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# MINUTES

International Scientific Advisory Committee Meeting  
Thursday 30 January, 2014  
2300hrs Melbourne, Australia time

## MEMBERS

Garry Cutting (Chair, USA)  
Richard Cotton (HVP Scientific Director, Australia)  
Raymond Dagleish (UK)  
Marc Greenblatt (President HGVS, USA)  
Finlay Macrae (Australia)  
Donna Maglott (USA)  
Raj Ramesar (South Africa)  
Peter Taschner (The Netherlands)  
Mauno Vihinen (Sweden)  
Michael Watson (USA )  
Martina Witsch-Baumgartner (Austria)

### Apologies:

John Burn (UK)  
Johan den Dunnen (The Netherlands)  
Stephen Lam (President IFHG, Hong Kong)  
Ming Qi (China)

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

## SUMMARY OF ACTIONS

Sec.	Action	Responsible
<a href="#">5a</a>	Consider individuals to include in the expansion of Committee	ISAC
<a href="#">5a</a>	Consider a process for ensuring greater professional and geographic diversity in future Committees	ISAC
<a href="#">5a</a>	Prepare advice on expanded Committee membership	ICO
<a href="#">5b</a>	Provide information to the Committee on the role and responsibilities of being a Director of HVPI	ICO
<a href="#">6b</a>	Ask Peter Robison to consider submitting Phenomizer and the Human Phenotype Ontology for RSS	Donna Maglott
<a href="#">6b</a>	Provide information on RSS process, examples of awarded systems and systems that could be proposed to the Committee	ICO
<a href="#">9b</a>	Members planning to attend HGM 2014 to inform the ICO by email	ISAC
<a href="#">10</a>	Send names of potential speakers for HVP5 to Heather Howard	ISAC

## MINUTES

### 1. Welcome & Appointment of Chair

Garry Cutting and John Burn will continue as Co-Chairs for this term of the Committee.

Garry Cutting welcomed members, particularly the new members of the Committee joining for the first time.

## 2. Apologies

Apologies were received from John Burn, Stephen Lam and Johan den Dunnen.

## 3. Confirmation of minutes of previous meeting

Minutes of the previous meeting were confirmed.

## 4. Issues arising from previous meeting

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

Garry Cutting deferred discussion until John Burn was able to participate.

## 5. HVP Governance Issues

### a. *Process for expanding ISAC membership – Heather Howard*

The Committee discussed a proposal from the ICO to consider expanding the membership of the ISAC. Helen Robinson provided background to the proposal, noting that quite a few people came close to being elected to the Committee in the last round of elections and that the Committee might consider it advantageous to make use of these obviously interested people. Expanding the Committee would also increase the diversity of the Committee, both in terms of the various professional perspectives and those from various parts of the world. The Committee was provided with a list of people who had been formally nominated, seconded and received votes in the last elections.

Following a discussion of the possible merits of expanding the membership of current and future Committees, members asked the ICO to prepare further information for consideration at the next meeting taking into account the following points:

- The need for the Committee to run effectively and efficiently
- The need for further information regarding the spread of professional roles and geographic locations of the various candidates in relation to existing members, and
- A suitable process and timeline for this taking into account the rules and regulations of Human Variome Project.

**ACTION:** Committee members to consider individuals to include in the expansion of Committee and a process for ensuring greater professional and geographic diversity in future Committees  
ICO to prepare advice in advance of the next meeting.

### b. *Appointment of Three Members to the Board – Heather Howard*

Heather Howard informed the Committee that it is required to appoint three members to the Board of Directors of HVPIL. As this is the first time that this will occur, there is no procedure in place to do so. The ICO will assist in the implementation of any process the Committee wishes to adopt.

Mike Watson requested that the Committee be provided with information on the role and responsibilities of being a Director. He also noted that the Committee may want to give some thought on this matter to the issues from the last matter regarding the mix of geographic location and professional expertise.

Garry Cutting directed the ICO to provide the Committee with the requested information before a decision to be made at the next meeting.

**ACTION:** ICO to provide information to the Committee on the role and responsibilities of being a Director of HVPIL.

## 6. Recommended System Status

Timothy Smith provided the Committee with some background on the process in place for awarding Recommended System Status and an update as to the current status of proposed systems for the benefit of the new members of the Committee.

**a. Applications under Review – Timothy Smith**

**i. VarioML**

Timothy Smith informed the Committee that the ICO is having trouble finding suitable reviewers for VarioML, but after some helpful suggestions from Mauno Vihinen, has had some positive communications in the past few days with some appropriate reviewers. It is expected that reports will be available for the next meeting.

Raymond Dalgleish noted that the trouble most likely stems from the fact that most of the people that understand VarioML were involved in its development.

**b. New Applications**

None formally received since last meeting. Members were asked if they knew of systems that could be put forward for review this year. Café Variome was suggested; Mauno Vihinen informed that now that the Variation Ontology publication had been accepted for publication, he would submit VariO for consideration. Mauno Vihinen also undertook to distribute a copy of the publication to members. Several members suggested that phenotype systems could be encouraged, so Phenomizer and the Human Phenotype Ontology were recommended as being timely for formal review as it may help with securing future funding. Donna Maglott offered to ask Peter Robinson to consider submitting these systems.

**ACTION:** Donna Maglott to ask Peter Robinson to consider submitting Phenomizer and the Human Phenotype Ontology

In response to a request for clarification of the types of systems that are eligible for Recommended System Status, it was advised that any system, tool or process that is available free of charge for non-commercial use could be considered. To assist members, particularly the new ISAC members, acquaint themselves with this process, the ICO will circulate information on the HVP process, together with examples of systems already awarded Recommended System Status, and a list of systems that could be proposed.

**ACTION:** ICO to provide information on the Recommended System Status process, examples of systems already awarded, and a list of systems that could be proposed.

**7. Activity Proposals**

None received since last meeting

**8. ISAC Priorities for 2014 – Helen Robinson**

Committee members were asked to consider the priority activities for 2014 – 2016. Helen Robinson indicated that the Project’s response to the Global Alliance for Genomics and Health was the most recent document to publicly set out HVP’s strategic directions and so might provide a framework for determining priority areas for 2014 and beyond. She also noted that the document presented a slight shift in thinking from the existing strategy document – HVP Roadmap 2012 to 2016 - and so a discussion was timely. Five areas were indicated to the HVP response:

- Promoting and assisting the shift from the science of the human genome to better understanding the biology of disease and its impact of delivery of health services
- Balancing the interests of research intensive countries and less developed countries when developing global standards and other initiatives
- Encouraging the harmonization of regulatory frameworks and governance between countries and regions by actively supporting the sharing of knowledge and expertise
- Developing and promoting the role of database curators as being crucial to ensuring data quality, standards and description, particularly for large datasets of genomic information
- Developing a shared understanding of the promise of genomic medicine, particularly among key interest groups including donors and funders

The Committee discussed how this might translate into more specific activities for 2014 and beyond and a preliminary list was developed:

- Encouraging and supporting small groups of Consortium members to seek joint funding for research projects focussed more on health service delivery or translating research into health service delivery
- Promoting mechanisms for improving data quality and consistent interpretation of variation data

- Encouraging more countries to join the ICCAC
- Promoting data storage and sharing and developing indicators to measure and report on progress
- Recruiting more curators for gene/disease specific databases
- Identifying resources to research the economics and financing, particularly cost-benefit analyses, of genomic medicine and the work of the Human Variome Project that could be used to present the business case for data sharing between countries to governments and funders
- Mapping the heterogeneity of ethical, regulatory and legal frameworks in different jurisdictions and determining how to manage these differences effectively in supporting international data sharing
- Continuing attention to sustainability and funding for the various activities the HVP members are involved in
- Determining and promoting agreed mechanisms for attribution and recognition of the work of gene/disease specific database curators when central databases aggregate or federate into multiple datasets

Marc Greenblatt noted that it would be very worthwhile to attempt to match the funding issue with the curators issue. He also noted that the ClinGen project in the US is now underway and it would be worthwhile for the HVP to consider developing standards out of the outputs of that project and promulgating these standards internationally.

Helen Robinson thanked the members for their input and agreed to condense these discussions into a short document and send this around for further discussion at the next meeting. Garry Cutting requested that this issue be moved up the agenda for the next meeting to allow for more time for discussion.

**ACTION:** ICO to develop and circulate a short discussion paper in advance of next meeting.

## 9. HVP Relationship with other Entities

### a. *Global Alliance*

Matter covered sufficiently in the previous item.

### b. *HUGO – Helen Robinson*

Helen Robinson reminded the Committee that there will be an HVP session during the 2014 HUGO meeting and asked members to let the ICO know if they were planning to attend this meeting.

**ACTION:** Committee members planning to attend HGM2014 to inform the ICO by email

### c. *UNESCO – Helen Robinson*

Matter not discussed.

### d. *WHO – Helen Robinson*

Matter not discussed.

### e. *H3Africa – Raj Ramesar*

Raj Ramesar informed the Committee about recent progress in the Human Hereditary and Health in Africa (H3Africa) Program. This program is funded by the NIH and the Wellcome Trust and includes a large number of projects involving 20 African countries. These projects all have genomics at their core, and include a broad range of areas, for example, infectious disease and chronic disease. Chief Investigators must be from Africa and projects must have at least three collaborators from other African countries. Raj Ramesar is promoting exchange of information between H3Africa and HVP, and would like to pursue closer ties between the two initiatives. He raised the possibility of showcasing a few H3Africa projects during a session of HVP5. He is keen to explore ways the HVP could assist in increasing the profile of African work with their own governments as a means to securing sustainable support beyond the H3Africa program.

Heather Howard reported that she has been informed by Casimiro Vizzini that UNESCO is very keen to host as many African delegates that are able to attend HVP5 and they will arrange meetings with members of UNESCO permanent delegations from African nations. This will be taken up in the planning of HVP5.

## 10. HVP5

Marc Greenblatt informed the Committee that the HGVS Council has offered assistance to review abstracts if, as there has been in the past, there are an overwhelming number of submissions. The Committee thanked the HGVS Council for their offer.

Heather Howard reported that the program for the meeting would be released in the coming month, but there was still time for Committee members to suggest individuals who should be invited to speak, particularly those who could offer one or two key presentations.

<b>ACTION:</b> Committee members to forward names of potential speakers for HVP5 to Heather Howard
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## 11. Scientific Director's Report

Richard Cotton presented his Scientific Director's report (see Attachment A). He made particular mention of the strong support for the HVP and an Indian Node on his recent trip to India. He also noted the recent progress towards a Canadian Node.

## 12. Other matters

No other matters discussed.

## 13. 2014 ISAC Meetings

- 27 February
- 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

## **ATTACHMENT A**

### **Human Variome Project - Scientific Directors Report – January, 2014**

Happy New Year to you all and a very warm welcome to the new members of the Committee, let's hope 2014 is a good year for us all.

#### **Meetings attended**

##### **India**

John Burn and I were guest speakers at the Indian Society of Human Genetics Conference 2014, Ahmedabad, India, January 23-25, 2014. I spoke on HVP and there was much interest John spoke amongst other things on his hand held analyzer. We both were featured in the Times of India and the Gudjarat Samachar circulation 20 million but in the Indian language! Samir Brahmachari was also present at the meeting who I know well and John and I briefed him regarding the HVP work and asked if he would assist in starting an Indian Node he agreed to take the Indian work forward.

Prior to the meeting Casimiro Vizzini from UNESCO organized a meeting with Indian government officials. The person I spoke with was the Indian secretary of education. The project was well received and will be raised at a planning meeting in 2 months' time, we hope to see some progress in India over the next few months.

#### **Progress**

##### **Canada**

We were pleased to hear a group led by Lerner Jordan Ellis had obtained funding for data collection and we look forward to interacting with them and congratulations.

#### **Regions**

##### **S E Asia**

We congratulate Falil on the establishment of this regional node a great achievement and precedent for HVP. We also wish him well in recovering from a recent illness.

#### **Upcoming Meetings**

- ACMG Annual Clinical and Genomics Meeting, March 25-29, 2014, Nashville, Tennessee, USA.  
We are pleased to report that HVP has been chosen as part of the ACMG Clinical Meeting Program in 2014. The session will be on "Making genomics data accessible: building shared genomic resources" and will be co-moderated by Richard Cotton and Marni Falk (The Children's Hospital of Philadelphia, USA).
- HVP will have a session at HUGO 2014, Geneva (27-30 April 2014).
- HVP 5 Biennial Meeting, UNESCO headquarter, Paris, May 19-23, 2014.
- 14th International Congress on Neuronal Ceroid Lipofuscinoses (Batten Disease). Richard Cotton is a member of the organising committee (in the Registries' Committee). Segolene Ayme is a speaker at the meeting.
- ASHG- San Diego 18th – 23rd October, 2014
  - HVP Meetings being held :
    - Editors Meeting
    - Scientific Advisory Committee Meeting
    - HVP Booth
- HVP session at Asia Pacific Society of Human Genetics meeting, 19-22 Nov 2014.