



sharing data · reducing disease

an NGO Official Partner of UNESCO

MINUTES

Interim Scientific Advisory Committee Meeting
Thursday 26th September, 2013 – 10.00PM Melbourne, Australia time

MEMBERS

Garry Cutting (Chair, USA)
Arleen Auerbach (USA)
Mauno Vihinen (Sweden)
Mona El Ruby (Egypt)
Marc Greenblatt (President HGVS, USA)
Finlay Macrae (Australia)

Richard Cotton (HVP Scientific Director, Australia)
Helen Robinson (HVPI Liaison Officer for WHO)
Tim Smith (HVPI Communications Officer)
Alfreda Soetopo (PA to the Scientific Director, Australia)

Apologies

Heather Howard (HVPI Operations Manager)
Stephen Lam (President IFHGS, Hong Kong)
Gert-Jan van Ommen (The Netherlands)
Johan den Dunnen (The Netherlands)
Mireille Claustres (France)
Aida Falcon de Vargas (Venezuela)
Ming Qi (Head China Node Partnership, China)
Richard Gibbs (USA)
Yoichi Matsubara (Japan)
John Burn (Alternate Chair, UK)

AGENDA

Meeting Opened 2205hrs

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

1. Welcome

2. Apologies

Apologies were noted from Heather Howard, Stephen Lam, Gert-Jan van Ommen, Johan den Dunnen, Mireille Claustres and Aida Falcon de Vargas.

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

Garry Cutting reported that Sir John and he would be meeting on 20th or 24th October and also in November to discuss the direction that the ethics committee will take and also to discuss the next step forward in establishing the committee. The discussion will be based on the information that Richard Cotton (Dick) provided regarding ethics-matters that are related to HVP. The plan is to make suggestions to ISAC members after the discussions.

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

b. Guidance on nomination and election procedures for ISAC – Richard Cotton

Richard Cotton reported that a few nominations had already been received and encouraged all current ISAC members to nominate. All details on the 2013 ISAC elections are available on the HVP website (<http://www.humanvariomeproject.org/index.php/about/scientific-advisory-committee/2013-isac-elections>). The deadline for receipt of nominations, including position statements, biographical information is **31 October 2013**. All ISAC members are reminded again to nominate as soon as possible. To prevent conflict, there is no further discussion regarding ISAC nomination (i.e. no disclosure regarding who have nominated).

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

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 3 applications and asked Committee members to advise the office of any potential reviewers. Tim mentioned that each application is in need of 3 independent reviewers to comment on the usability of the system. Reviewers are asked to comment on 2 categories: (1) compliance with HVP standard and guidelines that have been published to date; (2) usability factor.

Finlay Macrae suggested Bryony Thomson and John Paul Plazzer as reviewers of Mutalyzer. Marc Greenblatt commented that he will provide Tim with names from HGVS as suggested reviewers for Mutalyzer. Mauno Vihinen also remarked that he had experience in using Mutalyzer and commented that VariO is in the process of becoming an application and will be sent to HVP ICO as a recommended system.

Arleen Auerbach asked what is the use of reviewing LRG as a HVP recommended system since it is already used as standard. Tim replied that reviews are needed on the system itself to give an indication of its ease of use – i.e. usability, how well-described in terms of documentation, level of support. Arleen commented that as she has been using LRG, she will send Tim her comments on the LRG application.

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

b. New Applications

None received since last meeting.

c. Proposal for modification of review process – Tim Smith

Timothy Smith will present the ISAC with a full proposal for discussion at the next meeting.

6. Activity Proposals

a. Standards for disease and phenotype descriptions in gene variant databases (LSDBs) (submission by Peter Taschner) – Tim Smith

Timothy Smith commented that as the Activity Proposal (AP) (attached to the meeting agenda) was received by the ICO while he was away on leave, so he had not the time to review it in detail yet and to prepare the ICO position paper on it but from his brief reading, he thinks that it is a good AP to be taken up by the ISAC. It is then up to the ISAC to charter the AP to a Working Group (WG). Tim suggested that either: (1) decision be made at the next meeting, by which time the ICO position paper on the AP would be ready; or (2) a decision be made at this meeting

to either send the AP to the Gene/ Disease Specific Database Advisory Council (GDSDBAC) or International Confederation of Countries Advisory Council (ICCAC) or to retain it within ISAC. Garry commented that the AP is tailor made for the GDSDBAC and so it should go there if deemed worthy. All agree that it is clear-cut and that the AP should be sent to GDSDBAC.

Garry asked what the AP is designed to do since it seemed to be rather generic. Tim agreed that it is generic but explained that further details could be developed more in a WG when it is chartered. Marc commented that it is the role of the sponsoring council to develop the AP into something more concrete.

Garry also asked on how the AP is going to coordinate with other groups regarding writing of phenotype data. Marc commented that the ICO do the coordination with the sponsoring council. Dick suggested that the different groups get together to reach consensus and coordinate together (via position or discussion paper) with the goal of achieving a single recommendation for a criteria (i.e. deriving phenotype data) that represent the global viewpoint. Garry agreed and mentioned that HVP can help provide the global viewpoint to leverage the US centric/ North America approaches so as to have recognition that go beyond the USA/ North America/ Europe. Garry will talk to a member from his Institution to get an update on the discussion and plans between the different phenotype groups and will forward the discussion to Tim.

Tim commented that at this stage, no individuals are named in the AP but presumed that Peter Robinson is on the forefront of Peter Taschner's thinking when Peter was writing the AP. Nevertheless, Peter Robinson will be a suggested name for inclusion in the ICO position paper. Garry mentioned that as John Carey is also developing a similar system, his name should also be a suggested to the committee for review. The ICO will thus suggest in the position paper names to be included and other groups to be involved, such as IRDiRC/ rare disease group. All agree that the AP should be coordinated with other related groups/ efforts.

Garry mentioned that there will be a face to face meeting regarding Phenotype involving Kym Boycott (co-Director of IRDiRC), but is unsure on where it will be. He will find out. Arleen commented that some of the issues could be ironed out face to face if the meeting is in Boston (as there will be a lot of people attending ASHG). Dick remarked that Johan den Dunnen is now on the IRDiRC Diagnostic Committee and that Kym Boycott informed him twice before that the main thrust of IRDiRC is to 'find new genes'. Garry commented that IRDiRC is also exploring about phenotype description and that they have lots of funding, therefore it would not hurt to talk with IRDiRC so as to be aware of what else is being done by the other groups.

ACTION: ICO to send the AP on Standards for disease and phenotype descriptions in gene variant databases (LSDBs) to GDSDBAC and to provide the ICO position paper on the AP.

Garry Cutting to give an update on the situation between different phenotype groups and to find out the date of the next IRDiRC meeting re phenotype.

7. Redefinition of HVP Country Node – Helen Robinson

Helen reported that the country council (ICCAC) has been reviewing the definition of the country node for the past months and it is currently almost in the final stages. It is important that the ISAC be aware of this as the definition of HVP Country Node is important for the backbone work of HVP. The new definition of HVP Country Node consisted of 3 components: (1) data repositories – the new definition will be more flexible: rather than one data repository per country, there will be inter-related data repositories in collecting, storing and sharing data on variations in a country. (2) HVP Country Node to have governance structure for long term sustainability and to ensure that the node complies with the relevant national and international ethical, legal and social requirements. (3) HVP Country Node to have a set of policies and procedures to ensure that the repositories are maintained in a responsible and ethical manner that are consistent with HVP standards. Other than beefing-up up what the Country Node does, which is an important milestone, the ICCAC has lengthy discussions on how to assess the data quality in a Country Node i.e. whether data is: (a) to be of clinical standard or (b) to achieve clinical standard quality data over time, thus HVP needs to agree on which quality standard to measure and flag data against. Helen then asked if the ISAC is interested in seeing a draft proposal on the redefinition of HVP Country Node and if the ISAC is interested in commenting on the proposal and be involved in the discussion. All agreed that this exercise should be overseen by the ISAC.

8. HVP relationship with other entities

a. Global Alliance

i. Meeting in Boston

Garry commented that he had not heard much about Global Alliance (GA) in the USA. Helen reported that GA have just communicated an update in September with a list of partners (of which HVP is one) – i.e. they have set up a transitional committee and have appointed some staffing. The update by GA will be forwarded to the ISAC. Dick added that Martin Bobrow and Kathryn North and a couple of clinicians are included on the GA steering committee. Dick mentioned that a meeting in Boston with GA (David Altshuler and Peter Goodhand) has been set up on 24 October from 3 to 5pm to discuss the plans between HVP and GA. Garry expressed interest in attending, and mentioned that he will also try to find out the plans of the 3 groups (involving Mike Watson, Heidi Rehm/ Nussbaum (U41) and others) that were funded the recent NIH Clinically Relevant Variant Resource grants regarding GA. Garry is of the impression that they will be setting up standards (via internet committee) on data exchange that is more IT/ bioinformatics oriented rather than actual grassroots data collecting. Marc suggested that GA representatives be invited to the HGVS/ HVP annual meeting in Boston on 22 October and mentioned that those involved in the NIH grants such as Heidi Rehm, Carlos Bustamante, and Jonathan Berg will be there. Finlay Macrae requested that Garry's conversations in Boston highlighted the point that InSiGHT has offered its interpretation committee to the U41 grant application done by Heidi to assist in the interpretation of Variants of Uncertain Significance (VUS) in the pipeline between ClinVar and private labs in the USA.

b. HUGO – Helen Robinson

No further update at this time.

c. WHO – Helen Robinson

Helen reported things are progressing well with WHO and that she will be in Berlin in October with WHO at the World Health Summit to announce a new program that will be launched in April 2014. Helen will circulate further information on this.

d. UNESCO

See item 10 below. Dick also mentioned that John Burn will be presenting on HVP at the UNESCO-hosted World Science Forum in Rio de Janeiro in November.

9. BMJ Proposal – Richard Cotton

Richard Cotton reported that the ICO is still waiting for a proposal from BMJ and that he will be having a teleconference regarding this with Daniel Schorderet and Janet O'Flaherty (BMJ) in the next week.

10. HVP 5

a. African Satellite – Helen Robinson

Helen reported that we are currently proposing, with the assistance of both WHO and UNESCO, to host a meeting on the Monday before HVP5 with the H3Africa countries (H3Africa is a Wellcome Trust and NIH funded initiative of about 15 countries on the African continent who are developing genomic capacity with the hope of setting up databases). These countries will be invited to UNESCO to meet with their country delegation as well as with WHO to see if anyone is interested in establishing a HVP country node. There are positive responses from Nigeria and South Africa at this stage, and it is hoped that some of these African researchers will submit abstracts to HVP5 so that they can share their interesting research around the topics of infectious disease, cardiac disease etc. HVP 5 will highlight the research that is going on around these countries and try to promote the exchange between

developed and developing countries. Helen also mentioned that Raj Ramesar is going to raise the issues with the Africans as they are having a big meeting in the 1st week of October in Johannesburg.

11. Scientific Directors Report

Nothing to report at this time as the Scientific Director was away on leave for most of September. A list of upcoming meetings is attached for the information of the ISAC.

12. Other matters

Dates of upcoming relevant meetings are attached.

13. 2013 ISAC Meetings

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

Meeting closed 2258hrs AEST

Upcoming Meetings

- World Health Summit, Berlin (20-22 Oct) – Helen Robinson is an invited speaker.
- Human Genome Variation Society: “Clinically Relevant Variants: Identifying, Collecting, Interpreting, and Disseminating”, ASHG Satellite meeting, Boston 22nd October, 2013 - HVP Co-Organizer
- ASHG, 22nd – 26th October, 2013, Boston –
 - o Booth
 - o ISAC Meeting (23 October)
 - o Meeting with Global Alliance representatives (David Altshuler, Peter Goodhand) on 24th October 2013 (3-5pm)
 - o Editors Interest Group Meeting
 - o Sequence Variation Description Working Group Meeting, 25 October
- 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 .
- World Science Forum, hosted by UNESCO in Rio de Janeiro, Brazil, November 24-27 – John Burn will be speaking on HVP.
- INFRAFRONTIER - IMPC - IKMC Rome Meeting, December 2-4, 2013. Among the invited speakers for this meeting are: Kym Boycott, Paul Lasko and Peter Robinson. Johan den Dunnen is an invited speaker and will be representing HVP at the meeting.
- 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.