

- 8:00 - 8:55**      *Registration, Coffee*
- Speakers to give presentations to computer operator**
- 9:00 – 9:10**      **Prof. Richard G. H. Cotton**  
President's Welcome & brief report on projects of HGVS collaborators
- SESSION I: Chair: Dr. Alastair F. Brown**
- 9:10 – 9:35**      **Graham Taylor**  
A new online mutation repository for UK diagnostic laboratories
- 9:35 – 10:00**      **Thierry Soussi**  
Reassessment of the p53 mutation database in human disease by data mining with a library of p53 missense mutations
- 10:00 – 10:25**      **L. Roewer**  
The Y chromosome STR haplotype reference database (YHRD)  
an online repository of worldwide collected population samples
- 10:25 – 10:50**      **Anthony J. Brookes**  
A Phenotype Dimension to HGvbase
- 10:50 – 11:20**      **Coffee Break**
- SESSION II: Chair: Prof. Anthony J. Brookes**
- 11:20 – 11:45**      **Christoph Gille**  
Structural interpretation of mutations and SNPs in proteins using STRAP
- 11:45 – 12:10**      **Roger Horton**  
The MHC Haplotype project: Complete MHC Haplotype sequencing for SNP identification: Results from the first two Haplotypes
- 12:10 – 12:35**      **Peter Ahnert**  
Application of the GENOLINK<sup>(TM)</sup> Genotyping System in a Candidate Gene Association Study in Rheumatoid Arthritis
- 12:35 – 13:00**      **Damian Labuda**  
Genetic Variability In Gene Segments Upstream Of Coding Regions
- 13:00 – 14:00**      **Lunch**

**SESSION III: Chair: Dr. Christophe Beroud**

- 14:00 – 14:25**      **Pablo Marín-García**  
MutRes and LsdbRes: extracting mutations to a central database
- 14:25 – 14:50**      **Marcella Attimonelli**  
Statistical Prediction of Pathogenic Variant Sites In Human Mitochondrial Genomes
- 14:50– 15:15**      **Antonio Cavallo**  
Mapping SNPs and Locus specific Mutations to Protein Sequence Structure Data
- 15:15 – 15:25**      **Sigrid Beiboer**  
Rapid genotyping of blood group antigens using multiplex PCR and DNA microarray
- 15:25 – 15:45**      ***Coffee Break***

**SESSION IV: Chair: Dr. A. Jamie Cuticchia**

- 15:45 – 16:10**      **Ian Day**  
Identification of 'forme frustes' and 'paucimorphisms' by population mutation scanning by meltMADGE: proof-of-principle using LDLR gene
- 16:10 – 16:20**      **J. Smith**  
Expression cloning of T4 endonuclease VII and development of a MADGE-based heteroduplex cleavage protocol for economical high throughput mutation scanning
- 16:20 – 16:30**      **K. Alharbi**  
Definition of population 'reference range' for sequence diversity of MC4R gene using meltMADGE: two 'paucimorphisms,' occasional 'private' mutations and anthropometric consequences
- 16:30 – 16:55**      **H. Atashi Shirazi**  
Mutational Analysis of ATM Gene within Iranian Patients with Ataxia Telangiectasia
- 17:00**                **MEETING ENDS**