

Meeting report series

Report of the 3rd Interdisciplinary Scientific Committee (ISC) Meeting

Dublin, Mespil Hotel
15th April 2013

Participants

Prof Hanns Lochmüller, Newcastle, UK (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, Rome, Italy
Mr Alastair Kent, London, UK

Mrs Roseline Favresse, Scientific secretariat, France

Apologies

Dr Jeffrey Krischer, Tampa, USA (not attending the ISC but reporting on the WG discussions)
Dr Angel Carracedo, Santiago de Compostela, Spain
Prof Jamel Chelly, Paris, France (vice-chair)

Agenda

Meeting on the 15th April

1. Report by the chair on the Executive Committee meeting
2. Informal impressions from the working groups meetings
3. Discussion on IRDiRC road map
4. Other business

EXECUTIVE SUMMARY

The 3rd meeting of the IRDiRC Interdisciplinary Scientific Committee (ISC) took place in Dublin on 15th April 2013.

ISC members reported on the workings group (WG) meetings that took place on the same day. The four WG related to ISC (Registries and Natural history; Biobanks; Ethics and Governance; Data sharing and Bioinformatics) were invited to meet for the first time on the occasion of the IRDiRC Dublin conference. The mandate, composition, objectives and next steps for the WG were discussed.

The chair of the ISC reported that the IRDiRC Policies and Guidelines document was approved by the members of the Executive Committee when meeting in the afternoon of the 15th. The Executive Committee also defined the steps towards the efficient development and implementation of the IRDiRC roadmap.

The ISC members would like the WG to have the possibility to meet face-to-face sometimes as critical topics cannot always be discussed through teleconference. The means to find additional funding may need to be considered for this purpose.

The problem of the sustainability of registries was discussed in regard with the changes in regulation and examples provided by initiatives.

REPORT

1. Reports from Working Groups

WG “Registries and Natural History” – 1st meeting – 15th April

The goals of the meeting were to discuss and nominate Chair and co-Chair, to discuss and comment on topics and goals of the WG and to identify needs, gaps and opportunities

The following topics were discussed: the importance of using Common Data Elements, the need for interoperability, the Global Rare Diseases Repository project (GARD) and other funded projects (e.g., RD-Connect, Epirare, GRDR).

Quality assurance and data integrity (linked to core data set) were subsequently discussed. Additional ideas that were also discussed extensively:

- ▶ Sustainability plan enabling self reported data and clustering of registries. Funding will be a major issue and may affect the amount of data to be collected.
- ▶ Global unique patient identifier ID. Standardization of outcomes measures.
- ▶ Medical Records as a source of data.

Participants proposed potential collaboration with other Working Groups. Other WG members could be joining the next meeting of this group. A possible way to approach this task could be to initially establish principles, and next to collect and share existing tools and/or guidelines, and focus on implementation.

A follow-up phone call was planned to take place before the summer.

WG “Biobanks” / “Ethics & Governance” (the two WG merged) – 1st meeting – 15th April

Two main topics were discussed:

- ▶ The urgent need for IRDiRC to inform the EU Parliament’s discussions on the need to include an exception for biomedical research as concerns the need for an explicit specific consent for every sample/data use in the proposed Directive (regulation) on data protection. IRDiRC cannot function as an international collaborative research endeavour in the absence of such exception.
- ▶ The need to organise an IRDiRC sponsored meeting on the “Governance of international data sharing”. The WG are ready to assist in the organisation of such a meeting with a focus on the development of a “Governance Framework”.

Follow-up phone calls will take place before the summer for the 2 groups separately.

WG “Data Sharing” – 1st meeting – 15th April

A part of the time was spent on introducing participants and describing the setting in which they work. This was very helpful as many common areas of interest have been highlighted and work environments are in some aspects very similar.

In order to approach the main discussion goals - harmonization, data sharing, etc - it was proposed that use cases should be developed that could be circulated to the WG members. It was felt that it would be easier and more revealing to compare how each WG member would respond to each use case to describe the approach and methods.

Participants complemented each other in the sense of their approach to data whether they were coming from a genotype or phenotype direction. The data issues appear to be quite different in each setting and there was utility to discussing these perspectives separately.

Indeed there seemed to be value in considering issues from several different dimensions:

- ▶ Diagnostics: "omics", metabolomics and immunological testing (or more generally clinical testing)
- ▶ Genotype-phenotype correlations
- ▶ Therapeutics: including clinical trials; biomarkers; definitions and measures of response
- ▶ Etiologies: gene-environment interactions
- ▶ Regulatory: ethics, confidentiality, intellectual property
- ▶ Follow-up phone call will take place before the summer.

Main points from the WG meetings:

- ***4 WGs established with first face-to-face meeting***
- ***ISC members reported from the WG they attended to the ISC***
- ***A teleconference will be organised as a follow-up for each WG***

2. Report from Executive Committee Meeting

The chair of the ISC fed back to ISC members with regards to the Executive Committee meeting held in the afternoon of the 15th. One main achievement concerns the IRDiRC Policy and Guidelines document that was finalised and adopted by the Executive Committee members.

He explained that following the Executive Committee meeting in Barcelona (January 2013), the Executive Committee has requested a shortened and re-ordered version. Accordingly, the policy and guidelines document was substantially reworked, shortened and simplified by IRDiRC Scientific Secretariat and the 3 science committee chairs. The condensed and reordered version now lists a set of rules, which may be used as an annex to IRDiRC funders' agreements. The more extensive version in the original ordering system will be kept as a reference document while the shorter one will be systematically attached to contractual document from IRDiRC funders.

3. Discussion on the roadmap

The roadmap document - a document prepared by the IRDiRC Secretariat team - was discussed by the Executive Committee members for the first time. The “action list” that was presented was partly lifted from the policy document. The Executive Committee expressed its wish to first define a process to get to a roadmap. The next step is thus for the IRDiRC Secretariat team to provide an updated list of the action plan. The 3 science committees will then be invited to provide review and provide input to the executive committee in the fall. This should include feedback from the working groups.

4. Other issues discussed

Among other issues discussed during the ISC meeting, the following one has been highlighted:

- ▶ The issue of **funds’ allocation within the budget of the scientific secretariat**. The observation is that no budget is available to fund the WGs themselves. WG are expected to deliver and exchange on critical topics. Therefore, there may be a need to create the opportunity for people to meet in addition to the planned phone calls. If no money is available within the current budget, the means to find additional funding has to be considered.
- ▶ **Composition of WG:** It was questioned if the WG as they are constituted are the adequate organisation to advise on gaps and on what to do. Indeed, there is a gap between (i) finding and delineating the priorities and (ii) defining a framework for good practices.
- ▶ **Sustainability of registries:**
 - The Reg4all initiative, launched by Genetic Alliance and developed as self-reported registries, was discussed.
 - Complementary issues discussed after the meeting:
 - The opportunity to develop advocate actions towards the EC to include a call, under the IRDiRC initiative, for registries in order to ensure sustainability has been pointed out as a potential action for the ISC.
 - The example of the Public Health England who has established a national congenital abnormality register (which, it is intended, will grow into a national rare disease register in time) has also been quoted. The advantage of this initiative is that being part of the public health and service planning agenda, the tool is not vulnerable to changes in R&D funding priorities at the end of a grant.

Next ISC Meetings

October 17th-18th, 2013 in Montreal