What's new
this month?

May–June 2022

EJP RD HIGHLIGHTS

SAVE THE DATE

RE(ACT) Congress and IRDiRC Conference 2023

The joint event **"RE(ACT) Congress and IRDiRC Conference 2023"** aims to bring together **scientific leaders and experts and young scientists from various breakthrough scientific fields to present cutting-edge research, exchange ideas, and discuss rare diseases research policies**. Patients and patient organizations committed to research will also be in attendance to share their experiences and perspectives. This event represents an exciting program with outstanding speakers and an **in-person event with multiple ways of networking!** We look forward to welcoming you to **Berlin, Germany** from **March 15th - 18th, 2023**.

Registration open at: https://www.react-congress.org/attendees/registration-2/

On behalf of the organizing committee:

Dr. Olivier Menzel, Chairman and Founder of BLACKSWAN Foundation

Dr. Daria Julkowska, EJP RD Coordinator

Dr. David Pearce, IRDiRC Consortium Assembly Chair

More information

UPCOMING EVENT

EJP RD and IRDiRC support 11th European Conference on Rare Diseases and Orphan Products (ECRD) 2022

Registration open

The **11th European Conference on Rare Diseases and Orphan Products (ECRD) 2022** is being organized by **EURORDIS** and co-organized by **Orphanet**, with **EJP RD serving as a full partner and IRDiRC as an associate partner**. The ECRD is a **patient-led rare disease policy event** in which collaborative dialogue, learning and conversation takes place, forming the **groundwork to shape goal-driven rare disease policies**.

The **fully online conference** will take place on **June 27th – July 1st, 2022** from **14.00 – 18.00 CEST**.

Registration is still open for this event.

More information

RARE DISEASES TRAINING AND EDUCATION

Joint EJP RD & ERICA Workshop: Ethics and regulatory considerations for ERN Data Access Committee members

We are happy to invite you to take part in a workshop addressing **"Ethics and regulatory considerations for ERN Data Access Committee members"**.

This workshop is jointly organised by EJP RD and ERICA and will take place online on **June 30th, 2022** from **10.00 – 12:30 CEST**.

It primarily aims to train the **members of the ERNs Data Access Committee members on the legal and ethical aspects to consider when examining an incoming data access request.** All other interested stakeholders are welcome to join.

Registration & More information

MOOC on Diagnosing Rare Diseases: From the Clinic to Research and Back

Now open continuously for enrollment

The **MOOC (Massive Open Online Course) "Diagnosing Rare Diseases: from the Clinic to Research and back"** co-developed by EJP RD, ERN Ithaca, ERN GENTURIS and the French Foundation for Rare Diseases is **open continuously for enrollment**.

This means that you can begin learning as soon as you enroll, without waiting for a new 'run' to begin. **Facilitation will be guaranteed till July 3rd, 2022 by the mentoring team**.

Registration is free and open at this **link**.

We specifically encourage **medical and biomedical science students** to register and follow the MOOC.

The **topics covered** include:

- The diagnostic process and the types of genetic tests available for rare diseases
- The differences in rare genetic diseases patient pathways
- Technological advances for diagnostic research
- The role of collaborative studies and data sharing in rare diseases diagnosis
- The impact of having a diagnosis or lacking a diagnosis on patients' lives
- The role and place of physiopathology approaches as well as social sciences research in the context of rare diseases diagnosis.

More information

UPCOMING ERN WORKSHOPS

Modelling & Simulation: Research methodologies for small populations in rare diseases

Registration deadline: June 17th, 2022

In the context of **EJP RD's ERN Workshops**, a face-to-face workshop entitled **"Modelling & Simulation: Research Methodologies for Small Populations in Rare Diseases"** aimed at **facilitating discussion and exchange of knowledge on the M&S methodologies and strategies** as innovative and promising enough for facing complex multifactorial or rare diseases and conditions that require highly specialised treatments and resources is being organised by Donato Bonifazi of the Consorzio per Valutazioni Biologiche e Farmacologiche (CVBF).

The **in-person event** will take place over **two days** on **July 4th – 5th, 2022** at the Hotel Excelsior in **Bari, Italy**.

The workshop is open by prior registration and selection to **PhD students, post-doc researchers, senior scientists, young clinicians, investigators and academics** and who are employees of or affiliated to an ERN Full Member or affiliated Partner institution.

The training workshop is free of charge and the training methodology will be based on lectures, seminars, and practical sessions, aimed at providing concrete research skills.

Registration has been extended to **June 17th, 2022**.

More information

RDR CHALLENGES CALL

Rare Diseases Research (RDR) Challenge #4: Development of a novel pre-clinical assay (TRXassay) to detect triplet repeat expansions

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 fourth challenge issued under the RDR call was for the **development of a pre-clinical assay to detect**

instability of microsatellite repeat expansions. We are glad to announce the launch of the **TRXassay**, a novel pre-clinical assay to detect triplet repeat expansions, as a response to this challenge. The project is co-funded by the EJP RD and by LoQus23 Therapeutics Ltd of Cambridge, UK.

In this project, **three teams from universities in Ireland, Scotland and Wales** will work together to **develop novel technologies** to monitor how the number of repeats changes in cells grown in the laboratory. This system will then be used to identify new drugs that slow the rate at which the repeat grows. The hope is that one such drug could then be used to treat multiple inherited disorders, including Huntington's disease and myotonic dystrophy.

More information

EJP RD FUNDING OPPORTUNITIES

Next collection date: September 1st

Networking Support Scheme (NSS) Funding Opportunity
Call opens: July 1st, 2022
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 submission system for the Networking Support Scheme will open on July 1st, 2022.

The next collection date is September 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

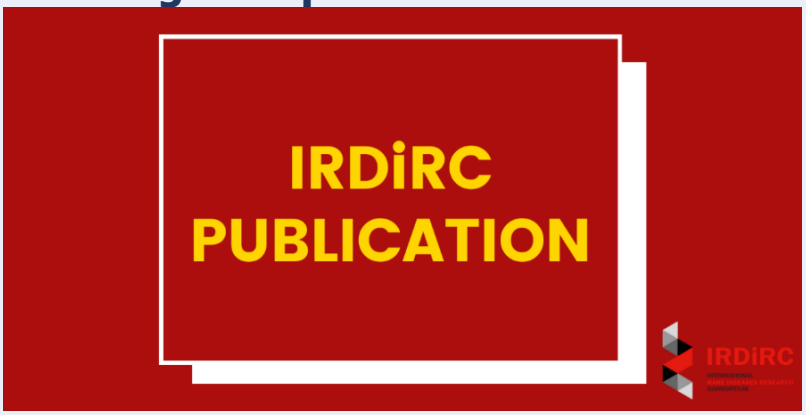
EJP RD IN EVENTS

During the months of April and May 2022, EJP RD was presented at the following events:

- **ERN-EYE presentation (April 26th, 2022)**
- **3rd ERA CoBioTech Hub Meeting (May 19th, 2022)**

NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

New Publication: Recommendations from the IRDiRC Working Group on Goal 3



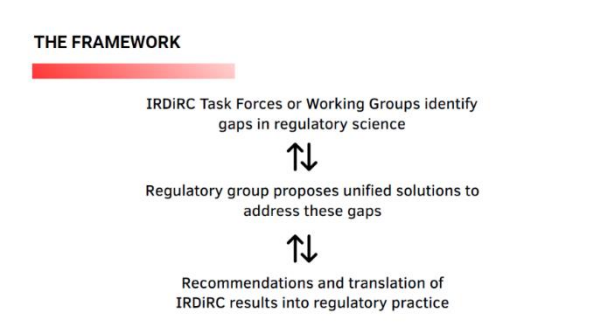
IRDiRC is proud to note that the **recommendations from the Working Group on Goal 3: Developing methodologies to assess the impact of diagnoses and therapies on rare disease patients** have been published in the *Orphanet Journal of Rare Diseases* in **May 2022**.

The Working Group characterized a set of metrics, tools and needs required for appropriate data collection and establishment of a framework of methodologies to analyze the socio-economic burden of rare diseases on patients, families and health care systems.

More information

IRDiRC Consortium Assembly Validates New Regulatory Scientific Committee

The IRDiRC Consortium Assembly has validated the **establishment of a new Regulatory Scientific Committee**. An open call for member nominations is currently ongoing. The Regulatory Scientific Committee is **part of the overarching IRDiRC goal to accelerate innovative medicines development for rare diseases**. The objectives of the Regulatory Scientific Committee will be to support the **development of IRDiRC activities to address specific regulatory challenges and identify additional topics for regulatory research**.



Health Europa Quarterly publishes interview with IRDiRC Chair

In its **April 2022** issue, **Health Europa Quarterly** **magazine published an interview with Dr. David Pearce** in an article titled **"Improving care for rare disease patients."** With a **focus on unmet needs in rare disease treatment** and how innovative therapies could offer patients new hope, Dr. Pearce addresses the following questions:

- *What is the role of the IRDiRC within the wider care landscape?*
- *What are the key challenges currently facing rare disease treatment and management?*
- *How has the EU Orphan Regulation introduced in 2000 helped to incentivise research and improve treatment options? What treatment gaps remain?*
- *How could digital or technological solutions help with the diagnosis and treatment of rare diseases?*
- *Are there any notable developments in the world of rare disease management that you think our readers should be more aware of?*

More information

RDI/EURORDIS Rare Barometer Global Survey on Journey to Diagnosis for Persons Living with a Rare Disease

Rare Diseases International (RDI) partnered with the **EURORDIS Rare Barometer Survey** project to launch the **Global Survey on the Journey to Diagnosis for Persons Living with a Rare Disease**. This survey aimed to identify factors that influence the process of obtaining a diagnosis and obstacles along the journey. This is a **global survey, available in 26 languages, and was open until June 15th, 2022.** The ambition is to collect relevant

responses at the national level and for specific disease areas, while developing an image of the diagnostic journey for our global community.

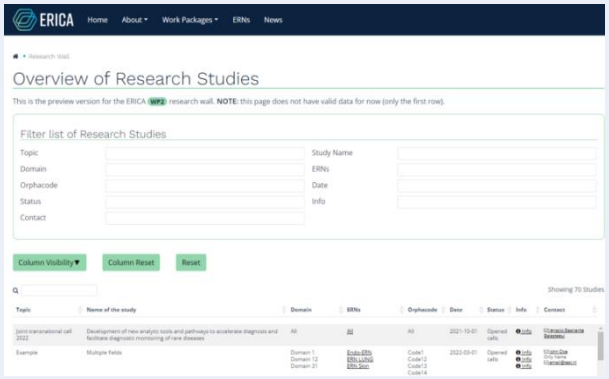
More information

Joint IRDiRC Consortium Assembly - Scientific Committees Meeting held in Paris

Joint IRDiRC Consortium Assembly and Scientific Committees Meeting: On **June 1st – 2nd, 2022**, an **in-person joint meeting** (with an online attendance option) of the **Consortium Assembly** and **Scientific Committees** was held in Paris, France, to discuss IRDiRC and its Committees' strategies and priorities, and provide updates on the activities of the Task Forces and Working Groups.

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

ERICA 2nd General Assembly to be held in Bologna, Italy

The **ERICA 2nd General Assembly** will be held on **June 20th - 22nd, 2022** in **Bologna, Italy**. All **ERICA beneficiaries, ERN representatives, Expert Group members, Advisors and partners** will gather for a **face-to-face symposium to discuss the progress and future of the ERN related Research activities and to participate in the WP-Specific Expert Working Group sessions**.

You will find the programme **here**.

Register **here**.

More information

OTHER NEWS

Innovative Health Initiative (IHI) hosted Kick-off and brokerage event

The **Innovative Health Initiative (IHI)**, a **European Union public-private partnership** funding health research and innovation, organised a **kick-off and brokerage event that provided pertinent information on upcoming IHI call topics and processes** and facilitated network building through face-to-face and virtual meetings as well as pitching sessions.

This event, which was targeted towards small and medium-sized enterprises, patient organisations, regulatory authorities, academic teams, industry, hospitals and other organisations with an interest in IHI projects, took place in **Brussels, Belgium** and **online** on **June 14th, 2022**.

More information

3rd International Symposium on Wiskott Aldrich Syndrome (WAS2022)

A funded networking event **"3rd International Symposium for Researchers and Clinicians on Wiskott Aldrich Syndrome (WAS2022)"** is being organised to offer access to the latest research and analysis related to this rare disease.

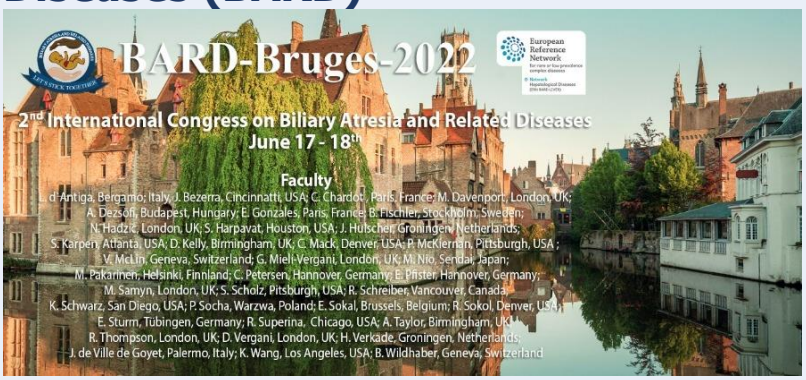
This event has received funding support from the EJP RD's **Networking Support Scheme (NSS) funding opportunity** as part of the first round of funding in March 2020.

The **in-person event** will take place on **June 17th, 2022** in **Munich, Germany** from **09.00 – 18.00 CEST**.

In this **networking conference**, participants will gain valuable insights into innovative perspectives in both basic and clinical research. The scientific programme draws together experts from around the world to discuss **breakthroughs in basic research, advances in clinical practice, novel therapeutic approaches and new insights into stem-cell and cellular therapies**.

More information

2nd International Congress on Biliary Atresia and Related Diseases (BARD)



A funded networking event **"2nd International Congress on Biliary Atresia and Related Diseases (BARD)"** is being organised with funding support from the EJP RD's **Networking Support Scheme (NSS) funding opportunity** as part of the second round of funding in September 2020. The **in-person event** will take place over two days on **June 17th – 18th, 2022** in **Bruges, Belgium**.

More information

European meeting on Phelan-McDermid syndrome

A funded networking event **"European meeting on Phelan-McDermid syndrome"** is being organised to d**iscuss the guideline recommendations and to strengthen the collaboration that was started in 2020 by the European consortium on Phelan-McDermid syndrome (PMS) and supported by ERN-ITHACA**, in order to tackle the knowledge gaps that they identified, as well as to discuss how best to proceed with the establishment of a European database enabling the collection of more data on the natural history of PMS, especially on the often observed mental health problems.

This event has received funding support from the EJP RD's **Networking Support Scheme (NSS) funding opportunity** as part of the seventh round of funding.

The **in-person event** will take place over three days on **June 20th – 22nd, 2022** in **Groningen, the Netherlands**. The meeting will be hosted by the centre of expertise for PMS of the UMC Groningen, the Netherlands, member of ERN-ITHACA.

More information

Recording of C-Path Webinar on Moving Global Regulatory Science Forward now available

In **April 2022**, the Critical Path Institute (C-Path) and the European regulatory, industry and research community were excited to introduce **"C-Path in Europe: Moving Global Regulatory Science Forward"** as a webinar.

The presentation highlighted how this nonprofit's new headquarters in Amsterdam will continue to improve public health, share expertise, data, risks and costs to move global regulatory science forward.

If you missed the webinar, the recorded presentation can be watched on demand at the link below.

More information

Article on Diamond-Blackfan anemia published in *Blood*

Diamond-Blackfan anemia is a rare disease characterized by a defect in the production of red blood cells. In a **plenary paper** published in the journal *Blood* on **May 26th, 2022** scientists highlighted a **new form of the disease linked to the loss of function of the *HEATR3* gene**, the product of which chaperones the synthesis and assembly of a ribosomal protein.

As a result of the **joint work of several genetic services within the RiboEurope consortium, funded by EJP RD through the Joint Transnational Call 2019**, variants of a new gene, HEATR3, were detected in six patients in the Netherlands, in Turkey and France.

More information

CAREERS

Job opportunities are available at EJP RD member institutions:

- EURORDIS is looking for a **Drug Repurposing Project Senior Manager**, **Patient Engagement & Training Manager**, **Open Academy Training Manager**, and a **Communications Manager**
- BBMRI-ERIC is looking for a **Project Management Officer** and a **(Scientific) Senior Project Manager**

EJP RD has received funding from the

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