



THE
HUMAN VARIOME
PROJECT
sharing data · reducing disease
an NGO Official Partner of UNESCO

MINUTES

International Scientific Advisory Committee Meeting
Thursday 27 February, 2014
2300hrs Melbourne, Australia time

MEMBERS

Garry Cutting (Co-chair, USA)
Richard Cotton (HVP Scientific Director, Australia)
Raymond Dalglish (UK)
Donna Maglott (USA)
Raj Ramesar (South Africa)
Peter Taschner (The Netherlands)
Mauno Vihinen (Sweden)
Michael Watson (USA)

Apologies:
John Burn (Co-chair, UK)
Johan den Dunnen (The Netherlands)
Marc Greenblatt (President HGVS, USA)
Stephen Lam (President IFHG, Hong Kong)
Finlay Macrae (Australia)
Ming Qi (China)
Martina Witsch-Baumgartner (Austria)

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

SUMMARY OF ACTIONS

Sec.	Action	Responsible
4b	Heather Howard to send a reminder to the Committee about nomination to the Board	Heather Howard
4b	Committee members to submit nomination forms to the Returning Officer	ISAC
6	Committee members to provide feedback on the document to Helen Robinson	ISAC
6	Helen Robinson to update the document as feedback is received and report progress	Helen Robinson
7b	Committee to consider the AP08-2014 at the next meeting	ISAC
10a	John Burn and Mike Watson to report on Global Alliance meeting at the next Committee meeting	John Burn, Mike Watson

MINUTES

1. Welcome

2. Apologies

Apologies were noted from, Johan den Dunnen, Stephen Lam and Martina Witsch-Baumgartner.

3. Confirmation of minutes of previous meeting

Minutes were confirmed unanimously

4. Issues arising from previous meeting

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

Not discussed.

5. HVP Governance Issues – Heather Howard

a. Process for expanding ISAC membership

Heather Howard reported that a list of the individuals who had been nominated to stand for election to the ISAC and had received votes in the election, but had not been elected, as well as their country of origin, had been sent to the Committee as requested. The opinion of the ICO—that all individuals on the list be invited to participate on the Committee as non-voting members—was relayed to the Committee.

The Committee discussed a process for expanding the membership. Members again expressed a desire for a broader membership, particularly in relation to geographic distribution of Committee members, but were reluctant to discriminate between the individuals on the list circulated by the ICO solely on the basis of geographic origin. The point was raised that all by inviting all individuals on the list to become members of the Committee, geographic diversity would be obtained, however then the concern would be the unwieldiness of such a large group. This issue could be mitigated by ISAC members identifying seconds from the expanded Committee membership.

An alternative process was discussed whereby the Committee would work towards greater geographic diversity in the future, rather than in the short term, with places on the Committee reserved for members from specific countries/regions, and perhaps, specific organisations that have complementary goals to those of the Human Variome Project. This may require changes to the constitution of HVP but would not be difficult to achieve.

Raymond Dalglish summarised the discussion to date, stating that the consensus was to invite all the individuals on the list circulated by the ICO to participate on the Committee. The issues of the status of the new members as voting or non-voting members, the use of seconds, and the process to use in the future to avoid a similar situation were still open. Given the limited number of ISAC members present on the call, which was less than a quorum at this point in time, Garry Cutting requested that the matter be resolved at the next meeting

b. Appointment of Three Members to the Board

Heather Howard reported to the Committee that a nomination form for the Committee's appointees to the HVPIL Board of Directors had been prepared as requested and sent to members. The Committee were asked to set a date by which the forms must be returned. The Committee determined that forms should be returned to the Returning Officer by 14 March, 2014.

ACTION: ICO to send a reminder email regarding the deadline, including the <i>Guide for Directors</i> publication to Committee members Committee members to submit nomination forms to the Returning Officer
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6. ISAC Priorities for 2014-2016

Helen Robinson presented a draft document distilling the discussions of the ISAC and ICCAC on priorities for the coming two years. The Committee provided feedback on the document. Peter Taschner suggested that the Human Variome Project should continue to work with journal editors to require the use of standard nomenclature and submission to a public database prior to submission. The Committee suggested that this might be actioned by creating and publishing a list of journals enforcing such requirements. Raymond Dalglish suggested that the International Society of Biocuration be recruited to assist with this activity. Timothy Smith reported on the progress of the WG04: Sequence Variant Description Committee and their progress towards establishing a joint committee with HGVS and HUGO to maintain and extend the nomenclature system in an ongoing and sustainable fashion and suggested that this joint committee could also monitor and report on journal requirements.

In the interests of time, Gary Cutting requested that further feedback be sent via email to Helen Robinson ahead of the next meeting of the Committee on 10 April, and a much more substantive discussion be held at that meeting. It was also requested that Helen Robinson send an updated version of the document in 2-3 weeks reflecting feedback received at that time. Helen Robinson reminded the Committee of the approaching face-to-face meeting in Paris in

May and that it would be beneficial to have this activity completed by this date to ensure that a robust discussion between the Committee and the two Councils can take place.

ACTION: Committee members to provide feedback on the document to Helen Robinson prior to next meeting
Helen Robinson to update the document as feedback received and report progress to the Committee in 2-3 weeks

7. Standards Development & Recommended Systems Status

a. *RSS Applications under Review – Timothy Smith*

VarioML - Timothy Smith reported that enough reviewers had not yet returned comments for the ISAC to consider at this meeting.

No new applications have been received.

b. *New Activity Proposals & Applications*

Timothy Smith presented the Committee with AP08-2014: Minimum Content of a Country Specific Variant Database and the accompanying comments and recommendations from the ICO. Peter Taschner noted the potential for overlap between this activity and the work of his Working Group on minimum content requirements for gene/disease specific databases and volunteered to be a member on any Working Group chartered to undertake the proposed activity to address the overlap. The Committee determined to consider the issue of Sponsoring Council for the Activity Proposal at their next meeting.

ACTION: Committee to consider the Activity Proposal at the next meeting

8. HVP5

Heather Howard reported to the Committee that the abstract submission deadline has passed and that all submitted abstracts have been reviewed. A draft programme is being prepared and will be circulated to the Committee once approved by the Scientific Programme Committee. Heather Howard further reported that, thanks to the efforts of Raj Ramesar, a full session on activities in Africa is looking very likely. Additionally, there will be a satellite meeting of numerous groups involved in breast and ovarian cancer genetics held during HVP5, with a view to consolidating the various databases that exist for these conditions.

Heather Howard reported that Martin Bobrow will be unable to attend the meeting to represent the Global Alliance. Peter Goodhand has been invited and the ICO is awaiting a response.

9. Scientific Director's Report

Richard Cotton reported on the following items:

The Human Variome Project will be hosting a session at the ACMG meeting along with Marni Falk, who is leading a major effort to database variants in mitochondrial genes. Marc Greenblatt and Johan den Dunnen will be representing Human Variome Project.

Peter Pearson is now working in Brazil and is working to assist in the creation of a Node there.

The Human Variome Project will be hosting a Session at HUGO HGM meeting in Geneva in April.

Human Molecular Genetics has taken up HVP recommendations on nomenclature and submission of variant data to public databases prior to publications.

The Dutch and Belgian breast cancer groups have submitted all their variants to the LOVD 3 shared installation.

A colleague in Brisbane is developing an International meeting on cardiac genetics in Brisbane that will be held soon. This might be a good time to encourage this group to establish a database on variants involved in cardiac diseases. Heidi Rehm has also started a similar group and it would be a good idea to bring those two groups together. There was to be a cardiac genetics meeting held in Cardiff run by Dhavendra Kumar, but it does not seem to have eventuated.

Donna Maglott noted that the cardiac myopathies are a major focus for the ClinGen project.

10. Other matters

a. Global Alliance Meeting

Garry Cutting noted that the Global Alliance meeting in the United Kingdom would be taking place next week and that the Human Variome Project would be represented there by John Burn and Mike Watson. He undertook to ask them to provide a report on the meeting to the Committee.

ACTION: John Burn and Mike Watson to report on Global Alliance meeting at the next Committee meeting

b. Relationship with WHO

Helen Robinson informed the Committee that the ICO has started the formal process of obtaining NGO status with WHO as a result of a meeting last week.

11. 2014 ISAC Meetings

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