Get to know the Regulatory Scientific Committee (RSC)!

The Regulatory Science Committee (RSC) is primarily focused on IRDiRC’s goal 2, the development of new therapies for people with rare diseases. The RSC includes a diverse group of experts from around the world from regulatory bodies, patient groups, biotech and pharmaceutical industry, not-for-profits and clinicians and scientists. They are involved in the regulatory process from different viewpoints, including from regulatory agencies and policy developers, the patient and caregiver perspective, drug developers and the treatment community. New to IRDiRC in 2022, the RSC has been exploring gaps in regulatory science with the aim to facilitate and accelerate the development, regulation and approval of rare disease therapies for the benefit of patients. Because regulatory science is involved in all areas of drug development, the RSC is also working to develop collaborations with other IRDiRC committees, and participate in IRDiRC task forces and work groups to better address regulatory questions that could increase the impact of IRDiRC activities.

Most recently the RSC has been:

- Working on a landscape analysis of global rare disease therapeutics approvals to understand possible differences among regions and identify areas of potential harmonization that could be leveraged
- Exploring existing models of global reciprocity for Orphan drugs
- Exploring current practices for advanced therapeutics to facilitate regulatory science excellence worldwide
Group priorities for 2024-2025 include better identifying and defining regulatory science topics that could be of most benefit for rare disease therapeutics, and to facilitate the work of other IRDiRC committees to assist their work in therapeutics development, identification of patient and caregiver priorities and facilitating the design, conduct and feasibility of good quality research for more rare diseases, especially those for very low prevalence and less attention in research. The feasibility of assessing approving advanced therapeutic products for small numbers of patients is one area of interest for cross-IRDiRC exploration that has been identified.

The RSC has recently welcomed two new members to the committee, Marco Rizzi and Claudia Saidman, from Australia and South America in order to more fully represent regulatory practices throughout the world.

**New Interdisciplinary Scientific Committee Members**

We're thrilled to introduce you the 5 new members of the Interdisciplinary Scientific Committee:

- **Chiu-Hui Mary Wang** - Global Programme Director at [Rare Diseases International (RDI)](https://www.rare-diseases.org), Italy 🇮🇹
- **Elmira Safarova** - Founder and CEO [Rarus Health SPA](https://rarushealthspa.com), Chile 🇨🇱
- **Robert Allaway** - Principal Scientist, [Sage Bionetworks](https://sagebionetworks.org), USA 🇺🇸
- **Ritu Jain** - Founder [DEBRA International](https://www.debrainternational.org), Singapore 🇸🇬
- **Chun-Hung Chan** - Director at [Sanford Biobank](https://biobank.sanfordhealth.org), USA (replacing Mahsa Shabani, former ISC member) 🇺🇸

We can't wait to start working with you!
New Regulatory Scientific Committee Members

We extend the warmest welcome to the newly appointed Regulatory Scientific Committee members:

- Claudia Saidman - Director of Clinical Research and Drug Authorization at ANMAT, based in Buenos Aires, Argentina
- Marco Rizzi - Associate Professor of Law, University of Western Australia, based in Perth, Australia

We are looking forward to tackling exciting projects together!

More Information

New IRDiRC Publications

We proudly announce two groundbreaking publications that help pave the way for the future of research data sharing and information governance that were developed by the Machine Readable Consent and Use Conditions Task Force in partnership with the European Joint Programme on Rare Diseases (EJP RD).

1) Getting your DUCs in a row – standardising the representation of Digital Use Conditions

Link: https://www.nature.com/articles/s41597-024-03280-6

This publication reports a robust, standard data structure that addresses the complex challenge of responsibly sharing research data, healthcare records, biosamples, and other biomedical resources. The Digital Use Conditions (DUC)
framework aims to streamline the management of these resources, while respecting applicable use conditions. The DUC data structure balances the need for clear rules with the flexibility required for diverse applications, ensuring that patient care and scientific discovery are advanced responsibly.

2) Common conditions of use elements. Atomic concepts for consistent and effective information governance

Link: https://www.nature.com/articles/s41597-024-03279-z

This publication introduces a core lexicon of 20 essential terms, refined from an initial set of 76, that digitally represent standardized ‘use condition’ concepts. Devised in conjunction with biobanks and registries, especially within the European Joint Programme for Rare Diseases, this new concept list will underpin data and sample sharing activities that are both practical and widely applicable. Specifically, by creating standardized Sharing Policy Profiles, one can manage and share biological resources more effectively, supporting software, training, and real-world projects.

Addressing Diagnostic Gaps and Priorities of the Global Rare Diseases Community: Recommendations from the IRDiRC Diagnostics Scientific Committee

A new paper developed by the IRDiRC Diagnostics Scientific Committee on “Addressing Diagnostic Gaps and Priorities of the Global Rare Diseases Community: Recommendations from the IRDiRC Diagnostics Scientific Committee”, has been recently published in the European Journal of Medical Genetics, as part of the Special Edition on “Globally Trending Topics in Rare Diseases”, having as guest editors IRDiRC Vice Chair, Samantha Parker, and former IRDiRC Chairs Lucia Monaco & Paul Lasko.
"People with rare diseases often face unmet health needs, including access to screening, diagnosis, therapy, and comprehensive health care. These challenges highlight the need for awareness and targeted interventions, including comprehensive education, especially in primary care. The majority of rare disease research, clinical services, and health systems are addressed with specialist care."

The Primary Care Task Force brought together representatives from the stakeholders required to identify the priority research areas in primary care that need to be addressed to deliver against the IRDiRC goals, identify current state of play, and identify challenges and opportunities in rare diseases primary care.

Read more about the Primary Care task force: [here](#).

**Newborn screening in Mexico and Latin America: present and future**

We're pleased to share a new publication on "Newborn screening in Mexico and Latin America: present and future", part of the Newborn Screening Initiative Real-World Applications and Technologies special edition, now fully available in the Rare Disease and Orphan Drugs Journal.

"The first newborn screening program (NBS) to be implemented in Latin America was in Mexico in 1974, eleven years after the initial NBS programs in other parts of the world. In the last 50 years, progress has been made in implementing and expanding NBS in Mexico and across Latin America, yet children across the region do not fully benefit from this effective public health strategy."
The publication highlights what is the progress in the implementation of expanded NBS in Latin America and what are the challenges faced by its complex ecosystem.

More publications on NBS: [https://www.oaepublish.com/specials/rdodj.1270](https://www.oaepublish.com/specials/rdodj.1270)

**IRDiRC EVENTS**

**Upcoming IRDiRC events:**

- **In-person IRDiRC Consortium Assembly Meeting:**
  - Location: Milan (Italy), on October 16-17, 2024 with the support of Fondazione Telethon.

- **In-person IRDiRC Consortium Assembly - Scientific Committees Meeting:**
  - Location: Brussels (Belgium), on March 3-4, 2025.

**Collaborative events:**

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

- **RE(ACT) Congress** - Location: Brussels, Belgium, on March 5-7, 2025, in partnership with BLACKSWAN Foundation & The European Rare Diseases Research Alliance (ERDERA).

**More Information:** [https://www.react-congress.org/](https://www.react-congress.org/)

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

**IRDiRC representation at events**

**Upcoming events**
We are delighted to join the ICMRA ‘Rare’ Symposium and Workshop 2024, a multi-stakeholder event focusing on rare diseases organized by the International Coalition of Medicines Regulatory Authorities (ICMRA) and hosted by Swissmedic. This exclusive symposium is open to selected stakeholder organizations and regulators, providing a unique platform to discuss and address the pressing challenges in the field of rare diseases. The symposium will be followed by a regulators workshop on September 17, which is open exclusively to regulators from ICMRA participating authorities. Regulatory Scientific Committee Vice Chair, Violeta Stoyanova-Beninska (EMA, The Netherlands), is part of the program committee and IRDiRC Chair, David Pearce (Sanford Research, USA), is among the confirmed participants.

- **Where?** LAC Lugano, Switzerland
- **When?** September 16, 2024, 12:00 – 18:30 (local time)

For more information about the event, including registration, hotel booking, and travel details, please visit the designated event website.
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

**Past events**
Great Plains Rare Disease Summit

In the United States, there are around seven thousand rare diseases that affect 25 to 30 million people.

The Great Plains Rare Disease Summit, organized by Sanford Research, took place on May 16-17, and brought together distinguished experts, clinicians, and dedicated community advocates to discuss about the learnings related to rare diseases. The gathering was designed to facilitate discussions on the latest developments in rare disease research and support opportunities for the RD community.

More Details

IHI session: ‘Breaking the Rare Disease Deadlock through Multi-Stakeholder Collaboration’

IRDiRC was represented at the BIO International Convention in San Diego (USA) by Deborah Requesens (FCC Member & Director, JumpStart Program, Orphan Disease Center/University of Pennsylvania, USA), who spoke in the session organised by the Innovative Health Initiative (IHI) on "Breaking the Rare Disease Deadlock through Multi-Stakeholder Collaboration" on Wednesday, June 5th. The session featured collaborative research model case studies on rare and orphan diseases from both sides of the Atlantic that are making practical contributions to the advancement of research and drug development in this highly challenging field.

More Details

NEWS FROM IRDiRC MEMBERS
Announcement of Grant Funding Opportunity for Rare Disease Research

The Food and Drug Administration’s (FDA) Office of Orphan Products Development (OOPD) is pleased to announce availability of funds for fiscal year (FY) 2025 to support clinical trials for rare diseases and conditions. The purpose of this funding opportunity announcement 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 submission: October 22, 2024

Additional Information and Contact: Katherine Needleman, Director, Orphan Products Grants Program. Email: katherine.needleman@fda.hhs.gov

Foundation for Rare Diseases (France) - Open call for grants applications

The Foundation for Rare Diseases has launched a series of calls for proposals available in 2024:

- Identifying new therapeutic molecules for rare diseases - focused on therapeutic molecules in order to develop new treatments for patients suffering from rare diseases
- Improving the quality of life of patients with sickle cell disease with MCGRE and Novo Nordisk
- Social and therapeutic innovations to improve the quality of life of young rare disease patients and their families with FE-IRCEM
- Human and Social Sciences & Rare Diseases
Moreover, the Foundation has organized its **2024 Annual Scientific Symposium** on June 18 at the Cordeliers campus in Paris (France).

**Global Genes RARE Advocacy Summit**
Registration for the **2024 Week in RARE** is officially open! Taking place **September 25-28 in Kansas City** (MO, USA), get ready for 4 days packed with sessions on Community & Capacity Building, Empowering the RARE Individual, Becoming a Research Ready Organization, and Promoting Equity in Healthcare. Week in RARE combines the **RARE Health Equity Forum** and **RARE Advocacy Summit**, in addition to the **RARE Champions of Hope** awards ceremony and annual membership meetings for Global Advocacy Alliance and RARE Corporate Alliance. This is a unique opportunity to gather and engage rare disease advocates and leaders in the same space for conversation. **Register here: [https://go.globalgenes.org/4bAyod5](https://go.globalgenes.org/4bAyod5)**

**Made in India, Missing in India: The Orphan Drug Access Challenge**
Boston, USA: By 2025, the Indian pharmaceutical industry is anticipated to hit the $100 billion mark. Despite this growth projection, accessing rare disease drugs remains a significant challenge in India. According to **Indian Organization for Rare Diseases (IORD)** CEO & President Prof **Ramaiah Muthyala**, although India produces all 450 of the world’s orphan medications (APIs), these drugs are often unavailable or unaffordable in India. Prof Muthyala highlighted this issue at the World Orphan Drug Congress 2024 held in Boston, USA on April 25, 2024.

During his presentation on **“Innovative & sustainable ways to increase access to treatments,”** Prof Muthyala emphasized the importance of innovation in addressing the scarcity of orphan medications, especially in impoverished nations like India. He acknowledged the efforts of the Indian government in promoting pharmaceutical innovation through schemes like the Pharmaceutical Technology Upgradation Assistance Scheme. However, he also cautioned against the misuse and abuse of invention and
innovation, distinguishing between the two as the emergence of new ideas or products and the creation of economic value through the intersection of discovery and invention.

Despite the challenges, India has been successful in providing affordable medications, partly due to its innovative approach, including the use of “Jugaad technology,” which focuses on finding the most economical solutions to problems. This approach has contributed to India’s prosperity and its position as a global leader in the pharmaceutical industry. With over 60,000 brands, India leads the world in generic brand supply, with four Indian companies among the top generic firms globally. However, there is still work to be done to ensure that rare disease drugs are accessible and affordable to all who need them in India.

More Information

The NINDS Nonprofit Forum
The National Institute of Neurological Disorders and Stroke (NINDS) Nonprofit Forum will take place July 23-24 virtually and in person at the Neuroscience Center Building in Rockville MD (USA).

The Nonprofit Forum, organized since 2005, brings together organizations that represent the many neurological diseases within the NINDS portfolio. It is a valuable venue for the organizations to collaborate and share lessons learned among themselves and for NINDS to understand how we can most effectively work with organizations to address research needs and challenges.

The Forum also welcomes poster submissions. Share your best practices, lessons learned, new programs, research, and successes on such topics as: the importance of direct engagement with basic researchers, lessons learned and case studies in advocacy led registries or natural history studies, non-profit research funding, public private partnerships, and other topics of interest to the audience. For additional instructions on poster submissions please contact Rebekah Corlew (Rebekah.corlew@nih.gov).

Check out the full agenda: https://nindsnonprofitforum.com/
OrphaDev4Kids project approved
The European Paediatric Translational Research Infrastructure (EPTRI) announces the launch of a collaborative project that aims to address the issue of Orphan medical devices (MDs) to be used in the paediatric population. The proposal plans to implement a complex innovation ecosystem in the orphan and paediatric MDs field in order to maximise the value-for-patients of the research and development projects that will support academic bodies, scientific societies, devices developers, particularly small/medium-sized companies (SMEs) and non-governmental organizations (NGOs) through all development phases from the idea conceptualization to the market access.

This proposal will focus on 3 specific orphan and paediatric case studies, one dealing with osteogenesis imperfecta and two concerning neonatal cardiology area, both referring to Cyanotic Congenital Cardiac Diseases.

Patient Advocacy Certificate Training
Professional Patient Advocates in Life Sciences (PPALS), in conjunction with Sanford Research created the Patient Advocacy Certificate Training (PACT) course of study for health and life science professionals and leaders of patient advocacy organizations (PAO) to enhance their professional development. This specially designed curriculum focuses on increasing the competencies and expertise of patient advocates in the life sciences and assisting PAO senior leadership and board members to work on their governance and infrastructure skills.

EURORDIS "Rare on Air" Podcast - Ayça Şahin's Story
Don't miss out the latest episode of "Rare on Air", of Ayça Şahin. Ayça, a PhD student living with Spinal Muscular Atrophy, shares her personal journey through the diagnostic process. Her story underscores the critical need for early and accurate diagnoses for rare disease patients across Europe, as well as the value of new treatments being researched, developed and made accessible.

Where to Listen:
• Spotify
  (https://open.spotify.com/episode/2Bs1lWNe5LWJFO5rq7MeUG?si=rMj8KnI2QTqDM6MnG3XnKq&nd=1)
• Amazon Music (https://music.amazon.fr/podcasts/d8ce0e9f-b50b-42b5-add9-b69dcb40e57/rare-on-air)
• YouTube (https://www.youtube.com/watch?v=V6QuQQkFyU)

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**OTHER NEWS**

**Driving Innovation in Rare Disease Research:**
**The EJP RD Final Conference 2024**

The EJP RD Final Conference was held on 27-28 May 2024 in Bari, Italy. This final conference brought together leading experts, researchers, and policymakers to discuss the advancements and future directions in rare disease research.

The conference not only celebrated the achievements of the EJP RD but also laid out a strategic path for future endeavours. Looking forward to continuing this journey of innovation and collaboration in rare disease research!
Exciting Job Openings at ERDERA: Project Managers for Rare Diseases

The European Rare Diseases Research Alliance (ERDERA) coordination team at INSERM, is thrilled to announce two openings for Senior Project Managers.

Hear directly from our team members about their experience in working in rare disease research coordination in this video (link: https://www.youtube.com/watch?v=S9-S-412DM8&t=1s).

Join ERDERA to make a significant impact in the field of rare diseases and collaborate with leading experts and institutions across Europe and beyond.

1. **Project Manager - Funding & Education**: Oversee collaborative research funding and training initiatives within the ERDERA programme. Your role will involve coordinating activities, supporting research teams, and ensuring the successful execution of funding and training programmes. Check the full job description: https://www.ejprarediseases.org/wp-content/uploads/2024/06/2024_ERDERA_Job-Description_Project-Manager_Funding-Education.pdf

2. **Project Manager - Data & Knowledge Hub**: Oversee data management and expertise services. Your responsibilities will include coordination and support of activities on rare diseases data infrastructures, as well as assistance to the methodological and regulatory expertise hub. Check the full job description: https://www.ejprarediseases.org/wp-content/uploads/2024/06/2024_ERDERA_Job-Description_Project-Manager_Data-and-Knowledge-Hub.pdf

More information on ERDERA.

**Apply Now**
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.

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