Education | Spring/Summer 2025

THE ACMG
Medical Geneticist
The Newsmagazine of the American College of Medical Genetics and Genomics

Q&A with ACMG's New President, Dr. Mira Irons on Priorities, Opportunities and Challenges Ahead

In a recent interview with *The ACMG Medical Geneticist*, new ACMG President Mira B. Irons, MD, FACMG, FAAP, discussed her priorities for the College and where she sees the greatest opportunity for growth and change.

The ACMG Medical Geneticist (ACMG): As you begin your term as ACMG President, what are your top priorities for the College over the next two years?

Dr. Mira Irons (MI): When I began this role, I came in with a set of clear priorities, but as often happens, circumstances evolved quickly, especially considering recent changes in federal agencies in the healthcare and research environments. While my core goals remain the same, there's now an increased urgency and depth to the work ahead.

My first priority is to support and expand ACMG membership, especially as many members face institutional and funding challenges. We must be a strong support system for clinical and laboratory geneticists and encourage cross-specialty collaboration, including genetic counselors, nurse practitioners and physician assistants. Given our workforce challenges and the increasing role that genetics and genomics will play in healthcare, the future of genetics lies in cross-specialty collaboration.

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From the **Editor-in-Chief**

Education is the theme of this newsmagazine, which recaps the activities of the 2025 ACMG Annual Clinical Genetics Meeting in Los Angeles. The meeting commenced with the Presidential Plenary on genetics in the media. This session was engaging, informative and entertaining. It is comforting to know that there are highly qualified geneticists promoting and protecting our specialty on social platforms. The ACMG Foundation for Genetic and Genomic Medicine recognized Dr. Stephen Cederbaum with the David L. Rimoin Lifetime Achievement Award in Medical Genetics. Dr. Cederbaum noted the importance of enduring friendships and spoke of the value of kindness, respect, curiosity and a sense of humor throughout his career. The complete listing of the 10 awards presented by the Foundation appears on page 24.

Students were afforded several opportunities to interact with genetic professionals at the conference. These included the Early Genetics Trainee and Faculty Forum to discuss the future of genetics, the Early Career Genetic Mentor Lunch to explore career options with established members of the college and the Student Workshop on careers in medical genetics and genomics. In addition, five of the 18 medical students who participated in the Summer Genetics Scholars Program (page 41) were in attendance. High school and college students participated in the E3 Genomics Pathway Program (page 37), connecting with more than 40 geneticists for an introduction and immersion into medical genetics. Additional student involvement in our field is encouraged through Student Interest Groups (SIGs). The Membership Committee describes the Student Interest Group (SIG) Program (page 40) and explains the many advantages of belonging to a SIG. Students and mentors alike benefit from involvement with these groups.

For Medical Genetics Awareness Week (March 18-21), we were reminded of 'The Joy of Working in Medical Genetics', with our members and others posting heartfelt messages and taking selfies to show their pride in our field (page 32).

In the Q&A (cover), Dr. Mira Irons shares the priorities, challenges and opportunities that lie ahead for the College during her upcoming term as our president. Finally, we close with a tribute to Dr. OJ Miller who passed away last year (page 42). OJ had an impressive genetics pedigree and a brilliant career during which he made numerous outstanding contributions to the fields of cytogenetics and medical genetics.

Until next time,

Katy Phelan

Katy Phelan, PhD, FACMG, Editor
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Ngoni Faya, PhD, Clinical Fellow at Cincinnati Children's Hospital, won an ACMG Genetics and Genomics Review Course Syllabus at the ACMG Annual Meeting.

Meet ACMG's Newest Staff Members

Judy Froehlich, MBA
Director of Communications and Public Relations

Judy Froehlich brings more than 25 years of communications and marketing experience across both the public and private sectors. She most recently served as the National Director of Marketing and Communications for the Hydrocephalus Association. Prior to that, she spent a decade at the University of Central Florida (UCF) as Director of Marketing and Communications, where she established and led the department from the ground up. Judy holds a Bachelor of Science in Marketing and an MBA from UCF. She is passionate about nonprofit work in the biomedical field and is deeply committed to making a meaningful impact through mission-driven communications and public relations.

"I'm honored to join ACMG and excited to support our mission through thoughtful, effective communications and public relations," says Judy. "I look forward to working collaboratively with the team to strengthen the College's voice and expand its reach."

Catherine Wattenberg
Director of Publications

Catherine Wattenberg brings more than 40 years of experience in scholarly publishing, beginning at *The Chronicle of Higher Education* and continuing with several academic journals. Most recently, she served as Director of Publications at the American Association of Immunologists, where she helped lead the transition from self-publishing to a partnership with Oxford University Press. Catherine is passionate about peer-reviewed science and the power of clear, credible communication.

"I am delighted to join the *Genetics in Medicine* and *Genetics in Medicine Open* journals' family. I look forward to collaborating with our dedicated community of members, authors, reviewers and editors as we navigate the challenges and opportunities ahead."

ClinGen Engages Students with Crowd-Sourced Literature Annotations

by **Pepper St. Clair, BSc; Justyne Ross, PhD** and **Courtney Thaxton, PhD**

The curation efforts of the Clinical Genome Resource (ClinGen) require extensive, time- consuming literature review. To increase the efficiency of ClinGen's expert panels, a crowd-sourced literature annotation activity called Baseline Annotation (https://clinicalgenome.org/curation-activities/baseline-annotation) was created. Using gene-specific protocols and a free web tool, hypothes.is (https://web.hypothes.is), volunteers contribute standardized and tagged summaries to case report repositories that are accessed by ClinGen biocurators. These annotations save curators time surveying the literature for cases with specific qualities.

Baseline Annotation is an opportunity for ClinGen to recruit and engage future genomics professionals. Students engaged in annotation build transferable skills in identifying and extracting key evidence. Annotation also serves as an introduction to data science and curation. This activity is virtual and asynchronous, which allows for students living or studying in rural areas to participate. Participation in annotation only requires access to a computer with Wi-Fi. ClinGen's annotation training is aimed at the fundamental level so students can participate without a pre-established background in genetics or biomedical studies. Students have been engaged in this activity as early as their junior year of high school.

University of North Carolina at Chapel Hill, a ClinGen grant institution and "Tier 1" research and medical school, and California State University, Monterey Bay, a designated Hispanic-Serving Institution (HSI), have incorporated Baseline Annotation into coursework, research for credit and training programs. Students who have completed annotation work as a part of these efforts have gone on to attend medical school, genetic counseling school, PhD programs and to work for ClinGen. Although students have benefited from this activity, anyone is welcome to participate. In fact, more than half of the ClinGen Baseline Annotation volunteers identify as "genetics professionals." Those who would like to engage their students or trainees through this activity are encouraged to reach out to the ClinGen Community Curation Working Group at volunteer@clinicalgenome.org.

The second key priority is advocacy. We're working on addressing hurdles like prior authorization for genetic testing, which delay care at a time when our diagnostic capabilities are rapidly advancing, and therapies are becoming more feasible as well as LDT legislation and advocating for PhD laboratory geneticists to bill for their services.

Another major advocacy concern is continued support for newborn screening. With the dissolution of the federal advisory committee (ACHDNC), we've formed a dedicated task force to determine how ACMG can best support the continuity and progress of newborn screening during this transition.
Finally, our advocacy efforts must also address threats to research funding. We're partnering with other societies, including ASHG and NSGC, to amplify our voice on shared challenges.

My third major priority is collaboration - working with other professional societies across the healthcare continuum. The full potential of genetics and genomics in clinical care and public health depends on collaboration with other specialties. If we want to improve access and equity in genomic medicine, building those partnerships will be essential.

ACMG: The landscape of medical genetics and genomics is evolving rapidly. Where do you see the most significant opportunities for ACMG and its members moving forward?

MI: There are several exciting opportunities on the horizon, and ACMG is well positioned to lead in this evolving space.

> **"The future of genetics lies in cross-specialty collaboration."**

First and foremost, we have a chance to provide meaningful guidance and leadership as genetics and genomics become more integrated into healthcare. That means focusing on access, ensuring equitable availability of genetic services and emphasizing collaboration across specialties to deliver the best care possible.

We're at a pivotal moment in our specialty as genetics shifts from a primarily diagnostic field to a therapeutic one. While biochemical geneticists have been able to provide biochemically informed treatment to patients, the same has not been as true for patients with rare disease or as-yet unknown conditions. Advances in molecular diagnostics now enable not only more precise identification of the genetic basis of conditions but also the emergence of genetically informed treatments. This progress also paves the way for population health applications, allowing earlier intervention for at-risk individuals.

For ACMG members, this transition represents a significant opportunity. The College can support professionals through education and training, helping them understand how to participate in clinical trials, how to access new therapies and how to collaborate with other specialties in managing genetically complex conditions. This new environment also provides opportunities to expand the educational activities of the ACMG Genetics Academy to provide even more CME activities to our members and other specialists as well as to patients and families.

Expanded newborn screening is a powerful example of population health. It gives every baby an equal chance at early detection and care before symptoms appear. It's a model we aim to extend across the lifespan.

ACMG: What do you see as the biggest challenges currently facing the field of medical genetics and genomics, and how can ACMG help address them?

MI: One of our biggest challenges is size. Our community includes roughly 3,000 board-certified medical and laboratory geneticists and about 7,100 genetic counselors nationwide. While our membership is "small but mighty," our members also often contribute to larger societies like AAP or ACOG. We must ensure that ACMG itself is recognized as the leading voice in clinical and laboratory practice of medical genetics and genomics.

Visibility is also a challenge. ACMG plays a key role in areas like newborn screening and policy, but our contributions are sometimes overlooked in favor of other larger professional specialty societies.

ACMG should be the go-to resource for insights on genetics and genomics in healthcare. Policymakers, journalists and specialty societies should instinctively reach out to us. We have made significant advances in this area over the years, and we've made this a priority in our strategic plan, but there's more to do to ensure we're on everyone's radar.

Finally, with the ongoing reorganization of federal agencies, ACMG must stay agile in advocating for policies that recognize the essential role of research and genetics and genomics in patient care.

ACMG: Advocacy and policy changes continue to impact our field. How do you see ACMG's role in shaping the future of medical genetics and ensuring continued progress?

MI: We're at a pivotal moment in medical genetics. Historically focused on the care and evaluation of patients with rare or unknown conditions, the field is now entering mainstream healthcare, not just for treatment, but for population health. ACMG plays a critical role in preparing members to lead this transformation in their hospitals and institutions, offering tools, education and support to empower them.

Collaboration is essential, as many organizations in our field face shared challenges. ACMG participates in numerous coalitions which help unify efforts and strengthen our collective voice. A recent example is the letter developed by our newborn screening coalition, which was signed by more than 200 organizations, including medical associations, hospitals, laboratories, pharmaceutical companies and patient advocacy organizations.

Equally important is ensuring we are at the table. Decision-makers need to hear directly from us about the vital role of genetics and genomics in healthcare, which means engaging with legislators and participating in public policy discussions.

We also need to improve communication with our members. Transparency and feedback are essential to keeping ACMG relevant, especially for younger professionals managing competing priorities.

Finally, we must make advocacy accessible. Not everyone feels comfortable engaging with lawmakers, but it's a skill we can

CEO CORNER

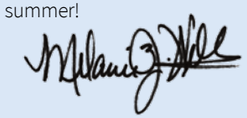
ACMG is dedicated to serving as your leader in genetics and genomics education, information and resources. Whether you are exploring the field as a career option, preparing for board certification exams or established in the field, ACMG has offerings that will support your continued professional development. For example, our highly successful Annual Clinical Genetics Meeting this March in Los Angeles offered a wide array of relevant content, networking opportunities and exposure to innovations in the field. In case you couldn't attend the Annual Meeting, missed a session or want to review the material covered in a session you attended, consider purchasing the 2025 Annual Clinical Genetics Meeting Digital Edition which offers more than 60 hours of content and is now available in our ACMG Genetics Academy.

We are continuing our informative quarterly webinar series coordinated by our journals, *Genetics in Medicine* and *Genetics in Medicine Open*, in partnership with Elsevier and Illumina. The next webinar entitled "A Revolution in Genomic Medicine: Transformative Treatments for Genetic Diseases" will be held in July and will address transformative treatments for genetic diseases. Let's continue the momentum from this year's first webinar, The Big Data Problem in Genomic Medicine: Acquisition, Processing, Interpretation, Storage and Retrieval presented by Kyle Retter, MS, which drew more than 1,300 registrants.

Are you studying for your board certification exam? The 2025 Genetics & Genomics Review Course (GGRC) is an excellent resource to help you prepare. The self-paced content and live question and answer sessions are now available as the 2025 GGRC Digital Edition in our ACMG Genetics Academy.

Our Summer Genetics Scholars Program launched in late May with 17 institutions welcoming students across the country for a 6-week program. This program exposes 1st year medical students to the field of genetics and genomics through unique experiences tailored specifically for them. In addition, our newest early exposure program, E3: Equip, Engage, and Empower, Genomics Pathways Program, introduced undergraduate and community college students to the field through monthly educational sessions and quarterly mentoring sessions which started in May.

As the interdisciplinary home for genetics and genomics professionals, ACMG is committed to ensuring that we adapt to the changes that are occurring in the field and keeping you well informed. I hope that you have a productive summer!



Melanie J. Wells, MPH, CAE
Chief Executive Officer

teach. With tools like talking points, meeting support and letter-writing campaigns, ACMG can empower members to speak up, especially on state-level issues like newborn screening.

ACMG: **Where does the College need more volunteer engagement, and what advice do you have for members looking to get more involved?**

MI: The College is always looking for greater member engagement, particularly in our advocacy efforts, committees and work groups. If you're interested in getting involved, the best first step is to reach out. Let us know your concerns, how the College can support you in patient care or where you see opportunities for improvement. You can email me directly at Mira.Irons@childrens.harvard.edu.

Each year, we call for volunteers and encourage members to submit their names and interests. While not everyone will be selected immediately, please don't be discouraged. Committee and work group positions rotate regularly, and we're always looking for engaged, enthusiastic members to step in.

I also encourage members to propose ideas for new work groups or committees—let us know where you see a need the College could address. Your input helps guide the direction of our efforts, and your voice truly matters.

ACMG: **With recent shifts in healthcare policy, technology and workforce dynamics, how do you see ACMG adapting to support its members and the broader genetics community?**

MI: ACMG has been working proactively to expand support for members as healthcare policy and technology evolve. One key area of focus has been education. We have introduced more webinars and learning opportunities than ever, thanks to the leadership of our Education and Professional Development Committee. Our annual meeting continues to grow and has become increasingly popular with sessions that include other specialties.

We're also prioritizing communication. Members can expect more frequent updates on advocacy, policy shifts and new resources. While we understand inbox fatigue, we want members to feel informed and have opportunities to provide feedback in real time. The issues we face are too important for anyone to feel disconnected.

ACMG: **Is there anything else you'd like to share with ACMG members as you step into this leadership role?**

MI: I am thrilled and honored to serve as ACMG President and deeply grateful for the trust our members have placed in me. Medical genetics has been my life's work, and this is a pivotal moment in our field. We have the opportunity to transform healthcare by advancing diagnoses, therapies and population health. However, we must always focus on ensuring equitable access to genetic and genomic services for all. I look forward to working with you all to continue the great work ACMG has done and to expand its impact.

Dr. Mira B. Irons Begins ACMG Presidency

Mira B. Irons, MD, FACMG, FAAP, is the new president of the American College of Medical Genetics and Genomics. Dr. Irons assumed this responsibility from Susan D. Klugman, MD, FACMG, FACOG, who completed her two-year term at the culmination of the 2025 Annual Clinical Genetics Meeting in Los Angeles this March.

"I am honored, humbled and thrilled to assume the presidency of the ACMG at this remarkable time in the history of our specialty, one that we have been anxiously anticipating," said Dr. Irons. "A time when rapid advancements in genomic testing increasingly informs not only the daily care of our patients, but also the development of therapies for conditions that were previously untreatable. We now increasingly have the ability to use the knowledge and tools of genetics and genomics to impact larger populations by working with our colleagues across all specialties, educating our patients and the public, and most importantly, advocating for the resources necessary to do so responsibly with a focus on equity, access, and improving disparities in healthcare."

Dr. Irons has held a number of positions within the ACMG, including as a member of the Finance Committee, the Board of Directors and the Nominations Committee. For the last two years, she served the College as president-elect. She has been a member of the ACMG Foundation Development Committee and the Task Force on Medical Genetics Education. She chaired the Conflict of Interest Subcommittee, the Education and Professional Development Committee and the Maintenance of Certification Committee and served as a member of the Professional Practices and Guidelines Committee, along with many more volunteer roles within the ACMG.

Recognized as a leading expert in medical genetics, Dr. Irons is associate chief of the Division of Genetics and Genomics at Boston Children's Hospital. She holds the Park Gerald Chair in Genetics in the Department of Pediatrics at Boston Children's Hospital. She also serves as the program director for the Medical Genetics Residency and Fellowship Programs at Boston Children's Hospital/Harvard Medical School, where she is a member of the faculty. She is a member of the Board of Directors of St. Christopher's Hospital for Children in Philadelphia, as well as a member of the Roundtable on Genomics and Precision Health at the National Academy of Medicine.

From 2013-2019 she served as senior vice president for academic affairs at the American Board of Medical Specialties. She was later group vice president and chief health and science officer at the American Medical Association from 2019-2021 and president and CEO and the Thomas W. Langfitt Chair at the College of Physicians of Philadelphia from 2021-2023.

Dr. Irons has participated in multiple local and national clinical trials and co-authored many peer-reviewed publications. Her research has focused on integrating new forms of genetic and genomic testing into clinical practice, identification of new biochemical genetic disorders and neurofibromatosis, type 1.

A graduate of the Northwestern University Feinberg School of Medicine, Dr. Irons completed a residency in Pediatrics at Children's Memorial Hospital in Chicago, a fellowship in Pediatrics (Genetics) at Massachusetts General Hospital in Boston and a second fellowship in Clinical and Biochemical Genetics at Children's Hospital in Boston.

ACMG Welcomes Six New Board Members

ACMG welcomed six new directors, including a new president-elect, to its Board of Directors during the 2025 ACMG Annual Clinical Genetics Meeting. The new Board members will serve as advocates for the organization and will assist in shaping and implementing the mission, vision and direction of the College. They will serve six-year terms from April 2025 to March 2031. Please join us in welcoming:

Christa L. Martin PhD, FACMG: President-Elect

Dr. Martin is currently the chief scientific officer at Geisinger and vice dean for research at Geisinger's School of Medicine. Her focus has been on integrating genomics into routine healthcare, particularly in the areas of neurodevelopmental and psychiatric disorders and population health genomic screening. She is the first PhD to serve the College as President-Elect.

Josh Deignan, PhD, FACMG: Clinical Molecular Genetics Director

Board-certified in Clinical Molecular Genetics and Genomics, Dr. Deignan is a clinical professor in the Department of Pathology and Laboratory Medicine at the David Geffen School of Medicine at the University of California, Los Angeles (UCLA).

Lauren J. Massingham, MD, FACMG: Clinical Genetics Director

Dr. Massingham is a board-certified medical geneticist who specializes in the care of pediatric and adult patients. She currently serves at Hasbro Children's Hospital and Rhode Island Hospital and is an associate professor in the Department of Pediatrics at The Warren Alpert Medical School of Brown University.

David T. Miller, MD, PhD, FACMG: Clinical Genetics Director

A board-certified clinical geneticist at Boston Children's Hospital, Dr. Miller directs the Neurofibromatosis Research Initiative. He is also an associate molecular pathologist at Brigham and Women's Hospital. At Harvard Medical School, Dr. Miller serves as an associate professor of Pediatrics and co-director of the Advanced Integrated Science Course in Human Genetics.

Douglas R. Stewart, MD, FACMG: Clinical Genetics Director

Dr. Stewart is a board-certified medical geneticist with more than 25 years of experience in clinical genetics, cancer risk assessment and genomics research. A senior investigator at the National Cancer Institute and adjunct investigator at the National Human Genome Research Institute, he focuses on the relationship between germline variants and cancer risk in both pediatric and adult populations.

William R. Wilcox, MD, PhD, FACMG: Clinical Biochemical Genetics Director

A board-certified clinical geneticist and biochemical geneticist, Dr. Wilcox is the medical director of the Emory Genetic Clinical Trials Center and the founder of the Medical Biochemical Genetics Training Program at Emory. He previously served as professor of pediatrics in residence at UCLA School of Medicine, where launched a combined Pediatrics-Medical Genetics Training Program based at Cedars-Sinai Medical Center.

Also, during the meeting, six board members completed their terms. The College thanks them for their dedicated service: Karen W. Gripp, MD, FACMG; Dietrich Matern, MD, PhD, FACMG; Michael F. Murray, MD, FACMG; Cynthia M. Powell, MD, FACMG; Heidi L. Rehm, PhD, FACMG; and Marc S. Williams, MD, FACMG. To read the news release about ACMG's new board members, visit acmg.net/ACMG/News/Newsroom. To view the full roster of the ACMG Board of Directors, visit acmg.net.

ACMG Honors Outgoing Committee Chairs

ACMG committees are vital to advancing the work of the College. These committees are composed of dedicated individuals from our membership who volunteer their time and expertise to serve the College and our profession. During the 2025 ACMG Annual Clinical Genetics Meeting, six committee chairs completed their terms. The College extends its sincere appreciation to these outgoing committee chairs for their dedicated service:

- **John W. Belmont, MD, PhD, FACMG - Economics of Genetic Services Committee**
- **Lindsay C. Burrage, MD, PhD, FACMG - Therapeutics Committee**
- **Jodi D. Hoffman, MD, FACMG - Program Committee for Annual Meeting**
- **Aditi S. Parikh, MD, FACMG - Education and Professional Development Committee**
- **Gordana Raca, MD, PhD, MS, FACMG - Laboratory Quality Assurance Committee**
- **Gopalrao V. Velagaleti, PhD, FACMG - Continuing Certification Program Committee**

To view the complete list of ACMG committees and their rosters, visit www.acmg.net.

More Important than Ever: ACMG Resources for State Advocacy

by **David Molina, ACMG Advocacy Manager** and **Michelle McClure, PhD, ACMG Director of Public Policy**

The first half of 2025 has been met with a flurry of federal government actions, including executive orders, confirmation of new cabinet officials, reorganization of federal agencies and termination of federal programs and funding. The rapid pace of changes and numerous related legal challenges have led to confusion and frustration, even among some members of Congress. Further, Congress has been tasked with implementing major budget priorities set by the Administration. As a result, Congress has been slow to pass legislation, especially those related to healthcare.

However, in contrast to the federal government, state governments are often able to respond more quickly and pass more legislation related to healthcare. Since state legislative sessions are often shorter, more productive and have stricter deadlines to finish legislative business, legislation is passed very quickly in some states. Further, some issues are specifically left to states to determine, such as medical licensure, conditions for newborn screening and many Medicaid coverage policies. Thus, it is crucial that healthcare professionals engage with their state legislators and establish themselves as a resource for them. To assist, the ACMG is continuing to update resources to keep our members informed and help them engage at the state level to become advocates for their medical policy priorities.

One new resource is the ACMG Policy Map, available on our website at acmg.net/advocacy. The Policy Map is an interactive tool to help track and share state legislation related to medical genetics. By being able to see what is going on in their state, members can more readily become advocates. The ACMG Advocacy website also contains helpful information about engaging in state-level advocacy, including information on how to find a state legislature's website and identify your legislators. There are numerous ways in which genetics professionals can engage with their state legislators, such as requesting meetings, attending virtual town halls, submitting letters about pending bills and notifying them about healthcare access issues in their state.

With the continued uncertainty and constantly fluctuating federal government policies, it is increasingly critical for genetics healthcare professionals to engage at the state level. We encourage members to explore the new ACMG Policy Map and additional information about engaging with legislators in the ACMG Advocacy Resource Center available at acmg.net/advocacy. If you have questions or recommendations for additional resources, including genetics-related topics for the ACMG Policy Map, please contact us at advocacy@acmg.net.

The first and only FDA-approved treatment for Rett syndrome[1,2]

Add more sparkle

to the world around us

DAYBUE™ is helping to advance the treatment paradigm in Rett syndrome (RTT)[1,2]

Clare, age 8, living with ***Rett syndrome***

Kate, age 9, living with ***Rett syndrome***

Maddy, age 20, living with ***Rett syndrome***

In the pivotal trial and open-label extension (OLE) studies, DAYBUE was evaluated for up to 3 years[1,3,4]

Daybue™ (trofinetide)

The efficacy and safety of DAYBUE were evaluated in LAVENDER™, a 12-week, randomized, double-blind, placebo-controlled, Phase 3 trial.[1,5]

Participants who completed the trial could enroll in the OLE studies for up to an additional 144 weeks.[3,4]

Both caregivers and clinicians evaluated improvements with DAYBUE across a range of RTT signs and symptoms[1]

- The **Rett Syndrome Behaviour Questionnaire (RSBQ)**, completed by caregivers, is the most widely used measure in RTT studies[5]
- The **Clinical Global Impression-Improvement (CGI-I)** scale, utilized by clinicians, provides meaningfulness to the caregiver measure[5]

Indication

DAYBUE is indicated for the treatment of Rett syndrome in adults and pediatric patients 2 years of age and older.

Important Safety Information

- **Warnings and Precautions**
  - **Diarrhea:** In a 12-week study and in long-term studies, 85% of patients treated with DAYBUE experienced diarrhea. In those treated with DAYBUE, 49% either had persistent diarrhea or recurrence after resolution despite dose interruptions, reductions, or concomitant antidiarrheal therapy. Diarrhea severity was mild or moderate in 96% of cases. In the 12-week study, antidiarrheal medication was used in 51% of patients treated with DAYBUE.

    Advise patients to stop laxatives before starting DAYBUE. If diarrhea occurs, patients should notify their healthcare provider, consider starting antidiarrheal treatment, and monitor hydration status and increase oral fluids, if needed. Interrupt, reduce dose, or discontinue DAYBUE if severe diarrhea occurs or if dehydration is suspected.
  - **Vomiting:** In a 12-week study, vomiting occurred in 29% of patients treated with DAYBUE and in 12% of patients who received placebo.

    Patients with Rett syndrome are at risk for aspiration and aspiration pneumonia. Aspiration and aspiration pneumonia have been reported following vomiting in patients being treated with DAYBUE. Interrupt, reduce dose, or discontinue DAYBUE if vomiting is severe or occurs despite medical management.
  - **Weight Loss:** In the 12-week study, 12% of patients treated with DAYBUE experienced weight loss of greater than 7% from baseline, compared to 4% of patients who received placebo. In long-term studies, 2.2% of patients discontinued treatment with DAYBUE due to weight loss. Monitor weight and interrupt, reduce dose, or discontinue DAYBUE if significant weight loss occurs.

RTT symptom areas evaluated

Using the RSBQ scale, caregivers assessed symptom areas such as[1,6]:

- Breathing
- Hand movements or stereotypies
- Repetitive behaviors
- Nighttime behaviors
- Vocalizations
- Facial expressions
- Eye gaze
- Mood

Because each individual with RTT is unique—with a unique set of symptoms—improvements with DAYBUE may be different for everyone.
Learn how DAYBUE has impacted these patients' lives.

View the long-term, open-label safety and efficacy data at DAYBUEhcp.com

Endorsed in clinical practice

>1500
patients initiated on DAYBUE treatment[7,a]

~3 in 10
individuals diagnosed with RTT in the US have been initiated on DAYBUE[7,8]

[a]As reported by ANOVO specialty pharmacy from March 10, 2023 through August 26, 2024.

Important Safety Information (cont'd)

- **Adverse Reactions:** The common adverse reactions (≥5% for DAYBUE-treated patients and at least 2% greater than in placebo) reported in the 12-week study were diarrhea (82% vs 20%), vomiting (29% vs 12%), fever (9% vs 4%), seizure (9% vs 6%), anxiety (8% vs 1%), decreased appetite (8% vs 2%), fatigue (8% vs 2%), and nasopharyngitis (5% vs 1%).
- **Drug Interactions: Effect of DAYBUE on other Drugs**
  - DAYBUE is a weak CYP3A4 inhibitor; therefore, plasma concentrations of CYP3A4 substrates may be increased if given concomitantly with DAYBUE. Closely monitor when DAYBUE is used in combination with orally administered CYP3A4 sensitive substrates for which a small change in substrate plasma concentration may lead to serious toxicities.
  - Plasma concentrations of OATP1B1 and OATP1B3 substrates may be increased if given concomitantly with DAYBUE. Avoid the concomitant use of DAYBUE with OATP1B1 and OATP1B3 substrates for which a small change in substrate plasma concentration may lead to serious toxicities.
- **Use in Specific Population: Renal Impairment**
  - DAYBUE is not recommended for patients with severe renal impairment.

DAYBUE is available as an oral solution (200 mg/mL).

Please read the Brief Summary of full Prescribing Information on the next page.

References: 1. Acadia Pharmaceuticals Inc. DAYBUE [Package Insert]. San Diego, CA, 2024. **2.** Acadia Pharmaceuticals announces U.S. FDA approval of DAYBUE™ (trofinetide) for the treatment of Rett syndrome in adult and pediatric patients two years of age and older. [press release]. Acadia Pharmaceuticals Inc. March 10, 2023. **3.** Percy AK, Neul JL, Benke TA, et al. Trofinetide for the treatment of Rett syndrome: results from the open-label extension LILAC study. *Med*. 2024;5(9):1178-1189.e3. **4.** Percy AK, Neul JL, Benke TA, et al. Trofinetide for the treatment of Rett syndrome: long-term safety and efficacy results of the 32-month, open-label LILAC-2 study. *Med*. 2024:S2666-6340(24)00253-8. doi:10.1016/j.medj.2024.06.007 **5.** Neul JL, Percy AK, Benke TA, et al. Trofinetide for the treatment of Rett syndrome: a randomized phase 3 study. *Nat Med*. 2023;29(6):1468-1475. **6.** Mount RH, Charman T, Hastings RP, Reilly S, Cass H. The Rett Syndrome Behaviour Questionnaire (RSBQ): refining the behavioural phenotype of Rett syndrome. *J Child Psychol Psychiatry*. 2002;43(8):1099-1110. **7.** Acadia Pharmaceuticals Inc. Data on file. DAYBUE Weekly Performance Update report. 2024. **8.** Acadia Pharmaceuticals Inc. Data on file. Diagnosed RTT patients in the US. March 2023.

ACADIA®

DAYBUE™ (trofinetide) oral solution
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Brief Summary: This information is not comprehensive. Visit www.DAYBUEhcp.com to obtain the full Prescribing Information or call 1-844-422-2342

1 INDICATIONS AND USAGE
DAYBUE is indicated for the treatment of Rett syndrome in adults and pediatric patients 2 years of age and older.

2 DOSAGE AND ADMINISTRATION
Administer DAYBUE orally twice daily, in the morning and evening, according to patient weight as shown in the table below. DAYBUE can be taken with or without food.

Recommended Dosage of DAYBUE in Patients 2 Years of Age and Older

Patient Weight	DAYBUE Dosage	DAYBUE Volume
9 kg to <12 kg	5,000 mg twice daily	25 mL twice daily
≥12 kg to <20 kg	6,000 mg twice daily	30 mL twice daily
≥20 kg to <35 kg	8,000 mg twice daily	40 mL twice daily
≥35 kg to <50 kg	10,000 mg twice daily	50 mL twice daily
≥50 kg	12,000 mg twice daily	60 mL twice daily

Administration Information
Administer DAYBUE orally or via gastrostomy (G) tube; doses administered via gastrojejunal (GJ) tubes must be administered through the G-port. A calibrated measuring device, such as an oral syringe or oral dosing cup, should be obtained from the pharmacy to measure and deliver the prescribed dose accurately. A household measuring cup is not an adequate measuring device. Discard any unused DAYBUE oral solution after 14 days of first opening the bottle.

Dose Modification for Diarrhea or Weight Loss
Advise patients to stop laxatives before starting DAYBUE. Interrupt, reduce the dosage, or discontinue DAYBUE if severe diarrhea occurs, if dehydration is suspected, or if significant weight loss occurs.

Dose Modification for Vomiting After Administration
If vomiting occurs after DAYBUE administration, an additional dose should not be taken. Instead, continue with the next scheduled dose. Interrupt, reduce dose, or discontinue DAYBUE if vomiting is severe or occurs despite medical management.

Dosage Recommendations in Patients With Renal Impairment
No dosage adjustment is recommended for patients with mild renal impairment (estimated glomerular filtration rate [eGFR] 60 to 89 mL/min for adult patients or 60 to 89 mL/min/1.73 m^2 for pediatric patients). The recommended dosage of DAYBUE for patients with moderate renal impairment (eGFR 30 to 59 mL/min for adult patients or 30 to 59 mL/min/1.73 m^2 for pediatric patients) is described in Table 2 of the Prescribing Information. DAYBUE is not recommended for patients with severe renal impairment (eGFR less than 30 mL/min for adult patients or less than 30 mL/min/1.73 m^2 for pediatric patients).

Missed Dose
If a dose of DAYBUE is missed, the next dose should be taken as scheduled. Doses should not be doubled.

5 WARNINGS AND PRECAUTIONS
Diarrhea
In a 12-week randomized, double-blind, placebo-controlled study (Study 1) and in long-term studies, 85% of patients treated with DAYBUE experienced diarrhea. In those treated with DAYBUE, 49% either had persistent diarrhea or recurrence after resolution despite dose interruptions, reductions, or concomitant antidiarrheal therapy. Diarrhea severity was of mild or moderate severity in 96% of cases. In Study 1, antidiarrheal medication was used in 51% of patients treated with DAYBUE.

Advise patients to stop laxatives before starting DAYBUE. If diarrhea occurs, patients should notify their healthcare provider, consider starting antidiarrheal treatment, and monitor hydration status and increase oral fluids, if needed. Interrupt, reduce dose, or discontinue DAYBUE if severe diarrhea occurs or if dehydration is suspected.

Weight Loss
In Study 1, 12% of patients treated with DAYBUE experienced weight loss of greater than 7% from baseline, compared to 4% of patients who received placebo. In long-term studies, 2.2% of patients discontinued treatment with DAYBUE due to weight loss.

Monitor weight and interrupt, reduce dose, or discontinue DAYBUE if significant weight loss occurs.

Vomiting
In Study 1, vomiting occurred in 29% of patients treated with DAYBUE and in 12% of patients who received placebo.

Patients with Rett syndrome are at risk for aspiration and aspiration pneumonia. Aspiration and aspiration pneumonia have been reported following vomiting in patients being treated with DAYBUE. Interrupt, reduce dose, or discontinue DAYBUE if vomiting is severe or occurs despite medical management.

6 ADVERSE REACTIONS
The following clinically significant adverse reactions are described elsewhere in labeling:

- Diarrhea *[see Warnings and Precautions]*
- Weight Loss *[see Warnings and Precautions]*
- Vomiting *[see Warnings and Precautions]*

Clinical Trials Experience
In controlled and uncontrolled trials in patients with Rett syndrome, 260 patients ages 2 to 40 years were treated with DAYBUE, including 109 patients treated for more than 6 months, 69 patients treated for more than 1 year, and 4 patients treated for more than 2 years.

Adult and Pediatric Patients With Rett Syndrome 5 Years of Age and Older
The safety of DAYBUE was evaluated in a randomized, double-blind, placebo-controlled, 12-week study of patients with Rett syndrome (Study 1). In Study 1, 93 patients received DAYBUE, and 94 patients received placebo. All patients were female, 92% were White, and the mean age was 11 years (range 5 to 20 years).

Adverse Reactions Leading to Discontinuation of Treatment
Eighteen patients (19%) receiving DAYBUE had adverse reactions that led to withdrawal from the study. The most common adverse reaction leading to discontinuation of treatment with DAYBUE was diarrhea (15%).

Common Adverse Reactions
Adverse reactions that occurred in Study 1 in at least 5% of patients treated with DAYBUE and were at least 2% more frequent than in patients on placebo are presented in the following table.

Adverse Reactions in at Least 5% of Patients Treated With DAYBUE and at Least 2% Greater than Placebo in Study 1

Adverse Reaction	DAYBUE (N=93) %	Placebo (N=94) %
Diarrhea	82	20
Vomiting	29	12
Fever	9	4
Seizure	9	6
Anxiety	8	1
Decreased appetite	8	2
Fatigue	8	2
Nasopharyngitis	5	1

Pediatric Patients With Rett Syndrome 2 to 4 Years of Age
In an open-label study in pediatric patients 2 to 4 years of age with Rett syndrome, a total of 13 patients received DAYBUE for at least 12 weeks and 9 patients received DAYBUE for at least 6 months. Adverse reactions in pediatric patients 2 to 4 years of age treated with DAYBUE were similar to those reported in adult and pediatric patients 5 years of age and older with Rett syndrome in Study 1.

Postmarketing Experience
The following adverse reactions have been identified during postapproval use of DAYBUE. Because these reactions are reported voluntarily from a population of uncertain size, it is not always possible to reliably estimate their frequency or establish a causal relationship to drug exposure.

Aspiration and aspiration pneumonia secondary to vomiting.

7 DRUG INTERACTIONS
Effect of DAYBUE on Other Drugs
Trofinetide is a weak CYP3A4 inhibitor; therefore, plasma concentrations of CYP3A4 substrates may be increased if given concomitantly with DAYBUE. Closely monitor when DAYBUE is used in combination with orally administered CYP3A4 sensitive substrates for which a small change in substrate plasma concentration may lead to serious toxicities.

Plasma concentrations of OATP1B1 and OATP1B3 substrates may be increased if given concomitantly with DAYBUE. Avoid the concomitant use of DAYBUE with OATP1B1 and OATP1B3 substrates for which a small change in substrate plasma concentration may lead to serious toxicities.

8 USE IN SPECIFIC POPULATIONS
Pregnancy
Risk Summary
There are no adequate data on the developmental risks associated with the use of DAYBUE in pregnant women. No adverse developmental effects were observed following oral administration of trofinetide to pregnant animals at doses associated with plasma exposures below those used clinically.

Lactation
Risk Summary
There is no information regarding the presence of trofinetide or its metabolites in human milk, the effects on the breastfed infant, or the effects on milk production. The developmental and health benefits of breastfeeding should be considered along with the mother's clinical need for DAYBUE and any potential adverse effects on the breastfed infant from DAYBUE or from the underlying maternal condition.

Pediatric Use
The safety and effectiveness of DAYBUE for the treatment of Rett syndrome have been established in pediatric patients aged 2 years and older. The safety and effectiveness of DAYBUE for the treatment of Rett syndrome in pediatric patients 5 years of age and older was established in Study 1, which included 108 pediatric patients age 5 to less than 12 years of age and 47 pediatric patients age 12 to less than 17 years of age. Use of DAYBUE in patients 2 to 4 years of age is supported by evidence from Study 1 and pharmacokinetic and safety data in 13 pediatric patients 2 to 4 years of age treated with DAYBUE for 12 weeks. Safety and effectiveness in pediatric patients less than 2 years of age have not been established.

Geriatric Use
Clinical studies of DAYBUE did not include patients 65 years of age and older to determine whether or not they respond differently from younger patients. This drug is known to be substantially excreted by the kidney. Because elderly patients are more likely to have decreased renal function, it may be useful to monitor renal function.

Renal Impairment
Mild renal impairment is not expected to impact the exposure of trofinetide; therefore, dosage adjustment is not necessary. Dosage adjustment of DAYBUE is recommended in patients with moderate renal impairment. Administration of DAYBUE to patients with severe renal impairment is not recommended *[see Dosage and Administration]*.

16.2 Storage and Handling
Store DAYBUE in an upright position refrigerated at 2°C to 8°C (36°F to 46°F). Do not freeze. Keep the child-resistant cap tightly closed. Discard any unused DAYBUE oral solution after 14 days of first opening the bottle.

17 PATIENT COUNSELING INFORMATION
Advise the caregiver or patient to read the FDA-approved patient labeling (Patient Information).

DAYBUE Administration
Advise the caregiver or patient that DAYBUE may be given orally or via gastrostomy (G) tube; doses administered via gastrojejunal (GJ) tubes must be administered through the G-port. DAYBUE may be taken with or without food.

Instruct the caregiver or patient to obtain a calibrated measuring device, such as an oral syringe or oral dosing cup, from the pharmacy to measure and deliver the prescribed dose accurately. A household measuring cup is not an adequate measuring device.

Instruct the caregiver or patient to discard any unused DAYBUE after 14 days of first opening the bottle.

Diarrhea
Advise the caregiver or patient that DAYBUE can cause diarrhea. Instruct the patient to stop taking laxatives before starting DAYBUE. If diarrhea occurs, patients should notify their healthcare provider, consider starting antidiarrheal treatment, and monitor hydration status and increase oral fluids, if needed *[see Warnings and Precautions]*.

Weight Loss
Inform the caregiver or patient that DAYBUE may cause weight loss and to notify their healthcare provider if weight loss occurs *[see Warnings and Precautions]*.

Vomiting
Advise the caregiver or patient that DAYBUE can cause vomiting and if vomiting occurs after DAYBUE administration, do not take an additional dose, but continue with the next scheduled dose *[see Warnings and Precautions]*. Instruct patients to notify their healthcare provider if vomiting does not stop despite medical management.

Storage
Keep bottles of DAYBUE oral solution upright and refrigerated before and after opening. Do not freeze *[see Storage and Handling]*.

Marketed by: Acadia Pharmaceuticals Inc.
San Diego, CA 92130 USA

2025 ACMG Annual Clinical Genetics Meeting Highlights

by **Jodi D. Hoffman, MD, FACMG**
2025 Program Committee Chair

Jane Radford, MHA, CHCP
Director of Education

Jane Dahlroth, CEM, CMP
Senior Director of Meetings and Exhibits

Penelope Freire, CMP, DES
Associate Director of Meetings, Exhibits and Digital Events

The 2025 ACMG Annual Clinical Genetics Meeting, held in Los Angeles in March, connected close to 3,300 medical, clinical and laboratory genetics professionals to experiencing the evolution of the specialty – from research to diagnostics to treatments and precision therapeutics. Attendees and exhibitors from around the world engaged in a dynamic exploration of the latest research, leading-edge innovations and impactful clinical advancements through insightful presentations led by topic experts. The meeting featured 49 accredited sessions and welcomed more than 250 expert presenters, further highlighting the depth and breadth of knowledge shared across the program.

The Annual Meeting was a tremendous success on all accounts. The attendance by 3,244 individuals, included 2,580 professional attendees, speakers, press and guests, along with 664 exhibit personnel. Notably, 920 participants were first-time attendees. The meeting welcomed attendees from 45 countries, with international participants comprising 14% of the total audience.

Highlights from the 2025 Meeting

Participants praised the diversity of educational opportunities, the well-organized format and the inclusion of a broad range

of topics that aligned with their varied interests and specialties. Attendee feedback reflected high satisfaction with both the content and overall meeting experience.

Attendees found the independently developed Corporate Educational Satellite Symposia offered by industry to be a valuable part of the overall education and appreciated the valuable insights and discussions on cutting-edge research and clinical advancements in genetics provided in these sessions. The content and speakers in these sessions were highly rated by those who attended.

Highlights of some of the more popular scientific plenary sessions, concurrent scientific sessions and platform presentations are summarized below.

The Presidential Plenary session, "Genetics in the Media – Entertainment, Public Education, Controversies and Ethical Dilemmas" was very well received. Los Angeles was the perfect city for this session. Speakers discussed genetics and science and medicine in several media formats, including television, film, newspapers, magazines and social media. Controversies and ethical dilemmas such as medical misinformation were explored. The role of the medical professional was reviewed as well as the importance of public perception.

With the ongoing integration of genetic testing in non-genetics clinics, participants enjoyed the cardio-genetics and ophthalmologic genetics sessions, learning about organ-specific syndromes and the partnership between genetics professionals and other specialists.

Other sessions wrestled with emerging ideas such as the identification of fetal fentanyl syndrome, cancer predisposition as a continuum, use of sex and gender identifiers in the laboratory setting, N of 1 ASO therapies and the idea of primary screening for some of the current actionable medical issues included in the ACMG list of secondary findings.

Participants learned about emerging laboratory testing methodologies, including long read RNA sequencing, sequencing advances in repeat expansion disorders and optical genome mapping. Additionally, the highly anticipated ACMG/AMP/CAP/ClinGen standards for Sequence Variant Classification were presented, including opportunities to pilot the guidelines prior to their expected release in the coming year.

This year, several especially strong prenatal sessions addressed timely topics such as issues to consider regarding the use of prenatal genome sequencing, possible ways to address the gap in teaching REI genetics, and the latest in cfDNA testing capabilities. An outstanding Genetic Counselor's Forum focused on reproductive counseling for individuals with disabilities, with many attendees highlighting the session's valuable inclusion of the patient perspective - featuring an interview with a cognitively challenged couple who expressed their desire to be heard, counseled and treated like other couples.

The focus on the importance of the patient/community perspective continued with the TED-Style Talks -- "Ethical Considerations in Genetics: Balancing Medical Care and Patient Needs in a Culturally Sensitive Way." During this

session, Dr. Holly Tabor, director of the Stanford Center for Biomedical Ethics, and a leading biomedical ethicist, framed the conversation in terms of the interactions between clinical care professionals and those with disabilities, the disability paradox and culturally sensitive care. Dr. Grace Meier, family medicine physician at the Clinic for Special Children in Lancaster, Pennsylvania, shared insights from her work with the Plain Community, offering guidance on how to build respectful partnerships with populations who maintain strong cultural identities and distinct approaches to illness and medical care. The session concluded with a powerful presentation by Lauren Maucere, deaf educator and curriculum & outreach supervisor at the California School for the Deaf, Riverside. She highlighted both the achievements of the Deaf community and the challenges posed by modern medical approaches, particularly in relation to the use of American Sign Language (ASL) and the preservation of Deaf culture. This session provided thought-provoking perspectives that encouraged all attendees to reflect deeply as they returned to their roles in genetics and genomics.

Opportunities for Young Professionals

The meeting provided many offerings for students and geneticists-in-training. Activities began on Tuesday evening when more than 300 young professionals engaged with ACMG leadership at a lively Welcome Reception.

The "Early Genetics Trainee and Faculty Forum - Clinical Genetics: Where Are We Headed?" featured a panel of experts who shared their insights on the future of genetics care. They addressed key challenges, including how genetics services will be delivered, how the interpretation of genetic testing results ordered by non-geneticists will be supported and who will be responsible for implementing emerging therapies. A lively and engaging discussion followed, centered around these critical questions.

The Early Career Genetic Mentor Luncheon offered the opportunity for more than 200 postdoctoral fellows, clinical fellows and residents to talk informally with senior members of the College about career options, goals and professional opportunities.

The Student Workshop, "Pathways to Careers within Medical Genetics and Genomics," provided medical, undergraduate, graduate, and genetic counseling students with the opportunity to learn more about careers in the field of medical genetics. They had the opportunity to meet with each speaker to gain deeper insights into why they chose their field, receive application advice, as well as make connections for future rotations.

The 3rd Annual GENEius Challenge, an exciting Jeopardy-style game for geneticists-in-training, featured three qualifying rounds in which nine contestants competed for a spot in the Final Championship Round, which was held onstage before the start of the closing plenary session. The winner was Pongtawat Lertwilaiwittaya, MD of the University of Alabama at Birmingham. In addition to a personal trophy, Dr. Lertwilaiwittaya received a cash prize and will have his name added to the GENEius Challenge trophy that will be brought to future Annual Meetings for display.

Special thanks to Drs. Keith Edelman (Program Chair 2023), Jenny King (Program Chair 2024) and Murugu Manickam for curating the questions and supporting and managing this interactive, educational contest, which brought genetics residents and attendees together for some genetics fun!

Exhibit Hall Highlights

When sessions were not taking place, the Exhibit Hall offered several opportunities for attendees to learn and engage. From the Opening Reception, Posters, Learning Lounges, Exhibit Theaters, the GENEius Challenge preliminary rounds and Speed Mentoring sessions, there was something for everyone. Rated highly by attendees, the exhibiting companies and organizations featured the latest advancements and products, therapeutics, and services in medical and clinical genetics and genomics.

With more than 900 posters on display, Poster Sessions offered a valuable opportunity for researchers to showcase their work and engage in discussions with peers. Posters continue to be available for viewing online as ePosters through the meeting app or Annual Meeting website.

Exhibit Theaters were once again a highly rated feature of the overall exhibition. These 30-minute sessions, presented by exhibiting companies, focus on the latest research being

conducted by industry, topics on collaborations with labs and clinical applications of their technologies.

The Learning Lounge featured several informative and interactive sessions, including a Genetics in Medicine–sponsored talk with editors offering insights on how to get published. The ACMG Advocacy Committee provided updates on the regulation of LDTs and other ongoing advocacy initiatives. Attendees also heard what's new from the ABMGG Continuing Certification Program and enjoyed the always-popular Rapid Fire Poster Presentations.

2025 Program Committee Chair Jodi D. Hoffman, MD, FACMG summarized the experience by saying "This year's ACMG meeting touched on ethical and patient care dilemmas, multi-disciplinary partnerships, work-force challenges and emerging technologies. We look forward to planning the 2026 meeting soon!"

Special thanks go to the 2025 Annual Meeting Program and Education Committees for their significant contributions towards the success of this year's meeting. ACMG is grateful to the industry partners that participate as exhibitors and provide support for many of the meeting's services and events.

Future ACMG Annual Clinical Genetics Meetings - Mark Your Calendar!
March 10 – 14, 2026 | Baltimore Convention Center | Baltimore, Maryland

The 2026 ACMG Annual Clinical Genetics Meeting will be held Baltimore, March 10-14. Baltimore – a new destination for ACMG! – is a vibrant community that honors the past while keeping a keen eye on the future. Baltimore is multifaceted and full of surprises, steeped in history (home of the birthplace of our national anthem) and culture, from the gritty graffiti alley to the vast collections at its art museums. Plus, an unexpected food scene and plenty of gorgeous views. Discover more about this scenic waterfront city at Visit Baltimore - baltimore.org

The 2027 and 2028 Annual Meetings will be held in these cities and on these dates:

- April 13 -17, 2027 * Minneapolis Convention Center * Minneapolis, Minnesota
- March 21-25, 2028 * Vancouver Convention Center * Vancouver, British Columbia

2026 | ACMG Annual Clinical Genetics Meeting

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

Save the Dates

ACMG 2026 in Baltimore! Located along an expansive waterfront, today's Baltimore is a vibrant community that honors the past while keeping a keen eye on the future. Fueled by cultural tastemakers and creative entrepreneurs, Charm City is home to a rich history that is constantly evolving.

Education

- Scientific Sessions and Platform Presentations
- Workshops
- Corporate Educational Satellite Symposia
- Posters featuring the latest research

Meeting Features

- Earn credits: CME, P.A.C.E.® and NSGC
- Network with peers and professional associates
- Discover what's new in genetics and genomics
- Exhibit Hall featuring exhibitors, Posters, Exhibit Theaters, Speed Mentoring sessions and Learning Lounge

Mark Your Calendar!

Detailed program, registration and hotel information available:
OCTOBER 2025

Abstract Submission Opens:
OCTOBER 2025

Online Submissions:
www.acmgmeeting.net

#ACMGmtg26
www.acmgmeeting.net

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ACMG Foundation Provides Customized Bicycles to Los Angeles Children with Down Syndrome

Always a highlight of the ACMG Annual Clinical Genetics Meeting, the ACMG Foundation for Genetic and Genomic Medicine hosted the 14th annual Day of Caring on March 21, presenting specially adapted bicycles and helmets to Los Angeles area children from the Down Syndrome Association of Los Angeles. The event coincided with World Down Syndrome Day.

A popular occasion that generated smiles and happy tears, this year's ACMG Foundation Day of Caring was supported by Revvity. ACMG Foundation President Nancy J. Mendelsohn, MD, FACMG and some parents of the children conducted media interviews.

If you missed this heartwarming event, please check the ACMG YouTube channel at youtube.com/theacmgchannel for a highlight video and scan the QR code to read an article from the San Fernando Valley Sun.

bit.ly/sanfernandosun-dayofcaring

ACMG Foundation Presents 10 Awards at the 2025 Annual Clinical Genetics Meeting

ACMG Foundation for Genetic and Genomic Medicine
Better Health Through Genetics™

Each year at the ACMG Annual Clinical Genetics Meeting, the ACMG Foundation for Genetic and Genomic Medicine (ACMG Foundation) honors outstanding individuals through a variety of awards and scholarships. The College and Foundation are pleased to recognize 10 exceptional recipients of the 2025 awards. To read the full announcements, visit acmgfoundation.org under "What We Do."

To learn more about the work of the ACMG Foundation and how you can support its mission, visit acmgfoundation.org or contact acmgfoundation@acmg.net.

Acclaimed clinical geneticist Stephen Cederbaum, MD, PhD, FACMG (pictured with ACMG Foundation President Dr. Nancy J. Mendelsohn) was honored with the 2025 ACMG Foundation's David L. Rimoin Lifetime Achievement Award in Medical Genetics.

The David L. Rimoin Lifetime Achievement Award in Medical Genetics

Celebrated clinical geneticist **Stephen Cederbaum, MD, PhD, FACMG**, has been named the recipient of the 2025 ACMG Foundation's David L. Rimoin Lifetime Achievement Award in Medical Genetics.

A founding member of the Society for Inherited Metabolic Disorders and the American College of Medical Genetics, Dr. Cederbaum is now a distinguished professor emeritus of Psychiatry, Pediatrics and Human Genetics at the University of California, Los Angeles (UCLA). During the height of his career at UCLA, he developed a clinical program in inborn errors of metabolism and, alongside it, became a leading expert on urea cycle disorders, most notably arginase deficiency, a disorder in which the body lacks the final enzyme in the urea cycle and is, therefore, unable to rid itself of excess ammonia. Dr. Cederbaum worked on enzyme therapy for the disorder, which remains a focus of his ongoing research.

"Congratulations to Dr. Stephen Cederbaum for this well-deserved award. Dr. Cederbaum's life and work epitomizes the objective characteristics of the Rimoin award. His passion for teaching, mentoring and care for patients and their families is admirable. He has championed the integration of clinical genomics care into the healthcare system. We are honored to be able to recognize his achievements," said Nancy J. Mendelsohn, MD, FACMG, president of the ACMG Foundation.

Even after 50 years in the field, Cederbaum remains engaged with colleagues and community members. "To me, it's very important [to recognize] that you can always be better, and it's important to make yourself better," he said. "You're never finished with your obligations to society."

The Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award

Monica Wojcik, MD, MPH, FACMG, FAAP is the recipient of the ACMG Foundation's 2025 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award—the "Watson Award"—named for the American College of Medical Genetics and Genomics' first and longstanding executive director, Michael S. Watson, MS, PhD, FACMG.

Dr. Wojcik is a neonatologist and clinical geneticist at Boston Children's Hospital and an assistant professor in pediatrics at Harvard Medical School who specializes in rare diseases affecting the fetus and neonate. Her research focuses on the application of genomic medicine in the perinatal setting, particularly related to genetic diagnosis in the neonatal intensive care unit, understanding genetic causes of perinatal mortality via genomic autopsy, and addressing inequities in rare disease genomics.

The Watson Award recognizes those who have demonstrated innovation in their work and developed or implemented a new concept, method or idea that has had significant impact on genetic and genomic medicine. The award was created to honor the role Dr. Watson played during his nearly 20 years at the helm of ACMG while the field of genetic and genomic medicine emerged and evolved into the far-reaching practice it is today.

"I am incredibly thrilled and humbled to receive this award, and I hope that my career will continue to honor the legacy of Dr. Watson, towards a future where all families can receive the care that they want and need, empowered by genetics and genomics," said Dr. Wojcik.

Monica Hsiung Wojcik, MD, MPH, FACMG, FAAP received the 2025 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award. The award is named for the College's first executive director, Michael S. Watson, PhD, MS, FACMG (right).

Ali H. Bereshneh, PhD (right), recipient of the ACMG Foundation/David L. Rimoin Inspiring Excellence Award, receives congratulations from Michael Rimoin, son of the late Dr. David L. Rimoin.

The ACMG Foundation/David L. Rimoin Inspiring Excellence Award

The ACMG Foundation is proud to present the ACMG Foundation/David L. Rimoin Inspiring Excellence Award to **Ali H. Bereshneh, PhD** for his featured platform presentation at the 2025 ACMG Annual Clinical Genetics Meeting, "Heterozygous De novo variants in *CDKL1* and *CDKL2* cause neuroregressive phenotypes in Human and Drosophila and are dominant negative alleles."

Dr. Bereshneh is currently a postdoctoral fellow in the Department of Molecular and Human Genetics at Baylor College of Medicine. He has been studying rare and undiagnosed disorders using sequencing data, bioinformatic analyses, molecular dynamic simulations and especially animal modeling. The research focuses on demonstrating that variants observed in individuals with rare neurological diseases are at the basis of their phenotypes in order to help end the diagnostic odyssey of these individuals.

"Ali Bereshneh is an exemplar individual for the 2025 ACMG Foundation David L. Rimoin Inspiring Excellence Award. Dr. Bereshneh's work teaches us more about the importance of clinical, research and family collaborators in expanding our understanding of new disorders," said Nancy J. Mendelsohn, MD, FACMG, president of the ACMG Foundation.

"It is a real honor to accept the ACMG Foundation David L. Rimoin Inspiring Excellence Award and I am extremely grateful. I would like to express my deepest appreciation to my mentors, Dr. Hugo Bellen and Dr. Oguz Kanca, our collaborators and the patients and families who participated in our research. Their support has been instrumental in the discovery of unknown human neurological diseases and identification of novel causative variants," said Bereshneh.

The Richard King Trainee Award

Kiely N. James, PhD, FACMG is the recipient of the 2025 Richard King Trainee Award for Best Publication by a Trainee in *Genetics in Medicine (GIM)* for her article, "Genome sequencing detects a wide range of clinically relevant copy-number variants and other genomic alterations," which was published online in *GIM* in January 2024.

Dr. James is currently an Associate Director of Medical Genetics and Genomics and Assistant Clinical Professor at the University of California, San Diego. Her interests include rare

Kiely N. James, PhD, FACMG (right) is the 2025 recipient of the Richard King Trainee Award. Accompanied by Nancy J. Mendelsohn, MD, FACMG, President of the ACMG Foundation.

diseases, somatic mosaicism and emerging genetic diagnostics technologies. “I’m very grateful to the ACMG Foundation and Genetics in Medicine for this award,” said Dr. James. “I hope it can spotlight the value of diligent assessment of our diagnostic tests as they evolve, as one way to improve patient outcomes. The work described in my publication was done in collaboration with many wonderful colleagues at Rady Children’s Institute for Genomic Medicine.”

The Richard King Trainee Award is given by the ACMG Foundation and is named for Dr. Richard King in recognition of his instrumental role in creating *Genetics in Medicine* and serving as the first and founding editor-in-chief of the journal.

The ACMG Foundation Genetic Counselor Best Abstract Award

Jessica Adsit, MS, CGC is the recipient of the 2025 ACMG Foundation Genetic Counselor Best Abstract Award for her platform presentation at the 2025 ACMG Annual Clinical Genetics Meeting, “Next Generation Sequencing in Blastocyst Stage Embryos: Results of over 41,000 Trophectoderm Biopsy Samples.”

A board-certified genetic counselor with 16 years of clinical experience, Ms. Adsit works as a manager of GC Services at Natera, Inc., specializing in preimplantation genetic testing and miscarriage testing. “Thank you so much to the ACMG Foundation for recognizing the work of myself and my colleagues. I am especially honored to receive an award that highlights the various contributions that genetic counselors make to research and clinical care,” she said.

“Jessica’s work highlights the importance of the role of genetic counselors in the field of genomic medicine. Her work underscores the power of data in the field of preimplantation genetic testing as this field expands,” said Nancy J. Mendelsohn, MD, FACMG, president of the ACMG Foundation.

The ACMG Foundation’s Genetic Counselor Best Abstract Award celebrates the contributions and accomplishments of licensed genetic counselors by presenting a cash prize to the genetic counselor judged as having the best abstract at ACMG’s Annual Genetics Meeting.

ACMG Foundation/ Revvity 2025 Early Career Travel Award

Isabelle B. Cooperstein, BS is the recipient of the ACMG Foundation/Revvity 2025 Early Career Travel Award. She was selected to receive the award for her platform presentation at the 2025 ACMG Annual Clinical Genetics Meeting, “SimPheny: Automated Patient Matching for Genetic Diagnosis in Rare Disease Cohorts.”

Ms. Cooperstein is a fifth-year PhD candidate in the Department of Human Genetics at the University of Utah in Salt Lake City. Her research is focused on developing computational tools to enhance the diagnostic process for rare disease patients. “I am honored to receive this award and…am deeply grateful to the ACMG Foundation for their continued support of my career as a trainee, as well as to my mentors and the participants in the Undiagnosed Diseases Network, whose contributions have made this work possible.”

The ACMG Foundation/Revvity Early Career Travel Award helps promising young genetics professionals to cover the costs associated with attending the ACMG Annual Clinical Genetics Meeting. Since 2008, more than one dozen early-career thought leaders in genetic and genomic medicine have been granted this honor.

Isabelle B. Cooperstein (right), a fifth-year PhD candidate at the University of Utah, received the ACMG Foundation/Revvity Early Career Travel Award. Accompanied by Madhuri Hegde, PhD, Revvity’s Senior Vice President and Chief Scientific Officer.

ACMG Foundation’s Next Generation Fellowship and Training Program Awards

Rafael Garrett da Costa, PhD; Atlas Sardoo, PhD; Emily S. Levine, MD, MTM; and Daniel R. Brooks, MD, each received 2025 Next Generation Fellowship and Training Program Awards. These “Next Gen Awards” will support these medical genetics professionals with one year of postgraduate training. Support for this year’s class of fellows was generously provided by Bionano, Pfizer, Sanofi, Spark Therapeutics, and Takeda.

Dr. Rafael Garrett da Costa received a Clinical Biochemical Genetics Fellowship Award and is currently a senior staff scientist at the Department of Laboratory Medicine, Boston Children’s Hospital, Harvard Medical School. For the past five years, his work has focused on inborn errors of metabolism, aiming to bridge innovative diagnostic approaches with patient care. “I am truly honored and excited to receive the Next Generation Fellowship and Training Award from the ACMG Foundation. This award offers a unique opportunity for me to deepen my knowledge in clinical biochemical genetics. I am committed to translating this experience into better patient testing and care,” said Dr. Garrett da Costa.”

Dr. Atlas Sardoo, recipient of a Laboratory Genetics and Genomics Fellowship Award, is currently a scientist at the National Institutes of Health (NIH) specializing in Next-Generation Sequencing (NGS) and translational research. Her work focuses on understanding the role of viral infections in liver cancer and developing risk prediction models. “I am deeply honored to receive the Next Generation Award. This recognition motivates me to continue advancing my work in clinical genetics and to make meaningful contributions to patient care through genomic research,” she said.

Dr. Emily S. Levine received an Ophthalmic Genetics Specialty Fellowship Award. She earned her medical degree with research honors from Tufts University School of Medicine, during which she dedicated an additional year as the Optical Coherence Tomography Research Fellow at the New England Eye Center. Dr. Levine currently serves as a chief ophthalmology resident at Dartmouth Hitchcock Medical Center and will begin a clinical fellowship at the Casey Eye Institute of Oregon Health and Science University in Portland this summer. “It is an immeasurable honor to receive the Next Generation Award and serve families affected by genetic eye conditions,” said Dr. Levine.

Dr. Daniel R. Brooks, who received a Medical Biochemical Genetics Subspecialty Fellowship Award, currently works as a research scientist at the Department of Pediatrics, UPMC Children’s Hospital of Pittsburgh. His contributions span research on prenatal genetic diagnostics, rare disease identification and expanding genetic services to underserved populations. “I am grateful for the ACMG Foundation Next Generation Fellowship award and the opportunity to be recognized among such distinguished peers,” said Dr. Brooks. “My work is rooted in the belief that genetic medicine should be accessible to everyone, not just a privileged few. This recognition reaffirms my mission to bridge diagnostic gaps and bring the benefits of genomic advances in inborn errors of metabolism to underserved communities.”

“These four recipients of the 2025 ACMG Foundation Next Generation fellowship awards truly exemplify the future of our field. These clinician scientists are the best and the brightest, and are doing work that is making significant impact, moving the field of genomics to the next level. I am indebted to their work ethic and enthusiasm,” said Nancy J. Mendelsohn, MD, FACMG, President of the ACMG Foundation.

Genetic Testing, Patient Education and Support: Recent Research from *Genetics in Medicine Open*

by **Morgan Richardson, PhD, MFA, Assistant Managing Editor of *Genetics in Medicine*** and ***Genetics in Medicine Open* and Bo Yuan, PhD, FACMG, Editor-in-Chief of *Genetics in Medicine Open***

In every area of the medical profession, educating patients is an essential part of care: providing context and information about how bodies work, how symptoms are interpreted, what test results mean and how conditions are managed naturally accompanies diagnosis and treatment. However, for healthcare professionals working in genetics and genomics, this requirement often goes further. Beyond the baseline task of explaining what genes are and how they function, there is added the complex and delicate next step of helping patients navigate that daunting question: how might taking the step to seek genetic testing shape their medical care moving forward?

In several articles published in Genetics in Medicine Open within the last year, different teams of authors have presented work on this question. The answers are always reliant on context in a variety of ways: does the patient have reliable access to health care? Does the rare condition of a potential diagnosis have available treatment options? Is early diagnosis associated with preventive care that can forestall more severe symptoms? Each patient with the possibility of learning more about their own genetic code faces an intricate matrix of reasons as to why seeking a diagnosis might be desirable and beneficial, and they look to their care providers for customized and up-to-date guidance on how to make the best possible decisions for themselves and their families.

In "Feasibility of an electronic patient-facing cancer family history tool in medically underserved populations" by Feigelson et al., the authors explored the use of a family history collection tool designed for use in primary care settings in a population with barriers to access. A unique element of the implementation of this tool was that "[b]ilingual staff were available to assist participants in completing the family history assessment if needed", and "all participants were immediately presented with a summary of their risk assessment results and next clinical steps" upon completion. The direct access to staff assistance and the provision of quick results not only increased the completion rate for this tool, but also meant that even participants who struggled with the process at certain points "endorsed the tool as easy to use." The discovery that customized and personalized support allowed more patients to complete the process (rather than giving up partway through) offers useful guidance to others seeking to build similar programs. Another digital health tool was described in the article "Genetics Adviser: The development and usability testing of a new patient digital health application to support clinical genomic testing" by Clausen et al., where the authors highlighted the importance of delivering "pre-test education, counseling, and post-test return of results adaptable to any population, test platform, and setting." The authors emphasized the way that "patient-facing digital tools can deliver genetics education asynchronously before meeting with a clinician, giving patients the ability to access necessary genetics educational and counseling content in the location and the time of their preference." In the case of this tool, feedback from participants revealed that the educational element of the experience was particularly valued, with users reporting that "they found the tool user-friendly and the content to be informative and easy to comprehend." In settings where an individual clinician may have limited time and expertise to spend one-on-one with each patient talking about genetics, this type of resource may provide a unique solution.

In "Pilot implementation study of a default genetic referral process for patients with early-onset colorectal cancer," Lau-Min et al. reported on a default genetic referral process for patients diagnosed with colorectal cancer between ages 40 and 49, where the researchers conducted exit interviews with a sample of patients who had undergone genetic testing as part of their cancer journey. One patient described the dual impulses to both want more information about a diagnosis, while also potentially feeling too overwhelmed to seek it out of their own accord: "I think if they [my oncologist] hadn't mentioned it, I probably wouldn't have thought of it [genetic testing] or asked for it… Your brain is swimming with so many thoughts around your diagnosis and your treatments, and the concept of getting more information to be better prepared for potential other cancers or to talk to your family about–it didn't even enter my mind…It probably wouldn't have happened if they hadn't recommended it." For patients in difficult circumstances, the momentum to seek testing on their own is understandably often absent, and this model of providing resources and education about the process might remove many barriers to testing.

Finally, one study explored not only the reasons why a person might agree to genetic testing, but the reasons why some parents might ultimately decide not to seek results for their child. In "Frequency and reasons that parents decline genetic testing for critically ill neonates" by Callahan et al., the authors "conducted brief interviews with parents who declined testing" to learn more about what influences that decision under particularly difficult circumstances. In these interviews, 83% of parents who declined testing responded that "the testing felt irrelevant to the problems they saw as most important," often because "parents believed the testing was unlikely to change their child's current treatment or ultimate outcome." The authors later wrote that "[p]arents' ability to clearly articulate their reasoning and their willingness to share their rationales, as reflected in a high consent rate for this study, suggest that declining reflects thoughtful analysis rather than misunderstanding." As clinicians work to educate patients about the potential benefits of genetic testing, this study offers an important reminder that patients who opt out of such testing are not necessarily making such a choice due to being uninformed, but rather that the decision is often being made from a position of both knowledge and nuance.

These papers are among many studies underscoring the critical role of personalized education and support in helping patients navigate the complex decision of whether to pursue genetic testing and information, particularly in diverse and challenging contexts. Together, these insights advocate for tailored, patient-centered approaches in genetics and genomics to enhance care and decision-making outcomes.

Meet the New CEO and Associate CEO of ABMGG

by **Azra H. Ligon, PhD, FACMG, Chief Executive Officer** and **Darrel Waggoner, MD, Associate Chief Executive Officer, American Board of Medical Genetics and Genomics**

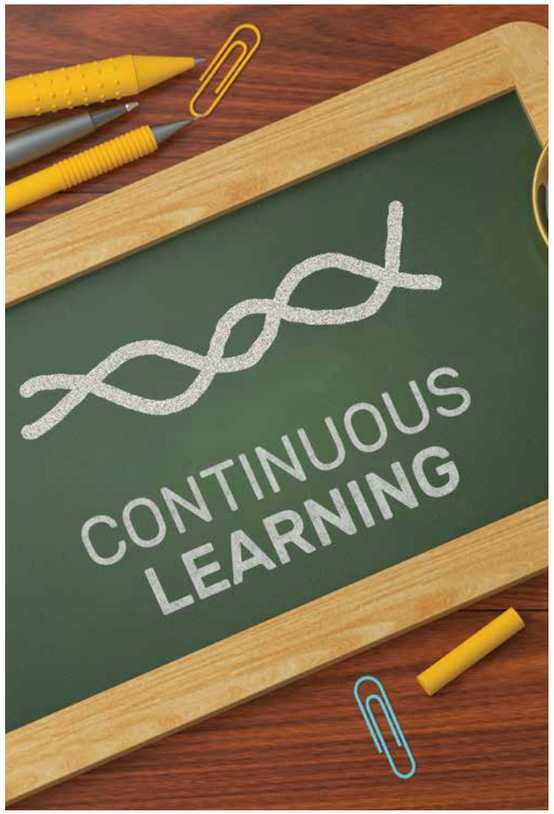
The last few months have been a whirlwind of activity as we both began formal onboarding for our new roles with the ABMGG. We are grateful to former CEO Dr. Mimi Blitzer for the countless hours she spent with us, and to the staff for keeping the ABMGG functioning smoothly during this transition. In anticipation of working with ACMG colleagues, we are excited to briefly introduce ourselves.

Dr. Ligon (CEO) received her PhD in Cancer Genetics from the University of Texas MD Anderson Cancer Center in Houston, trained in Clinical Cytogenetics at Baylor College of Medicine, and completed a postdoctoral research fellowship at Brigham and Women's Hospital (BWH) in Boston. She joined the Clinical Cytogenetics faculty at BWH, served as program director for the Clinical Cytogenetics Fellowship, and later was appointed division chief. Dr. Ligon was elected to the ABMGG Board of Directors and was involved in developing the Laboratory Genetics and Genomics specialty. Her tenure included two years as chair. She was the ABMGG representative to the ACGME Review Committee in Medical Genetics and Genomics when accreditation of laboratory specialties transitioned to this organization.

Dr. Waggoner (Associate CEO) received his MD from Washington University in St. Louis, completed pediatric residency at the University of Chicago and Clinical Genetics training at Washington University. He joined the Department of Human Genetics at the University of Chicago as medical director. Dr. Waggoner was elected to the ABMGG Board of Directors, where he led the development of continuing certification programs, served as the treasurer, and later as the Board representative to the ACMG Continuing Certification Committee.

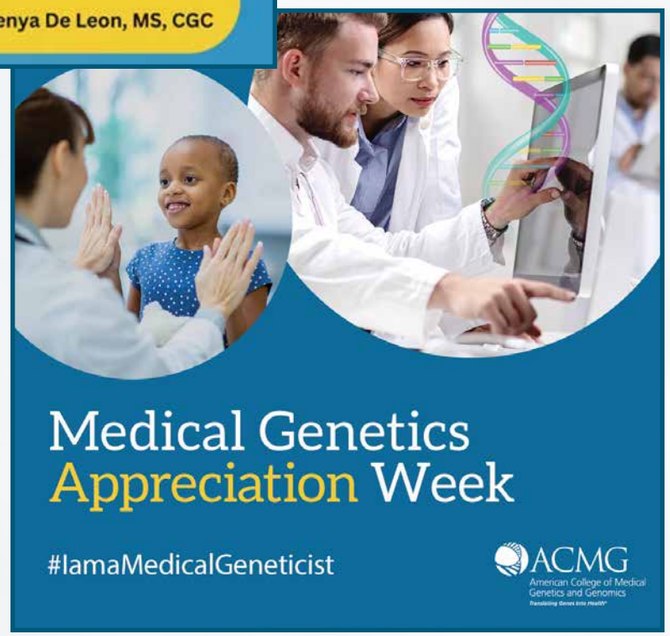
Over the last three decades the ABMGG and the ACMG built a relationship that led to many productive collaborations. These included work with the ABMGG, the ACMG Continuing Certification Program Committee and the ACMG Education Committee to create practice improvement modules; expanding opportunities to earn continuing medical education (CME) credits; selecting articles for CertLink; and developing patient safety modules. We both commit to building upon this foundation, including by advancing Competency Based Medical Education for trainees and diplomates. Another shared goal is the formation of focused practice designations, which will require building medical genetics curricula for physicians whose primary practice interfaces with our own.

As we both settle into our respective ABMGG roles, we look forward to additional opportunities to work with the leadership and members of the ACMG.

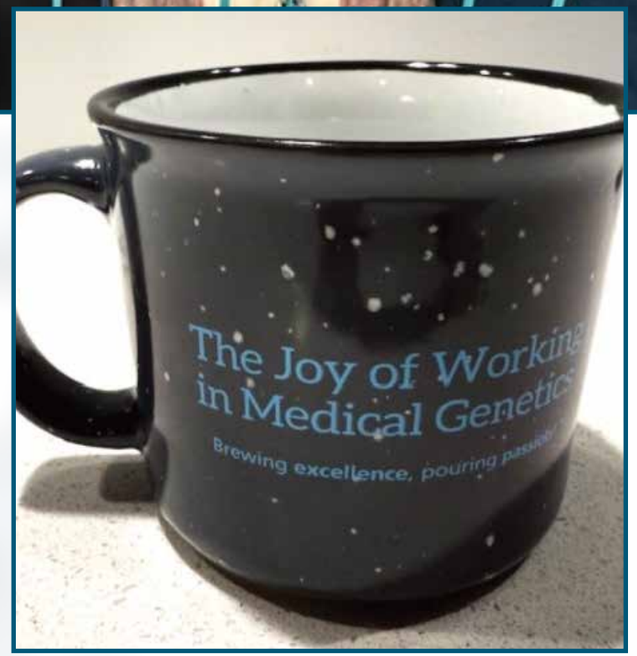


Seventh Annual Medical Genetics Awareness Week Celebrates Joy

The seventh annual Medical Genetics Awareness Week, held March 18-21, 2025, was a successful celebration of the medical genetics field, thanks to ACMG members and the global medical genetics community. We loved the enthusiasm focused on raising awareness of the importance of genetics in medicine. This year's theme, "The Joy of Working in Medical Genetics," highlighted the deep appreciation medical genetics professionals have for their patients and colleagues, as well as the excitement that comes from tackling complex challenges.



For the third consecutive year, we gave away a limited-edition ceramic mug at the ACMG Annual Clinical Genetics Meeting. This year's design was emblazoned with "The Joy of Working in Medical Genetics" and attendees waited in line to receive one to add to their collection and share their medical genetics pride at home and at work! Our "What Brings You Joy Working in Medical Genetics?" wall was filled with attendees' inspirational hand-written messages, and countless photos were taken in front of our Medical Genetics Awareness Week selfie wall. (Story continues after photos.)



A special thank you goes out to the ACMG Board Members, the Community, Outreach and Engagement Committee, the *Genetics in Medicine* and *GIM Open* Editorial Boards, several committee chairs and staff members who contributed social media posts or videos.

Next year, the eighth annual Medical Genetics Awareness Week will take place March 10-13, 2026. But why wait until then to promote medical genetics awareness? We invite you to visit our Medical Genetics Awareness Week web pages at acmg.net/MedicalGeneticsAwareness to find tools and tips to help you promote awareness all year long.

While social media engagement from across the US was most prevalent, we enjoyed seeing posts that displayed the spirit of the week from around the world and from as far away as Argentina, France, Germany, India, Indonesia, Spain, Uganda and the United Kingdom. Across X, Facebook, Instagram and LinkedIn, the #MedicalGeneticsAwareness hashtag earned more than 1.7 million impressions.

Mentoring Opportunities Galore at the ACMG Annual Clinical Genetics Meeting

by **Lauren B. Carter, MD, FACMG, Chair** and **Lauren Thompson, DO, FACMG, Vice-Chair, Education** and **Professional Development Committee**

Our newly named committee (formerly called the Education and CME Committee) had another successful year! Our group remained heavily involved with education and mentoring activities at the ACMG Annual Meeting, including hosting the Early Career Genetics Trainee and Faculty Forum, Genetic Mentor Luncheon and Student Workshop. These events have been well received, and we look forward to continuing these sessions at future meetings.

We hosted our 2nd annual Speed Mentoring Event this year as well. A few weeks before the meeting, all registered geneticists-in-training were invited to participate and make appointments to meet with mentors of their choosing. This year we had more than 40 mentors and 70 mentees sign up, leading to more than 230 appointments! We look forward to continually improving the process for this event and appreciate mentors offering their time on short notice for eager trainees who did not sign up in advance.

Our other accomplishments this past year include assisting ACMG Education with the digital course evaluations and analysis. We have tabulated the results and will publish a report on the feedback received about the digital courses. We also started assisting in the Education Webinar series. We have an ongoing list of scholarly projects that we are eager to implement including assessing needs for genetics residency education. Our Committee continues to coordinate the ACMG Genetics Monthly Challenge and contributes questions for the GENEius Challenge at the ACMG Annual Meeting. We are excited to continue ACMG's commitment to education and mentoring in our amazing field!

Engaging Future Leaders in Genomics: E3 Genomics Pathways Program — Phase 1 Recap

During the 2025 ACMG Annual Clinical Genetics Meeting in Los Angeles, the E3 Genomics Pathways Program welcomed local high school and college students to the Genetics Learning Hub for the inaugural Phase 1: Engage — a thoughtfully designed introduction to careers in genetics and genomic medicine. This immersive experience offered students a unique opportunity to connect with experts, explore real-world case studies and gain valuable insight into the collaborative nature of the genetics field.

The morning session began with a warm welcome from ACMG CEO Melanie Wells, MPH, CAE, who greeted the students and introduced them to the program. The session then began with an interactive survey designed to assess the students' baseline knowledge of genetics. Scholars were subsequently introduced to a panel of accomplished professionals representing a broad spectrum of roles within the genetics ecosystem, highlighting the diverse career pathways available in the field.

One of the morning's highlights was a case study presentation led by ACMG member, Pedro A. Sanchez-Lara, MD, MSCE, FACMG, FAAP, Chair of the E3 Program Committee. Dr. Sanchez-Lara serves as the director of pediatric clinical genetics at Cedars-Sinai Medical Center and associate clinical professor of pediatrics at the David Geffen School of Medicine at UCLA. His presentation centered on the diagnosis and care pathway for Down Syndrome (Trisomy 21). Students traced the clinical process from initial consultation with an obstetrician and genetic counselor, through laboratory analysis and bioinformatics, to clinical care and research teams — illustrating the highly collaborative, multidisciplinary nature of genomic medicine.

Following the case study, Scholars transitioned to the Exhibit Hall, where they participated in a series of enriching activities, including:

- The Day of Caring (see article, p. 22), with a special emphasis on Down Syndrome awareness.
- A Poster Walk highlighting ongoing research, laboratory innovations and clinical applications, where students engaged directly with poster authors and explored current developments in the field.
- A social media-based activity encouraging students to capture and share moments from their Exhibit Hall experience, designed to promote observational learning and peer-to-peer knowledge exchange.

At midday, students had the opportunity to observe the opening round of the GENEius Challenge, an academic competition celebrating problem-solving and collaboration in genetics.

The learning experience continued over lunch with a "meet-the-experts" speed networking session. During this structured exchange, students engaged with a diverse group of professionals to discuss academic pathways, career opportunities and real-world experiences. More than 20 ACMG members volunteered to connect with the E3 Scholars.

In the afternoon, the program offered tailored sessions designed to meet the distinct developmental needs of high school and college students:

- College students participated in the 2025 ACMG Student Workshop: Pathways to Careers within Medical Genetics and Genomics, where they explored specialized career pathways and professional development strategies, while continuing to network with experts in the field.
- High school students engaged in hands-on, didactic activities reinforcing key genetic concepts introduced earlier in the day, including a DNA card game and small-group reflections.

The program concluded with a joint reflection session, where college students met with second-year medical students to discuss academic pathways, professional challenges and perspectives on the future of genetic medicine.

Throughout the day, more than 40 ACMG members actively engaged with the E3 Scholars, demonstrating the organization's strong commitment to fostering the next generation of professionals and leaders in genetics and genomics.

The E3 Genomics Pathways Program — Phase 1: Engage successfully sparked curiosity, encouraged professional exploration and fostered meaningful connections between Scholars, mentors, and experts. Building on this strong foundation, Phase 2: Equip and Phase 3: Empower will be held during future ACMG Annual Meetings.

To learn more about the E3 Genomics Pathways Program, or if you are interested in becoming a mentor, please visit acmg.net/ACMG/Education/E3_Genomics_Pathways_Program.aspx or scan the QR code below.

This project was funded by the National Human Genome Research Institute (NHGRI) through a contract awarded by the National Heart, Lung, and Blood Institute (NHLBI) Contract No. HH75N92024P00281.

ACMG Education in Action: Compliance, Expertise and Continuous Improvement

by **Jane Radford, MHA, CHCP, ACMG Director of Education**

ACMG's nationally accredited education program continues to evolve with purpose and precision—grounded in compliance, driven by scientific rigor, and shaped by the needs of our learners. This three-part feature explores how ACMG upholds the highest standards of independence, addresses complex compliance scenarios, and invites members to actively contribute to the future of trusted continuing education. Together, these efforts reflect ACMG's enduring commitment to excellence in medical genetics education.

Join Us in Shaping the Future: Advancing Integrity and Innovation in ACMG Education

ACMG continues to lead the field of medical genetics education through its nationally accredited Continuing Medical Education (CME) program. With more than 25 years of continuous accreditation by the Accreditation Council for Continuing Medical Education (ACCME), ACMG is committed to providing timely, high-quality and independent education that strengthens professional practice and patient care.

During the 2021–2025 accreditation cycle, ACMG successfully implemented comprehensive reforms to align with the new ACCME Standards for Integrity and Independence in Accredited Continuing Education. These efforts included streamlining financial disclosure procedures through AAMC's Convey® platform, implementing rigorous peer review and content validation processes, and providing real-time monitoring of live events to uphold scientific balance and transparency.

A core strength of ACMG's program lies in its content. Our education program continues to prioritize case-based, applied learning that resonates with our diverse professional audience. Learners have responded strongly to offerings like the ClinGen Somatic Cancer Series, ACT Sheet Knowledge Nuggets, and our annual Gene Therapy Series, which together have drawn thousands of participants and elevated genomic literacy across disciplines.

To meet growing demand, ACMG expanded its Genetics Academy, launched competency-based programs like the ClinGen Curation Modules, and created new pathways for volunteer involvement through 13 specialty workgroups. These changes have broadened faculty engagement, enhanced member collaboration, and ensured alignment between our programming and the evolving needs of our learners.

Even amidst challenges—such as the loss of federal funding in 2024—ACMG responded by integrating essential resources into our CME portfolio in order to maintain continuity for topics such as newborn screening, policy and counseling.

Feedback from learners confirms our direction is making a difference:
- 94% report greater confidence applying new genetics knowledge in practice.
- 91% say ACMG's CME improves their clinical or laboratory decision-making.

These data reinforce what we hear every day: ACMG education is driving meaningful change.

Call for Members: Join Our Mission to Uphold Scientific Integrity

As we continue to expand our educational offerings, **we are seeking ACMG members with no relevant financial relationships** to support content review and mitigation.

If you
- Are free of disclosures related to ineligible companies, and
- Want to help safeguard the integrity of our accredited education

…then we would love to hear from you!
Reviewers assist with content validation, identify potential conflicts and ensure that our education meets the highest scientific and ethical standards.

Interested? Please contact Jane Radford at jradford@acmg.net. Your contribution will help ensure that ACMG continues to deliver trusted, independent genetics education to clinicians, laboratory professionals and learners across the field.

Together we are building a stronger, more transparent future for genomics in medicine.

Balancing Compliance and Expertise: Upholding Independence in Accredited Continuing Education

Continuing medical education (CME) plays a vital role in improving healthcare delivery, and its value depends on trust—trust that the education is independent, evidence-based and free from commercial influence. To preserve this integrity, the Accreditation Council for Continuing Medical Education (ACCME) has established the Standards for Integrity and Independence in Accredited Continuing Education. A core feature of these standards is the distinction between eligible and ineligible companies, and the clear expectations placed on accredited providers like ACMG.

What Is an Ineligible Company?
An ineligible company is one whose primary business is to produce, market, sell, re-sell or distribute healthcare products used by or on patients. This includes pharmaceutical manufacturers, device makers and diagnostic companies with proprietary products. A biomedical startup becomes ineligible once it initiates a governmental regulatory approval process—such as filing an IND or PMA—even before products reach the market. Subsidiaries of ineligible companies are also considered ineligible, regardless of internal firewalls.

What Is an Eligible Company?
An eligible company does not produce or market patient-facing healthcare products. Examples include academic medical centers, government agencies and labs that do not sell proprietary products.

Why It Matters
Owners and employees of ineligible companies cannot plan, deliver or evaluate accredited education unless they qualify for one of three narrow exceptions:
- The content is unrelated to their company's products
- The content is basic science research only
- They are demonstrating device use without care recommendations

ACMG's Process to Maintain Integrity
To ensure compliance and transparency, ACMG
- **Collects disclosures** using Planstone and AAMC Convey
- **Determines relevance and exclusions** based on company status and content alignment
- **Mitigates conflicts** via recusal, peer review and content restrictions
- **Discloses** all relevant financial relationships and mitigation steps to learners
- **Monitors sessions** in real-time and post-activity for signs of commercial bias

This structured approach ensures that ACMG's accredited activities remain rigorous, independent and aligned with the ACCME's standards of scientific and ethical excellence.

ACMG's Strategy for Content Integrity When Experts Work at Ineligible Labs

In the field of genetics and genomics, many of the most qualified experts work at diagnostic laboratories—some of which are considered ineligible under ACCME's definitions due to their development or sale of proprietary clinical tests. For ACMG, this presents a complex challenge: How do we protect the integrity of accredited continuing education while ensuring that our programming includes authoritative, field-leading expertise?

1. Content Before Speakers
ACMG begins with the educational need, not the speaker. Content gaps drive planning decisions, ensuring topics are selected based on clinical relevance, not personal affiliations.

2. Judicious Use of ACCME Exceptions
Faculty affiliated with ineligible companies may only participate if their role and content fall under one of ACCME's permitted exceptions—such as delivering basic science content or demonstrating technical device usage. These exceptions are applied sparingly and only after detailed review.

3. Use of Non-CME Formats When Necessary
When a speaker's expertise is essential, but their conflict cannot be mitigated, ACMG may offer the session as non-accredited and clearly distinguish it from accredited programming.

4. Peer Review Safeguards
All content is peer-reviewed by individuals with no relevant financial relationships. This ensures scientific rigor and neutrality, even when the content area is highly specialized.

5. Curated Roster of Independent Experts
ACMG maintains a robust pool of faculty and reviewers who are both highly experienced and free of industry conflicts.

This roster is regularly refreshed and expanded to include emerging leaders and early-career professionals.

6. Ad Hoc Review for Complex Cases
When a lab's eligibility is unclear, ACMG convenes an internal committee—including education and compliance leadership—to evaluate the company's structure, funding and business model. This allows consistent, well-documented decisions.

ACMG's dual commitment to the highest standards of integrity and to scientific excellence drives a thoughtful approach that meets ACCME's expectations without sacrificing educational value. Our careful vetting and mitigation procedures allow us to deliver content that is both compliant and credible, ensuring that our learners receive the most accurate and balanced education possible.

Support the ACMG Student Interest Group Program and Empower the Next Generation

by **Yuan Ji, PhD, MBA, FACMG, Chair; Mauro Longoni, MD, FACMG, Vice Chair** and **Wesley G. Patterson, PhD, PA-C**

The ACMG Student Interest Group (SIG) Program needs your support. Designed for medical, graduate and genetic counseling students at host institutions, SIGs provide opportunities to expand their knowledge and grow their interest in the field of medical genetics. The SIG Program is essential for cultivating the next generation of the genetic workforce and future ACMG members, ensuring the excellence and rigor that defines our community.

Early exposure to clinical genetics and active involvement with the College through the SIG Program provides students with a rich deposit of genetics educational resources, opportunities for networking and information about careers in medical genetics. By providing FREE Student memberships and numerous benefits, ACMG is committed to the next generation of genetics professionals.

The majority of ACMG members are affiliated with institutions where a SIG has not been established, presenting many opportunities to get involved and support students as they join the expanding network of SIG communities – a place to connect, grow, and make an impact.

There are many ways for you to get involved in the ACMG SIG Program, such as establishing a local SIG at your university or medical school; engaging in activities of an existing SIG; serving as a SIG advisor; or helping us spread the word!

Please visit the ACMG Student webpage for a list of currently active SIGs and information on how to establish an ACMG SIG.

We appreciate your commitment to the future success of our profession and the welfare of the patients we serve. Please contact us (membership@acmg.net) if you have additional questions about the ACMG SIG Program.

ACMG Foundation Supports Summer Scholars Through Immersive Genetics Training Program

Last summer, the ACMG Foundation's Summer Genetics Scholars Program (SGSP) provided 18 students from 11 institutions with a six-week immersive training program in genetics. Designed for students who have completed their first year of medical school, the program pairs each scholar with a medical geneticist mentor, offering hands-on experience in clinical care.

Five of the 18 participants were also awarded a travel scholarship to attend the 2025 ACMG Annual Clinical Genetics Meeting in Los Angeles. Awardees were selected based on their culminating experience essays. They received complimentary registration for the Annual Meeting, three nights of hotel stay and round-trip airfare to LA

While at the Annual Meeting, the travel scholarship awardees attended scientific sessions, the ACMG Foundation VIP Donor Reception and networked with a wide range of leaders in the field of genetics.

The ACMG Foundation thanks all of our individual donors, as well as BioMarin, Ultragenyx and all past contributors who specifically supported the Summer Genetics Scholars Program. If you are interested in donating to this program, please contact Karl Moeller at kmoeller@acmgfoundation.org.

Pictured from left to right: Emma Nussman, Kathleen Renna, Sarah Caffery (Director of External Medical Engagement at BioMarin), Katherine McDonald, Emma Ellis, Hunter Slosser

Orlando J. Miller, MD

by **Rhona Schreck, PhD, FACMG,** and **Art Brothman, PhD, FACMG**

Orlando J. Miller, MD, known as Jack or OJ, passed away on November 3, 2024, after a brief illness. Dr. Miller was the second president of the ABMGG, an AAAS fellow, an emeritus professor and founding chair of Molecular Medicine and Genetics at Wayne State University.

Jack, who was among the first cohort of medical geneticists in the United States, was born in Oklahoma City on May 11, 1927. He attended Yale University for undergraduate, medical school, and residency training in obstetrics and gynecology after serving for two years as a physician in the US Army's 11th airborne division.

He began his medical genetics training in 1958 with Lionel Penrose at the Galton Laboratory, University College, London. He was introduced to human cytogenetics there, and in collaboration with Penrose and Charles Ford, identified two males with 48 chromosomes: 48,XXY,+21 and 48,XXYY. He also worked with Henry Harris, as an NSF postdoctoral fellow, utilizing mouse/human somatic cell hybrid cells for gene mapping.

Jack went on to serve as Professor of Obstetrics & Gynecology and Human Genetics and Development at Columbia University, where his laboratory focused on chromosome organization, chromosome evolution and gene mapping in both humans and mice.

Together with his faculty colleague and wife, Dorothy Anne Miller (Sandy), Jack produced the first quinacrine banded mouse karyotype. He engaged many graduate students, post-doctoral fellows and junior faculty in these cytogenetic adventures, generating a zoological collection of karyotypes (including guinea pigs, rats, dogs, great apes, marsupials and the Indian muntjac with 6 chromosomes in females and 7 in males), and trips to the Bronx Zoo where he was tasked to determine if the newly acquired orangutang had Turner syndrome.

His long-standing collaboration with Roy Breg at Yale (Roy had demonstrated that the cells that grew from an amniotic fluid sample were of fetal origin) identified additional chromosomal causes of infertility and developmental delay, such as the involvement of Robertsonian translocations in familial Down syndrome. His studies of cancer cell lines demonstrated homogeneously staining regions (HSRs), evidence of gene amplification. The Miller group was the first to assign a gene to a specific autosome, using somatic cell hybrids.

Jack loved to travel and explore the world, doing sabbaticals in Oxford with Henry Harris, Edinburgh with Adrian Bird, and Melbourne with Jenny Graves. He and Sandy were active participants in national and international meetings and were consistent contributors to the Somatic Cell Genetics Meeting (now the American Cytogenetic Conference). In 2008 they were co-recipients of that organization's Distinguished Cytogeneticist award.

Jack was an exemplary mentor and teacher, enormously supportive of his numerous trainees, individuals from many different cultures and backgrounds, encouraging innovative thinking, the adoption of new technical skills and promoting the independent recognition of their achievements. We feel honored to have trained with this kind and creative man. He will be remembered fondly by both his three genetic and numerous scientific children.

New ACMG Publications: Policy Statements, Practice Resources, Clinical Guidelines

To facilitate the delivery of quality clinical and laboratory medical genetics and genomics services, the ACMG— through its Board of Directors, committees and workgroups— publishes policy statements, evidence-based or expert clinical and laboratory practice guidelines and descriptions of best practices in genomic medicine.

The following is a list of the College's recent publications since the Winter 2025 edition of *The ACMG Medical Geneticist*:

ACMG Statements, Guidelines, Technical Standards and Practice Resources

Marco L. Leung, et al. **A primer on regulation of laboratory-developed testing procedures: A points to consider statement of the ACMG**. *Genetics in Medicine* published online May 2, 2025; https://doi.org/10.1016/j.gim.2025.101391.

Patricia L. Hall, et al. **Biochemical testing for congenital disorders of glycosylation: A technical standard of the ACMG**. *Genetics in Medicine* published online February 13, 2025; doi: https://doi.org/10.1016/j.gim.2024.101328.

Tuya Pal, et al. **Management of individuals with heterozygous germline pathogenic variants in ATM: A clinical practice resource of the ACMG**. *Genetics in Medicine* published online December 4, 2024; doi: https://doi.org/10.1016/j.gim.2024.101243.

Wendy E. Smith, et al. **Phenylalanine hydroxylase deficiency diagnosis and management: A 2023 evidence-based clinical guideline of the ACMG**. *Genetics in Medicine* published online December 4, 2024; doi: https://doi.org/10.1016/j.gim.2024.101289.

Focused Revisions and Addenda

Ting Wen, et al. Addendum: **Yield of additional genetic testing after chromosomal microarray for diagnosis of neurodevelopmental disability and congenital anomalies: A clinical practice resource of the ACMG**. *Genetics in Medicine* published online January 28, 2025; doi: https://doi.org/10.1016/j.gim.2024.101335.

New Practice Performance Modules

Analysis of Acylcarnitines

This module is for clinical biochemical geneticists who are responsible for analysis, interpretation, and reporting of acylcarnitine profiles.

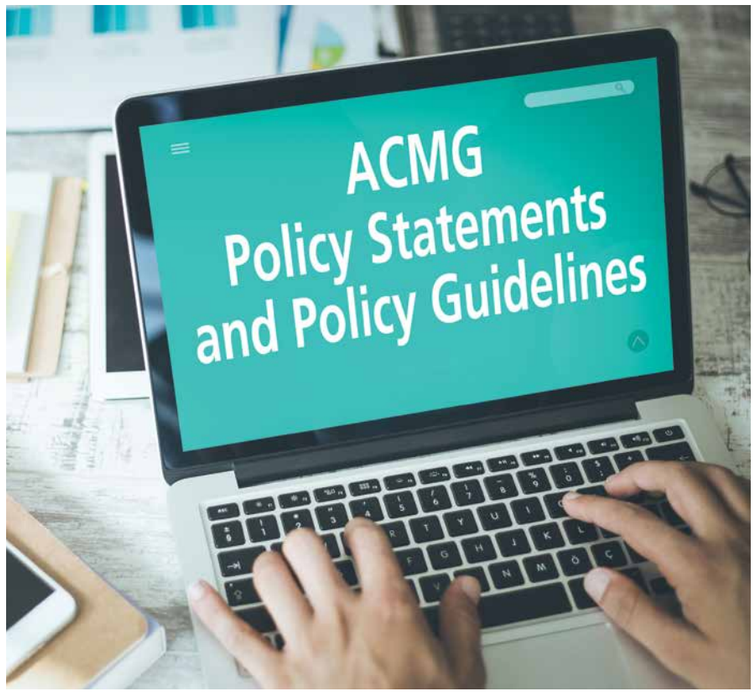
Management of Fabry Disease

This module assesses the appropriate monitoring and clinical management of individuals with Fabry disease according to published guidelines.

Revised Practice Performance Module

22q11.2 Deletion Syndrome (2024 version)

This module is for geneticists who participate in the initial and ongoing care of patients with 22q11.2 deletion syndrome. This does not include patients with known atypical deletions or who have duplications



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