What's new

this month?

July 2021

EJP RD HIGHLIGHTS

JULY 8TH

Strategy Meeting: Alignment of national rare disease strategies with the EJP RD

REGISTRATION DEADLINE: July 3rd

EJP RD is organising an integrative research and innovation strategy meeting on the theme of alignment of national rare disease strategies with the EJP RD. The fully online meeting will take place on **8th July 2021** from 09:30 to 17:00 CET. The workshop will focus on:

- Presentation of EJP RD advancement and analysis of national actions
- Review of country-level best practices and challenges identified from a survey
- Discussion of the way forward.

Online registration for the event is open until **July 3rd** at the link **here**.

More information

JULY 1ST

Webinar: Digitalization of the inborn errors of metabolism pathways

In this webinar (**July 1st**), **Denise Slenter** (Bioinformatics Department, Maastricht University) will showcase her project on **digitizing pathway figures** from the medical textbook *"Physician's guide to the diagnosis, treatment, and follow-up of inherited metabolic diseases"* by **N. Blau et al. 2019**. The **pathway drawings have been converted to fully machine readable pathway models, annotated with identifiers for proteins, metabolites and metabolic conversions**, which allows for analysis of various types of omics data.

More information

JULY 22ND

Resource Webinar: MetaboLights

As part of the **EJP RD Resource Webinar series**, the next webinar will be dedicated to **MetaboLights** and will be held on **July 22nd**.

MetaboLights is a **database for Metabolomics experiments** and derived information that is cross-species, cross-technique and covers metabolite structures and their reference spectra as well as their biological roles, locations and concentrations, and experimental data from metabolic experiments. MetaboLights is the recommended Metabolomics repository for several leading journals. Participants will be introduced to metabolomics and the EMBL-EBI MetaboLights resource. This will include the different types of data in the repository and how to search and access the data.

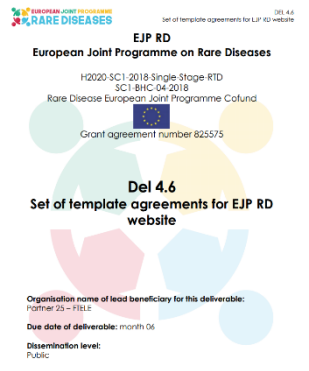
More information

EJP RD DELIVERABLE

EJP RD Template Agreements

EJP RD has released a **public deliverable** that reports progress on the **management of legal and IPR issues**:

- Preparation of an annotated template for a **Material and Data Transfer Agreement**;
- Preparation of an annotated template for an **Inter Institutional Agreement for the management of jointly owned results** generated by two or more beneficiaries within the action.



These templates can be consulted in the annexes of the deliverable.

More information

JULY 31ST

5th European Aniridia Conference

European **Aniridia** Conference

The 5th European Aniridia Conference is being organised to enable the **sharing of scientific knowledge about the rare genetic eye condition aniridia** to prevent sight loss and deal with aniridia's effects. This workshop has received funding support from the EJP RD's **Networking Support Scheme (NSS) funding opportunity**.

The fully online conference will take place over two days from **July 31st – August 1st** from 10.00 – 18.30 CET.

Online registration for the conference is currently open.

More information

FORTHCOMING

International Summer School on Rare Disease Registries and FAIRification of Data

REGISTRATION DEADLINE: July 11th

As part of the training activities proposed by the EJP RD, the **International Summer School on Rare Disease Registries and FAIRification of Data** is a **5-day online training programme** organised by Istituto Superiore di Sanità (ISS) in close collaboration with EJP-RD task partners, aimed at the international research community, clinicians,

medical specialists, registry curators, database managers, healthcare professionals and patients' representatives.

The fully online event will take place from **September 27th – October 1st** and is composed of **two learning modules**: a **three-day module** (**September 27th–29th**) that will cover rare disease registries and a **two-day module** (**September 30th–October 1st**) on the FAIRification of data.

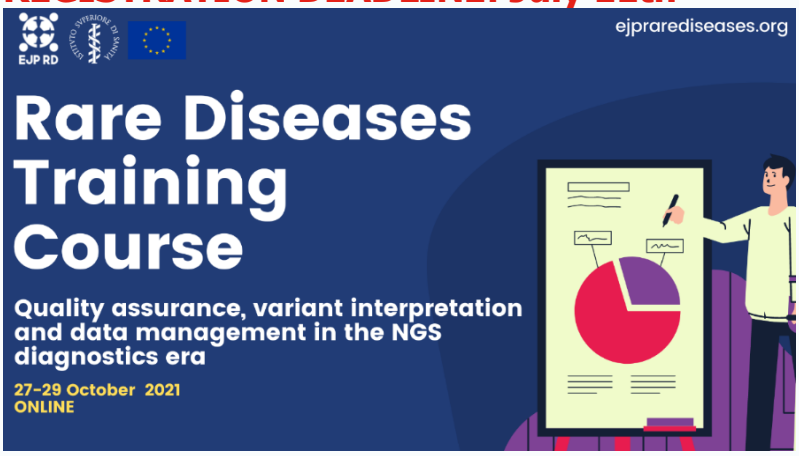
Online registration for the event is open until **July 11th** at the link **here**.

More information

FORTHCOMING

3rd Training Course: "Quality assurance, variant interpretation and data management in the NGS diagnostics era"

REGISTRATION DEADLINE: July 11th



As part of the training activities proposed by the EJP RD, a **3-day training course** titled **"Quality assurance, variant interpretation and data management in the NGS diagnostics era"** is being organised by Istituto Superiore di Sanità (ISS) in close collaboration with EJP-RD task partners. The course will build on expertise gained by EuroGentest and help in the **translation of research tools to diagnostic applications** (in line with the IRDiRC objectives). The impact is on the **quality and reliability of NGS results** obtained through rare disease research.

The fully online event will take place from **October 27th – 29th**.

Online registration for the event is open until **July 11th** at the link **here**.

More information

FORTHCOMING

Online Training: Orphanet nomenclature and Orphanet Rare Disease Ontology (ORDO) for clinicians/researchers

REGISTRATION DEADLINE: July 16th

A **one-day online training** workshop on **Orphanet nomenclature and Orphanet Rare Disease Ontology (ORDO)** is being organised by the Orphanet Network in close collaboration with EJP-RD task partners. This training is aimed at **improving knowledge and basic skills about Orphanet nomenclature** (including the ORPHA codes) and Orphanet Rare Disease Ontology (ORDO) for clinicians/researchers, and for the IT teams involved. The fully online event will take place on **September 10th**.

Please note that the training will be conducted in the Italian language.

Online registration for the event is open until **July 16th** at the link **here**.

More information

EJP RD FUNDING OPPORTUNITIES

Next collection date: September 2nd

Networking Support Scheme (NSS) Funding Opportunity
Next collection date: September 2nd, 2021

UPDATE: The NSS has been expanded to include online and hybrid networking events that can now be funded in addition to face-to-face events. A hybrid networking event consists of a group of participants networking face-to-face at a specific location together with other participants networking online.

The aim of the NSS call is **to encourage knowledge-sharing between health care professionals, researchers and patients** on rare diseases and rare cancers, as well as **to enable or increase the participation of usually underrepresented countries in Europe in new and existing research networks**. Eligible applicants are **health care professionals, researchers, and patient advocacy organisations** from the following countries involved in the EJP RD: Armenia, Austria, Belgium, Bulgaria, Croatia, Czech Republic, Denmark, Estonia, Finland, France, Germany, Georgia, Greece, Hungary, Ireland, Israel, Italy, Latvia, Lithuania, Luxembourg, Malta, Norway, Poland, Portugal, Romania, Serbia, Slovakia, Slovenia, Spain, Sweden, Switzerland, the Netherlands, Turkey, United Kingdom. There is no limit on the number of participants per event; however, the maximum budget that can be requested is **€ 30,000 per networking event**.

The next collection date is September 2nd at 14:00 (CET).

Selected past networking events are available **here**.

To get more information and to apply, click below.

More information

All EJP RD open funding opportunities here

EJP RD IN EVENTS

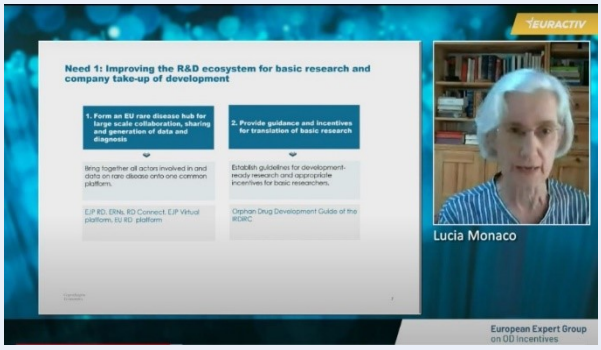
During the month of June, EJP RD was presented at the following events:

- During the **Third International Conference on Quality in Medicine, Global Health and Law** titled *"The Role of Interprofessional Collaboration in Improving Quality of Care in the Time of Pandemic"* **(June 15th).** Read more about this event here.
- During a meeting titled **"L'avenir des maladies rares en Europe - Révision du règlement européen et impact pour l'ensemble des acteurs français"** (**June 30th**) organised by France Biotech and the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE). The objective of this meeting is to discuss the ongoing review of the OMP Regulation from a French perspective and its impact on national policies for rare diseases and orphan medicinal products (OMPs).

NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

Orphan Drug (OD) Expert Group Webinar

During a webinar held on **June 15th**, **Dr. Lucia Monaco**, the Chair of the IRDiRC Consortium Assembly, presented two of the recommendations of the the **European Expert Group on Orphan Drug Incentives (OD Expert Group)** on how to address the **unmet needs of rare disease patients** by transforming the European Orphan Medicinal Products (OMP) landscape.



More information

Leadership and Membership Changes

IRDiRC congratulates **Marc Dooms** and **Birutė Tumienė** for their election as **Vice-Chairs** of the **Interdisciplinary Scientific Committee (ISC)** and the **Diagnostic Scientific Committee (DSC)**, respectively.

More information

OTHER NEWS

EC publishes draft Strategic Research & Innovation Agenda (SRIA) for the Innovative Health Initiative (IHI)

As one of the joint undertakings under Horizon Europe, a **draft Strategic Research & Innovation Agenda (SRIA)** for the **Innovative Health Initiative (IHI)** has been **published** by the European Commission (EC). IHI will build on the successes of and lessons learnt from the Innovative Medicines Initiative (IMI). The goal of IHI is to help create an EU-wide health research and innovation ecosystem that facilitates the translation of scientific knowledge into tangible innovations, with a **total proposed budget of €2.4 billion**.

The Commission welcomes feedback on the draft SRIA from members of the rare disease community. The draft SRIA will become a binding document of the IHI upon formal establishment and adoption by the Governing Board. Interested parties can send feedback by email: RTD-IHI@ec.europa.eu.

More information

ERN-Skin rare skin disease webinar

The European Research Network for rare skin diseases (ERN-Skin) is launching a series of **scientific webinars on rare skin diseases**. The next webinar will be held on **July 6th from 13.00 – 14.00 CET** on the topic of **combined cell and gene therapy for Epidermolysis Bullosa**.

More information

Share4Rare 1st open call for patient-driven research projects

The Share4Rare Project announces its **1st open call for patient-driven research projects targeted towards patient organisations and researchers** in the field of rare diseases to apply for conducting their patient-centred studies within the platform. The call is currently open but will **close** on **July 15th at 17:00 CET**.

More information

RDCA-DAP Webinar: How the platform can help inform optimal trial design in progressive rare disease

As part of the Rare Disease Cures Accelerator–Data and Analytics Platform (RDCA-DAP) **webinar series on the use of rare disease person-level data in drug development and regulatory decision making**, a webinar on the theme of How RDCA-DAP can help inform optimal trial design in progressive rare disease is being organised. The webinar will be held on **July 21st from 18.00 – 19.00 CET**.

Case Study: **Northstar Ambulatory Assessment as an outcome assessment in Duchenne muscular dystrophy**

More information

Open call for research proposals: Rare diseases in times of a pandemic

The **Eva Luise and Horst Köhler Foundation** in Germany announces its "Rare Diseases in Times of a Pandemic" care research project for 2021 in collaboration with the Alliance of Chronic Rare Diseases (ACHSE e.V.). In the context of this project, a **call for proposals is currently open** and will **close** on **August 15th**. The funding framework includes a **maximum of € 70,000** and the duration of the project is **12 months**.

More information

Call for research projects: MTFMT gene and Leigh syndrome

The **French Foundation for Rare Diseases (FFRD)** in parternship with Association contre les Maladies Mitochondriales (AMMi) are supporting and stimulating **biomedical research on MTFMT gene and Leigh syndrome**. In the context of the partnership, a **call for research projects is open** to develop research on Leigh syndrome and the MTFMT gene but will **close** on **August 31st at 17:00 CET**. All biomedical disciplines are eligible. This call for research projects provides for **financial support of up to € 10,000** for a maximum duration of **18 months**.

More information

CAREERS

Job opportunities are available at EJP RD and its member institutions:

- Department of Bioinformatics – BiGCaT, Maastricht University is looking for a **Postdoctoral Researcher in Bioinformatics**
- EURORDIS-Rare Diseases Europe is looking for a **Governance Manager** and a **Corporate and Donor Relations Assistant**
- Institute of Human Genetics, University Hospital Heidelberg is offering a **post-doc position**

EJP RD has received funding from the

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