

**Minutes of the Business Meeting of the
Human Genome Variation Society, Inc.**

23rd October 2007

4.35 PM – 6.23 PM

Room 29AB, San Diego Convention Centre

San Diego, CA, USA

The meeting was opened by Prof. Richard Cotton, President.

Members Present:

Anderson Paal Skytt
Andreson Brage Storstein
Auerbach Arleen
Beroud Christophe
Brown Alastair
Cotton Richard G.H
Cox Diane
Den Dunnen Johan T.
Giampietro Philip
Giardine Belinda
Gottlieb Bruce
Hadjisavvas Andreas
Horaitis Rania
Minoshima Shinsei
Oetting William
Paalman Mark
Povey Sue
Talbot Jr. Conover
Taylor Graham
Vihinen Mauno

Non-Members Present:

Barend Mons
Bean Colette
Maglott Donna
Mandel Jean Louis

1. Approval of Agenda – Presented by R. Cotton

No additions to agenda.

2. Minutes of the previous meeting for approval

No changes to minutes.

Motion to accept minutes of 2006

Minutes passed with no dissention.

3. Matters arising from the previous meeting

(a) Incentives to join the Society

(i) Decoupling *Human Mutation* from the Membership Dues

Discussion of possible incentives to entice new members to join the Society was made. Echoing comments also made at the Board meeting Alastair Brown suggested the Society decouple membership from journal, i.e. make *Human Mutation* subscription non-mandatory to joining the Society, thus dues could be reduced to US\$50 without the journal as the high dues hinder people from either joining or renewing their memberships. It was perceived that most people already receive *Human Mutation* via their libraries so do not need a subscription. A count was made of the people in the room who did not receive *Human Mutation* at their institute's library and only three people out of 23 did not receive it.

Arleen Auerbach asked Mark Paalman from Wiley if decoupling would have an effect on the Society's relationship with Wiley and if *Human Mutation* would still be the Official journal of HGVS. He said it was not essential for mandatory subscription to the journal for our relationship.

Richard Cotton pointed out the implications of this for people who do wish to subscribe to the journal; they will still be able to receive the journal at a reduced rate as members of the Society however it will cost more than what they pay now to the Society. Colette Bean representing Wiley said decoupling would affect the price the journal is sold to the Society and that Wiley would re-price it for the Society.

Motion to decouple subscription of *Human Mutation* from the Society dues.

Moved by Conover Talbot Jr.

2nd by Alastair Brown

Motion passed with no dissention

(ii) Revised Dues

Motion to make membership dues of the Society US\$50 for all

Moved by Alastair Brown

2nd by Conover Talbot Jr.

Motion passed with no dissention

(iii) Benefits of Membership

Mauno Vihinen initiated the discussion that Membership of the Society should include benefits as that is the reason to be a member. He said that if members have the same benefits as non-members why would people join. He suggested Membership dues are coupled to meeting participation. Rania Horaitis described the meeting discount to members of around US\$50 per meeting which is a substantial discount!

Bill Oetting proceeded to read benefits from a current HGVS flyer, they are:

- Subscription to *Human Mutation*
- Reduced rates for Meeting Registrations
- Voting Privileges
- Possibility to Serve on Board or as an Officer
- Receive Newsletter
- Serve on Committees
- Representation in Policy Making

Christophe Beroud suggested a good idea to increase membership of the Society is that if a non-member attends a meeting they automatically become a member of the Society by paying the higher registration fee.

Rania Horaitis pointed out that the higher registration fee for meetings for non-members at present offsets the *below cost* registration fee for members. By making non-members members in this way, yes the Society can still pay the cost of the meeting but the Society will be making no net income from dues unless some people join but never attend meetings. Alternatively, meeting registration fees could be increased across the board to counteract this effect, however it is not preferred as meeting fees could become too high.

Graham Taylor suggested incentives such as partnerships with other societies, possibly discounts with other society meetings should be investigated.

Motion to give every non-member meeting registrant a complementary membership to the Society for one year.

Moved by Christophe Beroud

2nd by Mauno Vihinen

Passed with no dissention.

(b) The HGVS should increase its presence amongst National Societies in Europe

To this end, Johan den Dunnen organised a session at the European Society of Human Genetics (ESHG) meeting in Nice this year with a focus on mutation databases and nomenclature. There was an attendance of around 250-300 people. He stated that in Europe a lot of people especially from Eastern European countries attend meetings to learn and so are very interested in the topics the Society offers, so the attendance was great. He suggests that the Society can achieve such a good attendance in Europe again.

A meeting will be held in Europe again in 2008, May 31, in Barcelona, Spain as a satellite to the ESHG. The theme of “Genotype, Phenotype and Databases” was suggested and Anthony Brookes was suggested as the Chair, as he was not present in San Diego he is to be asked.

(c) The Society should raise its profile

Graham Taylor suggested that more acknowledgment of the HGVS mutation nomenclature should be given as this is a unique selling point for the Society. Richard Cotton asked how we are going to do this. Conover Talbot Jr. suggested we encourage all the journals to make it a requirement of publication. Richard Cotton agreed that this will raise the profile and that the Society should write to the journals.

Johan den Dunnen said that when he sees a very strange mutation nomenclature he writes to the journal to inform them and they often do not realise there are recommendations!

Donna Maglott stated that NCBI Gene Reviews are now sending out requests that variants be named by HGVS standards.

Raising the profile alternatively to nomenclature, Sue Povey stated that there is an entry record for each gene on the gene nomenclature pages and each has a link to the HGVS Locus Specific Databases, and that is also a help to raise the profile of the Society.

Bruce Gottlieb stated that the Society must make more of an impact within the clinical field.

(d) Develop collaboration with HuGENet

Richard Cotton exchanged letters with HuGENet. That is all that has been done so far.

4. President’s Report

Richard Cotton summarised the past year. He stated that the Pharmacogenomics focussed meeting in Montreal was very successful. He welcomed Garry Cutting as the new Co-Editor of *Human Mutation*.

Regarding the Human Variome Project (HVP), he stated that the colon cancer group Insight, had dramatically entered the scene and desperately want to fix up their own flow of phenotype to databases. They are passionate about this and are collaborating with both the HGVS and the HVP to try to set up a system of flow to be used by other genes in other countries.

The *Nature Genetics* April issue published an Editorial by Myles Axton, the HVP Recommendations and another related paper on the HVP. Myles Axton was thanked for his encouragement and help on the HVP.

The Mutation Detection Workshop in Xiamen, China had a very good attendance of about 180 people. There are plenty of people in China discovering mutations. One interesting note is that in China, Real Time PCR spread around the country because of the SARS epidemic! The inventiveness of Chinese is fantastic so it was a great meeting overall.

SNP Workshop in Barcelona organised by Anthony Brookes was very well attended and heavily emphasis SNPs.

The HGVS has recently generated some recommendations:

- A structured simple form for ordering genetic tests is needed to ensure coupling of clinical detail (phenotype) with DNA variants (genotype) to ensure utility in publication and databases. *Hum Mutat* **28**, 931-2 (2007)
- Recommendations for Locus-Specific Databases and Their Curation. In Press due to be published in January 2008 issue of *Human Mutation*

5. Treasurer's Report

Bill Oetting described that the Society has two accounts, one in the United States to process credit cards etc, and a smaller one in Australia to be able to cash cheques and receive bank transfers.

The bank account has been slowly rising on a regular basis. An audit for both accounts will be performed at the end of the year and is to be arranged by Rania Horaitis in Melbourne. Tax reports were sent to the US tax office by Bill Oetting. He suggests we should think of what the funds should be used for.

Motion to accept Treasurer's Report

Moved by Arleen Auerbach
2nd by Christophe Beroud

6. Office Report

(i) Membership

Rania Horaitis presented the 2007 membership statistics. The main and serious problem with the Society seems to be retaining members. Although the Society does

manage to attract new members each year, they or others do not renew their memberships so the membership numbers are shrinking instead of growing.

As always, some of these non-renewing members are those who forget to renew their membership when asked and so wonder why their subscription to *Human Mutation* has stopped. This is despite many probably annoying reminders!

This year we attracted 10 new members to the Society. Suggestions of how to retain members once they join would be appreciated. It needs to be more than interesting meetings. Possibly the new dues just discussed at this meeting will change things.

Graham Taylor suggested the Society direct debit membership dues as some other Societies do. Sue Povey agreed.

Bill Oetting feels uneasy about doing this. Bruce Gottlieb stated that it is illegal in North America to do this; many, many documents and procedures need to take place to do this.

(ii) Elections

Rania Horaitis announced the Elections that will take place at the end of the year; the details are in item 8.

(iii) HGVS Meetings

Rania Horaitis described the meetings of 2007 and the planned meetings next year:

A Meeting with a Special Focus on “Pharmacogenomics” was held on 31st May 2007 in Montreal, Canada (a satellite of the main HUGO Human Genome meeting). Scientific papers were presented in a full day’s session. Bruce Gottlieb the Chair for this meeting, Alastair Brown, Conover Talbot Jr, and Christophe Beroud were thanked for all their help in the organization of this meeting.

In San Diego a Special Focus on “New DNA Sequencing Technologies” was the theme of the meeting. Johan den Dunnen, the Chair and Alastair Brown, Conover Talbot Jr, Christophe Beroud and Lauren Hardman were thanked for help in organization of this meeting.

The next meetings will be held as satellite meetings to the European Society of Human Genetics in Barcelona 31st May 2008 (Genotype to Phenotype) and the American Society of Human Genetics in Philadelphia 11th November 2008 (Clinical Genetics).

It was mentioned that in future Office Reports those people who were elected in January of that year be listed.

Johan den Dunnen commented that the HGVS does other things e.g. he himself replies to 100-200 nomenclature queries each year. There are the HGVS Newsletters, a regularly updated list of LSDBs and the HGVS website (user statistics unknown, which is a pity). These activities should be included in future reports.

7. Publications Report

Mark Paalman discussed in full the report in the handout. He described the biggest change for journal was Haig Kazazian's retirement after 15 years. Garry Cutting was taking over Co-Editorship and comes from a diagnostic background and is interested in mutations in general and complex diseases, he is a physician, scientist, clinician, and researcher and Wiley are pleased to have him on board.

The Impact Factor went down a little but that is because a couple of years ago the journal receive a big boost from the p53 issue. Even so, at 6.47 it is a very respectable IF especially with the increase in the number of journals in genetics and hereditary. *Human Mutation* remains one of the top five human genetics journals in this category. If you look at the five year trend for IF there is a consistent trend moving up significantly above the median trend for journals.

Wiley initiated email announcements to promote the pharmacogenetics symposium in Montreal, and got a couple of Special Articles from that symposium that were solicited for review and will be forthcoming in an issue soon.

The journal still averages around 650 – 700 submissions per year, the rejection rate is about 66% for all articles and for articles not invited about 71%. There were an increased numbers of invited manuscripts this last year.

The journal is receiving more association studies submissions, however a lot are not considered the type of articles *Human Mutation* should be publishing.

Mark Paalman thanked the authors, the Editors and the Reviewers for journal in the room!

Richard Cotton asked if Wiley could send a broadcast call for HGVS members through Wiley Interscience, and Mark Paalman said - why not? He suggested a membership form be attached to the PDFs, a new updated one of course.

8. Election of Council Members

Four positions on the Board of Directors of the HGVS will become vacant at December 31st, 2007. At this time, we are seeking individuals interested in sitting on the Board. All HGVS members are eligible to stand for nomination including those Board members whose term will expire; these are:

Arleen Auerbach
Alastair Brown
Bruce Gottlieb
Mauno Vihinen

Board members will serve a term of three years.

Please reply to Bill Oetting (bill@mail.ahc.umn.edu) and Rania Horaitis (rania@gdrc.hfi.unimelb.edu.au) by November 15th, 2007, to indicate your interest in being nominated, or to suggest other individuals. All nominees will be contacted by

the nominations committee to affirm their willingness to run for office. Electronic ballots will be sent out via email on December 1st, 2007 and will be due by December 31st, 2007.

Your nominations should include name, place of work, and a small paragraph describing work interests of the nominee.

9. Election of Office Bearers

A call for nominations for President-Elect will also be made at the time of Council Elections. The nomination committee will also propose and ask individuals for the election of President-Elect. Term as President will begin January 1, 2009. Please send in nominations for this very important office.

The nomination and election procedure will be the same as per the Council Elections in 8.

10. Other Business

(a) Inclusion of Non-Members in Policy Discussions

Richard Cotton asked how the Society should take into account opinions of non-members regarding any Policy generated. The idea is to circulate ideas and Policy to the whole mailing list and take any ideas from them. If a document comes from it, it would be purely agreed upon by the membership only.

Bill Oetting agreed to this as long as the HGVS maintains control of the document being written in the Society's name.

Diane Cox stated that if a non-member can still be considered in Policy, why would they become a member?

(b) Procedure of Policy Generation

The procedure of how to distribute Policy was discussed:

- It was suggested that Policy could be put in the free *Human Mutation* pages the Society has access to at the back of the journal.
- Policy could be put on the Website.
- It could be published in *Human Mutation* or another journal.

Richard Cotton favoured publishing as it gives the Policy more prominence. This was agreed by people in the room. If people put work into it, then it is good to get a publication out of it.

Any HGVS policy will also be emailed to the members as not all members will now get *Human Mutation*.

Mark Paalman reminded everyone that anyone seeing only an electronic copy of *Human Mutation* will not see the front and back pages of journal (re two free pages) so keep it in mind!

(c) Relationship of Human Variome Project and Genotype to Phenotype Project

Richard Cotton briefly presented it and it was agreed that these projects overlap and help each other. Both are projects of the members, there is no problem with it. No comments.

(d) Sharing of data between Locus Specific Databases and Central Databases

Richard Cotton circulated a document to the Board. This was initiated by fact that LSDB curators are receiving requests from browsers and central databases about putting their data on websites and people have been asking him what to do.

Johan den Dunnen has some strong ideas of what should and shouldn't be on central databases so he and Richard Cotton combined efforts and have written a document about this. This will then be sent to the membership to collect more opinions on this. In principle LSDBs should share with central databases only for the moment minimal information; the variant in the database and a link of how to get there and of course the reference sequence they use.

Diane Cox asked if each mutation be listed separately or each database listed separately with a link. Johan den Dunnen said a link of how to get to each variant to that variant in the LSDB.

Diane Cox asked what the clear advantage of a link is if someone can easily come into the database as this influences how people enter your database and can impact on funding agencies. If data is given up to central databases then there is no proof of use.

Johan den Dunnen said that as soon as there is a link there are more hits in to the LSDB.

Donna Maglott stated there are things other than the variant to increase local traffic on your LSDB.

Belinda Giardine stated that Phencode always asks permission for loading data. A summary is loaded; they don't try to load all data. This should be bringing more traffic to the LSDB as Phencode links back to the LSDB.

Bruce Gottlieb said that other issues should be looked at; legal, ethical etc. This issue should be circulated to members to receive feedback and then a formal discussion can take place.

Barend Mons agrees that hits will be better if the LSDB links to other databases e.g. Swissprot etc. Thoughts about how to make databases more formally recognised should be made.

Arleen Auerbach said the Society should formulate an opinion on reliability and accuracy of the data in LSDBs. Transcribed elsewhere there can be mistakes made and attributed to the original LSDB. It is best to have minimal data in larger databases, and the large database go back to the original source of the LSDBs to avoid errors. She said that LSDB curators are constantly updating the data and that updates may not be on the larger database. Pathogenicity for example, there may be new evidence for a variant to be pathogenic. It is better to give less rather than more and make the larger database link back to the LSDB.

Diane Cox stated that if a larger database links directly into a mutation she does not want to be responsible for a misdiagnosis because they have missed the disclaimer page!

Mauno Vihinen suggested putting a disclaimer on every mutation! Johan den Dunnen stated the LOVD database have a disclaimer on every page when they link to records.

Mauno Vihinen said additional information should to be put on databases about the validation of the variation.

Donna Maglott stated that trying to work out how many times a particular variant has been identified is good and it would be good to centralise this. Richard Cotton said that the HGVS said years ago that every instance of a variation should be reported.

Johan den Dunnen said that when information is shared, different LSDBs keep track of data differently. For example, a variant found in a family- can maybe have 20 incidences in one family –in one database counts only as 1 variant and in another database as 20 variants. Ethnicity is also very important in one database but not in another.

(e) Other business

Bruce Gottlieb formally thanked Richard Cotton to show appreciation for the work he does as he is the backbone of the organisation.

Richard Cotton suggested we have a LSDB report at future meetings so people can see what is happening. This was agreed.

Rania Horaitis thanked Conover Talbot Jr. for help in the HGVS listing of LSDBs.

Johan den Dunnen and Christophe Beroud were thanked for LSDB efforts in standardised software.

Sue Povey is to Chair a working group on ethics of LSDBs and is just getting it together. If anyone wants to join in please ask her. Arleen Auerbach stated that in the USA there are issues of what can and can't be put on the Web without specific prior consent. Sue Povey will try and have some guidelines put together. Richard Cotton said that in the past a lot of guidelines have been created from people not in the topic; ethicists and lawyers. Someone in the hot seat is needed so Sue Povey volunteering is wonderful!

In Summary

- *Human Mutation* to be decoupled from Membership Dues
- Membership Dues to be US\$50 with no journal for all
- Every non-member meeting registrant receives a complementary membership to the Society for one year
- “Genotype to Phenotype” themed meeting to be held 31 May 2008 in Barcelona, Spain as a satellite to ESHG to increase Society’s presence in Europe
- The Society should write to journal re nomenclature
- Wiley to send a broadcast call for HGVS members by attaching membership form to PDFs of articles

Meeting Adjourned 6.23 pm

Signed by Richard G.H. Cotton as a correct record:

Date
