



mtDNA variation, disease and databases

11th June 2012
UNESCO, Paris

PROGRAMME

Monday 11th June

- 8.30 – 9.00** **Registration outside security entrance**
UNESCO, 7 Place de Fontenoy
- 9.00 – 9.30** **Aims and Scope of the proposed database**
J. Elson (Newcastle, UK)
M. Sweeney (London, UK)
V. Procaccio (Angers, FR)
D.C. Wallace (Philadelphia, USA)
- 9.30 – 10.00** **InSiGHT: A pilot project on colon cancer mutation database:
Example of a successful database and consortium**
Finlay Macrae (Melbourne, AUS)
- 10.00 – 10.30** **Overview of the different mitochondrial databases: Mitomap,
Mitomaster, mtDB**
D. Wallace (Philadelphia, USA)-M. Lott (Philadelphia, USA) V.
Procaccio (Angers, FR)

10.30 – 11.00

Coffee Break

11.00 – 11.45 **Database Fields Clinical and Genetics data**
R. Pitceathly (London, UK) - M. Sweeney (London, UK)

11.45 – 12.30 **Assessing Mitochondrial Pathogenicity**
J. Elson (Newcastle, UK)

- Score of pathogenicity
- Assessing mitochondrial pathogenicity through functional tests

12.30 – 13.30 *Lunch – cafeteria 7th floor*

13.30 – 13.45 **Example of a mitochondrial LOVD database: A locus-specific database for mutations in GDAP1 allows analysis of genotype-phenotype correlations in Charcot-Marie-Tooth**
M. Ferre (Angers, FR)

13.50 – 14.40 **Work Flow and database organization, data collection, entry and access of data, database maintenance: Defining a curating committee**
R Ekong (London, UK)

14.45 – 15.15 **Ethical and Legal aspects: Ethics committee, informed consent**
M.-J. Sobrido (Santiago de Compostel, SP)

15.15 – 15.45 *Coffee Break*

15.45 – 16.15 **Funding Options**
V. Procaccio (Angers, FR)

16.15 – 17.30 Round table: J. Elson, M. Sweeney, V. Procaccio, D.C. Wallace
Review of key issues, Summary, Next steps and Conclusions

END OF MEETING