EJP RD
General Assembly and Consortium meeting 2020

Online

September 14th – 18th

Program at a glance:

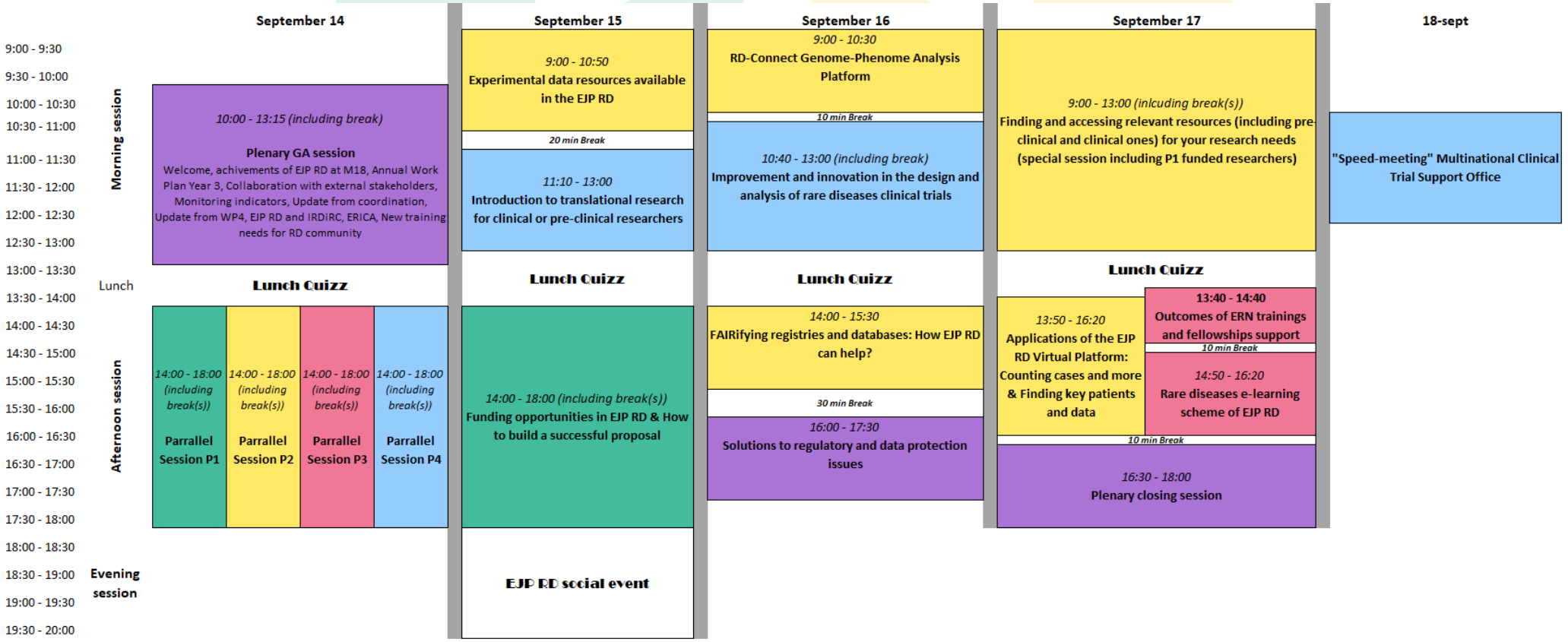
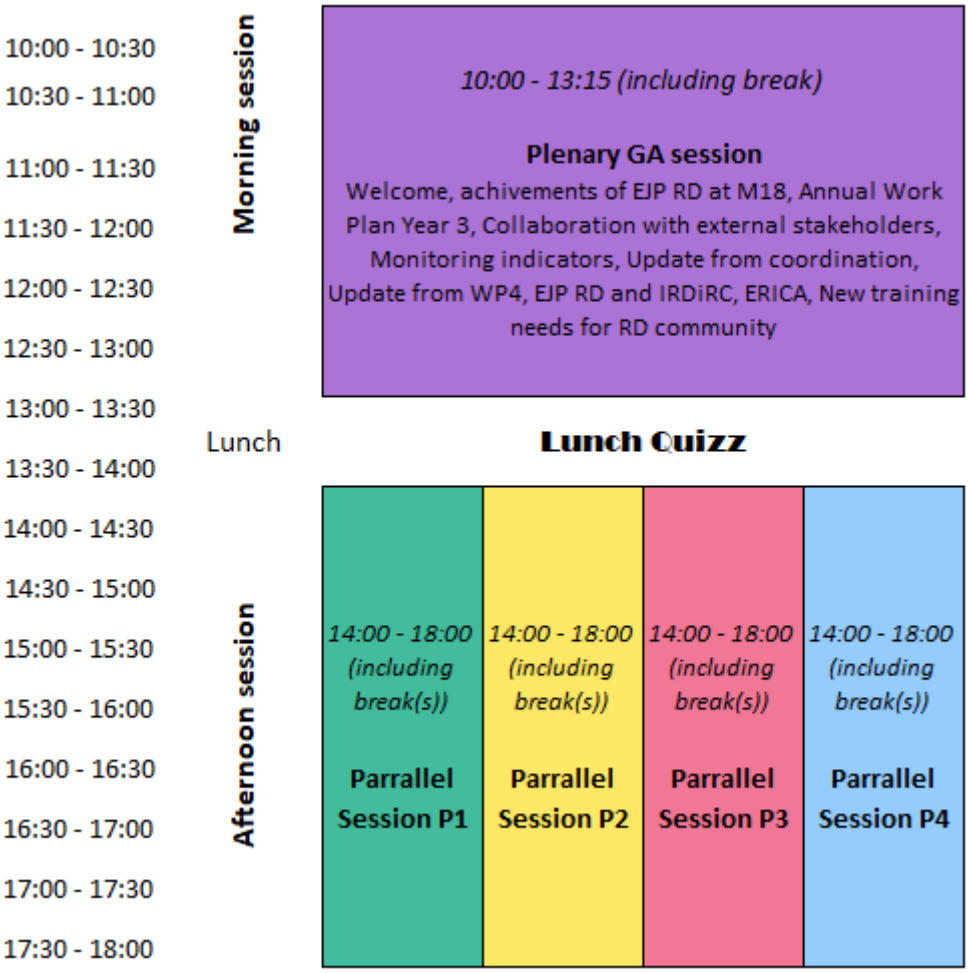


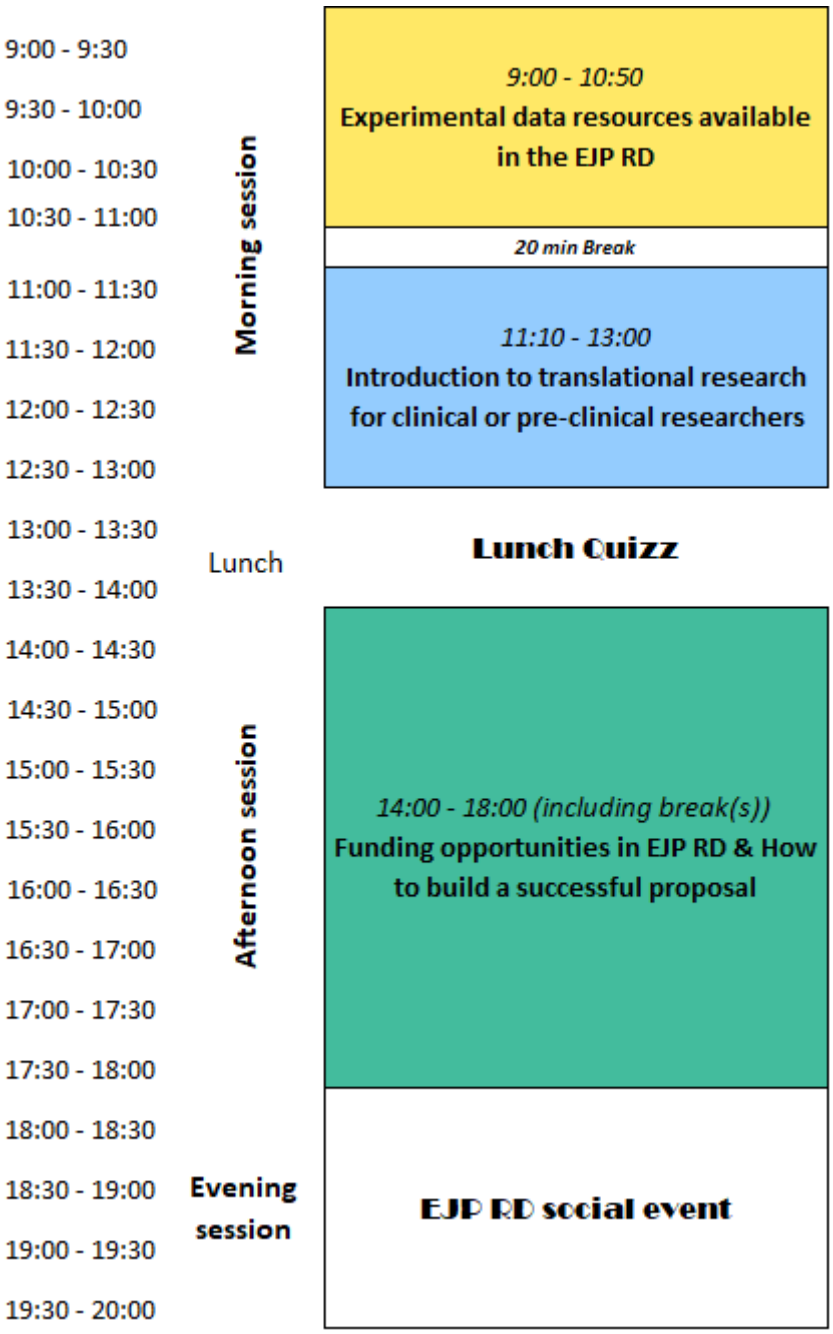
Table of contents

Program per day

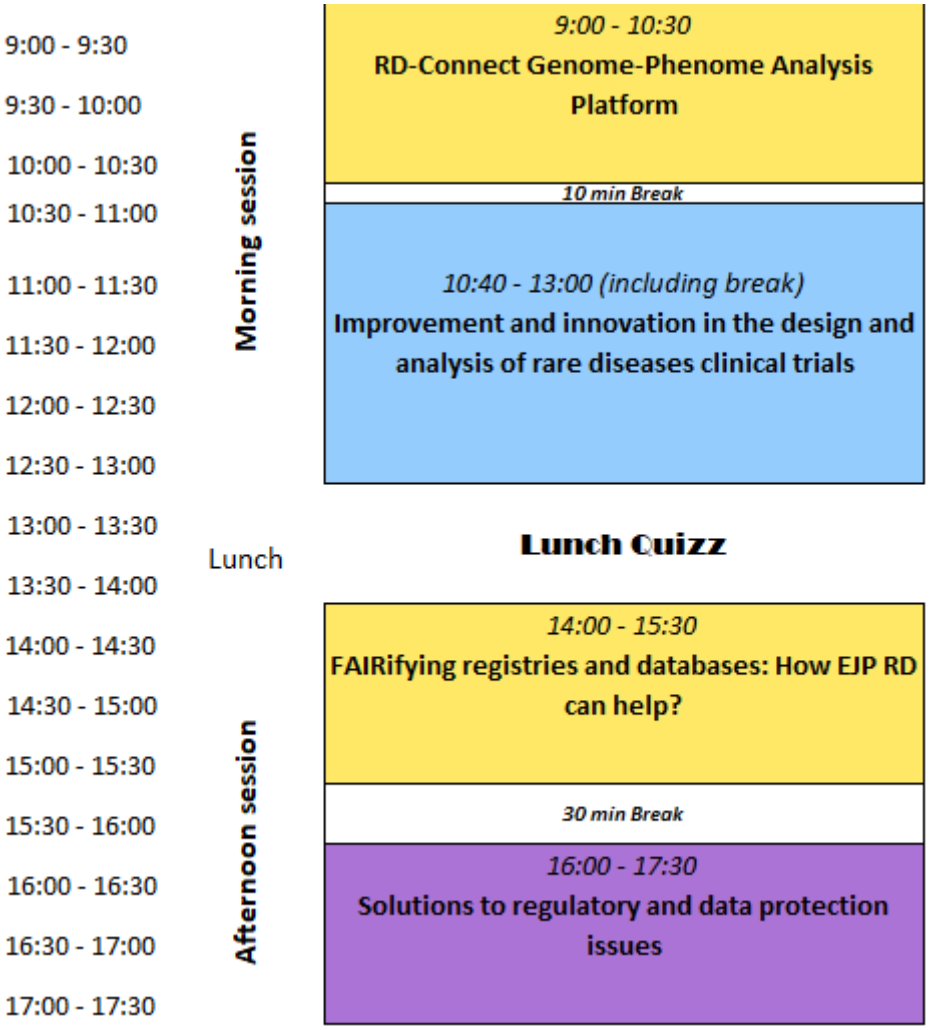
SEPTEMBER 14th



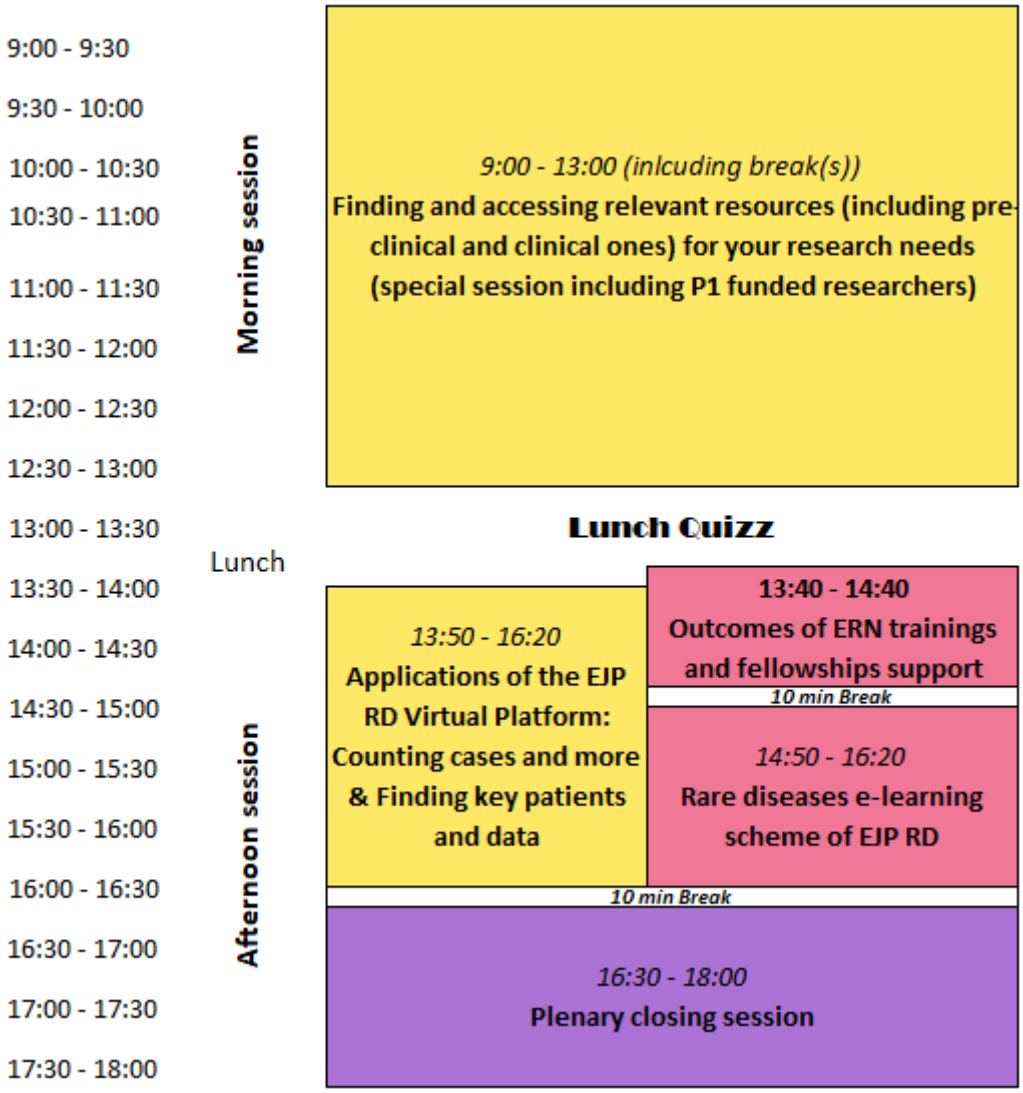
SEPTEMBER 15th



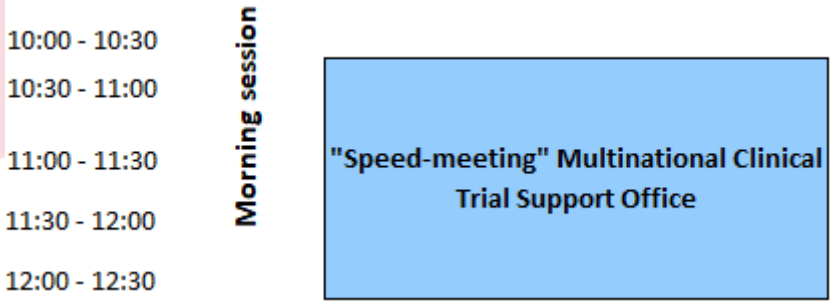
SEPTEMBER 16th



SEPTEMBER 17th



SEPTEMBER 18th



September 14th, 10:00 – 13:00

Plenary GA 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
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)
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
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 Beltrami, 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)

Objectives

- Pillar2 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:

- Reports on FAIRification and Query Builder activity eliciting progress and planned actions with the options foreseen for implementation as well as reporting issues
- 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

Speakers

Michael Nitzlnader : (technical Master Plan)

Bruna Dos Santos Vieira : FAIRification stewards report

Work Foci Leaders*: (Non-Technical Master Plan)

- Use Cases: Mary Wang, Marco Roos
- FAIRification: Marco Roos, Annika Jacobsen
- Resources for sharing experimental data and materials: Giselle Kerry, Daphné Jaoui
- Resources for experimental data analysis and interpretation: Sergi Beltran & Carles Garcia
- Pathway creation and curation: Chris Evelo, Friederike Ehrhart
- Networks: Eleni Mina
- Genetic Variants: Sarah Hunt
- Environment/Adverse Outcome Pathway: Anaïs Baudot, Alberto Mantovani

Anthony J. Brookes (Query Builder progress)

Karl Kreiner (First specifications related to non-functional requirements)

- David Lloyd (Quality)
- Esther van Enckevort: Consent Management, Data Use

- Rob Hooft (Sustainability)
- Michal Prochazka (AAI)

Program of the session	
14:00 – 14:20	**FAIRification Stewards report and next steps** (progress, identified issues)
14:20 – 14:40	Discussion
14:40 – 15:00	**Query Builder progress, planned tests and implementation options**
15:00 – 15:30	Discussion
15:30 – 15:45	*Break*

15:45 – 16:55	**Pillar2 Technical and non-technical Master Plans Finalization**
	Output: complete VP masterplan; working session: collaboratively complete the masterplan.
	Objective: make P2 partners find their place and identify where, with whom and when to work. Michael & Yanis navigating the timeline tool; WFs' As contributors:
	• WP13 WF
	  ○ Pathways creation & curation
	  ○ Networks
	  ○ Genetic Variants
	  ○ Environment/Adverse outcome pathway
	• Resources for sharing experimental data and materials
	• Resources for experimental data analysis and interpretation
	• FAIRification
	• Use cases

16:55 – 17:05	*Break*
17:05 – 18:00	**First specifications related to non-functional requirements (Quality, GDPR, Sustainability, Standards)** will be presented and endorsed *Overall WF session chair* (Karl Kreiner) *Content:* Presentation of the work performed so far: ▪ 8' Life Cycle Management (Karl Kreiner) ▪ 8' Quality: Criteria, Form and results obtained so far ▪ 8' Consent Management, Data Use: what has been done so far (use case work & end-choices ▪ 8' Sustainability: first specifications ▪ 8' AAI: current state of play

	▪ 15' Discussion (incl. a timeline to finalize written reference documents for the project and follow up on the work on endorsing standards in VP).

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:

- 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	

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.

September 15th, 9:00 – 10:50

Experimental data resources available in the EJP RD

Chair and Co-chair

Chair:

Carles Garcia is the Communications and Data HelpDesk Coordinator for GPAP. He has an academic background with a PhD in Genetics working in rare diseases, and then obtained a postgraduate in Project Management. He is currently involved in GPAP in tasks related to user communications, database maitenance and improvement, legal requirements and project management.

Co-chair:

Giselle Kerry is the EGA Helpdesk Team Lead and Project coordinator for EGA EMBL/EBI

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.

Program

9:00 – 9:05	**Introduction of resources**	
9:05 – 9:15	RD-Connect Genome-Phenome Analysis Platform	Sergi Beltran
9:15 – 9:25	European Genome-Phenome Archive (EGA)	Dylan Spalding
9:25 – 9:35	BBMRI-ERIC and Sample Catalogue	Esther van Enckevort
9:35 – 9:45	EJP-RD Cloud Sandbox	Lennart Johansson
9:45 – 9:55	hPSCreg	Nancy Mah
9:55 – 10:05	*Break*	
10:05 – 10:15	Cellosaurus	Amos Bairoch
10:15 – 10:25	Decipher	Julia Foreman
10:25 – 10:35	Metabolights	Keeva Cochrane

10:35 – 10:45	Infrafrontier	Montse Gustems
10:45 – 10:50	**Last questions**	

Speakers

Sergi Beltran (GPAP) is the Head of Bioinformatics Unit at the CNAG-CRG, which is devoted to the development and operation of tools and pipelines for the analysis and interpretation of sequencing data. He has over 15 years of experience with genomics and bioinformatics, he has participated in over 45 international publications and leads the development of the RD Connect GPAP platform.

Dylan Spalding(EGA) is the EGA coordinator. He has an MSc from Reading University and it was based on an investigation of DNA alignment using hybrid evolutionary algorithms, his PhD in Computer Science from Exeter University focused protein homology detection using kernel methods. In 2010 he joined the Peninsular College of Medicine and Dentistry as a bioinformatician, building a pipeline for exome sequence analysis with the primary aim of investigating monogenic causes of diabetes. Dylan joined EMBL-EBI in 2011 as part of the Database of Genomic Variants archive (DGVa) team, working with data on structural variation in all species. He became project lead for the European Genome-phenome Archive (EGA) in 2014 and coordinator of the EGA in 2017.

Esther van Enckevort (BBMRI-ERIC and Sample Catalogue) is a Project Manager at the Department of Genetics at the University Medical Center Groningen (UMCG). She is an expert in software development, data harmonisation, catalogues, and FAIR. She has a strong systems engineering background, including ontologies and semantic web. In recent years she has extended her expertise by leading the development of BBMRI-ERIC Directory, RD-Connect sample catalogue, TraIT studies catalogue, and LifeCycle data item catalogue. She has been involved in the EJP-RD since its inception and is the work package lead in the EUCAN-Connect project for the federated catalogue work package. Esther is a member of the IRDiRC Interdisciplinary Scientific Committee and co-chair of the ISBER Resource Locator Working Group.

Lennart Johansson (EJP-RD Cloud Sandbox) followed the biology and medical laboratory research track of the bachelor Life Sciences and Technology at the Hanze University Groningen and obtained a BASc in 2002. Then he obtained his PhD within the Innovation and Development section of the UMCG department of Genetics on the topic 'Novel algorithms for genetic variation detection. At that time he had already started to work as a project coordinator focusing on rare disease projects, such as EJP-RD, Solve-RD and CINECA, both on the side of infrastructure (HPC Computing, (meta-)databases) as data-analysis.

Nancy Mah (hPSCreg) has a background in molecular biology with a PhD in the field of Cell Biology, complemented with bioinformatics, and she has been

working in the stem cell field for over a decade. Since joining the hPSCreg project in 2014, she has contributed to the project's data structure and management, which entails not only biological cell line data but also the ethical provenance of these lines. Her goals within the EJP-RD project are to make the Human Pluripotent Stem Cell Registry (hPSCreg) interoperable with other resources in the Virtual Platform and to help educate others on FAIR data management through the biobank training workshops offered by EJP-RD.

Amos Bairoch (Cellosaurus) is professor of Bioinformatics at the University of Geneva, Amos Bairoch is head of the CALIPHO group of the SIB - Swiss Institute of Bioinformatics. Amos Bairoch's main work lies in the field of protein sequence analysis and the development of databases and software tools for this purpose. Currently his main activities are focused on the development of neXtProt, a web knowledge platform on human proteins and of the Cellosaurus, a cell line knowledgebase. Amos Bairoch has been awarded several distinctions, among which the European Latsis Prize, the Otto Naegeli Prize, the HUPO Distinguished Achievement Award and the ABRF Award for Outstanding Contributions to Biomolecular Technologies. He is an ISCB Fellow.

Julia Foreman (DECIPHER) is a Project Manager at the Sanger Institute, working in the DECIPHER web-based platform.

Keeva Cochrane (Metabolights) is a senior technical officer within the Metabolomics team at EBI, managing curation and outreach. She has a degree in biochemistry and PhD in molecular therapeutics. Keeva has significant experience of working with small and large biological and chemical datasets having worked in drug development R&D in academia and industry for many years. As part of her role at in the metabolomics team she supports submission of metabolomics data to the MetaboLights database.

Montse Gustems (INFRAFRONTIER) obtained her PhD in Biology from the University of Barcelona in 2008. After that, she moved to Munich and worked as a research scientist at the Helmholtz Zentrum München. In 2017, she joined the INFRAFRONTIER's scientific user support team.

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 – 11:30	Case Study on "Crossing the Valley of Death"	Toni Andreu
11:30 – 11:40	Questions to Audience	
11:40 – 11:45	Introductory remarks	
11:45 – 12:00	'The Development Process at a Glance'	Bernd Eisele
12:00 – 12:15	'Navigating the Regulatory Maze'	Giovanni Migliaccio
12:15 – 12:30	'Patents – A Necessary Evil'	Anton Ussi
12:30 – 13:00	Dialogue with Audience	

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– 17.10	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 GlaxoSmithKilne (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 and Neurology Departments of the Institute of Child Health and Great Ormond St. Children Hospital in London, UK. Prof. Elpeleg focuses on establishing proper diagnosis to children affected by developmental disorders. Her research is aimed at the identification of the genetic basis of monogenic disorders, using advanced technologies. The introduction of next generation sequencing enabled Prof. Elpeleg and her team to identify over 100 new disease causing genes, to delineate their phenotypic and molecular spectrum and to dissect disease mechanisms.

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.

Prof. Christina Has is a Professor for Dermatology at the University of Freiburg, Germany. Her research focus are genodermatoses. She is internationally recognized for her work on the genetic basis of genodermatoses, in particular of skin fragility disorders. With her group, she has identified new genes and characterized large cohorts of patients with genodermatoses, established genotype-phenotype correlations and explored the underlying disease mechanisms.

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.

September 16th, 9:00 – 10:30

RD-Connect Genome-Phenome Analysis Platform

Chair

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 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 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

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 heterogenicity 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		
10:40 – 10:50	Introduction to aims of WP20 activities	R. Nabbout
10:50 – 11:00	Overview of the clinical trials support office	M. d.Alamo
11:00 – 11:25	Presentation with hurdles/obstacles and methodological solutions with respect also to funded Demonstration Projects (Ideal, Asterix, Inspire)	R.-D. Hilgers
11:25 – 11:45	Question and Answers (Open discussion about methodologies)	
11:45 – 11:50	Break	
11:50 – 12:50	Discussion about Innovation Projects (completely interactive roundtable)	ECRIN (M. D.Alamo and Jacques Demotes), RD Hilgers, Statistician from IDEAL, R. Nabbout, clinician/Pharmacology person
12:50 – 13:00	Toolbox and educational program for clinical trials in rare diseases	M. d.Alamo, L. Sangiorgi

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.

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 Clincal 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 FAIRifcation procedures.

Program

Part I - informing		
14:00 – 14:10	general introduction: the generic FAIRification workflow	Marco Roos, Annika Jacobsen
14:10 – 14:20	A FAIRer registry - the VASCA pilot	Martijn Kersloot (tbc)
14:20 – 14:30	introduction to the FAIRification steward team	Bruna Dos Santos Vieira
14:30 – 14:40	the FAIRification work focus roadmap	Annika Jacobsen, Michael Nitzlnader [visualisation] (tbc)
14:40 – 14:50	*Break*	
Part II - Q&A		

14:50 – 15:00	Starter: FAIRification Toolbox for stewards: the FAIR standards mindmap & matrix, measuring FAIRness	Nirupama Benis, Annika Jacobsen, Bruna Dos Santos Vieira
15:00 – 15:30	Q&A	All

Speakers

Marco Roos' research focus is on making state-of-the-art computer science applicable to enhance biomedical research, particularly for rare human diseases and with knowledge discovery and data linking techniques. He has extended his original biological interest in the role of chromatin in the functioning of the cell to bridging between genotype and phenotype using data linking techniques and data science. After including computer science subjects in his MSc in molecular biology, Marco worked as a multidisciplinary researcher in research groups in life science and computer science. Now, as group leader of the Biosemantics research group of prof. Dr. Barend Mons, LUMC, he leads the research, development and application of knowledge discovery methods for human genetics research. He co-leads the rare disease community of the European life science data infrastructure ELIXIR, FAIR* 'at source' activities in the European Joint Program Rare Diseases, and initiated the Rare Diseases Global Open FAIR implementation network. An important aspect of Marco's work is advocating principles of FAIR and Linked Data to create a powerful substrate and world-wide robust infrastructure for knowledge discovery across heterogeneous data distributed over institutes and countries.

* FAIR principles: data should be findable, accessible, interoperable, reusable for humans and computers.

https://orcid.org/0000-0002-8691-772X

Annika Jacobsen
https://orcid.org/0000-0003-4818-2360

Martijn Kersloot
https://orcid.org/0000-0003-3357-3027

Bruna Dos Santos Vieira obtained her MSc degree in Public Health from the University of Porto, Portugal. The thesis, also developed in Portugal, focused on data quality analysis for hospital-based cancer registries. From 2011-2014, she studied Hospital Management in the University Feevale, in Brazil. While studying, she worked at Grupo Hospitalar Conceição and developed integrated projects involving process, people, logistics, strategic planning, and financial management. Currently at Radboudumc, Bruna works as a data steward for the CMBI and Radiology departments, collaborating in multiple projects and implementations such as EJPRD and the VASCA registry for Rare Vascular Anomalies.

Nirupama Benis
https://orcid.org/0000-0002-2101-6154

September 16th, 16:00 – 17:30

Solutions to regulatory and data protection issues

Chair/co-chair

Vivianna Giannuzi / Petr Holub

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 How can I collect data and sample from patients? How can I use or re-use sample I collected in a certain clinical study for my research? How shall I treat such data? can I use them in further research projects? GDPR provisions on informed consent and assent procedure will be discussed as well.

Description

How researchers can collect, use and re-use data for their research projects while abiding all ethical and law (GDPR) requirements?
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)

Program

16:00 – 16:10	Introduction of the topic	Viviana Giannuzzi, Petr Holub
16:10 – 16:25	Legal interpretation and application of GDPR in the biomedical research	Irene Schlünder
16:25 – 16:40	Informed consent and GDPR	Annalisa Landi

16:40 – 16:55	EJPRD - GDPR Guidelines	Petr Holub, Michaela Th. Mayrhofer
16:55 – 17:00	*Break*	
17:00 – 17:30	Question and Answers – Open discussion	All

Speakers

Viviana Giannuzzi: PharmD, PhD in cellular biochemistry and pharmacology, post-graduate degree master in Clinical Research of Medicines.
Her main areas of expertise are: ethics and regulatory, non-clinical and clinical research, ICH-GCP, clinical studies application, pharmacology, biomedical research for rare and paediatric diseases.
She performed non-clinical research in rare diseases affecting the skeletal muscle, such as genetic channellopathies and Duchenne Muscular Distrophy. She was the Secretariat Scientific Assistant of the Ethics Committee in Lecce (IT). From 2017 to August 2020, she was Patient Representative Delegate Alternate at Paediatric Committee, European Medicine Agency (EMA).
Currently, she is coordinator of research department at Fondazione per la Ricerca Farmacologica Gianni Benzi onlus, and acts as project manager of EuOrphan and researcher/ethical and regulatory expert in EU and national research projects.
Author/co-author of peer reviewed publications and abstracts and speaker at national and international congresses/meetings in the relevant fields of expertise.
She is member of WP4 Ethics Working Group of EnprEMA, Working Group on Rare Cancers (WGRC) of ECPC (European Cancer Patient Coalition) and the Italian Society of Pharmacology coordinating the paediatric pharmacology Working Group.

Petr Holub, Ph.D., is an Associate Professor of computer science at Masaryk University, the second largest university in the Czech Republic. Since 2015, he has become Senior IT/Data Protection Manager with BBMRI-ERIC, European Research Infrastructure Consortium for Biobanking and BioMolecular Resources. He has background both in sciences (MSc with specialization on computational quantum chemistry in 2001) and computer science (PhD in 2005, specializing on computer networks, self-organizing systems and multimedia processing). He was head of the Division of Communication Technologies at Institute of Computers Science, Masaryk University, as well as architect of advanced multimedia and collaborative systems of the Czech national e-infrastructure developed and operated by CESNET. His research in computer networks and multimedia processing has lead to more than 40 research papers in established computer science journals and ranked conferences, as well as to practical software systems and patents. He was a chief architect and team leader of UltraGrid, high-performance media processing and distribution framework, which received Best Open-Source Software Award by ACM Multimedia SIG. He is a co-founder of Comprimato Systems company, which further develops and commercializes research results in acceleration of compression algorithms. He has extensive collaboration with a number of research groups in the USA (e.g., EVL UIC, CalIT2 UCSD, CCT EVL, NLM NIH, RIT), Europe (e.g., i2cat, SurfSARA, NorduNET, SurfSARA), and Japan (AIST).

Since 2011, he has been responsible for design and development of IT infrastructure of the Czech BBMRI, as well as its subsequent operation using national research cloud e-infrastructure CERIT-SC. He is also responsible for the IT and data management

infrastructure of the ongoing EU THALAMOSS project, focusing on data collection and processing for stratification of patients suffering from thalassaemia. He has been a contributor to a number of international and national projects linking computer science and biomedicine, such as EU ITHANET or MediGrid, as well as a number of other international computer science oriented projects, e.g. EU GridLab, EU CoreGRID, or NSF. Strategies for Remote Visualization on a Dynamically Configurable Testbed (eaviv). https://orcid.org/0000-0002-5358-616X

Annalisa Landi: Researcher at Gianni Benzi Pharmacological Research Foundation in Bari, Italy. PharmD with a II level post-graduate Master in Regulatory Sciences at University of Pavia. She carried out an internship at Leiden University Medical Center in the FAIR team of Marco Roos. Member of the WG3 of the European Network of Centres for Pharmacoepidemiology and Pharmacovigilance (ENCePP). She collaborates in several national and international projects dealing with ethics in biomedical research. She is involved in scientific, ethical and regulatory activities particularly related to clinical studies, data protection and confidentiality, plan and management of patient registries and medicine databases, health data and accessibilities issues.

Irene Schlünder is a lawyer and expert in EU data protection law and database governance as well as IP rights. She works for TMF e.V., a member based German umbrella organisation for biomedical research in Berlin. She was involved in the IMI projects EHR4CR (co-author of the IMI Code of Conduct) and DO-IT (development of an Informed Consent Template for clinical trials including secondary use of the data for future research) as well as in H2020 projects like BioMedBridges, CORBEL and EOSC Life. She is a member of the Common Service ELSI of BBMRI and belongs to the core drafting group of the BBMRI lead initiative for a "Code of Conduct for health research" under the GDPR. She also collaborates with the German Ministry of Health to contribute to the development of the "European Health Data Space". She joined the EJP RD Advisory and Regulatory Ethics Board acting as a Legal Advisor.

Michaela Th. Mayrhofer serves as Head of ELSI Services & Research at BBMRI-ERIC. During her academic career, she held various positions at the Centre de Recherche Médecine, Sciences, Santé et Société, the University of Vienna, the Institute of Science, Technology and Society Studies at Alpen-Adria-Universität Klagenfurt/ Vienna/Graz, the Technical University of Vienna, the Medical University of Graz, and the University of Newcastle. Her research and implementation interests lie in the politics and governance of the life sciences. She is further spearheading the Code of Conduct for Health Research initiative and partake in several H2020 projects in various roles. She is part of the EJP RD Advisory and Regulatory Ethics Board.

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)

Chair, Co-chair

Chair: Giselle Kerry
Co-chair Ana Rath

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 showing what Pillar 2 partners learned from surveys addressing ERNs and researchers' needs in term of resources, and how can Pillar 2 partners help them. Presenters will take attendees through their services and will take their questions for on-session or later follow-up. Use cases will be presented to illustrate how researchers can interact with EJPRD partners.

Program

9:00 – 9:20	Research needs for RD: ERNs and researchers survey results	Mary Wang
9:20 – 9:35	EJPRD partner resources addressing researchers and ERNs' needs: Mindmap presentation	Ana Rath
9:35 – 10:05	Finding registries and biobbanks/biosamples: • Orphanet/Orphadata • ERDRI • BBMRI/RD-Connect Biobank and Registry Finder / Sample Catalogue	A Rath, Marc Hanauer, Holger Storf, Heimo Müller, Petr Holub
10:05 – 10:35	Finding tools for research: • Bio.tools • ELIXIR Bundle services • RD WikiPathways	Hans Ieanasescu, Gary Saunders, Chris Evelo
10:35 – 10:50	*Break*	

10:50 – 11:00	Finding animal models: • Infrafrontier	Sabine Fessele, Montserrat Gustems (tbc)
11:00 – 11:20	Finding cell lines: • Cellosaurus • hPSCreg	Amos Bairoch, Alain Gateau (tbc) Nancy Lynne Mah
11:20 – 11:40	Finding resources to deposit and analyse data (phenomics/genomics/multiomics) • RD-Connect GPAP • EGA	Sergi Beltran, Giselle Kerry
11:40 – 12:00	Finding support for translational and clinical research • EATRIS/IMT • ECRIN/WP20	Anton Ussi, Marta del Alamo
12:00 – 12:15	*Break*	
12:15 – 13:00	Use cases and general discussion	All

September 17th, 13:50 – 16:20

Applications of the EJP RD Virtual Platform: Counting cases and more & Finding key patients and data

Chairs

Part 1
Günter Schreier (task lead software development federated architecture)
Marco Roos (co-chair Use cases work focus, co-lead WP12)

Part 2
Anthony J. Brookes (task lead patient matchmaking, co-lead WP12)

Objectives

Part 1 - Towards a specification of the Virtual Platform using simple patient counting as driver & first test case: to introduce the counting case for defining primary virtual platform specifications from a user and technical point of view; obtain feedback from participants for obtaining counts and communication between components of the VP including ERN sources; to identify requirements for scaling up in terms of stakeholder expectations and technical capabilities.

Part II - Finding Key Patients and data: to 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

This interactive session covers two important aspects for developing the EJP RD Virtual Platform. The first considers the specification of the Virtual Platform that uses simple patient counting as driver & first test case. The second looks more deeply into techniques involved in patient matchmaking and querying.

Will it be possible for the VP to reproduce a measurement, a simple count, instantly from distributed source databases, with high and traceable quality, without sacrificing privacy?

In part I of this session, we explain the scope and the purpose of the counting case and gather feedback and suggestions from all participants, going back and forth between a user and technical point of view. We run through the counting case specified by the Use case work focus and look at where counts are generated by some ERN sources. This seemingly simple use case already challenges many components of the EJP RD VP infrastructure. Overall architecture WF members take participants through considerations regarding the specifications that are needed to allow VP components to interoperate.

Part II is an interactive session to establish user's needs and preferences for 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 session 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.

Program

13:50 – 14:40	Towards a specification of the Virtual Platform using simple patient counting as driver & first test case
14:40 – 14:55	Break
14:55 – 15:45	Patient matchmaking and querying
15:45 – 16:00	Break
16:00 – 16:20	Joint Q&A ('parking lot')

September 17th, 13:40 – 14:40

Outcomes of ERN trainings and fellowships support

Chair

Holm Graessner has graduated in Biomedical Engineering, Electrical Engineering, German Language and Literature, Philosophy as well as Business Administration. He received his PhD "Summa cum laude" in 2004 and, then, he obtained his MBA degree in 2008.
He has been Managing Director of the Rare Disease Centre, since 2010, at the University and University Hospital Tübingen, Germany. He is Coordinator of the European Reference Network for Rare Neurological Diseases (ERN-RND). www.ern-rnd.eu. Together with Olaf Riess, he coordinates the H2020 Solve-RD project on "Solving the unsolved rare diseases". www.solve-rd.eu
From 2003 until now, he has been coordinating and managing more than 10 EU funded collaborative projects. The main focus of these projects are rare and neurological diseases, among them EUROSCA, MEFOPA, SENSE-PARK, MULTISYN and NEUROMICS.
He has been co-leading one of the four working groups of the German Action Plan for Rare Diseases. Since 2020, he is a personal member of the Rare Disease Coordinating Panel of the EAN (European Academy of Neurology).

Objectives

- Give an update of the activities performed until now in the ERN training and support programm and the next milestones.
- Exchange with other WPs on the experiences regarding organization of workshops and conducting secondments.
- Discuss possible solutions for the successful organization of the trainings in view of the Covid-19 situation.
- Discuss the process of accreditation of the workshops (should start once the first round of selected workshops have been conducted).

Program

13:40 – 14:00	Presentation	Sanja Hermanns
14:00 – 14:40	Discussion	all

Speaker

Sanja Hermanns - After having obtained her PhD in Biology and Health, Sanja Hermanns has been working as a PostDoc for 3 years in the industry on an applied research project. She has then obtained an Certificate of Advanced Studies in didactics for university education and has been working for 9 months on the development of educational materials for STEM subjects and 2 years in

a simulation laboratory for training of medical students at the University Hospital Tübingen. Since 2018 she is working for the European Reference Network for Rare Neurological Diseases and is responsible for the Training activities and the Clinical Patient Management System (CPMS).

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 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

10:30 – 11:20 Interactive session	
10:30 – 10:55	presentation of the support office
10:55 – 11:20	general discussion
11:20 – 11:30	Break
11:30	**1 to 1 session**
	These sessions are dedicated to discussing 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.