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Orphanet Nomenclature and Ontologies National Trainings

Organisation name of lead beneficiary for this deliverable:

Partner 01 – INSERM-Orphanet

Contributors: Partner N° OUS-HF- INSERM ; Ciber ; OPBG;
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1. Introduction and Background

The EJP RD “Orphanet nomenclature and ontology for Rare Disease research” training programme was developed by Orphanet in the frame of the European Joint Programme on Rare Diseases (EJP RD). One of the tasks that Orphanet has been assigned within this project consists of diffusing a training module through the Orphanet Network. It is aimed at improving knowledge and basic skills about Orphanet nomenclature (including the ORPHAcodes) and Orphanet Rare Disease Ontology - ORDO for clinicians, researchers, and for the IT teams involved in RD research activities (EJP RD - WP14 - Task 14.1).

2. Rationale for the training

2.1. Content development

A training need assessment (results available [here](#)) was performed by Orphanet in 2019, then, based on this, the module content was prepared in collaboration with the Orphanet Scientific Team and the EJP RD Task14-1Scientific Committee (Sci-Comm reports available [here](#)).

The module content was adapted following the 3 pilot National Trainings (Norway, Spain, Italy) according to feedback from real-life users of the Orphanet nomenclature and ontology for RD research to complement the work initiated in the Training Needs Assessment (TNA) phase ([EJPRD_P3_D14.1_PU_First_Report_of_Orphanet_nomenclature_training_for_trainers_and_national_trainings.pdf](#)).

The module content is then adapted for each National Training as these are organised according to the participants' needs which are collected during the registration phase or assessed with the co-organisers in each country. Thus, the module is adapted each time, in terms of length and of pre-training tasks. Since 2022, the module can be complemented with more advanced material (both in terms of slide show presentations and hands-on exercises material) developed in the frame of the OD4RD project (see point 2.3).

In terms of translations, in 2022 the training module content was translated into Turkish in addition to the Spanish and Italian translations available since 2021.

Feedback survey answers from the participants and then the experiences (both difficulties and successes) as well as the participants feedback after each training are discussed with the training's organisers and the other teams (either via dedicated EJP RD T14-1 meetings or via the OD4RD forum). This with the aim of delivering the most effective training experience to the participants by adapting and improving the module content to the National and attendees needs but also to improve the Trainers toolkit that is made available to the Orphanet National Teams.

Thanks to this forum, the hands-on exercise content has been expanded and a no-excel based set of exercises has been developed for the participants that have no knowledge on this tool, as handling excel was one of the difficulties detected during the pilot phase. Finally, material produced in the frame of OD4RD project which is

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more “advanced level” can also be exploited in order to be able to pick and choose the modules and content that better fits the attendees needs and experience.

2.2. Programmed sessions

The “Orphanet nomenclature and ontology for RD research” training programme includes 2 different types of sessions:

1. A TRAINING FOR TRAINERS (TFT) SESSION: reserved to Orphanet Network members in order to empower them to be ambassadors of the Orphanet Nomenclature and ontology in their country and to deliver trainings in their countries in local languages. This training was delivered in November 2020.
2. NATIONAL TRAINING SESSIONS: The goal was to reach 10 sessions that will be open to researchers, clinicians and IT teams involved in RD research activities in Europe and in non-EU countries (present as Orphanet Third Linked Parties in the EJP RD project). As of today, 9 Trainings have been already held as indicated in the table 1 at the end of the document.

2.3 Co-Branding OD4RD

Since 2022, Orphanet Inserm, coordinator of this task, is also the beneficiary of a Direct Grant aimed at supporting the implementation of ORPHAcodes in Hospitals hosting ERNs in several Member States (www.od4rd.eu). Within this scope, the Network is developing National Orphanet Nomenclature Hubs that act as helpdesk for Nomenclature issues at National level with an umbrella scientific coordination by the Orphanet Coordinating team. Within this project National Hubs are also encouraged to train clinicians on the use of the ORPHAcodes (more from a clinical perspective). As the boundary between care and research is not clearly determined especially in the field of RD and because often in the frame of ERNs there is an overlap between clinical and research duties (i.e. coding in hospitals and entering data in registries), it was agreed with the EJP RD coordinating team to allow organisation of co-branded trainings for coders organised in the frame of the OD4RD project. This decision also allows to mutualise resources produced in the frame of the EJP RD Task 14-1 and the ones produced in the frame of OD4RD and to avoid redundancies. This strategy has proven effective and in 2022 and 2023 more advanced and practical sessions for trainees were organised thanks to the exploitation of both the material produced in the EJP RD project and OD4RD.

2.4 Development of an online module

The Orphanet Norway Team has developed a short e-learning module available at [ORPHA-koder for sjeldne diagnoser - Sjelden](#) in Norwegian, based on the EJP RD Module content and with interactive questions that allow the users to connect and test and improve their knowledge on ORPHAcoding whenever they want.

The module is being translated into English in order to make it available and exploitable by a larger resource and its release is planned by the end of 2023.

Indeed, this format could ensure the availability of the EJP RD module training online also beyond the length of the project, it could also obviously ensure reaching a higher number of trainees (as currently the National trainings are limited to 20-25 participants and clinicians have very busy schedules), and from all over the world not only limited to Orphanet Network countries. In addition to the wider audience, it could also facilitate the National trainings organisation itself and improve the number of clinician trainees reached. Indeed, the participants' feedback collected so far shows that, despite appreciating the content of the module, one of the trainees' constraints is often related to time, and the length of the full programme has often been mentioned as a limiting factor in their (or their colleagues) participation. Having the first part of module 1 as an online module could allow participants to follow it on their own schedule and then attend a shorter National training session more focused on the research related available tools and the hands-on session.

3. National Training Objectives

3.1 General objective

The General Objective of the National Trainings is to enable participants to improve knowledge on Orphanet nomenclature and Ontologies for RD research activities.

3.2 Specific Objectives

At the end of the training participants should be able to:

- 1- Explain what are the Orphanet nomenclature and the Rare Disease Ontology (ORDO)
- 2- Access Orphanet data that is relevant to their rare disease research activities
- 3- Contribute to Orphanet data by using dedicated forms and helpdesks

4. National Trainings held 2022-2023

4.1 Spain National Trainings

The Orphanet National team organized **two training sessions** (co-branding with OD4RD) together with the Catalonian Health Department, who oversaw dissemination, registration and follow up. The trainings consisted in two sessions to be followed in autonomy by the participants, followed by an online hands-on session which was held by the Orphanet Spanish Team. (November 25th where 12 participants attended & December 2nd where 15 participants attended). As far as the satisfaction rate of the trainings, 82% of participants manifested they were very satisfied with the course as far as quality/quantity/level of contents, organizational aspects, format and trainer's skills.

Detailed Programme of the training:

[November 8th – November 22nd: Estimated duration 3.5 h]

SESSION 1: ORPHANET NOMENCLATURE PRESENTATION

Objectives

ENHANCE knowledge on Orphanet Nomenclature

Description

1. Presentation of the webinar on Orphanet Nomenclature.
2. The first session of this online course will be followed by participants in an individual basis between November 8th and November 22th. Participants are expected to view all the presentations and complete a pre-training activity before attending Session 3 scheduled on November 25th or December 2nd 2022.

Contents

Presentation: ORPHANET NOMENCLATURE Chapter 1
Presentation: ORPHANET NOMENCLATURE Chapter 2
Presentation: ORPHANET NOMENCLATURE Chapter 3

Additional suggested videos:

- Orphanet presentation Short video
- Why ORPHAcoding? Slides and short video
- Orphanet tutorial

SESSION 2: ORPHANET DATA AND HOW TO ACCESS IT PRESENTATION

Objectives

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ENHANCE knowledge on Orphanet data available and tools including the Orphanet RD Ontology (ORDO) and the ORDO-HPO ontological module (HOOM).

Description

1. Presentation of the webinar on Orphanet data available for RD research and how to access it. It includes some slots to address any questions and doubts.
2. The second session of this online course will be followed by participants in an individual basis between November 8th and November 22nd 2022. Participants are expected to view all the presentations before attending Session 3 scheduled on November 25th or December 2nd 2022.

[November 25th / December 2nd, 2022 (morning)]:

SESSION 3: HANDS-ON

Objectives

ENHANCE knowledge on Orphanet Nomenclature and Ontologies

Description

Practical exercises

Program of the session

Time: 10:00- 14:00
Welcome & introduction
Presentations round
Presentation 1: Sessions 1 and 2 summary
Presentation 2: ORPHAcoding
Questions & Answers-1
Break
Practice time: HANDS-ON
Questions & Answers-2
Presentation of “Examples of Orphanet data exploitation” and Wrap-up

4.2 Italy National Training

The Orphanet National team organized the second edition of the Italian EJPRD Online Training Course on Orphanet Nomenclature and RD Ontologies on April 14 2022. This session was organised in order to include the over-booked participants from the training held in September 2021 (online courses are limited to 20 participants in order to ensure an optimal experience and knowledge gain). The training, aimed at improving knowledge and basic skills about Orphanet nomenclature (including the ORPHAcodes) and Orphanet Rare Disease Ontology (ORDO) was attended by 17 participants among clinicians and researchers. The Training consisted of two sessions: the first session was autonomously followed by the selected participants, provided with learning material; the second one was delivered as an online conference call on April 14 and was attended by the above mentioned 17 attendees.

The satisfaction rate of the training was of: 90%

Detailed Programme of the training is available here:

First session ("primo modulo" in italian)

Primo modulo: introduzione alla nomenclatura di Orphanet (entro il 14 aprile 2022)
<i>Webinar Chapter 1: ORPHANET NOMENCLATURE OF RARE DISEASES</i>
<i>Webinar Chapter 2: ORPHANET CLASSIFICATION</i>
<i>Webinar Chapter 3: ORPHACODES</i>
Hands on session

Second session ("secondo modulo" in italian)

Secondo modulo: classe online con il team di Orphanet (14 aprile 2022)		
Ora	Attività	Relatore
10:30- 10:50	Warm up-Working groups	S. Ciampa e M. Nutini
10:50- 11:10	Reminders first module	S. Ciampa e M. Nutini
11:10- 11:20	Orphanet Data Available, Tools and Ontologies	Video
11:20- 11:40	Q&A	S. Ciampa e M. Nutini

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11:40-11:55	Pausa	
12:00-12:45	Demo tools: API, ORDO - Q&A	Orphanet Management Team
12:45	Pausa pranzo	
14:15-14:30	Warm up - Summary	S. Ciampa e M. Nutini
14:30-15:15	Hands on session	S. Ciampa e M. Nutini
15:15-15:30	Pausa	
15:30-16:15	Hands on session	S. Ciampa e M. Nutini
16:15-16:45	Q&A & summary	S. Ciampa e M. Nutini

Final remarks: A survey was regularly delivered to the participants after every training. We calculated the satisfaction ratings on the basis of specific questions of the survey, focused on the following topics: 1. relevance of the contents; 2. educational quality; 3. usefulness of the training for the professional needs of the participant. Comparing the average rating of the 2022 training respect to the first edition, which was held on September 2021, we increased the satisfaction rating by 8 percentage points (90% in April 2022 vs. 82% in September 2021). We believe that this improvement is the result of the lessons learned from our first training experience. For example, we implemented strategies aimed at capturing the attendees' attention (i.e. by creating separate working groups during the exercise session).

4.3 France National Trainings

4.3.1 2022 Training

On the 14 and 24 November 2022, the Orphanet National team delivered a two-days e-training co-organised with the French National RD Registry- BNDMR (Banque Nationale de Données Maladies Rares) (this training was co-branded with OD4RD) . The training, aimed at improving knowledge and basic skills about Orphanet nomenclature (including the ORPHAcodes) was attended by 120 participants among clinicians and researchers over the 2 sessions. An article regarding this training session was published in the French Orphanews: https://france.orphanews.org/newsletter-fr/editorial/nl/fr20221122.html#or_nouveautes-orphanet-45197

The satisfaction rate of the training was of: 95%

Detailed Programme of the training is available here:

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BASIC session:

1. The Orphanet Nomenclature of Rare Diseases
 - Introduction to the Orphanet Nomenclature,
 - Where to find ORPHAcodes & Nomenclature data
2. The Orphanet Classification of Rare Diseases
 - Introduction to the Orphanet classification,
 - How is it organized?
 - How to navigate the Orphanet classification?
3. How is the nomenclature updated?
 - Creation, Modification and Inactivation of an ORPHAcode
 - Examples and special cases

ADVANCED session:

1. ORPHAcodes
 - General reminders
 - Genes linked to ORPHAcodes & Other Associated resources
2. A dedicated process for FSMRs
 - Fiche CMI Creation – Modification – Inactivation
 - Trello
 - BNDMR: the French National Registry for RD, a resource for research
3. Orphanet ERN collaboration
 - Methodology
 - State of play
4. Coding with ORPHAcodes
 - ORPHAcodes alignments with other terminologies
 - Coding recommendations
 - How to use the right ORPHAcode? Use cases

4.3.2 2023 Training

On the 16 and 20 November 2023, the Orphanet National team delivered a two-days e-training co-organised with the French National RD Registry- BNDMR (Banque Nationale de Données Maladies Rares) (this training was co-branded with OD4RD) . The training, aimed at improving knowledge and basic skills about Orphanet nomenclature (including the ORPHAcodes) was attended by 58 participants among clinicians and researchers over the 2 sessions. An article regarding this training session will be published in the French Orphanews in December 2023.

The satisfaction rate of the training will be available in December 2023, as the survey is still running at the time of this report production.

Detailed Programme of the training is available here:

MODULE 1 :

1. The Orphanet Nomenclature of Rare Diseases

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- Introduction to the Orphanet Nomenclature,

19.01.2023 Perşembe	10.00 Açılış
	10.15 İlanma Sınavı
	10.50 Orphanet Tanıtım
	11.00 Ara
	11.20 Orphanet Terminolojisi
	12.00 Soru&Cevap
	12.15 Öğle Yemeği
	13.00 Ar&Ge'deki Zorluklar EJPRD Videosu
	13.15 Geçmişten Günümüze Orphanet&Türkiye
	14.15 Orphanet Verilerine Nasıl Erişilir?
	14.40 Ara
14.55 Orphanet'in Oluşumu&Türkiye Terminolojisi	
15.50 Soru&Cevap	
20.01.2023 Cuma	09.30 İlk Günün Gözden Geçirilmesi
	10.00 Orphanet Hekim Deneyimi
	11.00 Ara
	11.20 Uygulamalı Deneyim
	12.30 Soru&Cevap

- Where to find ORPHAcodes & Nomenclature data?

2. The Orphanet Classification of Rare Diseases

- Introduction to the Orphanet classification,

- How is it organized?

- How to navigate the Orphanet classification?

3. How is the nomenclature updated?

- Creation, Modification and Inactivation of an ORPHAcodes

- Examples and special cases

- A dedicated process for FSMRs

MODULE 2 :

1. ORPHAcodes: General reminders

- Including: Coding undiagnosed patients; Inactivated ORPHAcodes in BaMaRa

2. Evolution of Orphanet Nomenclature: A dedicated process for FSMR

3. Orphanet ERN collaboration

- State of play: Ongoing & planned collaboration

4. Resources associated to ORPHAcodes

- Genes linked to ORPHAcodes & Genetic characterization in BaMaRa
- Other resources associated to ORPHAcodes
- ORPHAcodes alignments

5. New tools and knowledge databases

- Orphanet terminologies mapping services
- Clinical signs and symptoms
- New knowledge databases
- New BaMaRa tools

4.4 Turkey National Training

An Orphanet Nomenclature Training Session was held between 19-20 January 2023 as the training activities of the ISTisNA project in cooperation with Orphanet and its project partner Acıbadem University within the EJP RD scope.

The training, aimed at improving knowledge and basic skills about Orphanet nomenclature (including the ORPHAcodes) and Orphanet Rare Disease Ontology (ORDO) was attended by over 50 participants.

During the two-day training, all presentations were made interactively. As a result of the feedback received from the participants, the satisfaction rate from the training was determined as 92%.

Detailed Programme of the training is available here:

Agenda 19/01

- Opening
- Ice breaking
- Introduction to Orphanet

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- Orphanet Nomenclature
- Q/A session
- Lunch break
- [EJP RD Video on Challenges in RD resources](#)
- Orphanet Turkey history
- How to access Orphanet data
- Orphanet & Nomenclature in Turkish
- Q/A session

Agenda 20/01:

- Warming: recap day 1
- Orphanet clinicians experience
- Use cases
- Q/A session

4.5 Norway National Training

A training, aimed at improving knowledge and basic skills about Orphanet nomenclature (including the ORPHAcodes) and Orphanet Rare Disease Ontology (ORDO) was planned in Norway in Q4 2023, unfortunately because of an extended sick leave of the training manager it was cancelled. Still the availability of the online module (see section 2.4) ensures that Orphanet Nomenclature knowledge is accessible online.

5. Conclusion

Since the beginning of the project 9 National Trainings have already been held (table1), One training planned in Q4 of 2023 in Norway was unfortunately cancelled due to extended sick leave of the training manager. These represent 361 participants trained overall in 5 different countries (table 2). The impact of the training is higher as these numbers only include the participants to online sessions of National trainings and do not include the number of persons having taken the online module already available in Norwegian (see section 2.4).

Overall, we have a satisfaction rate of 85% for the 8 Trainings for which Satisfaction surveys results are already available (2023 National Training in France was held on the 16-20 November and the survey is still running at the time of this report preparation). And importantly for both countries (Italy and Spain) where additional sessions have been held in 2022 the satisfaction rate has increased compared to the Pilot sessions held in 2021.

	France	Italy	Norway	Spain	Turkey	Tft	Total
EJP RD		2	1	1	1	1	6

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EJP RD-OD4RD	2		1 (cancelled)	2			5
Total	2	2	1	3	1	1	10

Table 1. Number of trainings by country and according to Branding.

National Training	Number of participants	Satisfaction feedback regarding overall module content (% of respondents)
2021 Norway	35	77%
2021 Spain	37	80%
2021 Italy	17	82%
2022 Italy	17	90%
2022 Spain 1	12	82%
2022 Spain 2	15	82%
2022 France	120	95%
2023 Turkey	50	92%
2023 France	58	Satisfaction survey still open at the time of this report production
Total	361	

Table 2. Number of participants and overall satisfaction by Training.