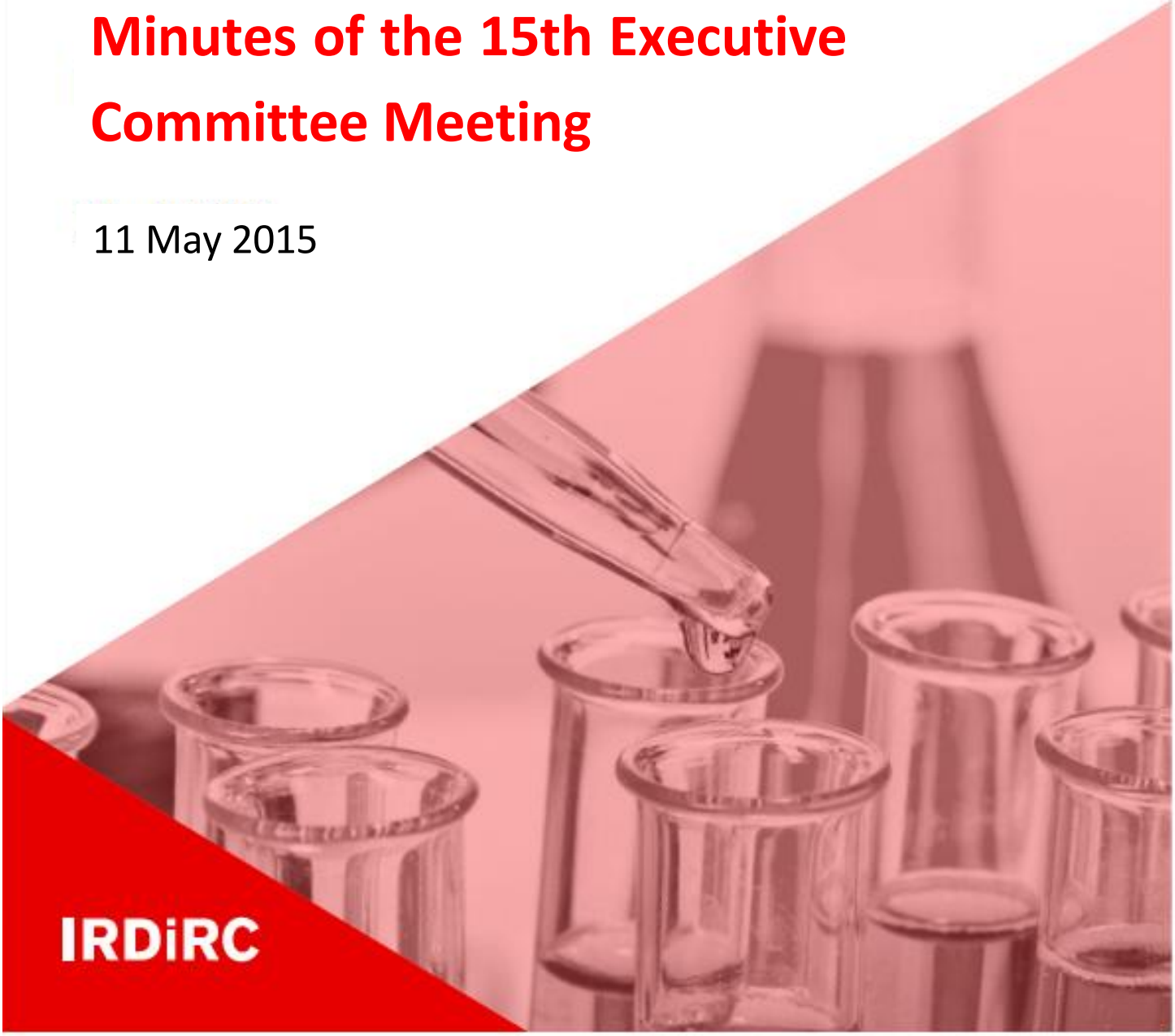
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Minutes of the 15th Executive Committee Meeting

11 May 2015



IRDiRC

EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 11 May 2015 by teleconference. The fifteenth meeting of the Exec Comm brought together 27 participants.

Members of the Exec Comm were informed of the progress made in preparation of draft background paper to "Patient Relevant/Reported Outcome Measures" and updated on the progress made on other Task Forces to date. A Task Force on Data Mining/Repurposing will be constituted based on nominees who will be contacted and agree to participate.

A number of specific questions were raised which the Scientific Secretariat (Sci Sec) will try to address in the next State of Play Report.

The Exec Comm agreed that tools/platforms, even if fairly new, that are useful in rare diseases and advancing IRDiRC goals, with an international connectivity and audience and a three year budget, should be accepted for review of "IRDiRC Recommended". The guideline on the website will be updated to include what it means (e.g. the lack of recommendation does not mean a tool is inferior to another). FDA will also submit a disclaimer text to be included on the website.

The IRDiRC website has been redesigned to better highlight its current activities and initiatives. A commenting system and a page to encourage involvement in IRDiRC will be added. The Sci Sec welcomes feedback to further improve it. The guideline to the use of IRDiRC logo was discussed, and a statement on its use will also be stated on the IRDiRC website.

IRDiRC will be a conference partner to the RE(ACT) Congress 2016. Additionally, the Sci Sec submitted a proposal of IRDiRC session to the AAAS 2016 Annual Meeting, and will submit another to the ESOF 2016 Manchester.

The Spring 2016 Exec Comm meeting will take place in the UK, on the premise that the host is able to secure the resources to fund the meeting and resolve an administrative issue.

The Exec Comm was asked to consider nominating active individuals to the Scientific Committees to replace members who will soon come to the end of their mandate.

Diane Goetz is retiring from PTC Therapeutics and a new representative to the Exec Comm will be nominated.

REPORT

Feedback: Draft Preparatory Document "Patient Relevant/Reported Outcome Measures"

The first draft of the background paper to "Patient Relevant/Reported Outcome Measures" (PROM) was circulated for information, to keep the Exec Comm informed on the relevant progress to date. Pre-workshop work methodology and its subsequent update on Task Force member nominations and appointments can be found in the reports of the 13th and 14th Exec Comm meeting respectively.

A member of the Exec Comm currently involved in two Innovative Medicines Initiative programmes, i.e. PRO-active and U-BIOPRED, suggested extending an invitation to the coordinators of these projects to participate in the PROM Task Force. Another member is the coordinator for patient powered research networks on PRO under a Robert Wood Johnson Foundation project and was invited to participate in the core group of PROM Task Force.

Incidental findings will not be addressed by the PROM Task Force. Nonetheless, a recent position statement by Canadian College of Medical Geneticists is of interest and fit IRDiRC general activities that it will be published on the website in the "recommendation" section.

The Chair of the Exec Comm asked that all draft background paper for workshops be circulated in real time for feedback. The Exec Comm will also be informed in real time of workshop dates and locations when they have been decided.

State of Play Report: Specific Questions to be Addressed

The next State of Play Report was discussed at the 14th Exec Comm meeting and one of the suggestions to improve the report involves the Exec Comm and Sci Comms generating specific questions they are searching answers to and that are forward looking, which the Sci Sec could try to address. The questions should be general in nature, rather than being disease-specific.

A number of questions were put forward:

- What are the genetic diseases with highest prevalence but currently without diagnostic tool available?
- What are the disease areas/approaches that are currently neglected/not researched?
- What are the statistics for diseases/therapeutic areas that companies are looking into the most?

The Sci Sec will incorporate answers to these questions to their best ability in the State of Play 2015. Moreover, Genetic Alliance (GA) has a beta algorithm for assessing diseases across four domains for "readiness" for drug development, and is ready to assist the Sci Sec in running relevant queries. This algorithm is currently available only in-house to GA but its second version is planned for web release. The Sci Sec will be in touch with GA to see how the algorithm can generate data for the next State of Play Report.

The Sci Sec will continue to accept and address additional specific questions from the members for the next State of Play within reasonable timeframe to its publication.

"IRDiRC Recommended"

Two applications have been received to date, and one of them was circulated to the Exec Comm to aid the discussion on assessment difficulties (e.g. of recently developed tools and single-institution tools).

In rare disease research where things are developing quickly, it is difficult to award the label solely to well-established tools/platforms, not to mention these will also continue to undergo changes and updates. The Exec Comm agreed that tools/platforms, even if fairly new, that are useful in rare diseases and advancing IRDiRC goals, with an international connectivity and audience, and a three-year budget, should be accepted for review.

The information page on IRDiRC website should be updated to include what it means (e.g. the lack of recommendation does not mean a tool is inferior to another) and to include the international criteria.

The review process should commence on the applications received. The evaluation should be made transparent, with a score card listing pros and cons and the percentage of criterion met to be provided as feedback to the applicants.

FDA Disclaimer

FDA is not able to neither vote nor endorse "IRDiRC Recommended", and a disclaimer text is currently in development to be included on the IRDiRC website, possibly in policy document too. The FDA is also due to provide a disclaimer text for the Therapies Sci Comm recommendations document.

Any other member of the Exec Comm who may have similar reservation should inform the Chair and the Sci Sec so appropriate discussion and action could take place.

Data Mining/Repurposing Task Force

The Exec Comm decided that a group of nominees will be contacted and a Task Force will be constituted of those who agree to participate.

Other Task Force Nominations/Acceptances

The Exec Comm was updated on the other Task Forces:

- PROM: two nominees have accepted their nominations, one will be having a further discussion to understand the scope prior to accepting, and Sharon Terry accepted the nomination to be part of the core group
- Small Population Clinical Trials (SPCT): all nominees have accepted their nominations although the EMA has yet to name the specific liaison to participate in the Task Force
- Matchmaker Exchange (MME): all nominees have accepted their nominations and a MME paper in preparation will be the basis of the workshop background paper
- Machine Readable Consent (MRC): the liaison with the Global Alliance (GA4GH) Task Team on MRC is in place

The Sci Sec will continue to compile names of potential participants to Task Forces and/or workshops received until it hands the list over to the core groups for their selection decisions. Members, particularly from the industry, are strongly encouraged to send in their nominations.

IRDiRC Website

The IRDiRC website (http://www.irdirc.org/) has been redesigned and this new version is now live. Most changes occurred on the front page to be more dynamic and to better highlight IRDiRC's current activities and initiatives. The Exec Comm congratulated the Sci Sec for the work well done.

The Chair of the Exec Comm would like to see the site to incorporate ability for commenting on IRDiRC's activities. This is a feature which the Sci Sec intended to implement but currently on hold until the contents are completely updated. Another feature planned is to create a page that encourages others to be involved in IRDiRC and explains how they could participate.

Members are encouraged to browse the website and let the Sci Sec have any additional feedback to further improve it.

IRDiRC Logo Guideline

The standard IRDiRC logo is permitted for use by:

- Relevant projects working on IRDiRC goals, funded by IRDiRC members
- Members giving oral or poster presentations in meetings and conferences
- Industry-based IRDiRC members

A statement re practices on the use of IRDiRC logo will be stated on the IRDiRC website; any other uses (e.g. by projects not funded by IRDiRC but working collaboratively with IRDiRC) is allowed if expressed permission to use is given.

Future Meetings/IRDiRC Sessions

AAAS 2016 Annual Meeting, 11-15 February 2016, Washington DC, USA

The Sci Sec has submitted a proposal for a 3-speaker session, following suggestion from the European Commission. The decision will be announced in June 2015.

RE(ACT) Congress 2016, 9-12 March 2016, Barcelona, Spain

E-Rare is co-organising RE(ACT) Congress 2016 with BLACKSWAN Foundation. They proposed using IRDiRC's logo thus seen as conference partner and in return, IRDiRC may add a speaker to each of their seven sessions (i.e. drug repositioning and personalised medicine, mapping diseases and genome instability, bringing treatments to the clinic, pathophysiology, next generation sequencing and undiagnosed rare diseases, neurological diseases, and patients and research). IRDiRC was not asked to financially support these speakers. IRDiRC also will not engage in a formal Memorandum of Understanding with BLACKSWAN Foundation, which accepted this condition. The Exec Comm agreed to participate in this initiative.

ESOF, 22-27 July 2016, Manchester, UK

EuroScience Open Forum is a biennial, interdisciplinary meeting with wide range of audience, aiming at, among others, promoting dialogue of science and technology in society and public policy. One of the sessions of this meeting is "Science for policy and policy for science" which may fit IRDiRC's activity in making a mark in policymaking in rare diseases. Moreover, this also falls within the European Commission recommendation that IRDiRC be active in community in which IRDiRC is not well known. The Exec Comm consented to the Sci Sec submitting a proposal for a session at the ESOF.

Exec Comm Face-to-Face Spring 2016

NIHR proposed to host the Spring 2016 face-to-face meeting in a dedicated conference centre but needs to secure the resources to pay for the hosting costs and resolve an administrative issue. An alternative venue may be needed.

Any Other Business

Membership of Scientific Committees

A large number of Scientific Committee members will come to the end of their mandate next year. Members who are active will be nominated for an extension of their terms, and the Exec Comm is also asked to consider nominating active individuals who can take on leadership roles on the Scientific Committees.

Diane Goetz's Retirement

Diane Goetz from PTC Therapeutics is retiring at the end of May, and a new representative to the Exec Comm will be nominated in due course. The Exec Comm and the Sci Sec would like to thank Diane for her contribution and wish her a happy retirement.

Next Exec Comm Teleconference

A teleconference of the Exec Comm will be organised in early August. The Sci Sec will send a Doodle to poll for the most suitable date.

Annex - List of participants

Members	**Representative**
Western Australian Department of Health, Australia	Hugh Dawkins
Canadian Institutes of Health Research, Canada	Paul Lasko
Genome Canada	Pierre Meulien
WuXi AppTec Co. Ltd., China	Mao Mao
European Commission, DG Research and Innovation, EU	Anders Colver
Academy of Finland, Finland	Heikki Vilen
AFM- French Association against Myopathies, France	Marie-Christine Ouillade
Lysogene, France	Karen Aiach
Children's New Hospitals Management Group, Georgia	Oleg Kvlividize
Federal Ministry of Education and Research, Germany	Ralph Schuster
Shire Pharmaceuticals, USA	Albert Seymour
Chiesi Farmaceutici S.p.A, Italy	Andrea Chiesi
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	James Reese
National Human Genome Research Institute (NHGRI), NIH, USA	Jeffery Schloss
PTC Therapeutics, USA	Diane Goetz
Sanford Research, USA	David Pearce

Invited Patient Advocacy Groups	
EURORDIS, Europe	Béatrice de Montleau
Genetic Alliance, USA	Sharon Terry
National Organization for Rare Diseases, NORD, USA	Peter Saltonstall

Scientific Committees	
Diagnostics	Kym Boycott
Interdisciplinary	Hanns Lochmüller

IRDIRC Scientific Secretariat	
SUPPORT-IRDIRC Project	Ségolène Aymé, Lilian Lau, Antonia Mills

Apologies

Members	Representative
BGI, China	Ning Li
Chinese Rare Diseases Research Consortium, China	Qing Wang
European Organisation for Treatment & Research on Cancer, EORTC	Denis Lacombe
E-RARE-2 Consortium, EU	Daria Julkowska
Agence National de la Recherche, ANR, France	Natalia Martin
Fondation Maladies Rares, France	Nicolas Lévy
Istituto Superiore de Sanita, Italy	Fabriozio Oleari
Saudi Human Genome Project, Kingdom of Saudi Arabia	Sultan Turki Al Sedairy
Prosensa, The Netherlands	Scott Clarke
Korea National Institute of Health, South Korea	Hyun-Young Park
National Institute for Health Research, UK	Willem Ouwehand
Genzyme, USA	Carlo Incerti
Isis Pharmaceuticals, USA	Brett Monia
National Cancer Institute, NCI/NIH, USA	Edward Trimble
National Center for Advancing Translational Sciences, NCATS/NIH, USA	Christopher Austin
National Eye Institute, NEI/NIH, USA	Santa Tumminia
National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIAMS/NIH, USA	Stephen Katz
National Institute of Child Health and Human Development, NICHD/ NIH, USA	Melissa Parisi
National Institute of Neurological Disorders and Stroke, NINDS/NIH, USA	Danilo Tagle
NKT Therapeutics, USA	Robert Mashal
Office of Rare Diseases Research, ORDR/NIH, USA	Pamela McInnes

Scientific Committee	
Therapies	Yann Le Cam

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