



THE HUMAN VARIOME PROJECT

Collection of Human Gene Variation Affecting Human Health

Monday 19th October 2009

8:30 AM - 5.30 PM

Coral Ballroom, Mid-Pacific Conference Centre
Hilton Hawaiian Village, Honolulu, Hawaii

7.30 – 8.30 **Registration**

8.30 – 8.35 **Welcome**
Richard G.H. Cotton / María-Jesús Sobrido

Session 1 Chair: Lars Bertram

8.35 – 8.50 **Richard G. H. Cotton**
The Human Variome Project - introduction

8.50 – 9.05 **Mike Woods**
Insights into InSiGHT and the HVP experience

9.05 – 9.20 **María-Jesús Sobrido**
LSDBs for Neurological Disorders: Is there a rationale for a Neurogenetics Database Consortium?

9.20 – 10.15 **Discussion**

10.15 – 10.45 Coffee Break

Session 2 Chair: María-Jesús Sobrido

10.45 – 11.00 **Andrea Haworth**
Everyday problems for a diagnostic Neurogenetics Laboratory

11.00 – 11.15 **John K. Fink**
Hereditary motor neuron disorders: genetic heterogeneity, genetic pleiotropy, and clinical overlap

11.15 – 11.30 **Sherifa Hamed**
Genetic Disorders among Egyptian Population: Experience from Epidemiologic & Molecular Data

11.30 – 12.30 **Discussion**

12.30 – 13.30 Lunch

Session 3**Chair: John K. Fink**

13.30 – 13.45

Antonio Salas*Guidelines towards a MITOvariome project*

13.45 – 14.00

Dianne W. Cox*Wilson disease Database: an Aid to DNA Diagnosis*

14.00 – 14.15

Birgitt Schuele*Update of the Parkinson's Disease Mutation Database*

14.15 – 14.30

Matthew J. Farrer*Who appropriated the PARK loci, and where were the geneticists?*

14.30 – 15.30

Discussion**15.30 – 16.00****Coffee Break****Session 4****Chair: Matthew J. Farrer**

16.00 – 16.15

Lars Bertram*Online encyclopedias and field synopses of genetic association studies in Alzheimer's and Parkinson's disease*

16.15 – 16.30

Marc Cruts*The Alzheimer Disease & Frontotemporal Dementia Mutation Database*

16.30 – 17.00

Discussion

17.00 – 17.25

Summary, Resolutions and Plans

María-Jesús Sobrido

17.25 – 17.30

Closing remarks

Richard G. H. Cotton

17.30**Meeting Ends**