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IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

IRDiRC HIGHLIGHTS

Get to know the Funders Constituent Committee (FCC)!

Identifying the right strategies for rare diseases research projects and enabling the mechanisms for funding collaborations represent a major area of focus in advancing the development and access to diagnosis, care and treatment for patients living with a rare disease and their families.

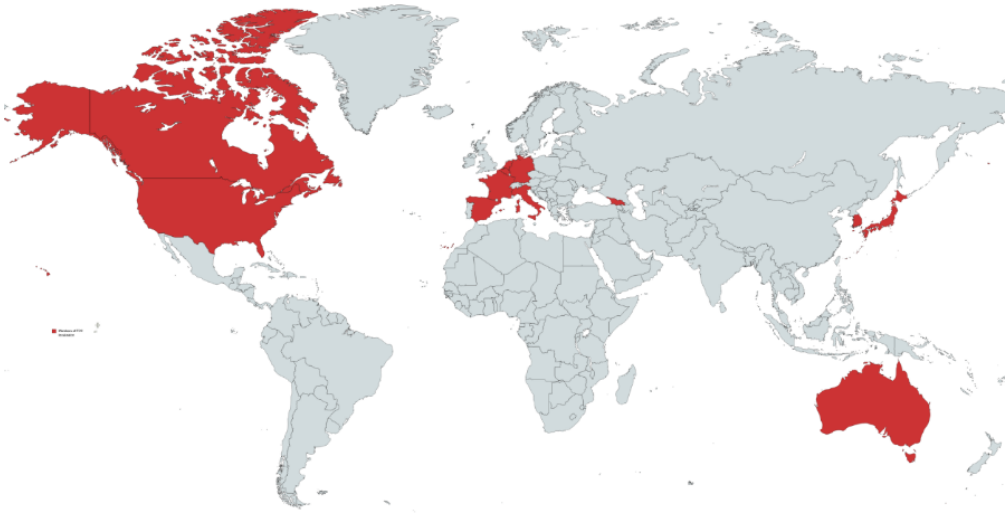
The **Funders Constituent Committee (FCC)** has played a pivotal role to this mission since the launch of the **International Rare Diseases Research Consortium** in 2011, and it continues to pursue new approaches in addressing different areas of research for improving and saving the lives of people with a rare disease around the world, being actively involved along the years in various **IRDiRC Task Forces and projects**: **Chrysalis Project** (focused on identifying key elements that make rare diseases research more attractive to companies in terms of research and drug development), **Primary Care** (addressing the prioritized research areas and challenges of RD primary care), **Enabling and Enhancing Telehealth for Rare Diseases Across the Globe** (performing a systematic review of telehealth models, their barriers and opportunities, as well as the creation of best practices for rare disease telehealth services), **Funding Models to Support the Spectrum of Rare Disease Research and Development** (determining the role of different factors that enhance or limit the funding at different stages of treatment development), **Polaris OS** (a solution that supports the long-term **Orphanet research platform** and IRDiRC ensuring the curation and automatic classification of funded research projects).

Having a strong motivation to work both on a national and global scale to help solve the many issues faced by rare disease patients, **Christopher McMaster**, **FCC chair** (2023-2026) expressed his ongoing interest in IRDiRC and his major trust in IRDiRC's mission: **"the international rare diseases research community is eager to share knowledge and experience and work collaboratively across borders in order to bring diagnoses and therapies to patients. In my role at the Canadian Institutes of Health Research the goals of IRDiRC have aligned with our strategic plan and that of the FCC to facilitate high-level coordination of funding initiatives to maximize the impact of rare diseases research projects by aligning strategies, avoiding unnecessary duplication, and identifying research funding gaps."**

"IRDiRC is the answer for the RD community to the complexity of the challenges we need to face. Thanks to the success of research on some RDs, I felt we are now all aware that ensuring treatments to most of the RD patients is feasible. But none of us could make it alone", says **Stefano Benvenuti**, **FCC vice chair** (Fondazione Telethon, 2023-2026). This year, **Fondazione Telethon** marked an exceptional accomplishment, **completing the transfer of the marketing authorization of Strimvelis for ADA-SCID in Europe** from Orchard Therapeutics, becoming the first non-profit organization to take on the commercialization of a gene therapy.

With 27 members currently, and continuing to grow, the FCC has a broad representation of research funding institutions from various regions around the world (North America, Europe, Asia, Australia).

*Legend - localization of FCC member institutions



More Information

New IRDiRC Publications

"Making Rare Disease Research Attractive to Companies" by IRDiRC Chrysalis Task Force

IRDiRC is delighted to announce the publication of the **Chrysalis Task Force** manuscript:**"Making Rare Disease Research Attractive to Companies"**. The Task Force worked on identifying key financial and non-financial factors that make rare disease research and development attractive to companies.

"It was a real pleasure working with such a talented IRDiRC Task Force. We also truly appreciate the critical input provided by the survey and interview respondents. All of them provided specific yet nuanced responses that were framed in an important general context. Their responses allowed the Task Force to compose and

frame what I consider to be an extremely informative report" says **Adam L. Hartman**, MD, co-chair of the Chrysalis Task Force and Program Director at the National Institute of Neurological Disorders and Stroke (NINDS) part of U.S. National Institutes of Health (NIH).

Read the publication

"Defining rare conditions in the era of personalized medicine"

IRDiRC proudly presents the new publication developed by the **Therapies Scientific Committee (TSC)** members **Daniel O'Connor, Annemieke Aartsma-Rus, Anneliene Jonker**, and **Regulatory Scientific Committee (RSC)** member, **Michela Galbado – "Defining rare conditions in the era of personalized medicine".**

"Despite these consensus features, there is currently no common global agreement on the impact and widespread application of advances in molecular sciences and pathology on the definition of a rare condition". The commentary article offers an insight on "the impact of defining rare conditions in the era of personalized medicine, including subsetting of common conditions, subsetting of rare conditions, individualized treatment options and shared molecular entity conditions".

Read the publication

New Member of the Consortium Assembly

IRDiRC is pleased to welcome **Nicklaus Children's Hospital** (USA), represented by **Daria Salyakina**, Director, Center for Precision Medicine, as the new member of the **Funders Constituent Committee (FCC)**.

Nicklaus Children's Hospital

Leadership Changes

IRDiRC is pleased to announce the election of **Vinciane Pirard**, Lead Scientific Advocacy and Insights, Global Medical Affairs Rare Diseases, at **Sanofi** (Belgium) as the new **chair of the Companies Constituent Committee (CCC)** for a mandate of 3 years, and the election of **Adriana Huertas-Vazquez**, Senior Director, Global Medical Affairs, at **Illumina, Inc.** (USA) as the **vice chair**.

IRDiRC would like to express its gratitude to the previously appointed chair, **Katherine Beaverson** (**Pfizer**, USA) for her contribution and involvement in the work of the Consortium over the past years.

IRDiRC wishes a warm welcome to the new chair and vice chair and it is looking forward to a fruitful collaboration on various rare diseases projects.

Membership Changes

Therapies Scientific Committee (TSC)

New members

- **Alaa Hamed**, Global Head of Medical Affairs Rare Diseases, **Sanofi**, USA
- **Diana Kwast**, Board member **Dutch Pituitary Foundation**, **ePAG Endo-ERN**, The Netherlands
- **Rajesh Krishna**, Distinguished Scientist and Global Lead, Rare Diseases Center of Excellence, **Certara**, USA
- **Martina Kawome**, Ophtalmologist, Department of Ophtalmology, **College of Health Sciences (Harare)**, Zimbabwe

We warmly thank **Dr. Maurizio Scarpa** and **Prajnya Ranganath**, former TSC members, for their invaluable contribution to IRDiRC during their mandate.

Diagnostics Scientific Committee (DSC)

New members

- **Charles Steward**, Head of Patient and Participant Engagement, Genomics England, UK
- **Florencia Braga Menéndez**, CO Director, Argentine Patient Alliance (ALAPA), Argentina

Funders Constituent Committee (FCC)

Change of representation

- **Sapna Mahajan**, Director, Research and Innovation, Genome Canada, replacing **Ivana Cecic**

IRDiRC wishes warmest welcome, and is looking forward to a successful collaboration!

IRDiRC EVENTS

Upcoming IRDiRC internal (closed) events:

- **In-person Consortium Assembly Meeting:** Location - **Montréal (Canada)**, on **October 3-4, 2023** with the support of **Canadian Institutes of Health Research - Institute of Genetics (CIHR-IG), Genome Canada, Canadian Organization for Rare Disorders;**
- **Online Consortium Assembly Meeting**: on **December 6-7, 2023 (3-5 PM CET)**;

Collaborative events:

- **Canadian Satellite Meeting** (in collaboration with the **Canadian Institutes of Health Research**): Location - **Montréal, Canada**, on **October 4, 2023**

IRDiRC representation at events

We are delighted to announce that **IRDiRC Therapies Scientific Committee (TSC)** Vice Chair, **Anneliene Jonker** (University of Twente, The Netherlands), will give a talk on the drug repurposing guidebook at the **RExPO23 Conference**, that will take place on **October 25-26, 2023** in Stockholm, Sweden. Anneliene will participate in **Session 4 – Regulatory Perspectives**, on the first day of the conference (October 25).

Check out additional details at the following link: https://repo4.eu/rexpo23/

NCL 2023

Dr David Pearce, IRDiRC chair, recently attended the scientific advisory board & board meeting at the **Global Genes: 2023 RARE Advocacy Summit** in **San Diego (USA)**. In addition, **Dr David Pearce** also presented at the **18th International Congress on Neuronal Ceroid Lipofuscinoses** in **Hamburg (Germany),** on **"BDSRA- General announcement about new granting program & US Batten disease Centers of Excellence".**

OrphanDrug Congress Europe 2023
30 October - 2 November 2023 | Fira de Barcelona
STRATEGY, ADVOCACY & PARTNERING FOR THE GLOBAL ORPHAN DRUG INDUSTRY

The World Orphan Drug Congress (Barcelona, Spain) is an award-winning event with an exhibition that has grown to become the largest and most established orphan drugs & rare diseases meeting of its kind across the globe. IRDiRC will be present during the whole duration of the event at **booth 122, together with many IRDiRC members** who will participate or chair different sessions, including one dedicated to IRDiRC: **Showcase 2: The International Rare Diseases Research Consortium (IRDiRC)** on **October 31st from 15:10 CET** (**Dave Pearce** & **Samantha Parker**).

Full programme available: here.

NEWS FROM IRDiRC MEMBERS

Fondazione Telethon (Italy) and Orchard Therapeutics complete transfer of marketing authorization of Strimvelis for ADA-SCID in Europe

Following a positive opinion from the **European Medicines Agency (EMA)**, the transfer of marketing authorization of **Strimvelis** from **Orchard Therapeutics** has been approved by the **European Commission**. **Fondazione Telethon** will now be responsible for providing the gene therapy to eligible patients in the European Union.

Fondazione Telethon, one of the main Italian biomedical charities, and Orchard Therapeutics, a global gene therapy leader, announced the completion of the transfer of the marketing authorization for **Strimvelis**, a gene therapy approved by the EMA in 2016 for the treatment of adenosine deaminase severe combined immunodeficiency (ADA-SCID).

The marketing authorization transfer was approved on July 17 by the European Commission following a positive opinion from EMA. The European manufacturing and distribution rights have been fully transferred to Fondazione Telethon from its former holder, Orchard Therapeutics, which previously announced it would discontinue investment in and seek strategic alternatives for its programs in rare primary immune deficiencies, including **Strimvelis**.

Read the full press release

FDA: Clinical Studies of Orphan Products Addressing Unmet Needs of Rare Diseases - Clinical Trials Required

The FDA Office of Orphan Products Development (OOPD) will have another receipt date for its **Clinical Trial Program**.

The purpose of this **funding opportunity announcement (FOA)** is to fund clinical trials of products evaluating efficacy and/or safety in support of a new indication or change in labeling to address unmet needs in rare diseases or conditions. Additionally, through the funding of collaborative, efficient, and/or innovative clinical trials, FDA expects to increase the number of approved treatments for rare diseases and exert a broad and positive impact on rare disease drug development.

Deadline for applications: October 23, 2023

Apply here

Canadian Institutes of Health Research (CIHR) - Institute of Genetics: Call for funding applications

The **CIHR - Institute of Genetics** is delighted to present **new funding opportunities** that started in August, 2023:

1. **Pan-Canadian Human Genome Library;**
2. **Bringing Rare Disease Gene Therapies to Clinical Trial Readiness;**
3. **National Paediatric Rare Disease Clinical Trials and Treatment Network;**
4. **Improving the diagnostic odyssey for patients with rare diseases;**
5. **Updating administrative data and monitoring across health facilities for rare disease.**

Check out all published or upcoming funding opportunities at the following link: https://cihr-irsc.gc.ca/e/51605.html

More details

EURORDIS Open Academy Schools 2024

The **2024 edition of the Open Academy Schools** will take place **in-person in June in Barcelona (Spain)**.

The face-to-face intensive week of the Open Academy School on Medicines Research & Development (formerly EURORDIS Summer School) and the Open Academy School on Scientific Innovation & Translational Research (formerly EURORDIS Winter School) take place during the same week, incorporating parallel sessions, focusing on School-specific topics, and joint sessions focusing on patient engagement and leadership. Knowledge-sharing and networking opportunities are fostered throughout the week.

The programme will include off-site visits to research facilities/hospitals, lots of contact time with faculty and EURORDIS staff and the chance to network with fellow patient advocates and researchers, supporting each other in learning and practice during the training and beyond. Schools are delivered in English in a blended format, comprising e-learning modules, pre-training webinars and face-to-face intensive days.

Apply here

Education: Sanofi launches the Gaucher disease curriculum as part of the Rare Diseases University

The Rare Diseases University (RDU) is a **physician-oriented, interactive, digital Medical Education program**. It was designed to address the urgent need for better patient diagnosis and care for patients living with rare diseases around the world.

The content is developed with the assistance of a faculty of doctors working in the field of rare diseases, and includes a mixture of bitesize content, videos, podcasts, and in-depth training material. It's accessible online for free to health care professionals always, no matter the time zone, and is regularly updated with new data.

The RDU now covers **Fabry disease**, **Gaucher disease**, and **ASMD**, with more to come very soon.

Register

Rare Diseases International: "Engaging the UN System and Member States to Achieve Universal Health Coverage for Persons Living with a Rare Disease: A Blueprint for Leaving No One Behind"

Rare Diseases International (RDI) and the **Permanent Mission of Spain to the United Nations in New York** organized a hybrid event **"Engaging the UN System and Member States to Achieve UHC for PLWRD: A Blueprint for Leaving No One Behind"** on **September 21, 2023**.

This event highlighted the continuation of years-long advocacy by RDI and its partners, and the objective was to bring to attention the national successful actions implementing Universal Health Coverage for rare diseases, to explore methods to reduce the out-of-pocket expenses shouldered by PLWRD and their families, and to draw some quick wins for policy makers and stakeholders to implement.

More Information

Game Changers in Health: Unpacking the Pharma Package with Marc Dooms

Don't miss out the interview with the **Interdisciplinary Scientific Committee (ISC) vice chair**, **Marc Dooms** (Senior Orphan Drug Pharmacist at University Hospitals Leuven, Belgium), in the new episode of **"Game Changers in Health"** about how the revisions within the **EU Pharma Package** will impact patients' ability to access care and information, especially for the orphan devices.

Watch the video

OTHER NEWS

Together4RD Position Statement on Collaboration between European Reference Networks and Industry

Despite the remarkable strides that ERNs have made in connecting medical professionals, researchers, and patients across Europe, we believe there is so much more we can achieve. Currently, ERNs are not fulfilling their full potential in research, largely due to lack of resources and support, and the lack of collaborations that harness relevant expertise,

including with industry.

Together4RD's paper outlines strategic recommendations to address these challenges and enhance the efficacy of ERNs in the rare disease landscape: **"We firmly believe that by fostering stronger public-private partnerships and encouraging cross-sector collaboration, we can amplify the impact of ERNs and improve the lives of those who need it most"**.

Read the Statement

Call for proposals: Establishing novel approaches to improve clinical trials for rare and ultra-rare diseases

Under the **Horizon Europe Framework Programme**, a new two-stage call for proposals for **"Establishing novel approaches to improve clinical trials for rare and ultra-rare diseases"** was published on **July 27, 2023**.

The project generated from the topic should not only develop capacities and capabilities to execute innovative trial designs, but also plan to identify solutions to address scientific gaps as well as technical and operational challenges, and to collaborate/find synergies with relevant existing initiatives to establish a new, dedicated, rare disease specific and sustainable infrastructure. The project is expected to support innovation and optimise drug development for rare diseases with high unmet medical needs by focusing on clinical trials conducted for small populations and clusters of diseases with commonalities.

The deadline for applications is November 8, 2023 (17:00 local Brussels time).

More Information

ERICA & EJP RD Joint Conference in Amsterdam

The **ERICA** & **EJP RD** **Joint Conference** is scheduled for **November 21, 2023**, in **Amsterdam, The Netherlands**. This event aims to unite stakeholders in **European Reference Networks (ERNs)** and **Rare Disease (RD) Research**. It will bring together medical professionals, researchers, patient advocates, policymakers, and industry representatives interested in improving care and advancing research for rare diseases.

A pre-conference dinner on November 20th provides a networking opportunity. The conference signifies collaboration and knowledge exchange in the realm of ERNs and rare disease research.

More information

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.