



## THE HUMAN VARIOME PROJECT

### COLLECTION OF HUMAN GENE VARIATION

Thursday 25<sup>th</sup> October 2007  
6.30 pm – 8.30 pm  
Room 32A, San Diego Convention Center  
San Diego, CA, USA

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The Human Variome Project was developed to give a unified voice and direction to those affected by and dealing with those with inherited disease and studies of variation in the human genome and their effects. This was done with the expectation of obtaining better funding for the field divided between thousands of faulty genes and improvement of genetic healthcare worldwide.

The Human Variome Project ([www.humanvariomeproject.org](http://www.humanvariomeproject.org)) was initiated in June 2006 out of the activities of the members of the HUGO Mutation Database Initiative and the Human Genome Variation Society ([www.hgvs.org](http://www.hgvs.org)). The main thrust of the activity is ultimately for collection of all variation in the human genome and its associated effects and to make it available publicly. This is an ambitious but essential activity and will probably require the collaboration of perhaps thousands of individuals worldwide as well as substantial funding.

**The vision** Anyone – patients, clinicians, researchers – will be able to determine reliably and quickly which mutations are causing disease. This will lead to improved genetic healthcare and research around the world.

**How?** The project will collect global information on all human genetic variation and how this variation affects health.

**The result** A curated public catalogue of variation for each of our 20,000 genes and their associated phenotypes/studies. It will have an automated **system** of submission and review so that everyone can have confidence in the information.

#### **Important questions to be answered by the Human Variome Project**

- Has the mutation in my patient been found before?
- If it has, what effect did it have on the patient?
- What are the most common mutations so a cheap test can be developed for them?
- What variation in symptoms occurs in 20 or so patients with the same mutation?
- What should parents be told regarding the quality of life for their children with this mutation?

The work can not be done in the high throughput mode, but protocols can and need to be developed to minimise the time to collect genotype and phenotype data and we can encourage this collection. With increased “medical re-sequencing”, gene or locus specific databases (LSDBs) are becoming more vital.

Thus, thousands or tens of thousands of people, new protocols and collaborators are needed to document the 100+ expected deleterious mutations in each of the 20,000 genes, which affect human health.

**We need the support and backing of as many individuals as possible. Thus, we would like for you to attend this meeting to allow you to be informed and learn how you can help the project achieve its goals.**

#### **References**

- Cotton, R.G. & Kazazian, H.H., Jr. Toward a human variome project. *Hum Mutat* **26**, 499 (2005).
- Patrinou, G.P. & Brookes, A.J. DNA, diseases and databases: disastrously deficient. *Trends Genet* **21**, 333-8 (2005).
- What is the Human Variome Project? *Nat Genet* **39**, 423 (2007).

Horaitis, O., Talbot, C.C., Jr., Phommarinh, M., Phillips, K.M. & Cotton, R.G. A database of locus-specific databases. *Nat Genet* **39**, 425 (2007).  
Cotton, R.G. et al. Recommendations of the 2006 Human Variome Project meeting. *Nat Genet* **39**, 433-6 (2007)

## PROGRAMME

<b>6.15</b>	<b>Registration of name and contacts for future mailings</b>
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<b>6.30 – 6.45</b>	<b>R. Cotton – Introduction to the Human Variome Project: Rationale and Current status</b>
<b>6.45 – 7.00</b>	<b>M. Axton – Microattribution</b>
<b>7.00 - 7.15</b>	<b>J. Burn – InSiGHT/Human Variome Project pilot project for collection of inherited colon cancer data</b>
<b>7.15 – 7.30</b>	<b>G. Pastores – A model for data collection worldwide; the MSPI experience</b>
<b>7.30 – 7.45</b>	<b>S. Richards – Lab perspective/ACMG perspective</b>
<b>7.45 – 8.00</b>	<b>D. Maglott – Variation collection at NCBI</b>
<b>8.00 – 8.30</b>	<b>General Discussion &amp; Closing Remarks</b>
<b>8:30</b>	<b>Meeting closes</b>

Those present representing Human Variome Project Working Groups will be asked to say a few words about their aims and intentions at the start of the General Discussion.

### **SPEAKERS**

Professor Richard G.H. Cotton, Head, Genomic Disorders Research Centre, Melbourne, Australia; Convenor, the Human Variome Project; Co-Editor, *Human Mutation*; President, Human Genome Variation Society (HGVS)

Dr. Myles Axton, Editor, *Nature Genetics*, New York, USA

Professor John Burn, Medical Director and Head of Institute, Institute of Human Genetics, International Centre for Life, Newcastle upon Tyne, United Kingdom

A/Prof. Gregory M. Pastores, Associate Professor of Neurology and Pediatrics, Director Neurogenetics Laboratory, Departments of Neurology (Genetics) and Pediatrics, NYU Medical Center, New York, USA

Professor (Carolyn) Sue Richards, Molecular & Medical Genetics, Director OHSU DNA Diagnostic Lab, Oregon Health & Science University

Dr. Donna Maglott, Curator OMIM, NCBI, Baltimore, USA

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