



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 20th Executive
Committee Meeting**

May 23, 2016



IRDIRC

EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on May 23, 2016, via web/teleconference, and attended by 28 participants representing 22 member organizations, the Diagnostics Scientific Committee (Sci Comm) and the Scientific Secretariat (Sci Sec).

1. IRDiRC Governance and Committees

- The name of the Exec Comm is changed to Consortium Assembly (CA).
- The Chair of the CA is also in the process of setting up a number of constituent committees analogous to the Sci Comms: Funders Comm, Companies Comm, and Patient Groups Comm.

2. Updates from the Chair

- “IRDiRC Recommended” will be rebranded as “IRDiRC Recognized Resources”.
- Currently held voluntary membership fees will be used to support travel of patient organization representatives and expenses related to the next IRDiRC conference; structure to support future collection is still being explored.
- Co-fund mechanism is being scoped as a model to support the Sci Sec after the SUPPORT-IRDiRC contract ends; the statement of work of the Sci Sec to be further discussed.

3. The 3rd IRDiRC Conference

- The next IRDiRC conference will take place in early February 2017 in Paris, and associated to a number of satellite events including meetings of IRDiRC committees and an E-Rare workshop.
- The constitution of a conference Planning Committee is in process.
- Funding sources include European Commission, voluntary membership fees and sponsorship.

4. IRDiRC Task Forces (TFs)

- Two new TFs of the Diagnostics Scientific Committee were approved: “Solving the Unsolved” and “Clinical Data Sharing for Gene Discovery”.
- Two current TFs will hold their workshops in winter 2016, two recently-met TFs are in writing phase, and two other TFs are not actively managed by the Sci Sec.

5. Updates from the Sci Sec

- The roles and responsibilities of each Sci Sec member were defined
- A document listing Sci Sec staff members and their specific responsibilities has been created and disseminated to IRDiRC members

6. Next Consortium Assembly face-to-face meeting will be hosted by E-Rare

- Date: September 23, 2016 – full day
- Location: Catania, Italy
- Hosted by E-Rare
- A joint IRDiRC-E-Rare meeting will be held on the afternoon prior to the CA meeting, September 22, 2016

7. Surveys for CA members to complete

- Survey on attendance to Catania: due back to Sci Sec on May 31, 2016
- Surveys on IRDiRC's impact and strategic priorities, and on State-of-Play of Research 2016 report: due back to Sci Sec on June 15, 2016

REPORT

1. IRDiRC Governance and Committees

1.1 Consortium Assembly

IRDiRC membership has grown in the past five years and so has the Executive Committee (Exec Comm); it has evolved into an assembly to exchange information with diverse input and representation. To accurately portray its structure and function, *the name of Exec Comm is changed to “Consortium Assembly” (CA)* without a change in how the organization works or its representation.

1.2 Additional constituent committees

The Chair is in the process of setting up committees analogous to the Scientific Committees (Sci Comms):

- ▶ Each committee will have a chair, committee meetings, goals; reports to the CA
 - Funders Comm: to globally coordinate rare disease efforts by international funders and provide new network opportunities
 - Companies Comm: view from companies, from collective standpoint, what stands in their way and holds back progress in rare disease progress relative to IRDiRC goals
 - Patient Groups Comm: view from patient advocates, similar aim as Companies Comm
- ▶ Added complexity to IRDiRC management and operation
 - Members need to reinforce commitments to participate and engage towards goals
 - Survey (c.f. Section 6) will help direct and optimize Sci Sec activities
- ▶ Operative process of the constituent committees
 - Implementation procedure not yet defined
 - Balanced representation and committee size not yet decided
 - Ideally same representatives in the committees as on the CA

→ A follow-up memo will be sent by the Chair to all members of the CA on the above two topics. Any suggestion or idea on implementation is also welcomed by the Chair.

2. Updates from the Chair

2.1 “IRDiRC Recognized Resources”

“IRDiRC Recommended” helps researchers identify reliable resources for rare diseases research

- ▶ “IRDiRC Recommended” will be rebranded as “IRDiRC Recognized Resources”
- ▶ Process remains the same, peer-reviewed by scientists of IRDiRC

The logo of “IRDiRC Recognized Resources”:



2.2 Voluntary membership fees management

Currently held fund at the CIHR will be used for:

- ▶ Supporting patient organization representatives to face-to-face CA meetings
- ▶ Pay for expenses related to IRDiRC conference (preferably one or two large invoices)

Structure to support future collection is still being explored:

- ▶ Genetic Alliance could help set up an account but needs account holder(s)
- ▶ For the long term, account should be held in a way that is independent of the identity of the Chair
- ▶ May roll into the next iteration of the Sci Sec, i.e. written into its statement of work
 - Note: IRDiRC will not become a legal entity

2.3 Supporting the Sci Sec

SUPPORT-IRDiRC contract ends in September 2018 but preparation for new contract is starting now:

- ▶ Co-fund mechanism is being scoped, looks promising as a model: the European Commission will fund part of contract, other organizations will co-fund the rest
- ▶ Members to reflect and include in survey the activities that should be included in the statement of work of the new contract, get the contract right and minimize post-contract modifications

→ The statement of work will be discussed in detail and prepared at the next CA face-to-face meeting.

3. The 3rd IRDiRC Conference

3.1 Some logistics

The next IRDiRC conference is an opportunity to celebrate its achievements and reflect on its future

- ▶ Expected attendance: 500-600 pax
- ▶ Venue: Paris (potential venues currently being scoped)
- ▶ Timeframe: early February 2017, ahead of Rare Disease Day
- ▶ Satellite events are envisaged:
 - IRDiRC: committee (i.e. CA, Sci Comms, constituent committees) meetings
 - E-Rare: Harmonization and data sharing workshop (organizer: CIHR)
 - Others: interested satellite event organizers should contact Daria Julkowska to include in venue price negotiation

3.2 Planning Committee

Constitution of Planning Committee (of about 10 people) is in process; it will include Operating Comm and representatives from all constituent committees.

3.3 Potential budget funding sources

A number of funding sources to support the conference:

- ▶ European Commission: travel and accommodation of invited speakers; amount to be confirmed
- ▶ Voluntary membership fees: for venue-related costs
- ▶ Sponsorship: to fill remaining budget; won't act on this until budget gaps identified

4. IRDiRC Task Forces (TFs)

4.1 New DSC proposal – Solving the Unsolved: New Approaches for Challenging Rare Genetic Disease Mechanisms

TF to get researchers together to identify top challenges in rare diseases where exome sequencing failed by at least two independent sets of investigators:

- ▶ Foster a coordinated approach to some difficult and unsolved rare diseases
- ▶ Approaches may be generalized, e.g. development of resources and tools
- ▶ Develop a scope of challenges and state-of-play, then identify targeted areas of priority
- ▶ Requirement: administrative support, workshop support for about 30 participants
- ▶ Outputs: review paper, generate collaborative groups for particular rare diseases
- ▶ The partners in this TF will bring their own resources too, e.g. cohort data

Additional notes:

- ▶ Proposed change to name: substitute “mechanisms” in title with “causes” to avoid confusion that it delves into downstream molecular mechanism
- ▶ Proposed refinement: add a sentence to the objective to clarify inclusion of results validation
- ▶ UDNI may be a TF participant but they are not particularly focused on these types of questions

4.2 New DSC proposal – Clinical Data Sharing for Gene Discovery

TF to facilitate access to clinically generated data from unsolved patients to enable discovery:

- ▶ Will not duplicate efforts of international consortia in the same space, e.g. GA4GH, G2MC
- ▶ Look at gaps between patient consent and data access in clinical care vs research setting, challenges to access, and addressing these challenges – be ahead of the curve
- ▶ Requirement: administrative support, workshop support for about 30 participants
- ▶ Outputs: position paper on needs and strategy for clinical data sharing, tools and resources to address challenge in different jurisdictions

Additional notes:

- ▶ Use case to define such data sharing policy is clear clinical need in rare diseases
- ▶ Healthcare funders to be brought together to develop relevant policy within their institutions
- ▶ Try to also involve people from personalized/precision medicine initiatives facing similar issue
- ▶ European Reference Networks (ERNs) representatives should also be involved in this TF

4.3 Update of current Task Forces

Six TFs to date:

- ▶ Active and ongoing:
 - Data Mining and Repurposing (TSC): workshop in Barcelona on 16 November 2016
 - Participant Unique Identifier (ISC): workshop tentatively in Paris in early December 2016
- ▶ In writing phase:
 - Patient-Centered Outcome Measures (TSC): restructure report, writing for publication
 - Small Population Clinical Trials (TSC): restructure report, writing for publication
- ▶ Not actively managed by IRDiRC:
 - Matchmaker Exchange (DSC): ongoing effort by MME partners
 - Automatable Discovery and Access (ISC): ongoing coordination by GA4GH

The two proposed TFs were approved by the CA:

- ▶ Process and timeline: Sci Sec will be able to commence meetings only later in the year (winter 2016), workshops next year (summer 2017)
- ▶ Budget is available to support both workshops
- ▶ [Post-meeting note: the involvement of Sci Sec in IRDiRC conference organization may affect the timeline of the TFs but Sci Sec will try to keep it as close to schedule as possible]

5. Updates of the Sci Sec

The roles and responsibilities of each Sci Sec member defined (c.f. Preparatory Document 4)

- ▶ The Coordinator will also ensure coordination with Orphanet for efficient provision of data
- ▶ A document listing contacts of Sci Sec staff members and their specific responsibilities has been created and disseminated to IRDiRC members
 - Please include cc to ensure efficient response
- ▶ Any CA member with question(s) about the status of the Sci Sec may contact the Chair

6. Next face-to-face meeting

The next CA face-to-face meeting:

- ▶ Location: Catania, Sicily, Italy (exact location to be announced in due course)
- ▶ Dates:
 - Thursday, September 22: half-day special joint IRDiRC-E-Rare meeting
 - Interested in meeting IRDiRC members
 - Present activities of E-Rare and IRDiRC
 - Joint lunch prior to the meeting and joint dinner after the meeting
 - Friday, September 23: full day IRDiRC meeting
- ▶ Hosted by E-Rare
- ▶ Members who haven't responded already were requested to complete the attendance survey sent out in late April to assist organization of the meetings

- ▶ The Chair encouraged as many members as possible to attend to keep the momentum of robust discussions going

→ A web/teleconference of the CA will also be organized for late July – early August 2016 to plan for the face-to-face meeting.

7. Surveys to complete

Three online surveys to complete:

- ▶ Attendance of CA meeting in Catania
 - Link already sent out, deadline May 31, 2016
- ▶ IRDiRC's impact and strategic priorities
 - Link and PDF will be sent out after the call, deadline June 15, 2016
- ▶ State-of-Play of Research 2016 report
 - Link and PDF will be sent out after the call, deadline June 15, 2016

→ Input to these surveys is very important so members were asked to take some time to reflect, including operational model of Sci Sec and IRDiRC on the whole, and respond to them.

Actions and deliverables

- ▶ Send follow-up memo on CA and constituent committees
- ▶ Send suggestion/idea to the Chair on implementation of constituent committees
- ▶ Contact Daria if wish to organize satellite event at IRDiRC conference
- ▶ Update TF proposals per feedback received
- ▶ Include cc in correspondence to the Sci Sec
- ▶ Organize web/teleconference of the CA
- ▶ Send survey links and questions in PDF to CA members
- ▶ Respond to surveys ahead of their deadlines

Annex - List of participants

<u>Members</u>	<u>Representative</u>
National Center for Advancing Translational Sciences, NCATS/NIH, USA	Christopher Austin, Christine Cutillo
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research, Canada	Paul Lasko
Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
E-RARE 2 Consortium, Europe	Daria Julkowska
European Commission, DG Research and Innovation, EU	Irene Norstedt, Iiro Eerola
Agence National de la Recherche, ANR, France	Daria Julkowska
French Foundation for Rare Diseases, France	Marc Tardieu
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Telethon Foundation, Italy	Lucia Monaco
Agency for Medical Research and Development (AMED), Japan	Kazuo Kawamura
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Akifumi Matsuyama, Makoto Hirose
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
National Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández
Food and Drug Administration, USA	Katherine Needleman
Genzyme, USA	Carlo Incerti
National Eye Institute, NEI/NIH, USA	Santa Tumminia
Pfizer, USA	Katherine Beaverson

<u>Invited Patient Advocacy Groups</u>	
EURORDIS, Europe	Béatrice de Montleau
National Organization for Rare Diseases, NORD, USA	Peter Saltonstall

<u>Scientific Committees</u>	
Diagnostics	Kym Boycott

<u>IRDIRC Scientific Secretariat</u>	
SUPPORT-IRDIRC Project	Ana Rath, Lilian Lau, Anneliene Jonker

Apologies

<u>Members</u>	<u>Representative</u>
European Organisation for Treatment & Research on Cancer, EORTC, Belgium	Denis Lacombe
Genome Canada, Canada	Cindy Bell
BGI, China	Ning Li
WuXi AppTec Co. Ltd., China	Mao Mao
Academy of Finland, Finland	Heikki Vilen
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Lysogene, France	Karen Aiach
Shire Pharmaceuticals, Ireland	Omar Francone
Istituto Superiore de Sanita, Italy	Gualtiero Ricciardi
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
Isis Pharmaceuticals, USA	Brett Monia
National Cancer Institute, NCI/NIH, USA	Edward Trimble
National Human Genome Research Institute, NHGRI/NIH, USA	Jeffery Schloss
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIAMS/NIH, USA	Stephen Katz
National Institute of Child Health and Human Development, NICHD/NIH, USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke, NINDS/NIH, USA	Danilo Tagle
Office of Rare Diseases Research, ORDR/NIH, USA	
PTC Therapeutics, USA	Ellen Welch
Sanford Research, USA	David Pearce

<u>Invited Patient Advocacy Groups</u>	
Genetic Alliance, USA	Sharon Terry

<u>Scientific Committees</u>	
Interdisciplinary	Hanns Lochmüller
Therapies	Yann Le Cam



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