

## Meeting report series

### Report of the 5th Interdisciplinary Scientific Committee (ISC) Meeting

Teleconference  
13 February 2014

#### Participants

Prof Hanns Lochmüller, Newcastle, UK (Chair)

Prof Jamel Chelly, Paris, France (co-chair)

Prof Bartha Maria Knoppers, Montreal, Canada

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

Dr Angel Carracedo, Santiago de Compostela, Spain

Ms Roseline Favresse, IRDiRC Scientific Secretariat, France

Dr Barbara Cagniard, IRDiRC Scientific Secretariat, France

Dr Sophie Höhn, IRDiRC Scientific Secretariat, France

#### Apologies

Prof Jack Goldblatt, Perth, Australia

Dr Petra Kaufmann, Bethesda, USA

#### Agenda

##### 2:00-3:00 pm

- ▶ Publication and status of the Montreal minutes
- ▶ Position papers to be issued from the ISC?
- ▶ Comments on Diagnostics SC Roadmap
- ▶ Meeting of the ISC in Berlin?
- ▶ Working group re-organisation and tasks
- ▶ Shenzhen planning

## REPORT

### Publication and status of the Montreal minutes

Short and full versions of the minutes were produced after the Montreal meeting. Full minutes have been shared with the Executive Committee. The Scientific Secretariat wanted to publish the full version of the minutes. A public version has been drafted.

As a whole, there is a consensus among the ISC members with the proposition to publish a full version of the minutes on the website and to be as transparent as possible.

### Position papers to be issued from the ISC?

Point whether the document drafted at the Montreal meeting could be published in a scientific review in order to disseminate better and give added value to the work/propositions developed by the ISC.

As a whole, there is agreement from all ISC members that it would be valuable to publish. This should be done in coordination with the Diagnostics and Therapies Committees in order to have an "IRDiRC moving forward paper". The Diagnostics SC intends to publish in the American Journal of Human Genetics.

**Action:** list topics that could be part of this common position paper (based on the work already done at the Montreal meeting and in agreement with WG). This short list of topics could be discussed at the next conference call/meeting and shared with the two other SC.

### Comments on Diagnostics SC Roadmap

The Diagnostics SC met in Prague in December and has produced its first roadmap. This document (DSC Report 4th meeting Prague) was sent to all ISC Members on 1 February. An additional document (PowerPoint Presentation) was presented at the latest Executive Committee meeting (3 February, by confcall). This document has been very appreciated by Executive Committee members. The chair of the committee emphasizes the interesting input and proposition to work towards a matchmaking tool for genome/phenome data.

**Action:** The DSC would be happy to receive comments from the ISC members.

- ▶ The DSC will also comment on the ISC roadmap.
- ▶ The Executive Committee would like that the three SC merge their roadmap in order to have a single document by the Berlin meeting (7-8 May).

## **IRDiRC Shenzhen conference**

Rumen Stefanov is the ISC representative in the organisational committee for the Shenzhen meeting. New dates have been defined for the Shenzhen meeting: 7-9 November 2014. This information will be included in the next IRDiRC newsletter to be sent to all IRDiRC members and participants soon.

Some ISC members are already willing to participate, the others should inform the group.

No recent developments have occurred with regard to the programme. The discussion on potential speakers is to be held next week.

Proposition of presentation: "The International Code of Conduct for Genomic and Clinical Data Sharing" on behalf of the Global Alliance for Genomics and Health. Since several China institutes and academies are members that should be of interest for the audience.

Questions were raised with regard to which topics would be of interest for the Chinese/Asian audience of the meeting : some topics may be less relevant than others. Registries for instance are at very early stages of development. Equally, orphan drug development is not a first issue at the moment in China. On the opposite, integrating activities are quite impressive (great capacity from Chinese groups to integrate biobanks, genomics data and clinical data into meaningful set of data).

## **Working groups re-organisation and tasks**

It is important to feedback the discussions held in Montreal to the Working Groups as well as to thank them for their inputs and ask for any additional inputs they may have.

Next actions for WG discussed within the ISC and to be further discussed at the WG level:

- ▶ **Bioinformatics and Data Sharing Working Group:** the Executive Committee was very positive about the idea to further work on the "Clearing House for data sharing" --> the WG should go on focusing on this subject and provide additional input/more detailed information on how such a Clearing House would work (including financial inputs; even if this is expensive). A group that could focus on this issue could also be identified.
- ▶ **Ethics and Governance Working Group:** need for the WG to be contacted by other WG with specific and concrete demands, willing to pursue its involvement.
- ▶ **Registries and Natural History Working Group:** advised to link with Therapies SC and WG. Group willing to pursue its involvement. Specific topics to be defined answering the main gaps and obstacles that need to be addressed by IRDiRC.
- ▶ **Biobanks Working Group:** it is necessary to define whether there is a distinct role for this WG, if the questions raised (historic collections, etc.) are sufficient to maintain the group alive and if an extension to new participants is needed. It was reminded that P3G has RD support and can contribute to IRDiRC. It is important to define the key questions regarding biobanks that can affect RD research developments.

## Meeting of the ISC at Berlin

There is a possibility to have one meeting of the three SC on the 8th May afternoon. The decision whether this meeting will take place will be taken next week (based on doodle answers). If a joint all-science committee meeting is not feasible, then the ISC would organize a meeting for the ISC only at the same time.

For information, the Therapies SC will meet in March in Paris.

## Deliverables

- ▶ Send comments on the public version of the Montreal report by 26 February.
- ▶ List topics that could be part of a position paper common to the 3 SCs all members.
- ▶ Send comments on DSC roadmap by 26 February.
- ▶ Send topics of interest for Shenzhen conference.
- ▶ Circulate roadmap and feedback from ISC to WGs.
- ▶ Organize WG teleconference.