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MINUTES

Gene Disease Specific Database Advisory Council

18th November, 2014

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

by Calliflower

ATTENDEES - COUNCIL MEMBERS

| | |
|------------------|---------------------------|
| Arleen Auerbach | Sarah E A Leigh |
| Raymond Dalglish | Finlay Macrae |
| Rosemary Ekong | Magali Olivier |
| Pascal Escher | Peter EM Taschner (Chair) |
| Mary Fujiwara | Mauno Vihinen |

ATTENDEES - NON COUNCIL MEMBERS

Richard Cotton
Rania Horaitis
Heather Howard

AGENDA:

1. Welcome

Meeting opened 9.05 PM

2. Apologies

Daniel Hampshire, Tamas Hegedus, Sue Povey

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 a progress report and work schedule was sent to the sponsoring Council. The membership comprises of Rosemary Ekong, Mauno Vihinen and Sarah Schlesinger. Gerado Jimenez from San Jose in Costa Rica has also expressed an interest to join the Working Group but it is not yet confirmed. The group was going to assess compliance according to the Povey *et al* paper and this will be done as a survey. The survey has not yet been designed or sent out yet but it is expected to go out before Christmas. Unfortunately Sarah Schlesinger has been unresponsive so Dr Ekong is concerned whether the Working Group can proceed with only two active members. Arleen Auerbach who knows Dr Schlesinger personally volunteered to find out what is happening. Peter Taschner suggested that a member of the Global Alliance Ethics committee involved with the BRCA Challenge might be interested in joining this Working Group. There was a call for members to join in the last HVP Bulletin. Richard Cotton suggested that an ethicist from UNESCO join the group. Heather Howard reported that Helen Robinson from the HVPI coordinating Office is in touch with an ethicist there so hopefully she is able join. There is also a possibility that an ethicist from the World Health Organisation may be able to join the working group too. Peter Taschner suggested that he asks Anne Cambon-Thompson who has been involved before via the Gen2Phen project if she would like to join the Working Group and help set up the survey. Finlay Macrae reported that InSiGHT has an already established ethics committee so maybe one or more of those people could join the Working Group and he suggests Julian Savaleiscu.

5.2 Merging existing lists of LSDBs

LOVD List, HGVS list, Gen2Phen list. Peter Taschner reported that unfortunately the merging of the three lists has not yet been accomplished. There was an agreement that there should be one list but there has not been a real decision yet of what that list should be. There is however an understanding about sharing information on the LOVD lists with the ClinVar databases. There will be an effort to combine all the information available. For the time being making sure the lists are available from the different websites is what needs to be done.

Peter Taschner suggested that if there was a GDSDAC page on the HVP Website he would be happy to begin adding content to the page.

5.3 Increase awareness of journals to nomenclature and data sharing

Peter Taschner sent suggested draft letters to Editors together with the meeting Agenda. Sarah Leigh made a suggested change to the end of the existing Editor's letter. She suggested that the last sentence "One of the advantages of this policy is that links from the public databases to the original publications lead to more citations" should be extended to say "which is obviously of benefit to authors and to journals

in improving their impact factor and reputation”, because that is what Editors are really interested in; impact factors and reputation. Peter Taschner has suggested he send these letters to any journal that publishes genetic data. He suggests that there be a “name and shame” list on the HVP Website of journals following the guidelines and journals not following much the same as HGNC lists those journals that do not use the correct gene nomenclature. Heather Howard stated that the HVP ICO is happy to send out the letters to the journals.

[Subsequently, Tim Smith has produced a list of journal which indicates the compliance, or not, with HGVS nomenclature and requirement for data to be deposited into a LSDB prior to publication. The data can be viewed at <http://www.humanvariomeproject.org/resources/genetics-and-genomics-journals.html>]

Raymond Dagleish reported further about the issue with Clinical Genetics. He stated that he had been in touch with the journal *Clinical Genetics* because in one paper he saw there were many errors of reporting the variants in the collagen genes that he is familiar with. As the journal had very vague guidelines of how data description should be given in their journal he contacted Michael Hayden the Editor in Chief who agreed to include a modified version of guidelines for reporting variants on the *Clinical Genetics* website. The paper that Raymond Dagleish questioned has now been published. The errors in reporting the collagen gene variants have been fixed, however there are several variants in other genes reported in the paper that are still incorrect. This shows the journal may have changed the instructions but is not policing the submissions.

6. Report from Chair

Peter Taschner reported that he and Jean-Pierre Bayley as curators of the TCA cycle gene database (<http://www.LOVD.nl/SDH>) have announced that the database is going to use the creative commons for non-commercial share-like license (CC4-NC-SA) and they have asked the submitters of the data if they agree; so far they have only received positive answers and no objections to applying this license to the contents of the database. The license information will be included on the database website with a link to information about obtaining a commercial license. The idea is that parties interested in a commercial license are asked to share their data as well. If they agree, they may either get a free license or a reduced-rate depending on the number of variants they want to include. Any income from these license fees will be used to make sure the data are curated and the quality is increased. The curators of the TCA cycle gene database will be open about what income there is and how it is applied. They would also like to include this in the Database Policy Statement, which will be finalized following the December 1, 2014 deadline for comments and objections regarding the license. Note added 18-12-2014: No additional comments and objections have been received, so the curators have decided to proceed as described above. The submitters will be asked to agree with new Database Policy Statement, which will become effective 1 month after the announcement by mail.

7. Gene/Disease Specific Database Activities

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

Arleen Auerbach reported that the Fanconi Anaemia database (all 16 genes) has moved from LOVD2 to LOVD3 and they are very grateful for the help from Leiden to do this.

Rosemary Ekong reported that the TSC1 and 2 database is at the final stage of a commercial agreement on variant sharing. The commercial company that cannot yet be named will be giving the TSC1 and 2 database their variants.

Finlay Macrae reported that InSiGHT are collaborating with Ian Tomlinson to curate 2 new specific database relating to POLD1 and POLE.

Magali Olivier reported that their TP53 database is merging with the database maintained by Thierry Soussi and hopefully this can be completed early 2015.

8. Updates from working groups

8.1 Copyright & Disclaimer Statements on LSDB websites

Arleen made a comment about the English for the copy of the Disclaimer statement sent around with the Council papers. "*Database curators may curate data to ensure that database formatting and quality standards are met; or share submitted data with external parties for research purposes or for sharing with other databases.*" This reads poorly and should be changed to "*Database curators may curate data to ensure that database formatting and quality standards are met. They may also share submitted data with external parties for research purposes or for sharing with other databases.*"

This will be discussed at the ISAC meeting this week. This should be fixed and then sent to the ISAC. All agreed to make the change.

8.2 Variant Database Quality Assessment Group

Both this Council and the ISAC have accepted the Variant Database Quality Assessment guidelines. Peter Taschner stated that every database curator should be made aware of what the requirements are before an assessment is started. Mauno Vihinen stated that the next step is how this should be implemented. He reported that at the ISAC meeting in San Diego it was decided that Genetic Alliance should be approached to implement this as they have many people who might be interested in this type of thing so the next step is to be in touch with Global Alliance. The Working Group should write an article about these guidelines (to be started as soon as possible).

9. Recommendations to the Scientific Advisory Council

Recommend acceptance of the modified Copyright and Disclaimer Statement.

10. Other matters

No other matters.

11. Next Meetings

- 20th January 2015 – 11:00 PM Melbourne / 1:00 PM Leiden

Meeting Adjourned 11:50 PM