

MINUTES

International Scientific Advisory Committee
Teleconference
Thursday 22nd June 2017
1200hrs UTC/GMT
Check your time zone at: <http://short.variome.org/isac-meeting-time>



MEMBERS

Present

Voting Members

John Burn
Raymond Dalglish
Raj Ramesar
Juergen Reichardt

Non-Voting Members

Arleen Auerbach
Qasim Ayub
Mona El Ruby
Marc Greenblatt
Melissa Landrum

ICO Staff

Amy McAllister
Helen Robinson

Apologies

Voting Members

Garry Cutting
Jordan Lerner-Ellis
Finlay Macrae
Ming Qi
Mike Watson
Martina Witsch-Baumgartner
Peter Taschner
Mauno Vihinen

Non-Voting Members

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

AGENDA

1. **Welcome** Apologies, and Administrative Matters
2. **Confirmation of record of previous meeting** - no changes required to the second version of the draft record that was circulated.
3. **Operational Issues - Reports from working groups**
 - a. **WG 10 – Proper Reporting of sequence variants in journal articles** –John Burn raised this verbally with Johan at EJHG Editorial Board Meeting but has yet to hear from him. John will get in touch with Johan once more, asking him for the official journal policy, and report back at the next meeting.
 - b. **Letter in Science 26 May** – from Stylianos Atonarakis (Immediate Past President of HUGO) on role of WHO in databases

Helen Robinson reports that she has been in touch with Stylianos since this letter was circulated to ISAC members (see [Genomic databases: A WHO affair](#)) members earlier in the month. It suggests that WHO should take an active role in database management. Helen said that there was a history to the letter and she had written to him congratulating him on the letter and the issues it contained. She and Stylianos had discussed this in recent years. She had also sent the letter to HVP's contacts at WHO in Geneva who had already seen the letter and are planning to meet with Stylianos.

John Burn reports that he recently spoke with Charles Lee of HUGO. During that call, they discussed the role of WHO and agreed to share information with a view to strengthening the relationship with WHO. It should be noted that HVP already has a MOU with WHO, but has lacked resources to assist in funding the work plan outlined in the MOU. GG2020 has WHO as a partner as a result of this collaboration with WHO. Helen indicated that HVP has been trying to find two or three governments to contribute financially to the WHO program on human genomics and public health. She will keep ISAC informed regarding the response to Stylianos and the relationship with WHO.

c. Links between HUGO and HVP – joint meeting 15 March 2018, Yokohama Japan

John indicated that Charles Lee and HUGO are currently approaching the Korean government regarding support for their global outreach as their office is based in Seoul. John flagged with Charles the need to think about WHO as one potential area where sponsorship from a national government could lead to the creation of a formal relationship for both of organizations. This was received in a positive manner.

It was also noted that as previously advised, HUGO and HVP will organized their meetings concurrently in mid-March 2018 at Yokohama Japan. John is a plenary speaker at the HUGO conference. HVP will cover cost of HVP's invited speakers and their registration. HVP will organize its own meetings as a side meeting of HUGO. Planning for this is becoming urgent. The HVP ICO will put together a draft program for circulation and comment, consisting of a data sharing session, where Ewan Birney will also be an invited speaker, plus a session dedicated to our project-wide initiatives - BRCA, GG2020- and any other projects we deem worthy. Members should also pursue their own abstracts for the rest of the program. There will also be meetings of ISAC, ICCAC and LSDC for those members in Yokohama.

d. Up-date on HVP Country Nodes – item requested at April meeting – see attached papers for reference

- i. Helen Robinson reported that in response to questions raised concerning the recent progress of country nodes at the April ISAC meeting and in the absence of Martina, she would up-date ISAC members. Every 2 years, ICO through the ICCAC asks the nodes to report on their activities. At the June meeting of ICCAC a draft template for the next report was presented. The report up-date administrative arrangement and contacts, and asks for details on what types of data are held in national databases, who is involved in this and same for data going into curated international databases. It should be noted that Nodes do not follow exactly the same path; while the eventual place for all variant information in is international databases, not all countries can do this owing to the ethical and regulatory frameworks operational in their country. For example, Brazil is a very active and comprehensive node, with its own national database; they will follow a federated approach. This is common with many countries wanting to hold information nationally, particularly those where the public health sector tends to dominate. Countries with an active private sector face greater complexities. The purpose of country nodes will be discussed further at the InSiGHT meeting in Florence next month, where the issue of using the HVP Nodes to expand the network of collaborators will be raised. Nodes are becoming increasingly active as data sharing raises its profile; the idea of the Node being comprised of a national network of interested people who can help promote data sharing by providing clear instructions of where good data should go. It is important to point out that the country nodes do not get any funding from HVP. The effort they make are within their national framework, many of them are already funded through their national health programmes. Those countries that have a mixture of private and public diagnostic labs tend to find it more challenging. However, it is usually not money they need; rather it is getting people to collaborate and providing clear steps to follow. Currently, ICCAC is encouraging nodes to have some form a steering committee or advisory group to increase engagement. It should also be noted that both BRCA and GG2020 are fulfilling their purpose as being entry points to international data sharing and providing concrete examples for countries to be involved in.
- ii. Melissa Landrum reports that Johan has reached out to ClinVar to start the process of getting the LOVD3 database into ClinVar. Melissa will report back with progress. ClinVar variants are single entry compared to LOVD; we then have two options to offer the world which will work differently in different settings, depending on individual local needs and aspirations.

4. Plans for ASHG – HVP meetings in Orlando

HVP routinely hosts several meetings in conjunction with ASHG:

- i. with journal editors, chaired by Garry
- ii. a face to face meeting of HVP members; while ASHG is very busy, a lot of members are there; it is hoped that this could be alongside Global Alliance which is immediately preceding ASHG.

Amy will liaise with Peter Goodhand's office to find a meeting time and venue that would be suitable for both of these meetings in Orlando in October. HGVS plans should also be considered as part of this. This will be discussed again.

5. Reports from Project-wide Initiatives

a. BRCA

John Burn reports that BRCA are heading towards having 20,000 variants. The recent HVP 14th International Symposium on Variants in the Genome meeting in Santiago was very successful with the BRCA session being very well received. Genomics England have given confirmation their data will be incorporated into our BRCA exchange – they have currently completed over 23,000 whole genome sequences. John reports that he has been invited by GA4HG to put together an outline bid for further funding for the BRCA Exchange based on its successful progress. John and Stephen Channock in charge of driving that forward. This bid should help to secure our curation, our administrative structure and help fund the app development

b. GG2020

- i. John reports that he will be giving a keynote speech at the Global Globin 2020 conference, taking place in Kuala Lumpur in July.
- ii. Helen Robinson and Raj Ramesar report that the Global Globin expert panel application has been accepted by Heidi Rehm and her colleagues which means we have representation of a global panel that includes representatives from Thailand, USA, South Africa, Mexico, Netherlands, Greece, France, Congo, Malaysia, Italy and Portugal and the Thallaemia International Federation (TIF). ClinGen steering committee has given us the go ahead for Stages 1 and 2 of a 3 stage process. This will allow the panel develop its procedures and contributions in a systematic manner
- iii. The relationship between HVP (through GG2020) and CING (through Ithamet and the Cyprus team) needs to be mapped out more clearly and formalized. This needs to be discussed further.

6. Current Issues – invitation to those on the call to report on issues or events that are topical

- i. Marc Greenblatt raised the issue of overlap between the following groups overlapping each other: HGVS; HVP; GA4GH; ClinGen; Decipher among others. John reassures that this is being addressed in several ways including by integrating meetings. However should everyone agree it a meeting could be convened at ASHG; it was also noted that some of the key people will be at InSiGHT in early July so informal discussions could take place there if required.
- ii. Raymond Dagleish raised the issue of proposed changes to how transcripts are recorded in RefSeek gene records. He had received an email from Terrance Murphy, the Refseek and gene team lead at NCBI, informing him that there is a proposal to change how exons are numbered in genes, such that the numbering will be independent of the gene itself but will be numbered on the basis of the exons in the individual transcript meaning that a variant may lie in exon5 in 1 transcript and exon 7 in the another transcript. If this is correct, it will have important implications. When referring the exon, people will thus have to refer to the transcript and version number. NCBI are claiming this is being driven by the diagnostic labs but does not appear to have been any discussion of these new proposals. It is concerning that this appears to come out of the 'blue'. Members confirmed that they had not been involved in the development of these proposals. It was agreed that all would try to find out more so that this can be discussed again at the next ISAC meeting when more information has been obtained.

7. Future meetings

- i. 20 July
- ii. 28 September
- iii. 19 October (in conjunction with ASHG – Orlando TBC)
- iv. 16 November
- v. 14 December

