EJP RD

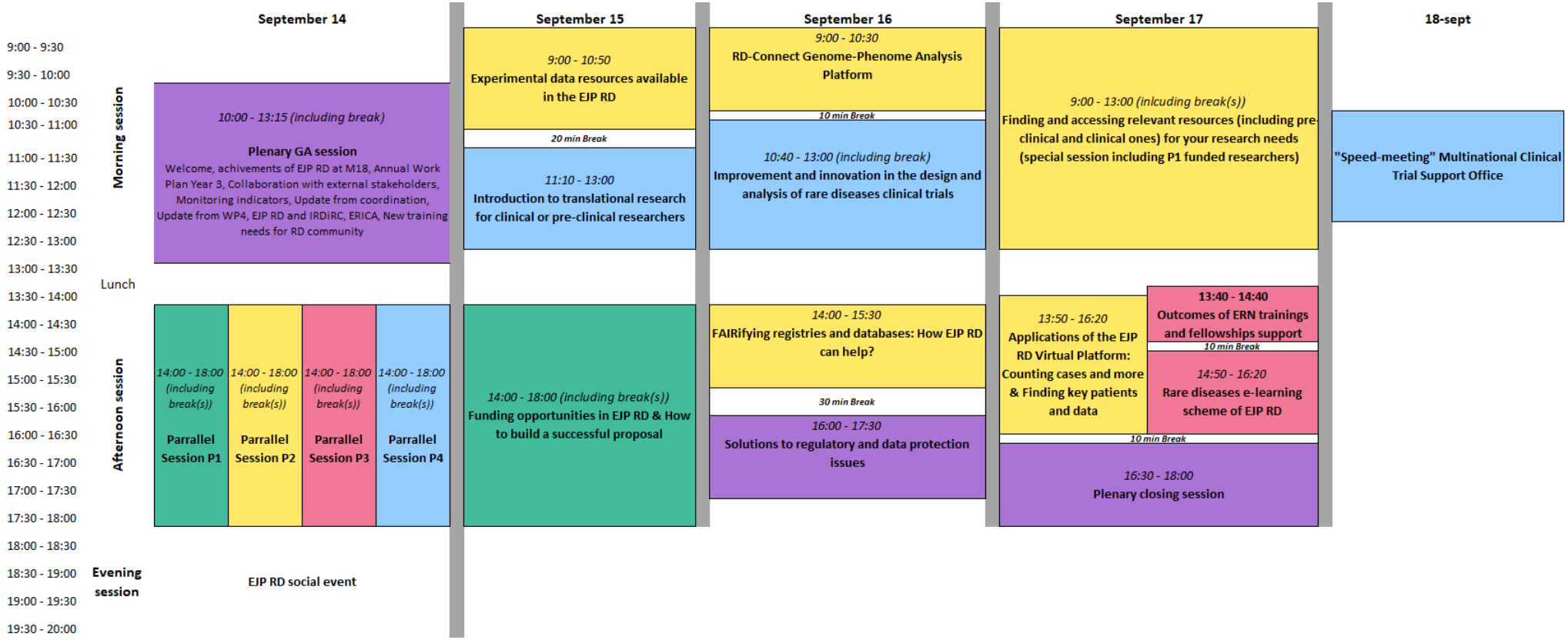
General Assembly and Consortium meeting 2020

Online

September 14th – 18th

Preliminary program

Program at a glance:



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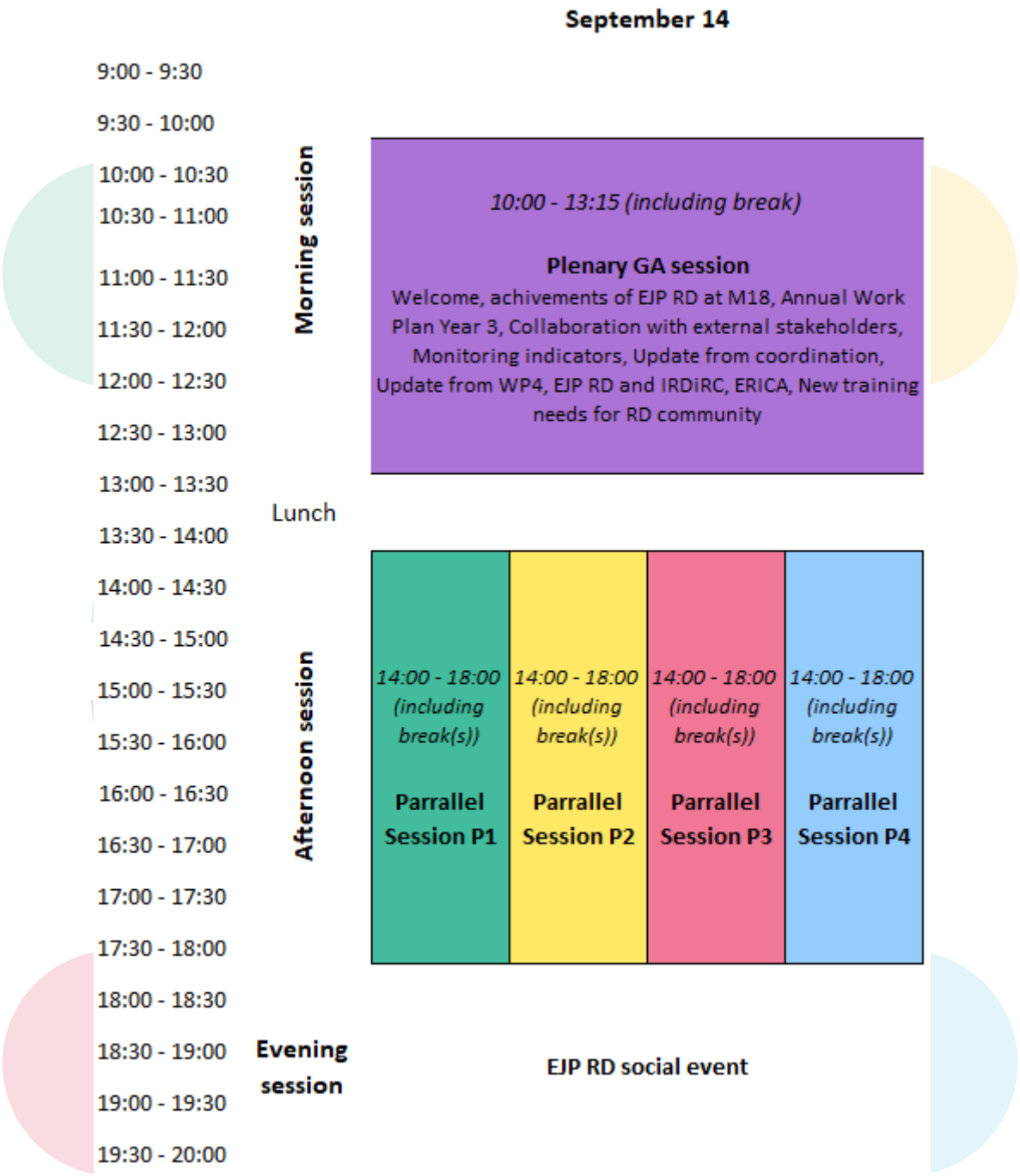

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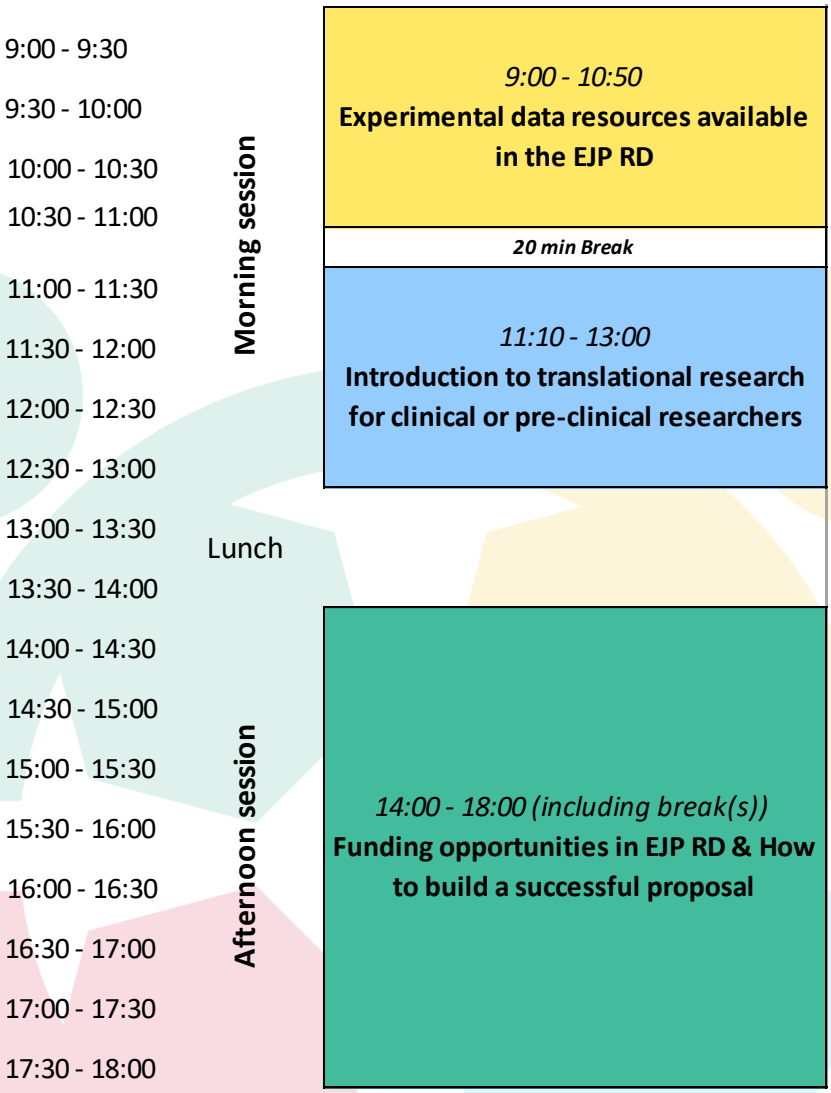

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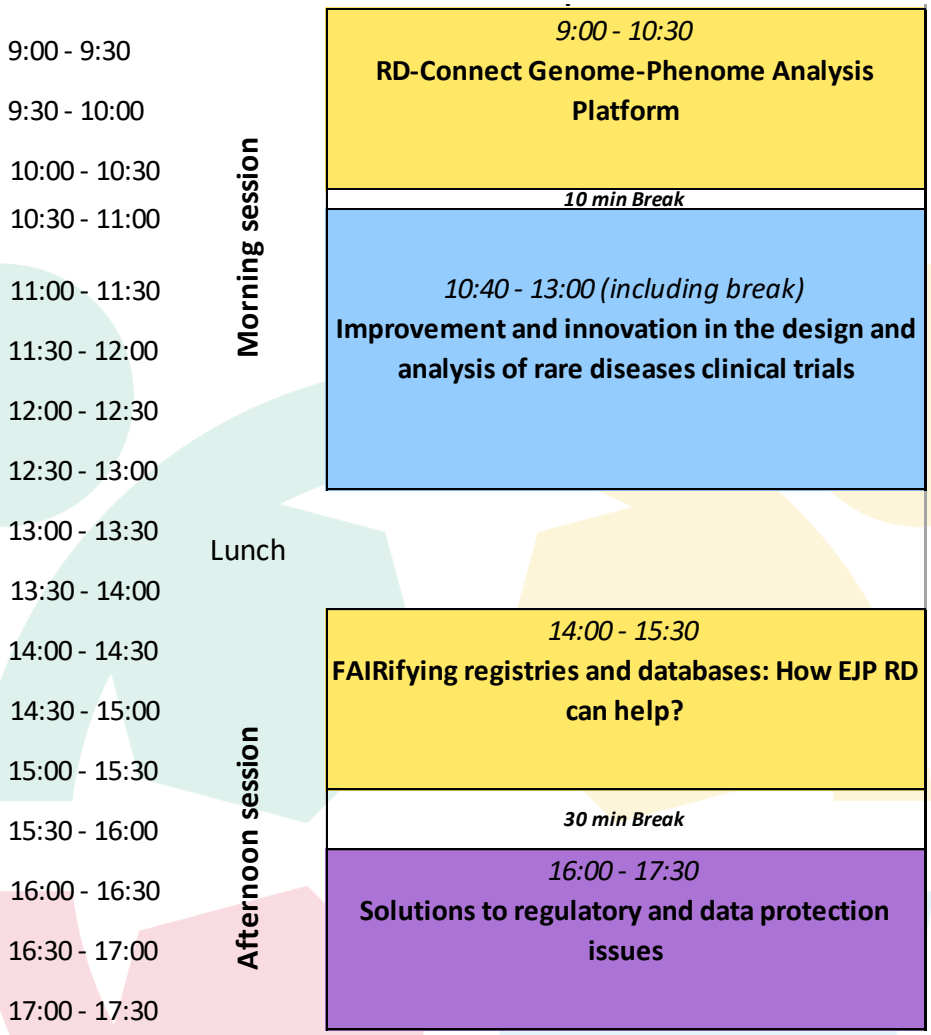

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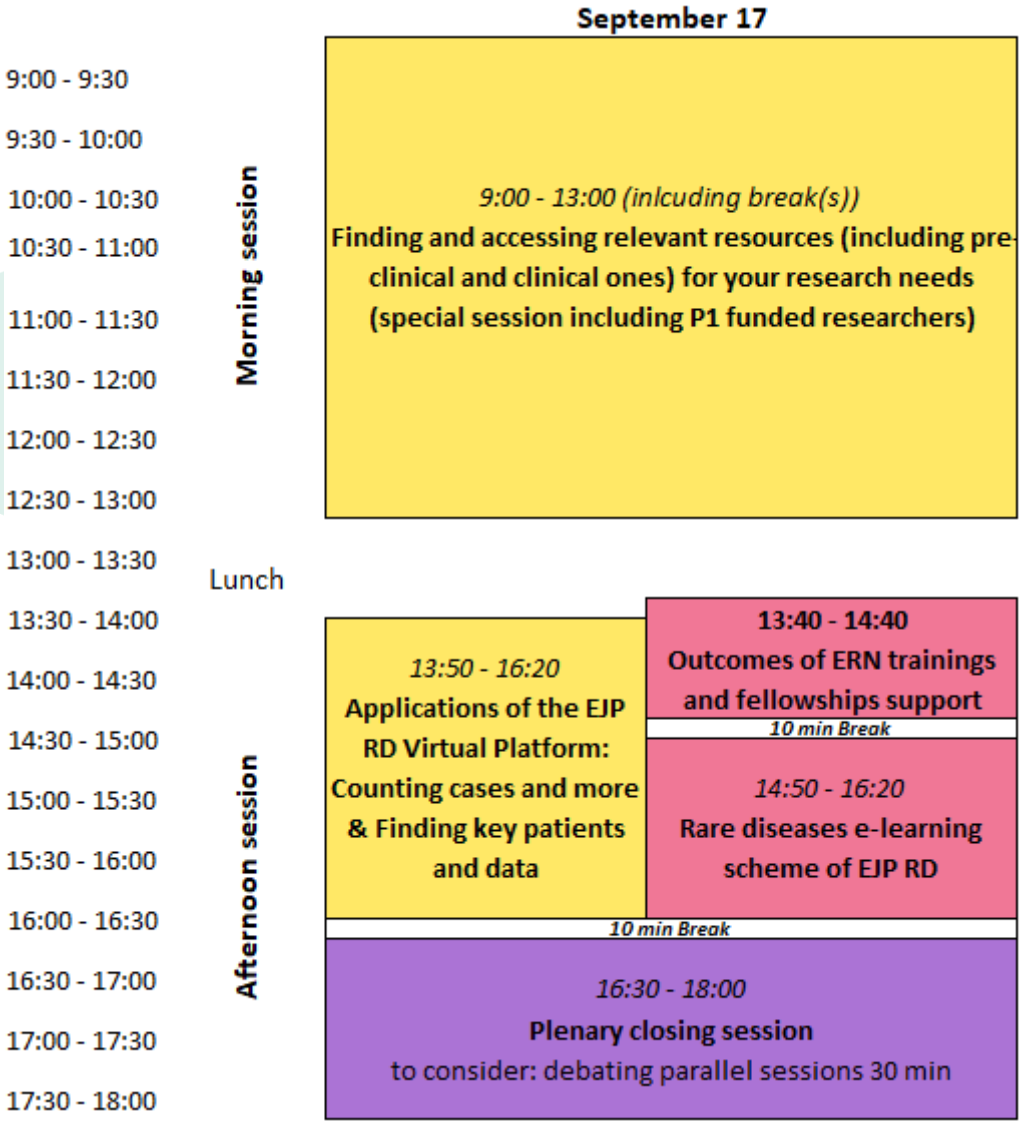

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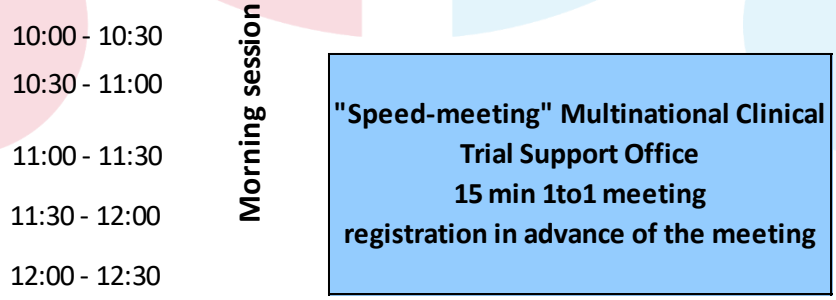

September 14th, 10:00 – 13:00

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 a 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

10:00 – 10:15	Welcome from the Coordination, general objectives of the meeting, EJP RD achievements at M18	Daria Julkowska (Inserm, Coo)
10:15 – 10:35	Annual Work Plan Year 3: - Major changes in the Pillars - Budget changes	Daria Julkowska, Blandine Castrillo (Inserm)
10:35 – 11:00	Collaboration with external stakeholders	Daria Julkowska (Inserm)
11:00 – 11:15	Monitoring indicators: identification of additional global indicators	Kejla Musaraj (tbc)
11:15 – 11:30	Update from coordination (lessons learned from reporting, etc.)	Blandine Castrillo (Inserm)

11:30 – 11:40	Break	
11:40 – 12:00	Update and presentation of the state of the art on ethical, legal and regulatory issues relevant for RD community	Viviana Giannuzzi (FGB), Stefano Benvenuti (FTELE) (tbc)
12:00 – 12:15	EJP RD and IRDiRC – update on Task Forces and joint activities	Carla d'Angelo, Galliano Zanello (Inserm)
12:15 – 12:30	ERICA – coordination and support action for ERN research strategy	Alberto Pereira (tbc)
12:30 – 13:15	New training needs for rare diseases community	Birute Tumiene (tbc)

September 14th, 14:00 – 18:00

Parallel Session Pillar 1

Chairs

Ralph Schuster, Sonja van Weely (Pillar 1 leaders)

Program of the session

Time	Title	Speaker
14:00 – 15:00	WP6 JTC 2020 update **(closed session, funders only)**	Florence Guillot, ANR
15:00 – 15:30	WP6 Update	Ralph Schuster, DLR
15:30 – 16:00	WP7 Update	Sonja van Weely, ZonMw
16:00 – 16:15	*Coffee break*	
16:15 – 16:45	WP8 Update	Christine Fetro, FFRD
16:45 – 17:15	WP9 Update	Irit Allon, CSO/MOH
17:15 – 17:45	WP19 translation/mentoring service update	Anton Ussi, EATRIS; Elena Bertrami, Telethon Italy - tbc
17:45 – 18:00	AOB	

September 14th, 14:00 – 18:00

Parallel Session Pillar 2

Chairs

Ana Rath, Franz Schaeffer (Pillar 2 leaders)

Description

Internal discussion on Pillar 2 advancements.

Program of the session

To be completed

September 14th, 14:00 – 18:00

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.

Program of the session

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

September 14th, 14:00 – 18:00

Parallel Session Pillar 4

Chairs

Rima Nabbout & Anton Ussi (Pillar 4 Leaders)

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

Time	Title	Speaker
14:00 – 14:15	P4 Overview from Co-Chairs	Rima Nabbout and Anton Ussi
14:15 – 15:00	Training opportunites (interactions with P3)	Speaker TBC
15:00 – 15:45	WP19 Update (Tasks by Task Leaders)	Task Leaders
15:45 – 16:00	*Coffee break*	
16:00 – 16:45	WP20 Update (Tasks by Task Leaders)	Task Leaders
16:45 – 17:15	Interactive Discussion - P4 strategy and integration with other Pillars	All
17:15 – 17:45	Interactions with P1 (integrate working with funded projects)	Speaker TBC
17:45 – 18:00	AOB	

September 15th, 9:00 – 10:50

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.

September 15th, 11:10 – 13:00

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

Time	Title	Speaker
11:10 – 11:25	Translational Research for rare diseases: the reverse planning perspective	A. Ussi or T. Andreu (tbc)
11:25 – 11:50	A translational success story: Lentiviral Gene therapy in children with X-linked Severe Combined Immunodeficieny (SCID-XI)	Speaker TBC
11:50 – 12:40	The paving stones of the translational pathway *(each one a 10 min presentation, providing an overview of their contribution to the translational pipeline identifying challenges and solutions)* - The scientist - The clinician - The regulator - The industry - The patient	
12:40 – 13:00	A dialogue with the audience (slide-driven)	All

September 15th, 14:00 – 18:00

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

14:00 – 17:10
Joint transnational calls (JTCs) - How to build a successful proposal

Session 1 – Introduction and scientific evaluation process

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

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

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)

September 16th, 9:00 – 10:30

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

9:00 – 9:15	Introduction to the RD Connect GPAP	Sergi Beltran
9:15 – 10:15	Hands on Workshop	Leslie Matalonga, Steven Laurie
10:15 – 10:30	Discussion	Sergi Beltran

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 other. 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.

September 16th, 10:40 – 13:00

Improvement and innovation in the design and analysis of rare diseases clinical trials

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 specific for rare diseases 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 capability 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 these novel methodologies as well as present the concepts proposed in the funded projects. A discussion on further innovation in methodologies applied to RD clinical trials will also take place. The proposed ideas will serve for the definition of the next EJP RD internal call that will foster innovation projects and shall open at the start of 2021.

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 FAIRifcation procedures.

September 16th, 16:00 – 17:30

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 fom 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)

September 17th, 9:00 – 13:00

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).

September 17th, 13:50 – 16:20

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, demograhics) 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). Simulteneously, 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

September 17th, 13:40 – 14:40

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.

September 17th, 14:50 – 16:20

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

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

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.

September 17th, 16:30 – 18:00

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

16:30 – 16:35	Announcement of the prize winner	Daria Julkowska, Inserm (Coo)
16:35 – 17:25	Major outcomes from Pillar parallel sessions and webinars	Coordination and Pillar Leaders
17:25 – 17:55	Feedback on questions gathered during the 4 days	Coordination, Pillar Leaders and Work Package Leaders
17:55 – 18:00	Closure of the meeting	Daria Julkowska, Inserm (Coo)

September 18th, 10:30 – 12:30

"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 treatments, drug or device repurposing, or diagnostic studies. Requests for this type of support are managed by ECRIN in collaboration with other EJP RD experts.

The workshop will focus on the presentation of the service through the practial excercise 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 solutions will be presented and discussed

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.