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# MINUTES

Gene/Disease Specific Database Advisory Council  
Tuesday 20 January, 2015  
1200-1300hrs UTC/GMT

## MEMBERS

### Present

Peter E M Taschner (Chair)  
Arleen Auerbach  
Mauno Vihinen  
Tamas Hegedus  
Daniel Hampshire  
Sarah E A Leigh  
Rosemary Ekong  
Mary Fujiwara  
Raymond Dalgleish  
Yves Sabbagh  
Marc Ferre

### ICO Staff

Timothy D. Smith

### Apologies

Ammar Al-Chalabi  
Stefan Aretz  
Timothy Barret  
David Baux  
Jean-Pierre Bayley  
Daniel Bichet  
Nenad Blau  
Nancy Braverman  
Paola Carrera  
Johan T den Dunnen  
Eamonn Maher  
Pascal Escher  
Bruce Gottlieb  
Raoul Hennekam  
Alex Hewitt  
Ammar Husami

Derek Lim  
Finlay Macrae  
Etienne Mornet  
Magali Olivier  
Olubunmi K D Abel  
Sue M Povey  
Judith Anne Savige  
Sarah Sim  
Carli Tops  
Ronald Trent  
Richard van Wijk  
Katarzyna Wertheim-Tysarowska  
Tom Winder  
Bing Yu  
Martina Witsch-Baumgartner  
Johannes Zschocke

## 1. Welcome

Peter Taschner welcomed members to the meeting.

## 2. Apologies

Apologies were noted as above.

## 3. Welcome new members

There were no new members.

## 4. Confirmation of minutes of previous meeting

The minutes of the previous meeting were confirmed unanimously.

## 5. Matters arising from the previous meeting

Peter Taschner advised the Council that he has moved to a new appointment at the University of Applied Sciences Leiden. Raymond Dalgleish proposed congratulations on behalf of the Council.

### **a. WG08: Ethics Checklist for Gene/Disease Specific Database Curators and Submitters**

Rosemary Ekong reported on the progress of the Working Group. A survey has been sent out to curators, seeking information on how ethical concerns are currently handled in databases and the extent to which the Povey et al. recommendations are being applied. To date approximately 10 responses have been received in the past week. A total of 177 curators have been invited to participate. Potential respondents will be given a total of five weeks to respond. Reminder emails will be sent by the ICO.

**b. Merging existing lists of LSDBs**

Peter Taschner reported that the International Scientific Advisory Committee have taken the position that the LSDB list maintained at LUMC is the most comprehensive list. This list is now linked to from the Project website (see <http://www.humanvariomeproject.org/resources/resources.html>). Further discussion regarding the merging of LSDB lists is dependent on the outcome of decisions by the HGVS Board.

**c. Increase awareness of journals to nomenclature and data sharing**

Peter Taschner reported that the ICO has published a list of journals in the genetics and genomics field on the HVP website that indicates whether their author instructions contain language regarding the use of the HGVS nomenclature and submission of data to public databases prior to publication. Peter suggested that the next step would be to also check if each of the journals enforces these instructions and track this information over time. He mentioned the possibility of engaging students from the University of Applied Sciences to assist with this work. Raymond Dalgleish requested the assistance of Council members in looking for changes in the author instructions of the journals listed and ensuring that the instructions are enforced. He also noted that some authors are reticent to have their data publicly visible in a database at the time of manuscript review, and that perhaps journals should merely require that the data be submitted but not public.

**6. Report from Chair**

Peter Taschner reported that at their last meeting in December, the International Scientific Advisory Committee approved *HVP/DA/002-02/EN: Disclaimer statements for Gene/Disease Specific Databases* for publication as an HVP Guideline. This version included changes requested by the Council to the language of the disclaimer statement. The published Guideline is available on the Project website (<http://www.humanvariomeproject.org/solutions/>)

**7. Gene/Disease Specific Database Activities**

**a. Rosemary Ekong**

Rosemary Ekong reported that Transgenomic have agreed to share data with the TSC database.

**b. Tamas Hegedus**

Tamas Hegedus reported that although the ABC protein mutation database is protein-centric there is a need to incorporate sequence data in non-coding regions and they are planning to incorporate this in the next couple of months. He also reported a new collaboration with a clinical group.

**c. Mauno Vihinen**

Mauno Vihinen informed the Council about a new general database Varisnp—a benchmark database that contains known benign variants from dbSNP. The variant descriptions include mapping to reference sequences on chromosomal, genomic, coding DNA and protein levels. The datasets will be updated with automated scripts on a regular basis and are freely available at <https://structure.bmc.lu.se/VariSNP>.

**d. Peter Taschner**

Peter Taschner reported that the curators of the TCA cycle gene database have not received any objection from submitters to the database contents being licensed under the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 International license (<https://creativecommons.org/licenses/by-nc-sa/4.0/>). The database website will be updated soon to reflect this change. In addition, the database will adopt the HVP Guideline on disclaimer statements.

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

No recommendations were made.

## 9. Other matters

Raymond Dagleish raised two issues for consideration.

In line with the article published by Garry Cutting in the *American Journal of Human Genetics* (see [Cutting, 2014, Am J Hum Genet 4:5-10](#)), the Council should consider recommending that the International Scientific Advisory Committee adopt a policy on behalf of the Human Variome Project on the terminology used to describe variants. Specifically, that the term “variant” be used, not the term “mutation.” Arleen Auerbach noted that the American Society of Human Genetics and the American Society of Medical Genetics and Genomics have similar policies. Mauno Vihinen noted that the HGVS nomenclature contains guidance on this issue, as well as guidance to avoid the term “polymorphism.”

The Council should consider recommending that the International Scientific Advisory Committee adopt a policy on behalf of the Human Variome Project on exon numbering. The HGVS nomenclature contains no guidance on exon numbering while the LRG specification allows for both a sequential numbering scheme and an alternate scheme.

The Council asked Raymond Dagleish to draft language to be considered on both of these issues for consideration at a later meeting.

<b>ACTION:</b> Raymond Dagleish to draft policy recommendations on variant terminology and exon numbering.
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## 10. Next Meetings

- Tuesday 17 March 2015 1200hrs GMT/UTC.

Council Members are asked to respond to the poll accompanying these minutes regarding preferred days of the week so that future meeting dates can be set.