

MINUTES

International Scientific Advisory Committee
Teleconference
Thursday 27 April 2017
1200hrs UTC/GMT



MEMBERS

Present

Voting Members

Garry Cutting
Raymond Dalgleish
Jordan Lerner-Ellis
Finlay Macrae
Juergen Reichardt
Mauno Vihinen
Mike Watson

Non-Voting Members

Qasim Ayub
Arleen Auerbach
Aida Falcon de Vargas
Melissa Landrum
Augusto Rojas Martinez
Ines Noher de Halac
Mona El Ruby
Tom Weber

ICO Staff

Amy McAllister
Helen Robinson

Apologies

Voting Members

John Burn
Ming Qi
Raj Ramesar
Peter Taschner
Martina Witsch-Baumgartner

Non-Voting Members

Mireille Claustres
Collet Dandara
Johan den Dunnen
Marc Greenblatt
Ada Hamosh
Helena Kaariainen
Dhavendra Kumar
Heidi Rehm
Katsushi Tokunaga
Ingrid Winship

AGENDA

1. Welcome & Apologies (noted as above)

Amy McAllister was welcomed to the meeting. Amy is a member of John Burn's team at the Institute of Genetic Medicine at Newcastle University. She is based at the International Centre for Life, Newcastle upon Tyne, UK. She will be assisting with the administration and management of GVL and its Councils, organizing meetings and communications so on. Members will have already seen her name on various messages and announcements; those attending ESHG will have an opportunity to meet her in person in Copenhagen.

2. Confirmation of the record of the previous meeting – there were no changes to the draft record circulated to members on 31 March, other than the corrections to those who were mistakenly omitted from the list of attendees.

3. Members' Up-date – several items raised at the previous meeting were up-dated the Chair:

- Funding for GVL was looking sound, with several of the discussions reported previously coming to fruition;
- The new administrative arrangements for the UK office were still being finalised by the Board and more information should be able to be provided after further meetings scheduled for the next few weeks;
- The relationship with UNESCO was now on a firm footing and GVL is continuing to fund several projects that are on-going; it was also mentioned that there would be no biennial meeting at UNESCO Headquarters in 2018 as HVP's next meeting will be held in conjunction with HUGO's meeting in Yokohama, Japan in March 2018 (further information on this is provided below – please see item 8);
- Further discussions are planned over the coming months with partners to further progress collaboration and improve co-ordination across the various societies, organizations and alliances working in the human genomics area; these will be reported on as more information becomes available; those on the call indicated their support

for these discussions as it would assist in a clearer focus, avoid duplication of effort and help strengthen the achievement of HVP's goals.

4. Operational Issues - Reports from Working groups

- a. **WG 07 – Joint Working Group on minimum content requirements for genomic variant databases** While neither Martina nor Peter were able to be on the call, they had sent a message indicating that there will be a meeting of WG 07 at ESHG; those involved in this group should contact either of them for information regarding the exact time and venue.
- b. **WG 09 – Database Quality Accreditation Scheme Development** – Mauno Vihinen reported that the testing on his own gene specific database (BTK gene) had been completed and this had led to some small refinements of the process; but it was very successful given that this was the first test of the scheme. The 'pilot' testing will be to be expanded to include a small number of databases before it can be more widely used. The next major task for the working group will be to develop a suitable scoring mechanism; this will need to include how to score each separate element of the scheme, plus the generation of an overall score. Defining these, then making a reliable and valid assessment will be addressed. This was not considered to be complex as there are many schemes that already operate in closely related fields that can be reviewed, but the more complex task will be to agree on how to do this, taking into account the time it takes to do it comprehensively and transparently. Those seeking more information should contact Mauno directly.
- c. **WG 10 – Proper Reporting of sequence variants in journal articles** - Garry Cutting asked for this to be taken up at a future meeting as further discussions have not yet taken place.

5. Reports from Project-wide Initiatives

GG2020 – as previously advised, there will be a GG2020 Challenge meeting in Copenhagen; unfortunately representatives from WHO will be unable to attend as the meeting coincides with the World Health Assembly in Geneva, the annual meeting of health ministers and there will be an election of a new Director-General for the WHO; UNESCO is supporting the meeting. GG2020 will be featured at the 10th African Society of Human Genetics and the National Society of Human Genetics in Cairo 9-11 November 2017. The theme of the conference is "**Human genetics and genomics in Africa: Challenges for both rare and common genetic disorders**"

6. Current Issues

The following issues were raised:

- Qaism Ayub reported that he had recently returned from Pakistan where he had further productive discussions with a group of researchers and clinicians centred around the University of Lahore who are ready to take steps to establish an HVP Country Node. Both Arleen Auerbach and Raymond Dagleish indicated that they had each independently received documents from researchers from University of Lahore for review, and were in discussion with the authors. It appeared that this provided an opportunity to work with the group to better coordinate and align their bio-molecular work, particularly the sequencing work, with international standards should they wish this. Qasim will follow up on this to seek further information and clarifications, then the process for establishing the Country Node can be initiated.
- This discussion of Country Nodes led to an exchange about Country Nodes in general as a result of which it was decided to discuss this in more depth at a subsequent meeting where Martina Witsch-Baumgartner as current Chair of ICCAC, might be asked to report on the activities of Country Nodes. Helen Robinson drew members' attention to the objectives in the HVP Roadmap 2016 -2020 that identified the need for better communication between ISAC, the ICCAC and GSDSC to achieve the overall goals set for HVP by 2020. The excerpt from Objective 1 is in the Box below.

OBJECTIVE 1: *to facilitate growth in the amount of quality curated genomic variant data that is publicly available in all regions of the world, through developing and consolidating effective partnerships that better co-ordinate data collection and sharing efforts across the world*

OUTCOMES:

1. *More countries and disease-focussed groups included in data collections, storage and sharing activities*
2. *A deepened commitment to better quality data collection, storage and sharing through participation in the generation of relevant standard and guidelines, their implementation and the provision of assistance to other to do so*

3. <i>An increased ability of project structures to sustainably resource their ongoing operations and implement the next coordination iteration of HVP</i>
ACTIONS
1. <i>Advocate for greater application of HVP standards, guidelines and recommendations</i>
2. <i>Help Country Nodes to be more proactive in identifying key individuals and organizational partners in government, health and research areas in their own country and in developing good sustainable relationships with them</i>
3. <i>Assist CN and G/DSDBAC database to work more closely and to link people in countries with appropriate international level databases</i>
4. <i>Make better use of data has already been generated</i>
5. <i>Define minimal content and quality if data sharing infrastructure and implementing this through all HVP Data Provider members</i>
6. <i>Define process for gaining full status as an HVP Data Provider member</i>
7. <i>Develop sustainability strategies that balance the requirement for ongoing funding with the HVP core principle of open data sharing</i>
8. <i>Promote the establishment of trustworthy public resources which in parallel to the databases used by and built for experts, explain the consequence of specific variants in lay terms</i>

Members are asked to consider these actions and this will be taken up in more detail, probably at the June ISAC meeting. In the meantime any questions, suggestions or comments can be sent to Helen Robinson directly – hmro@unimelb.edu.au

7. Other business

No other business to discuss at this time

8. Future meetings

- a. HVP/GVL's next face-to-face meeting will held in conjunction with **HUGO's meeting 13 – 15 March 2018, Yokohama Japan** while the exact details are yet to be finalised, it is expected that HVP will organize technical sessions within the main scientific program as well as having more operational Council meetings along-side the scientific program. Members are asked to put these dates into their schedules and wait for more specific information; any suggestions concerning how this should be organized can be sent to Helen Robinson hmro@unimelb.edu.au or to the Co-Chairs
- b. There are a range of other meetings of interest not only to ISAC members, but to all consortium members, consequently, a list of the most relevant 2017 -2018 meetings will be added to the HVP web-site and kept up-to-date by the ICO. The ICO will be happy to continue to circulate information on behalf of members, and to highlight events the members are involved in organising, or where they are making presentation and so on. This is expected to assist in networking and information sharing. Please contact – amy@variome.org
- c. Forthcoming meetings include:
 - i. ESHG - 27 – 30 May 2017 Copenhagen, includes HGVS and GG2020 meetings
 - ii. XIV Congress on Variants – 5 – 8 June 2017 Santiago, Spain
 - iii. 7th Biennial Meeting of the International Society for Gastrointestinal Hereditary Tumours (InSIGHT)– meeting –5 - 8 July 2017 Florence, Italy
 - iv. ASHG –17 – 21 October 2017 Orlando, USA
 - v. Asia-Pacific Congress on Human Genetics – 8 – 10 November 2017, Bangkok, Thailand
 - vi. GG2020 international meeting –16 – 19 July Kuala Lumpur Malaysia
 - vii. Asia-Pacific CHG – 8 – 10 November 2017, Bangkok, Thailand – theme; Genomic Medicine and Clinical Practice
 - viii. 10th African Society of Human Genetics and the National Society of Human Genetics in Cairo, Egypt 9-11 November 2017. The theme of the conference is "**Human genetics and genomics in Africa: Challenges for both rare and common genetic disorders**"
 - ix. World Science Forum, UNESCO, 7 – 11 November Amman, Jordan

9. Future ISAC Meeting Dates

- 25 May - Teleconference
- 22 June - Teleconference
- 27 July - Teleconference
- 28 September - Teleconference
- 19 October – Potential face-to-face during ASHG

- 16 November - Teleconference
- 14 December - Teleconference