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MINUTES

Interim Scientific Advisory Committee Meeting
Wednesday 7th of November, 2012
Room 270, West Mezzanine Level, Moscone Center, San Francisco, USA
12.45pm – 2.15pm

ATTENDEES

Garry Cutting (Chair – USA)	Gert-Jan van Ommen (The Netherlands)
Arleen Auerbach (USA)	
Johan den Dunnen (The Netherlands)	Richard Cotton (HVP Scientific Director, Australia)
Aida Falcon de Vargas (Venezuela)	Heather Howard (HVPI Operations Manager)
Marc Greenblatt (President HGVS, USA)	Tim Smith (HVPI Communications Officer)

APOLOGIES

John Burn (UK)	Finlay Macrae (Australia)
Mireille Claustres (France)	Yoichi Matsubara (Japan)
Mona El Ruby (Egypt)	Ming Qi (Head China Node Partnership)
Richard Gibbs (USA)	Mauno Vihinen (Sweden)
Stephen Lam (President IFHG, Hong Kong)	

AGENDA

1. Welcome

Garry Cutting welcomed members to the meeting and expressed his sadness at the passing of David Rimoin.

2. Confirmation of minutes of previous meeting

Passed unanimously

3. Issues arising from previous meeting

a. HVPI Board moving ahead with establishing an ethics committee

This committee will be only for oversight of the company ethics. It is suggested that the ISAC set up an Ethics committee for the Project.

- The Committee was reminded that the Human Variome Project has an Ethics Interest Group that could be drawn on.
- Gary Cutting raised the point that it was important to have an independent, internationally constituted group that is across the ethical issues that we sometimes forget as scientists.

- Arleen Auerbach reminded that Committee that different countries have different approaches to ethical and legal issues.
- Gert-Jan van Ommen raised the issue that few people have the necessary expertise for this and as other groups already have similar committees, do we really need to set up a new committee. Can ask committees from HUGO, ESHG, ASHG etc. to act for us as appropriate, possibly all three. Gary Cutting recognized that a committee from a different organization is by definition independent, but that these committees may not be representative of the Human Variome Project's international constituency. The possibility of approaching WHO or UNESCO committees was raised. Timothy Smith advised the Committee that we have been informed that these organizations are unable to act in this manner, but can provide access to experts that we could approach.

ACTION: Gary Cutting to seek advice from various groups (Bruce Korf, Carol Barash, Sue Povey, WHO, UNESCO)

- b. ICO to seek clarification from the Board about casual vacancies

The conclusion of the Board is that the ISAC fill any vacancies from those co-opted to the committee.

- Johan den Dunnen, previously and alternate for Gert-Jan van Ommen, is now a full member to fill vacancy by passing of David Rimoin.
- John Burn nominated and elected *in absentia* to fill vacancy left by resignation of Christine Van Broeckhoven.

4. Applications for Warrants

- Gary Cutting first sought clarification from the Committee that this was an activity that the Human Variome Project should be undertaking. The Committee determined that it was.
- Marc Greenblatt asked whether these "recommendations" would be exclusive: i.e. recommending one system over another. The Committee determined that the recommendations would not be exclusive. Gary Cutting recommended that alternative terms for "Warrant" and "Recommendation" be explored so as not to sound exclusive. He further suggested that the review process could include assessing other systems than those that apply.
- The Committee determined that a process was needed to assess applications.
- Gert-Jan van Ommen proposed that the process be similar to that used by an academic journal: peer review by 3 people. He further argued that we should not necessarily wait for people to apply, that the Committee review systems, processes etc. that they select – similar to how a consumer group works.
- Arleen Auerbach proposed that the Human Variome Project only recommend free software.

ACTION: International Coordinating Office to draft a process reflecting the issues discussed and circulate for the next meeting

5. Activity Proposals

- a. Assigning Pathogenicity to a Genetic Variant (Marc Greenblatt)
- The ISAC will act as Sponsoring Council for this activity. The chartering of a Working Group was deferred until the next meeting.

ACTION: International Coordinating Office to draft Working Group Charter and circulate for the next meeting

- b. Minimal content for gene variant databases – LSDB's (Johan den Dunnen)

- The ISAC will act as Sponsoring Council for this activity. The chartering of a Working Group was deferred until the next meeting.

ACTION: International Coordinating Office to draft a Working Group Charter and circulate for the next meeting

6. Reports – ICO Staff

- a. New & potential Nodes
 - Currently 16 Nodes. International Coordinating Office exploring a number of prospects
- b. Database Selection Sub-Committee
 - Last met in Paris. A review of clinically relevant genes and their database needs was done by Johan den Dunnen. Currently seeking clarification from China about what options they can provide for support of databases.
- c. Countries Development Program
 - A number of applications have been received. Three look likely to move to the next stage. The committee should expect to be asked to assess applications in the coming weeks.
- d. Country Council
 - International Coordinating Office working with Countries Council to produce a report assessing genetics and genomics capacity in individual countries: the *HVP Country Node Baseline Report*.
- e. Gene/Disease Database Council
 - Nothing to report.
- f. UNESCO
 - UNESCO are providing a lot of support. Holding a joint event in Paris during ESHG to celebrate 60 year anniversary of the discovery of the structure of DNA, 10 year anniversaries of the completion of the Human Genome Project and the adoption of the International Declaration on Human Genetic Data. Will also hold meetings of the Committee and Councils at UNESCO at this time.
- g. World Health Organisation
 - Discussing holding a joint project in 2013 to explore the possibility of a global programme on medical genetics and genomics within WHO. ICO holding a consultation on Friday 9 November in this room as a part of this process.
- h. Meetings and Events
 - Upcoming meetings include a satellite and session at APCHG2012, Mutation Detection in Canada, a session at HGM2013 in Singapore, and of course ASHG and ESHG next year.

7. Other matters

- a. Clarification of the term of the ISAC
 - Board is extending the term of the ISAC and Board until the end of 2013.
- b. Alternate Chair
 - Gary Cutting called for nominations via email

c. Collaboration with other groups

- Gert-Jan van Ommen urged the Human Variome Project to seek stronger ties with other groups in similar areas, especially IrDiRC. He specifically noted the therapeutic committee of IrDiRC. Richard Cotton assured the Committee that this was being done.

8. Next ISAC Meeting

A Skype/Phone meeting to be arranged each month starting in December, 2012

The next face to face meetings will be held at UNESCO in Paris 5-7th June, 2013

- Scientific Advisory Council Meeting - 5th June, 2013.
- International Confederation of Countries Advisory Council Meeting - 6th 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.