations by at least half of the participating countries during the project.



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Type of networking	Target				
	Contact type	HC Professionals	HC decision makers	Patient Organizations	Policy makers
 In-person meetings					
 E-mail correspondance					
 Attending events arranged by a different organization					
 Conference/direct calls					
	Attending events arranged by a different organization	16	10	15	13
	Conference/direct calls	16	11	12	13
	E-mail correspondance	19	15	12	10
	In-person meetings	19	13	11	14

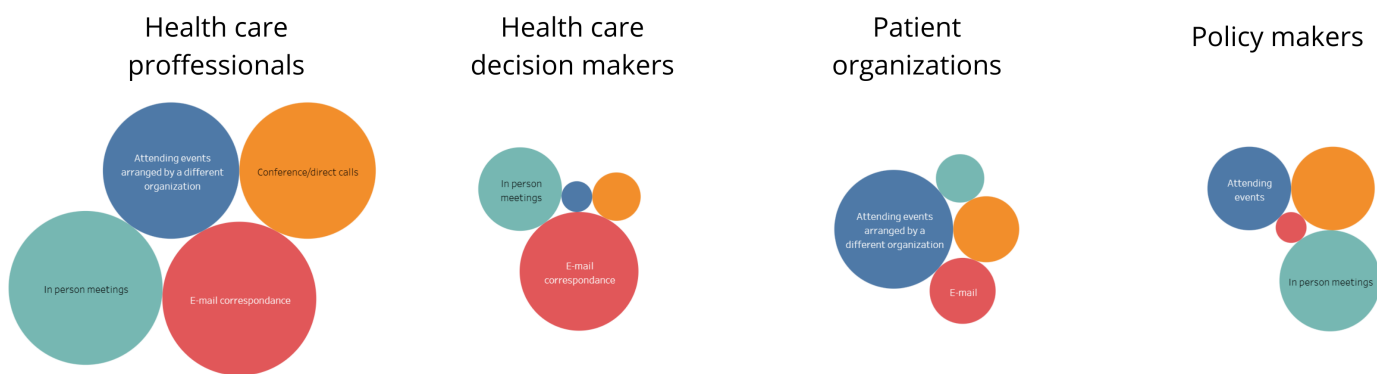


Figure 9) Successfully employed networking methods aimed at selected stake holders. Bubble chart visualizing NATIONAL HUBS responses on which networking methods had been successfully employed to reach intended target-organizations and/or professionals. The colour corresponds to one mode of contact while the size of the bubble corresponds to the number of National Hubs giving a specific response.

Communication

Communication materials

The survey examined which communication materials the National Hubs had successfully employed to increase awareness of RD and ORPHAcodes in selected target audiences. Web-based information campaigns were reported as the most successful form of communication overall, effectively reaching a diverse range of target audiences across participating countries, having been implemented by more than half of the National Hubs. The most successful targeting result was seen with Health care professionals, followed by patients, which was the most successful method employed to reach both of these groups. The second most successfully employed communication method was the production of tailored documents clarifying the need for ORPHAcodes. In contrast to the other communication methods, the most success for this material was reported with Health care decision makers closely followed by Policy makers, making targeted documents the most successful communication method for these target audiences.

Production of flyers was also a widely employed communication method and was used by more than half of the participating countries to reach professionals and patients. It was also used to reach policy

makers and Health care decision makers, although with less success. Scientific articles and posters were employed to a lesser extent, as was newsletters, news articles and non-scientific posters.

Across all communication methods, the most successful approach has been targeting Health care professionals. This mirrors the result from networking activities and illustrates the importance of ORPHAcodes in the clinical setting.

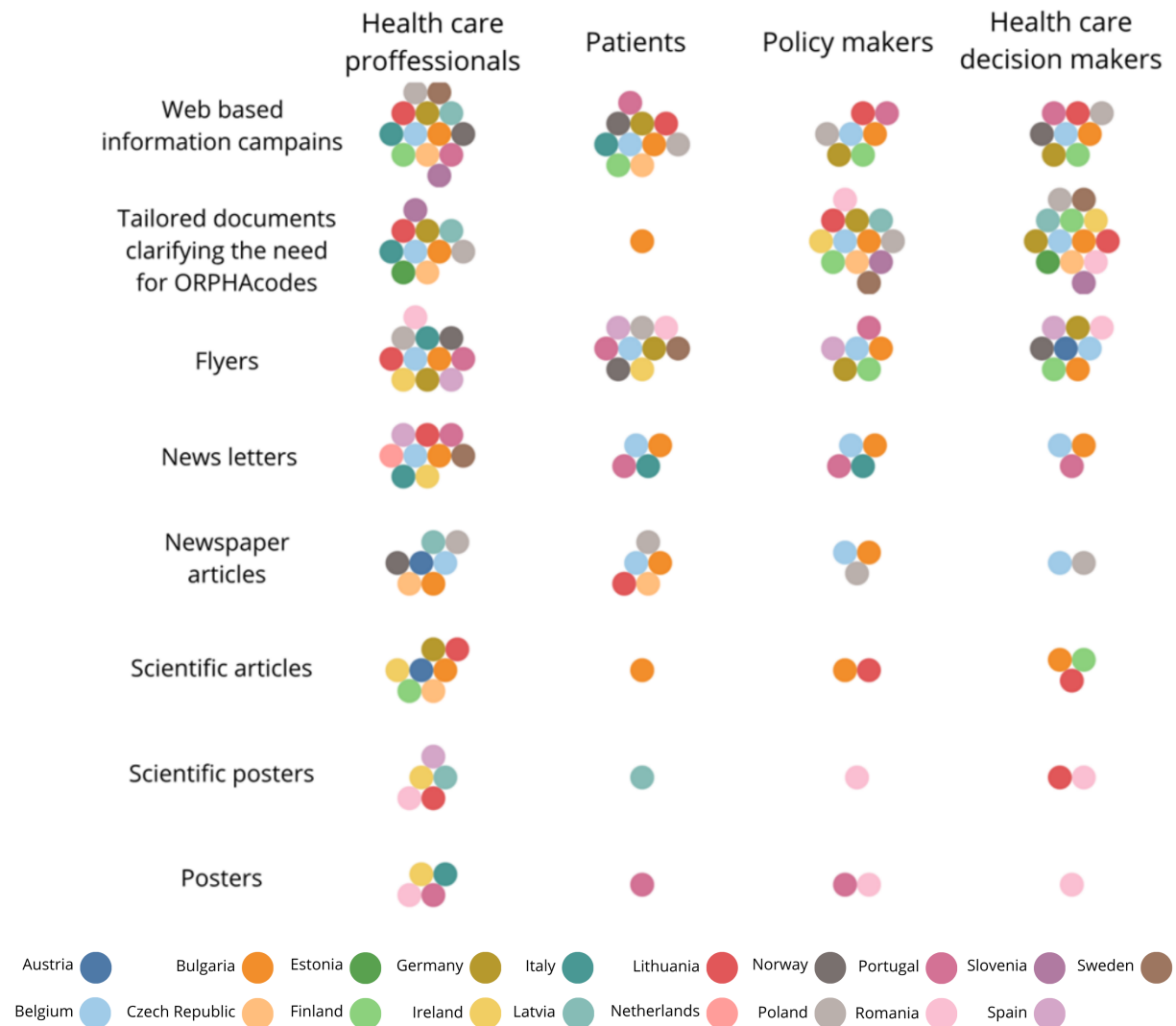


Figure 10) Successfully employed communication materials aimed at selected target audiences. Bubble chart visualizing National Hubs responses on which communication materials had been successfully employed to increase awareness of RD and ORPHAcodes in selected target audiences. Each bubble represents the response of a single country, with its colour corresponding to each specific country.

Additional successful communications

In addition to the communication materials specifically included in the survey, sixteen countries reported engaging in additional communication efforts to support the dissemination and implementation of ORPHAcodes. These additional initiatives included email campaigns, targeted meetings, presentations, and a variety of other activities aimed at engaging key stakeholders.

Action Plans

The survey included specific questions on the use of an action plan as a method to track national activities. Eighteen of the countries agreed that the use of Action Plans was a highly effective method for planning and structuring national activities related to ORPHAcode implementation. The Action Plans were regarded as a valuable tool for organizing tasks, prioritizing activities, and providing a framework to track country-level efforts. One country reported that adherence to the national implementation timeline took precedence over the schedule outlined in the action plan. This observation suggests that, while Action Plans offer a useful structure, flexibility is essential to accommodate country-specific circumstances and operational constraints.

Additional successful activities

Among the additional activities conducted during the project, several countries (Germany, The Netherlands, Norway) reported efforts focused on the integration and use of ORPHAcodes in electronic health record (EHR) systems. This approach reflects a strategic emphasis on embedding rare disease coding into routine clinical workflows, thereby promoting systematic data capture, interoperability, and long-term sustainability.

In addition, some countries highlighted the importance of collaborations with national stakeholders, including professional associations, healthcare institutions, and registries. These targeted partnerships were designed to enhance the adoption, acceptance, and practical utility of ORPHAcodes within existing healthcare infrastructures, ensuring that implementation is both feasible and effective.

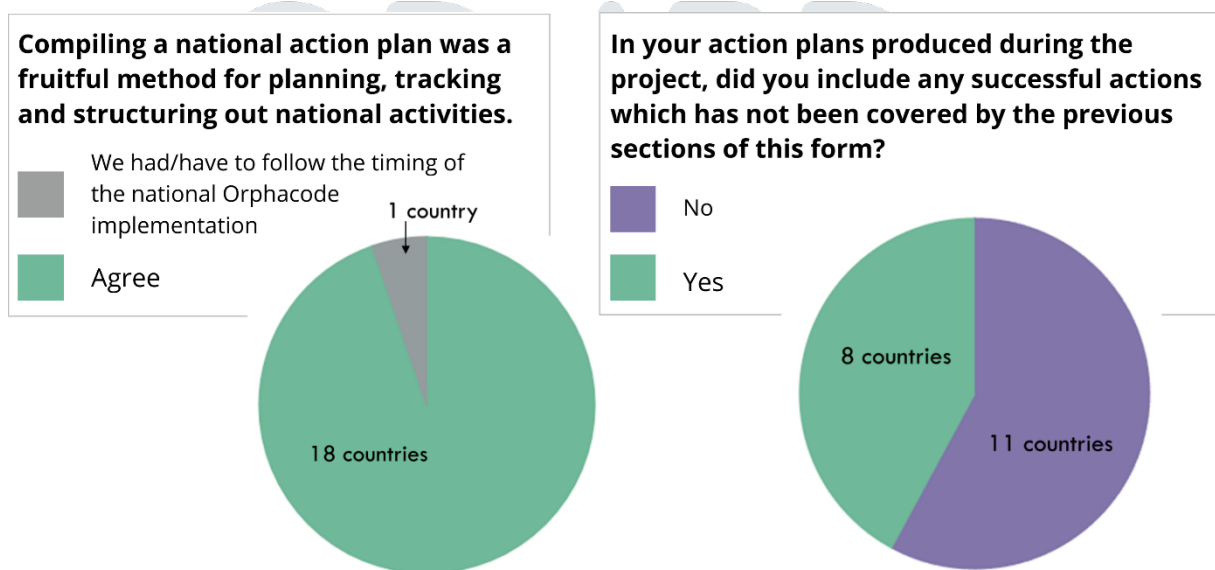


Figure 11) Action Plans use and activities

Left: Circle diagram visualizing National Hubs responses to the question “Compiling a national action plan was a fruitful method for planning, tracking and structuring our national activities” (N=19).

Right: Circle diagram visualizing National Hubs responses to the question “In your Action Plans produced during the project, did you include any successful actions which has not been covered by the previous sections of this form?” (N=19).

How would you improve?

In the survey's closing question, the National Hubs were invited to reflect on the question “If given the opportunity to redo the entire project, what would you do differently and why?”. Four respondents (Spain, Poland, Italy, Sweden) emphasized the importance of enhanced and closer collaboration with European Reference Networks (ERNs). Several countries (Spain, Norway, Romania) highlighted the need for earlier and increased engagement with policy makers and health care decision makers. A number of countries (Italy, Norway, Belgium, Ireland, Poland, Spain) also mentioned the need for policy decisions such as mandating ORPHAcodes use or connecting ORPHAcodes to reimbursement. Finally, several countries (Finland, Sweden, Slovenia, Norway, Czech Republic, Poland) mentioned the importance of supporting the technical implementation of ORPHAcodes into electronic health records (EHR) and health information systems (HIS) as well as the importance of collaboration with, and implementation in, registries (Sweden, Spain, Ireland, Norway, Portugal).

GitHub

The ORPHAcoding Main Helpdesk, accessible [here](#), is dedicated to responding to queries related to Orphanet nomenclature content and/or the implementation of ORPHAcodes in health information systems or other platforms. It serves as the official ORPHAcoding Helpdesk and is developed and maintained by Orphanet. In addition, a “[Questions and Answers](#)” section has been created on GitHub by the coordinating team based on users’ questions and reported issues. Its aim is to provide standardised and generalised answers for commonly raised topics such as: Orphanet nomenclature of rare diseases -ORPHAcodes, Why is an ORPHAcodes not available in my system?, Orphanet classification, Alignment with other terminologies, Good practice guidelines on Orphanet nomenclature, Orphanet tools for coders, Education and communication, Orphanet – ERN collaborations, ORPHAcodes technical implementation, Guidance documents for ORPHAcoding implementation and exploitation and why using the Orphanet nomenclature of RD (ORPHAcodes) to trace RD diagnosis.

Participation

The predominant share of the opened issues has been answered and closed, while a smaller number remains in-progress and are still open (see figure 12). The majority of the participating countries have been actively participating in the OD4RD2-Main Helpdesk found on GitHub (see figure 13), and the number of countries participating as well as the level of activity on the helpdesk has increased over time. The available labels have been used to flag the predominant share of the issues and the labels signal both which countries are concerned and the type of request described in the issue. The new number of issues being posted to the helpdesk has been steadily increasing for the duration of the project (see figure 14).

Type of requests

Across the timespan of the OD4RD2 project, creation requests were the most frequent type of demand on GitHub followed by nomenclature questions, cross-referencing issues and coding related issues. During the first year of the helpdesk, nomenclature requests were most common, followed by

cross-referencing and coding issues. The shift in type of request over time suggests that as implementation has progressed, the need for new ORPHAcodes have become more prominent.

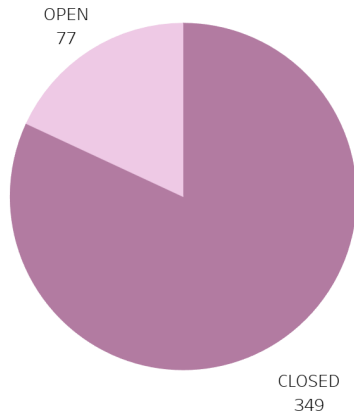


Figure 12) Status of posted GitHub issues

Circle diagram visualising the status of all issues posted in the OD4RD2 GitHub helpdesk.

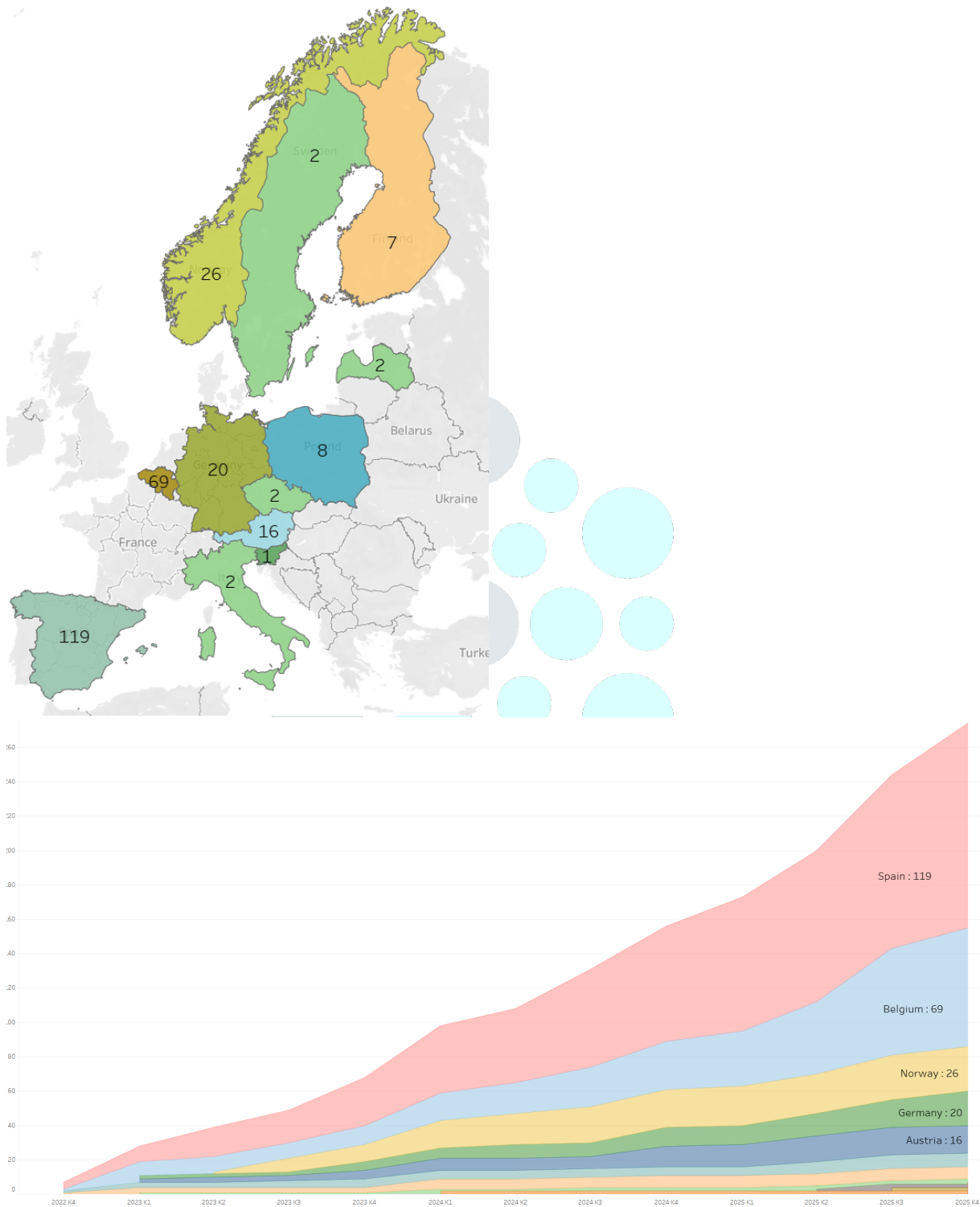


Figure 13) Number of GitHub tickets per country

Top: Map visualising the number of GitHub tickets flagged for each country in the OD4RD2-main helpdesk on GitHub. In total 426 GitHub issues had been posted of which 111 did not have a country flag. Bottom: Cumulative number of flagged issues per country in the OD4RD2-main helpdesk on GitHub.

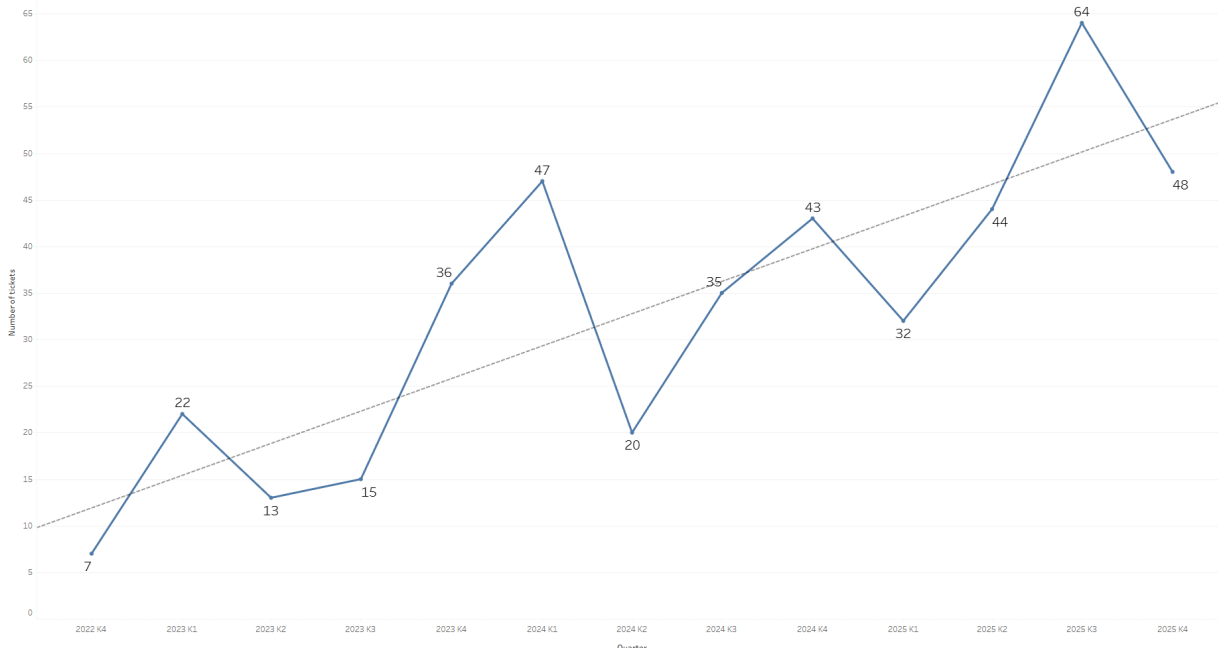
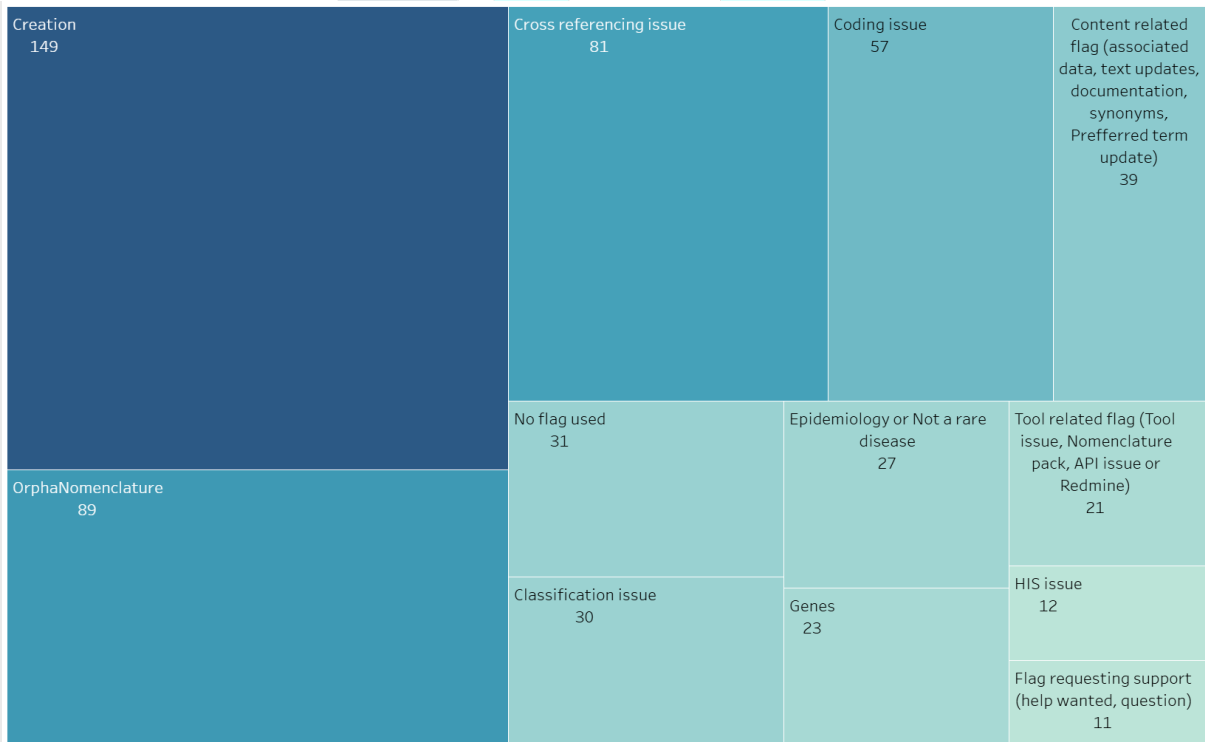


Figure 14) Number of new GitHub tickets per quarter
 Diagram visualising the number of novel issues posted on the OD4RD2 helpdesk on GitHub, per quarter (N=426), as well as the trend line indicating a steady increase in tickets for the duration of the project.



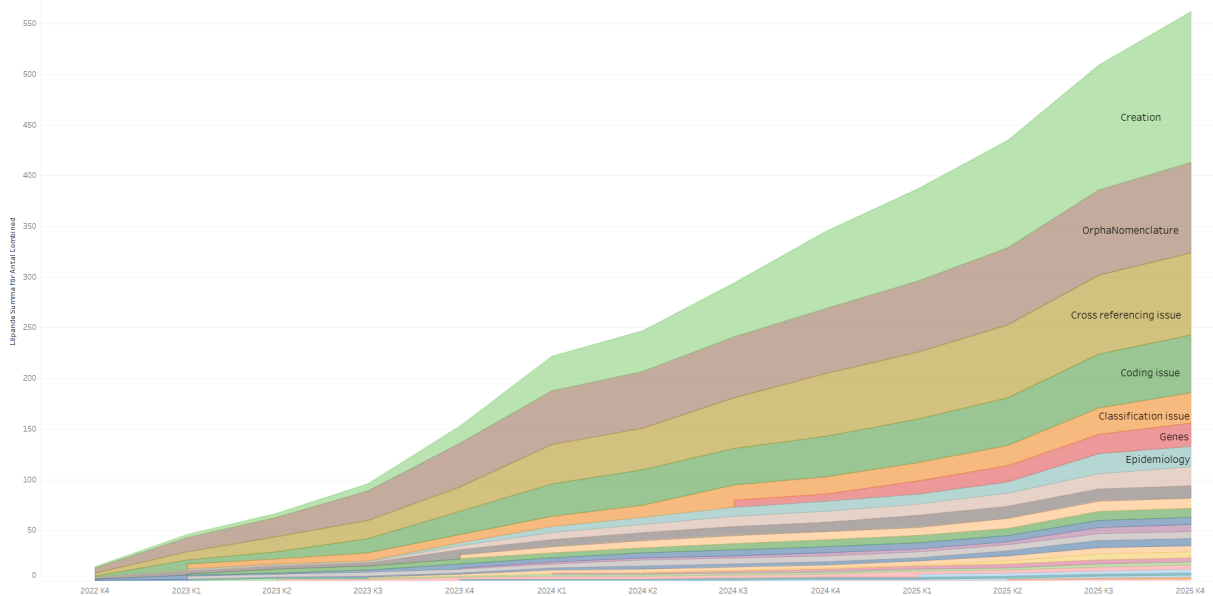


Figure 15) Distribution of labels on GitHub issues

Top: visualising the number of issues, per label, posted on the OD4RD2 helpdesk on GitHub. Labels used on less than 10 issues were excluded and similar labels were grouped. Bottom: Cumulative number of labels used for issues in the OD4RD2-main helpdesk on GitHub. The growing number of country-specific labels over time suggests that GitHub usage increased, and more countries became active in the GitHub helpdesk.

Conclusions

State of play

Reimbursement based incentives for ORPHAcodes use is uncommon among the participating countries. Five countries reported that linking reimbursement incentives to ORPHAcodes use had a positive effect on ORPHAcodes implementation. Three countries estimated that incentives through reimbursement have no effect while the majority consisting of eleven countries, were unable to estimate impact. These results demonstrate the uncertainty surrounding effectiveness of reimbursement incentives to promote national ORPHAcodes use. Reimbursement incentives may still be a positive contributing factor in promoting ORPHAcodes implementation, but it needs to be further explored in order to fully determine its effectiveness.

The effect of mandatory ORPHAcodes requirements is still somewhat unclear in regard to national impact, regardless if ORPHAcodes are nationally mandated or not. For countries with a ORPHAcodes mandate in place three out of seven reported this as beneficial. Countries without a mandate show similar numbers and five out of twelve countries reported the absence of a mandate as a hinder in implementation, and the same number reported uncertainty in regard to mandate impact on ORPHAcodes implementation. In total more than half of national hubs were unable to determine if mandating use of ORPHAcodes had an effect on implementation. This may to some extent mirror the differing state of play in the countries, it can be speculated that mandating ORPHAcodes use would have a larger impact in countries where the adoption of ORPHAcodes is not widely spread since it would be an incentive for ORPHAcodes implementation in electronic health care records and patient management systems in order to meet the regulatory obligations. If such support systems are already in place the mandatory status of ORPHAcodes use likely has a lesser impact.

From the collective results, the use of ORPHAcodes in registries provides support to ORPHAcodes implementation. Slightly more than half of the countries in which ORPHAcodes are in use in registries reported a positive effect from this fact. However, a large number of countries were unable to assess the impact, indicating that, similarly to reimbursement incentives and mandated ORPHAcodes use the contribution of use in registries is uncertain. However, the considerable number of countries experiencing a positive effect from use in registries cannot be disregarded and it can be tentatively concluded that registries probably have a supporting role in implementation.

When considering National Hubs engagement in European projects and ERNs that use ORPHAcodes, the results suggest that this supports national implementation efforts. The majority of the National Hubs reported positive effects both from involvement in these international initiatives and from having members in ERNs which use ORPHAcodes. This highlights the role of the ERNs as important collaborators and underscores the added value which arises from synergies with other European projects. ORPHAcodes constitute an important part of the rare disease landscape in Europe.

Activities

The activities part of the survey where the National Hubs were asked to estimate the impact and effort required for performing activities in the scope of the OD4RD2 project was included to evaluate if the included activities had an impact which was proportionate to the effort required in performing them. Evaluating which activities were most costly for the National Hubs as well as the results achieved by them can assist in prioritizing which activities to include, expand and discontinue moving forward. The selected actions spanned several core activities of the project, such as trainings,

networking, and communication in order to provide additional information on the real-world performance of said activities. For trainings the questions included both measures likely entailing a one-time investment of effort of which the result can be reused, such as adapting training material to new format, translating training materials, and tasks for which the effort required is recurring each time the activity is carried out, such as performing a previously prepared training. The latter is likely also the case for networking activities, producing communication materials, and providing tailored materials on request.

The results generally show a balance between the effort required and the impact achieved within the OD4RD2 activities. For most activities, the majority of responses indicated a high-impact and high-effort or intermediate-impact and intermediate-effort result, however a general estimate of the effort required for each activity of the impact cannot be identified. However, what can be observed is that the results indicate that all activities had an impact related to the invested effort. Only a minority of participating countries expressed any of the activities as having a low impact and those who did simultaneously reported having invested a low effort in the same activity. An exception to this was reported by one National Hub (Belgium), which evaluated the adaptation of training materials as requiring an intermediate level of effort but resulting in relatively low impact. This may reflect the fact that Belgium is a multilingual country and that as a direct effect of this all performed translation, and adaptations need to be performed multiple times.

Trainings

Trainings aimed at professionals using ORPHAcodes is a core activity of the OD4RD2 project which is mainly performed by the National Hubs. This allows the training to be delivered in the local language and adapted to the local needs. Both on-line and in-person trainings have been held on a large number of occasions across Europe. Data was then collected in order to evaluate the performance of on-line versus in-person trainings and determine how specific factors changed the effectiveness of the training sessions, and if this differed due to delivery method. The results reveal that group size affects both in-person and on-line trainings although the latter to a lesser extent. Tailoring the training for the specific audience contributes to its effectiveness and is beneficial regardless of delivery format. While online and in-person sessions were both noted as promoting professional connections by being an opportunity to network, this was seen as more effective for in-person settings. However online trainings were frequently viewed as more resource efficient and cost effective. From the results we can draw the conclusion that online trainings with tailored content and time requirement are most effective to reach large audiences, but in-person trainings foster better opportunity for networking and establishing connections with clinicians. Considering the added value of in-person events we conclude both delivery methods play important roles in the continued training efforts. The National Hubs were asked to estimate, for each training method, how long the optimal duration was for a training session. Most National Hubs indicated the optimal training duration for on-line trainings to be 60 minutes or less, while for in-person trainings the optimal training time is to be 60 minutes or more. This could be because audiences are more likely to allocate a shorter time to attend an online training while for example at work but need more time for travelling to a live event. The time required to participate in trainings has previously been defined as a significant barrier for professionals to attend trainings and this was confirmed in the current survey where seventeen countries agreed that this was indeed an obstacle. These results emphasize the need for flexible training formats available on demand that allows for inclusion in the busy environment of day-to-day clinical practice. Among the strategies suggested to minimize the effect of this obstacle developing on-line self-train materials such as e-courses was the most popular. This strategy has been planned or tried and implemented by at least two countries and should be further

explored in the future. The second most popular choice was making performed trainings available on demand by recording. Both these measures represent a high initial effort when producing the material, but the formats offer a significant reusability, leading to progressively higher returns over time.

Networking

The analysis of networking activities within the OD4RD2 project indicates that engagement with professionals was the most consistently successful networking activity. Nearly all participating countries reported positive outcomes for engaging this group, regardless of which networking method had been employed. Additionally, all networking methods included in the project were successfully applied to multiple target populations by at least half of the countries, demonstrating their effectiveness across different national settings. These findings suggest that a range of networking approaches can be successfully employed to connect with stakeholder groups.

These findings underscore the success of networking activities in fostering collaboration, facilitating knowledge exchange, and effective ORPHAcode implementation support. They also suggest that maintaining diverse networking strategies is a viable path when moving forward. Measures to ensure that networking continues to be a cornerstone in the endeavour to successfully implement ORPHAcodes.

Communication

The results evaluating communication strategies employed by the National Hubs indicate that web-based campaigns were the most effective and widely adopted approach for reaching diverse target audiences across participating countries. There was one exception, for reaching policy makers and health care decision makers tailored materials addressing the benefits and need for using ORPHAcodes were particularly effective. This highlights the value of content designed specifically to meet the needs and interests of selected stakeholder groups for whom more general approaches demonstrated limited usability. Similarly to the results on networking activities communication directed at professionals was the most successful activity, reflecting their significant role in the dissemination and practical application of ORPHAcodes.

The comparatively lower successful impact of scientific content suggests that highly specialized materials may have been used to a lesser extent or had a more limited reach. Taken together, these findings underscore that the strategic use of digital and flexible platforms, is essential for maximizing the reach, engagement, and effectiveness of ORPHAcode communication strategies while designed content targeting the specific needs of certain audiences still has its role to play. These insights provide guidance for the design of future communication initiatives, and ensuring both broad accessibility and relevance for key stakeholders remains a priority in future endeavours.

A large number of countries reported having used complementing communication materials and outreach activities, such as email campaigns, presentations, and targeted meetings. This adaptive multi-channel approach highlights the strength of having National Hubs which can be tuned in to the needs and opportunities to communicate at the local level. Overall, the results suggest that combining broad communication efforts with direct, tailored interactions adapted to local conditions can support more effective dissemination and implementation of ORPHAcodes across national settings.

The results indicate that scientific content was either used successfully to a lesser extent than more general informational materials or that it was less widely employed.

Action Plans

The use of Action Plans was widely recognized as an effective approach for planning and structuring national activities. The Action Plans offered a clear framework for prioritizing tasks, organizing activities, and ensuring progress across national activities. Eighteen countries agreed the use of an Action plan was a valuable tool while the remaining country experienced their activities were steered primarily by the national ORPHAcodes adoption timeline. The use of Action Plans allowed for incorporation and tracking of country-specific activities, such as collaboration with nationally important stakeholders and specific activities beyond trainings, networking, and communication needed in the country specific context. In addition to the core activities, trainings, networking, and communication, performed by all participating National Hubs eight countries reported that their Action Plans also included other activities. By integrating these additional measures, countries were able to address unique national challenges, enhance the reach and effectiveness of their initiatives, and strengthen the overall implementation process. The versatility of the Action Plans underscores the flexibility in the methodology. This adaptability allowed countries to respond to local priorities and address their unique local challenges. The use of Action Plans is a flexible instrument which accommodates diverse national contexts.

Since Action Plans have proven effective in structuring national activities, future projects should retain their use to ensure flexibility and to continue to accommodate country-specific timelines, priorities, and local initiatives. The adaptability allows countries to address unique challenges and capitalize on opportunities as they arise, enhancing effectiveness of the National Hubs activities.

How would you improve?

Reflections on areas for improvement highlighted the importance of closer engagement with European Reference Networks (ERNs), more strategic involvement of decision-makers and healthcare managers as well as continued emphasis on training and sustained support for the technical integration of ORPHAcodes into EHR/HIS systems. These observations emphasize that successful implementation depends not only on completing project tasks, but also on engaging with stakeholders and ensuring appropriate support. Addressing these areas would reinforce the advances gained so far in multiple dimensions of ORPHAcodes implementation, enhancing its effectiveness and long-term impact across participating countries.

In conclusion, the lessons learned from the OD4RD2 project demonstrate that a combination of structured planning, targeted and adaptable training, effective communication, networking, and flexible implementation strategies is critical for successful support to ORPHAcodes adoption. The lessons learned offer a framework for future initiatives and demonstrate a successful balancing of effort and impact for performed activities. In addition, they stress the importance of tailoring activities to stakeholder needs and maintaining flexibility in order to adapt to local needs in order to ensure sustainable integration of ORPHAcodes into national healthcare systems.

The responses also underscored the value of an early and sustained focus on training activities. This includes not only developing and adapting training materials to local contexts but also prioritizing the training-of-trainers program, which ensures the National Hubs capacity to provide wide and effective dissemination of knowledge to the professional community.

GitHub

The increased participation in the use of GitHub can be observed, as issues in GitHub labelled with additional countries appear progressively over the course of the project, indicating that more countries have become active contributors. The great variability in the number of issues belonging to

each country may also reflect the different state of play between the countries participating in the project. It appears likely that National Hubs in countries that have implemented ORPHAcodes to a greater extent will receive a larger number of questions from the larger number of users. We also know from previous LL and indicator collections that some countries are primarily contacted by phone or e-mail, which will contribute to a lesser number of issues submitted.

When looking at the number of new tickets posted on the GitHub help desk, a clear trend can be observed, which is that the number of novel issues increases over the course of the project. This trend of a rising number of novel issues indicates that activity on the platform has intensified and reflects the growing adoption of GitHub over the course of the project.

The most common requests over all were creation requests, which by far outnumbered the second and third most frequent requests, which were questions on nomenclature and terminology alignments issues. In the initial phases of the project, this was not the case. For the first years that the helpdesk was active, nomenclature requests were the most frequent, closely followed by terminology alignments issues and coding issues. In Q4 of 2024, creation requests became the most frequent demand, and its lead has since continued to increase. Creation requests becoming the leading type of request may reflect maturity in the process of ORPHAcode implementation in a number of participating countries. This highlights that the identification of rare diseases eligible for a designated ORPHAcode may increase as ORPHAcodes are more widely adopted and used in diverse clinical settings. This pattern indicates the potential to further evolve, improve and refine the nomenclature, as widespread use within diverse clinical fields provides opportunities for refinement and improvement. This may provide a complement to the evolutions performed in collaboration with the ERN: s and allows insight in to how the ORPHAcodes are used in clinical practice in diverse medical fields and health care settings across Europe.



Moving forward

The projects previously launched and completed in Europe to improve the quality of health and life situations of rare disease patients provide a solid foundation on which the continued work can build. The experiences and knowledge gained from RD-Action, RD-CODE, OD4RD and OD4RD2 ensure future activities can be directed where they are most necessary and ensure they are aligned with current real-world needs. The combined power between JARDIN and the OD4RD project working together has facilitated the identification of national authorities and hospital managers needs, which has enabled us to demonstrate the benefit of primary ORPHAcoding by comparing approaches in different settings. The progress achieved so far includes the full journey from development of coding guidance and standards shared across Europe to the full implementation of ORPHAcodes and their routine use in the everyday clinical setting. Moving forward, future projects must focus on expanding implementation across additional countries as well as strengthening long-term sustainability.

Future initiatives should place a strong emphasis on stakeholder engagement, encompassing European Reference Networks, policy makers, healthcare decision-makers, and institutional managers. Early and continuous involvement of these groups is critical to ensure alignment with national priorities, foster broad support and facilitate successful ORPHAcoding implementation.

The integration of ORPHAcodes into electronic health records and health information systems remains a critical component and providing countries with technical guidance, tools, and support will contribute to supporting ORPHAcoding implementation.

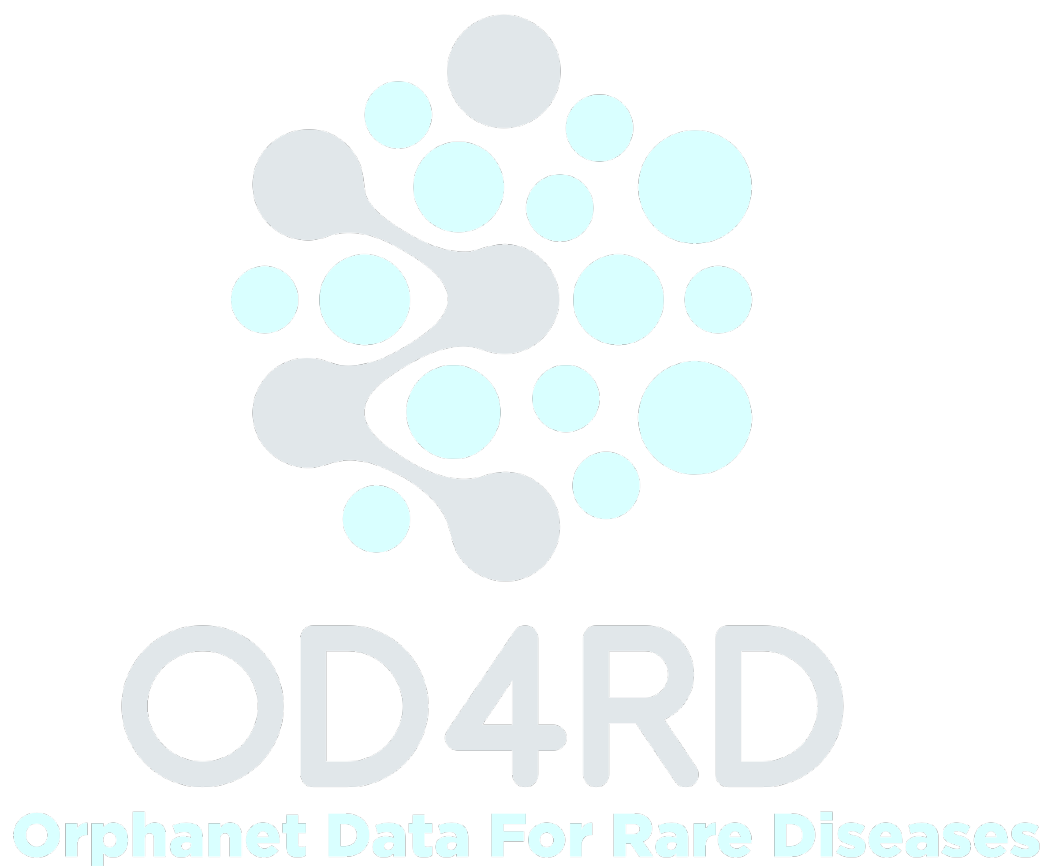
Building on the lessons learned, forthcoming efforts can prioritize supporting healthcare providers in the practical implementation of ORPHAcodes, enhancing data quality by training efforts, and ensuring that the Orphanet nomenclature continues to evolve in step with clinical practice. A key priority is stronger and closer engagement with European Reference Networks. Their expertise is invaluable in the effort to keep ORPHAcodes in line with the most recent scientific findings as well as for disseminating consensus on best clinical and coding practices. Leveraging the solid foundation already established will make it possible to accelerate progress on implementation, continue to promote harmonised RD data collection across Europe, and ultimately improve diagnosis, research, and care for rare disease patients.

Training activities remain crucial for ensuring homogenous implementation and use of ORPHAcodes within Europe. Trainings should continue to be a vital component of implementation strategies and focus on early and continuous capacity-building. Both online and in-person formats have been performed successfully and continue to play a significant role. Online trainings offer a cost-effective alternative to in-person trainings while still allowing for interaction and networking, however in-person trainings still have its advantages. At the same time, there is a clear need to develop novel training materials which can be made available on-demand. Suggestions on such materials are e-learning modules, recordings of classical training sessions, and other digital resources. In addition, developing and adapting training materials to suit local contexts have been identified as a success factor.

Limited availability of time to attend in-person training sessions has consistently been identified as a major barrier for participation by clinicians and this barrier also applies to on-line trainings although to a lesser extent. By creating access to flexible, on-demand training options, future efforts can reach a broader audience, accommodate varying schedules, and ensure that all users have the opportunity to enhance their skills and knowledge of ORPHAcodes. Development of flexible, digital resources as a

complement to classical training efforts can help participants overcome time constraints and make the content available to a larger audience.

Networking and communication efforts have been proven successful in promoting awareness and understanding of ORPHAcodes among both professionals, patients, and decision makers. Continuing such activities is essential to further disseminate information and maintain momentum in the adoption of ORPHAcodes. Sustained networking and communication will ensure that stakeholders remain informed and updated on RD developments in Europe, ultimately supporting the broader goal of harmonised rare disease data collection across Europe.



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