OD4RD
Orphanet Data for rare Diseases

Orphanet Data For Rare Disease

OD4RD

OD4RD&OD4RD2 Projects Progress Report
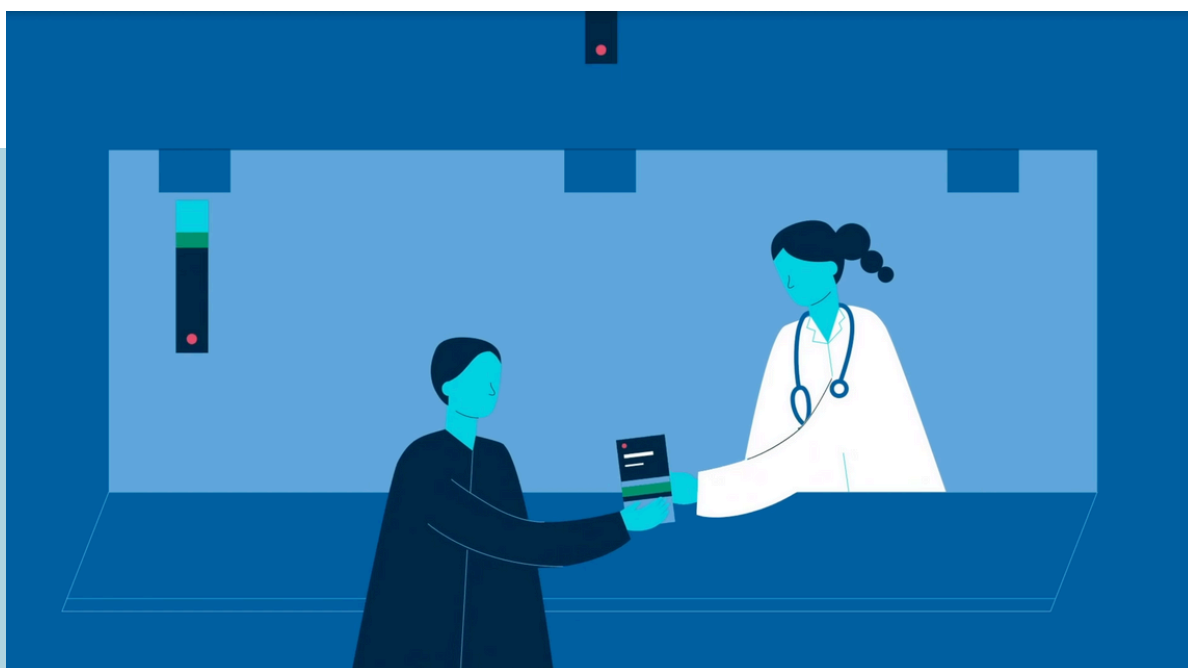
ORPHA **CODES**

ABOUT US

The ORPHANET DATA FOR RARE DISEASE-OD4RD 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

ORPHAcodes production & support

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 also provides coordinated support for ORPHAcodes implementation in Health Information Systems of 19 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
Finally we also provide evidence to support ERN coordination, BoMS and the EC's ERNs strategy and decision making

* https://health.ec.europa.eu/rare-diseases-and-european-reference-networks/rare-diseases_en
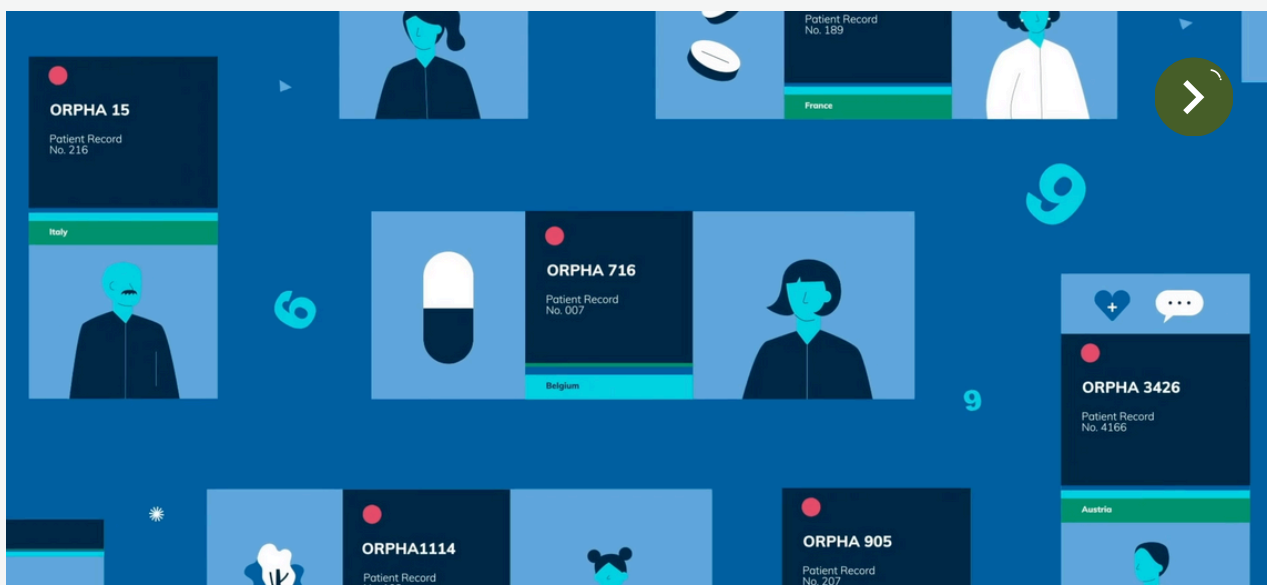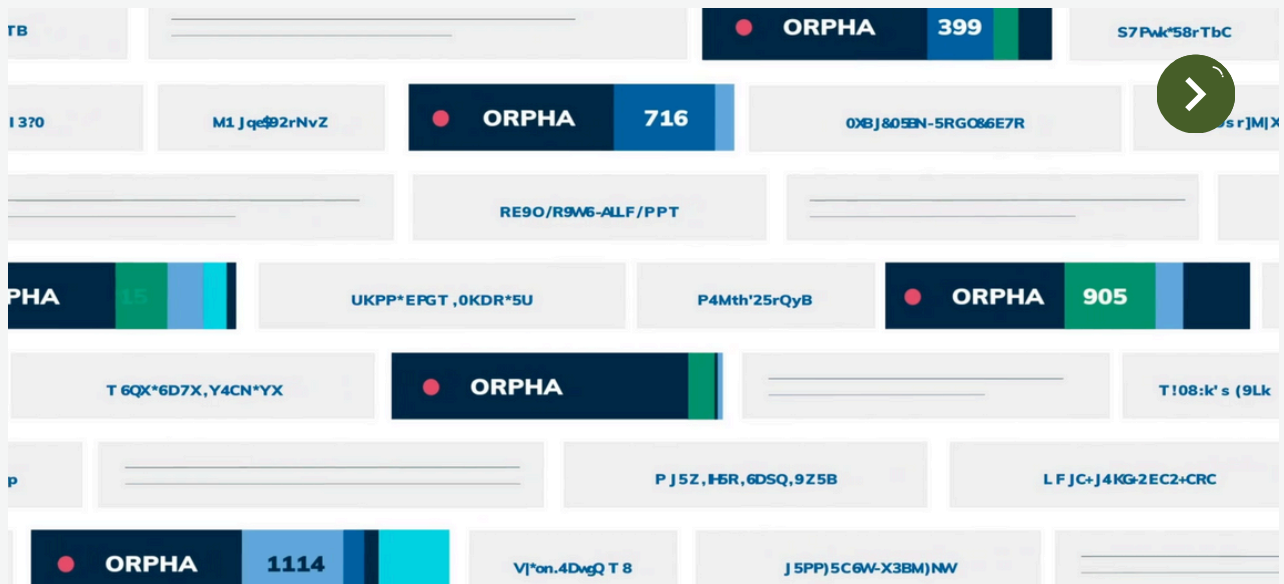
ABOUT OUR PROJECT

The OD4RD2 project is a 3-year project that builds on, and expands, the achievements of the previous OD4RD project, the specific expertise of Orphanet and its organisation as a long-lasting and well-established network, to fulfil the following 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, 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

OUR OBJECTIVES

ORPHA 399 · S7Pwk*58rTbC

M1 Jqe492rNvZ · ORPHA 716 · 0XBJ805BN-5RGO86E7R

RE9O/R9W6-ALLF/PPT

PHA · UKPP*EPGT ,0KDR*5U · P4Mth'25rQyB · ORPHA 905

T 6QX*6D7X,Y4CN*YX · ORPHA · T!08:k' s (9Lk

P J5Z,H5R,6DSQ,9Z5B · L F JC+J4KG-2EC2+CRC

ORPHA 1114 · V|*on.4DvgQ T 8 · J5PP)5C6W-X3BM)NW

OUR GOALS

- Increase the visibility of RD in Health Information Systems by supporting ORPHAcodes implementation in hospitals
- Increase the quality of data generated about RD patients by disseminating ORPHAcoding good-practices & guidelines;
- 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, both for primary use (by ensuring a better diagnosis and care of RD patients, and facilitating the assessment of current practices and results against gold standards of care) and for secondary use (by informing policy decision making and research).

IN A NUTSHELL

"Implementation of ORPHAcodes at the data source is essential : OD4RD2 provides support to facilitate this , the objective is to obtain RD exploitable data at the Member States (MS) level in order to be able to also understand the situation at the EU level"
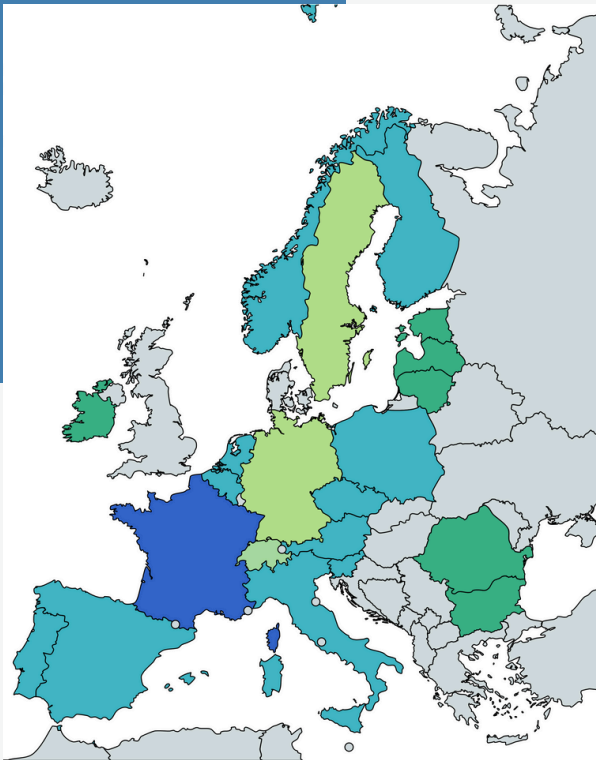
We want to build knowledge around ORPHAcoding to make RD visible in HIS and facilitate Evidence based decisions

The project provides coordinated support for ORPHAcodes implementation in Health Information Systems of 19 member states' hospitals hosting ERNs thanks to the growing Network of National Nomenclature Hubs, to harmonise ORPHAcoding practices across countries so as to reinforce the national level to add European value

PROGRESS OF OUR PROJECT

The Hubs make available a **National Helpdesk** and organise **trainings and events around ORPHAcodin**g to build capacity so that information is collected correctly and uniformly. The Network of Hubs meets regularly to mutualise the success stories and address common problems in a coordinated manner.

Capacity building events organised according to national needs since 2022
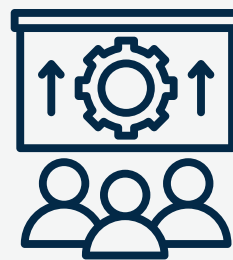
110 Trainings in National Language
2850+ participants
incl. 236 ERN Units
230 Ad Hoc Meetings

Support & advice on implementation & ORPHAcodes
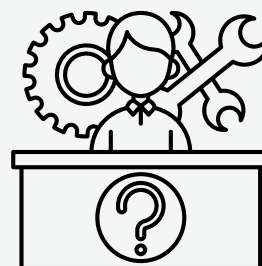
19 active National Helpdesk in National Language + a central helpdesk available at github

The operations around organisation of capacity building and support and advice are of satisfaction and match project goals, our next goal is to increase ERN attendance to national trainings
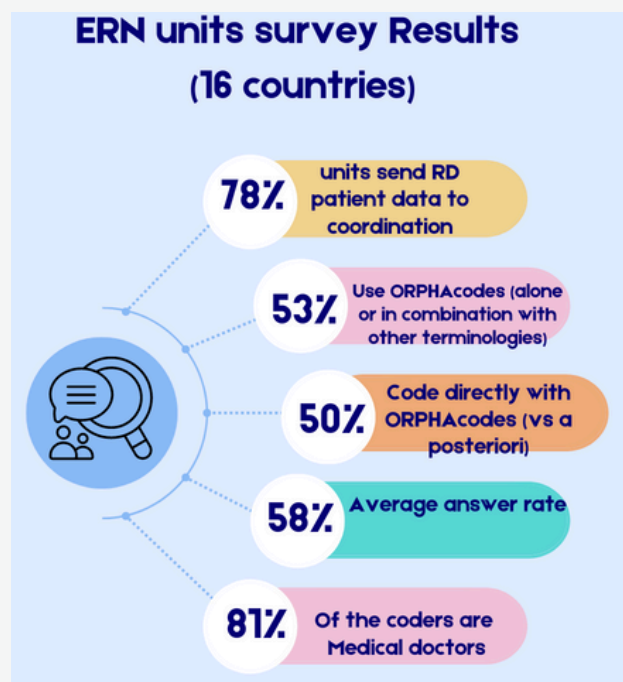
We want to assess users needs at national and ERN level

A Survey to evaluate the current situation of ORPHAcodes usage by health care providers (HCPs) linked
to ERNs in countries participating in OD4RD2 has been carried out, in order to be able to identify
ORPHAcoding practices and barriers in implementation to be able to fine tune both National Actions
but also transversal actions at the Coordinating team level within OD4RD & JARDIN

PROGRESS OF OUR PROJECT

A survey was sent to email contacts from each of the National ERN centres listed in the Orphanet Data Base of expert ressources ,
=> more than 500 units answers received in the 16 countries having launched the survey *.

ERN units survey Results (16 countries)

78% units send RD patient data to coordination

53% Use ORPHAcodes (alone or in combination with other terminologies)

50% Code directly with ORPHAcodes (vs a posteriori)

58% Average answer rate

81% Of the coders are Medical doctors

These results will be complemented with the JARDIN Joint Action survey ones, to obtain a complete picture of practices by ERN and across countries and identify barriers and needs. Results will be discussed with the ERNs during a dedicated day to raise awareness about ORPHAcoding benefits and to discuss around ERN data strategy so as to refine the project's national and transversal actions to improve harmonisation.

*https://od4rd.eu/03-deliverables/OD4RD2_ERN-Survey_11QC%20analysis_VF.pdf

We want to make sure that both PLWRD needs & the coders needs are met

The project supports the production of the ORPHAcodes, the only terminology that makes visible **ALL 6000+ RD** in Health Information Systems (HIS), recognised as a Best practice by the European Commission *.

ORPHAcodes update is carried out in collaboration with European Reference Networks (ERNs) to fit coders' needs**. ORPHAcodes are made available with annotations, definitions and transcoding information to facilitate the coding work and reduce its burden.

PROGRESS OF OUR PROJECT
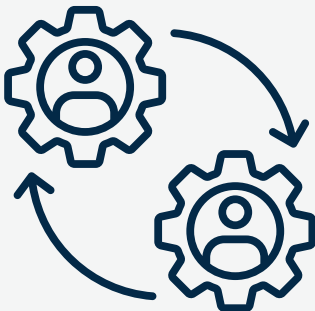
- 6300+ RD in the nomenclature delivered with a definition to facilitate the coders choice provide an accurate and comprehensive representation of knowledge.
- Updates of the gene-diseases relationships database are also input whenever relevant to further help the clinicians
- The work is carried out in collaboration with European Reference Networks: with 8 Nomenclature revision collaborations already delivered so far & 12 ongoing *

Exploitation of ORPHAcodes' annotated Health data increases the visibility of people living with a RD**** and contributes to ensure that needs of all RD Patients are met.

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* https://health.ec.europa.eu/rare-diseases-and-european-reference-networks/rare-diseases_en

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

eproc_Collaboration_networks_R1_Nom_Rev_Exp_EP_10.pdf

*** https://github.com/OD4RD/Main-Help-Desk/wiki/8.-Orphanet%E2%80%90ERN-collaborations

**** https://od4rd.eu/orphacodes-bibliography
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FACILITATED TRANSCODING

Alignment of ORPHAcodes with other terminologies in use in health systems and in registries is carried out by Orphanet to facilitate transcoding & reduce coding burden

99 %
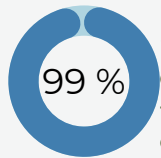
This process implies the identification by Orphanet of the best suitable ICD-10 code and the definition of a semantic relationship between both codes

% ORPHAcodes-ICD10 alignments

66 %
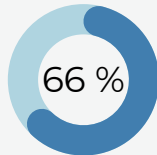
This process implies the identification by Orphanet of the best suitable ICD-11 code and the definition of a semantic relationship between both codes

% ORPHAcodes-ICD11 alignments

95 %
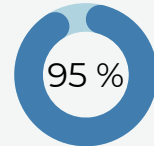
in the frame of the collaboration agreement Orphanet releases a human-readable file with the updated mappings while SNOMED-CT releases the machine readdable file

% ORPHAcodes-SNOMED CT alignments

Transcoding information produced by Orphanet aims at providing suggestions for the coders and automatise the correspondance between ORPHAcodes & the main medical terminology in use, this info is available in various machine readable formats easily adapted to the different HIS to decrease coding burden

TECHNICAL USERS NEEDS ASSESSMENT

We provide various technical solutions * & in December 2024 a workshop was organised to collect feedbacks from stakeholders in order to identify and prioritize possible technical improvements for the future releases of the tools in order to comply with the different settings and users' needs as much as possible

*www.orphadata.com/orphanet-nomenclature-for-coding/

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ORPHA C O D E S