ASHG ANNUAL MEETING 2024

PROGRAM-AT-A-GLANCE

DENVER, CO • NOVEMBER 5-9

CONTENT

"Welcome to the ASHG 2024 Annual Meeting in Denver, Colorado, for an opportunity to share ideas, inspire action, and push the boundaries of what's possible. Your presence here reflects the dedication we share toward achieving great things together. Let's make the most of this time by fostering collaboration, innovation, and, most importantly, progress in the field. I encourage each of you to bring your insights, ask questions, and engage fully. We're here not just to solve challenges but to build on our strengths and envision the future together. Thank you for being a part of this journey."

— Bruce D. Gelb, MD
ASHG President

"As your new CEO, I'm honored to be part of the ASHG community, dedicated to advancing human genetics and genomics. ASHG members are at the forefront of transforming lives through groundbreaking research, and together, we'll continue to focus on impactful growth and collaboration. I'm excited about the future of ASHG and welcome your ideas as we work together to realize the benefits of genetics and genomics research. I look forward to meeting you in person. Welcome to Denver!"

— Amanda Perl
ASHG Chief Executive Officer

The following office and attendee services locations are in the Colorado Convention Center:

ASHG Central & Membership Services	Booth 735, Exhibit & Poster Hall, Upper Level
Childcare Room *(Pre-Registration Required)*	Room 612, Street Level
Coat & Luggage Check	Registration Lobby A, Street Level
Exhibit & Poster Hall	Hall F, Upper Level
Family Room	Room 610, Street Level
First Aid	Near Room 507, Street Level Near Stout Street
Housing Assistance	Solutions Center Lobby A, Street Level
Information & Lost and Found	Solutions Center Lobby A, Street Level
Lactation Room with Private Pods	Room 610, Street Level Near Room 101 Behind Lobby A
Quiet/Prayer/Meditation Room	Room 101, Street Level
Registration & Badge Pick-Up	Lobby A, Street Level
Speaker Presentation Upload Room	Room 203, Street Level
Press Office & Registration	Room 207, Street Level

View ASHG Policies for Attendees, including Photography and Code of Conduct:
https://www.ashg.org/2024conduct

GENERAL INFORMATION

Wi-Fi Availability

Complimentary Wi-Fi is available in all public lobbies and meeting spaces. To access the Wi-Fi, select the wireless network **ASHG 2024**, password is **ASHG2024**. If you have an unlimited data plan, we encourage you to use it for better connectivity.

Continuing Education

Amedco LLC designates this live activity for a maximum of 25.25 *AMA PRA Category 1 Credits™* for physicians. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

The American Society of Human Genetics is approved as a provider of continuing education programs in the clinical laboratory sciences by the ASCLS P.A.C.E.® Program. This program is approved for up to 24.50 ASCLS P.A.C.E.® contact hours. All sessions at ASHG 2024 are offered at an intermediate level of instruction.

The National Society of Genetic Counselors (NSGC) has authorized American Society of Human Genetics to offer up to 2.525 CEUs or 25.25 Category 1 contact hours for the activity ASHG 2024.

Note: All credit and contact hour counts are subject to change.

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SCHEDULE-AT-A-GLANCE

	Tuesday November 5	Wednesday November 6
		Exhibit &
Morning		8:00–9:30 am **Platform Abstract Sessions**
		9:30–10:15 am **Coffee Break, Industry Events**
	10:00 am–12:00 pm **Interactive Workshops** (ticket required)	10:15–11:45 am **Platform Abstract Sessions**
Early Afternoon		11:45 am–1:15 pm **Break for Lunch, Networking, Industry Events**
		1:15–2:15 pm **Platform Abstract Sessions**
Mid-Afternoon	2:00–4:00 pm **Interactive Workshops** (ticket required)	2:30–4:30 pm **Poster Sessions & Industry Events**
	4:30–5:00 pm **Presidential Welcome & Address**	4:30–5:00 pm **Awards Recognition I**
	5:00–6:40 pm **Plenary Abstract Session I**	5:00–6:30 pm **Presidential Symposium**
Evening	7:00–8:30 pm **Trainee Reception** (ticket required)	7:00–8:30 pm **Diversity, Equity & Inclusion Reception** (ticket required)

Thursday November 7	Friday November 8	Saturday November 9
Poster Hall Hours 9:30 am–4:30 pm		
8:00–9:30 am **Featured Symposia**	8:00–9:30 am **Featured Symposia**	8:00–9:00 am **Platform Abstract Sessions**
9:30–10:15 am **Coffee Break, Industry Events**	9:30–10:15 am **Coffee Break, Industry Events**	9:30–10:30 am **Platform Abstract Sessions**
10:15–11:45 am **Platform Abstract Sessions**	10:15–11:45 am **Platform Abstract Sessions**	11:00 am–12:30 pm **Distinguished Speakers Symposium**
11:45 am–1:15 pm **Break for Lunch, Networking, Industry Events**	11:45 am–1:15 pm **Break for Lunch, Networking, Industry Events**	
1:15–2:15 pm **Platform Abstract Sessions**	1:15–2:15 pm **Platform Abstract Sessions**	
2:30–4:30 pm **Poster Sessions & Industry Events**	2:30–4:30 pm **Poster Sessions**	
4:30–5:00 pm **Awards Recognition II**	4:30–5:00 pm **Awards Recognition III**	
5:00–6:40 pm **Plenary Abstract Session II**	5:00–6:40 pm **Plenary Abstract Session III**	
7:00–8:30 pm **ASHG Social**		

- Scientific Sessions
- Coffee Breaks, Exhibits, Poster Sessions, and Industry Events
- Special Programming
- Receptions

Tuesday, November 5

10:00 AM – 12:00 PM

WORKSHOP Session 01: A Guide to Card Sort Methods for Engaging Participants and Patients
Room 712 (ticket required)

WORKSHOP Session 02: Getting Started with Biomedical and Genomic Data in the All of Us Researcher Workbench
Room 711 (ticket required)

WORKSHOP Session 03: Using the New Ensembl Genome Browser and Variant Effect Predictor (VEP) to Analyse and Interpret Genomic Variation Data
Room 706 (ticket required)

2:00 PM – 4:00 PM

WORKSHOP Session 04: Building Understanding and Practical Skills in Community Engagement for Genetic Research
Room 706 (ticket required)

WORKSHOP Session 05: Elevating Education: Engaging with Real Data and Tools in the Cloud
Room 712 (ticket required)

WORKSHOP Session 06: Hidden Features of the UCSC Genome Browser
Room 711 (ticket required)

4:30 PM – 5:00 PM

PLENARY Session 07: Presidential Welcome & Address
Mile High Ballroom

5:00 PM – 6:40 PM

PLENARY Session 08: Featured Plenary Abstract Session I
Mile High Ballroom

7:00 PM – 8:30 PM

NETWORKING Trainee Reception
Hyatt Regency Hotel, Capitol 1 Room (ticket required)

Wednesday, November 6

8:00 AM – 9:30 AM

PLATFORM Sessions 09-16: Concurrent Platform Sessions
See talks and rooms on pages 14-15

9:30 AM – 10:15 AM

COFFEE BREAK Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

NETWORKING Shared Interest Group Meetup: Phenotypes & Genotypes
Room 712

9:45 AM – 10:15 AM

INDUSTRY SOLUTIONS CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

10:15 AM – 11:45 AM

PLATFORM Sessions 17-24: Concurrent Platform Sessions
See talks and rooms on pages 16-17

11:45 AM – 1:15 PM

LUNCH & NETWORKING BREAK Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

SPECIAL SESSION Career Development Panel: How to Set Yourself Apart
Room 706 (ticket required)

SPECIAL SESSION Sustaining Diversity, Equity, and Inclusion Efforts in Human Genetics & Genomics Research and the Workforce during Polarizing Times
Room 711 (ticket required)

12:00 PM – 1:00 PM

INDUSTRY SOLUTIONS Industry Education Sessions Presented by Industry Experts
100s Hall

12:00 PM – 1:15 PM

INDUSTRY SOLUTIONS CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

Wednesday, November 6

1:15 PM – 2:15 PM
PLATFORM — Sessions 25-32: Concurrent Platform Sessions
See talks and rooms on pages 18-19

2:30 PM – 4:30 PM
COFFEE BREAK — Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

POSTERS — Poster Session I
Exhibit & Poster Hall/Upper Level

INDUSTRY SOLUTIONS — CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

3:00 PM – 4:00 PM
INDUSTRY SOLUTIONS — Industry Education Sessions Presented by Industry Experts
100s Hall

3:00 PM – 4:15 PM
SPECIAL SESSION — ASHG Policy Forum: The Next Frontier of AI/ML in Human Genetics/Genomics
Room 104

4:30 PM – 5:00 PM
AWARDS — Session 33: Awards Recognition I
Mile High Ballroom

5:00 PM – 6:30 PM
PLENARY — Session 34: Presidential Symposium: Mendelian Traits: Thinking about Complexity in the World of "Simple" Genetics
Mile High Ballroom

7:00 PM – 8:30 PM
NETWORKING — Diversity, Equity, and Inclusion Reception
Hyatt Regency Hotel, Capitol 1 Room (ticket required)

Thursday, November 7

8:00 AM – 9:30 AM

FEATURED SYMPOSIUM	Session 35: Advances in Artificial Intelligence Tools to Improve Clinical Diagnoses and Medical Genetics Education with an Emphasis on Diverse Datasets *Four Seasons Ballroom 2&3*
FEATURED SYMPOSIUM	Session 36: AJHG at 75: Looking to the Future of Human Genetics Research *Room 401*
FEATURED SYMPOSIUM	Session 37: Balancing Open Science and Patient Privacy in the Era of Precision Medicine *Room 405*
FEATURED SYMPOSIUM	Session 38: Contributions of Tandem Repeats to Human Variation, Traits, and Disease *Four Seasons Ballroom 4*
FEATURED SYMPOSIUM	Session 39: Human Genetic Mosaicism: Diversity within Individuals *Room 505*
FEATURED SYMPOSIUM	Session 40: The Alzheimer's Disease Sequencing Project (ADSP): A Paradigm for Identifying Genetically Driven Therapeutics for a Global Complex Disease *Four Seasons Ballroom 1*
FEATURED SYMPOSIUM	Session 41: TOPMed 10-Year Anniversary: Ongoing Success and Future Directions *Mile High Ballroom 2&3*
FEATURED SYMPOSIUM	Session 42: Unveiling Genetic Mysteries: RNA Editing's Breakthrough in Disease and Gene Therapy *Room 501*

9:30 AM – 10:15 AM

COFFEE BREAK	Visit Industry Partners and Posters *Exhibit & Poster Hall/Upper Level*
NETWORKING	Shared Interest Group Meetup: Bioinformatics & Computational Methods *Room 712*

Thursday, November 7

9:45 AM – 10:15 AM

INDUSTRY SOLUTIONS — CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

10:15 AM – 11:45 AM

PLATFORM — Sessions 43-50: Concurrent Platform Sessions
See talks and rooms on pages 20-21

11:45 AM – 1:15 PM

LUNCH & NETWORKING BREAK — Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

NETWORKING — Career Development Topics: A Networking Event
Room 706 (ticket required)

SPECIAL SESSION — Addressing the Challenges of Polygenic Scores in Human Genetic Research
Room 712 (ticket required)

12:00 PM – 1:00 PM

INDUSTRY SOLUTIONS — Industry Education Sessions Presented by Industry Experts
100s Hall

12:00 PM – 1:15 PM

INDUSTRY SOLUTIONS — CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

1:15 PM – 2:15 PM

PLATFORM — Sessions 51-58: Concurrent Platform Sessions
See talks and rooms on pages 22-23

2:30 PM – 4:30 PM

COFFEE BREAK — Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

POSTERS — Poster Session II
Exhibit & Poster Hall/Upper Level

INDUSTRY SOLUTIONS — CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

3:00 PM – 4:00 PM
INDUSTRY SOLUTIONS
Industry Education Sessions Presented by Industry Experts
100s Hall

4:30 PM – 5:00 PM
AWARDS
Session 59: Awards Recognition II
Mile High Ballroom

5:00 PM – 6:40 PM
PLENARY
Session 60: Featured Plenary Abstract Session II
Mile High Ballroom

7:00 PM – 8:30 PM
NETWORKING
ASHG Social
Hyatt Regency Hotel, Capitol Foyer

Friday, November 8

8:00 AM – 9:30 AM
FEATURED SYMPOSIUM
Session 61: Aging, Clonal Hematopoiesis, and Our Health
Room 505

FEATURED SYMPOSIUM
Session 62: Cross-Examining the Rare and Common Variant Architecture of Psychiatric Conditions, Brain Structure, and Function
Four Seasons Ballroom 1

FEATURED SYMPOSIUM
Session 63: Face the Facts: The Impact of Advances in Data Science on Translational Research
Room 401

FEATURED SYMPOSIUM
Session 64: For the Children: Genomics to Improve the Health of Pediatric Patients and Their Families
Room 501

FEATURED SYMPOSIUM
Session 65: How Do We Describe and Ascribe Clinical Significance to the Non-coding Genome?
Mile High Ballroom 2&3

FEATURED SYMPOSIUM
Session 66: Improving Health Equity in Genomics: Interventions and Implementation Efforts to Address Disparities in Genetic Services Research
Four Seasons Ballroom 2&3

Friday, November 8

8:00 AM – 9:30 AM

FEATURED SYMPOSIUM — Session 67: Model Organisms to the Rescue: Next Generation Animal Models for Precise Phenotyping of Complex Diseases
Four Seasons Ballroom 4

FEATURED SYMPOSIUM — Session 68: Not Only Transcription Intermediates: The Roles of R-loops in Genome Stability and Brain Disease
Room 405

9:30 AM – 10:15 AM

COFFEE BREAK — Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

NETWORKING — Shared Interest Group Meetup: Emerging Laboratory Technologies
Room 712

9:45 AM – 10:15 AM

INDUSTRY SOLUTIONS — CoLab Sessions Presented by Industry Experts
Exhibit & Poster Hall/Upper Level

10:15 AM – 11:45 AM

PLATFORM — Sessions 69-76: Concurrent Platform Sessions
See talks and rooms on pages 24-25

11:45 AM – 1:15 PM

LUNCH & NETWORKING BREAK — Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

SPECIAL SESSION — Behind the Scenes: Publications Workshop Luncheon
Room 712 (ticket required)

NETWORKING — Hiring Mixer in Career Hub
Booth 1130, Exhibit & Poster Hall/Upper Level

12:00 PM – 1:00 PM

INDUSTRY SOLUTIONS — Industry Education Sessions Presented by Industry Experts
100s Hall

1:15 PM – 2:15 PM

PLATFORM Sessions 77-84: Concurrent Platform Sessions
See talks and rooms on pages 26-27

2:30 PM – 4:30 PM

COFFEE BREAK Visit Industry Partners and Posters
Exhibit & Poster Hall/Upper Level

POSTERS Poster Session III
Exhibit & Poster Hall/Upper Level

4:30 PM – 5:00 PM

AWARDS Session 85: Awards Recognition III
Mile High Ballroom

5:00 PM – 6:40 PM

PLENARY Session 86: Featured Plenary Abstract Session III
Mile High Ballroom

Saturday, November 9

8:00 AM – 9:00 AM

PLATFORM Sessions 87-91: Concurrent Platform Sessions
See talks and rooms on pages 28-29

9:30 AM – 10:30 AM

PLATFORM Sessions 92-96: Concurrent Platform Sessions
See talks and rooms on pages 30-31

11:00 AM – 12:30 PM

PLENARY Session 97: Distinguished Speakers Symposium: The Promise and Payoff of Human Genetics and Genomics: Paths from Bench to Bedside
Mile High Ballroom

Wednesday, November 6

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 15: Decoding Structural Variation at Scale	**Session 10: Advancements in Molecular and Cytogenetic Diagnostics**	**Session 14: Cancer Risk: Novel Genes and Mechanisms**	**Session 13: Biobank Scale Genetic Data Resources for Studying Complex and Rare Human Diseases**
8:00 AM	The contribution of linked structural variants to recent positive selection in humans	Somatic overgrowth and vascular malformations: Unveiling novel pathogenic variants and clinical utility through comprehensive genetic testing at Genetic Diagnostic Laboratory (GDL)	An atlas of pan-cancer susceptibility genes revealed by intronic polyadenylation transcriptome-wide association study	100,000 genomes of Europe: Unlocking genetic variability across Europe for science and health
8:15 AM	Haplotype-informed analysis of structural variation in 490,414 genomes and its effects on human health ★	Alport syndrome: Genetic, clinical features and renal transplant outcomes ★	Human lung single-cell eQTL mapping identifies novel cell type-specific genetic control of lung cancer	The UAE genome program: Unique genetic insights from 43,608 individuals
8:30 AM	A phenome-wide association study of tandem repeat variation in 168,554 individuals from the UK Biobank	Assessing the utility of long-read genome sequencing in undiagnosed rare developmental disorders	Exploring gene-by-environment interactions in colorectal cancer risk using massively parallel reporter assays ★	Structural variant discovery with GATK-SV in 97,940 short-read whole genomes from the All of Us Research Program
8:45 AM	Contribution of copy number variation in disease related phenotypes risk in 23andMe research cohort	Inherited metabolic disorders in critically ill patients: Results of genome sequencing of 1,000 consecutive patients from a single centre	Genetic regulation of TERT splicing contributes to reduced or elevated cancer risk by altering cellular replicative potential ★	A complete telomere-to-telomere reference panel of 6404 human haplotypes improves imputation and phasing accuracy
9:00 AM	Pangenome-derived copy number variation maps with global diversity and association analysis in biobank scale data with ctyper	Advanced chromosomal and genomic abnormality detection in hematological malignancies: Leveraging genomic proximity mapping as a next-generation cytogenomics tool	Immune surveillance and cancer risk	Diversity in the NHGRI-EBI GWAS Catalog: Addressing disparities while promoting accessibility and data sharing
9:15 AM	Novel short tandem repeats on the telomere-to-telomere reference genome are associated with Alzheimer's disease neuropathology	Living with your dynamic genome: T2T-CHM13 reference genome identifies Robertsonian translocation carriers in healthy newborn cohorts	Prevalence and effect of inherited chromosomally integrated human herpesvirus 6 in 735,434 human genomes	Harmonizing the world's rare disease knowledge in Mondo

★ *denotes Trainee Research Excellence Award finalist, Resource-Limited Country Travel Award winner, or Human Genetics Scholar (HGSI)*

8:00 AM – 9:30 AM

Room 401	Room 405	Room 501	Room 505	
Session 09: A Heart to Heart on Cardiovascular Genetics in Health and Disease	**Session 16: I See Ghosts: Archaic DNA in Our Genomes**	**Session 11: All the Single Cells**	**Session 12: Beyond Genetic Discoveries: Novel Mechanisms of Neurodevelopmental Disorders**	
Genome-wide association study for resting electrocardiogram in the Qatar Biobank identifies 6 novel genes and validates novel polygenic risk scores	A refined analysis of Neanderthal-introgressed sequences in modern humans with a complete reference genome	Rare and common genetic variants regulate single-cell expression of immune cells from 2,000 individuals ★	Monoallelic de novo variants in DDX17 cause a novel neurodevelopmental disorder	**8:00 AM**
Multi-ancestry GWAS meta-analysis of TG/HDLC ratio in the Million Veteran Program and UK Biobank	Patterns of genomic and morphological variation in the mid-19th century burial remains of the Liberated Africans from St. Helena Island in the South Atlantic	Population-scale single-cell RNA-seq across five countries reveal Asian-specific genetic architecture of alternative splicing and complex disease	Maternally derived de novo variants in the non-coding spliceosomal snRNA RNU4-2 are a frequent cause of syndromic neurodevelopmental disorders	**8:15 AM**
Machine learning enables discovery of rare coding variants in 17 genes for coronary artery disease	Deciphering genetic contribution of ancient hunter-gatherer Jomon in Japanese populations	Interindividual cellular and transcriptional regulatory changes in human aging	Biallelic inactivating variants in DMAP1 underlie a syndromic neurodevelopmental disorder	**8:30 AM**
Deciphering rare non-coding LDL-C associations in over 246K individuals with whole genome sequencing	Characterize the nature of ghost archaic introgression in African populations	Single-cell long-read sequencing analysis in endemic pemphigus foliaceus	Loss-of-function of the Zinc Finger Homeobox 4 (ZFHX4) gene underlies a neurodevelopmental disorder	**8:45 AM**
Proteome-wide Mendelian randomization identifies candidate causal proteins for cardiovascular diseases	Indirect inheritance of archaic ancestry in modern Peruvians	Mapping the observable IBDverse: Identifying novel drivers of IBD susceptibility through population-scale, multi-tissue single-cell eQTL mapping	Biallelic UGGT1 gene variants cause a congenital disorder of glycosylation	**9:00 AM**
Characterising the role of 46 candidate genes in early-stage atherosclerosis using CRISPR/Cas9 and live fluorescence imaging	Archaic introgression in Samoans: Population structure, genetic admixture, and health associations	Single nucleus, multi-ancestry atlas of genetic regulation of gene expression in the human brain	Unveiling the crucial neuronal role of the proteasomal ATPase subunit gene PSMC5 in neurodevelopmental proteasomopathies	**9:15 AM**

Wednesday, November 6

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 22: New Frontiers in Multi-ancestry Methods for Complex Traits	**Session 18: Machine Learning and AI Applications in Human Genetics**	**Session 23: Not Only Genetics: Integrating Other Omics Approaches**	**Session 21: Multimodal Approaches to Interpreting the Non-Coding Genome: Evolution, Functional Genomics, and Machine Learning**
10:15 AM	Multi-trait and multi-ancestry genetic analysis of comorbid lung diseases and traits improves genetic discovery and polygenic risk prediction	CellPhenoX: An explainable cell-specific machine learning method to predict clinical phenotypes using single-cell multi-omics	Single nucleus multiome optimizations for postmortem human brain and large scale multiome profiling of Alzheimer's disease progression reveal novel gene regulatory mechanisms and effects of APOE	Evolutionary conservation and functional analysis of neuronal regulatory elements in mammals
10:30 AM	Dissecting ancestry-aware molecular causal effects for type 2 diabetes	scPrediXcan: Leveraging single-cell data for transcriptome-wide association studies at cell-type level through transfer learning	An atlas of protein quantitative trait loci in Olink and Somascan platforms uncover genetic insights into gastroenterological and hepatological diseases	Uncovering gene regulatory differences between human and chimpanzee neural progenitors
10:45 AM	Quantifying genetic effect heterogeneity across ancestral populations	MILTON: Disease prediction with multi-omics and biomarkers empowers case-control genetic discoveries in UK Biobank	Computational analysis of microbiome genetics in head and neck squamous cell carcinoma	Cross-species variant-to-function analyses implicate insomnia effector genes and reveal a highly conserved regulatory architecture at the MEIS1 locus
11:00 AM	AI-STAAR: An ancestry-informed association analysis framework for large-scale multi-ancestry whole genome sequencing studies	Human data, machine learning, and mouse models demonstrate that SPEN plays a critical role in cardiac development	Prioritizing Alzheimer's disease genetic risk variants with massively parallel reporter assays and 3D chromatin structure	Constructing cell type-specific enhancer-promoter regulatory interaction networks with massively parallel reporter assays
11:15 AM	A multi ethnic meta analysis of genome wide association studies identified additional novel genomic loci associated with cervical cancer ★	Using machine learning to predict noncoding variant associations with sulcal patterns in congenital heart disease	Multi-omic profiling in a 61 day pig kidney to human decedent xenotransplant reveals a concerted acute rejection immune response	Unbiasedly partitioning the heritability of scRNA-seq data reveals that the vast majority of cell type-specific gene regulation lies in trans
11:30 AM	A multi-ethnic reference panel to impute classical and non-classical HLA class II alleles: Enhancing HLA imputation accuracy in admixed populations	Whole exome sequencing shows cystic fibrosis risk variants confer a protective effect against inflammatory bowel disease	Targeted CRISPRa/CRISPRi screen identifies functional variants and novel target genes at multiple renal cell carcinoma (RCC) susceptibility loci	Predicting and interpreting functional non-coding regulatory variants with base-resolution deep learning models of chromatin accessibility

10:15 AM – 11:45 AM

Room 401	Room 405	Room 501	Room 505	
Session 20: Moving Polygenic Risk Scores Closer to Clinical Implementation	**Session 17: Creative Community Engagement: Gathering Data for Better Participatory Research**	**Session 19: Mapping the Brain in Health and Disease**	**Session 24: The Sex-Specific Landscape: Variation, Regulation, and Expression**	
Implementation of breast cancer polygenic risk scores in a personalized screening trial	From barbershop to biopsy: Improving access to genetic screening through the Cleveland African American Prostate Cancer Project	Common variants for migraine tested in 1,138,261 Europeans implicate biological processes with specific effects on head pain severity symptoms	CHD1-deficiency shows sexual dimorphism mediated by androgen exposure	**10:15 AM**
All of Us diversity and scale improve polygenic prediction contextually with greatest improvements for under-represented populations	Co-creating a story-based video collection to engage LGBTQIA+ community members with the All of Us Research Program: An engagement marketing and human entered design approach	Genetic regulation of the gene expression in fetal and adult brains explains GWAS signals from the East Asian population	Sex differences in brain cell-type specific chromatin accessibility in schizophrenia	**10:30 AM**
Polygenic risk score enriches for clinically significant prostate cancer in a screening program - the BARCODE 1 study results	Breaking barriers: Project GIVE's tele-genetic initiative for 100 children with rare diseases at the Texas-Mexico border	Single-cell atlas of transcriptomic vulnerability across multiple neuropsychiatric and neurodegenerative diseases	Disentangling mechanisms underlying sex differences in gene regulation using population-scale multi-omics	**10:45 AM**
Genetic and metabolomic determinants of disease in the UK Biobank	Assessing diverse communities' perspectives of precision research participation: The Precision rEsearCh pArticipatioN (PECAN) study	A nucleotide-scale map of brain cell effects of neurodegenerative GWAS variants reveals distinct and shared causal disease mechanisms	Let's talk about sex: How biological sex affects functional variation across the genome to alter risk of human disease	**11:00 AM**
Performance of contemporary polygenic risk scores for atherosclerotic cardiovascular disease in the All of Us Workbench ★	Genetic services in Africa: Evidence-based recommendations for policymakers and healthcare organizations ★	Cell-cell communication patterns in Alzheimer's disease dementia and mild cognitive impairment vary by cortical layers	Large-scale analyses of variants with sex-biased population allele frequencies, sex-biased association with phenotypes, and sex-biased allele penetrance in 43 human tissues	**11:15 AM**
PGS browser: a comprehensive analysis of 3,168 polygenic score models across 400,000 Finns ★	Balancing constitutional protections in genetic research: Addressing concerns of minoritized communities in non-consented tissue reuse	Mechanisms of presenilin2 driven neuroinflammation: Impact of PSEN2-N141I variant on microglial response to Alzheimer's disease-relevant stimuli	GenESIS: Enhancing transferability of polygenic scores with gene-by-sex interactions	**11:30 AM**

★ *denotes Trainee Research Excellence Award finalist, Resource-Limited Country Travel Award winner, or Human Genetics Scholar (HGSI)*

Wednesday, November 6

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 31: Therapies for Genetic Disorders	**Session 27: Interrogating Variant Function at Scale**	**Session 32: Unifying Multimodalities: Insights from Single Cell Analyses**	**Session 30: Novel Aspects of Modeling Genetic Architectures of Complex Traits**
1:15 PM	A drug repurposing screen identifies NSAIDs and COX1/2 enzyme inhibition as potential therapies for MAN1B1-CDG, a rare congenital disorder of glycosylation	High-throughput deep mutational scanning to determine pathogenicity of variants of uncertain significance in genes in the Sonic Hedgehog Pathway	Leveraging single-cell multi-omic profiling to investigate non-coding variants in Parkinson's disease ★	Selection, pleiotropy, and chance: Why rare and common variant association studies often implicate different genes
1:30 PM	NAGLU co-expressed with a modified phosphotransferase has increased mannose-6-phosphorylation and shows preclinical efficacy as a treatment for mucopolysaccharidosis IIIB (Sanfilipo B Syndrome)	PerturbVI: A scalable latent factor model to infer regulatory modules from large-scale CRISPR perturbation data	Single-cell eQTL analysis in >2,000 individuals in conjunction with single-cell multiomics analysis in 271 individuals infers causal disease mechanisms	Determining the driving factors shaping genetic architecture of complex traits in recently admixed populations
1:45 PM	Antisense oligonucleotide therapy in an individual with KIF1A-associated neurological disorder	Changes in Kv11.1 (hERG/KCNH2) protein interactomes from hiPSC-derived cardiomyocytes of individuals with extreme QT interval polygenic scores and CRISPR edited rare variants	A multiomics single cell atlas redefining the human maternal-fetal interface by spatial cellular mapping	Genomic and ethnolinguistic diversity in >40,000 eastern and southern Africans highlights the ongoing impact of cultural affiliation shaping genetic variation
2:00 PM	Rescue of Proteus syndrome lethality in mice with prenatal miransertib treatment	A shared autophagy pathway dysregulated in multiple neurodegenerative diseases revealed by phenotypic CRISPR screens of iPSC-derived neurons with familial mutations	Identifying noncoding regulatory variants by multiome single-cell sequencing in prostate cells	Detecting ongoing natural selection affecting allele frequencies across generations to uncover genetic variants contributing to disease susceptibilities

★ *denotes Trainee Research Excellence Award finalist, Resource-Limited Country Travel Award winner, or Human Genetics Scholar (HGSI)*

1:15 PM – 2:15 PM

Room 401	Room 405	Room 501	Room 505	
Session 25: Decoding Gene Expression Cis and Trans	**Session 28: Liver, Laugh, Love: New Insights into Liver Disease**	**Session 26: Genetic Approaches Informing Drug Targets and Mechanism**	**Session 29: Modeling Rare Neurodevelopmental Disorders in Human iPSCs and Mice**	
Colocalization of >1,200 skeletal muscle genes with GWAS loci for musculoskeletal and cardiometabolic traits: A muscle eQTL study of 1,002 individuals	Genetic determinants of liver function markers in African ancestry populations	Replication of genetic associations across diverse ancestry groups is indicative of drug target success in clinical trials ★	Rapid generation of mouse model mimicking VUS uncovers novel pleiotropy in neurodevelopmental disorders	**1:15 PM**
Identifying the molecular mechanisms of complex disease through a genome-wide trans-eQTL meta-analysis in 43,301 individuals	Investigating the role of LYPLAL1 loss-of-function in metabolic dysfunction-associated steatotic liver disease	Prioritising new antihypertensive drug targets and unravelling disease modulation by antihypertensive drugs using Mendelian randomisation	Loss of SZT2 leads to an increase in outer radial glia by hyperactivation of mTORC1 in human brain organoids	**1:30 PM**
Polymorphic short tandem repeats shape single-cell gene expression across the immune landscape	Characterizing 99 candidate genes for a role in MASLD and MASH using CRISPR/Cas9, in vivo imaging and deep learning in zebrafish larvae	Identification of plasma proteins as promising therapeutic targets to treat hypertension	Investigating NuRDopathies with GATAD2B-associated Neurodevelopmental Disorder (GAND): Clinical evaluations and modeling with patient-derived IPSCs and mice	**1:45 PM**
Variation and regulatory mechanisms of the small RNA transcriptome across human tissues	Machine learning-based subtyping and validation with longitudinal patient data in metabolic dysfunction-associated steatotic liver disease	Deep learning modeling of rare noncoding genetic variants in human motor neurons defines CCDC146 as a therapeutic target for ALS	Variants in cohesin release factors define a novel class of cohesin balance disorders	**2:00 PM**

Thursday, November 7

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 45: Disease Insights from Omic-Wide Approaches	**Session 44: Alzheimer's Disease from Gene Discovery to Multi-omics Integration**	**Session 50: The Context of All in Which We Live: Gene by Environment Interactions**	**Session 47: From Variant to Function: Prediction and Understanding Variants Function**
10:15 AM	Large-scale genome-wide association study meta-analysis across 1,962,069 individuals reveals insights into the genetic mechanisms of osteoarthritis	Discovering genes associated with Alzheimer's disease via multi-tissue and cell type transcriptome-wide association study	The genetic basis of environmental exposures in the personalized environment and genes study (PEGS)	Defining the function of disease variants with CRISPR editing and multimodal single cell sequencing
10:30 AM	Multi-ancestry proteome-wide Mendelian randomization offers a comprehensive protein-disease atlas and potential therapeutic targets	Integration of GWAS, 3D genomics, and CRISPRi screens in microglia implicates causal variants and genes at Alzheimer's disease loci, including at TSPAN14	Nature versus nurture of glucose homeostasis trajectories in children	Functional genomics applied to mapping the gene regulatory mechanisms downstream of neuron-astrocyte interactions
10:45 AM	Transcriptome-wide association study of early substance use reveals associations between tobacco use and predicted gene expression in adolescents	Deciphering single-cell genomic landscape of brain somatic mutations in Alzheimer's disease	Decomposing sex-different phenotypic correlations in the UK Biobank into genetic and environmental components	CircRNA mediated polyadenylation alteration contribute to Alzheimer's disease pathogenesis
11:00 AM	All by All of Us: Common and rare variant association testing in 245,000 whole genomes across diverse ancestry groups	Large-scale proteomic and genomic analysis identify plasma proteins influencing human brain structure and Alzheimer's disease risk	Neanderthal introgression modifies the response to environmental stimuli in modern humans	In silico module perturbation analysis unlocks a functional understanding of the dynamic gene networks in single-cell data
11:15 AM	Genome-wide association study and predictors of lymphocyte-related blood cell traits in Hispanic/Latino newborns	Unraveling the propagation of functional genetic effects in Alzheimer's disease on a population scale	Assessing cellular contexts of type 2 diabetes-associated variants at scale	De novo precise splice site predictor using deep learning and integration with minimap2 for enhanced long-read sequence alignment
11:30 AM	Omic risk scores are associated with cross-sectional and longitudinal chronic obstructive pulmonary disease-related traits across three cohorts	Astrocytes from diverse ancestries reveal key differences in APOE expression and other AD risk genes across populations	Alternative polygenic score approaches aid in detecting genetic modification of the relationship between adiposity and cardiometabolic risk	Classification of rare nonsynonymous variants to identify individuals at low risk of disease: Introducing variants of potential risk

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10:15 AM – 11:45 AM

Room 401	Room 405	Room 501	Room 505	
Session 48: Novel Genetic, Genomic, and Epigenetic Resources in the Era of Big Data	**Session 43: All about Implementation**	**Session 49: Polygenic Risk Scores: Novel Methods for Modeling Risk**	**Session 46: Diverse Epigenetic Marks in Health, Diagnosis, and Disease**	
The developmental Genotype-Tissue Expression projects	Implementation of hereditary cancer risk assessment in primary care settings: Strategies and proximal outcomes	JointPRS: A comprehensive framework for genetic prediction across populations incorporating genetic correlation and combining meta-analysis and tuning strategies	H3K36 methylation - a guardian of epigenome integrity	**10:15 AM**
The Clinical Genome Resource (ClinGen): Advancing genomic knowledge through global curation	The Million Veteran Program Return Of Actionable Results (MVP-ROAR) Study: Preliminary outcomes from participants receiving clinical genetic confirmation testing for familial hypercholesterolemia	A novel polygenic risk scoring framework integrating common and rare variants for enhanced genetic prediction across ancestries	m6A mediated epitranscriptomic dynamics in human brain development and disease	**10:30 AM**
The New York Genome Center ALS Consortium combines postmortem tissue transcriptomics with whole genome sequencing to empower biological discovery	Introducing an efficient framework to evaluate oncology and cardiology gene-disease validity leveraging clinicogenomic biobank data	Modeling diagnostic code dropout of schizophrenia in electronic health records improves phenotypic data quality and transferability of polygenic risk scores for a diverse Veteran cohort	Most genetic effects on DNA methylation are shared across tissues	**10:45 AM**
FILER 2.0: Unified access to >100,000 omics datasets across >1,000 cell types and tissues	Provider acceptance of patient-facing digital genetics service delivery tools: A qualitative study	A Deep Ensemble Encoder Network method for improved polygenic risk score prediction	Multi-platform long read genomics identifies methylation outliers in rare disease	**11:00 AM**
Whole exome sequencing of 44,028 British South Asians in Genes and Health uncovers 2,917 genes with putative human knockouts for systematic characterization	Does universal testing under payer medical policy equate with genetic testing coverage for patients with ovarian, pancreatic, male breast, and early-onset colorectal cancer?	Integrative polygenic score modeling with tissue-specific annotation improves polygenic scores transferability	A novel single-cell sequencing method for CHD2 variant classification in childhood epilepsies	**11:15 AM**
Enhanced genetic insights from brain region-specific GWAS using deep unsupervised learning derived endophenotypes on UK Biobank T1-weighted MRI data	Costs and outcomes of opportunistic genomic screening: Findings from the Incidental Genomics randomized controlled trial	Functional gene embeddings improve rare variant polygenic risk scores	In-utero rescue of neurological dysfunction in a mouse model of Wiedemann-Steiner syndrome	**11:30 AM**

Thursday, November 7

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 57: Population Genetics Methods Matter	**Session 56: Neurogenomic Approaches Translating Risk Variants to Disease**	**Session 51: 3D Chromatin and Epigenomics**	**Session 52: Computational Methods for Causal Variant Prioritization**
1:15 PM	Characterizing features affecting local ancestry inference performance in diverse admixed populations	Complex structural genome variation in the genetic architecture of neuropsychiatric disorders: Insights from human population analysis and from postmortem brains of individuals with psychiatric disorders	Dissecting the genetic underpinnings of chromatin loops and their relationship to transcriptional regulation	Footprint quantitative trait loci (fpQTLs) reveal non-coding causal variants associated with transcription factor binding for liver traits
1:30 PM	A genealogy-based approach for revealing ancestry-specific structures in admixed populations	Sex differences of the spatiotemporally dynamic FMRP-RNA interactome in the human brain	Comprehensive single-nucleus analysis of genetic regulation on gene expression and chromatin accessibility in human kidneys to understand of genetic basis of chronic kidney disease	A new variant-to-disease score prioritizing causal variants in GWAS
1:45 PM	Deep learning-augmented models of gnomAD v4 enable estimation of LoF mutational constraint for all human genes	ASXL1 mutations drive mitochondrial dysfunction, resulting in disrupted mTOR signaling and cellular proliferation in Bohring Opitz Syndrome	Genetic and epigenetic insights into the aging of the human retina	Robust fine-mapping in the presence of LD mismatch
2:00 PM	Genotype representation graphs: Enabling efficient analysis of biobank-scale data	Translating IGHMBP2 variants with a patient-specific neuromuscular junction system: Personalized medicine rescue	Single-cell genomics, QTLs, and regulatory networks for 388 human brains	Do deep genome language models help pinpoint causal variants in statistically fine-mapped loci?

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1:15 PM – 2:15 PM

Room 401	Room 405	Room 501	Room 505	
Session 58: Scaling Structural Birth Defects	**Session 53: Dysfunction at the Powerhouse: Molecules, Models, and Organisms**	**Session 55: Insights into Somatic Mosaicism and Human Diseases**	**Session 54: Expanding the Table: Considerations for Inclusion in Genetics and Genomics**	
Whole genome analysis of 137 trios with CHARGE-phenotype overlap	Investigating the role of seryl-tRNA synthetase (SARS2) in mitochondrial biology and human recessive disease	A personalized multi-platform assessment of somatic mosaicism in the human frontal cortex	Equity-focused implementation illuminates diverse perspectives in rare disease research	**1:15 PM**
Functional validation of a novel gene associated with orofacial clefts	COXFA4 dysfunction leads to ODC dysregulation: A link to mitochondrial disease mechanism	Reconstructing cell lineage in human brain using somatic mutations in microsatellites	Use of exclusion criteria to select critically ill newborns for rapid genome sequencing captures precise genetic diagnoses missed by use of conventional inclusion criteria	**1:30 PM**
Analysis of rare de novo variants in 5707 congenital heart disease (CHD) trios identifies three novel CHD genes	A multiomic approach to elucidate muscle-specific pathogenesis of SUCLA2-deficient mitochondrial myopathy	Somatic genomic changes in single ischemic human heart cardiomyocytes	Reprogenomics, ethics and inclusivity: Perspectives from sex and gender diverse communities	**1:45 PM**
Unraveling the diverse genetic architecture of structural birth defects	Identification and targeting of ABHD18 as a strategy to alleviate TAZ mutant phenotypes	Genotype-informed single-cell RNA-seq reveals somatic loss of heterozygosity in hemimegalencephaly with PIK3CA mutations	The NIH INCLUDE Project: Over five years of transformational research for people with Down syndrome	**2:00 PM**

Friday, November 8

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 74: The Non-coding Genome: From Nucleotide to Protein	**Session 73: Stats Just Wanna Have Fun: New Methods in Statistical Genetics**	**Session 71: Long-Read Transcriptomes in Health and Disease**	**Session 69: Complex Traits and Other Omics**
10:15 AM	Interpreting regulatory differences between species in terms of potential cis- and trans- mechanisms	Integrative statistical framework for detecting divergent selection and linking to disease	POISEN: A bioinformatics pipeline to identify poison exons in long-read transcriptomes	Genetically predicted leukocyte telomere length from 800,000 individuals identifies novel phenotypic associations
10:30 AM	Nanopore sequencing of chromatin accessibility	Genome-wide assessment of pleiotropy across >1000 traits among >1.5 million participants of diverse biobanks ★	The Spatial Atlas of Human Anatomy (SAHA) project: Unveiling cellular landscapes of health and diseases and orchestrating a new paradigm in precision medicine	Multiomics approach identifies novel genes for Skeletal Class III malocclusion
10:45 AM	BRAIN-MAGNET: A novel functional genomics atlas coupled with convolutional neural networks facilitates clinical interpretation of disease relevant variants in non-coding regulatory elements	Pleiotropic heritability quantifies the shared genetic variance of common diseases	Genome-wide profiling of highly similar paralogous genes using HiFi sequencing	Complex interactions of copy number variants on rare and common disorders
11:00 AM	Variable number tandem repeats (VNTRs) regulate epigenome and transcriptome in human prefrontal cortex	ENCODE cCRE-based WGS analysis of 100 traits in UK Biobank identifies 1,987 associations driven by rare-variants	Combining spatial transcriptomic with snRNA-seq data enhances differential gene expression analyses	Uncovering the nuclear genetic basis of mitochondrial DNA heteroplasmy
11:15 AM	CRISPRi perturbation screens and eQTLs capture different target genes for non-coding GWAS variants	Enhancing regulatory variant prioritization via long-range DNA sequences and multi-task learning	Benchmarking detection of technically challenging pathogenic variants with long-read sequencing and a head-to-head comparison with short-read sequencing in a clinical diagnostic laboratory	A phenome-wide association study of the structural variants in 467,152 UK Biobank genomes identifies non-coding structural variants associated with human diseases
11:30 AM	Connecting rare variation to extremes of plasma protein levels	Trans-modeling of large-scale proteomics data uncovers enriched protein-protein interactions and drug targets	A variety of molecular mechanisms cause copy number gains at 17p11.2 locus causing Potocki-Lupski syndrome: Understanding patients with CNVs that do not include RAI1 gene	Detecting large complex structural variants from human genome assemblies

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10:15 AM – 11:45 AM

Room 401	Room 405	Room 501	Room 505	
Session 76: Translating Genetics into Screening Programs	**Session 75: Tick-Tock: The Aging Genome**	**Session 70: Exploring the Genetic Spectrum of Obesity**	**Session 72: Pharmacogenomics: DNA and Drugs**	
Identification of actionable genetic variants in 4,198 volunteers from the Viking Genes research cohort and implementation of return of results	Cell-type-specific effects of aging on the human prefrontal cortex transcriptome across the lifespan	MRI-Based genetic studies reveal specific genetic variants and disease risks associated with fat distribution across anatomical sites	Incorporation of local ancestry (LA) in a GWAS of warfarin dose requirement in African Americans (AAs) identifies a novel CYP2C19 Splice QTL ★	**10:15 AM**
Early check genome sequencing of newborns to detect genetic risk of type 1 diabetes	Aging-related changes in gene regulation associate with risk, prognosis and therapy response in lung adenocarcinoma	An abdominal obesity missense variant in the transcription factor and thermogenesis gene TBX15 shows signals of adaptation to cold in Finns and affects downstream adipocyte expression in trans	Genome-wide association study on ACE-inhibitor switching identifies missense variants in NTSR1 and CACNA1H	**10:30 AM**
Combining gene genealogies and pedigrees to inform genetic screening programs	Characterization of de novo retrotransposition events in the aging germline	Functional characterization of 14 obesity-associated genes using CRISPR in human white adipose tissue implicates SLTM as a novel lipid accumulation gene	Epigenetic patient stratification via contrastive machine learning refines hallmark biomarkers in minoritized children with asthma	**10:45 AM**
Rate and profile of secondary findings in 381 participants in the DDD-Africa from the DR Congo ★	Linking rare non-coding variants associated with human longevity to cellular senescence via integrative functional genomic approaches	Prioritization of effector genes within body mass index loci yields molecular insight into the biology of body weight regulation	Understanding the impact of drug perturbations on disease-specific protein networks	**11:00 AM**
An association study without genotype sharing for uncovering germline susceptibilities in pediatric cancers	Longitudinal proteomic aging index construction using functional principal component analysis	The phenotypic variability, dose-response, and temporal effects in polygenic prediction of adiposity traits	Prioritization of icosapent ethyl for the potential reversal of metabolic dysfunction associated fatty liver disease using a genetically informed drug repurposing pipeline ★	**11:15 AM**
Biobank-scale genotype-to-phenotype analyses reveal the challenges in using exome sequencing for population screening	Epigenetic age acceleration across chronological age groups and its modifiable lifestyle risk factors in middle-aged and elderly adults	Weight loss with semaglutide is influenced by traditional metabolic risk factors and BMI-associated genetic variants	Combining genetics with real-world patient data enables ancestry-specific target identification and drug discovery	**11:30 AM**

Friday, November 8

	Four Seasons Ballroom 1	Four Seasons Ballroom 2&3	Four Seasons Ballroom 4	Mile High Ballroom 2&3
	Session 80: Linking Non-coding Variation to Function via Diverse Epigenetic Mechanisms	**Session 81: Rare Variants and Admixture Modeling in Diverse Population**	**Session 84: Strategies to Interpret Germline Variants in Cancer Predisposition Genes**	**Session 77: Exploring Omics: From Genomes to Microbiomes**
1:15 PM	Single cell multi-omics and 3D genome architecture reveals novel pathways and targets of metabolic dysfunction-associated steatohepatitis	Large-scale admixture mapping in the All of Us Research Program improves the characterization of cross-population phenotypic differences	Applying scalable machine-learning approaches to generate evidence to impact variants of uncertain significance in Lynch syndrome genes	Institution-wide access to a scalable, clinical grade genomic sequencing platform advanced rare disease research and improved clinical outcomes in a pediatric setting
1:30 PM	Machine learning identifies chromatin features that predict the sensitivity of regulatory sequences to inhibition of BAF chromatin remodeling activity	Multi-ancestry GWAS for hypermobile Ehlers-Danlos Syndrome	When clinical meets molecular: Why, when and how do CTNNA1 germline variants cause hereditary diffuse gastric cancer development	Project Baby Lion - Introducing ultra-rapid genome sequencing in German neonatal and pediatric ICUs
1:45 PM	Response sQTLs in primary human chondrocytes identify novel putative osteoarthritis risk genes	Rare variant associations and fine-scale population structure in the Genes & Health Study of >44,000 British South Asians	Comparing scalable and automated vs. ACMG/AMP variant interpretation strategies for BRCA1 and BRCA2 in a large clinicogenomic cohort from six US-based health systems	Assessing HiFi genome sequencing as first-tier test in rare disease genetics
2:00 PM	Massively parallel reporter assay highlights the importance of B cell activation in uncovering latent QTLs, especially for eQTLs	Network comparison of ancestry-specific genetically correlated diseases in a meta-analysis of phenome-wide association studies from 1 million individuals	Expanding the reach of paired DNA and RNA sequencing: Results from 450,000 consecutive individuals from a hereditary cancer cohort	Expanding the human gut microbiome atlas of Africa

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1:15 PM – 2:15 PM

Room 401	Room 405	Room 501	Room 505	
Session 78: Genetics of Human Brain: Regulation, Disease Risk, and Assortative Mating	**Session 79: Lessons from Height**	**Session 82: Read All about It: Transcriptomic Insights from New Sequencing Technologies**	**Session 83: Splice Splice Baby: Isoform Expression in Health and Disease**	
Establishing the molecular foundation of brain anatomy in living individuals	Leveraging whole-genome sequencing data from 750,000 diverse-ancestry individuals across biobanks to understand the genetic architecture of common anthropometric traits	Identifying pathogenic variants that cause Mendelian conditions using long-read transcript sequencing	An atlas of expressed transcripts in the prenatal and postnatal human cortex ★	**1:15 PM**
Mapping the regulatory effects of rare non-coding variants across cellular and developmental contexts in the brain	Impact of rare coding variants on height prediction in a diverse set of >1 million individuals	Single-Cell Omics for Transcriptome CHaracterization (SCOTCH): Isoform-level characterization of gene expression through long-read single-cell RNA sequencing	Uncovering the brain-specific genetic regulation of splicing by mapping splicing quantitative trait loci in 10,887 post-mortem brain RNA-seq samples	**1:30 PM**
The largest to-date exome study of autism spectrum disorder triples the number of autism-associated genes	Machine learning reveals 3D regulatory mechanisms for height-associated haplotypes	Applications of long-read RNA sequencing improves the design and interpretability of RNA-based therapeutics	A high throughput splicing assay for characterization of rare variants of unknown significance	**1:45 PM**
Assortative mating across nine psychiatric disorders: Consistency and persistence across cultures and generations	GWAS of infant and early childhood height in up to 70 000 children: Genetic influences on the early phases of childhood growth	Combined long- and short-read RNA sequencing of pathogen stimulated primary immune cells identifies the expression of uncharacterized genes and transcripts	Defining the landscape of poison exon splicing events in the human brain: Implications for neurodevelopmental and neurodegenerative disorders	**2:00 PM**

Saturday, November 9

	Four Seasons Ballroom 2&3	Room 401	Room 405
	Session 90: Tumor Genome Landscape Studies	**Session 91: Unraveling the Complexity of Polygenic Inheritance**	**Session 88: Keeping It REnAL! Genetic Studies of Kidney Disease**
8:00 AM	Integrated gene expression analysis reveals SIGLECs as new targets for immunotherapy in head and neck squamous cell carcinoma ★	Beyond known genes and relationships for craniofacial abnormalities	GWAS of multiple renal function biomarkers and kidney multi-omics prioritizes new chronic kidney disease genes
8:15 AM	A common missense polymorphism in the PARP1 gene is associated with distinct tumor transcriptomic, immune and clinical profiles in high grade serous ovarian cancers	Non additive interactions between rare variants and lifestyle factors contribute to obesity	Characterization of a novel ASAH2 variant associated with diabetes and kidney failure in Tongan and Samoan patients
8:30 AM	Characterization of the immunosuppressive microenvironment driven by HBV-infected tumor cells in hepatocellular carcinoma through single-cell sequencing ★	Polygenic risk of rheumatoid arthritis regulates the abundance of circulating regulatory T cells	KidneyGenAfrica: Putative novel genetic loci and improved polygenic prediction for kidney function derived from aggregating 10 continental African genome-wide association studies ★
8:45 AM	Single-cell RNAseq revealed multiple resistance mechanisms in patient-derived xenograft model of rectal cancer during treatment	An atlas of associations between plasma proteins biomarkers and polygenic risk scores for cancer and other complex human diseases	SLC6A19 loss of function is associated with improved kidney function and metabolic reprogramming of kidney cells

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8:00 AM – 9:00 AM

Room 501	Room 505	
Session 87: Framing Heritability for Complex Traits	**Session 89: Long-Read Sequencing Offering New Insights into Neurological Disease**	
Fine-mapped insertions and deletions disproportionately impact 78 diseases and complex traits	Long-read sequence and assembly of autism reference genomes	**8:00 AM**
Heritability and effect-size distribution of rare and de novo protein-coding variation	Long-read sequencing to diagnose autosomal recessive Parkinson's disease in diverse populations	**8:15 AM**
Uncovering the contribution of rare variants to the heritability of complex traits: Insights from the UK Biobank whole genome sequencing data	Mapping parent-of-origin methylation pattern during development by long-read 5-base HiFi sequencing reveals novel imprinting motifs and insight into human disease	**8:30 AM**
Partitioning genetic and non-genetic contributions to epigenetic-defined endotypes of allergic phenotypes in children	Identification of FXN protomutation alleles explains the unequal population distribution of Friedreich ataxia	**8:45 AM**

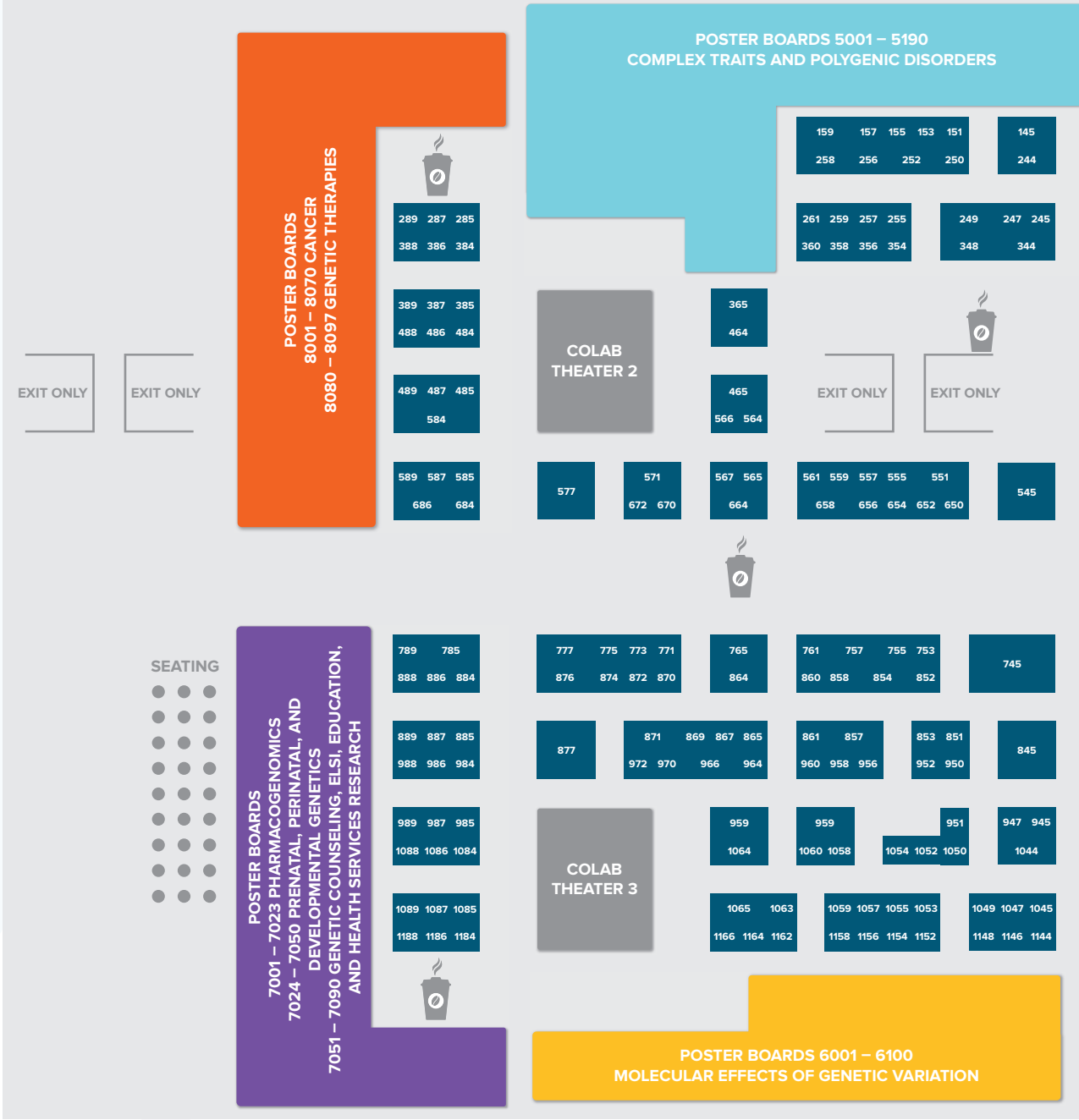
Saturday, November 9

	Four Seasons Ballroom 2&3	Room 401	Room 405
	Session 95: Phenomenal PheWAS	**Session 94: More than One Way to Break a Gene - Variant Effects on RNA**	**Session 96: Technology for Translation**
9:30 AM	Genome-wide association studies in a large Korean cohort identify novel quantitative trait loci for 36 traits and illuminate their genetic architectures	Unveiling the hidden role of RNA stability as a link between genetic variation and disease	A randomized study of a digital genetic health portal (MyCancerGene) for patients who have received germline cancer genetic test results as compared to usual care
9:45 AM	The phenomic landscape of gain- and loss-of-function genetic variants across diverse human populations	Splice Switch: An investigation on the effect of a sQTL on PAPR2 isoforms and subsequent influenza A virus susceptibility	GenAI-powered approaches in advancing genetic testing education and communication: An exploratory study in pharmacogenomics
10:00 AM	Phenome-wide study reveals multiple diseases and biomarkers causally associated with alcohol consumption	Modulation of the impact of genetic mutations on human health by transcriptional adaptation	Machine learning predictions to shorten diagnostic odysseys in Level IV NICUs
10:15 AM	Phenome-wide association of APOE alleles in the All of Us Research Program	Systematic analysis of nonsense variants uncovers peptide release rate as a novel modifier of nonsense-mediated mRNA decay efficiency	The All of Us Research Program data release 2024 (CDR v8): Powering genomic research through All of Us

9:30 AM – 10:30 AM

Room 501	Room 505	
Session 93: Modeling Ataxia and Neuropathy	**Session 92: Genetic Information in Breast Cancer Risk Assessment and Screening**	
Sumo1 mutation modifies behavioral performances in Fragile X associated tremor/ataxia mouse model	Comprehensive genetic risk assessment for breast cancer in a diverse cohort: Preliminary findings from the eMERGE study	**9:30 AM**
SNX13 and SNX14 influence neuronal lipid homeostasis and associate with spinocerebellar ataxia and intellectual disability syndromes	Investigating genotype-estrogen interactions in breast cancer through a combined molecular and epidemiological approach	**9:45 AM**
Investigating R-loop formation as a potential pathomechanism in spinocerebellar ataxia 27B using iPSC-derived GABAergic neurons	Finding the pathogenic variant in the haystack: Using breast-cancer-related family history from electronic health records to identify patients who should be prioritized for genetic testing	**10:00 AM**
Predictive modeling to define the locus heterogeneity of tRNA synthetase-related peripheral neuropathy	Polygenic risk score (PRS) significantly improves breast cancer (BC) risk assessment for diverse ancestries	**10:15 AM**

Exhibit & Poster Hall Map



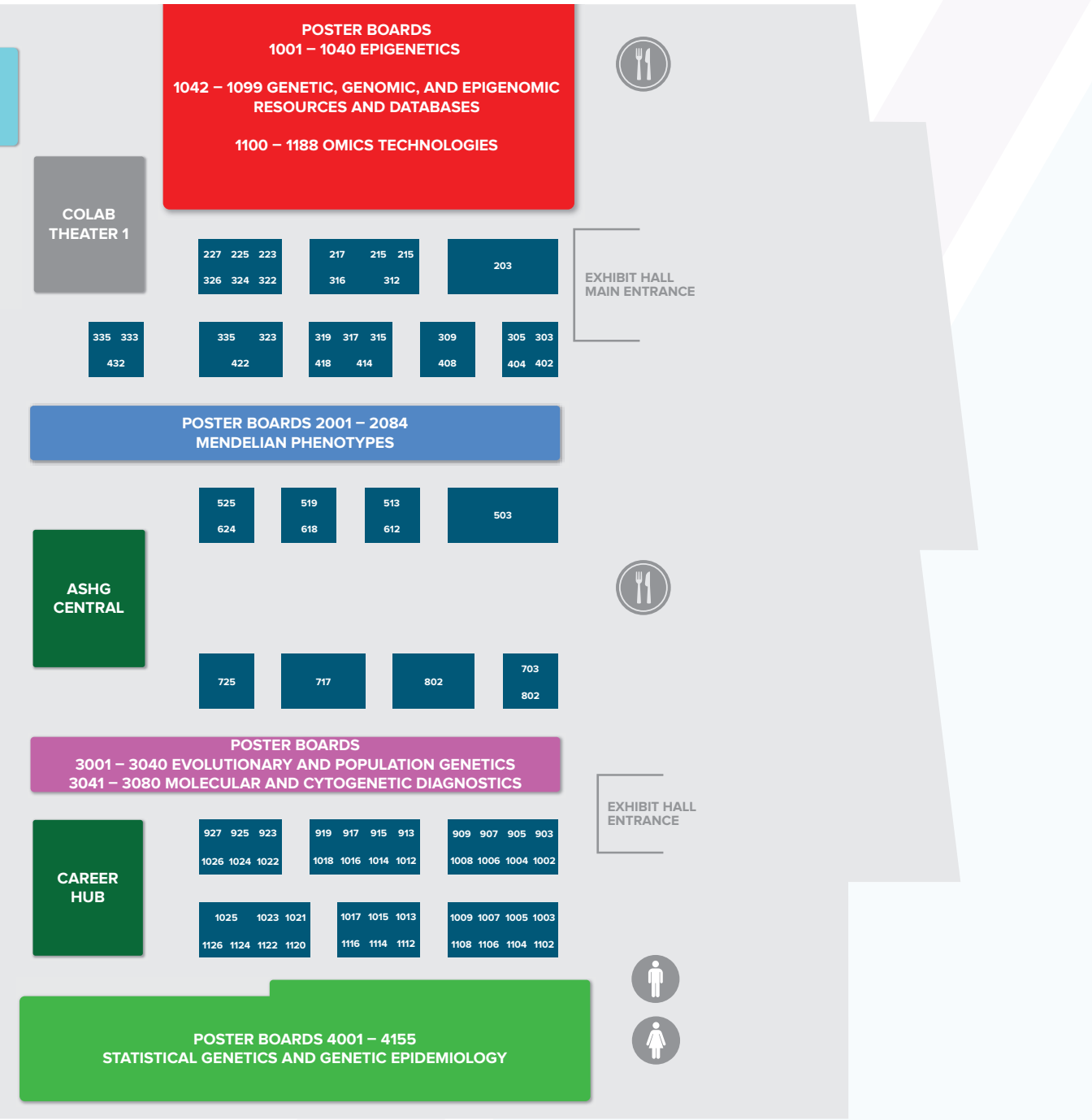

POSTER BOARDS
1001 – 1040 EPIGENETICS
1042 – 1099 GENETIC, GENOMIC, AND EPIGENOMIC RESOURCES AND DATABASES
1100 – 1188 OMICS TECHNOLOGIES
COLAB THEATER 1
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EXHIBIT HALL MAIN ENTRANCE
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POSTER BOARDS 2001 – 2084
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POSTER BOARDS
3001 – 3040 EVOLUTIONARY AND POPULATION GENETICS
3041 – 3080 MOLECULAR AND CYTOGENETIC DIAGNOSTICS
EXHIBIT HALL ENTRANCE
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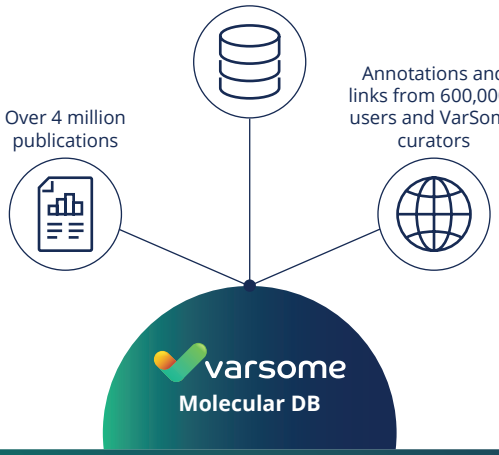
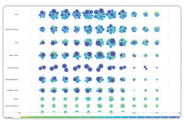


2 Powerful variant classification

Our classifiers provide pathogenicity predictions in **real-time** with **transparency** and **control.**

- each rule is clearly explained
- evidence presented for why a rule is triggered
- weightings of rules can be manually adjusted
- pathogenicity calculated on points-based system, in real-time
- save custom classifications

3 Intuitive and user-friendly data visualization tools



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