



sharing data · reducing disease

MINUTES

Interim Scientific Advisory Committee

12th October, Montreal Convention Centre, Room 513B

Attendees:

David Rimoin (Chair)
Arleen Auerbach
Mona El Ruby
Mireille Claustres
Johan den Dunnen
Mauno Vihinen
Garry Cutting

Gert-Jan van Ommen
John Burn
Richard Cotton (Scientific Director)
Heather Howard (Operations Manager)
Tim Smith (Communications Officer)
Rania Horaitis (Scribe)

Apologies:

Finlay Macrae
Marc Greenblatt
Yoichi Matsubara
Ming Qi (Head China Node Partnership)
Christine van Broeckhoven
Aida Falcon Vargas

Meeting Opened: 12.45pm

1. Introductions

2. Relationship with HGVS

Arleen related that the letter from HVP Board to the HGVS Board was discussed at the HGVS Board Meeting point by point at their recent Board Meeting and only a few minor points were suggested to be changed. The first item was would they continue as a Society in some form and they voted to continue. The HGVS Board indicated they would respond to the HVP Board in writing by the end of the month.

3. The impact of exome capture

The HVP must consider the complex new development of genomic/phenomic exome capture and how will this be integrated into the HVP infrastructure, as whole exome sequencing will be the norm now that it is getting cheaper. John Burn proposed that HVP develop a repository of all exomic data. If we

can interest our Chinese colleagues in taking on this sort of project it would be good as it would act as a clearing house. It will be also important to HVP to know what doesn't cause disease.

4. Standards Development Process

Tim Smith briefly outlined the Standards Development Process Policy Document. The document was circulated prior to the meeting and there were no changes required. This document is now recommended by this committee.

It is noted that there is a mechanism for an existing *de-facto* standard to be recommended and used as an interim HVP standard until the HVP process is completed. The Coordinating Office confirmed that there will be administrative assistance for the creation of standards and the website is currently being redesigned with this in mind.

5. HVP Roadmap

The first round of consortium comments has now closed and these will be distilled into the next version of the Roadmap and sent around for further comment. The final version will be agreed prior to the HVP Biennial Meeting in June, 2012.

6. Relationship with EC, NHGRI, Registry initiatives

Richard Cotton described the relationships: the EC has been interested for many years and has put funds towards the work via Gen2Phen. A grant has been put in for Coordination with Ségolène Aymé in the FP7 round. NHGRI and mutaDatabase and Heidi Reim are also involved in a NIH grant that HVP supports and Johan is listed as an advisor. HVP should work with IRDiRC to assist with global registries.

7. Mechanisms for assisting databases

Heather Howard outlined the Coordinating Office Position Paper *Mechanism for the establishment of new gene/disease specific databases under the Country Node and Cooperation Agreement with China* and advised that Ming Qi has commented on the content and had made the request that a Chinese representative be offered a seat on the committee. This was agreed unanimously.

Garry Cutting proposed that an independent committee of ethics representatives be appointed to provide advice if any unforeseen situations arises, particularly those arising from individual family situations. Seconded John Burn. All agreed.

Gert Jan van Ommen proposes that the HUGO group be approached to take on this task. Seconded John Burn. All agreed.

8. Discussion of Beijing Program

The Speakers were discussed and the program will be revised to include:

- Talks on whole genome sequencing from Hilger Ropers.

- A representative from Gen2Phen should be asked to speak as should someone from mutaDatabase in the global collection spot.

It was noted that Jean Jacques Cassiman would unfortunately not be attending the meeting.

9. Other matters

Dick Cotton proposed that Richard Gibbs be seconded onto the committee All agreed.

Dick Cotton asked the Committee to think of ways that we can work with both UNESCO and WHO.

John Burn suggested consanguinity as an area the HVP could have an effect especially on how to minimize consequences rather than stopping them.

Tim Smith reported that the HVP Consortium has reached 405 members.

Meeting adjourned: 14.30