



Program at a Glance

The following tables show session times and locations as of October 15. Exact rooms and speakers are subject to change. Please check ASHG's Annual Meeting app for the most up-to-date information about sessions and events.

Pre-Meeting Workshops and Courses

2:00 pm – 5:00 pm

Room 514BC/Level 5	Room 515AB/Level 5
Session 01 – Short Course: Active Learning Strategies for Teaching Variant Curation (Part 1 of 3) (Ticketed Event)	Session 02 – Short Course: UCSC Genome Browser (Part 1 of 3) (Ticketed Event)



Tuesday Ticketed Events

9:00 am – 12:00 pm

Room 513DEF/Level 5	Room 514BC/Level 5	Room 515AB/Level 5
Session 05 – Nextflow for Omics: Jumpstart your Analyses with nf-core Pipelines	Session 03 – Short Course: Active Learning Strategies for Teaching Variant Curation (Part 2 of 3)	Session 04 – Short Course: UCSC Genome Browser (Part 2 of 3)

10:00 am – 12:00 pm

Room 518/Level 5	Room 524AB/Level 5
Session 06 – Modeling Choices in Omics: Implications for Health Disparities in Human Genetics	Session 07 – Strategies for Communicating Your Science

10:00 am – 4:00 pm

Room 512ABCEFG/Level 5
Session 08 – Using AI-Enhanced, Multiomic Tools to Solve Rare and Undiagnosed Diseases

1:00 pm – 3:00 pm

Room 518/Level 5	Room 514BC/Level 5	Room 515AB/Level 5
Session 11 – Fine-mapping and Colocalization with Structural Variants: Practical Workflows Using GWAS and Multi-omic Summary Statistics	Session 09 – Short Course: Active Learning Strategies for Teaching Variant Curation (Part 3 of 3)	Session 10 – Short Course: UCSC Genome Browser (Part 3 of 3)

1:00 pm – 4:00 pm

Room 513DEF/Level 5	Room 524AB/Level 5
Session 13 – Best Practices for Working with AI in Variant Interpretation in Clinical Genome Analysis	Session 12 – Leadership Development Workshop: Leading from Where You Are

7:00 pm – 8:30 pm

720 Atrium/Level 7
Trainee Reception



Tuesday Plenary Sessions

4:30 pm – 5:00 pm

Room 517/Level 5

Session 14 – Finding Your Voice in Science: Reflections on Leadership and Resilience

5:00 pm – 6:15 pm

Room 517/Level 5

	Session 15 – Plenary Abstract Session I
5:00 pm	Automated interpretation of genomes for rare disease research with deep graph attention
5:19 pm	A whole-genome immunogenomics framework for African populations: Antigen processing, presentation, and clinically actionable diversity across the HLA, ERAP, TAP, and KIR loci
5:38 pm	Solving the unsolvable: A systematic multi-omic approach for rare disease diagnosis across Canada
5:57pm	CDCT1 Screening in 527K Participants: A 3-year Outcomes Study

Wednesday Concurrent Featured Symposia

8:15 am – 9:45 am

Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
Session 20 – Human Mutational Processes Underlying Genomic Signatures, Cancer, and Evolution	Session 19 – Genomics of Sex Differences and Reproductive State Across the Life Course: Context, Timing, and Biological Transitions	Session 16 – Adult DNA-Based Population Genomic Screening: Moving from Pilots to Practice	Session 18 – Beyond GWAS: Translating Human Genetic into Therapeutic Targets and Clinical Development	Session 22 – The All of Us Research Program At 10 Years: 500,000+ Genomes Advancing Precision Medicine	Session 17 – Autism Genetics: Integrating Biological Insights with Societal Impacts	Session 21 – Integrating AI and Knowledge Graphs for Human Genetics and Beyond: Innovations from the NIH Common Fund Data Ecosystem	Session 23 – Unique Considerations for Deceased Human Donor Tissue Biobanks to Support Innovative Genomic Studies
Introduction	Introduction	Introduction	Introduction	Introduction	Introduction	Introduction	Introduction
Pervasive lesion segregation and genetic background shape cancer evolution	Altered states: Biological complexity in sex, aging, health, and disease	DNA-based population screening (DNAPS) in adults – Building programs that prepare for the future and learn from the past	Advancing gene discovery and therapeutic translation through population-scale genomics in global biobanks	Building to 1 million genomes and more: How All of Us grew to the largest, most diverse public - omics resource and where it is going next	Autism genetic testing: Stakeholder perspectives in an era of expanding genomics	The Bridge2AI Consortium Cell Maps for AI (CM4AI) Project: Accelerating functional genomics within the Common Fund Data Ecosystem	Pediatric pathology and metadata management for biobanking
Segregating DNA lesions point to high selective advantage of tumor initiating cells	Sex differences in the genetic architecture of clinical quantitative traits in the electronic health record	How do we decide what to screen for? The ClinGen Population Genomic Screening Actionability Framework is here to help	Calibrated inference for translating human genetics into high-confidence therapeutic targets	Increasing discovery potential by adding multi-omics to the All of Us Research Program dataset	Deciphering phenotypic and genetic heterogeneity in autism: Examples from co-occurring neuropsychiatric conditions and quantitative symptoms	Advancing protein-level insight through the UniProt-Common Fund Data Ecosystem partnership	Using fibroblast-derived donor specific assemblies to call somatic mutation in a haplotype-resolved manner- an example of what you can do with a biobank

Comparative primate somatic genomics to understand human ageing and cancer risk	The genetic architecture of reproductive milestones within the lifespans of ancestrally-diverse women from the population architecture using genomics and epidemiology study	Scaling genetics services for population screening: Experiences, outcomes & impacts	Predicting trial results and informing clinical development with human genetic data: IL-6 signaling as a case study	Genetic and epigenetic landscape of self-identified Hispanics in All of Us	Personalized medicine and ethical intervention targets: A trait-centric approach to autism genomics research	The Common Fund Data Ecosystem Data Distillery Knowledge Graph: Unifying Common Fund insights through structured connectivity	A neurobiobank perspective: Challenges associated with brain banking for large scale projects
Collateral mutagenesis funnels disparate sources of DNA damage into a ubiquitous mutational signature	Sex modifies APOE ε4 effects on human microglial states across Alzheimer's disease progression	Implementing population genomics at scale: Global strategy, local reality, and lessons from a 14,000-participant pilot	Genetic prioritization frameworks to inform drug discovery and development	Contextualizing and integrating All of Us with other global biobank data in drug discovery	Co-creating genetics education resources matched to autism community contexts	REVEAL: AI-enabled synthesis of genetics and mechanism in the Common Fund Data Ecosystem	An integrated and cross-collaborative ethical, legal and social implications team model (ELSI 360°): Harnessing notes from two large-scale genomic studies involving deceased pediatric and adult donors
Panel discussion and Q&A	Panel discussion	Panel discussion	Panel discussion	Panel discussion	Panel discussion and Q&A	Panel discussion	Closing remarks
Closing remarks	Closing remarks	Closing remarks	Closing remarks	Closing remarks	Closing remarks		



Wednesday Events

9:45 am – 10:45 am

Exhibit & Poster Hall	Room 524AB/Level 5	Room 518/Level 5
Exhibit Hall Activities: Coffee Break, Concessions, Scavenger Hunt, Bark Park, Poster Viewing	Shared Interest Group Meetup: Genetic Counseling	Shared Interest Group Meetup: Phenotypes, Genotypes, and Disease Biology

10:00 am – 4:00 pm

ASHG Central, Exhibit & Poster Hall
Career Coaching (Pre-registration Required)

12:00 pm – 1:30 pm

Exhibit & Poster Hall	Room 518/Level 5
Exhibit Hall Activities: Concessions, Scavenger Hunt, Bark Park, Poster Viewing	Session 32 – Ethical and Cultural Implications: Navigating the Future of Genomics Research

2:30 pm – 4:30 pm

Exhibit & Poster Hall
Exhibit Hall Activities: Poster Presentations, Coffee Break, Concessions, Scavenger Hunt, Bark Park, Headshot Lounge, Photo Booth at ASHG Central

2:45 pm – 4:15 pm

Gene-ius Lounge, Exhibit & Poster Hall
Speed Mentoring (Pre-registration Required)

3:00 pm – 4:00 pm

ASHG Central, Exhibit & Poster Hall
Meet the ASHG Editors

4:30 pm – 6:15 pm

Room 517/Level 5
Session 39 – Awards Recognition & Presidential Symposium: Communicating Science in an Uncertain World: Evidence, Storytelling, and Public Trust

Wednesday Concurrent Platform Sessions

11:00 am – 12:00 pm

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 27 – Mice, Mutations, and Molecular Mysteries	Session 29 – Rare Variants, Big Insights: Illuminating Human Disease Genetics	Session 31 – The REnAL Deal: Genomic Insights into Kidney Disease	Session 30 – The Evolving Landscape of Genomic Newborn Screening with Global Experience and Emerging Challenges	Session 28 – Promise and Peril of Sequence-to- Function Models for Variant Effect Predictions	Session 25 – Detecting Human Adaptation: Methods, Drivers, and Health Impacts of Natural Selection	Session 24 – Clone Wars: Mutability and Its Consequences	Session 26 – Mapping Genetic Relatedness at Biobank Scale: Uncovering Hidden Signal from Rare Variants to Parent- of-Origin Effects
11:00 am	Rare variants in LRRFIP1 as a novel cause of severe early-onset scoliosis and common scoliotic phenotypes	Exome sequencing directly implicates 68 genes in inflammatory bowel disease	The genetics of inherited kidney disease in Africa: Disease-causing variants identified by whole-exome sequencing in a South African cohort	Emerging evidence for genomic newborn screening from over 45,000 newborn genomes from England	Improving gene expression prediction across diverse populations	Multi-ancestry selection and introgression maps in the NIH All of Us Biobank	Clonal hematopoiesis enables mutational epidemiology at biobank scale: Mutational signatures of 65,000 people connect environment to disease	Cryptic pedigree reconstruction from identity-by-descent maps ultra-rare disease variants at biobank scale
11:15 am	CNS-directed transferrin receptor-targeted enzyme replacement therapy for β -Mannosidosis	Rare coding and noncoding variants map 1,342 diseases and biomarkers in 490,549 whole genomes	Longitudinal GWAS reveals the genetic architecture of kidney function across the life course in the population and disease	Genomic newborn screening across diverse genetic ancestries: GUARDIAN screen-positive rates and variant findings among >19,000 newborns	In silico saturation mutagenesis of the human genome with AlphaGenome improves variant prioritization and interpretation	Ancient DNA reveals natural selection upregulating the human immune system over the last 10,000 years	Frequency of clonal hematopoiesis mimicking constitutional variants in cancer patients undergoing genetic testing	Accurate maternal and paternal haplotype inference reveals new parent-of-origin effects in the Finnish population

11:30 am	3D regulatory rewiring of SOX3 by an interchromosomal insertion causes spastic paraplegia	Large-scale whole-genome landscape of rare diseases in 15K Koreans	Multi-ancestry genome-wide association study meta-analysis on 3.5 million individuals expands the genomic landscape of kidney function	Rapid panel sequencing and automated pipeline for genetic hearing loss in newborns - The fasT genOmic heAring losS Testing (TOAST) study	Independent evaluation reveals advances in AI-driven variant impact prediction: Findings from CAGI 7, the Critical Assessment of Genome Interpretation	Heterogeneous selection signatures across West & Central African populations are associated with cardiometabolic, immune, and renal disease burden in African-ancestry populations	Molecular features of disorders of somatic mosaicism: Variant clustering and allele fraction patterns across 1,337 patients	Multi-phenotype high-resolution fast identity-by-descent mapping test
11:45 am	Rare, common, and repetitive variants and unexplored genetic features shape complex traits and nominate novel therapeutic targets in 67,609 Japanese whole genomes from Tohoku Medical Megabank	MAP3K7 loss-of-function variants cause isolated dilated cardiomyopathy	Distinct genetic architectures of kidney function decline quantified by multi-state Markov analysis of 3.4 million serial eGFR measurements across 13 health systems	Estimating false positives in genomic newborn screening using the Generation UK study	When to trust a genomic deep learning model: Effect magnitude as an intrinsic confidence signal for personalized expression prediction	A new statistical test for detecting multiple modes of positive selection from ancestral recombination graphs	Multimodal genomic profiling of solid tumors using optical genome mapping, long-read nanopore sequencing, and deep next-generation sequencing: Diagnostic, prognostic, and therapeutic significances	Understanding genetic and environmental complexity in phenotypic resemblance across relatedness in population-scale data

Wednesday Concurrent Lightning Talk Sessions

1:30 pm – 2:30 pm

	Room 510/Level 5	Room 511/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 35 – Advancing Precision Oncology with AI, Functional Genomics, and Multi-Omics: From Risk Prediction to Tumor Evolution and Therapeutic Discovery	Session 37 – Decoding Genetic Risk Across Ancestry and Sex in Polygenic Disorders	Session 38 – Genetic Variation: From Catalogs to Consequences	Session 34 – Advances in Population Genetics, Genetic Epidemiology, and Omics	Session 36 – Clinical Genetics Across the Lifespan: From Gene Discovery to Prenatal, Developmental, and Pharmacogenomic Insights	Session 33 – Advances in Clinical Genomics in the Era of AI, Long-Read Sequencing, Precision Medicine, and Genomic Care Delivery
1:30 pm	Mendelian randomization using genomic foundation models improves discovery power over additive QTLs and prioritizes cell-type specific therapeutic targets	G6PD deficiency and sickle cell trait have different impacts on HbA1c screening for abnormal glucose tolerance and monitoring diabetes treatment in continental Africans	The All of Us + AnVIL imputation service: The world's largest, most ancestrally diverse genomic reference panel for equitable variant discovery	Convergent human adaptation during parallel subsistence transitions across Eurasia	Variants in SFPQ underlie a novel dominant neurodevelopmental syndrome	Data-analysis biases are exacerbating GWAS representation gaps
1:35 pm	DeepRVAT2: Unified modeling of coding and regulatory rare variation at genome scale for enhanced gene discovery and diagnostics	Genetic architecture and polygenic risk score for intracerebral hemorrhage in 1.6 million individuals across trans-ancestry groups	Deploying long-read sequencing to bridge diagnostic and equity gaps in rare disease	Sampling alternative gene genealogies in an ancestral recombination graph	Analysis of 184,786 developmental disorder trios identifies over a hundred novel DD-associated genes and candidate haplolethal genes	Perceived understanding of results, autonomy, and security of care (PURASC): Patient experiences following whole-genome sequencing and standard genetic testing in adult neurogenetics
1:39 pm	Machine learning-based penetrance of genetic variants using metabolomics data	Cross-population genome-wide association meta-analyses highlight polygenetic, cellular, and spatial contrasts distinctly between dilated and hypertrophic cardiomyopathies	Robust annotation transfer across modern genome assemblies and species using synteny-aware coordinate projection	Characterizing differences in gene expression variability between humans and chimpanzees	Real-world prevalence of clinically significant RNU4-2 variants and the value of genotype-first reanalysis	Challenges in genomics: Lessons from patients and clinicians in Latin America
1:43 pm	Using Haplotype-Aware Analysis to help distinguish between germline, post-zygotic mosaic, and clonal somatic variants	Annotation-informed soft clustering of genome-wide association study signals identifies biologically interpretable factors and novel genes underlying type 2 diabetes heterogeneity	A human bone molecular QTL atlas reveals transcript-level regulatory mechanisms across skeletal and systemic traits	Proteomic prediction of disease risk is specific to short-term disease incidence	3D genome rewiring and lncRNA-associated cis-regulatory disruption of the FOXL2 region underlie blepharophimosis syndrome, a rare enhanceropathy	Closing the gap: A telehealth model with delivery of pediatric genomic sequencing in underserved border communities

1:47 pm	Choosing a clinical trial design to determine the real-world survival impact and health system value of tumor-agnostic next-generation sequencing	Dissecting obesity heterogeneity using cross-trait PRS and data-driven clustering for precision risk prediction	SNP-based heritability distinguishes genetically anchored and environmentally patterned methylation variation in the Black American postmortem brain	AFT-LDSC: The genetic architecture of disease timing reveals distinct drivers of onset and progression across the human phenome	A genetic screen for a model of PIGA-CDG identifies GSK3B as a candidate modifier	Whole genome sequencing and genetic counseling for people living with Parkinson's disease: The PD GENERation experience
1:51 pm	Cost-effectiveness of mono- and polygenic risk-guided breast cancer screening in the US	Proteomic signatures of lifestyle capture environmental risk and predict incidence of major chronic diseases independently of polygenic risk	Genetic architecture of plasma metabolites in highly autozygous South Asians	Mitigating cryptic and inevitable PGS overfitting in the era of million-person biobank cohorts	Long-read multi-omic sequencing for genetic diagnosis of neuromuscular disorders	Clinically integrated genomic screening yields reportable findings in >90% of individuals
1:55 pm	Risk of mutagenesis in near space	Sex-stratified analysis uncovers sex-dimorphic genetic modifiers of cardiac structure and function	Multi-omic profiling of the human caudate nucleus reveals sex-divergent epigenetic aging and its disruption in alcohol use disorder	Deep learning-based framework discovers adipose tissue -resolved plasma protein biomarkers for steatotic liver disease	Elucidating the genetic architecture of congenital heart defects in NICU through trio-based WGS	Accurate detection of Robertsonian translocations using Oxford Nanopore reads by estimating copy-number loss at acrocentric satellite junctions
1:59 pm	Polygenic risk stratification identifies hypothyroid patients with skin cancer risk comparable to family history and UV-related risk factors	Sex-stratified genome-wide association study of asthma and asthma-subtypes reveals age-dependent genetic differences and stronger sex-specific architecture	Convergent RFX3-dependent regulatory programs in language cortex across genetically heterogeneous autism	Validation of polygenic scores for complex traits in 490,000 our future health participants	Single cell multiomics of amniotic fluid uncovers molecular distinctions across spina bifida lesion types	Scalable, routine, methylation-aware long read genome sequencing of undiagnosed rare disease samples
2:03 pm	Circulating immune and cancer driver gene biomarkers precede prostate cancer diagnosis by over ten years	Genome-wide aggregated trans-effects analysis reveals B-cell activation and checkpoint signalling as therapeutic axes in primary biliary cholangitis	Deciphering causal variant effects in digestive disease via organoid-based allele specific open chromatin and CRISPRi	The genetic architecture of somatic passenger mutation burden in clonal hematopoiesis	Discovery of genetic risk factors in stillbirth highlights evolutionary constraint and incomplete penetrance	Concurrent long-read genome sequencing with methylation pattern analysis enhances diagnostic yields in the UCI-GREGoR rare disease cohort
2:07 pm	An integrative chromatin accessibility atlas of human gastric tissue links regulatory genetic variants to gastric cancer susceptibility	Dynamic genome elements are associated with infection risk and improve sepsis prediction in high-risk individuals	Population-scale immune multiome atlas of 10 million PBMCs reveals regulatory disease mechanisms in FinnGen	Pathway-specific proteomic aging clocks reveal disease-associated aging programs and protein drivers	Integrating GWAS with a multimodal atlas of the female reproductive system reveals critical cell types and pathways in gynaecological disorders	Gene- and domain-aware calibration increases the clinical utility of variant effect predictors

2:11 pm	Functional analysis of somatic variants across multiple tissues in SMAHT	Exposure to maternal human milk components interacts with infant polygenic risk and improves prediction of childhood atopy	Genetic architecture of pathway-specific metabolomic aging clocks in the Canadian Longitudinal Study on Aging	Integrating single-cell multi-omics with GWAS reveals hepatocyte-specific regulatory networks underlying circulating polyunsaturated fatty acids	Population-scale characterization of monogenic diabetes in 374,973 multi-ancestry All of Us participants reveals heterogeneous variant prevalence and variable penetrance	AI-based large language model agents for effective classification of clinical severity in 8,738 gene-disease pairs
2:15 pm	Interpretable multi-omic integration reveals novel epigenetic and transcriptomic subgroups in pancreatic ductal adenocarcinoma (PDAC)	Spatial transcriptomics of human knee osteoarthritis cartilage at single-cell resolution	Single-cell CRISPRi screens identify targets and gene programs promoting heart valve fibrosis	Saliva-derived DNA sequencing reveals higher mutation rates compared to blood tissues	Biobank-scale molecular profiling refines interpretation of rare recessive alleles in Mendelian disease genes	Protein language model embeddings predict CRIM status, cardiomyopathy, and disease onset from GAA genotype data in Pompe disease: A machine learning ablation study
2:19 pm	Bayesian ancestral state reconstruction of somatic evolution reveals hidden complexity of hematopoiesis	Rare variants in small nuclear RNA genes reveal a splicing related genetic mechanism in individuals ascertained for autism spectrum disorder	A rapid workflow for generating and characterizing mutations in mitochondrial DNA	The MoTrPAC Data Hub: Resources for the molecular map of exercise	Genome-wide association analysis of ACE inhibitor-induced angioedema in patients of African ancestry	Brain-wide non-viral genome editing for Angelman syndrome using STEP-RNP delivery in non-human primates
2:23 pm	Multi-modal spatial profiling of tumor-immune interactions in triple-negative breast cancer	Genome-wide association studies of psychotic disorders in diverse African populations reveal shared and population-specific genetic risk	A human IARS1 mechanism links deep intronic regulation and coding variation to tissue-specific myopathy	Two latent polygenic endophenotypes underly plasma proteomic levels	Phenome-wide association of pharmacogenomic metabolizer and functional status in the All of Us research program	From imbalance to equilibrium: antisense oligonucleotide therapy targeting allelic expression of PEX6 in Zellweger spectrum disorder
2:27 pm	Functional characterization of the multi-cancer risk locus 5p15.33 in normal pancreas cell types under endoplasmic reticulum stress	High-throughput functional characterization identifies astrocyte specific gene dysregulation at neuropsychiatric GWAS loci	De novo CNOT7 missense variants resulting in loss of deadenylation function for mRNA poly(A) tail shortening, cause a neurodevelopmental disorder	Characterizing the genetic landscape of neurodegeneration through large-scale long-read whole-genome sequencing	FGFBP1-mediated diabetogenic effects of statins: A proteome-wide Mendelian randomization study in the UK Biobank	BPN36964 corrects ELP1 splicing defects and reverses retinal neurodegeneration in familial dysautonomia mice even with delayed therapeutic intervention



Thursday Events

8:15 am – 9:30 am

Room 518/Level 5

Session 103 – Innovation Through Adversity: How Global Genomics Partnerships Are Evolving to Meet New Challenges

9:45 am – 10:45 am

Exhibit & Poster Hall

Room 518/Level 5

Room 524AB/Level 5

Exhibit Hall Activities: Coffee Break, Concessions, Scavenger Hunt, Bark Park, Poster Viewing

Shared Interest Group Meetup: Computational Genomics, AI, and Statistical Genetics

Community Conversations: Engagement and Belonging

10:00 am – 10:45 am

Room 517BC/Level 5

NHGRI 2026 Status Updates: What Researchers Need to Know

10:00 am – 4:00 pm

ASHG Central, Exhibit & Poster Hall

Career Coaching (Pre-registration Required)

12:00 am – 1:30 pm

Exhibit & Poster Hall

Room 518/Level 5

Exhibit Hall Activities: Concessions, Scavenger Hunt, Bark Park, Poster Viewing

Career Development Networking Luncheon (Ticketed Event)

1:30 pm – 2:15 pm

Room 519A/Level 5

Community Conversations: Scientific Communication

4:15 pm – 6:15 pm

Exhibit & Poster Hall

Snacks & SNPs: Afternoon PowerUP and Poster Presentations

5:00 pm – 6:00 pm

ASHG Central, Exhibit & Poster Hall

Meet the ASHG Editors

Thursday Concurrent Platform Sessions

8:15 am – 9:45 am

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 44 – Machine Learning-Driven Approaches to Variant Classification and Prioritization	Session 46 – Sequence Models Transforming Genetics and Genomics	Session 40 – Charting Human Genetic Variation Across Genomes, Populations, and Time	Session 41 – Chromatin in Motion: Epigenetic Control of Brain Development and Disease	Session 47 – The Long and Short of It: Long-Read Genomics from Biobanks to the Clinic	Session 45 – New Adventures in Molecular Trait Mapping	Session 42 – From Labels to Landscapes: Leveraging Local Ancestry and the Ancestry Continuum to Maximize Cross-Ancestry Association	Session 43 – Genetic Associations and Mechanisms in Cardiovascular Disease
8:15 am	Closing the equity gap in variant classification: Machine learning-driven evidence demonstrates differential impact across race and ethnicity	Predicting the molecular effects of common and rare variants across observed and unobserved biological contexts	What shapes early post-zygotic mutation burden? Family and parental age effects in nearly 1000 individuals from the large, four-generation CEPH/Utah pedigrees	A large-scale Perturb-seq map of haploinsufficient neurodevelopmental disorder genes in iPSC-derived neurons reveal convergence and divergence in transcriptional programs	Association analyses from Oxford Nanopore long read sequenced genomes of UK Biobank participants	An atlas of single cell eQTLs across multiple ancestries and different cell type annotations	Computationally efficient local ancestry-aware GWAS with GPU acceleration	Largest GWAS meta-analysis across congenital heart disease phenotypes identifies phenotype-specific loci and novel developmental candidate genes
8:30 am	predNMD: Prediction of nonsense-mediated mRNA decay for improved clinical variant pathogenicity classification	A multi-modal nanopore foundation model enables de novo single-molecule discovery of DNA and RNA modifications	Population-scale Y chromosome assemblies reveal constrained remodeling across 180,000 years of human evolution	Deep learning and massively parallel reporter assays determine non-coding risk for autism spectrum disorder	Haplotype-resolved structural variation and functional consequences across ancestrally diverse human populations	Scalable fine-mapping of single-cell eQTL data using variational count-based models	Moving beyond population labels: Personalizing polygenic risk scores with genetic neighbors	Whole-genome sequencing atlas of cardiac and adiposity imaging traits reveals shared and distinct genetic architectures and nominates therapeutic candidates for cardiometabolic disease

8:45 am	Evaluating performance and reproducibility of generative artificial intelligence models for autonomous variant classification	HiVerse: A multi-resolution genomic language model atlas integrating 5-methylcytosine for tissue- and cell-type-specific epigenome and transcriptome prediction	Ancient and present-day genomes reveal dual origins of Denisovan ancestry in Mainland Southeast Asia	Loss of Bap1 disrupts enhancer activity and 3D genome organization in the developing brain	Haplotype-resolved genomics at biobank scale in ~12,500 participants from the All of Us Research Program	The developmental GTEx resource enables the discovery of disease-associated genes	Decomposing cross-ancestry Mendelian randomization bias into structural, statistical, and architectural components: Robust estimators resolve most cross-ancestry attenuation	Sex-stratified cross-ancestry GWAS of coronary artery disease identifies shared and sex-specific genetic effect
9:00 am	Unlocking 5'UTR variant interpretation in 500,000 UK Biobank genomes	Big models cheat, small models learn: Interpretable splice-site prediction with Angler	Depletion of Neanderthal ancestry on the X chromosome reflects overlapping effects of sex bias, selection, and drift	Time-course Perturb-seq of epigenetic regulators reveal convergence on autism-risk transcriptional programs during neurodevelopment	The Human Pangenome Project: A new foundation for human genomics	From eQTL to function: Dissecting genetic control of Treg suppression	Betanfer: Estimation of linkage disequilibrium from GWAS summary statistics enables large-scale cross-biobank fine mapping	Mapping the genetic landscape of circulating fatty acids through integrative multi-trait GWAS analysis and single-cell CRISPRi screening
9:15 am	Continuous clinical prioritization of copy number variants (CNVs) through dynamic feature reconstruction	Long read RNA sequencing in primary lung cell types reveals functional principles and accurate sequence-based prediction of nonsense-mediated decay	Beyond population labels: A continuous PCA-based framework for ancestry-aware population genomics statistics in gnomAD	Haplotype-resolved long-read chromatin mapping reveals regulatory impacts of structural variation	Population-scale long-read sequencing for medical genomics: A UK population curated genomic dataset from NIHR BioResource	Local ancestry differentially impacts protein quantitative trait locus estimation in an ancestrally admixed cohort	Maximizing genetic association power across the ancestry continuum in large-scale biobanks and cohorts	Single-cell profiling of genetic risk for cardiovascular disease in endothelial cell 'populations-in-a-dish' under athero-prone and athero-protective shear stress
9:30 am	VCEPilot: Agentic AI execution of ClinGen VCEP specifications achieves expert-aligned variant classification	Deep variational models for genome-wide fine-mapping of thousands of complex and molecular traits with cell-type-specific, machine-learning-derived annotations	Quantifying individual genetic variation using data from >900,000 whole-genome sequences: Implications for screening	BRPF1 haploinsufficiency disrupts Wnt signaling during human neurodevelopment	Clinical long-read genome sequencing for rare disease diagnostics – a prospectively designed study of 1,000 genomes	Dissecting genetic effects on gene regulatory mechanisms with single-molecule footprinting	Genome-wide LD scores allow robust heritability and genetic correlation from summary statistics across ancestries	A CAD-associated SNP in the 9p21.3 region regulates the expression of MIR31HG, modulating the expression of several atherosclerosis-related genes

11:00 am – 12:00 pm

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 51 – Genomic Insights Across Reproductive Medicine and Prenatal Diagnosis	Session 49 – Cross-Disease Insights into Neuropsychiatric and Neurodegenerative Disorders	Session 52 – New Frontiers in Mitochondrial DNA Research	Session 55 – Uptown Funk(tion): Don’t Believe Me, Just Perturb	Session 53 – Pivoting from Gene-Level Analyses to Isoform-Resolved Biology	Session 50 – Gene Regulatory Networks Across Development, Evolution, and Disease	Session 54 – Polygenic Risk Score: From Modeling to Clinical Decision	Session 48 – Cancer Susceptibility: Germline Effects on Risk, Mechanisms, and Mutations
11:00 am	Genetic architecture and fitness costs of chromosomal abnormalities from 99,765 products of conception	Cell-type-resolved multiomic mapping of bipolar disorder risk in the human subgenual anterior cingulate cortex	Scaling mitochondrial variant calling in 30x short-read whole-genome sequencing for 535,000 All of Us Research Program participants	Functional dissection of promoter-proximal short tandem repeats reveals sequence class-specific regulatory mechanisms	Developmental splicing dynamics across human and non-human primate tissues in dGTEx and NHP-dGTEx	From human to mouse and back: Leveraging CNVs, GWAS, and in vivo experiments to uncover non-coding SHOX enhancers	RICEx integrates common and rare variants from summary statistics for multi-trait, multi-ancestry risk prediction	Germline polygenic susceptibility shapes somatic mutational signatures across common cancers
11:15 am	A comprehensive noninvasive fetal sequencing (NIFS) test reproduces 97.2% of fetal diagnoses identified by invasive diagnostic sequencing	Single-cell gene expression heritability informed by gene × cell-state interactions	Discovery and characterization of nuclear mitochondrial insertions across human tissues	High-throughput profiling of macromolecular interaction perturbations in human genetic disorders	The ageing human transcriptome: Longitudinal long-read RNA-sequencing in blood uncovers multi-level changes in isoform usage	Resolving the regulatory impact of non-coding disease risk alleles throughout human embryogenesis by single-cell multiomic profiling of advanced stem cell-derived models of post-implantation embryos	A scalable penalized regression framework improves accuracy and efficiency in multi-ancestry polygenic prediction	Real-world multi-ancestry sequencing uncovers germline determinants of somatic driver selection and cancer progression

11:30 am	Resolution of population structure affects carrier frequency estimates and the number of genes meeting ACMG tier 3 carrier screening panel criteria	Short- and long-read RNA sequencing revealed RNA fusions contributing to amyotrophic lateral sclerosis	Mitochondrial DNA variation shapes the human blood proteome and metabolome, highlighting ER stress, oxidative muscle biology, and pyruvate metabolism	Multimodal functional data reveals pathogenicity, pathomechanism, and genotype-phenotype relationships for ~75,000 MAP kinase signaling pathway variants	Long-read RNA sequencing reveals genetic effects on isoform expression in a globally diverse sample	Cis and trans contributions to cell type-specific regulatory evolution in primates	Polygenic Risk Methods Development (PRIMED) Consortium advances polygenic risk scores for diverse traits and ancestries by federated analysis of multiple large-scale biobanks	Gene-specific cancer patterns among pathogenic germline variant carriers
11:45 am	Perspectives of exome or genome sequencing as the first tier test in unexplained spermatogenic failure	Large-scale perturbational analysis of schizophrenia-associated genes in human neurons reveals the transcriptional consequences and convergence of disease genes and loci	Age- and tissue-dependent dynamics of mtDNA alterations clarify optimal specimen selection for clinical mitochondrial testing	Immune context unmasks regulatory effects of Neanderthal and Denisovan introgression	Single-cell full-length transcriptome of human lung reveals genetic effects on isoform regulation beyond gene-level expression	Loss of conserved regulatory elements in human brain evolution	Prospective performance of genetic and clinical risk prediction models to inform risk-based breast cancer screening: Experience from the WISDOM study	Single-cell lung eQTL dataset of Asian never-smokers highlights the roles of alveolar cells in lung cancer etiology

1:30 pm – 2:30 pm

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 60 – Lessons from Real-World Clinical Sequencing Across Neurodevelopmental, Neurological, and Movement Disorders	Session 57 – Elucidating Structural Variation in Human Health and Disease	Session 58 – Global and Cultural Perspectives on Genomic Risk and Implementation	Session 61 – Leveraging Pharmacogenomics for Drug Trial-Level Evidence	Session 59 – Integrative Genomics of Immune Regulation Across Infection, Inflammation, and the Lifespan	Session 56 – EHR-Based Computational and AI Approaches in Clinical Genetics
1:30 pm	Factors influencing molecular diagnostic yield in a clinical autism exome sequencing cohort of over 68,000 individuals	gnomAD-LR: A population-scale, long read reference resource for variant discovery and haplotype-resolved episignatures	Final results of a randomized non-inferiority trial evaluating digital return of actionable genetic research results as compared to return by a genetic counselor (RESPECT3)	Thiazide diuretics are associated with reduced risk of incident dementia: A phenomic-genomic study in All of Us, Mount Sinai Million, and UK Biobank	Dynamic genetic regulation of immune gene expression across inflammatory and infectious disease contexts revealed by single-cell eQTLs	Applying a phenotype risk score to expedite identification of heart failure patients at risk for variant transthyretin cardiac amyloidosis

1:45 pm	Hereditary neurological disorders in Mali, West Africa: Findings from a 12-year research experience	Characterization of de novo structural variants in 1,561 trios with orofacial clefts	Germline predisposition and re-evaluating age-based screening criteria across Asian ethnicities: A 12-year inception cohort study of 10,616 hereditary cancer patients	Rare GLP1R variants selectively attenuate weight loss with semaglutide but not tirzepatide, implicating GIP receptor agonism as a compensatory mechanism	Viral stimulation reveals age- and sex-dependent transcriptional programs shaped by genetics and evolutionary constraint	A multimodal agentic knowledge graph system for screening undiagnosed transthyretin amyloid cardiomyopathy
2:00 pm	Gene-STEPS: Rapid genome sequencing improves diagnostic yield and management for infants with epilepsy	HiFi long read genome sequencing after negative clinical exome sequencing in a consecutive cohort increases diagnostic yields	Clinical genomics in Africa: A CFIR-informed hybrid model for neurodevelopmental disorders in Cameroon	Genetic negative control reveals confounders and disease subtypes in drug trial emulations	Genetic architecture of lung cell-type specific gene expression and cell-cell interaction patterns underlying idiopathic pulmonary fibrosis	Systematic evaluation of diagnostic delay in rare disease in an electronic health systems and the United States
2:15 pm	Diagnostic yield of genome sequencing for individuals with movement disorders	Surveying the structural variant landscape in genetic testing: Prevalence and diagnostic yield in a clinical cohort of 1.8 million individuals	Living with the unknown: Longitudinal psychosocial outcomes of VUS and variant reclassification in clinical oncology	Trial entry, not genetics: Collider bias can create apparent PRS-modified treatment response	Differential methylation patterns in nasal mucosa linked to asthmatic and chronic stress phenotypes in the pediatric Black community	Predicting cancer type progression in dermatomyositis patients using large language model-derived embeddings and multi-label machine learning classifiers



Thursday Plenary Session

2:30 pm – 4:15 pm

	Room 517/Level 5
	Session 62 - Awards Recognition & Plenary Abstract Session II
2:30 pm	Genetic drivers of Human Leukocyte Antigen (HLA) regulatory variation across the pangenome
2:49 pm	Charting mutation heterogeneity across human tissues: Early results from SMaHT
3:08 pm	Long-read sequencing reveals telomere inheritance patterns across human trios and fetal-parental quads with sperm
3:27 pm	Coding and noncoding risk variants for congenital heart defects map to gene regulatory networks in valve cells

Friday Concurrent Featured Symposia

8:15 am – 9:45 am

Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
Session 68 – Recent Advances and Opportunities Across the African Disease Genomics Landscape	Session 64 – Emergence of Complex Traits at the Intersection of Genomics, Exposomic, and Time	Session 70 – Translational Models for Substance Use Disorders	Session 63 – AI Across the Diagnostic Odyssey: From Variant Interpretation to Computable Phenotypes	Session 69 – The IGVF Consortium: Deciphering the Impact of Genomic Variation on Genome Function	Session 67 – Pharmacogenomics: Moving Beyond Gene-Drug Pairs	Session 65 – Epi-Mutations: The Underrepresented Paradigm in Genetics	Session 66 – Host-Pathogen Coevolution: Shaping Genetic Diversity and Disease Resistance
Introduction	Introduction	Introduction	Introduction	Introduction	Introduction	Introduction	Introduction
Characterizing the impact of APOL1-mediated kidney disease in Africa	Gene-environment interactions shape complex disease and trait architecture	Using large-scale genetics in C. elegans to decipher opioid regulatory networks	Using specialist AI to identify variants in DNA sequence data and generalist AI to predict effects	Coding variants: Experimental and predictive evidence to re-classify variants of unknown significance	Multiomic analysis of hepatocyte models – discovery of pharmacogenomics insights in African Americans	Beyond the base pairs: Scoring protein-encoding genomic variants in epigenomic regulation at an atomic resolution level	Two species, one disease: Host-pathogen coevolution in infectious diseases
Blood regulatory variation in South Africans enables GWAS interpretation: Applications to HIV susceptibility and quantitative traits	High-dimensional exposomics: Enabling systematic gene-environment discovery	The hybrid rat diversity panel as a valuable strategy to model stages of opioid use disorder	Agentic reasoning across genomes, RNA, and phenotype for rare disease diagnosis	Noncoding variants: Deciphering variant function using gene regulatory maps from CRISPR perturbations and predictive modeling	Massively parallel functional investigation of the impact of regulatory variants on glucocorticoid response	New advances in epigenomic analysis for precision medicine	Pathogen-driven evolution of human immunity across time and function
Bridging the genomic divide: Rare developmental disorder discovery in an African healthcare setting	Gene-environment in gene regulation mediates phenotypic plasticity over changing environments	Genetics of cocaine consumption and preference in the drosophila genetic reference panel	Precision automated phenotyping for genomic discovery and clinical translation	Molecular networks: A shared CRISPRi resource for mapping transcription factor function across human cell lineages	The use of the All Of Us to identify genetic drivers of platelet factor 4/heparin antibody production in diverse populations	Leveraging DNA methylation signatures in developmental and epileptic encephalopathies: From precision diagnostics to biological insights	The genomic architecture of immune variation across populations

A blueprint for expanding rare disease genetics in Africa: The African Rare Disease Initiative	Do modern diseases arise from genomes living in the past?	Genetic studies of substance use disorders reveal convergent gene regulatory mechanisms in mice and humans	From clinical phenotypes to mechanism: AI-based stratification of patients for genomic discovery	Complex disease: Integrating maps of genome function with genetic risk scores to improve cardiovascular disease prevention	Clinical applications of social determinants of health in pharmacogenomics research	Epi-mutations as drivers of cellular architecture: Organelle convergence in chromatinopathies	A longstanding co-evolutionary interaction between sickle haemoglobin and the Plasmodium falciparum genome
Panel discussion and Q&A	Closing remarks	Panel discussion	Panel discussion and Q&A	Panel discussion	Panel discussion	Panel discussion and Q&A	Panel discussion
Closing remarks		Closing remarks	Closing remarks			Closing remarks	Closing remarks



Friday Events

9:45 am – 10:45 am

Exhibit & Poster Hall	Room 518/Level 5	Room 524AB/Level 5
Exhibit Hall Activities: Coffee Break, Concessions, Scavenger Hunt, Bark Park, Poster Viewing	Shared Interest Group Meetup: Emerging Omics and Clinical Translational Research	Advocates in Action: ACGT Presentations and Networking Event

12:00 pm - 1:30 pm

Exhibit & Poster Hall	Room 524AB/Level 5
Exhibit Hall Activities: Concessions, Scavenger Hunt, Bark Park, Poster Viewing	Session 78 – Behind the Scenes: ASHG Publications Workshop (Ticketed Event)

1:30 pm - 2:30 pm

Room 518/Level 5
Session 87 – The Future of Gene Therapy: Policy Pathways for Safe, Scalable Innovation

2:30 pm - 4:30 pm

Exhibit & Poster Hall
Exhibit Hall Activities: Poster Presentations, Coffee Break, Concessions, Scavenger Hunt, Bark Park, Headshot Lounge, Photo Booth at ASHG Central

2:45 pm – 4:15 pm

Gene-ius Lounge, Exhibit & Poster Hall
Speed Mentoring (Pre-registration Required)

3:00 pm – 4:00 pm

ASHG Central, Exhibit & Poster Hall
Meet the ASHG Editors

Friday Concurrent Platform Sessions

11:00 am – 12:00 pm

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 73 – New Tricks for Tackling Unsolved Neurodevelopmental and Neuromuscular Disorders	Session 71 – Genetics of Cardiometabolic Traits	Session 76 – Single-Cell, Multi-Omics, and Liquid Biopsy Approaches to Decode Lung Disease Genetics and Biomarkers	Session 74 – Resolving Rare Disease: Transcriptional, Functional, and Population-Scale Approaches	Session 104 – What's Next in Genetics and Genomics: Late Breaking Science	Session 75 – RNA Unbound: New Regulatory Layers from Modifications to Circular RNA in Disease	Session 77 – Splicing in Disease Etiology	Session 72 – New Methods for Detecting Clonal Hematopoiesis and Somatic Variation Across Scales
11:00 am	Brain imaging endophenotypes unlock novel gene discovery in isolated dystonia	Placental expression quantitative trait loci (eQTLs) shed light on the biology of the prenatal origins of anthropometric measurements and cardiometabolic complex traits	Elucidation of novel and cell-type specific eQTL in single-nuclear sequencing of lung tissue	Comprehensive characterisation of non-coding and coding effects of de novo variants in a large-scale rare disease case-control cohort	Semantic fragment representations for coordinate-free analysis of genomics data	Genetic effects of common variants on RNA modifications in induced pluripotent stem cells	LARIAT (long-read amplicon RNA-based identification of aberrant transcripts): A clinically validated laboratory developed test for splicing VUS resolution in rare disease	mCA Explorer: An interactive catalog of mosaic chromosomal alterations detected in large population-based genomic investigations
11:15 am	Loss of function of ATAD2B disturbs brain development in mouse and human	From phenome-wide genetic clustering to shared cardiometabolic gene architecture in UK Biobank	Cross-disease single-cell mapping of 2 million lung cells reveals macrophage-centered fibrotic signaling networks	Standardized transcriptome analysis improves rare disease diagnosis in large rare disease consortia	Genome-wide association study of dreams and nightmares identifies over twenty loci	Multi-omic time-lapse dissection of acute miRNA biogenesis disruption reveals temporal and cellular identity signatures	Integrating long-read RNA sequencing with genomics and phenomics in a birth defects cohort to discover disease-relevant splice-altering variants	Somatic chromatin epimutations cap genetic determinism in the human diploid chromatin epigenome
11:30 am	De novo TUBA1B variants cause a syndromic neurodevelopmental disorder associated with brain, intestinal, renal, and immunological defects	Large-scale multi-omics analyses of cardiometabolic traits in the Middle East	Proteome-wide linear and non-linear association study of systemic inflammation indices and Mendelian randomization identifies causal proteins for idiopathic pulmonary fibrosis	Homozygosity gnomAD: Maximizing the clinical value of gnomAD through the power of autozygosity	Genetic dissection of human islet ER stress responses uncovers context-specific T2D variant effects and mechanisms	Multi-omic profiling identifies MALAT1-driven splicing and circular RNA dysregulation in CD8+ T cells from multiple sclerosis patients	Characterization of novel isoforms from long-read trio RNA sequencing of whole blood in rare disease	Single-cell multi-omic dissection of ASXL1-mutant clonal hematopoiesis: Integrating genotype, transcriptome, and protein in myeloid disease progression

11:45 am	Genome-wide tandem repeat expansions in regulatory regions contribute to epilepsy risk	Lipoprotein(a): Monogenic or polygenic?	Beneath the variants: Fragmentomics unmasks epigenomic signal from existing mutation profiling of cfDNA for lung cancer classification and prognosis	A tiered functional evidence framework for rare disease diagnosis in the NIH Undiagnosed Diseases Program	Differentiate molecular hallmarks in prostate tumours linking ancestry and geography	MicroRNA-mediated regulatory flux at the thermal stress-immunity nexus	Integration of dilated cardiomyopathy genomics with transcriptomics from the human heart implicates regulatory molecular mechanisms	Ultra-sensitive profiling of clonal haematopoiesis in an apparently healthy West African cohort reveals dominance of DNMT3A and TET2 mutations
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1:30 pm – 2:30 pm

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 517A/Level 5	Room 517BC/Level 5	Room 520ABDE/Level 5	Room 710A/Level 7	Room 710B/Level 7
	Session 86 – Returning ‘Omics Results to Patients to Improve Health	Session 83 – New Insights into the Genetic Basis of Neurodegenerative Conditions	Session 85 – Operationalizing Precision: Frameworks, Platforms, and Target Discovery for Individualized Genomic Therapies	Session 84 – New Machine Learning Approaches in Genetics and Genomics	Session 80 – MAMO: Multi-Ancestry-Aware Multi-Omics Analysis for Complex Diseases	Session 79 – Decoding Malformation Syndromes: New Disease Genes Across Organ Systems	Session 82 – Multi-Omics Perspectives on Immune Regulation, Adaptation, and Inflammatory Disease	Session 81 – Modeling Genetic Disorders with Organoids
1:30 pm	A longitudinal review of the outcomes of genetic testing in a multi-disciplinary prostate cancer genetics clinic	Powerful SNP and STR association study of young-onset Alzheimer’s disease identifies new genes and infers novel disease biology	Providing ethical guidance for the development of individualized genomic medicine as rare as n-of-1: Results from the GENIE study	Transformer-based modeling of longitudinal health records uncovers genetically distinct trajectories of major depressive disorder	Multi-ancestry genome-wide association and multi-omics analyses elucidate spatio-cellular features of multiple sclerosis genetics	Novel human variants and zebrafish models confirm the critical role of Zinc Finger Homeobox 4 (ZFHX4) in structural eye disorders	Genetic architecture and evolutionary origins of immune trait variation in Europeans	Rapid individualized drug repurposing for the treatment of rare genetic diseases using patient-derived organoids
1:45 pm	One in ten individuals receives a medically actionable result through direct-to-consumer genetic testing: Insights from 6 million research participants	Population-scale whole-genome burden analysis identifies rare coding risk genes for Alzheimer’s disease and Parkinson’s disease	Building the N=1 Collaborative Gene Registry to advance discovery, assessment, and collaboration for bespoke genomic therapies	Accurate genomic prediction in ultra-small patient cohorts	Multi-omics quality control in the Trans-Omics for Precision Medicine (TOPMed) Program using scalable genetically-anchored strategies	Rare variants in DSTYK contribute to vertebral malformations and idiopathic scoliosis	Atlas-level multi-omics data reveals the genetic impact of cell type-specific gene regulatory cascade in human immune diseases	Modeling CHARGE syndrome uncovers disrupted cranial neural crest migration and fate specification

2:00 pm	Behavioral impacts of genome-informed risk assessment (GIRA) on participants in the eMERGE-IV network	Somatic deletions in the brain preserve the reading frame of APOE in Alzheimer's disease	An atlas of human pathogenic variants for splice-switching oligonucleotide therapy	Integrating 730,947 exomes with agentic clinical literature curation improves gene discovery	Genome-wide and multi-omic analyses of irritable bowel syndrome across 25 cohorts implicate immune, circadian, and gut-brain signaling pathways	Haploinsufficiency of RNPS1, a component of the exon junction complex, causes a syndromic neurodevelopmental disorder with skeletal anomalies	Mission aware multi omics profiling of early human adaptation to spaceflight	DNA methylation signatures distinguish pancreatic ductal adenocarcinoma prognostic subtypes of patient-derived organoids
2:15 pm	Nationwide clinical implementation of metabolomic risk scores: Evidence from Finnish occupational healthcare	Large scale analysis of transposable element insertions in Alzheimer's disease across different ancestries	A scalable platform for exon-skipping antisense oligonucleotide therapy development for inborn genetic diseases	A spatially resolved genomic-molecular atlas of human white-matter microstructure	The largest multi-ancestry fine-mapping in over 2 million participants improves credible set resolution and enrichment in breast regulatory elements	Identification of somatic mutations in surgically resected tissues from individuals with craniofacial microsomia (CFM)	Integrating computational genetics and functional genomics reveals palmitoylation as a therapeutically targetable pathway in asthma	High-throughput in vitro and in vivo AAV screen uncovers short, strong, and tissue-specific regulatory elements for gene therapy



Friday Plenary Session

4:30 pm – 6:15 pm

	Room 517/Level 5
	Session 88 – Awards Recognition & Plenary Abstract Session III
4:30 pm	Transcriptomics profiling identifies signatures associated with progression of precancerous gastric lesions
4:49 pm	CRISPR-Cas9 screening reveals ATAD5 as a tissue-specific modifier of CAG instability in Huntington’s disease mice
5:08 pm	Heritability of germline mutagenesis in 40 large three- and four-generation pedigrees
5:27 pm	Fine-mapping gene-gene and gene-environment interactions at phenotype-associated loci

Saturday Concurrent Platform Sessions

8:15 am – 9:15 am

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 520ABDE/Level 5	Room 710B/Level 7
	Session 93 – Translational and Technological Advances in Genetic Therapies	Session 89 – Decomposing Diabetes: Approaches for Understanding Diabetes Heterogeneity	Session 90 – From Autoimmune Risk Variants to Cell States and Target Genes: Integrative Genetic Dissection of Immune-Mediated Disease	Session 91 – Rethinking Genomic Discovery: New Methods for a Complex Genetic Landscape	Session 92 – The Selection Paradox: Why Disease Variants Persist
8:15 am	Genetic and therapeutic characterization of FIG4- linked neurodegeneration	Dissecting the genetic architecture of diabetes subgroups in mexican individuals	A distinct subset of autoimmune diseases defined by profound co-prevalence and genetic risk association with the IL12 pathway	OPERA: A scalable framework for identifying disease-mediating gene programs from perturb-seq data	Why do high-effect common variants persist in human populations?
8:30 am	Characterization and targeting of intronic splicing variants in neurological disease with an AI-informed prime-editing framework	Disentangling adiposity-related and non-adiposity-related genetic pathways for type 2 diabetes	Direct functional characterization of autoimmune risk variants reveals cell state-specific allelic effects in human CD4+ T cells	Genomic foundation models do not yet boost common variant discovery	Transcriptome-wide Mendelian randomization reveals widespread antagonistic pleiotropy between reproductive fitness and complex diseases
8:45 am	Development of a compact bidirectional promoter for coordinated multi-gene expression	Genome-wide polygenic mapping of latent mechanisms underlying type 2 diabetes integrated with CRISPR high-content imaging	Long-read sequencing and site-directed mutagenesis show the validity and non-redundancy of allele-specific CpG methylation for post-GWAS mapping in autoimmune inflammatory diseases	Genome haplotype orchestration via synthetic trajectory (GHOST): Reconstructing population-specific linkage disequilibrium for unrepresented ancestries using only summary statistics	Integrative analysis of variant effects on gene dosage, complex traits, and fitness
9:00 am	AAV gene addition therapy for MMACHC-related combined methylmalonic acidemia and homocystinuria, cobalamin C type (cbLC)	Functional dissection of regulatory variants in type 2 diabetes using massively parallel reporter assays in two cell types	Deciphering the genetic causes of autoimmunity: From non-coding GWAS variants to target genes	A heritability-optimized method for functional prioritization of rare coding variants in complex traits	An end-to-end differentiable, inferential model of stabilizing selection for molecular quantitative traits

9:45 am – 10:45 am

	Room 510/Level 5	Room 511/Level 5	Room 516BC/Level 5	Room 520ABDE/Level 5	Room 710B/Level 7
	Session 101 – X, Y and Why? - Mechanism of Sex Bias in Neuronal Disorders	Session 100 – Move Over SNPs: Structural Variants, Repeat Expansions, and More	Session 98 – Decoding Sensory Diseases: Germline, Somatic, and Regulatory Mechanisms	Session 99 – Genomic Medicine in Underrepresented and Diverse Populations	Session 97 – Cancer Under Pressure: Genomics of Therapy Response and Resistance
9:45 am	An APOE*4-informed genomic atlas of the X chromosome in Alzheimer's disease	Federated analyses yield almost 2 million structural variants from 147,994 genomes from the Genome Aggregation Database and the All of Us Research Program	Novel role of DDX41, a cancer-associated RNA helicase, in inherited retinal dystrophies	Genomics for KIDS in ASEAN: Improving access to genomic testing through regional collaborations in Southeast Asia	Circulating proteomic signatures predict immune checkpoint blockade resistance and immune-related toxicity across cancers
10:00 am	Sex-biased transcriptional programs in the fetal enteric nervous system and Hirschsprung disease susceptibility	Genotype imputation and functional characterization of centromeric structural variants using long-read assemblies	High-throughput splicing assay to functionally characterize genetic variants with splicing effect	Genomic and environmental factors related to neurodevelopmental disorders in Rwanda	Dissecting evolutionary and drug response dynamics in metastatic breast cancer via long-read single-cell transcript isoform, fusion, and mutation analysis
10:15 am	Cell-state-resolved sex differences and APOE genotype interactions in dementia and ADRD: A systematic review of single-cell studies and the structural evidence gap	PAV 3: Comprehensive variant discovery across a range of variant types and sizes	Somatic second-hit detection in retinoblastoma by aqueous humor liquid biopsy	Beyond the primary indication: Incidental findings in apparently healthy individuals from a Jewish - Mexican cohort	Genome-scale screening identifies lncRNA vulnerabilities in triple-negative breast cancer
10:30 am	Sex-specific genetic architecture and liability thresholds in amyotrophic lateral sclerosis	Phenome-wide association studies of short tandem repeats in the UK Biobank identify novel pathogenic repeat expansions and common repeat polymorphisms underlying diverse human traits	Large-scale integrative whole-genome analyses revealed a high prevalence of dominant MME variants and a rare NEFH promoter mutation in idiopathic peripheral neuropathy	Addressing Latin American underrepresentation in ClinVar through a quality-controlled variant governance and data-sharing framework from Brazilian clinical genomics	Logic-gated CAR-T cells for the treatment of relapsed acute myeloid leukemia post-haploidentical bone marrow transplant



Saturday Events

8:15 am – 10:15 am

Room 523/Level 5	Room 524AB/Level 5
Session 95 – Beyond the Blueprint: GENE-ius Interactive Strategies for Engagement (Ticketed Event)	Session 96 – The Human Pangenome Release 3: Data, Tools, Applications, and Ethics (Ticketed Event)

8:15 am – 11:15 am

Room 518/Level 5
Session 94 – Assessing Genetic Variants for RNA and Gene Editing Therapeutic Approaches (Ticketed Event)

11:15 am – 12:45 pm

Room 517/Level 5
Session 102 – Globalizing Human Genetics: From Discovery Through Implementation