

2026

ACMG Annual  
Clinical Genetics Meeting

MARCH 10-14 • EXHIBIT DATES: MARCH 11-13  
BALTIMORE CONVENTION CENTER • BALTIMORE, MD

Pocket Guide  
SCHEDULE AT-A-GLANCE

Sponsored by:



Welcome to ACMG 2026!



Come visit us at booth  
**#2122**

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# 2026 | ACMG Annual Clinical Genetics Meeting

## 2026 ACMG Annual Clinical Genetics Meeting **SCHEDULE AT-A-GLANCE**

All Scientific Plenary sessions, Scientific Concurrent sessions and Workshops will offer the option to use Artificial Intelligence assisted Closed Captioning. ACMG does not guarantee and does not accept responsibility for the accuracy, reliability, or performance of this service nor the limitations provided by this service, such as the inability to transcribe specific scientific words.

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## Event Schedules

### MEETING REGISTRATION

*Baltimore Convention Center, Pratt Street Lobby, Level 300*

**Tuesday, March 10** | 11:00 am – 7:00 pm

**Wednesday, March 11** | 6:30 am – 6:00 pm

**Thursday, March 12** | 7:30 am – 6:00 pm

**Friday, March 13** | 7:30 am – 6:00 pm

**Saturday, March 14** | 7:30 am – 12:00 pm

### SPEAKER CHECK-IN & READY ROOM

*(Audiovisual Preview)*

*Baltimore Convention Center, Room 327-328*

**Tuesday, March 10** | 8:00 am – 7:00 pm

**Wednesday, March 11** | 6:30 am – 5:00 pm

**Thursday, March 12** | 7:00 am – 6:00 pm

**Friday, March 13** | 7:00 am – 6:00 pm

**Saturday, March 14** | 7:00 am – 12:30 pm

## POSTER VIEWING HOURS

*Baltimore Convention Center, Exhibit Hall C-E*

**Wednesday, March 11** | 5:00 pm – 7:00 pm

**Thursday, March 12** | 10:30 am – 11:30 am  
(odd number presentations)

**Friday, March 13** | 10:30 am – 11:30 am  
(even number presentations)

## EXHIBIT HALL HOURS

*Baltimore Convention Center, Exhibit Hall C-E*

**Wednesday, March 11** | 5:00 pm – 7:00 pm

**Thursday, March 12** | 10:00 am – 4:30 pm

**Friday, March 13** | 9:30 am – 1:00 pm



## INDUSTRY SUPPORTED SATELLITE SYMPOSIA

*Hilton Baltimore Inner Harbor, Holiday Ballroom*

### Tuesday, March 10 | 4:00 pm – 5:30 pm

Sponsor	Room
Denali Therapeutics	Ballroom 6
Oxford Nanopore Technologies	Ballroom 1-3
Rhythm Pharmaceuticals, Inc.	Ballroom 4-5

### Tuesday, March 10 | 6:15 pm – 7:45 pm

Sponsor	Room
BioMarin Pharmaceutical, Inc.	Ballroom 6
UCB	Ballroom 4-5

### Wednesday, March 11 | 8:00 am – 9:30 am

Sponsor	Room
Acadia Pharmaceuticals, Inc.	Ballroom 1-3
Amicus Therapeutics	Ballroom 4-5
Sanofi	Ballroom 6

## INDUSTRY SUPPORTED SATELLITE SYMPOSIA

*Baltimore Convention Center, Level 300*

**Tuesday, March 10 | 4:00 pm – 5:30 pm**

<i>Sponsor</i>	<i>Room</i>
BioMarin Pharmaceutical, Inc.	339-342

**Wednesday, March 11 | 8:00 am – 9:30 am**

<i>Sponsor</i>	<i>Room</i>
Mirum Pharmaceuticals	339-342

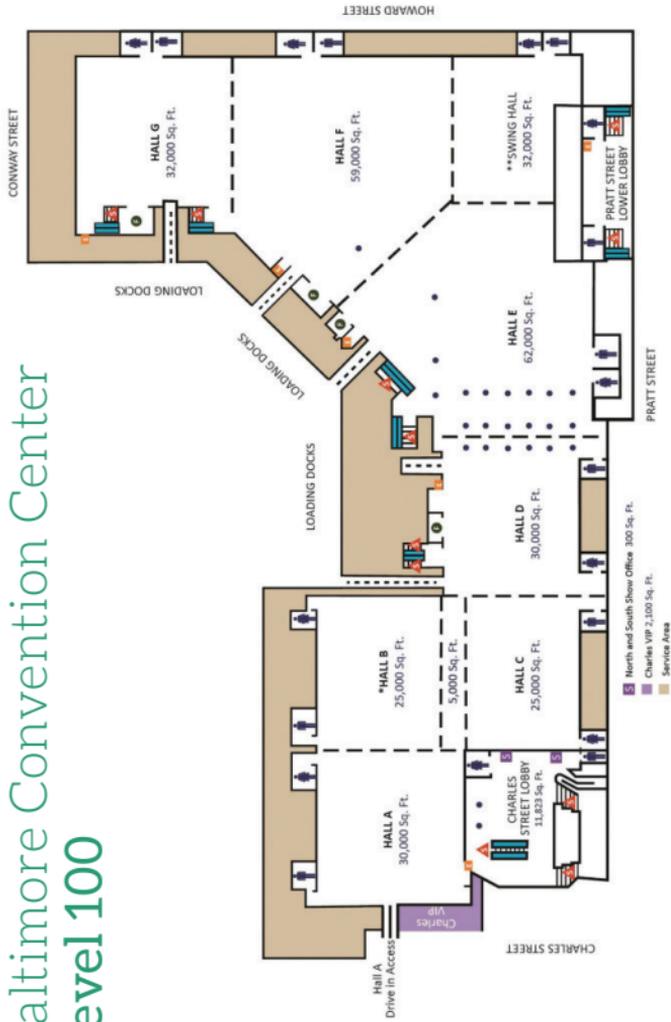
## ACMG FOUNDATION CORPORATE PARTNER INSIGHTS SESSIONS

*Baltimore Convention Center, Level 300*

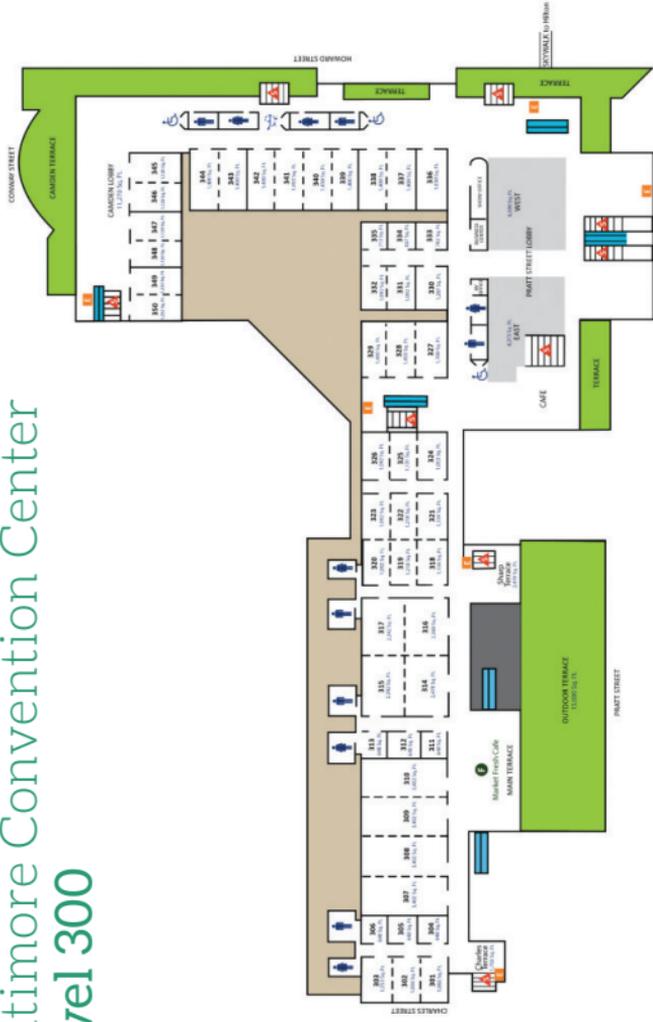
**Wednesday, March 11 | 12:15 pm – 1:15 pm**

<i>Sponsor</i>	<i>Room</i>
BioMarin Pharmaceutical Inc.	337-338
Illumina Inc.	339-340
MyOme	341-342
Natera	343-344
PTC Therapeutics, Inc.	324-326

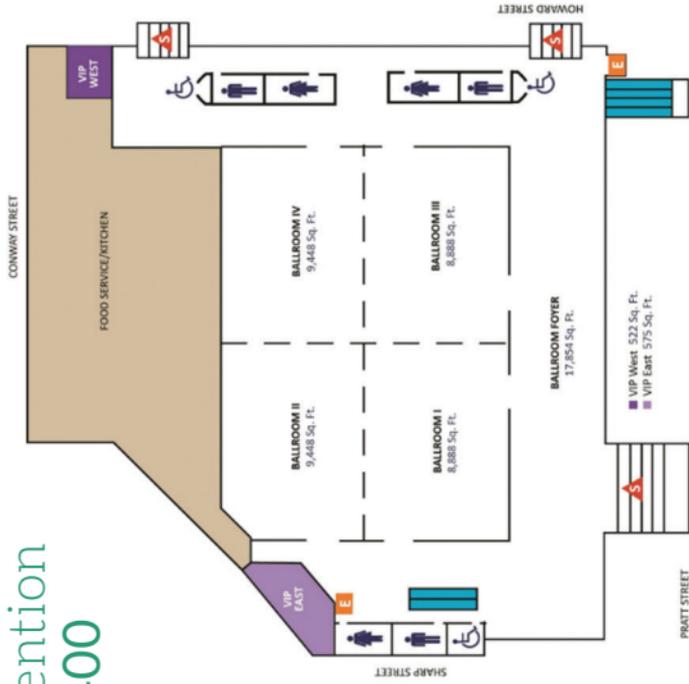
# Baltimore Convention Center Level 100



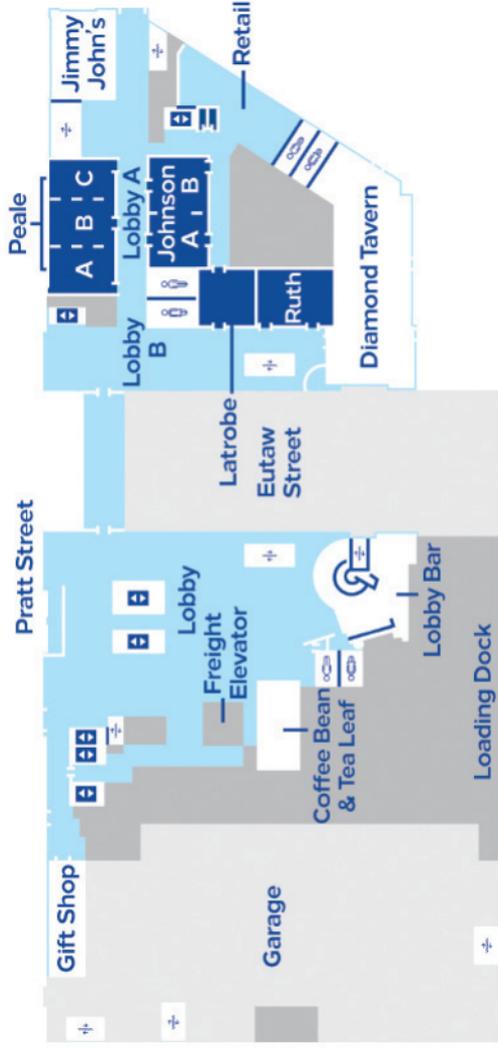
# Baltimore Convention Center Level 300



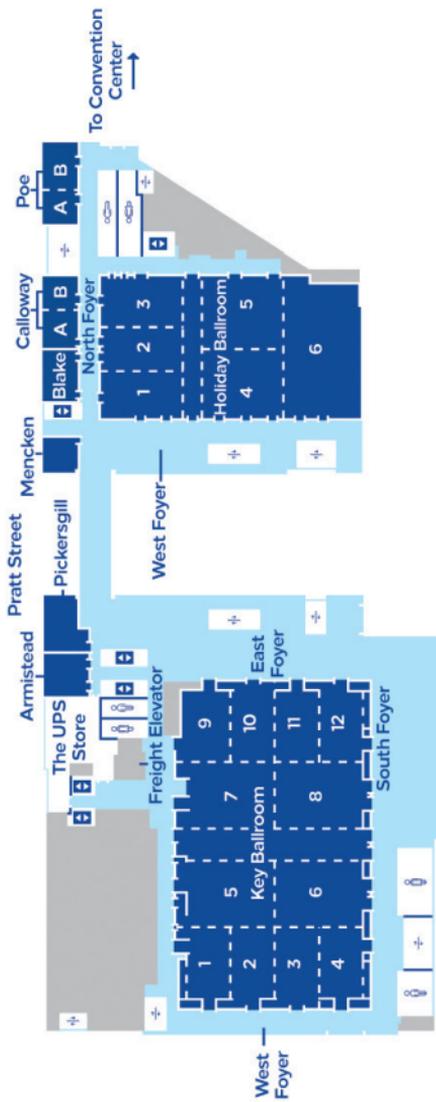
# Baltimore Convention Center – Level 400



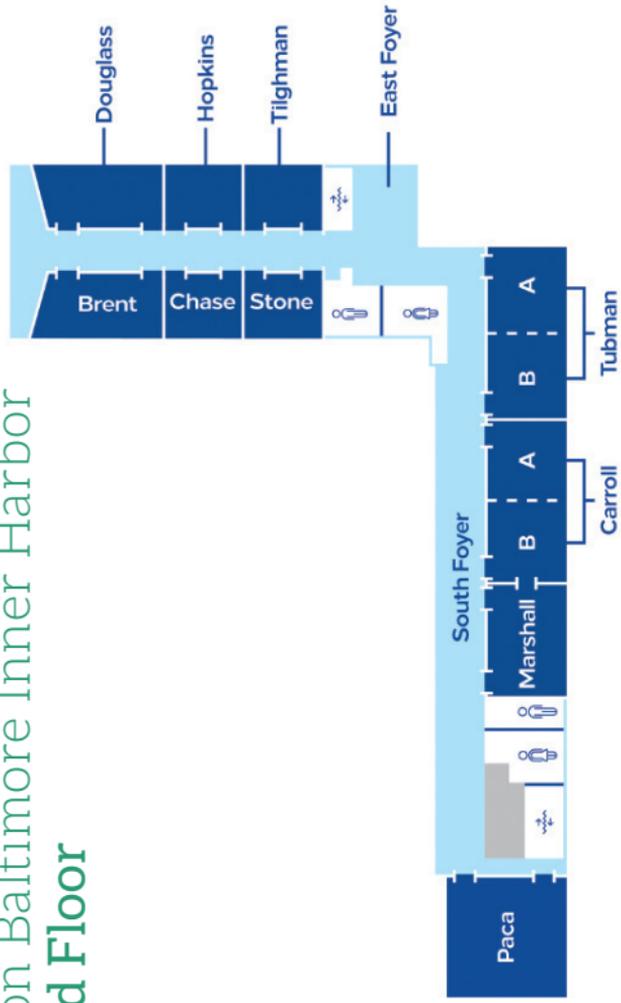
# Hilton Baltimore Inner Harbor – First Floor



# Hilton Baltimore Inner Harbor – Second Floor



# Hilton Baltimore Inner Harbor Third Floor





## **TUESDAY, MARCH 10**

**12:00 pm – 5:00 pm**

### **WORKSHOP**

- College of American Pathologists (CAP) Inspector Training Seminar (pre-registration through CAP is required)

*Baltimore Convention Center, Level 300, Room 336*

**12:30 pm – 5:00 pm**

### **WORKSHOPS**

- Genetic Therapies Bootcamp: Foundations, Frontiers, and Clinical Engagement (additional registration fee applies)
- Medical Genetics in Ophthalmology: Through the Lens of Genetics-Informed Therapies (additional registration fee applies)

*Baltimore Convention Center, Level 300, Room 318-323*

*Baltimore Convention Center, Level 300, Room 324-326*

**1:30 pm – 3:30 pm**

### **SCIENTIFIC SESSION**

- Beyond Uncertainty: Using Functional Assays for Clinical Variant Classification

*Baltimore Convention Center, Level 300, Room 307-310*

**4:00 pm – 5:30 pm**

### **INDUSTRY SUPPORTED SATELLITE SYMPOSIA**

*(Pre-registration through the sponsoring company is required)*

- Exploring the Clinical Spectrum of Hypochondroplasia: From FGFR3 Variants to Multidisciplinary Management (This activity is supported by an independent educational grant from BioMarin Pharmaceutical, Inc.)  
*Baltimore Convention Center, Level 300, Room 339-342*
- Transforming Patient Care in MPS II (This activity is sponsored by an independent educational grant from Denali Therapeutics)  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 6*
- Unrestricted Disease Insights with Comprehensive Long-read Sequencing (This activity is sponsored by an independent educational grant from Oxford Nanopore Technologies)  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 1-3*
- Update on Bardet-Biedl Syndrome (BBS) Diagnostic Criteria (This activity is sponsored by an independent educational grant from Rhythm Pharmaceuticals, Inc.)  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 4-5*

### **SPECIAL INTEREST GROUP FORUMS**

**4:00 pm – 5:00 pm**

- Public Health SIG Meeting  
*Baltimore Convention Center, Level 300, Room 337-338*
- Quality Improvement SIG Meeting  
*Baltimore Convention Center, Level 300, Room 343-344*



### **5:15 pm – 6:15 pm**

- **Adult Genetics SIG Meeting**  
*Baltimore Convention Center, Level 300, Room 337-338*
- **Medical Directors SIG Meeting**  
*Baltimore Convention Center, Level 300, Room 343-344*

### **5:15 pm – 7:00 pm**

#### **JOINT KEYNOTE SESSION AND RECEPTION**

- **The Evolution of Gene Therapy – Milestones, Challenges, and Future Directions (Registration fee applies)**  
*Baltimore Convention Center, Level 300, Room 314-317*

### **6:00 pm – 7:30 pm**

#### **RECEPTION**

- **Welcome Reception for Trainees, Residents, Fellows, and Students**  
*Baltimore Convention Center, Level 300, Camden Lobby*

### **6:15 pm – 7:15 pm**

- **Digital Genetics SIG Meeting**  
*Baltimore Convention Center, Level 300, Room 337-338*

### **6:15 pm – 7:45 pm**

#### **INDUSTRY SUPPORTED SATELLITE SYMPOSIA**

*(Pre-registration through the sponsoring company is required)*

- **Achondroplasia: Current Viewpoints on the Importance of Early Management (This activity is supported by an independent educational grant from BioMarin Pharmaceutical, Inc.)**  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 6*

- Illuminating Advances in Early-Onset TK2d: A Discussion on Genetic Testing & the Patient Journey (This activity is sponsored by an independent educational grant from UCB)  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 4-5*

## WEDNESDAY, MARCH 11

**8:00 am – 9:30 am**

### **INDUSTRY SUPPORTED SATELLITE SYMPOSIA**

*(Pre-registration through the sponsoring company is required)*

- Genetic Mirages: Navigating Pseudodeficiency in the Quest for Diagnostic Clarity  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 6*
- Advancements in Rett Syndrome (RTT) Care: Caregiver and Expert Panel Discussion (This activity is sponsored by an independent educational grant from Acadia Pharmaceuticals, Inc.)  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 1-3*
- Exploring Long-term Data in Certain Adults with Lysosomal Storage Disorders (This activity is sponsored by an independent educational grant from Amicus Therapeutics)  
*Hilton Baltimore Inner Harbor, 2<sup>nd</sup> floor, Holiday Ballroom 4-5*



- Case-Based Approaches in CTX, SLOS & ZSD: Pathways, Tests, and Clinical Decisions (This activity is sponsored by an independent educational grant from Mirum Pharmaceuticals)

*Baltimore Convention Center, Level 300, Room 339-342*

### **WORKSHOP**

- Early Genetics Trainee Forum – Mastering the Challenges of Diagnosing and Managing Overgrowth, Connective Tissue, and Mitochondrial Disorders

*Baltimore Convention Center, Level 300, Room 318-323*

**10:00 am – 12:00 pm**

### **SCIENTIFIC PLENARY SESSION**

- 2026 ACMG Foundation Awards and Presidential Plenary Session - The Future of Expanded Newborn Genomic Screening: Promise and Practice

*Baltimore Convention Center, Level 100, Exhibit Hall AB*

**12:15 pm – 1:15 pm**

### **ACMG FOUNDATION CORPORATE PARTNER INSIGHTS SESSIONS**

- Advancing Genetic Insight from Prenatal Screening Through Rare Disease Diagnosis (sponsored by Natera)

*Baltimore Convention Center, Level 300, Room 343-344*

- Sponsored by PTC Therapeutics, Inc.

*Baltimore Convention Center, Level 300, Room 324-326*

- A New Lens in Rare Disease Diagnosis (sponsored by MyOme, Inc.)  
*Baltimore Convention Center, Level 300, Room 341-342*
- Development and Deployment of Strategies to Aid Genome Interpretation (sponsored by Illumina, Inc.)  
*Baltimore Convention Center, Level 300, Room 339-340*
- Unlocking Possibilities in PKU - A Review of New Clinical Trial Data (sponsored by BioMarin Pharmaceutical, Inc.)  
*Baltimore Convention Center, Level 300, Room 337-338*

#### **PLATFORM SESSION A (NON-ACCREDITED)**

##### *Abstracts presented*

- **O54** - Molecular Insights and Cautionary Tale of Duplications in Duchenne and Becker Muscular Dystrophy Across a Large Cohort
- **O31** - Rare Pathogenic, Polygenic, and Structural Variant Analysis in Sarcomere-negative HCM Patients of African and European Genetic Ancestry From Health Systems
- **O45** - VUS Is not a Four-Letter Word: A Better Understanding of Copy-Number Variants of Unknown Significance From Examining a Large Dataset
- **O26** - From Uncertain to Actionable: Significant Reduction in Variants of Uncertain Significance in Hereditary Germline Testing via Multi-Institutional Real-World Evidence

*Baltimore Convention Center, Level 300, Room 318-323*



**1:30 pm – 3:00 pm**

**CONCURRENT SCIENTIFIC SESSIONS**

- Genetic Counselor Forum - Consanguinity: Counseling, Laboratory and Ethical Considerations in the Genomic Era

*Baltimore Convention Center, Level 400, Ballroom I*

- The New Era of Genomic Oncology: Technology, Complexity, and Clinical Impact

*Baltimore Convention Center, Level 400, Ballroom IV*

- Uniting Science, Standards, and Policy: Collaborative Approaches for Accelerating Pharmacogenomics Adoption

*Baltimore Convention Center, Level 400, Ballroom III*

- When the Trial Ends Early: Perspectives on Premature Clinical Trial Closures in Rare Disease Therapeutics

*Baltimore Convention Center, Level 400, Ballroom II*

**3:00 pm – 3:30 pm**

**REFRESHMENT BREAK**

*Baltimore Convention Center, Level 400, Ballroom Foyer*

**3:30 pm – 5:00 pm**

**CONCURRENT SCIENTIFIC SESSIONS**

- Cardinal Signs

*Baltimore Convention Center, Level 400, Ballroom I*

- Dilemmas of Omic Technologies for Molecular Diagnosis of Rare Disease

*Baltimore Convention Center, Level 300, Room 307-310*

- **False Positives in Biochemical Genetics: Challenges and Cautionary Tales**  
*Baltimore Convention Center, Level 400, Ballroom II*
- **Leading the Field, Coming Out in Front**  
*Baltimore Convention Center, Level 400, Ballroom III*
- **Polygenic Scores and Embryo Screening: Navigating the Clinical and Ethical Applications**  
*Baltimore Convention Center, Level 400, Ballroom IV*

**5:00 pm – 7:00 pm**

**EXHIBITS OPEN**

- **Opening Reception and Exhibits**  
*Baltimore Convention Center, Level 100, Exhibit Hall C-E*
- **First GENEius Challenge Semi-final Round**  
**(6:00 pm – 6:30 pm)**  
*Baltimore Convention Center, Level 100, Exhibit Hall C-E, Learning Lounge*

**THURSDAY, MARCH 12**

**8:00 am – 9:00 am**

**SCIENTIFIC PLENARY SESSIONS**

**Featured Platform Presentations**

*Abstracts presented*

- **O37** – Demystifying Base Large Language Model  
- Reproducibility and Accuracy in ACMG/AMP Variant Classification



- **O18** - Deciphering DHX9 Gene Pathogenicity in Neurological Disorders with Distinct Phenotypes
- **O01** - Clinical Efficacy and Safety Data in Adolescents Treated with Repinatrabit for Phenylketonuria
- **O11** - Prevalence and Tumor Characteristics of Patients with TMEM127 Pathogenic Variants in a Large, Pan-Cancer Cohort

*Baltimore Convention Center, Level 100, Exhibit Hall AB*

**9:00 am - 10:00 am**

### **TED-STYLE TALKS**

- Shoumita Dasgupta, PhD: Mentorship Matters: Harnessing Authentic Connection to Cultivate Tomorrow's Leaders in Genomic Medicine
- Rebecca Ahrens-Nicklas, MD, PhD: It Takes a Village to Advance Genomic Therapies
- Sharon Terry, MA: Beyond Borders and Bias: Genomics for Health in the Hands of Those Who Need It

*Baltimore Convention Center, Level 100, Exhibit Hall AB*

**10:00 am - 4:30 pm**

### **EXHIBITS AND POSTERS**

- Refreshment Break • 10:00 am - 10:30 am
- Speed Mentoring Session • 10:15 am - 11:30 am
- Poster Presentations (odd numbers) • 10:30 am - 11:30 am
- Learning Lounge • 10:30 am - 1:15 pm
- Exhibit Theaters • 10:45 am - 1:00 pm
- Food Court Concession Lunch in the Exhibit Hall • 10:30 am - 1:00 pm

- Refreshment Break • 3:00 pm – 3:45 pm
- Second GENEius Challenge Semi-final Round (3:15 pm – 3:45 pm) in the Learning Lounge

*Baltimore Convention Center, Level 100, Exhibit Hall C-E*

**11:45 am – 1:00 pm**

### **LUNCHEON**

- Early Career Genetic Mentor Luncheon

*Baltimore Convention Center, Level 300, Room 314-317*

**12:00 pm – 1:00 pm**

### **PLATFORM SESSION B (NON-ACCREDITED)**

#### **Clinical Studies and Test Utilization**

*Abstracts presented*

- **O19** – Long-term Efficacy and Safety of DTX401, an AAV8-mediated, Liver-directed Gene Therapy, for the Treatment of GSDIa: Phase 1/2 End-of-Study Results
- **O29** – Results From a Phase 1/2 Open-label Trial of GTX-102, an Investigational Antisense Oligonucleotide for Treatment of Patients With Angelman Syndrome
- **O47** – Utilization and Impact of Exome and Genome Sequencing by Non-Genetics Providers
- **O48** – Insights from Screening Newborns by Genome Sequencing

*Baltimore Convention Center, Level 400, Ballroom I*



## **PLATFORM SESSION C (NON-ACCREDITED)**

### **Laboratory Topics (Methods)**

*Abstracts presented*

- **O50** – Fractional Clues: Decoding NF1 Variant Pathogenicity Through Low Variant Allele Fraction
- **O52** – Long-read Sequencing Enables Accurate Detection and Discovery of Pharmacogenomic Haplotypes
- **O21** – A Rapid, Novel Approach to Rare Disease and Clinical Genetic Variant Discovery Using On-flowcell Proximity Sequencing and Haplotype-resolved Variant Calling
- **O20** – Benchmarking Comprehensive Variant Detection in Challenging Genomic Regions Using Constellation Mapped Read Technology

*Baltimore Convention Center, Level 400, Ballroom III*

**12:30 pm – 1:30 pm**

### **SCIENTIFIC PLENARY SESSION**

- Late Breaking Open Forum: Expanded Newborn Genomic Screening—What's Next?

*Baltimore Convention Center, Level 100, Exhibit Hall AB*

**1:45 pm – 3:15 pm**

### **CONCURRENT SCIENTIFIC SESSIONS**

- Advances in Genomics With AI: From Patient Selection to Variant Analysis and Beyond

*Baltimore Convention Center, Level 300, Room 307-310*

- Do Not Miss Treatable Inborn Errors of Metabolism  
*Baltimore Convention Center, Level 400, Ballroom IV*
- Non-invasive Fetal Sequencing (NIFS) Is Coming: Are You Ready?  
*Baltimore Convention Center, Level 400, Ballroom II*

**4:00 pm – 5:30 pm**

### **CONCURRENT SCIENTIFIC SESSIONS**

- Beyond the Genetics Clinic: Frontline Perspectives on Integrating Genomics in Specialty Care  
*Baltimore Convention Center, Level 300, Room 318-323*
- The Laws of the Ring: Diagnostic Algorithm, Formation Mechanisms, and Clinical Consequences  
*Baltimore Convention Center, Level 300, Room 307-310*

### **PLATFORM SESSION 1**

#### **Hereditary Cancer and Adult Genetics**

*Abstracts presented*

- **O12** – Comprehensive Pan-Cancer Assessment of the APC I1307K Variant Reveals Ancestry-Independent Colorectal Cancer Risk
- **O15** – Cancer Phenotypes of Hypomorphic Versus Non-Functional TP53 Variants in Li-Fraumeni Syndrome
- **O13** – Prevalence of Cancer Predisposition Variants in a Pediatric Pineoblastoma Cohort
- **O16** – Expanded Germline Cancer Predisposition Testing Increases Diagnostic Yield Across Cancer Types

- **O28** - Hidden Genetic Risk: Unanticipated Indications for Testing in an Adult Genomic Health Clinic
- **O43** - High Diagnostic Yield, Unequal Access, and Emerging Guidelines in Adult Genetics: Results and Insights From an 8-year, 7,000-Patient Analysis

*Baltimore Convention Center, Level 400, Ballroom I*

## **PLATFORM SESSION 2**

### **Biochemical / Metabolic**

*Abstracts presented*

- **O02** - Survival, Clinical, and Biomarker Data Support the Efficacy of Intrathecal Adrabetadex in Niemann-Pick Disease Type C
- **O05** - Defining the Genotype-CRIM Status Relationship in a Large Cohort of 215 Patients With Infantile-onset Pompe Disease
- **O07** - Inhibiting Wnt/ $\beta$ -catenin-mTOR Signaling Enhances Porphyrin Clearance in Porphyrrias with Hepatic Involvement
- **O06** - Multi-Omic Evaluation of Glycosylation Restoration Therapy in SLC35A2-CDG
- **O09** - Sepsiapterin Responsiveness Over 14 Days in Children and Adults With Phenylketonuria: Pooled Results From Three Phase 3 Clinical Trials
- **O63** - Reproductive Carrier Screening for Metabolic Conditions With Heterozygous Manifestations

*Baltimore Convention Center, Level 400, Ballroom II*

### PLATFORM SESSION 3

#### Integrative Genomics and Translational Discovery

##### *Abstracts presented*

- **O30** - GATAD2B-Associated Neurodevelopmental Disorder (GAND): Clinical Insights and Investigations With Animal and Human Cellular Models of NuRDopathies
- **O39** - Genome x Environment Analysis of Sudden Unexpected Infant Death Unveils Etiologic Heterogeneity and Strong Cannabis and Genetic Disease Risks
- **O44** - Pharmacogenomics Standardization to Promote Global Uptake
- **O64** - Genomic and Functional Dissection of Fetal Brain Abnormalities
- **O59** - Combined Donor and Immune Cell-free DNA Signatures to Enhance Accurate Detection of Cardiac Allograft Rejection
- **O56** - Enhanced Genomic and Prognostic Understanding of Thoracic Aortic Aneurysms/Dissections (TAAD): A Study of 1,340 Early-Onset Patients With Extended Follow-Up

*Baltimore Convention Center, Level 400, Ballroom III*

## PLATFORM SESSION 4

### Genomic Analysis Tools and Workforce

#### Abstracts presented

- **O24** - STELLAR-AIM: State-of-the-Art Genetic Differential Diagnosis by AI Using Knowledge Extraction From Literature, and Metabolomic, Genomic, and Phenotypic Data
- **O60** - A Semi-quantitative Framework Curating Mechanism of Pathogenicity in Monogenic Conditions: A Consensus Effort of the Gene Curation Coalition
- **O57** - The AnVIL Clinical Resource: Empowering Investigators on NHGRI's Cloud Infrastructure
- **O61** - Improving Variant Classification for Genomic Medicine Through Evidence Calibration
- **O38** - Patient-Centered, Multidisciplinary, Centralized, Longitudinal Clinical and Genomic Follow-up: Development and Evaluation in the Undiagnosed Diseases Network (UDN)
- **O40** - The E3 Genomics Pathways Program: Building a Sustainable Workforce in Medical Genetics and Genomics

*Baltimore Convention Center, Level 400, Ballroom IV*

**5:45 pm - 6:45 pm**

### MEETING

- ACMG Membership Business Meeting (members only)  
*Baltimore Convention Center, Level 300, Room 324-326*

**7:30 pm – 9:00 pm**

**RECEPTION**

- ACMG Foundation Donor Recognition Reception  
(by invitation only)  
*The Royal Sonesta Hotel, 2nd Floor, Whitehall Ballroom*

**FRIDAY, MARCH 13**

**8:00 am – 9:30 am**

**CONCURRENT SCIENTIFIC SESSIONS**

- Beyond the List – Scope and Implementation of ACMG Secondary Findings  
*Baltimore Convention Center, Level 400, Ballroom IV*
- Clonal Hematopoiesis, Somatic and Post-Zygotic Mosaicism in Multigene Cancer Testing for Geneticists  
*Baltimore Convention Center, Level 400, Ballroom III*
- Diagnostic Dilemmas From the Undiagnosed Diseases Network  
*Baltimore Convention Center, Level 300, Room 307-310*
- Hot Topics in Washington, DC and Impacts to Medical Genetics  
*Baltimore Convention Center, Level 400, Ballroom II*

**9:30 am – 1:30 pm**

**EXHIBITS AND POSTERS**

- Refreshment Break • 9:30 am – 10:15 am
- Day of Caring Bicycle Presentation • 10:00 am – 10:30 am
- Speed Mentoring Session • 10:30 am – 11:45 am



- Poster Presentations (even numbers) • 10:30 am – 11:30 am
- Learning Lounge • 10:30 am – 1:15 pm
- Exhibit Theaters • 10:45 am – 1:00 pm
- Food Court Concession Lunch in the Exhibit Hall • 10:30 am – 1:00 pm
- Third GENEius Challenge Semi-final Round (12:00 pm – 12:30 pm) in the Learning Lounge

*Baltimore Convention Center, Level 100, Exhibit Hall C-E*

### **1:30 pm – 3:00 pm**

#### **CONCURRENT SCIENTIFIC SESSIONS**

- Demystifying Neurology in Genetic Diagnosis: A Practical Approach for Geneticists  
*Baltimore Convention Center, Level 300, Room 318-323*

#### **WORKSHOP**

- 2026 Student Workshop: Pathways to Careers Within Medical Genetics and Genomics  
*Baltimore Convention Center, Level 300, Room 314-317*

#### **PLATFORM SESSION 5**

##### **New Laboratory Techniques (Long-read, OGM, RNA, PRS)**

*Abstracts presented*

- **O49** – WAFL: A Comprehensive, Multi-omic Computational Workflow for Rare Disease Diagnostics Using Long-read Sequencing
- **O53** – Two Years of Clinical Long-read Sequencing: Yes We Can Phase That – and Yes, It Is Reimbursable

- **O55** - High-Throughput Functional Characterization of Splicing Variants in Inherited Retinal Degenerations Using a Multiplexed Assay Platform
- **O35** - Challenges of RNA Sequencing Data Interpretation for the Reclassification of Genomic Variants of Uncertain Significance
- **O58** - Increased Diagnostic Yield with Optical Genome Mapping in Pediatric Rare Disease Setting: 1-year Clinical Experience at the Greenwood Genetic Center
- **O33** - An LDL-C Polygenic Risk Score Enables Early Identification of Individuals at High Risk for Hypercholesterolemia Who May Otherwise Be Missed

*Baltimore Convention Center, Level 400, Ballroom I*

## **PLATFORM SESSION 6**

### **Cancer and Aneuploidy**

*Abstracts presented*

- **O14** - Improving Detection of Variant MECOM Rearrangements Through Optical Genome Mapping and Repurposed Targeted RNA-Seq
- **O51** - The Co-occurring Cytogenetic Landscape in Pediatric t(12;21)(p13;q22)/ETV6::RUNX1 Positive Acute Lymphoblastic Leukemia
- **O36** - Optical Genome Mapping for Cytogenetic Analysis in Multiple Myeloma: Real-world Evidence
- **P066** - Integrating Tumor-Normal Testing to Clarify the Significance of Adult-Onset Cancer Predisposition Genes in Pediatric Cancer Patients

- **O34** – A Single Center Review of Survival and Clinical Phenotype Among Children With Trisomy 18 and 13
- **O23** – Network for Advancing Sex Chromosome Aneuploidy Research Readiness (NASCARR): A New Consortia of the Rare Disease Clinical Research Network (RDCRN)

*Baltimore Convention Center, Level 400, Ballroom II*

## **PLATFORM SESSION 7**

### **Biochemical / Metabolic**

*Abstracts presented*

- **O25** – Treatment for AD Disorders: Amplifying Protein Expression With Gene-specific Antisense Oligonucleotides Targeting a Common 3'UTR Motif
- **O04** – Growth Outcomes and Safety of Navepegritide in Children With Achondroplasia: Results of the ApproaCH Trial Open Label Extension
- **O03** – Mitochondrial Function in Fibroblasts From a Patient With Congenital NAD Deficiency Due to Biallelic NADSYN1 Variants
- **O22** – A Novel Clinical-Translational Model of Leukoencephalopathy with Calcifications and Cysts Recapitulates the Human Phenotype
- **O08** – Re-evaluation of Genotype-Phenotype Correlations in D-Bifunctional Protein Deficiency in a Phenotypically Diverse Cohort of 27 Natural History Study Participants

- **O10** - Evaluating the Role of Mitochondrial Dysfunction in Muscle Cell Differentiation in an iPSC-derived Skeletal Muscle Model of Barth Syndrome

*Baltimore Convention Center, Level 400, Ballroom III*

## **PLATFORM SESSION 8**

### **Rapid WGS and Newborn Screening**

*Abstracts presented*

- **O46** - Diagnostic Yield of Rapid Genome Sequencing in Critically Ill Infants with Cardiac Indications
- **O62** - Comprehensive Reanalysis Increases the Diagnostic Yield of Rapid Genome Sequencing in Infantile Epilepsy
- **O27** - Diagnostic Yield and Clinical Utility of Rapid Genome Sequencing in an Unrestricted NICU Cohort
- **O32** - BeginNGS: Updates on Advancing Newborn Screening by Genome Sequencing Trial With Federated Learning Models
- **O41** - Genomic and Clinical Correlation of G6PD Deficiency Identified by the GUARDIAN Expanded Genomic Newborn Screening Study
- **O42** - Challenging Paradigms to Advance Rare Disease Screening and Early Diagnosis: Development of a Targeted Metabolomics Panel to Expand Newborn Screening

*Baltimore Convention Center, Level 400, Ballroom IV*



**3:00 pm – 3:30 pm**

**REFRESHMENT BREAK**

*Baltimore Convention Center, Level 300, Charles Terrace & Level 400, Ballroom Foyer*

**3:30 pm – 5:00 pm**

**SCIENTIFIC PLENARY SESSION**

- Gene Therapy in Action: Real-World Implementation in Clinical Genetics

*Baltimore Convention Center, Level 100, Exhibit Hall AB*

**5:30 pm – 7:00 pm**

**DIAGNOSTIC CHALLENGES SESSIONS**

- Challenge the Experts - Pediatric and Prenatal Diagnostic Dilemmas (Rare Knowns and Unknowns)  
*Baltimore Convention Center, Level 300, Room 307-310*
- Laboratory Diagnostic Challenges: Constitutional and Neoplastic Cases in Molecular, Cytogenomic, and Biochemical Genetics Specialties  
*Baltimore Convention Center, Level 300, Room 318-323*
- Medical Diagnostic Challenges (Adult and Cancer)  
*Baltimore Convention Center, Level 300, Room 324-326*

## **SATURDAY, MARCH 14**

**8:00 am – 9:30 am**

### **CONCURRENT SCIENTIFIC SESSIONS**

- **Bridging Diagnosis and Therapy: The Expanding Role of Somatic Testing in Vascular Anomalies**  
*Baltimore Convention Center, Level 400, Ballroom III*
- **Reproductive Carrier Screening: Is It Time for Whole Genome Testing?**  
*Baltimore Convention Center, Level 400, Ballroom IV*
- **Rewriting the Reference: Laboratory and Clinical Applications of the New Human Pangenome Reference**  
*Baltimore Convention Center, Level 300, Room 318-323*

**9:30 am – 10:15 am**

### **REFRESHMENT BREAK**

*Baltimore Convention Center, Level 400, Ballroom I - I*

**9:45 am – 10:15 am**

### **GENEius Challenge Championship Round**

*Baltimore Convention Center, Level 400, Ballroom I - II*

**10:15 am – 12:15 pm**

- **R. Rodney Howell Symposium: Delivering Pharmacogenomics at Scale: Implementation Strategies That Work**  
*Baltimore Convention Center, Level 400, Ballroom I-II*

# 2026 David L. Rimoin Lifetime Achievement Award in Medical Genetics

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2026

ACMG Annual  
Clinical Genetics Meeting

## Resources

*All located in the Baltimore Convention Center*

### Meeting Information Desk

*Level 300, Pratt Street Lobby*

### ACMG Office

*Level 300, Room 301-302*

### Speaker Check-in and Ready Room

*Level 300, Room 327-328*

### Press and Media Room

*Level 300, Room 304*

### Prayer and Meditation Room

*Level 300, Room 311*

### Quiet Room

*Level 300, Room 312*

### Lactation Room

*Level 300, Room 313*

A large graphic at the top of the page features a blue and white DNA double helix on the left, overlapping a teal circle. To the right, a photograph of a city skyline with skyscrapers is visible, partially obscured by a light green circle. In the foreground of the city photo, there are pink cherry blossom trees and green grass.

# 2027 | ACMG Annual Clinical Genetics Meeting

APRIL 13-17 • EXHIBIT DATES: APRIL 14-16  
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## Call for Proposals

Proposal Submission Opens:

**MARCH 14, 2026**

Proposal Submission Closes:

**MAY 8, 2026**

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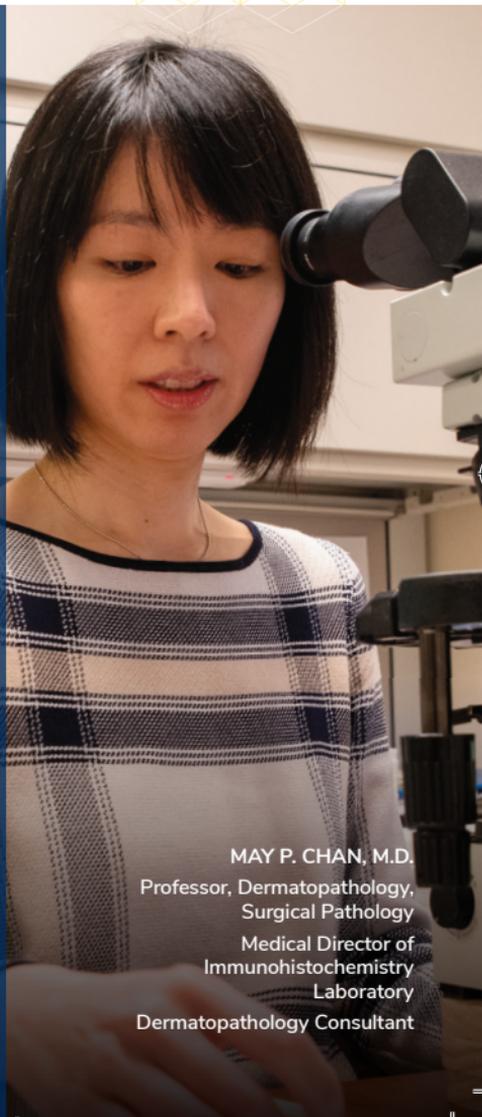
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