IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

Register for the Second International Conference on Clinical Research Networks for Rare Diseases

With just two weeks to go, registrations are still open for the **2nd International Conference on Clinical Research Networks**, a two-day hybrid meeting focused on aligning global efforts in **data, diagnostics, and clinical trials for rare diseases**. Co-organised by the **European Rare Diseases Research Alliance (ERDERA)**, **IRDiRC**, and **Rare Diseases International**, the event will take place in Heidelberg (Germany) and online on 9-10 December, 2025.

IRDiRC Chair, **Dr. David Pearce** will set the scene and open the conference on the 9th of December, together with **Alexandra Heumber Perry** (Chief Executive Officer, Rare Diseases International) and **Baptiste Eluard** (Senior Scientific Project Manager, ERDERA). IRDiRC Vice Chair, **Samantha Parker** will lead the workshop on **"Rules of engagement in multi-stakeholder pre-competitive**

environments" (10 November, 9h00 CET), while Dr. Pearce will chair the plenary session on **"Global Networks and Models of Care in high-income and LMIC - The Duchenne experience"** (10 November, 15h30 CET).

Sessions will explore key themes including regulatory-ready real-world evidence, multinational data initiatives, and emerging diagnostic approaches.

Registration remains open just for virtual attendance until 5th December.

Full Programme & Registration

New Leadership at the Funders Constituent Committee

We are pleased to announce that **Stefano Benvenuti**, Head of Public Affairs at Fondazione Telethon, based in Italy, and **Naveed Aziz**, Vice President of Research and Innovation at Genome Canada, based in Canada, have been elected as the Chair, respectively, Vice Chair, of the **Funders Constituent Committee (FCC)**.

The IRDiRC Funders Constituent Committee's principal mission is to ensure high-level coordination of funding initiatives in order to maximize the impact of rare disease research. This is achieved by aligning strategic priorities, preventing unnecessary duplication, and systematically identifying gaps in research funding.

We are sending our sincere congratulations to the new chairs and we are looking forward to working with them.

More Information

New Leadership at the Therapies Scientific Committee

IRDiRC is delighted to announce the appointment of **Shekhar Natarajan** as the Chair of the Therapies Scientific Committee (TSC) and of **Diana Kwast** as Vice Chair.

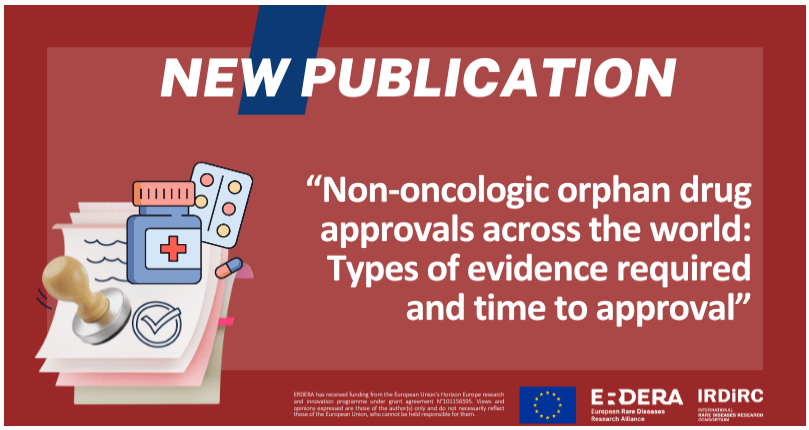
Shakar Natarajan and Diana Kwast bring complementary leadership to IRDiRC's Therapies Scientific Committee (TSC). As Chair, Shakar draws on his deep regulatory experience at Dyne Therapeutics to steer the committee's mission of accelerating rare-disease therapy development. As Vice Chair, Diana contributes a strong patient-led perspective: she is Executive Director of the (Un)limited Forward! foundation, an EUPATI and EUCAPA Fellow, and a PhD candidate, combining advocacy, academic insight, and lived experience. Together, they will guide the TSC in advancing IRDiRC's goal of delivering 1,000 new treatments for rare diseases by 2027.

We send our warmest welcome to our new TSC leaders!

More Information

IRDiRC PUBLICATIONS

Non-oncologic orphan drug approvals across the world: Types of evidence required and time to approval



IRDiRC is pleased to announce the release of a new publication developed by the **Regulatory Science Committee (RSC)**: **"Non-oncologic orphan drug approvals across the world: Types of evidence required and time to approval."**

This analysis examines 53 orphan medicines approved between 2021–2022 across six major regulatory regions. The findings highlight persistent delays in global access to innovative therapies for people living with rare diseases, despite substantial alignment in regulatory evidence requirements.

Key Insights

- Significant approval delays: An average 3-year gap between the first and subsequent regulatory approvals.
- Limited global submissions: Although 99% of products were approved in two or more regions, few were submitted across all major jurisdictions.
- Aligned evidence requirements: 69% of approvals relied on identical or highly similar evidence packages.
- Streamlined data needs: Most medicines were authorized on the basis of one adequate and well-controlled study, supported by confirmatory evidence.
- Opportunity for acceleration: Results underscore the importance of collaborative regulatory pathways to shorten global approval timelines.

With more than 300 million people worldwide affected by rare diseases, ensuring timely access to effective therapies remains crucial. These findings highlight the need for strengthened international coordination to reduce delays and support equitable access to treatment across regions.

Read the Publication

What matters ethically about how the UDN has changed since its inception?

For individuals living with undiagnosed rare diseases, the journey toward a diagnosis is often long, uncertain, and emotionally taxing. A new IRDiRC publication takes a deep look at the **Undiagnosed Diseases Networks (UDNs)**, how they have evolved over time, and the ethical dimensions that accompany these changes.

The article highlights the importance of international collaboration, thoughtful data sharing, and the development of diagnostic best practices. Together, these efforts can help expand access, support equity, and ultimately improve outcomes for patients and families worldwide.

Dr. Dave Pearce joined Ethics Talk to discuss the publication's insights and implications for the rare disease community. Listen to the interview: https://journalofethics.ama-assn.org/podcast/author-interview-what-matters-ethically-about-how-udn-has-changed-its-inception

Read the Publication

Regulatory sandboxes: A new frontier for rare disease therapies

Regulatory sandboxes are emerging as a promising tool to support the development and approval of innovative technologies, including therapies for rare diseases. By creating a controlled yet flexible environment for testing new regulatory approaches, sandboxes can help accelerate the development and accessibility of life-saving treatments that often fall outside traditional regulatory pathways.

With more than 400 million people worldwide affected by rare diseases, about half of them children, the need for faster, more adaptable solutions is urgent. Diagnosis and treatment are frequently delayed due to limited data, variable disease presentation, scarce expertise, and the high cost and complexity of drug development. These challenges highlight the need for more collaborative, interdisciplinary, and responsive regulatory mechanisms.

As global regulators explore new models to keep pace with scientific progress, regulatory sandboxes offer a pathway toward greater harmonization and cross-border cooperation. This article outlines their purpose, key features, current initiatives, and potential impact on improving rare disease therapy development and approval.

Read the Publication

Applying the international rare disease research consortium (IRDiRC) N-of-1 therapy task force eligibility criteria for individualised therapies use case: Duchenne muscular dystrophy

IRDiRC announces publication of a new manuscript, developed as part of N-of-1 Task Force: **"Applying the IRDiRC N-of-1 Therapy Task Force Eligibility Criteria for Individualised Therapies – Use Case: Duchenne Muscular Dystrophy"**.

Nucleic acid-based therapies, including antisense oligonucleotides (ASOs), are opening new doors for the treatment of rare diseases. With over 30 individuals currently receiving N-of-1 ASO therapies, there is increasing interest in expanding these personalised approaches. However, the complexity of these treatments calls for a clear and ethical framework.

This publication tests the eligibility roadmap developed by the IRDiRC Task Force by applying it to a real-world use case: Duchenne muscular dystrophy (DMD).

Read the Publication

IRDiRC EVENTS

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

- **Online Consortium Assembly Meeting** on 15 December, 2025.
- **In-person Consortium Assembly and Scientific Committee Meeting** in Sofia, Bulgaria on 18-19 March, 2026.

Collaborative events:

- **The 2nd International Clinical Research Networks Conference,** co-organized with Rare Diseases International & ERDERA: Location - Heidelberg, Germany, on 9-10 December, 2025**.**
- **The European Conference on Rare Diseases & Orphan Products (ECRD)** in Prague, Czech Republic on 3-4 June 2026, as an **associated partner** of the event. More information about the event: https://www.rare-diseases.eu/
- **World Orphan Drug Congress (WODC) USA** in Boston, MA, USA, on 9-11 June, 2026. More information about the event: https://www.terrapinn.com/conference/world-orphan-drug-congress-usa/index.stm

**Photo taken at the Preventive Medicines Task Force workshop in Paris, France (October 2025).*

IRDiRC REPRESENTATION AT EVENTS

UPCOMING EVENTS

ECRD 2026 – IRDiRC Proud to Participate as an Associated Partner

IRDiRC is delighted to announce its participation as an Associated Partner in the next **European Conference on Rare Diseases & Orphan Products (ECRD)**, taking place 3–4 June 2026 in Prague. The 2026 edition will be held under the theme:

"Rare Diseases in a Changing & Competitive Europe: Shaping policies to address the unmet needs of people living with rare diseases."

ECRD is Europe's largest patient-led, policy-shaping event for the rare disease community. By bringing together people living with a rare disease, patient advocates, policymakers, clinicians, regulators, industry representatives, and Member State officials, the conference creates a powerful platform for advancing policies that improve the lives of those affected by rare diseases.

As an associated partner, IRDiRC looks forward to contributing to this important milestone, reinforcing our shared commitment to accelerating rare disease research and improving outcomes for patients worldwide.

Mark your calendar for 3–4 June 2026 and join us in shaping the future of rare disease policies in Europe.

More information

IRDiRC at World Orphan Drug Congress USA 2026

IRDiRC is pleased to announce its participation in the upcoming **World Orphan Drug Congress (WODC) USA**, taking place 10-11 June, 2026 at the Boston Convention & Exhibition Center in Boston, MA.

WODC USA is one of the premier global gatherings for the rare disease and orphan drug community, bringing together researchers, industry leaders, patient advocates, regulators, and policymakers to advance innovation and improve outcomes for people living with rare diseases.

Programme Highlights

- Keynote Panel: ***Sustainability & Access for Innovative Therapies for Rare Diseases***

Moderator: Dr. David Pearce (IRDiRC Chair, USA)

Representatives will include experts from the National Institutes of Health (NIH), the Food and Drug Administration (FDA), the Centers for Medicare & Medicaid Services (CMS), as well as a parent advocate who will bring the perspective of families affected by rare diseases.

- Featured Presentations:

(1) Bridging Diagnostics to Therapy and Care

Presenter: Lisa Emrick, Baylor College of Medicine (USA)

(2) Preventative Medicine Strategies for Rare Diseases

Presenter: Jon Brudvig, Amicus Therapeutics (USA)

(3) Fireside Chat: Where Is N-of-1 Going?

Moderator: Dr. David Pearce (IRDiRC Chair, USA)

Panelists: Sarah Glass (n-lorem Foundation, USA) and Timothy Yu (Boston Children's Hospital, USA)

And more to be confirmed!

More information

PAST EVENTS

Global Highlights in Rare & Undiagnosed Diseases

We are proud to share that **Dr. David Pearce**, the Chair of IRDiRC, participated in the **14th International Conference on Rare and Undiagnosed Diseases (UDNI)** in Rio de Janeiro (Brazil) on 28-31 October, contributing to international efforts to advance diagnosis, care, and collaboration for individuals with undiagnosed conditions. More information about UDNI: https://www.udninternational.org/

In addition, Dr. Pearce joined the **17th International Collaboration on Rare Diseases (ICORD)**, which took place on 14-16 November, in Izmir, Turkey. ICORD brings together global leaders working to improve outcomes for people living with rare diseases through research, policy, and patient-centered innovation. Dr. Pearce delivered a keynote address, ***"A global approach to rare and undiagnosed diseases,"*** and also served as Session Chair for Patient Centered Care and Advocacy. More information about ICORD: https://www.icord2025.com/

IRDiRC at ERDERA's General Assembly

The International Rare Diseases Research Consortium, represented by Scientific Project Manager **Alexandra Tataru**, participated in **ERDERA's 2nd General Assembly Meeting** held in Amsterdam, Netherlands, from 29–31 October 2025. The event offered a valuable opportunity to connect with European and international partners in rare disease research and to learn about the latest initiatives and strategic developments presented by ERDERA.

More information

IRDiRC at WODC Europe 2025

IRDiRC demonstrated a strong and visible participation at this year's World Orphan Drug Congress (WODC) Europe in Amsterdam, underscoring its central role in shaping global rare disease research and policy. Across the 3 days of congress, IRDiRC leaders contributed with their expertise to high-level discussions addressing research, regulation, public–private collaboration, and future therapeutic landscapes. Their involvement reflected both the breadth of IRDiRC's scientific and policy leadership and its commitment to driving forward solutions for the rare disease community.

The programme commenced on Monday, 27 October, with an important pre-congress workshop, ***Rare Disease Private Public Partnerships*** organised by Sanofi's representative **Vinciane Pirard**, Chair of

the IRDiRC Companies Constituent Committee (CCC), who contributed to IRDiRC's perspective on strengthening multi-stakeholder collaboration to accelerate innovation.

On Tuesday, 28 October, IRDiRC's presence was prominent across multiple scientific tracks. The day opened with Working Group 6 – ***Competition of regulatory ecosystems in approving medicines: policy implications in the case of Europe***, featuring **Violeta Stoyanova-Beninska**, Senior Scientific Specialist at the European Medicines Agency (EMA) and Chair of the IRDiRC Regulatory Scientific Committee (RSC). Later sessions included **Fondazione Telethon** ***not-for-profit model, from research to distribution of ATMPs for ultra-rare conditions*** by **Stefano Benvenuti**, Chair of the IRDiRC Funders Constituent Committee (FCC); ***Orphan Drugs Development: Productivity and Probability of Success*** by **Samantha Parker**, Vice Chair of IRDiRC; and the ***IRDiRC Task Force on Prevention: Opportunities and Challenges in the shift from treatment to prevention*** delivered by **Dan O'Connor,** former Chair of the IRDiRC Therapies Scientific Committee (TSC). The day concluded with ***Engagement of young people in rare diseases research: a roadmap for action presented*** by **Anneliene Jonker**, former Vice Chair of the IRDiRC Therapies Scientific Committee (TSC).

IRDiRC's contributions culminated on Wednesday, 29 October, during the high-profile ***Keynote Panel: New treatments on the block – 2025's rare disease pipeline***. This session explored emerging therapeutics, policy evolution, and sustainability challenges in rare disease drug development. IRDiRC was represented by two members: **Dan O'Connor** and **Violeta Stoyanova-Beninska**. Their joint participation reinforced IRDiRC's voice in guiding the future landscape of rare disease therapies.

NEWS FROM IRDiRC MEMBERS

EURORDIS: Registrations for the Black Pearl Awards

Held each February to coincide with Rare Disease Day, the annual **EURORDIS Black Pearl Awards** honour the outstanding achievements of people living with a rare disease and those who work tirelessly on their behalf, celebrating hard work, innovation, and dedication across the rare disease community. Organized since 2012 by EURORDIS–Rare Diseases

Europe, the event recognises the significant contributions of patient advocates, organisations, policymakers, scientists, companies, and others driving progress in the field. Since February 2023, it has evolved into a fully hybrid experience, enabling participants to join the ceremony from anywhere in the world, creating a unique evening of celebration, learning, shared experiences, and artistic performances.

Registrations for the **15th annual EURORDIS Black Pearl Awards** are now open. The event will take place in Brussels (Belgium) and online on 24 February 2026.

Don't miss this inspiring evening of live performances, celebrating stories of courage, collaboration, and community!

Registration

Other Publications Co-Authored by IRDiRC Members

New Publication by Dr. Gareth Baynam: Functional characterization of the MED12 p.Arg1138Trp variant in females: implications for neural development and disease mechanism

IRDiRC is pleased to highlight that **Gareth Baynam**, Chair of Interdisciplinary Scientific Committee, has co-authored a newly published study, **"Functional characterization of the MED12 p.Arg1138Trp variant in females: implications for neural development and disease mechanism"**. This manuscript provides the first functional evidence linking the rare MED12 p.Arg1138Trp variant to disrupted neural development in females with congenital anomalies and developmental delay. Using patient-derived iPSCs engineered to express either the wildtype or variant MED12 gene, researchers identified significant gene expression changes affecting RNA polymerase II regulation, transcription, pre-mRNA processing, and neural development, alongside reduced MED12L expression. Pathway analyses revealed delayed axon growth, impaired forebrain differentiation, altered neural cell specification, and strong upregulation of pre-ribosome complex pathways. Together, these findings demonstrate how the MED12 p.Arg1138Trp variant affects neural cell biology and contribute new mechanistic insight into MED12-related disorders.

Read the Publication

New Publication by Prof. Sally Ann Lynch: What is risk in clinical genetics? Designing and piloting tools to evaluate risk in clinical genetics using failure modes and effects analysis

Prof. Sally Ann Lynch, Chair of Diagnostics Scientific Committee, was senior author on a new study introducing the first systematic risk assessment tools tailored for Clinical Genetics, addressing a longstanding gap in how risk is monitored and managed across services. Using a retrospective review of 115 adverse events and near misses in Ireland, researchers developed a process map, audit tool, and adapted severity scoring system aligned with Failure Modes and Effect Analysis methodology. These tools were piloted across five European clinical genetics centres, revealing that adverse events occurred in more than 3% of appointments in most sites, with common issues involving consent, sample processing, and patient communication. Re-audits in 2024 showed improvements following targeted interventions, underscoring the utility of these tools. The study demonstrates how standardized monitoring can enhance risk reduction and support higher-quality, safer clinical genetics services.

Read the Publication

New Publication by Dr. David Pearce & Dr. Gareth Baynam: Digital health technology use in clinical trials for rare diseases (a systematic review)

Dr. Pearce and **Dr. Baynam** co-authored a new publication on digital health technology use. Rare disease trials often face low enrollment and limited retention, but digital health technologies (DHTs) are helping bridge these gaps. An analysis of 262 studies across the ten most-studied rare diseases shows rising adoption of DHTs for recruitment, monitoring, treatment, and follow-up. Data monitoring and collection was most common (31.3%), while digital treatment appeared in 21.8% of studies. From 2021–2024, nearly all diseases saw increased digital integration, with cystic fibrosis leading at 29.7%.

These findings highlight a growing shift toward decentralized, patient-centric research models that reduce burden and broaden access. DHTs enable more continuous data collection, flexible participation, and stronger patient engagement. To guide future progress, researchers propose the "4A" framework: Accessibility, Agility, Awareness, and Adaptability. Together, these principles point to a more scalable and equitable future for rare disease care and therapeutic development.

Read the Publication

New Publication by Tudor Groza, Gareth Baynam and Saumya Jamuar: Information content as a health system screening tool for rare diseases

IRDiRC Interdisciplinary Scientific Committee member and Chair**, Tudor Groza** and **Gareth Baynam,** together with **Saumya Jamuar**, Vice Chair of Diagnostics Scientific Committee, co-authored a new publication on information content as a new health system screening tool applied for rare diseases.

Rare diseases pose major challenges for patients and health systems, with individuals often waiting years for a diagnosis due to fragmented records, inconsistent coding, and limited specialist access. Because many rare conditions lack clear ICD identifiers, undiagnosed patients frequently remain invisible in electronic health records. SNOMED-CT, with its richer clinical detail and strong coverage of rare disease concepts, offers a promising alternative, especially when paired with advanced analytics. This study explores the use of information content (IC) and entropy-based methods to detect atypical clinical patterns in a dataset of 1.2 million Singaporean patients. Findings show that IC can differentiate rare disease profiles from the first encounter, offering a scalable, high-sensitivity approach to identifying potential rare disease patients earlier in their care journey.

Read the Publication

OTHER NEWS

ERDERA 2026 Joint Transnational Call

On 10 December 2025, ERDERA will launch its **2026 Joint Transnational Call**, titled: **"Resolving unsolved cases in rare genetic and non-genetic diseases".** This call will invite multinational research teams to submit proposals aimed at improving diagnostic clarity for patients with rare genetic and non-genetic conditions that currently remain unsolved. The initiative seeks innovative, collaborative approaches that can move the field closer to timely and accurate diagnoses for underserved patient groups.

Information Webinar

Researchers interested in applying are encouraged to join the dedicated webinar:

- Date: 16 December 2025
- Time: 15:00–17:00 CET

Registration: https://erdera.org/event/information-webinar-erdera-joint-transnational-call-2026/

The session will cover the call's objectives, application procedures, and opportunities for international collaboration.

More information

CONTACT & SOCIAL MEDIA

LinkedIn: International Rare Diseases Research Consortium (IRDiRC)

X (Twitter): IRDiRC

Website: https://irdirc.org/

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