

Meeting report series

Report of the 9th Interdisciplinary Scientific Committee Meeting

Teleconference
15 May 2015

Participants

Prof Hanns Lochmüller, Newcastle, UK (Chair)
Dr Angel Carracedo, Santiago de Compostela, Spain
Prof Jack Goldblatt, Perth, Australia
Dr Stephen Groft, Bethesda, USA
Dr Petra Kaufmann, Bethesda, USA
Prof Bartha Maria Knoppers, Montreal, Canada
Dr Jeffrey Krischer, Tampa, USA
Ms Samantha Parker, Paris, France
Prof Rumen Stefanov, Plovdiv, Bulgaria
Dr Domenica Taruscio, Roma, Italy

Dr Lilian Lau, Scientific Secretariat, Paris, France

Apologies

Prof Jamel Chelly, Strasbourg, France
Mr Alastair Kent, London, UK

Agenda

1. Update from the Executive Committee and Operating Committee
2. Update on Task Forces
3. Glasgow Agenda: Interdisciplinary Scientific Committee Meeting
4. Glasgow Agenda: Joint Scientific Committees Meeting

REPORT

Update from the Executive Committee and Operating Committee

The Executive Committee (Exec Comm) met twice recently: a face-to-face meeting on 16 March in Madrid, Spain, which the Chair of the Interdisciplinary Scientific Committee (ISC) was unable to attend, and a teleconference call on 11 May. The Chair also attended a teleconference of the Operating Committee. Most discussions revolve around newly constituted Task Forces and their implementations.

Update on Task Forces

General steps of Task Force formation:

- ▶ Nomination of experts by the Exec Comm and Sci Comms
- ▶ Compilation of all names received by the Scientific Secretariat (Sci Sec)
- ▶ Provision of compiled names to the Exec Comm
- ▶ Selection of core group members by the Exec Comm (c.f. 14th Exec Comm Report)
- ▶ Expansion of Task Force membership by core group members

The formation of Task Forces may differ in some circumstances, e.g. when working in collaboration with Global Alliance for Genomics and Health (GA4GH), where there are already pre-formed groups.

The Task Forces which are currently being formed:

- ▶ Patient Relevant/Reported Outcome Measures (PROM) – core group selected
- ▶ Small Population Clinical Trials (SPCT) – core group selected
- ▶ Data Mining/Repurposing (DMR) – core group not yet selected
- ▶ Matchmaker Exchange (MME) – with GA4GH
- ▶ Machine Readable Consent (MRC) – with GA4GH

The Chair was slightly concerned that ISC nominees were not selected for core groups but was assured:

- ▶ that there will be more individuals who will be called to contribute to Task Forces, review documents and attend the workshop,
- ▶ that there will be information on all individuals asked to contribute, and
- ▶ the core group members are mainly people running large consortia related to the topics, and of which the nominations by the ISC do not fit the description.

The Sci Sec will assist the Task Forces by preparing background paper when required, discussing and completing the background paper with members of Task Forces, and organising planned workshops or face-to-face meetings. This role is also extended to Task Forces in joint action with GA4GH.

Members of the ISC who wish to put forward additional names to the Task Forces may do so by sending the names to the Sci Sec, copying the Chair of the ISC. The Sci Sec continues to accept nominations as they may be invited to be a part of the non-core Task Force members, attendees of workshops, or reviewers of workshop papers.

Machine Readable Consent Task Force

Three members of the ISC have agreed to join the MRC Task Force. This Task Force will try to meet at the ASHG, or preferably, later in Paris, after a number of initial teleconference to set up the landscape of the work needed. Members participating in this Task Force were reminded to keep this project on agenda for IRDiRC and report back to the Exec Comm to let them know of progresses and actions.

Glasgow Agenda: Interdisciplinary Scientific Committee Meeting

The ISC members will be lodged and will meet at Premier Inn on 377 Argyle Street, Glasgow, G2 8LL, UK. The specific meeting rooms are:

- ▶ ISC meeting: Argyle Room
- ▶ Joint meeting: Clyde Room

The ISC will meet for a full day on 5 June in Glasgow, starting at around 9am. The main topics of discussion would be on future Task Forces to present to the Exec Comm. The Chair will complete the full agenda in due course.

Members were asked to think of potential Task Forces for future actions which would be the starting point for discussion and subsequent identification of the order of prioritisation among the proposed topics. Some of the suggestions during the teleconference are listed below.

Additional suggestions for Task Forces are welcomed and should be submitted to the Chair of the ISC, copying Sci Sec, within the next 10 days. Proposers are to submit a small paragraph to define what the Task Force should be about, what it aims to achieve and what is/are its expected outcome(s). Proposers are expected to take a lead in the discussion during the Glasgow meeting and present their Task Force idea in brief, in particular how this is related to the IRDiRC objectives, previous work of the committee and working group, whether a meeting would be useful/appropriate, approximate timing, and deliverable/outcome.

Suggested Task Force: Targeted Newborn Screening

GA4GH's Paediatrics Task Team – co-led by Jan Friedman and Martina Cornel – recently put out a call for membership. This Task Team's first priority is to examine regulatory and ethical issues in genomic newborn screening, and to develop policies that would provide international guidance which can later be extended to children and adolescent. [Post meeting comment from the Sci Sec: this topic falls outside the scope of IRDiRC which focuses on research issues, not on services.]

Suggested Task Force: Gene Therapy in Children

The time is ripe to investigate gene therapy in children, to see what is needed now in terms of policy, to examine new safety issues, etc.

Suggested Task Force: Electronic Health Records Integration

This project aims to look into the integration of data from different sources, specifically the use of data generated in a clinical context and captured in the electronic health record for clinical research purposes. Increasing penetration of electronic health records and advances in data technology means that there will be opportunities to accelerate rare disease research. Future data integration will require advanced planning and the development of new policies, interoperable research data warehouses and standard terminologies.

Suggested Task Force: Research Participant (Global Unique) Identifier

Global unique identifier (GUID) should be coordinated to facilitate and advance rare diseases research while alleviating privacy concern by data sharing that is detached from personally identifiable information. The NIH is making use of GUIDs, but not a “single unique” exists.

Suggested Task Force: Best Practices in Patient/Stakeholder Engagement

This is an overarching issue that looks into best practices in patient engagement, to promote the engagement of patients, families and other relevant communities early on and as active partners, rather than engaging them as “consumers” of research results. Best practices could be discussed in a forum like IRDiRC and this may lead to a paper with recommendations to adopt.

Suggested Task Force: Medical Devices in Rare Diseases

With large portfolio of medical devices available, it is important, and timely, to refine what medical devices are of particular interest to rare diseases and rare disease research. NCATS has been looking into development of better devices for intrafoetal delivery of therapeutics and recently organised a workshop with the FDA and other stakeholders on this topic. Conforming to regulatory processes is another issue to be addressed in order to develop right devices to deliver therapies in rare diseases. [Post-meeting comment from the Sci Sec: there is a US policy of medical devices for rare diseases but there is none in Europe.]

Glasgow Agenda: Joint Scientific Committees Meeting

The ISC will be joined by the Diagnostics Sci Comm (DSC) and the Therapies Sci Comm (TSC) for a half-day meeting on 6 June in Glasgow.

Potential agenda items to put forward:

- ▶ Feedback of PROM Task Force draft background paper
- ▶ Future Task Forces and joint committee input
- ▶ Defining mandate of the Sci Comms
- ▶ Publication of position paper and further publications

PROM draft background paper

A member of the ISC was excited and pleased to see a good inventory of patient outcome measure initiatives included in the draft paper, and suggested the inclusion of NeuroQOL, an initiative of the NIH. The joint Sci Comm meeting could be a good opportunity to discuss the background paper further and provide a more complete landscape, e.g. to add interdisciplinary aspects to help patients report on therapeutics through natural history. The Sci Sec was asked to make the background paper available to all members of Sci Comms ahead of the meeting.

Main action points

- ▶ Send paragraphs on potential Task Force topics
- ▶ Compilation of Task Force ideas, listed in this report and any additional submission(s) by email, as a preparatory document for Glasgow meeting
- ▶ Forward background paper on PROM to members of Sci Comms
- ▶ Agenda(s) for Glasgow meetings