



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 2<sup>nd</sup> Executive  
Committee meeting**

25-26 September 2012



**IRDIRC**

## EXECUTIVE SUMMARY

The Interim Executive Committee (Exec Com) of the International Rare Diseases Research Consortium (IRDiRC) met on 25-26 September 2012 in Evry France. This second meeting of the Exec Com was kindly hosted by IRDiRC member AFM and brought together eighteen members and three of the newly elected Chairs of the IRDiRC Scientific Committees (Sci Com).

The Exec Com discussed the next steps and tentative timelines with regard to finalising a first version of the IRDiRC Policies and Guidelines Document (Policy Document) and the preparation of the IRDiRC conference 2013. In addition, a proposal for a first list of working groups to be set up under each Sci Com was approved (see list below) and a number of other governance issues, including how to ensure a balanced representation on the Exec Com, were discussed.

Moreover, the Exec Com was presented with an indicative outline of the support action "Support-IRDiRC" funded by an EU grant (subject to signature) and dedicated to running the IRDiRC secretariat.

The Exec Com also decided that Dr Paul Lasko, Scientific Director, Canadian Institutes of Health Research (CIHR), should become the next Chair of the Exec Com as the interim period comes to an end in December 2012. The Chairmanship should be handed over during a transition period ending in April 2013. A Vice Chair should be appointed and nominations should be sought among the members of the Exec Com.

The next Executive Committee meeting will be organised in Barcelona in January 2013.

### List of initial IRDiRC Working Groups

The Exec Com agreed on the initial set up of the Working Groups as follows:

#### **Diagnostics committee:**

- ▶ Ontologies and disease prioritisation
- ▶ Sequencing
- ▶ Model systems
- ▶ Genome / Phenome

**Interdisciplinary committee:**

- ▶ Ethics and governance
- ▶ Biobanks
- ▶ Registries and natural history
- ▶ Bioinformatics and data sharing

**Therapeutics committee:**

- ▶ Biomarkers for disease progression and therapy response
- ▶ Re-purposing+ small molecules
- ▶ Advanced therapies
- ▶ Regulatory aspects and bottlenecks hampering orphan drug development

## REPORT

### IRDiRC policy document

The Chairs of the Sci Coms provided an update on the timelines for completing a first revision of the IRDiRC Policy Document. The document will require substantial revisions and the Chairs of the Sci Com will agree on a process that should ensure timely and balanced input from all Committees. It was suggested that the final Document should contain twelve sections and that each section should be followed by not more than two policies and four guidelines. A draft document will be provided to the Exec Com in December for discussion at its next meeting in January 2013 (see *next meeting* section). A final first version of the document will be approved by the Exec Com in April 2013 (by a majority vote).

### Annual Conference

The first annual IRDiRC conference, with the aim to network projects funded in line with IRDiRC objectives and to take stock of progress toward IRDiRC goals, will be held in Dublin, Ireland on 16-17 April 2013. The conference will be organised under the auspices of the Irish EU Presidency.

With regard to the conference programme it was decided that the Chairs of the Sci Coms will work up a proposal for the thematic sessions and that the current and incoming Chairs of the Exec Com will be responsible for the plenary sessions. The Chairs of the Exec Com and Sci Com will take care of the overall coordination of the conference programme.

A draft programme should be ready for approval by the Exec Com by end of November and the final programme should be approved by the Exec Com meeting in January 2013.

It was agreed that during a first period (end of November 2012 – January 2013) conference registrations will be by invitation only whereafter registration will be open to the public. Each Exec Com member is responsible for identifying and inviting suitable conference participants (members of funded projects). A "save-the-date" message for dissemination will be sent to the Exec Com members by the Chair in October and an invitation message giving access to the conference on-line registration facility will be circulated in the end of November (subject to the completion of the draft conference programme).

A poster session will be organised and participants will be able to submit project posters. It will be investigated if a prize for the best posters could be arranged by one of the members of the Executive Committee.

*Post-meeting note:* Following a suggestion to extend the length of the conference programme, the IRDiRC interim secretariat can confirm that it would not be possible to start the conference on 15 April. However, the programme of the second conference day (17 April) could be extended if needed.

### **IRDiRC Working groups**

It was decided that the mandate of each IRDiRC Working Group will be established by the Sci Coms. Working Groups should be time limited and objective focused, however, their roles could change over time responding to the needs in the respective sectors. To be operational, each Working Group should have about 5-10 members. The Working Groups should be composed of representatives from projects funded by the Exe Com members which adhere to IRDiRC policies, other specific invited experts and patient representatives. Expressions of interest could be launched to look for suitable candidates. Exec Com members can also suggest names for Working Groups. Exec Com will agree on the composition of the Working Groups based on the proposals by the Sci Coms. Working Groups can organise their meetings via tele- and videoconferencing or webcasting. All Working Groups should meet face to face in the IRDiRC Conference to be organised in Dublin on 16-17 April 2013.

The Exec Com agreed on the initial set up of the Working Groups as follows:

#### **Diagnostics committee:**

- ▶ Ontologies and disease prioritisation
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- ▶ Model systems
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#### **Interdisciplinary committee:**

- ▶ Ethics and governance
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### **Therapeutics committee:**

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### **Governance – Scientific Committees**

The Exec Com pointed out that the Sci Com members have been nominated in their personal capacity. Replacements of Sci Com members in their meetings are strongly discouraged. Replacements could be however accepted exceptionally upon the approval of the Sci Com chairs. In cases of no material contribution or consecutive absences by a Sci Com member, the Sci Com chairs should alert the Exec Com which should then consider action. Nomination of the new member of the Therapeutic Scientific Committee was postponed.

The Exec Com decided that individuals from regulatory bodies can be invited as advisors to the Scientific Committees as appropriate. Sci Com chairs shall send proposed nominations/invited advisors to the Exec Com for approval.

### **Governance – Executive Committee**

The Exec Com noted that replacements of Exec Com members are possible as Exec Com members represent their organisations. Nominations of alternate representatives should be made by the authorising person in the organisation.

IRDiRC Secretariat will circulate a list of Exec Com members and their e-mail addresses among the Exec Com members.

Related to the new Exec Com member E-rare ERA-NET, it was noted that funding by the organisations already members of IRDiRC is additional to already committed funds to IRDiRC. It was agreed that in addition to the designated representative, a proxy-delegate will be invited on rotational basis.

Related to the inclusion of regulatory bodies in the Exec Com, it was pointed out that the Exec Com decided already in January 2012 to invite regulatory bodies in advisory capacity to the Exec Com. It was noted that FDA's funding program is a member of Exec Com as a research funder not as a regulator.

The Exec Com discussed also the potential need for the limitation of the proportion of for-profit organisations to public organisations in the Exec Com. It was concluded that flexibility should be built in the Governance document to facilitate flexibility of representation at the Exec Com to ensure balanced representation of different stakeholders.

On the wider participation of public health care authorities in IRDiRC, the Exec Com acknowledged that the issues such as reimbursement and uptake of innovations in health care are important, but emphasised that the IRDiRC should focus on R&D issues.

### **Chairmanship of Executive Committee**

The mandate of the Interim Chair of the Exec Com, Dr Ruxandra Draghia-Akli (Director, Health Directorate, European Commission) will end in December 2012. Following a request for nominations, Dr Paul Lasko (Scientific Director, CIHR) was elected to take over the Chairmanship of the Exec Com. It was decided that the transition period to the new Chairmanship would end in April 2013 to coincide with the IRDiRC annual conference. It was also decided that a Vice Chair should be appointed among the members of the Exec Com.

### **IRDiRC support action**

A project outline of the support action dedicated to running the IRDiRC secretariat was presented by the coordinator Dr Ségolène Aymé. The support action would kick off on 1 October 2012 for a period of six years and ensure the smooth running of IRDiRC committees and working groups. It would also be responsible for communication and dissemination activities. The project will moreover create a database with projects funded under the IRDiRC umbrella. The Exec Com welcomed the presentation and also decided to adopt the IRDiRC logo presented by the coordinator as part of the support action's communication plan.

### **General Policy Issues**

Discussion took place also on how to facilitate public-private collaborations in translational research and attract investments in the area. The Exec Com concluded that once IRDiRC is more mature, one of the annual IRDiRC meetings could be dedicated to partnering IRDiRC projects, companies, Venture Capitals, business angels and other funders. Support IRDiRC could also tailor outreach activities to VCs, bio-techs and conferences.

Diverse prevalence criteria are used in the definition of the rare diseases in different regions of the world. The Exec Com concluded that each region's definition will be respected and ranges of prevalence of the disease could be presented in communications on funded projects.

The last item discussed was how to report funding contributing to IRDiRC. The Exec concluded that the investments by each funder should be tracked and highlighted at the IRDiRC website (to be done by Support IRDiRC). It should be stated that different funding bodies are using different cost models for reporting direct costs and overheads.

### **Next meetings**

The next Exec Com meeting should be held in January 2013. The Chair will follow up with the Exec Com members to find suitable meeting dates and a meeting venue. A second meeting in 2013 is planned for the afternoon of 15 April in connection with the IRDiRC annual conference.

### **Acknowledgments to the host AFM**

The Exec Com was very grateful to IRDiRC funding member AFM for hosting the meeting and for arranging a visit to the labs of Généthon BioProd laboratories (the world's first centre for the production of gene therapy drugs for the treatment of rare diseases). The Chair and the IRDiRC interim secretariat wish to thank AFM for their hospitality.



**Annex – List of participants**

<b><u>Members</u></b>	<b><u>Representative</u></b>
IRDiRC Chair Interim Executive Committee, European Commission, DG Research and Innovation, EU	Ruxandra Draghia-Akli
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research, Canada	Paul Lasko
Genome Canada, Canada	Cindy Bell
European Commission, (DG Health and Consumer Protection), EU	Antoni Monserrat
EURORDIS (Patient Advocacy Group), Europe	Beatrice de Montleau
E-RARE-2 (ERA-net group of funders), Europe	Sophie Koutouzov
French National Research Agency, France	Natalia Martin
French Association against Myopathies, France	Marie-Christine Ouillade/Francoise Rouault
Lysogene, France	Karen Aiach
Federal Ministry of Education and Research, Germany	Ralph Schuster
Shire, Ireland	Philip Vickers
Telethon Foundation, Italy	Lucia Monaco
The Netherlands Organisation for Health Research and Development	Janna de Boer
Carlos III Health Institute, Spain	Rafael d'Andres
Sandford Research, USA	David Pearce
National Human Genome Research Institute NIH, USA	Jeff Schloss
Office of Rare Diseases, NIH, USA	Stephen Groft
<b><u>Scientific Committees</u></b>	
Chair Therapies Sci Com	Josep Torrent-Farnell
Chair Interdisciplinary Sci Com	Hanns Lochmueller
Chair Diagnostics Sci Com	Kym Boycott
<b><u>IRDiRC Secretariat</u></b>	
European Commission, (DG Research and Innovation),EU	Irene Norstedt
European Commission, (DG Research and Innovation), EU	Iiro Eerola

European Commission, (DG Research and Innovation),EU	Anders Colver
<b><u>Invited</u></b>	
SUPPORT IRDiRC project	Segolene Ayme



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