

What's new this month?

January 2022

EJP RD HIGHLIGHTS

EJP RD FUNDING OPPORTUNITY

Joint Transnational Call 2022: A funding opportunity for research projects

Pre-proposal submission deadline: February 16th

The fourth **EJP RD Joint Transnational Call (JTC) 2022**, aimed at **funding multilateral research projects on rare diseases under the EJP-COFUND action**, is currently open.

Call topic: "**Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases**"

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

- 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



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
16 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

submitting a proposal **must involve four to six eligible principal investigator partners from at least three different participating countries.**

The pre-proposal submission deadline is [February 16th.](#)

[More information](#)

OPEN FUNDING CALL

Innovative Health Initiative (IHI) announces upcoming funding opportunities for 2022

The [Innovative Health Initiative \(IHI\)](#), a European Union public-private partnership funding health research and innovation, has published **draft information on upcoming funding opportunities in 2022** in order to give potential applicants additional time to find or build a consortium and prepare a strong proposal.

The first call for proposals will be launched in the first half of 2022. The following draft themes could be included in the future calls:

Single stage calls

- Innovative patient-facing care pathways for patients with neurodegenerative diseases and comorbidities
- Next generation imaging and image-guided diagnosis and therapy for cancer
- Precision oncology: Innovative patient-centric, multi-modal therapies against cancer
- Access and integration of heterogeneous health data for improved health care in diseases areas of high unmet public health need.

Two stage calls

- New tools for prediction, prevention and monitoring of cardio-metabolic diseases including secondary manifestations to enable timely intervention
- Strengthening EU clinical development excellence and innovation attractiveness: Harmonised methodology to promote the uptake of early feasibility studies (EFS)

Please check the IHI website and social media for regular updates related to the upcoming calls.

[More information](#)

NETWORKING SUPPORT SCHEME (NSS)-FUNDED EVENT

Networking event on vascular liver diseases and associated complications





A **funded networking event on vascular liver diseases and associated complications** is being organised to give **an update on current clinical and scientific concepts**, as well as open questions in the field of vascular liver diseases. This workshop has received funding support from the EJP RD's [Networking Support Scheme \(NSS\)](#) funding opportunity.

The **hybrid meeting** will take place over two days on **February 3rd – 4th** both **online** and at the **Medical University Vienna, Vienna, Austria**.

During these two days, specialists in hematology, hepatology, pathology and (interventional) radiology will not only provide **state-of-the-art lectures** but will also participate in **case discussions** in order to improve patient management and to develop multinational strategies to answer current scientific questions.

[More information](#)

NETWORKING SUPPORT SCHEME (NSS)-FUNDED EVENT

Networking event – The anti-IgLON5 disease: Challenges in diagnosis, treatment and pathogenesis of the disease



An **EJP RD funded networking event** on the **rare neurological disease caused by anti-IgLON5 antibodies**, discovered in 2014 by researchers from the Hospital Clínic-IDIBAPS, is being organised in the context of the [Networking Support Scheme \(NSS\)](#) funding opportunity.

The **in-person meeting** will take place over two days on **February 24th – 25th** at the Esteve Auditorium of the **Esther Koplowitz Center, Barcelona, Spain**.

Spain.

[More information](#)

UPCOMING ERN WORKSHOP

Trans-ERN Working Group for spina bifida (spinal dysraphism): Workshop for future research on innovative diagnostics and interdisciplinary treatment

Registration Deadline: February 4th

In the context of [EJP RD's ERN Workshops](#), an in-person workshop titled "**Trans-ERN Working Group for Spina Bifida (Spinal Dysraphism): Workshop for future research on innovative diagnostics and interdisciplinary treatment**" aimed at **sharing participants' expertise on research in different areas of spinal dysraphism and opening the way to new research projects** is being organised by Centre de Référence Spina Bifida – Dysraphismes C-MAVEM, Centre Hospitalier Universitaire de Rennes.

The **in-person workshop** will take place over two days on **March 31st – April 1st** at the **Centre Hospitalier Universitaire de Rennes** in **Rennes, France**.

The workshop is **open by prior registration and selection to geneticists, fetal medicine experts, neurosurgeons, rehabilitation medicine specialists, pediatric and adult urologists, pediatricians** who are employees of or affiliated to an ERN-Full Member or affiliated Partner institution.

The training workshop is free of charge. The workshop will consist of **interactive presentations and discussions** on different areas of interest.

Registration closes on February 4th and those selected to participate from among the applicants will be informed by **February 15th** of their selection.

[More information](#)

UPCOMING ERN WORKSHOP

ERN Workshop on contemporary outcome measures in neuromuscular diseases

Deadline extended to February 22nd

The poster is green with white and yellow text. At the top left is the EJP RD logo. The main title 'WORKSHOP' is in large yellow letters. Below it, the workshop title is written in white. The dates '31 MARCH - 1 APRIL 2022' are in yellow. The location 'Centre Hospitalier Universitaire de Rennes' and address are in white. At the bottom, there are logos for the European Joint Programme for Rare Diseases, the European Union flag, and the European Reference Network. An illustration of a person pointing at a whiteboard is on the right side.

WORKSHOP
TRANS-ERN WORKING GROUP FOR SPINA BIFIDA (SPINAL DYSRAPHISM): WORKSHOP FOR FUTURE RESEARCH ON INNOVATIVE DIAGNOSTICS AND INTERDISCIPLINARY TREATMENT
Organizers: Andrea Manunta & Giovanni Mosiello
31 MARCH - 1 APRIL 2022
Centre Hospitalier Universitaire de Rennes
2 Rue Henri le Guilloux
35033 Rennes (FRANCE)

ERN WORKSHOPS
THE EJP RD "ERN RESEARCH TRAINING WORKSHOPS CALL"

EUROPEAN JOINT PROGRAMME FOR RARE DISEASES
European Reference Network



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** 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 for physical attendance is closed but registration for online attendance has been extended to February 22nd.

[More information](#)

UPCOMING ERN WORKSHOP

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

Deadline extended to March 7th

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** 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 has been extended to [March 7th](#).



[More information](#)

RARE DISEASES TRAINING COURSE

Training on strategies to foster solutions of undiagnosed rare disease cases



As part of the [Training activities proposed by EJP RD](#), an online training course on "**Training on strategies to foster solutions of undiagnosed rare disease cases**" is being organised by Istituto Superiore di Sanità (ISS) in close collaboration with EJP RD partners. The **online event** will take place **over three days** from on **[April 11th – 13th](#)**.

Through the presentation of sample use cases that have long eluded diagnosis, the **course will provide participants with with useful tools, instruments and knowledge on novel strategies to foster solutions of undiagnosed rare diseases cases**. Moreover, the course will facilitate networking among professionals involved in undiagnosed rare conditions.

The **course is open to the international research community, to clinicians and to medical specialists** who have experience and concrete interest in the diagnosis and research on rare diseases.

To ensure active participation and exchange with teaching staff and participants, a **maximum of 30 attendees** will be admitted.

Registration closes on March 6th.

[More information](#)

ANNOUNCEMENT

EURORDIS Black Pearl Awards Ceremony 2022

The **EURORDIS Black Pearl Awards** celebrate the **inspirational qualities of people living with a rare disease** along with those who go that extra mile to make a difference to their lives.

The eleventh edition of the Awards will take

place **online** on **February 8th** from **18:00 – 19:30 CET** and

will bring together persons living with a rare disease, patient advocates, policy makers, scientists, healthcare professionals, industry representatives, and more.



[More information](#)

ANNOUNCEMENT

Update: Third International Summit on Human Genome Editing postponed to 2023



The **Third International Summit on Human Genome Editing**, organised jointly by 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) has been postponed from its originally scheduled date of March 2022.

The **in-person event (with an option to attend online)** will take now place in **2023** at the **Francis Crick Institute in London** over three days from **March 6th – 8th, 2023**.

The original meeting window of **March 7th – 9th, 2022** will be used to host a number of small, themed online discussions on priority areas that will inform the Summit agenda for 2023.

[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 months of November-December 2021, EJP RD was presented at the following events:

- During the EJP RD-organized [TEDDY paediatric expert patients training workshop \(November 26th\)](#) by Clément Moreau
- During the [BBMRI-ERIC Fourth National Scientific and Training Conference of Polish Biobanks \(December 6th – 7th, 2021\)](#) by Dr. Daria Julkowska



NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

Call for Members: Task Force on Disregarded Rare Diseases (PLUTO PROJECT)

IRDiRC's [Therapies Scientific Committee](#)

(TSC) is establishing a **Task Force to characterize specific commonalities amongst a large group of "disregarded" rare diseases**, with the potential secondary aims to identify removable roadblocks that may foster future research and development.

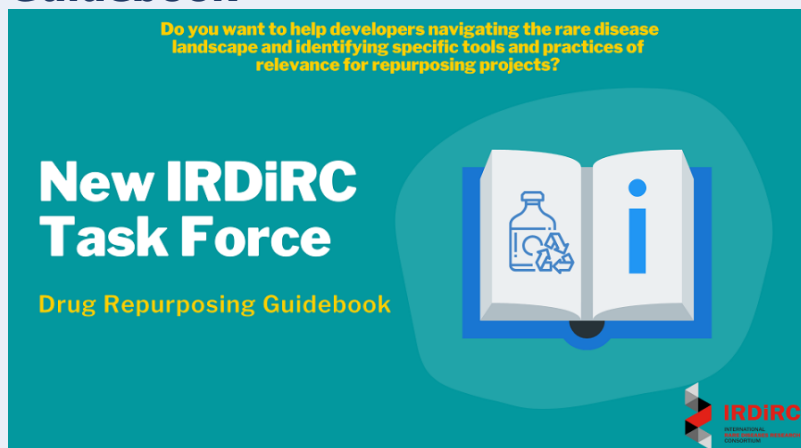
Interested candidates can **submit their application (CV, biosketch and letter of**

motivation, one paragraph each) to the Scientific Secretariat before [February 11th](#).

[More information](#)



Call for Members: Task Force on Drug Repurposing Guidebook



IRDiRC's [Therapies Scientific Committee](#)

(TSC) is establishing a **Task Force on Drug Repurposing Guidebook** aimed at helping developers **identify specific tools and practices of relevance for repurposing projects**, by focusing on repurposing approaches, following the same successful methodology used for the Orphan Drug Development Guidebook.

Interested candidates can **submit their application (CV, biosketch and letter of motivation, one paragraph each) to the Scientific Secretariat before [February 25th](#).**

[More information](#)

Commentary on 10 years of IRDiRC / Results of COVID-19 survey

- [A commentary on 10 years of progress and challenges of IRDiRC](#) has been published in the prestigious journal *Nature Reviews Drug Discovery*. The article **summarizes IRDiRC's vision and goals and**



highlights its achievements and prospects after its first decade (2011–2021).

- The [results of IRDiRC's COVID-19 survey](#) have been published in the *Rare Disease and Orphan Drugs Journal*. The article aims to **identify the needs and challenges of the RD community during the COVID-19 pandemic.**

[More information](#)

Leadership and Membership Changes

- **Dr David Pearce**, President of Innovation, Research & World Clinics, Sanford Health (USA) has been elected as the next **Chair of the IRDiRC Consortium Assembly (2022–2024)**. Dr Pearce is replacing Dr Lucia Monaco, ex Lead of the Research Impact and Strategic Analysis team at Fondazione Telethon (Italy). **Ms Samantha Parker**, Chief Patient Access Officer at InnoSkel (France), has been elected **Vice Chair of the IRDiRC Consortium Assembly (2022–2024)** and replaces Dr. Pearce in his former role.
- **Dr. Virginie Hivert**, Therapeutic Development Director, replaces Dr. Virginie Bros-Facer as **EURORDIS representative for PACC**.
- **Dr. Stefano Benvenuti**, Public Affairs Manager, replaces Dr. Lucia Monaco as **Fondazione Telethon representative for FCC**.



[More information](#)

OTHER NEWS

European Research and Innovation Days 2021 report released

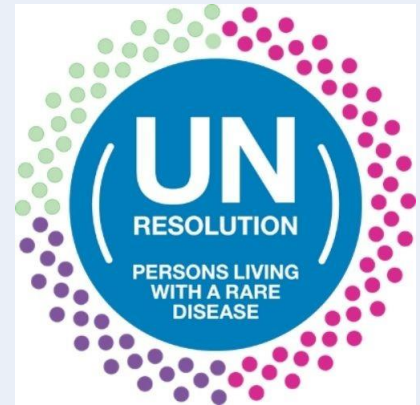


The **recently released report of the [2021 European Research and Innovation Days](#)** organised in June 2021 by the European Commission presents the main conclusion from the 21,000 participants and 200 high-level speakers. The report contains **summaries and quotes from the event's 70 online live sessions and workshops, as well as from over 200 consultations** between participants and Commission representatives.

[More information](#)

UN General Assembly formally adopts Resolution on Rare Diseases

Following a **sustained campaign by rare disease patient advocacy organisations such as Rare Diseases International** and with the support of several Member States, the United Nations (UN) General Assembly has formally adopted on **December 16th, 2021** with the consensus of all 193 UN Member States the **UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families**. This is the **first ever UN text to give full visibility to the over 300 million persons living with a rare disease worldwide** and calls for action to address the specific challenges faced by these individuals and their families.



[More information](#)

FDA announces 2022 grant funding opportunity for rare disease research



The Food and Drug Administration's (FDA) Office of Orphan Products Development (OOPD) has announced **availability of funds to support natural history studies for rare diseases and conditions**. These studies are intended to provide acceptable data to the FDA that will **substantially contribute to the approval of new products, or new indications for already marketed products**. The next receipt date is **February 15th, 2022**.

[More information](#)

Dravet Syndrome Foundation releases 2021 Therapeutic Pipeline report

The Dravet Syndrome Foundation Spain works to improve the quality of life of people with Dravet syndrome and their families. The foundation recently released its **end-of-year 2021 Dravet Syndrome Therapeutic Pipeline Report**. In this report, the Foundation undertook a **comprehensive review of the approved and investigational drugs and therapies for Dravet syndrome**. The report also summarises the activities and achievements in 2021 as a Foundation dedicated to research and the patient.



[More information](#)

EMA's Clinical Trials Information System (CTIS) goes live



The European Medicines Agency's (EMA) **Clinical Trials Information System (CTIS)** has gone live as of **January 31st**. CTIS is the backbone of the **Clinical Trials Regulation that will harmonise the assessment and supervision of clinical trials in the European Union**, ensuring better outcomes for patients and supporting the attractiveness of the European Union as a location for clinical research.

Sponsors, patients, healthcare professionals and the general public can visit the public Clinical Trials website at <https://euclinicaltrials.eu/home>.

[More information](#)

CAREERS

Job opportunities are available at EJP RD member institutions:

- The **EJP RD Coordination Team** (Paris, France) is looking for a **Project Assistant (Office Manager) (bilingual French and English)**
- UNIVERSITAETSKLINIKUM AACHEN (UKA) is looking for a **Research Associate (Biostatistics)**

Other job opportunities:

- The **Swiss Rare Disease Registry** (Berne, Switzerland) is offering a **3-year funded PhD position** as part of the [Screen4Care project](#).



EJP RD has received funding from the European Union's Horizon 2020 research and innovation programme under GA N°825575

