25 Sept 2017

HGVS
HUMAN GENOME
VARIATION SOCIETY

Methods & Tools for Assessing the Impact of Genetic Variations

Lake Nona Room, Lobby Level
Hilton Orlando
Orlando, FL, USA

17th October 2017

(a satellite of ASHG)

8:00 - 8:55 Registration

8:55 - 9:00 Welcome

Session 1 **Moderator: Marc Greenblatt**

9:00 - 9:40 **KEYNOTE SPEAKER**

Prioritizing somatic variants

Mark Gerstein
Prof of Biomedical Informatics & Prof of Molecular Biophysics & Biochemistry, & of Computational Biology & Bioinformatics, Yale University, CT, USA

9:40 - 9:55 **Presentation from selected Abstract**

GRASP v3: an updated GWAS catalog and contrast to similar catalogs

Ben Rodriguez
Population Sciences Branch, Division of Intramural Research, National Heart, Lung and Blood Institute, 73 Mt Wayte Ave, Framingham, MA, USA

9:55 - 10:35 **KEYNOTE SPEAKER**

Predicting the pathogenicity of rare missense variants

Weiva Sieh
Senior Faculty, Population Health Science & Policy & Genetics and Genomic Sciences, Mt Sinai, NY, USA

10:35 - 10:50 **Presentation from selected Abstract**

MutPred2 enables probabilistic interpretations of pathogenicity and impact on protein structure and function

Vikas Pejaver
Department of Biomedical Informatics and Medical Education and the eScience Institute, Univ. of Washington, Seattle, WA, USA

10:50 - 11:20 **Coffee Break & Poster Session**

Session 2 **Moderator: Steven Brenner**

11:20 - 12:00 **KEYNOTE SPEAKER**

Predicting the impact of mutations on splicing signals

Christophe Béroud
Genetics and Bioinformatics, Aix-Marseille University, Marseille, France

12:00 - 12:15 **Presentation from selected Abstract**

Rethinking the 5 splice site algorithms used in clinical genomics

Gabe Rudy
Golden Helix, 203 Enterprise Blvd, Suite 1, Bozeman, MT 59718, USA

12:15 - 12:55 **KEYNOTE SPEAKER**

Evaluating the evaluation of cancer driver genes

Rachel Karchin
Institute for Computational Medicine, Johns Hopkins Biomedical Engineering, Baltimore, MD, USA

12:55 - 13:10	**Presentation from selected Abstract** **The Ensembl Variant Effect Predictor (VEP)** **Benjamin Moore** *European Molecular Biology Laboratory, European Bioinformatics Institute, Wellcome Genome Campus, Hinxton, UK*
13:10 - 14:15	**Lunch & HGVS Annual General Meeting**
Session 3	**Moderator: Christophe Béroud**
14:15- 14:55	**KEYNOTE SPEAKER** **Validating and calibrating computational and functional approaches in BRCA and MMR** **Sean Tavtigian** *Oncological Sciences, Huntsman Cancer Institute, Salt Lake City, UT, USA*
14:55 - 15:35	**KEYNOTE SPEAKER** **Predicting the molecular mechanisms of genetic disease for protein coding variants** **Predrag Radivojac** *Department of Computer Science and Informatics, Indiana University Bloomington, IN, USA*
15:35 - 15:50	**Presentation from selected Abstract** **Findings from CAGI, the Critical Assessment of Genome Interpretation, a community experiment to evaluate phenotype prediction** **Steven Brenner** *Department of Plant and Microbial Biology, University of California, Berkeley, CA 94720, USA*
15:50 - 16:00	Closing Remarks
16.00	**MEETING END (in time for ASHG Plenary @ 16.30)**