HGVS SCIENTIFIC MEETING

NEW DNA SEQUENCING TECHNOLOGIES & HUMAN GENOME VARIATION

PROGRAMME

Ballroom 29, San Diego Convention Centre
San Diego, CA, USA

23rd October 2007

8.00	**Speakers arrive and hand in presentations** **Registration**
9.00 – 9.05	**President's Welcome** Richard G. H. Cotton
Session I	**NEW DNA SEQUENCING TECHNOLOGIES** **Chair: Johan T. den Dunnen**
9.05 – 9.15	**Introduction** Johan T. den Dunnen
9.15 - 9.45	**Speaker to be confirmed**
9.45 – 10.20	**Michael D. Rhodes** Applied Biosystems Use of SOLiDtm system in variation studies
10.20 – 10.55	**David R. Bentley** Illumina/Solexa Solexa sequencing technology
10.55 – 11.25	***Coffee Break***
Session II	**NEW DNA SEQUENCING TECHNOLOGIES (continued)** **Chair:**
11.25 – 12.00	**Michael Egholm** 454 Life Sciences/Roche Discovering & screening genetic variations within the human genome: from whole human genome sequencing to amplicon resequencing
12.00 – 12.20	**Bruce Gottlieb** Development of DNA Sequencing Techniques to Investigate the Presence of Multiple forms of Specific Genes, Including "Minority DNA Species", Within Tissues

12.20 – 12.40	**Shamil Sunyaev** Computational analysis of the prospect for unbiased identification of genes underlying human phenotypes by re-sequencing at phenotypic extremes
12.40 – 13.00	**Graham Taylor** Clonal sequencer evaluation: applications in re-sequencing
13.00 – 14.00	***Lunch***
Session III	**GENERAL VARIATION** **Chair:**
14.00 – 14.20	**Christophe Beroud** TREAT NMD patients' registries: an international initiative to develop LSDBs for neuromuscular diseases and clinical trials
14.20 – 14.40	**Susan M. Kenney** The Wilson disease database: disease causing vs normal ATP7B variants
14.40 – 15.00	**Kathleen Claes** High resolution melting analysis (HRM) for rapid and sensitive detection of mutations in *BRCA1&2*: comparison of Lightscanner™ (Idaho Technologies) and LightCycler 480 (Roche)
15.00 – 15.20	**Isaksson M** Selector Probes for multiplex targeted copy-number and sequence analyses
15.20 – 15.40	**Barend Mons** Wikiproteins – A web tool enabling community annotation of proteins and their (inter)actions
15.40 – 15.55	***Concluding remarks***
15.55 – 16.15	***Coffee Break***
16.15	**HGVS ANNUAL GENERAL MEETING** **Non-members welcome but are unable to vote**
17.30 –	***Mixer***

Poster Presentations

1. Fokkema I.F.A.C. Facilitating DNA diagnostics by collecting human disease gene variation using an open source LSDB-in-a-Box platform – LOVD 2.0
2. Giampietro P.F. Genetic variation in pattering genes associated with vertebral development
3. Belinda Giardine PhenCode: Linking Human Mutations and Phenotype
4. Shinsei Minoshima *MutationView*, A database for human disease-associated mutations: archiving and distribution of custom softwares
5. Nobuyoshi Shimizu A shotgun DNA sequencing method for mutation analysis of the complicated Filaggrin (FLG) gene