



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 4<sup>th</sup> Executive  
Committee meeting**

15 April 2013



**IRDIRC**

## EXECUTIVE SUMMARY

The Executive Committee (Exec Com) of the International Rare Diseases Research Consortium (IRDiRC) met on 15 April 2013 in Dublin, Ireland. The fourth meeting of the Exec Com was kindly hosted by the European Commission and brought together 33 participants including 23 Exec Com members and the chairs of the 3 Scientific Committees (Sci Com).

The Exec Com discussed and approved the IRDiRC Policy Document.

The Exec Com discussed the next steps with regard to the elaboration of the Roadmap document that aim to guide IRDiRC members on priorities areas for future funding/actions. The Action Plan and Gap Analysis documents - presented for the first time to the Exec Com - should be further elaborated by the scientific Secretariat in collaboration with the Sci Com and Working Groups to be presented at the next Exec Com meeting for discussion.

The Exec com decided not to agree on a common definition of rare diseases but to use the official definition of the different regions.

Moreover, the Exec Com welcomed three new members among them and approved the nomination of 3 new members for the Therapeutics Sci Com and one new member for the Diagnostics Sci Com.

The chairmanship of the Exec Com was officially transferred to Dr Paul Lasko, Scientific director of the Institute of Genetics, CIHR, Canada.

## REPORT

### Welcome of new members

Ruxandra Draghia-Akli and Paul Lasko welcomed the participants and the new members of the Exec Com:

- ▶ Dr Ning Li, Beijing Genomics Institute
- ▶ Ms Diane Goetz, PTC Therapeutics
- ▶ Dr Robert Mashal, NKT Therapeutics

### IRDiRC policy document

The chairs of the Sci Com presented the history of the Policy document. Since the last Exec Com meeting in Barcelona in January 2013, the document was significantly shortened and the policies and guidelines reordered in 2 major sections entitled 'for researchers' and 'for funders' by the Sci Sec and the 3 Sci Com chairs. The Exec Com discussed the policies and guidelines point by point before adopting the document. Editorial revisions included rephrasing, reorganization and deletion. The extensive version of the document will be kept as a reference document.

### Roadmap and Gap Analysis

The purpose and validity of the roadmap and gap analysis documents – drafted by the Sci Sec by extraction of relevant information from the policy document – were discussed. The road map should aim to guide IRDiRC members on priority areas for future funding/actions. The next step is thus to further elaborate the 'Action plan' to include priorities and suggestions, taking into consideration feasibility. The scientific secretariat will work with the Sci Com/Working Groups to present the updated document at the next Exec Com Meeting for further discussion and possible adoption.

### Work plan and communications issues

The Sci Sec presented a work plan with objectives at 12 months for the Exec Com, Sci Com and Sci Sec, emphasizing the importance of adopting key policies documents, finalizing the composition and mandate of the Working Groups, providing the list of the IRDiRC funded project for trend analysis, and extending the consortium. The Exec Com decided not to agree on a common definition of rare disease but to use the official definition of the different regions – USA, Asia and Europe. Moreover, the 2015 IRDiRC conference will be discussed at the next Exec Com meeting.

Regarding the communication strategy, the Sci Com should suggest possible indicators of success to be presented on the IRDiRC website as well as congresses where IRDiRC posters/sessions could be presented /organized. In the near future, an IRDiRC session will be held at the next BIO International Convention in Chicago on April 23, 2013, and a poster on IRDiRC will be presented at the European Society of Human Genetics meeting held in Paris in June.

For communication purpose, a flyer, poster and set of presentation slides about IRDiRC will be available to the Exec Com members on the private section of the website. In addition, a cover page for IRDiRC documents was approved by the Exec Com.

### **Governance issues**

The nomination of 4 new members of the Sci Com was approved by the Exec Com for a 3-year mandate. The new members are:

- ▶ Therapeutics Sci Com:
  - Dr Robert George Schaub (nomination by NKT Therapeutics)
  - Dr Ellen Welch (nomination by PTC Therapeutics)
  - Dr Karin Rademaker nominated to replace Dr Marianne de Visser (nomination by ZonMw)
  
- ▶ Diagnostics Sci Com:
  - Prof Jun Wang (nomination by BGI)

It was also stated that Dr Marcela del Rio (CIEMAT-CIBER) has left the Therapies Sci Com

### **Chairmanship of Executive Committee**

The chairmanship of the Executive Committee was officially transferred from Dr Ruxandra Draghia-Akli (Director, Health Directorate, European Commission) to Dr Paul Lasko (Scientific Director of the Institute of Genetics, CIHR) during the working dinner held at the Burlington Hotel.

### **Acknowledgments to the host**

The Exec Com is very grateful to the European Commission for hosting the meeting. The Chair and the IRDiRC secretariat wish to thank the European Commission representation in Ireland for their generosity and hospitality.

**Annex - List of participants**

<b>Members</b>	<b>Representative</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	Pierre Meulien
BGI, China	Ning Li
European Commission, (DG Health and Consumer Protection), EU	Stefan Schreck and Jaroslaw Waligóra
EURORDIS ( Patient Advocacy Group), Europe	Beatrice de Montleau
E-RARE-2 (ERA-net group of funders), Europe	Daria Julkowska and Iris Fortmann
ANR- French National Research Agency, France	Natalia Martin
AFM- French Association against Myopathies, France	Marie-Christine Ouillade
Federal Ministry of Education and Research, Germany	Ralph Schuster
Shire, Ireland	Philip Vickers
Telethon Foundation, Italy	Lucia Monaco
Carlos III Health Institute, Spain	Rafael De Andres-Medina
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
U.S. Food and Drug Administration, USA	Katherine Needleman
Genetic Alliance, USA	Terry Sharon
National Center for Advancing Translational Sciences, NIH, USA	Christopher Austin
National Human Genome Research Institute, NIH, USA	Jeff Schloss
National Cancer Institute, NIH, USA	Ted Trimble
Office of Rare Diseases, NIH, USA	Stephen Groft
PTC Therapeutics, USA	Diane Goetz
<b>Scientific Committees</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 and Iiro Eerola
<b><u>Invited</u></b>	
SUPPORT-IRDiRC project	Ségolène Aymé and Barbara Cagniard

### Apologies

<b><u>Members</u></b>	<b><u>Representative</u></b>
Lysogene, France	Karen Aiach
Instituto Superiore de Sanita, Italy	Enrico Garaci
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
NORD, USA	Peter Saltonstall
Sanford Research	David Pearce



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