



THE  
**HUMAN VARIOME**  
PROJECT  
sharing data · reducing disease  
an NGO Official Partner of UNESCO

# MINUTES

Gene/Disease Specific Database Advisory Council  
Tuesday 8 September, 2015  
1200-1300hrs UTC/GMT

## MEMBERS

### **Present**

Peter E M Taschner (Chair)  
Arleen Auerbach  
Raymond Dalgleish  
Daniel Hampshire  
Sarah E A Leigh  
Yves Sabbagh  
Mauno Vihinen

### **ICO Staff**

Timothy D. Smith

### **Apologies**

Olubunmi K D Abel  
Ammar Al-Chalabi  
Stefan Aretz  
Timothy Barret  
David Baux  
Jean-Pierre Bayley  
Daniel Bichet  
Nenad Blau  
Nancy Braverman  
Paola Carrera  
Johan T den Dunnen  
Rosemary Ekong  
Pascal Escher  
Marc Ferre  
Mary Fujiwara  
Bruce Gottlieb  
Tamas Hegedus  
Raoul Hennekam

Alex Hewitt  
Ammar Husami  
Derek Lim  
Finlay Macrae  
Eamonn Maher  
Etienne Mornet  
Magali Olivier  
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

## 2. Apologies

Apologies were noted as above.

## 3. Confirmation of minutes of previous meeting

Minutes were confirmed unanimously.

## 4. Matters arising from the previous meeting

### ***a. Policy recommendations on variant terminology and exon numbering – Raymond Dalgleish***

The Council discussed the Working Draft prepared by Raymond Dalgleish and approved its move to the next stage of the Standards Development Process. The Council asked the ICO to publish the Working Draft as an Exposure Draft and facilitate the required public consultation.

## 5. Report from Chair – Peter Taschner

Peter Taschner reported on the progress of the manuscript on the database quality scheme that was submitted to *Human Mutation*. The journal has asked for the manuscript to be resubmitted for their 25<sup>th</sup> anniversary special issue,

with a deadline of November 1.

Peter Taschner further reported on ongoing efforts within Council and ISAC to move the issue of database quality accreditation forward. The ISAC are currently looking at what other groups and individuals should be a part of this effort.

#### **6. Project Roadmap 2016-2020 – Timothy Smith**

Timothy Smith provided the Council with an update on the Roadmap Development process. He thanked the Council members for their participation in the process so far by providing feedback on the list of “thoughtstarters” sent to them by the HVPIL Board. The feedback received will be incorporated into a consultation document by the ICO that will be released to the Consortium early in October.

#### **7. Solution Blueprint Edition 1 – Timothy Smith**

The Council considered the draft content for the first edition of the Solution Blueprint. The Council asked to consider the matter again once comments received from Council members had been incorporated into the document.

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

Arleen Auerbach reported that the Fanconi Anemia database has been expanded to cover 19 genes.

Mauno Vihinen reported that his team is in the process of translating the ID bases to LOVD; a timeframe for this process is not yet available.

Peter Taschner reported that the LOVD team is working on tools to migrate databases from version 2 to version 3. There are still some issues to resolve how to map between the data models, primarily with respect to custom fields. Assistance is still available to curators wishing to migrate

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

The Council resolved to inform the ISAC of the status of the draft Guidelines for variant terminology and exon numbering.

#### **10. Other matters**

No other matters discussed.

#### **11. Next Meetings**

- Tuesday 10 November 2015 1200hrs GMT/UTC