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What's new this period?

July–August 2022

EJP RD HIGHLIGHTS

OUR SERVICES

EJP RD launches Innovation Management Toolbox (IMT)

EJP RD is delighted to announce the launch on **June 30th, 2022**, of the **Innovation Management Toolbox (IMT)**, a **free-to-use and curated reference library of resources in rare disease translational medicine** that will provide investigators with self-help resources specific to their needs.

The database will be maintained actively!

- **Learn about the IMT:** <https://www.ejprarediseases.org/innovation-management-toolbox/>
- **Access the IMT:** <https://imt.ejprarediseases.org/>



[More information](#)

RARE DISEASES TRAINING AND EDUCATION

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

Registration deadline: August 15th, 2022

As part of the [training activities proposed by EJP RD](#), a **3-day training course** titled "Quality



assurance, variant interpretation and data management in the NGS diagnostics era” is being organised by [Universitätsklinikum Tübingen](#), in close collaboration with EJP RD task partners, targeted at the **international research community: clinicians, medical specialist, laboratory scientists (EBMG-registered), junior laboratory scientists, clinical geneticists, policy makers and assessors for**

laboratory accreditation, and patient representatives with a basic knowledge in biology or medicine.

The **fully online event** will take place from **October 19th – 21st, 2022**.

Registration is currently open until **August 15th, 2022**.

As the training course is limited to 30 attendees, respondents who are not selected will be kept on a waiting list until October 7th, 2022.

Registration & More information

Past EJP RD training videos now available on YouTube

We are glad to announce that **videos from past EJP RD training/webinars** are being made available on our [official YouTube channel](#).

[LATEST UPDATES]

(1) The webinar on **“Composite endpoints including patient relevant endpoints (Quality of Life)”** is now available [here](#), covering the following:

- The procedures to combine multiple endpoints and its limitations
- The properties and the flexibility of the class of generalized pairwise comparison tests
- The potential advantages and disadvantages of designing a clinical trial in rare disease with generalized pairwise comparisons primary analysis

(2) You can also watch the webinar on **Randomization procedures in Rare Disease Clinical Trials** [here](#).



Watch EJP RD training videos

UPCOMING ERN WORKSHOPS

Rhabdoid tumors in clinic and research: From basic biology to the patient bed

Registration deadline: July 31st, 2022

In the context of [EJP RD’s ERN Workshops](#), a **face-to-face**

WORKSHOP
RHABDOID TUMORS IN CLINIC AND RESEARCH: FROM BASIC BIOLOGY TO THE PATIENT BED

Organizers: Pascal Johann

5 NOVEMBER 2022
3-5 Place des Marseillais
84220 Charenton-le-Pont, France

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

EUROPEAN JOINT PROGRAMME ON RARE DISEASES
European Reference Network
The European Joint Programme on Rare Diseases is an initiative that has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement 101019729

workshop entitled "Rhabdoid tumors in clinic and research: From basic biology to the patient bed" aimed at **discussing the current clinical standard of treatment in rhabdoid tumors, as well as the controversies in the rhabdoid tumors regimen** is being organized by Dr. Pascal Johann of the [Universitätsklinikum Augsburg](#).

The **in-person event** will take place on **November 5th, 2022** in **Charenton-le-Pont, France**.

The workshop is open by prior registration and selection to **both PhD or Master students from the life sciences and MD students or residents, although advanced-stage PIs or physicians with more extensive clinical experience are invited to join**. All participants must be affiliated to an ERN Full Member or affiliated Partner institution.

The training workshop is **free of charge**.

Registration is open and closes on **July 31st, 2022**.

Those selected to participate from among the applicants will be informed by **August 15th, 2022**, of their selection.

[Registration & More information](#)

RDR CHALLENGES CALL

Rare Diseases Research (RDR) Challenge #2: Intranasal Device for Neonates (INDENEO)

EJP RD's innovative [Rare Diseases Research \(RDR\) Challenges](#) call in partnership with the [French Foundation for Rare Diseases](#) was aimed at **facilitating and funding collaboration between industry, academia, SMEs, and patient organisations to solve specific research challenges in rare diseases**.

The second challenge issued under the RDR Challenges call was for the **development of a delivery system for intranasal administration of biological drugs to neonates**. We are glad to

announce that the **project INDENEO (INtraNasal Device for NEOnates)** started at the end of **April 2022** in response to this challenge, bringing together an international consortium of four partners: EVEON, the Chiesi Group, Les Cliniques universitaires Saint Luc (Belgium) and the CEA (Fontenay aux Roses site – France). The project is co-funded by the Chiesi Group.

The **INDENEO project aims to develop a dropper system from nose to brain for biological drugs and advanced therapies in neonates**. The project will last **18 months** with **two main milestones**: the design and development of a functional prototype, then the pre-clinical validation.



[More information](#)

EJP RD FUNDING OPPORTUNITIES

NEXT COLLECTION DATE: SEPTEMBER 1ST

Networking Support Scheme (NSS) Funding Opportunity

OPEN CALL

Next collection date: **September 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.

Your event should take place between February 15th, 2023, and September 1st,

2023.

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 1st, 2022, at 14:00 (CEST)

Selected past networking events are available [here](#) providing insights on using this funding opportunity.

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 June and July 2022, EJP RD was presented at the following events:

- [Address to the Spanish Parliament \(June 21st, 2022\)](#)
- [ERICA General Assembly \(June 20th – 22nd, 2022\)](#)
- [11th European Conference on Rare Diseases and Orphan Products \(June 27th, 2022\)](#)
- [General Assembly Meeting of the TEDDY Network \(July 7th, 2022\)](#)



NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDiRC)

Redesigned IRDiRC website and State of Play 2019–2021 now available!



A [newly redesigned user-friendly IRDiRC website](#) was launched on **June 1st, 2022**, with an **easier navigation panel** to keep up with the latest rare diseases updates.

On **July 5th, 2022**, IRDiRC published the online version of the **2019–2021 edition of the State of Play**, a report that aims to inform stakeholders at large of **developments in the field of rare**

diseases research in order to support decisions of policy makers and research funders, based on a systematic survey of published articles in scientific journals and press releases.

[More information](#)

Diagnostics Scientific Committee (DSC) Nomination Call

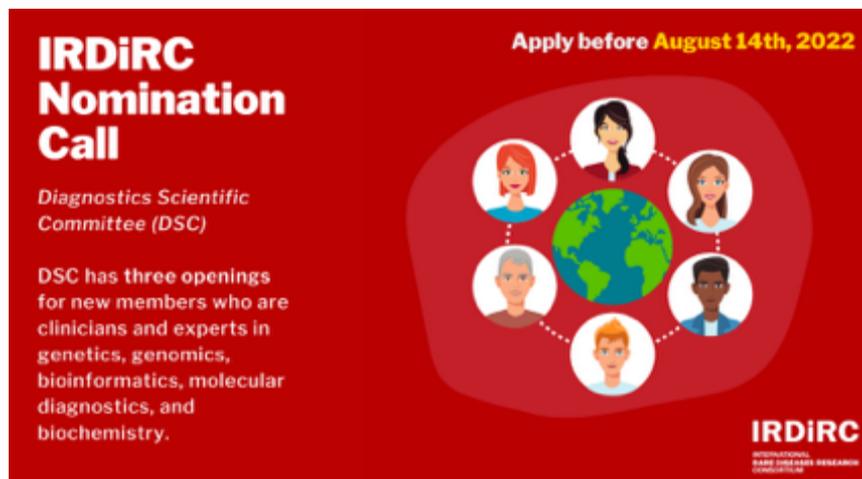
IRDiRC's [Diagnostic Scientific](#)

[Committee \(DSC\)](#) identifies current and future bottlenecks to rare disease gene

discovery, addresses challenges and roadblocks in rare disease diagnosis, and collaborates with international partners to develop tools and resources to facilitate genomic data discovery, analyses and sharing.

DSC has three openings for new members who are clinicians and experts in genetics, genomics, bioinformatics, molecular diagnostics, and biochemistry.

Apply before **August 14th, 2022.**



IRDiRC Nomination Call

Apply before **August 14th, 2022**

Diagnostics Scientific Committee (DSC)

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IRDiRC
INTERNATIONAL RARE DISEASE RESEARCH CONSORTIUM

[More information](#)

IRDiRC launches the Newborn Screening (NBS) initiative

IRDiRC is launching a discussion on the importance of **Newborn Screening (NBS)** with a focus on covering the topic by including examples of **implementation, usage of results, ethics, future state**. As different rules and regulations apply in different countries, IRDiRC can bring a major contribution in articulating the status of the science and its applications for all stakeholders, including patients, physicians, researchers, companies.

Information on this initiative will be updated on the IRDiRC website in due course.



Consortium Assembly Chair Dr. Pearce presents IRDiRC



IRDiRC member [Sanford Research](#), in conjunction with the [Professional Patient Advocates in Life Sciences \(PPALS\)](#), hosted the **2022 Patient Advocate Certificate Training (PACT)** on **May 16th – 19th, 2022**. Dr. David Pearce, Chair of the IRDiRC Consortium Assembly, presented both Sanford Research and also gave an overview of IRDiRC.

Dr. Pearce also presented at the **ERICA 2nd General Assembly** in **Bologna, Italy** on **June 20th – June 22nd, 2022**, on the topic of **"International Rare Disease Research Cooperation from the IRDiRC perspective"**. The presentation can be accessed at the following link: [ERICA Presentation - David Pearce](#).

Meeting Summary of the RDI Informal Side-Event to the 75th World Health Assembly

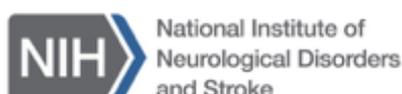
[Rare Diseases International \(RDI\)](#) would like to thank those who attended the **RDI Informal Side-Event to the 75th World Health Assembly**, which took place in **Geneva, Switzerland**, on **May 24th, 2022**.

The event was opened by the [World Health Organization](#) and discussed how **health systems around the world would be strengthened by the development of a Global Network for Rare Diseases**. The **WHO also recognised the rare disease community's proposed framework for the Network and affirmed its commitment to addressing rare diseases** to achieve Universal Health Coverage.



[More information](#)

NINDS creates the Ultra-Rare Gene-based Therapy (URGenT) Network



IRDiRC member [National Institute of Neurological Disorders and Stroke \(NINDS\)](#) recently created the [Ultra-Rare Gene-Based Therapy \(URGenT\) Network](#).

The URGenT Network supports the **development of state-of-the-art gene-based therapies for ultra-rare neurological diseases**, which affect as few or fewer than one in fifty thousand people. It supports **studies and clinical testing of gene-based or transcript-directed therapeutics** and **provides funding and resources to advance gene-based therapies for ultra-rare neurological diseases from late-stage pre-**

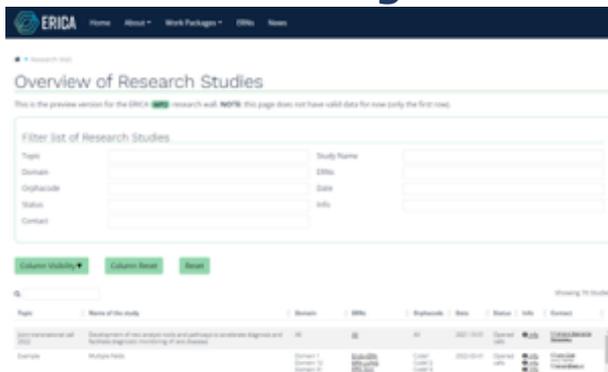
clinical development into first-in-human clinical testing, to accelerate the development of a promising clinical candidate with robust biological rationale.

[More information](#)



NEWS FROM THE EUROPEAN RARE DISEASE RESEARCH COORDINATION AND SUPPORT ACTION (ERICA)

New Matchmaking Tool: Collaborative Inter-ERN Research Wall



ERICA aims to **promote collaborative inter- ERNs research projects**. It is therefore crucial to have a **centralised location to announce any new project and search for collaborators**. A specific web page has been created for this purpose on the ERICA website [Research Wall](#), which provides **basic information about the open calls for collaboration as well as contact details of the project PI**. Please note that only inter-ERNs collaborative projects will be advertised via this research wall. New projects will be regularly announced and promoted. If you wish to

receive updates directly by email, we kindly ask you to [REGISTER HERE](#).

[More information](#)

Presentations from the ERICA 2nd General Assembly held in Bologna, Italy

The **ERICA 2nd General Assembly** was held on **June 20th - 22nd, 2022**, in **Bologna, Italy** as a hybrid meeting. **ERICA beneficiaries, ERN representatives, Expert Group members, Advisors and partners** had **three productive days full of interactive sessions**. All the presentations are now available at the ERICA website.



[More information](#)

OTHER NEWS

Science Webinar series – Combating the fragmentation of data and disciplines: Innovation hubs to address rare diseases

The recording of [Science's](#) webinar on **innovation hubs as a viable option to address rare diseases**, that took place on **May 26th, 2022**, is now available to watch on their website.

This webinar brings together key opinion leaders to **discuss the current and future needs of patients, and how innovators, doctors, scientists, drugmakers, and policymakers can work efficiently together to deliver solutions** to the millions of

diagnosed and undiagnosed patients in need.

[More information](#)

EMA grants marketing authorisation to rare disease therapies

In the months of **May and June 2022**, the [European Medicines Agency \(EMA\)](#) recommended granting various forms of marketing authorisation in the European Union (EU) for the following rare disease-targeted therapies:

1. First therapy to treat two types of Niemann-Pick disease, a rare genetic



metabolic disorder

2. First treatment for children with Progeria or progeroid like syndromes (rare premature aging syndromes)
3. First therapy to treat rare genetic nervous system disorder AADC deficiency
4. First gene therapy to treat severe haemophilia A

[More information](#)

World Duchenne Awareness Day 2022: Duchenne and Women



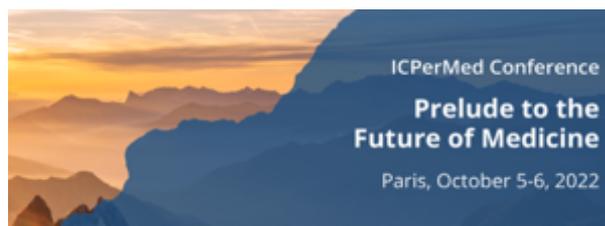
The **World Duchenne Awareness Day**, taking place each year on **September 7th**, is the **official campaign to raise awareness for people living with Duchenne & Becker muscular dystrophy**. Each year, an educational theme is chosen; this year's theme is **Women and Duchenne**. Through the creation of specific educational materials, the event will highlight all the **interconnected**

aspects of Duchenne and the female world.

[More information](#)

ICPerMed Conference 2022: Prelude to the Future of Medicine

The [International Consortium for Personalised Medicine](#) is pleased to invite you to the **ICPerMed Conference: Prelude to the Future of Medicine**, which will provide insight into what future of medicine will look like by **presenting concrete examples of personalised medicine approaches and aspects already in practice.**



The **hybrid event (in-person + virtual conference)** will take place over two days on **October 5th – 6th, 2022**, in **Paris, France.**

Registration is free but mandatory for virtual or in-person participation.

[More information](#)

Save the Date: Europe Biobank Week Roadshow 2022

The [European, Middle Eastern & African Society for Biopreservation and Biobanking](#) ([ESBB](#)) and [BBMRI-ERIC](#) are organising the **Europe Biobank Week Roadshow 2022 (EBW22).**

The **second stop of the EBW22 will be held as an in-person event in Rome, Italy** from **October 13th – 14th, 2022.**



Paediatric (research) biobanking is a European priority. Accordingly, the **critical starting point of the second stop will be an overall view of European commitment to paediatric research and healthcare and an in-depth exchange on good practices and priorities of various actors.**

[More information](#)

Questionnaire on health system resilience in the context of rare diseases

Gelareh Emami, a PhD student at the Department of Health Management at [Scuola Superiore Sant'Anna](#), under the supervision of Prof. Giuseppe Turchetti and Dr. Valentina Lorenzoni, is **investigating health system resilience in the context of rare diseases.**

She needs the help of **healthcare professionals (Medical doctors, Nurses, Physiotherapists, etc.) involved in the treatment of rare diseases to complete a questionnaire for this investigation.**

The results of this study will be used in future publications and in the student's PhD thesis.



[More information](#)

CAREERS

[Job opportunities](#) are available at EJP RD member institutions:

- ERKNet is looking for a **Project Manager** and a **Data Scientist**
- EURORDIS is looking for a **Patient Engagement Manager in HTA** & an **Events and Nominations Intern**
- BBMRI-ERIC is looking for a Head of **Biobanking Development**
- Izmir Biomedicine and Genome Center (IBG) is looking for an **ERA Chair and Head of the Unit for Rare Diseases at IBG**



**EJP RD has received funding from the
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