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

Letter From the President

Advocacy is defined as the act or process of supporting a cause or proposal. It was first used in the 14th century tribunal court for one who pleads the cause of another. While public policy is "government policies that affect the whole population," and was first used in the 15th century where policy was considered prudence or wisdom in the management of affairs and may have come from the Middle French word police or *policie*.

As geneticists, we are advocating every day and hopefully influencing policies. We advocate for our patients, for our specialty, for our trainees and for the public. We advocate for our patients by ensuring their counseling, testing and potential treatments are available and, hopefully, covered by insurance. We advocate for our specialty by supporting our patients, trainees and funding of genetic research and new therapies. We advocate for the public by shaping laws related to genetics, supporting genetic education for patients and non-genetics healthcare professionals and addressing ethical concerns related to new technologies – as well as ensuring access to genetics for underserved populations.

In 2015 I started on the ACMG Board of Directors, and around the same time, I was asked to serve on the ACOG District II (NY) Legislative Committee. The Committee reviews proposed NY State laws with the dedicated District office staff and marks up bills for review. In 2016 I heard an Advocacy Grand Rounds given by Dr. Thomas Gelhaus, a former President of ACOG. He discussed all he has done in his life and how ACOG has been influential in their advocacy efforts. He encouraged attendings to apply for ACOG's McCain Advocacy Fellowship. In my letter of intent, I spoke about cases where I had advocated for coverage and reimbursement for genetic testing as a medical geneticist and an obstetrician gynecologist. I described a 70 year-old sister of a patient with a *BRCA1* pathogenic variant who was denied coverage because her insurance was Medicare. I reviewed the importance and availability of *in vitro* fertilization and pre-implantation embryo testing for a couple with a child with Sickle Cell disease who were planning a subsequent pregnancy. And I discussed the inequities of coverage for prenatal screening tests. I was thrilled to be selected and spent two weeks in Washington, DC in April and November of 2018, working with ACOG's

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From the **Editor-in-Chief**

This newsmagazine reflects the College's far-reaching efforts in advocacy. In the Letter From the President (page 1), Dr. Susan Klugman defines advocacy and reviews the many ways geneticists advocate for patients, trainees, the public and our profession. She provides several examples of advocacy efforts from her own illustrious career, including the formation of the Advocacy and Government Affairs Committee (AGA). ACMG CEO Melanie J. Wells (page 11) describes the E3 Genomic Pathway Program, launching in spring, 2025 to address the genomic workforce shortage. Various activities during and following the 2025 ACMG Clinical Genetics Meeting in Los Angeles will engage and mentor future genetics professionals.

ACMG Committees play a huge role in advancing our advocacy efforts. For example, in the past year, the AGA Committee (page 16) employed numerous avenues to keep our membership informed about the FDA actions regarding LDTs, including advising members how to express their concerns to elected officials. At our Annual Meeting, the AGA Committee provides timely sessions on a variety of topics involving services, policies, coverage and more. The Economics Committee (page 18) is concerned with reimbursement for both clinical and laboratory genetic services. They collaborate with other committees – such as Therapeutics and Professional Practice and Guidelines – to establish medical necessity and efficacy of genetic tests. Making sure the correct test is ordered and is medically necessary would certainly help us manage our test load and reduce waste. The Ethics, Legal, and Social Implications Committee (formerly SELI) (page 15) supports our membership with timely publications on expert witness testimony and the use of polygenic risk scores for embryo selection. They recently partnered with the Diversity, Equity, and Inclusion (DEI) Committee on a document regarding biases affecting equitable access to genomic testing and are collaborating with DEI and AMP on a statement concerning the use of sex/gender identifiers in clinical and lab genomics/genetics.

The In Memoriam (page 22) remembers Dr. Vic Riccardi, a brilliant and colorful geneticist who made neurofibromatosis his personal crusade. Finally, please join me in thanking Dr. Mimi Blitzer for her tireless dedication as CEO of the ABMGG and congratulating her on her retirement (page 19). ABMGG has come a long way under her thoughtful leadership and careful guidance, and we greatly appreciate her service!

Until next time,

Katy Phelan

Katy Phelan, PhD, FACMG, Editor
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Government Affairs staff, meeting with lawmakers, attending legislative hearings, and interacting with staff in regulatory agencies. I gained insight into the development of laws and the legislative process.

During my second week, I met with Michelle McClure, PhD, ACMG's Director of Public Policy and Allison Kassir, our Government Relations Advisor. I told them how excited I was to be in DC and work with the staff and how refreshing it was to advocate for patients and hopefully make a difference. I told them it was my dream to bring this type of fellowship to ACMG in the future. We discussed student interns, increasing staff and forming an Advocacy and Government Affairs Committee to help in these efforts.

Late in 2019, a call for volunteers for the newly formed Advocacy and Government Affairs Committee was announced and we formed the Committee with many passionate volunteers, led by Sheila Dobin, PhD as the Chair and Mary Beth Dinulos, MD as the Vice Chair. Dr. Dobin, a former chair and vice chair of the Economics Committee noted, "Statements help us draft our own ideas on legislation or rule making by government agencies… and help the decision makers of health systems on what to prioritize… when resources are limited." Dr. Dinulos also commented that, "It is extremely important that we, as a College, have an active and participatory role in influencing public policies that impact the utilization of genetic and genomic services in healthcare." Michele McClure was the staff liaison and I was appointed Board Liaison. The Committee has reviewed policy issues pertaining to genetics including LDTs, telehealth and the CLIA proposed rule, answered multiple RFIs (Request for Information) from various governmental agencies, had several presentations at our annual meetings and developed several publications, some in the workgroup stage. (The current chair and vice chair, Marco Leung, PhD, FACMG and David Viskochil MD, PhD, FACMG have a Committee update in this issue.) It is hard to believe this Committee has only been active for a little over 4 years! Many of ACMG's advocacy efforts are featured in our publications, on our website and on our social media pages. David Molina joined the team in 2024 as Advocacy Manager and was instrumental in setting up our first Virtual Hill Day where ACMG members met with Congressional staff members telling their stories and advocating for genetics and genomics.

As ACMG's representative to the House of Delegates at the American Medical Association, I am able to advocate for the advancement of healthcare on a national level with multiple specialties. Many of the policies discussed on the floor of the AMA are brought to Congress and congressional members do seek the support and guidance of the AMA. This past year, I was thrilled to introduce a resolution, along with my alternate delegate Jerry Vockley, MD, PhD, FACMG advocating for support of Rare Disease Advisory Councils in every state. Some of the topics we are working on with colleagues across the country include equality in healthcare services, reimbursement, protection of reproductive rights and elimination of pre-authorization. AMA membership has its benefits and I encourage all Fellows to consider AMA membership.

By the end of my ACOG fellowship, I saw the possibilities of genetic coverage for all of the patients in my letter of intent. Since then, multiple bills have been introduced to expand genetic testing, including the Hereditary Cancer Act (Debbie Wasserman Schulz in 2021).

On a personal note, in the airport on my last trip to DC, I spotted then Senator Kamala Harris sitting in a corner waiting for a plane. She had just challenged Brett Kavanagh during his Supreme Court confirmation hearing with a simple question, "Can you think of any laws that give the government the power to make decisions about the male body?" I thanked her for all that she had done and she proceeded to ask me what I was doing in DC. We discussed the advocacy fellowship and my work as a geneticist and obstetrician gynecologist. When we said goodbye, she parted with, "Dr. Klugman, thank you for all you do!" And yes, I do regret not asking for a selfie!

Healthcare is a profession and a daily grind, but the benefits of working on advocacy- related matters can be tremendously rewarding! I hope all of our members feel the same and get to participate in advocacy on a local, regional, national or even international level.

Susan D. Klugman, MD, FACOG, FACMG

Meet the New NIH-ACMG Fellows: Training to Lead Medical Genetics Programs

The NIH-ACMG Fellowship in Genomic Medicine Program Management has been offered since 2017. A two-year opportunity, the fellowship program seeks to increase the number of healthcare practitioners trained to lead research and implement programs in genomic medicine.

NIH-ACMG fellows participate in rotations at the ACMG and the four participating components of the NIH including the National Human Genome Research Institute (NHGRI), National Heart, Lung, and Blood Institute (NHLBI), National Institute on Minority Health and Health Disparities (NIMHD), and the All of Us Research Program. Fellows also design an elective rotation that comprises the final six months of their second fellowship year.

Rachel Nusbaum, MS, LCGC is a genetic counselor with 19 years of experience in clinical genetics, social and behavioral research, variant interpretation and genetic counseling program leadership. She was formerly on the faculties of Georgetown University and the University of Maryland School of Medicine. Through the Alliance to Increase Diversity in Genetic Counseling grant, Rachel coordinated programming to provide full scholarships and stipends to genetic counseling students from underrepresented backgrounds and created a summer internship program for diverse undergraduate students. In anticipation of her NIH-ACMG Fellowship experience, Rachel said, "I am excited to work alongside brilliant people and learn about new resources, while I find ways to utilize my skills to chart new pathways for genetic counselors."

Nicole Thompson, MS, LGC is a genetic counselor specializing in cancer genetics, with more than a decade of experience in clinical and research genetics, program development, community outreach and advocating for underserved communities. She founded and established the first Hereditary Cancer Genetic Counseling Clinic at the Howard University Cancer Center, where the focus of her work was to provide education at the physician and community level, making genetic counseling more accessible and a standard of care for patients in need. Nicole earned her undergraduate degree in Family & Child Sciences at Florida State University and received her master's degree in Human Genetics & Genetic Counseling from Howard. Thinking about her plans as an NIH-ACMG Fellow, Nicole said, "Genetics and genomics are at the forefront of healthcare and I am thrilled to have the opportunity to make meaningful connections, gain mentorship, learn from the best in the field and expand my skill set to further the advancement of genomic healthcare."

Q&A with Our Leadership Team on New Strategic Plans That Will Maximize Success for the College and the Foundation

Both the ACMG and the ACMG Foundation boards of directors have now approved new strategic plans that will synergize and streamline work between the two organizations during the coming five years. Full text of those strategic plans is available to members on the College and Foundation websites. In a recent interview with *The ACMG Medical Geneticist*, Susan Klugman, MD, FACMG, FACOG, who serves as ACMG President and directs the Division of Reproductive and Medical Genetics and the Clinical Medical Genetics Residency at Montefiore Medical Center; Nancy Mendelsohn, MD, FACMG, who serves as President of the ACMG Foundation and is the principal of NJMendelsohn Consulting; and Melanie Wells, MPH, CAE, who serves as CEO for both the College and the Foundation, gave us an overview of these new plans, as well as what members and partners can look forward to next.

ACMG Medical Geneticist (ACMG): Why was this the right time to develop new strategic plans for both the College and the Foundation?

Susan Klugman (SK): A strategic plan is important for every organization because it kind of resets you. It allows you to reflect on your successes from the past years and to prioritize what you want to do in the years coming. The College has had strategic plans in the past. The last one was developed in 2018, and I was fortunate to be on the board and participate in the development of that plan. But genetics and genomics have changed. There are some complicated issues we are facing in taking care of patients and in healthcare in general. We would have liked to roll this out five years after the last strategic plan, but we wanted to be able to gather information in person, and COVID delayed us. Doing this now, in person, allowed us to have deeper conversations than we likely would have had previously on Zoom.

> **"It was good for us to pause and clarify our mission, our vision and our strategy for the years ahead."**

Nancy Mendelsohn (NM): Yes, and this is also a perfect time because we're at an inflection point in terms of planning for growth, with new relationships that can be established within the broader community. It was good for us to pause and clarify our mission, our vision and our strategy for the years ahead. And unlike the College, the Foundation has never really had a formal strategic plan.

Melanie Wells (MW): It was vitally important for us to develop synergistic strategic plans for both the College and the Foundation so we could help our organizations work together to improve general knowledge of the field, awareness of the work that we're doing and to streamline efforts and prioritize what is most important for our members in the field. We also have a completely different leadership model now. For the first time in the history of these organizations, we are led by an all-female team, including me, a non-geneticist and a non-PhD. This means there are new opportunities for our members and our volunteers to engage with the work that we're doing.

ACMG: What was the process for developing these plans?

MW: The process we followed was the same for both organizations. Twenty individuals were identified who had key roles in each distinct organization, and they participated in 30-minute to hour-long interviews with our consultant. The consultant also developed a survey with specific questions for each organization to identify what the respondents' priorities are, and what are the things they feel are relevant for the future, that we should be paying attention to. Those surveys were distributed to 500 people, and we had about a 35 percent response rate, which is great for this type of work. Our consultant evaluated those responses and provided a summary to both boards at the end of February 2024. This served as the foundation for our strategic planning retreat in March 2024, where we spent about nine hours in person going through that information and identifying the priorities. The boards created summary reports that were then reviewed last April by task force representatives from each board, and then the boards voted separately to approve their respective strategic plans.

> **"...we hear you, and we're focusing our efforts on the things that are most important to you."**

SK: As I mentioned, we would have liked to roll out these plans in 2023, but we had to put them on hold because of COVID, and because we had a CEO transition. After a nationwide search, we are thrilled that Melanie is now our CEO. It would have been hard to start the process for developing a strategic plan when she was only a couple of months into her new role, so we waited until January 2024 to begin.

ACMG: Why should readers be excited about these new plans?

SK: Ultimately much of what we talked about during the strategic planning process was how we can benefit our members, whether that has to do with advocacy, helping them take care of their patients, helping them navigate genetic testing reimbursement or preauthorization. We're trying to make the practice of genetics and genomics a little bit smoother and easier. We are a membership organization, and we also want to create ways for our members to get involved, whether it's through a work group or committee, presenting at a meeting or submitting a proposal for a session at a meeting.

NM: On the Foundation side, there is such a large opportunity in the broader medical community, as well as with corporations, insurers, payors, really with everybody, to have a clearer understanding of who we are and of our leadership role in the field of genetics and genomics. One goal of the Foundation is to make a clearer, more effective voice for all of us. We know what we do, we know that we are the experts and there's an opportunity for us to let everyone else know and for us to lean into that.

MW: I would just add that on the Foundation side, the strategic plan will provide clear information for representatives to share with their corporate leadership. They will be able to easily show how, if a corporation partners with ACMGF, they can help make a difference and benefit patient populations that we are committed to serving. Then, on the College side, where the focus is on benefiting our members, we will be able to provide meaningful opportunities for them to engage, to help ACMG and help the field of medical genetics and genomics. Those are different target audiences, but the message is shared: You have a role here, and this is how you can help us make things better for the populations that we serve. We've also built indicators into these strategic plans so that we can continue to report back to our members. We want them to know that this is an organization that you should be happy to be a part of. There's a place and a home for you here, and we hear you, and we're focusing our efforts on the things that are most important to you.

ACMG: How will the strategic plans be implemented?

MW: The next phase will be developing implementation plans with our senior staff so we can break things down on a quarterly basis during the next five years to identify the goals each board

has established, the resources we need to pursue those goals and the partners we need to strengthen our relationships with to meet the objectives for those goals. After we create the implementation plans, we will create a budget, and then we will be prepared to launch the plans in January of 2025. Thereafter, we will include consistent updates for our Foundation donors, sponsors, and corporate partners during quarterly roundtable discussions to let them know how we are performing on the things we said we would do in terms of the specific pillars of our strategic plan. And we encourage College members to pay attention to the e-zines as they come out, because there will be updates on our progress, with opportunities for them to participate.

NM: Everything will be included in our quarterly and end-of-year reports that will refer to the accomplishments as they relate to the pillars of the strategic plans.

ACMG: What can members look forward to next?

NM: For the Foundation's rollout, the first step is to look for people who can help us raise money starting in 2025. We would appreciate any kind of help we can get. If somebody wants to partner with us to reach corporations, charitable organizations, and individuals, please let us know. It's an opportunity for both our PhDs and our MDs, and we will provide training for those who would like to work with us. We also have scholarships available to College members, whether they are medical students, medical residents, or young investigators.

MW: Again, our members need to watch for the monthly *ACMG in Action* e-zines because those will include opportunities to get involved, even this fall, and particularly as we start planning activities for our 2025 annual meeting in Los Angeles. One of the initiatives for the meeting that I am particularly excited about relates to an issue that the College has been involved with for a number of years, which is the expansion of the workforce. How do we do that? Where should we start? Susan and I have participated in calls with NHGRI and a number of other groups to discuss this issue, and what we have found is that there is a population currently untapped in this effort: undergraduate and community-college students. At the 2025 annual meeting, we will be able to utilize an in-place program that funnels high school students and community college students from surrounding communities to STEM-specific meetings at the Los Angeles Convention Center. We have already planned ways to include these students in our meeting, with hands-on didactic activities and our Day of Caring. We also have a "Genetics Challenge" planned where fellows in training will participate in something like the game *Jeopardy*, and we'll pair our Summer Genetics Scholars with students during lunch so they can give them their "why": Why did I choose this field? What drew me here? What have I learned since I got started? Our goal is to help our members engage with the students and show them that there's a place for them here, whether they decide to become a clinical geneticist, a laboratory geneticist, a genetic counselor, or whether they decide to participate in industry.

SK: We don't want to get too much into the details of the strategic plans here, because members can read those details for themselves online or in this edition of their member newsmagazine. But I would just add that a key finding from the survey used to develop this plan for the College was that our members want more opportunities to network. There will be no better way to network in the field of genetics and genomics than by getting involved with ACMG and the ACMG Foundation as we roll out these new strategic plans in 2025.

> **"There will be no better way to network in the field of genetics and genomics than by getting involved with ACMG and the ACMG Foundation as we roll out these new strategic plans in 2025."**

ACMG 2025 – 2029 Strategic Plan Pillars

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

1. **Establish ACMG as the primary genetics and genomics educational and informational resource.**
2. **Define and promote ACMG's role as a leader in the genetics and genomics specialty.**
3. **Advocate for policies and initiatives that advance the interests of genetics and genomics.**
4. **Expand the workforce in genetics and genomics.**

ACMG Foundation for Genetic and Genomic Medicine 2025 – 2029 Strategic Plan Pillars

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

ACMG Foundation Mission Statement

The ACMG Foundation for Genetic and Genomic Medicine is committed to advancing the field of medical genetics and genomics by providing philanthropic funding to support the priorities of the American College of Medical Genetics and Genomics.

1. **Establish initiatives to support ACMG strategic planning priorities.**
2. **Expand, maintain, and maximize current funding streams to support ACMG.**
3. **Enhance visibility of the Foundation and its relationship to the College.**

DAYBUE™ (trofinetide) is the first and only FDA-approved treatment for Rett syndrome (RTT), a complex neurodevelopmental disorder with multisystem impact[1-3]

Daybue™ (trofinetide)

In a 12-week clinical trial, changes in signs and symptoms of RTT were evaluated, including[1]:

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

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 of mild or moderate severity in 96% of cases. In the 12-week study, antidiarrheal medication was used in 51% of patients treated with DAYBUE. Patients should 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. Interrupt, reduce dose, or discontinue DAYBUE if severe diarrhea occurs or if dehydration is suspected.
  - **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.

Statistically significant improvements in signs and symptoms of RTT were seen with DAYBUE[1]

The pivotal Phase 3 LAVENDER™ trial (NCT04181723) was a 12-week, randomized, double-blind, placebo-controlled study that evaluated the efficacy and safety of DAYBUE in 187 female patients (aged 5 to 20 years) with RTT. Two co-primary endpoints evaluated changes in signs and symptoms of RTT, as assessed by caregivers and clinicians.[1,4,a]

Caregiver completed scale[1]

Co-primary Endpoint: Change from baseline to Week 12 in the Rett Syndrome Behaviour Questionnaire (RSBQ) total score. Lower score reflects lesser severity in signs and symptoms of RTT. The maximum possible score is 90.

Results:

- LSM change from baseline (SE) to Week 12 was -4.9 (0.94) for DAYBUE and -1.7 (0.90) for placebo. The LSM placebo-subtracted treatment difference (drug minus placebo) was -3.2 (95% CI: -5.7, -0.6; *P*=0.018)
  - Mean baseline RSBQ score (SE) was 43.7 (1.21) for DAYBUE and 44.5 (1.26) for placebo
- **Almost 3x greater mean score reduction** from baseline vs placebo

Clinician completed scale[1]

Co-primary Endpoint: Clinical Global Impression-Improvement (CGI-I) score at Week 12. A decrease in score indicates improvement.

Results:

- Mean score (SE) of 3.5 (0.08) compared with 3.8 (0.06) for placebo. The LSM placebo-subtracted treatment difference was -0.3 (95% CI: -0.5, -0.1; *P*=0.003)

[a]Patients had a diagnosis of typical Rett syndrome with a documented disease-causing mutation in the *MECP2* gene. Patients were randomized to receive DAYBUE (N=93) or matching placebo (N=94) for 12 weeks.[1]

CI=confidence interval; LSM=least squares mean; SE=standard error.

Demonstrated safety and tolerability profile of DAYBUE

Adverse reactions seen in at least 5% of patients treated with DAYBUE and at least 2% greater than placebo in LAVENDER 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%).[1]

Important Safety Information (continued)

- **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 moderate or severe renal impairment.

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

Please see the accompanying Brief Summary of Prescribing Information on the following page.

Discover an opportunity to spark meaningful improvements in your patients with RTT.

Learn more at **DAYBUEhcp.com**

References: 1. Acadia Pharmaceuticals Inc. DAYBUE [Package Insert]. San Diego, CA, 2023. **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.** Fu C, Armstrong D, Marsh E, et al. Consensus guidelines on managing Rett syndrome across the lifespan. *BMJ Paediatr Open*. 2020;4(1):e000717. **4.** Neul JL, Percy AK, Benke TA, et al. Design and outcome measures of LAVENDER, a phase 3 study of trofinetide for Rett syndrome. *Contemp Clin Trials*. 2022;114:106704.

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CEO CORNER

ACMG will accelerate efforts to Engage, Equip and Empower tomorrow's genomic workforce through the E3 Genomic Pathways Program, a pilot launching March 2025 to address critical worker shortages amidst higher patient demands by engaging community college and undergraduate students pursuing STEM degrees.

ACMG's Board of Directors has prioritized developing and diversifying the genomic workforce pipeline in our 2025-2029 strategic plan. We're pursuing this work in response to documented studies that demonstrate patient demand outweighs the workforce's capabilities as the number of geneticists has remained constant while wait times and new patient caseloads have increased.[1]

The E3 Genomic Pathways Program pilot will help broaden the genomic workforce and enhance our capabilities by developing talent that better reflects diverse patient populations. It's well-documented that a diverse workforce can enhance health care providers' cultural competence. This is an imperative to foster trust and engagement with patients from historically marginalized communities that ultimately improves patient outcomes.[2]

"The E3 Genomic Pathways Program pilot will help broaden the genomic workforce and enhance our capabilities by developing talent that better reflects diverse patient populations."

Join us to launch the E3 Genomic Pathways Program pilot on March 21, 2025, at the ACMG Clinical Genetics Annual Meeting in Los Angeles. The E3 Genomic Pathways Program pilot goals are to:

- enhance historically marginalized students' understanding of clinical and laboratory genetics and genomics;
- engage students in targeted career development activities and real-world applications of clinical genetic principles; and
- provide opportunities for students to meet and network with clinical and laboratory genetics and genomics professionals.

During the Annual Meeting, pilot participants will engage in didactic activities, a mentoring luncheon and a guided poster walk. After the meeting, students will be invited to monthly virtual experiences that are developed for all ACMG students and Student Interest Group members. Pilot participants will also attend quarterly virtual mentoring sessions to help students navigate challenges that they may encounter during their academic journey.

Get involved in supporting the next generation of the genomics workforce. Email e3program@acmg.net with your name, area of expertise and how you'd like to support the program as either a leader of a didactic activity, luncheon mentor or virtual mentor during the upcoming year.

Together we will expand the interest in our field and the number of individuals equipped to meet the needs of future genetics patients.

Melanie J. Wells, MPH, CAE
Chief Executive Officer

[1]Maiese DR, Keehn A, Lyon M, Flannery D, Watson M. Current conditions in medical genetics practice. *Genet. Med* 21:1874–1877 (2019). https://doi.org/10.1038/s41436-018-0417-6.

[2]National Academies of Sciences, Engineering, and Medicine. 2022. Improving diversity of the genomics workforce: Proceedings of a workshop—in brief. Washington, DC: The National Academies Press. https://doi.org/10.17226/26478.

2025 Legislative Outlook and the Importance of Member Engagement

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

As the 118th Congress comes to a close, ACMG is looking forward to the 119th Congress starting on January 3, 2025. The 118th Congress has been interesting in that it has passed an unusually small number of pieces of legislation into law compared to previous Congresses. As frustrations have grown, many members of Congress have decided to retire and pursue other opportunities. As of August 2024, 45 House representatives and eight senators had announced that they are retiring or seeking another office at the end of the 118th Congress.

Those retiring included many longstanding members like Representative Anna Eshoo, Chair of the Energy and Commerce Subcommittee on Health, and Representative Cathy McMorris Rodgers, Chair of the full Energy and Commerce Committee. Representative Eshoo has been a fierce supporter of access to healthcare, and Representative Rodgers played a critical role in stopping the controversial Verifying Accurate Leading-edge IVCT Development (VALID) Act from passing in 2023. While there are always unknowns going into elections, this time it is increased by the number of longstanding members leaving Congress. Thus, it is difficult to predict whether the 119th Congress will be more or less productive in passing legislation, especially those related to improving healthcare.

“While members of the US Congress typically have large, full-time staff who help research and inform them about a multitude of issues, state legislators often have little to no staff support. Thus, it is crucial that healthcare professionals engage with their state legislators and establish themselves as a resource for them.”

As a result, state legislation will continue to play an increasingly significant role in healthcare. Genetics professionals are important players in influencing state policies, and ACMG encourages members to familiarize yourselves with your state’s legislature and session schedule. For example, in 2025 many states will be starting a new legislative session. While some sessions run throughout the year, others are extremely short such as Utah, which is only 45 days long. This means that legislation can move very quickly in some states. While members of the US Congress typically have large, full-time staff who help research and inform them about a multitude of issues, state legislators often have little to no staff support. Thus, it is crucial that healthcare professionals engage with their state legislators and establish themselves as a resource for them.

Visit https://www.congress.gov/state-legislature-websites to find your 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. Additional information about engaging with legislators is available on ACMG’s Advocacy Resource Center at www.acmg.net/advocacy.

Lastly, don’t forget to get out and vote! Election day is Tuesday, November 5, 2024. For information on how to register to vote, voter registrations deadlines and other state-specific voting information, please visit https://www.usa.gov/voter-registration.

2023 *ACMG Salary Survey Report* Now Available

ACMG is pleased to announce that the 2023 *ACMG Salary Survey Report* is now available in the Members Only section of the ACMG website.

This biennial ACMG publication has been a vital source of industry salary information for over a decade. The report contains the findings of the web-based salary survey that was conducted in the fall of 2023. The survey queried American Board of Medical Genetics and Genomics (ABMGG)- and Molecular Genetic Pathology (MGP)-certified MD, DO and PhD ACMG members practicing in the United States. The report also includes, where appropriate, comparisons with data from the 2021 salary survey.

> **"ACMG provides the *Salary Survey Report* FREE to ACMG members, as a valuable member benefit."**

As in previous years, the 2023 survey captured targeted demographic information about respondents, information on salary increases and decreases in the current and prior fiscal years, and data on salary as a function of relative value units (RVUs) generated as a medical geneticist. The report includes analyses that stratify the salaries of those working in high-cost areas with those in other areas and an analysis of salaries as a function of time spent performing various responsibilities such as laboratory direction, direct patient care, administration, research, and teaching.

ACMG provides the *2023 Salary Survey Report* FREE to ACMG members, as a valuable member benefit. The report is also available for purchase by nonmembers at a cost of $400 in the publications section of the ACMG Store.

ACMG thanks all who participated in the 2023 survey and welcomes suggestions for future surveys. ACMG also thanks the members of the 2023 ACMG Salary Survey Subcommittee including: Natasha Strande, PhD, FACMG; Miriam Blitzer, PhD, FACMG; Yu-Wei Cheng, PhD, FACMG; Melissa Crenshaw, MD, FACMG; Jinbo Fan, PhD, FACMG; Scott McLean, MD, FACMG; Sheetal Parmar, MS, CGC; Tatiana Yuzyuk, PhD, FACMG, as well as Michelle McClure, PhD and Nataly Schwartz, MS, MBA (ACMG Professional Staff Liaisons).

If you are interested in volunteering to join the 2025 Salary Survey Subcommittee, please contact Nataly Schwartz at nschwartz@acmg.net.

Acadia is New Exclusive Sponsor of ACMG Membership Newsmagazine

ACMG is excited to welcome Acadia, a pharmaceutical company headquartered in San Diego, CA, as the new exclusive annual sponsor of ACMG's popular member newsmagazine, *The ACMG Medical Geneticist*.

Editor-in-Chief Katy Phelan, PhD, FACMG said, "*The ACMG Medical Geneticist* boasts a very high level of readership with 86% of ACMG members reporting they read at least some of each issue. Thank you to Acadia for helping to make our member newsmagazine possible."

Acadia has developed therapeutics for disorders such as Rett syndrome, while Prader-Willi syndrome is among the company's current targets.

Bridging Law, Ethics and Genetics: The ELSI Committee's Milestones and Collaborations

by **Mahmoud Aarabi, MD, PhD, FACMG, Chair** and **Dena Matalon, MD, FACMG, Vice Chair**
Ethical, Legal and Social Issues Committee

ACMG's Ethical, Legal and Social Issues Committee (ELSI, formerly known as SELI) consists of members from diverse backgrounds with expertise in clinical and laboratory genetics, genetic counseling, bioethics and law. The ELSI Committee has addressed emerging and hot topics in medical genetics including recent Points to Consider Statements on providing expert witness testimony for the specialty of medical genetics (2024) and the clinical utility of polygenic risk scores for embryo selection (2024).and stewardship of patient genomic data (2022).

In addition to publications, ELSI prioritizes member education through sponsored sessions at the ACMG Annual Meeting and educational sessions in the ACMG Genetics Academy. An ELSI Education Subcommittee, formed in 2022, has supported the 2024 Annual Meeting session about the "Impact of Abortion Laws on Genetics and Genomics Training & Practice," as well as a Genetics Academy session on "The Ethical Principles Every Geneticist Needs to Know," planned for November 19, 2024.

Our Committee strives to collaborate with other ACMG Committees and professional societies to advocate for ELSI initiatives. We collaborated with the ACMG Diversity, Equity and Inclusion (DEI) Committee on a recently published statement on "Clinical, technical, and environmental biases influencing equitable access to clinical genetics/genomics testing" (2023). ELSI, DEI and the Association for Molecular Pathology (AMP) are currently working on a policy statement on the use of sex/gender identifiers in clinical and laboratory genetics and genomics. ELSI and DEI are also co-sponsoring a session at the 2025 ACMG Annual Meeting to discuss this important topic. We are now working on further collaborations with the ACMG International Outreach and Engagement Committee, American Academy of Pediatrics (AAP) and the Society for Inherited Metabolic Disorders (SIMD).

ELSI appreciates contributions from its committee and workgroup members and support from the ACMG Board of Directors. Our Committee members in 2024-2025 include Mahmoud Aarabi, MD, PhD, FACMG (Chair); Dena Matalon, MD, FACMG (Vice Chair); George Khushf, PhD, MA (Past Chair); Robert Hufnagel, MD, PhD, FACMG (Board Liaison); Perry Chan, PhD, JD, MBA, FACMG; Bimal Chaudhari, MD, MPH, FAAP, FACMG; Tanya Eble, MS, CGC; Alison Elliott, PhD, MS; Patricia L. Gordon, MD, FACMG; Maria J. Guillen Sacoto, MD, FACMG; Samuel Huang, MD, FACMG; Jesse M. Hunter, MS, DSci, FACMG; Shagun Kaur, MD, FACMG; Jaime Lopes, PhD, FACMG; Chaya N. Murali, MD, FACMG; Kathleen Pope, MD, MS, FAAP, FACMG; Asha N. Talati, MD, MS, FACMG; Tatum Vilaboy, MGC (Student Member); Alessandra Sugrañes, MD, FACMG (DEI Liaison); Lynn Fleisher, PhD, JD, FACMG (Ex-Officio Representative); David Molina (Staff).

Annual Highlights of the Advocacy and Government Affairs Committee

by **Marco L. Leung, PhD, FACMG, Chair** and
David H. Viskochil, MD, PhD, FACMG, Vice Chair

The ACMG Advocacy and Government Affairs (AGA) Committee continues to engage members and elected officials on the latest policies that pertain to medical genetics. Composed of laboratory and medical geneticists, the Committee has addressed a variety of topics over the past year, with the most notable being the regulation of laboratory-developed tests (LDTs).

Since the US Food and Drug Administration (FDA) released their proposed rule on the regulation of LDTs in September 2023, and the subsequent final rule in April 2024, ACMG staff and the AGA Committee have kept members informed on this subject through policy alert emails, updates in the monthly *ACMG in Action* e-newsletter, and web postings. To advocate for our members, we submitted official comments to the FDA proposed rule in November 2023. In May 2024, ACMG launched a letter-writing campaign to help ACMG members reach out to their elected officials asking them to support a resolution disapproving the FDA rule. In the same month, the AGA Committee hosted a webinar on the summaries of the FDA Final Rule and Medical Device Regulations. In February 2024, the ACMG Board of Directors and AGA Committee participated in a virtual Hill Day, speaking with congressional staffers on LDT regulation, as well as the Reducing Hereditary Cancer Act and the Expanded Genetic Screening Act.

The AGA Committee also engaged members during the 2024 ACMG Annual Meeting in Toronto. The Committee hosted a Concurrent Session titled "Providing Genetic Services at a National Level – Workforce, Telegenetics, Payer Coverage Policies, and Beyond" and a learning lounge session titled "Genetic Professionals' Experience Engaging with Lawmakers." At the 2025 Annual Meeting in Los Angeles, we will have another Concurrent Session titled "Navigating the Current Landscape in Prenatal Genetics – Coverage, Politics and Laboratory Practice" and a workshop titled "Rare Disease Advisory Councils – State Advocacy for the Rare Disease Community."

As the legislative landscape continues to evolve for the medical genetics field, ACMG staff and the AGA Committee will continue to provide comments to policymakers and elected officials on proposed legislation and policies, while also continuing to engage members through a variety of ACMG communication channels.

Left to right: David Molina; Mary Beth Dinulos, MD; Michelle McClure, PhD; and Marco Leung, PhD.

Mary Beth is now the past chair of the Advocacy and Government Affairs Committee and Marco is the current chair. At the time of this photo they were chair and vice chair.

Medical Necessity in Genetic Services

by **John Belmont, MD, PhD, Chair of the ACMG Economics of Genetic Services Committee**

The Economics of Genetic Services Committee advises the College on appropriate reimbursement for clinical genetic services. We are concerned with both physician consultations and diagnostic laboratory testing. Genetics professionals must monitor changes in coding and new reimbursement policies as they arise both from private payers and state Medicaid organizations. The Committee also helps the College respond to the payer community as evidence for the medical necessity and efficacy of genetics care develops.

Recently, the Economics Committee helped the College review a new framework for establishing medical necessity put forward by the Patient-centered Laboratory Utilization Guidance Services (PLUGS®). Responsible laboratory stewardship is key to maintaining the confidence of payers.

One of our key recommendations was for further refinement in the standards for publication of evidence for both diagnostic efficacy and clinical utility. The Economics Committee can play an important role in facilitating use of materials produced by other ACMG committees, such as the Therapeutics Committee, which is tasked with a systematic analysis of up-to-date treatments for genetic disease, as well as the Professional Practice and Guidelines Committee, which carries out systematic literature reviews and makes recommendations for best practices.

The Economics Committee continues to monitor advances in medical coding. Over the past year the Committee has evaluated new ICD-10 codes for rare genetic diseases and provided support for the College positions on these codes. The Committee is also looking ahead to the adoption of ICD-11 codes. Our attention will be on synonyms and prompts for more accurate automated coding as well as codes and code combinations that might suggest appropriate diagnostic testing for rare genetic disorders.

Looking Back on a Fruitful Partnership

by **Miriam G. Blitzer, PhD, FACMG, ABMGG, Chief Executive Officer**

The ACMG and ABMGG have a history of fruitful collaboration, regularly featured in *The ACMG Medical Geneticist* through contributions by the ABMGG Directors. The first of these columns appeared six years ago, when I was interviewed about the newly established specialty of Laboratory Genetics and Genomics – today a firmly established and successful laboratory specialty. Our field continues to advance at a remarkable pace. Now, as I prepare to step down after a 15-year tenure as ABMGG CEO, I would like to take this opportunity to look back at some of our key accomplishments:

1. **Changing our name to American Board of Medical Genetics and Genomics (2014):** In recognition that "genomics" is part of our medical specialty and our professional identity, both ABMGG and ACMG took the necessary steps to incorporate "Genomics" into their names. For ABMGG, this involved a rigorous process for approval by the American Board of Medical Specialties and its member boards.

2. **Establishing Laboratory Genetics and Genomics (LGG) as a primary specialty (2016):** In response to rapid technological advancements and following careful deliberations within our community, LGG was established as a new primary medical specialty. The first LGG certifying exam was offered in 2019. To facilitate the transition, ABMGG developed a time-limited alternative pathway for diplomates seeking LGG certification who were already certified in Cytogenetics and Genomics or Molecular Genetics and Genomics, utilizing instructional materials developed by ACMG.

Pictured from left to right during ACMG's 2024 Annual Meeting: ACMG CEO Melanie J. Wells, MPH, CAE; ACMG President Susan Klugman, MD, FACMG, FACOG; ABMGG CEO Miriam (Mimi) Blitzer, PhD; and ACMG Membership Committee Chair Yuan Ji, PhD, MS, MBA, FACMG.

ABMGG CEO Dr. Miriam (Mimi) Blitzer (left) and ACMG President Dr. Susan Klugman, after Mimi was honored at the 2024 ACMG Annual Meeting for her 15-year tenure at the helm of ABMGG.

3. **Transitioning our laboratory training programs to accreditation by the ACGME (2017):** In recognition of the rigorous training in our specialties, the move of LGG and CBG (Clinical Biochemical Genetics) to ACGME accreditation represents the first time ACGME has accredited medical training programs other than MD residencies or fellowships.

4. **Implementing Continuing Certification for ABMGG diplomates (2018):** ABMGG diplomates now maintain their certification through an ongoing program rather than in 10-year cycles. In 2020, this was expanded to incorporate the Longitudinal Assessment Program (CertLink), a twice-yearly assessment that replaces the previous, 10-year, high-stakes recertification exam.

5. **Offering certifying exams annually (2025):** Beginning in Augst 2025, certifying exams will be offered yearly for all specialties except CBG and MBG (Medical Biochemical Genetics), which will remain on a two-year cycle.

It is gratifying to see how far our medical specialty, medical genetics and genomics, has grown and matured in the last 15 years. The ABMGG and ACMG continue to work together on many important topics, with workforce challenges at the top of the list. It's been an amazing journey these last 15 years, and I can't wait to see where this trajectory leads us next!

Mentoring the Next Generation: Summer Genetics Scholars Program

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

This past summer, the ACMG Foundation for Genetic and Genomic Medicine (ACMGF) sponsored the 14th annual Summer Genetics Scholars Program (SGSP). Eighteen first-year medical students spent six weeks in a "hands on" training program at one of the 11 institutions around the country that were selected to participate in the program this year. Eleven of the students were directly sponsored by the ACMGF, while an additional seven were sponsored by their hosting institution.

The goal of the SGSP is to increase the genetics workforce and inspire participating medical students to learn more about genetics, whether they ultimately go into the field or not. The Foundation thanks BioMarin, Ultragenyx and all past contributors who supported students and our ongoing efforts to expand the genetics workforce pipeline.

Established in 2011, SGSP has sponsored more than 230 scholars at more than 60 institutions around the country. The program teaches medical students about the field of genetics and provides them with practical experience in many different facets of genetics. One scholar stated "I have learned so much during my Summer Genetics Scholars Program…we often do not get much interaction with medical genetics as medical students while on rotations, and thus it can be difficult to get a grasp on the field during that time. However, this summer program gave me the opportunity to experience the breadth of medical genetics – from pediatric and metabolic genetics to cancer genetics to prenatal genetics."

"This experience did a lot to confirm my interest in genetics, and therefore I want to continue learning more," explained another scholar. "I plan to attend the weekly genetics case conferences when my class schedule allows… I am certain that everything I learned this summer will serve me well as I continue my medical education." These students are the future of the genetics workforce and the ACMGF is thrilled to be able to be able to provide them with this unique opportunity.

If you would like more information about the Summer Genetics Scholars Program, visit the Foundation website at https://bit.ly/ACMGF-SGSP.

2024 ACMG Education Needs Assessment Report

The ACMG conducted a comprehensive Education Needs Assessment to understand the current landscape and future needs of its members. The survey gathered responses from 303 participants, revealing key insights into the demographics, educational preferences and professional challenges of the genetics community.

Demographics and Professional Background

- A majority of respondents (67.33%) are current ACMG members.
- The primary roles represented include medical/clinical geneticists (40.92%), genetic counselors (11.88%) and other healthcare professionals such as laboratory scientists and educators.
- Participants work predominantly in academic medical centers, hospitals and commercial laboratories.

Educational Needs and Preferences

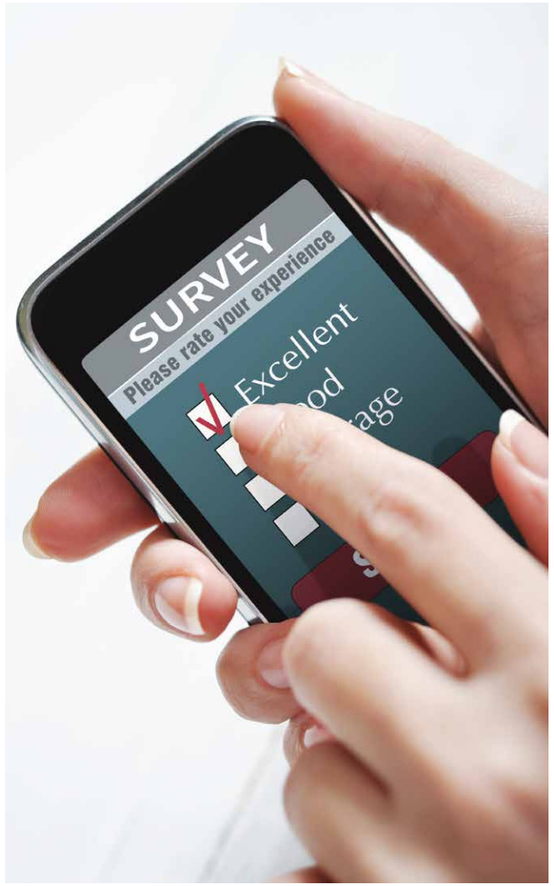
- The top credits sought by members include AMA PRA Category 1 Credits™ and educational credits for other healthcare professionals.
- In terms of educational content, respondents expressed a strong preference for in-person courses, webinar series, and OnDemand courses through the ACMG Genetics Academy. Many also value podcasts and other flexible learning formats.

Key Findings

1. **Educational Impact:** The ACMG's educational programs have significantly contributed to the professional growth of participants with 88.45% affirming positive changes in their clinical practices. Many respondents highlighted the importance of staying updated with the latest research, technologies and clinical guidelines.
2. **Challenges and Gaps:** There is a notable demand for more content on emerging technologies, complex case management and practical applications. Additionally, members expressed a need for more affordable educational opportunities and better support for diverse professional roles, including genetic counselors and laboratory specialists.
3. **Future Focus Areas:** Key areas for future educational content include advancements in molecular genetics, regulatory and legal updates in genomic medicine and enhanced interdisciplinary collaboration. Participants also emphasized the need for improved coverage of adult genetics and prenatal genetics.

The ACMG Education Needs Assessment highlights the organization's crucial role in providing comprehensive and up-to-date educational resources for the medical genetics community. The findings underscore a strong commitment to continuous learning and professional development among members, with a call for more tailored and accessible educational opportunities. The College aims to address these needs by refining its educational offerings and expanding its support for all healthcare professionals in the field of genetics.

This article provides a snapshot of the ACMG's ongoing efforts to enhance the knowledge and skills of its members, ensuring they are equipped to meet the evolving challenges in the field of medical genetics and genomics.



Vincent M. Riccardi, MD, FACMG

by **Judith L. Benkendorf, MS, CGC** and **Wayne W. Grody, MD, PhD, FACMG**

The medical genetics community lost a giant, a mentor and friend with the death of ACMG founding fellow Vincent (Vic) Riccardi on July 2, 2024 in La Crescenta, CA; he was 83. Vic will be remembered by patients, families and colleagues as a supremely influential name in neurofibromatosis (NF) care and research for more than half a century and the first physician to make NF a personal subspecialty. He was also a Renaissance man, published poet, astute observer, longtime student of interpersonal communication and the philosophy of language, with special interests in medical communication and counseling, equitable access, human biology, and the origin of life. ACMG appointed him History Editor of *Genetics in Medicine* (2013-2017), where he created a Genetic Legacy column, inviting selected leaders to share first-person narratives on their professional journeys in medical genetics.

A graduate of UCLA with a major in zoology, Vic received his MD from Georgetown University then completed an internal medicine residency at the University of Pittsburgh Medical Center, followed by clinical genetics fellowship at Mass General. This led to faculty positions at the University of Colorado, Medical College of Wisconsin, and Baylor College of Medicine where he directed the Research Cytogenetics Laboratory and founded the Neurofibromatosis Institute. It was during this time (1977-90) that his seminal contributions to NF flourished: publishing two key books on the disease, launching a dedicated journal, co-sponsoring the first international NF research workshop, initiating the first therapeutic trial in the disorder, and prominently including patient advocates in these efforts.

Returning to California, Vic worked as voluntary faculty in UCLA's Medical Genetics Clinic while continuing his lifelong inquiry into NF, earning an MBA and founding American Medical Consumers, a service designed to help patients navigate the US healthcare system. Vic took advantage of his proximity to Hollywood, working as an "extra" in television and films, sometimes showing up to clinic "in costume"!

We are two of many colleagues and trainees who benefitted immeasurably from Vic's skills as a physician, scientist and mentor. He never forgot the people behind every diagnosis and was beloved as a caring, almost parental, figure to generations of NF patients and families. Vic embraced his life and work with passion. He overcame an early adulthood spinal cord injury, manifesting a lifelong perseverance in the face of adversity. He was truly one of a kind.

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.

Additionally, the ACMG and the National Coordinating Center for the Regional Genetics Network (NCC) publish ACTion (ACT) Sheets, which serve as clinical decision tools for healthcare providers without genetics expertise. ACT Sheets are categorized by subject areas including newborn screening, carrier screening, diagnostic testing, family history, transition and secondary findings.

The following is a list of the College's recent publications since the Summer 2024 edition of *The ACMG Medical Geneticist*:

ACMG Statements, Guidelines, Technical Standards and Practice Resources

Adam S. Gordon, et al. **Consideration of disease penetrance in the selection of secondary findings gene-disease pairs: A policy statement of the ACMG.** *Genetics in Medicine* published online May 30, 2024; doi: http://doi.org/10.1016/j.gim.2024.101142

Saurav Guha, et al. **Laboratory testing for preconception/prenatal carrier screening: A technical standard of the ACMG.** *Genetics in Medicine* published online May 30, 2024; doi: http://doi.org/10.1016/j.gim.2024.101137

Bryce A. Seifert, et al. **Myotonic dystrophy type 1 testing, 2024 revision: A technical standard of the ACMG.** *Genetics in Medicine* published online June 5, 2024; doi: http://doi.org/10.1016/j.gim.2024.101145

ACMG ACT Sheets and Algorithms

ACMG and NCC. **Congenital Adrenal Hyperplasia ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Cogenital-Adrenal-Hyperplasia-ACT-Sheet.pdf

ACMG and NCC. **Congenital Adrenal Hyperplasia Algorithm.** Available at https://www.acmg.net/PDFLibrary/Elevated-17-OHP-Algorithm.pdf

ACMG and NCC. **Congenital Hearing Loss ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Hearing-Loss-ACT-Sheet.pdf

ACMG and NCC. **Congenital Hearing Loss Algorithm.** Available at https://www.acmg.net/PDFLibrary/Hearing-Loss-Algorithm.pdf

ACMG and NCC. **Cystic Fibrosis High IRT+ 0 Variants ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/CF-High-IRT-ACT-Sheet.pdf

ACMG and NCC. **Cystic Fibrosis High IRT+ 0 Variants Algorithm.** Available at https://www.acmg.net/PDFLibrary/CF-High-Variant-Algorithm.pdf

ACMG and NCC. **Cystic Fibrosis High IRT+ 1 Variant ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/CF-1-Variant-ACT-Sheet.pdf

ACMG and NCC. **Cystic Fibrosis High IRT+ 1 Variant Algorithm.** Available at https://www.acmg.net/PDFLibrary/CF-1-Variant-Algorithm.pdf

ACMG and NCC. **Cystic Fibrosis High IRT+ 2 Variants ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/CF-2-Variants-ACT-Sheet.pdf

ACMG and NCC. **Cystic Fibrosis High IRT+ 2 Variants Algorithm.** Available at https://www.acmg.net/PDFLibrary/CF-2-Variants-Algorithm.pdf

ACMG Therapeutics Bulletins

Pavalan Selvam, et al. **Trofinetide approved for children and adults with Rett syndrome (RTT): A therapeutics bulletin of the ACMG.** *Genetics in Medicine* Open published online July 15, 2024; https://doi.org/10.1016/j.gimo.2024.101856

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