THE HUMAN VARIOME PROJECT

sharing data · reducing disease

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MINUTES

Gene/Disease Specific Database Advisory Council
Tuesday 15 March, 2016
1200-1300hrs UTC/GMT

MEMBERS

Present
Peter E M Taschner (Chair)
Arleen Auerbach
Raymond Dalgleish
Daniel Hampshire
Tamas Hegedus
Magali Olivier

ICO Staff
Helen Robinson

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
Johan T den Dunnen
Rosemary Ekong
Pascal Escher
Marc Ferre
Mary Fujiwara
Bruce Gottlieb
Raoul Hennekam
Alex Hewitt
Ammar Husami
Derek Lim
Sarah E A Leigh
Finlay Macrae
Eamonn Maher
Etienne Mornet
Sue M Povey
Yves Sabbagh
Judith Anne Savige
Sarah Sim
Carli Tops
Ronald Trent
Mauno Vihinen
Richard van Wijk
Katarzyna Wertheim-Tysarowska
Tom Winder
Bing Yu
Martina Witsch-Baumgartner
Johannes Zschocke

AGENDA

1. **Welcome**

2. **Apologies**

   Apologies were noted as above.

3. **Confirmation of minutes of previous meeting**

   The minutes of the previous meeting were confirmed.

4. **Report from Chair – Peter Taschner**

   Peter Taschner reported that on DNA Day, he ran a "Check the Experts" event that involved a group of 25 students who checked that HGVS nomenclature had been correctly used and data submitted to public databases in 130 different papers published in January editions of several journals. The results will be presented at HVP6.

One of the findings from this event that there was a high number of papers that specifically mention that variants were found during specific studies but that didn't provide any details of what these variants were.

The Council discussed how this situation could be improved. One idea was for the Council, in collaboration with the International Confederation of Countries Advisory Council, or the delegates at HVP6, to issue a statement after HVP6 calling on Human Variome Project Members to ensure that over the next 12 months these issues are adequately addressed in their own papers, the papers that they review and the instructions to authors issued by the journals that they are involved with. The Council also discussed the possibility of asking the International Confederation of Countries Advisory Council to write to the national human genetics societies of the countries represented on the Council to get their support for addressing these issues. Raymond Dalgleish volunteered to write an editorial for Human Mutation repeating the points that Human Mutation and the Human Variome Project have made in the past. He noted that he would need assistance from others. Peter Taschner and Mauno Vihinen were suggested.

Magali Olivier made the point that when approaching journals to discuss this issue, it would be wise to have a broad approach and not just target journals that specifically report on human genetics. She further noted that to ensure wide-spread use of the HGVS nomenclature, a tool that converts variants in VCF files to the correct HGVS nomenclature would be extremely useful.

5. Project Roadmap 2016-2020 & Work Plans – Helen Robinson

Helen Robinson reminded the Council that a consultation on the final draft was currently underway and urged members to comment. She also noted that the Council needs to think about how it will contribute to meeting the objectives of the Roadmap over the coming period.

6. Working Group Reports

a. WG03: Minimal content for gene variant databases (LSDBs) – Peter Taschner

Peter Taschner reported that work is ongoing but a bottleneck has been reached on how to describe the ethnicity of patients. The Working Group is suggesting that this field not be included in the minimal content set as it is becoming a political and ethical issue rather than a technical one. Arleen Auerbach suggested capturing self-declared ethnicity. Helen Robinson suggested that rather than take a position, several options could be provided.

b. WG07-Minimum content of a country specific database – Peter Taschner

Peter Taschner noted that this Working Group is asking for more time. A more substantive report will be made in the future.

c. WG08: Ethics Checklist for Gene/Disease Specific Database Curators and Submitters – Rosemary Ekong

Rosemary Ekong reported via email that the Working Group is we are working on the last draft before public consultation. She further reported that have a new member has joined the Working Group: Professor Michael Parker of the Ethox Centre at the University of Oxford.

7. Gene/Disease Specific Database Activities

Magali Olivier reported that a new version of the TP53 database has been released. Previously, the database focused on germline variants, however this data is out of date. New data is being included, primarily from induced mutations. She further reported that she is working on a paper comparing somatic variant data from Sanger and NGS.

8. Recommendations to the Scientific Advisory Committee

Peter Tacschner raised the need for a formal specification of the HGVS nomenclature in a format that is useable by computers to enable the creation of automated tools. One issue preventing this from happening is that the nomenclature, as currently specified, includes ambiguities that have not yet been resolved. He noted that a continued lack of usable tools will lead to NGS tool developers coming up with their own solutions. Any work in this area will have to be inclusive of these groups and a dialogue should be established. Members of the Council provided suggestions of

who could be approached: the Broad and Sanger Institutes, the Global Alliance. Raymond Dalgleish volunteered to talk to Fiona Cunnigham at EMBL-EBI about getting alignment between the HGVS nomenclature and GA4GH efforts.

9. Other matters

Peter Taschner noted that there will be an election for the Chair of the Council before the next meeting.

10. Next Meetings

- 3 June – Face-to-face at HVP6
- 12 July - Virtual
- 13 September - Virtual
- 8 November - Virtual