



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 6<sup>th</sup> Executive  
Committee meeting**

25 November 2013



**IRDIRC**

## EXECUTIVE SUMMARY

The Executive Committee (Exec Com) of the International Rare Diseases Research Consortium (IRDiRC) met on 25 November by teleconference. The sixth meeting of the Exec Com brought together 31 participants including 22 Exec Com members and representatives of the 3 Scientific Committees (Sci Com).

The Interdisciplinary Sci Com presented recommendations for the IRDiRC roadmap to the Exec Com to discuss. These recommendations are divided in three parts:

- ▶ Recommendations on IRDiRC activities and structures
- ▶ Recommendations for implementing IRDiRC policies
- ▶ Recommendations for funding priorities directed at IRDiRC funded organizations

A few recommendations were discussed during the teleconference and members of the Exec Com and the Sci Com were invited to provide further comments on the document offline. A revised version of the documents will be submitted to a subsequent meeting for approval.

It was decided that the next international IRDiRC conference will be held in Shenzhen, China, in November 2014, hosted by BGI, to improve the profile of IRDiRC and rare diseases research in the Asia-Pacific region. A committee gathering the chair of the Exec Com, a representative of each of the Sci Com and representatives of BGI will prepare the scientific program of the conference.

A generic letter of intent will be developed for private partners willing to join IRDiRC. This letter will address issues specific to the private sector such as intellectual property, data sharing, etc.

The chair of the Exec Com proposed the implementation of an annual membership fee based on voluntary participation to establish a funding pot to support travel of patient organizations to attend Exec Com face-to-face meetings, outreach activities and cost of hosting Exec Com meetings. The proposition will be open to vote at a subsequent meeting.

It was confirmed that the next face-to-face meeting of the Exec Committee will be held in Berlin on May 7-8, hosted by the German Federal Ministry of Education and Research (BMBF).

## REPORT

### Report from the Interdisciplinary Scientific Committee

The chair of the Interdisciplinary Scientific Committee presented to the Exec Com recommendations for the IRDiRC roadmap, drafted by this Sci Com during their last meeting in October in Montreal, and based on the inputs received from the 4 ISC Working Groups which had 1-2 teleconferences (Biobanks; Data sharing and Bioinformatics; Ethics and Governance; Registry and Natural History). These recommendations should be consolidated in function of the recommendations of the Diagnostics Sci Com and Therapies Sci Com.

The recommendations are divided in 3 chapters and some recommendations from each chapter were discussed.

#### **Chapter 1: Recommendations on IRDiRC activities and structures (for the IRDiRC Executive Committee and Secretariat)**

Recommendation 1: *IRDiRC executive committee should take a more proactive approach regarding proposed data protection legislation in the EU, as current draft will jeopardize many of their funded programmes in health, especially rare disease research (arguments: waste of tax payers' money, lost opportunities for academic and commercial research and development, contrary to patients' expectations). This is for immediate consideration!*

An action was already taken on that item. The Interdisciplinary Sci Com wrote a letter to the Committee on Civil Liberties, Justice and Home affairs (LIBE committee) to express concern regarding certain amendments in the Compromise Text of the General Data Protection Regulation. This letter, co-signed by the chair of the Exec Com and the chair of the Interdisciplinary Sci Com, was sent without the consent of the Exec Com as it was a time sensitive matter.

It was decided to take down this letter from the IRDiRC website as it creates a conflict of interest for some IRDiRC members. Moreover, in the future, any standpoint in the name of IRDiRC should get approval by the Exec Com.

IRDiRC members are however encouraged to individually contact key members of the European Parliament if they wish to influence the decision of the European Parliament on the topic. In addition, a letter of concern signed by the Interdisciplinary Sci Com, whose members are nominated as individuals and not representative of an institution, could be publicized on the IRDiRC website.

### Recommendation 2:

- ▶ *Review further objectives/remit of the 4 WGs related to the ISC. Some WGs may consider merging or have joint meetings as there are several transversal issues (avoid redundancy/duplication),*
- ▶ *Merging WGs in one single working group related to ISC and implementing ad hoc groups should be discussed with the WGs if many overlaps are identified. Focused/ad hoc groups within this newly merged WG should be considered and may operate in a task-specific and time-limited way.*

The Interdisciplinary Sci Com raised the question of the longevity of the Working Groups (WG) as the topics of the different WGs are interlinked and it may be necessary in the future to merge, split or create new groups.

The Exec Com members reminded the Sci Com that the organization of the WGs was always destined to be dynamic and that the reorganization of the WGs depends on each Sci Com.

Recommendation 3: *IRDiRC has signed up to the global alliance, which is seen as a major opportunity for RD research (along with cancer). Executive committee should determine their role in the global alliance; a MoU (Memorandum of Understanding) will be requested by the global alliance in early 2014.*

The Exec Com considers that it is important to keep communicating with global alliance but that there is not enough information for the moment to adopt a standpoint regarding the role of IRDiRC with the global alliance. This item will be further discussed when more information are available. A member of the Exec Com, who is also a member of the global alliance steering committee, specified that global alliance wishes to stay a collective of institutions and other organizations, and thus will not be as well-formed and structured as IRDiRC and that there will certainly not be any policies documents besides the white paper.

## **Chapter 2: Recommendations for implementing IRDiRC policies (directed at all IRDiRC associated organizations)**

Recommendation 4: *Support a “Clearing House” for data standards (genome, phenome, clinical trial data, etc). Rather than being prescriptive what the best standard is that everybody should be using, the Clearing House will collect information about the standards, and make them publically available (and give people the opportunity to submit their standards, and possible use cases).*

The WG on Data sharing and Bioinformatics recommend an initiative to collect and publicize existing standards, which would help data sharing across projects funded by IRDiRC members.

Members of the Exec Com suggested inquiring what already exists about data sharing to determine what can be used and improved to have a more global view. Existing initiatives were mentioned: Institute of Medicine Consensus Committee (standards for data sharing), International Consortium of Human Phenotype Terminology (common terminology for human phenotype) and another initiative working on a search tool to help with unsolved exome sequencing.

The Exec Com supports the idea of this project and recommends the use of the IRDiRC website to publicize these standards and use cases.

***Recommendation 5:** Promote innovative approaches to licensing that favour the attribution of Intellectual Property (IP) (e.g. non-exclusive licensing) and allow RD patients to access treatments; investigate the role of IP to translate findings/knowledge into accessible treatments for RD patients.*

The input from the private sector would be highly valuable on that topic.

### **Chapter 3: Recommendations for funding priorities directed at IRDiRC funding organizations**

***Recommendation 6:** Specific funding should be dedicated to explore the impact of revealing “incidental (unanticipated) findings” to patients and their families.*

The Interdisciplinary Sci Com thinks that it would be highly beneficial to fund social research on patients’ opinion on sequencing and data sharing, clinical trials children in research, etc. These studies should include the point of view of patients but also researchers.

Members mentioned that these societal aspects are being studied or in the process of being studied in some programs (Care for Rare, NIH return on research).

The difficulty of launching international calls for funding such research was raised, although a few IRDiRC members may be able to fund international calls.

- ▶ The recommendations document drafted by the Interdisciplinary Sci Com will be accessible on the private section of the IRDiRC website for comments from the Exec

Com members and the other Scientific Committees. A revised version will be presented at a subsequent Exec Com teleconference for approval.

### **Update on the IRDiRC conference in Shenzhen, Nov 2014**

The possibility of holding an IRDiRC conference in China in November 2014 was discussed. BGI proposed to sponsor and host the conference in Shenzhen, their core location, and will take the financial risk of the organization of this conference. China appears to be a good location to improve the profile of IRDiRC and rare diseases research in the Asia-Pacific region as a few initiatives developed recently such as the creation of the Chinese Rare Diseases Research Consortium, the creation of the Chinese Genetic Alliance, etc.

In addition to the location of the meeting, the Exec Com agreed that the conference should be international, and based on the same concept as the IRDiRC conference held in Dublin, with mostly international speakers. A committee composed of the Exec Com chair, one representative of each Scientific Committee and representatives of BGI will develop the scientific program. This committee will meet once in Shenzhen but mostly work by teleconference. The Chinese Rare Diseases Research Consortium, member of IRDiRC, should be included in the organization of the conference. The date of the conference should be formalized soon.

### **Modification of the 'letter of intent' for Industry**

In the past few months, there was ongoing discussion with a biotechnology company that showed interest in joining IRDiRC but was concerned about the language in the IRDiRC policies and guidelines document concerning intellectual properties, patient and data privacy, etc. At the request of the Exec Com chair, this company submitted a letter of intent with modifications to address these issues. These modifications were reviewed by the subcommittee on industry's position, created in Miami in September, and IRDiRC representatives for the European Commission. None reported conflict with the wording employed by this company, which was thus invited to formally apply to IRDiRC.

Considering the situation, the Exec Com conceded that a generic letter of intent should be developed for private partners willing to join IRDiRC and that current IRDiRC members from the private sector are allowed to modify their letter of intent to reflect these issues. The Sci Sec will draft this letter of intent for industry.

### **Discussion on the possibility to establish a funding pot**

It was previously mentioned that a small amount of IRDiRC funding would be useful for various purpose, such as funding travel for patient organizations to attend Exec Com face-to-face meetings, funding outreach activities and funding cost of hosting Exec Com meetings. A possibility is to create an annual membership fee – couple thousands dollar per year – based on voluntary participation. This funding would be administered by the Sci Sec, independently of the Support-IRDiRC funding. Although a few members already expressed their willingness to participate, the subject will be further discussed at the next meeting to give time to the IRDiRC members to reflect on the proposition.

### **Other topics**

- ▶ It was confirmed that the next face-to-face Exec Com meeting will be held in Berlin on May 7-8, hosted by the German Federal Ministry of Education and Research (BMBF).
- ▶ The members of the Exec Com are supportive of the idea of holding regular teleconferences as it is productive.

**Annex - List of participants**

<b>Members</b>	<b>Representative</b>
Western Australian Department of Health, Australia	Hugh Dawkins
IRDiRC Chair Executive Committee, Canadian Institutes of Health Research, Canada	Paul Lasko
Genome Canada, Canada	Pierre Meulien
BGI, China	Ning Li
E-RARE-2 (E-Rare Group of Funders), Europe	Daria Julkowska
European Commission, (DG Health and Consumer Protection), EU	Stefan Schreck
European Commission, (DG Research and Innovation), EU	Iiro Eerola and Anders Colver
EURORDIS ( Patient Advocacy Group), Europe	Béatrice de Montleau and Valentina Bottarelli
Academy of Finland, Finland	Aki Salo
ANR- French National Research Agency, France	Natalia Martin
AFM- French Association against Myopathies, France	Marie-Christine Ouillade
Lysogene, France	Fanny Vincent
Federal Ministry of Education and Research, Germany	Ralph Schuster
Children's New Hospitals Management Group, Georgia	Oleg Kvlividze
Korea National Institute of Health	Hyun-Young Park
Carlos III Health Institute, Spain	Rafael de Andreas Medina
Food and Drug Administration, USA	Katherine Needleman
Genetic Alliance, USA	Sharon Terry
National Center for Advancing Translational Sciences, NIH, USA	Christopher Austin
National Human Genome Research Institute, NIH, USA	Jeff Schloss and Wang Lu
NORD, USA	Peter Saltonstall
Office of Rare Diseases, USA	Stephen Groft
<b>Scientific Committees</b>	
Diagnostic Sci Com	Milan Macek
Chair Interdisciplinary Sci Com	Hanns Lochmüller
Therapies Sci Com	Maria Mavris
<b>IRDiRC Scientific Secretariat</b>	
SUPPORT-IRDiRC project	Ségolène Aymé , Barbara Cagniard and Sophie Höhn

**Apologies**

<b><u>Members</u></b>	<b><u>Representative</u></b>
Chinese Rare Disease Research Consortium, China	Qing Wang
Instituto Superiore de Sanita, Italy	Enrico Garaci
Shire, Ireland	Phil Vickers
Telethon Foundation, Italy	Lucia Monaco
Prosensa, The Netherlands	Luc Dochez
The Netherlands Organisation for Health Research and Development, The Netherlands	Sonja van Weely
National Institute for Health Research, United Kingdom	Willem Ouwehand
National Cancer Institute, NIH, USA	Edward Trimble
National Eye Institute, NIH, USA	Santa Tumminia
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIH, USA	Stephen Katz
National Institute of Child Health and Human Development, NIH, USA	Alan Guttmacher
National Institute of Neurological Disorders and Stroke, NIH, USA	Danilo Tagle
NKT Therapeutics, USA	Robert Mashal
PTC Therapeutics, USA	Diane Goetz
Sanford Research, USA	David Pearce



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