Deliverable 4.2

OD4RD2 National Action Plans Year 1

24.06.2024

This document represents deliverable 4.2 of the OD4RD2 project, which has received funding from the European Union. It contains the national action plans by the members of OD4RD2 work package 4, developed from April 2023 to March 2024. The document has been produced by the leaders of the OD4RD2 - Work Package 4. The OD4RD2 project has been launched in April 2023 for a 33 months period.

More information on the activities of the OD4RD and OD4RD2 can be found at www.OD4RD.eu

Disclaimer:

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.

Despite the fact that the Orphanet nomenclature already exists and is freely available in computable formats, the real-life implementation in health information systems is challenging due to the heterogeneity of coding systems and practices, and tools. The experience from the mentioned RD-CODE project taught us that local support in local language for coders and technical teams is necessary to achieve proper implementation in compliance with good practice guidelines for coding and to therefore increase data quality and comparability. To address those needs, it is important to maintain the Orphanet nomenclature of rare diseases, and to improve it building on the well organised and structured rare disease expertise laying in European Reference Networks (ERNs), to increase its interoperability with other terminologies in use in different countries and in registries, and to contribute to the adoption and implementation, starting by the hospitals involved in ERNs. Building on the well-established Orphanet network, the latter is facilitated by setting up a Network of National Orphanet Nomenclature Hubs, nowadays acting in 19 member states.

Objective of OD4RD work package 4

The objective of work package 4 (WP4) is to ensure coordinated local support for the local implementation of ORPHAcodes in national HCPs hosting ERNs and national HCPs linked to ERNs by establishing Orphanet national nomenclature support hubs.

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 ORPHAcodes 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 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 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 WP4 of the OD4RD1 project 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 health care providers (HCPs) hosting ERNs and national HCPs linked to ERNs. This template is also used during the OD4RD2 project and is 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 OD4RD2 WP4 website. During the bimonthly 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 March 31st 2024. For the achievments of each national nomenclature hub please consult deliverable 1.3, evaluation report: (https://od4rd.eu/03-deliverables).

Participating countries:

Austria, Belgium, Bulgaria, Czech Republic, Estonia, Finland, France*, Germany, Ireland, Italy, Latvia*, Lithuania, Netherlands, Norway, Poland, Portugal, Romania*, 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; Latvia officially joined the OD4RD2 project January 1st 2024; Romania was still not able to carry out activities as of June 2024, discussions are ongoing to assess their capacity to start their activities as of September 2024 for the last year and a half of the project.

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)

Ursula Unterberger has also been responsible for the implementation of ORPHAcoding in national centers of expertise on behalf of the Ministry of Health (MoH) over the last two years. In addition to Orphanet activities, Till Voigtländer is coordinating the European Joint Action JARDIN. Ursula

Unterberger is co-leading work package 7 on national reference networks and undiagnosed diseases in this Joint Action.

National action plan of Austria

PLANNED ACTION	TIMELINE
Trainings/Workshops: Implementation of ORPHAcodes in Austrian Centers of Expertise (CoE) and Associated National Centers (ANC) in ERNs was/is ongoing, and advice has been provided to involved stakeholders. Trainings have been provided to the teams at CoE/ANC.	Trainings started Q4 2023
Networking: We are working on the implementation of ORPHAcodes on behalf of and together with the MoH, and in this context collaborating with several clinical departments and hospitals to implement ORPHAcoding on a pilot basis in a number of CoE and ANC. 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.	ongoing
Helpdesk Implementation: Orphanet Austria is taking questions and requests, as well as providing assistance on ORPHAcoding under the email address ursula.unterberger@meduniwien.ac.at Use of the helpdesk is promoted among CoE/ANC who are already using ORPHAcodes. A separate section was installed on the new Orphanet country page.	ongoing

Further Activities: Orphanet Austria supports the implementation of ORPHAcodes in Austria on behalf of the MoH (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). A coding list adapted from the Orphanet nomenclature pack is provided to the MoH. ORPHAcoding will be mandatory for CoEs beginning 2026 (this was achieved by the MoH in close collaboration with Orphanet Austria).	ongoing

National hub of Belgium

The Belgian OD4RD hub is managed by the Belgian Rare Diseases Team hosted by Sciensano, the Scientific Institute of Public Health. It is composed of two staff members:

Annabelle Calomme (Master in molecular biology, information scientist for the Orphanet Belgium database, OD4RD1/OD4RD2 Belgian Hub project manager)

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

The Belgian Rare Diseases team has more than 15 years of experience with performing tasks related to Orphanet and has expertise in developing and implementing other RD projects that are part of the Belgian Plan for Rare Diseases. The service that hosts the team manages national patient registries as the Central Registry of Rare Diseases and registries for specific rare diseases, e.g. cystic fibrosis, neuromuscular diseases and rare bleeding disorders. The team also participates in several European RD projects related to rare diseases (EJP-RD, JARDIN JA, etc.). The Belgian RD team is in charge of translating the rare diseases nomenclature into Dutch (OrphaDutch project).

National action plan of Belgium

PLANNED ACTION	TIMELINE
Trainings/Workshops: • **Delivery of trainings on implementation and use of ORPHAcodes to university hospitals with a RD function:**	

UZ Leuven: online, in English and Dutch (two-hour session, basic training) IPG-GHdC: on site, in French and English (three-hour session, basic training) UZ Gent: online, in English and Dutch (two-hour session, basic training) • **Presentation on Orphanet and OD4RD for a GP working on a RD awareness project in collaboration with the "Société Scientifique de Médecine Générale" (SSMG)** • **Presentation on coding of undiagnosed RD patients for the Working Group Central Registry Rare Diseases (CRRD)** • **Update of the PPT document used for the OD4RD trainings**, with description of new documents and tools (OD4RD wikipage, RDK™, etc.) • **Webinar on Orphanet and RD coding for GPs from the SGPs Network**	15 May 2023 (session 1, in EN), done Participants: clinicians, RD coders and coordinator RD (n=13) 06 Jun 2023 (session 2, in NL), done Participants: clinicians, RD coders and coordinator RD (n=8) 24 Nov 2023, done Participants: clinicians, RD coordinator and IT specialists (n=8) 07 Dec 2023, done Participants: clinicians, study coordinators and data managers (n=7) 08 Jun 2023 (session 1, in FR), done 27 Oct 2023 (session 2, in FR), done 1 participant (Dr A.M., scientific coordinator of the SSMG Rare Diseases project) 30 Nov 2023, 04 Dec 2023 (2 sessions), done Participants: clinicians, RD coordinators (n=10) Dec 2023, done 01 Mar 2024, done (n=27)
Networking: • **Meetings with HCPs (RD Functions)** to promote the use of ORPHAcodes • **Promotion of ORPHAcodes to the Working Group ERN & RDs** (ERN members, clinicians from university hospitals, RD coordinators, PO representatives, FPS Health) • **Creation of a newsletter** sent to the coordinators of the 8 Belgian university hospitals with a RD Function, as well as other key people active in the RD field (reminder of the opportunity to benefit from trainings on RD-coding, promotion of the OD4RD GitHub, etc.)	08 Sep 2023; 01 Dec 2023, done 16 Oct 2023; 11 Dec 2023, done 8 Jan 2024, done (n=34), done

• **Promotion of the Orphanet website and the ORPHAcodes via a wobbler containing an overview of resources available in Belgium in the field of RDs** (in collaboration with RaDiOrg, the Belgian umbrella patient organization for people living with a RD). This "marketing tool" was distributed on the occasion of Rare Disease Day 2024 by two Belgian information magazines intended for both GPs and specialists ("Artsenkrant" and "Le Journal du Médecin")	Week of 29 Feb 2024, done
• **Participation in "Rare Diseases in the EU: Joint Action shaping the future of ERNs" JARDIN kick-off meeting**	06-08/03/24, Brussels, done
• **Collaboration with the Terminology Centre of the FPS Health regarding the coding of RD patients in the Belgian electronic health records**	Throughout the year
Helpdesk Implementation: Sciensano RD team members act as a national helpdesk for RD coding issues. A process is in place to manage questions addressed by ORPHAcode users by phone or email (orphacodes.belgium@sciensano.be). If necessary, a conference call is organized to deepen the discussion with the experts.	
• **19 questions addressed to the helpdesk** (Coding: 9, Nomenclature: 6, Others: 4), received from clinicians (10), coders (4), managers of patient registries (3), patients (2)	Replies provided by the helpdesk; creation of 12 GitHub posts (GitHub #68, 72, 102, 117, 129, 142, 150, 155, 165, 175, 187, 192)
• **Promotion of the GitHub to RD experts**	In the newsletter sent on 18 Jan 2024 (n=34); during the OD4RD trainings, done
• **Participation in Nomenclature/Coding Open sessions for National Hubs**	6 sessions (19/10/23, 22/11/23, 14/12/23, 18/01/24, 22/02/24, 21/03/24), done
Further Activities:	
• **ERN-linked HCPs survey preparation (deliverable n°12, survey on the RD coding practices within ERN-centres):** identification of the Belgian RD experts to	Nov-Dec 2023, done

be invited to participate, creation of an online Lime survey (FR, NL, EN)	
• **ERN-linked HCPs survey promotion:** email sent to the coordinators of the RD Functions to inform them of the objectives of the survey and ask them to promote it in their hospitals	14 Dec 2023, done (n=23)
• **ERN-linked HCPs survey launch:** sending of invitations	18 Dec 2023, 23 Jan 2024 (reminder), done (282 emails sent to (sub)-representatives of 135 Belgian ERN-units and to the 8 RD coordinators)
• **ERN-linked HCPs survey:** analysis of the replies	Feb-Mar 2024, done
• **ERN-linked HCPs survey:** results report	Mar 2024, done
• **Creation of the OD4RD project description for the Sciensano website**	Jan 2024, done
• **Contribution the RD Memorandum to Belgian political parties**, in collaboration with RaDiOrg and the Collège belge de génétique et des maladies rares	Jan-Feb 2024, done
• **Promoting the use of ORPHAcodes to health ministry and other national health institutions** (non-university hospitals, ERNs expert centres, Flemish Network Rare Diseases, etc.)	Throughout the year
• **Creation of a take home leaflet with description of the Belgian hub (FR, NL)**	Postponed (Q3 2024)

National hub of Bulgaria

The national hub of Bulgaria is built of:

Prof. Rumen Stefanov, medical doctor, professor in public health at the Medical University of Plovdiv (Bulgaria), Director of Institute for Rare Diseases, Bulgaria and President of ICORD: 2023-2025. He has successfully launched projects on RDs in Bulgaria and Eastern Europe: the Information Centre for Rare Diseases and Orphan Drugs (2004), the Eastern European Conferences for RDs (2005-2011), the "RareDis" Medical Centre (2009), the Centre for Health Technology Assessment and Analysis (2013), as well as RDs initiatives in Russia, Romania, Turkey, Ukraine and other countries in the region. At national level, he is currently the chair of the Committee for rare diseases at the Bulgarian Ministry of Health. He has chaired expert working groups, responsible for the creation and implementation of the

Bulgarian national plan for RDs (2006- 2008) and the Ordinance for establishment of a registry and expert centers for RDs in Bulgaria (2013).

Elena Mitova, MD, MBA is medical manager, Institute for Rare Diseases, BAPES (Bulgaria) and project manager for Screen4Care, OD4RD2. She has master's degrees in medicine (MD), specialty in pediatrics, pediatric nephrology and business administration (MBA). Elena has 20 years of experience in healthcare, in regulatory affairs, sales, marketing, public affairs and general management, as well as 13 years' work in academia – chief assistant professor in the University Children's Hospital, Nephrology department, Sofia.

Kostadin Dimitrov, MD is a project manager at BAPES, Institute for Rare Diseases for OD4RD2. He is a medical doctor with four years of clinical experience, a member of the Bulgarian Medical Union and the Bulgarian Scientific Society of Public Health. Currently pursuing a PhD in public health at the Medical University of Plovdiv.

Georgi Stefanov is project manager at BAPES, Institute for Rare Diseases (Bulgaria). He has master's degrees in medicine (MD), business administration (MBA), PhD in public health, clinical and academic experience and has passed successfully courses on Innovation, IESE, Spain and Sharpening Strategic Skills, INSEAD, France. He initiated and led under the auspice of the Pharmaceutical Group of EU the development of pharmacy guidance for patients with pain (2012). He has been member of the EU pharmaceutical working group contributing to the EC & EMA Biosimilars in EU - Information guide for HCPs (2017), Screen4Care, BUR-EB and OD4RD2 EU funded projects, National rare disease awareness campaign "Like All of Us", HTAs and rare disease partnership projects.

BAPES, as part of Institute for Rare Diseases, is situated in Medical center "RareDis", Plovdiv and has an affiliate in Sofia.

Their additional tasks are the close collaboration in the RD field with medical professional, patients, health authorities, caregivers and industry, participation in international and national conferences dedicated to rare diseases, including HTA trainings for health authorities, academia, patient representatives and industry, participation in 10 national rare diseases registries and numerous European RD projects.

National action plan of Bulgaria

PLANNED ACTION	TIMELINE
<u>Trainings/Workshops:</u> 1) Information meetings with chief of departments of the current 24 RD Expert reference centers, aiming to highlight the relevance of ORPHAcodes and assess specific needs. We will offer meetings for each department or smaller groups within the departments if requested.	1) Sept – Dec 2023

2) Launch OD4RD2 session during XIV National conference for Rare Diseases and Orphan Drugs: 29-30 Sept 2023, targeting expert centers participating in the conference	2) 29-30 Sept 2023
3) Trainings/Workshop specifically targeting clinicians and other HCPs involved in Expert centers: Basic/Advanced trainings provided by Orphanet in 2023, adapted to local needs	3) Jan – Apr (will continue beyond Apr) 2024 / ongoing
Networking:	
1) Survey on the use of ORPHAcodes and bottlenecks/challenges met by stakeholders (utilizing Spain best practice and adding country specific items). Assessment of specific needs per expert center and plan activities accordingly.	1) May - Oct 2023: Launched, ongoing
2) Meetings with responsible stakeholders: Ministry of Health, National Health Insurance Fund, hospital managers, for advocating ORPHAcodes towards national decision-makers, as well as with software developers and National Aliance for Rare Diseases patient representatives to highlight the relevance of ORPHAcodes and to promote OD4RD2 WP4 objectives	2) Jan – Apr (will continue beyond Apr 2024) / ongoing
Helpdesk Implementation:	
1) Map other countries examples / solutions and deciding on solution	1) Oct 2023
2) Setting up the technical necessities for the solution	2) Till Dec 2023
3) Training of the team by Orphanet to be in charge of answering the queries	3) Till Dec 2023

4) Launching the helpdesk	4) Q1/2024 depending on the system readiness
Further Activities: 1) Publication in medical journal „Rare Disease and Orphan Drugs "(ISSN 1314-3581) related to 2023 survey results	1) H1 2024
2) Map, select and adapt from Orphanet, Norway, Spain, Italy, Belgium, other countries best practices like information brochure, fact sheet, take home leaflet, satisfaction survey, video in local language, etc.	2) Q1 2024
3) Distribute created materials: information brochure, fact sheet, etc., through www, mailing to targeted stakeholders	3) During 2024 / ongoing

National hub of Czech Republic

The national hub of the Czech Republic is built by two members of the Orphanet Czech Republic team, located at the Department of Biology and Medical Genetics of Praque University hospital, and two 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 team 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: Further trainings at university hospitals according to their needs, especially in Prague and Brno (all ERN centers are based there). We also offer prepared materials from previous trainings and workshops online.	Offering and negotiating with more HCPs
Networking: Two institutions already involved in the project: • National Coordination Centre 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 Interdepartmental Commission for Rare Diseases at Ministry of Health (MEKOVO). Cooperating with Czech Association for Rare Diseases and other patients organizations on promotion of ORPHAcodes among patients. We are offering support to vendors/developers of health information systems.	Cooperation is already established on level of Interdepartmental Commission, this year we will try to promote ORPHAcodes more on genetic conferences and to IT companies related to health information systems. Ongoing Ongoing Ongoing
Helpdesk Implementation: We are already providing support over e-mail and phone and with direct contact and demonstration with clinicians. Contacts are on local page at orphanet.cz. On this page we will offer a special contact form with some kind of issue/request tracking in local language to have it as user friendly as possible.	Already working and being used by our clinicians. First half of 2024.

Further Activities:	
• Updating the Czech translation of the nomenclature and providing this to health care providers and health insurance companies	Ongoing – whole year
• Promoting usage of ORPHAcodes at different national conferences (e. g. conferences of Society of Human Genetics)	Providing the coding table - June
• Promoting of ORPHAcodes during pregradual and postgradual education of health-care professionals	Continuous (eg. DNA diagnostics conference – May 2024)
• Promoting ORPHAcodes in local medical journal	Ongoing – whole year
• Promoting ORPHAcodes in local genetic textbook	First half of 2024
• Updating and promoting the translation of Human Phenotype Ontology (HPO) in Czech language	Ongoing – whole year
• Cooperation on creating pilot Czech Rare Disease registry	Ongoing – whole year
• Cooperation with companies providing hospital and laboratory information systems and providing them support	Ongoing – whole year
• Official methodology for ORPHAcoding in Bulletin of Czech Ministry of Health	Approved on 15th March 2024 by the minister and then published

National hub of Estonia

The national hub of Estonia consists of:

Vallo Tillmann, head of Orphanet Estonian team, Professor in paediatrics, University of Tartu, consultant in Paediatric Endocrinology, Tartu University Hospital.

Sille Vahtra, information scientist of Orphanet Estonian team, coordinator, Department of Paediatrics, University of Tartu.

They are situated at the University of Tartu, Institute of Clinical Medicine, Department of Pediatrics.

Their tasks besides the work for Orphanet are the support of ORPHAcode implementation at Tartu University, the teaching of doctors at TUH and different HCPs in Estonia where the coding is done by doctors, clinical work at TUH (VT - Consultant in Paediatric Endocrinology), data management of ENDO-ERN registries, coordination of the establishment of national RD plan and strategy (2016), the participation in EU projects (EURO-WABB) and in COMP work at EMA (VT - national representative at COMP), and the coordination of clinical trials (PREVALL and DIABIMMUNE longitudinal study).

National action plan of Estonia

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
• Presentation about the aims of OD4RD2 to the Chairs of ERN-s at Tartu University Hospital	• September 2023: **Done,** (representatives from 20 ERNs at TUH participated)
• Rare Disease Day meeting/webinar (presenatation on Orphanet and ORPHAcodes)	• February 2024: **not done** as the program of the Rare Disease conference on 29th Febr 2024 was too busy and on other main topic (Orphan drugs and their reimbursement)
• Trainings/workshop for doctors how to use ORPHAcodes for coding at TUH	• Within 2-3 months after ORPHAcodes are implemented into the system of Electronic Case Report of TUH: **ongoing**
• Presentation/workshop of ORPHAcodes implementation process and use at TUH to the other HCP in Estonia (North Estonian Regional Hospital, Tallinn Children‘s Hospital)	• Within 12 months after ORPHAcodes are implemented into the system of Electronic Case Report of TUH: **as scheduled**
Networking:	
• With IT-team of Tartu University Hospital (TUH) to implement ORPHAcodes at TUH	• Through the whole period : **ongoing**
• With Board members of other HCP-s in Estonia about the implementation of ORPHAcodes in their hospitals	• Within 12 months after ORPHAcodes are implemented into the system of Electronic Case Report (EHL) of TUH: **as scheduled**

Helpdesk Implementation: • Presentation about the role of helpdesk in the coding of rare diseases (introducing the experience from other countries) to the board of TUH • Presentation about the role of helpdesk in the coding of rare diseases (introducing the experience from other countries) to the CEOs or board members of HCP-s in Estonia who have implemented ORPHAcodes in their hospital	• Within 2-3 months after ORPHAcodes are implemented into the system of Electronic Case Report of TUH- **not done** • Within 12 months after ORPHAcodes are implemented into the system of HCP: **as scheduled**
Further Activities: • Providing the updated ORPHAcode nomenclature file to the IT- department of TUH for the update within the Electronic Case Report (EHL) system	• Annually in September: **ongoing**

National hub of Finland

The national hub team of Finland contains of

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

Iiro Toikka (background in political science, Development Manager)

Satu Wedenoja (medical doctor, Senior Medical Officer)

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 third Finnish national plan and stategy for RD.

National action plan of Finland

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
ORPHAcode information card (in Finnish, Swedish and English)	2023, done
Building up an online course of rare diseases (includes basic data on ORPHAcodes)	2023, done
Building up an online course of ORPHAcodes (in Finnish and possibly translated in Swedish and English)	2023-2024, in process
Visits/online meetings with RD sepcialists of the university hospitals: discussions on the status and possible obstacles related to the implementation of ORPHAcodes in clinical practice	2023, done
Networking:	
Piloting the use of ORPHAcodes as part of a patients report system in all university hospitals	2023, done
Increasing knowledge of ORPHAcodes through the national expert group and network of RD, a special meeting	2023, done
Providing statistics on the use of ORPHAcodes at the Finnish University Hospitals and the data transferred to the national Care Registry	2023-2024, not completed yet
Providing data on the activity of ORPHAcoding by different hospitals to the Finnish ERN members	2023-2024, not completed yet
Survey on the use of ORPHAcodes in the Finnish University Hospitals and ERN centres: motivation, obstacles, frequently asked questions	2024, done
THL Code Service	

-maintenance of ORPHAcodes -annual update of ORPHAcodes	2023-2024, done
Assessing possibilities for Nordic collaboration related to ORPHAcodes	2023-2024, in process
Helpdesk Implementation:	
Building up a national helpdesk for ORPHAcodes	2023, done
Writing the most relevant and difficult questions from the national Helpdesk to the GitHub Helpdesk	2023, done
Further Activities:	
Assessing possibilities to get ORPHAcodes obligatory in electronic patient report systems	2023-2024, in process
Advertising ORPHAcodes through a special national campaign by the Finnish Institute for Health and Welfare	2023-2024, not completed yet
Gathering data on emergency guidelines of RD and the related ORPHAcodes available online (in Finnish)	2023-2024, in process
Implementation of ORPHAcodes in all hospitals providing specialized care	2023-2025, in process

National hub of Germany

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

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

Dr. Kathrin Rommel (PhD, project manager of Orphanet Germany)

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

Sven Rütz (information scientist of Orphanet Germany)

Marius Guddorp (information scientist of Orphanet Germany)

Dr Stefanie Denger-Israel (PhD, information scientist of Orphanet Germany)

Orphanet Germany is part of the department „Code systems and registers“ 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 development and release of the Alphabetical Index of the ICD-10-German Modification, 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 “Coding of rare diseases”/ Meeting of the steering committee of NAMSE (National Action League for people with RD)	▪ 25.04.23 (21 participants)
▪ Coding training for CORD MI (Collaboration on RD/Medical Informatics Initiative Germany) members	▪ 20.10.23 (50 participants) (done)
▪ Presentation “Status Quo Alpha-ID-SE Version 2024” / Meeting of the steering committee of NAMSE (National Action League for people with RD)	▪ 14.11.23 (30 participants) (done)
▪ Presentation “Improving the visibility of Orphanet in Germany” / Meeting of the steering committee of NAMSE (National Action League for people with RD)	▪ 14.11.23 (30 participants) (done)
▪ Webinar on Rare Disease Day: “Navigation through the rarity”: RD patient pathways in the European Network for Rare Neurological Diseases	▪ 29.02.24 (92 participants) (done)
▪ Alpha-ID-SE training video (45 min., online since March 28th 2023)	▪ 1.252 accesses 01.04.23-31.03.24

Networking: ▪ CORD MI (Collaboration on RD/Medical Informatics Initiative Germany) ▪ NAMSE (National Action League for people with RD) ▪ Interaction with other governmental departments ▪ Interaction with ACHSE (Alliance of Chronic Rare Diseases)	 ▪ participating at final monthly CORD workshops (21.4.23, 19.5.23,16.6.23) (done) ▪ Participation in several Workshops/Meetings of NAMSE ▪ Throughout the whole year ▪ Throughout the whole year
Helpdesk Implementation: ▪ 41 questions addressed to helpdesk (Coding: 37, Nomenclature: 4, Others: 0) ▪ Questions by: Coders (3), Clinicians (14), IT personnel (4), Hospital managers (1), Controllers (16), Others (3)	 ▪ answered by national helpdesk, 1 question forwarded via GitHub
Further Activities: ▪ Further development of German Alpha-ID-SE data file (version 2024); ORPHAcode connection with diagnostic terms of Alphabetical Index: 17.369, inclusion of 7.022 different ORPHAcodes in data file;	 ▪ published on October 5th, 2023

National hub of Ireland

The national hub of Ireland consists of:

Prof. Eileen Treacy MD, FRCPC, FRCPI, FCCMG, Clinical Professor of Medicine in Rare Disorders, TCD; Orphanet Ireland's representative and project lead for OD4RDs-WP4; Clinical Lead National Rare Disease Office (Coordinating Hub for ERNs) and Orphanet Operating Committee member.

Geraldine Sweeney Providing project management support on OD4RD2-WP4; Business Manager, National Rare Disease Office

Aileen Timmons PhD, information scientist: OD4RD2-WP4**;** Orphanet Ireland and National Rare Disease Office

The Irish team is located at HSE National Rare Diseases Office (NRDO), Mater Misericordiae Univeristy Hospital, Dublin 7 and at HSE Acute Operations Division Heuston South Quarter, Dublin 8.

Besides their Orphanet responsibilities, the team works on the collaboration of national eHealth projects to implement ORPHAcodes, care pathway development for Rare Diseases and is the ERN Coordination Hub. They also provide the National Rare Diseases Information Helpline.

National action plan of Ireland

Background:

- Ministry (Department of Health) is currently working on developing Ireland's second plan for Rare Diseases.
- To date, ORPHAcodes have not been included in Health Information Systems (HISs) in Ireland. SNOMED-CT is the coding system currently being implemented at national level for primary and secondary data usage.
- The national electronic data strategic plan is in development to align with the European Health Data Space initiative and the introduction of a unique patient identifier.

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Project Initiation/Planning meeting for Irish OD4RD team	Aug 2023 (complete)
Attendance at training for trainers sessions - 2 Information Scientists attended	Sept 2023 (complete)
Stakeholder Presentations / Information Sessions on ORPHAcodes	
National Genetics and Genomics Office (NGGO)	Dec 2023 (complete)
Childrens Health Ireland (CHI) - Electronic Health Record Project Team	Dec 2023 & Jan 2024 (complete)
Royal College of Surgeons Ireland (RCSI) – Genetics Society (30 participants)	February 2024 (complete)
University College Dublin (UCD) – Medical Students (35 participants)	March 2024 (complete)

DIS Stockholm – American undergradatestudents (37 particpants)	March 2024 (complete)
<u>Networking:</u> **National Genetics and Genomics Office (NGGO)** In 2023 Ireland developed a new strategy for National Genetics and Genomics Medicine in Ireland. The National Rare Diseases Office (NRDO) and NGGO are working together on shared objectives such as the implementation of ORPHAcodes for the NGGO Test Directory results database **Childrens Health Ireland (CHI)** are developing an electronic health record (EHR) with EPIC (Electronic health record provider). • The NRDO are meeting with CHI to advise on the importance of including ORPHACodes in the planned new CHI EHR • Meeting held with CHI, The Netherlands Orphanet team and EPIC to learn from the Netherlands experience of implementing ORPHAcodes in EPIC **Collaborations with ERN Irish leads to support the implementation of ORPHAcodes to ERNs and national registry development** Currently targeting: o ERN ERKNet o ERN Guard Heart o ERN EYE o ERN-ENDO	Dec 2023 (ongoing) Dec 2023 (ongoing) Feb 2024 (complete) Jan 2024 (ongoing)
<u>Helpdesk Implementation:</u> **Description:** The helpdesk is a function of the NRDO. The Information Scientist for Orphanet manages queries raised by telephone or email on a part-time basis **Helpdesk** – Queries received The information scientist has received and responded to a number of queries in relation to	Feb 2024 (ongoing) Mar 2024 (ongoing)

ERN ORPHAcode matching and ORPHAcode and HPO alignment **GitHub (Ireland)** set up	 Sept 2023 (complete)
<u>Further Activities:</u> **Development of new Rare Disease Plan for Ireland** RD coding and implementation of ORPHAcodes is a key area of focus • NRDO prepared a position paper on 'Requirements for effective rare disease coding in Electronic Health Records and Registries in Ireland' to support stakeholder meetings and discussions • Presentation by NRDO Clinical Lead to national Rare Diseases Plan Steering Group on ORPHAcodes and Registry use. Further meeting planned for June 2024 • Meeting with Dept. of Health escalating the necessity of ORPHAcodes in the CHI HER **JARDIN WP6 National Care Pathways** • Ireland leading on Task 6.1 – to develop a sign posting tool for expertise and multidisciplinary care pathways for the more prevalent RDs based on 2020 study, with the use of ORPHAcodes ref: (Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, Murphy D, Le Cam Y, Rath A. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020 Feb;28(2):165-173). **Rare Diseases Research and Innovation Catalyst Award (HRB RDCat) 2023** This project aims to increase support and capacity for Rare Diseases pre-clinical, clinical, population health and health services research,	 Jan 2024 (ongoing) Mar 2024 (complete) Feb 2024 (ongoing) Apr 2024 (ongoing)

and to promote clinical trial readiness targeting the ERN sites • The NRDO is collaborating with RD Cat WP Leads where there is specific overlap with OD4RD such as WP2: Data Support **OD4RD Stakeholder Information / communication pack** • Development of OD4RD information for dissemination across stakeholders to introduce the project and to support upcoming information / training sessions • Circulated information to ERN clinical leads, co leads and hospital managers on ORPHAcodes and undiagnosed RDs **ERN HCP Survey** • Circulated ERN survey to 35 ERN clinical leads and co leads • Reminder communication issued • Helpdesk responding to queries as they arise and assisting CLs to complete the survey	Mar 2024 (ongoing) Mar 2024 (complete) Mar 2024 (in progress)

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)

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 the Italian healthcare providers, by training the professionals, many of which members of ERNs, on the Orphanet nomenclature and tools.

National action plan of Italy

PLANNED ACTION	TIMELINE
Trainings/Workshops: • **XXVI Congress of the Italian Society of Human Genetics (S.I.G.U.)** Orphanet booth. Presentation of the Orphanet team/OD4RD project. • **OD4RD2-branded training for the implementation ad usage of ORPHAcoding.** The course was planned for autumn 2023 but remains to be done. We are thinking of a smarter version of the training, easier to manage for the trainer and less time consuming for the participants.	Done. To be done
Networking: Italian Society of Human Genetics (S.I.G.U.) Italian Inter-regional Technical Board for Rare Diseases Ministry of Health Press agency O.M.a.R.- Rare Diseases Observatory: dissemination of OD4RD project aims National Centre for Rare diseases (Istituto Superiore di Sanità). The ELIXIR Italy All Hands Meeting, held on 28 November 2023, has been the occasion to get in touch with this institutional figure. The possibility to collaborate, in order to	Ongoing. To be done. 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 ORPHAcodes and to the Orphanet classification system. Ongoing. Ongoing.

give visibility to our respective events, has been discussed.	
Helpdesk Implementation: Dissemination of the Italian GitHub helpdesk via social media and OrphaNews Italy	Ongoing.
Further Activities: Preparation of informative material about the Orphanet activity and the OD4RD2 project for the S.I.G.U. Congress. The material prepared will be updated, if necessary, to disseminate our activity on future occasions.	Done.

National hub of Latvia

The national hub of Latvia is located at the Childrens University Hospital (CCUH) in Riga. It consists of the following members:

Madara Auzenbaha, MD, PhD, national coordinator Orphanet, pediatrician, chief physician in Rare Diseases

Gita Taurina, MD, senior adviser Orphanet, medical geneticist, head of Medical Genetics and Prenatal diagnostics clinic

Lauma Vasilevska, MD, information scientist Orphanet, pediatrician with competence in rare diseases

Inese Ledina, MHRM, BS psych., CM, rare disease coordination center coordinator, helpdesk

Anna Kursa, business Psychology BS, information scientist Orphanet, assistant to the Head of Medical Genetics and Prenatal diagnostics clinic

Their main task besides the work of Orphanet is the involvement in the national rare disease registry.

National action plan of Latvia

PLANNED ACTION	TIMELINE
Trainings/Workshops: Meeting with the Ministry of Health and National Health Service: Presentation on the importance	Q2

of ORPHAcoding and current situation in Latvia and its inclusion possibilities in E-health	
28.02. "ORPHAcoding Awareness Day" (in collaboration with The Rare disease alliance)	28.02.2024
On-site training in university and regional hospitals of Latvia: Orphanet and coding of rare diseases (n=4)	Q4
Presentation at Riga Stradinš University/Rare Disease Forum	29.02.2024
Online session for general practitioners (n=3)	Q4
Networking:	
Implementation of ORPHAcodes in CCUH inner information system	Q1-Q2
Additional demand for ORPHAcoding in case of usage specific to ICD10 codes	Q4
CCUH presentation on the importance of ORPHAcode usage	08.03.2024
Handouts about ORPHAcoding	Q4
Helpdesk Implementation:	
Creation of the helpdesk	Created
Dissemination of information	Q4
Further Activities:	
Create a video “Coding Recommendations” in Latvian	Q3
Modify document "Registration cards for patients with congenital malformations" (must be approved in the regulations of the Cabinet of Ministers)	Q4
Create WG on new Rare disease registry	Q2

National hub of Lithuania

The national hub of Lithuania is located at Vilnius University Hospital Santaros Klinikos (VUHSK). It consists of the following members:

Birutė Tumienė, Assoc. professor, MD, PhD, Orphanet Lithuania national coordinator, Orphanet Operating Committee member, head of the Coordinating Center for Rare Diseases, VUH Santaros Klinikos, head of the Clinical Genetics and Genomics Department, VUH Santaros Klinikos, ERN Board of Member States, LT Representative, Chair, European Joint Program on Rare Diseases, Pillar 3, co-leader IRDiRC Diagnostic Scientific Committee, member European Society of Human Genetics, board member

Evelina Marija Vaitėnienė, MD, Orphanet Lithuania information scientist, clinical geneticist, Clinical Genetics and Genomics Department, VUHSK, senior specialist, Coordinating Center for Rare Diseases, VUHSK

Besides the work of Orphanet their tasks are clinical work (timely, efficient, extensive diagnostics; comprehensive, holistic, coordinated care provided by multidisciplinary teams; RD education and information, patient empowerment), participation in ERNs, RD registries and several European RD projects.

National action plan of Lithuania

PLANNED ACTION	**TIMELINE**
<u>Trainings/Workshops:</u>	
Trainings for the implementation of ORPHAcoding	6 training sessions for healthcare professionals
Webinar and supplementary informational resources on ORPHAcoding, accessible in the Lithuanian language	achieved/ongoing
ORPHAcoding trainings for graduates and postgraduates	ongoing
<u>Networking:</u>	
Meetings with Rare Disease Coordination Center, cooperation with ERNs Meetings with hospital management Meetings with Ministry of Health	Throughout the year
ERN survey	March 2024
Rare disease day events	3 events (2024-02/03)

	"United for Rare Diseases: from Patient to System" "Patient and family-oriented multidisciplinary integrated health care for patients with rare diseases" "Real help for a child with a rare disease" (on the occasion of the opening of the day ward for children's rare diseases)
Helpdesk Implementation: Helpdesk through e-mail and phone	 Full implementation achieved by 2023 Q4 Questions received by clinicians, IT team
Further Activities: ORPHA classification translation into Lithuanian Language ORPHAcoding system integration into the patient's e-health record system at VUH Santaros Klinikos Rare disease registry at VUH Santaros Klinikos ORPHAcoding integration into the National ehealthcare system Updating the content on orpha.net Lithuania website	 achieved 2023-04 achieved 2023-06 achieved, new functions added 2023-11 ongoing to be done

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)

Simone Dusseljee (BSc, project manager 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:	
Translation of the e-learnings developed by Orphanet Norway into English. Necessary steps:	
1. Basic translation of text in English	completed Q2/2023
2. Revision of text	completed Q4/2023
3. Implementation of text in e-learnings	ongoing Q1-Q3/2024
4. Testing e-learning	Q3-Q4/2024
Development of additional e-learning(s) – technical & clinical - specifically for the Netherlands: Script is in development, based on existing training material on ORPHAcodes, generated insight and material on registration in EHR and implementation of ORPHAcodes in the Netherlands.	
Necessary steps:	
1. Final script generated	Q2/2024
2. Implementation script in online e-learning tool	Q2/2024
3. Testing and updating e-learning	Q3/2024
4. Available for the Netherlands	Q4/2024
Networking:	
Regular meetings with the following organisations are in place:	
1. Dutch Ministry of Health, Welfare and Sport	Regular meetings; every 4-6 weeks
2. NFU (The Netherlands Federation of University Medical Centres)	Regular meetings; every 2-4 weeks
3. DHD (Dutch Hospital Data)	Regular meetings; monthly

With the following organisations a meeting is scheduled whenever necessary to discuss DT-SNOMED CT-ORPHAcode mapping issues: 1. DHD (Dutch Hospital Data) & Nictiz (The Dutch competence centre for electronic exchange of health and care information) Additional meetings: 1. At symposia organised by the NFU, DHD demonstrated the „ORPHA-viewer“ to encourage and improve registration of rare diseases in the EHR. 2. To improve effective registration with ORPHAcodes in the EHR relevant rare disease entries should be added to the different specialism-specific diagnosis lists. To enable the responsible Dutch Societies of Medical Specialists to achieve this goal the NFU and DHD started a campaign in which the help of officially designated expert centres is instrumental.	Meeting whenever necessary Completed Q1/2024; a symposium was held in every UMC, except Amsterdam UMC. For this UMC a digital meeting with the most important stakeholders was held. Q1-Q4/2024; various meetings will be held with expert centres to explain and coordinate the campaign.
<u>Helpdesk Implementation:</u> 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 - 20 questions were received regarding the nomenclature and 1 regarding a coding issue. The questions were submitted by clinicians (11), coders (4) and others (6). 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 DHD professionals are unable to answer ORPHAcode related questions, these are redirected to us - 1 question was 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	Implemented Routing from DHD to Orphanet NL helpdesk was provisionally implemented. After evaluation in Q4/2023 formal implementation was deemed not necessary. Account in place

this helpdesk entry point is not actively promoted - 1 question was received via this route.	
Further Activities: Since April 2023, 286 new links with ORPHAcodes have been created in DT. In total now 5500 ORPHAcodes are linked in the DT, representing ~87% of the 6305 disorder level ORPHAcodes.	
- Possibly the NFU/DHD campaign described in the „Networking“ paragraph will lead to the addition of new ORPHAcodes and disease terms in the DT.	Q2-Q4/2024
A „light“ version of the ORPHA-viewer will go live end of April 2024. Several discussions on „de-doubling“ of patients and technical adjustments to the ORPHAviewer led to delays and the going live without (for the moment) the „de-doubling“ functionality.	Q2/2024
Currently patients can only be coded with ORPHAcodes of the disorder level via the DT. Discussion is ongoing to also enable registration for group and subtype levels. Issues with the exact mapping between group ORPHAcodes and SNOMED CT entries are the major holdback at the moment. Different solutions are being explored.	Q2/2023 – Q4/2024
A local pilot will be conducted at the Radboudumc with the goal to make ORPHAcodes visible in the EPD.	Q1-Q4/2024

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, M.Sc., biomedical laboratory scientist, 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 (Registered nurse, Norwegian Registry on Rare Disorders)

Ingrid B. 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 Oslo University Hospital (OUS). We offer meetings for each department or smaller groups within the departments if requested: • The Lipid Clinic • Department of clinical neuroscience for children • Paedriatic Medicine outpatient clinic - endocrinology • Department of Ophthalmology • Department of Rheumatology • Division of Paediatric and Adolescent Medicine • Dept. of Neurology • Centre for Rare Diseases Online training/webinar on Orphanet nomenclature and the Norwegian registry on rare	 27.04.2023 (10 participants), done 29.08.2023 (1 participant), done 10.09.2023 (5 participants), done 20.09.2023 (1 participant), done 05.10.2023 (7 participants), done 08.11.2023 (25 participants), done 08.11.2023 (10 participants), done 05.02.2024 (20 participants), done 12.06.2023 (approx. 100 participants), done

disorders for network of professionals in the field of rehabilitation E-learning course on ORPHAcodes: • Contribute to the English translation of the Norwegian version. • Updated version of the Norwegian e-learning course	 18.04.2023, 14.12.2023, ongoing and throughout the year (planned launch: June 2024) 09.06.2023, done
<u>Networking:</u> Interaction with governmental departments and Health Authorities concerning measures in The Norwegian National Strategy for Rare Diagnoses: • the Directorate of e-health • the Directorate of Health • Regional Health Authorities South-Eastern Norway Regional Health Authority: Follow-up on request to implement ORPHAcodes in the new version of the EHR software (DIPS Arena) Western Norway Regional Health Authority: Meeting on implementation of ORPHAcodes in EHR – Fabry disease Outpatient clinic Participation in the annual Karen Helene Ørstavik's Dysmorphology Meeting, Telemark Hospital, Skien, Norway Participation in the Nordic conference on rare diseases, Stockholm, Sweden 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	 Ongoing (until June 2023) Ongoing and throughout the year 07.06.2023, done 23.-24-08.2023 (2 participants), done 2.-3.10.2023 (4 participants), done Postponed to Q4 2024, awaiting implementation of ORPHAcodes in the hospital EHR systems

• University Hospital of North Norway	
Helpdesk Implementation: Answer all requests to the national helpdesk (web form and e-mails) Participation in Orphanet Nomenclature and coding open sessions	 65 requests received, of which 45 answered by the national helpdesk and 20 forwarded to the coordination team via GitHub (done) Attended 6 sessions (19.10.2023, 22.11.2023, 14.12.2023, 18.01.2024, 22.02.2024, 21.03.2024), done
Further Activities: Development of new solution for ORPHAcodes in EHR (DIPS Arena): extensive collaboration (meetings and workshops) with the software provider DIPS Distribute information flyer about ORPHAcodes Survey on the experiences with ORPHAcodes and bottlenecks/challenges met by the professionals at Oslo University Hospital The Norwegian Registry on Rare Disorders: • Distribution of information flyer to patient organisations • Development of an electronic data collection form for clinicians in hospitals other than Oslo University Hospital ERN survey (Survey on the use of ORPHAcodes in ERN-linked HCPs): • Set-up of survey questionnaire • E-mail invitations to Norwegian ERN unit representatives (n=35) • Result analysis and report writing	 Q2 2023- Q2 2024 (approx. 20 meetings/ workshops), ongoing and throughout the year) Launch: October 2024 Postponed to Q4 2024, awaiting the new solution for ORPHAcodes in HER September 2023, done On hold, due to governmental evaluation of a National registry on rare diseases On hold, due to development of the ORPHAcodes implementation in EHR (DIPS Arena), which can be further developed to support automated data collection from EHR to the registry database January 2024, done 17.01.2024 (reminder sent 01.02.2024), done February-March 2024, done

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**
<u>Trainings/Workshops:</u> ORPHAcoding – dedicated training sessions and as a part of thematic courses for medical specialist in training (in total over 2000 participants)	
1. Meeting of the working team for the Rare Diseases Registry – 10 participants	17.05.2023
2. Course of Medical Centre for Postgraduate Education (MCPE) „Introduction to specialization in neurology" – ORPHAcoding in scope of lecture " Genetic testing in neuropediatrics" – 30 participants	14.06.2023
3. Course of MCPE „Introduction to specialization in clinical genetics", two lectures "Orphanet nomenclature and classification of rare diseases with ORPHAcoding" and "Rare diseases" – 4 participants	13.11.2023
4. Course of MCPE "Diagnostics and treatment of cardiac conduction and rhythm disorders in children", ORPHAcoding in scope of lecture	11.03.2024

"Genetic testing in hereditary heart rhythm disorders" – 23 participants	
5. Course of MCPE „Introduction to specialization in paediatric endocrinology and diabetology" – 12 participants	22.03.2024
6. Scientific monthly meeting of Children's Memorial Health Institute "ORPHAcoding" – 90 participants, including representatives of 7 ERN HCPs from the CMHI	12.03.2024
7. "Orphanet nomenclature and classification of rare diseases with ORPHAcoding" – Project of the Polish Medical Research Agency: Digital medicine centres, Wrocław (online) – 14 participants	23.04.2024
8. Individual trainings for physicians attending our Centre for Rare Diseases/Genetic Outpatient's Clinic in scope of their specialization training in clinical genetics/ neonatology/ diabetology/endocrinology	April 2023 – March 2024
<u>Networking:</u>	
1. Healthcare Policy Summit – Rare Diseases, lecture „Rare diseases – diagnostics and treatment"	05.07.2023
2. Round Table of the Medical Reason of State "Patients-Experts-System" - Quality of life in rare and neurological diseases	13.07.2023
3. IX Congress of Polish Society of Human Genetics, lecture: "Rare diseases – successes, pitfalls, challenges and perspectives"	03-06.09.2023
4. Conference of patient's advocacy group STOP Duchenne, lecture: "Registry of rare diseases" (including ORPHAcoding)	07.09.2023
5. Meeting of the Parliamentary Team for Rare Diseases: Rare diseases - opening balance 2024	01.02.2024
6. International Symposium Rare Disease Day 24.02.2024 - Warmia and Mazury, ERN ITHACA Olsztyn „We are the 300 million – we are all	24.02.2024

fighters!", lecture: "The wide and growing range of laminopathies" (including ORPHAcoding)	
7. Meeting of the Council of Experts on Rare Diseases for the Medical Reason of State, Round Table debate; Topics: Plan for Rare Diseases – current status and main challenges	28.02.2024
8. Rare Disease Day - National Forum for the Therapy of Rare Diseases ORPHAN	29.02.2024
9. Conference „Rare diseases – challenges for Poland 2024", Discussion on National Plan for Rare Diseases - implementation and challenges, Registry of Rare Diseases, Information Platform – role of Orphanet and ORPHAcoding in rare disease reporting	01.03.2024
Other activities	
1. Regular meetings of the Expert Councils on the Registry of Rare Diseases and Information Platform of Rare Diseases	Done, April 2023 – March 2024
2. Regular meetings of the Council of Experts of the Medical Ration of State	Done, April 2023 – March 2024
3. Cooperation with ERNs on mechanisms for the dissemination and updating of the nomenclature and Orphanet knowledge, identification of complementarities and gaps in the areas of knowledge of interest to individual ERNs	Done, April 2023 – March 2024
<u>Helpdesk Implementation:</u>	
Orphanet Poland National Helpdesk in the Memorial Children's Health Institute, Warsaw – solving problems and providing support with ORPHAcoding / answering questions on Orphanet nomenclature, classification and translation – contact via e-mail and phone (in Polish) (a.madej-pilarczyk@ipczd.pl; k.chrzanowska@ipczd.pl)	Implemented, active
GitHub account for Polish helpdesk	Implemented, active

Further Activities:	
1. Expansion of Orphanet resources in Polish language: Curation of nomenclature before release of the Nomenclature Pack 2023	Done, ongoing (April 2023 – March 2024)
Translation of Orphanet diseases summaries and full descriptions of RD to Polish (all nomenclature and over 2000 disease summaries have been currently translated and updated; approx. 250 disease summaries were translated and uploaded to POLOR in the period of April 2023 – March 2024)	Done, ongoing (April 2023 – March 2024)
2. Cooperation with e-Health Centre in creation of IT Platform for the Registry of Rare Diseases, Platform of Rare Diseases and Passport for patient with Rare Disease	Done, ongoing (April 2023 – March 2024)
The Information Platform of Rare Diseases has been already implemented on the governmental website: https://chorobyrzadkie.gov.pl/pl	
Contains:	
- Orphanet short description	
- Orphanet nomenclature (Polish translation) and short description of RD (partially translated to Polish) with respective link to Orphanet website and Orphanet logo	
3. Diagnostic recommendations in rare diseases	
Sections dedicated to each rare disease/group of rare disease contain following information:	Ongoing (April 2023 – March 2024), publication planned in 2024
Name of RD/group of RD, Incidence, Inheritance, Codes: OMIM, ICD10, ICD11, ORPHA, Recommended genetic diagnostics	
4. Constant cooperation with the Expert and IT team of the Ministry of Health in supporting the construction of the Polish Register of Rare Diseases with knowledge on rare diseases delivered by Orphanet (nomenclature, classification, ORPHAcoding, etc.)	

"Orphanet nomenclature and classification of rare diseases with ORPHAcoding" for Polish Ministry of Health, meeting dedicated to Polish Registry of Rare Diseases in scope of National Plan of Rare Diseases – 20 participants (Ministry of Health, IT team E-Health, policy makers)	Planned and realized 20.05.2024
5. Questionnaire on ORPHAcoding usage in Polish ERNs centres	Done, March 2024

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**
<u>Trainings/Workshops:</u> **Training sessions** Workshops will be held online and targeted to Reference Centres as ORPHAcodes users. 1. Revise existing material created for trainings by OD4RD project on ORPHAcoding and nomenclature	1. Dec. 23 (ongoing)

2. Translate and adapt new material for training sessions 3. Organize 2-3 online training sessions 4. Contact the Board of HCPs, namely Expert Centres members of the European Reference Network and Reference Centre Coordinators to send training invitations **ORPHAcodes and OD4RD project promotion materials** Flyer, infographics, and translated video versions to be distributed/emailed/presented in meetings and made available on the Orphanet national website, with the involvement of the National Commission of Reference Centres. 5.Revise and present Orphanet promotion materials in scientific meetings and conferences 6.Make available Orphanet promotion materials on Orphanet national website 7.Meet the new National Commission of Reference Centres after its nomination in 2023 8.Email promoting materials to Reference Centre Coordinators and collaborators	2. Dec. 23 (ongoing) 3. Dec. 23 (planned: sep/oct´24) 4. Dec. 23 (planned: sep/oct´24) 5. Jun-Dec.23(done) 6. Jun-Dec.23 (done) 7. Jun-Dec.23 (done) 8. Jun-Dec.23 (ongoing)
<u>Networking:</u> **OD4RD2 project formal presentation to the Ministry of Health** 1.Present OD4RD2 project and the national Action Plan to the New Ministry of Health. 2.Present OD4RD2 project and the National Action Plan, to the National Commission of Reference Centres and other Commissions on Rare Diseases **Promotion visibility of ORPHAcodes** 3.Expert Centres integrated or not in any ERN or Reference Centre will be encouraged to use ORPHAcodes	1. Jun-Dec.23 (planned: ´24) 2. Jun-Dec.23 (done) 3. Jun-Dec.23 (done)

Umbrella Patient Organization 4.Anual meetings between National Team and umbrella PO to be promoted in order to highlight the relevance of ORPHAcodes **RD Annual Event** 5.During the Rare Diseases Day celebrations the use of ORPHAcodes will be highlighted by the National Hub presentation	4. Feb.24 (done) 5. Jan.2024 (done)
<u>Helpdesk Implementation:</u> 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. 1.The Orphanet National Team routinely answers to requests/clarifications on Orphanet nomenclature within of Rare Disease Card new registries 2.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 Centre (competence centre/network focused on the use of clinical terminologies in Information Systems within the institutions of the National Health Service). 3.IT official helpdesk to be planned and promoted. (SNOMED and Orphanet interoperability will be an important help to achieve this action)	1. Jun-Dec.23 (done) 2. Jun-Dec.23 (ongoing) 3. Dec.23 (awaiting the requested IT implementation)
<u>Further Activities:</u> **Annual report on the implementation of the Rare Disease Card** 1.The 2022 Annual Report on the implementation of the Rare Disease Card to be published online on the Directorate-General of Health website 2.An English translation of the Report to be published on Orphanet national website	1. Jul.23 (done) 2. Sep.23 (done)

Maintain an up to date translation of Nomenclature ORPHA Provide an updated translated Portuguese version of ORPHAcodes to the ERN and national Reference Centres.	
3.Routine translation activities and their clinical validation have been maintained throughout the year	3. Jun-Dec.23 (done)
4.An extensive review and harmonization of nomenclature clinical terms translation has been taken place	4. Jun-Dec.23 (done)

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
<u>Trainings/Workshops:</u> Basic training on Orphanet coding dedicated for genetics team (medical & laboratory, including researchers, and registry)	September 2023-ongoing

Mandatory trainings will be held for personnel of all ERNs (online/in person). The trainings will start soon, since we finally implemented ORPHAcodes in the official hospital information systems in pilot department (CIMG, UMCL)	
Networking: Successful meetings with the Ministry of health to support our activities – project on education on RD (Orphanet team participates) Kleefstra conference in Ljubljana, 1-2.6.2023 – presentation 2.6.: Innovative approach towards the national organization of rare disease management – Slovenian National plan assist. prof. Luca Lovrecic, MD, PhD, Ljubljana UMC, Slovenia 14th Balkan Congress on Human Genetics & 9th Rare Disease SEE Meeting, Macedonia, 5-7th October	Ongoing May 2023 - CRP 2023 – 3.2.1 Topic title: Innovative education for the implementation of national plan for rare diseases June 2023 https://idefine-europe.org/conference/ Orphanet activities at our Dpt were mentioned October 2023 Luca Lovrecic presented: Innovative approach towards the national rare disease management – Slovenian National Plan (Orphanet activities have been emphasized)
Helpdesk Implementation: As we employed 1 administrative person (a person dedicated for RD team covering ERN-national hub and Orphanet support) we introduced a dedicated help desk line/e-mail for questions	We believe that Slovenia is too centralized and too „small“ for establishing a ticketing system and additional platform, therefore we started with a simple e-mail. If this proves to be insufficient, we will implement changes.
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

Spain’s National Hub is integrated by Orphanet Spain, hosted by the Center for Biomedical Network Research on Rare Diseases (CIBERER, Institute of Health Carlos III).

Team members are:

Francesc Palau (Medical doctor, PhD, national coordinator of Orphanet-Spain)

Virginia Corrochano (PhD in 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 it 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. The team has also participated in a working group in charge of translating the HPO terminology into Spanish, and it has collaborated with the MoH to implement ORPHAcodes in a terminology server. 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**
<u>Trainings/Workshops:</u> 1. Organize “Orphanet coding and nomenclature“ training sessions. 2. Translate any new training material produced by Orphanet-France or by any other partner. 3. Produce training material as needed 4. Plan some onsite visits as required.	1. Two sessions concluded (13th and 20th June 2023)* - 46 participants from 6 Hospitals and from RD Registries/ Health Authority Departments from 4 Autonomous Regions (Castilla-León, Murcia, Navarra and Valencia) 2.Flyers translated: National Hubs; ORPHAcodes IT; and OD4RD Achievements booklet. 3.Training material: *ad hoc* presentations broadening the scope with additional Orphanet related information. *The number of courses celebrated is lower than expected as one of the Information Scientists has not been available for over a year.

Networking:	
5. Contact the Ministry of Health (MoH) regarding the use of the Orphanet nomenclature pack at a national nomenclature server.	5. In April, the project was presented to the team from the MoH in charge of implementing OC at the nomenclature server. Follow up messages exchanged in Aug23 and Jan24.
6. Disseminate the project among POs.	6. The project has been presented at several events organized by: > The Spanish Federation of Rare Diseases, FEDER (Oct23; Nov23); > Principado de Asturias Regional Health Authorities (Feb24); > Infanta Sofia Hospital in Madrid (ORPHAcode photograph exhibition; Feb24); > Center for Biomedical Network Research (CIBER), Área of Rare Diseases (Mar24).
7. Disseminate the project among ERN's HCPs: re-contact all HCPs linked to ERNs to raise awareness about the project and the support provided by the National Hub.	7. Mass mailing (attaching National Hubs flyer): November 2023 > 103 HCPs from 24 ERNs (mainly clinicians, HCP medical informatics teams and coders) were contacted.
8. ERN survey: Launching, results' analysis and reporting	8. Launched in Jan. 2023. A report summarizing the results was produced (March 24)
9. Re-contact Spanish main regional RD registries to try to get more	9. Done. The action was not successful. No replies were received.
Helpdesk Implementation: The helpdesk was implemented in 2022 and it is integrated by the Orphanet-Spain team and a working group (WG) constituted by previous RD-CODE partners and collaborators.	
11. Provide support to nomenclature users by responding to their queries.	11. A total of 23 requests were attended.
12. Collaborate in the update of the Master File to include the ICD-10-ES mapping to the latest nomenclature pack available.	12. Continuous update of the MF to include ICD-10-ES alignments for the differential file from July 22 and July 23.
13. Incorporate new members to the WG as needed.	13. New contacts were made with 4 Regional Registries (February 24). However, we failed to recruit new members.

Further Activities: 14. Nomenclature translation to national language: processing of abstracts and disease names (translation, validation process and upload) 15. Promoting the Orphanet nomenclature: > Dissemination and political incidence, through contact with main patient federations in Spain. > Dissemination and political incidence, through participation of the National Hub members in the Scientific Coordination Group of the Spanish National Rare Diseases Strategy.	14. 322 abstracts/definitions and 1.282 names translated and uploaded since April 2023 15. (please refer to the networking section)

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, PhD, Assoc. professor, 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. They coordinate ERNs in Sweden and lead the EUHA RD-ERN network.

National action plan of Sweden

PLANNED ACTION	TIMELINE
Trainings/Workshops: Presentation of ORPHAcodes and the need for specific RD coding held at Visby Hospital, Gotland. Presentation directed towards healthcare professionals, patient organisation representatives and patients with a RD.	Oktober 2024 - Done

Production of a wider encompassing presentation on Orphanet, ORPHAcodes, rare disease, ERN:S and more. Production of educational videos on ORPHAcodes and surrounding topics which will be used in webinars and published on the internal e-learning portal for Karolinska University Hospital.	November 2022- ongoing Planned for 2024-2025
Networking: **ORPHAcode implementation:** Contact established with the hospital IT-dept. 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. Meeting with IT department and prescicion medicine department regarding the next step, which is an ORPHAcode implementation pilot at Karolinska. Orphanet Sweden created a customized mapping directed towards the National Board of Social Affairs and Health, using the information shown on their information pages for rare diseases. This was made to highlight the existing lack of congruity with ICD-10 codes and ORPHAcodes, and how ORPHAcodes provide a better coverage of RD. This mapping was presented to professionals from the unit for classification, the department for highly specialized care and the department of central production control the at Karolinska University Hospital. Five meetings held with Hospital IT-dept to further the implementation process.	 2022 - Done 2022 - Done 2022 - Done 2023 -Done 2024 - Done April 2024 – Done 2023-2024 - Done

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.	Postponed
Further Activities: Orphanet Sweden helped organize and create social media content for Rare Disease day 2024 in Stockholm. Orphanet Sweden was present at two hospitals in Stockholm to hand out flyers on Orphanet, ORPHAcodes as well as to provide information and answer questions from healthcare professionals, patients and the public. Promotional material in the form of pamphlets, flyers and presentations have been produced continually during 2024 as well as social media posts on the public instagram account for Karolinska University Hospital.	February - Done 2024 - Done

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 National Action Plans presented.

More than 60 trainings and workshops with a total of more than 1,300 participants have already been conducted or prepared. A number of national hubs have translated training materials and videos into their respective national languages to make their 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 national health administrations and national institutions in charge of coding to communicate the benefits of using ORPHAcodes. Numerous activities were aimed to ERN network clinics, RD expert centers, human genetic societies and other national expert groups. Also contacts were sought with providers of health information systems, hospital managers and umbrella patient organisations. The usage of ORPHAcodes has been promoted at national and international conferences. Furthermore activities were carried out to support the development of national plans for RD in countries where these do not yet exist or are being further developed.

From the 19 active national hubs, almost all have already implemented a national helpdesk. 15 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. The remaining 4 helpdesks (all in smaller population size countries) have opted for organising their helpdesk activities either via phone or via email.

A lot of further activities have been implemented and are still being pursued like the development of promotion material like leaflets, flyers, fact sheets or videos in national language, using them for national promotional campaigns. Also the translation of the Orphanet nomenclature in more local languages has been extended.

Annex: National hub and national action plan of Switzerland

Switzerland is no official member of the OD4RD2 project but has been accepted as guest country. The Swiss team has participated in all OD4RD2 work package 4 activities and has built 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**
<u>Trainings/Workshops:</u>	
Short presentations to several services at the Geneva University Hospital (Neurology, Orthopaedics, ENT), where our team is based, to encourage and support the clinicians to code their patients with a rare disease.	Throughout the year
Training on the nomenclature to the responsible of the Centre for Rare Diseases at the Lucerne Cantonal Hospital.	March 2024

Presentation to the students of the Biomedical Sciences Master from the Faculty of Medicine of the University of Geneva.	September 2023
Networking: The network at the political and institutional level is already established, mainly with: National coordination of rare diseases (kosek): The Orphanet national coordinator is member of several working groups. Kosek is in the process of designating reference centres at the national level which has clearly fostered the coding of rare disease patients. Swiss Rare Disease Registry: The Orphanet national coordinator is member of the Steering Board. We are working to strengthen the collaboration with the above partners and the Federal Office of Public Health. The Orphanet Swiss team is directly involved in the practical implementation of the Orphanet nomenclature at the Geneva University Hospitals (HUG), dealing with the professionals and performing a quality control on their coding: Patients coded in the period April 2023-March 2024: 710 (for the period April 2022-March 2023: 545)	Throughout the year
Helpdesk Implementation: The monthly meeting of the "Coding Working Group", launched in July 2021 in the framework of the Swiss Rare Disease Registry (SRDR), constitutes a helpdesk where national coders bring doubts and questions that are directly answered by the Orphanet Swiss team or transmitted to the Orphanet coordinating team. • 48 issues addressed to helpdesk (13 requests to create new entities in the nomenclature).	Ongoing

• Issues by Coders (45), Clinicians (3). • 32 issues submitted to the GitHub; 16 answered by the national helpdesk.	
Further Activities: Communication activities where we had the opportunity to discuss ORPHAcoding: Rare Disease Day: https://www.proraris.ch/fr/journee-maladies-rares-samedi-mars-2024-532.html Leaflet focused on the Swiss Rare Disease Centres and Helplines:	02.03.2024 March 2024