



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 Solve<sup>RD</sup>

[www.eurordis.org](http://www.eurordis.org)

# Monday March 19, 2018

## Setting the Landscape

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

## Tuesday March 20, 2018 Diagnosis

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

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

# Wednesday March 21, 2018

## Data

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

**Optional group dinner 19:30**  
**Sign up onsite!**

**Thursday March 22, 2018**

**Diagnosis and Therapies**

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

**Optional group dinner 19:30**

**Sign up onsite!**

# Friday March 23, 2018

## European Reference Networks

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