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## MINUTES

International Confederation of Countries Advisory Council  
19<sup>th</sup> February, 2013 - 11PM Melbourne, Australia time  
Via GoToWebinar

### ATTENDEES

Martina Witsch-Baumgartner	Austria	<b>Apologies</b>	
Andreas Hadjussavvas	Cyprus	Graham Taylor, John Coghlan	Australia
Sherifa Ahmed Hamed	Egypt	Helene Antoine-Poirel	Belgium
George Patrinos	Greece	Milan Macek	Czech Republic
Fahd Al Mulla (Chair)	Kuwait	Ming Qi	China
Makia Marafie	Kuwait	Maria Loizidou	Cyprus
Augusto Rojas-Martinez,	Mexico	Michael B Petersen	Greece
Tilak Shresta	Nepal	Zilfalil bin Alwi, Amir Feisal Merican	Malaysia
Maria Jesus Sobrido	Spain	Luis Figuera-Villanueva	Mexico
Andrew Devereau	UK	Rebati Man Shreshta	Nepal
		Angel Carracedo	Spain
		Chí Dŭng Vŭ	Vietnam
		Sir John Burn	UK
		Mike Watson, Bruce Korf	USA
Heather Howard (HVPI Operations Manager)		Richard Cotton (HVP Scientific Director)	
Timothy Smith (HVPI Communications Officer)		Helen Robinson (HVPI Liaison – WHO)	
Fan Li (Liaison Officer – China)			

### AGENDA:

Meeting Opened 11pm Melbourne, Australia time

**1. Welcome**

**2. Welcome new Members**

No new members since last meeting.

**3. Confirmation of minutes of previous meeting – UNESCO Headquarters**

The minutes were approved without modification.

**4. Issues arising from previous meeting**

None

## 5. Report from Chair

Fahd thanked everyone for electing him Chair of the Council. Since June, several meetings have been held with ICO staff. There are great challenges ahead:

- i) **Baseline Report:** There are some questions still to be answered, eg. What is a Node? What are the duties for people overseeing a node? What should be taken to start a Node? Some Nodes are doing better than others. For example, we started Kuwait Node in 2004 but has not moved forward much, whereas some other Nodes (e.g. Australia) are doing better. Therefore we need a mean to measure the baseline activities and capability of each country. A survey has been sent to all Nodes and some other countries to complete baseline report. We hope to analyse the data and generate a manuscript. More importantly the data will allow us to understand the baseline (who has got what) and then develop guidelines. People who contribute to the survey will be on the manuscript.
- ii) **WHO/UNESCO partnership:** Helen Robinson is driving an important initiative aiming to establishing a case for an international program on genomics and public health, possibly linked in some way to WHO. This program will have global connections and be able to attract funders. We hope WHO will be able to promote the program to governments in different countries.
- iii) **Funding mechanisms to expand Country Node:** Grant applications for the HVPCCDP are in progress.

## 6. Countries Baseline reports

Timothy Smith reported that the ICO had received survey responses from half of the countries in this Council. Reports were also received from a few of the other countries that have not yet formed HVP Nodes but were also invited to participate. A total of 16 – 20 reports received so far. Nodes are encouraged to complete the report ASAP. Timothy is willing to assist as much as he can if any help is needed. He will follow up the outstanding reports in the next several days. The ICO expects to complete a draft summary report for consultation in this Council within the next two months. We hope to present the results at ESHG in June.

This is the initial baseline report. The ICO hopes to follow up this on regular basis (eg. yearly) to see how everything develops. Therefore this is something countries can use and build on for the genetics service improvement and node development.

Timothy Smith will send the abstract submitted to ESHG to the Council members. People who contribute to the survey will be listed as authors on the final manuscript. Fahd Al-Mulla suggests, which is agreed by the Council, that summarised data should be sent to everyone for comments.

<b>ACTION:</b> Timothy Smith to send abstract to Council members Timothy Smith to send summarised data to Council members for comment
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### Updates from country nodes

**Australia:** Australian Node is up and running a small set of tests as pilots for breast cancer, colon cancer and Huntington's disease, and currently collects data from three labs. Have funding from the government until end of 2013 to support collection of data from a further 12 labs. There are a total of 41 labs doing approximately 300 diagnostic tests, mostly paid by patients. Australia has robust processes to train clinical geneticists. It is debated whether this should be integrate into the undergraduate and graduate medical training schemes.

**Austria:** A meeting of the working group was held to develop ideas about the pilot phase to create databases. A PhD student is working on the LSVD. Presentations were given in a number of meetings. Try to apply funds from government. The application to Country Development Program is to work together with Czech and India on databases development.

**Cyprus:** Currently investigate what people do in terms of depositing their findings/variants into databases. There is strong interest from everyone to share the data. Funding is the issue. Try to have postgraduate students to create and maintain databases for the next 1-2 years.

**Greece:** In the process to migrate Greek National Genetic Database to the upgraded version of the Ethnos Software, which will be completed within the next month or so. Generate interest from general public and professionals in genetics and pharmacogenomics with educational activities funded by other sources.

**Kuwait:** Focus on normal population first – exome genome sequencing in normal Arabs. Plan to deposit data into local databases and generate publications. Apply grant to employ at least two people to enter the data from Kuwait, UAE and Qatar. Funding is the major problem.

**Mexico:** Node was started in June 2013 and four meetings held. People are very enthusiastic to establish and work in the node. Most of the patient cares in Mexico are based on government institutions. There are twelve institutions (seven in Mexico City and five outside) participating in the project. Plan to focus on certain diseases first, such as Muscular Dystrophy, hemophilia, etc. One curator employed and another two volunteer curators available. Plan to hold a workshop this year.

**Nepal:** Difficult to move forwards without funding. Applying for funds from HVPCCDP now.

**Spain:** Move forward slowly due to the funding issue. Two Master students work on HVP related works (meetings on centralised databases). Annual meeting of Spanish Society of Human Genetics will be held in April. A workshop is organised to communicate about HVP.

**UK:** The strategic review of UK genetics has moved into the 100,000 genome project, announced last Nov. Now high level talks take place to design the network for the genetics medicine in UK. HVP work (proposal submitted last year) has not yet been able to attract funding support. Efforts are being made to include HVP in the overall UK strategic plan. UK node continues to develop databases, create links to automatically collect data from labs by tools/software and work on the quality assurance of the data. Phenotype data are also collected.

## **7. Human Variome Project/China Country Development Program**

To date, two applications (Austria/Czech/India and Belgium/D.R.Congo) have gone to the review panels set up by the International Scientific Advisory Committee, pending final decision. Several other applications are in the process of being put in the form that will be reviewed by the International Scientific Advisory Committee. There will be more applications coming in due course.

Discussions were held in regards to the process of the applications. It was suggested that a clear guideline is needed for future applicants, especially in the areas of application requirements and budget. It was also suggested that all applications should go to the International Scientific Advisory Committee for review. Timothy Smith explained that, as stated in the guidelines, an initial assessment was conducted by ICO staff to make sure the application met the guideline requirements. ICO staff will review and refine the guidelines over during the next 2 months.

A question was raised about whether a deadline was needed for application submissions. Timothy Smith explained that the reason for not having a cut-off date was to allow countries to form long-term partnerships but not rush into looking for funding partners only.

*Supplementary information from Helen Robinson has been provided as an attachment to these minutes.*

## **8. Standards Development Process/Activity Proposals**

No Working Groups currently chartered

No Activity Proposals received

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

None

**10. World Health Organization consultations**

Timothy Smith reported briefly that the ICO had embarked on a joint project with the Foundation for Genomics and Population Health (PHG Foundation) in the UK and the WHO Secretariat in Switzerland to attempt to establish a case for a global program on genomics and public health. There is the possibility that if a case for such a program can be made, WHO will establish such a program. The project is being overseen by a steering committee consisting of staff from each of the three participating organisations. As part of this joint project, staff from the ICO have been conducting public consultations at major human genetics meetings in 2012 (ASHG 2012, APCHG 2012), with more planned in 2013. In addition, a series of one-on-one telephone interviews will be conducted with invited individuals over the next few months. The outcomes of these consultations and interviews will be distilled into a report for the WHO Assistant Director General - Health Systems and Innovation.

*A more complete report by Helen Robinson has been provided as an attachment to these minutes.*

**11. Travel subsidies for country node representatives**

A question was raised regarding travel subsidies for Node and database representatives to attend face-to-face meetings. Heather Howard explained that it is not possible to subsidize members of both councils to attend meetings into the future as already the number is over 60. There could however, in the future, be some funds allocated to assist representatives from low- and middle-income countries to attend.

**12. Other matters**

No other matters discussed.

**13. Next Meeting**

16 April, 2013 via GoToWebinar

Meeting Closed 12:55AM Melbourne, Australia time

## **Attachment A: HVPCCDP**

The HVPCCDP is a very new grant program, being launched in July 2012. It is designed to assist in the establishment of HVP Country Nodes. When the program was created, the information package, the application form and the process for handling applications was circulated for comment among members on HVP committees and structures including the ISAC and ICCAC at the time. The final design incorporated these comments and suggestions. Final approval was given by the HVP Board and the donor of the funds.

The current Call for Applications is dated April 2012 (see attached) and contains all the information relating to the program and sets out the process for making applications. The process for assessing applications is set out on page 2 of the document.

To date, nine applications have been received; two have been considered by Panels; two are currently being considered; two have been withdrawn by applicants and the remainder are being redrafted by the applicants before going to a Panel. There have also been inquires from nine other groups requesting advice before making an application, of which we believe five are actively preparing their applications at the moment.

Being a new program, the application package can always be improved. The April 2012 version is designed to be used for 12 months to gain some experience before reviewing the process and the quality of the information it contains. April 2013 might be a good time to do this.

DRAFT

## Attachment B: HVP and World Health Organization (WHO)

Since the establishment of HVP, it has had a good relationship with WHO, with WHO representatives attending HVP meetings for example they co-hosted the Melbourne meeting in 2006, but the relationship has never been formalized. Many HVP members would like to see a more formalized relationship because that would help promote activities related data sharing with their national ministries of health. Since 2011, HVP has sought to develop a more formal relationship with WHO.

WHO admits that it has not had a strong human genetics/genomics and public health program and would like to address this gap in its programs. WHO undertook its *Grand Challenges in Genomics and Public Health in Developing Countries* project in 2010 as a means of gathering information from the broader research community as a basis for defining the mandate for a human genomics and public health program at WHO. Many HVP members are involved in this process.

In 2012, HVP approached WHO and offered assistance in defining an international human genomics and public health program at WHO with a view to strengthening the relationship between HVP members and WHO. WHO established a Steering committee involving HVP, and PHG Foundation UK, and chaired by WHO. This committee agreed to carry out a joint consultative project. This project began in September 2012 and activities undertaken so far include:

- 1 WHO's Grand Challenges survey was promoted through the HVP web-site; a draft report has been prepared by WHO and HVP has been invited to make comments
- 2 A consultation was held at ASHG in San Francisco in November 2012, chaired by Garry Cutting, Chair of HVP ISAC
- 3 A second consultation was held at APHG in Kuala Lumpur in December 2012, chaired by Richard Cotton, Scientific Advisor to HVP
- 4 A series of structured interviews has begun to further develop ideas from the consultations; HVP is conducting some of these interviews
- 5 Further consultation sessions are planned for HUGO (Singapore) and Mutation Detection Symposium (Canada) both in April, at EHGS in Paris in June, and at WHO in June; these consultations will incorporate the results from the key informant interviews

The outcome of this project will be recommendations regarding the mandate for an international program on human genomics and public health, how the program might work, who should be involved and how such a program could be made sustainable. The project will report to Assistant Director EIP, Dr Marie-Paule Kierny, at WHO sometime in late 2013 or early 2014.