What's new
this month?

October–November 2021

EJP RD HIGHLIGHTS

EJP RD FUNDING OPPORTUNITY

ERN Research Mobility Fellowship

Open until November 28th

The **ERN Research Mobility Fellowship** is now open. The funding opportunity **aims to support PhD students, Postdocs and medical doctors in training to undertake scientific visits fostering specialist research training outside their countries of residence**.

The exchange can be carried out **(1) within the same ERN (Full Members and Affiliated Partners), (2) between different ERNs (Full Members and Affiliated Partners), and (3) between ERN Full Members / Affiliated Partners and non-ERN institutions**.

Either home or host (secondment) institution must be a Full Member or Affiliated Partner of an ERN at the time when the application is submitted, as well as during the proposed period of the training stay.

Successful applicants should acquire new competences and knowledge related to their research on rare diseases, with a defined research plan and demonstrable benefit to the ERN of the home and/or host institution.

The research mobility fellowships are meant to cover stays of 4 weeks to 6 months duration.

More information

NEW FUNDING CALL

European Commission (EC) opens funding call on development of new effective therapies for rare diseases

The European Commission (EC) has opened a new funding call titled **"Tackling diseases (Two Stage - 2022) (HORIZON-HLTH-2022-DISEASE-06-two-stage)"** in the context of the Horizon Europe Framework Programme on the development of new effective therapies for rare diseases.

The **call is currently accepting proposals covering several different stages in the continuum of the innovation pathway** (i.e., translational, pre-clinical, clinical research, validation in the clinical and/or real-world setting etc.), as relevant.

The funding call has the following deadlines: **February 1st, 2022**, and **September 6th, 2022.**

The EC recommends that proposals take stock of the FAIR guidance and other models and strategies developed by the EJP RD and relevant EU-funded projects. Therefore, the EJP RD has made available multiple services for prospective applicants.

More information

EJP RD DELIVERABLE

First version of the Virtual Platform Specification (VIPS)

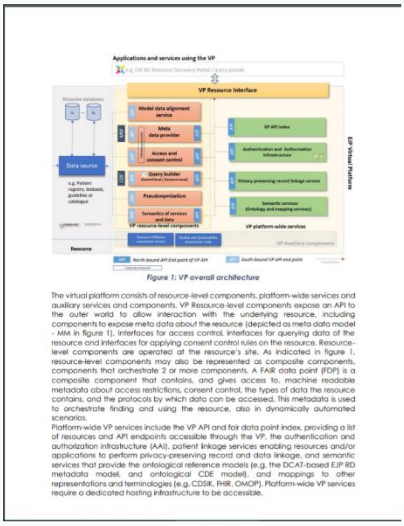

Figure 1: VP overall architecture

The virtual platform consists of resource-level components, platform-wide services and auxiliary services and components. VP Resource-level components expose an API to the outer world to allow interaction with the underlying resource, including components to expose meta data about the resource (depicted as meta data model - MM in figure 1), interfaces for access control, interfaces for querying data of the resource and interfaces for applying consent control rules on the resource. Resource-level components are operated at the resource's site. As indicated in figure 1, resource-level components may also be represented as composite components, components that orchestrate 2 or more components. A FAIR data point (FDP) is a composite component that contains, and gives access to, machine readable metadata about access restrictions, consent control, the types of data the resource contains, and the protocols by which data can be accessed. This metadata is used to orchestrate finding and using the resource, also in dynamically automated scenarios.

Platform-wide VP services include the VP API and fair data point index, providing a list of resources and API endpoints accessible through the VP, the authentication and authorization infrastructure (AAI), patient linkage services enabling resources and/or applications to perform privacy-preserving record and data linkage, and semantic services that provide the ontological reference models (e.g. the DCAT-based EJP RD metadata model, and ontological CDE model), and mappings to other representations and terminologies (e.g. CDISK, FHIR, OMOP). Platform-wide VP services require a dedicated hosting infrastructure to be accessible.

The first version of the **Virtual Platform Specification (VIPS)** has been published.

This deliverable **describes the overall architecture of the EJP RD virtual platform (VP) of data, resources, and tools**. It identifies and describes components and services provided to EJP RD resources including **services for FAIRification, data discovery, consent management and privacy preserving record linkage**.

The document **elaborates on standards for discovery, representation of data and meta-data, authentication as well as how VP components come to life** through an iterative use-case driven process, supported by quality and sustainability guidelines.

Please be advised that the current deliverable represents the first draft of the architecture that is now explored through the implementation of dedicated use-cases and pilots.

More information

ANNOUNCEMENT

First rare disease registry that is FAIR from its conception is now online!

All ERNs are tasked to set up patient registries that follow the FAIR Principles, as these 'FAIR registries' are essential for enabling efficient analysis of data across multiple sources. Existing methods to make clinical trial and registry data (more) machine-readable and FAIR are usually carried out after a research project is conducted and data are collected (post hoc).

The Registry of Vascular Anomalies (VASCA, part of VASCERN ERN) has implemented de novo FAIRification, thereby making all data FAIR automatically and in real-time upon collection. In practice, this means that all the hands-on work for the FAIRification is conducted before data collection. Subsequently, data is made FAIR through entering them into the Electronic Data Capture (EDC) system Castor EDC. This has the advantage that clinical data is made FAIR without any intervention from data management and data entry personnel. **Due to the generic approach and developed tooling, the VASCA working group believes that the method can be used in other registries and clinical trials as well.**

More information

OPEN CONSULTATION

Open consultation of the Strategic Research and Innovation Agenda (SRIA) of the European Partnership ERA4Health

Closes October 29th

The European Commission (EC) is inviting interested stakeholders to participate in the **open consultation of the Strategic Research and Innovation Agenda (SRIA) of the European Partnership "Fostering an ERA for Health" (ERA4Health)** by providing feedback through this **online survey**.

The deadline for providing feedback to the Draft SRIA is **October 29th, 2021.**

Relevant stakeholders include: decision makers, research and innovation funding bodies, relevant national and regional stakeholders and experts (e.g. Health and Care providers, Public Health experts, researchers, Health and Care Innovators, enterprises, civic and patient associations, Health and Care professionals and formal and informal carers associations).

More information

OCTOBER 27TH

Resource Webinar: Sample Catalogue & BBMRI-ERIC Directory & Negotiator

As part of the **EJP RD Resource Webinar series**, a webinar dedicated to **Sample Catalogue & BBMRI-ERIC Directory & Negotiator** will be held on **October 27th**. Participants will learn how to use the Sample Catalogue and the BBMRI-ERIC Directory to **identify biobanks and collections of samples of interest** for their research and how they can effectively negotiate access to these samples using the Negotiator. As a secondary objective, they will learn **how to list their own collections** in these tools to enable reuse of samples and data.

More information

NOVEMBER 9TH

Rare Conversations Conference – European Rare Disease Ecosystem: A Collaborative Path Forward

EJP RD is co-organising the "**Rare Conversations – European Rare Disease Ecosystem: A Collaborative Path Forward**" conference in cooperation with **Alexion**, **EURORDIS**, **EUCOPE**, and **EuropaBio**. The conference is targeted towards representatives of the different communities active in rare diseases: patients, researchers, clinicians, regulators, investors, payers, and industry.

The **fully online conference** will take place on **November 9, 2021** from **14.00 – 18.00 CET**.

This **high-level conference will be the occasion to discuss the rare disease ecosystem in its whole spectrum of policies and stakeholders at the national, EU and international level**. Starting from a general discussion on how to enable the ecosystem and needs to fulfil, the conference will then develop through three different areas: Research and Development, regulatory approval, and access.

More information

OCTOBER 28TH & NOVEMBER 4TH

Drug repurposing for rare disease workshop series

EJP RD is co-organising a **series of 3 workshops dedicated to drug repurposing for rare diseases**. Registration is open here. Registration is free but places are limited.

Webinar #1 (concluded October 21st): An introduction to drug repurposing for rare diseases – the benefits, the process and patient perspective

Webinar #2 (**October 28th**): Towards commercialization of a repurposed drug – patenting strategies, access to data, active ingredients, and collaborations between academia, patients and industry

Webinar #3 (**November 4th**): Getting a repurposed drug the to the patient – regulatory processes, reimbursement and prescription scenarios for repurposed drugs

More information on Webinar 2
More information on Webinar 3

NEW EU RESEARCH PROJECT

Launch of new Innovative Medicines Initiative project: Screen4Care

October marked the **official launch of Screen4Care,** a new Innovative Medicines Initiative (IMI) project focused on **accelerating diagnosis for rare disease patients through genetic newborn screening and advanced analysis methods such as machine learning and Artificial Intelligence**. The project will run for a period of five years with a **total budget of EUR 25 million** provided by the Innovative Medicines Initiative (IMI 2 JU), a joint undertaking of the European Union (represented by the European Commission) and the European Federation of Pharmaceutical Industries and Associations (EFPIA).

More information

EJP RD FUNDING OPPORTUNITIES

Next collection date: December 2nd

Networking Support Scheme (NSS) Funding Opportunity
Next collection date: December 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 December 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 October, EJP RD is being presented at the following events:

- During the **VASCERN Days 2021** workshop **(October 8th) by Dr. Yanis Mimouni**
- During the **AOP Orphan Round Table** entitled ***Research to Market*** (**October 14th**) by Dr. Daria Julkowska

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NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

Call for experts for Primary Care Task Force

The IRDiRC Diagnostics Scientific Committee (DSC), Interdisciplinary Scientific Committee (ISC), and Funders Constituent Committee (FCC) have set up a **joint Task Force to identify challenges and opportunities in rare diseases research focusing on primary care**. IRDiRC is currently assembling a team of experts to populate this Task Force and is specifically looking for members with expertise/experience in one or more of the following areas:

- **Primary care health provider with experience/research in rare disease**
- **Involvement in strategic planning for rare disease research**

If you are interested in taking part in this Task Force, please send a short biography and letter of motivation (1 page) to the Scientific Secretariat (scisec-irdirc@ejprarediseases.org) before **October 29th**.

More information

New IRDiRC Recognized Resource: Cellosaurus

IRDiRC has recently accepted a new Recognized Resource, the **Cellosaurus**, a knowledge resource on cell lines aiming to describe all cell lines used in biomedical research. The Cellosaurus provides **information for about 130,000 cell lines and 25% of these cell lines are established from rare disease patients**. For each cell line the Cellosaurus provides a wealth of information, cross-references and literature citations. The Cellosaurus is available on the ExPASy server (**https://web.expasy.org/cellosaurus/**) and can be downloaded in a variety of formats.

More information

Leadership and Membership Changes

The IRDiRC **Consortium Assembly (CA)** is in the process of electing a new **Chair** and **Vice Chair**. The new leadership of the CA will be presented during the **next CA Meeting** to be held in Paris, France, and online (hybrid meeting) on **December 9th – 10th**.

More information

OTHER NEWS

BioData World Congress 2021: Big Data and Digital Transformation in Pharmaceuticals and Healthcare

The **BioData World Congress 2021** organised by **Terrapinn** is Europe's largest congress covering **big data in pharmaceutical development and healthcare** aimed to showcase innovation, demonstrate success and break through the obstacles and barriers to ensure that **innovations in genomics and big data enter the clinic with speed and efficiency**.

This **in-person event** will take place in **Basel, Switzerland** over three days from **November 2nd–4th**.

With over **250 senior executive speakers across 19 stages**, the Congress will feature the involvement of EJP RD members for the following sessions:

Day 1 (**Nov 2nd**): 17:20 Empowering Data Analysis: Lessons from Rare Diseases (Marco Roos, Chris Evelo, Sergi Beltran, Christine Durinx, Andrea Splendiani, Peter Goodhand [tbc])

Day 2 (**Nov 3rd**): 16:40 FAIR in Action – Successful Implementation of Principles (Marco Roos)

Day 3 (**Nov 4th**): 11:50 EJP RD – How creating the rare diseases research ecosystem can help in providing diagnosis to all RD patients within one year of coming to medical attention? (Dr. Daria Julkowska, Coordinator of the EJP RD)

More information

EATRIS Winter School 2021: Translational Medicine Explained (TMex)

EATRIS is organising the **TMex (Translational Medicine Explained) Winter School** from **November 8th – 12th** targeted to PhD students in the second half of their PhD, early postdocs and other **young scientists who are involved in biomedical research**. The TMex course (1) **provides a birds eye view of the medicine discovery & development process**; (2) **Raises awareness** of obstacles and challenges involved, and (3) **Introduces the different players** (academia, SME, pharma, regulatory) and career options.

More information

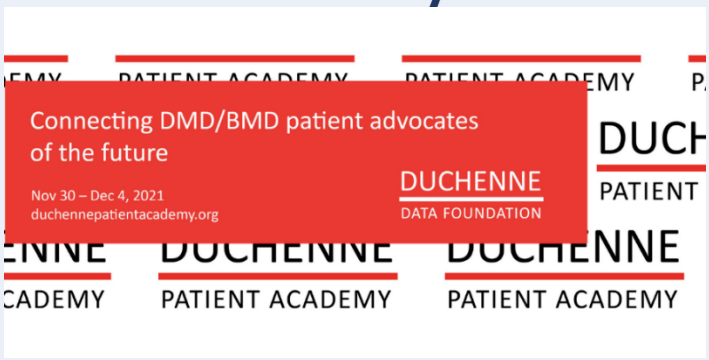
Virtual Congress: Europe Biobank Week 2021

The **European, Middle Eastern and African Society for Biopreservation and Biobanking (ESBB)** and the **European Research Infrastructure on Biobanking (BBMRI-ERIC)** are jointly organising the **Europe Biobank Week 2021** as a **Virtual Congress** with this year's theme "**Biobanking for our Future – Opportunities Unlocked**" over three days from **November 8th – 10th**.

The conference programme features a variety of workshops, presentations, and interactive industry sessions and covers themes like **regulatory implications, patient engagement, biological quality, paediatric biobanking, biobanking in personalised medicine, novel and advanced IT solutions for biobanking**, and so on.

More information

Applications open for Duchenne Patient Academy 2021



The **Duchenne Data Foundation**, in collaboration with the **World Duchenne Organization**, is launching the 2021 edition of the **Duchenne Patient Academy (DPA)** from **November 30th – December 4th**.

In this intensive training session, **patient advocates receive training and updates to build a strong base for current and future global advocacy**. Applications are now open and will close on **November 9th**.

More information

EHDEN 5th Open Call for Data Partners

The **EHDEN Consortium** announces its **5th Open call** targeted towards **data custodians** of Electronic Health Records, Claims, Hospital and Registry data across Europe, **supporting the mapping of their data to the OMOP common data model** to accelerate research and healthcare decision-making. The **current open call** will close on **November 15th**.

Data Partners can benefit from up to a **maximum of € 100,000 funding** from a **call budget of €3 million**.

More information

World Orphan Drug Congress 2021: Strategy, advocacy and partnering for the orphan drug industry

The **World Orphan Drug Congress** organised by **Terrapinn** is a **global, multi-stakeholder orphan drugs & rare diseases meeting** that aims to provide attendees with a **one-stop progressive scientific and strategic solution to the orphan drugs industry**.

The **in-person event** will take place in **Sitges (Barcelona), Spain** over four days from **November 15th – 18th**.

Patient groups can ask for free registration (subject to approval).

Dr. Daria Julkowska, Coordinator of the EJP RD, will moderate the panels on **"Responding to the EU OMP review – how to strengthen the EU rare disease ecosystem"** (**November 15th**) and **"The European Expert Group on Orphan Drug Incentives – How do we develop a sustainable European ecosystem?"** (**November 16th**).

More information

EMA Webinar on Clinical Trial Regulation and Clinical Trials Information System (CTIS)

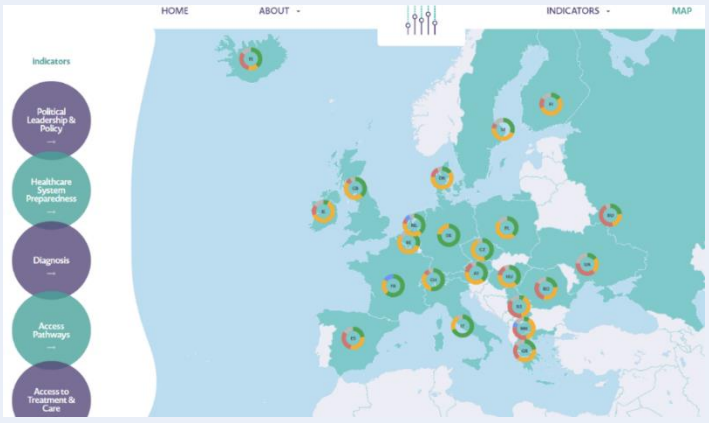
The **European Medicines Agency (EMA)** is organising a webinar for small and medium enterprises (SMEs) and academia on the **Clinical Trial Regulation (Regulation (EU) No 536/2014)** and the **Clinical Trials Information System (CTIS)**.

The webinar will take place on **November 29, 2021** from **09.00 – 13.15 CET**.

Attendance at the webinar is by registration only and open to companies that have been assigned SME status by EMA, academia and representatives of stakeholder organisations.

More information

Biogen introduces Spinal Muscular Atrophy (SMA) Policy and Access Trackers



Biogenhas collaborated with **SMA Europe** to develop a tool called the **Spinal Muscular Atrophy (SMA) Policy and Access Tracker**, which offers an **in-depth assessment of how 23 European countries are performing** in terms of access to SMA treatment, care and supporting policies, **identifies the major areas for improvement**, and **provides policy recommendations** to tackle existing gaps.

The Tracker has a **dedicated website** featuring an interactive map and a White Paper summarising findings and presenting targeted policy recommendations.

More information

C-Path and Pulse Infoframe establish patient-centered data harmonisation partnership

The **Critical Path Institute (C-Path)**, an independent nonprofit organisation, and **Pulse Infoframe**, a real-world evidence generation, health informatics and insights company, have announced their **collaboration to advance technologies and**

tools to further rare disease research and drug development. The two organisations will identify opportunities to **combine Pulse Infoframe's ambispective data with retrospective data in C-Path's Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP®)**, an FDA-funded initiative to support rare disease drug development.

More information

New Report: How patient organisations can drive FAIR data efforts to facilitate research and healthcare

Last March, the **World Duchenne Organization** in collaboration with **Duchenne Data Foundation** organised the second **Meeting on FAIR Data Sharing for Duchenne**. During this meeting, **120 participants from 22 countries discussed how they could drive FAIR data efforts to facilitate research and healthcare**.

The report of the second virtual meeting summarises the presentations and discussions of the meeting In addition to this, it provides an overview of the key lessons learned since the first meeting, and outlines the next steps.

More information

CAREERS

Job opportunities are available at EJP RD member institutions:

- The **EJP RD Coordination Team** (Paris, France) is looking for two **Scientific Project Managers for Rare Diseases**
- Centre for Molecular and Biomolecular Informatics (CMBI), Radboud UMC is looking for a **Post-doctoral Data Scientist**
- UNIVERSITAETSKLINIKUM AACHEN (UKA) is looking for a **Research Associate (Biostatistics)**
- EURORDIS is looking for an **Events Junior Manager** (Paris), **Communications Junior Manager** (Paris), **Patient Data Director** (Barcelona or Brussels), **Patient Engagement Junior Manager** (Barcelona) and a **Governance Manager** (Paris).

- Department of Genetics, AP-HP (Paris University Hospital Trust) is looking for a **Pedagogical Engineer/Moodle Developer**

EJP RD has received funding from the

European Union's Horizon 2020 research and innovation programme under GA N°825575