



Deliverable

D3.8 First summer school for ePAGs delivered

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Explanation according to GA Annex I:

Training ERN patient representatives in genome diagnostics.

Abstract:

This deliverable report covers the first edition of the EURORDIS Winter School, the capacity building training programme for rare disease patient representatives on scientific innovation and translational research, which was held at the Imagine Institute for Genetic Diseases in Paris from 19th to 23rd March 2018.

Introduction:

The EURORDIS Winter School 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 re-search translates into real benefits for rare disease patients. 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.

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 first edition of the face-to-face training of the EURORDIS Winter School was held in March 2018. 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 and included laboratory visits. 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 synergy that encourages 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

29 participants attended the first edition of the Winter School. Participation was on an invitation basis; the invitations were sent to potential participants to ensure meaningful feedback and constructive criticism in order to better tailor future editions to the needs of patients.

The trainees represented different organizations and networks, including European Patient Advocacy Group (ePAG) representatives from 6 European Reference Networks including those represented in Solve-RD (ITHACA, NMD, RND and GENTURIS), EURORDIS task forces and working groups (DITA, Genome editing), members of EURORDIS Board of Directors and representatives from the undiagnosed community (SWAN UK, SWAN Europe).

METHODOLOGY

The Winter School applied a mixed approach to deliver the training. The training programme included a face to face training with interactive plenary sessions, discussion groups and case studies. Since this was the first edition of a capacity building training on scientific innovation and translational research, the thematic sessions and the training methodology were designed according to the aforementioned objectives, to be tested and to be tailored for the future editions according to the feedback received from the participants, in order to suit trainees' needs and to ensure that the training objectives are met.

PRE-TRAINING

Trainees received reading materials about genetic diagnosis, genome editing technologies, drug repurposing and outcome measurements as well as video links on genome editing in order to maximize the effectiveness of the onsite training and familiarize participants with the key concepts and the terminology that will be used during the onsite course. In addition, the speakers were asked to define the learning objectives of their session. 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 received a preparatory webinar one month before the Winter School, in order to present the training programme in general and answer any questions that the participants might have.

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

- **Setting the landscape:** This session was devoted to familiarize 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.

The presentations and videos from the training sessions were recorded and uploaded on the EURORDIS Open Academy online platform: <https://openacademy.eurordis.org/>

CHALLENGES FOR FUTURE EDITIONS

- Ensure that there is a well-developed and comprehensive online pre-training webpage on the EURORDIS Open Academy website;
- Brief the speakers on the type of pre-training readily available, the educational approach required, the various level of knowledge of the trainees attending and overarching as well as specific training goals;
- To incorporate additional sessions for discussion and networking as well as break-out activities in smaller groups.

Conclusion:

A substantial majority of participants 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 disciplines and backgrounds. Moreover, 75 percent of the respondents reported that most of their questions were answered during the training.

Implementation of the received feedback within the next editions of the Winter school will increase active participation of trainees in the sessions, maximize their understanding of scientific concepts and increase their capacity as patient advocates therefore contributing to meaningful patient involvement in scientific research.



EURORDIS Winter School 2018
IMAGINE Institute
Hôpital Necker
Paris, France
19 to 23 March, 2018

Capacity-building programme
for patients
on scientific innovation
and translational research.

Held in cooperation with SolveRD

www.eurordis.org

Monday March 19, 2018

Setting the Landscape

12:30 13:00	Registration	Nancy Hamilton (EURORDIS)
13:00 13:30	Welcome	Yann Le Cam (EURORDIS) Stanislas Lyonnet (Institut Imagine)
13:30 14:30	Ice breaking exercise	Virginie Bros-Facer (EURORDIS)
14:30 15:00	Coffee Break	
15:00 16:00	Existing resources, funding opportunities at EC and national levels, strategies for patient groups & partnership with industry	Daria Julkowska (Agence Nationale de la Recherche France)
16:00 16:45	Patient Generated Data	Elin Haf Davies (Aparito)
20:00 22:00	Welcome Dinner L'Apollo Montparnasse Restaurant 106 boulevard du Montparnasse 75014 Paris (Métro Vavin)	

Tuesday March 20, 2018 Diagnosis

09:00 09:45	History in Genetics – from Watson & Cricks to NGS	Ségolène Aymé (ICM Brain and Spine Institute Hôpital Pitié Salpêtrière)
09:45 10:30	State-of-play in Diagnosis	Olaf Riess (Institute of Medical Genetics & Applied Genomics U of Tuebingen)
10:30 11:00	Coffee Break	
11:00 11:30	Solve RD: new EU project for solving the unsolved	Holm Graessner (Institute of Medical Genetics and Applied Genomics U. of Tuebingen)
11:30 12:30	How to deal with results – variants – 23andme...- Genetic Counselling - consent for data sharing	Laurence Faivre (CHU Dijon Department of Medical Genetics)
12:30 13:30	Lunch Break	
13:30 14:30	Case scenario discussions on secondary findings	Laurence Faivre (CHU Dijon Department of Medical Genetics)
14:30 14:50	Genetic research: new discoveries!	Sandrine Marlin (Imagine Institute)
15:15 16:00	Visits to genetic sequencing labs and discussion with geneticist on site at <i>Imagine Institute</i> (Building Lavoisier, rue de Vaugirard: see map)	Jean-Paul Bonnefont , (Hôpital Necker) Tania Attié-Bitach (Hôpital Necker)

Optional group dinner 19:30 for those who want to sign up! « Crêperie » close by.

Wednesday March 21, 2018

Data

09:00 09:45	ORPHANET what it is and how to use it.	Ana Rath (ORPHANET)
09:45 10:30	Agreeing what outcomes to measure: the importance of core outcome sets and the crucial roles that patients have in the development and use of core outcome sets	Bridget Young University of Liverpool (PoPPiE)
10:30 11:00	Coffee Break	
11:00 11:45	Drug repurposing as a strategy for rare disease charities	Rick Thompson (Findacure)
11:45 13:00	Break-out groups: Starting down the road of repurposing for your rare condition	Rick Thompson (Findacure) Annemieke Aartsma-Rus (Leiden University Medical Center) Virginie Hivert (EURORDIS)
13:00 14:00	Lunch Break	
14:00 14:45	Data sharing and analysis within and beyond RD platforms (RD-Connect, MatchMaker Exchange)	Sergi Beltran (Centre Nacional d'Anàlisi Genòmica)
14:45 16:00	Bioinformatic lab and discussion	Patrick Nitschke Nicolas Garçelon (IMAGINE Hôpital Necker)

Optional group dinner 19:30

Sign up onsite!

Thursday March 22, 2018

Diagnosis and Therapies

09:00 09:45	Evolution of practice in animal research for the development of new therapies	Ian Ragan (National Centre for the Replacement, Refinement and Reduction of Animals in Research)
09:45 10:30	Translational Research: what, why, how, and with whom?	Annemieke Aartsma-Rus (Leiden University Medical Center)
10:30 11:00	Coffee Break	
11:00 11:45	Gene Therapy MPS IIIB Clinical Trials at Pasteur Institute	Marc Tardieu (Université Paris Sud et Hôpitaux universitaires Paris Sud)
11:45 12:30	Gene/Advanced Therapies on the market	Diego Ardigo (Chiesi Pharmaceuticals)
12:30 13:30	Lunch Break	
13:30 14:30	Strategy of Patient Organisations in driving research from the lab to therapies.	Ana Mingorance (Madrid)
14:30 16:00	Preclinical studies in a murine model and visit of an animal facility in three groups.	Catherine Caillaud Emilie Panafieu (IMAGINE Hôpital Necker)

Optional group dinner 19:30

Sign up onsite!

Friday March 23, 2018

European Reference Networks

09:00 09:45	Genetic therapies: from AAV to exon skipping and the CRISPR revolution	Dominik Fischer (University Eyeclinics, Tuebingen)
09:45 10:30	From Care to Research / From Research to Care: ERNs	Vicky Hedley (University of Newcastle)
10:30 11:00	Coffee Break	
11:00 11:45	ERN-ITHACA: Diagnosis for the undiagnosed	Jill Clayton-Smith (Manchester Centre for Genomic Medicine)
11:45 12:30	Feedback session & Networking	Virginie Bros-Facer (EURORDIS) Nancy Hamilton (EURORDIS)
12:30	End of programme	