

Meeting report series

Report of the 20th Interdisciplinary Scientific Committee Meeting

Teleconference
November 6, 2017

Participants

Dr Petra Kaufmann, Bethesda, USA (Chair)

Dr Domenica Taruscio, Roma, Italy (Vice Chair)

Ms Gema Chicano, Murcia, Spain

Dr Stephen Groft, Bethesda, USA

Dr Jeffrey Krischer, Tampa, USA

Prof Hanns Lochmüller, Newcastle, UK

Ms Samantha Parker, Paris, France

Dr Marlène Jagut, Scientific Secretariat, Paris, France

Dr Anneliene Jonker, Scientific Secretariat, Paris, France

Dr Lilian Lau, Scientific Secretariat, Paris, France

Apologies

Prof Jack Goldblatt, Perth, Australia

Prof Bartha Maria Knoppers, Montreal, Canada

Dr Angel Carracedo, Santiago de Compostela, Spain

Prof Ken Ishii, Tokyo, Japan

Dr Edmund Jessop, London, UK

Prof Rumen Stefanov, Plovdiv, Bulgaria

Agenda

1. Welcome and introduction
2. ISC membership – document 1
 - a. Results from poll re nominations
3. Action template submission results – document 2
4. Data sharing – document 3
5. Task Force update
 - a. Model Consent Clauses proposal – document 4

- b. International Collaboration on RD Clinical Research Networks
 - c. Patient Engagement for Rare Diseases – document 5
6. Any other business

REPORT

1. Welcome and introduction

The Chair welcomed members who joined the teleconference call, and presented the agenda for the call.

2. Proposal of ISC members

ISC composition

- ▶ Currently 13 members in the ISC
- ▶ In February 2018, mandate of 5 members will end after serving two consecutive terms
- ▶ Ideally, should be composed of 12-15 members, with relevant expertise to interdisciplinary topics and from various geographical areas.
- ▶ ISC members voted for prioritization nominations ahead of the call
- ▶ Nominations with most votes were discussed in order to make a decision regarding new members

Nominees will be approached by the Sci Sec, to ask for their CV and letter of motivation, while keeping the nominating ISC member on copy. Thereafter, these nominations will be send to the OpComm and CA for approval.

→ Sci Sec to reach out to nominees for CV and Letter of Motivation

3. Action template submission results

ISC members have provided feedback and comments to the action template. The ISC Chair and Vice Chair have reviewed these comments, and prioritized them for discussion.

Goal 1 – Theme: Integrate Research into care

- ▶ Connect care to UDNI
- ▶ Train rare diseases experts in research
- ▶ Promote registries

Goal 2 – Theme: Setting the stage for clinical trials

- ▶ Promote high impactful natural history studies
- ▶ Tools, protocol templates, data standards
- ▶ Biomarkers

- ▶ Clinical research infrastructure
- ▶ Non-traditional trial designs (adaptive, platform) and clinical outcome measures– work on this aspect has been done by the Therapies Scientific Committee and their Task Forces, but potential for collaboration

Goal 3 – Theme: assess impact – how do we measure disease impact?

- ▶ Post-approval data collection
 - Task force, publication on methods
- ▶ Impact and burden
 - Task force, publication on methods
- ▶ Training
 - Create online resources
 - Link training programs internationally
- ▶ Sustainability and Health Systems: upcoming theme in Europe

Overarching actions

- ▶ Data sharing – standards clearing house and consent guidelines: Major focus of the ISC
- ▶ Biobanking - standards and ELSI (potential for collaboration with FCC), needs to be tightened in with actions on biomarkers
- ▶ Developing countries – connect with ICORD
- ▶ Patient engagement – patient committee
- ▶ Transitional Care – important, but not IRDiRC mandate
 - IRDiRC - central
 - Provide information (GARD and Orphanet)
 - Enhance outreach (Social media)
- ▶ In progress
 - Promote ADAM
 - Promote PPRL
- ▶ Harmonization of regulatory agencies
 - Better harmonization at approval level, post approval level but also at research points
 - Country to country differences on regulatory perspective – what is allowed with samples and so on
- ▶ Integrate data at per patient level
 - Make data actionable for patients

→ Chairs of the ISC to present these action items in the ISC update at the CA meeting in Tokyo.

4. Data sharing

There has been a longstanding focus of the ISC on data sharing, and this continues to be a top priority. In the week prior to the ISC TC, a call was held to discuss the 2014 project proposal on a Data Sharing Clearing House.

- ▶ Database of standards with use cases
- ▶ Goals to facilitate data sharing, to promulgate standards, to create awareness of standards, and to promote their implementation
- ▶ To create something with continued value (will need updates)
- ▶ Potential issue on how to launch this project, as estimation is that 1 fte is needed for 1 year
 - This has previously been the reason that this was not launched, and a mechanism needs to be found to fund this project

- ▶ Potential products/outcomes
 - Paper on data standards: this will not get to the heart of the issue, as many elements will need to be addressed
 - Training courses on different data standards
 - Ongoing manual or policy document with different data standards, to be updated continuously
 - List on the IRDiRC website with the different standards
 - Website dedicated to this initiative, with standards and use cases, maintained continuously. The use cases element is expected to be the most essential part of this initiative.

The discussion on common data elements will be postponed till next TC.

5. Task Force update

Model Consent Clauses for RD Research

- ▶ Proposal by Bartha Knoppers and Petra Kaufmann
- ▶ Develop RD research consent clauses that are
 - International
 - Interoperable
- ▶ Multi-stakeholder group to conduct workshop
 - Researcher, patient and funder perspectives
 - Clinical, data, legal, ethical and policy expertise
- ▶ Disseminate working with IRDiRC
 - Website
 - Training material
 - Funder to use in calls for application and contract language
- ▶ Will be discussed for approval at CA meeting in Tokyo

Patient Engagement in Research

- ▶ Proposal by TSC and ISC, approved last year
- ▶ Needs to have major drivers from patient advocates
- ▶ Initiations send out and accepted; first leadership call held in the summer
- ▶ Chair selected, possible Co-Chair from patient advocacy group

International Collaboration in RD research

- ▶ Task Force proposal on clinical research networks in rare diseases
- ▶ Idea from previous TSC Chair, now written up by current TSC Vice Chair and FCC Chair, but high involvement expected from ISC
- ▶ Strong support from the ISC
- ▶ Will be discussed for approval at CA meeting in Tokyo

Main action points

- ▶ Contact nominees for CV and Letter of Motivation
- ▶ Present action template and update ISC to CA in Tokyo
- ▶ Present TF proposal Model Consent Clauses for RD Research to CA
- ▶ Discuss common data elements in next TC
- ▶ Organize the next teleconference of the ISC + invited observers