

## Meeting report series

### Report of the 4th Interdisciplinary Scientific Committee (ISC) Meeting

McGill Faculty Club Centre and Conference  
17<sup>th</sup> and 18<sup>th</sup> October 2013

#### Participants

Prof Hanns Lochmüller, Newcastle, UK (Chair)  
Prof Jamel Chelly, Paris, France (co-chair)  
Prof Bartha Maria Knoppers, Montreal, Canada  
Prof Jack Goldblatt, Perth, Australia  
Mrs Samantha Parker, Paris, France  
Prof Rumen Stefanov, Plovdiv, Bulgaria  
Dr Domenica Taruscio, Roma, Italy  
Mr Alastair Kent, London, UK  
Dr Jeffrey Krischer, Tampa, USA  
Ms Roseline Favresse, IRDiRC Scientific Secretariat, France

#### Apologies

Dr Angel Carracedo, Santiago de Compostela, Spain  
Dr Petra Kaufmann, Bethesda, USA

#### Agenda

##### Day 1 - 17 October (9:00-17:00)

- 09:00 09:15 - Introduction from ISC Chair and co-Chair  
09:15 09:45 - Feedback from the latest Executive Committee Meeting (Miami, Sept. 2013)  
09:45 10:45 - Governance and organizational issues (roundtable, discussions)
- ▶ Executive Committee expectations for the ISC
  - ▶ WGs role and composition: clarification, chairing
  - ▶ Interactions with the Scientific Secretariat
  - ▶ Interactions with the other 2 Science Committees
  - ▶ ISC composition: new member P. Kaufmann; chairing
- 10:45 11:00 - *Coffee break*  
11:00 13:00 - Round-table on the Working Groups related to the ISC
- ▶ Feedback from ISC members who took part to the first WG conference calls
  - ▶ Review of WG minutes and summary papers

13:00 14:00 - Lunch (*Old Montreal Room*)

14:00 15:30 - Gap analysis and recommended actions: data sharing

15:30 17:00 - Gap analysis and recommended actions: governance and ethics

19:00 *Dinner*

**Day 2 - 18 October (9:00-12:00)**

09:00 10:30 - Gap analysis and recommended actions: biobanks

10:30 12:00 - Gap analysis and recommended actions: registries and natural history

## EXECUTIVE SUMMARY

The 4<sup>th</sup> meeting of the IRDiRC Interdisciplinary Scientific Committee (ISC) took place in Montreal, Canada on 17<sup>th</sup> and 18<sup>th</sup> October 2013. ISC members addressed the Working Groups outputs (Registries and Natural History; Biobanks; Ethics and Governance; Data Sharing) after all of them had held one or two teleconferences. ISC members also discussed the gap analysis and recommendations for actions to be included in the IRDiRC road map. A tentative list of preliminary recommendations for consideration by the other parties, in particular the Working Groups and the two other Scientific Committees was developed. The Co-Chair also fed back discussions of the Executive Committee meeting held in September in Miami. Other issues discussed encompassed the WG role, composition and objectives, organisational and internal governance aspects as well as interactions with the Scientific Secretariat.



## REPORT

### **Governance and organizational issues**

#### **Road map and gaps' analysis**

A first draft of the road map and gap analysis is expected by the end of November when the Executive Committee holds a teleconference. This road map is to be drafted jointly with the two other SC. It is expected to be signed off by the Executive Committee at its next face-to-face meeting in Berlin, in May, as the Executive Committee will meet before the European Conference on Rare Diseases and Orphan Products (ECRD), organised by Eurordis from 8th to 10th of May. This document is to be scientifically oriented. The Executive Committee expects feedbacks and recommendations from the three SC which should advise the Executive Committee members on what they should support financially.

#### **Working Groups role and composition**

WGs were awaiting directions and clearer instructions to further develop their activities. ISC members themselves expressed that more orientation and feedback from the Executive Committee would help setting the priorities. It was proposed that flexible solutions could be considered for the WGs.

#### **Interactions with the Scientific Secretariat**

Interactions with the Scientific Secretariat are now operational. With regard to the fact that WG have no funding to develop their activities, it has been underlined that the Scientific Secretariat can be asked to support some efforts. The secretariat may help centralising and collecting information. Comparative studies on specific tools or initiatives can be initiated by the Secretariat.

#### **Interactions with the Scientific Committees**

Interactions with the two other SC should be developed and maintained as the ISC is aimed at supporting the major objectives set by IRDiRC in the two other SC. In view of the roadmap, interactions will be developed in the coming weeks with the two SC Chairs.

#### **ISC composition**

Petra Kaufmann, who was unable to attend the Montreal meeting, is now an active member of the ISC. No additional member is expected for the moment in the ISC.

#### **Funding opportunities to develop IRDiRC objectives**

Potential funding opportunities are expected from the forthcoming Framework research programme of the European Commission: "Horizon 2020".

## Working Groups contribution to the ISC

### Ethics and Governance

- ▶ There is a need to create a questionnaire to capture the sensitivity, aspirations and hopes of the RD patients and families about data sharing. The perception of patients with rare diseases might be different from the ones with common diseases as the former may be more willing to share data if it would help the discovery of therapies.
- ▶ It is recognised that rules and laws are different from one country to another in Europe and North America. Nevertheless, cross continental data sharing requires **a minimal consensus about standards, guidelines and policies amongst countries.**
- ▶ With regard to **clinical trials**, it was stressed that a common framework is needed for ethics and governance approval. The level of risks could be distinguished between information risks vs. physiological risks when dealing with clinical trials. Central ethics review would clearly help multicentric clinical trials. Whereas it is done in the US (being a prerequisite before the trial starts), it is found difficult to implement at the EU Level.
- ▶ An international consensus on **giving back the information to the patients is not a possible goal** as of today, this is a matter of medical judgement and what is medically actionable may rule what information is given back to patients.
- ▶ The **EU Data Protection Regulation** was addressed as an area requiring further action.
- ▶ The P3G (Public Population Project in Genomics and Society) newly created **IPAC service (International Policy interoperability and data Access Clearinghouse)** is an office working on the harmonization of data sharing laws around the world and also to a separate project to create an **Ethic Safe Harbour**, a research project whose objective is to recognize ethics review process equivalency.
- ▶ An update and clarification on the **Global Alliance** initiative was also made. The initiative was launched in 2012 with the idea to promote the sharing of clinical and genetic data. A New York meeting with in-person representation (i.e. not institutional) was held in January 2013. A signed Letter of Intent was made public in June 2013 with declared intention to work on the idea and ideal of a Global Alliance. Organizations from 40 countries signed this document. As of today, an administrative and Scientific Secretariat team, with three principal hubs, supports the Global Alliance: (i) California dealing with the governance structure; (ii) Toronto with the expert working groups; (iii) Boston with technical and computing issues. The ICGC is already considering developing pilot projects with the Global Alliance. Currently, no funding will be made available from this initiative but opportunities to test and create a prototype on rare diseases could be envisioned. A MoU will become available for signature in 2014 to all interested parties, including IRDiRC.
- ▶ Incentives should be found to increase and encourage the entering of information in databases. Reimbursement may be an option. The MDA (Muscular Dystrophy Association) was cited as an example of co-funding such initiatives. The question is whether this can be done internationally for all rare diseases.
- ▶ ISC members agreed that a Minimal Data Set or standard is needed. Best practices guidelines on databases and standard ontologies are also needed.

## Registries and Natural History

- ▶ WG has dedicated efforts to GUID (Global Unique Identifier). The GUID relies on the observation that different datasets are used for the same patient in different institutions. The concept of GUID is to develop identifiers that would be similar at all places for the same patients as it has been developed for Huntington for instance. With the GUID, data are collected, crypted and de-identified through the generation of codes. Adopting an IRDiRC “GUID” is to be questioned and discussed as it is a cross subject over the different SC and WG. Questions arose with regard to the implication in the public domain and to the possibility for the consent form to be included in this system.
- ▶ Different gaps for registries were identified: the lack of sustainable funding, the critical issue of the entry point for filling the data (both time and funding are necessary to fill the data), the resources needed to explore what exists, the need for a clearing house, the need for criteria and quality standards. The question of quality assessment should be investigated..

## Data sharing and bioinformatics

- ▶ Technologies are dramatically changing the way we are thinking registries and generating new ways of collecting information and reliable data seem unavoidable.
- ▶ There is not necessarily a need to build new registries but to collect daily data differently (DVT technology, PCORI (patients centred outcome research initiative) and PPRN should be envisioned in order to develop new funding and business models). Feasibility studies would be needed in that perspective.
- ▶ The WG agreed with the principle that researchers funded by IRDiRC should submit their data. One key idea of the WG is the “sharability” of data for the IRDiRC projects.
- ▶ It is clearly stressed that it is important to deposit things that are useful and that standards should be shared, not only information.
- ▶ A common place is needed where it is possible to communicate and exchange with patients and groups involved in similar studies, a Clearing House that would facilitate researchers’ goals. Scientists could monitor the information. Settling a Clearing House will require time and resources. ISC discussed the opportunity to seek extra funding for a Clearing House. The requirements and specifications of such a Clearing House (How resource intensive will it be? How many standards and sources?) should be explained prior to any further activity or application for funding.

## Biobanks

- ▶ Unravelling the unique challenges in rare diseases research and explicating the specificities of RD biobanking (particularities with regard to the existing standards, specificities in collecting samples and data for rare diseases, encouraging some areas more than others to share data due to the scarce amount of data)
- ▶ Anticipating the analysis that might be undertaken, making sure that the samples are fit for purpose.
- ▶ Addressing the issues of quality assessment, biobanks’ certification/accreditation. A scoring system for biobanks would certainly find consensus within WG members. The main question is

how to implement it. Preparatory work could be envisioned with representative from BBMRI in the WG as BBMRI has already developed those scoring systems for more common diseases.

- ▶ Addressing the specific issue of rare diseases paediatrician research. Requirements are usually more rigorous when research deals with children. This is not necessary significant when rare diseases are concerned.
- ▶ Unlocking the value of legacy biobanks.
- ▶ Addressing the legal constraints of ownership.