

Meeting report series

Report of the 3rd ISC Working Group on Registries and Natural History teleconference

9 July 2014

Organization

Organized by: IRDiRC Scientific Secretariat
Teleconference

Participants

Dr Domenica Taruscio, Rome, Italy (Chair)
Ms Liz Donohue Wheeler, Sanford Research, USA (Co-Chair)
Dr Veronica Alonso (representing Manuel Posada), Madrid, Spain
Dr Anna Ambrosini, Milan, Italy
Mr Kyle Brown, San Mateo, USA
Dr Joshua Burns, Sydney, Australia
Dr Sabina Gainotti, Rome, Italy
Ms H el ene Moussu (representing Virginie Picard), Paris, France
Ms Samantha Parker, Puteaux la D efense, France
Dr Yaffa Rubinstein, Bethesda, USA
Dr Rumen Stefanov, Plovdiv, Bulgaria

Dr Barbara Cagniard, Scientific Secretariat
Dr Sophie H ohn, Scientific Secretariat

Apologies

Prof Odile Boespflug-Tanguy, Paris, France
Dr Jan Friedman, Columbia, Canada
Dr Stephen Groft, Bethesda, USA
Dr Micheil Innes, Calgary, Canada
Ms Anna Kole, Paris, France
Prof Eugenio Mercuri, Rome, Italy
Prof Marshall Summar, Washington, USA
Prof Bernard Zabel, Freiburg, Germany
Dr Cornelia Zeidler, Hannover, Germany

Agenda

- Introductions of new working group (WG) members
- IRDiRC Conference
- ISC Roadmap
- The JRC platform on registries
- Core implementation WG of RD-Connect
- Next steps

REPORT

Introduction of new WG members

Stephen Groft recently joined this WG but was not able to attend this teleconference. H  l  ne Moussu, project manager of the French muscular dystrophy association, attended for the first time this teleconference as she was replacing Virginie Picard.

IRDiRC Conference

The second IRDiRC Conference will take place in Shenzhen, China, on 7-9 November 2014. Lysogene and NCATS will be represented.

Interdisciplinary Scientific Committee (ISC) Roadmap

The ISC Roadmap was sent to the WG members prior to the teleconference.

Four WGs are under the umbrella of the ISC: the WG on Data Sharing and Bioinformatics, the WG on Ethics and Governance, the WG on Registries and Natural History, and the WG on Biobanks. This roadmap was discussed during the ISC meeting in Montr  al and shows future actions for 2014, 2015 and 2016.

The WG on Data Sharing and Bioinformatics proposed to create a Clearinghouse for Data Standards. They prepared a paper about this proposition which was discussed and accepted during the Executive Committee (EC) that took place in Berlin in May. They are now looking for funds in order to progress with this activity. The first version of the Clearinghouse should be available in 2015, and the final version in 2016.

The WG on Ethics and Governance will produce an International Code of Conduct for Genomics and Clinical Data Sharing as well as Consent Template Clauses. Seed funding is required.

The WG on Registries and Natural History will pilot unique rare disease patient identifiers to enable linking different datasets from the same patient across institutions and countries. This a key point for building registries. It will also need to be linked with biobanks and OMIM. No additional funds would be required.

The WG on Biobanks overlaps with the WG on Registries on several points. However, they will not be mixed in order to discuss their specific topics. Nevertheless, common sessions could be scheduled. The WG on Biobanks will propose standards for biobanks and develop a catalogue and scoring system for biomaterial quality in biobanks.

The Joint Research Centre (JRC) platform on registries

The European Commission decided to build a platform for registries at the JRC. They established a formal agreement that the JRC will be in charge of the governance of all registries in Europe. It would be a good idea to invite them in this WG in order to not duplicate the work.

Core implementation WG of RD-Connect

A list of tasks was sent to the potential participants that they should accept in order to be member of the RD-Connect WG. 16 registries and 3 networks of biobanks accepted and the WG was established in June 2014. The registries involved have been used for many years in the field of rare diseases.

Their starting point was the creation of a profile in the RD-Connect catalogue (a tool which will be soon available to the research community) named “identity card”. This catalogue gives basic information on the content of registries and biobanks (people and institutions involved, number of patients included, standards for data collection and management, procedures to access data). Through this catalogue, researchers will be aware of how many patients for one rare disease are registered into several registries. This catalogue is a first step towards data sharing. The WG members have also provided the documents they use as standards operating procedures, study protocols and format consent templates. Next steps will be the testing of the facilities provided by the online catalogue as well as the guidelines on informed consent made for RD-Connect.

Next steps

- The Global unique identifier (GUID) is important and it would be interesting to see how other groups have implemented it.
- Share information on the different initiatives in the domain of registries and realize a policy document.
- Send inputs to structure the future activities of this WG to help reaching IRDiRC objectives.
- Update the gap analysis document and create subgroups regarding the topics found in this document to work in parallel.

Main deliverables

- Invite the JRC members to join this WG
- Send inputs to structure the future activities of this WG
- Update the gap analysis document
- Schedule the next teleconference in September