



**INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM**

**Minutes of the 9th Executive
Committee meeting**

7-8 May 2014



IRDIRC

EXECUTIVE SUMMARY

The Executive Committee (Exec Com) of the International Rare Diseases Research Consortium (IRDiRC) met on 7-8 May 2014 in Berlin, Germany. The ninth meeting of the Exec Com brought together 31 participants including Exec Com members and representatives of the three Scientific Committees (Sci Com).

The Scientific Secretariat reported on its activities to support IRDiRC Committees, to monitor and disseminate information on IRDiRC activities for the past few months. Members of the Exec Com agreed that IRDiRC communication strategy should be further discussed and that a R&D trends analysis will be an essential contribution in the field of rare diseases. Communication will be further strengthened through publication on IRDiRC and its activities. It was decided that acknowledgment of IRDiRC in publication will be sufficient and that IRDiRC as a consortium should not be included as an author. In addition, a label “IRDiRC recommended” will be created to highlight tools and standards identified as key resources.

The Chair of the Exec Com summarized his recent activities related to recruitment for IRDiRC and representation of IRDiRC at conferences. In a related discussion, Exec Com established that the strategy to approach potential new members should include a geographic approach, a balance of the different stakeholders and include members able to fund clinical trials.

The Chairs of the Sci Com presented the roadmap of their respective committees. The first part of the roadmap related to the milestones of the Working Groups, i.e., tools, standards and position statements to develop in the next 3 years. The second part addressed the immediate actions to improve the efficiency of IRDiRC activities and actions to implement IRDiRC policies. Finally, the Chairs of the Sci Com expounded several funding priorities to reach IRDiRC goals. Exec Com recognized the importance of developing tools/infrastructures to facilitate research in rare diseases and Sci Com Chairs were asked to prepare proposals for the financing of projects they consider to be important and urgent. Possible mechanisms for international funding of such projects will be discussed by Exec Com in a subsequent meeting.

Members of the Exec Com reached an agreement on the benefit of developing formal associations with not-for-profit association/organization with complementary and related mandates to IRDiRC's, to help coordination of overlapping activities.

It was also decided that:

- ▶ The next IRDiRC conference should be organized in 2016 in a region where IRDiRC is not yet well known.
- ▶ The next face-to-face meeting of the Exec Com will be held in Shenzhen, China, on 6 November 2014.

REPORT

Report on activities by the Scientific Secretariat

The Scientific Secretariat reported on its activities since the last face-to-face Exec Com meeting in September 2013.

Support to Committees and WGs

- ▶ From October 2013 to May 2014, the Scientific Secretariat organized 23 teleconferences and five face-to-face meetings, all Committees and WGs included.
- ▶ Analysis of overlaps and identification of trends to be supported in the future, which were planned in the first 18 months of the SUPPORT-IRDIRC contract, were delayed due to a difficulty to obtain the list of research projects funded by IRDiRC members. However, the Secretariat can now start analyzing the trends.

Dissemination

Dissemination of information is conducted through the public website, private website, and the monthly newsletter.

Recent improvements of the public website consist of the integration of a sitemap, a search engine and a link to the private website. The website is visited in average 1500-2000 times per month and the number of visits increases every month.

IRDIRC private website was launched on October 2013. This website contains information and documents useful to each committee and working group.

Exec Com funding agencies are encouraged to regularly update the Secretariat with:

- ▶ List of research projects funded.
- ▶ Calls for funding.

All members are encouraged to send suggestions on:

- ▶ Publications on IRDiRC, IRDiRC session in conferences, and so on, to be published in the section 'IRDIRC in the news'.
- ▶ Research projects to highlight on the website.
- ▶ Potential interviews of research project leaders or other.
- ▶ Selected reports and guidelines relevant to the field to publish on the website.

Monitor the activities

The webpage showing indicators monitoring progresses towards goal is still a work in progress. It should be ready by this summer.

Collection of research projects funded by IRDiRC members took over a year (not all yet received) and most of them are now registered in Orphanet database to allow an analysis of trends. Limitation on data registration is the lack of work power to register the 3000 or so projects funded by the eight NIH members of the consortium, as there is no Orphanet team in USA.

Comments from Exec Com:

- ▶ The possibility for NIH to send information on the research projects for injection in Orphanet database will be discussed.
- ▶ IRDiRC communication strategy should be further discussed as it is very important. The strategy should be as ambitious as IRDiRC goals. This topic will be an item for a teleconference.
- ▶ R&D trends analysis: it will be an essential contribution as it will be the 1st synthesis of both EU and USA. Purpose is to help decision on funding priorities. The result should be published with the following process: preparation by Scientific Secretariat of the trend analysis, review of this draft by the scientific committees, and preparation of recommendations by Sci Coms.

Report from the Exec Com Chair

The Chair of the Committee summarized his recent activities related to recruitment for IRDiRC and representation of IRDiRC at conferences:

- ▶ 2 private-sector members were recently recruited: Genzyme and Isis Pharmaceuticals.
- ▶ There are ongoing discussion with 3 other private-sector members/ biotechs and a member of the Japanese National Institute of Public Health.
- ▶ The Chair presented IRDiRC in 9 conferences between October 2013 and May 2014 and 2 presentations are scheduled for June 2014.

IRDiRC roadmap

The Chairs of the Sci Com presented the roadmap of their respective committees. The first part of the roadmap related to the milestones of the Working Groups, i.e., tools, standards and position statements to develop in the next 3 years. The second part addressed the immediate actions to improve the efficiency of IRDiRC activities and actions to implement IRDiRC policies. Finally, the Chairs of the Sci Com expounded several funding priorities to reach IRDiRC goals. Exec Com recognized the importance of developing tools/infrastructures to facilitate research in rare diseases and Sci Com Chairs were asked to prepare proposals for the financing of

projects they consider to be important and urgent. Possible mechanisms for international funding of such projects will be discussed by Exec Com in a subsequent meeting.

Presentation by new members

Genzyme

Genzyme, a company funded in 1981, became a Sanofi company in 2011. Genzyme's mission is to discover and deliver transformative therapies for patients with rare and special unmet medical needs. Genzyme focuses on rare diseases (genetic, endocrinology and cardiovascular diseases) and multiple sclerosis, and has eight major marketed products.

French Foundation for rare diseases

The French Foundation for rare diseases is a non-profit structure created in 2012. The Foundation aims to coordinate, federate and fund rare diseases research to accelerate scientific, medical and social innovations in France but also internationally by collaborating with other initiatives. Actions include assessing national landscape and needs in research, accelerating the translation of research into clinical development, developing rare diseases community networks, detecting business development opportunities, and facilitating cross-sector partnerships.

Discussion on the types of IRDiRC membership

IRDiRC has now 39 members spread across 13 countries in four continents, and it is necessary to establish a strategy to approach potential new member to obtain a good representation of the rare diseases research community, keeping in mind that there may be a problem of governance if number of members increases significantly.

Members currently include:

- ▶ Public funding agencies issuing calls for proposals.
- ▶ For-profit companies investing in research.
- ▶ Charities issuing calls for proposals.
- ▶ Non-for-profit Foundation supporting their own research projects.
- ▶ Academic institutions spending a significant budget in research activities.
- ▶ Consortium of academic institutions spending significant budget in research activities.
- ▶ Invited patient advocacy groups.

Members of the Exec Com agreed that:

- ▶ A geographic approach should be continued (South America is an area of interest).

- ▶ A balance of the different stakeholder should be kept.
- ▶ Considering the aims of IRDiRC, members able to fund clinical trials should be included as several members of the consortium are not able to do so.

Composition of the Scientific Committee

New process to nominate members

The procedure to nominate new members of the Scientific Committees, which was proposed at the last meeting of this committee, was approved.

New procedure:

- ▶ Accept nomination of new members for Scientific Committees and Working Groups to have a pool of potential candidates.
- ▶ Call for application to IRDiRC members when renewal of Sci Com members.

Nomination for the TSC

The nomination of Dr Shuling Guo (Isis Pharmaceuticals) as a new member of the Therapies Sci Com was approved by the Exec Com for a 3-year mandate.

Membership fee

The Scientific Secretariat is in discussion with Inserm to set up an account independent of SUPPORT-IRDiRC contract for the purpose of the membership fee. If negotiation with Inserm failed, there is the possibility to host the account at the University of McGill, Canada.

In the meantime, each member will be contacted by the Secretariat to inquire their willingness to contribute through the membership fee or by in-kind participation, in order to prepare a financial plan.

Publication about IRDiRC

IRDiRC communication strategy includes dissemination of information on IRDiRC and its activity through publication in peer-reviewed journals. Publications would generally consist of perspective papers such as IRDiRC aims and policies, IRDiRC roadmap, and papers on Sci Coms and WGs outcomes.

The question was raised to determine if IRDiRC as a consortium should be listed as an author. Although in principle this should be possible, to avoid any potential conflict of interest, members of the Exec Com decided that IRDiRC as a consortium should not be included as an author and that an acknowledgement of IRDiRC in the publication will be sufficient.

Proposal for the creation of the label “IRDiRC recommended”

The possibility to create a label to highlight tools and standards generated through IRDiRC activities but also tools and standards not generated by but identified by IRDiRC as key resources was discussed. To avoid any conflict of interest, Exec Com agreed on the terminology ‘IRDiRC recommended’.

Process for adoption was defined:

- ▶ Scientific Committee to produce 1-2 pages of rationale, including list of other most important tools or standards, and contentious issues.
- ▶ Consultation of the two other Scientific Committees.
- ▶ Submission to the Executive Committee for approval.

Any recommended tools/standards/guidelines will be highlighted on the IRDiRC website. A logo will be designed and a regulatory disclaimer posted. Moreover it was requested that visitors should be able to submit comments on the website for critical review of these items.

Mechanism for formal association

Chair of the Executive Committee received formal request for association by ICORD. Other formal association would be beneficial, such as GA, IHEC, etc.

Members reached an agreement that such formal association would be beneficial to IRDiRC to help among others coordination of overlapping activities. As for new membership, request for formal association should be screened by the Chair of the committee, with the possibility to consult the committee if uncertainty.

A few criteria for acceptance were decided:

- ▶ Association/organization should be not-for-profit
- ▶ National or umbrella groups of patients will be accepted.

- ▶ Association/organization should have complementary and related mandates to IRDiRC's.

Update on the IRDiRC conference in Shenzhen, November 2014

The conference to be held from 7-9 November 2014 in Shenzhen in China will include:

- ▶ Educational track on 7-8 November.
- ▶ Training course on genetics on 9 November.
- ▶ Matchmaking workshop to facilitate collaboration between clinicians and scientists.

The website of the conference was presented (<http://irdirc-shenzhenconference2014.org/>).

The most convenient date to organize the Executive Committee meeting in Shenzhen is 6 November 2014, the day before the IRDiRC conference.

Next IRDiRC conference

The initial plan was to organize an IRDiRC conference every 2 years, i.e., the 2nd conference should have been scheduled in 2015, following the Dublin conference in 2013. However, BGI generously proposed to host a conference in Shenzhen in November 2014.

The 3rd conference should be organized in 2016. Two strategies are possible:

- ▶ Organizing the conference in Spring 2016, jointly with E-rare and RE(ACT) International Congress on Research of Rare Diseases in Barcelona.
- ▶ Organizing a conference in fall 2016 in a geographical area where IRDiRC is not yet well known, such as South America or Middle East. However, it would be necessary to find sponsorships, which would be easier in Middle East, but would require time.

Any proposal from a member to sponsor and contribute of funding an IRDiRC conference in 2016 would be considered.

Acknowledgments to the host

The Exec Com is very grateful to Ralph Schuster for hosting the meeting. The Chair and the IRDiRC Secretariat wish to thank Ralph Schuster and the Federal Ministry of Education and Research for their generosity and hospitality.

Annex - List of participants

Members	Representative
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
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	Irene Nordsted
EURORDIS (Patient Advocacy Group), Europe	Béatrice de Montleau
AFM- French Association against Myopathies, France	Marie-Christine Ouillade and Françoise Rouault
ANR- French National Research Agency, France	Natalia Martin
FFRD – French Foundation for rare diseases, France	Roseline Favresse
Federal Ministry of Education and Research, Germany	Ralph Schuster
Children’s New Hospitals Management Group, Georgia	Oleg Kvlividize
Shire, Ireland	Emmanuel Chantelot
Istituto Superiore de Sanita, Italy	Domenica Taruscio
Telethon Foundation, Italy	Lucia Monaco
The Netherlands Organisation for Health Research and Development, The Netherlands	Sonja van Weely
Carlos III Health Institute, Spain	Pedro Cortegoso Fernandez and Antonio Andreu Periz
Food and Drug Administration, USA	Debra Lewis
Genetic Alliance, USA	Sharon Terry
Genzyme, USA	Carlo Incerti
National Human Genome Research Institute, NIH, USA	Jeff Schloss
Office of Rare Diseases, USA	Pamela Mc Innes and Stephen Groft
PTC Therapeutics, USA	Diane Goetz
Sanford Research, USA	David Pearce
Scientific Committees	
Chair Diagnostic Sci Com	Kym Boycott
Chair Interdisciplinary Sci Com	Hanns Lochmüller

Therapies Sci Com	Yann Le Cam and Josep Torrent i Farnell
IRDiRC Scientific Secretariat	
SUPPORT-IRDiRC project	Ségolène Aymé and Barbara Cagniard

Apologies

<u>Members</u>	<u>Representative</u>
BGI, China	Ning Li
Chinese Rare Disease Research Consortium, China	Qing Wang
Academy of Finland, Finland	Heikki Vilen
Lysogene, France	Karen Aiach
Korea National Institute of Health, Korea	Hyun-Young Park
Prosensa, The Netherlands	Luc Dochez
National Institute for Health Research, United Kingdom	Willem Ouwehand
Isis Pharmaceuticals, USA	Brett Monia
National Cancer Institute, NIH, USA	Edward Trimble
National Eye Institute, NIH, USA	Santa Tumminia
National Center for Advancing Translational Sciences, NIH, USA	Christopher Austin
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
NORD, USA	Peter Saltonstall



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