



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 5th Consortium
Assembly Meeting**

October 6, 2017



IRDIRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on October 6 2017, via web/teleconference. It was attended by 34 participants representing 24 member organizations, the Scientific Committees (Sci Comms) 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 Spring and Fall 2018
- ▶ Five new “IRDiRC Recognized Resources” were approved
- ▶ Six new IRDiRC 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) and Rare Diseases South Africa (RDSA)
- ▶ Various changes for representatives of members in the Companies Constituent Committee (CCC)
- ▶ Hugh Dawkins is re-elected for his second term as CA Vice Chair

2. Draft Tokyo Agenda

- ▶ Meeting will be held on Friday-Saturday November 10-11
- ▶ For round table discussion, members are asked to send in key actions to Sci Sec, using the new round table template
- ▶ Session dedicated to strengths and challenges in RD research in Japan will be held in order to take advantage of the diverse geographical locations of CA meetings
- ▶ Parallel Constituent Committee breakouts will also take place, followed by a wrap-up/next steps discussion, in which the priorities and strategies are discussed on the second day

3. IRDiRC Constituent Committees

- ▶ Patient Advocates Constituent Committee (PACC)
 - 6 new members joined, targeting other umbrella organizations to encourage to join shortly
 - Working on new actions for the committee by establishing the PACC roadmap
- ▶ Funders Constituent Committee (FCC)
 - Starting an iSearch pilot, to determine use of this tool
 - Working on three different topics for face-to-face meeting
 - How FCC tackles the new IRDiRC goals
 - International collaboration in rare disease research networks
 - Ethical, legal and social implications of rare disease funding
- ▶ Companies Constituent Committee (CCC)
 - Looking for a Chair to lead the committee
 - Working on new actions for the committee by establishing the CCC roadmap

4. IRDiRC Scientific Committees

- ▶ Diagnostics Scientific Committee (DSC)
 - 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
 - Working on new actions for the committee by establishing the DSC roadmap
- ▶ Interdisciplinary Scientific Committee (ISC)
 - Working on strengthening the committee, due to departure of various members; opportunity to bring on members with informatics knowledge
 - Working on new actions for the committee by establishing the ISC roadmap
 - Taking part in the Patient Engagement in Research Task Force
- ▶ Therapies Scientific Committee (TSC)
 - New members joined the committee, representing the European Reference Networks and registries
 - Working on a Gap analysis to set up new actions for the committee, and establishing the TSC roadmap
 - New counter metrics for orphan drugs identified; will be presented in Tokyo
 - Publication accepted on the topic of Patient-Centred Outcome Measures (PCOM)
 - Patient Engagement in Rare Diseases Research Task Force is about to start; leadership and Steering Committee members have been identified
- ▶ SC memberships vote
 - One new DSC member was approved following a recent online vote
 - Two DSC and one TSC renewals were approved during the call

5. Members' commitment

- ▶ Members have reported back on high-level member investment, reporting the total costs of rare diseases investment and the number of projects they invested in
 - All members that provided data have met the minimum commitment required
 - Members that have not yet provided their data, are requested to do so

6. IRDiRC Scientific Secretariat

- ▶ The Sci Sec presented recent activities, including supporting teleconferences, tracking of the rollout plan for the goals, article writings, etc
- ▶ The Sci Sec will focus on, among other activities, the organization of the next face-to-face meetings, article writings, and a poster presentation at rare 2017.
- ▶ A number of organizational changes were recently made to the Sci Sec team to better serve IRDiRC members and committees

REPORT

1. Updates by the Chair of Consortium Assembly (CA)

1.1 Operating Committee (OC) activities and progress and June - September 2017

- ▶ IRDiRC Goals roll-out plan
 - 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
 - Articles were released simultaneously with a press release by the IRDiRC Sci Sec
 - Final versions of papers will be distributed when available
 - ▶ Members disseminated the new goals to their networks, via various forms of communication, in order to maximize publicity
 - ▶ The Scientific Secretariat tracked the result of the rollout plan, including interviews, web pages, articles and social media – *see Supplementary doc circulated for this call*
- Sci Sec will distribute the final versions of the papers, when available

1.2 IRDiRC CA 2018 meetings

- ▶ Investigation ongoing regarding best dates and locations for the in-person spring 2018 meeting, which will be a joint CA/SC meeting
- ▶ Host is still being sought for the fall CA meeting
- ▶ Generally, the fall meetings are CA meetings, with CA members and Chairs/Vice Chairs of the Scientific Committees present, and Spring meetings are joint CA and Scientific Committees meetings
- ▶ Potential to look into organizing the fall CA meeting in Australia, but there were concerns about distance

1.3 Approval of “IRDiRC Recognized Resources”

- ▶ 5 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

1.4 IRDiRC membership growth

- ▶ Patient Advocacy Constituency Committee has grown substantially, with 6 new members, thanks to dynamic leadership of PACC Chair
 - Chinese Organization for Rare Disorders (CORD): Rufang Huang
 - Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid): Yukiko Nishimura
 - Indian Organization for Rare Diseases (I-ORD): Ramaiah Muthyala
 - Canadian Organization for Rare Disorders (CORD): Durhane Wong-Rieger
 - Rare Voices Australia (RVA): Nicole Millis
 - Rare Diseases South Africa (RDSA): Kelly du Plessis
- ▶ By welcoming these new members, IRDiRC has formal representation in new regions and continents
- ▶ New members will introduce themselves formally at the Japan CA meeting

1.5 IRDiRC membership changes

- ▶ Two changes in the Companies Constituent Committee
- ▶ Companies Committee could benefit from additional representation
- ▶ Companies Committee currently has no Chair, and a volunteer is requested to step forward

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

→ Members asked to submit suggestions for Chair to Sci Sec

1.6 Vice Chair mandate

- ▶ CA Vice Chair's mandate will come to an end in November. He has been re-elected for an additional term, and his mandate is extended for an additional three years.

2. Draft Tokyo Agenda

2.1 Agenda

- ▶ Meeting will be held on Friday-Saturday November 10-11
- ▶ Day 1 is dedicated to the roundtable (morning) and strengths and challenges in RD research in Japan (afternoon)
- ▶ Day 2 will have parallel Constituent Committee breakouts (morning), followed by a wrap-up/next steps discussion (afternoon), in which the priorities, strategies, and roadmap are discussed.

2.2 Round Table

- ▶ During the round table, members are asked to present 2-3 activities that made significant headway in the last six months that advance the IRDiRC mission
 - For example, results of a new funding call, or a new RD drug brought to the market
- ▶ Round table discussion time has been extended in order to adequately provide the opportunity for members to present their work
- ▶ Round table topics should be presented thematically around the goals, so that progress on the goals can be tracked systematically
- ▶ In order to structure the discussion, a round table template is created, in which members are asked to send in their key events/ actions, classified by the IRDiRC goal
 - Template is simple, and will allow the Sci Sec to collect and interpret actions more easily

→ CA members asked to submit the information that they plan to update during the round table to Sci Sec, by October 30

2.3 Japan-focused session

- ▶ In this session, specific items and overarching questions pertaining to RD research in Japan will be discussed; an initiative to make use of the diverse geographical locations of the CA meetings
- ▶ Overall, the session is supposed to be a two-way information exchange: informing IRDiRC on the state of rare disease research in Japan, and supporting Japan in their rare disease research activities.
 - Listening and learning in addition to providing suggestions/thoughts on how to further rare disease research development in Japan

2.4 Action template

- ▶ Each Committee Chair was charged to discuss methodologies, strategies and timeline of activity implementation with their Committees to move toward the three new goals for 2017-2017.
- ▶ Based on the Funders Constituent Committee template, an action template has been created, which the Sci Sec has developed to instigate discussion
- ▶ Once each of the Committees has designated its activities, overlap and shared interests will be determined and IRDiRC as a whole will move forward with the top priority actions after taking into consideration the limited resources of the Sci Sec. The coalesced information will be the foundation for the IRDiRC roadmap.
- ▶ Committee Chairs will be asked to present the outcome of the action methodology discussions and their proposed roadmap of activities for their respective Committee during the face-to-face meeting in Tokyo

→ Each committee except the FCC, who has previously completed the exercise, has been asked to fill out the template by different due dates, prior to **October 23**

3 IRDiRC Constituent Committees

3.1 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
 - Great step forward to welcome several new umbrella patient groups from previously unrepresented countries
- ▶ At present, the PACC is working on the template, determining how they can impact the goals in a measurable and meaningful way
 - Align and integrate activities of PACC with existing efforts of other organizations
 - Identify gaps in current landscape and identify points along the translational spectrum where there are strengths/weaknesses.
 - Such an exercise could point to areas where tools/technologies/approaches would improve the process of rare diseases research.
- ▶ A PACC conference call is organized in mid-October to
 - Introduce new members
 - Work on action template to determine priority actions
 - Determine how the PACC will work, as a whole, moving forward

3.2 Funders Constituent Committee (FCC)

- ▶ A FCC call was held at the end of September
- ▶ A working group was set up to pilot and determine if iSearch is useful for FCC's purpose
 - Ask a number of questions and do a limited number of analyses of the funding landscape
 - Piloting working group to come up with first questions and identify how to utilize tools
- ▶ Agenda in preparation for Tokyo by various volunteers
 - How the FCC tackles the new goals?
 - What are the new programs and ongoing initiatives?
 - Occasion to present the European Joint Programme on Rare Diseases
 - International collaboration on RD Clinical Research Networks
 - 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
 - Ethical, legal and social implications (ELSI) of RD research funding, RD healthcare, and therapies
 - A number of countries have funding calls on ELSI aspects
 - Discuss ways to reference ELSI in RD research
 - Study use cases
 - Opportunity to learn and see how this can be projected to other countries

3.3 Companies Constituent Committee (CCC)

- ▶ CCC is currently without a Chair, but CCC members are asked to fill out the action template, to identify topics for action shortly

4 IRDiRC Scientific Committees

4.1 Diagnostics Scientific Committee (DSC)

- ▶ Two Task Forces are currently ongoing, 1 active, 1 about to launch
 - Solving the Unsolved (STU) is currently active
 - First TC was held end of September
 - Members are expert in various areas of unsolved RD
 - Several TCs are organized, prior to a workshop on March 25 in Cambridge, UK
 - Clinical Data Sharing for Gene Discovery will launch shortly
- ▶ DSC will have a TC in mid-October to discuss the action plans moving forward, that will be presented in Japan
- ▶ [Post-meeting note: DSC now includes representations from South Africa and Ecuador]

4.2 Interdisciplinary Scientific Committee (ISC)

- ▶ Membership
 - Looking to expand membership to get strong representatives with diverse disciplines on the committee
 - Add members with informatics and/or data expertise
- ▶ Opportunity for ISC to promote data sharing, coordinated with other initiatives, such as GA4GH
 - Ethical/legal: what can be included in funding announcements, to facilitate data sharing
 - Harmonization of consent clauses: proposal in preparation for submission to the CA
- ▶ Taking part in the Patient Engagement Task Force

4.3 Therapies Scientific Committee (TSC)

- ▶ Membership
 - TSC has grown with new members, expanding to include regulatory expertise and representation from the European Reference Networks
- ▶ Task Force activities recently completed
 - Patient-Centered Outcome Measures
 - Small-Population Clinical Trials
 - Data Mining and Repurposing

- ▶ Opinion piece by Thomas Morel accepted in the OJRD describing the outcomes of the PCOM Task Force
 - ▶ Article will be circulated upon publication
- ▶ Two Task Forces about to start
 - Patient Engagement in Rare Diseases Research
 - Activities about to commence
 - Leadership and Steering Committee defined
 - Joint effort of several committees: ISC, TSC, PACC, FCC
 - Workshop planned in quarter 1, 2018
 - Clinical Rare Diseases Research Networks
 - Proposal about to be finalized
 - Will be submitted for approval in Tokyo
- ▶ Gap analysis ongoing to set up a new action plan for the TSC
 - First results will be presented in Tokyo
- ▶ Counter metrics redefined
 - Proposal will be presented in Tokyo

5 Membership Commitment

- ▶ In June, a survey was launched to track high-level member investment, and to ensure member organizations continue to meet the minimum commitment (a criterium of membership)
- ▶ At present, 22 out of 42 members (public and private funders) reported the total costs of rare diseases investment – *document 5*
 - All members that provided a full dataset have met the minimum membership requirement (USD 10 million over 5 years).
 - Data collection is incomplete; therefore, data analyses cannot be properly interpreted as present.
- ▶ A significant increase in funding is shown between 2012 and 2013, partly due to the arrival of new members, and the availability of data covering these years.

→ CA members that have not provided their funding data are requested to send this to Sci Sec

6 IRDiRC Scientific Secretariat

6.1 Activities update June-September 2017

- ▶ 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
- ▶ Task Forces: assist with write-up or proofing articles

- Article re: recommendations of Data Mining and Repurposing TF
- Preparation of resubmission re: Small Population Clinical Trial TF
- Publication re: Patient-Centered Outcome Measures TF accepted

6.2 Activities planned for October-November 2017

- ▶ Teleconferences
 - Monthly call of the OC
 - SC: with the full committees, and/or with Chairs and Vice Chairs to plan work and monitor progress of activities of the SCs and the Task Forces
 - CC: with the full committees of the PACC and FCC
 - Task Forces: to progress work carried out to date, and commence new actions for Privacy-Preserved Record Linkage and STU
 - ▶ Organization of F2F meeting in Tokyo
 - ▶ Organization of next F2F meeting for joint CA/SCs in Spring 2018
 - ▶ Poster presentation at Rare 2017
 - Present new goals at French Rare 2017 conference
 - ▶ Members with calls for proposals: please forward to IRDiRC's Communication Manager
 - ▶ Members presenting on IRDiRC in meetings/conferences: please inform Sci Sec
- CA members asked to inform Sci Sec of calls for proposals
- CA members asked to inform Sci Sec when presenting on IRDiRC

Actions and deliverables

- ▶ Complete and submit to Sci Sec round table template by October 30
- ▶ Send in membership commitment data for members that have not previously done so
- ▶ Send suggestions for additional companies to join IRDiRC and CCC
- ▶ Inform the Sci Sec of any current/upcoming calls for proposals
- ▶ Inform the Sci Sec when presenting on IRDiRC
- ▶ Send final goals perspective articles to CA members
- ▶ Send round table template to CA members
- ▶ Send action template to all SC and CCC/PACC
- ▶ Organize next face-to-face meeting in Spring 2018

Annex - List of participants

<u>Members</u>	<u>Representative</u>
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin (Chair)
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research (CIHR), Canada	Paul Lasko
Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
European Commission, DG Research and Innovation, EU	Iiro Eerola
French Foundation for Rare Diseases, France	Roseline Favresse
Children's New Hospitals Management Group, Georgia	Oleg Kvlivdize
Indian Organization for Rare Diseases, India/USA	Ramaiah Muthyala
Shire Pharmaceuticals, Ireland	Bryan Goodwin
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
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	Senkei Umehara
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
Loulou Foundation, UK	Daniel Lavery
Genetic Alliance, USA	Sharon Terry
National Eye Institute (NEI), USA	Santa Tumminia
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	Katherine Beaverson
Recursion Pharmaceuticals Inc, USA	Tim Considine
Sanford Research, USA	David Pearce

Scientific Committees	
Diagnostics	Kym Boycott, Gareth Baynam
Interdisciplinary	Petra Kaufmann, Domenica Taruscio
Therapies	Diego Ardigò

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Marlene Jagut, Anneliene Jonker
NIH/NCATS	Christine Cutillo, Lilian Lau

Apologies

Members	Representative
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
E-Rare Consortium, Europe	Daria Julkowska
Genome Canada, Canada	Cindy Bell
BGI, China	Ning Li
WuXi AppTec Co. Ltd., China	James Wu
Rare Diseases Europe-EURORDIS, Europe	Béatrice de Montleau
Agence National de la Recherche (ANR), France	Daria Julkowska
Academy of Finland, Finland	Heikki Vilen
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Lysogene, France	Karen Aiach
Federal Ministry of Education and Research, Germany	Ralph Schuster
Istituto Superiore de Sanita, Italy	Gualtiero Ricciardi
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Yoshihiro Yoneda
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Korea National Institute of Health, South Korea	Hyun-Young Park
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Ionis Pharmaceuticals, USA	Brett Monia
Food and Drug Administration (FDA), USA	Katherine Needleman
Genzyme, USA	Carlo Incerti
National Cancer Institute (NCI), USA	Jack Welch

National Institute of Arthritis and Musculoskeletal and Skin Diseases, (NIAMS), USA	Stephen Katz
National Organization for Rare Diseases (NORD), USA	Peter Saltonstall
NKT Therapeutics, USA	Robert Mashal
PTC Therapeutics, USA	Ellen Welch



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