Concurrent Featured Symposia, 8:30-10:00 AM

Ballroom West, Level 3	Session 17 - Integration of Long-Read Sequencing with Multi-Omics Data to Identify Hidden Causal Variants
Room 258ABC, Level 2	Session 18 - NIH/NIA-ADSP Artificial Intelligence and Machine Learning Consortium: Gene Discovery and Drug Development for Alzheimer's Disease
Room 253ABC, Level 2	Session 15 - Genes, Traits, and Identities: Navigating Sex, Gender, and Sexuality in Modern Research
Room 210AB, Level 2	Session 12 - Beyond Transcriptomics: Can Imaging Bridge the Gap in Variant Interpretation?
Room 210C, Level 2	Session 14 - From Data to Diagnosis: Advancing Rare Disease Research through Collaborative Genomics
Room 206AB, Level 2	Session 13 - Filling the Placenta Gap
Room 205ABC, Level 2	Session 11 - Authentic Engagement Beyond the Lab: Engaging Communities in the Future of Genetic Medicine
Room 156ABC, Level 1	Session 16 - Genomics at the Incubator: Advancing Neonatal Care Through Translational Insights

10:00 – 10:45 Coffee Break in Exhibit & Poster Hall
SIG Meetup: Phenotypes & Genotypes, Room 104AB
SIG Meetup: Genetic Counselors, Room 102AB

Wednesday, October 15

ASHG ANNUAL MEETING 2025
BOSTON, MA • OCTOBER 14-18

Concurrent Platform Sessions, 10:45-11:45 AM

	Ballroom, Level 3	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2	Room 156ABC, Level 1
	Session 19 - Alz in the Details: Piecing Together the Alzheimer's Puzzle	**Session 26 - Seq-ing Function with Single Cells and CRISPR Perturbations**	**Session 21 - From Identification to Consequence: Structural Variant Detection, Mapping, and Effects**	**Session 22 - From Interaction to Insight: Leveraging Gene-Environment and Multi-Omics Interactions Across Populations**	**Session 20 - Evolution of the Coding and Non-Coding Genome**	**Session 24 - Loop Logic: 3D Chromatin Architecture and Functional Genomics**	**Session 23 - It Is What It Isn't, Non-Canonical Drivers of Disease Gene Expression**	**Session 25 - Mendelian Mysteries Through a Global Lens**
10:45 AM	Single-cell-resolved phenotypes for Alzheimer's disease across 599 individuals and 3.4 million cells	Systematic targeting and inhibition of bone mineral density GWAS loci in single cells identifies novel regulatory networks for human bone formation	Marker-based tumor-normal haplotypes matching for accurate genome-wide recombination identification and haplotype specific somatic structural variation discovery using GIAB tumor-normal assemblies	Novel method for scalable non-additive GWAS reveals 781 new loci across 2,329 traits in FinnGen	Expanded models of gene constraint improve selection estimates from multi-ancestry population data	3D genomic approaches mapping accessible chromatin and architecture implicate causal risk variants in primary open-angle glaucoma pathogenesis	Genetic control of alternative transcriptional initiation reveals a pan-cancer atlas of susceptibility genes	A decade of Initiative on Rare and Undiagnosed Diseases (IRUD): a nation-wide diagnostic and research network in Japan
11:00 AM	Spatial transcriptomic of hippocampal region-specific pathology in Primary Age-Related Tauopathy and Alzheimer's Disease	PerTurbo: A scalable Bayesian framework for whole-transcriptome differential expression analysis and optimal design of single-cell CRISPR screens	Personalized Mapping of Allele-Specific Chromatin Accessibility at Single-Cell Resolution	Context-specific polygenic scores improve prediction and portability in complex human traits	The cumulative effect of protein-truncating variants on human life expectancy and overall health	Subtype-Specific Differences in 3D Genome Architecture Reveal Regulatory Mechanisms in Medulloblastoma	Population-scale single-cell long-read sequencing reveals transcriptome variations and genetic effects on alternative splicing across lung cell types	Novel coding and noncoding rare variant association and in vivo functional validation across the developmental continuum
11:15 AM	A genome-wide epitranscriptomic regulatory landscape in the aged human brains with Alzheimer's disease	PhenoBridge: A Unified Framework for Interpreting Molecular Effects on Complex Phenotypes through Integration of Perturb-seq and Phenotypic Data	Bridging long- and short-read sequencing to accelerate population scale structural variant association studies	Genetic Ancestry and Environmental Differences in Somatic Alterations and Clinical Outcomes for Five Common Cancers	Centromere-spanning haplotypes reveal deep ancestry and evolutionary dynamics in the human genome	Comprehensive Dissection of Alzheimer's Disease Genetic Variants Using a Neuron-Tuned 3D DNA Foundation Model	Improving isoform-level eQTL and integrative genetic analyses of breast cancer risk and mammographic density using long-read RNA transcript assemblies	Biallelic Variants in KMO Cause a Novel Form of Congenital NAD Deficiency
11:30 AM	Single-Cell Multi-Omic Profiling Reveals an Alzheimer's Disease-Associated Somatic Mutation Burden in the Human Brain	Single-cell, Trimodal Omics Atlas of Ileal and Colonic Mucosal T Cells in Crohn's Disease Highlights $CD4^+$ Regulatory T Cell Plasticity in Inflammation Progression	Structural and rare variants underlying psoriasis susceptibility identified by whole-genome sequencing: discovery of CERCAM as a novel risk gene	Incorporating UV exposure into functional genomic analyses identifies additional key cancer drivers and pathways at melanoma susceptibility loci	Co-evolution of lactase persistence and glucose homeostasis in ethnically diverse Africans	Transformer-enhanced Hi-C Upscaling Unveils 3D Genome Architecture at Low Sequencing Depth	Topoisomerase IIb binding underlies driver alterations and frequently mutated regulatory elements in cancer genomes	Advancing National Genomic Initiatives: Abu Dhabi's Model for Newborn Genetic Screening and Lifelong Precision Medicine

ASHG ANNUAL MEETING 2025
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Concurrent Platform Sessions, 1:30-2:30 PM

	Ballroom West, Level 3	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2	Room 156ABC, Level 1
	Session 34 - Single-Cell Multiomic Dissection of Gene Regulation Across Disease States	**Session 27 - Advanced Technologies Uncover Dosage, Structure, and Phenotypic Complexity**	**Session 29 - Cancer Risk: Gene Discovery and Polygenicity**	**Session 31 - Making a Connection: Pleiotropy in Neurologic and Psychiatric Conditions**	**Session 28 - Advances in Long-Read Sequencing Studies**	**Session 30 - Hijacked Immunity: Navigating the Balance Between Host Defense and Immune Dysregulation**	**Session 33 - RNA Functions Beyond Coding Sequence**	**Session 32 - Precision Risk for Every Brain**
1:30 PM	Integrated single cell map of the human pancreas and immune-related tissues provides novel insights into diabetes risk	More is more: Shared phenotypes amongst sex chromosome trisomies hints dosage-sensitive effect of pseudoautosomal regions in genetic males and females	Cell type-specific risk gene discovery in colorectal cancer through single-cell transcriptome-wide association study and regulatory element integration	Gene discovery and pleiotropic architecture of chronic pain in a genome-wide association study of over 1.2 million individuals	Novel variant discovery and interpretation from long-read sequencing on 1,027 African Americans in All of Us	Modeling Viral Evolution and T Cell Evasion through HLA-Informed Deep Learning	mcDETECT: Characterizing mRNA Localization in Polarized Neuronal Compartments With Spatial Transcriptomics	Genome-wide multi-trait and multimodal single-cell analyses of tobacco smoking and schizophrenia reveal novel risk loci and regulatory mechanisms
1:45 PM	Integrative analysis of single-cell chromatin accessibility and GWAS statistics to prioritize enhancers and partition trait heritability in lung development	Completely resolved structural variants by optical genome mapping with adaptive sampling from CNV discovery	Single-cell Transcriptome-wide Association Study Identifies Cell-Type-Specific Genes for Lung Cancer	Mapping the Pleiotropic Genetic Landscape of Brain Imaging Traits Using DIMPLE-GWAS and SuSie-GeneSet	Long story short: discovery of new complex mutations and new candidate genes in Parkinson's disease using long-read sequencing	East Asian GWAS of serum immunoglobulin and subclass levels provides insights into the genetic architecture of humoral immunity implicated with immune cell type specificity	Sequence-dependent regulators of transposable-element-associated alternative splicing	Genome Wide Association Study of Schizophrenia identifies African-enriched variants in PTPRD and TMEM150C
2:00 PM	Enhancer-Driven Gene Regulatory Networks Reveal Distinct Transcription Factor Hierarchies in Myeloid Cell Fate Decisions in Crohn's Disease	A multi-omic diagnostic framework leveraging long-read sequencing for unsolved rare disease	The most comprehensive characterisation of polygenic risk for cutaneous melanoma to date	A cell-type-resolved deep learning atlas of human cis-regulatory evolution	Cue2: deep learning framework for structural variant discovery with long reads	Using functional genomics to understand variation in response to infection in northeast Thailand	Saturation mutagenesis of 37 human splicing factor genes using pooled prime editing	Rare Genetic Variation Illuminates Bipolar Disorder Risk Across Diverse Populations
2:15 PM	Disease Genetic Scorecard Reveals Convergent Coding and Regulatory Variants in Complex Diseases	Comparative genomic analysis of syndromic and nonsyndromic forms of orofacial clefting	Identification of 33 new germline risk loci for clonal hematopoiesis by integrating genetic data on blood cell counts and indices	A deep cellular atlas of the human ventral substantia nigra in Parkinson's identifies a genetic and molecular overlap with Type 2 Diabetes	Sharing valuable rare disease and multiomic data through the GREGoR Consortium	Gene regulation in T cells depends on the severity of atopic dermatitis	Non-coding snRNA variants in RNU4-2 and RNU6 paralogues are a recurrent cause of retinitis pigmentosa	Leveraging genome assembly and pangenome graphs to investigate an African-origin protective haplotype for Alzheimer's Disease in APOEε4 Carriers

2:30 – 4:30 PM	Poster Sessions (Wed.), Coffee, CoLab Sessions, Exhibit & Poster Hall
	Brain Break with Museum of Science, Gene-ius Lounge in Exhibit & Poster Hall
2:45 – 3:45 PM	ASHG Advocacy in Action: Driving Science Policy Forward in Room 206AB, Level 2
3:00 – 4:00 PM	Meet the Editors at ASHG Central, Booth 854
4:30 – 5:00 PM	Awards Recognition I: Mentorship & Lifetime Achievement Professional Awards in Ballroom, Level 3
5:00 – 6:30 PM	Presidential Symposium: Unraveling the Genetic Foundations of Human Disease: Insights from the Past, Present, and Future in Ballroom, Level 3

ASHG ANNUAL MEETING 2025
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Concurrent Platform Sessions, 8:30-10:00 AM

	Ballroom, Level 3	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2	Room 156ABC, Level 1
	Session 36 - Artificial Intelligence and Machine Learning Tools Reshaping Modern Genomics	**Session 37 - Choose Your Own Adventure: Novel Approaches in Statistical Genetics**	**Session 40 - Expanding the RNAverse in Health and Disease**	**Session 42 - Massively Parallel Experimental and AI Approaches for Variant Characterization**	**Session 39 - Delivering on Precision Medicine: Final Outcomes and New Insights on Returning Genetic Results Across Diverse Cohorts**	**Session 38 - Computational Tools for Precision Therapeutics**	**Session 43 - Statistical Genetics: Multi-Omics to Improve Gene Discovery**	**Session 41 - Human Diversity and Evolution**
8:30 AM	Decoding gene expression in cellular and disease states with deep learning	Computation and resource efficient genome-wide association analysis for large-scale imaging studies	Deep reinforcement learning tuned self-attention for nucleotide resolution mapping of regulatory variants in long non-coding RNA	Variant pre-classification using ~280,000 variant effect measurements in 42 disease genes...	The All of Us Research Program's Return of Research DNA Results Program: A Complete Review	Machine learning-enabled genetic discovery and therapeutic targeting of hepatic IRS1 in MASLD	Integrating Genomic and Proteomic Data Improves Complex Trait Prediction in Diverse Populations	Is Directional Selection Rare in the Recent Human Past? Insights from Ancient DNA Time Series
8:45 AM	Integrating genetics, multimodal medical imaging, and AI for high-precision risk prediction of type 2 diabetes	A phenome-wide map of pleiotropy: Connecting genetic signals to disease mechanisms and drug discovery	First Comprehensive Long-Read Transcriptome Assembly of the Human Placenta Reveals Novel Molecular Mechanisms of Maternal-Fetal Health	T cell-specific causative variants underlying immune-mediated inflammatory disorders	RESPECT3: A randomized study evaluating return of actionable genetic research results from the Penn Medicine Biobank	Cas9 cytosine base editor proteins for precision genome editing with high specificity and activity	Integrative multi-omics approaches identify key molecular pathways and improve risk prediction of Late-Onset Alzheimer's Disease	Spatiotemporal dynamics of adaptive evolution in Neolithic East Asia
9:00 AM	G2DBridge: A Two-Step Machine Learning Framework for Disease Risk Prediction Using...	Parent-of-origin inference and its role in the genetic architecture of complex traits: evidence from ~235,000 individuals	GTEx and GENCODE 47 integration expands the role of long-noncoding RNAs in complex traits and disease	Global identification of inflammatory bowel disease risk variant allele-dependent gene regulatory activity	Return of hereditary cancer research results to Black women: Results of a non-inferiority trial...	Leveraging Open-Source Large Language Models to Identify Undiagnosed Patients with Rare Genetic Aortopathies	Integrating multi-omics and multi-context QTL data with GWAS reveals ...	A comprehensive map of introgressed and adaptive structural variation in diverse humans
9:15 AM	Machine learning-derived exosomal circRNA signature enables early lung cancer detection and reveals metabolic dysregulation	mFABIO: Multi-tissue TWAS fine-mapping to prioritize causal tissues and genes for binary traits	Comprehensive long non-coding and circular RNA atlas of human brain regions across multi-ancestry cohorts	Cell2net: A Deep Learning Framework to Decode Genotype-Specific Gene Regulatory Networks from Single-Cell Multi-Omics	The GUARDIAN expanded newborn screening study: Short- and medium-term follow-up...	Enhanced Prediction of Antisense Oligonucleotide Therapy Eligibility for Rare Genetic Diseases Using a Semi-Automated Bioinformatics Pipeline	Integrating multi-omics data to identify lung-tissue-specific susceptibility proteins for lung cancer risk	PHATE Derived Ancestry Coordinates Can Effectively Capture Continuous Population Structure in Human Genomic Data
9:30 AM	Distinguishing causal variants from linked hitchhikers in the human genome using deep learning	Leveraging cis and trans genetic data to improve protein level prediction in proteome-wide association studies	Identification of long non-coding RNAs potentially involved in inherited retinal disease pathogenesis	AlphaGenome: advancing regulatory variant effect prediction with a unified DNA sequence model	A post-test counseling model accelerates access to cardiovascular genetic test results while ...	DeepDR-ADT: Multi-Modal Deep Learning for Drug Repurposing with Auxiliary Dosage-Time Task Learning	Integrated Mendelian randomisation and multi-omic validation identify causal serum proteins...	Stable genetic risk for myopia over 15,000 years in East Asian and European populations...
9:45 AM	PubMind: A Large Language Model Framework for Scalable Extraction of Variant and Disease Associations from Biomedical Literature	causalR2M: causal Mendelian randomization mediation analysis with high-dimensional multi-omics mediators...	A novel human β cell microprotein from a lincRNA controls insulin secretion and harbors rare null variants associated with type 2 diabetes	Functional annotation of over 25,000 type 2 diabetes-associated variants	Implementing polygenic risk scores in clinical care...	Building a 100 million cell single cell atlas of drug perturbations across 50 human cell models for AI driven drug discovery	Multiomic analysis of circadian and seasonal biomarkers	Machine learning identifies the genomic and statistical features that predict cross-population replication of GWAS signals

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Concurrent Platform Sessions, 10:45-11:45 AM

	Ballroom, Level 3	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2	Room 156ABC, Level 1
	Session 45 - Dissecting the Biology of Neurodegenerative Disorders	**Session 48 - In Situ Insight: Uncovering Cellular Regulation with Spatial-Omics**	**Session 47 - Genetic Findings in African Genetic Ancestry Populations**	**Session 51 - Resolving Complex Genomic Variation in Rare Disease**	**Session 49 - Methylation Mechanisms: New Insights into DNA-Methylation in Health and Disease**	**Session 44 - Beyond the Bean: Genetic Studies of Kidney Disease**	**Session 46 - Functional Dissection of Variants at Scale**	**Session 50 - Precision in Chaos: Exploring the Intricacies of Immune Regulation and Autoimmunity**
10:45 AM	Localizing MS GWAS SNPs to pathogenic cell types reveals convergent mechanisms of microglia dysfunction	BRIDGE: Inferring 3D Molecular Tissue Structure from Sparse Spatial Omics and Histological Sections	Proteogenomic analysis of BMI in ~200,000 continental Africans and admixed African identifies 28 loci and enhances cross-ancestry incident obesity prediction using proteomic scores	Comprehensive characterization and analysis of nuclear mitochondrial DNA segments (NUMTs) from long-read sequencing data	Epigenome-Wide and Causal Mediation Analyses Identify DNA Methylation Pathways Mediating Longitudinal Glycemic Effects on Diabetic Retinopathy	Novel genetic insights into kidney function in countries with a high prevalence of chronic kidney disease: A meta-analysis of GWAS from 596,624 East Asians in Japan, South Korea, and Taiwan	Multi-modal functional mapping of non-coding sequences regulating fetal hemoglobin	A multi-omics resource of B cell activation reveals genetic mechanisms for immune-mediated diseases
11:00 AM	Age at onset GWAS of Parkinson's Disease in the FinnGen study suggests links with genetic risk for earlier age at onset and known prodromal symptoms for Parkinson's	Applying the Novel Illumina Spatial Technology in Pig-Kidney to Human Xenotransplant biopsies allows deep profiling of infiltrating human immune cells	Characterizing the genetic and environmental factors that impact vitamin D metabolism in sub-Saharan Africa	Identification of de novo variants from parent-proband duos via long-read sequencing	Widespread non-Mendelian inheritance of DNA methylation patterns in mice	Ancestry-specific genetic loci for kidney function identified in the largest African-ancestry meta-analysis	A comprehensive MPRA of both trans-ancestral genome-wide and suggestively-associated childhood obesity loci implicates a role for CBLN4 in microglia	Multi-Trait GWAS and Fine-Mapping of 14 Traits Identify 17 Ancestry-Specific Immune Loci in Fibrotic and Inflammatory Diseases
11:15 AM	Large-scale transcriptomic profiling across brain regions in Progressive Supranuclear Palsy	Mitochondrial lineage tracing within spatially intact human tissues	Dissecting Genetic Contributors to eGFR Bias: Effects of African-Enriched GATM Regulatory Variants on Detailed Renal Phenotypes	Improved Transcriptome Diagnostics in the Undiagnosed Diseases Network Using the Telomere-to-Telomere CHM13 Genome	Differential Bulk and Cell-Type Specific DNA Methylation in Asthma with Severe Exacerbations in Pediatric Populations of Diverse Ethnicities	Bridging Genomic Inequities: Chronic Kidney Disease Susceptibility and Pharmacogenomic Variation in Indigenous Australians	Deciphering causal variant effects in Digestive Disease via Organoid-based allele specific open chromatin and CRISPRi	T cell receptor repertoire-wide association study identified potential biomarkers for rheumatoid arthritis
11:30 AM	Genomic and epigenomic insights into purkinje and granule neurons in Alzheimer's disease and related dementia using single-nucleus multiome analysis	Subcellular spatial transcriptomics with spatial neighborhood analysis framework reveals immune–stromal crosstalk within the synovium of patients with juvenile idiopathic arthritis	Gene expression associated with impending severe asthma exacerbations in non-Hispanic Black individuals with asthma	Characterization of Rare Genomic Structural Variants Across 2,981 Genomes Reveals Significant Involvements in Recessive Conditions	Therapeutic epigenome editing for Prader-Willi Syndrome using a novel non-viral, non-nanoparticle delivery platform for CRISPR/dCas9-based gene regulation	Uncovering the inconsistencies and potential risks of AI: inaccuracies in nephrogenetic counseling provided by five LLMs	Multimodal CACNA1C deep mutational scanning to assess the impact of all missense variants on ion channel function	Post-GWAS analysis in autoimmune-inflammatory diseases: strength and patterns of allele-specific DNA (ASM) vary between disease-relevant cell types

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Concurrent Platform Sessions, 1:30-2:30 PM

	Ballroom, Level 3	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2	Room 156ABC, Level 1
	Session 56 - Genetic Insights Fueling Drug Discovery and Precision Health	**Session 58 - Mendelian Disorders of the Epigenetic Machinery: New Cellular and Phenotypic Insights**	**Session 54 - Function of Complex Variation and Repeats**	**Session 53 - Every Story Has a Beginning and an End: About UTRs and Poly(A) Tails**	**Session 57 - Genetic Mechanisms of Cancer Risk**	**Session 55 - Gene Editing and Gene Replacement for Rare Disease**	**Session 59 - Shaking The Evolutionary Tree: Multi-Species Studies to Understand Human Biology**	**Session 52 - Autism Spectrum Disorder: From Genetic Variation to Neurobiology**
1:30 PM	Human genetics evidence supporting ANGPTL4 inhibition to reduce remnant cholesterol and triglycerides informs successful Phase 2a trial outcomes	Cell type-specific dysregulation of gene expression due to Chd8 haploinsufficiency during mouse cortical development	A phenome-wide association study of CNVs genotyped from genome sequencing data in the UK Biobank	Genome-wide detection of human 5'UTR variants of functional significance in translation	Comprehensive characterization of functional pleiotropy at the 5p15.33 multi-cancer risk locus	Corrective editing to treat the connective tissue disorder pseudoxanthoma elasticum	Single Cell Multiomics Across Nine Mammals Reveals Cell Type Specific Regulatory Conservation in the Brain	The role of topologically associating domains in defining the pathogenicity of de novo non-coding variants on autism spectrum disorder risk
1:45 PM	Associations between ultra-rare loss-of-function variation and 454 traits in the Mexico City Prospective Study	ASXL1 mutations drive metabolic alterations through mitochondrial dysfunction	Identification of genetic modifiers of somatic instability across multiple repeat expansion disease loci using in vivo CRISPR–Cas9 genome editing	MicroRNA Perturb-Seq Reveals Genome-wide Functional Targets and Deleterious 3'UTR Variants	Genome-wide characterization of clonal hematopoiesis reveals extensive non-coding putative driver mutations	High-Coverage Whole-Genome Sequencing Uncovers Complex CRISPR-Induced Genomic Rearrangements in Human Hematopoietic Stem Cells	Genetic control of regulatory dispersion across human and chimpanzee cell types	Rare coding variants identify novel genes and phenotypic context of autism
2:00 PM	GLP1R and GIPR coding variants reveal differential impacts on metabolic and mental health – Implications for drug response in incretin receptor agonist treatment	Defining the source of motor deficits in KMT5B-linked syndrome	Computational analysis of tandem repeats for genome-wide identification of novel repeat expansions	Modulating gene expression in human cells via high-throughput prime editing of regulatory elements	Detection and quantification of cancer-associated mutagenic processes in normal human somatic cells	A one and done therapeutic strategy to correct ELP1 splicing defect in familial dysautonomia	Elucidating the metabolic mechanisms of cellular homeostatic resilience	Shared and distinct genetic architectures of autism and neuropsychiatric disorders
2:15 PM	Hypercholesterolemia polygenic score modulates response to statin therapy	Rare genetic variants in histone modification pathways implicate epigenetic dysregulation in ADHD	Going Viral: Targeted Detection and Profiling of Endogenous Retrovirus Transcripts in the Human Genome	Atlas of cell type-specific genetic impacts of alternative polyadenylation in immune-related diseases	A multiplex assay of variant effect (MAVE) of MSH6 enables accurate, prospective Lynch Syndrome clinical variant interpretation	Mouse models of, and AAV gene therapy for, cobalamin C (cblC) deficiency	The human and non-human primate developmental GTEx projects reveal tissue-specific gene expression across developmental stages	Chd8-dependent molecular and functional vulnerabilities in the developing and adult mouse cerebellum

10:00 – 10:45 Coffee Break in Exhibit & Poster Hall

SIG Meetup: Bioinformatics and Computational Methods, Room 104AB

2:30 – 4:30 PM Poster Sessions (Thu.), Coffee, CoLab Sessions, Exhibit & Poster Hall

3:00 – 4:00 PM Meet the ASHG Editors at ASHG Central, Booth 854

4:30 – 5:00 PM Awards Recognition II: Early Career & Leadership Professional Awards, Ballroom

AJHG Award for Outstanding Trainee Publication

HGG Advances Award for Outstanding Early Career Publication

5:00 – 6:30 PM Featured Plenary Abstract Session II, Ballroom

Concurrent Featured Symposia, 8:30-10:00 AM

Ballroom West, Level 3	Session 65 - Deep Learning for Non-coding Variant Interpretation
Room 258ABC, Level 2	Session 64 - Decoding Human Aging: From Single-Cell Resolution to Population-Scale Insights
Room 253ABC, Level 2	Session 66 - Genomes from the Global South: Population Genomics Projects and their Impact on Precision Medicine in Global Populations
Room 210AB, Level 2	Session 67 - Machine Learning-Derived Phenotype Refinement for Genomic Discovery and Prediction
Room 210C, Level 2	Session 62 - Breakthroughs in Down syndrome Research in the Age of the NIH INCLUDE Project
Room 206AB, Level 2	Session 63 - Clinical and Translational Aspects of Genetic Hearing Loss
Room 205ABC, Level 2	Session 68 - The Cell Village: A Platform for Population-Scale Genetic Studies in a Dish
Room 156ABC, Level 1	Session 61 - Bench to Bedside: The Clinical Impact of the Latest Advances in Epilepsy Neurogenetics

10:00 – 10:45 Coffee Break in Exhibit & Poster Hall
SIG Meetup: Emerging Laboratory Technologies, Room 104AB

Friday, October 17

ASHG ANNUAL MEETING 2025
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Concurrent Platform Sessions, 10:45-11:45 AM

	Ballroom, Level 3	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2	Room 156ABC, Level 1
	Session 75 - The Utility of AI in Clinical Genomics Workflows	**Session 70 - Contextualizing Somatic Variation: From Clonal Expansion to Molecular Mechanisms**	**Session 72 - Functional Consequences of Transcriptome Variation Across Tissues and Populations**	**Session 76 - Unraveling Disease Mechanisms Through Multi-Omics: From Genetics to Microbiome and Chromatin Regulation**	**Session 73 - Grey Matters: The Science of Aging**	**Session 71 - From Classroom to Community: Shaping the Future of Genomics Education**	**Session 69 - Advances in Cell-Free DNA Applications from Prenatal Genomics to Cancer Diagnostics**	**Session 74 - Pan-Genome Perspectives on Cardiovascular Diseases**
10:45 AM	Automated Phenotyping Using NLP Rivals Human Expert Curation and Enables Scalable Gene Prioritization in Clinical Diagnostic Workflows	Landscape of somatic mutations across genomes, tissues, and individuals	Multi-modal dissection of coronary artery disease GWAS loci reveals transcript splicing-mediated risk mechanisms in vascular smooth muscle cells	Microbiome and host genetic effects on gene regulation in Inflammatory Bowel Disease	Linking single-cell transcriptomic and genomic changes in the aging human brain	PaRtnEring to build understanding oF gEnomics Responsibly: the PREFER CHW research education program	Noninvasive fetal exome sequencing from maternal plasma: Large-scale validation and optimization for prenatal genetic use	Uncovering Hundreds of New Loci from a Multi-Ancestry Study of Blood Pressure Traits in Over 2.5M Individuals
11:00 AM	Artificial intelligence cannot replace human intelligence: publicly available generative AI is not suitable for variant classification	Clonal Hematopoiesis of Indeterminate Potential (CHIP) Mutations Are Enriched in Alzheimer's Disease Patients with APOE ε3/ε3 Genotype	Isoform-Level Transcriptomic Signatures of Glycemic Traits	Multi-omics integration nominates disease-relevant cell types and novel effector genes for Crohn's disease and ulcerative colitis	Epigenetic mosaicism acquired in human embryogenesis and aging	Psychometric Validation of the Education and Assessment of Genetic Literacy (EAGL) Scale	Performance of prenatal cell-free DNA screening to identify sex chromosome abnormalities: Experience of a commercial laboratory	Cigarette smoking modifies genetic associations with serum lipids in cross-population meta-analyses of 920,966 individuals
11:15 AM	The Community Texome Project: Advancing Rare Disease Diagnosis Through Genomic Medicine, AI, and Functional Modeling in Texas communities	Multi-ethnic Transcriptomic Analysis of Clonal Hematopoiesis Reveals Driver-Specific Pathways and Causal Associations	Allele-specific transcriptome variation identification at single-cell resolution in human retina enabled by single-cell long-read RNA sequencing	Integrating GWAS and CRISPR screens reveals context-specific T cell cytokine regulation in inflammatory bowel disease	Epigenome-Wide Analysis of Aging in the Largest Nanopore Methylation Cohort of the Emirati Genome Program	Learning Competencies and Pedagogical Considerations for the Future Public Health Genetics Workforce	Prenatal cell-free DNA screening detects fetal genetic conditions linked to uniparental disomy	Genetic underpinnings of stroke in Hispanic/Latino populations
11:30 AM	Benchmarking genAI tools for literature retrieval and summarization for genomic variant interpretation	Population scale single-cell multi-omic analysis of clonal haematopoiesis	Multi-omics in 10,000 diverse individuals expands mapping genetic mechanisms in health and disease	Resolving genome regulatory complexity with new multiomic long-read sequencing tools for chromatin state, DNA methylation, and genetic variation	Non-linear molecular QTLs in the human brain reveal context-dependent effects on aging and neurodegenerative disorders	How Epigenetics is Communicated on YouTube: A Content Analysis of Science Communication Strategies	Genome-Wide Variations of End Motif in Cell-Free DNA Fragments Distinguish Immunotherapy Responders from Non-Responders in Head and Neck Cancer: A Multi-Institute Prospective Study	Global Evaluation of Congenital Heart Disease-Associated Non-Coding Variant Effect on Cardiac Transcription Factor Regulation

ASHG ANNUAL MEETING 2025
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Concurrent Lightning Talks, 1:30-2:30 PM

	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 205ABC, Level 2	Room 206AB, Level 2
	Session 79 - Genetic Landscapes: Innovations in Population Genetics, Epidemiology, and Genomic Tools	**Session 80 - Interpreting Human Variation in Cancer Genomics and Pharmacogenomics**	**Session 77- Beyond Sequencing: Genetic Architecture of Complex Traits**	**Session 78 - From Bedside to Bench: Delineating Novel Genomic Etiologies of Mendelian disorders**	**Session 81 - Linking Genetic Variation to Molecular, Cellular, and Phenotypic Function**	**Session 82 - Scaling Genomic Medicine: From Patient-Centered Tools to Population and Therapeutic Insights**
1:35 PM	Fine-mapping of loci under directional selection reveals functional architecture of human adaptation	Rare Variant Burden Analysis Identifies Novel Germline Susceptibility Genes in Early-Onset Colorectal Cancer	Proteomic Signatures as Biomarkers of Atherosclerosis Burden	KAT14 Deficiency as a Novel Genetic Cause of Diamond-Blackfan Anemia	Using pangenomes to characterize protein-coding and mutational constraint in high-identity duplicated genes	The Patient-reported Genetic testing Utility InDEx (P-GUIDE): A novel measure of personal utility for pediatric clinical genetics
1:40 PM	A novel framework to characterize monogenic disease by leveraging founder populations in biobank-scale data	Clonal Hematopoiesis of Indeterminate Potential: A Modifiable Risk Factor for Prostate and Bladder Cancer	Beyond heritability: Multimodal AI integrating imaging and genetics enables population scale precision coronary artery disease risk prediction	Detection of Allele-Specific DNA Methylation of X chromosome in a Female Patient With a Truncating MED12 Variant Using Long-Read Sequencing	A scalable framework to link rare human variants to disease phenotypes using pooled prime editing	Population-Scale Genomic and EHR-Driven Insights into Inherited Retinal Disease Burden and Family-based Risk in the Emirati Population
1:45 PM	GATE-GENE: Rare Variant Survival Testing for Time-to-Event Phenotypes Identifies Genes Associated with Disease Onset and Progression Missed by Binary Trait Analyses in Large-Scale Biobanks	Integrative analysis identifies multiple germline genetic variants and biologic pathways for chronic lymphocytic leukemia, yielding a predictive genetic risk score	An Integrated Multi-Trait PRS and Proteomic Approach Identifies Biomarkers and Therapeutic Targets in Chronic Obstructive Pulmonary Disease	Saturation genome editing of the non-coding snRNA RNU4-2 reveals distinct dominant and recessive neurodevelopmental disorders	Comprehensive multimodal single-nucleus profiling of an iPSC reprogramming trajectory reveals variable inter-individual epigenetic differences	When Recurrent Copy Number Variants Are Not the Whole Story: Diagnostic Yield from Alternative Etiologies in a Pediatric Cohort
1:50 PM	PRS Without Population Labels: Accurate Multi-Ancestry Risk Prediction with ARGs	Meta-Analysis of Rare Cancers Identifies Germline Risk Variants and Functional Mechanisms in Anal Cancer and Gastrointestinal Stromal Tumor	Genetic architecture of fat and lean mass across body compartments in men and women	Transcriptomic signatures in postmortem brains uncover tau-related neurotoxicity as a novel disease mechanism in X-linked dystonia-parkinsonism	Single-Cell Multiomic Mapping Uncovers Cell-Type-Specific Regulatory Architecture of Chronic Kidney Disease	Uncovering Genomic Diversity in the African Diaspora via Long-Read Nanopore Sequencing
1:55 PM	Association of a multi-trait polygenic risk score with type 2 diabetes in the UK Biobank	Saturation genome editing of BRCA1 across cell types resolves variant pathogenicity and improves cancer risk estimation	HLA Allele Frequencies and Phenome-Wide Associations in a Diverse Cohort from the All of Us Research Program	Deficiency of the cationic amino acid transporter SLC7A1 causes spastic cerebral palsy & hereditary spastic paraplegia	A High-Resolution Examination of Genetically and Environmentally Induced Methylation Changes in the Million Veteran Program	Integrating 76,000 Whole Genome Sequences from ADSP Release 6 (R6) to Accelerate Alzheimer's Disease Genetics Research

Concurrent Lightning Talks, Continued

	Room 258ABC, Level 2	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 205ABC, Level 2	Room 206AB, Level 2
2:00 PM	Genetic variants regulating immune cell-cell interactions are key drivers for autoimmune disease risk	Identification of target genes at inherited risk loci in chronic lymphocytic leukemia	Defining the Molecular Effect of the Autoimmune-Risk Allele on Surface CD40 Expression in B Cells	Transcriptome-wide approach to gene and variant discovery in Mendelian disorders of the minor spliceosome	Epigenome-wide association study of lifecourse socioeconomic position with cardiovascular and cognitive health in US Hispanics/Latinos from the Hispanic Community Health Study/Study of Latinos	OTC-HOPE: The first in vivo, liver directed, AAV-mediated, gene insertion clinical trial in infants with Ornithine Transcarbamylase Deficiency
2:05 PM	Integrative Multi-omics Analyses to Identify Divergent Genetic Loci and Molecular Regulators of Crohn's Disease and Ulcerative Colitis with Real-world Data Applications	Single-cell eQTL dataset of lung tissues from Asian never-smokers highlights the roles of alveolar cells in lung cancer etiology	MSH3 is a modifier of repeat somatic expansion, somatic contraction and age-at-onset in X-linked dystonia parkinsonism (XDP)	Gene knockouts across 138,000 individuals for novel rare disease gene discovery	Genetic regulation of cell-type–specific chromatin accessibility shapes immune function and disease risk	Imsidolimab, a novel high affinity IgG4 IL-36 Receptor Antagonist, was Effective and Well- Tolerated in Patients with Generalized Pustular Psoriasis: Results from Phase 3 trials, GEMINI-1 and GEMINI-2
2:10 PM	Spatial Multi-omics at Scale: A Unified Framework for Human Biology, from Molecular Atlases to Clinical and Spaceflight Applications	Going the Extra Mile to Understand Chronic Lymphocytic Leukemia	Targeting glutamatergic pathways: genetic insights into comorbid neurodevelopmental disorders	Beyond the Germline: Mapping Somatic Causes of Rare Inflammatory Disease in Adults	Isogenic induced pluripotent stem cells uncover distinct effects of TCF7L2 and a common diabetes GWAS variant on pancreatic beta cell differentiation and function	Construct Interpreted Literature Database using LLM
2:15 PM	Single-Cell Multi-Omic Drug Response Profiling of 1 Million Cells Across 500 PRISM-Multiplexed Cancer Cell Lines Sequenced with SBX	Identification of CPT1A as a Blood-Based Biomarker of Mifepristone Efficacy in Alcohol Use Disorder	Uncovering Ancestry-Specific Genetic Architecture in Admixed Individuals of African Descent with a Scalable Tractor GWAS Workflow	Aberrant splicing prediction across human organ development	Comprehensive analysis of splicing snRNA genes and pseudogenes reveals new associated rare diseases genes	The impact and challenges of more data: retraining of a Bayesian genotype-phenotype model on nearly one million additional patients to resolve VUS
2:20 PM	Mapping the Exposome: An integrative meta-reference across Child and Parent diverse cohorts from Human Health and Exposure Resource (HHEAR)	GWAS of creatinine response to RAAS inhibition in 32,907 Finns identifies regulatory variant near LINC01267	Improved glaucoma polygenic risk score enables clinical utility and risk prediction across major ancestries	Genetically imputed ancestry in a large clinical cohort undergoing carrier screening identifies new genes to target, but reveals potential disparity	Addressing RNA-seq alignment bias against gene-altering SVs improves structural characterization and expression quantification	3ASC 2.0: A unified deep learning model for comprehensive variant prioritization across WES and WGS in rare diseases

2:30 – 4:30 PM	Poster Sessions (Fri.), Coffee, CoLab Sessions, Exhibit & Poster Hall
	Hiring Mixer, Gene-ius Lounge in Exhibit & Poster Hall
3:00 – 4:00 PM	Meet the Editors at ASHG Central, Booth 854
4:30 – 5:00 PM	Awards Recognition III: Education, Advocacy, and Scientific Achievement Professional Awards, Ballroom
5:00 – 6:30 PM	Plenary Abstract Session III, Ballroom

Concurrent Platform Sessions, 8:30-9:30 AM

	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2
	Session 85 - Better Together: Multi-Ancestry Approaches for Gene Discovery	**Session 84 - All That and a Bag of Variants: Functional Genomics of Brain Disorders**	**Session 88 - Using Disease Models to Assess Variant Affect**	**Session 87 - Prenatal Genetics and Reproductive Health**	**Session 86 - Genomic Causes and Consequences in Drug Treatment**
8:30 AM	Multi-ancestry genome-wide and transcriptome-wide association analyses identify new risk loci and genes for Inflammatory Bowel Disease	Optimally matching healthy and disease-specific adult brains onto the EN-TEx personal epigenome resource	Exploring the Utility of A Knockout Mouse Resource for Insights into Human Disease	Anticipating Futures: Stakeholder Perspectives on Ethical Governance of Prenatal Gene Editing	Genome-wide association study for Glucocorticoid-induced Ocular Hypertension
8:45 AM	Largest multi-ancestry genetic study of 99 infectious disease traits identifies hundreds of novel disease-associated loci across the whole genome	Genetic atlas of the human brain revealed by a 2-Million-Voxel GWAS at millimeter resolution	Digenic variants that affect an actin-mitochondria-glutamate pathway cause epilepsy	Utility of Genomic Autopsy for Infant Mortality	High-throughput Identification and Genetic Analysis of Adverse Drug Effects in Individuals of African Ancestry
9:00 AM	Multi-ancestry genome-wide association study of endometriosis and its clinical manifestations in ~1.4 million women: translating gene discovery into pathogenic mechanisms and therapeutic targets	Uncovering molecular mechanisms of neuropsychiatric risk via functional genomics and AlphaFold 3	Investigating cilia-autophagy crosstalk in skeletal development and FGFR3-mediated pathogenesis of achondroplasia	Prospective comparative evaluation of optical genome mapping, mate-pair genome sequencing, and chromosomal microarray analysis in prenatal genetic diagnosis of fetuses with ultrasound anomalies	Genetic control of gene expression in whole blood of childhood cancer survivors
9:15 AM	Next generation multi-population GWAS of Coronary Artery Disease	SingleBrain: A Meta-Analysis of Single-Nucleus eQTLs Linking Genetic Risk to Brain Disorders	ZIC1 is a transcription factor that intersects Idiopathic Hypogonadotropic Hypogonadism and Craniosynostosis	Lifecourse Genetic Risk of Cardiovascular Disease in Women with Reproductive Health Conditions	Genomic and transcriptomic insights into antipsychotic-induced changes in total cholesterol in a multi-ancestry cohort of 58,000 veterans

Concurrent Platform Sessions, 10:00-11:00 AM

	Room 253ABC, Level 2	Room 210AB, Level 2	Room 210C, Level 2	Room 206AB, Level 2	Room 205ABC, Level 2
	Session 93 - The Metabolic Matrix: Untangling Complex Conditions	**Session 89 - Biobank-Scale Genomic Data Resources of Diverse Populations**	**Session 90 - Cardiometabolic Traits: Insights from Multi-Omics Approaches**	**Session 92 - Pathogenic Mechanisms in Neurological Disease: Gene to Network and Everything In Between**	**Session 91 - End Results—Telomere Resolution and Involvement in Genomic Instability**
10:00 AM	Molecular Risk Scores Enhance Type 2 Diabetes Prediction Across Clinical Contexts	Large-scale metabolomic profiling enables genome-wide and phenome-wide association studies in the Million Veteran Program	Large scale metabolomics study in 69,671 South Asians identifies novel rare variant associations with lipids	Dissecting MECP2 cis-regulatory architecture with massively parallel reporter assays to assess its contribution to male-biased autism	Telomere dysfunction promotes remote recombination events between telomeres, centromeres and ribosomal DNA that are catalyzed by long-range chromatin interactions
10:15 AM	Male-specific genetic regulation across germline and somatic variation on the Y chromosome contributes to type 2 diabetes	Our Future Health: The World's Largest Cohort Study is Open for Genetic Research	Connecting proteomics and genomics to identify sex-specific plasma biomarkers for calcific aortic valve stenosis	CYP2D6 contributes to cognitive dysfunction and promotes excessive synapse loss	A genomic foundation model for ALT detection reveals new therapeutic vulnerabilities in pediatric cancer lacking actionable drug targets
10:30 AM	Signatures of positive selection in Pacific-ancestry genomes reveal adaptive signals relevant to metabolic diseases	The All of Us Research Program data release 2026 (CDR v9): Powering genomic research through All of Us	Proteomic discovery in an inception cohort of acute myocardial infarction survivors	Synergistic Loss of Regulatory and Protein-Coding Genes Drives Neuronal and Mitochondrial Dysregulation in Prader-Willi Syndrome	FLT3 regulates telomere maintenance and suppresses senescence in acute myeloid leukemia
10:45 AM	Phenome-wide Genetic Colocalization and Mendelian Randomization of eQTLs and pQTLs against complex traits in up to 2.5 million participants across multiple Biobanks	Uncovering genetic etiologies in 30,000 rare disease genomes from a nationwide Chinese cohort	Adipose single nucleus transcriptomic and epigenomic sequencing discovers 192 colocalized regulatory variants for eight cardiometabolic disease traits	Transcriptomic Profiling of Extracellular Vesicles from Autism Patient-Derived Forebrain Organoids Reveals Novel Signatures	Determinants of chromosome-specific telomere lengths estimated from long reads among 2,573 All of Us participants

11:15 – 12:45 PM Session 94: AI Powered Genomics: Transforming Data into Insights, Ballroom