EJP RD
General Assembly and Consortium meeting 2020
Online
September 14th – 18th

Preliminary program
Program at a glance:

**September 14**
- 10:00 - 12:15 (including break) Plenary GA Session: Welcome, achievements of EJP RD at M18, Annual Work Plan Year 3, Collaboration with external stakeholders, monitoring indicators. Update from coordination, updates from VPA, EJP RD, and RDRRC, ERICA, New training needs for RD community.

**September 15**
- 9:00 - 10:30 Experimental data resources available in the EJP RD
- 11:10 - 13:00 Introduction to translational research for clinical or pre-clinical researchers
- 14:00 - 18:00 (including break(s)) Funding opportunities in EJP RD & How to build a successful proposal

**September 16**
- 9:00 - 10:30 RD Connect Genome-Phenome Analysis Platform
- 10:40 - 13:00 (including break) Improvement and innovation in the design and analysis of rare diseases clinical trials
- 14:00 - 16:00 (including break) FAIRifying registries and databases: How EJP RD can help?
- 16:00 - 17:30 Solutions to regulatory and data protection issues

**September 17**
- 9:00 - 13:00 (including break(s)) Finding and accessing relevant resources (including pre-clinical and clinical ones) for your research needs (special session including PI funded researchers)
- 13:00 - 16:00 Applications of the EJP RD Virtual Platform: Counting cases and more & Finding key patients and data
- 16:30 - 18:00 Plenary closing session

18-sept
- "Speed meeting" Multinational Clinical Trial Support Office
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Program per day

SEPTEMBER 14th

9:00 - 9:30
9:30 - 10:00
10:00 - 10:30
10:30 - 11:00
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18:00 - 18:30
18:30 - 19:00
19:00 - 19:30
19:30 - 20:00

Morning session

10:00 - 13:15 (including break)

Plenary GA session
Welcome, achievements of EJP RD at M18, Annual Work Plan Year 3, Collaboration with external stakeholders, Monitoring indicators, Update from coordination, Update from WP4, EJP RD and IRDRC, ERICA, New training needs for RD community

Lunch

Afternoon session

14:00 - 15:00
14:00 - 18:00 (including break(s))
14:00 - 18:00 (including break(s))
14:00 - 18:00 (including break(s))

Parallel Session P1
Parallel Session P2
Parallel Session P3
Parallel Session P4

Evening session
EJP RD social event
SEPTMBER 15th

9:00 - 9:30
9:30 - 10:00
10:00 - 10:30
10:30 - 11:00
11:00 - 11:30
11:30 - 12:00
12:00 - 12:30
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14:30 - 15:00
15:00 - 15:30
15:30 - 16:00
16:00 - 16:30
16:30 - 17:00
17:00 - 17:30
17:30 - 18:00

Morning session

9:00 - 10:50
Experimental data resources available in the EJP RD

10:50 - 11:10
20 min Break

11:10 - 13:00
Introduction to translational research for clinical or pre-clinical researchers

Afternoon session

14:00 - 18:00 (including break(s))
Funding opportunities in EJP RD & How to build a successful proposal
SEPTEMBER 16th

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<td>9:00 - 10:30 RD-Connect Genome-Phenome Analysis Platform</td>
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<td>9:30 - 10:00</td>
<td>10:00 - 10:30 10 min Break</td>
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<tr>
<td>10:00 - 10:30</td>
<td>11:30 - 12:00 Improvement and innovation in the design and analysis of rare diseases clinical trials</td>
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<td>10:30 - 11:00</td>
<td>12:00 - 12:30 FAIRifying registries and databases: How EJP RD can help?</td>
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<td>16:00 - 16:30</td>
<td>16:00 - 17:30 Solutions to regulatory and data protection issues</td>
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<td>17:00 - 17:30</td>
<td>Lunch</td>
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Morning session

Afternoon session
SEPTEMBER 17th

Morning session

9:00 - 9:30
Morning session

9:30 - 10:00
Morning session

10:00 - 10:30
Morning session

11:30 - 12:00
Morning session

12:00 - 12:30
Morning session

12:30 - 13:00
Morning session

SEPTEMBER 18th

Morning session

10:00 - 10:30
Morning session

10:30 - 11:00
Morning session

11:00 - 11:30
Morning session

11:30 - 12:00
Morning session

12:00 - 12:30
Morning session

"Speed-meeting" Multinational Clinical Trial Support Office
15 min 1to1 meeting registration in advance of the meeting

September 17

9:00 - 13:00 (including break(s))
Finding and accessing relevant resources (including pre-clinical and clinical ones) for your research needs (special session including P1 funded researchers)

13:50 - 16:20
Applications of the EJP RD Virtual Platform: Counting cases and more & Finding key patients and data

13:40 - 14:40
Outcomes of ERN trainings and fellowships support

14:50 - 16:20
Rare diseases e-learning scheme of EJP RD

16:30 - 18:00
Plenary closing session to consider: debating parallel sessions 30 min
Plenary Session

Chair/co-chair:
Daria Julkowska, Inserm, coordinator of the EJP RD

Speakers
Daria Julkowska (Coo, INSERM), Blandine Castrillo (Coo, INSERM), Kejla Musaraj (CVBF), Viviana Giannuzzi (FGB), Stefano Benvenuti (FTELE), Carla d’Angelo (Coo, INSERM), Galliano Zanello (Coo, INSERM), Alberto Pereira (LUMC), Biruté Tumiene (VUHSK)

Objectives
- Update from coo and partners on EJP RD progress: achievements, monitoring, lessons learned from reporting
- Short presentation of the Annual Work Plan for year 3 and collaborations initiated by EJP RD with external stakeholders
- Annual state of the art on ethics, regulatory and legal aspects
- Update from the International Rare Diseases Research Consortium
- Short presentation of ERICA - the coordination and support action of ERNs
- Introduction to the discussion on new training needs for rare diseases community

Description
This opening plenary session will help all participants to dive in EJP RD activities. The 18 months of EJP RD achievements will be shortly presented and lessons learned from monitoring and reporting actions shared with EJP RD members. Even though the official vote of the Annual Work Plan for year 3 will take place in the week after the EJP RD GA, the summary of the activities planned and major changes will be presented. In addition, the coordination will feature new collaborations with external stakeholders initiated by EJP RD. As every year an update on IRDiRC, ERNs and ethical/regulatory/legal state of the art will be presented. Finally, the session will terminate with short introduction to start the discussion on the new training needs for rare diseases community, which is the goal of work package 18.

Program of the session

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<th>Speaker</th>
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<td>10:00 – 10:15</td>
<td>Welcome from the Coordination, general objectives of the meeting, EJP RD achievements at M18</td>
<td>Daria Julkowska (Inserm, Coo)</td>
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<tr>
<td>10:15 – 10:35</td>
<td>Annual Work Plan Year 3: - Major changes in the Pillars - Budget changes</td>
<td>Daria Julkowska, Blandine Castrillo (Inserm)</td>
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<tr>
<td>10:35 – 11:00</td>
<td>Collaboration with external stakeholders</td>
<td>Daria Julkowska (Inserm)</td>
</tr>
<tr>
<td>11:00 – 11:15</td>
<td>Monitoring indicators: identification of additional global indicators</td>
<td>Kejla Musaraj (tbc)</td>
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<tr>
<td>11:15 – 11:30</td>
<td>Update from coordination (lessons learned from reporting, etc.)</td>
<td>Blandine Castrillo (Inserm)</td>
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<tr>
<td>Time</td>
<td>Session</td>
<td>Presenter(s)</td>
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<tr>
<td>11:30 – 11:40</td>
<td>Break</td>
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<tr>
<td>11:40 – 12:00</td>
<td>Update and presentation of the state of the art on ethical, legal and regulatory issues relevant for RD community</td>
<td>Viviana Giannuzzi (FGB), Stefano Benvenuti (FTELE) (tbc)</td>
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<tr>
<td>12:00 – 12:15</td>
<td>EJP RD and IRDiRC – update on Task Forces and joint activities</td>
<td>Carla d'Angelo, Galliano Zanello (Inserm)</td>
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<tr>
<td>12:15 – 12:30</td>
<td>ERICA – coordination and support action for ERN research strategy</td>
<td>Alberto Pereira (tbc)</td>
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<tr>
<td>12:30 – 13:15</td>
<td>New training needs for rare diseases community</td>
<td>Birute Tumiene (tbc)</td>
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Parallel Session Pillar 1

Chairs
Ralph Schuster, Sonja van Weely (Pillar 1 leaders)

Program of the session

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<td>WP6 JTC 2020 update (closed session, funders only)</td>
<td>Florence Guillot, ANR</td>
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<td>15:00 – 15:30</td>
<td>WP6 Update</td>
<td>Ralph Schuster, DLR</td>
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<td>15:30 – 16:00</td>
<td>WP7 Update</td>
<td>Sonja van Weely, ZonMw</td>
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<td>16:00 – 16:15</td>
<td>Coffee break</td>
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<td>16:15 – 16:45</td>
<td>WP8 Update</td>
<td>Christine Fetro, FFRD</td>
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<td>16:45 – 17:15</td>
<td>WP9 Update</td>
<td>Irit Allon, CSO/MOH</td>
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<td>17:15 – 17:45</td>
<td>WP19 translation/mentoring service update</td>
<td>Anton Ussi, EATRIS; Elena Bertrami, Telethon Italy - tbc</td>
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<td>17:45 – 18:00</td>
<td>AOB</td>
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Parallel Session Pillar 2

Chairs
Ana Rath, Franz Schaeffer (Pillar 2 leaders)

Objectives
- Pillar 2 Technical and non-technical Master Plans Finalization
- FAIRification Stewards report and next steps (progress, identified issues)
- Query Builder progress, planned tests and implementation options
- Address the Virtual Platform Non-Functional Requirements

Description
Pillar 2 Clarity Afternoon where:
- Master Plans as activity planning and visual progress reporting tools will be finalized for both the technical development of the alpha version of the virtual platform and the non-technical development (non-software development)
- First specifications relating to Quality, GDPR, Sustainability & Standards will be presented and endorsed
- Organization and Alignment relating to how some activity and resources should be performed and used
- Reports on FAIRification and Query Builder activity eliciting progress and planned actions with the options foreseen for implementation as well as reporting issues

Speakers
Michael Nitznader : (technical Master Plan)
Bruna Dos Santos Vieira : FAIRification report
Work Foci Leaders (Non-Technical Master Plan)

Program of the session

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<td>14:00 – 14:20</td>
<td>FAIRification Stewards report and next steps (progress, identified issues)</td>
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<td>14:20 – 14:40</td>
<td>Discussion</td>
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<tr>
<td>14:40 – 15:00</td>
<td>Query Builder progress, planned tests and implementation options</td>
</tr>
<tr>
<td>15:00 – 15:20</td>
<td>Discussion</td>
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<tr>
<td>15:20 – 15:35</td>
<td>Break</td>
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<tr>
<td>15:35 – 16:00</td>
<td>Organization and Alignment relating to how some activity and resources should be performed and used</td>
</tr>
<tr>
<td>16:00 – 16:55</td>
<td>Pillar 2 Technical and non-technical Master Plans Finalization</td>
</tr>
<tr>
<td>16:55 – 17:05</td>
<td>Break</td>
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<tr>
<td>17:05 – 18:00</td>
<td>First specifications relating to Quality, GDPR, Sustainability &amp; Standards will be presented and endorsed</td>
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Parallel Session Pillar 3

Chairs
Virginie Bros-Facer & Birute Tumiene (Pillar 3 Leaders)

Speakers
WP14: Claudio Carta, Marie Verrey/Sylvie Maiella, Gert Matthijs/Liliane Geyskens, Mary Wang
WP15: Raquel Castro, Mariangela Lupo
WP16: Roseline Favresse
WP17: Holm Graessner/Sanja Hermanns
WP18: Birute Tumiene, Krystyna Chrzanowska, Virginie Bros-Facer

Objectives
This session aims to provide 1) a summary of all training activities in 2020; 2) Highlights of plans for 2021 and 3) discussion on recent improvements and remaining challenges. Finally, this session will strive to increase visibility of tools and resources helpful for the organisation of the training courses and will end with an open discussion on different ways training organisers can support each other moving forward to ensure better efficiency and cohesion between the training activities on offer in Pillar 3.

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<td>14:00 – 14:30</td>
<td>WP14 updates (5min per task)</td>
<td>respective task leaders</td>
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<td>14:30 – 14:50</td>
<td>Interactive discussion</td>
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<tr>
<td>14:50 – 15:15</td>
<td>WP15 updates (5min for 15.1; 15.2 and 15.3 and 10min for 15.4)</td>
<td>respective task leaders</td>
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<td>15:15 – 15:30</td>
<td>Interactive discussion</td>
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<td>15:30 – 16:00</td>
<td>Break</td>
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<td>16:00 – 16:15</td>
<td>WP16 updates</td>
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<td>16:15 – 16:25</td>
<td>Interactive discussion</td>
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<td>16:25 – 16:35</td>
<td>WP17 updates</td>
<td>WP leader</td>
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<td>16:35 – 16:45</td>
<td>Interactive discussion</td>
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<td>16:45 – 17:15</td>
<td>WP18 plans including interactive discussion</td>
<td>WP leader</td>
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<tr>
<td>17:15 – 18:00</td>
<td>Toolkit updates and open discussion on how to support each other for organisation of training courses</td>
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Parallel Session Pillar 4

Description
This session aims to provide for Pillar 4:
• A summary of all activities in 2020;
• Highlights of plans for 2021 and beyond;
• Discussion of interactions with Pillar 3 for training opportunities;
• Discussion of interactions with Pillar 1 for integrated working with funded projects;
• Opportunity for all participants to give input for P4 strategy and integration with other Pillars.

Program of the session

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<td>14:00 – 14:15</td>
<td>P4 Overview from Co-Chairs</td>
<td>Rima Nabbout and Anton Ussi</td>
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<td>14:15 – 15:00</td>
<td>Training opportunities (interactions with P3)</td>
<td>Speaker TBC</td>
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<td>15:00 – 15:45</td>
<td>WP19 Update (Tasks by Task Leaders)</td>
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<td>15:45 – 16:00</td>
<td>Coffee break</td>
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<td>16:00 – 16:45</td>
<td>WP20 Update (Tasks by Task Leaders)</td>
<td>Task Leaders</td>
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<td>16:45 – 17:15</td>
<td>Interactive Discussion - P4 strategy and integration with other Pillars</td>
<td>All</td>
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<tr>
<td>17:15 – 17:45</td>
<td>Interactions with P1 (integrate working with funded projects)</td>
<td>Speaker TBC</td>
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<td>17:45 – 18:00</td>
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Speakers
Rima Nabbout is Professor of Paediatric Neurology at Paris Descartes University and Director of the French center for Rare Epilepsies at Necker Enfants Malades, Imagine Institute (INSERM U1136), Paris, France. She received her medical degree from Saint Joseph University, Beirut, Lebanon; her paediatric board from Descartes University, Paris; and a PhD in Neurosciences from University Pierre et Marie Curie, Paris, France. She is a member of the steering committee of EPICARE (European reference network on rare and complex epilepsies), of 3 task force groups of ILAE (Nosology TF, transition TF and the regulatory affairs TF), of the EJP-RD program and of scientific committees of patient’s groups on rare epilepsies. Pr Nabbout areas of research include electro clinical delineation of childhood epilepsies, guidelines on epilepsy syndromes nosology and transition from childhood into adulthood, causes and mechanisms of rare epilepsies, orphan drugs trials for rare epilepsies with development of patients’ centered end points and innovative methodologies. She
has authored more than 190 peer-reviewed papers and received H2020 and FP7 grants.

**Anton Ussi** is Operations & Finance Director at EATRIS ERIC, the European infrastructure for translational medicine. Joining EATRIS in 2010 as Head of Operations, he was co-responsible for the operational design and statutory incorporation of the infrastructure, and has been in his current role since 2015. With a background in mechanical engineering in the automotive industry, small business administration, and later in technology transfer focused on molecular imaging, Ussi specialises in public private collaboration and academic translational research in medicine. Ussi is co-leader of Pillar 4 of the EJPRD and WP 3 & 19 co-leader.
Experimental data resources available in the EJP RD

Objectives
The audience would:
- know which resources are available in the EJP RD to deposit and/or analyse their experimental data (e.g. genomes, exomes, metabolomes, etc.)
- find other data for their research.
- and learn how to find these resources and how to propose new features and development to meet their needs.

Description
Interactive session to present the available data resources, how they can be exploited to address the needs of the RD community and how to find them.
Introduction to translational research for clinical or pre-clinical researchers

Chair/Co-Chair
Anton Ussi, Toni Andreu

Objectives
Capacity building, helping researchers go from unknown unknowns to known unknowns with regards to developing their new findings towards the clinic, particularly for early- or late-career, pre-clinical and clinical researchers with little or no experience in therapy development.
Indicating what are the help and resources available, how they can and do facilitate the process, and how to find and access them.

Description
Workshop dedicated to introducing to participants how a new therapy is developed. What has to be done to turn a new finding into a high potential therapy? What/who are the main resources available (within and external to EJP RD) that are available to support them?

Program of the session

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<td>Case Study on “Crossing the Valley of Death”</td>
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<td>11:30 – 11:40</td>
<td>Questions to Audience</td>
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<td>11:40 – 11:45</td>
<td>Introductory remarks</td>
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<td>11:45 – 12:00</td>
<td>‘The Development Process at a Glance’</td>
<td>Bernd Eisele</td>
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<td>12:00 – 12:15</td>
<td>‘Navigating the Regulatory Maze’</td>
<td>Giovanni Migliaccio</td>
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<td>12:15 – 12:30</td>
<td>‘Patents – A Necessary Evil’</td>
<td>Anton Ussi</td>
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<td>12:30 – 13:00</td>
<td>Dialogue with Audience</td>
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### Funding opportunities in EJP RD & How to build a successful proposal

**Chair/co-chair**
Ralph Schuster, DLR  
Florence Guillot, ANR

**Objectives**
- Improve understanding of ongoing EJP RD funding mechanisms  
- Support the community to increase quality of applications  

**Description**
What are the funding opportunities in EJP RD? How to write a successful grant application for EJP RD transnational calls? How are applications reviewed? How to involve patients in research project?  
The workshop includes presentations from funders, scientific evaluators, patient representatives and lessons learned from successful applicants to improve understanding of ongoing EJP RD funding mechanisms and support the community to increase the quality of applications.

**Program of the session**

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<th>Time</th>
<th>Session 1 – Introduction and scientific evaluation process</th>
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</table>
| 14:00 – 14:15 | Welcome to the session & Introduction to EJP RD / E-Rare calls  
Ralph Schuster, DLR & Florence Guillot, ANR  |
| 14:15 – 14:25 | Introduction to the evaluation process  
Ralph Schuster, DLR  |
| 14:25 – 14:40 | Do’s and Don’t’s - Recommendations from Scientific Evaluation Committee chairs  
Jacques Beckmann, Lausanne & Orly Elpeleg, Jerusalem  |
| 14:40 – 14:50 | Statistical study design  
Armin Gemperli, Lucerne  |
| 14:50 – 15:10 | Q&A Session 1  |

**Session 2 – Transversal aspects of the evaluation**

<table>
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<tr>
<th>Time</th>
<th>Topic</th>
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| 15:10 – 15:25 | Translational applicability  
Anton Ussi, EATRIS  |
| 15:25 – 15:40 | How to facilitate patient engagement / involvement  
Virginie Bros-Facer, EURORDIS  |
| 15:40 – 15:50 | Ethics review  
Ralph Schuster, DLR  |
| 15:50 – 16:10 | Q&A Session 2  |
| 16:10 – 16:30 | Coffee break  |

**Session 3 – Lessons learned and success stories from funded applications**

<table>
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<tr>
<th>Time</th>
<th>Topic</th>
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</table>
| 16:30 – 16:40 | GENOMIT  
Holger Prokisch, Munich  |
16:40 – 16:50  MuTaEB  Cristina Has, Freiburg
16:50 – 17.00  MYOCITY  Fabien Le Grand, Lyon
17:00 – 17.10  Q&A Session 3

17:10 – 17:35  
**EJP RD Joint Transnational Call 2021 - Social sciences and Humanities Research to improve health care implementation and everyday life of people living with a rare disease (Diana Desir-Parseille, FFRD)**

17:35 – 18:00  
**Network support scheme (Sonja van Weely, ZonMw)**

**Speakers**

**Dr. Ralph Schuster** received his PhD in 2000 at the University of Giessen after studying biology in the field of molecular virology. Since 2001 he works at the DLR project management agency as a research program manager. Since then he has been involved in the planning and implementation of various research funding programs of the German Ministry for Education and Research (BMBF), including rehabilitation, genomics and research into rare diseases. Since 2004 he has been the research program coordinator of the National Research Funding Program on Rare Diseases. He was involved in the development of the European research funding network for rare diseases E-Rare, which was founded in 2006, and coordinates the German efforts in this network. In the current European Joint Program on Rare Diseases (EJP RD), he is co-coordinator of the Pillar 1 research funding activities.

**Dr Florence Guillot** holds a PhD in Neurosciences from the King’s College of London where she studied axonal transport mechanisms in Charcot Marie Tooth disease. She also performed a Marie Skłodowska fellowship at GlaxoSmithKline (UK) where she studied the effect of pharmacological drug repositioned/repurposed in neurodegenerative disease models. She then worked as a post-doc researcher for University Paris-Sud on Alzheimer’s disease. She also worked as consultant for an IP firm before becoming a scientific officer in different European research programmes. She is currently working at ANR (the French National Agency for Research) where she is the coordinator of the ERA-Net cofund E-Rare-3 and is involved as partner of EJP RD.

**Prof. Jacques S. Beckmann** was, from 2013 till 2016, head of clinical bioinformatics at the SIB Swiss Institute of Bioinformatics. From 2003-2012, he was Professor of Human Genetics and Director of the Dept. of Medical Genetics at the Univ. of Lausanne (UNIL) and of the Medical Genetics Service of the Centre Hospitalier Universitaire Vaudois (CHUV). His recent research interests included genomic disorders as well as the genetic basis of complex traits. He is or has been a board member or chairman of many evaluation committees including the Italian Telethon, INRA, E-Rare / EJPRD, La Fondation des Maladies Rares and the Jérôme Lejeune Foundation.

**Prof. Orly Elpeleg** is the Director of the Department of Genetics at the Hadassah Medical Center in Jerusalem. She is a Professor of Pediatrics in the Faculty of Medicine, Hebrew University, Jerusalem, in the same place where she obtained her M.D. degree. Prof. Elpeleg completed a Pediatric residency in Jerusalem, followed by a research fellowship at the Royal Hospital in Copenhagen, Denmark. She had spent her sabbaticals at the Genetic
Prof. Armin Gemperli holds a master’s degree in mathematical statistics from the University of Bern and a PhD in biostatistics from the University of Basel. He did research in spatial disease modeling as a postdoc at the Johns Hopkins University, Baltimore, and in environmental and genetic epidemiology at the University of Basel. From 2008 to 2012 he worked as statistician and group leader in clinical trials, first at a clinical research organization in Basel, later at the clinical trials unit of the University of Bern. Since 2012 he is professor in health sciences at the University of Lucerne, Switzerland, where he leads the research unit in health sciences research. Since 2012 Armin Gemperli is also senior researcher and group leader at Swiss Paraplegic Research, Nottwil, Switzerland, an extra-university, state-supported research organization dedicated to spinal cord injury research.

Anton Ussi MSc is Operations & Finance Director at EATRIS ERIC, the European infrastructure for translational medicine. Joining EATRIS in 2010, he was co-responsible for the operational design and statutory incorporation of the infrastructure, and has been in his current role since 2015. With a background in mechanical engineering in the automotive industry, small business administration, and later in technology transfer focused on molecular imaging, Ussi specialises in public private collaboration and deployment of infrastructure for translational research in medicine.

Diana Desir-Parseille has an academic research background with a Master degree in Biochemistry and Molecular Biology, from the Pierre et Marie Curie University, Paris. She has been working in the field of rare diseases for almost 15 years, first as a research engineer for the European Network for the study of Orphan Nephropathies and for the French Foundation for rare diseases since its creation in 2012. Acting as a research administration manager, she implements and co-coordinates the Foundation’s calls for proposals both in Biomedical Sciences and in Social Sciences and Humanities. She is involved in every step of the calls for proposals, from the topics’ definition to the financial support and monitoring of the funded projects. For the EJPRD project, she participates in WP8 and in WP6, both dedicated to research support and funding.

Dr. Sonja van Weely worked for more than 10 years as scientist in the field of biochemistry of rare diseases. Thereafter she became scientific officer of the multidisciplinary Dutch Steering Committee on Orphan Drugs that was installed by the Minister of Health (2001-2011) that e.g. initiated the national plan on rare diseases. Subsequently, she was appointed as senior programme officer dedicated to research and networking programmes on rare diseases and orphan drugs at ZonMw, the Netherlands Organisation for Health Research and Development (2011-now). As representative of ZonMw she is involved in E-Rare from 2006 and coordinates the Dutch efforts in this network. She represents ZonMw in the International Rare Disease Research Consortium (IRDiRC) from 2011. Sonja is co-pillar 1 leader and work package leader of the Networking Support Scheme in the EJP RD.
**RD-Connect Genome-Phenome Analysis Platform**

**Chair**

**Sergi Beltran:** Sergi Beltran holds a PhD in Biology and is the Head of the Bioinformatics Unit at the National Center of Genomic Analysis in Barcelona (CNAG-CRG) since 2012. Sergi’s group is devoted to the development and operation of sequencing data analysis and management tools and pipelines. The group collaborates with several national and international projects, mostly related to human health. Specifically on Rare Diseases, he leads the RD-Connect platform development (platform.rd-connect.eu) and is a partner in Solve-RD (www.solve-rd.eu), EJP-RD (www.ejprarediseases.org), URD-Cat (www.urdcat.cat), the ELIXIR Rare Disease Community and MatchMaker Exchange (www.matchmakerexchange.org). Sergi also collaborates with the Navarra 1000 Genomes project (www.nagen1000navarra.es), GA4GH and the IRDiRC Diagnostics Scientific Committee.

**Objectives**

In this workshop the audience will learn to:
- Filter and prioritize variants (SNVs, indels and CNVs) using common annotations and on-the-fly gene panels associated to diseases, phenotypes and pathways (OMIM, ORDO, HPO, PanelApp, DisGeNET, Mendelian.co and Reactome).
- Interpret genomic variants according to ACMG guidelines using integrated tools such as Exomiser/Genomiser, ClinVar, Varsome, Intervar, etc.
- Define ad hoc phenotypically related cohorts for gene discovery.
- Identify similar cases through MatchMaker Exchange (MME) and variants identified in other resources through GA4GH Beacon.

**Description**

Hands-on workshop to train participants in the usage of the system to diagnose difficult RD cases with exome/genome data. We would also like to engage with ERNs to understand how we can support them (e.g. developing templates to facilitate the capture of their phenotypic/clinical data for analysis of genomes). The key concepts for variant filtering and prioritization will be introduced. The audience will then be challenged to apply these concepts to solve rare disease cases using the online RD-Connect GPAP.

**Program of the session**

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
<th>Presenter(s)</th>
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<tbody>
<tr>
<td>9:00 – 9:15</td>
<td>Introduction to the RD Connect GPAP</td>
<td>Sergi Beltran</td>
</tr>
<tr>
<td>9:15 – 10:15</td>
<td>Hands on Workshop</td>
<td>Leslie Matalonga, Steven Laurie</td>
</tr>
<tr>
<td>10:15 – 10:30</td>
<td>Discussion</td>
<td>Sergi Beltran</td>
</tr>
</tbody>
</table>
Speakers

Leslie Matalonga: Leslie Matalonga obtained a PhD in Biomedicine at the Hospital Clínic de Barcelona. Leslie is the Clinical Genomics Specialist of the CNAG-CRG Bioinformatics Unit. She has been key in coordinating the RD ELIXIR community and the EXCELERATE RD Use Case. Currently she is very much involved with H2020 Solve-RD, where she is leading on exome re-analysis through APIs. She is an everyday user of the GPAP to diagnose RD cases and coordinates many of the new features to be added. She participates in the NAGEN 1000G project, among others. Leslie is an author of 16 international publications.

Steven Laurie: Steven Laurie is a Bioinformatician with a PhD in Biomedicine. He has authored 13 peer-reviewed publications. Steve has implemented the genomics data analysis pipeline used in the RD-Connect GPAP and other customised versions of the system. He is currently responsible for co-ordinating the submission, processing, analysis, and return of results for 1000s of data sets which are being submitted to the GPAP via the SolveRD and EJP-RD projects. Independently he is also leading the CNV benchmarking and analysis working Group within SolveRD, and is involved in similar activities for ELIXIR and TransBionet. Steve is in direct contact with many of the RD-Connect GPAP users.
Improvement and innovation in the design and analysis of rare diseases clinical trials

Chairs
Rima Nabbout and Ralf-Dieter Hilgers

Objectives
Improvement of knowledge and value of novel methodologies for CTs and collection of ideas for innovation call

Description
During the last 5 years, three unique EU funded projects asterix, IDeAl, and InSPIRe, developed innovative statistical methodologies to improve the design and analysis of small population clinical trials (CT) aimed at efficient evaluation of novel therapies useful in rare diseases research.

At present trials are often performed with standard classical methodologies not adapted to the heterogeneity and small number of RD patients resulting in a loss of power to show positive effects. That is why EJP RD implemented a dedicated call for DEMONSTRATION projects aiming to show the usability and applicability of these innovative statistical methodologies for clinical trials in rare diseases. The goal of the demonstration projects is to re-evaluate data (from previous CTs) that lacked efficiency because it was analysed with classical statistical methodology, which might be not feasible for trials in the rare disease context.

The proposed workshop will introduce where we stand today with CTs in rare diseases and identify hurdles for which the clinical trial support office is available to help. We will present a few examples of some of the obstacles with respect to methodological issues and how the funded Demonstration projects are aiming to overcome. These novel methodologies as well as present the concepts proposed in the funded projects. We will also tackle the challenge of the urgent need of further innovation in methodologies applied to RD clinical trials. The proposed ideas during this discussion will help to finalize the innovation topics for the coming EJP RD internal call that will foster innovation projects and shall open before the end of 2021. Furthermore, aspects of the work of the clinical trial support office included in the toolbox and the planned educational program on advanced methodology in rare disease clinical trials will be discussed.

Program

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<tr>
<th>Time</th>
<th>Session</th>
<th>Presenter</th>
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<tr>
<td>10:40 – 10:50</td>
<td>Introduction to aims of WP20 activities</td>
<td>R. Nabbout</td>
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<tr>
<td>10:50 – 11:00</td>
<td>Overview of the clinical trials support office</td>
<td>M. d.Alamo</td>
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<tr>
<td>11:00 – 11:25</td>
<td>Presentation with hurdles/obstacles and methodological solutions with respect also to funded Demonstration Projects (Ideal, Asterix, Inspire)</td>
<td>R.-D. Hilgers</td>
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<tr>
<td>Time</td>
<td>Event</td>
<td>Speaker(s)</td>
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<tr>
<td>11:25</td>
<td>Question and Answers (Open discussion about methodologies)</td>
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<tr>
<td>11:45</td>
<td>Break</td>
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<tr>
<td>11:50</td>
<td>Discussion about Innovation Projects (completely interactive roundtable)</td>
<td>ECRIN (M. D.Alamo and Jacques Demotes), RD Hilgers, Statistician from IDEAL, R. Nabbout, clinician/Pharmacology person</td>
</tr>
<tr>
<td>12:50</td>
<td>Toolbox and educational program for clinical trials in rare diseases</td>
<td>M. d.Alamo, L. Sangiorgi</td>
</tr>
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</table>

**Speakers**

**Rima Nabbout** is Professor of Paediatric Neurology at Paris Descartes University and Director of the French center for Rare Epilepsies at Necker Enfants Malades, Imagine Institute (INSERM U1136), Paris, France. She received her medical degree from Saint Joseph University, Beirut, Lebanon; her paediatric board from Descartes University, Paris; and a PhD in Neurosciences from University Pierre et Marie Curie, Paris, France. She is a member of the steering committee of EPICARE (European reference network on rare and complex epilepsies), of 3 task force groups of ILAE (Nosology TF, transition TF and the regulatory affairs TF), of the EJP-RD program and of scientific committees of patient’s groups on rare epilepsies. Pr Nabbout areas of research include electroclinical delineation of childhood epilepsies, guidelines on epilepsy syndromes nosology and transition from childhood into adulthood, causes and mechanisms of rare epilepsies, orphan drugs trials for rare epilepsies with development of patients’ centered end points and innovative methodologies. She has authored more than 190 peer-reviewed papers and received H2020 and FP7 grants.

**Ralf-Dieter Hilgers** studied mathematics at RWTH Aachen University. He finished his doctoral thesis at the statistical faculty of the University of Dortmund in 1991. In 2000 he received the Venia Legendi for Medical Statistics at the University of Cologne. Since 2001 he is Professor of Biometrie and Chair of the Department of Medical Statistics at the Medical Faculty, RWTH Aachen University. His research interest is in optimal design of experiments, randomizations procedure and clinical trials. Since 1987 he gives biostatistical advice to clinical and experimental trials in all clinical and preclinical areas. Professor Hilgers teaches 300 students in different bio-scientific areas per year and is responsible for the education of investigators in clinical trials. He also acts as reviewer for methodological and clinical journals with main focus on surgical trials. He was coordinator of the “Integrated Design and Analysis of Small Population Clinical Trials (IDEAl)” project funded by the European Union’s Seventh Framework Programme for research, technological development and demonstration from 2013-2017, which established new methodologies for small population group trials.
Luca Sangiorgi, Head of Department of Rare Skeletal Disorders, is the coordinator of the Rare Diseases Centre of IOR since 2006 (when it was established). He’s been nominated as the Italian Government Delegate for the European Research Infrastructure BBMRI working on Rare Disease Biobanks. From March 1st, 2017, is Coordinator of the European Reference Network on Rare Bone Disorders (BOND ERN).

Jacques Demotes MD, PhD, MBA is Director General of ECRIN and Professor of cell biology, clinical neurologist. Dr Demotes was the coordinator of ECRIN’s four FP6- and FP7-funded projects, which supported the sustainable development of ECRIN as a pan-European infrastructure with “ERIC” status (awarded in 2013). He has participated in more than 60 FP6, FP7, H2020 and IMI-funded projects.

Marta del Alamo is clinical project manager at ECRIN, coordinating ongoing multinational clinical trials and providing support to clinical investigators preparing clinical studies/projects for European funding. She holds a PhD in Molecular Biology and a post-graduate degree in Clinical Trials Management. As a previous research scientist in the field of molecular biology and virology, she has worked as post-doctoral scientist at research centers in Spain and USA, authoring 11 peer-reviewed publications.

Mark Turner is Professor of Neonatology and Research Delivery at the University of Liverpool, UK. He has been instrumental in setting up five international paediatric research collaborations involving academia, regulators, industry, and families. He co-leads the largest ever European public-private partnership in paediatric research (€140million Innovative Medicines Initiative 2 project, “conect4children”, c4c). He has contributed to publicly funded and industry evaluations of 16 active pharmaceutical ingredients and 3 excipients. He is European Co-Director of the International Neonatal Consortium and President of the European Society of Developmental Perinatal and Paediatric Pharmacology.
September 16th, 14:00 – 15:30

**FAIRifying registries and databases: How EJP RD can help?**

**Objectives**
- to help EJP RD stakeholders adopt and shape the interoperability considerations and enroll in EJP RD collaboration structures in order to FAIRify their resources to become part of the EJP RD ecosystem.
- to identify additional requirements and candidate tools for FAIRification procedures.

**Description**
Bringing rare disease research to the level of efficiency that is required for efficient, computational use of resources (registries, biobanks, molecular databases) is a comprehensive challenge. The VP can only be as good as its resources.
In this session the interoperability considerations that the EJP RD made to support ERNs that aspire to make their registry (and sometimes also their underlying resources) interoperable and FAIR, will be explained.
First experiences with their implementation will be discussed. This encompasses project planning, FAIRification procedures and decision support for data stewards.
Tools and public resources, such as WikiPathways and NextProt, that make submitted data FAIR will be highlighted.
EJP RD stakeholders will be invited to share their experiences and bottlenecks, and candidate tools for inclusion in FAIRification procedures.
Solutions to regulatory and data protection issues

Objectives
The audience will learn about specificities to consider during both the set-up of new projects and the data (re)use of running projects with regards to regulatory and data protection issues. Consent and assent procedure to process lawfully personal data from adults, children and vulnerable populations will be discussed.

Description
Discussion about regulatory and ethical issues
Legal interpretation of GDPR for medical research is varying from one EU country to another. Some legal issues create an impasse for research use. Some examples:

- Data processor and controller definition are paralleled by legal agreements
- The role of data processor/controller depends also on the information included in the informed consent signed by the patient.
- Uploading data to European platform would require (according to strict interpretation) co-controller agreements with all parties accessing the data uploaded on the platform, and reobtaining consent from patient to this end; waiver for medical benefit is under debate (not its usefulness of course, but the legal interpretation)

This session will present guidance diagrams on research data under GDPR and how they can be used according to the different contexts proposed by the audience (interactive part)
Finding and accessing relevant resources (including pre-clinical and clinical ones) for your research needs (special session including P1 funded researchers)

**Objectives**
Learn about existing resources, how to use EJP RD website and Helpdesk, learn about possibilities offered by the Virtual Platform as the project makes progress.

**Description**
Interactive session starting from researchers’ needs already identified (survey, annual retreat), establish priority needs by interactive means so as to orient on a use-case basis how to use the mind map resources overview that is planned and how to query the VP (from the current version at that time).
Applications of the EJP RD Virtual Platform: Counting cases and more & Finding key patients and data

Objectives

Counting cases and more: Demonstration of the process including stakeholder needs; starting from the community surveys and towards the Virtual Platform. Identify requirements to scale up from the current implementation, in terms of stakeholder expectations and technical capabilities.

Finding Key Patients and data: Create a prioritisation plan and ecosystem design for how to start deploying tools emerging from EJP RD, some ERN groups, registries, and other teams that want to work together to set up demonstrators.

Description

Counting cases and more: In this interactive session the audience will walk through the case of counting patients using the emerging EJP RD Virtual Platform (VP). Is it possible for the VP to produce a count instantly from all databases that may contain relevant cases, and in such way that the count is of high quality, that the quality can be checked, and that privacy was preserved? This seemingly simple case challenges many components of the EJP RD VP infrastructure. Participants will be taken through the steps that were implemented so far to address these challenges, discuss what challenges lie ahead, and how this case is helping to shape the infrastructure beyond the capability of counting.

Finding Key Patients and data: Interactive session to establish user’s needs and preferences re finding specific ‘entities’ of interest, such as (but not limited to) distinct patients (by mutations, phenotypes, demographics) including the ‘advanced matchmaking’ challenge; particular datasets (by content, scope, consent, use conditions); potential collaborators/researchers (by location, expertise); and biosamples (by location, use conditions, anatomy, disease). Simultaneously, the audience will be engaged in the discussion if such services should be set up within ERNs or across RD as a whole, and whether EU based or global. The workshop will begin with high-level intro of tools, technologies and approaches being devised in EJP RD, then gather questions/topics from audience, and work through a discussion on each
Outcomes of ERN trainings and fellowships support

**Objectives**
ERN RD Training and Support Program is to fill the gap in the available education on rare diseases research by creating and implementing a comprehensive and cohesive program of education and empowerment for different target groups or stakeholders such as researchers and young clinicians. This session will present the outcomes of the first rounds of selection of Research workshops and Research mobility fellowships as well as upcoming opportunities.
Rare diseases e-learning scheme of EJP RD - How to increase collaboration, avoid overlaps and support stakeholders in the best way

Objectives
Improved coordination, pulling efforts and exchanging on best practices
Objectives are three-fold:
- Update interested parties about the WP16 developments of academic-like online courses on RD research topics
- Make sure WP16 developments are in line with community needs and building upon existing and also, most critically, available expertise
- Identify any potential synergies to be developed in this field, especially in the Covid-19 situation and the increasing demand of online learning

Description
Interactive session with ERN training coordinators/focal points and EJP RD relevant stakeholders and any other interested stakeholders to discuss on the development of the RD e-learning scheme of the EJP RD in order to align actions, propose most suitable solutions and strengthen collaborations.

Program

<table>
<thead>
<tr>
<th>14:50 – 15:20</th>
<th>Presentation of the EJP RD academic online course scheme</th>
<th>Roseline Favresse</th>
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<tbody>
<tr>
<td>15:20-15:30</td>
<td>Ideas &amp; opportunities to foster wider cooperation: plans vs reality when developing online learning materials</td>
<td>Roseline Favresse</td>
</tr>
<tr>
<td>15:30-16:20</td>
<td>round-table, discussion and Q/A on how to increase interaction and impact with interested stakeholders</td>
<td>All</td>
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</table>

Speaker
Roseline Favresse has an academic background in Social Sciences and Humanities in France and Canada (MA in Geopolitics from Sorbonne University and Ecole Normale Supérieure (Paris). She is specialized in the set-up, development and management of international research projects & capacity-building programs. She worked for international organizations, NGOs and consulting companies and developed knowledge of EU public policies and funding schemes/instruments. For 10 years now, she has been working in the RD field, first as a consultant by setting-up and managing FP6/FP7/H2020 projects (incl. TREAT-NMD) in answer to EC Calls for proposals; then, at the French Foundation for Rare Diseases since 2012 as a Regional Coordinator helping clinicians and researchers speeding up their RD development projects for new medicines. She is coordinating EJP RD WP16.
Plenary Closing Session

Chair
Daria Julkowska, Inserm, coordinator of the EJP RD

Speakers
Daria Julkowska (Coordination) + Ralph Schuster, Sonja van Weely, Ana Rath, Franz Schaefer, Virginie Bros-Facer, Biruté Tumiene, Rima Nabbout, Anton Ussi (Pillar 1,2,3,4 leaders)

Objectives
• Summarize the 3 days of the meeting: major conclusions from different sessions
• Provide feedback to participants through gathered questions-answers
• Next steps for EJP RD
• Announcement of the prize winner

Description
After 3 days of intense meeting the session will be focused on summarizing the ideas and feedback that will feed the planning of the EJP RD. It will be also an occasion to announce the winner of the EJP RD General Assembly prize (modalities to be announced separately in advance of the meeting).

Program of the session

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<th>Speaker/Leaders</th>
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<tbody>
<tr>
<td>16:30 – 16:35</td>
<td>Announcement of the prize winner</td>
<td>Daria Julkowska, Inserm (Coo)</td>
</tr>
<tr>
<td>16:35 – 17:25</td>
<td>Major outcomes from Pillar parallel sessions and webinars</td>
<td>Coordination and Pillar Leaders</td>
</tr>
<tr>
<td>17:25 – 17:55</td>
<td>Feedback on questions gathered during the 4 days</td>
<td>Coordination, Pillar Leaders and Work Package Leaders</td>
</tr>
<tr>
<td>17:55 – 18:00</td>
<td>Closure of the meeting</td>
<td>Daria Julkowska, Inserm (Coo)</td>
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</table>
"Speed-meeting" Multinational Clinical Trial Support Office

Format
Interactive session followed by virtual 1to1 meetings

Objectives
Increased knowledge of the Clinical Study Support Office service and how it can be used in practice

Description
The EJP RD has launched an online Clinical Study Support Office (CSSO) assisting European Reference Networks (ERNs) and other clinical teams involved in rare diseases. This support is intended for clinical investigators for the preparation of multinational clinical studies for the development of new drugs, therapies or devices, in addition to repurposing, or diagnostic studies. Requests for this type of support are managed by ECRIN in collaboration with other EJP RD experts within WP20 of the EJP-RD.

The workshop will focus on the presentation of the service through the practical exercise on use cases. In addition to the presentation of the pre-existing examples of the CSSO support, the participants will be invited to propose "use cases" - issues, type of help they need for their specific clinical study and the possible solutions will be presented and discussed. Issues requiring the expertise of CSSO advisors not present at this session will be followed up after the GA.

Program

<table>
<thead>
<tr>
<th>10:30 – 11:20 Interactive session</th>
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<td>10:30 – 10:55 presentation of the support office</td>
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<tr>
<td>10:55 – 11:20 general discussion</td>
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<tr>
<td>11:20 – 11:30 Break</td>
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11:30 1 to 1 session

These sessions are dedicated to discuss confidentially real life problems that researchers faced preparing a clinical study or a request for advice in a study that is under preparation. Each session will have a dedicated time of 20 minutes and Dr M Del Alamo will send upon registration further information for the applicants for these sessions.

Speaker
Marta del Alamo holds a PhD in Molecular Biology and a post-graduate degree in Clinical Trials Management. She is Clinical Project Manager at ECRIN in Paris, coordinating ongoing multinational clinical trials in several European countries and providing support to clinical investigators preparing clinical studies/projects for European funding. Previously Marta worked at SCReN, the Spanish Clinical Research Network, as project manager and was a member of the Ethics Committee at
Hospital Ramon y Cajal in Madrid. As a previous research scientist in the field of molecular biology and virology, she has worked as post-doctoral scientist at research centers in Spain and USA, authoring 11 peer-reviewed publications.