IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Meeting report series

Report of the 7th Interdisciplinary Scientific Committee (ISC) Meeting

Charité-Campus Virchow-Klinikum, Berlin, Germany
8 May 2014

Participants

Prof Hanns Lochmüller, Newcastle, UK (ISC Chair)
Dr Kym Boycott, Ottawa, Canada (DSC Chair)
Dr Angel Carracedo, Santiago de Compostela, Spain
Prof Jamel Chelly, Paris, France
Prof Jack Goldblatt, Perth, Australia
Dr Stephen Groft, Bethesda, USA
Dr Adam Heathfield, Sandwich, UK (TSC member)
Dr Petra Kaufmann, Bethesda, USA
Dr Jeffrey Krischer, Tampa, USA
Mrs Samantha Parker, Paris, France
Prof Peter Propping, Bonn, Germany (DSC member)
Prof Rumen Stefanov, Plovdiv, Bulgaria
Prof Josep Torrent iFarnell, Barcelona, Spain (TSC member, replacing the TSC Chair)

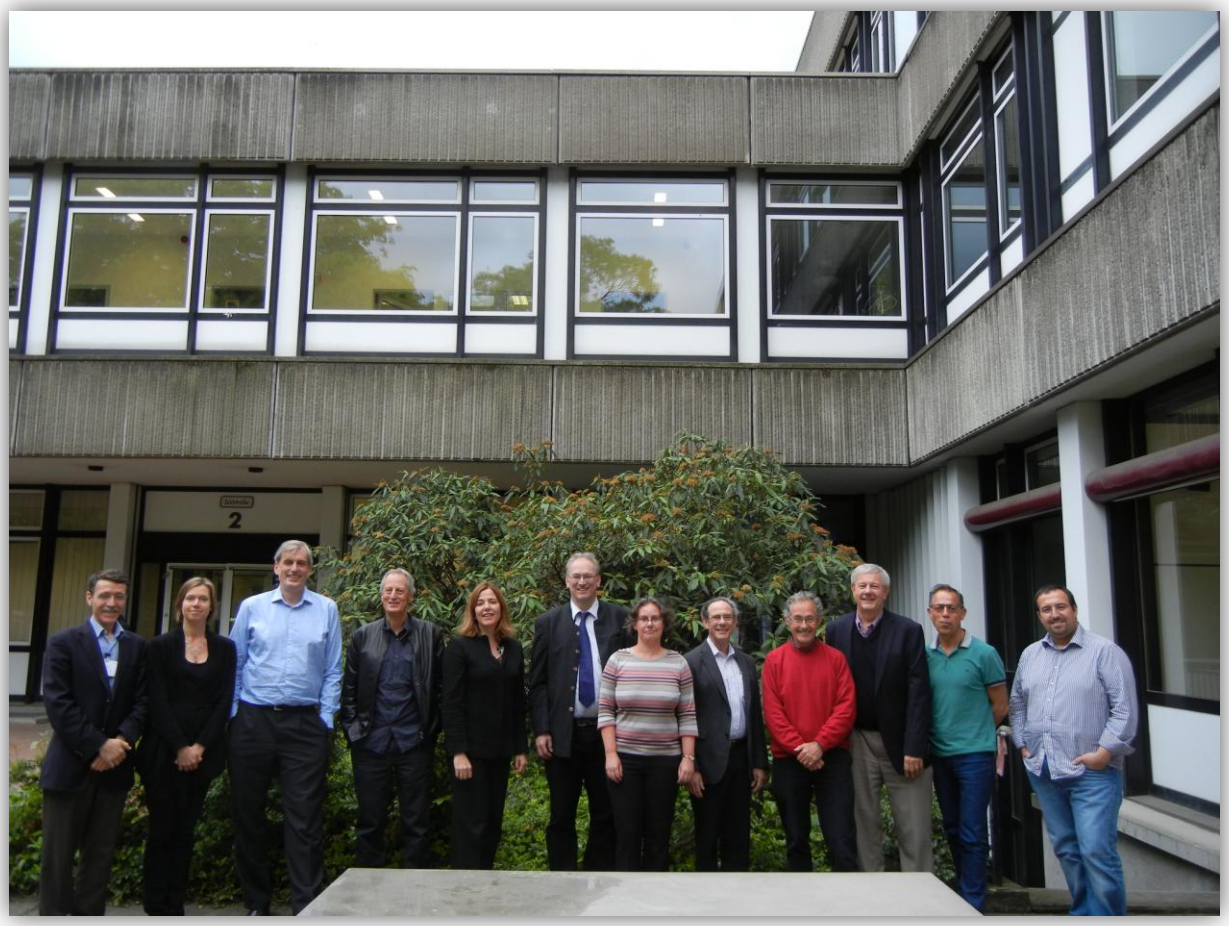
Dr Barbara Cagniard, IRDiRCScientificSecretariat, France
Dr Sophie Höhn, ScientificSecretariat, France

Apologies

Mr Alastair Kent, London, UK
Prof Bartha Maria Knoppers, Montreal, Canada
Dr Domenica Taruscio, Roma, Italy

Agenda

- Introduction
- IRDiRC Roadmap
- ISC Working Groups
- Cross-cutting work and interactions between Working Groups and Scientific Committees
- IRDiRC 2nd Conference in Shenzhen, China
- Next meetings
- Other topics



REPORT

Introduction

This Interdisciplinary Scientific Committee (ISC) Meeting was organized in conjunction with the European Conference on Rare Diseases and Orphan Products which took place on 8-10 May 2014 in Berlin, Germany. Thanks to this, some members of the two other Scientific Committees were also present to the ISC meeting.
All the present members introduced themselves.

IRDiRC Roadmap

The main topic of the latest Executive Committee (EC) meeting that took place on 7-8 May 2014 in Berlin, Germany, was the feedback from the EC on the roadmap elaborated by the Scientific Committees with the inputs of their Working Groups.
The three Scientific Committees have worked on different paths. The Therapies Scientific Committee (TSC) will need more time to finish its road map. Consequently, particular focus was devoted to the Diagnostics Scientific Committee (DSC) and ISC roadmaps which were merged into one. The shorter term objectives of this common roadmap were named "Milestones" and the longer term goals represent the funding objectives. During this presentation, the ISC had the opportunity to ask questions and identify areas of common interest. The TSC recommendations were also presented.

ISC Working Groups

For the past 6 months, most of the WGs of the ISC were inactive, awaiting for feedback from the Scientific Committees and the Executive Committee regarding their roadmap contributions. This is now time to schedule the next teleconferences of the WGs.

The WG on Registries and natural history may need to be divided at some point into one WG on Registries and one WG on Natural history if they continue to grow.

Cross-cutting work and interactions between Working Groups and Scientific Committees

Contact people from the 3 Scientific Committees will need to be nominated for facilitating these interactions. They will attend to the WG/SC teleconferences and read/share the required documentation.

The following interactions could happen in the future:

- WG on Model systems (DSC) – WG on Biomarkers for disease progression and therapy response (TSC)
- WG on Data sharing and bioinformatics (ISC) – WG on Sequencing (DSC)
- WG on Biobanks (ISC) – TSC
- WG on Registries and natural history (ISC) – TSC

The WG on Model systems will report to the DSC and the TSC.

IRDiRC 2nd Conference in Shenzhen, China

The second conference organized by the IRDiRC in collaboration with the BGI is gathering top scientists from Europe, North America and Asia for dynamic exchanges on knowledge and expertise. It will be held on 7-9 November 2014, with two days of conference - where four parallel tracks will take place in the topics of Diagnostics, Interdisciplinary-Technologies, Therapies, and Educational - and one day of training course.

The program is currently composed as follows:

- Plenary Session 1: Opening Session
- Rare Disease Research in 2014 – an overview
  - Genomics: achievements and challenges
  - Therapies for rare diseases: achievements and challenges
  - Role of patient organization in R&D
  - An universal platform for rare disease in China: from omics research to patient care
- Track 1: Diagnostics
  - Sequencing and other omics technologies
  - Interpretation of sequence data
  - Impact on Diagnosing Rare Diseases on Patients and on the Healthcare System
  - Sharing Information on Rare Diseases
- Track 2: Interdisciplinary – Technologies
  - Registries and biobanks
  - National Plans and Policies on Rare Diseases
  - Patient Involvement and Ethics in Rare Disease Research
  - Health Technology Assessment and Access to Orphan Drugs
- Track 3: Therapies
  - Gene / Cell therapy
  - A View of Therapeutic Development from the Industry
  - Regulatory Challenges for Drug Development in Rare Diseases
  - Clinical Trials: Challenges and Possibilities
- Track 4: Educational
  - Principle of Genetic & Genomic Medicine I
  - Principle of Genetic & Genomic Medicine II
  - Principle of Genetic & Genomic Medicine III
  - Principle of Genetic & Genomic Medicine IV

No additional international speaker is required. China proposed a list of Chinese speakers which will be prioritized.

Next meetings

The next joint face-to-face meeting of the 3 SCs will probably take place in conjunction with the ESHG meeting (6-9 June 2015) that will be held in Glasgow, United Kingdom, unless there would be an IRDiRC Conference in 2015. A joint meeting with the DSC and the TSC could be organized at this occasion.

Other topics

The EBiSC project (http://www.ebisc.org/) was mentioned by Adam Heathfield. 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. Rare diseases may not be dominant in this project but the IRDiRC could push for it.