IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Meeting report series

Report of the 12th Therapies Scientific Committee Teleconference

Teleconference
October 26, 2018

Participants

Dr Diego Ardigo, Parma, Italy (Chair)
Dr Virginie Hivert, Paris, France (Vice Chair)
Dr Seng H. Cheng, Framingham, USA
Dr Robin Conwit, Bethesda, USA
Dr Michela Gabaldo, Milan, Italy
Dr Anne Pariser, Bethesda, USA
Dr Maurizio Scarpa, Wiesbaden, Germany

Ms Christine Cutillo, Bethesda, USA
Dr Anneliene Jonker, Paris, France

Apologies

Prof Annemieke Aartsma-Rus, Leiden, The Netherlands
Dr Adam Heathfield, Sandwich, UK
Dr Sandrine Marreaud, Brussels, Belgium
Dr Akifumi Matsuyama, Osaka, Japan
Mr Yann Le Cam, Paris, France
Dr Karin Rademaker, Utrecht, the Netherlands
Prof Josep Torrent i Farnell, Barcelona, Spain

Agenda

1. Welcome
2. Introduction new TSC members
    a. Anne Pariser
    b. Maurizio Scarpa
3. Updates on ongoing Task Forces
    a. PCOM publication

4. Updates on new Task Forces
    a. Patient Engagement – *Doc 1*
    b. Clinical Research Network
5. Brainstorming and proposals for the strategic plan based on new goals – *Doc 2*
    a. Results gap-mapping exercise

REPORT

1. Welcome and introduction

The Chair welcomed all participants of the teleconference call and explained that the objective of the call was to provide an overview of ongoing activities and to continue on the brainstorming for strategic plans.

2. Introduction new TSC members

The new members were welcomed to the TSC, and shortly introduced themselves

Anne Pariser

- Deputy Director at the Office of Rare Diseases Research (ORDR), NCATS, NIH
- Previously worked at FDA

Maurizio Scarpa

- Paediatrician, working on rare diseases
- Coordinator European Reference Network on Hereditary Metabolic Diseases
- Chair to the board of the European Reference Network Coordinators
- Director of the Gene Transfer Laboratory and Lysosomal Diagnostic Unit

3. Updates on ongoing Task Forces

Patient-Centred Outcome Measures Task Force had a Workshop in late 2015

- Recommendations resulted from this Task Force were published March 2016
- Thomas Morel, member of this Task Force, wrote a commentary based on these recommendations, including both recommendations and his personal perspective
- Article accepted by Orphanet Journal of Rare Diseases
- When published, will be send to TSC members

4. Updates on new Task Forces

- Patient Engagement in Rare Diseases Research
  - Two focus points to tackle through best practices and guiding principles
    - Patient-industry partnership in medical product development
    - Patient engagement to improve quality of research
  - Official invitations send by end of the month *[post-meeting note: invitations are send]*

- The Scientific Secretariat (Sci Sec) will start preparing the background document, also based on discussion with the Task Force Steering Committee
- The Task Force workshop is expected to take place in Spring 2018, and will hopefully result in recommendations and a paper

- Clinical Research Network
  - Not yet formally approved by the IRDiRC Consortium Assembly (CA), proposal will be discussed by the CA in Tokyo
  - Basic premise: take existing initiatives (e.g. US's Rare Disease Clinical Research Network, the newly launched European Reference Networks in the EU, etc) and use their experience to identify common features, build common objectives and develop global framework to facilitate clinical research
  - Identify key people in every geographical region, thereby mapping the potential for more collaborations, and to maximize the potential from these networks

5. Brainstorming and proposals for the strategic plan based on new goals

In order to create new strategic plans to achieve the new goals, a general framework has been created for the generation of the TSC strategy.

- Framework
  - to identify the gaps potentially preventing the achievement of the 1000 drugs
  - to identify key stakeholders
  - to identify target behaviour of stakeholders
  - → to set up a strategic action plan
- Members previously asked what the gaps are
  - from a technical standpoint?
  - from legal/ regulatory standpoint?
  - from a political standpoint?
  - from a society standpoint?
  - from other standpoints?

- Great response from members on the different gaps to overcome.
  - Great source to identify points that are crucial
  - Time to prioritize the different ideas, in order for key gaps to be addressed first
  - Members are asked to judge all gaps by October 27
    - its potential impact, i.e. how much positive impact is expected when the gap is overcome
    - its manageability, i.e. how much the gap can be tackled by IRDiRC and its members
- Some trends could be identified across different gaps

- Task Forces are still a useful instrument, but probably not the only one (e.g. workshops)
- Follow-up of previous TFs represents a major opportunity
  - Design and feasibility of clinical trials in RDs: create a roadmap on how to conduct clinical trials in RD
  - Definition of a business model for repurposing: continuation on creating opportunities after the previous Task Force
  - Continuation of small population clinical trials: uptake of new methodologies, determination of feasibility
- Importance of disease understanding and research networks (Task Force proposal up for approval)
- Definition of a specific development and access framework for advanced therapies
- Price trends and access to orphan drugs
- Data use and sharing (in combination with other committees)

- Essential points in the strategic work plan for the TSC are:
  - Awareness/lobbying
    - Dissemination of best practices, deliverables of existing project, and of use cases
    - IRDiRC is not a lobbying organization per se; discussion with national and international governments to make sure RD funding does not decline further
  - Multi-stakeholder dialogue
    - Create structured safe-harbor discussions among stakeholders on contentious topics
    - Engage with new stakeholders and with other Consortia
  - Guidance/Recommendations
    - Creation of specific guidelines for each relevant stakeholder on relevant topics
    - Define the new roadmap/ master plan for drug development incorporating the new initiatives specific for RD
  - Operational activities
    - Education, train collaborative network personnel in “agile” methodologies emphasizing early multi-disciplinary collaboration, transparency and early deliverables
- Horizon scanning might be needed in order to anticipate and contribute to trends in the field of therapies

Main action points

- Prioritize gap analysis scoring by October 27
- Collate responses gap scoring, prepare for presentation to CA meeting in Tokyo
- Review of prioritized gaps