Deliverable 4.2
OD4RD National Action Plans
20.12.2022

This document represents deliverable 4.2 of the OD4RD project, which has received funding from the European Union. It contains the national action plans by the members of OD4RD work package 4, developed from April to November 2022. 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 www.OD4RD.eu

Disclaimer:

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

Content summary

Context

There are currently an estimated 30 million people in the European Union who suffer from one of the more than 6000 different rare diseases. Since 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, advances in diagnostics and therapy in the field of rare diseases are particularly important.

Since only a few patients suffer from a particular rare disease, 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, which applies to rare diseases only in very few cases if using generic terminologies. Therefore, additional appropriate identifiers for coding rare diseases become necessary. The Recommendation of the Council of the European Union of 8.6.2009 for action in the field of rare diseases advises that: "An appropriate classification and codification of all rare diseases is necessary in order to give them the necessary visibility and recognition in national health systems." 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. ORPHAcodes were considered as "Best practice" by the European Commission in 2017 (see European Best Practices portal: https://webgate.ec.europa.eu/dyna/bp-portal/transfered).

The RD-Action project (CHAFEA Grant Nr.677024) 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. These guidelines and recommendations were applied in the follow-up project RD-CODE (CHAFEA Grant Nr.826607) in the introduction of ORPHAcodes into the national coding systems of four European countries and the guidelines were further refined according to real life experience.

The current OD4RD project aims to support and facilitate the implementation of ORPHAcodes in hospitals belonging to 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 will build on the specific Orphanet expertise, and on its organisation as a long-lasting, well-established network, to fulfil the following general objectives:

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

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

data source level.

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

Objective of OD4RD work package 4

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

Objectives of the national action plans

A survey among participating countries was launched at the beginning of the project to assess each national situation in terms of ORPHAcoding implementation (available at: https://od4rd.eu/03-deliverables). This revealed that the overall picture of RD coding with ORPHAcodes is very different between work package 4 participating countries. The range extends from already implemented ORPHAcodes with linkeage to ICD-10 in national coding systems used in all hospitals to not yet using ORPHAcodes at all.

Taking into consideration the different situation and needs of end users in regards 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 the following tasks:

1) Provide training sessions in local language for coders, hospital information managers and statistical services and other stakeholders involved, as well as clinicians in ERN centers.
2) Actively participate 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 coordination and adapting or translating it when needed.
3) Establish 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.

Methodology

BfArM and INSERM as coordinating partners of work package 4 developed a template that enabled the participating countries to document the development of their measures to ensure support for the local implementation of ORPHAcodes in national HCPs hosting ERNs and national HCPs linked to ERNs. The template has been divided into four sections: trainings/workshops, networking, helpdesk implementation and further activities. The respective further developed versions of the national teams were available to all participants at all times on an internal OD4RD WP4 website. During the monthly WP4 conference calls these topics were discussed and experiences of the different countries were shared in order to benefit from each other experiences and to address common topics in a coordinated manner. Finally, the developed national action plans were presented and discussed.

This document presents the national action plans as by November 2022.

Participating countries:

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

*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 a national action plan.

National hubs and national action plans

National hub of Austria

The Austrian national hub is made up of the team of Orphanet Austria. It is located at the Medical University of Vienna and consists of 2 staff members:

Prof. Dr. Till Voigtländer (medical doctor, Orphanet country coordinator)

Dr. Ursula Unterberger (medical doctor, Orphanet deputy country coordinator, project manager, information scientist)

Orphanet Austria is in part financed by the Austrian Ministry of Health. In addition to Orphanet activities their members are also working on establishing and integrating of national reference networks for RD, establishing a national undiagnosed rare diseases program and conception of a national RD registry.

National action plan of Austria

PLANNED ACTION	TIMELINE
Trainings/Workshops: Mandatory trainings will be held for Centers of Expertise (CoE) and Associated National Centers (ANC) in ERNs, either online or in person. These trainings will start once ORPHAcodes have been implemented in the official hospital information systems in pilot HCPs (General Hospital Vienna, Private Medical University Salzburg, presumably Medical University Graz). Facultative trainings will be offered to centers and other institutions, as well as interested experts which are currently already using ORPHAcodes in registries.	Q1/23 for facultative trainings Mandatory trainings according to the timeline of the implementation in hospital information systems
Networking: Orphanet Austria is represented in the National Advisory Board on RD, and the National Office for RD is identical to the Austrian Orphanet team. Therefore, the use of ORPHAcodes is already mentioned in the national action plan for RD, which was mostly drafted by the Austrian Orphanet team on behalf of the Ministry of Health. In the context of the implementation of ORPHAcodes, we have been working closely with Pro Rare Austria (umbrella organization of PO) to implement ORPHAcodes in electronic health records (ELGA) and in the planned patient information card. This project is currently waiting for the official implementation of ORPHAcodes in hospital information systems (a project that is included in the Orphanet contract between the Ministry of Health and Orphanet Austria). In this context, we are closely working together with several clinical departments and hospitals to implement Orphacoding on a pilot basis in a number of CoE and ANC, before a general roll-	ongoing

out in all official centers, which is planned for 2023. Regular meetings are held between Orphanet Austria, the MoH, and the centers and the persons responsible for the respective hospital information systems. Official designation as CoE includes the commitment to implement ORPHAcodes, and experts/centers, as well as hospitals are highly motivated and compliant.	
Helpdesk Implementation: Orphanet Austria is (and has been for a long time) taking questions and requests, as well as providing assistance on Orpha Coding for a long time under the email address ursula.unterberger@meduniwien.ac.at The interactions have been intensifying as regular meetings are being held (see above) and networking is expanded and structured.	done
Further Activities: Orphanet Austria is responsible for the implementation of ORPHAcodes in Austria in general, so all activities necessary in this context are performed by us (see above; advice beyond the Orpha classification and nomenclature, including advice on possible applications, implementation strategy, technical issues, go-between for institutions within the healthcare system plus organization of regular meetings).	ongoing

National hub of Belgium

The national hub of Belgium is built by the Belgian Rare Diseases team. It is hosted by Sciensano, the Scientific Institute of Public Health and contains the following staff members:

Elfriede Swinnen (PhD Medical Science, National coordinator of Orphanet Belgium)

Kim Van Roey (PhD Biochemistry and Bio-informatics, in charge of the Dutch translation activities (Orphanet RD nomenclature and encyclopedia, Orphanet international and national website))

Annabelle Calomme (MD Molecular Biology, Information Scientist for the Orphanet Belgium database management, OD4RD national Hub project manager)

Katrien Van Der Kelen (PhD Biotechnology, Manager of the Belgian Central Registry for Rare Diseases)

The Rare Diseases team is part of the Service “Health Services Research”. The current team has 10 years of experience with performing tasks related to Orphanet, and also has expertise in developing and implementing other rare disease projects that are part of the Belgian plan of rare diseases. The Service that hosts the team also manages national patient registries e.g. for cystic fibrosis, neuromuscular diseases and hemophilia.

National action plan of Belgium

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Analysis of available material and development of ad hoc material for national trainings (PowerPoint document, quiz, video in local languages: FR, NL)	Done (March-November 2022)
Delivery of national trainings, online/on site, to university hospitals with an RD function:	
- UCL Saint-Luc: online, in French and English (two-hours session, basic training on nomenclature and codification, theoretical part, 15 participants: clinicians and RD coders);	Done (April 29, 2022)
- UCL Saint-Luc: online, hands-on session based on the advanced quiz developed by the Orphanet coordinating team;	Done (August 29, 2022)
- Ghent University Hospital: online, in Dutch (two-hours session, basic training on nomenclature and codification, theoretical part, 15 participants: clinicians and RD coders);	Done (September 19, 2022)
- CHU Liège: online, in French and English (one-hour session, Q&A on ORPHAcodes. Participants: clinicians and RD coders, advanced level (ORPHAcodes already used in the patient files).	Done (October 21, 2022)

- Other Belgian university hospitals (UZ Brussel, UZ Antwerpen, UZ Leuven, ULB Erasme) Delivery of national trainings, online/on site, to non-university hospitals, ERN-centres and any institutions involved in the RD field	To be planned with the healthcare professionals (Q1-Q2 2023) To be planned with the healthcare professionals (by the end of 2023)
Networking: Communication of the participation of Belgium as a pilot-country during the "Rare disease accompanying committee" meeting. This committee is made up of representatives of different Belgian health authorities (including SPF-FOD and INAMI-RIZIV), a representative of the Ministry of health and several members of officially-designated genetic centres. Contact by email with the coordinators of the 8 Belgian hospitals with a RD Function as well as other key people active in the rare disease field in our country to inform them about the OD4RD project, its goals and anticipated actions. Publication of information on training opportunities for professionals on the Orphanet Belgium website (FR, NL). Presentation of the OD4RD project at a meeting with the "Belgian Working Group Rare Disease Functions/College Genetics & Rare Diseases". Invitation by paper letter (FR, NL) to the coordinators of the 8 Belgian university hospitals with a RD Function, as well as other key people active in the RD field to promote the trainings (about 200 Belgian professionals have been identified) Meetings with the SPF-FOD terminology centre in order to support the implementation of ORPHAcodes in Belgian electronic health records.	Done (April 28, 2022) Done (May 12, 2022) Done (May 12, 2022) Done (October 7, 2022) Planned: January 2023 Done (November 16, 2022: first meeting) Done (December 09, 2022: follow-up meeting)
Helpdesk Implementation: Members of the rare disease team at Sciensano act as a national helpdesk for coding matters. A process is already in place to manage questions addressed by emails ('Orphacodes.Belgium@sciensano.be') or by phone by ORPHAcode users. If necessary, a	Ongoing and throughout the year

conference call is organized to deepen the discussion with the professionals. <u>Description of the structure of the helpdesk:</u> It consists of three people: - **Katrien Van Der Kelen**, Manager of the Belgian Central Registry for Rare Diseases; - **Annabelle Calomme**, Orphanet Belgium Information Scientist for the database management; - **Kim Van Roey**, in charge of the Dutch translation activities. <u>GitHub tool for Orphanet national hubs:</u> Creation of posts related to questions received regularly via the national helpdesk Promotion of the tool to the professionals.	Ongoing and throughout the year Ongoing and throughout the year
<u>Dissemination activities:</u> Mention of the OD4RD project in the 2021 activity report of the Orphanet Belgium database (available in FR, NL, EN) published on the website of Sciensano and on the Orphanet Belgium website. Creation of a SharePoint to make documents available to external partners. Creation of new resources: leaflets and satisfaction survey (FR, NL) Translation of the video on ORPHAcodes in Dutch. The video is available on: - YouTube: https://www.youtube.com/watch?v=fJV2JHll1Ng - RD-CODE website: http://www.rd-code.eu/wp-content/uploads/2022/10/orphacodes_final_dutch.mp4-1080p.mp4 Promoting the use of ORPHAcodes to government and other health institutions (non-university hospitals, ERNs centers of expertise, Flemish Network Rare Diseases,…).	Done (April 2022) Done (May 2022) Planned: by the end of 2023 Done (September-November 2022) Ongoing and throughout the year

Further Activities:	
Translation and update of the Orphanet nomenclature and Human Phenotype Ontology (HPO) terms into Dutch; collaboration with Belgian Terminology Centre with regard to Dutch terms.	Ongoing and throughout the year
Evaluation of the progress and room for improvement, based on the feedback received from the participants.	Ongoing and throughout the year

National hub of Czech Republic

The national hub of the Czech Republic is build by 2 members of the Orphanet Czech Republic team, located at the Department of Biology and Medical Genetics of Praque University hospital, and 2 members of the Institute of Health Information and Statistics:

Prof. Milan Macek (M. D., Ph.D., MHA, National coordinator of Orphanet Czech Republic)

Marek Turnovec (M. D., Information scientist of Orphanet Czech Republic)

Miroslav Zvolský (M. D.)

Kateřina Hanušová (MSc.)

Their national Orphanet teams main areas of work include developing and implementing of strategy and action plans for rare diseases, translation of rare diseases nomenclature and Human Phenotype Ontology (HPO) into Czech language and support of implementation of ORPHAcodes into information systems.

National action plan of Czech Republic

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Trainings at University Hospitals at Prague and Brno where are almost all Czech ERN centers based At University Hospital Motol ORPHAcodes were implemented in September 2022, two trainings followed, one for representatives of ERNs and one for representatives of all clinics. We will then continue at other hospitals according their needs.	Done, we are prepared to make another training when needed by the clinicians (probably during the year 2023).

We already have prepared materials from previous workshops.	
Networking: Two institutions involved: • National Coordination Center for Rare Diseases at Department of Biology and Medical Genetics, 2nd Medical Faculty of Charles University & University Hospital Motol • National Centre for Digitisation of Healthcare at Institute of Health Information and Statistics Cooperation with all ERNs in Czech Republic via Interdeparmental Commission for Rare Diseases at Ministry of Health Cooperating with Czech Association for Rare Diseases and other patients organizations on promotion of ORPHAcodes among patients	Cooperation already established.
Helpdesk Implementation: We are already providing support over e-mail and phone and with direct contact and demonstration with clinicians. We are still discussing about usage of GitHub on national level. We are considering the possibility to develop our own website with contact forms with simple issue tracking that can be localized into Czcech language, because GitHub is not localized. But we can still use it internally and also refer to it the clinicians who can understand English.	Ongoing Our own (localized) issue tracking system could be launched in 1st quarter of 2023
Further Activities: Updating the Czech translation of the nomenclature and providing this to health care providers and health insurance companies Promoting usage of ORPHAcodes at different national conferences (e. g. conferences of Society of Human Genetics)	Ongoing (and neverending) process Planned to conferences during 2023

Cooperation on revisions of Czech translation of ICD-11 (with usage of previous translation of Orphanet nomenclature)	Mostly done during 2022
Translation of Human Phenotype Ontology (HPO) in Czech language	Ongoing
Cooperation on creating pilot Czech Rare Disease registry	Pilot version of the registry is currently developed
Cooperation with companies providing hospital and laboratory information systems and providing them support	We already prepared specifications for developers of one hospital information system in 2022, we are prepared to collaborate with other companies in 2023

National hub of Finland

The national hub team of Finland contains of

Professor Helena Kääriäinen (medical doctor, country coordinator of Orphanet)

Mikko Härkönen (M.Soc.Sc, Development Manager of the Code Service)

Iiro Toikka (political science, Development Manager)

Satu Wedenoja (medical doctor, Chief Physician)

The team is located at the Finnish Institute for Health and Welfare (THL). They are supporting the adding of ORPHAcodes to the national Code Service of THL and to national health registries of THL. They also coordinate the national work in the field of RD, implement the use of ORPHAcodes and participate in expert networks (Nordic collaboration, ERNs, Orphanet, national networks) within the scope of the second Finish national plan and stategy for RD.

National action plan of Finland

PLANNED ACTION	TIMELINE
Trainings/Workshops: Information on ORPHAcodes delivered to Finnish ERN centers and departments of clinical genetics at the university hospitals	February 2022

National expert group on rare diseases: Survey about piloting and implementing ORPHAcodes at the university hospitals	September 2022
Translating the promotion video of ORPHAcodes in Finnish	October 2022
Information on ORPHAcode to clinicians and patient organisations	
Promotion material of ORPHAcodes for hospital districts and companies responsible for electronic patient reports in Finland -THL, an information card	February 2023
THL electronic news (when pilot project starts in Helsinki area)	2022-2023
-Generating a list of the most common RD and related ORPHAcodes to the national Terveysportti health portal	2022-2023
Networking:	
THL Code Service	
-maintenance of ORPHAcodes	2022
-annual update of ORPHAcodes	August 2022
-emergengy guidelines (in Finnish) for certain diseases available for clinicians	2022-2023
Piloting the use of ORPHAcodes as part of a patients report system (Epic-based Apotti)	
-Hospital District of Helsinki and Uusimaa	January 2023-
-Other university hospitals	2023-
Increasing knowledge of ORPHAcodes through the national expert group and network of RD	2022-
Helpdesk Implementation:	
Assessing national needs for Helpdesk	Done
Setting up national Helpdesk in Github	2022

Further Activities:	

National hub of Germany

The national hub of Germany is represented by the team of Orphanet Germany. Their members are:

Dr. Stefanie Weber (medical doctor, National coordinator of Orphanet Germany)

Carina Thomas (medical doctor, Deputy national coordinator of Orphanet Germany)

Dr. Kathrin Rommel (Dr.rer.nat., Project manager of Orphanet Germany)

Kurt Kirch (medical doctor, Information scientist of Orphanet Germany)

Sven Rütz (Information scientist of Orphanet Germany)

Orphanet Germany is part of the department „Code systems“of the Federal Institute for Drugs and Medical Devices (BfArM), an independent federal higher authority within the portfolio of the Federal Ministry of Health. Besides their Orphanet responsibilities the team works on the development and release of Alpha-ID-SE data file for clinical coding, the support of national projects like Collaboration on rare diseases by the German Medical Informatics Initiative, the collaboration with stakeholders for rare diseases in Germany (e.g. NAMSE, ACHSE etc.) and the participation in European RD projects.

National action plan of Germany

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Presentation in frame of an event of the german “bvitg” (The German Health IT Association)	January 2022 (done)
Webinar Rare Disease Day (presentations on Orphanet and Coding of Rare Diseases)	February 2022 (done)
Advisory of the “German hospital federation” and “The National Association of Statutory Health Insurance Funds”	several Web-Meetings February/March 2022 (done)

Presentation/Workshop in frame of CORD-MI (German Medical Information Technology Initiative)	May 2022 (done)
Trainings/Workshops for the Implementation and usage of the german Alpha-ID-SE	08.11.22 (ca. 160 participants, done),13.12.22 and following in January 2023
Workshop “BfArM in Dialogue”	24. November 2022 (191 participants, done)
Creating Alpha-ID-SE information material (Flyer)	December 2022
Publication of essay in professional journal	done, Publication November 2022
Development of a new version of a training presentation of ORPHAcoding with Alpha-ID-SE and publication on German Orphanet website	December 2022
Presentation/Workshop for Coders of a German Hospital Association	07. December 2022
Networking:	
CORD MI (Collaboration on Rare Diseases by the Medical Informatics Initiative Germany)	Throughout the whole year, participating at monthly CORD workshops (mostly done)
NAMSE (National Action League for people with RD)	Participation in several Workshops/Meetings of NAMSE (mostly done)
Interaction with other governmental Departments	Throughout the whole year
Helpdesk Implementation:	
There is an existing Helpdesk concerning Orphanet Germany and all issues in frame of ORPHAcoding in Germany. Coders or other people can address their needs via phone or mail to get information or to get their problems solved.	implemented
Github account for German helpdesk	implemented

Further Activities:	
Work on Production of the RD-Code Video	Beginning of the year (done)
Translation Video ORPHAcodes	Beginning of the year (done)
Further development of German Alpha-ID-SE data file (version 2023)	done, publication in October (includes 11.625 diagnostic terms connected with ICD-10-GM and ORPHAcodes, representing 6.655 different ORPHAcodes)

National hub of Italy

The national hub of Italy is represented by the team of Orphanet Italy. It is hosted by the Bambino Gesù Children's Research Hospital (OPBG), in Rome and consists of the following members:

Prof. Bruno Dallapiccola, MD (National coordinator Orphanet Italy)

Dr. Rita Mingarelli, MD (Project Manager Orphanet Italy)

Serena Ciampa (Biologist, Information Scientist Orphanet Italy)

Michele Nutini (Biologist, PhD, Information Scientist Orphanet Italy)

Fabiana Battisti (IS, Social media manager)

Antonella Longhi (Translator)

They have been involved in drafting both the first and the second National Plan for Rare Diseases (2013 and 2021). They supported the integration of ORPHAcodes into the health and research information systems of 10 Italian Regions/Autonomous Provinces.

National action plan of Italy

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Contact the centers of expertise to inform them of the service that Orphanet Italy offers (new editions of the training "Orphanet Nomenclature and Ontologies of Rare Diseases for Research", workshops)	Done. Each registered professional linked to an ERN expert center has been contacted and informed about the incoming OD4RD training, planned on December 15. This new course

The training material has been prepared in advance in the context of the EJP-RD project, with addition of new documents provided by the OD4RD project, and has been translated and adapted to the audience.	follows the last EJP-RD-branded course held on 14 April 2022. Done.
An updated version of the post-training material (a summary with brief guidelines) has been prepared.	Done.
Networking:	
Rete Idea (Network of pediatric research hospitals)	Done. This collaboration is already in place, the upcoming OD4RD training (December 15th) has been disseminated through the Rete Idea website.
S.I.G.U.	Ongoing. This collaboration is in place and further implemented to launch the 2023 OD4RD trainings.
National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy	Planned for 2023
Italian Inter-regional Technical Board for Rare Diseases	Planned for 2023
Regional coordinators of the Italian National Network for Rare Diseases	Planned for 2023
Ministry of Health	Ongoing. The country Coordinator, Bruno Dallapiccola, participated as expert in the working group who drafted the Second National Plan of Rare Diseases (LEGGE 10 novembre 2021, n. 175). The article 4 states that the updated list of rare diseases/groups of rare diseases included in the Essential Levels of Care must refer to the ORPHA codes and to the Orphanet classification system.

Helpdesk Implementation: Establish the Italian helpdesk (using the GitHub platform) to answer questions related to nomenclature and the implementation of ORPHAcodes	Ongoing. The GitHub account has been created, but it has not been used so far. Standard Email exchange was promoted by our team as the main tool to contact Orphanet-Italy. We plan to use emails for communicating between professionals and Orphanet-Italy, and GitHub for communicating between Orphanet-Italy and CT.
Further Activities: Dissemination of the OD4RD project aims. Mapping the italian centers of expertise, members of a European Reference Network (ERN). Registration of the italian centers of expertise, members of a ERN, in the italian Orphanet database.	Done. We informed the subscribers (about 5500 people) of the Italian version of OrphaNews about the objectives of the new direct grant OD4RD (see the editorial section of 13 May 2022). Done. All available information about Italian ERNs' full members have been collected. Ongoing. More than 80% of Italian ERNs' HCPs (respect to the total number provided by a dedicated EC website) are implemented in the Italian database.

National hub of the Netherlands

The national hub of the Netherlands consists of the Dutch Orphanet team. It is located at the Radboud University Medical Centre, Nijmegen and has the following members:

Prof. Wendy van Zelst-Stams (MD PhD, National coordinator Orphanet Netherlands)

Judith Carlier-de Leeuw van Weenen (PhD, Information Scientist Orphanet Netherlands)

Besides the work for Orphanet they participate in the national assessment of expert centres for rare diseases, the Undiagnosed Diseases Network International and at the board of Member States for ERNs. They also support the implementation of ORPHAcodes in Dutch Health Care Records.

National action plan of the Netherlands

PLANNED ACTION	TIMELINE
Trainings/Workshops: The implementation of ORPHAcodes in the Dutch health information system is commissioned by the NFU (The Netherlands Federation of University Medical Centres) so it was decided the NFU would be in the lead when it comes to communications concerning the implementation of ORPHAcodes. The NFU will launch a communication campaign in which a training/workshop on ORPHAcodes will be integrated. We are evaluating the existing training material on ORPHAcodes to determine what will be included in the training/workshop for Dutch professionals.	Q1/2023 Q4/2022
Networking: Regular meetings with the following relevant organisations are in place: 1. Dutch Ministry of Health, Welfare and Sport – *observer* 2. NFU (The Netherlands Federation of University Medical Centres) – *client* 3. DHD (Dutch Hospital Data) - *in the lead of the actual implementation of ORPHAcodes* 4. Nictiz (The Dutch competence centre for electronic exchange of health and care information) – *participating in the implementation of ORPHAcodes* Organisations still needing to be involved:	1. Regular meetings; every 4-6 weeks 2. Regular meetings; every 2-4 weeks 3. Regular meetings; monthly 4. Regular meetings during implementation phase / in later phase: contact when necessary

1. All Dutch Societies of Medical Specialists	1. Contact via individual members which will be target of the NFU communication campaign
Helpdesk Implementation: Dutch professionals are able to reach the Dutch Orphanet helpdesk via 3 routes: 1. Via an e-mail to orphanet@radboudumc.nl. This route is already in place since several years and is well established. 2. Via the DHD helpdesk. Due to the way ORPHAcodes are implemented the first contact point for coding questions is the DHD (Dutch Hospital Data) helpdesk. If the questions concern ORPHAcode matters that DHD professionals can't answer, the question will be redirected to us. 3. Via Github. An Orphanet NL account is created so GIthub can be used to reach the Dutch Orphanet helpdesk, but for the moment this helpdesk entry point will not be actively promoted.	Implemented Routing from DHD to Orphanet NL helpdesk provisionally implemented. In Q4/2023 an evaluation will follow to determine if formal implementation is necessary. Account in place
Further Activities: Currently almost 5000 ORPHAcodes are linked to disease terms in the Diagnosis thesaurus. This means that still ~1200 ORPHAcodes are not mapped to a DT term. Of the ~5000 ORPHAcodes 1853 could be mapped to existing DT terms and for 3125 ORPHAcodes new DT terms had to be created. To not overload medical professionals with new DT terms it was decided to map the new ORPHAcodes only when necessary, meaning: 1. If specifically requested by medical specialists/hospitals 2. If expert centres received designation for ORPHAcodes that are not yet mapped DHD will develop an „ORPHA-viewer", a dashboard with which per ORPHAcode the number of registered patients can be seen. This	Throughout the year Q1/2023

can e.g. be used to compare the number of patients seen in one hospital compared to the number seen in all hospitals for a certain ORPHAcode.	

National hub of Norway

The national hub of Norway is located at Oslo University Hospital under the National Advisory Unit on Rare Disorders and consists of the following members:

Stein Are Aksnes (Leader, biochemist, medical genetic councelor, National Coordinator Orphanet Norway)

Lene Martinsen (PhD. Biology/Genetics, Information Scientist Orphanet Norway)

Maria Emilie Gresslien (M.Sc. Public health, Information Scientist Orphanet Norway)

Linn Bjørnstad (PhD. Molecular biology, Norwegian Registry on Rare Disorders)

Mette Salomonsen (Nurse, Norwegian Registry on Rare Disorders)

Ingrid Helland (MD, PhD, Medical advisor)

Mari Bakken (MD, Medical advisor)

Besides their work on Orphanet they are also active in empowering RD patients and strengthening advocacy, strengthening RD networks and collaborations of professionals, participation in research networks and relevant ERNs and education of professionals, patients, organizations and the general public.

National action plan of Norway

PLANNED ACTION	TIMELINE
Trainings/Workshops: Information meetings, including basic training of clinicians, at all the 12 divisions at Oslo University Hospital (OUS) started autumn 2021 (see attachment 1 for details). We will offer meetings for each department or smaller groups within the departments if requested.	Ongoing

• Division of Peadiatric and Adolescent Medicine at OUS • Department of Neurology • Department of Dermatology • Division of Laboratory Medicine (Section for Medical Genetics) • Department of Endocrinology • Department of Pulmonary Medicine • Division of Technology and Innovation (IT) • Coding advisors: One from each division at Oslo University Hospital • Department of Hematology, Akershus University Hospital and Lovisenberg Diaconal Hospital	January 1st 2021-December 31st 2022 (mostly done)
Spring 2023 we will expand our focus area to other divisions. First e-mails have been sent out.	Ongoing
Survey on the use of ORPHAcodes and bottlenecks/challenges met by the professionals.	Q1 2023
Training/Workshop specifically targeting clinicians involved in ERNs: 3 hour training based on the EJP-RD training we arranged in March 2021 and Basic/Advanced trainings provided by Orphanet in 2022.	March 15th and March 30th, 2023 (postponed from October 2022 due to key staff absent)
• Decide on training curriculum. • Adapt training material to March 15th training.	By December 15th 2022 By February 15th 2023
E-learning course: • Finalizing the online content • Launching	By October 15th 2022 (done) By December 1st 2022
Networking: National Strategy for Rare Diseases from 2021: Contribute to Action point 6 - evaluation of the needs concerning coding of rare disorders, and Action point 7 – investigation of a National Registry on Rare Diseases. Meetings with responsible authorities (the Directorate of e-health, the Directorate of Health, and Regional Health Authorities) so far: • Regional health authorites	Ongoing (until June 2023) May 5th, 2022, November 9th 2022

• Directorate of eHealth and Directorate of Health • All three mentioned above – project group meeting to coordinate the follow-up on both action point 6 and 7.	November 23rd 2022 November 28th 2022
Southern and Eastern Norway Regional Health Authority: Follow up on request to implement ORPHAcodes in the new version of the EHR software (DIPS). This will also benefit hospitals in other health reagions using the same software.	Autumn 2022
• Project meetings for developing solution	August 31, September 12, September 14, September 19 (done)
Meeting with Molgenis and the Norwegian ERN-ITHACA node about possibility for local installation of the ILIAD registry as platform for the Norwegian Registry on Rare Disorders.	June 23rd 2022 (done)
Poster at ECRD 2022	June 27-July 1st (done)
Participation at annual meeting for diagnostics of syndromes (Karen Helene Ørstavik's Dysmorphology Meeting) (Syndromdiagnostikkmøte), Haukeland University Hospital	September 14-15 (done)
National coordinator Orphanet present at genetics seminar at Telemark Hospital	September 22nd (done)
National coordinator Orphanet present at the 13th Norwegian National Genetics Meeting, Tromsø	November 9-10 (done)
Establish contact points for future trainings in other hospitals than Oslo University Hospital, starting with the university hospitals: • Akershus University Hospital • Haukeland University Hospital • Stavanger University Hospital • St. Olav's Hospital • University Hospital of North Norway	Ongoing

Helpdesk Implementation:	
Deciding on solution (GitHub not possible)	Done
Setting up the technical necessities for the solution	Done
Launching the helpdesk	Done
Connect national helpdesk to GitHub	By December 15th 2022
Further Activities:	
Description of the structure of the national hub: Consists of 3 teams described on the hub's website. In addition, communication advisor Kari Andresen is responsible for information campaigns.	Done
Decide on solution for electronic registration form for the Norwegian Registry on Rare Disorders for clinicians outside Oslo University Hospital.	Q1 2023
Development of information brochure	Done
Distribution of information brochure	Ongoing
Develop information brochure about the Norwegian Registry on Rare Disorders for patient organisations.	Done
Distribute information brochure on the Norwegian Registry on Rare Disorders for patient organisations.	Ongoing

National hub of Poland

The national hub of Poland operates within the Children's Memorial Health Institute (CMHI) in Warsaw. It consists of the following team members:

Prof. Krystyna Chrzanowska (specialist in clinical genetics and pediatrics, National coordinator of Orphanet Poland)

Dr. Agnieszka Madej-Pilarczyk (specialist in clinical genetics and internal medicine, Information scientist of Orphanet Poland)

Prof. Jolanta Wierzba (specialist in clinical genetics, pediatric metabolism and pediatrics)

Besides the work on Orphanet they are active in developing a National Registry of Rare Diseases, participating in European RD projects, coding work regarding ICD-10 codes, ORPHAcodes and OMIM numbers and diagnostics of a wide spectrum of rare genetic diseases/syndromes to provide complete genetic counseling for families at risk.

National action plan of Poland

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Establishment of the Polish National Plan for Rare Diseases (the first version)	2021 (done)
In scope of the Polish National Plan for Rare Diseases the Ministry of Health established 3 councils:	
- for Rare Diseases	
- for Registries of Rare Diseases	
- for Information Platform „Rare Diseases"	
Cooperation with e-Health Centre – creating of an IT platform for the Registry of Rare Diseases, Passport of Patient with Rare Disease and the Information Platform	ongoing
- establishing of data and identifiers being processed	
- selection of expert centres and genetic outpatient clinics which will implement ORPHA codes	
Translation to Polish of the training materials for ORPHA coding	September-October 2022 (done)
Pilot individual training for representatives of metabolic and neuromuscular departments, members of ERNs	November 2022 (done)
Group training in scope of OD4RD	By the end of December 2022

Next trainings (individual and group) after launch of the Polish National Plan for Rare Diseases – according to needs	By the end of March 2023 and beyond…
Networking: Two meetings of the Council of Experts of the Medical Ration of State - dedicated for rare diseases: Krystyna Chrzanowska presented the National Plan for Rare Diseases, in particular focused on the creation of the Polish Registry of Rare Diseases.	15.02.2022 and 22.09.2022
Helpdesk Implementation: National Helpdesk in the Memorial Children's Health Institute, Warsaw – solving problems and providing support with ORPHA coding / answering questions on Orphanet – contact via e-mail and phone (in Polish) GitHub account for Polish helpdesk	done done
Further Activities: Translation and updating of the Orphanet nomenclature Translation of definitions and full descriptions of Orphanet disorders Correction of translation to Polish of ICD-11 (developmental anomalies and inborn errors of metabolism) Updated list of Polish ERN expert centres published on the Orphanet website (with support of Orphanet expert centres team) 6th Polish Congress of Genetics: oral presentation by Krystyna Chrzanowska "Rare diseases in Europe and in Poland" (with information on Orphanet activity)	ongoing and throughout the year ongoing and throughout the year November 2022 July – September 2022 27-30.06.2022

National hub of Portugal

The national hub of Portugal is formed by the team of Orphanet Portugal, which is located at the Department for Quality in Health, in the Directorate-General of Health (Ministry of Health). It consists of the following members:

Carla Pereira (PhD in Public Health, Master in Health Services Management, National coordinator of Orphanet Portugal)

Rui Gonçalves (MD in Clinical Genetics, Information Scientist of Orphanet Portugal)

Cristina Rocha (PharmD, DVM, Information Scientist of Orphanet Portugal)

Besides their activities for Orphanet they are managing the Portuguese Rare Disease Card, participating in the Integrated Strategy for Rare Diseases 2015-2020, participating in the EJPRD Policy Board and Mirror Group and the ERN Board of Member States.

National action plan of Portugal

PLANNED ACTION	TIMELINE
Trainings/Workshops: Training sessions: Workshops will be held online targeted to Reference Centres as ORPHAcodes users - Collect and select existing material created for trainings by Orphanet and EJP-RD on ORPHA coding and nomenclature - Translate and adapt new material and quizz created for training sessions - Map the national centres of expertise, namely Expert Centres members of the European Reference Network - Organize 2-3 online training sessions - Contact the Board of HCPs and Reference Centre Coordinators to send training invitations ORPHAcodes and OD4RD project promotion materials: Flyer, infographics and translated video version to be distributed/emailed/presented in meetings	 Sep. 22 (done) Sep. 22 (done) Jun-Oct. 22 (done) Nov. 22 (done and ongoing) Nov. 22 (ongoing)

and made available on the Orphanet national website, with the involvement of the National Commission of Reference Centres - Translate and present Orphanet promotion material in scientific meetings and conferences - Make available Orphanet promotion materials on Orphanet national website - Contact and meet the new President of National Commission of Reference Centres after his nomination in September 13th - Email promoting materials to Reference Centre Coordinators and collaborators	 Mar-Nov.22 (done) Nov. 22 (ongoing) Nov. 7th 22 (done) Nov. 22 (ongoing)
Networking: <u>OD4RD project formal presentation to the Ministry of Health</u> - OD4RD project and the national Action Plan were approved by the Ministry of Health <u>OD4RD project formal presentation to national commissions (to inform and involve)</u> The National Commission of Reference Centres - The new President of National Commission of Reference Centres was informed The Coordinating Commission for the Treatment of Lysosomal Overload Diseases The Coordinating Commission for the Treatment of Cystic Fibrosis The National Hemophilia Commission - Other Commissions Presidents will be contacted about OD4RD project and invited to Orpha training sessions - Other institutions: - INFARMED (National Medicines Agency) - INSA (National Health Institute) - CTC (Centre for Clinical Terminologies)	 June 22 (done) Nov. 7th 22 (done) Nov. 22 (ongoing) May-Oct. (done)

Umbrella Patient Organization Semestral meetings between National Team and umbrella PO to be promoted in order to highlight the relevance of ORPHAcodes - One dedicated meeting with the Umbrella Patient Association (RD Portugal) for dissemination of OD4RD project and the importance of completeness of Orphanet Website. - RD Annual Event During the Rare Diseases Day annual event the use of ORPHAcodes will be highlighted by the National Hub presentation	Nov.14th 22 (done) Feb. 23 (planned)
Helpdesk Implementation: In a previous stage the National Team will take the helpdesk role while answering questions related to ORPHAcoding within the scope of the Rare Diseases Card. - The Orphanet National Team routinely answers to requests/clarifications on Orphanet nomenclature within of Rare Disease Card new registries - Implement and promote GitHub tool for Orphanet national hubs In a later stage it is expected that the helpdesk will be assured by the Clinical Terminology Center (competence center/network focused on use of clinical terminologies in Information Systems within the institutions of the National Health Service). - IT official helpdesk to be developed. (SNOMED and Orphanet interoperability will be an important help to achieve this action)	Jun. 22 (ongoing) Q1 23 (planned) 2023 (planned)

Further Activities:	
Annual report on the implementation of the Rare Disease Card:	
Translation in English to be made available on the Orphanet national website	
- The 2021 Annual report on the implementation of the Rare Disease Card was published online on Directorate-General of Health website	Jul. 14th 22 (done)
- An English translation of the Report is ongoing	Dec. 22 (planned)
- Also an English translation of the Report to be published on Orphanet national website	Dec. 22 (planned)
Promotion visibility of ORPHAcodes:	
Expert Centres not integrated in any ERN or Reference Centre will be encouraged to use ORPHAcodes	2023 (planned)
Maintain an up-to-date translation of Nomenclature ORPHA:	
Provide an updated translated Portuguese version of ORPHAcodes to the ERN and nacional Reference Centres	
- Routine translation activities and their clinical validation have been maintained throughout the year	Jan.-Dec. 22 (ongoing)
- An extensive review and harmonization of nomenclature clinical terms translation has been taken place	Sep.-Oct. 22 (ongoing)

National hub of Slovenia

The national hub of Slovenia is located at the University Medical Center Ljubljana and consists of the following members:

Luca Lovrecic (MD, PhD, Assist. Prof. of Human Genetics in Med. fac., National coordinator Orphanet Slovenia)

Borut Peterlin (MD, PhD, Prof. of Human Genetics in Med. fac.)

Nuša Trošt (PhD in biosciences, Information scientist Orphanet Slovenia)

Ana Nyasha Zimani (MD)

Esada Kerić (administrative support)

Their activities additionally to the Orphanet tasks include participation in the creation of the Slovenian national plan for rare diseases, together with the Ministry of health, clinical and laboratory work, participation in a national RD registry and participation in European projects (e.g. eHealth).

National action plan of Slovenia

PLANNED ACTION	TIMELINE
Trainings/Workshops: Meeting with clinical and laboratory geneticists at Clinical institute of Genomic Medicine, UMCL (CIMG, UMCL) – the main national institution in the field of diagnosing rare diseases. Basic training on Orphanet coding dedicated for genetics team (medical & laboratory, including reserchers, and registry) Mandatory trainings will be held for personel of all ERNs (online/in person). The trainings will start after the implementation of ORPHAcodes in the official hospital information systems in pilot department (CIMG; UMCL) National meeting – ERN-HUB, all ERN national coordinators and collaborators.	September 2022 – done – main result: discussed implementation of ORPHA codes into the HIS Depending on the timeline of implementation of the codes into the HIS Same After HIS implementation and first results
Networking: Our Orphanet team is part of the National Advisory Board on rare diseases and ERN-Hub, having links to all ERN partners. The use of ORPHAcodes is mentioned in the national action plan for RD. We have been collaborating closely with the Ministry of health in the generation of a	ongoing

minimal data set for National registry for rare diseases which is now in place and ORPHAcodes are mandatory in this registry.	
Helpdesk Implementation: Hopefully we will manage to have additional RD specialized team by the end of 2022 which will include helpdesk activities.	December 2022
Further Activities: Our aim is to create a system/team for all these activities to become an ongoing, self-sustained, permanent action	ongoing

National hub of Spain

The national hub of Spain is formed by the team of Orphanet Spain and is located at the Center for Biomedical Network Research on Rare Diseases, CIBERER. It is integrated by the following members:

Francesc Palau (Medical doctor, PhD, National Coordinator of Orphanet Spain)

Virginia Corrochano (PhD biology, Project manager of Orphanet Spain)

María Elena Mateo (Grad. Information Sciences, Information scientist of Orphanet Spain)

Noelia Millán (UG. Information Sciences, Information scientist of Orphanet Spain)

Besides the tasks related to Orphanet´s daily activities, the team leads the scientific coordination of the RD Strategy of the Spanish NHS, and contributes to CIBERER by being involved in management tasks related to national and international projects as well as being responsible for the attention to the consultations received by CIBERER from patients and patients organizations. In addition, Francesc Palau leads a research group centered in the genetics and physiopathology of neuromuscular diseases and neurodegenerative disorders at the Sant Joan de Deu Hospital (Barcelona).

National action plan of Spain

PLANNED ACTION	TIMELINE

Trainings/Workshops: Identifying and contacting HCPs Preparing a survey to gather information about the state-of-play that will guide actions After assessing HCPs specific needs, organize online training sessions Translate new material produced in the frame of the EJP-RD nomenclature training courses Organize 2-3 *"Orphanet coding and nomenclature"* training sessions in 2022-23 Plan some onsite visits as required.	June: - contact with HCPs - elaborate and send survey July: - survey analysis - update EJP-RD training material - elaborate training proposal September 22 – March 23: - training sessions
Networking: Elaborate project flyer or fact sheet. Contact all HCPs linked to ERNs to raise awareness and assess their ORPHAcode implementation degree and specific needs. Contact Spanish participants in RD-CODE and main regional RD registries to set up a working group that provides support to Spain's help-desk. Dissemination and political incidence, through contact with main patient federations in Spain and through participation of the national hub members in the Scientific Coordination of the Spanish National Rare Diseases Strategy.	June – November: - Flyer - Mailing and call round - Spanish National Rare Diseases Strategy meetings
Helpdesk Implementation: Set up a Github account. Elaborate a basic information doc (*or translate if available*) regarding the Helpdesk and disseminate among HCPs. Constitute a working group led by the Orphanet national team. Training of the team to be in charge of answering the queries.	June 22– March 23: - The times will depend on when the ticketing system is ready for use.

National hub of Sweden

The national hub of Sweden is represented by the team of Orphanet Sweden and is located at the Centre for rare diseases at Karolinska University Hospital, Stockholm. It consists of the following members:

Rula Zain (National coordinator of Orphanet Sweden)

Terese Bodérus (Information scientist of Orphanet Sweden)

Elsa Ekblom (Information scientist of Orphanet Sweden)

Besides their work on Orphanet they are also active in supporting rare disease expert teams and contributing to a national infrastructure for care of rare disease patients, participating in the national action program for rare diseases as well as in patient empowerment.

National action plan of Sweden

PLANNED ACTION	TIMELINE
Trainings/Workshops: A presentation of Orphanet focusing on ORPHAcodes and the need for implementation of a specific coding system for rare diseases has been prepared. The ORPHAcode presentation is planned for the following events: 1. Course on rare diseases for medical doctors 2. Seminar on rare diseases for dental nurses 3. Undergraduate course for medical doctors in genetics	February 2022 – Completion of the presentation for future use – Done March 2022 – Presentation for medical doctors - Done June 2022 – Presentation for dental nurses. (Unfortunately postponed to the fall)
Production of a wider encompassing presentation on Orphanet, ORPHAcodes, rare disease and ERNS	November 2022 and ongoing
Networking: Support for specialist staff within the dental care profession regarding rare diseases is being provided. (They have been experiencing	Ongoing

difficulties with identifying patients with rare diseases.) We have contact with the hospital IT-dept and are waiting for feedback regarding the estimated cost of making ORPHAcoding a possibility in the existing HIS. We have received information regarding the technical possibilities and a cost estimate for implementing ORPHAcodes in the HIS. Since a policy decision is required on the regional level, we have initiated contact with the unit in charge of coding. The IT department responsible for implementing the ORPHAcodes into the electronic care system has said that we have to pilot the ORPHAcode implementation internally at Karolinska University Hospital firstly. Then we can start thinking about and planning the expansion to other hospitals, since this is the easiest route to take for the implementation to be successful. Within the OOC Nordic countries, exchange of information and documents is ongoing.	Planned for May/June 2022. Postponed until we can production and collection of all necessary documents and information will be completed. Done Done Ongoing Ongoing
Helpdesk Implementation: The helpdesk implementation in progress, but will probably not be fully active until we get ORPHAcodes launched in the electronic system at Karolinska university hospital.	Planned for 2023
Further Activities: Production of code-sheets containing ERN-specific ORPHAcodes and corresponding ICD-10 codes for use by regional and national expert teams. From this a subset only containing the codes relevant for each expert team will be sent out. As there were further interest in more encompassing cheat-sheets that were not specific to each expert centre, the production of	April-May 2022: An Excel template, which autofill ORPHAcode and ICD-10 corresponding to the activity of each team. June-August: Specific code-sheets for all regional and national teams November 2022 - ongoing

complete ERN sheets have begun, one for each ERN. At this stage, ERN-RITA has been finished and sent out to the interested parties.	
Production and distribution of pamphlets in Swedish and English with information about ORPHAcodes for the general public.	May-August 2022
We are currently in communication with the media department at Karolinska to display the ORPHAcode pamphlets locally on media screens within Karolinska University Hospital.	August 2022 - ongoing
Production of supporting documentation for regional implementation of ORPHAcodes.	June-September 2022

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 summarised in the NAPs presented.

Numerous trainings and workshops have already been conducted or prepared. 3 national teams have conducted surveys to identify the needs of ERN network members. A number of national hubs have translated training materials and videos into their respective national languages to make the trainings more effective. A lot of national teams have also developed their own training materials to tailor the trainings to the needs of the target groups.

Extensive activities have been undertaken to raise awareness of RD coding among both ERN network clinics and national health administrations and to communicate the benefits of using ORPHAcodes.

From the 13 national hubs, 11 have already implemented a national helpdesk. 7 of them have also created a github account which enables transparent traceability of the requests as well as their forwarding to the coordination team if their expertise is needed for answering the user´s questions. 3 more github accounts are in preparation, while in 2 countries implementation is currently not possible due to regulatory restrictions. The remaining 2 national hubs are preparing to implement their national helpdesks at the end of 2022 or in 2023.

A lot of further activities have been implemented and are still being pursued like creation of satisfaction surveys, cooperation with companies providing hospital and laboratory information systems, further development of ORPHAcoding data files, development of a dashboard which determines the number of patients per ORPHAcode, development of electronic registration forms of national RD registries, production of code sheets containing ERN specific ORPHAcodes corresponding with ICD-10 codes to mention just a selection.

The activities will be continued with the release of a lessons learned document until the end of the project in March 2023. The aim is to continue these activities in a follow-up project OD4RD2 beginning in April 2023 and to extend them to other participating countries.

Annex: National hub and national action plan of Switzerland

Switzerland is no official member of the OD4RD project but has been accepted as guest country. The Swiss team has participated in all OD4RD work package 4 activities and has build a national hub and developed a national action plan.

The national hub of Switzerland is represented by the team of Orphanet Switzerland, located at the University Hospitals of Geneva (HUG). It consists of the following members:

Dr. Loredana D'Amato Sizonenko (medical doctor, National coordinator of Orphanet Switzerland)

Martin Arles (Project manager of Orphanet Switzerland)

Béatrice Geissbühler (Information scientist of Orphanet Switzerland)

Besides their work for Orphanet they are also active in medical supervision of the rare disease helpline for the French part of Switzerland, participating in several committees (national coordination for rare diseases, steering board of the Swiss Rare Disease Registry, "Coding Working Group" created in the framework of the SRDR).

Their national action plan looks as follows:

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
First Orphanet nomenclature training at the national level already delivered. The attendants were mostly coders from the different Swiss hospitals and representatives of the Swiss Rare Disease Registry: the training had a good geographical coverage.	April 2022
The possibility of having more trainings on demand was discussed but none foreseen so far.	Ongoing
Networking:	
The network at the political and institutional level is already established, mainly with:	Ongoing

- National coordination of rare diseases (kosek) - Swiss Rare Disease Registry The objective is to strengthen it.	
Helpdesk Implementation: The monthly meeting of the "Coding Working Group", launched in July 2021 in the framework of the Swiss Rare Disease Registry (SRDR), includes helpdesk activities where national coders bring doubts and questions that are directly answered by the Orphanet Swiss team or transmitted to the Orphanet coordinating team. They have already been requested by several teams in charge of implementing the nomenclature at different Swiss hospitals.	Monthly meetings Ongoing
Further Activities: Communication activities to inform the clinicians on the Orphanet nomenclature and the SRDR at the HUG. The Orphanet Swiss team is directly involved in the practical implementation of the Orphanet nomenclature at the HUG, dealing with the professionals and performing a quality control on their declaration.	June 2022 Should be a routine in the first semester 2023