

## Meeting report series

### Report of the 21<sup>st</sup> Interdisciplinary Scientific Committee Meeting

Teleconference

April 23, 2018

#### Participants

Dr Petra Kaufmann, San Diego, USA (Chair Interdisciplinary Scientific Committee (ISC))

Dr Domenica Taruscio, Roma, Italy (Vice Chair)

Dr Dixie Baker, Los Angeles, USA

Dr Stephen Groft, Bethesda, USA

Dr Melissa Haendel, Portland, USA

Dr Elizabeth McNeil, Cambridge, USA

Prof Hanns Lochmüller, Freiburg, Germany

Mr David van Enckevort, Groningen, The Netherlands

Dr Marlène Jagut, Scientific Secretariat, Paris, France

Dr Anneliëne Jonker, Scientific Secretariat, Paris, France

Dr Lilian Lau, Scientific Secretariat, Paris, France

#### Apologies

Ms Gema Chicano, Murcia, Spain

Dr Angel Carracedo, Santiago de Compostela, Spain

Prof Ken Ishii, Tokyo, Japan

Dr Edmund Jessop, London, UK

Prof Matt Might, Birmingham, USA

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#### Agenda

1. Welcome and introduction
2. Introduction new ISC members
3. Task Forces
  - a. IRDiRC – GA4GH TF Model Consent Clauses for Rare Diseases
  - b. Clinical Research Networks for Rare Diseases proposal – [document 1](#)
4. Topics for discussion at ISC Vienna meeting

## REPORT

### 1. Welcome and introduction

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

### 2. Introduction new ISC members

Representatives present on the call were asked to introduce themselves shortly and to indicate their history with IRDiRC.

Dixie Baker

- ▶ Senior Partner with Martin, Blanck & Associates
- ▶ Involved in health information technology, electronic health records, privacy and security technology, and the sharing and protection of genomic data
- ▶ Co-Chair of the Security Working Group of the Global Alliance for Genomics and Health
- ▶ Recently joined the ISC

David van Enckevort

- ▶ Working at the Genomic Coordination Centre as Technical Project Lead at the University Medical Centre Groningen
- ▶ Involved in development of software for genetic data interpretation
- ▶ Involved in several European projects, such as RD-Connect, Solve-RD and the future European Joint Program for Rare Diseases
- ▶ Recently joined the ISC

Stephen Groft

- ▶ Senior advisor, National Center for Advancing Translational Sciences (NCATS), NIH
- ▶ Former Director Office of Rare Diseases, NCATS, NIH
- ▶ Has been working on rare diseases for over 40 years
- ▶ Involved in IRDiRC in different roles since its start

Melissa Haendel

- ▶ Director of the Ontology Development Group at Oregon Health and Science University (OHSU)
- ▶ Associate Professor at OHSU
- ▶ Formerly a bench scientist, now involved in the development of ontologies and data standards
- ▶ Member of the Data and Clinical Working Groups of the Global Alliance for Genomics and Health
- ▶ Recently joined the ISC, but previously been involved in one of IRDiRC's Working Groups

Petra Kaufmann

- ▶ Chair of the ISC
- ▶ Vice President of R&D Translational Medicine, AveXis

- ▶ Former Director Office of Rare Diseases, NCATS, NIH and former Director Division of Clinical Innovation, NCATS, NIH

#### Hanns Lochmüller

- ▶ Formerly Professor of Experimental Myology, University of Newcastle
- ▶ Former scientific coordinator of the RD-Connect project in 2012-2017
- ▶ Currently attached as researcher to the University of Freiburg, in the department of neuropediatrics and muscle disorders
- ▶ Involved in IRDiRC since the start of the Scientific Committees

#### Elizabeth McNeil

- ▶ Senior Director of Clinical Development, BlueBird Bio
- ▶ Works on a project to develop therapy for adrenoleukodystrophy
- ▶ Formerly a member of the Therapies Scientific Committee, now back at IRDiRC and recently joined the ISC

#### Domenica Taruscio

- ▶ Vice Chair of the ISC
- ▶ Director of the National Center for Rare Diseases, Istituto Superiore di Sanità (ISS)
- ▶ Leader of the Workpackage “Databases and Registries” in RD-Connect, and actively involved in linking rare disease patient registries and development of the Registry & Biobank Finder
- ▶ Involved in IRDiRC since the start of the Consortium and of the Scientific Committees

### 3. Task Forces

#### 3.1 Model Consent Clauses for Rare Diseases

The overarching purpose of this Task Force is to gather international policy researchers in rare diseases to address specific consent needs. The objectives of the Task Force:

- ▶ Develop a series of model consent clauses for rare disease researchers based on robust bioethical and legal approaches, addressing the complexity of the scientific, ethical and legal issues that arise when conducting rare disease research
- ▶ Address issues such as participant privacy, use of identifiable images, and return of results to participants
- ▶ Enable rare disease researchers to use clauses specific to their research context and participant populations, consequently assisting in the customization of their own consent documents

Task Force will have a series of teleconferences, followed by a face-to-face meeting in September 2018. This Task Forces, is as all IRDiRC Task Forces, composed of multi-stakeholders, such as lawyers, ethicists, expert patients, patient representatives, researchers, health policy advisors, and industry representatives from around the world.

First steps in the Task Force has been to define a set of core elements that are indispensable for model consent clauses:

- ▶ Objective
- ▶ Return of results
- ▶ Matchmaking databases (opt-in/opt-out?)
- ▶ Ongoing analysis/research (indefinite use of data/samples)
- ▶ Limiting research to the "cause of the disease" only (exclude other results)
- ▶ Participation in registries
- ▶ Commercialization/for profit
- ▶ International data/sample sharing
- ▶ Withdrawal mechanisms
- ▶ Facial imaging
- ▶ Identifiability
- ▶ familial/pedigree consenting
- ▶ De-duplication
- ▶ Re-contact/re-consenting
- ▶ Patient responsibility to maintain contact (update coordinates and contact info)
- ▶ Specific clauses for pediatric/incompetent adults

The Task Force works now on verifying what is available regarding these elements, and determining request for these specific elements.

Suggested ideas for the Task Force to keep in mind are:

- ▶ To look at the new *General Data Protection Regulation (GDPR)* law very carefully, and determine its potential impact on research, not yet from a retro-active approach
- ▶ Consider publishing consent elements on IRDiRC website, thereby rating samples of consent model language that is considered as really useful
- ▶ What are the implications for consent on different data repositories, a centralized data repository versus a federated repository? Where does the data live and how will it be used?
- ▶ To bring synergy between company and academic research

The ISC will be kept informed of the ongoing discussions and next steps in the Task Force.

### **3.2 Clinical Research Networks for Rare Diseases proposal**

Worldwide, there are several Clinical Research Networks for Rare Diseases, such as the Clinical Research Networks in the US, the ERN in Europe and the research networks in Japan.

- ▶ The objective of the Task Force on Clinical Research Networks for Rare Diseases will be to develop recommendations on guiding principles for national/supra-national policies on clinical research networks within an international context for collaboration and interoperability, and the related funding recommendations.
- ▶ Will be built on the experience gained and ongoing initiatives in the US and EU, but also Australia, Japan and other countries willing to take an active part in identifying policy recommendations as

well as recommendations to funders in support of the development and adoption of new diagnostic tools and new therapies for rare diseases

- ▶ Task Force that will be joined by IRDiRC Funders Constituent Committee and IRDiRC Therapies Scientific Committee

Comments on the Task Force proposal are:

- ▶ How to best integrate the different perspectives of the clinical research networks; patient care, research, clinical discovery, etc
- ▶ There are many variables in existing research sites. Attempting to create an interoperable research network will require several considerations if we are to reach the patients and health care providers in need and many times this community extends beyond active research networks/consortia to the community level. For many patients travel to a distant research site can be problematic due to the manifestations of their disease and the need for more local research sites to improve recruitment, retention, and patient participation.
  - Identifying and Qualifications of Research-Naïve Institutions will require special considerations of necessary training programs for investigators and staff, adherence to Good Clinical Practices, Code of Federal Regulations and IRBs in USA for research investigations, participation in recruitment outreach activities, and gaining sufficient resources to initiate and sustain the research sites.
  - Consideration of existing infrastructure to develop and adhere to study protocol with attention to local collaborative hospitals, access to advocacy groups and patients without advocacy groups, and referral procedures from partners.
  - Support development of information on planned and existing clinical trials to be included in ClinicalTrials.gov and other searchable databases to expand access to this information.
  - Develop Clinical Trials Training Modules for investigators, health care providers, patients, families, and caregivers
  - Emphasize methods to identify and interact with patients not represented by an organized patient advocacy groups including social media and health care systems.
  - Identify resources and services to provide travel to local and distant research sites.
- ▶ The European Reference Networks have just celebrated their first birthday. How can this experience be shared and improved upon?
- ▶ Expand organizations identified on list of potential groups to participate in networks to include:
  - H3Africa
  - Undiagnosed Diseases Network in USA
  - Undiagnosed Diseases Network International (UDNI, <http://www.udninternational.org>)
- ▶ A part of this Task Force should be dedicated to patients
  - Approach to patients
  - How to build registries that support patient engagement?

#### **4. Topics for discussion at ISC Vienna meeting**

Several topics have been suggested for the meeting of the ISC in Vienna:

- ▶ Topic dedicated to repositories and databases

- Specifically, to the sustainability of databases
- Specifically, to the collaboration of existing databases, to push them to think beyond building collaboration
- ▶ Topic dedicated to social media and rare diseases
  - Social media influences more and more in our lives
  - Time to do a comprehensive review of possibilities of social media in rare diseases and rare diseases research
- ▶ Topic dedicated to data sharing
  - How do we define data sharing for informatics purposes?

In addition, several topics were suggested for the Joint Scientific Committees meeting in Vienna, that will follow the ISC meeting:

- ▶ Interlink with different Task Forces
  - How to link the Task Forces and their outcomes more efficiently?
- ▶ IRDiRC Recognized Resources
  - IRDiRC Recognized Resources exists since a few years, and an article has been published in *EJHG*
  - Time to bring it more to the attention
  - Time to think about the sustainability of the label

### **Main action points**

- ▶ Send suggestions for the Model Consent Clauses Task Force to Sci Sec
- ▶ Send suggestions for the Clinical Research Network for Rare Diseases to Sci Sec
- ▶ Send suggestions for ISC Vienna Agenda to Sci Sec

### **Document history**

Version 1. Report drafted by Anneliene Jonker, April 26, 2018

Circulated to Chair and Vice Chair of the ISC, April 26, 2018

Version 2. Report edited by Domenica Taruscio, May 3, 2018

Circulated to all members, May 4, 2018