



IRDiRC HIGHLIGHTS

IRDiRC releases 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 our **YouTube channel!**

[Watch the video](#)

European Commission produces IRDiRC interview series

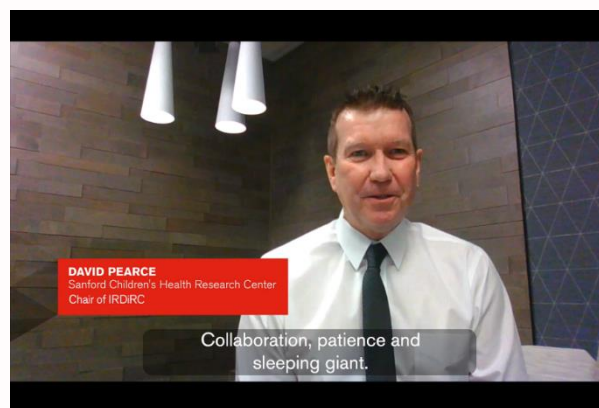
On the occasion of **10 years of IRDiRC (2011 – 2021)**, the European Commission (EC) worked with IRDiRC to produce a **video series** featuring **six interviews with past and present IRDiRC leaders and members**. These videos are being released this month on the EC's DG Research & Innovation **Twitter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

Videos already released:

February 28th: [Interview with Dr. David Pearce](#)

March 4th: [Interview with Dr. Lucia Monaco](#)

March 10th: [Interview with Irene Norstedt](#)



[Watch the videos](#)

IRDIRC supports 11th European Conference on Rare Diseases & Orphan Products



The **11th European Conference on Rare Diseases and Orphan Products (ECRD) 2022** is being organized by [EURORDIS](#) and co-organized by [Orphanet \(INSERM\)](#), with IRDiRC serving as an associate partner. 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**. **Registration is currently open!** **Poster abstract submissions for ECRD 2022 are open until [March 31st](#).**

[More Information](#)

IRDIRC at PFUE 2022 Scientific Symposium on Rare Diseases

In the framework of the **[French Presidency of the Council of the European Union \(PFUE 2022\)](#)**, IRDiRC member [INSERM \(French National Institute of Health and Medical Research\)](#) organised a **Scientific Symposium on Rare Diseases** on **February 28th** in **Paris, France**. IRDiRC Consortium Assembly Chair **Dr. David Pearce** participated in a **round table on challenges of public-private collaborations for therapeutic innovation in rare diseases** along with representatives from Sanofi France and Pfizer USA.



At this event, IRDiRC Scientific Coordinator **Dr. Daria Julkowska** also presented the the **concept of the future Rare Diseases Partnership under Horizon Europe**.

[More Information](#)

Horizon Magazine publishes interview with IRDiRC leaders



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 leaders**. 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 for an article published in the **February 2022** issue of *Medical Technology* magazine. The article, "**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](#)

Leadership and Membership Changes

- Agence National de la Recherche (ANR) has appointed **Prof. Philippe Bouvet** (Head of the Biology-Health Department) to replace Dr. Dominique Dunon-Bluteau on the FCC
- National Center for Advancing Translational Sciences (NCATS) – NIH has appointed **Dr. PJ Brooks** (Deputy Director, Office of Rare Diseases Research) to replace Dr. Anne Pariser on the FCC
- Lysogene has appointed **Dr. Marie Trad** (Chief Medical Officer) to replace Dr. Ralph Laufer (Chief Scientific Officer) on the CCC

We warmly thank **Prof. Anthony Brookes** (Leicester University, UK), who has ended his second mandate on the Diagnostics Scientific Committee (DSC), for his invaluable contribution to IRDiRC during his tenure.

IRDIRC IN EVENTS

IRDIRC held the following meetings:

- 9-10 March 2022: An online meeting of the **Consortium Assembly** was held to provide updates on the activities of the Committees, Task Forces, and Working Groups, and start planning for upcoming in-person events in 2022 and 2023.
- 9 March 2022: An in-person **strategic meeting** of the **Scientific Secretariat** was held in Paris in the morning before the Consortium Assembly.



IRDIRC was presented at the following events:

- 28 February 2022: During the **PFUE 2022 Scientific Symposium on Rare Diseases** in a session entitled "**Challenges of public-private collaborations for therapeutic innovation in rare diseases**" (see above)

NEWS FROM IRDIRC MEMBERS



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

On **February 25th**, Western Australia Department of Health announced the 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.

[**More Information**](#)



Genetic Alliance launches RFI (Request for Information) for iHope Genetic Health

The **iHope™ Genetic Health (iGH)** program launched by **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 announces two patient-partnered Requests for Application (RFA) with rare disease focus

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](#)

European Commission (EC) 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**,



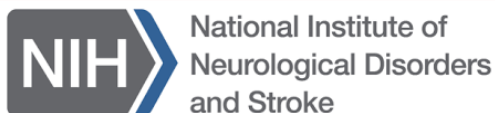
notably one on the development of RNA-based therapies and diagnostics for complex or rare genetic diseases.

The currently open ***EIC Transition 2022 (HORIZON-EIC-2022-TRANSITION-01)*** call 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](#)



Funding opportunities: Ultra-rare Gene-based Therapy (URGenT) Network

The **Ultra-rare Gene-based Therapy (URGenT)** program at **NINDS, NIH (National Institutes of Health)** will support the development of **state-of-the-art gene-based therapies for ultra-rare neurological diseases** through funding and resources for **late-stage pre-clinical development into first-in-human clinical testing**. Two funding calls are currently open.

[More Information](#)

Coordination of Rare Diseases at Sanford (CoRDS) program at Sanford

Based at **Sanford Research, Coordination of Rare Diseases at Sanford (CoRDS)** is a **centralized international patient registry** for all rare diseases that is free for both participants and researchers. The registry works with patient advocacy groups, individuals and researchers to capture health information from individuals with a rare diagnosis, undiagnosed patients, unaffected carriers or at-risk patients. The registry **connects researchers and patients and notifies participants of emerging clinical trials**.



[More Information](#)



Ultragenyx matches donations to Rare Disease Innovations Institute and Genetic Alliance's Baby First Test program

Genetic Alliance's **Baby's First Test Program** and the **Rare Disease Innovations Institute (RDII)** are organizations focused on **expanding access to newborn screening** so that individuals can be diagnosed and treated much earlier for rare diseases. **Ultragenyx will match donations of up to a total of \$25,000** for each of these organizations.

[More Information](#)

OTHER NEWS



EJP RD is officially on Instagram for

Rare Disease Day!

A new social media profile has been added to the European Joint Programme on Rare Diseases (EJP RD) online presence: Instagram!

To commemorate **Rare Disease Day (February 28th)**, EJP RD undertook **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 took over the EJP RD Instagram** account and posted stories throughout the day, which you can find in the "Highlights" on EJP RD's Instagram account. Prof. Bloom also hosted a live with EJP RD member Sonja van Weely, which you can watch or re-watch [here](#).

[Follow EJP RD on Instagram](#)

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.

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