HUMAN GENOME VARIATION SOCIETY

Leveraging the Electronic Medical Record for Genomic Interpretation

Room 30CD, Upper Level
San Diego Convention Center
San Diego, CA, USA

16th October 2018

(a satellite of ASHG)

8:00 - 8:55	Registration
8:55 - 9:00	Welcome

Session 1	**Moderator: TBA**
9:00 - 9:45	**KEYNOTE SPEAKER** **EHR and Genomics to Find Mendelian Disease** Lisa Bastarche *Biomedical Informatics, Vanderbit University Medical Center*

9:45 - 10:00	**Presentation from selected Abstract** **Proactive variant effect maps for MTHFR and CBS accurately predict homocystinuria patient phenotypes and response to therapy** Song Sun *The Donnelly Centre and Departments of Molecular Genetics and Computer Science University of Toronto, Toronto, Ontario M5S 3E1, Canada & Lunenfeld-Tanenbaum Research Institute, Mount Sinai Hospital, Toronto Ontario M5G 1X5, Canada*
10:00 - 10:45	**KEYNOTE SPEAKER** **Phenotopic ontologies and clinical integration with CD2H, the Monarch Initiative and/or the NCATS Translator (*exact title to be advised)*** Melissa Haendel *Translational Data Science, Linus Pauling Institute, Oregon Health & Science University (OHSU)*
10:45 - 11:15	**Coffee Break**
Session 2	**Moderator: TBA**
11:15 - 12:00	**KEYNOTE SPEAKER** **Fast Healthcare Interoperability Resource** Atul Butte *UCSF School of Medicine Paediatrics*
12:00 - 12:15	**Presentation from selected Abstract** ***In Silico* Algorithms Vary in Pathogenicity Predictions For APC Missense Variants Reported in ClinVar** Alexander Karabachev *University of Vermont, Burlington VT*
12:15 - 12:30	TBA
12:30 - 14:00	**Lunch & HGVS Annual General Meeting**

Session 3	Moderator: TBA
14:00 - 14:45	**KEYNOTE SPEAKER** **Informatics & Phenotype** Marylyn Ritchie *Department of Genetics, Director, Center for Translational Bioinformatics, Institute for Biomedical Informatics (IBI), Associate Director for Bioinformatics, Institute for Biomedical Informatics (IBI), Associate Director, Center for Precision Medicine*
14:45 - 15:00	**Presentation from selected Abstract** **Identifying potential pleiotropy across cardiovascular and neurological diseases in the Electronic Medical Records and Genomics (eMERGE) network** Xinyuan Zhang *Genomics and Computational Biology Graduate Group, University of Pennsylvania, Philadelphia, PA*
15:00 - 15:45	**KEYNOTE SPEAKER** **Clinical Sequencing Exploratory Research (CSER)** Brian Shirts *Department of Laboratory Medicine, University of Washington Medical Center, Associate Director, Genetics Division, Laboratory Medicine, Assistant Director, Informatics Division, Laboratory Medicine*
15:45 - 16:00	**Closing Remarks**
16.00	**MEETING END (in time for ASHG Plenary @ 16.30)**