Report

IRDiRC Consortium Assembly Meeting

13 June 2025
Online

MEETING REPORT CONTENTS

13 June 2025:

THE REPORT

The International Rare Diseases Research Consortium (IRDiRC) organized an online meeting of the Consortium Assembly (CA) on June 13, 2025.

13 June 2025

1. Welcome and presentation of the meeting agenda

2. New members and changes of representation

New members Therapies Scientific Committee (TSC) - IRDiRC welcomes 4 new members in TSC:

- **Shekhar Natarajan**, Vice President, Head of EU and International Regulatory Affairs, Dyne Therapeutics, USA
- **Dorota Zgodka,** Pharmaceutical and Healthcare Executive for the European Rare Diseases Research Alliance (ERDERA), Multi-stakeholder Advisory Board member, Switzerland
- **Joao Rocha**, Associate Professor of Pharmacology and Pharmacotherapy, University of Lisbon, Portugal
- **Jill Weimer,** Senior Director of Therapeutic Development, Sanford Research, USA

Changes of representation

Companies Constituent Committee (CCC)

- **UCB: Estelle Michael**, Policy and Public Affairs Lead, Belgium
- **Alexion AstraZeneca Rare Disease: Rohita Sharma**, Senior Director, Global Strategic Alliances, USA

3. Leadership Changes

Patient Advocacy Constituent Committee (PACC)

- PACC chair: **Kelly du Plessis**, Rare Diseases International, South Africa
- PACC vice chair: **Tammi Shipowick**, Program Director at GlobalSkin, Canada

Diagnostic Scientific Committee (DSC)

- DSC vice chair: **Saumya Jamuar**, Medical Director at KK Women's and Children's Hospital, Singapore

4. Progress of IRDiRC activities in 2025 (Q1-Q2)

Task Forces and initiatives

Five activities have been ongoing in 2025.

(1) Toward clarifications, improvement and international convergence in regulatory approvals of rare disease therapies with a focus on Advanced Therapy Medicinal Products (TF)
(2) Facilitating the development of Preventive Medicines at scale for rare diseases (TF)
(3) Bridging diagnosis to therapies and care (TF)
(4) Stigma and Rare Diseases (TF)
(5) Engagement of Young People Living with Rare Diseases in Therapy Development (WG)

(1) Toward clarifications, improvement and international convergence in regulatory approvals of rare disease therapies with a focus on Advanced Therapy Medicinal Products (ATMPs)

Chairpersons: Christopher McMaster (FCC), Violeta Stoyanova-Beninska (RSC), Vinciane Pirard (CCC)

Background

The current regulatory and manufacturing environment for innovative rare disease therapies is extremely complex and differs between jurisdictions. The current scheme struggles with adapting to the peculiarity of ultra rare diseases, more and more often treated with gene therapies, and this lengthens the route to patients and increases costs of therapies. Moreover, the need to navigate different regulatory procedures in different jurisdictions further complicates the picture. Consequently, there is an increasing disparity in medicines available to treat rare diseases between different countries. The overall objective of the task force is to identify similarities and differences in the approval of therapies by different jurisdictions and propose concrete solutions. Chemistry, Manufacturing, and Controls (CMC) will be considered as first hurdle, but also any other regulatory requirements.

Objectives

- Identify similarities and differences in the approval of therapies (both regulatory and CMC) by different jurisdictions (initial focus on ATMPs, adeno-associated virus (AAVs) based gene therapies)
- Use Real World Evidence (RWE) examples from companies and philanthropic organizations to point out pain points in the approval process (time, cost, confusion, *etc*)
- Propose solutions toward regulatory convergence - including engagement with regulatory agencies
- Create a map/flowchart for researchers/funders with guidance on navigating regulatory approval for RD therapies
- Publish in a peer reviewed journal and highly promote these findings

Task Force Progress

Two working groups were established that work in parallel, every 3 weeks, and update common meetings (4 online teleconferences + 1 in-person workshop):

Group 1:

- Mapping and analyzing current regulatory collaborative frameworks and identifying discrepancies

Group 2:

- Use case approach: Investigating scientific, regulatory and CMC pathways
- Preparing a survey/interview to gather information on the use cases

An in-person workshop is planned on 26-27 November 2025 in Paris, France.

Comments from audience:

IRDiRC chair highlighted that, based on the discussion led by the Regulatory Scientific Committee chair at the Consortium Assembly – Scientific Committee meeting in Bruxelles, a manuscript has been produced and submitted to a journal. The group is waiting for the journal's revision. The chair of the Therapies Scientific Committee discussed how the platform technology designation from Food and Drug Administration (FDA) is more applicable for gene therapy and gene editing. This represents a huge topic and there is an emerging opportunity to harmonize some of these requirements and incentives and help inform the regulators. On another level, there is a publication produced by IRDiRC on shared molecular etiologies using basket trials which raised some regulatory issues on acceptability and therapeutic development.

(2) Facilitating the development of Preventive Medicines at scale for rare diseases

Chair: Dan O'Connor (TSC)

Background

Amongst patients, industry, and policymakers there is a growing interest in disease prevention in order to improve population health – huge opportunity in rare diseases. The majority of orphan designations are for 'treatment', clustered around just a few conditions - clearly demonstrates that more must be done to develop a proactive approach to developing preventative medicines at scale, rebalancing the delivery of healthcare away from acute care into prevention.

Objectives

- Map the current preventative medicine pathway(s) and create recommendations that recognizes and manage the additional uncertainties of earlier intervention and small population research, in order to raise the visibility of prevention, and encourage development and access opportunities

Task Force Progress

- Kick off meeting held and monthly TF meetings in place – good proactive engagement with members
- Scope of 'prevention' problem statement agreed with TF (what it can and should cover), with recognition of some key areas to include (primary and secondary, rare versus ultra rare, Low-Middle Income Countries (LMIC))
- Outline of a paper drafted with key sections in place – contact with Nature Reviews Drug Discovery (NRDD) to align on the structure and submission timeline
- Moving towards technical discussion and areas to develop
- Considering what an in-person meeting might involve and the timeline

Comments from audience:

The RSC chair inquired whether when mentioning primary or secondary prevention, does this mean looking at existing medicines or potential substances which can be used for early diagnosis prevention? Another point specified is pregnancy prenatal diagnosis. Interested to know more details about data collection. *Response:* The development programs are still positioned around treatment. The orphan drug designations highlight the missed opportunities, because the main focus remains on the treatment of the condition rather than on the opportunities for prevention. Designations for prevention are also clustered around a very small number of conditions.

(3) Bridging diagnosis to therapies and care

Chairs: Clara Van Karnebeek (former DSC vice chair), Anneliene Jonker (TSC)

Background

This task force is the cumulative work of 3 committees, Diagnostics, Interdisciplinary and Therapies Scientific Committee, as individualized care and therapies play a fundamental and essential role in the journey of a person with any medical condition, but specifically in rare diseases.
Provision of individualized care is a fundamental goal in the journey of a person with a longstanding medical condition. In rare diseases, therapeutic care encompasses a wide variety of potential options: pharmaceutical agents, nutritional therapy, genomic and cell therapies, medical devices, medical procedures, supportive therapy, education and many more. A considerable number of RD patients suffer from two or more distinct diseases, mandating adjustments of therapeutic approaches. Personalized outcomes and reliable monitoring for effect and safety are essential for personalized medicine. Taken all together, the need exists to link and accelerate diagnosis to targeted therapy and individualized care and establish a better understanding of this trajectory.

Objectives

- Create a list of critical elements required for the transition from diagnosis to therapy, including barriers, opportunities and recommendations for implementation;

- Exhaust opportunities to connect newly diagnosed individuals to existing therapies, both approved and on a research basis (SoC procedures/disease management guidelines; approved, experimental, compassionate use or off-label therapy; towards self-management; psychological support; educational, disability, social and work support);
- Prioritization of case-specific tasks needed to instate available therapies;
- Prioritization work needed to bridge existing gaps between newly diagnoses and effective therapy or research.

Task Force Progress

The task force group has worked towards a definition of therapy and pathway and started to map the pathway and potential information sources.

By creating a map of trajectory, the group discussed on the best ways to link the diagnosis with the case management path, taking into consideration the clinical decision that supports each step of the way. When moving towards a therapy it is necessary to have some decisive steps including companion diagnostics, biomarkers, enzyme assays, genetic tests, but also to define which therapy would be appropriate and when to start or stop it. In addition, there is also the secondary support: psychological, social support, symptomatic treatment.

(4) Engagement of Young People Living with Rare Diseases in Therapy Development

Chairpersons: Anneliene Jonker (TSC), Maria Cavaller Bellaubi (TSC)

Background

Engaging young people in research is essential for advancing therapies for rare diseases and from a research perspective has been shown to improve inclusion and retention in clinical trials and research outcomes. Despite the existence of different global initiatives supporting youth involvement, there is a lack of clear and harmonized guidance on how to effectively engage young people in rare disease research, including in the design and execution of clinical studies.

Objectives

- Understand and map out different patient engagement activities in therapy development by age groups and determine if there are differences if the young people lived with the condition their entire life or if they were diagnosed at a later age;
- Understand existing methodologies, tools and processes for young people engagement and identify the gaps;
- Create new opportunities for engaging young people in the whole process of therapy development.

Working Group Progress

- Discussion scope of WG

- Discussion on best practice cases
- Start of checklist/ decision tree making for young people engagement

Comments from audience:

A companies constituent committee representative mentioned that there are many initiatives around patient engagement, including ones dedicated to young patients and requested additional clarifications on mapping the practice, if this is expected to be a compendium of good practice in young patients engagement in general, as such meta-analysis has been done at one point. *Response:* The most severe pediatric diseases are rare. There are also representatives from the previous Innovative Health Initiative (IHI) project, connect4children (c4c) in the working group. When studying the typical methodology applied in adults for drug development, we observe differences compared with the pediatric population, so it is needed a different approach.

(5) Stigma and rare diseases

Chairs: Gareth Baynam (ISC), Ritu Jain (ISC), Marc Dooms (ISC), Samuel Wiafe (PACC)

Background

For many People Living with a Rare Disease (PLWRD), stigma is a critical health and social barrier with a high unmet need, and it is expressed in approximately 1 in 3 PLWRD. Contributing to inequality, marginalization, discrimination and exclusion, and highly impacting the access to health, social and community participation and services, stigma is a likely a key contributor to the lower quality of life reported by PLWRD. The Task Force on Stigma aims to articulate the particular facets of stigma that come with rarity and benefit from the learnings and solutions from other domains in which stigma has been a concerted focus of attention (such as HIV or birth defects).

Objectives

- Identify the State of Play and unlock new knowledge to address stigma to accelerate diagnosis and to enhance equitable care and support;
- Deliver a survey and white paper to identify technology and human factor solutions;
- Guide and accelerate co-designed patient driven research that also harnesses the power of technology for inclusivity and scale.

Task Force Progress

- Kick-off meeting held and monthly teleconferences fixed
- Identification of potential missing expertise in the group – targeted invitations sent to some additional experts
- Review of literature on the topic and of existing frameworks (e.g. for HIV), analysis of health-related stigma at the intersection with race, sexual orientation, culture and religion, visible-invisible conditions; identification of tools

- Contribution to the EURORDIS Rare Barometer Survey on Mental Health & Wellbeing
- Discussions around the areas to be covered and developed; creating the 'RD Stigma framework'
- Timeline of the Task Force in-person workshop

5. Publications

Link: https://irdirc.org/activities/irdirc-publications/

Publication Name	Authors	Journal
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, Daniel O'Connor, Annemieke Aartsma-Rus	Therapeutic Advances in Rare Disease
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
Funding Models Toolbox	Lucia Monaco, Adam Hartman	Resource available on IRDiRC website
Realising the potential impact of artificial intelligence for rare diseases – A framework	Tudor Groza, Chun-Hung Chan, David A. Pearce, Gareth Baynam	Rare 3
What matters ethically about how the UDN has changed since its inception	David A. Pearce, Elena-Alexandra Tataru	AMA Journal of Ethics *(accepted for publishing)*

6. Next meetings and events

In-person Consortium Assembly Meeting

- Dates: 15-16 October, 2025
- Location: Paris, France
- Venue: Plateforme Maladies Rares, 96 Rue Didot, 75014, Paris, France
- Preparation of agenda ongoing - 3 special topics; Confirmed session - Rare disease advancements in China

World Orphan Drug Congress (WODC) Europe

- Dates: 27-29 October, 2025
- Location: Amsterdam, Netherlands
- Venue: RAI Conference Center Amsterdam

Comments from the audience:

Several IRDiRC members, including the IRDiRC vice chair, have worked closely with the WODC organizer for the agenda preparation, suggesting a restructure of the agenda themes and sessions, various topics and speakers. The Scientific Secretariat will have a booth at the congress. WODC will be back-to-back with ERDERA 2nd General Assembly, planned from 29-31 October in Amsterdam, Netherlands.

Clinical Research Networks Conference

- Dates: 9-10 December, 2025
- Location: Heidelberg, Germany
- Venue: Marsilius College
- Invitation-only event
- Event co-organized by ERDERA and Rare Diseases International

7. AOB – No AOB was reported.

Abbreviations

ATMPs - Advanced therapy medicinal products

AAVs – Adeno-associated viruses

c4c – connect for children

CA – Consortium Assembly

CCC – Companies Constituent Committee

CMC – Chemistry, Manufacturing and Controls

DSC – Diagnostics Scientific Committee

ERDERA – European Rare Diseases Research Alliance

FCC – Funders Constituent Committee

FDA – Food and Drug Administration

IHI – Innovative Health Initiative

IRDiRC – International Rare Diseases Research Consortium

ISC – Interdisciplinary Scientific Committee

LMIC – Low-Medium Income Country

NRDD – Nature Reviews Drug Discovery

PACC – Patient Advocacy Constituent Committee

PE – Patient Engagement

PLWRD – Persons Living With a Rare Disease

RD – Rare Disease

RSC – Regulatory Scientific Committee

RWD – Real World Data

SaME – Shared Molecular Etiologies

SC – Scientific Committee

SoC – Standard of Care

TF – Task Force

TSC – Therapies Scientific Committee

UDN – Undiagnosed Disease Network

WG – Working Group

WODC – World Orphan Drug Congress

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