INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the Consortium Assembly Meeting

07-08 June 2023
Online

IRDiRC

EXECUTIVE SUMMARY

The International Rare Diseases Research Consortium (IRDiRC) held a meeting of the Consortium Assembly (CA) on 7-8 June 2023 via web/teleconference. It was attended by 51 participants representing 30 member organizations, the chairs and vice-chairs of the Scientific Committees and the Scientific Secretariat.

1. **New Leadership and Membership**
   - FCC leadership
     - New FCC Chair: FCC Chair: Christopher McMaster, Canadian Institutes of Health Research (CIHR), Canada
     - New FCC Vice Chair: Stefano Benvenuti, Fondazione Telethon, Italy
   - ISC membership
     - New member: Claudia Gonzaga-Jauregui**,** Assistant Professor at National Autonomous University of Mexico, Mexico

2. **The Scientific Secretariat under the Rare Diseases Partnership (2024-2031)**
   - The Scientific Secretariat will be transferred from Inserm to the French Foundation for Rare Diseases thus allowing more flexibility in the administrative tasks managed by the Secretariat.

3. **IRDiRC 2024 Roadmap**
   - New IRDiRC Task Forces can only be implemented within the Rare Diseases Partnership which starts in September 2024. Therefore, 2024 Task Forces will have to run online exclusively

4. **Updates on IRDiRC Committees**
   - Funders Constituent Committee
   - Companies Constituent Committee
   - Patient Advocates Constituent Committee
   - Diagnostic Scientific Committee
   - Interdisciplinary Scientific Committee
   - Therapies Scientific Committee
   - Regulatory Scientific Committee

5. **Updates on Task Forces, Working Groups and initiatives**
   - Machine Readable Consent and Use Conditions
   - Working Group on MedTech for Rare Diseases
   - Pluto Project
   - Drug Repurposing Guidebook
   - Funding Models to Support the Spectrum of RD Research and Development
   - Operationalizing a Comprehensive Framework to Assess the Impacts of Diagnoses and Therapies in Rare Disease Patients
   - Functional Analysis
   - N-of-1+ Therapies

- Initiative on Newborn Screening
- Parallels Between COVID19 and Rare Diseases
- Demystifying Gene Therapy

6. Upcoming meetings

- In-person Consortium Assembly meeting, 3-4 October 2023, Montreal, Canada
- Online CA meeting, 6-7 December 2023
- In-person Consortium Assembly and Scientific Committee meeting, 27-28 March 2024, Shanghai, China

7. Communication Sub-Committe

- This Committee was created to identify communication issues, potential solutions and tactics to improve IRDiRC visibility. The first meeting was organized on the 24th of May 2023.

8. Collaboration with the European Journal of Medical Genetics

- Special Issue in the Journal presenting articles authored by IRDiRC members. Submission of the articles at the end of 2023.

9. World Orphan Drug Congress – Barcelona 30 October – 02 November

- Several talks by IRDiRC members. IRDiRC will be showcased.
- https://www.terrapinn.com/conference/world-orphan-drug-congress/agenda.stm

10. Definition of IRDiRC Membership

- This discussion will be held in the October meeting of the Consortium Assembly.
- Some organizations do not fit the categories of IRDiRC (funders, companies, patient advocates) but significantly contribute to rare disease research. How can we onboard them?

REPORT

1. Summary of the last CA-SC meeting and outputs

- CA-SC members met in Berlin on the 14th-15th of March 2023. This event was attended by 53 in-person participants and 16 online participants. It allowed the committees to meet individually and hold cross-committee sessions. Updates on IRDiRC activities were provided
- Four topic sessions organized were:
  - Parallels of COVID19 and RD
  - Models for Data and Registries Support Within Regular Funding Mechanisms
  - Clinical Research Networks – Focus on Collaboration
  - Demystifying Gene Therapies for Better Understanding and Accessibility
- Outputs of the meeting were:
  - Invite IRDiRC members to increase the dissemination of IRDiRC activities
  - Create a sub-committee on communication
  - A white paper on "Parallels between Covid-19 and RD"
  - A collection of resources of gene therapies to be published on the IRDiRC website

2. FCC Leadership

- New FCC Chair: Christopher McMaster, Canadian Institutes of Health Research (CIHR), Canada
- New FCC Vice Chair: Stefano Benvenuti, Fondazione Telethon, Italy

3. IRDiRC Scientific Committee Members

- New ISC member: Claudia Gonzaga-Jauregui**,** Assistant Professor at National Autonomous University of Mexico, Mexico

4. Future of the Scientific Secretariat Under the Rare Diseases Partnership

- The European Commission will continue to support and fund the Scientific Secretariat during the RD Partnership (2024-2031)
- The Coordination of the Scientific Secretariat will be transferred from Inserm to the French Foundation for Rare Diseases thus allowing more flexibility in the administrative tasks managed by the Secretariat.

5. Constituent Committee Updates

Funders Constituent Committee

- Election for the FCC Chair and Vice Chair role was implemented in April to May.
- Last FCC meeting was May 24th with the following agenda:
  - Presentation of the New FCC Chairs
  - Updates on FCC Task Forces
  - Funded Projects Database Platform (Polaris) and the Need for Re-Commitment by FCC Members
- 2-hrs online training will be provided by Orphanet for FCC members on the new feature and how to use the platform

- FCC Chairs proposed the below points for FCC members consideration:
  - Implementation of funding model ideas by funders
  - Implementation of mouse to man workbooks/pathways by funders
  - Thoughts on priorities moving forward for the FCC
- Online survey was launched to gather FCC members feedback regarding FCC priorities and meeting frequency

Comments:

- The resources used to develop the funder database come originally from EU. How do you foresee its sustainability?
  - It has to be discussed if and how we can insert it in the future Rare Diseases Partnership
  - Very few funders are utilizing it. We have to have a discussion about its use in the close future.
- IRDiRC members need more training to understand the value of the platform. Does Polaris include AI functionality for text mining aiming to do the curation of the projects?
  - The platform offers 80% of accuracy. The quality control needs to be done for 35000 projects.

Companies Constituent Committee

- Meeting on the next strategic steps for CCC took place on 23 May 2023
- **Key points identified:**
  - Strategic plan on how to increase the number of "companies" in CCC is needed, and how to actively involve companies' representatives
  - Evaluate the member definition, engagement language and interests, and criteria of selection
  - Deploy an outreach strategy to company representatives who would engage based on a shared interest (natural history studies, data sharing, etc.); demonstrate value and expand reach
- Discussions with different companies have been taking place to explore their interest in joining IRDiRC (Biogen [LoI already submitted], BridgeBio, Genomenon, Vigil Neuro)
- There is an on-going call for Chair and Vice Chair election

Comments:

- The criteria entry for companies is 10 million USD investment in RD research. There is a need for a strategic approach. The challenge is to show what is the value for companies to join IRDiRC and participate in the Consortium activities.

Patient Advocacy Constituent Committee

- The last PACC meeting was organized on March 14th in Berlin.
- Key discussion points in PACC

- Introduction of new representatives
- Sharing of activities
  - Rare Disease Day awareness and advocacy activities
  - Other training, research and collaboration events
  - Upcoming events, including regional and international conferences
- Group discussion on role(s) of PACC
  - Origin and history of PACC
  - Engagement of PACC (members) in IRDiRC Task Forces and Working Groups
  - Recruitment, training and support of "expert" patients to participate in IRDiRC and other groups and projects
  - Mentorship and Capacity Building for PACC members

➢ Proposal for online "needs assessment" to identify potential roles for PACC members and benefits to member organizations

Comments:

➢ Umbrella groups were originally selected because of their ability to think broadly, having no conflict of interest regarding a particular disease/treatment. Several umbrella groups were in Iceland at the founding of International Rare Diseases Research Consortium. That preceded the PACC. Umbrella groups participated for years before the formation of the PACC and have created methods and rubrics for companies and patient groups to work together. Disease specific patient group leaders can participate under the umbrellas - as they have so far. So people with expertise in R&D can be part of the groups.

6. Task Forces, Working Groups and other initiatives

➢ **Updates were provided for the following activities**

- Machine Readable Consent and Use Conditions
- Working Group on MedTech for Rare Diseases
- Pluto Project
- Drug Repurposing Guidebook
- Funding Models to Support the Spectrum of RD Research and Development
- Operationalizing a Comprehensive Framework to Assess the Impacts of Diagnoses and Therapies in Rare Disease Patients
- Functional Analysis
- N-of-1+ Therapies
- Initiative on Newborn Screening
- Parallels Between COVID19 and Rare Diseases
- Demystifying Gene Therapy

➢ **Machine Readable Consent and Use Conditions**

- Objectives: The objective of the Task Force is to create machine-readable profiles for consent and use for registries and biobanks, building on the data structures and semantics of the Global Alliance for Genomics and Health (GA4GH) + IRDiRC standards.

- Expected Output: The Task Force aims to work with ontology developers to define new classes and properties that fill gaps identified in the ontologies. They also plan to extend the concepts of the Consent Codes Exchange (CCE) to provide a basis for consent and use conditions at the record level, in Data Use Consent (DUC) format.
- Progress Update: The Task Force has two papers on DUC and CCEs that are in the last phase for submission. They are currently working on creating an individual-level consent template that is mapped to the DUC-CCE structure and verifying its applicability and compatibility with different parts of the record-level approach. They are also evaluating the need for consent and use conditions at the record level for biobanking data and defining the objectives. The development of Real-World Pilots has started, but it is still in the early phases.

➢ **Working Group on MedTech for Rare Diseases**

- Objectives: To understand and map the current incentives, supportive frameworks, and unmet technical and functional needs for developing medical devices for rare diseases. The group would also like to identify the regulatory landscape in the different regions for medical devices.
- Expected Output: The Working Group intends to publish a comprehensive overview of incentives and frameworks for medical devices worldwide, opportunities for harmonization approaches in the regulatory space, and the involvement of patients in medical device development.
- Progress Update: The Working Group is currently preparing an article that summarizes the state-of-the-art in rare diseases and medical technologies. They have also engaged in discussions on patient involvement in MedTech development and participated in few speaking engagements.

➢ **Pluto Project on Under-Researched Rare Diseases**

- Objectives: To identify those rare diseases that appear to have attracted virtually no interest from academic researchers and industrial developers; determine the characteristics they have in common; and understand roadblocks in developing effective treatments for such diseases.
- Progress Update: A new working definition of under-researched rare disease has been drafted as a disease that has a “4-Zero Concept”: No clinical trial activity, No scientific publication available, No regulatory authority orphan drug designation, and No approved medicine/therapy. 4-Zero Concept is now questioned as initial methodological approach has shown that databases are either incomplete with redundant terminology and or duplications. Requirement for further database integration to identify groups of diseases based on variables that can be consistently and robustly measured drawing valid conclusions. Therefore, the TF will develop new methodological approaches to measure the characteristics of under-researched rare diseases.
- Expected Output: The Task Force is preparing two articles (1) Methodology paper describing the Pluto planets of under-researched rare diseases, (2) Policy Recommendations paper describing identified commonalities between

disregarded RD, roadblocks for therapy development, and opportunities to overcome them and foster research and development.

- **Drug Repurposing Guidebook**
  - Objectives: To create a guidebook focused on repurposing approaches (incentives, regulatory tools, initiatives, development tools, etc.).
  - Progress Update: The group agreed on a definition of repurposing and identified four model cases for the guidebook which are on/off patent and with/without Marketing Authorisation Applications (MAA). The group finalized the preparation of the materials including, the building blocks, the activity chart, the START checklist. A manuscript was submitted to Nature Review Drug Discovery
  - Expected Output: The Orphan Drug Development Guidebook will be completed with the materials developed by the Task Force.

- **Funding Models to Support the Spectrum of RD 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. To identify the key influencing factors and models for effective public-private partnerships at different stages of a treatment's life cycle.
  - Progress Update: The group is collecting literature on funding models and public private partnerships. Interviews with key opinion leaders are under preparation.
  - Expected Output: A summary paper of findings and a Funding Model/Good Practices Toolbox.

- **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 rare disease.
  - Progress Update: The group started a literature review search to identify the elements of the framework. Both quantitative and quantitative measures have been identified.
  - Expected Output: Framework to assess impact. White paper.

- **Functional Analysis**
  - Objectives: To create a framework for the robust and effective ecosystem of functional analyses in rare diseases
  - Progress Update: The group is currently looking to identify the challenges and opportunities in functional analysis. A workshop will be organized on November 4th-5th in Barcelona, back-to-back with World Orphan Drug Congress and N-of-1 Task Force Workshop.
  - Expected Output: Framework for the robust and effective ecosystem of functional analyses in rare diseases. White paper.

- **N-of-1+ Therapies**

- 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
- Progress Update: The group reviewed the literature and drafted the outline for the state-of-the art paper. The therapy roadmap for N-of-1 therapy development is drafted. A workshop will be organized on November 2th-3th in Barcelona, back-to-back with World Orphan Drug Congress.
- Expected Output: Framework for the robust and effective ecosystem of functional analyses in rare diseases. White paper.

➢ **Newborn Screening (initiative)**

- Special edition in the Rare Disease and Orphan Drugs Journal
- Theme 1: Real World Applications and Technologies

Manuscript type	Title (final/*tentative*)	Authors	Status
1. Systematic review	A systematic review of real-world applications of genome sequencing for newborn screening	Magnifico Giuditta, Artuso Irene, Benvenuti Stefano	**Final draft in last internal review**
2. Perspective	Could federated data analysis be the catalyst accelerating the introduction of newborn genome screening for the detection of genetic disease?	Petros Tsipouras, Maria Chatzou Dunford, Hadley E. Shepherd, Hannah Gaimster, Theoklis Zaoutis	**Final draft in last internal review**
3. Case report	*Horizon scan of Next-Generation Sequencing-based Newborn Screening initiatives in Europe*	Virginie Bros-Facer , Maria Martinez-Fresno, Adriana Huertas-Vazquez, & Christine Patch (TBC)	Survey (57 questions) sent to 14 genomic NBS initiatives in Europe (April/May) Responses collected & data analyzed (May) **Draft in development (ready end of June)**
4. Perspective	*Success and challenges in European NBS Alliance on SMA*	Marie-Christine Ouillade	Draft in development
5. Expert Opinion	*Pitfalls and limits of NBS for inborn errors of metabolisms and how NGS could be the next complementary test*	Guillem Pintos-Morell, Clara Van Karnebeek, Francjan van Spronsen, Raquel Yahyaoui, Maria Iascone, Giorgio Casari and Stefano Benvenuti	Scope and outline done First draft in development
6. TBC	*Real World Data Standards and its significance for the extension of high-quality NBS practice*	Mengchun Gong and colleagues	TBC

- Theme 2: Policy, Ethics and Patient Perspectives

Manuscript Type	Topic/Title	Authors	Status
1.Commentary	Bridging the NBS gap between HIC & LMIC	Helen Malherbe & Mary Wang TBC: Melissa Parisi, Manuel Posada, Domenica Taruscio, Carmencita Padilla, Luis Alejandro Barrera Avellaneda, EURORDIS	Confirmed. Outline sent 6 June to finalise authors.
2.	NBS in Spain	Manuel Posada, ISCIII, other members of the Dr. Serrano's team	Confirmed, Article submitted 5 June.
3.	The Australian landscape of newborn screening in the genomics era	Sanduni (Sandi) Kariyawasam & Michelle A Farrar,Sarah Norris, Belinda Burns, Bruce Bennetts, Kaustuv Bhattacharya, Tiffany Wotton, Alex Brown, Louise Healy, Nicole Millis (RVA), Didu Kariyawasam	Confirmed. Outline received 16 May & article draft circulating with authors (7 June).
4.	NBS in the Philippines	Carmencita Padilla	Confirmed. Awaiting further details.
5.	NBS in South Africa	Helen Malherbe, George van der Watt, Marco Zampoli, Elne Conradie, Surita Meldau, Chris Vorster	Confirmed. Based on NBS4SA meeting report (Feb '23). First draft end June
6.	NBS in Japan	Yukiko Nishimura	Confirmed. First draft end of June

Manuscript Type	Topic/Title	Authors	Status
7.Review/ research	Overview/status of NBS in SSA: Scoping review	Helen Malherbe, others TBC e.g. Marli Derckson, Surita Meldau, Solomon Ofori-Acquah (Ghana); Professor Obiageli Nnodu (Nigeria)	Confirmed. Outline sent 6 June to finalise authors.
8. Commentary/ Perspective	Update on NBS in Asia	Professor Luo Xiaoping, Mengchun Gong, Carmencita Padilla, Yukiko Nishimura, Ritu Jain	Confirmed. Awaiting details (19 May & 6 June)
9. Commentary/ Perspective	Update on NBS in Europe?	Marie-Christine Ouillade, Inputs from other European based members, Gulcin Gumus, EURORDIS	Confirmed: Follow up with after 9 May
10. Case studies/ Commentary	Patient advocacy in action for NBS The SMA example	Vicky Antoniadou (+2 colleagues: 1x clinician & 1x patient rep) Marie-Christine Ouillade, Mary Wang, Durhane Wong-Reiger, Ritu Jain	Confirmed: Article outline received 27 April.
11.	ELSI of Genetics in NBS	Sarah Norris, Michelle Farrar	Confirmed. Awaiting details
12. Review	NBS in the rural community	Elizabeth McNeil	Confirmed. 1st draft end June.

- **Parallels Between COVID19 and Rare Diseases (initiative)**
  - The main goal is to publish a position paper drawing parallels between COVID pandemic and "Operation Warp Speed" and the urgent and ongoing silent epidemic of Rare Diseases.
  - The draft of the article is completed. It is expected to be submitted in the fall 2023 as a position paper making the case for ongoing Rare Diseases emergency.

- **Demystifying Gene Therapies**
  - The information submitted by IRDiRC members on gene therapies was collected and collated.
  - A review article will summarize this knowledge and will be submitted to the European Journal of Medical Genetics.

7. IRDiRC Roadmap 2024

- EJP RD extension was officially approved by the European Commission which includes the IRDiRC Scientific Secretariat, but new IRDiRC Task Forces can only be implemented within the Rare Diseases Partnership which starts in September 2024. Therefore, 2024 Task Forces will have to run online exclusively.

8. Upcoming IRDiRC meetings

- 03-04 October 2023: In person Consortium Assembly Meeting in Montreal (Canada)
  - 1.5-day meeting; Back-to-Back with Canada RD Meeting
  - Meeting organization supported by CIHR Institute of Genetics, Genome Canada, and CORD
  - Hotel rooms available in the meeting venue with discount for all IRDiRC members, applicable until 03 July 2023
  - 28 confirmed in-person participants; 11 Maybe
- 06-07 December 2023: Online Consortium Assembly Meeting at 15:00-17:00 CET
- 27-28 March 2024: In person Consortium Assembly Meeting in Shanghai (China)
  - 1.5-day meeting; Back-to-Back with China RD Research and Translational Medicine Annual Conference

- Meeting organization supported by Chinese Organization for Rare Disorders (CORD) and Hope for Rare Foundation
- Airport transfer, hotel rooms, some lunches, and some dinners are covered by CORD and Hope for Rare Foundation for all IRDiRC members from 26-31 March 2024
- 50 confirmed in-person participants; 20 Maybe

9. Scientific Committee Updates

Diagnostic Scientific Committee

- End of mandate
  - Juergen Reichardt, Professor, Australian Institute of Tropical Health and Medicine (AITHM), James Cook University, Australia
  - Francois van der Westhuizen, Professor of Biochemistry, North-West University, Potchefstroom, South Africa
- The DSC launched a nomination call for rare disease diagnostic expert from Africa or Latin America and a rare disease patient/patient advocate from anywhere in the world with strong interest and involvement in activities or initiatives to improve rare disease diagnosis. The call was closed on the 9th of June.
- Overview of the DSC activities:
  - Productive meeting in Berlin on March 2023 with the two new chairs
  - Functional Analysis Task Force developed by departing chairs is underway
  - Berlin discussion focused on large goals
    - Select task for priorities from large set of prior proposals
    - Set as a tentative goal an annual publication of diagnostic priorities
- Follow-up items for future meetings:
  - Preliminary interest in "Transitions from diagnosis to therapy."
    - Consider joint task force with other scientific committees
    - Funding delays will provide additional time for deliberation
  - Establish a working group for diagnostic priorities manuscript
    - Schedule rapid-review session for triage of existing literature
  - Work with program staff to schedule future meetings (done)
    - July, September, December
  - Work on recruiting replacements for graduating DSC members

Interdisciplinary Scientific Committee

- Online monthly teleconferences have been occurring since September 2022
- The committee proposed 2 Task Forces for the IRDiRC Roadmap 2024-2025:
  - **"G-Force"** (**G**enerative AI for Rare Disease Task**FORCE**) – Task Force draft has been completed and discussed by email with the Operating Committee
  - **"Stigma" Task Force** - a final draft has been submitted to the Operating Committee
- Next steps for the committee:

- Evaluate the members activity and involvement in the committee's activities on yearly basis
- Stronger involvement of patient advocacy groups in ISC: possible recruitment of a patient advocate in the committee

Therapies Scientific Committee

- End of mandate:
  - Annemieke Aartsma-Rus
  - Michela Gabaldo (joined the RSC)
- The TSC launched a nomination call for 3-4 rare disease experts from Africa, Asia, Australasia or North- or South America (could be Industry, Regulatory, Research, Health Technology Appraisal / Payer, Clinical care or Patient group)
- The TSC brainstormed on topics to work on, within and outside the committee in the next tear
  - Drug device combinations
  - Patient engagement in pediatric therapy development
  - Guidance for small populations
  - Guidelines for digital biomarkers
  - Pre-competitive space working
  - Patient-relevant regulatory endorsed endpoints
  - AI in therapy development (this topic will join the Operating Committee proposal)
  - HTA and health economics of therapy development
- The TSC developed a manuscript to discuss the definition of rare disease in the era of precision medicine

Regulatory Scientific Committee

- Online conference organized every month.
- The RSC defined its mission and developed specific actions:
  - Mission: "To benefit all rare disease patients through the development, communication and transparency of Regulatory Science"
  - Short term actions: Define current regulatory environment for rare diseases around the world through case examples and precedent; communicate flexibility; identify gaps.
  - Mid-term actions:
    - Landscape analysis by region in the last 5-7 years, focus on precision/genetic approaches
    - Develop case examples of what worked/what didn't
- Work in progress:

- Mapping of Orphan Medicinal Products approved throughout regions in the world. Synergy with the ongoing mapping of the TSC working with Orphanet and comparing it with the regulatory science network in the Netherlands.
- Landscape analysis of Orphan/Rare approvals 2018 – 2022, focus on precision medicine approaches – what can we learn from these?
- Examine differences by region; Contribution by RSC members in the context of existing (divergent) jurisdictions

➢ Publications expected:
- Landscape analysis late 2023
- Comparisons by region late 2023
- Case examples 2023
- Identify gaps, propose solutions 2024

10. Communication-Sub Committee Updates

➢ The first group meeting held on the 24th of May 2023
- Introductions and identification of communication issues, potential solutions and tactics to improve IRDiRC visibility

➢ The ongoing work is divided into two phases:
- Phase 1: Create a strategic framework to be agreed with the communications sub-committee
- Phase 2: Build out the messages and tactics

Comments:

➢ The possibility to subcontract some activities will depend on the resources of the Scientific Secretariat

11. Collaboration with the European Journal of Medical Genetics

➢ Timeline: 2-3 weeks to select the topics and end of the year (November) to submit the papers

➢ The following articles were proposed for the Special Issue:
- **Dave Pearce** (Sanford Research, USA): "Demystifying Gene Therapies"
- **Andrea Gropman** (Children's National, USA): "*From biomarker to study to basket clinical trials. Advancing science from the bedside or bench to trials: two models in academia*"
- **Gareth Baynam** (Rare Care Centre, Australia): "*Rare Care - holistic cross-sector care coordination*"
- **Sally Ann Lynch** (University College Dublin, Ireland): "*Cataloguing rare genetic disorders found amongst the Roma population*"
- **Claudia Fuchs** (EURORDIS, Italy): "*Drug Repurposing in Neurodevelopmental Disorders*"

- **Anneliene Jonker** (University of Twente, The Netherlands) and **Marc Dooms** (University Hospitals Leuven, Belgium): "Devise – ways forward for medical devices for rare diseases"
- **Ruxandra Draghia Akli** (Johnson & Johnson, USA): "Title TBD"

Comments:

- DSC Chair is interested to submit the DSC paper on priority topics in diagnosis.
- This collection may also be of interest for the RSC case manuscript.

12. World Orphan Drug Congress – Barcelona Edition

- The WODC will be organized from 30th October – 2nd November, in Barcelona.
  - Pre-congress workshop on October 30th
  - Main congress days 31st October – 2nd November
- Several IRDiRC members will participate as speakers
  - Virginie Hivert, Daneil O'Connor, Marjon Pasmooij, Anneliene Jonker, Violeta Stoyanova-Beninska, Cesar Hernandez, Virginie Bros-Facer, Samantha Parker, David Pearce, Daria Julkowska, Durhane Wong-Rieger, PJ Brooks, Anne Pariser, Maria Cavaller Bellaubi
  - Congress agenda: https://www.terrapinn.com/conference/world-orphan-drug-congress/agenda.stm
- IRDiRC will be showcased and will have a booth

13. IRDiRC Membership Definition

- Some organizations do not fit the categories of IRDiRC (funders, companies, patient advocates) but significantly contribute to rare disease research. How can we onboard them? This point will be discussed in Montreal.
- Some organizations such as companies have people with very different expertise and struggle to identify a person who could bring the full perspective of their organization and address the different needs of the Consortium. Is it valuable to have more than one representative per organization during the meetings?

Comments:

- When IRDiRC was founded, it was decided that only one person would represent an organization to ensure continuity, avoid repetitions and be more effective decision makers.
- On the other side, having two people may be very interesting to avoid gaps induced by people turnover.
- The Scientific Secretariat will send a form to Consortium Assembly to get the feedback of members.