

Meeting report series

Report of the 14th Interdisciplinary Scientific Committee Meeting

Teleconference
8 September 2016

Participants

Prof Hanns Lochmüller, Newcastle, UK (Chair)
Dr Petra Kaufmann, Bethesda, USA (Co-Chair)
Ms Gema Chicano, Murcia, Spain
Prof Jack Goldblatt, Perth, Australia
Prof Bartha Maria Knoppers, Montreal, Canada
Dr Jeffrey Krischer, Tampa, USA
Prof Rumen Stefanov, Plovdiv, Bulgaria
Dr Domenica Taruscio, Roma, Italy

Dr Lilian Lau, Scientific Secretariat, Paris, France
Dr Anneliene Jonker, Scientific Secretariat, Paris, France

Apologies

Dr Angel Carracedo, Santiago de Compostela, Spain
Dr Stephen Groft, Bethesda, USA
Ms Samantha Parker, Paris, France

Agenda

1. Updates by Chair of the ISC
2. Membership of the ISC and potential new nomination(s)
3. Update on Task Forces
4. 2017 Conference in Paris
5. Setting goals for the next 5 years

REPORT

1. Updates by Chair of the ISC

The Chair of the Interdisciplinary Scientific Committee (ISC) welcomed its members and outlined some changes within IRDiRC structure since its last meeting in Lyon.

- ▶ The Executive Committee has been renamed Consortium Assembly (CA)
- ▶ As the CA grows, in order to work efficiently, constituent committees were created
 - Funders Committee
 - Patient Advocates Committee
 - Companies Committee
- ▶ Operating Committee (Op Comm) runs monthly meeting, consists of
 - Chair and Vice Chair of CA
 - Co-Chairs of Scientific Committees (Sci Comms)
 - Scientific Secretariat (Sci Sec)

2. Membership of the ISC and potential new nomination(s)

- ▶ Upcoming renewal
- ▶ New nominations

3. Update on Task Forces (TFs)

Two TFs aligned to ISC are joint efforts with Global Alliance for Genomics and Health (GA4GH), which made much progress and could present their work in upcoming IRDiRC meetings/conference.

3.1 Participant Unique Identifier (PUID) TF

- ▶ Co-Chaired by Dixie Baker, Petra Kaufmann and Bartha Knoppers
- ▶ Aim: to look at internationally recognised PUID
 - Background document available
 - 6 use cases have been developed
 - Two subcommittees: one to look at ethical and legal issues, one on technical issues
- ▶ Will meet in Paris on 8-9 December for a face-to-face workshop
 - Sci Sec due to start organising travel for selected participants
- ▶ Any ISC member wishing to have more information, documents or report, please contact Sci Sec

3.2 Automatable Discovery and Access (ADA) TF

- ▶ Co-Chaired by Anthony Brookes and John Wilbanks
 - Formerly Machine Readable Consent (MRC)
- ▶ Recently completed ADA-Matrix (ADA-M); currently in 30-day public consultation period

- On IRDiRC site: <http://www.irdirc.org/draft-version-of-ada-matrix-open-for-comments/>
- Larger public demonstrations will take place at the GA4GH meetings prior to the ASHG in Vancouver, on 16 and 18 October; will be tested and validated in October
- Once finalised will be put on IRDiRC website as a tool for the community
- ▶ Very useful prospective tool but use with retrospective consent may be limited

3.3 Development of guidelines: Post-Marketing Industry-Independent Surveillance Registry

- ▶ This was previously suggested for formation of a TF, idea further developed
 - An article has been submitted for publication
 - Emphasis on modular, easy-to-use, web-based, patient-centred RD registry
 - Would be model to develop such registry independent of industry
- ▶ Could be further discussed in the next meeting
 - In the mean time, the original manuscript will be shared with members

4. 2017 Conference in Paris

- ▶ Important – members encouraged to save these dates:
 - 6-7 February: IRDiRC meetings of the CA and Sci Comms
 - 8-9 February: 3rd IRDiRC Conference
- ▶ Logistics for Sci Comm members
 - Travel and accommodation for IRDiRC meetings will be covered by Sci Sec
 - Sci Sec also agreed to cover additional nights to attend conference
 - Reimbursement of meals and local transfers will be covered too
 - Modest conference registration fee may apply; Sci Sec currently reviewing if it's possible to exempt Sci Comm members (vs conference running costs)
- ▶ Conference sessions
 - There will be plenary sessions and poster session
 - Programme overseen by Conference Planning Committee (Conf Comm)
 - One session specifically targeting young investigators
 - Exciting new and innovative approaches
 - Aim for gender balance, so think of female speakers
 - Also geographical balance, so think of speakers outside of Europe and US
 - Send proposals to Sci Sec latest by Monday, 12 September cob
 - Sufficient to provide name, and one sentence on why he/she fits the criteria
 - Need not check for availability either, as selection will be done by Conf Comm

5. Setting goals for the next 5 years

- ▶ IRDiRC now about half way through its first phase (2011-2020)
 - At conference, will evaluate progress, how achievable are goals, if to set new goals
 - Initial goals:

- Diagnostics: to diagnose most RD but how to quantify “most” against unknown number of RD?
- Therapies: 200 new therapies seemed ambitious but will now be achieved this year, should it be expanded?
 - For consideration: what about setting a goal to improve access for patients?
- ▶ All members encouraged to read Annexes in the document circulated to select or think of ideas
- ▶ More weights if goals come from the ISC as a whole rather than individual members
 - A one-page primer will be started; aim to circulate by 9 Sept
 - Members to review and provide blitz feedback over the next 72 hours, i.e. by 12 Sept
 - Finalise goal proposals by 13 Sept, and send to Sci Sec

Main action points

- ▶ Contact Chair ADA to verify utility of ADA-M with respect to retrospective consent
- ▶ Share with members manuscript re post-marketing surveillance registry
- ▶ Send proposal of conference speaker names to Sci Sec by 12 Sept
- ▶ Start and send a document re goals for the next 5 years from the ISC
- ▶ Provide feedback to document on goals by 12 Sept
- ▶ Finalise goal proposals and send to Sci Sec by 13 Sept

Document history

Version 1. Report drafted by Lilian Lau, 8 September 2016

Circulated to Hanns Lochmüller, 8 September 2016

Circulated to all members, 9 September 2016