

HGVS SCIENTIFIC MEETING

“Clinical Genetics & Human Genome Variation”

PROGRAMME

Rooms AB & CD, Hilton Garden Inn Centre City
Philadelphia, PA, USA

11th November 2008

Schedule Subject to small Change

7.45 - 8.30 *Speakers arrive and hand in presentations*
Poster Mounting
Registration

8.45 – 8.50 **President’s Welcome**
Richard G. H. Cotton

Session 1

CLINICAL GENETICS
Chair: Marc S. Greenblatt

8.50 – 9.20 **Invited Speaker**
William Isaacs
Advances in Prostate cancer genetics

9.20 – 9.50 **Invited Speaker**
Sharon Plon
Sequence variant classification and reporting: recommendations for improving the interpretation of cancer susceptibility genetic test results

9.50 – 10.05 **Ute Geigenmuller**
Systematic Scoring of Variant Significance for Mendelian Disorders

10.05 – 10.35 **COMPANY LECTURE**
Idaho Technologies Inc.
Jason McKinney
Mutant Allele Amplification Bias using Rapid Cycle-Real Time PCR and Hi-Res Melting[®] with LunaProbes[™] on the LightScanner 32[™]

10.35 – 11.00

Coffee Break & Poster Session

Poster presenters to stand by their poster

Odd number posters (1,3 etc.): 10.35 – 11.00

Session 11

CLINICAL GENETICS & GENERAL VARIATION

Chair: Christophe Beroud

11.00 – 11.30

Invited Speaker

James Hudziak

Genetic and Environmental Contributions to Human Behavior

11.30 – 11.45

Marc Greenblatt

TBA

11.45 - 12.00

Melissa Southey

Mutation scanning in the breast cancer susceptibility gene *PALB2*: are some female carriers at high risk of breast cancer?

12.00 – 12.15

Leiah Luoma

Predictive and functional analyses of variants in the Wilson Disease gene, *ATP7B*

12.15 – 12.30

Graham Taylor

Cost and performance of high throughput mutation detection in tumour suppressor genes using the Illumina GA II clonal sequencer

12.30 – 12.45

COMPANY LECTURE

Affymetrix

B. Kelley

Integrating SNPs, CNPs and rare CNVs with the Affymetrix SNP 6.0 platform

12.45 – 13.45

Lunch – to be served in the restaurant

13.45 – 14.15

Poster Session

Poster presenters to stand by their poster

Even number posters (2,4 etc.): 13.45 – 14.15

Session III

CLINICAL GENETICS & GENERAL VARIATION

Chair: Conover Talbot Jr.

14.15 – 14.45

Invited Speaker

Greg Feero

Health care providers and human genetic variation: between a rock and data overload

14.45 – 15.00

Giovanna Spinella

The Collaboration, Education and Test Translation (CETT) Program: A model for collecting genotype and phenotype data from clinical laboratories

15.00 – 15.15

Donna Maglott

Integrating Variation Information in NCBI's Databases

15.15 – 15.30

Marianna Yakubovskaya

Comparative reactivity of all mismatched and unpaired bases for interpretation of CCM results

15.30 – 15.45

Shinsei Minoshima

Construction of independent LSDBs using *MutationView* softwares, *MV4LSDB*

15.45 – 16.15

Coffee Break

Session IV

CLINICAL GENETICS & GENERAL VARIATION

Chair: Alastair F. Brown

16.15 – 16.30

Bruce Gottlieb

The clinical implications of human genome variation for locus-specific genetic databases

16.30 – 16.45

B. D'Haene

A deletion of 7,5 kb disrupting a distant non-coding RNA (ncRNA) *PISRT1* and eight highly conserved non-coding sequences (CNCs) upstream of *FOXL2* cause BPES

16.45 – 17.00 Philip Giampietro

Genetic variation in *IL6* and *LRP5* is associated with osteoporosis in the Marshfield Clinic Personalized Medicine Cohort: evidence for interaction with smoking

17.00 – 17.10 Concluding Remarks

17.10 – 17.20

Short Break

17.20 – 18.30 HGVS ANNUAL GENERAL MEETING

Non-members welcome

18.30 – 20.00

Mixer – all registrants welcome

Poster Presentations

Poster Number

Posters are displayed in Room CD

Authors will present their posters at the following times:

Odd numbers: 10.35 – 11.00

Even numbers: 13.45 – 14.15

- 1** Towards a web-based collection of genotype/phenotype data for Fanconi anemia
Arleen D. Auerbach
- 2** Two paths to central repositories: PhenCode and dbSNP
Belinda Giardine
- 3** Molecular studies in patients with OTC deficiency
Lenka Dvorakova
- 4** Investigation of BDNF polymorphisms with SSRI response in Major Depressive Disorder
Stephanie Hall

- 5 Novel pharmacogenomic markers associated with Paclitaxel resistance in NCI60 cancer cell lines
Irada Ibrahim-zada
- 6 The role of apoptotic genes in the resistance to anthracyclines-based chemotherapy in breast cancer
Irada Ibrahim-zada
- 7 The application of high-density genotyping microarrays in a rare autosomal recessive syndrome of neonatal diabetes
Hui-Qi Qu
- 8 Creating ad HOC metadata to compare clinical trial Datasets
Gregg Silk
- 9 Germline Alterations within MH1 and MH2 Domains of SMAD3 and SMAD4 and their Impact on Breast Cancer Susceptibility
E. Tram
- 10 ATP7B splice variants identified in patients with Wilson Disease: a minigene approach
Anna Wilson
- 11 Higher than Expected Frequency of a Third Mutant Allele in a Multiethnic Population Affected with Bardet-Biedl Syndrome
Gail Billingsley
- 12 Genome-wide identification of Ca ion transport regulator genes in breast and prostate cancer
A. Lindenmaier