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# MINUTES

Interim Scientific Advisory Committee Meeting  
Thursday 9<sup>th</sup> May, 2013 - 11PM Melbourne, Australia time  
Check your time zone at <http://short.variome.org/isac-meeting-time>

## ATTENDEES

John Burn (UK)  
Arleen Auerbach (USA)  
Mona El Ruby (Egypt)  
Mauno Vihinen (Sweden)  
Richard Cotton (HVP Scientific Director, Australia)  
Heather Howard (HVPI Operations Manager)  
Tim Smith (HVPI Communications Officer) for 30 mins

### Apologies

Garry Cutting (Chair, USA)  
Mireille Claustres (France)  
Johan den Dunnen (The Netherlands)  
Aida Falcon de Vargas (Venezuela)  
Richard Gibbs (USA)  
Marc Greenlatt (President HGVS, USA)  
Finlay Macrae (Australia)  
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 - John Burn Chair in the absence of Garry Cutting

### 2. Apologies

### 3. Confirmation of minutes of previous meeting

- a. 28<sup>th</sup> February, 2013 - moved Arleen Auerbach - unanimously accepted
- b. March, 2013 – moved John Burn – unanimously accepted

### 4. Issues arising from previous meeting

- a. Establishment of an Ethics Committee

Richard Cotton gave background to this issue and spoke about discussion he had had with HUGO, and suggested that we explore a joint HUGO/HVP ethics committee. Ruth Chadwick who heads the HUGO Ethics Committee and the new president of HUGO, Stylianos Antonarakis, are in favor of this as HUGO would like to have closer ties with HVP.

- b. Database Quality Interest Group

Mauno Vihinen provided the background to the proposal. The ISAC accepted this activity as a working group to develop the standard and determined that the Gene/Disease Specific Database Advisory Council should be the Sponsoring Council for this activity.

- c. HVP statement in line with ACMG statement

“Ensuring the Free and Open Sharing of Clinically Relevant Genome Variants”

The Committee asked for the statement to be opened to public consultation amongst the Consortium with comments received to be discussed at the next meeting in June.

A statement on incidental finding needs more thought and will be discussed at the June meeting.

- d. Publishing of short reports (BMJ) - R Cotton

The committee endorses this in principle so it can move forward however it should not in any way conflict with UNESCO or potential WHO relationships or with the work that Myles Axton is doing on microattribution. We must also make sure that a submission is credited as a publication.

## 5. Applications for Recommended System Status

- a. Applications under Review

- i. Application for Recommended System Status– HGVS variation nomenclature – Accepted
- ii. Application for Recommended System Status - Mutalyzer – Still under review
- iii. Application for Recommended System Status - LOVD – Accepted

- b. New Applications

- i. VarioML – accepted by committee, to be sent for review

## 6. Activity Proposals

- a. None received since the last meeting - however see 4b re new working group.

## 7. ISAC Sponsored Working Group - Reports

- a. Assigning Pathogenicity to a Genetic Variant – Marc Greenblatt (Chair) – none received
- b. Minimal content for a gene variant database – Peter Taschner (Chair) – none received
- c. Sequence Variant Description Committee – Johan den Dunnen (Chair) – none received

## 8. Scientific Directors Report

Attached – unanimously accepted

## 9. Meetings and Events

IRDIRC Meetings – Tim Smith spoke about IRDiRC meetings.

He stressed it was important we work together to eliminate the possibility of duplication of efforts and that this should be readily possible.

John Burn let it be known that he was looking at the possibility of establishing a European office of HVP via RD-Connect funding in his Centre.

## 10. China Node Funding

The letter from Professor Li was received by the committee. Heather Howard confirmed that Professor Li will be attending the Paris Meetings and that he has indicated that he is trying to send funds before then.

## 11. International Confederation of Countries Advisory Council

It was reported that Martina Witsch-Baumgartner has accepted the role of interim Chair until the next Council elections at HVP5 in May 2014 and Andrew Devereau has accepted the role as alternative Chair. The members expressed their appreciation for them stepping in to these roles.

John praised the efforts of the ICO in handling the difficult situation regarding the resignations of Fahd and George. He also expressed the wish of all for both Fahd and George to be able to reconsider their positions in the future.

R. Cotton stated "How To" papers were being developed for country Nodes and GDSD initiatives.

## 12. Other matters

Genome Centre for New York starting with a grant for cooperation.

## 13. 2013 ISAC Meetings

- June face to face meetings will be held at UNESCO in Paris 7 & 10th June, 2013
  - Scientific Advisory Council Meeting - 10th June, 2013.
  - International Confederation of Countries Advisory Council Meeting - 7th June, 2013
  - Gene/Disease Specific Advisory Council Meeting – 7th June, 2013
  - UNESCO Ambassadors Information Seminar – 10th June, 2013 followed by a Cocktail Party in the evening.
- July – Thursday 25th
- August – Thursday 22nd
- September – Thursday 26th
- October – Face to Face ASHG Boston Wednesday 23rd
- November – Thursday 21st
- December – Thursday 12<sup>th</sup>

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

### **a. Consortium Members**

There are currently 1018 Consortium Members from 72 Countries

### **b. New & Potential Country Nodes**

Venezuela has now joined as a Node. Thank you to Aida Falcon de Vargas for your hard work in making this happen.

### **c. Database Selection Sub-Committee**

A draft of the “Process for establishing and assisting International Gene/Disease Specific Databases (LSDB’s) by China” has been reviewed for a second time by the sub-committee and the changes have been recirculated it is due to be finalised.

### **d. Countries Development Program**

The applications from Austria and Belgium have been sent to the panels for review and both have made the decision to fund these projects. Funding agreements have been drawn up and the agreement is waiting to be finalised. No other application has reached the point of being put to a panel. Other countries and regions are still in the process of putting forward applications.

### **e. Country Council (ICCAC)**

A meeting of the ICCAC was held on the 19<sup>th</sup> February. The draft minutes of which are available on the website. A meeting is now scheduled for every 2 months.

On the 9<sup>th</sup> March Fahd Al Mulla the Chair of the ICCAC sent a letter to the Council resigning his position as he felt that he was unable to continue in his role as he was having a strong disagreement with the way the HVP is being run. This also sparked a letter from George Patrinos from Greece who said he felt the same. To clear the air and to provide a forum for any grievances that members of the ICCAC might have a special meeting was called and held on the 19<sup>th</sup> March

The HVP Country Node Baseline Report has been sent to 42 countries and 11 replies have been received to date. We are still following up on some outstanding reports which have been promised. The report will be finalised over the next few months and we hope will be presented in Paris.

Richard Cotton has drafted a ‘how to start a country node’ document which is now being revised before being sent to the ISAC for approval.

**f. Gene/Disease Database Council (G/DSDAC)**

A meeting was held on the 13<sup>th</sup> February, 2013 with Raymond Dagleish, Chair of the G/DSAC. Draft minutes of this meeting are available on the website. A full meeting is scheduled to take place on the 3<sup>rd</sup> April with meetings to follow every 2 months.

The council currently has 1 Working Group under its purview “Disclaimer Statement on LSDB websites.”

The ICO is working towards increasing the membership of the Council with a new mail out to known non HVP databases is underway.

Richard Cotton has drafted a ‘how to start a Gene/Disease Database’ document which is now being revised before being sent to the ISAC for approval.

**g. UNESCO**

Work is continuing on the Meetings to be held in Paris surrounding the celebrations of the discovery of DNA and the Mapping of the Genome to be held 5 – 10<sup>th</sup> June, 2013 together with our scheduled committee and council meetings.

**h. World Health Organisation**

More telephone interviews were conducted on what future role the WHO may play in the future of Global Genetic Healthcare. Larger group consultancy’s will be held at the HUGO, Mutation Detection, Canada and ESHG meetings.

**i. Meetings and Events attended**

***Meetings at which the HVP has been involved***

- ICCG Reception at ACMG, Phoenix Convention Center, Arizona, USA, March 20. Arleen Auerbach represented HVP.
- ACMG Laboratory Quality Assurance Committee and the Interpretation of Sequence Variations Workgroup, Phoenix Convention Center, Phoenix AZ, March 22. Arleen Auerbach represented HVP.
- BioVision – Lyon, France, 24-26th March – Invitation to present HVP by R. Cotton in the Catalyser section.
- 6th International Biocuration Conference, Churchill College, Cambridge, UK, April 7-10. Richard Cotton Invited speaker.

- Human Variome Project Workshop/Session at Human Genome (HUGO) Meeting/ International Congress of Genetics 2013, Singapore, April 16. R. Cotton, Helen Robinson and Graham Taylor attended
- IRDiRC Meeting – Dublin, Ireland, 16-17 April – Garry Cutting and Tim Smith attended
- IRDiRC 'RD-SymPathI Meeting – Dublin, Ireland, 18-19 April – R Cotton on organising committee and Tim Smith attended, other HVP members invited.
- Health Informatics Society of Australasia (HISA) Big Data 2013 Conference – “Big Data: addressing the challenges of the data deluge in health”, Rydges Melbourne, Australia, April 18-19. R Cotton Organizer (member of Scientific Program Committee).
- 12th International Symposium on Mutation in The Genome (International Mutation Detection Meeting), Lake Louise, Canada, April 22-26 R Cotton Organiser

***Other meetings and events for 2013***

- 2013 International Collaboration for Clinical Genomics (ICCG) Conference, Bethesda Marriott (Pooks Hill), Bethesda MD, USA, May 9-10. Garry Cutting and Tim Smith attending
- HVP Scientific Advisory Committee Meeting - UNESCO Headquarters, Paris, France, 10<sup>th</sup> June, 2013.
- HVP International Confederation of Countries Advisory Council Meeting, UNESCO Headquarters, Paris, France, June 7th.
- HVP Gene/ Disease Specific Advisory Council Meeting, UNESCO Headquarters, Paris, France, June 7th.
- UNESCO Headquarters, Paris, France, June 10<sup>th</sup>, 2013 - UNESCO/HVP Celebration of the discovery of DNA (60 years), the completion of the sequencing of the Genome (10 years) and the UNESCO declaration on Genetics (10 years).
- Human Genome Variation Society: Clinical Applications of Next Generation Sequencing, ESHG Satellite meeting, Paris, France, June 8. HVP Co-Organizer
- ESHG, 8<sup>th</sup> – 11<sup>th</sup> June, 2013, Paris –
  - Booth
  - WHO Consultancy lunch session
- OESO 12th World Conference on Cancers of the Oesophagus, UNESCO Headquarters, Paris, France, August 27-30. R Cotton – Co President.
- InSiGHT Annual Meeting – Cairns Australia – 25th 28th August includes Joint HVP day - T. Smith speaker

- Human Genome Variation Society: Clinical Applications of Next Generation Sequencing, ASHG Satellite meeting, Boston 22<sup>nd</sup> October - HVP Co-Organizer
- ASHG, 22nd – 26th October, 2013, Boston –
  - Booth
  - ISAC Meeting
  - Editors Interest Group Meeting,
  - WHO/HVP consultation