

## HGVS Scientific Meeting - Programme

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- 8:00**                      **Registration & Coffee**
- 8:30 – 8:35**                **President's Welcome**  
Richard G. H. Cotton
- SESSION I: Chair: Arleen Auerbach**
- 8:35 – 8:50**                **Marc S. Greenblatt**  
CDKN2a Database Project: Integrating Allelic Variants with Evolution, Structure, Function, and Disease-Associated Mutation
- 8:50 – 9:05**                **Anna-Kaisa Anttonen**  
findis.org – Finnish Disease Database
- 9:05 – 9:20**                **Mauno Vihinen**  
How to curate 100 LSDBs
- 9:20 – 9:35**                **Christophe Beroud**  
The UMD software to Create Locus Specific Databases
- 9:35 – 9:50**                **Christophe Beroud**  
The UMD Central System: A New Tool to Query Simultaneously Multiple UMD Locus Specific Databases
- 9:50 – 10:05**              **Bruce Gottlieb**  
Locus Specific Databases in the 21<sup>st</sup> Century
- 10:05 – 10:15**              **Alastair F. Brown**  
LSDB in a Box – Status Report
- 10:15 – 10:25**              **Steve Sherry**  
Developments in dbSNP for 2003
- 10:25 – 10:55**              **Coffee Break**
- SESSION II: Chair: Nobuyoshi Shimizu**
- 10:55 – 11:25**              **INVITED SPEAKER**  
**Prakash M. Nadkarni**  
Database Representation of Phenotype: Research Issues

- 11:25 – 11:40**      **A. Jamie Cuticchia**  
Reinventing the GDB Human Genome Database
- 11:40 – 11:55**      **Heikki Lehvälaiho**  
MutRes and parsing LSDB data into HGVbase
- 11:55 – 12:10**      **Stefan White**  
Detecting copy number changes in genomic DNA using multi-colour multiplex ligation-dependent probe amplification
- 12:10 – 12:25**      **Nancy Carson**  
Field inversion gel electrophoresis (FIGE) for the analysis of the CCTG repeat in the ZNF9 gene causing myotonic dystrophy type 2
- 12:25 – 12:40**      **Marianna G. Yakubovskaya**  
Structural Polymorphism of DNA in Solution of Homologous Duplexes and Its Consequences for Heteroduplex Formation Procedure
- 12:40 – 12:55**      **Emma Ashton**  
An innovative protocol combining multiplex QF-PCR and fluorescent conformation sensitive capillary electrophoresis (FCSCE) for unified dosage analysis and point mutation screening
- 13:00 – 14:00**      ***Buffet Lunch in Heinsbergen room***
- SESSION III: Chair: Johan T. den Dunnen**
- 14:00 – 14:10**      **Saeed Teebi**  
The Genome Variation WayStation Now Open for Submissions
- 14:10 – 14:25**      **Steve Callaghan**  
Current Mutation Database Quality
- 14:25 – 14:40**      **Peter S. White**  
Extraction of cancer-specific genomic variation from the biomedical literature
- 14:40 – 14:55**      **Trevor Clancy**  
Text mining of sequence variations and linking them to diseases, genes and proteins

**14:55 – 15:10**

**S. C. Yau**

Analysis of dystrophin mRNA show nonsense, splice and cryptic splice site mutations cause Becker muscular dystrophy

**15:10 – 15:25**

**Jonathan Liu**

Mutation Quantifier Software to Detect DNA Variants and to Measure Allele Frequency with DNA Pooled Samples by DNA Sequence Method

**15:25 – 15:40**

**Iris Grossman**

Autoimmunity Related Genes and their Evolutionary Fate as Reflected in Three Inbred Ethnic Groups

**15:40 – 16:10**

**Coffee Break**

**SESSION IV: Chair: Anthony Brookes**

**16:10 – 16:25**

**Shinsei Minoshima**

MutationView : An Integrated Knowledge-Base for Variation in Human Disease Genes - Intelligent Search System and Genome-wide Data Construction

**16:25 – 16:40**

**Frank H. Ruddle**

Williams Syndrome Animal Model

**16:40 – 16:55**

**Seyed Javad Mirhassani Moghaddam**

MEFV Gene Mutations in Iranian Patients with Familial Mediterranean Fever (FMF)

**SHORT PRESENTATIONS IN –LIEU OF POSTERS**

**16:55 – 17:00**

**Kyusang Lee**

An Automated Data storage and Analysis System for Diagnostic SNP Marker Discovery using High-throughput Genotyping

**17:00 – 17:05**

**Jian-Min Chen**

Rapid Detection of Genomic Rearrangements in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene by Quantitative Multiplex PCR Amplification of Short Fluorescent Fragments: Extensive Allelic Heterogeneity and Diverse Mutational Mechanisms

**17:05 – 17:10**

**Jian-Min Chen**

Two Novel Severe Mutations in the Pancreatic Secretory Trypsin Inhibitor Gene (*SPINK1*) Cause Familial or/and Hereditary Pancreatitis

**17:10 – 17:15**

**E. K. Khusnutdinova**

Title to be advised

**17:20 – 17:30**

***Short Break***

**17:30- 19:00**

**HGVS ANNUAL GENERAL MEETING**

**Agenda attached**

**Non-members welcome but are unable to vote**

**19:00 – 20:00**

***Mixer- all registrants for this meeting welcome***