

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

4th November 2003

Los Angeles, CA, U.S.A.

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

Members Present:

Auerbach Arleen
Beroud Christophe
Brookes Anthony
Brown Alastair
Christodoulou John
Compton John
Cotton Richard G. H.
Cox Diane
den Dunnen Johan
Gottlieb Bruce
Greenblatt Marc
Horaitis Rania
Hovig Eivind
Lehväslaiho Heikki
Oetting William
Paalman Mark
Povey Sue
Talbot Jr. Conover
Teebi Saeed
Vihinen Mauno

Non-Members Present:

Amberger Joanna
Bean Colette
Beroud Gwenaelle
Chen Jian-Min
Clancy Trevor
Ganguly Arupa
Kazazian Jr. Haig
White Peter
White Stephen
Yakubovskaya Marianna

1. Approval of Agenda – Presented by R. Cotton

No additions to agenda
Moved by Saeed Teebi

Passed with no dissention

2. Minutes of the previous meeting for approval

No changes to minutes

Motion to accept minutes of 2002
Moved by Alastair Brown

Minutes passed with no dissention

3. Matters arising from the previous meeting

(a) Tax-Free status

A 501 (c) 3 status has been applied for on November 2001 and an advanced ruling has been granted until December 31, 2005.

(b) Affiliation of HGVS with HUGO

Letters were sent to HUGO, but there has been no official response received by HGVS. R. Cotton and R. Horaitis are to pursue this matter further.

(c) Affiliation with International Federation of Human Genetics Societies (IFHGS)

The HGVS Board recommended affiliation and we should go forward with this. R. Cotton and R. Horaitis are to pursue this matter further.

4. President's Report – Presented by R. Cotton

Richard Cotton gave a small presentation regarding the goals and mission statement of the Society and how the Society must strive for coordination of efforts between groups for the common good of the community. Prof. Cotton stated:

- HGVS holds 2 yearly meetings at HUGO and ASHG meetings.
- HGVS is active in setting up LSDBs
- Rania's salary is now funded – The Buckland Foundation (Australia)
- There are no office costs to the HGVS
- The journal *Human Mutation* plays a major role in mutation collection
- *Human Mutation* is the greatest source of published mutations
- HGVS members doing a lot of work in Central databases and LSDBs
- Consortium of HGVS members invited to put in an NIH grant

A discussion of the NIH grant submitted by a consortium of HGVS members proceeded.

Bruce Gottlieb suggested that the Society looks at its goals and what it is actually doing. Should the Society play a role in the NIH grant? Four to five years ago there was a lot of excitement within this group, what is happening now?

John Compton stated that if funding is a problem then new approaches are required to obtain it. The HGVS should go to the NIH and say "We want you to send out a request for applications (RFA) on X".

Dick Cotton stated that David Rimoin, Charles Scriver, Victor McKusick and he wrote to Francis Collins that variations were being ignored; however there was no reply from Collins.

Tony Brookes stated that the NIH does appreciate the need for variation collection however they think these applications are not fundable.

Joanne Amberger asked if other institutions such as NCI and NLM have been approached.

Saeed Teebi stated that the groups who applied for funding to NIH did not and still do not understand quite what the NIH actually wants.

Tony Brookes stated that this is talk of individuals' PROJECTS not the Society.

A discussion about stagnation of membership then proceeded.

Bruce Gottlieb has some suggestions re the stagnation of the membership:

- Where is the Society going?
- The message of the Society is not getting out.
- Does the HGVS want to do something?
- An official session at the ASHG meeting should be devoted to this.

Dick Cotton stated that he applies for such a session for each ASHG meeting but is constantly turned down by them.

Dick Cotton proposed to approach the American College of Medical Genetics.

Rania Horaitis stated that the focus of the LSDBs which were the original Mutation Database Initiative objective has been lost as the group has become preoccupied with the NIH grant. The LSDB curators thought they would get more recognition. It should be noted that most of them chose not to join the Society.

Eivind Hovig stated that it seems like the Society will disband once it has achieved its goal.

Marc Greenblatt commented that we should be looking at money for "facilitation" rather than projects. For example sponsor an informatics extraction meeting; a focus meeting for people to do the work.

Alastair Brown stated that societies are meeting facilities, they do not have projects.

Bruce Gottlieb stated that there are organisations such as the Keystone symposia that can sponsor such meetings, e.g. lets raise the issue of phenotype this is better than focussing on one project.

Haig Kazazian Jr. commented that we are concentrating on databases but only a limited number of individuals are interested in them. For the Society to grow we need to expand our interests.

Tony Brookes stated that genetic variation is not the goal it is the whole spectrum that genetic variation encompasses.

Sue Povey asked how many clinical geneticists are members.

Arupa Ganguly stated that in diagnostic laboratories when they find a mutation they have not place to go. We should bring this information into the clinical world.

Bruce Gottlieb stated that this is a good suggested for a conference for clinicians and diagnosticians only.

Richard Cotton stated that this could be a theme for the meeting next year.

Bruce Gottlieb asked how we are going to attract the clinicians and diagnosticians.

Mark Paalman said we should collaborate with a company for example a diagnostic company.

Marc Greenblatt suggested Myriad and asked if we are targeting any particular group e.g. the cancer world.

Bill Oetting stated we make connections between the diagnostic labs and the LSDBs. For example send diagnostic labs an email telling them to submit mutations to an LSDB.

Richard Cotton said Graham Taylor in the UK was trying to do this.

HGVS must try to make connection between diagnostic laboratories and LSDBs

Move to accept President's report
Moved and accepted

5. Treasurer's report – Presented by W. Oetting

William Oetting reported that the Society is doing well. It was officially incorporated November 2001 and this is now the second year.

501(c) (3) status was obtained with an advanced ruling from November 6th 2001 until December 31st 2005. The Society is now considered tax-exempt.

The balance of accounts for 2003 is a positive balance. An Audit was done for 2002 and Passed. There was a Final balance of AUD\$ 14,418.17. Tax forms for 2002 have been submitted.

Move to accept Treasurer's report
Moved and accepted

6. Membership report and Annual subscription 2004 – Presented by R. Horaitis

Rania Horaitis reported that after an encouraging start in 2001 the Society recorded no net growth in 2003. Although most members renewed their membership for 2003, the number of those choosing not to renew equalled the number of new memberships attracted, this is despite extensive advertising campaigns. This problem should be addressed.

It was noted that those members who failed to pay their dues on time last year had an interruption to their subscription of *Human Mutation*. To avoid any interruption and to continue to receive the journal on time, dues should be paid promptly after the notice is sent.

The Society continues to attract an international membership with almost half of all members from Europe a number that has shrunk slightly since last year, 36 % from North America, and 26% the from the rest of the world.

Christophe Beroud asked if it would be possible that people from labs can join without a journal subscription since one lab member already receives the journal.

Colette Bean representative of Wiley the publisher of *Human Mutation* stated that the Society voted for mandatory subscription with membership in order to receive substantial discount of the journal. An optional subscription for members will make the journal more expensive at approximately double the rate. Wiley is flexible and can change the subscription method but a decision needs to be made by April/May 2004.

Tony Brookes asked if the journal is an incentive to join HGVS or not as many small labs will never afford the US\$200 to join.

Sue Povey said she thinks it a disincentive as she has online access. Bruce Gottlieb and Joanne Amberger agreed.

Arleen Auerbach and Saeed Teebi stated they thought the journal was an incentive to join.

Motion for an email vote of the membership be done to decide if subscription of *Human Mutation* should remain mandatory or not.

Moved by Diane Cox
2nd by Conover Talbot Jr.

Passed with no dissention

7. Publication report – Presented by C. Bean & M. Paalman

Wiley representatives C. Bean and M. Paalman stated that *Human Mutation* had a stellar year with several major milestones in 2003.

The journal's impact factor continued to climb - now to an all time high of 6.894 mainly due to high quality review and mutation update articles along with special issues. Due to increased impact factor a rise in submissions was made. Two special issues were published with a large number of downloads for the p53 issue.

In August, Wiley launched a sophisticated new interface for *Wiley InterScience*, the service that provides online access to *Human Mutation*. The new interface also provides prominent Society branding.

8. Web report – Presented by R. Horaitis

Rania Horaitis reported that the HGVS formally registered the domain name www.hgvs.org in 2001 and continues to maintain this URL.

The extensive lists of databases available on the HGVS website show a high volume of use, especially the lists of LSDB links with well over 1000 hits weekly.

This year, the Mutation Nomenclature pages were moved from the Leiden Muscular Dystrophy pages and added to the HGVS Website under the "Recommendations for nomenclature" link and are constantly updated by Dr. Johan T. den Dunnen and include numerous examples of naming mutations. A new feature to this site is the "mutation checklist" which is useful to anyone wishing to name the mutations they have discovered when writing publications. See: <http://www.hgvs.org/mutnomen/>

Abstracts for the 2003 AGM were available online before the meeting after acceptance and we will continue to do this for all future meetings.

9. Office Report – Presented by R. Horaitis

Rania Horaitis the Executive Secretary of the HGVS briefly reported on the office activities. The HGVS Office recently relocated to brand new office space and handles administration of the Society funds and day to day business such as membership processing, newsletters, Website management (see Website report), organisation of two meetings per year, exhibitor's booths and handling member and other queries. Other projects undertaken include solicitation of *Human Mutation*, Mutation Updates and Reviews and NCBI monographs, encouragement of new LSDBs and more.

10. Election of council members, 3 places for election

Three positions on the Board are to be vacated. Since no nominations were received the current rotating Board members are to stand for re-election. They are Christophe Beroud, A. Jamie Cuticchia and Conover Talbot Jr. A vote should probably occur anyway.

11. Committees for membership

The HGVS committees listed in the meeting booklet are not very active and something should be done about this.

12. Any other business

The use of the two Society pages per issue in Human Mutation was briefly discussed with no outcome.

Meeting adjourned

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

Date
