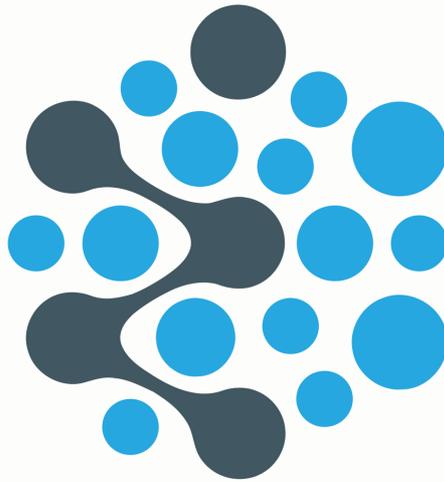


The Orphanet logo features the word "orphanet" in a lowercase, sans-serif font. A blue horizontal line is positioned below the "o", and a blue swoosh underline is positioned below the "n".

orphanet



The OD4RD/OD4RD2 projects tackle the invisibility of rare diseases in European member states' health systems, promotes harmonisation of practice and facilitate generation of standardised interoperable data around RD, thus contributing to meet the ambitions set by RARE2030 concerning data.



OD4RD

Orphanet Data For Rare Diseases

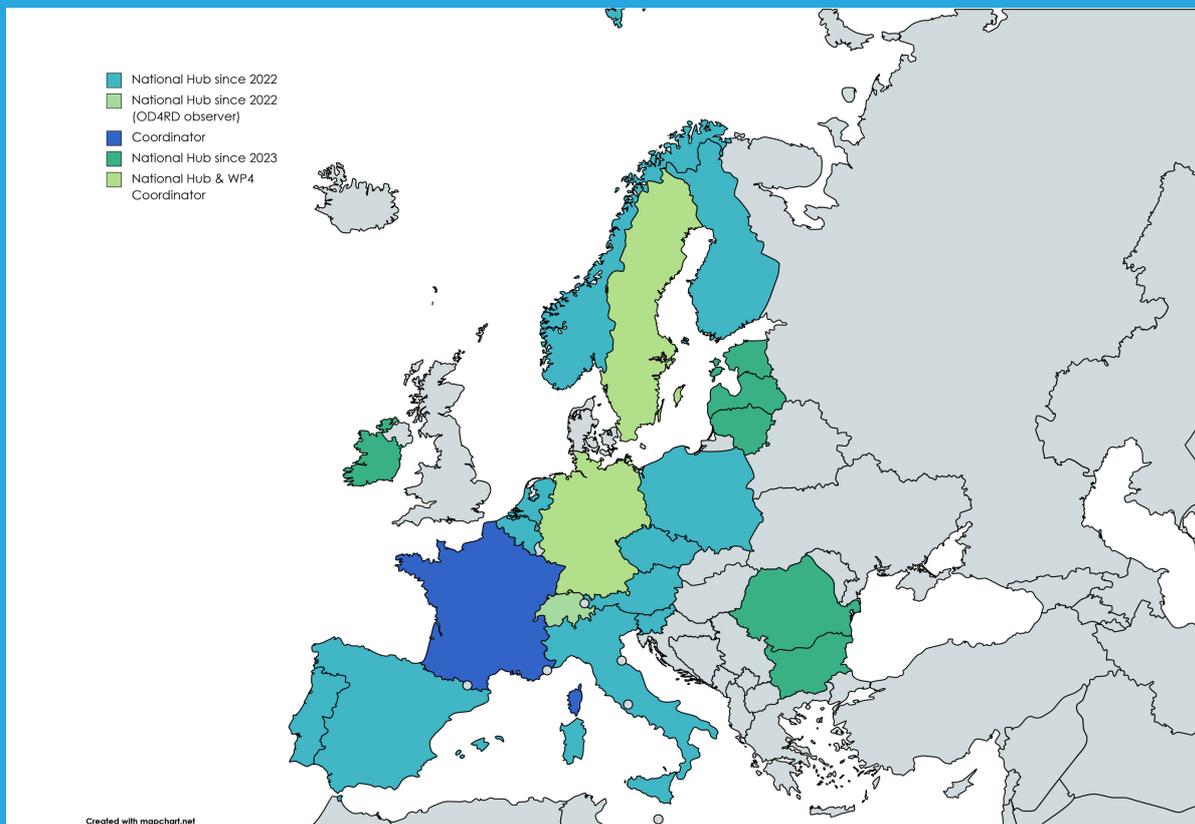
The project produces and maintains the ORPHAcodes, recognised as a Best practice by the European Commission, and makes them available with annotations and transcoding information in a variety of formats that can be adapted to the different codification settings and needs of the different stakeholders

The project provides coordinated support for ORPHAcodes implementation in Health Information Systems of 20 member states' Hospitals hosting ERNs thanks to the growing Network of National Nomenclature Hubs so as to reinforce the national level to add European value

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

PARTNERS

1. **France** – Institut National de la Santé et de la Recherche Médicale (INSERM).
2. **Austria** - Medical University of Vienna (MUW)
3. **Belgium** - Sciensano
4. **Czech Republic** - Charles University Prague and its 2nd Faculty of Medicine
5. **Germany** – BfArM The Federal Institute for Drugs and Medical Devices (BfArM)
6. **Spain** - CIBER
7. **Finland**- Kehitysvammaisten Tukiliitto (Norio Centre)
8. **Italy** – IRCCS Ospedale Pediatrico Bambino Gesù (OPBG)
9. **Poland** - The Children’s Memorial Health Institute (CMHI)
10. **Portugal** - Directorate-General of Health (DGS)
11. **Sweden** - Karolinska University Hospital (KS)
12. **Slovenia** - University Medical Centre Ljubljana (UMCL)
13. **Norway**- Oslo University Hospital- OUS-BAR
14. **The Netherlands**- Stichting Radboud universitair medisch centrum
15. **Estonia** – University of Tartu (UTartu)
16. **Lithuania** – Vilnius University Hospital Santaros Klinikos (VUH)
17. **Romania** – University of Medicine and Pharmacy GR.T.Popa Iasi Orphanet center (*starting 2024*)
18. **Ireland**- Health Service Executive (HSE)
19. **Bulgaria** – Bulgarian association for promotion of education and science/Rare disease institute (BAPES)
20. **Latvia** – Center of Rare Diseases (BKUS) (*starting 2024*)
21. **Switzerland**- University Hospital of Geneva (HUG) (Observer)



The coordinated Network of Orphanet Nomenclature National Hubs

The project produces and maintains the ORPHAcodes, recognised as a Best practice by the European Commission, and makes them available with annotations and transcoding information in a variety of formats that can be adapted to the different codification settings and needs of the different stakeholders

**6300+
RD**

Orphanet
Classification
ongoing
revisions with
the ERN
to meet coder
needs

Aligned with
ICD10, ICD11,
SNOMED-CT &
OMIM
to facilitate
transcoding

With definitions
& genetic
annotations to
facilitate coding
choice

Available in
different formats
adaptable to the
different
codification
settings

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

All info available on www.orpha.net & www.orphadata.com

The project also provides coordinated support for ORPHAcodes implementation in Health Information Systems of 20 member states' Hospitals hosting ERNs thanks to the growing Network of National Nomenclature Hubs



Assess users' needs
at national level



Organise capacity
building
around the
Orphanet
Nomenclature &
tools



Support & advice
on
implementation
& ORPHAcodes use

Coordinated approach to ensure mutualisation & build capacity of the network

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

Preliminary report on analysis of disease coverage by ERN available

2023 ACTIVITIES

.....

- **Nomenclature Helpdesk Operational in 20 countries**
- **New content of the Nomenclature Pack**

with 6,300 disorders of which 96% with a definition and improved ICD11 alignment files with +4,000 new alignments with MMS weblinks and URI

- **SNOMED CT-Orphanet nomenclature map available:**

under a collaboration agreement between the INSERM/Orphanet & SNOMED International. Access to the human readable map @ orphadata & RF2 version from SNOMED's Member Licensing and Distribution Service.

- **State of play of ORPHAcoding in 20 countries**
- **Ongoing ORPHAcoding trainings in ERNs centres across the Network**
- **follow @orphacodes (twitter & Instagram)**

WHAT'S NEXT

.....

- **Survey of coding habits in ERNs centres from Hospitals in 20 countries**
- **Organisation of dedicated events for ERNs and Hospital Managers: stay tuned!**
- **Release of the updated Nomenclature Pack in July 2024 with new features to fit users needs!**
- **Analysis of ERNs RD coverage, complementarities & gaps**

FURTHER INFO

 www.od4rd.eu  pmt.orphanet@inserm.fr  [@orphacodes](https://www.instagram.com/orphacodes)

 <https://github.com/OD4RD/Main-Help-Desk> @ [GitHub country alias]  [www.orphadata.com/
orphanet-nomenclature-for-coding](http://www.orphadata.com/orphanet-nomenclature-for-coding)

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**KNOW
THE
RARE**
FOR
BETTER
CARE



OD4RD
Orphanet Data For Rare Diseases