

MINUTES

Gene/Disease Specific Database Advisory Council

Teleconference
Tuesday 6th June 2017
1200hrs UTC/GMT



MEMBERS

Present

Peter E M Taschner (Chair)
Arleen Auerbach
Nenad Blau
Mary Fujiwara
Daniel Hampshire
Finlay Macrae
Magali Olivier
Yves Sabbagh
Mauno Vihinen

Amy McAllister
Helen Robinson

Apologies

Olubunmi K D Abel
Ammar Al-Chalabi
Stefan Aretz
Timothy Barret
David Baux
Jean-Pierre Bayley
Daniel Bichet
Nancy Braverman
Paola Carrera
Raymond Dagleish
Johan T den Dunnen

Rosemary Ekong
Pascal Escher
Almudena Fernández
Marc Ferre
Bruce Gottlieb
Tamas Hegedus
Raoul Hennekam
Alex Hewitt
Ammar Husami
Sarah E A Leigh
Derek Lim
Eamonn Maher
Lluis Montoliu
Etienne Mornet
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

AGENDA

1. Welcome - Apologies, and Administrative Matters

2. Confirmation of record of previous meeting – Minutes confirmed as correct

3. Operational Issues

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

Peter Taschner reported that he and Martina met with a group at the ESHG in Copenhagen to discuss some of the details regarding this topic. The results of this discussion are in the process of being used to draft a meaningful document, and as there has been little time between those discussions and this call, unfortunately, it was not yet available. There is still work to be done on simplifying the document and integrating material that is currently in appendices. The draft will include examples drawn from both LOVD2 and LOVD3 and the aim is to circulate a good draft to members for comment before the northern summer starts.

b. WG08: Ethics Checklist for LSDB Curators and Submitters – Up-date on comments received from circulation of final draft - Rosemary Ekong

Rosemary Ekong was not able to join the call but had advised that she has received the helpful comments and feedback on the final draft. The comments were very encouraging but a few suggestions were more substantive so she will refer those back to the working group. Once this work has been done, a new version will come back to Council for review. It is expected that this can be done quite quickly so that can be put on the agenda for a decision at the next meeting.

Finlay Macrae also reported that InSiGHT, through its Governance Committee, has developed an Ethics and Privacy Policy based on the *Framework for Ethical and Privacy Principles in relation to Responsible Sharing of Genomic and Health--Related Data*. This document was based on work done in conjunction with Global Alliance's (GA4HG) Ethics Working group, of which HVP is a contributing member (through Helen Robinson – refer to: <http://genomicsandhealth.org/working-groups/regulatory-and-ethics-working-group> for more information), and has been tailored to fit the work of InSiGHT. Fin and Helen agree that this work by InSiGHT on the Framework might be of interest to both Rosemary's group and other members of the Council who are working on these important issues. The InSiGHT document is being passed through the InSiGHT Database Governance Committee and then to Council for approval. Anyone wanting more information should contact Fin or Helen directly.

4. Reports from Database Curators

Finlay Macrae reports that the InSiGHT database operations continue to evolve not only with ethics and privacy mentioned above, but with their operational procedures (SOPs). As they deal with clinical interpretation and assignment of pathogenicity they are anxious to avoid duplication of effort but also have consistent outcomes. He reports that they have good communication with ClinGen who has helped by developing an interface software tool loosely based on the guidelines of the American College. Also InSiGHT continues with its involvement with the Global Alliance and the BRCA Challenge for the development of some kind of mutually beneficial frontend interface to LOVD.

Several other members reported on their interactions with ClinGen and ClinVar in the recent past. Helen reported on the recent very positive experience with GG2020 when they were invited by ClinGen to form a global expert panel for haemoglobinopathies. While the process had been time consuming, it had been a very professional process with much assistance and support. It was suggested that the advice on how to best to interact with them should be sought at the next ISAC meeting, given the representation on that Committee. Peter and Helen will ensure that the matter is raised there.

5. Other Business

- i. LOVD team has come up with a method to present and view variant data uploaded so far using a country view. This was recently demonstrated by Johan and had been sent to members of the Country Node Council. It will be circulated to members of this Council as well – look for separate email. Members are encouraged to view their own country page as well as those of other countries as it is surprising to see the many different individuals/ organizations are currently providing the data. The data provider information is kept, and can be displayed. There is often a much wider set of data presented than would otherwise be expected. This led to a discussion of the links between members of this Council and the activities of Country Nodes, and how they could work together in a more systematic way. It was noted that the current HVP Roadmap for 2016-2020 refers to the need to increase the amount and quality of variant information that is shared in curated databases, and highlights that one way of addressing this is by encouraging members of this council to work more closely with the country node network, and look for opportunities for sharing good variant information.
- ii. Helen reported that she was a member of a panel at a community genetics workshop at ESHG focusing on population diversity in genomics and variant data collection. Her presentation was based on the learnings from GG2020 where it was increasingly clear that there was a need to ensure that all sub-populations, minority communities, and different ethnic groups are included in genomic analyses. She asked members to consider how they might address this in their current activities as it was an issue that is likely to be of increasing concern to those managing locus specific databases as the demands for information relevant to population background is often needed for interpretation.

6. Future Meeting Dates for 2017

Members agreed to *cancel* the meeting of 1 August 2017 as it would coincide with the northern summer holidays. It was suggested that rather than October and December calls it would be better to have calls in September and November.

The following dates are suggested:

- i. Tuesday 19 September
- ii. Tuesday 21 November