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MINUTES

Gene Disease Specific Database Advisory Council
23rd May – 11.30 AM
Salle IV
UNESCO, 7 place de Fontenoy, Paris, France

ATTENDEES: COUNCIL MEMBERS

Arleen Auerbach	Sarah E A Leigh
Raymond Dalgleish (Co-Chair)	Finlay Macrae
Rosemary Ekong	Peter Taschner (Co-Chair)
Daniel Hampshire	Mauno Vihinen

ATTENDEES: NON-COUNCIL MEMBERS

Akin Abayomi	Marc Greenblatt
John Burn	Rania Horaitis
Garry Cutting	John- Paul Plazzer

AGENDA:

1. Welcome

Meeting opened – 11.30 AM

2. Apologies

Marc Ferre, Tamas Hegedus, Sue Povey.

3. Welcome new Members

No new members to welcome.

4. Confirmation of minutes of previous meeting – 1st April, 2014

All agreed, minutes accepted.

5. Matters arising from the previous meeting

5.1 Roadmap summary and role of the GDSDAC

The Roadmap summary form with commentary or information was circulated to Council before the meeting; comments and suggestions were received by Raymond Dalgleish. He wishes to particularly acknowledge the contribution of Sue Povey to this document.

It was agreed that the recommendations should be implemented and become a core part of what the Council does. If any Council member has anything to present to the Council and the ISAC they should indicate this as soon as possible, for example new technologies.

Raymond Dalgleish indicated that in discussions with Helen Robinson she indicated that the Council seems to be ignoring the ethics issues of databasing. Ethics is not just about confidentiality; it is also about behaving ethically and avoiding conflicts of interest. These issues impinge on all activities of the Human Variome Project. [Subsequent discussions between Helen and Raymond resulted in a proposal for example scenarios to be described where ethical issues might arise. These exemplar scenarios will be circulated for further discussion.]

There is a paper in *Nature Reviews Genetics* (Ehrlich and Narayanan (2014) Nat Rev Genet 15:409-421: <http://www.ncbi.nlm.nih.gov/pubmed/24805122>) about methods for circumventing patient confidentiality and methods for mitigating these methods; in the absence of identifiers for patients you have to go to a lot of effort to identify patients from data. The essence is how much information is required to identify a patient? Perhaps a statement could be drawn up that the Council and the HVP can endorse about what sort of information is safe to release to databases.

Further suggestions for the Role of the Council in the Roadmap: Finlay Macrae suggested that a cost-benefit analysis for an LSDB be made. Raymond Dalgleish suggested that this is not just specific to the Council but it should be overarching for the HVP.

5.2 Ethics and Legal Issues

Rosemary Ekong stated that in the Sue Povey ethics paper they found all the points valid but some of these are impossible to fulfill and so they (including Sue Povey) withdrew from this activity as they are

database curators and so there is a conflict of interest. So in essence, what they are advocating in the paper is in reality difficult to implement.

Raymond Dagleish suggested that a short list of aspirations in order of importance be drawn up and then curators could implement as many of these as possible in practical terms. The InSiGHT database is the best there is in terms of screening and ethics.

Peter Taschner stated that it is impossible for curators to check if ethical guidelines are followed and it is up to the submitters to follow the guidelines and indicate they did so.

Mauno Vihinen suggested to take this further and create a checklist for LSDB curators of what they have to do regarding ethics. He said he is happy to be part of a group but is not able to lead it.

Rosemary Ekong agreed that she will chair this group and work on the checklist with the help of others.

Action: Activity proposal for ethics checklist for LSDBs to be made

6. Report from Chair

Raymond Dagleish had nothing in particular to report. He thanked Rania Horaitis for her help as the Council liaison to the HVP.

Elections for a new Council Chair were held during the HVP5 meeting in Paris. All HVP consortium members were eligible to vote. It was with great pleasure that Raymond Dagleish handed over the Chair of the Council to the newly elected Chair Peter Taschner.

7. Transition to Newly Elected Chair

Peter Taschner thanked Raymond Dagleish for his work as the 1st Chair of the Council. He asked him if he would be willing to assist as Deputy-Chair, an invitation that was accepted. Peter Taschner also asked if Arleen Auerbach would be willing to participate in the leadership and she agreed.

8. Gene/Disease Specific Database Activities

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

There were no reports at this time.

9. Updates from working groups

9.1 Copyright & Disclaimer Statements on LSDB websites

John Paul Plazzer reported at the main HVP5 meeting. A draft Disclaimer Statement for Websites has been drawn up and was presented. The exposure draft is now available on the HVP website in the for comment by the wider community. This statement has been designed so that it can be edited to suit various databases. Council members are asked to make comments if they wish when the call is made.

9.2 Variant Database Quality Assessment Group

Mauno Vihinen reported this at the main HVP5 meeting. The draft Variant Database Quality Assessment document has been sent to HVP members. The comments and suggestions received in the 2-month public consultation period have been incorporated in the revised draft version that has to be approved by this council.

Action: The revised draft Variant Database Quality Assessment Group document is to be sent to all members of Council to give comments. This document needs to be scrutinized. Decision to be taken at next tele-meeting in July.

10. NGS Data to LSDBs

Peter Taschner stated that this topic will be handled and discussed in the Global Alliance Working Groups.

Rosemary Ekong asked how curators are handling data from NGS, what is the coverage and quality?

Peter Taschner stated that in many cases there is confirmation by Sanger sequencing however Rosemary Ekong stated that people are reporting that there is no verification by Sanger Sequencing as they do it twice and are confident with the depth and coverage of the NGS data.

John Burn stated that having to validate NGS data with Sanger sequencing is like shining a red lantern in front of a train! People should step away from this.

Peter Taschner stated that it is a matter of time to see what is trustworthy and what is not.

11. Merging Existing Lists of LSDBs

At present there are three lists of LSDBs, all of them good but none of them comprehensive. They are the:

- HGVS list
- LOVD list
- Gen2Phen list

It has been suggested that these three lists be merged into one definitive list where people can submit or view LSDBs. There have been issues of maintenance with the other lists and these can be avoided with one list. The Council should take steps to obtain agreement from the three organizations to merge the lists.

It was agreed by the Council that Co-Branding with every organization who had their list, and including the HVP should be on this new definitive list. The domain name "TBA" .

Arleen Auerbach asked if there would be vetting of databases on this list.

Peter Taschner stated that in the end there should be a quality assessment of each database but the first step is to merge the lists.

12. Membership Requirements of an HVP Database

It was suggested that old legacy databases that have been inactive for some time be written to. The curators should be thanked for their participation and efforts but it should be made clear that they are no longer active. The legacy data should then be incorporated into a new LSDB. After approach, and if there is no response then absorbing that data can proceed. Rosemary Ekong stated that this is what they did for the TSC databases.

13. Recommendations to the Scientific Advisory Council

Recommending to absorb legacy data into new LSDBs once attempt to contact curators has been made.

14. Other items

14.1 Orchid ID

Peter Taschner asked who has an ORCID ID (Open Researcher and Contributor ID, <http://orcid.org/>); half of the meeting attendees answered yes. LOVD is asking all submitters to obtain an ORCID ID. This will allow easy identification of submitters and can be used to authenticate submissions; it will also make it easier to contact them when they move.

14.2 Increase awareness of this Council

Peter Taschner suggested that awareness of this Council should be made to other groups; the Council should seek endorsement from other groups such as HUGO and the HGVS. International genetics communities should be asked to become curators.

All agreed.

14.3 Increase awareness of journals to nomenclature and data sharing

Peter Taschner suggested that the Council make sure the journals are using the correct HGVS nomenclature and that they request data be submitted to databases as a condition of publication to authors. The HVP has already begun this step by hosting a Genetics journal Editors' meeting during the annual ASHG meeting where these and other issues have been discussed.

Peter Taschner volunteered to send letters to all journal Editors making these requests and invited all council members to follow his example, especially when asked for new submissions or when authors fail to provide sufficient information/evidence to include in gene variant databases. Example letters will be provided. He stated that the EJHG had agreed and is paying editorial support checking manuscripts and helping authors to fulfill the requirements. If authors do not meet these requirements their paper will not be published.

Finlay Macrae suggested that a list of the least compliant journals be made against the number of citations as a guide to entice them to comply.

14.4 Endorsement of Courses

Peter Taschner asked if the Council could endorse the LSDB Curation Course and the Nomenclature Courses.

All agreed this was a good idea and these courses could be part of the educational component of the HVP.

15. Next Meetings

It was agreed that bimonthly meetings should continue as previously.

Next meeting will be Tuesday 29th July 2014.

Meeting Adjourned 1.00 PM