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)

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<td>8:00 - 8:55</td>
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**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*
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<td>9:55 - 10:35</td>
<td><strong>KEYNOTE SPEAKER</strong> Predicting the pathogenicity of rare missense variants</td>
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|              | Weiva Sieh<br>
|              | *Senior Faculty, Population Health Science & Policy & Genetics and Genomic Sciences, Mt Sinai, NY, USA* |
| 10:35 - 10:50| **Presentation from selected Abstract**<br>MutPred2 enables probabilistic interpretations of pathogenicity and impact on protein structure and function |
|              | Vikas Pejaver<br>
|              | *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**                                                  |
| 11:20 - 12:00| **KEYNOTE SPEAKER** Predicting the impact of mutations on splicing signals         |
|              | Christophe Béroud<br>
|              | *Genetics and Bioinformatics, Aix-Marseille University, Marseille, France*          |
| 12:00 - 12:15| **Presentation from selected Abstract**<br>Rethinking the 5 splice site algorithms used in clinical genomics |
|              | Gabe Rudy<br>
|              | *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<br>
|              | *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)