

Meeting report series

Report of the 17th Interdisciplinary Scientific Committee Meeting

Teleconference
22 May 2017

Participants

Dr Petra Kaufmann, Bethesda, USA (Chair)
Ms Gema Chicano, Murcia, Spain
Dr Stephen Groft, Bethesda, USA
Prof Ken Ishii, Tokyo, Japan
Dr Jeffrey Krischer, Tampa, USA
Prof Hanns Lochmüller, Newcastle, UK
Ms Samantha Parker, Paris, France
Prof Rumen Stefanov, Plovdiv, Bulgaria

Dr Lilian Lau, Scientific Secretariat, Paris, France
Dr Takeya Adachi, AMED, Tokyo, Japan

Apologies

Dr Domenica Taruscio, Roma, Italy (Vice Chair)
Dr Angel Carracedo, Santiago de Compostela, Spain
Prof Jack Goldblatt, Perth, Australia
Dr Edmund Jessop, London, UK
Prof Bartha Maria Knoppers, Montreal, Canada

Agenda

1. Welcome and introduction
2. Operating Committee update
3. Feedback from Task Forces
4. External request
5. ISC membership
6. Coordination to facilitate data sharing
7. Any other business

REPORT

1. Welcome and introduction

The Chair welcomed members who joined the teleconference call, and each participant briefly introduced themselves.

2. Operating Committee update

The new IRDiRC goals have been finalised and articles are currently being written to disseminate them to a larger audience. A concerted dissemination is envisaged, and members will be contacted to assist in this effort:

- ▶ Distribute press release to their network of contacts on the day of papers release
- ▶ Spread the news through social media within their network
- ▶ Present the goals when giving presentation – slides will be provided by the Scientific Secretariat

Timeline of actions is provisional; the dissemination timeline will be revised shortly and members will be notified of when their actions will be requested, together with template text.

3. Feedback from Task Forces

3.1 Automatable Discovery and Access (ADA)

The ADA Task Force has developed a matrix (called ADA-Matrix, ADA-M) to make consent and data use agreements machine readable, to enable discovery and access of such information, and to facilitate data exchange. There is no further update since IRDiRC Scientific Committees meeting in Paris, France, in February 2017. The ADA-M software, together with detailed documentation, is available to download and use; feedback is welcomed.

3.2 Privacy-Preserving Record Linkage (PPRL)

Two aspects are being addressed: ethico-legal recommendations and technical solution, to enable de-duplication of research participants while respecting their privacy, with a (optional) possibility to link back in event of the need to return results/findings to participants. A workshop of the Task Force was held in Paris, France, in December 2016; two primers have been drafted for this meeting. The Task Force will meet next week on a teleconference to advance their work.

It is important to deliver results of this Task Force within a short timeframe given multiple ongoing initiatives could buy-in the clear recommendations and adopt the proposed technical solution. The EUPID system is a secured, third generation solution which has gone one step further on identifiers, opening the possibility of deployment within context of federated international research database.

The ISC should reach out to the European Reference Networks (ERNs) which will be using a software platform for the management of the ERNs. A collaborative effort could see the PPRL system be integrated, thus influencing how research database can be linked while protecting privacy.

4. External request

The ISC were asked to weigh in on an external request to participate in an international rare diseases research project. Her proposal to share data from their research is of relevance to the ISC.

A number of proposed actions following internal Secretariat discussions:

- ▶ Potential invitation to join a Scientific Committee or a Task Force
- ▶ Check if they wish to publish their results (e.g. submit to the Orphanet Journal of RD)
- ▶ If results already published, send a summary to OrphaNews for further dissemination
- ▶ Invite to submit epidemiology data for inclusion in the Orphanet database

5. ISC membership

Upcoming renewal of mandate will be put through to the Consortium Assembly for approval. A few members will rotate off in early 2018 when their mandates come to an end. In order to ensure smooth transition while retaining key expertise, the Chair invited proposals of names of scientific experts who may be good fit in the ISC.

6. Coordination to facilitate data sharing

The ISC should help promote data sharing and one proposed way to do so is by drafting a set of recommendations and a template of core data sharing language. For this initiative:

- ▶ Product: core language that can be shared via IRDiRC website or through a published article
- ▶ Content: “what do we do upstream to enable downstream data sharing”, e.g. to have the right consent, data use agreement, data standards, etc.

ISC members were requested to send to the Chair any data sharing language that they have access to, e.g. from different programs members are involved in, and the information received will be compiled in order to identify a set of core module.

Future step: consider setting up a Task Force with the involvement of the TSC to work on harmonisation language for therapeutic regulatory purposes.

7. Any other business

The NCATS’ Global Rare Diseases Patient Registry Data Repository (GRDR) is being refreshed in order to align to other registry initiatives as well as progress in informatics and smart use of data. Interested members should contact the Chair.