

MINUTES

Gene/Disease Specific Database Advisory Council
Teleconference
Tuesday 7 February, 2017
1300hrs UTC/GMT

MEMBERS

Present

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

ICO Staff

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

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
Raymond Dagleish
Johan T den Dunnen

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

ACTION SUMMARY

Section	Action	Responsible
4	Prepare 2017 Workplan discussion paper	Timothy Smith
5c	Share Draft Working Group output with Council	Mauno Vihinen

MINUTES

1. Welcome, Apologies, and Administrative Matters

2. Confirmation of minutes of previous meeting

Minutes were confirmed unanimously.

3. Roadmap Metrics

The Council noted the Roadmap Metrics.

4. 2017 Workplan – Timothy Smith

The Council members discussed what they hoped to achieve in 2017 against the *Project Roadmap 2016-2020*.

Timothy Smith reported on the results of the survey of members undertaken at the end of last year. He noted that three themes of activities emerged:

1. Defining best practices
2. Skills development
3. Engagement with other Project components

Timothy Smith undertook to summarize and expand on the suggestions raised in this meeting in a discussion paper that will be shared with all Council members.

5. Defining Best Practices

a. Joint Working Group: Minimum Content Requirements – Peter Taschner & Martina Witsch-Baumgartner

Peter Taschner reported that the two Working Groups have been combined and are currently reviewing the previously undertaken work. Peter Taschner further reported that Germany has decided that proper reporting is required before reimbursement will be made for diagnostic tests, so all the clinical genetics centres are actively looking at how to store the data that they generated. He noted that this will inevitably lead to requests for more suitable databases and tools, and that this could be potential source of sustainable funds for the development and maintenance of these tools. The Netherlands have also decided to develop a national database, and will eventually share this data worldwide, but the blocking issue is concerns over sharing interpretations opening them up to legal liability if the data is used incorrectly. They are currently only doing so for BRCA1 and 2, through the LOVD 3.0 Shared Installation.

b. WG08: Ethics Checklist for LSDB Curators and Submitters – Rosemary Ekong

Rosemary Ekong reported that a new draft has been prepared incorporating the feedback generated during the public consultation phase and will be sent to the other Working Group members today so that a Draft for Approval can be prepared.

c. WG09: Database Quality Measures

Mauno Vihinen reported on the work of the Database Quality Measures Working Group that is being overseen by the ISAC. He reported that this is an extension of the previously published HVP Guideline on Database Quality Criteria. This Working Group is focused on practical implementation of a quality accreditation scheme. The Working Group has assembled an almost final list of measures and are planning to move to an alpha testing phase soon, where a number of databases are manually scored. He undertook to share the draft document with the Council.

6. Reports from Database Curators

Arleen Auerbach reported that her team is now responsible for curating 22 Fanconi Anemia genes on LOVD.

Peter Taschner reported on two recent publications on somatic variants. One is regarding synonymous variations that are observed in cancer. This is [available now in Human Mutation Early View](#). The other paper is a review paper that discusses requirements for reporting somatic variants, specifically making a clear distinction between germline variants observed in tumors and true somatic variants.

7. Other Business

a. Sharing data between LOVD and ClinVar

The Council members discussed how data is planned to be shared between ClinVar and LOVD and noted that there is still no automated means to do so. They further noted that the ClinVar team are no longer able to provide the same level of support to LOVD curators wishing to make their data available in ClinVar, although it is still possible for individual curators to submit their own data. Peter Taschner reported that the current LOVD development focus is centered on migrating version 2 databases to version 3. Timothy Smith reported that on the last ISAC call, Johan den Dunnen undertook to investigate the feasibility of providing downloadable summary data files of variant interpretations, which would enable all data in LOVD databases flagged as being available for download to be bulk imported into ClinVar and any other database or pipeline.

8. Future Meeting Dates

- 4 April – Teleconference
- 6 June – Teleconference

- 1 August – Teleconference
- 3 October – Teleconference
- 5 December – Teleconference