

MEETING RECORD

International Confederation of Countries Advisory Council
Teleconference

Tuesday 13 June , 2017

Call 1: 0800 UTC/GMT Call 2: 2200hrs UTC/GMT

Check the time in your time zone at <http://short.variome.org/iccac-call-1>

MEMBERS

Apologies

Martina Witsch-Baumgartner	Austria
Peter E.M. Taschner	Netherlands
Marielle E. van Gijn	Netherlands
John Burn	UK
Amy McAllister	

On the Call

Angela Solano	Argentina
<i>Vincent Harley</i>	<i>Australia</i>
Iscia Lopes-Cendes	Brazil
Matthew Lebo	Canada
Andreas Hadjisavvas	Cyprus
Domenico Coviello	Italy
<i>Fahd Al-Mulla</i>	<i>Kuwait</i>
<i>Zilfalil bin Alwi</i>	<i>Malaysia</i>
Augusto Rojas-Martinez	Mexico
Luis Figuera-Villanueva	Mexico
Luz Berenice Lopez Hernandez	Mexico
Denise Brito	Mozambique
<i>Tilak Shrestha</i>	<i>Nepal</i>
<i>Oyekanmi Nash</i>	<i>Nigeria</i>
<i>Hadiza Mahey Rasheed-Jada</i>	<i>Nigeria</i>
Catherine Lynn Silao	Philippines
<i>Aida Falcón de Vargas</i>	<i>Venezuela</i>

ICO Staff

Helen Robinson

* those in ITALICS had difficulties with connections or times

Not on call

Johan Rung	Sweden
Malin Melin	Sweden
Michael Watson	US
Chí Dũng Vũ	Vietnam
Kathleen Claes	Belgium
Milan Macek	Czech Republic
Propser Lukusa	D.R. Congo
Sherifa Ahmed Hamed	Egypt
Mahmoud Raafat Kandil	Egypt
Ming Qi	China
Raj Ramesar	South Africa
Maria Jesus Sobrido	Spain
Sharif Akhteruzzaman	Bangladesh
Dylan Mordaunt	New Zealand
Rosemary Ekong	Nigeria
Celeste Bento	Portugal
Xavier Estivill	Qatar

AGENDA

1. Welcome, Apologies, and Administrative Matters

As Martina was unable to join the call and that Falil, our Deputy Chair, was to be on the call but unable to be heard, Helen R acted as call convener. Helen said that for the rest of the year there will be only one call for this Council, instead of two, mainly because the second call was not well attended and so it was not a good use of resources; however, it will be monitored and any feedback or suggestions would be most welcome.

Helen alerted members to the next face-to-face meeting of the ICCAC will be at the HVP/HUGO meeting in Yokohama, Japan in March 2018. See Agenda Item 7 for more information on this.

She also indicated that two more countries – Pakistan and Qatar – are likely to join the group as new HVP Country Nodes; hopefully the process will be completed before the next call and they can be welcomed then.

2. Confirmation of minutes of previous meeting

There were no changes to the meeting record.

3. Operational Issues

a. Letters National Human Genetics Societies– progress report

- Brazil – Ischia indicated that there will be a meeting of the national society of medical genetics later in the year and as there are planned lectures and courses covering related topics, it should be not be difficult to obtain a message of support
- Italy – Domenico said that their national society was meeting later in the month and it should be possible to obtain a formal and positive response there
- Netherlands – not on this call but it has been advised at ESHG that a letter will be forthcoming from them
- Canada – positive also but Matt will follow up; he also advised of the statement from the American College of Medical Genetics and Genomics as many countries follow the ACMGG their statement might influence other organizations
- Argentina – Angela indicated that this is in train
- Mexico – Augusto indicated that this will be taken up at meetings planned for October and that it is not expected to be a problem.

Members were reminded that if a letter was too difficult to obtain, an email or some other record or expression of support would be good. **Individual countries receiving replies to Letters National Human Genetics Societies should forward them to the ICO please.**

b. Joint Working Group: Minimum Content Requirements

Many members are anxiously waiting for the results of this group and their discussion paper. In Martina and Peter's absence, Helen reported that there had been a meeting of the group at ESHG and that it was hoped that the paper would be available in draft format before the northern summer. There is still work to be done on simplifying the document and integrating material that is currently in appendices. The draft will include examples drawn from both LOVD2 and LOVD3. ICO will circulate the paper as soon as it becomes available.

4. BRCA Challenge Report –

The question was asked: who has forwarded their data recently? Ischia indicated that in Brazil they will be taking a federated approach to both BRCA and GG2020 national data, using their national database, and linking this, where appropriate, to international databases. They are using LOVD3 and are involved in the Beacon project. Their database is public and a link to it is here – www.bipmed.org. Ischia very generously agreed to respond to any questions or queries about what is happening there – please contact her directly: icendes@unicamp.br) or lopescendes@gmail.com

Matt reported on Canada and said that there was some Canadian data in BRCA Exchange as well and they were also involved in Beacon project, as well as ClinVar.

Andreas asked that he delay reporting on Cyprus as he was only now rejoining the Council and was more interested in hearing what others had to say.

Helen also reminded members of the message from Johan den Dunnan sent to members on 17 May and repeated here below -

"...informing of the new country pages developed in LOVD3 related to the topic of "country nodes", please note a new general service that LOVD offers. Using any country's two-letter code (e.g. nl, de, jp, us, ...) you can now see which variants in the LOVD3/shared are linked to either an Individual (patient) or a Submitter. He highlight what he has done for Angela in Argentina also. Members are encouraged to investigate their pages as you will see not only data you have submitted but anything submitted that can be linked to a particular country. This highlights the role of the Country Node to know what is happening across their country – what everyone is doing, and how to co-ordinate and involve others.

<https://protect-au.mimecast.com/s/z4n0BMSQQKNOiD?domain=de.lovd.org> (5019 vars / 9425
 subms)
<https://protect-au.mimecast.com/s/rNKkB3t008Qoia?domain=be.lovd.org> (4405 / 4249)
<https://protect-au.mimecast.com/s/ZXg6BmSKKLD9Hn?domain=jp.lovd.org> (2470 / 751)
<https://protect-au.mimecast.com/s/m4klBrSNNrzVcR?domain=au.lovd.org> (1095 / 7790)
<https://protect-au.mimecast.com/s/krZOBli11Np2tM?domain=at.lovd.org> (321 / 1511)
<https://protect-au.mimecast.com/s/pLGZBoi44lGmiO?domain=my.lovd.org> (103 / 41)
 ...

We are working on a page showing numbers per country.

These pages might be interesting to use as a starting point for a country node (with doing nothing there might already be something). For some countries, e.g. Argentina (<https://protect-au.mimecast.com/s/Ld1ZBmS33JW5SN?domain=argentina.lovd.org>) we are currently building this a step further where the start page is showing the information in another language (start page to be controlled by the country node).

Best regards,

Johan den Dunnen"
 LOVD-team

<https://protect-au.mimecast.com/s/GN1VBvtnnJALIZ?domain=facebook.com>

Any other countries not on the call but who would like to report progress, please do so by sharing a message with the group or by sending to Helen – hmro@unimelb.edu.au

5. Global Globin 2020 Report

In Falil's absence, Helen reported on :

- GG2020 is now formally linked to ClinGen/Clinvar as a global Expert Panel for haemoglobinopathies;
- There was a meeting of GG2020 working group at ESHG and a report will be available soon; next meeting will be in Malaysia on 16 July – closed meeting for GG2020 participants and 17 – 18 July for open registration – see gg2020conference@gmail.com for more information or <http://hvpmalaysia.kk.usm.my/gg2020>
- all those Council members who are involved in GG2020 and in thalassaemias in particular should contact their national Thalassaemia society regarding participation at the TIF meeting to be held in Greece in 17 – 19 November; members should consider presenting their work at this important meeting.

6. Gaining Financial Support for your Node

Paper with suggestions for funding was discussed – any further ideas can be added to the list – what has worked for you?

Links with UNESCO and WHO/PAHO were raised. It seems that these organizations are more important in different parts of the world. Role of WHO/PAHO in data sharing and with variant databases was raised in connection to the letter from Stylianos Antonarakis, Past-president of HUGO, to the Editors of *Science* on 26 May 2017 suggesting that WHO oversee them in some way. This is a topic meriting thought and debate. Helen said that she had been in touch with WHO over the letter and would receive their thoughts and reactions.

7. Process for becoming a Country Node

There are two items here:

- *Process for becoming a HVP Country Node* – these forms and papers need to be up-dated for the change of address to the UK so they were circulated to see if any other changes were needed. Ischia, who had completed the process most recently, said that she had found it very clear and straightforward. As there are some new nodes about to commence the process, any comments for suggestions to hmro@unimelb.edu.au by **end of June** please, or we assume that you support the process as is.
- **Country Node Profile – 2017** – Helen noted that the draft was circulated for comment but those on the call said that it was straightforward and clear. She also informed members that these documents, when completed, provided the basis for the biennial reporting of Node activities that will be scheduled at the face-to-face meeting of ICCAC at Yokohama in March 2018. It will also be used to up-date the HVP website; comments and questions to Helen by **end of July** please. Forms will be completed by end of 2017 and based on the 2017 situation; those, like Brazil, who had so recently completed them would not need to do so again

8. Reports/Questions/Issues from Individual Countries

Most of the country news had been taken up in previous items. Augusto gave more details about the meetings planned in Mexico. All are welcome to attend. John Burn and Johan den Dunnen will be speaking about HVP and LOVD. Luz also talked about the activities to involve students and early career professionals – this fits with HVPs' Objective 5 for the Roadmap 2016-2020 – to actively promote ways to involve the next generation in genomic data sharing activities. This is something that all members are encouraged to work on.

9. Feedback from recent meetings of interest:

i. ESHG – Copenhagen

Helen raised the topic of the panel in which she participated at ESHG – that of how to ensure that there is more sequencing and bio-molecular data from sub-populations and minority communities. This is a big topic for almost all countries, especially those with diverse and ethnically mixed populations, like Brazil, Canada, USA and Australia to name but a few, but really all countries face this issue of needing diverse references. This is an issue that has come to prominence in GG2020 where not only is it important to know the range of variants, but also the frequency of occurrence of markers and so on. It was agreed by those on the call that this topic needs further thought and discussion.

ii. Variant Detection – Spain

Angela shared her views on the Spain meeting, indicating that she found it very interesting and helpful. It was a very large group but the focus was on the practical – “what to do, and how to do it”; the sessions on sequencing methodologies and bio-informatics were helpful for keeping up-to-date

10. Future Meeting Dates

- **15 August – PLEASE NOTE that this meeting will be cancelled. There will be only one call for the rest of 2017.**
- 10 October – Teleconference
- 12 December - Teleconference