

## New President Elected

An election was held in September to elect a President as Professor Richard Cotton's term as President of the Society was coming to an end. Professor Graham Taylor has been elected as President and will begin his term 1st January 2009, the term will end 31st December 2011.

Graham R Taylor PhD FRCPath (Molecular Genetics) is Professor of Molecular Medicine at the University of Leeds and Head of Genomic Services, Cancer Research UK. He is an Honorary Consultant Clinical Molecular Geneticist and a UK Health Professions Council Registered Clinical Scientist (Genetics).

Research activities include development and improvement of mutation detection technologies, development of high throughput sequencing platforms, diagnostic applications of clonal sequencing, molecular cytogenetics, molecular pathology and diagnosis of

rare recessive genetic disease in consanguineous families. He leads the pilot project and is a current member of the steering group for the UK Diagnostic Mutation Database (DMuDB.net).

He has organised several national and international meetings and edited several books on the topic of mutation detection. He is a Course co-ordinator for the HUGO mutation detection workshops 1998-2008. Chair of the Cancer Research UK SNP committee 2006-2008, an advisory committee for genome wide association (GWA) and other large-scale association studies. The GWA studies were considered to be one of CR-UK's major research achievements of 2007/8. Lead in Cancer Research UK clonal sequencing evaluation on the cost and performance of next generation sequencers (2007) Lead a review of Cancer

Research UK bioinformatics demand and capacity (2007). Established a Next Generation Sequencing Facility at Leeds from April 2008.

Honorary Member of the NIHR Faculty (UK National Institute of Health Research). Honorary membership of the Faculty is offered to highly regarded researchers who do not receive NIHR or PRP funding but who are carrying out or supporting people or patient-based research. This includes researchers employed by partner organisations such as charities or industry.

Member of Caddy Committee lead by Professor Brian Caddy, for the review of low copy DNA testing for forensic testing (Caddy Report 2008).

### HGVS News :

- New President Elected
- Meeting held in Barcelona
- Mutation Databases List Updates
- OMIM links out to HGVS LSDB list
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## Meeting in Barcelona

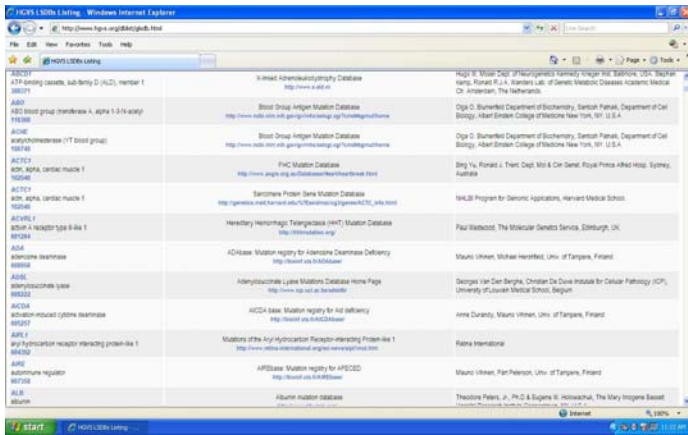
The HGVS meeting held in Barcelona, Spain with the theme of "How to explore human genotype to phenotype relationships" was very well attended with 110 registrants. This is the 1st time the HGVS held a meeting as a satellite to the European Society of Human Genetics annual meeting and we were very pleased with its success as many people attended the meeting for the 1st time. Thus, we will be holding another satellite meeting to ESHG next year in Vienna on May 23rd.



Mutation Databases List Updates

One component of the HGVS-supported WayStation project is to maintain the central database of LSDBs, and to produce the web list displayed on the HGVS and WayStation's respective websites (<http://www.hgvs.org/dblist/glsdb.html> and <http://www.centralmutations.org/Lsdb.php>). This resource provides one-point access to all known LSDBs. LSDB curators and others can also submit information about novel LSDBs for inclusion in the database.

We would like to announce an LSDB Submission Tool for the LSDB list. If you would like to submit a new LSDB to the list you can now fill in the form with the details. Once checked and approved, the new database will be added to the list. The list will be updated online fortnightly.



**“NCBI is now using the HGVS central database of LSDBs service to link OMIM gene entries to their related LSDBs”**

OMIM Links out to HGVS LSDB list

NCBI is now using the HGVS central database of LSDBs service to link OMIM gene entries to their related LSDBs. For example, OMIM entry +261600 (Phenylketonuria, PKU), displays the HGVS link under LinkOut on the left side of the page. This link takes users to the HGVS's LSDB list entry for Charles Scriver's PAH LSDB, which users follow to access the knowledgebase. NCBI generates and updates these OMIM links from files provided by

the HGVS and WayStation, and mounts them on their website. Conversations are now under way to extend NCBI-to-HGVS LSDB links to include NCBI's Entrez Gene Database, thereby providing its users access to the best curated gene variation information available.

Exhibitor's booth at ASHG

The Society will be hosting an exhibitor's booth at ASHG in Philadelphia from 12<sup>th</sup>-14<sup>th</sup> November. Please drop by the booth and meet fellow members.

Booth Number: 122

Website Undergoing Update and Changes

The HGVS Website is in the process of being completely updated and will also have a new look. The new look Website should be completed in the next few weeks. Apologies if you happen to come across any pages that have not been updated during the changeover.

Election of Board Members

The Society will hold an election for Board Members beginning December 1<sup>st</sup>. We would therefore like to call for nominations to the Board. Any member of the Society wishing to be nominated or nominate someone else should send in their nomination by the 15<sup>th</sup> of November to Rania ([rania@variome.org](mailto:rania@variome.org)). There will be four positions available. Terms are for three years and begin 1<sup>st</sup> January 2009. An official notice will be sent.



New website coming soon...

## Forthcoming Scientific Meeting & Annual General Meeting

The next one-day HGVS Scientific Meeting with the theme of "Clinical Genetics & Human Genome Variation" will be held as a Satellite to the American Human Genetics Meeting in Philadelphia, PA, USA on the 11th November 2008.

The remarkable progress in the basic science and technology of genetics over the last decade is rapidly being translated into the clinic. The promise of "individualized genetic medicine" based on an understanding of inter-individual human genetic variation has attracted both popular and scientific attention. The genetic community faces challenges in scientific discovery, in applying discoveries to clinical disorders, and in making these advances comprehensible to physicians and patients. This HGVS special session will feature

speakers who are at the forefront of each of these areas. We will explore the science of genetic variation and cancer, genetic variation and non-malignant diseases, and the presentation of information on genetic variation to physicians and to the public.

### Invited Speakers:

#### Greg Feero, NHGRI, NIH

Presenting genetic/genomic data to practitioners and patients

#### Bill Isaacs, Johns Hopkins, Baltimore

Advances in Prostate cancer genetics

#### James J. Hudziak, University of Vermont, College of Medicine

Genetic Variation in Behavioural Genetics

#### Sharon Plon, Baylor Cancer Genetics Clinics, Houston

Classifying genetic variants in cancer susceptibility genes

In addition to the invited speakers, we have two related company lectures; Idaho Technology and Affymetrix and some high-quality abstract submissions that will be presented orally and in a poster session.

The program will be available in a few days on the meeting Website: <http://www.hgvs.org/Philly08/>

The afternoon portion of the day from 5.30 PM will be devoted to the Annual General Meeting of the Society. All members are encouraged to attend if they are in Philadelphia.

"Anyone wishing to make an announcement of interest to members in these newsletters please send a short paragraph to Rania at ([rania@gdrc.hfi.unimelb.edu.au](mailto:rania@gdrc.hfi.unimelb.edu.au)) to be included in the next issue"

## Correspondence

### Gene & Genome Viewers

from Ray Dalgleish

**NCBI Genome Workbench** (<http://www.ncbi.nlm.nih.gov/projects/gbench/>).

#### Application

This application is available for PC, Mac and Linux. Although not advertised on the homepage, there are 32-bit and 64-bit Linux RPMs available at <ftp://ftp.ncbi.nlm.nih.gov/toolbox/gbench/ver-1.1.3/>. I manage to successfully translate the 32-bit RPM, using alien, into a 32-bit DEB package that installed correctly into Ubuntu Linux 8.04.

This is currently my favourite gene browser.

**GeneWindow** - <http://genewindow.nci.nih.gov>

#### Browser based viewer

The GeneWindow tool is a great product from the National Cancer Institute. People looking for "browser" type tools to view genes, exons, coding regions, and translation sequences should take a look.

This runs directly Windows in Internet Explorer if you allow it to run an Active-X Control that installs Adobe SVG plug-in version 6. It only runs in Firefox on a PC if you manually install the plug-in. Unable to get it to work with Ubuntu Linux even though Firefox 3 has built-in support for SVG. I cannot comment on whether or not it works with a Mac.

**NCBI GraphicalViewer** - <http://www.ncbi.nlm.nih.gov/sites/entrez>

#### Browser-based viewer

This appears to work with any browser that can handle Javascript. If you search for a nucleotide sequence using Entrez, you will see a blue-coloured link called

"Reports" just to the right of the accession number for each result in the list. Select "Graphic" from the menu that drops down to view the sequence graphically. The view already is fairly feature-rich and appears to be improving from week to week.

In addition to these viewers, there are tools for checking mutation

nomenclature:

**Mutalyzer** - <http://www.humgen.nl/mutalyzer/>

**Mutation Checker** - <http://mutation.sanbi.ac.za/checker>

## Correspondence continued

### LSDB Data Integration Project

Dear LSDB Curators,

On behalf of the HGVS and the GEN2PHEN project we are writing about an international collaborative project to integrate LSDB data with central repository and genome browser information. The project builds on previous efforts including projects such as PhenCode and Sue Povey's providing an extract of some of her TSC2 data to NCBI. Indeed, Donna Maglott or Ray Tully at the NCBI may have already contacted you regarding reference gene sequences. An international collaboration, which includes the EBI, the NCBI, and the GEN2PHEN project, aims to create integrated data resources and visual interfaces based on consolidating core information from LSDBs with genome resources such as dbSNP and Ensembl. The overall goal is to create a useful public resource, while

giving full acknowledgement and links to the Locus Specific DataBases that will continue to maintain their own detailed records.

To participate, we ask that you submit merely the core content from your LSDB to either the NCBI or the EBI. Suggested data elements for exchange are listed below, but additional or fewer or alternative items may be submitted based on the needs of your LSDB. The question of 'what is core LSDB content' has been addressed by Johan den Dunnen in a document recently sent to HGVS members for comment (see next page). It is not necessary for the data from your LSDB to be made public as part of participation in this project. However, if you are happy for us to release the core data from your LSDB to the public, that content will be integrated and released through public genome browsers and other resources.

If you are interested in participating, please send a message to either Paul Flicek (flicek@ebi.ac.uk) or Donna Maglott (maglott@ncbi.nlm.nih.gov) to discuss data transfer. Data previously provided to either the EBI or the NCBI does not need to be resubmitted.

With best regards,

Anthony Brookes  
(ajb97@le.ac.uk)  
on behalf of the GEN2PHEN Consortium  
(<http://www.gen2phen.org>)

Richard Cotton  
(cotton@unimelb.edu.au)  
on behalf of the HGVS  
(<http://www.hgvs.org/>)

**"An international collaborative project to integrate LSDB data with central repository and genome browser information"**

DATA EXCHANGE, LSDB TO BROWSER: Suggested core content

- 1) Contact details for PI(s) running the LSDB
- 2) Gene Name(s)/Symbol(s) [from HGNC] and utilised Reference Sequence(s)
- 3) Name and URL of the LSDB
- 4) URL syntax for linking to individual LSDB records
- 5) Core Content to exchange per variant
  - a) HGNC Gene Symbol (e.g. 'CAPN3')
  - b) Variant in HGVS nomenclature (e.g., c.-766A>G)
  - c) Variant ID (for direct URL linking to this record in your LSDB)
  - d) dbSNP ID (e.g., rs1801496) [if exists / optional]
  - e) OMIM ID (e.g., OMIM114240:0004) [if exists / optional]
  - f) PubMed ID for paper describing variant(s) [if exists / optional]

Notes:

- If a particular variant exists as multiple independent records in your LSDB, then submit those variants multiple times with their own specific linking information for point 5)c).
- Phenotype and pathogenicity data are not considered core information at this stage of the project as standards for this must first be agreed to avoid any risk of misinterpretation.

**Correspondence continued**

**Human Variome Project Planning Meeting**

We were delighted with the high profile attendance at the Human Variome Project planning meeting ([www.humanvariomeproject.org/HVP2008/](http://www.humanvariomeproject.org/HVP2008/)) held at Costa Brava, Spain, May 25-29, 2008. Equally impressive were the numerous collaborative exercises initiated and the juxtaposition of Saudi Clinical Geneticists and US informaticists, for example, promising a full global collaboration as 21 countries were represented. Key representatives of EBI, NCBI, NCI, Genetic Alliance, American College of Medical

Genetics and the Editor of Nature Genetics were present. A manuscript will be produced from the meeting for a high profile publication. Almost a majority of the Council of the inherited colon cancer consortium InSiGHT ([www.insight-group.org](http://www.insight-group.org)) were present and they held two meetings there. This group is so important because, in piloting the collection of mutations worldwide in the four genes which cause colon cancer for their own purposes, they have agreed this be a pilot for collection of mutation in all genes worldwide causing inherited disease. This activ-

ity will greatly simplify the task of the Human Variome Project participants.

*Richard Cotton  
Convenor  
Human Variome Project*



**Calendar of Meetings of Interest [all linked]**

[The 10th International Meeting on Human Genome Variation - 15th - 17th October, 2008, Toronto, Canada](#)

[HGVS Scientific & Annual General Meeting - 11th November 2008, Philadelphia, PA, USA \(ASHG 2008 satellite\)](#)

[Mutation Detection 2009: 10th International Symposium on Mutations in the Genome - 28 May - 1st June 2009, Paphos, Cyprus](#)

**Mutation Detection Symposium Cyprus 2009**

[6th HUGO Mutation Detection Training Course - 24th - 28th October 2008, Rotterdam, The Netherlands](#)

[1st Golden Helix Symposium: Copy number variation and genomic alterations in health and disease - 28-29 November, 2008 Athens, Greece](#)

[Human Variome Project Forum - 10th November 2008, Philadelphia, PA, USA \(ASHG 2008 satellite\)](#)

**Human Genome Variation Society Newsletter**

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The Society is an Affiliate of the International Federation of Human Genetics Societies ([IFHGS](#)) and also the Human Genome Organisation ([HUGO](#)).

Members of the Society have published a number of papers in relation to human genome variation and in particularly mutation databases, these are listed in the Publications link left.

The Society maintains a substantial collection of links to mutation databases (link left) and has made recommendations for nomenclature of variations, and content of mutation databases.

To join the Society and more information such as Board of Directors, Bylaws, Membership etc. see our website.

We're on the Web!  
[www.hgvs.org](http://www.hgvs.org)