OD4RD2 internal Mid-term Workshop Report

June 2024

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Content summary

Executive summary

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:

1. 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).
2. To contribute to the harmonisation of data collection amongst settings (health records, registries) and amongst countries, through the dissemination of coding good practices at the data source level.

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3. 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.

The OD4RD2 mid-term internal Workshop was an online event held on the 6th and 7th of June 2024. It was intended as a check point of the ongoing tasks and activities in order to define mitigating actions if needed and also fine-tune the transversal work of the different Workpackages for the last months of the project.

The first day gathered 46 participants (from 18 Orphanet Nomenclature National Hubs (NH)) and was dedicated to discuss in depth, the OD4RD2 project achievements by Workpackage (WP) (www.od4rd.eu), the relevant indicators were also presented so as to fine tune the activities, discuss possible improvements as well as mitigating actions if needed. The last session of the day centered on success stories and problems encountered by the NH Network and sticky notes were used to capture ideas, thoughts and results (What have been your positive experiences during project year 1? What challenges did you face during project year 1? What are your expectations for Project year 2?).
The second day gathered 45 participants (from 16 NH), opened with a session dedicated to develop the thoughts (recorded thanks to the sticky notes session) and to plan the transversal activities such as the cross links with the JARDIN project, the organisation of an ERN Day, a Hospital Manager Day and the update of the communication Plan.

The final closing session ensured that agreement was reached on an updated and « user needs-orientated » action plan.

From the participants' perspective, collected thanks to a feedback survey launched during the meeting (17 answers out of 46 participants received! 100% of respondents were either satisfied (strongly agree or agree). 100% considered that the workshop helped them better understand the activities carried out in the workpackages they are not directly involved in. 100% of the respondents considered that the workshop inspired them for the next activities that will be carried out by their team.

Meeting Slides can be accessed here.

Mid-Term Internal Workshop Report

DAY 1

Participants

Orphanet Nomenclature National Hubs: Austria, Belgium, Bulgaria, Czech Republic, Germany, Estonia, Finland, France, Ireland, Italy, Lithuania, Netherlands, Norway, Portugal, Poland, Slovenia, Sweden, Switzerland, Spain.

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Opening

The Partners were welcomed by the Orphanet Network Coordinator and OD4RD2 project coordinator. The global aim of the project was reminded: to tackle the invisibility of rare diseases in European member states' health systems, promote harmonisation of practice and facilitate generation of standardised interoperable data around RD, thus contributing to meet the ambitions set by RARE2030 concerning data.

In particular, the project aims to:

- 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, 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).

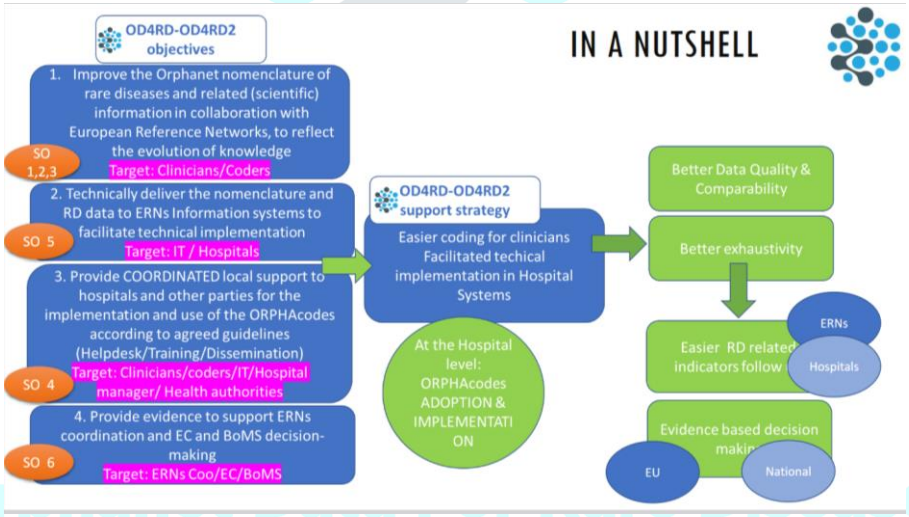

Figure 1 OD4RD-OD4RD2 project objectives linked with support strategy and final impact

The Objectives and strategy of the project were presented (fig.1) in relation to the specific objectives together with information on new partners (fig.2). Implementation of ORPHAcodes at the data source is essential and we provide support to facilitate this within OD4RD2, the objective is to have RD exploitable data at the Member States (MS) level in order to be able to also understand the situation at the EU level.

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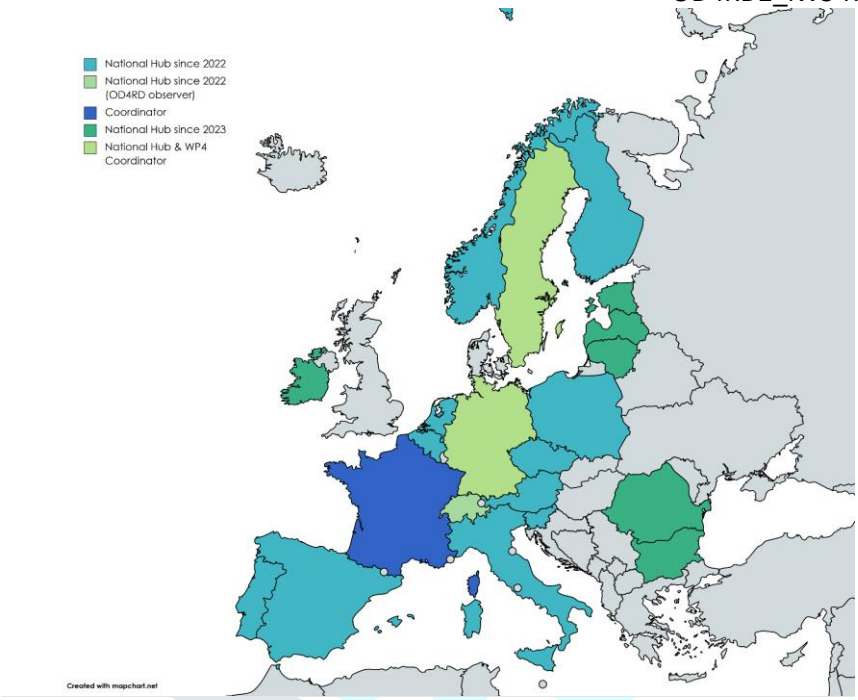

Figure 2 Orphanet Nomenclature National Hubs Network

The audience was reminded that 5 out of 6 new countries have carried out national hub activities since the beginning of the project, thus representing a network of 19 NH active in OD4RD2 as of today. Interactions with other projects were presented together with the expected output and impact of the project.

Figure 3 OD4RD2 interactions with other projects

The project's conditions for success were discussed together with the audience. These include:

- A nomenclature that fits coding needs and as such is used by the clinicians;
- A nomenclature that allows for transcoding in Health information System to avoid double coding and save time;
- To implement well-trained National hubs, in capacity to support local implementation;

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- To establish very good connections at the decisional level: Ministry of Health, Hospital managers and ERN coordination facilitating local level contacts and interactions;
- Awareness and support from ERNs governing bodies such as the ERN Coordination, the Board of Member States and the European Commission is of paramount importance.

Audience members were reminded of the governance structure of the project as well as administrative issues.

Finally, the expectations for the Workshop were presented:

- □ Have a common understanding on past, current and planned actions
- □ Share good practices
- □ Identify difficulties or bottlenecks and decide on mitigating actions
- □ Decide on ways to increase our reach at the national level
- □ Clearly define the interactions with JARDIN

Orphanet nomenclature and classification of RD update and maintenance (WP2)

The objective of the Workpackage 2 was presented: to develop, update and continuously maintain the Orphanet nomenclature (ORPHAcodes) and classification of RD, in collaboration with ERNs, in line with the evolution of knowledge.
The presentation of the objectives of the collaborations with ERNS followed. These objectives are:

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

The audience was reminded of the main achievement carried out in the scope of this workpackage during the OD4RD project:

The recruitment of an ERN collaboration project manager who acts as the main contact point for ERNS and coordinates and priorities all the ERN collaborations of the disease team as per figure below.

Figure 4 ERN project Management @ Orphanet

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- A methodology on the workflow behind a collaboration with an ERN was made available during the OD4RD project and is available here and recapped in the figure below. In this procedure all the different steps of the workflow are detailed as is the prioritisation of ERN collaborations , including how the complexity of each collaboration is assessed (table 1). Each Orphanet nomenclature manager works on one or two revision projects in parallel according to their workload.

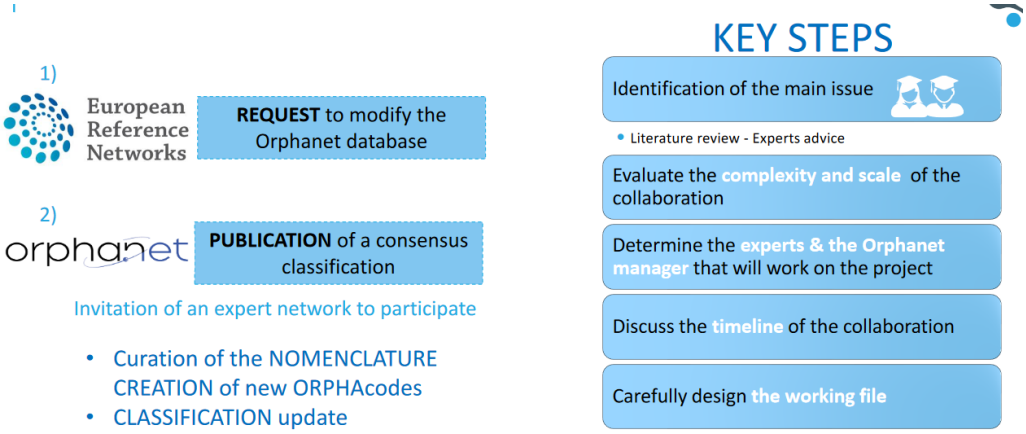

Figure 5 ERN collaborations key steps

Achievements were presented:

- 4/18 of the projects started in 2023 or 2024 have been completed.
- 2/18 of the projects started in 2023-2024 are ongoing and will be finalized in 2024.
- 5 new collaborations started in 2024 and are already ongoing.
- There are currently 6 new collaborations planned for the second half of 2024/beginning of 2025.
- a follow up table and status of ongoing Orphanet-ERN collaborations is available online and it is updated every six months: https://github.com/OD4RD/Main-Help-Desk/wiki/8.-Orphanet%E2%80%90ERN-collaborations
- A report on the collaborations carried out was also released (here).

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Follow up table and status of ongoing Orphanet-ERN collaborations [as for December 2023]

This table will be biannually updated in December/July in order to help users to follow current collaborations

ERN	Group Revised/ to be revised		weight of the collaboration	Collaboration main goal	Started	Status (August 2023)	Finalized (expected date)
ITHACA	ORPHA:183757	Rare genetic intellectual disability	High complexity	Classification revision and creation of new entities	March 2019	Finalized in January 2022	
ERN-BOND	ORPHA:364559	Dysostosis	High complexity	Classification revision and creation of new entities	December 2020	Ongoing	Early 2024
	ORPHA:364526	Primary bone dysplasia	High complexity	Classification revision and creation of new entities	December 2020	Ongoing	Early 2024
EuRRECa	ORPHA:90503	Pituitary hormone deficiency of tumoral origin	Medium-low complexity	Creation of new entities	February 2021	Ongoing	2024
ITHACA-eUROGEN / Spina Bifida and other dysraphisms	ORPHA: 268357	Neural tube closure defect	Medium-low complexity	Classification revision and creation of new entities	February 2019	Ongoing	December 2023
VASCERN	ORPHA:68419	Vascular anomaly or angioma, step 1: Rare vascular tumor, ORPHA: 211237	High complexity	Classification revision and creation of new entities	March 2019	Ongoing	August 2024
ERN-SKIN	ORPHA:79869	Autoimmune bullous skin disease	Low complexity	Classification revision and creation of new entities	June 2019	Ongoing	October 2023
EpiCARE	ORPHA:101998	Rare epilepsy, step 1: Epilepsy syndromes, ORPHA:166482	High complexity	Classification revision and creation of new entities	August 2019	Ongoing	August 2024
MetabERN	ORPHA:68380	Mitochondrial disease	High complexity	Classification revision and creation of new entities	January 2024	Starting	January 2029
	ORPHA:738	Porphyria	Low complexity	Classification revision and creation of new entities	January 2024	Starting	June 2024
ERN-BLOOD		Integration of Pediatric thrombotic diseases	Low complexity	Creation of new entities	August 2023	Ongoing	November 2023
TransplantChild	ORPHA:565779	Rare disorder potentially indicated for transplant	Medium-high complexity	Classification revision and creation of new entities	August 2023	Starting	2024
		Complication after transplantation	Low complexity	Classification revision and creation of new entities	January 2024	Starting	2024
PaedCan-ERN & SIOPE		Integration of Pediatric oncology data	High complexity	Classification revision and creation of new entities	End of 2023	Starting	2024
ERN-EYE	ORPHA:499047	Autoimmune/inflammatory optic neuropathy	Medium-low complexity	Classification revision and creation of new entities	October 2023	Starting	2024
	ORPHA:519315	Rare retinal disorder	Medium-high complexity	Classification revision and creation of new entities	September 2023	Ongoing	2024
ERNICA	ORPHA: 104012	Rare inflammatory bowel disease	Low-medium complexity	Classification revision, creation of new entities, nomenclature update	January 2024	Starting	September 2024
ERN-CRANIO	ORPHA:98026	Rare odontologic disease	Medium-high complexity	Classification revision and creation of new entities	January 2024	Starting	2024
	ORPHA:2014	Cleft palate	under evaluation	under evaluation	2024		
ERN-LUNG	ORPHA:182095	Interstitial lung disease	not yet evaluated	not yet evaluated	2024		
ERN-RITA		Pediatric rheumatology	under evaluation	under evaluation	2024		
		Autoimmune diseases	not yet evaluated	not yet evaluated	2024		

The revision is usually referring to a group into a classification **rather than the whole classification.** The changes are applied progressively and released in the Nomenclature Pack.

Figure 6 ERN collaboration table as of June 2024 (https://github.com/OD4RD/Main-Help-Desk/wiki/8.-Orphanet%E2%80%90ERN-collaborations)

The audience was then given insight into how 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.
So far, the following collaborations are in progress or are finalized (details in following tables):

3 Low complexity
5 Medium-low complexity
8 Medium- high complexity
3 High complexity

Low complexity (0,5)	Medium-low complexity (1)	Medium-high complexity (1,5)	High complexity (2)
Small group, no structure revision	Medium group, little to no structure revision	Medium group, structure revision	Large group to entire classification, structure revision

Table 1 Complexity assessment per ERN collaboration

As well as weight and timeline of the collaboration

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ERN	GROUP	WEIGHT	START	END
ITHACA-eUROGEN	Spina Bifida and other dysraphisms	1,5	February 2023	December 2023
ERN EYE	Autoimmune/inflammatory optic neuropathy	0,5	September 2023	December 2023
ERN BOND	Dysostosis	2	December 2020	August 2024
ERN BOND	Primary bone dysplasia	2	December 2020	July 2024
ERN VASCERN	Rare vascular tumor	1,5	March 2023	May 2024
ERNMetabERN	Porphyria	1	June 2023	January 2024
EuroBloodNet	Pedriatic thromboses	0,5	August 2023	May 2024
ERN EYE	Rare retinal disorder	1,5	September 2023	September 2024
TransplantChild	Rare disorder potentially indicated for liver transplant	1	August 2023	2025
TransplantChild	Rare disorder potentially indicated for lung transplant	1	August 2023	2025
TransplantChild	Rare disorder potentially indicated for heart transplant	1	August 2023	2025
TransplantChild	Rare disorder potentially indicated for bowel transplant	1	August 2023	2025
TransplantChild	Rare disorder potentially indicated for HSC transplant	1,5	August 2023	2025
TransplantChild	Rare disorder potentially indicated for kidney transplant	1,5	August 2023	2025
ERN EpiCARE	Epilepsy syndrome	2	September 2023	2025
ERN ERNICA	Rare Inflammatory Bowel Disease	0,5	January 2024	2025
ERN CRANIO	Rare odontologic disease	1,5	January 2024	2025
ERN CRANIO	Clef palate	0,5	January 2024	2025
ERNMetabERN	Organic Acidurias	1,5	February 2024	2025
ERN VASCERN	Rare vascular malformation	1,5	May 2024	2025
ERN EURO NMD	Neuromuscular Junction disease	1	March 2024	
Inter ERN Mito-WG	Mitochondrial disease	2	May 2024	
ERN RND	Hereditary spastic paraplegia	1,5	*to be determined*	

Table 2 Complexity assessment per ERN existing collaboration with estimated length date

The indicators linked to WP2 activities were presented

- WP2 output indicator Target : 8.5/year

Cumulative weight of ERN projects finalized in 2023: 2 (2 collaborations started in 2023)
Cumulative weight of ERN projects finalized in 2024: 8,5 (6 collaborations started in 2020 or 2023)
Cumulative weight of ERN projects planned for 2025: 14,5 (12 collaborations ongoing started in 2023-2024)

- WP2 impact indicator

Number (%) of finalised ERN collaboration projects, after which the related ERNs/ERN thematic group(s) have expressed their intention to effectively implement and use the revised ORPHAcodes in their activities including, for instance, monitoring, coding activities, publications, registries, etc.
→ Currently working on a better survey to collect the ERN feedback at the end of a collaboration

The audience was also presented with the number of ERNs collaborations that have been initiated, closed and still ongoing (table 2 to 5).

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ERN	Diseases group	Status	Year
ERKNet	Renal tubular diseases & ciliopathies	Finalized	2022
ERN CRANIO	Cranial malformations	Finalized	2022
ITHACA	Intellectual disabilities - OrphaID	Finalized	2022-23
ERN-ITHACA + eUROGEN	Spina Bifida and other Dysraphisms	Finalized	2023
ERN-EYE	Autoimmune/inflammatory optic neuropathy	Finalized	2023
VASCERN	Rare vascular tumors	Finalized	2024
ERN-EuroBloodNet	Rare pediatric thrombotic diseases	Finalized	2024

Table 3 ERNs finalised collaborations for Nomenclature Revision

Detailed examples were given (see WP3 slides)

ERN	Diseases group	Status	Year
ERN-BOND	Primary skeletal dysplasias & Dysostoses (in collaboration with ISDS)	Ongoing	2022-24
VASCERN	Rare vascular anomalies	Ongoing	2023-24
MetabERN	Inborn errors of metabolism (finalized: Porphyrias)	Ongoing	2023-24
EpiCARE	Rare epilepsies: epilepsy syndromes	Ongoing	2023-24
ERN-EYE	Rare retinal disorder	Ongoing	2023-24
TransplantChild	Rare disorder potentially indicated for transplant	Ongoing	2023-24
ERN-CRANIO	Cleft palate	Ongoing	2024

Table 4 ERNs ongoing collaborations for Nomenclature Revision

Detailed examples were given (see WP3 slides)

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ERN	Diseases group	Status	Year
ERNICA	Rare inflammatory bowel disease	Initiated	2024
Inter ERN Mito-WG	Mitochondrial diseases	Starting soon – under discussion	2024
ERN EURO-NMD	Neuromuscular junction diseases	Initiated	2024
ERN PaedCan	Pediatric tumors	Not yet initiated (under discussion)	2024
ERN-RND	Possible revision of spastic paraplegias (solicited by Orphanet)	Not yet initiated	2024
ERN-LUNG	Possible revision of Interstitial lung disease	Not yet initiated	2024

Table 5 ERNs planned collaborations for Nomenclature Revision

An overview of the OD4RD lessons learned about successful collaborations for Nomenclature revision was presented to the audience:

- A responsive contact point in the working group (either a project manager or an expert) helps with coordinating efforts;
- Establishing a priorisation list is fundamental so as to respect the work capacity of experts and nomenclature managers;
- Assessing the burden of a project and determining a precise methodology helps planning and meeting deadlines, especially when treating high complexity projects, that need to be subdivided into smaller blocks;
- Training experts as to the rules of the Orphanet Nomenclature at the beginning of a revision facilitates the collaborative work;
- The lessons learned on the ERN side will be discussed during the day's interactive sessions.
- Training experts to the rules of the Orphanet Nomenclature 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 nomenclature managers.

Next steps were presented:

- ⇨ Work on a better survey to collect the ERN feedback at the end of a collaboration in order to improve the process.
- ⇨ Maintain the rate of completion of collaborative projects.
- ⇨ Accept new collaborations with ERNs.
- ⇨ Plan to contact those ERNs that have not yet collaborated with us to evaluate the bi-directional needs.

* Actions will be taken to the extent that there are Nomenclature agents available to do so.

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During the discussion it was pointed out that the content of the survey to assess the impact of the ERN collaboration should be brainstormed and also the training with the ERN experts could be an opportunity to remind the NH work as well as the helpdesks. The final report could also contain a paragraph with these items so as to strengthen the links with the NH.
It was also precise that the experts involved in the collaboration vary according to its scope. Coders can be involved in the discussion if collaboration is tackling coding issues

Develop Orphanet Knowledge and information base around RD in collaboration with ERNs (WP3):

The interactions between WP2 and 3 were presented to the audience.
WP2 and WP3 are closely interrelated since updates performed on the Orphanet Nomenclature naturally lead to quality control and updates on associated information (texts, genes, alignments), and vice- versa.

The WP3 objectives aim to ensure better comparability and interoperability in the RD domain and to provide up to date information on RD validated by ERN RD experts, as a mean to aid accurate assignment of ORPHAcodes were presented:

- To develop and continuously maintain the data associated with RD, and in particular:
  - Genetic data (*OMIM, HGNC…..*);
  - Other alignments of ORPHAcodes with the main terminologies in use in health information systems and registries (including *ICD-10, ICD-11, SNOMED-CT and OMIM*);
- To create and update the textual information associated with RD.

The action plan for this WP was presented and discussed as per fig.7 below:

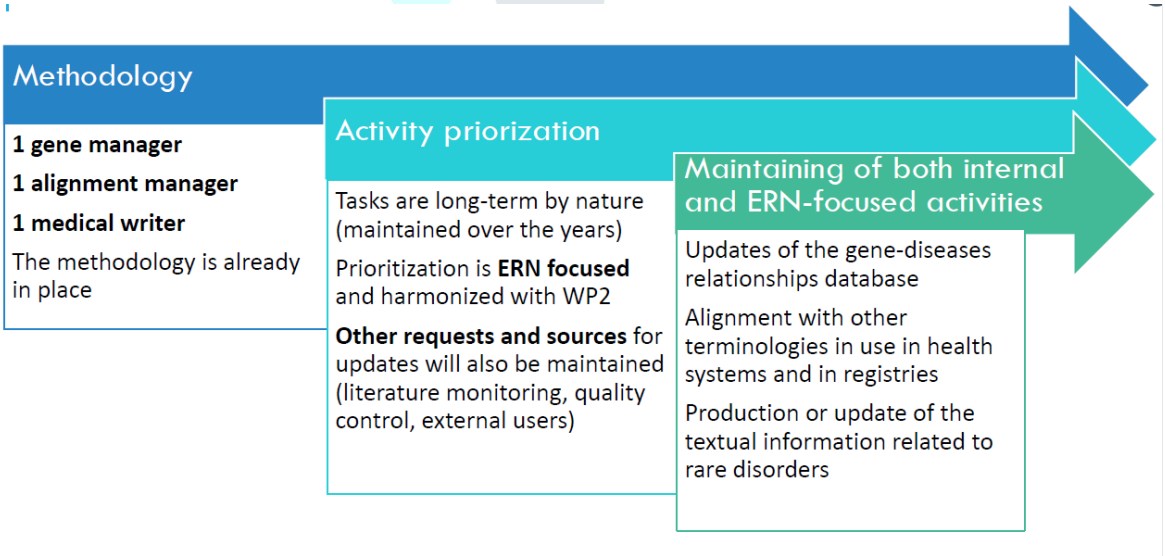

Figure 7 WP3 Action Plan

Moving forward the audience was reminded of impact of the Orphanet Knowledge base:

- For patients: understand the manifestations and genetic background of their disease.
- For professionals: facilitate examination, diagnosis and evaluation of patients.
- For policy makers: obtain reliable figures for evidence-based policy planning.

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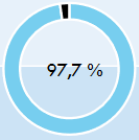
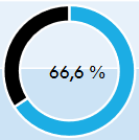
- For researchers: re-use of combinations of data for hypothesis making.
- For industry: facilitated knowledge of market size; reuse of combinations of data for hypothesis making in R&D (pre-competitive).

Then the Genetic data annotation process procedure was presented (here) and the OD4RD achievements regarding this task were presented (table 6):

(April 1 2023- April 30 2024) (compared to the previous period in OD4RD)	Output
Cumulative number of genes linked to rare diseases with a disease-causing relationship	**4,606 (+144)**
Newly created disease-causing genes (i.e. genes were absent from Orphanet and were created with a disease-causing relationship)	**66 (+5)**
New disease-causing relationships attributed to unknowingly disease-causing genes (i.e. genes were present in Orphanet as parts of diagnostic panels and were subsequently identified as disease-causing)	**No more pertinent**
Newly added disease-causing relationships attributed to knowingly disease-causing genes (i.e. genes already present in Orphanet linked to other diseases as disease-causing)	**225 (+95)**
Modification of gene-disease relationships (e.g. disease-causing germline mutation was updated to disease-causing germline mutation loss of function)	**8 (+1)**
Suppressed gene-disease relationships	**218 (+99)**

Table 6 Data Report: OD4RD genetic data production

The alignments process procedure was also presented to the audience (Procedure for ICD-10 alignments here) and OD4RD2 achievements regarding this task were presented (table 7):

ORPHAcodes aligned to ICD-10:	Total:	7478	(was 7288)
ICD-10 disorder level	Disorders:	6361	(99,7% aligned)
	Subtypes of disorders:	995	(96,6% aligned)
	Groups of disorders:	122	(5,6% aligned) *
ORPHAcodes aligned to ICD-11:	**Total:**	**5550**	**(was 1711)**
ICD-11 disorder level	Disorders:	4251	(66,6% aligned)
	Subtypes of disorders:	884	(85,8% aligned)
	Groups of disorders:	415	(19,2% aligned) *

* Please note that for group of disorders, Orphanet only aligns the exact concepts in all terminologies

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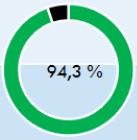
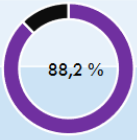
ORPHAcodes aligned to SNOMED-CT:	Total:	6552	(was 6397)
SNOMED CT disorder level	Disorders:	6016	(94,3% aligned)
	Subtypes of disorders:	305	(29,6% aligned)
	Groups of disorders:	231	(10,7% aligned) *
ORPHAcodes (genetic entities only) aligned to OMIM:	**Total:**	**4742**	**(was 4526)**
OMIM disorder level	Disorders:	4061	(88,2% aligned)
	Subtypes of disorders:	630	(80,5% aligned)
	Groups of disorders:	51	(3,8% aligned) *

* Please note that for group of disorders, Orphanet only aligns the exact concepts in all terminologies

Table 7 Data Report: OD4RD alignments data production

Challenges and next steps were presented:

- GENES: continue with the routine workflow, as per standard procedure;
- ALIGNMENTS: having achieved 99,7% coverage in ICD10 (disorder level), work will focus on maintenance of the alignment and an effort will be done to complete the ICD11 alignment. This is particularly challenging, as the codes that remain to be aligned fall mostly in the "attributed code" category (when no matching term exists), as the strategy that was put in place to align ICD11 started from specific codes, moving then to index terms.

Collaboration with SNOMED-CT still ongoing for 2024, work on the 2024 release is being finalized, according to the established process:

1. Orphanet produces the updates of nomenclature of rare diseases according to standardized procedures & collaboration with international rare disease expert networks

2. Knowledge is transmitted to and evaluated by SNOMED-CT according to their own guidelines

3. Matches are validated and approved on both sides, mismatches lead to a rejected mapping

The audience was reminded that Orphanet also works on aligning ORPHAcode of other relevant terminologies (fig.8).

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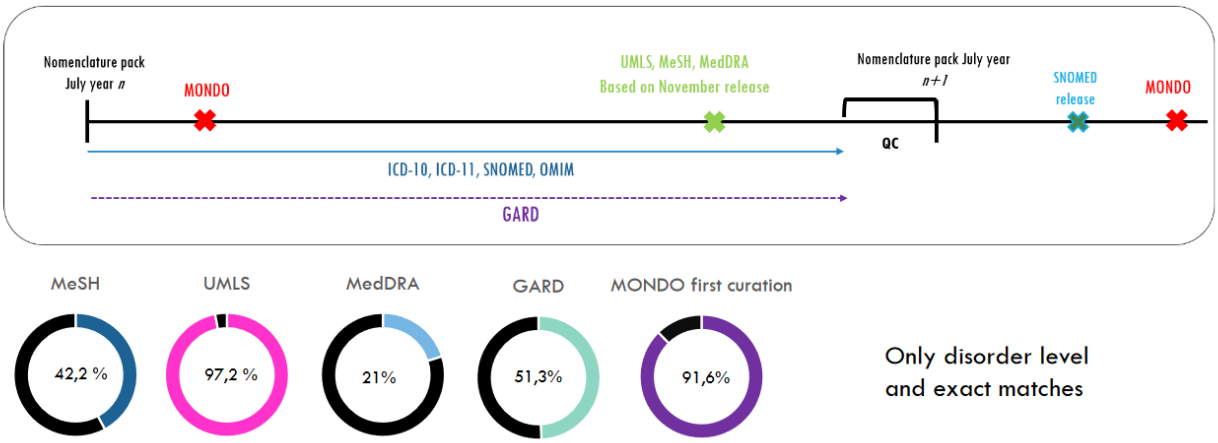

Figure 8 ORPHAcodes alignement to other terminologies (June 2024)

Discussion: The audience was reminded that the ICD10 represent different concepts and not only diagnosis, as such the codes capture a given patient in a given moment while OC are for RD diagnosis which are stable.

It was also noted that for clinicians the alignment code “Broder To Narrow” are difficult to use from a clinical point of view and that direct transcoding is only possible for exact mappings. The German team has carried out a transcoding work and establishes a file with 1:1 mapping between ICD10-GM and ORPHAcodes for implementation into the HIS.

The audience was also reminded that for all the OC not represented in ICD11: requests will be made to include these RD in the foundation to have an URI (which is a link not a unique identifier) , this will increase the representation of RD in ICD11 , improve ICD11 content and confirm OC as the interoperability back bone for RD.

The methodology behind the production of definitions and abstracts was also presented (here) and the detail of the ongoing structured collaborations with ERNs was presented as below:

Ongoing structured collaborations:

- ERN ITHACA
- EpiCARE
- MetabERN

Punctual collaborations have been carried on with:
Endo-ERN, ERN CRANIO, ERN EURO-NMD, ERN GENTURIS, ERN RARE-LIVER, ERN RITA, ERN-EYE, ERNICA, ERN- BOND, ERN-LUNG, ERN-RND, EURACAN, ERN-SKIN, EuroBloodNet, ERN-PeadCan, ERKNet, and ERN-eUROGEN.

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Indicators (April 1 2023-April 30 2024)			Total
Published texts			**358**
Abstracts (80 with ERNs)	Newly produced	26 (19 with ERNs)	
	Updated	107 (61 with ERNs)	
			133
Definitions (7 with ERNs)	Newly produced	153	
	Updated/Quality controlled	72 (7 with ERNs)	
			225

Table 8 *Data report Orphanet published texts (Abstracts vs definitions, newly produced vs updates)*

Challenges linked to this activity were presented:
Definitions:
Major limitation: Medical validation (difficulty of recruiting regular and volunteer experts to dedicate time for monthly validations);
Additional limitations: definition update/reformatting usually is shifting to creating a new definition for the old texts, thus they invade certain amount of time dedicated to entities without any definitions;
Abstracts:
Setting up collaborations is particularly difficult with the texts of very rare diseases where number of experts are few and extremely busy.

NEXT STEPS: To overcome the limitation of medical validation and increase the productivity, in the frame of classification & nomenclature revisions with experts, missing texts/texts updates will be included to the revision to facilitate/speed up the validation process.

Support to the EC in its ERN strategy including dedicated IT systems (WP5):

The objectives of the IT support tasks of WP5 were presented as per figure below:

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Figure 9 WP5 IT support tasks objectives

Evidence-based reports on ERNs

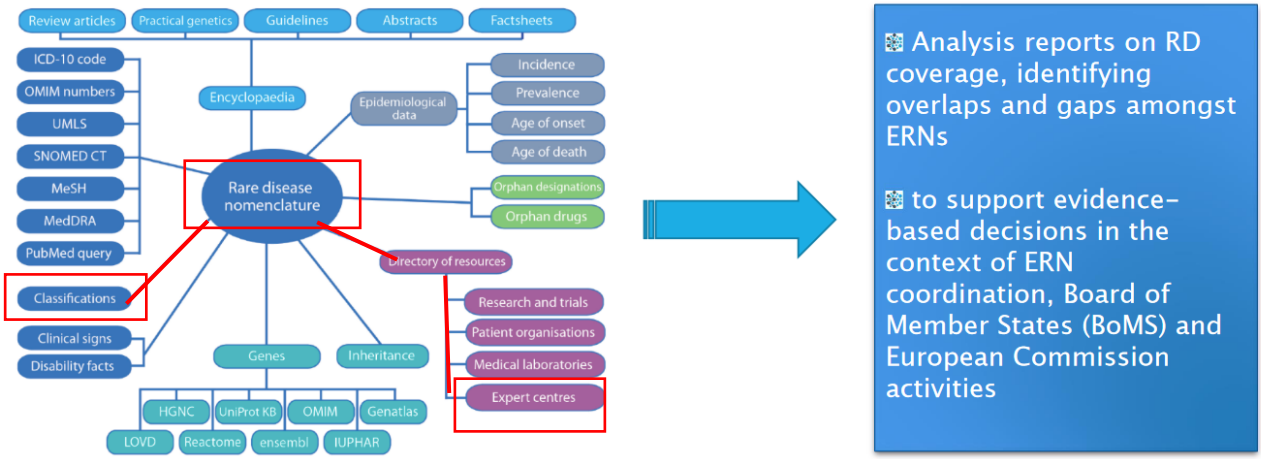

Figure 10 Orphanet Knowledge base content & evidence based reports

The results of the quantitative RD coverage and gap analysis by comparing ERNs covered thematic groups with the Orphanet classification of RD were presented to the audience.
The main objective of the analysis is to identify groups of RD that are still left out by the ERN system and to suggest possible thematic extensions.

The sources used were:

- Application files (EC): thematic groups, sub thematic areas, exemplar RD Orphanet classifications of RD
- ERN websites
- Orphanet ERNs centres database linked to the classifications (~99% complete) (as per highlight in fig.10).

Methodology

1) mapping ERNs' units to ORPHA classification:

ERNs' centres data collection and mapping to the Orphanet classification of RD were extracted from the Orphanet database. (N.B. 99% of ERN units inventory is completed & these inventories are quality controlled: check center by center by Orphanet network information scientists and CQ by Orphanet coordination)

2) thematic/ sub thematic areas were mapped to ORPHAcodes.

The audience was reminded that the underlying presupposition/hypothesis to this exercise is that all the diseases in a group are covered by the center linked to the group, even when they are not listed in the expert centres application files within the sub-thematic areas (so as to include ultra-rare RD).

The limitations of this analysis were discussed:

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1) The first would be a too permissive underlying presupposition: It is important to stress that if only RD mentioned in the application forms were considered « covered », most RD would have been in a «gap». However we believe in leaving no one behind, and by using this pre-supposition patients with the rarest diseases for which there is no single specific expert center would still benefit for entering the ERN corresponding to their family of diseases because this constitutes the best available response to provide them.

2) The analysis was performed taking into consideration declared thematic and sub-thematic groups, and ERNs websites exclusively and **not considering** the center declaration when registering to Orphanet if different from ERN application.

The Results in terms of GAPS were presented.
Compared to the ORPHA classifications : 11 /24 ERNs provide either a complete/almost complete coverage

- ~10% of RD (disorder level) seem not to be covered (after exclusion of infectious and toxicants effects-related RD)
- In particular for 25/392 most prevalent RD there is no ERN out of their mapping with thematic/sub thematic groups
- While for 16/392 of the most prevalent diseases could be indirectly mapped.

⇨ Some gaps could « easily » be proposed as extensions of ERN centes but need qualitative analysis of participating centers

- Rare benign tumors (i.e. Cardiac tumors, rare benign gynecological tumors)
- Rare developmental anomalies of arteries (i.e. Internal carotid absence, Congenital renal artery stenosis)
- Rare dyslipidemias
- Rare neurological disorders
  - Rare sleep disporders
  - Paraneoplastic neurologic syndrome
  - Rare pain-related disorders (i.e. Paroxysmal extreme pain disorder, Complex regional pain syndrome)
  - Some rare autonomic disorders (i.e. Primary orthostatic hypotension)
  - Rare prion diseases
  - Rare medullar diseases (not NTD)
  - Rare specific language disorders
  - Rare miscellaneous neurological disorders (i.e. Gerstmann syndrome, Locked-in syndrome…)

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- Rare ophthalmic disorders
  - Sclera disorders
  - Endophthalmites
- Rare thrombotic diseases of hematological origin
- Rare osteonecroses and Rare osteolysis
- Miscellaneous vascular, systemic diseases: (i.e. Calciphylaxis, Hyaline fibromatoses)
- Non-Langerhans cell histiocytosis
- Rare inner-ear disorders: vestibulopathies (i.e. Idiopathic bilateral vestibulopathy, Semicircular canal dehiscence syndrome)
- Rare infertility
- Rare pregnancy-related disorders & Rare non-malformative gynecologic or obstetric disease
- Rare acquired disorders
  - of cardiac rythm
  - dermis elastic tissue disorder
  - rare hemorrhagic disorder due to an acquired platelet anomaly
- Rare urticaria
- Rare lichen planus
- Rare skin appendages disorders
- Rare skin pigmentation disorders
- Non-inflammatory vasculopathies (i.e. Segmental arterial mediolysis, Neonatal compartment syndrome)
- Others

The results in term of OVERLAPS & COMPLEMENTARITIES were presented:
In terms of diseases we observe Complete group overlaps between multiple ERNs, such as:
EuroBloodNet & PaedCan;
ERKnet & eUROGEN;
EndoERN & eUROGEN
…

For Multi-classified RD we observe overlaps of whole groups, such as Metabolic disorders (i.e. MetabERN & EuroNMD, …); but also some individual diseases such as Noonan syndrome (5 ERNs,), CHARGE (4 ERNs), …

In terms of centres we observe that the vast majority of centres pertains to only 1 ERN. But there are some examples of multi- ERNs (besides the RD Hubs). **These could be explored further to understand complementarities.**
Some examples from different countries:
Centre for rare diseases of the metabolism of calcium and phosphate: ERN-BOND & Endo-ERN
Center of Expertise for Congenital Malformations of the Intestinal Tract: ERNICA & eUROGEN
Rare Dermatological Conditions Clinic: VASCERN & ERN-Skin
Center of Expertise for Systemic Autoimmune Diseases and Primary Immune Deficiencies: ReCONNET & RITA

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PROPOSED NEXT STEPS

1) To collect ERN coordinators feedback: the lists of ORPHAcodes and of the full classifications matching thematic/subthematic groups will be now sent.

Perform a Qualitative analysis: the Complementarities analysis will also be performed based on centers' declaration and ERN coo feedback (i.e Pediatric vs adults; Diagnosis & multidisciplinary management vs interventions…)

⇨ JARDIN WP6: we will then be able to contribute this information to design pathways

Then the audience was presented with the specific tools and services developed during the OD4RD project such as:

- The Nomenclature dedicated pages on Orphadata were overall viewed 9,112 times in 2023 by 4,284 users.

Regarding the local instances of services such as the *API, « dataviz », mappings…* the audience was reminded that the ORPHAcodes API and Dataviz update follows the Nomenclature Pack release; their month usage stats were presented.For the Dtaviz tool, access by country was also presented (Fig.11).

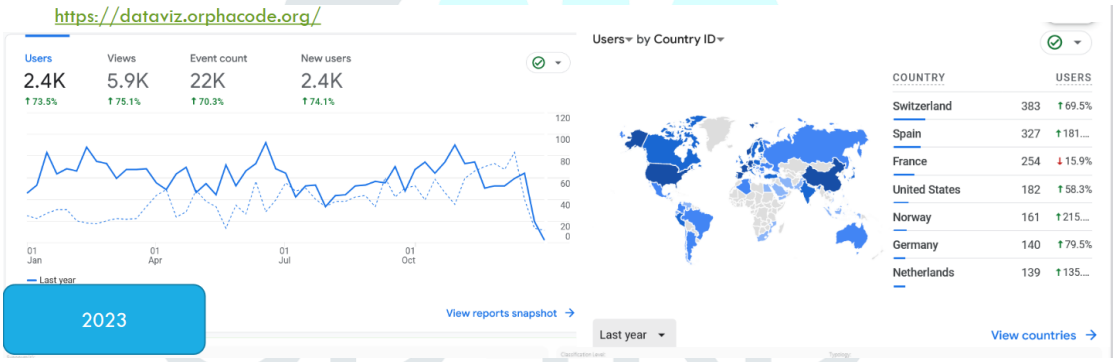

Figure 11 Data visualisation Tool use in 2023 by country.

A further reminder was done regarding these data sets which are a partial extraction of the Orphanet data base, freely accessible at www.orpha.net for consultation purposes only. The data sets are available in nine languages: English, French, German, Italian, Portuguese, Spanish, Dutch, Polish and Czech;

An online mapping service Mapping files to facilitate the transcoding is also available (fig.11).

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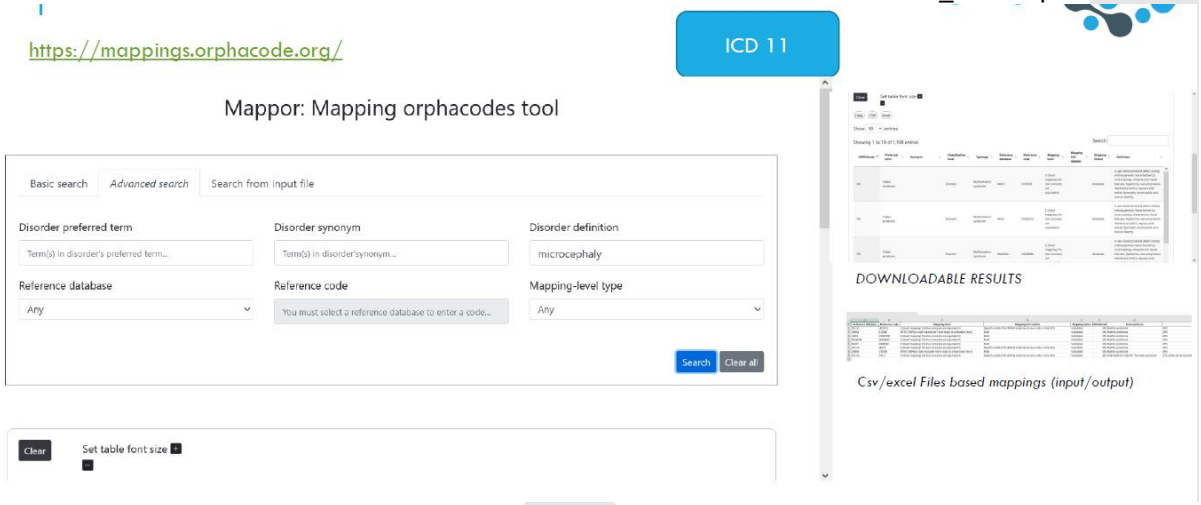

Figure 12 Online Mapping service

The Clinical Patient Management Systems (CPMS) developed by the EC for ERNs timeline was presented to the audience (Fig12)

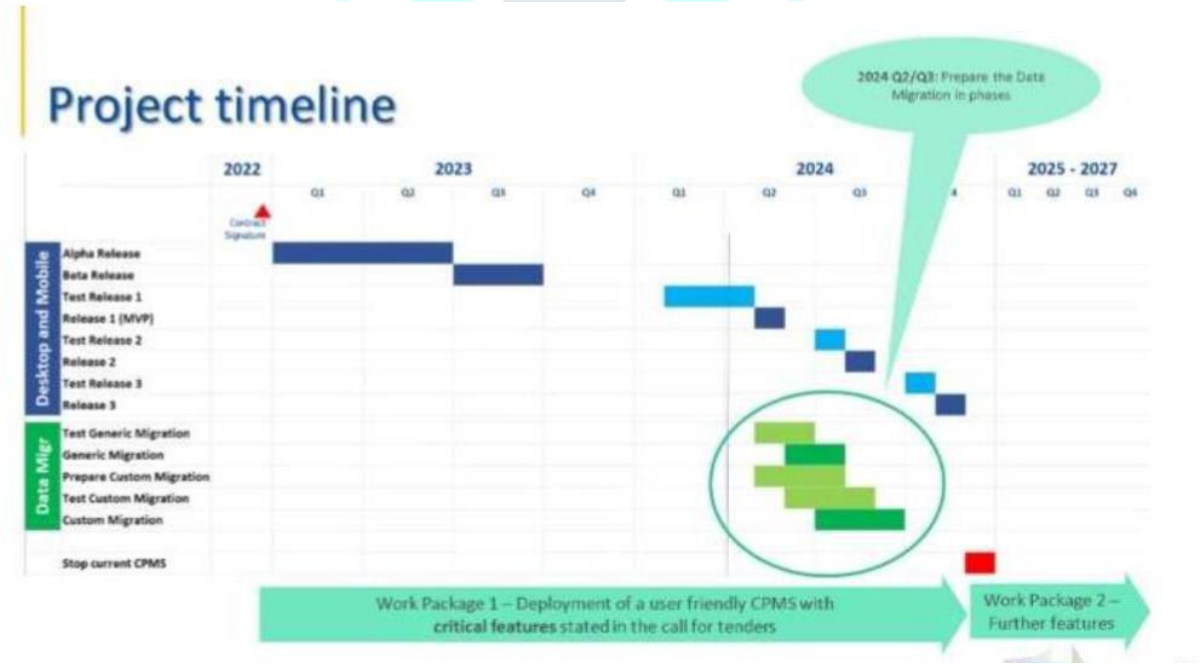

Figure 13 CPMS Project Timeline

The MVP (minimum valuable product) release was planned in the first trimester 2024 and the testing platform is available at this address: https://cpms2-acceptance.ern-net.eu.

While Data migration is planned in 2024 (including critical features deployment) and possible further functionalities will be developed in 2025.

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=> It is an opportunity for us to Explore possible interaction/integration during the 2 semester 2024

Technical feedbacks:

Content wise the https://api.orphacode.org & Nomenclature pack share the same content & same release timeline. This is true also for the Data visualisation tool: https://dataviz.orphacode.org/ as it is « plugged » to api.orphacodes.org.

=> Because of the yearly release users don't need to check for frequent content changes (N.B. this was a *desideratum* from the pilot Hospitals where pilot technical ORPHAcodes implementation was performed (first via the RD-ACTION project then via the RD-CODE project)

Also, a warning was given regarding the api.orphacode.org: it is not meant to be used by « production » front end directly as there is no SLA, no access restriction and would provide risk of high dependencies

- ⇨ it is perfect to build any « backend », and load datasets once a year!

Some users' requests received by the coordination were discussed:

"Could the Date field only be updated when some information about the code was changed, it would drastically change how many requests we had to do when checking for changes"

This would be pointless, as there are no changes in between. The good practice is to check for changes, once a year.

The difference with « Orphadata Sciences »: genes, HPO annotations, more mappings…was stressed: these datasets are updated twice a year.

Discussion:

It was noted that some IT queries reach directly the Coo team via the GitHub and they skip the National Hub, whenever this happens the relevant NH is put in copy of the exchanges.

The audience discussed whether we should include additional content in the Nomenclature pack and the tools derived by it (i.e. some hospitals would like to include genes or HPO in the datasets) or should we keep routing people to the available Orphadata datasets for genes & HPO etc. (and be clear about advantages and limitations). It will be nice to assess technical use case implementation in order to decide which way to go

- ⇨ We could plan an Hackathon/bring your own data session
- ⇨ The possibility of sundering the Nomenclature Pack from Orphadata (as a stand-alone website) for the purpose of clarity will be discussed

Some NH observe that HIS or registries where the Orphanet Nomenclature is implemented do not update annually their content: what can be done to ensure they follow the annual update?

The ARBOR tool is helpful in understanding the Classification structure of the Orphanet Nomenclature however it is an internal tool and moreover it is not aligned with the nom pack:

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content, thus it is not recommended to use it for visualisation of the classification for the clinicians as it can generate confusion

- ⇨ Need for a classification browser open to the public/clinicians

Finally during the discussion, much frustration expressed by the NH regarding the IT questions:

- ⇨ Organise a Webinar to build capacity on this matter
- ⇨ Provide more educational material
- ⇨ Make available a Best practice in wiki on how to use the tools

Consolidate and Expand national Orphanet nomenclature hubs (WP4):

The objective of the WP4 was presented: to ensure coordinated local support for the local implementation of ORPHAcodes in national HCPs hosting ERNs and national HCPs linked to ERNs by establishing a coordinated network Orphanet national nomenclature support hubs.
The Network of Orphanet Nomenclature National Hub is coordinated by BfArM and Karolinska while the scientific coordination of NH activities is ensured by the Orphanet Coordinating team (Inserm).

The next presentation revolved around the activities carried out in the frame of the Coordinated Helpdesk and support for local implementation of ORPHAcodes in national HCPs. These activities are carried out the Scientific Coordination of the national nomenclature hubs network, the coordinating team continues to provide the national Orphanet hubs with central support and material ensuring the same level of service quality in each country.

➢ A regular Training –or-trainers (TfT) program
➢ Monthly Nomenclature & Coding Open Sessions
➢ 24/7 Central GitHub helpdesks and wiki page

The TfT programme consists of a 3h Basic training program and a 3h Advanced Training program:

- The basic module was held on the 12/09/2023 and gathered 26 participants from 14 National Hubs (of which 6 new NH). A Quiz was carried out at the end of the session and only 16 answers were received (12NH, of which 5 of the new NH)
- The ADVANCED training module was held on the 19/09/2023 and gathered 32 participants from 18 NH (of which 5 new NH). A quiz was carried out at the end of the session and only 12 answers were received (9NH, of which 3 of the new NH)

A Satisfaction Survey was launched regarding these trainings and 12 answers were received (28/09/2023): overall 11/12 of the participants were satisfied or very satisfied and 1/12 was unsatisfied.
The respondents found the presentations more structured, understandable, updated presentations when compared to the 2022 material. They particularly liked the live tools demonstration and website navigation, the nomenclature alignments presentation, the examples and use cases and the new content (new tools), the exercises/quiz. The respondents evaluated a good communication for sessions organization.
When asked what they disliked, some mentioned that the pace was too fast; the short time left for discussions, the short time left for use case discussion (advanced training), the short time on some topics, the short time for the quiz as well as the little interactivity between/with participants (in particular for new NH).

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The respondents suggestions to improve the TfT were the following: more practical examples; more time for specific topics; review training presentations; revise wording of quiz questions; avoid change of schedule last minute; remove ERN collaborations from the TfT (keep it for Open sessions), sharing already diffused national trainings, more time for training.

All the relevant feedback will be considered for the next TfT organisation.

Nomenclature & coding Open sessions: these are held monthly as 1h sessions, are open to National Hubs and they revolve around discussions on burning topics the NH may face during their day-to-day work.
8 sessions have been held so far and the themes are indicated below:

- o #1 Oct 23, Nomenclature pack 2023 overview, coding non-chronic RD
- o #2 Nov 23, Scientific content updates, decisional tree to trace missing ORPHAcodes in HIS, Orphanet news
- o #3 Dec 23, Orphanet e-learning module EN, coding prenatal diagnosis
- o #4 Jan 23, GitHub overview
- o #5 Feb 24, Journal Club Angin et al., (2024) "Coding undiagnosed RD patients in HIS, recommendations from RD-CODE project"
- o #6 Mar 24, Coding healthy/asymptomatic carriers
- o #7 Apr 23, Coding rare cancers
- o #8 May 24, new Orphanet disease meeting report 2024, WP4 & Orphanet news

Central helpdesk
This OD4RD ticketing tool for National Hubs was launched in July 2022. It allows to track and assign the issues around ORPHAcoding received by the National Hubs and at central level.

As of today, there are 16/19 National Hubs on GitHub. The 3 not having implemented a national Github organise the helpdesk via telephone and mail and correspond to 1 senior hub (i.e. Hubs having started their activity with OD4RD in 2022), 2 junior hubs (i.e. having started their activity in 2023 with OD4RD2)).

Currently a total of 219 GitHub issues are tracked, of which

- 166 issues are closed & 53 currently open (6 months delay from? creation)
- ⇨ Of these 190 issues from NH to Orphanet coord. Team + ~30 issues/creation suggestions from external collaborator (NORD)

More precisely, for OD4RD2 year 1 [April 2023-March2024] there are 111 issues from NH to Orphanet coo.

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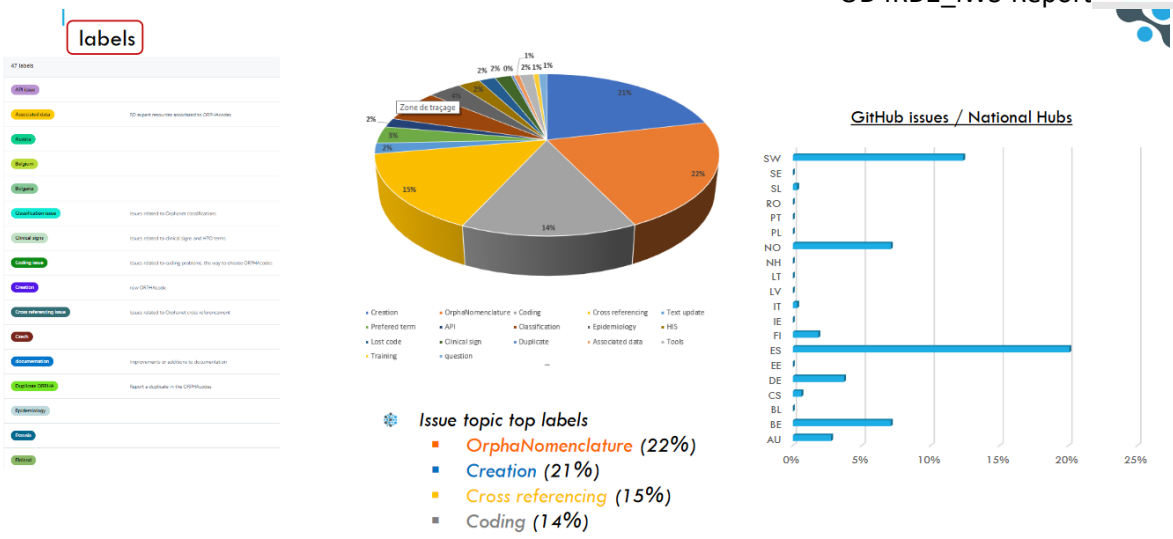

Figure 14 GitHub issues per categories and per National Hub since 2022

GitHub Wiki (FAQ)

To provide a sustainable and homogeneous, standardised support, a 'Questions and Answers' section within the GitHub has been developed by the coordinating team based on users' questions. It provides standardised and general answers among 7 main topics: alignments with other terminologies, coding recommendations, education and communication, epidemiology of RD, ORPHAcodes and nomenclature, Orphanet classification and Orphanet tools. This Wiki page was developed during the OD4RD pilot year and it is continuously upgraded.

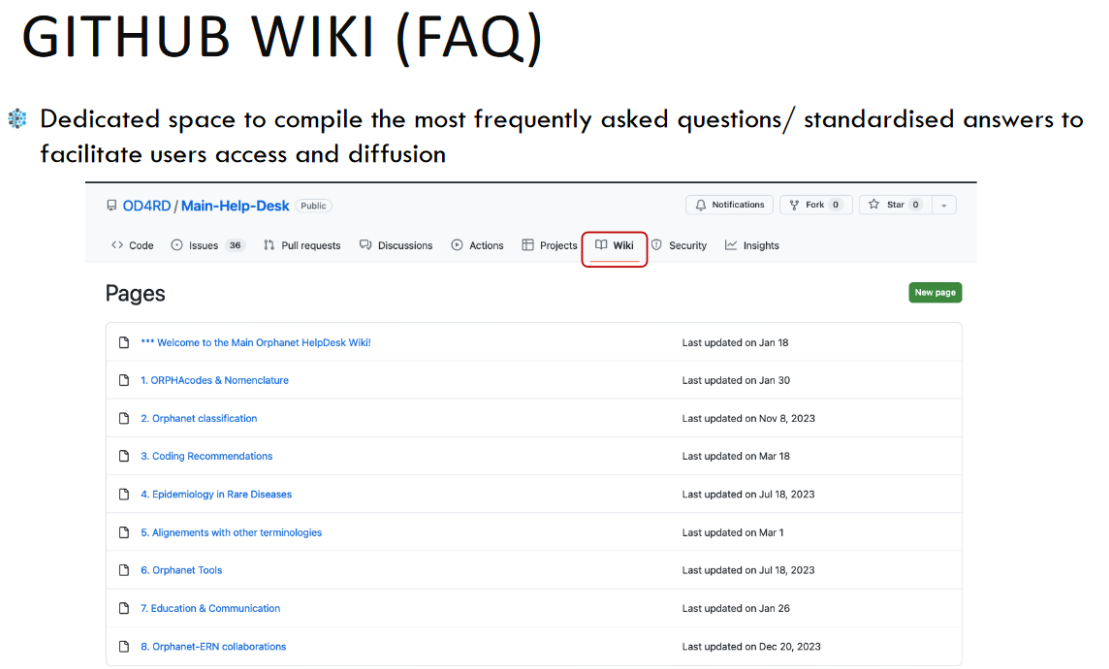

Figure 15 GitHub Wiki/FAQ: a dedicated space to compile the frequently asked questions standardised answers to facilitate access and diffusion

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New & Next

More dissemination will be organised around the GitHub tool! We want to reach more inter-intra national hubs / external community posts

- ⇨ a Wiki section about Orphanet technical queries for IT will be added
- ⇨ a Training for trainers will be organised in the fall 2024/early 2025
- ⇨ An Orphanet e-learning module (English version) will be available online (Norwegian platform) for all national trainings
- ⇨ An ORPHAcodes best practices Advisory Committee will start working during the fall 2024: a small medical experts group (M.D., coordinator clinicians, coding clinicians) from Orphanet National Hubs countries (OD4RD2) are invited to join Orphanet scientific and medical coordination members, for consultation and ad hoc meetings only, to provide advice on specific best practices through clinical use cases (Rare diseases list) & Encourage best usage of ORPHAcodes in their networks and national HCPs

To ensure that the same quality support is provided in each country, a coordination support to the national hubs work is organised by BfArM & Karolinska University to coordinate the work and ensure the project deliveries.

The main highlights of the activities carried out were presented such has:

- ➢ 9 meetings of 90 minutes each organised and held
- ➢ participation ratio of 93,4 %
- ➢ 19 presentations of national hubs
- ➢ 5 presentations on national ORPHAcode usage
- ➢ 14 presentations on national action plans
- ➢ presentations on TfT program, GitHub tool, technical support, documents & training material, promotional material, ERN collaboration

The main deliveries carried out during OD4RD2 so far were presented:

- MS12: NATIONAL HCP-SURVEY: 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.
  At least 75% of national hubs have performed a survey and have presented written results. These have been provided to the ExCom through the projects
  internal website. 16 out of 19 national teams provided a survey report (79%) and 11 teams performed a QC of the data results.
  Preliminary results were presented during the meeting: these results already allow to confirm that our strategy responds to the « field » barriers. The survey results will be complemented with the JARDIN survey results, to obtain a complete picture of practices by ERNs 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, the National Hubs work and to discuss around ERN data strategy so as to refine the project's national and transversal actions to improve harmonisation. During the discussion it was agreed that an additional effort needed for the final analysis and to ensure quality of data.
    - All NH should provide the info on number of units contacted (deduplicated)

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• All NH should indicate the number of answers received per question so as to make sure that the adding up of the single categories matches the total number (some discrepancies were observed). If some answers are discarded, it should be indicated
• For the analysis by ERN: Deduplicate if several answers from the same unit and report on eventualdiscrepancies)

A final report of this analysis has been issued following the quality control performed by 11 teams , and it is available here: https://od4rd.eu/03-deliverables/OD4RD2_ERN-Survey_11QC%20analysis_VF.pdf

The Lesson learned for the next survey were also assessed:

- ⇨ a Glossary should accompany it so has to make all concepts clear
- ⇨ Absolute numbers and percentages should be indicated to facilitate reading, also for histograms, showing the values of each can facilitate reading
- ⇨ Multiple choice questions results can be presented as Venn Diagrams to have a full picture
- ⇨ Always indicate the total of respondents by question as these may vary (and also provide a quick quality control)

- END USERS' STATE OF PLAY SURVEY: a report compiled from the survey on the state of play in each of the Orphanet Nomenclature National Hubs was updated in 2023

- **Participating countries:**
  Austria, Belgium, Bulgaria, Czech Republic, Estonia, Finland, Germany, Ireland, Italy, Latvia, Lithuania, Netherlands, Norway, Poland, Portugal, Slovenia, Spain, Sweden and Switzerland*

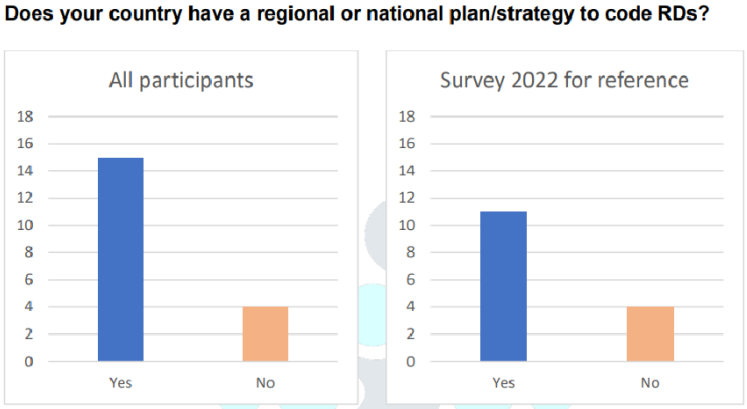

Figure 16 Does your country have a regional or national plan/strategy for RD?

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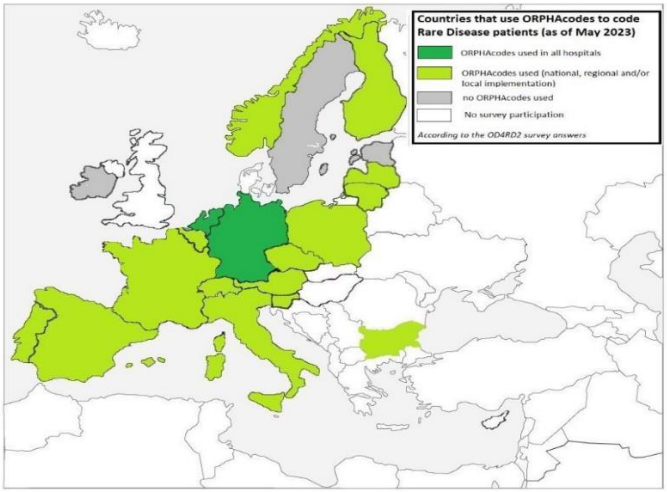

Figure 17 Countries using ORPHAcodes to code RD diagnosis

In two participating countries ORPHAcodes are used to produce data or statistics for RD in all hospitals. In most cases ORPHAcodes are used in registries, either national registries (10) and/or regional registries (3). In most countries ORPHAcodes are used in centers of expertise for RD, either nationally (5) or regionally (7). In three countries ORPHAcodes are not yet used for that purpose, but two of them are preparing for implementation. Among the five countries that participated in 2022 that did not use ORPHAcodes at the time of last year's survey, only one is not yet using ORPHAcodes, but is preparing to launch a national registry using ORPHAcodes in 2023.

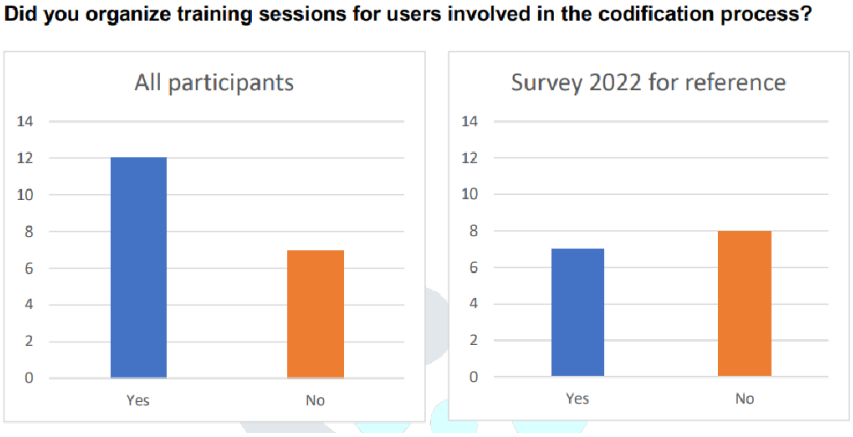

Figure 18 Did you organise training sessions for users involved in the codification process?

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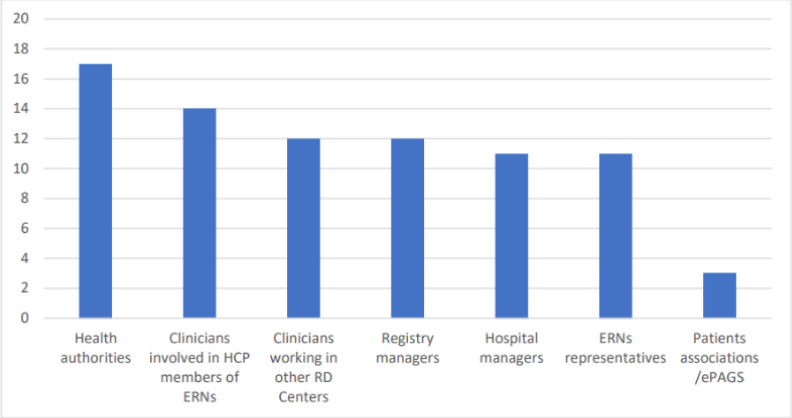

Figure 18: Crucial need of involvement of the following stakeholders for a successful ORPHAcodes implementation

Figure 19 Crucial need of involvement of the following stakeholders for a successful ORPHAcodes implementation

Main drivers identified for successful ORPHAcode implementation:

- ⇨ Effective training activities, performed by the national teams and Orphanet experts
- ⇨ "local" helpdesk managing requests from users during the implementation process
- ⇨ convincing national health authorities of the benefits of ORPHAcoding to introduce or expand it
- ⇨ providing IT expertise or a dedicated tool to facilitate the implementation of ORPHAcodes into health information systems
- ⇨ sharing of experience, useful information material and documents between participating national teams essential to successfully achieve the project goals

- ESTABLISHMENT, DELIVERY AND REPORT OF ACTION PLAN FOR EACH NATIONAL HUB: National action plans, based on the state of play in the country, have been written and are provided as a PDF document to the executive committee (ExCom). BfArM collected reports from all national hubs and compiled them for the ExCom in a PDF document.
- Mid-term REPORT ON LESSONS LEARNED: a compilation from the items addressed during the WP4 monthly meetings and from the FAQ-collections. This report should be used by other countries starting National Hub activities and implementing ORPHAcodes. The list of FAQs can also be used to enhance the Orphanet website (30th March 2023).

The pooled indicators of all NH were presented:

National helpdesk GitHub accounts: 15	15
Number of tickets received: 299	299
Type	➢ Coding: 135 ➢ Nomenclature: 97 ➢ Others: 65
Submitted by	➢ Clinicians: 126

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	➢ Hospital management: 3 ➢ Coders: 98 ➢ IT personnel: 20 ➢ Others: 48
Number of demands/tickets answered by NH in autonomy:	178
Demands/tickets forwarded to coordination	121 (72 via GitHub)

Table 9 Indicators (pooled for all NH) related to helpdesk activities

In terms of ARTICLES/COMMUNICATION, PROMOTIONAL MATERIAL produced by the NH we have:

- A Orphanet nomenclature Quiz (Belgium)
- ORPHAcoding scientific session / National conference for rare diseases and orphan drugs (Bulgaria)
- ORPHAcode information card (Finland)
- Training video “Coding of Rare Diseases” (Germany)
- Media communication and RD articles (Latvia)
- Flyers and leaflets (Sweden, Italy, Ireland)

Then the audience was presented with the work that is ahead of us:

- MS 15 - White-paper on implementation issues of ORPHAcodes in relation to other code systems (March 2025)
- D4.4 Annual Action plans Y 2-3 (June 2025)
- D4.5 Final Report on lessons learned (December 2025)
- Task 4.2: workshop with hospital managers (see day 2 discussions)

The Lessons learned by the NH work was presented, it was compiled from the analysis of the NH action plans, the above-mentioned deliveries and the discussion during WP4 meetings.

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- Based on the collected lessons learned
- Big difference in starting position from country to country
- Some countries = further in the implementation process than others
- Support from authorities and a clear structure = important for successful implementation
- Within lessons learned = common denominators
- Many of the countries = face the same problem, despite the varying conditions in each country.

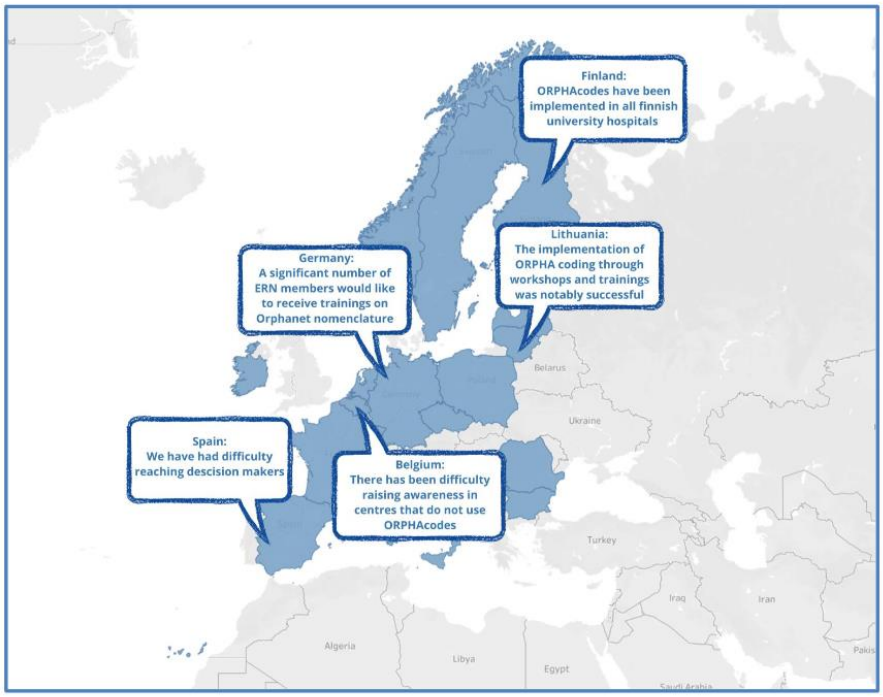

Figure 20 ***Feedback on ORPHAcoding national situation by NH***

- In regards to ORPHAcoding implementation it appears that official support is key as well as the existence of incentives
- The structure of RD care at national level also has a great impact on the choices made (fig 20)

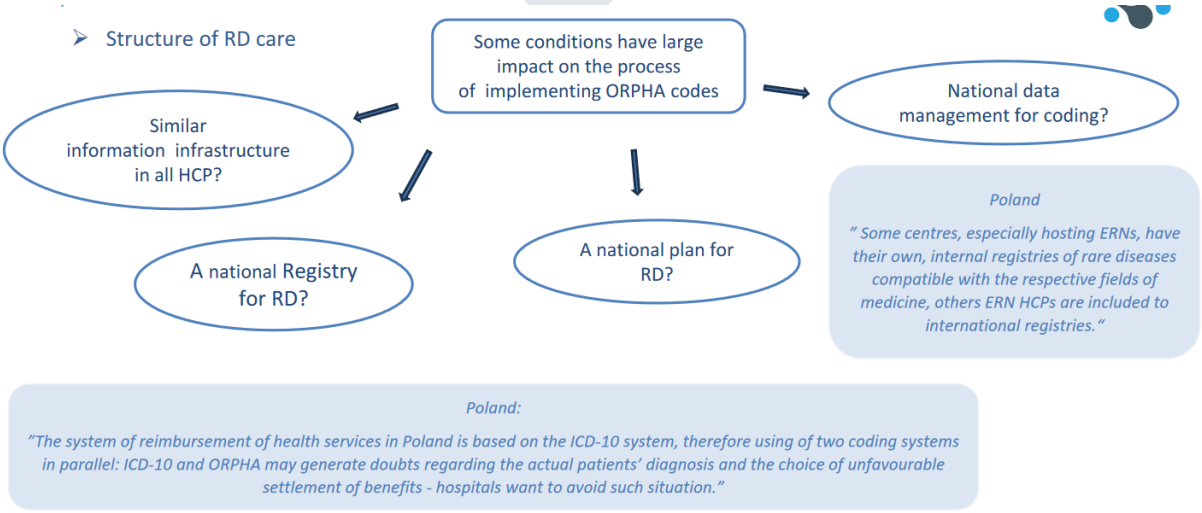

Figure 21 ***Conditions that have an impact on ORPHAcodes implementation***

- Another key factor would be the possibility of expanding upon existing framework
- A recurrent topic is the need to creating incentives for attending trainings and ORPHAcodes usage
-

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Some lessons learned on how to overcome obstacles to facilitate ORPHAcodes learning and usage were discussed (fig21)

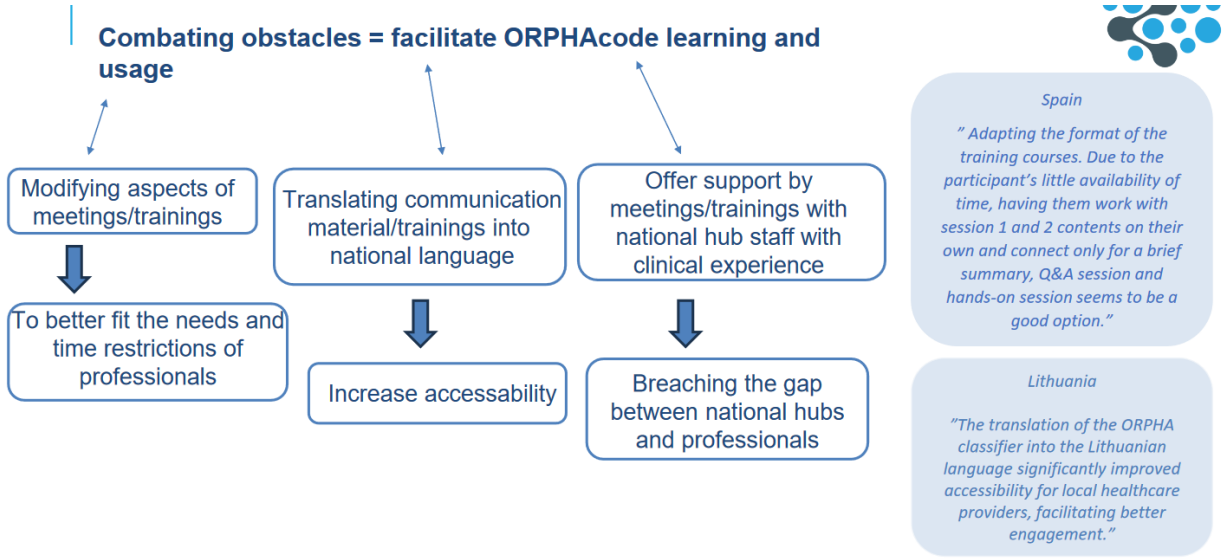
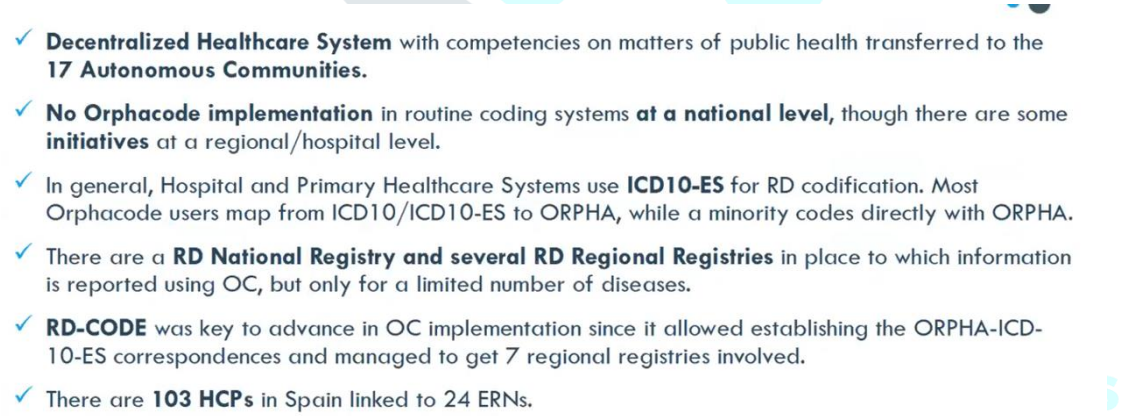


Figure 22 *Overcoming obstacles*

A presentation around a success story was given by the Spanish team:
First an overview of the Spain context was provided:



The team was the pilot having launched an ERN units survey already in OD4RD and inspired the collective work launched in OD4RD2.
They observe an increase in respondents since the initial survey (50 in 2022 vs 81 in 2024)

In terms of trainings, the team started with the Catalonia Region and the Madrid region has they have the highest number of HCPs and these 2 regions are the one that officially support ORPHAcodes implementation.
they have had 198 participants, belonging to 20 different ERNs from 20 hospitals, 8 Regional Health departments and 5 RD registries (fig 23).

And have already a planned course and several requests from other hospitals

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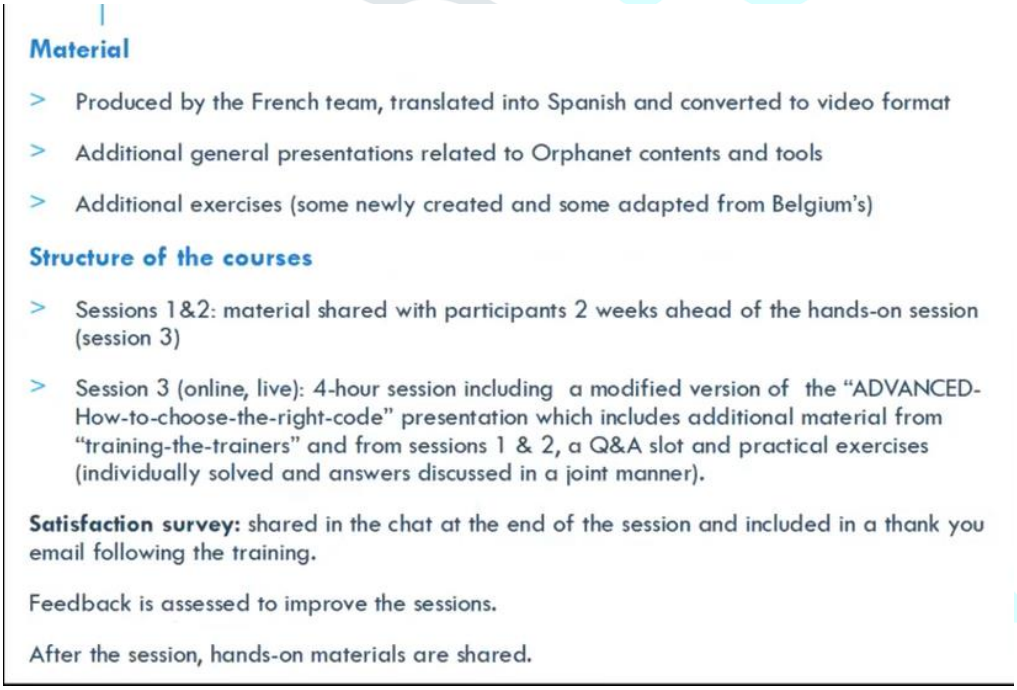
Figure 23 Orphanet Nomenclature National Hub Spain Training activity

The NH has also translated the content and adapted the structure of the course according to the users 'needs:



Finally, the team compared the difference between the two approaches top-down, bottom up which each need different strategies, and presented the lesson learned from these experiences. (fig. 24).

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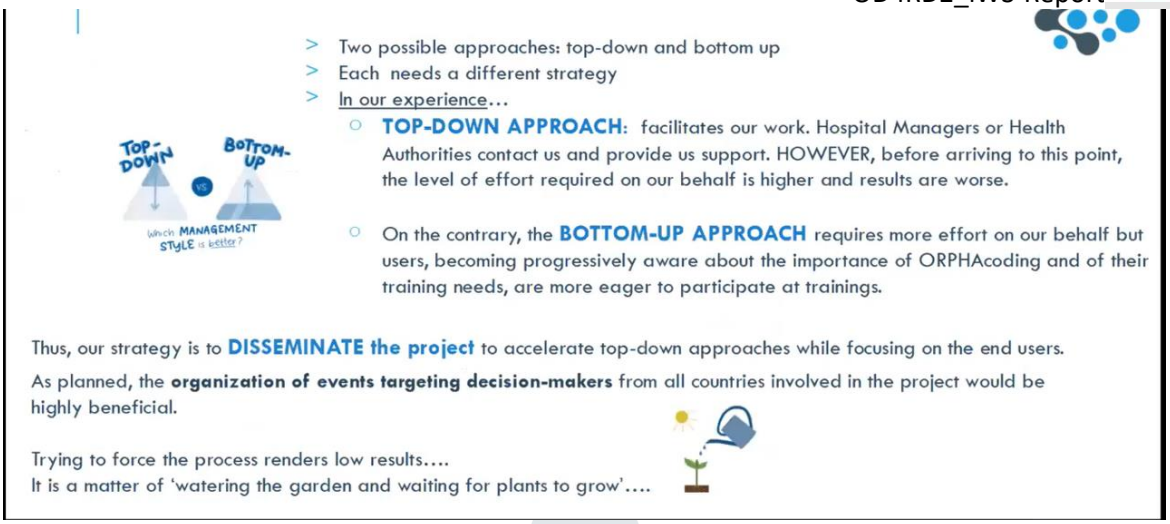

Figure 24 Lesson Learned regarding trainings Spanish NH

A challenges story was also presented by the Belgium National Hub:
Firstly, the audience was reminded of the situation in Belgium in terms of coding:
The officially designated Genetic centres send the data to the Belgian Central Registry for RD (N.B. not all RD expert centres), most send data via ORPHAcodes but some use other terminologies fig.25.
However, registrations concern roughly 2% of the RD population.

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

Figure 25 Coding System used to encode the RD diagnosis in the Genetic centres

The ERN survey also confirmed that overall ORPHAcodes are underused in ERN units also as the majority of respondents said they do not use ORPHAcodes to send data to the ERN coordination.

Finally, the obstacles identified were presented:

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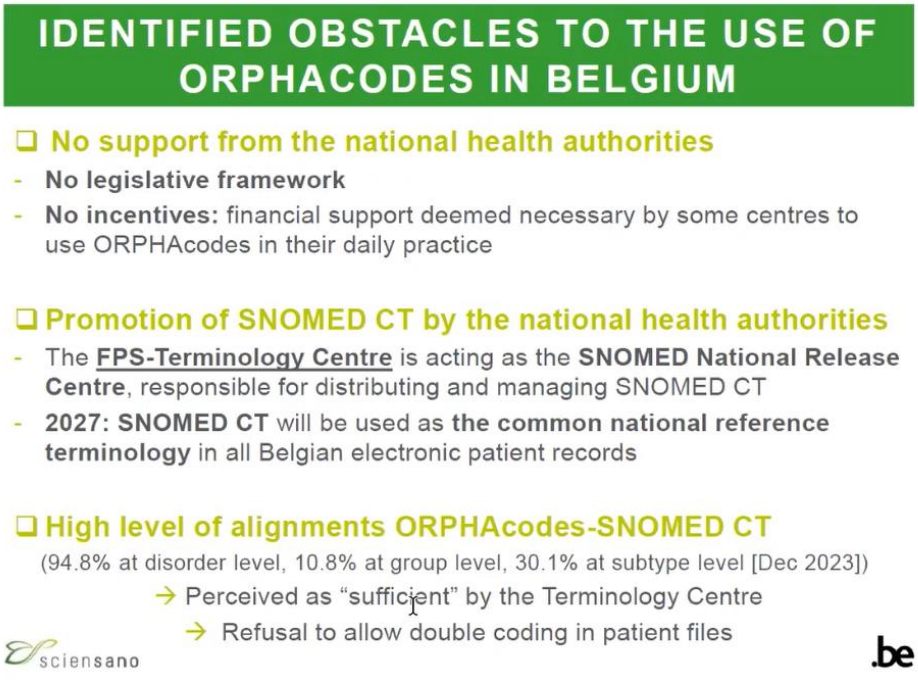
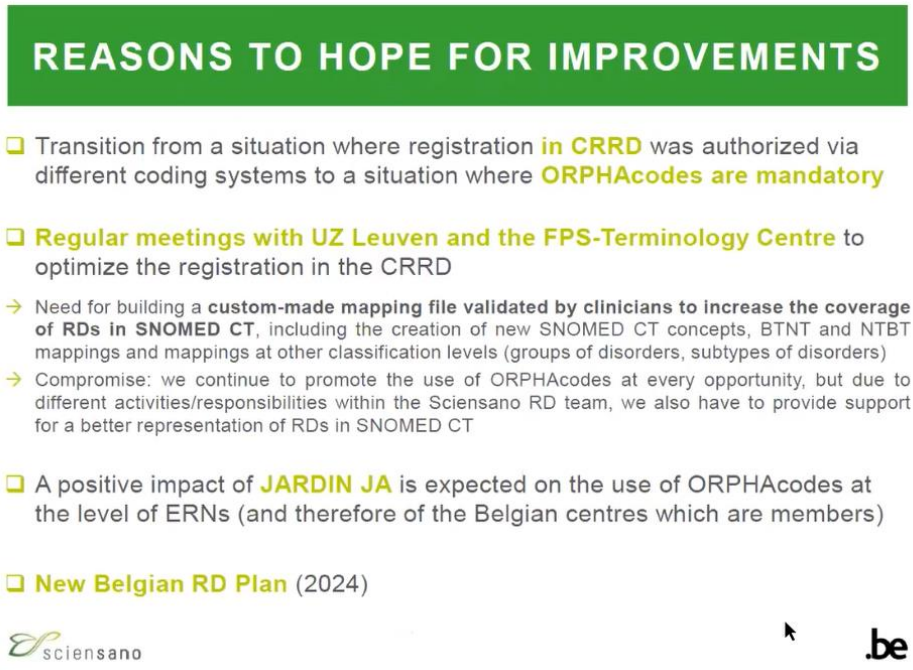


Followed by the reasons to hope for improvements:



Discussion revolved around the EC support for implementing SNOMED CT and reticence of the MS to use OC in complement for RD diagnosis as the coverage in terms of alignment OC-SNOMED CT is pretty high.

It is true that the EC covers partially the SNOMED CT license, however costs linked to the implementation of SNOMED CT are high and this is the reason maybe some countries do not want to invest in implementing an additional coding system at the source.

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The problems are known:

- By using SNOMED CT, you will never have 100% coverage => additional transcoding work will be needed
- Delay in adding new RD as the alignments are provided by OR => additional transcoding work will be needed
- Not possible to analyse by RD or groups of RD as it is a generic terminology not RD specific and with no hierarchy => additional transcoding work will be needed

Transcoding will be needed for Registries so the work will be carried out twice & additional work/effort will be needed also for data accuracy because all transcoding needs to be validated, in short, to have accurate and comparable data for data sharing at the EU and ERN level they will need to invest more resources for the exploitation.

Also, it was noted that the additional mappings of SNOMED-CT to ORPHAcodes (i.e missing codes, group codes etc.) will not be carried out by the coders but by highly specialized experts, who will not have the time nor the resources to do so.

It is important to highlight whether the country either chooses to implement the OC in the HIS and this as a cost or chooses to invest in transcoding, which also as a cost.

- ⇨ We need to document this economic aspect (what are the secondary level of expenses if ORPHAcodes are not available in the HIS for RD diagnosis coding) as financial risk is a strong argument for decision makers
- ⇨ We need to explain in simple terms what are also the costs linked to having these two terminologies both available in the HIS/HER
- ⇨ We also should document the clinical risks
- ⇨ We need to invest in JARDIN so to have an EU recommendation on the matter so that ORPHAcoding can be incentivised in National Plans

WORKSHOP: STICKY NOTES SESSION

1. During this interactive session, all the teams were invited to brainstorm around 3 main topics:

- What have been your positive experiences during project year 1? (fig.26)
- What challenges did you face during project year 1? (fig.27)
- What are your expectations for project year 2? (fig.28)

Thanks to this exercise many interesting ideas emerged from the collective brainstorming and have been added in the OD4RD2 action plan (see section *Wrap up-take home messages*), also the ideas that could not be discussed during the meeting because of time constraints will be further analysed and discussed during the forthcoming WP4 monthly meetings.

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Figure 26 ***What have been your positive experiences during project year 1?***

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Figure 27 What challenges did you face during project year 1?

Figure 28 What are your expectations for project year 2? (fig.24)

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There are 3 ways to raise awareness at a higher level

- ⇨ JARDIN joint action via lobbying and capacity building with competent authorities
  Different incentives can be organised by National authorities:
  France: to address the lack of time of clinicians who could not contribute to the Registry data managers salaries are covered by a specific funding via the NP.
  Austria: the NH worked with the Ministry of health and now via a short paragraph brought into the law it is mandatory for the hospital to send info coded with ORPHAcodes for their reimbursement, therefor everyone is onboard.
  Many countries: patient organisations have been a great and effective support and they can leverage Ministry of Health

- ⇨ JARDIN Joint action: demonstration that the ORPHAcodes help in making practical and cheap care pathways, this way everyone will be onboard.
- ⇨ Our new recurrent point @BoMS (however the person in charge in the health data strategy is not necessarily the person attending the BoMS)
- ⇨ EU RD plan as Overall EU should incentivise technical implementation of OC (as they do for SNOMED-CT) as it will be an epidemiological bias if only few countries send the data, it will not be representative

Day 2

Participants :

Orphanet Nomenclature National Hubs: Austria, Belgium, Bulgaria, Czech Republic, Germany, Finland, France, Ireland, Italy, Netherlands, Norway, Portugal, Poland, Slovenia, Sweden, Switzerland, Spain.

Opening

Recap of 1st day discussions and mitigating actions and unplanned actions to include in the roadmap (see next paragraph)

TRANSVERSAL ACTION: EVENTS FOR EXTERNAL PARTICIPANTS

• ERN Day will be organised by the end of 2024:

To raise awareness about ORPHAcoding benefits, around the National Hubs Work and to discuss around ERN data strategy so as to refine the project's national and transversal actions to improve harmonisation.

Proposal to do it back-to-back with the next ERN/BoMS meeting in September (under discussion) and the proposed agendas were discussed (according to the available time).

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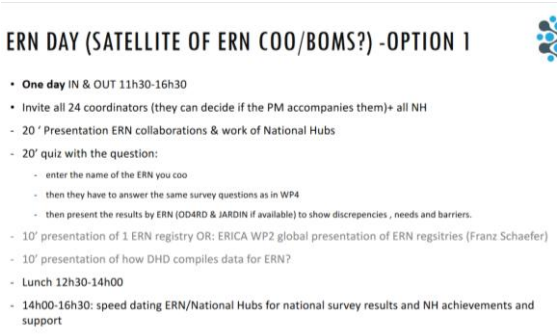

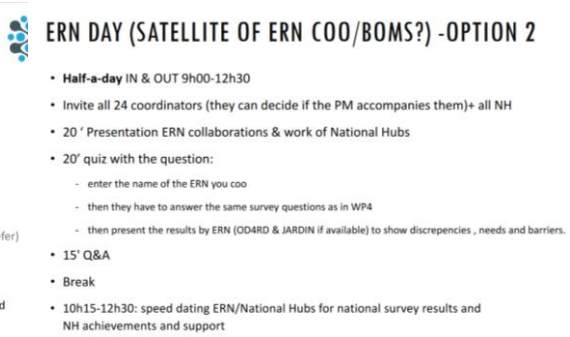

Hospital manager day will be organised in 2025:

The Lessons-learned from National Hubs who reported having met with Hospital managers were presented:

Austria: the deputies were targeted in the biggest Paediatric Hospital; these managers were initially hostile but the NH had the support of the clinicians. Regular *de visu* meeting with 15-20 persons were organised and after a few sessions once they started to know each other it got better and now they are ORPHAcoding, the managers have made it mandatory. They would also want OC everywhere i.e. discharge letters but it was difficult form a technical pov and this was difficult to understand for them. They do not make the difference between ORPHAcoding and technical issues.

- ⇨ It is very important to identify the relevant person(s): in JARDIN we have as target the managers of the second level i.e. responsible for coding, responsible for IT
  - list of identified contact via JARDIN : We can ask JARDIN to send an invitation
- ⇨ It is very important to connect in person

Norway: the same strategy was used, in persons meetings with the medical manager of the Oslo University Hospital. The new HER will be available to all the south-eastern region of Norway

=> The support of Network and RD experts has been crucial

Poland: big paediatric department with 7 different ERNs. Participants of the meeting were the second level managers: Scientific manager + International Coo+ IT specialist manager + Digital medicine manager

They wanted to have a HIS compatible with ERN requirements in terms of registry & create a registry including for RDs

Then the proposed agenda for the Hospital Managers day was discussed:

o State-of-play of ORPHAcoding

o Background: Needs for ORPHAcoding for RD

o Testimonials: 2 Hospital Managers from 2 different countries (benefits, barriers, experience)

o What OD4RD can do for you: support proposed by national hubs.

o Ending survey: seeking for commitment

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Then followed an interactive session: all the teams were invited to brainstorm around the agenda of these events and the target participants

Thanks to this exercise many interesting ideas emerged from the collective brainstorming and will be implemented:

- Target participants: Second level managers
- Use JARDIN mailing lists
- Onboarding of EUHA and ECHO
- Explore co-branded event JARDIN-OD4RD

Collaboration with JARDIN

Oprhanet is involved in many activities of JARDIN (fig.29), in particular within workpackage 8.

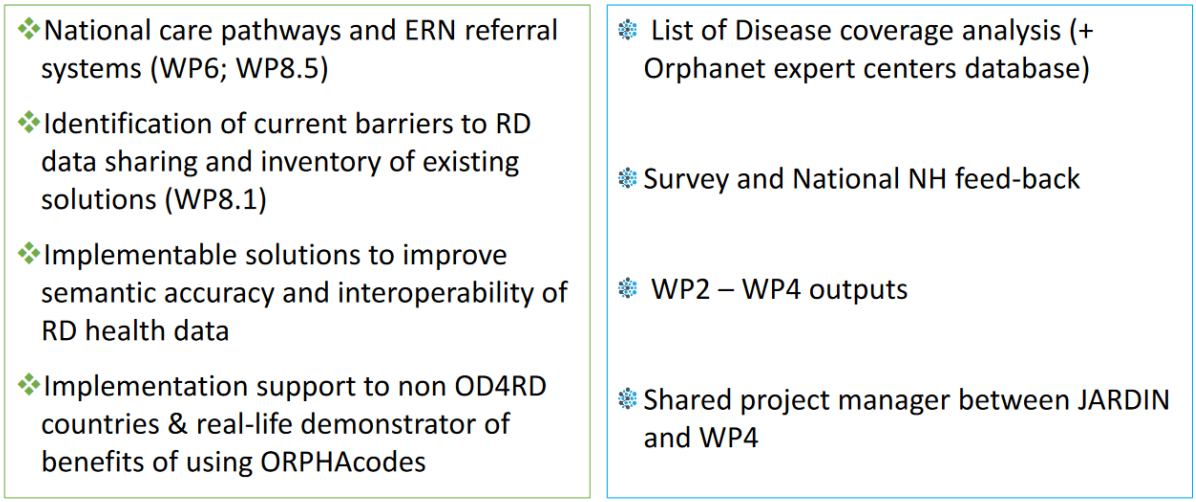

Figure 29 Orphanet implication in JARDIN activities

An OVERVIEW OF WORK PACKAGE 8 activities was given to the audience.

The Leaders of this WP are the French Ministry of Health & Dutch Ministry of Health, Welfare, and Sport. The Key Topic is Data Management and its Objective is to develop recommendations ensuring the interoperability of data structures on MS level (local, regional, national) and ERN level. Its methodology is described in fig. 30

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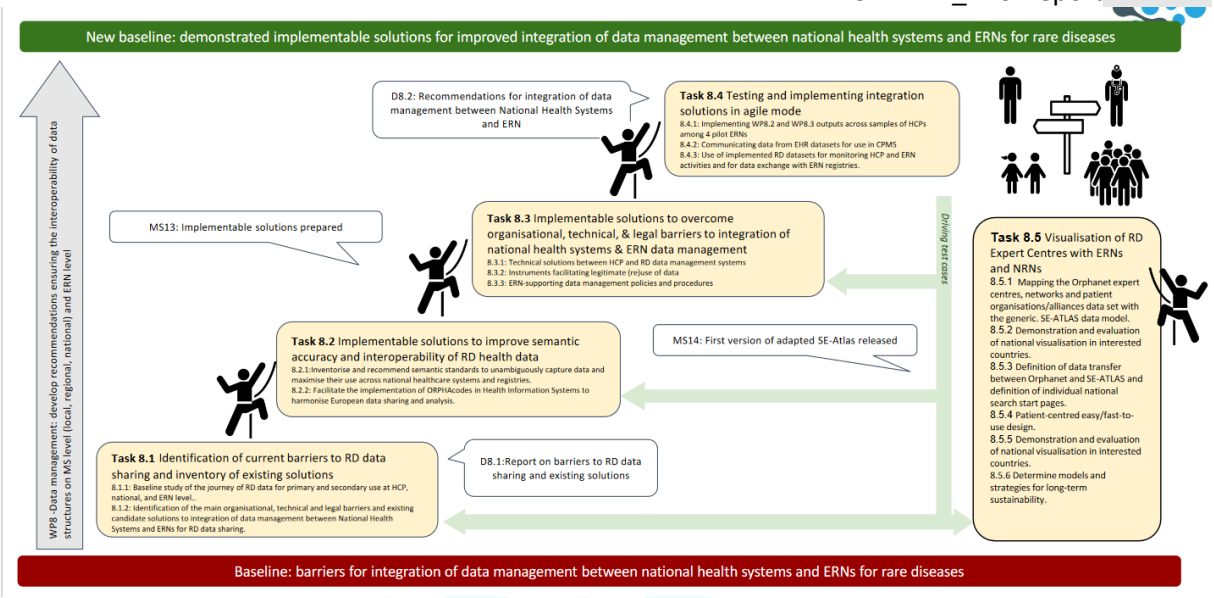

Figure 30 JARDIN WP8 methodology

Orphanet is leading task 8.2 which consist in identifying Implementable solutions to improve semantic accuracy and interoperability of RD health data. The task has been organized into 2 subtasks:

Task 8.2.1 Propose/develop functional specifications for the implementation of a standardised common RD dataset in health information systems (HIS).

This will consist in Inventorise / recommend semantic standards to capture data for RDs, building on (i) previous projects (e.g. EU projects, X-eHealth network, EJP- RD, etc.) (ii) agreed minimum data sets / core data elements (iii) Results of surveys and to Maximise use of semantic standards across national systems and ERN Registries

Two workshops with MS representatives will be organized to (i) Discuss the MDS recommendations, with focus on 1ary and 2dary use , (ii) Reality check for recommended specifications, (iii) provide Feedback from demonstrators (Task 8.4: demo of benefits of ORPHAcoding)

- Compile recommendations on MDS and CDEs
- Compile existing implementation solutions
- 1st version of semantic implementation guidelines M12

Thus, the first step will consist in solving the RD invisibility (fig 31), Moving towards general semantic interoperability for RD

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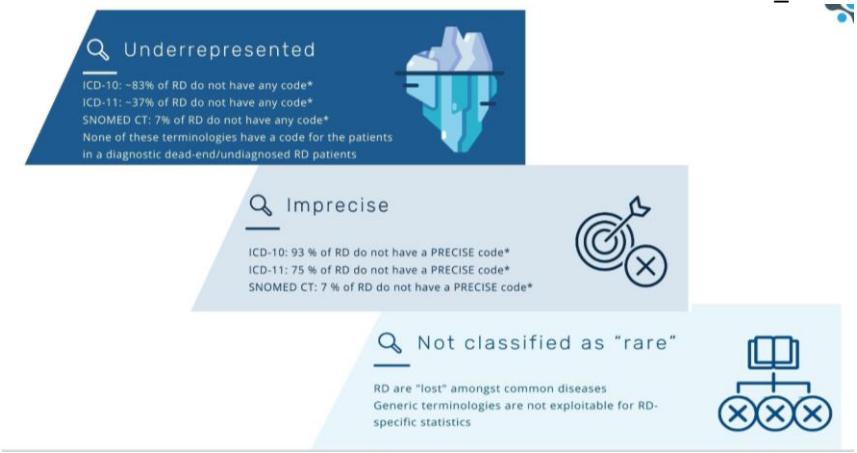

Figure 31 Solving RD Invisibility

The Second step will consist on agreeing on data elements

- EUCERD Joint Action (2015):
  - Agreement on a Minimum Data Set (MDS) for RD registries
  - Based on French MDS
- Common Data Elements
  - Subset of MDS recommended for RD registries
  - EU Platform on RD Registration (JRC)
  - Patient-centered data model elaborated by the EJP RD
- French Minimum Data Set (2013)
  - Elaborated by a multistakehoder committee with the input of 131 Centres of expertise
  - Intended for data collection in health records
  - Now deployed in EHRs (*dossier patient informatisé*) and feeding the National registry (BNDMR)
  - Technical format: CDA r2 3rd level
- EHealth Network (2023):
  - the electronic exchange of health data under Cross-Border Directive 2011/24/EU: Patient Summary

Task 8.4 “Testing and implementing integration solutions in agile mode”. 3 pilot projects will ensure that the deliverables of 8.2 and 8.3 are relevant and can be fully deployed (fig 32). This task is led by APHP and Orphanet co-leads task 8.4.3.

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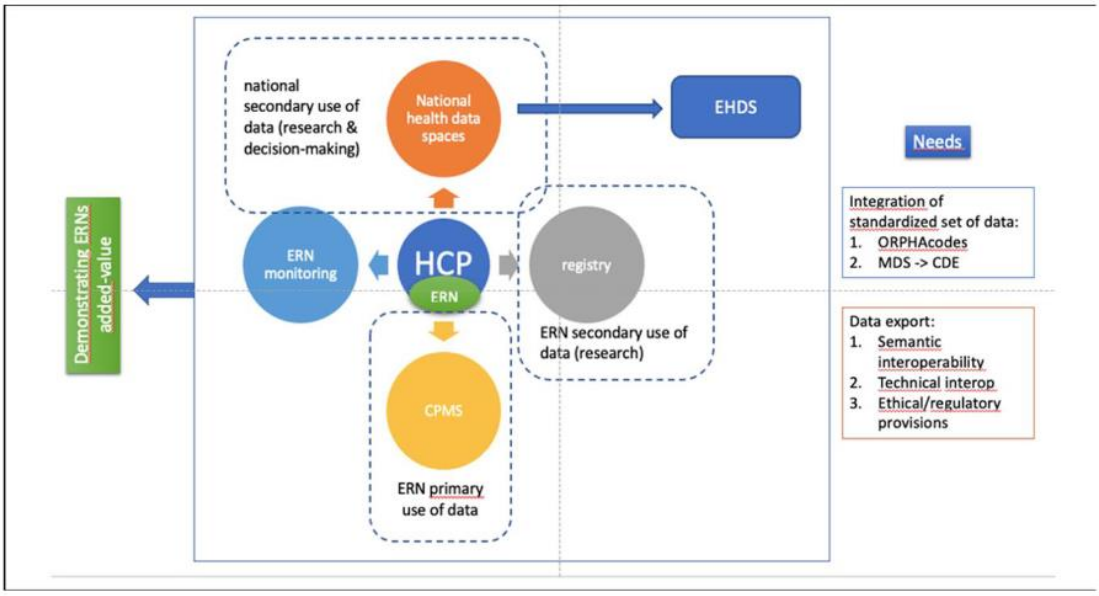

Figure 32 Task 8.4 methodology

• 8.4.1: Implementing T8.2 and 8.3 outputs across samples of HCPs among 4 pilot ERNs (EpiCARE, EURO-NMD, ERN-RND, ITHACA).

• A call for volunteers will be launched among HCPs involved in the 4 ERNs and the corresponding National Health Data Spaces (NHDS) using T8.1 to ensure that selected HCPs sufficiently represent the variety in Europe.

• 8.4.2: Communicating data from EHR datasets for use in CPMS. In parallel to the development of the new CPMS, data required for case discussions (i.e., data from the RD common data set, DICOM files, etc.) will be mapped to available sources in EHRs of ERN members and national clinical case consultation systems. A transfer of data from EHRs of HCPs into the new CPMS will then be piloted in 6-7 HCPs from 4-5 countries, in addition to transfer from the German clinical case consultation system KONSIL-SE. Data transfers will be adjusted as the new CPMS evolves. National privacy preserving requirements and GDPR will be followed.

Task 8.4.3 description: Use of implemented RD datasets for monitoring HCP and ERN activities and for data exchange with ERN registries.

Participants: INSERM +ARRHUS (+/- GREECE) & for year 3 RUMC & SCIENSANO +VENETO REGION (TBC) + OUS

Scope: This demonstrator will showcase the added value of ERN and will contribute to improving data collection, data quality and monitoring at the source. As such it is key in order to facilitate further integration of the ERN in the national health care system in all MS, as per timeline in fig 33.

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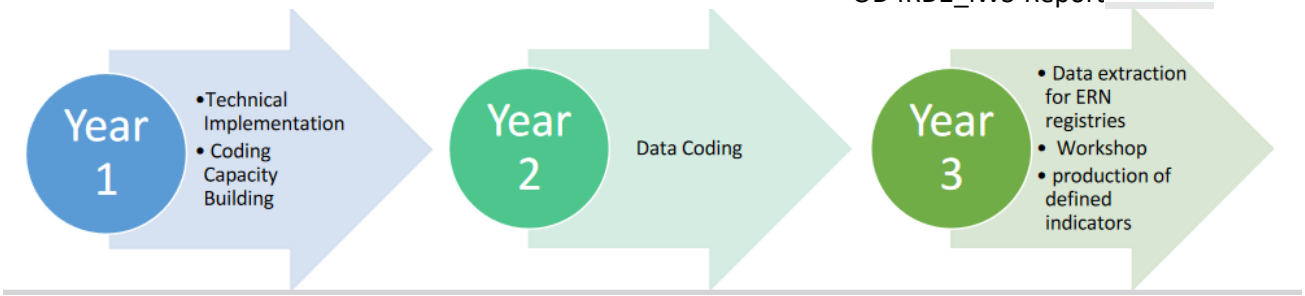

Figure 33 JARDIN Demonstrator 8.4.3 Timeline

Description: The demonstrator will work on implementing ORPHAcodes into national HER as well as CDE, and consequently focus on:

1. The re-use of prospective and retrospective data to assess the added-value of ORPHAcode implementation and the impact of ERN centres in hospital's activities by identifying a number of KPIs of interest. (ex: increase in the number of RD identified over the years during the JA and comparing with the same data extracted using ICD10; comparing Length of Stay (LOS) in RD inpatients vs non-RD patients or number/length of outpatient consultations).

2. The implementation of recommendations issued from X-eHealth for EHRs data extraction for registries (D8.5): by extracting data from the EHR into selected ERN registries, therefore eventually contributing to Demonstrator described in 8.4.1 if pertinent (need to select which ERN according to local maturity) enabled by the use of ORPHAcodes.

Communication & Dissemination

The available material was presented to the audience

Communication Available material	Dissemination available material
https://od4rd.eu/communication-material	https://github.com/OD4RD/Main-Help-Desk/wiki
https://od4rd.eu/orphacodes-bibliography	Tackling the invisibility of RD in European Member States: the OD4RD project contribution, ECRD 2024 Poster
https://od4rd.eu/03-deliverables	
https://www.youtube.com/channel/UCKMLSL9hlrxz6zKFod5IlnA	Tackling the invisibility of RD in European Member States: the OD4RD project contribution, ECRD 2024 Abstract to be published in special issue of OJRD
N.B. Canvas template for translation into your national language available for NH leaflet & on demand for other material	

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N.B. Your translated flyers/leaflet can be published on OD4RD website if you wish so!	
Insta: https://www.instagram.com/orphacodes/	

Table 10 Communication & Dissemination material available

Then progresses compared to the dissemination plan were discussed and a call of additional items was launched.

COMMUNICATION

- ❑ 4 Instagram Publications & 7 followers (oO): please follow with accounts of your institution or any other really ;)
- ❑ 2 Flyers & 2 booklets
- ❑ 2024 Achievements leaflet
- ❑ Add info on NH in the ERN collaboration trainings (new)…
- ❑ ….

…….Additional ideas ?

DISSEMINATION

- ❑ WIIKII with FAQ and ERN collaborations
- ❑ ECRD 2024 Poster & Abstract
- ❑ Comparison of Implementation in FR, DE, NL & NO….more if needed
- ❑ SNOMED CT strong argument: new
- ❑ WHITE PAPER
- ❑ WEBINAR for tekkies : new….
- ❑ Update Wiiki as per discussions
- ❑ OPRHAcodes at BoMS and Coo meetings: new

…….Additional ideas ?

During the discussion held during day one it had emerged that Official info whenever an ERN collaboration is complete should be made, in addition to the information provided in the wiki table, we could publish some post on social media and Orphanews.

Meeting Wrap up and revised OD4RD2 action plan

WP1:

- To be added in the communication plan: notify via social media and Orphanews whenever a revision collaboration is finalised
- Hospital Manager day: identify the relevant persons liaise with JARDIN (i.e. second level manager)
- ERN day co-event with BoMS
- EU RD plan as Overall EU should incentivise technical implementation of OC (as they do for SNOMED-CT) as it will be an epidemiological bias if only few countries send the data , it will not be representative

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WP2:

o to assess the impact of ERN collaborations an end-of-project survey must be launched: a CALL FOR IDEAS was open

o WP2-WP4 interactions: good opportunity to raise awareness on NH coding support activities (during the Collaboration kick off meeting, via the survey & in the final report)

WP3:

o WP3-WP2 interactions: Annotations and texts as part of WP2 ERN collaborations (AMAP): improve interaction mechanisms

o WP3-WP4 interactions: describe transcoding use cases

WP4:

o Compile a library of use cases fed by National Hubs (for re-use in trainings / exercises)

o National Hubs to liaise with National Health Data authorities for introduction of ORPHAcodes as medical terminology

o increase the discussion time in WP4 meetings as our Network of National Hubs has a real added value and having the time to discuss success stories, challenges and ways to overcome them is of paramount importance

o Build strong argumentation to support ORPHAcodes vis-à-vis National authorities from both an economic and clinical point of view with regards to SNOMED CT: Writing group to be launched

WP5:

o IT

▪ How can we highlight Nomenclature Pack in Orphadata and its annual release (color/ORPHAcodes, or stand-alone website?) => brainstorm on actions to decrease confusion on what to use and frequency of updates

▪ Include Mappor (terminologies mapping tool) in Orphadata (Nomenclature Pack version?)

▪ WP5-WP4 interactions: ORPHAcodes for Tekkies : Wiki modules

▪ WP5-WP4 interactions: ORPHAcodes for Tekkies: include a module in TfT or stand-alone webinar and increase the educational material for increased capacity building of the NH

▪ WP5-WP4 interactions: organise BYO UseCase sessions / trainings

▪ Classifications browser tool allowing for extraction (Nom. Pack version): launch a working group

▪ Develop use cases (together with WP4) for Dataviz-like tools including other data (i.e. genes, HPO)

▪ Introduce SNOMED CT to DataViz and Mappor (if agreed)

▪ DATAVIZ stats by country / other?

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o Evidence-based analysis:

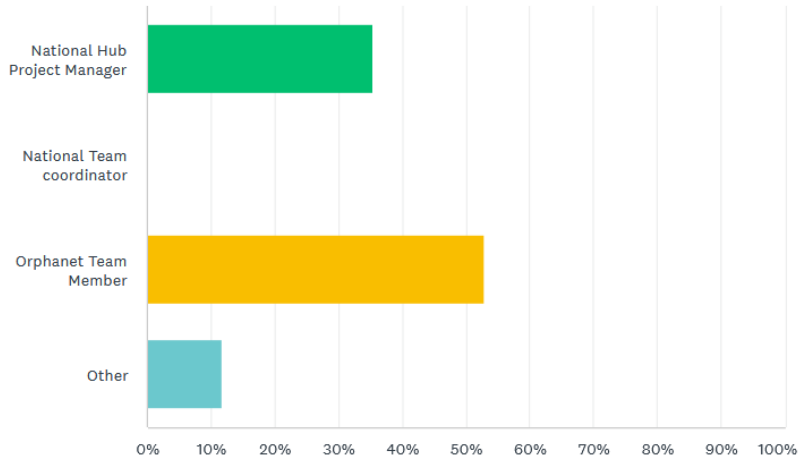
- Qualitative analysis + ERN coordinators
- WP5-WP2 interaction: message regarding Classifications / ORPHAcodes revision in ERN files

Annex 1 Feedback Survey Results

A feedback survey was shared with the participants during the meeting and then sent by email. It stayed open for 7 days. Questions aimed at collecting insight into the knowledge regarding the OD4RD2 project and its activities prior to the meeting as well as satisfaction regarding the future planned activities and on the meeting itself. 17 answers out of 46 participants were received. 100% of participants were either satisfied (strongly agree or agree). 100% considered that the workshop helped them better understand the activities carried out in the work packages they are not directly involved in. 100% of the respondents considered that the workshop inspired them for the next activities that will be carried out by their team.

Respondents:

17 answers out of 46 participants were received at feedback survey



ANSWER CHOICES	RESPONSES	
National Hub Project Manager	35.29%	6
National Team coordinator	0.00%	0
Orphanet Team Member	52.94%	9
Other	11.76%	2
TOTAL		17

Of these, all knew the project in general and only 4 did not know the specific activities carried out.

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	STRONGLY AGREE	AGREE	DISAGREE	STRONGLY DISAGREE	I DON'T KNOW	TOTAL	WEIGHTED AVERAGE
I was aware of the OD4RD/ OD4RD2 project before attending this workshop	100.00% 17	0.00% 0	0.00% 0	0.00% 0	0.00% 0	17	1.00
I was aware of the ALL Workcpackages specific activities of OD4RD/ OD4RD2 project before attending this workshop	41.18% 7	35.29% 6	23.53% 4	0.00% 0	0.00% 0	17	1.82
I learned about the OD4RD/ OD4RD2 project attending this workshop	35.29% 6	47.06% 8	5.88% 1	11.76% 2	0.00% 0	17	1.94
I learned about the Workpackages specific activities in which I am not involved during the workshop	47.06% 8	41.18% 7	5.88% 1	5.88% 1	0.00% 0	17	1.71

	STRONGLY AGREE	AGREE	DISAGREE	STRONGLY DISAGREE	I DON'T KNOW	TOTAL
I am overall satisfied with the Workshop	76.47% 13	23.53% 4	0.00% 0	0.00% 0	0.00% 0	17
The Workshop helped me better understand the activities carried out in the Workpackages I am not directly involved in	58.82% 10	41.18% 7	0.00% 0	0.00% 0	0.00% 0	17
The workshop has inspired me for the next activities that will be carried out by my team	58.82% 10	41.18% 7	0.00% 0	0.00% 0	0.00% 0	17
The workshop helped me better understand the other National Hubs Activities	52.94% 9	35.29% 6	11.76% 2	0.00% 0	0.00% 0	17
I think there was enough time for discussion during day 1	17.65% 3	47.06% 8	23.53% 4	0.00% 0	11.76% 2	17
I think there was enough time for discussion during day 2	29.41% 5	64.71% 11	5.88% 1	0.00% 0	0.00% 0	17
The pace of the workshop was appropriate during day 1	29.41% 5	58.82% 10	11.76% 2	0.00% 0	0.00% 0	17
The pace of the workshop was appropriate during day 2	31.25% 5	68.75% 11	0.00% 0	0.00% 0	0.00% 0	16

The respondents were also asked to list the things that went well during the meeting to be kept for further meetings, as well as the items that were less liked were traced and will be discussed for improvements.

What participants Liked:

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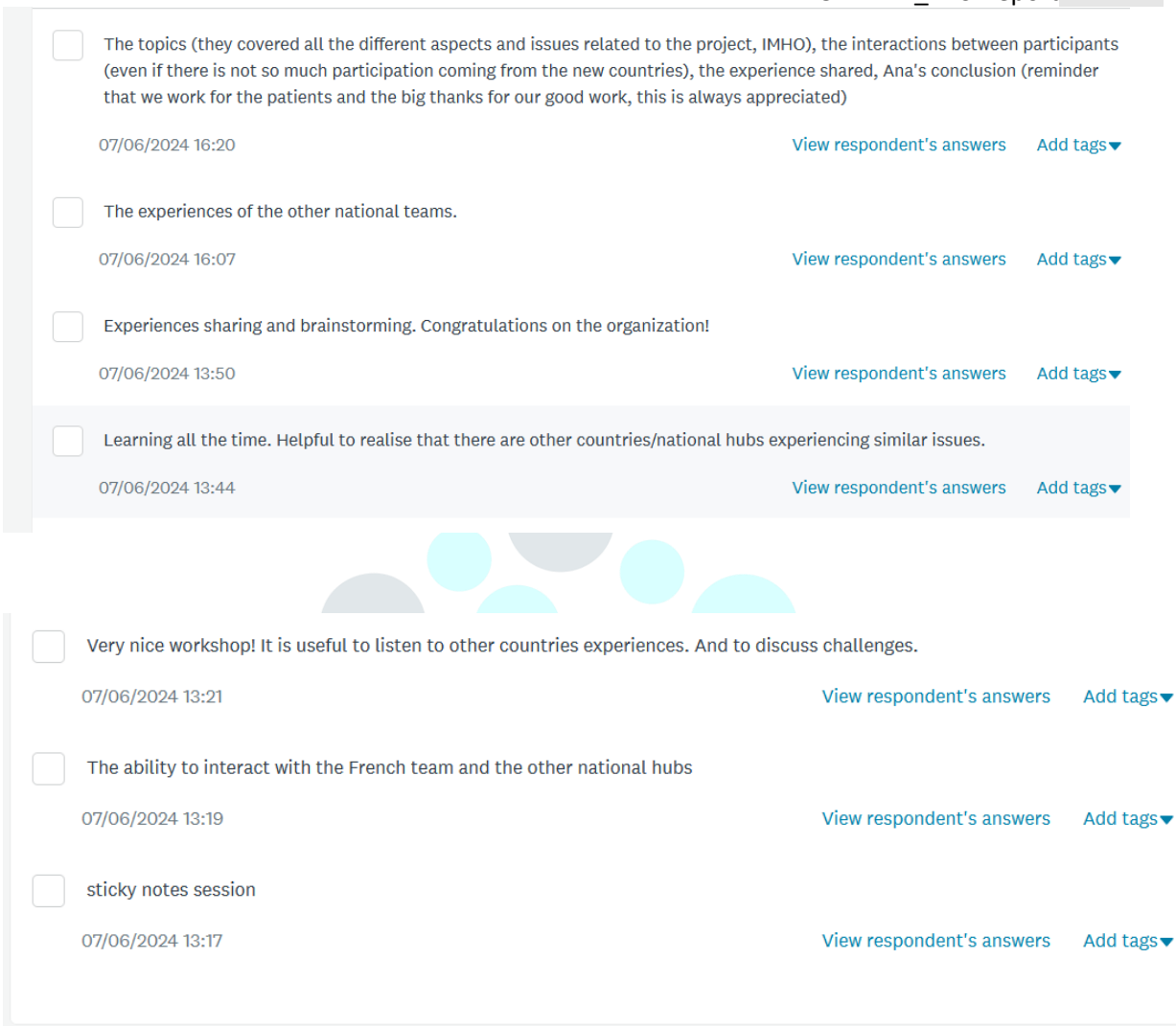
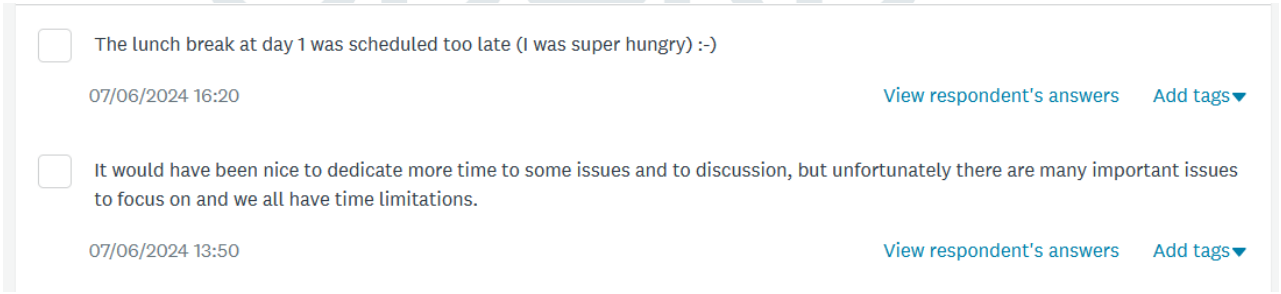


What participants disliked:



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Annex 2 OD4RD 2 iWS Agenda

6-7 June 2024

Agenda 6 June 2024

10h Opening and Welcome (AR)

10h45 Orphanet nomenclature and classification of RD update and maintenance (WP2) : What has been done (including update on indicators &Problems encountered) , What is planned & Discussion on new ideas (WP 2 speaker)

11h30 15 min Break

11h45 Develop Orphanet Knowledge and information base around RD in collaboration with ERNs (WP3): What has been done (including update on indicators &Problems encountered) , What is planned & Discussion on new ideas (WP3 speaker)

12h30 Support to the EC in its ERN strategy including dedicated IT systems (WP5): What has been done (including update on indicators & Problems encountered) , What is planned & Discussion on new ideas (TV, AR & MH)

13h15-14h30 Lunch Break

14h30 Consolidate and Expand national Orphanet nomenclature hubs (WP4): What has been done (including update on indicators &Problems encountered), What is planned & Discussion on new ideas (moderator RZ)

- ERN survey (SM)
- GitHub helpdesk (MCG)
- Indicators (KK)
- Lessons learned (Sweden)

15h30- 15h35 Short Coffee break

15h35 Workshop Network of National Hubs (WP4) activities (problems encountered and success stories) facilitated by Bfarm and Karolinska

- Presentation on successful experience
- Presentation on challenges
- Sticky-notes session
- Experiences from year 1
- Expectations on year 2

16h05 Coffee break 5 min

- Overview of sticky notes and discussion

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16h45 Closing: presentation of tomorrow's agenda (AR)

Agenda 7 June 2024

10h Opening and welcome (AR)

10h15 Recap of yesterday's discussions around sticky notes (KK) and mitigating actions and unplanned actions to include in the roadmap (AR)

10h45 Transversal actions planning I (SM-AR)

- Organisation of Workshops for external partners (ERN day and Hospital managers day….)

11h45 15 min break

12h Transversal actions planning II (SM/AR)

- Collaboration with JARDIN and other projects (AR)

- Communication: (Posters for 2024 and 2025, Publications for 2024 and 2025, Social media strategy…) (SM)

13h15 Closing

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