

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

**11<sup>th</sup> November 2008**

**5.45 PM – 6.58 PM**

**Rooms A and B, Hilton Garden Inn Centre City  
Philadelphia, PA, USA**

The meeting was Chaired by Professor Graham R. Taylor, President- Elect.

**Apologies:**

Professor Richard G.H. Cotton, President

**Members Present:**

Anderson Paal Skytt  
Auerbach Arleen D.  
Beroud Christophe  
Brown Alastair F.  
den Dunnen Johan T.  
Giardine Belinda  
Gottlieb Bruce  
Greenblatt Marc S.  
Hadjisavvas Andreas  
Hardison Ross  
Horaitis Rania  
Oetting William  
Paalman Mark  
Polychronakos Constantin (arrived 6.30 PM)  
Qi Ming  
Rogan Peter K.  
Talbot Jr. Conover  
Taylor Graham R.  
Utsunomiya Joji  
Vihinen Mauno

**Non-Members Present:**

Billingsley Gail  
Bruford Elspeth (arrived 6.30 PM)  
Cox Diane  
Kojima Toshio  
Luoma Leah  
Maglott Donna (arrived 6.23 PM)  
Minoshima Shinsei  
Silk Gregg (arrived 6.23 PM)  
Wilson Anne  
Yakubovskaya Marianna  
Young Kim

## 1. Approval of Agenda

Addition to Agenda.

### 10. Other Business

(b) Committee to develop a minimal requirement for Locus Specific DataBases, ask for volunteers.

Motion to approve agenda.

Moved by Conover Talbot

2<sup>nd</sup> – Alastair Brown

Motion passed with no dissention

## 2. Minutes of the previous meeting for approval

No changes to minutes.

Motion to accept minutes of 2007.

Minutes passed with no dissention.

## 3. Matters arising from the previous meeting

### (a) Incentives to join the Society

Peter Rogan commented that individuals who are not Board members should actually do something for the Society, as that is an incentive to join.

Graham Taylor stated members should be involved or support be requested from members, e.g. minimum requirements for databases, put out for support from the membership.

Bruce Gottlieb commented that the Society did as much as it could in regards to the membership dues yet there are still people who attend meetings who decide not to become members.

Rania stated they choose not to join despite the large registration fee discount that is clearly indicated to them.

Mark Paalman came up with another idea as in 2007 he said that Wiley-Blackwell can put a membership form in PDFs of the article proofs for *Human Mutation* however they have not done that yet, but it is on a shortlist of things to do- so then all *Human Mutation* authors will get the form.

Graham Taylor suggested partnerships with other groups such as ASHG, ESHG and give members a discount.

Bruce Gottlieb however stated that this was tried once before and there were some problems, that he can't remember.

Conover Talbot stated that HGVS does have a relationship with them but it has not been taken to the next step.

### (b) Develop a collaboration with HuGENet

There was no comment about this as the collaboration seems to have come to a standstill.

### **(c) The Society should write to journals regarding nomenclature standards**

Conover Talbot Jr. suggested that journals could make a statement that submitting data into Locus Specific DataBases would not jeopardize their publication.

Alastair Brown stated that the people in the room who are on Editorial Boards should bring pressure on those journals to use the HGVS mutation nomenclature.

Johan den Dunnen commented that he does it for every paper he reviews for any journal; he writes a specific paragraph about the use of correct nomenclature in his review.

Graham Taylor said that NCBI is now supporting what the HGVS does. He suggested that a letter be put on the website regarding the use of correct nomenclature.

Bill Oetting suggested that the HGVS write a letter to the Editor regarding use of nomenclature.

Johan den Dunnen said that Human Mutation should make more stringent requests to authors. Is that something you share with other journals?

Mark Paalman stated he was thinking that Richard Cotton who is so good at getting people together and talking to them, has already tried to do this with medical genetics journals in the context of clinical data. If the Co-Editors of *Human Mutation*; Richard Cotton and Garry Cutting wrote letters to other Editors of journals to try to get a discussion going at the Editor level maybe there would be some action taken by the other journals.

Ross Hardison asked if *Nature Genetics* requires the use of HGVS nomenclature and if not, Myles Axton should be pressured to do something about this. Myles Axton is keen on microattribution, one component of it is that a database ID needs to be cited, if dbSNP is the place where the database ID comes from and it is using HGVS nomenclature then it is moving in the right direction and *Nature Genetics* and *Nature* using the nomenclature is great! Ideally we want high profile journals to use it as this will make a big difference.

Mark Paalman stated that getting journals to use the HUGO HGNC nomenclature was a lot easier to do and this is a lot more complicated.

#### Actions

The Co-Editors of *Human Mutation* are to write letters to other Editors more formally regarding the use of HGVS nomenclature.

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

The project of sharing data between Locus Specific DataBases and Central Databases is going well. NCBI is constructive and Phencode is an interesting way as well.

Diane Cox stated that sharing of data is beneficial and asked what curators are doing regarding obtaining funding for their Locus Specific DataBases. She asked if there has been any progress in getting Locus Specific DataBases funded as it is such a costly effort.

Mark Paalman stated that HGMD has recently announced that they have become fully self sustaining but they have restrictions, including a 2 year embargo on data.

Mauno Vihinen said that HGMD download databases and they use the data.

Diane Cox said that HGMD download the data from other databases for free.

Graham Taylor stated that it is similar to the Craig Venter and public sequencing effort and there is nothing anyone can do about it.

Diane Cox asked if anyone has found a source of money for their Locus Specific DataBase.

Mark Paalman said that the only one he knows of is the one sponsored by a company called MC Global as part of the Human Variome Project "Adopt a Gene" project.

Christophe Beroud stated that patient organizations allow curators to obtain some money and support. Patient organizations can fund Locus Specific DataBases and pay for curators. A Locus Specific DataBase must try to become self sustainable after five years of financial support. The curator should allow the approach by companies for clinical trials and patient selection and make the company pay for these requests. This is a great way to get some income especially if your gene of interest has any specific genotyping related interest.

Bruce Gottlieb stated that the only person in Canada that has ever been funded for a Locus Specific DataBase is Charles Scriver. He said that he knows of a database that has been funded in Europe, and received substantial funding, it is based in Germany.

Christophe Beroud said that the European Union could fund for five years. To get that funding, you need to show how the database will become self sustaining after those first five years. Thus, curators need to think about the later five years and so linking with companies is a way towards funding. Another idea is to try and obtain funding from Governments by going to registries for example funding for a "French database".

Graham Taylor stated that obtaining funding for Locus Specific DataBases is difficult because governments think that they are already funding databases. Should more pressure be applied to governments?

Bruce Gottlieb stated that as the majority of HGVS members well know that many groups have applied for money for example from NIH but none of them ever get funded. One of the reasons is that governments do not want to fund something that is universal as opposed to something that is based in one place.

Christophe Beroud again stated that a key point regarding funding is that the database needs to contain the clinical information. For example the DMD gene, there is more than one database in the USA through NIH funded. NIH is not interested in small things like rare diseases and so on.

Mauno Vihinen stated that Locus Specific DataBases were something new 10 years ago. Now they have become infrastructures and money for that comes from other sources such as NCBI, EBI etc.

Graham Taylor stated that institutes such as NCBI and EBI provide the hardware but not the curation.

Alastair Brown and Mauno Vihinen stated it is the curation that needs funding, not the hardware.

Johan den Dunnen stated that the European Union funds hardware for the LOVD and Gen2Phen project when they don't need any funding for hardware!

Graham Taylor stated that NCBI and EBI don't want to fund the actual people doing the curation; Paul Flicek from the EBI who attends the Human Variome Project meetings seems to be understanding the to problems of funding database curation.

Christophe Beroud stated that when you apply for money and you say you need an expert for curation of genotype data and the institution says you don't need an expert, you have to convince them that it is not so easy and that you have a specific gene, you need to convince then you need an expert. This is one point where we can say the curation process cannot be performed by anyone anywhere.

Bruce Gottlieb stated that the phenotype section is not extensive in most databases. You can send the genotype information but the phenotype information needs to be curated. Gen2Phen is working at this issue now.

Ross Hardison states that the problem is how the salaries for curation are going to be paid. He was never optimistic that they were going to get a grant for an LSDB only. Is this a critical issue with the data sharing with central repositories?

Graham Taylor stated that the LSDB data sharing project is going well and that Johan den Dunnen has already put a document together. This has been submitted to *Human Mutation* for publication and is now under review.

Peter Rogan referring to Greg Feero's presentation at the scientific meeting earlier stated that companies are mining the literature and re selling the data with high profit margins, this is the information they want to get out the community.

Everyone exclaimed they are getting the data for free!

Graham Taylor asked if anyone was in favour of getting together with the companies.

Peter Rogan suggested we get them to sponsor!

Ross Hardison asked if the HGVS has endorsed the idea of Locus Specific DataBases sharing data with central repositories using the mechanisms that Donna Maglott explained.

Graham Taylor answered that yes there has been a formal endorsement.

Donna Maglott stated that NCBI will help people find out how to do it. They will help people find out about how to submit their data as they don't know how to get started.

Thus, sharing LSDB data with central repositories is a recommendation of the HGVS and a paper is under review about this at *Human Mutation*.

Ross Hardison suggested that the paper be an official statement from the HGVS.

#### **(e) Relationship between the Human Variome Project and the Society**

This topic was only briefly mentioned where it was generally agreed the two should run in parallel.

#### **4. President's Report**

As the President Richard Cotton was absent due to illness, there was no report other than what was published in the meeting booklet.

#### **5. Treasurer's Report**

Bill Oetting stated that the Society's two bank accounts (United States and Australia) were both audited through 31<sup>st</sup> December 2007 and all was found to be in order by the auditor. The report was included in the meeting documents.

He went on further to say the Society is still in the black with some money in the bank, the main income being membership dues and meeting registrations and some sponsorship too. The expenditure is mainly meeting costs, bank fees and journal subscription costs with a few miscellaneous items such as purchase of a projector and exhibitor booth costs.

Therefore, the Society is doing well financially, however it has taken a long time to accumulate such little funds.

## 6. Office Report

Rania Horaitis briefly discussed a few items from the Office Report. She reported that the Society attracted 47 new members in 2008 and 3 on the HGVS meeting day bringing the total number of members to 101. Still, some members still fail to renew their memberships. However the lower membership dues seems to be working, although most new members join to obtain the discounted meeting registration fee.

Rania continued to report on the Election of the President – Elect, Graham Taylor and announced that a call for nominations of officers of the Society will be made shortly.

She continued to report about the successful meetings held. The one in Barcelona attracting 110 registrants, was the 1<sup>st</sup> time the Society had held a satellite meeting to the European Society of Human Genetics meeting. It is hoped that Vienna should be equally successful.

Rania reported that the Society had some promotion during the year, exhibitor's booths at ESHG and ASHG, promotional mouse mats with variation nomenclature on the back and the Wiley Interscience broadcast email.

She announced the Website was being revamped to a new look and the LSDB listing had undergone some updates.

All in all, it was a busy year.

## 7. Locus Specific Database Report

The report was presented in the booklet as required by the minutes of the 2007 Annual General Meeting however it was not discussed.

## 8. Publications Report

Mark Paalman, the Managing Editor of *Human Mutation* briefly summarized his report as presented in the Annual General Meeting papers the main points being the new article design that will feature the HGVS logo on the top right hand corner of every paper and the new feature in the journal called "In this Issue" which will highlight selected papers from each issue each month. The journal continues to be a successful respected journal and maintains its standing as one of the top five genetics journals devoted solely to human genetics research and reviews.

## 9. Election of Council Members

There will be an election of Council Members to serve a term (2009-2011).

The nominations committee is requesting suggestions for nominations for officers of the Society. Four positions on the Board of Directors of the HGVS will become vacant at December 31<sup>st</sup>, 2008. 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:

- Anthony Brookes
- Johan den Dunnen
- Marc Greenblatt
- Heikki Lehvälaiho

Current Board members with expiring terms need to indicate if they wish to stand again for election. Board members will serve a term of three years.

Rania Horaitis will send an email announcement to all members. Please reply to Bill Oetting (oetti001@umn.edu) and Rania Horaitis (rania@variome.org) by November 30<sup>th</sup>, 2008, 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 8<sup>th</sup>, 2008 and will be due by December 31<sup>st</sup>, 2008.

## **10. Election of Office Bearers**

At this time the Society is calling for candidates to run for Secretary/Treasurer. This person needs to be a United States permanent resident due to the tax forms that need to be filled out and submitted yearly. Any member interested should contact Rania Horaitis, who will also make an email call to all members. If no nomination is received, Bill Oetting will remain as acting Secretary/Treasurer until a replacement is found.

## **11. Other Business including items from the Board Meeting**

### **(a) Themes for future meetings**

Possible themes for the forthcoming meetings in Vienna and Honolulu were actively discussed.

#### **Scientific Meeting: 23<sup>rd</sup> May 2009, Vienna, Austria (Satellite of European Society of Human Genetics)**

(i) "Epigenetics, mutations and cancer".

Diane Cox was concerned that that topic is too focused.

Marc Greenblatt stated that he tried to spread this out in the clinical genetics theme of the day by inviting two cancer focused speakers and two non- cancer focused speakers.

Everyone thought that the theme "Clinical genetics and human genome variation" was great as it was not too focused. A topic that allows for that is needed.

Alastair Brown commented that the topic is only part of the day as there is always space in the program open to "variation general topics" to attract the regular attendees.

Graham Taylor asked if cancer is too restrictive.

Diane Cox stated that a number of people would not want a whole day on cancer however an afternoon, a session would be great.

Arleen Auerbach stated that at the Barcelona meeting, people who don't normally come to the HGVS meetings attended so the Society should be aware of the new group of people who would attend. .

(ii) Genome Wide Association Studies

Peter Rogan stated that people will be drawn to Vienna with an interest in genome wide association studies, and that the Society should grab onto that opportunity, and specifically last mile problem of genome wide association studies. Hence a title could be "Genome wide association, the last mile problem"

(iii) Variants affecting RNA

Johan den Dunnen suggested Variants affecting RNA. It was not discussed.

A vote was taken in the room with “Genome wide association the last mile problem” being the topic decided for the Vienna meeting. The Chair of the meeting will be Peter Rogan.

**Scientific Meeting and Annual General Meeting: 20<sup>th</sup> October 2009 Honolulu, HI, USA (satellite of American Society of Human Genetics)**

Two topics were suggested the first coming out of the Board meeting.

(i) Outcomes of Next Generation sequencing

(ii) Variants affecting RNA, RNA Sequencing

A vote was taken in the room with “Outcomes of Next Generation sequencing” being the topic decided for the Honolulu meeting. The Chair of the meeting will be Graham Taylor; the Co-Chair will be Bill Oetting.

**(b) Committee to develop a minimum requirement for Locus Specific DataBases.**

This topic was added to the Agenda at the Board meeting but failed to be discussed here. The Chair of the Committee is Johan den Dunnen and an email call for volunteers will be made.

**(c) Other Business**

Motion to formally thank Richard Cotton and find some way of honouring him

Moved – Bruce Gottlieb

2<sup>nd</sup> Arleen Auerbach

Motion passed with no dissention

Graham Taylor stated what a superb job Richard Cotton has done and with limited support, and that he must be very pleased, so thanks to Dick.

All thanks.

Officially thank Rania Horaitis for her work for the Society.

All thanks

In Summary:

- Encourage non-Board members to do something for the Society
- The Co-Editors of *Human Mutation* are to write letters to other Editors more formally regarding the use of HGVS nomenclature.
- Election of Council Members
- Nomination of Secretary/Treasurer
- “Genome wide association the last mile problem” is the topic decided for the Vienna meeting 23<sup>rd</sup> May 2009. The Chair of the meeting will be Peter Rogan.
- “Outcomes of Next Generation sequencing” is the topic decided for the Honolulu meeting 20<sup>th</sup> October 2009. The Chair of the meeting will be Graham Taylor; the Co-Chair will be Bill Oetting.

- Develop Committee to develop a minimum requirement for Locus Specific DataBases; Johan den Dunnen the Chair; call for Volunteers.
- Formally thank Richard Cotton and find some way to honour him.

Meeting Adjourned 6.58 pm

Signed by Graham R. Taylor as a correct record: \_\_\_\_\_

Date: \_\_\_\_\_