D1.5 OD4RD2 Closing Meeting Executive Summary

November 2025

OD4RD
Orphanet Data For Rare Diseases

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Content summary

Introduction

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

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

The OD4RD2 Final meeting was held on the 5th and 6th November 2025.

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Day 1

Day 1 Participants (restricted to project partners)

Orphanet teams: Austria, Belgium, Czech Republic, Finland, France, Germany, Greece, Iceland, Italy, Latvia, Lithuania, Netherlands, Norway, Portugal, Poland, Sweden and Spain.

Day 1 Executive summary

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 past 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 reflect on areas for enhancement.

Day 2

Day 2 Participants

Orphanet teams: Austria, Belgium, Czech Republic, Finland, France, Germany, Greece, Iceland, Italy, Latvia, Lithuania, Netherlands, Norway, Portugal, Poland, Sweden, Switzerland and Spain.

ERN representatives: ERN PaedCan ; RITA ; eUroGEN ; ERKnet ; ERN-EYE ; ERN CRANIO ; RARE LIVER ; ERN BOND ; ERN ENDO, ERN LUNG ; ERN TransplantChild, ERN ERNICA, GUARD-HEART, ITHACA,RITA, ERN-RND, ERN SKIN

Other stakeholders/project representatives: French Ministry of Health-DGOS - JARDIN Representatives; LUMC-ERDERA representative, HADEA, DG-SANTE B3 UNIT

Day 2 Executive summary

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.

The morning session was dedicated to presenting and discussing the OD4RD2 project’s achievements and their impact on different stakeholders, particularly regarding the ERNs’ disease coverage analysis. Particular attention was also given to the lessons learned from the Network of National Hubs. The National Hubs have ensured continuous and tailored support for the local implementation of ORPHAcodes in national Health Care Providers hosting ERNs. They facilitate the work of coders by

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providing training and maintaining helpdesk services for day-to-day coding needs in national language. The meeting also served as an opportunity to present the proposed objectives of the new OD4RD3 project and to reflect on areas for enhancement with external stakeholders. This next project aims to continue ensuring the production and delivery of Orphanet nomenclature of RD that is regularly updated and aligned with evolving knowledge and coding requirements, including the need for interoperability of rare disease (RD) data across health and research systems. Another key objective is to further support and promote the expansion of RD codification in all European Member States by providing both human and technical support for implementation, adoption, and harmonised use of ORPHAcoding standards across Europe. Continued close collaboration with the ERNs will ensure that their expertise and registry data contribute to knowledge generation and dissemination through an ever-improving knowledge base.

In addition, insights were shared on synergies with the JARDIN Data Management work package activities.

These extensive presentations set the stage for the afternoon program, which consisted of two rounds of parallel working groups. The objective was to brainstorm with ERNs on potential unmet needs and gaps that should be addressed in the final months of OD4RD2 and in OD4RD3. Discussions focused on:

- Improving scientific collaboration with ERNs, including nomenclature and scientific information: from the discussions with the audience this process seems to be already mature and no additional suggestions were received if not trying to increase the number of collaborations per year, which is the already planned intention in OD4RD3 if the current proposition is received.
- Supporting ERN registries, particularly in transforming their data into reusable knowledge: this item needs to be further addressed in a more focused working group (with ERN registry group and Registry managers) which will be organized in Q1-Q2 of 2026 to better assess needs and agree on an implementable action plan.
- How to enhance ORPHANET's contribution to the dissemination of ERN coding guidelines and other knowledge: the discussions pointed out the need to further assess the state of play of existing guidelines across the 24 ERNs and the need for Orphanet to propose a more structured support on this aspect.
- How to further support the implementation and adoption of ORPHAcodes in Member States in parallel with NH actions and JARDIN WP8 actions: during this working group actions on how ERNs coordinators can also join forces were discussed and will be further discussed with the ERN coordinators group.

Conclusion

Across both days, the meeting provided a comprehensive and forward-looking platform for reflection, exchange, and strategic planning. The first day enabled OD4RD2 project partners to engage in detailed technical discussions, consolidating lessons learned and aligning on the refinements needed to optimise ongoing activities. The essential role of the National Hubs in supporting ORPHAcodification efforts and maintaining high-quality, locally adapted implementation was clearly reaffirmed.

The second day broadened the dialogue to include a wide range of external stakeholders, whose contributions enriched the discussions on project achievements, impacts, and future directions. The

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collaborative working group sessions proved particularly valuable in identifying remaining gaps and emerging needs to be addressed in the final months of OD4RD2 and in the design of OD4RD3.

Overall, the meeting successfully strengthened collaboration among all actors involved—from ERNs and National Hubs to broader stakeholders. It laid a solid foundation for the next OD4RD3 project, ensuring that future work will continue to advance harmonised RD codification across Europe, enhance scientific collaboration with ERNs and data usability, and support high-quality, interoperable knowledge generation for the benefit of the entire rare disease community.

From the participants' perspective—collected through a feedback survey administered during the meeting (20 respondents)—the results were highly positive. All participants (100%) agreed or strongly agreed that the meeting met their expectations. While all respondents were already familiar with the project in general prior to attending, only four were unaware (uncertain) of the specific activities carried out. Moreover, 95% indicated that the meeting helped them gain a better understanding of the project's objectives and plans. Satisfaction levels were consistently high: 100% of respondents were satisfied with the OD4RD2 project activities as well as with the planned activities under OD4RD3. Additionally, all participants (100%) felt that the pace of the meeting was appropriate, and 95% believed there was sufficient time for discussion. Finally, 90% stated that the collaborative sessions met their expectations, while two respondents were uncertain.

Annex 1 OD4RD2 Closing Meeting Agenda Day1 & Day2

Orphanet Data 4 Rare Diseases-OD4RD2 Final Meeting

Day 1: 5th November 2025
Collège Franco-Britannique of the Cité Universitaire -9 Boulevard Jourdan 75014 Paris
N.B. This half day is only for OD4RD2 project partners & Orphanet Network

16h00 -16h30 Networking Coffee

16h30-17h00 Presentation of OD4RD3

17h00 -17h15 Q/A

17h15-18h30

- **OD4RD2 Lesson learned focus on WP4**
- **Presentation of next day's agenda and organisation of Working groups**

19h30 Networking Dinner

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Orphanet Data 4 Rare Diseases-OD4RD2 Final Meeting

Day 2: 6th November 2025

Collège Franco-Britannique of the Cité Universitaire - 9 Boulevard Jourdan 75014 Paris

9h30-10h15 Welcome Coffee-Networking session

10h15-12h30 Session 1

- Welcome message (DG Santé and HADEA representatives, Project Coordinator)
- OD4RD2 achievements
- RD Codification: lessons learned
- Synergies with JARDIN
- Next Direct Grant Plan: Focus on gaps yet to cover

12h30-13h30 *Lunch*

13h30-16h30 Session 2
13h30-14h30 Parallel working sessions to identify gaps and needs 1

- Explanation
- Scientific collaborations with ERNs: nomenclature and scientific information
- ERN registries: how to transform their data in reusable knowledge

14h30-15h15 Parallel working sessions to identify gaps and needs 2

- How Orphanet can help to disseminate ERN coding guidelines & other knowledge
- Implementation & adoption of ORPHAcodes in MS

15h15h15h45 *Coffee break*
15h45 -16h15 Plenary: Restitution of breakout sessions
16h15 Participant survey: knowledge on project and satisfaction+ Closing remarks

16h30: End of the meeting

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