

MINUTES

Gene/Disease Specific Database Advisory Council
Tuesday 12 July, 2016
Teleconference
1200-1300hrs UTC/GMT



MEMBERS

Present

Peter E M Taschner (Chair)
Arleen Auerbach
Raymond Dagleish
Johan T den Dunnen
Rosemary Ekong
Mary Fujiwara
Tamas Hegedus
Sarah E A Leigh
Yves Sabbagh
Bing Yu

ICO Staff

Timothy D. Smith
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
Pascal Escher
Almudena Fernández
Marc Ferre
Bruce Gottlieb
Daniel Hampshire
Raoul Hennekam
Alex Hewitt

Ammar Husami
Derek Lim
Finlay Macrae
Eamonn Maher
Luis Montoliu
Etienne Mornet
Magali Olivier
Sue M Povey
Judith Anne Savige
Sarah Sim
Carli Tops
Ronald Trent
Mauno Vihinen
Richard van Wijk
Katarzyna Wertheim-Tysarowska
Tom Winder
Martina Witsch-Baumgartner
Johannes Zschocke

AGENDA

1. Welcome

Peter Taschner welcomed members to the meeting and made particular note of two new members: Luis Montoliu and Almudena Fernández, representing the Albinism Database.

2. Apologies

Apologies were noted as above.

3. Confirmation of minutes of previous meeting

Minutes of the previous meeting were confirmed.

4. Report from Chair – Peter Taschner

Peter Taschner reported that Working Group WG03 will merge with Working Group WG07 from the ICCAC as discussed at the joint meeting of the two Councils in Paris in June. He further reported that the project being undertaken by one of his students of checking papers from journals that include a requirement to use correct nomenclature and submit data in their author instructions to make sure that these requirements were being complied with, was almost complete. Preliminary results from this project were reported at HVP6. He committed to running a similar project next year. He noted that the action discussed at the Join Council meeting about a comparison table of available tools would also make a suitable student project. Members wishing to provide this opportunity to a student should contact Timothy Smith (tim@variome.org).

5. Contacting Human Genetics Societies regarding use of HGVS Nomenclature and data submission – Timothy D. Smith

Timothy Smith reminded the Council that this matter had first been raised during the May meeting of the Council. The Council discussed the matter further and resolved to produce a template letter asking national societies and professional organisations with an interest in the accurate reporting of variants to take action. They further resolved that the ICCAC should be asked to participate in this activity.

The template letter would be available to be used as a base for Council members to write as an individual, on their own or their institution's letterhead, to the relevant societies in their country. The letter will cover issues including:

- Requiring the use of HGVS nomenclature in publications and clinical reports;
- Encouraging the submission of variant data to public databases;
- Becoming a contact person for the Society back to the Human Variome Project.

The base letter will be provided in English, and members are encouraged to produce versions in their own languages. The letter will also include a version of the Human Variome Project logo denoting that the authors are representatives of Human Variome Project Data Provider Members. Copies of any letters sent should be forwarded to the ICO as a record.

The ICO undertook to prepare a draft letter in time for members to comment prior to the matter being included on the agenda for the 2nd of August meeting of the ICCAC. Comments should be sent to Timothy Smith by 25 July.

6. Gene/Disease Specific Database Activities – Brief Reports from Members

Johan den Dunnen asked members to put him in touch with any colleagues in China that might be interested in an exchange program that is operating between the Netherlands and China.

He further reported that his team is working on improving the exchange of data between ClinVar and LOVD. Data exchange is taking place from the LOVD shared installation. If curators of other LOVD installations would like to be included, they should contact Johan. The solution being developed will only be available for LOVD3, but he reminded members that his team is able to provide support for a migration from LOVD2 to LOVD3. He further mentioned that the dbSNP export button in LOVD2 might be able to be used to share data with ClinVar. He further reported that a Country specific view for LOVD has been developed by his team for the Argentinean Node and translated into Spanish by the Node. Finally, he has started a collaboration with the supplier of MLP kits in the Netherlands to include information on how to properly describe the results using the HGVS nomenclature.

7. Recommendations to the Scientific Advisory Committee

No recommendations were made.

8. Other Matters

Johan den Dunnen asked members to put him in touch with any colleagues in China that might be interested in an exchange program to work on the LOVD gene variant databases that is operating between the Netherlands and China. He contacted Ming Qi but he did not yet reply.

Johan reported that his team is working on improving the exchange of data between ClinVar and LOVD. Data exchange is taking place from the LOVD shared installation. If curators of other LOVD installations would like to be included, they should contact Johan. The solution being developed will only be available for LOVD3, but he reminded members that his team is able to provide support for a migration from LOVD2 to LOVD3. He further mentioned that the dbSNP export button in LOVD2 might be able to be used to share data with ClinVar.

Johan reported that a Country specific view for LOVD has been developed by his team for the Argentinean Node and translated into Spanish by the Node (current url <http://courses.lovd.nl/countrynode/argentina/>).

Johan stated he has started a collaboration with the supplier of MLPA kits in the Netherlands to include information on how to properly describe the results of the deletions/duplications detected using correct HGVS nomenclature.

9. **Next Meetings**

- 13 September - Virtual
- 8 November – Virtual