



THE HUMAN VARIOME PROJECT

Collection of Human Gene Variation Affecting Human Health

Monday 10th May 2010
8:30 AM – 4.30 PM
Salle VII, UNESCO Headquarters
Paris, France

8.30 – 9.00

Registration

9.00 – 9.15

Welcome and Update from 1st consortium meeting
María-Jesús Sobrido

Session 1

Chair: María-Jesús Sobrido

9.15 – 9.30

Francisc Palau
A proposal for a neurogenetics database in Spain: the Charcot-Marie-Tooth Disease Initiative

9.30 – 9.45

Paola Carrera
Acting locally, thinking globally: development of an integrated international curated database for collection and management of clinical and genetic data in hereditary spastic paraplegia (HSP)

9.45 – 10.00

Marc Cruts
Title TBC

10.00 – 10.30

Discussion Topic – Linking and Integration with other databases

Non attendee abstracts

Direct Patient Submission of Longitudinal Phenotype Data
Dennis R. Johnson and Rebecca Sutphen

10.30 – 11.00

Coffee Break

Session 2

Chair: Andrea Haworth

11.00 – 11.15

Dipa Raja Rayan
Skeletal Muscle Channelopathies – Utilising databases in clinical practice

11.15 – 11.30

Henry Houlden
Clinical and genetic heterogeneity in hereditary sensory neuropathy

11.30 – 11.45

Mary Sweeney
Mitochondrial mutations: what do we need to know?

11.45 – 12.00

Joanna Elson

Mitochondrial mutations: What changes cause disease, which can be inherited?

12.00 – 12.15 Speaker TBA
Dystonia topic Title TBA

12.15 – 13.30 **Discussion Topic – Funding of the HVP Neurogenetics Consortium**

13.30 – 14.30 *Lunch*

Session 4 **Chair: TBA**

14.30 – 14.45 Johan T. den Dunnen
Developments in gene variant database software - the LOVD platform

14.45 – 15.00 Corey D. Braastad
Challenges with Real-Time Variant Investigation and Reporting in a private Clinical Molecular Genetics laboratory

15.00 – 15.15 Peter Robinson
Human Phenotype Ontology: Introduction and Applications in Neurogenetics

15.15 – 15.45 **Discussion Topic – Further progress towards a consortium**

15.45 – 16.10 **Summary, Resolutions and Plans**
Andrea Haworth

16.10 – 16.15 **Closing remarks**
María-Jesús Sobrido

16.15 **Meeting Ends**