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«First Report on International course on undiagnosed diseases» (Deliverable 14.5)

Introduction and Objective

This document is a report on the training activity on undiagnosed rare disease cases that took place on 27-29 April 2020, organized by Istituto Superiore di Sanità (ISS) in close collaboration with the partners of the European Joint Programme on Rare Diseases (EJP RD), [EKUT, LBG (LBI-RUD), ACU/ACURARE, ISCI, INSERM (AMU), FTELE, UMCG, IMAGINE, CNAG-CRG, IPCZD (CMHI)] and with the endorsement of ICORD.

The International course titled “International Course: Training on Strategies to foster solutions of undiagnosed rare disease cases” is the training activity of task 14.3 which is part of Work Package 14 (WP14), led by ISS, “Training on data management and quality”, that falls under the action of Pillar 3 “Capacity Building & Empowerment” of the 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 planned as face-to-face courses hosted in different countries, with a focus on (i) Standardization of RD data; (ii) Standards and quality genetic testing; (iii) Strategies to foster solution of undiagnosed RD cases; (iv) Biological RD sample data management and sharing; (v) Establishment and long term sustainability of International, FAIR and high quality RD registries and (iv) Learn components and interoperability tools of the European RD Registry Infrastructure.

In particular the relevance of the proposed international training course on undiagnosed rare disease cases lies in the provision of newly developed tools and strategies based on phenotypic and molecular analysis to foster solutions for undiagnosed rare disease cases for researchers and clinicians.

ISS, leader of this task, has organized this training course in close collaboration with task partners, “Solve-RD”, and the “Undiagnosed Diseases Network International” (UDNI).

Methodology

Definition of training programme

The International course was planned to take place in Rome, Italy, hosted in ISS at the end of April 2020, as a face-to-face 3-days training.

Due to the emergency situation given by the Covid 19 pandemic and the extraordinary measures to limit the spread of the virus, the course has been adapted to fit an online format.

The course has been structured to maintain its specific aims in guiding participants to:

- discover novel molecular strategies, functional analysis, integrative multi-omics interpretation and clinical utility;
- identify clinical pathways and innovative methods to reach diagnosis;
- learn methodologies and tools that are already internationally used, providing useful examples for the resolution of undiagnosed rare disease cases;
- integrate useful tools, instruments and knowledge on novel strategies to foster solutions of undiagnosed RD cases through the presentation of sample use cases that have long eluded diagnosis;

- facilitate networking among professionals involved in undiagnosed conditions.

Training method, selection of speakers and dissemination of the event

The selection of the speakers and facilitators has been done in close collaboration with task partners.

Trainers came from 10 different institutions and were mainly EJP RD partners. Collaboratively, the trainers prepared the outline of the course, redefining the contents based on previous experiences and received feedback.

The information regarding the training course has been diffused through the EJP RD website and on various social media channels including Twitter.

The Problem-Based Learning (PBL) was planned to be adopted as training method. After discussion between the course organizers and Pillar 3 members, it has been agreed that the PBL method was not suitable for a three days online training course. To maintain a highly interactive setting of the training course, the PBL has been replaced by 30 minutes to 1 hour question and answer (Q&A) sessions that followed the online presentations held by the experts. Moreover, speakers in order to maintain the interaction with the participants, organized their presentations with interactive questions for the participants. On the third day, tool demo and hands-on exercises have been programmed.

Participants and registration

To ensure active participation and exchange between teaching staff and participants, a maximum of 30 participants was foreseen to be selected.

The course was open to the research community, to clinicians, medical specialists, interested in rare diseases. Selection criteria were based on the participants' professional background and experience in i) genotypic and/or phenotypical identification of rare disorders, ii) deep phenotyping; interfering variants, iii) digital technologies in rare diseases. Priority has been given to participants involved in ERNs, national and international rare disease programmes and projects (UDNI, Solve-RD).

The EJP RD funding included provision of four travel fellowships for participants living and working in an EU13 Country.

Online registration was done using LisyLime at Inserm.

Registration confirmations were sent to the selected participants including those to whom the fellowship had been assigned one week after registration closed (26 January 2020).

A waiting list for applicants who were not initially selected was foreseen in case of renunciations of the selected participants, for unexpected reasons. The reserve list was kept open until 27 February 2020.

At the end of the training course participants have been asked to fill a 28 item multiple choice learning assessment questionnaire, and a satisfaction questionnaire composed of 9 multiple choice and two open questions. Both questionnaires had to be compiled and then returned by email.

Results and discussion

60 applicants filled in the registration survey correctly and completely. Applications submitted after the deadline were not considered eligible.

37 instead of 30 participants were selected including 4 fellowships. 23 participants were assigned to the reserve list.

28 of the selected participants accepted their place including 4 participants with granted travel fellowships and 6 participants who had applied for a fellowship but accepted to participate without a fellowship.

Taking into account the number of replies and the COVID situation, 5 additional participants were selected from the reserve list.

Finally of the 33 participants admitted to attend the training course, 26 did participate in the online training course.

The course was delivered online on 27-29 April 2020 due to the emergency situation given by the COVID 19 pandemic and, as a consequence, the travel fellowships have not been used.

Moreover, even if the course was adapted as online some of the attendants were not able to join the training course, for example, 2 participants had to cancel their participation on the starting day of the training course because they were recalled to work in their hospitals for the Covid 19 emergency situation.

The 26 participants came from: Spain (3), Italy (9), UK (1), Austria (1).

Eu13 represented Countries: Bulgaria (3), Romania (3).

Other Countries: Turkey (1), North Macedonia (1), Ukraine (1), Georgia (3).

Among the selected participants there were representatives of 12 ERNs (50% of ERNs) Due to the Covid 19 situation some of the participants had to cancel

their participation unexpectedly so that at the end representatives belonging to 9 different ERNs attended the training course.

4 of the participants were already involved in national or international programmes and projects related to unsolved RD cases. All the participants were involved since 1 to more than 30 years in the rare diseases field. 6 participants were EJP RD Partners/Linked Third Parties.

Results of the satisfaction questionnaire:

21 participants returned the filled satisfaction questionnaire. Below are reported the percentage of participants who "Agree/Strongly agree with the following questions:

- "The course increased my knowledge": 100%
- "The number of speaker/teacher was sufficient": 100%
- "The speakers/teachers were competent and prepared": 100%
- "The course was well organized": 100%
- "The teaching method was effective": 95,24%
- "The learning materials were sufficient and of good quality": 95,24%
- "The contents were appropriate to my level of knowledge": 80,96%
- "The objectives were clearly defined": 76,19%
- "Time dedicated to individual/group exercises is sufficient": 57,15%

The analysis of the satisfaction questionnaires shows a general positive evaluation of the entire training course.

Other comments included:

- "Most recent and reliable information "
- "Friendly atmosphere, interaction, international speakers, new perspectives, current topics "
- "Given the conditions (COVID 19), the course was well designed and organized "
- "Speakers were so great; the organization in this virtual context was impressive, was so great to find people present on the course from all over Europe, we thank you so much for the 3 places you offer to Romania, really much appreciated "
- "Innovative and up-to-date information on rare and undiagnosed diseases; successful symbiosis of clinic and management issues;

authoritative and competent speakers; wide geography of speakers representing not only European but also North American continent; good attendance of the course, almost all participants were constantly online for three days; intensive interactive communication between speakers and participants; atmosphere of peer-to-peer professional communication created by course organizers; no one was tired in a three days of the course, at least I can speak about myself and the representatives of my country; the course was organized brilliantly; I will participate in the following conferences on undiagnosed diseases

- The effort to organize the course despite the difficult situation; up-to date information and many useful things
- Bright spectrum of clinical cases; meeting new specialists, working in round table, very good organization and structure
- Really experienced and competent speakers, good atmosphere, good opportunity to connect with so many specialists and to learn from them
- Highly international context, with broad knowledge and possibility to continuously interact with teachers and organizers; the content was ambitious and I not only had the opportunity to update my skills but also to confront my daily work with everyone else's across Europe
- Friendly approach, excellent speakers, good feed-back from participants
- Quality of speakers, excellent organization, although the need to do it online, useful messages to take home
- High scientific content, promotion to networking
- The overall environment that was created during the course, friendly and characterized by high level
- Sharing experience in rare diseases is really important. In our lab we have undiagnosed rare disease cases and different methods and approaches would be really helpful for solving them
- Presentations and cases were selected properly and leaved the desired message; all speakers were experienced and presented the information in a friendly and easy acceptable way; lections and cases were informative; I appreciate it

Positive comments were shared also on the social media.

Conclusions and next actions

The next edition of the training course will take participants' and speakers feedback into account to elaborate a new, enhanced version of the course.

Moreover, the progress of the ongoing EJP-RD project will be taken into account in order to transfer promptly any improvement related to the aims of this course.

The dates for the next edition of the training course have been confirmed, the international course will take place from 12-14 April 2021, ISS, Rome Italy. In case the COVID 19 situation continues, or a new emergency situation arises, the course will become an online course.

