



THE HUMAN VARIOME PROJECT

Collection of Human Gene Variation Affecting Human Health

Friday 22nd May 2009

8:30 AM - 5.00 PM

Room Linz

Hilton Vienna Danube

DRAFT PROGRAMME – SUBJECT TO CHANGES

7.30 – 8.30	Registration
8.30 – 8.35	Welcome Richard G.H. Cotton/Johan T. den Dunnen
Session 1	Chair: Myles Axton
8.35 – 8.55	Johan T. den Dunnen The mutation nomenclature standard
8.55 – 9.15	Mauno Vihinen Status of variation ontologies standards
9.15 – 9.35	TBA
9.35 – 9.55	Raymond Dalgleish The Locus Reference Genomic (LRG) DNA sequence format for LSDBs
9.55 – 10.30	Discussion
10.30 – 11.00	<i>Coffee Break</i>
Session 2	Chair: Mauno Vihinen
11.00 – 11.20	Peter Robinson Towards a Standard for Phenotype Ontology for Inherited Disease
11.20 – 11.40	TBC Towards a standard for description of morphology

11.40 – 12.00	Rolf Sijmons Towards standards in clinical documentation
12.00 – 12.20	TBA
12.20 – 13.00	Discussion
13.00 – 14.00	<i>Lunch</i>
Session 3	Chair: Johan T. den Dunnen
14.00 – 14.20	TBA Quality control standards relevant to databasing
14.20 – 14.40	Myles Axton Standards in variation reporting and attribution
14.40 – 15.00	Robert Hoffman Wiki genes- Towards standards in microattribution
15.00 – 15.20	Christophe Beroud Databases and current law
15.20 – 15.45	Discussion
15.45 – 16.15	<i>Coffee Break</i>
Session 4	Chair: TBA
16.15 – 16.35	Sean Tavtigian Towards standards for unclassified variants
16.35 – 16.55	Sylvie Tuffery-Giraud Towards a standards for gene specific collection in a country – DMD in France
16.55 – 17.15	Sue Povey Towards a standard in ethics of variation databasing
17.15 - 17.45	Discussion & Summary
	Meeting Ends

TBA = To be announced

TBC = To be confirmed