



sharing data · reducing disease

## MINUTES

### International Confederation of Countries

#### Advisory Council

15<sup>th</sup> July, 2012, UNESCO Headquarters, Paris, France

#### Attendees

##### HVP Country Node Representatives

Heather Howard	Australian Node Representative
Martina Witsch-Baumgartner	Austrian Node Representative
Willy Lissens	Belgian Node Representative
Ming Qi	Chinese Node Representative
Xitao Li	Chinese Node Representative
Andreas Hadjusavvas	Cypriot Node Representative
Sherifa Ahmed Hamed	Egyptian Node Representative
Makia Marafie	Kuwaiti Node Representative
Zilfalil bin Alwi	Malaysian Node Representative
Tilak Shresta	Nepalese Node Representative
Maria Jesus Sobrido	Spanish Node Representative
Chí Dũng Vũ	Vietnamese Node Representative

##### Human Genetics Society Representatives

Feliciano Ramos	President, Spanish Society of Human Genetics
Raj Ramesar	Liaison & Representative, African Society of Human Genetics
Vaidutis Kučinskis	Lithuanian Society of Human Genetics
Thomy de Ravel de l'Argentiere	President, Belgian Society of Human Genetics

##### Human Variome Project Interim International Scientific Advisory Committee

Mona El-Ruby	Egypt
Mauno Vihinen	Finland
Mireille Claustres	France
Aida Falcón de Vargas	Venezuela

##### Human Variome Project Staff

Richard GH Cotton	Scientific Director, Human Variome Project
Timothy Smith	Communications Officer, Human Variome Project
Julia Hasler	UNESCO Liaison
Helen Robinson	WHO Liaison

Fan Li Liaison Officer - China

### **Others**

Peter Taschner The Netherlands

Mitali Mukerji India

Rosemary Ekong UK

Maurizio Genuardi Italy

Thong Meow-Keong Malaysia

Collet Dandara South Africa

Jun Chen China

### **Apologies**

George Patrinos Greek Node Representative

Fahd Al Mulla Chair

Meeting Opened 1030

### **1. Welcome New Chair**

It was reported that Fahd Al Mulla was elected Chair of the Council, for the next 2 years. As Fahd was unable to attend the meeting, Andreas Hadjisavvas (Cyprus Node) served as Chair in his absence.

### **2. Welcome new Members**

It was reported that 3 new Country Nodes have joined the HVP since the last meeting: United States of America, United Kingdom, and Czech Republic.

### **3. Confirmation of minutes of previous meeting – Beijing**

The minutes were approved without modification.

### **4. Issues arising from previous meeting**

It was reported that during HVP4 a number of bilateral meetings were held between permanent delegations to UNESCO and consortium members from relevant countries in conjunction with the International Coordinating Office and the Natural Sciences Sector of UNESCO lead by Dr Nalecz. These meetings were a great success and it should be continued and extended at other meetings. It was also encouraged that HVP country nodes representative should contact UNESCO delegates in their countries to work together and get their help for actions towards promoting the specific country node. E.g. Dr. Ramos and Dr. Sobrido, who are organizing a session on the HVP to be held within the meeting of the Spanish Society for Human Genetics in 2013, commented that they will invite the UNESCO delegate in Spain to join that meeting.

## **5. Report from Country Node Development Workshop**

Timothy Smith (ICO) provided a brief report from the Country Node Development Workshop that was facilitated by the International Coordinating Office on the 11th June, 2012.

The goal of the Country Node Development Workshop was to assist countries wishing to initiate an HVP Country Node. The Workshop aimed to begin a conversation towards establishing a definition for HVP Country Nodes, and to provide an overview of available systems and organisational models. The Workshop revealed that more work is needed before a definition can be proposed to the International Confederation of Countries Advisory Committee.

Based on the discussions and outcomes from the Workshop, the International Coordinating Office will undertake to develop a discussion paper that proposes a draft definition of an HVP Country Node and the steps required to initiate a Node. This Discussion Paper, with the consent of the ICCAC will undergo a formal consultation process of the Human Variome Project Consortium before being finalised and presented to the ICCAC for further action.

In the meantime, the ICO will begin assembling a comprehensive list of systems, materials and information that Country Nodes, both existing and potential, may find useful. Such a list would cover: database management systems, regulatory frameworks operating in various regions, potential solutions for operating under such frameworks, lobbying and educational materials, etc. The ICO will also continue its work with both UNESCO and WHO to encourage member states to develop proactive policies around variant information collection and sharing. However, Consortium members from these member states will also need to be active in their own countries, if effective policy changes are to occur. To do so, Consortium members should organise themselves and other stakeholders in their country: i.e. clinical genetics and human genetics societies and their members, diagnostic laboratories, clinical geneticists, patient groups and government officials.

## **6. Report from HVP 4 Meeting**

The session on HVP Country Nodes (CNs) at the HVP4 meeting had the main objectives of working towards a conceptualization of HVP CNs, and the requirements for initiating, accrediting and monitoring accomplishments. Helen Robinson and Timothy Smith from the International Coordinating Office presented a draft position paper<sup>1</sup> that proposed a system for assessing the development of CNs, including potential stepwise indicators to monitor progression within the framework of *Project Roadmap 2012-2016*. This position paper was discussed for refinement and produce summary recommendations for the ICCAC and the International Scientific Advisory Committee.

In answer to the first objective, “Does the Country Node approach help the HVP?” the need was uniformly recognized for developing a structure within the HVP that is representative of geographical

and political organizations (at country, region and continental levels), and so these organizational elements (i.e. Country Nodes) will be very important to achieve the goals of the HVP. However during the discussions important challenges were identified, the first of which was to come to an agreement of what an HVP CN is. It seems that CNs may have different goals in different regions of the world. In rich countries, where genetics services and diverse administrative and scientific infrastructures related to genetics are already in place, it is more likely that CNs will mainly exert a catalysing or coordinating role (promoting and coordinating variant collection, encouraging variant submission by the laboratories to existing data repositories, fostering participation in disease-centred consortia, etc.). It is unlikely that all HVP-related activities will be centralised in developed countries, however specific collecting activities may be centralized (the French muscular dystrophy organization, explained by Dr. Claustres, is a good example of this). In developing countries however, HVP CNs will likely need to have a primary role in improving the delivery of genetic services; here the CNs will probably have a role as genetic education centres and a more direct action in actually carrying out variant collection in those countries in a more centralized manner.

This heterogeneous concept of what an HVP CN should be and do, leads to the subsequent problem of defining what form accreditation should take and the requirements by which to accredit CNs. Care must be taken to ensure that CNs are supported in their heterogeneous needs when monitoring their achievements. In response to the why do CNs have to be accredited, it seemed clear from the discussions that some type of "official" certification procedure is clearly needed in order to regulate the good functioning of the HVP, also emphasizing that this accreditation as HVP CNs will not only serve as internal quality control within the HVP and an internal measure of progress, but such an "accreditation stamp" will also serve the "accredited" CNs to be recognizable spokespeople for the HVP in their regions, to address the national administrations, etc. However, given that the general goals and principles of an accreditation process for the HVP CNs still need final clarification, the specific instruments, procedures, the "what" and "how" to measure, frequency of evaluation, scoring method and resources needed for such a compliance monitoring system will only come afterwards. Regardless of what the accreditation and evaluation process of CNs will entail, it was suggested that words with an implied negative meaning of "not passing" should be avoided and that this process should be oriented to encourage participation.

Two keynote conferences were presented by Drs. David Weatherall (Weatherall Institute of Molecular Medicine) and Myles Axton (editor, Nature Genetics). From very different professional routes, both speakers exemplified the key role of sharing genomic knowledge for the progress of health worldwide, and very especially the moral obligation to an open sharing of the results of genomics research carried out in rich countries to facilitate the return of advancements and building of capacities in resource-poor areas, from which often so much knowledge is obtained.

## **7. Standards Development Process**

The draft matrix for Assessing Genomic and Genetic Services in a Country that was presented during HVP4 was discussed. The Council recommended that all members attempt to use the tool to establish the base line for each individual country.

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

1. The state of genetics - and of research and healthcare provisions in general - in individual countries was presented by participants at length and, as expected, there is a broad variability. The role of the HVP should obviously not be to substitute for the tasks and objectives corresponding to other administrative, medical and scientific, national and international organizations (including national health and educational systems) that already exist. Instead, the HVP should work in coordination with these existing organizations for the improvement of the use of genetics in healthcare, specifically in the task of collecting and sharing genetic knowledge to improve healthcare and, since there is an exponential increase in ELSI issues related to the portentous speed of data collection and IT capabilities for storing and sharing, the HVP should take a world leadership role in the ELSI issues of Molecular Medicine.

Although this first recommendation may not be a specific outcome from the Country Node session, but more of a general recollection from the whole HVP4 meeting, keeping that in mind is key in order to being able to discuss the CNs, their shape and role. Since most relevant stakeholders the HVP will have to partner with to accomplish the HVP's main goals will be country-level organizations, the development of HVP country (or regional) nodes will be very important.

2. Although it should be possible to have a simple, operational definition of what a HVP CN is, it will probably be much more difficult (perhaps not possible) to have only one set of goals they should accomplish, and at what pace.
3. It is unlikely that there will be just one centralized repository of genetic information in developed countries, or even that all HVP-related activities will be centralized in these countries. Thus, in some cases the possibility of having more than one HVP node in a given country (e.g. with differentiated roles) might be appropriate.
4. Given the above, it will be difficult to define a unique and simple process for accreditation and evaluation of CNs, what will be measured, who will be accrediting, etc. Different "layers" of evaluation might be a solution.
5. It is necessary to avoid too strict requirements to receive the "stamp" of an accredited HVP-CN as this and subsequent evaluation requirements might be discouraging to potential participants. At this point of the HVP evolution, everyone is needed and the developing of country and regional nodes should be encouraged.
6. Perhaps a "self-evaluation" type of process might be more appropriate at this point.
7. Actions aimed at helping to start CNs, tools to assist members in their work, and to help secure sustainability of CNs should be among the priorities of the HVP.

8. Promoting engagement with appropriate stakeholders in each country / region is among the first and foremost actions that CNs should undertake, with the help of the HVP ICO. For this purpose, functionalization of the CNs (identifying a specific, measurable, short-term goal to show the usefulness of this endeavour) may be the best way to capture the interest of local authorities and engagement of relevant stakeholders. Regardless of the time it may take to come to an eventual agreement on a definition of a CN and the accreditation process, being "accredited" or "entitled" as HVP spokesperson /spokesgroup in a country will be important for the HVP representatives in that given country, so that they can efficiently approach local authorities and stakeholders.

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

China will make US\$1 million dollars available for dispersal by HVPI to assist the development of Nodes with an emphasis on developing Nations. The grant options were discussed and a number of member reported that they will be forming partnerships to apply for grants. The application process is open on the 1st July.

#### **10. Other Matters**

No other matters discussed.

#### **11. Next Meeting**

A further meeting was proposed for those members attending the American Society of Human Genetics in December. The ICO also promised to look into the possibility of conducting virtual meetings.

Meeting Closed 1230