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IRDiRC HIGHLIGHTS

The Foundation for Rare Disease welcomes IRDiRC Scientific Secretariat

The Foundation for Rare Diseases (France) will host the International Rare Diseases Research Consortium (IRDiRC) Scientific Secretariat for a period of 7 years, funded under the European Rare Diseases Research Alliance (ERDERA), starting the 1st of September 2024.

About the Foundation for Rare Diseases (Fondation Maladies Rares)

The Foundation for Rare Diseases is a non-profit scientific cooperation foundation dedicated to rare diseases research, created on 7 February, 2021, by decree of the French Ministry of Higher Education and Research, by 5 funding members:

- AFM Téléthon

- The Rare Disease Alliance (Alliance Maladies Rares)
- The National Institute of Health and Medical Research (INSERM)
- The Conference of General Directors of University Hospitals (CHU France)
- The Conference of University Presidents (CPU)

The foundation acts as a strategic hub to coordinate, federate and fund rare disease research.

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.

New IRDiRC Publications

The complexity of funding rare disease research: an IRDiRC assessment of the landscape

The complexity of the funding landscape for rare diseases (RD) research is due to many factors. Each type of funder has strategic goals guiding its investments. The International Rare Diseases Research Consortium (IRDiRC) convened RD experts to explore contributing factors and investment principles in RD funding. Similar to IRDiRC's previous work on motivating factors for company investments in RD research, the current Task Force found that return on investment (ROI) was a guiding principle. However, within the broader RD funding landscape, the definition of ROI varied between types of funders. While they shared funding requirements (e.g., scientific quality, health economics), funders employed both major investment and venture-type

instruments, demonstrating the ongoing need for flexibility in supporting RD funding. These observations warrant further analysis of the interactions and partnerships among all actors of RD research and the sustainability of RD research funding.

More about the **Funding Models to Support the Spectrum of Rare Disease Research and Development** Task Force: here.

Read the publication

Patient organizations: advocating for timely newborn screening & improved quality of life

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IRDiRC announces the release of a new publication on newborn

screening, coordinated by the Interdisciplinary Scientific Committee member, **Chiuhui Mary Wang** (Global Programme Director at Rare Diseases International): **"Patient organizations: advocating for timely newborn screening & improved quality of life"**. This paper, published in the Rare Disease and Orphan Drugs Journal, provides a unique exploration of the crucial role patient organizations play in advocating for NBS across different national, regional, and disease-specific contexts.

Have a look at the complete special edition of **Newborn Screening on "Policy, ethics and Patient Perspectives"**, now available open access: https://www.oaepublish.com/specials/rdodj.1271

More about NBS initiative: here.

Read the Publication

Leaving no patient behind! Expert Recommendation in the Use of Innovative Technologies for Diagnosing Rare Diseases

IRDiRC Task Force "Integrating New Technologies for the Diagnosis of Rare Diseases" has investigated the technological advances in the field of metabolomics, genomic sequencing and machine learning algorithms.

The group has recently published a paper in the Orphanet Journal: **"Leaving No Patient Behind! Expert Recommendation in the Use of Innovative Technologies for Diagnosing Rare Diseases"** which provides an overview of a broad spectrum of emerging diagnostic technologies involving bioinformatics and Artificial Intelligence (AI), showcasing their advantages, limitations and the current landscape of clinical adaption.

More about **Integrating New Technologies for the Diagnosis of Rare Diseases** Task Force: here.

Read the Publication

IRDiRC EVENTS

Upcoming **IRDiRC internal (closed) events**:

- **In-person Consortium Assembly Meeting:** Location - **Milan (Italy)**, on 16-17 October, 2024 with the support of Fondazione Telethon.
- **In-person IRDiRC Consortium Assembly - Scientific Committees Meeting (CA-SC)**: Location - **Brussels (Belgium)**, on 3-4 March, 2025.

Collaborative events:

- **Workshop on Funding Models for Rare Disease Research**: Location - **Milan, Italy**, on 17 October, 2024**.** Online registration available: here.

**Photo from the CA-SC Meeting in Shanghai, China (May, 2024)*

RE(ACT) Congress 2025 and IRDiRC Conference

A new edition of the **RE(ACT) Congress** will take place on 5-7 March 2025 in **Brussels (Belgium)**, organized jointly by the BLACKSWAN Foundation, International Rare Diseases Research Consortium and ERDERA.

The congress will hold 6 sessions, various plenary talks as well as poster exhibition:

- Session A: Diagnosing rare diseases: from NBS to machine learning
- Session B: Empowering data
- Session C: ATMPs: challenges and opportunities of today
- Session D: Impacts of rare diseases on patients, families, and society
- Session E: Funding models
- Session F: Drug repurposing in rare diseases

Check out the tentative programme at the following link: https://www.react-congress.org/program/full-program/

Register now

IRDiRC representation at events
Upcoming events

IRDiRC at 2024 World Orphan Drug Congress Europe

World Orphan Drug Congress Europe is the largest and most established orphan drug & rare disease event worldwide. Meet over 2000 attendees, hear from 250 leading speakers, and connect with 130 exhibitors as we bring together experts from the start-to-finish of orphan drugs, from regulation and policy to global pricing and gene therapy. The attendees include the most senior individuals across the industry,

including: R&D, Gene Therapy and Rare diseases, Precision/Personalised Medicine, Genetic Disorders, Patient Advocacy and Engagement, Associations in ATMPs, Investors, Regulatory Affairs Professionals, Governmental Bodies and many more. If you are a member of the European Reference Networks (ERNs), patient group organization, charity or government (including public health bodies, HTAs, regulators), you are eligible for a free VIP pass. IRDiRC Vice Chair, **Samantha Parker**, is part of the organizing committee, and multiple IRDiRC members will participate in different sessions and panels.

More information about the World Orphan Drug Congress here.

Dates:

🗓️22nd October 2024 | Pre-Congress Workshops | Hotel Catalonia Barcelona Plaza (Barcelona, Spain)

🗓️23rd – 25th October 2024 | Main Congress Days | Fira de Barcelona Montjuïc (Barcelona, Spain)

Follow **#WODC** on social media to stay up to date:

LinkedIn:https://www.linkedin.com/company/world-orphan-drug-congress-europe/

Twitter: https://twitter.com/orphan_drugs

Registration and More Information

ICoNS Meeting 2024

The International Consortium on Newborn Screening (ICoNS) 2024 Meeting will take place on 9-10 October, 2024, at the New York Academy of Medicine (New York, USA) and it will gather global thought-leaders and stakeholders from over 50 countries.

IRDiRC will be represented at the event by its **Companies Constituent Committee (CCC)** member, **Illumina** representative, **Adriana Huertas-Vazquez** (Senior Director Medical Affairs, Genetic Diseases, and Vice Chair of CCC).

More Details

Past events

13th International Conference on Rare and Undiagnosed Diseases

The 13th Conference of Undiagnosed Diseases Network International took place on 5-7 September, 2024, in Seoul, South Korea and marked the 10th anniversary of UDNI's establishment in 2014. IRDiRC was represented at the event by its chair, **Dr. David Pearce**, who presented on **"International Collaboration leading to successes in rare diseases"**.

The conference emphasized the importance of international cooperation and funding in rare disease research, and brought together over 282 experts from 30 countries.

This year's UNDI conference was co-hosted by the Wilhelm Foundation of Sweden and the Child Cancer and Rare Disease Project at Seoul National University Hospital (SNUH).

More Details

2024 ESG Global Leaders Conference

The ESG Global Leaders Conference is an event organized by United Nations that gathers leaders and decision-makers to advance sustainable development. This year, the third edition of the conference was focused on **"ESG Synergy: Uniting for Global Prosperity"**. The event took place in Shanghai, China, from 16-19 October, and IRDiRC was represented by **Dr. David Pearce**, who offered an online presentation on **"Global Collaboration and Joint Action on Rare Diseases"**.

In addition, **Dr. David Pearce** also participated in the Global MND Research Roundtable, in Melbourne, Australia (28-30 August), and presented the **"Global Collaboration for Clinical Trials"**.

NEWS FROM IRDiRC MEMBERS

Congressionally Directed Medical Research Programs - New Funding Opportunities

The Glioblastoma Research Program (GBMRP) released program announcements for the following funding opportunities:

- **Resource Development Award**
- **Hypothesis Development Award** (with Partnering Principal Investigator Option)

To view the funding opportunity announcement and submit a pre-application visit: https://ebrap.org/eBRAP/public/ProgramFY.htm?programFYId=629101

FDA-OOPD: Call for Grant Applications

The U.S. Food and Drug Administration, Office of Orphan Products Development (OOPD) supports and advances the development and evaluation of new treatments for rare diseases. The OOPD is pleased to announce that two calls for clinical studies are now open until 22 October, 2024.

a. **Clinical Studies of Orphan Products Addressing Unmet Needs of Rare Diseases** (RFA-FD-23-001: https://grants.nih.gov/grants/guide/rfa-files/RFA-FD-23-001.html)
b. **Clinical Trials Addressing Unmet Needs of Rare Neurodegenerative Diseases** (RFA-FD25-001: https://grants.nih.gov/grants/guide/rfa-files/RFA-FD-25-001.html)

Moreover, the OOPD held receipt dates for applications for clinical trials for rare diseases, natural history studies for rare diseases, and rare neurodegenerative studies in Fiscal Year (FY) 2024, and received 51 clinical trial applications, 53 natural history applications, and 15 rare neurodegenerative applications. Awards will be announced later in October.

More information

French National Research Agency (ANR): Call for Projects 2025

The **Generic Call for Proposals 2025 (AAPG 2025)** is the **French National Research Agency's (ANR)** main call. All scientific communities and all public and private actors involved in French research are concerned. It should allow

researchers from different scientific fields to access, in addition to their recurrent funding allocated, co-funding on a large number of research themes, whether finalized or not.

he AAPG 2025 call is structured into **57 research themes**, each corresponding to a scientific evaluation panel (CES).

- 38 research themes covering 7 main scientific fields (including 1 new theme in Physics);
- 19 research themes covering trans- or interdisciplinary challenges and integrating issues from various scientific fields.

The deadline of the call is 15 October, 2024.

More information

CIHR-IG - New Funded Programs

The Canadian Institutes of Health Research - Institute of Genetics is proudly announcing the latest funded programs:

- National Network Grant: **National Pediatric Rare Disease Clinical Trials and Treatment Network;**
- Team Grants: **Improving Diagnosis for Rare Disease Patients;**
- Team Grants: **Improving Health and Administrative Data and Monitoring for Rare Diseases;**
- Operating Grants: **Bringing Rare Disease Gene Therapies to Clinical Trial Readiness;**
- National Network Grant: **Bioinformatics, Computational Biology and Health Data Science Training Platform.**

In addition, the institute organized a two day workshop on **DNA on Loan 2**, led by **Indigenous peoples and scholars** on biobanking and genomics, and how to best try to ensure there are no inequities in research and healthcare for Indigenous peoples in Canada, while taking into account CARE and OCAP principles.

EFPIA - Data Sharing Playbook

The European Federation of Pharmaceutical Industries Association (EFPIA), developed together with the European Infrastructure for Translational Medicine (EATRIS), Teamit, Lygatureand Information Technology for Translational Medicine, the **Data Sharing Playbook**, a tool that aims to help all participants in pre-competitive projects by facilitating internal processes and decisions to accelerate the provision of data in IMI/IHI projects. The Playbook intends to be a user-friendly and comprehensive tool for all parties involved in data sharing, and to provide strategies and resources for navigating common challenges associated with data provision with the scope to advance collaborative and impactful research and to improve health outcomes.

Check out the Data Sharing Playbook at the following link:

https://www.ihi.europa.eu/sites/default/files/uploads/Documents/ProjectResources/IMI_IHI_DataSharingPlayBook_2024.pdf

Read the Press Release

New Publication: "Orphan and paediatric medical devices in Europe: recommendations to support their availability for on-label and off-label clinical indications"


Perspective

Orphan and paediatric medical devices in Europe: recommendations to support their availability for on-label and off-label clinical indications

Tom Melvin, Marc M. Dooms, Berthold Koletzko, Mark A. Turner, Damien Kenny, Alan G Fraser ...show all

Received 25 Jul 2024, Accepted 10 Sep 2024, Published online: 20 Sep 2024

Cite this article https://doi.org/10.1080/17434440.2024.2404257

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IRDiRC Interdisciplinary Scientific Committee (ISC) Vice Chair, **Marc Dooms**, and Therapies Scientific Committee (TSC) Vice Chair, **Anneliene Jonker**, have co-authored a publication in *Expert Review of Medical Devices* that aims to examine the **use of medical devices in orphan and paediatric conditions** with relevant regulatory concerns and associated guidance that would recommend possible policy and practice interventions to ensure continued availability of essential devices for children and persons with rare diseases.

Read the Publication

New Publications on Rare Diseases

The chair of the Interdisciplinary Scientific Committee, **Dr. Gareth Baynam**, has co-authored several publications on rare diseases:

Operational description of rare diseases: a reference to improve the recognition and visibility of rare diseases - together with **Chiuhui Mary Wang** (ISC member), **Diego Ardigo** (CCC member), **Helen Malherbe** (ISC member) and **Anne Pariser** (RSC member); Access the publication: https://pubmed.ncbi.nlm.nih.gov/39261914/

- **Increasing Diversity, Equity, Inclusion, and Accessibility in Rare Disease Clinical Trials** - with Charles Steward (DSC member) and Anne Pariser (RSC member); Access the publication: https://pubmed.ncbi.nlm.nih.gov/38977611/
- **FastHPOCR: pragmatic, fast, and accurate concept recognition using the human phenotype ontology;** Access the publication: https://pubmed.ncbi.nlm.nih.gov/38913850/
- **Indigenous-led precision public health: a new starting point;** Access the publication: https://pubmed.ncbi.nlm.nih.gov/39267644/

Innovative Health Initiative 2025 Call

IHI has just pre-published its January 2025 call for projects, including for rare diseases. The call is directed at public-private consortia where industry and public partners, including academia, regulators, physicians and small companies collaborate to address common challenges.

While previous IHI calls defined challenges applicants should address, call 9 will invite applicants to identify untapped opportunities in the **IHI Strategic Research and Innovation Agenda**.

Proposals will have to address an unmet public health need, require a large-scale, ambitious and cross-sector public-private partnership with clearly described impacts on society, economy and science.

An **informative online session** will take place on 10 October, which will cover the scope of IHI Call 9 and how to get the most of the platform. A **brokerage event** will be organized in Brussels, Belgium, on 12-13 November and online on 14 November.

More information

OTHER NEWS

Call for candidates to ERDERA's Multistakeholder Advisory Board (MAB)

The European Rare Diseases Research Alliance (ERDERA) is seeking experts to join its **Multistakeholder Advisory Board (MAB)**. This is the chance to help shape the future of rare disease research across Europe.

The programme is looking for professionals in the following fields:

1. Rare Disease Researchers
2. Healthcare Professionals
3. Public-Private Partnership and Industry Professionals
4. Digital Health and Data Scientists
5. Regulatory, HTA and Policy Groups
6. Funding and Grant Management Bodies

7. Education and Training Personnel
8. Strategic Coordinators
9. Ethics and Patient Consent Groups
10. Legal Experts in Healthcare and Researchers

Find out more information: https://erdera.org/news/call-for-nominations-join-the-erdera-multistakeholder-advisory-group/

Deadline for applications submission: **October 31, 2024**.

More information

DeCODe Project Kick-off: Advancing Paediatric and Orphan Medical Devices

The European commission co-funded **DeCODe consortium** held its kick-off meeting in Brussels on 9-10 September 2024. This two-year ground-breaking initiative aims to catalyse innovation and address the unique healthcare needs of people living with rare diseases (PLWRD), specifically children. This collaborative group, comprising clinicians, researchers, industry experts, and regulatory authorities, will develop a platform for developing safe and effective paediatric and orphan medical devices.

Read the Press Release

The Scientific Secretariat of IRDiRC is supported by the The European Rare Diseases Research Alliance (ERDERA) that has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.

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