

MINUTES

International Scientific Advisory Committee
Teleconference
Thursday 25 May 2017
1200hrs UTC/GMT



MEMBERS

Present

Voting Members

John Burn
Garry Cutting
Raymond Dalglish
Finlay Macrae
Juergen Reichardt
Mike Watson
Martina Witsch-Baumgartner

Non-Voting Members

Arleen Auerbach
Mona El Ruby
Ada Hamosh
Melissa Landrum

ICO Staff

Amy McAllister
Timothy Smith

Apologies

Voting Members

Jordan Lerner-Ellis
Ming Qi
Raj Ramesar
Peter Taschner
Mauno Vihinen

Non-Voting Members

Qasim Ayub
Mireille Claustres
Collet Dandara
Johan den Dunnen
Aida Falcon de Vargas
Marc Greenblatt
Helena Kaariainen
Dhavendra Kumar
Rita Ines Noher de Halac
Heidi Rehm
Augusto Rojas Martinez
Katsushi Tokunaga
Tom Weber
Ingrid Winship

AGENDA

- 1. Welcome & Apologies (noted as above)**
- 2. Confirmation of the record of the previous meeting** – there were no changes to the draft record circulated to members.
- 3. Up-date on recent events** - Co-Chairs to report

John Burn and Garry Cutting report they had a discussion via teleconference over the issue of branding.

Both chairs recognise the concerns of various committee members. The Human Variome Project is the recognised brand, which will not be dismissed. Thus, Global Variome is simply set up as holding company. The name Global Variome was needed in order to keep our UNESCO NGO status. It was also needed to distance ourselves from the organisation in Australia that was shut down, leaving with it its financial entanglements. Another positive additional benefit is the opportunity for a closer relationship to Global Alliance. HVP and other collaborating organisations, (eg. HUGO), can now be managed more efficiently under the GV holding group. Global Variome will simply facilitate interactions between each group. Both John and Garry plan to write a document with more details.

- 4. Operational Issues - Reports from Working groups etc**
 - a. Closer ties between HVP and IRDiRC** - Timothy Smith to cover topic

You will recall from the previous meeting March the discussion about reciprocal recognition of IRDiRC recommended systems and HVP recommended systems. The administrative matters have all been signed off by both sides now and our website has been updated to reflect the IRDiRC systems that we are providing recognition of. Tim has been informed that the IRDiRC website is in the process of being updated. We will continue to have a relationship with IRDiRC by keeping each other updated.

i. HVP representation on IRDiRC DSC

Previously HVP has been invited to appoint a representative to the IRDiRC diagnostic subcommittee. That role was filled by Johan den Dunnen but his term finished at the end of last year. They have just come back to us after prompting several times to ask if we wanted to appoint another member to that committee. A decision needs to be made on this soon on who the right person should be. Johan has not ruled himself out at this point, but he has made it clear that he thinks fresh ideas would be beneficial to both parties. He also stated that the work is limited, just one meeting a year and sometimes a teleconference combined with an organised meeting. ie. ESHG/ ASHG. Juergen Reichardt has put himself forward for nomination. All on the call agreed to this appointment.

ii. IRDiRC representation on ISAC

If IRDiRC are offering a seat of one of their committees, we might like to extend the same courtesy seat to them. The ISAC seems like the most obvious choice.

b. WG 10 – Proper Reporting of sequence variants in journal articles – held over from last meeting, Garry Cutting and Tim Smith to report

Garry Cutting has nothing new to report as he has assumed the sole editorship of Human Mutation. Human Mutation will be bringing in some deputy editors soon which will ease the workload. They have an outstanding Agreement with HGVS, the society we were working with; further discussions are needed regarding that relationship. Part of these discussions have already touched upon the process for sequence/ variant entry and we should produce more clear documents and forms that can be used as templates for other journals. Garry will start approaching journals as the editor instead of an HVP representative. A collation of editors might be a more effective way of approaching these guidelines that we are producing.

Tim Smith reports that progress by the working group has been made on producing the guidelines. They have identified the types of information that we believe should be included within a report of a sequence variant within an academic paper. Tim emailed the working group for their final thoughts, there were no responses so Tim will take that as confirmation that is the list to be used moving forward. The next project was to define some acceptable values for each of those attributes, this has almost been completed. Once it has been confirmed by the working group then the writing up of the text of the guidelines. At this point, journal editors should be more formally involved. Garry mentions that the lists may have to be modified to appease all journals if the editors are going to abide by these guidelines. Tim reports that if we use the current list of attributes and acceptable values against each one, by the next ISAC teleconference.

John Burn will be attending the ESHG Editorial board meeting on 29th May. John will present the latest version to the board and will report back on what is said at that meeting.

c. WG 08 - Checklist For Gene/Disease Specific Database Curators To Enable Ethical Data Management – Note that this document is currently being finalised by the Gene and Disease Specific Database Advisory Council and will be sent to ISAC in preparation for discussion and approval at the June meeting of ISAC.

Tim Smith reports that the council are confirming their draft for approval by email and it will be sent to the ISAC for discussion and approval at the June meeting.

5. Reports from Project-wide Initiatives

a. BRCA – report will be postponed until the next meeting. John Burn will report on his meeting with Global Alliance.

John Burn reports that as expected Ewan Birney is driving Global Alliance to be more of a big data science organisation and there is now a strong opportunity for us to be more visible. They still have the BRCA exchange as the primary driving project. He has raised with them the possibility of broadening that by making HVP a driver project for variant databases more generally. Not surprisingly, there was a muted reaction but they didn't refuse. John will write a document on how to integrate with them more formally. There is a real opportunity for us to migrate our work on the BRCA exchange for front end inter connectivity with the InSiGHT database and expand our work to include the mismatch repair genes in collaboration with InSiGHT. Software work has been done by David Hausler's group on the BRCA exchange, this could relate to the InSiGHT database as it relates to LOVD and ClinVar. This would make it more user friendly, people could come to a 'cancer exchange' website and access variants on both mismatch and on BRCA. This would be a migration to a broader front end software. As long as we keep LOVD at the front as a primary source, Johan is happy to combine with Global Alliance. This will in turn broaden our appeal and incorporate HVP much more into mainstream thinking of Global Alliance; providing more formal support for curation. It seems that Global Alliance's current restructuring and shifting objectives will be positive for HVP. John Burn will report further once Global Alliance have had time to reflect on their meeting. Ewan Bernie is confident that the restructure will strengthen Global Alliance finances and in turn should strengthen our position.

b. **GG2020** - report will be postponed until after meeting on 30 May

6. **Current Issues** – Nothing more for individual members to report at this time

7. **Other business** - No further business

- i. Finlay Macrae reports he has a teleconference coming up between the ClinGen Bowel Cancer Working Group and the Executives of Variant Interpretation Committee of InSiGHT to align our activities.
- ii. Members should be encouraged to attend the Global Alliance meeting immediately preceding ASHG in Orlando in October. We should make a conscious effort to integrate. Amy will circulate the information on Global Alliance leadership once they have changed their restructuring.

8. **Future meetings**

ISAC scheduled teleconferences for 2017:

- i. 22 June
- ii. 20 July
- iii. ~~24 August~~ **(It has been decided that this will be cancelled due to the holiday period)**
- iv. 28 September
- v. 19 October (in conjunction with ASHG – Orlando, Florida)
- vi. 16 November
- vii. 14 December