

European Joint Programme on Rare Diseases

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Rare Disease European Joint Programme Cofund



Grant agreement number 825575

EJP RD Final Conference

Agenda

May 27-28, 2024

Bari, Italy

Program at a glance:

CEST	Day 1 (27/05/2024)	CEST	Day 2 (28/05/2024)
08:30-09:00	Registration		
9:00 - 09:30	Welcome (Coo, Host, EC)		
9:30 - 10:30	Keynote "Can AI overcome the curse of small numbers in RD research"	9:00 - 11:00	Health and research policy - collaboration perspectives
10:30 - 11:00	<i>Coffee Break</i>		
11:00 - 12:30	The Virtual Platform: a data ecosystem for RD research	11:00 - 11:30	<i>Coffee Break</i>
12:30 - 13:30	Networking Lunch Break	11:30 - 13:00	From collaboration to enterprise
13:30 - 15:30	Building research capacity through training and mentoring	13:00 - 14:00	Networking Lunch Break
15:30 - 16:00	<i>Coffee Break</i>		
16:00 - 17:30	Variety is the spice of life – how EJP RD research funding addresses a multitude of stakeholders and topics		
17:45	<i>Short Break</i>		
17:45 - 19:15	Impact of EJP RD in clinical research		
20:00 - 00:00	Social Event		

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May 27, 9h00 – 9h30 CEST

Welcome Introduction

Time (CEST)	Title	Speaker
9:00-9:10	Welcome from the EJP RD Coordinator	Daria Julkowska (INSERM)
9:10 – 9:20	Welcome from the host	Fedele Bonifazi (President of Fondazione per la Ricerca Farmacologica Gianni Benzi onlus)
9:20 – 9:30	Welcome from the European Commission (EC)	Irene Norstedt (EC)

May 27, 9h30 – 10h30 CEST

Keynote - "Can AI overcome the curse of small numbers in RD research"

Chair/co-chair

Daria Julkowska (INSERM)

May 27, 11h00 – 12h30 CEST

The Virtual Platform: a data ecosystem for RD research

Chair/co-chair

Ana Rath (INSERM), Marco Roos (LUMC)

Objectives

- Learn about how EJP RD helped out making resources for RD research more FAIR compliant by integrating a virtual federated ecosystem, The Virtual Platform
- Learn about how FAIR data facilitates data re-use for analysis from a single resource or from several resources in a federated way
- Learn about tools, methods and facilities developed by EJP RD to make resources and data FAIR

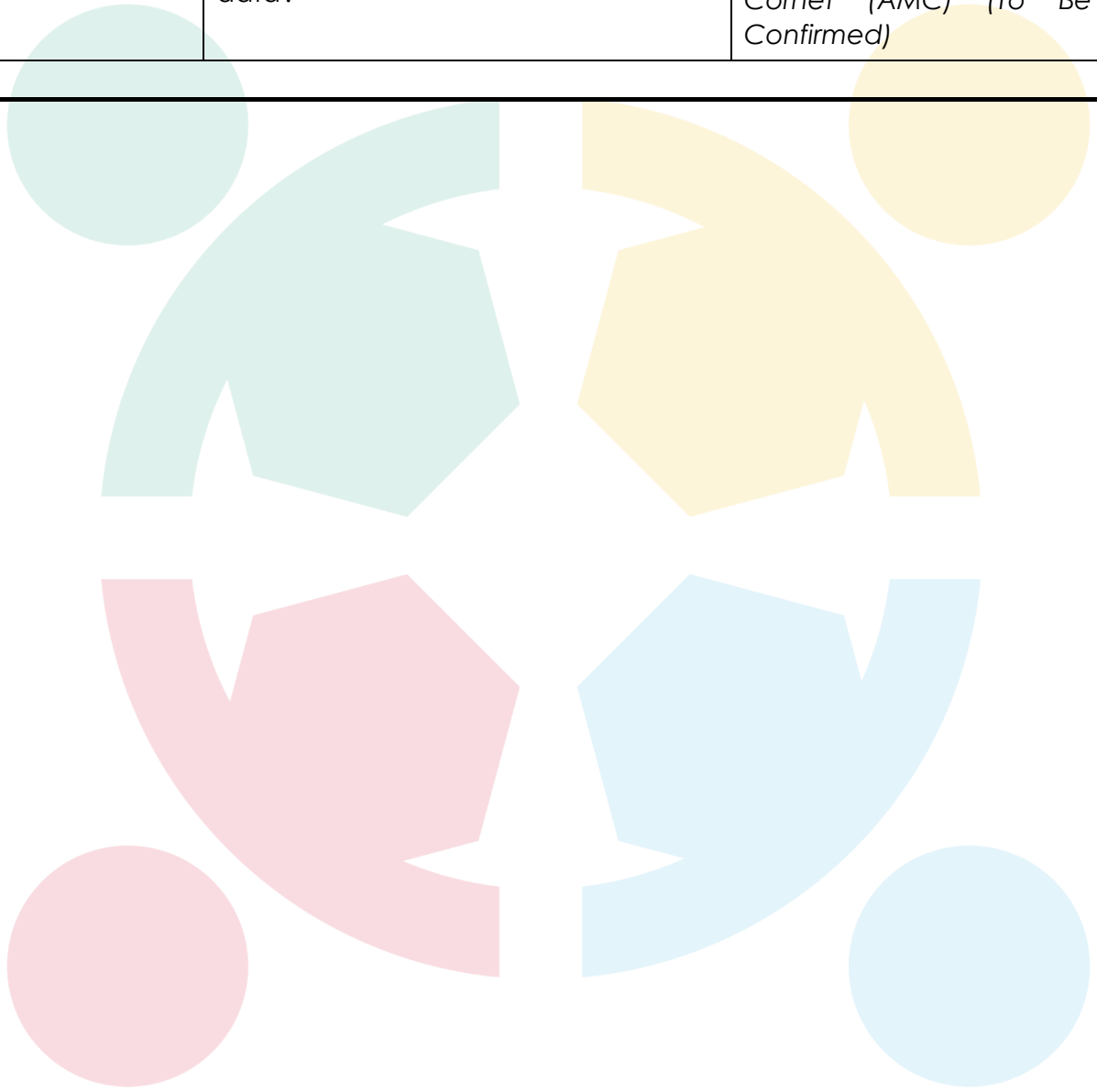
Description

In this session Pillar 2 partners will show different levels of becoming FAIR for a data source thanks to methods and tools developed during the project in the context of the Virtual platform ecosystem of resources for RD research. It will be exemplified by use cases' demonstrations and experiences within and beyond EJP RD. Feedback interactive panel will allow to understand the benefits, the difficulties and the bottlenecks yet to be addressed in the future for continue building and expanding the VP ecosystem.

Program of the session

Time (CEST)	Title	Speaker / Panellists (TBC)
11-11:10	Introduction to the session "From data sources fragmentation to a data ecosystem: the Virtual Platform"	Marco Roos (LUMC)/Ana Rath (INSERM)
11:10-11:50	Demonstration of exploiting the VP network: how collaborative interdisciplinary research on data allows for finding and using data sources	Luiz Bonino Da Silva (LUMC) (To Be Confirmed) VP discoverability connexion levels (demonstration) VP federated query and analysis demonstration
11:50-12:10	How do data sources enable federated research scenarios?	Bruna Dos Santos Vieira (Radboudumc) (To Be Confirmed)

<p>12-10-12:30</p>	<p>Moderated interactive session: How do we continue to build the VP network together?</p> <p>How do you envision to use the network of VP compliant sources? How the VP network will serve a larger ecosystem of health and research data?</p>	<p>Chairs: Marco Roos (LUMC), Ana Rath (INSERM)</p> <p>Panellists: <i>all speakers, Sergi Beltran (CNAG) (To Be Confirmed), Ronald Cornet (AMC) (To Be Confirmed)</i></p>
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May 27, 13h30 – 15h30 CEST

Building research capacity through training and mentoring

Chair/co-chair

Anton Ussi (EATRIS) & Birute Tumiene (VUHSK)

Objectives

- Learn about EJP RD training and mentoring programme's impact
- Demonstrate how training and mentoring have an impact on onboarding research, in developing research projects, networks as well as on research efficiency.
- Build upon successful examples for the upcoming ERDERA partnership.
- Showcase examples of trainees and mentees who have been able to initiate or pursue research thanks to the support provided by EJP RD in terms of training/mentoring.

Program of the session

Time (CEST)	Title	Speaker
13:30-13:40	EJP RD training and mentoring activities overview	Roseline Favresse (EURORDIS) & Anton Ussi (EATRIS)
13:40-13:50	Case example: fellow having applied to NSS successfully - SCN8A and SCN2A European Network Conference	Vincenza Scarcelli (Associazione SCN8A Italia ODV)
13:50-14:00	Case example: mentoring fellow	Paula Videira (Universidade NOVA de Lisboa)
14:00-14:10	Case example: ERN mobility fellow	Aleksandre Tavadze (Medical Center Mrcheveli)
14:10-14:20	Case example: WP14 train the trainer workshop?	Rasa Traberg (LHUS hospital Kauno klinikos)
14:20-14:30	EJP RD MOOC mentor and past trainee	Marie-Charlotte Villy (Institut Curie)
14:30-15:30	Roundtable with panellists	All above speakers + panellists: Franz Schaeffer (UKL-HD), Harm Peter

May 27, 16h00 – 17h30 CEST

Variety is the spice of life – how EJP RD research funding addresses a multitude of stakeholders and topics

Chair/co-chair

Sonja van Weely (ZonMw), Ralph Schuster (DLR)

Objectives

- To share success stories in several EJP RD funding schemes from different stakeholder and research topic perspectives
- To discuss the lessons learned and needs to improve research.

Description

Several research funding schemes have been implemented in the EJP RD. The Joint Transnational Calls aim to fund European consortia to perform research on specific topics and to improve the involvement of patients in these projects. The Networking Support Scheme aims to support networking between clinicians, researchers and patients and to better include stakeholders from underrepresented countries. The Rare Diseases Research Challenges is a funding scheme to encourage public-private partnerships in research. Successful examples of these funding schemes will be presented in this session next to lessons learned and challenges to overcome.

Program of the session

Time (CEST)	Title	Speaker
16.00 - 16.10	Overview research funding	Ralph Schuster (DLR)
16.10 - 16.20	Impact of patient leadership and guidance in consortium and proposal development	Michela Onali (Gli Equilibranti HIBM)
16.20 - 16.30	Congenital Disorders of Glycosylation – A New Generation	Hana Hansikova (CUNI)
16.30 - 16.40	Building a better mousetrap: the TRX assay for triplet repeat expansions	Robert Lahue (University of Galway)
16.40 - 16.50	Social and psychological long-term impact of autoimmune encephalitis: Advocating for Multinational Collaboration and Patient Involvement	Carsten Finke (CHARITE)
16.50 - 17.30	Panel discussion – lessons learned, needs to improve research possibilities	All speakers, Sonja van Weely (ZonMw), Christine Fetro (FFRD), H�el�ene Le Borgne (EC)

May 27, 17h45 – 19h15 CEST

Impact of EJP RD in clinical research

Chair/co-chair

Franz Schaefer (UKL-HD), Rima Nabbout (AP-HP)

Objectives

- To showcase the wide range of clinical research funding activities applied in EJP RD
- To review researchers' experience with the different funding formats
- To appraise the efficacy and impact of clinical research funding provided by EJP RD

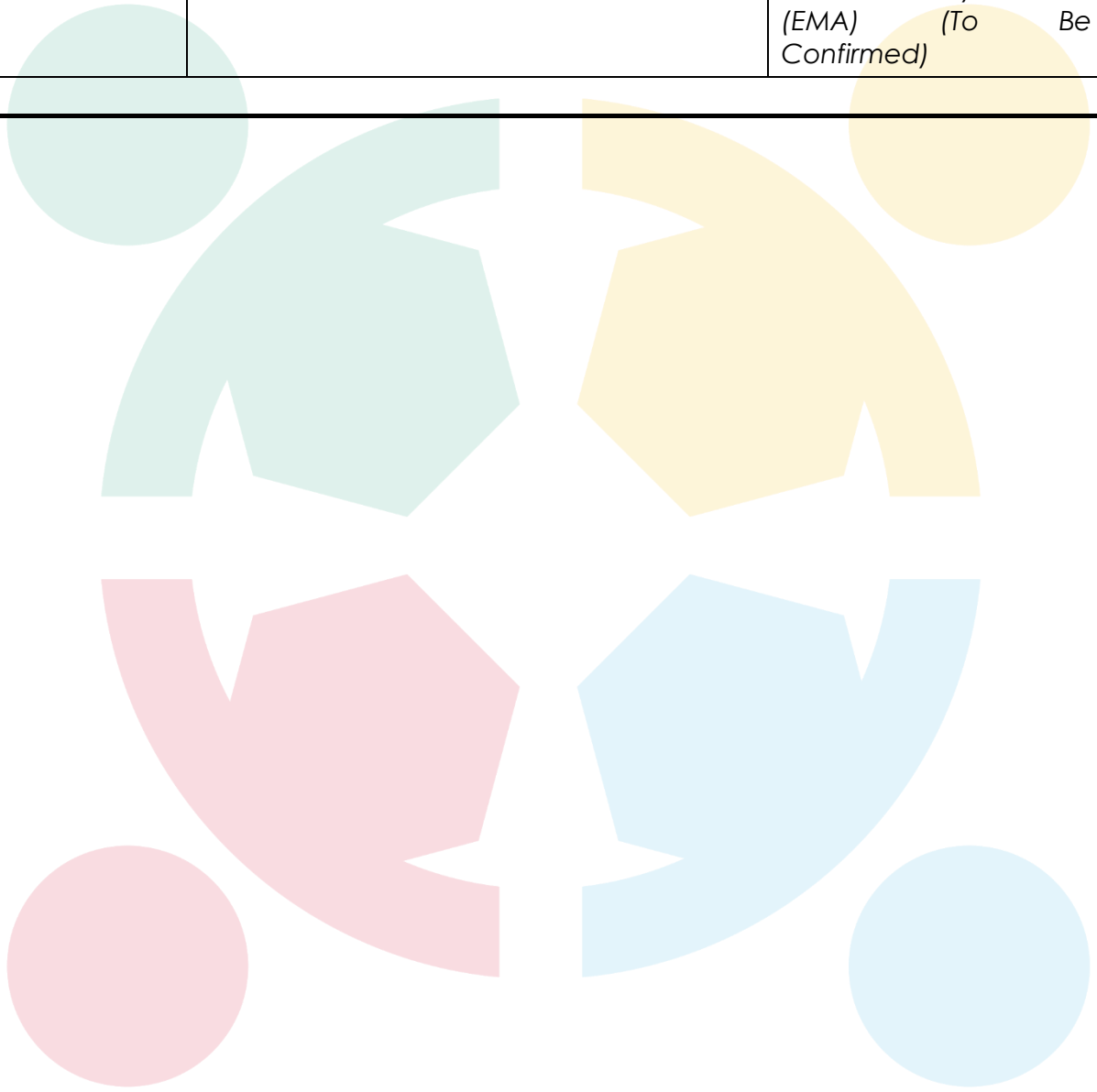
Description

The session will use illustrative examples to provide an overview of the range of formats by which clinical research was promoted in EJP RD. The efficacy and impact of the different funding streams will be discussed.

Program of the session

Time (CEST)	Title	Speaker
17.30-17.40	Overview clinical research	Franz Schaefer (UKL-HD), Rima Nabbout (AP-HP)
17.40-17.50	<i>Novel therapy development for APS1</i>	Speaker To Be Confirmed
17.50-18.00	LAMA2 Muscular Dystrophy: Paving the road to therapy	Bert Smeets (Maastricht University)
18.00-18.10	Congenital Anomalies of the Kidneys and Urinary Tract: Novel etiological concepts emerging from gene network analysis	Franz Schaefer (UKL-HD)
18.10-18.20	IDEAL-EPISTOP: clinical trials data reuse and re-analysis in rare diseases to increase the results robustness: the example of a collaboration EPISTOP-IDEAL in Tuberous sclerosis extended beyond EU	Katarzyna Kotulska (IPCZD)
18.20-18.30	Novel statistical approaches for Individual outcome prediction: The example of	<i>Mathys Synofzik (Universitätsklinikum</i>

	hereditary movement disorders (from WP20)	<i>Tübingen) (Speaker To Be confirmed)</i>
18.30-19.00	Panel discussion – lessons learned: Efficacy and impact of clinical research funding instruments in EJP RD Role of EJP RD to research within and outside ERNs	All speakers, Alexandre Méjat (AFM), Sheela Upadhyaya (Together4RD), Carla Fladrowski (TSC EU) (To Be Confirmed), Kit Roes (EMA) (To Be Confirmed)



May 28, 9h00 – 11h00 CEST

Health and research policy - collaboration perspectives

Chair/co-chair

Clément Moreau (INSERM), Viviana Giannuzzi (FGB)

Objectives

- Demonstrate the EJP RD work and role in the health policy framework
- To focus on the national Level and provide examples on how to influence national strategies
- To share results from EJP RD as a framework for inclusion of underrepresented countries and comparison to other EU programmes and future prospects
- To discuss on possible impacts of future EU initiatives

Description

This session is meant as a multi-stakeholder forum where EU and national authority representatives, Policy Board Members, researchers and patients share their experience on shaping health policies and expectations.

Program of the session

Time (CEST)	Title	Speaker
9:00 – 9:10	The impact of EJP RD outcomes in EU health policies and public consultations	Annalisa Landi (FGB)
9:10 – 9:25	Impact of the EJP RD National Mirror Groups on the development of national strategies: the example of France	Anne-Sophie Lapointe (French Ministry of Health)
9:25 – 9:40	The inclusion of underrepresented countries in EU programmes and future perspectives	Anabela Isidro (AICIB)
9:40 – 9:55	The point of view of patients on the impact of future EU initiatives and programme in RDs	Dimitrios Athanasiou (World Duchenne Organization)
9:55 – 10:10	Exploring how to have engaging, inspiring, and useful communication between Rare Diseases stakeholders	Claudio Cinquemani (CureRare)

10:10 – 10:25	Factors that establish European research programmes as decision-making tools in public health policy: perspectives for ERDERA	Yanna Petton (INSERM)
10:25 – 11:00	Discussion	Panelists: Paola Facchin (Azienda Ospedaliera di Padova) Dimitrios Athanasiou (World Duchenne Organization) Claudio Cinquemani (CureRare) Zivile Ruželè (LMT) H��l��ne Le Borgne (EC)

May 28, 11h30 – 13h00 CEST

From collaboration to enterprise

Chair

Anton Ussi (EATRIS)

Objectives

Look at the EJP RD deliverables through the industry lens and discuss path to scale and how collaboration within ERDERA can facilitate deployment in the industrial research and regulatory context.

Description

This session will build on the presentation of key achievements in the previous sessions and discuss in more detail

- the impact or potential impact of these tools on de-risking and enabling research at scale, regulatory and HTA submissions.
- Path to scale and deployment – what is ready? What needs more work that ERDERA can do?
- Most effective ways to collaborate (different models and their value)?

Program of the session

Time (CEST)	Title	Speaker
11:30 – 11:50	Presentation of 4 most relevant EJP RD tools that can de-risk and accelerate R&D in industrial and regulatory context	Daria Julkowska (INSERM)
11:50 – 12:50	Panel discussion: four 15-min rounds to look at each deliverable and discuss value and limitations in the industrial and regulatory context, what would it take to deploy them and what ERDERA can do and where public private collaboration can be most helpful for achieving that goal: <ul style="list-style-type: none"> - The Rare Diseases Virtual Platform - "Innovation Management Toolbox & translational mentoring services" - The Customised expert Training support service - Innovation enablers community (supporting fundamental science, enabling translation, regulatory compliance and industrialisation) 	Moderator: Anton Ussi (EATRIS) Panellists: <ul style="list-style-type: none"> - Ruxandra Draghia (Johnson & Johnson Innovative Medicine) - Solange Rohou (AZ) - Tom Metcalfe (Roche) - Alexandre Méjat (AFM) - Malwina Mejer (CE) - Giorgio Iotti (Chiesi)
12:50 – 13:00	Conclusions and recommendations for ERDERA	Panellists and moderators