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MINUTES

Gene Disease Specific Database Advisory Council

16th September, 2014

9.00 PM Melbourne, Australia time, 1.00 PM Leiden, Netherlands time

by Calliflower

ATTENDEES COUNCIL MEMBERS

Arleen Auerbach	Finlay Macrae
Nenad Blau	Peter EM Taschner (Chair)
Raymond Dagleish	Mauno Vihinen
Sarah E A Leigh	

ATTENDEES NON COUNCIL MEMBERS

Richard Cotton
Rania Horaitis
Heather Howard

AGENDA:

1. Welcome

Meeting opened 9.05 PM

2. Apologies

Paola Carrera, Marc Ferre, Rosemary Ekong, Pascal Escher, Daniel Hampshire, Yves Sabbagh, Bing Yu

3. Welcome new Members

No new members to welcome.

4. Confirmation of minutes of previous meeting – 16th September, 2014

Minutes confirmed.

5. Matters arising from the previous meeting

5.1 Activity proposal for ethics checklist for LSDBs to be made

Rosemary Ekong reported that she had an initial meeting with Mauno Vihinen and Sarah Schlesinger who agreed to be members of this committee. She would like to see what LSDB curators have implemented so far. For many LSDB curators, they probably have been unable to check if informed consent has been achieved. This can be elaborated on in database policies. There is also an unmentioned 4th member who has expressed an interest in joining this committee. The council will have to wait for further developments.

5.2 Merging existing lists of LSDBs

LOVD List, HGVS list, Gen2Phen list. There is an initial understanding about sharing information on the lists with the ClinVar databases. There will be an effort to combine all the information available. In principle a single list should be available from different websites. This will be discussed during the HGVS Board meeting and a decision should be made of what to do. Heidi Rehm (ClinVar) and Anthony Brookes (Gen2Phen) as well as Peter Taschner (LOVD) will also be at the meeting so between them a consensus should be reached about what to do with the lists.

Finlay Macrae stated that InSiGHT has an interest in genes related to inherited colorectal cancer with a particular interest in MMR genes. InSiGHT would like to identify on LOVD genes that InSiGHT are interested in but they are not necessarily ready to curate these genes yet. Peter Taschner stated that this should be possible.

5.3 Increase awareness of journals to nomenclature and data sharing

Peter Taschner had promised to send suggested letters to Editors. A draft was sent to Raymond Dagleish for his comments. Raymond Dagleish stated that he had been in touch with the journal *Clinical Genetics* because they had very vague guidelines of how data description should be given in their journal. On the 2nd Sept Michael Hayden the Editor in Chief wrote and said "Thank you for your input on the genome variant sequence description guidelines, we will be including a slightly modified version on the *Clinical Genetics* website soon. This will certainly be a good resource for authors who are preparing their submissions." This refers to guidelines put together based upon the guidelines that *Human Mutation* uses and, with the approval of Mark Paalman, they were modified and extended. This went to *Clinical Genetics*, and they will use them as instructions to authors. This could be a set of guidelines that could go out to all journals. If not already circulated it should go around to the Council for any additional comments. Raymond Dagleish would then be happy to have a look at any such comments.

5.4 *Human Genome Variation* journal's new database

Johan den Dunnen had objected to the setting up of a new database. Peter Taschner also sent a mail to the Editor but had no response. All members in contact with the Editorial board should at least ask what their plans are and mention there is no need for additional databases as it would only increase any confusion that exists.

Mauno Vihinen stated that he is on the Editorial Board and Macmillan is the publisher, and they wish to have the data from papers published in the journal in their database so that the data is under their control. They wish to have a much more extensive database so there will be an additional database. The data will be freely available.

Sarah Leigh asked if they will be offering micro-attributions.

Mauno Vihinen stated that all data published in that journal will go into the database, but that they will not be collecting data from elsewhere.

6. Report from Chair

Peter Taschner reported that he has been in contact with several databases that have agreed to move their data to LOVD. There are still some people setting up databases in Excel and they were surprised to hear that there was a database system available. So it will not be very difficult to move to a real database system.

7. Gene/Disease Specific Database Activities

Brief report from Council members – activities & plans (*2 minutes each only if they wish*)

Rosemary Ekong reported 500 new entries in the last four months for the TSC 1 and 2 databases.

8. Updates from working groups

8.1 Copyright & Disclaimer Statements on LSDB websites

The period for comment is now over. When this Council agrees that this is ready to go, it is to be sent to the ISAC as a recommendation. This document should be ready for the next meeting of this Council. John-Paul Plazzer who is Chair of this group is to be asked to have the document ready.

8.2 Variant Database Quality Assessment Group

This Council has accepted the proposal. It is with the ISAC but it has not yet been accepted, but it will be discussed this week at the ISAC meeting. If moved ahead, it will become official, and also the 1st HVP Standard to be adapted. It is published on the HVP Website and sent to all the members. There will also be a publication.

9. Recommendations to the Scientific Advisory Council

No items to recommend except to make a decision on the recommendation of Variant Database Quality Assessment Group.

10. Other matters

Arleen Auerbach asked about the BRCA challenge, and what is happening in regards to merging the various databases.

Peter Taschner stated that this initiative is being led for the HVP by John Burn together with the Global Alliance. The proposal to merge all the databases using the LOVD system is still standing but it is unknown who will take the lead in this and if the Global Alliance agreed. Some of the participants involved have already agreed.

Heather Howard stated that there will be a meeting of Global Alliance and the BRCA Challenge at ASHG from the BRCA perspective. The HVP will be putting up a funding proposal for funding from an unknown source just for the HVP component and this will be for the integration of the BRCA information through the Country Node system, also some funding to assist with the interpretation committee and LOVD.

Regarding the Globin Initiative; Helen Robinson is working on that particular proposal. HVP is seeking funding for that project as well. Helen Robinson should be able to give a further update on the Globin Initiative at the ISAC meeting.

11. Next Meetings

- November 18th

Meeting Adjourned 9.57 PM