



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 16th Executive  
Committee Meeting**

4 August 2015



**IRDIRC**

## EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 4 August 2015 by teleconference. The sixteenth meeting of the Exec Comm brought together 27 participants, including two new members of the consortium – Pfizer and Japan Agency for Medical Research and Development (AMED), and two replacements in the Exec Comm.

The Chair of the Exec Comm reported his recent and upcoming activities, and the success of recent Scientific Committee (Sci Comm) meetings in Glasgow. The Chairs of Sci Comms provided updates from these meetings, outlined their upcoming activities and presented a refined mandate of the Sci Comm. The Exec Comm was also asked to consider organising another IRDiRC meeting, pending available financial support.

An update on all five Task Force (TF) activities to date was provided by the Scientific Secretariat (Sci Sec). The Exec Comm was also informed that a number of workshops will take place in the coming months:

- ▶ Matchmaker Exchange: 6-7 October 2015 (Baltimore, USA)
- ▶ Machine Readable Consent: 9-10 November 2015 (Paris, France)
- ▶ Patient-Centred Outcome Measures: 30 November 2015 (Paris, France)

GA4GH Framework will be invited to submit an “IRDiRC Recommended” application, and four other applications were assessed by the Exec Comm:

- ▶ International Charter of Principles for Sharing Bio-Specimen and Data: approved
- ▶ PhenomeCentral: required further clarification on points raised by reviewers
- ▶ Orphanet: approved
- ▶ Registro Español de Pacientes con Déficit de Alfa-1 Antitripsina (REDAAT): not supported

The Sci Sec gave an update on the preparation of State of Play 2015 report, which the Chair of the Exec Comm welcomed but wished to open another robust discussion on its content and utility before a 2016 report is prepared.

The intended discussions on Exec Comm meeting in March 2016 and endorsement of conference were postponed till the Exec Comm meets next month in Montreal.

## REPORT

### 1. Update from the Chair of the Exec Comm

The Chair of the Exec Comm welcomed two new members to IRDiRC: Pfizer and AMED, represented by Dr Katherine Beaverson and Dr Kazuo Kawamura respectively on behalf of the official Exec Comm representatives, Dr Kevin Lee and Dr Shigeki Kuzuhara. Both organisations will be introduced more formally through presentations at the next Exec Comm meeting in Montreal.

The Chair also welcomed new representatives for Genome Canada, Dr Cindy Bell, who replaces Dr Pierre Meulien following his appointment to the Innovative Medicine Initiatives (IMI), and Dr Ellen Welch who replaces Dr Diane Goetz following her retirement from PTC Therapeutics.

The Chair informed the Exec Comm of his attendance to the successful Sci Comm meetings in Glasgow, and has also presented IRDiRC as a model for international collaboration at the European Union-Latin America and the Caribbean (EU-LAC)-Health conference held in Brussels; both events took place in June 2015. The Chair will next attend two meetings in Australia in the coming weeks.

### 2. Updates following Sci Comm meetings in Glasgow

#### 2.1 Report from the Chair of the Diagnostics Sci Comm (DSC)

The Chair of the DSC summarised the meeting of the committee which took place over a full day, the first half for reviewing day-to-day work and the second half to draft a commentary.

The manuscript should be ready for internal review by the time the Exec Comm meets in Montreal. The Exec Comm agreed that the Chair and the Vice Chair will review the manuscript on their behalf and provide rapid feedback to DSC. The aim is to publish the commentary ahead of the ASHG meeting in Baltimore in October 2015. Both AJHG and EJHG have indicated their interest in publishing a joint commentary.

The Chair of the Exec Comm will also look into the possibility of reaching out to an Asia-based journal to gauge their interest in jointly publishing this commentary piece.

#### 2.2 Report from the Chair of the Interdisciplinary Sci Comm (ISC)

The meeting of the ISC also took place over a full day, where its members discussed various proposals that will be put forward to the Exec Comm in Montreal for approval to constitute Task Forces.

The Chair of the ISC also informed the Exec Comm of the resignation of Alastair Kent. The Exec Comm was asked to identify potential replacement to provide patient voice to the committee; otherwise the ISC would be happy to make some suggestions for selection and approval by the Exec Comm. [Post

*meeting note: according to the Governance document, page 7, “Individuals representing funders in the Executive Committee cannot be nominated or selected to also participate in a Scientific Committee”.]*

The Chair of the ISC also emphasised the importance of the joint Sci Comms meeting for overarching scientific views and achievements, and requested that such joint meeting be a part of future Sci Comm meetings.

### **2.3 Future IRDiRC meeting/conference**

Another point of discussion during the Sci Comm meetings was the possibility of organising another IRDiRC meeting/conference in late 2016, in conjunction with a reference to the milestone of reaching 200 therapies, and taking advantage of the momentum in place to set new goals for the second phase of IRDiRC. Members are conscious that this achievement should be celebrated with appropriate credits and attribution towards all stakeholders/organisations involved in culminating this success.

However, hosting a large-scale meeting will require financial support beyond what is available through Support-IRDiRC and membership fees. Should there be any/several funders on the Exec Comm open to take on this responsibility, an organising committee needs to be set up soon and the venue decided, as the timeline is relatively short.

### **2.4 Refinement of the mandate of the Sci Comm**

The mandate of the Sci Comm was discussed and refined in Glasgow, and this update had been endorsed by the Chairs of Sci Comms. The changes include clear Sci Comm roles in Task Forces, preparation of the annual State of Play report, and review of “IRDiRC Recommended”. On the latter, the Sci Comm members will abstain from reviews of resources which may pose a conflict of interest. The Sci Sec will also assign reviewers appropriately to respect non-conflict of interest.

The updated mandate was approved by the Exec Comm and the Governance document will be updated accordingly.

## **3. Updates on Task Forces (TFs)**

### **3.1 Matchmaker Exchange (MME)**

The MME TF Steering Comm has been operating actively, meeting by teleconference approximately once a month to discuss the content of the workshops, plan the agendas, finalise its invitation lists and organise meeting logistics.

The workshops will be held on 6-7 October 2015, at the ASHG in Baltimore. The full-day meeting on 6 October is for invited participants only, while the meeting on the evening of 7 October is a community-

engagement event, open to all and particularly targeted towards researchers interested in knowing more about MME and using its API.

No background paper is being prepared by the Sci Sec, as a special issue of Human Mutation on MME will serve as its background document. The covers for the special issue are currently in the works, and within, there will be a quarter-page advertisement bearing IRDiRC's logo and a brief description of IRDiRC. Copies of the MME issue will be printed and distributed in upcoming events.

A print run of 1,000 copies will be made for distributions at:

- ▶ MME community-engagement workshop (150-200)
- ▶ At IRDiRC and GA4GH booths at the ASHG, October 2015 (a couple of hundreds each)
- ▶ RE(ACT) meeting in Barcelona, March 2016
- ▶ ACMG meeting in Tampa, Florida, March 2016
- ▶ ICHG meeting in Kyoto, April 2016
- ▶ ECRD meeting in Edinburgh, May 2016

The printing and shipping cost will be supported by SUPPORT-IRDiRC; additional support is available through IRDiRC membership fee and GA4GH.

### **3.2 Patient-Centred Outcome Measures (PCOM)**

The PCOM TF Steering Comm has been constituted and invitation of general TF members had also been carried out. The Sci Sec is currently conducting one-on-one discussion with Steering Comm members to improve the background document, of which the second version has now been circulated to the Exec Comm. Refinement of this document is ongoing, and with each discussion, new ideas have been brought in as there have been high level of interest from the members of the TF. Members of the Exec Comm were asked to please read through the document and provide additional feedback, if any.

A workshop of PCOM TF will take place in Paris on 30 November 2015, and as soon as meeting logistics are ironed out, Steering Comm members and selected general members will be invited to attend. The Coordinator of SUPPORT-IRDiRC is confident that this TF will achieve its goals and advance the development of patient-centred outcome measures for rare diseases.

### **3.3 Small Population Clinical Trials (SPCT)**

The SPCT TF Steering Comm has been constituted and a preliminary background document had been produced by the Sci Sec. This background document will soon be circulated to the Steering Comm members for their first comments and for one-on-one discussion.

The dialogue with EMA on organising this workshop is ongoing, with the EMA agreeing to host the event in London. Meeting room availability is currently under investigation, and a poll will be conducted to identify the best date(s) for the workshop.

### **3.4 Data Mining and Repurposing (DMR)**

The DMR TF Steering Comm has been constituted. However, the Sci Sec has not yet been able to commence the preparation of its background document, due to staff change that leads to temporary shortage in the team. This will be remedied as soon as possible.

### **3.5 Machine Readable Consent (MRC)**

Led by GA4GH, this TF is due to hold their workshop in Paris on 9-10 November 2015. A number of ISC members are involved in this TF. The Sci Sec provides meeting organisation and financial support of this workshop.

## **4. “IRDiRC Recommended” application reviews**

A page has been set up on the IRDiRC private website to provide real-time information and updates of applications to the Exec Comm.

### **4.1 Procedural notes**

- ▶ Reviewing Sci Comm members should ask for clarification from applicants on any ambiguous point
- ▶ Future reviews sent to the Exec Comm for decision-making will also include a brief description at the start of the summary document
- ▶ Further discussion will be held in Montreal in relation to single disease registry applications

### **4.2 International Charter of Principles for Sharing Bio-Specimens and Data**

This application relates to a position paper/recommendation – the effort was led by Mats Hansson and included international groups – that provides a framework which takes into account ethical and legal considerations in sharing samples from biobanks and relevant data. The Charter can also be adapted accordingly by institutions adopting it. The Exec Comm voted in majority voice to approve this application as “IRDiRC Recommended”.

### **4.3 PhenomeCentral**

Reviewers’ comments were largely positive; however, reviewer #3 raised a few points for discussion. The sustainability of PhenomeCentral – one of the most innovative tools available – is assured with current resources available and funding for the next three years has also been secured. As it was unclear, according to reviewer #3, if PhenomeCentral adheres to ethical and privacy policies and requirements, the applicant will be asked to address this concern. This communication will be expedited to ensure PhenomeCentral receives an outcome following the Exec Comm meeting in Montreal.

#### **4.4 Orphanet**

Following assessment of reviews and discussion, the Exec Comm acknowledged the importance of Orphanet as knowledge source of rare diseases and that this should be the gold standard to which other resources should aspire. The Exec Comm voted in majority voice to approve this application as “IRDiRC Recommended”.

#### **4.5 Registro Español de Pacientes con Déficit de Alfa-1 Antitripsina (REDAAT)**

While the importance of REDAAT cannot be understated, it is, however, a national registry that doesn't meet the criteria of multi-national connectivity and audience, and lacks site and data accessibility to international community at large. The Exec Comm did not support this application as “IRDiRC Recommended”, and the reviews will be provided to the applicants to shed light on this decision.

### **5. State of Play 2015 update**

The Sci Sec provided an update on projected timeline in the writing and review of the State of Play 2015 report. A first draft is currently in preparation and will require a few more extra days than initially planned to finish, before being sent to the Exec and Sci Comms for review and edition.

The Chair of the Exec Comm wished to have a robust discussion of the content and utility of the State of Play report for upcoming years in another meeting following the publication of the 2015 report, to take into account the work needed, and done, on preparation of background documents and workshops for various TFs.

### **6. GA4GH Framework**

A request for endorsement of the GA4GH Framework for Responsible Sharing of Genomics and Health-Related Data was submitted via email. The Exec Comm asked that this requested be submitted via “IRDiRC Recommended” procedure for decision-making in Montreal.

### **7. Exec Comm face-to-face meeting: Spring 2016**

An initial plan for the Exec Comm to meet face-to-face in April 2016 near London had been cancelled. Two new propositions were received, and will be further discussed in Montreal.

#### **7.1 Barcelona, in conjunction with RE(ACT), March 2016**

E-Rare proposed to host the Exec Comm meeting in Barcelona, Spain, back-to-back to its membership meeting and the RE(ACT) Congress. Its proposed meeting dates are Sunday 6 March – Monday 7 March.

## 7.2 Lyon, in conjunction with Myology, March 2016

AFM-Téléthon offered to host the Exec Comm meeting in Lyon, France, back-to-back to Myology 2016 Congress. Access to Myology for Exec Comm will also be made available to members wishing to attend. A full day meeting on Monday 14 March was raised as possibility, which the host will confirm if meeting room is available.

## 8. Conference endorsement: ECRD 2016

IRDiRC has been invited to be an associate partner of the next European Conference on Rare Diseases and Orphan Products (ECRD), which will be held in Edinburgh, Scotland, on 26-28 May 2016. Due to time constraint, a full discussion could not take place during this call and will be properly discussed in Montreal.

## 9. Staff change at the Sci Sec

Antonia Mills has left the Sci Sec on 31 July 2015, and will be replaced by Anneliene Jonker who will commence on 1 September 2015. [*This information was shown on screen but not formally announced.*]

## 10. Agenda of Exec Comm meeting in Montreal

The following items will be included in the agenda of Exec Comm in Montreal:

- ▶ Presentations by Pfizer and AMED by representing members
- ▶ Updates on Task Forces
- ▶ Proposals of new Task Forces by Chairs of Sci Comms
- ▶ “IRDiRC Recommended” reviews and discussion re single disease registry
- ▶ Upcoming Exec Comm face-to-face meeting(s)
- ▶ Conference endorsement: ECRD 2016

[*Post-meeting note: members of the Exec Comm who would like to propose item(s) to the agenda please do so by emailing the Chair of the Exec Comm, copying the Sci Sec.*]

## 11. Next steps and actions

- ▶ Editorial review of DSC commentary
- ▶ Enquiry re Asia-based journal of interest in publishing DSC joint commentary
- ▶ Consideration of financing an IRDiRC meeting by interested funder(s)
- ▶ Update IRDiRC Governance document to reflect Sci Comm mandate
- ▶ Communicate “IRDiRC Recommended” decisions to applicants
- ▶ Invitation to apply “IRDiRC Recommended” for GA4GH Framework
- ▶ Review of State of Play 2015 Report when circulated
- ▶ Identification of available date(s) to host Exec Comm in March 2016

**Annex - List of participants**

<b><u>Members</u></b>	<b><u>Representative</u></b>
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research, Canada	Paul Lasko
Genome Canada	Cindy Bell
WuXi AppTec Co. Ltd., China	Mao Mao
E-RARE-2 Consortium, EU	Natalia Martin
Agence National de la Recherche, ANR, France	Juliane Halftermeyer
AFM- French Association against Myopathies, France	Marie-Christine Ouillade
Fondation Maladies Rares, France	Roseline Favresse
Federal Ministry of Education and Research, Germany	Ralph Schuster
Shire Pharmaceuticals, Ireland	Albert Seymour
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Telethon Foundation, Italy	Lucia Monaco
Agency for Medical Research and Development (AMED), Japan	Kazuo Kawamura
The Netherlands Organisation for Health Research and Development	Sonja van Weely
National Institute of Health Carlos III, Spain	Pedro Cortegoso Fernández
Food and Drug Administration, USA	Katherine Needleman
National Eye Institute, NEI/NIH, USA	Santa Tumminia
National Human Genome Research Institute (NHGRI), NIH, USA	Jeffery Schloss
Pfizer, USA	Katherine Beaverson
PTC Therapeutics, USA	Ellen Welch

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

<b><u>Scientific Committees</u></b>	
Diagnostics	Kym Boycott
Interdisciplinary	Hanns Lochmüller

<b><u>IRDIRC Scientific Secretariat</u></b>	
SUPPORT-IRDIRC Project	Sékolène Aymé, Lilian Lau

## Apologies

<b>Members</b>	<b>Representative</b>
BGI, China	Ning Li
Chinese Rare Diseases Research Consortium, China	Qing Wang
European Organisation for Treatment & Research on Cancer, EORTC	Denis Lacombe
European Commission, DG Research and Innovation, EU	Anders Colver, Iiro Eerola
European Commission, DG SANTE, EU	Stefan Shreck, Jarek Waligora
Academy of Finland, Finland	Heikki Vilen
Lysogene, France	Karen Aiach
Children's New Hospitals Management Group, Georgia	Oleg Kvlividze
Istituto Superiore de Sanita, Italy	Gualtiero Ricciardi
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Prosensa, The Netherlands	Scott Clarke
Korea National Institute of Health, South Korea	Hyun-Young Park
National Institute for Health Research, UK	Willem Ouwehand
Genzyme, USA	Carlo Incerti
Isis Pharmaceuticals, USA	Brett Monia
National Cancer Institute, NCI/NIH, USA	Edward Trimble
National Center for Advancing Translational Sciences, NCATS/NIH, USA	Christopher Austin
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
NKT Therapeutics, USA	Robert Mashal
Office of Rare Diseases Research, ORDR/NIH, USA	Pamela McInnes
Sanford Research, USA	David Pearce

<b>Scientific Committee</b>	
Therapies	Yann Le Cam



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