ORPHANET DATA FOR RARE DISEASES

NCPs/HCPs meeting– March 19 2025

OD4RD 2
Orphanet Data For Rare Diseases

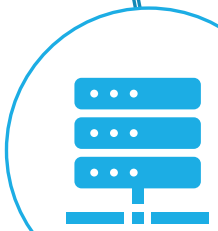
This presentation is part of the project OD4RD2 which has received funding from the European Union's Health Programme. The content of this presentation represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission and/or 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

- Introduction to OD4RD 2 project – Ana Rath (3 min.)
- Interactive Mentimeter session 1 (3 min.)
- ORPHAcodes: why and how? – Ana Rath (12 min.)
- The Orphanet nomenclature national hubs netwrok – Terese Bodérus (10 min)
- Interactive Mentimeter session 2 (3 min.)
- Discussion (10 min.)

OD4RD2 GENERAL OBJECTIVES

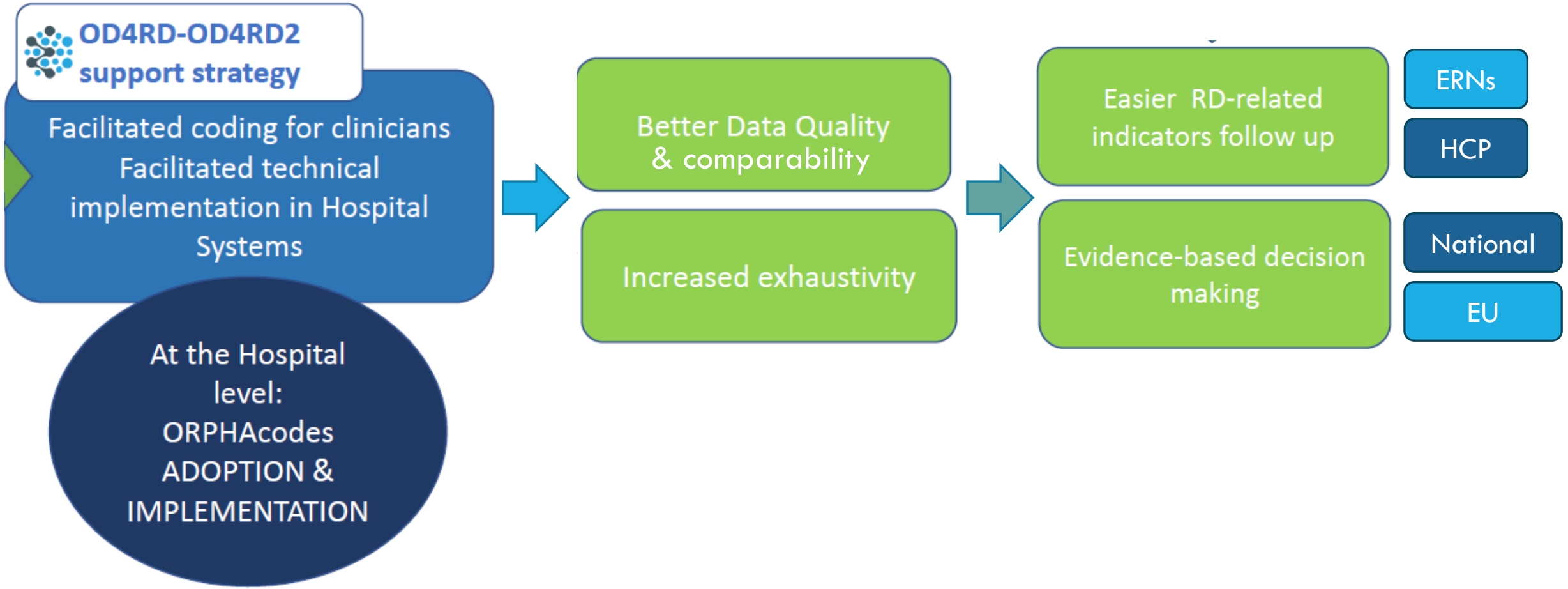
To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, by the maintenance and the support to the implementation of the Orphanet nomenclature of RD



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

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

IMPACT


OD4RD-OD4RD2 support strategy
Facilitated coding for clinicians
Facilitated technical implementation in Hospital Systems
At the Hospital level:
ORPHAcodes
ADOPTION & IMPLEMENTATION
Better Data Quality & comparability
Increased exhaustivity
Easier RD-related indicators follow up
Evidence-based decision making
ERNs
HCP
National
EU

OD4RD STRATEGY

ORPHANET/ERN COLLABORATION

Scientific expertise

Data validation

orphanet ⇄ European Reference Networks

Expertise on **databasing and classification**

→ Standardized structure of clinical information

Data for **analysis** of RD

OD4RD STRATEGY

REINFORCE THE NATIONAL LEVEL TO ADD EUROPEAN VALUE

orphanet

European Reference Networks

JARDIN

EHDS

National hubs

- Information & data
- ORPHAcode implementation support

National nodes

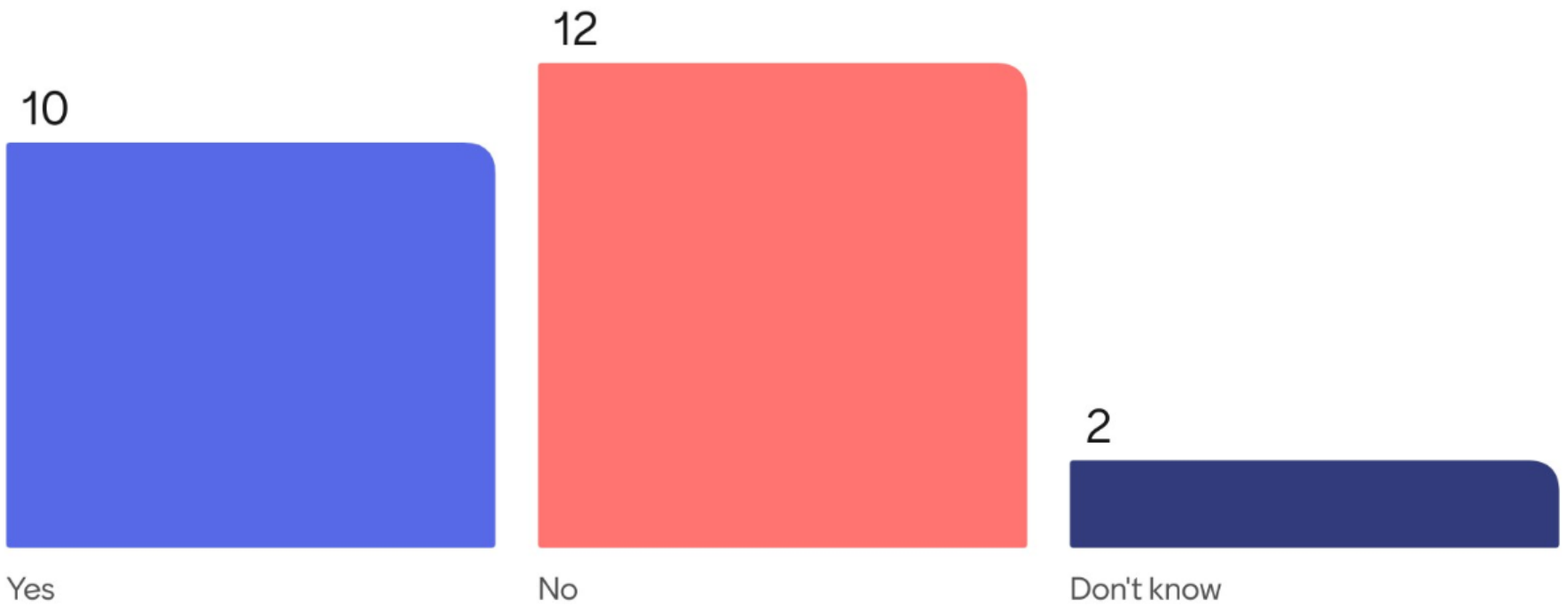
- Care and research activities
- CPGs
- ORPHAcode implementation

National nodes

- Hospitals' data
- EHRs
- ORPHAcode implementation

Mentimeter

Have you implemented ORPHAcodes in your Hospital?



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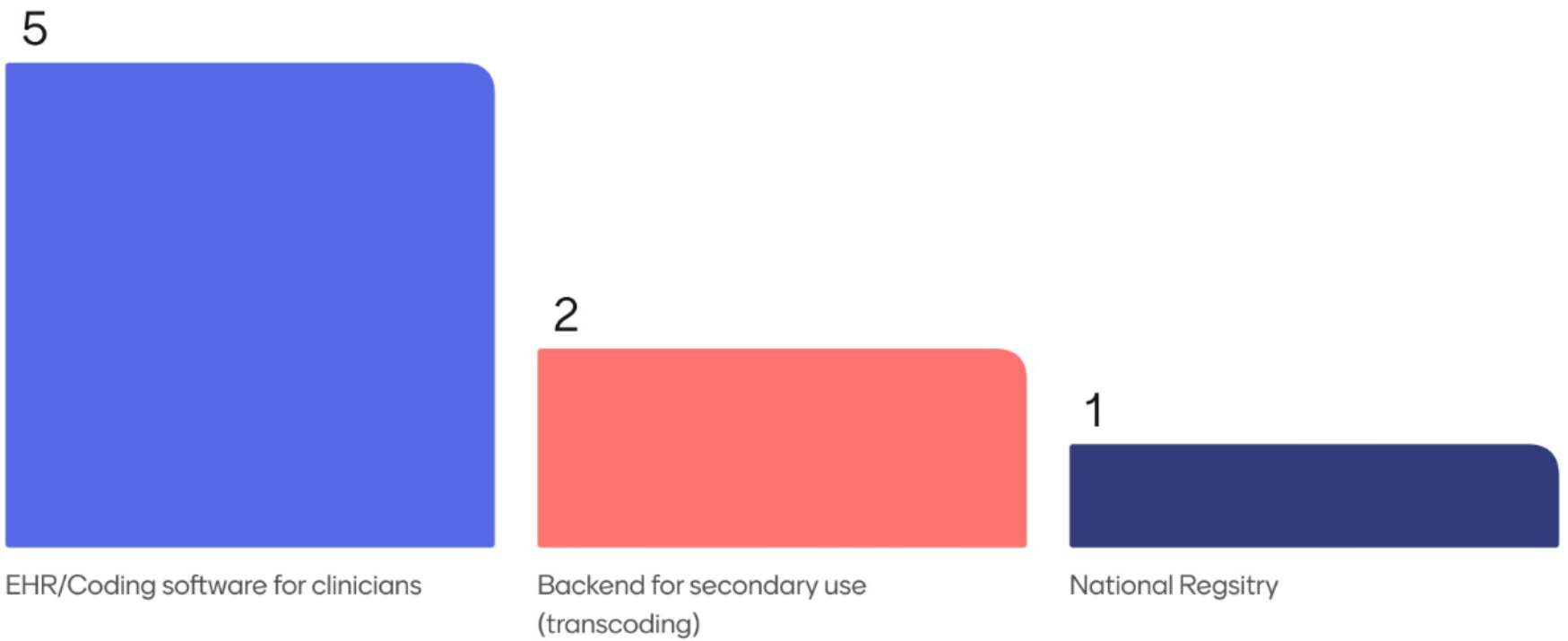
Mentimeter

If yes, where are they implemented?



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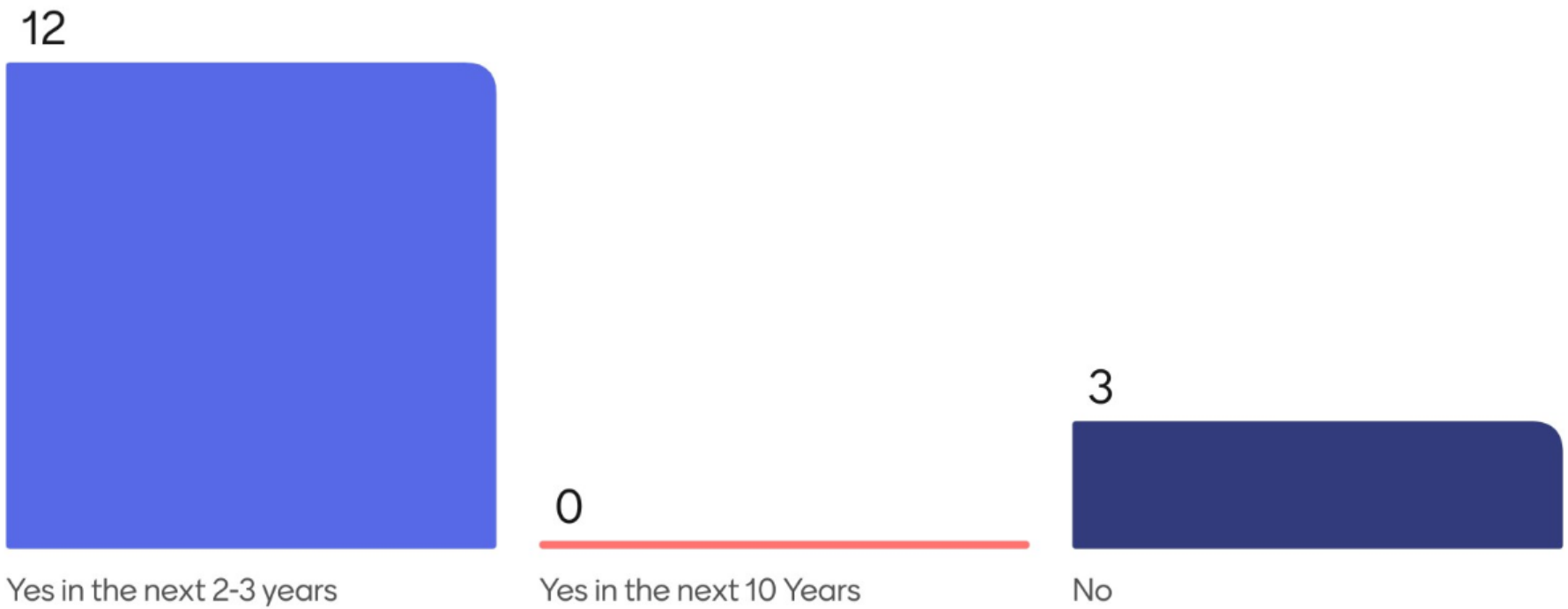
Mentimeter

If no, are there plans to implement ORPHAcodes?



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THANK YOU VERY MUCH!

WHY ORPHACODES

NCPs/HCPs meeting– March 19 2025

OD4RD 2
Orphanet Data For Rare Diseases

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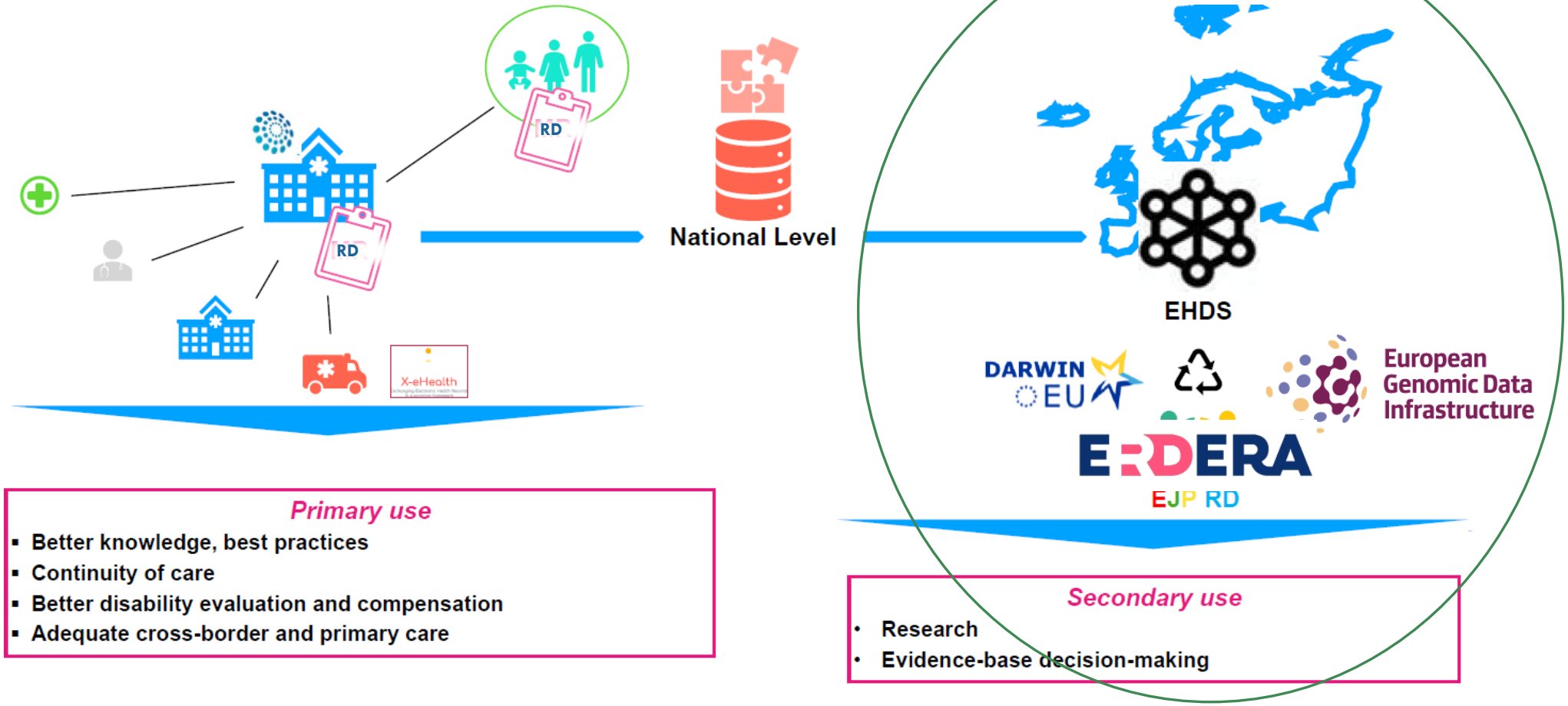
THE FUTURE: RD IN THE DATA ECOSYSTEM



Primary use

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

Secondary use

- Research
- Evidence-base decision-making

FIRST STEP: SOLVING RD INVISIBILITY

Underrepresented

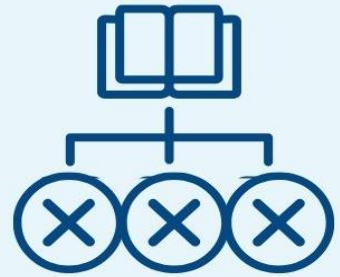
ICD-10: ~83% of RD do not have any code*
ICD-11: ~37% of RD do not have any code*
SNOMED CT: 7% of RD do not have any code*
None of these terminologies have a code for the patients in a diagnostic dead-end/undiagnosed RD patients

Imprecise

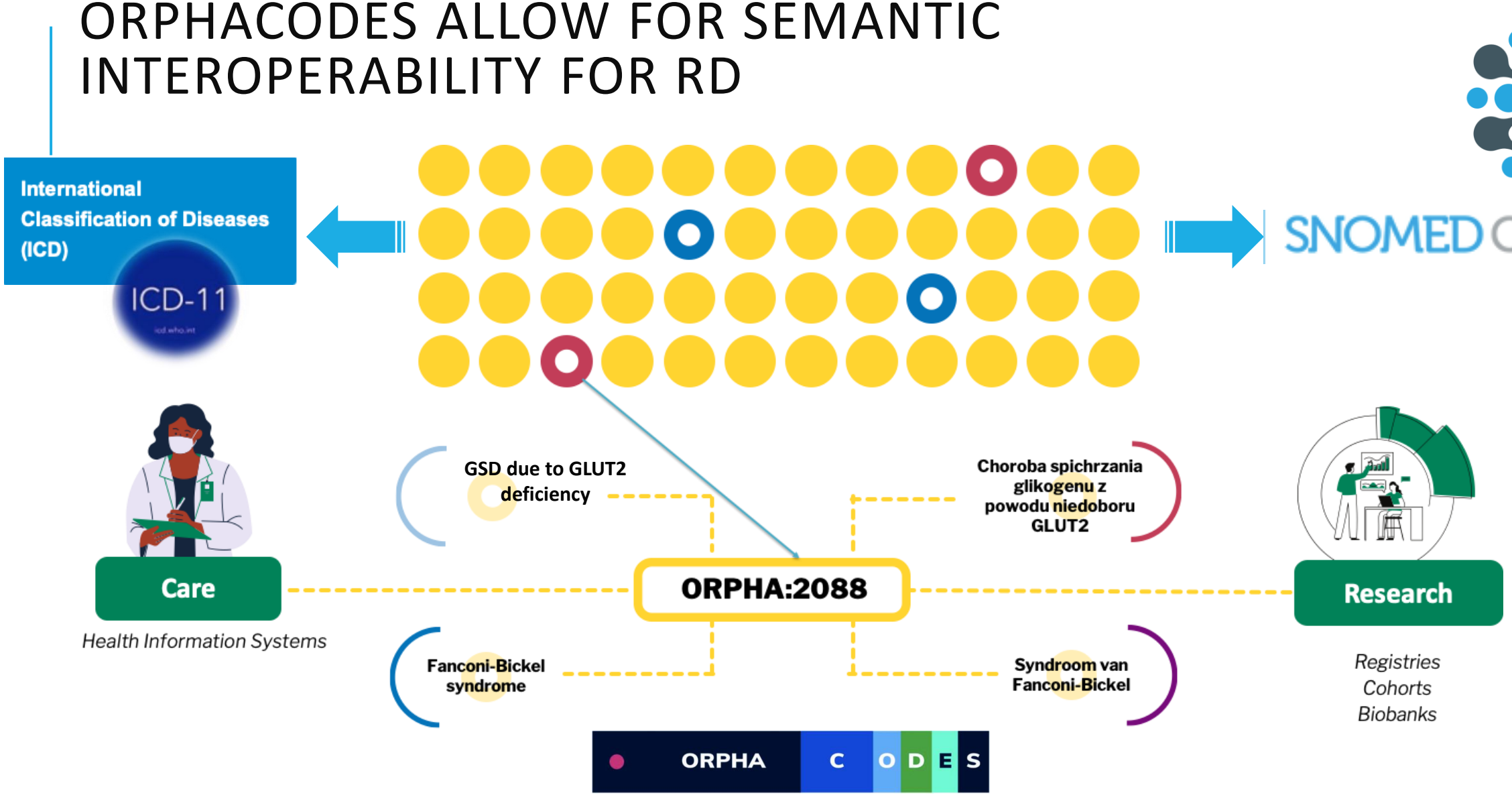
ICD-10: 93 % of RD do not have a PRECISE code*
ICD-11: 75 % of RD do not have a PRECISE code*
SNOMED CT: 7 % of RD do not have a PRECISE code*

Not classified as “rare”

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



ORPHACODES ALLOW FOR SEMANTIC INTEROPERABILITY FOR RD



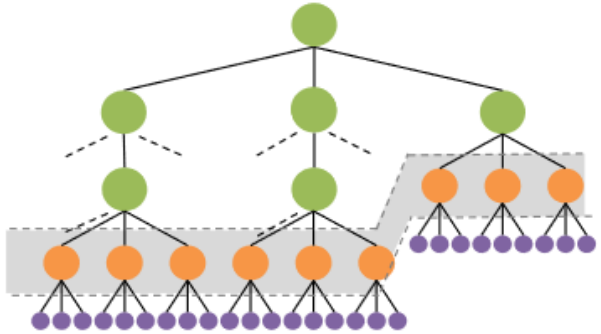
EC Best practice 2017

A RD-SPECIFIC CLASSIFICATION SYSTEM ADAPTED TO THE NEEDS OF DIFFERENT USERS

ORPHA CODES

Clinical definition:

Disorders are clinically homogeneous entities described in at least two independent individuals, confirming that the clinical signs are not associated by fortuity.

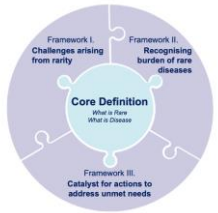


Group
Disorder
Subtype

Examples of use

Public health
Diagnosis, epidemiology
Research

"Classification level"



ENG FR SP DE IT NL PG PL CS UE CHN TUR

Comprehensive, standardized, evidence-based, interoperable, versioned, computable and free (CC-BY 4.0)

NEW ORPHACODE ORPHA:616874

Rare disorder without a determined diagnosis after full investigation

Suggest an update

Disease definition

A rare disorder for which all reasonable efforts have been done by rare diseases experts to determine a diagnosis according to the state of the art and available diagnostic capabilities, but did not enable to conclude on a clinically known concept. It is recommended to restrict the use of this entity for coding purposes to rare disease experts.

ORPHA:616874

Classification level: Disorder

Synonym(s):

Fully investigated rare disorder without a determined diagnosis

Prevalence: -

Inheritance: -

Age of onset:

A summary on this disease is available in Français (2021) Español (2021) Deutsch (2021) Italiano (2021) Português (2021) Nederlands (2021)

A code for specific and unambiguous designation of undiagnosed patients

Available in the Orphanet nomenclature, together with the guidelines

Publication:
https://link.springer.com/article/10.1186/s13023-024-03030-2

Guidelines video ->

http://www.rd-code.eu/guidelines-for-coding-undiagnosed-patients-video/

IMPLEMENTING ORPHACODES ALLOWS FOR 1ARY AND 2DARY USE OF DATA

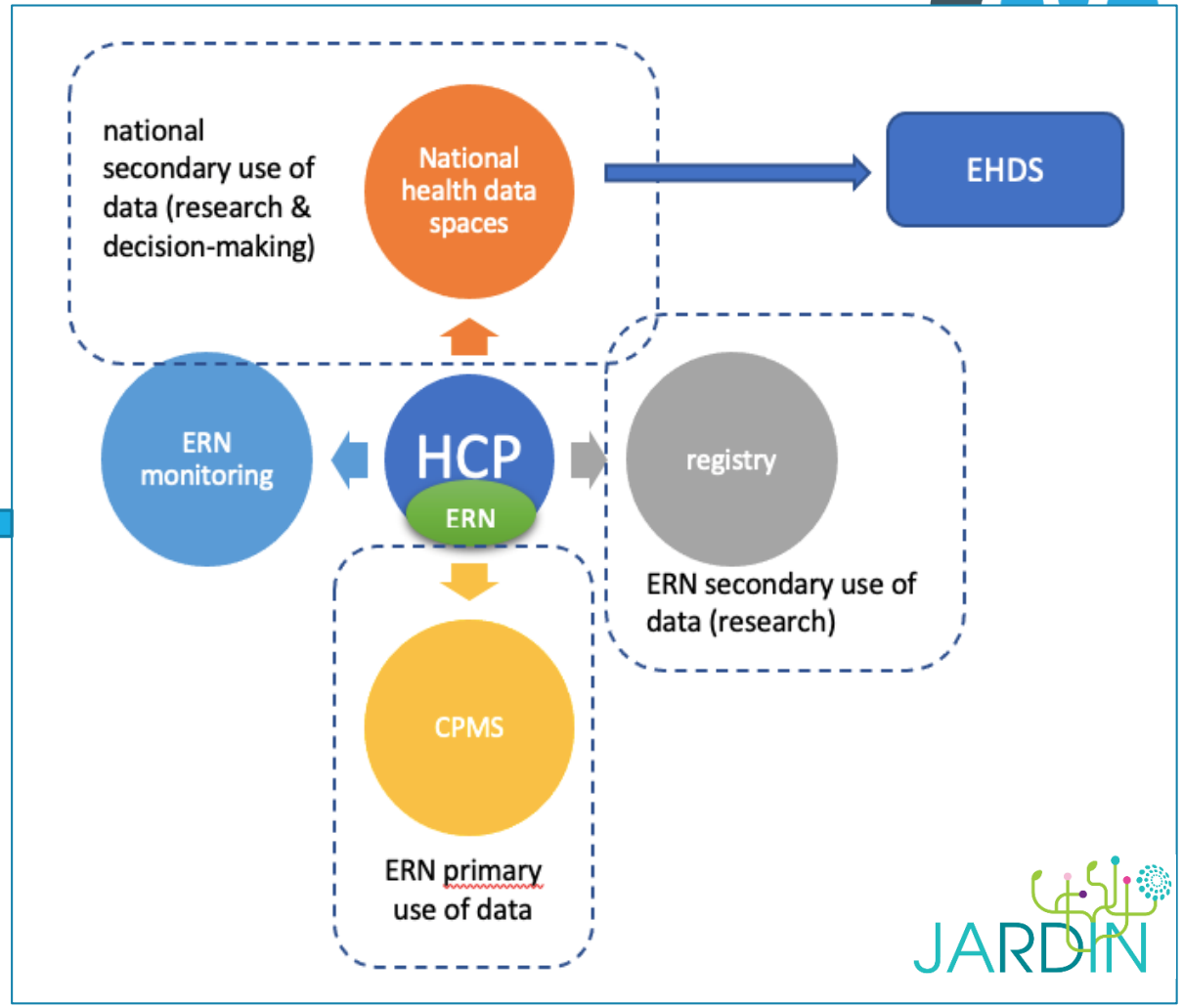
RD data use cases:

- Primary use
  - Planned care
  - Unplanned care
  - CPMS
- Secondary use

Evidence -based decision-making
  - HCP indicators
  - MoH indicators
  - ERN monitoring

Research
  - (ERN) registries
  - EHDS
  - Undiagnosed programmes

Demonstrating ERNs added-value

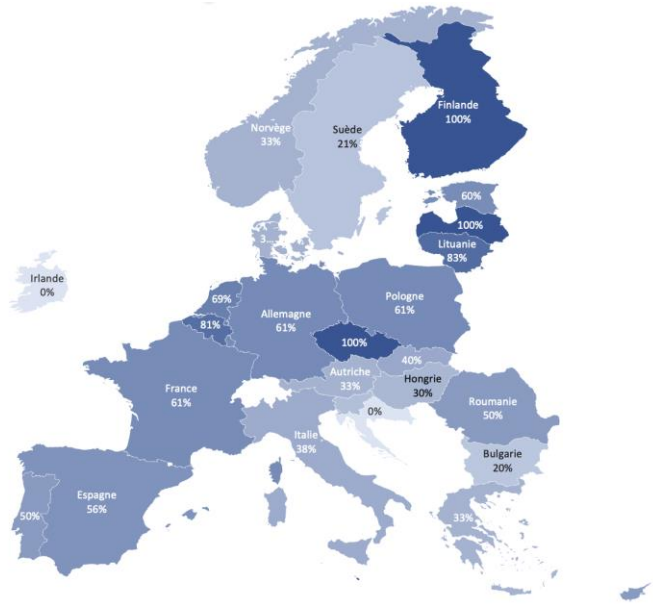


WE ARE READY

Codification and datasets

- **2009:** Codification as priority in the Council Recommendation for RD
- **2013**: CEGRD recommendations on Minimum Data Sets for RD
- 2014: CEGRD recommendations on Orphacodes
- **2015-2018:** RD-ACTION guidelines on codification for harmonized statistics on RD in Europe
- JRC's CDE for RD registration: ORPHAcodes
- 2017: ORPHAcodes as Best practice by the EC
- **2017:** creation of ERNs: need for data generation to monitor ERN activity and added-value
- **2018-2021:** RD-CODE: pilot ORPHAcode implementation in 4 countries; renewed recommendations; code for undiagnosed patients
- **2019:** ERN registries
- **2019:** European Joint co-fund programme for RD (EJP-RD): ORPHAcodes in standardized data models for data exploitation
- 2020-2023: X-eHealth: approved specification for ePatient summary V3 including RD for cross-border unplanned care (ORPHAcodes)
- **2022-2025:** OD4RD & OD4RD2: Network of national Orphanet nomenclature hubs in 20 countries: local support for ORPHAcodes implementation and codification
- **2023- 2024**: EHDS Pilot2 project: Orphanet participation to data models and interoperability workpackages (after having participated in TEHDaS)
- **2024 - ongoing:** JARDIN JA (Orphanet leads task 8.2 on semantic harmonization for RD data in European health information systems)

Implementation



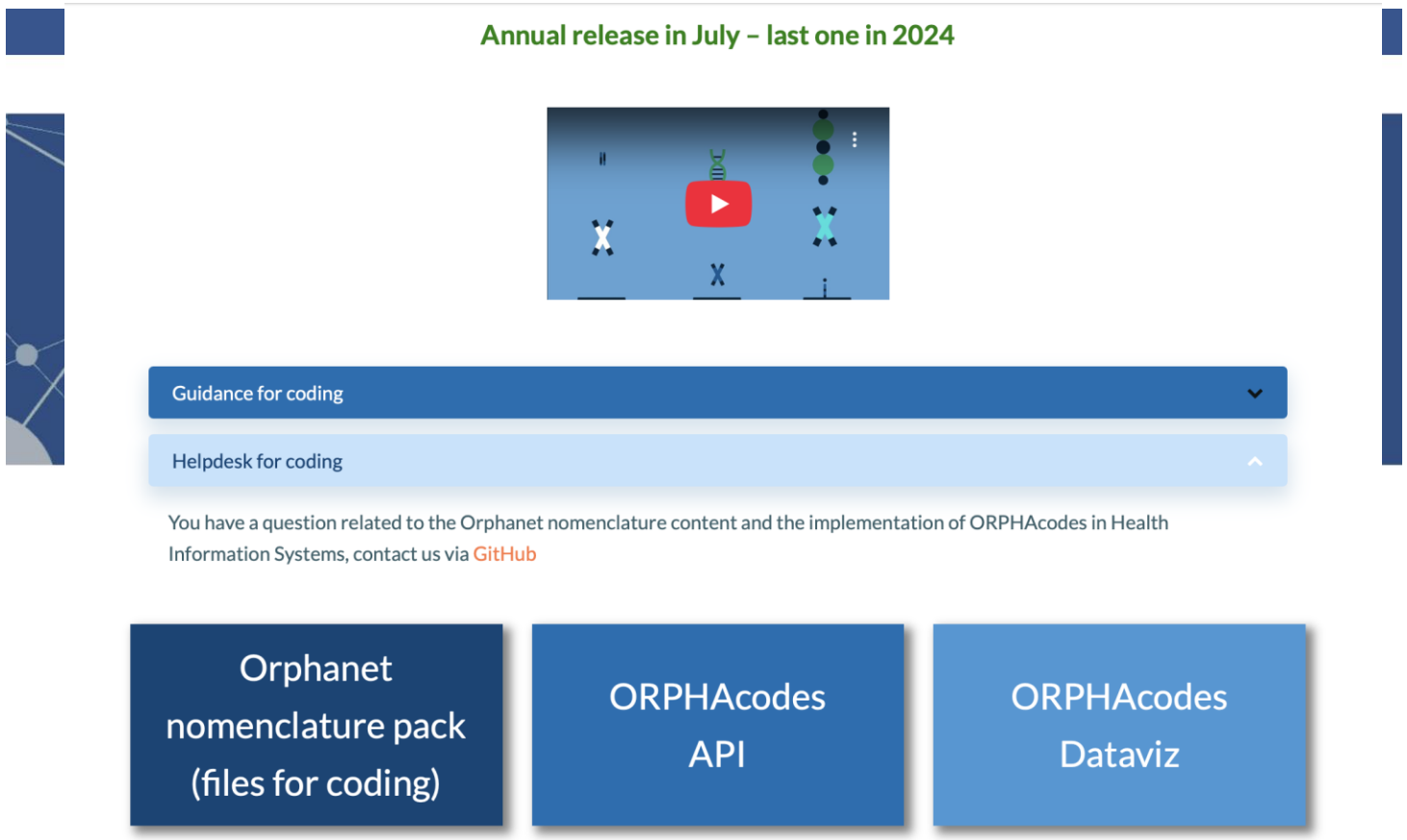
ORPHAcodes National Hubs

- Trainings
- Workshops
- Helpdesks

https://www.orphadata.com/orphanet-nomenclature-for-coding/

WHERE TO START?

https://www.orphacodes.org



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Inserm
La science pour la santé From science to health
Co-funded by the Health Programme of the European Union
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TOOLS

FILES

ORPHACODES

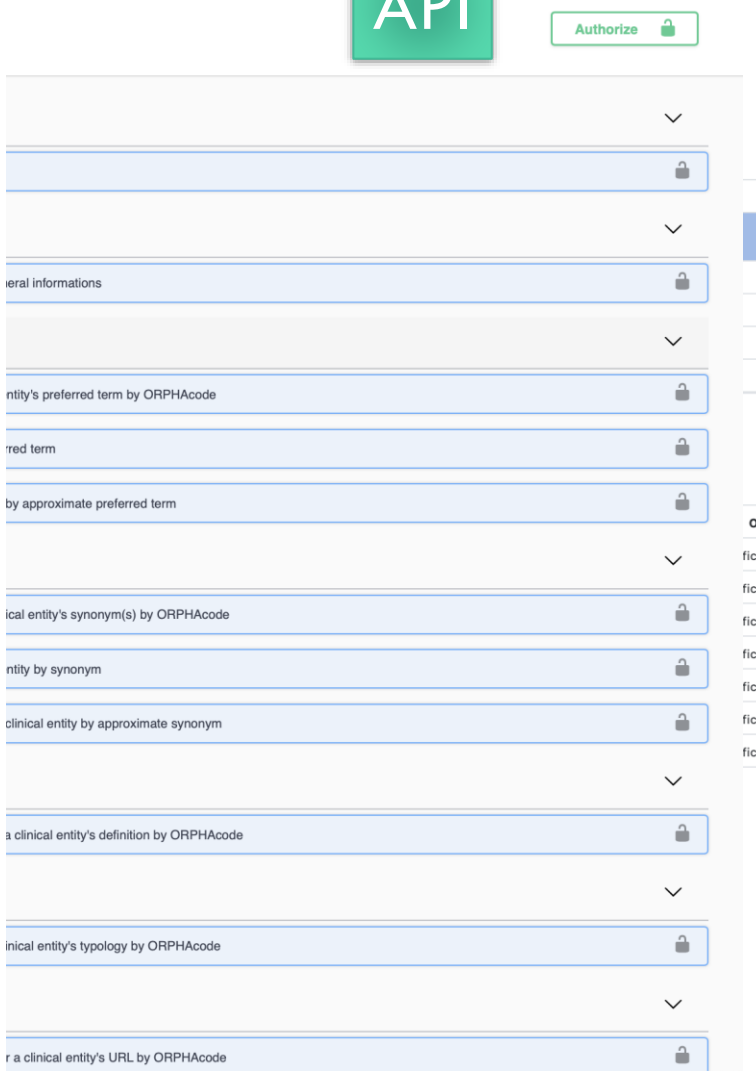
The Orphanet nomenclature pack compiles various files (listed below) which provide the computable information necessary to achieve implementation of ORPHAcodes in health information systems, and ensure easier and accurate coding. These files are updated once a year, in 9 different languages: Czech, Dutch, English, French, German, Italian, Polish, Portuguese and Spanish. Excel and PDF files are available in English and are common to all Orphanet nomenclature pack.

Each year the Orphanet nomenclature pack includes :

- **Orphanet nomenclature file** (XML Schema Definitions and JPEG representations for this file).
- **Orphanet to ICD-10** (XML Schema Definitions and JPEG representations for this files) and **ICD-11 mapping files** (XML Schema Definitions and JPEG).
- **Orphanet classifications** files by medical specialities (XML Schema Definitions and JPEG representations for these files).
- **Linearisation file** attributing one preferential medical specialty to every clinical entity (XML Schema Definitions and JPEG representations for this file).
- **Master file** (Excel file), the minimal set of ORPHAcodes, aligned with ICD-10 codes, that should be used for data sharing and statistical purposes at EU-level. More information about the Master file can be found here.
- **Orphanet nomenclature differential file** (Excel file) which provides the key changes made to the nomenclature compared to the previous version.
- **Description file** (PDF file describing the xml files enclosed in the Orphanet nomenclature pack for coding). More information about the Aggregation level can be found here.
- Recommendations and helpdesk for coding are available below in the dropdown menu.

The nomenclature is also available through dedicated APIs and a human-readable view is provided through Dataviz.

API



DATA VIZ

GUIDELINES FOR IMPLEMENTATION

Guidance for coding

- Description of the Orphanet nomenclature files for coding (PDF)
- Recommendation for routine maintenance of codification resources for rare diseases (PDF)
- Specification and implementation manual of the Master file for statistical reporting with ORPHAcodes (PDF)
- Specifications for an integrated coding application with ORPHAcodes (PDF)
- Know more about ORPHAcodes

https://www.rd-code.eu/wp-content/uploads/2022/01/826607_D5-4_Standard-procedure-and-guide-for-coding-with-Orphacodes_final.pdf

STANDARD PROCEDURES

Guideline 1 - Several tools and strategies could be set at MS level to produce data or statistics for RD, nevertheless each country should set this strategy accordingly to a standard principle of maximizing exhaustiveness as well as possible re-use of existing data collections.

Guideline 6 - If ORPHAcodes are used together with another national coding system for morbidity coding, the two systems should be linked in a standardized way to ensure that code combinations are standardized and the coding effort for the user is minimized.

Maximize exhaustivity:

Use the ORPHAcode alongside other terminologies in use.

Prefer primary ORPHAcoding to secondary transcoding.

Use the reference mappings provided when transcoding

Guideline 2 - Code the data in a way that the reporting can compile to the granularity of the international recommended list of ORPHAcodes (MF-

Disorder level is the statistical aggregation level to count RD patients.

Disorder level codes are compiled in a « master file » (list of active rare disorders codes)

STANDARD PROCEDURES

Guideline 3 - Whenever possible capture the diagnostic assertion for all RD cases. Use the Options "Suspected rare disease", "Confirmed rare disease" and "Undetermined diagnosis". Additional options might be helpful.*

Guideline 5 - Keep track, for each patient file, of the different ORPHAcodes and associated versions that were used to describe the patient's diagnostic pathway.

Providing space for metadata on diagnosis assertion status (and ideally diagnostic history) is useful in the complex process of diagnosing a RD.

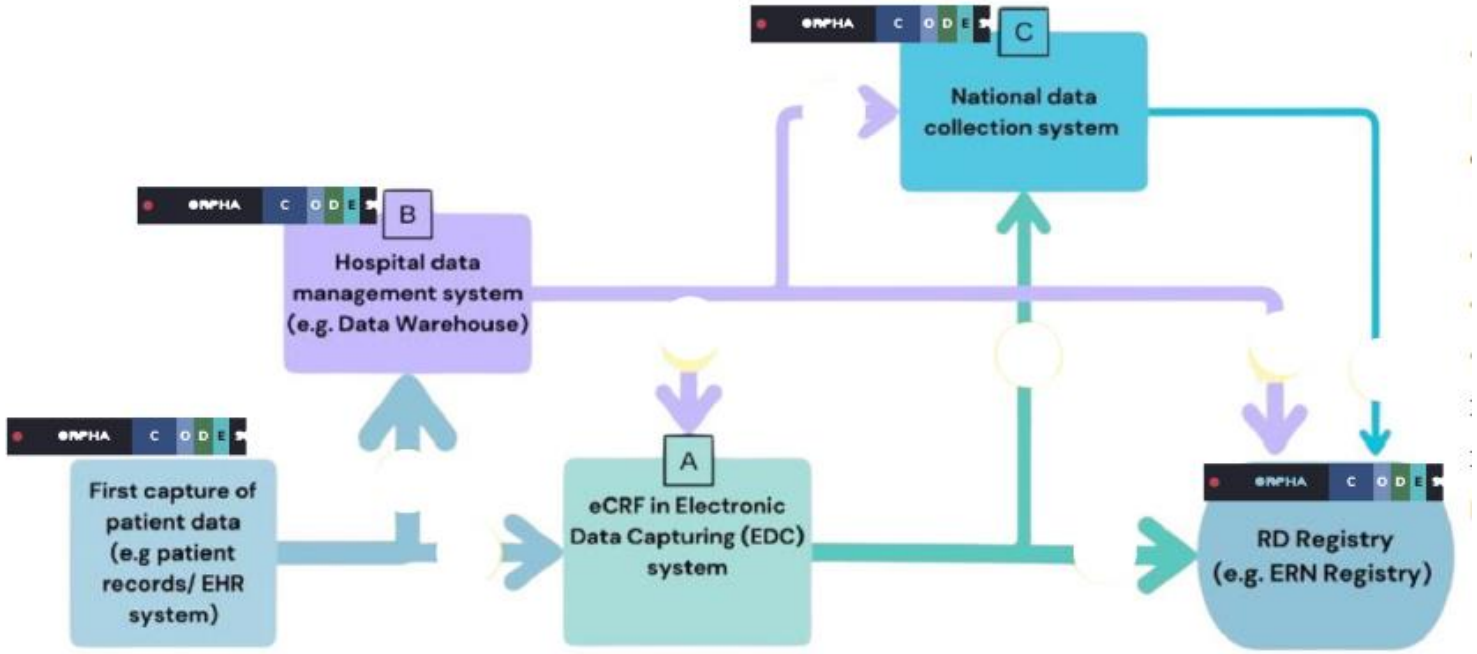
Guideline 4 - Update your coding resource according to the internationally agreed cycle annually in order to have the most recent coding file and to ensure comparability.

ORPHAcodes are released in July every year.

Differential tracking files are provided.

Be aware that ORPHAcodes are created every month that are made available in the next nomenclature version.

DIRECT ORPHACODING



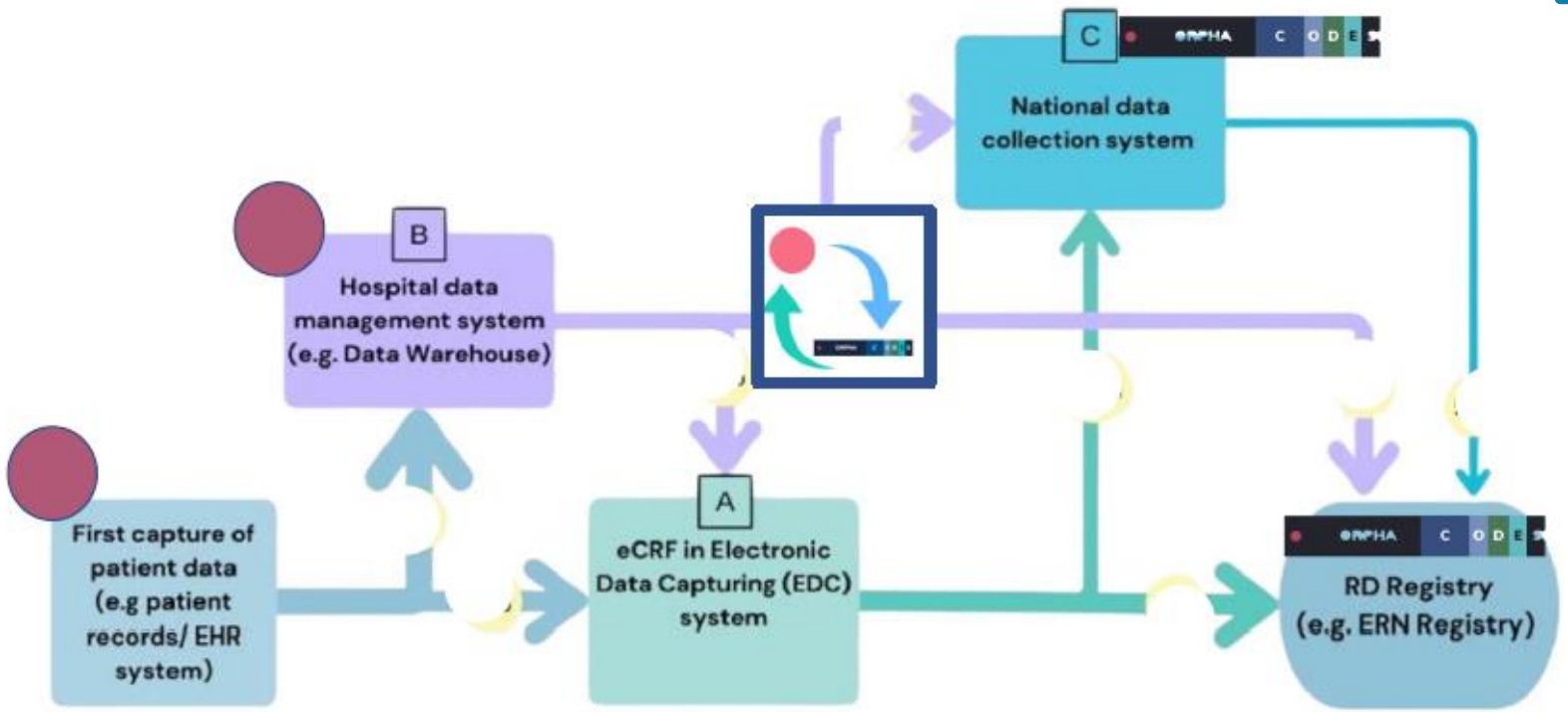
- Possibility to provide electronic support in the health care pathway of the patient thanks to Orphanet Knowledge linked to OC
- **Visibility of all RD diagnosis**
- **Including undiagnosed**

Precise/Accurate

⇒ **Complete epidemiological data**

⇒ **Better patient care, research and management**

TRANSCODING ORPHACODING (A)



Extra burden of transcoding step

- Not all RD diagnosis are traced
- In particular undiagnosed

are not traced

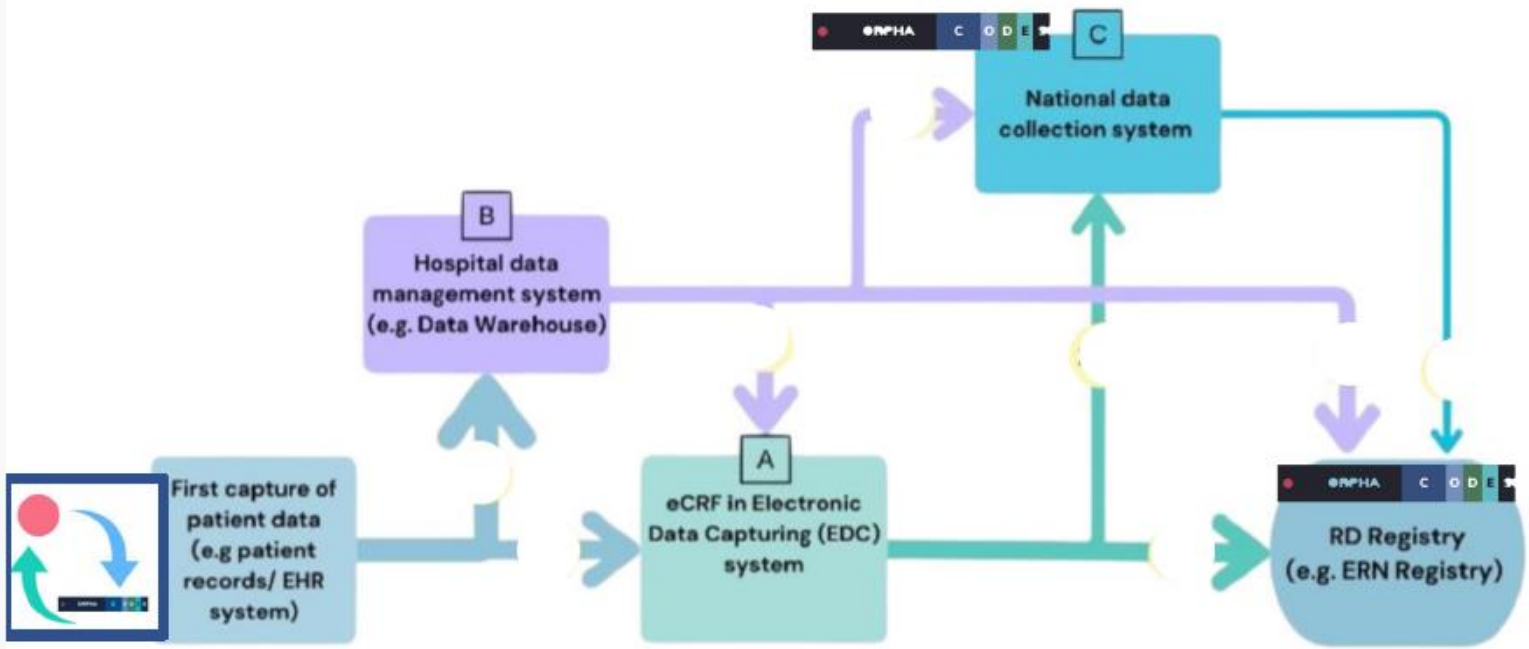
- Imprecise

⇒ Incomplete epidemiological data

⇒ Impaired capacity of Healthcare Systems to address the SPECIFIC/vital NEEDS

⇒ Inadequate Health planning for the non traced diseases

TRANSCODING ORPHACODING (B)



Extra burden of transcoding step

- Not all RD diagnosis are traced
- In particular undiagnosed

are not traced

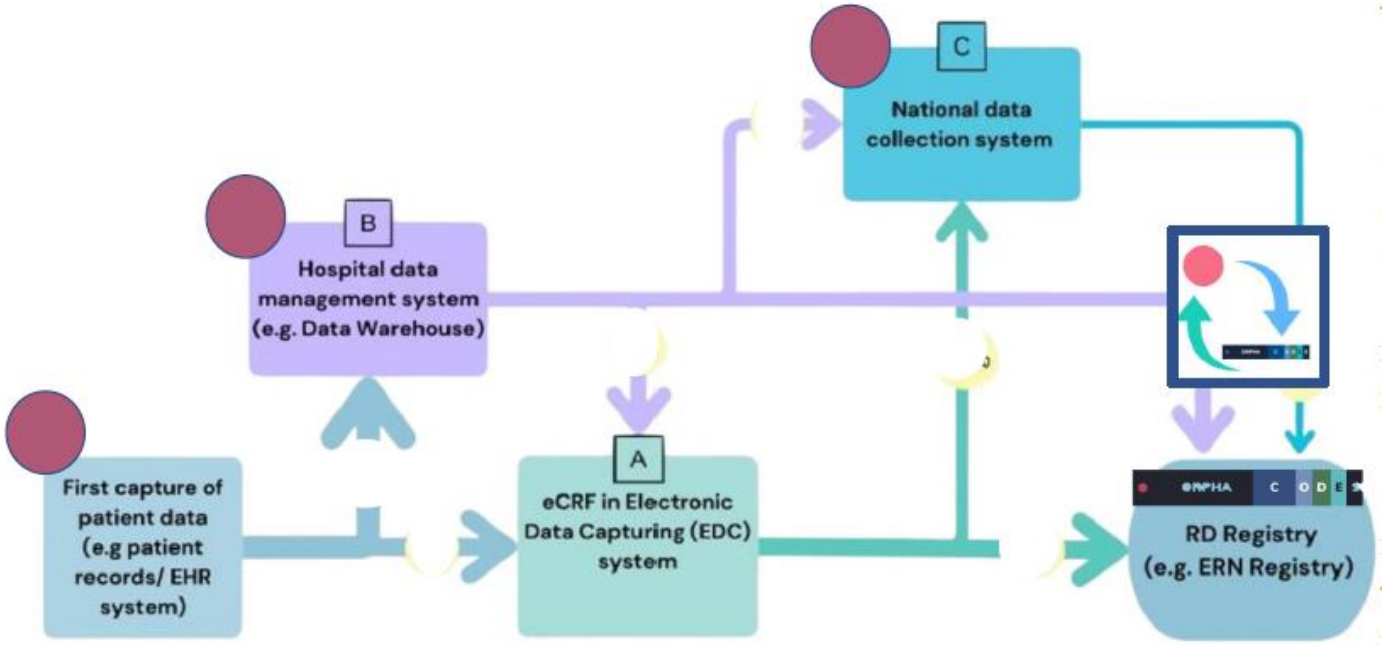
- Imprecise

⇒ Incomplete epidemiological data

⇒ Impaired capacity of Healthcare Systems to address the SPECIFIC/vital NEEDS

⇒ Inadequate Health planning for the non traced diseases

NO ORPHACODING



Extra burden of transcoding step for
⇒ **ERN monitoring/registry**
=> Difficult & time consuming
to produce RD indicators

- Not all RD diagnosis are traced
- In particular undiagnosed

are not traced

- Imprecise

⇒ Incomplete epidemiological data
⇒ Impaired capacity of Healthcare Systems to address the SPECIFIC/vital NEEDS
⇒ Inadequate Health planning for the non traced diseases
⇒ Limited healthcare monitoring in regard to a given RD or a group of RD, or RD as a whole.

https://youtu.be/2intXbSgKAI

OD4RD2
ORPHANET NOMENCLATURE
NATIONAL HUB ACTIVITIES

Brussels, March 19th, 2025

OD4RD
Orphanet Data For Rare Diseases

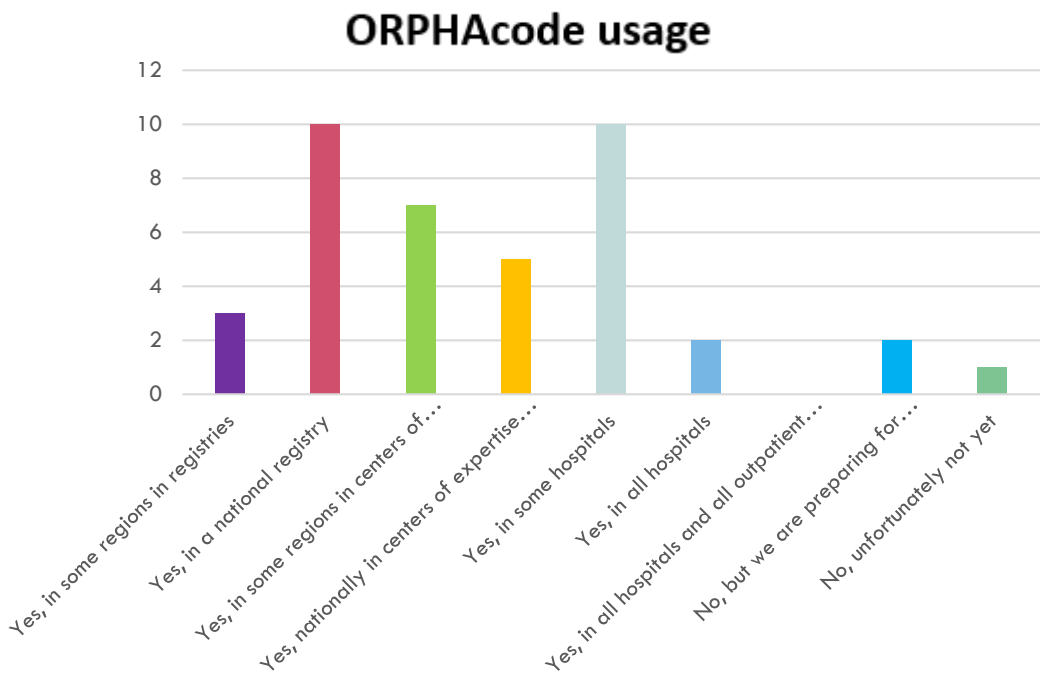
OBJECTIVE
NETWORK OF ORPHANET NOMENCLATURE NATIONAL HUBS

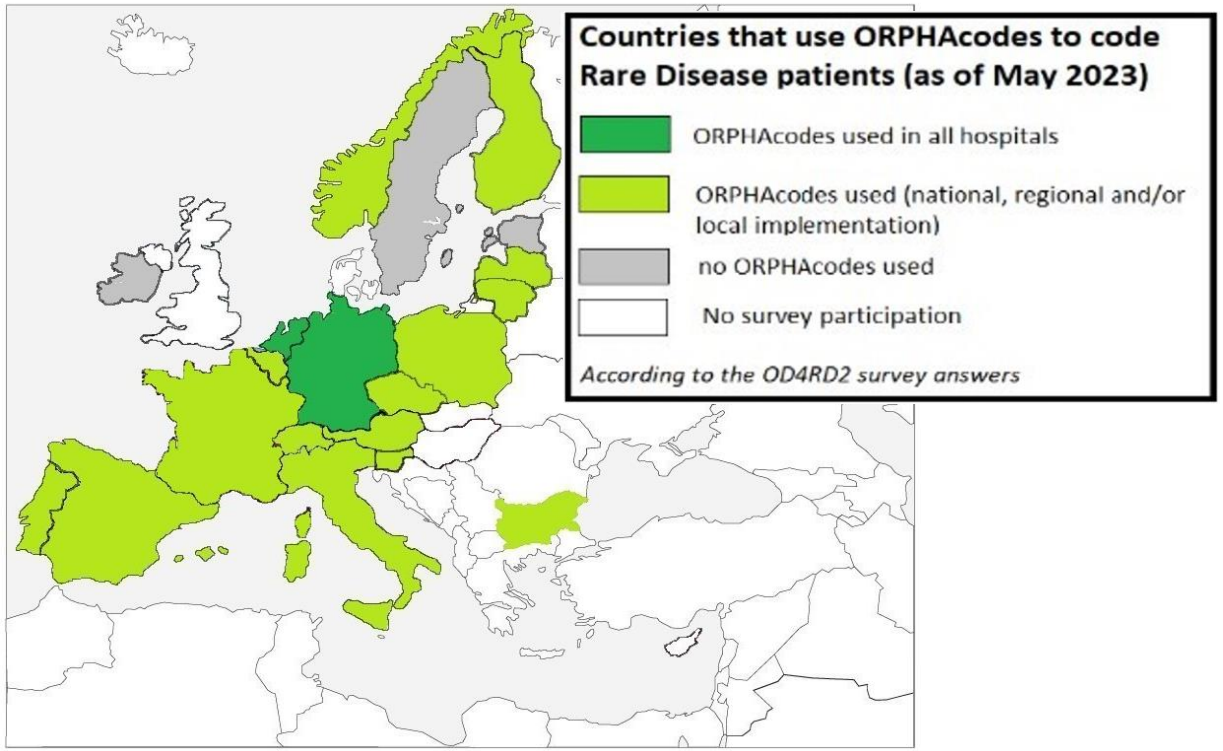
- Provide coordinated support for ORPHAcodes implementation in Health Information Systems of Hospitals hosting ERNs in the participating countries and to harmonise ORPHAcoding practices across countries.

REINFORCE THE NATIONAL LEVEL TO ADD EUROPEAN VALUE

STATE OF PLAY SURVEY 2023

- A survey was launched to the project's National Hubs to assess the state of play regarding ORPHAcodes implementation in HCPs in their country
https://od4rd.eu/03-deliverables/D4.1_OD4RD2_state-of-play-survey_final.pdf





STATE OF PLAY SURVEY 2025

➢ ONGOING WORK : In order to assess an exhaustive State of Play of ORPHAcodes implementation in European countries, complementary approaches have been used to assess the implementation status but also to describe the existing national legal frameworks & real-life implementation cases.

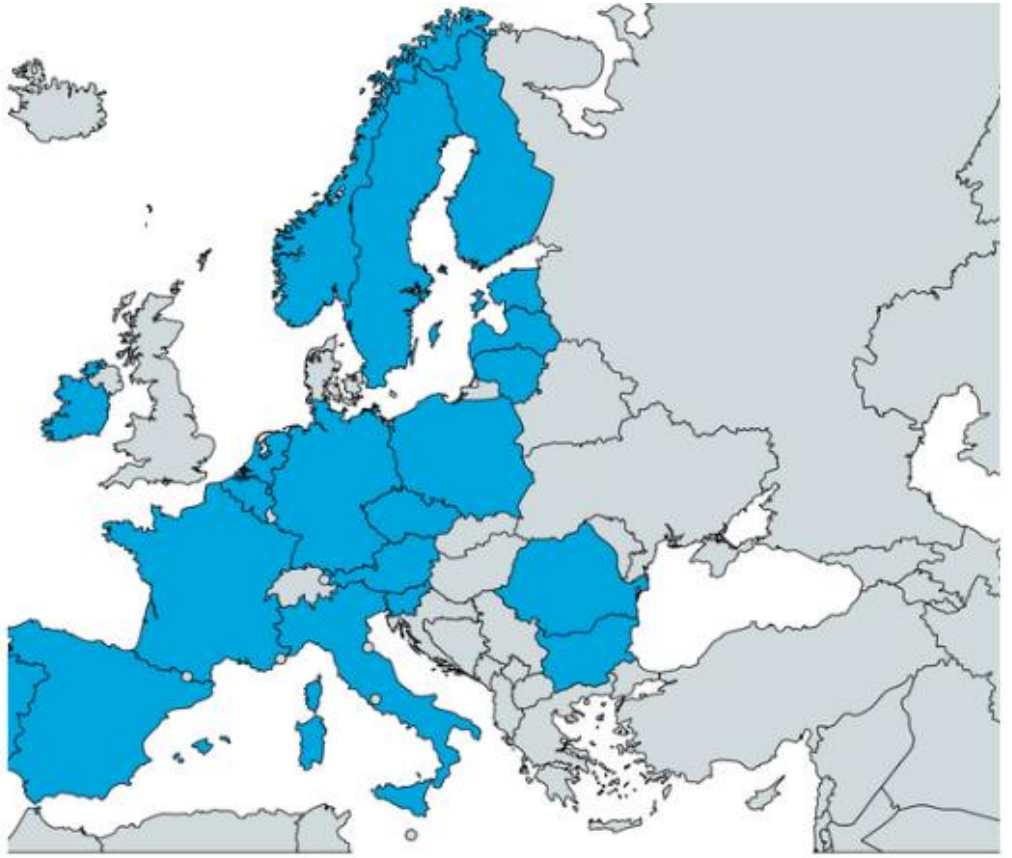
- Feedback from the Orphanet National Nomenclature Hubs
- Feedback from users :

- OD4RD2 ERN survey & JARDIN survey
- Scientific Publications

- This document will be submitted for revision and approval to the MS representatives working group of the JARDIN project during the 19 June Workshop

OD4RD ORPHANET NOMENCLATURE NATIONAL HUBS

- France
- Austria
- Belgium
- Czech Republic
- Germany
- Spain
- Finland
- Italy
- Poland
- Portugal
- Sweden
- Slovenia
- Norway
- The Netherlands
- Bulgaria
- Estonia
- Ireland
- Latvia
- Lithuania
- Romania



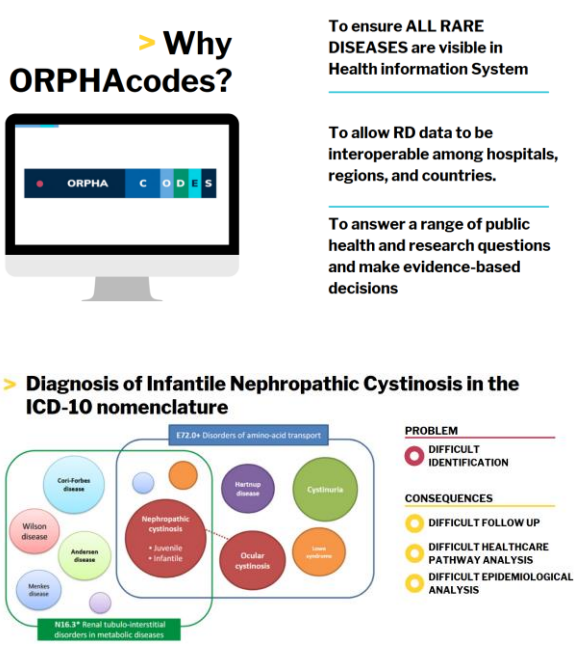
20 COUNTRIES

NATIONAL HUBS

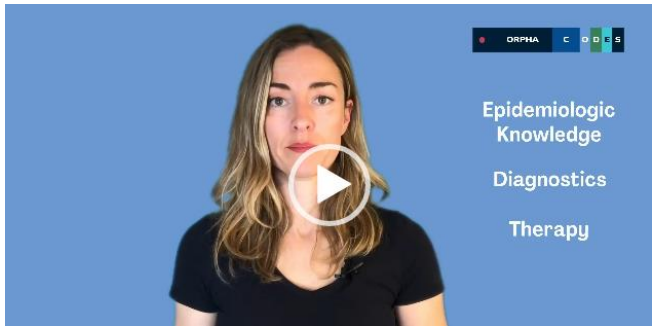
WHAT THEY CAN OFFER:

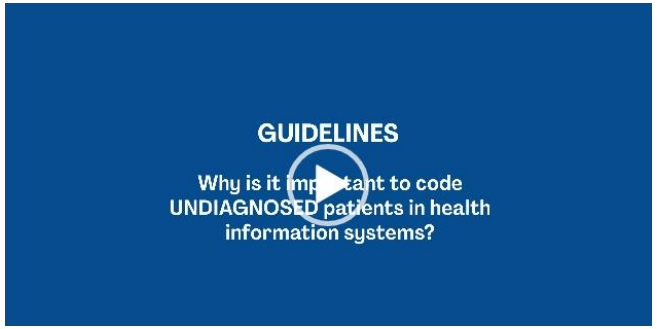
ONLINE TRAINING TOOLS

PRESENTATIONS

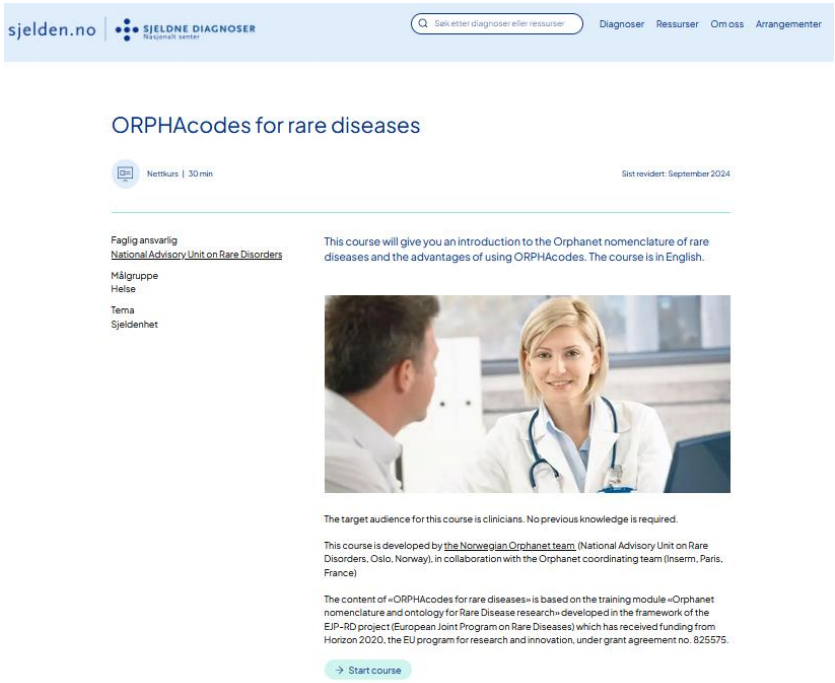


VIDEOS





E-LEARNING



[ORPHA Coding – RD-CODE]

[Orphanet - YouTube]

[ORPHAcodes for rare diseases - Sjelden]

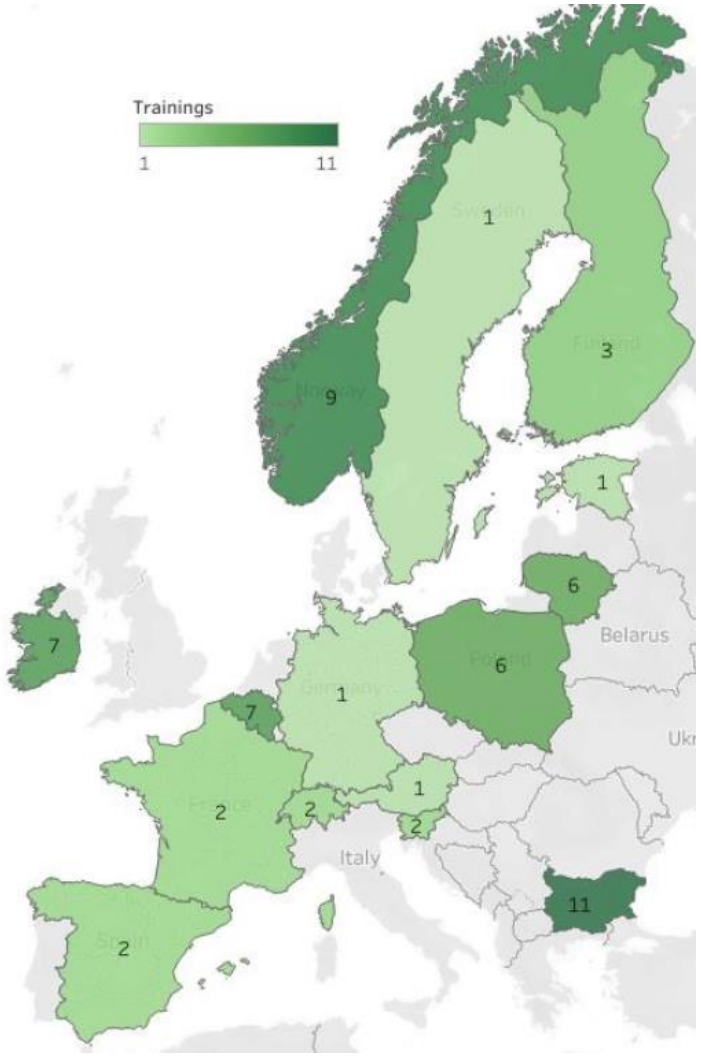
TRAININGS / WORKSHOPS

OD4RD1: >60 Trainings/Workshops

- >1.900 participants

OD4RD2 1st year: 60 Trainings/Workshops

- >1.200 participants



TRAININGS / WORKSHOPS

WHAT

Correct usage of ORPHAcodes

Orphanet Nomenclature

Coding of undiagnosed patients

ORPHAcodes implementation process

RD patient pathways

HOW

Onsite trainings

Distant trainings

Webinars

Onsite Workshops

Distant workshops

To WHOM

Clinicians

Coders

Medical informatics personnel

HOW

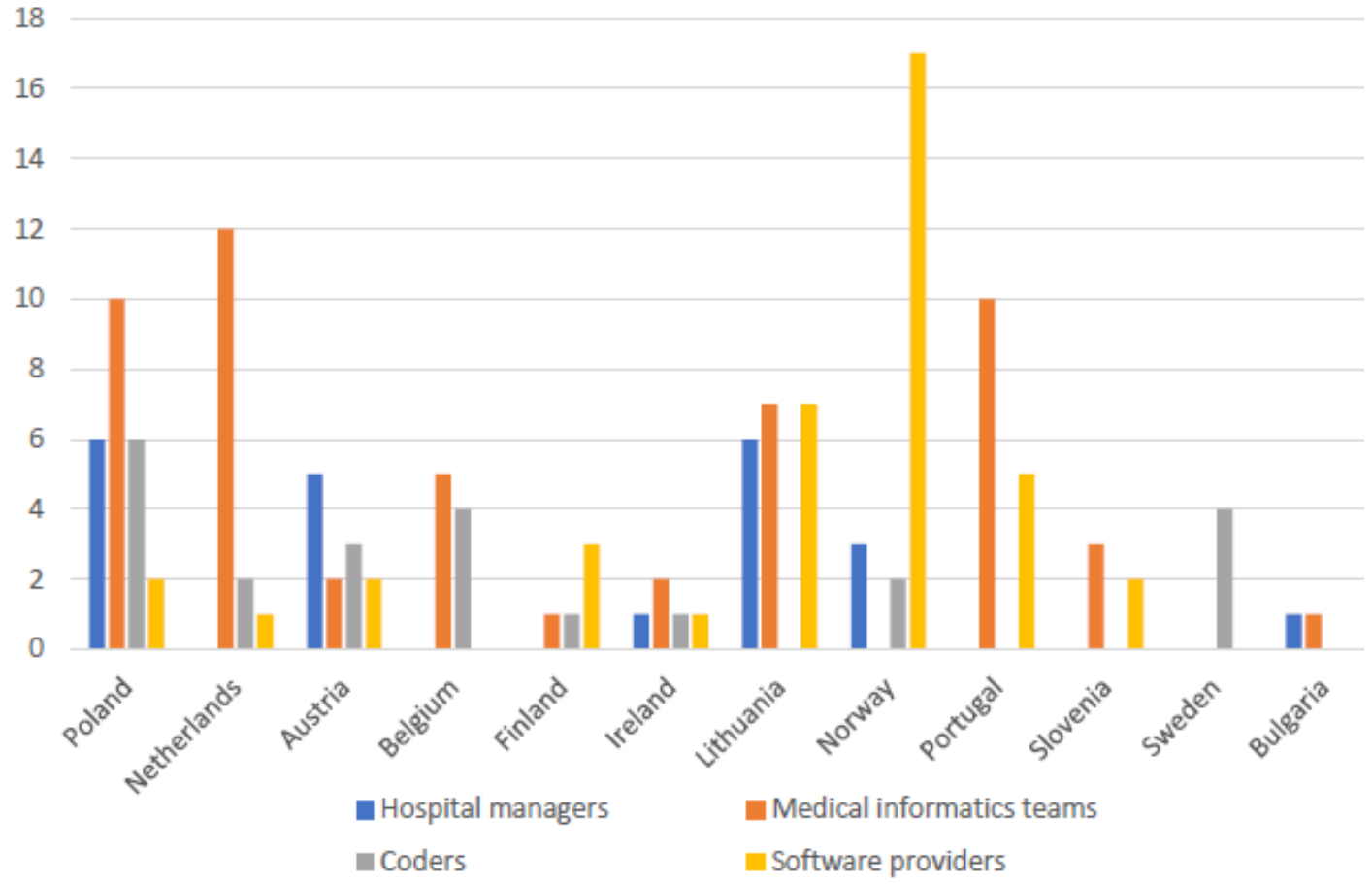
Basic / advanced trainings

Training sessions at national conferences

English

National language

MEETINGS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16
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12
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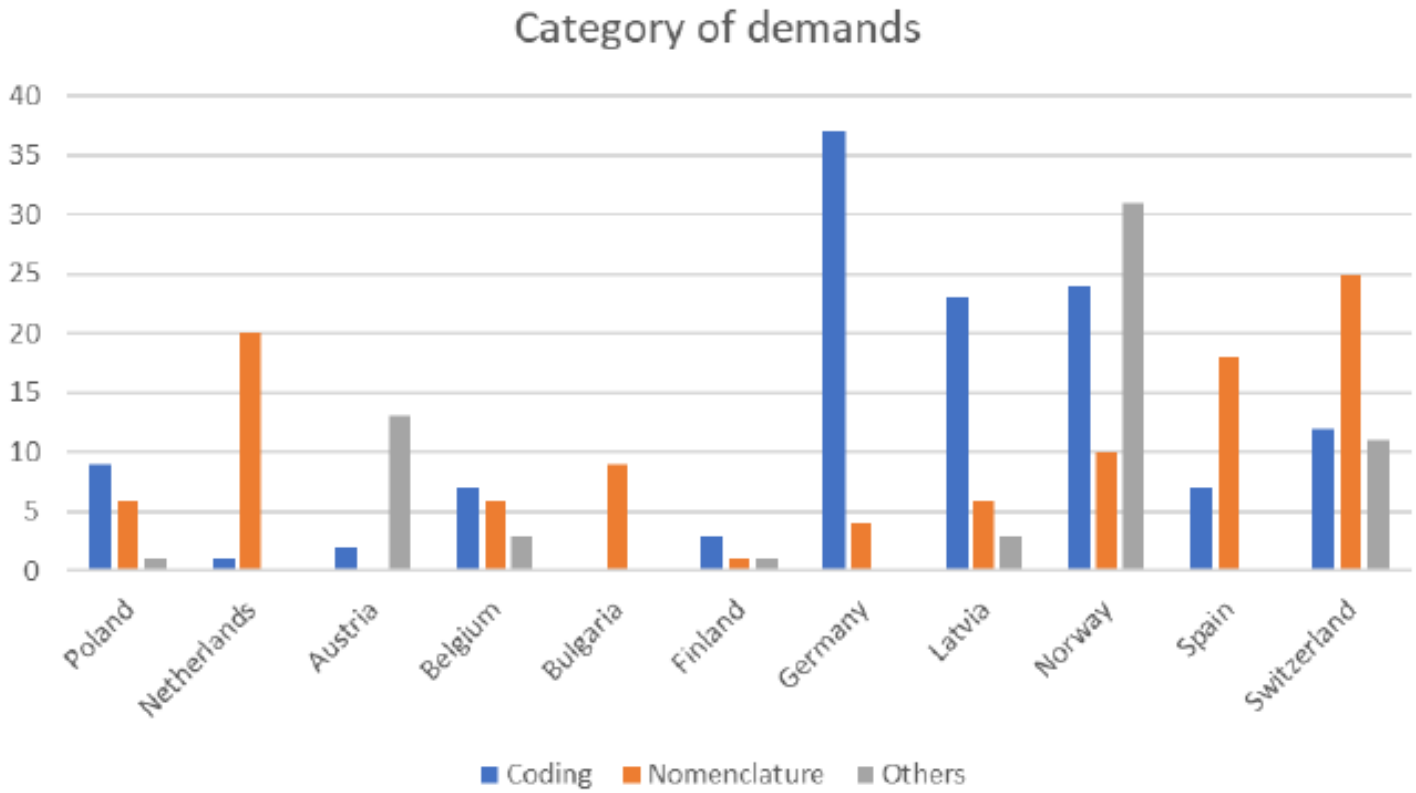
Poland
Netherlands
Austria
Belgium
Finland
Ireland
Lithuania
Norway
Portugal
Slovenia
Sweden
Bulgaria
Hospital managers
Medical informatics teams
Coders
Software providers

NATIONAL HELPDESKS



GITHUB CONTACT WORKFLOW

OD4RD / Main-Help-Desk

User

Other EU countries

orphanet

National Helpdesk

By Orphanet national team, in national language

Wiki

Central Helpdesk

By Orphanet coordinating team, in English

TICKETING TOOL: GITHUB

issues

Filters | is:issue is:open | Labels 40 | Milestones 0 | New issue

16 Open | 45 Closed | Author | Label | Projects | Milestones | Assignee | Sort

- Transthyretin amyloid polyneuropathy: missing ORPHAcode? Belgium Creation Preferred Term update — #68 opened 2 weeks ago by Orphabe — 2
- CAKUTHED - PBX1-associated malformation syndrome Coding issue OrphaNomenclature Switzerland — #64 opened on Apr 27 by OrphanetSwitzerland — 2
- A diagnosis with the word "asymptomatic" in name and definition Clinical signs Norway OrphaNomenclature — #63 opened on Apr 26 by LeneMartinsen — 1
- Sifrim-Hitz-Weiss syndrome Coding issue Cross referencing issue Norway — #62 opened on Apr 26 by LeneMartinsen — 2
- ORPHA:1872 Cone rod dystrophy - add OMIM 617547? Coding issue Cross referencing issue Norway — #61 opened on Apr 26 by LeneMartinsen — 1
- Cleft palate, cardiac defects, and mental retardation - MEIS2 gene Coding issue Creation OrphaNomenclature Switzerland — #58 opened on Apr 17 by OrphanetSwitzerland — 3
- New syndrome associated to variants in the GLDN gene Creation Spain — #53 opened on Mar 7 by Orphanet-ES — 6
- ERKNet orphacodes Coding issue Epidemiology OrphaNomenclature Spain — #50 opened on Feb 21 by Orphanet-ES — 5
- Searching for ORPHAcode in Dataviz Cross referencing issue Spain Tool issue — #48 opened on Jan 24 by Orphanet-ES — 2
- Hypothalamic Hamartoma Coding issue Switzerland — #44 opened on Dec 2, 2022 by MartinArles — 2
- I would like to be referenced for Interstitial Lung Diseases (ORPHA:182095) in Orphanet but also specifically for idiopathic pulmonary fibrosis (ORPHA:2032) and alveolar proteinosis. Is it possible? Belgium Classification issue Coding issue

WIKI/FAQ: GITHUB

OD4RD / Main-Help-Desk

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Home

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sylmaiOR edited this page now · 14 revisions

ORPHA CODES

To provide a sustainable and homogeneous, standardised support to the Rare Diseases community, a 'Questions and Answers' section within GitHub has been developed by the coordinating team based on users' questions and issues. It aimed to provide standardised and generalised answers among 8 main topics:

- ORPHAcodes and Nomenclature,
- Orphanet classification,
- Coding recommendations,
- Epidemiology in RD,
- Alignments with other terminologies,
- Orphanet Tools,
- Education and Communication,
- Orphanet - ERN collaborations
- ORPHAcodes Technical Implementation
- Guidance documents for ORPHAcoding implementation and exploitation

+ Add a custom footer

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- 8. Orphanet-ERN collaborations
- 9. ORPHAcodes Technical Implement...
- Guidance documents for ORPHAcodi...

NATIONAL HUB FLYER

https://od4rd.eu/communication-material

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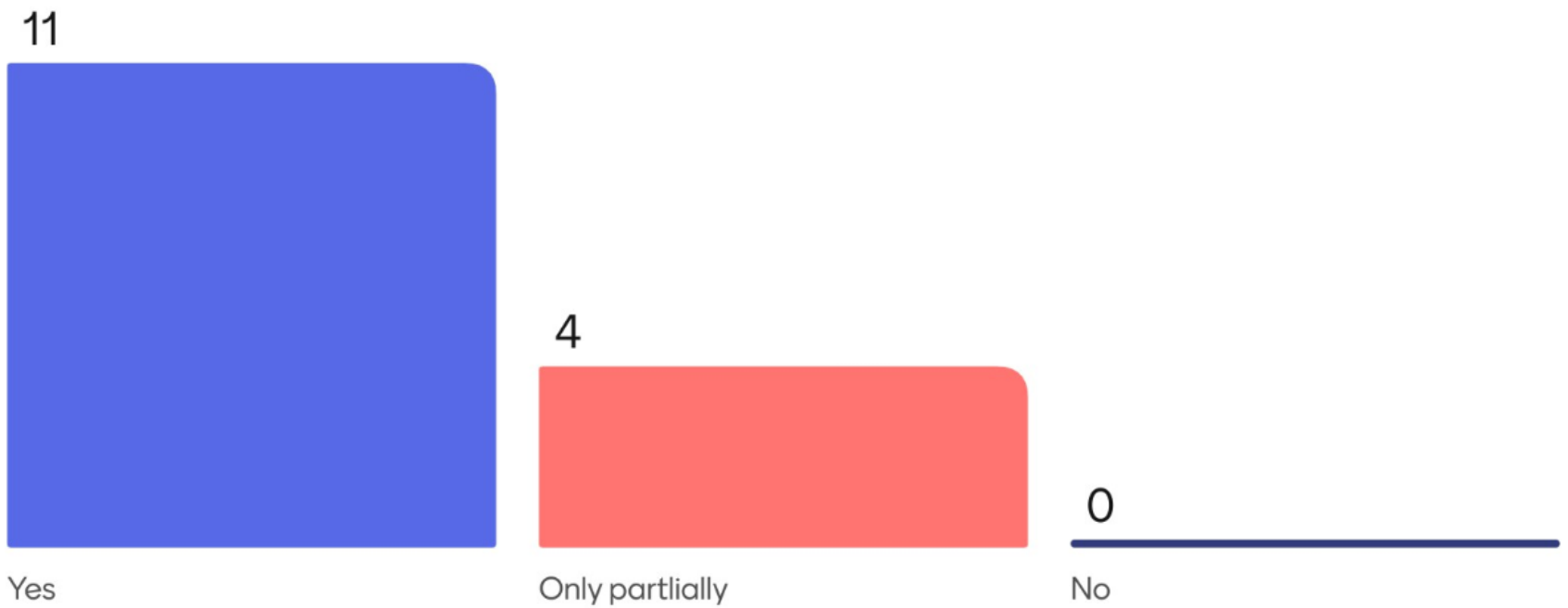
Mentimeter

What about your National Orphanet Nomenclature Hub

Mentimeter

Do the activities presented today cover your needs in terms of ORPHAcoding?



1 15

Mentimeter

If not or partially, what are your ORPHAcoding needs?

national policy in place

Awarness on a national basis

recognition by the national insurrance company

To train countries - as there can occurre some changes in the team

Better correlation with molecular diagnostic

We will be rolling out snomed ct with orpha code mappings and building a new national register. We will need to ensure we integrate and automate as much of this as possible while maintaining quality

National policy in place

Correct diagnostic of rare diseases

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OD4RD

Thank you for your attention!