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# MINUTES

International Confederation of Countries Advisory Council  
9:30AM – 12:30PM, 7<sup>th</sup> June, 2013 Paris  
Room 16 Miollis Building, 1 Rue Miollis, 75732 Paris, France

## ATTENDEES

Martina Witsch-Baumgartner (Chair)	Austria	<b>Apologies</b>	
Thomy de Ravel	Belgium	Graham Taylor, John Coghlan	Australia
Ming Qi	China	Helene Antoine-Poirel	Belgium
Andreas Hadjussavvas	Cyprus	Milan Macek	Czech Republic
Zilfalil bin Alwi	Malaysia	Sherifa Ahmed Hamed	Egypt
Andrew Devereau (alternate Chair)	UK	Mahmoud Raafat Kandil	Egypt
Aída Falcón de Vargas	Venezuela	Makia Marafie, Fahd Al Mulla	Kuwait
		Amir Feisal Merican	Malaysia
<b>ICO:</b>		Augusto Rojas-Martinez	Mexico
Richard Cotton (HVP Scientific Director)		Luis Figuera-Villanueva	Mexico
Helen Robinson (HVPI Liaison – WHO)		Tilak Shrestha, Rebati Man Shrestha	Nepal
Heather Howard (HVPI Operations Manager)		Angel Carracedo, Maria Jesus Sobrido	Spain
Timothy Smith (HVPI Communications Officer)		Chí Dũng Vũ	Vietnam
Fan Li (Liaison Officer)		Sir John Burn	UK
Julia Hasler (HVPI Liaison – UNESCO)		Mike Watson, Bruce Korf	USA
Rania Horaitis (HVPI Conference & Events Officer)			
<b>Also present:</b>			
Finlay Macrae	Australia		
Xitao Li	China		
Jun Chen	China		
Mona El Ruby	Egypt		
Mireille Claustres	France		
Peter Taschner	The Netherlands		
Arleen Auerbach	USA		

## AGENDA:

Meeting Opened 9:40am Paris time

- 1. Welcome**
- 2. Apologies**
- 3. Welcome new Members**

No new members since last meeting.

#### 4. Confirmation of minutes of previous meeting – 16th April, 2013

The minutes were approved without modification.

#### 5. Issues arising from previous meeting

Note that all issues arising from the previous meeting have been taken up in the items below.

#### 6. Country Node activities

##### 6.1 Country node reports: each Country Node representative to give a report on recent activities involving their node

**Cyprus:** Baseline report was completed. It is planned to input data into LOVD by the end of this year. A master's student was recruited to set up the database.

**Venezuela:** Following several meetings the Venezuelan HVP Node has been formally established. Diagnostic laboratories are interested in the project. There are some concerns about giving and sharing information, but these can be dealt with particularly by using the examples from other successful countries. The node also met with members in the Latin American Society (RELAGH) to discuss common issues of data collection. Spain and other countries have also been approached to identify further opportunities for collaboration.

**Egypt:** There is work to do in promoting the need to collect and share data and the issues are not well understood at the national level. The Node is moving forward to collect data from genetics departments and also to obtain WHO support. The recent naming of a key institute as a Centre of Excellence for genetics and genomics will assist.

**Belgium:** The Country Node has been involved in developing a national plan with the Ministry of Health. The role of clinical genetics services is recognized in this plan. The Belgian Node is involved in developing the capacity of genetics and genomics in several central African countries including the DR of Congo. At a recent meeting in Kinshasa the local society was re-launched with the active support of the Belgian Node and DR Congo is expected to request status as an HVP Country Node in the near future.

**China:** A national network for retinal diseases has been established, with BGI and three medical schools involved. The eye disease registry in Zhejiang is used as prototype to build up a national system. A hearing loss database has also been established based on the eye model. China is reorganizing its infrastructure following the restructuring of the Ministry of Health. This is a lengthy process and has impact on a lot of activities. The project to run a genetic counsellor training program with the MOH has begun but will be delayed by approximately six months until the restructuring of the MOH has been completed. Academic projects are conducted as usual.

**Malaysia:** A workshop was organised in December 2012 that generated a lot of interest from the government, including the MOH, and the broader genetics community. This has helped to promote the activities of the Node. Since then, three further meetings have been held. The Australian node is working with the Malaysian Node to build capacity and share knowledge. The node continues to collect data from published information and through support from the host university, a full time student has been engaged to collect and store data. It is intended to establish a South-East regional network to assist the many countries in the region who are at various stages of establishing national Nodes and to help encounter obstacles that may arise. A meeting has been organised to meet with the UNESCO Malaysia delegates to get support at ministerial level.

**Australia:** Graham Taylor now works in the University of Melbourne as the Herman Professor of Genomic Medicine and Director of the HVP Australian Node. The Node gained funding support from the Australian federal government to develop web-based tools for data collection from labs. A pilot study has been conducted involving three laboratories as a first stage. It will expand to 15 laboratories by the end of this year with coverage of approximately 75% of diagnostic services. This is in conjunction with a national program (BioGrid Australia) to develop a national system of linked clinical data, including births, deaths, and other clinical records, using de-identified data. The legal

and ethical issues related to sharing this internationally are under consideration. Some very useful automatic configuration tools have also been developed as part of this effort and are available for other Nodes to use.

**UK:** The UK government has established and funded a major project to run and collect information from sequencing 100,000 genomes. This will help the government to plan and support the infrastructure necessary to support health service delivery across the whole of the UK. The past ten years of work by HVP colleagues is being integrated into this project and will form the core for future activities. As this project will have a big impact on how the UK Node will operate, decisions about the establishment of Nodes in Great Britain will need to move in parallel with this project. In the mean time, works continue on data collection and connection, development of tools for displaying data from LOVD, quality control and phenotype data collection.

**Austria:** The Country Development Program grant with Czech Republic and India has been approved, pending the funds being made available. This will network activities across the three countries and support exchange of knowledge and capacity building. It is possible to get some funds from the Austrian government for database development. Seven new LOVD were established.

**France,** while not yet a formally established node, also reported on activities, indicating that the French government has set aside funds to organize a series of national databases. This is still in the planning phase, but it will be important to follow progress and it may encourage other countries to do the same.

**Spain (written report):** A national general variant database is being constructed, with participation of the main public genetic diagnostic centers. A conference on the HVP was presented by John Burn and Maria Jesus Sobrido at this year's meeting of the Spanish Human Genetics Association. After the conference, a couple of geneticists came to offer their help for country node activities.

## **6.2 Report on new nodes likely to be established in next 6 to 12 months**

This was taken up under the following item 6.3.

### **6.3 HVP Country Node Baseline Report**

ICCAC members were thanked for their responses to the request for information on their current activities as outlined in the HVP Country Node Baseline Report. The original intention was that these reports would be collated and the contents analysed by the ICO to produce a single report to provide an overview of data collection and storage, and diagnostic testing around the globe with 2012 being the baseline for the report. However the information received was in a variety of formats and came from many more countries than there are HVP Nodes. Given the complexity of the information and the richness of the materials received, the ICO proposes that this project take a slightly different direction. It is proposed that the ICO –

- Create a simple electronic database with a file on each country undertaking some form of genetic testing irrespective of their current formal association with HVP, to contain any relevant information, reports and publications of nationally based testing and data collection and sharing
- Write to each country requesting any further information be sent for inclusion in the database
- Analyse the information from each country with a view to identifying priority issues for consideration by the ICCAC, for example: how to best categorise the various types of testing being carried out in various regions of the world; how to report on testing in a meaningful way; how to report on aspects of quality control of data sharing.

For this to happen, each Node would need to agree that the information that they originally provided in confidence, can be shared among HVP members in an electronic database.

Members welcomed the proposal and agreed to proceed in this manner. They also decided that ICCAC should develop a status report on Country Node activity to be published every two years.

This approach raised the issue of how to engage those countries active in human genetics and genomics and with diagnostic laboratories providing services in HVP activities and as new Country Nodes. Following discussion, it was agreed that all members of the ICCAC should assist whenever and wherever possible in promoting the responsible collection, storage and sharing of data on variation among their colleagues and professional networks. It was also agreed that it would be timely to develop a more useful system for categorising HVP Country Nodes. Members asked the ICO to develop a model for their consideration that includes different types of national membership of ICCAC for new members and suggested terms like observer, interim and full node be included in this proposal.

**ACTION:**

- **ICO to send message to each country regarding permission to share the information they have provided in an electronic repository available to members**
- **ICO to conduct preliminary analysis of this data to identify priority issues for discussion at future ICCAC meetings**
- **ICO to develop a proposal for discussion among ICCAC members defining three levels of Country Node status for new members: observer, interim, full**
- **ICO to work with ICCAC members to produce a report on Country Node activities every two years**

#### **6.4 Human Variome Project/China Country Development Program: up-date on progress**

Two proposals have been formally approved. There are two more proposals that could be reviewed by the International Scientific Advisory Committee. It was noted that the current guidelines and information package for applicants was scheduled for closure at the end of June 2013.

Prof Li and Ming Qi informed the Council the funding situation for HVPCCDP. Ming, representing Prof Li, promised that the funds would be transferred to ICO before the end of June.

ICO confirmed that HVPCCDP application would remain open until the funds are used up. A new information package for applicants will need to be prepared when the funding is available.

**ACTION: China Node to transfer funds to ICO**

#### **6.5 Guidelines for Country Node formation: discussion of attached draft paper**

The Council members were asked to review the draft paper and send ICO their feedback before the next meeting. ICO will then produce the final draft.

**ACTION: Council members to review the draft paper and send ICO their feedback before the next meeting**

### **7. Working towards 2016 HVP Roadmap**

#### **7.1 Roadmap summary and role of ICCAC: Discussion of attached action summary**

ICO hopes that the council starts to work out how ICCAC can assist with achieving the goals defined in the HVP roadmap 2016. This will be further discussed in the next meetings.

**ACTION: Council members to review the action summary for discussion in the next meeting**

#### **7.2 Standards Development Process/Activity Proposals**

No Activity Proposals received

#### **7.3 Working Groups under the purview of the Council**

None

A new item will be added to the standing agenda: 7.4 Interest Groups. It was agreed to explore the feasibility of creating an Interest group to work on resource mobilisation for establishing HVP Country Nodes.

**ACTION: Chair of ICCAC to canvass interest for such a group.**

## **8. HVP5 program 2014: Discussion of attached draft program**

Heather Howard explained the draft program developed by ICO. The program includes sessions of presentations based on open call for abstracts, posters, brief pre-conference meetings of the Councils and formal Council meetings. The Council members commented that the draft program was too long and could work better if over a weekend (possibly in two venues since UNESCO is not open over weekends) and/or having more concurrent sessions. More comments are welcome before the next meeting.

**ACTION: Council members to make comments on the draft program before the next meeting**

## **9. Items for information**

### **9.1 Up-date on World Health Organization consultation**

An up-date on the consultations on establishing an international program to deal with human genetics and genomics and public health together with WHO was provided. Since November 2012, six formal consultations had been held in many parts of the world and involving more than 200 clinicians, researchers, diagnosticians from more than 75 countries. The response was overwhelmingly positive, indicating that the time was right to establish such a program according to the genetics and genomics community itself. Three main issues are emerging as priorities for action:

- Health care delivery issues – as advancements in testing, particularly genome-wide testing had lowered the costs and increased the levels of testing in most parts of the world; this has the consequence that many national health systems are grappling with how to integrate these services into delivery; as many tests are carried out across national borders there is a need for more formal international collaboration and standard setting
- Ethical, social and legal issues associated with the provision of quality health services in diverse cultural, ethnic and religious settings
- Need to engage health systems planners and policy makers in understanding the implications that advances in human genetics and genomics have for public health

These results will be presented to WHO headquarters at a meeting later in the month with a view to gaining agreement for WHO to establish such a program in the near future.

### **9.2 UNESCO**

HVP is an official NGO partner of UNESCO. Julia Hasler explained how UNESCO could help member countries. Each country has a national commission and a permanent delegation to UNESCO, both important for making contact and developing relationships. UNESCO has strong interest in science development, international collaboration and networking. HVP is a global, multi-disciplinary project that fits well in UNESCO's agenda. Organisers for international meetings can apply for UNESCO patronage.

### **9.3 Population-specific variant display**

Peter Taschner, on behalf of Jula Muilu, Anne Polvi, Ivo Fokkema, etc. presented information on a new FinDis portal. This approach could be used to display population-specific variants from LOVD on a Country Node site without the need of setting up new databases.

**10. Recommendations to the Scientific Advisory Committee**

None

**11. Other matters**

None

**12. Next Meetings - all via Calliflower unless otherwise indicated**

- 20th August
- 15th October
- 17th December

Meeting closed 12:25PM Paris time