

Deliverable 1.3

OD4RD2 Evaluation Report

June 2024



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Background

This report represents the Deliverable 1.3 of the OD4RD2 project (www.OD4RD.eu). It contains a quantitative evaluation with (1) the analysis of the project KPIs and (2) the analysis of all relevant, task-related indicators as outlined in section (“Specific objectives of the action”), methods and means, and a qualitative evaluation presenting the results of the planned intended satisfaction surveys which include questions to assess users’ further needs and suggestion of improvements.

Quantitative Evaluation

Project Key Performance Indicators

All project milestones (a) and deliverables (b) planned by the end of June 2024 have been achieved on schedule, for milestones a +/- 1 month flexibility was applied. Project objectives (c) were fulfilled.

a) Milestones :

Milestone Number	Milestone Name	Description	Lead Beneficiary	Achievd on time (+/- 1 month)	Date
MIs1	KOM Preparation	Participants list and location available	INSERM	Y	01/04/2023
MIs10	End-users’ state-of-play survey		BFARM	Y	04/05/2023
MIs11	Establishment and delivery of - annual action plan for each national hub	At least 75% of national action plans, based on the state of play in the country, have been written and are provided as pdf document to the executive comittee (ExCom).	BFARM	Y	30/06/2023
MIs18	Specifications for data exchange workflow with the EC ERN information system	OD4RD functional specifications document revised/updated	INSERM	Y	31/07/2023

Milestone Number	Milestone Name	Description	Lead Beneficiary	Achieved on time (+/- 1 month)	Date
MIs6	ERN collaborations planning (nomenclature) WP2 1-INSERM	Timelines communicated to involved ERNs.	INSERM	Y	12/09/2023
MIs19	Feasibility study and specifications for data analysis towards strategic report(s)	Specifications document validated by EC and Orphanet teams	MUW	Y	11/01/2024
MIs3	Target specific survey launched	Link sent to all ERNs and to HCPs Hospital managers in participating countries.	INSERM	Y	31/01/2024
MIs8	First Annual checkpoint	Indicators checkup	INSERM	Y	29/03/2024
MIs12	HCP survey WP4 5-	At least 75% of national hubs have performed a survey and have presented written results. These are provided as pdf document to the executive	BFARM	Y	07/05/2024
MIs4	Mid-project workshop Preparation	Participants list and location available	INSERM	Y	14/05/2024
MIs13	Mid-term Report on lessons learned and on improvement of capacity and implementation	Report on lessons learned prepared	BFARM	Y	24/06/2024
MIs14	Establishment and delivery of - annual action plan for each national hub Y2	At least 75% of national action plans, based on the state of play in the country, have been written and are provided as pdf document to the executive committee (ExCom)	BFARM	Y	14/06/2024

Table 1. OD4RD2 Project milestone status

b) Deliverables :

/N.B\ All the project deliverables listed below are Public and available at: <https://od4rd.eu/03-deliverables>

Deliverable #	Deliverable Name	Description	Lead Beneficiary	ontime Delivery
D1.1	KOM Executive summary	This executive summary includes all items discussed during the meeting/workshop, as well as the agreed work plan. It has been delivered as a pdf in English.	INSERM	Y 30/06/2023
D1.2	Dissemination plan	A Dissemination plan detailing all the material is made available to disseminate project results, the planned events for the project dissemination. It has been delivered as a pdf in English.	INSERM	Y 30/06/2023
D4.1	State of play survey	A report compiled from the survey on the state of play in each participating country regarding ORPHAcodes implementation in HCPs has been delivered as pdf document on the project's website.	BFARM	Y 31/08/2023
D1.3	Mid-term Evaluation Report	This report contains (1) the analysis of all relevant, task-related indicators as outlined in section (“Specific objectives of the action”), methods and means, as well as (2) the analysis of the KPI and of the planned intended satisfaction surveys. It has been delivered as a pdf in English.	INSERM	Y 30/06/2024
D2.2	Mid-term report on Orphanet-ERN collaborations	Written summary report of completed and ongoing collaborations with ERNs for the concerned period of reporting. It provides an overview of the number of projects and the groups of diseases they covered, the ERNs involved (and more precisely the thematic groups within the ERNs when pertinent), the issues raised and lessons learned (especially those that may benefit other ERN projects). It also includes annexes containing the lists of entities revised (by activity).	INSERM	Y 30/06/2024

Deliverable #	Deliverable Name	Description	Lead Beneficiary	ontime Delivery
D3.1	Mid-Term Report on Genetic Annotations (new and updated gene-disease relationships), ORPHAcodes alignments with other terminologies, and newly produced or updated texts (definitions and abstracts)	This report contains 3 sections- New discoveries around genes related to RD and their representation in the Orphanet database, with links to where these relationships are available for reuse. - RD coverage in target terminologies and the relationships between ORPHAcodes to each of the target terminologies, with links to where these alignments are available for reuse - list of newly produced definitions and of newly produced or updated RD summary information realised in collaboration with ERNs	INSERM	Y 30/06/2024
D4.2	Annual Action plans Year 1	A report compiling all national hubs annual action plans delivered as a pdf document. This also includes a description of the structure of each national hub.	BFARM	Y 30/06/2024
D4.3	Mid-term Report on lessons learned	A report on lessons learned has been delivered as pdf document, including a compilation from the FAQ collected at a mid-term meeting"	BFARM/Karolinska	Y 30/06/2024
D1.4	Mid-project workshop report	This executive summary includes all items discussed during the meeting/workshop, as well as the agreed work plan. It has been delivered as a pdf in English.	INSERM	Y 31/07/2024

Table 2. OD4RD2 project deliverable status and availability

c) Project objectives:

This project is fulfilling 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,
2. To contribute to the harmonisation of data collection amongst settings (health records, registries) and amongst countries, by the dissemination of coding good practices at the data source level.
3. To contribute at supporting evidence-based decision-making in the frame of the European strategy around ERNs, by supporting the exploitation of reference corpus of data and information on RD.

Six specific objectives were also defined together with Process, output and outcome indicators have been defined for the specific objectives of the project as well as their targets as indicated in the table below and fully described in the next chapter.

Colonne1	Number planned & pertinent to M18 (A)	Number of indicators having reached the planned target (B)	Of which Target Exceeded (D)	% Indicators achievement
Process indicators	13	13	3	100%
Output indicators	15	12	4	81%
Outcome indicators	6	5		92%

Table 3. OD4RD2 project Percentage of Achievement of Process, output and outcome Indicators' Targets

N.B. Target deviations are explained in the section below

Reporting of impact/output/outcome indicators per specific objectives

The proposed specific objectives of the Orphanet Direct Grant are as below.

1. To continue developing, updating and maintaining continuously the Orphanet nomenclature (ORPHAcodes) and classification of RD, in collaboration with ERNs, in line with the evolution of knowledge.
2. To expand the semantic interoperability with the main international terminologies in use, and in particular with SNOMED CT and with WHO's ICD11, alongside with other medical and genetic resources (i.e. OMIM, UMLS)
3. To continue developing and continuously maintaining the data and information associated with RD, and in particular genetic data and textual information associated with RD
4. To continue ensuring support for the local implementation of ORPHAcodes in national HCPs hosting ERNs by establishing and enlarging a Network of Orphanet national nomenclature hubs across Europe.
5. To put in place and to maintain the necessary mechanisms to deliver and update the Orphanet nomenclature and knowledge base (as reference body of data) in the IT system related to ERNs (i.e. in HCPs, at the EC, etc).

6. To analyse and produce reports on RD coverage, identifying complementarities, as well as 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, by taking the lessons-learned from the pilot OD4RD project.

Process, output and outcome indicators have been defined in the Grant Agreement per each specific objective. Target reached for each indicator are indicated in the table below. Deviations are explained in the next paragraph.

Specific Objective Number	1.	
Specific Objective	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.	
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Collaboration workflow's steps completed for each ERN collaboration and justified in the final report	100%	100%
Number of internal nomenclature validation cycles completed (each cycle includes an internal pre-validation meeting, an internal validation meeting, and the production of a final decision report disseminated within the Orphanet Network).	8/8	8
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Cumulative weight of ERN collaboration projects completed. Each project is attributed a "weight" according to its scale and complexity (from smallest size/lowest complexity to larger size/highest complexity) among these values: 0,5 ; 1 ; 1,5 ; 2.	25.5 cumulative weight	2
Number of clinical entities (ORPHAcodes) created, modified or inactivated.	800/year	411 in 2023
Number (%) of completed ERN revision projects for which a final collaboration report has been produced and sent to the ERN representatives.	100%	100%
Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
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. Measured by a short satisfaction survey sent after the project	90%	This process will be formalized in Y2. N.A.
Specific Objective Number	2	
Specific Objective	To expand the semantic interoperability with the main international terminologies in use, and in particular with SNOMED CT and with WHO's ICD11, alongside with other medical and genetic resources (i.e. OMIM, UMLS)	
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Validation meetings with the scientific officer for ICD-10, ICD-11 and SNOMED-CT mappings	30/year	>30
Survey of OMIM (phenotypes) updates	10 /year	10
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
% of ORPHAcodes (at the disorder level) aligned with ICD-10, including inexact mappings	100%	99,7%
% of ORPHAcodes aligned with ICD-11 terms, including inexact mappings	100% by Year 3	66,6%

Number of ORPHA-SNOMED CT human-readable mapping files released	3 (1/Year)	1
Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Satisfaction and utility of the nomenclature mappings according to end users (assessed through online surveys and ERN proactive surveys)	Increase compared to 2021 figures	7 % increase. <i>In 2023 81% of online users who knew the mapping files were either very satisfied or satisfied of this product/service. Compared to 74% in 2021</i>
Number of downloads of the nomenclature with alignments and of the human-readable ORPHA-SNOMED CT files in Orphadata		40% increase. 28 654 in 2023 vs 16 458 in 2021 baseline (orphadata nom et alignements+ Nom Pack). Nom Pack downloads detail: 689

Specific Objective Number	3	
Specific Objective	To develop and continuously maintain the data associated with RD, and in particular genetic data. In addition, to create and update the textual information associated with RD	
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Survey of OMIM (genes) updates	10 /year	4
Collaboration progress as assessed by completion of agreed texts and calculated as a percentage of agreed diseases (including both definition quality controls and production/updates of disease information summaries) completed per cycle. Calculated annually based on the cycles completed during the year.	95%	127% <i>The target of production is 350 and during Y1 the team produced 445 texts as the process is working very well (more collaborations with ERNs, more reactivity form the epxerts..)</i>
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
New or updated disease-gene associations per year	450	517
Number of disease summaries/definitions produced or updated per year	250	358
Number of definitions reviewed for quality controlled per year	100	72
Outcome/Impact Indicator(s)	Target	
Satisfaction and utility of the relevant category (gene-disease database and encyclopaedia) according to end users (assessed through online surveys and ERN proactive surveys)	Increase compared to 2021 figures	A small decrease is observed: -4%.

<p>Number of downloads of the Gene-disease associations file in Orphadata</p> <p>Additional indicator: Views of the https://id-genes.orphanet.app/ithaca/ page which allows Search for intellectual disability-related genes and has been developed in collaboration with ITHACA ERN (this is a baseline)</p>		<p>In 2023 91% of online users who knew the encyclopaedia were either very satisfied or satisfied of this product/service. Compared to 95% in 2021</p> <p>A small increase can be observed for the gene database: +3%.</p> <p><i>in 2023 87% of online users who knew the gene disease database were either very satisfied or satisfied of this product/service. Compared to 84% in 2021.</i></p> <p>40% decrease for genes, Almost 150% increase for texts (in 2023 6633 genes downloads, while in 2021 approx. 11096 downloads. In 2023: 663 Texts downloads vs 274 in 2021</p> <p>7,667 (over a 12 months period)</p>
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Specific Objective Number	4	
Specific Objective	To ensure support for the local implementation of ORPHAcodes in national HCPs hosting ERNs by establishing a Orphanet national nomenclature hubs network.	
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Survey of State of Play in new involved countries at start of project (to show where in the spectrum they do align to the others)	1 report in first half of project	Available
Meetings of the national hubs for alignment on implementation questions and to build capacity within the network of hubs	6	9 WP4 meetings plus 9 Open sessions
Survey each year about improved capacity in national hubs	1 /year	Available in the paragraph below
Attendance of each hub to Train-for-Trainers (TfT) sessions	1/hub	<p>In total, these trainings gathered 58 participants (26 at the Basic from 14 National Hubs and 32 at the</p> <p>Advanced' session from 18 National Hubs,) At least 1 person from each</p>

		hub attended either one of the sessions.
Implementation of a clear and traceable workflow for helpdesks activities (from reception to closure of demands to the national hubs) using a ticketing tool	1/hub	19/19 , repr. 100% of active hubs
National action plan described and published in the project intranet, with annual revisions	1/hub	19/19, repr. 100% of active hubs
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Availability of the local GitHub or similar ticketing system for national hub helpdesks demands	1/hub	19/19 , repr. 100% of active hubs
Autonomous treatment of first-level end-users' demands (based on basic-level training received) at end of year 1 of hubing	100%/hub from year 1	100%
Autonomous treatment of second-level demands (based on advanced-level training received) at the end of year 2 of hubing	100%/hub from year 2	N.A.
Organisation of Training-workshop or other event by each hub	1/hub	61 trainings & 151 ad Hoc meetings or awareness events in 18 countries (95% of active hubs)
Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Compiled report on implementation questions to serve as a lessons learned base for other countries joining the network of hubs in the future	1 (M33)	N.A.
Feedback by national training/events participants on knowledge gain after the training/event (short survey)	85% have gained knowledge	No formalised surveys were carried out
Feedback on satisfaction by users on the tool used to received demands by the national hub (short survey)	N.A. Will be carried out at the end of the project Goal 90% users are satisfied	N.A
Feedback on quality of interaction by national users with national hubs (short survey)	N.A. Will be carried out at the end of the project Goal 90% users are satisfied	N.A.
Feedback by TfT Participants on knowledge gain after the training event (short survey)	85% have gained knowledge	93% of them were satisfied or very satisfied with the program delivered. https://network.orphanet.org/wp-content/uploads/E_Com/Int_Com/ONW_News/ONW_2023/ONW_2023_3_VF.pdf

Specific Objective Number	5	
Specific Objective	To put in place and to maintain the necessary mechanisms to deliver and update the Orphanet nomenclature and knowledge base (as reference body of data) in the IT systems related to ERNs (i.e. in HCPs, at the EC, etc).	
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Meetings with national hubs on regular basis in order to assess the evolving needs in terms of nomenclature / datasets delivery	N.A.	N.A.
Output Indicator(s)	Target	
Technical advice provided as requested per national hubs including proposal of dedicated services to hospitals	100% of requested advices	100%
Outcome/Impact Indicator(s)	Target	
Statistics on access of the dedicated API, measurement of the usage by ECT IT /HCPs systems through API gateway and systems metrics (Servers usage).	API's call management indicator through the API Gateway and servers resources consumption>0	>0
Specific Objective Number	6	
Specific Objective	To produce analysis and reports on RD coverage, overlaps, insufficiencies in coverage and gaps within each ERNs in order to support the ERN coordination, Board of Member States (BoMS) and European Commission in their work	
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Meetings with EC, ERN chairs and BoMS to assess the needs organized	1-2/year	B3 unit Meeting Luxembourg Jan 2024; ERN Chairs & BoMS Meeting 22 May 2024
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
Short reports or communications about the agreed tools and strategy for the coverage and gap analysis at the relevant meetings of the BoMS and the ERN coordinators group	Minimum 1 for each group (BoMS, ERN-CG)	ERN Chairs & BoMS Meeting 22 May 2024
Report analysis on overlaps, insufficiencies and gaps on RD coverage / ERN (to be replaced by another report analysis if more priority)	1	Preliminary report sent for validation to ERNs as of July 2024
Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)
End-users' satisfaction on the report	N.A. will be collected after report delivery goal: >95%	N.A.

Table 4. OD4RD2 project Process, output and outcome Indicators' Targets achievement at M13

Indicators deviation explanation & mitigating actions planned:

- 1) Specific objective 1. 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 number of clinical entities (ORPHAcodes) created, modified and inactivated has been lower than expected (411 in 2023 vs 800/year). It is important to note that these figures do not include all the requests that are launched but ultimately need no action on the database but still need to be assessed and document and evaluated by the Nomenclature project managers.
 - The Cumulative weight of ERN collaboration projects completed in Y1 is also a bit low, however the current prevision of the final cumulative weight of the already planned collaborations is of 23, indicating that we are aligned with the final target of 25,5.

- 2) Specific Objective 3: To develop and continuously maintain the data associated with RD, and in particular genetic data. In addition, to create and update the textual information associated with RD
 - Survey of OMIM genes: 4 out of 10 were carried out as there has been a few months staff gap on this activity and a new script was tested and set up when the new staff was recruited. We expect to reach the expected target in Y2
 - Number of definitions reviewed for quality control per year was a little less than planned but the target for Abstracts was outreached.
 - Satisfaction and utility of the relevant category:
 - o gene-disease database we observe a decrease compared to 2021 regarding the satisfaction of website users while we observe an increase of the Orphadata downloads
 - o encyclopaedia we observe an increase compared to 2021 regarding the satisfaction of the website users while we observe a small decrease of the orphadata downloads.
 - These figures testimony of different utilisations of the same data and deserves monitoring over a longer period to confirm a trend.

An additional Indicator will be followed up: the number of views of the <https://id-genes.orphanet.app/ithaca/> page which allows to search for intellectual disability-related genes and has been developed in collaboration with ITHACA ERN, the number of views here reported over a 12 Month period is a baseline for next report;

- 3) Specific Objective 4: To ensure support for the local implementation of ORPHAcodes in national HCPs hosting ERNs by establishing a Orphanet national nomenclature hubs network.
 - Romania was still not able to carry out activities as of June 2024, discussions are ongoing to assess their capacity to start their activities as of September 2024 for the last year and a half of the project
 - 2 National Hubs did not organise trainings in Y1 (IT and CZ): organisation is planned in year 2, No formalised gain of knowledge survey carried out in National Trainings: a possibility would be to include a gain of knowledge auto evaluation question in the final satisfaction survey.
 - No gain of knowledge measure during the Training for Trainers (TfT): this will be formally organised for the TfT in Y2
 - Indicators glossary will be prepared in Year 2 so to have a clear common understanding of the Indicators to report, as some confusion was observed between the National Hubs reporting.

Raised capacity of National Hubs

Over the course of the OD4RD project a threshold was defined to assess “Raised capacity of National Hubs”: at least one third of the national participating hubs should have conducted trainings and/or set up an active helpdesk.

During OD4RD: 19 out of the 19 active National Hubs have set up a helpdesk and 18 out of the 19 National hubs have held at least 1 training session (overall 61 training sessions were held during OD4RD2 and 151 ad Hoc meetings and 2 awareness events), this represents more than 2/3 of the hubs having showed raised capacity.

National Hubs specific Indicators

Moreover, during the OD4RD project indicators have been defined to follow the National Hubs activities more granularly and will be analysed at the end of the first OD4RD2 year in order to define thresholds so that in the following years each team will set up a self-evaluation process of national hub activities by assessing raised capacity and this will allow to make evidence-based decisions to adjust their activities and priorities and to improve the quality of their services.

The table of additional indicators to follow up or definition of a baseline t is indicated below:

Indicator	Target	Achieved Y1 OD4RD2
National action plan delivered/updated	1/Year	18 countries (+Switzerland)
Number of training sessions in local language	min 1/year, number should increase over the years	17 countries, see indicators table in the annex for baseline for Y2
Number of HCPs involved in training sessions:	will depend on number of HCPs present in the countries, should be >70% by the end of OD4RD2	Number of medical departments involved: 184
Number of ERNs present at the training sessions	will depend on number of ERNs present in the countries, should be >70% of ERNs present in the country by the end of OD4RD2	Of which 131 ERN units
Results of feedback surveys for training events and also of knowledge gain if available	Should be at min 70% of satisfied users and 70% of knowledge gain	To set up for Y2
Number of meetings and workshops with hospital managers, HCP medical informatics teams, software providers, and coders (also indicate with which of these categories there have been contacts)	to be assessed according to first year results and the threshold should increase yearly	151 Ministry of Health meetings: 2 Hospital Managers Meetings: 22 Medical Informatic teams meetings: 43 Coders meetings: 35 Software providers meetings: 35 Other 14
National helpdesk activity	to be assessed according to first year results and the threshold should increase yearly	N.A. Will be calculated starting next year, Y1 is a threshold
- how many tickets/demands were received (by category)/!\ these figures	-	299 • Coding:135 • Nomenclature: 97

Indicator	Target	Achieved Y1 OD4RD2
include all demands not only those traced via the Github platform		<ul style="list-style-type: none"> • Others: 65
- Analytics of demands according to type of users and typology of demands: /!\ these figures include all demands not only those traced via the Github platform	-	<ul style="list-style-type: none"> • User, clinician: 126 • User, Hospital management: 3 • User, coders: 98 • User, IT personnel: 20 • User, other: 48
- Number of demands answered by national helpdesk /!\ these figures include all demands not only those traced via the Github platform	- should increase over the years (Y1 as a baseline)	- 193 (baseline)
- Number of demands required support of coordination team /!\ these figures include all demands not only those traced via the Github platform	- should decrease over the years for basic and intermediate level demands - (Y1 as a baseline)	- 106 (baseline)

Table. Compiled specific National Hubs indicators OD4RD2 Year 1

Indicator	Average/total	Countries	AT	BE	BG	CH	CZ	DE	ES	EE	FI	IE	IT	LT	LV	NL	NO	PL	PT	SE	SI
Ste of play Survey	19	19	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
ERN survey	17	16	Y	Y	Y	N/A		Y	Y	Y	Y	Y	Y	Y	yes	Y	Y	Y	Y	Y	N
1st action Plan	19	19	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
Number of training sessions/workshops/webinars in local language	61	18	1	7	11	2		1	2	1	3	7	0	6	0	0	9	6	2	1	2
Awareness events	2	2											1		1						
Number of participants	1320	14	30	45	164	5		92	46	18	110	147		200			140	169	100	35	19
Number of medical departments involved	184	14	3	13	10	2			6	16	5	1		23			6	30	50	15	4
registries	1	1							1												
Number of ERN units involved	131	11	11	8	5				2	16	5	1		18			6	8	48		3
Results of feedback surveys	0				Y				82%								Y				
MoH meetings	2	1										2									
Hospital managers meetings	22	6	5	0	1							1		6			3	6			
Medical informatic teams meetings	43	8	2	5	1						1	2		7		12	10				3
Coders meetings	35	9	3	4		12					1	1				2	2	6		4	
Software providers meetings	35	8	2								3	1		7		1	17	2			2
Other				14																	
Github created	15		yes	YES	yes	yes		yes	yes	no	yes	yes	yes	yes	yes	yes	yes	YES	NO	YES	NO
Number of tickets received (or demands, these figures include all demands not only those received via github)	299	10	15	16	9	48		41	25	2	5	4	0	uncertain number (will be traced in the future)	32	21	65	16		0	
ticket type: coding	135		2	7	9	12		37	7		3	1			23	1	24	9			
ticket type:nomenclature	97			6	0	25		4	18		1	1			6	20	10	6			
ticket type:others	65		13	3		11					1	2			3		31	1			
Ticket submittor: clinicians	126			7		3		14	3		5	3			16	11	55	9			
Ticket submittor:	3							1										2			

Indicator	Average/total	Countries	AT	BE	BG	CH	CZ	DE	ES	EE	FI	IE	IT	LT	LV	NL	NO	PL	PT	SE	SI
Hospital management																					
Ticket submittor: Coders	98		15	4	8	45		3	14							5	4				
Ticket submittor: IT personnel	20				1			4								7		6	2		
Ticket submittor: Others	48			5				19	7			1				4	6	4	2		
Number of demands/tickets answered by NH in autonomy	193		13	16		16		40	10	1	0	2				32	7	45	11		
Demands/tickets forwarded to coordination			2	12	9	32		1	18	1	5	2					14	20	5		
Demands/tickets added to GitHub			2	12	0	32		1		1	3	0					1	20			
Demands/tickets closed at national level				16		0															

Table. Specific National Hubs indicator by country OD4RD2 Year1

N.B. The Inserm located in France is the Coordinating team of the project and of the Orphanet Network. France has not developed an Orphanet Nomenclature National Hub in the frame of the OD4RD project as supporting activities for the implementation of ORPHAcodes have started since the 2 National Plan on RD and therefore organised national support had been already in place before the beginning of OD4RD in 2022. In particular annual trainings are organised with the RD National Reference Networks (Filières de Santé Maladies Rares) to ensure capacity building of coders and helpdesk activities exists since 2019 with the creation of a position working half time at the National RD registry and half time at Orphanet, in charge of following up users needs in terms of codes, trainings, tools etc.

Qualitative evaluation

General Users evaluation

- I. Project activities and results have been made available through the online www.orpha.net website : nomenclature, genetic information and textual information per disease.

In January 2023 a satisfaction survey of the Orphanet website users was conducted in one phase, launched via a pop-up window appearing on the first page users landed on. The survey was translated in 8 out of 9 languages of the website (i.e. English, French, Spanish, Italian, Portuguese, Dutch, German and Polish) and was displayed in the language of consultation via the pop-up. The survey was closed after 6 weeks of display on the website. This survey was composed of 13 questions split into 3 sections: Knowledge of and reasons for using the Orphanet website; Your opinion on Orphanet’s products / services and its website ; More information about you.

The on-line survey was designed using the online survey tool Survey Monkey, the questions focused on the professional activity of the users, their habits when they visit the Orphanet website, their opinion of the content as well as their overall satisfaction and their suggestions for improvement. A total of 9,852 users gave answers to the questions this year. This is slightly more than last year (9,389 respondents).

The results from all of the languages of the survey were consolidated and then analysed. The results of this analysis are presented in a report available online (https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Orphanet_survey2023.pdf) with elements of comparison as regards last survey’s results.

In this section we highlight the results which are pertinent to the project related activities, i.e. nomenclature, classifications, alignments, genetic info and texts. For this we exploited the results from Question 6 How useful would you rank the following Orphanet services for your own use? . This question aims to determine the utility and users’ knowledge of the various services/products offered by Orphanet. Respondents were asked to rate (++, +, -, --) each service/resource or to specify that they did not know this service/product. Answer was not required for all services / products and 3,458 respondents replied to this question, i.e. most regular users (82%) representing 37% of the total respondents. For the purpose of this analysis we subtracted the users that did not know the service. Overall 87% of the users that knew the service are either very satisfied or satisfied with the products listed in the table below, this is stable compared to our previous analysis carried out thanks to the 2022 Survey. Percentage is also indicated by product in the table below.

Product	% users that know the product and are either very satisfied or satisfied with it. Survey 2022	% users that know the product and are either very satisfied or satisfied with it. Survey 2023
Disease summaries	96%	91%
Classifications	88%	88%
Inventory of genes	88%	87%
Orphanet nomenclature	83%	87%
Cross referencing	80%	81%
Average	87%	87%

Table 5. % of users that know the product and are either very satisfied or satisfied with it, comparison 2022 vs 2023

II. Nomenclature, classification, linearization genetic information and textual information per disease is also available from the data download platform Orphadata (www.orphadata.com)

Nomenclature downloads were unusually high in 2022, whereas we observe higher results between 2023 and 2021.

Data	Downloads 2023	Downloads 2022	Downloads 2021
Classifications	86,363	105,517	106,827
Genes	6,633	11,902	11,096
Linearisation	1,026	1,432	1,473
Nomenclature	28,654	255, 872	16,458
Texts	633	279	274
Total	123,309	375,002	136,128

Table 6. Orphadata downloads by type of data and by year (2023 vs 2022 vs 2021)

User satisfaction surveys sent to ERNs,

To assess ERNs’ feedback as end-users a survey to evaluate the current situation of ORPHAcodes usage by health care providers (HCPs) linked to ERNs in countries participating in OD4RD 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

The Minimum Data Set of surveyed was set as the email contacts from each of the National ERN centres listed the Orphanet Data Base of expert resources by country (https://www.orpha.net/en/expert_centres/european-reference-network), and each Hub could add additional contacts if needed

A Minimum data set of questions was also established and each Hub could add questions if needed at national level.

16 Hubs provided a report (Austria, Belgium, Bulgaria, Germany, Spain, Ireland, Italy, Finland, Estonia, Lithuania, Latvia, the Netherlands, Norway, Poland, Portugal and Sweden) which describes in detail the methodology used. The preliminary results are summarised below and available here togetherwith an analysis by ERN.

In total, more than 1447 ERN units were contacted out of the 1523 units registered in the Orphanet database (95%) However, these number varied across countries from 8 to 629 [but this is an underestimation as only 15 hubs out of 16 have provided this info] and 544 answers were received (37.6%, 16%-88% depending on the country) It must be noted that 1 country has not provided information of the numbers of units contacted and that the Netherlands decided to contact only the units present at RUMC).

78% of the respondents confirmed providing RD patient data to the ERN coordination (n= 424/544)

53% confirmed using ORPHAcodes or ORPHAcodes coupled with another terminology for this purpose (n=262/494).

When asked what professionals are in charge of codification, most of the units answered medical doctors (n=393).

When specifically asked whether they used ORPHAcodes to send data to coordination, 49% said yes (n=231/471), for annual monitoring (n=190), 5-year evaluation (n=106), registries (n= 98).

50% confirmed codifying directly with ORPHAcodes (n=183/363)

An open question aimed at understating the main reason for not using ORPHAcodes was also present in the survey. Answers for this question were variable according to the users but they have been categorized by theme and the results show that the main reasons for not using ORPHAcodes are (in decreasing importance order): Unavailability of ORPHAcodes in Health Electronic Records, ORPHAcodes use not being mandatory, Lack of time/resources, ORPHAcodes not suitable, Lack of knowledge.

A detailed report of the OD4RD2 ERN survey is available here: <https://od4rd.eu/03-deliverables>

Organisation of trainings and workshops to disseminate coding best practices by the National Hubs, at national level and in local language

61 trainings had been held by the national hubs and 151 ad hoc meeting and 2 awareness events, with more than 1,320 participants representing more than 131 ERN expert centers and overall, the national hubs were able to report that the feedback from the participants after the training sessions has been very positive.

Satisfaction surveys of Face-to-face and online events organised to exchange with ERN coordinators.

Kick off Meeting satisfaction survey

As of M 15 the Coordinators of ERNs were invited to the OD4RD2 kick off meeting. 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 OD4RD and OD4RD2 project and its activities prior to the meeting as well as satisfaction regarding the future planned activities and on the meeting itself. 29 answers out of 70 participants were received. 93% of participants were either satisfied (strongly agree or agree) and all the respondents knew the project in general before attending and only 4 did not know the specific activities carried out. 93% considered that the meeting was helpful in further better understanding the project objectives and plans. 29 answers out of 70 participants were received at feedback survey of which only 6 ERN representative and 2 Hospital managers, 1 Unknown and the others being Orphanet team representatives.

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

	STRONGLY AGREE	AGREE	DISAGREE	STRONGLY DISAGREE	I DON'T KNOW	TOTAL	WEIGHTED AVERAGE
▼ I was aware of the OD4RD/OD4RD2 project before attending this meeting	72.41% 21	27.59% 8	0.00% 0	0.00% 0	0.00% 0	29	1.28
▼ I was aware of the specific activities of OD4RD/OD4RD2 project before attending this meeting	48.28% 14	37.93% 11	13.79% 4	0.00% 0	0.00% 0	29	1.66

Moreover 27/29 were satisfied with the meeting.

	STRONGLY AGREE	AGREE	DISAGREE	STRONGLY DISAGREE	I DON'T KNOW	TOTAL
▼ I am overall satisfied with today's meeting	55.17% 16	37.93% 11	6.90% 2	0.00% 0	0.00% 0	29
▼ We have busy schedules, meetings should be shorter to facilitate attendance	14.81% 4	29.63% 8	37.04% 10	7.41% 2	11.11% 3	27
▼ The meeting helped me better understand the activities carried out in OD4RD2	50.00% 14	42.86% 12	0.00% 0	3.57% 1	3.57% 1	28
▼ I think there was enough time for discussion during the Morning interactive session	41.38% 12	37.93% 11	10.34% 3	6.90% 2	3.45% 1	29
▼ I think there was enough time for discussion during the afternoon interactive session	46.43% 13	32.14% 9	10.71% 3	7.14% 2	3.57% 1	28

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.

Globally, it was noted that communication with ERNs should be improved.

- ⇒ Several actions have been organised in order to improve this aspect (i.e. attendance of OD4RD officer to ERN events to present the project, project presentation at the BoMS and ERN-Coo meeting as well as organisation of an ERN Day to raise awareness about the project and ORPHAcoding.

During a brainstorming session (connecting ideas), the Kick off meeting audience was also asked a series of questions which allow to identify the barriers encountered and the opportunity to suggest solutions which have been integrated in the OD4RD2 action plan revision.

Some questions were specifically oriented to ERNs collaborators to have their feedback related to passed and ongoing projects.

The main conclusions and feedback of this brainstorming session were:

- The majority of the participants (78%) knew the ORPHAcodes related to their field, some commented about the ongoing revisions, and others about the needs of ORPHAcodes mentioning that the clinicians know it most based on their practice and patient registries (at the minima, know where and how to find specific RD codes)
- The majority of the participants (64%) found that the Orphanet Classification is complete but could be more accurate or updated. Some examples were provided: new described syndromes should be included in a quicker delay in the Nomenclature; a few disorders are missing and it causes specific problem as for RD there is often no 'other/unspecified' provided code.
- Through any context (previous individual experts' solicitation or ERN/Orphanet classification revision), 42% of the attendees of the session already contributed one way or the other to collaborative work with Orphanet.
- Most participants attended a specific Orphanet training session either (30%) through an ERN first training to initiate a collaboration or (40%) during another context/European RD project. The large majority of participants (70%) find it helpful or very helpful.
- In the framework of ongoing or passed ERN/Orphanet collaborations, most participants (86%) are sharing achievements within ERN centres through publications or periodic updates to the steering committee and annual meetings. 14% mentioned they did not yet since too early in the collaboration, but they will share once they have established plans for local ORPHAcodes implementation and guidelines.
- Half of the ERN centres representatives (56%) present during the session mentioned they are commonly sharing coding guidelines within centres, largely mentioning it happened partially and heterogeneously

depending on the units, working groups, hospital documentation or internal using ORPHAcoding documentation

- The main added values of ORPHANET-ERN collaborations according to participants are: *i.* the communication (67%), *ii.* the Nomenclature and Classification methodology of the revision (67%), and *iii.* the available material (67%).
- Finally, including Orphanet managers into the ERN discussions in the early phases (64%), having access to on-line free training material (57%) and organising more frequent meetings between Orphanet managers and experts (43%) were the three items more valuable for the participants to end with a successful collaboration.

Overall satisfaction of all the KOM participants with Orphanet collaboration (past-present-future) is positive, rating above 6 to 10 on a 10 scale their satisfaction.

Conclusions

Overall, the project is progressing according to plans (100% of process indicators reached as of year 1) with satisfactory outputs (81% of Output indicators fully reached) and outcomes (92% of outcome indicators fully reached) so far.

All project milestones and deliverables planned by the end of June 2024 have been achieved on schedule, for milestones a +/- 1 month flexibility was applied.

Project objectives have been fulfilled so far.

Whenever needed, mitigating actions to address the few difficulties and barriers encountered identified thanks both to the quantitative and qualitative evaluations carried out as per description in previous paragraphs, have been planned “on the go” by the Executive committee and also more collectively during the WP4 meetings and thanks to the feedback gathered at the internal mid-Term workshop to which participated 20 out of the 21 partners*.

**for more info on the mid-Term Workshop, please consult the page A detailed report of the OD4RD2 ERN survey is available here: <https://od4rd.eu/03-deliverables> where it will be available at the end of July 2024*