

Meeting report series

Report of the 8th Interdisciplinary Scientific Committee Meeting

Teleconference
4 February 2015

Organization

Organized by: Scientific Secretariat
Teleconference

Participants

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Agenda

1. Decision of the Executive Committee at Shenzhen (short summary)
2. Priorities, task forces and workshops in relation to the Interdisciplinary Scientific Committee (discussion)
3. Meeting of the Interdisciplinary Scientific Committee in Glasgow June 5, 2015

REPORT

Decision of the Executive Committee at Shenzhen (short summary)

The Chair reported on the latest developments and key decisions of the Executive Committee (Exec Comm) of IRDiRC. The Exec Comm made a restructuring decision in Shenzhen, where all Working Groups (WGs) would be discontinued and new Task Forces (TFs) be constituted for more refined remits. An official letter was discussed with the chairs of the three Scientific Committees (Sci Comms) to be sent to all members of the Sci Comms to explain these changes, and another to be sent to all members of WGs to thank them for the contributions. Further details can be found in the minutes of the Exec Comm meetings of the 6th November 2014 and 26th January 2015 (reports available on IRDiRC public website, under IRDiRC activities).

IRDiRC Task Forces

The Chair provided a brief introduction to the organisation of TFs:

- ▶ All Sci Comms may suggest topics/projects for *ad hoc* TF formation to the Exec Comm, although implementation may not be immediate as only a limited number of TFs can be managed at the same time.
- ▶ A TF is usually associated with a workshop where experts on a particular topic would be invited to propose solutions or recommendations that will advance IRDiRC's goals [*Post-meeting note: all workshops will be organised under the umbrella of IRDiRC and will not be attached to specific Sci Comms*].
- ▶ All Exec Comm and Sci Comm members would be invited to nominate experts, be it individuals already involved with IRDiRC or not, to be part of the TFs.

The modalities on how TF members and participations are selected are described by the secretariat:

- ▶ Suggestions from all members of the Exec and Sci Comms using nomination forms, which contain brief descriptions of the selected topics, sent out by the Scientific Secretariat (Sci Sec).
- ▶ Compilation of nominated names by the Sci Sec.
- ▶ Development of a list of “must” and “potential” participants.
- ▶ Appointment decision by all members of Sci Comms through a vote organised by the Sci Sec.
- ▶ Notification to appointed members.
- ▶ Selection of a Chair and a Co-Chair for each TF by its members.

Selected topics put forward by the Interdisciplinary Sci Comm

The Interdisciplinary Sci Comm proposed two high priority activities last year for seed funding, but as the Exec Comm was unable to directly fund these projects, the following has been proposed:

- ▶ Data standards clearinghouse: to create a (static) resource database.
- ▶ Computable consent form: to create a TF that will lead a workshop.

Data standards clearinghouse

The premise of this proposal is to create a public clearinghouse that would lead to the creation and adoption of data standards and common data elements across the community of stakeholders in rare diseases. The proposed plan includes a dynamic dimension, where new data standards may be input by users, and use cases and references to standards would also be available.

The Sci Sec has been tasked to create a resource database of data standards. However, this is only part of the original proposal and a static database would be of limited use. In essence, it will require solicitation of information to be provided to the Sci Sec but this duplicates existing and available resources. One example of a list of standards already available can be found on NINDS website; it does not, however, provide information on who used these standards, how they were used and the experience in using them.

A dynamic clearinghouse is a more valuable, innovative and long term approach that requires more substantial commitment and resources. New work could be built on existing results when users could access not only available standards but also associated use cases. This would facilitate and encourage data sharing, which is much needed in rare disease research. However, given the limited resources currently available, external funding and human resources to update the tools would be necessary.

A possible alternative would be to pilot a few standards and use cases to make a stronger argument of the utility of the dynamic clearinghouse, and then seek more resources to expand and make it more comprehensive. The committee also explored the possibility of redirecting the financial resources, should a workshop not be organised, to funding the project.

Over the next weeks, further discussions will be held to explore repositioning this project in the context of personalised medicine initiatives to seek funding. The secretariat will move this priority project forward with the support of the Exec Comm and ISC.

Computable consent form

The initial proposal set out to develop model consent clauses for rare disease research, with a focus on model consents in the area of paediatric rare disease research that is particularly lacking. Following the discussion at the Exec Comm meeting, this proposal was altered to include a computable element, with the aim of setting up a workshop to establish elements to computerise based on standards and to create clear records of what patients agree upon as well as to identify which samples are usable in the future, in coordination with the Global Alliance for Genomics and Health (GA4GH) and other initiatives (e.g. RD-Connect).

Within GA4GH, a new Task Team (TT) on machine-readable consent forms was to be formed in mid-February 2015. It is not a paediatrics-specific TT and the consent would be geared for data sharing. It is important that IRDiRC be included in the TT and accordingly, there has been expression of interest from Sci Comm members. Members of the Interdisciplinary Sci Comm interested in this GA4GH TT should inform the project leader for inclusion in the TT.

Other selected topics for IRDiRC workshops

Matchmaker Exchange

This is an initiative co-led by the Diagnostics Sci Comm co-Chair, in joint effort with GA4GH. Members of this TT met recently to try to finalise an API to connect different matchmaking databases, including Phenome Central, Gene Matcher, GEM.app and DECIPHER. Follow up meetings have been planned, among them an IRDiRC workshop in conjunction with the ASHG conference in Baltimore in October 2015. There is keen interest from several members of the ISC and previous working groups to be informed and included in this initiative.

Small population clinical trials

This is an initiative suggested by the Therapies Sci Comm and the TF will plan a workshop that includes discussion with the EMA/FDA on guidelines for small clinical trials that are particularly relevant to rare diseases. It will also touch upon adaptive design and statistical methods. It will be vital to nominate key people on this TF. Both FDA and EMA are respectively drafting and updating guidelines related to clinical trials, and the outcome of the workshop may be recommended for incorporation into their guidelines. EMA will jointly host this workshop with IRDiRC. A member of the ISC has expressed an interest to be included.

Patient relevant/reported outcome measures

This is another topic suggested by the Therapies Sci Comm, which aims to set up a workshop to make use of case studies and advance the HTA process, especially where biomarkers are absent in diseases. Three members of the ISC have expressed an interest to be included. This topic is also of relevance to patient registries that frequently use patient reported outcomes, and interest from members of the former working group on registries and natural history studies is expected.

The Interdisciplinary Sci Comm face-to-face meeting

The next face-to-face meeting of this committee will take place in Glasgow, Scotland, on 5 June 2015 for a full day. On the morning of 6 June 2015, a joint meeting with the Diagnostics and Therapies Sci Comms will take place. The IRDiRC meetings are followed by the ESHG conference in Glasgow (<https://www.eshg.org/eshg2015.0.html>).

Main action points

- ▶ Letter sent to all members of Interdisciplinary Sci Comm from the Exec Comm (Sci Sec)
- ▶ Send letter to all members of WGs to thank them for the contributions (Sci Sec)
- ▶ Nomination forms for TF participation sent to all members of the ISC (Sci Sec)

- ▶ Organise the next steps on two priority topics with members of the ISC (Sci Sec)
- ▶ All members to suggest possible TF topics
- ▶ Organise the next teleconference (Sci Sec)