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# MINUTES

Interim Scientific Advisory Committee Meeting  
Thursday 22<sup>nd</sup> August, 2013 – 10.00PM Melbourne, Australia time

## MEMBERS

John Burn (Alternate Chair, UK)  
Arleen Auerbach (USA)  
Mona El Ruby (Egypt)  
Mauno Vihinen (Sweden)  
Gert-Jan van Ommen (The Netherlands)

Richard Cotton (HVP Scientific Director, Australia)  
Heather Howard (HVPI Operations Manager)  
Tim Smith (HVPI Communications Officer)

### Apologies

Garry Cutting (Chair, USA)  
Mireille Claustres (France)  
Finlay Macrae (Australia)  
Helen Robinson (HVPI Liaison Officer for WHO)  
Ming Qi (Head China Node Partnership, China)  
Johan den Dunnen (The Netherlands)  
Aida Falcon de Vargas (Venezuela)  
Richard Gibbs (USA)  
Marc Greenblatt (President HGVS, USA)  
Yoichi Matsubara (Japan)  
Stephen Lam (President IFHGS, Hong Kong)

## AGENDA

Meeting Opened 2209hrs

Meeting inquorate. All decisions and actions are considered advisory until confirmed.

### 1. Welcome

### 2. Apologies

Apologies were noted from Garry Cutting, Mireille Claustres and Finlay Macrae.

### 3. Confirmation of minutes of previous meeting

Minutes accepted unanimously.

### 4. Issues arising from previous meeting

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

Sir John reported that Garry Cutting and he would be meeting in Baltimore in October to discuss this issue.

**ACTION:** Garry Cutting and John Burn to progress work on establishing an Ethics Committee

#### *b. Guidance on nomination and election procedures for ISAC – Heather Howard*

Heather Howard reported that the interim committee will continue until the end of year. A call for nominations will be sent to all Consortium members on 1 September. This has already been sent to the Committee. Heather remarked that she hoped all current members will renominate for election.

Heather briefly outlined the election process. All Consortium members are eligible to stand for election. Each nominee must be nominated and seconded by a Consortium member. Each of the two Advisory Councils will elect six people to the Committee. Nominees must indicate which council they wish to be voted on their candidacy.

Arleen asked how many positions are on the interim Committee now. Heather replied that there are twelve elected positions plus ex-officio positions for the Presidents of the IFGHS and HGVS and the Project's Scientific Director. Additionally, a few people had been co-opted by the Committee in a non-voting capacity.

Gert-Jan noted that it was understandable that there was a desire to maintain some continuity to the Committee, but that this runs the risk of the Committee only possessing a limited number of views. Heather responded that while there was no maximum limit to the number of terms an individual could serve, any one term is capped at four years. However she noted that in the first term of the full Committee, the constitution requires six individuals to retire and their positions filled by another election so that terms are staggered.

In response to a question from Arleen, Heather clarified that the time served on the current interim Committee does not count towards a term limit for the full Committee if a current member was to be elected to the full Committee.

Arleen further queried if the Board is being reconstituted at the same time. Heather responded that the constitution requires the interim Board to be replaced by the full Board by the 31st of December as well. In the New Year, the full Board will be asking the full Committee to nominate its representatives to the Board.

Sir John asked what the timescale for this process was. Heather responded that by the 31st of December, the full Board and the full Committee have to be in place.

**ACTION:** Committee members to nominate for election to the new Committee by filling the nomination form and have it seconded and then returned to [returningofficer@variome.org](mailto:returningofficer@variome.org)

## 5. Recommended System Status

### *a. Applications under Review*

- i. Application for Recommended System Status - Mutalyzer – Tim Smith
- ii. VarioML – Tim Smith
- iii. LRG as a reference standard for variant reporting – Tim Smith.

Timothy Smith reported to the Committee that the ICO was still working on finding suitable reviewers for these applications and asked Committee members to advise the office of any potential reviewers.

**ACTION:** Committee members to send names of potential reviewers to Timothy Smith ([tim@variome.org](mailto:tim@variome.org)).

### *b. New Applications*

None received since last meeting

### *c. Proposal for modification of review process – Tim Smith*

Timothy Smith spoke to a memo he had prepared for the Scientific Director and that had been shared with the Committee about the problems the ICO had encountered in implementing the Recommended System Status and a proposal for modifying the proposal.

The Committee were generally receptive of the proposal and asked the ICO to work up a more thorough treatment of the new concept for Recommended System Status for discussion at the next meeting.

## **6. Activity Proposals**

### ***a. Diagnostic grade VCF specification – Tim Smith***

Timothy Smith spoke to the Activity Proposal that had been submitted by Graham Taylor and expanded on the ICO comments contained in PP-ICO-10-2013.

Arleen Auerbach asked if the ACMG were working on a similar idea. Timothy Smith responded that the ICO had no knowledge on ACMG activities in this area, but that they are aware of a number of activities in this space, particularly those of Illumina and the CDC, and that the activity being proposed was for the Human Variome Project to facilitate collaboration amongst these groups.

Mauno Vihinen remarked that he believed that this activity was a good thing for HVP to do. He further noted that GEN2PHEN had a lot of experience in standards development work and what they discovered was, for it to be successful, the major players have to be involved in the development work and not just expected to adopt something they had no say in producing.

Arleen remarked that entities such as EBI and NCBI are better funded and have more experience in this type of activity. She asked how the Human Variome Project is the best party to lead this activity. Mauno noted that the Human Variome Project is regarded as an impartial broker. Gert-Jan remarked that the Human Variome Project being involved would be good, but also wondered if the Project was best placed to lead this. He noted that the investment and the work are going to come from the other people, and wondered how they might react to us claiming stewardship.

The Committee determined that the planned meeting in Boston should continue and they would make a decision on how to proceed based on the discussion there.

## **7. ISAC Sponsored Working Groups – Reports**

Reports from the Working Groups were noted by the Committee.

## **8. HVP relationship with other entities**

### ***a. IRDiRC***

#### ***i. Representation on IRDiRC Diagnostic Committee***

Johan den Dunnen has agreed to be the HVP representative on the IRDiRC Diagnostics Committee.

### ***b. Global Alliance – Heather Howard***

The ICO has requested a meeting with the Global Alliance during ASHG in Boston. Garry Cutting will be attending this meeting.

## **9. BMJ Proposal – Richard Cotton**

Richard Cotton reported that the ICO has asked BMJ to come back with a proposal that will form the basis of the negotiations. He suspects that the holiday period means that the proposal will take some time to arrive.

## **10. Scientific Directors Report**

The Committee took note of the Scientific Director's report (see attached).

## **11. Other matters**

## **12. 2013 ISAC Meetings**

- September – Thursday 26th
- October – Face to Face ASHG Boston Wednesday 23rd
- November – Thursday 21st
- December – Thursday 12th

Meeting closed 2302hrs AEST.

## Human Variome Project - Scientific Directors Report – August, 2013

### a. Meetings attended

#### i. 2013 ISMB/ ECCB Meeting, 21 July 2013, Berlin

Johan den Dunnen spoke at a session organised by Doron Lancet at the 2013 ISMB/ ECCB Meeting on 21 July 2013 in Berlin. The session was entitled “Human Disease Bioinformatics”. Besides Doron and Johan (who spoke on gene variant databases/ LOVD, his activities in HVP/HGVS), the other speakers were Warren Kibbe (Northwestern University, USA) whose presentation was on Disease Ontology, Alan Scott (John Hopkins University, USA) whose presentation was on OMIM, and Lars Jensen (University of Copenhagen, Denmark) whose presentation was on networks of proteins and diseases – STRING database to work towards systems-level analysis of disease.

We thank Doron for this opportunity.

### b. UNESCO update

#### i. International Decade for the Rapprochement of Cultures (2013-2022)

David Abraham submitted a letter to UNESCO for HVP (as an NGO Official Partner of UNESCO) to contribute to the elaboration of the Action Plan for the International Decade for the Rapprochement of Cultures (2013-2022) and to associate HVP with this important Decade. In his letter to UNESCO, David pointed out the role of Science and of HVP in joining many countries as part of our quest.

#### ii. Participation in the life of the NGO/ UNESCO Liaison Committee in 2013

Julia Hasler, the HVP Liaison Officer for UNESCO, will represent HVP in the activities of the NGO/UNESCO Liaison Committee. She will also be attending the Quality Education Forum on 23 September 2013 in Paris.

#### iii. 8th Session of the Intergovernmental Bioethics Committee (IGBC), UNESCO, Paris, 5th to 6th September 2013.

Julia Hasler will represent the HVP as an observer at the 8<sup>th</sup> session of the Intergovernmental Bioethics Committee (IGBC) at UNESCO headquarters from the 5<sup>th</sup> to the 6<sup>th</sup> September 2013.

**c. Assisting Nepal/ Dr Tilak Shreshta**

For some years, we have been anxious that HVP has not been able to assist Dr Tilak Shreshta in Nepal and the HVP Nepalese Node with genetic analysis and its documentation. We are delighted that with some publicity from the HVP, there have been many generous offers of help. We really thank those people and it looks like Tilak will be working with Dr Zilfalil bin Alwi from Malaysia at least.

**d. New HVP Country Node Application – Korea**

The Republic of Korea (South Korea) has formally registered as an interim member of the Human Variome Project International Confederation of Countries Advisory Council. Dr Myungchull Rhee (President of the Genetic Society of Korea) and Dr Jong Bhak (Director, Genome Research Foundation; Director, Variome Data Center, National Center for Standard Reference Data, Ministry of Trade, Industry, and Energy (MOTIE)) are the nominated representatives on the council.

**e. Upcoming Meetings**

- OESO 12th World Conference on Cancers of the Oesophagus, UNESCO Headquarters, Paris, France, August 27-30, 2013. R Cotton – Co President. I am honoured to be one of the 3 co-Presidents for the OESO conference this year. The aim of the organiser was to inject some informatics into the meeting. I am the organiser of the following sessions: Grand Forum 2 on the HVP and Symposium 4 on genetic data collection and library (databasing).
- InSiGHT Annual Meeting – Cairns Australia – 25th 28th August, 2013, includes Joint HVP day.
- Human Genome Variation Society: “Clinically Relevant Variants: Identifying, Collecting, Interpreting, and Disseminating”, ASHG Satellite meeting, Boston 22<sup>nd</sup> October, 2013 - HVP Co-Organizer
- ASHG, 22nd – 26th October, 2013, Boston –
  - o Booth
  - o ISAC Meeting
  - o Editors Interest Group Meeting

- Sequence Variation Description Working Group Meeting
- Medica Conference, November 20-22, 2013, Dusseldorf – John Burn will be speaking.
- We will be holding an "HVP Country Node Development Workshop" on the 20th of November as a satellite meeting of the 3rd Congreso Latinoamericano De Genética Humana (RELAGH) and the 38th Congreso Nacional De Genética Humana (AMGH) meetings (20-23 November, 2013).
- Japan Society of Human Genetics Meeting 2013, Sendai, Japan, November 21-23, 2013. Richard Cotton invited speaker. It was hoped that through the meeting, a HVP Japanese Node can be initiated and for Japan to become a core member of HVPI .
- INFRAFRONTIER - IMPC - IKMC Rome Meeting, December 2-4, 2013 – Richard Cotton invited to speak. Among the invited speakers for this meeting are: Kym Boycott, Paul Lasko and Peter Robinson.
- Indian Society of Human Genetics Conference 2014, Ahmedabad, India, January 23-25, 2014. Richard Cotton invited speaker. It was hoped that through the meeting, a HVP Indian Node can be initiated and for India to become a core member of HVPI .
- 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 5 Biennial Meeting, UNESCO headquarter, Paris, May 19-23, 2014.