HGVS
HUMAN GENOME VARIATION SOCIETY

Pathogenicity Interpretation in the Age of Precision Medicine

Chesapeake Room
Holiday Inn Baltimore Inner Harbour
Baltimore, MD, USA
6th October 2015

7.30 – 8.15	Registration Speakers arrive and hand in presentations
8.15 – 8.20	**Introduction** Marc Greenblatt
Session 1	**Chair: Marc Greenblatt**
8.20 – 8.45	**Computational methods for pathogenicity assessment (*exact title TBA*)** Carlos Bustamante Stanford University, USA
8.45 – 9.10	**Towards increasing the clinical relevance of computational methods to predict the consequence of human genetic variation** Rachel Karchin Johns Hopkins, USA
9.10 – 9.25	Discussion
9.25 – 9.35	**Semi-supervised learning for clinical variant interpretation** Matthew D. Rasmussen Counsyl, South San Francisco, USA
9.35 – 9.40	**Findings from the Critical Assessment of Genome Interpretation, a community experiment to evaluate phenotype prediction** Steven Brenner University of California, Berkeley, CA, USA

9.40 – 9.50	**Diagnostic Role of Exome Sequencing in Immune Deficiency Disorders** Steven Brenner University of California, Berkeley, CA, USA
9.50 – 10.00	**The European Variation Archive: assessing data quality, integration with LOVD and addition of clinically relevant data** Garry Saunders EMBL-EBI, Wellcome Trust Genome Campus, Hinxton, Cambridge, UK
10.00 - 10.10	**Description Extractor: Automated HGVS-recommended sequence variant description** Peter Taschner Depts of Human Genetics and Molecular Epidemiology, LUMC, and LIACS, University Leiden3, Leiden, Nederland
10.10 – 10.20	**Rapid Fire Poster Presentations**
10.20 – 10.30	**COMPANY LECTURE** **Qiagen** **Title TBA**
10.30 – 11.00	**Qiagen Coffee Break & Poster Session**
Session 2	**Variant Nomenclature** **Chair: Steven Brenner**
11.00 – 11.15	**The HGVS recommendations to describe DNA, RNA and protein sequence** Johan T. den Dunnen
11.15 – 11.30	**Variant representation in a world of change** Deanna Church
11.30 – 11.45	**Working towards a combined ISCN and HGVS standard for the description of chromosome rearrangements** Jean McGowan-Jordan
11.45 – 12.40	**Evolution of nomenclature systems to create a standard that incorporates traditional HGVS nomenclature and genomic systems** Panel Discussion: • Johan T. den Dunnen (LUMC), Chair • Deanna Church (Personalis) • Fiona Cunningham (EBI) • Reece Hart (Invitae)

- Jean McGowan-Jordan (Chair, ISCN)

12.40 – 13.40	**Lunch & HGVS Annual General Meeting**
Session 3	**Chair: Christophe Beroud**
13.40 – 14.05	**Predicting the pathogenicity of genetic variants in the DNA double-strand break repair pathway** Harry Ostrer Albert Einstein College of Medicine, USA
14.05 – 14.30	**Functional assays for assessment of variants of uncertain significance (VUS) in breast and ovarian cancer predisposition genes** Fergus Couch Mayo Clinic, USA
14.30 – 14.45	Discussion
14.45 – 14.55	**Rare non-synonymous variations in the human ferroportin iron transporter gene (haemochromatosis type 4): the quest for causal mutations** Gérald Le Gac U1078, Brest, France
14.55 – 15.20	**Experimental methods for pathogenicity assessment (*exact title TBA*)** Nicholas Katsanis Duke University, USA
15.20 – 15.45	**Systematically identifying pathogenic human variants using yeast** Fritz Roth University of Toronto, Canada
15.45 – 15.55	**Discussion**
15.55 – 16.00	**Meeting Summary**
16.00	**MEETING END IN TIME FOR ASHG PLENARY**