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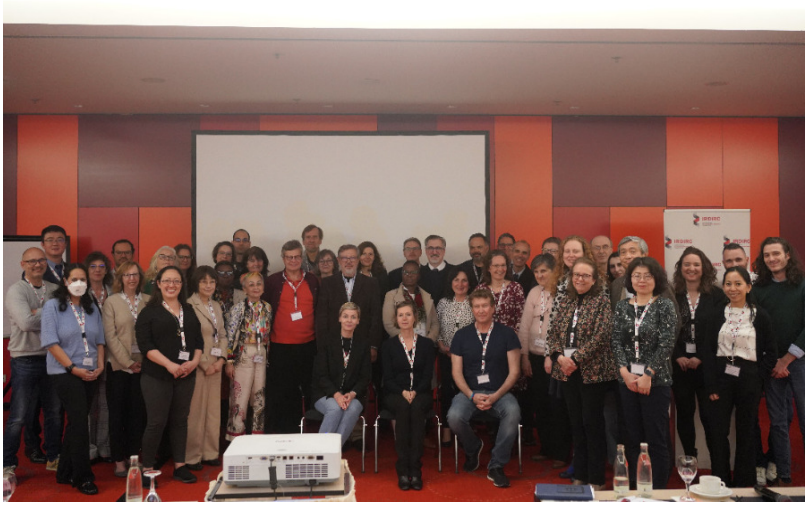
IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

IRDiRC Conference & RE(ACT) Congress held in Berlin, Germany

Berlin, Germany.The IRDiRC Conference & RE(ACT) Congress (March 14-18, 2023) gathered approximately 150 people in Berlin, and presented the latest advancements in knowledge, understanding, and innovation in **Rare Disease research.** The meeting offered an expanded presentation of health systems for rare diseases, and was a great opportunity to understand the challenges and opportunities for collaboration for promoting advancement in various areas of the Rare Disease ecosystem.



Read the full **Press Release**: **here**.

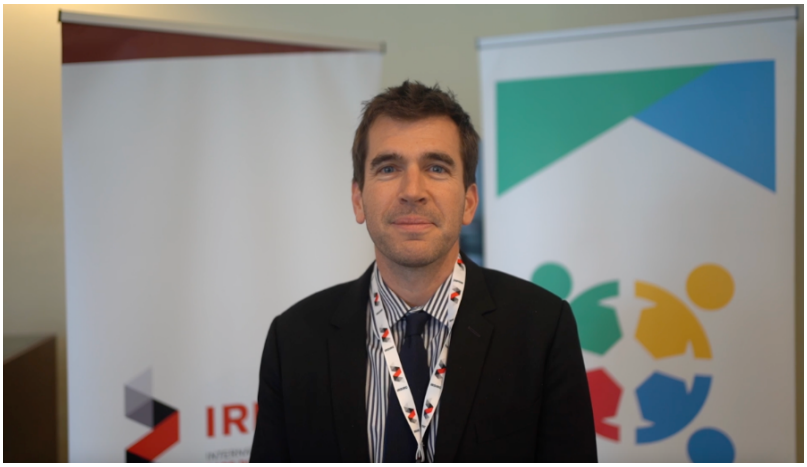
IRDiRC Members Describe IRDiRC in One Word

Rare Disease Day takes place worldwide, typically on or near the last day of February each year (28 February or 29 in leap years), to **raise awareness among policymakers and the public about rare diseases and their impact on patients' lives**.

This year we asked 9 of our members what **IRDiRC** means for them.

Watch the video

Meet Prof. Guillaume Canaud (MD at Necker Hospital for Children, France) & Speaker at RE(ACT) Congress



IRDiRC is delighted to share a short interview with **Prof. Guillaume Canaud (Necker Hospital for Children, AP-HP, France)** who offered an exceptional presentation on **"Targeted therapy for patients with PIK3CA-related overgrowth spectrum"** at the **RE(ACT) Congress** in **Berlin (Germany)**.

Check out his impressions of the **IRDiRC Conference x RE(ACT) Congress** below!

Watch the interview

New Members of the Consortium Assembly

IRDiRC is pleased to welcome the **European Federation of Pharmaceutical Industries and Associations (EFPIA),** represented by **Magda Chlebus** (Executive Director Science Policy and Regulatory Affairs, Belgium) as a new member of the **Companies Constituent Committee (CCC)**.

IRDiRC also wishes a warm welcome to the new member of the **Patient Advocates Constituent Committee (PACC)**, the **International Alliance of Dermatology Patient**

Organizations (also known as **GlobalSkin**), represented by **Tammi Shipowick** (Programs Director, Canada).

New Members of the Scientific Committees

Therapies Scientific Committee (TSC)

- **Emilio J.A. Roldan**: Scientific Director at Qualix DoT, Argentina;
- **Michelle Farrar**: Professor of Neurology at School of Clinical Medicine/Sydney Children's Hospital, Australia;
- **Trudy Nyakambangwe**: Founder at Child And Youth Care Zimbabwe, Zimbabwe.

Regulatory Scientific Committee (RSC)

- **Caroline Pothet**: Head of Advanced Therapies, Human Medicines Division, European Medicines Agency (EMA), The Netherlands.

IRDiRC wishes a warm welcome to its new members and it is looking forward to a fruitful collaboration on various rare diseases projects and initiatives.

IRDiRC EVENTS

Upcoming IRDiRC internal (closed) events:

- **Online Consortium Assembly Meeting**: on **June 7-8, 2023 (3-5 PM CET)**;
- **In-person Consortium Assembly Meeting:** Location - **Montreal (Canada)**, on **October 3-4, 2023**;
- **Online Consortium Assembly Meeting**: on **December 6-7, 2023 (3-5 PM CET)**;
- **In-person Consortium Assembly - Scientific Committees Meeting**: Location - **China** (Tbc), in **March 2024.**

IRDiRC representation at events

6th Annual Gene Therapy for Rare Disorders

IRDiRC Chair, **Dr. David Pearce** participated at the **6th Annual Gene Therapy for Rare Disease** (Boston, MA, USA) in the session: **"Understanding the Impacts to Various Stakeholders in the Rare Disease Community"**, and chaired several panel discussions. **Dr. David Pearce** also represented **IRDiRC** at the **Peds Symposium** (San Jose, Costa

Rica) in the session **"From the bench to treatment: The complex landscape of rare diseases"**.

NEWS FROM IRDiRC MEMBERS

FDA announces new Funding Opportunities for Rare Diseases

Two new funding opportunity announcements (FOA) for rare neurodegenerative diseases were recently posted by FDA/OOPD.

A. FOA Number: RFA-FD-23-028.

The purpose of this FOA is to **support efficient natural history studies and/or biomarker studies** that fill unmet needs for rare neurodegenerative diseases for children and adults. Through the support of prospective natural history and/or biomarker studies with high quality and interpretable data elements, FDA expects to address critical knowledge gaps, remove major barriers to progress in the field, exert a significant and broad impact on a specific rare neurodegenerative disease or multiple rare neurodegenerative diseases with similar pathophysiology, and facilitate rare disease product development. The receipt date for applications is **May 4, 2023** and top scoring grant applications will be funded in FY 2023.

B. FOA Number: RFA-FD-030.

The purpose of this funding opportunity is to solicit applications for **UH2/UH3 cooperative agreements to support and inform the future development and use of COAs for cBCIs in patients with amyotrophic lateral sclerosis (ALS)**.

The UH2/UH3 cooperative agreement involves **two milestone driven phases**:

1) the **UH2 Phase** will include a **systematic landscape analysis of the available literature**, **relevant data sources**, and i**nterviews with key opinion leaders (KOLs)** to document COAs for cBCIs used clinically and identify gaps between current COAs used in cBCI studies and other outcome measures that could demonstrate functional benefits for ALS patients with severe communication limitations;

2) the **UH3 phase** will consist of **patient and caregiver focus groups to collect information about symptoms**, **functional status**, and **perceived benefits/risks of cBCIs**. The receipt date for applications is **May 24, 2023** and the top scoring grant application will be funded in FY 2023.

Fondation Maladies Rares launched several Calls for Proposals

Fondation Maladies Rares (France) joined forces with different partners and launched several calls for proposals.

- Launched the call for proposals **"Innovative therapeutic proofs of concept in rare diseases"** funded by the **AFM-Téléthon** (France). The deadline for submission of the letter of interest is **April 13, 2023, 5pm CEST**;

- Launched the call for proposals **"Human and Social Sciences in Overgrowth syndroms"** funded by the **RHU COSY**. The deadline for submission is **May 11, 2023, 5pm (Paris time);**
- Launched the research price **"Pain in rare diseases"** funded by the **APICIL Foundation**. Call for projects open until **April 20, 2023, 5pm (Paris time)**.

Furthermore, **Fondation Maladies Rares** organised a **scientific meeting on regional research on rare diseases** that took place in **Strasbourg (France)** on **February 28th, 2023**.

More Information

AFM-Téléthon Calls for Proposals 2023

Through its annual calls for proposals, **AFM-Téléthon (France)** supports new research projects each year in France and abroad, in particular for young researchers. After the evaluation by its Scientific Board, the association finances the most relevant or innovative initiatives in the development of therapeutic concepts and understanding of the rare and neuromuscular diseases. Discover the current funding opportunities: **here**.

Dr. David Pearce (Sanford Research, USA) co-authored an article in Nature Medicine

Dr. David Pearce co-authored a correspondence in **Nature Medicine** discussing a new Task Force on Rare Diseases launched by the **European Burden of Disease Network**. The goal of this task force is to identify existing unmet needs related to the burden of rare diseases and perform research on the population health impact of rare diseases. ***"An EU task force to assess the burden of rare diseases."***

Link: https://pubmed.ncbi.nlm.nih.gov/36759674/

Rare disease education in Europe and beyond: time to act

IRDiRC members Biruté Tumiene (Lithuania), **Maurizio Scarpa** (Italy) and **Gareth Baynam** (Australia), shared their contribution in the development of a position statement that was recently published in the **Orphanet Journal of Rare Diseases**: ***"Rare disease education in Europe and beyond: time to act"***. This position statement was developed through common discussions of many stakeholders (from **ERNs**, **EURORDIS**, **UEMS**, **ESHG**, **Association of Medical Schools in Europe**) and aims to set the stage for further discussions on the common

strategy for rare disease education and training that encompasses the whole educational pyramid from students and general practitioners to highly-specialized experts (learners) and the whole ecosystem of education and training providers (as universities, hospitals, research institutions and infrastructures).

Link to publication

"Challenges in Pediatric Neuroscience Research Webinar Series" organised by NINDS

The next installment of the **"Challenges in Pediatric Neuroscience Research Webinar Series"**, will be on **May 12th, 2023 from 2-3 PM EST**. In this episode of the ongoing series, the discussion will be with **Dr. Heather Hazlett**, a pediatric neuropsychologist at the University of North Carolina at Chapel Hill, about **remote neuropsychological assessments**. This type of assessment (used increasingly by rare diseases researchers) became more frequent in research studies during the pandemic. However, there are some significant challenges associated with their use. They also offer an opportunity for research in the future.

This event is the sixth in a series and provides an overview of issues that clinical researchers face today. Future webinars will feature guest speakers who will cover specific challenges and how **NINDS** programs strategically address them.

Registration open: here.

More information

The Chan Zuckerberg Initiative grants award

The Chan Zuckerberg Initiative (CZI) is proud to work alongside several prominent organizations that share the goal of finding treatments and cures for rare diseases. At the end of last year (2023), the organization awarded **general operating support grants** to the following international partners:

- **Asia Pacific Alliance of Rare Disease Organisations;**
- **Enfermedades Raras en Caribe y America Latina;**
- **EURORDIS;**
- **Rare Diseases International;**
- **Wilhelm Foundation;**

Further, applications from teams bringing together patient-led rare disease organizations and research groups for research projects were also invited, that aimed at:

- advancing our understanding of the biological mechanisms of rare neurodegenerative and neurological disorders;
- supporting the application of single-cell biology methods to rare inflammatory pediatric diseases.

Each of the **ten winning teams** was awarded **USD 2,000,000 for 4 years** (grants started in December 2022):

- **PCH2cure**
- **Closing the Knowledge Gaps in Lafora, a Fatal Neurodegenerative Disease**
- **CADASIL-centered Modeling of Immunovascular Neurodegenerative Disease**
- **A Cell Atlas of Batten Pathobiology & Therapeutic Response**
- **Investigating ATP1A3 Diseases in Cell and Animal Models**
- **Integrating Patients to Accelerate the Science Towards a World without NEC**
- **Understanding the Cellular Ecosystem of Primary Sclerosing Cholangitis**
- **Creating an Inflammatory Childhood Interstitial Lung Disease Cell Atlas**
- **Deciphering RUNX1-Familial Platelet Disorder at Single-Cell Resolution**
- **Developing a Single-Cell Atlas to Accelerate Precision Medicine in Juvenile Dermatomyositis**

More Information

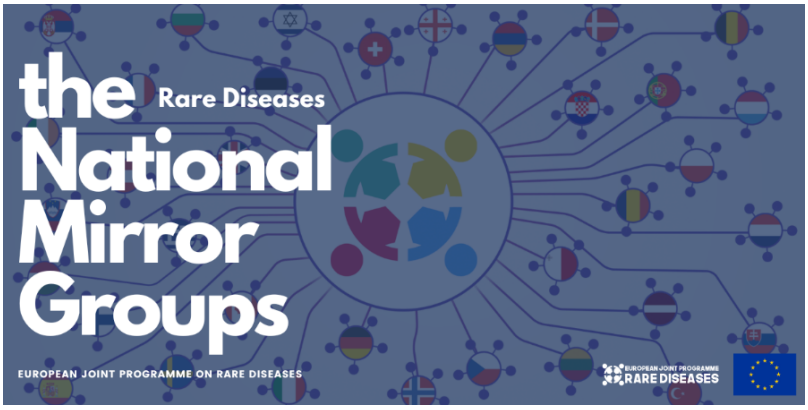
OTHER NEWS

Announcement of the Joint Transnational Call for proposals "Healthcare of the Future"

The **European Partnership on transforming health and care systems (THCS)**, a Cofund action under the **Horizon Europe Programme** designed to support coordinated national and regional research and innovation programmes along with capacity building, networking, dissemination and other key activities to support health and care systems transformation, will launch its **first Joint Transnational Call** (entitled **"Healthcare of the Future"**) for proposals aiming to encourage the **optimization of patient care pathways** and contribute to the transition towards more sustainable, efficient, resilient, ethical, high-quality, and accessible person-centered healthcare systems.

More information

New EJP RD video on the Rare Diseases National Mirror Groups (NMGs)



For the **Rare Disease Day**, **the European Joint Programme on Rare Diseases (EJP RD)** launched a video focusing on the connection between the expertise and knowledge from the Rare Diseases Community of a specific country with the EJP RD countries.

The less-than-3min video explains the **Rare Diseases National Mirror Groups (NMGs)** and their objectives, as well as a few testimonies from the **NMG Netherlands**, **NMG Portugal**, and **NMG UK**.

Watch the video

Solve-RD Final Meeting to take place on April 24-26 in Prague (Czech Republic)

Solve-RD, a **research project** funded by the **European Commission** for five years **(2018-2022)**, having the ambitious goals set out by the **International Rare Diseases Research Consortium (IRDiRC)** to deliver diagnostic tests for most rare diseases by 2020, invites its partners to attend the **Solve-RD Final Meeting 2023 in Prague (Czech Republic)**. The meeting will take place on **April 24-26, 2023** and the **registration for online participation is still open** until **April 23rd, 2023**.

Register **here** to attend the meeting online.

More information

The National Center for Genomic Analysis releases a new version of GPAP

The **CNAG-CRG** (Spain) released a new version of the **RD-Connect Genome-Phenome Analysis Platform (GPAP)** on **April 3rd**. The new interface is more intuitive and user friendly. Among other things, it provides more information and facilitates analysis reproducibility. The release also includes additional features for both family and cohort analysis. The **RD-Connect GPAP** is accessible online at **https://platform.rd-connect.eu**.

The Scientific Secretariat of IRDiRC is supported by the European Union through the European Joint Programme on Rare Disease under the European Union's Horizon 2020 research and innovation programme Grant Agreement N°825575.