2018 Poster Talks: Two Concurrent Sessions

Session I (Room 6C): Clinical and Mendelian Genetics
Listed in order of presentation; order subject to change.

659F  Assessing the prevalence and clinical implications of secondary findings for hereditary cancer predisposition among patients with chronic kidney disease. E. Groopman

3145T  Overview of the NHLBI Trans-Omics for Precision Medicine (TOPMed) program: Whole-genome sequencing of >100,000 deeply phenotyped individuals. C. Laurie

2022W  Somatic activating mutations in MAP2K1 cause melorheostosis. J.C. Marini

2783F  Interpreting variants of uncertain significance using common variation in non-human primates. H. Gao

593T  Long-term impact of presymptomatic genetic testing for HD: Family narratives. J.M. Bollinger


428T  A de novo CNV deletion of the FOXF1 enhancer in 16q24.1 unmasks a rare noncoding SNP that likely prevented a lethal ACDMPV phenotype. P. Szafranski

1122W  Inherited peripheral neuropathies: Analysis of PDXK gene identifies a new treatable disorder. V. Chelban

1260W  Missense variants in PLS3 in patients with X-linked congenital diaphragmatic hernia. F. Petit

2959T  The complex genetic architecture of mitochondrial disease: Exploration of 1500 cases by whole exome sequencing. S.L. Stenton

3010T  Mate pair sequencing: Ushering cytogenetics into the era of personalized medicine. N. Hoppman

1774T  Returning unanticipated genomic results in a hospital-based research biobank. R.C. Green

533T  When is a disease too rare? A statistical framework for estimating clinical sensitivity for expanded carrier screening panels. R. Ben-Shachar

521T  Full-gene sequencing based expanded carrier screening of over 150 disorders. C. Gijavanekar
**Session II (Room 6D): Complex Traits and Statistical Genetics**

*Listed in order of presentation; order subject to change.*

1519T  Meta-analysis of vaginal microbiome data provides new insights into preterm birth. I. Kosti

1474T  Deep linear mixed models for structured, high-dimensional trait GWAS. F.P. Casale

742T  Increased penetrance of acute lymphoblastic leukemia susceptibility loci in children with Down syndrome. A.J. de Smith

2432F  Towards an understanding of the genetic architecture of the human cortex: Examining the effects of common variants on cortical thickness and surface area. S.E. Medland

2061W  Using *Xenopus laevis* as a model for studying genes associated with intellectual disabilities. M. Lasser

2012F  A limited set of transcriptional programs define major histological types and provide the molecular basis for a cellular taxonomy of the human body. A. Breschi

2769W  Mapping African ancestry among African Americans. C.C. Jones

432F  Global protein-protein and protein-DNA interactions of DNA topoisomerases. L. Uuskula-Reimand

1714T  Fine-mapping autoimmune disease variants in cytokine induced cell states. B. Soskic

3340T  Genome-wide association studies of brain structure and function in the ~20,000 UK Biobank participants. K.J. Sharp

3445T  Hepatic gene expression and DNA methylation associates with African ancestry: Uncovering the role of the genome in disease disparities in African Americans. C.S. Park