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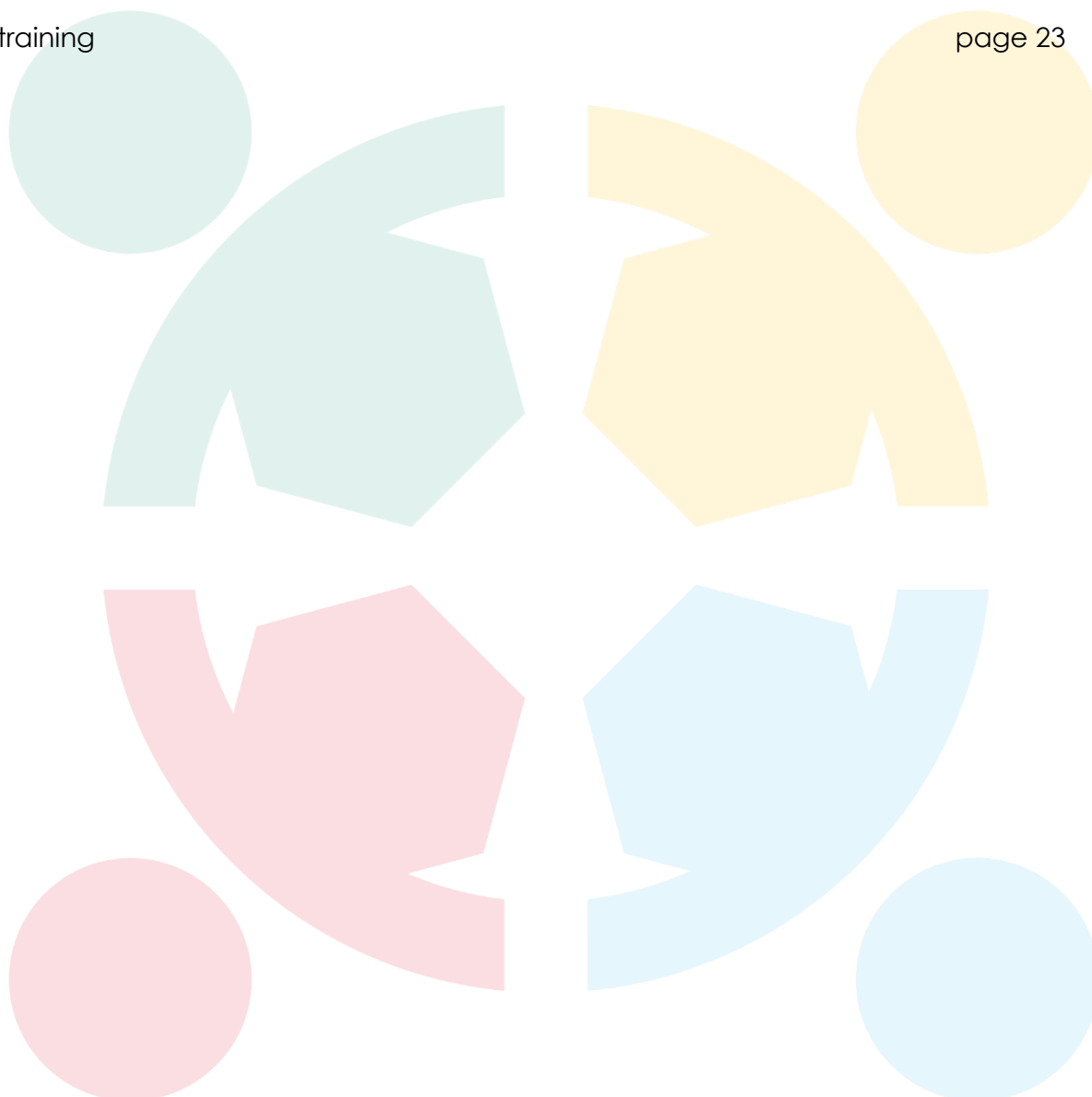
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Executive Summary

The “International Summer School on Rare Disease Registries and FAIRification¹ of data” has been organised in each year of activity of the European Joint Programme on Rare Diseases (EJP RD), hence in five editions.

The trainings addressed the need to decrease RD data fragmentation and increase data quality in order to raise the level of capacities and help data sharing and networking within the RD community.

The deployment of this specific training foresaw the integration and implementation of an existing and successful training organised by the National Centre for RD of Istituto Superiore di Sanità (ISS) on RD and orphan drugs registries, as well as of existing workshops organised by the same Institution on the FAIRification of RD registries.

Over the five editions of the trainings a high number of RD specialists, such as researchers, database managers, clinicians and medical specialists, European Reference Network (ERN) representatives, in addition to patient representatives, has been trained.

Speakers and participants taking part in the training came from many different countries. The participation to the free course of patient representatives and attendants living and working in a EU-13 Country/Turkey has been eased by the awarding of travel and accommodation fellowships dedicated to these specific targets.

The training foresaw two different training modules “RD Registries” and FAIRification of data”, and the possibility for the participants to register for only one or both training modules.

Given the outbreak of the Covid-19 pandemic there has been the need to adapt three of the five editions from a residential to an online training, namely editions 2, 3 and 4. During these editions no fellowships have been awarded.

The training kept in all editions its highly interactive character and the objective to promote the establishment of networks between different RD stakeholders involved in RD registries, even in the online adaptation.

The quality of the course has been highly appreciated by the participants, as shown by the feedback collected through the satisfaction surveys.

¹ FAIR: Findable, Accessible, Interoperable, Reusable (for humans and computers)

Introduction and Objective

This document is a report on the training activities on RD Registries and FAIRification of data at the source that represent a specific goal of Work Package (WP) 14, “Training on data management and quality”, led by ISS, and falling under the action of Pillar (P) 3 “Capacity Building & Empowerment” of the European Joint Programme on Rare Diseases (EJP RD).

The organisation of the course titled “International Summer School on RD Registries and FAIRification of data” has been carried out yearly by ISS for the whole duration of the project, in close collaboration with Task (T) 14.5² Partners. Task 14.5 Partners are LUMC³ (also for Endo-ERN); Bond-ERN (IOR)⁴, Metab-ERN (ASUIUD)⁵, EURORDIS⁶, ISCIII⁷, INSERM⁸ (RaDiCo); UMCG⁹, DTL-Projects¹⁰ (ELIXIR-NL), CNR¹¹ (ELIXIR -IT), AMC¹², ELIXIR/EMBL-EBI (UNIMAN)]¹³. The premises standing behind the organisation of the EJP RD training courses of WP 14, and of T14.5 in particular, as well as the topics covered during the course, have been extensively described in Deliverable (D) 14.9 “First Report on International course on RD Registries and FAIRification of data”, available at the following [link](#) and will not be further described in the present document.

Methodology

The results of the five editions of the “International Summer School on Rare Disease Registries and FAIRification of data” organised by ISS in collaboration with Task 14.5 Partners as residential and online trainings have been reported and broad conclusions have been drawn.

More in detail the following aspects have been analysed for each edition of the training:

- the number of registrations
- the number of selected attendants

² Task 14.5 “Training on rare disease registries and FAIRification of data at the source”

³ LUMC Leiden University Medical Centre

⁴ IOR Istituto Ortopedico Rizzoli

⁵ Metab-ERN (ASUIUD) European Reference Network for Hereditary Metabolic Disorders, University Hospital of Udine

⁶ EURORDIS European Organisation for Rare Diseases Association

⁷ ISCIII Instituto de Salud Carlos III

⁸ INSERM Institut National de la Santé et de la Recherche Medicale

⁹ UMCG Academisch Ziekenhuis Groningen

¹⁰ DTL Dutch Techcentre for Life Sciences

¹¹ CNR Consiglio Nazionale delle Ricerche

¹² AMC Academisch Medisch Centrum bij de Universiteit van Amsterdam

¹³ University of Manchester

- the effective number of participants
- the EJP RD countries represented by the effective participants
- the number of awarded fellowships
- the number of participants belonging to an ERN and the ERNs represented in the training
- the number of speakers and their affiliation, as well as their country of employment/residence
- the number of training hours
- the percentage results of the scoring from the satisfaction surveys for each training module and some highlights from the free comments

Results

Summary of the first edition of the International Summer School on RD Registries and FAIRification of data

The residential training course took place at ISS from 23 to 27 September 2019 and foresaw the participation of 30 attendees for each of the two training modules composing the course: "RD Registries" and "FAIRification of data".

72 applicants filled in the registration form on the LisyLime Platform of INSERM correctly and completely. Of these, 58 applied for both training modules, 8 for the first and 7 for the second training module only.

3 fellowships were foreseen for participants living and working in a EU-13 Country a 3 for patient representatives.

Among the 28 applicants that required to receive a travel and accommodation fellowship, 4 lived and worked in a EU-13 Country (2 applicants from Cyprus, 1 from Lithuania and 1 from Slovakia; one of the applicants from Cyprus applied for the first module only), 14 were patient representatives and 10 applied without having the requisites.

Given the high number of applications, it has been decided to allow the admission of more than 30 participants per module.

Overall, 38 participants have been admitted taking part in the course: 30 for both training modules, 3 for the first training module only and 5 for the second training module only.

5 fellowships have been assigned.

The fellowships for the participants living and working in a EU-13 Country have been awarded to the applicants of Cyprus and Lithuania. The third fellowship has not been assigned as the remaining two eligible applicants declined. The 3 fellowships reserved to patient representatives were assigned following a scoring matrix defined by EURORDIS. The 3 awarded participants came from the following affiliations:

- Institute of Neurology, Psychiatry and Narcology of the National Academy of Medical Sciences of Ukraine
- Pro Retina Germany/Retina International Dublin

- Osteogenesis Imperfecta Federation Europe (OIFE).
- Two of these belonged also to an ERN (ERN EYE and ERN BOND).

A total of 34 attendees participated in the training, although 23 attended the entire module(s) to which they had registered: 17 followed both modules, 3 the first module only and 3 the second module only.

20 of the selected participants belonged to an ERN, representing 50% of the ERNs (12/24 ERNs), namely:

- VASCERN (3 participants)
- ERN EPICARE (2 participants)
- ERN ReCONNET (2 participants)
- ERN Euro-NMD (2 participants)
- ERN TRANSPLAN-CHILD (2 participants)
- ERN ITHACA (2 participants)
- ERN EYE (2 participants)
- ERN Eurogen
- ERN EURACAN
- ERN Skin
- ERN BOND
- ERN Euro Blood Net

4 participants were patient representatives.

The selected participants were from 10 different EJP RD/EU-13 countries: Cyprus, France, Germany, Italy, Lithuania, Norway, Spain, The Netherlands, Turkey, and UK.

The 22 Speakers of the course belonged to the following Institutions from 8 different countries (Bulgaria, France, Germany, Italy, Spain, The Netherlands, UK, USA):

- University of Glasgow, UK (EndoERN)
- Università degli Studi di Padova, Italy (ERN PaedCan)
- EURORDIS France
- National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Global Medical Operations & Patient Registries Amicus Therapeutics, USA
- University Hospital Heidelberg (MetabERN), Germany
- Orphanet, INSERM, France
- Leiden University Medical Centre, The Netherlands
- Fondazione per la Ricerca Farmacologica Gianni Benzi, Italy
- European Commission's Joint Research Centre, Ispra, Italy
- Institute of Health Carlos III, Madrid, Spain

- BioSemantics group, Leiden University Medical Centre, The Netherlands
- Medical University of Plovdiv, Bulgaria
- University of Bolzano, Italy
- University Medical Centre Groningen, The Netherlands
- Institute of Molecular Biology and Pathology, National Research Council, Italy
- Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP), Spain
- Catholic University “Sacro Cuore” Rome, Former Coordinator AIFA registries, Italy

21 participants followed a non- mandatory preparatory webinar that was delivered at the beginning of September 2019, dedicated to participants attending the second training module.

For the first training module on RD Registries the Problem-Based Learning (PBL) methodology was adopted¹⁴.

In the second training module on FAIRification of data, plenary and break-out sessions, active exercises, work on tutorials, on sample datasets and lightweight tools were used to carry out the hands-on “Bring Your Own Data” (BYOD) experiences. Participants had the possibility to work on their own mock datasets. This training module too foresaw group work to stimulate networking. At the end of each of the two training modules the participants presented the group solutions to the experts and received feedback on their work.

The duration of the entire training course was 40 hours, divided in morning and afternoon session in all five training days. The first training module foresaw 24 training hours and the second module 16 training hours.

The satisfaction level of the participants has been collected through a satisfaction questionnaire composed of nine multiple choice items and two open questions.

19 participants of the first training module and 22 participants of the second training module agreed to fill in the satisfaction questionnaire.

The analysis of the satisfaction questionnaires showed a general positive evaluation of the entire training course. For both modules 100% of the participants answered “agree/strongly agree” to the item “The course increased my knowledge”.

¹⁴ The PBL is a student-centred approach in which students learn by working in groups to solve an open-ended problem. This method uses complex real-world problems and provides opportunities for working in groups and for finding and evaluating research materials.

Below the percentage of participants answering “agree/strongly agree” to the other items of the questionnaire for module 1:

- 74% to item “The contents were appropriate for my level of knowledge”
- 90% to item “The objectives were clearly defined”
- 84% to item “The teaching method was effective”
- 95% to item “Time dedicated to individual/group exercises is sufficient”
- 63% to item “The learning materials were sufficient and of good quality”
- 100% to the item “The number of speakers was sufficient”
- 95% to item “The speakers were competent and prepared”
- 90% to item “The course is well organized”.

Percentage of participants answering “agree/strongly agree” to the other items for module 2:

- 68% to item “The contents were appropriate for my level of knowledge”
- 82% to item “The objectives were clearly defined”
- 77% to item “The teaching method was effective”
- 90% to item “Time dedicated to individual/group exercises is sufficient”
- 68% to item “The learning materials were sufficient and of good quality”
- 90% “The number of speakers was sufficient”
- 100% “The speakers were competent and prepared”
- 87% “The course is well organized”.

Among the free comments on positive aspects the participants highlighted the interactivity and high quality of the training, and the large range of covered topics. Among the suggestions an introduction of the basics standing behind the presented topics, more breaks and the sharing of training material have been reported.

Summary of the second edition of the International Summer School on RD Registries and FAIRification of data

The second edition of this residential training course had to be adapted to an online version, given the outbreak of the Covid-19 pandemic and the relative restriction measures adopted to limit the outspread of the virus.

The course has been delivered on 28 September-2 October 2020, on the Microsoft Teams Platform (MS Teams Platform) of the EJP RD.

The registration was carried out using the LisyLime Platform of INSERM, and was possible for the first training module (RD Registries, 28-30 September), for the second training module (FAIRification of data, 1-2 October) or for the entire training course. The registration form has been enhanced, in order to avoid the high number of uncompleted registrations that have been observed in the previous edition of the training.

132 completed registrations have been submitted. 131 respondents applied for both training modules, 15 for the first training module only and six for the second training module only. 37 participants have been admitted to attend the online course: 31 were admitted to attend both training modules, 5 the first training module only and 1 the second training module only.

The admission of more than 30 participants per module has been motivated by the high number of registrations.

28 of the 37 selected participants joined all or most of the training hours of the module to which they were registered. 22 followed both training modules, 5 the first training module only and 1 the second training module only.

The participants lived and worked in the following 16 EJP RD/EU-13 countries: Belgium, Canada, Czech Republic, France, Germany, Hungary, Ireland, Israel, Italy, Lithuania, Norway, Portugal, Spain, The Netherlands, Turkey, UK.

24 participants belonged to an ERN. The represented 16 ERNs (67% of the ERNs) in the training were:

- ERN BOND (3 participants)
- ERN Cranio (2 participants)
- ERN EURO NMD (2 participants)
- ERN EYE (2 participants)
- ERN RARE-LIVER (2 participants)
- ERN ReCONNET (2 participants)
- ERN Skin (2 participants)
- ERN RND
- MetabERN
- ERN RITA
- VASCERN
- ERN GENTURIS
- ERN EUROGEN
- ERK-Net
- ERN EURACAN
- ERN EuroBloodNet

3 participants were patient representatives.

The 19 Speakers belonged to the following Institutions, spread in 7 Countries:

- University of Glasgow, (EndoERN), UK
- National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Global Medical Operations & Patient Registries Amicus Therapeutics, USA
- EURORDIS, France
- Orphanet, INSERM, France
- Leiden University Medical Centre, The Netherlands
- Academic Medical Center, The Netherlands
- Leiden University Medical Centre, GO FAIR, The Netherlands
- European Commission's Joint Research Centre, Ispra, Italy
- Institute of Health Carlos III, Madrid, Spain
- University Hospital Frankfur, Germany
- University of Bolzano, Italy
- University Medical Centre Groningen, The Netherlands
- Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP), Spain

In the first training module the PBL methodology has been substituted by plenary presentations and interactive question & answers sessions between speakers and participants.

In the second training module, participants deepened their knowledge on the single steps of the FAIRification of data and discovered the potential of FAIR registries. For each step of the FAIRification process, there was a plenary session followed by hands-on exercises and a wrap up session. Also, the FAIR data management and FAIR project planning were addressed in this training module.

The duration of the online course has been of 24 hours: 9,5 hours for the first training module that foresaw afternoon session only, and 14,5 hours for the second training module that was composed on both morning and afternoon sessions.

The adaptation of the number of hours, when compared to the residential edition of the training course, has been motivated by the online delivery of the course, that needed to take into account the time in which the participants could keep their attention and active participation.

The analysis of the satisfaction questionnaires, that were completed and returned by 23 participants for the first training module and 18 for the second training module evidence a good satisfaction rate. For both modules a high

percentage of participants answered “agree/strongly agree” to the item “The course increased my knowledge” (80% for the first training module, 89% for the second training module).

For module 1, the percentage of participants answering “agree/strongly agree” to the other items of the questionnaire have been:

- 74% to item “The contents were appropriate for my level of knowledge”
- 78% to item “The objectives were clearly defined”
- 66% to item “The teaching method was effective”
- 52% to item “Time dedicated to individual/group exercises is sufficient”
- 60% to item “The learning materials were sufficient and of good quality”
- 79% to the item “The number of speakers was sufficient”
- 96% to item “The speakers were competent and prepared”
- 87% to item “The course is well organized”.

Percentage of participants answering “agree/strongly agree” to the other items for module 2:

- 50% to item “The contents were appropriate for my level of knowledge”
- 61% to item “The objectives were clearly defined”
- 72% to item “The teaching method was effective”
- 56% to item “Time dedicated to individual/group exercises is sufficient”
- 56% to item “The learning materials were sufficient and of good quality”
- 99% “The number of speakers was sufficient”
- 94% to item “The speakers were competent and prepared”
- 89% to item “The course is well organized”.

The free comments underlined the quality of the speakers and the good, organisation of the course, as well as the interactivity. The points of improvement focused mainly on shortening the sessions, especially for an online training.

Summary of the third edition of the International Summer School on RD Registries and FAIRification of data

Given the persistence of the Covid 19 pandemic, also the third edition had to be adapted to an online version. The course took place from 27 September to 1 October 2021 on the MS Teams Platform of the EJP RD.

The applications have been gathered through the MS Forms of the EJP RD, where. 123 applicants registered for both training modules, 19 registered to be admitted to the first training module only and 4 for the second training module

only. Given the high number of requests and the online delivery of the training course, 39 applicants have been selected for both training modules and 2 for the second training module only. No candidates have been selected to participate in the first module only.

Altogether 34 participants connected to the online course, two of which participated in the second training module only.

Taking stock of the lessons learned in the first online edition, it has been decided to improve the interactivity of the training and the work in small groups.

More in detail, greater attention has been dedicated to questions and answers in the first training module, while for the second training module virtual breakout rooms have been carried out, where participants had the possibility to experience in small groups the importance of the FAIRification of data, with the assistance of IT Trainers.

At the end of the first training module an online “social event” has been proposed, where the participants had the possibility of a free networking, to better know each other, exchange experiences and create professional links.

The duration of the course has been of 24 hours: 10,5 hours for the first and 13,5 hours for the second training module. For this training course too, the first training module foresaw only afternoon sessions, while the second module has been composed of morning and afternoon sessions.

The participants lived and worked in the following 17 EJP RD/EU-13 Countries: Austria, Belgium, Czech Republic, Estonia, France, Germany, Greece, Hungary, Italy, Latvia, Portugal, Slovenia, Spain, Switzerland, The Netherlands, Turkey, UK. As for the 2020 edition no fellowships could be awarded. Nevertheless, participants from Turkey and from 6 EU-13 Countries (Bulgaria, Czech Republic, Estonia, Hungary, Latvia, Slovenia), took part in the course. No fellowships were awarded for the online course.

In this edition 22 participants belonged to and ERN, and 9 ERNs (58% of the ERNs) have been represented:

- ERN-RND (3 participants)
- ERN-NMD (3 participants)
- ERN LUNG (2 participants)
- MetabERN (2 participants)
- Endo-ERN (2 participants)
- ERN eUROGEN (2 participants)
- ERN EpiCare
- ERN EYE
- ERN RITA
- ERN BOND

- ERN RARE-LIVER
- ERN GENTURIS
- ERN ITHACA
- ERN EURACAN

4 participants were patient representatives.

21 Speakers have been involved in the course, and came from the following institutions, of 7 different countries:

- University of Glasgow, (EndoERN), UK
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Endo ERN; The Netherlands
- Global Medical Operations & Patient Registries Amicus Therapeutics, USA
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- EURORDIS, France
- Directeur technique Orphanet, INSERM, France
- Leiden University Medical Centre, The Netherlands
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Ministry of Health Directorate-General for Health Planning Government Organisation, Italy
- Azienda Ospedaliero Universitaria Pisana, Italy
- Leiden University Medical Centre, GO FAIR, The Netherlands
- European Commission's Joint Research Centre, Italy
- Leiden University Medical Centre, The Netherlands
- GO FAIR, The Netherlands
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- University of Bolzano, Italy
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- University Medical Centre Groningen, The Netherlands
- Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP), Spain

The course received the endorsement of the International Conference for Rare Diseases and Orphan Drugs (ICORD).

With regard to the satisfaction questionnaires, 23 participants completed the questionnaire for the first training module and 19 for the second training module. 91% of the respondents answered "agree/strongly agree" to the item "the course increased my knowledge" for the first training module, and 95% for the second training module.

The percentages of participants answering “agree/strongly agree” to the other items for the first module have been:

- 48% to item “The contents were appropriate for my level of knowledge”
- 63% to item “The objectives were clearly defined”
- 63% to item “The teaching method was effective”
- 100% to item “Time dedicated to individual/group exercises is sufficient”
- 84% to item “The learning materials were sufficient and of good quality”
- 100% “The number of speakers was sufficient”
- 95% to item “The speakers were competent and prepared”
- 95% to item “The course is well organized”.

An additional question has been added to the satisfaction questionnaire for the online trainings, regarding the easiness of the connection process.

To the question “The connection to the event was easy”, 83% answered “agree/strongly agree” for the first training module, and 90% for the second training module.

The free comments highlighted the appreciation of the involvement of patients, the relevance of the topics for establishing a new registry, the interactivity of the sessions, although online and the networking opportunities. Among the points for enhancement, the proposal to end the course with a final summarising wrap-up session, the suggestion to stress more practical examples from the medical field and the sharing of the presentations before the start of the training course have arisen.

Summary of the fourth edition of the International Summer School on RD Registries and FAIRification of data

After consultation of Task Partners and considering the persistence of the uncertainty linked to the Covid-19 pandemic and of the international situation it has been decided to keep the online format of the training for the 2022 edition.

The training has hence been held on the MS Teams Platform of EJP RD on 26-30 September 2022.

76 applications have been collected through the MS Forms of the EJP RD, of which 65 were for attending both training modules, 8 for the first training module only and 3 for the second training module only.

41 applicants have been selected to attend the training, 39 for attending both training modules, one for the first training module only and one for the second training module only.

25 attendees connected to the training, 7 of which discontinuously. All 25 had been selected to attend both training modules.

The training envisaged a duration of 22 hours, 9 hours for the first and 13 hours for the second training module.

In this edition of the training too, an online social event took place at the end of day 3, given the success of this initiative in the previous edition of the course.

Again, special attention has been posed on the interactivity and hands-on parts of the course, to overcome the limits posed by the online delivery of the course.

ICORD endorsed the 2022 edition of the course, too.

Participants lived and/or worked in the following 10 EJP RD/EU-13 Countries: Austria, Denmark (country of residence, Belgium country of employment), Georgia, Germany, Norway, Portugal, Switzerland, The Netherlands, Turkey, UK.

No fellowships were awarded for the online course.

8 participants belonged to an ERN, representing 6 ERNs (25% of the ERNs):

- ERN RARE-LIVER (2 participants)
- EURO-NMD
- ERN ReCONNET
- ERN GUARD-HEARTH
- ERN EYE
- ERN ITHACA

4 of the participants were patient representatives.

The Speaker were from the following institutions, of 7 different countries:

- University of Glasgow, (EndoERN), UK
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Endo ERN; Dutch Pituitary Foundation, The Netherlands
- Global Medical Operations & Patient Registries Amicus Therapeutics, USA
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- EURORDIS, France

- Orphanet, INSERM, France
- Leiden University Medical Centre, The Netherlands
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Ministry of Health Directorate-General for Health Planning Government Organisation, Italy
- Azienda Ospedaliero Universitaria Pisana, Italy
- Leiden University Medical Centre, GO FAIR, The Netherlands
- European Commission's Joint Research Centre, Italy
- Leiden University Medical Centre, The Netherlands
- GO FAIR, The Netherlands
- University of Bolzano, Italy
- University Medical Centre Groningen, The Netherlands
- Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP), Spain

The satisfaction surveys have been filled by 22 participants for the first training module, and by 18 participants for the second training module.

Looking at the results of the satisfaction questionnaire 95% of the respondents answered “agree/strongly agree” to the item “the course increased my knowledge” for the first training module, and 94% for the second training module.

The percentages of participants answering “agree/strongly agree” to the other items for the first module have been:

- 80% to item “The contents were appropriate for my level of knowledge”
- 72% to item “The objectives were clearly defined”
- 72% to item “The teaching method was effective”
- 92% to item “Time dedicated to individual/group exercises is sufficient”
- 80% to item “The learning materials were sufficient and of good quality”
- 96% “The number of speakers was sufficient”
- 74% to item “The speakers were competent and prepared”
- 95% to item “The course is well organized”
- 95% to the item “The connection to the event was easy”.

The percentages of participants answering “agree/strongly agree” to the other items for the second module have been:

- 56% to item “The contents were appropriate for my level of knowledge”
- 75% to item “The objectives were clearly defined”
- 83% to item “The teaching method was effective”
- 87% to item “Time dedicated to individual/group exercises is sufficient”
- 75% to item “The learning materials were sufficient and of good quality”
- 88% “The number of speakers was sufficient”
- 88% to item “The speakers were competent and prepared”

- 100% to item “The course is well organized”
- 100% to the item “The connection to the event was easy”.

The positive aspects from the free comments stressed the presence of a good mix of stakeholders at the course, the broad overview given by the speakers and by the interactions between participants with different background and the highlights provided on the importance of the FAIRification process. Among the points of enhancements, the participants proposed to foresee more time for the practical sessions.

Summary of the fifth edition of the International Summer School on RD Registries and FAIRification of data

The last edition of the International Summer School on RD Registries and FAIRification of data, has been held as residential training course at ISS from 25 to 29 September 2023.

As for all the editions of this training course, 30 attendees for each training modules were foreseen. 68 completed applications have been submitted via the MS Teams Platform of the EJP RD, 66 of which for attending both training modules, 5 for the first training module only and 7 for the second training module only.

3 fellowships were reserved for participants living and working in a EU-13 Country or in Turkey, and 3 to patients' representatives.

7 applicants applied for a fellowship reserved to participants living and working in a EU-13 Country or in Turkey, of which one for the first training module only (3 applicants from Turkey, 2 applicants from Romania, 1 applicant from Cyprus and 1 applicant from Malta) and 9 for a fellowship reserved to patient representatives. Moreover, 13 applicants asked for a fellowship, but without having the eligibility criteria.

Finally, 4 fellowships have been assigned, 3 to participants living and working in a EU-13 Country/Turkey, and namely to participants living and working in Malta, Romania and Turkey, and one fellowship to an ePAG, co-lead of a cross-ERN framework on healthcare transition and active member of two ERNs (ERN Rare Liver and ERKNet).

All fellowships have been awarded to attend both training modules.

Overall, 35 attendees have been selected, 23 to attend both training modules, 3 to attend the first module only and 9 the second module only.

To the residential course 19 of the selected participants attended both training modules, 4 the first module only and 7 the second module only.

Of the present participants 9 belonged to an ERN, representing 6/24 ERNs (25% of the ERNs), namely:

- ERN ReCONNET (2 participants)
- ERN EuroBloodNet (2 participants)
- ERN EURO-NMD (2 participants)
- ERN ITHACA
- ERN Rita
- ERN TransplantChild

4 participants were patient representatives.

The EJP RD/EU-13-Countries represented in the course were: Austria, Belgium, Cyprus, France, Germany, Italy, Latvia, Malta, Romania, Slovenia, Spain, The Netherlands, Turkey.

The 20 Speakers of the course belonged to the following institutions from 6 different countries (France, Italy, Spain, The Netherlands, UK, USA)

- University of Glasgow, UK (EndoERN)
- Centro de Biotecnología y Genómica de Plantas (CBGP, UPM-INIA), Spain
- The University of Manchester, UK
- Leiden University Medical Centre, The Netherlands
- BBMRI-ERIC ELSI Service & Research Unit, Italy
- National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- Academic Medical Center, Universiteit van Amsterdam, The Netherlands
- Center for Molecular and Biomolecular Informatics, Radboud University Medical Center, The Netherlands
- Global Medical Operations & Patient Registries Amicus Therapeutics, USA
- EURORDIS, France
- Orphanet, INSERM, France
- ERN Guard Heart, The Netherlands
- European Commission's Joint Research Centre, Italy
- University of Trento, Italy
- Groeningen University Medical Center, The Netherlands

3 of the Speakers held their presentation online.

In the first training module in plenary presentations, work in small groups and hands-on exercises have been carried out, in a highly interactive setting, while in the second training module also demos of specific tools have been performed.

The duration of the course has been of 34 hours, divided in morning and afternoon sessions in all five training days. The first training module has been composed of 21 hours and the second of 13 hours.

The course received the endorsement of ICORD.

The satisfaction questionnaires, filled by 15 participants for the first and for the second training module, registered a high satisfaction rate with 100% of the participants declaring "agree/strongly agree" to the statement "The course increased my knowledge" for the first training module and 87% for the second training module.

For the first training modules, the responses to the other items of the satisfaction questionnaire have been:

- 77% to item "The contents were appropriate for my level of knowledge"
- 93% to item "The objectives were clearly defined"
- 87% to item "The teaching method was effective"
- 100% to item "Time dedicated to individual/group exercises is sufficient"
- 80% to item "The learning materials were sufficient and of good quality"
- 88% "The number of speakers was sufficient"
- 100% to item "The speakers were competent and prepared"
- 100 to item "The course is well organized"
- 93% to the item "The connection to the event was easy".

For the second training module the responses to the other items of the satisfaction questionnaire have been:

- 53% to item "The contents were appropriate for my level of knowledge"
- 93% to item "The objectives were clearly defined"
- 73% to item "The teaching method was effective"
- 87% to item "Time dedicated to individual/group exercises is sufficient"
- 87% to item "The learning materials were sufficient and of good quality"
- 80% "The number of speakers was sufficient"
- 93% to item "The speakers were competent and prepared"
- 93% to item "The course is well organized"
- 94% to the item "The connection to the event was easy".

Free comments from the satisfaction survey indicated a good balance of the topics addressed during the training course, the networking and interactions between speakers and participants, and the understanding of the FAIRification process allowed by the course. The points of enhancement referred to structure more the hands-on sessions and that some sessions were too specific for "beginners".

Discussion

The five editions of the “International Summer School on RD Registries and FAIRification of data” have given place to two in site and three online editions of the training course. The adaptation to the online versions has been made necessary by the outbreak of the Covid-19 pandemic, that limited travels, as well as face-to-face events, in many countries, in the period from March 2020 to December 2022.

Along the five editions of the “International Summer School on Rare Disease Registries and FAIRification of data”, 151 participants have been trained, of which 126 in both training modules “RD Registries” and “FAIRification of data”, 12 in the first training module only and 13 in the second training module only. Altogether, 27 different EJP RD/EU-13 Countries have been represented by the participants. The 27 countries are: Austria, Belgium, Canada, Cyprus, Czech Republic, Denmark, Estonia, France, Germany, Georgia, Greece, Hungary, Ireland, Israel, Italy, Latvia, Lithuania, Malta, Norway, Portugal, Romania, Slovenia, Spain, Switzerland, The Netherlands, Turkey, UK (as shown in figure 1). Of these, 8 were EU-13 Countries (Cyprus, Czech Republic, Estonia, Hungary, Latvia, Lithuania, Romania, Slovenia), and Turkey (as shown in figure 2), that represented the countries who could benefit from travel and accommodation fellowships.

Figure 1: EJP RD/EU-13 Countries represented by the participants of the five editions of the training course

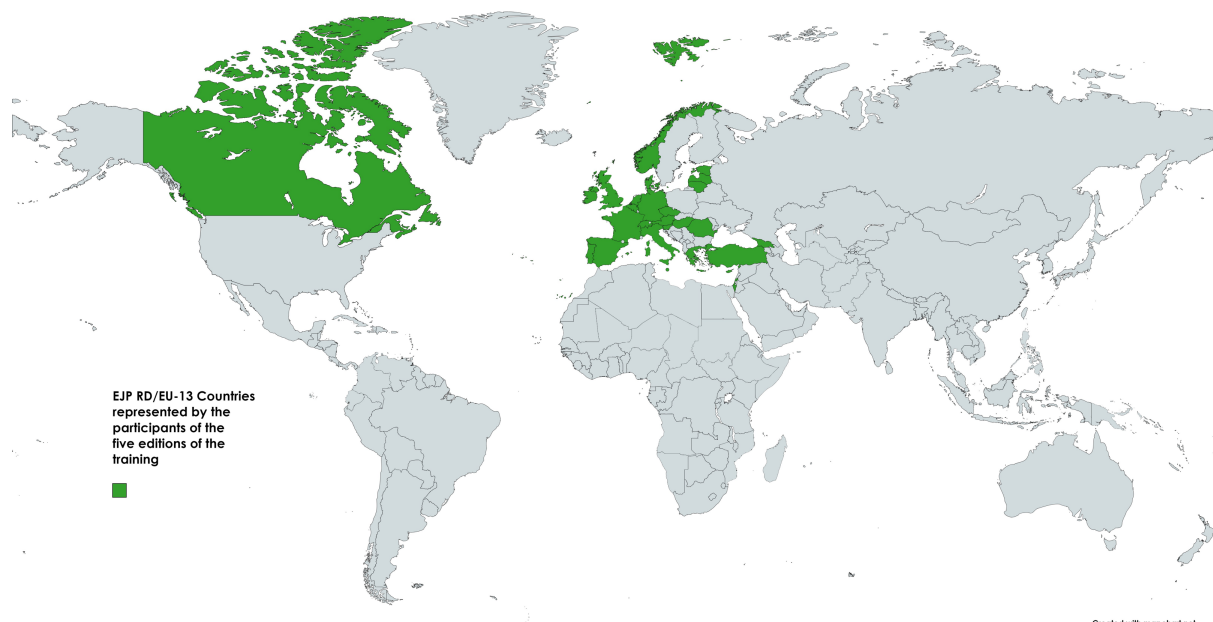


Figure 2 shows the countries of residence and employment of the participants that received a travel and accommodation fellowship reserved to EU-13 Countries (and Turkey in the last edition of the course). Overall, 5 participants have been awarded by this kind of fellowship. The countries of residence and employment of these participants were Cyprus, Lithuania, Malta, Romania and Turkey.

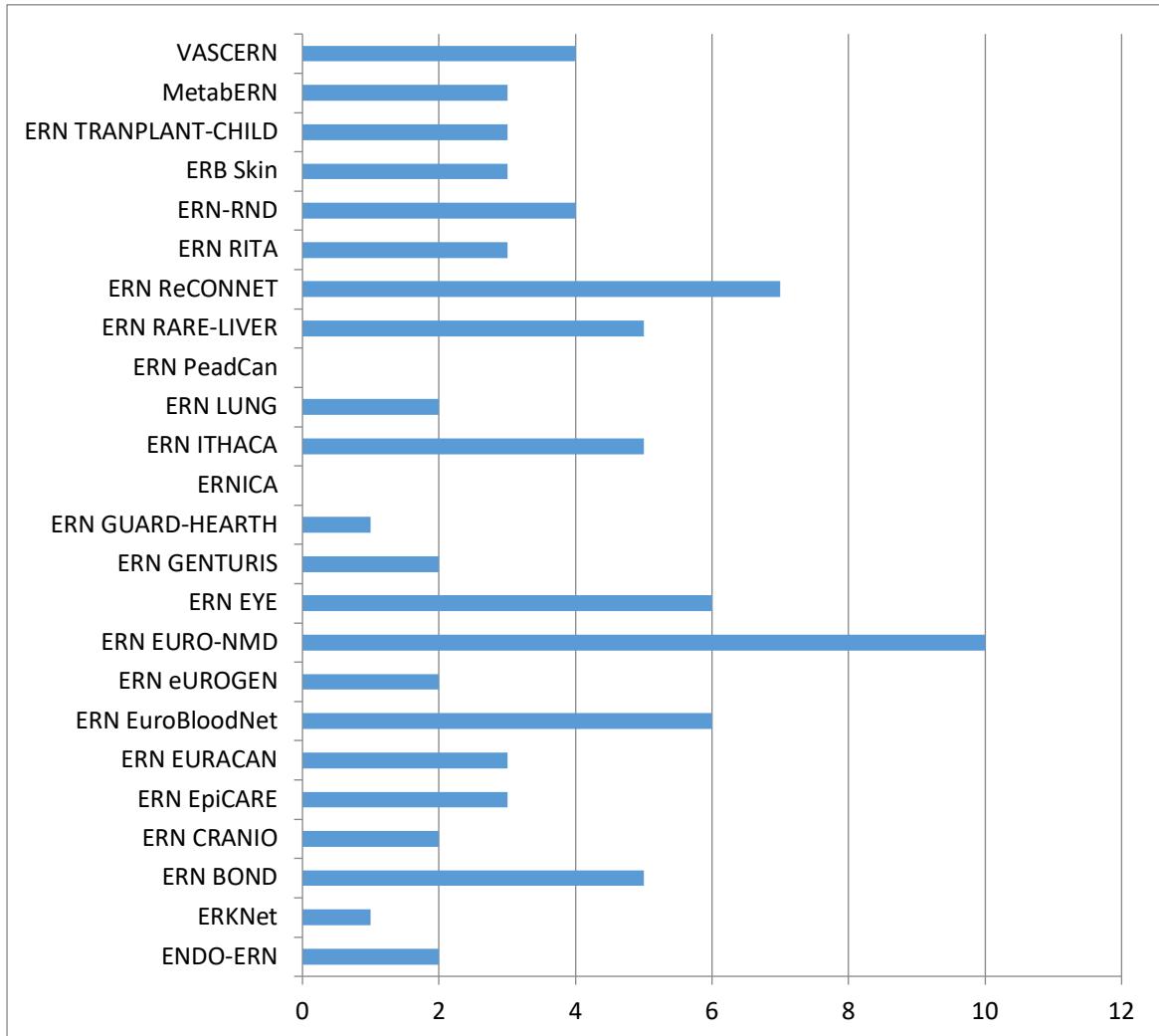
Figure 2: countries that received travel and accommodation fellowships reserved to participants living/working in a EU-13 Country/Turkey



With regard to the participation of patient representatives to the course, 19 have participated over the five years of the training, 4 of which received a travel and accommodation fellowship.

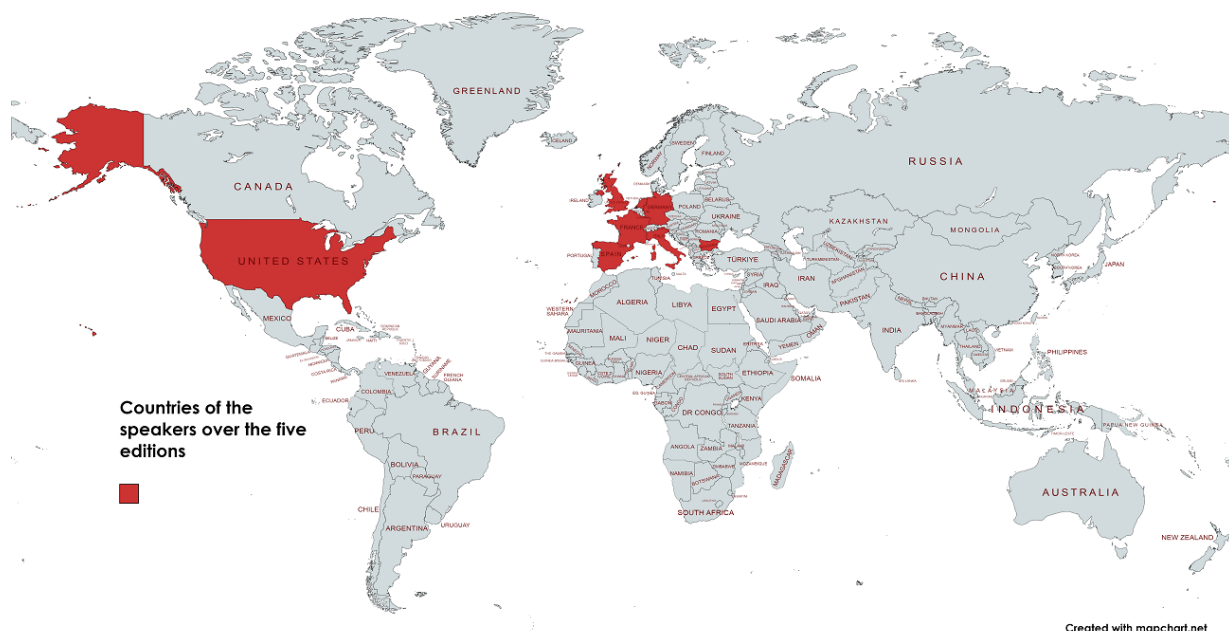
Coming the ERNs, 83 of the participants belonged to an ERN. Figure 3 shows the participation of ERNs in the five editions, with 92% of the ERNs represented along the five editions of the training.

Figure 3: participation of the ERNs in the five editions of the training



49 Speakers participated in the training over the five years, belonging to 8 different countries (as shown in figure 4): Bulgaria, France, Germany, Italy, Spain, The Netherlands, UK, USA).

Figure 4: countries of the speakers that participated over the 5 years of the training



A total of 144 of training hours has been delivered.

For each edition of the course training material including scientific papers and the presentations of the speakers that agreed to share the presented material has been shared with the participants of the course.

Certificates of attendance have been handed to the participants that attended 100% of the training modules (or were excused for eventual absences).

All the satisfaction surveys highlight a positive appraisal of the course, with percentages between 80%-100% of the participants declaring that the course increased their knowledge, for both training modules. It can be stated that for 4 of the course editions these satisfaction rates range from 87% to 100%, and that the lower rates (80%) regard only the 2020 edition of the course, in coincidence with the outbreak of the Covid-19 pandemic and the first online adaptation of the course. Special efforts have been undertaken in each preparatory phase of the course to enhance the quality of the training, taking particularly the feedback provided by the participants into account.

Conclusions

The “International Summer School on Rare Disease Registries and FAIRification of data” figures as an expansion of existing training courses organised by ISS titled “International Summer School on Rare Disease and Orphan drug Registries” and editions of the workshops on the FAIRification of the registries.

The adaptation and expansion of the training meets the key intention of Pillar 3 of the EJP RD to delivering a seamless flow of training programmes on key aspects relevant for rare disease research across the RD community.

The five editions of the "International Summer School on Rare Disease Registries", organised by ISS in close collaboration with Task 14.5 Partners in the period end of September/beginning of October 2019-2023 provided training to clinicians, medical specialists, registry curators, database managers, healthcare professionals and rare disease patients' representatives of different EU and non-EU countries.

The trainings allowed free of charge education, as well as the possibility to receive travel and accommodation fellowships, in line with Pillar 3 specific attention to the needs of ERNs and EU-13 Countries. Turkey has been added as target country to receive travel and accommodation fellowships.

The speakers of the training courses have been selected among the leading experts of international standing in the field of RD Registries and FAIR data.

The trainings were based on an interactive teaching methodology, that required some special adaptation after the outbreak of the Covid-19 pandemic and the consequent impossibility to deliver residential trainings in the years 2020, 2021 and 2022.

Despite the online delivery of three of the five editions of the training, all editions allowed to establish networks among participants and among participants and experts.

All the training editions were evaluated very positively from the participants, as shown by the results of the satisfaction surveys. Moreover, the hints pointed out by the participants in these surveys were taken into account to enhance the different editions of the training course over the years.