

ASHG 2023 NHLBI Research Presentations

Thursday, November 2

Keynote: Diversity, Equity, and Inclusion Luncheon

Gary H. Gibbons, M.D.

12:15 – 1:45 p.m.

Conv Ctr/Room 151AB/Level 1

Poster Presentation: 033 – Rare variant contribution to the heritability of coronary artery disease based on 22,443 whole genome sequences from the NHLBI TOPMed Program. [Board No. PB1756]

NHLBI TOPMed Atherosclerosis Working Group

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 039 – Expanding the mutational spectrum in elastin-driven disease through a gene-first approach. [Board No. PB4749]

Teresa Luperchio, Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 041 – Identification of an elastin long-range cis-regulatory element. [Board No. PB1144]

Sara Procknow, M.D., Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 045 – cellSTAAR: Incorporating single-cell-sequencing-based functional data to boost power in rare variant association testing of non-coding regions [Board No. PB4082]

NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 033 – Multi-omic integration uncovers biological pathways underlying HIV viral load [Board No. PB1678]

Brad Aouizerat, Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Post Presentations: 045 – The genetically regulated transcriptome better predicts plasma protein levels than the observed transcriptome and reveals mechanisms underlying autoimmune diseases [Board No. PB4388]

NHLBI TOPMed Consortium

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Post Presentation: 045 – Whole genome sequencing interaction study identifies loci with sex-specific effects underlying platelet aggregation traits [Board No. PB4430]

Ming-Huei Chen, Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Friday, November 3

Poster Presentation: 074 – Genetic predictors of Venous Thromboembolism in Europeans [Board No. PB1514]

Florian Thibord, Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 074 – Transcriptome-wide association study for frequent cocaine use identifies gene dysregulation in CD4+ T cells linked to immune response and cancer pathways. [Board No. PB1835]

Brad Aouizerat, Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 074 – Whole Genome Sequencing (WGS) Meta-analysis identifies Loci Associated with Non-alcoholic Fatty Liver Disease (NAFLD) across ancestries [Board No. PB1877]

NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 080 – Investigating the role of Rbfox2 in Splicing and the Transcriptional Network of the SWI/SNF Complex in myoblast fusion [Board No. PB4816]

Thirupugal Govindarajan, Ph.D. and Sachiyo Kawamoto, Ph.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 086 – Bayes factors for linear mixed models in genetic association tests [Board No. PB4068]

Yongtao Guan, Ph.D. and Daniel Levy, M.D.

3 - 5 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Saturday, November 4

Platform Presentation: 110 - Catalog and Genetic Architecture of Circulating Metabolites from Trans-Omics for Precision Medicine (TOPMed) Program

NHLBI Trans-Omics for Precision Medicine (TOPMed) Metabolomics Working Group

1:45 – 2 p.m.

Conv Ctr/Ballroom A/Level 3

Poster Presentation: 113 – Identifying individuals at risk for surgical supra-avalvular aortic stenosis in Williams-Beuren syndrome by polygenic score [Board No. PB1590]

Delong Liu, Ph.D.

2:15 - 4:15 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 113 – Quantifying individual-level mosaic chromosomal alteration fitness: genetic determinants and clinical consequences [Board No. PB1743]

NHLBI TOPMed Consortium

2:15 – 4:15 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 121 – Cross-cohort eQTL analyses of 6,602 multi-ancestry TOPMed whole blood RNA-seq samples uncover regulatory relationships [Board No. PB1065]

NHLBI TOPMed Consortium

2:15 – 4:15 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 122 – Proteomics of Urinary Exosomes Reveals Impaired Protein Trafficking in HPS-1 Renal Epithelial Cells. [Board No. PB3456]

Bernadette R. Gochuico, M.D.

2:15 – 4:15 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 124 – GWAS meta-analysis on heavy menstrual bleeding in three large biobanks reveals 28 loci suggesting multiple contributing pathways [Board No. PB2077]

Andrew Johnson, Ph.D.

2:15 - 4:15 p.m.

Conv Ctr/Exhibit & Poster Halls AB

Poster Presentation: 125 – Systematic Integration of Multi-omics Data for the Study of Coronary Artery Disease and Subclinical Atherosclerosis [Board No. PB4381]

George John Papanicolaou, Ph.D.

2:15 - 4:15 p.m.

Conv Ctr/Exhibit & Poster Halls AB