

**Minutes of the Business Meeting of the
HUGO Mutation Database Initiative**

October 12, 2001

San Diego, California, U.S.A.

The meeting was opened by Prof. Richard Cotton.

Present:

Auerbach Arleen
Bean Colette
Beroud Christophe
Brown Alastair
Christodoulou John
Compton John G.
Cotton Richard G.H.
Cruts Marc
Cuticchia A. Jamie
Darlison Matthew
Fung David
Glavac Damjan
Gottlieb Bruce
Greenblatt Marc S.
Horaitis Rania
Houle Jean-Francois
Kazazian Jr. Haig
Kraus Jan
Lehväslaiho Heikki
Li Jinlong
Maheshwari Manjula
McKusick Victor A.
Oetting William
Paalman Mark
Ravine David
Scaringe William A.
Silk Gregg W.
Talbot Jr. C. Conover
Teebi Saeed
Van Ommen Gert-Jan
Vihinen Mauno

Chairman's Report

The Mutation Database Initiative N.I.H. grant was submitted on the 15th of June. An international team of MDI members has submitted one grant together with subcontracts for different parts. The general aim of the application is to establish a public domain system for the capture of human gene-related variations, and phenotypes.

A campaign to recruit orphan gene editors has begun. We have recruited around 80 GE's for over 100 genes and will be recruiting more at the ASHG meeting through our booth (no. 737).

Secretary/Treasurer's report

There are several issues in setting up accounts that transfer international funds. Using credit card accounts appear to be the most efficient and cost effective way to do this. There is currently a credit card account that is being used by MDI. We will begin to use this account system for the Society. As the Society grows, other means and mechanisms may be put into place. Using the current MDI VISA account will minimize the initial costs of setting up the Society.

Acceptance of the Board and Council members

Initial candidates for officers and council members were presented. They are as follows:

President – Richard G. H. Cotton
Secretary/Treasurer – William S. Oetting
Council Members

- A. Auerbach
- C. Beroud
- E. Beutler
- A. Brookes
- A. Brown
- C. Conover Talbot Jr.
- A. J. Cuticchia
- M. Darlison
- J.T. Den Dunnen
- B. Gottlieb
- H. Lehv slaiho
- V. McKusick
- C. Scriver

N. Shimizu
G. Taylor
E. Tuddenham
M. Vihinen

A motion to accept the officers as presented except R. Dalgleish and D. Wallace who are accepted after stating a willingness to serve was made.

Moved by Victor McKusick
Second by Arleen Auerbach
Passed with no dissention

Objectives of Society

The objectives and mission statement of the Society were presented for discussion. They are as follows.

Objectives:

To foster discovery and characterization of genomic variations, including population distribution and phenotypic association.

To promote collection, documentation, annotation and free distribution of genomic variation information and associated clinical variation.

To foster the development of necessary methodology and informatics.

Mission statement:

To enhance human health through identification and characterization of changes in the genome that lead to susceptibility to illness. To this end, to collate the genomic information necessary for molecular diagnosis, research on basic mechanisms and design of treatments of human ailments

A motion to accept objectives and mission statement as stated was made.

Passed with no dissention

Ratification of name of Society

Name of society

Human Genome Variation Society (HGVS)

Moved Connie Talbot
Second Alastair Brown
Passed with no dissention

A motion was made to obtain the domain name www.hgvs.org and to look into a trademark for the Human Genome Variation Society.

No vote was taken

Jamie Cuticchia will look into obtaining a domain name.

A possible competition for a logo was proposed

Formal Establishment of Society

The bylaws of the Society were presented and discussed. These bylaws were initially from the PanAmerican Society for Pigment Cell Research. These have been looked at in USA by March of Dimes lawyers and minor corrections were made.

Incorporation within the United States for the Society will be looked into by Connie Talbot and Arleen Auerbach. A possible site for incorporation is Maryland

A motion to incorporate the Society with the presented bylaws allowing for changes as necessary to comply with state laws was made.

Moved by Bruce Gottlieb
Second by Victor McKusick
Passed with no dissention

A motion to formally establish the Society was made.

Moved by Arleen Auerbach
Second by Mark Paalman
Passed with no dissention

Human Mutation and Society agreement

A document stating that Human Mutation will be the official Journal of the Society has been signed with Wiley, subject to approval by Society membership. Only those members who pay their dues will get the Journal as part of this agreement.

Colette Bean, Senior Editor Life Sciences Division John Wiley and Sons Inc.

Suggested mandatory subscription would be best for the journal and society.

Will explore discounts to other Wiley New York journals.

Will explore colour printing discounts to society members.

Stated that any colour change to the *Human Mutation* cover must be limited to two colours only. Possibly it can be redesigned to society colours and will explore this.

The January 2002 issue will contain a statement about the new Society.

Answering Bruce Gottlieb who asked about publicity for the Society, Colette commented that Wiley Interscience "genetics" members have already been spammed about the new Society affiliation.

Reasonable to review Wiley/Society agreement after three years.

Wiley is willing to give a discount to select poorer countries but only a select few. The society must nominate who would receive the discount dependant on location, electronic journal access is preferred.

Colette also mentioned that the Society will have to pay for postage of the journal, Wiley will not include this in the \$100 subscription. This can be very expensive when posted outside the US. Postage to Canada also incurs a 7% government surcharge. The society will have to take out the postage costs per issue from the \$200 membership dues. If this is \$3 per issue this amounts to \$36 per year! Greater than 50% of the MDI members and probably future Society members do not reside in the US so postage is an important issue.

Haig Kazazian Jr. suggests that Wiley and the Society split the cost of postage for each issue.

There was no motion made regarding the postage issue.

A potential reduction in membership dues was proposed for student and/or post-doctoral trainee only if the head of laboratory is a member of the Society.

A motion was made that the society go with the mandatory subscription rate of US\$100.00 including the print journal and on-line access and add to dues. (Option B in the Wiley agreement).

Moved by Bruce Gottlieb
Second by Haig Kazazaan Jr.
Passed with no dissention

Formalize what was agreed upon last year the *Human Mutation* is the formal journal of the Society. Society has pages in the journal that can be used as needed. A three year contract is stated.

A motion was made to ratify the signature of the Society – Wiley contract

Moved Jamie Cuticchia
Second by Arleen Auerbach
Passed with no dissention

Setting Society membership fees and subscription rate for Human Mutation

A motion was made that the regular membership of the society is US\$200.00, including the journal subscription for *Human Mutation*.

Move by Bruce Gottlieb
Second by Saeed Teebi
Passed with no dissention

Human Mutation and MDI collaboration regarding publication of Gene Variation Reports (GVRs)

Mark Paalman, Managing Editor Human Mutation, Wiley-Liss

A move to change name of Gene Variation Reports (GVR) to Genome Variation Reports (GVR). No action on this.

What specifically is a GVR? What type of sequence variation is appropriate? The Society should take the lead on this as well as standards for acceptance of publication.

Comments on publication. These will be published in an electronic format only. Hopefully GVS will have a PubMed ID. GVRs will be HTML documents and will

include hyperlinks to other databases including the WayStation. These will be free access publications.

Questions: Are GVRs going to be part of the impact number for *Human Mutation*?—GVRs as part of a table in *Human Mutation* or possibly some other way to publish their existence in *Human Mutation*.

Get XML format finalized and get an adhoc review board, and enter submissions into the WareHouse, or at least put the submissions into the WayStation database.

Agree to begin the beta test for GVR submission. Also that Dick Cotton and Haig Kazazaan write an editorial on the society/journal agreement, as well as the concept of GVR submission. The Society will have two pages per issue. One page will be a membership application and a second page will contain the mission statement for the Society and any other business or announcements.

Other Business

Relationship of Society to HUGO.

We would like HUGO to acknowledge the role of the Society.
MDI has been a standing committee within HUGO. We would like the Society to continue to have a role with HUGO.

Gert-Jan van Ommen

As a Society we would like to state a continued connection with HUGO.
Suggests HUGO members become HGVS members and vice-versa.

A motion was made that HGVS seeks to continue to have a formal affiliation with HUGO and that the HGVS continue to provide its expertise to HUGO for issues of mutual interest.

The following statement was made “Recognising the historical relationship with HUGO stemming from the MDI the HGVS seeks to continue a formal relationship with HUGO as an independent society”.

Moved by Arleen Auerbach
Second by Victor McKusick
Passed with no dissention

It was stated that the Society should continue a relationship with the March of Dimes, and Dick Cotton will undertake to explore this.

A motion was made to thank The March of Dimes for their help and role in supporting the HGVS.

“We appreciate the role of the MOD helping our organization through its formative stages”.

Moved by: ?

Seconded by: ?

Passed with no dissention

Ethical Standards

The society should continue to work with HUGO on ethical aspects of sequence variation databases. This includes privacy and data ownership. It was stated that we need to have a Society statement on the ethics involved in databases containing sequence variation, and continue the relationship with the HUGO ethics committee.

Clinical Mutations

The Society encourages individuals to work on obtaining mutations from clinical databases. Also for individuals to try and obtain funding to obtain these mutations.

Sustainability

People should think about trying to obtain private funds to help support the Society.

Advisory Committee for NIH Grant application

The following individuals are members of the N.I.H. grant advisory committee.

Stylianos E. Antonarakis, Christophe Beroud, Ernest Beutler, Alastair Brown, Aravinda Chakravarti, Raymond Dalglish, Matthew Darlison, Bruce Gottlieb, Haig H. Kazazian Jr., William Oetting, Nobuyoshi Shimizu, David Valle, Gert-Jan Van Ommen, Mauno Vihinen.

Feedback from NIH states we should not add any additional names.

Electronic lists. Heikki Lehväslaiho will set up some listservers for the Society membership.

Other

It was stated that we should send out a formal request for membership. The initial membership will be seen as a Founder membership of the Society.

Next Business Meeting

The next Business meeting will be at the ASHG meeting at Baltimore October 15, 2002.

Meeting adjourned

Respectively Submitted

William S. Oetting

SIGNED R. G. H. COTTON PRESIDENT

DATED