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# MINUTES

International Confederation of Countries Advisory Council  
4 August 2015 1200hrs GMT/UTC  
Check your time zone at <http://short.variome.org/iccac-meeting-time>

## MEMBERS

### **Present**

#### ***Interim Members:***

Martina Witsch-Baumgartner (Chair)	Austria
Andreas Hadjisavvas	Cyprus
Domenico Coviello	Italy
Zilfalil bin Alwi	Malaysia
Marielle E van Gijn	Netherlands
Mike Watson	USA

#### ***Observers:***

Denise Brito	Mozambique
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#### ***ICO:***

Helen Robinson (HVPI Liaison – WHO)  
Timothy Smith (Communications Officer)

### **Apologies**

#### ***Interim Members***

Graham Taylor	Australia
Thomy de Ravel	Belgium
Kathleen Claes	Belgium
Jordan Lerner-Ellis	Canada
Matthew Lebo	Canada
Ming Qi	China
Maria Loizidou	Cyprus
Milan Macek	Czech Republic
Sherifa Ahmed Hamed	Egypt
Mahmoud Raafat Kandil	Egypt
Maurizio Genuardi	Italy

Makia Marafie	Kuwait
Amir Feisal Merican	Malaysia
Augusto Rojas-Martinez	Mexico
Luis Figuera-Villanueva	Mexico
Tilak Shrestha	Nepal
Rebati Man Shrestha	Nepal
Peter Taschner	Netherlands
Oyekanmi Nash	Nigeria
Hadiza Mahey Rasheed-Jada	Nigeria
Raj Ramesar	South Africa
Collet Dandara	South Africa
Myungchull Rhee	South Korea
Jong Bhak	South Korea
Angel Carracedo	Spain
Maria Jesus Sobrido	Spain
Richard Rosenquist	Sweden
Johan Rung	Sweden
Andrew Devereau	UK
Sir John Burn	UK
Bruce Korf	USA
Aída Falcón de Vargas	Venezuela
Chí Dǔng Vǔ	Vietnam

#### ***Observers:***

Peter Pearson	Brazil
Celeste Bento	Portugal

#### ***Invited Attendees:***

Rosemary Ekong	Nigeria
Vera Lopes	Brazil

## 1. Welcome

## 2. Apologies

Apologies were noted as above.

## 3. Confirmation of minutes of previous meeting

Minutes were confirmed unanimously.

#### **4. Matters arising from the previous meeting**

All matters taken up in the items below.

#### **5. Global Globin 2020 – Zilfalil bin Alwi & Raj Ramesar**

Zilfalil bin Alwi reported on the GG2020 meeting held in Paris in June. Sixteen countries were represented at the meeting, bringing the total number of interested countries to twenty. A Steering Committee has been formed, members of which are Zilfalil bin Alwi (Malaysia), Raj Ramesar (South Africa), Carsten Lederer (Cyprus), Ming Qi (China) and Helen Robinson (Australia). Garry Cutting and Michael Watson (USA) have been providing valuable advice. The Steering Committee will be expanded moving forward. Zilfalil detailed future meetings dates for the GG2020 Challenge, including Hanoi in September as part of the Asia Pacific Congress of Human Genetics, Budapest in November for the World Science Forum and Paris in May 2016 immediately prior to HVP6.

Helen Robinson reported that the Challenge is in the process of identifying which international database they would be contributing data to. They know of two currently available data and are assessing options. A budget is also being drawn up that will allow participants to meet to set plans, but also to support some infrastructure development in low- and middle-income countries.

Domenico Coviello asked for a list of information about national activity that country coordinators could collect. Helen Robinson reported that the project would be writing to all those who had expressed interest with a progress report and some suggestions in the coming weeks. Helen Robinson also promised to send the Country Checklist.

#### **6. BRCA Challenge – John Burn**

Timothy Smith reported, on behalf of John Burn, recent progress of the BRCA Challenge. The current focus of the BRCA Challenge is to prepare a website with all known public, pathogenic variants in BRCA1 & BRCA2 for release at ASHG in October. This website will bring together data in different database including BIC, LOVD, ClinVar and others. The next step will be to begin classifying and publishing on this website variants of unknown significance using the ENIGMA classification processes.

Timothy Smith encouraged those Council members that were interested in participating in the BRCA Challenge to contact the HVP ICO.

#### **7. Project Roadmap 2016-2020**

The Council discussed the list of thought-starters for issues to be considered during the development of the next Project Roadmap. Council members raised additional issues, including:

- Developing easier means of data submission for diagnostic laboratories;
- Increasing the participation in the Human Variome Project of young professionals and the role of HVP Country Nodes in promoting this; and
- The development of guidelines for generating quality data, including variant naming, selecting transcripts and classifying pathogenicity.

Helen Robinson notified the Council that a draft of the Roadmap would be ready in time for the December meeting of the Council, and that sufficient time would need to be set aside for discussion at this time.

#### **8. Recommendations to the Scientific Advisory Committee**

No recommendations were made.

#### **9. HVP Country Node Progress Updates**

##### ***a. Italy - Domenico Coviello***

Domenico Coviello reported that Italy had made progress setting up an initial network of laboratories. Domenico asked about the data model for HVP Country Nodes and what would be considered the best Node to collaborate with in developing a data model for Italy. Helen Robinson responded that it would depend; that the Human Variome Project is using Project-wide initiatives like the GG2020 and the BRCA Challenge to answer these

questions for each disease and that there may not necessarily be a single data model for every disease. Historically, the Project used to advocate for a single Node in each country that would collect all the data and transfer it to the relevant international gene./disease specific database. However, the Project now recognizes the need to be more flexible in its approach.

**b. Malaysia – Zilfalil bin Alwi**

Zilfalil bin Alwi reported that the Malaysian Node of the Human Variome Project was in the process of producing a new newsletter. Previous editions are available at <http://hvpmalaysia.kk.usm.my/>. He invited members of the Council to contribute content to this publication.

**10. Other matters**

No other matters were discussed.

**11. Next Meetings**

- 29 September (1 week earlier than usual because of ASHG)
- 1 December