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IRDiRC HIGHLIGHTS

IRDiRC Announces Creation of Regulatory Scientific Committee to Tackle Regulatory Challenges in Rare Disease Research

22 September 2022 – Paris, France –

IRDiRC announced the formation of the Regulatory Scientific Committee (RSC).

"IRDiRC recognizes that the fast-moving pace of rare disease innovation and therapeutics creates many considerations in the regulatory space.

This committee will be pivotal to

translate progress into effective, high quality and accessible therapies," said **Samantha Parker, Vice-Chair of IRDiRC and Chief Patient Access Officer at Innoskel (France).**

"The new RSC brings broad expertise across different stakeholders and geographies allowing for a balanced synergy that will provide insights and guidance into the advancement of IRDiRC's mission," said **David Pearce, Chair of IRDiRC, President of Innovation, Research and World Clinics at Sanford Health (USA).**

The RSC includes **11 experts** from around the world, representing **multiple geographies** and **stakeholders** from **regulatory bodies, patient groups, biotech** and **pharmaceutical industry, public** and **non-for-profit organizations, clinicians** and **scientists**.

Read the full Press Release [here](#).



[More information](#)

IRDiRC appoints new ISC members



IRDiRC is proud to announce the appointment of **eight new members** for the **Interdisciplinary Scientific Committee (ISC)**, bringing together experts from multiple fields to work on **cross-cutting emerging aspects of rare disease research, data sharing and ontologies, industry, social health impact and policies.**

We wish a warm welcome to our new members and we look forward to working with them:

Eva Bermejo-Sanchez - Director and Scientist at Instituto de Salud Carlos III, Spain

- **Claudio Carta** - Researcher at Instituto Superiore di Sanita, Italy
- **Helen Malherbe** - Director of Research and Epidemiology at Rare Diseases South Africa NPC, South Africa
- **Maria del Mar Mañú-Pereira** - Head of Rare Anemia Disorders Research Laboratory at Vall d'Hebrón Research Institute, Spain
- **Vassili Soumelis** - Chief Medical Officer at Owkin, France

- **Lindsey Murray** - Executive Director at Critical Path Institute (C-Path), USA
- **Gareth Baynam** - Medical Director at Rare Care Centre, Western Australian Department of Health, Australia
- **Kazuko Wada** - M.D. and Director at Osaka Women's and Children's Hospital, Japan

[More information](#)

Four new members of the IRDiRC DSC

The **IRDiRC Diagnostics Scientific Committee (DSC)** welcomes **four new members** to address the challenges of **rare disease diagnosis** and to **collaborate with international partners in developing tools and finding resources to foster data discovery, analysis, and sharing.**



We look forward to working with our new **DSC team members**:

- **Virginie Bros-Facer** - Associate Director, Medical Affairs at Illumina, EMEA
- **Victor Faundes** - Assistant Professor, Faculty of Medicine at University of Chile, Chile
- **Saumya Shekhar Jamuar** - Medical Director at KK Women's and Children's Hospital, Singapore
- **Sally Ann Lynch** - Consultant Clinical Geneticist at Children's Health Ireland, Ireland

[More information](#)

Learn more about IRDiRC from its members!



IRDiRC proudly launched **a series of interviews** with its members to discover how the passion for rare diseases research, policy and advocacy drives our members to make an impact globally and serve better the rare diseases community.

Don't miss out the interview with **Stefano Benvenuti** (IRDiRC FCC member) representing **Fondazione Telethon (Italy)**.

[Watch the video](#)

New Orphanet Journal of Rare Diseases series looking for authors on MedTech

As one of the outcomes of the **MedTech for Rare Diseases Working Group** led by IRDiRC members **Anneliene Jonker** and **Marc Doms**, the **Orphanet Journal of Rare Diseases** has decided to focus a thematic issue on the questions regarding the **research and development of orphan devices and medical technologies** that are being developed for Rare Diseases patients.



Find more information about the *Orphanet Journal of Rare Diseases*' upcoming issue [here](#). Submission guidelines can be found [here](#).

To submit an article to the series, please use the *Orphanet Journal of Rare Diseases*' [submission system](#).

Commentary on the 10 years of IRDiRC is now available as full text

We are delighted to announce that the **commentary on the 10 years of progress and challenges of IRDiRC** that was previously published in the prestigious journal *[Nature Reviews Drug Discovery](#)* is now **available as a full text**.

The article **summarizes IRDiRC's vision and goals and highlights its**

achievements and prospects after its first decade (2011–2021).

The commentary has been authored by **Lucia Monaco** (former IRDiRC Consortium Assembly Chair 2018–2021) along with **David Pearce, Galliano Zanello, Gareth Baynam, Anneliene Jonker, Daria Julkowska, Adam Hartman, Daniel O'Connor, Chiuhui Mary Wang and Durhane Wong-Rieger.**

The **supplementary information** included with the commentary provides a **visual summary** of IRDiRC's vision and goals, its international reach, the organization of its committees, Working Groups, and Task Forces, including an overview of its communication activities, and some rare diseases metrics.

- The **full article is available** [here](#).
- The **supplementary information** can be downloaded [here](#).



Leadership and Membership Changes

- IRDiRC member **[Iberoamerican Alliance for Rare Diseases \(ALIBER\)](#)** has appointed **Karla Ruiz de Castilla** as their new representative. **Karla Ruiz de Castilla** is the current **Secretary of ALIBER** and **President of [ESPERANTRA](#)**. She is based in Peru and will replace **Alba Ancochea** on the **[Patient Advocates Constituent Committee \(PACC\)](#)**.
- **[Japan Agency for Medical Research and Development \(AMED\)](#)** named **Susumu Kusunoki, Professor** and **Chairman of the Department of Neurology Faculty of Medicine at Kindai University** as the **[new AMED representative](#)** in the **[IRDiRC Funders Constituent Committee \(FCC\)](#)**.

- IRDiRC member **Ultragenyx Pharmaceutical Inc** has appointed **Grania Crowley** as their new representative. She is the **Head of EU Patient Advocacy and Public Affairs** at **Ultragenyx (Switzerland)**, and is replacing **Tom Pulles** as the company representative in the **IRDiRC Companies Constituent Committee (CCC)**.

IRDiRC would like to express its gratitude to the previously appointed members for their contribution and involvement in the work of the Consortium over the past years. We hope that the collaboration with IRDiRC will continue in the future through the work of Task Forces and Working Groups.

IRDiRC wishes a warm welcome to the new representatives and we are looking forward to collaborate with them on various projects and initiatives.

IRDiRC IN EVENTS



Register now to attend online the International Conference on Clinical Research Networks (CRN) for Rare Diseases

The **International Rare Diseases Research Consortium (IRDiRC)** and the **European Joint Programme on Rare Diseases (EJP RD)** are jointly organizing a conference on **Clinical Research Networks (CRNs) for Rare Diseases in Paris (France)**.

CRNs offer a tremendous opportunity for **patients** and **multi-disciplinary teams** to **collaborate, share expertise, gain better understanding on rare diseases**, and **accelerate clinical research** and **innovation**. The objective of this conference is to gather experts from different continents to increase mutual knowledge on CRNs structure, activities and identify pathways to

stimulate collaboration and interoperability of these clinical research networks.

The conference will start on **December 1st, 2022** at **1:45 PM CEST**, and will end on **December 2nd, 2022**, at **1:00**

PM CEST.

Register to online CRN conference

Other IRDiRC events:

- Consortium Assembly In-Person Meeting: In Paris, France on November 30th –December 1st, 2022, back-to-back with the

International Conference on Clinical Research Networks (CRN) for Rare Diseases on December 1st – 2nd, 2022

- Consortium Assembly In-Person Meeting: In Berlin, Germany on March 14th – 15th, 2023

- RE(ACT) Congress In-Person: In Berlin, Germany on March 15th – 18th, 2023. Register at <https://www.react-congress.org/attendees/registration-2/>



NEWS FROM IRDiRC MEMBERS

FONDAZIONE



Fondazione Telethon (Italy) joins the **AGORA (Access to Gene therapies for RAre disease) Consortium**, a Europe-wide group focused on boosting access to life-saving gene therapies.

“Participating in AGORA is a further step, which will allow us to involve these figures at a European level so that we can implement joint actions at different levels to simplify the development of these complex drugs,

also reducing production costs, making the therapies more sustainable”, said **Francesca Pasinelli**, General Director at Fondazione Telethon.

“What we are doing with **Strimvelis** is the concrete implementation. Fondazione Telethon has in fact decided to take over

the commercialization of the **gene therapy for ADA-SCID** from Orchard Therapeutics, creating an ad hoc non-profit organization and thus bearing all the costs of management and maintenance on the market. Not only that, with a non-profit organization we will be able to 'donate' the therapy to less affluent patients who are not eligible for reimbursement by the National Health Service as non-European citizens or who do not have medical insurance. However, with six gene therapies abandoned by the industry from the market in the last two years alone, it will be necessary to mobilize a very broad commitment to reverse this trend: Fondazione Telethon and AGORA will work towards this goal on several fronts."

[More Information](#)

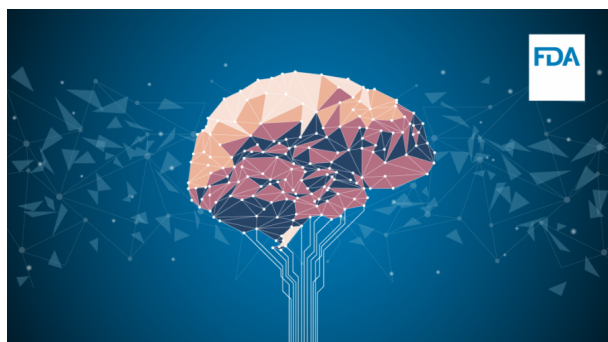
NIH: Grant opening for Pilot Projects Investigating Understudied Proteins

Associated with Rare Disease



The **US National Institutes of Health (NIH)** announced the **opening of a grant opportunity** that receives applications for **pilot projects to elucidate a role for understudied proteins associated with rare diseases**. Awards will support **generation of preliminary data and/or tools** around eligible understudied protein(s). This **funding opportunity announcement (FOA)** is intended to **jumpstart research on understudied proteins** that are associated with rare diseases and provide applicants with sufficient funding to perform basic biochemical and/or biological work to further the characterization of understudied proteins associated with rare disease. The earliest submission date is **September 17th, 2022**.

[More Information](#)



FDA launches a plan to Accelerate Access to Critical Therapies

As part of its requirements under the **Accelerating Access to Critical Therapies for Amyotrophic Lateral Sclerosis (ACT for ALS) legislation**, **FDA** has developed a plan describing actions the FDA intends to take to advance innovation that promotes and accelerates drug development for the treatment of **rare**

neurodegenerative diseases including **ALS**. For more information, visit **FDA's ACT for ALS webpage**.

The FDA Office of Orphan Products Development (OOPD) is pleased to announce **availability of funds to support clinical trials for rare diseases and conditions**. With a focus on developing **collaborative, efficient, and innovative clinical trials**, FDA launches this grant opportunity in support of a **new indication** or **change in labeling to address unmet needs in rare diseases**. Read more about this **Grant Funding Opportunity here**.

Takeda-sponsored Economist Impact Report - Connecting the dots:

Embedding progress on rare disease into healthcare

IRDiRC members **Takeya Adachi**, **Gareth Baynam** and **Durhane Wong-Reiger**

shared their contribution as an expert panel in the development of the **Economist**

Impact Report, sponsored by **Takeda** (**IRDiRC CCC** member). The report

focuses on **seven markets** in **Europe** and the **Asia-Pacific region** (Australia,

France, Germany, Japan, South Korea, the UK and Taiwan) and focuses on the analysis of the **speed of reimbursement**

decisions for eight treatments for rare diseases across the mentioned markets.



[More Information](#)



Indian Organization for Rare Diseases (IORD) partners with Global Players to improve Rare Diseases Visibility

The **Indian Organization for Rare Diseases (IORD)** partnered with **Project Y** and the **Rare Care Centre** at **Perth Children's Hospital (Australia)** to work on **improving the visibility** and **recognition of rare diseases** at a global scale.

Following **strong advocacy** from IORD, the health commissioner of India's state of Telangana approved a ground-breaking proposal to count rare disease patients within their state. The project is in collaboration with India's Accredited Social

Health Activist (ASHA) workers and it is aiming to include **rare diseases** in the **Indian National Census**.

[More Information](#)



Genetic Alliance

Genetic Alliance is seeking partners who are ready to deploy cWGS for undiagnosed individuals in LMIC

On the verge of a **number of Memorandum of Understanding (MOUs)** with a variety of **South East Asian, Indian, and African collaborators** to deploy millions of dollars in sequencers and reagents, **Genetic Alliance** is looking for contributors to deploy **clinical whole genome sequencing (cWGS)** for **undiagnosed patients** in **low- and middle-income countries**. The funding will be focused on operationalizing the >\$150M in instruments, reagents, software, technical assistance, participant data management, and cloud storage.

For more information, visit <https://ihopegenetichealth.org/> or contact **Sharon Terry** (Chief Executive Officer, Genetic Alliance) at sterry@geneticalliance.org.

Illumina supported the "**Rapid whole genome sequencing of critically ill**



pediatric patients from genetically underrepresented populations study in the **United Arab Emirates (UAE)**. Dr Tayoun and colleagues from **Al Jalila Children's Specialty Hospital (Dubai, UAE)**, recently published a **paper** in **Genome Medicine Journal** detailing a study of five critically ill newborns and infants in their hospital's intensive care unit (ICU). At the Genomics Centre, the team performed rapid whole-genome sequencing (rWGS) for trio analysis using samples from each patient as well as their parents. The average turnaround time was about 37 hours, and three out of five patients received a diagnosis.

[More Information](#)

OTHER NEWS



European Joint Programme on Rare Diseases (EJP RD)

The Fourth **EJP RD** General Assembly (GA) and Consortium Meeting took place last **September 13th-15th, 2022** in **Porto, Portugal**.

The **GA meeting** was the perfect occasion to bring together **all EJP RD members** to have an open discussion on the **progress performed** and **to define the new road map** for next year(s).

Search for **#EJPRDGeneralAssembly2022** on **Twitter** for updates!

[More information](#)

Upcoming Beacon for Rare Diseases Conference in London (UK) on October 10th

The **Rare Beacon's Drug Repurposing for Rare Diseases 2022** is shifting its programme to match the **new drug repurposing landscape**. This year's programme will highlight those who dared to

believe in drug repurposing and are **bringing repurposed medicines to patients at an accelerated rate**. Be sure to **register** to hear from:

- **The Centre for Drug Development,**
Cancer Research UK
- **Medicines and Healthcare products**
Regulatory Agency (MHRA)
- **NHS England and NHS Improvement**
- **REMEDI4ALL**
- and more!



[More information](#)

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