Meeting Report

Joint IRDiRC Consortium Assembly and Scientific Committees Meeting

22-23 May 2024
Shanghai, China

ACRONYMS

ANMAT	National Administration on Drugs, Foods, and Medical Devices
ATMP	Advanced Therapy Medicinal Product
CA	Consortium Assembly
CCC	Companies Constituent Committee
DSC	Diagnostics Scientific Committee
DUC	Digital Use Conditions
ECRD	European Conference on Rare Diseases
EFPIA	The European Federation of Pharmaceutical Industries and Associations
EHC	European Haemophilia Consortium
EJP RD	European Joint Programme on Rare Diseases
EMA	European Medicines Agency
ERDERA	The European Rare Diseases Research Alliance
ERN	European Reference Network
FCC	Funders Constituent Committee
FDA	US Food and Drug Administration
KPIs	Key Performance Indicators
FEPER	Federación Peruana Enfermedades Raras
ICORD	International Collaboration on Rare Diseases and Orphan Drugs
INSERM	French National Institute of Health and Medical Research
IRDiRC	International Rare Diseases Research Consortium
ISC	Interdisciplinary Scientific Committee
PACC	Patient Advocacy Constituent Committee
PE	Patient Engagement
PLWRD	Persons Living with a Rare Disease
PPP	Public-Private Partnership
QoL	Quality of Life
RD	Rare Disease
RDI	Rare Disease International
RSC	Regulatory Scientific Committee
RD Moonshot	Rare Disease Moonshot
SC	Scientific Committees
SciSec	IRDiRC Scientific Secretariat
TF	Task Force
TSC	Therapies Scientific Committee
UDNI	Undiagnosed Diseases Network International
WODC	World Orphan Drug Congress

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THE REPORT

The International Rare Diseases Research Consortium (IRDiRC) recently held a 1.5 day of hybrid meeting for the Joint IRDiRC Consortium Assembly (CA) and Scientific Committees (SC) Meeting on May 22-23, 2024, in Shanghai, China, in partnership with **Hope for Rare Foundation (China)** and **Fudan University (China)**. The meeting took place through a combination of teleconference and face-to-face in Shanghai. A total of 9 participants representing CA and SC members attended online, while 33 participants representing 17 members of the CA, 7 members of the DSC, 4 members of the TSC, 1 member of the RSC, 3 members of the ISC, 1 member of the Scientific Secretariat (Sci Sec).

1. Presentation of New IRDiRC Members, Leaders, and Representatives

IRDiRC is pleased to announce the addition of new members and representatives to the Consortium. We extend a warm welcome to all.

- **New Members of IRDiRC Constituent Committee (1)**
  - **Centre-Alliance for Rare Disease in Rwanda**
    - Official Representative: Manzi Ndamukunze, Chairman and Executive Director, Rwanda

- **New Members of IRDiRC Scientific Committees (4)**
  - **ISC**
    - Chiu-Hui Mary Wang – Global Programme Director at Rare Disease International (RDI), Italy
    - Elmira Safarova - Founder and CEO Rarus Health Spa, Chile
    - Robert Allaway – Principal Scientist, Sage Bionetworks, USA
    - Ritu Jain – Founder DEBRA International, Singapore

2. Scientific Committees Parallel Session - *discussion updates included at Point 3*

3. Constituent Committee and Scientific Committees Reporting

Constituent Committees

- **Funders Constituent Committee Updates**

  - The FCC met several times during the first six months of 2024 through teleconferences. Among the topics brought to attention were the Task Force proposal preparation on

Regulatory Convergence, and the current challenges associated with the data collection and processing for the funding database (POLARIS OS).

- **New Task Force Proposal for 2024:**

  Moving rare disease medicines to first in human trials – the current approval regulations are not fit-for-purpose for therapies designed for ultra-rare diseases, especially in the case of ATMPs. In consequence, it is important to explore and convene on RD medicine approval reciprocity between different regulatory bodies. Moreover, the current manufacturing approvals are not adequate for ATMPs for N-of-1 (or "N-of-few" medicines). The proposal is planned to be co-developed with the Regulatory Scientific Committee and Companies Constituent Committee.

- **Other discussions:** AFM Téléthon France representative mentioned that the organization funds projects of 100 million euros on annual basis. One of the members of the Therapies Scientific Committee mentioned that funding for RD projects could come also from patient advocacy organizations through funds raising.

➢ **Patient Advocacy Constituent Committee Updates**

- **New PACC Member:** Federación Peruana Enfermedades Raras (FEPER), represented by Pilar Estremadoyro Reyes, Vice President of the organization, based in Peru, Latin America.
- **Change of representation for Global Genes:** Charlene Son Rigby, CEO, based in USA, is the new IRDiRC representative for Global Genes.
- **Next actions planned for PACC**
  The Patient Advocacy Committee expressed their interest in inviting the members of PACC to present their affiliated organization research impact in rare diseases and the areas of research interest. To further improve interactions and work with the other committees, PACC plans to invite Task Force representatives to present their project's results, as well as members of the other constituencies to display some of the projects developed with the goal to facilitate the uptake and understanding the outcomes of IRDiRC activities by the global patient community.
- **Other Discussions:** A PACC representative brought up the issue that for some PACC members it can be difficult an active participation to IRDiRC meetings due to the complexity of the work discussed, on this basis, it is important to ensure translations of different outputs in an accessible manner, including a broader dissemination of results with local patient organizations. It was proposed that PACC might be the right committee to help with the drafting of such documents and their dissemination through different communication channels. Another PACC member representative expressed her concern if it would be possible to obtain more concrete outcomes from IRDiRC work applying a shorter-term view, which is more aligned with the patients' expectations in terms of timelines. Among the topics that could raise interest of patients or other

patient advocacy groups to join the consortium would be how the acceleration of therapies development can be improved and understand their motivation for joining a global organization such as IRDiRC. One possibility to better understand the patients' needs would be to encourage them to participate in different RD congresses sessions to openly express their needs. FCC representative of Fondazione Telethon Italy mentioned that PACC can help in identifying the projects to be prioritized for funding, without following a country-based approach. Another FCC member mentioned that a solution for helping PACC on long-term basis would be setting of services that can offer consultancy services. Capturing the patients' needs could also be done through surveys, another PACC representative underlined. The representative of Child and Youth Care Zimbabwe specified that the connection with IRDiRC can be bringing back the learnings from the work performed in the consortium in one's own community in a simplified, visually focused concrete content, explaining the knowledge and results obtained.

- **Companies Constituent Committee updates**

  - In the second quarter of 2024, the Companies Constituent Committee reported the progress on the ongoing projects developed within the committee.

    a) **Public-Private Partnership (PPP) Project** – with The European Federation of Pharmaceutical Industries and Associations (EFPIA) and Rare Disease Moonshot (RD Moonshot) – For the development of this project, the literature review has been completed and multiple interviews were taken with several IRDiRC members and members of the RD Moonshot group. In addition, 10 case studies were identified and consolidated. In the second stage of the project, in-depth interviews will be compiled in a report, which is planned to be delivered and presented at the Consortium Assembly Meeting in Milan, Italy (16-17 October, 2024).

    b) **Global Convergence** – the preparation of the Global Convergence Task Force is ongoing together with FCC and RSC.

    c) **Basket trials for rare bleeding disorders** – the companies constituent committee is actively involved in the development of basket trials for rare bleeding disorders project in partnership with the European Haemophilia Consortium (EHC), the European Reference Network (ERN) EuroBloodNet (focused on haematological diseases) and several external clinicians.

Scientific Committees

- **Diagnostics Scientific Committee updates**

  - The Diagnostics Scientific Committee has actively worked on the preparation of the Task Force proposal on **Linking Diagnosis to Therapy**, in collaboration with the Therapies Scientific Committee. An initial draft has been developed and distributed to DSC and TSC members for comments and input. Additional important points were discussed during the DSC parallel session in Shanghai. The next steps discussed is to gather any additional input via email and present it to the IRDiRC leadership in late summer or early autumn.

- **Future of DSC** – the committee has discussed what are the barriers for achieving IRDiRC goal 1, including the barriers that are marked differently among settings depending on geography, income level and resource availability. Another question has been raised if the committee should shift their focus on equity and access. Among other topics discussed were:
  - For undiagnosable individuals, are limitations driven by resource scarcity, communication limitations or other factors?
  - Where genome, exome and other advanced testing are available, what would be the next steps for evolution towards clinically unrevealing tests?
  - Phenotyping – has not kept the pace with gene matching. The resources and standards for natural history studies are lacking, which impairs the diagnosis and therapy development. However, there are broad opportunities to mine medical records, support reserves phenotyping and expand the ontologies to include longitudinal data.
  - Diagnosis to therapy – what are the emerging therapies that will be critical for addressing future needs and what diagnosis methods can be mapped to those? Diagnosticians need to be empowered to answer, "Now what?" following a successful diagnosis.
- **Comments:** several members underlined the problem of misdiagnosis, which is being underreported and in consequence is more difficult to capture in real setting. Another FCC representative pointed out that in South and Eastern Europe a fast diagnosis is very rare as there are key factors that delay the process, while at the same time a faster diagnosis is not always a correct one. ISC vice chair raised the importance of acting up as a community to better showcase the "red flags" linked to misdiagnosis.

➢ **Therapies Scientific Committee Updates**

- **Task Force proposal:** TSC discussed about the development of a consolidated methodological guidance for small population research with a focus on analyzing existing resources to baseline and consolidating new or enhancing existing tools to improve small population research (e.g. creating proposals for 'SPIRIT' rare extension).
- Discussions for a Working Group to **optimize patient engagement in rare paediatric therapy development** are ongoing in partnership with the Patient Advocacy Constituent Committee.
- **Additional topics considered for development:** joint collaboration with DSC for exploring areas of common interest.
- **Topics under development/planned for the close future:**
  - **Digital endpoints –** working on creating a white paper focused on the current landscape and emerging perspectives in the application of digital endpoints in rare diseases (activity led by Rajesh Krishna & Dan O'Connor);
  - **Regulatory endorsed patient relevant endpoints** – aim to showcase the current landscape (subgroup let by Michelle Farrar & Anneliene Jonker);
  - **Drug-device combinations –** working on an overview paper focused on the current landscape, definitions and incorporation of several case studies; activity developed together with ISC with a deadline for paper

submission to journal July 2024 (subgroup led by Marc Dooms & Anneliene Jonker).

- **Regulatory Scientific Committee Updates**
  - **Summary:** The committee has worked on increasing their regional representation and organized a membership call for 2 positions in the Q2 of 2024, with a special focus on geographical areas such as Australia, South-East Asia, Africa, South America. The call received 16 applications: 4 from Africa, 7 from South America, 3 from Australia, and one from Europe and from North America.*

    **Two members were selected after the CA-SC meeting took place: Claudia Saidman, Director of Clinical Research and Drug Authorization at National Administration on Drugs, Foods, and Medical Devices (ANMAT), Argentina, and Marco Rizzi, Associate Professor of Law at University of Australia.*

  - **Work Group proposal:** Together with the Funders' Committee, RSC has been investigating the topic of complex manufacturing, an area which is expensive and considered technically difficult. The two groups have discussed where flexibility in manufacturing, regulatory oversight, and inspections can be applied.

- **Interdisciplinary Scientific Committee Updates**

  - The Interdisciplinary Scientific Committee has 4 end of mandate terminations in Q2 2024. Due to this reason a call for new members was launched in April and 4 new members were recruited in the second quarter:

    - Mary Wang – Rare Diseases International, based in Italy
    - Ritu Jain – DEBRA International, based in Singapore
    - Robert Allaway – Sage Bionetworks, based in USA
    - Elmira Safarova – Rarus Health Spa, Chile

  - The committee has been working on two publications, one on Basket trials for rare bleeding disorder, in partnership with the **European Haemophilia Consortium** and **ERN EuroBloodNet**, and another paper on **Drug-device combinations** together with members of the Therapies Scientific Committee.
  - In addition, the committee is defining the topics for the Task Force proposals for the Roadmap 2025, among the selected areas of interest is stigma and rare diseases with a focus on pharma funding, genetic testing and how to assess the impact and offer solutions for stigma related issues in the life journey of people living with rare diseases and their families. The project aims to bring together multiple partner organizations.
  - **Comments:** A Diagnostic Scientific Committee member mentioned that stigma attached to having a rare disease should be separated from genetics, as the common genetic diseases are

not rare. The member also raised the importance of educating every stakeholder to know how to report to this issue.

4. Joint Constituent and Scientific Committee Session

Paediatric engagement in therapeutic development (TSC led)

The session was led by Anneliene Jonker, TSC Vice Chair, and Maria Cavaller Bellaubi, TSC member, who highlighted the importance of young patient engagement (PE), being considered at this moment a gap in the field of PE. The discussion exemplified the importance of including the perspectives from a wide range of stakeholders, including nurses, carers, siblings and to define to what extent everyone has capabilities and possibilities to provide input (patient, children, depending on different age groups). The issue of consent from parents was also brought up, if it is necessary to be applied when working with young people, as well as the requirement for ethics committee approval. It was recommended that IRDiRC should connect with the work performed by other networks such as TEDDY, connect4children, etc. Another aspect showcased was the Quality of Life (QoL) difference between adults and children with rare diseases.

Drug-device combinations for patients with rare diseases (ISC led)

The ISC Vice Chair, Marc Dooms, led this session and started the discussion by a comparison between medical technology versus medicinal products for RD. In the case of medicinal products, the costs accumulate over the course of the treatment, while the application is often controlled by the patient or carer. In addition, the basic molecule is not modified, and the product has a long-life cycle. From regulatory perspective, it has a basic patent or fewer incremental patents. For the medical technology, there is a higher product cost which is amortized over service life, and the patients might use complex instructions. In this case, it needs a continuous refinement to improve the effectiveness and costs.

Highlighting what are the unmet needs from the patients', healthcare and society perspective is crucial. The majority of clinicians have indicated the need for new device as the existing ones have limitations. The current legislations provide options but also faces certain challenges, especially for paediatric combination products.

The orphan devices have been defined as products or equipment intended for the prediction, prevention, diagnosis, support, treatment and management of life-threatening or chronically debilitating diseases that have a low prevalence or incidence. In consequence, the orphan medical technology is considered as a combination of device (defined by tool or equipment) and the connection part of the device. One example is the combined ATMP – MACE, another one is Flolan.

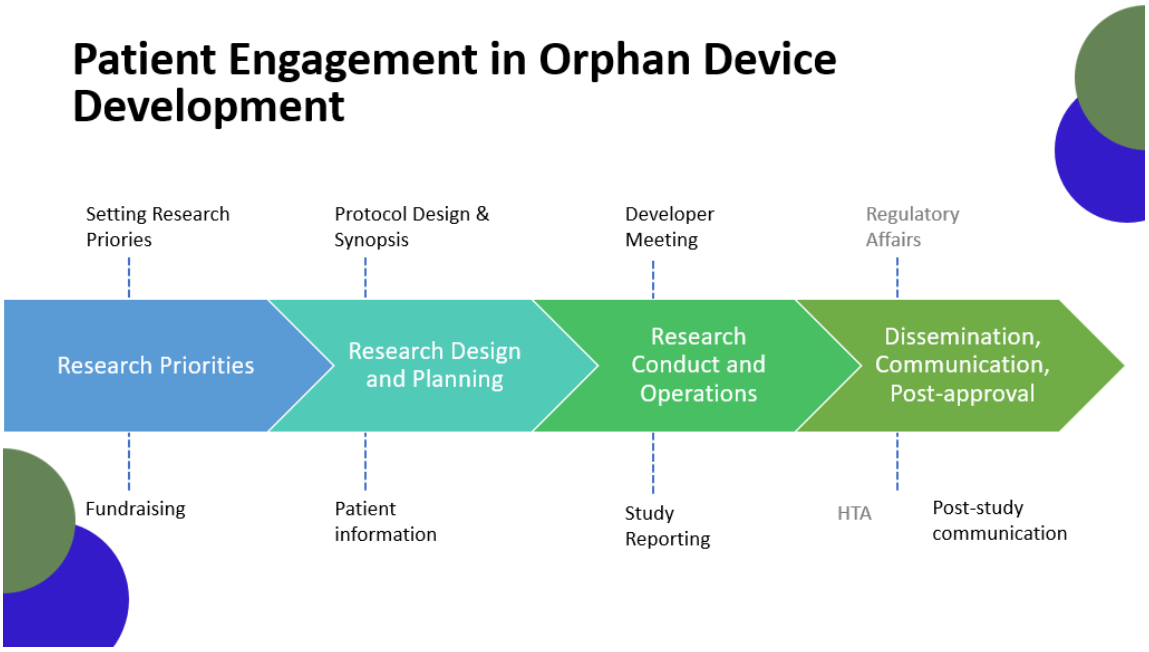

Figure 1. Schema of the patient engagement in the orphan device development based on current research priorities, the research design and planning, operational activities and post-approval actions, including communication and dissemination.

Other examples of drug-device combinations include the neuro-pixels to unravel the brain over weeks, wearable device-smart contact lens and 3D-printed medical devices.

5. Summary of Day 1

IRDiRC Chair, Dr. Dave Pearce, presented a summary of Day 1 of the Consortium Assembly and the agenda for the second day of the meeting.

6. IRDiRC Activities

Task Forces

- **Operationalizing a Comprehensive Framework to Assess the Impacts of Diagnoses and Therapies in Rare Disease 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 did 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. The group is now developing a coherent framework for the manuscript and is looking to include the impacts of experience along the patient journey. The manuscript is being restructured to present the ecosystem determinants, the methodologies and performance indicators, and the lived experience impacts in an integrated way that will link to the patient journey.

- **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 rare diseases.
  - **Planned Output:** Framework for the robust and effective ecosystem of functional analysis in rare diseases; White Paper
  - **Progress Update:** An in-person workshop was held in Paris (France) on March 11-12, 2024. A final teleconference with the group took place on March 29th to discuss the finalization of the manuscript. The Task Force was split in different Working Groups with a focus on specific topics. *Working group 1* has worked on the diagnostic, therapeutic, and scientific drivers that contribute to the improvement of generation and coordination of knowledge bases, tools and platforms. Another focus of the group has been education and awareness of functional analysis in primary and specialist care, from prevention to diagnosis and treatment, and basic research. *Working group 2* has performed a compilation of current functional analysis knowledge, from retrospective and perspective views, and a review of what already exists and what is missing. In addition, the group investigated the matching genes and variants to functional analysis. The focus of *Working group 3* has been on standardization, quality control and regulation, how to best analyze and integrate multi-omics, how to overcome challenging molecular mechanisms and what is the criteria for clinical use, cut-offs, need or not for orthogonal methods for confirmation. *Working group 4* has investigated the cost, access and sustainability, the perspectives of PLWRD and equity, and the increasing throughput and disease/multiplexing/gene agnostic methods.

- **Funding Models to Support the Spectrum of Rare Disease Research and Development**
  - **Objectives:** To identify key motivating factors for different types of funders of rare disease 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 Manuscripts:**

- The first manuscript that includes a **literature review** on existing and **Key Opinion Leader (KOL) interviews results** is planned to be submitted to a scientific journal; The paper was divided in 11 blocks including:
  1) Mission-oriented investments: Public Funders and Foundations
  2) Mission-oriented investments: Patient Groups and Foundations
  3) Mission-oriented investments: Private Investors and Companies
  4) Mission-oriented investments: Cautionary Comment regarding IP
  5) Health Economics
  6) Market Timing
  7)Partnerships: Public-Private, Industry Partnerships with Foundations, Roles and Opportunities for Patient Advocacy Group Investments, Venture Philanthropy
  8) Funding Mechanisms
  9) Downstream Requirements
  10) Human Resources
  11) Scientific Quality
- A second manuscript, planned to be developed as business article, will include the Orphan Drug Database Analysis Results based on US-Food and Drug Administration (FDA) and European Medicines Agency (EMA) databases.

- The close-out meeting of the Task Force took place on April 10th.

➢ **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.
- **Progress Update:** The State-of-the-Art of N-of-1 therapies paper has been submitted to Nature Reviews Drug Discovery. A last Task Force online meeting with the group took place in April.
- **Outputs:** The Task Force collected the list of the available literature resources on the topic. The State-of-Art publication has been re-submitted to journal, addressing the reviewers' comments. A list of educational resources is available on the IRDiRC website.

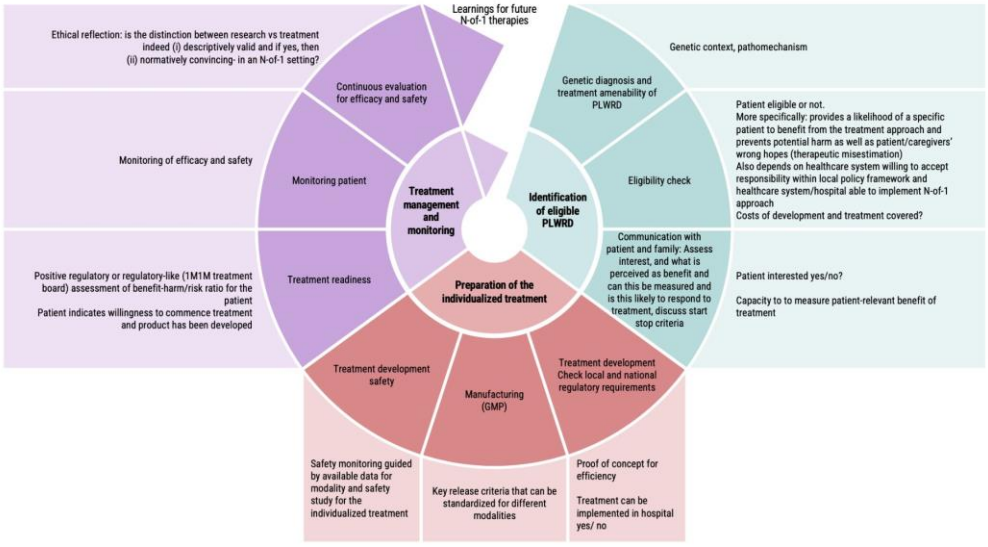

Figure 2: Roadmap for N-of-1 development.

Other IRDiRC initiatives

- **Newborn Screening Initiative**

Progress Update

Four publications from the **Newborn Screening I – Real World Applications and Technologies** are now available open access on the Rare Disease and Orphan Drugs Journal.

1) A systematic review of real-world applications of genome sequencing for newborn screening. Authors: Magnifico Giuditta, Artuso Irene, Benvenuti Stefano
Link: 10.20517/rdodj.2023.17

2) Federated databases: An approach to enhance secure data sharing in newborn genomic screening. Authors: Petros Tsipouras, Maria Chatzou Dunford, Paul R. Billings, and Theoklis Zaoutis
Link: 10.20517/rdodj.2023.15

3) Next-Generation Sequencing-Based Newborn Screening Initiatives in Europe: An Overview. Authors: Virginie Bros-Facer, Stacie Taylor and Christine Patch
Link: 10.20517/rdodj.2023.26

4) Towards genomics in Newborn Screening for Inherited Metabolic Disorders: Expert opinion from IRDiRC-Newborn Screening Initiative. Authors: Guillem Pintos-Morell; Maria Iascone; Giorgio Casari; Raquel Yahyaoui; Elena-Alexandra Tataru; Clara D.M. van Karnebeek; Francjan J. van Spronsen
Link: 10.20517/rdodj.2023.52

Moreover, two publications are currently in review by journal.*

At the time of the writing of this report, the two papers have been launched and are openly accessible on the journal website: https://www.oaepublish.com/specials/rdodj.1270

The Special Edition **Newborn Screening II – Policy, ethics and patient perspectives** has four open access publications:

1) Development of newborn screening policies in Spain 2003-2022: what do we actually need to reach an agreement? Authors: Cristina Valcárcel-Nazco, Lidia García-Pérez, Renata Linertová, Carmen Guirado- Fuentes, Aránzazu Hernández-Yumar, Lucinda Paz-Valiñas, Paula Cantero-Muñoz, Manuel Posada de la Paz, Pedro Serrano-Aguilar
Link: https://www.oaepublish.com/specials/rdodj.1271

2) The Australian landscape of newborn screening in the genomics era. Authors: Charli Ji, Michelle A Farrar, Sarah Norris, Belinda Burns, Bruce Bennetts, Kaustuv Bhattacharya, Tiffany Wotton, Alex Brown, Louise Healy, Nicole Millis (RVA), Didu Kariyawasam
Link: https://www.oaepublish.com/articles/rdodj.2023.30

3) Overcoming challenges in sustaining newborn screening in low-middle-income countries: the Philippine newborn screening system. Authors: Carmencita D. Padilla, Michelle E. Abadingo, Katherine V. Munda, Bradford L. Therrell
Link: https://www.oaepublish.com/articles/rdodj.2023.38

4) Newborn screening in South Africa: the past, present, and plans for the future. Authors: Helen Malherbe, Jim Bonham, Michelle Carrihill, Kamy Chetty, Elne Conradie, Marli Derckson, Sithembile Dlamini-Nqeketo, Hilary Goieman, Marianne Gomes, Brenda Klopper, Neil McKerrow, Carmencita Padilla, Tahir Pillay, Bronwyn Roussot, Tueloa Satekge, Mike Urban, Geroge van der Watt, Heleena Vreede, Dianne Webster, Marco Zampoli, Chris Vorster
Link: https://www.oaepublish.com/articles/rdodj.2023.49

In addition, one publication is under review by the journal and one in preparation.

- **Strategic Communication Sub-Committee Updates**

- In Q1/Q2, the focus has been on the revision of the website structure and content, including the adding of the Key Performance Indications (KPIs). The communication slide deck was updated to include also IRDiRC outputs and task forces. A more sustained activity has been reported on the social media channels: Twitter, LinkedIn and website traffic.
- The planned activities for Q2/Q3 include a better promotion of IRDiRC publications and other outputs, especially on LinkedIn and the creation of a set of operating procedures for communication and dissemination. *The implementation of the new website will be delayed to Q4 due to the change of host of the Scientific Secretariat starting September 2024.*

7. IRDiRC Priorities and Strategies

➢ IRDiRC Timeline for 2024

Past events

- IRDiRC Strategic Meeting, France: 12 January *(IRDiRC internal event)*
- Online Consortium Assembly Meeting: 6-7 March *(IRDiRC internal event)*
- Functional Analysis Task Force in-person workshop, France: 11-12 March *(IRDiRC internal event)*
- Genomics of Rare Disease, UK: 25-27 March
- Orphan Drugs & Rare Disease Global Congress, UK: 11-12 April
- World Orphan Drug Congress (WODC), USA: 23-25 April
- European Conference on Rare Diseases (ECRD) Conference, Belgium: 15-16 May
- In-person Consortium Assembly – Scientific Committees Meeting, China: 22-23 May *(IRDiRC internal event)*
- Global Rare Disease Research Symposium, China: 23-25 May
- European Joint Programme on Rare Diseases (EJP RD) Final Conference, Italy: 27-28 May
- BIO International Convention, USA: 5 June
- DIA Global Annual Meeting, USA: 16-20 June

Upcoming events

- International Collaboration on Rare Diseases and Orphan Drugs (ICORD), Argentina: 24-25 July
- Undiagnosed Diseases Network International (UDNI) Conference, South Korea: 5-7 September
- In-person Consortium Assembly Meeting: 16-17 October *(IRDiRC internal event)*
- WODC, Europe: 22-25 October
- Online Consortium Assembly Meeting: 9-10 December *(IRDiRC internal event)*

➢ 2024 Priorities

- The transition of the Scientific Secretariat from French National Institute of Health and Medical Research (INSERM) to the Foundation for Rare Diseases (France) is planned for September 2024, and its funding will be provided by the European Commission through The European Rare Diseases Research Alliance (ERDERA).
- The Task Force proposals submissions for the IRDiRC Roadmap 2025 is ongoing.
- The State of Play Report 2022-2024 is planned to be delivered the first semester of 2025.
- The preparation of the 2025 IRDiRC conference and RE(ACT) Congress is ongoing, in partnership with ERDERA and BLACKSWAN Foundation.
- An open call for CA Chair and Vice Chair renewal will be launched in September 2024.

➢ New IRDiRC Publications

- **Getting your DUCs in a row - standardising the representation of Digital Use Conditions**. Francis Jeanson, Spencer J. Gibson, Esther van Enckevort, Anthony J. Brookes, et al. 2024 *Scientific Data.*

Link: https://www.nature.com/articles/s41597-024-03280-6

- **Common conditions of use elements. Atomic concepts for consistent and effective information governance.** Maria del Carmen Sanchez Gonzalez, Pim Kamerling, Anthony Brookes, et al. 2024 *Scientific Data*.
  Link: https://www.nature.com/articles/s41597-024-03279-z
- **Analysis of genomics implementation in newborn screening for inherited metabolic disorders: an IRDiRC initiative.** Guillem Pintos-Morell, Maria Iascone, Francjan J. van Spronsen, et al. 2024 RDODJ.
- *Link:* https://www.oaepublish.com/articles/rdodj.2023.52

➢ **Upcoming IRDiRC Meetings in 2024/2025**

Consortium Assembly Meetings

- In-person CA Meeting, October 16-17, 2024, Milan, Italy
  The Consortium Assembly meeting is planned as one day and a half meeting, and it will be hosted by Fondazione Telethon Italy. The conference will be followed by the **workshop on Funding mechanisms** on the afternoon of October 17th.
- Online CA Meeting, December 9-10, 2024 (2 hours on each day)
- In-person CA-SC Meeting, March 3-4, 2025, Brussels, Belgium

RE(ACT) Congress and IRDiRC Conference 2025

- Back-to-back with the IRDiRC CA-SC Meeting, March 3-4, Brussels, Belgium
- RE(ACT) Congress, March 5-7, Brussels, Belgium. More information: https://www.react-congress.org/

IRDiRC at WODC Europe

For a second consecutive year, IRDiRC will participate at the World Orphan Drug Congress Europe at Fira de Barcelona Montjuïc, Spain, from October 22-25, 2024. The WODC is the largest and most established orphan drugs and rare diseases meeting of its kind across the globe and it brings together top pharmaceutical companies, government and regulatory authorities, patient advocacy groups, payers and investors. The conference provides a forum to discuss and advance orphan drugs development and enhance the access to life-saving therapies for patients with rare diseases. Link: https://www.terrapinn.com/conference/world-orphan-drug-congress/index.stm.

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