



THE HUMAN VARIOME PROJECT

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1. The international colon cancer group (InSiGHT; www.insight-group.org) has developed a plan to collect variations to their databases to better serve their patients and research. It is also intended that this be a major pilot for the Human Variome Project.
2. Johan den Dunnen and Christophe Beroud held a highly successful and well attended Workshop (250-300 people) at the ESHG meeting in Nice (www.eshg.org/eshg2007). The topic was mutation nomenclature and locus specific databases.
3. Charles Scriver chaired a project report on the Human Variome Project at HUGO's HGM2007 in Montreal on May 24th. Those speaking were Dick Cotton, Tony Brookes, Johan den Dunnen, Ruth Chadwick and Bernard Brais. This was well attended despite being the last session of the meeting.
4. Dr. Tom Weber arranged a key meeting with the top officials of the New York State Health Department on May 25th, where the importance of collection of patient variation and chemical data was emphasized in any future planning.
5. Arleen Auerbach, Michael Katz (via phone), Tom Weber, Connie Talbot (via phone) and Dick Cotton met with Myles Axton to discuss further plans for publication of mutations.
6. Mike Watson, Mendel Tuchman, Hiroki Morizono, Connie Talbot, Dick Cotton and Michael Katz met with Francis Collins, Lisa Brooks and Mark Guyer to update the NHGRI on the Human Variome Project on May 29th and potential pilot projects were discussed.
7. Dick Cotton and Connie Talbot met Dr. Jim Herrington (Fogarty International Center) and others on May 29th and found several areas of common ground for further discussion.
8. Meetings and lobbying of the State and Federal Australian Governments continues with the energetic assistance of the CASS Foundation (David Abraham and Daniel Rechtman).

9. Ethics Working Group. There have been at least two papers on ethics in relation to LSDBs. Sue Povey, curator of the TSC LSDB and HUGO Trustee, has agreed to head the HVP Ethics Working Group to establish practical guidelines and will rely on community and ethical input.

10. Publications. The last Newsletter indicated four articles related to the Human Variome Project in the April issue of Nature Genetics.

- 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).
- Gout, A.M. et al. Analysis of published PKD1 gene sequence variants. *Nat Genet* 39, 427-8 (2007).
- Cotton, R.G. et al. Recommendations of the 2006 Human Variome Project meeting. *Nat Genet* 39, 433-6 (2007).

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