D1.1 OD4RD2 Kick Off Meeting Executive Summary
April 2023

Content summary

Introduction

The OD4RD2 project is a 3-year project that builds on, and expand, 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 Kick Off meeting was an online event held on the 13th and 14th April 2023.

DAY 1

Participants (restricted to project partners)

Orphanet teams: Austria*, Belgium*, Bulgaria*, Czech Republic*, Germany*, Estonia*, Finland*, France*, Ireland*, Italy*, Georgia, Latvia*, Lithuania*, Netherlands*, North Macedonia, Norway*, Portugal*, Poland*, Romania*, Serbia, Slovenia*, Sweden*, Switzerland, Spain* and Turkey.

Observer: Iceland

Executive summary

The first day gathered 70 participants and was restricted to OD4RD2 project partners & Orphanet Network, in order to discuss in depth, the OD4RD project achievements (www.od4rd.eu) and the new OD4RD2 project objectives and its timeline so as to fine tune the activities and discuss possible improvements. Detailed presentations around each Workpackages activities were given :

In the frame of Workpackage 2, the Orphanet nomenclature (ORPHAcodes) and classification of RD will be produced and maintained, in collaboration with ERNs, in line with the evolution of knowledge. The procedure describing the workflow of collaborations was presented (here) together with the prioritization criteria. Followed by the state of play of the collaborations achieved and the lessons learned from these experiences together with the planning for 2023.

In the frame of Workpackage3, Orphanet Knowledge and information base around RD will be fed in collaboration with ERNs in order 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..) to facilitate cross—coding. The data and information associated with RD will be also expanded, in particular genetic data and textual information associated with RD.

The meeting was also the occasion to kick off the Workpackage 4 work dedicated to scale up and to further develop the activities of the Orphanet Network of Nomenclature Hubs in 19 MS across the EU as well as Norway and Switzerland. The National Hubs will continue ensuring support for the local implementation of ORPHAcodes in national Health Care Providers s hosting ERNs to facilitate the work of coders by providing trainings and by ensuring helpdesk activities for day to day ORPHAcodes support, in a more tailored way thanks to the lessons learned from OD4RD and the national users'needs assessment. With OD4RD2, they will also develop specific support for different target groups such as IT and hospital managers amongst others.

The objective of Workpackage 5 is to provide support to the EC in its ERN strategy including dedicated IT systems. The tools and services around ORPHAcodes that have been developed were presented (https://www.orphadata.com/orphanet-nomenclature-for-coding/) as well as results of the analysis of disease coverage by ERN centres, including complementarities (overlaps) and gaps in terms of disease fields.

Day 2

Participants

Orphanet teams: Austria*, Belgium*, Bulgaria*, Germany*, Estonia*, Finland*, France*, Ireland*, Italy*, Georgia, Latvia*, Netherlands*, Norway*, Portugal*, Poland*, Romania*, Slovenia*, Sweden*, Switzerland and Spain*.

Observer: Iceland

ERN representatives: ERN PaedCan ; RITA ; MetabERN ; eUroGEN ; ERKnet ; ERN-RND ; EURO-NMD ; ERN-EYE ; ERN CRANIO ; RARE LIVER ; ERN GENTURIS ; RECONNECT; ERN EYE ; ERN BOND ; EUROBLOODNET ; ERN LUNG ; ERN SKIN ; ERN TransplantChild.

Other stakeholders/project representatives: French DGoS ; French BNDMR, ERASMUS MC (invited speaker), DHD (invited speaker), ECHO, SIOP, Oslo University Hospital (invited speaker)

Executive summary

The second day was open to external stakeholders, it gathered 107 participants including all project partners as well as representatives from 19 ERNs and other stakeholders.
This second day wanted to expand the meeting scope to be as exhaustive as possible, by presenting and discussing the OD4RD project achievements and their impact on the different stakeholders from their own perspectives. Amongst others, this included the German case study of implementing ORPHAcodes in hospitals, ERN-CRANIO feedback on the Nomenclature Revision Collaboration with Orphanet, feedback from a Neurologist from the Oslo University Hospital having participated in a training organised by the Norwegian National Hub, as well as the Netherlands case study of ORPHAcode implementation in hospitals and data exploitation.

It also provided an occasion to present and discuss with external stakeholders the planned OD4RD2 activities aimed at scaling up those started during OD4RD, such as increasing the number of nomenclature revision collaboration projects with ERNs, further expanding the scientific annotations of RDs, increasing the network of national hubs and widening the scope of their activities; as well as continuing to supporting evidence-based decisions in the context of ERN coordination, Board of Member States (BoMS) and European Commission activities. Results of the analysis of disease coverage by ERN centres, including complementarities (overlaps) and gaps in terms of disease fields were also presented to the audience.

Two brainstorming sessions were organised after each session to allow project partners and the audience to brainstorm together on the way forward for a better identification of RD in national health systems and how to empower ERNs, hospitals and the EC to strengthen their understanding of RD-related activities.

Conclusion

The first day of this collaborative meeting ensured that project partners are on the same page and onboard regarding the planned actions. Moreover the users'feedback and exchanges with external stakeholders during the second day meeting allowed the project partners to better understand the users'needs, consider new ideas and discuss new tasks for implementation by the action. A final closing session allowed to agree on a updated and « more fitting the users'needs » action plan.

From the participants perspective, collected thanks to a feedback survey launched during the meeting (29 respondents): 93% of participants were satisfied with the meeting (strongly agree or agree) ; all the respondents knew the project « in general » before attending and only 4 did not know the specific activities carried out. Moreover, 93% of the respondents considered that the meeting was helpful in further better understanding the project objectives and plans.

 Meeting Slides can be accessed here.

Annex 1 OD4RD 2 KOM Agenda

Thursday 13 April from 13h30 to 17h30

N.B. This half day is only for OD4RD2 project partners & Orphanet Network

13h30 Welcome – *Orphanet Network Coordinator*

13h45 Introduction – *Orphanet Network Coordinator*

14h15 Orphanet nomenclature and classification of RD update and maintenance (WP2) : What has been done, What is planned & Discussion (including items to discuss the day after with the external stakeholders*) – Orphanet Director - ERN collaboration Manager*

14h45 Develop Orphanet Knowledge and information base around RD in collaboration with ERNs (WP3): What has been done, What is planned & Discussion (including items to discuss the day after with the external stakeholders)- *Orphanet director-Alignement Manager*

15h15 Support to the EC in its ERN strategy including dedicated IT systems (WP5) : What has been done, What is planned & Discussion (including items to discuss the day after with the external stakeholders) – *WP5 leaders*

15h30 Break

15h45 Consolidate and Expand national Orphanet nomenclature hubs (WP4) : What has been done, What is planned & Discussion (including items to discuss the day after with the external stakeholders) – *WP4 Leaders*

16h15 Workshop: kick off of Network of National Hubs (WP4) activities – *WP4 Leaders*

17h15 Closing : tomorrow's agenda presentation and items to be adressed *Orphanet Network Coordinator*

Friday 14 April from 10h to 17h30

N.B. This meeting is open to extrenal stakeholders from 10h to 16h30.

For Orphanet members the meeting will start at 10h and end at 17h30 (the last Planning session is only for OD4RD2 project partners)

10h Welcome – *Ana Rath, Orphanet Network and OD4RD2 Coordinator*

10h15 Introduction and brief presentation of OD4RD achievements – *Orphanet Network and OD4RD2 Coordinator*

10h45 Session 1 : OD4RD2 & ERNS *Chair: Orphanet OD4RD2 Coordinator-Orphanet National Hubs Scientific Coordinator*

- Implementation of ORPHAcodes in Hospitals: German usecase *(ERN-RND)*
- Collaboration feedback from ERNs perspective *(ERN CRANIO)*
- Orphanet database results analysis: Disease coverage by ERN centres – *(Orphanet Director,)*

11h30 Break

11h45-12h30 Session 1: Connecting Ideas : brainstorming to find consensus solutions to adress the difficulties encountered by ERNs/OD4RD and discuss new ideas for implementation by the action

12h30 Lunch Break

14h00- ORPHAcodes Implementation Support *Chairs: OD4RD2 National Hubs Coordinators*

- National Hubs activity feedback from an Hospital Clinicians perspective *(Norway)*
- ORPHAcodes Implementation at National Level Hospitals Managers perspective : the Netherlands use case *(MSc -EUHA & Erasmus MC Representative; Dutch Hospital Data)*
- Interactions with the ERNs Integration Joint Action *(JARDIN Coordinator)*
- Storyline: What are the process/ tools available *(Orphanet CTO)*

15h15 Session 2: Connecting Ideas : brainstorming to find consensus solutions to adress the difficulties encountered by Hospitals/OD4RD and discuss new ideas for implementation by the action

16h00 Closing (for external partners only)

16h15 Break

16h30-17h15 [**only for project partners / wp leaders]:** Adjustement of plans according to feedback received

17h15 Closing for internal partners