2019 ACMG Annual Clinical Genetics Meeting

APRIL 2–6 | EXHIBIT DATES: APRIL 3–5
APRIL 6 | ACMG/SIMD JOINT SESSION
WASHINGTON STATE CONVENTION CENTER | SEATTLE

PROGRAM GUIDE

2019 ACMG Annual Clinical Genetics Meeting • Program Guide

Sponsored by the American College of Medical Genetics and Genomics

Joint Closing Plenary Session with the Society of Inborn Errors of Metabolism
Keep up with RAVICTI® (glycerol phenylbutyrate) Oral Liquid

Have you heard the latest about the expanded indication?

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Navigate the 2019 event like a pro with the Annual Conference mobile app, powered by Core-apps.

With the mobile app, you can:

- **Stay organized** with up-to-the-minute Exhibitor, Speaker, and Event information
- **Build a personalized schedule** and bookmark exhibitors
- **Locate sessions and exhibitors**
- **Read about the speakers**
- **View the abstracts**
- **Network and connect** with your colleagues through Friends
- **Stay in-the-know** and join in on social media
- **Play the Genetics Quest** to win prizes!

**Downloading the App is Easy!**
Search the App store or Google Play for ACMG Annual Meetings

For all other device types (including BlackBerry, Windows, and all other web browser-enabled devices): While on your smartphone, point your mobile browser to http://app.core-apps.com/acmg2019 to be directed to the proper download version for your device.

Should you have any questions, please contact support@core-apps.com
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2019 ACMG Annual Clinical Genetics Meeting 3
Medical Genetics Awareness Week

Through National Medical Genetics Awareness Week, the American College of Medical Genetics and Genomics (ACMG) aims to promote the understanding of the importance of medical genetics professionals, including medical geneticists, laboratory geneticists and genetic counselors on the healthcare team. Medical Genetics Awareness Week is being celebrated for the first time in 2019 to recognize the invaluable contributions that medical genetics healthcare professionals make in the diagnosis, management and prevention of genetic diseases and the difference these professionals make in the lives of patients and families. During this week, which will coincide annually with the ACMG Annual Clinical Genetics Meeting, we take time to recognize the skills and commitment of all those on the healthcare team who translate genetic and genomic discoveries into better patient care.

#MedicalGeneticsAwareness
#IamaMedicalGeneticist
#FutureGeneticsProfessional

www.ACMG.net
### TUESDAY, APRIL 2, 2019

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
<th>Location</th>
</tr>
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<tbody>
<tr>
<td>7:00 am – 12:00 pm</td>
<td>ABMGG Board Exam</td>
<td>205 – Washington State Convention Center</td>
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<tr>
<td>8:00 am – 1:30 pm</td>
<td>CAP Inspection Training Seminar (Ancillary Event)</td>
<td>2AB – Washington State Convention Center</td>
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<tr>
<td>8:00 am – 4:00 pm</td>
<td>ACMG Board Meeting*</td>
<td>Jefferson – Sheraton Grand Seattle</td>
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<tr>
<td>10:00 am – 6:00 pm</td>
<td>Nursing Mothers Room</td>
<td>302 – Washington State Convention Center</td>
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<tr>
<td>10:00 am – 6:00 pm</td>
<td>Prayer and Meditation Room</td>
<td>309 – Washington State Convention Center</td>
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<tr>
<td>10:00 am – 7:00 pm</td>
<td>Registration Open</td>
<td>Atrium Lobby – Washington State Convention Center</td>
</tr>
<tr>
<td>10:00 am – 7:30 pm</td>
<td>Speaker Ready Room</td>
<td>619-620 – Washington State Convention Center</td>
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<tr>
<td>12:30 pm – 5:30 pm</td>
<td>ACMG Short Course: NAMA at the ACMG 3.0 (pre-registration required)</td>
<td>602-604 – Washington State Convention Center</td>
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<tr>
<td>12:30 pm – 5:30 pm</td>
<td>ACMG Short Course: Skeletal Dysplasias Across the Human Lifespan (pre-registration required)</td>
<td>611-614 – Washington State Convention Center</td>
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<tr>
<td>12:30 pm – 5:30 pm</td>
<td>ACMG Short Course: Somatic Mutation Testing in Cancer: Implications for the Oncologist and Geneticist (pre-registration required)</td>
<td>606-609 – Washington State Convention Center</td>
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<tr>
<td>4:15 pm – 5:00 pm</td>
<td>First Time Attendee Orientation</td>
<td>6A – Washington State Convention Center</td>
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<td>5:00 pm – 6:30 pm</td>
<td>ACMG Committee Chairs Meeting*</td>
<td>208 – Washington State Convention Center</td>
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<tr>
<td>5:45 pm – 7:30 pm</td>
<td>Public Health SIG Forum and Business Meeting - Hot Topics 2019: Challenges, Controversies, Conversations</td>
<td>2AB – Washington State Convention Center</td>
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<tr>
<td></td>
<td>Adult Genetics SIG Forum and Executive Committee Meeting – Novel Therapeutics in Adult Genetic Disease</td>
<td>3A – Washington State Convention Center</td>
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<tr>
<td>6:00 pm – 7:30 pm</td>
<td>Welcome Reception for Trainees, Fellows, Residents and Students</td>
<td>2AB Lobby – Washington State Convention Center</td>
</tr>
<tr>
<td>6:00 pm – 9:00 pm</td>
<td>AAP Council on Genetics Executive Committee Meeting (Ancillary Meeting)</td>
<td>Seneca – Sheraton Grand Seattle</td>
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</table>
6:00 pm – 7:30 pm  **SATELLITE SYMPOSIA** (pre-registration required)
Note: Satellite Symposia are not a part of the official ACMG Annual Clinical Genetics Meeting programming.
- Exploring A New Frontier in Cancer Genetics – Clinical Insights and Evolving Concepts in The DNA Damage Response Pathway
  (This activity is sponsored by Creative Educational Concepts and supported by an educational grant from AstraZeneca)
  *Location: 6B – Washington State Convention Center*
- Ex Vivo Gene Therapy for Lysosomal Storage Disorders – The Path from Promise to Patient
  (This activity is sponsored by Avrobio. Educational credits are not provided)
  *Location: 6A – Washington State Convention Center*
- Fabry Disease: Know When to Treat
  (This activity is sponsored by Sanofi Genzyme. Educational credits are not provided)
  *Location: 602-604 – Washington State Convention Center*

7:15 pm – 10:00 pm
- ACMG Lab QA Cytogenetics Subcommittee Meeting*
  *Location: Issaquah B - Sheraton Grand Seattle*
- ACMG Lab QA Biochemical Genetics Subcommittee Meeting*
  *Location: Issaquah A - Sheraton Grand Seattle*
- ACMG Lab QA Molecular Genetics Subcommittee Meeting*
  *Location: Kirkland – Sheraton Grand Seattle*

7:30 pm – 9:00 pm
- ACMG Program Committee Dinner*
  *Location: Loulay Restaurant*
- ACMG Workshop: Using Quality Improvement Tools to Enhance Patient Care and Training in Genetics
  *Location: 611-614 – Washington State Convention Center*
- ACMG Workshop: Addressing Literacy and Language to Equitably Deliver on the Promise of Precision Medicine
  *Location: 606-609 – Washington State Convention Center*

**WEDNESDAY, APRIL 3, 2019**

6:30 am – 6:00 pm  Registration Open
  *Location: Atrium Lobby – Washington State Convention Center*

6:30 am – 6:00 pm  Speaker Ready Room
  *Location: 619-620 – Washington State Convention Center*

6:30 am – 8:30 am  ACMG Maintenance of Certification Committee Meeting
  *Location: 208 – Washington State Convention Center*

7:00 am – 8:00 am  ACMG Genetics and Genomics Review Course Faculty Meeting*
  *Location: 209 – Washington State Convention Center*
7:00 am – 8:30 am  ClinGen Actionability Workgroup Meeting  
**Location:** 303 – Washington State Convention Center

7:00 am – 9:00 am  ClinGen Sequence Variant Interpretation Workgroup Meeting*  
**Location:** 304 – Washington State Convention Center

7:00 am – 9:00 am  ACMG Lab QA Full Committee Meeting  
**Location:** 205 – Washington State Convention Center

7:00 am – 9:30 am  ACMG Professional Practice and Guidelines Committee Meeting  
**Location:** 204 – Washington State Convention Center

7:00 am – 10:00 am  ClinGen Somatic Workgroup Meeting  
**Location:** 307 – Washington State Convention Center

7:30 am – 9:30 am  ACMG Social, Ethical and Legal Issues Committee Meeting  
**Location:** 203 – Washington State Convention Center

8:00 am – 9:30 am  Trainee Workshop: Publishing Your Scholarly Work: Soup to Nuts  
**Location:** 611-614 – Washington State Convention Center

**SATELLITE SYMPOSIA** (pre-registration required)  
Note: Satellite Symposia are not an official part of the ACMG Annual Meeting Programming.

- **Exploring the Genetics and Management of Fabry Disease**  
  (This activity is sponsored by Excel Continuing Education and supported by an educational grant from Sanofi Genzyme)  
  **Location:** 606-609 – Washington State Convention Center

- **The Medically Actionable Genome: Clinical Utility of WGS Through Reproduction, Rare Disease and Oncology**  
  (This activity is sponsored by EXCEL Continuing Education and supported by an educational grant from Illumina)  
  **Location:** 6AB – Washington State Convention Center

- **Diminishing the Burden of Phenylketonuria During Adulthood: New Science & Encouraging Stories Highlighting the Critical Role of Adjunctive Therapy**  
  (This activity is sponsored by Medical Learning Institute, Inc. and PVI, Peerview Institute for Medical Education, Inc. and supported by a grant from BioMarin Pharmaceutical Inc.)  
  **Location:** 602-604 – Washington State Convention Center

- **Are You Ready for Gene Replacement Therapy? Example from Spinal Muscular Atrophy**  
  (This activity is sponsored by The France Foundation and supported by an educational grant from AveXis)  
  **Location:** 6C – Washington State Convention Center

8:00 am – 4:00 pm  Press and Media Room  
**Location:** 618 – Washington State Convention Center

8:00 am – 6:00 pm  **Nursing Mothers Room**  
**Location:** 302 – Washington State Convention Center

- **Prayer and Meditation Room**  
  **Location:** 309 – Washington State Convention Center

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*INDICATES CLOSED OR BY INVITATION ONLY COMMITTEE, BOARD MEETING OR EVENT. IF A MEETING IS NOT CLOSED, ACMG MEMBERS ARE WELCOME TO ATTEND AND OBSERVE IT.*

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2019 ACMG Annual Clinical Genetics Meeting 7
10:00 am – 12:00 pm

**OPENING PLENARY SESSION**
Presentation of the 2019 ACMG Foundation Awards
- ACMG Presidential Plenary Session: Perspectives and Possibilities: Partnering for the Future of Medical Genetics
  Location: Exhibit Hall 4E – Washington State Convention Center

12:00 pm – 1:00 pm

Lunch on your own
AAP Council on Genetics Joint Awards Luncheon (by invitation only)
Location: 201 – Washington State Convention Center

12:00 pm – 1:15 pm

- ACMG Membership Committee Meeting
  Location: 305 – Washington State Convention Center
- ACMG Secondary Findings Maintenance Workgroup Meeting
  Location: 303 – Washington State Convention Center
- ClinGen Ancestry and Diversity Workgroup Meeting
  Location: 307 – Washington State Convention Center

12:15 pm – 1:15 pm

- Industry Platform Presentations
  Location: 6C – Washington State Convention Center
- ACMG Finance Committee Meeting
  Location: 304 – Washington State Convention Center

1:30 pm – 3:30 pm

**CONCURRENT SESSIONS**
- Prenatal Diagnostic Challenges
  Location: 6B – Washington State Convention Center
- The Eyes Have It: Genetics of Ophthalmological Disorders
  Location: 6E – Washington State Convention Center
- Social, Ethical and Legal Perspectives on Emerging Genomic Population Health Initiatives
  Location: 6A – Washington State Convention Center
- Pharmacogenomics – Promises, Potential, Pitfalls, and Practice
  Location: 6C – Washington State Convention Center
- ACMG Interpreting Sequence Variation Guideline Revision Workgroup – Open Forum
  Location: 606-609 – Washington State Convention Center

3:30 pm – 4:00 pm

Refreshment Break
Location: 6ABC and E Lobbies – Washington State Convention Center

4:00 pm – 5:30 pm

**CONCURRENT SESSIONS**
- New ACMG Practice Guidelines from the Professional Practice and Guidelines (PP&G) Committee
  Location: 606-609 – Washington State Convention Center

**ORAL PLATFORM PRESENTATIONS**
- Genetic Counseling Platforms
  Location: 6B – Washington State Convention Center
- Clinical Genetics and Therapeutics Platforms
  Location: 6E – Washington State Convention Center
### Schedule At A Glance

- **Laboratory Genetics and Genomics Platforms**  
  *Location: 6C – Washington State Convention Center*

- **Legal and Ethical Issues; Education**  
  *Location: 6A – Washington State Convention Center*

#### 5:30 pm – 7:30 pm  
**Exhibits Opening and Opening Reception**  
*Location: Exhibit Halls 4ABCD – Washington State Convention Center*

#### 7:30 pm – 9:00 pm  
**GIM Editorial Board Meeting**  
*Location: 209 – Washington State Convention Center*

**ClinGen Reception (Ancillary Event)**  
*Location: Room 606-609 – Washington State Convention Center*

### Thursday, April 4, 2019

#### 6:45 am – 8:00 am  
**ClinGen Hereditary Cancer Workgroup Meeting**  
*Location: 305 – Washington State Convention Center*

#### 7:00 am – 6:00 pm  
**Speaker Ready Room**  
*Location: 619-620 – Washington State Convention Center*

**Registration Open**  
*Location: Atrium Lobby – Washington State Convention Center*

#### 7:00 am – 8:00 am  
- **ACMG Exome Sequencing Clinical Utility Workgroup Meeting**  
  *Location: 204 – Washington State Convention Center*
- **ClinGen Metabolism Workgroup Meeting**  
  *Location: 303 – Washington State Convention Center*
- **ACMG Economics of Genetic Services Committee Meeting**  
  *Location: 205 – Washington State Convention Center*

#### 8:00 am – 4:00 pm  
**Press and Media Room**  
*Location: 618 – Washington State Convention Center*

#### 8:00 am – 6:00 pm  
- **Nursing Mothers Room**  
  *Location: 302 – Washington State Convention Center*
- **Prayer and Meditation Room**  
  *Location: 309 – Washington State Convention Center*

### Plenary Session

#### 8:00 am – 9:00 am  
**Featured Platform Presentations**  
*Location: Exhibit Hall 4E – Washington State Convention Center*

#### 9:00 am – 10:00 am  
**TED-Style Talks**  
*Location: Exhibit Hall 4E – Washington State Convention Center*

- Multimomic Investigation from the Urban Biome to Outer Space
- Next Generation Education Can Save Our Species
- Defective? A Somatic Carrier’s Strategy to Impact Duchenne Muscular Dystrophy

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*Indicates closed or by invitation only committee, board meeting or event. If a meeting is not closed, ACMG members are welcome to attend and observe it.*
9:30 am – 4:30 pm  Exhibit/Poster Hall Open

Exhibit Halls 4ABCD – Washington State Convention Center

10:00 am – 10:30 am  Refreshment Break
10:00 am – 11:30 am  Poster Presentations (Odd Numbered Posters)
10:30 am – 12:45 pm  Exhibit Theater Presentations
10:30 am – 1:15 pm  Learning Lounges
11:30 am – 1:00 pm  Concession Lunch
3:30 pm – 4:00 pm  Refreshment Break

11:00 am – 1:00 pm  ■ ACMG Therapeutics Committee Meeting
Location: 204 – Washington State Convention Center

■ ACMG Foundation Board of Directors Meeting
Location: 209 – Washington State Convention Center

■ ACMG Lab QA Workgroup on the Risk Categorization of LDTs (closed)
Location: 203 – Washington State Convention Center

■ ClinGen Neurodevelopmental Workgroup
Location: 205 – Washington State Convention Center

11:45 am – 1:15 pm  ■ Genetic Counselor’s Luncheon and Forum: Risk Alleles - Discovery, Classification, and Reporting (pre-registration required)
Location: Metropolitan Ballroom AB – Sheraton Grand Seattle

■ Trainee / Mentor Luncheon (Pre-registration required)
Location: 606-609 – Washington State Convention Center

1:30 pm – 3:30 pm  50th Annual March of Dimes Clinical Genetics Conference – From Causes to Care: State of the Art Update of Common Birth Defects
Location: Exhibit Hall 4E – Washington State Convention Center

4:15 pm – 5:45 pm  ■ CONCURRENT SESSIONS

■ Next Level Genomic Data Interpretation: Machine Learning, Methylation Analysis and 3D Mapping
Location: 6E – Washington State Convention Center

■ Advances in the Genetics of Inherited Heart Disease
Location: 6A – Washington State Convention Center

■ The Chicken and the Egg Dilemma in Public Health: Timing and Scope of Genetic Screening as Innovative Therapeutic Interventions Incubate – R. Rodney Howell Symposium
Location: 6B – Washington State Convention Center

■ Seeing Into the Future: Gene Therapy Has Arrived
Location: 6C – Washington State Convention Center

6:00 pm – 7:00 pm  ACMG Annual Membership Business Meeting (ACMG Members Only)
Location: 611-614 – Washington State Convention Center

7:30 pm – 9:00 pm  ACMG Foundation and the David L. Rimoin Donors Recognition Reception (by invitation)
Location: Cirrus Ballroom – Sheraton Grand Seattle
### FRIDAY, APRIL 5, 2019

<table>
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<tr>
<th>Time</th>
<th>Event</th>
<th>Location</th>
</tr>
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</table>
| 7:00 am – 6:00 pm  | **Speaker Ready Room**  
*Location: 619-620 – Washington State Convention Center*  
**Registration Open**  
*Location: Atrium Lobby – Washington State Convention Center* |                                                                         |
| 7:00 am – 8:00 am  | **Informational Session: Laboratory Genetics and Genomics Alternative Pathway Training Program**  
*Location: 602-604 – Washington State Convention Center*  
**ACMG Genomic Screening of Asymptomatic Patients Committee Meeting**  
*Location: 304 – Washington State Convention Center* |                                                                         |
| 7:00 am – 8:30 am  | **ACMG Education and CME Committee Meeting**  
*Location: 307 – Washington State Convention Center*  
**ACMG Population Screening PTC Workgroup Meeting**  
*Location: 303 – Washington State Convention Center* |                                                                         |
| 7:15 am – 8:30 am  | **ClinGen Expert Panel Information Workshop (pre-registration required)**  
*Location: 3AB – Washington State Convention Center* |                                                                         |
| 7:15 am – 8:45 am  | **CONCURRENT SESSION**  
*Note: Satellite Symposia are not an official part of the ACMG Annual Meeting Programming.*  
**Towards Precise Genetic Diagnosis of Primary Mitochondrial DNA Diseases**  
*Location: 6C – Washington State Convention Center* |                                                                         |
| 8:00 am – 4:00 pm  | **Press and Media Room**  
*Location: 618 – Washington State Convention Center* |                                                                         |
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<th>Time</th>
<th>Event</th>
<th>Location</th>
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| 8:00 am – 6:00 pm | Nursing Mothers Room<br>
*Location: 302 – Washington State Convention Center* |                                   |
|              | Prayer and Meditation Room<br>
*Location: 309 – Washington State Convention Center* |                                   |
| 9:00 am – 10:00 am | **PLENARY SESSION**<br>-
**A Tribute to Arno G. Motulsky, MD: A Giant of Medical Genetics**<br>
*Location: Exhibit Hall 4E – Washington State Convention Center* | ACMGLIVE   |
| 9:30 am – 1:00 pm | **Exhibit/Poster Hall Open**<br>
*Exhibit Halls 4ABCD – Washington State Convention Center* |                                   |
|              | 10:00 am – 10:30 am **Refreshment Break**<br>10:30 am – 11:00 am **ACMG Day of Caring Bicycle Presentation**<br>10:30 am – 12:00 pm **Poster Presentations (Even Numbered Posters)**<br>10:30 am – 12:30 pm **Learning Lounges**<br>10:45 am – 1:00 pm **Exhibit Theater Presentations**<br>11:30 am – 1:00 pm **Concession Lunch** |                                   |
| 11:30 am – 1:00 pm | **ACMG Board of Directors Meeting 2**<br>
*Location: 208 – Washington State Convention Center* |                                   |
| 1:00 pm – 3:30 pm | **Student Workshop: Careers in Medical Genetics – An Informational Session for Students and Trainees (Pre-registration required)**<br>
**Lunch – Sponsored by Illumina (1:00 pm – 1:30 pm)**<br>
*Location: 602-604 – Washington State Convention Center* |                                   |
| 1:30 pm – 3:30 pm | **CONCURRENT SESSIONS**<br>
**Complexities in Cancer Risk Assessment and Management**<br>
*Location: 6B – Washington State Convention Center* | ACMGLIVE   |
|              | **Laboratory Diagnostic Challenges (Constitutional and Neoplastic Cases in Molecular, Cytogenomic, and Biochemical Genetics Specialties)**<br>
*Location: 6C – Washington State Convention Center* |                                   |
|              | **Cardinal Signs of Common and Uncommon Disorders**<br>
*Location: 6E – Washington State Convention Center* | ACMGLIVE   |
| 3:30 pm – 4:00 pm | **Refreshment Break**<br>
*Location: 6ABC and E Lobbies – Washington State Convention Center* |                                   |
| 4:00 pm – 5:30 pm | **CONCURRENT SESSIONS**<br>
**Innovative Uses of Technology for Practicing Genetic Clinicians and Counselors**<br>
*Location: 606-609 – Washington State Convention Center* |                                   |
| 4:00 pm – 5:30 pm | **ORAL PLATFORM PRESENTATIONS**<br>
**Biochemical/Metabolic Genetics Platforms**<br>
*Location: 6C – Washington State Convention Center* |                                   |
|              | **Prenatal Genetics Platforms**<br>
*Location: 6B – Washington State Convention Center* |                                   |
SCHEDULE AT A GLANCE

2019 ACMG Annual Clinical Genetics Meeting

CANCER GENETICS AND THERAPEUTICS PLATFORMS
Location: 6E – Washington State Convention Center

PUBLIC HEALTH, HEALTH SERVICES AND IMPLEMENTATION PLATFORMS
Location: 611-614 – Washington State Convention Center

6:00 pm – 7:30 pm
- Pediatric Diagnostic Dilemmas (Rare Knowns and Unknowns)
  Location: 6E – Washington State Convention Center
- ACGME Accreditation Transition of Clinical Biochemical Genetics and Laboratory Genetics and Genomics Training Programs
  Location: 611-614 – Washington State Convention Center

7:00 pm – 7:30 pm
- ClinGen Hearing Loss Workgroup Meeting
  Location: 203 – Washington State Convention Center

7:30 pm – 9:00 pm
- Shabbat Dinner (Ancillary Event)
  Location: 201 – Washington State Convention Center
- ACMG Program Committee Meeting
  Location: 204 – Washington State Convention Center

SATURDAY, APRIL 6, 2019

7:30 am – 3:00 pm
- Speaker Ready Room
  Location: 619-620 – Washington State Convention Center
- Registration Open
  Location: Atrium Lobby – Washington State Convention Center

8:00 am – 3:00 pm
- Nursing Mothers Room
  Location: 302 – Washington State Convention Center
- Prayer and Meditation Room
  Location: 309 – Washington State Convention Center

8:00 am – 9:30 am
- CONCURRENT SESSIONS
  - Adult & Cancer Diagnostic Challenges
    Location: 6C – Washington State Convention Center
  - Phenotypes of “Single” Gene Disorders Are Complex Traits: Lessons from Inherited Metabolic Disorders for Personalized and Precision Medicine
    Location: 6E – Washington State Convention Center
  - Using Telegenetics to Improve Access to Clinicians and Researchers for Families
    Location: 6B – Washington State Convention Center

9:30 am – 9:45 am
- Refreshment Break
  Location: Skybridge – Washington State Convention Center

*INDICATES CLOSED OR BY INVITATION ONLY COMMITTEE, BOARD MEETING OR EVENT. IF A MEETING IS NOT CLOSED, ACMG MEMBERS ARE WELCOME TO ATTEND AND OBSERVE IT.
<table>
<thead>
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<th>Time</th>
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| 9:45 am – 10:45 am | **PLENARY SESSION**  
  - Hot Topics: Emerging Fetal Stem Cell and Molecular Therapies  
  *Location: Exhibit Hall 4E – Washington State Convention Center* |
| 10:45 am – 11:00 am | **ACMG / SIMD JOINT PLENARY SESSION (Part 1)**  
  - “Thinking Metabolic” for the Genetics Clinician. You Took the Call from the Department of Health: What Now?  
  *Location: Exhibit Hall 4E – Washington State Convention Center* |
| 11:00 am – 12:30 pm | **ACMG / SIMD JOINT PLENARY SESSION (Part 2)**  
  - “Thinking Metabolic” for the Genetics Clinician: Could This Really be Metabolic? Dysmorphic Features of IEM  
  *Location: Exhibit Hall 4E – Washington State Convention Center* |
| 1:30 pm – 3:30 pm | **ACMG / SIMD JOINT PLENARY SESSION (Part 2)**  
  - “Thinking Metabolic” for the Genetics Clinician: Could This Really be Metabolic? Dysmorphic Features of IEM  
  *Location: Exhibit Hall 4E – Washington State Convention Center* |
ABSTRACTS
Abstracts of the platform and poster presentations may be viewed online at acmgmeeting.net and via the ACMG Mobile App.

NOTE: Speakers in the plenary and concurrent sessions are not required to provide abstracts.

ACMG
Stop by the ACMG booth in the Exhibit Hall (Booth 300) to learn more about ACMG’s current programs, events and membership. Submit your membership application on-site and receive a $50 application fee waiver and a $75 rebate coupon to apply towards registration for the 2020 ACMG Annual Meeting or the Genetics and Genomics Review Course, a $125 value. The ACMG Booth will feature:

- ACMG membership information
- ACMGLive and OnDemand education information
- ACMG’s annual limited-edition T-shirt
- Social media promotion during the Opening Exhibit
- Branded gift distribution
- The ACMG Foundation for Genetic and Genomic Medicine information
- Genetics in Medicine information
- Current ACMG publications and resources
- Medical Genetics Awareness Week giveaways
- 2019 Annual Meeting Mobile App assistance

ACMG EMPLOYMENT RESOURCES
Stop by ACMG’s Employment Resources area near the ACMG booth where you can post your job opportunities on our job boards for free or expand your reach by posting to ACMG’s Online Employment Resource Center that offers options to cross post to more than 290 healthcare employment sites.

ACMG FOUNDATION EVENT – DAY OF CARING
The Day of Caring is a special, heartwarming event hosted by the ACMG Foundation for Genetic and Genomic Medicine and sponsored through the generous support of genetics professionals. Once again, in our small way, we will show others how much we care by giving bicycles and helmets to local children living with genetic diseases. Be sure to join us for this special event on Friday, April 5 at 10:30 am in the Exhibit Hall.

ACMG REGISTRATION AND BADGES
ACMG Registration is located in the fourth floor Atrium of the Washington State Convention Center and is open during the following hours:

- Tuesday, April 2 .......... 10:00 am – 7:00 pm
- Wednesday, April 3 ...... 6:30 am – 6:00 pm
- Thursday, April 4 ........ 7:00 am – 6:00 pm
- Friday, April 5 .......... 7:00 am – 6:00 pm
- Saturday, April 6 ......... 7:30 am – 3:00 pm

Name badges are required for admittance to all educational sessions, exhibits and other functions and events. ID will be requested when picking up your badge. If you registered a guest, pick up their badge when obtaining your registration materials. Badges contain a bar code that holds the attendee’s name, address, email, National Provider ID and demographic information provided when registering. Exhibitors will scan badges to send information after the meeting.

Employers: Post a job opportunity to our online site while attending the meeting and receive a $50 discount.
Job Seekers: Post a resume for free and view employment opportunities.
ATTENDEE INTERACTION PLATFORM

Many sessions held during the meeting will feature Sync by Freeman™, a second-screen platform that creates two-way dynamic participation between attendees and speakers. Using Sync attendees will be able to:

- Post comments and questions to the speakers;
- Respond to Audience Response and polling questions, provide feedback, give insights and more;
- View PowerPoint slides and follow along with the content via your tablet, smartphone or laptop;
- Take and save notes, the notes are emailed to you.

There will be a link to Sync for those sessions utilizing the platform within the Mobile App and additional instructions and a direct link to the platform will be provided in each session. **Please note that some slides may be withheld from viewing on a device due to restrictions by the presenter.**

BREAKS AND CONCESSIONS

Morning and afternoon refreshment breaks will be located in the following areas of the Convention Center:

- Wednesday afternoon  
  Sixth floor 6ABC and E Lobbies
- Thursday morning/afternoon  
  Exhibit Hall
- Friday morning  
  Exhibit Hall
- Friday afternoon  
  Sixth floor 6ABC and E Lobbies
- Saturday morning  
  Fourth floor Skybridge

Concession/Cash lunches are available for purchase in the Exhibit Hall on Thursday and Friday. Some exhibitors will distribute lunch coupons from their booth. See the Addendum in the registration bag for a list of those companies.

COAT AND LUGGAGE CHECK

A coat and bag check area will be available near the ACMG booth on the fourth floor of the Atrium of the Washington State Convention Center from Tuesday through Saturday. Please do not take luggage into the session rooms.

COMMITTEE AND ANCILLARY MEETINGS

ACMG committee meetings are listed in the Schedule at a Glance, the Daily Schedules and the ACMG Mobile App. All ACMG members are invited to attend and observe committee meetings unless designated as a “closed meeting.” Ancillary meetings are organized by groups other than ACMG and may have restricted attendance.

DRESS CODE AND EVENT POLICIES

- **Dress Code:** Appropriate dress is business casual. A light jacket or sweater is recommended, as meeting rooms may be cool. Badges will be required for entry into all sessions and events.
- **Consent for Use of Photographic Images:** Registration and attendance at, or participation in, ACMG meetings and other activities constitutes an agreement by the registrant to permit ACMG’s use and distribution (both now and in the future) of the registrant or attendee’s image or voice in photographs, videotapes, electronic reproductions, audiotapes of such events and activities.
- **Use of Recording Equipment:** Recording audio, video, livestreaming or photographs of speaker presentations, poster presentations, or exhibits is strictly prohibited, unless pre-approved by ACMG, as is the case with authorized recording and photography vendors. Copyright and intellectual property law is observed during all ACMG presentations, sessions and events.
- **Children and Guests:** Children and guests are not permitted in educational sessions, and children under age 12 are not allowed into the Exhibit Hall.
• **Mobile Devices:** As a courtesy to presenters and other attendees, mobile devices must be turned off or switched to vibrate or silent during sessions.

### EXHIBITS AND POSTERS

**Exhibit Hours:**

- Wednesday, April 3... 5:30 pm – 7:30 pm *(Opening Reception in the Exhibit Hall)*
- Thursday, April 4........ 9:30 am – 4:30 pm
- Friday, April 5 ............ 9:30 am – 1:00 pm

Be sure to visit the exhibit hall to learn about the latest industry developments, see demonstrations of new products and services, view the over 900 poster presentations that will be on display, and experience Exhibit Theater and Learning Lounge Presentations. Exhibits are located in Exhibit Halls AB and C located on the fourth floor of the Convention Center.

Exhibitor information can be found in the Exhibits Section of this program guide. The ACMG Mobile App contains the detailed descriptions of each company, product categories, booth number, and the exhibit hall floor plan.

Your barcoded badge will allow you to request additional information from exhibitors. Contact information provided during the registration process, including email addresses, is provided to exhibitors via the barcode on the badge.

**Poster Hours:**

Poster presenters will be present at their boards during the following times:

- Thursday, April 4......... 10:00 am – 11:30 am *Odd-numbered posters*
- Friday, April 5 ............ 10:30 am – 12:00 pm *Even-numbered posters*

**ePosters:** Posters uploaded by authors will be available during and after the meeting in ePoster format. Authors may upload their poster file during the meeting at the kiosk provided in the Exhibit Hall.

### LEARNING LOUNGES

Learning Lounges located in the Exhibit Hall will feature 30 minute sessions and offer participants bite-sized education in a variety of formats. Session listings and descriptions are available in the online program, on the meeting website and in the Exhibit Hall.

### EXHIBIT THEATERS

Several exhibiting companies will present state-of-the-art sessions in the two Exhibit Theaters in the Exhibit Hall showcasing recent developments and applications of the products and/or services offered by the company. Each session is 30 minutes long. Sessions are open to all attendees and seating is limited. The Exhibit Theater schedule is listed in the Exhibits Section of this guide and in the Mobile App.

### HOTEL INFORMATION

ACMG’s housing partner, CMR, will be available at the Housing Desk in the Registration Area In the fourth floor Atrium of the Washington State Convention Center to assist attendees with any housing and hotel needs.

### LOST AND FOUND

Lost items may be turned in to the ACMG Office located in Room 615/616 of the Washington State Convention Center. All unclaimed items will be turned over to the Convention Center Security Department at the end of the meeting.

### MEDIA/PRESS RELATIONS

The Press Room, located in Room 618 of the Washington State Convention Center is open for all working, credentialed media throughout the Meeting. ACMG and exhibitor press kits are available here.

Arrangements for interviews and inquiries concerning news releases should be arranged through Kathy Moran, ACMG Senior Director of Communications & Public Relations, in the Press Room or by email at kmoran@acmg.net.
MOBILE APP

A smartphone mobile app will be available for attendees during the Annual Meeting, and up to one year post meeting. This app will give attendees a convenient way to navigate the 2019 ACMG Annual Clinical Genetics Meeting. Information about how to download and utilize this app is available in the registration materials. Visit the ACMG booth for assistance with the app.

MEDICAL GENETICS AWARENESS WEEK

The first-ever Medical Genetics Awareness Week will be celebrated April 2-6, 2019. Through Medical Genetics Awareness Week, ACMG aims to promote the understanding of the importance of medical genetics professionals, including medical geneticists, laboratory geneticists and genetic counselors on the healthcare team. The theme of the week is “Celebrating the Contributions of the Entire Medical Genetics Team to Patient Care and Public Health.”

Medical Genetics Awareness Week is being celebrated for the first time in 2019 to recognize the invaluable contributions that medical genetics healthcare professionals make in the diagnosis, management and prevention of genetic diseases and the difference these professionals make in the lives of patients and families. During this week, which will coincide annually with the ACMG Annual Clinical Genetics Meeting, we take time to recognize the skills and commitment of all those on the healthcare team who translate genetic and genomic discoveries into better patient care.

People are invited to use the following hashtags for posts related to the celebration of Medical Genetics Awareness Week and to tag @TheACMG in Twitter posts:

#MedicalGeneticsAwareness
#IamaMedicalGeneticist
#FutureGeneticsProfessional

NURSING MOTHERS ROOM

Room 302 in the Washington State Convention Center is available for nursing mothers. The room is set up to provide privacy and a refrigerator will be available for storage.

PRAYER AND MEDITATION ROOM

The Prayer and Meditation Room located in Room 309 allows attendees of all religious faiths and non-religious beliefs to experience a place for peace, prayer, meditation and/or reflection throughout the day. This area is intended for individual use and not for any organized group activity or meetings.

RECORDED ON-DEMAND SESSION CONTENT

Over 30 scientific sessions and platform presentations are being captured for distribution as videos or audio synched with slides. They will be available in late June with CME, P.A.C.E.®, general CEU’s and NSGC credits.

Attendees who did not purchase the content recordings when registering and who wish to order access to the recorded content capture may do so at the Content Recording Sales kiosk located in the ACMG Booth #300 in Hall C. If you did not order the content recordings during the registration process, the on-site cost is $229. The price will increase to $349 (member) and $399 (nonmember) after the meeting – so be sure to place your order while at the meeting. CME, P.A.C.E.®, general CEU’s and NSGC credits are available.

SAFETY AND SECURITY

Meeting room attendants and security personnel will check for meeting badges at the entrance of sessions, exhibits and Annual Meeting events. Attendees should wear badges at all times during the meeting and should not dispose of badges in public trash containers.
Be sure to follow common sense safety precautions throughout the meeting, when out in the city and in your hotel. Remove your badge when leaving the Convention Center. Do not share your hotel room number with anyone. Establish the quickest route to emergency exits and fire escapes from your room, utilize hotel safety deposit boxes, and do not leave cash, credit cards, or other valuable items in your hotel room.

SOCIAL MEDIA

ACMG has several social media sites and you are invited to become a fan, group member or follower on our Facebook (American College of Medical Genetics and Genomics), LinkedIn (ACMG Annual Clinical Genetics Meeting) and Twitter (www.twitter.com/theacmg) sites to get the latest ACMG and Meeting news, information, updates, schedule or room changes and more.

Please use the ACMG 2019 Annual Meeting hashtag #ACMGMtg19 on your posts related to the annual meeting so that others can follow the discussion thread. The ACMG video series “Careers in Genetics” and “Genetics & Your Health” are also available on YouTube at www.youtube.com/TheACMGChannel. Not an ACMG member yet? Watch the new “ACMG – Be a Part of It!” membership video on The ACMG Channel on YouTube to see why ACMG membership is a must for any professional in genetics and genomics.

#ACMGMtg19 – Social Media Promotion: Be a Part of it!

Visit the ACMG Booth #300 during the Exhibit Hall Opening Reception on Wednesday, April 3 from 5:30 – 7:30 PM and become part of the ACMG Social Media Experience! Whether you are attending a meeting session, visiting exhibits or catching up with colleagues, we want to hear about it on Twitter, Facebook, or our NEW Instagram page @theacmg. Post on your social media account(s) using our meeting hashtag #ACMGMtg19 or one of the Medical Genetics Awareness Week hashtags (#MedicalGeneticsAwareness, #IamaMedicalGeneticist or #FutureGeneticsProfessional) and show us your social media post or take a fun photo using our Selfie frame at The ACMG booth and join the conversation to receive a free limited edition 2019 meeting t-shirt during the Exhibit Hall Opening Reception (while supplies last).

ACMG Social Media Policy for the ACMG Annual Clinical Genetics Meeting

ACMG Meeting attendees and participants are encouraged to participate in social media activities about the ACMG Annual Clinical Genetics Meeting by using the hashtag #ACMGMtg19. Discussion of general topics, speakers, and presentations is permitted as a way to briefly summarize or highlight material and the conference in general as long as you respect copyright and other intellectual property laws. For example, taking photos, videos, audiotapes or livestreaming of slides, posters and presenters or questions and answers are NOT permitted and will be requested to be removed at the discretion of the ACMG.

ACMG Social Media Code of Conduct

INFORMATION PROVIDED BY ACMG

ACMG social media channels were created to provide our members and other interested parties with information in order to build visibility and awareness of ACMG and its programs, products and positions. Although ACMG tries to ensure the accuracy and reliability of information presented by or on our behalf on these channels, it does not guarantee the accuracy, completeness, efficacy or timeliness of such information, and therefore, makes no representation or warranty regarding such information.

INFORMATION PROVIDED BY OTHER USERS

The ACMG social media channels were not created to allow other organizations or companies to advertise products for commercial gain. ACMG does not endorse,
guarantee, warrant, or recommend any products or services discussed or advertised on its social media channels. Reliance on any information appearing on these channels is solely at your own risk.

Any content and/or opinions posted by a third party, including all materials, articles, hyperlinks, documents, spreadsheets, presentations, images, individual user profiles, comments, opinions, dialogue, any other content or opinions in any form and responses to questions (hereinafter “User-Posted Content”) are solely the opinions and responsibility of the person or entity posting them and do not necessarily reflect the opinions of the ACMG. ACMG makes no representation or warranty regarding User-Posted Content that is posted on its social media channels. ACMG expressly disclaims any and all liability related to such posts. By using the social media channels, you agree to hold ACMG harmless from any and all claims arising out of User-Posted Content.

Frequent off-topic posts by the same individual or organization, or posts that are blatantly designed to generate commercial gain rather than discussion, may be deleted at the sole discretion of the ACMG platform administrators. ACMG does not condone uncivil dialogue on its social media channels, including but not limited to bullying, threatening language, intimidation, profanity, obscenity, spamming, ethnic slurs or personal attacks. By using the ACMG social media channels, you agree not to post any such uncivil dialogue. ACMG may, but does not accept any responsibility to, monitor or delete posts or take other steps to ensure that the dialogue on its social media channels conforms to this policy. ACMG will be the sole judge of what is appropriate for this purpose.

SUNSHINE ACT – WHAT ATTENDEES NEED TO KNOW

Under the Physician Payment Sunshine Act (Section 6002 of the Patient Protection and Affordable Care Act), medical device, biologic, and drug companies are required to track all payments and transfers of value (TOV) to U.S. healthcare providers made on or after January 1, 2012.

To assist 2019 Annual Meeting exhibitors in complying with the federal mandate associated with the Physician Payment Sunshine Act, the attendee’s NPI number (provided during registration) will be imbedded in the bar code data within the badge. ACMG Annual Meeting attendees should understand that, although supplying NPI information as part of Annual Meeting registration is voluntary, the reporting of transactions by companies under the Sunshine Act is mandatory. Beginning in 2013, companies must report any payment or TOV (transfers of value) with a minimum value of $10/payment or a cumulative value of $100 or more per year to the Department of Health & Human Services (HHS) and publicly disclose the information on their websites.

Badges may also be scanned at the entrance of the Satellite Symposia so the industry supporters can track and report their transaction in relation to these sessions.

SUSTAINABILITY INITIATIVES

Washington State Convention Center has been an industry leader in conservation and recycling for more than two decades, and is dedicated to environmental building practices and “green” operations.

They continue to reduce their carbon footprint with sustainable culinary practices (such as locally sourced food and beverages), energy-efficient lighting, environment-friendly cleaning products, and a first-rate recycling and composting program. They continue to invest significant resources in new and innovative projects that make the facility even more efficient.

ACMG kindly requests that attendees help out by disposing of trash, recycling, and compost in the appropriate receptacles.
2019 Annual Meeting
Program Committee
ACMG thanks the 2019 Program Committee for their dedication and hard work.

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Barrie Suskin Kaplan, MD, FACMG
Tracey Weiler, PhD, MS
Janson White, Student Member

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Claudia Barnett, Education Program Coordinator
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Melanie Wells, MPH, Director of Administration
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Kathy Moran, MBA, Senior Director, Communications and Publications
Janae Collins, Senior Administrative Assistant
Lynn Fleisher, PhD, JD, FACMG, Legal Counsel
Program Information

PURPOSE
The 2019 Annual Clinical Genetics Meeting will present both research and clinical topics that promote the science and the practice of clinical genetics and genomics. Sessions will focus on the latest discoveries of the etiology and the pathogenesis of genetic disorders, the latest developments in genetic testing and screening, the laboratory’s role in the diagnosis of genetic disorders, the treatment of genetic disorders in children and adults, the delivery of genetic services and more.

AUDIENCE
- Medical and clinical geneticists; genetic counselors; pediatric, obstetric, and maternal fetal specialists; and all medical practitioners who are providing comprehensive diagnostic, management, and counseling services for patients with, or at risk for, genetically influenced health problems
- Laboratory directors and technicians who conduct genetic testing
- Researchers involved in the discovery of genetic disorders and treatments
- Clinical, laboratory and research trainees of genetics and all biomedical sciences
- Any healthcare and public health professionals who have an interest in medical and clinical genetics and genomics
- Advocates for patients with genetic conditions and their families

LEARNING OBJECTIVES
After attending the 2019 ACMG Annual Meeting, participants should be able to:
- Describe the advances in the detection of genomic variants using current and emerging technology
- Recognize cardinal clinical signs and symptoms of specific syndromic condition
- Manage initial care of new disorders on the recommended uniform screening panel
- Describe the latest advances in the field of cancer genetics
- Review the differential diagnoses of unknown cases
- Formulate differential diagnoses for inborn errors of metabolism with physical manifestations
- Examine the potential and the challenges of prenatal genetics and newborn screening
- Summarize the indications for genetic testing
- Interpret pharmacogenomics reports to understand patient-specific clinical implications
- Review the latest advances in targeted and personalized treatments for genetic conditions
- Formulate innovative strategies to enhance healthcare delivery in the field of genetics medicine
- Describe pre- and post-test counseling for secondary findings and variants of uncertain significance

MEETING FORMATS
- Plenary and Concurrent Sessions
  These sessions have been developed based on proposals from members and non-members of ACMG and the Program Committee. The sessions highlight a wide range of topics of interest to genetics practitioners, researchers, laboratory directors and technicians, and counselors. If submitted, abstracts from speakers are available in their online session description.
- Learning Lounges
  Learning Lounges located in the Exhibit Hall will feature 30-minute sessions and offer participants bite-sized education in a variety of formats. Session listings and descriptions are located in the Mobile App and online at www.acmgmeeting.net. Don’t miss the “Rapid Fire Poster Presentations” featuring the top twenty posters, each presented in one minute.
Tech Bar:
Experts with their fingers on the pulse of the latest technologies will be available throughout the exhibition to field questions and inquiries from attendees about how technology can and will benefit you on a personal and professional level. The Tech Bar provides an immersive experience of best practices, trends and knowledge that every professional needs in today's technological and social media environment.

Abstract Presentations
The presentation of cutting-edge research is an essential feature of the ACMG Annual Clinical Genetics Meeting. Abstract presentations are selected from abstracts that were submitted by genetics professionals (ACMG members and non-members). Submitted abstracts were peer reviewed and the Program Committee has selected the highest scoring abstracts for either a poster or a platform presentation.

Platform Presentations
ACMG's Program Committee has assembled 60 abstract-driven platform presentations. Each presenting author will give a 10-minute presentation followed by 5 minutes of discussion. On Wednesday, April 3 from 12:15 pm – 1:15 pm there will be an Industry Platform Presentation session. Please note that educational credits are not available for this session. Platform sessions in Genetic Counseling, Clinical Genetics and Therapeutics, Laboratory Genetics and Genomics, and Legal and Ethical Issues and Education will be presented concurrently on Wednesday, April 3 from 4:00 pm – 5:30 pm. Platform sessions in Biochemical/Metabolic Genetics, Cancer Genetics and Therapeutics, Prenatal Genetics, and Public Health, Health Services and implementation will be presented concurrently on Friday, April 5 from 4:00 pm – 5:30 pm. The plenary session, Featured Platforms, will showcase four top-rated abstracts and be presented on Thursday, April 4, from 8:00 am – 9:00 am.

Poster Presentations
To encourage scientific discussions and to facilitate the exchange of information, posters are grouped by related topics (selected by the first author at the time of abstract submission). Abstract listings include titles of abstracts and author names, preceded by abstract numbers. The first author listed is the presenting author, unless a co-author has been designated to present. Posters will be on display in the Exhibit/Poster Hall during all exhibit hours on Wednesday evening (Opening Reception in the Exhibit Hall), Thursday, and Friday. The Top Rated Posters in each topic area are recognized with a rosette. Search for them in the ACMG mobile app or www.acmgmeeting.net. Poster presenters for odd-numbered posters will be present on Thursday, April 4, from 10:00 am – 11:30 am; presenters for even-numbered posters will be present Friday, April 5, from 10:30 am – 12:00 pm. The number on each poster board corresponds to the abstract number that may be accessed on the ACMG meeting website (www.acmgmeeting.net) and via the ACMG mobile app.

Poster topics:
- Clinical genetics and therapeutics
- Cancer genetics and therapeutics
- Biochemical and metabolic genetics
- Laboratory genetics and genomics
- Genetic counseling
- Prenatal genetics
- Public health, health services and implementation
- Legal and ethical issues, education
SIG Forums, Workshops and Luncheons
In addition to sessions selected via a submission process or developed by the Program Committee, the ACMG Annual Meeting also includes a variety of other educational opportunities including:

- Public Health SIG Forum and Business Meeting – Hot Topics 2019: Challenges, Controversies, Conversations
- Adult Genetics SIG Forum – Novel Therapeutics in Adult Genetic Disease
- Using Quality Improvement Tools to Enhance Patient Care and Training in Genetics
- Addressing Literacy and Language to Equitably Deliver on the Promise of Precision Medicine
- ClinGen Expert Panel Information Workshop
- ACGME Accreditation Transition of Clinical Biochemical Genetics and Laboratory Genetics and Genomics Training Programs
- First Time Attendee Orientation
- Trainee Workshop: Publishing Your Scholarly Work: Soup to Nuts
- Fellow/Resident/Trainee Mentor Luncheon
- Genetic Counselors Luncheon
- Special Student Session: Careers in Medical Genetics
- ACMG Interpreting Sequence Variation Guideline Revision Workgroup – Open Forum
- Informational Session: Laboratory Genetics and Genomics Alternative Pathway Training Program

Diagnostic Challenges Sessions
These sessions provide attendees with an opportunity to bring cases to the attention of a panel of experts and an audience of geneticists and genetics professionals for discussion of differential diagnosis, treatment, and counseling opportunities.

- Prenatal Diagnostic Challenges
  Wednesday, April 3
  1:30 pm – 3:30 pm

- Laboratory Diagnostic Challenges
  (Constitutional and Neoplastic Cases in Molecular, Cytogenomic and Biochemical Genetics Specialties)
  Friday, April 5
  1:30 pm – 3:30 pm

- Pediatric Diagnostic Dilemmas
  (Rare Knowns and Unknowns)
  Friday, April 5
  6:00 pm – 7:30 pm

- Adult and Cancer Diagnostic Challenges
  Saturday, April 6
  8:00 am – 9:30 am

Satellite Symposia
These symposia, held on Tuesday evening and Wednesday and Friday mornings, are noncommercial scientific sessions supported by ACMG’s industry partners that broaden the educational experience for meeting attendees. These symposia are not a part of the official ACMG Annual Clinical Genetics Meeting planned and developed by the Program Committee. They are not endorsed by ACMG, nor do they offer CME credits by ACMG. Some sessions are accredited by independent CME providers, and ACMG has obtained approval for continuing education units (CEUs) for genetic counselors from the National Society of Genetic Counselors (NSGC) and P.A.C.E.® credits. While these sessions are open to all ACMG attendees, pre-registration is required. Many sessions reached maximum capacity prior to the start of the meeting.

Exhibit Theater Presentations
These 30-minute presentations are held in theaters in the Exhibit Hall during exhibit hours and are presented by exhibiting companies. These presentations were not planned by the Program Committee and are not approved for educational credit. These sessions showcase the companies’ recent developments and applications of their products, techniques, and/or services, and they demonstrate how the products or services are used in practical, laboratory and/or clinical settings.
Continuing Education Information
CME, MOC, P.A.C.E.®, and NSGC
Educational Credits

Each attendee seeking credit must claim their time spent in a session by using the online Self-Reporting system. The number of credits awarded for each session are included in the session description in the Program Guide and on the meeting website (www.acmgmeeting.net).

The number of education credits vary by the type of credit awarded. For future reference and access to the evaluation and education credit reporting system you need to login with your badge number, so please make a note of your number.

Deadlines:
- NSGC credits must be claimed by June 6, 2019. Credits claimed after this deadline will be reported at the end of December 2019.
- All other credits must be claimed by December 1, 2019.

Continuing Medical Education (CME)

Accreditation
The American College of Medical Genetics and Genomics is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.

Credit Designation
The American College of Medical Genetics and Genomics designates this live activity for a maximum of 28.5 AMA PRA Category 1 Credits™. Physicians should claim only the credit commensurate with the extent of their participation in the activity. (24 AMA PRA Category 1 Credits™ if you are not attending a Short Course) The American Medical Association (AMA) defines physicians as those individuals who have obtained an MD, DO, or equivalent medical degree from another country. Non-physicians may request a certificate of attendance for their participation at the registration desk.

ABMGG Lifelong Learning and Self-Assessment (MOC Part II) – Short Course Participants Only
ABMGG diplomates must demonstrate involvement in continuing education as part of the Lifelong Learning and Self-Assessment (Part II) requirement of the ABMGG Continuing Certification (Maintenance of Certification) program. The ABMGG has approved participation in the Short Courses offered at the 2019 ACMG Annual Clinical Genetics Meeting as alternative activities for the ABMGG literature review modules. Successful completion of a Short Course, including participation in the evaluation component, enables the participant to earn a certificate of completion for submission via the ABMGG Portal to fulfill this requirement.
Genetic Counselor Credit
The National Society of Genetic Counselors (NSGC) has authorized ACMG to offer up to 3.7 CEUs or 37 Category 1 contact hours for the activity 2019 ACMG Annual Clinical Genetics Meeting (Live Meeting). The American Board of Genetic Counseling (ABGC) will accept CEUs earned at this program for the purposes of genetic counselor certification and recertification. (32.5 CEUs if you are not attending a Short Course).

P.A.C.E.® CEU’s
ACMG is approved as a provider of continuing education programs in the clinical laboratory sciences by the American Society for Clinical Laboratory Science (ASCLS) Professional Acknowledgment for Continuing Education (P.A.C.E.®) Program. ACMG is approved by the Florida Board of Clinical Laboratory Personnel as CE Provider (50-11878). This course is registered # 20-696574 with CEBroker. ACMG is approved by the California Department of Health Services through the ASCLS P.A.C.E.®. This activity has been approved for 33 P.A.C.E.® CEU’s (28.5 CEU’s if you are not attending a Short Course).

DISCLAIMER: ACMG educational programs are designed primarily as an educational tool for health care providers who wish to increase their understanding of the application of genomic technologies to patient care. The ACMG does not endorse, or recommend the use of this educational program to make patient diagnoses, particularly by individuals not trained in medical genetics. Adherence to the information provided in these programs does not necessarily ensure a successful diagnostic outcome. The program should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed at obtaining the same results. In determining the propriety of any specific procedure or test, a healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Questions regarding CE credit should be directed to education@acmg.net.
Congratulations

Roger E. Stevenson, MD, FACMG
Greenwood Genetic Center

Recipient of the 2019 David L. Rimoin Lifetime Achievement Award in Medical Genetics in recognition of the life and career of the late Dr. David L. Rimoin.

The David L. Rimoin Lifetime Achievement Award in Medical Genetics is given in recognition of the life and career of the late Dr. David L. Rimoin and is the ACMG Foundation’s most distinguished award. This distinction is presented to an individual in the medical genetics profession who exemplifies a lifetime of achievements and personal qualities embodied by Dr. Rimoin including his passion for teaching and mentoring, his care of patients and their families and his enthusiasm for integrating the medical genetics and genomics community into mainstream healthcare.

www.ACMGFoundation.org
March of Dimes Birth Defects Foundation
2019 Samuel Pruzansky Memorial Lecture

Maximillian Muenke, MD, FACMG
Senior Investigator, Chief, Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health

Thursday, April 4
Hall 4E – Washington State Convention Center

The Samuel Pruzansky Memorial Lectureship, which was established in memory of the founder and director of the world’s first center for craniofacial anomalies, is now sponsored by the March of Dimes Birth Defects Foundation.

Samuel Pruzansky, DDS was an internationally renowned research scholar, clinician, and teacher, starting at the National Institute of Dental Research. In 1959, he chaired the Gatlinburg Conference, the transformative meeting on craniofacial anomalies. It was during his academic career at the University of Illinois, that Dr. Pruzansky developed the Center for Craniofacial Anomalies, serving as Founding Director. He was also President of the American Cleft Palate Association, the Teratology Society, and the International Society for Craniofacial Biology. His achievements have been published as a memorial tribute, “Craniofacial Dysmorphology: Studies in Honor of Samuel Pruzansky” (New York, Alan R. Liss, Inc., 1985).

Previous Pruzansky Memorial Lecturers are listed below.

2018 Hope Northrup, MD, FACMG
2017 William Gahl, MD, FACMG
2016 Suzanne Cassidy, MD
2015 Eric Vilain, MD, PhD, FACMG
2014 Leslie Biesecker, MD, FACMG
2013 Brendan Lee, MD, PhD, FACMG
2012 George M. Martin, MD
2011 Harry Dietz, MD
2010 Christopher A. Walsh, MD, PhD
2009 James R. Lupski, MD, PhD, FAAP, FACMG, FAAAAS
2008 Malcom A. Ferguson-Smith, FRS
2007 David L. Rimoin, MD, PhD, FACMG
2006 David C. Page, MD
2005 Irene H. Maumenee, MD
2004 Robert J. Gorlin, DDS
2003 Christine Petit, PhD
2002 James F. Gusella, PhD
2001 Eric N. Olson, PhD
2000 Douglas C. Wallace
1999 Huntington F. Willard, PhD
1998 Henry A. Erlich, PhD
1997 Connie Eaves, PhD
1996 Clifford J. Tabin, PhD
1995 Savio L. Woo, PhD
1994 Scott F. Gilbert, PhD
1992 Grant Sutherland, PhD, DSc
1991 D. Carleton Gajdusek, MD
1990 Francis S. Collins, MD
1990 Lap-Chee Tsui, PhD
1989 Louis M. Kunkel, MD
1988 Elizabeth D. Hay, MD
1987 Drew Noden, MD
2019 ACMGF Awards and the David L. Rimoin Award Presentation

Presented during the Presidential Plenary Session on Wednesday, April 3, 2019

The David L. Rimoin Lifetime Achievement Award

The David L. Rimoin Lifetime Achievement Award in Medical Genetics is the ACMG Foundation’s most distinguished award. This distinction is presented to an individual in the medical genetics profession who exemplifies a lifetime of achievements and personal qualities embodied by Dr. Rimoin, including his passion for teaching and mentoring, his care of patients and their families, and his enthusiasm for integrating the medical genetics and genomics community into mainstream healthcare. The David L. Rimoin Lifetime Achievement Award is the most prominent award given on behalf of ACMG and the ACMG Foundation.

The 2019 award recipient is
Roger E. Stevenson, MD, FACMG, Greenwood Genetic Center

Presented during the ACMGF and David L. Rimoin Donor Recognition Reception on Thursday, April 4, 2019

The David L. Rimoin Inspiring Excellence Award

Since 2015, the David L. Rimoin Inspiring Excellence Award has been given to a selected student, trainee, or junior faculty ACMG member whose abstract submission is chosen as a platform presentation during the ACMG Annual Clinical Genetics Meeting. In recognition of their selected presentation, the Rimoin Fund provides a cash award and covers the cost of travel to the meeting.

The 2019 award recipient is
Sarah Brnich, BS, University of North Carolina School of Medicine

The Rimoin “Lifetime Achievement” and “Inspiring Excellence” awards are generously supported by The ACMG Foundation David L. Rimoin Fund.
About the ACMG Foundation “Next Generation” Fellowship and Residency Awards

The ACMGF “Next Generation” concept was implemented to fund one- and two-year competitive fellowships and residency opportunities in clinical genetics, clinical laboratory, and medical biochemical genetics subspecialty.

The program is made possible through corporate partnerships between Takeda, Pfizer, Sanofi Genzyme, and the ACMG Foundation for Genetic and Genomic Medicine to foster a new generation of geneticists around the country who will serve a critical role in the advancement of research, diagnosis and patient care of those living with rare diseases.

The 2019 ACMGF “Next Generation” Awards are generously funded by the following corporate partners:

The Sanofi Genzyme ACMGF “Next Generation” in Medical Biochemical Subspecialty Award is a one year fellowship designed to support the sustainable development of clinically interested and qualified clinical geneticists with an expertise in biochemical genetics.

The award sponsors a trainee’s first year of a fellowship in biochemical genetic diseases following residency. The fellowship year will commence in summer 2019. The purposes of the program are to advance education, research, and standards of practice in medical genetics; develop and expand clinical and laboratory expertise in medical genetics in the United States; and to initiate and develop a broad-based infrastructure for industry funding of high-quality projects in medical genetics.
ACMGF Takeda and Pfizer “Next Generation” Fellowship and Residency Awards

In 2016, through the ACMG Foundation Corporate Partners Program (CPP), Takeda Pharmaceuticals, a global biotechnology company and Pfizer, a premier biopharmaceutical company; developed a residency and fellowship initiative to address the shortage of medical geneticists in the United States (US).

The partnership with ACMGF is focused to train, develop and deliver innovative medicines for people living with rare diseases, and highly specialized conditions. The awards are intended to provide a unique mentoring opportunity for trainees and emerging professionals pursuing diverse genetic and genomic specialties.

**Takeda Medical Biochemical Subspecialty recipients are:**

Brian Shayota, MD Baylor College of Medicine  
Kevin Glinton, PhD, Baylor College of Medicine  
Joshua Baker, D.O., FAAP, Boston Children’s Hospital  
William Brucker, MD, PhD, Children’s Hospital Corporation  
Filippo Ingoglia, PhD, University of Utah, School of Medicine

**Takeda Clinical Genetics and Genomics Residency Fellowship Award**  
Elizabeth Jalazo, MD, University of North Carolina, Chapel Hill

**Pfizer Clinical Genetics and Genomics Residency Fellowship Award**  
Samuel Huang, MD, University of Washington School of Medicine

**Sanofi Genzyme Medical Biochemical Subspecialty recipient is:**  
Jessica Gold, MD, Children’s Hospital of Philadelphia
The Carolyn Mills Lovell Genetic Counselors Award

The Carolyn Mills Lovell Genetic Counselors Award will be presented on Thursday, April 4, 2019 at 11:45 am – 1:15 pm during the Genetic Counselors’ Luncheon.

Since 2015, the Carolyn Mills Lovell Genetic Counselors Award has been given to a selected genetic counselor whose abstract submission is chosen as a platform or poster presentation at the ACMG Annual Clinical Genetics Meeting.

The award winner is
Jin Yun Helen Chen, MS, CGC, Massachusetts General Hospital.

The PerkinElmer Travel Award

PerkinElmer Company and the ACMG Foundation for Genetic and Genomic Medicine offer the “Signature Genomic Travel Award” which supports one recipient to attend the ACMG Annual Clinical Genetics Meeting. The Award winner is selected from a list of competitive first authors, who are ACMG members who submit abstracts for the scientific program. The award has been given since 2008, with the exception of 2018.

The 2019 award recipient is
Cynthia Gubbels, MD, PhD, Boston Children’s Hospital.

Richard King Trainee Award for Best Publication in Genetics in Medicine Journal

The Richard King Trainee award was instituted in 2009 and is given by the ACMG Foundation to encourage ABMGG trainees in their careers and to foster the publication of quality research in Genetics in Medicine. The award is named for Dr. Richard King in recognition of his instrumental role in creating Genetics in Medicine and serving as the first Editor-in-Chief of the journal.

The 2019 award recipient is
Bryce Seifert, PhD, Duke University School of Medicine Durham, NC.
2019 ACMGF Corporate Partners

The ACMG Foundation for Medical Genetic and Genomic Medicine (ACMGF) and ACMG gratefully acknowledge the generous support of all donors throughout 2018-2019, and especially the following companies who participate in our Corporate Partners Program.

### Diamond Champion Partner
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- Myriad Women’s Health

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($15,000 – $24,999)
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($10,000 – $14,999)
- GeneDx
2019 ACMGF Genetics Leadership Society

A very special thank you to the members of the ACMG Foundation Genetics Leadership Society, who contributed $1,000 or more from January 1, 2018 – January 31, 2019

“We make a living by what we get; but we make a life by what we give.”

• WINSTON CHURCHILL

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Miriam G. Blitzer, PhD, FACMG
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Yuan Ji, PhD, MS, FACMG
Susan D. Klugman, MD, FACMG
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Presenters, Moderators, Program and Education Committees’ Financial Disclosures

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This disclosure pertains to relationships with ACCME-defined commercial interests whose products or services may be related to the subject matter of the presentation topic. Any real or apparent conflicts of interest related to the content of the presentations must be managed prior to the educational activity.

NOTE:
- ACMG will follow the ACCME’s expectation that no employees or owners of commercial interests will be involved as planners/faculty/presenters of a CME accredited activity.
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- The ACCME does not consider providers of clinical service directly to patients to be commercial interests - unless the provider of clinical service is owned, or controlled by, an ACCME-defined commercial interest.
- Diagnostic laboratories are not considered commercial interests unless they are owned by or have a sister organization which is a commercial interest.

Presenters and moderators have disclosed the following:

Al-Zaidy, Samiah – AveXis Inc, 2
Blout, Carrie – 23andMe, 6
Bober, Michael – Biocarein, Shire, MedLife Sciences, Therachon, Ascendis, Alexion, 5
Bodamer, Olaf – Perkin Elmer, 2
Borrell, Antoni – Illumina, 5
Caleshu, Colleen – Clear Genetics, 2;
Gene Matters, 2; MyoKardia, 2; Personalis, 3; Phosphorus, 2
Cunniff, Christopher – Biogen (member of the Pregnancy Advisory Board), 10
Dhar, Shweta – Acer Therapeutics, 9;
PanGenomics, INDIA, 3

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9. Membership on an entity’s board of directors, speaker’s bureau, or its advisory committees
10. Any other financial relationship and list organization and relationship
Elsea, Sarah – Baylor Miraca Genetics Laboratories, 3; Smith-Magenis Syndrome Research Foundation, 2

Flannery, David – Salus Telehealth, 3

Gelb, Bruce – GeneDx, Prevention Genetics, Correlegan, LabCorp, 7

Gibbons, Melissa – Sarepta, AveXis, 10

Harding, Cary – Biopta, AveXis, 2

Hopkin, Robert – Genzyme Sanofi, 9

Howell, R. Rodney – AveXis, Baebies, Sanofi-Genzyme, 6

Korf, Bruce – Envision Genomics, 9; Genome Medical, 9; Novartis, 5

Lincoln, Stephen – Illumina and Thermo Fisher, 4

Litton, Jennifer – ASCO, 10; AstraZeneca, 10; EMD Serono, 5; Genentech, 5; Glaxo Smith Kline, 5; medLearning Group, 9; NCCN, 10; Pfizer, 10; Physician's Education Resource, 9; UpToDate, 7

MacKenzie, Tippi – Ultragenyx, 5

Manzi, Shannon – Global Gene, Inc., 3

Mason, Christopher – Illumina, Abbvie, Takeda, DNA Genotek, Zymo Research, Biotia, Onegevity, 2

McCandless, Shawn – Aeglea Biotherapeutics, 9; Biogen, 9; Department of Justice Vaccine Injury Compensation Program, 8

Medin, Jeffrey – Avrobio, Inc., 4, 7; Rapa Therapeutics, 9; Sanofi Genzyme, 6; Shire, 6

Moore, Kathleen – Aravivie, 2; Astra Zeneca, 2; Clovis, 2; Genentech/Roche, 2; Janssen, 2; Merck, 2; Tesaro, 2; VBL Therapeutics, 2

Park, Ben – Loxo Oncology, 2

Pennesi, Mark – Spark Therapeutics, AGTC, Nightstar, ProQR, Sanofi, Gensight, 2

Picker, Jonathan – Global Gene Corp, 3

Rao, Vamshi – Avexis, 9

Raymond, Gerald – Bluebird, 2; Viking, 2

Robson, Mark – Abbvie, 5; AstraZeneca, 2, 5, 6; InVitae, 5; McKeeons, 2; Medivation, 5; Myriad Genetics, 5; Pfizer, 5, 6; Tesaro, 5

Roth, Frederick – Ranomics, 3; SeqWell, 3, 9

Sacharow, Stephanie – Bi marin, 5, 9

Schaaf, Christian – Springer, 3, 7

Schmidt, Ryan – Veritas Genetics, 2

Schneider, Holm – EspeRare Foundation, 7, 9; EspeRare Foundation (non-profit organization), 5

Shen, Jun – Counsyl/Myriad Genetics, 2

Skotko, Brian – Band of Angels Foundation, 9; expert legal witness, 8; Gerson Lehrman Group, 2; Massachusetts Down Syndrome Congress, 9; National Center for Prenatal and Postnatal Down Syndrome Resources, 9; Woodbine House, Inc., 7

Slavotinek, Anne – OptumRx, 2; Retrophin, 2; Oxford University Press, 7

Steiner, Robert – Acer Therapeutics, Censa Pharmaceuticals, 4; Acer Therapeutics, Censa Pharmaceuticals, Retrophin, Raptor, Biogen, Alexion, E-Scape Bio, Health Advances, Precision for Value, and Best Doctors, 2; Alexion, 5; Pfizer, 10

Swoboda, Kathryn – Biogen 5; AveXis, 5

Trzupek, Karmen – InformedDNA, 1; Spark Therapeutics, 2

Van den Veyver, Ignatia – Illumina, 5

Wilcox, William – Amicus, 5, 10; Freeline, 10; Genzyme-Sanofi, 2, 5, 10; Protalix, 10; Shire, 2, 5, 10

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8. Paid expert testimony
9. Membership on an entity’s board of directors, speaker’s bureau, or its advisory committees
10. Any other financial relationship and list organization and relationship
The following list of presenters and moderators have no disclosures to report:
(Report date Feb. 25, 2019)

Aaron, Kathleen
Adam, Margaret
Adam, Shelin
Arnold, Georgianne
Bedoukian, Emma
Berry, Gerard
Berry, Susan
Best, Robert
Billington, Charles
Birgmeier, Johannes
Brnich, Sarah
Burke, Leah
Bush, Lynn
Caggana, Michele
Chang, Jiyoo
Chaudhari, Bimal
Chen, Jin Yun
Chung, Wendy
Dang Do, An N.
Demmer, Laurie
Dipple, Katrina
DiStefano, Marina
Dubuc, Adrian
Eddleman, Keith
El-Gharbawy, Areeg
Esplin, Edward
Farnaes, Lauge
Fiesco-Roa, Moises
Fishler, Kristen
Funke, Birgit
Glinton, Kevin
Gropman, Andrea
Grzymski, Joseph
Gubbels, Cynthia
Hassold, Terry
Hisama, Fuki
Hodge, Jennelle
Hudgins, Louanne
Hulick, Peter
Joseph, Galen
Kass, Stephanie
Kelley, Whitley
King, Jennifer
Kishnani, Priya
Korson, Mark
Kronn, David
Liu, Ning
Luo, Xi
MacFarland, Suzanne
Magoulas, Pilar
Makhnoon, Sukh
Manace, Leslie
McCabe, Edward
Mikhail, Fady
Miller, David
Muenke, Maximilian
Oglesbee, Devin
Pal, Tuya
Pickarski, Justine
Plon, Sharon
Prows, Cynthia
Rajagopalan, Ramakrishnan
Regier, Debra
Reis, Linda
Riley, Catharine
Roberts, Amy
Robertson, Alison
Rocha, Heather
Sanchez-Valle, Amarilis
Scaglia, Fernando
Schindewolf, Erica
Schleit, Jennifer
Schmidlen, Tara
Segal, Jeremy
Sharp, Richard
Smith, Jessica
Stewart, Douglas
Talkowski, Michael
Tan, Wen-Hann
Toriello, Helga
Truty, Rebecca
Tvrdik, Tatiana
van Karnebeek, Clara
Vockley, Jerry
Vonesh, Marie
Vora, Neeta
Vossaert, Liesbeth
Wang, Catharine
Watson, Elise
Whitehead, Matthew
Wick, Myra
Wien, Katie
Wiesner, Georgia
Williams, Eli
Worthey, Liz
Yang, Shan
Zelley, Kristin
Members of the ACMG Program Committee (PC), Education Committee (EC) and Staff (S) have the following disclosures:

Bernat, John (EC) – Sanofi Genzyme, Shire and Protalix, 5
Cunniff, Christopher (PC) – Biogen (member of the Pregnancy Advisory Board), 10
Dhar, Shweta (EC) – PanGenomics India, 3
Elsea, Sarah (PC) – Vanda Pharmaceuticals, 2; PRISMS, Inc, 9
Hudgins, Louanne (PC) – Oxford Press, 7
Mito, Yoshiko (EC) – Coriell, 2
Mullen, Thomas (EC) – Good Start Genetics Inc., 6
Schaaf, Christian (PC) – Springer 3, 7
Slavotinek, Anne (PC, EC) – Oxford University Press and UptoDate, 7
Stevenson, David (PC) – Alexion, Lineagen, GLG, 2; PWSA, CFC syndrome MAB, and Costello syndrome MAB, 9
Toriello, Helga (PC) – Oxford Press, 7

The following members have nothing to disclose:

Barnett, Claudia (PC, EC, S)  Nguyen, Joanne (PC)
Berry, Sue (PC, SIMD Liaison)  Pal, Tuya (PC)
Bodurtha, Joann (EC)  Pappas, Kara (EC)
Chang, Irene (EC)  Parikh, Aditi (EC)
Cusmano-Ozog, Kristina (PC)  Radford, Jane (PC, EC, S)
Dahlroth, Jane (PC, EC, S)  Roberts, Amy (EC)
Eddleman, Keith (PC)  Smith, Jessica (PC)
Freire, Penelope (PC, S)  Suskin Kaplan, Barrie (EC)
Giovanni, Monica (PC, EC)  Watson, Michael (PC, EC, S)
Hisama, Fuki (PC)  Weiler, Tracey (EC)
Hodge, Jennelle (PC)  White, Janson (EC)
King, Jennifer (PC)  Wick, Myra (PC)
Magoulas, Pilar (PC)  Williams, Eli (PC)

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Daily Schedule – Educational Sessions, Events and Meetings

Indicates this session will be live streamed during the meeting.

Tuesday, April 2

**COMMITTEE & BOARD MEETINGS**

8:00 am – 4:00 pm
ACMG Board of Directors Meeting (Closed Meeting)
*Location: Jefferson – Sheraton Grand Seattle*

5:00 pm – 6:30 pm
ACMG Committee Chairs Meeting (Closed Meeting)
*Location: Room 208 – 2nd floor, Washington State Convention Center*

7:15 pm – 10:00 pm
ACMG Lab QA Biochemical Genetics Subcommittee Meeting (Closed Meeting)
*Location: Issaquah A – Sheraton Grand Seattle*

7:15 pm – 10:00 pm
ACMG Lab QA Cytogenetics Subcommittee Meeting (Closed Meeting)
*Location: Issaquah B – Sheraton Grand Seattle*

7:15 pm – 10:00 pm
ACMG Lab QA Molecular Genetics Subcommittee Meeting (Closed Meeting)
*Location: Kirkland – Sheraton Grand Seattle*

7:30 am – 9:00 pm
ACMG Program Committee Dinner (Closed Meeting)
*Location: Loulay Restaurant*

**ANCELLARY EVENTS**

7:00 am – 12:00 pm
ABMGG Board Exam
*Location: Room 205 – 2nd floor, Washington State Convention Center*

8:00 am – 1:30 pm
CAP Inspection Training Seminar
*Location: Room 2AB – 2nd floor, Washington State Convention Center*

6:00 pm – 9:00 pm
AAP Council on Genetics Executive Committee Meeting
*Location: Seneca – Sheraton Grand Seattle*
SHORT COURSES

12:30 pm – 5:30 pm

Skeletal Dysplasias Across the Human Lifespan
Location: Room 611-614 – 6th floor, Washington State Convention Center

This session is eligible for up to (4.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Course Directors: Keith Eddleman, MD, ABMGG, ABOG, FACMG, Icahn School of Medicine at Mount Sinai; Jennifer King, MD, Albert Einstein College of Medicine

Faculty: Deborah Krakow, MD, FACMG, UCLA; Sheila Unger, MD, Centre Hospitalier Universitaire Vaudois; Michael Bober, MD, PhD, FACMG, Nemours/A.I. duPont Hospital for Children; Sandesh Nagamani, MD, FACMG, Baylor College of Medicine; Ericka Okenfuss, MS, CGC, Kaiser Permanente

12:30 pm – 5:30 pm

Somatic Mutation Testing in Cancer: Implications for the Oncologist and Geneticist
Location: Room 606-609 – 6th floor, Washington State Convention Center

This session is eligible for up to (4.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Course Directors: Georgia Wiesner, MD, MS, FACMG, Vanderbilt University Medical Center; Eli Williams, PhD, FACMG, UVA School of Medicine

Faculty: Ben Park, MD, PhD, Vanderbilt University Medical Center; Ken Walsh, PhD, UVA School of Medicine; Thomas Slavin, MD, FACMG, City of Hope National Medical Center

12:30 pm – 5:30 pm

NAMA at the ACMG 3.0
Location: Room 602-604 – 6th floor, Washington State Convention Center

This session is eligible for up to (4.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Course Directors: Georgianne Arnold, MD, FACMG, University of Pittsburgh Medical Center; Mark Korson, MD, VMP Genetics

Faculty: Jerry Vockley, MD, PhD, FACMG, University of Pittsburgh School of Medicine; Amy Goldstein, MD, Children’s Hospital of Philadelphia

WORKSHOPS

4:15 pm – 5:00 pm

First Time Attendee Orientation
Location: Ballroom 6A – 6th floor, Washington State Convention Center

7:30 pm – 9:00 pm

Addressing Literacy and Language to Equitably Deliver on the Promise of Precision Medicine
Location: Room 606-609 – 6th floor, Washington State Convention Center

7:30 pm – 9:00 pm

Using Quality Improvement Tools to Enhance Patient Care and Training in Genetics
Location: Room 611-614 – 6th floor, Washington State Convention Center
SPECIAL INTEREST GROUP ACTIVITIES AND FORUMS

5:45 pm – 7:45 pm

Public Health SIG Forum and Business Meeting – Hot Topics 2019: Challenges, Controversies, Conversations
Location: Room 2AB – 2nd floor, Washington State Convention Center

5:45 pm – 7:45 pm

Adult Genetics SIG Forum and Executive Committee Meeting – Novel Therapeutics in Adult Genetic Disease
Location: Room 3A – 3rd floor, Washington State Convention Center

SATELLITE SYMPOSIA (PRE-REGISTRATION REQUIRED)

6:00 pm – 7:30 pm

Exploring a New Frontier in Cancer Genetics – Clinical Insights and Evolving Concepts in The DNA Damage Response Pathway
Location: Ballroom 6B – 6th floor, Washington State Convention Center

6:00 pm – 7:30 pm

Ex Vivo Gene Therapy for Lysosomal Storage Disorders - The Path from Promise to Patient
Location: Ballroom 6A – 6th floor, Washington State Convention Center

6:00 pm – 7:30 pm

Fabry Disease: Know When to Treat
Location: Room 602-604 – 6th floor, Washington State Convention Center

Satellite Symposia are scientific/clinical programs supported by commercial or not-for-profit entities to broaden the educational experience for meeting attendees and offer educational content through speaker presentations, including CME and non–CME accredited activities.

NOTE: These activities have been independently organized and/or accredited by an organization other than the American College of Medical Genetics and Genomics (ACMG) and are not affiliated with the official ACMG scientific program.

Complete details for these sessions can be found on the ACMG Meeting website and the Program Supplement “Satellite Symposia” found in the registration packet.

EVENING EVENTS

6:00 pm – 7:30 pm

Welcome Reception for Trainees, Fellows, Residents and Students
Location: 2AB Lobby – 2nd floor, Washington State Convention Center

Fellows, Residents, Trainees and Students are invited to attend a welcome reception where they will have a chance to meet other young professionals and network with the ACMG and ABMGG Leadership.
Daily Schedule – Educational Sessions, Events and Meetings

Indicates this session will be live streamed during the meeting.

Wednesday, April 3

COMMITTEE & BOARD MEETINGS

6:30 am – 8:30 am
ACMG Maintenance of Certification Committee Meeting
Location: Room 208 – 2nd floor, Washington State Convention Center

7:00 am – 8:00 am
ACMG Genetics and Genomics Review Course Faculty Meeting
Location: Room 209 – 2nd floor, Washington State Convention Center

7:00 am – 8:30 am
ClinGen Actionability Workgroup Meeting
Location: Room 303 – 3rd floor, Washington State Convention Center

7:00 am – 9:00 am
ClinGen Sequence Variant Interpretation Workgroup Meeting (Closed Meeting)
Location: Room 304 – 3rd floor, Washington State Convention Center

7:00 am – 9:00 am
ACMG Lab Quality Assurance Full Committee Meeting
Location: Room 205 – 2nd floor, Washington State Convention Center

7:00 am – 9:30 am
ACMG Professional Practice and Guidelines Committee Meeting
Location: Room 204 – 2nd floor, Washington State Convention Center

7:00 am – 10:00 am
ClinGen Somatic Workgroup Meeting
Location: Room 307 – 3rd floor, Washington State Convention Center

7:30 am – 9:30 am
ACMG Social, Ethical and Legal Issues Committee Meeting
Location: Room 203 – 2nd floor, Washington State Convention Center

12:00 pm – 1:15 pm
ACMG Membership Committee Meeting
Location: Room 305 – 3rd floor, Washington State Convention Center

12:00 pm – 1:15 pm
ACMG Secondary Findings Maintenance Workgroup Meeting
Location: Room 303 – 3rd floor, Washington State Convention Center
12:00 pm – 1:15 pm
**ClinGen Ancestry and Diversity Workgroup Meeting**
*Location: Room 307 – 3rd floor, Washington State Convention Center*

12:00 pm – 1:15 pm
**ACMG Finance Committee Meeting**
*Location: Room 304 – 3rd floor, Washington State Convention Center*

7:30 pm – 9:00 pm
**GIM Editorial Board Meeting**
*Location: Room 209 – 2nd floor, Washington State Convention Center*

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**ANCILLARY EVENTS**

12:00 pm – 1:00 pm
**AAP Council on Genetics Joint Awards Luncheon**
*Location: Room 201 – 2nd floor, Washington State Convention Center*

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**SATELLITE SYMPOSIA (PRE-REGISTRATION REQUIRED)**

8:00 am – 9:30 am
**Exploring the Genetics and Management of Fabry Disease**
*Location: Room 606-609 – 6th floor, Washington State Convention Center*

8:00 am – 9:30 am
**The Medically Actionable Genome: Clinical Utility of WGS Through Reproduction, Rare Disease, and Oncology**
*Location: Ballroom 6AB – 6th floor, Washington State Convention Center*

8:00 am – 9:30 am
**Diminishing the Burden of Phenylketonuria During Adulthood: New Science & Encouraging Stories Highlighting the Critical Role of Adjunctive Therapy**
*Location: Room 602-604 – 6th floor, Washington State Convention Center*

8:00 am – 9:30 am
**Are You Ready for Gene Replacement Therapy? Example from Spinal Muscular Atrophy**
*Location: Ballroom 6C – 6th floor, Washington State Convention Center*

Satellite Symposia are scientific/clinical programs supported by commercial or not-for-profit entities to broaden the educational experience for meeting attendees and offer educational content through speaker presentations, including CME and non-CME accredited activities.

**NOTE:** These activities have been independently organized and/or accredited by an organization other than the American College of Medical Genetics and Genomics (ACMG) and are not affiliated with the official ACMG scientific program.

Complete details for these sessions can be found on the ACMG Meeting website and the Program Supplement “Satellite Symposia” found in the registration packet.
WORKSHOPS

8:00 am – 9:30 am

Trainee Workshop: Publishing Your Scholarly Work: Soup to Nuts
Location: Room 611-613 – 6th floor, Washington State Convention Center

1:30 pm – 3:30 pm

ACMG Revision to Interpreting Sequence Variation Laboratory Practice Resource Workgroup – Open Forum
Location: Room 606-609 – 6th floor, Washington State Convention Center

PLATFORM PRESENTATIONS

12:15 pm – 1:15 pm

Industry Platform Presentations
Location: Ballroom 6C – 6th floor, Washington State Convention Center

No Educational Credits are available for this session

Moderator: Christian Schaaf, MD, PhD, FACMG, University of Heidelberg, Germany

Abstracts:

#227 Positive Yield of Genetic Testing for Patients with Epilepsy is Associated with Age of Seizure-Onset, Presence of Neurological Features, and Trio Testing
Dianalee McKnight, PhD, GeneDx

#166 Characteristics and Clinical Correlations of Somatic Mosaic Large Rearrangements Identified by Pan Cancer NGS Panel Test
Shujuan Pan, PhD, FACMG, Myriad Genetic Laboratories, Inc.

#611 Use of a Chatbot to Offer Pre-test Education for Expanded Carrier Screening
Shivani Nazareth, MS, CGC, Clear Genetics

#659 Three-quarters of Upgraded Negative Reports Due to Reclassification are Due to Lab-initiated Proactive Reanalysis
Kelly Radtke, PhD, Ambry Genetics
SCIENTIFIC PLENARY SESSION
10:00 am – 12:00 pm

Presentation of the 2019 ACMG Foundation Awards

Presidential Plenary – Perspectives and Possibilities: Partnering for the Future of Medical Genetics

Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E./NSGC Contact Hours

Moderator: Louanne Hudgins, MD, FACMG, Stanford University School of Medicine

Presentations:

- 10:00 am – 10:15 am
  2019 ACMG Foundation Awards

- 10:15 am – 10:30 am
  Perspectives and Possibilities: Partnering for the Future of Medical Genetics
  Louanne Hudgins, MD, FACMG, Stanford University School of Medicine

- 10:30 am – 10:55 am
  Advocacy and Public Policy – How the Geneticist Can Help
  Susan Klugman, MD, Albert Einstein College of Medicine/Montefiore Medical Center

- 10:55 am – 11:20 am
  The All of Us Research Program: Implications for Medical Genetics and Genomics
  Bruce Korf, MD, PhD, FACMG, University of Alabama at Birmingham

- 11:20 am – 11:45 am
  “We’ve Got a Pill for That”: The New Reality of Treatable Genetic Disorders
  Shawn McCandless, MD, FACMG, FAAP, University of Colorado Denver and Children’s Hospital Colorado

- 11:45 am – 12:00 pm
  Audience Response Questions
SCIENTIFIC CONCURRENT SESSIONS

1:30 pm – 3:30 pm

■ Pharmacogenomics – Promises, Potential, Pitfalls, and Practice
  Location: Ballroom 6C – 6th floor, Washington State Convention Center

This session is eligible for up to (2) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderators: Shannon Manzi, PharmD, BCPPS, FPPAG, Harvard Medical School, Boston Children's Hospital; Wen-Hann Tan, BMBS, Harvard Medical School, Boston Children's Hospital

Presentations:
  1:30 pm – 1:55 pm
  Pharmacogenomics – Resources and Implementation Models
  James Hoffman, PharmD, St. Jude Children’s Research Hospital
  1:55 pm – 2:20 pm
  Pharmacogenomics: Knowledge Resources to Support Patient Care
  Teri Klein, PhD, Stanford University
  2:20 pm – 2:45 pm
  Multiple Case Discussions, Including Human Leukocyte Antigens (HLAs) and Serious Skin Reactions, Cytochrome P450 Enzymes (CYPs) and Drug Toxicity, Age Based Maturation, and N-Acetyltransferase 2 (NAT2) and Drug Efficacy
  Shannon Manzi, PharmD, BCPPS, FPPAG, Harvard Medical School, Boston Children’s Hospital
  2:45 pm – 3:10 pm
  Setting Up a Pharmacogenomics Service
  Jonathan Picker, MBChB, PhD, FACMG, Harvard Medical School, Boston Children’s Hospital
  3:10 pm – 3:30 pm
  Discussion and Q&A

1:30 pm – 3:30 pm

■ Prenatal Diagnostic Challenges
  Location: Ballroom 6B – 6th floor, Washington State Convention Center

This session is eligible for up to (2) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderators: Jennifer King, MD, Albert Einstein College of Medicine, NY; Myra Wick, MD, PhD, Mayo Clinic, MN

This session to be presented in the Diagnostic Dilemma format was created in an effort to increase the quantity and quality of Prenatal genetic educational opportunities. The session will be an interactive session which will allow genetics professionals to present cases of rare knowns and unknowns.
1:30 pm – 3:30 pm

**The Eyes Have It: Genetics of Ophthalmological Disorders**

*Location: Ballroom 6E – 6th floor, Washington State Convention Center*

This session is eligible for up to (2) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderator:** Fuki Hisama, MD, FACMG, FAAN, University of Washington School of Medicine

**Presentations:**

1:30 pm – 2:00 pm
- Genetics of Congenital Cranial Dysinnervation Disorders
  
  *Elizabeth Engle, MD,* Boston Children’s Hospital, Harvard Medical School / HHMI

2:00 pm – 2:30 pm
- Clinical and Genetic Evaluation of Retinal Dystrophies
  
  *Mark Pennesi, MD, PhD,* Oregon Health & Science University, Casey Eye Institute

2:30 pm – 3:00 pm
- Utilization of Next Generation Sequencing in Molecular Diagnosis of Inherited Retina Disorders
  
  *Jennifer Schleit, PhD, FACMG,* Blueprint Genetics

3:00 pm – 3:30 pm
- Non-Coding Variants in Retinal Dystrophies
  
  *Timothy Cherry, PhD,* University of Washington/Seattle Children’s Research Institute

1:30 pm – 3:30 pm

**Social, Ethical and Legal Perspectives on Emerging Genomic Population Health Initiatives**

*Location: Ballroom 6A – 6th floor, Washington State Convention Center*

This session is eligible for up to (2) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderators:** Robert Best, PhD, FACMG, University of South Carolina School of Medicine, Greenville, SC; Leslie Manace, MD, MPhil, Kaiser Permanente Oakland Medical Center, CA

**Presentations:**

1:30 pm – 1:50 pm
- Genomic Population Health - Defining Value in Tier 1 Tests
  
  *Jonathan Berg, MD, PhD, FACMG,* University of North Carolina at Chapel Hill

1:50 pm – 2:10 pm
- Ethics at the Interface of Genomics and Population Health; Achieving the Quadruple Aim
  
  *Lisa Lee, PhD, MA, MS,* Virginia Tech

2:10 pm – 2:30 pm
- Challenges to Prevailing Legal Systems Inherent to Genomic Medicine
  
  *Leslie Francis, JD,* University of Utah

2:30 pm – 2:50 pm
- Social Perspective: Engagement of Patients and Participants as Partners
  
  *Marc Williams, MD, FAAP, FACMG, FACMI,* Geisinger

2:50 pm – 3:00 pm
- Discussion and Q&A
SCIENTIFIC CONCURRENT SESSIONS

4:00 pm – 5:30 pm

New ACMG Practice Guidelines from the Professional Practice and Guidelines (PP&G) Committee

Location: Room 606-609 – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderators: David Miller, MD, PhD, FACMG, Boston Children’s Hospital; Douglas Stewart, MD, National Cancer Institute

Presentations:

4:00 pm – 4:10 pm
The ABC’s of ACMG’s PP&G: Introduction, Goals and Summary of 2018 Session
David Miller, MD, PhD, FACMG, Boston Children’s Hospital

4:10 pm – 4:25 pm
The Latest and Greatest from PP&G: Highlights from Recent PP&G Documents – Chromosomal Microarray Testing
David Miller, MD, PhD, FACMG, Boston Children’s Hospital

4:25 pm – 4:40 pm
The Latest and Greatest from PP&G: Highlights from Recent PP&G Documents – Clinical Utility of Exome and Genome Sequencing for Congenital Anomalies
Jun Shen, PhD, FACMG, Brigham and Women’s Hospital

4:40 pm – 4:55 pm
The Latest and Greatest from PP&G: Highlights from Recent PP&G Documents – Points to Consider in Tumor/Germline Sequencing
Marilyn Li, MD, Children’s Hospital of Philadelphia

4:55 pm – 5:10 pm
The Latest and Greatest from PP&G: Highlights from Recent PP&G Documents – Neurofibromatosis Type 1 Clinical Practice Resources for Adults and Children
Douglas Stewart, MD, National Cancer Institute

5:10 pm – 5:30 pm
ACMG PP&G Brainstorming: Solicitation of Ideas for New Document Ideas Important to ACMG Membership
David Miller, MD, PhD, FACMG and Douglas Stewart, MD, National Cancer Institute
ORAL PLATFORM PRESENTATIONS

4:00 pm – 5:30 pm

Genetic Counseling

Location: Ballroom 6B – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.@/NSGC Contact Hours

Moderator: Pilar Magoulas, MS, Baylor College of Medicine

Abstracts:

4:00 pm – 4:15 pm
#1 Phenotype Genotype Variability Among Sibships with Spinal Muscular Atrophy: Implications for Newborn Screening and Early Treatment Intervention
Jin Yun Chen, MS, CGC, Massachusetts General Hospital

4:15 pm – 4:30 pm
#2 The Importance of Genetic Counselling for Genome-wide Sequencing: Measuring its Impact on Knowledge and Decisional Conflict
Shelin Adam, BSc, MSc, University of British Columbia

4:30 pm – 4:45 pm
#3 Self-Reported Personal and Family History among Participants in a Population-Based Genomic Research Initiative
Whitley Kelley, MS, CGC, HudsonAlpha Institute for Biotechnology

4:45 pm – 5:00 pm
#4 Uptake of Genetic Counseling and Testing after Positive First Trimester Screening in the Pre- and Post- Non-Invasive Prenatal Screening Eras
Kathleen Aaron, MS, CGC, Albert Einstein College of Medicine/Montefiore Medical Center

5:00 pm – 5:15 pm
#5 Impact of Genetic Counseling and Patient Follow-up Calls on Uptake of Risk-Reducing Actions among Biobank Participants Receiving Actionable Sequencing Results
Tara Schmidlen, MS, LGC, Geisinger

5:15 pm – 5:30 pm
#6 When Should We Make Pre-test Genetic Counseling Optional? Survey Results from Participants in a Large Genomic Sequencing Study
Richard Sharp, PhD, Mayo Clinic
4:00 pm – 5:30 pm

Clinical Genetics and Therapeutics

Location: Ballroom 6E – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderator: Anne Slavotinek, MBBS, PhD, University of California, San Francisco

Abstracts:

4:00 pm – 4:15 pm
#7 Physical Abnormalities Driven by the Genotype in Patients with Fanconi Anemia
Moises Fiesco-Roa, MD, Mexican Board of Human Genetics; National Cancer Institute, NIH

4:15 pm – 4:30 pm
#8 Findings of the Whole Genome Sequencing Core of the Undiagnosed Diseases Network
Liz Worthey, PhD, HudsonAlpha Institute for Biotechnology

4:30 pm – 4:45 pm
#9 Rapid Turnaround Whole Exome Sequencing for Critically Ill Neonates
Cynthia Gubbels, MD, PhD, Boston Children’s Hospital

4:45 pm – 5:00 pm
#10 Factors Affecting Outcomes in Classic Infantile Pompe Disease Patients Treated with Enzyme Replacement Therapy: Lessons Learned from the Pompe Registry
Priya Kishnani, MD, MBBS, Duke University Medical Center

5:00 pm – 5:15 pm
#11 Diagnostic Yields of Clinical Testing for Skeletal Dysplasia Disorders Using Different Methods
Tatiana Tvrdik, PhD, MS, University of Utah School of Medicine; ARUP Laboratories

5:15 pm – 5:30 pm
#12 AVXS-101 Gene-Replacement Therapy for Spinal Muscular Atrophy: A Comparative Study With a Prospective Natural History Cohort
Richard Shell, MD, The Ohio State University Wexler Center
4:00 pm – 5:30 pm

- **Laboratory Genetics and Genomics**
  
  *Location: Ballroom 6C – 6th floor, Washington State Convention Center*

  This session is eligible for up to (1.5) CME/CEU/P.A.C.E®/NSGC Contact Hours

  **Moderator:** *Eli Williams, PhD, FACMG, UVA School of Medicine*

  **Abstracts:**

  4:00 pm – 4:15 pm
  
  #13 *Making sense of missense: a pathway-based approach to functional assays for improved clinical variant interpretation*
  
  *Sarah Brnich, BS, University of North Carolina School of Medicine*

  4:15 pm – 4:30 pm
  
  #14 *Analysis of Mosaicism for Sequence and Copy Number Variants in a Broad Diversity of Hereditary Disorders in a Large Clinical Cohort*
  
  *Rebecca Truty, PhD, Invitae*

  4:30 pm – 4:45 pm
  
  #15 *The Gene Curation Coalition: A Global Effort to Harmonize Gene-level Resources*
  
  *Marina DiStefano, PhD, Harvard Medical School*

  4:45 pm – 5:00 pm
  
  #16 *A Rigorous Examination of the Need for Sanger Confirmation in Clinical Genetic Testing*
  
  *Stephen Lincoln, BS, Invitae*

  5:00 pm – 5:15 pm
  
  #17 *Systematic Benchmarking of Copy-number Detection using Exome Sequencing Data in a Clinical Setting*
  
  *Ramakrishnan Rajagopalan, MS, The Children’s Hospital of Philadelphia*

  5:15 pm – 5:30 pm
  
  #18 *ACMG/CGC Technical Laboratory Standards for Interpretation and Reporting of Acquired Copy Number Abnormalities and Copy-Neutral Loss of Heterozygosity in Neoplastic Disorders*
  
  *Fady Mikhail, MD, PhD, FACMG, University of Alabama at Birmingham*
4:00 pm – 5:30 pm

**Legal and Ethical Issues; Education**  
*Location: Ballroom 6A – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderator:** Monica Giovanni, MS, CGC, ACMG Education Committee Chair and Liaison to the Program Committee

**Abstracts:**

4:00 pm – 4:15 pm  
**#19 Undiagnosed or Underdiagnosed: Is Insurance Denial Impacting Clinical Diagnosis in Level IV Neonatal Intensive Care Units?**  
Kristen Fishler, MS, University of Nebraska Medical Center

4:15 pm – 4:30 pm  
**#20 Discordant Genomic Choices among Adolescents and Parents**  
Cynthia Prows, MSN, APRN, Cincinnati Children’s Hospital Medical Center

4:30 pm – 4:45 pm  
**#21 Relationship between genomic knowledge and familial communication of genomic information: Insights from eMERGE III**  
Sukh Makhnoon, MS, PhD, MD, Anderson Cancer Center

4:45 pm – 5:00 pm  
**#22 Medical Genetics for Millennials: Innovation and Modification of the Baylor College of Medicine Curriculum**  
Kevin Glinton, MD, PhD, FAAP, Baylor College of Medicine

5:00 pm – 5:15 pm  
**#23 Optimizing Genetics Online Resources for Diverse Readers**  
Jiyoo Chang, BA, University of California San Francisco

5:15 pm – 5:30 pm  
**#24 Physician Perceived Utility and Preparedness for Newborn Genomic Sequencing: Findings from the BabySeq Project**  
Carrie Blout, MS, CGC, Brigham and Women’s Hospital

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**EXHIBIT/POSTER HALL**

5:30 pm – 7:30 pm

**Exhibits Opening and Opening Reception**  
*Location: Exhibit Halls 4ABCD – 4th floor, Washington State Convention Center*

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**EVENING EVENTS**

7:30 pm – 9:00 pm

**ClinGen Reception (Ancillary event)**  
*Room 606-609 – 6th floor, Washington State Convention Center*
Daily Schedule – Educational Sessions, Events and Meetings

Indicates this session will be live streamed during the meeting.

Thursday, April 4

**COMMITTEE & BOARD MEETINGS**

6:45 am – 8:00 am

**ClinGen Hereditary Cancer Workgroup Meeting**  
*Location: Room 305 – 3rd floor, Washington State Convention Center*

7:00 am – 8:00 am

**ACMG Economics of Genetic Services Committee Meeting**  
*Location: Room 205 – 2nd floor, Washington State Convention Center*

7:00 am – 8:00 am

**ClinGen Metabolism Workgroup Meeting**  
*Location: Room 303 – 3rd floor, Washington State Convention Center*

7:00 am – 8:00 am

**ACMG Exome Sequencing Clinical Utility Workgroup Meeting**  
*Location: Room 204 – 2nd floor, Washington State Convention Center*

11:00 am – 1:00 pm

**ACMG Foundation Board of Directors Meeting**  
*Location: Room 209 – 2nd floor, Washington State Convention Center*

11:00 am – 1:00 pm

**ClinGen Neurodevelopmental Workgroup Meeting**  
*Location: Room 205 – 2nd floor, Washington State Convention Center*

11:00 am – 1:00 pm

**ACMG Therapeutics Committee Meeting**  
*Location: Room 204 – 2nd floor, Washington State Convention Center*

11:00 am – 1:00 pm

**ACMG Lab QA Workgroup on the Risk Categorization of LDTs (closed)**  
*Location: Room 203 – 2nd floor, Washington State Convention Center*
Sessions and Events

SCIENTIFIC PLENARY SESSION

8:00 am – 9:00 am

Featured Platform Presentations

Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center

This session is eligible for up to (1.0) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderator: Christian Schaaf, MD, PhD, FACMG, University of Heidelberg, Germany

Abstracts:

8:00 am – 8:15 am

#25 Healthy Nevada Project Population Health Genetic Screening in Northern Nevada: At Risk Individuals are Missed Using Current Guidelines

Joseph Grzymski, PhD, Renown Health

8:15 am – 8:30 am

#26 All That Glitters Is Not Gold: Circulating TP53 Mutations and Li-Fraumeni Syndrome (LFS)

Alison Robertson, LGC, Dana Farber Cancer Institute

8:30 am – 8:45 am

#27 Limitations of HBOC Direct-To-Consumer Genetic Screening: False Positives, False Negatives and Everything in Between

Edward Esplin, MD, PhD, FACMG, FACP, Invitae

8:45 am – 9:00 am

#28 ClinPhen Extracts and Prioritizes Patient Phenotypes Directly from Medical Records to Expedite Genetic Disease Diagnosis

Johannes Birgmeier, MSc, Stanford University
SCIENTIFIC PLENARY SESSION

9:00 am – 10:00 am

TED-Style Talks

*Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center*

This session is eligible for up to (1.0) CME/CEU/P.A.C.E./NSGC Contact Hours

*Moderator: Christopher Cunniff, MD, FACMG, FAAP, Weill Cornell Medical College, Weill Cornell Medicine, NY*

9:00 am – 9:06 am

**Welcome to the ACMG TED-Style Talks**

*Christopher Cunniff, MD, FACMG, FAAP, Weill Cornell Medical College, Weill Cornell Medicine*

9:06 am – 9:24 am

**Multiomic Investigation from the Urban Biome to Outer Space**

*Christopher Mason, PhD, Weill Cornell Medicine, Mason Lab*

9:24 am – 9:42 am

**Next Generation Education Can Save Our Species**

*Debra Regier, MD, PhD, Children’s National Medical Center, Rare Disease Institute*

9:42 am – 10:00 am

**Defective? A Somatic Carrier’s Strategy to Impact Duchenne Muscular Dystrophy**

*Patricia Furlong, RN, BSN, Parent Project Muscular Dystrophy*

EXHIBIT/POSTER HALL

9:30 am – 4:30 pm

Exhibit Hall Open

*Location: Exhibit Halls 4ABCD – 4th floor, Washington State Convention Center*

10:00 am – 10:30 am

**Refreshment Break**

10:00 am – 11:30 am

**Poster Presentations – odd numbers**

*This session is eligible for up to (1.5) NSGC CEU Contact Hours*

10:30 am – 1:15 pm

**Learning Lounges**

10:30 am – 12:45 pm

**Exhibit Theater Presentations**

11:30 am – 1:00 pm

**Concession Lunch available for purchase in the Exhibit Hall**

3:30 pm – 4:00 pm

**Refreshment Break**
LUNCHEONS

11:45 am – 1:15 pm

Genetic Counselor’s Luncheon and Forum: Risk Alleles - Discovery, Classification, and Reporting (pre-registration required)
Location: Metropolitan Ballroom AB, 3rd floor, Sheraton Grand Seattle

This session is eligible for up to (1.0) NSGC CEU Contact Hours

Moderator: Ryan Schmidt, MD, PhD, Keck School of Medicine, University of Southern California, Children’s Hospital Los Angeles

Presentations:

12:00 pm – 12:15 pm
Welcome and Presentation of the Carolyn Mills Lovell Award
Pilar Magoulas, CGC, Baylor College of Medicine; David Flannery, MD, FACMG, Cleveland Clinic

12:15 pm – 12:40 pm
Where to Draw the Line? Clinical Reporting of Low Penetrance Variants in the Context of Whole Genome Sequencing in Healthy Adults
Birgit Funke, PhD, FACMG, Harvard Medical School, Veritas Genetics

12:40 pm – 1:10 pm
Clinical Challenges with Results with Different Pathogenicity Classifications and Risk Levels
Heather Hampel, MS, LGC, The Ohio State University, Wexner Medical Center

1:10 pm – 1:15 pm
Discussion and Q&A
Ryan Schmidt, MD, PhD, Keck School of Medicine, University of Southern California, Children’s Hospital Los Angeles

11:45 am – 1:15 pm

Trainee/Mentor Luncheon (pre-registration required)
Location: Room 606-609 – 6th floor, Washington State Convention Center

This luncheon provides an opportunity for trainees (graduate students, postdoctoral fellows and clinical fellows) to meet and talk informally with senior members of the College about career options, goals, and professional opportunities.
SCIENTIFIC PLENARY SESSION
1:30 pm – 3:30 pm

50th Annual March of Dimes Clinical Genetics Conference – From Causes to Care: State of the Art Update of Common Birth Defects

Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center

This session is eligible for up to (2.0) CME/CEU/P.A.C.E./NSGC Contact Hours

Moderator: Maximilian Muenke, MD, FACMG, NHGRI, NIH

Presentations:

1:30 pm – 1:35 pm
Welcome and Introduction of Pruzansky Lecture

1:35 pm – 2:05 pm
Holoprosencephaly: Causes and Care from Preconception to Adulthood (Pruzansky Lecture)
Maximilian Muenke, MD, FACMG, NHGRI, NIH

2:05 pm – 2:30 pm
Congenital Cardiac Anomalies from Birth to Adulthood
Bruce Gelb, MD, Icahn School of Medicine at Mount Sinai

2:30 pm – 2:55 pm
Renal Ciliopathies and Congenital Anomalies of the Kidneys and Urinary Tract: From Whole Exome Sequencing to Precision Medicine
Friedhelm Hildebrandt, MD, Boston Children’s Hospital, Harvard Medical School

2:55 pm – 3:20 pm
Identifying Genetic and Environmental Factors Causing Birth Defects: Disease-gene Discovery and Roles for Oxygen, and Vitamin B3
Sally Dunwoodie, PhD, University of New South Wales, Victor Chang Cardiac Research Institute

3:20 pm – 3:30 pm
Discussion
SCIENTIFIC CONCURRENT SESSIONS

4:15 pm – 5:45 pm

The Chicken and the Egg Dilemma in Public Health: Timing and Scope of Genetic Screening as Innovative Therapeutic Interventions Incubate – R. Rodney Howell Symposium

Location: Ballroom 6B – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderators: Lynn Bush, PhD, MS, MA, Columbia University Medical Center, NY; R. Rodney Howell, MD, FAAP, FACMG, University of Miami, Miller School of Medicine, FL

Presentations:

4:15 pm – 4:25 pm
Introduction, Ethical and Psychosocial Challenges
Lynn Bush, PhD, MS, MA, Columbia University Medical Center

4:25 pm – 4:35 pm
Panethnic Expanded Carrier Screening - How Early, How Much, and for Whom?
Ignatia Van den Veyver, MD, Baylor College of Medicine

4:35 pm – 4:45 pm
Fifty Years’ Wisdom of Wilson-Jungner – Point
Susan Berry, MD, FACMG, FAAP, University of Minnesota

4:45 pm – 4:55 pm
Why Thoughtful Expansion of Newborn Screening is Now Needed More Than Ever – Counterpoint
Olaf Bodamer, MD, PhD, Boston Children’s Hospital/Harvard Medical School

4:55 pm – 5:10 pm
Q&A

5:10 pm – 5:20 pm
A Tale of Genetic Newborn Hearing Screening from One Million
Jun Shen, PhD, FACMG, Brigham and Women’s Hospital

5:20 pm – 5:30 pm
Genetic Screening and Novel Therapies for Spinal Muscular Atrophy
Kathryn Swoboda, MD, FACMG, FAAN, Massachusetts General Hospital

5:30 pm – 5:45 pm
Q&A
4:15 pm – 5:45 pm

**Seeing Into the Future: Gene Therapy Has Arrived**  
*Location: Ballroom 6C – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E./NSGC Contact Hours

**Moderators:** *Emma Bedoukian, MS, CGC, Children’s Hospital of Philadelphia, PA; Linda Reis, MS, CGC, Medical College of Wisconsin, WI*

**Presentations:**

- **4:15 pm – 4:30 pm**  
  **Overview of Gene Therapy Techniques**  
  *Mark Pennesi, MD, PhD, Oregon Health & Science University, Casey Eye Institute*

- **4:30 pm – 4:45 pm**  
  **Ophthalmic Genetics Overview: The Eye as a Target for Gene Therapy**  
  *Dana Schlegel, MS, MPH, CGC, Kellogg Eye Center*

- **4:45 pm – 5:00 pm**  
  **The First FDA Approved Gene Therapy: Voretigene Neparvovec**  
  *Karmen Trzupek, MS, InformedDNA*

- **5:00 pm – 5:20 pm**  
  **Gene Therapy for Fabry Disease: From Concept to Clinic to Company**  
  *Jeffrey Medin, PhD, Medical College of Wisconsin*

- **5:20 pm – 5:35 pm**  
  **Gene Therapy for Spinal Muscular Atrophy**  
  *Melissa Gibbons, MS, CGC, University of Colorado School of Medicine / Children’s Hospital Colorado*

- **5:35 pm – 5:45 pm**  
  **Question and Answer with Panel and Conclusion**
4:15 pm – 5:45 pm

**Next Level Genomic Data Interpretation: Machine Learning, Methylome Analysis and 3D Mapping**

*Location: Ballroom 6E – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E./NSGC Contact Hours

**Moderators:** Adrian Dubuc, PhD, Brigham and Women’s Hospital, Harvard Medical School, MA; Jennelle Hodge, PhD, FACMG, Indiana University, IN

**Presentations:**

- 4:15 pm – 4:45 pm
  - **Impact of Machine Learning on Variant Interpretation**
    - Jochen Lennerz, MD, PhD, Massachusetts General Hospital and Harvard Medical School
  - **Implications of Structural Alteration to Three-Dimensional Genome Organization**
    - Michael Talkowski, PhD, Massachusetts General Hospital / Harvard Medical School / Broad Institute
- 5:15 pm – 5:45 pm
  - **Evaluation of the DNA Methylome for Classification of Mendelian Disorders**
    - Bekim Sadikovic, PhD, Western University, London Health Sciences Centre and St. Joseph’s Health Care London

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4:15 pm – 5:45 pm

**Advances in the Genetics of Inherited Heart Disease**

*Location: Ballroom 6A – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E./NSGC Contact Hours

**Moderators:** Amy Roberts, MD, Boston Children’s Hospital/Harvard Medical School, MA

**Presentations:**

- 4:15 pm – 4:35 pm
  - **What have ExAC and gnomAD Taught us About the Genetics of Inherited Heart Disease?**
    - Colleen Caleshu, MS, CGC, Stanford Center for Inherited Cardiovascular Disease
  - **Ion Channel Disease: Phenotype and Interpretation of Genetic Findings**
    - Andrew Krahn, MD, University of British Columbia
- 4:55 pm – 5:15 pm
  - **Arrhythmogenic Cardiomyopathies: Phenotype and Interpretation of Genetic Findings**
    - Dominic Abrams, MD, MRCP, MBA, Boston Children’s Hospital
  - **Management of Secondary Findings in Genes Associated with Inherited Heart Disease**
    - Robyn Hylind, MS, CGC, Boston Children’s Hospital
- 5:35 pm – 5:45 pm
  - Discussion and Q&A
**EVENING EVENTS**

6:00 pm – 7:00 pm

**ACMG Annual Membership Business Meeting (ACMG Members Only)**  
*Location: Room 611-614 – 6th floor, Washington State Convention Center*

7:30 pm – 9:00 pm

**ACMG Foundation and the David L. Rimoin Donors Recognition Reception**  
*(by invitation)*  
*Location: Cirrus Ballroom – Sheraton Grand Seattle, 35th floor of the Pike Tower*
Daily Schedule – Educational Sessions, Events and Meetings

Indicates this session will be live streamed during the meeting.

Friday, April 5

**COMMITTEE & BOARD MEETINGS**

7:00 am – 8:00 am

**ACMG Genomic Screening of Asymptomatic Patients Committee Meeting**
Location: Room 304 – 3rd floor, Washington State Convention Center

7:00 am – 8:30 am

**ACMG Education and CME Committee Meeting**
Location: Room 307 – 3rd floor, Washington State Convention Center

7:00 am – 8:30 am

**ACMG Population Screening PTC Workgroup Meeting**
Location: Room 303 – 3rd floor, Washington State Convention Center

11:30 am – 1:00 pm

**ACMG Board of Directors Meeting 2**
Location: Room 208 – 2nd floor, Washington State Convention Center

7:00 pm – 7:30 pm

**ClinGen Hearing Loss Workgroup Meeting**
Location: Room 203 – 2nd floor, Washington State Convention Center

7:30 pm – 9:00 pm

**ACMG Program Committee Meeting**
Location: Room 204 – 2nd floor, Washington State Convention Center

**ANCILLARY EVENTS**

7:30 pm – 9:00 pm

**Shabbat Dinner (Ancillary Event)**
Location: Room 201 – 2nd floor, Washington State Convention Center
**SATELLITE SYMPOSIA (PRE-REGISTRATION REQUIRED)**

7:15 am – 8:45 am  
**Improving Care for Patients with Pompe Disease: A Look at Emerging Therapies and Multidisciplinary Management Strategies**  
*Location: Room 606-609 – 6th floor, Washington State Convention Center*

7:15 am – 8:45 am  
**A Precision Medicine for Adult Patients with Amenable Variants and Fabry Disease**  
*Location: Room 611-614 – 6th floor, Washington State Convention Center*

7:15 am – 8:45 am  
**Effects of Hyperammonemia on Brain Function: What Urea-lly Need to Know**  
*Location: Ballroom 6B – 6th floor, Washington State Convention Center*

Satellite Symposia are scientific/clinical programs supported by commercial or not-for-profit entities to broaden the educational experience for meeting attendees and offer educational content through speaker presentations, including CME and non–CME accredited activities.

NOTE: These activities have been independently organized and/or accredited by an organization other than the American College of Medical Genetics and Genomics (ACMG) and are not affiliated with the official ACMG scientific program.

Complete details for these sessions can be found on the ACMG Meeting website and the Program Supplement “Satellite Symposia” found in the registration packet.

**Sessions and Events**

**SCIENTIFIC PLENARY SESSION**

9:00 am – 10:00 am  
**A Tribute to Arno G. Motulsky, MD: A Giant of Medical Genetics**  
*Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center*

This session is eligible for up to (1.0) CME/CEU/P.A.C.E.@/NSGC Contact Hours

**Moderator:** Fuki Hisama, MD, FACMG, FAAN, University of Washington School of Medicine

**Presentations:**

<table>
<thead>
<tr>
<th>Time</th>
<th>Presentation</th>
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<tbody>
<tr>
<td>9:00 am – 9:10 am</td>
<td><em>In Memoriam: Arno G. Motulsky, MD (1923 -2018)</em></td>
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<td>Gail Jarvik, MD, PhD, University of Washington</td>
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<td>9:10 am – 9:35 am</td>
<td><em>Arno’s Influence: Serendipity, Research Opportunity, Care for Families, and Biological Insights</em></td>
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<td>Peter Byers, MD, University of Washington</td>
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<td>9:35 am – 10:00 am</td>
<td><em>Structural Variation, Disease and the Evolution of the Human Genome</em></td>
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<td>Evan Eichler, PhD, University of Washington School of Medicine &amp; HHMI</td>
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EXHIBIT/POSTER HALL

9:30 am – 1:00 pm

Exhibit Hall Open
Location: Exhibit Halls 4ABCD – 4th floor, Washington State Convention Center

10:00 am – 10:30 am
Refreshment Break

10:30 am – 12:00 pm
Poster Presentations – even numbers
This session is eligible for up to (1.5) NSGC CEU Contact Hours

10:30 am – 11:00 am
Day of Caring Bicycle Presentation

10:30 am – 12:30 pm
Learning Lounges

10:45 am – 1:00 pm
Exhibit Theater Presentations

11:30 am – 1:00 pm
Concession Lunch available for purchase in the Exhibit Hall

SCIENTIFIC CONCURRENT SESSIONS

7:15 am – 8:45 am

Towards Precise Genetic Diagnosis of Primary Mitochondrial DNA Diseases
Location: Ballroom 6C – 6th floor, Washington State Convention Center

Educational credits are not available for this session

Moderator: Renkui Bai, MD, PhD, GeneDx, MD; Marni Falk, MD, FACMG, University of Pennsylvania Perelman School of Medicine, Children’s Hospital of Philadelphia, PA

Presentations:

7:15 am – 7:42 am
Clinical Diagnostic Laboratory and Molecular Approach to Classify Mitochondrial DNA Variant Pathogenicity
Renkui Bai, MD, PhD, GeneDx

7:42 am – 8:04 am
Mitochondrial Medicine Clinic Evaluations to Identify and Assess the Pathogenicity of Mitochondrial DNA Variants for Individual Patients and Families
Marni Falk, MD, FACMG, University of Pennsylvania Perelman School of Medicine, Children’s Hospital of Philadelphia

8:04 am – 8:26 am
Bioinformatics Tools and Approaches to Analyze the mtDNA Genome and Assess the Pathogenicity of Mitochondrial DNA Variants
Xiaowu Gai, PhD, Children’s Hospital Los Angeles

8:26 am – 8:45 am
Panel Discussion
STUDENT SESSION

1:00 pm – 3:30 pm

Careers in Medical Genetics – An Informational Session for Students and Trainees (pre-registration required)
Location: Room 602-604 – 6th floor, Washington State Convention Center

Moderator: Anita Beck, Seattle Children’s Hospital, University of Washington, WA

1:00 pm – 1:30 pm

Lunch – Sponsored by Illumina

Presentations:

1:30 pm – 1:40 pm
Welcome and Introductions
Anita Beck, Seattle Children’s Hospital, University of Washington

1:40 pm – 1:50 pm
A Day in the Life of a Clinical Geneticist
Erin Cooney, MD, University of Texas Medical Branch at Galveston

1:50 pm – 2:00 pm
Clinical Laboratory-based Career Paths
Yuan Ji, PhD, FACMG, ARUP Institute for Clinical & Experimental Pathology, University of Utah School of Medicine

2:00 pm – 2:10 pm
A Career Path in Genetic Counseling
Laura Baker, MGC, CGC, Nemours/Alfred I. duPont Hospital for Children

2:10 pm – 2:20 pm
Tips and Perspectives From a Recent Medical Genetics Residency Training Program Graduate
Anna Hurst, MD, MS, FACMG, University of Alabama

2:20 pm – 2:30 pm
All About the ABMGG
Miriam Blitzer, PhD, FACMG, American Board of Medical Genetics and Genomics

2:30 pm – 3:30 pm
Round Table Discussions
SCIENTIFIC CONCURRENT SESSIONS

1:30 pm – 3:30 pm

- Complexities in Cancer Risk Assessment and Management
  Location: Ballroom 6B – 6th floor, Washington State Convention Center
  This session is eligible for up to (2.0) CME/CEU/P.A.C.E./NSGC Contact Hours
  Moderator: Tuya Pal, MD, FACMG, Vanderbilt University Medical Center, TN

Presentations:

  1:30 pm – 1:45 pm
  Welcome and Session Goals

  1:45 pm – 2:15 pm
  Current Understanding of Risks Associated with Pathogenic Variants in “Moderate Penetrance” Genes
  Mark Robson, MD, Memorial Sloan Kettering Cancer Center

  2:15 pm – 2:45 pm
  Polygenic Risk Scores: Development and Validation
  Amanda Toland, PhD, FACMG, Ohio State University

  2:45 pm – 3:15 pm
  Management of Individuals at Increased Risk for Breast and Ovarian Cancer Who Do Not Have BRCA1/2 Mutations
  Jennifer Litton, MD, The University of Texas MD Anderson Cancer Center

  3:15 pm – 3:30 pm
  Panel Discussion and Q&A
1:30 pm – 3:30 pm

**Cardinal Signs of Common and Uncommon Disorders**

*Location: Ballroom 6E – 6th floor, Washington State Convention Center*

This session is eligible for up to (2.0) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderators:** Leah Burke, MD, FACMG, FAAP, University of Vermont; Helga Toriello, PhD, FACMG, Michigan State University

**Presentations:**

1:30 pm – 1:50 pm

*What Exactly is Amyoplasia?*

Judith Hall, MD, BC Children’s and Women’s Hospital, UBC

1:50 pm – 2:10 pm

**Cardinal Signs of HDAC4-Related Disorders**

Sarah Elsea, PhD, FACMG, Baylor College of Medicine

2:10 pm – 2:30 pm

**Saul-Wilson Syndrome and its Cardinal Manifestations**

Carlos Ferreira, MD, FACMG, NHGRI, NIH

2:30 pm – 2:50 pm

**Cardinal Signs of Stormorken Syndrome**

Danielle Monteil, MD, Naval Medical Center Portsmouth

2:50 pm – 3:10 pm

**POLR1-Related Disorders and Their Cardinal Signs**

Kathryn Weaver, MD, FACMG, FAAP, Cincinnati Children’s Hospital Medical Center

3:10 pm – 3:30 pm

**Update on the Clinical Manifestations of VACTERL**

Virginia Kimonis, MD, MRCP, FACMG, University of California-Irvine Medical Center
1:30 pm – 3:30 pm

**Laboratory Diagnostic Challenges (Constitutional and Neoplastic Cases in Molecular, Cytogenomic, and Biochemical Genetics Specialties)**
*Location: Ballroom 6C – 6th floor, Washington State Convention Center*

This session is eligible for up to (2.0) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderators:** Jennelle Hodge, PHD, FACMG, Indiana University, IN; Devin Oglesbee, PhD, Mayo Clinic Biochemical Genetics Laboratory, MN

This session to be presented in the Diagnostic Dilemma format was created in an effort to increase the quantity and quality of Laboratory genetic educational opportunities. The session will be an interactive session which will allow genetics professionals to present cases of rare knowns and unknowns.

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**SCIENTIFIC CONCURRENT SESSIONS**

4:00 pm – 5:30 pm

**Innovative Uses of Technology for Practicing Genetic Clinicians and Counselors**
*Location: Room 606-609 – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderator:** Brian Skotko, MD, MPP, FACMG, Massachusetts General Hospital, MA

**Presentations:**

- **4:00 pm – 4:20 pm**
  **Down Syndrome Clinic To You (DSC2U): An Online Automated Clinic for Patients and Families**
  *Brian Skotko, MD, MPP, FACMG, Massachusetts General Hospital*

- **4:20 pm – 4:40 pm**
  **Telemedicine to Improve Efficiency and Increase Access to Genetic Counseling Services**
  *Katie Stoll, MS, LGC, Genetic Support Foundation*

- **4:40 pm – 5:00 pm**
  **Exceptional Lives: Unique Online Disability Platform Uses Innovative Technology to Help Families Connect to Services and Benefits**
  *Julie McKinney, MS, Exceptional Lives, Inc.*

- **5:00 pm – 5:20 pm**
  **Electronic Health Record Integration: Using Technology to Improve Patient Care**
  *Stephanie Santoro, MD, Massachusetts General Hospital*

- **5:20 pm – 5:30 pm**
  **Panel and Q&A**
ORAL PLATFORM PRESENTATIONS

4:00 pm – 5:30 pm

Biochemical/Metabolic Genetics

Location: Ballroom 6C – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E./NSGC Contact Hours

Moderator: Sarah Elsea, PhD, FACMG, Baylor College of Medicine, TX

Abstracts:

4:00 pm – 4:15 pm
#29 CLN3-Batten Natural History: Deep Phenotyping of a Lysosomal Storage Condition
An N. Dang Do, MD, PhD, Eunice Kennedy Shriver National Institute of Child Health and Human Development, NIH

4:15 pm – 4:30 pm
#30 A Characterization of Non-Immune Hydrops Fetalis in Lysosomal Storage Diseases
Stephanie L. Kass, BS, Sidney Kimmel Medical College at Thomas Jefferson University

4:30 pm – 4:45 pm
#31 Phase 3 PRISM Studies: Update of Efficacy and Safety of Pegvaliase for the Treatment of Adults with Phenylketonuria
Cary Harding, MD, Oregon Health & Science University

4:45 pm – 5:00 pm
#32 Proactive missense effect maps for MTHFR and CBS accurately predict homocystinuria phenotypes and therapeutic response
Frederick Roth, PhD, University of Toronto

5:00 pm – 5:15 pm
#33 Broad-Scale Untargeted Metabolomic Profiling as an Initial Approach to Clinical Diagnosis of Inborn Errors of Metabolism
Ning Liu, PhD, Baylor College of Medicine

5:15 pm – 5:30 pm
#34 GOT2 Deficiency: Expanding the Number of Treatable Metabolic Epilepsies with a Malate-Aspartate Shuttle Related Encephalopathy
Clara van Karnebeek, MD, PhD, Amsterdam University Medical Centre
4:00 pm – 5:30 pm

**Prenatal Genetics**

*Location: Ballroom 6B – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderator: Myra Wick, MD, PhD, FACMG, Mayo Clinic, MN**

**Abstracts:**

4:00 pm – 4:15 pm

**#35 What Is the Rate of Aneuploidy in Twin Reversed Arterial Perfusion Sequence?**

*Jessica Smith, MD, University of Michigan*

4:15 pm – 4:30 pm

**#36 Single Fetal Cell Testing for Prenatal Diagnosis: Two Pilot Validation Studies**

*Liesbeth Vossaert, PhD, Baylor College of Medicine*

4:30 pm – 4:45 pm

**#37 Clinical Utility of postmortem Whole Exome Sequencing in fetal cases of multiple congenital anomalies**

*Erica Schindewolf, MSc, LCGC, The Children’s Hospital of Philadelphia*

4:45 pm – 5:00 pm

**#38 Recombination Failure in Human Female Meiosis**

*Terry Hassold, PhD, FACMG, Washington State University*

5:00 pm – 5:15 pm

**#39 Systematic Evaluation of Yields from Whole-Genome Sequencing of 8,954 Individuals Compared to Conventional Technologies for Prenatal and Pediatric Diagnostics**

*Michael Talkowski, PhD, Massachusetts General Hospital / Harvard Medical School / Broad Institute*

5:15 pm – 5:30 pm

**#40 Cell-Free DNA-Based Testing Can Be Used to Detect Aneuploidy in Early Pregnancy Loss**

*Antoni Borrell, MD, PhD, BCNatal – Hospital Clinic Barcelona*
4:00 pm – 5:30 pm

**Cancer Genetics and Therapeutics**

*Location: Ballroom 6E – 6th floor, Washington State Convention Center*

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

**Moderators:** Jennelle Hodge, PhD, FACMG, Indiana University School of Medicine, IN; Tuya Pal, MD, FACMG, Vanderbilt University Medical Center, TN

**Abstracts:**

4:00 pm – 4:15 pm

#41 Analysis of Breast Cancer Surveillance Guidelines for Moderate Penetrance Breast Cancer Genes  
*Elise Watson, MS, CGC, UT Southwestern*

4:15 pm – 4:30 pm

#42 APC Variant Identification in an Unselected Patient Population: Where are the Polyps?  
*Heather Rocha, MS, LGC, Geisinger*

4:30 pm – 4:45 pm

#43 Germline Variants Associated with Cancer Predisposition Identified in a Large Cohort of Pediatric Cancer Patients through Somatic Tumor Sequencing  
*Suzanne MacFarland, MD, Children’s Hospital of Philadelphia*

4:45 pm – 5:00 pm

#44 Myeloid Malignancy Variant Curation Expert Panel: an ASH-sponsored ClinGen Expert Panel to optimize and validate ACMG/AMP variant interpretation guidelines for genes associated with inherited myeloid neoplasms  
*Xi Luo, PhD, Baylor College of Medicine and Texas Children’s Hospital*

5:00 pm – 5:15 pm

#45 Comprehensive Multigene Panels in Cancer Patients: Increased Diagnostic Yield and Clinical Utility  
*Shan Yang, PhD, Invitae*

5:15 pm – 5:30 pm

#46 Genetic Predisposition in Pediatric Rhabdomyosarcoma: When to Worry?  
*Kristin Zelley, MS, CGC, Children’s Hospital of Philadelphia*
Public Health, Health Services and Implementation

Location: Room 611-614 – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderators: Pilar Magoulas, MS, Baylor College of Medicine, TX

Abstracts:

4:00 pm – 4:15 pm
#47 Initial Outcomes for a Genetic Health Assessment Tool Implemented Across a Health System Primary Care Network
Peter Hulick, MD, MMSc, NorthShore University HealthSystem

4:15 pm – 4:30 pm
#48 Children with Genetic Conditions in the United States: Estimates from the 2016 and 2017 National Survey of Children’s Health
Catharine Riley, PhD, MPH, Health Resources and Services Administration

4:30 pm – 4:45 pm
#49 Implementing digital systems to facilitate ascertainment and referral for hereditary cancer syndromes: Variation in genetic counseling and testing uptake
Catharine Wang, PhD, Boston University

4:45 pm – 5:00 pm
#50 Newborn Screening for X-linked Adrenoleukodystrophy in Minnesota: Novel Challenges and Observed Incidence
Katie Wiens, MS, LG, University of Minnesota Health-Fairview

5:00 pm – 5:15 pm
#51 Electronic Health Records Identify Potential Undiagnosed Cases of Hypophosphatasia
Bimal Chaudhari, MD, MPH, FAAP, Nationwide Children’s Hospital

5:15 pm – 5:30 pm
#52 After the Pilot: The Commencement of Universal Newborn Screening for Spinal Muscular Atrophy in New York State
Michele Caggana, ScD, FACMG, NYSDOH/Wadsworth Center
WORKSHOPS

7:00 am – 8:00 am

Informational Session: Laboratory Genetics and Genomics Alternative Pathway Training Program
Location: Room 602-604 – 6th floor, Washington State Convention Center

7:15 am – 8:30 am

ClinGen Expert Panel Information Workshop (pre-registration required)
Location: Room 3AB – 3rd floor, Washington State Convention Center

6:00 pm – 7:30 pm

ACGME Accreditation Transition of Clinical Biochemical Genetics and Laboratory Genetics and Genomics Training Programs
Location: Room 611-614 – 6th floor, Washington State Convention Center

6:00 pm – 7:30 pm

Pediatric Diagnostic Dilemmas (Rare Knowns and Unknowns)
Location: Ballroom 6E – 6th floor, Washington State Convention Center
Daily Schedule – Educational Sessions, Events and Meetings

Indicates this session will be live streamed during the meeting.

Saturday, April 6

SCIENTIFIC CONCURRENT SESSIONS

8:00 am – 9:30 am

- **Adult & Cancer Diagnostic Challenges**
  
  *Location: Ballroom 6C – 6th floor, Washington State Convention Center*
  
  This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours
  
  **Moderators:** Shweta Dhar, MD, MS, FACMG, Baylor College of Medicine TX; Fuki Hisama, MD, FACMG, FAAN, University of Washington School of Medicine, WA
  
  This session to be presented in the Diagnostic Dilemma format was created in an effort to increase the quantity and quality of Adult and Cancer genetic educational opportunities. The session will be an interactive session which will allow genetics professionals to present cases of rare knowns and unknowns.

8:00 am – 9:30 am

- **Phenotypes of “Single” Gene Disorders Are Complex Traits: Lessons from Inherited Metabolic Disorders for Personalized and Precision Medicine**
  
  *Location: Ballroom 6E – 6th floor, Washington State Convention Center*
  
  This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours
  
  **Moderators:** Katrina Dipple, MD, PhD, FACMG, University of Washington, Seattle Children’s Hospital, WA; Edward McCabe, MD, PhD, FACMG, University of California Los Angeles, CA
  
  **Presentations:**
  
  8:00 am – 8:30 am
  
  Lessons for Complexity from Rare Genetic Disorders: Thresholds, Modifiers, Network Architecture, and “Moonlighting” Functions of Proteins
  
  *Katrina Dipple, MD, PhD, FACMG, University of Washington, Seattle Children’s Hospital*
  
  8:30 am – 9:00 am
  
  Metabolite Flux: A Dynamic Concept for Inherited Metabolic Disorders as Complex Traits
  
  *Edward McCabe, MD, PhD, FACMG, University of California Los Angeles*
  
  9:00 am – 9:30 am
  
  A Next Generation, Multi-omic View of Inborn Errors of Metabolism
  
  *Carmen Argmann, PhD, Icahn School of Medicine at Mount Sinai*
8:00 am – 9:30 am

Using Telegenetics to Improve Access to Clinicians and Researchers for Families

Location: Ballroom 6B – 6th floor, Washington State Convention Center

This session is eligible for up to (1.5) CME/CEU/P.A.C.E.®/NSGC Contact Hours

Moderator: David Flannery, MD, FAAP, FACMG, Cleveland Clinic

Presentations:

8:00 am – 8:05 am
Introduction of Telegenetics
Sylvia Mann, MS, CGC, Hawaii State Department of Health

8:05 am – 8:15 am
What You Didn’t Learn in School: Telegenetics Training Opportunities for Genetics Providers
Sylvia Mann, MS, CGC, Hawaii State Department of Health

8:15 am – 8:30 am
Improving Access to Telegenetics for Medically Underserved Populations
Janet Thomas, MD, FACMG, University of Colorado School of Medicine

8:30 am – 8:40 am
Requesting Technical Assistance for Telegenetics
Joann Bodurtha, MD, MPH, FACMG, FAAP, McKusick-Nathans IGM, Johns Hopkins University

8:40 am – 8:55 am
A Genetics Provider’s Experience Using Telegenetics in Multiple States
Gerald Schaefer, MD, FACMG, University of Arkansas for Medical Sciences

8:55 am – 9:05 am
A Supportive Environment of Policies to Support Telegenetics
David Flannery, MD, FAAP, FACMG, Cleveland Clinic

9:05 am – 9:30 am
Panel Discussion and Q&A
SCIENTIFIC PLENARY SESSION

9:45 am – 10:45 am

- **Hot Topics: Emerging Fetal Stem Cell and Molecular Therapies**
  
  *Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center*

  This session is eligible for up to 1.0 CME/CEU/P.A.C.E./NSGC Contact Hours

  **Moderator:** Pilar Magoulas, MS, Baylor College of Medicine, TX

  **Presentations:**
  
  - 9:45 am – 10:15 am
    *Prenatal Correction of X-linked Hypohidrotic Ectodermal Dysplasia*
    
    Holm Schneider, MD, University Hospital Erlangen
  
  - 10:15 am – 10:45 am
    *Emerging Fetal Stem Cell and Molecular Therapies*
    
    Tippi MacKenzie, MD, University of California San Francisco

11:00 am – 12:30 pm

- **ACMG/SIMD JOINT PLENARY SESSION (Part 1)**

  “Thinking Metabolic” for the Genetics Clinician: You Took the Call from the Department of Health: What Now?

  *Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center*

  This session is eligible for up to 1.5 CME/CEU/P.A.C.E./NSGC Contact Hours

  **Moderators:** Susan Berry, MD, FACMG, FAAP, University of Minnesota, MN; Fernando Scaglia, MD, PhD, FACMG, Baylor College of Medicine, Texas Children’s Hospital, TX and Prince of Wales Hospital, Hong Kong SAR

  **Presentations:**
  
  - 11:00 am – 11:30 am
    *You got the NBS call for presumptive positive Pompe. What now?*
    
    Priya Kishnani, MD, MBBS, Duke University Medical Center
  
  - 11:30 am – 12:00 pm
    *You got the NBS call for presumptive positive XALD. What now?*
    
    Gerald Raymond, MD, FACMG, Penn State Medical Center
  
  - 12:00 pm – 12:30 pm
    *You got the NBS call for presumptive positive MPS1. What now?*
    
    Melissa Wasserstein, MD, Albert Einstein College of Medicine and the Children’s Hospital at Montefiore
1:30 pm – 3:30 pm

ACMG/SIMD JOINT PLENARY SESSION (Part 2)

“Thinking Metabolic” for the Genetics Clinician: Could This Really be Metabolic? Dysmorphic Features of IEM

Location: Exhibit Hall 4E – 4th floor, Washington State Convention Center

This session is eligible for up to (2.0) CME/CEU/P.A.C.E./NSGC Contact Hours

Moderators: Gerard Berry, MD, FACMG, Boston Children’s Hospital, Harvard University, MA; Amarilis Sanchez-Valle, MD, University of South Florida, FL

Presentations:

1:30 pm – 2:00 pm
Dysmorphism in Sterol Disorders
Robert Steiner, MD, FACMG, University of Wisconsin School of Medicine and Public Health

2:00 pm – 2:30 pm
Dysmorphism in Congenital Disorders of Glycosylation
Eva Morava-Kozicz, MD, PhD, Mayo School of Medicine

2:30 pm – 3:00 pm
Dysmorphism in Mitochondrial Disorders
Rebecca Ganetzky, MD, Children’s Hospital of Philadelphia

3:00 pm – 3:30 pm
Dysmorphism in Disorders of Peroxisome Biogenesis
Nancy Braverman, MD, MSc, FACMG, Research Institute-McGill University Health Center
Abstract Information

In accordance with ACMG’s green-meeting initiatives, abstracts are available for online viewing and print on demand only. Abstracts are indexed by keyword and author, and they may be viewed online at www.acmgmeeting.net or via the ACMG mobile app.

FEATURED PLATFORM PRESENTATIONS: Washington State Convention Center – Hall 4E

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<tr>
<td>THURSDAY, APRIL 4</td>
<td>8:00 AM – 9:00 AM</td>
<td></td>
</tr>
<tr>
<td>25</td>
<td>Healthy Nevada Project Population Health Genetic Screening in Northern Nevada: At Risk Individuals are Missed Using Current Guidelines</td>
<td>Joseph Grzymski, PhD Renown Health</td>
</tr>
<tr>
<td>26</td>
<td>All That Glitters Is Not Gold: Circulating TP53 Mutations and Li-Fraumeni Syndrome (LFS)</td>
<td>Alison Robertson, LGC Dana Farber Cancer Institute</td>
</tr>
<tr>
<td>27</td>
<td>Limitations of HBOC Direct-To-Consumer Genetic Screening: False Positives, False Negatives and Everything in Between</td>
<td>Edward Esplin, MD, PhD, FACMG, FACP Invitae</td>
</tr>
<tr>
<td>28</td>
<td>ClinPhen Extracts and Prioritizes Patient Phenotypes Directly from Medical Records to Expedite Genetic Disease Diagnosis</td>
<td>Johannes Birgmeier, MSc Stanford University</td>
</tr>
</tbody>
</table>

PLATFORM PRESENTATIONS: (Abstracts 1-24 and 29-52)

<table>
<thead>
<tr>
<th>PLATFORM PRESENTATION TOPICS</th>
<th>ABSTRACT NUMBERS</th>
<th>WSCC ROOM</th>
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</thead>
<tbody>
<tr>
<td>WEDNESDAY, APRIL 3</td>
<td>4:00 PM – 5:30 PM</td>
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</tr>
<tr>
<td>Genetic Counseling</td>
<td>1–6</td>
<td>6B</td>
</tr>
<tr>
<td>Clinical Genetics and Therapeutics</td>
<td>7–12</td>
<td>6E</td>
</tr>
<tr>
<td>Laboratory Genetics and Genomics</td>
<td>13–18</td>
<td>6C</td>
</tr>
<tr>
<td>Legal and Ethical Issues, Education</td>
<td>19–24</td>
<td>6A</td>
</tr>
<tr>
<td>FRIDAY, APRIL 5</td>
<td>4:00 PM – 5:30 PM</td>
<td></td>
</tr>
<tr>
<td>Biochemical/Metabolic Genetics</td>
<td>29–34</td>
<td>6C</td>
</tr>
<tr>
<td>Prenatal Genetics</td>
<td>35–40</td>
<td>6B</td>
</tr>
<tr>
<td>Cancer Genetics and Therapeutics</td>
<td>41–46</td>
<td>6E</td>
</tr>
<tr>
<td>Public Health, Health Services and Implementation</td>
<td>47–52</td>
<td>611-614</td>
</tr>
</tbody>
</table>

Taking photographs or recording posters is strictly prohibited.
INDUSTRY PLATFORM PRESENTATIONS*

<table>
<thead>
<tr>
<th>PRESENTATION TITLE</th>
<th>ABSTRACT NUMBERS</th>
<th>TIME</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive Yield of Genetic Testing for Patients with Epilepsy is Associated with Age of Seizure-Onset, Presence of Neurological Features, and Trio Testing</td>
<td>227</td>
<td>12:15 pm – 12:30 pm</td>
</tr>
<tr>
<td>Characteristics and Clinical Correlations of Somatic Mosaic Large Rearrangements Identified by Pan Cancer NGS Panel Test</td>
<td>166</td>
<td>12:30 pm – 12:45 pm</td>
</tr>
<tr>
<td>Use of a Chatbot to Offer Pre-test Education for Expanded Carrier Screening</td>
<td>611</td>
<td>12:45 pm – 1:00 pm</td>
</tr>
<tr>
<td>Three-quarters of Upgraded Negative Reports Due to Reclassification are Due to Lab-initiated Proactive Reanalysis</td>
<td>659</td>
<td>1:00 pm – 1:15 pm</td>
</tr>
</tbody>
</table>

Taking photographs or recording posters is strictly prohibited.
Presenter/Poster Information

For poster topic locations, see the fold out map at the front of this guide.

894 Poster Presentations (954 Abstracts)
Odd Numbered Posters: Thursday  •  10:00 am – 11:30 am
Even Numbered Posters: Friday  •  10:30 am – 12:00 pm

<table>
<thead>
<tr>
<th>POSTER TOPICS</th>
<th>ABSTRACTS NUMBERS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical genetics and therapeutics</td>
<td>7 - 12</td>
</tr>
<tr>
<td></td>
<td>227 - 610</td>
</tr>
<tr>
<td>Cancer genetics and therapeutics</td>
<td>41 - 46</td>
</tr>
<tr>
<td></td>
<td>167 - 226</td>
</tr>
<tr>
<td>Prenatal genetics</td>
<td>35 - 40</td>
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<tr>
<td></td>
<td>873 - 946</td>
</tr>
<tr>
<td>Biochemical and metabolic genetics</td>
<td>29 - 34</td>
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<tr>
<td></td>
<td>100 - 165</td>
</tr>
<tr>
<td>Genetic counseling</td>
<td>1 - 6</td>
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<tr>
<td></td>
<td>612 - 658</td>
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<tr>
<td>Public health, health services and implementation</td>
<td>47 - 52</td>
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<tr>
<td></td>
<td>947 - 1009</td>
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<tr>
<td>Legal and ethical issues; education</td>
<td>19 - 24</td>
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<tr>
<td></td>
<td>842 - 872</td>
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<tr>
<td>Laboratory genetics and genomics</td>
<td>13 - 18</td>
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<td>659 - 841</td>
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</tbody>
</table>

Taking photographs or recording posters is strictly prohibited.
# Learning Lounge Schedule

## Thursday, April 4

<table>
<thead>
<tr>
<th>LOUNGE</th>
<th>START/END TIME</th>
<th>TITLE/PRESENTER(S)</th>
</tr>
</thead>
</table>
| 1      | 10:30 am – 11:00 am | Producing Powerful Podcasts  
Presenter: Tess Vismale, CMP |
| 2      | 10:30 am – 11:00 am | Developing a Stable Clinical Version of HGVS Nomenclature  
Presenter: Ryan Schmidt, MD, PhD, Children’s Hospital, Los Angeles |
|        | 11:15 am – 11:45 am | Policy Engagement with NCC  
Presenters: Susanna Ginsburg, MSW; Alisha Keehn, MPA, ACMG; Megan Lyon, MPH, ACMG |
| 1      | 11:15 am – 11:45 am | Alpha Thalassemia Major - Not a Lethal Diagnosis  
Presenter: Billie Liangolou, CGC, UCSF Fetal Treatment Center |
|        | 12:00 pm – 12:30 pm | Highlights from Skeletal Dysplasia Through the Human Lifespan  
Presenters: Keith Eddleman, MD, FACMG, Icahn SOM at Mount Sinai; Jennifer King, MD, Albert Einstein College of Medicine |
| 2      | 12:00 pm – 12:30 pm | Steps to Building a Stellar Personal and Professional Brand  
Presenter: Tess Vismale, CMP |
|        | 12:45 pm – 1:15 pm | Rapid Fire Poster Presentation  
The top 20 posters will be presented in “Rapid Fire” format  
Moderator: Marc Williams, MD, FACMG, Geisinger |
|        | 12:45 pm – 1:15 pm | Collaboratively Transforming Access to Genetic Testing  
Presenters: Adam Buchanan, MS, MPH, LGC, Geisinger; Miranda Hallquist, MSc, LGC, Geisinger; Kelly Ormond, MS, LCGC, Stanford |
### Friday, April 5

<table>
<thead>
<tr>
<th>LOUNGE</th>
<th>START END TIME</th>
<th>TITLE/PRESENTER(S)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>10:30 am – 11:00 am</td>
<td><strong>AI in Healthcare: Navigating the Hype from the Hope</strong>&lt;br&gt;Presenter: <em>Dekel Gelbman, FDNA</em></td>
</tr>
<tr>
<td>2</td>
<td>10:30 am – 11:00 am</td>
<td><strong>Top 10 Must-have Apps for Medical Professionals</strong>&lt;br&gt;Presenter: <em>Tess Vismale, CMP</em></td>
</tr>
<tr>
<td>1</td>
<td>11:15 am – 11:45 am</td>
<td><strong>After Chat with the Pruzansky Lecturer</strong>&lt;br&gt;Presenter: <em>Max Muenke, MD, FACMG, NGHRI, NIH</em></td>
</tr>
<tr>
<td>2</td>
<td>11:15 am – 11:45 am</td>
<td><strong>Ethical Considerations in Newborn Screening Research</strong>&lt;br&gt;Presenter: <em>Amy Brower, PhD, NBSTRN, ACMG</em></td>
</tr>
<tr>
<td>1</td>
<td>12:00 pm – 12:30 pm</td>
<td><strong>Top Tips for Creating Eye-popping Brand Content</strong>&lt;br&gt;Presenter: <em>Tess Vismale, CMP</em></td>
</tr>
<tr>
<td>2</td>
<td>12:00 pm – 12:30 pm</td>
<td><strong>Presentation and Book Signing, The Gene Machine, by Bonnie Rochman</strong></td>
</tr>
</tbody>
</table>
# Exhibitor Theater Schedule

## Thursday, April 4

<table>
<thead>
<tr>
<th>Start/End Time</th>
<th>Theater</th>
<th>Company</th>
<th>Title/Speaker</th>
</tr>
</thead>
<tbody>
<tr>
<td>10:30 am – 11:00 am</td>
<td>1</td>
<td><strong>Ambry Genetics</strong></td>
<td>RNA Genetic Testing Improves Variant Classification and Patient Management</td>
</tr>
<tr>
<td></td>
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<td></td>
<td>Speaker: Rachid Karam, MD, PhD, Ambry Genetics</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td><strong>Oxford Nanopore Technologies, LTD</strong></td>
<td>Same Day-point of Care Aneuploidy Testing Using a Hand-held Nanopore Sequencer</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Speaker: Zev Williams, MD, PhD, Columbia Medical Center</td>
</tr>
<tr>
<td>11:05 am – 11:35 am</td>
<td>1</td>
<td><strong>SciGene</strong></td>
<td>Automating Slide Preparation and Processing in the Cytogenetics Laboratory</td>
</tr>
<tr>
<td></td>
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<td></td>
<td>Speaker: James Stanchfield, PhD, SciGene</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td><strong>Thermo Fisher Scientific</strong></td>
<td>Exon Level CNV Analysis in Fetuses with Structural Anomalies</td>
</tr>
<tr>
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<td></td>
<td>Speaker: Brynn Levy, MSc, PhD, New York Presbyterian Hospital, Columbia University Medical Center</td>
</tr>
<tr>
<td>11:40 am – 12:10 pm</td>
<td>1</td>
<td><strong>BioDiscovery, Inc.</strong></td>
<td>See What You’ve Been Missing in Your NGS Data With NxClinical</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Speaker: Soheil Shams, PhD, BioDiscovery, Inc.</td>
</tr>
<tr>
<td></td>
<td>2</td>
<td><strong>Prevention Genetics</strong></td>
<td>Value of Genome Sequencing</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Speaker: James Weber, PhD, PreventionGenetics</td>
</tr>
<tr>
<td>12:15 pm – 12:45 pm</td>
<td>1</td>
<td><strong>Roche Sequencing Solutions, Inc.</strong></td>
<td>Cell-Free DNA Prenatal Testing in 2019: What Really Matters</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Speaker: Liza Kunz, MD, Roche Sequencing Solutions</td>
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<tr>
<td></td>
<td>2</td>
<td><strong>Agilent</strong></td>
<td>Implementation of Genomic Variant Interpretation Through Vendor Collaboration In Genovic: The Melbourne Genomics Health Alliance Shared Clinical System for Genomics</td>
</tr>
<tr>
<td></td>
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<td></td>
<td>Speaker: Natalie Thorne, PhD, Melbourne Genomics Health Alliance</td>
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</tbody>
</table>
## Friday, April 5

<table>
<thead>
<tr>
<th>Theater</th>
<th>Company</th>
<th>Title/Speaker</th>
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</thead>
<tbody>
<tr>
<td></td>
<td><strong>10:45 am – 11:15 am</strong></td>
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<tr>
<td>1</td>
<td>Centogene</td>
<td>Genome First. Last. Fast: The One Step Approach to Genetic Testing</td>
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<tr>
<td></td>
<td></td>
<td>Speaker: Peter Bauer, MD, PhD, Centogene</td>
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<tr>
<td>2</td>
<td>Color</td>
<td>Leveraging NGS Technologies Beyond Monogenic Applications</td>
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<tr>
<td></td>
<td></td>
<td>Speaker: Alicia Y. Zhou, PhD, Color Genomics</td>
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<tr>
<td></td>
<td><strong>11:20 am – 11:50 am</strong></td>
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<tr>
<td>1</td>
<td>Integrated Genetics</td>
<td>When a Negative cfDNA isn’t the Whole Story: The Value of Genome-wide Analysis</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Speaker: Ron McCullough, PhD, MBA, Integrated Genetics</td>
</tr>
<tr>
<td>2</td>
<td>Baylor Genetics</td>
<td>Clinical Validation of Whole Genome Sequencing</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Speaker: Pengfei Liu, PhD, Baylor Genetics</td>
</tr>
<tr>
<td></td>
<td><strong>11:55 am – 12:25 pm</strong></td>
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</tr>
<tr>
<td>1</td>
<td>PerkinElmer Genomics</td>
<td>Unlocking the Power of Genome Sequencing</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Speakers: Madhuri Hegde, PhD, FACMG, PerkinElmer Genomics; Alka Chaubey, PhD, FACMG, PerkinElmer Genomics</td>
</tr>
<tr>
<td>2</td>
<td>Catalyst Pharmaceuticals</td>
<td>Congenital Myasthenic Syndromes: What You Need to Know as a Genetics Professional</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Speaker: Stanley Iyadurai, MD, PhD, FAAN, Ohio State University, Wexner Medical Center</td>
</tr>
<tr>
<td></td>
<td><strong>12:30 pm – 1:00 pm</strong></td>
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</tr>
<tr>
<td>1</td>
<td>MNG Laboratories</td>
<td>Genome Scale Genetic Testing</td>
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<td></td>
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<td>Speaker: Trey Langley, PhD, MNG Laboratories</td>
</tr>
<tr>
<td>2</td>
<td>Blueprint Genetics</td>
<td>Improved Diagnostic Yield in Rare Hereditary Disorders: Meeting Clinical Needs Using Optimized Sequencing Solutions for Copy-Number Variations and Difficult-to-Sequence Regions</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Speaker: Kim Gall, MCs, CGC, Blueprint Genetics</td>
</tr>
</tbody>
</table>
CELEBRATE 60 YEARS AND COUNTING

The McKusick Short Course remains the most influential annual training event in medical genetics. Each July, more than 60 scientists and 100 trainees gather in beautiful Bar Harbor, Maine to learn the latest genetics and genomics in experimental animals and humans.

In 2019 the course will include a special symposium: The Future of Human and Experimental Genetics on Saturday, July 20. Leaders in genetics and genomics will describe a vision for the field well into the 21st Century.

SCHOLARSHIPS

As many as 40 scholarships of $1,900 are available through generous support from the Howard Hughes Medical Institute and the NIH. Trainees and junior faculty who are underrepresented in the biomedical sciences are encouraged to apply.

LEARN MORE

For more information visit www.jax.org/bhshortcourse.

WHERE & WHEN

The Jackson Laboratory
Bar Harbor, Maine
July 15-26, 2019

REGISTRATION

$2,100 Standard Registration
$1,900 Trainee Registration
Scholarships Available

CONTACT

Erin McDevitt
erin.mcdevitt@jax.org
207-288-6659
GIVE YOUR ADULT PATIENTS WITH UNCONTROLLED PKU

A difference they can see in Phe

1450 µmol/L
mean blood Phe level at pretreatment baseline
n=29

597 µmol/L
at RWP baseline
n=29

553 µmol/L
at RWP week 8
n=26

897 µmol/L (15 mg/dL; 62%) reduction in blood Phe levels for patients in clinical trials receiving PALYNZIQ 20 mg subcutaneously once daily**

Learn more at BioMarin Booth 501

*Patients who were randomized to receive PALYNZIQ® (pegvaliase-pqpz) Injection 20 mg subcutaneously once daily and who achieved a ≥20% reduction in blood Phe levels from pretreatment baseline had a 62% reduction in mean blood Phe levels from pretreatment baseline to randomized withdrawal period (RWP) week 8.1

Indication: PALYNZIQ® (pegvaliase-pqpz) Injection is a phenylalanine-metabolizing enzyme indicated to reduce blood phenylalanine concentrations in adult patients with phenylketonuria (PKU) who have uncontrolled blood phenylalanine concentrations greater than 600 µmol/L on existing management.

Important Safety Information

BOXED WARNING: RISK OF ANAPHYLAXIS

• Anaphylaxis has been reported after administration of PALYNZIQ and may occur at any time during treatment with PALYNZIQ

• Administer the initial dose of PALYNZIQ under the supervision of a healthcare provider equipped to manage anaphylaxis, and closely observe patients for at least 60 minutes following injection. Prior to self-injection, confirm patient competency with self-administration, and patient’s and observer’s (if applicable) ability to recognize signs and symptoms of anaphylaxis and to administer auto-injectable epinephrine, if needed

• Prescribe auto-injectable epinephrine to all patients treated with PALYNZIQ. Prior to the first dose, instruct the patient and observer (if applicable) on its appropriate use. Instruct the patient to seek immediate medical care upon its use. Instruct patients to carry auto-injectable epinephrine with them at all times during treatment with PALYNZIQ

• PALYNZIQ is available only through a restricted program under a Risk Evaluation and Mitigation Strategy (REMS) called the PALYNZIQ REMS. Further information, including a list of qualified pharmacies, is available at www.PALYNZIQREMS.com or by telephone at 1-855-758-REMS (1-855-758-7367)

Please see additional Important Safety Information on the next page in this program guide and full prescribing information at the BioMarin Booth.
IMPORTANT SAFETY INFORMATION

Indication
PALYNZIQ™ [pegvaliase-pqpz] Injection is a phenylalanine-metabolizing enzyme indicated to reduce blood phenylalanine concentrations in adults with phenylketonuria (PKU) who have uncontrolled blood phenylalanine concentrations greater than 600 μmol/L, on an ongoing management.

Important Safety Information

BOXED WARNING: RISK OF ANAPHYLAXIS
• Anaphylaxis has been reported after administration of PALYNZIQ and may occur at any time during treatment with PALYNZIQ.
• Administer the initial dose of PALYNZIQ under the supervision of a healthcare provider equipped to manage anaphylaxis, and closely observe patients for at least 60 minutes following injection. Prior to self-injection, confirm patient competency with self-administration, and patient’s and observer’s (if applicable) ability to recognize signs and symptoms of anaphylaxis and to administer auto-injectable epinephrine, if needed
• Prescribe auto-injectable epinephrine to all patients treated with PALYNZIQ. Prior to the first dose, instruct the patient and observer (if applicable) on its appropriate use. Instruct the patient to seek immediate medical care upon its use. Instruct patients to carry auto-injectable epinephrine with them at all times during treatment with PALYNZIQ.
• PALYNZIQ is available only through a restricted program under a Risk Evaluation and Mitigation Strategy (REMS) called the PALYNZIQ REMS. Further information, including a list of qualified pharmacies, is available at www.PALYNZIQREMS.com or by telephone at 1-855-758-REMS (1-855-758-7367)

WARNINGS AND PRECAUTIONS
Anaphylaxis
• Signs and symptoms of anaphylaxis reported include syncope, hypotension, hypoxia, dyspnea, wheezing, chest discomfort/chest tightness, tachycardia, angioedema (swelling of face, lips, eyes, tongue), throat tightness, skin flushing, rash, urticaria, pruritus, and gastrointestinal symptoms (vomiting, nausea, diarrhea)
• Anaphylaxis generally occurred within 1 hour after injection; however, delayed episodes occurred up to 48 hours after PALYNZIQ administration
• Consider having an adult observer for patients who may need assistance in recognizing and managing anaphylaxis during treatment with PALYNZIQ. If an adult observer is needed, the observer should be present during and for at least 60 minutes after administration of PALYNZIQ, and should be able to administer auto-injectable epinephrine and call for emergency medical support upon its use
• Anaphylaxis requires immediate treatment with auto-injectable epinephrine. Prescribe auto-injectable epinephrine to all patients receiving PALYNZIQ and instruct patients to carry auto-injectable epinephrine with them at all times during treatment with PALYNZIQ. Prior to the first dose, instruct the patient and observer (if applicable) on how to recognize the signs and symptoms of anaphylaxis, on how to properly administer auto-injectable epinephrine, and to seek immediate medical care upon its use. Consider the risks associated with auto-injectable epinephrine use when prescribing PALYNZIQ. Refer to the auto-injectable epinephrine Prescribing Information for complete information
• Consider the risks and benefits of readministering PALYNZIQ following an episode of anaphylaxis. If the decision is made to readminister PALYNZIQ, administer the first dose under the supervision of a healthcare provider equipped to manage anaphylaxis and closely observe the patient for at least 60 minutes following the dose. Subsequent dose titration of PALYNZIQ should be based on patient tolerability and therapeutic response
• Consider premedication with an H1-receptor antagonist, H2-receptor antagonist, and/or antipyretic prior to administration of PALYNZIQ based upon individual patient tolerability

Other Hypersensitivity Reactions
• Hypersensitivity reactions other than anaphylaxis have been reported in 196 of 285 (69%) patients treated with PALYNZIQ
• Consider premedication with an H1-receptor antagonist, H2-receptor antagonist, and/or antipyretic prior to PALYNZIQ administration based upon individual patient tolerability

ADVERSE REACTIONS
• The most common adverse reactions (at least 20% of patients in either treatment phase) were injection site reactions, arthralgia, hypersensitivity reactions, headache, generalized skin reaction lasting at least 14 days, pruritus, nausea, abdominal pain, oropharyngeal pain, vomiting, cough, diarrhea, and fatigue
• Of the 285 patients exposed to PALYNZIQ in an induction/titration/maintenance regimen in clinical trials, 31 (11%) patients discontinued treatment due to adverse reactions. The most common adverse reactions leading to treatment discontinuation were hypersensitivity reactions (6% of patients)—including anaphylaxis (3% of patients) and angioedema (1% of patients)—arthralgia (4% of patients), generalized skin reactions lasting at least 14 days (2% of patients), and injection site reactions (1% of patients)
• The most common adverse reactions leading to dosage reduction were arthralgia (14% of patients), hypersensitivity reactions (9% of patients), injection site reactions (4% of patients), alopecia (3% of patients), and generalized skin reactions lasting at least 14 days (2% of patients)
• The most common adverse reactions leading to temporary drug interruption were arthralgia (13% of patients), hypersensitivity reactions (13% of patients), anaphylaxis (4% of patients), and injection site reactions (4% of patients)

Blood Phenylalanine Monitoring and Diet
• Obtain blood phenylalanine concentrations every 4 weeks until a maintenance dosage is established
• After a maintenance dosage is established, periodically monitor blood phenylalanine concentrations
• Counsel patients to monitor dietary protein and phenylalanine intake, and adjust as directed by their healthcare provider

DRUG INTERACTIONS
Effect of PALYNZIQ on Other PEGylated Products
• In a single-dose study of PALYNZIQ in adult patients with PKU, 2 patients receiving concomitant injections of medroxyprogesterone acetate suspension (a formulation containing PEG 3350) experienced hypersensitivity reactions, and 1 of the 2 patients also experienced anaphylaxis
• The clinical effects of concomitant treatment with different PEGylated products is unknown. Monitor patients treated with PALYNZIQ and concomitantly with other PEGylated products for hypersensitivity reactions

USE IN SPECIFIC POPULATIONS
Pregnancy and Lactation
• PALYNZIQ may cause fetal harm when administered to a pregnant woman
• If PALYNZIQ is administered during pregnancy, or if a patient becomes pregnant while receiving PALYNZIQ or within 1 month following the last dose of PALYNZIQ, healthcare providers should report PALYNZIQ exposure by calling 1-866-906-6100
• Monitor blood phenylalanine concentrations in breastfeeding women treated with PALYNZIQ

Pediatric Use
• The safety and efficacy of PALYNZIQ in pediatric patients have not been established

Geriatric Use
• Clinical studies of PALYNZIQ did not include patients aged 65 years and older
You are encouraged to report suspected adverse events to BioMarin at 1-866-906-6100 and the FDA at 1-800-FDA-1088 or www.fda.gov/medwatch. Please see enclosed full Prescribing Information, including Boxed Warning, or at PALYNZIQ.com/hcp.
WE ARE BREAKING BARRIERS

AveXis is relentlessly focused on bringing gene therapy out of the lab and into the clinical setting.

We are a clinical-stage gene therapy company dedicated to developing novel treatments for patients suffering from rare and life-threatening neurological diseases such as spinal muscular atrophy (SMA).

For more information about AveXis, gene therapy, or SMA, visit avexis.com.
# Exhibits Directory

The following list includes exhibitors as of February 22, 2019. Additional exhibitors may be listed in the Program Guide Addendum. For detailed company descriptions and product categories, please refer to the ACMG Mobile App and the online Exhibit List at [www.acmgmeeting.net](http://www.acmgmeeting.net).

<table>
<thead>
<tr>
<th>Exhibitor Name</th>
<th>Address</th>
<th>Phone</th>
<th>Website</th>
<th>Booth#</th>
</tr>
</thead>
<tbody>
<tr>
<td>23andMe</td>
<td>899 W. Evelyn Avenue, Mountain View, CA 94041</td>
<td>301-718-9603</td>
<td><a href="http://www.23andme.com">www.23andme.com</a></td>
<td>920</td>
</tr>
<tr>
<td>American College of Medical Genetics and Genomics (ACMG)</td>
<td>7101 Wisconsin Avenue, Suite 1100, Bethesda, MD 20814</td>
<td>301-718-9603</td>
<td><a href="http://www.acmg.net">www.acmg.net</a></td>
<td>300</td>
</tr>
<tr>
<td>Admera Health</td>
<td>126 Corporate Blvd., South Plainfield, NJ 07080</td>
<td>908-222-0533</td>
<td><a href="http://www.admerahealth.com">www.admerahealth.com</a></td>
<td>1108</td>
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<tr>
<td>ADS BIOTEC</td>
<td>7409 Irvington Road, Omaha, NE 68122</td>
<td>402-800-3200</td>
<td><a href="http://www.adsbiotec.com">www.adsbiotec.com</a></td>
<td>734</td>
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<tr>
<td>Agilent Technologies</td>
<td>2850 Centerville Road, Wilmington, DE 19808</td>
<td>302-636-1950</td>
<td><a href="http://www.genomics.agilent.com">www.genomics.agilent.com</a></td>
<td>629</td>
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<tr>
<td>Alexion Pharmaceuticals</td>
<td>121 Seaport Blvd., Boston, MA 02210</td>
<td>617-335-4818</td>
<td><a href="http://www.alexion.com">www.alexion.com</a></td>
<td>1200 and 1202</td>
</tr>
<tr>
<td>Allele Diagnostics</td>
<td>120 N Pine Street, Suite 152, Spokane, WA 99202</td>
<td>844-255-3532</td>
<td><a href="http://www.allelediagnostics.com">www.allelediagnostics.com</a></td>
<td>717</td>
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<tr>
<td>Alnylam Pharmaceuticals</td>
<td>300 Third Street, #3, Cambridge, MA 02142</td>
<td>978-340-4850</td>
<td><a href="http://www.alnylam.com">www.alnylam.com</a></td>
<td>1109</td>
</tr>
</tbody>
</table>
Ambry Genetics, A Konica Minolta Company
15 Argonaut
Aliso Viejo, CA 92656
Phone: 949-900-5500
Toll Free: 866-262-7943; Fax: 949-900-5501
www.ambrygen.com
Booth# 525

American Board of Genetic Counseling (ABGC)
4400 College Blvd., Suite 220
Overland Park, Kansas 66211
Phone: 913-222-8661
Booth# 305

American Board of Medical Genetics and Genomics
6120 Executive Blvd., Suite 525
Rockville, MD 20852
Phone: 301-634-7315; Fax: 301-634-7320
www.abmgg.org
Booth# 304

American Porphyria Foundation
4915 St. Elmo Avenue, Suite 105
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Phone: 301-347-7166
www.porphyriafoundation.com
Booth# 202

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www.ashg.org
Booth# 308

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www.amicustherapeutics.com
Booth# 601

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www.artel-usa.com
Booth# 1207

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Toll Free: 800-242-2787; Fax: 801-584-5209
www.aruplab.com/genetics
Booth# 815

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www.creatineinfo.org
Booth# 200

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www.amp.org
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www.audentestx.com
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www.avaxis.com
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www.bbrauncegat.com
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www.biogen.com
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Phone: 415-827-8129
www.bmrn.com
Booth# 500

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www.bgi.com/us
Booth# 636

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www.blueprintgenetics.com
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www.centogene.com/
Booth# 1015

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www.cgcgenetics.com/en/
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Los Angeles, CA 90027
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www.chla.org/pathology-and-laboratory-medicine
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www.cleargenetics.com
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www.clinicalgenome.org
Booth# 529

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www.color.com
Booth# 928

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www.concertgenetics.com
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www.ctgt.net
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Phone: 973-369-5714
www.coopersurgical.com
Booth# 834

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Brescia, 25121 Italy
Phone: 3427738606
www.copangroup.com
Booth# 634

Coriell Institute for Medical Research
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coriell.org
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www.childrensdayton.org
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www.ddccliniclab.org
Booth# 1105

Diagnostics Inc.
5795 Kearny Villa Road
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www.diagnomics.com
Booth# 1107

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www.dsdiagnosisnetwork.org
Booth# 104

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Plymouth Meeting, PA 19462
Phone: 610-825-6000; Fax: 610-834-1275
www.ecri.org
Booth# 1115
Inozyme’s No-Cost Genetic Testing for Rare Calcification Disorders

**Early Detection is Key**

In partnership with PreventionGenetics, Inozyme Pharma provides a no-cost, global genetic testing program to improve detection and understanding of two rare and debilitating calcification disorders, **ENPP1 Deficiency** and **ABCC6 Deficiency**. In both deficiencies, the body lacks the ability to produce critical proteins that regulate pyrophosphate, or PPI, levels in plasma. Low levels of plasma PPI have been linked to an increased risk of vascular calcification and bone mineralization defects. Depending on the gene mutation, the following conditions may occur:

<table>
<thead>
<tr>
<th>Genes</th>
<th>Deficiency</th>
<th>Condition</th>
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| **ENPP1** | ENPP1 Deficiency | • Generalized arterial calcification of infancy (GACI) type 1  
• Autosomal recessive hypophosphatemic rickets type 2 (ARHR2) |
| **ABCC6** | ABCC6 Deficiency | • GACI type 2 in infants  
• Pseudoxanthoma elasticum (PXE) |

**No-Cost Third Party Genetic Testing**

Inozyme Pharma and PreventionGenetics introduced the genetic testing program to increase awareness, reduce barriers to genetic testing, and help people and their healthcare providers make more informed decisions about these rare conditions.

Individuals who meet eligibility criteria can receive a no-cost third-party genetic test to determine if they have the condition or are a carrier for ENPP1 Deficiency and ABCC6 Deficiency.

**Information for Your Healthcare Professionals**

Your HCP can contact PreventionGenetics at 1-715-387-0484 and/or mail clinicaldnatesting@preventiongenetics.com or visit https://www.preventiongenetics.com/specialOrders?sp=sp051.

For more information, visit PreventionGenetics at **Booth 707**
Inozyme’s No-Cost Genetic Testing for Rare Calcification Disorders

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|       |           | • Autosomal recessive hypophosphatemic rickets type 2 (ARHR2) |
| ABCC6 | ABCC6 | • GACI type 2 in infants  
|       |           | • Pseudoxanthoma elasticum (PXE) |

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<table>
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<tr>
<th>Company</th>
<th>Address</th>
<th>Phone</th>
<th>Fax</th>
<th>Website</th>
<th>Booth#</th>
</tr>
</thead>
</table>
| EGL Genetic Diagnostics | 2460 Mountain Industrial Blvd.  
   Tucker, GA 30084 | 470-378-2193 | www.eeglgenetics.com | 533                      |
| ELLKAY           | 200 Riverfront Blvd.  
   Elmwood Park, NJ 07407 | 201-791-0606; 201-791-0605 | www.ELLKAY.com | 519                      |
| Elsevier         | 1600 John F. Kennedy Blvd., Suite 1800  
| Emedgene         | 2625 Middlefield Road, #451  
   Palo Alto, CA 94306-2516 | 408-507-4257 | www.emedgene.com | 403                      |
| Enzo             | 10 Executive Blvd.  
   Farmingdale, NY 11735 | 800-942-0430; 631-694-7501 | www.enzolifesciences.com | 417                      |
| Fabric Genomics, Inc. | 1611 Telegraph Avenue, Suite 500  
   Oakland, CA 94612 | 510-595-0800; 510-588-4523 | www.fabricgenomics.com | 923                      |
| FamHis, Inc.     | 604 NE Venezia Lane  
   Boca Raton, FL 33487 | 866-484-7890; 561-631-9277 | www.famhis.net | 516                      |
| FDNA             | 186 South Street, Floor 5  
   Boston, MA 02111 | 973-513-2520 | www.fdna.com | 1011                     |
| Food & Drug Administration Office of Women’s Health | 10903 New Hampshire Avenue  
   Silver Spring, Maryland 21666 | 301-796-9463 | | 1210                     |
| Fulgent Genetics | 4978 Santa Anita Avenue, Suite 205  
   Temple City, CA 91780 | 626-350-0537; 626-454-1667 | www.fulgentgenetics.com | 1128                     |
| GBF, Inc.        | 2427 Penny Road  
   High Point, NC 27265 | 336-369-5234; 336-665-0209 | www.gbf-inc.com | 932                      |
| Geisinger        | 100 N Academy Avenue  
   Danville, PA 17821 | 570-214-6918; 800-845-7112 | www.geisinger.org | 628                      |
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Toll Free: 888-682-5252
www.gene42.com
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www.gene-matters.com
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**Genome Medical**
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www.genomemedical.com
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www.genomenon.com
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www.globalgenes.org
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D301 Chuang Xin Mansion Zhongguancun Life Science Park
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www.ggc.org
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Phone: 608-775-4242
www.gundersenhealth.org
Booth# 735

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741 Sunset Avenue
Honolulu, HI 98616
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www.hawaiigenetics.org
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hcunetworkamerica.org
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www.physiciancareers.kp.org/scal
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lettercase.org/
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www.nccrcg.org
Booth# 302

National Organization for Rare Disorders, Inc.
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www.rarediseases.org
Booth# 105

NCBI Medical Genetics and Human Variation
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Booth# 531

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careers.osfhealthcare.org/
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www.ogt.com
Booth# 1020

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raregenomes.org
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<td>The Focus Foundation</td>
<td>2772 Rutland Road, Davidsonville, MD 21035</td>
<td>443-223-7323</td>
<td>thefocusfoundation.org</td>
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<td>Therapak, LLC</td>
<td>651 Wharton Drive, Claremont, CA 91711</td>
<td>470-655-3170; 760-614-2938</td>
<td>therapak.com</td>
<td>1209</td>
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<td>Thermo Fisher Scientific</td>
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<td>thermofisher.com/</td>
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<td>Translational Software</td>
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<td>206-777-4063</td>
<td>translationalsoftware.com</td>
<td>1018</td>
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<td>Twist Bioscience</td>
<td>455 Mission Bay Blvd., South, Suite 545</td>
<td>415-508-7956</td>
<td>twistbioscience.com</td>
<td>830</td>
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<td>UCSF Fetal Treatment Center</td>
<td>1855 4th Street, 2nd Floor, A-2432, San</td>
<td>800-793-3887; 415-502-0660</td>
<td>fetus.ucsf.edu/</td>
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<td>Ultragenyx Pharmaceutical</td>
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<td>800-719-0671</td>
<td>ultragenyx.com</td>
<td>1035</td>
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<td>Undiagnosed Diseases Network (UDN)</td>
<td>10 Shattuck Street, Boston, MA 02115</td>
<td>617-432-5788</td>
<td>udnconnect.org</td>
<td>201</td>
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<td>University of Chicago Genetic</td>
<td>5841 S Maryland Avenue, MC0077, Chicago, IL</td>
<td>773-834-0555; 773-702-9130</td>
<td>dnatesting.uchicago.edu</td>
<td>824</td>
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<tr>
<td>Services Laboratories</td>
<td>985440 Nebraska Medical Center, Omaha, NE</td>
<td>402-559-6935; 402-559-6391</td>
<td>unmc.edu/geneticslab</td>
<td>917</td>
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<td>Variantyx, Inc.</td>
<td>1671 Worcester Road, Suite 300, Framingham,</td>
<td>617-209-2090</td>
<td>variantyx.com</td>
<td>820</td>
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<td>WORLD Symposium</td>
<td>6001 Broken Sound Parkway NW, Suite 340,</td>
<td>561-609-0438</td>
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**6.00pm–7.30pm, 2 April, 2019**

Ballroom 6A, Washington State Convention Center, Seattle

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This symposium is supported by an education grant from AVROBIO, Inc.

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<th>Registration Refund</th>
<th>Application Fee Waiver</th>
<th>Rebate Coupon</th>
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<td>MD’s, PhD’s, DO’s, Laboratory Directors, and Corporate Executives</td>
<td>$325</td>
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<td>$775</td>
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<td>Genetics Counselors, Nurses, Physician Assistants, Dietitians, Laboratory Technologist/Technician, Advocate Leaders, and Non-profit Advocacy Representatives</td>
<td>$215</td>
<td>$50</td>
<td>$75</td>
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<td>Residents, Trainees and those in Fellowships</td>
<td>$30</td>
<td>$50</td>
<td>$75</td>
<td>$30</td>
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<td>Students</td>
<td>FREE MEMBERSHIP</td>
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Proposition Submission Opens for Scientific Sessions, Workshops, Short Courses and other presentations: April 2019
Proposal Submission Deadline: May 20, 2019
Online Submissions: www.acmgmeeting.net
Detailed program, registration and hotel information available September 2019
Abstract Submissions: Open: September 2019 | Deadline: November 5, 2019
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When a negative cfDNA isn’t the whole story: the value of genome-wide analysis
A review of cases referred through the Genome-Flex high risk pathway. Presented by Ron McCullough, PhD, MBA

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