To consult the online version, click here

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

Get to know the Interdisciplinary Scientific Committee (ISC)!

Having a team of experts working on cross-cutting aspects of rare disease research, bridging the gaps between data sharing, ontologies, biobanking and registries have been some of the ongoing priorities of the **Interdisciplinary Scientific Committee (ISC)**. During the last years, the group has worked on guiding the **policies for privacy-preserving identifiers, interoperability, and model consent clauses for rare disease research**. A major milestone represented the investigation of **basket trials** to evaluate single treatment intervention for multiple diseases that share a common molecular alteration, largely described in the **"Targeting shared molecular etiologies to accelerate drug development for rare diseases"** paper [link: https://www.embopress.org/doi/full/10.15252/emmm.202217159].

Another important achievement for the group constituted the collaboration with the **Therapies Scientific Committee (TSC)** in creating a review of the **orphan devices used in rare diseases**, 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].

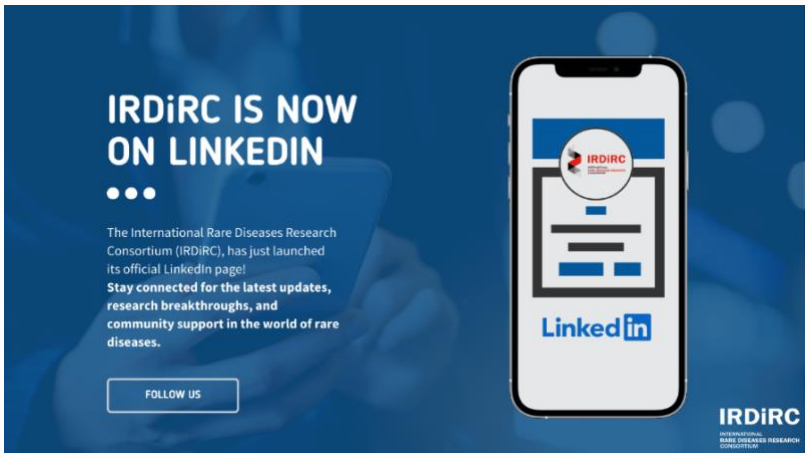
Setting up the group priorities for 2024, some of the topics planned to be further investigated include: **Basket trials for ultra rare bleeding disorders** (together with **ERN-EuroBloodNet** and the **European Haemophilia Consortium**, following the shared molecular etiologies example), **Drug-**

eluting devices for rare diseases (in partnership with **TSC**), and addressing **Stigma in Rare Diseases** (a key challenge to overcome to end the patient diagnostic, therapeutic and care odyssey). Increasingly, the ISC is focussing on key cross-cutting issues like **mental health and wellbeing, human rights, biomarkers** and **outcome measures**.

Four membership openings are foreseen for the Interdisciplinary Committee in the next months. (Stay tuned!) The ISC will be looking for those with bright minds and big hearts, thinkers and doers that are committed and action oriented.

More Information

IRDiRC has now a LinkedIn Page



We are delighted to announce that **IRDiRC has officially launched its LinkedIn page** where we will provide timely and relevant updates on the work of IRDiRC and its members.

Subscribe here: https://www.linkedin.com/company/international-rare-diseases-research-consortium-irdirc/about/

Advancing diagnosis and research for rare genetic diseases in Indigenous peoples

IRDiRC proudly presents the second paper developed by the members of the **Indigenous Population Task Force** now available in **Nature Genetics, "Advancing diagnosis and research for rare genetic diseases in Indigenous peoples".** The commentary focuses on the importance of achieving a diagnosis for Indigenous people living with a rare, often genetic disease, which is crucial for equitable healthcare.

Read the publication

New Members of the Consortium Assembly

IRDiRC is delighted to present its new members of the **Consortium Assembly**:

Companies Constituent Committee (CCC)

- **Alexion AstraZeneca Rare Disease**, represented by **Wendy Erler**, VP Patient Experience (based in USA)
- **ITALFARMACO SpA**, represented by **Samantha Parker**, Patient Advocacy and Communication Lead Rare Diseases Europe (based in France)

Funders Constituent Committee (FCC)

- **Congressionally Directed Medical Research Programs (part of USAMRDC)**, represented by **Kristie Lidie**, Deputy Director for Program Management (based in USA)

Patient Advocacy Constituent Committee (PACC)

- **Federación Peruana de Enfermedades Raras - FEPER**, represented by **Pilar Estremadoyro Reyes**, Vice President (based in Peru)

IRDiRC wishes a very warm welcome to its new members!

Change of representation

IRDiRC announces the change in representation for **Takeda** in the **Companies Constituent Committee (CCC)**. **Laura Rosen**, VP & Head, Translational Clinical Sciences - Neuroscience (USA) is replacing **Ceri Davies**, former Takeda representative in the Consortium.

Welcome aboard, Laura!

IRDiRC EVENTS

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

- **Online Consortium Assembly Meeting**: on **March 6-7, 2024**;
- **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

Upcoming events

2024 World Orphan Drug Congress USA

As part of our commitment in advancing initiatives related to rare diseases, **IRDiRC** is a proud partner at the **World Orphan Drug Congress USA**.

This will be a defining event for the rare disease and orphan drug space globally, bringing together top key opinion leaders from around the globe.

The upcoming congress will cover priority topics in the rare disease environment, including **next generation therapies, clinical development and regulatory, global market & patient access, advanced therapies, rare disease advocacy**, and many more! Among the confirmed speakers at the congress are **David Pearce**, IRDiRC chair, and **PJ Brooks** (FCC member, NIH/NCATS).

- **Where? Boston Convention & Exhibition Center (MA, USA)**
- **When? April 23-25**

IRDiRC is happy to share a **50% discount code** to its network for registering for the USA edition: **IRDIRC50**.

More details on the agenda and speakers coming soon.

Registration and More Information

#iDR24 - International Drug Repurposing Conference

IRDiRC members **Anneliene Jonker**, **Daniel O'Connor** and **Marjon Paasmoij** (Therapies Scientific Committee) will be speakers at the the first international drug repurposing conference, **#iDR24** (co-organised by **REMEDI4ALL**, **Beacon** and **MeRIT** on **6-7 March** in Barcelona, Spain) in the Opening plenary - **"The challenge of translation"**, in the Methodologies session **"Mind the gap: What are the blockers and how can we smoothen the transition from discovery to clinic?"** and in the **Rare diseases path** **"The evolving rare disease landscape: past, present, and future".**

Registration and More Information

POLITICO LIVE: Rare Diseases and the battle to define unmet medical needs

A roundtable of EU and national policymakers, industry experts and patients will gather together to discuss the best approach to define unmet medical needs and Belgium's multi-criteria approach.

The POLITICO event will convene EU and national policymakers, industry experts and patients to discuss what is the best approach to define unmet medical needs and Belgium's multi-criteria approach. Among the confirmed speakers are also the IRDiRC **Interdisciplinary Scientific Committee** Vice Chair, **Marc Dooms** (University Hospitals Leuven, Belgium) and **Regulatory Scientific Committee**Vice Chair, **Violeta Stoyanova-Beninska** (European Medicines Agency, the Netherlands).

When? **Wednesday, March 6, 2024 at 3:30 - 6:00 PM CET**

More Information

Past events

RARE-X 2024 Conference

Dr. David Pearce, IRDiRC Chair, represented the International Rare Diseases Research Consortium at the **RARE-X 2024 Conference** held in **Johannesburg, South Africa**, where he presented on the organization and its overall goals and initiatives.

More Details

NEWS FROM IRDiRC MEMBERS

FDA presents 2024 Grant Proposals

The U.S. Food and Drug Administration (FDA) has two upcoming grant receipt dates, for the following topics:

1. Efficient and Innovative Natural History Studies Addressing Unmet Needs in Rare Diseases

More information: https://grants.nih.gov/grants/guide/rfa-files/RFA-FD-22-001.html

2. Natural History and Biomarker Studies of Rare Neurodegenerative Diseases

More information: https://grants.nih.gov/grants/guide/rfa-files/RFA-FD-24-024.html

In addition, FDA awarded **10 new clinical trial studies** through the **Orphan Products Grants Program** and **6 new studies to support research for rare neurodegenerative diseases**.

More grant applications

Scientific Calls for Proposals AMED 2024

The Japan Agency for Medical Research and Development (AMED) announced the opening of the calls for proposals in 2024.

Among the topics, are present:

- **Evidence generation** (adult, paediatric, ultra-rare, Software as a Medical Device);
- **Registry information utilisation research;**
- **Undiagnosed Diseases Initiative (IRUD).**

Apply here

Foundation for Rare Diseases announces the launch of Calls for Proposals in 2024

The Foundation for Rare Diseases (France) announces the calls for proposals in 2024:

- "**GenOmics**" - a next generation sequencing to investigate genetic and molecular bases of rare diseases;
- **Alnylam Pharmaceuticals Price 2024** - RNAi and rare diseases.

Additional details about the deadlines and requirements in the link below.

More Information

Join the 12th European Conference on Rare Diseases and Orphan Products in Brussels!

The 12th edition of the European Conference on Rare Diseases and 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://download2.eurordis.org/ecrd/2024/Programme_at_a_glance.pdf
- **Registration** categories and fees: https://www.rare-diseases.eu/register/

More Details

IRDiRC Chair, David Pearce, is elected in the Board of Undiagnosed Diseases Network International

IRDiRC is excited to share the fantastic news that IRDiRC Chair, **David Pearce** (Sanford Research, USA), was elected in the **Undiagnosed Diseases Network International (UDNI) Board**.

UDNI is an international rare disease network, modelled after the **National Institutes of Health's (NIH) Undiagnosed Disease Program (UDP)** (established in 2008), to tackle undiagnosed diseases as a global health issue, calling for an international scientific and healthcare effort, fostering also a strong patient involvement through the **Patient Advisory Group**.

More Information

Orphan medicines development - Ask the European regulator

Don't miss out the **European Medicines Agency's Orphan Medicines Office** interactive webinar organized on the **Rare Disease Day**, 29 February, to answer questions raised by the developers of products for rare diseases or the researchers in this field might have on orphan designations and rare disease development.

Among the panellists at the webinar will be the **Regulatory Scientific Committee Vice Chair**, **Dr. Violeta Stoyanova-Beninska** (EMA, the Netherlands), who will share additional details on the

background of orphan designation and the benefits it has brought to patients. The webinar will include a live question-and-answer session enabling participants to ask their questions on orphan medicines development.

More information: https://www.ema.europa.eu/en/events/orphan-medicines-development-ask-european-regulator

Register here

SANOFI announces two recent partnerships to generate Real-World Evidence

The International Niemann-Pick Disease Registry (INPDR) announced a new collaboration giving **Sanofi**controlled access to anonymized registry information via INPDR's research "Gateway" platform. This collaboration will provide Sanofi with Real-World Evidence that will support evaluation and decision making on the use of **olipudase alfa** to manage **Acid Sphingomyelinase Deficiency (ASMD)**.

INPDR is the largest active database about the diagnosis, management, and progression of the inherited lysosomal storage disorders known as Acid Sphingomyelinase Deficiency and Niemann-Pick type C Disease (NPC).

Read the press release: https://inpdr.org/wp-content/uploads/2024/02/INPDR-Sanofi-Press-Release-V5.pdf

Launch of Together4RD's first pilot project

Together4RD's groundbreaking initiative has taken off with the official launch of its first pilot project! Proudly spearheaded by **Sanofi** and the **European Reference Network on Rare Bone Diseases (ERN BOND)**, the collaborative efforts are committed to gaining a deeper understanding of the **Osteogenesis Imperfecta burden**, leveraging the wealth of data already gathered to also develop innovative outcomes measures.

More Information

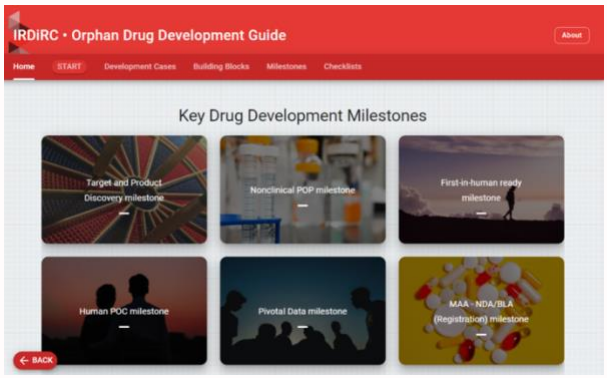
ORDI - Annual Awareness Run "RACEFOR7"

The Organization for Rare Diseases India (ORDI)coordinated a new edition of the major initiative, **Annual Awareness Run "RACEFOR7"** - A run of 7 kilometres symbolically representing the 7000 rare diseases with an estimated 70 million affected people. RACEFOR7 is a successful annual awareness run since 2016 in Bengaluru & has become a PAN India Event.

RACEFOR7 helps to raise Awareness & recognize the challenges faced by people with rare diseases, by driving public policy, accelerating research and improving care for people living with rare diseases.

The 9th edition of Racefor7 was held on 25th February (the last Sunday of February) across multiple towns in India.

More Information



New Tutorial for the Orphan Drug Development Guidebook

Check out the new tutorial, now available, for the **Orphan Drug Development Guidebook!** The ODDG is a patient focused guide that describes the available tools, incentives, resources and practices for developing traditional and innovative drugs or therapies for rare diseases indications and how to best use them.

The platform can be used by various stakeholders including: academics, NGOs, small/large biotechs and drug developers.

More Information

OTHER NEWS

Save the date for the EJP RD Final Conference in Bari!

The **final European Joint Programme on Rare Diseases (EJP RD) Conference** is scheduled to take 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.

Note: *the event is hybrid, however it is open for in person participation only by invitation.*

The registration deadline for online participation for the general public is **March 31, 2024**. Link: https://forms.office.com/pages/responsepage.aspx?

More Information

X-OMICS Festival - "The future of multi-omics research is now!"

On **April 15th 2024** the **6th edition of the X-omics Festival** will take place in **Nijmegen, the Netherlands**. The program of this edition will contain different types of multi-omics sessions with a keynote, presentations, poster pitches sessions and opportunities to network with the X-omics community and other (multi-)omics researchers.

Registration is free of charge (but is required): https://www.aanmelder.nl/x-omicsfestival2024

More Information

EJP RD-EFPIA Joint Advanced Webinar on Real-World Data, Machine Learning, and Deep Analytics

We are delighted to announce the successful outcome of the **EJP RD-EFPIA Joint Advanced Webinar on Real-World Data, Machine Learning, and Deep Analytics in rare diseases**, which accumulated enthusiastic participation from approximately **300 attendees**.

For those unable to partake in the live event, we are pleased to inform you that the recorded sessions, featuring keynote speaker presentations and insightful discussions, are now accessible.

Watch the Recording

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.