

Meeting report series

Report of the 1st Interdisciplinary Scientific Committee Meeting

Paris, Agence Nationale de la Recherche
May 30th and 31st, 2012

Organization

Hosted by: Agence Nationale de la Recherche, 212, rue de Bercy, Paris 75012

Participants

Dr Denise Avard, Montreal, Canada (representing Prof BM Knoppers)

Dr Angel Carracedo, Santiago de Compostela, Spain

Prof Jamel Chelly, Paris, France (vice-chair, chair-elect)

Prof Jack Goldblatt, Perth, Australia

Mr Alastair Kent, London, UK

Dr Jeffrey Krischer, Tampa, USA

Prof Hanns Lochmüller, Newcastle, UK (chair)

Dr Natalia Martin, ANR, Paris, France (host)

Mrs Samantha Parker, Paris, France

Prof Rumen Stefanov, Plovdiv, Bulgaria

Dr Domenica Taruscio, Rome, Italy

EXECUTIVE SUMMARY

The IRDiRC Interdisciplinary Scientific Committee (ISC) was selected by the Executive Committee (EC) and met for the first time on May 30/31 in Paris, hosted by the Agence Nationale de la Recherche. As suggested by the Executive Committee, a “policies and guidelines” document (draft April 2012) was extensively reviewed, the remit and composition of working groups were defined, and a roadmap including deliverables for the ISC was agreed. Specifically, Prof Hanns Lochmüller and Prof Jamel Chelly will serve as chair and chair-elect (vice-chair) respectively for a 1 year period. The ISC concluded that the “policies and guidelines” document requires extensive revision, and is happy to lead on the revision process for several sections, and assist the 2 other Scientific Committees (Diagnostics and Therapies) in revising the remaining sections. The ISC suggests that the chairs of the 3 Committees (once established) will discuss and agree on a process and workflow for the revisions to provide the EC with a consolidated document. The ISC requests clarification whether all EC members (funders) are required to agree to the document and implement the policies in their funding rounds. The ISC believes that working groups with membership from IRDiRC-funded projects may be in an excellent position to identify important gaps and opportunities towards the IRDiRC goals. The ISC requests further clarification on composition, remit and funding of the working groups by the EC. The ISC suggests having close interactions with the following working groups once established: registries and natural history, biobanks, ethics and governance, bioinformatics and data sharing, and possibly biomarkers. The ISC identified gaps in IRDiRC policies and activities which should be implemented in order to facilitate achieving the IRDiRC 2020 goals, specifically ethics and governance, implementation with healthcare professionals, patients and wider public, and sustainability of resources and infrastructures. The ISC suggests to share information and to streamline activities with the other 2 Scientific Committees, requests clarification on the timelines and role of the IRDiRC Secretariat once established, and recommends – as a priority – publishing the names and core data of all IRDiRC projects on a dedicated, publicly accessible website. The ISC is ready to help with setting up working groups and to contribute to the programming of the spring 2013 IRDiRC meeting once these actions have been approved by the EC in their September meeting. Involvement of the ISC and its members in developing and reviewing policies, in establishing and supporting working groups, and further activities may have resource implications that need to be considered by the EC.

REPORT

May 30th 2012 (9.30-18.30)

1. Introduction

The host thanked members for their participation in the Interdisciplinary Scientific Committee (ISC) meeting. The message from the Chair of the IRDiRC Interim Executive Committee was projected, and a presentation was given by the host, where the following points were highlighted:

- ▶ Objectives of IRDiRC and IRDiRC Governance structure /modus operandi
- ▶ Introduction of mandate, expectations and deliverables of the Interdisciplinary Scientific Committee, including an introduction to the role, responsibility and term of the chair

Some questions were raised during and after the presentation. The host sent an email to the EC representative. A reply was received during discussion on the second day (see appendix 1).

2. Roundtable, vote and election of the Chair of the Scientific Committee

The ISC members introduced themselves and the role and responsibilities of the chair were discussed. It was agreed to appoint a chair (Prof. Lochmüller) and a chair-elect/vice-chair (Prof. Chelly) for the duration of one year. The chair and chair-elect can replace each other in respect of communicating the views of the ISC. After the first year, the chair-elect assumes the position as chair, and a new vice-chair is elected by the committee. The ISC will review the process annually and may adopt changes where necessary. The chair will represent the ISC at the EC meeting in September. The ISC members asked the host to remind them of the type of IRDiRC projects that will potentially (under negotiation for funding) be funded through the FP7 2012 call for proposals. The host presented this information, which was completed with the input of the participants. Specifically, several projects are currently under negotiation with the European Commission, that are labeled as IRDiRC projects and highly relevant to the work of the Scientific Committees: RD-Connect (Newcastle) aims to harmonize biobanks, databases and clinical bioinformatics for rare diseases (www.rd-connect.eu), Eurenomics (Heidelberg) and Neuromics (Tübingen) will apply –omics approaches to rare kidney and rare neuromuscular/neurodegenerative disorders, and SUPPORTIRDiRC (Paris) will provide secretariat support for IRDiRC. Additional projects labeled as IRDiRC under the same Health call include “natural history” and “development of orphan drugs”. Moreover, Rare-bestpractices (Rome) will include a work package on IRDiRC as requested by the European Commission during the negotiation. The negotiation deadline is the 25th of June, and – provided successful conclusion of the negotiations – these projects are expected to start before the end of the year. The IRDiRC projects will be expected, by the EC, to send delegates to the working groups and to assist the Scientific Committees. Other projects funded from 2010 and national projects (e.g. 2 IRDiRC-labeled projects already approved in Spain) will also be asked to support the committees. The ISC has little information about these projects, particularly with regard to US NIH-funded projects. *The ISC agreed that it is of high importance that more information is provided, to the ISC, about these projects (name of PI, summary, key deliverables). Furthermore this*

information could also be made available on a dedicated, publicly available website (it was pointed out that this information is not available or incomplete on the Orphanet website). The ISC would also appreciate information on the role of the secretariat, provided by SUPPORT-IRDiRC, especially in assisting the work of the scientific committees and the working groups.

3. Brainstorming of ideas to define the borders of the Interdisciplinary Scientific Committee regarding the other Scientific Committees: Diagnostics, Therapies

After a brief discussion concerning the working groups, the ISC members highlighted the difficulty in identifying the issues that should be considered by this particular SC with respect to the other SCs: Diagnostics or Therapies. The ISC understands that the Diagnostics Committee has already met, but minutes were not yet available, and the Therapeutics Committee is due to meet in July. The chair suggested a brainstorming session to help identify the main issues pertinent to this Committee compared to the other two. The chair also took note of a position paper on registries by EURORDIS and NORD which was provided to the Committee (appendix 2). Important issues raised during the brainstorming session included:

- ▶ Implementation issues should also be considered in the IRDiRC management bodies: regulatory issues, potential to facilitate implementation through support (or reimbursement) procedures by social security/health delivery systems (potential difficulties related to the diversity of the systems), socio-economic issues.
- ▶ Medical professionals need to be better involved, informed and educated about IRDiRC research, especially relating to counseling on informed consent and the provision of genetic results from Next Generation Sequencing (NGS).
- ▶ The recognition of the importance, and the acceptance of, IRDiRC research must be ensured through improved communication and interaction with the wider public, a website was not considered sufficient.
- ▶ Public private partnerships and involvement of industry in the development of Orphan Drugs is a priority in IRDiRC funding. If the company successfully brings an OD to the market as a direct result of this investment we would like to reflect on how and if there should be a mechanism for refunding the community for the public sector investment.
- ▶ A policy needs to be developed that puts the patients at the center of drug development when developing ODs. Patients should be represented and play an active role in all IRDiRC activities.
- ▶ IRDiRC recommends that if a patient group participates in a research project a representative of the patient group could be invited to participate in the working group.
- ▶ In order to avoid duplication of efforts it is necessary to make an inventory of available means and ongoing national projects which can link to the IRDiRC objectives (e.g. Fondation Maladies Rares and RADICO projects in France). It would be useful that responsible/representatives of these projects are also involved in the discussions or working groups. The ISC suggests that this issue should be discussed at the EC level.
- ▶ The ISC considered the importance of harmonizing international resources (such as registries, biobanks, patient resources) to facilitate achieving the diagnostic and therapeutic goals.

In summary, the ISC identified gaps in IRDiRC policies and activities which should be addressed in order to facilitate achieving the IRDiRC goals by 2020, specifically ethics and governance, implementation with healthcare professionals, patients and wider public, sustainability and harmonization of resources and infrastructures.

4. Review of the Policies & Guidelines document (first draft April 2012)

General comment and question:

The policy document was reviewed in depth (full afternoon session), although some sections of the document may fall under the remit/lead of the Diagnostics and/or Therapies Committees. Specific suggestions for modification of the policies and guidelines are found in appendix 3. The ISC would like to learn more about the workflow for the completion of this document, and confirmation on which policies they should focus, in order to avoid duplication of the work with other SCs. The ISC discussed the exact purpose of the policy document, in particular whether all EC members (funders) are required to agree and implement the policies in their funding rounds.

The ISC made the following general findings:

There are chapters/issues missing in the policy document, specifically a chapter on bioethics policies (mentioned in several chapters, but incoherent, should form a separate chapter) and a chapter on policies addressing pathophysiological studies (could potentially be included in “D.2. Models and resources for future research”). The content and style of the document is variable, and sometimes contradictory, requiring major revisions. The current draft of the document appears to be the product of different experts, but there is no indication who generated the draft (no document history). *The ISC suggests including a document history for any further work on the document. The ISC is happy to lead on the revision process for several sections, and assist the 2 other Scientific Committees (diagnostic and therapeutic) in revising the remaining sections. The ISC suggests that the chairs of the 3 Committees (once established) will discuss and agree on a process and work-flow for the revisions to provide the Executive Committee with a consolidated document. Involvement of ISC members in developing and reviewing policies may have resource implications. In addition to administrative support (through SUPPORT-IRDiRC), funding needs to be made available for the committee’s work itself. The ISC requests clarification whether all EC members (funders) are required to agree to the document and implement the policies in their funding rounds. This should be made explicit.*

May 31st 2012 (8.30-12.00)

5. Identification of working groups that the ISC would like to follow/work with.

Working groups will consist of members of IRDiRC funded research projects. Clear guidance to the number of working groups, the (maximum/minimum) number of members for each working group, the number of in-person or virtual meetings per year, etc has not been defined. More importantly, the remit and financing of the working groups needs to be defined by the EC. For example, participation by patient group representatives will need explicit funding for them to be able to participate if they are not funded through project budgets. Other external experts, including members of the 3 science committees, may also need to have their costs covered to enable participation in the working groups where required. There was particular concern about the representation of US scientists in working groups, as current NIH funding streams may not provide appropriate and sufficient mechanisms and means to fund participation of US scientists. The ISC believes that working groups may be in an excellent position to identify important gaps and opportunities towards the IRDiRC goals. The ISC suggests that working groups share their meeting results (e.g. minutes) with the ISC. The ISC identified the following working groups that the ISC would be keen to collaborate with and made a start in identifying potentially relevant topics.

Registries/Natural History

(large overlap between registries and natural history, but could also form 2 WG if necessary).

- ▶ The list of policy areas from the IRDiRC paper will form the key topics to be considered by this working group
- ▶ Consideration of the Eurordis/NORD position paper
- ▶ Phenotype delineation (disease trajectory)
- ▶ Clinical management
- ▶ Socio-economic
- ▶ Harmonization

Biobanks

- ▶ Access to samples and data
- ▶ International transfer of samples
- ▶ Cataloguing of samples
- ▶ Harmonization of consenting issues
- ▶ Networking

Ethics and Governance

- ▶ Return of research results, benefit sharing
- ▶ Pediatric issues and transition through life stages

- ▶ Harmonization of consenting issues
- ▶ Data protection (related to new EU regulation)
- ▶ Patient representation (appropriate ways to have patient representation in research)
- ▶ Intra-familial communication
- ▶ Access to samples and data

Datasharing/Bioinformatics

(this WG is expected to be closely linked to the other 2 SC as well)

- ▶ Harmonization
- ▶ Core data sets
- ▶ Data release
- ▶ Data access
- ▶ Technical aspects

Biomarkers

Should be under the responsibility of the Therapies SC. Could be taken by the ISC if necessary (strong relationship with biobanks).

In summary, the ISC suggests establishing close relationships with working groups on registries and natural history, biobanks, ethics and governance, bioinformatics and data sharing and possibly biomarkers. Cross-talk with the other scientific committees will be required (e.g. exchange of minutes, teleconference of the committee chairs). The ISC is ready to help with setting up working groups once these actions have been approved by the EC in their September meeting. Involvement of ISC members in setting up and running working groups may have resource implications that need to be considered. The ISC requests further clarification on composition, remit and funding of the working groups by the EC.

6. Preparation of a roadmap: Working methodology, expected deliverables and Timelines

Main deliverables for the ISC

- ▶ The minutes of the first ISC meeting completed and submitted to the EC: June 14th 2012
- ▶ Final list of working groups. To be provided after discussion with the other Scientific Committees and final approval of the Executive Committee (end September 2012)
- ▶ Revised “policies & guidelines” document. As mentioned in the general comments of section 5, the ISC would like to have instructions on a formal way to work on this document: what is the exact workflow for the completion of the document and who takes the responsibility for the document. The ISC found it important to coordinate this task with the other two scientific committees in order to avoid duplication of efforts (i.e. more than one SC working at the same time on the same policies).
- ▶ Programme for the Interdisciplinary session(s) for the first IRDiRC Conference. The ISC would like to have more details on the scope, content and targeted participants for this conference. This

programme could be achieved after the next Executive Meeting and the next ISC meeting (expected in October-November 2012)

- ▶ Identify and prioritize gaps in interdisciplinary aspects. Discussions concerning the gaps in interdisciplinary aspects of RD research were launched during this meeting. A first draft could be achieved after the next ISC meeting (expected in October-November 2012), once a better definition of the ISC's responsibilities compared to the other SCs is achieved.

Main milestones

- ▶ Meetings, or exchanges by other means (access to minutes of the other SC meetings, email exchanges, etc), with the other Scientific Committees before September 2012.
- ▶ Executive Meeting in September 2012.
- ▶ Next ISC meeting (expected in October-November 2012). The date will be fixed by a Doodle poll.

Working methodology

- ▶ The ISC considered it would be important to learn more about the exact role and responsibilities of the IRDiRC Secretariat (Support action) in order to better define their working methodology.
- ▶ Scientific Committee meetings. Taking into consideration the potential overlap of issues that should be considered by the ISC with the other two SC, it was suggested that some meetings are held jointly with the Therapies and Diagnostics committees to facilitate interaction between members of different committees on issues of mutual interest.
- ▶ Access to information (abstracts, deliverables and involved researchers) concerning the IRDiRC funded projects. The ISC found it important to have access to this information as a basis for the establishment of the working groups, identification of gaps, etc. This task may be included in the Communication work package of the IRDiRC Secretariat (Support action).
- ▶ The ISC found it important to establish a certain level of interaction with other initiatives (at national and international levels) with the same goals as IRDiRC regarding the coordination, and avoidance of duplication, of efforts in the field of RD research.
- ▶ The exchanges with the working groups could be done by different means: attendance of an ISC representative at the working groups' meetings, make the working group's chairs attend the ISC meetings, and/or exchange of information (minutes, *ad-hoc* email exchanges, etc).
- ▶ The issue of an "Inventory" of existing means and ongoing projects not supported by IRDiRC committed members (funding organizations) but related to its activities.