

HGVS SCIENTIFIC MEETING

“How to Explore Human Genotype to Phenotype Relationships”

Room ‘Mar’
Hotel Vincci Maritimo
Barcelona, Spain

31st May 2008

PROGRAMME 2nd May '08 – Schedule subject to change

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|------------------|---|
| 7.00 – 7.45 | Speakers arrive and hand in presentations (or have emailed them) |
| 7.15 – 8.00 | Registration |
| 8.00 – 8.05 | President’s Welcome Richard G. H. Cotton |
| Session I | Session I Chair: Anthony J. Brookes |
| 8.05 – 8.35 | Stephan Beck Reverse Phenotyping: Towards an integrated (epi)genomic approach to complex phenotypes and common disease |
| 8.35 – 9.05 | John Burn DNA diagnostics: the lessons of the urban motorway |
| 9.05 – 9.25 | Bruce Gottlieb When Unique Locus-Specific Disease Genotypes Are Not Associated With Identical Phenotypes - Is It Time To Consider A New Genetic Hypothesis? |
| 9.25 – 9.45 | Steven Brenner The Genome Commons and The Genome Commons Navigator |
| 9.45 – 10.05 | Peter Rogan Common splicing mutations detected by individual information analysis of single nucleotide polymorphisms |
| 10.05 – 10.35 | COMPANY LECTURE Illumina Inc. Debora Bailey Optimizing genotyping arrays for genome-wide associated studies and |

focused replication studies

10.35 – 11.00

Coffee Break & Posters - Sponsored by SAGE Publications

Session II

Session II

Chair: Conover Talbot Jr.

11.00 – 11.30

Michael Stratton

Systematic resequencing of the coding exons of the X chromosome in X-linked Mental Retardation

11.30 – 12.00

John Rioux

Genome-wide association studies in inflammatory diseases - taking stock of what we have learned thus far

12.00 – 12.20

Paul Flicek

The European Genotype Archive

12.20 – 12.40

Anthony Brookes

Summary, Software, and Speculation

12.40 – 13.10

COMPANY LECTURE

SAGE Publications

Human Genomics to Proteomics: Open Access Journal

George P. Patrinos

National/Ethnic Mutation Databases: Recording Populations' Genography

13.10 – 14.10

Lunch - Sponsored by SAGE Publications

14.10 – 14.40

Poster Session

Session III

Session III

Chair: Richard G.H. Cotton

14.40 – 14.55

COMPANY LECTURE

Affymetrix

Dirk Juergensen

A genome-wide association platform designed with copy number in mind

14.55 – 15.10

Peter Propping

The German HNPCC consortium: structure, aims and accomplishments

15.10 – 15.25

Tom LaFramboise

A genomic survey of allele-specific selection in lung cancer amplicons

- 15.25 – 15.40** **Paul Schofield**
An informatics approach to relating human and mouse phenotypes and diseases
- 15.40 – 15.55** **Timothy Smith**
Varivis: A Visualisation Toolkit For Variation Databases
- 15.55 – 16.00** **Concluding remarks**
Meeting Ends so delegates may attend ESHG Plenary Session

Poster Presentations

- 1 Bernd Auber**
An unusually large 5Mb deletion on chromosome 21q21.2-q21.3 in a German family: implications of the deletion for a family member with a Silver-Russel phenotype and mental retardation
- 2 Arleen D. Auerbach**
Phenotyping genetic diseases using an extension of μ -scores for multivariate data.
- 3 Bibhuranjan R. Das**
Lack of association between the Arg753Gln single nucleotide polymorphism in the TLR2 gene and Tuberculosis risk
- 4 Yoon Shin-Cho**
Genome-wide association study of 10000 Koreans from epidemiological cohorts to identify genetic factors determining the individual difference in anthropometry
- 5 Yeun-Jun Chung**
Copy number variations associated with rheumatoid arthritis in Korean population
- 6 Yeun-Jun Chung**
Copy number variations associated with autism spectrum disorder in Korean population
- 7 Richard G.H. Cotton**
Pilot studies of the Human Variome Project
- 8 Andrew D. Devereau**
DMuDB – the UK Diagnostic Mutation Database: collecting genotype and phenotype data within the UK National Health Service

- 9 Anne Friedrich**
MS2PH-db: toward the integration of protein structural behaviour to study phenotypic impacts of mutations
- 10 Ming Qi**
Bio-bank and genetic database of major diseases in China
- 11 Judy Savige**
The use of a COL4A3-5 gene map in the analysis of genotype-phenotype relationships in Alport and related syndromes
- 12 Paul N. Schofield**
An informatics approach to relating human and mouse phenotypes and diseases.
- 13 Rolf H. Sijmons**
Clinical interpretation of mismatch repair gene unclassified variants: the Dutch study
- 14 Thomas K. Weber**
A Q324H Germline Sequence Variant of the MutY Homolog (MYH) Gene is Associated with Increased Risk of Early Age Onset Colorectal Polyp Formation independent of MYH Y165C and G382D mutation status and is Characterized by Decreased Enzymatic Activity *In Vitro*
- 15 Thomas K. Weber**
The "InSiGHT Panel" Provides a Novel Tool to Facilitate a Resource Conserving Analysis of the POPULATION Frequency of Pathogenic MMR Alleles and Their Contribution to the HNPCC Phenotype
- 16 Bertram Weiss**
Using Phenotype for Gene Function Prediction