



# THE HUMAN VARIOME PROJECT

## Human Variome Project Bulletin No. 13

Mar 08 - Jun 08

---

### 1. Major News:

The HVP Planning Meeting was held in San Feliu de Guixols, Costa Brava, Spain May 25-29 2008 ([www.humanvariomeproject.org/HVP2008/](http://www.humanvariomeproject.org/HVP2008/)). It attracted over 100 registrants and speakers and there were many supportive comments afterwards. Some highlights were as follows:-

1. Attendance of most key individuals was pleasing, many of whom had been at the Melbourne meeting and a large percentage of the HVP Planning Group was there. There were representatives of 21 countries and key bodies such as NCBI, EBI, RIKEN, OMIM, *Nature Genetics*, GEN2PHEN, InSiGHT, Genetic Alliance, KOBIC, Genome Commons, Nutrigenomics, NCI, *Human Mutation*, IARC, Sanger Centre, ACMG, and key universities worldwide. Last minute cancellations were from S. Brahmachari (CSIR-India), K. Stefansson (deCODE) and J. Solbakk (UNESCO). Many clinical geneticists were present.
  2. The mixing of many disparate nationalities was notable and the juxtaposition of US informaticists and Saudi clinical geneticists was noted as stunning.
  3. Conversations and presentations indicated that a massive number of new collaborations and understandings were being developed.
  4. The impact of the InSiGHT/HVP collaboration was dramatic. 10-15 members were present and they had two separate meetings and developed minutes and a roadmap and linked up with those where expertise was needed. This initiative as a model for all genes in collecting mutations and phenotype from their four genes worldwide is proving to be highly stimulating to mutation collection strategies.
  5. Currently we are writing a manuscript with a general piece for publication in a major journal with detail in supplementary material on the web with all participants as authors.
  6. Major discussions included semantics regarding naming, project coverage and governance.
  7. The cooperation of NCBI and EBI was still visible and a great highlight.
  8. The excellent EC funding of the GEN2PHEN project will be a great help for some informatics aspects of the project and they are fully collaborative, but with a European focus.
2. Follow up meetings to the Spain HVP planning meeting is being planned for November 2009 and a number of locations are being considered including Cyprus and Singapore. Also, specific topic meetings are planned in Japan, Korea and Australia to discuss local data collection and methods of doing it.

3. HVP Forums are planned on September 27 in Hyderabad, India as a satellite to HUGO's HGM and on November 10 in Philadelphia as a satellite of the Annual American Society of Human Genetics meeting and associated with the HGVS meeting on November 11. These are not just further meetings on the crowded international schedule, but information meetings where speakers and attendees at the main meeting can contribute and be informed. Speakers and attendees are being sought from those attending both conferences. Volunteers welcome.
4. Conference of interest; The Seventh Asia-Pacific Bioinformatics Conference (APBC2009), Beijing, China, 13-16 January 2009 (<http://bioinfo.au.tsinghua.edu.cn/apbc2009/>).
5. Leiden scientists sequence first female DNA. More information from the Leiden University Medical Centre (LUMC) can be found here [http://www.nwo.nl/nwohome.nsf/pages/NWOA\\_7EZAY6\\_Eng?Opendocument](http://www.nwo.nl/nwohome.nsf/pages/NWOA_7EZAY6_Eng?Opendocument). We are pleased our energetic colleague Johan den Dunnen was the major participant showing such sequencing is possible for a diagnostic lab with the current equipment.
6. Papers of interest:
  1. Chen, Y. H., C. K. Liu, et al. (2008). "GenoWatch: a disease gene mining browser for association study." Nucleic Acids Res.
  2. Forbes, S. A., G. Bhamra, et al. (2008). "The Catalogue of Somatic Mutations in Cancer (COSMIC)." Curr Protoc Hum Genet **Chapter 10**: Unit 10 11.
  3. Richards, C. S., S. Bale, et al. (2008). "ACMG recommendations for standards for interpretation and reporting of sequence variations: Revisions 2007." Genet Med **10**(4): 294-300.
  4. Estivill, X., et al., *SNPs meet CNVs in genome-wide association studies: HGV2007 meeting report*. PLoS Genet, 2008. **4**(4): p. e1000068.
  5. Smith, T.D. and R.G. Cotton, *VariVis: a visualisation toolkit for variation databases*. BMC Bioinformatics, 2008. **9**: p. 206.
  6. George, R.A., et al., *Response to Stenson et al on the review of general mutation databases*. J Med Genet, 2008. **45**(5): p. 319-20.
  7. Hu, J. and C. Yan, *Identification of deleterious non-synonymous single nucleotide polymorphisms using sequence-derived information*. BMC Bioinformatics, 2008. **9**(1): p. 297.
  8. Houdayer, C., et al., *Evaluation of In Silico Splice Tools for Decision-Making in Molecular Diagnosis*. Hum Mutat, 2008. **29**(7): p. 975-982.

---

Contact:

Prof. Richard G.H. Cotton

Convenor, The Human Variome Project

Tel: +613 8344 1893

Fax: +613 9347 6842

Email: [cotton@unimelb.edu.au](mailto:cotton@unimelb.edu.au)