

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 15.4

Second Report on training of patient representatives on scientific innovation and translational research in RD

Organisation name of lead beneficiary for this deliverable:

Partner 77 – EURORDIS

Due date of deliverable: month 60

Dissemination level: Public

Table of Contents

1.	2
2.	3
3.	3
4.	4
5.	4
6.	5
7.	6
8.	6
9.	7
10.	7
11.	8
12.	8

1. Introduction and Objectives of the training

The advances toward the diagnosis, prevention, and treatment of rare genetic diseases are amongst the most innovative in medical research and science is moving faster than ever. New technologies such as genome editing and exon skipping tools are being developed in rare conditions, introducing new paradigms in diagnostics and therapeutics that will ultimately advance the science of more common diseases. Furthermore, rare disease research is trailblazing in patient engagement, with many rare disease patient organizations actively involved in research. In this environment, patients must be educated on the fundamentals of research in addition to drug development to express their needs, expectations and more importantly aspirations in the most knowledgeable and meaningful fashion.

In 2018, EURORDIS created the EURORDIS Winter School on Scientific Innovation and Translational Research (now Open Academy School on Scientific Innovation and Translational Research) in recognition of patients' need for support and training on these topics. The first two editions of this training in 2018 and 2019 have been funded by the EU project Solve-RD. From 2021 onwards, the programme of the course was enriched by the diverse and excellent expertise and knowledge within the EJP RD and the addition of faculty members among the EJP partners.

This School aims to provide patients with the knowledge and skills needed to understand translational research, genetics, the latest advances in diagnostic tools and pathways including those developed within Solve-RD, disease model systems, data (collection, sharing, and protection), patient-reported outcomes and the use of novel technologies in the context of rare diseases research.

The training sessions are delivered by experts in research coming from academia and industry, along with patient groups who will provide case studies in successful research engagement. The course also provides information on research funding in the EU, research funding strategies for patient groups and corporate collaborations.

The training originally took place at the Imagine Institute for Genetic Diseases in Paris, to enable daily on-site visits to relevant laboratories and interactive discussion between research teams and participants. However, the 2021 and 2022 editions had to take place online due to the COVID-19 spread across Europe. The 2023 edition was held in conjunction with the Open Academy School on Medicines Research & Development to foster greater knowledge-sharing and networking opportunities.

2. Training programme: definition of training content and methods

Since 2018, specific training needs and speakers have been identified by expert patients, EURORDIS, academic researchers and industry to shape the training programme.

The programme has online (available for free to anyone) and face-to-face training components. The face-to-face portion trains a group of expert patients during an intensive 5-day course, or 5 half-days online for the exceptional years of 2021 and 2022. Over 20 trainers provide the training each year.

The programme has been updated and improved over the years, according to the feedback of participants and to the latest developments in the area of translational research and scientific innovation.

3. Programme Committee and speakers

An informal Programme Committee has supported the development of the programme for the period 2020-2021 and again for the period 2022-2023, via Programme Committee calls and ad-hoc consultations. Over 20 EURORDIS Summer School trainers provide the training each year and the faculty is mostly recurrent. Some new faculty members were identified specifically for the most recent editions, with the support of the Programme Committee experts and of other faculty members.

4. Participants: applications and selection method

Participation is usually limited to 30-35 patient advocates and 10 researchers, whilst additional applicants are put on a waiting list. 2-3 patient advocates based outside of Europe are also permitted to attend as observers.

The application form is composed of over questions divided into the following main areas:

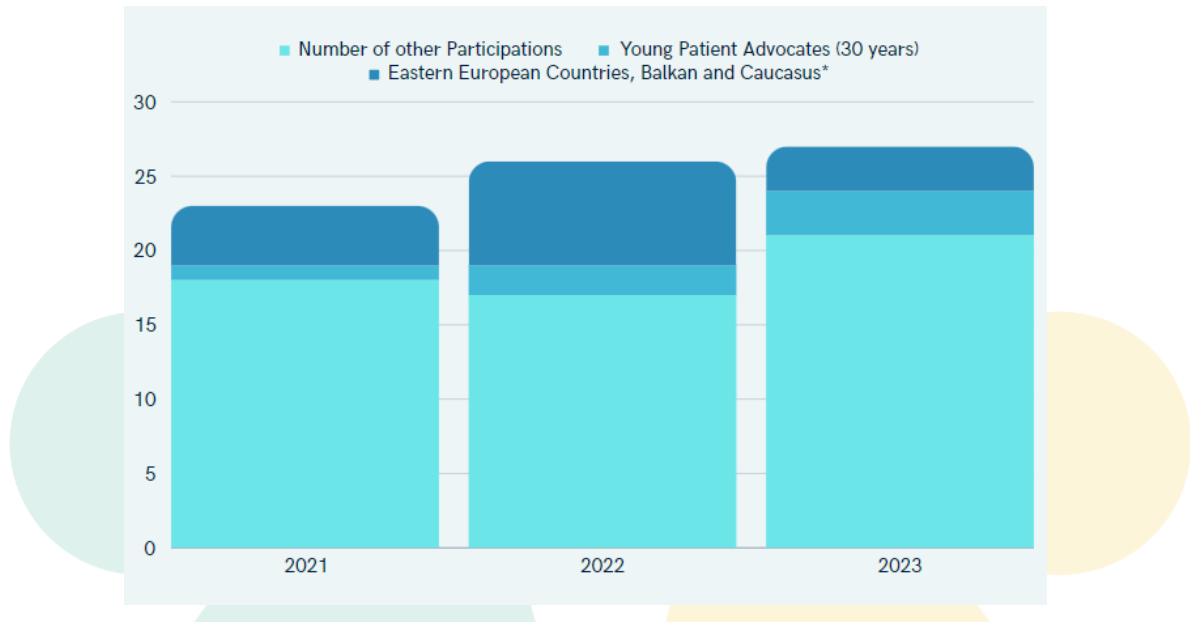
- Personal information, including disease/country represented and roles as patient advocate/researcher;
- Experience/knowledge in the training areas, previous training experience and motivation to attend the training;
- Information about the applicant's organization and its relations to national alliances/European federations for rare diseases;
- Agreement to fully attend pre-training and face-to-face session;
- Consent for data processing in the scope of the review of applications.

The applications' review of this EURORDIS Summer School consisted of 3 phases:

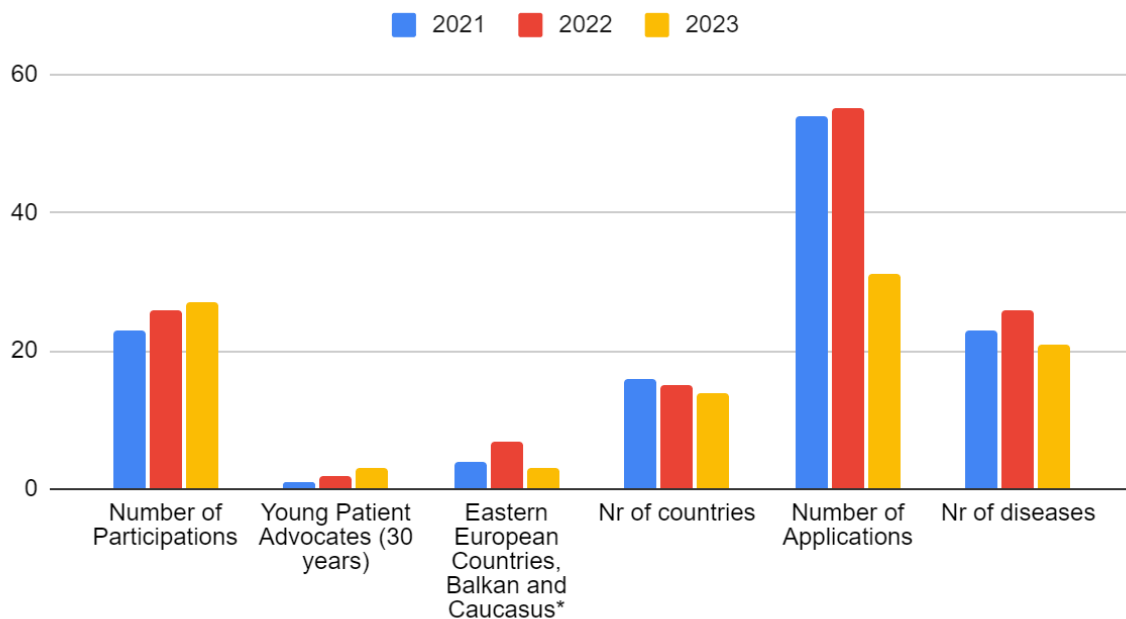
- Exclusion of non-eligible applications (e.g. non-EU; no affiliation to patient organisations);
- Scoring of applications (from 1-5 based on the main sections underlined above) by EURORDIS staff working with therapeutic development, research and training; researchers' applications were also reviewed by Professor Aartsma-Rus from Leiden University Medical Center (WP15.1 task partner);
- Final selection: selecting final list and waiting list from the highest scored applicants, also considering disease and country diversity.

5. Participants: profile

	2021	2022	2023
Number of Participations	23	26	27
Number of Applications	54	55	31
Young Patient Advocates (30 years)	1	2	3
Eastern European Countries, Balkan and Caucasus*	4	7	3
Number of countries	16	15	14
Number of diseases	23	26	21



2021, 2022 and 2023



6. Fellowships: process for attribution and results

5 fellowships are offered to patient advocates each year to attend the training in-person. This was not offered for the online editions.

The fellowship applications take place following the selection of attendees. The fellowships were attributed in accordance to the EURORDIS [fellowship evaluation scale available here: Fellowship application evaluation scale updated090922.docx](#)

7. Pre-training

The pre-training allows trainees to familiarize themselves with the concepts and terminology that is used during the training week, in view of optimising the onsite training and allowing for more fruitful discussions.

The pre-training for the School takes place in the 6 month preceding the intensive training week. It included 3 webinars and mandatory e learning courses (composed of video presentations, reading materials and case studies) available online (<https://openacademy.eurordis.org/winter-school/>), for example:

- Unit 1: Setting the landscape:
 - [Course 1: History in Genetics](#),
 - [Course 2: Existing resources, national and EC funding](#)
 - [Course 3: Translational research: what, why, how, and with whom?](#)
 - [Course 4: From care to research and research to care: ERNs](#)
- Unit 2: Genetic research to clinical diagnosis of Rare Diseases:
 - [Course 1: State-of-play in diagnosis](#)
 - [Course 2: Solve RD: solving the unsolved](#)
 - [Course 3: How to deal with results](#)
 - [Course 4: Case scenarios on secondary findings](#)
 - [Course 5: ERN-ITHACA: Diagnosis for the undiagnosed](#)
- Unit 3: Sharing patient data:
 - [Course 1: Patient generated data](#)
 - [Course 3: Agreeing what outcomes to measure](#)
 - [Course 5: Bioinformatic laboratory and discussion](#)
- Unit 4: From research to therapies:
 - [Course 3: Gene/Advanced therapies on the market](#)
 - [Course 4: Strategy of POs in driving research from lab to therapies](#)
- Unit 5: Genome editing:
 - [Course 1: Introduction](#)

8. Training delivery: programme outline

The face-to-face training (or online for the years 2021 and 2022) lasts 5 days and consists of a series of plenary, breakouts, and visits.

As the programme was developed for a face-to-face delivery, a series of adjustments had to be made to adapt it to ensure a successful online delivery. The daily programme was shortened and limited to 4 hours per day, including 2 comfort breaks. All the sessions were maintained, except the visits

to laboratories and one session that was considered the least relevant in the review of the previous editions.

When face to face, an example programme outline of the School training week is as follows:

- Monday: Setting the Landscape.
- Tuesday: Diagnosis.
- Wednesday: Use of Data.
- Thursday: Research & Therapies.
- Friday: Taking action!

When online, several interactive tools were used during the training – i.e. chat tool, polls, word clouds, breakout sessions and plenary networking sessions – to ensure the highest interactivity between participants and with the faculty. Special attention was paid to participants with visual impairments and other impairments to eliminate barriers that could be caused by the online delivery.

9. Training materials available

The e-learning courses used for the pre-training and additional courses on scientific innovation and translational research are available online [here](#). The pre-training webinar recordings and presentations (including recordings in the case of the online editions) were made available to the participants after the training. These are available to EJP partners on demand.

10. Evaluation: methodology and results

The evaluation of the programme is conducted via feedback questionnaires, filled in online and anonymously. At the end of the training day, participants receive a short questionnaire via which they are required to indicate, for each presentation, if it was “informative”, “relevant” and “too technical”. On the last training day, participants also receive the overall feedback questionnaire, including questions on the impact/value of the training, on the pre-training and on the preparation/logistics. In 2022, we adapted the questionnaire to be sent only after the full training, which increased the response rate and to adapt to changes in the programme.

	2021	2022	2023
% of participants who recommend the OA's schools	100%	100%	100%
Rate of satisfaction with the OA's schools: I am satisfied with my experience of the OA Schools			
Strongly disagree	0%	0%	5%
Disagree	0%	0%	0%
Neither Agree nor disagree	0%	4%	0%

Agree	9%	14%	5%
Strongly agree	91%	82%	90%
Rate of response to evaluation questionnaires	95%	84%	89%

Quotes on participants' key takeaways from the training:

"Meet people who convinced me to stay in the CAB that I was considering quitting. The conference about CABs helped that decision".

"The only way to advance in research and innovation is through networking. Rare diseases could represent a great value for scientific innovation, and we have to communicate it well to all the stakeholders".

"Getting organized as a patient organization or empowered as a patient/patient representative to be involved in research and make a difference".

"Patients can change a lot of things".

"Very good for networking not only with fellow patient advocacy colleagues but also with researchers and other professionals. Very good information to put into practice. However, there could be more exercises on how to do this practical part and not just be theory".

"The positive can-do attitude no matter what size the mountain is".

"I do not think I have 1 main take-away, but I think our Winter school group was very good. People were engaged, attentive, informed and really interested in learning as well as in sharing their experiences and expertise with others. I found that really stimulating and it made the networking more worthwhile"!

11. Dissemination

The applications for the trainings were widely disseminated via EURORDIS partners, social media as well as via the EJP's communication tools and partners.

12. Conclusion and Next Steps

The next edition of the School will take place in June 2024 during the extension period of the EJPRD project in the same in-person format as 2023.

Applications took place September-October 2023 and generated 102 potential candidates. We are in the final stages of finalizing the participants list and the pre-training stage will start in January.

We will introduce several changes as of 2025 with the advent of the ERDERA project, including different levels to the training (basic, intermediate, advanced), different tracks for participants to follow, translation of e-learning courses, a more blended format with more online elements to increase accessibility and more interactive in-person sessions. The advanced level will eventually entail access to a buddy/mentoring programme.

