IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Meeting report series

Report of the 3rd ISC WG on Ethics and Governance teleconference

3 July 2014

Organization

Organized by: IRDiRC Scientific Secretariat
Teleconference

Participants

Prof Bartha Maria Knoppers, Montreal, Canada, co-chair
Ms Megan Fookes, Sydney, Australia
Mrs. Jasjote Grewal, Hanover, Germany
Prof Ingrid Holm, Boston, USA
Prof Matthias Kretzler, Ann Arbor, USA
Mrs Marie-Christine Ouillade, Paris, France
Mrs Minh Thu Nguyen, Montreal, Canada
Dr Simon Woods, Newcastle, UK

Dr Barbara Cagniard, Scientific Secretariat
Dr Lilian Lau, Scientific Secretariat

Apologies

Prof Jack Goldblatt, Perth, Australia, co-chair
Prof Nils Hoppe, Hanover, Germany
Dr Tsveta Schyns, Austria
Dr David Towndend, Maastricht, the Netherlands
Kurt Zatloukal, Graz, Austria

Agenda

1. Summary and update of last WG teleconference
2. Interdisciplinary Scientific Committee/IRDiRC roadmap and feedback to WG
3. Other topics

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

REPORT

Summary and update of last WG teleconference

This WG did not convene since August last year as the Interdisciplinary Scientific Committee (ISC) decided to submit its roadmap to the IRDiRC Executive Committee approval before resuming the activity of the WGs.

An update of most of the topics discussed at the last teleconference was provided.

Global Alliance for Genomics and Health (GA4GH)

GA4GH is an entity gathering around 200 candidate members (institutions, centers, university, funders, patient organizations, and diverse companies) over 40 countries. The purpose is to facilitate the sharing of genomic and health-related data for biomedical research to ensure further progress in research.
A steering Committee was created and is now working on a constitution to be signed by institutions wishing to become a member.

European Data Protection directive

The document went through the parliament and the Commissioner has reassured the biomedical research community that the research exemption might be reinstated in the document.

Creation of a questionnaire to capture the sensitivity of RD patients and families about data sharing

RD-Connect had a series of group discussion with patient representatives (focus group) in collaboration with Eurordis. Analysis of these discussions is ongoing and results should be published in the format of a report later this year. Focus group is supportive of data sharing but displays anxiety about the security of data sharing and reports issues around presence or absence of consent. A Delphi's study will be conducted next year, including patient associations and professionals.
WG suggested that the study should be expanded to other part of the world such as the Asia-Pacific area and not limited to Europe.

Another initiative is the Australian Rare Diseases Survey for Adults, launched in July 2014, by the Office for Population Health Genomics, Department of Health WA in partnership with Rare Voices Australia, The Genetic and Rare Disease Network, Genetic Support Network Victoria and the Association of Genetic Support Australasia.

The Electronic Medical Records and Genomics (eMERGE) Network is developing a large survey to obtain the perspective of patients of the network institutes on topics such as broad data sharing, broad consent, etc. The survey should be released in 6-9 months. The diffusion of this survey through the Rare

Diseases Clinical Research Network (RDCRN) patient registry was previously discussed between leaders of both initiatives. This would allow the comparison of perspective of rare diseases patients vs. general patient population.

- ⇨ WG would like to be kept informed of the results of all initiatives as it will be precious information to develop policies integrating patient perspective.
- ⇨ Interdisciplinary Scientific Committee would like a 2-3 pages synthesis of all the initiatives (including work from Mats Hansson) with the latest data.

Creation of the International Policy Interoperability and Data Access Clearinghouse (IPAC)

IPAC was launched in fall 2013 and provide since January 2014 a database of generic clauses for consent forms and generic agreement (data access, genetic data transfer, etc.). IPAC is producing baseline products for researchers to customize and use. There is no section for rare diseases at the moment. This could be part of the consent work (see later section).

Funding of oversight committee to monitor all the guidelines and assure the sustainability of databases

The IRDiRC Scientific Secretariat recently started to collect guidelines and procedures framing rare diseases. The list of gathered guidelines will be circulated to the WG for feedback.

Interdisciplinary Scientific Committee/IRDiRC roadmap and feedback to WG

The ISC generated a roadmap summarizing funding priorities and tasks to be conducted by each of the four ISC WGs for the next 3 years. A fifth WG - on Patient Involvement and Societal Outreach - was included in the ISC roadmap but the WG was not yet created.

The Interdisciplinary and Diagnostics Scientific Committee then combined their respective roadmap defining standards, tools and position statements to be developped by the WGs that were divided in function of the goals and core activities of IRDiRC:

- Diagnose Most rare Diseases by 2020
  - Phenotype and Genotype Data Generation
  - Data Interpretation for Gene Discovery
  - Core Infrastructure
- 200 New Therapies by 2020

The Therapies Scientific Committee's roadmap will also be integrated once finalized.

Three deliverables for the WG on Ethics and Governance are listed on the roadmap.

Consent template clauses

A proposal for funding was presented to the Executive Committee for this task. The purpose is to develop template consent clauses specific to rare diseases for the researchers. Topics would include international data sharing, international samples sharing, pediatrics return of results, etc.
Consent clauses will be available on the IRDiRC and IPAC website.
Once the proposal will be funded, WG members will be contacted for examples of consent forms (RD-Connect, Genethon, etc.), and comments and validation of the newly developed clauses.

International Data Safe Havens and e-health Consents

The purpose is to provide machine readable consent to optimize data sharing including cloud computing projects across countries and jurisdictions. Types of attributes necessary to be considered to be a safe haven should be defined to be equivalent to attributes that ethic committees, access committees, security and privacy commissions would consider as substantially equivalent to the level of protection that they are legally required to offer in their countries.
The project is not yet completely defined as it is planned for 2015-2016.

There are two current consultations in UK to establish data safe haven: one by the Department of Health - Protecting Health and Care Information: a consultation on proposals to introduce new Regulations - and one by the Health and Social Care Information Centre.

International framework for Genomics and Clinical Data Sharing

The 7th draft of the 'Framework for Responsible Sharing of Genomic and Health-Related Data' was developed under the auspices of the Global Alliance for Genomics and Health. Ideally, this framework should be referenced in their constitution.
The word 'Code of conduct' was replaced by the term 'framework' to indicate that it includes principles for governance and is not a law as the word 'code of conduct' suggests in some countries.
The next version of the document will be released on 18 October 2014, the day before the ASHG meeting.
A series of specific polices – security, privacy, consent, governance – will be then prepared, with the participation of IRDiRC.

Comments on the 7th draft:

- 'Preamble' is unique, based on an approach of Human Rights complementary to bioethics. It is based on 3 rights of the 1948 'Universal Declaration of Human Rights': right to benefit from advances in research, right to proper attribution, and right to scientific freedom.
- The field of application is extremely broad to include as many entity types as possible. Any entities missing in the list?
- 'Foundation Principles' are general, short and translated into core elements in the following section.
- 'Core Elements of Responsible Data Sharing'. There is still discussion on the sentence "This Framework applies to data that has been approved for use by competent bodies in compliance

with national and international laws and that respects restrictions on downstream uses". Downstream uses will depend on restrictions to consent in each country.

Other topics

- Next teleconferences of this WG will be 1-hour in duration.
- The European Bank for Induced Pluripotent Stem Cells (EBiSC) project (http://www.ebisc.org/) was mentioned in the 7th meeting of the ISC. EBiSC is designed to address the increasing demand by induced Pluripotent Stem Cell (iPSC) researchers for quality-controlled, disease-relevant research grade iPSC lines, data and cell services. Its goal is to demonstrate an operational banking and distribution service of iPSC lines after 3 years and to establish subsequently for Europe a centralized, not-for-profit bank providing all qualified users with access to scalable, cost-efficient and customized products. This project is interesting as similar to what IRDiRC wish to accomplish. Should IRDiRC get involved?
- The NIH Undiagnosed diseases program, launched in 2006, is now expanding into a network of six institutes. There is an ethics component of the network that is mostly focused on the return on results.

Main deliverables

- Send comments in bullet-point on the 7th draft of the 'Framework for Responsible Sharing of Genomic and Health-Related Data'.
- Provide the Scientific Secretariat with sources of surveys, etc. for synthesis.
- Send information on the UK consultation initiatives on data safe havens to the Scientific Secretariat for circulation to WG members.