



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 6th Consortium  
Assembly Meeting**

November 10-11, 2017



**IRDIRC**

## EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on November 10-11, 2017 in Tokyo, Japan. It was attended by 40 participants in person and 3 via teleconference, representing 29 member organizations, the Scientific Committees (SCs) and the Scientific Secretariat (Sci Sec).

1. Updates by the Chair of Consortium Assembly (CA)
  - ▶ The new IRDiRC Goals 2017-2027 were published in three articles and a press release
  - ▶ IRDiRC is planning the next face-to-face meetings for Q1-2 and Q3-4 2018
  - ▶ Six new “IRDiRC Recognized Resources” were approved
  - ▶ Nine new IRDiRC members: Recursion Pharmaceuticals, Loulou Foundation, Chinese Organization for Rare Disorders (CORD), Advocacy Service for Rare and Intractable Diseases’ multi-stakeholders in Japan (ASrid), Indian Organization for Rare Diseases (I-ORD), Canadian Organization for Rare Disorders (CORD), Rare Voices Australia (RVA), Rare Diseases South Africa (RDSA), and Cydan II
  - ▶ Various changes for representatives of members in the Companies Constituent Committee (CCC)
  - ▶ Investigation is currently ongoing regarding best dates and locations for the in-person Q1-2 2018 meeting, which will be a joint CA/SC meeting, most likely in Vienna, Austria, in May 2018
2. New member presentations
  - ▶ Members were introduced to Recursion Pharmaceuticals, CORD (China), ASrid, I-ORD, and CORD (Canada).
3. Round Table
  - ▶ IRDiRC members presented the 2-3 key things which happened in the past six months in relation to the new IRDiRC goals.
4. Updates from the Scientific Secretariat
  - ▶ The Sci Sec presented recent activities, including supporting teleconferences, tracking of the rollout plan for the goals, article writings, and the workplan for the next year, including the efforts for the upcoming Task Forces.
  - ▶ The IRDiRC website is currently undergoing a redesign and will be released shortly.
5. Next CA face-to-face meeting: request for hosts
  - ▶ Hosts are requested for the next face-to-face meetings, i.e. for the meetings in Q3-4 2018, Q1-2 2019 and Q3-4 2019
6. IRDiRC Constituent Committees
  - ▶ The IRDiRC Constituent Committees reported back from their break-out sessions, and presented their top priority actions

- ▶ Patient Advocates Constituent Committee (PACC)
  - Top priority actions:
    - Creation of a federated DB of metadata from registries
    - Creation of a position statement including specific recommendations for how to apply the second new IRDiRC goal (1000 new therapies) to all nations
    - Identification of barriers to patient/participant engagement in RD research and developing recommendations to remove them
- ▶ Companies Constituent Committee (CCC)
  - Top priority actions:
    - Better and more natural history and registry studies
      - 2-day workshop to build simulated cases and discuss recommendations on how to build an “ideal” system for use in the real world
      - Build a universal, broad-based platform that can be used by everyone based on existing case studies, covering different technologies and therapeutic areas
  - Common infrastructure to drive research in new rare diseases
- ▶ Funder Constituent Committee (FCC)
  - Top priority actions:
    - Investigation of tools to get a better overview of the full rare diseases funding landscape, in order to build a coordinated, strategic research funding agenda
    - Develop recommendations in the context of clinical research networks for rare diseases
  - The FCC discussed current funding opportunities for ethical, legal and social implications of research, and potential gaps to overcome in the future

## 7. IRDiRC Scientific Committees

- ▶ Diagnostics Scientific Committee (DSC)
  - Top priority actions:
    - Access to testing is an important topic, and should be included in the DSC roadmap
    - Solving the Unsolved Task Force is launched, and had its first conference call
    - Clinical Data Sharing for Gene Discovery Task Force is expected to launch shortly
  - Potential Task Force proposal is currently being developed on Indigenous Populations and Developing Countries
- ▶ Interdisciplinary Scientific Committee (ISC)
  - Working on strengthening the committee, due to departure of various members; opportunity to bring on members with informatics knowledge
  - Top priority actions:
    - Integrating research into care
    - Setting the stage for clinical trials
    - Assessing impact and overarching actions
- ▶ Therapies Scientific Committee (TSC)

- Top priority actions:
  - Definition of a new master plan for the development and registration of innovative drugs specific for rare diseases
  - Definition of a research framework and a business model for repurposing
  - Definition of standards for use of data collected in health care practice for real-world evidence generation
  - Reframing of the current international research agenda for rare diseases

## REPORT

### 1. Welcome and Chair's activity report

#### 1.1 Welcome

The Chair of the Consortium Assembly (CA) thanked the Japan Agency for Medical Research and Development (AMED) for hosting the 6<sup>th</sup> Consortium Assembly face-to-face meeting and welcomed participating members.

#### 1.2 Activities and progress: June-November 2017

- ▶ IRDiRC Goals roll-out
  - Team effort for write-up of three articles, including a high-profile short Nature Commentary, and two longer CTS papers: a CTS Past Perspective and a CTS Future Perspective – *See Supplementary documents for final versions of the CTS papers circulated for this meeting*
    - Papers are not only important for IRDiRC, but also for rare diseases research, in general
    - Articles were released simultaneously with a press release by the IRDiRC Sci Sec
  - Members disseminated the new goals to their networks, via various forms of communication, in order to maximize publicity
  - Sci Sec tracked the result of the rollout plan, including interviews, web pages, articles and social media – *see Supplementary doc circulated for this meeting*

#### 1.3 IRDiRC CA 2018 meetings

- ▶ Investigation ongoing regarding best dates and locations for the in-person Q1-2 2018 meeting, which will be a joint CA/SC meeting
  - Sci Sec will investigate the possibility of organizing the next CA face-to-face meeting connected to ECRD, on May 14-16 in Vienna, Austria, due to the convenience of the many CA members that are planning to attend this conference.
- ▶ Generally, the Q3-4 meetings are CA meetings, with CA members and Chairs/Vice Chairs of the Scientific Committees present, and Q1-2 meetings are joint CA and Scientific Committees meetings

#### 1.4 Approval of “IRDiRC Recognized Resources”

- ▶ 6 new “IRDiRC Recognized Resources” applications have been approved
- ▶ New resources via collaboration with the Human Variome Project:

- Mutalyzer
- Leiden Open Variant Database (LOVD)
- Human Genome Variation Society (HGVS) Nomenclature
- Genetic/Disease Specific Variant Database Quality Parameter guidelines
- ▶ Approval of “Guidelines for diagnostic NGS”, applied by Gert Matthijs
- ▶ Approval of “FAIR Guidelines”, applied by Marco Roos

### 1.5 IRDiRC membership changes

- ▶ Two changes in the Companies Constituent Committee
- ▶ Companies Committee could benefit from additional representation

→ Members asked to submit suggestions for additional companies to target to Sci Sec

### 1.6 IRDiRC membership growth

- ▶ 9 new members since the last face-to-face meeting
- ▶ Companies Constituency Committee has two new members
  - Recursion Pharmaceuticals
  - Cydan II
- ▶ Funders Constituency Committee has a new member
  - Loulou Foundation
- ▶ Patient Advocacy Constituency Committee has grown substantially, with 6 new members
  - Chinese Organization for Rare Disorders (CORD)
  - Advocacy Service for Rare and Intractable Diseases’ multi-stakeholders in Japan (ASrid)
  - Indian Organization for Rare Diseases (I-ORD)
  - Canadian Organization for Rare Disorders (CORD)
  - Rare Voices Australia (RVA)
  - Rare Diseases South Africa (RDSA)
- ▶ By welcoming these new members, IRDiRC has formal representation in new regions and continents
- ▶ New members present at this meeting introduced themselves formally later in the meeting

## 2. New member presentations

### 2.1 Recursion Pharmaceuticals

- ▶ Recursion Pharmaceuticals
  - Mission statement: to decode biology, to improve human lives
  - Drug discovery through artificial intelligence, with ultimate goal of having 100 new treatments by 2025
  - Cellular phenotype for many diseases are accessible via cellular images

- Several rare diseases can be diagnosed accurately by automatized phenotypical discovery
- Discovery can be done at cellular level
- Extraction of image-based phenotypes using computer vision, chosen because of low cost of imaging
  - About 20 terabytes of data created every week
- ▶ Platform
  - Loss-of-functions screening, by siRNA and compounds, to compare healthy versus disease cells
    - Fluorescent imaging: 1000 parameters of cellular parameters measured
    - Data analytics: machine learning approach to quantify the differences
    - Phenomics profiling: quantify what changes take place, and to determine which drug would create a similar effect
- ▶ Results
  - Technology validated by un-biased re-discovery of multiple clinical stage drugs
  - Scale, scope and pace for accelerated drug discovery, with increases expected in the next years
    - Portfolio currently 305 monogenic loss-of-function diseases
    - Screening of approximately 8 diseases a week
    - Working on hits reported by 65 diseases so far

## 2.2 Chinese Organization for Rare Disorders

- ▶ Chinese Organization of Rare Disorders (CORD)
  - Founded in 2012
  - In 2011, there were only 11 patient's organizations in China, currently there are over 80, more than half are incubated by CORD
  - Recently participated ICORD meeting in Beijing, in September 2017
- ▶ Mission and goals
  - Hope to contribute to rare diseases drug development
  - To help other patient's organizations in China
  - Very large rare disease population in China, with many people not well characterized
    - Reach out and find patients: currently 30000 to 40000 patients registered via CORD
    - Over 60% of rare disease patients in China are undiagnosed
  - Promote active patient participation in research, in order to speed up therapeutic development
- ▶ Initiatives
  - CORD launched a start-up company to help patients get a faster diagnosis
  - Involved in the newly formed Chinese Orphanet, in order to provide information to patients
  - Development of a rare disease patient registry in China

## 2.3 Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan

- ▶ ASrid
  - Founded on November 4, 2014
  - Members are researchers from academia and industry, patients and their families, entrepreneurs, engineers, physicians, medical professionals
    - Gives ASrid a position as intermediate organization, connecting different stakeholders
  - Motto: to patients, for patients, beside patients
- ▶ Goals and activities
  - Formulate guidelines
    - Quality of life of rare disease patients
    - Collaboration and cooperation in rare disease patients
  - Work on ethical, legal and social implications of rare diseases
  - Operate the J-RARE, a patient's data platform
  - Introduce global discussion to patients
    - Function as intermediary organizations as IRDiRC, RDI, ICORD, UDNI and the Japan Patients' Association and the Nanbyo support network
  - The organization of Rare Disease Day
  - Promote collaboration and cooperation

## 2.4 Indian Organization for Rare Diseases (I-ORD)

- ▶ I-ORD
  - Established in 2003, in both India and US
  - All volunteer run
  - Mission: Improve the quality of lives of people with rare diseases
  - Member of several international organizations, such as RDI, ICORD, and partner to many more, as CODR, EURORDIS, RareConnect
- ▶ Goals
  - Provide support to rare diseases' patients and patient groups;
  - Organize, develop, connect to patients-to-patients, - researchers, healthcare providers, policy makers drug developers etc.
  - Advance rare disease programs such as diagnosis, preventions, access to therapies, research collaborations
  - Gain support from public to promote rare diseases
  - Learn from International RD Advocacy groups and disseminate the information to our patients
- ▶ Challenges to RD research in India
  - Massive number of rare disease patients in India
  - Large number (> 300) of languages spoken in India



- 60% rural population
- >40% adult illiteracy
- Costs of drugs

## 2.5 Canadian Organization for Rare Disorders (CORD)

### ▶ CORD

- Canada's national network for organizations representing all those with rare disorders
- Founded in mid 1990's, originally as patient support group
- Currently contains about 100 member organizations in Canada
- Emphasis on building a cohesive rare disease community in Canada, that is partnered with various national and international patient organizations

### ▶ Goals and activities

- Represents the orphan disorders community in the development of Canadian Orphan Drug Policy
  - Focus on policy development for rare diseases, as there is still not an orphan drug framework in Canada
  - Working on strategies for better reimbursement of drugs used to treat rare diseases
- Promoting innovative research, in order to encourage research and development in Canada
  - Encouraging investments in clinical trials in Canada, and working to ensure Canada's Clinical Trials Registry works effectively for those with rare disorders
  - Development of better research protocols and clinical trials protocols
  - CORD is a small funding partner for research, collaborating with CIHR
  - Stimulating the establishment of research sites
- Improving early detection and prevention
  - Assisting in the registration of Canada's rare disease population
  - Development registries for rare diseases
- Patient's participation to research
  - Base of patients that can participate in research

### 3. Round Table

Representative of each member organization was asked to present 2-3 key things which happened in the past six months that this group could benefit from knowing and relevant to the new IRDiRC goals.

### 4. Updates from the Scientific Secretariat (Sci Sec)

#### 4.1 Activities update February-November 2017

The Coordinator of the Sci Sec updated members of activities from the past 9 months, including:

- ▶ Teleconferences: organization, document preparation, report writing (on average 1 per week)
- ▶ Fortnightly calls with the Chair of CA: exchange of updates and information on activities
- ▶ Workshops and meetings: venue and logistics, travel organization, document preparation, report writing, reimbursements
- ▶ Metrics tracking: therapies, new genes and diagnostic trackers
- ▶ State-of-Play Report: 2 parts, delayed due to changes in the Sci Sec
- ▶ Communication: redesign website, updated standard slide decks
- ▶ Press release and coordinated roll out plan articles Goals 2017-2027
  - Systematic tracking of outcome roll out plan – *supplementary document*
- ▶ Proofing, submission, and acceptance of IRDiRC Policies and Guidelines Paper

In the coming months:

- ▶ Task Forces: work started on STU Task force, work to start on potentially 4 new Task Forces while supporting ongoing ones
- ▶ Publications: ongoing efforts for a dozen or so articles/commentaries
  - Dissemination of outcomes critical

#### 4.2 Scientific Secretariat team update

- ▶ Anneliene Jonker is replacing Lilian Lau as Project Manager
- ▶ Marlène Jagut will continue as Communication Manager
- ▶ Lilian Lau will continue working with IRDiRC on specific projects, as contractor of Dr Austin, in tandem with Christine Cutillo

#### 4.3 Re-design of IRDiRC website

- ▶ Re-organization of content
  - Pages dedicated to members and activities
  - Set-up FAQ page
- ▶ Creation of scientific contributions page
  - Collection and compilation of list of experts that show an interest in IRDiRC

- ▶ New features
  - Search box
  - Possibility to connect to Twitter
  - Possibility to subscribe to newsletter
- ▶ New website is expected to go live in January

## 5. Next CA face-to-face meetings: request for hosts

Hosts are needed for upcoming face-to-face CA meetings, in particular:

- ▶ Q3-4 2018
- ▶ Q1-2 2019
- ▶ Q3-4 2019
  - The Dept of Health Western Australia proposes to host a meeting, if survey is positive regarding the willingness of CA members to travel to Australia

Hosts will cover the costs of meeting room(s), equipment (and technicians), and hospitality. Participants will cover their own travel and accommodation. Potential host(s) are requested to contact the Sci Sec.

→ Sci Sec will run a doodle poll to determine willingness of CA members to travel to Australia

## 6. IRDiRC Constituent Committees

### 6.1 Patient Advocates Constituent Committee (PACC)

- ▶ Last few months have been focused on bringing on new members to the committee, and more members are expected to join shortly, to further strengthen the committee
  - Members attending from Australia, North America, Europe, Asia
- ▶ PACC presented their action plans for the new IRDiRC Roadmap. Top priority actions:
  - Creation of a federated DB of metadata from registries, that is people centered, global, cross condition, cross-funder, cross-country, cross-company
    - Steps in this proposal are the creation of a list of registries to be assessed, consensus on what constitutes an adequate database, an analysis of registries and registries and resulting data, the identification of gaps and consultant to determine fields
    - This proposal will require the hiring of a consultant to assist with the set-up
  - Creation of a position statement including specific recommendations for how to apply the second new IRDiRC goal (1000 new therapies) to all nations
    - Steps in the writing of this position statement are an assessment of the state of approvals in the various regions, building on existing harmonization consortiums, and the write-up of recommendations for reducing the need to approve therapies in multiple nations

- Identification of barriers to patient/participant engagement in RD research and develop recommendations to remove them
  - Steps in this identification procedure are the creation of a survey to patients and clinicians with attention to barriers to participation, a second survey on patient engagement in research, which should lead to the determination of an incentive structure for encouraging responses to the surveys, mindful of cultural differences
- ▶ Due to the newly defined plans of the PACC and the Consortium as a whole, the PACC thinks that the activities included in the current form of the Patient Engagement Task Force are within the context of the PACC and part of a wider, more expansive effort in the patient involvement arena.

## 6.2 Companies Constituent Committee (FCC)

- ▶ Focus of the CCC is to bring more and better therapies to the market, what are the gaps that need to be overcome in order to achieve that goals
  - CCC presented an action plan on key activities they think they need to invest in, in order to contribute to this focus
  - Four focus areas were identified that can be implemented in the pre-competitive space
- ▶ CCC presented their action plans for the new IRDiRC Roadmap. Top priority actions:
  - Better and more natural history and registries studies are needed for drug development, in order to be of use for clinical trials
    - Build an universal, broad-based platform that can be used by everyone, ideally based on existing cases, covering different technologies and therapeutic areas, that can actually drive decisions
    - Recommendations on how to build an ideal system for use in the real world, and recommendations of standards and best practices in data collection
      - Possibility of cooperation
        - Partnership possibilities between TSC, CCC, and FCC
        - Partner with organizations involved in providing such infrastructure, i.e. focus on this becoming a multi-stakeholder adventure
  - Building of a central clinical trial data repository, to submit clinical trials including failed investigations
    - Many companies have data sitting in the company that is no longer useful for them, but can contribute to other future or ongoing investigations
    - Potential to use data as placebo dataset
  - Set-up of a common infrastructure to drive research in new rare diseases
    - Currently many factoids are surrounding rare diseases: time to get clear picture of information, including prevalence data, that has often been incorrect
      - Factoid data allows the same 100-200 diseases to be consistently selected for drug development, whereas other diseases, with good quality data also presents opportunities for development

- Possibility to build a complete list of rare diseases
  - Build a complete list of rare diseases, based on existing databases, as OMIM and Orphanet
  - Build additional information including prevalence, geographical area, tissues affected
- To inform areas of focus for research, and thereby spread the field for drug development possibilities, by providing well-controlled data

### 6.3 Funders Constituent Committee (FCC)

- ▶ The Funders Constituent Committee (FCC) would like to get a better overview of the full rare diseases funding landscape
  - They would like to create a real-time tracking database for past, current, and upcoming calls in order to build a coordinated, strategic, international research funding agenda
  - A potential collaborative platform, between Orphanet and Uber Research was presented, costing 60K euros in Y1 and 25K euros in Y2-5
    - If approved by the CA, this activity could be included in the current SUPPORT-IRDiRC contract. If so, it will require a contract amendment and will need to follow subcontracting protocols, meaning that it will not begin until mid-late 2018. FTE to support this work is built into the current SUPPORT-IRDiRC contract, but unsure about the future EJP funding.
- ▶ Future activities to tackle the goals were presented, primarily focused on the presentation of the European Joint Programme Cofund (EJP) on Rare Diseases
  - The IRDiRC Scientific Secretariat will be part of the future EJP
  - The EJP gives the possibility to do a RD Challenge, an opportunity for companies to collaborate to the EJP
- ▶ International collaboration for clinical research networks is more and more important
  - Presentation of plans for Clinical Rare Diseases Research Networks: RDCRNs in the US, ERNs in Europe, other equivalents in different parts of the world
  - How to establish connections and bring international partners in?
  - How to establish durable international collaborations, which are the obstacles now
  - A Task Force proposal will be proposed for approval by the CA
- ▶ Ethical, legal and social implications (ELSI) are aspects of rare diseases research that has recently received increased attention
  - A number of countries have funding calls on ELSI aspects
  - Discuss ways to reference ELSI in RD research
  - Opportunity to learn and see how this can be projected to other counties, and opportunity to reduce redundancy
  - The FCC discussed current funding opportunities for ELSI research, and potential gaps to overcome in the future.

## 7 IRDiRC Scientific Committees

### 7.1 Diagnostics Scientific Committee (DSC)

- ▶ DSC members presents currently all continents in the world
- ▶ Four items were presented that prevents goal 1 from being achieved, as well as a Task Force proposal that will undergo further development. Top priority actions:
  - Access to testing
    - How do we create pathways to testing, in order to diagnose rare disease patients?
      - Referral to subspecialists: how to create red-flags in the testing system, to allow for an efficient referral procedure?
      - Indigenous population: set up culturally safe procedures, taking into account the differences in languages
      - Developing countries: issues and items are overlapping with indigenous populations
    - Reimbursement
      - Evidence collection for HTA
      - Qualitative and quantitative health economics
      - Importance of rare disease coding, as only 3.5% of rare diseases have a specific code
  - Clinical data sharing is an already approved, but not yet started Task Force
    - Majority of genome-wide sequencing is now occurring in the clinic – data is being siloed
    - Suggested tools in this are (some of them originating from the interdisciplinary field):
      - Consent templates
      - Dynamic consents
      - Clinical interface development e.g. Phenotips, Patient Archive
      - Data exchange developments
      - Patient facing interfaces – interaction with Patient Advocates Committee
    - Possibility collaborate with other Task Forces
  - Solving the Unsolved (STU) is currently active
    - Bring together experts in each of the areas we suspect are contributing to rare diseases and patients/families that remain unsolved following WES analysis
    - First TC was held end of September
    - Members are expert in various areas of unsolved rare diseases
    - Several TCs are organized, prior to a workshop on March 25 in Cambridge, UK
  - Translation of new technologies
    - Horizon scan of new technologies that will be introduced to the clinic for either diagnostics or screening

- Potential Task Force proposal on Indigenous Populations and Developing Countries
  - Globally this includes most rare disease patients
  - Mapping of existing programs, tools and access
  - Stimulate local genomic centers/ capacity building
  - Interdisciplinary UDP as a “low cost model” – patient centric phenotyping, workflow and ethos

## 7.2 Interdisciplinary Scientific Committee (ISC)

- ▶ Membership
  - Currently 6 new members proposed that bring expertise in data and informatics, as particularly important in the field of interdisciplinary
- ▶ The ISC presented action plans around the three goals, and overarching actions. Top priority actions:
  - ▶ Integrate Research into care
    - Possibility to connect care to UDNI
    - Setting up training for rare diseases experts in research
    - Promote registries and natural history studies, as presented also by CCC
    - Promote newborn screening - Short windows to diagnosis, collaborate with Diagnostics Committee?
  - ▶ Setting the stage for clinical trials
    - Promote high impact natural history studies
    - Clinical research infrastructure
    - Non-traditional trial designs (adaptive, platform) – Therapeutics Committee is already currently addressing
  - ▶ Assess impact, by:
    - Post-approval data collection
    - Assessing impact and burden
    - Creating training opportunities
    - Setting up sustainability and health Systems
  - ▶ Overarching actions that are prioritized are
    - Data sharing – standards clearing house, when and how to share patient-level data, and consent guidelines. This is a proposal by an ISC member that connects standards to use cases
    - Biobanking - standards and ethical/legal guidelines
    - Regulatory harmonization – IRDIRC to engage international regulatory agencies, in collaboration with other committees

## 7.3 Therapies Scientific Committee (TSC)

- ▶ The TSC presented their extensive methodology, through which they reached their action plans, through a gap prioritization exercise

- Prioritization asked for each key action and assessment on manageability and impact
- ▶ Strategic action plans of the TSC are grouped around four themes:
  - Support the definition of a new master plan for the development and registration of innovative drugs specific for rare diseases
    - TF on patient engagement in research
    - Follow-up WS on small population trials with case studies and simulated cases to be “solved” in a multi-stakeholder group
    - Follow-up workshop on PROMs focusing on how to build an endpoint that satisfies all stakeholders
    - TF for drawing the master plan by development phase (what to do by when) and for the identification of key elements to be considered for rare diseases R&D
    - Follow-up workshop to deliver a rare diseases drug development handbook
    - Selected follow-ups on specific key elements (~ to PCOM or Patient Engagement TFs)
  - Support the definition of a research framework and a business model for repurposing of existing drugs specific for rare diseases
    - Follow-up WS on repurposing with case studies and simulated cases to create a “taxonomy” of repurposing cases
    - TF on how to fund and conduct drug development based on repurposing
    - Follow-up activities on specific topics raised by the first two activities.
  - Support the definition of standards for use of data collected in health care practice for real-world evidence generation, in particular for disease understanding and treatment monitoring
    - TF on scientific value and required standards for real-world evidence -based medical research
    - Follow-up action: generation of an IRDiRC position statement on value of real-world evidence research
    - TF on the use of real-world evidence data for conducting natural history studies valuable for multiple stakeholders
    - TF of the use of real-world evidence in drug development (with case studies, simulated cases, cost gain analyses, data ownership and handling models, etc.)
    - WS on the medical decision making value of real-world evidence research (requirements and mode of use of these data)
  - Support the reframing of the current international research agenda for rare diseases pushing for focusing research efforts and funding
    - Internal IRDiRC consultation to identify the perceived priorities in rare disease research to enable achievement of goal 2 and requiring public-private partnerships
    - Workshop with selected external experts to identify:
      - Key needs for each of the prioritized areas of research
      - Suitable model for multi-stakeholder collaboration in the area
    - Creation of a recommended list of priorities and mode of funding/ collaboration



- Periodic monitoring and revision of the IRDiRC priority recommendations
- ▶ Task Force updates
  - Patient-centered outcome measures in rare diseases
    - Recommendations published by Thomas Morel and Stefan Cano
  - Small population clinical trials and Data Mining and Repurposing
    - Articles pending by Simon Day and Scientific Secretariat, Virginie and Diego respectively
  - Patient-Engagement in Research Task Force
    - Members identified, leadership transferred to PACC

## 8. Task Force proposals for discussion and consideration

### 8.1 Model Consent Clauses for RD Research Taskforce proposal

- ▶ Proposal by ISC
- ▶ Objective of Task Force is to develop rare diseases research consent clauses that are
  - International and interoperable
- ▶ Multi-stakeholder group to conduct workshop, with presence of:
  - Researcher, patient and funder perspectives
  - Clinical, data, legal, ethical and policy expertise
- ▶ All CA members voted in favor of the proposal, and a request for nominations for Task Force members will be send out shortly

### 8.2 Clinical Research Networks for Rare Diseases

- ▶ Proposal by FCC Chair, TSC Vice-Chair and ISC Chair, Task Force to be headed by ISC
- ▶ Objective of the Task Force is to develop recommendations on guiding principles for policies on clinical research networks within an international context for collaboration and interoperability
- ▶ Build on experience gained and ongoing initiatives:
  - CRN US
  - ERN Europe
  - E-Asia
  - Australia
- ▶ Members suggested that the proposal, in its current form, does not include recent developments of the European clinical research networks
  - Task Force proposal needs to be re-scoped in order to take into account changes, before brought for vote again

## Actions and deliverables

- ▶ Members asked to submit suggestions for additional companies to target to Sci Sec
- ▶ Inform the Sci Sec of any current/upcoming calls for proposals

- ▶ Inform the Sci Sec when presenting on IRDiRC
- ▶ Discuss the results of the presented key actions from all CC/SC
- ▶ Send out poll re possibility to travel to Australia
- ▶ Organize next face-to-face meeting in Q1-2 2018

**Annex - List of participants**

<b><u>Members</u></b>	<b><u>Representative</u></b>
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin (Chair)
Western Australian Department of Health, Australia	Hugh Dawkins (Vice Chair)
Rare Voices Australia, Australia	Nicole Millis
Canadian Institutes of Health Research (CIHR), Canada	Paul Lasko
Canadian Organization for Rare Disorders, Canada	Durhane Wong-Rieger
BGI, China	Ning Li, Nicolas Li
Chinese Organization for Rare Disorders, China	Kevin Huang, Rachel Yang
E-Rare Consortium, Europe	Daria Julkowska
European Commission, DG Research and Innovation, EU	Iiro Eerola
Academy of Finland, Finland	Heikki Vilen
EURORDIS-Rare Diseases Europe, France	Béatrice de Montleau
Federal Ministry of Education and Research, Germany	Ralph Schuster
Indian Organization for Rare Diseases, India/USA	Ramaiah Muthyala
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore di Sanità, Italy	Domenica Taruscio
Telethon Foundation, Italy	Lucia Monaco
Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid), Japan	Yukiko Nishimura
Japan Agency for Medical Research and Development (AMED), Japan	Makoto Suematsu, Takeya Adachi
Saudi Human Genome Project, Saudi Arabia	Sultan Turki AlSedairy
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
National Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández
Roche, Switzerland	Mathew Pletcher
Genetic Alliance, USA	Sharon Terry
National Human Genome Research Institute (NHGRI), USA	Lu Wang, Teri Manolio
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman
Pfizer, USA	Sabine Gilliam
Recursion Pharmaceuticals Inc, USA	Tim Considine
Sanford Research, USA	David Pearce

<b>Scientific Committees</b>	
Diagnostics	Kym Boycott, Gareth Baynam
Interdisciplinary	Petra Kaufmann, Domenica Taruscio
Therapies	Diego Ardigò, Virginie Hivert

<b>IRDIRC Scientific Secretariat</b>	
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