

Meeting report series

Report of the 11th Diagnostics Scientific Committee Meeting

Teleconference

11 May 2016

Participants

Prof Kym Boycott, Ottawa, Canada – Chair
Assoc Prof Gareth Baynam, Perth, Australia – Vice Chair
Prof Anthony Brookes, Leicester, UK
Prof Gert Matthijs, Leuven, Belgium
Dr Feng Zhang, Shanghai, China

Dr Lilian Lau, Scientific Secretariat, Paris, France

Apologies

Prof Fowzan Sami Alkuraya, Riyadh, Kingdom of Saudi Arabia
Prof Michael Bamshad, Seattle, USA
Prof Han Brunner, Nijmegen, Netherlands
Prof Johan den Dunnen, Leiden, Netherlands
Prof Xavier Estivill, Barcelona, Spain
Prof Milan Macek, Prague, Czech Republic
Prof Woong-Yang Park, Seoul, Republic of Korea
Prof Pak-Chung Sham, Hong Kong, China
Prof Hendrik Stunnenberg, Nijmegen, Netherlands

Agenda

1. Task Force proposals
 - Solving the unsolved
 - Clinical data sharing for gene discovery
2. Update on commentary
3. Vice Chair of the DSC

REPORT

1. Task Force proposals

Two Task Force proposals were refined (see attached documents for the refined proposals) and they will be submitted to the Executive Committee for approvals in order to commence work. In identification of aspects to act upon, the ideal, in order to optimise work and reduce overlaps/reinvention of the wheel, as well as maximising impact, IRDiRC and GA4GH activities should be complementary.

Potential Task Force members for both proposals were suggested, and ***all Diagnostics Scientific Committee (DSC) members with additional suggestion(s) are encouraged to put forward name(s) to the Chair.***

2. Update on DSC commentary

The DSC commentary is still in progress. Related OMIM analysis for 2015 is being prepared for inclusion in the paper, while the status of data analysis from Orphanet is unknown. ***The Chair will contact Orphanet and the Scientific Secretariat will follow up on this.***

3. Vice Chair of the DSC

Following a call of interest among members to serve as Vice Chair, Gareth Baynam is confirmed as the Vice Chair of the DSC and will assist the Chair in coordinating the DSC and its activities.

Action points

- ▶ Put through suggestions of Task Force members
- ▶ Contact Orphanet regarding 2015 data analysis