

Deliverable 1.6

OD4RD2 Evaluation Report

March 2026



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Background

This report represents the Deliverable 1.6 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 March 2026 have been achieved on schedule, for milestones a +/- 1 month flexibility was applied. Project objectives (c) were fulfilled.

a) Milestones :

MIs No	Name	WP no	Lead	Means of verification	Expected date	Achievement date	Verified
1	KOM Preparation	WP1	INSERM	Participants list and location available	31 May 2023	1 Apr 2023	Y
2	Mid-term ExCom meetings assessment	WP1	INSERM	6 ExCom meeting reports available	30 Sep 2024	9 Jul 2024	Y (n=13)
3	Target specific survey launched	WP1	INSERM	Link sent to all ERNs and to HCPs Hospital managers in participating countries	31 Jan 2024	31 Jan 2024	Y
4	Mid-project workshop Preparation	WP1	INSERM	Participants list and location available	31 May 2024	14 May 2024	Y
5	Final Meeting Preparation	WP1	INSERM	Participants list and location available	30 Nov 2025	21 Oct 2025	Y
6	ERN collaborations planning (nomenclature)	WP2	INSERM	Timelines communicated to involved ERNs.	30 Sep 2023	12 Sept 2023	Y
7	2nd ERN collaborations planning (nomenclature)	WP2	INSERM	Timelines communicated to involved ERNs.	30 Sep 2024	31 Juillet 2024	Y
8	First Annual checkpoint	WP3	INSERM	At M12 the established indicators for tasks 3.1/3.4 should be calculated to assess the progress; indicators should display 1/3 progress: Indicators calculated	31 Mar 2024	29 Mar 2024	Y
9	Second Annual checkpoint	WP3	INSERM	At M24 the established indicators for tasks 3.1/3.4 should be calculated to assess the progress; indicators should display 2/3 progress.	31 Mar 2025	31 Mar 2025	Y
10	End-users’ state-of-play survey	WP4	BFARM	Delivery of the survey	31 May 2023	4 May 2023	Y

MIs No	Name	WP no	Lead	Means of verification	Expected date	Achievement date	Verified
11	Establishment and delivery of - annual action plan for each national hub	WP4	BFARM	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)	30 Jun 2023	30 Jun 2023	Y
12	HCP survey	WP4	BFARM	At least 75% of national hubs have performed a survey and have presented written results. These have been provided to the executive committee (ExCom) through the project's internal website	31 Mar 2024	7 May 2024	Y 15 reports or 79%
13	Mid-term Report on lessons learned and on improvement of capacity and implementation	WP4	BFARM	Report on lessons learned prepared	30 Jun 2024	24 Jun 2024	Y
14	Establishment and delivery of - annual action plan for each national hub Y2	WP4	BFARM	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)	30 Jun 2024	14 Jun 2024	Y
15	White paper on implementation issues of ORPHAcodes in relation to other code systems		BFARM	White paper prepared	31 Mar 2025	31 Mar 2025	Y released online 31/03/2025
16	Establishment and delivery of - annual action plan for each national hub Y3	WP4	BFARM	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)	30 Jun 2025	13 Jun 2025	Y
17	Final Report on lessons learned and on improvement of capacity and implementation	WP4	BFARM	Report prepared	31 Dec 2025	31 Dec 2025	Y
18	Specifications for data exchange workflow with the EC ERN information system	WP5	INSERM	OD4RD functional specifications document revised/updated	31 Jul 2023	31 Jul 2023	Y
19	Feasibility study and specifications for data analysis towards	WP5	MUW	Specifications document validated by EC and Orphanet teams	31 Dec 2023	11 Jan 2024	Y

MIs No	Name	WP no	Lead	Means of verification	Expected date	Achievement date	Verified
	strategic report(s)						

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> , with the exception of the Del. 5.1 & D5.2 which were sensitive and therefore only shared with the B3 unit and the ERN Coordination as well as BoMS (at the ERN coordination trio request).

Deliverable Related No	Deliverable Name	Lead Beneficiary	Delivery Date
D1.1	KOM Executive summary	INSERM	30 Jun 2023
D1.2	Dissemination plan	INSERM	30 Jun 2023
D1.3	Mid-term Evaluation Report	INSERM	16 Jun 2025
D1.4	Mid-project workshop report	INSERM	26 Jul 2024
D1.5	Final meeting report	INSERM	28 Nov 2025
D1.6	Final Evaluation Report	INSERM	31 Mar 2026
D2.1	Publication of an update of the standardised methodology for the revision of the nomenclature in collaboration with networks of expertise.	INSERM	18 Jun 2025
D2.2	Mid-term report on Orphanet-ERN collaborations	INSERM	28 Jun 2024
D2.3	Global report on Orphanet-ERN collaborations	INSERM	31 Mar 2026
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)	INSERM	28 Jun 2024
D3.2	Final Report on Genetic Annotations (new and updated gene-disease relationships), ORPHAcodes alignments with other terminologies, and newly produced or updated texts (definitions and abstracts)	INSERM	31 Mar 2026
D4.1	State of play survey	BFARM	18 Jun 2025
D4.2	Annual Action plans Year 1	BFARM	18 Jun 2025
D4.3	Mid-term Report on lessons learned	BFARM	18 Jun 2025
D4.4	Annual Action plans Y 2-3	BFARM	02 Jul 2025
D4.5	Final Report on lessons learned	Karolinska	31 Mar 2026
D5.1	Mid-term Report on disease coverage by ERNs	INSERM	07 Nov 2024
D5.2	Final Report on disease coverage by ERNs	INSERM	18 Dec 2025

Table 2 OD4RD2 project deliverable status

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 for each one of them, as well as their target values as indicated in the table below and fully described in the next chapter.

N.B. In this report, the indicators have been calculated by December 2025 (at 33 months). The final project KPIs, as for March 2026 (36 months) will be reported in the Technical Report. Therefore, the values in this report should be considered provisional

Type of KPI	Number planned & pertinent to M33	Number of indicators having reached the planned target	Of which Target Exceeded	% Indicators achievement
Process indicators	14	10	0	71%
Output indicators	15	11	4	73%
Outcome indicators	10	9		90%

Table 3 OD4RD2 project Percentage of Achievement of Process, output and outcome Indicators' Targets at M33 (3 mo ahead of the end of the project)

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

Reporting of process/output/outcome indicators per specific objectives

The proposed specific objectives of the OD4RD2 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. Targets reached for each indicator are indicated in the table below. **N.B. Any deviation from the expected target is explained in the next paragraph.**

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.		
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached Dec25
Collaboration workflow's steps completed for each ERN collaboration and justified in the final report	100%	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).	24	8	16*
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached Dec25
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 as of end of March 2026	2	21
Number of clinical entities (ORPHAcodes) created, modified or inactivated.	800/year	411 in 2023	2013 (732/year)*
Number (%) of completed ERN revision projects for which a final collaboration report has been produced and sent to the ERN representatives.	100%	100%	100%
Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached Dec25

<p>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</p>	<p>90%</p>	<p>This process will be formalized in Y2. N.A.</p>	<p>95%</p>
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<p>Specific Objective 2</p>	<p>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)</p>		
<p>Process Indicator(s)</p>	<p>Target planned</p>	<p>Target Reached at M13 (end of April 2024)</p>	<p>Total Target Reached at Dec25</p>
<p>Validation meetings with the scientific officer for ICD-10, ICD-11 and SNOMED-CT mappings</p>	<p>30/year</p>	<p>30</p>	<p>56*(~21/year)</p>
<p>Survey of OMIM (phenotypes) updates</p>	<p>10 /year</p>	<p>10</p>	<p>20*(~7/year)</p>
<p>Output Indicator(s)</p>	<p>Target planned</p>	<p>Target Reached at M13 (end of April 2024)</p>	<p>Total Target Reached at Dec25</p>
<p>% of ORPHAcodes (at the disorder level) aligned with ICD-10, including inexact mappings</p>	<p>100%</p>	<p>99,70%</p>	<p>96,10%*</p>
<p>% of ORPHAcodes aligned with ICD-11 terms, including inexact mappings</p>	<p>100% by Year 3</p>	<p>66,60%</p>	<p>73,50%*</p>
<p>Number of ORPHA-SNOMED CT human-readable mapping files released</p>	<p>3 (1/Year)</p>	<p>1</p>	<p>3 (1/Year)</p>
<p>Outcome/Impact Indicator(s)</p>	<p>Target planned</p>	<p>Target Reached at M13 (end of April 2024)</p>	<p>Total Target Reached at Dec25</p>
<p>Satisfaction and utility of the nomenclature & mappings according to end users (assessed through online surveys and ERN proactive surveys)</p>	<p>Increase compared to 2021 figures (74%)</p>	<p>7 % increase. In 2023 81% of online users who knew the mapping files were either very satisfied or satisfied of this product/service.</p>	<p>8% increase compared to 2021. In 2025 82% of online users who knew the nomenclature, classification & mapping files were either very</p>

<p>Number of downloads of the nomenclature with alignments and of the human-readable ORPHA-SNOMED CT files in Orphadata</p>		<p>2023 Nom Pack downloads: 1,157</p>	<p><i>satisfied or satisfied of this product/service.</i></p> <p>Nomenclature Pack downloads and ORPHA-SNOMED CT : 5,548 downloads in 2025</p>
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<p>Specific Objective 3</p>	<p>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</p>		
<p>Process Indicator(s)</p>	<p>Target planned</p>	<p>Target Reached at M13 (end of April 2024)</p>	<p>Total Target Reached at Dec25</p>
<p>Survey of OMIM (genes) updates</p>	<p>10 /year</p>	<p>4</p>	<p>14 (target achieved against revised expectations approach)*</p>
<p>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.</p>	<p>95%</p>	<p>127% <i>This indicator was miscalculated at RP1</i></p>	<p>90%*</p>
<p>Output Indicator(s)</p>	<p>Target planned</p>	<p>Target Reached at M13 (end of April 2024)</p>	<p>Total Target Reached at Dec25</p>
<p>New or updated disease-gene associations</p>	<p>450/y</p>	<p>517</p>	<p>1487 (~540/y)</p>
<p>Number of disease summaries/definitions produced or updated</p>	<p>250/y</p>	<p>358</p>	<p>791 (~287/y)</p>
<p>Number of definitions reviewed for quality control</p>	<p>100/y</p>	<p>72</p>	<p>165* (~60/y) target achieved against revised expectations approach *</p>
<p>Outcome/Impact Indicator(s)</p>	<p>Target</p>	<p>Target Reached at M13 (end of April 2024)</p>	<p>Total Target Reached at Dec25</p>

Satisfaction and utility of the relevant category (gene-disease database and encyclopaedia) according to end users (assessed through online surveys and ERN proactive surveys) Encyclopaedia	Increase compared to 2021 figures	A small decrease is observed: -4%. 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	12% increase compared to 2021 and 6% increase compared to 2023. In 2025 97% of online users who knew the encyclopaedia were either very satisfied or satisfied of this product/service.
Satisfaction and utility of the relevant category (gene-disease database and encyclopaedia) according to end users (assessed through online surveys and ERN proactive surveys) Genes	Increase compared to 2021 figures (84%)	A small increase can be observed for the gene database: +3%. In 2023 87% of online users who knew the gene disease database were either very satisfied or satisfied of this product/service.	A small increase can be observed for the gene database: +3%. In 2025 87% of online users who knew the gene disease database were either very satisfied or satisfied of this product/service.
Number of downloads of the Gene-disease associations file in Orphadata	Increase compared to 2021 figures (11,096)	40% decrease for genes, (in 2023: 6,633 genes downloads, while in 2021 approx. 11,096 downloads).	4,578 downloads for genes in 2025=> 41% decrease compared to 2021 *

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.		
Process Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached at Dec25
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	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	20 WP4 meetings plus 14 Open sessions
Survey each year about improved capacity in national hubs	3	1 (detail Available in the paragraph below)	3 (detail Available in the paragraph below)*

Attendance of each hub to Train-for-Trainers (TfT) sessions	1/hub	At least 1 person from each hub attended either one of the sessions. In total, these trainings gathered 58 participants (26 at the Basic from 14 National Hubs and 32 at the Advanced' session from 18 National Hubs,)	At least 1 person from each hub attended either one of the sessions. In total, these trainings gathered 105 participants [58 participants in 2024 (see left cell) and 47 participants in 2025 (basic module: 15 participants; advanced module 19 participants; hands-on in-person training: 13 participants)].
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, (1/hub)	19/19, (1/hub)
National action plan described and published in the project intranet, with annual revisions	1/hub	19/19, (100% of active hubs)	1/Hub including annual revision
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached at Dec25
Availability of the local GitHub or similar ticketing system for national hub helpdesks demands	1/hub	19/19, (1/hub)	19/19, (1/hub)
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%	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.	Not achieved: mitigating action evolution of the TfT content with hands on session to increase confidence
Organisation of Training-workshop or other event by each hub	1/hub	61 trainings & 151 ad hoc meetings or awareness events in 17 countries (90% of active hubs)	173 trainings or workshops & 434 ad Hoc meetings or awareness events in 19 countries (100% of active hubs) => (target achieved against revised expectations approach) *
Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached at Dec25
Compiled report on implementation questions to serve as a lessons learned base for other countries joining the network of hubs in the future	M33	FAQ and lessons learned shared	FAQ and lessons learned shared
Feedback by national training/events participants on knowledge gain after the	85% have gained knowledge	No formalised surveys were carried out	Satisfaction surveys were carried out by 4 NH, see paragraph below

training/event (short survey)			
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	N.A	To be reported in Technical report
	Goal 90% users are satisfied		
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	N.A.	To be reported in Technical report
	Goal 90% users are satisfied		
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	This indicator has been changed into Self-assessment results carried out via a survey at the end of the Tft held in September 2025 showed a clear improvement: participants' confidence in delivering the training increased from an average of 3.7/5 at the beginning to 4.7/5 at the end, while confidence in handling advanced questions improved from 3.1/5 to 3.7/5=> (target achieved against revised expectations approach)*

Specific Objective 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 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)	Total Target Reached at Dec25
Meetings with national hubs on regular basis in order to assess the evolving needs in terms of nomenclature / datasets delivery	N.A.	N.A.	14 Open Sessions & 20 WP4 meetings & github always open
Output Indicator(s)	Target		Total Target Reached at Dec25

Technical advice provided as requested per national hubs including proposal of dedicated services to hospitals	100% of requested advices	100%	100% including multi-stakeholder workshop December 2024 and cross links with JARDIN WP8 T8.3 and 8.4
Outcome/Impact Indicator(s)	Target		Total Target Reached at Dec25
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	>0
Specific Objective 6	To produce analyses 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)	Total Target Reached at Dec25
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	4 B3 meeting calls and 2 F2F in Luxembourg and in Paris, 3 ERN & BoMS meetings, 1 ERN coordinators group F2F=> ~3.6/year
Output Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached at Dec25
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	ERN Chairs & BoMS Meetings 22 May 2024; 22 October 2024; 22 April 2025; 11 Feb 2026 (ERN Co group)
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	Available

Outcome/Impact Indicator(s)	Target planned	Target Reached at M13 (end of April 2024)	Total Target Reached at Dec25
End-users' satisfaction on the report	N.A. will be collected after report delivery goal: >95%	N.A.	B3 Satisfaction assessment ongoing=> will be reported in technical report ?

Table 4. OD4RD2 project Process, output and outcome Indicators' Targets achievement at M33

Indicators deviation explanation & mitigating actions :

1) Specific objective 1. NTR

While the cumulative production over the three-year period fell short of the original target due to underperformance in Year 1, performance in Years 2 and 3 met the annual targets. This demonstrates that the underlying issues were addressed and that the current production model is capable of consistently achieving the required targets moving forward.

2) Specific objective 2

- Validation meetings with the scientific officer for ICD-10, ICD-11 and SNOMED-CT mappings: the target number of meetings was not reached at the end of December 2025, but early 2026 the process runs as usual
- Survey of OMIM (phenotypes) updates: less surveys performed because of technical problems with the script that runs OMIM surveys. However, the OMIM T0 analysis comparing OMIM entities with ORPHAcodes to identify potential missing codes or mapping was finalised, which compensates the months in which the script could not be run, for it provides a complete update of the comparison between OMIM numbers and ORPHAcodes.
- % of ORPHAcodes (at the disorder level) aligned with ICD-10, including inexact mappings : We observe a decrease compared to mid-term as at the end of December 2025 there has been an important increase in newly created disorders and limited validation meetings, please note that the foreseen validation meetings were held during the first trimester of 2026 and this percentage has reached: 97.8% for ICD-10 in March 2026.
- % of ORPHAcodes aligned with ICD-11 terms, including inexact mappings: the target of 100% has not been reached, for the number of newly created disorders increased significantly during the project. However, 100% of RD present in ICD-11 (in the MMS and/or in the foundation) have been aligned, and as a mitigation measure, a systematic approach has been adopted, proceeding classification by classification: at the end of the project, 25 classifications out of 29 have been aligned at 100%.

3) 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) updates: this process indicator did not meet the initially defined quantitative targets as during the project implementation, it became evident that the planned number of survey meetings was overestimated due to a more efficient coordination mechanisms and staff maturity/seniority. The project demonstrated that a lower number of survey meetings was sufficient to ensure quality without compromising outputs (corresponding output indicator reached).
- The global editorial production and quality control indicator target is 350/y of which approx. 100 quality controlled definitions and 250 abstracts/definitions produced. Overall this global target has been reached with more abstracts and definition produced in collaboration with ERNs.

- 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: overall the target achieved is 90%, however for 4% of the agreed list of texts the process was stopped because of a nomenclature revision action was needed.
- Satisfaction and utility of the relevant category:
 - o gene-disease database: we observe an increase compared to 2021 regarding the satisfaction of website users while we observe a decrease of the Orphadata absolute downloads numbers. However, when compared with the other Orphadata downloadable categories, gene-disease associations constantly increased, from 3% of all downloaded data in 2022 up to 15% in 2024.
- These figures testimony of different utilisations of the same data and deserves monitoring over a longer period to confirm a trend.

4) 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.

- 1 training/hub: While the indicator initially set a target of one training per country, implementation experience showed that this assumption did not fully reflect actual country needs. In countries where coders do not directly use ORPHAcodes (i.e. transcoding in the backoffice is in place), no training sessions were conducted due to a lack of demand from end-users, however additional capacity-building activities were conducted (ad hoc meetings). This reflects a context-responsive and demand-driven approach rather than a shortfall in delivery. Consequently, the original target is considered to have been overestimated. When assessed against revised expectations grounded in actual users' demand, the indicator can be considered achieved, as training was delivered where needed and resources were allocated to activities with a clear added value (ad hoc meetings).

- Gain of knowledge measure: this indicator was adapted based on evidence from the evaluation of previous TtT and open sessions and the analysis of GitHub queries. Findings showed that the primary challenge was not a lack of knowledge among National Hubs (NH), but rather a lack of confidence in delivering content. NH members generally demonstrated correct understanding but were often hesitant when presenting or responding to users' questions, frequently seeking validation from the coordination team for already accurate answers. In response, the Training-of-Trainers (TtT) approach was adjusted to include blended learning exercises specifically designed to strengthen participants' confidence in delivering training and addressing advanced questions on ORPHAcodes (e.g. role-play exercises simulating real coders questions scenarios). Self-assessment survey results collected at the end of the TtT showed a clear improvement: average confidence in delivering training increased from 3.7/5 at baseline to 4.7/5 at completion, while confidence in handling advanced questions improved from 3.1/5 to 3.7/5. These results suggest that the adapted training format effectively supported participants in translating their knowledge into more confident delivery. However, further efforts are needed to strengthen capacity in handling advanced questions, which will be a focus of the next project phase (OD4RD3).

5) Specific Objective 5: no deviations

6) Specific Objective 6: no deviations

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 OD4RD2: 19 out of the 19 active National Hubs have set up a helpdesk and 17 out of the 19 National hubs have held at least 1 training session (overall 173 training sessions were held during OD4RD2 and 434 ad hoc meetings/events), this represents more than 2/3 of the hubs having showed raised capacity.

National Hubs specific Indicators

Moreover, during the previous OD4RD project indicators have been defined to follow the National Hubs activities more granularly and have been analysed at the end of the first year of OD4RD2 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. When considering all NH, all targets have been achieved.

Indicator	Target	Achieved in Y1	Total Achieved in Y3
National action plan delivered/updated	2/hub	18 countries (as Romania started in Y2)	2/hub
Number of training sessions in local language	min 1/year per hub, number should increase over the years	17 countries: 61 trainings and 151 at m13 (first evaluation Report (~195 trainings or events/y)	More than 1 event per year per hub has been reached and overall the number has increased: Training / Events: ~221/y Total events 434 in 19 countries Total trainings 18 countries: 173
Number of unique HCPs with ERN units	will depend on number of ERNs HCPs present in the countries, should be >70% of ERNs/HCPs present in the country by the end of OD4RD2	131 ERN units (<i>this indicator was miscalculated at RP1 and report ERN unit instead of unique HCPs</i>)	28% (110/389)* <i>But 601 HCPs have been trained including those hosting ERNs</i>
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	Only 4 NH carried out satisfaction surveys, see results below.
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 See details in table below	434 See details in table below
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	Increase :42% since Y1
- how many tickets/demands were received (by category)! these figures include all demands not only those traced via the Github platform	-	299 • Coding:135 • Nomenclature: 97 • Others: 65	426 • Coding:137 • Nomenclature: 239 • Others: 65
- Analytics of demands according to type of users and	-	• User, clinician: 126	• To be reported in technical report

Indicator	Target	Achieved in Y1	Total Achieved in Y3
typology of demands: /!\ these figures include all demands not only those traced via the Github platform		<ul style="list-style-type: none"> 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)	- To be reported in technical report
- 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)	- To be reported in technical report

Table 4 Compiled specific National Hubs indicators OD4RD2 Year 1-3

*Coverage of National HCPs with at least 1 ERN centre for trainings: the target of 70% has not been reached and the achieved target is significantly lower: 28%. It must be clarified that some NH organised online training sessions and invited all the HCPs present in the territory and did not collect info on HCPs affiliation from the participants and thus resulting in lower numbers and under-reporting (this indicator collection will be clarified in the next project) but also some countries, where SNOMED CT is the primary coding system including for RD, or if a thesaurus terminology is in place and transcoding to ORPHAcodes happens in the back-office, reported a problem related to training organisation : HCPs show little or no interest in receiving ORPHAcoding training.

Details Information about trainings

Country	Number of training sessions/workshops/webinars:	total number of participants	Number of medical departments	ERNs	Total Number of unique HCPs with ERN units	Total number of HCP(Hospitals) present at the training sessions:
Austria	3	41	3	EYE, ERNICA, eUROGEN, BOND, EpiCARE, MetabERN, RND, LUNG, ERKNet, RITA, GUARD-HEART, GENTURIS, PaedCAN	1	1
Belgium	15	197	NA	GENTURIS, MetabERN, ITHACA, ERN RND, ERN LUNG, ERKNet, ERN Skin, EURACAN, VASCERN, ERN CRANIO, Endo-ERN, EuroBloodNet, ERN RITA, ERN BOND	6	7
Bulgaria	43	1276	34	Euro NMD; MetabERN, ERN RND, EuroBloodNet, EndoERN, PeadCan: Total 6 (six)	54	487 (all HCPs from Rare Disease Expert centers and 50% from HCPs from Symposiums, Conferences)

Country	Number of training sessions/workshops/webinars:	total number of participants	Number of medical departments	ERNs	Total Number of unique HCPs with ERN units	Total number of HCP(Hospitals) present at the training sessions:
Czech RP1	2	100		Endo-ERN, ERKNet, ERN BOND, ERN CRANIO, ERN EpiCARE, ERN EURACAN, ERN EuroBloodNet, ERN EURO-NMD, ERN GENTURIS, ERN GUARD-HEART, ERN PaedCan, ERN RITA, ERN-EYE, ERN-ITHACA, ERN-RND, ERN-Skin, MetabERN	3	
Estonia	3	100	13	ERKNet, ERN EpiCARE, ERN EURACAN, ERN EuroBloodNet, ERN GENTURIS, ERN GUARD-HEART, ERN RITA, ERN TRANSPANT-CHILD, ERNICA, ERN-ITHACA, ERN-LUNG, ERN-RND, MetabERN, ENDO-ERN, ERN EURO-NMD, ERN-EYE, ERN PaedCan, ERN RARE-LIVER, ERN ReConnct (total 19)	4	6
Finland	1	110	5	No data collected		5
Germany	1	92	no data collected			
Ireland	11	207	no data collected	ERKNet, MetaB ERN, ERN BOND, Endo ERN, LUNG ERN, ERN EYE, ERN SKIN	5	6
Italy	3	54	Information not requested to participants	Endo-ERN, MetabERN, EuroBloodNet, ERKNet, Transplantchild, RND, ReCONNET (7*). *Please note that the number of ERNs is a conservative <u>estimate</u> (the affiliation was not requested to the participants, so it was not possible to accurately determine the ERNs).	NA	<u>21</u> Please note that this number is a conservative <u>estimate (the affiliation was not requested to the participants, so it was not possible to accurately determine the number of HCPs).</u>
Latvia	6	630	Data not collected	Data not collected		
Lithuania	8	210	23	ERKNet, ERN CRANIO, ERN EpiCARE, ERN EURACAN, ERN EuroBloodNet, ERN eUROGEN, ERN GENTURIS, ERN GUARD-HEART, ERN RITA, ERN TRANSPANT-CHILD, ERNICA, ERN-ITHACA, ERN-LUNG, ERN-RND, MetabERN, Vascern		1
Netherlands	0	0	0	NA		
Norway	28	1366	49	No data collected	2	13
Poland	26	552 (representatives of >150 depts during specialist training courses)		ERN ERKNet, ERN ERNICA, ERN EURO-NMD, ERN EpiCARE, ERN PaedCan, ERN RITA, ERN RARE-LIVER, ERN ReCONNECT, ERN TRANSPANT-CHILD, ERN-ITHACA, MetabERN	6	7 (>50 HCPs was represented during specialist training courses)
Portugal	4	83	no data available	no data collected	7	10
Romania	1					
Slovenia	5	40	8	ERKNet, ERN CRANIO, ERN eUROGEN, ERN GUARD-HEART, ERNICA, ERN-ITHACA, ERN-RND, ERN-NMD, MetabERN, RARE-LIVER	1	1

Country	Number of training sessions/workshops/webinars:	total number of participants	Number of medical departments	ERNs	Total Number of unique HCPs with ERN units	Total number of HCP(Hospitals) present at the training sessions:
Spain	10	249	NA	RITA, BOND, PaedCan, EURACAN, GUARD-HEART, RECONNET, CRANIO, ENDO, EYE, EURO-BLOODNET, RARE-LIVER, ITHACA, ERNICA, RND, EURO-NMD, LUNG, ERKNet, eUROGEN, TRANSPLANT-CHILD, EpicARE, Metab ERN	16	31
Sweden	3	63		ENDO-ERN, MetabERN, ERN Epicare, ERN EURACAN, ERN EURO-NMD, ERN eUROGEN, ERN-LUNG, ERN ERNICA, ERN PaedCan, ERN TRANSPLANS-CHILD, ERN-EYE, ERN-Skin, Vascern	5	5
Total	173	5371	135	0	110	601

Table 5 Information on Trainings by country (number of trainings, number of participants, number of medical departments, ERNs involved, number of unique HCPs hosting ERNs, total HCPs)

: Other events

Number of ad hoc meeting, events and workshops	with hospital managers:	with medical informatics teams:	with medical coders:	with software providers:	Awareness event	Total
Austria	13	4	9	2	2	30
Belgium	0	8	16	1	5	30
Bulgaria	8	6	0	4	8	26
Czech RP1	3	3	1	2		9
Estonia	2	2			2	6
Finland	11		11	1	1	24
Germany			1		1	2
Ireland	2	2	1	2		7
Italy		1			4	5
Latvia			6		6	12
Lithuania	8	9	7	7	3	34
Netherlands	8	27	3	1	7	46
Norway	1	8	29	39	1	78
Poland	12	17	20	15	10	74
Portugal	3		3		3	9
Romania					5	5
Slovenia RP1	1	2	3	2		8
Spain	3	3	5	0	8	19
Sweden		2	8			10
Total	75	94	123	76	66	434

Table 6 Other events : ad hoc meetings/workshop by type of stakeholders & awareness events

Indicator	Countries	AT	BE	BG	CH ¹	CZ	DE	ES	EE	FI	IE	IT	LT	LV	NL	NO	PL	PT	RO	SE	SI
State of play Survey	18	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
ERN survey	16	Y	Y	Y	N/A		Y	Y	Y	Y	Y	Y	Y	yes	Y	Y	Y	Y	N.A.	Y	N
1st action Plan	19	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y.	Y	Y
2 nd Action Plan	19	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y.	Y	Y
Github created			Y	Y		Y	Y	Y	N	Y	Y	Y	Y	Y	Y	Y	Y	N	N	Y	

Table 7 Specific National Hubs indicator by country OD4RD2 April 2023-December 2025

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 Plans 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 coding, trainings, tools etc.

Qualitative evaluation

Evaluation of Orphanet and Orphadata users satisfaction and use

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

Every year, a satisfaction survey of the Orphanet website users is launched via a pop-up window appearing on the first page users landed on. The survey is translated in 8 out of 9 languages of the website (i.e. English, French, Spanish, Italian, Portuguese, Dutch, German and Polish) and is displayed in the language of consultation via the pop-up. The survey is closed after a target amount of answers is reached. This survey is 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 results from all of the languages of the survey are consolidated and then analysed. The results of these analyses are presented in a report available online with elements of comparison as regards previous survey’s results:

- [Orphanet 2025* Users Satisfaction Survey](#)
- [Orphanet 2024* Users Satisfaction Survey](#)
- [Orphanet 2023* Users Satisfaction Survey](#)
- [Orphanet 2022* Users Satisfaction Survey](#)

In this section of the evaluation report, we highlight the results which are pertinent to the project-related activities, i.e. nomenclature, classifications, alignments, genetic info and texts. For this purpose, 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 number of respondents who replied to this question, % of regular users and % of total responders is indicated in the table. For the purpose of this analysis, we subtracted the users that did not know the service. Over 4 years (2022-2025), 86% of the users, in average, that knew the services are either very satisfied or satisfied with the products listed 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	% users that know the product and are either very satisfied or satisfied with it. Survey 2024	% users that know the product and are either very satisfied or satisfied with it. Survey 2025	Average per type of product
Numbers of responders to this question	3,458	3,847	2,372	2,294	N.R.
% of regular users	82%	82,5%	82%	96%	N.R
% of the total responders	37%	39%	47%	68%	N.R

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	% users that know the product and are either very satisfied or satisfied with it. Survey 2024	% users that know the product and are either very satisfied or satisfied with it. Survey 2025	Average per type of product
Disease summaries	96%	91%	98%	99%	97%
Classifications	88%	88%	87%	91%	88%
Inventory of genes	88%	87%	82%	87%	87%
Orphanet nomenclature	83%	87%	77%	78%	81%
Alignments	80%	81%	75%	77%	79%
Average	87%	87%	82%	87%	86%

Table 8 % of users that know the product and are either very satisfied or satisfied with it, comparison 2022 vs 2023 vs 2024 vs 2025

I. Orphadata downloads

Nomenclature Pack, classifications, linearization, and genetic information per disease is also available from the data download platform Orphadata (www.orphadata.com) and the www.orphacode.org website.

Data	Downloads in 2025	Downloads in 2024	Downloads in 2023
Nomenclature Pack & associated files (Master File+Differentials+SNOMED-CT mapping file)	5,548	3,335	1,157
Classifications	Not available yet	10,000	86,363
Genes	4,578	5,769	6,633
Linearisation	Not available yet	1,154	1,026

Table 9 Orphadata downloads by type of data and by year

User satisfaction surveys sent to ERNs

Regarding ORPHAcodes implementation:

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 2024, in order to be able to identify ORPHAcoding practices and barriers in implementation to be able to fine tune both national actions and 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 database of expert resources by country (https://www.orpha.net/en/expert_centres/european-reference-network), and each National Orphanet Nomenclature Hub could add additional contacts if needed

A minimum set of questions was also established and each National Orphanet Nomenclature Hub could add questions if needed at national level.

16 National Orphanet Nomenclature 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 together with an analysis by ERN.

In total, 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 [information available for only 15 hubs out of 16] and 544 answers were received (average 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>

Regarding Scientific Collaborations with Orphanet:

As part of our ongoing effort to improve the collaboration process, we recently introduced in our methodology a short survey sent to ERNs once the collaboration project for revising the Orphanet nomenclature is completed. We received a total of 10 responses since several ERNs provided more than one feedback (one has 2 projects and in other cases more than one of the experts involved replied).

The feedback is summarized in table 10. The majority of contacted ERNs have expressed a high level of overall satisfaction with the collaboration process, the channels used to communicate, and the regularity of discussion sessions. Satisfaction with the deadlines is slightly lower, and this issue will be examined in more detail. Nevertheless, the feasibility of achieving this objective is contingent upon the availability of experts in the field.

The majority of the respondents intend to use the ORPHAcodes resulting from the collaborative revision: in communications, in patient registries, and in national applications asking for an expertise center on RD. One limitation of the survey is that in some cases the response was provided by the ERN project manager, that explains the 'No', 'Not possible to evaluate' and 'I have no direct say in coding' answers in questions 8 and 10.

The results of this survey suggest that the current workflow has a positive impact on different levels. There was a consensus amongst the respondents regarding the impact of the collaboration on improving the accuracy of the Nomenclature and Classification of the RDs, and the positive effect on coding. The willingness of collaborators to engage in RD texts revision, future follow-up revisions, and the initiation of new projects was also demonstrated. Furthermore, some ERNs have expressed a notable enthusiasm for the dissemination of the co-constructed scientific knowledge through international publications.

Overall, the survey validated the proposed Orphanet methodology for ERN collaborations and revealed the multiple benefits of Orphanet-ERNs scientific interactions.

Number of ERNs/ number of feedbacks received 6/10		
1.	How do you rate globally the collaboration process?	Excellent (90%)/Acceptable (10%)
2.	How is the level of satisfaction with the channel used to communicate?	Excellent (90%)/Acceptable (10%)
3.	How is the level of satisfaction with the frequency of discussion between the Orphanet Manager and the ERN contact point?	Excellent (90%)/Acceptable (10%)
4.	How is the level of satisfaction with the deadlines?	Excellent (70%)/Acceptable (30%)
5.	If you chose "Not possible to evaluate", please can you briefly explain why?	NA
6.	Has the collaboration improved the accuracy of Nomenclature and Classification of RD?	Yes (100%)
7.	Do you think that the changes will have a positive impact on the coding?	Yes (100%)
8.	Do you intend to use the newly revised codes to code your patients?	Yes (60%)/No (20%)/ Not possible to evaluate (20%)
9.	If you intend to use the newly revised codes to code your patients: in which context do you plan to use the codes?	1/6 "In our national application expertise center for rare disease" 2/6 "In communications" 1/6 "In our hospital and ERN" 1/6 "Out- and inpatient care, registry" 1/6 "Clinical practice and research (national data base for dysraphisms)"
10.	If you chose 'Not possible to evaluate', please can you briefly explain why?	1/3 "Clinically not relevant" 1/3 "Not currently employed in this role" 1/3 "I have no direct say in coding"
11.	Would you be available to contribute to the writing of the abstract of the clinical entities related to your clinical specialty? (That means being available to work again with Orphanet to update the clinical information shown in Orpha.net)	Yes (90%), No (10%)
12.	Would you be available to revise the recently modified classification to help Orphanet in their quality control process? (That means being available to work again with Orphanet to confirm the adequacy and accuracy of the classification in five-year timeline)	Yes (90%), No (10%)
13.	Do you plan to contact Orphanet for other projects in the future?	Yes (80%), No (20%)
14.	If you plan to contact Orphanet for other projects in the future: what would be your request (a revision of the classification, a nomenclature update, ORPHAcodes creation, ORPHAcodes inactivation...)	1/6 "Not sure yet" 3/6 Yes, revision of a classification 1/6 "All" 2/6 Nomenclature changes 1/6 "Writing of manuscript about the revision"

Table 10 Preliminary results of the survey sent to ERNs to evaluate the satisfaction of the results and steps of the revision process with Orphanet

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

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

173 trainings had been held by the national hubs and 434 ad hoc meetings and awareness events, with more than 5,371 participants representing more than 110 Hospitals hosting ERN expert centers and 601 HCPs overall, the national hubs were able to report that the feedback from the participants after the training sessions has been very positive.

N.B. Some countries, where SNOMED CT is the primary coding system including for RD, or if a thesaurus terminology is in place and transcoding to ORPHAcodes happens in the back-office, reported a problem related to training organisation : HCPs show little or no interest in receiving ORPHAcoding training.

Four countries reported the following feedback collected after the training sessions, of which 1 (Belgium)'s results are not exploitable. In that case, the survey was sent by mail after the training session which resulted in a too small response rate. Italy, Spain and Sweden collected feedback after the training sessions in a systematic way, through questionnaires that were organised differently but that can be summarized as follows:

The overall satisfaction was high (4.44/5 in Spain, 77.2% of respondents in Sweden). In particular, training relevance was highly rated (5.5/6) in Italy, but results were less positive in Spain (62% did not respond to this question and 31% judged the training relevant; as did 45% of Swedish respondents while 41% remain neutral). These results can be attributed to the fact that ORPHACodes are not widely adopted in these 2 countries. The quality and format of the trainings were rated very high in the 3 countries (5.4/6 in Italy, 4.3/5 in Spain, 86.3% in Sweden satisfied or very satisfied with the format, and 95.4 with the quality of the trainer). In terms of difficulty, 79% of Italian respondents thought that they need further trainings for extra clarifications, and only 27% of Swedish respondents judged the training difficult, underlying the need for advanced sessions in complement. Regarding the impact of the trainings, 84% of Italian respondents judged that it will be positive, and 59% of Swedish respondents said that they significantly gained knowledge. Finally, in Sweden, the topic the most appreciated was the Orphanet classification system (63% of respondents), whereas the less appreciated were the mappings to other terminologies (9%). The detailed results of each satisfaction survey are given in Annex.

Satisfaction and content evaluation surveys of face-to-face and online events organised during the project

A series of events and workshops gave us the opportunity to evaluate the adequacy of the project's outputs compared to end-users' needs.

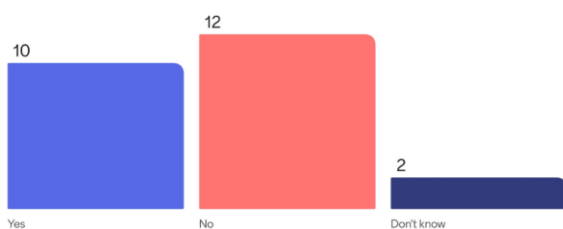
Workshops :

The 19 March 2025, an OD4RD2 session took place at Jardin Day For Hospital Managers And Policy Group, to present the added-value of ORPHACoding and the support and tools available at national level for ORPHACodes implementation and use.

A survey was launched live during the presentation in order to collect live feedback from the Hospital Managers present at the meeting.

The first questions aimed at assessing whether ORPHACodes were already implemented in their hospitals: 50% responded positively, and in that cases, half of the respondents have ORPHACodes implemented in their EHR systems. If their answer was no, we further investigated if there were actual plans to implement ORPHACodes, which was the case in 80% of the cases..

Have you implemented ORPHACodes in your Hospital?



If no, are there plans to implement ORPHACodes?



Figure 1 Q1 & Q2 Survey results Jardin Day For Hospital Managers And Policy Group

Then we investigated the audience awareness of the Orphanet Nomenclature National Hubs which was high (80%).

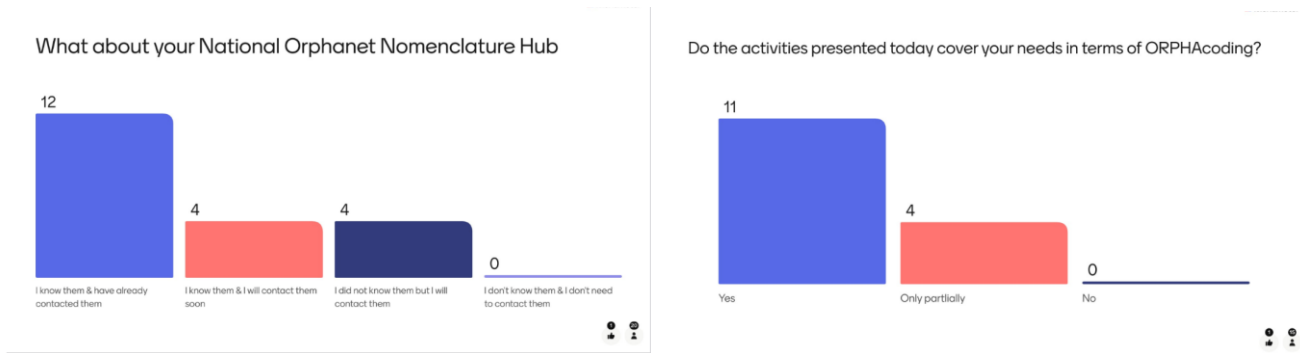


Figure 2 Q3 & Q4 Survey results Jardin Day For Hospital Managers And Policy Group

Finally, the audience was asked to express ORPHAcoding needs in order to eventually adapt OD4RD2 delivery plan accordingly. Expressed needs converged with the already planned deliveries and action in place in OD4RD2 and JARDIN, reflecting that the National Nomenclature Hubs Network organisation is well fit for purpose, known and useful. However, this also shows that there is need for further disseminating actions in order to increase awareness on the project outcomes and available services.

Mentimeter

If not or partially, what are your ORPHAcoding needs?

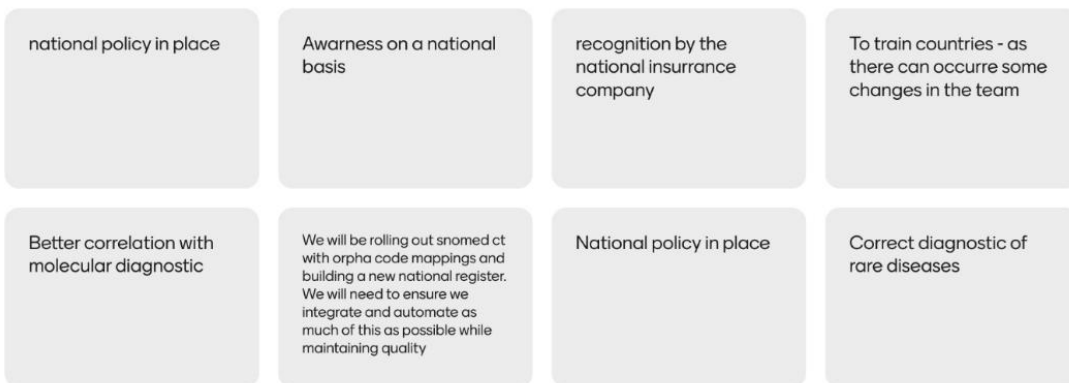


Figure 3 Q5 Survey results Jardin Day For Hospital Managers And Policy Group

The 18 December 2024, OD4RD2 organised an online ORPHAcodes technical implementation workshop. At that occasion, we conducted a needs assessment survey that also confirmed.

A report has been issued after the Workshop: https://od4rd.eu/communication-material/OD4RD2_Technical-Workshop_181224_ReportVF.pdf

Project Meetings:
Kick off Meeting satisfaction survey

As of M1 the ERNs Coordinators were invited to the OD4RD2 kick off meeting that took place on the 13 and 14 of April 2023. 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 (41.4%) of which 6 ERN representative and 2 hospital managers, 1 unknown and the others being Orphanet team representatives. 93% of participants were satisfied (strongly agree or agree) and all the respondents knew the project in general before attending with only 4 not knowing the specific activities carried out. 93% considered that the meeting was helpful in better understanding the project objectives and plans.

	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

Figure 4 Answers regarding the level of satisfaction Kick Off Meeting

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 during the duration of the project in order to improve this aspect (i.e. attendance of OD4RD2 officers 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 Kickoff 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 clinicians' needs regarding ORPHAcodes (at minimum, 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 timeliness problems as the Orphanet classification does not include 'other/unspecified' codes as does ICD for instance.

- 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 sessions 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%) found trainings 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 it is too early in the collaboration, but they will share the outputs once they have established plans for local ORPHAcodes implementation and guidelines.
- More than 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 of 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 achieve 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 of satisfaction.

Mid-Term Internal Workshop Satisfaction Survey

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 (WP) for the last phase 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 WP . The relevant indicators were also presented so as to help fine tuning the activities, discuss possible improvements as well as mitigating actions if needed. The last session of the day focused 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-oriented » action plan. From the participants' perspective, collected thanks to a feedback survey launched during the meeting (17 answers out of 46 participants received (37%), distribution by category of participants (fig.5) 100% of respondents were either satisfied (strongly agree or agree) (fig.6). 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. Of these, all knew the project in general and only 4 did not know the specific activities carried out (fig. 7).

Respondents:

17 answers out of 46 participants were received at feedback survey

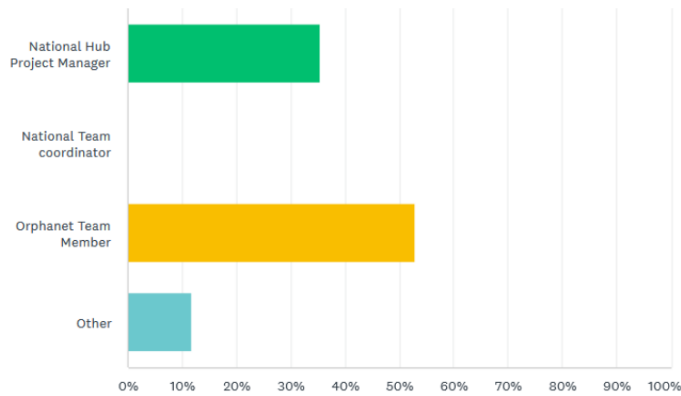


Figure 5 Type of Participant

	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 Workpackages 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

Figure 6 Project Awareness Measurement

	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

Figure 7 Workshop Satisfaction

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 the most included sharing experiences amongst National hubs, the brainstorming (sitcky notes) session, the interaction with the coordinating team and the coordinator’s final remarks.

What participants disliked was that there was enough time to deep dive in some topics (1 participant) and lunch time being scheduled too late (1 participant)

OD4RD2 Final Meeting Satisfaction Survey

The OD4RD2 Final Meeting was held in Paris on 5 & 6 November 2025. The first day gathered 46 participants and was limited to OD4RD2 project partners and the Orphanet Network, allowing for an in-depth discussion of project achievements and the timeline of the next few months. The goal was to fine-tune upcoming activities and identify potential improvements. Particular attention was given to the lessons learned from the Network of National Hubs. The meeting also served as an opportunity to present the proposed objectives of the new OD4RD3 project and to brainstorm on areas for enhancement.

The second day was open to external stakeholders and gathered 61 participants, including all project partners, representatives from 17 ERNs, and HADEA, DG SANTE B3 UNIT, ERDERA and JARDIN representatives. The aim of this second day was to broaden the scope of the meeting and ensure comprehensive coverage of all relevant topics with regards to rare disease data by fostering collaborative brainstorming and actively inviting stakeholder feedback to capture all needs.

A survey was launched to collect general feedback on the meeting and suggestions for improvement, 20 answers were received (33%).

Participants were asked whether they strongly agree/agree/uncertain/disagree/strongly disagree with the following sentences, in order to assess previous knowledge of the project and project activities, whether the meeting was effective in helping deeper understanding and also to collect satisfaction on OD4RD2 activities and activities planned in OD4RD3. Finally, the last 4 questions aimed at assessing whether the agenda of the meeting was appropriate and allowed discussion and if expectations were met. Satisfaction rate is indicated next to each question.

- I was aware of the existence of the OD4RD-OD4RD2 projects before attending this meeting (100% Agree or strongly agree)
- I was also aware of the different OD4RD2 activities before attending this meeting (80% Agree or strongly agree)
- The meeting helped me better/further understand the activities carried out (95% Agree or strongly agree)
- I am satisfied with the OD4RD2 activities (95% Agree or strongly agree)
- I am satisfied with the planned OD4RD3 activities (100% Agree or strongly agree)
- The pacing of the meeting was appropriate (100% Agree or strongly agree)
- I think there was enough time for discussion (85% Agree or strongly agree)
- The collaborative working sessions met my expectations (100% Agree or strongly agree)
- The meeting met my expectations (100% Agree or strongly agree)

When asked what they particularly liked, we received the following answers:

- Very well structured
- Everyone was very welcoming and open, the discussions were really nice
- Nice to meet all the ERN coordinators.
- Thank ORPHANET’s team for your work, organization, and support!
- Strengthening of networking
- The friendly atmosphere; AR and her team!
- The morning session
- That ERNs were present

When asked if there is something you particularly disliked during the meeting, participants said no.

A final question to collect further feedback, the following comments were received:

- I would highly recommend the use of badges with the name of the person and the institution. While a lot of faces were somewhat familiar, I have seen most of them through a camera in online meetings so the transition to in person can be a bit disorienting. From experience, having visible badges can help people connect more easily in those situations.
- The breakout sessions were too focused on the ERN representatives.
- Dinner was a bit late

All these remarks will be taken into consideration when organizing upcoming meetings.

Conclusions

Overall, the project has progressed according to plans (71% of process indicators reached) with satisfactory outputs (73% of Output indicators fully reached) and outcomes (90% of outcome indicators fully reached). Although some process and outcome indicators are not yet fully achieved, they remain on track to be met by project completion, supported by effective mitigating actions. While some process and output indicators remain below target, the intended outcomes have been fully achieved, therefore, the project has effectively delivered its intended results.

All project milestones and deliverables planned by the end of December 2025 have been achieved on schedule, while 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. That proves that regular monitoring of activities and a mid-term state of play are fundamental to limit eventual deviations and to adapt to our evolutive environment.

Extensive actions have been undertaken to assess the needs of the different project user groups, both at national and transnational level, including ERNs, Hospital technical teams, and Hospital managers in different meetings organised within the project or by other projects were sessions dedicated to OD4RD2 or to ORPHAcoding were held. Overall, the needs identified through these assessments are aligned with the actions and deliverables already planned within the OD4RD2 project. This suggests that the project has reached a high level of maturity and demonstrates a solid capacity to anticipate and address stakeholder requirements effectively. In addition, when new or emerging needs have been identified during the course of the project, these have been carefully evaluated and, where more relevant, redirected to other appropriate projects such as JARDIN or myHealth@EU, rather than being addressed within OD4RD2. This approach ensures that each need is handled in the most suitable context, while also promoting mutualisation of efforts and fostering synergies across related initiatives. Such coordination helps to avoid duplication, optimise resources use, and strengthen overall impact at a broader programme level.

At the same time, this situation highlights an important area for improvement to be addressed in OD4RD3. While the project appears to be responding appropriately to users’ needs, it also indicates that not all different stakeholders/users may be fully aware of the activities, outputs, and solutions being developed. This points to a need for strengthening dissemination and communication efforts, in particular towards decision-makers outside the rare disease sphere as for example, national health data digitalisation offices, and in particular in countries where other generic terminologies (i.e. SNOMED CT) are mandatory. Enhancing these activities will help ensure that all relevant stakeholders are adequately informed, engaged, and able to benefit from the project’s results, ultimately maximizing its impact and uptake.

Annex Feedback survey from NH trainings

Belgium: Feedback surveys conducted since 2025. Only 4 responders (form sent after the training, via email).

Italy: Results of the surveys delivered after the two trainings delivered in 2025 (July and October)

Total number of respondents=19 (49% of the participants)

- How do you rate the relevance of the topics covered to your learning needs? Average= 5,5 (from 0 to 6)
- How do you rate the educational quality of this program? Average= 5,4 (from 0 to 6)
- How do you rate the usefulness of this event for your training/updating needs? Average= 5,7 (from 0 to 6)
- How do you rate the event, in terms of duration? Right duration= 84,2%; Too short= 15,8%; Too long= 0%
- Have you encountered a situation similar to the one you experienced during the training event in your daily practice? No= 68%; Yes= 26%; Other= 0% (options: Yes/No/Other).
- Do you think the course you took will have a real impact on your daily work? No= 16%; Yes= 84% (options: Yes/No).
- Do you think you need further clarification? No= 21%; Yes= 79% (options: Yes/No).

Spain: Feedback from 131 participants (53% of the total)

Questions:

1. Overall satisfaction related to (highest positive score=5):
 - a) Enrolment: 4.67
 - b) Contents: 4.52
 - c) Difficulty level: 4.10
 - d) Format of the courses: 4.5

2. Satisfaction related to the online material facilitated (highest positive score=5):
 - a) Difficulty level of the recorded videos: 3.97
 - b) Time allowed to review the material: 4.37
 - c) Quality of material: 4.44
 - d) Quantity of material: 4.42

3. Satisfaction related to the live session (highest positive score=5):
 - a) Format of the practical session: 4.56
 - b) Practical exercises: 4.42
 - c) Quality of material: 4.44
 - d) Quantity of material: 4.54
 - e) Clarity of material: 4.37
 - f) Difficulty level: 4.37
 - g) Session schedule: 4.18

h) Session length: 4.08

4. After taking the course, do you consider that the knowledge acquired is sufficient to be able to work with this coding system?

- Yes: 31
- No: 14
- No answer: 62
- Other comments: 24

Sweden: Feedback from 22 out of 28 participants from the only training that had evaluation survey (Percentages are out of 100% of the participants whom answered the evaluation survey)

1. Which part of the training did you appreciate most?

- a. The Orphanet Classification system: 63,6%
- b. The introduction to Orphanet: 13,6%
- c. Tools for coding: 13,6%
- d. The mapping of ORPHAcodes: 9%

2. To what extent do you feel that your knowledge in the subject has increased after the training? (1 (no increase) to 5 (definitive increase))

- a) 1: 0%
- b) 2: 22,7%
- c) 3: 18,1%
- d) 4: 36,3%
- e) 5: 22,7%

3. How relevant was the training content to your work or role? (1 (not relevant) to 5 (relevant))

- a) 1: 0%
- b) 2: 13,6%
- c) 3: 40,9%
- d) 4: 18,1%
- e) 5: 27,2%

3. How difficult was the training? (1 (not difficult) to 5 (difficult))

- a) 1: 0%
- b) 2: 54,5%
- c) 3: 18,1%
- d) 4: 27,2%
- e) 5: 0%

4. How satisfied are you overall with the training? (1 (not satisfied) to 5 (very satisfied))

- a) 1: 0%
- b) 2: 9%
- c) 3: 13,6%
- d) 4: 40,9%
- e) 5: 36,3%

5. How well did communication and interaction work in the digital format? (1 (badly) to 5 (very well))

- a) 1: 0%
- b) 2: 0%
- c) 3: 13,6%
- d) 4: 31,8%
- e) 5: 54,5%

6. How satisfied are you with the trainer(s)' competence and ability to convey the content? (1 (not satisfied) to 5 (very satisfied))

- a) 1: 0%
- b) 2: 0%
- c) 3: 4,5%
- d) 4: 40,9%
- e) 5: 54,5 %