EJP RD

European Joint Programme on Rare Diseases

H2020-SC1-2018-Single-Stage-RTD
SC1-BHC-04-2018
Rare Disease European Joint Programme Cofund

Grant agreement number 825575

Del 14.9

First Report on International course on RD Registries and FAIRification of data at the source

Organisation name of lead beneficiary for this deliverable:

26 – ISS

Due date of deliverable: month 12

Dissemination level:

PU

Introduction and Objective

This document is a report on the training activity on Rare Disease Registries (RD Registries) and FAIRification of data at the source, that took place in Rome, at Istituto Superiore di Sanità (ISS) from 23 to 27 of September 2019.

The International course titled "International Summer School on RD Registries and FAIRification of Data" is the training activities of the task 14.5 which is part of Work Package 14, WP14, "Training on data management and quality", both led by ISS, which falls under the action of Pillar 3 "Capacity Building & Empowerment" of the European Joint Programme on Rare Diseases (EJP RD).

Pillar 3 through a common joint education & training programme aims to raise the level of knowledge and know-how within the RD research and care community, including ERNs and RD patient representatives in order to ensure equity and optimize outcomes for people living with rare diseases

To this purpose, the activities of Pillar 3 concentrate on developing and delivering a seamless flow of training programmes on key aspects relevant for RD research and challenges across the RD community.

Overall, the main objectives of WP 14 are to decrease RD data fragmentation and increase data quality through training activities on data management & quality which will raise the level of capacities and help data sharing and networking within the RD community.

The training activities, organized by this WP, are face-to-face courses hosted in different countries, with a focus on (i) Orphanet nomenclature and RD ontologies for RD research (ii) Standardization of RD data; (iii) Standards and quality genetic testing; (iv) Strategies to foster solution of undiagnosed RD cases; (v) Biological RD sample data management and sharing, (vi) Establishment and long term sustainability of International, FAIR and high quality RD registries and (vii) Learn components and interoperability tools of the European RD Registry Infrastructure.

In particular, the relevance of the proposed international training course lies in the fact that registries are key resources to increase timely and accurate diagnosis, improve patients' management, tailor treatments, facilitate clinical trials, support healthcare planning and speed up research.

ISS, leader of this task, has gained vast experience by organizing numerous courses focused on rare disease registries with the support of key partners. In particular, since 2013 ISS, with the support of RD-Connect and other key partners, has organized and hosted the "International Summer School on Rare Disease and Orphan Drug Registries" and since 2014 the "Bring Your Own Data To Link Rare Disease Registries". The EJP RD has enabled further development of the programme and has provided three travel fellowships for patients and three fellowships for participants living and working in EU13 countries

Methodology

Definition of training programme

The International course was organized and hosted in ISS, Rome, Italy at the end of September, in close collaboration with task partners [LUMC (Endo-ERN), UoG (Endo-ERN), IOR (Bond-ERN), HSK (Metab-ERN), EURORDIS, ISCIII, LUMC, INSERM (RaDiCo), UMCG, DTL-Projects (ELIXIR-NL), CNR (ELIXIR -IT), AMC].

The course was structured as a residential face-to-face 5-day training, articulated in two training modules with specific aims:

- The first training module (23-25 September 2019) dedicated to: (a) what resources are needed for the establishment/maintenance of a high quality FAIR registry, (b) the features of successful strategies to ensure (i) long-time sustainability of the registry, (ii) quality, (ii) legal and ethical issues in compliance with the EU General Data Protection Regulation and (iv) FAIR principles.
- The second training module (26-27 September 2019) dedicated to: (a) FAIRification of case data; the potential of a FAIR registry as the basis for cross resource questions; execution of a query across the use cases that become FAIR. (b) discussing FAIR data management and FAIR project planning.

Training method, Selection of speakers and dissemination of the event

The selection of the speakers, IT-trainers and Facilitators was done in close collaboration with task partners.

Trainers came from 20 different institutions and were mainly EJP RD partners. Collaboratively, the trainers prepared the programme of the course based on previous experiences.

An optional (not mandatory) preparatory webinar for the selected participants attending the second training module was delivered at the beginning of September 2019.

During the first training module on RD Registries the Problem-Based Learning (PBL) was applied as training method. This method uses complex real-world problems to drive a student-centred learning process and provides opportunities for working in groups and for finding and evaluating research materials.

The second training module on FAIRification of data, was based on hands-on "Bring Your Own Data" (BYOD) experiences, consisting of both plenary and break-out sessions, active exercises, work on tutorials, on sample datasets (fake, anonymized data) and lightweight tools. The course was disseminated through

the EJP RD website and different websites of the EJP-RD partners including media channels of ERNs and patient associations

Participants and registration

To ensure active participation and exchange with teaching staff and between participants a maximum of 30 attendees was planned.

The course was open to the research community, to clinicians, medical specialists, healthcare professionals and RD patient representatives involved in or intending to establish RD registries, mainly inside the ERNs.

Selection criteria were based on the participants' background, their role with reference to registry activities, involvement in ERNs.

The EJP RD grant allowed three fellowships for participants living and working in an EU13 Country and three fellowships for selected rare disease patient representatives.

The registration was carried out using LisyLime of Inserm.

The confirmation of the acceptance was sent to the selected participants, including those to whom the fellowship has been assigned one week after the registration was closed.

A waiting list for applicants who were not selected was foreseen in case of cancellations made by selected participants for unexpected reasons. The reserve list was kept open until one month before the start of the course.

Participants could choose to submit their request for attending one or both training modules.

At the end of the two training modules participants presented the group solution to the expert.

The assessment also covered the satisfaction level of the participants, which was collected through a satisfaction questionnaire composed of nine multiple choice items and two open questions.

Results and discussion

72/212 prospective applicants filled in the registration survey correctly and completely. The high number of non-completed registration forms through the survey will be taken into account and the survey will be reviewed accordingly for the next edition.

Applications that were submitted after the established deadline were not taken into account.

Following the high request to attend the course in the end 38 instead of 30 participants were selected of which 5 received fellowships.

Three fellowships were assigned to patient representatives based on a scoring matrix established by EURORDIS.

There were three fellowships available for participants from EU13 countries, however in the end only two were assigned. They were given to participants working and living in Cyprus and Lithuania. The third fellowship available for EU13 countries had been assigned a first time, then re-assigned to a different applicant a second time. However, both of these applicants cancelled their participation.

Overall, 30 candidates for both training modules were selected, as well as 3 candidates for the first training module only, and 5 candidates for the second training module only.

Three of the participants selected to attend both training modules, as well as one participant selected for the first training module only, cancelled their participation close to the start of the training course.

A total of 34 participants attended the course (comprising both training modules/ first training module only / second training module only).

21 of 34 participants that attended the second training module followed the preparatory webinar that was held on the 3rd of September 2019.

50% (12/24 ERNs) of ERNs were represented with 20 selected participants, and in more detail:
- 3 participants from
  VASCERN;
- 2 participants from
  ERN EPICARE, ERN ITACA, ERN Euro Blood Net, ERN Re CONNET, ERN-Euro-NMD, ERN TRANSPLAN-CHILD;
- 1 participant from
  ERN Eurogen, ERN EURACAN, ERN SKIN, ERN EYE, ERN BOND.

Participants were from 12 different countries of employment, precisely from Cyprus, France, Germany, Hong Kong, Italy, Lithuania, Norway, Spain, The Netherlands, Turkey, Ukraine, and United Kingdom

13/34 attendees were already involved in a RD registry (1 did not answer) and 13/34 intended to establish a RD registry in the future.

Four of the participants are involved in the EJP RD and four participants were Patients' Representatives.

The training course was delivered 23-27 September 2019 in ISS, Rome. The duration of the entire training course was 40 hours.

For the first 3-day module on RD registries, the attendees were divided into 4 working groups for the PBL sessions. Expert presentations took place at day 2 in a plenary session.

At the end of the first training module, all of the working groups presented the problem solution successfully in a plenary session and got feedback from the experts.

During the second training module on FAIRification of data, participants saw the potential of a FAIR registry by answering cross resource questions and executing a query across the use cases that became FAIR. Participants had the possibility to work on their own anonymized datasets.

A time slot was dedicated to FAIR project planning.

All the participants successfully completed the hands-on experiences.

19 participants in the first training module and 22 participants in 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".

Responses to the other items for module 1:

Participants that answered "agree/strongly agree":

- 73, 70% to item "The contents were appropriate for my level of knowledge";
- 89, 50% to item "The objectives were clearly defined";
- 84, 40% to item "The teaching method was effective";
- 94, 80% to item "Time dedicated to individual/group exercises is sufficient";

- 63, 20% to item "The learning materials were sufficient and of good quality";
- 100% to the item "The number of speakers was sufficient";
- 94, 80% to item "The speakers were competent and prepared";
- 89, 50% to item "The course is well organized".

Responses to the other items for module 2:

Participants that answered "agree/strongly agree":

68, 20% to item "The contents were appropriate for my level of knowledge";

81, 90% to item "The objectives were clearly defined";

77, 30% to item "The teaching method was effective";

90% to item "Time dedicated to individual/group exercises is sufficient";

68, 20% to item "The learning materials were sufficient and of good quality";

90, 90% "The number of speakers was sufficient";

100% "The speakers were competent and prepared";

87, 30% "The course is well organized".

Free comments in the satisfaction questionnaire were: "Time allocated to each speaker for questions was perfect, tutorials were well presented and well understood"; "I learned new skills and knowledge, there were interesting discussions"; "The entire course was extremely useful"; "A lot of examples and hands-on experience".

The pdfs of the training materials were sent in order to be published with the consent of the invited speakers on EJP-RD website and ISS website.

Moreover, the course received a good feedback through social media, having, for example, on twitter, a good number of Retweets, Likes, and Interactions. This helped the EJP-RD account increase its number of followers

Conclusions and next actions

A follow up organized by task partners and speakers is foreseen in the next few months to assess participants' long-term learning and application to professional challenges.

The next edition of the training course will take participants', speakers', IT-trainers' and facilitators' feedbacks into account to elaborate an improved programme of the course. Moreover, the progress of the ongoing EJP-RD

project will be taken into account in order to transfer promptly any advances and progress related to the aims of this course.

Dates have been already set. The international course will take place at ISS, Rome from 28 September to 2 October 2020.

The results of 2019's edition of the training course were presented at the 14th Meeting of the International Conference on Rare Diseases and Orphan Drugs (ICORD), 11-13 November 2019, Rishon-Lezion, Israel.