ASHG GenomeCast 2025 Annual Meeting Virtual Schedule*

October 14, 2025

4:30pm Opening Presidential Address- <u>Stronger Together: Advancing Human</u> Genetics Through the Power of Community

Special recognition of former National Institutes of Health Director Francis Collins, M.D., Ph.D.

5:00pm Featured Plenary Abstract Session I

- Measuring the functional impact of 5'UTR variants in human disease
- Comprehensive Proteomic Characterization of GLP1R Therapy: Leveraging Mendelian Randomization and Colocalization with Validation in Clinical Trials
- Integrative multi-omics analysis of spaceflight-induced physiological adaptations
- Identifying candidate genes and potential drug targets for central obesity through a multi-population GWAS meta-analysis in the GIANT Consortium
- Comparing the Diagnostic Capability of Large Language Models and Clinical Geneticists

October 15, 2025

8:30am Feature Symposium- Integration of Long-Read Sequencing with Multi-Omics Data to Identify Hidden Causal Variants

This session will be an overview of the benefits of long-read sequencing to identify complex variants and explore the integration of multi-omics data to uncover how these variants are having an effect on gene regulation and disease.

10:45am Platform Session-<u>Alz in the Details: Piecing Together the Alzheimer's</u> Puzzle

This session covers recent advances linking transcriptional states to clinical phenotypes to enhance our understanding of pathogenesis in Alzheimer's disease through epigenetic, single cell, and spatial -omics methods.

1:30pm Platform Session- <u>Single-Cell Multiomic Dissection of Gene Regulation</u> <u>Across Disease States</u>

This session will highlight latest advances in applying single-cell multiomic techniques to uncover the genetic mechanisms of cell-type specific gene regulation within human tissues.

5:00pm Presidential Symposium- <u>Unraveling the Genetic Foundations of Human</u>
<u>Disease: Insights from the Past, Present, and Future</u>

October 16, 2025

7:30am Update on Recent Changes at NIH and NHGRI: What Researchers Need to Know

NHGRI's extramural leadership will highlight recent changes in structure, policies, and priorities at NHGRI and NIH, with a focus on their impact on the research community. The session will cover updated guidance on key topics such as foreign collaborations, data sharing, and funding opportunities, followed by a Q&A.

8:30am Featured Symposium- <u>Artificial Intelligence and Machine Learning Tools</u> <u>Reshaping Modern Genomics</u>

The capabilities of Artificial Intelligence and Machine Learning tools have rapidly progressed, facilitating adoption in discovery and analysis applications in human genomics. This session covers a broad range areas of such application, from disease state prediction and imaging analysis to literature mining and causal variant prediction.

10:45amPlatform Session- Dissecting the Biology of Neurodegenerative Disorders This session will showcase studies investigating the biology of neurodegenerative disorders through different analytical designs.

1:30pm Platform Session- <u>Genetic Insights Fueling Drug Discovery and Precision</u> Health

This session highlights how human genetics is accelerating therapeutic discovery and advancing precision health. Talks will feature cutting-edge approaches--from large-scale exome sequencing to multimodal genetic analyses--that are driving target identification, drug development, and patient stratification.

5:00pm Featured Plenary Abstract Session II

 Supernumerary X chromosomes shape brain organoid architecture and functions in a dose-dependent fashion

- Identification of Unannotated mRNA Isoforms Driving Gene Regulation and Disease from Large-Scale RNA-seq data
- Reimbursement of germline genetic testing for cancer is impacted by socioeconomic characteristics and insurer type
- Pervasive interaction between HLA, KIR, and TCR for autoimmune diseases in > 1 million individuals across diverse populations
- Human-specific tandem repeat in CACNA1C modulates responses to neuronal stimulation

October 17, 2025

8:30am Featured Symposium- <u>Deep Learning for Non-coding Variant</u> <u>Interpretation</u>

Deep learning models are a powerful approach for predicting regulatory activity from genomic sequence and interpreting the cis-regulatory code. In this session, four leading scientists will present their work on leveraging such models to interpret the effects of regulatory variation. These talks will highlight recent advances in deep learning for variant effect interpretation, limitations in existing models, and strategies for improvement.

10:45am Platform Session- The Utility of Al in Clinical Genomics Workflows

This session explores the evolving role of artificial intelligence in clinical genomics, highlighting both its potential and limitations. Presentations will cover applications ranging from literature summarization, variant prioritization and classification to real-world integration in diagnostic workflows.

5:00pm Featured Plenary Abstract Session III

- Haplotype-resolved chromatin differences and genome structural variation
- Accurate representation of globally diverse human haplotypes in the second release of the human pangenome reference
- Local ancestry-specific genetic architecture of breast cancer risk in 40,000 women of African ancestry from the AABCG Consortium
- Age- and BMI-dependent genetic architecture of blood lipids in 2.5 million individuals from globally diverse populations
- The impact of rare deleterious mutations on human lifespan

October 18, 2025

11:15am ASHG Distinguished Speakers Symposium- *Al-Powered Genomics: Transforming Data into Insights*

Artificial intelligence (AI) is revolutionizing the field of genomics, unlocking breakthroughs in both research and healthcare that were once unimaginable. By accelerating the analysis of large genomic datasets, AI uncovers critical patterns that illuminate gene networks, highlight risks for common diseases, and help pinpoint the causes of rare conditions. AI also plays a key role in advancing precision medicine by developing novel treatments, customizing therapies to individual genetic profiles, and predicting patient responses to therapies, all of which enhance diagnostic accuracy and treatment outcomes. However, the growing integration of AI in genomics raises important ethical considerations, including concerns about data privacy, informed consent, and the potential for biased algorithms to perpetuate existing inequalities. As AI continues to shape genomics, it is essential to balance innovation with ethical responsibility to ensure equitable access and outcomes for all patients.