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## MINUTES

Interim Scientific Advisory Committee Meeting  
Thursday 12<sup>th</sup> December, 2013  
11.00PM Melbourne, Australia time

### MEMBERS

Arleen Auerbach (USA)

John Burn (UK)

Marc Greenblatt (President HGVS, USA)

Finlay Macrae (Australia)

Mauno Vihinen (Sweden)

Richard Cotton (HVP Scientific Director, Australia)

Heather Howard (HVPI Operations Manager)

Timothy D. Smith (HVPI Communications Officer)

Helen Robinson (HVPI Liaison Officer for WHO)

#### **Apologies:**

Garry Cutting (Chair, USA)

Mireille Claustres (France)

Johan den Dunnen (The Netherlands)

Aida Falcon de Vargas (Venezuela)

Richard Gibbs (USA)

Mona El Ruby (Egypt)

Yoichi Matsubara (Japan)

Gert-Jan van Ommen (The Netherlands)

Ming Qi (Head China Node Partnership, China)

Stephen Lam (President IFHG, Hong Kong)

### AGENDA

#### 1. Welcome

#### 2. Apologies

#### 3. Confirmation of minutes of previous meeting

Minutes from the 23rd October 2013 meeting were approved.

#### 4. Issues arising from previous meeting

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

Discussion was postponed until next meeting.

#### 5. Recommended System Status

##### *a. Applications under Review*

##### *i. Application for Recommended System Status - Mutalyzer – Tim Smith*

Reviewer reports were presented to and discussed by the Committee. The Committee approved the award of Recommended System Status on the proviso that the Mutalyzer development team makes explicitly clear where Mutalyzer does not fully comply with HGVS nomenclature guidelines.

**ii. *VarioML – Tim Smith***

The ICO is still working on recruiting suitable reviewers for this application.

**iii. *LRG as a reference standard for variant reporting – Tim Smith***

Reviewer reports were presented to and discussed by the Committee. The Committee approved the award of Recommended System Status.

**b. *New Applications***

None received since last meeting

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

No further discussion on this item was deemed necessary following the outcome of discussions from the previous meeting.

**6. *Activity Proposals***

No Activity Proposals have been received in this period.

**7. *Redefinition of HVP Country Node – Tim Smith***

The Committee members were advised of the progress of this activity being undertaken by the ICCAC. The proposal was passed at the November meeting of the ICCAC and will take effect from the 1<sup>st</sup> of January, 2014.

The Council has modified the classes of membership for the ICCAC. A new class, Observer Status, has been created to allow countries that are not yet in a position to create a Country Node to participate. The definition of a Country Node has been expanded to include an operating organization and a set of policies for collection, storage and sharing, in addition to a data repository. While it may make it slightly more difficult for Country Nodes to be established, it makes what they need to do much clearer. It also creates a need to determine some standards around the quality of data held by a Country Node.

Timothy Smith and Helen Robinson used the new definition at an HVP Country Node Development Workshop in Mexico in November and it was very well received.

John Burn commented that it was essential that Country Nodes operate as an integrated part of the health system so that it is clear that the data contained in them has been generated by accredited diagnostic laboratories. Helen Robinson mentioned that as part of HVP's discussions with WHO, integration of genomic knowledge sharing into health systems will be a big component of future work with them and a key requirement of becoming a Full HVP Country Node. Part of the requirement of signing up to be an Interim HVP Country Node is a commitment to move a country in that direction.

The Committee discussed the issue that a large number of countries don't have formalized processes within their health systems to accredit laboratories or individual tests. The HVP may have a role to play in assisting countries establish these processes and incentivizing their establishment.

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

**a. *Global Alliance***

Heather Howard informed the Committee of the outcome of a meeting between HVP representatives (Garry Cutting, Michael Watson, Heather Howard and Timothy Smith) and representatives of the Global Alliance (Peter Goodhand and David Altschuler) during the ASHG meeting in Boston. The Global Alliance does not yet have a great deal of infrastructure in place and are working on developing this. There was a general agreement that the two groups should collaborate, but it was left unclear about how this collaboration would work. Further thought will be given to this by the ICO and ISAC members. It is understood that several ISAC members will be approached by the GA in their own right.

b. HUGO

Timothy Smith reminded the Committee that there was a meeting of the nomenclature working group in Boston. Stylianos Antonarakis is on this working group as a representative of HUGO and HUGO has been invited to cosponsor this working group.

c. UNESCO

Nothing to report.

**9. Comments on NIH genomic data sharing policy**

The HVP response to the draft NIH sharing policy has been received by the NIH.

**10. HVP Interpretation Group and ACMG Interpretation of Sequence Variations Committee**

Marc Greenblatt provided an update on the U401 grant that has been funded to create a clinically relevant variant resource. This involves both interpretation of variants and the informatics to share them. The group, now called ClinGen, has been really active and there is a lot of good will towards HVP within the group. They are still identifying an official liaison. Plan is to share any standards developed by the ClinGen group with HVP for ratification and discussion.

**11. HVP 5 – Helen Robinson**

Helen Robinson informed the Committee that the ICO will be canvassing the Committee during January for their top strategic priorities for the HVP to inform the development of the meetings around HVP5.

**12. Scientific Directors Report**

The committee noted the Scientific Director's report.

**13. Other matters**

Nothing discussed.

**14. 2014 ISAC Meetings**

New committee members will be polled for appropriate dates and times.

## **Human Variome Project - Scientific Directors Report – November, 2013**

The last Scientific Advisory Committee meeting was held during the ASHG meeting in Boston where the HVP also hosted, co-hosted or attended many key meetings and facilitated the HVP booth. All meetings were judged highly successful.

Joint satellite meeting with HGVS on “Clinically Relevant Variants: Identifying, Collecting, Interpreting, and Disseminating”

Helen Robinson and Tim Smith left on November 18th for the South and Central American Human Genetic Society Meeting in Cancun. Aida Falcon de Vargas is arranging HVP sessions including a satellite meeting to discuss the formation of HVP Country Nodes.

Richard Cotton has been invited to speak on HVP at the Japanese Society Human Genetics November 21-23rd. The formation of the Japanese Node is expected to be discussed at this meeting.

Sir John Burn has been invited to speak on HVP issues at the World Science Forum in Rio November, 24-27 and again we regard this as an honour and thank UNESCO for their assistance with this. Casimiro Vizzini will also attend on behalf of UNESCO to present the exhibition of the 60 years of DNA.

Casimiro Vizzini will attend and present at the Portuguese Genetic Society Meeting in November where they will discuss the formation of a HVP Country Node.

We are pleased to announce that Nigeria has formally joined as a node of HVP.

Finally Chris Arnold represented the HVP board at the initiation of the South East Asian node of HVP. Countries represented in Malaysia were Vietnam Thailand Sri Lanka and Singapore. We thank Zifalil bin Alwi for his energetic pursuit of this objective