Report

IRDiRC Consortium Assembly Meeting

15 December 2025
Online

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

MEETING REPORT CONTENTS

15 December 2025:

ABBREVIATIONS

AI – Artificial Intelligence

ATMPs - Advanced therapy medicinal products

CA – Consortium Assembly

CA-SC – Consortium Assembly and Scientific Committees

CCC – Companies Constituent Committee

DSC – Diagnostics Scientific Committee

ERDERA – European Rare Diseases Research Alliance

FAIR – Findable, Accessible, Interoperable, Reusable

FCC – Funders Constituent Committee

FDA – Food and Drug Administration

GDPR – General Data Protection Regulation

HTA – Health Technology Assessment

IRDiRC – International Rare Diseases Research Consortium

ISC – Interdisciplinary Scientific Committee

LMIC – Low-Medium Income Country

PACC – Patient Advocacy Constituent Committee

RD – Rare Disease

RWD – Real World Data

TF – Task Force

TSC – Therapies Scientific Committee

WG – Working Group

WODC – World Orphan Drug Congress

THE REPORT

The International Rare Diseases Research Consortium (IRDiRC) organized an online meeting of the Consortium Assembly (CA) on 15 December, 2025.

15 December 2025

Agenda

1. New constituent committee members

Patient Advocacy Constituent Committee

COMBINEDBrain, the Consortium for Outcome Measures and Biomarkers for Neurodevelopmental Disorders, a non-profit organization based in the USA, joined the IRDiRC Patient Advocacy Constituent Committee (PACC). The organization will be represented by Terry Jo Bichell, Chief Executive Officer, translational neuroscientist and parent advocate. COMBINEDBrain is a non-profit consortium of approximately 120 patient advocacy groups for rare genetic neurodevelopmental disorders.

2. New leadership

Funders Constituent Committee (FCC)

IRDiRC Funders Constituent Committee appointed **Stefano Benvenuti**, Head of Public Affairs at Fondazione Telethon, Italy, as Chair of FCC, and **Naveed Aziz**, Vice President Research & Innovation at Genome Canada, Canada, as Vice Chair of the same committee.

Therapies Scientific Committee (TSC)

IRDiRC Therapies Scientific Committee appointed **Shekhar Natarajan**, Vice President and Head of EU and International Regulatory Affairs at Dyne Therapeutics, UK based, as Chair of the committee, respectively **Diana Kwast-Hoekstra,** President and Founder of (Un)limited Forward! Foundation and PhD candidate at University Medical Center Groningen, based in Netherlands**,**as Vice Chair.

3. Roadmap 2026 presentation

For Roadmap 2026, the selected Task Forces (TFs) and Working Group (WG) will launch an open call for membership in January 2026. Each Task Force is comprised of approximately 15 members, with a balanced expertise and geographical distribution across stakeholder types. The Task Forces will convene on a regular basis throughout the year to ensure continued dialogue, coordination and progress towards shared objectives, and they are expected to hold an in-person workshop. The usual deliverables of a Task Force are white papers/publications, along with any additional outputs that may support a broader dissemination of results, such as reports, factsheets, etc.

The Working Groups take place for a shorter period of time, usually 6 months with the possibility of an extension of extra 3 months. The WG will convene just through online teleconferences. The usual deliverable of a WG is a white paper or report.

According to the IRDiRC Governance, the Consortium Assembly must approve the proposed TFs through an online anonymous voting form. An email will be sent by the Scientific Secretariat to all Consortium Assembly (CA) members requesting validation of the new activities, and more than half of the members must respond in order to proceed with the 2026 Roadmap.

3.1 Task Force – Models of Care Coordination (proposed by Interdisciplinary Scientific Committee)

Background

IRDiRC's vision includes not only diagnosis and therapy, but also timely, effective care. The Interdisciplinary Scientific Committee noted that models of care and care coordination remain under-studied and unevenly implemented across rare diseases. As most rare diseases still lack disease-modifying therapies, how care is organized has major impact on quality of life and burden for patients/families.

Proposed focus

- Map and analyze current care coordination approaches across rare diseases and health systems.
- Develop a conceptual framework to design, implement, and evaluate models of care coordination, incorporating:
  - Equity
  - Patient-centred outcomes
  - Links to research participation and knowledge generation
  - Potential technology-enabled coordination
- And ensure that they enhance the way care coordination is implemented to reduce patient and family burden

Context

- Intended to complement broader initiatives (such as Global Nursing Network for Rare Disease and Lancet Commission on Rare Diseases) by focusing specifically on care coordination rather than only care pathways.

Discussion highlights

- Consider including learnings from the International Clinical Research Networks (CRN) meeting (Heidelberg), including examples of care coordination in Duchenne Muscular Dystrophy (Czech Republic/Poland) and parallels to "service line/navigation" approaches used in other fields.

Resources shared on the chat

- https://pubmed.ncbi.nlm.nih.gov/41026964/
- https://onlinelibrary.wiley.com/doi/10.1111/jir.70049
- https://www.hse.ie/eng/services/list/5/rarediseases/transition-guide-for-rare-diseases.pdf

3.2 Task Force – Data Sources and Registries (proposed by Companies Constituent Committee)

Background

Increasing reliance on real-world data and AI/modeling heightens the need for data that are high quality, accessible, and fit-for-purpose—yet rare disease data remain scarce, fragmented, and inconsistently standardized, with persistent issues around access and regulatory usability. There are growing concerns about "wasted data": collections exist but cannot be used due to design/logistical/standardization barriers.

Proposed focus

- Not to reinvent existing registry standards, but to provide guiding principles and practical navigation in a complex standards landscape.
- Identify proven/promising strategies for building or leveraging a "high-quality standardized ecosystem" (which may include but is not limited to registries).
- Potential outputs include:
    - A taxonomy clarifying what different "data sources/registries" mean in practice
    - Compile lessons learned and emerging strategies for registries, Real-World Data, and novel data sources
    - Approaches to legacy systems
    - Consideration of differences across health systems and geographies
    - Success criteria and best-practice compilation tailored to rare diseases

3.3 Task Force – Digital Twins (proposed by Therapies Scientific Committee)

Background

Digital twins are described as dynamic, data-driven virtual representations of individual patients, integrating multi-modal data: genomic, clinical, imaging, biomarkers, behavioral, etc. to simulate disease and treatment response. Potentially valuable in rare diseases where cohorts are very small and heterogeneity is high.

Key challenges

- Data scarcity increases sensitivity to bias, privacy risk, and disproportionate influence of single individuals on models.
- Need for explicit ethical governance and responsible implementation approaches.

Proposed objectives and deliverables

- Produce a dedicated landscape and gap analysis for digital twins in rare diseases.
- Map initiatives and align with relevant standards/frameworks (e.g., Findable, Accessible, Interoperable, Reusable (FAIR) principles, interoperability approaches, General Data Protection Regulation (GDPR) considerations were referenced).
- Engage stakeholders (patients, regulators, technology developers) via consultations and a webinar.
- Deliver peer-reviewed publication(s) and actionable recommendations for responsible and equitable implementation.

The Task Force will explicitly align its analyses and recommendations with IRDiRC goals on diagnosis, therapy development, and assessment, particularly in contexts where traditional endpoints are difficult to establish.

Discussion highlights

- Strong recommendation to include multiple regulatory perspectives, and to consider Health Technology Assessment (HTA)/payer viewpoints as stakeholders.

Resources shared on the chat

- https://www.virtualhumantwins.eu/manifesto

3.4 Working Group – Digital Biomarkers (proposed by Therapies Scientific Committee)

Background

This activity builds on prior IRDiRC work published in a *Nature Reviews Drug Discovery* paper on how digital biomarkers could change rare disease drug development https://pubmed.ncbi.nlm.nih.gov/39681697/.
It was noted that qualification/validation of digital biomarkers can take many years; one example referenced was a digital endpoint in Duchenne (Stride Velocity 95th Centile) with a long development runway.

Proposed focus

- Use a survey-based, multi-stakeholder analysis (drug developers, technology developers, regulators, patients/patient organizations) to define what a credible "weight of evidence" could look like to accelerate qualification/validation and adoption.
- Identify barriers and potential enabling technical standards.
- Intended outputs: frameworks/processes to support adoption and clearer qualification pathways.

Discussion highlights

- Ongoing work using 3D photography/Cliniface as a digital biomarker in certain disorders. Potential to connect with other Task Forces.

4. 2026 Timeline

- *January*
  - Open call for membership for approved Task Forces and Working Group to be published on the website and social media.
  - 9th January – IRDiRC Yearly Strategic Meeting in Paris, France.

- *18-19 March*
  - Annual Consortium Assembly and Scientific Committees (CA-SC) Meeting in Sofia, Bulgaria.
  - Planned as a back-to-back with the European Rare Diseases Research Alliance (ERDERA) meeting National Mirror Groups (NMGs) annual meeting. The CA-SC will

include a joint session with ERDERA's NMG in the morning of 18 March (agenda to be finalized and shared early 2026).

- *June*
  - Online Consortium Assembly Meeting (*date to be confirmed*)
  - 9-11 June – IRDiRC's participation at World Orphan Drug Congress in Boston (MA), USA

5. Summary of Scientific Secretariat Activities in 2025

Meetings

Throughout the year, the IRDiRC convened 25 internal operational meetings and one in-person strategic meeting in January 2025. The Secretariat organized 12 meetings with the IRDiRC Operating Committee (one per month), two in-person Consortium Assembly (+ one Scientific Committee) Meetings in Bruxelles (Belgium) and in Paris (France) and two online. In addition, the Scientific Secretariat organized 42 committees meeting, 38 Task Forces and Working Group meetings and 2 in-person workshops for Preventive Medicines Task Force (October), and for Regulatory Convergence Task Force (November). In parallel, the Secretariat organized 25 meetings for the planning, development or organization of other activities, including but not limited to: Pre-competitive space working publication, World Orphan Drug Congress scientific programme, the 2nd International Clinical Research Networks Conference, Drug Repurposing, ERDERA International WorkStream, etc.

Events Organization and Participation

RE(ACT) Congress and IRDiRC Conference, 5-7 March 2025, Bruxelles, Belgium

From March 5 to 7, 2025, Brussels hosted the RE(ACT) Congress & IRDiRC Conference, an international event dedicated to advancing research on rare and orphan diseases. Organized by the International Rare Diseases Research Consortium (IRDiRC) in collaboration with the BLACKSWAN Foundation and ERDERA, the conference brought together researchers, clinicians, policy-makers, and patient organizations to address scientific progress and ongoing challenges in the field. Discussions focused on innovative diagnostic approaches combining genomic sequencing and artificial intelligence, improved use and sharing of health data, advances in gene and cell therapies (ATMPs), drug repositioning strategies, sustainable funding models, and the profound impact of rare diseases on patients and their families, with particular attention to care pathways and quality of life.

World Orphan Drug Congress Europe, 27-29 October 2025, Amsterdam, Netherlands

The World Orphan Drug Congress 2025 in Amsterdam brought together global leaders from the rare disease community for three days of collaboration and forward-looking discussion focused on advancing research and improving patient access to therapies. Representatives from the International Rare Diseases Research Consortium (IRDiRC) played a central role, contributing to policy, research, industry, and patient perspectives. IRDiRC had a booth at the congress and 2 members of the Scientific Secretariat joined on site.

ERDERA General Assembly, 29-31 October 2025, Amsterdam, Netherlands

The European Rare Diseases Research Alliance marked its first anniversary through a 3-days event in Amsterdam, Netherlands. The Assembly featured an open session for wider audience, presenting two keynote speeches by Fanny Mochel, Director at the Reference Centre for Neurometabolic Diseases at La

Pitié-Salpêtrière Hospital (France) and Bojana Mirosavljevic, Founder of Zivot-Life, a rare disease patients association from Serbia.

Second International Clinical Research Networks Conference, 9-10 December, Heidelberg, Germany

During 9-10 December, the second edition of the Clinical Research Networks Conference took place in Heidelberg, Germany, and online, gathering approximately 85 rare disease expert leaders, regulators, clinicians and researchers on site. IRDiRC was co-organizer of the conference together with ERDERA and Rare Diseases International (RDI). Read the conference summary: https://irdirc.org/clinical-experts-and-patient-advocates-gather-in-heidelberg-to-turn-discussion-into-action-on-global-rare-disease-research/

6. Publications in 2025

Publication Name	Authors	Journal
Non-oncology orphan drug development: Productivity and probability of success	Samantha Parker, Jida El Hajjar, Anneliene H. Jonker, Susan R. Kahn, Persefoni Kritikou, Christina Kyriakopoulou, Anthony Haight	Drug Discovery Today
Non-oncologic orphan drug approvals across the world: Types of evidence required and time to approval	Anne R. Pariser, Violeta Stoyanova-Beninska, Oxana Iliach, Reda Jundi, Kerry Jo Lee, Hanako Morikawa, Samantha Parker, Caroline Pothet, Marco Rizzi, Julie Vaillancourt, Ana Hidalgo-Simon	Drug Discovery Today
Regulatory sandboxes: A new frontier for rare disease therapies	Galliano Zanello, Violeta Stoyanova-Beninska, Oxana Iliach, Daniel Scherman, Samantha Parker, David A. Pearce	Rare
What matters ethically about how the UDN has changed since its inception?	David A. Pearce, Elena-Alexandra Tataru	AMA Journal of Ethics
Applying the international rare disease research consortium (IRDiRC) N-of-1 therapy task force eligibility criteria for individualised therapies use case: Duchenne muscular dystrophy	Annemieke Aartsma-Rus, Anneliene H Jonker, Daniel O'Connor	Neuromuscular Disorders
From roadmap to a sustainable end-to-end individualized therapy pathway	Anneliene H Jonker, Elena-Alexandra Tataru, David P Dimmock, Alison Bateman-House, Holm Graessner, Gareth Baynam, Erika F Augustine, Adam Jaffe, Anna M G Pasmooij, Oxana Iliach, Richard Horgan, James Davies, Shruti Mitkus, Larissa Lapteva, Matthis Synofzik, Timothy W Yu,	Therapeutic Advances in Rare Disease

	Daniel O'Connor, Annemieke Aartsma-Rus	
Drug-device combinations in rare diseases: Challenges and opportunities	Elena-Alexandra Tataru, Marc Dooms, Claudia Gonzaga-Jauregui, Anna Maria Gerdina Pasmooij, Daniel J. O'Connor, Anneliene H. Jonker	Drug Discovery Today

IRDiRC publications are available at the following link: https://irdirc.org/irdirc-publications/

7. Communication

In 2025, IRDiRC produced 5 newsletters, and engaged a stronger presence on LinkedIn, and to a lesser extent on X, former Twitter.

Website enhancements are currently in progress. Among the changes that were deployed: improved user interface, closure of outdated/inactive pages, new plugins, and restructuring of tabs to better highlight IRDiRC work.

Members are encouraged to submit institutional updates via a monthly Microsoft Form, enabling dissemination via website and social channels.

A communications strategic document is in preparation, targeted for completion by end of 2025.

Acknowledgements

This report was prepared by IRDiRC scientific project managers Alexandra Tataru and Galliano Zanello (Fondation Maladies Rares, Paris, France).

Disclaimer

Certain elements of the meeting discussion have been intentionally omitted from this report, as the details relate to sensitive or confidential matters. This document is intended for public dissemination. The opinions presented in the report do not reflect the position of member organizations.