OD4RD Final Meeting Report

October 2022

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Introduction

The Final OD4RD meeting was a hybrid event organised by the Orphanet Network Coordinating team (INSERM), it was held in Paris and online on the 6th October 2022. It gathered 83 participants including all project partners as well as ERNs and other stakeholders. The objectives of this meeting were multiple: allowing ERNs and the external stakeholders to learn about the project in general, and its current and planned outcomes, but also and importantly to discuss on potential improvements, additional tasks and challenges to be addressed in the next months of the project and in its next phase (OD4RD2 project which is currently under evaluation).

Report and slides can be accessed here

Participants

Orphanet teams: Austria*, Belgium, Czech Republic, Germany*, Finland, France, Ireland*, Italy, Kazakhstan, Luxembourg*, North Macedonia, Netherlands, Norway, Portugal, Poland, Romania*,Serbia*, Slovenia*, Sweden*, Switzerland, Spain* and Turkey*

ERNs: RITA, eUROGEN, Transplantchild, EUROBLOODNET, ERN-RND*, ERN EYE*, GENTURIS*, CRANIO*, RARE LIVER*, ENDO ERN*, ITHACA*, ERNICA*

Other projects: ERICA*

Other stakeholders: French Ministry of Health

N.B. The asterisk Indicates online attendance

Opening

The Orphanet Data for Rare Disease project (www.od4rd.eu) started on the first January 2022. It is a 15 months long project that builds on Orphanet's specific expertise, and on its organisation as a long-lasting, well-established network, to fulfil the following general objectives:

1. To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, through maintenance of the Orphanet nomenclature of RD in collaboration with ERNs, and active support for its implementation in hospitals hosting ERNs,

2. To contribute to the harmonisation of data collection amongst various settings (health records, registries) and amongst countries, through dissemination of coding good practices at the source (health records, registries, etc)

3. To support evidence-based decision-making in the frame of the European strategy around ERNs, by providing an exploitable reference corpus of data and information on RD.

In particular the project aims at:

- Increase the visibility of RD in Health Information Systems by supporting real implementation in hospitals
- Increase the quality of data generated about RD patients by disseminating good coding practice
- 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 the EU health data strategy, by connecting the dots with structuring initiatives around EHR formats and health data spaces (EHDS). Both for primary use: better diagnosis and care of RD patients, assessment of current practices and results against gold standards of care, and for secondary use: informing policy decision-making and research

For these endeavors to be successful we need first and foremost to provide a nomenclature that fits coding needs and allows for transcoding in Health Information Systems. But also, well-trained National Nomenclature hubs, in capacity to support local implementation of ORPHAcodes. And in parallel we also need very good connections at decisional level such as Ministries of Health and Hospital managers and with the ERN coordination to ensure facilitated local level contacts and interactions. Finally, it is of the upmost importance to reach awareness and obtain support from ERNs governing bodies such as the ERN Coordination, the Board of Member States and the European Commission.

By helping implementing ORPHAcodes in hospitals, this project has the ambition to contribute to the health data strategy, by connecting the dots with other health data related projects in which Orphanet is participating, such as X-eHealth, TEHDaS joint action, the EHDS Pilot 2 project and the European Joint Co-fund Program on rare diseases (EJP RD) in which a federated, virtual platform of data and resources for research is being created. This of course will benefit the ERN registries strategy, and the EU RD Platform at the Joint Research Centre. ORPHAcodes are the *sine qua non* condition for RD data interoperability, for RD patients' visibility and for linking healthcare and research settings for RD. Ultimately, this project will contribute to better diagnosis and care of RD patients, because it will allow comparability of data, and therefore assessment of current practices and results against gold standards of care, necessary for taking actions and improvement.

The audience was reminded that 2 Boards are in place the ERN Board and the Data advisory Board (more info here: Boards | OD4RD). The Data Advisory Board recommendation on the project was presented.

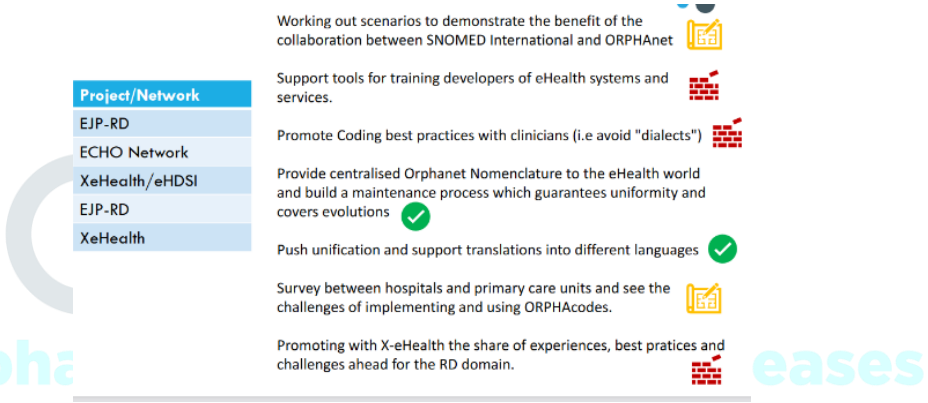


Figure 1 Data Advisory Board Recommandations for OD4RD and OD4RD2

Session 1 Network of Orphanet Nomenclature National Hubs

Network of National Hubs and coordination activities

The actions in this project aim not only at maintaining and developing further the ORPHAcodes, but also to help lowering the barrier for rare diseases patients' visibility and data generation for both primary and secondary purposes. We build upon the lessons learned in previous projects around rare disease codification (RD-ACTION, RD-CODE) and upon a the well-established Orphanet network to provide local support, at each hospital level, to implement ORPHAcodes in their information systems and ensure accurate use of the ORPHAcodes. Furthermore, this network will spread good coding practices, so as to contribute to quality of the data generated. Such strategy will bring a very practical piece in the gear of collecting rare disease data, making recommendations come to the reality. It is

expected to be a long-lasting effort, in which ERNs and hospital managers will be associated, as well as the European Commission.

With this pilot year, the Network of National Hubs has been developed in 13 MS countries (Austria, Belgium, Czech Republic, Finland, Germany, Italy, the Netherlands, Norway, Poland, Portugal, Spain, Sweden, Slovenia) and Switzerland as an observer.

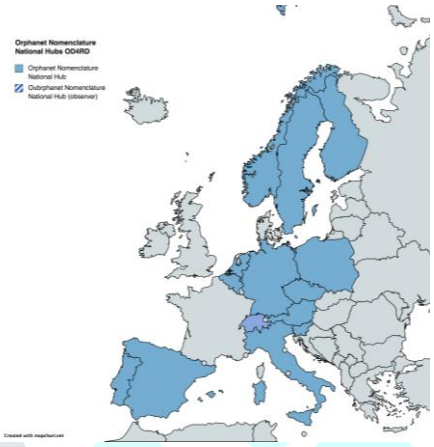


Figure 2 Current Nomenclature National Hub Net work

The objectives of the Network of Hubs is to ensure support for local implementation of ORPHAcodes in national HCPs hosting ERNs and national HCPs linked to ERNs. Each Hub will be the national helpdesk and contact point for all questions related to ORPHAcodes and ORPHAcoding, it will promote ORPHAcodes and ORPHAcoding to the different stakeholders and organize trainings for coders to ensure accurate and standardized coding. As of today, each Hub has established its annual action plan regarding three main lines of action, and these are being implemented at different levels by the different hubs.

To ensure that the same quality support is provided in each country, a scientific and coordination support to the national hubs is organized by BfArM & Inserm. In particular:

- to empower each National Hub a 'Train the trainers program' adapted from the EJP-RD project was developed and 41 persons were trained, with the objective to reach advanced training level for all national hubs.
- to set up a central and federated helpdesk in the form of an issue-tracking tool.
- to coordinate monthly meetings with all national hubs allowing a common promotion strategy by sharing experience between hubs, and to further learn about ORPHAcoding with practical examples from real users of ORPHAcodes from different settings and countries. These meetings also allow to discuss development of users-oriented common communication material (training, quiz, survey), discuss on best ways of bringing professionals together (over-booked schedules) and how to reach other stakeholders (*i.e. Ministries of Health, Directories of Health, hospital managers, decision makers...*) and ERN representatives. And overall, they facilitate structuring and leading national helpdesk efficiently.

During the next phase of the project the network will be expanded to 6 new national hubs (Bulgaria, Ireland, Estonia, Latvia, Lithuania and Romania). With the objective by the end of the 33 months to have 19 +1 observer fully autonomous National Hubs.

Spotlight on Belgian National Hub

In Belgium the National Hub is hosted by Sciensano. The RD coding state of play is extremely variable as there is no legal basis for RD coding. Currently in the **Central Registry of Rare Diseases (CRRD), different coding systems** (ORPHAcodes, SNOMED-CT, OMIM, HPO, ICD-10, …) are allowed (fig.).

Coding system used to encode the disease diagnosis							
BELGIAN HOSPITAL/CENTRE	UZA	CHU de Liège	IPG	UZ Gent	UZ Leuven	UCL St-Luc	UZ Brussel
ORPHAcodes	92.7	100.0	98.0	99.0	0	97.1	0
OMIM	1.4	0	0.3	0.2	0	1.0	0
ORPHAcodes + OMIM	5.8	0	1.9	0	0	1.0	0
ICD-10	0	0	0.1	0.2	0	0	0
HPO	0	0	0	0.1	0	0	45.5
SNOMED-CT	0	0	0	0.1	100	0	0
ORPHAcodes + ICD-10	0	0	0	0.1	0	0	0
HPO + OMIM	0	0	0	0.1	0	0	29.9
No disease code	0.2	0	0	0.4	0	1.0	1.3

Table 1 CRRD data received from the GC of university hospitals (2022)

The National Hub in Belgium has been very active in terms of promotion activities and in organising National Trainings, with three sessions already organised in University Hospitals and a fourth scheduled before the end of the year.

They are also working on production of new material and in the next months will start targeting Governement and other Health Institutions.

Spotlight on Spanish National Hub

The Spanish National Hub is hosted by CIBER. There is no ORPHAcode implementation in routine coding systems at a national level, though there are some isolated initiatives in some regions and hospitals. Also, there is a RD National Registry in place to which information is reported using ORPHAcodes (OC), but only for a very limited number of diseases (22 at present; plus another 12 in 2023; and close to 40 are under study)

The RD-CODE project participation was key to advance in ORPHAcode implementation since it allowed to establish the ORPHA-ICD-10-ES mapping and it managed to get 7 regional registries involved.

Given the complexity of the situation, the Spanish Hub felt that they could not come up with an action plan adapted to the reality of Spain without conducting a survey to provide some information about the State of Play of RD codification at reference centers.

So, the first steps have been to create a working group integrated by some former RD-CODE members and to launch a survey. As of today, Hub members are in this second phase corresponding to the analysis of data that will lead them to planify future action.

A summary of the 112 HCPs participating in the 24 ERN in Spain was presented. They are distributed in 26 hospitals from 10 Autonomous Communities, and some of them participate through a mixed unit integrated by an adult and a pediatric hospital.

Although the situation is complex, in spite of having 17 regions in Spain, only 10 of them participate in at least one ERN and 85% of HCPs concentrate in Catalonia and Madrid, which are the 2 of the communities with greatest involvement and with who we are working in close collaboration.

The preliminary results from the survey to assess ORPHAcodes use in HCPs were ERNs are hosted shows that:

- 80% of the centres replied that they do send epidemiological information to the ERNs coordinating team but not all use ORPHAcodes.
- It appears that the coding habit is not homogenous inside the same ERN.
- for those replying that they DO NOT use ORPHAcodes, reasons were multiple and included "the codes have not been implemented in the hospital" or "lack of knowledge and staff" but other respondents pointed to the ERN lack of request, such as "we comply with the instructions of the ERN".

Next steps to be followed include:

- Setting up the helpdesk
- Organisinge training
- Starting to provide technical support
- Initiating political action

Session 2 Highlights on collaboration with ERNs

Collaboration with ERNs are an important aspect of this project, in order to:

- improve the quality of the Orphanet Nomenclature and classification of RD reflecting the evolution of the medical knowledge;
- provide a high-quality coding system that reflects the needs of the users;
- create and update the textual information associated with RD as a mean to aid accurate assignment of ORPHAcodes;
- valorize the ERNs RD expertise, combining it with the complementary Orphanet expertise in databasing, to produce the highest quality, interoperable Nomenclature of RD.

OD4RD collaborates with ERNs both from the Nomenclature classification production point of view but also for the production of abstracts of the Orphanet encyclopaedia.

Collaboration for Nomenclature and classification revision

A procedure describing this collaboration has been delivered in the frame of the OD4RD project and it is available online : Collaboration with networks of expertise for the revision of the Orphanet nomenclature and classification of rare diseases

During the presentation, particular emphasis was given to how prioritisation of demands for modification to the Nomenclature coming from ERNs were done (fig.3). It is a multi-factor decision that considers both if the lack (or modification) of the code is blocking coding activities, if there is a

publication involved, are also taken into consideration the complexity and scale of the demand versus the available resources staff wise in the Orphanet team but also whether a reactive « contact point » is available at the ERN level.

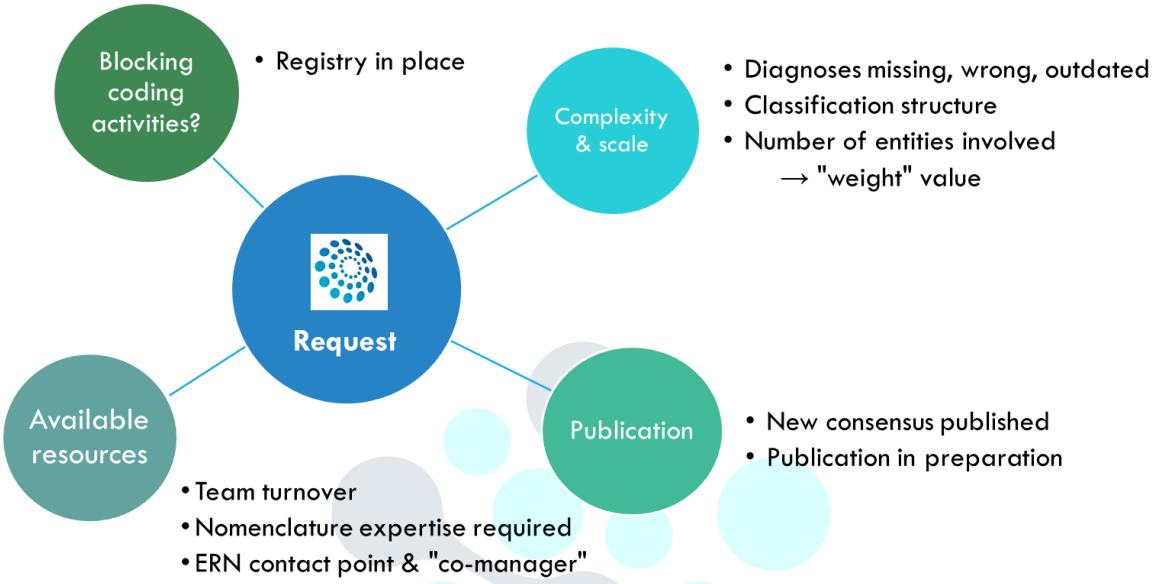


Figure 3 Demands prioritisation for creation/modification of ORPHAcodes entries

The complexity and scale of a collaboration is estimated considering the number of ORPHAcodes to be revised and the changes that need to be done to the classification structure (table 2);

Low complexity	Medium-low complexity	Medium-high complexity	High complexity
Small group, no structure revision	Medium group, little to no structure revision	Medium group, structure revision	Large group to entire classification, structure revision

Table 2 Complexity and scale of a collaboration

Finalised collaborations prior to 2022 were presented (table 3).

In 2022 were completed 1 medium-high complexity project and 1 medium- low complexity project. 5 more collaborations have been initiated (details in following tables) (table 4);

In the following 3 years we aim to complete 7 collaboration projects per year, ranging from low complexity to high complexity (details in following tables).

ERN	Diseases group	Status	Year
ERN-EYE	Rare ophtalmic diseases (first Orphanet-ERN collaboration)	Finalized	2018
ERKNet	Global revision with focus on Thrombotic microangiopathies, Glomerular diseases, and Rare causes of hypertension	Finalized	2019
ERN-SKIN	Epidermolysis bullosas	Finalized	2020
VASCERN	Primary lymphedema	Finalized	2020
eUROGEN	Anorectal malformations	Finalized	2021
RARE-LIVER	Vascular liver diseases	Finalized	2021

Table 3 Finalised ERNs collaboration for nomenclature revision prior to 2022

ERN	Diseases group	Status	Year	Weight
ERKNet	Renal tubular diseases & ciliopathies	Finalized	2022	**Medium-low complexity** Revision of ~60 ORPHAcodes + small structure changes
ERN CRANIO	Cranial malformations	Finalized	2022	**Medium-high complexity:** Revision of ~80 ORPHAcodes + structure

Table 4 2022 finalised ERNs collaboration for nomenclature revision (with weight)

ERN	Diseases group	Status	Year	Estimated weight
ERN-BOND	Primary skeletal dysplasias & Dysostoses (in collaboration with ISDS)	Ongoing	2022-23	High-complexity
VASCERN	Vascular anomalies	Ongoing	2022-23	High-complexity
ERN CRANIO	Orodental malformations	Ongoing	2022-23	Medium-high complexity
ENDO-ERN	Pseudohypoparathyroidism (PTH/PTHrp signaling pathway)	Ongoing	2022-23	Medium-low complexity
ITHACA	Intellectual disabilities	Ongoing	2022-23	Medium-high complexity

Table 5 Ongoing ERNs collaboration for nomenclature revision (with weight)

ERN	Diseases group	Status	Year	Weight
MetabERN	Inborn errors of metabolism	Initiated	2023	High-complexity
EpiCARE	Rare epilepsies	Initiated	2023	High-complexity
ERN CRANIO	ENT disorders	Not yet initiated	2023	Not yet estimated
EuroBloodNet	Under discussion	Not yet initiated	2023	Not yet estimated
ERN-RND	Possible revision of spastic paraplegias (to be solicited by Orphanet)	Not yet initiated	2022-2023	Not yet estimated

Table 6 Planned ERNs collaboration for nomenclature revision (with weight)

Collaborations for Encyclopaedia texts revision

The methodology for collaboration was presented (fig.4). It has been established based on the experience with ITHACA, ERKNet and EpiCARE, and is adaptable to the needs and resources of each network. It is published on the Orphanet website: Creation and Update of Disease Summary Texts in English for the Orphanet Encyclopaedia for Professionals

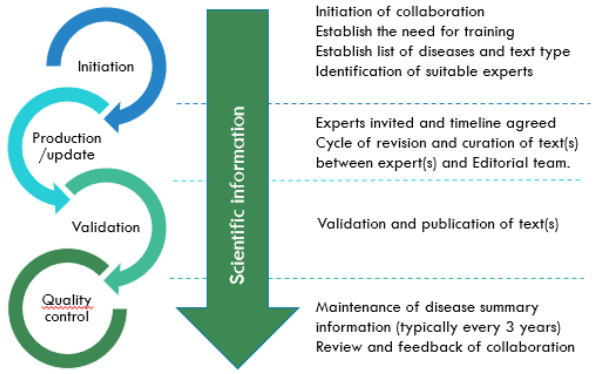


Figure 4 Methodology for Encyclopaedia text revision

Structured collaborations are ongoing with ERKNet; ITHACA ; EpiCARE ; ERN-Skin and MetabERN.

Punctual collaborations have been carried on with Endo-ERN, EURO-NMD, EuroBloodNet, ERN RARE-LIVER, ERN-LUNG, ERN-Skin, eUROGEN, ERNICA, ERN-RND, ERN-EYE.

FURTHER COLLABORATIONS are open for discussion, especially in the frame of the Nomenclature revision in harmonization with WP2.

Lessons learned

The Lesson learned from this multi-years collaboration are multiple and allowed to improve the exchange and accelerate the delivery of the work. In particular :

- Explaining and training experts to the rules of the Orphanet Nomenclature and classification at the beginning of a revision facilitates the collaborative work;
- A responsive contact point in the working group (either a project manager or an expert) helps with coordinating efforts;
- Assessing the weight of a project and determining a precise methodology helps planning and meeting deadlines;
- Establishing a prioritisation list is fundamental to respect the working capacity of experts and Orphanet nomenclature managers;

Next steps and objectives

- In the second phase of the project, every Orphanet nomenclature manager should be able to work on one or two revision projects according to their workload (increased weight of the target collaborations to take into account the new members of the Orphanet Nomenclature team)
- In 2023, complete the ongoing projects and the planned projects as seen in previous prioritization table;
- Collection of new collaboration requests to plan for the second half of 2023/beginning of 2024

In parallel what we plan to investigate is:

- Are ERNs using ORPHAcodes and if not what is the blocking problem? Lack of codes? Outdated classification?
- Are ERNs using the new codes created together with Orphanet and if not, what is the blocking problem?
- How can Orphanet better engage with experts or WG inside the ERN ?
- In a highly complex revision process, is there a point to stop revising? Where is this "good enough" target? Does it correspond to the possibility of coding a patient?
- Would experts involved in the classification revision process be interested in continuing the collaboration by engaging in abstract production?

Discussion

During the discussion it emerged that in many Hospitals, clinicians use HPO to code patients rather than using the ORPHAcodes. This results in inaccuracies as HPO signs describe phenotypic traits while ORPHAcodes describe a specific diagnosis. The audience was informed that this problem is known and it is clearly addressed in the Nomenclature trainings which are provided by the National Hubs.

Furthermore, in many Hospitals the mandatory coding system to be used is ICD-10 as it is exploited for reimbursement purposes. While it is clear that accurate ORPHAcoding at the bedside is more important from a diagnostic point of view and for primary and secondary use of data, this code has to be used often in addition to a generic and mandatory Coding system. This can be perceived as a burden by the coder. The audience was re-assured that also this problem is known and a solution can be implemented to avoid this additional burden. Indeed Orphanet makes available in the Nomenclature

Pack alignments files, also in the form of API and mapping services so that an automatic process can be set up to automatically retrieve the aligned ICD-10 upon entering the ORPHAcode.

From an IT perspective a process can be put in place to re-use the data and make them directly available in registries, depending on the structure of electronic health record, so that the coder only codes once at the bedside. The audience insisted on the importance on making sure that these possibilities are understood at the Hospital manager level and implemented at the IT level.

A summer school on ORPHAcoding for all ERNs was suggested, as well as the need for educating each ERNs coordinating team.

The importance of ORPHAcoding in the monitoring activities of ERNs liaison with the ERN monitoring working group was suggested as better quality of the monitoring could benefit from better knowledge on ORPHAcoding.

The future joint action on ERN integration at the national level was mentioned and it was clarified that the OD4RD and OD4RD2 projects are intented to provide support in ORPHAcodes implementation while the actual uptake and implementation by the Hospital should be considered and included in the next joint action.

The importance about coding undiagnosed patients was also evoked. The current available recommendation was reminded to the audience (http://www.rd-code.eu/workpackage-5-enhancements/). Moreover, since July 2022 a specific ORPHAcode for undiagnosed RD after full investigation is available; it must be used under very clear conditions, in addition to HPO terms to further describe the phenotypic traits. In conclusion the audience suggested that a dedicated training course on the subject should be developed to ensure a standardised approach at European level.

Session 3: Support and services

Analyse and deliver Reports

One of the objectives of the project is also to analyse and produce reports on RD coverage, identifying overlaps, insufficiencies and gaps amongst ERNs in order to support evidence based decisions in the context of ERN coordination, Board of Member States (BoMS) and European Commission activities.

The elements to be analysed have been defined as below:

- Disease group coverage of ERN-HCP
- Disease group complementarity between ERN-HCP (within and across)
- Gaps in disease group coverage in ERN
- Insufficient disease group coverage in ERN
- Other topics thay may arise from EC, ERN CG, BoMS

An initial virtual meeting with EC (former unit B3), chairs of the ERN Coordinating Group and BoMS co-chair and Orphanet representative was held to explain the project tasks, the methodology and define additional items if necessary. A first draft of a new survey to ERN HCPs requesting their impression on insufficient coverage was also presented during this meeting.

Currently, OD4RD is working on the preparation of the coverage and gap analysis and on the launch of the “insufficiencies” survey.

Regular information with updates and discussions of the ERN-Coordinating Group and the Board of Member States will be given and in parallel close collaboration with the ERICA project has been established in particular regarding the integration of information on ERN research and Clinical Practice Guidelines activities into the Orphanet database.

As of today the ERN centers designated in 2021 have been integrated into the Orphanet database as well as their relevant link to the ORPHA classification. The analysis of disease coverage by ERN is ongoing, based on the Primary and secondary thematic areas declaration by ERNs. The first report for discussion will be available in the next few months. Preliminary results are shown in the table below, they are compared with the results from the GAP Analysis carried out in 2019 which was based on ERN declaration. The aim was to asses if some GAPs from the first designation campaign have been covered thanks to the second designation campaign:

Remaining gaps	Covered gaps
- ERNICA: Rare non-malformative gastroesophageal diseases - ERN-RND: Rare autonomic system disorders Rare sleep disorders Rare headaches Other neurological pain-related disorders Some neurovascular malformations and diseases - eUROGEN: Rare (male) infertility - Rare (non cancer) gynecological and obstetric diseases	- Disorders of lipid metabolism / dyslipidemias (MetabERN) - Paraneoplastic neurologic syndromes (ERN-RND) - Lipodystrophies (EndoERN) - Autoimmune polyendocrinopathies (RITA) - Non-histaminic angioedema (RITA) - Amyloidosis (EuroBloodNet, ERKnet, GUARD-HEART, RITA, EURO-NMD) - Systemic mastocytosis (EuroBloodNet) - Peritoneal tumors (EURACAN)

N.B. Gap Rare infectious diseases are not included nor mental diseases apart from the neurological part.

These very preliminary quantitative results show already that some gaps have been covered while some remain to be addressed.

Next steps

It is important to note that this is a quantitative analysis based on the declaration of the ERNs and that a qualitative assessment of the accuracy of the data has to be set up with each relevant ERN coordinating team. To define a methodology for quality analysis OD4RD will work with 3 exemplar ERNs. The exchanges will enable to assess if the mapping exercise has really captured all relevant codes as sometimes the files are not complete. If this is the case, the additional diseases covered will be included so to obtain the most exhaustive representation of disease coverage. This exercise will enable ultimately to have sets of ORPHAcodes by ERNs that can be a starting point to establish coding guidance to be shared intra the ERN and it will also allow us to deliver a report by the end of the project (March 2023) encompassing both quantitative disease coverage and qualitative POC report on 3 ERNs

Furthermore, we are currently planning to request a meeting with the new EC team in charge of the ERN file (since 1st October) and be able to make a presentation of the draft disease coverage report by the end of the year.

Development of Services

In the frame of the OD4RD project we also want to facilitate the implementation of the ORPHAcodes from a technical point of view by providing tools and services that can be adapted to the different settings and situations. Indeed, the objective of the IT support task is to make from Orphanet Knowledge base “actionable items” for stakeholders and provide tools to feed RD community needs.

An overview of the new look and feel of the Orphadata website was presented. From this platform you can download all the necessary computable files for implementation of ORPHAcodes in It systems as well as all the informative human readable documentation (https://www.orphadata.com/orphanet-nomenclature-for-coding/) .

The ORPHAcode API developed in the frame of the RD-CODE project as well as the data visualisation tool continue to be available and their content updated yearly in the frame of the OD4RD project. Both are available from the Orphadata website (https://api.orphacode.org/ & https://dataviz.orphacode.org/).

Some news services were also presented:

- additional APIs available for annotation and research
- A online mapping service Mapping files to facilitate the transcoding (fig.5)

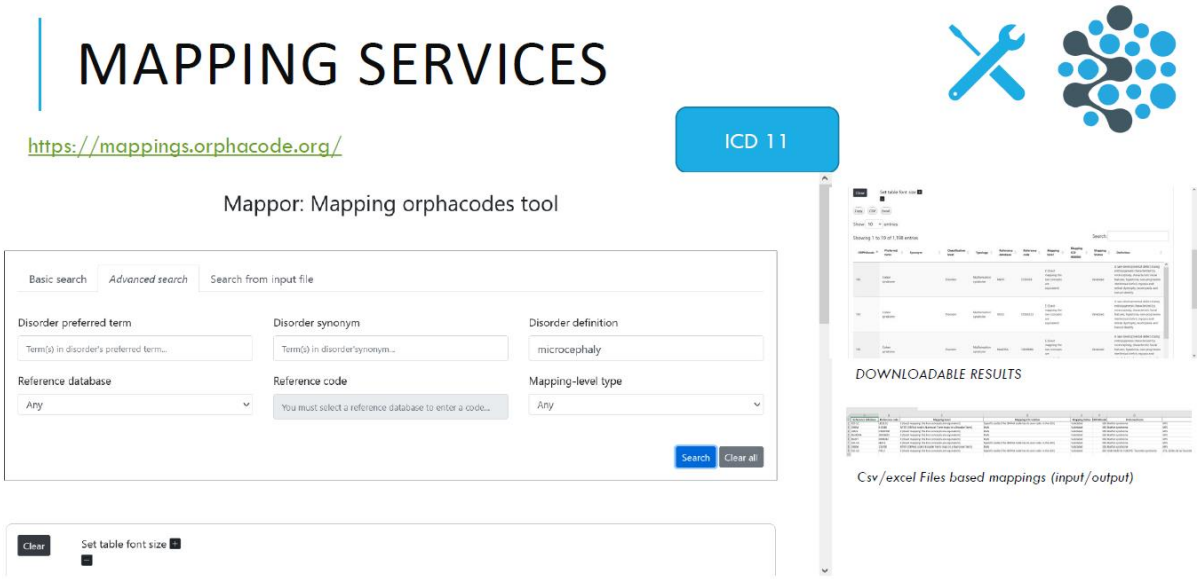


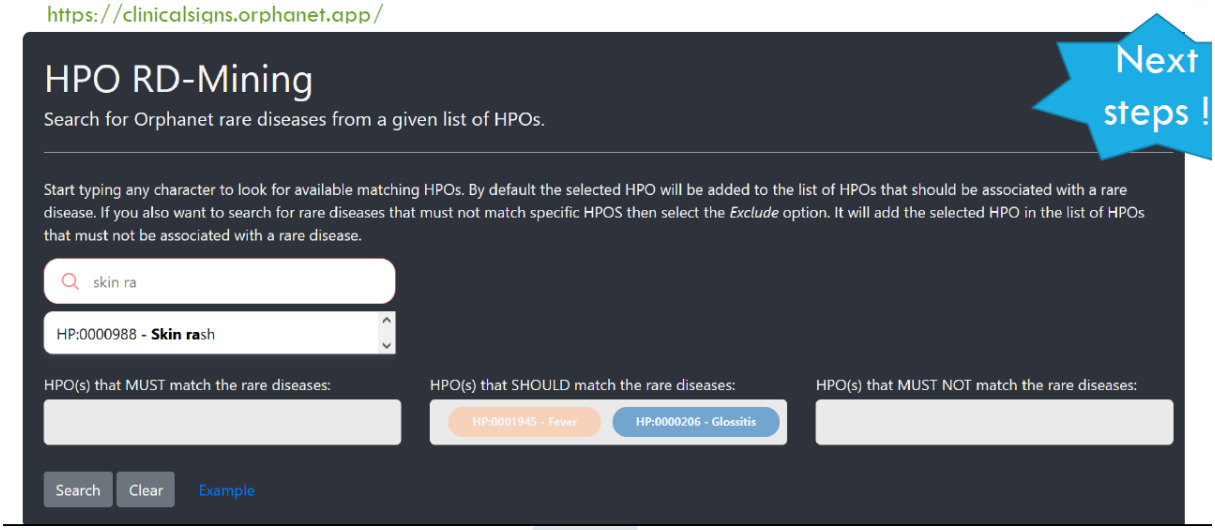
Figure 5 Online Mapping service

- HPO RD-Mining (fig.6) : to Search for Orphanet rare diseases from a given list of HPOs.

Start typing any character to look for available matching HPOs. By default, the selected HPO will be added to the list of HPOs that should be associated with a rare disease. If you also want to search for rare diseases that must not match specific HPOS then select the *Exclude* option. It will add the selected HPO in the list of HPOs that must not be associated with a rare disease.

N.B. It it not a differential diagnosis tool!

https://clinicalsigns.orphanet.app/



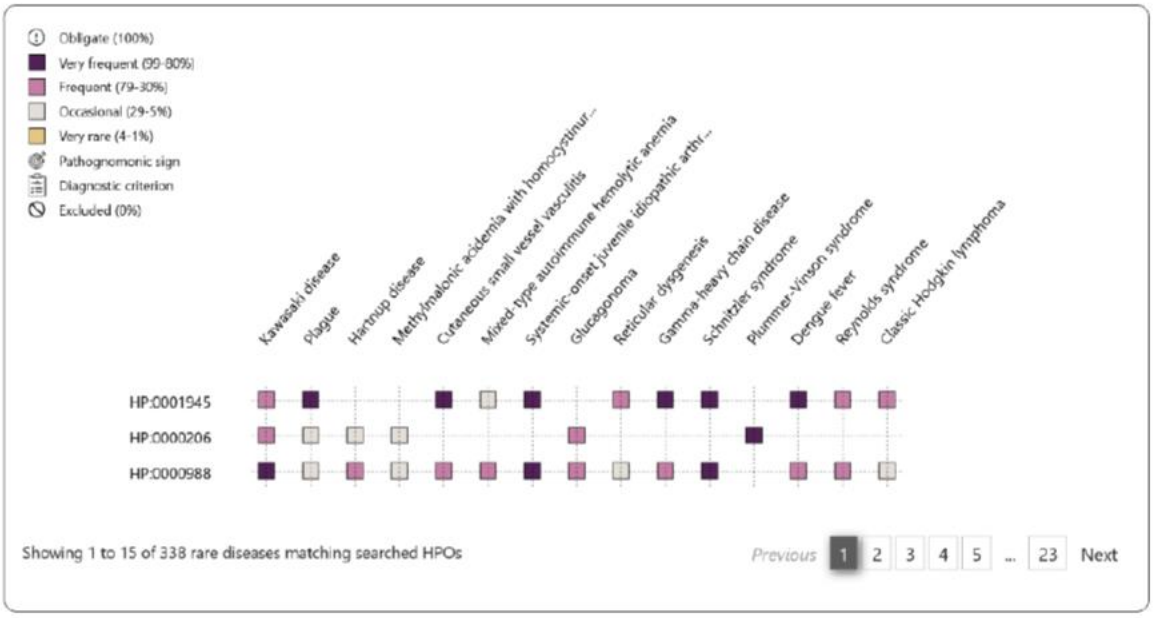


Figure 6 HPO RD-Mining

- Also a dynamic data visualisation service (fig.7) have been developed with ITHACA ERN, and thank to its support, as shown in the fig below as an example of Orphanet-ERN collaboration :

https://id-genes.orphanet.app/ithaca/

Figure 7 Dynamic data visualisation service : ITHACA/ORPHANET collaboration example

Next steps

Orphanet will continue making available local instances of services (API, « dataviz », mappings…) and Finalize “legal” management of APIs gateway

Orphanet will continue to work on Expanding the tools & services to facilitate technical implementation of ORPHAcoding and ease transcoding

Discussion :

When ORPHAcodes are already in place and used at the Hospital level then all the monitoring data is already available and ready to use. This includes re-use for further Reports and analysis. (only for full members of ERNs).

As the topics ERNs and Orphanet have been separated from the data unit in EC, the audience wondered whether the new team can be assisted with a briefing of previous activities. It was also noted that it is important they are aware of the importance of ORPHAcoding and of having real life data which is important and something very valuable and precious in the RD field. Data also helps to prioritise and planify ERN work and across ERN work. The audience wondered also how to guide this new Unit (which is primarily Public Health oriented) to be sensible to the « data » aspect of both networks. It was agreed to put in place a tri-partite advocacy (from ERNs, Orphanet and BoMs) to brief the new team about the importance of the data side at the Orphanet and ERN registry level.

Concerning HPO translations, the processes are being set up for standardised HPO translation into French and then there will be extensible to other languages. The main burden is to maintain it and follow its versioning which is frequent.

It is important to promote the message that ORPHAcoding does not mean changing the current and mandatory coding habits in a Hospital. Indeed, Mapping files and services and tools are available so that Hospitals can use them to faciliate transcoding. It was also noted that it is also possible for some Hospitals that still want to code in ICD-10 to obtain the corresponding ORPHAcode a posteriori also exploiting the mapping file. It was stressed that this is of course possible and already in place in some hospitals but this type of « reverse » ORPHAcoding approach needs a step of assessment for every assigned code.

Conclusion and take away messages

OD4RD is working on supporting ORPHAcode implementation not only by developing a network of hubs that will be able to train coders locally and setting up local high-quality helpdesks but aims at facilitating the technical implementation process by providing tools and services that can adapt to different settings and situations.

The OD4RD2 project, which is currently under evaluation, will continue this work and will capitalize on this pilot phase and has the ambition to expand the network, increase the number of ERNs collaborations and develop material to better understand transcoding.

We will continue contributing to the strategy of leaving no one behind in analysing the gaps in expertise at the ERN level and at the national level. We will also work hand in hand with the new joint action for ERNs integration and with all the health data strategy projects at the EU and national level to ensure the presence of the RD use case.

Take away messages

Regarding the RD data strategy :

OD4RD is providing coding resources and support for implementation and exploitation

- Building on the pre-existing Orphanet organisation
- Building on Orphanet-ERN complementarity

Synergies and uptake of OD4RD results should happen in:

- ERN National integration
- ERN data strategy

European leadership: EU experience can be transposed to other geographies.

Furthermore, listed below are some actions to be carried out in the future as emerging from discussion:

1. At the coordination level :
   - Increase direct connections with each ERN coordinating team for awareness, including communication on planned training dates to increase the number of participants by pushing also the information through the ERNs channels. Consider a summer school type event organised by the Orphanet coordinating team for all the coordinating teams of ERNs and/or plan more F2F events
   - Provide ERN coordinators with the list of ORPHAcodes in their area of expertise to faciltate ORPHAcoding guidelines in their centres
   - Increase the connections with the Hospital Alliance managers for awareness on the tools and service available to decrease coding burden and facilitate transcoding
   - Raise awareness within the new EC unit (common strategy Orphanet-BoMS and ERNs)
   - Liaise with EURORDIS ERN team
2. Ideas of material to be developed centrally:
   - Dedicated Training to the undiagnosed code use
   - Training for trainers : ensure that all hubs can fully explain the benefits of ORPHAcoding vs other general terminologies and the methodology Alignments with other terminologies as well as the different services and tools available, so that they can discuss at the Hospital level what solution can be implemented to ease the burden of the coders and facilitate transcoding.

Annex 1: Meeting Feedback and evaluation survey results

Feedback survey

A survey was launched live and stayed open for 3 days in order to allow participants to share additional feedback, in addition to suggestions already highlighted during the discussions and available in this report :

- It was suggested to organise an additional meeting before the end of the OD4RD project in March with a dedicated session on users feedback (i.e of ERN/Orphanet collaboration presented by both sides and have a ERN presenting their implementation of OC)
- It was suggested to share contacts of national hubs with all ERN coordinating team

Meeting and project Evaluation Survey

In order to collect insight into the knowledge regarding the OD4RD project and its activities prior to the meeting as well as satisfaction regarding the future planned activities and on the meeting itself the audience was asked to what extent they agreed to the 5 questions below [36 answers collected] (fig.8).

These results that the audience was well aware of the OD4RD project before the meeting, slightly less aware of the specific activities carried out.

The audience was satisfied with the outputs of the activities carried out as well as the activities planned in the next phases.

Finally, the meeting met the expectations of the audience.

Annex 2 : Agenda

10h30-11h Welcome Coffee

11h Welcome & Opening Ana Rath

11h45-13h Session 1 Network of Orphanet Nomenclature National Hubs (chair: S. Weber)

11h45-12h45 Highlight on National Hubs networks achievements (M.C Gaillard ; Orphanet National Hub Belgium, Orphanet National Hub Spain)

12h45-13h Open discussion/exchange with the audience about next steps for national Hubs

13h-14h Lunch Break

14h 15h10 Session 2 Collaboration with ERNs (chair: T. Voigtlander)

14h-14h30 Highlight on ERNs collaboration including next steps plans (C.Lucano)

14h30-14h50 Spotlight on a ERN collaboration (TBC)

14h50- 15h10 Open discussion/exchange with the audience about next steps for ERNs collaboration

15h10-16h30 Session 3: Support and services (chair: M.Hanauer)

15h10-15h45 Highlight on OD4RD Analysis reports and ambition for next year (T.Voigtlander and A.Rath)

Coffee break

16h00-16H20 Highlight on tools for implementation in Hospitals and Knowledge base in other systems and ambition for next year (M. Hanauer)

16h20-16h30 Open discussion about next steps

16h30-17h00 Final survey and Closing remarks