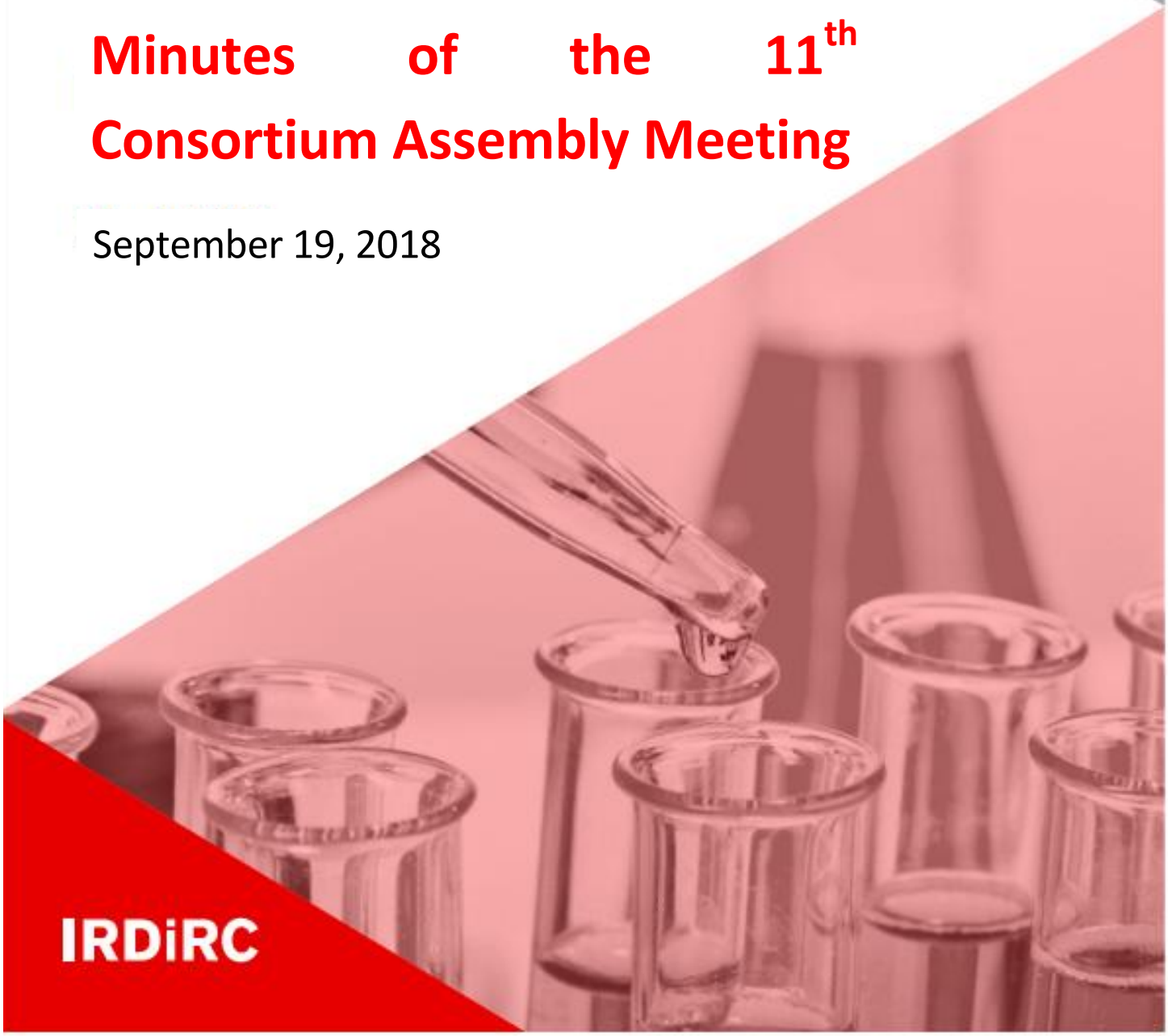
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 11th Consortium Assembly Meeting

September 19, 2018



IRDiRC

EXECUTIVE SUMMARY

The Consortium Assembly (CA) of the International Rare Diseases Research Consortium (IRDiRC) met on September 19, 2018, via web/teleconference. It was attended by 30 participants representing 21 member organizations, the Scientific Committees (SC) and the Scientific Secretariat (Sci Sec).

1. Consortium Assembly
   - Chairmanship and election
     - Current CA Chair will not seek re-election at the end of his term (December 31, 2018)
     - We will launch elections that will assure a new Chair to be in place by the end of the year, therefore a call for nominations will be officially launched in October 2018
     - Members are encouraged to start thinking about (self-)nominations
   - Face-to-face meeting in Brussels
     - Face-to-face meeting will take place in Brussels, Belgium December 6-7, 2018 at the European Commission offices
     - Meeting will last two days, and will include, among others, a roundtable discussion, parallel breakout sessions, and a CA wrap-up session
     - Members are asked to start thinking about topics they want to discuss in the face-to-face meeting, so they can be included in the agenda.
2. Major Activity updates
   - CCC
     - Mostly working on Activity H *the Penumbra project* – aimed at addressing the gaps of data to expand the list of rare diseases that companies are willing to work on.
   - FCC
     - Currently reviewing a call for tenders to fulfill the primary objectives of Activity A – *Global database and platform for RD funding analysis and collaboration*.
   - PACC
     - Selected external experts for Activity B – *Identify barriers to patient participation in RD R&D and recommendations to remove them* – Task Force; and investigating/welcoming other participants from the CCC, FCC and Scientific Committees.
   - DSC
     - Wrapping up *Solving the Unsolved (STU)* Task Force by writing a commentary on strategies to solve unsolved genetic diseases after exome sequencing and starting to draft a proposal for an *Underrepresented Population* Task Force.
   - ISC
     - Drafting the proposal for Activity D – *National history and registry platform for use in real world evidence data collection* – with input from the TSC, FCC and CCC; starting up Activity G – *Clinical Research Networks for Rare Diseases*; and wrapping up the *Model Consent Clauses for Rare Diseases Research (MCC)* Task Force
   - TSC
     - Organizing the workshop for Activity C – *Orphan Drug Development Guidebook* – and wrapping up previous Task Forces.
3. Scientific Committees

- ISC leadership update
  - Domenica Taruscio and Dixie Baker have been elected as Chair and Vice Chair, respectively.

4. Membership updates
   - National Institute for Dental and Craniofacial Research (NIDCR), NIH, joined as new IRDiRC member
   - Sanofi-Genzyme is now represented by Daniel Gruskin
   - NORD is now represented by Vanessa Boulanger

REPORT

1. Consortium Assembly

1.1 Chairmanship and Election

The current CA Chair, Christopher Austin, will not seek re-election, as he feels like much progress has been made during his tenure and it is time for someone else to take the reins.

- IRDiRC is on an extremely productive path – with many continuing and upcoming activities planned; and protocols and procedures in place to sustain the overall IRDiRC research infrastructure.
- The next Chair will be tasked with continuing on our positive and fruitful trajectory.
- Current CA Chair declares that it has been a privilege to lead the organization, and that he considered it the highlight of his career, so far.

The transition of the current to new Chair

- Official details:
  - Chair and a Vice Chair of the Consortium Assembly are elected for a period of 3 years that can be renewed once.
  - Official Chair period Christopher Austin: January 1, 2016 – December 31, 2018.
- Current CA Chair will ensure that a new Chair is situated and the Sci Sec hand-off settled prior to stepping away.
- We will start the procedure of a vote for the Fall with the aim to have a final Chair chosen by the Brussels meeting
- Current CA Chair will still run the Brussels meeting, but want to ensure the new Chair present to facilitate an easy hand-off of duties in January 2019

Procedure to elect a new Chair

- Announce election: today
  - Members are asked to start thinking about (self-)nominations
- Collection of nominations: early-mid October
  - Nomination includes bio and short statement of motivation from interested parties (via email)
- Fall teleconference: late October-early November
  - Each candidate will be asked to provide a very short (5 min) presentation stating why they are seeking Chairmanship and to answer any questions
- Electronic vote open: early-mid November
- Announce outcome: mid-late November
- Ceremonial hand-off: December 7, 2018 (Brussels CA face-to-face meeting)
- Official start of new Chairmanship: January 1, 2019

→ Members are asked to start reflecting on (self-) nominations for CA Chair

1.2 Face-to-face meeting in Brussels

The next CA face-to-face meeting will take place on December 6-7, 2018 in Brussels, Belgium, at the offices of the European Commission.

- Sci Sec will send detailed logistics information following the call
  - Will include the address and hotel suggestions for members
  - CA members are responsible for booking their own accommodations
- For travel arrangements, please aim to:
  - Arrive prior to meeting start on Thursday, December 6 at 8 AM (recommend flights arriving day prior or by 6 AM on the same day)
  - Stay until evening of Friday, December 7 (recommend flights departing after 8 PM)

The provisional meeting format is the following:

- Thursday, December 6
  - Morning: CA meeting, including the round-table discussion
  - Afternoon: CA meeting continued, potentially a "Rare Diseases Research in the Benelux" session
  - Evening: celebratory session for the European Joint Programme on Rare Diseases (5-7 PM), followed by an informal CA dinner (7:30 PM)
- Friday, December 7
  - Morning: Parallel CC meetings
  - Afternoon: Wrap-up CA meeting

Members are asked to start thinking about topics they want to discuss in the face-to-face meeting, so they can be included in the agenda. These ideas for topics should be send to the Sci Sec.

→ Sci Sec will send out detailed practical information regarding the Brussels CA meeting
→ Members should book and arrange their own accommodation for the Brussels CA meeting
→Send in topics for inclusion in the agenda of the Brussels CA meeting

2. Activity updates

2.1 Activity A tender outcome

- A call for tenders was launched in July to initiate Activity A -- *Global database and platform for RD funding analysis and collaboration* – through the creation of a comprehensive database and platform.
  - Aim is to systematically track and analyze global rare disease funding landscape, and coordinate that of all IRDiRC members
  - Expectation is that this effort will provide a tool that allows for in-depth analysis of funded projects and the rare diseases research funding landscape at an international level – to enable better understanding, address the gaps in research, and provide a basis for further funding coordination.
- Two applications received

- Proposals evaluated by CA Chair, FCC Chair, and current Sci Sec Coordinator
- Currently in the process of asking several follow-up questions to both applicants
- Decision will be made by next week

2.2 FCC activity update

- Activity A -- *Global database and platform for RD funding analysis and collaboration*
  - Call for tenders is currently under evaluation (see above for more details).
- Progress on the FCC survey for funding collaboration
  - Some answers still missing, but the final results are expected to be presented at the next FCC teleconference, and then be further discussed at the Brussels meeting.
- Ethical, Legal and Social Issues (ELSI) of Rare Disease Research
  - ELSI WG is drafting a paper based on the analysis of the survey.
  - E-Rare had a discussion on organizing a strategic workshop on ELSI issues.
    - Discussed current calls on this topic, and the possibility of organizing a joint call in three years.
- Draft proposal Activity D -- *Natural history and registry (NH/R) platform for use in the real world evidence (RWE) data collection*
  - Led by ISC, with involvement of TSC, FCC and CCC. ISC will provide an update.
- Activity G – *Clinical Research Networks for Rare Diseases*
  - Led by ISC, with involvement of TSC and FCC. ISC will provide an update.
- Research in developing countries
  - Currently no progress, but awaiting the DSC proposal for under-represented populations.

2.3 CCC activity update

- Regular calls initiated
  - Last call was the best attended, so far.
  - New members and old members with renewed interest attended.
- Currently working on recruiting additional companies' members to IRDiRC
  - Reached out to several companies to see if they are interested in becoming a member, which will hopefully lead to some applications soon.
- Activity D -- *Natural history and registry (NH/R) platform for use in the real world evidence (RWE) data collection*
  - Led by ISC, with involvement of TSC, FCC and CCC. ISC will provide an update.
- Activity H – *the Penumbra project*
  - The central aim of Activity H is to address the gaps of data to expand the list of rare diseases that companies are willing to work on. This is an area of research severely lacking and limiting the set of diseases companies are investing in.
    - Goal is to find data on the top 200-500 rare diseases, and find/generate the data to make them more attractive and accessible.
    - Particularly data on which decisions are made to continue research in companies.

        - The first step in this activity is to clearly define all data needed.
    - CCC members will investigate the possibility of mining and analyses of sequenced data to generate the information needed to improve the rare disease knowledgebase
        - To get a better understanding of disease prevalence, one of the biggest drivers of decision making
    - CCC members will contact coordinators from ERNs for a discussion and to learn about the ERNs data work flow
        - What data is available in the ERNs and what is a possible way to access that?
    - CCC Chair is preparing to draft a full activity proposal.

2.4 PACC activity update

- Activity B -- *Identify barriers to patient participation in RD R&D and recommendations to remove them*
    - Aim is 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.
    - First step is setting-up a Task Force to define implementation
        - The recent call for experts generated ~6 nominations with varied expertise (methodology, qualitative research methods, patient-centered outcome research); all nominees will be invited to take part in the activity.
        - The TF will aim to be relatively small, but PACC would like representation from (most) committees and therefore is still looking for input from the CCC, FCC and the Scientific Committees.
    - The initial activity of the Task Force, once in place, will be to define the process, implement the focus groups, perform analysis on data received, and generate recommendations.
- Activity F -- *Issue position statement including specific recommendations on a model for applying Goal 2 (therapy development) internationally and a model for inclusion of patients' perspectives in that therapy development*
    - The PACC members had previously decided to table this activity until the outcomes of Activity B are determined.
    - However, it seems important for PACC members to make a statement on the application of Goal 2 while the others IRDiRC Committees are working on its implementation strategies, so a decision has been made to create a strawman statement to get this out to all CA members
        - Statement is expected to be ready in the next couple of months.
    - Will be important to address the different challenges and opportunities in a worldwide setting, not only Europe and the US.

2.5 DSC activity update

- Wrapping-up of the *Solving the Unsolved (STU)* Task Force
    - Had a face-to-face meeting earlier this year, and still continues to do work together

  - First output expected later this year, with a special issue in the American Journal of Medical Genetics
  - Overview commentary on how to approach "unsolved" diseases after exome sequencing on strategies, technologies, resources that are needed
- Start of development of a proposal for an *under-represented populations* Task Force
  - In order to meet IRDiRC's Goal 1, special efforts are required for under-represented populations as well as populations in resource-poor areas, with a particular aim of decreasing health system access inequities in the field of rare diseases.
  - A Task Force proposal will be refined and circulated among DSC members for feedback shortly.
- Working on an overview of IRDiRC and its policies and guidelines
  - Invited review by EMBO report, to be submitted later this month.
- Working on carrier screening testing
  - No full Task Force proposal, but working on an overview review.

2.6 ISC activity update

- Activity D -- *Natural history and registry (NH/R) platform for use in the real world evidence (RWE) data collection*
  - Aim is to support the definition of standards for use of RWE data generation and collection – in particular for disease understanding and treatment monitoring.
  - Led by ISC, with involvement of FCC, TSC, CCC.
  - Will take into consideration activities planned in the EJP for Rare Diseases.
  - Proposal drafted and currently being reviewed by members SC/CC.
- Activity G – *Clinical Research Networks for Rare Diseases*
  - Approved at CA meeting in Vienna
  - Aim is to map the existing ecosystem of clinical research networks for rare diseases worldwide and to produce guiding principles.
  - Currently analyzing potential Task Force composition.
- *Model Consent Clauses for Rare Diseases (MCC)* Task Force
  - Workshop held in Paris on September 6-7, 2018
  - Gathered rare disease patient advocates, lawyers, researchers, and clinicians.
  - Discussed progression and history of consent forms in rare disease research.
  - Drafted a series of model consent clauses.
  - Paper will be written by next month.

2.7 TSC activity update

- Draft proposal Activity D -- *Natural history and registry (NH/R) platform for use in the real world evidence (RWE) data collection*
  - Led by ISC, with involvement of TSC, FCC and CCC. See above for update.
- Activity G – *Clinical Research Networks for Rare Diseases*
  - Led by ISC, with involvement of TSC and FCC. See above for update.

- Working on finishing manuscript of the *Data-Mining and Repurposing* Task Force
  - Activity C – *Orphan Drug Development Guidebook*
  - Currently the primary focus of the TSC.
  - Exercise to map all initiatives, tools, incentives and shortcuts that are available for drug development for rare diseases (with Europe, US, and Japan as geographical scope).
  - Tracking and highlighting the initiatives; creating fact sheets for all initiatives.
  - Workshop by the end of the year to put all initiatives into a coherent development plan.
- Activity E – *Support the reframing of the current international research agenda for RD pushing for focused research efforts and funding*
  - Activity will start after completion of Activity C

3. Scientific Committees

3.2 ISC leadership update

- ISC held elections for Chair and Vice Chair
    - Domenica Taruscio (Italy), the previous ISC Vice Chair is now elected as Chair, and follows in the footsteps of Petra Kaufmann
    - Term will run through the end of her mandate (end of February 2019).
  - Dixie Baker (US) elected as Vice Chair
    - Term will run through the end of her mandate (end of April 2021).

4. Any other business

4.1 Member updates since July

- New member:
  - National Institute for Dental and Craniofacial Research (NIDCR), NIH
    - Jason Wan, Director of Mineralized Tissue Physiology Program
- Change of representation:
  - Genzyme
    - From Carlo Incerti to Daniel Gruskin (Head of Global Rare Diseases Medical Affairs)
  - NORD
    - From Peter Saltonstall to Vanessa Boulanger (Director of Research Programs)

New members will be asked to present themselves at the CA face-to-face meeting in Brussels

Actions and deliverables

- CA

- Start reflecting on (self-)nominations for CA Chair
- Book and arrange own accommodation for the Brussels CA meeting
- Send in topics for inclusion in the agenda of the Brussels CA meeting
- Vote on DSC nominations via the electronic survey

- Sci Sec
  - Send out detailed practical information regarding the Brussels CA meeting

Annex - List of participants

Members	**Representative**
Western Australian Department of Health, Australia	Hugh Dawkins
Genome Canada, Canada	Cindy Bell
BGI, China	Jiong Zhang
European Commission, DG Research and Innovation, EU	Iiro Eerola
EURORDIS-Rare Diseases Europe, Europe	Gulcin Gumus
French Muscular Dystrophy Association (AFM-Téléthon), France	Alexandre Mejat
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Istituto Superiore de Sanità, Italy	Domenica Taruscio
Telethon Foundation, Italy	Lucia Monaco
Japan Agency for Medical Research and Development (AMED), Japan	En Kimura
Rare Diseases International (RDI), Singapore	Ritu Jain
Korea National Institute of Health, South Korea	Younjhin Ahn
Roche, Switzerland	Mathew Pletcher
Cydan II, USA	James McArthur
Food and Drug Administration (FDA), USA	Katherine Needleman
Genetic Alliance, USA	Sharon Terry
Global Genes, USA	Maureen McArthur Hart
National Center for Advancing Translational Sciences (NCATS), USA	Christopher Austin
National Institute of Dental and Craniofacial Research (NIDCR), USA	Jason Wan
PTC Therapeutics, USA	Ellen Welch
Sanford Research, USA	David Pearce

Scientific Committees	
Diagnostics	Gareth Baynam, Kym Boycott
Interdisciplinary	Domenica Taruscio
Therapies	Diego Ardigo, Virginie Hivert

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

Apologies

Members	**Representative**
Rare Voices Australia, Australia	Nicole Millis
European Organisation for Treatment & Research on Cancer, Belgium	Denis Lacombe
Botswana Organization for Rare Diseases (BORDIS), Botswana	Eda Selebatso
Canadian Institutes of Health Research (CIHR), Canada	Christopher McMaster
Canadian Organization for Rare Disorders (CORD), Canada	Durhane Wong-Rieger
Chinese Organization for Rare Disorders (CORD), China	Kevin Huang
Chinese Rare Diseases Research Consortium, China	Qing Kenneth Wang
WuXi AppTec Co. Ltd., China	James Wu
Academy of Finland, Finland	Heikki Vilen
E-Rare Consortium, Europe and Agence National de Recherche, France	Daria Julkowska
French Foundation for Rare Diseases, France	Roseline Favresse
Lysogene, France	Karen Aiach
Federal Ministry of Education and Research, Germany	Ralph Schuster
Organization for Rare Diseases India (ORDI), India	Prasanna Kumar Shirol
Indian Organization for Rare Diseases (I-ORD), India/USA	Ramaiah Muthyala
Shire Pharmaceuticals, Ireland	Madhu Natarajan
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (ASrid), Japan	Yukiko Nishimura
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Yoshihiro Yoneda
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Rare Diseases South Africa, South Africa	Kelly du Plessis
National Institute of Health Carlos III, Spain	Manual Posada
Ultragenyx, Switzerland	Tom Pulles
Loulou Foundation, UK	Daniel Lavery
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Genzyme, USA	Daniel Gruskin
Ionis Pharmaceuticals, USA	Brett Monia
National Cancer Institute (NCI), USA	Edward Trimble
National Eye Institute (NEI), USA	Santa Tumminia
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	Melissa Parisi

(NICHD), USA	
National Institute of Neurological Disorders and Stroke (NINDS), USA	Adam Hartman
National Organization for Rare Diseases (NORD), USA	Vanessa Boulanger
NKT Therapeutics, USA	Robert Mashal
Pfizer, USA	Katherine Beaverson
Recursion Pharmaceuticals Inc, USA	Chris Gibson

Scientific Committees	
Interdisciplinary	Dixie Baker

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