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IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

Membership Call - Therapies Scientific Committee

IRDiRC brings together global stakeholders across research, diagnostics, therapies, funding and patient advocacy to accelerate progress for people living with rare diseases.

The Therapies Scientific Committee (TSC) is an international expert committee that helps guide priorities in rare disease therapy development. Through its Task Forces and Working Groups and in close

collaboration with the other IRDiRC Committees, the TSC contributes to the development of new ideas and collaborative initiatives across the rare disease field.
Current recruitment focuses on strengthening the committee's expertise in scientific, technical and regulatory priority areas.

We are looking for candidates with a proven track record of professional experience in the field of rare diseases and demonstrated **expertise in one or more of the following areas**:

- Simulation and modelling, including innovative trial design for small populations;
- Digital health and data systems;
- Regulatory science and global regulatory frameworks;
- Strong understanding of international research, development and regulatory landscapes, including experience working across jurisdictions;
- Experience working in academia, industry, regulatory agencies or other relevant organisations involved in therapy development;
- Experience contributing to research, innovation or policy in the rare disease field.

Key competencies and requirements

- Ability to work effectively in multidisciplinary and international environments and to collaborate constructively across sectors and cultures;
- Good command of English in both spoken and written contexts;
- Motivation and ability to contribute to the strategic goals and ongoing work of the TSC;
- Availability and willingness to actively participate in committee activities, including regular monthly online meetings, and contributions beyond scheduled meetings.

Commitment and expectations

Membership of the TSC is a voluntary role within an active and collaborative international group.
Members are expected to:

- Participate in monthly online meetings, including preparation, and contribute to ongoing committee activities and outputs;
- Attend one annual in-person meeting.

The appointment is for a three-year term, with the possibility of a one-time extension.

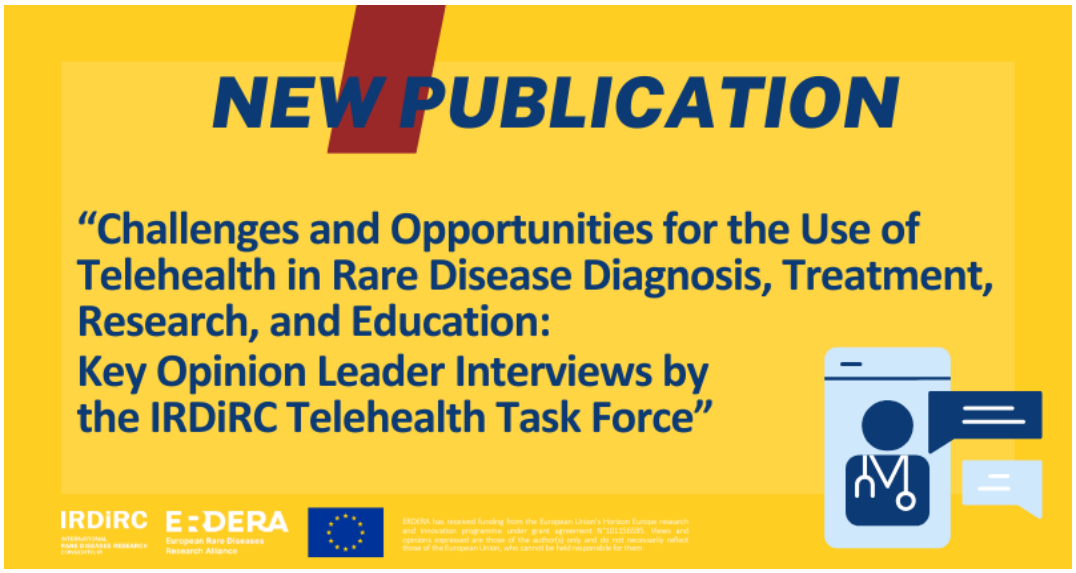
Application process

Interested candidates are invited to submit their curriculum vitae, a short biosketch and a letter of motivation. Applications should be sent to scientific.secretariat@irdirc.org by **11 May 2026**.

More about TSC

IRDiRC PUBLICATIONS

Challenges and opportunities for the use of telehealth in rare disease diagnosis, treatment, research, and education: key opinion leader interviews



A new IRDiRC publication, **"Challenges and opportunities for the use of telehealth in rare disease diagnosis, treatment, research, and education: key opinion leader interviews"**, was produced by the **IRDiRC's Enabling and Enhancing Telehealth for Rare Diseases Across the Globe Task Force**, which explores how **telehealth (TH)** can transform diagnosis, care, research, and education for rare diseases (RDs) worldwide.

Through interviews with 23 key opinion leaders (KOLs) across Europe, the United States, Sub-Saharan Africa, and Asia, we gathered diverse perspectives spanning clinicians, researchers, patient advocates, regulatory experts, and families.

Highlights:

• Telehealth has strong potential to improve access to timely and accurate diagnosis for rare disease patients;

• Remote care models can enhance treatment and long-term management, especially for underserved populations;

• Telehealth enables more inclusive and efficient rare disease research through remote assessments;

• Innovative virtual education and mentoring models can expand expertise among healthcare providers globally.

While barriers to large-scale implementation remain, there is strong consensus: telehealth is a powerful tool to bridge gaps in access, expertise, and equity in rare disease care. This paper synthesizes these insights and highlights critical evidence and knowledge gaps that future research must address to fully unlock telehealth's potential.

Read the Publication

Translating multi-omics into healthcare: requisites for scalable and equitable implementation

We are pleased to announce a new publication produced by the IRDiRC **Functional Analysis Task Force, "Translating multi-omics into healthcare: requisites for scalable and equitable implementation"**. This work explores the transformative potential of multi-omics in rare disease research and precision medicine. By integrating diverse omics datasets with advanced computational methodologies, it reveals deeper molecular insights. The paper highlights improvements in diagnostics, disease prevention, and personalized treatment strategies.

This work emphasizes multidisciplinary collaboration across researchers, clinicians, policy makers, and patient representatives.

Read the Publication

IRDiRC EVENTS

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

- **Online Consortium Assembly Meeting on** 17 June, 2026.

Collaborative events:

- **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
- **World Orphan Drug Congress (WODC) Europe** in Amsterdam, Netherlands, on 26-27 October, 2026. More information about the event: https://www.terrapinn.com/conference/world-orphan-drug-congress/index.stm

**Photo taken at the Consortium Assembly and Scientific Committees Meeting in Sofia, Bulgaria (March, 2026).*

IRDiRC REPRESENTATION AT EVENTS

UPCOMING EVENTS

RNDF Spring Assembly

Dr. David Pearce, IRDiRC Chair, will serve on the Scientific Advisory Board for the Rare Neurological Disorder Foundation (RNDF). The inaugural event will take place on 2 May 2026, at the Mayborn Museum Complex, Baylor University (USA).

Founded in 2021, RNDF is an emerging education and advocacy organization dedicated to improving understanding and support for individuals and families affected by neurological conditions, with an emphasis on rare neurological disorders.
Among the speakers from Baylor College of Medicine, two former IRDiRC Task Force members were confirmed, **Dr. Shinya Yamamoto** (Functional Analysis Task Force) and **Dr. Lisa Emrick** (Bridging diagnosis to therapies and care Task Force).

The 13th European Conference on Rare Diseases and Orphan Products (ECRD 2026)

ECRD 2026 stands as the largest patient-led policy-shaping event on rare diseases in Europe, acting as an instrumental hub to advance goal-driven policies which improve the lives of those living with rare diseases.

Organised by EURORDIS, ECRD will take place on 3-4 June 2026 in Prague, Czech Republic, and online under the theme **"Rare Diseases in a Changing & Competitive Europe: Shaping policies to address the unmet needs of people living with rare diseases."**

Expected to welcome 500+ participants onsite and 300+ additional online attendees, this event leverages the strength of the unique rare disease network, bringing together people impacted by rare diseases and patient advocates alongside policy makers, healthcare industry representatives, clinicians, regulators, and Member State officials. Its hybrid format creates a powerful platform for connection and collaboration, enabling participants across the rare disease community to exchange knowledge, share experiences, and build partnerships.

ECRD 2026 will serve as a pivotal milestone in the inclusive, multi-stakeholder effort to launch a **European Blueprint for Rare Diseases**, aiming to cultivate collective ownership and deliver lasting impact across Europe, while strengthening Europe's contribution to the WHO Global Action Plan on Rare Diseases. The conference will delve into the multifaceted challenges and key opportunities shaping the future of the rare disease ecosystem, including therapy development and access to treatments, timely and accurate diagnosis, advances in holistic care, specialised healthcare, health technology assessments, and mental health. To guarantee an accessible and inclusive experience for all, the sessions will feature closed captioning and translation into more than 60 languages.

Event details:

📍 Prague, Czech Republic

🗓️ 3-4 June, 2026

🔗 In-person registration: here.

Full programme

PAST EVENTS

IRDiRC at ERDERA National Alignment Board meeting

IRDiRC was present at **ERDERA National Alignment Board**

(NAB) meeting, a two-day event that gathered European national representatives and international partners to reinforce collective alignment and empower research across all ERDERA participants, including Underrepresented Countries (UCs).

Having the NAB meeting alongside IRDiRC's annual Consortium Assembly and Scientific Committee meeting created space to explore international perspectives on National Mirror Groups (NMGs) and to identify synergies between ERDERA's country-level structures and IRDiRC's global research missions.

Read the article

DIA Europe - What could healthcare look like in 2035 - if we act with the urgency that rare diseases demand?

IRDiRC was present at **DIA 2026 panel session,** chaired and convened by **Estelle Michael** (UCB, member of IRDiRC Companies Constituent Committee), which set the scene for a discussion on a future healthcare ecosystem, and why time matters. During the discussion, **Fleur Chandler** (Duchenne UK) reminded us of a stark reality: around 70% of rare diseases affect children, and approximately 30% of those children die before the age of five. The implication is clear - if progress takes another decade, we risk losing entire generations of children while we deliberate.

Against this backdrop, the panel explored a shared vision for a healthcare ecosystem that learns continuously and acts earlier:

- Every patient generates standardised outcomes data that can be used by regulatory and reimbursement agencies
- Every therapy generates reusable evidence
- Every regulatory and reimbursement decision improves the next one
- Every new diagnosis triggers a defined therapy pathway
- And prevention replaces rescue as the dominant paradigm

IRDiRC members on the panel contributed perspectives from across regulation, industry, and patient organisations - highlighting how quality data, shared evidence frameworks, improved access, and collaboration are essential to making this vision achievable.

Thank you to the panellists for a thoughtful and forward-looking exchange: **Samantha Parker** (Italfarmaco, IRDiRC Vice-Chair), **Violeta Stoyanova-Beninska** (European Medicines Agency,

IRDiRC Regulatory Scientific Committee Chair), **Fleur Chandler** (Duchenne UK), **Lovisa Berggeren** (UCB), and to **Estelle Michael** (UCB) for bringing the session together and guiding the conversation.

At IRDiRC, this vision reflects a direction of travel already underway - through practical tools, shared frameworks, and global collaboration across the rare disease ecosystem.

NEWS FROM IRDiRC MEMBERS

Save the Date

| 79th World Health Assembly RDI Side Event

Rare diseases expose the stress points of our health systems, revealing gaps in diagnosis, care coordination, innovation, data, financing, and access. More importantly, they offer a powerful opportunity: **If health systems are designed to deliver for rare diseases, they can deliver for everyone.**

During the 79th World Health Assembly, **Rare Diseases International (RDI)** will host a side-event in Geneva titled **"Rare Diseases: A Catalyst for Global Health Transformation"**, which will bring together policy makers, patient advocates, industry leaders, researchers, and global health institutions to explore how achieving UHC requires reaching those who are hardest to reach.
This side event builds on the landmark **WHA Resolution "Rare Diseases; a global health priority for equity and inclusion"** adopted at the 78th WHA, which recognized rare diseases as a global health priority, and called for the development of a 10-Year Global Action Plan on Rare Diseases (GAPRD). As we

move toward implementation of these commitments, it is important to reflect on the transversality of rare diseases and the importance of developing people-centred health systems.

This is about improving the lives of over 300 million persons living with rare diseases worldwide, but it's about scaling innovative solutions and designing systems that are more precise, integrated, person-centred, and equitable.

Event details:

📍 Geneva Graduate Institute (Geneva, Switzerland)

🗓️ 20 May 2026, 18:00–20:00 (CEST) (Cocktail reception to follow until 21:00 CEST)

🔗 In-person registration: here.

More Information

Join Undiagnosed Day 2026!

Undiagnosed Day 2026 is a two-day, clinician-led meeting in Gdansk (Poland) bringing together specialist clinicians, clinical geneticists and invited experts to support phenotype-led diagnosis through live case discussion, shared clinical reasoning and practical exchange.

The event is organised by the **European Rare Diseases Research Alliance (ERDERA)**, the **Wilhelm Foundation** (IRDiRC Patient Advocates Constituent Committee member), the **Medical University of Gdansk (GUMed)**, and the **University Clinical Centre in Gdansk (UCK)**.

Although it draws on the same collaborative spirit as international Undiagnosed Hackathons, where patients who have previously failed to receive a diagnosis through standard genetic testing are re-

analysed using more advanced techniques, the Gdansk event is designed around specialist clinical assessment and multidisciplinary discussion with patients and families present.

The meeting will focus on practical approaches to phenotyping and diagnosis in undiagnosed conditions, including how to define next steps when a diagnosis remains uncertain, and how to strengthen pathways and collaboration around undiagnosed care.

Event details:

📍 Gdansk, Poland

🗓️ 29-30 April, 2026

🔗 Agenda and more information: here

Registration

The third Hope for Rare Science Conference in Shanghai

The third Hope for Rare Science Conference will be held in Shanghai, China from 25-27 June, 2026. The conference is hosted by **Hope for Rare Foundation**, with **National Children's Medical Center/ Children's Hospital of Fudan University** and **Chinese Organization for Rare Disorders (CORD)** as co-hosts.

The conference will last for 3 days, featuring 22 parallel sessions and several satellite meetings. More than 80 speakers from over 10 countries have been invited, including representatives from academia, industry, regulatory agencies, and foundations. We warmly welcome you to Shanghai to exchange ideas and promote global rare disease research and translation together.

Event details:

📍 Shanghai, China

🗓️ 25-27 June, 2026

🔗 Agenda and more information: www.hrsc.org.cn.

More Information

Major updates from iHope program

IRDiRC Patient Advocates Constituent Committee (PACC) member, **Genetic Alliance**, is pleased to announce that 9 new clinical sites and 1 new laboratory joined the **iHope** program network, further strengthening the ability to reach families with rare genetic conditions. In addition, Dr. Jennifer Troyer, has joined the staff to support the continued growth and impact of iHope.

Thanks to the dedication of iHope partners and community:

• More than 3,000 children have now been sequenced;

• More than 1,500 diagnoses have been reached, helping families finally get answers.

Every diagnosis represents a child, a family, and a step forward for the rare disease community.

More Information

RARE Drug Development Symposium 2026 "Accelerating Rare Disease Progress: Aligning Advocates, Science & Industry"

The landscape of medical research is shifting: rare disease patient advocates are no longer just participants, they are the architects of discovery. Driven by an urgent need for life-saving treatments, these leaders are spearheading research initiatives with unprecedented momentum. To support this movement, Global Genes (IRDiRC Patient Advocates Constituent Committee member), in partnership with Boston Children's Hospital and the Termeer Institute, will be hosting the annual RARE Drug Development Symposium.

Returning to Boston for its second year following a sold-out 2025 event, this 2.5 day symposium is specifically designed to equip advocates, industry leaders, and academic experts with the tools to navigate early-stage research with clinical precision and confidence.

Attendees will have a mix of main stage sessions, targeted breakouts, group presentations, and hands-on workshopping. Whether you're initiating research efforts or looking to refine your strategy, this symposium offers practical insights to accelerate progress in research strategies and activities.

Event details:

📍 David Rubenstein Treehouse at Harvard University, Boston, MA (USA)

🗓️ 9-11 September, 2026

🔗 More information: https://globalgenes.org/rdds-2026/

European

Parliament Event on Rare Diseases: Placing Patients at the Centre of a Future European Framework

A European Parliament Event on Rare Diseases, under the theme **"Placing Patients at the Centre of a Future European Framework"** took place on 23 April 2026 at the European Parliament in Brussels and online. Organised by **EURORDIS** (member of IRDiRC Patient Advocates Constituent Committee) in collaboration with **MEP Nicolás González Casares**, the event offered a dedicated space for policy makers, patient representatives, advocates, and healthcare experts to contribute to shaping a stronger, more equitable and coordinated European approach to rare disease policy.

Set against a pivotal time in EU health policy, the meeting aimed to place patients' lived experiences at the centre of discussions on a future European framework for rare diseases, and to identify key gaps in diagnosis, care and access to treatments across Europe; explore the role of innovation, including genomics, and bridge the gap between EU institutions and the rare disease community.

More Information

IRDiRC DSC Chair, Sally Ann Lynch, invited to join the Minority advisory board of Genome of Europe

We are pleased to learn that **Dr. Sally Ann Lynch**, Chair of IRDiRC Diagnostics Scientific Committee, has been invited to be a member of the recently formed **Minority advisory board** of Genome of Europe (GoE).

MAB comprises of external experts who will advise GoE consortium on matters involving minority groups, e.g., population subgroups from non-EU countries and those not represented as a country within EU (e.g., Roma, Sinti and Sami).

Other Publications Co-Authored by IRDiRC Members

IRDiRC PACC member, Yukiko Nishimura (ASrid, Japan), co-authors "Quality of life and psychosocial well-being in thyroid eye disease in Japan: Pre-teprotumumab National Survey" publication

Thyroid Eye Disease (TED) is known to cause functional and psychosocial challenges, yet patient-experienced burden in Japan has been poorly understood. In the first nationwide cross-sectional survey in Japan, researchers evaluated health-related quality of life (HRQOL) and psychological wellbeing among 56 patients with TED.

The study highlighted the substantial psychosocial burden faced by Japanese patients with TED, particularly during active disease and when structural eye symptoms are present. While overall physical and mental health measures may appear normal, patients still experience significant disruptions to daily roles and social life. Improving patient-centered care in TED requires more than treating the disease itself. Integrating psychosocial support and targeted symptom management may meaningfully improve quality of life for patients living with this condition.

Read the Publication

IRDiRC ISC Chair, Gareth Baynam, co-authors "TrialR: critical enablers and the need for reusable Rare Disease Clinical Trial infrastructure in Western Australia" publication

More than 400 million people worldwide live with one of over 10,000 identified rare diseases, making rare diseases collectively a major global health challenge. Despite advances in diagnostic technologies, many patients—particularly children—continue to experience significant physical, emotional, and financial burdens, with limited treatment options available. Encouragingly, progress in individualized therapeutics is opening new possibilities for conditions that previously had no definitive treatment. However, inequitable access to rare disease clinical trials remains a major barrier for many families. The TrialR program aims to address this gap by identifying key enablers for rare disease clinical trial access and developing scalable, reusable infrastructure to support trial participation in Western Australia.

Read the Publication

OTHER NEWS

Help pinpoint where consultancy makes the biggest difference in rare disease research

The European Rare Disease Research Alliance (ERDERA) has re-opened its survey to better understand where the rare disease research community most needs external consultancy support.

The goal is simple: turn practical insights from researchers and stakeholders into targeted consultancy services that help projects overcome common bottlenecks and generate robust, timely evidence.

Launched in September 2024 and co-funded by the European Union and participating countries and organisations, ERDERA is assessing demand for expert support in areas such as:

• Ethics and regulatory alignment

• Data readiness and governance

• Patient and public involvement (PPIE)

• Intellectual property and innovation management

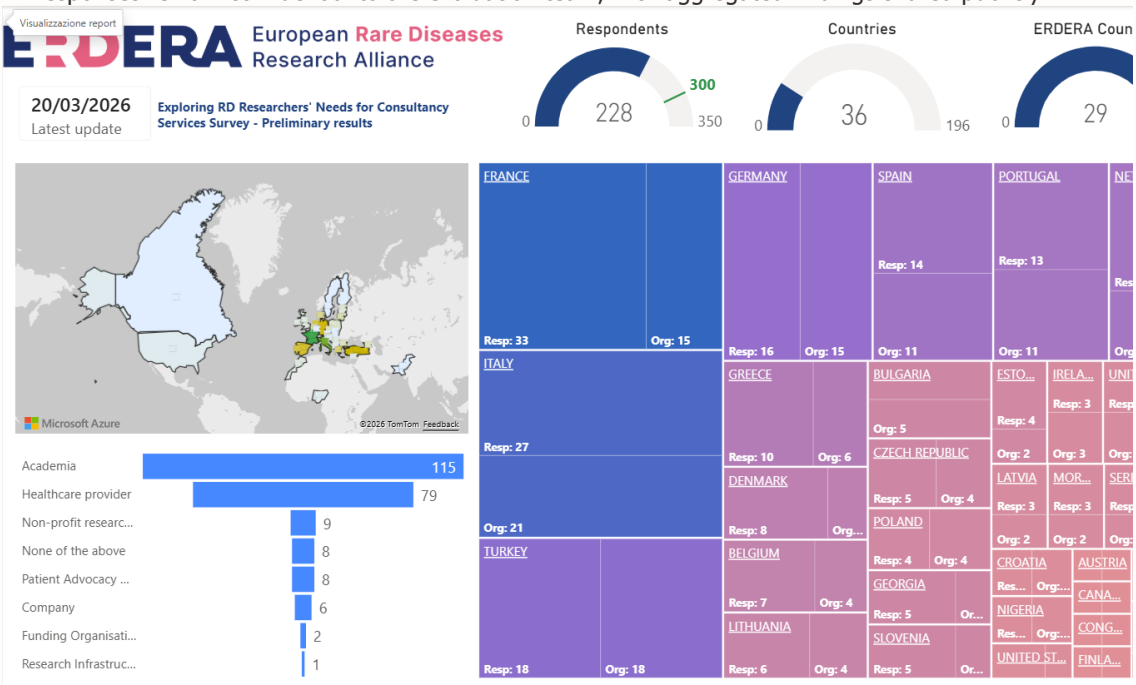
• Study start-up operations

By extending the response window, ERDERA aims to capture more diverse perspectives from researchers, clinicians, project managers, data stewards, and patient partners across Europe and beyond.

The Exploring RD Researchers' Needs for Consultancy Services survey will help:

- Identify which ERDERA service areas should be prioritised
- Define the expertise profiles needed from external advisers
- Inform how ERDERA Expertise Services can remain sustainable beyond the partnership's funding period

All responses remain confidential to the evaluation team, with aggregated findings shared publicly.



Complete the Survey

CONTACT & SOCIAL MEDIA

LinkedIn: International Rare Diseases Research Consortium (IRDiRC)

X (Twitter): IRDiRC

Website: https://irdirc.org/

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