



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 13th Executive  
Committee meeting**

26 January 2015



**IRDIRC**

## EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 26 January 2015 by teleconference. The thirteenth meeting of the Exec Comm brought together 27 participants.

Members of the Exec Comm were reminded that they could nominate a replacement to represent their organisations if they are unable to attend any of the meetings.

The Exec Comm discussed the activity plan for 2015:

- ▶ Members of Working Groups (WGs) will shortly be informed of IRDiRC restructuring
- ▶ Nominations of Task Force (TF) members will be requested in the coming days thus to begin work on selected topics and the relevant workshop organisations
- ▶ Work on small population clinical trials, matchmaker exchange and patient relevant/ reported outcome measures are ready to commence
- ▶ Work on computable consent forms is on hold until further Interdisciplinary Scientific Committee (Sci Comm) discussion; it may be replaced by a workshop on repurposing and data mining should the committee decide to postpone or cancel it
- ▶ The steps of pre-workshop methodology were outlined
- ▶ The “IRDiRC Recommended” process is adopted, and the finalised common phenotype terminology completed by the ICHPT will be awarded the “IRDiRC Recommended” label.
- ▶ The finalised common phenotype terminology will soon be publicly available

A number of modifications to the IRDiRC Governance document were proposed to reflect changes in the consortium. The revised document has been adopted, provided the final text incorporates point-by-point discussions agreed during the teleconference call.

The draft State-of-Art will be discussed in the next Exec Comm face-to-face meeting in Madrid. Members of the Exec Comm are requested to review the document, its goal and methodology, and provide feedback, be it in person or by email in advance of the meeting if unable to attend.

Two nominations of members to the Therapies Sci Comm were approved by the Exec Comm for a 3-year mandate. Nine renewals of members from all three Sci Comms were also approved.

The Exec Comm explored the idea of admitting a new patient organisation to be member of IRDiRC and will return to this topic in Madrid. The Chair suggested exploring the subject of other organisations wishing to be associated/affiliated with IRDiRC in Madrid.

The Sci Sec is currently preparing an anonymous performance review survey which will be sent out shortly to all members of the Exec Comm and Sci Comms; it is important that all members take a few minutes to complete this survey as they constitute the Scientific Advisory Board of “Support IRDiRC”.

In terms of IRDiRC communications, the Exec Comm agreed on:

- ▶ Making the IRDiRC newsletter available to individuals wishing to receive it
- ▶ Poster presentation on “IRDiRC Recommended” at the ESHG meeting in Glasgow
- ▶ Reorganisation/update of IRDiRC website to present successful project-based initiatives more prominently e.g. Matchmaker Exchange, common phenotype terminology

The Exec Comm also briefly discussed other topics that would be included in the agenda of the face-to-face meeting in Madrid.

## REPORT

### IRDiRC activity plan for 2015

#### Finalisation of Working Group activities

It was decided during the Exec Comm meeting in Shenzhen to bring the two-year period of brainstorming to a close and turn towards specific, actionable priority topics.

The Chair of the Diagnostics Sci Comm has spoken to all but one of the Chairs of the WGs to communicate the decision made, proposed to reorganise active members of the Genome/Phenome WG into the Matchmaker Task Force (TF), and suggested one further teleconference call for the Population Controls Variant Datasets WG to make its product available before winding down. *[Post meeting note: a last teleconference of Ontologies WG will also be held.]*

The Chair of the Therapies Sci Comm is following up on two elements: one on orphan drug guidelines which will involve further discussion with EMA and FDA, and one on discussion with the Chair of Orphan Drug-Development and Regulatory Processes WG.

The Chair of the Interdisciplinary Sci Comm plans to discuss the decision of the Exec Comm with its committee in early February, through a teleconference, before informing the WGs of the upcoming changes.

An official letter to the WGs, to thank them for their contributions and to inform them of future plans, is being drafted and will be sent out in the coming days. TFs will be constituted after the WG members receive the letter.

#### Priority projects and establishments of Task Forces

##### 2.1 Computable consent forms

This project was selected for action in 2015 and is based on a proposal related to consent forms previously submitted to the Exec Comm; however, the Chair of the Interdisciplinary Sci Comm has not yet discussed this with the other committee members to obtain their feedback on the modification to this proposal and to make the consent forms computable.

A potentially strategic move would be to explore a joint workshop with Global Alliance (GA4GH) to accelerate the work already done so far. The Interdisciplinary Sci Comm will investigate the feasibility of creating a dynamic and computable system; it will be difficult given the different

international/federal/local laws applicable as well as personal wishes of individuals, but it is nonetheless an important initiative if it is developed.

## 2.2 Small population clinical trials

The project is set to start once the TF can be constituted. The EMA is ready to jointly organise this workshop at its facility. As there is a continuous discussion in EMA/FDA regarding coordination of their guidelines on small population clinical trials, the IRDiRC workshop will be based on exploring the evolution of these guidelines and their possible convergence, and the next steps of scientific projects involved in this pipeline.

## 2.3 Data standards clearinghouse

This is a project that the Scientific Secretariat (Sci Sec) is working on, under the supervision of a member of the Interdisciplinary Sci Comm, who initiated this project and is the project leader.

## 2.4 Matchmaker Exchange

A primary workshop took place in Miami on 20-21 January, aiming at finalizing the API to connect matchmakers via Matchmaker Exchange (<http://matchmakerexchange.org/>). The pilot API was launched a week prior and needs further work. A second workshop may be planned for June/July 2015, before the IRDiRC workshop in conjunction with the ASHG meeting in October 2015 which will have broader engagement with different parties and advertise the project.

Currently, there are resources to solve the technical issues on the version 1 of the API, but as Matchmaker Exchange gets larger, further resources are required and funding opportunities are currently being explored. There is also ongoing work on a special issue of Human Mutation which will describe the Matchmaker API and talk about the other matchmakers, slated for publication also in conjunction with the ASHG meeting and IRDiRC workshop.

## 2.5 Patient relevant/reported outcome measures

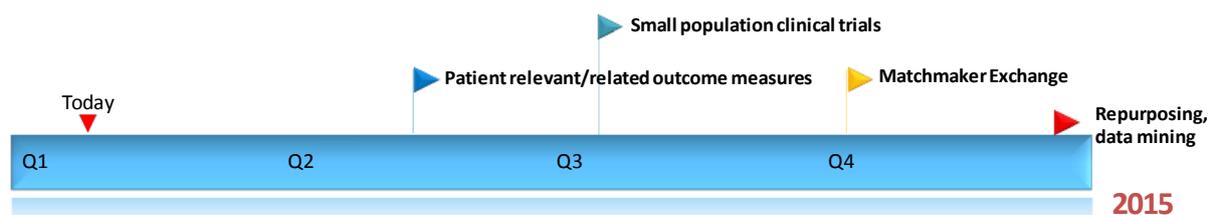
This project is also ready to start. This is an area where IRDiRC should target key players including regulatory bodies, outcome measure centres and industry, and invite them to act on outcome measures for rare diseases, develop necessary concepts and guidelines, and advance the HTA process.

## 2.6 Repurposing and data mining

This topic was initially selected for action in 2016, but included in the activity plan as potential replacement to Computable Consent Forms, should the latter be postponed.

### IRDiRC workshops

The proposed workshop timeline for 2015:



As workshops get organised, the Exec Comm will be informed in real time of the location and the time when they will take place. Members of the Exec Comm may attend as observers and keep a closer tab of these exciting areas.

The following steps were outlined as pre-workshop methodology:

- ▶ Nomination of TF members by the Exec Comm, Sci Comms and Sci Sec
- ▶ Appointment of TF members by Sci Comms following organised voting
- ▶ Sci Sec to carry out analysis and develop draft background paper
- ▶ Sci Sec to contact each TF member to discuss and finalise the concept paper
- ▶ Sci Sec to assist TF members in the development of the programme
- ▶ Finalised report and programme to be validated by TFs

The request for nomination of TF members will be issued as soon as WG members have been informed of the restructuring, together with relevant descriptions. In the meantime, members of the Exec Comm could give a thought to potential nominees based on the broad scope of the TFs, as described in the "Proposed IRDiRC Activity Plan for 2015".

*[Post-meeting note: the workshops will be organised under the IRDiRC umbrella and not attached to a specific Sci Comm, therefore Chairs of all Sci Comms will be equally consulted.]*

## **Dissemination of the common phenotype terminology (ICHPT)**

The final version of the terminology by the International Consortium for Human Phenotype Terminologies (ICHPT) has been received, and could be launched after the information is made available on a dedicated website. An announcement will be made on the IRDiRC website and to the rare disease community, and this will also be written up as an article.

## **Change of IRDiRC Governance document**

A revision to the IRDiRC Governance was proposed in light of recent changes, including the maturation of IRDiRC from high level discussion on principles and areas to coordinate activities to more specific and focused activities thus the decision to restructure WGs and to introduce TFs, the voting in of a Vice Chair, the admission to the Exec Comm of the Project Coordinator of the Sci Sec, and the changing mandates of the Sci Comms and the Sci Sec.

The revised IRDiRC Governance is adopted, with the understanding that the text of the draft document would be edited to incorporate all point-by-point discussions agreed during the call. The document will be made available on the IRDiRC website together with the previous version.

## **“IRDiRC Recommended” process**

Following a discussion at the Exec Comm meeting in Shenzhen, the “IRDiRC Recommended” process was refined. A summary of the review process, the technical requirements and the plan to open the call of submissions was provided for discussion. The Exec Comm agreed to adopt this document.

## **Feedback on draft State-of-Play report**

The preparation of an annual State-of-Play (SoP) report is one of the deliverables of the Support-IRDiRC contract. A draft SoP was disseminated to the member of the Exec Comm prior to the face-to-face meeting in Shenzhen; however, there has been limited feedback on its content, relevance and usefulness, and the Sci Sec would appreciate guidance from the Exec Comm.

This will be a discussion item at the next face-to-face Exec Comm meeting in Madrid. Members of the Exec Comm are requested to review the document, its goal and methodology, and provide their comments at the meeting; members unable to attend should send the comments in advance of the meeting to the Sci Sec, who will compile the received feedback.

### **Nominations of Sci Comm members**

The nomination of two new members of the Therapies Sci Comm was approved by the Exec Comm for a 3-year mandate. The new members are:

- ▶ Dr Sandrine Marreaud (nomination by EORTC)
- ▶ Dr Diego Ardigò (nomination by Chiesi Farmaceutici)

### **Renewals of Sci Comm members**

The following members of the Sci Comms were renewed for a 3-year mandate:

Diagnostics Sci Comm:

- ▶ Han Brunner
- ▶ Michael Bamshad
- ▶ Milan Macek

Interdisciplinary Sci Comm:

- ▶ Bartha Knoppers
- ▶ Jack Goldblatt
- ▶ Jeffrey Krischer
- ▶ Samantha Parker

Therapies Sci Comm:

- ▶ Gert-Jan van Ommen
- ▶ Adam Heathfield

### **Admission of new patient advocacy group**

When IRDiRC was founded, there wasn't an active and prominent patient advocacy group in Asia to represent the region at the Exec Comm. As IRDiRC is going global, the Chair proposed the admission of an emerging key patient group in China, and for it to be granted similar status as that of EURORDIS, NORD and Global Alliance.

However, as the number of patient groups is increasing and becoming more organised, it may be better to identify a representative that has been agreed upon among the different patient groups active in their geopolitical regions (e.g. Asia-Pacific, Latin America), instead of targeting specific organisations. As not everyone has the same sense of which organisations exist and have been active advocates, it may be useful to first build a map of patient organisations and provide an overview of their landscape today; only then can an informed choice be made.

This topic will be further discussed in Madrid and a mapping of the patient organisations will be provided. Members of the Exec Comm were asked to think of possible ways to achieve globalising patient representations in IRDiRC.

### **Outreach and international relations**

Additional requests from organisations wishing to be linked to IRDiRC will be further discussed in the next Exec Comm face-to-face meeting in Madrid.

### **IRDiRC membership fees**

The Chair thanked the members for the responses received so far to the request of voluntary membership fees. However, some members have not yet responded whether they will put through a contribution or if they are not in the position to do so. The Chair requested, in order to facilitate budget planning, that members inform him of their position if they have not already done so.

The funds raised will be used to pay for representatives of patient organisations to attend the Exec Comm meetings in person, and potentially for various outreach activities although none is specially planned at present. The use of funds will be accounted to the Exec Comm members on an on-going basis.

### **Support-IRDiRC performance review survey**

The submission of a performance review report is one of the deliverables of the Support-IRDiRC contract. An anonymous survey is currently in preparation and will be shortly sent to all members of the Exec Comm and the Sci Comms to evaluate the performance of the Sci Sec. It is important that all members take a few minutes to complete this survey as they constitute the Scientific Advisory Board of “Support IRDiRC”.

### **IRDiRC communications**

#### **IRDiRC newsletter**

Until present, the IRDiRC newsletter is an internal newsletter, sent to all members of the Exec Comm, Sci Comms and WGs. The Sci Sec recently received requests to be included in the mailing list in order to be informed of IRDiRC activities. The Exec Comm supported the provision of newsletter to interested individuals, either via a registration form or an email request.

## **Publication of IRDiRC Roadmap**

It would be desirable to publish the new roadmap of IRDiRC involving plans on priority topics and actions, both on the IRDiRC website and in a peer-reviewed journal.

## **Poster presentation at ESHG**

The poster to be presented at ESHG would focus on the “IRDiRC Recommended” endorsement.

A question of work attribution was raised and members were asked to consider how will IRDiRC identify the origins and authorship of poster and papers for publication. Members were asked to reflect on this and further discussion will be carried out on this issue in Madrid.

## **Evolution of IRDiRC website**

As IRDiRC is moving in the direction of project-based initiatives to achieve its goals and facilitate collaborations, the website should be updated to reflect on its activities. Information on successful initiatives such as Matchmaker Exchange and ICHPT’s Common Phenotype Terminology should be made prominent. Presentations and posters related to these activities should also be made available if the authors/presenters give permission to do so.

A website review and reorganisation has been planned by the Sci Sec.

## **Agenda of face-to-face meeting in Madrid**

Members of the Exec Comm were reminded that they could nominate a replacement to represent their organisations if they are unable to attend any of the meetings.

In addition to the topics agreed for discussion (i.e. feedback of SoA, globalisation of patient organisations, affiliation to IRDiRC, authorship attribution), these were suggested:

- ▶ Roundtable with “state of play” of funding initiatives
- ▶ Update re Task Force formations and workshop organisations
- ▶ Update re “IRDiRC Recommended” introduction and implementation

The meeting in Madrid will be a full day meeting, starting at 9.00/9.30 am and running until around 6.00 pm.

**Annex - List of participants**

<b><u>Members</u></b>	<b><u>Representative</u></b>
Western Australian Department of Health, Australia	Hugh Dawkins
European Organisation for Treatment & Research on Cancer, EORTC	Denis Lacombe
Canadian Institutes of Health Research, Canada	Paul Lasko
Genome Canada	Pierre Meulien
WuXi AppTec Co. Ltd., China	Mao Mao
E-RARE-2 Consortium, EU & ANR, France	Daria Julkowska
European Commission, DG Research and Innovation, EU	Anders Colver
European Commission, DG Health and Consumer Protection, EU	Jaroslawn Waligóra
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Shire Pharmaceuticals, USA	Albert Seymour
Telethon Foundation, Italy	Lucia Monaco
The Netherlands Organisation for Health Research and Development	Sonja van Weely
Carlos III Health Institute, Spain	Pedro Cortegoso Fernández
Food and Drug Administration, USA	Katherine Needleman
National Eye Institute, NIH, USA	Santa Tumminia
National Human Genome Research Institute (NHGRI), NIH, USA	Jeffery Schloss, Lu Wang
PTC Therapeutics, USA	Diane Goetz
Sanford Research, USA	David Pearce

<b><u>Invited Patient Advocacy Groups</u></b>	
EURORDIS, Europe	Béatrice de Montleau
Genetic Alliance, USA	Sharon Terry

<b><u>Scientific Committees</u></b>	
Diagnostics	Kym Boycott
Interdisciplinary	Hanns Lochmüller
Therapies	Yann Le Cam

<b><u>IRDIRC Scientific Secretariat</u></b>	
SUPPORT-IRDIRC Project	Ségoène Aymé, Lilian Lau, Antonia Mills

## Apologies

<b><u>Members</u></b>	<b><u>Representative</u></b>
BGI, China	Ning Li
Chinese Rare Diseases Research Consortium, China	Qing Wang
Academy of Finland, Finland	Heikki Vilen
AFM- French Association against Myopathies, France	Marie-Christine Ouillade
Fondation Maladies Rares, France	Nicolas Lévy
Lysogene, France	Karen Aiach
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
Istituto Superiore de Sanita, Italy	Fabrizio Oleari
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Prosensa, The Netherlands	Luc Dochez
Korea National Institute of Health, Korea	Hyun-Young Park
National Institute for Health Research, UK	Willem Ouwehand
Genzyme, USA	Carlo Incerti
Isis Pharmaceuticals, USA	Brett Monia
NIH National Center for Advancing Translational Sciences, NCATS, USA	Christopher Austin
National Cancer Institute, NIH, USA	Edward Trimble
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIH, USA	Stephen Katz
National Institute of Child Health and Human Development, NIH, USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke, NIH, USA	Danilo Tagle
NKT Therapeutics, USA	Robert Mashal
Office of Rare Diseases, USA	Pamela McInnes
<b><u>Invited Patient Advocacy Groups</u></b>	
National Organization for Rare Diseases, NORD, USA	Peter Saltonstall



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