INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the Consortium Assembly Meeting

06-07 December 2023
Online

IRDiRC

ACRONYMS

CA	Consortium Assembly
CCC	Companies Constituent Committee
CORD	Chinese Organization for Rare Disorders
DSC	Diagnostics Scientific Committee
EFPIA	European Federation of Pharmaceutical Industries and Associations
EJMG	European Journal of Medical Genetics
EJP RD	The European Joint Programme on Rare Diseases
EMA	European Medicines Agency
ERDERA	European Rare Diseases Research Alliance
FCC	Funders Constituent Committee
FDA	US Food and Drug Administration
HTA	Health Technology Assessment
Inserm	Institut National de la Santé et de la Recherche Médicale
IRDiRC	International Rare Diseases Research Consortium
ISC	Interdisciplinary Scientific Committee
KOL	Key Opinion Leader
LMICs	Low- and Middle-Income Countries
NBS	Newborn Screening
NIH	US National Institutes of Health
ODDG	Orphan Drug Development Guidebook
PACC	Patient Advocacy Constituent Committee
RD	Rare Disease
RDI	Rare Diseases International
RSC	Regulatory Scientific Committee
TBC	To Be Confirmed
TSC	Therapies Scientific Committee
WODC	World Orphan Drug Congress

MEETING REPORT CONTENTS

THE REPORT

The International Rare Diseases Research Consortium (IRDiRC) held a two-days online meeting of the Consortium Assembly (CA) on 06-07 December 2023, via teleconference. It was attended by 43 online participants representing 26 member organizations, Chairs and Vice Chairs of the Scientific Committees, and members of the Scientific Secretariat.

1. Introduction and Welcoming

- **Summary of Montreal Consortium Assembly Meeting**

  The Consortium Assembly and Scientific Committee members convened in Montreal on 03-04 October 2023, for a significant event that drew the participation of 37 in-person attendees and 20 members who joined online. This meeting provided a platform for committee members to conduct both individual and cross-committee sessions, fostering collaboration and exchange of ideas. Comprehensive updates on the activities of IRDiRC were presented during the event. Two featured sessions centered on the following topics were held: "Advancing RD Research and Development through Public-Private Partnerships" chaired by Samantha Parker and "Revolutionizing RD Research: Harnessing the Power of Artificial Intelligence" chaired by Gareth Baynam. Additionally, a keynote presentation by Anne Pariser and Elizabeth McNeil titled "FDA, NIH & Industry: Lessons learned along the way" enriched the program. The event served as a forum for discussions and initiatives aimed at advancing RD research and development.

- **Summary of IRDiRC participation at the World Orphan Drug Congress in Barcelona (30 October – 02 November 2023)**

  Twenty-two members of IRDiRC Consortium Assembly, Scientific Committee, and Task Force were invited to present or be involved in a panel discussion in 31 sessions, providing valuable insights and fostering collaborative discussions with the congress attendees. In addition, IRDiRC further extended its presence at the congress through an exhibition booth. The booth served as a hub for engaging with diverse audience of the congress, attracting 56 congress attendees who signed up for newsletters or indicating interest in becoming an IRDiRC member (either through the Consortium Assembly, Scientific Committee, or Task Force).

- **New Members**
  - CCC: **UCB**, represented by **Gabriella Almberg**, Global Head Policy & Public Affairs, Rare Disease Organisation, Denmark

- **Change of Representation**
  - PACC: **Charlene Son Rigby**, CEO Global Genes, replaced **Maria Della Rocca**, Senior Director Support & Education Programs, USA

2. Updates from the IRDiRC Constituent Committees

- **Funders Constituent Committee (FCC)**

The FCC has been actively engaged in various initiatives. The Chrysalis Task Force, which has been completed, successfully identified key criteria to enhance the attractiveness of RD research for industry in terms of both research and development. Similarly, the completed Primary Care Task Force sought to unite representatives from diverse stakeholders to pinpoint priority research areas, challenges, and opportunities in RD research within primary care. The Enabling and Enhancing Telehealth for RD Across the Globe Task Force, also concluded, which conducted KOL interviews and systematic review of existing telehealth models, addressing barriers and opportunities, and formulating best practices for the integration of telehealth services into communities. The ongoing Funding Models to Support the Spectrum of RD Research & Development Task Force endeavours to understand the decision-making processes of different funders at various stages in a treatment's development.

Looking ahead to 2024, Christopher McMaster, Chair of the FCC, mentioned there is potential for a new Task Force focusing on moving RD medicines to first in human trials. It was raised that there are inadequacies of regulatory systems in the context of RDs, and that existing regulatory processes in various countries, including the FDA, EMA, Health Canada, and others, are primarily designed for more common diseases that involve small-molecule drugs manufactured on a large scale. The traditional regulatory approval process, which often requires large-scale double-blind placebo-controlled trials, does not seem to be practical for RDs with limited patient populations. Additionally, the cost and administrative burden of keeping a medicine on the registry in Europe are issues that challenge the development and approval of therapies for RDs. It was suggested that the regulatory sciences and processes need to be adapted to the unique challenges of RD drugs. This discussion highlights the need for changes in the regulatory framework to better serve RD patients and encourages alignment between regulatory agencies to address these issues on a global scale. A joint Task Force is foreseen which acknowledges that current HTA approval regulations may not be suitable for RD therapies and aims to explore reciprocity in RD medicine approval between HTAs. Additionally, it addresses manufacturing approvals for essentially N-of-1 (or N-of-many) medicines, collaborating with Regulatory Scientific Committees, and potentially with the Companies Constituent Committee.

Another prospective initiative by the FCC for 2024 is centered on Rare Diseases (RD) in Low- and Middle-Income Countries (LMICs), with a specific emphasis on exploring the utility of the iHOPE Illumina initiative for LMICs and Indigenous peoples worldwide. Ryan Taft, Vice President of Scientific Research at Illumina, presented the iHope philanthropic program to the FCC members, a project initiated by Genetic Alliance and Illumina, during an FCC parallel session in Montreal. The program has two primary objectives: (1) to provide clinical whole genome sequencing to as many patients as possible each year, and (2) to assess

the program's impact across different geographic regions and income levels. He emphasized the importance of democratizing whole genome sequencing globally, not limiting it to only a small percentage of individuals who can afford it.

Lastly, the FCC presented to the Consortium Assembly a survey on their top five funding opportunities and priorities in the past and next three years, including allocation of funding opportunities and interest in co-programming some research with other FCC members. The survey will be re-launched within the FCC to collect more data. The copy of the survey results is available only to the Consortium Assembly members via the Scientific Secretariat.

- **Companies Constituent Committee (CCC)**

2023 – A Year of Change for the Companies Constituent Committee (CCC) that redefined the role of CCC in IRDiRC and its members expectations. Along the year, three new members joined the CCC: The European Federation of Pharmaceutical Industries and Associations (EFPIA) in February, Biogen in June, and UCB in November. The committee worked on defining the priorities for the next months and preliminary discussions took place on the execution aspects.

The mission of the companies in IRDiRC is to represent diagnostic and therapy developers' perspectives, but also to be a solid partner and sounding board to the other IRDiRC committees, and to ensure that a broad representation of pharma and biotech perspectives on RDs are aligned with the rest. Another goal of CCC is to identify and share with IRDiRC the issues faced by therapeutic developers, at the same time highlighting the challengers in the RD research ecosystem that are roadblocks to the achievements of IRDiRC goals from the industry perspective.

With the new leadership changes, a set of priorities have been set up to guide the group, among them: public-private partnerships, global convergence, and contribution to dissemination and implementation of existing solutions recommended by IRDiRC outputs, as well as improving the patient access to trials, diagnosis, treatments, improvement of natural history of disease and validation of biomarkers and clinical endpoints.

- **Patient Advocates Constituent Committee (PACC)**

Members discussed how PACC can play a better role in engaging patients/patient groups into IRDiRC's activities and facilitate the sharing and the understanding of IRDiRC outcome activities within the patient community.

The proposed actions include:

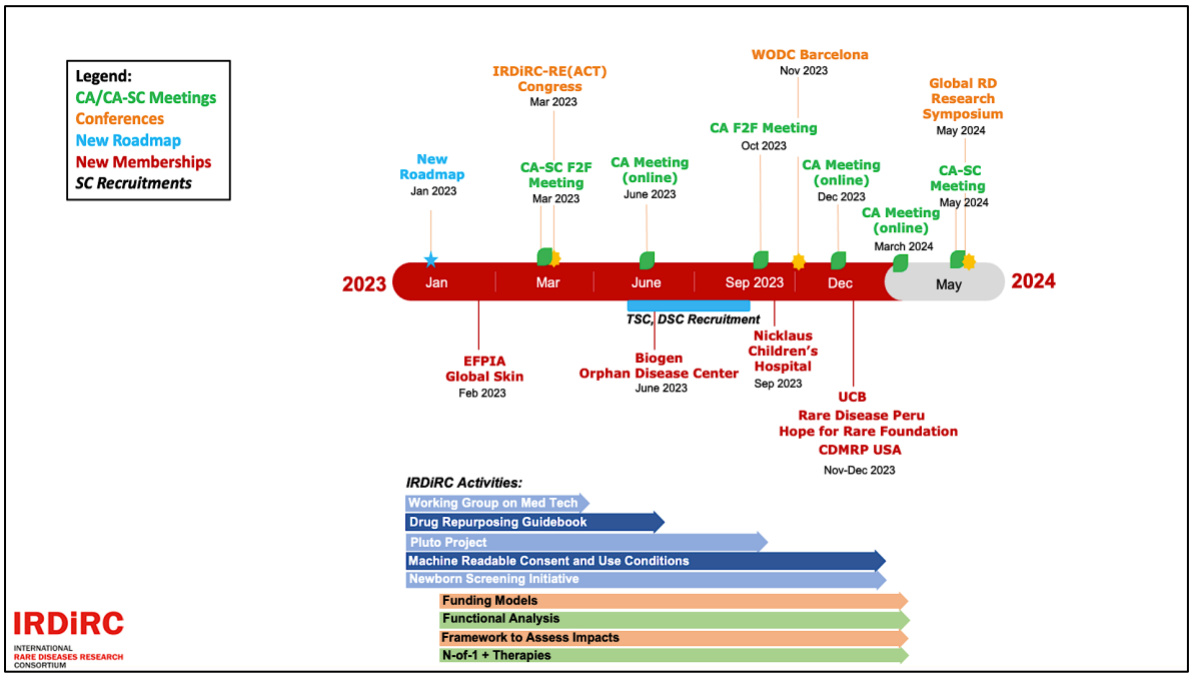
- Invite members of PACC to present their organization and what they do in research.
- Invite members of Task Forces and other constituencies in PACC meetings to present their work.
- Summarize and provide feedback on the outcomes of IRDiRC activities to facilitate their uptake and understanding by the global patient community.

The members have also clarified the mission of the PACC which is to improve value of RD research through active participation of patient community. The mission includes three main points:

- Understand what is important to patients and bring it to IRDiRC to inform our research.
- Translate research findings into real benefits for the patients.
- Develop a life cycle approach from diagnosis to therapies and care follow-up.

The development of patient data platform to capture real world data (natural history studies, patient-reported outcomes, clinicians entering data, etc.) for evidence generation for HTAs and to facilitate access to medicine was discussed and recognized as of significant importance.

3. Annual Overview of IRDiRC Activities and Publications in 2023



➢ Task Forces and Working Groups

- **Preparing for Genetic N-of-1 Treatments of Patients with Ultra-Rare Mutations**

  Objectives: To connect different N-of-1 efforts to reduce duplication, achieve global consensus, and create a roadmap towards development and implementation of N-of-1 treatment; to raise awareness of the N-of-1 concept and challenges with all stakeholders, and identify major challenges hampering N-of-1 therapy development and timely patient access, which can potentially

lead to development of proposed solutions and create better opportunities for strategic planning and delivery.

Planned Output: Two White Papers on (1) State-of-the-Art paper, including the N-of-1 roadmap, and (2) Future perspectives for N-of-1 therapies.

Progress Update: Seven teleconferences with the group took place in 2023 and an in-person workshop in Barcelona (Spain) that was attended by 18 members out of the 22.

- Compilation of the current literature resources on N-of-1.
- State of the Art paper – under internal review to incorporate the N-of-1 therapy roadmap, per feedback received from the journal, planned for Q1 2024.
- Second paper on future perspectives is planned in Q2 2024.
- A curated list of educational resources available on IRDiRC website.

- **Operationalizing a Comprehensive Framework to Assess the Impacts of Diagnoses and Therapies in RD Patients**

Objectives: To develop, operationalize, and test a comprehensive framework of holistic, multidimensional, and evolving life-long experiences of patients and families living with a RD, (derived from or leading to a natural history study); to develop, operationalize, and validate multidimensional indicators and measures (qualitative and quantitative) of impacts associated with diagnosis, treatment, support, and community integration that can be used to capture changes along the patient "journey"; and to investigate qualitative case studies to represent a number of parameters that could inform on impacts

Expected Output: Framework to assess impacts; White Paper**.**

Progress Update: The group started a literature review search to identify the elements of the framework. Four clusters have been identified and are currently investigated by the Task Force members: clinical impacts, care impacts, socio-economic impacts, and RD ecosystem impacts.

- Clinical Impacts: Diagnostic, Disease knowledge, treatment
- Care Impacts: Mental health, Care coordination, Socio-cultural determinants
- Socio-economic Impacts: Economic determinants, Socio-economic determinants, Informed burden of disease.
- RD Ecosystem impacts: Patient group support, RD ecosystem & landscape, Patient and family roles and expectations

The aim is to identify the role of the patients and the families, identify gaps, and select quantitative indices and qualitative measures that can be mapped

on the patient pathway. Currently investigating the literature for each cluster to identify measures of impacts, gaps, and map them on the patient pathway.

- **Functional Analysis**

  Objectives: To further development, standardization, and quality improvement of the experimental and computational methods of functional assays ; to foster ecosystem building, infrastructure development and partnerships for the effective chain from fundamental research to clinical applications of functional assays; and to foster equity in RD diagnostics and treatment through the application of indiscriminative multiplexed assays of variant effect and variant effect maps to the fundamental research and clinical practice in RDs.

  Planned Output: Framework for the robust and effective ecosystem of functional analysis in RDs; White Paper.

  Progress Update: The group has established a framework that will serve as the foundation of their future white paper, including step-by-step diagram of functional analysis that involves education, policy, and sustainability. Simultaneously, preparations are underway for an in-person Task Force workshop in 11-12 March 2024 in Paris, France.

- **Funding Models to Support the Spectrum of RD Research and Development**

  Objectives: To identify key motivating factors for different types of funders of RD research and how different types of funders decide at which point in a research study's lifecycle they will provide support; identify the key influencing factors for effective public-private partnerships at different stages of a treatment's life cycle and models of public-private partnerships, including means of sharing information (with attention to tech transfer issues and regulatory requirements).

  Planned Output: Summary Paper of Findings and Funding Model/Good Practices Toolbox

  Progress Update: The group has successfully completed a literature review and conducted online interviews with 21 selected key opinion leaders which help obtain insights into research funding strategies and decision-making processes. Analysis of collected data and review of interview summaries are currently being performed. Simultaneously, an in-depth analysis of a database containing orphan drug designations from EMA and FDA is also ongoing. This analysis includes an examination of the number of products in the preclinical stage at the time of designation, the progression of these into clinical trials, reasons for product failure, the distribution of public and private grants across different development stages, and the involvement of small or medium-sized companies and academic researchers in partnerships with larger pharma/biotech entities.

Finalization of the work and writing of the manuscript is foreseen in Q1-Q2 2024.

- **Newborn Screening Initiative**

Two special editions in the Rare Disease and Orphan Drugs Journal.

Theme 1 - **Real World Applications and Technologies** (associate editor: Virginie Bros-Facer):

1) A systematic review of real-world applications of genome sequencing for newborn screening (systematic review) – *published.*
2) Federated databases: An approach to enhance secure data sharing in newborn genomic screening (opinion paper) – *published.*
3) Next-Generation Sequencing-Based Newborn Screening Initiatives in Europe: An Overview (original research) – *published.*
4) Enhancing the Efficiency and Efficacy of Newborn Screening by using Real-World Data and relevant technologies – *under revision by authors following Journal's peer-review.*
5) Towards genomics in Newborn Screening for Inherited Metabolic Disorders: Expert opinion from IRDiRC-Newborn Screening Initiative – *submitted and under review by the journal.*
6) Incorporating a new disease in the Newborn Screening programmes in Europe: The Spinal Muscular Atrophy case study (opinion paper) – *final draft under review before submission.*
7) Newborn Screening in Mexico: Present and Future (perspective paper) – *draft in development.*

Theme 2 - **Policy, Ethics and Patient Perspectives** (associate editors: Helen Malherbe & Mary Wang):

1) Introductory Editorial – *to be drafted during review process.*
2) More evidence is not enough to reconcile equity and efficiency in neonatal screening policies: Analysis of the trend in Spain, 2003-2022 – *published.*
3) The Australian landscape of newborn screening in the genomics era – *published*.
4) Patient advocacy in action for Newborn Screening: role of patient organizations in the timely diagnosis of RDs and quality of life – *under revision by authors following Journal's peer-review*.
5) Overcoming challenges in sustaining NBS: the Philippine model – *published.*
6) Newborn screening in South Africa – *submitted and under review by the journal.*
7) NBS in the rural community – *submitted and under review by the journal.*

- **Parallels of COVID-19 and RD**

  Established during the 2023 CA-SC Conference in Berlin, Germany, the initiative is comprised of IRDiRC members Gareth Baynam (ISC), PJ Brooks (FCC), and Anne Pariser (RSC). The primary goal of this collaborative effort is to publish a position paper that draws parallels between the expedited response to the COVID-19 pandemic, particularly the "Operation Warp Speed" initiative, and the ongoing but often overlooked silent epidemic of RDs. The group has set clear goals, and outlined the content of the manuscript. The first draft has been completed, with the team currently engaged in the process of refining and editing through videoconferences and email communication. Anticipated to bring attention to the ongoing emergency in the realm of RDs, the position paper is expected to be finalized in the first or second quarter of 2024, with the target journal for publication yet to be determined.

- **Demystifying Gene Therapies**

  The information on gene therapies submitted by the session participants in Berlin (Germany) was collected and collated. A review article summarizes this knowledge was submitted to the European Journal of Medical Genetics (EJMG) for the IRDiRC Special Issue as review article.

- **IRDiRC Publications**

  Published:

  - **Drug Repurposing for Rare: Progress and Opportunities for the Rare Disease Community**. Anneliene Hechtelt Jonker, Daniel O'Connor, Maria Cavaller-Bellaubi, Christine Fetro, Maria Gogou, Peter A.C. 't Hoen, Martin de Kort, Heather Stone, Nivedita Valentine, Anna Maria Gerdina Pasmooij. Frontiers in Medicine. Access the paper here.

  - **IRDiRC Drug Repurposing Guidebook: Making Better Use of Existing Drugs to Tackle RDs.** Anneliene Jonker, Simon Day, Michaela Gabaldo, Heather Stone, Martin de Kort, Daniel J. O'Connor & Anna Maria Gerdina Pasmooij.
    Access the paper here.

  - **Defining Rare Conditions in the Era of Personalized Medicine.** Daniel J. O'Connor, Michela Gabaldo, Annemieke Aartsma-Rus and Anneliene Hechtelt Jonker. Access the paper here.

  - **The IRDiRC Chrysalis Task Force: Making Rare Disease Research Attractive to Companies.** Katherine L. Beaverson, Daria Julkowska, Mary Catherine V. Letinturier, Annemieke Aartsma-Rus, Jennifer Austin, Juan Bueren, Simon Frost, Misako Hamamura, Jane Larkindale, Greg LaRosa, Rita Magenheim, Annamaria Merico, Anna Maria Gerdina Pasmooij, Vinciane Pirard, Nicholas

Ekow Thomford, Michihiko Wada, Durhane Wong-Rieger, and Adam L. Hartman. Access the publication here.

- **How to START? Four Pillars to Optimally Begin Your Orphan Drug Development**. Anneliene Hechtelt Jonker, Liliana Batista, Michela Gabaldo, Virginie Hivert & Diego Ardigo on behalf of the IRDiRC ODDG TF and IRDiRC TSC. Access the paper here.

- **Targeting Shared Molecular Etiologies to Accelerate Drug Development for Rare Diseases.** Galliano Zanello, Macarena Garrido-Estepa, Ana Crespo, Daniel O'Connor, Rima Nabbout, Christina Waters, Anthony Hall, Maurizio Taglialatela, Chun-Hung Chan, David A Pearce, Marc Dooms, Philip John Brooks. Access the paper here.

- **Towards the International Interoperability of Clinical Research Networks for Rare Diseases: Recommendations from the IRDiRC Task Force**. Rima Nabbout, Galliano Zanello, Dixie Baker, Lora Black, Isabella Brambilla, Orion J.Buske, Laurie S. Conklin, Elin Haf Davies, Daria Julkowska, Yeonju Kim, Thomas Klopstock, Harumasa Nakamura, Kim G. Nielsen, Anne R. Pariser, Jose Carlos Pastor, Maurizio Scarpa, Maureen Smith, Domenica Taruscio, Stephen Groft. *Orphanet Journal of Rare Diseases, 2023.* Access the paper here.

- **Sustainable Approaches for Drug Repurposing in RD: Recommendations from the IRDiRC Task Force**. Galliano Zanello, Diego Ardigò, Florence Guillot, Anneliene H. Jonker, Oxana Iliach, Hervé Nabarette, Daniel O'Connor, Virginie Hivert. *Rare Disease and Orphan Drugs Journal, 2023*. Full text available here.

In Preparation or Under Review:

- **Indigenous Population: "**Advancing rare genetic diseases diagnosis and research for indigenous people" – *Manuscript accepted in Nature Genetics for publication.*

- **IRDiRC-RDI Global Access Working Group Part II: "**Barriers to access essential medicines for rare diseases – Case Studies" – *Manuscript under review.*

- **Primary Care: "**Global Health for Rare Diseases through Primary Care" – *Manuscript under review.*

- **Integrating New Technologies for RD Diagnosis**: "Leaving no patient behind! Expert recommendations for the use of innovative technologies in the diagnosis of rare diseases" – *Manuscript under review*.

- **Enabling and Enhancing Telehealth for RDs Across the Globe: "**Telehealth for rare disease care, research and education across the globe: a review of the literature by the Telehealth Task Force" – *Manuscript under review.*

- **Preparing for genetic N-of-1 treatments of patients with ultra-rare mutations:** State-of-Art review on N-of-1 therapies with a focus on genetic therapies and illustrative cases when it was applied – *Manuscript under review.*

- **Machine readable consent and use conditions:** "Getting Your DUCs in a Row – Standardising the Representation of Digital Use Conditions": using Digital Use Conditions as a framework that facilitates advancing scientific discovery and sharing of research data, healthcare records, biosamples, and biomedical resources – *Manuscript under review*.

- **Machine readable consent and use conditions:** "Common Conditions of Use Elements. Atomic Concepts for Consistent and Effective Information Governance": standardized digital representation of diverse use conditions – *Manuscript under review.*

- **Medical Technologies for RD in Orphanet Journal:** "Developing orphan medical devices for use in rare diseases: a landscape overview".

- **Pluto Project on Under-Researched RD.** – *Manuscript under preparation.*

- **Interoperability of Clinical Research Networks.** – *Manuscript under preparation.*

- **Special Issue Collaboration with the European Journal of Medical Genetics:** Special bundle issue, nine articles were submitted in October 2023. – *Manuscript under review.*

4. Scientific Committees Updates

- **Diagnostics Scientific Committee (DSC)**

  The group worked on a white paper titled "Addressing Diagnostic Gaps and Priorities of the Global Rare Diseases Community: Recommendations from the IRDiRC Diagnostics Scientific Committee" and submitted it to the European Journal of Medical Genetics for the IRDiRC Special Issue. The white paper covers clinical aspects, laboratory testing and technology, ongoing care and transition to therapy. DSC is planning a joint task force proposal with the TSC for the IRDiRC Roadmap 2025. Quarterly online meetings are planned for 2024.

- **Therapies Scientific Committee (TSC)**
  The TSC is preparing a task force with the aim of developing consolidated guidance for small population research, potentially building on the SPIRIT items. Additionally, they plan to set up a working group with the aim of optimizing patient engagement, specifically in the development of rare paediatric therapies. Other ongoing initiatives they would like to organize include the development of guidelines for digital biomarkers, with a paper to be drafted by a small subgroup. Similarly, another subgroup will draft a paper on patient-relevant regulatory-endorsed endpoints. Collaboration is encouraged in the realm of drug-device combinations, where the task force plans to join forces with the ISC. Furthermore, the integration of artificial intelligence into therapy development is being explored. Notably, certain topics are currently under consideration, including work in the pre-competitive space and discussions related to health technology assessment (HTA) and the health economics of therapy development.

- **Regulatory Scientific Committee (RSC)**
  The RSC was established last year and has successfully gained momentum. They worked on defining their mission and spent the first half of 2023 shaping their strategic vision while developing short and midterm action items. RSC's initial focus involved conducting a landscape analysis of orphan and rare product approvals from the past few years, recognizing the significant therapeutic changes during this period. Gathering data has been time-consuming but enlightening, with core information collected from some regions and the remaining ones still being completed. RSC's goal is to draft and submit a paper in early 2024 addressing issues such as regulatory approval reciprocity and the global nature of these trials, leading to discussions on streamlining processes. Identifying case examples and considerations for upskilling regulators emerged as crucial points for discussion. Excitingly, RSC hopes to leverage the lessons learned from the oncology field to propose solutions and best practices. Looking ahead, the RSC plans to engage in discussions about manufacturing and regulatory flexibility in 2024, seeking collaboration and partnerships with other committees, such as the FCC, as we continue our work toward advancing the field.

- **Interdisciplinary Scientific Committee (ISC)**
  In 2023, ISC had six online meetings that highlighted their participation in different conferences, such as the World Orphan Drug Congress, the European Hemophilia Consortium Conference, and other notable gatherings. Gareth Baynam, ISC Chair, chaired a session at the Montreal meeting centered around "Revolutionizing RD Research: Harnessing the Power of Artificial Intelligence." Additionally, he holds the first authorship position on a publication addressing the stigma associated with genetic testing, as well as a submitted commentary. ISC's planned activities in 2024 involve further exploration with the members how artificial intelligence can be applied to enhance RD research in care and living experiences. ISC aims to delve into addressing stigma as an integral aspect of the overall life experience of individuals with RD. Furthermore, the ISC is actively exploring a potential

collaboration with ERN-EuroBloodNet for the development of a publication focusing on basket trials for hematological disorders.

5. Scientific Secretariat Updates

- **Upcoming IRDiRC Meetings**
  - 06-07 March 2024: Online Consortium Assembly Meeting
  - 22-23 May 2024: In person Consortium Assembly Meeting in Shanghai (China)
    - A 1.5-day meeting back-to-back with the China RD Research and Translational Medicine Annual Conference.
    - Meeting organization will be supported by the Chinese Organization for Rare Disorders (CORD), Hope for Rare Foundation, and Fudan University Children's Hospital.
  - 16-17 October 2024 (TBC): In person Consortium Assembly Meeting in Milan, Italy, hosted by the Fondazione Telethon.
- **Task Force Timeline in 2024**

  Collection of Task Force proposals is planned for September-October 2024.
- **Future of the Scientific Secretariat**

  Starting September 2024, and for a period of 7 years, the Scientific Secretariat will be supported by the European Commission through the European Rare Diseases Research Alliance (ERDERA) programme. The European Commission will contribute 2.4 million over 7 years.

  The Scientific Secretariat will be hosted by the Foundation for Rare Diseases (France) and will be coordinated by Prof. Daniel Scherman.

  The contribution of ERDERA will be reflected in:
  - Supporting the salary cost for two project managers.
  - Funding of one CA-SC meeting per year. Another CA meeting will be supported with the help of other member organizations.
  - Four annual Task Forces will be launched on yearly basis and two in person workshops will be funded.
  - The communication activities will be partially supported by the ERDERA communication manager together with the help of the two project managers.

6. Communication Sub-Committee Updates

The objective of this group is to improve the visibility of IRDiRC for internal and external stakeholders to have a credible and trusted communication approach, at the same time highlighting the impactful changes that IRDiRC produces.

Outputs:

- Creation of communication slide deck for external events.
- Review of the current website and create a list of suggested changes for improvement.

Next Steps:

- Complete the IRDiRC slide deck with the Task Forces information and restructuring of the available tools in a comprehensive way.
- Ongoing review of the website structure and content – *planned for Q1 2024.*

7. References Shared by Meeting Participants During the Meeting

- Genomics England, the Medicines and Healthcare products Regulatory Agency (MHRA), Oxford-Harrington Rare Disease Centre, Mila's Miracle Foundation and the Association of the British Pharmaceutical Industry (ABPI) launched a pilot to support children with rare conditions to access personalized therapies: https://www.genomicsengland.co.uk/news/pilot-launched-to-support-children-with-rare-conditions-to-access-personalised-therapies#:~:text=The%20Rare%20Therapies%20Launch%20Pad%20aims%20to%20do%20more%20for,for%20them%2C%20quickly%20and%20safely.
- Nature Article: Regulatory Watch – The Orphan Drug Pipeline in Europe: https://www.nature.com/articles/nrd.2016.96.pdf
- Human Gene Therapy Article: The Lived Experience of Pediatric Gene Therapy – A Scoping Review: https://www.liebertpub.com/doi/10.1089/hum.2023.157
- Project HERCULES (Health Research Collaboration United in Leading Evidence Synthesis) by Duchenne UK: https://www.duchenneuk.org/project-hercules/
- Health Technology Assessment (HTA) International on Rare Diseases: https://htai.org/interest-groups/rare-diseases/
- Innovative Genomics Institute's Report on "Making Genetic Therapies Affordable and Accessible": https://innovativegenomics.org/atf-report/