

Meeting report series

Report of the 8th Diagnostics Scientific Committee Meeting

Teleconference

12 May 2015

Participants

Dr Kym Boycott, Ottawa, Canada, Chair
Prof Michael Bamshad, Seattle, USA, Co-Chair
Prof Han Brunner, Nijmegen, Netherlands, Co-Chair
Prof Xavier Estivill, Barcelona, Spain
Prof Gert Matthijs, Leuven, Belgium
Prof Milan Macek, Prague, Czech Republic

Dr Lilian Lau, Scientific Secretariat, Paris, France
Ms Antonia Mills, Scientific Secretariat, Paris, France

Apologies

Prof Fowzan Sami Alkuraya, Riyadh, Kingdom of Saudi Arabia
Prof Johan den Dunnen, Leiden, Netherlands
Prof Woong-Yang Park, Seoul, Republic of Korea
Prof Pak-Chung Sham, Hong Kong, China
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Agenda

1. Update on Task Forces
2. Update on process for “IRDiRC Recommended”
3. Update on IRDiRC goals
4. Update from the Executive Committee
5. Review of DSC Roadmap and planning its dissemination
6. Focus of face-to-face meeting in Glasgow

REPORT

Update on Task Forces

Five Task Forces have been constituted by IRDiRC to work on selected prioritised topics:

- ▶ Matchmaker Exchange (MME)
- ▶ Patient Relevant/Reported Outcome Measures (PROM)
- ▶ Small Population Clinical Trials (SPCT)
- ▶ Machine Readable Consent (MRC)
- ▶ Data Mining-Repurposing (DMR)

MME is underway as a joint action with Global Alliance (GA4GH) and from the DSC, possible project/workshop for 2016 may be on ontologies and/or functional analysis.

Update on process for “IRDiRC Recommended”

“IRDiRC Recommended” was recently launched and call for applicants for this label is ongoing. Two applications have been received to date, and review process is due to begin.

The steps involved in “IRDiRC Recommended” review:

- ▶ Receipt of submission of tool/standard/guideline for review
- ▶ Appointment of experts to review the submission by the Scientific Secretariat (Sci Sec)
- ▶ Rapporteur’s report from the Scientific Committee (Sci Comm) following experts’ review
- ▶ Assessment of experts’ and rapporteurs’ reports by the Executive Committee (Exec Comm)
- ▶ Vote collection for acceptance and rejection of tool/standard/guideline

The format of the rapporteurs’ report needs to be defined in order for the Sci Comm to perform its duty efficiently. The Chair also encouraged members to submit an application for recognition of their tools.

Update on IRDiRC goals

The IRDiRC website (www.irdirc.org) has just undergone a reorganisation and now better promotes its current activities as well as displaying the progress towards IRDiRC goals. Data from Orphanet enabled the visualisation of number of new genes and genetic tests available, but the methodology is not described in detailed on the relevant page; the Chair requested that Sci Sec provides this information. The DSC was also informed that the Sci Sec intends to update the way these numbers are displayed to better represent the progress made.

Centers for Mendelian Genomics (CMG) used to keep a list of genes discovered but this is not currently the case; the next phase of CMG will probably include a more thoroughly curated list than what is currently available on OMIM. Mike could, however, provide information on the pace of gene discovery based on next generation and conventional sequencing strategies.

Update from the Executive Committee

The Exec Comm met in March 2015 in Madrid, Spain. The DSC was represented by a member who gave an update from the DSC, including the nominations for Task Forces and plans for the upcoming meeting in Glasgow. The most recent Exec Comm teleconference in May 2015 was largely a process discussion.

Review of DSC Roadmap and planning its dissemination

The DSC Roadmap was put together at the meeting in Prague in December 2013. The committee wish to disseminate something about IRDiRC, centred on the gaps identified through the Working Groups (WGs) in order to reach the goal of diagnosing most diseases by 2020. As funders need time to consider and pick up projects that work towards the goal, to set the motion in action, an opinion piece needs to be written and disseminated without further delay.

Suggested content of the paper:

- ▶ Title the manuscript around the goal of IRDiRC/DSC, i.e. diagnosing most rare diseases by 2020
- ▶ Introduction centred around the pace of gene discovery since 2010, what is the trend, and predict how this will go in the coming years
- ▶ Overview on WGs and why each priority was identified as important to achieve the diagnostic goal
- ▶ List critical steps necessary to move toward the goal, which may or may not indicate the potential actors to act upon them
- ▶ Key messages to include integration into healthcare system and eligibility for reimbursement, and research funding needed to enable large scale data sharing and functional analyses
- ▶ Inclusion of estimated cost involved to enable the critical steps with emphasis on what should and could be achieved in the next few years within the indicated budget

Authorship of the paper will include any contributing members including past WG Chairs. The Chair wishes to also include Orphanet and OMIM, resources from which data was obtained, to make a more powerful statement. The ideal would be to engage different members of the rare disease community to be implicitly involved at all levels. The DSC is looking into collaborative dissemination of the opinion paper by both AJHG and EJHG.

Focus of face-to-face meeting in Glasgow

The following will be focus items for the DSC face-to-face meeting:

- ▶ Defining the role of Sci Comm in Task Forces
- ▶ Discussion and opinion paper writing re DSC roadmap and recommendations

The Chair will start on the first version to introduce IRDiRC and the goals of the Sci Comm, the current state of play, and be assisted in formulating the critical next steps. No extensive references are required, and the first draft may be circulated to all members for additional input ahead of the face-to-face meeting.

Main deliverables

- ▶ Definition of elements in rapporteurs' report
- ▶ Provision of methodology used to calculate the number of new genes and genetic tests
- ▶ Information on the pace of gene discovery
- ▶ First version of paper with introduction and critical next steps
- ▶ Contact ASHG/ESHG re building bridges session, collaborative dissemination of opinion paper
- ▶ Identify any additional points to be included in the opinion paper