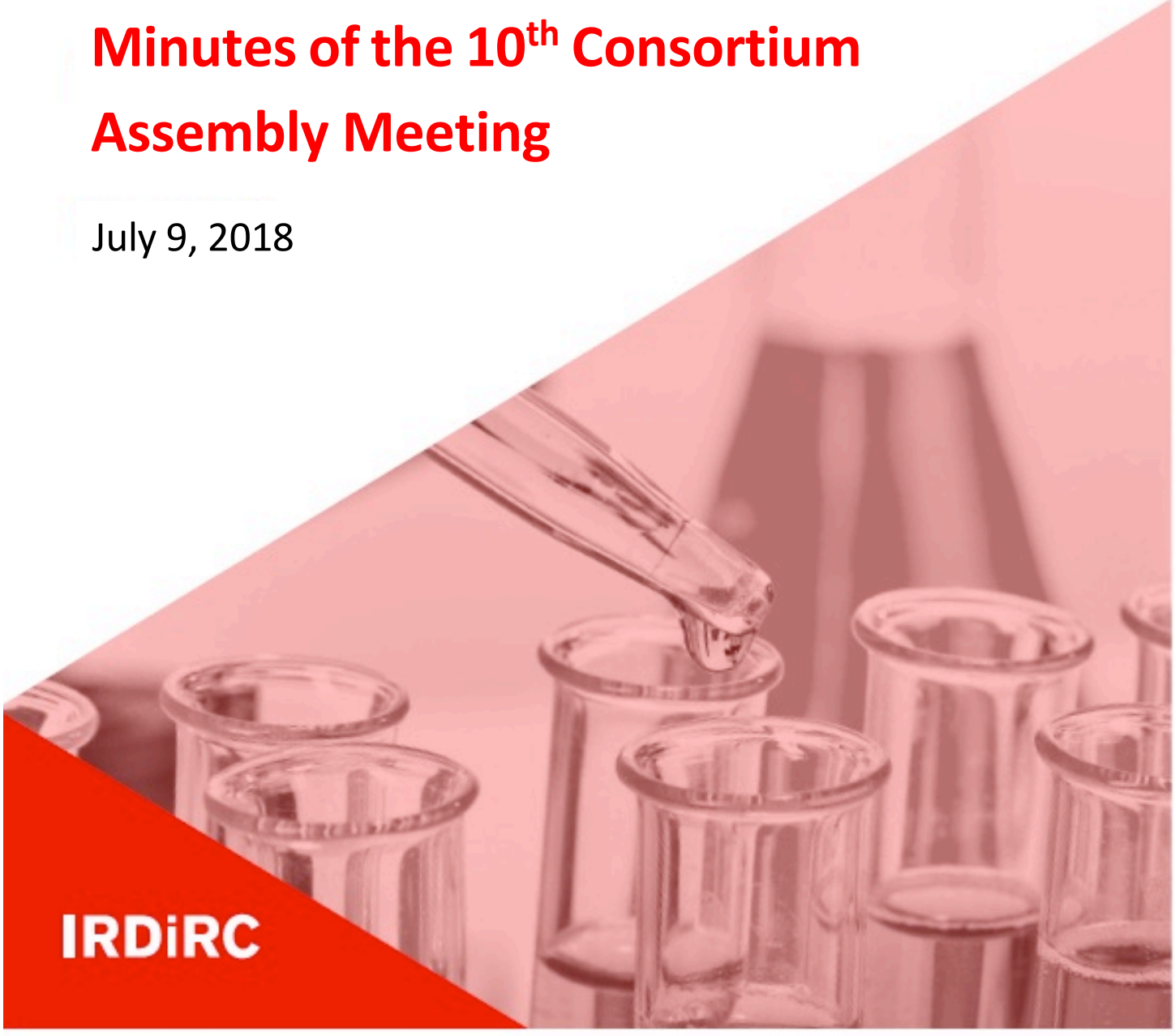
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 10th Consortium Assembly Meeting

July 9, 2018



IRDiRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on July 9, 2018, via web/teleconference. It was attended by 36 participants representing 28 member organizations, the Scientific Committees (SC) and the Scientific Secretariat (Sci Sec).

1. Activity proposals
   - Activity B – Identifying barriers to patient participation in RD R&D
     - Activity by Patient Advocates Constituent Committee (PACC), aimed to leverage IRDiRC's stakeholder and geographic representation to conduct a complementary environmental scan of barriers to and recommendations for patient participation in RD research
     - Activity will last through the end of 2019, but intermediate outcomes and results will be released throughout the activity
     - Expected outcomes are the identification of barriers from the perspectives of all stakeholders within IRDiRC, recommendations to better align efforts and needs, facilitate better patient engagement, determine strategic areas for funding, and inform future activities of IRDiRC and the entire RD community
     - Final proposal was presented and will be brought to subsequent electronic vote
   - Activity G – Clinical Research Networks for RD
     - Activity by Funders Constituent Committee (FCC), aimed to map and analyze the existing ecosystem of national/supranational clinical research networks, and to develop policy recommendations for collaboration of these networks
     - Activity will last through mid-2019, with a workshop in early 2019
     - Expected outcomes are a landscape paper on the different plans for international rare diseases clinical research networks, and recommendations on guiding principles for national/supra-national policies on clinical research networks
     - Final proposal was presented and will be brought to subsequent electronic vote

2. Governance proposal – updated membership criteria
   - Due to the influx of patient advocacy organization interested in becoming an IRDiRC member, patient advocacy membership criteria was revisited.
     - Specific points that were proposed for an update are the addition of: research focus, "direct experience" representation, non-represented regions, and Letter of Intent/Letter of Motivation requirements
     - Proposed language was refined based on previous CA, PACC, and OpComm discussions; it was presented and will be brought to subsequent electronic vote

REPORT

1. Activity Proposals

Two Activity proposals were presented to the IRDiRC CA members, Activities B and G. Hereafter, the Sci Sec will send out a survey, asking CA members to vote electronically.

1.1 Activity B - Identifying barriers to patient participation in RD R&D

- Committee: PACC
- Goal: Leverage IRDiRC's stakeholder and geographic representation to conduct a complementary environmental scan of barriers to and recommendations for patient participation in RD research
- Background
  - There is a need to determine how best to empower and enable patient participation in RD research
  - Initial discussion
    - Centered around the creation of a multi-stakeholder survey to be sent around to patients and other stakeholders
    - After further consideration within PACC breakout discussion, the consensus was multifaceted but that a simplification was needed
  - PACC now would like to take advantage of the players within IRDiRC who represent a broad swath of RD research stakeholders and are already committed to IRDiRC's mission
- Timeline
  1. Develop proposal within PACC – *done*
  2. Present proposal to CA for approval – *today*
  3. Initiate PACC-led TF – *Sept 2018*
     - Members will include internal representation (from across IRDiRC), and external experts in methodology, qualitative data analysis, data protection, etc.
  4. Start PACC-led TF work to define implementation details – *Q3-4 2018*
     - Define design, questions, and instrument
  5. Initiate PACC focus groups – *Q1 2019*
     - Questions around: what are the distinct barriers that your patients face, what are the solutions that have been successful, etc.
  6. Initiate Funders, Companies scan – *Q1 2019*
     - Questions around: what are the methodologies or strategies that your organization uses to conduct patient engagement in research, what are your solutions, etc.
  7. Initiate SCs scan to represent academic researchers across jurisdictions – *Q1 2019*
     - Questions around: how your sponsors deal with patient engagement, are those methods effective/appropriate, etc.

8. Intermediate review of results from three streams (PACC focus groups, FCC/CCC scan, SCs scan) – *Q2 2019*
    - Take advantage of May 2019 CA F2F
9. Analyze results and develop recommendations – *Q3 2019*
    - Publish/disseminate recommendations – *Q4 2019*

- Product and potential outcomes
    - Identify barriers from the perspectives of all stakeholders within IRDiRC
    - Determine whether existing strategies for improvement align with barriers identified by stakeholders
    - Create list of the suite of methodologies/strategies used in patient engagement (from Funders and Companies perspective)
        - Disseminate/publish to enable developing countries to view the list; determine whether they can potentially use or implement
    - Develop recommendations to:
        - Better align efforts and needs
        - Facilitate better patient engagement across geographic areas with shared resources
        - Determine strategic areas for new funding initiatives
        - Inform future activities of IRDiRC (and wider RD research community)
    - Publish/disseminate recommendations
- People involved in activity
    - PACC-led Task Force
    - PACC members
    - FCC, CCC, SC members
    - Sci Sec

Comments:

- Work is well needed, and can build further on existing knowledge
    - Several existing initiatives, such as Clinical Trials Transformation Initiative (CTTI), European Patient Academy (EUPATI), should be investigated as pre-work
    - Many of the people who will be included in the PACC TF have been involved in the creation of these existing initiatives, as well. Their help in the design and implementation details will be helpful and crucial.
- How is research defined in this context? The question is currently rather broad; how can it be narrowed around "research?"
    - Part of the proposed timeline is dedicated to this question -- to define "research", to investigate other engagement initiatives, and to define our questions that build on the previous advances and progress the field.
- Many different groups are involved in this proposal – how to ensure that it appropriately involves all stakeholders and avoid overlap between different groups?

- The PACC, through the PACC-led TF, has a dedicated time to define the implementation details (including these types of issues) in Q3-4 2018, prior to the start of the scans in Q1 2019.
- The TF will need to ensure to tailor all the questions to the various stakeholder groups (and to the variation within the stakeholder groups) participating in the scan/exercise either via focus groups or interview (or other instrument defined by the TF).

- What is the timing on the outcomes of this activity, and the overlap with other activities?
  - The PACC would like to finish as quickly as possible; the proposal includes the shortest timeline that could be imagined by the PACC. To ensure that the outcomes of this activity can be of influence and appropriately integrated into other IRDiRC activities, results and outcomes will be shared several times throughout this activity, prior to the finalization.

1.2 Activity G - Clinical Research Networks for RD

- Committee: ISC, FCC, TSC (ISC to lead). This proposal has been presented twice, in Tokyo and Vienna, and has been refined with comments from the CA and different CC/SC
- Objective:
  - To map and analyze the existing ecosystem of national/supranational clinical research networks
  - 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
- Background
  - This activity would build on different international initiatives
    - US - Rare Diseases Clinical Research Network (RDCRN)
    - EU - European Reference Networks (ERNs)
    - EU – Solve-RD project
    - International - Undiagnosed Diseases Network International
    - United Kingdom – 100 K Genome project
    - Japan – IRUD
    - Western Australia - Strategic Rare Diseases Framework
- Timeline
  1. Adapt proposal within ISC, FCC, TSC – *done*
  2. Present proposal to CA for approval – *today*
  3. Assemble a Task Force team with two Co-Chairs (preferably from different continents) – *Q3 2018*
  4. Develop a scope of the challenge and state-of-play document which includes an overview of the different clinical plans for clinical research networks for rare diseases – *Q3 2018*
  5. Identify one workshop that addresses an area(s) of priority – *Q4 2018*
  6. Develop recommendations and a scientific paper from the workshop – *Q1 2019*

- Product and potential outcomes
  - Landscape paper on the different plans for international rare disease CRNs, reviewing the experience gained and ongoing initiatives
  - Recommendations on guiding principles for national/supra-national policies on clinical research networks
  - Recommendations for alignment of protocols for data collection, infrastructures, centralized data repository, longitudinal studies, clinical trials, natural history studies
- People involved in activity
  - Interested members of the SCs and CCs
    - Interdisciplinary Scientific Committee (ISC)
    - Therapies Scientific Committee (TSC)
    - Funders Constituent Committee (FCC)
  - External experts
    - Representatives from worldwide CRNs for RD

Comments:

- Comes at a good time, after the launch of the ERNs, and around the time of the renewal of the US RDCRN.
- There are several other initiatives in the process of setting up, or developing further, so the activity could be of importance on an international scale.
- In the list of organizations that could be involved, there should be a clinical research network added on rare cancer that has developed intergroup policies for rare cancer (www.irci.info)
- Will these recommendations be addressed to funders?
  - At present, it is difficult to say based on the preliminary gaps that were identified
  - A scoping exercise could be performed at the beginning of this activity, clarifying where further funding is needed, on what topics, and in which geographical areas. The recommendations could be based on the results of such an exercise.
- Which standards are to be shared? Will these take into account the various international regulatory and legislative frameworks?
  - Harmonizing standards is not always possible, but the TF can collect best practices and guidelines, and facilitate the sharing of them.
  - Many clinical research networks already have been working on such guidelines for a while. It might be best to map best practices to understand all recent developments and diminish the variability of information.
  - The new General Data Protection Regulation (GDPR) legislation should be taken into account throughout the activity.

[post-meeting comment: In Europe, the European Commission's Joint Research Centre (JRC) in collaboration with DG SANTE is developing the European Platform on Rare Diseases Registration (EU RD Platform) which, for the first time, will make RD patient data searchable and findable at EU level. This is based on the European Rare Disease Registry Infrastructure (ERDRI), the EUPID psedonymisation tool and

EU-level standards for RD data collection and data sharing. Thus, the Platform will facilitate epidemiological, clinical, pharmacological, translational, etc. studies and research in the field of RD.]

→ Vote on Activity Proposals B and G via the electronic survey

2. Governance proposal – updated membership criteria

2.1 Patient advocacy groups

- IRDiRC has continuously evolved with regards to its membership, scope, goals, and activities
  - Given that and influx of applications, decided to revisit PACC membership criteria
  - Draft presented to OC in early March, then presented to the CA, and refined with comments from OC, CA and PACC
  - Current proposal broadly supported by all PACC members, after extensive discussion
- Current actual verbiage re Patient Advocacy Group membership in IRDiRC Governance:
  - *"To be considered as an IRDiRC Patient Advocacy Group member, the umbrella organization must be a patient organization representing broad patients' interests for all rare diseases in at least one country or larger area. In general, IRDiRC expects patient groups that are members of an umbrella organization already a member of IRDiRC to be represented appropriately by that organization within IRDiRC. Once approved, each umbrella organization can nominate one representative to the Consortium Assembly, who will also serve on the Patient Advocates Constituent Committee."*
- This includes:
  - Umbrella or pan-rare disease focus (i.e., not single disease or organ specific)
  - Country or greater representation
  - Groups that are members of an umbrella organization already a member of IRDiRC are expected to be appropriately represented by that organization within IRDiRC
- Suggested additions to Governance verbiage:
  - Add research focus
    - *"…the umbrella organization must be a patient organization (1) representing broad patients' interests for all rare diseases in at least one country or larger area and (2) contributing to research that shares and will advance the IRDiRC vision and goals (e.g., developing and providing tools to accelerate research, diagnostic and therapeutic development, evaluation of processes)."*
  - Encourage "lived experience" representation
    - *"IRDiRC recommends that the organization nominate a representative who has direct experience with an RD and worked in the interest of patients for at least a year."*
  - Encourage non-represented regions to join
    - Broaden to be applicable to all of IRDiRC, not just the PACC (add to governance, not in the PACC portion but the general membership portion)

        - *"IRDiRC actively seeks organizations from non- or under-represented geographical regions to join due to the critical importance for IRDiRC to be representative of the global RD community. It is essential that all individuals affected by rare conditions have a voice to enhance visibility and international collaboration, and mutual exchange of knowledge and experience."*
    - Add Letter of Intent/Letter of Motivation (LOI/LOM) requirements
        - Ask applicants to answer the following questions within the LOI/LOM:
            - *"Why does your organization want to join IRDiRC?*
            - *How does your organization contribute to research on rare diseases? Please provide examples of your research focus.*
            - *What will your organization specifically contribute to IRDiRC to help advance IRDiRC's vision, goals, and activities?"*
        - Make all LOI/LOM requirements consistent (i.e., once the above questions are finalized, propagate them to the Funders/Companies LOI/LOM).

- Comments and suggestions of CA
    - Research focus
        - How do we address research diversity and capability?
            - There has been a lot of discussion on this topic with the PACC members, they themselves having different involvements with research. Therefore, decided not to codify it in the governance and instead leave it up to the PACC and OpComm to review submitted application materials for appropriateness within IRDiRC.
        - What are experiences that count as research?
            - As discussed above, it was decided by the PACC to not define the research focus too much in the governance and instead leave it up to the PACC and OpComm to review submitted materials for appropriateness within IRDiRC.
            - A few examples of research focus are included in the updated governance language.
    - Encourage non-represented regions to join
        - Is there a degree of flexibility for membership?
            - Previously there was a discussion to create different criteria for developed and developing countries, but a decision was made that this would be more discriminatory. Therefore, a decision was made that membership criteria should be broad and inclusive enough, which the PACC thinks these criteria are.

→Vote on the governance updates via the electronic survey

3. Any other business

For Activity C (*Guidebook for drug developers describing available tools/initiatives specific for RD and how to best use them)*, a list of Building Blocks has been developed, and sent out to all CA members. Members are asked to review the list and send additional Building Blocks to the Sci Sec.

→Review list of Activity C Building Blocks, and send in additional Building Blocks, if needed, to the Sci Sec

Actions and deliverables

- CA
  - Vote on Activity Proposals B and G via the electronic survey
  - Vote on the governance update via the electronic survey
  - Review list of Activity C Building Blocks and send in additional Building Blocks, if needed

Document history

Version 1. Report drafted by Anneliene Jonker, July 16, 2018
Circulated to Chair of the CA, July 16, 2018
Version 2. Report edited by Christine Cutillo, July 18, 2018
Circulated to members of the CA, July 30, 2018

Annex - List of participants

Members	Representative
Western Australian Department of Health, Australia	Hugh Dawkins
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
Canadian Institutes of Health Research (CIHR), Canada	Christopher McMaster
European Commission, DG Research and Innovation, EU	Iiro Eerola
E-Rare Consortium, Europe and Agence National de Recherche, France	Daria Julkowska
EURORDIS-Rare Diseases Europe, Europe	Virginie Bros-Facer
French Foundation for Rare Diseases, France	Roseline Favresse
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Organization for Rare Diseases India (ORDI), India	Prasanna Kumar Shirol
Shire Pharmaceuticals, Ireland	Madhu Natarajan
Istituto Superiore de Sanità, Italy	Domenica Taruscio
Telethon Foundation, Italy	Lucia Monaco
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
Rare Diseases International (RDI), Singapore	Ritu Jain
Korea National Institute of Health, South Korea	Younjhin Ahn
National Institute of Health Carlos III, Spain	Manual Posada
Ultragenyx, Switzerland	Tom Pulles
Food and Drug Administration (FDA), USA	Ilan Irony, Gumei Liu
Genetic Alliance, USA	Sharon Terry
Global Genes, USA	Maureen McArthur Hart
National Center for Advancing Translational Sciences (NCATS), USA	Christine Cutillo
National Human Genome Research Institute (NHGRI), USA	Teri Manolio
National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), USA	Faye Chen
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

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Interdisciplinary	Petra Kaufmann, Domenica Taruscio
Therapies	Diego Ardigo

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Marlene Jagut, Anneliene Jonker, Anne-Laure Pham-Hung d'Alexandry d'Orengiani
NIH/NCATS	Lilian Lau

Apologies

Members	**Representative**
Rare Voices Australia, Australia	Nicole Millis
Botswana Organization for Rare Diseases (BORDIS), Botswana	Eda Selebatso
Genome Canada, Canada	Cindy Bell
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Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
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NKT Therapeutics, USA	Robert Mashal
PTC Therapeutics, USA	Ellen Welch
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Sanford Research, USA	David Pearce

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Diagnostics	Kym Boycott
Therapies	Virginie Hivert

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