Deliverable 4.4

OD4RD2 National Action Plans Year 2+3

30.06.2025

This document represents deliverable 4.4 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 2024 to May 2025. 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 month period.

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

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Content summary

Context

Currently an estimated 30 million people in the European Union are suffering from one of the more than 6000 different rare diseases (RD). 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 RD are particularly important.

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

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 RD only in very few cases if using generic terminologies. Therefore, additional appropriate identifiers for coding RD become necessary. The Recommendation of the Council of the European Union of 8.6.2009 for action in the field of 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 of RD 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 RD, and to improve it building on the well organized and structured RD 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 with 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 health care providers (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 among work package 4 participating countries. The range extended 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 centres.
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 Orphanet nomenclature of RD and using the nomenclature and classification for statistical aggregation analysis for coded data.

The participating countries (except for Romania due to a late project entry) developed national action plans for the time period April 2023 until March 2024 (OD4RD2 National Action Plans Year 1, available at: https://od4rd.eu/03-deliverables). This document contains the continuation of their national action plans until May 2025.

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 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 on an internal OD4RD2 WP4 website. During the bimonthly WP4 conference calls these topics were discussed, and experiences of the different countries were shared to benefit from each other´s experiences and to address common topics in a coordinated manner. Finally, highlights of the developed national action plans were presented and discussed

This document represents the national action plans as of May 31st 2025. For the achievements 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 and Sweden

*France participated in WP4 only as coordinating partner and did not develop a national action plan; Latvia officially joined the OD4RD2 project on January 1st 2024; Romania officially joined the OD4RD2 project on September 1st 2024;

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 centres 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: Trainings and individual advice on the implementation of ORPHAcoding have been provided to Centres of Expertise (CoEs) and other stakeholders (e.g. hospital management, hospital IT etc.).	Individual advice has been provided since before the beginning of OD4RD2 (~2021). Official trainings for larger groups started in Q4 2023. A higher number of trainings will be carried out in the second half of 2025, since ORPHAcoding will become mandatory in officially designated CoEs on 01/01/26.
Networking: We are working on the implementation of ORPHAcodes on behalf of and together with the Ministry of Health (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 centres and the persons responsible for the respective hospital information systems.	Ongoing since 2021.
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.	Implemented in the frame of OD4RD.

Further Activities: Orphanet Austria supports the implementation of ORPHAcodes in Austria on behalf of the MoH (see above; advice beyond the Orphanet 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 has been developed according to the needs of the MoH and was provided the first three times. Subsequently, a detailed instruction for preparing the list was provided to the MoH. ORPHAcoding will be mandatory for CoEs beginning 2026 (this was achieved by the MoH in close collaboration with Orphanet Austria).	Started in the frame of OD4RD and OD4RD2, activities are ongoing.

National hub of Belgium

The Belgian OD4RD hub is part of the Rare Diseases Team hosted by Sciensano, the Scientific Institute of Public Health. The OD4RD2 hub is composed of two staff members:

Annabelle Calomme (Master in Molecular Biology, Country Coordinator of Orphanet Belgium, OD4RD1/OD4RD2 Belgian Hub project manager, member of the Sciensano Rare Diseases Team).

Laura Debouverie (Master in Pharmacy, member of the Sciensano Rare Diseases Team).

The Sciensano Rare Diseases team has expertise in developing and implementing RD projects that are part of the Belgian Plan for Rare Diseases (first national plan, Dec 2013). It has more than 20 years of experience with performing tasks related to Orphanet (data collection, validation and update of the Belgian specialized resources in the RD field, see The Belgian contribution to the international Orphanet database for rare diseases and orphan drugs | sciensano.be; translation of the Orphanet encyclopedia, terminology and structural Orphanet webpages into Dutch, see: OrphaDutch project).

The service hosting the Belgian OD4RD hub also manages national patient registries as the Central Registry for Rare Diseases and registries for specific RD, e.g. cystic fibrosis, neuromuscular diseases and rare bleeding disorders. The team participates in several European projects related to RD (EJP-RD, JARDIN JA, ERDERA, etc.). Sciensano was designated as the national contact point for the Belgian National Mirror Group on Rare Diseases (ERDERA, Work Package 23).

National action plan of Belgium

PLANNED ACTION	TIMELINE
Trainings/Workshops: **Delivery of trainings on implementation and use of ORPHAcodes to university hospitals and centres involved in rare diseases:**	
• Hôpital Universitaire de Bruxelles (H.U.B), Erasme, Endocrinology service: on site, in French, to clinicians	11/06/2024 (6 participants). DONE.
• Hôpital Universitaire de Bruxelles (H.U.B), Erasme, HUDERF, Bordet: on site, in French and English, to clinicians	18/09/2024 (4 participants). DONE.
• Sciensano: on site, in English, to registry managers	10/12/2024 (3 participants). DONE.
• University Hospital Brussels (UZ Brussel), session 1: online, in Dutch and English, to clinicians	21/03/2025 (5 participants). DONE.
• University Hospital Antwerp (UZA): on site, in Dutch and English, to clinicians	27/03/2025 (5 participants). DONE.
• University Hospital Brussels (UZ Brussel), session 2: online, in Dutch and English, to clinicians	01/04/2025 (13 participants). DONE.
Contribution to the scientific symposium entitled "Registry-Day, Advancing patient care and research through real-world data" organized by Sciensano: oral presentation of the Orphanet nomenclature of RD	17/05/2024 (ca. 80 participants). DONE.
Contribution to the conference entitled "The State of Health in Europe" organized by the Civil Society Organisations' Group of the European Economic and Social Committee (EESC): oral presentation of the challenges faced by PLWR and health professionals, emphasizing the need for specific RD coding in health information systems	04/06/2024 (ca. 135 participants). DONE.
Participation in a Policy Forum entitled "Equal opportunities in healthcare, including for rare diseases. Patients, experts and policymakers in	07/11/2024 (ca. 100 participants). DONE.

dialogue", at the Chamber of Representatives (report): several interventions to highlight the need for a legal framework to make the use of ORPHAcodes compulsory in Belgium (at least at the level of the centres of reference)	
Contribution to a workshop on rare diseases at the Cliniques Universitaires Saint-Luc (CUSL): oral presentation entitled “Fighting against the invisibility of rare diseases thanks to the Orphanet nomenclature of RD” OD4RD leaflet, the national flyer and promotion of ORPHAcodes	27/02/2025 (ca. 80 participants). DONE.
Networking:	
Meetings with representatives of Belgian political movements to present the Memorandum on rare diseases containing policy proposals aimed at improving the life of people affected by rare diseases in Belgium	21/05/2024. Done.
Participation in a "Rare Disease Brainstorming" at the Belgian Ministry of Public Health	25/06/2024. Done.
Meeting with the Terminology Center of the Federal Public Service (FPS) Health, Food Chain Safety and Environment regarding the implementation of ORPHAcodes in Belgian electronic health records	24/07/2024. Done.
Training of RD Patient Experts, in collaboration with the Patient Expert Centre: organization of certified training for Belgian patients to become experts in rare diseases (delivery of two modules: 1 on rare diseases in general; 1 on Orphanet, codification of rare diseases and patient registries)	17/09/2024, 15/10/2024. Done.
Participation in the 2024 RaDiOrg (Belgian umbrella association for people with rare diseases) Members Day	09/11/2024. Done.
Meeting with the RD coders at the Cliniques Universitaires Saint-Luc (CUSL): discussion on the	27/02/2025. Done.

use of ORPHAcodes at hospital level using the Epic HER software	
Holding a stand at the 2025 annual Belgian Society for Human Genetics (BeSHG) conference: distribution of the OD4RD leaflet, the national flyer and promotion of ORPHAcodes	14/03/2025 (ca. 200 participants). Done.
Participation in the workshop for ERN Hospital Managers and National Policy Contact Points Representatives in the context of the Joint Action on Integration of ERNs into the National Healthcare Systems (JARDIN): distribution of the OD4RD2 Belgian hub flyer, contact with the Belgian Hospital Managers Representative (Gert Van Assche, UZ Leuven)	19/03/2025. Done.
Participation in the federal working group on rare diseases: collaboration established between the ministerial cabinet, the NIDHI, the public health FPS, RaDiOrg, the representatives of the Rare Disease Functions, the Sciensano RD team and the Fund for Rare Diseases and Orphan Medicines in order to design the new Belgian rare diseases plan (expected by the end of 2025)	24/03/2025. Done.
Meeting with the software engineer in charge of the management of RD data at UZ Brussel: discussion on the use of ORPHAcodes to identify RD cases from the hospital EHR using the REDCap data management platform	25/03/2025. Done.
Meeting with the Belgian Rare Disease/College Genetics Working Group	04/04/2025. Done.
Kick-off meeting of the Belgian National Mirror Group Rare Diseases - ERDERA (WP 23)	20/05/2025. Done.
Helpdesk Implementation: The Belgian OD4RD2 hub acts 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.	

• Creation of GitHub posts (n=33: #210, 211, 231, 238, 242, 254, 257, 268, 283, 284, 286, 288, 291, 297, 298, 303, 310, 312, 327, 328, 333, 340, 341, 345, 347, 348, 349, 350, 353, 355, 356, 367, 370)	Continuously
• Provide answers to questions received by emails (without the need to create a GitHub post)	Continuously
• Promotion of the GitHub to RD experts	Continuously, during the OD4RD trainings and via emails
• Participation in Nomenclature/Coding Open sessions for National Hubs	7 sessions (18/04/24, 30/05/24, 27/06/24, 19/09/24, 12/12/24, 20/03/25, 12/05/25)
Further Activities: Accreditation of the OD4RD2 training sessions: the training given to Belgian doctors as part of the OD4RD2 project is accredited, this means that it entitles participants to a financial advantage in the context of continuing training	Since January 2025
Creation of a leaflet with description of the Belgian OD4RD2 hub (FR, NL)	March 2025. Done.
Update of the advanced quiz on the Orphanet nomenclature of RD (described in OD4RD GitHub #355)	April 2025. Done.
Drafting of a White Paper on the need for a legal framework for the implementation of ORPHAcodes, adapted to the Belgian context	June 2025 (planned)
Offering new training courses to hospitals that have already received training under OD4RD1 (in 2022 and early 2023)	Q3-Q4 2025 (planned)

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 centres 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 centre "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 RD, including HTA trainings for health authorities, academia, patient representatives and industry, participation in 10 national RD registries and numerous European RD projects.

National action plan of Bulgaria

PLANNED ACTION	TIMELINE
Trainings/Workshops:	**April 1st 2024-May 31st 2025: In total 34 meetings with 889 participants executed**

1. Training meetings with Health Care Professionals (HCPs) from current 34 RD Expert centres, aiming to highlight the relevance of ORPHAcodes, assess specific needs; Basic/Advanced trainings provided by Orphanet, adapted to local needs	Apr 24th – May 2025: Meetings executed with 22 RD Expert centres with 112 HCPs: main focus on awareness and usage of ORPHAcodes with relevant to the respective expert centre practical cases
2. OD4RD2 awareness / training sessions during:	
2.1 10th International HTA symposium organized by BAPES targeting HCPs (RD Expert centres), Clinical experts, Health authorities	April 5th 2024, Sofia, Bulgaria: Awareness session on ORPHAcoding benefits with 50 registered participants
2.2 44th National Medical Students Association meeting: targeting medical students from the 7 Medical Universities in Bulgaria	April 20th - 21st 2025, Stara Zagora, Bulgaria: Awareness session on ORPHAcoding benefits 120 registered medical students
2.3 25 National pediatric conference targeting pediatricians from all over the country	May 23rd – 26th 2024, Nessebar, Bulgaria Awareness session on ORPHAcoding benefits 250 registered pediatricians
2.4 XV National conference for Rare Diseases and Orphan Drugs: targeting HCPs in Bulgaria managing RD	September 13th – 14th 2025, Plovidv, Bulgaria: 280 registered HCPs
2.5 Training workshop for PhD medical doctors in haematology	January 10th – 11th 2025, Sofia, Bulgaria: 12 PhD attended the training workshop: session on ORPHAcoding benefits
2.6 1st National RD Expert Centres meeting with Orphanet France video session / presentation	April 4th 2025, Sofia, Bulgaria: 50 experts (RD expert centres, health authorities, clinical experts, patient group representatives, software providers: dedicated session on ORPHAcoding benefits
2.7 Meetings with software providers: National Health Information System and Hospital software providers with the objective of educating, training and potentially piloting implementation of ORPHAcoding in selected hospitals / frame plan for National information system implementation	Six meetings on: January 16th; February, 6th, 12th and 18th; April 4th and May 13th 2025: 15 representatives of software providing companies attended meetings
<u>Networking:</u>	
Meetings with responsible stakeholders: Ministry of Health, National Health Insurance Fund, Hospital Managers, National Health Information System, Hospital software providers for	April 1st 2024 – May 31st 2025 Ongoing

advocating ORPHAcodes, as well as with National Alliance for Rare Diseases patient representatives to highlight the relevance of ORPHAcodes and to promote OD4RD2 WP 4 objectives	
Helpdesk Implementation: Map other countries examples / solutions and deciding on solution Setting up the technical necessities for the solution Training of the team by Orphanet to be in charge of answering the queries Launching the helpdesk Conducting regular meetings between one of the main healthcare IT providers for hospitals and medical professionals working with RD expert centres to discuss and align on technical needs and integration of RD coding systems. Number of tickets/demands received: 4 Category of demands: Coding: 4 Demands from what type of users: IT personnel: 4 Number of demands answered by national helpdesk: 4 Number of demands required support of coordination team: 4 Of which, forwarded by GitHub to coordination team: 0 The most frequent question revolved around understanding the specific technical requirements of expert centres was regarding RD coding. To address this, we actively facilitated IT professionals in designing a questionnaire aimed at capturing the detailed needs and challenges faced by these centres. Furthermore, we organized and facilitated a series of collaborative meetings between IT providers and medical experts from the RD expert centres. These sessions were essential to foster clear	January 2025 – ongoing

communication, ensure mutual understanding, and align technical solutions with clinical realities, ultimately improving the integration and use of RD coding systems within healthcare workflows.	
Further Activities: 1.1) 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. 1.2) Distribute created materials: information brochure, fact sheet, etc., through national hub website, mailing to targeted stakeholders Educational materials covering the Orphanet nomenclature of RD, the utilization of the Orphanet database, and coding practices have been published on the national website in local language. The resources are available in Bulgarian at *Национален център – Orphanet България*, and in English at *National Hub - Orphanet Bulgaria* "Standard Procedure and Guide for the Coding with ORPHAcodes – RD-CODE (2nd update)" has been uploaded and is accessible on the national hub website. The Norwegian web-based approach for the Guide for Coding is planned to be adapted and implemented on the Bulgarian Orphanet National Hub website by September 2025 - *ORPHA-kodeverket - ORPHA-koder for sjeldne diagnoser*. Orphanet training videos with Bulgarian subtitles are planned to be uploaded to the Bulgarian Orphanet National Hub website by September 2025 - *Orphanet Tutorials*.	Executed and 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 Prague 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: We are offering trainings to ERN members and other HCPs at different meetings and conferences. We also offer prepared materials from previous trainings and workshops online.	Ongoing
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).	Ongoing

Cooperation with Czech Association for Rare Diseases and other patients support groups on promotion of ORPHAcodes. We are offering support to vendors/developers of health information systems.	
Helpdesk Implementation: We already provide support over e-mail and phone and with direct contact and demonstration with clinicians. Contact details are on local page at orphanet.cz. More sophisticated system with issue/request tracking in local language is still under consideration.	Ongoing Later in 2025
Further Activities: Updating the Czech translation of the Orphanet nomenclature of RD 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) Promoting of ORPHAcodes during pregradual and postgradual education of health-care professionals Promoting ORPHAcodes in local medical journal Promoting ORPHAcodes in local genetic textbook Updating and promoting the translation of Human Phenotype Ontology (HPO) in Czech language Cooperation on creating pilot Czech Rare Disease registry Official methodology for ORPHAcoding in Bulletin of Czech Ministry of Health Regulation 373/2016 Coll. on rare disease registry implementing ORPHAcodes and outcomes of genetic testing - by Czech Ministry	Ongoing From 10/2025

of Health, in final negotiations in the Czech parliament	

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). The University of Tartu is a partner from Estonia in OD4RD2.

National action plan of Estonia

PLANNED ACTION	**TIMELINE**
<u>Trainings/Workshops:</u> Trainings/workshop for doctors how to use ORPHAcodes for coding at TUH TUH: • Is the main teaching hospital in Estonia • Is the only competence centre of rare diseases in Estonia • provides genetic testing and clinical genetics services all round Estonia, • has an out-patient clinic also in Tallinn A national meeting to introduce ORPHAcodes to other HCP in Estonia who are part of ERNs. A presentation (given by Prof. Vallo Tillmann)	On 31st May 2025 7 clinics at Tartu University Hospital (TUH) are using ORPHAcodes (ORPHAcodes were not in use in any hospital in Estonia in June 2023)

about ORPHAcodes, OD4RD2 project, our experience at TUH and about helpdesk Trainings for doctors how to use ORPHAcodes in other 9 clinics at TUH who has not started to use ORPHAcodes yet	On 3rd October 2024 a conference about ERNS-s in Estonia was held with more than 100 participants from different HCP-s represented at ERN-s in Estonia Sept 2025- Febr 2026 (planned)
Networking: With IT-team of Tartu University Hospital (TUH) to Implement ORPHAcodes at TUH	ORPHAcodes have been implemented into Electronic Health Record (eHL) at TUH On May 8th 2025 3441 cases have had their ORPHAcode at TUH. Other HCP-s in Estonia have no plan to introduce ORPHAcodes in 2025 or 2026 →thus, some activities planned for other HCP-s in OD4RD2 action plan (training, workshops) will be unlikely to be met as there is no need for that.
Helpdesk Implementation: Information about the possibility to ask help to find right ORPHAcodes have been distributed to the physicians at TUH. At the board meetings (held quarterly, last one on 8.5.2025) of Rare Disease Competence Centre (RDCC) at TUH the Helpdesk possibility have been introduced Actually (as on 31st of May 2025) 7 clinics at TUH are using ORPHAcodes. However, the need for the advice from helpdesk has been small so far, most likely due to the fact that the coding in Estonia is done by physicians.	Quarterly, latest on 8.5.2025
Further Activities: Providing the updated ORPHAcode nomenclature file to the IT department of TUH for the update within the Electronic Case Report (eHL) system	Last on in September 2024 The next is planned in September - October 2025

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 strategy for RD.

National action plan of Finland

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Training webinar on RD and ORPHAcodes to professionals	April 2024, done
Training on RD and ORPHAcodes to government officials	August 2024, done
Finnish European Health Data webinar, ORPHAcodes included	October 2024, done
ORPHAcode training for government officials	November 2024, done
Planning Finnish ERN Ithaca seminar	November 2024, done
ERDERA JARDIN Collaboration Workshop	January 2025, done
PROPHET workshop on pharmacogenomics	January 2025, done
National Finnish RD Day event	January 2025, done
Training event/workshop on RD and OPRHAcodes for patient organisations	April 2025, done

Workshop on RD, ORPHAcodes and vasculitis, politicians, government officials, patients and professionals in attendance	April 2025, done
Training session on rare cancers	May 2025, done
Networking:	
Discussion about OPRHAcodes between the Finnish members of the Nordic Network of Rare Diseases	April 2024, done
Meeting of the Nordic Network of Rare Diseases, ORPHAcodes discussed	April 2024, done
Meeting of the Finnish National Advisory Group on RD Coordination	May 2024, done
Meeting of the RD Units of Finnish University Hospitals	May 2024, done
Meeting of the Finnish National Advisory Group on RD Coordination	June 2024, done
Meeting with the Office of the Parliamentary Ombudsman of Finland, RD and ORPHAcodes discussed	August 2024, done
Meeting of the Finnish National Advisory Group on RD Coordination	August 2024, done
Meeting with the Nordic Welfare Centre, RD and ORPHAcodes discussed	September 2024, done
Meeting of the Finnish Delegates to the Nordic Network of Rare Diseases	September 2024, done
Meeting of the Nordic Network of Rare Diseases in Reykjavik	September 2024, done
Meeting with Ministry officials regarding RD and ORPHAcodes	September 2024, done
Meeting with Ministry officials regarding RD and ORPHAcodes	October 2024, done

Meeting of the Finnish National Advisory Group on RD Coordination	October 2024, done
Meeting of the Finnish National Advisory Group on RD Coordination	November 2024, done
Meeting between THL, the ministry and hospital head physicians	November 2024, done
Meeting of the Finnish National Advisory Group on RD Coordination	December 2024, done
Meeting with basic healthcare professionals regarding ORPHAcodes	December 2024, done
Meeting with Inclusion Finlands Norio Centre about RD and ORPHAcodes	December 2024, done
Meeting of the Finnish National Advisory Group on RD Coordination	December 2024, done
Meeting with basic healthcare professionals regarding ORPHAcodes	January 2025, done
Helpdesk Implementation: ***Note:*** *Finish national hub did not provide a comment on helpdesk implementation. Quote from their NAP of Year 1: "Building up a national helpdesk for ORPHAcodes, 2023, done."*	
Further Activities:	

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 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**
<u>Trainings/Workshops:</u>	
Presentation “Coding of rare diseases, Alpha-ID-SE update, OD4RD2 project”/ Meeting of the steering committee of NAMSE (National Action League for people with RD)	03.12.24 (done)
Alpha-ID-SE training video updated version (40 min.)	done (online since November 26th, 2024)
Orphanet nomenclature of RD training for German ERN members	planned for Nov/Dec 2025
<u>Networking:</u>	
NAMSE (National Action League for people with RD)	Participation in several Workshops/Meetings of NAMSE
Interaction with other governmental Departments	Throughout the whole year
Interaction with ACHSE (Alliance of Chronic Rare Diseases)	Throughout the whole year

Helpdesk Implementation: 63 questions addressed to helpdesk 04/24-05/25 (Coding: 49, Nomenclature: 14, Others: 0 Questions by: Coders (5), Clinicians (22), IT personnel (6), Hospital managers (1), Controllers (24), Others (5)	answered by national helpdesk, 5 questions forwarded to coordinating team via GitHub
Further Activities: Further development of German Alpha-ID-SE data file (version 2025); ORPHAcode connection with diagnostic terms of Alphabetical Index: 18.916, inclusion of 7.233 different ORPHAcodes in data file;	published on October 1st, 2024

National hub of Ireland

The national hub of Ireland consists of:

Dr Atif Awan (Consultant Nephrologist, Clinical Lead for the National Rare Diseases Office and Country Coordinator for Orphanet Ireland)

Geraldine Sweeney (Business Manager, National Rare Diseases Office)

Aileen Timmons (PhD, Information Scientist for Orphanet Ireland and National Rare Diseases Office).

The Irish team is located at the National Rare Diseases Office in the Mater Misericordiae University Hospital, Dublin. Besides their Orphanet responsibilities, it is responsible for the National Rare Diseases Information Service, leading a pilot project developing care pathways for RD, supporting ERN integration into the health system, provision of RD education and awareness activities and initiatives and participating in EU/EC-funded joint action grants such as JARDIN and OD4RD and contributing to national research in RD. The National Rare Diseases Office will play a key role in supporting the implementation of the new National Rare Disease Strategy 2025-2030.

National action plan of Ireland

PLANNED ACTION	TIMELINE
<u>Trainings/Workshops:</u> University College Dublin (UCD) – Medical Students (35 participants). Included information on Orphanet and ORPHAcodes **Information/training sessions on ORPHAcodes and Orphanet Nomenclature of RD and RD Classification system** Development of information and training materials **Workshops planned** for: RD Research Coordinators RDCat Project Members ERN Teams	 Feb 2025 (complete) Jan 2025 (ongoing) June 2025 (ongoing)
<u>Networking:</u> **Department of Health** Since December 2023, the NRDO has been a key contributor to the development of the **second Rare Diseases Strategy for Ireland 2025 – 2030**. The NRDO presented several times to the RD Strategy Steering Committee on key topics including ORPHAcodes and the importance of ORPHAcodes in Health Information Systems and Registries. **Children's Health Ireland (CHI)** Information Session on ORPHAcodes and importance of ORPHAcodes in EHRs Meetings with CHI to discuss progress of implementation of ORPHAcodes in the EHR currently under development at CHI **National Genetics and Genomics Office (NGGO)**	 Dec 2023 (ongoing) Second Rare Disease Strategy 2025-2030 (pending publication) April 2024 (ongoing) Dec 2023 (ongoing)

The NRDO and NGGO are collaborating on shared objectives	
Helpdesk Implementation: 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 ORPHAcode use **GitHub (Ireland**) monitoring	Set up in Feb 2024 (complete) April 2024 (ongoing) April 2024 (ongoing)
Further Activities: **ORPHAcoding Success:** **Children's Health Ireland (CHI)** - Electronic Health Records (EHRs) are being introduced in the new national Children's Health Ireland hospital (CHI). • The NRDO/Orphanet Ireland team have met with CHI to advise on the importance of including ORPHAcodes in the planned new CHI EHRs (EPIC) • 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 **Interim Solution** • ORPHAcodes are being implemented in EHRs in CHI- based on existing IT systems and SNOMED-CT mapping with ORPHAcodes using the nomenclature pack to allow extraction of RD data **National Rare Disease Strategy 2025-2030**	 Feb 2024 (ongoing) Jan 2024 (ongoing) May 2024 (ongoing)

• Ireland's second National Plan will be published in Q3 2025. • The new Strategy recommends the implementation of ORPHAcodes as the optimal clinical coding system for RD • It recommends the long-term vision for the development of EHRs and associated IT systems should include the full integration of ORPHAcodes. A stepwise and an incremental approach to the integration of ORPHAcodes may be required to achieve this.	Pending publication
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 See: 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).	Feb 2024 (ongoing)
Rare Diseases Research and Innovation Catalyst Award (HRB RDCat) 2023 Aims to increase support and capacity for Rare Diseases pre-clinical, clinical, population health and health services research, 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 • Five Research Coordinators have been appointed and have a role in supporting ERN Registries.	April 2024 (ongoing) April 2025
OD4RD2 Stakeholder Information / communication pack Development of OD4RD information for dissemination across stakeholders to introduce	April 2024 (ongoing)

the project and to support upcoming information / training sessions **OD4RD2 Technical Workshop** in attendance with CHI colleagues	 Dec 2024

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 of RD and tools.

National action plan of Italy

PLANNED ACTION	**TIMELINE**
<u>Trainings/Workshops:</u> • XXVII Congress of the Italian Society of Human Genetics (S.I.G.U.) Orphanet boot. Dissemination of the activity of the Orphanet Italy national hub. Updated leaflets of the Orphanet resources (i.e. Orphadata website) were prepared and available for the attendees. • OD4RD2-branded training for the implementation and use of the ORPHAcoding. A shortened version of this training course,	 Done Done

focusing on the hands-on session, was delivered online on 19th December 2024.	
Networking: Italian Society of Human Genetics (S.I.G.U.) Ministry of Health. 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. Press agency O.M.a.R.- Rare Diseases Observatory: dissemination of OD4RD project aims National Centre for Rare diseases (Istituto Superiore di Sanità).	 Ongoing Ongoing Ongoing Ongoing
Helpdesk Implementation: Dissemination of the Italian GitHub helpdesk: • via social media and OrphaNews Italy. • on the occasion of the XXVII Congress of the Italian Society of Human Genetics (S.I.G.U.)	 Ongoing Done
Further Activities: • On the occasion of the S.I.G.U. National Congress, USB keys with informative material on ORPHAcodes use (basic level) were prepared and freely given to the participants working in the rare disease field. • On February 25th 2025, in collaboration with OPBG, OMAR and two patient organizations (Hopen Foundation, IMI Committee), we promoted a webinar for the creation of a national network of outpatient clinics for listening and	 Done Done

taking care of undiagnosed and ultra-rare disease patients, and to endorse the use if ORPHAcode 616874	

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 centre 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**
<u>Trainings/Workshops:</u>	
Meeting with the Ministry of Health and National Health Service: Presentation on the importance of ORPHAcoding and current situation in Latvia and its inclusion possibilities in E-health	Q2 2024, done
On-site training in university and regional hospitals of Latvia: Orphanet and coding of rare diseases (n=4) On-site training in university and regional hospitals of Latvia: Orphanet and coding of rare diseases (n=4)	Q4 2024, done
Online session for general practitioners (n=3)	Q4 2024, done
Onsite lecture about ORPHAcodes for GPs	14.03.2025, done

Onsite and online presentation about the usage of ORPHA codes in clinical practice for FMR doctors	24.04.2025, done
Online training about the usage of ORPHAcodes in clinical practice	Q4 2025, planned
Networking:	
Additional demand for ORPHAcoding in case of usage specific to ICD10 codes	Q3 2025, planned
Handouts about ORPHAcoding	Q4 2024, done
Implementation of an ORPHAcoding information summary in the CCUH website for general practitioners in Latvia (aiming to simplify the workload for doctors that are not as well versed in rare disease protocols)	Q4 2025, planned
Helpdesk Implementation:	
Promoting helpdesk activities	Q1-Q4 2025
Dissemination of patient rare disease cards including information about the respective ORPHA code of a particular patient	Q1-Q4 2025
Actively following registered ORPHA codes and suggesting registering any additional diagnoses in the patient registry file if needed	Q1-Q4 2025
Regular reminders for doctors to consider coding their patients, based on used manipulation codes	Q1-Q4 2025
Quality control of ORPHAcode usage	Q1-Q4 2025
Further Activities:	
Creating an online information brochure about ORPHAcodes in Latvian for the general public	Q3 2025, planned
Participation in the creation of a new RD patient registry platform	Q4 2025, planned
Modify document "Registration cards for patients with congenital malformations" (must be approved in the regulations of the Cabinet of Ministers)	Q4 2025, planned
Radio interview about ORPHAcode " Zināmais nezināmajā" (Known in the unknown)	26.02.2025, done

Television interview including ORPHAcoding "Zilonis studijā" (Elephant in the studio) Handouts about ORPHAcoding published online	24.04.2025, online (recorded 6.03.3025.), done Q3 2025, planned

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 Centre 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 Centre 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 Dissemination of updated guidance materials on ORPHAcoding in Lithuanian language Integration of ORPHAcoding principles into academic curricula (undergraduate and postgraduate levels)	 Regular training sessions for healthcare professionals throughout the year Updated guidance available / in use Ongoing

Networking:	
Continued collaboration with Rare Disease Coordination Centre and ERNs Bilateral and group consultations with hospital administrators on Rare disease services group under the Ministry of Health	Throughout the year
Participation in inter-institutional working groups and thematic roundtables	
Policy dialogue on RD strategy implementation with the Rare Disease Policy	
Development of the new National Plan on Rare Diseases was commenced	
Rare disease day events	2 events (2025-02/03) 1) “Paths of Rare Disease Care: Opportunities and Challenges”; International Scientific–Practical Conference to Mark Rare Disease Day 2) “Sanfilippo Syndrome – What’s New?” Academy of Rare Diseases, International Seminar
Helpdesk Implementation:	
Continued operation of helpdesk via email and phone	Ongoing support provided since Q4 2023
Regular response to inquiries from healthcare professionals and technical staff	Inquiries continue to be addressed by the designated team
Monitoring of common issues to support future updates in ORPHA coding tools	
Further Activities:	
Orphanet classification translation into Lithuanian language	achieved 2023-04 but implementation into the national e-health system is still ongoing
Rare disease registry at VUH Santaros Klinikos	enhanced with new interactive functionalities (since 2024), including data monitoring tools
National-level integration of ORPHAcoding into e-healthcare system	ongoing, trial version tests passed 2025 Q1

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), until September 1st, 2024

Besides the work for Orphanet they participate in the national assessment of expert centres for rare diseases, and Prof. Van Zelst-Stams is part of the Undiagnosed Diseases Network International and at the board of Member States for ERNs. They also take part in the implementation of ORPHAcodes in Dutch Health Care Records.

National action plan of the Netherlands

PLANNED ACTION	**TIMELINE**
<u>Trainings/Workshops:</u> Development of 2 e-learnings – one technical & one clinical - specifically for the Netherlands: E-learnings will be based on existing ORPHAcode training material and acquired insight from, and experience with, implementation of ORPHAcodes and registration in EHR. Necessary steps: 1. Creation of first version 2. Testing phase 3. Improving and updating e-learning 4. Available for the Netherlands	 Q3-Q4/2025 Q1/2026 Q2/2026 Q3/2026
<u>Networking:</u> Regular meetings with the following organizations 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 organizations 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)	Meeting whenever necessary
Additional meetings/networking actions:	
1. To improve effective registration with ORPHAcodes in the EHR relevant rare disease entries should be linked to the correct codes for reimbursement and added to the different specialism-specific diagnosis lists. Therefore, the NFU, together with DHD, launched a campaign with the following steps:	
- Officially designated expert centres propose links between ORPHAcodes and codes for reimbursement.	Q2-Q3/2025; until the end of this phase regular meetings take place with the hospital rare disease teams.
- The results are presented to the relevant Dutch Societies of Medical Specialists for validation.	Q3-Q4/2025
- DHD will add ORPHAcodes to the different specialism-specific diagnosis lists based on the lists validated by the Dutch Societies of Medical Specialists.	Q4/2025-Q1/2026
2. At the National Conference on Rare Diseases the Orpha-viewer will be presented and the ORPHAcode for undiagnosed patients briefly discussed.	04/04/2025
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. - 5 questions were received regarding the nomenclature and 13 regarding a coding issue. The questions were submitted by clinicians (7), coders (10) and others (1).	Implemented
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. - 2 questions were redirected to us.	Routing from DHD to Orphanet NL helpdesk was provisionally implemented. After evaluation in Q4/2023 formal implementation was deemed not necessary.
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 is not actively promoted. - no questions were received via this route.	Account in place
Further Activities: Since April 2024, 232 new links to ORPHAcodes have come available in the DT. In total now 5732 ORPHAcodes are linked in the DT: 176 groups of disorders 5400 disorders 165 subtypes Of the 6417 disorders present in the Orphanet nomenclature of RD (July 2024) ~84% is linked in the DT.	Q2/2024-Q1/2025
Further development of the Orpha-viewer: • First testing-phase by small user group ended	Q4/2024-Q1/2025
• Based on feedback the user-friendliness will be improved	Q1-Q2/2025

• "De-doubling" of patients will be fully implemented	Q1-Q2/2025
• A second testing-phase with a larger user group will be performed	Q3-Q4/2025
Visibility ORPHAcodes in EHR (Epic):	
• The local Epic service team at the Amsterdam UMC created specific fields for ORPHAcodes in the EHR	Q4/2024
• A project with similar goal is ongoing at the Radboudumc	Q1-Q4/2025
In the Erasmus MC a pilot project will be initiated to provide medical specialists in the EHR the option to search for medical diagnosis ONLY in the subset of ORPHAcodes – linked DT terms.	Q3/2025-Q3/2026
Some DT terms used in clinical practice are more specific than available ORPHAcodes (e.g. terms for certain tumors in specific locations). To keep these terms available for patient coding while also being able to link these patients to ORPHAcodes, the option to align these specific DT terms to the more general ORPHAcodes will be explored.	Q2-Q4/2025

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., MHA, biomedical laboratory scientist, medical genetic counselor, national coordinator Orphanet Norway)

Lene Martinsen (PhD. Biology/Genetics, 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. Moreover, they manage the Norwegian Registry on Rare Disorders.

National action plan of Norway

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Information meetings, including basic training of clinicians, offered to departments, centres and networks based at Oslo University Hospital (OUS). Some of the units display national functions:	
National Network on Congenital limb malformation	21.05.2024, done
Department of neurohabilitation	23.04.2025, done
Centre for rare disorders	19.05.2025, done
National Centre for Rare Epilepsy-related Disorders	22.05.2025, done
Training of healthcare professionals associated with the expert centre on Neuromuscular Diseases, University Hospital of North Norway	12.11.2024, done
Information about ORPHAcodes and the Orphanet nomenclature of RD to the Northern Norway Regional Health Authority ICT Trust in regard to ORPHAcodes implementation in the EHR system of hospitals in Northern Norway Regional Health Authority	24.01.2025, done
ORPHAcodes e-learning course: • Development of English version, in collaboration with Orphanet coordinating team France (ORPHAcodes for rare diseases - Sjelden) • Revision of Norwegian version (ORPHA-koder for sjeldne diagnoser - Sjelden)	23.09.2024 (published online), done 31.03.2025 (last revision), done
<u>Networking:</u>	

Establish contact points for ORPHAcodes implementation in the EHR systems and future trainings of the four Health Regions: • South-Eastern Norway Regional Health Authority • Western Norway Regional Health Authority • Central Norway Regional Health Authority • Northern Norway Regional Health Authority	 Q2 2024, ongoing and throughout the year Q2 2024, ongoing and throughout the year 27.05.2025, planned Q4 2024-Q1 2025, done
Interaction with governmental departments and Health Authorities concerning measures in The Norwegian National Strategy for Rare Diagnoses: • the Norwegian Directorate of Health • The Norwegian Institute of Public Health • Regional Health Authorities	Q2 2024, ongoing and throughout the year
Dialogue with the Norwegian Institute of Public Health, Department of Patient registries Data Management and Quality, on possibility of including ORPHAcodes in the dataset of Norwegian Patient Registry (NPR)	Q2 2024, ongoing
Dialogue with Lovisenberg Diaconal Hospital, on possible implementation of ORPHAcodes in EHR system	Q2 2025, planned
Association of General Practitioners –Rare Diseases Interest Group	Q3 2024, done
National ERN networking day, Aarhus, Denmark: Presentation “Norway’s implementation of ORPHAcodes	20.01.2025, done
Nordic University Hospital Alliance: Participation in meeting “NUHA knowledge sharing ORPHAcodes”	19.05.2025, done
Participation in JARDIN WP8 Data Management	Q1 2024-Q2 2025, ongoing and throughout the year
Dialogue with Norwegian registry on POLG-related mitochondrial disease	29.04.2025

Helpdesk Implementation:	
Reply to incoming inquiries to the national helpdesk (via e-mail/web form/in person contact)	Q2 2024, ongoing and throughout the year
Participate in OD4RD2 Nomenclature and coding open sessions	Q2 2024-Q2 2025, done
Further Activities:	
Collaboration with the EHR software provider DIPS on development of new solution/product for ORPHAcodes in the hospital EHR system DIPS Arena	Q2 2023-Q4 2024, done
Collaboration with the regional ICT provider of the South-Eastern Norway Health Region, Sykehuspartner, regarding implementation in hospitals in this region	Regular meetings Q1-Q2 2025, ongoing
Public hearing from the Directory of Health, regarding a national information model (Helse-NIM) comprising ORPHAcodes in electronic patient summary: supportive response given on consultation paper	19.03.2025, done
The Norwegian Registry on Rare Disorders: • Participation in working group on investigation of how to further development the registry into a national registry (national coverage) • Follow-up work concerning further development into a national registry	Q3 2024-Q1 2025, done Q1 2025, ongoing and throughout the year
Participation in working group on establishment of a national registry on childhood visual impairment	31.10.2024, 10.01.2025, 04.04.2025, ongoing and throughout the year

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, translator and information scientist of Orphanet Poland)

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

Jagoda Błaszkiewicz (information scientist, web developer)

Alina Belska (information scientist)

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:	
ORPHA coding – dedicated training sessions and as a part of thematic courses for medical specialist in training (in total over 300 participants):	
1.ORPHAcodes, Wrocław, ABM Digital Medicine program team (14 participants)	23.04.2024
2.ORPHAcodes, IPCZD Digital Medicine team, representatives of the Science Department and IT Department (5 participants)	27.05.2024
3.Postgraduate course in paediatric neurology course “Genetic testing in neuropediatrics” (32 participants)	05.06.2024
4.ORPHAcodes – The Warsaw Medical University, Paediatric Neurology Clinic, Centre for Rare Diseases, hospital administration (3 participants)	10.06.2024
5.ORPHAcodes, Dept of Medical Services Contracting, CMHI (3 participants)	02.09.2024
6.ORPHAcodes, Dept of Gastroenterology CMHI, ERN-Liver (20 participants)	11.09.2024

7.ORPHAcodes, Dept of Neurology and Epileptology Clinic CMHI, ERN EpiCare (20 participants)	23.09.2024
8.ORPHAcodes, Dept of Immunology CMHI, ERN RITA (7 participants)	01.10.2024
9.Postgraduate course “Genetics in Pediatric Neurology” Elements of ORPHA coding rules as a part of the lecture “Genetic Counseling” (10 participants)	21.10.2024
10.Postgraduate course “Introduction to Specialization in Genetics” Nomenclature and Classification of Rare Diseases According to Orphanet (3 participants))	04.11.2024
11.ORPHAcodes, Dept of Pediatrics and Metabolic Diseases CMHI, MetabERN (40 participants)	30.01.2025
12.ORPHAcodes, Dept of Science CMHI (15 participants)	19.02.2025
13.Postgraduate course “Pediatric neurology” Elements of ORPHA coding rules as a part of the lecture “Genetic Testing in arrhythmology” (10 participants)	24.02.2025
14.ORPHAcodes, Dept of Pediatric Surgery and Organ Transplantation CMHI, ERN TransplantCHILD (20 participants)	26.02.2025
15.ORPHAcodes, Dept of Nephrology, Kidney Transplantation and Arterial Hypertension CMHI (13 participants)	26.03.2025
16.ORPHAcodes, University Pediatric Hospital Cracow-Prokocim - representatives of 6 ERNs in Cracow (70 participants)	15.04.2025
17.ORPHAcodes, secretaries and coders from ERN units of CMHI (20 participants)	23.04.2025

18.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 1st 2024 – May 31st 2025
Networking: 1.Conference „Rare diseases – problems not only systemic" – lecture: „Cooperation of Orphanet with European Reference Networks – challenges, common standards and common language" 2.Health Policy Summit – Rare Diseases Expert debate of the Institute for the Development of Social Affairs 3.National Forum for Rare Disease Therapy ORPHAN – celebration of the World Rare Disease Day 2025 4.Conference "Rare diseases - More than you can imagine"; discussion panel "Common goal, two perspectives" (register of rare diseases - promotion of the use of ORPHA codes), Warsaw	18.05.2024 27.06.2024 28.02.2025 01.03.2025
Helpdesk Implementation: Orphanet Poland National Helpdesk in the Memorial Children's Health Institute, Warsaw – solving problems and providing support with ORPHA coding / answering questions on Orphanet nomenclature of RD, classification and translation – contact via e-mail and phone (in Polish) (a.madej-pilarczyk@ipczd.pl; k.chrzanowska@ipczd.pl) GitHub account for Polish helpdesk	Implemented, active Implemented, active
Further Activities: 1.Expansion of Orphanet resources in Polish language: Translation of Orphanet diseases summaries and full descriptions of RD to Polish (all	

nomenclature and over 2000 diseases summaries have been currently translated and updated; approx. 488 disease summaries (short abstracts) were translated / modified and uploaded to POLOR in the period of 1st April 2024 – 31st May 2025)	Done, ongoing (April 1st 2024 – May 31st 2025)
Preparation of handy lists of RD with ORPHAcodes divided into medical fields	Done
Orphanet Poland website www. https://orphanet.site/poland with translated content regarding Orphanet, the principles of naming and classification of RD together with the assignment of Orpha and links to important resources (including the National Plan for Rare Diseases, the Rare Disease Information Platform, the European Reference Networks).	Done, ongoing
2.Regular meetings of the Council of Rare Diseases, established by the Minister of Health	12.11.2024, 16.12.2024, 09.01.2025,23.01.2025. 27.02.2025, 08.05.2025
3.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 RD 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 of RD (Polish translation) and short description of RD (partially translated to Polish) with respective link to Orphanet website and Orphanet logo	Done
4.Diagnostic recommendations in RD	Publication planned in 2025
Sections dedicated to each rare disease/group of RD contain following information: Name of RD/group of RD, Incidence, Inheritance, Codes: OMIM, ICD10, ICD11, ORPHAcode, Recommended genetic diagnostics	08.04.2024, 22.04.2024, 27.05.2024, 12.06.2024
Regular meetings of the Editorial Committee:	25.06.2024, 09.07.2024, 16.07.2024

5.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 RD delivered by Orphanet (nomenclature, classification, ORPHA coding, etc.)	23.07.2024, 30.07.2024, 06.08.2024 27.08.2024, 15.10.2024, 29.10.2024
6.Implementation of ORPHAcodes into the IT system of the National Health Fund, integration with the IT system of the e-Health Centre, method of reporting ORPHAcodes (17 participants form the Ministry of Health and IT team E-Health)	07.05.2025
7.Questionnaire on ORPHAcoding usage in Polish ERNs centres – presentation during bi-monthly WP4 meeting together with NAP for 2-3 year	23.05.2024
8.ORPHAcode usage in Poland during bi-monthly WP4 meeting	28.11.2024
9.ORPHAcodes implementation in Poland - use-cases – phone call	16.01.2025

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 and ERN National members as ORPHAcodes users. 1. Update existing material from trainings within OD4RD project on ORPHAcoding and nomenclature 2. Organize 2-3 online training sessions 3. 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 presented in meetings and made available on the Orphanet national website. 4. Present Orphanet promotion materials in scientific meetings and conferences 5. Make available Orphanet promotion materials on Orphanet national website	 2024 (done) Sep.2024 (done), Sep.2025 (planned) 2024 (done), 2025 (planned) 2024 (done), 2025 (ongoing) 2024 (done), 2025 (ongoing)
<u>Networking:</u> OD4RD2 project formal presentation to the Ministry of Health • Present OD4RD2 project and the national Action Plan to the New Ministry of Health. Promotion visibility of ORPHAcodes • Expert Centres members of ERNs or Reference Centres will be encouraged to use ORPHAcodes	 2024 (done), 2025 (planned) 2024 (done), 2025 (ongoing)

• To contribute to a new Integrated Strategy for Rare Diseases and promote its implementation	2024-2025 (ongoing)
Umbrella Patient Organization • To highlight the relevance of ORPHAcodes during the annual meeting organized by the umbrella Patients Organization	Nov.2024 (done)
RD Annual Event • During the Rare Diseases Day celebrations, the use of ORPHAcodes will be highlighted by the National Hub presentation	Feb.2025 (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 of RD within of Rare Disease Card new registries	2024 (done)
2. Implement and promote GitHub tool for Orphanet national hubs	2024 (done)
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)	2025 (awaiting the requested IT implementation)

Further Activities:	
Annual report on the implementation of the Rare Disease Card	
• The Annual Report on the implementation of the Rare Disease Card to be published online on the Directorate-General of Health website	2024 (done), 2025 (planned)
• An English translation of the Report to be published on Orphanet national website	2024 (done), 2025 (planned)
Maintain an up-to-date translation of the Orphanet nomenclature of RD	
Provide an updated translated Portuguese version of ORPHAcodes to the ERN and national Reference Centres.	
• Translation activities of the nomenclature and their clinical validation have been maintained throughout the year	2024 (done), 2025 (ongoing)
• An extensive translation of Orphanet abstracts on RD to take place	2024 (done), 2025 (ongoing)

National hub of Romania

The national hub of Romania consists of:

Cristina Rusu (MD, PhD, Prof., Pediatrician and Clinical geneticist, national coordinator of Orphanet Romania)

Monica Panzaru (MD, PhD, assoc. Prof., clinical genetist, information scientist Orphanet Romania)

The Romanian team is part of the University of Medicine and Pharmacy “Grigore T. Popa” Iasi (UMF Iasi), and part of Iasi Regional Medical Genetics Centre belonging to “Sfanta Maria” Paediatric Emergency Hospital Iasi. Prof. Rusu is a member of the Romanian National Council of rare diseases, consultative body of the Health Ministry, as Orphanet representative. Besides providing specialized healthcare for patients with congenital anomalies and intellectual disability, they participate in local research projects, train medical students, residents, PhD students in the field of rare genetic diseases, promote the expansion of newborn screening in Romania and the introduction of a national registry for RD.

National action plan of Romania

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Presentation "The benefits of genetic testing and the use of ORPHAcodes in rare diseases" at the National Conference of Modern Neurosciences, "Rare & Autoimmune Diseases."	March 21-22, 2025
Discussions with the representatives of the current RD Expert reference centres, to highlight the relevance of ORPHAcodes and to establish a workshop schedule.	ongoing
We also offer materials from previous trainings and workshops online.	
Networking:	
We are working on implementing ORPHAcodes in other Centres of Rare Diseases. Regular meetings are held between Orphanet Romania and the centres and the personnel from hospital information systems.	ongoing
Promotion of ORPHAcodes to the Working Group on Rare Diseases from the Ministry of Health.	
Cooperating with the National Alliance for Rare Diseases to promote ORPHAcodes among patients.	ongoing
A memorandum regarding the need to introduce ORPHAcodes was submitted to the Ministry of Health.	October 2024
Helpdesk Implementation:	
We already provide support over e-mail and phone, and direct contact (email orphanet.romania@gmail.com)	ongoing
Map other countries' examples/solutions.	ongoing

Further Activities:	
Orphanet Romania supports the implementation of ORPHAcodes in Romania (guidance beyond the Orphanet classification and nomenclature). A coding list adapted from the Orphanet nomenclature pack is provided to other centres.	ongoing
Select and adapt from other countries' best practices, like an information brochure, take-home leaflet, satisfaction survey, etc.	ongoing
Promote the use of ORPHAcodes at different national conferences.	ongoing

National hub of Slovenia

The national hub of Slovenia is located at the Clinical Institute of Genomic Medicine, University Medical Centre Ljubljana and consists of the following members:

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

Borut Peterlin (MD, PhD; MD in Clinical Genetics and Neurology, Prof. of Human Genetics in Med. fac.)

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

Mateja Vinkšel (MD in Clinical Genetics)

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, participation in a national RD registry development and participation in European projects (e.g. eHealth).

The main activity of the team is clinical and genetic evaluation / diagnostics of RD patients.

At our institute, Borut Peterlin and Luca Lovrecic with some other team members also established a National Centre for Undiagnosed RD patients.

National action plan of Slovenia

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
Basic trainings for the personnel of all ERNs (online/in person).	Ongoing, several trainings – we have had trainings for national teams of ERN-RND, ERN-NMD, MetabERN, ERN RARE-LIVER, ERN-Skin, ERN-ITHACA, ERN eUROGEN, ERN-EYE
Advanced training on Orphanet coding dedicated for genetics team (medical & laboratory, including researchers)	At the Clinical Institute of Genomic Medicine, UMCL, the main diagnostic unit and outpatient clinic, we have had 2 trainings for coding and more are planned – the main challenge is that we regularly encounter diseases with lacking ORPHAcodes
Multidisciplinary team (MDT) for rare diseases	Implementation of ORPHAcode usage in the MDT patient management and evaluation
Basic trainings for the personnel of national RD registry (in person).	April 2025 and to be continued when they implement IT solution
Networking:	
Interaction with the Ministry of Health concerning The Slovene National Strategy for Rare Diseases	Ongoing, several meetings
National project on the education on RD (Orphanet team participates)	October 2023 – June 2026 - CRP 2023 – 3.2.1 Project title: „The innovative approach in education in the field of rare diseases“
Helpdesk Implementation:	
We introduced a dedicated help desk line/e-mail for questions	Slovenia is too centralized and „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.
Should our OD4RD2 project address / suggest how to keep up with the rapid progress of new gene discoveries and the lack of ORPHAcodes?	

The main challenges we encounter are non-existing ORPHAcodes – the field of RD and gene discoveries is so vibrant and changing quickly. We alone have participated in the discovery of several new genes in the past 3 years, which cannot be coded in the system – we would be keen to discuss options generating the ORPHAcodes in such scenarios	
Further Activities: Active promotion of ORPHAcodes at all points Further meetings with hospital management to implement ORPHAcodes at other departments. Active collaboration with national RD registry to implement ORPHAcodes	ongoing

National hub of Spain

Spain's National Hub is integrated by Orphanet Spain, hosted by the Centre 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 all the tasks related to Orphanet´s daily activity, the team leads the Scientific Coordination of the RD Strategy of the Spanish NHS, and it contributes to CIBERER by being involved in the management of national and international projects as well as being in charge of attending the consultations received 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), and he is the Editor in Chief of the Orphanet Journal of Rare Diseases.

National action plan of Spain

PLANNED ACTION	TIMELINE
Trainings/Workshops:	
1. Organize and deliver "Orphanet coding and nomenclature" training sessions in national language. Ad hoc training courses as requested by specific Institutions or by Regional Health Authorities.	4 training sessions delivered (17th Apr 2024, 23rd Apr 2024, 10th Jun 2024, 5th Feb 2025, 30th May 2025), to a total of 134 participants from La Paz Hospital (Madrid), Aragon Health Sciences Institute - IACS, Basque Country RD Registry, CIBERER's RD Biobank.
2. Translate new training material produced by OD4RD2 partners	Norway's e-learning course has been translated into Spanish. It is being formatted to be included in an e-learning platform.
3. Produce training material as needed	New ppts and exercises to be used in the trainings have been prepared.
Networking:	
1. Dissemination and political incidence, through contact with main patient federations/organizations in Spain.	The project has been presented at events organized by the Spanish Federation of Rare Diseases (FEDER): • III Foro de Enfermedades Raras – Bellvitge Hospital, 9th May 2024 • Foro Enfermedades Raras Andalucía- University Hospital Virgen del Rocío (Sevilla), 2nd Dec 2024.> Centre for Biomedical Network Research (CIBER), Área of Rare Diseases (Mar24).
2. Disseminate the project and promote the Orphanet nomenclature of RD among other stakeholders: Health/Research centres, National and Regional Health Authorities and through participation of the National Hub members as Scientific Coordinators of the Spanish National Rare Diseases Strategy.	• Meetings held with La Paz Hospital (Madrid) to approach the implementation of ORPHAcodes (May 14th 2024, June 28th 2024) • Contact with the Regional Government of Castile and Leon (phone and email contacts to provide support with implementation). • Contact with the Aragon Health Sciences Institute - IACS (phone and email contacts to provide support with implementation).

	• Contact with Catalonia Regional Health Authorities (phone and email contacts to discuss future training). • Meeting with the representatives of the National RD Registry (ReeR) – Ministry of Health on July 1st 2024 to explore possible collaboration opportunities. • Meeting with all Regional RD Registries and the MoH on Jan 28th 2025 to disseminate the OD4RD2 project and request collaboration.
Helpdesk Implementation: Helpdesk was implemented in 2022, and it is integrated by the Orphanet-Spain team. Aims: 1. Provide support and advice on ORPHAcodes implementation and use. Operation of National helpdesk. 2. Collaborate in the update of the MF to include the ICD-10-ES mapping to the latest nomenclature pack available.	A total of 69 requests have been attended in the period April 2024-May 2025. Update of the MF to include ICD-10-ES /ORPHA alignments for the differential files were concluded for the differential file published in July 2022 and partially finished in the case of the file released in July 2023. Unfortunately, our efforts to have our collaborators working on the ICD-10-ES/ORPHA mapping to keep it up to date has not been successful, and neither to involve new codifiers in spite of our dissemination attempts at trainings, email invitations and a dedicated presentation at a meeting held between the MoH and the Regional RD Registries (28th Jan 2025).
Further Activities: Nomenclature translation to national language: processing of abstracts and disease names (translation, validation process and upload).	404 abstracts/definitions and 2.550 names translated and uploaded between 1st April 2024 and 30th May 2025.

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

Carolin Wanntorp, information scientist of Orphanet Sweden

Besides their work on Orphanet, they are also active in supporting RD expert teams and contributing to a national infrastructure for care of RD 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:	
Webinar on Orphanet tool usage, Basic training	December 2024 (completed)
Production of education on ORPHAcodes and surrounding topics which will be published on the internal e-learning portal for Karolinska University Hospital.	In production, estimated release August 2025
Production of educational videos on ORPHAcodes and tool usage.	In production, estimated release August 2025
Web based ORPHAcode trainings aimed at ERN members	2 sessions planned for October 2025
Coding practice with clinicians from Clinical genetics at Karolinska, on specific cases	February-March 2025 (completed)
Presentation of Orphanet database and ORPHAcodes at Rare disease conference for Primary care at Karolinska	March 27th, 2025
Networking:	

Orphanet Sweden has ongoing collaboration with RaraSwed, the national RD registry in regard to ORPHAcoding.	2025 – ongoing
Ongoing collaboration with the Swedish rare disease patient organization regarding visibility of ORPHAcodes	2025 – ongoing
Ongoing collaborations with the national highly specialized care and ERN hub at Karolinska	2025 – ongoing
Meetings with selected ERN members to determine key points driving their need for ORPHAcode implementation in HIS	January 2025 (completed)
Meetings with the Swedish Board of Health and Welfare in order to communicate the need for adopting ORPHAcodes as a medical terminology and to align to the EHDS activities	June 2025 (planned)
Review of mapping between ORPHAcodes and the published articles in the Swedish rare disease knowledge database	Autumn 2024
Helpdesk Implementation:	
Promotion of GitHub helpdesk to clinicians	2025
Further Activities:	
ORPHAcode implementation:	
Finalization of documentation clarifying the need for ORPHAcode implementation in HIS.	March 2025
Contact with IT-dept at Karolinska to improve visibility of the Orphanet nomenclature of RD and communicate the need for implementation in the HIS.	October 2024 (completed), June 2025 (planned)
Promotion of Orphanet tools to clinicians	2024-2025

Discussion and conclusion

Depending on the state of coding in their respective countries, participants have continued to undertake extensive measures to achieve the WP4 goals. These measures have been summarized in the National Action Plans of Year 2 and 3, that are presented in this document. Meanwhile

ORPHAcodes are used in all the participating countries, either in all hospitals, in centres for RD, or in hospitals participating in European Reference Networks.

From April 2024 until May 2025 participating countries continued to conduct numerous trainings, workshops and webinars on ORPHAcoding and the Orphanet nomenclature of RD, both on-site and virtual. These training courses were provided to health care professionals like clinicians, general practitioners, medical students, medical coders and other clinical staff, hospital managers, IT specialists, software providers, ministry members and members of patient organizations. National hub members also participated in health conferences and congresses, as well as in scientific symposiums and health policy forums. Several 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. Examples include the preparation of online training courses in national languages.

Extensive activities have furthermore been undertaken to raise awareness on the importance 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 centres, human genetic societies and other national expert groups and medical societies. Contacts with providers of health information systems were intensified. The benefits of using ORPHAcodes were resented to hospital managers, umbrella patient organizations, national working groups and advisory groups on RD. 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.

All national hubs have implemented a national helpdesk. Almost all 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. Helpdesks without GitHub account have opted for organizing their helpdesk activities via phone and/or via email.

A lot of further activities were carried out. Various training and information materials have been produced and distributed like leaflets, flyers, fact sheets, information brochures, training or information videos in national language or Orphanet nomenclature of RD quiz, using them for education or national promotional campaigns. Furthermore, the translation of the Orphanet nomenclature of RD in more local languages has been extended. More activities were the publication of articles in medical journals, the cooperation in creating or supporting national RD registries, the production of coding files for national usage of RD coding or the support of ORPHAcode integration into e-health systems.

In addition to the measures already implemented, a whole series of further measures are planned until the end of the project in order to achieve the project objectives.