Orphanet Data for Rare Diseases 2

Final Meeting

OD4RD 2
Orphanet Data For Rare Diseases

This presentation is part of the project OD4RD2 which has received funding from the European Union's Health Programme. The content of this presentation represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission and/or the HADEA or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.

Agenda

9h30-10h15: Welcome Coffee-Networking session

10h15-12h30: Session 1

- Welcome messages

  DG Santé and HADEA representatives, OD4RD2 coordinator

- OD4RD2 achievements
- RD Codification : lessons learned
- Synergies with JARDIN
- Next Direct Grant Proposal: OD4RD3

12h30-13h30 Lunch break

13h30-16h30: Session 2

13h30-14h30: **Parallel working sessions (1)** to identify gaps and needs

1. Scientific collaborations with ERNs: nomenclature and scientific information
2. ERN registries: how to transform their data in reusable knowledge

14h30-15h15: **Parallel working sessions (2)** to identify gaps and needs

3. How Orphanet can help to disseminate ERN coding guidelines & other knowledge
4. Implementation & adoption of ORPHAcodes in MS

15h15-15h45: Coffee break

15h45-16h15 Plenary: Restitution of breakout sessions

16h15: Participant survey and Closing remarks

16h30: End of the meeting

Orphanet Data for Rare Diseases 2

Achievements-Final Meeting

OD4RD 2
Orphanet Data For Rare Diseases

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OUR OBJECTIVES

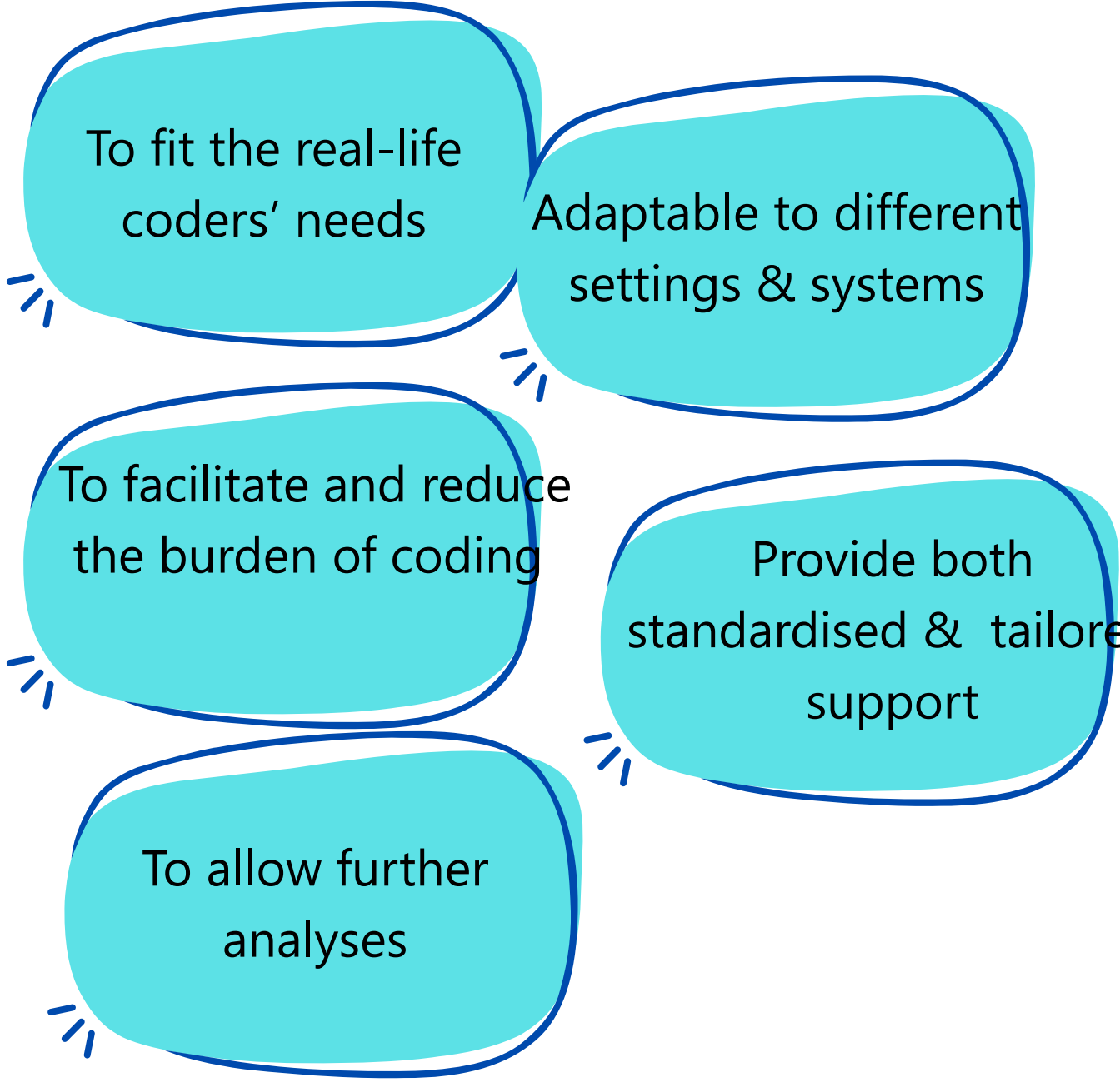
- To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, by the maintenance and the support to the implementation of the Orphanet nomenclature of RD, in collaboration with the European Reference Networks (ERNs).

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

- To contribute to supporting evidence-based decision-making in the framework of the European strategy around ERNs, by supporting the use of the reference corpus of data and information on RD


What do we want?
@OD4RD
OD4RD
Orphanet Data For Rare Diseases
ORPHA CODES

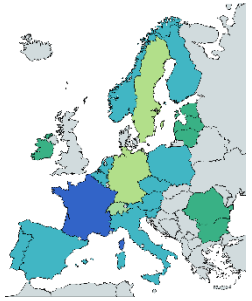
- ORPHAcodes production & maintenance , in collaboration with ERNs and experts
- Genetic annotations, definitions & transcoding information
- Delivered in different formats
- Network of Orphanet National Nomenclature Hubs: coordinated support for ORPHAcode implementation in HIS of 19 MS Hospitals that host ERNs
- Wiki and central Helpdesk accessible to the wider community



Reinforce the national level to add value at the European level

Generation of standardised exploitable RD data at the national level contributing also for EU-level insights

orphanet ⇄ European Reference Networks ⇢ EHDS

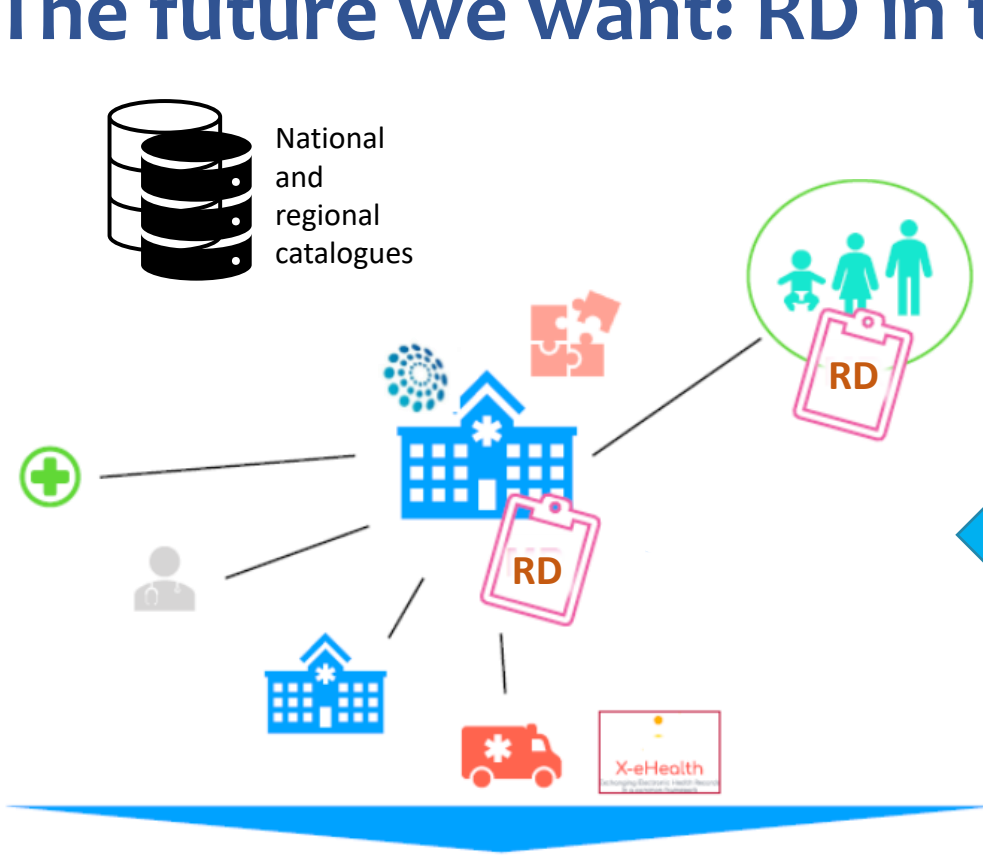


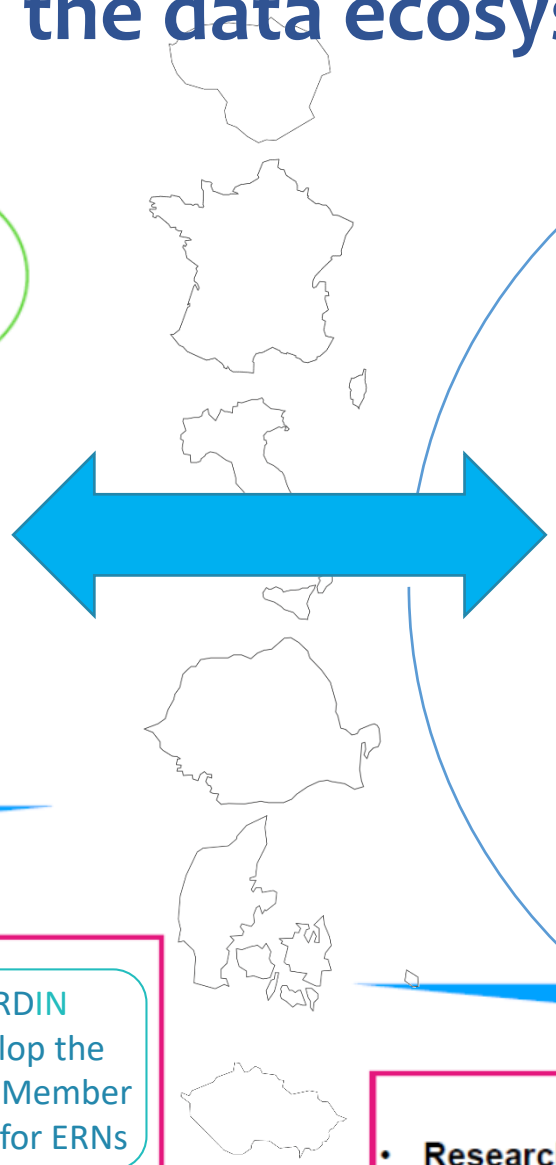
- National hubs
  - Information & data
  - ORPHAcode implementation support

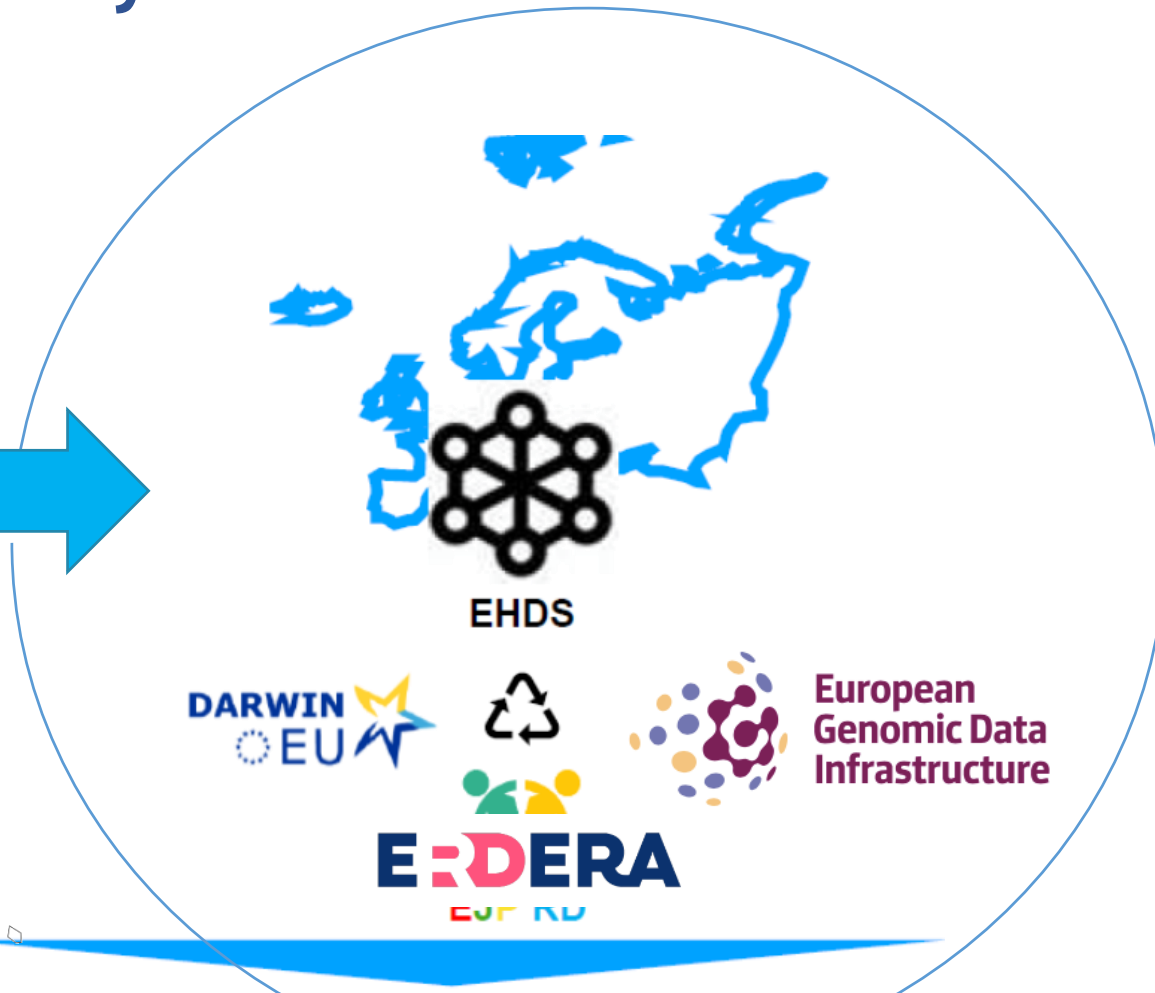
National nodes
- Care and research activities
- CPGs
- ORPHAcode implementation

National nodes
- HCPs' data
- EHRs
- ORPHAcode implementation

The future we want: RD in the data ecosystem







Primary use

- Better knowledge, best practices
- Continuity of care
- Better disability evaluation and compensation
- Adequate cross-border and primary care

JARDIN
Develop the role of Member States for ERNs

Secondary use

- Research
- Evidence-base decision-making

Connecting the dots for a seamlessly RD data ecosystem… and ERN integration in MS

ERDERA European Rare Diseases Research Alliance

orphanet

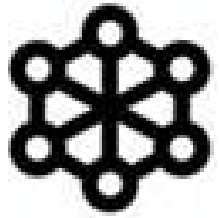
JARDIN

Rare2030
Foresight in Rare Disease Policy
Recommendation #7

EEHRxF

MyHealth@EU

HealthData@EU



EHDS

http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

ORPHACODES

Accurate representation of current knowledge & meeting users' needs thanks to daily literature survey & to the experts in ERNs' feedback.

6500+ Disorders

Classification revisions
15 revisions with 10 ERNs

1628 Genes-disorders associations

Disorders definitions & mappings to generic terminologies

Available in xml & API at a single-stop shop

NONH

Coordinated approach to ensure mutualisation & build capacity of the network while providing standardised and tailored support

Assess users' needs at national level

Organise capacity building around the Orphanet Nomenclature and tools

Support and advice implementation and use of ORPHAcodes

Coordinate partners to mutualise knowledge and build capacity across the network

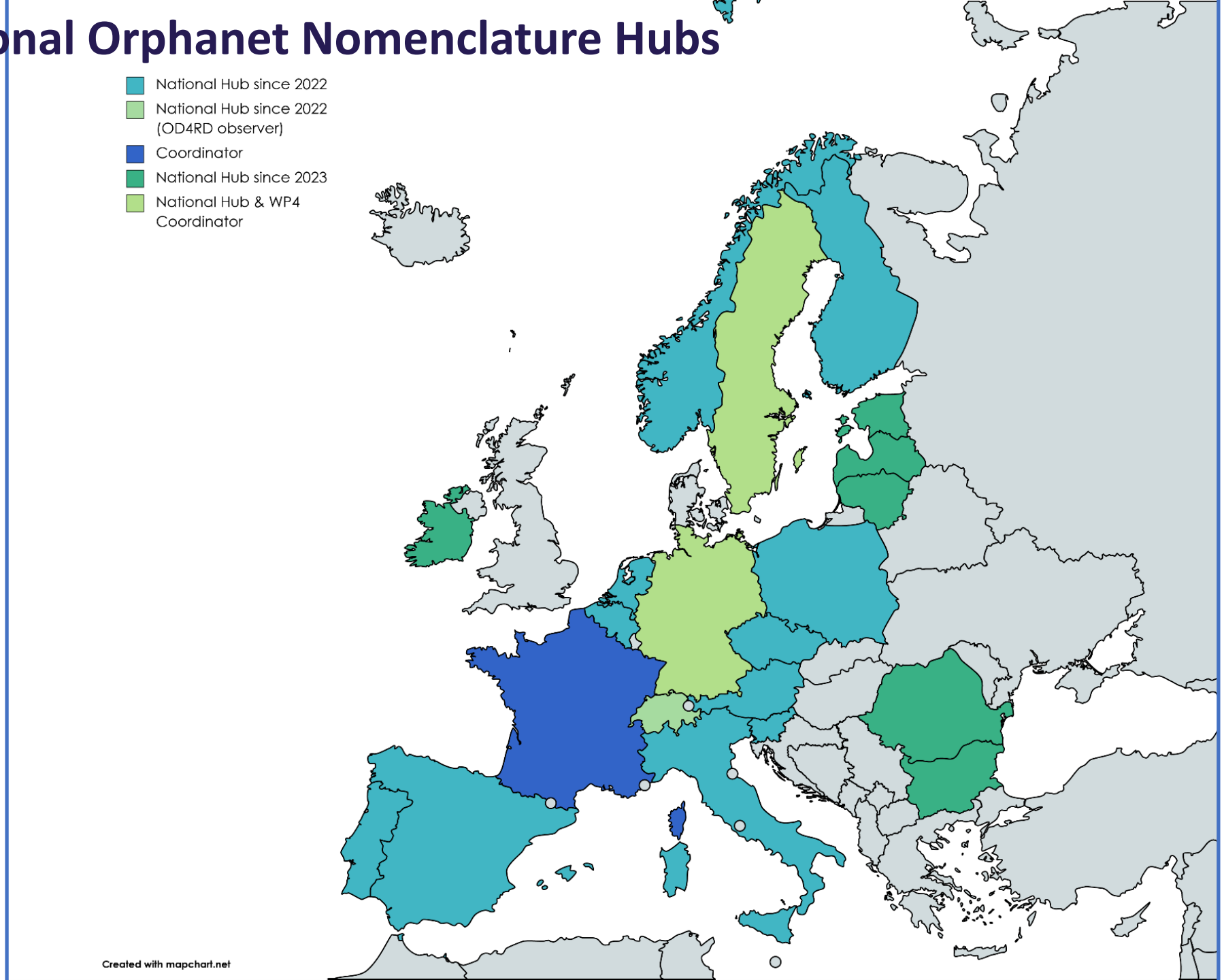
State of Play & survey of the ORPHAcodes usage in HCPs hosting ERNs

95 Trainings, 1800+ participants & 220 ad hoc events in national language in 19 countries

200+ demands received via the national helpdesk feeding the shared FAQ & Bets practices

2 TfT & a hands-on session + bi-monthly meetings and open sessions

OD4RD2 National Orphanet Nomenclature Hubs



National Hubs contacts

Austria: ursula.unterberger@meduniwien.ac.at

Belgium: Orphacodes.Belgium@sciensano.be

Bulgaria: info@raredis.org

Czech Republic: info@orphanet.cz

Finland: harvinaiset@thl.fi

Germany: orphanet@bfarm.de

Ireland: orphanet.ireland@mater.ie

Estonia: Vallo.Tillmann@ut.ee Sille.vahtra@ut.ee

Italy: orphanetitalia@opbg.net

Latvia: retasslimibas@bkus.lv

Lithuania: retosligos@santa.lt

Netherlands: orphanet@radboudumc.nl

Norway: Sjeldne-diagnoser@ous-hf.no

Portugal: info.orphanet@dgs.min-saude.pt

Poland: k.chrzanowska@ipczd.pl a.madej-pilarczyk@ipczd.pl

Romania: orphanet.romania@gmail.com

Slovenia: luca.lovrecic@kclj.si

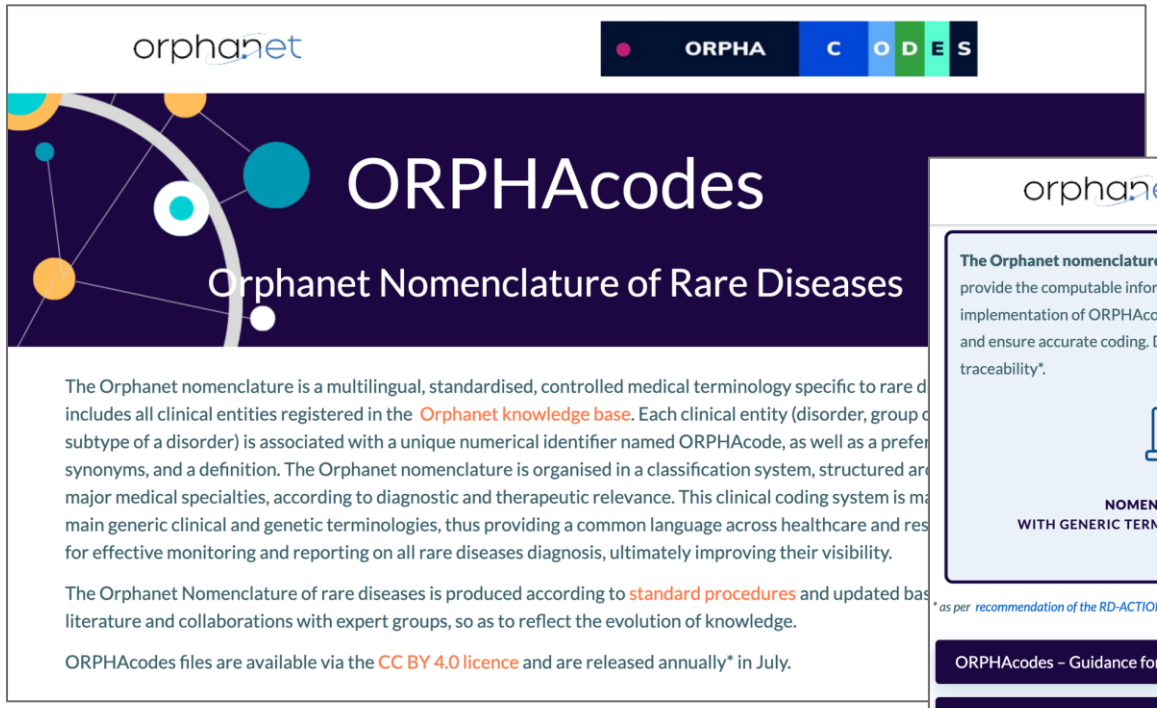
Spain: helpdesk.orphanet@ciberer.es

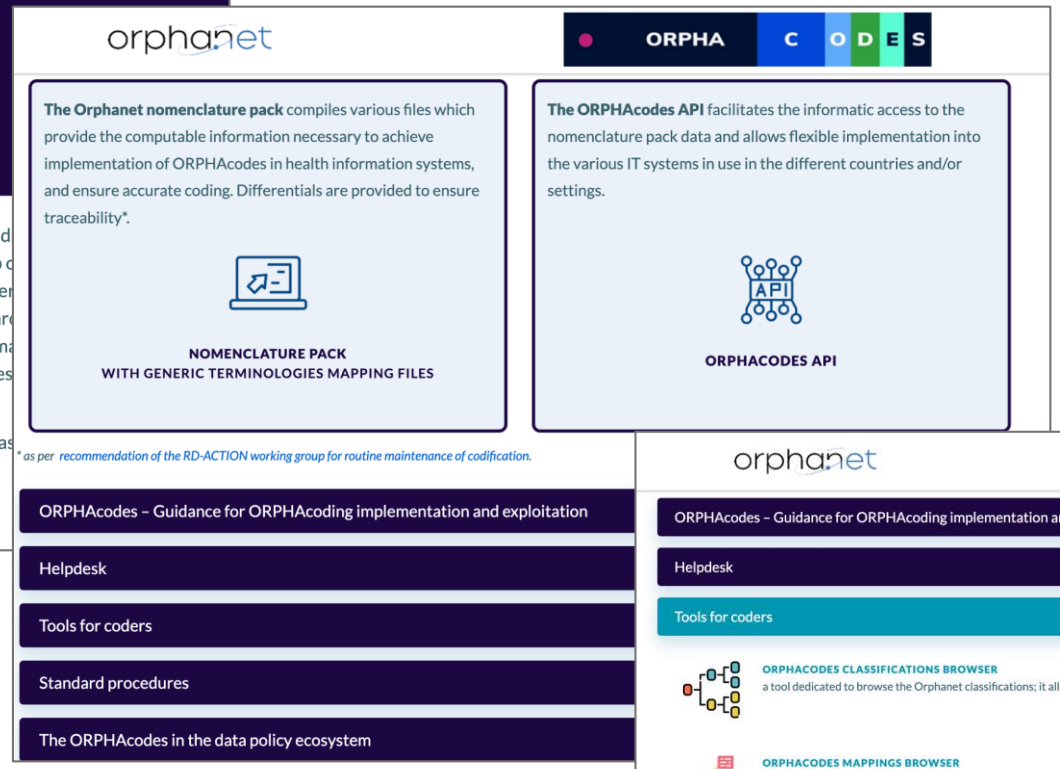
Sweden: orphanetsweden.karolinska@regionstockholm.se

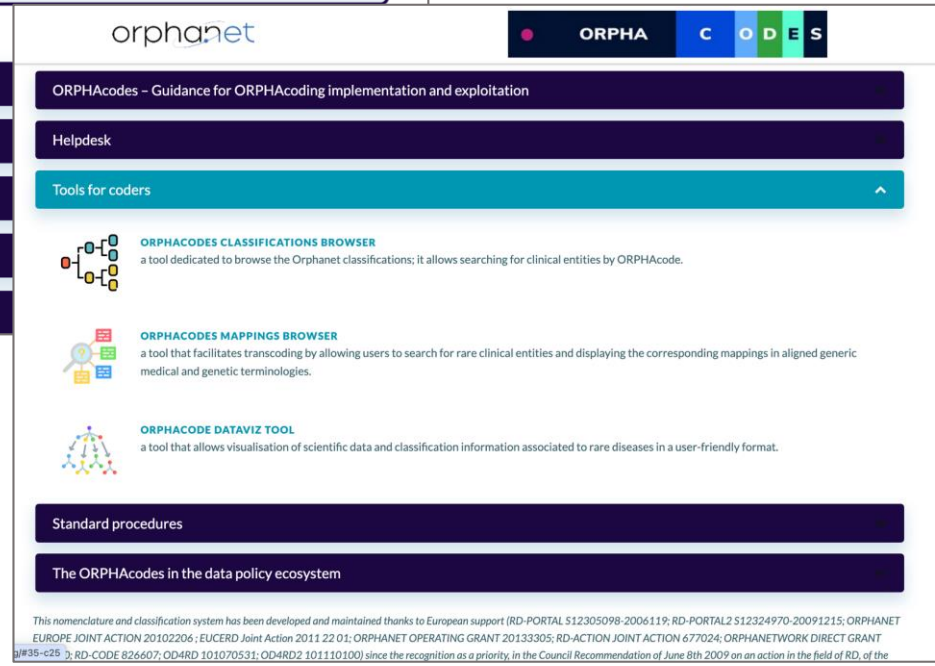
Switzerland: contact@orphanet.ch

Single-stop shop

https://www.orphacodes.org

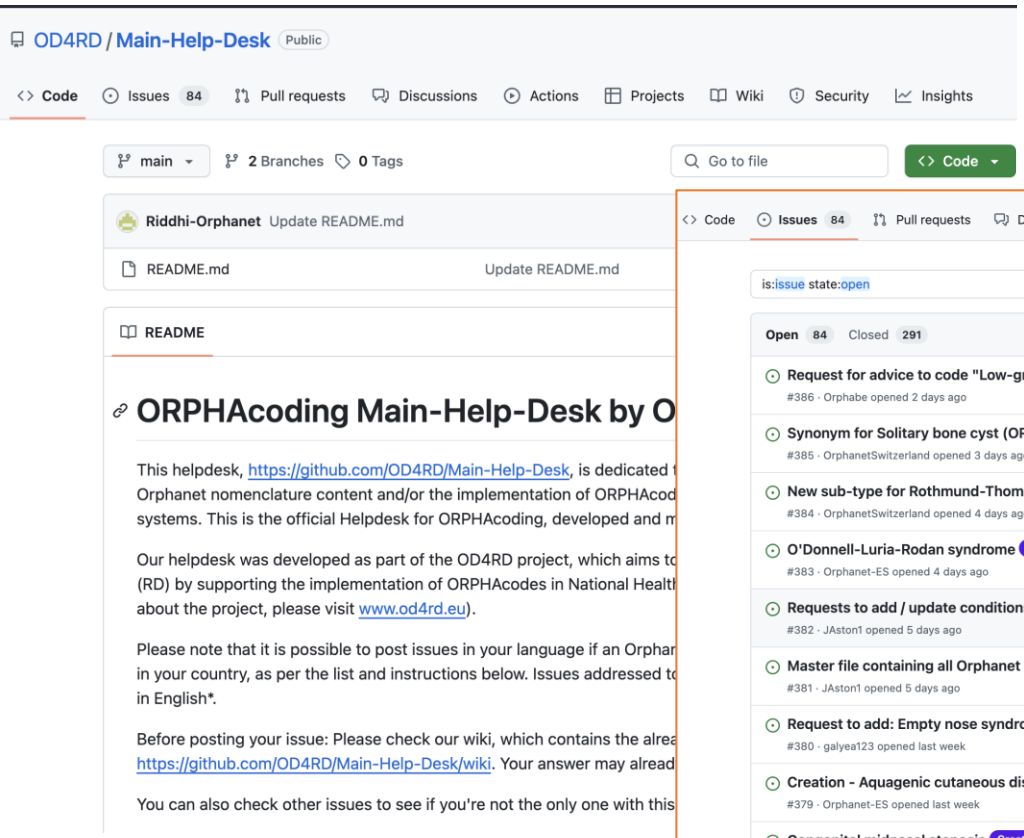


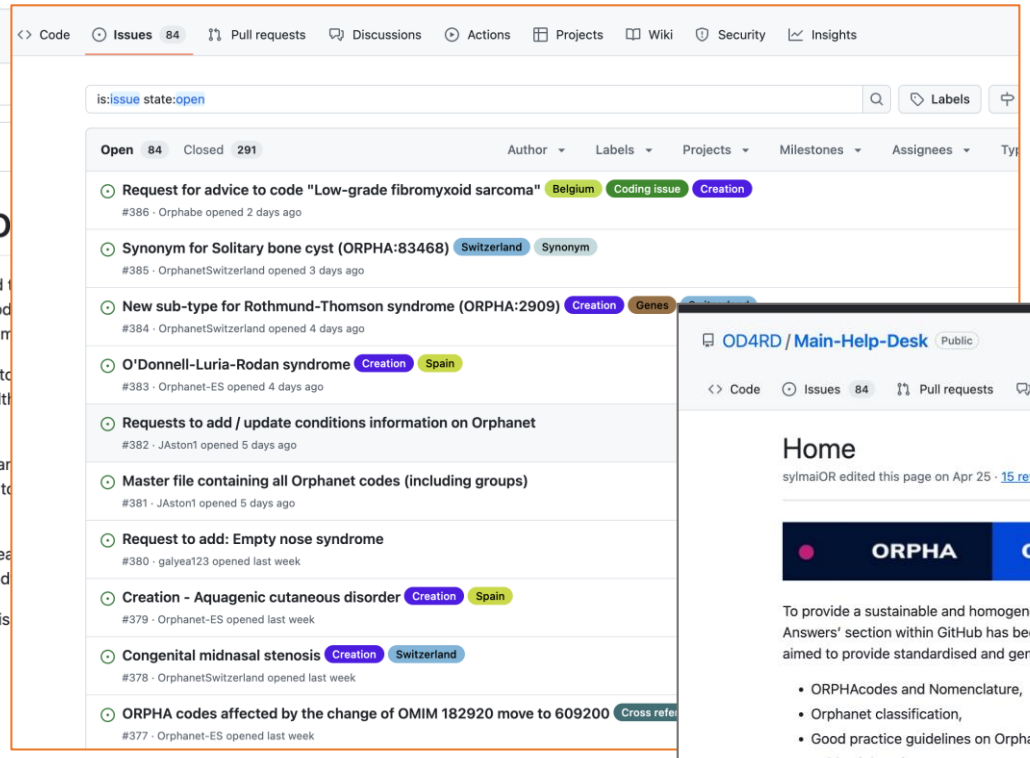


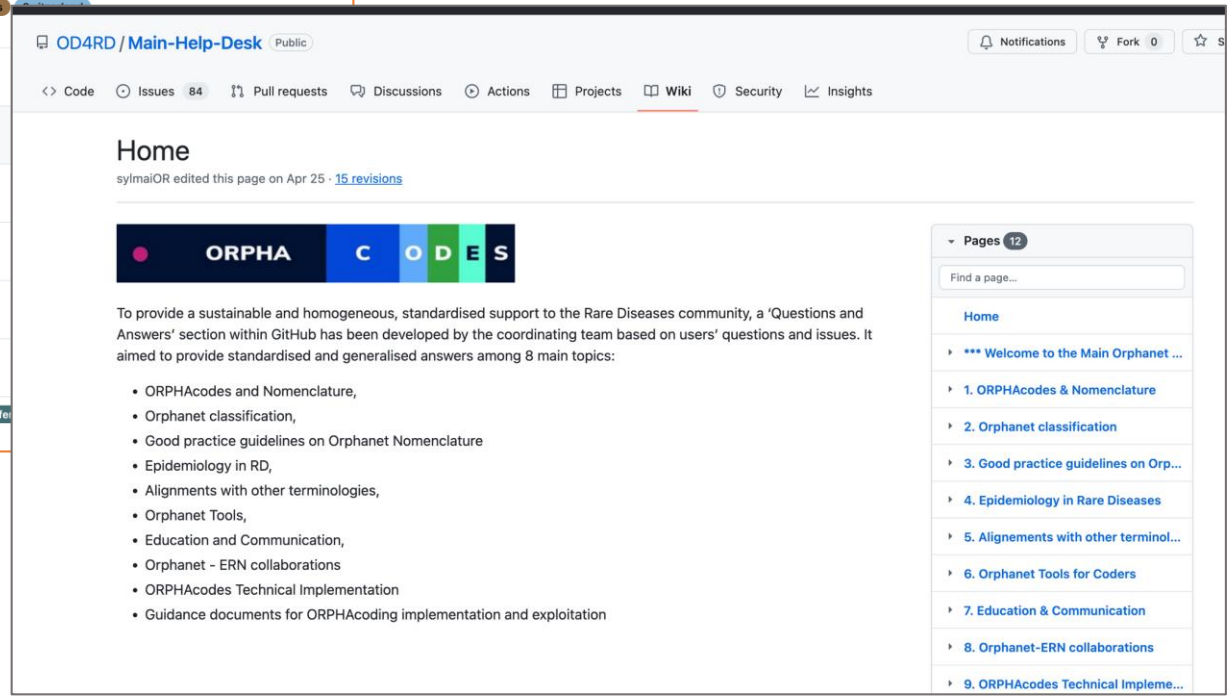


Helpdesk

https://github.com/OD4RD/Main-Help-Desk

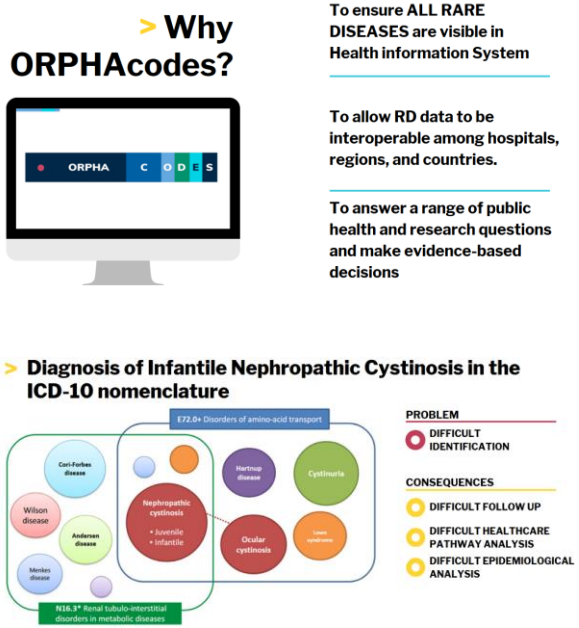






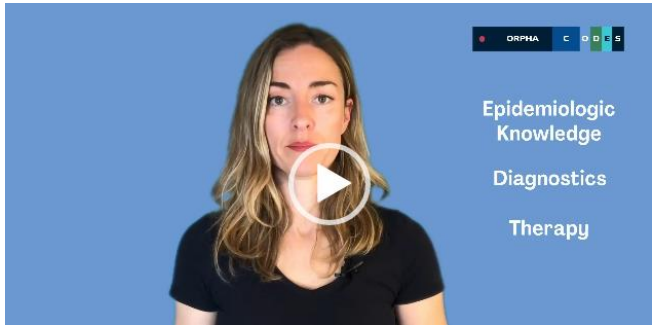
Training modules

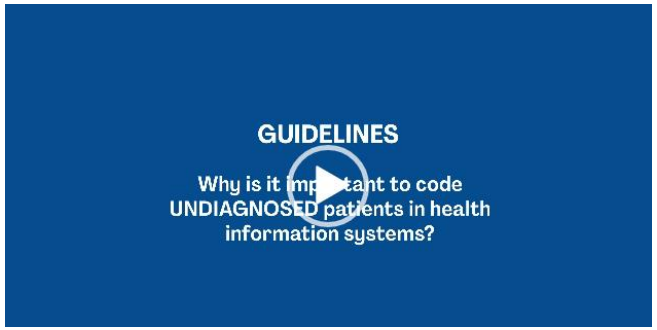
PRESENTATIONS



[ORPHA Coding – RD-CODE]

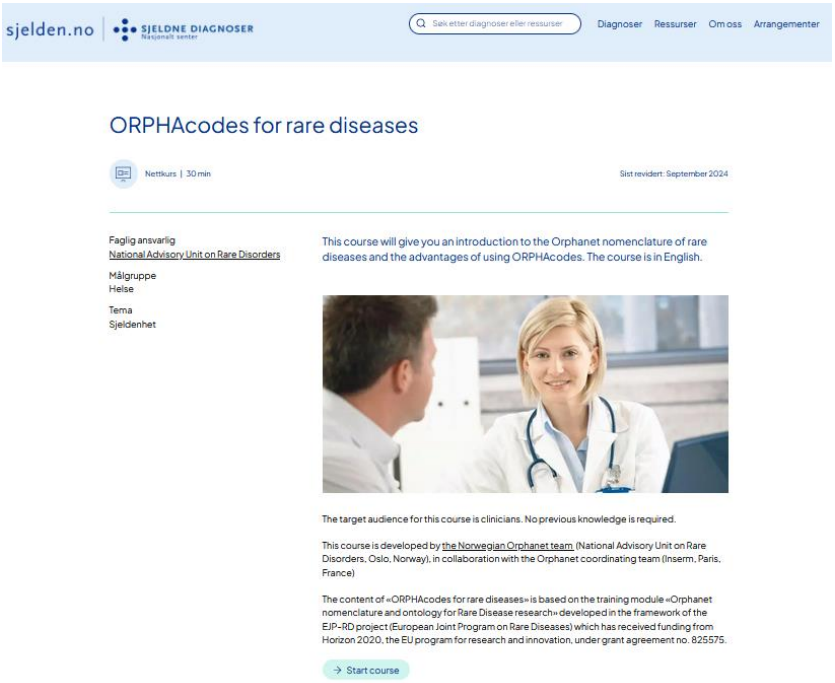
VIDEOS



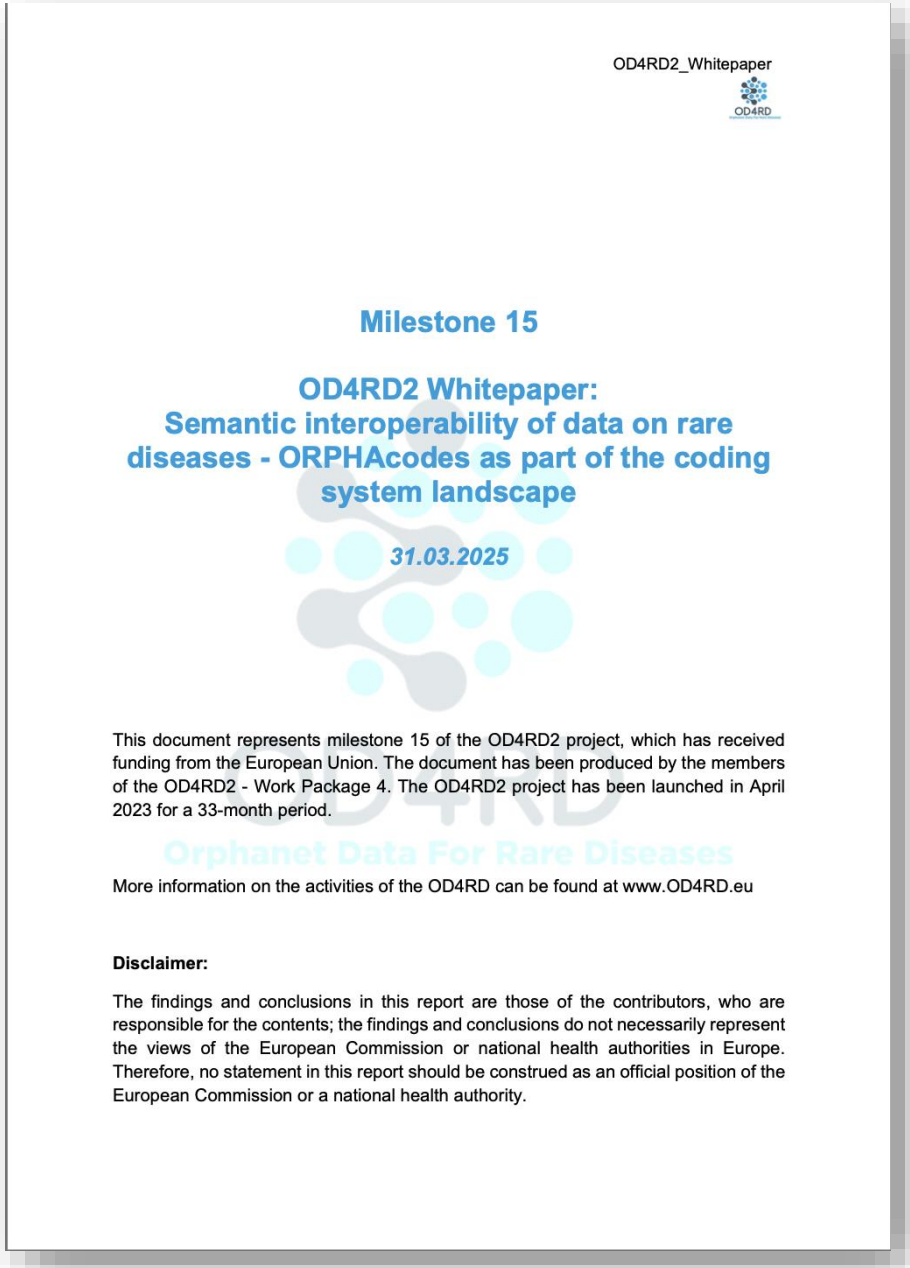


[Orphanet - YouTube]

E-LEARNING



[ORPHAcodes for rare diseases - Sjelden]


OD4RD2_Whitepaper

Milestone 15

OD4RD2 Whitepaper:
Semantic interoperability of data on rare diseases - ORPHAcodes as part of the coding system landscape

31.03.2025

This document represents milestone 15 of the OD4RD2 project, which has received funding from the European Union. The document has been produced by the members of the OD4RD2 - Work Package 4. The OD4RD2 project has been launched in April 2023 for a 33-month period.

Orphanet Data For Rare Diseases

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

Disclaimer:

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

OD4RD2 White paper

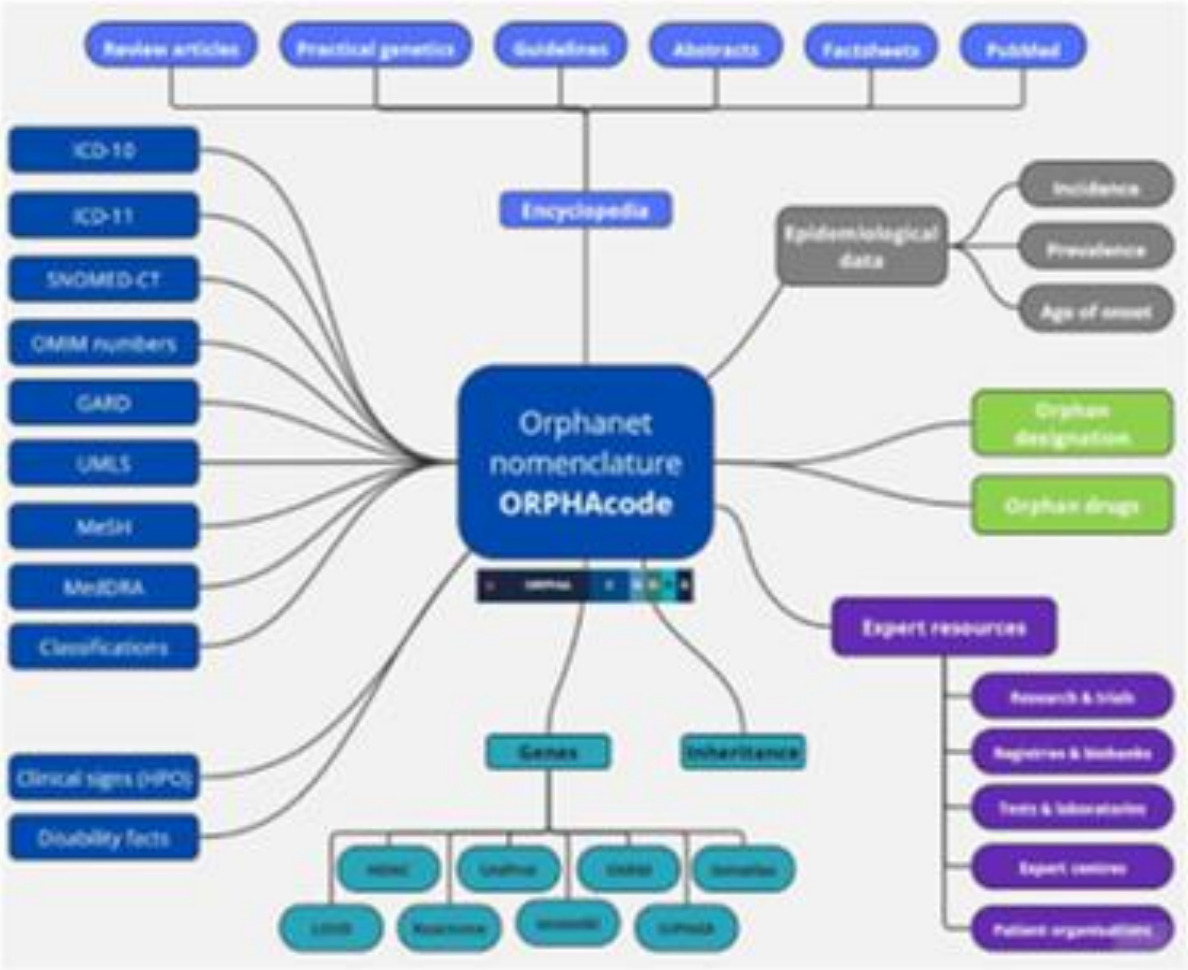
1. It is recommended to capture the respective **ORPHAcode for patients with RD at the point of care** and to enable the **inclusion of the ORPHAcode in the consecutive data flow.**

2. In case of a **joint use of two or more coding systems**, it is recommended to link the Orphanet nomenclature of RD to the other coding system(s) as much as possible and using **standardized, curated mappings so that at the point of coding, codes from both coding systems can be captured.**

3. To avoid future misalignment of the different coding systems, it is recommended to the **developers and guardians of the different coding systems** used in the disease space **to continue and enhance collaboration** on aligning the coding systems and enabling the joint use.

https://od4rd.eu/communication-material/OD4RD2%20MS%2015%20White%20Paper_final-V02.pdf

Evidence-based reports on ERNs' RD coverage



- Analysis reports on RD coverage, identifying overlaps and gaps amongst ERNs
- to support evidence-based decisions in the context of ERN coordination, Board of Member States (BoMS) and European Commission activities

The project also provides evidence to support ERN coordination, BoMS and the EC ERN strategy & decision making

Exploitation of the Orphanet classification and database

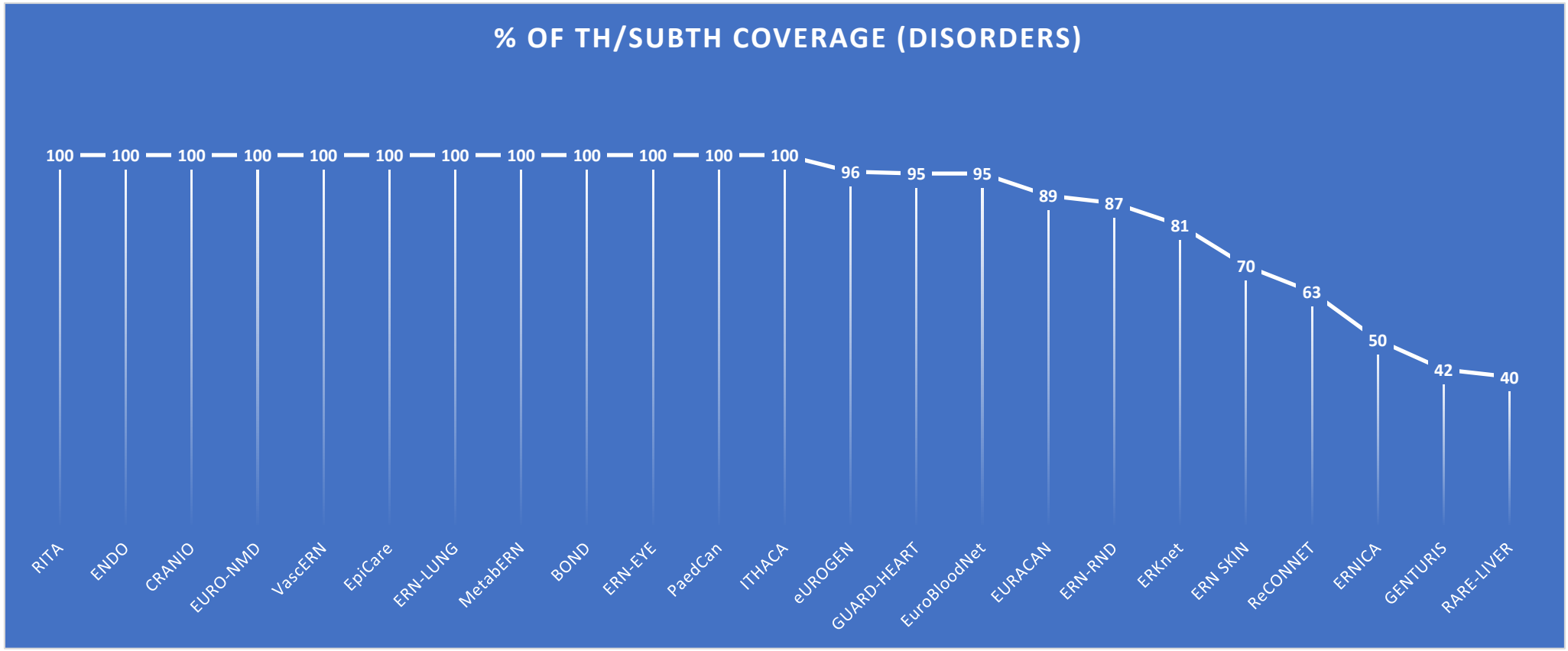
Mapping ERNs' thematic and subthematic areas to ORPHAcodes (groups of disorders)

Assuming all RD in a covered group are covered

Several validation rounds by 23 ERN* coordinators and WGs

* Excluding TransplantChild

Declared coverage of thematic/subthematic areas



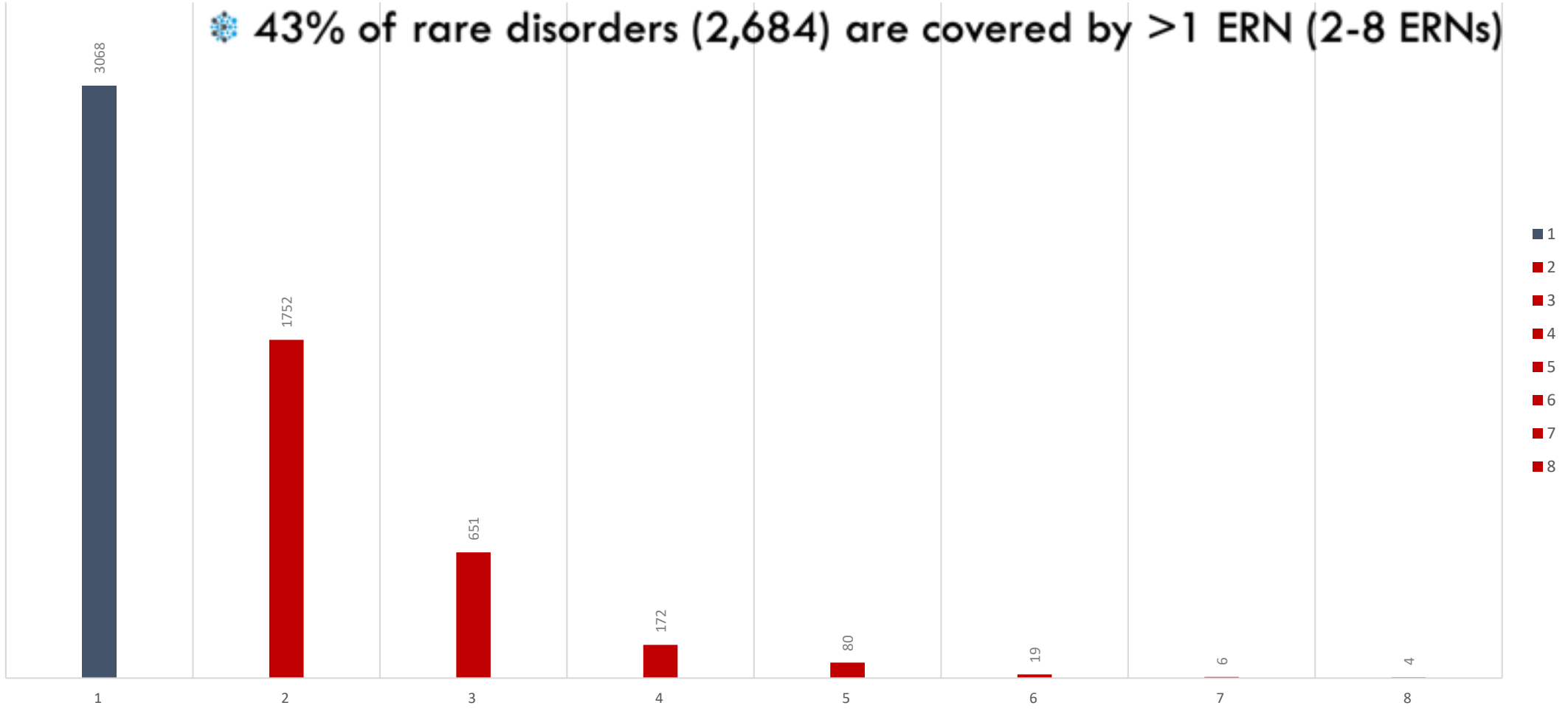
GAPS and overlaps

- After exclusion of infectious and toxic-related disorders:
  - Total gap, defined as RD not being covered by an ERN, is:
  - 7.45% (467/6,246 disorders, April 2025 version), not considering the declared demands for extension
  - 7% (442/6,246 disorders) considering the declared demands for extension
    - some ERNs asked for extensions for groups already covered by another ERN (ex. Vasculitis for ReCONNET, already covered by RITA)
  - 180 RD out of 442 RD not covered (41%) have prevalence data (EU or worldwide)

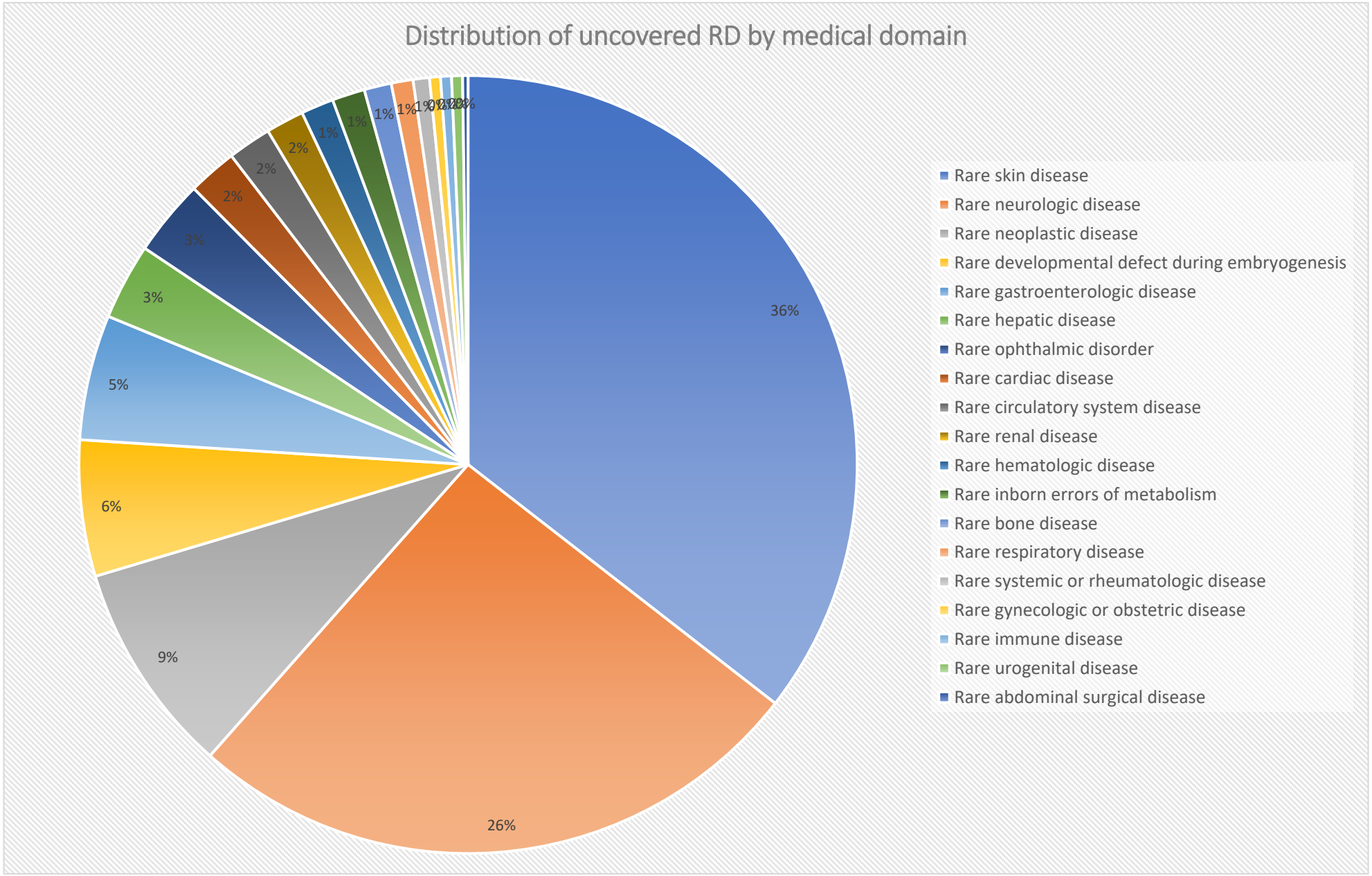
= 1.5 M persons in EU

- 6 other RD have point prevalence data by country and 8 RD have birth prevalence data (giving a total extrapolation of **1,760,000 persons** in EU not covered by an ERN)
- 32 RD have other epidemiological data (annual incidence; lifetime prevalence)
- For 114 out of 442 RD not covered (26%) the prevalence is still unknown in the literature, and 102 are yet to be documented

RD covered by >1 ERN



MAIN GAP AREAS



OD4RD2 outputs and impacts

- **Increase the visibility of RD** in Health Information Systems
  - By achieving real implementation in hospitals
- **Increase the quality of data** generated about RD patients
  - By disseminating good coding practices
- Empower ERNs, hospitals and the EC's **understanding on RD-related activities**
  - By providing means to generate accurate data for exploitation and analysis
- **Contribute to ERN integration** at the national level
  - By collaborating with National Hospitals and JARDIN
- **Contribute to the EU health data strategy**
  - By connecting the dots with structuring initiatives around EHR formats and health data spaces (EHDS)
    - For primary use: better diagnosis and care of RD patients, assessment of current practices and results against gold standards of care
    - For secondary use: informing policy decision-making and research

Conditions reasons of success

- A nomenclature that fits coding needs
- A nomenclature that allows for transcoding in HIS
- Well-trained National hubs, in capacity to support local implementation
- Very good connections at
  - Decisional level: MoH – Hospital managers
  - ERN coordination facilitating local level contacts and interactions
- Awareness and support from ERNs governing bodies
  - ERN Coordination
  - Board of Member States
  - European Commission

THANK YOU FOR YOUR ATTENTION!

OD4RD^2

Orphanet Data For Rare Diseas

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OD4RD
Orphanet Data For Rare Diseases

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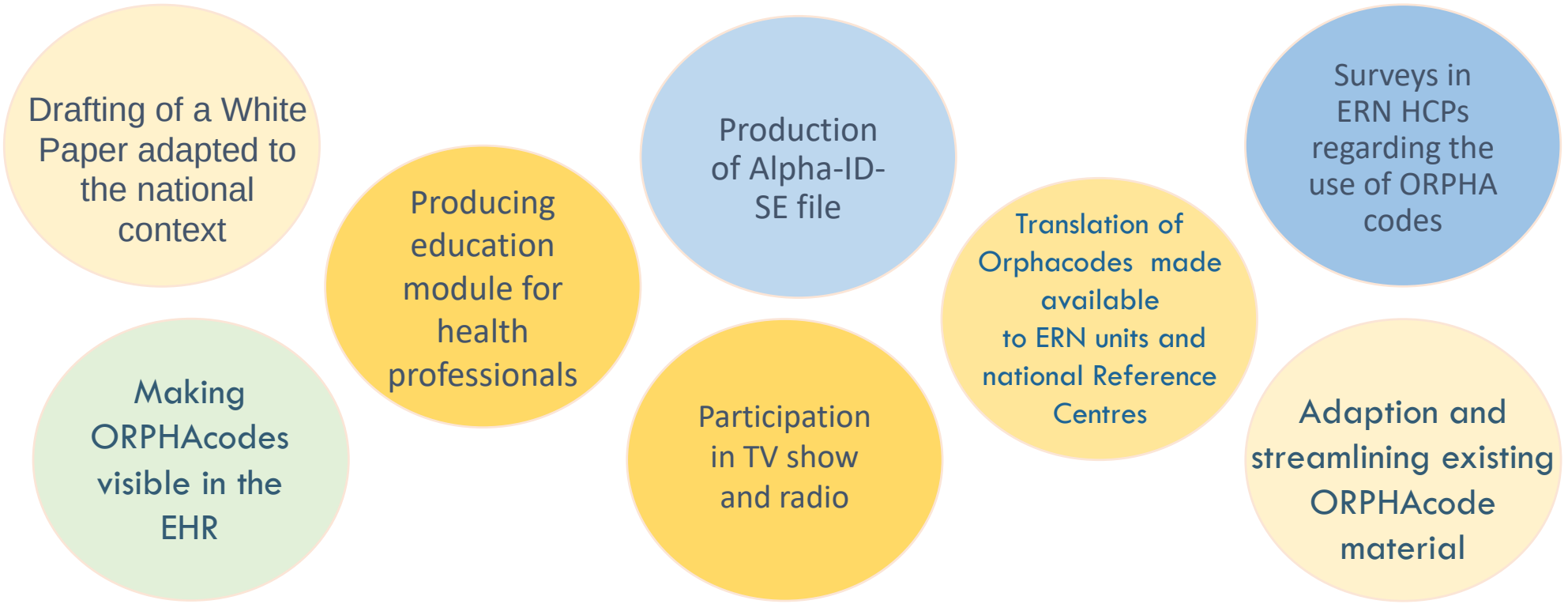
Objective WP4

- Provide coordinated support for ORPHAcodes implementation in Health Information Systems of Hospitals hosting ERNs in the participating countries and to harmonise ORPHAcoding practices across countries.



Activities

- Trainings
- Networking
- Communication
- Help desk

OD4RD ORPHANET National Hubs

LL data collection topics

State of Play – Impact

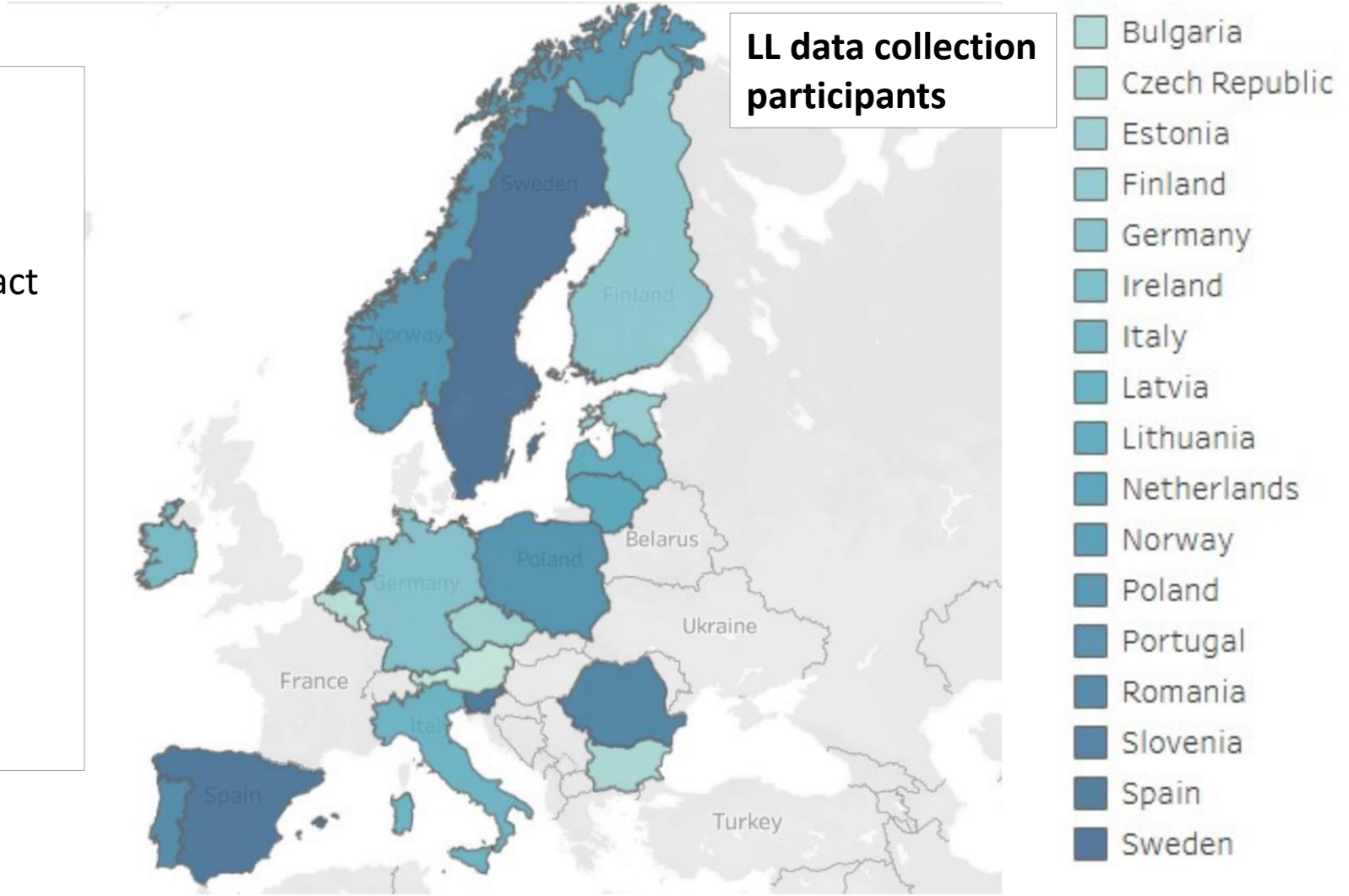
Activites – Effort and Impact

Trainings

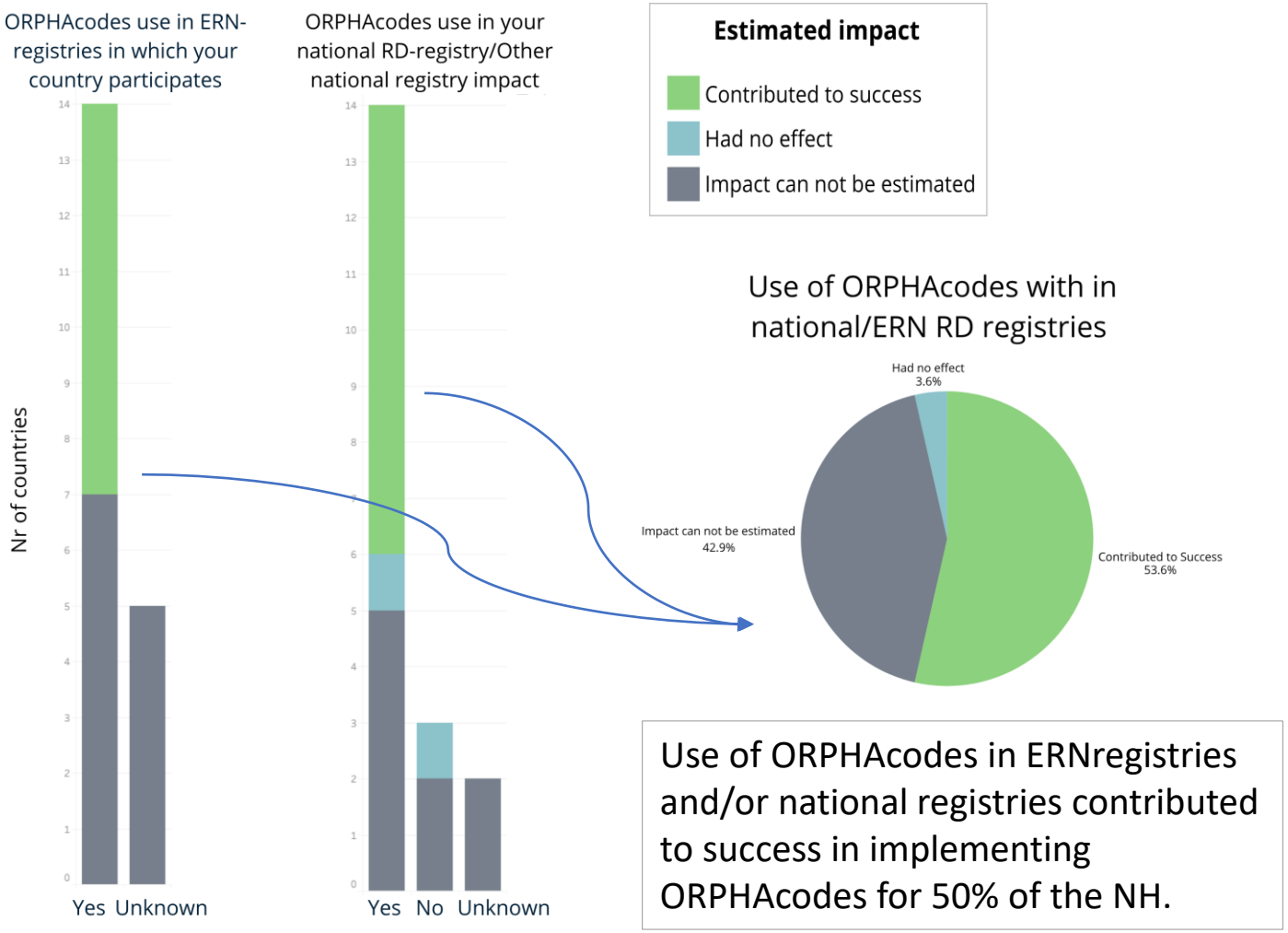
Communication

Networking

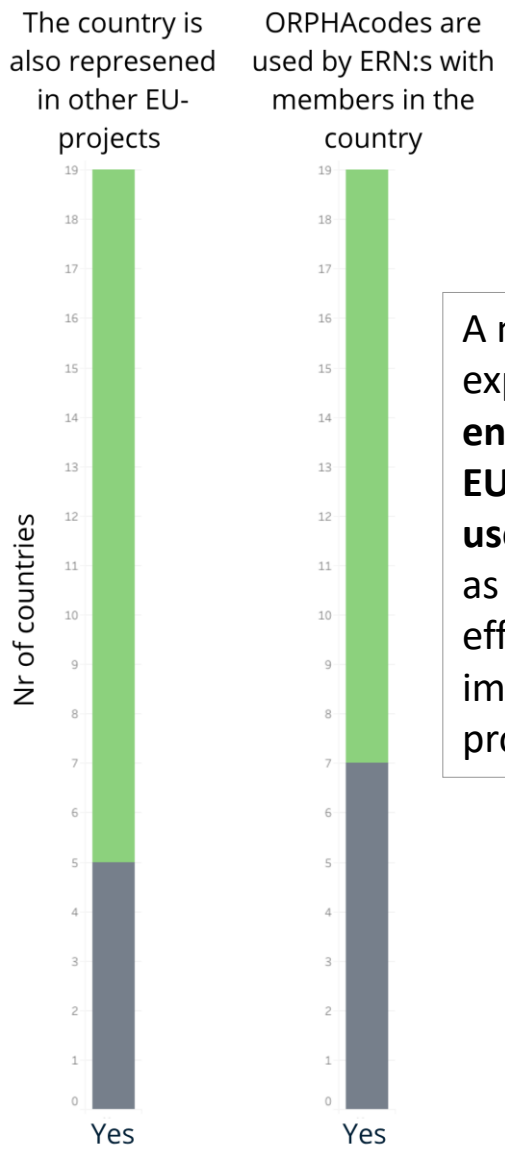
Action plans



State of Play Success factors



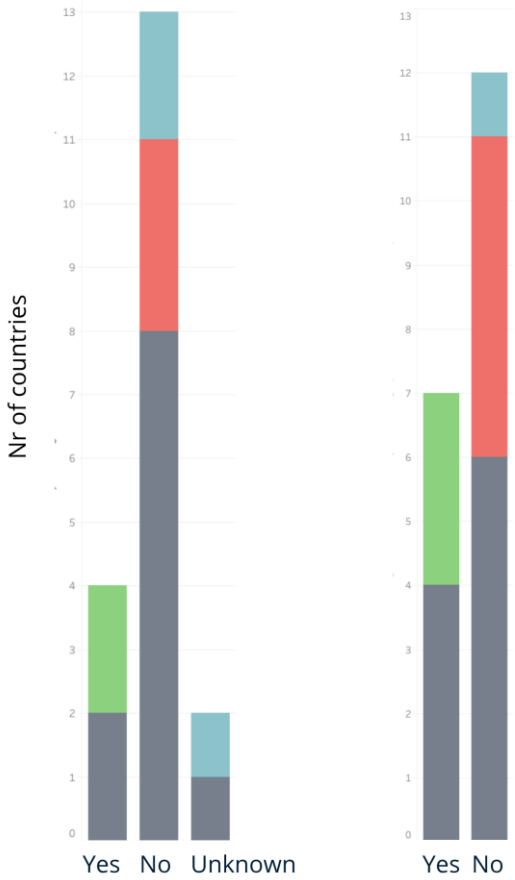
Use of ORPHAcodes in ERNregistries and/or national registries contributed to success in implementing ORPHAcodes for 50% of the NH.



A majority of the NH experienced **engagement in other EU-projects** and **ERN use of ORPHAcodes** as having a positive effect on the implementation process.

State of Play Success factors

ORPHAcode use incentiviced by affecting reimbursment calculation for healthcare services



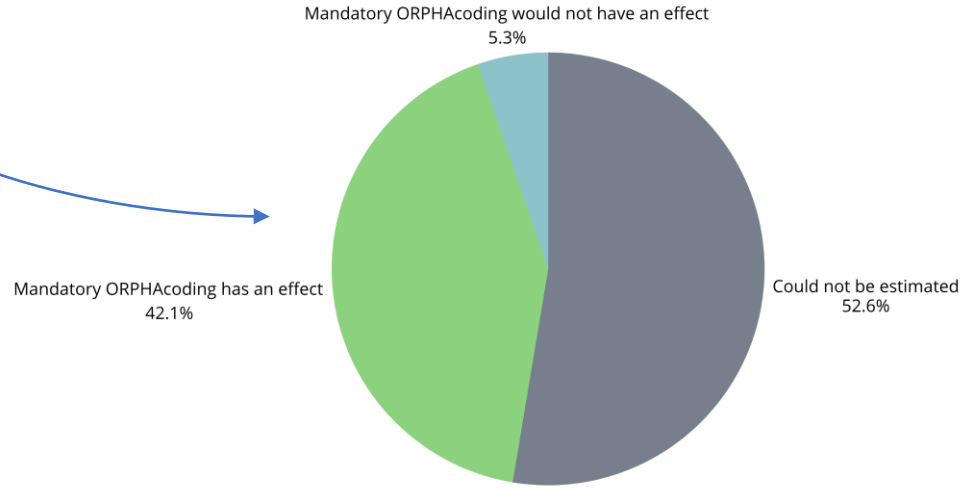
Using ORPHAcodes is mandatory when coding a patient with a rare disease

Estimated impact

- Contributed to success
- Had no effect
- Had a negative effect
- Impact can not be estimated

Incentivizing ORPHAcoding by linking it to reimbursement for health services is estimated to have an effect on implementation of ORPHAcodes by 25.3% of the NH

Estimated effect of mandating ORPHAcode use for patients with RD



Mandating ORPHAcoding is estimated by 42% of the NH to have an effect on ORPHAcode implementation

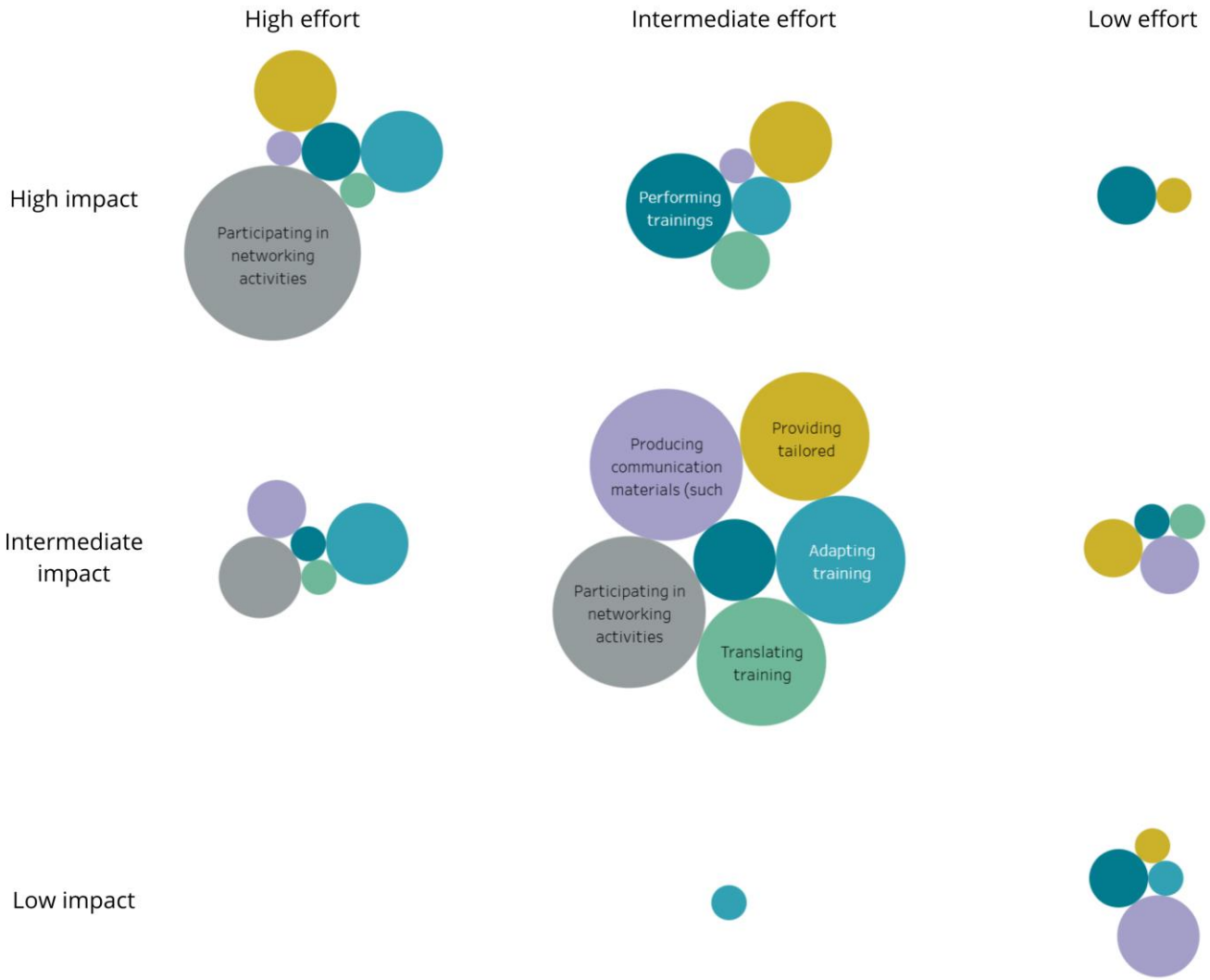
State of play - summary

- Engagement in European projects, use of ORPHAcodes by ERN:s and in registries have a positive impact on ORPHAcode implementation.

- If incentivizing ORPHAcode use by reimbursement has a positive effect on ORPHAcode implementation can not be determined.

- The experiences of the National hubs do not allow for firmly establishing that mandating ORPHAcode use for RD-patients has a positive impact on ORPHAcode implementation.

Activities- effort and impact

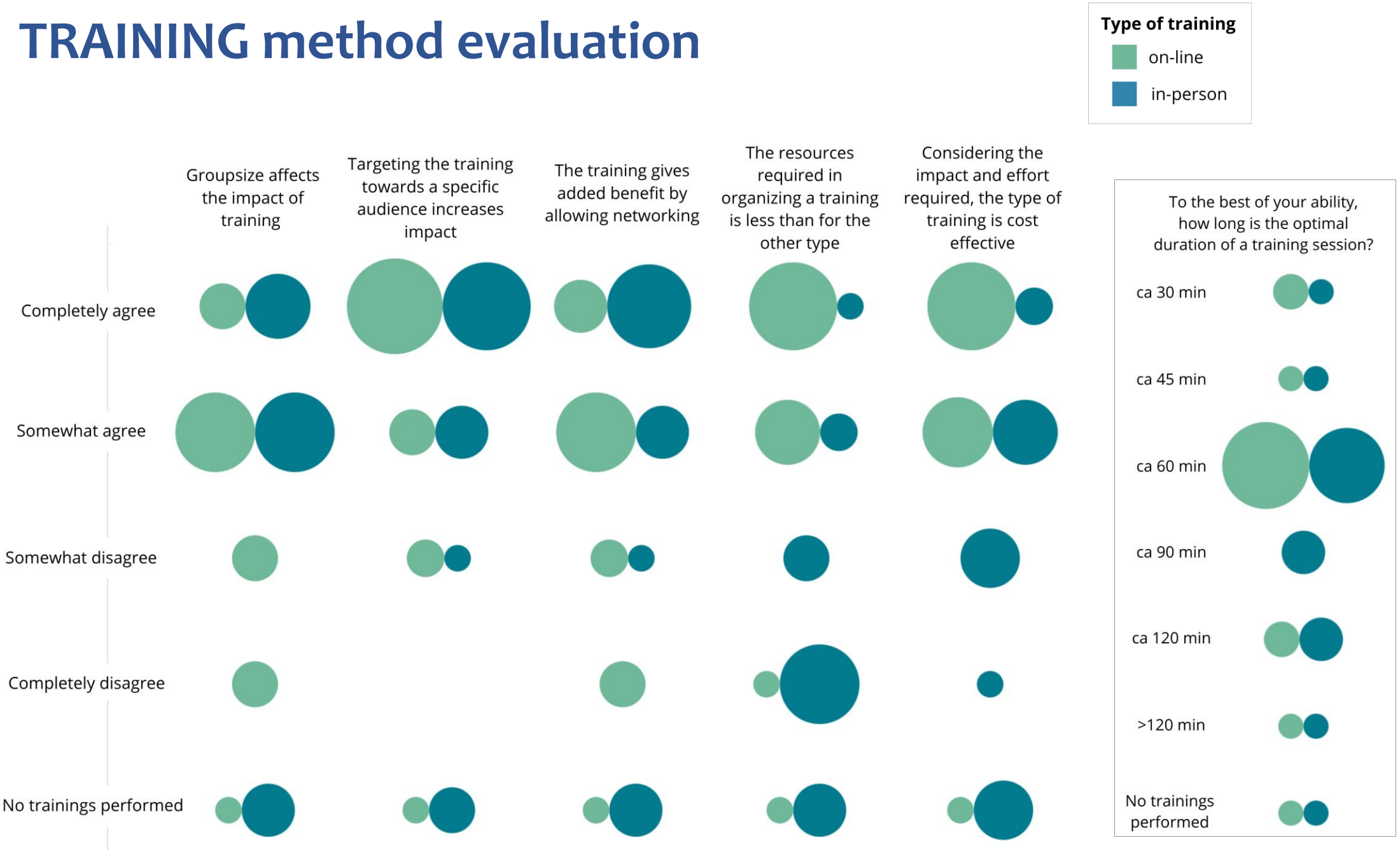


Activity

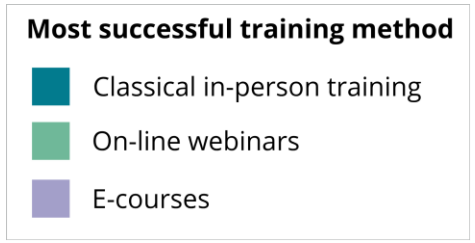
- Performing trainings which have been previously prepared
- Adapting training materials to new format
- Translating training materials
- Participating in networking activities
- Producing communication materials (such as flyers, porters, articles, etc)
- Providing tailored materials on request (such as lists, mappings, etc)

Results indicate a maintained balance between effort required and impact achieved for performed OD4RD2 activities

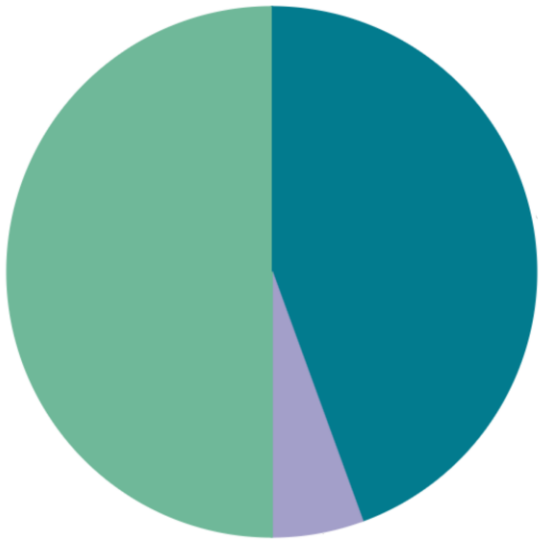
TRAINING method evaluation

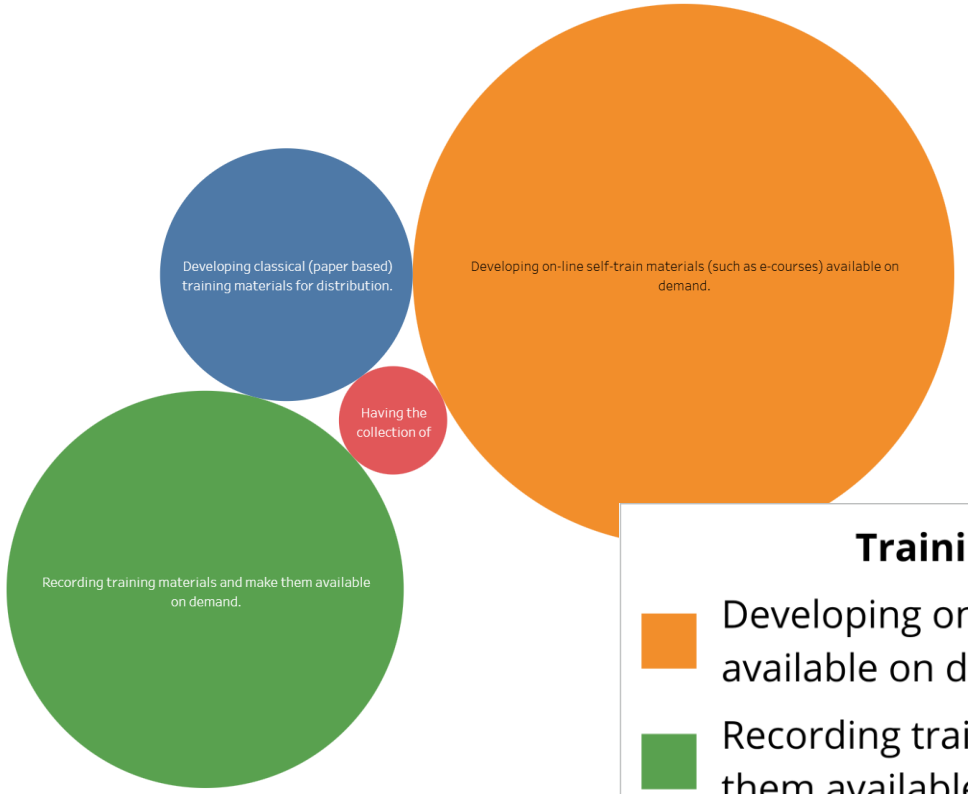


Training methods

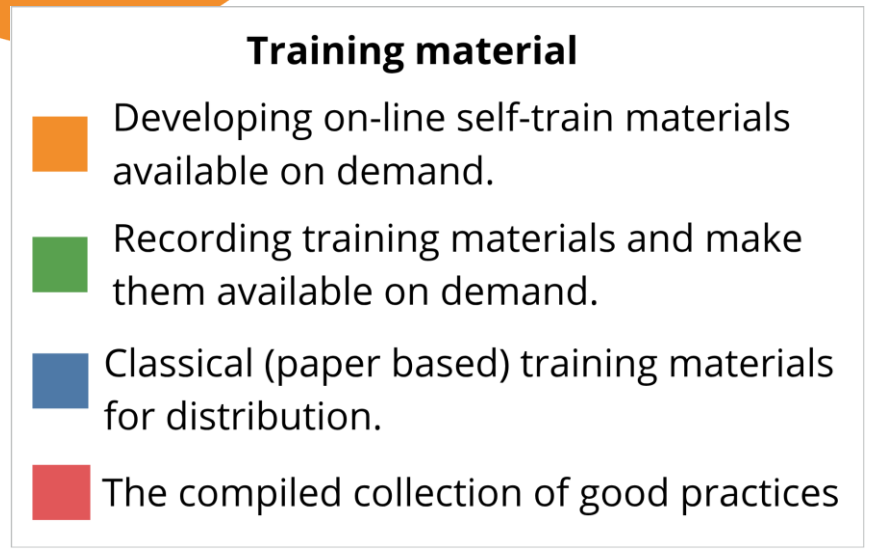


Most successful training method





Which additional training materials would help circumvent the obstacle of required time?



The time required to attend trainings is identified as an obstacle to professionals participating in trainings by 17 of the NH.

Activities & Trainings - Summary

- The national hubs' assessment of effort and impact for OD4RD2 activities indicates a **well-balanced relationship between input and outcome.**
- Targeting trainings towards the intended recipients is a key success factor for all types of trainings.
- On-line trainings are considered cost-effective by a majority of the National Hubs and require less effort than in-person events.
- Groupe size affects the effectiveness of on-line trainings, but to a lesser extent than for in-person events.
- On-line trainings provide an opportunity to network, but less so than in-person events.
- The optimal duration of an in-person training is 60 or more, while being 60 minutes or less for on-line trainings.
- The time required for attending trainings is an obstacle to participation, this can in part be mitigated by developing training materials available on demand such as on-line self training and/or recordings

Networking

most successsfull

Least successfull

Type of networking

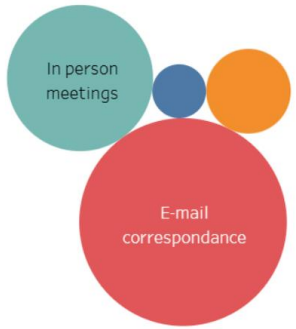
- In-person meetings
- E-mail correspondance
- Attending events arranged by a different organization
- Conference/direct calls

All networking methods have been successfully employed to connect with the target population by more than 50% of the countries.

Health care proffessionals

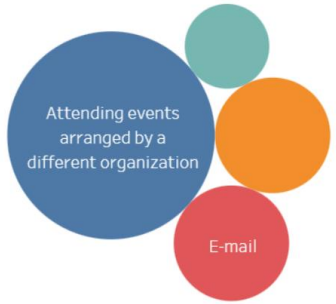
Target with most success

Health care decision makers

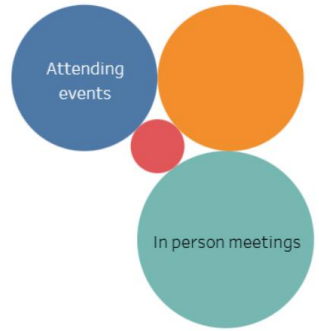


Target with least success

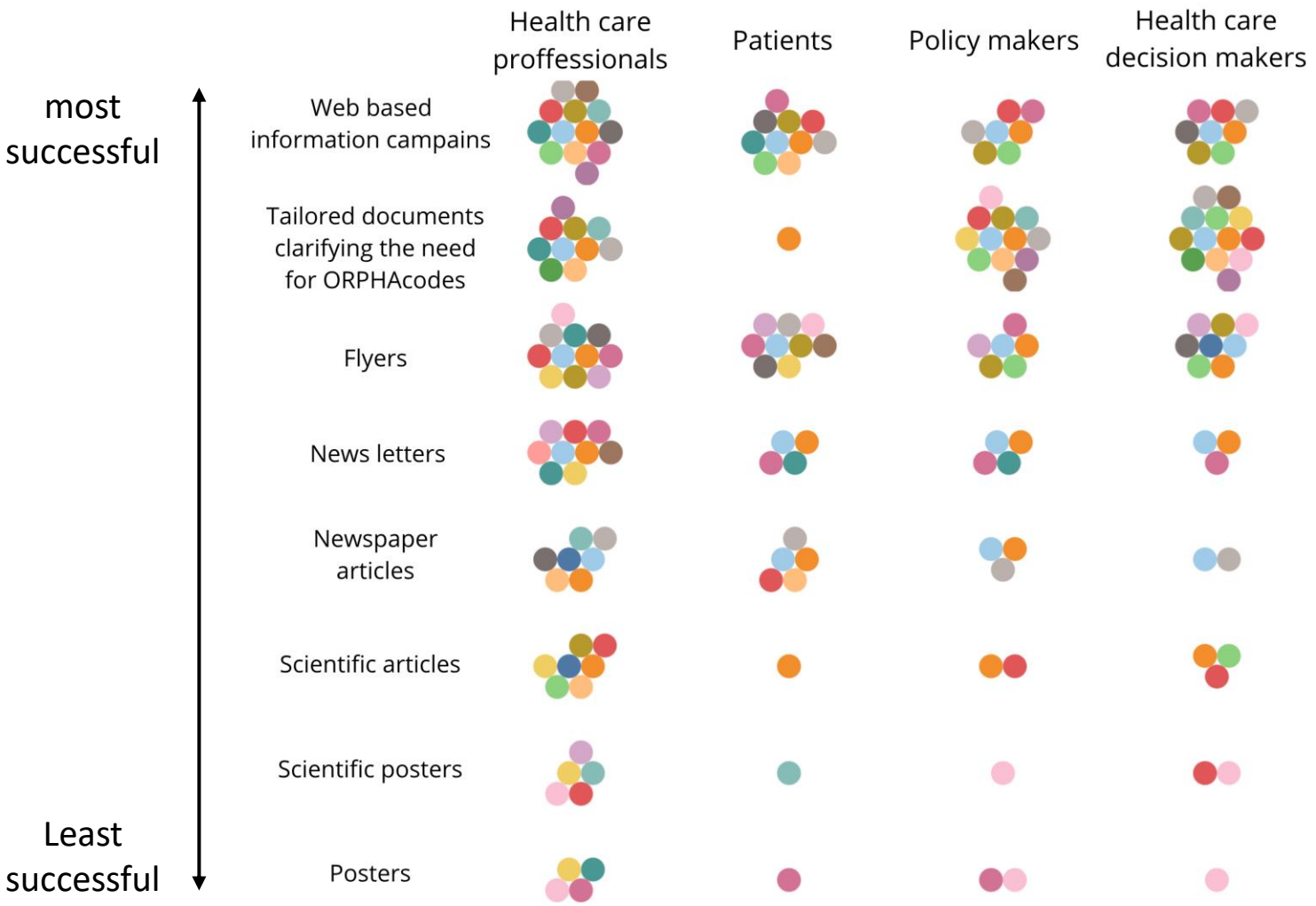
Patient organizations



Policy makers



Communication materials



Additional successful communications

The most important method of communication to HCPs are direct in-person interactions, trainings, RD and ORPHA coding presentations included to specialization courses, dedicated scientific meetings / to patients - direct in-person interactions, presentation during PAG meetings / to policy makers and heath care decision makers - dedicated
presentations on RD and ORPHA coding / for all audience - RD Information Platform on governmental website

Participation in TV show and radio

Public hearings, social media posts

Drafting of a White Paper adapted to the Belgian context and sent to all decision-makers (Ministry, Terminology Centre, FPS Public Health, RIZIV-INAMI). In response, we have been invited to several meetings to address this topic. But unfortunately, it is still SNOMED CT that is promoted by the Belgian authorities as the terminology for primary coding in all electronic files

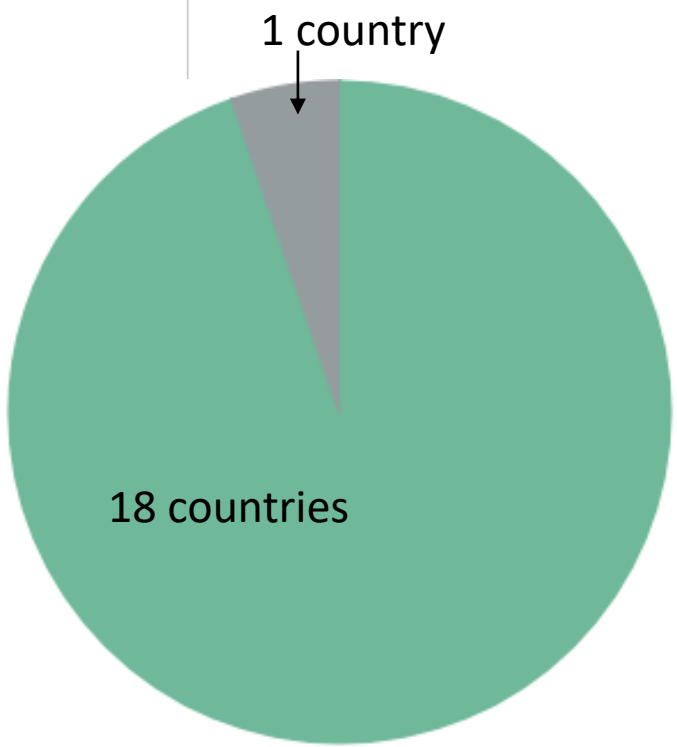
Rare Disease education module for health professionals published in Mar 2025 - included subsection on Orphanet and Orphacodes. Presentation on importance of orphacodes to MoH during development of new rare disease strategy (published Aug 2025)

meetings with groups of clinicians in HCP belonging to ERNs; involve patients in communication campaigns

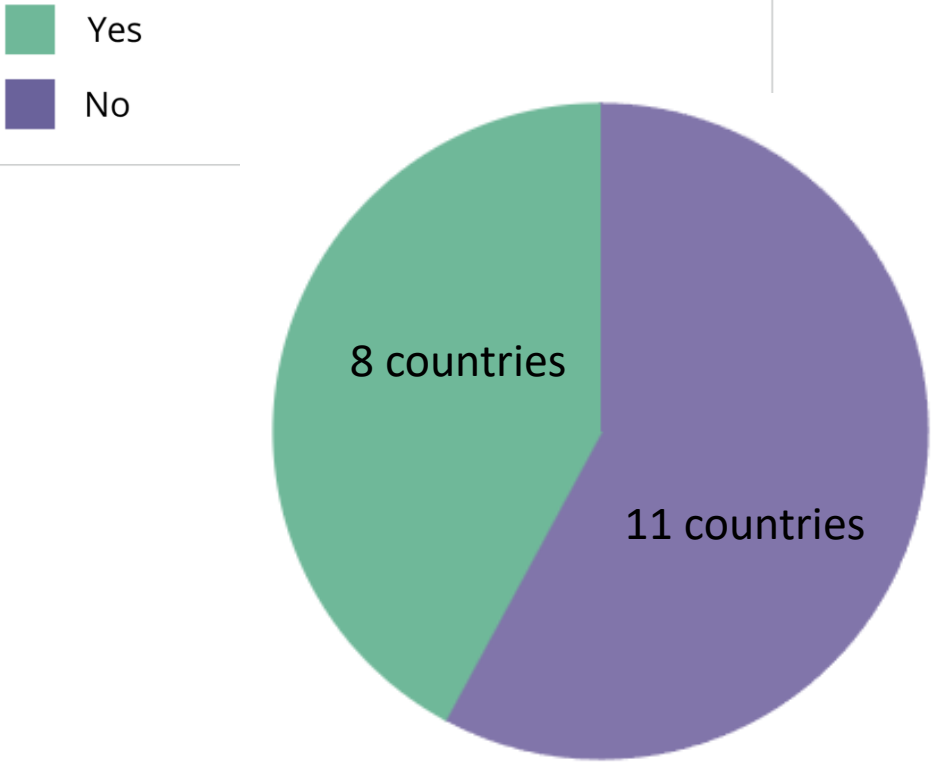
National conference for rare diseases, National RD expert centers annual meetings

Action plans

Compiling a national action plan was a fruitful method for planning, tracking and structuring our national activities.



In your Actionplans produced during the project, did you include any successful actions which has not been covered by the previous sections of this form (networking, training, information materials?



Additional successful activities

Bulgaria

Applied best practices from other countries, questionaire development for RD expert centers, online trainings, scientific publications

Portugal

To provide an up to date translation of Orphacodes available to the ERN units and national Reference Centres

The Netherlands

Getting ORPHAcodes made mandatory in the to apply for designation as an expert centre for rare diseases. Local EHR service teams have added specific fields for ORPHAcodes to the HER (as such field isn't provided by the EHR provider)

Belgium

Accreditation of the OD4RD2 training sessions: the training given to Belgian doctors as part of the OD4RD2 project is accredited since 2025

Poland

ERN HCPs surveys on use of ORPHAcodes gave opportunity to identify these centres, which are currently only ones in Poland to have been nominated by the Ministry of Health for ECRD). Establishing cooperation helped promote idea of ORPHAcoding, provide trainings and lobby for development of Common strategy for RD patients

Finland

Collaboration with national registries (malformation registry, abortion registry, Kanta registry, national care registry for healthcare)

Germany

Production of Alpha-ID-SE file , use mandatory for RD cases in inpatient sector

Norway

Development of ORPHAcoding module in EHR system, and its implementation in two of four health regions. E-learning course

Networking, communication & action plan - Summary

- All modes of networking (e-mail, meetings, attending events, direct calls) have been successfully employed by at least 50% of national hubs for each intended target (professionals, decision makers, policy makers, patient organizations)

- Non scientific communications materials were successfully employed by all national hubs with web-based information campaigns being the most reported successful measure.

- Targeted communications clarifying the need for ORPHAcodes were the most successful communication used for reaching policy makers and healthcare decision makers.

- A wide array of different communication activities have been successfully employed during the project!

- **Using an Action plan to organize national activities was a great success!**

How would you improve?

Key areas for success moving forward

- Closer collaboration with ERN:s
- Even stronger targeting of the HCP decision makers/policy makers
- Sustained emphasis on training
  - E-courses, videos, TfT
- Continued focus on implementation in HIS/EHR

Spain

a higher interaction with ERNs representatives at national level would be desirable but, to be more effective, it should be encouraged by the ERN coordinators following a top-down approach.

Norway

Reimbursements for orphacoding.

Poland

Promoting Orphanet-ERN collaboration

Romania

prioritizing the involvement of decision-makers

Italy

Communication campaigns regarding the importance of the Orpha Codes implementation in HCPs

Finland

Stronger participation of hospital and electronic patient record experts in the project at all levels

The Netherla

We would expand the national team and dedicate more time to creating e-leanings

Austria

There can never be enough training and communication

Italy

initiatives to strenghten the relationships with ERN representatives /members

Spain

Perhaps the only change I would make it would be to prioritize the activities targeting decision makers.

Norway

higher priority for activities targeting health authorities in order to make orphacoding mandatory.

Poland

Training for trainers at an earlier stage.

Poland

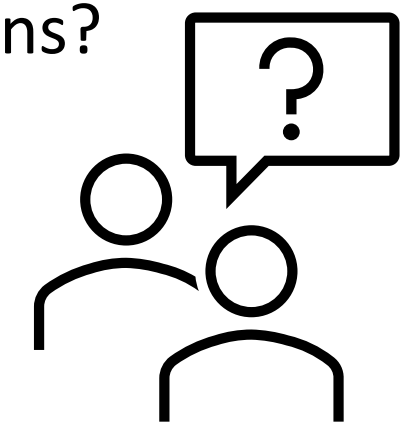
Strengthening training activities

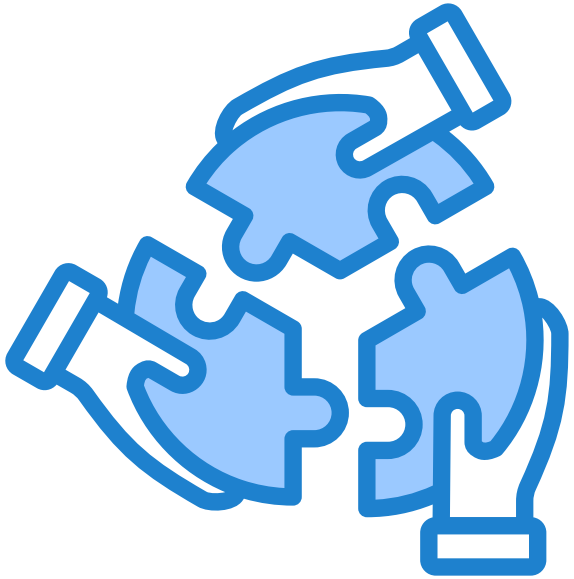
Bulgaria

adapting / developing educational videos and tools for medical students

Thank you!

Questions?





Collaboration with JARDIN
WP8

JARDIN

Joint Action on Integration of ERNs into National Healthcare Systems

Introduction to Work Package 8 and Future Cross-Project Collaborative Strategies

Co-funded by the European Union

“Co-funded by the European Union. Views and opinions expressed are however those of the author(s) only and do not necessarily reflect those of the European Union or European Health and Digital Executive Agency. Neither the European Union nor the granting authority can be held responsible for them.”

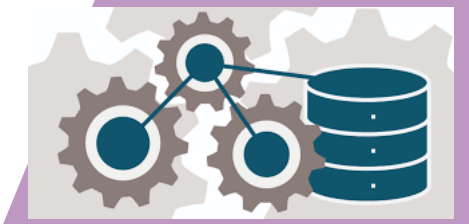
The Role of Data in RD Care

- Data has both primary and secondary uses in patient care
  - Secondary uses include policymaking and research

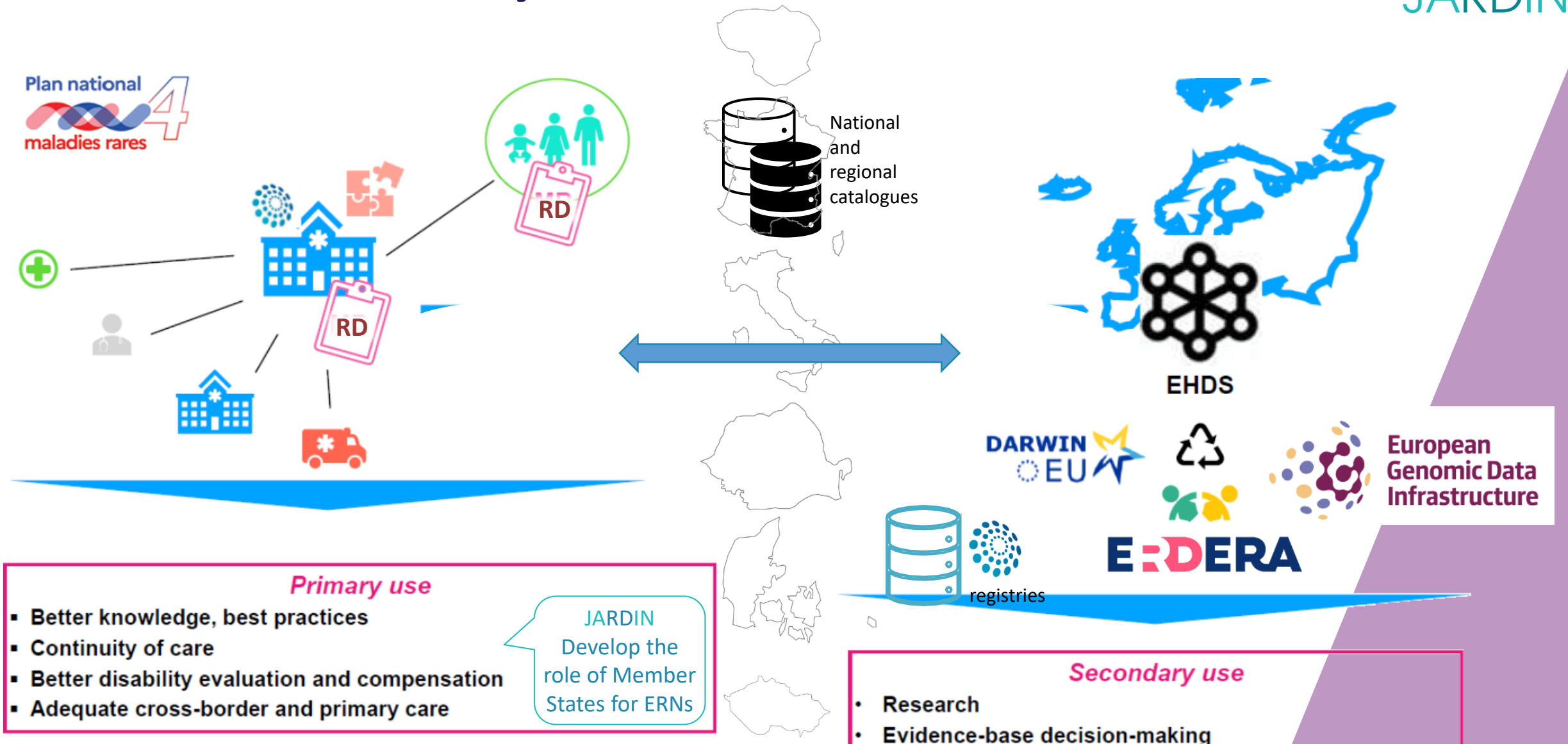
- RD data and expertise is complex, heterogenous, and scarce
  - Therefore insufficient to achieve these objectives

- Strategies such as data cumulation, collaboration, and sharing across HCPs, registries, and countries are thus indispensable

- To achieve this, data must be FAIR to improve semantic accuracy and interoperability
  - Will necessitate improvements in technical, legal, organisational, and political frameworks



RD in the Data Ecosystem: What is at Stake?



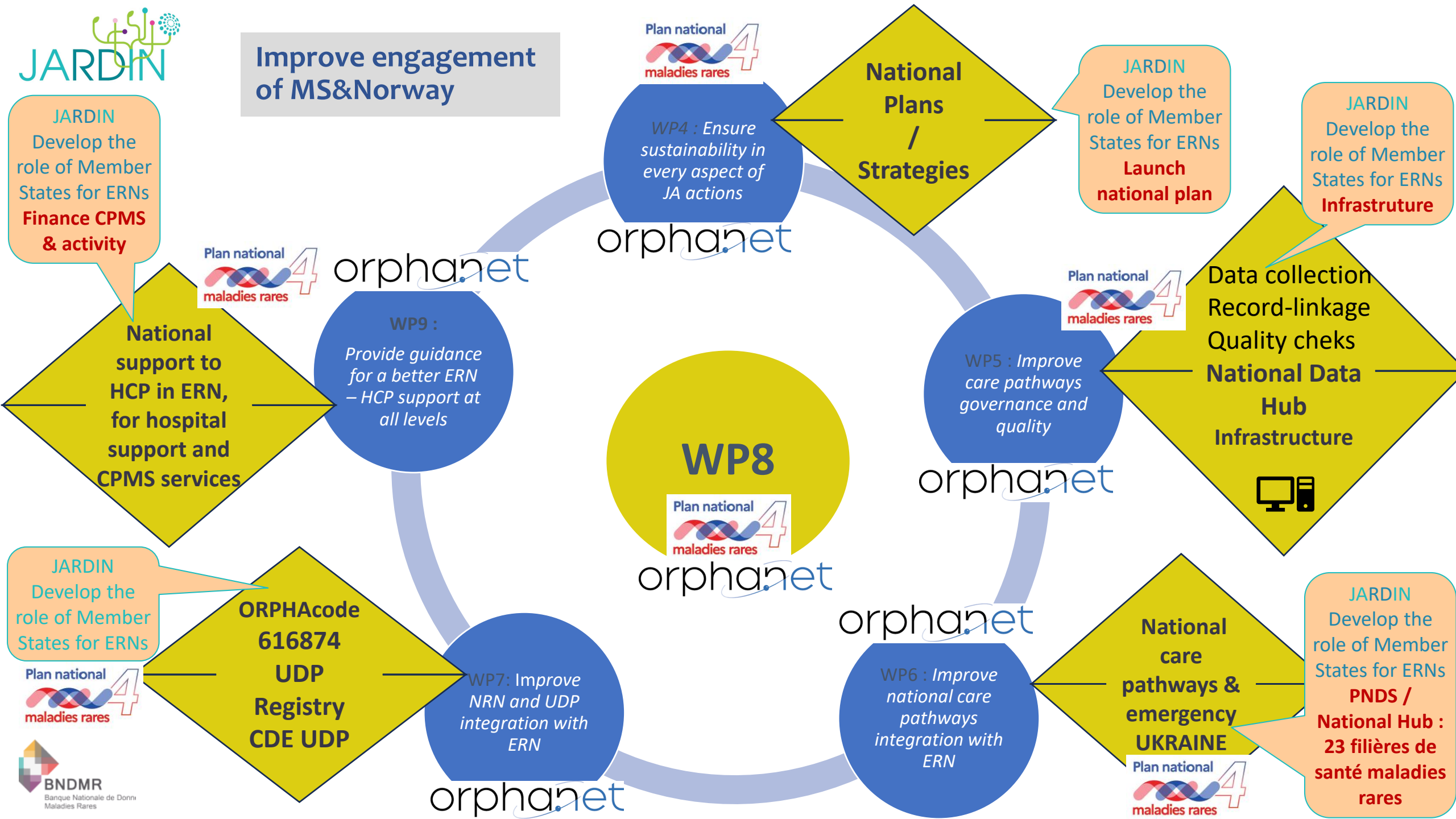
Primary use

- Better knowledge, best practices
- Continuity of care
- Better disability evaluation and compensation
- Adequate cross-border and primary care

JARDIN
Develop the role of Member States for ERNs

Secondary use

- Research
- Evidence-base decision-making


JARDIN
Improve engagement of MS&Norway
WP4 : Ensure sustainability in every aspect of JA actions
National Plans / Strategies
JARDIN Develop the role of Member States for ERNs Launch national plan
JARDIN Develop the role of Member States for ERNs Infrastruture
Data collection
Record-linkage
Quality cheks
National Data Hub
Infrastructure
WP5 : Improve care pathways governance and quality
WP6 : Improve national care pathways integration with ERN
National care pathways & emergency UKRAINE
JARDIN Develop the role of Member States for ERNs PNDS / National Hub : 23 filières de santé maladies rares
WP7: Improve NRN and UDP integration with ERN
ORPHAcode 616874 UDP Registry CDE UDP
JARDIN Develop the role of Member States for ERNs
WP9 : Provide guidance for a better ERN – HCP support at all levels
National support to HCP in ERN, for hospital support and CPMS services
JARDIN Develop the role of Member States for ERNs Finance CPMS & activity
WP8
Plan national maladies rares
orphanet
BNDMR
Banque Nationale de Donn
Maladies Rares

A short introduction to JARDIN and the ERNs

- The EU has long recognised that the scattered distribution of RD patients renders the acquisition of knowledge and expertise difficult

- The 24 European Reference Networks were established in 2017
  - Designed to:
    - Improve patient access to high quality care
    - Promote research
    - Advance clinical training and therapies
    - Inform healthcare policy

JARDIN Joint Action

- Joint Action of the Integration of ERNs into National Healthcare Systems

- Running in parallel to other synergistic initiatives
  - European Rare Disease Research Alliance (ERDERA): RD research
  - European Rare Disease Research Coordination and Support Action (ERICA): ERNs research
  - Orphanet Data for Rare Disease Projects (OD4RD/OD4RD2): RD codification and data
  - European Health Data Space (EHDS) and related projects (TEHDAS, Xt-EHR)

- ERNs and RD data are at present fragmented and unintegrated into national health systems

JARDIN Joint Action

'To improve the accessibility and support the long-term sustainability of the ERN system by contributing to the effective integration of ERNs in the national health systems (while always respecting the autonomy of MS in this regard), thereby, on the other hand, also strengthening the resilience of the national health systems'

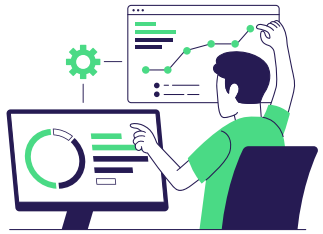
- JARDIN WP8 looks to bridge this gap through technical and semantic operability via consistent legal, operational and governance frameworks

JARDIN Survey

- Conducted to better understand the barriers that exist in the journey of RD health data as well as what solutions exist to overcome said barriers

- Addressed to three groups of targeted respondents
  - HCP Unit Leads: 457 responses from 27 countries
  - IT Support: 100 responses from 24 countries
  - National Authority IT Experts: 28 responses from 23 countries

- Covered topics such as the first capture of data, data sharing, and data FAIRness



Focus on ORPHAcoding – Current State of Play

- **ORPHAcodes implementation in hospital EHRs**
  - MS coverage remains incomplete
- **ORPHAcodes implementation in RD registries**
  - Complete in most of the MS
- **Huge heterogeneity in ORPHAcodes implementation**
  - Subset of ORPHAcodes only
  - Inpatients or outpatients only
  - Lack of portability of ORPHAcodes
  - Secondary ORPHAcoding from hospital to registry

Semantic, Technical, & Infrastructural

Barriers

- Lack of digital infrastructure
- Non-standardised datasets
- Limited interoperability between local, national, and European systems
- A lack of automation, leading to labour-intensive processes
- Limited integration of ORPHAcodes

Solutions

- Common or interoperable standardised EHR formats at the national level that allow for the consistent use of ORPHAcodes and other coding standards (e.g. EEHRxF, HL7 FHIR)
- Implementation of FAIR Principles and use of semantic terminologies and tools (e.g., the ORPHAcodes) to harmonise coding
- Use of standardised minimum datasets to simplify data collection

Legal & Governance

Barriers

- Absence of binding national policies enforcing ORPHAcode usage
- Fragmented legal frameworks for data sharing and reuse
- Complexity of obtaining consent and approvals for secondary use

Solutions

- National legislation mandating ORPHAcodes in (at least) designated centres or hospitals
- Clear and centralised governance frameworks for RD data protection and sharing
- Central ethics approval for reuse of pseudonymised data
- Adoption of opt-out models
- Harmonised Consent Frameworks

Organisational & Resource-Based

Barriers

- Limited human or technical resources for registry participation
- Administrative delays in data sharing agreements

Solutions

- Establishment of central coordinating support units
- Engagement in national or regional initiatives to support structured RD data capture and international interoperability
- Capacity-building Initiatives
- Enhanced Data Governance and Policy Support

Minimum Datasets

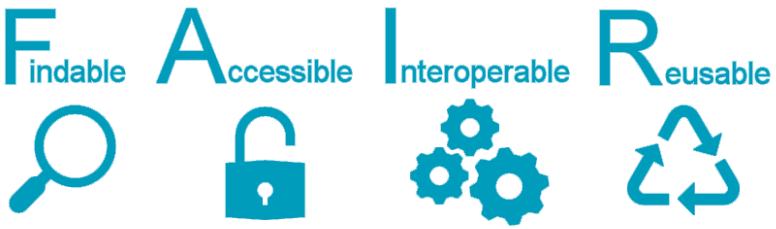
- Inconsistent and non-standardised RD coding and data capture make it difficult to identify RD patients in hospital systems
  - Negatively impacts RD patients and healthcare systems

- Conversely, harmonised standards confer many benefits
  - Improvement of patient care
  - Clinical research
  - Epidemiological insights
  - Resource allocation
  - Public health policy

Minimum Datasets

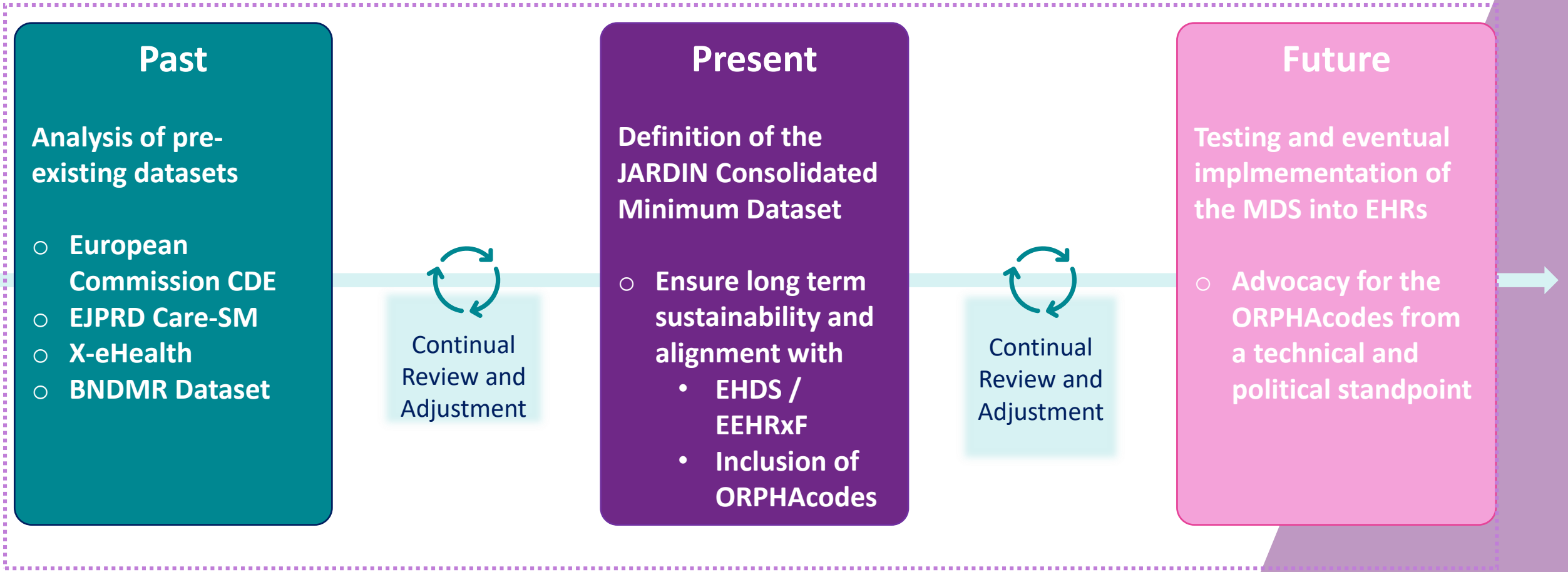
- A minimum dataset (MDS) defines a consistent list of data elements to be collected for all RD patients
  - Ensures homogeneity across HCPs, countries, registries and databanks

- MDS render collected data interoperable and reusable (FAIR) for both primary and secondary data use
  - By using known and accepted standards, data can be shared, reused and understood across EHRs, software and registries



JARDIN Chronology

Flow Chart of JARDIN WP8 Methodology



JARDIN Subtask 8.4.3

Use of implemented RD datasets for monitoring HCP and ERN activities

Capacity Building
Coders ORPHAcoding Training

Data Management
Workshop T8.2

Capacity Building:
Data Exploitation Workshop

Y1

Technical implementation of the ORPHAcodes into EHR

Aarhus University Hospital
Inserm

Y2

Active coding of RD diagnoses

Helpdesk provided by Inserm

AUH (DK)
Inserm (FR)
TUH (EE)
Sciensano (BE)
Veneto region (IT)

Y3

Production of RD releated KPIs at ERN and HCP levels

AUH (DK)
Inserm (FR)
TUH (EE)
Sciensano (BE)
Veneto region (IT)

Demonstrate the added value and use-cases of the ORPHAcodes*

** i.e. increase in the number of RD identified (OC vs ICD10) ; comparing LOS in RD inpatients vs non-RD patients*

Data Management Workshop

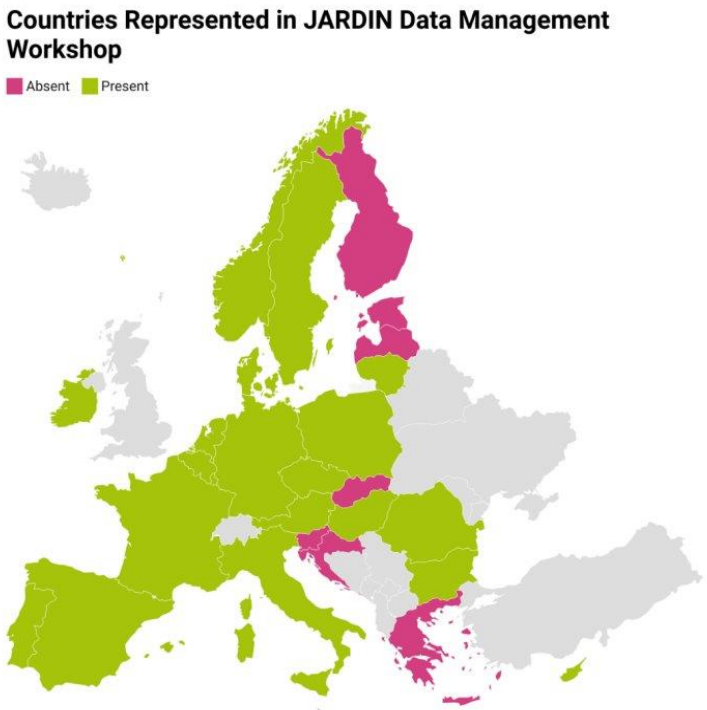
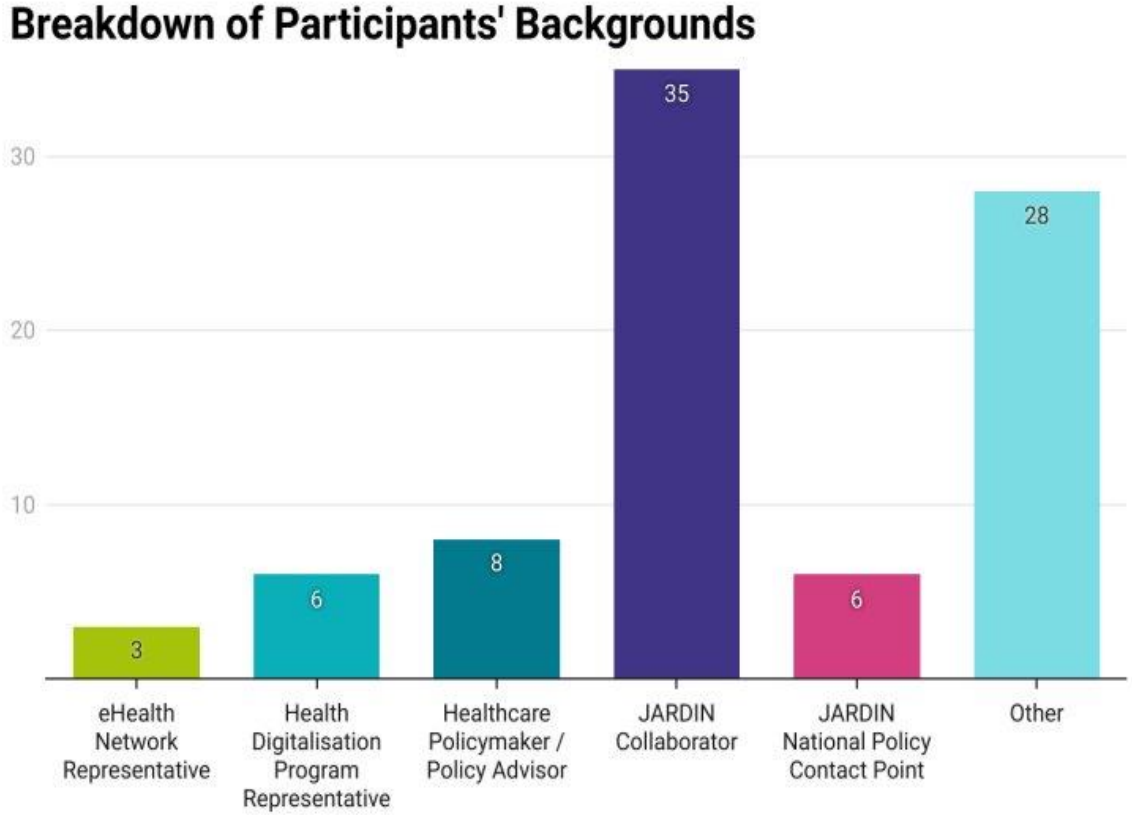


Figure 1 Countries Represented in the JARIDN Data Management Workshop: The map indicates the countries who were present at (green), or absent from (magenta), the JARDIN Data Management Workshop. Countries not implicated in the workshop are in silver.



Data Management Workshop - Key Messages

- Promote **policies** commanding the capture, sharing and re-use of RD data at national level
- **Incentivise** through benefits (for HCPs, for health professionals, for patients) and education
- **Finance** the data capture, harmonization and technical/legal support
- Enforce and empower HCPs IT departments to implement **ORPHAcodes and MDS**, and use **IT implementable solutions and guidelines (automation)**
- Provide **capacity building** tailored for IT, health professionals, coders, managers
- Facilitate **helpdesks/helplines** to support them

ORPHAcode Implementation Policy Guidance

- Will require consideration of how the ORPHAcodes are utilised at the policy level in addition to the HCP level.
  - There are numerous use-cases for coding with the ORPHAcodes

- Therefore, a better understanding coding practices is important as these practices will have policy implications.
  - This may require further research or a survey of the member states to gain a better understanding of their ORPHAcoding practices.
  - General recommendations that can be adopted at the national level would be the ultimate goal.

Opportunities to Collaborate: JARDIN / OD4RD2

- JARDIN to provide the framework and guidelines for ORPHAcode implementation from a political standpoint
  - Can further advocate for this through our final workshop and documentation

- OD4RD2 can provide the groundwork through capacity building, practical assistance, and the empowerment of coders and IT personnel

THANK YOU!

Orphanet Data for Rare Diseases 3

Proposal

OD4RD3

Orphanet Data For Rare Diseases

Contract information

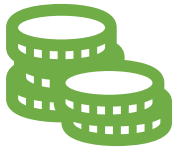
Direct grant

- No competition
- Orphanet *de facto* monopoly

Mono-beneficiary (INSERM) + 23 affiliated entities(+4: GR; IS; DK; *LU*)

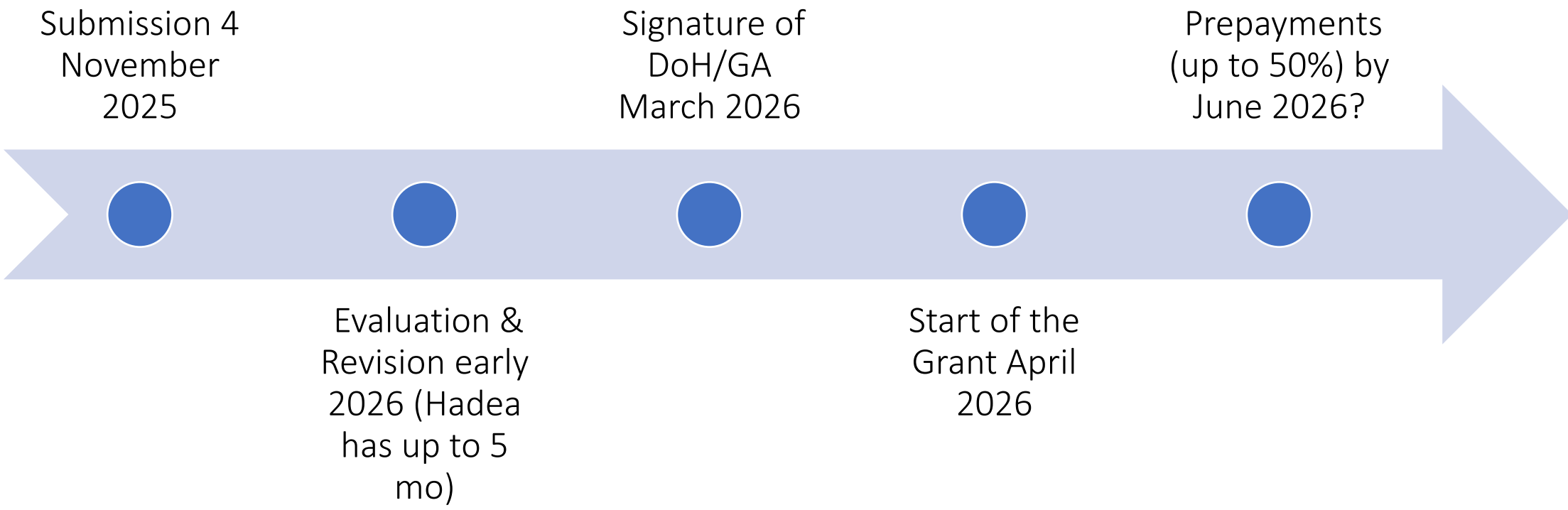
33 months

- March 2026 – Dec 2028

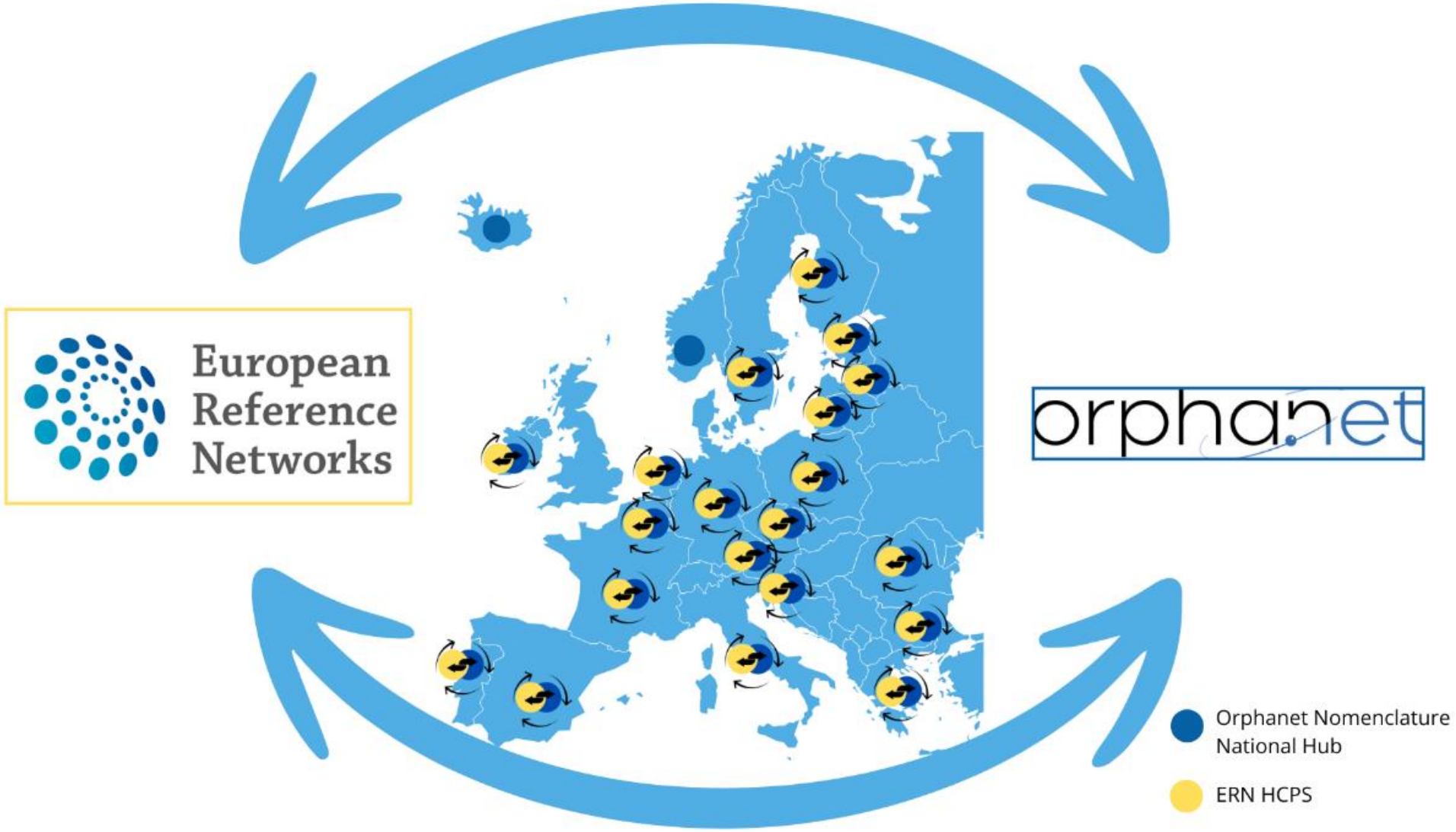


Estimated call budget 3.5 M€

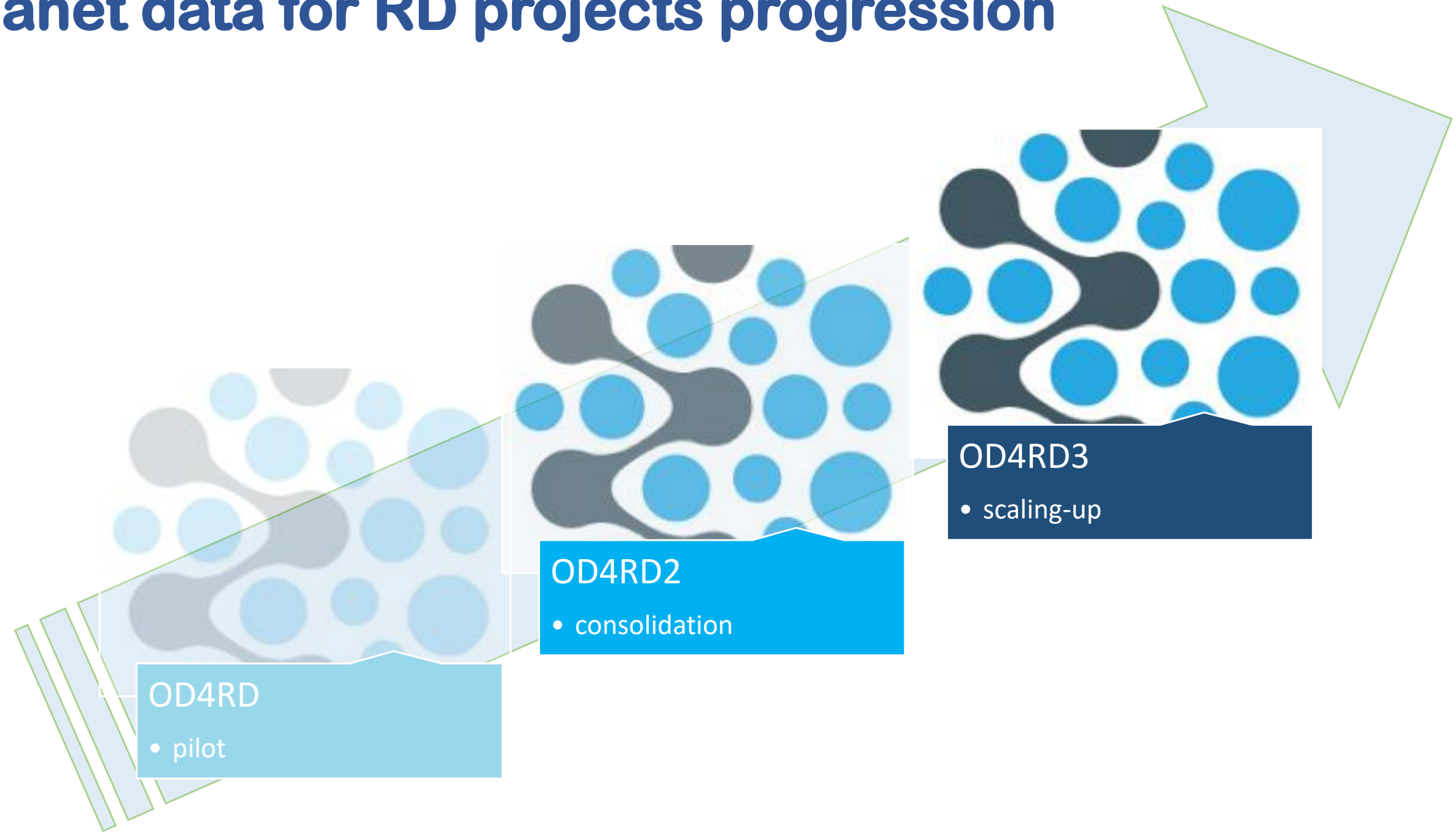
Expected Timeline



Main principle



Orphanet data for RD projects progression



Challenge for RD Data	Description	Needs	Solutions	Foreseen actions
Rarity and invisibility; insufficient representation of RD in medical terminologies	There are 6500+ RD and 85% touch less than 1 person in 1 M. This implies that no country or system can alone generate a critical mass data necessary to build knowledge + no generic terminology can as of today specifically represent EACH and ALL RDs, except ORPHAcodes.	To generate a critical mass of the data all countries must collect RD data in a standardized way using a specific terminology	ORPHAcodes implemented in medical records in Hospitals, coding at the bedside. Clinicians should be supported and coding facilitated for data accuracy and quality IT departments should be supported so as ORPHAcodes are effectively implemented	Continuous production of specific RD Nomenclature in collaboration with ERNs (WP2) Delivery of a ready-to-use ORPHAcodes files and tools to ease implementation (WP2) Support and training to coders and IT by national Hubs (with umbrella coordination) (WP3)
Rapid evolutivity of the field, and of the knowledge especially in genetics	New diseases are described, already known diseases are better characterised and either regrouped or splited. Knowledge of each RD evolves (etiology and physiopathology, natural history, management and therapies) ERNs hold the clinical expertise, increase the knowledge on RD through their registries and provide clinical practice guidelines and standards of care	To have an up-to date nomenclature, textual information and scientific data on each RD available for consultation and re-use for free to all the stakeholders	Update the Orphanet database to follow up the evolution of knowledge in collaboration with ERNs Develop facilities to capture experts/users' feedback	Production of specific RD Nomenclature in collaboration with ERNs (WP2) Production of genetic annotations of RD (WP4) Production of updated information on RD (WP4)
RD touch every specialty and the border between care 1ary use) and research and policy-making (2dary use) is very faint	Good quality data produced at the point of care is necessary for improved care as well as research & for Indicators production , allowing for evidence-based decision making in health systems. However, in most countries it is impossible to answer the most basic questions such as how many RD patients we have in our health system.	To ensure interoperability between these different domains. To demonstrate the added-value of good quality data for healthcare provision and planning.	Accurate alignments between terminologies are available and delivered in computable formats. Support for data exploitation at local level (HCPs, registries, etc) by using the Orphanet RD classification system.	Production of specific RD Nomenclature in collaboration with ERNs (WP2) Quality controlled and inter-terminologies consistent alignments (WP2) Developments of data stewardship programme (WP3) Easy to use tools (WP2-3)
Need for evidence to support policy actions in the ERNs sphere	ERNs are aimed to bring coordinated expertise and standard of care for all RD including rare cancers across MS. No systematic analysis on this coverage is available. It is difficult nowadays to design patient care pathways by RD within and among ERNs.	To have up-to-date quantitative and qualitative reports on coverage of all RD by ERNs to inform future actions within and between ERNs To have a clear picture of the geographical distribution of ERN expertise across MS.	Analyse RD field coverage using the Orphanet classification, analyse qualitative coverage by co-analysing Orphanet and ERNs' data, inclusing the Orphanet database of expert centers.	Production of reports on ERN RD coverage (WP4) Identification of gap expertise at national and EU level (WP4) Specify and provide a user-friendly way to capture the updated ERN coverage of RD

General Objectives

1.Ensure the production and delivery of the **Orphanet nomenclature of RD** (ORPHAcodes) in a way it is updated and adapted to evolution of knowledge and coding needs including the need for interoperability of RD data in health and research systems.

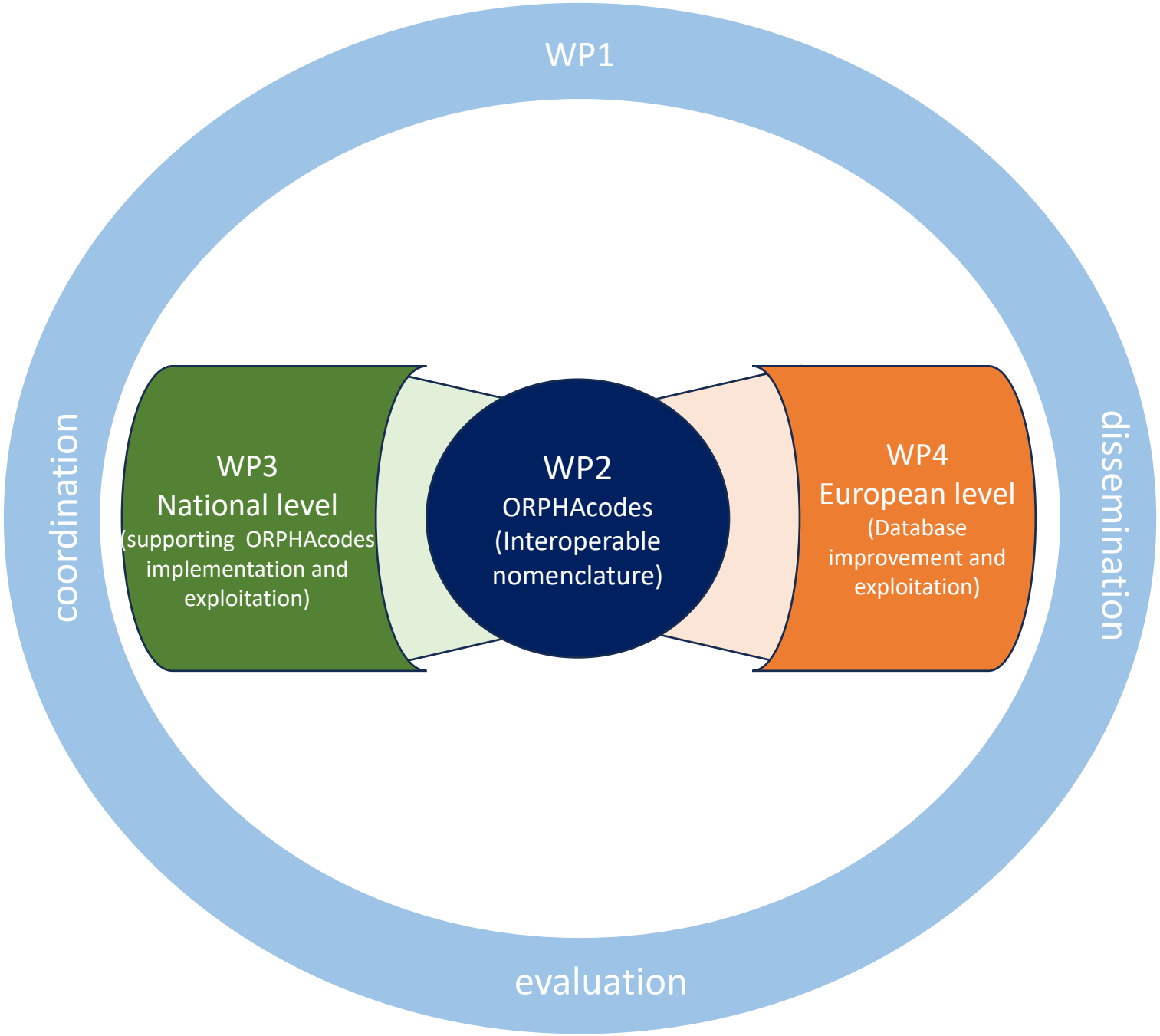
2.Accompany and promote the **RD codification expansion** in ALL European MS, by providing human and technical support for implementation, actual adoption and codification in a harmonised standardized way across Europe

3.Collaborate closely with ERNs so their expertise and data collected in registries contributes to **knowledge generation and dissemination** through a constantly improved Orphanet knowledge base.

4.Accompany the **RD data exploitation** at scale, for primary and secondary use (including monitoring activities) by disseminating best practices, tools and services, and making use of the Orphanet reference data produced in collaboration with ERNs.

Specific Objectives

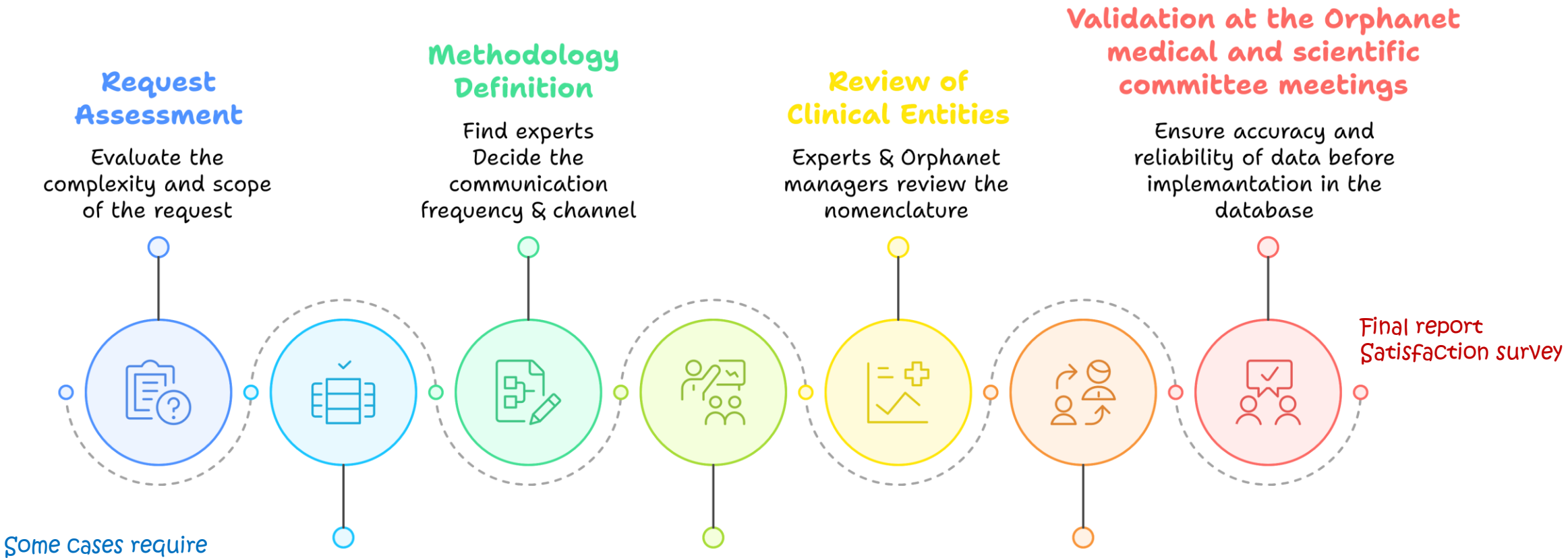
1. Ensure the continuous production and delivery of Orphanet **nomenclature** so as to follow the continuous evolution of knowledge and to adapt to coding needs, in particular by enhancing collaborations with ERNs
2. Ensure the **interoperability** between ORPHAcodes and other terminologies in use (in particular ICD-10, ICD-11 and SNOMED-CT) so as to provide an accurate and consistent resource for transcoding.
3. **Scale-up** the support for ORPHAcodes implementation in MS by expanding the capacity and the geographical coverage of **the Network of Orphanet Nomenclature Hubs (NONH),** so as to cover all EU MS, and Ukraine by adapting the National hubs action plans to the local situations
4. Empower the NONH to provide **support for data exploitation** using the Orphanet open-science knowledge base for the production of monitoring indicators and/or to respond to research questions through the creation of a data stewardship programme
5. Expand and update the **Orphanet knowledge base** content in collaboration with ERNs so as to contribute to its exploitation for primary use (improved patients' healthcare pathways) and secondary use (data exploitation)
6. Contribute to the European RD policy by providing **support** to the European Commission, the Board of Member States and the ERN coordination **for evidence-based decision-making**.


WP1
coordination
dissemination
evaluation
WP3
National level
(supporting ORPHAcodes implementation and exploitation)
WP2
ORPHAcodes
(Interoperable nomenclature)
WP4
European level
(Database improvement and exploitation)

OD4RD3 structure and activities

- **WP1. Project management and coordination (Inserm)**
  - Coordination
  - Dissemination
  - Evaluation
- **WP2. Orphanet interoperable nomenclature and classification of RD update and maintenance**
  - Production & release of the nomenclature; Collaboration with ERNs (Inserm)
  - Maintenance and quality control of ORPHAcodes alignments with ICD and SNOMED) (Inserm & BfArM)
- **Work Package 3: Supporting ORPHAcodes implementation & exploitation at the National Level**
  - Scientific and organisational coordination to scale up (Inserm & OUS)
    - **Support to implementation and coding + good coding practices**
    - **Support to data exploitation using Orphanet classification & knowledge base**
  - National hubs action plans (all)
- **WP4. Database improvement and exploitation at the European level (Inserm)**
  - Expand and improve RD information (genes and texts) in collaboration with ERNs
  - Specifify and (co)develop services for the EC and ERN coordination
  - Update and expand the RD coverage analysis for better patient pathways

WP1. Orphanet nomenclature
Orphanet and ERN collaboration steps



https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/eproc_disease_inventory_R1_Nom_Dis_EP_05.pdf

ERN collaboration procedural document

Orphanet and ERN collaboration projects

ERN	Group Revised/ to be revised		Collaboration main goal	Status
ITHACA-eUROGEN / Spina Bifida and other dysraphisms	ORPHA: 268357	Neural tube closure defect	Classification revision and entities inclusion	Finalized
MetabERN	ORPHA:738	Porphyria	Entities inclusion	
ERN-EYE	ORPHA:499047	Isolated optic neuritis	Classification revision and entities inclusion	
VASCERN	ORPHA:211237	Rare vascular tumor	Classification revision and entities inclusion	
ERN-EuroBloodNet	NA	Pediatric thrombotic diseases	Entities inclusion	
ERN SKIN	ORPHA:79373	Ectodermal dysplasia syndrome	Entities inclusion	
ERN GENTURIS	ORPHA:104010	Intestinal polyposis syndrome	Entities inclusion	
ERN EuroBloodNet	ORPHA:68364	Hemoglobinopathies	Classification revision and entities inclusion	
ERN EURO-NMD	ORPHA:599	Distal myopathy	Classification revision and entities inclusion	
ERN EURO-NMD	ORPHA:98482	Idiopathic inflammatory myopathy	Classification revision and entities inclusion	
ERN-Transplantchild	ORPHA:506210	Rare disorder potentially indicated for liver transplant	Classification revision	
ERN-Transplantchild	ORPHA:506222	Rare disorder potentially indicated for lung transplant	Classification revision	
ERN-Transplantchild	ORPHA:506225	Rare disorder potentially indicated for heart transplant	Classification revision	
ERN-Transplantchild	ORPHA:506213	Rare disorder potentially indicated for kidney transplant	Classification revision	
ERN-EYE	ORPHA:519311	Rare disorder of the posterior segment of the eye	Classification revision and entities inclusion	
ERN RND	ORPHA:282	Frontotemporal dementia	Classification revision and entities inclusion	
ERN EpiCARE	ORPHA:101998	Rare epilepsy, step 1: Epilepsy syndromes, ORPHA:166463	Classification revision and entities inclusion	Finalized before 2026
ERN VASCERN	ORPHA:211237	Rare vascular malformation: simple	Classification revision and entities inclusion	
ERN VASCERN	ORPHA:211237	Rare vascular malformation: complex	Classification revision and entities inclusion	
ERN PaedCan/SIOPE	NA	Pediatric cancer - high grade glioma - CNS	Creation of the classification	
ERN EURO-NMD	ORPHA:98491	Neuromuscular Junction disease	Classification revision and entities inclusion	
ERN ERNICA	ORPHA :104012	Rare Inflammatory Bowel Disease	Classification revision and entities inclusion	

Orphanet and ERN collaboration projects

ERN	Group Revised/ to be revised		Collaboration main goal	Status
ERN GUARD-Heart	ORPHA:218436	Rare cardiac rhythm disease	Classification revision and entities inclusion	Ongoing
ERN RND	ORPHA:68356	Leukodystrophy	Classification revision and entities inclusion	
ERN RND	ORPHA:306719	Chorea	Classification revision and entities inclusion	
Inter ERN Mito-WG	ORPHA:68380	Mitochondrial disease	Classification	
ERN PaedCan/SIOPE	NA	Pediatric cancer - renal tumors	Creation of the classification	
ERN PaedCan/SIOPE	NA	Pediatric cancer - Very rare tumors	Creation of the classification	
ERN PaedCan/SIOPE	NA	Pediatric cancer - CNS tumors	Creation of the classification	
ERN RND	ORPHA:98006	Hereditary spastic paraplegia	Classification revision and entities inclusion	
MetabERN	ORPHA:289899	Organic aciduria	Classification revision and entities inclusion	
ERN EURO-NMD	ORPHA:97245	Congenital myopathy	Classification revision and entities inclusion	Pending
ERN-NMD	ORPHA:98006	Rare neurological diseases, step1: Hereditary spastic paraplegia	Classification revision and entities inclusion	
ERN-Transplantchild	ORPHA:506216	Rare disorder potentially indicated for bowel transplant	Classification revision	
ERN-Transplantchild	ORPHA:506219	Rare disorder potentially indicated for HSC transplant	Classification revision	
ERN-CRANIO	ORPHA:2014	Cleft palate	Classification revision and entities inclusion	
ERN SKIN	ORPHA:79354	Ichthyosis	Classification revision	Starting in early 2026
ERN SKIN	ORPHA:79357	Hereditary palmoplantar keratoderma	Classification revision	
ERN SKIN	ORPHA:79373	Ectodermal dysplasia syndrome	Classification revision	
ERN EuroBloodNet	ORPHA:182040	Rare aplastic anemia	Entities inclusion	
ERN EuroBloodNet	ORPHA:98363	Rare hemolytic anemia	Classification revision and entities inclusion	
ECLip & Lipodystrophy international experts	ORPHA:90970	Primary lipodystrophy	Classification revision and entities inclusion	Starting in late 2026

WP1. Interoperability of the Orphanet nomenclature

Alignments workflow: ensure interoperability between ORPHAcodes and other generic terminologies

Cardinality:

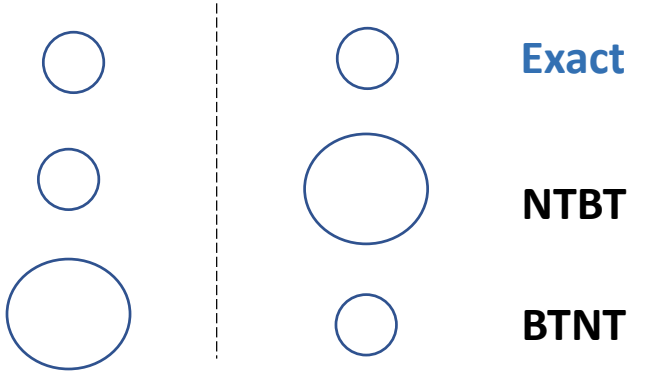
ORPHAcode → Target terminology

Except : ORPHAcode ↔ SNOMED-CT

Alignments are qualified by:

A proximity relationship

ORPHAcode → Targeted terminology



Exact

NTBT

BTNT

A specificity relationship (WHO terminologies)

Specific code

Inclusion term *(only ICD-10)*

Index term

Attributed code

Alignments workflow:
ensure interoperability between ORPHAcodes and other generic terminologies

SNOMED CT

- Reciprocal collaboration:
1. Orphanet produces the updates of RD nomenclature
2. Knowledge is transmitted to and evaluated by SNOMED-CT according to their own guidelines

- Only exact matches are aligned

SNOMED browser:
https://browser.ihtsdotools.org/?

OMIM
Human Genetics Knowledge for the World

- Survey of OMIM phenotypes (once a month)
- Orphanet genetic entities are aligned with phenotypic OMIM numbers
- Alignment is qualified by proximity relationship: Exact matches, NTBT or BTNT.

https://www.omim.org/

World Health Organization ICD-10 ICD-11

- Aim to cover all of the entities at the disorder level
- ICD-11 includes: MMS & foundation
- RDs are more represented in ICD-11
- Alignments qualified by:
  Proximity (Exact, NTBT, BTNT)
  Specificity (Specific, Index, Inclusion for ICD-10, Attributed)

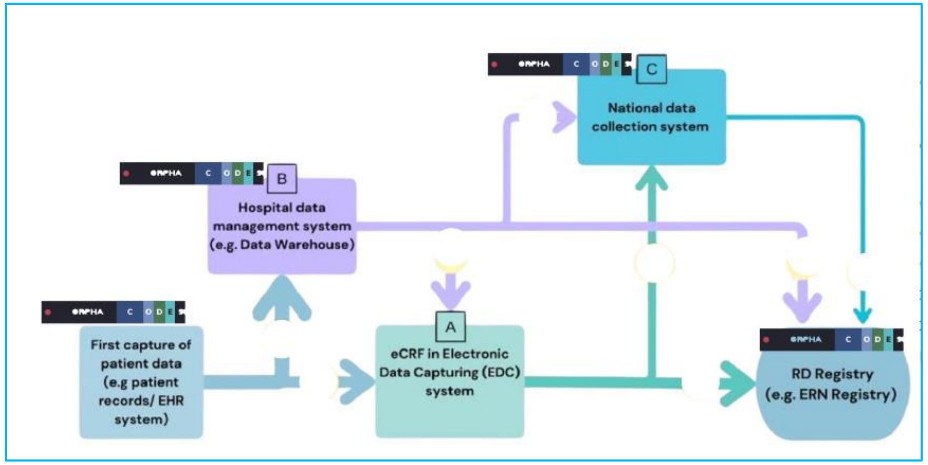
ICD-10: https://icd.who.int/browse10/2019/en
ICD-11 MMS: https://icd.who.int/browse/2025-01/mms/en

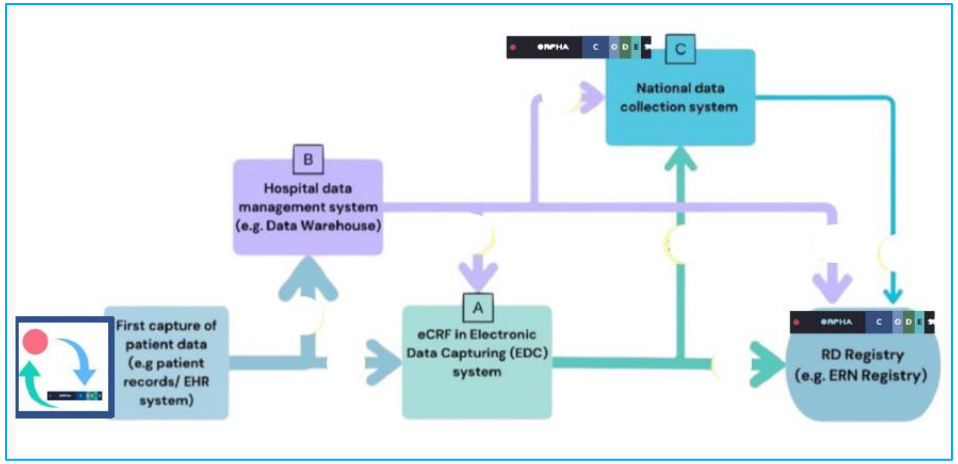
https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Orphanet_ICD10_coding_rules_R1_Nom_ICD_EP_07.pdf

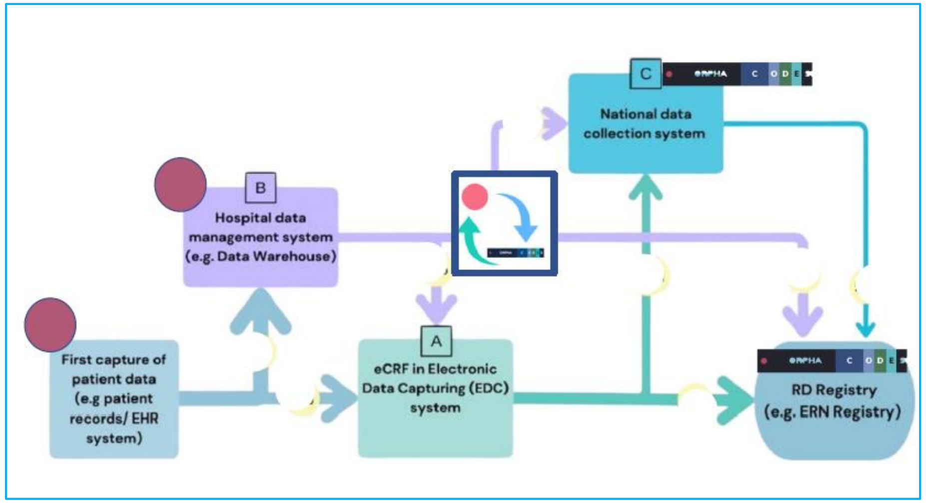
Alignment activity indicators

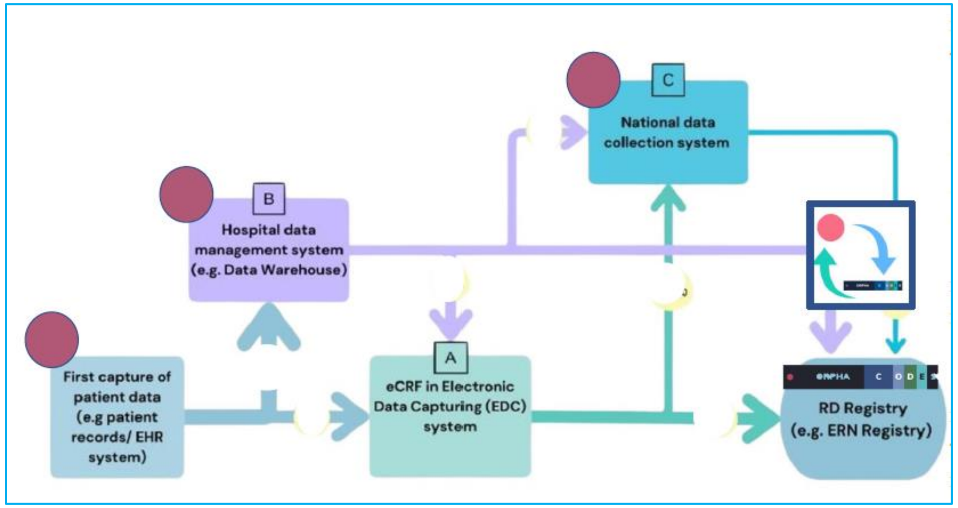
Indicator	Target reached (2022-2025)
% of disorder ORPHAcodes aligned with ICD-10, including inexact mappings	97.3%
% of disorder ORPHAcodes aligned with ICD-11 terms, including inexact mappings	71.7%
Number of ORPHA-SNOMED CT human-readable mapping files released	1/year, last release October 2025
Validation meetings with the scientific officer for ICD-10, ICD-11 and SNOMED-CT mappings	2/month
Survey of OMIM (phenotypes) updates	1/month

Inter-terminologies alignment: why it matters?

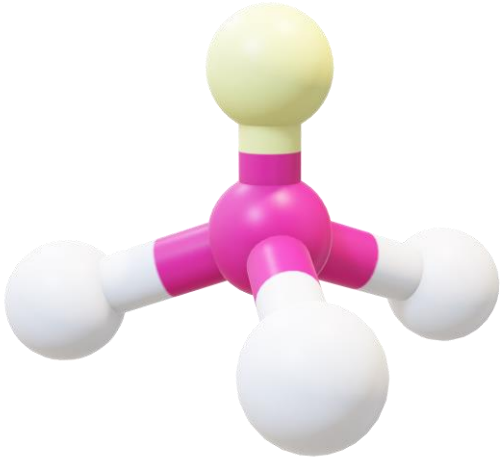








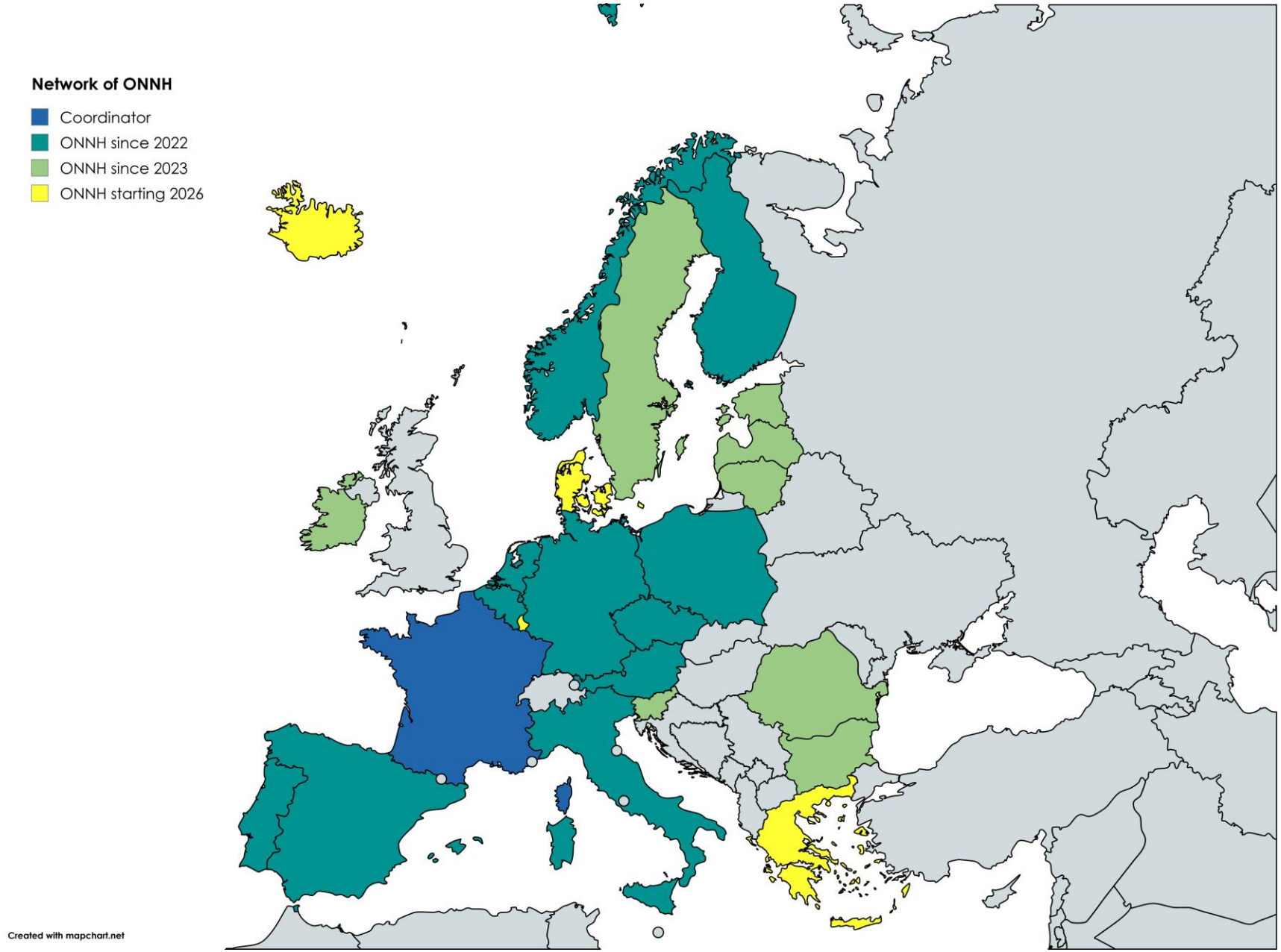
What's new in OD4RD3



- Cross-terminology alignment consistency
- By quality-controlling ORPHAcodes-to-n terminologies mappings
- Comparing to transcoding results from the ground (SNOMED CT mappings, Germany & other countries transcoding practices)

WP3. Supporting ORPHAcodes implementation & exploitation at the National Level

- Based on OD4RD2 and JARDIN lessons learned
- Increase the number of NOHN
  - Denmark
  - Greece
  - Iceland
  - Luxembourg
- Provide distant support to non-covered countries & Ukraine
- Increase the number of HCPs/country
- Provide support for data exploitation using the Orphanet classification and knowledge base
  - To produce indicators
  - To produce evidence for decision-making


Network of ONNH
Coordinator
ONNH since 2022
ONNH since 2023
ONNH starting 2026
Created with mapchart.net

Creation of a data stewardship programme for data exploitation support

- Year 1: forming
  - (constitution and capacity building of the coordinating team)
- Year 2: norming
  - (harmonizing practices based on first exploitation projects)
- Year 3: performing
  - (expanding the projects both within and across the borders to serve European Commission, MS, HCPs and ERNs aims)

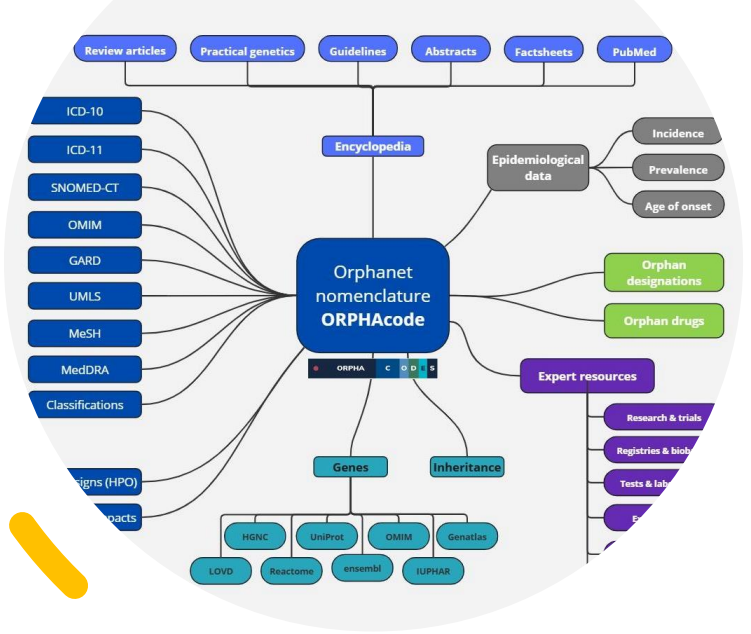
NOHN capacity building

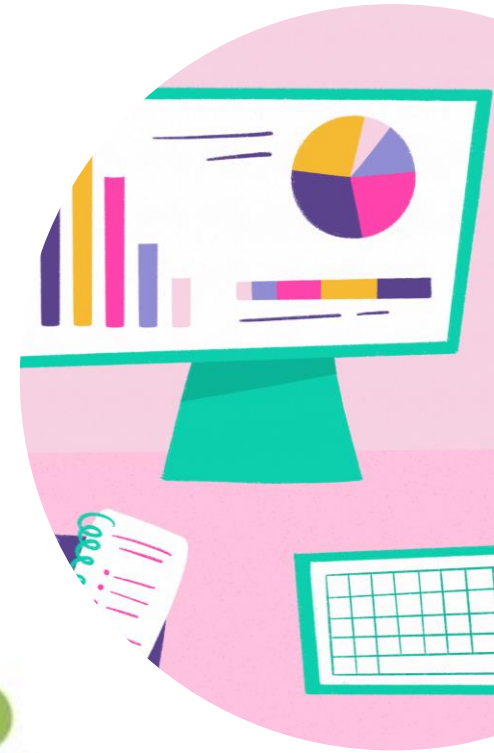
Provide support to make use of the Orphanet classification graph

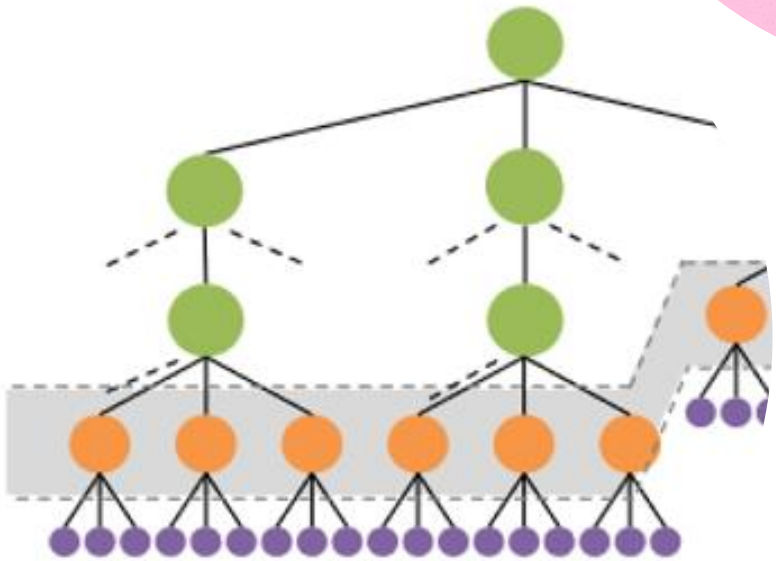
and the Orphanet knowledge base content organised around it

To analyse users' data in a meaningful way

- Indicators
- Using JARDIN lessons learned and use cases

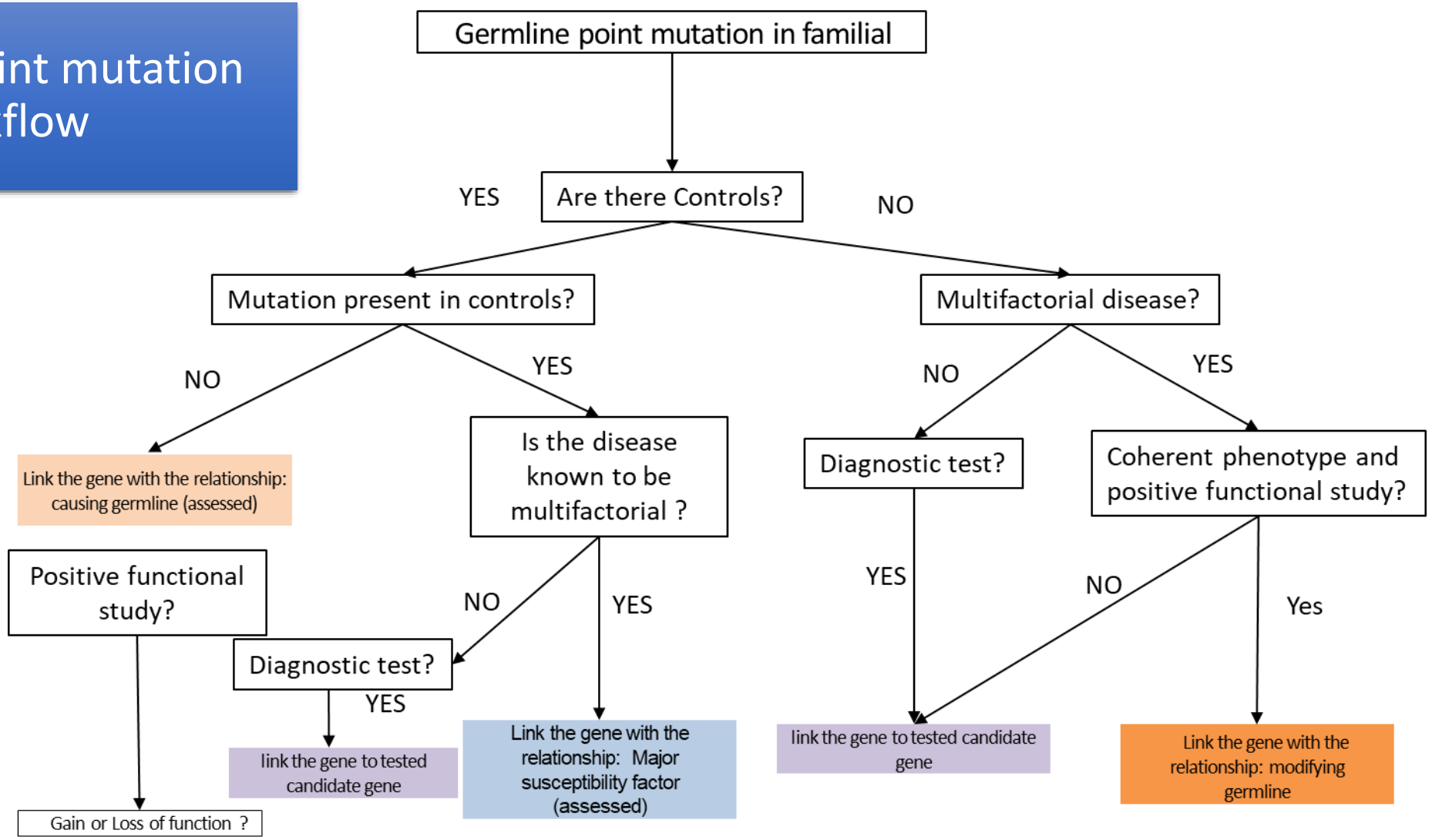






WP4. Orphanet content: gene-disease database

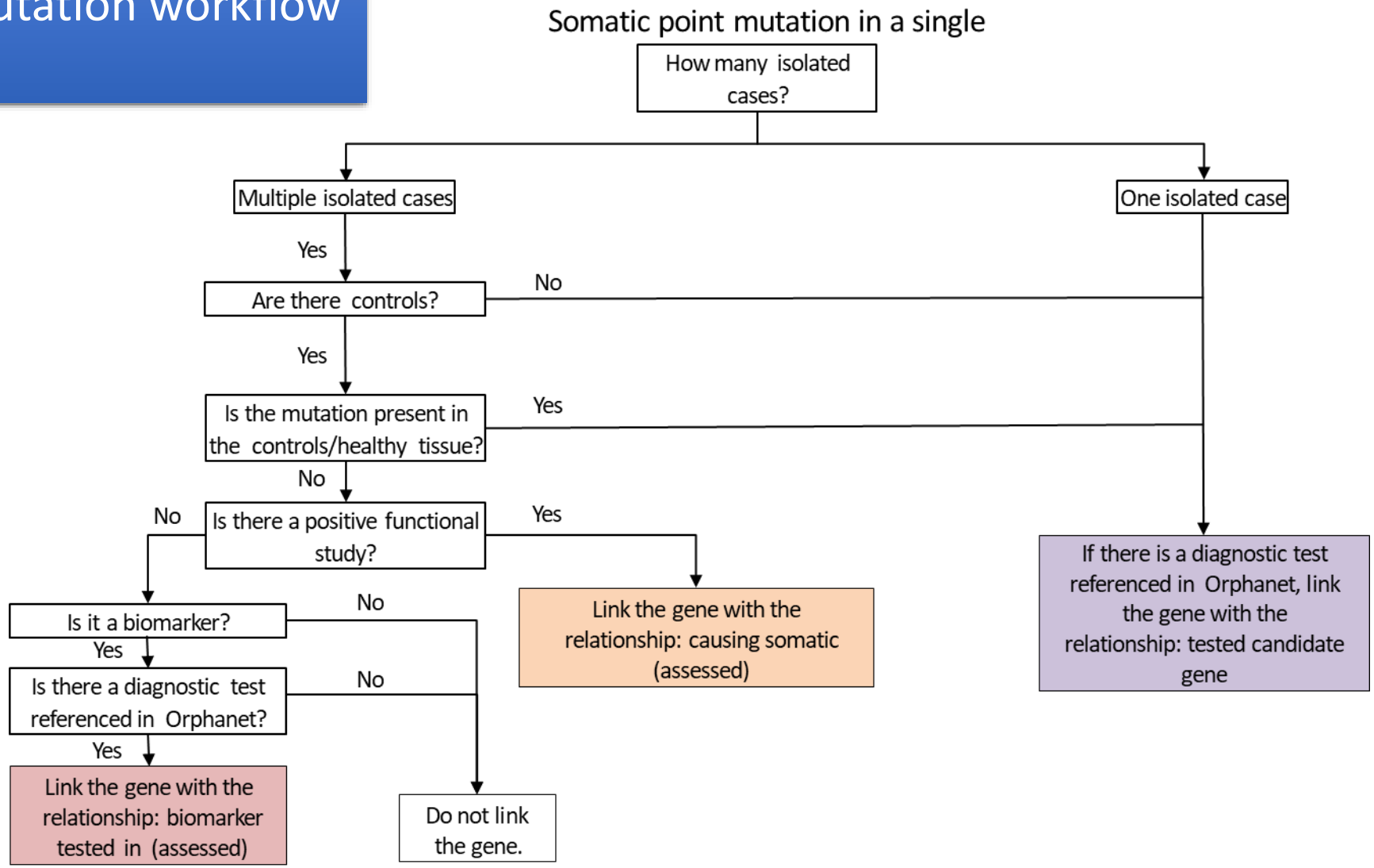
Germline point mutation workflow



https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Orphanet_Genes_inventory_R1_Ann_gen_EP_02.pdf

WP4. Orphanet content: gene-disease database

Somatic mutation workflow



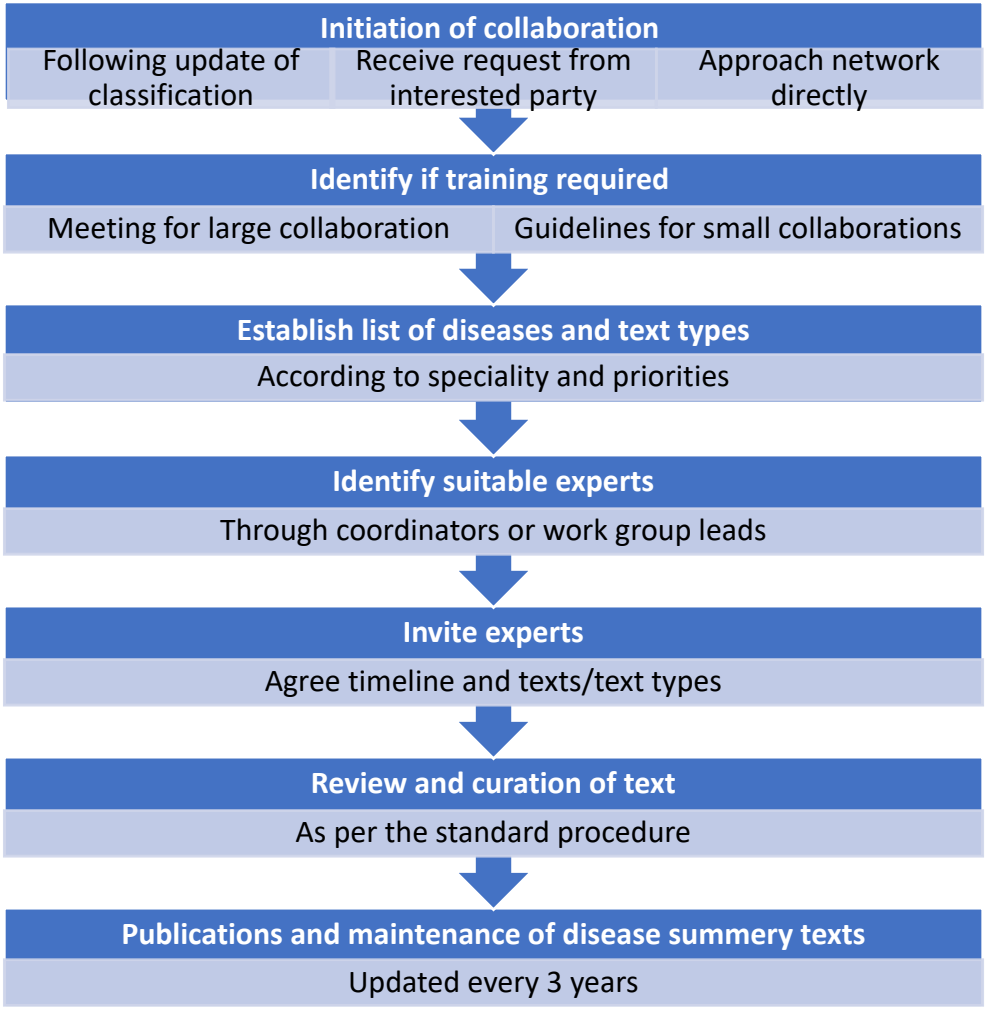
OD4RD/OD4RD2 gene database activity

- Since Feb 2022 to July 2025

Indicator	Number of corresponding associations
New genes or genes-disease associations	895
Modifications of gene-disease asssociations	61
Gene-disease associations suppressions	672
Total = new or updated gene-disease associations	**1628**

WP4. Orphanet content: definitions and abstracts

Workflow for collaboration with experts & expert networks



https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Orphanet_DiseaseSummaryText_R1_Prod_sum_EP_02.pdf

Current status of ERN-Orphanet professional encyclopedia collaboration

- Since Feb 2022 to July 2025

ERN	Number of text produced	ERN	Number of text produced
Endo-ERN	36	ERN-EuroBloodNet	28
EpiCARE	10	eUROGEN	3
ERKNet	13	ERN-EURO-NMD	19
ERN-CRANIO	18	ERN-ITHACA	43
ERN-RITA	7	MetabERN	36
ERN-BOND	4	VASCERN	11
ERN-EYE	3	**Texts co-signed by inter-ERN-collaboration**	
ERN-ERNICA	4	Endo-ERN & ERN-BOND	1
ERN-LUNG	2	eEUROGEN & ERN-ITHACA	4
ERN-RND	1	ERN-ITHACA & ERN-CRANIO	2
ERN-SKIN	25		
EURACAN	7	**TOTAL number of texts**	**277 (with 18 ERNs)**

WP4. Support the ERN governance

- Update the quantitative RD coverage analysis
- Perform a qualitative RD coverage analysis
  - Per country
  - To support patients' healthcare pathways understanding and design
  - To map further expertise needed to increase RD coverage
- Contribute to the ERN governance discussions and decisions
- To co-develop user-friendly tool for EC to keep RD coverage mapping up-to-date
  - Understand the needs
  - Specify the tool in collaboration with DG Santé IT colleagues
  - Implement data workflows

Lunch time

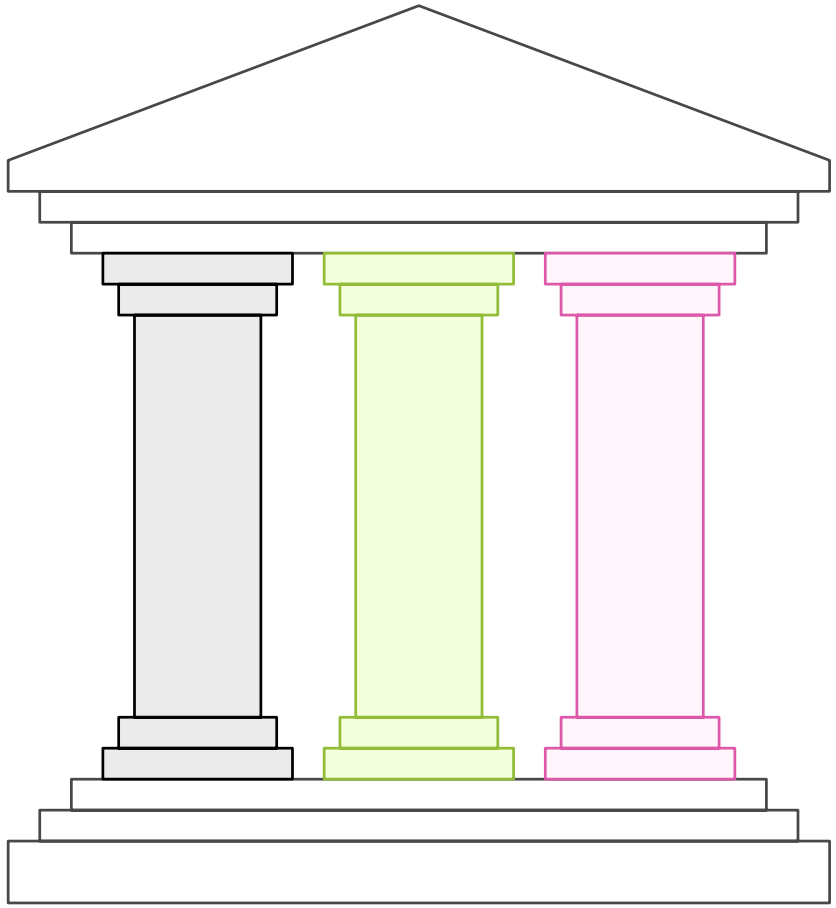
Thank you & please answer the survey ;)

OD4RD

Orphanet Data For Rare Diseases

Registries: a useful set of data

Registries: Pilar of the ERN needs



FAIR Principles

Ensures data quality and usability through findability, accessibility, interoperability, and reusability.

Common Data Elements

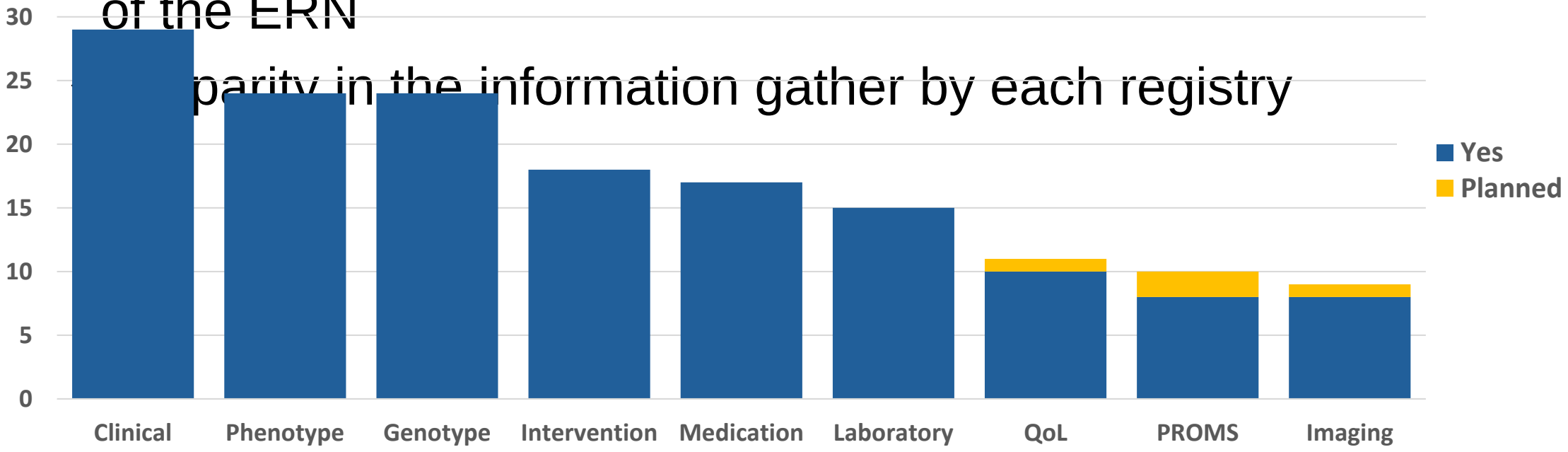
Facilitates interoperability by sharing standardized data elements across registries.

ERN-Specific Data Elements

Tailors data to specific clinical expertise areas, enhancing relevance and precision.

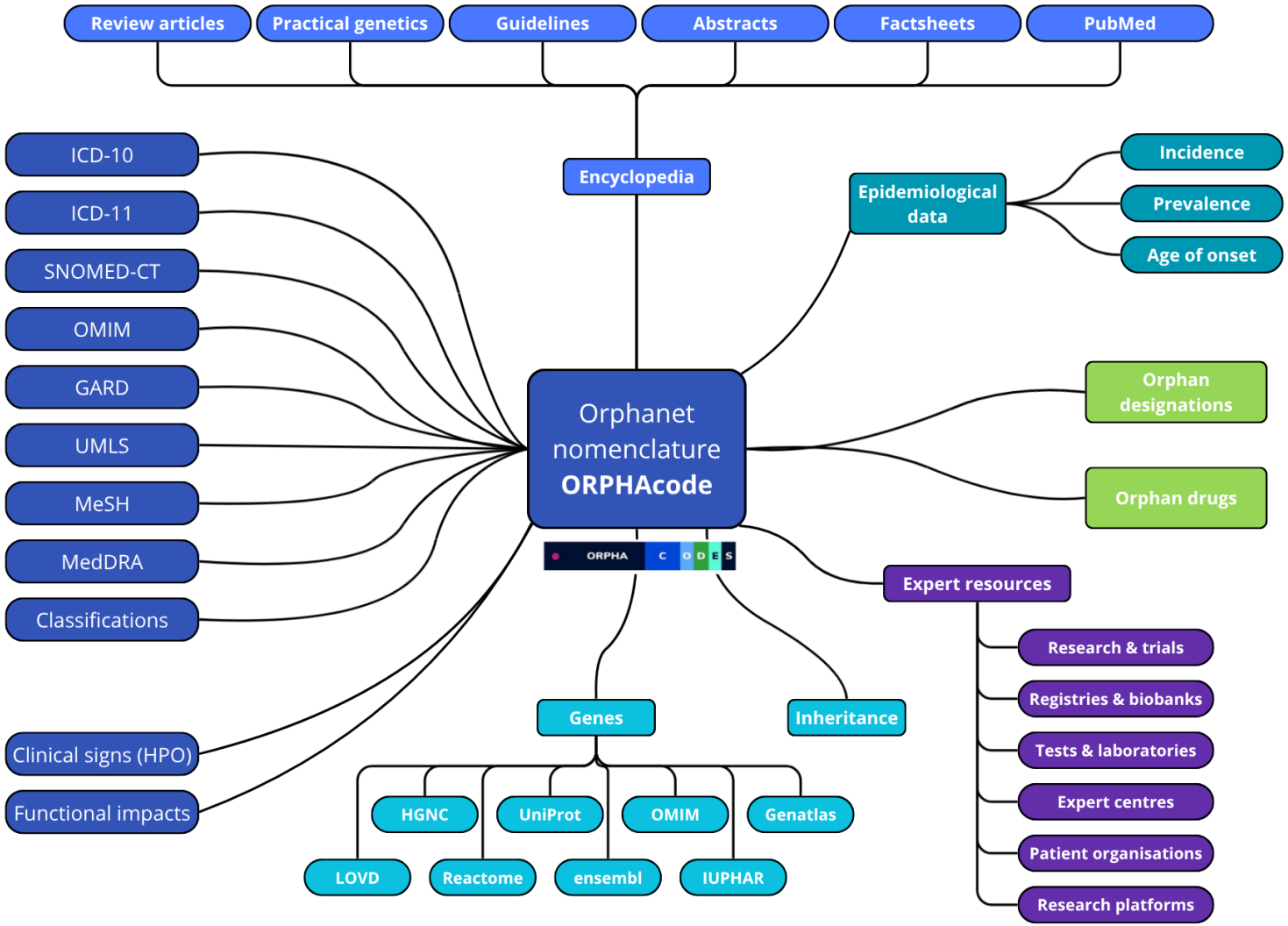
ERN Registries: State-of-the-art

- 29 registries for 24 ERNs with several ERNs with disease-specific registries
- Disparity in the number of centers participating depending of the ERN
- Disparity in the information gather by each registry



Modified from Jose Ramírez-García

Orphanet datasets



Round table

European Reference Networks

orphanet

Round table

European Reference Networks

orphanet

- What specific support or resources can Orphanet offer to improve coding practices and coding guidelines in the registries?
- What are your registries main challenges and needs related to the exploitation of Orphanet data?
- How can Orphanet help turn registry data into shareable knowledge about rare diseases?

Thank you for participating!

OD4RD2 FINAL MEETING

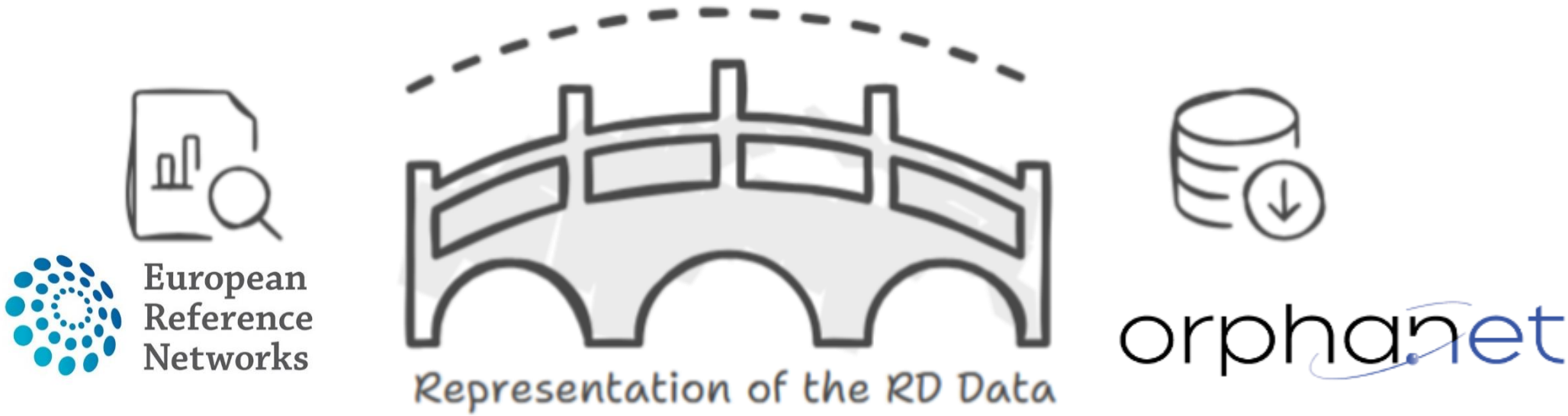
Scientific collaborations with the ERNs: nomenclature and scientific information

Paris, November 6th 2025

Animators: Marta FRUCTUOSO, Aysegul DILSIZOGLU

OD4RD
Orphanet Data For Rare Diseases

Orphanet and ERN collaboration improves high quality RD data availability



Scientific and clinical

- Clinicians' needs
- Accuracy of the data
- Coding coverage
- Updated terminology

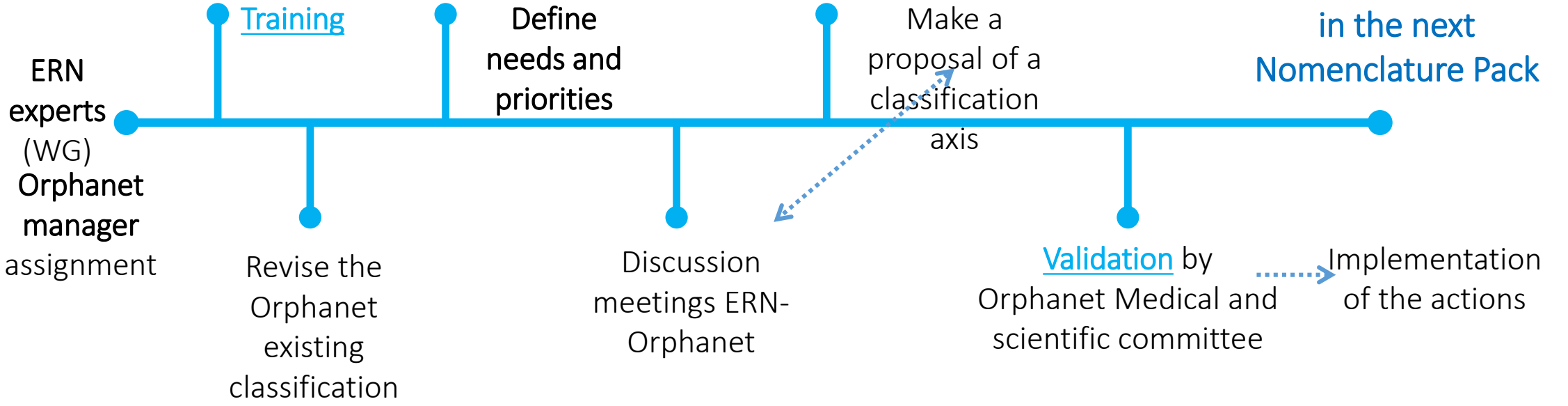
Standartized database

- High-quality, interoperable nomenclature
- Inter and intra-classification consistency
- Transpose RD information

• Orphanet and ERN collaboration steps

ERN collaboration procedural document

- To achive the goal…



- … progress isn't linear

Sometimes, the final completion of the project may take longer than planned

Difficulties in finding **The experts**

Training
More than one session is needed

Define needs and priorities
Not always straightforward

Make a proposal of a classification axis

Release of the changes in the next Nomenclature Pack
can be delayed

Difficulties in finding **Orphanet manager**

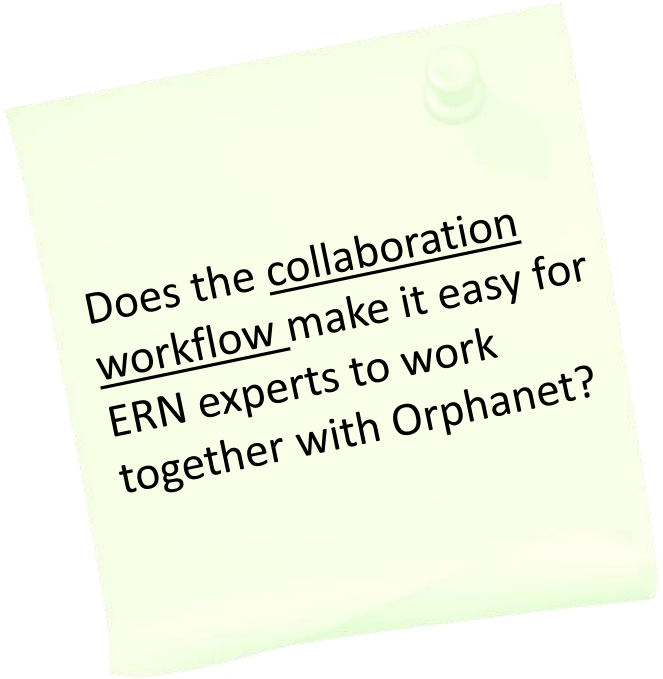
Revise the Orphanet existing classification

Discussion meetings ERN-Orphanet

Validation may need several cycles to be finalized

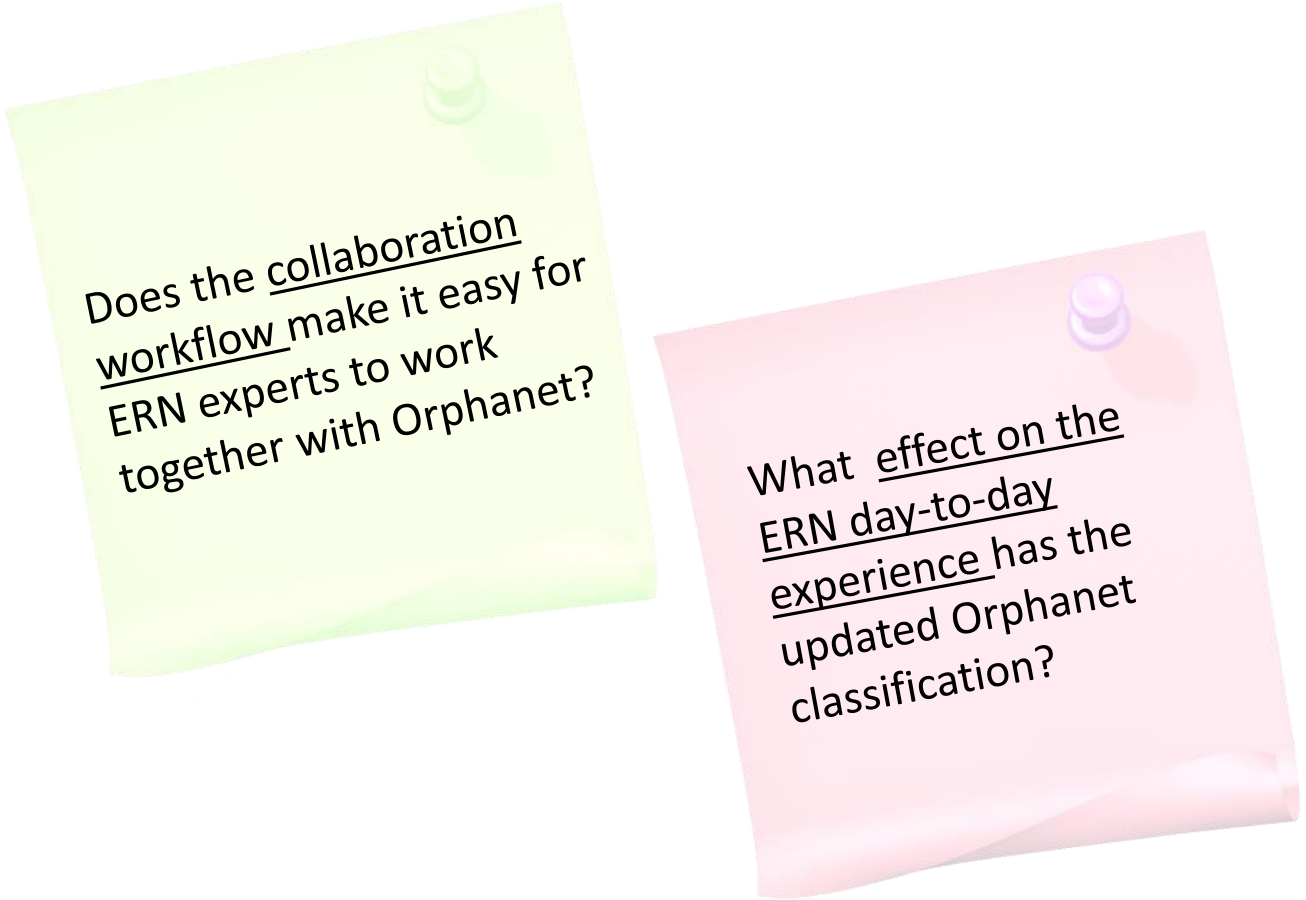
Break-out session

Round table



Break-out session

Round table



Break-out session

Round table

Does the collaboration workflow make it easy for ERN experts to work together with Orphanet?

What effect on the ERN day-to-day experience has the updated Orphanet classification?

Do the Orphanet rules allow enough flexibility to take into account the reality of the clinical settings?

Break-out session

Round table

Does the collaboration workflow make it easy for ERN experts to work together with Orphanet?

What effect on the ERN day-to-day experience has the updated Orphanet classification?

Do the Orphanet rules allow enough flexibility to take into account the reality of the clinical settings?

Break-out session

Round table

What changes do you think should be made to make the collaborative process better?

3 / 3 3

It's already very good Face to face meetings better if it's a complex area eg spina bifida

Again, budget to allow for some in person meetings for large scales revisions would be saving a lot of time and effort from everyone involved

To have a clear contact person for orphanet related matters in each ERN. Disseminate workflow, some ERNs don't know about it or at least it's not at the top Of their heads

Break-out session

Select the key steps for a succesful collaboration

1. Availability of experts to discuss issues, answer questions and/or provide references
2. Availability of enough Orphanet Nomenclature managers to handle requests in a timely manner
3. Easy and reactive communication
4. Respect the timeframe for planned actions
5. Agreement on the communication mode frequency
6. Identify the priorities
7. Decide on the steps of the revision
8. Recruit the experts
9. Include the experts in the writing process
10. Good understanding of the Experts request
11. Good understanding of the Orphanet rules
12. Number of face-to-face meetings

Break-out session

Select the key steps for the straightforward revision of the RD nomenclature with ERNs

Break-out session

Select the key steps for the straightforward revision of the RD nomenclature with ERNs

3 / 3

- 1 Good understanding of the Experts request
- 2 Availability of enough Orphanet Nomenclature managers to handle requests in a timely manner
- 3 Identify the priorities
- 4 Availability of experts to discuss issues, answer questions and/or provide references
- 4 Recruit the experts
- 5 Easy and reactive communication
- 5 Good understanding of the Orphanet rules

Break-out session

- 5 Good understanding of the Orphanet rules
- 6 Include the experts in the writing process
- 7 Decide on the steps of the revision
- 8 Number of face-to-face meetings
- 9 Respect the timeframe for planned actions
- 9 Agreement on the communication mode frequency

key steps

Conclusion

This is a **collaborative project**, and **bidirectional** at every step:
there is an ERN counterpart and an Orphanet counterpart

Orphanet Scientific collaborations with the ERNs:

The four main points to respect are:

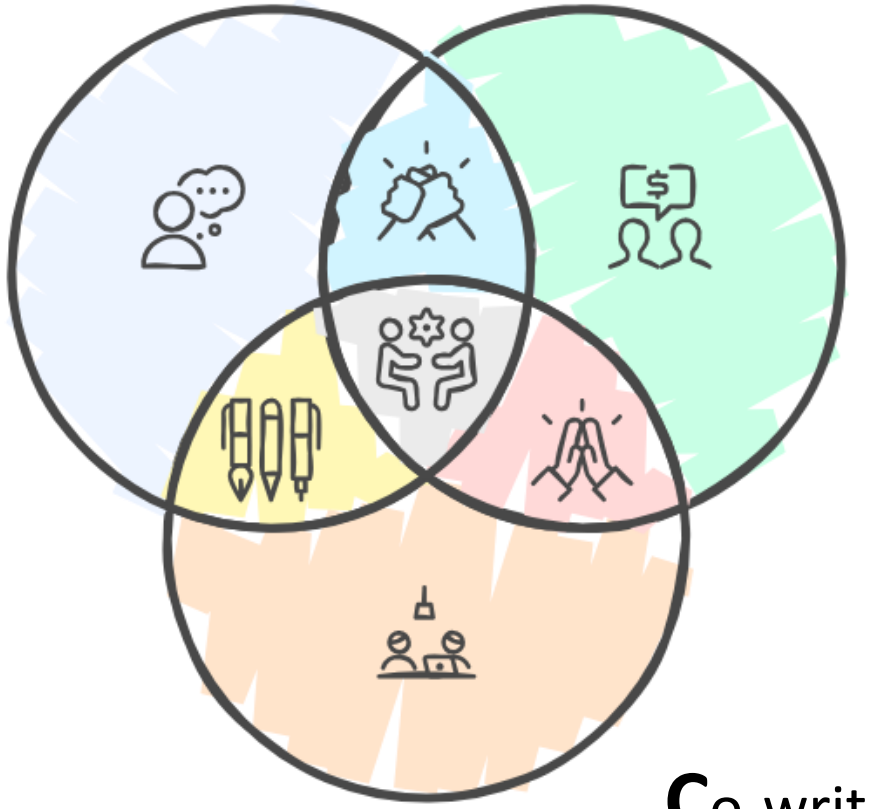
- ✓ Training on the Orphanet standards
- ✓ Fluid and frequent exchanges
- ✓ Engagement in accomplishing the delays
- ✓ Flexibility to apply/accept the most suitable actions

Common limitations observed:

- X Number of people involved
- X Fluid communication
- X Intrinsic time constraints due to Orphanet validation cycles and the high volume of demands to treat

Orphanet Scientific collaborations with the ERNs:

Communication



Commitment

Co-writing

Orphanet and ERN collaborations are **Co-Constructive projects**

Thank you for participating!

OD4RD2 FINAL MEETING

Implementation & adoption of ORPHAcodes in MS

Paris, November 6th 2025

Animators: Ashray VOHORA, Seed MIHIC

OD4RD
Orphanet Data For Rare Diseases

Rare diseases are not visible when using generic terminologies only

Underrepresented

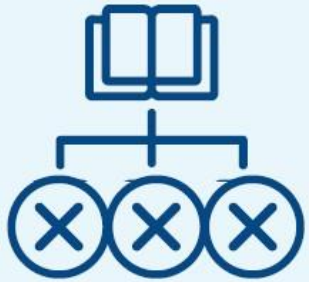
ICD-10: ~83% of RD do not have any code*
ICD-11: ~39% of RD do not have any code*
SNOMED CT: 6% of RD do not have any code*
None of these terminologies have a code for the patients in a diagnostic dead-end/undiagnosed RD patients

Imprecise

ICD-10: 93% of RD do not have a PRECISE code*
ICD-11: 84% of RD do not have a PRECISE code*
SNOMED CT: 6% of RD do not have a PRECISE code*

Not classified as “rare”

RD are "lost" amongst common diseases
Generic terminologies are not exploitable for RD-specific statistics



WHY ORPHAcoding
https://od4rd.eu/communication-material/WHY%20ORPHAcoding%20vs%20Other%20Terminologies_VF.pdf

Benefits of ORPHAcoding

Using ORPHAcodes ensures visibility of rare diseases in health data
This visibility helps:

- facilitate the primary and secondary use of RD data
- adequate cross-border care
- equity in healthcare access
- epidemiological monitoring and better understanding of disease burden

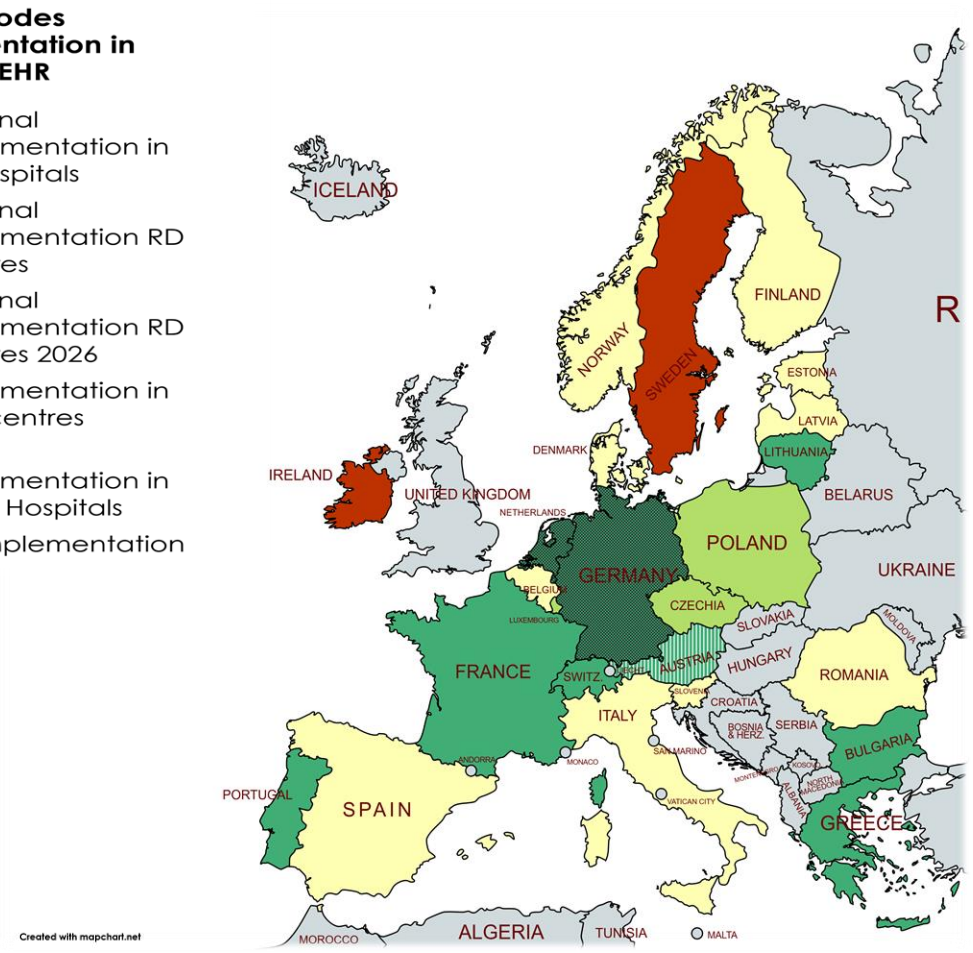
ORPHAcodes also enable interoperability

Recognised as best practice by the European Commission since 2017
Recommended in the 78th WHA resolution on RD

State of play across Member States

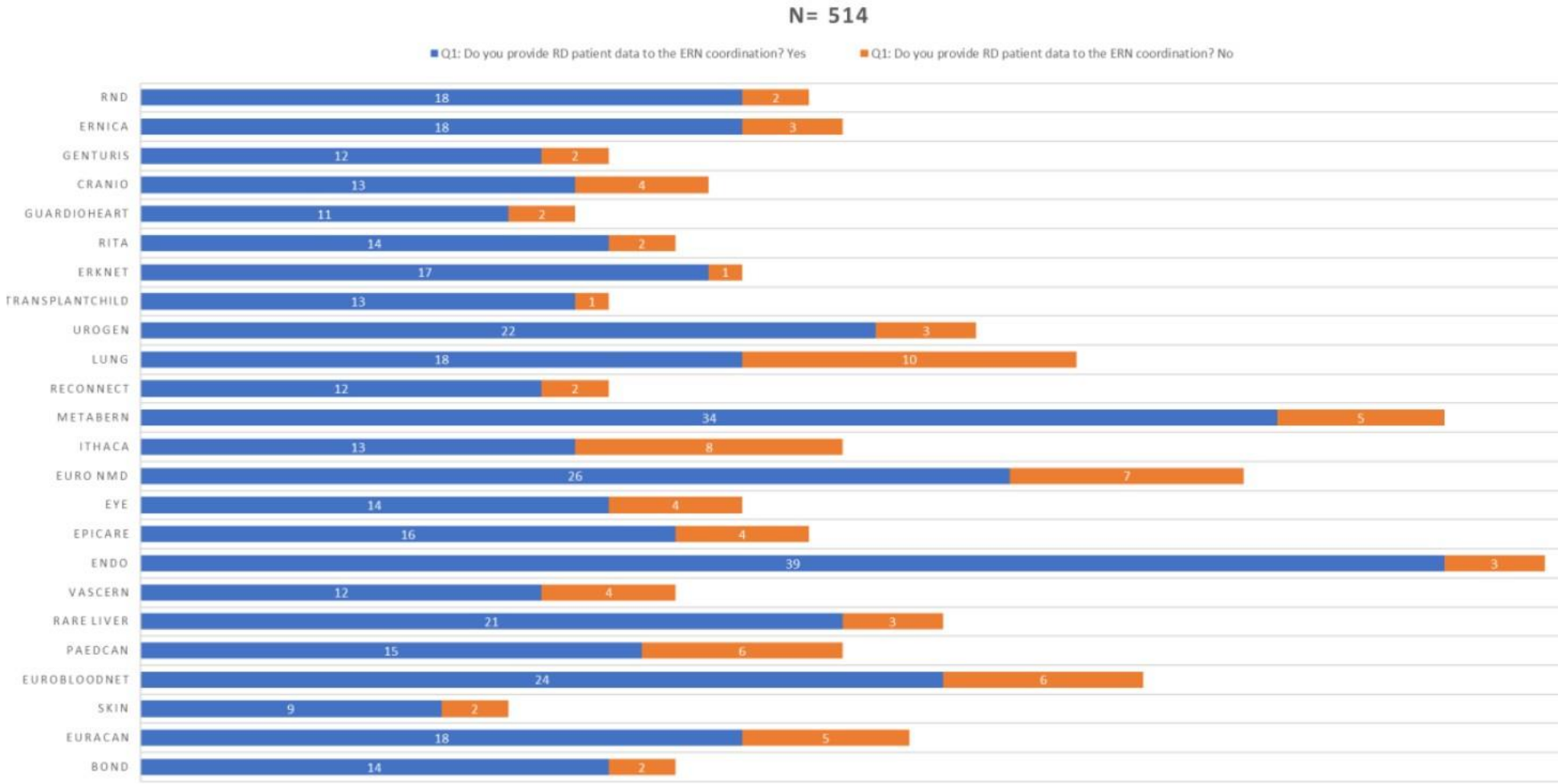
Heterogeneous

- Implementation: some MS have national frameworks or legal mandates for ORPHAcode use
- ORPHAcode usage: some use primary coding with ORPHAcodes, while others rely mainly on transcoding from other terminologies, which limits consistency



ORPHAcodes implementation in EHRs of hospitals or RD centres

Do you provide RD patient data to ERN coordination?

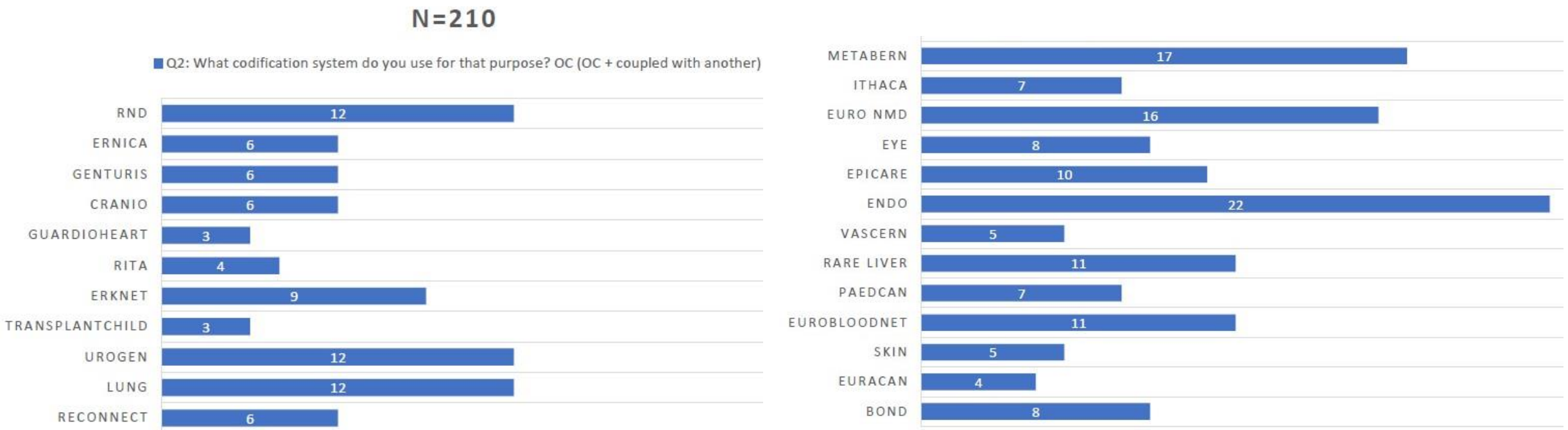


Yes

No

ERN survey analysis: https://od4rd.eu/03-deliverables/OD4RD2_ERN-Survey_11QC%20analysis_VF.pdf

What codification system do you use for what purpose: pooled answers “ORPHAcodes” and “ORPHAcodes coupled with another system”



ERN survey analysis: https://od4rd.eu/03-deliverables/OD4RD2_ERN-Survey_11QC%20analysis_VF.pdf

OBJECTIVE of the workshop session

What can OD4RD and ERNs do to increase ORPHAcode adoption and implementation, to ensure that RD patients are visible across healthcare systems and that consistent, high-quality information is collected across Europe?

mentimiter

Round table

1. What can ERNs do for advocacy in their network to support the implementation and adoption of ORPHAcodes, on a national and local (hospital) level?
2. What can OD4RD do to support the ERN networks in this mission?
3. Please briefly explain the state of play in your network - share success stories or troubles to be addressed
4. For those that have not implemented ORPHAcodes - why? Can OD4RD help you to change this?

Take-home message

ORPHAcodes are recognised as the best practice for coding RD patients and as a policy priority. The focus should now shift from recognition to consistent implementation and exploitation, thereby ensuring that every person living with RD is visible in data and care pathways.

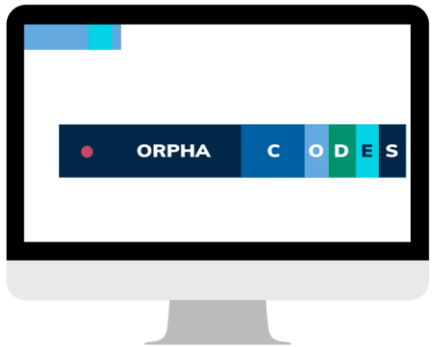
Thank you for participating!

OD4RD

Orphanet Data For Rare Diseases

CODING GUIDELINES FOR RD

> Why ORPHAcodes?



To ensure ALL RARE DISEASES are visible in Health information System

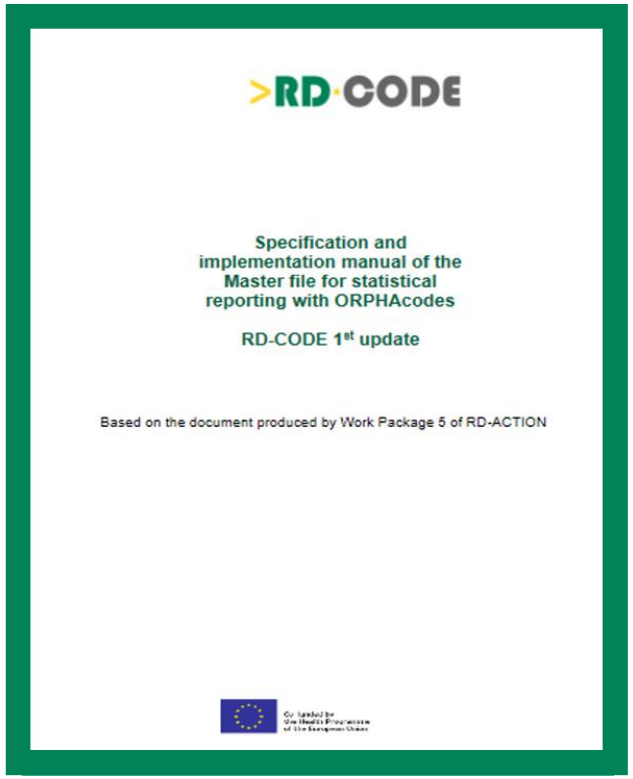
To allow RD data to be interoperable among hospitals, regions, and countries.

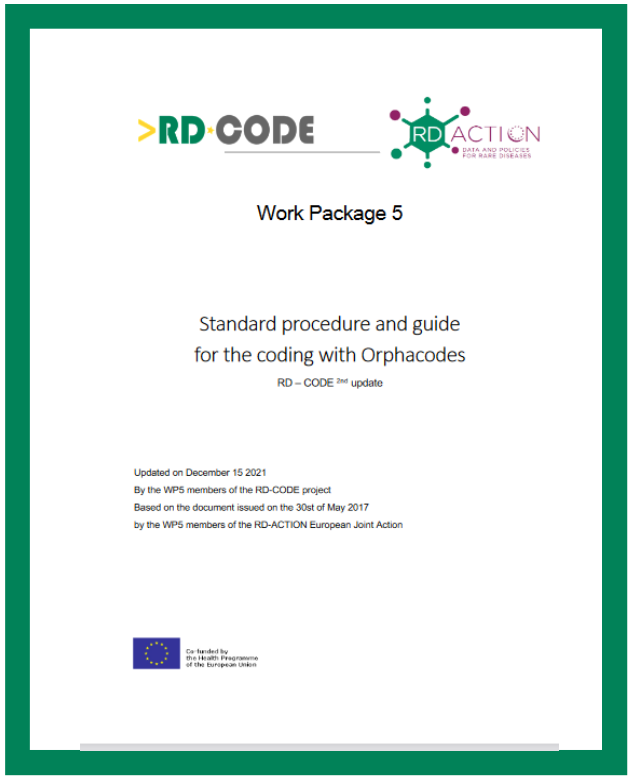
To answer a range of public health and research questions and make evidence-based decisions

- **Coding guidelines** are a set of rules and best practices for translating clinical documentation into standardized diagnosis **codes**, so data are accurate for reimbursement, quality metrics, epidemiology, etc…
- **Why standardizing coding guidelines matters:** accuracy, legal compliance, fair reimbursement, comparable data at European level, etc…
- **Benefits of harmonized ORPHAcodes coding guidelines**: European consistency, better clinical and research data, stronger ERN visibility, increased trust in ORPHAcodification.

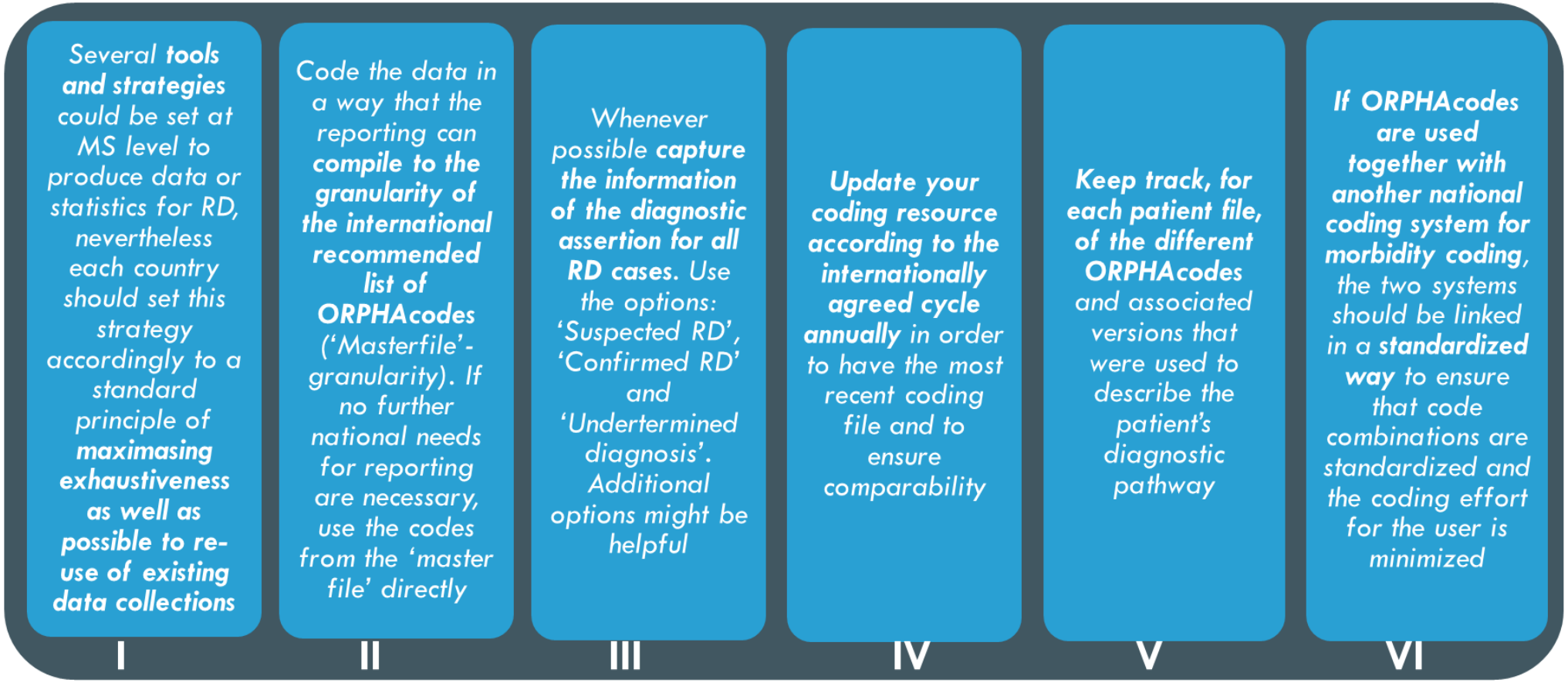
CODING GUIDELINES

- RD-CODE Guidelines





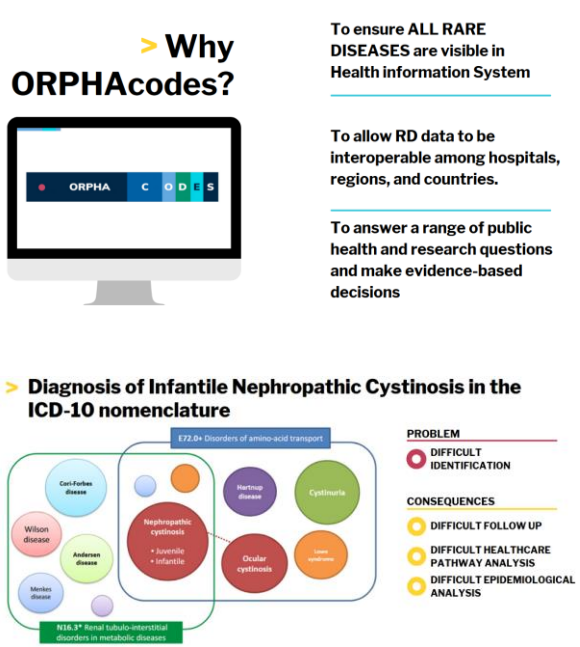
CODING GUIDELINES



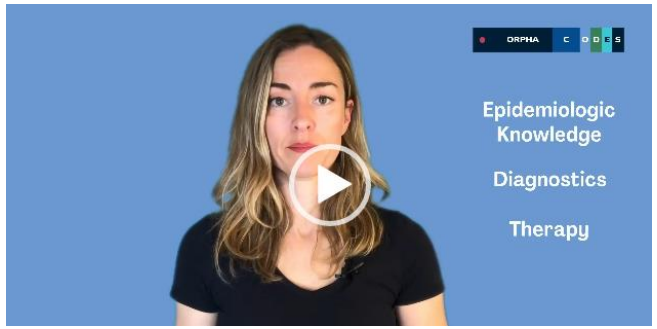
www.rd-code.eu

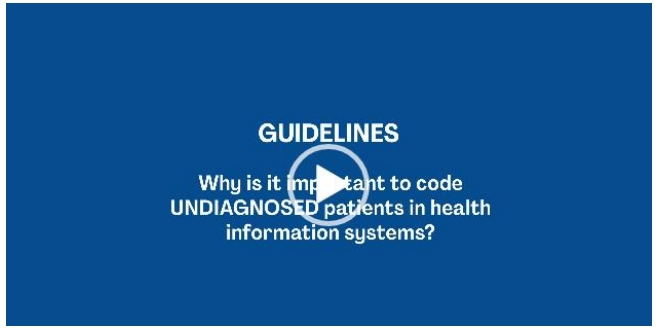
CODING TRANINGS AND RESOURCES: training modules

PRESENTATIONS

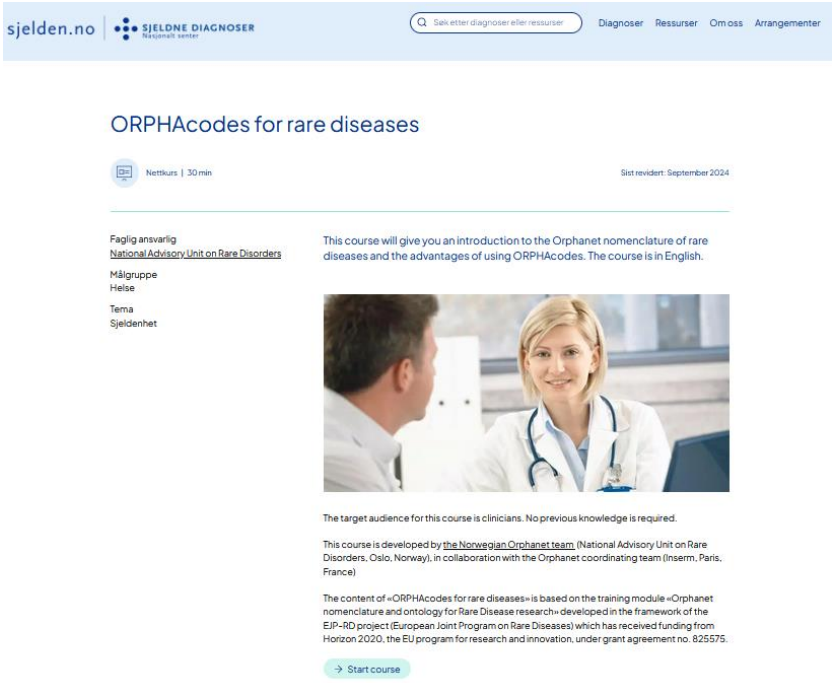


VIDEOS





E-LEARNING



[ORPHA Coding – RD-CODE]

[Orphanet - YouTube]

[ORPHAcodes for rare diseases - Sjelden]

CODING TRANINGS AND RESOURCES: OD4RD trainings

USE CASE: CLINICAL MANIFESTATION OF A MAIN DIAGNOSIS (1)

Example: a patient with Classical-like E
also presents with mitral valve prolapse

- Cardiac abnormalities such as MVP are
  Danlos syndrome
- Coding with MVP would imply a « part
- 2 ORPHAcodes would exist for the sam
  next example)

/!\ However, clinicians might need to indicate t
management and statistical analysis

Solution: Use the patient's main diag
descriptive field if available
* in this case ICD-10 code as the standard car

USE CASE: CODING AFTER ORPHACODES INACTIVATION (2)

***Example:** Coding issue addressed to the Orp*
'A national expert centre contacted us becaus
puberty (CPP).
Considering that ORPHA:759 'Central precoc
ORPHA:169615 'Idiopathic CPP' have been
*suggest to the physician to use the **ORPHA6***
though the patient has not been genetically t
Moreover, in our country the CPP is considered rare.

Solution: Despite the recent nomenclature chan
ORPHAcodes, it is not recommended to use an ORP
desc
The mention "genetic" implies that this ORPHAcode mu
- **If there is a genetic etiology suspiscion: use**
- **If there is no genetic testing confirmed o**

USE CASE: A PATIENT WITH TWO CO-EXISTING MAIN DIAGNOSES (4)

Example: a patient in a consangui
SLC34A3 mutation) ORPHA:1572
ORPHA:598. One of his siblings ha
has been diagnosed only with Multi

/!\ This is a case of a joint occurrence

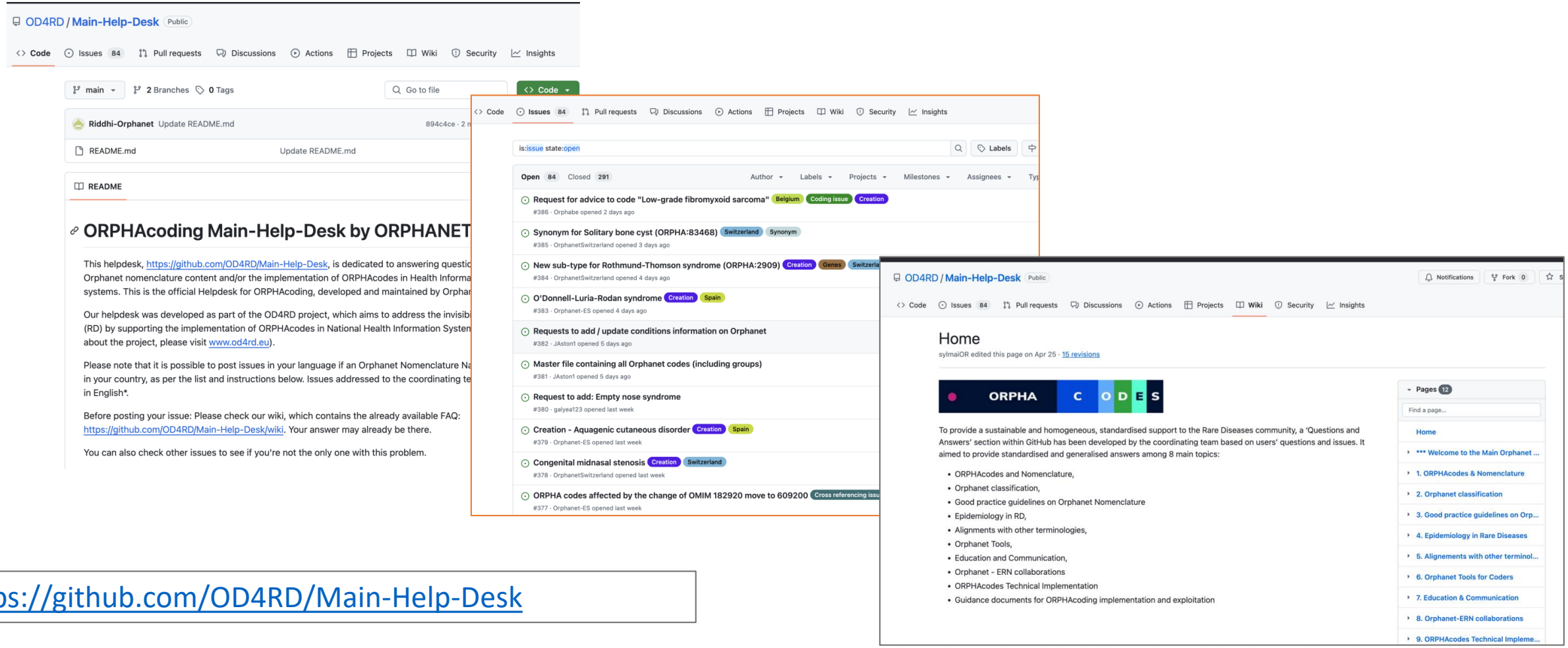
Solution: Use both
diagnosis observ

USE CASE: USE OF ORPHACODES BELONGING TO A GROUP OF DISORDERS (5)

Example:

- ☑ **Non confirmed diagnosis [diagnosis assertion option available in the system]**
- ☑ **National official requirements**
- ☒ **Insufficient knowledge of the classification system by the user**

CODING TRANINGS AND RESOURCES: OD4RD helpdesk



https://github.com/OD4RD/Main-Help-Desk

CODING COMPLEX CASES

Coding of patients with both structural and numeric genetic abnormnalities #330

OrphanetSweden opened on Feb 20

Dear colleagues!

We were hoping you could offer some guidance on how to ORPHAcode patients with deletion-duplication syndromes as well as offer some clarity on coding patients with either deletion or duplications. In most instances the Orphanet nomenclature seems to be organized by grouping disorders first by if they are caused by deletion OR duplication. For instance in the group ORPHA:98142 Partial autosomal deletion and its children which consist of a designated group for each relevant chromosome.
How should a patient with a del/dup rearrangement be coded, especially if both rearrangements are deemed to contribute symptoms?

A similar question concerns the coding of patients having both structural and numeric abnormalities.
Example: a patient with 45X/46X,idic(Y)? InWould the code ORPHA:1772 45,X/46,XY mixed gonadal dysgenesis be applicable in spite of the rearranged Y or would perhaps the group ORPHA:96325 Isochromosome Y be a better fit?

I was also hoping you could offer some guidance on how to code patients with either deletion or duplication which does not correspond to the breakpoints specified is not found in the Orphanet nomenclature. EsFor instance the cases where the rearrangement might either encomapass the breakpoints specified in an ORPHAcode or have a partial overlap.

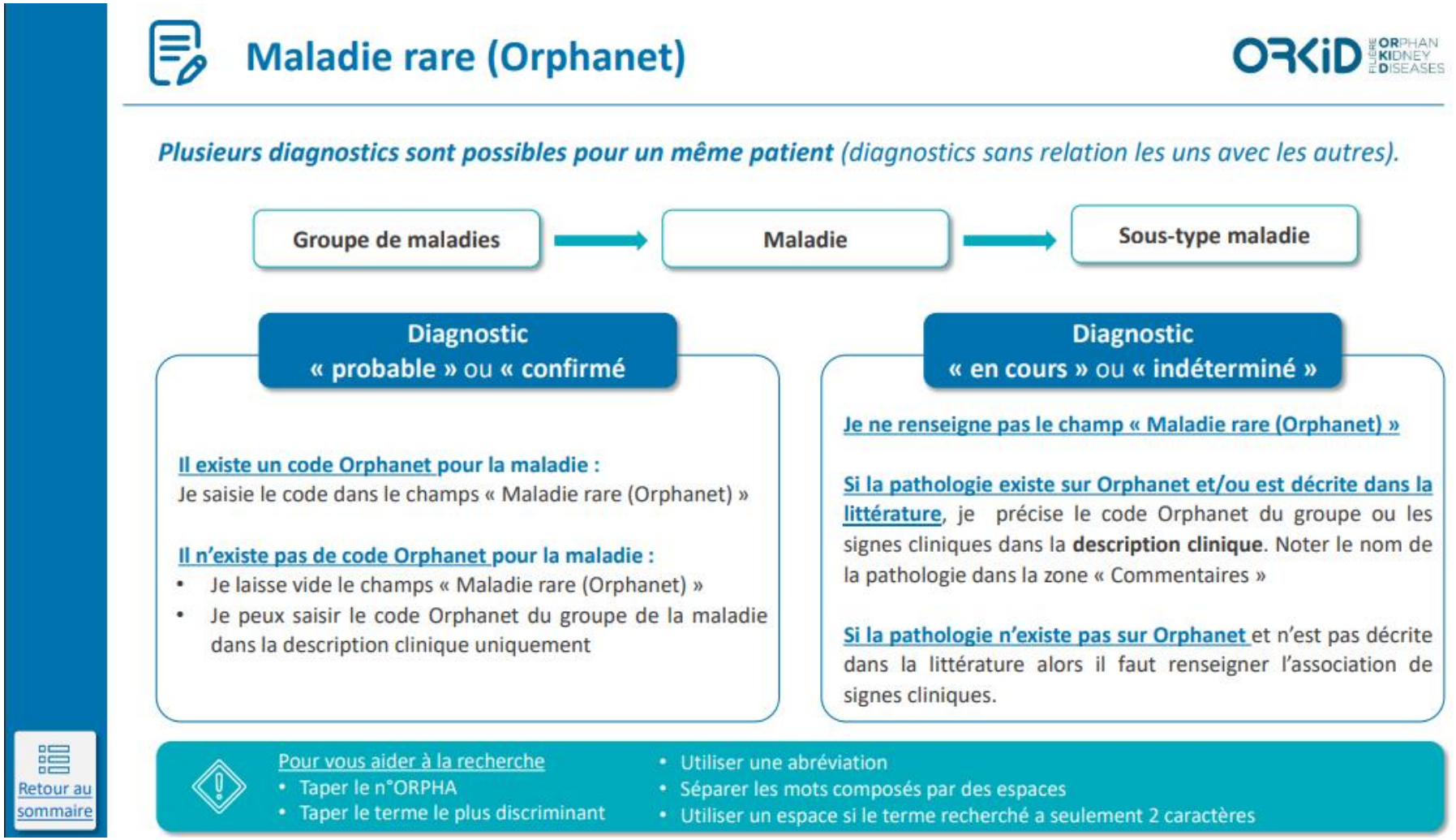
Example: ORPHA:96121, 7q11.23 microduplication syndrome, can this code be used also for patients with a duplication which is larger in size than the specified band (q11.23) or smaller in size, not containing the whole band (q11.23)?

I did not manage to find clarity on this topic in the FAQ and was hoping you could offer some general principles on how to reason about similar cases where an available ORPHAcode is describing a specific genetic alteration.

Best Regards,
Orphanet Sweden

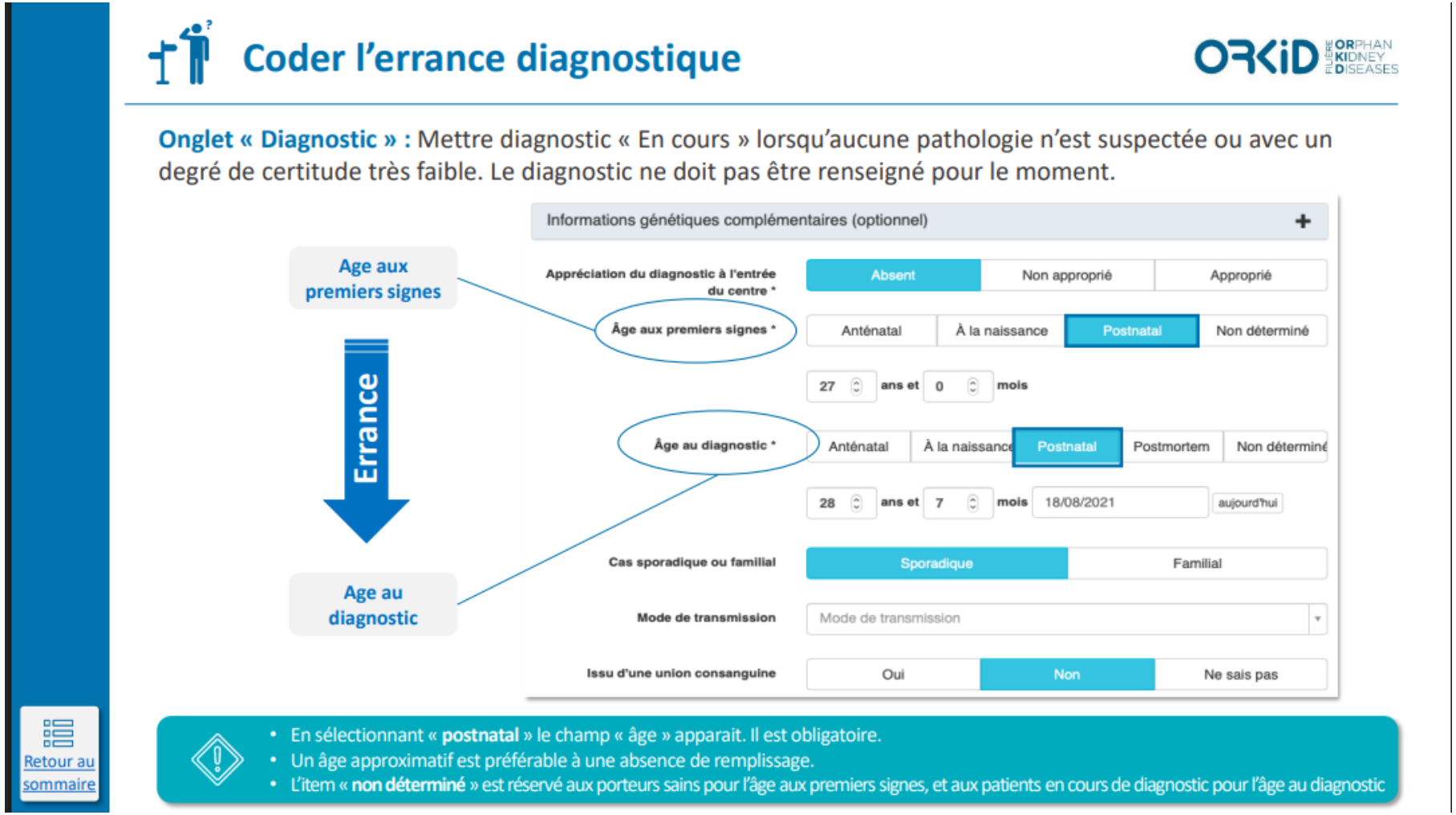
Complex cases for which an ERN guideline would probably be needed

CODING GUIDELINES FSMR



filière de santé maladies rares

CODING GUIDELINES FSMR



CODING GUIDELINES FSMR

4- Cas par maladie :

→ **Cholangite auto-immune** : mettre le statut du diagnostic « En cours » jusqu'à la détermination du type exact de cholangite.

→ **Overlap Syndrome (HAI+CSP) / (HAI+CBP)**: Un code ORPHA a été attribué mais ne répond pas à nos attentes. En attendant sa corre
voici la démarche à suivre :

- Renseigner les deux diagnostics sur BaMaRa (diagnostic 1, diagnostic 2).
- Créer deux fiches sur ORBIS, une par maladie.

→ **Maladies vasculaire porto-sinusoïdales** : Code ORPHA : 596937 récemment implémenté dans BaMaRa

Dans le cas où une VPO (veinopathie portale oblitérante) a été renseignée à la place de la maladie porto-sinusoïdale avant l'implémentation du code ORPHA, il est nécessaire de les corriger. Ceci permet une visibilité plus exhaustive sur le plan épidémiologique.

→ **Cholangite ischémique :** pas de code ORHA pour le moment

Mettre le statut de diagnostic « confirmé » et renseigner la maladie dans le champ commentaire. **La fiche ne sera pas valide (en rouge) mais sera bien comptabilisée dans le calcul des items PIRAMIG.**

⚠ **Pour le champ commentaire : attention à bien orthographier la « Cholangite ischémique » comme indiqué ici.** Ceci nous permet de revenir sur les dossiers si besoin.

FILFOIE
FILIÈRE DE SANTÉ MALADIES RARES DU FOIE DE L'ADULTE ET DE L'ENFANT

Guide des règles de codage BNDMR - Filfoie

CODING GUIDELINES FSMR

GUIDE DE CODAGE: HÉMOCHROMATOSES

Hémochromatose rare : hémochromatose autre que homozygotes C282Y HFE : donc liée à SLC40A1, TF, CP, HAMP, HJV, BMP6, TFR2, variant privés HFE (hors H63D et S65C) ou sans cause génétique identifiée pour l'instant

- **un consensus international** récent sur la définition d'une hémochromatose et donc par extension des hémochromatoses non HFE C282Y. Qui peut nous aider à "coder" ce qui relève de l'anormal.
- Ce qui veut dire:
  - Surcharge en fer: CHF IRM significative (malheureusement pas de consensus sur la valeur, on peut dire 100µmol/g pour simplifier).
  - Et pas de pathologies hémato ou alcool ou métabolique prédominante sur le tableau
- En ce qui concerne la maladie hémato ou alcool prédominante c'est assez "simple" cliniquement de savoir si cela peut être la seule explication pour la surcharge. Pour le métabolique pour que cela soit prédominant c'est plus délicat que chacun doit juger selon son habitude.

ON NE CODE DANS BAMARA QUE:

l'hémochromatose HFE C282Y homozygote pour laquelle il y a :

- soit la nécessité de réaliser un traitement inhabituel (chélateur du fer par voie orale ou par injection)
- soit la nécessité d'une étude génétique exhaustive, sur observation d' anomalies phénotypiques (survenue anormale d'une cirrhose chez un sujet jeune, symptômes cliniques anormaux)

Les HFE 282Y seul ou composite qui ne rentre pas dans les deux catégories ci-dessus et dans la définition des hémochromatoses rares ci contre **ne relèvent pas des maladies rares.**

MALADIES HÉRÉDITAIRES MÉTABOLIQUES
Filière nationale de santé

Round table

- **Goal of this session**: identify concrete ways Orphanet can help harmonize ERN coding guidelines across networks, integrate ERN feedback into the Orphanet tools, and support shared visibility and dissemination via OD4RD3 trainings, webinars, GitHub, documents…

Round table

Challenge	Possible actions
Heterogenity between ERNs Depends on the complexity of the classification may be (?) not always needed?	ERN coordination should lead and work with WG of experts
Undiagnosed code usage would differ in different countries even in EU (again heterogenity)-national differences needs to be taken into account	InterERN would be considerd
	Special issue for guidelines :)

Round table

Challenge	Possible actions
Scientific societies have monopol of producing guidelines in general	A backbone on how to produce guidelines
Gathering experts from many different thematic groups to give their input	Standart templates/formats
Coding guidelines needs to repect international classification	Guidance in granulartiy level
Convincing everyone their importance	Feedbcak from early stage of the guidline to be sure that it serves the purpose it is supposed to
Time of experts	Hirachy oder
	An example of a guideline
That every ERN follows the same principles when producing guidelines	User friendly online tool
Difficulty in collecting and classifying evidence	

Thank you for participating!