



THE  
**HUMAN VARIOME**  
PROJECT

sharing data · reducing disease

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# MINUTES

Gene/Disease Specific Database Advisory Council  
Tuesday 15 March, 2016  
1200-1300hrs UTC/GMT

## MEMBERS

### **Present**

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

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

Eamonn Maher  
Etienne Mornet  
Magali Olivier  
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

### **ICO Staff**

Helen Robinson  
Catherine Carnovale

### **ICO Staff**

Timothy D. Smith

### **Apologies**

Olubunmi K D Abel

## AGENDA

### 1. Welcome

### 2. Apologies

Apologies were noted as above

### 3. Confirmation of minutes of previous meeting

Raymond Dalgleish raised a concern with the use of the word "polymorphism" in Item 5 of the previous minutes which read:

**Mary Fujiwara noted that further clarification is required when describing the frequency of a polymorphism. While the report provides a frequency of 0.01%, this should be clarified to be 0.01% within a defined population due to differences in prevalence of certain polymorphisms between populations. Mauno Vihinen agreed with this point, however he noted that the current recommendation was not to use the term polymorphism. It was**

**agreed that the term would remain, however Mauno would make a clarification about the population such that it is used correctly.**

Raymond requested the minutes be revised to have the word variant in brackets. Furthermore it was requested that the term polymorphism be avoided in the minutes, and that further clarification is required for this term.

#### **4. Report from Chair – Peter Taschner**

Peter Taschner updated the Council on the progress of the University of Applied Sciences' investigation on the use of correct nomenclature and submission to databases. At this stage, some of the information is currently only in Dutch, however the plan is to have this material translated into English.

#### **5. Project Roadmap 2016-2020 - Helen Robinson**

Helen Robinson informed the Council that the ICO has received comments on the current draft of the Project Roadmap. All comments received are resolvable and do not require major changes to be made. The most notable change will be to include an effort on develop a greater emphasis on using variant data for research. Peter Taschner suggested the mention of FAIR data principles (Findable, Accessible, Interoperable, and Re-usable) in the draft. Helen said that this was suggested by other Consortium members and would be included in the next iteration. Helen invited comments from the Council regarding the suggestion that they work more closely with the ICCAC on certain issues to further activities.

#### **6. WG07-Minimum Content Requirements for Country Specific Databases - Request for assistance - Peter Taschner**

Peter Taschner circulated the draft document describing the Minimum Content of a G/DSDB, developed by WG07 for comments and suggestions however he did not receive any feedback prior to the meeting. Helen Robinson suggested that the draft be recirculated to the Council with the minutes and that replies be sent to Peter prior to the next meeting of the ISAC on 24<sup>th</sup> March 2016. If comments are not received, it will be assumed that members of the Council are in agreement with the draft.

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

Raymond Dagleish informed the Council that he has been involved in the development of a new tool to verify variant descriptions called variantvalidator (found at [variantvalidator.org](http://variantvalidator.org)). He encouraged Council members to visit the site and provide feedback. Raymond Dagleish explained that it is not a replacement for Mutalyzer. Peter Taschner mentioned that a current nomenclature related issue for Mutalyzer relates to the current lack of formal specification. He noted that many groups are encountering similar issues due to the lack of formal rules which can be clearly applied to each case. This has implications for the prediction of variants, and could create instances where different results are generated from using different tools (particularly when checking combinations of variants).

Raymond Dagleish mentioned that the 2016 update of HGVS Nomenclature has been published online in Human Mutation for download. He went on to mention his concerns about a paper which was published in Genome Medicine in July 2015 titled "*CSN and CAVA: variant annotation tools for rapid, robust next-generation sequencing analysis in the clinical setting.*" He shares his concerns that the group have attempted to develop a new nomenclature system, which is being promoted as an alternative to HGVS. Raymond offered to circulate the link for the paper to the Council.

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

Peter Taschner expressed his wish that the ISAC formulate a new Working Group concerning the formalization of nomenclature guidelines. Raymond Dagleish asked for clarification of the new Working Groups goals and how they will differ from the Sequence Variant Descriptions Working Group. This matter is to be discussed at the next meeting after Peter has developed these ideas.

## **9. Other matters**

Helen Robinson asked if any Council members were going to be present at ICHG in Kyoto. While none of the members present on the call were planning to attend, Helen informed the Council that there are various HVP related events scheduled as part of the ICHG2016 program and any member planning on going is welcome to attend.

Helen raised the issue of whether the Council would like to meet to discuss various issues at HVP6. Helen will liaise with members of the Council and the ICO to work out the best time.

Tamas Hegedus informed the Council on the progress of updates to the ABC transporter mutation database. The plan is to update the C1-inhibitor database next, with the intention to progress to an LOVD installation.

Helen Robinson updated the Council on the activity of Global Globin 2020. The plan is to update the existing database in Cyprus, moving it to LOVD3 with the help of Johan den Dunnen and present it at the GG2020 session at HVP6. It is hoped that approximately 25 countries will contribute to this database.

## **10. Next meetings**

10 May - Virtual

3 June – Face-to-face at HVP6

12 July - Virtual

13 September - Virtual

8 November - Virtual