



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 18th Executive
Committee Meeting**

11 January 2016



IRDIRC

EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 11 January 2016 by teleconference. The eighteenth meeting of the Exec Comm was attended by 37 participants, representing 27 member organisations, the three Scientific Committees (Sci Comms) and the Scientific Secretariat (Sci Sec).

Dr Christopher Austin was elected Chair of IRDiRC Exec Comm in December 2015. The Exec Comm thanked the former Chair, Dr Paul Lasko, for his guidance and leadership in the past three years.

Dr Ségolène Aymé, Project Coordinator of SUPPORT-IRDiRC tendered her resignation in early January 2016. The SUPPORT-IRDiRC is undergoing a technical review and the Exec Comm will be updated of its progress in due course. Changes notwithstanding, the current Sci Sec team will continue to support the work of IRDiRC as usual. It will also redefine its role to support the vision of the new Chair and IRDiRC committees. The Sci Sec will present its work plan and communication strategy at the Exec Comm meeting in Lyon.

A revised IRDiRC organogram, which better represents transitions within IRDiRC including establishment of Task Forces and Operating Committee (Op Comm), was approved. The Governance document was also amended to take into account agreed changes from previous meetings. A motion of amendment was proposed to withdraw the voting right of Chairs of Sci Comms at the Exec Comm but the motion did not carry. Additional refinement to the Governance document is required in relation to description of Op Comm, term duration of Vice Chair, and Task Forces nominations and appointments, all of which the Op Comm will work on before presenting an updated draft for discussion at the next Exec Comm meeting.

Scientific leadership of Task Forces which previously lay with the Project Coordinator of the Sci Sec needs to be transferred; the Chairs of Sci Comms will identify key members from the Sci Comms and Task Force Steering Committees to take over the lead and ensure delivery of work as planned. The Exec Comm was also briefed on activities of five Task Forces to date, and approval was given to establish a new Task Force on Participant Unique Identifiers, proposed by the Interdisciplinary Sci Comm (ISC).

Two nominations to the Diagnostics Sci Comm (DSC) were approved for a 3-year mandate: Gareth Baynam and Anthony Brookes. Additionally, two members of the DSC were renewed: Xavier Estivill and Gert Matthjis. Two members of the ISC were also renewed: Angel Carracedo and Domenica Taruscio. The ISC is seeking to nominate new member with patient voice perspective and will write a half-page, identifying other expertise requirement to complement the existing committee in place. The membership and requirements of the Therapies Sci Comm (TSC) are still under review by its Chair.

Members of Exec Comm were solicited for topics they would like to discuss in Lyon. Currently topics include confirming a host for the Exec Comm meeting in Fall/Autumn 2016, and consideration of the venue of Exec Comm meeting in Spring 2017 to which the potential 3rd IRDiRC Conference could be coupled to. Members were also asked to consider the future and sustainability of the Sci Sec beyond 2018. A request was also made to identify the level of financial investment to IRDiRC to date.

REPORT

1. Announcements

1.1 Chairmanship of IRDiRC

Following an election which took place in late 2015, Dr Christopher Austin from the National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH), USA, was elected the new Chair of the IRDiRC Executive Committee (Exec Comm).

On behalf of the Exec Comm, the Vice Chair voiced the committee's gratitude to the former Chair for his guidance and leadership in the past three years, a sentiment echoed by the Chair-Elect, who is currently awaiting NIH ethical clearance in order to commence his work fully. Consequently, the Vice Chair presided over the teleconference on his behalf.

The Chair-Elect thanked the members for their confidence in electing him and informed the members that he has spoken to several members of leadership to get acclimated and brought up to date with IRDiRC activities. His plan for the initial few months is to listen to what members are looking for from IRDiRC. He expressed his enthusiasm to make headway on rare disease therapeutics and diagnostics, which have been a major focus of his career for almost 30 years.

1.2 Changes at the Scientific Secretariat (Sci Sec)

Dr Ségolène Aymé announced her resignation as Project Coordinator of SUPPORT-IRDiRC in early 2016. Members of the Exec Comm thanked her for her contributions to IRDiRC and SUPPORT-IRDiRC, and acknowledged her outstanding contribution and commitment in raising the profile of rare diseases research over the years.

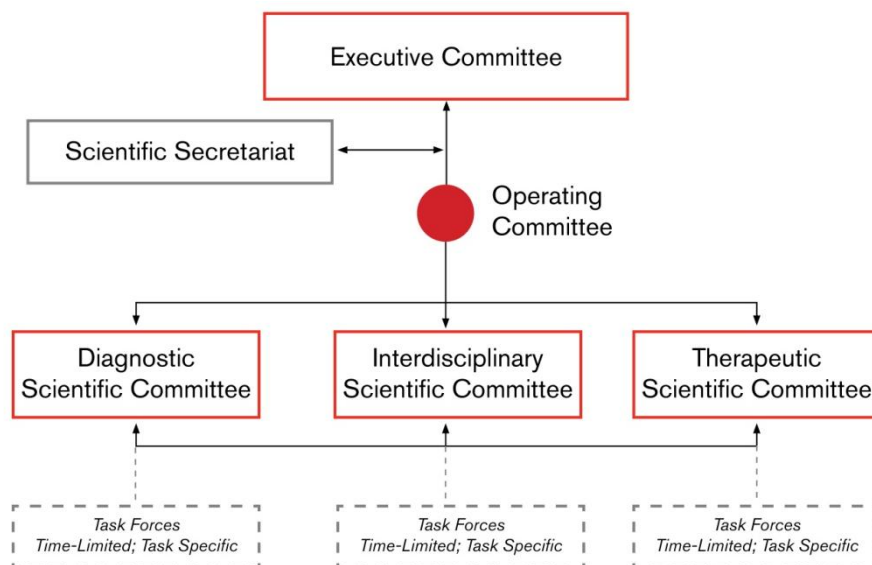
A standard technical review of the SUPPORT-IRDiRC instigated by the European Commission (EC) is currently ongoing and the Exec Comm will be updated of its outcome when it has been concluded. Inserm, the organisation to which the SUPPORT-IRDiRC is contracted, is in continuous dialogue with the EC; the Director of Service Unit 14 (US14) is an interlocutor as the US14 hosts the Sci Sec in Paris. The resignation of the Project Coordinator also triggers the need for an amendment of SUPPORT-IRDiRC contract, which will be carried out in consultation with the EC.

These changes notwithstanding, members were assured that work will be carried out as usual by the Sci Sec team currently in place and the transition will progress with as little disruption as possible. The Sci Sec will support the vision of the Chair and of all IRDiRC committees, and redefine its role accordingly. The Sci Sec will present a work plan, including its communication strategy – to be developed together with the Operating Committee (Op Comm) – to the Exec Comm at its next meeting in Lyon.

2. IRDiRC Organogram and Governance

2.1 IRDiRC Organogram

The Vice Chair presented, for approval of the Exec Comm, an IRDiRC organogram redrafted by the Op Comm to better represent the transitions which the IRDiRC underwent in the past 6-12 months, in particular the development of Task Forces and recently established Op Comm.



It was noted that the Sci Sec is not an entity in the organogram. Its dynamic interaction and complex reporting relationship with all IRDiRC committees is hard to capture, therefore its link is represented by double-sided arrow which feeds into the chain of communication of the main structure. More specific details (e.g., Sci Sec reporting to the Exec Comm) should be captured in the Governance document.

The Op Comm was instituted following the Exec Comm meeting in Montreal. It is composed of the IRDiRC Exec Comm Chair and Vice Chair, Chairs of Scientific Committees (Sci Comms), and the Sci Sec. The Op Comm is not an executive decision-making body; it was set up to ensure IRDiRC was able to manage and respond to issues in an agile manner and to assist operationally to deal with activities which require time-sensitive consideration, e.g., responding to the needs of Task Forces, and to streamline operational matters, e.g., polishing documents prior to bringing them to the Exec Comm for discussion. The Op Comm fills an important gap and has improved communications between the Sci Comms while keeping the Chair and Vice Chair of Exec Comm updated with ongoing activities. The Governance document will be updated with a description of the Op Comm to maintain coherence in the document.

2.2 IRDiRC Governance document

A number of changes to the Governance document were also discussed. Changes which reflect previous agreed discussions were highlighted and put forward as proposed changes. These were accepted, with additional points regarding Task Forces, e.g., nominating step described but not appointment step, to be

re-reviewed by the Op Comm. An updated draft will be presented to the Exec Comm at the next meeting in Lyon for discussion and approval.

AFM-Téléthon emphasised the importance of the presence of Sci Sec at the Exec Comm, and proposed an amendment to withdraw the voting rights of Chairs of Sci Comms, who are advisory members of the Exec Comm. Most members maintained that the current Governance document has a clearly defined provision in relation to potential conflict of interest,¹ and therefore the Chairs of Sci Comms should not be excluded from voting on decisions given the key role they play for the organisation. The motion for this amendment therefore did not carry.

A further change proposed to the Governance document is the inclusion of text to clarify the term duration of the Vice Chair, which is three years.

3. Task Forces

3.1 Reassignment of scientific leadership of Task Forces

Task Forces are complicated in terms of structure, along with nomination and appointment processes. Among five Task Forces established to date, two have been set up in conjunction with the Global Alliance for Genomics and Health (GA4GH) thus leadership of these Task Forces operates differently from the others.

Three other IRDiRC Task Forces, originated from the Therapies Sci Comm (TSC), have been delegated to the Sci Sec for management and operation. The former Project Coordinator of Sci Sec, who has rich scientific knowledge and expertise, had been steering the discussions; this expertise is now no longer available to IRDiRC. It is therefore imperative to identify key members from the Sci Comms and Task Force Steering Committees who are willing to step up and lead the work.

The Chair of the TSC will meet with the Sci Sec during the week following the Exec Comm teleconference to review and revise the work process for these Task Forces. He is confident that with the right leads for the Patient-Centred Outcome Measures (PCOM) and Small Population Clinical Trials (SPCT) Task Forces, the Sci Sec would continue to produce quality deliverables. For the Data Mining and Repurposing (DMR) Task Force, the first teleconference of the Steering Comm is due to take place on 20 January 2016. A decision whether to put the DMR Task Force on hold or to continue work will be made after an assessment at the next Op Comm on 14 January 2016. [*Post-meeting note: this teleconference will go ahead as planned.*]

IRDiRC could adopt a structure usually employed in other organisations, i.e., Chairs of Sci Comms decide on Steering Comms that are constellated around expertise, and work closely with key leads identified. The Op Comm will refine the work steps of the Task Forces accordingly.

¹ The Institute of Medicine has produced a good guideline “Conflict of Interest in Medical Research, Education and Practice”, available from [the National Academies Press](#).

3.2 Matchmaker Exchange (MME)

The Chair of the Diagnostics Sci Comm (DSC), who is also a co-lead of the MME, updated the Exec Comm of the recent MME workshop and community engagement event in Baltimore in conjunction with the ASHG 2015 conference. The workshop was attended by key MME members where discussions were centred on the service of the MME and its envisioned next steps, i.e., single-sided hypothesis matching and hypothesis-free matching; a document to outline these will be prepared in the coming months. The community engagement event was attended by individuals wishing to know more about the MME, and a special Human Mutation issue on MME was distributed.

Printed copies of the Human Mutation MME issues are still available for any members interested in providing them at future meeting. Some can be retrieved from the Johns Hopkins Institute of Genetic Medicine in the USA, and others from the Sci Sec office in Paris. [*Post-meeting note: Sci Sec will be providing the copies from the IRDiRC booth at the RE(ACT) Congress in Barcelona in March 2016.*]

3.3 Automatable Discovery and Access (ADA); formerly Machine Readable Consent (MRC)

ADA is a GA4GH-led Task Team and its members had a successful workshop in Paris on 9-10 November 2015. The group is now developing an Automatable Discovery and Access Matrix (ADAM), a tool which they hope to pilot in a few weeks. Use categories are currently being defined, and ADAM will enable query of data/cohorts based on set of criteria, e.g., for disease X at specific age group that is available for commercial use, and return a result without a person having to contact the PI for consent condition.

The work of ADA Task Team is closely followed by Interdisciplinary Sci Comm (ISC) members, and should anyone have any question, the Sci Sec is available to relay questions through to the Co-Leads. The work is running smoothly with active participation of its members who continue to work diligently by emails.

3.4 Patient-Centered Outcome Measures (PCOM)

The PCOM Task Force met in Paris on 30 November 2015 for a one-day workshop, which unfortunately not all members could attend due to travel advisory notice issued by their organisations following the 13 November attack in Paris. Nonetheless, feedback of the workshop background document were forthcoming from members who could not attend as well as other interested individuals who carried out a review as part of public consultation of the document. These comments were circulated to Task Force members for inclusion in their discussions at the workshop.

The focus of the workshop was on knowledge and experience gained so far in the field of outcome measures with common diseases that are transferable to rare diseases. The post-workshop report and recommendations are currently being drafted by the former Project Coordinator of Sci Sec, who is committed to see the completion of this work. A plan was also in place to interview PCOM members not in attendance of the workshop individually after they review the post-workshop report. There will be another public consultation period, and with it opportunities to enrich the report.

A lead for the PCOM Task Force needs to be identified in order to complete the work started and take charge of preparation of a scientific paper to present the workshop conclusion and the recommendations of the Task Force. The aim is to close the work before summer 2016.

3.5 Small Populations Clinical Trials (SPCT)

The SPCT Task Force is also fairly advanced in its work, and a workshop jointly organised with the EMA will take place in London on 3 March 2016. To date, a background document has been prepared by the Sci Sec and reviewed by the Steering Comm. Following a Steering Comm teleconference, additional discussion points were added to the background document, the workshop format decided, and members of the Task Force invited. In parallel, Sci Sec has been working with the EMA on logistical arrangements.

All members of the SPCT Task Force have been sent the pre-workshop background document for review, and they were also solicited for three questions which they would like to address and discuss at the workshop; these questions will be provided to the Steering Comm to aid preparation of the agenda of the workshop. The background document is also publicly available on the IRDiRC website for consultation by IRDiRC Exec and Sci Comms, as well as any interested stakeholders.

A lead for the SPCT Task Force needs to be identified to lead the discussions at the workshop and subsequent report and recommendations writing. The Chair of the TSC will review the composition of the Task Force and discuss with the Sci Sec appropriate next steps.

3.6 Data Mining and Repurposing (DMR)

The work of the DMR Task Force is still in the early stages. A background document prepared by the Sci Sec was sent to the Steering Comm for review, and they are due to meet in a teleconference on 20 January 2016 to provide feedback in relation to the document, identify questions and points of actions for discussions at its workshop, discuss Task Force membership, and identify a potential workshop date and venue. A Steering Committee suggested by email to hold the workshop in November 2016, coupled to a workshop in Barcelona, Spain.

A lead for the DMR Task Force needs to be identified to lead the discussions at the workshop and subsequent report and recommendations writing. The Chair of the TSC will review the composition of the Task Force and discuss with the Sci Sec appropriate next steps.

3.7 Participant Unique Identifiers (PUID)

The PUID proposal was discussed initially in Glasgow by members of the ISC and then subsequently presented to other Sci Comm members, before presentation to the Exec Comm in Montreal. Based on feedback received, the proposal was refined, integrating a number of suggestions for improvement, i.e., inclusion of data protection perspective and opportunity for co-funding with other projects interested in

participant identifiers. The PUID Task Force will be closely associated with IRDiRC Sci Comms whose members are leading the group. An informal expression of interest from GA4GH to collaborate was also received.

The Exec Comm approved the establishment of this Task Force, with additional feedback to take into consideration:

- ▶ Issue of data ownership, security and access
- ▶ Collaborations with other groups also working on solving this problem, e.g., European Reference Networks (ERNs) and European Platform on Rare Disease Registration (JRC-Joint Research Centre, European Commission)
- ▶ Solutions that also address the needs of less developed communities in order to be truly global

4. Scientific Committees membership

4.1 Diagnostics Sci Comm (DSC)

The following members were renewed for a 3-year mandate:

- ▶ Xavier Estivill
- ▶ Gert Matthjis

The nomination of two new members was also approved for a 3-year mandate:

- ▶ Gareth Baynam
- ▶ Anthony Brookes

4.2 Interdisciplinary Sci Comm (ISC)

The following members were renewed for a 3-year mandate:

- ▶ Angel Carracedo
- ▶ Domenica Taruscio

As previously brought up in Montreal, the ISC is seeking a new member with patient voice perspective. A few candidates were being considered by the members and a nomination will be presented to the Exec Comm for approval in due course.

The Chair of ISC also asked for guidance from the Exec Comm on the addition of a member whose expertise lies in health technology assessment. The Exec Comm cautioned that the topic is sensitive in nature and the ISC could focus on research development and global health economics aspect in a broad context; the ISC needs to frame this need more precisely, on questions it wants to address, before exact competency can be identified.

The ISC will prepare a half-page on its need and circulate it to the Exec Comm for comment. This will be an agenda item for discussion at the Op Comm as well as continuous discussion in Lyon.

4.3 Therapies Sci Comm (TSC)

The Chair of TSC would like more time to review the state of the Sci Comm and frame the competences needed at the next Op Comm prior to seeking approval of the Exec Comm. He also suggested approval of renewals via written procedures ahead of the Exec Comm meeting in Lyon.

5. Exec Comm face-to-face meetings

5.1 Lyon, 14-15 March 2016

Members of the Exec Comm were asked to contact the Sci Sec should they have any topic they would like to discuss in Lyon, be it for the Exec Comm meeting or for the joint Exec/Sci Comms meeting.

At present, the Sci Sec is funded through the SUPPORT-IRDiRC contract financed by the EC. However, this contract will terminate in 2018. The EC asked Exec Comm members to think of the future of the Sci Sec and its sustainability, and this will be brought for discussion in Lyon.

5.2 Venue for Fall/Autumn 2016

A host for the Exec Comm meeting in Fall/Autumn 2016 is needed. Any member with a proposition, please contact the Sci Sec as soon as possible. *[Post-meeting note: time is of essence as some members require considerable advance notice in order to request for travel authorisation.]*

5.3 Venue for Spring 2017

Members were asked to consider a venue for the Exec Comm meeting in Spring 2017. This could be coupled with an IRDiRC conference, an event which has been requested by many in the community and presents an opportunity for IRDiRC to celebrate its achievements and formulate new aspirational goals.

6. Any other business

6.1 Progress of investment level

An interest was shown in seeing the track record on financial investment to IRDiRC, i.e., investment level in projects to support rare diseases research and its progress over the year. This is similar to a request made by the Chair of the DSC in Montreal. This information is currently not available, and the Op Comm will look for a way to work on addressing this.

6.2 Reminder re attendance in Lyon

Members of the Exec Comm were reminded to provide information to the Sci Sec via a questionnaire provided of his/her attendance to facilitate meeting logistics organisation.

7. Agenda of the Exec Comm in Lyon

The following will be among the topics of discussion in Lyon; additional suggestions are welcomed:

- ▶ Update of technical review of SUPPORT-IRDiRC
- ▶ Sci Sec work plan and communication strategy
- ▶ The future and sustainability of the Sci Sec
- ▶ Updated IRDiRC Governance document
- ▶ Update on Task Forces leadership and progress of work
- ▶ Interdisciplinary Sci Comm expertise requirement
- ▶ Proposal for a 3rd IRDiRC Conference

8. Next steps and actions

- ▶ Prepare work plan and communication strategy
- ▶ Revise IRDiRC Governance document
- ▶ Review Exec/Sci Comm mandates in relation to Task Forces
- ▶ Review progress and identify leads of PCOM, SPCT, DMR Task Forces
- ▶ Launch work of the PUID Task Force
- ▶ Prepare short document on ISC expertise requirement
- ▶ Review TSC membership and expertise requirement
- ▶ Notify relevant Sci Comm members of renewals and acceptance of nominations
- ▶ Propose to host the Exec Comm in Fall/Autumn 2016
- ▶ Identify way to address the question of investment level in IRDiRC
- ▶ Inform the Sci Sec of (non)attendance in Lyon

Annex - List of participants

<u>Members</u>	<u>Representative</u>
National Center for Advancing Translational Sciences, NCATS/NIH, USA	Christopher Austin
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, Anders Colver, Iiro Eerola
European Commission, DG SANTE, EU	Jarek Waligora
Academy of Finland, Finland	Heikki Vilen
French Foundation for Rare Diseases, France	Marc Tardieu, Roseline Favresse
French Muscular Dystrophy Association, AFM-Téléthon, France	Marie-Christine Ouillade
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Telethon Foundation, Italy	Lucia Monaco
Agency for Medical Research and Development (AMED), Japan	Kazuo Kawamura, Senkei Umehara
The Netherlands Organisation for Health Research and Development, the Netherlands	Sonja van Weely
Korea National Institute of Health, South Korea	Hyun-Young Park
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
National Human Genome Research Institute, NHGRI/NIH, USA	Jeffery Schloss
National Institute of Child Health and Human Development, NICHD/NIH, USA	Melissa Parisi
Pfizer, USA	Katherine Beaverson
Sanford Research, USA	David Pearce

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

Scientific Committees	
Diagnostics	Kym Boycott
Interdisciplinary	Hanns Lochmüller
Therapies	Yann Le Cam

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Anneliene Jonker, Lilian Lau, Ana Rath

Apologies

Members	Representative
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
Agence National de la Recherche, ANR, France	Bertrand Schwartz
Lysogene, France	Karen Aiach
Shire Pharmaceuticals, Ireland	Albert Seymour
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore de Sanita, Italy	Gualtiero Ricciardi
National Institutes of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan	Yoshihiro Yoneda
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
National Institute for Health Research (NIHR), UK	Willem Ouwehand
Isis Pharmaceuticals, USA	Brett Monia
National Cancer Institute, NCI/NIH, USA	Edward Trimble
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIAMS/NIH, USA	Stephen Katz
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	
PTC Therapeutics, USA	Ellen Welch



IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM