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MINUTES

International Scientific Advisory Committee Meeting
Thursday 10 April, 2014
2200hrs Melbourne, Australia time

MEMBERS

Garry Cutting (Co-chair, USA)
Richard Cotton (HVP Scientific Director, Australia)
Raymond Dalglish (UK)
Marc Greenblatt (President HGVS, USA)
Finlay Macrae (Australia)
Donna Maglott (USA)
Peter Taschner (The Netherlands)
Mauno Vihinen (Sweden)

Apologies

John Burn (Co-chair, UK)
Johan den Dunnen (The Netherlands)
Stephen Lam (President IFHG, Hong Kong)
Ming Qi (China)
Raj Ramesar (South Africa)
Michael Watson (USA)
Martina Witsch-Baumgartner (Austria)

Heather Howard (HVPI Operations Manager)
Timothy D. Smith (HVPI Communications Officer)
Helen Robinson (HVPI Liaison Officer for WHO)

AGENDA

1. Welcome

2. Apologies

Apologies were noted as above.

3. Confirmation of minutes of previous meeting

The minutes of the previous meeting were confirmed unanimously.

4. Issues arising from previous meeting

a. Establishment of an Ethics Committee – Garry Cutting/John Burn

This issue was not discussed.

b. Process for expanding ISAC membership - Heather Howard

The last meeting decided that the membership of the ISAC would be expanded by inviting those individuals who had been duly nominated and received votes in the election last December to become non-voting members. Due to the number of ISAC members that were not present on the call, those absent were given the opportunity to object to this decision via email. No objections were received. The ICO is seeking clarification that they can now proceed to issue the invitation to these individuals. The Committee directed the ICO to issue the invitations.

The issue of two or more individuals from the same institution or location being members of the ISAC was discussed. The possibility of seconds or substitutes was again discussed but further discussion was postponed until the face-to-face meeting in Paris in May.

5. ISAC Priorities for 2014-2016 – Helen Robinson

Helen Robinson spoke to the paper that was distributed to the Committee. She noted that additional comments had been received from Mauno Vihinen that had not yet been incorporated into the paper.

Raymond Dagleish noted that running education and training courses is expensive. He also spoke to how the distinction between the database curator and administrator role is handled in the database that he is responsible for: the roles are separate but performed by the same individual. Peter Taschner spoke to how these two roles are separated in the LOVD installations hosted at Leiden: Leiden provides the database administration resources, the curators are distributed all around the world. Finlay Macrae provided a different perspective from the InSiGHT database, where the principal curator is chiefly a bioinformatics specialist and handles the administrative role, but through experience has acquired gene-specific knowledge and is also able to draw on the knowledge and experience of a larger committee of curators. Garry Cutting queried if there was an opportunity here to perhaps define new terms for the different roles to assist with differentiating between them.

Peter Taschner requested that the issue of Country Portals be added to the document.

Donna Maglott commented on the 'good quality databases' issue. The ClinGen group is asking ClinVar to get an explicit statement of the methods used to evaluate the clinical significance or the consequences of a variant relative to a disorder. This could be something that the HVP considers when developing metrics for curation that would help identify good quality databases. Marc Greenblatt reported that the ACMG laboratory committee is preparing a set of guidelines that will address this interpretation issue for laboratories, but would also be applicable to databases. He suggested that as several people from the various groups involved in developing interpretation guidelines would be present at HVP5 it will be a good opportunity to develop recommendations for standards setting moving forward that will apply to laboratories, databases and disease interest groups.

Finlay Macrae stated that developing a business case for database support was a very important issue. Peter Taschner questioned how reliable information could be gathered on how many times a database was accessed in order to reach a medically actionable conclusion. Raymond provided some information on how he uses Google Analytics on his database to identify users and how that information could be used to request statements about the value of the database.

Garry Cutting noted that the six aims presented on the paper were well stated but some overlap exists and questioned whether they could be restated in a more general or global way or perhaps combined.

Helen Robinson suggested that the document she presented be updated based on these discussions for further discussion at the next Committee meeting.

6. Reports from Working Groups

Working Group chairs were asked to provide a report by email to the Committee.

7. Standards Development & Recommended Systems Status

a. RSS Applications under Review – Timothy Smith

i. VarioML

Discussion postponed until the next meeting.

b. Activity Proposals & Applications

i. AP08 – Minimum Content Requirements for HVP Country Nodes – Timothy Smith

Peter Taschner advised the Committee that Martina Witsch-Baumgartner would be submitting a revised proposal. Discussion was postponed until the revised proposal is received.

ii. AP06 – VCF File Format

Discussion postponed until the next meeting.

8. HVP5

The Committee discussed the timing of the ISAC meeting and joint meetings with the two Advisory Councils during HVP5.

9. Scientific Director's Report

Richard Cotton advised the Committee of the following matters:

- The ICO is facilitating the preparation and lodging of a joint grant application to the University of Melbourne between InSiGHT and some colleagues in Brazil that will initiate the HVP Brazilian Node.
- Richard Cotton, Helen Robinson and Peter Robinson would be speaking at a special Human Variome Project session at the upcoming HUGO meeting in Geneva.
- Marc Greenblatt and Johan den Dunnen has spoken about the Human Variome Project at the recent ACMG meeting.

10. Other matters

a. Global Alliance Meeting

This issue was not discussed.

b. Phenotype Group

This issue was not discussed.

11. 2014 ISAC Meetings

- 23 May (in person at HVP5)
- 26 June
- 31 July
- 28 August
- 25 September
- 21 October (in person at ASHG)
- 27 November
- 18 December