What's new

this month?

February 2022

EJP RD HIGHLIGHTS

RARE DISEASE DAY 2022

EJP RD launches Instagram for Rare Disease Day 2022!

INSTAGRAM LIVE EVENT with Lara Bloom on February 28th at 2:30pm CET

To commemorate **Rare Disease Day (February 28th)**, EJP RD is proud to announce the launch of our **Instagram account** and an **exciting social media collaboration** with rare disease influencer **Prof. Lara Bloom, President and CEO of the Ehlers-Danlos Society.** On that day, **Prof. Bloom will take over our Instagram account** and post stories relating to EJP RD as well as the important advocacy work that she's engaged in.

At **2:30 pm CET**, **Prof. Bloom will come LIVE on EJP RD's Instagram all the way from Dubai!**

More information

OPEN FUNDING CALL

European Commission funding call for RNA-based therapies and diagnostics for rare genetic diseases

The European Commission (EC) has adopted the **2022 work programme of the European Innovation Council (EIC)**, opening **funding opportunities** worth over €1.7 billion in 2022 for breakthrough innovators to scale up and create new markets. This includes **€60.5 million to tackle three Transition Challenges**, notably one on the **development of RNA-based therapies and diagnostics for complex or rare genetic diseases**.

The **EIC Transition 2022 (HORIZON-EIC-2022-TRANSITION-01)** call, which has a **planned opening date of March 1st**, is intended to meet the following specific objectives:

- Advance, beyond the state-of-the-art, RNA delivery methods, including robust mRNA formulations, that would enable effective and safe delivery of mRNA into the cells;
- Design, develop and preclinical validate of novel miRNAs (miRNA lncRNA, tRNA or siRNA-based) therapies for complex or rare genetic diseases;
- Develop and validate novel RNA-based diagnostics and RNA-based predictive biomarkers that would allow for early and more accurate diagnosis and for favourable or non- post-treatment prognosis, respectively.

The funding call has the following deadlines: **May 4th** and **September 28th**.

More information

RARE DISEASE DAY 2022 EVENTS

PFUE 2022 Scientific Symposium on Rare Diseases

In the framework of the **French Presidency of the Council of the European Union (PFUE 2022)**, and under the high patronage of the French Ministry of Higher Education, Research and Innovation (MESRI), **INSERM (French National Institute of Health and Medical Research)** is organising a **scientific symposium on Rare Diseases**.

The **hybrid conference** (**in-person** for specially invited attendees and **online** for other attendees) will be held on **February 28th** from **09.00 – 12.30 CET** in **Paris, France**.

The objective of this conference will be to **review the current state of research and the means implemented for the diagnosis, care and treatment of rare diseases, and the expected prospects for increased collaboration** – between Member States of the European Union, public and private partners, research actors – on a European scale.

Please register to attend the event virtually. EJP RD Coordinator Dr. Daria Julkowska and other EJP RD members will be presenting the concept of the future Rare Diseases Partnership under Horizon Europe.

Register

RARE DISEASE DAY 2022 EVENTS

2022 Global Rare Disease Day Event at the World Expo Dubai

The **NGO Committee for Rare Diseases**, **Ågrenska Foundation**, **Rare Diseases International (RDI)**, and **EURORDIS-Rare Diseases Europe** are jointly organising their **2022 Global Rare Disease Day Event** (**February 28th**) at the **World Expo, Dubai**.

The event will take place from **07.30 – 13.30 CET (10.30 – 16.30 local time)** at the Sweden Pavilion (Sustainability District), World Expo, Dubai (United Arab Emirates). **You can also join the event virtually (registration required)!**

The aim of the event is to **support rare diseases as a global priority for equity**, discuss **impact and implementation of the UN Resolution on Rare Diseases at the global and local level**, and **build a roadmap for the future of rare diseases beyond health**, with a focus on Africa and low- and middle-income countries.

Register

UPCOMING EVENT

2nd International Conference on Rare Diseases: Greek Chapter

The **2nd International Conference on Rare Diseases** is being organized by **95, Rare Alliance Greece** with the theme of **"The Balancing Act between Equity and Sustainability"**.

The **fully online conference** will take place over two days on **March 1st – 2nd** from **09.00 – 17.30 CET**.

Targeted towards **healthcare stakeholders** such as government officials, pharmaceutical industry executives, public and private health services providers, health economists, health policy experts, healthcare professionals, nurses, academics, researchers, and journalists, the conference will address the following **themes**:

- Building a Sustainable Healthcare System based on equity, equality and patient rights
- European vs National RD Action Plans
- Advancing Policy Discussion on Prevention and Newborn Screening as Pillars of Public Health
- Data Wealth in Rare Diseases
- Thinking out of the box: Alternative Access and Funding Models for Rare Diseases
- Rare Disease as a National Priority in Greece

Registration is currently open!

Register

RDR CHALLENGE PROJECT

Rare Diseases Research (RDR) Challenge #1: Digital Tools for Rare Disease (DT4RD)

EJP RD's innovative Rare Diseases Research (RDR) Challenges call in partnership with Fondation Maladies Rare aimed at **facilitating and funding collaboration between industry, academia, SMEs, and patient organisations to solve specific research challenges in rare diseases**.

The **first challenge** issued under the RDR call was for the **development of a non-invasive tool for measuring rare disease patient mobility in daily living**. We are pleased to announce the launch of the **three-year Digital Tools for Rare Disease project (DT4RD)** as a response to this challenge.

The DT4RD project consortium will develop a **digital platform capable of assessing upper limb function remotely and continuously**, capturing the point between being ambulant and non-ambulant, a pivotal juncture in the progression of rare diseases.

More information

COLLABORATION

EJP RD and C-Path to Expand Global Impact and Partnership

In **February 2022**, EJP RD announced a **collaboration** with the Critical Path Institute (C-Path), an independent global nonprofit organization, to **advance technologies and methodologies that are fit for regulatory purposes to further global rare disease research and drug development**.

In this context, EJP RD and C-Path have successfully established a **collaboration using a shared data model to demonstrate that key datasets from both efforts can be queried simultaneously**. Building on the experience from the past year, both organizations will **support the creation of an interoperable global data ecosystem** for rare

disease to expedite effective development of new therapies. The partnership will benefit patients, regulators, advocacy stakeholders, researchers and industry.

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

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. Sklodowska-Curie National Research Institute.

The in-person event will take place on **May 4th** at the **M. Sklodowska-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

UPCOMING ERN WORKSHOP

ERN Workshop: Translational research on bone impairment in rare diseases

Registration deadline: March 27th

In the context of **EJP RD's ERN Workshops**, a face-to-face workshop on **"Translational research on bone impairment in rare diseases"** aimed at giving an update on translational research on bone impairment in rare diseases and **bringing together experts and trainees to facilitate collaborations** is being organised by Justine Bacchetta of the Faculty of Medicine of Lyon.

The **in-person event** will take place over **two days** on **June 9th – 10th** at the **Faculty of Medicine of Lyon** in **Lyon, France**.

The workshop is open by prior registration and selection to **senior scientists, senior physicians, postdocs, medical fellows, and PhD students** who are employees of or affiliated to an ERN Full Member or affiliated Partner institution.

The training workshop is free of charge and consists of interactive presentations and discussions on different areas of interest. On the second day, a "meet the experts" session will encourage small group talks, exchanges, and networking.

Registration closes on March 27th.

More information

ANNOUNCEMENT

Prof. Franz Schaefer conferred EURORDIS Scientific Award 2022

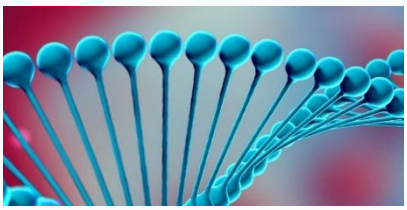
EJP RD is proud to announce that EJP RD member **Prof. Franz Schaefer was conferred the EURORDIS Scientific Award 2022** during the online award ceremony on **February 8th**.

The EURORDIS Scientific Award, which forms part of the annual **Black Pearl Awards**, recognises **Prof. Schaefer's scientific excellence and comprehensive reach**, the **outstanding dedication** he has demonstrated in the rare disease community and the positive impact he has made on rare disease research and patient community on an international level.

More information

ANNOUNCEMENT

Update: Online events announced in the run up to Third International Summit on Human Genome Editing



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 2023. The original dates of **March 7th – 9th, 2022** are being used to host a **three-part series of online events**. Registration for these events is not needed. **All three events will be live streamed** and the recorded presentations will be made available:

March 7th: Looking Ahead to the Science

March 8th: Looking Ahead to the Equity & Access

March 9th: Looking Ahead to the Governance

More information

EJP RD FUNDING OPPORTUNITIES

Next collection date: March 1st

Networking Support Scheme (NSS) Funding Opportunity
Next collection date: March 1st

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 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 January and February 2022, EJP RD was presented at the following events:

- **During the** Fourth Roundtable on Ethics of Genome Editing: Equal Access and Governance **(January 20th)**
- **During the ERA-LEARN** workshop **entitled** *"Horizon Europe Workshop European Partnerships – involvement of Associated Countries"* **(February 3rd)**
- **During the upcoming** PFUE 2022 Scientific Symposium on Rare Diseases **(see description above) (February 28th)**

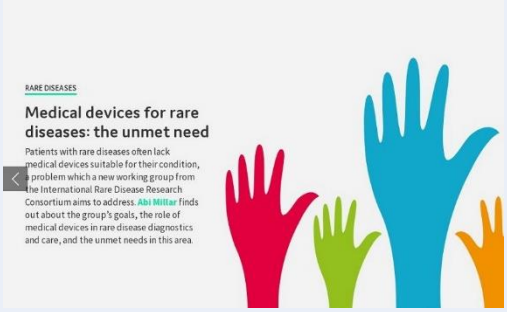
NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

Horizon Magazine publishes interview with IRDiRC leadership

On **February 7th**, Horizon Magazine, issued by the European Commission's Directorate-General for Research and Innovation, **published an interview with past and present IRDiRC leadership**. The article features wide-ranging comments from **Dr. Lucia Monaco**, outgoing IRDiRC Consortium Assembly Chair, and **Dr. David Pearce**, the newly elected IRDiRC Consortium Assembly Chair (2022–2024). Through her comments, Dr. Monaco provides a **succinct overview of IRDiRC's major accomplishments to date**, while Dr. Pearce concentrates on IRDiRC's **future potential**.

More information

TSC Vice Chair interviewed for article on medical devices for rare diseases



Dr. Anneliene Jonker, Vice Chair of the IRDiRC Therapies Scientific Committee (TSC), has been interviewed extensively for an article published in the **February 2022** issue of *Medical Technology* magazine.

The article, titled **"Medical devices for rare diseases: the unmet need"**, presents the goals of the **IRDiRC Working Group on MedTech for Rare Diseases**, the role of medical devices in rare disease diagnostics and care, and the unmet needs in this area.

More information

Chan Zuckerberg Initiative announces two patient-partnered Requests for Application (RFA) with rare disease focus

IRDiRC member **Chan Zuckerberg Initiative (CZI)** is **inviting applications from collaborative teams** bringing together patient-led rare disease organisations and research teams for **4-year research projects** aimed at advancing understanding of the fundamental science of rare diseases across two requests for applications (RFAs):

- The **Patient-Partnered Collaborations for Single-Cell Analysis of Rare Inflammatory Pediatric Disease RFA** aims to support the **application of single-cell biology methods to rare inflammatory pediatric diseases** in order to clarify cellular mechanisms of disease, improve understanding of disease heterogeneity, identify biomarkers, and improve diagnosis. **Patient organizations are expected to be active collaborators** on this research opportunity and full partners in the development of the grant application.
- The **Patient-Partnered Collaborations for Rare Neurodegenerative Disease RFA** aims to advance the understanding of the pathophysiology and mechanistic underpinnings of rare neurodegenerative and neurological disorders.

The **deadline** for submitting applications is **May 24th**.

More information

OTHER NEWS

EC Horizon Impact Award 2022 open for applications

The **European Commission (EC)** has launched the 2022 edition of its **Horizon Impact Award**, a prize dedicated to **EU-funded projects whose results have created societal impact across Europe and beyond**. **Six winners** will be chosen, and **each will receive €25,000**. The contest is open for applications until **March 8th**. The winners will be announced at a dedicated award ceremony in Brussels in 2022. The prize acknowledges and rewards the **most influential**

and impactful project results under Horizon 2020 (2014-2020) and its predecessor, the 7th Framework Programme (FP7, 2007-2013). The prize will highlight concrete achievements that have a demonstrable value for society, and will celebrate the people who made it happen.

IMPORTANT: *The contest is open only to FP7 and Horizon 2020 projects that have ended by the close of the contest and that can provide proof of their impact.*

More information

Dravet Syndrome Foundation organises Scientific and Family Days 2022

The **Dravet Syndrome Foundation Spain (FSD)**, a non-profit organisation that promotes, encourages, and connects the world's leading research centers for **Dravet syndrome and related diseases**, is announcing its upcoming conferences taking place in 2022. These **in-person events** will take place in **Madrid, Spain** over three days from **March 31st – April 2nd**.

1. **Dravet Syndrome Conference 2022** – *March 31st*
2. **European Dravet Syndrome Advanced Therapies (EDSAT) 2022** – *April 1st*
3. **FSD Family Meeting 2022** – *April 2nd*

More information

1st World Congress on Rare Skin Diseases to be organised in Paris

Organized by the **Rare Skin Diseases Network of the René Touraine Foundation**, in association with **ERN SKIN**, the **1st World Congress on Rare Skin Diseases** is an event that brings together the best experts, patient representatives and industry around the same cause: improving the care of patients with rare skin diseases.

This **in-person event** will take place in **Paris, France** over **three days** from **June 7th – 9th**.

The program includes the following:

- **4 plenary sessions** discussing on multidisciplinary management of rare disorders, clinical trials, orphan drugs, active research, and rising stars
- **28 parallel workshops** on the various diseases and topics
- **1 poster session** (there will be a call for posters)
- **2 training sessions**

More information

International Scientific Symposium on the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders

The **Ehlers-Danlos Society** is organising the **International Scientific Symposium on the Ehlers-Danlos Syndromes (EDS) and Hypermobility Spectrum Disorders (HSD)**, a state-of-the-art meeting in which **new research on clinical advances and the molecular and pathogenic mechanisms underlying EDS** and related syndromes will be discussed.

This **hybrid event** will take place in **Rome, Italy** and **online** over **five days** from **September 14th – 18th**. The event will bring together leading experts, clinicians, and scientists in the field of the EDS and HSD for a high quality, scientific program with a focus on **"Translational Medicine in EDS and HSD – from Basic Science to Community."**

More information

Dates announced for Innovation Bootcamp in Rare Diseases 2022 (IBRD2022)

The dates for the **2022 edition of the Innovation Bootcamp in Rare Diseases (IBRD2022)** have been announced. The event is targeted towards all professionals involved in the prevention, treatment and diagnosis of rare diseases and orphan drug research and development, including researchers, clinicians, pharma, policy makers and patient representatives.

This **in-person event** will take place in **Brussels, Belgium** on **October 11th**.

More information

EU-STANDS4PM publishes review article on computational modelling for personalised medicine

EU-STANDS4PM, a European standardization framework for data integration and data-driven in silico models for personalised medicine, has published a **review article on computational models for clinical applications** in the *Journal of Personalized Medicine*.

In the article, the authors discuss the **most relevant computational models for personalised medicine in detail that can be considered as best-practice guidelines for application in clinical care**. They define specific challenges and provide applicable guidelines and recommendations for study design, data acquisition, and operation as well as for model validation and clinical translation and other research areas.

More information

Scientify Research publishes curated list of rare disease grants and funding opportunities

Scientify Research, a Swedish company providing an **open, curated and structured research funding database** that focuses on aggregating information on funding opportunities across several research areas, has published a **curated list of grants and funding opportunities available in the field of rare diseases.** The website also collates information about research funders, and users are allowed to submit new grant and funding opportunity information for eventual inclusion in the list.

More information

CAREERS

Job opportunities are available at EJP RD member institutions:

- UNIVERSITAETSKLINIKUM AACHEN (UKA) is looking for a **Research Associate (Biostatistics)**

EJP RD has received funding from the

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