

# MINUTES

Gene/Disease Specific Database Advisory Council  
Teleconference  
Tuesday 13 September 2016  
1200hrs UTC/GMT



## MEMBERS

### **Present**

Peter E M Taschner (Chair)  
Raymond Dalgleish  
Rosemary Ekong  
Mary Fujiwara  
Mauno Vihinen

### **ICO Staff**

Timothy D. Smith (Scientific Programs Manager)  
Helen Robinson (Consultant)

### **Apologies**

Olubunmi K D Abel  
Ammar Al-Chalabi  
Stefan Aretz  
Arleen Auerbach  
Timothy Barret  
David Baux  
Jean-Pierre Bayley  
Daniel Bichet  
Nenad Blau  
Nancy Braverman  
Paola Carrera  
Johan T den Dunnen  
Pascal Escher  
Almudena Fernández

Marc Ferre  
Bruce Gottlieb  
Daniel Hampshire  
Tamas Hegedus  
Raoul Hennekam  
Alex Hewitt  
Ammar Husami  
Sarah E A Leigh  
Derek Lim  
Finlay Macrae  
Eamonn Maher  
Lluis Montoliu  
Etienne Mornet  
Magali Olivier  
Sue M Povey  
Yves Sabbagh  
Judith Anne Savige  
Sarah Sim  
Carli Tops  
Ronald Trent  
Richard van Wijk  
Katarzyna Wertheim-Tysarowska  
Tom Winder  
Bing Yu  
Martina Witsch-Baumgartner  
Johannes Zschocke

## AGENDA

### **1. Welcome**

### **2. Apologies**

Apologies were noted as above.

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

Minutes were confirmed unanimously.

### **4. Reports of actions since last meeting**

#### **a. Letter to National Human Genetics Societies**

Members were reminded of the importance of this issue to all database curators, and how action can be taken by using the template letter and instructions. It was noted that the topic of promoting the use of standardized descriptions of human genome sequence variant data was increasing in

importance as governments in various parts of the world – USA, Australia, EU among others – were raising the issue of regulating the collection of variant information in different ways. The collection of information in data systems requiring international collaboration and curation was likely to have a much higher profile in the future, hence laying out the foundations for enabling international sharing was increasingly important.

Several members of the Council have sent letters as have members of all other HVP Councils. During September and October the topic will be taken up societies in the Netherlands, UK, with dates already set for discussion. ICO will keep records of who has sent letters, together with contact details to up-date the HVP data base. Members were asked to ensure that ICO was informed of all actions taken. It is also important to monitor any of the barriers to action that is any arguments that are raised that indicate why the relevant professional bodies cannot support this need to be reported to this Council so that these can be overcome.

The template letter and accompanying instructions will be circulated with the record of this meeting for convenience of members.

## **5. Project Roadmap 2016-2020 Objectives and Actions**

### **a. Action 1.3: Building relationships between Council members – Timothy D. Smith**

The need to promote more effective relationships between the members of the various HVP Councils was raised. This action was agreed as part of a series of activities that will strengthen the ties between the group of international database curators and those generating data in various parts of the world. It is known that many of the HVP Country Nodes are looking to make places to house data that is generated in their countries and that they are looking for the right people to connect with to make this happen. This will be taken up in more detail at the next meeting but in the meantime members are asked to consider if there are any countries that they would particularly like to connect with in order to capture new sources of data.

### **b. Objective 5: The next generation of scientists – Helen Robinson**

This item was introduced for discussion at the last meeting and has since been discussed at each of the HVP Councils. With the start of the new academic year underway in the Northern hemisphere, several members noted that they were about to commence lectures and seminars with various student groups where the importance of human genome sequence variant data generation and sharing could be both raised and discussed. The various skills required to do this can also be taught and assessed. Members agreed to share information on presentations, assessment tools and the like directly between themselves. In accordance with the plan supporting Objective 5, the ICO will be keeping a list of all those actions that HVP members are currently undertaking as part of the mapping exercise (Stage 1). Any members of this Council who were unable to join the call are asked to send a short description of any relevant activities that they are involved in – for undergraduate students, post-graduate students and early-career education - to Helen Robinson – hmro@unimelb.edu.au - so that they can be added to the list for future discussion of this issue.

## **6. Standards Development Process**

### **a. WG08: Ethics Checklist for LSDB Curators and Submitters – Rosemary Ekong**

Council members were reminded of the ongoing consultation on the Working Draft produced by this Working Group and were asked to submit any comments through the Human Variome Project website.

## **7. Responses to public consultations/calls for comments**

### **a. FDA (USA): Use of Public Human Genetic Variant Databases to Support Clinical Validity for Next Generation Sequencing (NGS)-Based In Vitro Diagnostics**

GA4HG draft response to be circulated; other Councils have been discussing this and an HVP response was being developed with the following issues being included:

- The quality focus mentioned is to be commended and supported
- Need for resources to carry out the functions being suggested, particularly international curation activities
- Need to address the problem of existing databases that may not meet any new standards and requirements, with the suggestion of that rather than expend resources on up-grading old data collecting systems, they be 'grandfathered'

Any other suggestions are to be sent to Tim Smith [tim@variome.org](mailto:tim@variome.org) before Monday 26 September.

## **8. Future Meeting Dates**

- HVP Members' Meeting – 20 October 1800-2000hrs (local) – Cheakamus Room, Fairmont Waterfront, Vancouver, Canada
- 8 November – Teleconference