

# MINUTES

Gene/Disease Specific Database Advisory Council  
Tuesday 10 November, 2015  
1200-1300hrs UTC/GMT

## Members

### **Present**

Peter E M Tashner (Chair)  
Arleen Auerbach  
Raymond Dalgleish  
Nenad Blau  
Rosemary Ekong  
Sarah E A Leigh  
Mauno Vihinen

### **ICO Staff**

Timothy D. Smith  
Catherine Carnovale

### **Apologies**

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

Ammar Husami  
Derek Lim  
Finlay Macrae  
Eamonn Maher  
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 Zschock

## Agenda

### 1. **Welcome**

### 2. **Apologies**

Apologies were noted as above

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

Minutes were confirmed unanimously

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

Peter Taschner reported on the progress of the preparation of a manuscript on the data quality parameters (HVP/GL/001-01/EN). He noted that Mauno Vihinen has made some changes and that the submission deadline is on Sunday—further updates are to be made before this time.

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

Timothy Smith provided the Council with an update on the Project Roadmap development. During October, the ICO ran consultations for the Project Roadmap asking for contributions from consortium members on a range of

issues. Using these responses, the ICO will prepare a draft version for the new Project Roadmap during November which will be made publically available for comment upon completion.

It was noted that while approximately 30 members made comments through the online portal, this was combined with comments gathered from the consultation event run at ASHG as well as other informal events which overall provided a good cross section of opinions.

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

Timothy Smith asked the Council to review the content of the Solution Blueprint now that the requested changes from Raymond Dalgleish have been incorporated. He provided the Council with credentials to view the content online, but asked them to keep in mind that the site is currently very barebones, with no design elements yet in place. Formatting and content updates will be made over the new few weeks. Council members were encouraged to make comments on the content and submit them via the “Help improve this page” link on each page of the site.

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

Arleen Auerbach reported that the Fanconi Anemia database has been expanded to 19 genes with help from Johan’s group.

Mauno Vihinen reported that his group is continuing with the transfer of the ID databases into LOVD; BTKbase has been released already and they are working on the other 130 databases.

Peter Taschner reported that he has been in contact with Thierry Soussi and that the TP53 field has decided to prepare a manuscript for a Nature Reviews Clinical Oncology about the reporting and description of variants, as well as the screening of TP53 gene. They hope to achieve greater standardization of the field. He reported that he has been advising them on issues regarding the use of HGVS nomenclature, in particular the reporting of somatic variants.

Rosemary Ekong reported on progress of the ethics checklist working group, which has been slow in recent months, but will be renewed as of now. Rosemary Ekong noted that the membership of the Working Group had reduced to only include Mauno Vihinen and herself. She further noted that Dr Mats Hansson of the Centre for Research Ethics & Bioethics in Uppsala (Sweden) was contacted, however he felt that he would not be able to participate due to time constraints.

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

The Council resolved to share the data quality parameters manuscript with the ISAC once it has been finalized.

#### **9. Other matters**

Sarah Leigh reported to the Council on a new process implemented by the Elsevier journal that she is involved with. Submitting authors now have an option during submission to declare if they have submitted the data contained in the publication to a database. Sarah Leigh noted that she is planning to review the process shortly to determine how many authors are actually submitting their data, and will report to the Council.

Peter Taschner mentioned that European journals may have a stronger preference for data to be submitted to databases prior to publication although this will be up to the individual editors and journals.

Timothy Smith mentioned that in the journal editors meeting at ASHG, the editors that were present gave broad support for encouraging, but not requiring, submission of data prior to publication.

#### **10. Next meetings**

The ICO undertook to poll members to determine if Tuesdays are still suitable and put together a list of proposed dates for next year’s meetings.