



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 20th Consortium  
Assembly Meeting**

October 1 & 2, 2020



**IRDIRC**

## EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on October 1 & 2, 2020, via web/teleconference. The event was attended by 50 participants on October 1<sup>st</sup> and 46 participants on October 2<sup>nd</sup>.

- ▶ Task force/working group leaders presented updates on the six ongoing activities.
- ▶ Task forces/working groups, especially when nearing completion, were invited to implement a dissemination and exploitation strategy for the project outcomes to maximize reach to stakeholders and advancement of IRDiRC goals.
- ▶ Eight new proposals for the 2021 Roadmap were presented; the Scientific Secretariat will have the capacity to accommodate two new activities in 2021.
- ▶ Activity proposers were encouraged to consider merging their activities or to seek collaboration with outside organizations whenever possible, to allow more activities to be undertaken during the year.
- ▶ Advancements towards IRDiRC goals were discussed and areas of attention identified.
- ▶ The IRDiRC-RE(ACT) Congress was confirmed as an online event on 13-15 January 2021.
- ▶ A celebratory event for the IRDiRC 10-year anniversary was proposed to be held during the second semester of 2021.

## REPORT – 01 October 2020

### 1. Introduction

- Samantha Parker (Lysogene) was elected Vice Chair of the Companies Constituent Committee.
- The current IRDiRC leadership is constituted as follows:
  - Chair, Consortium Assembly (CA): Lucia Monaco
  - Vice Chair, Consortium Assembly (CA): David Pearce
  - Chair, Funders Constituent Committee (FCC): Adam Hartman
  - Vice Chair, Funders Constituent Committee (FCC): Catherine Nguyen
  - Chair, Companies Constituent Committee (CCC): Katherine Beaverson
  - Vice Chair, Companies Constituent Committee (CCC): Samantha Parker
  - Chair, Patient Advocacy Constituent Committee (PACC): Durhane Wong-Rieger
  - Vice Chair, Patient Advocacy Constituent Committee (PACC): Yukiko Nishimura
  - Chair, Diagnostics Scientific Committee (DSC): Gareth Baynam
  - Vice Chair, Diagnostics Scientific Committee (DSC): Sarah Bowdin
  - Chair, Therapies Scientific Committee (TSC): Diego Ardigo
  - Vice Chair, Therapies Scientific Committee (TSC): Virginie Hivert
  - Chair, Interdisciplinary Scientific Committee (ISC): Philip John Brooks
  - Vice Chair, Interdisciplinary Scientific Committee (ISC): Dixie Baker

### 2. Update on ongoing activities

- Updates on ongoing activities were presented by task force/working group leaders.
- As a general recommendation, task forces/working groups, especially when nearing completion, were invited to implement a dissemination and exploitation strategy for the project outcomes to maximize reach to stakeholders and advancement of IRDiRC goals. Elements to be taken into account:
  - Who will benefit (potential users)?
  - How will they benefit?
  - How to maximize reach (methods and tools)?
  - Identification of dissemination opportunities

#### ***Indigenous Populations (Gareth Baynam, DSC)***

- The goal is to explore the access barriers to diagnosis of rare genetic diseases faced by Indigenous peoples, with a view of developing recommendations to overcome them
- Progress summary: activities are expected to be completed by the end of year: a review paper focused on the barriers regarding access to genomic technologies and research by Indigenous populations was submitted for publication in the journal *Frontiers in Pediatrics*; the revised manuscript is about to be re-submitted and should be accepted within the next few weeks.

- A second paper is under development and will focus on the recommendations to overcome identified barriers and to advance RD diagnosis.
- Discussion:
  - What are the main barriers? --> Difficult to access, be it geographical, financial, cultural, or linguistic barriers, lack of genomic datasets, lack of efforts directed at community capacity building and intercultural engagement with communities.
  - What were the major concerns highlighted by the manuscript reviewers? --> These were minor, mostly focused on financial barriers and capacity building.
  - How much do barriers differ across different geographies? --> Barriers don't differ much per geographical region. However, this first paper was mostly focused on Indigenous Populations from high income countries (HIC), where Indigenous peoples are a relatively small minority group, whereas Indigenous peoples are a significant part of the population in middle- and low-income countries (MLIC). On these parts of the globe, there are particular challenges as to rare diseases vs other disorder groups to be considered (e.g., infectious diseases).
  - Pursue discussion with CA Vice Chair on how the metrics developed by the working group on Goal 3 could be applied to address the socio-economic aspects mentioned above.

***Chrysalis Project (Adam Hartman, FCC)***

- Main goal is to identify key criteria that would make rare diseases research more attractive to industry for R&D and identify opportunity gaps in the current funding opportunity landscape to develop recommendations
- The team is currently developing the survey targeting companies; the output should be a whitepaper .
- Discussion:
  - Who are the end users of the survey findings? --> Too early to define, it will depend on the findings but certainly funders, patient advocates, and perhaps companies.
  - Which type of industry partners will be contacted? --> The goal is to reach the full spectrum, ranging from big pharma to potentially start up focused on individual RDs.
  - Survey methodology: is it going to be only online or completed by semi-structured interviews? --> currently under discussion.
  - Do you consider asking questions to funders? --> The survey is focused primarily on corporate interests.
- Suggestion to provide a pdf version of the survey in advance to respondents so they can take time to consider answers and connect with collaborators if needed.

***Clinical Research Networks (Stephen Groft)***

- The goals are:
  - To map and analyze the existing ecosystem of national/supranational clinical research networks (CRNs).
  - To develop policy recommendations on guiding principles for an international framework of collaboration of these networks in respect to best practices, interoperability, tools and common goals.
  - To develop relevant recommendations for funders based on gaps identified through the mapping exercise.
- Progress summary: CRNs landscape have been mapped and a survey developed to characterize the CRNs attributes, identify the tools/resources/standards used to reach

international interoperability and also to identify the barriers preventing the networks from reaching this goal.

- Discussion:
  - The outcomes of the survey are very promising in terms of identification of the needs. It will be possible to propose different solutions that can be supported by IRDiRC to accelerate the development of interoperability capacities between the networks.
  - Any systematic barriers already identified? --> The geographical distribution of the networks who responded to the survey is broad. It will allow the identification of what are the barriers and how they have been overcome by some groups.
- The Rare Diseases Clinical Research Network (RDCRN) has an initiative in which one of the goals is to establish a global RD clinical network.

#### ***Machine Readable Consent (Anthony Brooks, DSC)***

- The objective of this Task Force is to transform the generalized consent clauses developed in previous work of IRDiRC into a machine readable and computable consent framework.
- This activity is being developed within the framework of EJP RD and is supported by IRDiRC.
- Contribution to the further development and aligning of
  - DUO (<https://www.ga4gh.org/news/data-use-ontology-approved-as-a-ga4gh-technical-standard/>)
  - and ADA-M (<https://irdirc.org/activities/task-forces/automatable-discovery-and-access/>)
- Discussion
  - Is IRDiRC working collaboratively with Global Alliance for Genomics and Health (GA4GH) or is it trying to consume DUO? --> they are tightly related but different. The machine readable and computable consent framework will add data structure and will not use of new semantics.
  - Members of GA4GH participated in the initial team meeting organized by EJP RD and were invited to collaborate in the project.
  - Could it be used in any disease or only RD? ADA-M was not designed to be specific but rather general. This new structure/design will be the same. We want it to be generically applicable.

#### ***Rare Diseases Treatment Access (Durhane Wong-Rieger, PACC)***

- The goal of this activity is to improve global access to rare disease medicines by creating a list of standard-of-care medicines and identifying the systemic and idiosyncratic barriers to access, especially in low-and-middle income countries (not excluding high-income countries)
- Discussion
  - Did you encounter hurdles in industry participation? --> Companies did not get involved in the creation of the list to avoid any conflict of interest.
  - Possibility to consider interaction with regulatory groups (ICMRA, ICH) for the identification of the barriers. IFPMA were also approached and might be partners.
  - Did you develop guiding principles for identifying the drugs? --> Effectiveness and safety are the primary criteria.

#### ***Alternative Business Models in Drug Repurposing (Virginie Hivert, TSC)***

- The Task Force on Alternative Business Models in Drug Repurposing was established with the goal of identifying key take-aways and potential recommendations to IRDiRC stakeholders regarding the suitability and key recurring elements on Alternative Business Models for the development and commercialization of orphan drugs. The Task Force intends to:
  - Review successful cases of academic and industrial drug repurposing;
  - Identify their characteristics and similarities;
  - Characterize the specificity of their business models and their long-term outcomes;
  - Develop recommendations to IRDiRC stakeholders and the RD community.
- The Task Force was launched in August 2020 to fill the gap left by the delayed start of the Task Forces on Shared Molecular Etiologies and New Technologies for the Diagnostic of RD. These activities will start in Q1 2021.

### 3. Overall vision

#### ***Where do we stand toward the achievement of IRDiRC goals?***

##### Key points addressed in the discussion:

- More needs to be done in terms of diagnosis, access to genetic testing, getting approval from insurances in the US is very challenging. --> Working group on Goal 3 is encompassing all activities.
- The Undiagnosed Diseases Network International (UDNI) activated a new working group dedicated to undiagnosed RDs in underdeveloped countries – to be considered for potential collaborations.
- Dissemination and development of the Orphan Drug Development Guidebook (ODGG) will be sustained through partnership with other programs. What about the other task force outcomes? How do we want to revise them? What complementary actions could we do to advance them and make them useful for the community over time?
- IRDiRC needs to better understand the policy makers position and how to reach them and convince them.
- If IRDiRC really wants to make a difference, it will be important to work closely with health policy makers and health care providers.

#### ***IRDiRC 10-year anniversary***

- Proposals for the celebration of IRDiRC's 10th year anniversary:
  - Anniversary event
    - 2nd semester 2021
  - Anniversary publication
    - IRDiRC impact on the RD community
    - Prospect towards the 2027 goals

### 4. Update from the EJP RD Consortium Assembly on September 14-18, 2020 (Daria Julkowska)

- No questions/comments

## REPORT – 02 October 2020

### 1.COVID survey

- The goal of the survey is to gauge the impact of COVID-19 on the RD research community and to understand how IRDiRC can react to such changes
- 50% respondent:
  - Impact of the pandemic on the achievement of IRDiRC goals: 1/3 pessimists vs 2/3 optimistic
- 20/24 IRDiRC members who have set up a survey on the topic are willing to share key findings
- Possibility to issue a white paper on pandemic preparedness for the RD research community.
- No specific recommendations regarding further actions IRDiRC could take to move forward.

### 2.Roadmap 2021

- Timeline & strategy:
  - The full proposals will be presented during the next CA meeting in January 2021
  - The vote will be set up after the meeting to prioritize the activities that will be launched in 2021
- Key questions for open discussion on each proposal:
  - Is this activity responding to urgent needs and gaps to advance toward the IRDiRC goals?
  - Is this activity relevant to address these needs and gaps?
  - Are there any activities sufficiently overlapping to be merged into one action?
  - Are there any activities that may benefit from joint efforts with outside organisations?

### Proposals discussion

#### ***Drug repurposing Guidebook - Big Bang project (part 2)***

- Discussion:
  - Will you involve regulatory authorities? --> Some TSC members are representatives of regulatory agencies; it will be important to have them in the Task Force to benefit from what has been done in EU and US (e.g. STAMP initiative on drug repurposing in the EU)
  - Do we keep data on the use of the ODGG? --> it's an ongoing process, some metrics are being developed to monitor the use of the guidebook. The tutorial on how to use the guidebook was communicated during the ECRD congress. A webinar will be organized with the EC and the guidebook will be presented at EMA. The YouTube video has been seen more than 400 times.
  - The guidebook material is currently in the transition phase from IRDiRC to EJP RD.
  - Publication, considering open journal? --> yes and all materials are publicly available in the IRDiRC website and fully accessible to the external community.

#### ***Disregarded RD Working Group (PLUTO PROJECT)***

- Proposition for working group
- IT part is being provided by Chiesi
- Discussion:



- There is a huge amount of RD for which there is no research; how to deal with this challenge and identify common data elements? --> For the majority of the diseases, there's a minimum knowledge usually available in Orphanet (e.g., gene, cellular location, organ) and also other databases. Only ultrarare diseases have absolutely no indications.
- Can the task be achieved in one year? This seems to be a big work first to gather enough information and then to assess the evidence behind each data/information found.
- One of the major problems is that there is no financial interest from companies to develop drugs for very low prevalence diseases. Also, the shared molecular etiologies approach is actually designed to address this problem. It redefines disease and enrolls patients in clinical trials based on molecular etiology, rather than traditionally defined diseases.

### ***MedTech for Rare Diseases***

- Discussion:
  - Consumer health tech becoming very sophisticated and widely used. Will the task force include consumer and health regulated devices? --> both are in the scope
  - Suggested reading: <https://www.nature.com/articles/s41746-020-00324-0>

### ***Facilitating and enabling global access to RD treatments***

- Comments: n/a

### ***Understanding impact of RD on Carers across diseases and countries***

- May start as a working group
- Discussion:
  - Could be done in collaboration with projects that could be funded under the next EJPRD JTC 2021 call on SHS topics (<https://www.ejprarediseases.org/index.php/early-announcement-jtc2021/>) --> pursue discussion within PACC
  - Pursue discussion for connection with working group on Goal 3 (i.e., socio- economic impact on caregivers and families)

### ***Measuring impact of diagnosis and treatment of disease***

- Discussion:
  - Companies are asked to put these data together for payers; they want the voice of patients to be included in their decision making but they need to access these data. The manuscript should be oriented toward them.
  - For some diseases, there is already a lot of material regarding direct/indirect cost (e.g., SMA).
  - The following papers can clarify the current knowledge regarding cost of diseases:
    - The burden, epidemiology, costs and treatment for Duchenne muscular dystrophy: an evidence review S. Ryder, R. M. Leadley, N. Armstrong, M. Westwood, S. de Kock, T. Butt, M. Jain, J. Kleijnen Orphanet J Rare Dis. 2017; 12: 79. Published online 2017 Apr 26. doi: 10.1186/s13023-017-0631-3 PMID: PMC5405509



- Social and economic costs and health-related quality of life in non-institutionalised patients with cystic fibrosis in the United Kingdom Aris Angelis, Panos Kanavos, Julio López-Bastida, Renata Linertová, Elena Nicod, Pedro Serrano-Aguilar, BURQOL-RD Research Network BMC Health Serv Res. 2015; 15: 428. Published online 2015 Sep 28. doi: 10.1186/s12913-015-1061-3. PMCID: PMC4587726

### **Primary Care**

- Discussion:
  - Important to engage with electronic health record companies to integrate clinical decision support.
  - Interoperability is also important since not all of them have access to Electronic Health Records.
  - Collaboration with ERN working group on national integration (entire care pathways from GPs, referrals, etc) would be valuable.
  - It might be interesting looking at what has been developed by Medics4Rare in UK and also the UK Royal College of Nurses (they developed some online education program on at least one RD).

### **Enabling and enhancing telehealth for RD across the globe**

- Discussion:
  - Rural health systems are equipped with telehealth. Good example for implementation of telehealth. Need to develop the infrastructure to support telehealth.
  - Need to include ERN in the reflection since telehealth is part of their mandate
  - Are drug development and clinical trials part of this effort? If so, it is worth including industry --> The proposal is to expand the reach to difficult patients (all aspects of RD research and manifestation, including drug development).

### **3.Overall discussion**

- The border between research and health care is floating. It's important to think whether IRDiRC could be reinforced by including additional stakeholders that may merge these two borders --> this may require reconsidering the nature and the goals of IRDiRC (currently a research consortium).
- Eight proposals were presented but the Scientific Secretariat has the capacity to accommodate only 2 activities for the next year. One solution is to create connections between proposals --> speakers can get in touch with the Scientific Secretariat to help making those connections.
- All participants will send their comments/observations/suggestions within one week from today.

[LINK](#) to the list of publication shared during the call

### **4.IRDiRC Conference & RE(ACT) Congress: 13-15 January 2021**

- Save the date for a virtual meeting

**Participants – 01 October 2020**

Lucia Monaco – CA Chair, Telethon Foundation, Italy  
David Pearce – CA Vice Chair, Sanford Research, USA  
Adam Hartman – National Institute of Neurological Disorders and Stroke (NINDS, NIH), USA  
Alexandre Mejat – French Muscular Dystrophy Association (AFM – Telethon), France  
Alaa Hamed – Sanofi Genzyme, USA  
Anne Pariser – National Center for Advancing Translational Sciences (NCATS, NIH), USA  
Anthony Brooks – Leicester University, UK  
Catherine Nguyen – National Institute of Health and Medical Research (INSERM), France  
Catherine Berens – European Commission, Belgium  
Christian Rubio – Global Genes, USA  
Christina Kyriakopoulou – European Commission, Belgium  
Christopher McMaster – Canadian Institutes for Health Research (CIHR), Canada  
Cindy Bell – Genome Canada, Canada  
Diego Ardigò – Chiesi Group, Italy  
Dixie Baker – Martin, Blanck & Associates, USA  
Domenica Taruscio – Istituto Superiore di Sanità (ISS), Italy  
Durhane Wong-Rieger – Rare Diseases International (RDI), Canada  
En Kimura – Japan Agency for Medical Research and Development (AMED), Japan  
Faye Chen – National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS, NIH), USA  
Florence Guillot – E-RARE Consortium, France  
Gareth Baynam – Western Australia Department of Health, Australia  
Hélène Le Borgne – European Commission, Belgium  
Hotake Takizawa – Japan Agency for Medical Research and Development (AMED), Japan  
Ivana Cecic – Genome Canada, Canada  
Jason Wan – National Institute of Dental and Craniofacial Research (NIDCR, NIH), USA  
Katherine Beaverson – Pfizer Inc., USA  
Kelly du Plessis – Rare Diseases South Africa, South Africa  
Marie-Christine Ouillade – French Muscular Dystrophy Association (AFM-Téléthon), France  
Melissa Parisi – National Institute of Child Health and Human Development (NICHD, NIH), USA  
Oleg Kvlividze – Georgian Foundation for Genetic and Rare Diseases (GeRaD), Georgia  
Philip John Brooks – National Center for Advancing Translational Sciences (NCATS, NIH), USA  
Ramaiah Muthyala – Indian Organization for Rare Diseases (I-ORD), India  
Roseline Favresse – French Foundation for Rare Diseases, France  
Samantha Parker – Lysogene, France  
Samuel Agyei Wiafe – Rare Diseases Ghana Initiative (RDGI), Ghana  
Shi Yin Foo – Cydan II  
Sonja van Weely – The Netherlands Organisation for Health Research and Development (ZonMw), The Netherlands  
Stephen Groft – National Center for Advancing Translational Sciences (NCATS, NIH), USA  
Vanessa Boulanger – National Organization for Rare Disorders (NORD), USA  
Virginie Bros-Facer – EURORDIS-Rare Diseases Europe, France  
Virginie Hivert – EURORDIS-Rare Disease Europe, France  
Volker Liebenberg – Illumina Inc., Germany  
Younjhin Ahn – Korea National Institute of Health (KNIH), South Korea  
Yukiko Nishimura – Advocacy Service for Rare and Intractable Diseases’ multi-stakeholders in Japan (ASrid), Japan

Daria Julkowska – Coordinator, IRDiRC Scientific Secretariat, France  
Carla D’Angelo – Senior Project Manager, IRDiRC Scientific Secretariat, France  
Galliano Zanello – Senior Project Manager, IRDiRC Scientific Secretariat, France  
Katerina Tzima – Project Assistant, IRDiRC Scientific Secretariat, France  
Mary Wang – Special assistant to CA Chair, Telethon Foundation, Italy  
Tanguy Onakoy – Communication Manager, IRDiRC Scientific Secretariat, France

**Apologies – 01 October 2020**

Alba Ancochea – Ibero-American Alliance for Rare Diseases (ALIBER), Spain  
Christopher Gibson – Recursion Pharmaceuticals Inc., USA  
Daniel Lavery – Loulou Foundation, UK  
Dominique Dunon-Bluteau – Agence Nationale de la Recherche (ANR), France  
Eda Selebasto – Botswana Organization for Rare Diseases (BORDIS), Botswana  
Katherine Needleman – FDA, Office of Orphan Products Development (OOPD), USA  
Kristen Nowak – Western Australian Department of Health, Australia  
Lisa Chadwick – National Human Genome Research Institute (NHGRI, NIH), USA  
Madhu Natarajan – Takeda Pharmaceuticals, USA  
Manuel Posada de la Paz – Instituto de Salud Carlos III (ISCIII), Spain  
Nicole Millis – Rare Voices Australia, Australia  
Ning Li – BGI Group, China  
Oxana Iliach – Canadian Organization for Rare Disorders (CORD), Canada  
Prasanna Kumar Shirol – Organization for Rare Diseases India (ORDI), India  
Qi Sun – Chinese Organization for Rare Disorders (CORD), China  
Ralph Schuster – Federal Ministry of Education and Research (BMBF), Germany  
Ritu Jain – Asia Pacific Alliance of Rare Disease Organizations (APARDO), Singapore  
Sarah Bowdin – Addenbrooke's Hospital, Cambridge, UK  
Satish Gopal – National Cancer Institute (NCI, NIH), USA  
Sharon Terry – Genetic Alliance, USA  
Shefa Gordon – National Eye Institute (NEI, NIH), USA  
Sultan Turki Alsedairy – Saudi Human Genome Project, Saudi Arabia  
Tom Pulles – Ultragenyx, USA  
Willem Ouwehand – National Institute for Health Research (NIHR), UK  
Shuyang Zhang – National Rare Diseases Registry System of China (NRDRS), China

**Participants – 02 October 2020**

Lucia Monaco – CA Chair, Telethon Foundation, Italy  
David Pearce – CA Vice Chair, Sanford Research, USA  
Adam Hartman – National Institute of Neurological Disorders and Stroke (NINDS, NIH), USA  
Alexandre Mejat – French Muscular Dystrophy Association (AFM – Telethon), France  
Anne Pariser – National Center for Advancing Translational Sciences (NCATS, NIH), USA  
Anneline Jonker – TechMed Centre, University of Twente Enschede, The Netherlands  
Catherine Nguyen – National Institute of Health and Medical Research (INSERM), France  
Catherine Berens – European Commission, Belgium  
Christian Rubio – Global Genes, USA  
Christina Kyriakopoulou – European Commission, Belgium  
Christopher McMaster – Canadian Institutes for Health Research (CIHR), Canada  
Cindy Bell – Genome Canada, Canada  
Diego Ardigò – Chiesi Group, Italy  
Dixie Baker – Martin, Blanck & Associates, USA  
Domenica Taruscio – Istituto Superiore di Sanità (ISS), Italy  
Dominique Dunon-Bluteau – Agence Nationale de la Recherche (ANR), France  
Durhane Wong-Rieger – Rare Diseases International (RDI), Canada  
En Kimura – Japan Agency for Medical Research and Development (AMED), Japan  
Faye Chen – National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS, NIH), USA  
Florence Guillot – E-RARE Consortium, France  
Hélène Le Borgne – European Commission, Belgium  
Ivana Cecic – Genome Canada, Canada  
Katherine Beaverson – Pfizer Inc., USA  
Katherine Needleman – FDA, Office of Orphan Products Development (OOPD), USA  
Manuel Posada de la Paz – Instituto de Salud Carlos III (ISCIII), Spain  
Marie-Christine Ouillade – French Muscular Dystrophy Association (AFM-Téléthon), France  
Melissa Parisi – National Institute of Child Health and Human Development (NICHD, NIH), USA  
Oleg Kvlividze – Georgian Foundation for Genetic and Rare Diseases (GeRaD), Georgia  
Oxana Iliach – Canadian Organization for Rare Disorders (CORD), Canada  
Philip John Brooks – National Center for Advancing Translational Sciences (NCATS, NIH), USA  
Ramaiah Muthyala – Indian Organization for Rare Diseases (I-ORD), India  
Roseline Favresse – French Foundation for Rare Diseases, France  
Samantha Parker – Lysogene, France  
Sonja van Weely – The Netherlands Organisation for Health Research and Development (ZonMw), The Netherlands  
Stephen Groft – National Center for Advancing Translational Sciences (NCATS, NIH), USA  
Vanessa Boulanger – National Organization for Rare Disorders (NORD), USA  
Virginie Bros-Facer – EURORDIS-Rare Diseases Europe, France  
Virginie Hivert – EURORDIS-Rare Disease Europe, France  
Volker Liebenberg – Illumina Inc., Germany  
Younghin Ahn – Korea National Institute of Health (KNIH), South Korea  
Yukiko Nishimura – Advocacy Service for Rare and Intractable Diseases’ multi-stakeholders in Japan (ASrid), Japan

Daria Julkowska – Coordinator, IRDiRC Scientific Secretariat, France  
Carla D’Angelo – Senior Project Manager, IRDiRC Scientific Secretariat, France  
Galliano Zanello – Senior Project Manager, IRDiRC Scientific Secretariat, France

Mary Wang – Special assistant to CA Chair, Telethon Foundation, Italy  
Tanguy Onakoy – Communication Manager, IRDiRC Scientific Secretariat, France

**Apologies – 02 October 2020**

Alaa Hamed – Sanofi Genzyme, USA  
Alba Ancochea – Ibero-American Alliance for Rare Diseases (ALIBER), Spain  
Christopher Gibson – Recursion Pharmaceuticals Inc., USA  
Daniel Lavery – Loulou Foundation, UK  
Eda Selebasto – Botswana Organization for Rare Diseases (BORDIS), Botswana  
Gareth Baynam – Western Australia Department of Health, Australia  
Jason Wan – National Institute of Dental and Craniofacial Research (NIDCR, NIH), USA  
Kelly du Plessis – Rare Diseases South Africa, South Africa  
Kristen Nowak – Western Australian Department of Health, Australia  
Lisa Chadwick – National Human Genome Research Institute (NHGRI, NIH), USA  
Madhu Natarajan – Takeda Pharmaceuticals, USA  
Nicole Millis – Rare Voices Australia, Australia  
Ning Li – BGI Group, China  
Prasanna Kumar Shirol – Organization for Rare Diseases India (ORDI), India  
Qi Sun – Chinese Organization for Rare Disorders (CORD), China  
Ralph Schuster – Federal Ministry of Education and Research (BMBF), Germany  
Ritu Jain – Asia Pacific Alliance of Rare Disease Organizations (APARDO), Singapore  
Samuel Agyei Wiafe – Rare Diseases Ghana Initiative (RDGI), Ghana  
Sarah Bowdin – Addenbrooke's Hospital, Cambridge, UK  
Satish Gopal – National Cancer Institute (NCI, NIH), USA  
Sharon Terry – Genetic Alliance, USA  
Shefa Gordon – National Eye Institute (NEI, NIH), USA  
Shi Yin Foo – Cydan II  
Sultan Turki Alsedairy – Saudi Human Genome Project, Saudi Arabia  
Tom Pulles – Ultragenyx, USA  
Willem Ouwehand – National Institute for Health Research (NIHR), UK  
Shuyang Zhang – National Rare Diseases Registry System of China (NRDRS), China