Get to know the Therapies Scientific Committee (TSC)!

The Therapies Scientific Committee (TSC) is primarily focused on helping to deliver IRDiRC’s goal 2, the development of new therapies for people with rare diseases. The TSC is a team of experts with different experience, such as industry, academics, clinicians, patient engagement and regulatory bodies. They work on a variety of aspects of therapy development, whether it is understanding which diseases are under researched, to developing tools for therapy development (such as the Orphan Drug Development Guidebook, but also on developing recommendations for patient-entered outcome measures, small population clinical trials, and repurposing). As such the aim of the committee is to analyse gaps and opportunities throughout the therapy development process and to provide solutions. An important recent evolution in the focus of the TSC has been to include medical devices as well as medicines in the scope of the work programme.

Most recently the group has worked on:

- Developing a guidebook for repurposing for rare diseases (Publications: Drug repurposing for rare: progress and opportunities for the rare disease community)
Contributing to a review on the legislation for orphan medical devices, highlighting the functional, technical, medical and regulatory issues around the research and development of such devices (Publication: MedTech Innovation & Rare Diseases, link: https://www.biomedcentral.com/collections/MTIRD);

Supporting the development of genetic N-of-1 therapies;

Defining rare diseases in the era of personalised medicine (link: https://www.nature.com/articles/d41573-023-00145-2).

Setting up the group priorities for 2024, some of the topics planned to be further investigated include: digital biomarker development, regulatory endorsed patient-relevant endpoints, drug-device combination products (together with the Interdisciplinary Scientific Committee), paediatric patient engagement (together with the Patient Advocacy Constituent Committee), and guidance for small population clinical trials. Some of this work is done conducted within the committee, while some is tackled at a larger scale with a Task Force or Working Group. Increasingly, the TSC is focussing on therapy development at large, in which a therapy can either be a drug, a medical device, a drug-device combination or a non-pharmaceutical-non-device solution, such as a nutritional therapy.

One membership position opening is foreseen for the Therapies Committee in the next months. The TSC will be looking for a committed candidate, a team player, ideally from outside Europe, that has fresh ideas on therapy development for patients with rare conditions.

Apply to join IRDiRC's Regulatory Scientific Committee
The International Rare Diseases Research Consortium’s (IRDiRC) Regulatory Scientific Committee (RSC) has two openings for new members who are from the following regions: Africa, Australasia or South America.

We are looking for rare disease experts from these regions with the following qualities:

- Expertise in drug and biological product regulation, regulatory review, oversight, or policy development/legal expertise;
- Knowledge on the medicines regulatory landscape in their region;
- Expertise from an industry, regulatory agency, academia or patient advocacy;
- Able to commit to monthly teleconferences, a yearly meeting and regular committee activists and email correspondence.

Interested candidates are invited to send their resume, biosketch and letter of motivation to scisec-irdirc@ejprarediseases.org before the 30th of April 2024.
Upcoming IRDiRC internal (closed) events:

- **In-person Consortium Assembly- Scientific Committees Meeting:** Location - Shanghai (China), on May 22-23, 2024 with the support of Hope for Rare Foundation, the Chinese Organization for Rare Disorders (CORD), and Fudan University.

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

Collaborative events:

- **2024 Global Rare Diseases Research Symposium & The Second China Rare Diseases Research and Translational Medicine Annual Conference:** Location - Shanghai, China, on May 23-26, 2024.

IRDiRC representation at events
Global Rare Diseases Research Symposium in Shanghai (China)

IRDiRC is co-hosting together with Hope for Rare Foundation and Fudan University, with National Children’s Medical Center/Children’s Hospital of Fudan University and Chinese Organization for Rare Disorders, the Global Rare Diseases Research Symposium & The Second China Rare Disease Research and Translational Medicine Annual Conference on May 23-25 in Shanghai (China).

- Where? Greenland Convention Center (Shanghai, China)
- When? May 23-25, 2024

The conference will last for 2.5 days with 21 parallel sessions and several satellite meetings. It will bring together over 100 speakers from global universities, hospitals, academic institutions, and pharmaceutical companies. Topics will cover basic research on rare diseases, gene and cell therapies, Investigator Initiated Trials (IIT), clinical studies, drug development, and international research collaborations. The conference will present the latest developments and original findings in rare diseases research, highlighting China’s contributions in the context of a global perspective.

Registration and More Information

Past events
IRDiRC at 2024 World Orphan Drug Congress USA

The World Orphan Drug Congress (WODC) USA (April 23-25) presented a golden opportunity to showcase the impactful work we’re doing and explore how collaborative efforts are shaping the future of rare disease research. Multiple IRDiRC members (David Pearce, PJ Brooks, Samuel Agyei Wiafe, Pamela Gavin, Alaa Hamed, Ramaiah Muthyala, Deborah Requesens, Durhane Wong-Rieger, Oxana Iliach) were present at the congress, which also featured a special panel on "Sustainability of N-of-1" and a "IRDiRC: A multi-stakeholder approach to understanding the needs in developing rare diseases diagnostics and therapies" session.

In addition, Ben Forred, from member organization Sanford Research, presented "Coordination of Rare Diseases at Sanford (CoRDS): Centralized international patient registry for all rare diseases". CoRDS is a a nonprofit research institution and the largest, free, international rare disease registry that connects patients, patient advocacy organizations and researchers.

Genomics of Rare Disease Conference
Dr. Dave Pearce, IRDiRC chair, attended the Genomics of Rare Disease Conference at the Wellcome Genome Campus on 23-25 March in Hinxton, England. This year's theme was focused on "Cutting-edge genomic science and rare disease clinical medicine". The conference included discussions on the use of a 'pangenome' to improve diagnostic yield, the role of common variants in rare disease, and new developments in prenatal and neonatal genomic screening, advances in therapeutics and common disease studies with implications for rare disease research.
In addition, Dr. Pearce presented at the 21st Orphan Drugs & Rare Diseases Conference in London, UK, in the session "International Collaboration in Research and Development in Rare Diseases" and participated in the panel discussion on "Strategies to promote research discoveries and development of orphan products to improve the health of people with rare disease". Have a look at the topics discussed here: https://www.orphandrugscongress.com/

IRDiRC Scientific Secretariat Coordinator, Daria Julkowska, attended the Rare Diseases Forum, a two-day conference held on 16-17 April in Brussels, Belgium. Against the backdrop of the Belgian EU Council Presidency, the forum brought together senior leaders from across patient organisations, industry and policy to address how and why innovation in tackling rare diseases can support the EU’s competitiveness agenda and strengthen the case for EU Action on Rare Diseases.

NEWS FROM IRDiRC MEMBERS

Foundation for Rare Diseases (France) - Open call for grants applications
The Foundation for Rare Diseases has launched a series of calls for proposals:
Call for projects with Association Strümpell-Lorrain/HSP France (ASL-HSP) – "Research on hereditary spastic paraplegia";

Joint call for proposals with French Friedreich’s Ataxia Association (AFAF) – "Research on Friedreich’s ataxia";

Call for projects – “Human and Social Sciences & Rare Diseases”;

Call for projects with Vaincre La Mucoviscidose – "Human and Social Sciences & Cystic Fibrosis”.

In addition, the Foundation for Rare Diseases, the Expertise Platform on Rare Diseases Bourgogne-Franche-Comté, the Health Network for Rare Diseases AnDDI-Rares and the centers of reference and resource are pleased to invite you to their scientific symposium "From research to care for rare diseases” to be held on Monday, April 29, 2024 at the Centre for Taste and Feeding Behaviour (CSGA) in Dijon (France). This event is open to all, and will highlight the unique regional ecosystem of research and care in the Bourgogne-Franche-Comté region.


More grant applications

FDA - New Grant Applications

The U.S. Food and Drug Administration (FDA) is holding review panels for its clinical trial grant applications received this Fiscal Year (FY). If studies are meritorious, funding will occur prior to September 30, 2024.

In addition, FDA is reviewing the natural history study grant applications submitted to RFA-FD-22-001. The FDA Office of Orphan Products Development (OOPD) next receipt date is May 6, 2024, for applications for the Rare Neurodegenerative Disease Grants Program.

The purpose of this funding opportunity announcement (FOA) is to support efficient natural history studies alone or in conjunction with the development and validation of clinical outcome assessments (COAs) and/or biomarker studies to address the unmet needs in rare neurodegenerative diseases for
children and adults. Through the support of studies with high quality and interpretable data elements, FDA expects to address critical knowledge gaps, remove major barriers to progress in the field, exert a significant and broad impact on a specific rare neurodegenerative disease or multiple rare neurodegenerative diseases with similar pathophysiology, and facilitate rare disease product development.

**CDMRP - Open Funding Opportunities**

The Department of Defense's (DoD's) Congressionally Directed Medical Research Programs (CDMRP) has a well-established process for managing the review and selection of funding applications that it receives for its medical research programs. New Research Funding Opportunities spanning the research spectrum from mechanistic research to clinical trials are now available in Rare Disease. All organizations, institutions and companies, both international and domestic, are encouraged to apply.

**Alexion AstraZeneca Rare Diseases Updates**

We are proud to share the kick-off of the Companies Constituent Committee member Alexion AstraZeneca Rare Diseases project RARE REV-inar Women in Rare science and research that marks the International Women's Day 2024, by hosting a webinar celebrating women in rare disease science and research:

Check out more details about this programme: [https://bit.ly/WomenInRareScienceAndResearch](https://bit.ly/WomenInRareScienceAndResearch)

**Neuromyelitis Optica Spectrum Disorder (NMOSD) Mental Health Study**

"My care team consists of over 30 healthcare professionals and I can’t recall even one time where one of them asked me about my mental health" (60 y.o. female patient).
Alexion is partnered with Thomas Jefferson University to assess and address mental health among NMOSD patients. Phase 1 of the partnership was a research study titled "The Psychological Burden of NMOSD", which has recently been published in PLOSOne after a successful abstract debut at the European Committee for Treatment and Research in Multiple Sclerosis and Americas Committee for Treatment and Research in Multiple Sclerosis (ECTRIMS-ACTRIMS) Meeting in Milan. This study is the first effort by Alexion to generate evidence on the unique mental health challenges of rare disease patients.

The spark for this study came from an Alexion workshop where colleagues listened to lived patient experiences. In this workshop, Alexion heard directly from patients that their mental health journey living with NMOSD was a very important moment that matters key research objectives in Phase 1 included understanding and characterizing the psychological burden of NMOSD patients and caregivers, developing a disease-specific scale to measure this burden, assessing the appropriateness of various mental health interventions, developing self-serve tools that fit the user needs of NMOSD patients and caregivers, and planning for developing and testing NMOSD telehealth-based intervention for mental health.

Findings from this phase reveal that patients with NMOSD and caregivers are experiencing markedly heightened anxiety, depression, medical trauma, mistrust in the medical system, perceived abandonment at the time of diagnosis, avoidance-based coping, and a sense of not living according to their values. All these psychological barriers can negatively impact how patients experience living with the disease and treatment adherence.

**Global Genes - RARE Drug Development Symposium**

The RARE Drug Development Symposium, hosted by Global Genes and the Orphan Disease Center, equips advocates with the knowledge, skills and connections they need to advance therapy.
development for their communities. This year’s theme is Innovative Ideas from Next Generation Change-Makers.

**When?** April 29 - May 1, 2024

**Where?** Sheraton Philadelphia Downtown, North 17th Street, Philadelphia, PA, USA

Don’t miss out on this unique opportunity to be a catalyst for change in the field of rare disease research. As advocates, the time is now to actively participate in the exploration of innovative therapies for rare diseases, your community, and loved ones.

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**LaunchR - A new network of rare disease experts**

LaunchR is a new network of Western Australian (WA), national, and international rare disease experts dedicated to developing, providing access to, and manufacturing of, precision medicines for rare disease. In addition, LaunchR stimulates the pathways to translate these to impact for common diseases. The network leverages WA’s expertise in rare disease precision medicine, particularly, but not only, in antisense oligonucleotides to pioneer personalised therapies for rare diseases.

LaunchR brings together initiatives for integrated pipelines for diagnosis, drug discovery, trials and treatment access, and new local (drug) manufacturing industry. As part of LaunchR’s initiatives, through the Rare Care Centre and a network of public-private partnerships, an expanding paediatric rare diseases clinical trials and treatment ecosystem, called TrialR, to administer these cutting-edge medicines, is in deployment. Additionally, LaunchR will prioritise equity by advancing access to treatments and clinical trials for underrepresented populations, including First Nations Australians, for example through Lyfe Languages (the Universal Indigenous Medical Translator), and for those living in remote regions, through teletrials partnerships.

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Read the Press Release
OTHER NEWS

The European Joint Programme on Rare Diseases Final Conference - Consolidating the rare disease research ecosystem

The final European Joint Programme on Rare Diseases (EJP RD) Conference is taking place from May 27th to May 28th in Bari (Italy), and is set to bring together leading experts, professionals, and enthusiasts from around the world. The registration deadline has been extended to May 22nd, 2024!

Join us online on May 27-28, 2024 for engaging sessions on rare disease research.

Check out the agenda here for a sneak peek into the sessions and plan your conference experience!

Learning from conect4children: A Collaborative Approach towards Standardisation of Disease-Specific Paediatric Research Data

Check out how public-private partnerships foster research in Rare Diseases! Connect4Children, funded by the Innovative Medicines Initiative 2 Joint Undertaking, a public–private partnership between the European Union and the European pharmaceutical industry, has just released a publication on the need for standardized disease-specific data and reviewed multiple initiatives, data resources, and data standards that could be utilised for this purpose. Have a look at their plan of action that can act as a steppingstone to the long-term standardisation goals.
Orphan Products (ECRD) organized by EURORDIS-Rare Diseases Europe will take place this year on May 15-16 in Brussels (Belgium). The ECRD is the largest, patient-led, rare disease policy-shaping event held in Europe.

The primary policy objective of ECRD 2024 is to ensure that rare diseases remain a top priority for the upcoming European political leadership, as the Conference sets a decisive platform to champion the integration of a comprehensive European Action Plan on rare diseases into the new EU legislative agenda, rooted in the recommendations of Rare 2030.

Check out the programme at a glance here: https://www.rare-diseases.eu/programme-7/