



# Deliverable

## D3.9 Second training for ePAGs delivered

Version   Status	V1   final
Work package	WP3
Lead beneficiary	EURORDIS
Due date	31.12.2020 (M36)
Date of preparation	09.12.2020
Target Dissemination Level	Public
Author(s)	Gulcin Gumus
Reviewed by	Holm Graessner (EKUT)
Approved by	Gisèle Bonne (INSERM-CRM)



The Solve-RD project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 779257.

**Explanation according to GA Annex I:**

Training ERN patient representatives in scientific innovation and genome diagnostics.

**Abstract:**

This deliverable report covers the second edition of the EURORDIS Winter School, which is the capacity building training programme for rare disease patient representatives on scientific innovation and translational research. The EURORDIS Winter School consists of one week face-to-face training and of online training modules. The second edition of the EURORDIS Winter School on scientific innovation & translational research took place from 11-15 March 2019 at the Imagine Institute in Paris, France.

**Introduction:**

The EURORDIS Winter School on Scientific Innovation and Translational Research is part of the EURORDIS' Open Academy which brings together various capacity-building trainings for patient advocates. The EURORDIS Winter School aims to deepen patient representatives' understanding of how pre-clinical research translates into real benefits for rare disease patients. The training equips participants with knowledge and skills in order to empower them to effectively participate in discussions with researchers, policy makers and companies responsible for research or research infrastructures.

**Objectives:**

The main objectives of the Winter School are to improve RD research and innovation and to enhance the uptake of research results by building the capacity of the patient community and promoting the inclusion of patients in rare disease research specifically related to genome diagnostics.

The specific objectives of the Winter School include:

- Providing the knowledge and skills required for RD patients to become legitimate collaborators in RD scientific & translational research;
- Empowering the RD patient community and specifically patient representatives in their roles as equal, valued, and efficient partners in research and scientific project / ERNs governance bodies;

In order to meet these objectives, the second edition of the face-to-face training of the EURORDIS Winter School was held in March 2019. The training was delivered by expert speakers and researchers from across Europe and covered a broad range of topics on scientific innovation and translational research; including the history of genetics, the latest advances in diagnostic tools and pathways including those developed within the Solve-RD, new technologies in gene therapy, and drug repurposing. The programme also included laboratory visits and discussion with researchers. The Winter School also provided information on research funding in the EU, research funding strategies for patient groups and corporate collaborations.

**Report:**

**Venue:** The 5-day onsite training workshop took place at the Imagine Institute for Genetic Diseases in Paris which is a research and innovative healthcare institute, bringing together researchers, doctors and patients, with a common goal: better understand genetic diseases to better treat them.

The Institute is located on the campus of the Necker Enfants Malades Hospital, gathering all those concerned in the treatment of genetic diseases to create a synergy that encourages the transfer of knowledge, to speed up the discovery of new treatments and diagnoses.

Imagine institute, an official partner of EURORDIS Winter School, provided the venue for the Winter School and enabled daily onsite visits to relevant laboratories and interactive discussion between research teams and participants.

**Participants:** The applications' review of the EURORDIS Winter School consisted of three phases:

- Exclusion of non-eligible applications (e.g. non-EU; no affiliation to patient organisations);
- Scoring of applications (from 1-5 based on the experience/knowledge in the training areas, previous training experience and motivation to attend the training) by EURORDIS staff working with research and training.
- Final selection: selecting the final list and waiting list from the highest scored applicants, considering the disease represented and country diversity.

Out of 136 applications received, 30 patient representatives from 18 countries were selected to participate in the second edition of the Winter School.

The selected participants represented different organisations and networks, including European Patient Advocacy Group (ePAG) representatives from 6 European Reference Networks (ERNs) including ERN ITHACA and ERN EURO-NMD which are represented in Solve-RD and other ERNs (ReCONNET, VASCERN, ERN-Eye, ERN-BOND, ERN-LUNG and ERN-Skin).

Rare diseases represented included Sjögren Syndrome, Charcot Marie Tooth, Succinic semialdehyde dehydrogenase (SSADH) deficiency, Rett Syndrome, Duchenne & Becker Muscular Dystrophy, Pyruvate Kinase Deficiency (PKD), Marfan Syndrome, Cavernoma, Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency, Hereditary hemorrhagic telangiectasia (HHT), Mucopolysaccharidosis type III, Pulmonary Hypertension, LGMD2I, GNE Myopathy, Lymphoma, Cystic Fibrosis, Ectodermal Dysplasia, Alkaptonuria (AKU) and Leber's Hereditary Optic Neuropathy.

**Methodology:** The Winter School applied a mixed approach to deliver the training. The training programme included an online pre-training and a face-to-face training with interactive sessions, discussion groups, case studies, visits to laboratories and meeting with rare disease researchers. The thematic sessions and the training methodology were designed according to the aforementioned objectives and tailored according to the feedback from the first edition of the Winter School, in order to suit participants' needs and to ensure that the training objectives are met.

**Pre-Training:** In order to maximise the effectiveness of the onsite training and to familiarise the participants with the key concepts and the terminology that will be used during the onsite course, the participants were invited to the e-learning platform to complete the online training which consists of the following units:

- Setting the Landscape
- Genetic research to clinical diagnosis of Rare Diseases
- Sharing patient data
- From research to therapies
- Genome editing

The online training included presentations and videos from the training sessions from the first edition, glossary, useful links and reading materials on genetic diagnosis, genome editing technologies, drug repurposing and outcome measurements, video links on genome editing and quizzes at the end of each unit to assess participants' own knowledge. The online training can be found at the e-learning platform in [the EURORDIS Open Academy website](#).

In addition, the speakers were asked to define the learning objectives of their session and these learning objectives were sent to the trainees before the onsite training in order to manage expectations and highlight key elements and concepts to be learnt from the different sessions. Moreover, trainees attended two preparatory webinars that took place from January to February 2019. The webinars were organised in order to introduce participants to the programme, to the pre-training and to important logistics information. Webinar recordings were shared with all the participants.

**Onsite training:** The onsite training was delivered by experts in scientific and clinical research coming from academia and industry, along with patient groups who provided case studies in successful research engagement. The speakers were informed to prepare their materials for delivering complex information to lay audience. They were also briefed on the disease areas that trainees represented so that they could use disease specific cases and pertinent examples.

The training programme included plenary sessions, interactive breakout sessions and visits onsite to genetic sequencing laboratories and animal facilities. The programme was divided into 5 thematic sessions:

- **Setting the landscape:** This session was devoted to familiarise the trainees with the history of genetics, the concept of translational research and the research funding landscape including the role of patient engagement;
- **Diagnosis and therapies:** Introduced the basis of genetics, interpretation of genetic sequencing and the state of play in diagnosis;
- **Sharing patient data:** Focused on the use of data, data-sharing platforms and EU projects
- **Genetic Research to clinical diagnosis of rare diseases:** Aimed to bridge the gap between research and therapies by explaining the potential of genomics and disease models for diagnosis and development of therapeutic strategies;
- **European Reference Networks:** Provided information on the roles of European Reference Networks in healthcare and research.

Specific topics covered included genetics and diagnosis, genome editing tools, translational research, pre-clinical models, artificial intelligence for diagnosis, drug repurposing, IRDiRC activities, EJP-RD, the evolution of practice in animal research for the development of new therapies, patient participation in research projects and ERNs.

**Evaluation:** The evaluation of the programme is conducted via feedback questionnaires, anonymously filled in online.

At the end of each training day participants receive a short questionnaire in which they were 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 in 2019 was of 83%. 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 (81% strongly agreed, 19% agreed);
- 89.2% have indicated that they would share the knowledge received during the training with their organization and other patient advocates (82.1% strongly agreed; 7.1% agreed);
- 89.2% 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 (50% strongly agreed; 39,2% agreed).

#### **Addressing challenges from the previous edition:**

- The Winter School pre-training webpage on the EURORDIS Open Academy website has been updated and the content has been reviewed to ensure that there is a well-developed and comprehensive online pre-training.
- All the speakers were briefed on the educational approach required, the various level of knowledge of the trainees attending and overarching as well as specific training goals. The speakers were also advised to design their session in an interactive manner, by adding polls, quizzes, breakouts or other approaches to increase participant engagement and active participation.
- The programme has also been organised to be more interactive and engaging by allowing more time for breakouts, case studies and hands-on training on data sharing platform.

**Challenges for future editions:** To evaluate the overall quality of the Winter School and to ensure that the Winter School meets the real need of patients, EURORDIS decided to establish a programme committee for the future editions of the Winter School. The results of a detailed satisfaction questionnaire including feedback on the face to face training programme, the pre-training, as well as evaluation of speakers will be communicated to the programme committee. The programme committee will review this feedback, will provide input on potential speakers; and will contribute to the drafting of the face to face programme of the following year.

#### **Conclusion:**

A substantial majority of participants agreed that the Winter School effectively builds the capacity of patient advocates on scientific innovation and translational research and reported that the training experience will be useful in their work, that the goals of the training have been met and that the training provided an opportunity to meet others from different disci-

plines and backgrounds. All the respondents stated that they would recommend the Winter School to other patient representatives.

Evaluation and implementation of the feedback from the participants and the speakers within the next editions of the Winter School will support the development of the training programme for the future editions. The use of interactive techniques can increase active participation in the sessions, maximise their understanding of scientific concepts and increase their capacity as patient advocates, therefore, contributing to the added value of patient involvement in scientific research.

**Annex 1: EURORDIS Winter School 2019 Programme**



# EURORDIS Winter School

on Scientific Innovation & Translational Research

Imagine Institute, Paris

11-15 March 2019

*A capacity-building programme for patients  
on scientific innovation and translational research*

Co-organised by

imagine  
INSTITUT DES MALADIES GÉNÉTIQUES

Organised with the support of the EU-funded project

Solve<sup>RD</sup>

**Organisers:**

The EURORDIS Winter School is organised by:

EURORDIS-Rare Diseases Europe  
 Plateforme Maladies Rares  
 96 rue Didot 75014 Paris, France  
[www.eurordis.org](http://www.eurordis.org)

Raquel Castro

Virginie Bros-Facer

Aline Schnieder

Open Academy Director

Scientific Director

Open Academy Junior Manager

[raquel.castro@eurordis.org](mailto:raquel.castro@eurordis.org)

[virginie.bros-facer@eurordis.org](mailto:virginie.bros-facer@eurordis.org)

[aline.schnieder@eurordis.org](mailto:aline.schnieder@eurordis.org)

**The EURORDIS Winter School is co-organised by:**

*Imagine* Institute, Hôpital Necker – local organiser  
 Paris, France  
 24, Boulevard de Montparnasse  
 75015 PARIS  
[www.institutimagine.org/en/](http://www.institutimagine.org/en/)

## Venue:

*Imagine* Institute, Hôpital Necker  
 Paris, France  
 24, Boulevard de Montparnasse  
 75015 PARIS

## Funding:

The EURORDIS Winter School 2019 is funded by the European project “Solve-RD - solving the unsolved rare diseases”. <http://solve-rd.eu/>



*The Solve-RD project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 779257.*

**Financial support is also provided by:**

AFM-Téléthon

<http://www.afm-telethon.com/>

## Introduction

The **EURORDIS Winter School on Scientific Innovation and Translational Research** consists of one week face-to-face training and of online training modules, available for free.



EURORDIS launched the training with the aim of deepening patient representatives' understanding of how pre-clinical research translates into real benefits for rare disease patients. Expert speakers and researchers from across Europe deliver the face-to-face training sessions.

The training equips participants with knowledge and skills so they are empowered to effectively participate in discussions with the researchers, policy makers and companies responsible for research or research infrastructures.

The first edition of the face-to-face training of the EURORDIS Winter School was held in March 2018 in Paris. It covered topics including the history of genetics, diagnostics, new technologies in gene therapy, and drug repurposing and included laboratory visits.

### EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 800 rare disease patient organisations from more than 70 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

EURORDIS works across borders and diseases to improve the lives of people living with a rare disease. EURORDIS' vision is to enable better lives and cures for people living with a rare disease.

### Imagine Institute

*Imagine* is a research and innovative healthcare institute of a new type, bringing together researchers, doctors and patients, with a common goal: to cure genetic diseases.

The new Institute is housed in a 19,000 m<sup>2</sup> building located on the campus of the Necker Enfants Malades Hospital, and brings together over 900 scientists, doctors and healthcare personnel, with an innovative vision: gathering all those concerned in the treatment of genetic diseases to create synergy that encourages transfer of knowledge, to speed up the discovery of new treatments and diagnoses to meet the expectations of the patients and their families.

### Solve-RD

"Solve-RD - solving the unsolved rare diseases" is a research project funded by the European Commission for five years (2018-2022). It echoes the ambitious goals set out by the International Rare Diseases Research Consortium (IRDIRC) to deliver diagnostic tests for most rare diseases by 2020. The current diagnostic and subsequent therapeutic management of rare diseases is still highly unsatisfactory for a large proportion of rare disease patients – the unsolved RD cases. For these unsolved rare diseases, we are unable to explain the etiology responsible for the disease phenotype, predict the individual disease risk and/or rate of disease progression, and/or quantitate the risk of relatives to develop the same disorder.

Solve-RD's main ambitions are thus:

- To solve large numbers of rare disease, for which a molecular cause is not known yet by sophisticated combined omics approaches, and;
- To improve diagnostics of rare disease patients through contribution to, participation in and implementation of a "genetic knowledge web" which is based on shared knowledge about genes, genomic variants and phenotypes.

## Monday March 11, 2019 - Setting the Landscape

Room: 601/602 (6<sup>th</sup> floor)

Time	Session and Trainer(s)
11:30-12:00	<b>Registration</b> <i>Aline Schnieder, Open Academy Junior Manager, EURORDIS</i>
12:00- 12:30	<b>Welcome</b> <i>Raquel Castro, Open Academy Director, Social Policy Director, EURORDIS</i> <i>Virginie Bros-Facer, Scientific Director, EURORDIS</i> <i>Stanislas Lyonnet, Director, Imagine Institute</i>
12:30-13:30	<b>Lunch</b>
13:30-14:30	<b>Ice breaking exercise</b> <i>Virginie Bros-Facer, Scientific Director, EURORDIS</i>
14:30-15:00	<b>Coffee Break</b>
15:00-16:15	<b>History in Genetics – from Watson &amp; Cricks to NGS</b> <i>Jean-Louis Mandel, Researcher, Institut de Génétique et Biologie Moléculaire et Cellulaire, Strasbourg/Fondation Maladies Rares</i>
16:15-17:30	<b>Translational Research: what, why, how, and with whom?</b> <i>Annemieke Aartsma-Rus, Professor at the Department of Human Genetics, Leiden University Medical Center</i>
19:30-21:30	<b>Welcome Dinner</b> <i>Restaurant Jardin Secret</i>

## Tuesday March 12, 2019 - Diagnosis

Room: 601/602 (6th Floor)

Time	Session and Trainer(s)
09:00-10:00	<p><b>State-of-play in Diagnosis and Solve-RD new EU project for solving the unsolved</b></p> <p><i>Holm Graessner, Coordinator of Solve-RD and Coordinator of ERN-RND, Institute of Medical Genetics &amp; Applied Genomics, University of Tuebingen</i></p>
10:00-11:00	<p><b>How to deal with results – variants – 23andme...</b></p> <p><i>Laurence Faivre, Head of French Reference Network AnDDI-Rares, Professor in Genetics, CHU Dijon Department of Medical Genetics</i></p>
11:00-11:30	<p><b>Coffee Break</b></p>
11:30-12:30	<p><b>Interactive session: Case scenario discussions on secondary findings</b></p> <p><i>Laurence Faivre, Head of French Reference Network AnDDI-Rares, Professor in Genetics, CHU Dijon Department of Medical Genetics</i></p>
12:30-13:30	<p><b>Lunch</b></p>
13:30-14:45	<p><b>Artificial Intelligence for Diagnosis Health29</b></p> <p><i>Julian Isla, Chairman and founder, Foundation 29/Chief Scientific Officer, Dravet Syndrome European Federation</i></p>
14:45-15:15	<p><b>Genetic Bingo</b></p> <p><i>Gulcin Gumus, Research and Policy Project Manager, EURORDIS</i></p>
15:15-16:00	<p><b>Visits to genetic sequencing labs and discussion with geneticist on site at Imagine Institute</b></p> <p><i>Tania Attié-Bitach, Geneticist, Hôpital Necker-Enfants Malades &amp; Giulia Barcia, Geneticist, Hôpital Necker</i></p>

## Wednesday March 13, 2019 – Use of Data

Room: 601/602 (6th Floor)

Time	Session and Trainer(s)
09:00-10:00	<b>ORPHANET what it is and how to use it</b> <i>Ana Rath, Director, ORPHANET</i>
10:00-11:00	<b>Drug repurposing as a strategy for rare disease charities</b> <i>Rick Thompson, CEO, Findacure</i>
11:00-11:30	<b>Coffee Break</b>
11:30-13:00	<b>Break-out groups: Starting down the road of repurposing for your rare condition</b> <i>Rick Thompson, CEO, Findacure</i> <i>Annemieke Aartsma-Rus, Professor at the Department of Human Genetics, Leiden University Medical Center</i> <i>Nick Sireau, Chairman of Trustees, AKU Society</i>
13:00-14:00	<b>Lunch</b>
14:00-15:30	<b>Data sharing: why and how?</b> <i>Sergi Beltran, Head of the Bioinformatics Unit, Centro Nacional de Análisis Genómico (CNAG-CRG)</i> <i>Leslie Matalonga, Clinical Genomics Specialist, Centro Nacional de Análisis Genómico (CNAG-CRG)</i>
15:30-16:30	<b>Activities in the International Rare Diseases Research Consortium (IRDiRC)</b> <i>Diego Ardigo, Project Leader of advanced therapy medicinal products (ATMP) and biologics, Chiesi Pharmaceuticals/Chair of Therapies Scientific Committee of IRDiRC</i>
16:30-17:15	<b>GENIDA – Genetics of Intellectual Disability and Autism spectrum disorders</b> <i>Jean-Louis Mandel, Researcher, Institut de Génétique et Biologie Moléculaire et Cellulaire, Strasbourg/Fondation Maladies Rares</i>
17 :15-18 :15	<b>Dr Warehouse – a biomedical data warehouse</b> <i>Nicolas Garcelon, Head of Data Science Platform, Imagine Institute, Hôpital Necker</i>

## Thursday March 14, 2019 - Research & Therapies

Room: 330 (3rd floor)

Time	Session and Trainer(s)
09:00-10:00	<b>How to get research done on your rare disease?</b> <i>Daniel Lewi, Chief Executive, CATS Foundation</i>
10:00-11:00	<b>Genome editing: a tool to advance rare disease research</b> <i>Matteo Bovolenta, Senior Researcher, Marie Curie Fellow, Généthon</i>
11:00-11:30	<b>Coffee Break (Press Room 6th Floor)</b>
11:30-12:15	<b>European Joint Programme on Rare Diseases (EJP-RD)</b> <i>Daria Julkowska, EJP RD Coordinator, Inserm</i>
12:15-12:45	<b>Q&amp;A on patient involvement in research</b> <i>Virginie Bros-Facer, Scientific Director, EURORDIS Researcher, Imagine Institute</i>
12:45- 14:00	<b>Lunch (Press Room 6th Floor)</b>
14:00-15:00	<b>Evolution of practice in animal research for the development of new therapies</b> <i>Ian Ragan, Neuropharmacologist and former Board Member, National Centre for the Replacement, Refinement and Reduction of Animals in Research</i>
15:00-17:00	<b>Background info and visit of the animal facility in three groups</b> <i>Pierre Cherel, Director, Veterinary &amp; Emilie Panafieu, Operational Manager, Veterinary, Imagine Institute, Hôpital Necker</i>

## Friday 15 March 2019 - European Reference Networks

Room: 330 (3rd floor)

Time	Session and Trainer(s)
09:00-10:00	<b>From Care to Research / From Research to Care: ERNs</b> <i>Victoria Hedley, Rare Disease Policy Manager, University of Newcastle</i>
10:00-11:00	<b>Genetic therapies, ERN Eye (ERN on eye diseases)</b> <i>Dominik Fischer, Professor of Ophthalmology, University Eye Hospital, Tuebingen</i>
11:00-11:30	<b>Coffee Break (Press Room 6th Floor)</b>
11:30-12:30	<b>ERN-ITHACA (ERN for Rare Congenital Malformations and Intellectual Disability): Diagnosis for the undiagnosed</b> <i>Jill Clayton-Smith, Consultant in Clinical Genetics and Honorary Professor in Medical Genetics, Manchester Centre for Genomic Medicine/ERN ITHACA</i>
12:30-13:00	<b>Feedback session</b> <i>Virginie Bros-Facer, Scientific Director, EURORDIS</i> <i>Raquel Castro, Open Academy Director, Social Policy Director, EURORDIS</i>
<b>End of the training programme</b>	