Report of the 19th Interdisciplinary Scientific Committee Meeting

Teleconference
September 18, 2017

Participants

Dr Petra Kaufmann, Bethesda, USA (Chair)
Dr Domenica Taruscio, Roma, Italy (Vice Chair)
Dr Jeanine d'Armiento, Bethesda, USA
Dr Ken Gersing, Bethesda, USA
Prof Jack Goldblatt, Perth, Australia
Prof Bartha Maria Knoppers, Montreal, Canada
Dr Jeffrey Krischer, Tampa, USA
Prof Rumen Stefanov, Plovdiv, Bulgaria

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

Apologies

Dr Angel Carracedo, Santiago de Compostela, Spain
Ms Gema Chicano, Murcia, Spain
Dr Stephen Groft, Bethesda, USA
Prof Ken Ishii, Tokyo, Japan
Dr Edmund Jessop, London, UK
Prof Hanns Lochmüller, Newcastle, UK
Ms Samantha Parker, Paris, France

Agenda

1. Welcome and introduction
2. Proposals of ISC members – *Doc 1*
3. Preparation ahead of Tokyo Consortium Assembly meeting – *Doc 2*
4. Coordination to facilitate data sharing – *Doc 3*
5. Patient Engagement Task Forces – *Doc 4*

REPORT

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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.

Proposals of potential members
- ▶ 4 nominations to date
- ▶ Expertise in informatics or big data knowledge needed, but should also define which specialty as bioinformatics expertise can be very diverse
- ▶ ISC members were asked to reflect on membership further and email in their nominations

- → Submit additional nominations **by October 2** for consideration by the OpComm

For information, nomination procedure:
- ▶ Call for nominations – can be among ISC members, as well as asking CA to provide nominations
- ▶ Select most suitable candidates among nominations received
- ▶ Invite candidates to submit his/her CV and letter of motivation
- ▶ Discuss candidacy with the OpComm
- ▶ If no particular concern, will go to the CA for final vote

3. Preparation ahead of Tokyo Consortium Assembly (CA) meeting

At a recent OpComm, Chairs of SCs and CCs were tasked to identify – together with respective committee members – actions that contribute towards IRDiRC's new goal and vision.

A template has been generated for ISC members to provide ideas and feedback
- ▶ 18 actions have been suggested for the ISC
  - ○ Some were previously discussed at the Paris 2017 meeting
  - ○ List is not definitive nor exhaustive; members are free to propose additional actions
- ▶ Template to be used to seek feedback on methods, tools and metrics for each action
  - ○ Compiled results will effectively provide a new roadmap for the ISC

Using the template provided – explanation of columns:

- ► Proposed actions: list of proposed actions, and members can add if have more ideas
- ► Timeline: indicate time-based priority, e.g. if a proposed action is for immediate action or more long-term action; can also propose time to carry out a proposed action, e.g. in 2018
- ► How: consider what method to implement in order to carry out a proposed action
- ► Tools: propose tools/resources, if available, that can advance a proposed action
- ► Metrics: how to measure the success and/or progress of a proposed action
- ► Task Force: if a Task Force is set up for a proposed action, who should be involved
- ► Any additional comments

- ➔ Each member to fill in the template and return to the Sci Sec by **October 20**
- ➔ Also send template to Chairs of PPRL and ADA TFs for feedback
- ➔ Sci Sec to summarise feedback received and present to ISC Chair and Vice Chair

4. Coordination to facilitate data sharing

4.1 Language on general principles

- ► Funders use general language in funding opportunities
- ► Potential for immediate action if can introduce language that will improve data sharing, e.g. that research projects need to generate data that respect FAIR principles

4.2 Technical update

Data sharing is integral to RD research and there is overwhelming amount of data available, along with data standards and data sharing tools. Instead of creating more new tools or standards, IRDiRC should focused on helping users make informed decision regarding available resources.

Proposed steps to promote good resources and help users make informed decision:

- ► Perform a landscape analysis of available tools and standards
- ► Create a consumable format to "read" the features of the resources, i.e. a matrix of attributes
- ► Convene expert panels to evaluate the features/attributes and complete the matrices
- ► Possibility of users to provide feedback and improve the matrices
- ► Note: will require programming and maintenance support
- ► Note: IRDiRC provides a comparison tool, not dictate which resource to use

Data standards clearinghouse

- ► A proposal was submitted to IRDiRC in summer 2014 but not acted on
  - ○ Time to revisit, given the many (overlapping) data standards available
  - ○ There are some coordinated actions and want to address duplicated efforts
- ► Premise of this clearinghouse:

- - Users can deposit standards and describe it
  - Standards can also be indexed via use cases
  - Will not define new standards but better communicate and promote existing standards

➔ A teleconference to discuss potential TF on this clearinghouse

4.3 Ethical-legal update

A landscape on ethical-legal and consent aspects of data sharing were carried out in collaboration with the P3G, which has covered pretty much the groundwork needed.

A key action arises from this:
- ▶ RD-specific consent clauses are needed
- ▶ IRDiRC members will be solicited contribute clauses specific to RD
- ▶ Will then prepare model RD generic template that typify RD consent form

Supporting this action:
- ▶ Matching fund request
- ▶ In-person meeting to consolidate collected clauses and write up the consent language

➔ Exact request wording to be provided ahead of CA meeting in Tokyo

5. Patient Engagement Task Force

A joint TSC-ISC Task Force on Patient Engagement has been set up. The work on this Task Force is about to start, with invitations of Steering Committee due to be issued shortly.

A briefing document has been prepared
- ▶ It outlines timeline, potential Steering Committee members, and potential Task Force members
- ▶ Initial comments regarding the briefing document
  - On the WSJ article: should not focused on "experts not believing the drug works"
  - To approach with a focus on driving the cost of therapy down
  - Need paradigm on approval of rare diseases therapies
  - Strongly highlight the need for industry-independent, post-marketing registry
- ▶ ISC members were asked to provide feedback, if any, to the Sci Sec

➔ Provide feedback to Patient Engagement TF briefing document

6. Any other business

The NCATS' toolkit for patient-focused therapy development was recently launched. ISC members are encouraged to promote its use in rare diseases research: https://rarediseases.info.nih.gov/toolkit

The next teleconference of the ISC will be organised for the end of October.

Main action points

▶ Send in nominations, with brief bios, of potential ISC members **by October 2**
▶ Complete the charge template **by October 20**
▶ Summarise feedback of the charge template
▶ Discuss potential Task Force on data standards clearinghouse
▶ Provide request wording to IRDiRC for support on RD-specific consent clauses
▶ Provide feedback to the Patient Engagement TF briefing document
▶ Organize the next teleconference of the ISC + invited observers