

What's new this month?

November–December 2021

EJP RD HIGHLIGHTS

EJP RD FUNDING OPPORTUNITY

Joint Transnational Call 2022: A funding opportunity for research projects

Call opens December 14th

Following the **successful implementation of three Joint Transnational Calls since 2019**, EJP RD is glad to announce the upcoming launch on **December 14th** of the fourth **EJP RD Joint Transnational Call (JTC) 2022**, aimed at **funding multilateral research projects on rare diseases under the EJP-COFUND action**.

The aim of the call is to **enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project**, with the call topic being **"Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases."**

An **information webinar** will be held on **December 16th** from **14.00-15.30 (CET)**.

Partners belonging to one of the following categories may request funding under a joint research proposal (according to country/regional regulations):



THE
EUROPEAN JOINT PROGRAMME
RARE DISEASES
OPENS THE
JOINT TRANSNATIONAL CALL 2022
A FUNDING OPPORTUNITY FOR RESEARCH PROJECTS

Try our matchmaking tool to find your ideal project partner!

14 DECEMBER: Launch of the call
14 DECEMBER: Information webinar for potential applicants
14 FEBRUARY: Pre-proposal submission deadline

Topic: Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases

ejprarediseases.org

- academia
- clinical/public health sector
- enterprises when allowed by national/regional regulations
- patient advocacy organizations.

The **maximum duration** of the project is **three years**. **Only transnational projects will be funded**. Each consortium submitting a proposal **must involve four to six eligible principal investigator partners from at least three different participating countries**.

[More information](#)

OPEN FUNDING CALL

EJP RD Support Services for Horizon Europe Call for Proposals: 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**.

SUPPORT FOR PROPOSALS FROM EJP RD

As part of EJP RD's support for rare disease research, **multiple EJP RD services are available** to aid in the above call for proposals:

- **Data FAIRification:** making resources Findable, Accessible, Interoperable and Reusable for humans and machines.
- **Clinical Studies Support Office** can assist with the design of clinical trials both in terms of project planning (site selection, cost evaluation etc) as well as study design (methodological and statistical methods).
- **Mentoring** for translational research to design proposals from bench to bedside. Our range of experts can assist on a number of topics and review proposals to increase their impact.

Depending on your needs, we may be able to assist with other services.

[More information](#)



DECEMBER 14TH

Training Webinar: Does Randomization matter in RD clinical trials?



As part of the [EJP RD Training series](#), an advanced-level training webinar on the topic "*Does Randomization matter in RD clinical trials?*" will be held on **December 14th**.

The training consists of two parts: a 1.5-hour training in less frequently applied randomization procedures and their

performance on the level of evidence, followed by 1 hour of panel discussion with experts from regulatory, pharmaceutical, academic, and clinical fields. The event is **open to the international research community, clinicians, medical specialists, healthcare professionals and advocacy patient groups** with knowledge of rare disease clinical trials.

Registration closes on **December 10th**.

[More information](#)

DECEMBER 15TH

Resource Webinar: INFRAFRONTIER

As part of the [EJP RD Resource Webinar series](#), a webinar dedicated to **INFRAFRONTIER** will be held on **December 15th**.

Participants will be given an overview of INFRAFRONTIER (the **European Research Infrastructure for the generation, phenotyping, archiving and distribution of model mammalian genomes**) and

the European Mouse Mutant Archive (EMMA) and what these can offer to rare disease researchers.

The webinar will include **live demos on searching for mouse models** for a certain rare disease and placing an online order, **submitting a mouse strain to EMMA for cryopreservation** or **looking for available phenotype information** for a certain mouse strain.



[More information](#)

DECEMBER 15TH

ERN Workshop on anonymisation and pseudonymisation of rare disease patients: Legal and technical aspects



The [European Rare Disease Research Coordination and Support](#)

[Action consortium \(ERICA\)](#) is organising a workshop on **anonymisation and pseudonymisation of rare disease patients: legal and technical aspects** as

the first workshop of the **ERN data strategy workshop series**, on **December 15th, 2021**.

The speakers include Spencer Gibson (University of Leicester), Guillaume Byk (DG-Santé – European Commission) and Antonio Cutillo (Joint Research Center). During this first workshop, the speakers will discuss the **techniques and algorithms to anonymise and pseudonymise healthcare data from patients with rare diseases**.

[More information](#)

UPCOMING ERN WORKSHOP

ERN Workshop on contemporary outcome measures in neuromuscular diseases



In the context of [EJP RD's ERN Workshops](#), a hybrid workshop on “**contemporary outcome measures in neuromuscular diseases**” aimed at **understanding the advancement of technologies with digital outcome measures in neuromuscular diseases** is being organised by Prof. Sabrina Sacconi of University Hospital Nice. The **hybrid event (in-person on site and online by videoconference)** will take place over two days on **March 4th – 5th, 2022** at the **Pasteur 2 Hospital in Galet, Nice, France**.

The workshop is open by **prior registration and selection** to different target groups, especially those who want to deepen their knowledge in clinical outcomes measures and innovative digital outcomes, consisting of **researchers and clinicians affiliated to an ERN Full Member or Affiliated Partner Institution, physiotherapists and students**. The training workshop is free of charge. The workshop will consist of

both **presentations by experts in the field of neuromuscular diseases** as well as **interactive panel discussions and quiz sessions** to train participants in performing and interpreting various outcome measures.

Registration closes on January 10th, 2022.

[More information](#)

UPCOMING ERN WORKSHOP

ERN Workshop on endocrine cancer: A challenge in adults and children

In the context of [EJP RD's ERN Workshops](#), a face-to-face workshop on “**Endocrine cancer: A challenge in adults and children**” aimed at **giving an update on the molecular background and clinical management of rare endocrine malignancies** is being organised by Prof. Barbara Jarzab of the M. Skłodowska-Curie National Research Institute.

The **in-person event** will take place on **May 4th, 2022** at the **M. Skłodowska-Curie National Research Institute in Gliwice, Poland.**

The workshop is open by **prior registration and selection to endocrinologists, oncologists, surgeons, radiotherapists, internists and pediatricians, pathologists, radiologists, nuclear medicine specialists who are employees of or affiliated to an ERN Full Member or affiliated Partner institution.**

The training workshop is free of charge. Travel and hotel expenses will be reimbursed for all selected (max 20) ERN participants. Lunch and dinner will be provided on site for all participants.

Registration closes on January 28th, 2022.

[More information](#)



ANNOUNCEMENT

Submissions open for EURORDIS Photo Award

The **EURORDIS Photo Award** is an opportunity to **visually express what it means to live with a rare disease** and to **share your story with the rare disease community and beyond.**

The contest is open to all nationalities, ages, and diseases.

Every year, hundreds of people from all around the world submit their photos, each reflecting the drive of people living with a rare disease.

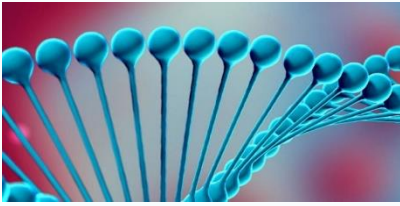
Submissions for the 2022 Photo Award will be open until January 16th, 2022.

[More information](#)



UPCOMING EVENT

Planning Committee named for the Third International Summit on Human Genome Editing (March 2022)



The [UK Royal Society](#), [Academy of Medical Sciences](#), the [US National Academies of Sciences and Medicine](#) and [UNESCO-The World Academy of Sciences for the advancement of science in developing countries \(TWAS\)](#) are jointly organising the **Third International Summit on Human Genome Editing**.

The **in-person event (with an option to attend online)** will take place in at the **Francis Crick Institute in London** over three days from **March 7th – 9th, 2022**.

We are pleased to announce that **Dr. Daria Julkowska, Coordinator of the EJP RD, is a member of the multidisciplinary planning committee** comprising representatives from eleven nations.

[More information](#)

EJP RD FUNDING OPPORTUNITIES

Next collection date: December 2nd

Networking Support Scheme (NSS) Funding Opportunity

Next collection date: March 1st, 2022

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 March 1st, 2022 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 November, EJP RD was presented at the following events:

- During the [Biodata World Congress \(November 2nd – 4th\)](#) by Dr. Daria Julkowska in a session entitled *"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?"*
- During the [Rare Conversations – European Rare Disease Ecosystem: A Collaborative Path Forward](#) conference ([November 9th](#)), an event co-organised by EJP RD
- During the [World Orphan Drug Congress 2021 \(November 15th – 18th\)](#) by Dr. Daria Julkowska
- During the [1st International Conference on Rare Diseases and Paediatric Research \(November 18th – 19th\)](#), an event supported by EJP RD



NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDiRC)

Working Group Opening: MedTech for Rare Diseases

IRDiRC's [Therapies Scientific Committee](#)

[\(TSC\)](#), [Interdisciplinary Scientific](#)

[Committee \(ISC\)](#) and the [University of](#)

[Twente](#) are jointly establishing a **Working**

Group to explore the role and value of

medical devices in rare diseases. The

Working Group aims to create a **better**

understanding and enhanced awareness

of device developer's needs, the

standardized outcomes to define user needs for devices, and to offer a groundwork for developing

solutions to improve the (regulatory) landscape of MedTech use for rare disease patients.

Interested candidates can **submit their application (CV, biosketch and letter of motivation, one paragraph each)**

to the Scientific Secretariat before [December 14th](#).

Do you want to explore the role and value of medical devices in rare diseases?

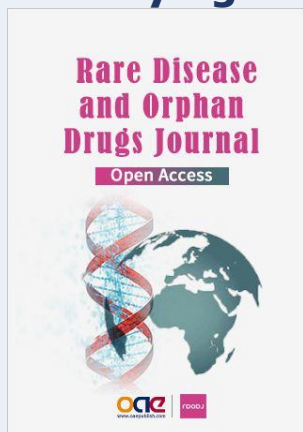
New IRDiRC Working Group

on MedTech for Rare Diseases

Apply before 14 December

[More information](#)

Publication: 1000 new rare diseases treatments by 2027, identifying and bringing forward strategic actions



A manuscript entitled **IRDiRC: 1000 new rare diseases treatments by 2027, identifying and bringing forward strategic actions** has been published in the open-access **Rare Disease and Orphan Drugs Journal**.

The manuscript describes the recent key steps undertaken by the IRDiRC **Therapies Scientific Committee (TSC)** to support the future approval of 1000 new therapies for rare diseases.

[More information](#)

Leadership and Membership Changes

The IRDiRC **Consortium Assembly (CA)** has now completed the process of electing a new **Chair** and **Vice Chair** for a **three-year term** beginning **January 01, 2022**. The new leadership of the CA is being presented to IRDiRC members during the **Consortium Assembly Meeting** being held in Paris, France, and online (hybrid meeting) on **December 9th – 10th**.

IRDiRC warmly thanks the current Consortium Assembly Chair and Vice Chair, Lucia Monaco and David Pearce respectively, as their terms draw to a close at the end of December 2021.

[More information](#)

OTHER NEWS

XIV Foresight Training Course (FTC): “The health emergency: Regulatory crash and future perspectives”

The **Fondazione Gianni Benzi** is organizing the **XIV Foresight Training Course (FTC)** on the theme of “**The health emergency: Regulatory crash and future perspectives**” as a virtual meeting that will take place on **December 10th, 2021** from **11.20 – 18.30 CET**. The event will focus on the **extraordinary regulatory measures put in place by the international and national authorities** to face the emergency, the lessons learnt from the pandemic and their impact on future actions, and an **insight on the value of the use and sharing of health data**.



[More information](#)

AKU Society launches crowdfunding campaign for Black Bone Disease



The [AKU Society](#), a UK-based charity, has launched a crowdfunding campaign to **raise funds for “Leave No Patient Behind – A Global Black Bone Disease Registry”** in order to gather information about alkaptonuria (AKU) patients around the world in one single place. The funds raised will allow the AKU Society to **make the case for access to the life-changing drug nitisinone** in the many countries where it is

not yet available. It will also help **prepare plans for gene therapy** and other new treatment studies for alkaptonuria, also known as AKU or Black Bone Disease.

[More information](#)

EMA publishes guideline on registry-based studies

The [European Medicines Agency \(EMA\)](#) has announced the publication of a **guideline on registry-based studies** following its adoption by the EMA’s cross-Committees Task Force on registries and the [Committee for Medicinal Products for Human Use \(CHMP\)](#).



The guideline provides pharmaceutical organisations with **key methods and good regulatory practices on the planning and conduct of studies using patients registries’ data collection infrastructure or population** to inform on medicines’ impact on public health. Additionally, the Annex proposes **aspects of good practice in the establishment and management of patient registries** considered relevant to their use for registry-based studies and other possible regulatory purposes.

[More information](#)

EMA launches pilot project on drug repurposing



The [European Medicines Agency \(EMA\)](#) and the [Heads of Medicines Agencies \(HMA\)](#) are launching a pilot project to support the repurposing of medicines. The aim of this initiative is to **support not-for-profit organisations and academia to gather or generate sufficient evidence on the use of an established medicine in a**

new indication with the view to have this new use formally authorised by a regulatory authority. The pilot is open to not-for-profit stakeholders and academia.

The EMA is **accepting submissions from sponsors** until **February 28th, 2022**.

[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** and a **Project Assistant (bilingual French and English)**
- 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**



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