



## THE HUMAN VARIOME PROJECT

### Human Variome Project Bulletin No. 10

June – July 2007

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1. The Genomic Disorders Research Centre has reserved a room for two hours from 6:30 pm – 8:30 pm on Thursday October 25<sup>th</sup> at the San Diego ASHG Meeting, to discuss the Human Variome Project. The Planning Group will be consulted as to how to use the time but in the mean time, ideas please.
2. Interest has been expressed from the German Society of Human Genetics in joining the Human Variome Project effort.
3. Professor Utsoumiya has reported that he has received agreement on support for the Human Variome Project activities from the Japanese Ministry of Education, Culture, Sport, Science and Technology (MEXT).
4. Johan den Dunnen and Dick Cotton were invited to attend a meeting at the International Agency for Research on Cancer (IARC) in Lyon July 9-10. This involved teaching for one day and discussing strategy for databasing somatic mutation in cancer, the next step is a discussion paper for *Human Mutation*. Representatives of COSMIC and NIH were present and discussions were lead by Magali Olivier and Pierre Hainaut.
5. The Human Variome Project will get further exposure in Dubai at the Second Pan Arab Human Genetics Conference Meeting in November.
6. Two outline pilot projects have been put to Francis Collins (NHGRI/NIH). One on the international colon cancer group, InSiGHT, as a model study for data flow as a model for the Human Variome Project and another to submit LSDB content to NCBI.
7. A debate has been initiated about what LSDB content should be uploaded to central databases/browsers and under what conditions.
8. Professor Charles Rotimi would like our readership's attention drawn to the African Society of Human Genetics Meeting to be held in Cairo, November 3-5, 2007 (see [www.afshg.org](http://www.afshg.org)).

9. Papers of interest:

- Giardine, B. et al. PhenCode: connecting ENCODE data with mutations and phenotype. *Hum Mutat* **28**, 554-62 (2007).
- Chan, P.A. et al. Interpreting missense variants: comparing computational methods in human disease genes CDKN2A, MLH1, MSH2, MECP2, and tyrosine (TYR). *Hum Mutat* **28**, 683-93 (2007).
- van Baal, S. et al. FINDbase: a relational database recording frequencies of genetic defects leading to inherited disorders worldwide. *Nucleic Acids Res* **35**, D690-5 (2007).
- Woods, M.O. et al. A new variant database for mismatch repair genes associated with Lynch syndrome. *Hum Mutat* **28**, 669-73 (2007).

10. Please send in any relevant contribution for the next Bulletin.

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