What's new
this month?

March 2022

EJP RD HIGHLIGHTS

OPEN FUNDING CALL

ERN Research Training Workshops Call
Submission Deadline: April 25th

The **ERN Research Training Workshops funding opportunity** is now open for applications until **April 25th**. The goal of the workshops is to **train researchers and clinicians affiliated with ERN Full Members or Affiliated Partners in relevant topics on research in rare diseases**.

The workshops will be delivered as two-day events. The **costs for the workshop organization will be covered up to a limit of €25,000.**

Organizer's profile: The applicant submitting workshop topics must fulfil one of the following conditions:

- Affiliated to any EJP RD beneficiary institution
- Affiliated to an ERN Full Member
- Affiliated to an ERN Affiliated Partner institution at the time when the application is submitted, as well as during the period of the execution of the workshop.

More information and application

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** from **14.00 – 18.00 CET**.

Poster abstract submissions for ECRD 2022 are open until March 31st.

The organizers encourage patient groups, academics, health care professionals and all other interested parties having conducted research or studies on rare diseases or public health projects to submit a poster abstract.

More information

EJP RD SERVICES

Explore the Rare Diseases Clinical Trials Toolbox

The **Rare Diseases Clinical Trials Toolbox** has been developed by EJP RD as a **practical aid for developers of clinical trials on medicinal products for human use regardless of therapeutic area**. The toolbox aims to collect the accumulated knowledge, experience, and resources (collectively termed as 'tools') generated by previous projects and/or research infrastructures and other organizations into a practical and guided toolbox to **help clinical trialists and R&D managers understand the regulations and requirements for conducting trials**, with special focus on investigator-initiated trials for rare diseases and applicable in Europe.

Check out the Rare Diseases Clinical Trial Toolbox today!

More information

EJP RD presents the FAIRopoly board game: FAIRification Guidance for ERN Patient Registries

The **board game FAIRopoly developed by EJP RD** illustrates the **FAIRification steps followed by the ERNs registries** to make their data more **F**indable, **A**ccessible, **I**nteroperable, and **R**eusable.

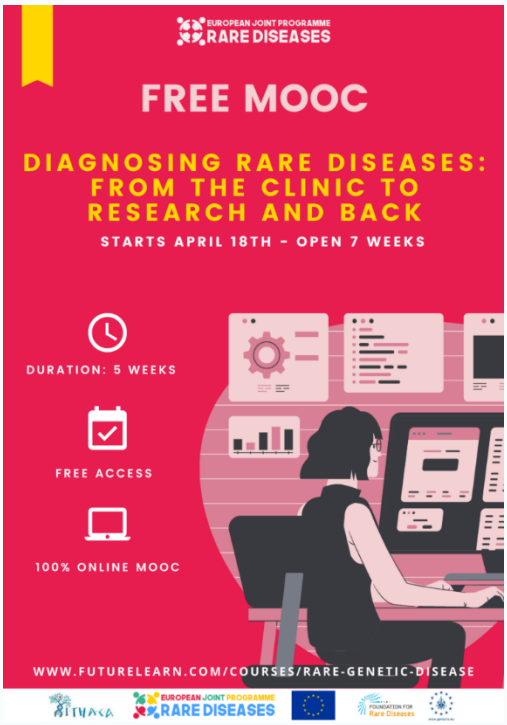
The player starts the game on the top left corner of the board and moves around clockwise one tile at a time. *Follow the link to fully explore the game!*

Explore FAIRopoly

RARE DISEASES TRAINING AND EDUCATION

MOOC on Diagnosing Rare Diseases: from the Clinic to Research and back

Third run from April 18th!



The **third run** of 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 will start on **April 18th**.

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

International Summer School 2022: Rare Disease Registries & Data FAIRification

Registration deadline: April 13th

As part of the **Training activities proposed by EJP RD**, the **International Summer School on Rare Disease Registries and FAIRification of Data** is a **5-day training programme** organised by Istituto Superiore di Sanità (ISS) in close collaboration with EJP RD task partners, aimed at the international research community, clinicians, medical specialists, registry curators, database managers, healthcare professionals and rare disease patients' representatives.

The **in-person training** will take place from **September 26th – 30th** at ISS in **Rome, Italy**.

Registration is currently open but will close on **April 13th**.

More information

Training for patient representatives and advocates on leadership and communication skills

Registration deadline: April 13th

As part of the **Training activities proposed by EJP RD**, an **international course** entitled **"Training for patient representatives and advocates on leadership and communication skills"** is a **2-day training programme** organised by Istituto Superiore di Sanità (ISS) in close collaboration with EJP RD task partners, open to **patient representatives** involved in the 24 European Reference Networks (ERNs), including members of the European Patients Advisory Groups (ePAGS), and other RD patient advocates.

The **in-person training** will take place from **November 10th – 11th** at ISS in **Rome, Italy**.

Registration is currently open but will close on **April 13th**.

More information

UPCOMING ERN WORKSHOPS

Translational research on bone impairment in rare diseases

Registration deadline extended to April 10th

In the context of **EJP RD's ERN Workshops**, a 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 has been extended to **April 10th**.

More information

From high throughput sequencing to diagnosis in immune mediated disorders

Registration deadline: April 14th

In the context of **EJP RD's ERN Workshops**, a face-to-face workshop entitled **"From high throughput sequencing to diagnosis in immune mediated disorders"** aimed at educating young researchers and clinician on which **techniques and functional tests are available and appropriate to solve challenging clinical cases in the field of immune mediated disorders and inborn errors of immunity (IEI)** is being organised by the Genetics Working Party (WP) of the European Society for Immunodeficiencies (ESID) together with the Molecular Testing Working Group (WG) of ERN RITA.

The **hybrid event (in-person with an option to participate online)** will take place over **two days** on **May 30th – 31st** at the **Imagine Institute, 24 Boulevard du Montparnasse, Paris, France**.

The workshop is open by prior registration and selection to **clinical and laboratory immunologists, internist-infectiologists, pediatricians, geneticists and pediatric and adult rheumatologists** 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.

Registration closes on **April 14th**.

More information

Ectodermal dysplasias: Training course and update

Registration deadline: April 14th

In the context of **EJP RD's ERN Workshops**, a face-to-face training course on **"Ectodermal dysplasias: Training course and update"** aimed at informing and training participants in **basic and practical aspects on Ectodermal Dysplasias (ED) and Incontinentia Pigmenti (IP)** with specific focus on recent research update research and innovative care is being organised by Smail Hadj-Rabia of ERN-Skin, Department of Dermatology, Necker-Enfants Malades Hospital.

The **in-person event** will take place on **June 10th** at the **Necker-Enfants Malades Hospital in Paris, France**.

The workshop is open by prior registration and selection to **residents in dermatology, ophthalmology, dentistry, surgery, neurology, general practice, pediatrics, researchers, and patient representatives** 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 participation in the conference with other specialists (with feedback on their observation and key learnings of the day) as well as classes on ED and IP. Real-life situations and ill-defined problems will be proposed.

Registration closes on **April 14th**.

More information

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

Registration deadline: May 10th

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** 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 closes on **May 10th**.

More information

Comprehensive gene profiling, molecular tumor board (MTB) and artificial intelligence in the diagnosis and treatment of patients with rare adult cancers

Registration deadline: June 2nd

In the context of **EJP RD's ERN Workshops**, a face-to-face workshop entitled **"Comprehensive gene profiling, molecular tumor board (MTB) and artificial intelligence in the diagnosis and treatment of patients with rare adult cancers"** aimed at increasing physicians' awareness on the **possibilities of comprehensive genomic profiling in gene-guided planning of modern cancer treatment** and addressing multidisciplinary aspects between specialists who are working in ERN-EURACAN and ERN GENTURIS is being organised by Pia Vihinen of the FICAN West Cancer Centre and Turku University Hospital.

The **in-person event** will take place over **two days** on **September 29th – 30th** at the **FICAN West Cancer Centre and Turku University Hospital, Turku, Finland**.

The workshop is open by prior registration and selection to **physicians and cancer researchers who want to deepen their knowledge in working within MTB and innovative genomic profiling** and 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 lectures and discussion.

Registration closes on **June 2nd**.

More information

EJP RD FUNDING OPPORTUNITIES

Next collection date: September 1st

Networking Support Scheme (NSS) Funding Opportunity
Call opens: July 1st
Next collection date: September 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.

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.

The next collection date is September 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 March 2022, EJP RD was presented at the following events:

- 2nd International Conference on Rare Diseases: Greek Chapter **(March 1st – 2nd)**
- ERN Workshop "Contemporary outcome measures in neuromuscular diseases" **(March 4th – 5th)**
- EURORDIS Winter School **(March 21st)**

NEWS FROM THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

IRDiRC released new video for Rare Disease Day 2022

On **Rare Disease Day 2022** (**February 28th**), IRDiRC was delighted to release an **introductory** video that **presents an outline of the Consortium and its goals**. This video also marks the occasion of **10 years of IRDiRC (2011 – 2021)**.

Don't forget to like, subscribe, and share to the **IRDiRC YouTube channel**!

Watch the video

European Commission produces IRDiRC interview series

On the occasion of **10 years of IRDiRC (2011 – 2021)**, the European Commission (EC) worked with IRDiRC to produce a **video series** featuring **six interviews with past and present IRDiRC leaders and members**. These videos have been released this month on the EC's DG Research & Innovation **Twitter account** and will also be made available on IRDiRC's **YouTube channel**.

- **February 28th**: Interview with Dr. David Pearce
- **March 4th**: Interview with Dr. Lucia Monaco
- **March 10th**: Interview with Irene Norstedt
- **March 17th**: Interview with Dr. Katherine Beaverson
- **March 22nd**: Interview with Dr. Gareth Baynam
- **March 28th**: Interview with Kevin Huang

Watch the videos

Last chance to respond to Genetic Alliance RFI for iHope Genetic Health

The **iHope™ Genetic Health (iGH) program** launched by IRDiRC member **Genetic Alliance** and supported by Illumina aims to **expand access to whole-genome sequencing to low- and middle-income communities around the world**, with more than one-third of funds being allocated to patients in Africa. iGH is requesting information from stakeholders to formulate a Request for Proposals (RFP) from laboratories and their associated clinical sites to serve the needs of undiagnosed patients. They **request information from all stakeholders**, especially patients and families, genomic laboratories, hospitals and/or clinics that care for patients with genetic disorders, but also from advocacy organizations, individual clinicians, healthcare administrators, genetic disease researchers, governmental agencies, and policy makers.

The deadline for responses is **April 1st**.

More information

Chan Zuckerberg Initiative hosts Networking Expo for Patient-Partnered Collaborations (PPC) for Rare Disease

Earlier this year, IRDiRC member **Chan Zuckerberg Initiative (CZI)** launched ***two Requests for Applications (RFA) for grant opportunities*** that will fund collaborative teams bringing together patient-led rare disease organizations and research teams for **4-year research projects aimed at advancing our understanding of the fundamental science of rare diseases**.

The **Networking Expo is a two-day ideation and matchmaking workshop** specifically designed as an opportunity for researchers and patient-advocates/organizations who are looking to develop a new collaboration to connect around potential projects for the Patient-Partnered Collaborations (PPC) for Rare Neurodegenerative Disease RFA.

The **virtual event** will take place over **two days** on **April 20th – 21st**.

Registration is open until **April 8th**.

More information

New Rare Care Centre will coordinate rare disease care at Perth Children's Hospital, Western Australia

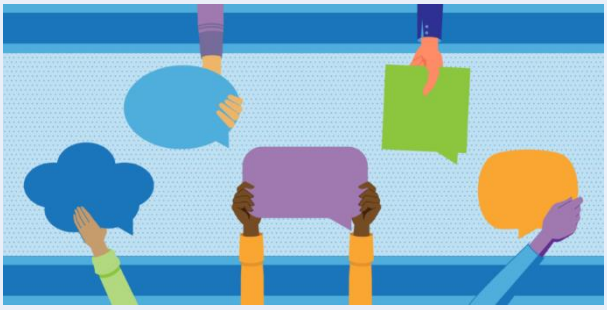
IRDiRC member **Western Australia Department of Health** announced the upcoming launch of a new **Rare Care Centre in Perth, Western Australia**, which will provide a **holistic model of care for children with rare and undiagnosed diseases** by delivering improved awareness and early identification of children with potential rare diseases and enhanced referrals to support earlier and more accurate diagnosis.

The Centre has secured funding from philanthropists and foundations of **10 million Australian dollars over 5 years**.

More information

OTHER NEWS

GA4GH seeks feedback through Community Survey and Town Hall Discussions



The **Global Alliance for Global Health (GA4GH)** is looking for feedback on its strategic approach via a **Community Survey** and **eight live Town Hall meetings** with members of the executive team.

The **Community Survey** is open until **May 13th**.

The **Town Hall meetings** are being proposed in **March** and **April**, and are open to everyone.

Every two years, GA4GH asks the international genomics community to advise on how well the organization meets its needs. This year, the **focus is on three "community imperatives"** that emerged in the most recent review: integration, implementation, and clinical engagement. The **plan is to refresh — rather than overhaul — GA4GH's strategic approach**.

More information

Rare Conversations 2022 webinar series: Game changing opportunities for the R&D community to address existing challenges in rare diseases

Rare Conversations invites you for the **inaugural episode of the new Rare Conversations event series in 2022**, aimed at powering the next decade of rare disease innovation in Europe. The first episode will be focused on **game-changing opportunities for the R&D community to address existing challenges in rare diseases**. The aim is to understand what is the basis needed to build the foundations for the future innovative rare disease ecosystem.

The **webinar** will take place on **April 6th** from **14.00 – 15.15 CET**.

The event, organized by **Alexion, AstraZeneca Rare Disease**, in cooperation with the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE), will revolve around discussions on how the revision of the OMP Regulation can create a space for opportunities for scientific progress.

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

Registration open for Innovation Bootcamp in Rare Diseases 2022 (IBRD2022)

Registration is now open for the **2022 edition of the Innovation Bootcamp in Rare Diseases (IBRD2022)**. 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

Policy paper available on unmet needs in Neuromyelitis Optica Spectrum Disorders (NMOSD) in Europe

The **Global Alliance for Patient Access** has made available a **policy paper on unmet needs in Neuromyelitis Optica Spectrum Disorders (NMOSD) in Europe,** published in multiple languages. A **rare autoimmune condition of the central nervous system**, NMOSD causes deterioration to a person's optic nerve and spinal cord. It **affects over 10,000 people in Europe** but is most common in women in their 30s and 40s.

More information

CAREERS

Job opportunities are available at EJP RD member institutions:

- BBMRI-ERIC is looking for a **Scientific Stakeholder Specialist** and a **Developer and Service Operator**
- Fondazione Telethon is looking for a **Head of Scientific Research**
- UNIVERSITAETSKLINIKUM AACHEN (UKA) is looking for a **Research Associate (Biostatistics)**

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