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IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

Nomination Call for Experts - Therapies Scientific Committee

IRDiRC has four Scientific Committees. The Therapies Scientific Committee (TSC) is a multi-stakeholder, multi-disciplinary group of experts in medical research and therapy development in rare diseases.

Specifically, the TSC is devoted to pursuing the therapeutic development of IRDiRC, supporting the rare diseases research community in achieving goal 2, **"1000 new rare disease treatments by 2027."**

We would like to broaden the committee membership, to represent the worldwide rare diseases therapies community in its full width, from early innovation towards regulatory approval and therapeutic support of patients. As such, the TSC has **three openings** for new members. We are looking for rare disease experts from different backgrounds with the following qualities:

- Demonstrable track record in contributing to rare disease therapeutic development in either medicines and/ or medical devices (could be Industry, Regulatory, Research and Academia, Health Technology Appraisal/Payer, Clinical care).
- Expertise will be considered from across the therapeutic spectrum, including from prevention, management, quality of life support to curative care.
- Commitment to active contribution at monthly teleconferences, a yearly face-to-face meeting and regular TSC activities, article drafting, and email correspondence.

If you are interested, please send before the **6th of April 2025** a CV, biosketch and letter of motivation to the scientific.secretariat[at]irdirc.org.

More Information

IRDiRC RSC Chair, Violeta Stoyanova-Beninska receives EURORDIS Black Pearl Award for Leadership

We extend our sincere congratulations to the IRDiRC Regulatory Scientific Committee Chair, **Dr. Violeta Stoyanova-Beninska**, who received the EURORDIS **2025 Leadership Award**. This award is presented each year to leaders whose exceptional dedication and commitment have made a significant difference to the rare disease community.

Through her mandate as the Chair of the Committee for Orphan Medicinal Products (COMP) at the European Medicinal Agency (EMA), Dr Stoyanova-Beninska's leadership was fundamental in influencing patient-centred rare disease health policy and legislation, thereby shaping orphan drug policies and enhancing patient involvement in regulatory activities.

RE(ACT) Congress and IRDiRC Conference 2025

From March 5 to 7, 2025, the RE(ACT) Congress and IRDiRC Conference was hosted in Brussels, an event focused on rare and orphan diseases research. Organized by the IRDiRC in collaboration with the BLACKSWAN FOUNDATION and ERDERA (European Rare Diseases Research Alliance), the congress brought together researchers, clinicians, policymakers, and patient organizations to discuss scientific advances and key challenges in the field.

The event covered several important themes, including:

- ***Diagnosing rare diseases:*** new approaches combining genomic sequencing and artificial intelligence.
- ***Health data utilization:*** structuring and sharing information to advance research.
- ***Advanced therapy medicinal products (ATMPs):*** developments in gene and cell therapies.
- ***Drug repurposing:*** identifying new uses for existing treatments.
- ***Funding models:*** strategies to support rare disease research.
- ***The impact of rare diseases on patients and families:*** improving care and quality of life.

The discussions highlighted the importance of international cooperation to accelerate diagnosis and the development of treatments. The shared goal remains to improve access to care and therapies for people living with rare diseases.

The RE(ACT) Congress & IRDiRC Conference will return in 2027 to continue these discussions and track progress in the field.

More Information

IRDiRC EVENTS

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

- **Online Consortium Assembly Meeting:** on 13 June 2025.
- **In-person Consortium Assembly Meeting:** in **Paris, France**, on 15-16 October, 2025.

Collaborative events:

- **International Rare Disease Conference of China (IRDCC),** organized by **CHARD**: Location - Haikou, Hainan Province, China, on 24 May, 2025**.**

**Photo from the CA-SC Meeting in Brussels, Belgium (March 2025)*

World Orphan Drug Congress Europe 2025

The World Orphan Drug Congress Europe is the largest and most established event globally for orphan drugs and rare diseases.

Joined by over 2,000 attendees, from 250+ industry leaders, it is an opportunity to connect with over 130 exhibitors and experts from the entire orphan drug lifecycle, and discuss areas such as regulatory affairs, policy, global pricing and cutting-edge gene therapies.

The attendees represent the most senior individuals across the industry, including leaders in R&D, gene therapy, rare diseases, precision/personalised medicine, genetic disorders, patient advocacy and engagement, associations in ATMPs, investors, regulatory affairs professions, governmental bodies, and many more.

📅27th October 2025 | Pre-Congress Workshops

📅28th - 29th October 2025 | Main Congress Days

📍**RAI Congress Centre, Amsterdam, Netherlands**

See you there!

Register now

NEWS FROM IRDiRC MEMBERS

SANT launches public consultation on rare diseases

On 28 February, marking the 2025 Rare Disease Day, European Parliament's public health committee released its survey on rare diseases.

The purpose of the public consultation is to provide a basis for the forthcoming work of the **Committee on Public Health (SANT)** in bringing a better understanding and detailed knowledge of the challenges of persons affected by rare diseases and views of persons working with or involved in rare diseases.

The online survey can be filled in by all interested parties and is open for one month. Citizens, but also stakeholders (patient organisations, representatives of industry, NGOs etc.) have the opportunity to provide anonymous input with information about their difficulties, needs and expectations regarding further EU actions in the field of rare diseases.

More information

GA4GH Clinical Impact

Genetic Alliance will be hosting a **GA4GH Connect session** in **Boston (US)**, titled **"Towards Clinical Application of GA4GH Products"**, and they are looking for real-world clinical use cases that GA4GH products could help address, including input from:

- Clinical providers
- Translational researchers
- Testing laboratories
- Software vendors
- Biopharma industry
- Government initiatives
- Standards development organisations
- Regulatory and policy groups

Please **share your ideas** at the following link: **here**.

Have a look at the key questions they're hoping to answer:

1) What clinical use cases could be addressed by GA4GH products? *(e.g. identifying the most relevant genetic test(s) to order, interpreting tests results, decision support at point of care, clinical trial matching, patient/provider education, finding an expert for a specific genetic condition)*

2) Which GA4GH products can or should support those clinical use cases?

3) What opportunities exist to improve GA4GH products to address gaps or pain points?

NIH - Newborn Screening by Whole Genome Sequencing Collaboratory Research Opportunity Announcement

The US National Institutes of Health announces a new collaboratory research opportunity on **Newborn Screening by Whole Genome Sequencing**.

Eligible organisations are invited to submit their application before Friday, April 4th, 2025, local time of applicant organisation. LOIs are required to submit a full application.

With significant community involvement and input, this initiative will assess the feasibility of developing a collaborative model to allow incorporation of genomic sequencing, as a screening tool for select monogenic diseases that are actionable in early childhood, into the existing state-based US public health newborn screening program.

More Information

Webinar - Challenges in Defining Elements of Value in DMD for Decision Making

Don't miss out an upcoming ISPOR webinar, sponsored by Italfarmaco SpA.

This session will explore the challenges of traditional HTA frameworks in evaluating rare, paediatric, degenerative diseases, using Duchenne Muscular Dystrophy (DMD) as a case study. We have assembled an exceptional panel of experts who will share innovative, multi-perspective insights on assessing orphan drugs.

This is a unique opportunity to gain valuable knowledge and join the discussion on shaping the future of HTA for rare diseases.

The Learning Objectives are :

- Introduce the current challenges with traditional value assessment frameworks, with a focus on rare diseases and DMD; and understand the factors influencing societal willingness and ability to pay for treatments.
- Understand the relevance of caregiver burden and why it should be considered an element of decision making for the assessment of new DMD treatments, from a patient and payer perspective including impact on costs and well-being.
- Describe potential methods for capturing these factors in the HTA context for DMD, including impact on caregiver costs and well-being.

Webinar details:

- Date: Tuesday, March 25, 2025
- Time: 12:00PM EDT | 4:00PM UTC | 5:00PM CET

Free registration.

More Information

OTHER NEWS

RealiseD
compRehensive mEthodological and operational Approach to cLinical trialS in ultra-rarE Diseases

From the Rare Disease Moonshot's vision to the launch of the RealiseD project

The RealiseD project, funded under the Innovative Health Initiative (IHI), emerged in direct response to the strategic priorities set by the Rare Disease Moonshot. The Moonshot's **Clinical Trials Research Needs Recommendations** provided a structured foundation that helped shaping RealiseD's objectives, methodologies, direction and expected outcomes.

The origins of RealiseD can indeed be traced back to the EFPIA-EURORDIS Joint Statement on Patient Access to Medicines for Rare Diseases released in June 2022. Proposal 6 of this statement called for a Rare Disease Moonshot, recognising the need for a coordinated and ambitious approach to tackling persistent gaps in rare disease research and drug development. This call to action led to the establishment of the Rare Disease Moonshot launched in December 2022 at the European Health Summit, and which in turn produced research needs recommendations, particularly to optimise clinical trials. These recommendations directly informed the design and implementation of RealiseD.

"The Rare Disease Moonshot made it clear that we needed more than just recognition of the problem—we needed structured solutions that could be implemented at scale. RealiseD was conceived as a direct response to that need, building on the recommendations to create practical frameworks for optimising clinical trials and regulatory pathways in rare diseases." – Solange Corriol-Rohou (AstraZeneca and RealiseD lead)

A key feature of RealiseD is the development of playbooks—structured guides aimed at improving the design and execution of clinical trials for rare diseases. It is also planned to identify solutions to address scientific gaps and technical and operational challenges, and find synergies with relevant existing initiatives (e.g., ERDERA, ERAMET or IDEAL) to establish a new, dedicated, rare-disease-specific and sustainable infrastructure. These playbooks, shaped by the RD Moonshot's research recommendations, will provide standardised methodologies for researchers and industry stakeholders. The project also prioritises regulatory-grade data integration, responding to the RD Moonshot's emphasis on embedding regulatory science in trial design to facilitate approval processes and patient access. Furthermore, RealiseD embraces the Moonshot's model of cross-sector collaboration, bringing together academic institutions and industry leaders under the IHI framework to strengthen translational research and clinical implementation. The public-private spirit of this project is also embodied with the in-kind contribution from the industry matching the public EU funding (with a total budget over 17M€). This model should not be the future. The needs are for today.

The RealiseD project demonstrates the tangible impact of the Rare Disease Moonshot. By transforming research recommendations into funded initiatives, it highlights how strategic collaboration, regulatory integration, and innovative trial design can help overcome the challenges of rare disease research.

We call on all research funders, public, private, or charities, to prioritise funding opportunities that centre on public-private partnerships and push beyond existing frameworks (such as the IHI). These partnerships have consistently proven their effectiveness and impact. Given the urgent need for innovation in rare disease research and the implementation of its outcomes, it is crucial to utilise these mechanisms. The evidence is clear: strategic multistakeholder collaboration accelerates progress and delivers tangible benefits to patients.

More information

Don't forget to follow us!

LinkedIn: International Rare Diseases Research Consortium (IRDiRC)

X (Twitter): IRDiRC

Website: https://irdirc.org/

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