

MINUTES

International Scientific Advisory Committee
Teleconference Thursday 20th July 2017 1200hrs UTC/GMT
Check your time zone at <http://short.variome.org/isac-meeting-time>

MEMBERS

Present

Voting Members

John Burn
Garry Cutting
Jordan Lerner-Ellis
Raj Ramesar
Juergen Reichardt

Non-Voting Members

Arleen Auerbach
Johan den Dunnen
Rita Ines Noher de Halac
Heidi Rehm

ICO Staff

Amy McAllister
Helen Robinson

Apologies

Voting Members

Raymond Dalgleish
Finlay Macrae
Ming Qi
Mike Watson
Martina Witsch-Baumgartner
Peter Taschner
Mauno Vihinen

Non-Voting Members

Qasim Ayub
Mireille Claustres
Collet Dandara
Aida Falcon de Vargas
Mona El Ruby
Marc Greenblatt
Ada Hamosh
Helena Kaariainen
Dhavendra Kumar
Melissa Landrum
Augusto Rojas Martinez
Katsushi Tokunaga
Tom Weber
Ingrid Winship

AGENDA

1. Welcome John Burn chaired the call as Garry Cutting was delayed and joined the discussions part way through.

2. Confirmation of record of previous meeting – confirmed without comment

3. Up-date on recent events - Co-Chairs to report
Discussions with HGVS, HUGO, GA4HG

Discussions are continuing with these various partners; John Burn and Marc Greenblatt discussed informally opportunities for HVP and HGVS to work more closely together at the recent InSiGHT meeting. Closer ties between HVP and InSiGHT were also discussed informally as well.

There was Global Variome Board meeting in early July at which the HUGO conference in Yokohama was discussed. It was agreed that HVP would become a formal sponsor of the meeting as the basis for greater co-operation in the future. There will be an HVP session with four speakers with each covering a different aspect of HVP's work: Gunnar Rättsch to present on the BRCA Exchange with emphasis on developing the BRCA app; Carsten Lederer will present on GG2020 with emphasis on HVP and Thalassemia; Johan will present LOVD3; and Ingrid Winship has offered to talk about cardiac genetics with reference to Dhavendra Kumar's development of the Global Familial Heart Challenge for HVP.

Anyone with ideas regarding the content of the session should contact John Burn by email directly within the next few weeks. All HVP members are asked to make plans to attend this meeting in March 2018. Details will be provided via the HVP web-site.

Linking up with GA4HG meetings in Yokohama is still to be followed up, and should perhaps wait the release of the GA4GH strategic plan as it may make it more clear how the various cross-cutting themes proposed by the new Chairperson might provide opportunities for discussing the way ahead. Discussion of this followed and it was suggested that the most important issue to focus us on was suggesting concrete projects with named PIs as the basis of collaboration. It is understood that following the GA4HG April 2016 meeting in Kyoto that an announcement regarding the placement of Japanese data is likely to be made in Yokohama.

See also Item 4. f. below.

4. Operational issues - reports from working groups and other follow up

a. WG 07 – Joint Working Group on minimum content requirements for genomic variant databases –

As neither Martina nor Peter were on the call to report this will be held over for the next call.

b. WG 09 – Database Quality Accreditation Scheme Development – As Mauno was not on the call to report, this will be held over for the next call.

c. WG 10 – Proper Reporting of sequence variants in journal articles – up-date on discussions regarding EJHG policies– Johan, John, Garry

HVP has held a meeting of the Journal Editors each year at ASHG and all agreed that this meeting should continue as it is a productive exercise. Garry and Juergen will work with Amy in the ICO to contact the relevant Journal Editors and provide them with an invitation to the meeting. Details of the venue and the time need to be finalised as soon as possible. Any member with connections to any journal of interest and having suggestions of who to invite to this meeting should contact Amy directly with names as soon as possible.

d. ClinVar and LOVD3 – up-date on progress of database up-grade – Melissa L to report

Melissa was unable to make the call, however she did provide an up-date for the meeting indicating that regarding LVD3 and ClinVar, she is working with Johan to define a submission process that is both feasible for Johan and his team, and also gives proper attribution to each database and curator. They have not yet reached the stage of submitting data.

Heidi also added that the coordination is progressing slowly. This week LOVD has been given a submitter account which means that we can more accurately share the data. A general discussion of this followed indicating that these issues point to key decision making areas regarding the way forward for HVP. Exactly how to organize across networks of collaborators, how to link genotype and phenotype information across a range of diseases is not yet clear. A key decision point seems to be emerging as the role of the curator and whether or not the bulk of decision making should be done via a machine-generated process or logarithm as opposed to having greater emphasis on 'human' intervention of the curator. More effort needs to be applied to determining how to address this efficiently and cost-effectively. As well, the amount of time it may require to fully resolve this is not well understood.

It was also noted that some of the features of Ithonet, for example the mapping, might be worth considering as additional useful features for LOVD3 at some point.

e. RefSeq NCBI changes to procedures – Raymond D was not on the call to report, but he will be asked to circulate a short report up-dating this topic.

f. Plans for ASHG and HVP – up-date on plans, dates, meeting location etc

John encourages all to attend ASHG to meet with people at GA4GH. He will have a one-on-one conversation with Ewan Birney to discuss the relationship between GV and Global Alliance before ASHG. It seems that HVP should be mapping and documenting more effectively the tremendous efforts that are going on around the world in gene specific areas, for example, Garry's CFTR2 Project, InSiGHT and Arleen's work with Fanconi anemia. A lot of people are unaware these databases exist. HVP needs to publicise these critical efforts.

There will be a closed members' meeting at ASHG – details will be provided as soon as possible and registration details will be announce through the HVP web-site.

5. Reports from Project-wide Initiatives

a. BRCA – is making good progress and has been adopted as a driver project for GA4GH. HVP place in this is still to be clarified. GV/HVP could potentially take the lead in running the BRCA Exchange expanding that into other genes and bringing in other groups like InSiGHT and so on.

b. GG2020 – report on recent Malaysian meeting

Bin Alwi Zilfalil hosted a very successful meeting sponsored by the Malaysian Ministry of Higher Education. The Deputy Minister opened the meeting and a process for developing a formal memorandum of understanding with the government was established. The Malaysian government seemed very much engaged with the efforts of HVP and GG2020. Positive discussions with the government were focussed on the possibility of rolling out thalassaemia screening across sections of the Malaysian population. Patient groups were involved in the meeting and gave some excellent presentations. Malaysia is currently examining closely the approach used in Cyprus of systematically determining carrier status in adolescence and using this information to make informed decisions regarding reproduction. The Malaysian government has started to roll out Thalassaemia testing on all 16 year olds at school with consent from their parents. This makes it possible to put together a bid to assist by working with the Malaysian team to develop a point of care thalassaemia test for all the common variants in Malaysia.

It was also noted that there was a meeting of the American Society of Hematology (ASH) in December in Atlanta GA that has a focus on hemoglobinopathies in low and middle income countries. GG2020 has an invitation to meet the key officers of ASH through colleagues from NIH/ NHGRI following discussions at ESHG. These connections will be followed up with a view to hoping that GG2020 can be profiled at the December 2017 meeting.

5. Other business

There was discussion of the situation in South America regarding representation and formal links to HVP. As several members of ISAC are based in South America and aware of the situation, plus others will be visiting the region for meetings and workshops later in the year, it was felt that this can be managed in a positive way.

Planned technical meetings and conferences involving members for noting include:

- i. ASHG – 17 – 21 October 2017, Orlando USA
- ii. HVP: 2nd Variant Effect Prediction Training Course – 6 – 8 November 2017, Prague
- iii. A-PCHG – 8 – 10 November 2017, Bangkok Thailand
- iv. African Society of Human Genetics – 7 - 10 November Cairo, Egypt
- v. World Science Forum – 7 – 10 November Amman Jordan
- vi. HUGO - GVL meeting 13 – 15 March 2018, Yokohama Japan

ISAC scheduled teleconferences for remainder of 2017 –

- i. 28 September
- ii. 19 October (in conjunction with ASHG – Orlando)
- iii. 16 November
- iv. 14 December

