

Meeting report series

Report of the 2nd Funders Constituent Committee Meeting

Teleconference

July 12, 2017

Participants

Dr Daria Julkowska, E-RARE-3 Consortium, France (Chair)
Prof Hugh Dawkins, Western Australian Department of Health, Australia (Vice Chair)
Dr Pedro Cortegoso Fernández, Carlos III Health Institute, Spain
Ms Christine Cutillo, National Center for Advancing Translational Sciences, NCATS/NIH, USA
Dr Iiro Eerola, European Commission, DG Research and Innovation, Belgium
Ms Roseline Favresse, Fondation Maladies Rares, France
Dr Adam Hartman, National Institute of Neurological Disorders and Stroke, NINDS/NIH, USA
Dr Daniel Lavery, Loulou Fondation, USA
Dr Teri Manolio, National Human Genome Research Institute, NHGRI/NIH, USA
Dr Lucia Monaco, Telethon Foundation, Italy
Dr Melissa Parisi, National Institute of Child Health and Human Development, NICHD/NIH, USA
Dr Senkei Umehara, Japan Agency for Medical Research and Development (AMED), Japan
Dr Sonja van Weely, The Netherlands Organisation for Health Research and Development (ZonMW), the Netherlands

Dr Lilian Lau, Paris, France – Scientific Secretariat, France
Dr Marlène Jagut, Paris, France – Scientific Secretariat, France

Apologies

Dr Christopher Austin, National Center for Advancing Translational Sciences, NCATS/NIH, USA
Dr Cindy Bell, Genome Canada, Canada
Dr Oleg Kvlividze, Children's New Hospitals Management Group, Georgia
Dr Paul Lasko, Canadian Institutes of Health Research, Canada
Dr Katherine Needleman, Food and Drug Administration, USA
Ms Marie-Christine Ouillade, AFM-Téléthon, France
Prof Willem Ouwehand, National Institute for Health Research, UK
Dr Hyun-Young Park, Korea National Institute of Health, South Korea
Dr David Pearce, Sanford Research, USA
Dr Gualtiero Ricciardi, Istituto Superiore de Sanità, Italy
Dr Ralph Schuster, Federal Ministry of Education and Research, Germany

Dr Sultan Turki AlSedairy, Saudi Human Genome Project, Kingdom of Saudi Arabia

Dr Edward Trimble, National Cancer Institute, NCI/NIH, USA

Dr Santa Tumminia, National Eye Institute, NEI/NIH, USA

Dr Heikki Vilen, Academy of Finland, Finland

Dr Yoshihiro Yoneda, National Institute of Biomedical Innovation, Health and Nutrition (NIBIOHN), Japan

Agenda

1. Priority actions
 - A. Action 3: short update from Chair on the discussion with iSearch and IRDiRC Secretariat
 - B. Action 6: RD research in developing countries
 - C. Action 7: natural history studies
2. Actions that need immediate discussion
 - A. Action 11: guidelines on patients engagement in RD research and product development
 - B. Action 14: RD methodology for health technology assessment
3. Any other business

REPORT

The Chair of the Funders Constituent Committee (FCC) thanked and welcomed participating members to the teleconference, aimed to discuss the survey results on FCC actions prioritization. The outcomes for the 14 action points have been collected in a document sent to all FCC members on 6 July 2017. This call focused on actions that should start in 2017 and on those which may need further clarification.

1. Priorities actions

1.1 Action 3: Establish process whereby funders can coordinate and prioritize efforts on research and avoid unnecessary duplication

- ▶ Several tools available for analyses of research funding, e.g. iSearch, HERO, Uber Research
 - To have better overview of what are being financed, where, and how to align strategy
- ▶ Chair of FCC held discussions and explored different platforms' suitability for IRDiRC
 - iSearch (same info as per Uber Research)
 - Useful and impressive tool, with extra functions specific to the NIH (e.g. projects not funded, so can perform additional analyses)
 - However, data from Uber Research, while much mastered, not exhaustive
 - Funder members' effort needed to complete curation
 - In particular, national funding of European countries not yet captured
 - Potential collaboration between Orphanet/ Uber Research
 - Will be sharing data (e.g. Uber Research lack rare diseases annotation)
- ▶ Pilot action prior to committing to a large scale action, via a small working group
 - Establish of a roadmap of what needs to be done
 - Ask providers (iSearch, Uber Research) for a portfolio analysis/testing opportunity
 - Ask providers if they have capacity to capture information from public domain
 - At large scale, difficult for funders to feed additional information)
 - Ask providers which type of information they require to update the database if needed
 - E.g. is a link to the funders' public website sufficient?
 - Ask providers what kind of customization can they provide to fit IRDiRC's use
 - Ask funders for a list of questions to ask the providers and see what type of answer retrieved in return

→ Action: set up a piloting committee and evaluate iSearch and Uber Research for FCC's needs

1.2 Action 6: Foster RD research, diagnosis and treatment in developing countries

- ▶ In February 2017, FCC members agreed to engage in this action on the short term
- ▶ But, contradictory prioritisation following survey vote

- Approximately 40% of members think this should be a short-term action (start in 2017), while another 40% as long-term action (needs additional support)
- It is obviously a project that should run over long term, however, time is needed to initiate fruitful connection, therefore should start action now
- ▶ To first develop a small working group
 - Get in touch with various organizations involved in related activities, e.g. H3Africa
 - Identify, together with these organisations, their needs and what IRDiRC can do collaboratively with them
 - Only after this, will determine how IRDiRC can contribute in the long term

→ Action: set up working group to identify collaborating partners

1.3 Action 7: Formulate and disseminate standards and guidelines including promotion of natural history studies

- ▶ Natural history studies
 - Problem of financing these studies due to their length
 - Innovative ways could circumvent this issue or exploit resources already collected
- ▶ A Task Force to be set up
 - “Which innovative mechanisms would allow natural history studies to stay short”
 - Recommendations of the TF can then be integrated by FCC members into their funding opportunities
 - To collaborate with the SCs on this topic (there used to be a WG on Natural Histories)
 - To discuss who will lead the TF and also identify the agenda for this TF

→ Action: identify FCC members who wish to lead this TF

→ Action: contact SCs for expression of interest to work on this action

2. Actions that need immediate discussion

2.1 Action 11: Formulate and disseminate standards and guidelines on patient engagement in RD research and product development

- ▶ Survey outcome shows a split of 1/3 each for short, medium and long-term implementation
- ▶ The Therapies and Interdisciplinary SCs are launching TF on patient engagement in RD research
 - Patient Advocacy CC also participant to this TF

→ Action: Sci Sec to transmit interest of FCC to the TSC leadership

2.2 Action 14: Promote RD methodology development for health technology assessment and health economics research to evaluate disease burden and access of patients to diagnostics and therapies

- ▶ This action – connected to Goal 3 – was also identified as priority in February 2017
 - However, survey outcome now indicated a majority voted this as long-term priority
 - Some FCC members have impression they're lacking expertise on this topic and therefore need more preparatory work before launching such funding opportunities
- ▶ The Interdisciplinary SC has recently started to look at this issue
 - FCC would like to contribute to the discussions
 - Decision on action is postponed until more information is available

Action Items and deliverables

- ▶ Complete Doodle re teleconference in September
- ▶ Express interest to participate in TFs/WG
- ▶ Prepare agenda and items for discussions in September
- ▶ Check with iSearch / Uber Research on piloting opportunity
- ▶ Express expectations for the November meeting in Tokyo