

## **EJP RD**

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## **Del 15.3**

# **First Report on training of patient representatives on scientific innovation and translational research in RD**

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## 1. Introduction and Objectives of the training

*Based on EJP RD Annex1 description and Annual Work Plan for year 2:*

This task enables further development of a new 5-day annual residential training workshop for 30 patient representatives, focused on scientific innovation and translational research, organised by EURORDIS since 2018.

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 are being developed in rare conditions, introducing new paradigms in diagnostics and therapeutics. Furthermore, rare disease research is trailblazing in patient engagement, with many rare disease patient organisations 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 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 2020 onwards, the programme of the course is enriched by the diverse and excellent expertise and knowledge within the EJP RD and the addition of faculty members among the EJP RD partners.

This Winter 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 EURORDIS Winter School normally takes 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, this 2020 edition had to take place online due to the escalating COVID-19 spread across Europe during March 2020.

## **2. Training programme: definition of training content and methods**

The 2020 edition of the EURORDIS Winter School, built on the programme developed over the two previous training sessions.

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

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**

In September 2019, a formal Programme Committee for the EURORDIS Winter School has been established, to support the upcoming editions (2020-2022). This Programme Committee takes part in two annual Programme Committee calls, in view of reviewing the draft training programme and suggesting speakers or content to help address any identified gaps in the programme/faculty.

Around 20 experts provide the training each year. Some new faculty members were identified for the 2020 edition, with the support of the Programme Committee experts and of other faculty members.

## 4. Participants: applications and selection method

The applications for the EURORDIS Winter School 2020 took place in July-August 2019. EURORDIS received a total of 85 applications from 31 countries.

30 patient advocates were initially selected to attend, while additional 5 were put on a waiting list.

The application form of the EURORDIS Winter School 2020 was composed of over 30 questions/fields, divided into the following main areas:

- Personal information, including disease/country represented and roles as patient advocate;
- Experience/knowledge in the training areas, previous training experience and motivation to attend the training;
- Information about the applicant's organisation and its relations to national alliances/European federations for rare diseases;
- Agreement to fully attend pre-training and the intensive training sessions;
- 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 training, research and patient engagement;
- Final selection: selecting final list and waiting list from the highest scored applicants, considering also disease and country diversity.

## 5. Participants: profile

28 participants attended the EURORDIS Winter School in 2020. These represented 11 countries and over 22 rare diseases.

The countries represented included: Belgium, France, Germany, Ireland, Italy, Netherlands, North Macedonia, Romania, Serbia, Spain, United Kingdom.

Rare diseases represented included e.g.: ADNP Syndrome, Angelman Syndrome, Arrhythmogenic right ventricular cardiomyopathy (ARVC), Congenital Diaphragmatic Hernia, Cutis Laxa, Cystinosis, Duchenne and Becker Muscular Dystrophy, GSD Glycogen storage diseases, Hereditary Hemorrhagic Telangiectasia (HHT), Inherited bleeding disorders, Inherited Metabolic Disorders, LUPUS, Lymphoma and all haematological diseases, Osteogenesis Imperfecta, PTEN Hamartoma Tumor Syndrome, Pulmonary Hypertension, Retina diseases, SCN2A-related disorders, Waldenström's Macroglobulinemia.

## 6. Fellowships: process for attribution and results

5 patient advocates received a fellowship to attend the EURORDIS Winter School 2020. These were from Ireland, Italy, North Macedonia, Romania and Serbia.

Although the training took place online, these fellowships were still used, as the face-to-face event was cancelled on the week prior to the Winter School, due to the COVID-19 outbreak. Only one of the fellowship receivers was able to cancel his travel costs and did not need a travel fellowship.

The fellowship applications took place in November 2019.

The fellowships were attributed in accordance to the EURORDIS [fellowship evaluation scale available here: https://openacademy.eurordis.org/wp-content/uploads/2019/07/EURORDIS-Open-Academy-Fellowship-application-evaluation-scale.pdf](https://openacademy.eurordis.org/wp-content/uploads/2019/07/EURORDIS-Open-Academy-Fellowship-application-evaluation-scale.pdf)

## 7. Pre-training

The pre-training allows trainees to familiarise 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 EURORDIS Winter School 2020 took place from December 2019 to March 2020. It included 3 webinars and the following 15 mandatory e-learning courses (composed of video presentations, reading materials and case studies; [available online here https://openacademy.eurordis.org/winterschool/](https://openacademy.eurordis.org/winterschool/)):

- 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 training week of the EURORDIS Winter School 2020 took place on 9-13 March 2020, online.

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 the onsite laboratories at the Imagine Institute and one session that was considered the least relevant in the review of the previous editions.

The programme outline of the Winter School 2020 training week was the following:

- Mon, 9: Setting the Landscape;
- Tues, 10: Diagnosis;
- Wed, 11: Use of Data;
- Thurs, 12: Research & Therapies;
- Friday, 13: Taking action!

The [full programme of the online training week is available here](#):

<https://openacademy.eurordis.org/wp-content/uploads/2020/12/EURORDIS-Winter-School-2020-Online-programme.pdf>

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.

The sessions were video recorded and some of the recordings may be used to update the pre-training of future Winter Schools and the related e-learning courses available within the EURORDIS Open Academy Programme.



## 9. Training materials available

The e-learning courses used for the pre-training and additional courses on scientific innovation and translational research (total of 21 courses) are [available online here https://openacademy.eurordis.org/winterschool/](https://openacademy.eurordis.org/winterschool/). The pre-training webinar presentations were shared with all the participants.

All the presentations of the EURORDIS Winter School were made available to the participants ahead of the training and to all the faculty. These presentations 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 each 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.

The rate of response to the evaluation questionnaires of the EURORDIS Winter School 2020 was of 86%. The main results include:

- 100% of the respondents stated that they would recommend the Winter School to other patient representatives;
- 100% have found that the EURORDIS Winter School effectively builds the capacity of patient advocates on Scientific Innovation & Translational Research (75% strongly agreed, 25% agreed);
- 100% have found the topics relevant for their activity as patient advocate (58,33% strongly agreed; 41,67% agreed);
- 100% considered that the training provided them with essential knowledge and tools to support them in representing patients in activities related to Scientific Innovation & Translational Research (66,67% strongly agreed; 33,33% agreed);
- 91% of the respondents have found the pre-training useful to prepare for the Winter School (60,87 strongly agreed; 30,43% agreed).

The report with the full feedback from the participants has been used to improve the programme outline for the 2021 edition.

## 11. Dissemination

The applications for the EURORDIS Winter School were widely disseminated via EURORDIS media and social media as well as via the EJP's communication tools. A series of social media posts were disseminated during the training, under the hashtag #EURORDISWinterSchool.

## 12. Conclusion and Next Steps

The EURORDIS Winter School 2021 will take place on 12-16 April 2021. The training will again take place online, due to the COVID-19 pandemic.

Applications for the 2021 edition took place from July to September 2020. 54 applications were received, out of which 26 were selected. These participants represent 16 countries and over 20 rare diseases.

The programme for the 2021 edition was elaborated over the summer of 2020, considering the feedback from participants and in collaboration with the Programme Committee.

The first pre-training webinar took place on 2 December 2020. The pre-training will run until March.



### 13. Indicators

WP	Action/Activity	Result	Indicator	How is the indicator measured?	Target/threshold	Results Year 2, 2020	Indicator type	Who measures the indicator?	Who reviews the Indicator
WP 15.2	Training delivery	Pre-training and training webinars and online courses	All planned webinars and online courses	Number of webinars and courses actually delivered	2 webinars 2 courses	3 webinars 15 courses	KRI	EURORDIS Open Academy Director	Pillar 3 and WP15 leader (EURORDIS Scientific Director)
WP 15.2	Training quality	Programme Committees engagement	All planned consultations with Programme Committees	Number of calls, email consultations, etc. to Programme Committees	2 calls 1 email consultation	2 calls 1 email consultation	KRI	EURORDIS Open Academy Director	Pillar 3 and WP15 leader (EURORDIS Scientific Director)
WP 15.2	Training quality	Programme Committees contribution to training programmes	All recommendations implemented in online and/or face to face training courses	Number of reviews of the programmes integrating programme committees' feedback	1 annual review	1 annual review	KPI	EURORDIS Open Academy Director	Pillar 3 and WP15 leader (EURORDIS Scientific Director)
WP 15.2	Training quality	Review of pre-training and online courses according to alumni's feedback	All planned evaluation and annual follow-up with participants	Rate of satisfaction of participants regarding usefulness of the pre-training ( <i>Q: The pre-training was useful to prepare for the Winter School. Rate of satisfaction combines "Strongly Agree"/"Agree"</i> )	60%	91%	KPI	EURORDIS Open Academy Director	Pillar 3 and WP15 leader (EURORDIS Scientific Director)
WP 15.2	Training impact	Review of impact of the training on individual participants	All planned evaluation and annual follow-up with participants	Percentage of participants considering that the training effectively builds their capacity and/or provides essential knowledge and tools for their own activities ( <i>Q: The Winter School effectively builds the capacity of patient advocates on scientific innovation and translational research. Rate combines "Strongly Agree"/"Agree"</i> )	70%	100%	KRI	EURORDIS Open Academy Director	Pillar 3 and WP15 leader (EURORDIS Scientific Director)