Deliverable 4.3
OD4RD Lessons learned
March 2023

This document represents deliverable 4.3 of the OD4RD project, which has received funding from the European Union. It contains the lessons learned by the members of OD4RD work package 4. The document has been produced by the leaders of the OD4RD work package 4. The OD4RD project has been launched in January 2022 for a 12 months period, and has been extended until March 31st 2023.

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

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Content summary

1. Context

There are currently an estimated 30 million people in the European Union who suffer from one of the more than 6,000 different rare diseases (RD). A large proportion of these diseases are chronic conditions that often manifest in childhood, are often diagnosed very late and are often associated with lifelong impairment of those affected, therefore advances in diagnostics and therapy in the field of RD are particularly important.

Since only a few patients suffer from each particular RD, the critical amount of data needed to improve knowledge and action can only be achieved through cumulative data collection in different countries in a standardized way.

In order to be able to generate basic epidemiological data on diseases, one falls back on the codes of the coding systems used in the respective countries. However, this is only successful if the diseases under consideration have specific codes in these common coding systems, which applies to RD only in very few cases. Therefore, additional appropriate identifiers for coding RD become necessary. The recommendation of the Council of the European Union of 8.6.2009 for action in the field of RD states: "An appropriate classification and codification of all RD is necessary in order to give them the necessary visibility and recognition in national health systems." (Official Journal of the European Union, 2009/C 151/02). In 2014, the Commission Expert Group on Rare Diseases listed measures to improve the coding of RD and recommended the use of ORPHAcodes to specifically code all RD. Furthermore, ORPHAcodes were selected as a best practice by the EC in 2017 (https://webgate.ec.europa.eu/dyna/bp-portal/transfered).

The RD-Action project developed guidelines and recommendations for the introduction of ORPHAcodes into national coding systems and defined a level of granularity suitable for merging RD data at the European level to reach a critical mass of data for RD knowledge. These guidelines and

recommendations were applied in the follow-up project RD-CODE in the introduction of ORPHAcodes into the national coding systems of four European countries, and further refined according to the real-life experience gained from the follow-up of the implementation.

The current OD4RD project aims to advance the implementation of ORPHAcodes in hospitals hosting the European Reference Networks (ERNs) of numerous European countries and to use the expertise of the ERNs to further improve the Orphanet nomenclature.

The OD4RD project builds 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 standardized, interoperable data on RD diagnosis for primary and secondary use, by the maintenance and the support to the implementation of the Orphanet nomenclature of RD.

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

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

2. Objective of OD4RD work package 4

The objective of Work Package 4 (WP4) is to ensure support for the local implementation of ORPHAcodes in national HCPs hosting ERNs and national HCPs linked to ERNs by establishing Orphanet national nomenclature support hubs in order to facilitate ORPHAcodes implementation (from a technical point of view) and use (from a coding and data exploitation point of view).

A survey among participating countries at the beginning of the project revealed that the overall picture of RD coding with ORPHAcodes has been very different among WP4 participating countries (deliverable 4.1, https://od4rd.eu/user/pages/04.03-deliverables/D4.1_OD4RD_State-of-play-survey.pdf). 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.

Taking into consideration the different situations and needs of end users in regard to ORPHAcodes implementation in the WP4 participating countries, it has been necessary to develop individual

national action plans to achieve the project goals. These action plans focus on providing training sessions in local language for coders, hospital information managers, statistical services and other stakeholders involved, as well as clinicians in ERN centers. Furthermore, the hubs' national action plans focus on actively participating in advocating ORPHAcodes towards national decision-makers (e.g. Ministry of Health, Directorate of Health, hospital managers, etc.) making use of the promotional material provided by the Orphanet coordination and adapting or translating it when needed. And finally, on establishing a helpdesk in local languages within the central Orphanet GitHub or within the national hub. It will provide guidance for both implementing the nomenclature and using the

nomenclature and classification for statistical aggregation analysis for coded data. The elaboration and the first implementation steps of the national action plans (deliverable 4.2, https://od4rd.eu/03-deliverables/Deliverable_4.2_OD4RD_National_Action_Plans.pdf) led to a number of lessons learned. These are summarised in this document, together with the most important questions asked by users via the provided GitHub tool and answered by Orphanet experts.

3. Methodology

BfArM and INSERM as coordinating partners of WP4 organised 10 WP4 virtual meetings between March 2022 and January 2023 where the elaboration and implementation of the national action plans have been presented and discussed. At the final meeting all national hubs received a template by the coordinating team. Here, all teams were given the opportunity to summarise their most important successful implementations as well as experienced hurdles. These summaries have been brought together in this document.

The end users had the opportunity to ask questions related to RD coding or the Orphanet nomenclature via a GitHub tool that has been provided and supported by the team of OD4RD WP5. The answers are continuously provided by the coordinating team to support national helpdesks and end users. For each recurrent question, a dedicated Q&A has been developed in a generalised and standardised way in order to facilitate common knowledge and provide a sustainable Orphanet reference resource.

Participating countries:

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

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*France participated in WP4 only as coordinating partner and did not develop a national action plan; Switzerland is participating in WP4 as observer and has been given the opportunity to voluntarily develop their national action plan.

4. Lessons learned

4.1. Lessons learned on trainings

During the project trainings have emerged as an important component with regard to the acceptance of ORPHAcoding. As a first step a training for national hub trainers has been provided. The Orphanet nomenclature team led four training sessions of 3 hours each based on the needs that have been assessed by a survey among national hub members. All the training materials including the nomenclature exercises from these sessions have been made available to the OD4RD network as a reference and as a guide for following trainings.

After receiving the 'training for trainers' program, more than 60 local trainings or workshops had been held by the 14 national hubs, with more than 1,900 participants representing more than 110 ERN expert centers. This led to the following lessons learned.

First of all, it has been important to contact the health care professionals of the ERN centers and motivate them to participate in the trainings. Support from regional health authorities or regional associations, if available, has often proven helpful in this regard.

The number of participants in the trainings conducted for one center has not always been satisfactory. On the one hand, this was due to the fact that the information about the trainings did not always reach all potential participants despite the use of mailings, advertising on Orphanet national websites etc.

The experience has not been surprising that raising awareness in centers that do not use ORPHAcodes turned out to be more difficult than in those that already use the ORPHAcodes and are convinced of their necessity. It might be useful to explain in detail the objectives of the project and the needs of the RD community in the invitation mails, and to use paper letters in addition to create more attention. Also, it should be useful to broaden the targeted audience by also raising awareness among hospital managers. Finally, one should consider the possibility of publishing links to Orphanet online courses on the ERN websites.

Also, the lack of time available for participation has been a reason why a number of possible participants could not attend. Recording training sessions can be very helpful in engaging those who are unable to attend training sessions due to time constraints as well as developing online self-training material .

In order to provide targeted and satisfactory trainings, it was first important to determine the specific needs of the potential participants. Conducting a survey can certainly provide the most comprehensive and detailed information on participant needs. The materials developed by Orphanet in the European joint programme on rare diseases (EJPRD) project have proven to be a valuable basis for designing the trainings.

Basically, it was evident that training in local language with translated supporting materials (exercises, videos etc.) could contribute significantly to the success of the training activities. It should be noted that in multilingual countries like Belgium communication and trainings must be done in several languages which means that more trainers are needed compared to other countries and that more time has to be invested in the creation of new material.

Regarding the length of the training sessions, a duration of no more than 90-120 minutes has been found most efficient. Trainings with a significantly longer duration were often judged to be too long and also represented a hurdle to attracting additional participants to the training due to a lack of available time. In the case of extensive training content, it should therefore be considered to offer the training in several partial trainings, possibly flanked by additional online quizzes or exercises. However, should it be necessary to convey theoretical and practical contents in one training session, a duration of 3 hours should be considered.

Both online and on-site trainings have shown their benefits. The on-site trainings have distinguished themselves above all by the fact that an intensive training with reference to the individual needs of the participants was possible. There was also the possibility of interactive training with extensive immediate feedback. The limitations were the significantly higher effort and the relatively small number of participants. The online trainings were distinguished by the fact that a large number of participants (up to 450) could be included and thus the range of the trainings was significantly higher.

Also, the effort required to conduct these trainings was lower. A disadvantage was the reduced possibility of tailoring the training content to individual needs and the limited possibility of direct feedback. Based on the experience gained, the use of both forms of training can be considered ideal, whereby online training is certainly advantageous for conveying the general topic of ORPHAcoding and the Orphanet nomenclature. On-site training is of particular importance if the implementation of RD coding is to be prepared or accompanied on-site.

In general, the national hubs were able to report that the feedback from the participants after the training sessions has been very positive.

While in terms of train the trainers' feedback (where National Hubs staff is trained by the Orphanet Scientific team), the participants also provided a very positive feedback. Also, a recurrent comment concerns the need to develop regular ORPHAcoding live sessions to address coding issues, share experiences and gain knowledge on this topic within national hubs with support of the coordination leaders. This will be taken up in OD4RD2 by scheduling additional meetings in-between the regular WP4 meetings to address these issues. The experience gained should lead to a continuous 'training for trainer' program that benefits the members of the national helpdesks.

4.2.   Lessons learned on networking

The most straightforward way to achieve coding of RD using ORPHAcodes is the implementation of ORPHAcodes into national coding systems combined with mandatory coding for centers for RD or even all inpatient facilities in the country, as it will be implemented in Germany beginning in April 2023, and as it was implemented in France during the current national plan for RD.

It might be favourable to begin with addressing the coding of RD with ORPHAcodes in a National Rare Disease Plan. This creates awareness of the RD issues and explains the preferred way in the European Union to implement the coding of RD diagnoses with ORPHAcodes. It may also highlight the need that unique coding of RD has as a basis for advances in diagnosis and treatment as urgently needed to improve the care situation of people with RD. This should be used to make other stakeholders, such as medical societies, aware of the issue and get their support. Making clear the added value of using ORPHAcodes is a critical factor in convincing decision makers to adopt its use. Tools such as the *Orphaviewer*, a dashboard developed by the Dutch team with which per ORPHAcode the number of registered patients can be seen, can be helpful in this regard (see figure 1). Also, the tools of the ORPHAcoding promotion pack that was released during the RD-CODE project can be reused and adapted.
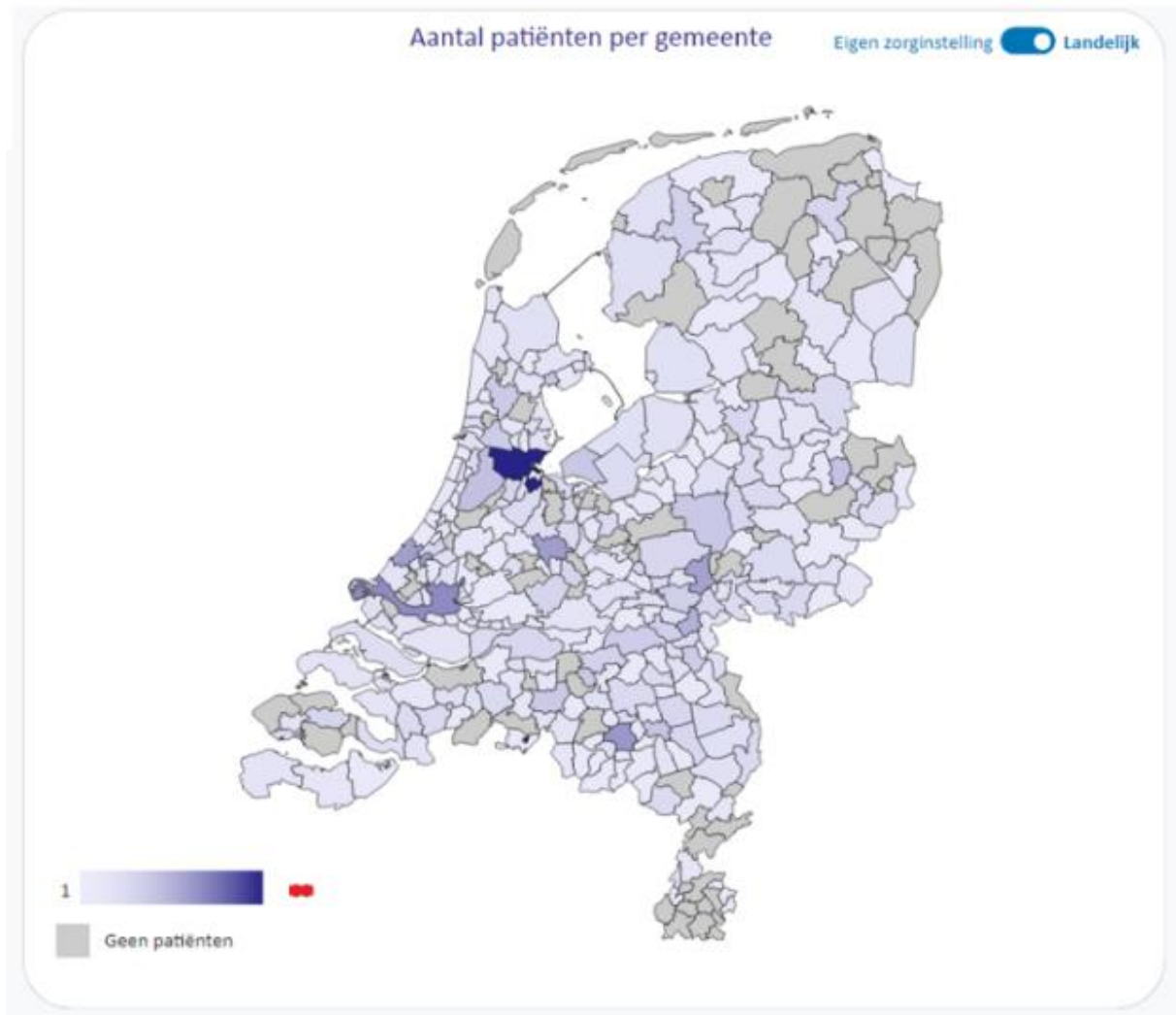
Figure 1: Proportion of RD patients (in blue scale) per community in the Netherlands for a group of RD by Orphaviewer (grey: no patients).

Another way to establish ORPHAcoding in specialised centers for RD is via creating national registries or ERNs' registries of RD, as ORPHAcodes are needed as one of the mandatory common data elements for RD registration. The offering of information events or trainings for medical specialists to create awareness for added value among members of the scientific societies might create some movement towards adaptation and involvement in the ORPHAcode registration implementation.

In addition to support from medical societies, the involvement of RD patient organisations can be helpful to support information dissemination and political lobbying for the use of ORPHAcodes.

The lack of legislative framework to use ORPHAcodes and the lack of financial incentives/reimbursement for treatment of RD patients significantly complicates the interest to implement ORPHAcodes.

Also, the use of SNOMED-CT as the common national reference terminology in electronic health records (EHR) in several European countries does not lead to facilitation of ORPHAcode implementation because often the benefit of additional use of ORPHAcodes is not sufficiently recognised. In addition, there is often no willingness to integrate another code system in addition to SNOMED-CT due to the additional effort involved. In this case, it is necessary to explain the limitations

of SNOMED-CT with respect to RD and the additional gain for patients and health care professionals that ORPHAcoding allows. In the OD4RD2 project a white paper on implementation issues of ORPHAcodes in relation to other systems such as SNOMED-CT or ICD-11 will be developed that will provide joint use guidelines, highlighting the complementarity of both systems.

If the use of ORPHAcodes in registries or national code systems is considered as a future option it is necessary to raise awareness for ORPHAcoding with the developers and users of the health information systems. In each country involved, the integration of information systems at the level of the Ministry of Health is essential to guarantee the use of ORPHAcodes.

4.3.   Lessons learned on national helpdesks

Within the project, the establishment of a national helpdesk (NH) for questions regarding RD coding and Orphanet nomenclature was a key issue. Some countries already had corresponding helpdesks, while others were setting them up.

In order to have a centralised, transparent, traceable, organised tool for all the issues a GitHub system was provided by the coordinators for procedural implementation as a free, open access and available tool (https://github.com/OD4RD). This enables user questions to be addressed directly to the NH, supported and shared with other NHs as well as to be forwarded to the coordinators if the answer cannot be provided by the NH.

During the first year, 26 people joined the GitHub tool progressively (11 from the coordinating team and 15 from NHs) representing 12 out of the 14 participating countries (85%), sometimes with more than one national representative by helpdesk. In total 46 questions have been posted from 7 NHs and one external: 37 have been answered and closed as complete and 9 are still open with ongoing answers. 40 flagging labels corresponding to either the topic or the country of the question have been developed to help identification.

The GitHub helpdesk tool was adopted by most national hubs. Only 2 (15%) do not have an NH account due to IT security requirements of their institutions. Initially the need to set up a new account and the unfamiliar handling meant that some users were not in favor to use the system. Alternatively, NHs can still use other communication channels such as e-mail to facilitate information access to the external users without increasing their workload. In this case, questions that cannot be answered by the NH should be forwarded to the coordinators via GitHub for precise answer and to ensure traceability.

To provide a sustainable and homogeneous, standardised support, a 'Questions and Answers' section within GitHub has been developed by the coordinating team based on users' questions. It provides standardised and generalised answers among 7 main topics: alignments with other terminologies, coding recommendations, education and communication, epidemiology of RD, ORPHAcodes and nomenclature, Orphanet classification and Orphanet tools. It is open access to any end users or national helpdesks.

To complement the national expertise of the national hub it may be beneficial to bring in experts to the NH in an advisory capacity. This might help especially with regard to country specific coding questions. The establishment of national working groups with coding expertise has proven helpful in this respect.

A current list of exhaustive questions and answers by categories is presented in chapter 5 of this document and can be found at: https://github.com/OD4RD/Main-Help-Desk/wiki. The coordinating team is regularly adding updated content and corresponding answers.

4.4. Lessons learned on RD coding implementation

Implementation of ORPHAcodes in national health information systems (HIS) usually turns out to be a complex process.

Capacity building and direct support by Orphanet experts is extremely useful in order to avoid inappropriate implementation, as choosing the right tools is difficult without in-depth knowledge of the classification. Alignment with parallel classification/nomenclature systems has to be considered. It may be useful to start with simple solutions for implementation, as adaptation of the entire IT-system and all cross-links is difficult and may delay the first implementation of ORPHAcodes.

Due to the numerous annual updates of the Orphanet nomenclature, that closely follow the evolutions of knwoledge, it is essential to consider that the maintenance of the coding systems containing the ORPHAcodes requires sufficient available capacity, which must be ensured in the long term.

When introducing ORPHAcodes into registries, care must be taken also to ensure that the codes can be used correctly. One problem encountered in local registries are different "dialects" of ORPHAcodes when letters or numbers are added to the codes for different purposes. This practice should be avoided, because it can hamper the cross-national comparability. Also, the difference between coding diagnosis and phenotype description is very often not understood, which can lead to the misapplication that two or more ORPHAcodes are used for one and the same patient with only one RD.

It is also important to provide feedback to Orphanet in the case of structural features or questions about the designation of entities or entity groups in the Orphanet nomenclature that may hinder or create isolated problems when introducing ORPHAcodes into national coding systems. Here, the Orphanet team may consider whether certain adaptations to the needs of the countries can be implemented to facilitate the use of ORPHAcodes.

4.5. Additional lessons learned

It should be ensured that the OD4RD objectives are aligned with other European projects focused on RD to avoid compromising of the success of the project. As an example, we highlight the future Joint Action for Rare Diseases Integration (JARDIN) and the new European Partnership for Rare Diseases (RDP). This will be ensured by the active Orphanet partners participation in these initiatives.

Nevertheless, the extension of dissemination and implementation of ORPHAcodes by all health care providers still remains to be done and efforts on that direction continue to be a priority to the national hubs.

5. Orphanet questions and answers

An up to date and detailed FAQ is available here: https://github.com/OD4RD/Main-Help-Desk/wiki

6. Discussion and conclusion

Depending on the state of coding in their respective countries, participants have undertaken extensive measures to achieve the WP4 goals. These measures have been summarized in the national action plans (https://od4rd.eu/03deliverables/Deliverable_4.2_OD4RD_National_Action_Plans.pdf).

Numerous trainings and workshops have been conducted or prepared. The use of training and training material in the national language, the restriction to a compact time format and the context-optimised use of online and on-site training in each case proved to be advantageous. The identification of concrete training needs through a survey in the ERN centres should provide an optimal basis for the design of the training measures.

The involvement of administrative bodies with the support of professional associations and patient organisations is crucial to drive the implementation of ORPHAcodes in national coding systems and national registries. The elaboration of the concrete additional benefits of the use of ORPHAcodes for patients and HCPs have emerged as important measures.

The establishment of national helpdesks is of great importance to support the introduction and monitoring of ORPHAcodes and to support user feedback to improve the orphanet nomenclature.

Orphanet's support of the implementation of ORPHAcodes in coding systems is of great importance, as the complexity of the process as well as the alignment with parallel classification/nomenclature systems has to be considered.

The lessons learned will provide valuable guidance for further work in OD4RD2, both as a guideline for newly joining national hubs and for the further development of the national action plans and their implementation by the already participating national hubs.