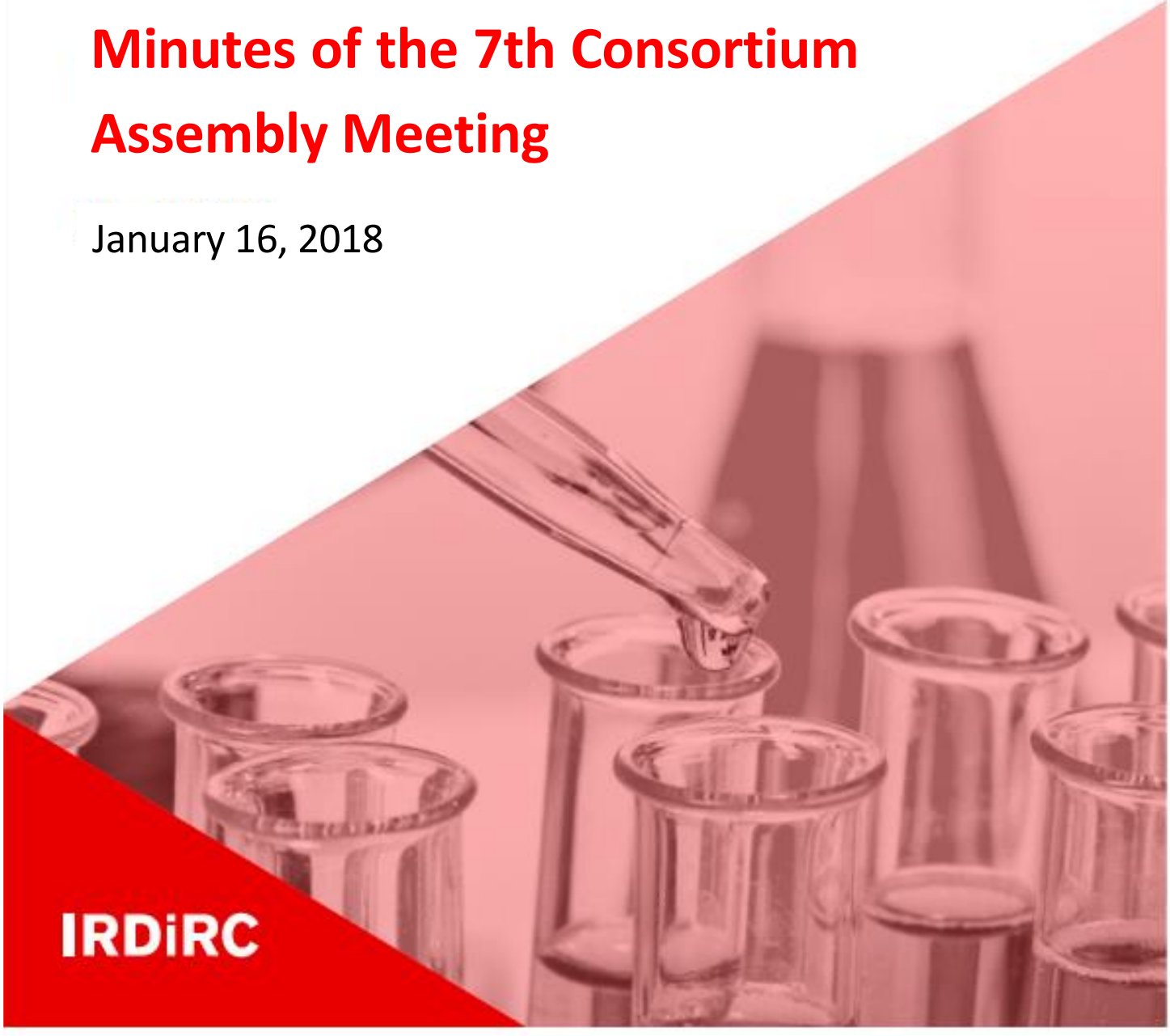
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 7th Consortium Assembly Meeting

January 16, 2018



IRDiRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on January 16, 2018, via web/teleconference. It was attended by 38 participants representing 26 member organizations, the Scientific Committees (Sci Comms) and the Scientific Secretariat (Sci Sec).

1. IRDiRC Roadmap 2018
   - With the publication of the new IRDiRC goals, embarked on process to establish a roadmap on how to move towards the new goals
   - The Draft Roadmap 2018 was presented to the CA
     - The rationale behind the creation of the Draft Roadmap 2018 was presented
     - A discussion of prioritized actions took place
     - Feedback received will be incorporated to the final Roadmap 2018
     - CA members will be contacted for vote on the final Roadmap 2018
2. Updates from the Chair
   - Membership updates
   - The IRDiRC CA/SC face-to-face meetings are taking place on May 14-16, 2018 in Vienna, Austria, right after ECRD 2018 (May 11-12, 2018)

REPORT

1. IRDiRC Roadmap 2018

1.1 Introduction IRDiRC Roadmap 2018

- Starting point: new IRDiRC Goals in 2017 and goals roll-out
  - Team effort for write-up of the three articles
    - Papers are not only important for IRDiRC, but also for rare diseases research
    - 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
- Next steps: Roadmap and action planning
  - Based on new goals, embarked on planning process for strategically identifying top priority actions for the Consortium for the coming years
    - Developed process/procedure
    - Initiated process for identifying actions of highest priority
    - Aim of determining the best activities for each Committee to work on in order for IRDiRC, as a whole, to progress toward the new goals

- Process of action planning
  - September-October 2017
    - Brainstorming at Committees level
  - November 2017
    - Refining actions with Committees
    - Review of actions by Sci Sec, together with Chair and Vice Chair of CA
  - December 2017:
    - Sci Sec worked with the Operating Committee (OpComm) to develop:
      - Prioritization and approval procedures
      - Capacity and scope of Sci Sec document
      - Criteria for action prioritization
    - Sci Sec worked closely with Committee Chairs to:
      - Ensure that submissions were represented accurately
      - Merge actions, where appropriate
      - Shortlist actions of highest priority, based on capacity and scope
  - January 2018
    - OpComm discussion of the Draft Roadmap 2018
    - Presenting Draft Roadmap 2018 to the CA for discussion and deliberation
  - Next steps, Sci Sec will
    - Incorporate feedback received on the CA call

- Send out final version of the Roadmap 2018 for vote by the CA
- Amend Sci Sec contract, where appropriate
- Start work on 2018 actions with relevant Committees

1.2 Prioritization and Approval Procedure

A prioritization and approval procedure was developed to facilitate the planning of future actions of the Scientific Committees (SC) and Constituent Committees (CC).

Main aspects of the procedure include:

1. Committee brainstorming
    - For current set of actions, this was organized prior to the Tokyo CA meeting
2. Actions refined within the Committee
    - Actions sent to the Sci Sec for compilation
3. Review of actions by Sci Sec, together with Chair and Vice Chair of CA
    - Analyze proposed actions submitted, identify overlaps, and merge overlapping actions
    - Determine capacity to take on actions given overall work plan
    - Propose shortlist of highest priority actions in accordance to resources and scope
4. Review by OpComm
    - Discuss highest priority actions, and prioritize actions for the Consortium as a whole
    - Review actions in context of overall work plan
5. Review by CA and vote ← *We are currently here*
    - Presentation to CA for discussion and feedback
    - Integration of feedback and finalize roadmap
    - Vote for approval by CA

1.3 Sci Sec Capacity and Scope

A capacity and scope document of the Sci Sec was presented with regard to Task Forces and Activities for the planning of ongoing and future actions by the SCs and CCs. The document provided an overview of the Sci Sec charge, scope and capacity, particularly with respect to the Draft Roadmap 2018.

- Sci Sec Charge and Scope
    - To better understand the scope of the Sci Sec contract, the contractual language is provided for Sci Sec activities in support of the CA, SCs, CCs, OpComm, and Task Forces
    - Sci Sec has a maximum capacity to take on 6 Task Forces, if they are not in the same heavy workload phase, in addition to the normal workload of the Sci Sec
    - As a ballpark figure, a Task Force requires about 10-15 weeks of Sci Sec time
- Task Forces phases
    - Not all Task Forces follow the same procedure; the various phases with different Sci Sec involvement were presented

- Current status of Task Forces, with regards to Sci Sec involvement (N.B., Phases 4-6 are heavy workload phases):
  - Phase 0: Approved, not yet started -- *CDS*
  - Phase 1: Request for nominations -- *MCC*
  - Phase 4/5: Workshop organization, TC preparation and reporting phases -- *STU*
  - Phase 6: Report and article writing -- *SPCT, ADA, DMR, PPRL*
  - Phase 7: Finalization -- *PCOM, MME*
- BioRxiv was suggested as a potential paper submission site, to enable preprinted articles prior to peer review journal publication be accessed in a timely manner

1.4 Criteria for Prioritization

- In order to arrive at the proposed Draft Roadmap, actions for 2018 were separated based on the below criteria:
  - (1) The activity is **within** the current scope and capacity of the Sci Sec (e.g., TFs)
    - Separated out into the below based on Sci Sec assessment and Comm Chair feedback:
      - Those that are currently **prioritized (a)**
      - Those that are tentatively **deprioritized (b)**
        - For those that are (b) deprioritized, these are saved in the hopper for future activities and years
  - (2) The activity **outside** the current scope and capacity of the current contract of the Sci Sec (e.g., subcontracts for database creation)
    - Separated out into the below based on Sci Sec assessment and Comm Chair feedback:
      - Those that are currently **prioritized (a)**
        - For these activities, the contract will need to be amended, in order to allow the project to be done
      - Those that are tentatively **deprioritized (b)**
        - For those that are (b) deprioritized, these are suggested as potential actions for Consortium Assembly members to pick up

1.5 Draft Roadmap 2018

The Excel document with all actions collated, containing three separate tabs, was presented.

- The "All requests for funding" tab
  - In the most right column, the Sci Sec has noted how each action was dealt with according to the prioritization criteria described above, with additional notes included
- The "2018 requests sorted by criteria" tab
  - Contains all requests are sorted by the prioritization criteria
  - Actions that fall into categories (1a), (2a) and (2b) were reviewed with the CA

- Actions that are deprioritized and within scope (1b), will be saved for future years
- Actions that are outside scope and deprioritized (2b) would be best picked up by CA members if possible; otherwise, these will also be saved for future years

- The "Draft Roadmap 2018" tab
  - Shows current activities and suggested prioritized actions for 2018
  - Tab is separated into
    - Current actions (those already approved and ongoing) above the grey line,
    - Potential actions for 2018 (those currently up for discussion) below the grey line
    - To the left of frozen frame, activity descriptions
    - To the right of frozen frame, timeline and capacity planning
  - The Draft Roadmap 2018 also shows ongoing major actions for the year, such as the IRDiRC CA and SC meetings, and contract reporting
  - A few actions proposed by multiple Committees were merged to form single actions
  - Activities are spread out over the year to enable a consistent workload as much as possible
  - For each activity, the estimated cost and Sci Sec effort is noted

Novel actions included in the Roadmap 2018 are:

Committee	Action number	Action
FCC	A	Establish process to coordinate and prioritize research funding efforts
PACC	B	Identify barriers to patient participation in RD R&D and develop recommendations to remove them
TSC	C	Defining new process for drug development and registration of innovation therapies for RD
CCC, TSC, FCC	D	Natural history and registry (NH/R) platform for use in real world evidence (RWE) data collection
TSC	E	Support the reframing of the current international research agenda for RD pushing for focusing research efforts and funding
PACC	F	Issue position statement including specific recommendations on: *Model for applying Goal 2 (therapy development) internationally *Model for inclusion of patients' perspectives in that therapy development
ISC, FCC	G	Clinical Research Network (pending re-scoping activities by Comm Chairs)
CCC	H	Background internal work on common knowledge base to drive RD research

1.6 Discussion on Draft Roadmap

- Overall discussion points:
  - Concern on the phrasing of certain actions was raised as IRDiRC is not a lobby group (e.g. rather than "creating a position statement for" it should "develop recommendations on")

        - Actions are in good spirit, but vocabulary needs to be rephrased
    - After approval of actions, a spreadsheet of the actions should be created, clearly portraying what IRDiRC is doing in a tangible way, divided by goal
        - Define metrics to define progress, in collaboration with CC/SC Chairs
    - Possibility to reinstate a "voluntary membership fund" to take on actions that cannot be taken on by Sci Sec?
        - Reviewing possibilities of including voluntary membership fund in EJP – not yet confirmed
        - Nevertheless, members with resources and capacity are encouraged to take on actions individually, and report back to CA

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

2.1 IRDiRC membership changes

- A change in the Patient Advocates Constituent Committee
    - Virginie Bros-Facer represents EURORDIS-Rare Diseases Europe
- A change in the Companies Constituent Committee
    - Madhu Natarajan represents Shire

2.2 Joint CA/SC face-to-face meeting in Austria, Vienna

- Next joint CA/SC F2F meeting will be held on May 14-16, 2018 in Austria, Vienna, contemporaneous to the European Conference on Rare Diseases (ECRD) on May 10-12, 2018
- Details:
    - May 14 – IRDiRC SC Meetings
    - May 14 (*evening*) – Joint informal IRDiRC CA/SC Dinner
    - May 15 (*morning*) – Joint IRDiRC CA/SC Meeting
    - May 15-16 – IRDiRC CA Meeting
- A survey will be forthcoming to gauge the number of participants

Actions and deliverables

- Provide comments to Draft IRDiRC Roadmap 2018 by January 17, 2018
- Complete the survey to vote on the final IRDiRC Roadmap 2018 by January 21, 2018
- Complete the survey on attendance to the IRDiRC CA/SC meetings in Vienna, Austria
- Send out survey to vote for final Roadmap 2018
- Send out survey for attendance IRDiRC CA/SC meetings in Vienna, Austria
- Organize next IRDiRC CA/SC meetings in Vienna, Austria

Annex - List of participants

Members	**Representative**
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin (Chair)
Western Australian Department of Health, Australia	Hugh Dawkins
Rare Voices Australia, Australia	Nicole Millis
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, Irene Norstedt
E-Rare Consortium, Europe / Agence National de la Recherche (ANR), France	Daria Julkowska
Rare Diseases Europe-EURORDIS, 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
Indian Organization for Rare Diseases, India/USA	Ramaiah Muthyala
Shire Pharmaceuticals, Ireland	Madhu Natarajan
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore de Sanita, 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
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
Cydan II, USA	James McArthur
Food and Drug Administration (FDA), USA	Katherine Needleman
Genetic Alliance, USA	Sharon Terry
National Eye Institute (NEI), USA	Santa Tumminia
National Human Genome Research Institute (NHGRI), USA	Lu Wang
National Institute of Child Health and Human Development (NICHD), USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman
Recursion Pharmaceuticals Inc, USA	Tim Considine

Scientific Committees	
Diagnostics	Kym Boycott
Interdisciplinary	Petra Kaufmann, Domenica Taruscio

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

Apologies

Members	**Representative**
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
Genome Canada, Canada	Cindy Bell
Canadian Organization for Rare Disorders, Canada	Durhane Wong-Rieger
BGI, China	Ning Li
WuXi AppTec Co. Ltd., China	James Wu
Chinese Organization for Rare Disorders, China	Kevin Huang, Rachel Yang
Academy of Finland, Finland	Heikki Vilen
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Lysogene, France	Karen Aiach
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
Loulou Foundation, UK	Daniel Lavery
Ionis Pharmaceuticals, USA	Brett Monia
Genzyme, USA	Carlo Incerti
National Cancer Institute (NCI), USA	Edward Trimble
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

Pfizer, USA	Katherine Beaverson
PTC Therapeutics, USA	Ellen Welch
Sanford Research, USA	David Pearce

Scientific Committees	
Diagnostics	Gareth Baynam
Therapies	Diego Ardigò, Virginie Hivert

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