

THE ACMG Medical Geneticist

The Newsmagazine of the American College of Medical Genetics and Genomics

with ACMG's Incoming President, Dr. Marc S. Williams, on Priorities, Opportunities, and Challenges Ahead

In a recent interview with *The ACMG Medical Geneticist*, Marc S. Williams, MD, FAAP, FACMG, FACMI, described his priorities for the College as he begins in his new role as ACMG president, the challenges he expects, and the coming opportunities he sees for volunteers to help shape the practice of medical genetics and genomics in the future.

ACMG Medical Geneticist (ACMG): What are your top priorities for ACMG while you serve as its president during the next two years?

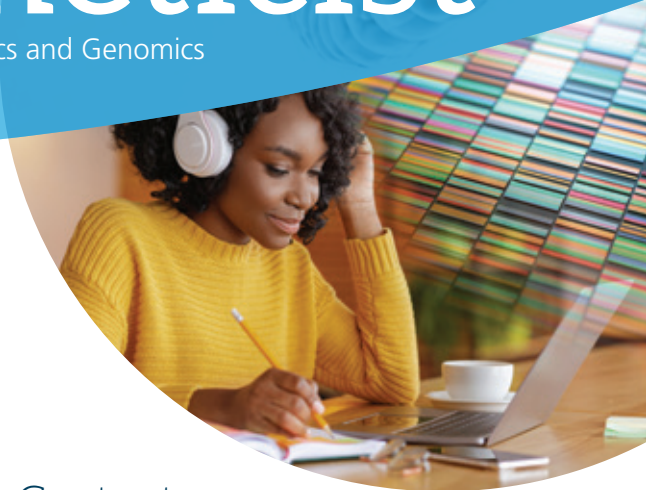


Marc Williams (MW): One of the important responsibilities I have as ACMG president is not only to explore new initiatives, but also to continue supporting critical initiatives and programs that are already in place. I plan to continue supporting the College's commitment to developing evidence-based guidelines,

which are our most important products as a professional society. I'm excited to not only maintain this initiative, but also to expand it going forward.

Among our highest priority initiatives is our focus on Diversity, Equity and Inclusion (DEI). Every day, we see the need for this in our patient population, our workforce, throughout medicine and society, and even within ACMG itself. I'm committed to advancing, expanding and accelerating our efforts in DEI. Our DEI Committee, chaired by Dr. Katy Phelan and co-chaired by Dr. Cinthya Zepeda,

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From the Editor-in-Chief

The theme of this edition of your member Newsmagazine is education, a very fitting topic following such a successful virtual meeting in April. To deliver such a valuable educational experience was an impressive

achievement on the part of the Program Committee and the ACMG Education and Meetings team. I hope many of you were able to participate – the sessions, posters and other offerings were outstanding and, for someone as computer-challenged as I am, the site was very easy to navigate. The summary on [page 12](#) gives details of the *Virtual Experience* and on [page 13](#) you can read comments from some of the attendees. Along this same line, in the CEO column, Max Muenke reviews the ways that ACMG contributes to the education of members and non-members throughout their careers through various platforms and programs.

The Q&A features an introduction to ACMG's new president, Dr. Marc Williams, who discusses the priorities, opportunities, and challenges facing the College. Along with pursuing new initiatives, Marc stresses the importance of supporting our current programs, including accelerating our DEI efforts which could not be more crucial in genetics, in healthcare, and in society. The Advocacy and Government Affairs (AGA) Committee is a relatively new committee of the College that keeps its eye on legislation that can affect the application

of genetics and genomics in healthcare. You can read about what this committee has achieved in its first year and learn what activities are currently on their radar on [page 34](#).

We typically include many photos from the meeting in this edition, but lacking those due to the virtual format, maybe you will recognize someone you know in the images from Medical Genetics Awareness Week ([page 24](#)) and the I Got the Shot campaign ([page 28](#)). These two activities are special ways to highlight our colleagues who are supporting genetics and genomics and doing their part to end the pandemic.

This is also a time of transition, so we thank the outgoing committee chairs and board members for their drive and dedication, and we welcome the incoming board members. We also congratulate the recipients of the ACMG Foundation Awards and the Summer Genetics Scholar fellowships.

Until next time –

Katy

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Meet ACMG's Newest Staff Members



David Carpenter
Advocacy Manager

David Carpenter joined ACMG in December 2020. He brings a broad base of political, policy, and government affairs experience to the College. A graduate of American University in Washington D.C., he has

worked on several political campaigns, including for then Senate Majority Leader Harry Reid's successful reelection campaign, and in multiple congressional offices, including on the floor of the House of Representatives for Speaker Nancy Pelosi. His policy portfolio prior to medical genetics policy predominantly focused on financial regulatory policy as well as time spent in hydrogen energy policy. "I'm excited and grateful to come aboard ACMG and work with leading professionals in the field. Moving into health policy has been a longtime goal and I look forward to growing with the organization as we work together on important policy priorities."



Mame Diaw, MS
Accountant

Mame Diaw joined ACMG at the beginning of January as a staff accountant. Prior to working at ACMG, Mame was the senior accountant at American General Supplies. During her tenure at AGS, she helped the company

automize, integrate and streamline its accounting system and processes. Mame received her master's degree in financial management from the University of Maryland. She has also worked on contracts with Discovery Communications and Otsuka. Since joining ACMG, Mame says, "I am excited to have joined ACMG and grateful for the opportunity to be part of a great accounting team. I am looking forward to continued learning at ACMG."



Nataly Schwartz, MBA, MS
Governance and Operations Manager

Nataly joined ACMG as governance and operations manager in December 2020. Her role involves working on oversight of the Foundation's programs, activities and awards, building effective

relationships with donors and companies, and supporting both the Foundation Board of Directors and the College Board to advance the mission of ACMG. Nataly has an MBA and a Master of Science in Nonprofit and Association Management from the University of Maryland. Nataly brings to ACMG over 15 years of experience working in medical nonprofit associations. She has held leadership roles in program management, business development, communications, and volunteer management. Nataly said,

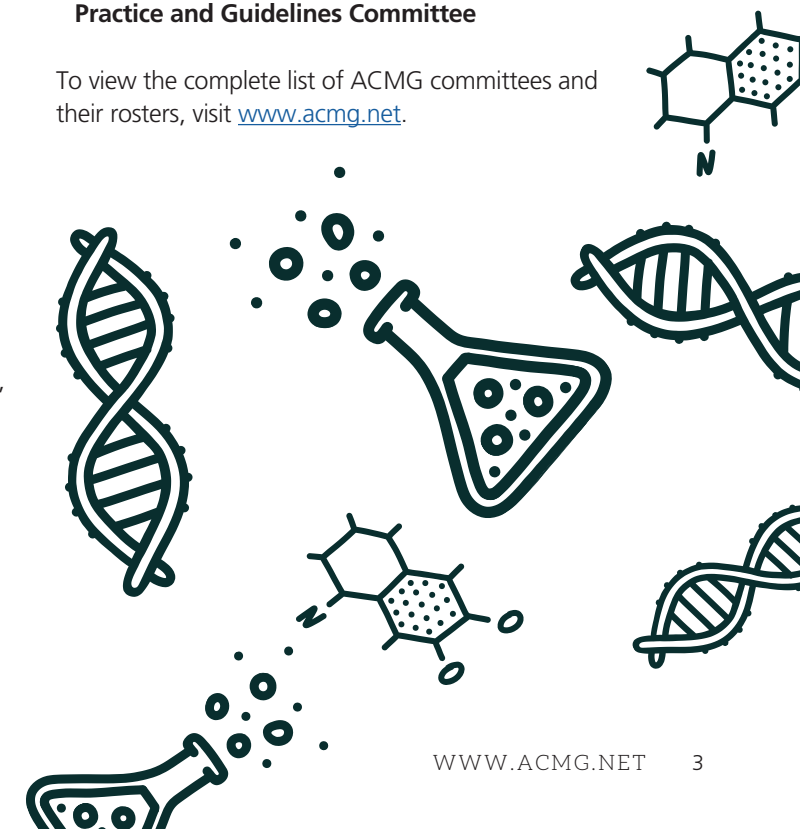
"It is very exciting to work with such an accomplished and dedicated staff and prestigious board of directors. It is an honor to be able to support the ACMG in achieving its mission, especially given the current attention on breakthroughs in medical genetics and genomics. I look forward to being a part of developing something greater than myself."

ACMG Thanks Outgoing Committee Chairs, Vice Chairs

ACMG committees are vital to advancing the work of the College. Committees are made up of ACMG members who volunteer their time to serve the College and our profession. During the 2021 ACMG Annual Clinical Genetics Meeting – a *Virtual Experience*, six ACMG committee chairs and vice chairs completed their terms. The College thanks these outgoing committee chairs and vice chairs for their dedicated service:

- **Michael T. Bashford, MD, FACMG: Vice Chair, Professional Practice and Guidelines Committee**
- **M. Laura L. Cremona, PhD, FACMG: Vice Chair, Economics of Genetic Services Committee**
- **Kim L. McBride, MD, MS, FACMG: Chair, Therapeutics Committee**
- **Fady M. Mikhail, MD, PhD, FACMG: Chair, Laboratory Quality Assurance Committee**
- **Anne M. Slavotinek, MBBS, PhD, FACMG: Chair, Education and CME Committee**
- **Douglas R. Stewart, MD, FACMG: Chair, Professional Practice and Guidelines Committee**

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



developed a Diversity Statement that was approved by the ACMG Board of Directors in 2020. We've had initiatives toward DEI in the past, but nothing of the magnitude the committee is now pursuing. We're doing work on this with other societies aligned, including the American Society of Human Genetics, the National Society of Genetic Counselors, and the National Human Genome Research Institute (NHGRI) at NIH. We're open to other collaborations with groups working on these important issues. I hope to have an action plan with an associated timeline for these efforts approved by the board during our summer meeting. This will be publicly available so that we can be accountable for our commitments.

In terms of new initiatives, I'm very interested in how we can leverage technology to improve the care of patients. In addition to being board certified in pediatrics and genetics, I am also board certified in clinical informatics. The volume of information we now have at our disposal with sequencing, and with rich electronic health record data, allows us to do things with patients who have ultra-rare diseases that just had not been possible before. ACMG receives funding through the NHGRI ClinGen



“We need to think creatively going forward about how we can provide genetic services that are better integrated with care delivery in general and reduce health disparities.”

project, one of the priorities of which is to improve the availability of genetic resources for clinicians. And this is a critical effort because as a genetics workforce, there will never be enough of us to ensure that every patient with a genetic disorder has face-to-face access to a geneticist or a counselor. We must develop methods to distribute our knowledge so it can be used effectively by our non-genetics colleagues.

ACMG: Where do you see the greatest opportunities for the College and its members looking ahead?

MW: I think we are at a point now where genetics is becoming much less exceptional in medicine than it was in previous

decades. Clinicians of all specialties and levels of training understand now that genetics is part and parcel to the patients they see, and that it is important to know about it and pay attention to it. So, another opportunity we have been discussing at the board level is how we can take some of the knowledge that we have as geneticists and provide additional training to individuals who aren't interested in practicing as geneticists, per se, but still want to improve the care of patients who have genetic diseases in their practices. A project like that would involve partnering with other societies, maybe through mini fellowships or other types of innovative approaches that offer specialty-specific content that could be used by a cardiologist or nephrologist, for example, or anyone else who wants to. There are a lot of challenges with an endeavor like this, but there are opportunities, as well. I know the American Board of Medical Genetics and Genomics has begun conversations with sister boards about how our certification processes could be coordinated to achieve this.

Another opportunity, which I touched on previously, relates to technology and patient management. When I was on the ACMG board previously, in the context of newborn screening we started developing ACT sheets, and I helped to develop flow charts so providers could visually see what to do if they had a child that tested positive on newborn screening. We've expanded this now to encompass secondary findings and other things, and we find these flow charts to be very useful. The interesting opportunity is that these diagrams are potentially computable. So, we could embed these as



clinical decision support tools in electronic health records to help clinicians provide care. We've explored this a little in terms of collaborations with technology groups and vendors, but Dr. Muenke and I have begun thinking more about how ACMG might be able to disseminate our guidelines and best practices through enhanced technologies.

One final opportunity worth commenting on—and it is one that's near and dear to my heart, because my first involvement with it was through my work on the Economics Committee—is reimbursement. I chaired that committee for several years, and it was my first chance to provide leadership in the College. I have maintained an interest in these issues of coverage and reimbursement and continue to participate in dialog with payors and employers and technology

assessment organizations to make the case that genetics is important and needs to be reimbursed without additional barriers or impediments. I think we are getting to a point where this is becoming less of an ongoing issue. It's certainly not a solved problem, there's lots more work to do. But we have a much better situation for coverage and reimbursement now than we did 20 years ago, when I first started to work on this issue.

ACMG: What are the biggest challenges facing the field of medical genetics and genomics currently?

MW: Our workforce is very uniform, at least from the perspective of race and ethnicity, and we need to be aggressive about expanding its diversity. We also have a challenge with the number of people who go into genetics with formal training. I can't imagine a more exciting field to be in, but that has not become a general belief among medical students and residents who are considering what to do with their lives. A number of reasons have been put forward for this, and frankly a lot of the non-procedural specialties are having trouble attracting trainees because of the cost and debt load, which directs people toward specialties that are more remunerative. Because we have a small workforce, we also have fewer role models to put forward to medical students.

If we move even farther back and think about when people are beginning to direct career ambitions in high school and college, what can we do to inspire the excitement of genetics, genetic medicine, and career opportunities in this field? For those who are interested

in contributing to new knowledge, there are so many opportunities to pursue interesting science in a clinical genetics role. Back when I was a solo practice geneticist, based on patients I saw in the clinic I was able to participate in gene discovery and definitions of clinical conditions and syndromes. It's really thrilling to do this work, and we need to get that story out. So, we have opportunities and initiatives going on, funding for summer fellowships in our genetics units, and special interest groups

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at our medical schools, and we'd like to expand these programs. While we have had these programs for several years, we've never done an analysis of how they actually influenced career choices. Are the students who participate more likely to pursue genetics? We now have enough time to look at residency and fellowship choices to see if we're making a difference.

The final challenge I see relates to models of delivering genetic services. Because genetics and genomics are now affecting medicine so broadly, the way we have done things in the past is probably not going to be sustainable going forward. It's too inefficient, it doesn't scale well, and we know when you have a situation with a rapidly changing landscape and current practices aren't well adapted to that landscape, there's an opportunity for disruption. That's why we're seeing direct-to-consumer testing now, and people offering services around a genetic condition, because these are ways to deal with the increasing need to use genetics in medicine using alternative models. But in some ways it's disrupting the role of geneticists as thought leaders in terms of how this should be done. So, we need to think creatively going forward about how we can provide genetic services that are better integrated with care delivery in general.

ACMG: Where does the College need more volunteer support and how can members get more involved?

MW: The truth is that we need volunteer support for each of the opportunities and challenges I've just described. When that call for volunteers comes out, please don't be shy about being willing to sign up. We have been listening very carefully to our members about some of the ways we can improve the process of volunteering, and we are definitely going to be implementing those suggestions, so we hope this will make it easier for people to volunteer in the future.

When I was on the ACMG Board of Directors previously, it was not uncommon for members to

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come up and say, “You have to fix this problem or that problem.” I have had a lot of conversations with these people explaining that the problem they asked us to fix is not one we actually can fix, because you're talking about an individual problem with an individual insurer, and you are much more likely to solve that problem than we are as long as you use the materials we provide. So, in some ways I think we need to recognize that there is

a need for people to not look to someone else to solve their problem, but instead bring their problems forward and ask, “What is the role of the College and what role can I play to lead efforts to solve this issue?”

That's how I first became involved with leadership at ACMG. It was in raising discussions with the executive director about problems with coverage and reimbursement. We decided it would be important to have information available to members, so we created a coverage and reimbursement

manual, and I worked with colleagues to write it. The manual wasn't only available to our College members, but also frequently it was purchased by the billing units of healthcare systems, because they were running into the same sorts of problems. And I didn't start out as an expert in reimbursement, I was just someone who said, “We've got a problem, how can we fix it?” So, it's a matter of being willing to get your hands dirty. It's not easy, it's extra work, and it's almost all volunteer. But it's the passion we bring to the field that is critically important, and the College's role is to try to get people passionate about the issues that affect us, but also channel that passion into efforts that can benefit individuals and our College as a whole.

ACMG: How have new lessons learned from the COVID-19 pandemic shaped the future direction of ACMG and opportunities for our members?

MW: No one wanted the pandemic to occur, but it has provided opportunities we can use as a specialty. The transformation of our molecular laboratories into high-throughput viral testing facilities was incredible. ACMG has published a number of stories about that. And to see the rapidity with which people were able to turn around their systems to accommodate the need for testing, I think it is a testament to the quality and vision of people who choose to be geneticists. So that was one takeaway from the pandemic.

The second thing that is really interesting is that the pandemic forced healthcare systems to move toward increased use of telemedicine. Genetics in

CEO CORNER



Dear Members of the ACMG Family, Friends, and Colleagues,

Education is the focus of my CEO Column, of this entire issue, and of ACMG's Strategic Plan Update, which aims “to provide best-in-class education to members and non-members of the College.” Prior to joining ACMG, I was the Director of the NIH Medical Genetics and Genomic Medicine Residency and Fellowship program. In this position, my ultimate goal was to train clinical and laboratory geneticists to provide outstanding, optimal care to patients and families. As the CEO of ACMG, I am extremely pleased to see how the College contributes to educating its members and non-members at various stages of their careers, including undergraduate, graduate, and medical students, clinical residents and laboratory fellows, and board-certified medical geneticists. ACMG contributes to education through 1) our annual meeting either via in-person, virtual or hybrid format, 2) a wide-ranging online ACMG Genetics Academy, and 3) ACMG's journal, *Genetics in Medicine*. Many of these resources are free with ACMG membership, and access to much of our online educational material is free of charge for undergraduate, genetic counseling, medical, and graduate students. Whether you are interested in student and trainee resources, preparing for the ABMGG exam in Laboratory Genetics and Genomics, would like to know about point-of-care ACT sheets or evidence-based guidelines, or are a practicing clinician, laboratory geneticist or genetic counselor who wants to learn more about cutting-edge topics while collecting continuing education credits, ACMG can meet your educational needs. Enjoy reading this issue of *The ACMG Medical Geneticist*.

Enjoy.

Max Muenke

Maximilian Muenke, MD, FACMG





many ways has been a pioneer in this area. We have had publications going back 30 years about use of telemedicine because of the relatively small number of us who need to cover large geographic areas where patients have poor access. So genetic services in mountain states and the western region, particularly for Pacific islands, have been the exemplar. I don't think any of us knew this would lay the groundwork to seamlessly convert our practices from in person to telegenetics, but the reality is that because we have so much experience and resources, we may have had an easier time adjusting to the restrictions of COVID-19 than other specialists.

I expect that we are never going back to fully in-person medicine. There is so much that can be done with telemedicine to prevent the problems healthcare systems suffer from every day: lack of space, efficiency, patients having to find a place to park, sitting in waiting room, etc. Things that do not need to be done in person should not be done in person. We need to think about how this change will impact our practice and how we can utilize

new types of care delivery, and then we need to measure the outcomes to make sure those are equivalent to what we saw with in-person care delivery.

On the negative side, the pandemic has starkly identified the health disparities that are inherent in our delivery system. The disproportionate impact on underserved and underrepresented communities is a call to action for medicine. Of particular interest to the College is the impact of COVID-19 on individuals with disabilities, many of whom have genetic conditions. This will be another area to address as part of our ongoing efforts around diversity.

Medicine is also moving toward more of a value-based system in terms of reimbursement. In other words, up to this point, the more work you did the more you were paid, but that is not sustainable, and it hasn't resulted in optimum quality. There have been initiatives in the last 10 years looking at use of value-based reimbursement. What that means for genetics is we need to define the

New ACMG ACT Sheets NCC, in partnership with ACMG, recently published two new non-invasive prenatal screening (NIPS)– Trisomy 18 and 21 ACT Sheets.	ACMG ACT Sheets and Algorithms Access other ACT Sheets, and accompanying algorithms, for newborn screening, secondary findings, and more at https://www.acmg.net/act	Knowledge Nugget Series NCC develops short animated videos that offer CME to accompany the ACT Sheets. The first in the series, SMA, is available in the ACMG Genetics Academy.
	Genetics and Genomics Education Resources	Access all of these materials at nccrcg.org
Healthcare Interpreter Training To support healthcare interpreters, NCC offers modules to train interpreters on genetics. Access the prenatal or pediatric genetics curriculum on our website.	Policy Education NCC offers a number of resources related to policy education. Learn more about your state's Medicaid programs policies towards genetic services by visiting the Medicaid database on our website.	Resource Repository In addition to our educational resources, NCC has the Resource Repository (ReRe) which has over 450 educational resources from our system and other national partners.

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #U49AC000770 from 8/2020–5/2024 for \$806,000 per award year. The information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

value we provide, the outcomes that lead to high value, and we need to commit to measuring what we do to ensure that, at the individual practitioner level, we are demonstrating high value. This is an evolving area. The system as a whole is gradually moving in this direction, and we need to be aware of that and take advantage. And frankly, it should be at the core of what we do every day: What can we do to improve the outcomes of patients who come to see us?

Value still needs to be defined, however. I think it will be a mixture tied to satisfaction and to patient-reported outcomes. We have not done much in genetics to begin looking at patient-reported outcomes, and it's something we need to be thinking about. Also, we have relied on general patient satisfaction, or generic patient satisfaction—I was privileged to lead some of these projects when I was chairing the Quality Improvement in Clinical Genetics Special Interest Group—but we should continue to do more work. Our colleagues in genetic counseling have developed some specific satisfaction metrics for genetic counseling services. I think that is a good start, but we need to do more than that, and we need to include the perspectives of payors and employers. Because if we do not define the quality measures, someone will define them for us. And they will probably get them wrong.



ClinGen Partners with Human Heredity and Health in Africa (H3Africa) Rare Disease Working Group

by **Danielle Azzariti, MS, CGC**, on behalf of the **H3Africa RDWG/ClinGen Workshop Organizing Committee**

ACMG is a proud partner of the National Institutes of Health (NIH)-funded Clinical Genome Resource (ClinGen, www.clinicalgenome.org), which is building resources that define the clinical relevance of genes and variants for use in precision medicine and research. The Human Heredity and Health in Africa consortium (H3Africa, <https://h3africa.org/>) aims to facilitate fundamental research into diseases on the African continent. Last year, ClinGen and the H3Africa Rare Disease Working Group established monthly calls to work towards collaborative goals, including developing a workshop on clinical genomic interpretation for rare disease. The three-day workshop was held in February 2021. Members of the H3Africa consortium were invited to participate; 220 individuals representing 23 African countries enrolled.



The workshop consisted of lecture-based learning focused on the fundamentals of genomic analysis, gene and variant curation, and genomic data sharing. The third day offered more detailed sessions for participants to choose from based on interest and applicability to their work. All workshop recordings and slides were made available through a website (<https://www.clinicalgenome.org/tools/h3africa-rdwg-workshop/>), which is now publicly available.

Of the 85 respondents who completed the post-workshop evaluation, 30% were students, trainees or fellows, 19% research lab staff, 12% bioinformatics and the remaining 39% included a smaller number of clinical and laboratory roles. Half of the respondents were either familiar (37.5%) or very familiar (12.5%) with genomic interpretation, performing it routinely or sometimes as part of their work, while the remaining half was either learning about genomic interpretation for the first time or rarely performing it in their work. Over 90% of respondents indicated they were Very Satisfied/Satisfied with the workshop content and indicated they were Very Likely/Likely to use it in their work. Interested participants were invited to contribute to ClinGen curation activities, as either biocuration volunteers or disease area experts. This workshop will serve as a model to develop other collaborative international workshops and increase diversity in ClinGen.

Human Heredity and Health in Africa (H3Africa)

H3Africa Rare Disease Working Group and ClinGen Resource Workshop

ClinGen
Clinical Genome Resource

Updates from the ABMGG on Training Programs and Trainees

by **Fuki M. Hisama, MD, FACMG, FAAN**, Credentials Committee Chair, ABMGG and **Miriam G. Blitzer, PhD, FACMG**, Chief Executive Officer, ABMGG

Accreditation Transitions to ACGME

In years past, the American Board of Medical Genetics and Genomics (ABMGG) played a dual role in both accrediting *training programs* in the medical genetics specialties and certifying *individuals* who had completed training. In 1993, when the ABMGG joined the American Board of Medical Specialties (ABMS), the role of accrediting residency training programs transitioned to the Accreditation Council on Graduate Medical Education (ACGME). Now, with the transition of laboratory genetic and genomics (LGG) and clinical biochemical genetics (CBG) programs, the ACGME assumes responsibility for accrediting all medical genetics and genomics training programs.



It is exciting that there has been remarkable growth in combined residency programs offering opportunities for dual board certification. The increased numbers of combined medical genetics and genomics (MGG)/pediatrics programs is particularly impressive, increasing from 16 in 2015 to 23 in 2021. Other currently available combined genetics training programs include MGG/internal medicine, MGG/maternal fetal medicine, and MGG/reproductive endocrinology/infertility.

Within the year, all laboratory training programs will have transitioned to ACGME accreditation. The number of current training programs continues to grow.

ACGME-ACCREDITED TRAINING PROGRAMS

MGG Residency (categorical)	46
Medical Biochemical Genetics Subspecialty	20
Molecular Genetic Pathology Subspecialty	42
Clinical Biochemical Genetics	19
(16 ACGME accredited; 3 in transition)	
Laboratory Genetics and Genomics	41
(34 ACGME accredited; 7 in transition)	

COMBINED CLINICAL TRAINING PROGRAMS

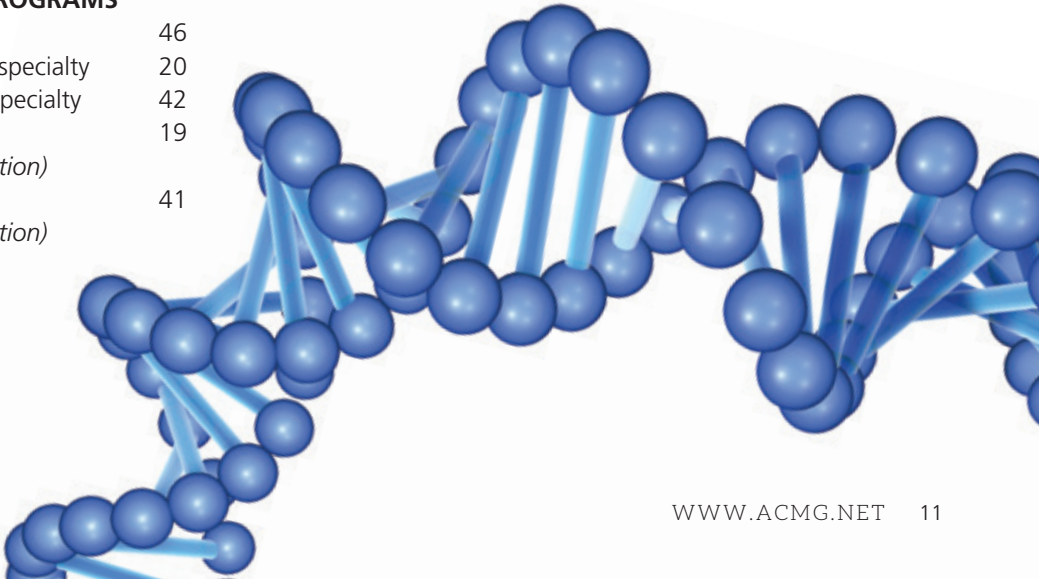
(approved by ABMGG and corresponding board)	
MGG/Pediatrics	23
MGG/Internal Medicine	6
MGG/Maternal Fetal Medicine	8
MGG/Reproductive Endocrine & Infertility	3

Diversity, Equity and Inclusion

The ABMGG is committed to increasing both the number and diversity of trainees entering the field of medical genetics and genomics as the number of trainees entering each specialty has increased over the last 10 years. Our diversity, equity and inclusion efforts include collecting baseline demographic information for ABMGG diplomates and current trainees. Collaborating with the ACMG, we are focusing outreach to medical schools without ABMGG-certified faculty to continue to support the workforce and diversity activities of the other human and medical genetics professional societies.

Highlights for Trainees, Program Directors, Diplomates

- During the COVID-19 pandemic, the ABMGG modified logbook requirements to allow some flexibility in case numbers and to include cases collected by telemedicine.
- It is important for all trainees to go to the ABMGG portal to 1) complete the personal profile page and 2) list both a work and personal email address to ensure timely receipt of all ABMGG notifications—both as a trainee and once a diplomate.
- The number of clinical geneticists, medical biochemical subspecialists, and laboratory geneticists is increasing, which is great news. Our goal is to continue to attract excellent clinicians and laboratorians to our field.
- The ABMGG supports a diverse medical genetics workforce and is committed to making sure our Board and volunteers demonstrate diverse and inclusive representation.
- The 2021 certification examinations are scheduled for August 2021. We wish you all success!
- For more information, please visit our website at <http://www.abmgg.org/> or contact us at abmgg@abmgg.org.



2021 ACMG Annual Clinical Genetics Meeting – a Virtual Experience Highlights

by **Tuya Pal, MD, FACMG**
2021 Program Committee Co-Chair

Jennelle C. Hodge, PhD, FACMG
2021 Program Committee Co-Chair

Jane Dahlroth, CEM, CMP-HC
Senior Director of Meetings & Exhibits

Jane Radford, MHA, CHCP
Director of Education

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



The 2021 Annual Clinical Genetics Meeting was held April 13-16 as a *Virtual Experience*. With face-to-face conferences and travel still on hold due to the COVID-19 pandemic, the ACMG Education and Meetings Teams secured a vendor with an innovative virtual platform. Best-in-class educational content developed by the ACMG Annual Meeting Program Committee was interwoven with opportunities for virtual networking, poster chats, collaboration and an Industry Showcase.

Feedback throughout the week indicated that the virtual platform delivered a great learning and networking experience

on many levels and the overall response from participants was decidedly positive.

Anticipation for the *Virtual Experience* ramped up on April 8 with the presentation of the

Laboratory Diagnostic Challenges. Over 250 learners participated in the live broadcast, and the recording has since been viewed by an additional 850 individuals. This was a terrific interactive session that kept the learners engaged in the multifaceted cases presented.

Then, Tuesday, April 13 marked the official start of the *Virtual Experience*, and for four days nearly 2,800 medical genetics professionals participated in the outstanding educational offering. More than 60 sessions were presented during the live broadcast days in combination with six Online Exclusive sessions that provided additional content for learners to view on demand. The virtual meeting sessions remain accessible to attendees until July 16th, when the platform will close.

ACMG Staff (Jane Dahlroth, Penelope Freire and Jane Radford) were on site in Orlando, Florida in a studio with 20 producers and production managers of the platform vendor CNTVNow. Speakers logged into their “speaker ready room” 30 minutes before each session started and were coached by the production team. As each session started, the staff and speakers experienced the excitement of the countdowns from standby to live. It was almost like producing a live television show...“lights, camera, action”.



“The freedom to watch sessions whenever I want to. It allows me to work and attend the conference.”



Comments from attendees about the virtual offering included appreciation for the ability to watch sessions on their own schedule, often from the comfort of home. While many expressed that they were happy not to travel, they certainly would have liked to see old friends and colleagues and make new face-to-face contacts. Many also communicated that they missed the energy of the in-person crowd. The chat dialogue during sessions helped alleviate these challenges and supported making connections, with many enthusiastically using the platform to greet fellow geneticists and discuss the scientific content as it was shared.

“Seeing all the excellent content from a variety of presenters, including a large number of genetic counselors. GCs with a great knowledge base and variety of experiences seem integral to the success of the ACMG meeting.”

Some highlights of the *Virtual Experience* include:

Scientific Sessions, Platform Presentations and Short Courses

Sessions were in a live and simulative format (presentations pre-recorded; Q&A live), and all were available on demand within five minutes of the original broadcast. Sessions during and after the live meeting have been viewed over 23,800 times.

Abstracts and Posters

Of 548 accepted abstract submissions, 60 were presented as platform presentations and the remaining 488 as poster presentations with an audio tour which could be viewed through the Poster Showcase. Featuring chat capability within the Poster Showcase, attendees could pose questions and provide comments to the poster presenters and have discussions regarding the research presented.

Online Exclusives

Six sessions determined to be of very high interest to attendees were selected as bonus Online Exclusive presentations with an ongoing chat feature. To date, over 1,300 participants have accessed these sessions.

Satellite Symposia

Ten Satellite Symposia were offered during the meeting and have been viewed over 3,600 times. Originally broadcast



2021 ACMG Annual Meeting Available OnDemand

Those who registered for the 2021 *Virtual Experience* will have access to all sessions and the entire virtual platform through July 16. After that date, purchasing the 2021 Digital Edition will allow you to view the content for two years. Those who did not register for the meeting can still access the content at a reduced rate through June 30th by visiting www.acmgmeeting.net for pricing and registration.



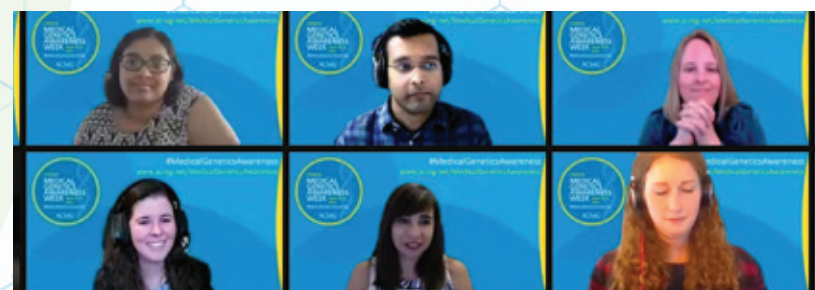
each morning, these sessions are now available on demand through July 16.

Product Theaters

A popular feature at the regular ACMG Meeting, Product Theaters were pre-recorded and have been viewed nearly 1,200 times. All of these presentations are available on demand.

Industry Solution Center

The Industry Solution Center provided opportunities for participants and industry partners to interact and connect. Industry colleagues shared their technologies, products and services via downloadable brochures and videos. They were able to schedule live video chats with attendees or



to chat via a chat box on each sponsor's page. The Sponsor Pages have been viewed over 5,025 times.

Networking and Evening Events

Networking and Entertainment events were also held each evening. From the Trainee/Postdoc/Student Welcome Reception to Los Angeles-based entertainers, subject matter experts or a concert from Nashville (the destination of the 2022 meeting), these events were enjoyed by many.

ACMG Foundation Awards

A page in the virtual platform features the 2021 ACMG Foundation Award winners. This section includes recordings of the awards presented by Dr. Bruce Korf, Dr. David Flannery and Dr. Robert Steiner. The David L. Rimoim Lifetime Achievement Award was presented to Ada Hamosh, MD, FACMG, and her acceptance speech is also viewable.

As Co-Chair of the 2021 Annual Program Committee, Jennelle Hodge, PhD, extends her gratitude to the many talented people who contributed to the *Virtual Experience*. "Our meeting was defined by diversity-centric topics, clinical practice-altering subjects, cutting edge science and thought-provoking discussions, with the content balanced across many different specialties. Importantly, the entire ACMG community should

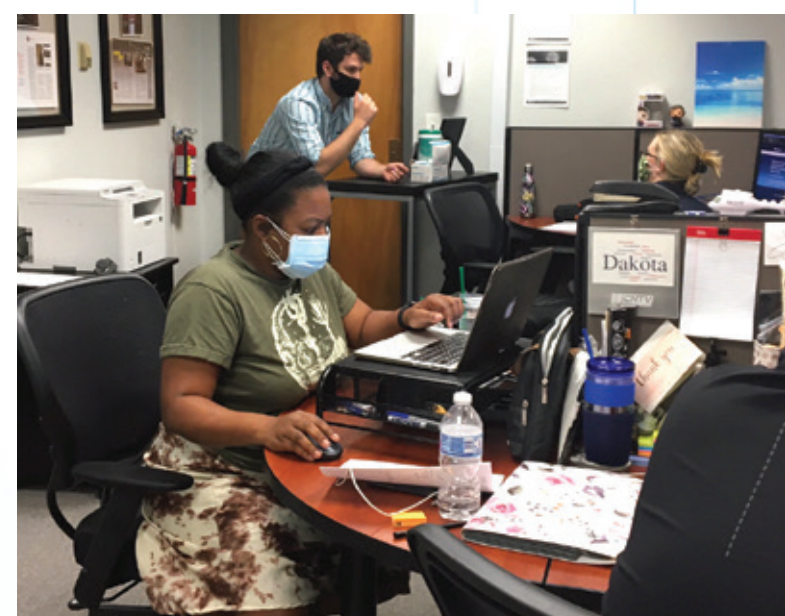
be proud of the high level of science still being produced as well as the care we've given each other during this challenging time." Fellow Co-chair, Tuya Pal, MD, echoes these sentiments and adds that, "Despite the challenges, we are proud to have risen to the challenge of putting together a highly successful virtual meeting, while continuing to try to innovate and expand the reach of our meeting and are tremendously grateful to all contributors and attendees."

ACMG is pleased that we were able to provide outstanding educational content virtually as we all look forward to the return to face-to-face meetings. Special thanks go to the 2021 Program and Education Committees, all of the presenters, and the many industry supporters for their significant contributions towards the success of this year's meeting.



2022 ACMG Annual Clinical Genetics Meeting * March 22-26 * Nashville, Tennessee – Save the dates!

ACMG returns to Nashville! It's a place where haute cuisine and down-home cooking are equally lauded, where urban offerings meet Southern charm, and where the craft of songwriting is as revered as the superstars who bring the music to life. With a state-of-the-art convention center, music scene that is second to none, rock star chefs, and so much more to offer, we look forward to returning to Nashville. The current plan is to offer the 2022 meeting in a hybrid format – with both a face-to-face live option and a digital offering for those unable to travel to the meeting.



“Although I thought I was saturated with Covid-19 issues, I found the diverse approaches of the speakers and their interaction (even their disagreement) very interesting and I was better (scientifically) informed.”

ACMG Welcomes Five New Board Members

ACMG welcomed five new directors, including a new president-elect, to its Board of Directors during the 2021 ACMG Annual Clinical Genetics Meeting – a *Virtual Experience*. 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. The five newly elected directors will serve six-year terms from April 2021 to March 2027. Please join us in welcoming:

- **Susan D. Klugman, MD, FACMG: President-Elect**
- **Shweta Dhar, MD, MS, FACMG: Clinical Genetics Director**
- **Hutton M. Kearney, PhD, FACMG: Cytogenetics Director**
- **David A. Stevenson, MD, FACMG: Clinical Genetics Director**
- **Jerry Vockley, MD, PhD, FACMG: Biochemical Genetics Director**

Also, during the virtual meeting, four Board members completed their terms. The College thanks these outgoing Board members for their dedicated service: Tina M. Cowan, PhD, FACMG; Louanne Hudgins, MD, FACMG; Katy Phelan, PhD, FACMG; and Amy E. Roberts, MD, FACMG.

To read the news release about ACMG’s new board members, visit https://www.acmg.net/PDFLibrary/NewBoardMembers_PresElect2021.pdf. To view the full roster of the ACMG Board of Directors, visit www.acmg.net.



Susan D. Klugman, MD, FACMG
President-Elect



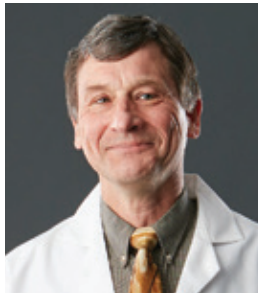
Shweta Dhar, MD, MS, FACMG
Clinical Genetics Director



Hutton M. Kearney, PhD, FACMG
Cytogenetics Director



David A. Stevenson, MD, FACMG
Clinical Genetics Director



Jerry Vockley, MD, PhD, FACMG
Biochemical Genetics Director

ACMG Diversity Statement

As the chair of the ACMG Diversity, Equity and Inclusion (DEI) Committee, it is a pleasure for me to introduce the College’s new Diversity Statement. The DEI Committee is eager to hear your ideas and to facilitate activities that will bring us closer to a diverse and inclusive organization. Please feel free to provide feedback as we move toward achieving an environment of diversity, inclusion, and equity. You will also find the Diversity Statement posted on our website at <https://www.acmg.net/PDFLibrary/ACMG%20Diversity%20Statement.pdf>.

Scientific diversity and the understanding of genomic variation among populations are cornerstones to medical advancement and development of better healthcare systems. The commitment to bolster equity and celebrate diversity is now, more than ever, a necessary step towards addressing our country’s systemic racism, and confronting the neglect and oppression of targeted racial, ethnic, and social groups in our population.

As a medical genetics society and community, the American College of Medical Genetics and Genomics (ACMG) embraces diversity in our members and our patients. ACMG welcomes and encourages participation by all individuals in our organization and the genetics field, regardless of age, sex, gender, sexual orientation, race, religion, ability,



ethnicity, national origin, or economic status. We believe in diversity, both in knowledge of human genome variation and the composition of our membership, to cultivate an interdisciplinary society built on respect, encouragement, and mentorship.

ACMG has championed diversity among our members and joined other professional organizations in their pledge to promote inclusivity and diversity in the genetics and medical fields. Through the creation of the Diversity, Equity, and Inclusion (DEI) Committee, ACMG will endeavor to ensure representation of diverse genetic backgrounds in genetic population databases and study cohorts, and to increase representation amongst our membership from historically underrepresented groups. Through the actions of the DEI Committee, ACMG will work to identify and facilitate activities that promote an environment in which the inherent worth and dignity of all people are recognized, respected, and accepted.

Plan of action regarding diversity, equity, and inclusion
ACMG’s Diversity, Equity and Inclusion (DEI) Committee is responsible for promoting activities to achieve DEI within

ACMG and the medical and genetics fields in our country. To achieve ACMG’s pledge to promote diversity and inclusivity, the DEI Committee will advise the ACMG Board with respect to:

1. Identifying strategies to increase diversity at all levels within ACMG, including membership, representation at the annual meeting (as presenters, panelists, and moderators), and bolstering leadership roles within ACMG through committees and as part of the Board of Directors;
2. Ensuring that medical genetic principles are addressed appropriately and with equity in ACMG policies, procedures, and guidelines;
3. Reducing potential barriers that may sustain advantage to some and disadvantage to others in the genetics and medical fields;
4. Creating educational opportunities for students from underrepresented social groups to perform genetics research at the graduate and undergraduate level; and
5. Reaching out to other professional societies to increase awareness of genetics among underrepresented social groups in science.

The ACMG Foundation for Genetic and Genomic Medicine Presents Eight Awards during the 2021 ACMG Annual Clinical Genetics Meeting – a Virtual Experience



The ACMG Foundation for Genetic and Genomic Medicine is committed to advancing the field of medical genetics and genomics and growing the size of the medical genetics workforce. In support of these aims, the Foundation offers awards that honor excellence in clinical and laboratory practice, teaching and research, and that fund the training of residents and fellows.

During the 2021 ACMG Annual Clinical Genetics Meeting – a *Virtual Experience*, the ACMG Foundation recognized eight individuals with awards and scholarships. These awards included the ACMG Foundation for Genetic and Genomic Medicine David L. Rimoin Lifetime Achievement Award in Medical Genetics, the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award, the ACMG Foundation/David L. Rimoin Inspiring Excellence Award, the Richard King Trainee Award for Best Publication by a Trainee in *Genetics in Medicine*, the ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award, and the ACMG Foundation's Next Generation Fellowship & Training Program Award.

In this issue of *The ACMG Medical Geneticist*, we highlight these awards, the award winners, and recognize and thank Pfizer for their support in the training of the next generation of medical geneticists. To learn more about the vital work of the ACMG Foundation and how you can support our mission to bring more students into the field of medical genetics, visit www.acmgfoundation.org or contact Karl Moeller at acmgfoundation@acmg.net.

The ACMG Foundation for Genetic and Genomic Medicine David L. Rimoin Lifetime Achievement Award in Medical Genetics

Renowned clinical geneticist and pediatrician Ada Hamosh, MD, MPH, FACMG, is the recipient of the 2021 ACMG Foundation for Genetic and Genomic Medicine David L. Rimoin Lifetime Achievement Award in Medical Genetics. Dr. Hamosh, who is the Dr. Frank V. Sutland Professor of Pediatric Genetics and Clinical Director of the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins



Ada Hamosh, MD, MPH, FACMG

University School of Medicine, is being recognized with this award because of her extraordinary commitment to teaching and mentoring, her remarkable energy and dedication while helping patients and their families, and her leadership of online resources—most notably as the Scientific Director of Online Mendelian Inheritance in Man® (OMIM®) and co-creator of PhenoDB and GeneMatcher—that have transformed the use of genetic information in mainstream healthcare worldwide.

The news that she had received the David L. Rimoin Lifetime Achievement Award came to Dr. Hamosh as a tremendous surprise. “You could have knocked me over,” she admitted. “I’m a very behind-the-scenes person, so

the idea that I would get this award is truly overwhelming, and that it is named after David Rimoin makes it even more meaningful to me. David Rimoin was such a kind, generous, thoughtful person, just a wonderful mentor, doctor and human being.”

“Dr. Hamosh is an extraordinarily accomplished medical geneticist and mentor. Aside from the influence she has had on the lives of the patients she has seen and the trainees she has mentored, there are countless others who have benefitted from her contributions to resources such as OMIM® and GeneMatcher, which are practically universally used by practitioners in genetics and genomics,” said ACMG Foundation President Bruce R. Korf, MD, PhD, FACMG. “Her dedication, knowledge, and compassion make her an ideal recipient of this award.”

“The Rimoin family is grateful for Dr. Hamosh’s extraordinary achievements in her research on Mendelian disorders as well as her dedication and commitment to teaching and mentoring junior faculty, fellows and residents,” said Dr. Rimoin’s widow, Dr. Ann Garber-Rimoin. “David would have been particularly impressed with her establishment of PhenoDB and GeneMatcher to advance gene identification, as they share similar



features with the International Skeletal Dysplasia Registry, which enhances the ability to diagnose and characterize skeletal dysplasia disorders. Our family offer whole-hearted congratulations to Dr. Hamosh as the recipient of the David L. Rimoin Lifetime Achievement Award."

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

Noura Abul-Husn, MD, PhD, FACMG is the recipient of the 2021 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award. Dr. Abul-Husn is an associate professor of Medicine and Genetics, founding chief of the Division of Genomic Medicine in the Department of Medicine, and clinical director of the Institute for Genomic Health at the Icahn School of Medicine at Mount Sinai. She is a physician-scientist whose research focus is to uncover the clinical impact of human genetic variation in diverse and unselected populations. Her scientific contributions include pioneering genome-first approaches in population-based biobanks to provide new clinical insights and inform genome-guided therapeutic discovery. She is an

expert at leveraging large-scale genomic data linked to electronic health records, and her work has been published in leading journals, including *Science*, *Cell*, and the *New England Journal of Medicine*. Dr. Abul-Husn's translational research and clinical goals are to integrate leading-edge genomic applications into routine clinical care and drive the equitable implementation of genomic medicine. She recently launched a genomic screening program for medically actionable conditions tailored to ancestrally diverse participants of the BioMe Biobank in New York City.

"I am truly honored and delighted to receive this award from the ACMG Foundation," said Dr. Abul-Husn. "I am deeply committed to growing an innovative program that translates genomic findings into improved patient care for diverse populations."

"We are delighted to announce that Dr. Noura Abul-Husn is the 2021 recipient of the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation. "Dr. Abul-Husn has been a pioneer in the application of genomics to improve health in diverse populations, helping to pave the way towards equitable access to genomic medicine for all people."

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, a period during which Dr. Watson helped ACMG assume its position at the forefront of policy and guideline development.

The ACMG Foundation/ David L. Rimoin Inspiring Excellence Award

Catherine A. Ziats, MD is the recipient of the 2021 ACMG Foundation/David L. Rimoin Inspiring Excellence Award. Dr. Ziats is in her second year of Medical Genetics training at the Greenwood Genetic Center in Greenwood, South Carolina. She received the award for her platform



Catherine A. Ziats, MD

presentation, "Alterations in respiratory epithelial gene *SPDEF* segregate with severe disease in a family with variable response to COVID-19 infection," which she presented during the 2021 ACMG Annual Clinical Genetics Meeting – a *Virtual Experience*.

Upon receiving the award, Dr. Ziats said, "I am very honored and grateful to have received the prestigious David. L Rimoin Inspiring Excellence Award and for the recognition by the ACMG. Importantly, I am extremely grateful to the family members who agreed to participate in this study, who despite their difficult time found it important to attempt to advance our knowledge on host genetic factors associated with severe COVID-19 infection. I am also very grateful for the support and training I have received at the Greenwood Genetic Center, and for the help and collaboration of other GGC researchers who helped with this project. To help promote further similar endeavors and ongoing work, I intend to donate the award money to the Greenwood Genetic Center Foundation."

Dr. Ann Garber-Rimoin, Dr. Rimoin's widow, said, "Dr. Ziats has combined her medical and scientific research interests with COVID-19, the most critical public health care concern in recent history. Her decision to donate the award money to the Greenwood Genetic Center Foundation further demonstrates her generous character, which will go far with her future collaborative efforts. The Rimoin family is happy to acknowledge her accomplishments and pleased to congratulate her as the recipient of the David L. Rimoin Inspiring Excellence Award."

The Richard King Trainee Award

Adam Guenzel, PhD is the recipient of the 2021 Richard King Trainee Award for Best Publication by a Trainee in *Genetics in Medicine (GIM)*. Dr. Guenzel is a fellow in Laboratory Genetics and Genomics at the Mayo Clinic in Rochester, Minnesota. He received the award for his published article, "The critical role of psychosine in screening, diagnosis, and monitoring of Krabbe disease," which was published online in *GIM* in February 2020.

"I am very excited to receive the 2021 Richard King Award. I would like to thank the ACMG Foundation for this honor and also recognize the collaborators that have contributed to and improved this work. Together we have enhanced testing for the diagnosis and monitoring of patients with Krabbe," said Dr. Guenzel upon receiving the award.

"Every year we are amazed at the outstanding quality of articles authored by finalists for the prestigious King award and this year was no different. We are thrilled to announce that Dr. Adam Guenzel's article was chosen as the top article by a trainee this past year and congratulate the author for an excellent manuscript. The caliber of papers published by King awardees reflects the tremendous accomplishments of trainees in genetics and genomics," said Robert D. Steiner, MD, FAAP, FACMG, editor-in-chief of *GIM*.

The Richard King Trainee Award is named for Dr. Richard King in recognition of his instrumental role in creating *GIM*



Adam Guenzel, PhD



Noura Abul-Husn, MD, PhD, FACMG

and serving as the founding editor-in-chief of the journal. The award was established by the ACMG Foundation to encourage American Board of Medical Genetics and Genomics (ABMGG), international equivalents, or genetic counseling trainees in their careers and to foster publication of the highest quality research in ACMG's official journal, *Genetics in Medicine*.

The ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award

Adrienne Bailey, MS, CGC and Renee S. Jones, MS, LCGC are the recipients of the 2021 ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award. Ms. Bailey, a manager of Genetic Counseling at Labcorp Genetics, received the Lovell award for her 2021 ACMG Annual Meeting platform presentation, "A Tale of Two Years: Effects of a Shift in Genetic Counseling Modality on Genetic Testing Uptake and Follow-through." Ms. Jones, a Senior Genetic Counselor at Roche Diagnostics Solutions, received the Lovell award for her 2021 ACMG Annual



Adrienne Bailey, MS, CGC



Renee S. Jones, MS, LCGC

Meeting platform presentation, "Patient Education for Prenatal Aneuploidy Testing using a Chatbot: A Multicenter Randomized Controlled Trial."

Upon receiving the award, Ms. Bailey said, "Thank you to the ACMG Foundation for selecting my abstract to receive the 2021 ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award. I am honored to have my work and the work of my colleagues recognized by the ACMG Foundation."

Ms. Jones said, "We hope that this study will encourage the adoption of alternative platforms to educate patients and obtain meaningful informed consent in the rapidly expanding arena of genetic testing. I am very grateful to the ACMG Foundation for this recognition."

"Congratulations to Adrienne Bailey and Renee S. Jones, as the recipients of this year's award. This year is extraordinary because for the first time two genetic counselors, Ms. Bailey and Ms. Jones received the same exact score in the review process, consequently they each will receive a \$1000 award," said David Flannery, MD, FAAP, FACMG. "I established this award to honor Carolyn Mills Lovell, MAT, MS, CGC, who was an exemplary genetic counselor. It was created to recognize the important contributions of genetic counselors to patient care and to research in the field of medical genetics."

ACMG Foundation's Next Generation Fellowship & Training Program Award

Christina Tise, MD, PhD, of Stanford University, and Daniel Pomerantz, MD, of Children's Hospital of Michigan and Wayne State University, received two 2021 Next Generation Fellowship & Training Program Awards. These "Next Gen Awards" will support Drs. Tise and Pomerantz for one year of postgraduate training in clinical laboratory biochemical genetics and medical biochemical genetics, respectively.

Upon receiving the award, Dr. Tise said, "I am beyond fortunate in having amazing mentors who have cultivated my love for genetics and have provided me incredible support and encouragement at each step of my academic journey thus far. The field of genetics has grown immensely in the past century due to the hard work and dedication of



Daniel Pomerantz, MD

so many amazing geneticists, clinicians, and researchers, but there is still so much to discover! I cannot thank Pfizer and the ACMG Foundation enough for supporting this endeavor and providing me the honor and opportunity to contribute to the field."

Dr. Pomerantz said, "I am ecstatic and deeply humbled to be a recipient of this Next Gen Award in a Medical Biochemical Subspecialty. I am also deeply grateful for the exceptional support by my mentor at Harvard, Dr. Gerry Berry, as well as our esteemed collaborators at the NIH, Dr. Charles Venditti and Dr. Oleg Shchelochkov. With the guidance and mentorship of the outstanding clinical and research faculty members within the Harvard Medical Genetics Training Program, as well as our collaborators at the NIH, I enthusiastically look forward to joining the biochemical genetics community to serve, care and advocate for our patients and their families."

"We greatly appreciate Pfizer's continued support of this year's Next Gen Program. In the past 20 years, Pfizer has committed well over \$1,000,000 and played a major role in helping our foundation provide several dozen students more than 60 years of highly technical training in medical genetics and genomics. Training up-and-coming leaders in medical biochemical genetics is a critical investment in the future of our field that will benefit patients with complex biochemical disorders for years to come," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation.



Christina Tise, MD, PhD

Thanks to You, the Third Annual Medical Genetics Awareness Week Was a Great Success

Through Medical Genetics Awareness Week, ACMG promotes understanding of the importance of medical genetics professionals on the healthcare team—including clinical and laboratory geneticists, genetic counselors, nurses, nurse practitioners, physician assistants, and public health geneticists. This commemorative week was created to raise awareness of the indispensable contributions that

medical genetics healthcare professionals make in the diagnosis, management and prevention of genetic diseases, and the difference these professionals make in the lives of patients and families. During the week, we honor the skills and commitment of all of those on the healthcare team who translate genetic and genomic discoveries into better patient care and public health.

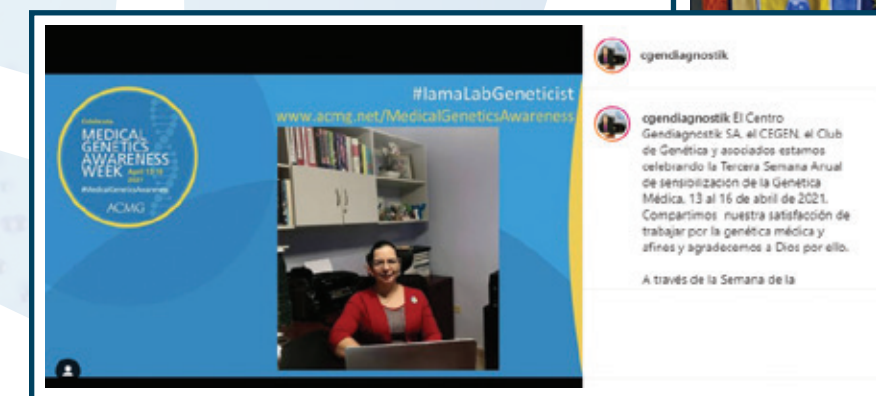
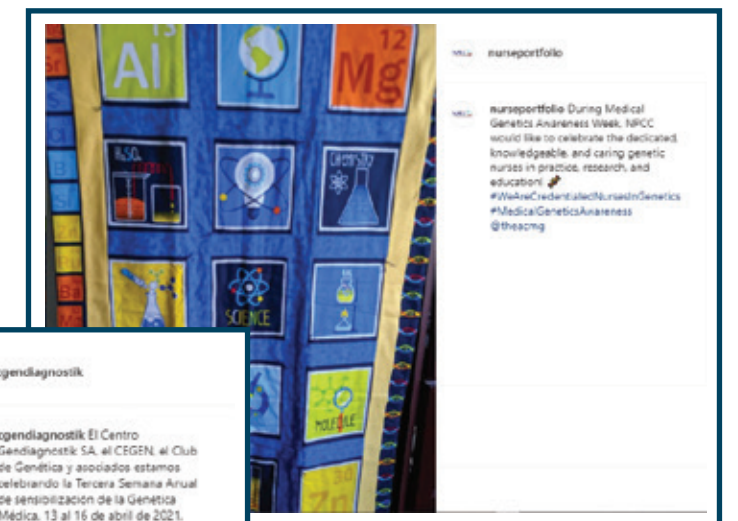
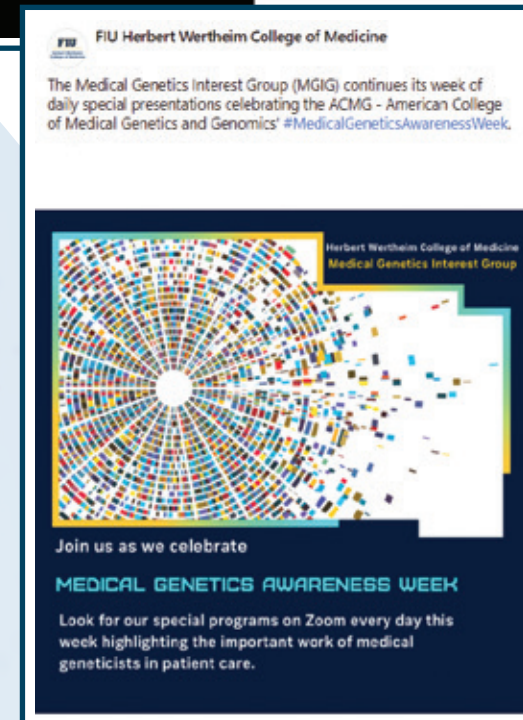
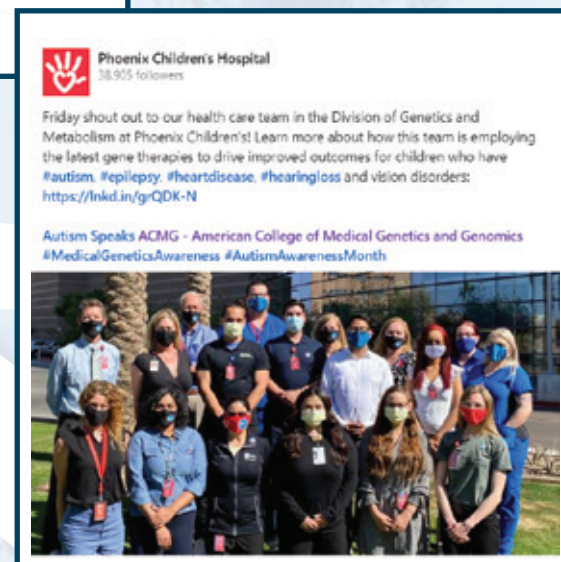
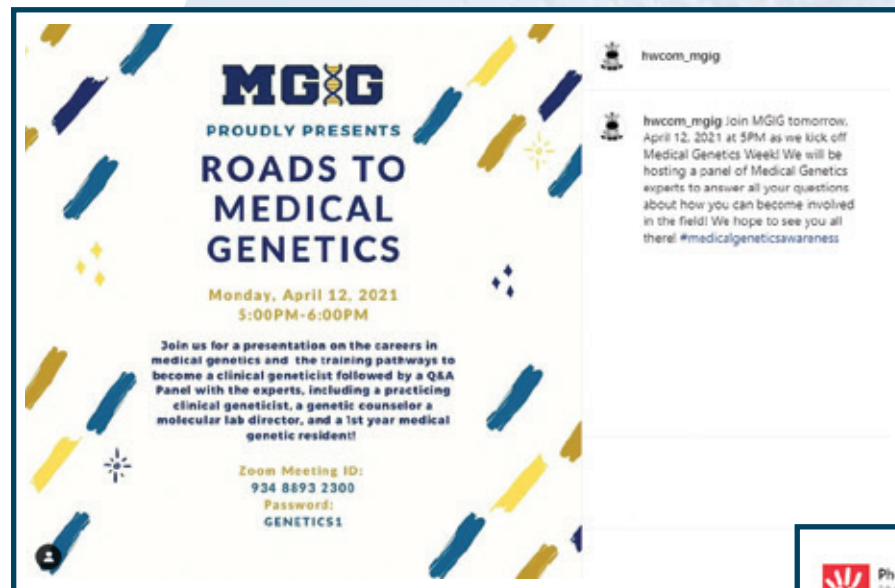
Thanks to you, this year's Medical Genetics Awareness Week was once again a great success! We distributed hundreds of Medical Genetics Awareness Week face masks and nearly 4,000 hashtag buttons and stickers. Our Zoom virtual backgrounds were frequently seen on the ACMG Annual Meeting virtual platform; our profile picture frames could be seen across Facebook; and our **Medical Genetics Awareness Week** hashtags have earned more than 1.9 million impressions so far this year!

We loved seeing the energy and enthusiasm focused on raising awareness of the importance of genetics in medicine. The celebrations and engagement—especially around **Diversity Day**, **Student and Trainee Day** and **ACMG Resources Day**—inspired us and we hope it did the same for you. We also loved seeing your social media posts, just a few of which we are pleased to share here.

As we look forward to next year's Medical Genetics Awareness Week, which will take place March 22-26, 2022, we invite you to visit our Medical Genetics Awareness Week web pages at www.acmg.net/MedicalGeneticsAwareness. On these pages, you will find tools to help you share your medical genetics pride and tips on how you can continue to raise awareness about medical genetics all year long.

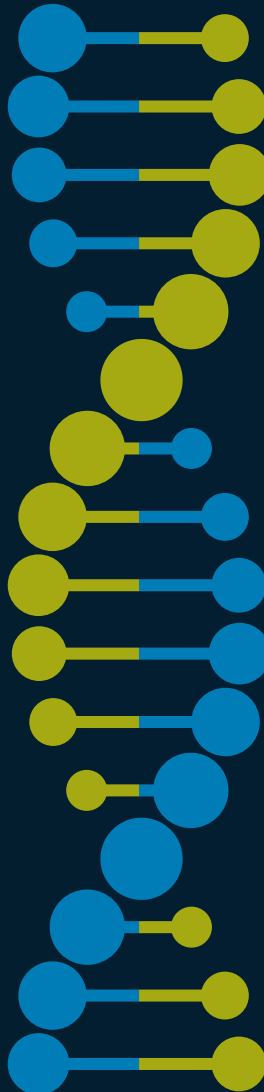
New for 2021—Theme Days, Face Masks and Zoom backgrounds Helped You Share Your Medical Genetics Pride

New this year, we held theme days including a Student and Trainee Day, Diversity Day and ACMG Resources Day. We hope you enjoyed learning how ACMG engages in these important topics. We also created Medical Genetics Awareness Week face masks, colorful Medical Genetics Awareness Week Zoom virtual backgrounds, and two new hashtag buttons. The Zoom virtual backgrounds remain available for free on our Medical Genetics Awareness Week web pages. Face masks (free to members) and hashtag buttons (free to all) are also available, while supplies last, upon request to ACMG Communications Coordinator Reymar Santos (rsantos@acmg.net).



ACMG Membership Has Its Benefits

Save more than \$1,000 per year on a wealth of ACMG resources developed to enhance your professional growth



Education for Your Professional Development and Certification Needs

ACMG provides a full range of learning experiences from introductory content for students to advanced education for medical genetics professionals. Learn in the environment that meets your needs whether it's live, live-streamed or on-demand.

The ACMG Annual Clinical Genetics Meeting featuring the latest developments and research in clinical genetics and genomics and opportunities to meet, network and collaborate with your peers. **Members receive substantial discounts on registration.**

The ACMG Genetics and Genomics Review Course prepares those sitting for medical genetics certification, but can also be used as a means to fulfill CCP requirements for those already certified. **Members receive substantial discounts on registration.**

The ACMG Student page provides access to medical genetics and genomics career information, resources, videos and links to ACMG Student Interest Groups across the country.

The ACMG Genetics Academy, your online learning site dedicated exclusively to medical genetics and genomics education including on-demand ACMG meetings and webinars, Continuing Certification Part II and IV modules and the Genetics Review Course. **Members receive substantially discounted or free access to these offerings.**

Visit www.acmg.net/education to start your learning experience today.

Collaborate and Share Your Knowledge and Expertise (Network, Volunteer, Comment, Advocate)

Network with medical genetics professionals from around the world through our LinkedIn, Facebook, Twitter and Instagram pages.

Connect with the leaders in medical genetics by joining an ACMG committee, workgroup or special interest group.

Take action – look for ACMG Advocacy alerts or visit our Advocacy page and help with our public policy activities.

Provide input on draft ACMG standards and guidelines.

Products, Tools and Resources to Help Your Practice, Lab and Career

ACMG's official journal, *Genetics in Medicine* (GIM), provides members with timely research and original reports, which enhance the knowledge and practice of medical genetics.

Tune-in to **GenePod** for **free podcasts** featuring GIM authors discussing their latest research in medical genetics and genomics.

The bi-annual **ACMG Salary Survey Report**, a **free member resource**, get the data you need to get that raise, negotiate a new job, advise graduating trainees or simply compare your compensation to standards in the field.

The **ACMG Find a Genetics Clinic Directory** helps you locate genetics clinics across the United States.

ACMG's ICD-10 Pocket Guides, **free to ACMG members**, are handy pocket-sized reference sheets to help College members determine appropriate codes for genetic services. Download your copies today from the Members Only website.

The **ACMG in Action** electronic newsletter and **The ACMG Medical Geneticist** news magazine provide members with timely College and industry news, tools, advocacy updates and resources.

Post your resume, search for available positions or announce a new employment opportunity on the **ACMG Employment Resource Center** site. Visit careers.acmg.net to learn more.

ACMG/NBSTRN connects researchers to essential tools with the **Longitudinal Pediatric Data Resource** (LPDR), offering access to more than 60 conditions for secondary analysis.

Join NBSTRN's monthly conference calls to discuss conditions recently added to nationwide screening including **Pompe Disease** and **Spinal Muscular Atrophy** (SMA). Visit www.nbstrn.org to learn more.

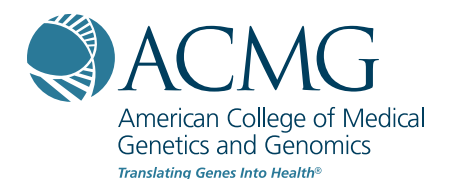
ACMG ACT Sheets and Algorithms, available on our website, help inform the actions a non-genetics provider should take for a variety of genetic conditions including those found in newborn screening, adults and non-invasive prenatal screening

Looking for current information about genetic service policies and legislation? Visit the NCC website (bit.ly/NCCHAF) to access state legislation and regulation reports, subscribe to the **Legislative, Insurance, and Finance Tracking** (LIFT) newsletter and learn about your state's Medicaid genetic services policies.

Not An ACMG Member Yet?

Don't miss out on these benefits any longer!

Contact membership@acmg.net or visit www.acmg.net/join.



ACMG Launches #IGottheShot Social Media Campaign to Encourage COVID-19 Vaccination

Over the past year, ACMG has been inspired by the example our members have set in supporting their colleagues, families, and friends, and caring for and protecting their patients during the pandemic. In January of this year, we announced ACMG's #IGottheShot public relations campaign, which featured photographs of ACMG members getting the COVID-19 vaccine. The goal of the campaign is to show examples of healthcare professionals safely getting vaccinated against COVID-19 and in so doing, encourage others to get vaccinated. Our sincere thanks go out to all those who have participated in the campaign. We hope you enjoy seeing their posts here.



Mahmoud Aarabi, MD, PhD, FACMG
ACMG member Mahmoud Aarabi, MD, PhD, FACMG says, "Today, I got the COVID vaccine in a mobile vaccine clinic at one of the long-term care homes affiliated with my hospital. Wishing for the health and well-being of all of our dear seniors, we love you!"



Jane Dahlroth, CEM, CMP-HC
Jane Dahlroth, CEM, CMP-HC, senior director of meetings and exhibits at ACMG, says, "I got the shot because as the director of the ACMG meetings department I look forward to us meeting again face to face. I have missed seeing ACMG members and meeting attendees this last year. I hope too this will encourage my fellow business event professionals as we all want to see the return to live events, conferences and trade shows."



Elfride De Baere, MD, PhD
ACMG member Elfride De Baere, MD, PhD says, "I got the shot today in Belgium. Thank you!"

Jenna Cohen
ACMG Meetings Manager Jenna Cohen says, "I got the shot" while sporting her Medical Genetics Awareness face mask! She says, "Getting the COVID vaccine is one way we can practice Tikkun Olam and help heal the world."



Tony Gregg, MD, FACMG
ACMG past president Tony Gregg, MD, FACMG says, "I got the shot, thanks to the scientists and other experts who developed the vaccine against COVID-19."



Jennelle Hodge, PhD, FACMG
Jennelle Hodge, PhD, FACMG, past co-chair of the 2021 ACMG Annual Clinical Genetics Meeting Program Committee, says, "Getting the COVID-19 vaccine was a wonderful experience. Inspiring artwork, friendly volunteers excited to help give the life-saving vaccine, and a statement sticker at the end. I'm very grateful for the opportunity to help protect everyone."

Andrea Gropman, MD, FACMG, FAAN, FAAP
ACMG member Andrea Gropman, MD, FACMG, FAAN, FAAP says, "I got the shot because I am a pediatrician first and we believe in the power of vaccines."



Susan Klugman, MD, FACMG
Susan Klugman, MD, FACMG, ACMG's new president-elect, and reproductive genetics editor for *Genetics in Medicine*, says, "I got the shot!"



Jillene Kogan, MD, PhD, FACMG
ACMG member Jillene Kogan, MD, PhD, FACMG says, "I am so grateful to have this opportunity to get the COVID-19 vaccine! Please get your vaccine when it's your turn. This is our way out of this pandemic!"





Kathleen A. Leppig, MD, FACMG

Kathleen A. Leppig, MD, FACMG, cytogenetics/clinical genetics editor for *Genetics in Medicine*, says, "We all need to do our part. Even if we weren't among those called upon to serve on the front lines of the COVID-19 pandemic, we all can support vaccination and moving past the pandemic."

Marco L. Leung, PhD, FACMG

ACMG member Marco L. Leung, PhD, FACMG says, "Science rocks! Grateful to have gotten the second dose of COVID-19 vaccine!"



Dietrich Matern, MD, PhD, FACMG

Dietrich Matern, MD, PhD, FACMG, a member of the ACMG Board of Directors, says, "While I don't know if it is fair that I—a laboratory director—already got vaccinated, I am happy I did as it not only protects myself but also helps protect those I come in contact with. But the mask stays on for now."

David T. Miller, MD, PhD, FACMG

David T. Miller, MD, PhD, FACMG, deputy editor-in-chief of *Genetics in Medicine* and medical geneticist at Harvard Med, Boston Children's Hospital, says, "I got the shot and I'm feeling great!"



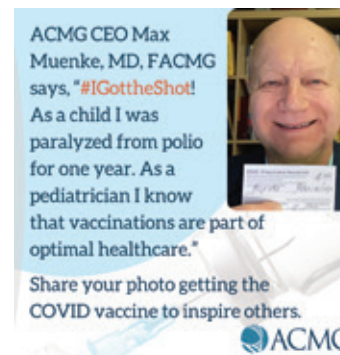
Marilena Melas, MSc, PhD

ACMG trainee member Marilena Melas, MSc, PhD, says, "I got the shot of first dose of Moderna COVID vaccine! Easy, quick, painless! I am a scientist who believes and trusts the miracle of science!"



Max Muenke, MD, FACMG

ACMG CEO Max Muenke, MD, FACMG says, "I got the shot! As a child I was paralyzed from polio for one year. As a pediatrician I know that vaccinations are part of optimal healthcare."



Katy Phelan, PhD, FACMG

Katy Phelan, PhD, FACMG, chair of ACMG's Diversity, Equity and Inclusion Committee, says, "I got the vaccine to protect myself and others from COVID-19 and to end the pandemic. I am ready to get back to living with the freedom I had before the pandemic—traveling, visiting friends and family, attending social events—I want it all back!"

COVID-19 VACCINATION



Beth A. Pletcher, MD, FACMG

ACMG member Beth A. Pletcher, MD, FACMG says, "Be well, stay safe and have a wonderful 2021. Hold your loved ones close and roll up your sleeve!"



Cynthia M. Powell, MD, FACMG

ACMG board member and clinical geneticist Cynthia M. Powell, MD, FACMG says, "I got the shot."



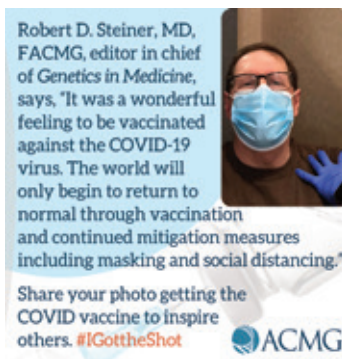
Michele A. Lloyd-Puryear, MD, PhD, FAAP, FACMG

NHGRI's Teri Manolio MD, PhD and Michele A. Lloyd-Puryear, MD, PhD, FAAP, FACMG have been administering the COVID vaccine as volunteer members of the Maryland Medical Reserve Corps. Dr. Puryear says, "I want everyone to be vaccinated!" as she receives her shot from Dr. Manolio.



Robert D. Steiner, MD, FACMG

Robert D. Steiner, MD, FACMG, editor in chief of *Genetics in Medicine*, says, "It was a wonderful feeling to be vaccinated against the COVID-19 virus. The world will only begin to return to normal through vaccination and continued mitigation measures including masking and social distancing."



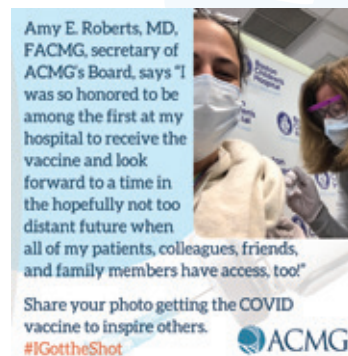
Douglas R. Stewart, MD, FACMG

Douglas R. Stewart, MD, FACMG, cancer genetics editor for *Genetics in Medicine* says, "I 'got the shot' to help protect not only myself and family but the entire community in which I live."



Amy E. Roberts, MD, FACMG

Amy E. Roberts, MD, FACMG, past secretary of the ACMG Board, says, "I was so honored to be among the first at my hospital to receive the vaccine and look forward to a time in the hopefully not too distant future when all of my patients, colleagues, friends, and family members have access, too!"



Sam Strom, PhD, FACMG

ACMG member Sam Strom, PhD, FACMG shared a picture of his vaccination card after getting the COVID vaccine saying, "I got the shot."



Myra Wick, MD, PhD, FACMG

Myra Wick, MD, PhD, FACMG, now chair of ACMG's Program Committee, says, "I got the shot!"



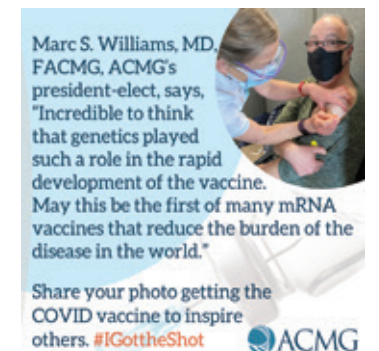
Alessandra Sugrañes, MD

Alessandra Sugrañes, MD, a member of ACMG's Diversity, Equity and Inclusion Committee, says, "I got vaccinated against COVID-19 to help protect my loved ones and my community."



Cynthia J. Tifft, MD, PhD, MGC, FACMG

ACMG member Cynthia J. Tifft, MD, PhD, MGC, FACMG says, "To borrow from Maryland Governor Larry Hogan's comment on mask wearing, 'Just get the d--- shot!'"



Marc S. Williams, MD, FACMG

Marc S. Williams, MD, FACMG, ACMG's new president, says, "Incredible to think that genetics played such a role in the rapid development of the vaccine. May this be the first of many mRNA vaccines that reduce the burden of the disease in the world."



It's A Bit Like Running on a Treadmill: ACMG's Advocacy and Government Affairs Committee – A Very Busy First Year!

by **Sheila Dobin, PhD, FACMG, Chair, Advocacy and Government Affairs Committee** and **Mary Beth Dinulos, MD FAAP, FACMG, Vice Chair, Advocacy and Government Affairs Committee**

The ACMG Advocacy and Government Affairs (AGA) Committee was launched in March 2020, and has been very busy since then. The committee is charged with exploring and reporting on public policies developed by government entities, such as the U.S. Congress, state legislatures, and federal agencies, that impact the application of genetics and genomics in healthcare. We work closely with ACMG's public policy staff to develop recommended positions and comments on legislation and rules provided by rulemaking authorities. The committee studies, evaluates, and prepares information for dissemination to members of the College, policymakers, and the public. All work is subject to approval by the Board of Directors before dissemination.

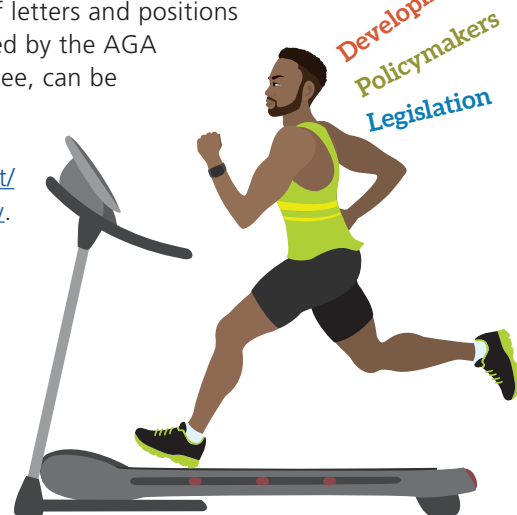
Throughout our first year, the AGA Committee has been very active! We assessed and responded to legislation that would impact the regulation of laboratory developed tests (LDTs). These bills, the VALID and VITAL Acts, did not pass during the last legislative session but are expected to be reintroduced in the current session. We supported the Expanded Genetic Screening Act which would require Medicaid programs to cover noninvasive prenatal genetic screening for all pregnant patients, not just those considered to be high risk. We reviewed and developed a position on the Reducing Hereditary Cancer Act, legislation that would allow Medicare to cover genetic testing for hereditary cancer and certain screening and preventative surgeries as recommended by National Comprehensive Cancer Network guidelines. We are in the process of evaluating numerous telehealth bills that have been introduced by the current Congress. We have also provided requested feedback to members of Congress currently developing

legislation to improve Medicaid coverage of genetic sequencing for children and public payer coverage of genetic testing for rare cancers.

We commented on the Office of the National Coordinator for Health Information Technology (ONC) rule requiring the immediate release of test results and how it might harm our patients, the inconsistent language in the bill surrounding exceptions, and problems posed with reference laboratories releasing the results before the ordering healthcare professional might be notified. While we agree with the basic premise that patients should be informed before testing and thus understand the results, we also know that there are special cases where this may cause harm, especially when there are unexpected findings.

Currently we are working with other ACMG groups on reimbursement of laboratory geneticists for their professional interpretations. While a legislative fix will be necessary, current emphasis is placed on gaining support from other medical associations. Securing this support prior to pursuing legislation is necessary to have any chance of passage of legislation. Additional information about ACMG's position is available on ACMG's website at https://www.acmg.net/PDFLibrary/PhDs_QHPs_Approved_Final_WebPosting08272019.pdf.

The AGA Committee has many other efforts in the works, including developing resources about issues in genetics to guide policymakers. As you can see, the committee is quite busy, and this work could not be done without the dedicated public policy staff at ACMG and our hard-working committee members. Individuals interested in joining the committee should be on the lookout for ACMG's call for volunteers that will go out later this year. If you have any concerns or comments about legislation or rules, please feel free to contact ACMG's policy team at advocacy@acmg.net or contact me directly. Additional information about ACMG's advocacy efforts, including copies of letters and positions developed by the AGA Committee, can be found at www.acmg.net/advocacy.



The ACMG Membership Committee - Growing the Genetics Workforce: Increasing Interest and Diversity in Genetics

by **Jennifer A. Bassetti, MD, FACMG, Chair, ACMG Membership Committee** and **Alessandra Sugrañes, MD, Member, ACMG Membership Committee**

The global COVID-19 pandemic and the ever-growing demand for specialized medical expertise have reinforced the need for a larger and more diverse medical genetics and genomics workforce. The College and its committees are committed to making this a priority. Since 2012, the ACMG Membership Committee has curated and hosted the Student/Trainee Workshop, entitled "Careers in Medical Genetics: An Informational Session for Students and Trainees" held during the annual ACMG Annual Clinical Genetics Meeting. This workshop is designed as an interactive event intended to expose college and higher-level students and genetics

trainees to the array of careers available and to provide attendees the opportunity to network with leaders in the field of genetics and genomics. This year, to promote and highlight efforts on increasing diversity within medical genetics and genomics, the Membership Committee worked in collaboration with ACMG's new Diversity, Equity, and Inclusion (DEI) Committee to engage a diverse group of genetics professionals as our featured speakers. Natario Couser, MD, FACMG presented on a clinical genetics career path, Ruben Bonilla Guerrero, MD, FACMG discussed a clinical-laboratory-based career path approach, and Andrea Schelhaas, MS, CGC shared her path into genetic counseling. Tips and perspectives from recent training program graduate, Cinthya Zepeda Mendoza, PhD, were also very enlightening. Of course, our session would not be complete without details on how to achieve certification in medical genetics and genomics presented by the American Board of Medical Genetics and Genomics CEO Miriam Blitzer, PhD, FACMG. We sincerely thank these speakers who made this year's event a great success!

Traditionally, the Student/Trainee Workshop is held in person at the ACMG annual meeting. Due to the COVID-19 pandemic, its format had to be reimaged. While last year's speakers pre-recorded

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their presentations to be accessed through the ACMG Genetics Academy, this year we shifted to a virtual meeting platform in hopes of mimicking the interactive environment of our traditional in-person workshop. While there were concerns that the virtual platform might hinder attendance, the outcome was quite the opposite! More people attended this year than in the past, with over 250 attendees from 30 different countries representing medical students, genetics trainees, genetic counseling students, and graduate and undergraduate students. This year's careers workshop provided an incredible way for a large number of students and trainees interested in genetics to interact and network with genetic professionals. The virtual platform allowed us to expand and reach a much larger and more diverse audience than ever before.

If you were unable to participate the 2021 workshop and would like to know more about careers in medical genetics and genomics, or want to share this opportunity with students and trainees, the speaker presentations are available on ACMG's YouTube Channel and also posted on the ACMG Genetics Academy under Student/Trainee Resources.

The ACMG Membership Committee is committed to continue its work to encourage and build diversity, equity, and inclusion within the ACMG membership. We encourage you to motivate and support your colleagues, students, and trainees to join ACMG to help move their careers forward in the field of medical genetics.

ACMG Publications: Providing Education and Tools for Medical Geneticists, Other Health Professionals, and the Public

Among the priorities outlined in ACMG's strategic plan are facilitating the delivery of quality clinical and laboratory medical services to patients and their families and providing best-in-class education to members and nonmembers. The publication of timely and expert policy and position statements and clinical and laboratory practice resources, standards and guidelines supports these aims. The College is pleased to announce the following recent publications at the time this edition of the member magazine went into layout and print.

ACMG Statements, Guidelines, Technical Standards and Practice Resources:

Murray MF, Giovanni MA, Doyle DL, Harrison SM, Lyon E, Manickam K, Monaghan KG, Rasmussen SA, Scheuner MT, Palomaki GE, Watson MS and ACMG Board of Directors. **DNA-based screening and population health: a points to consider statement for programs and sponsoring organizations from the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 16 March 2021; doi: <https://doi.org/10.1038/s41436-020-01082-w>.

Bean LJH, Scheuner MT, Murray MF, Biesecker LG, Green RC, Monaghan KG, Palomaki GE, Sharp RR, Trotter TL, Watson MS, Powell CM and ACMG Board of Directors. **DNA-based screening and personal health: a points to consider statement for individuals and health-care providers from the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 31 March 2021; doi: <https://doi.org/10.1038/s41436-020-01083-9>.

Spector E, Behlmann A, Kronquist K, Rose NC, Lyon E, Reddi HV and ACMG Laboratory Quality Assurance Committee. **Laboratory testing for fragile X, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 01 April 2021; doi: <https://doi.org/10.1038/s41436-021-01115-y>.

Chao EC, Astbury C, Deignan JL, Pronold M, Reddi HV, Weitzel JN and ACMG Laboratory Quality Assurance Committee. **Incidental detection of acquired variants**

in germline genetic and genomic testing: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* published online 16 April 2021; doi: <https://doi.org/10.1038/s41436-021-01138-5>.

Focused Revisions and Addenda:

Mintz CS, Seaver LH, Irons M, Grimberg A, Lozano R; ACMG Professional Practice and Guidelines Committee. **Focused Revision: ACMG practice resource: Genetic evaluation of short stature.** *Genet Med* published online 29 January 2021; doi: <https://doi.org/10.1038/s41436-020-01046-0>.

Bhatt S, Taylor AK, Lozano R, Grody WW, Griffin JH; ACMG Professional Practice and Guidelines Committee. **Addendum: American College of Medical Genetics consensus statement on factor V Leiden mutation testing.** *Genet Med* published online 05 March 2021; doi: <https://doi.org/10.1038/s41436-021-01108-x>.

ACT Sheets and Algorithms:

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Noninvasive Prenatal Screening via Cell-Free DNA ACT Sheet: Trisomy 13: Positive Cell Free DNA Screen.** Available at: <https://www.acmg.net/PDFLibrary/Trisomy-13.pdf>.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Noninvasive Prenatal Screening via Cell-Free DNA ACT Sheet: Trisomy 18: Positive Cell Free DNA Screen.** Available at: <https://www.acmg.net/PDFLibrary/Trisomy-18.pdf>.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Noninvasive Prenatal Screening via Cell-Free DNA ACT Sheet: Trisomy 21/ Down Syndrome: Positive Cell Free DNA Screen.** Available at: <https://www.acmg.net/PDFLibrary/Trisomy-21.pdf>.



Helping to Shape the Future of Newborn Screening Research: NBSTRN Educational Resources

by Amy Brower, PhD, Associate Project Director, Co-Principal Investigator; Kee Chan, PhD, MBA, Scientific Strategy Manager, and Ross Wiebenga, BS, Marketing Analyst

New discoveries in newborn screening (NBS) research have the potential to profoundly impact the health of four million babies born in the United States each year. In addition, in NBS the translation from discovery to population-based screening to clinical practice is rapid. These factors result in NBS research being one of the most rewarding, interesting and impactful areas of biomedical research. Medical geneticists and specialists who care for newborns with an NBS condition often lead these amazing efforts, and the NBSTRN is helping to shape the future of NBS research by providing innovative data tools and online tutorials.

NBSTRN’s new website features three key data tools designed to accelerate rare genetic disease research and educate the next generation of clinical investigators. First, the NBSTRN’s new data tool called the NBS Virtual Repository of States, Subjects, & Samples (NBS-VR, <https://nbstrn.org/tools/nbs-vr>) educates researchers on the policies and practices used by state NBS programs. The policies and practices may accelerate or slow research and the NBS-VR provides data on the number of conditions screened in each state/territory, the number of expected cases, incidence rates of conditions currently part of nationwide screening and conditions that are candidates for research pilots to expand NBS, and much more.

Next, to help the NBS community of researchers,



healthcare professionals, state NBS programs, families and advocacy groups better understand the genetic diseases that are part of NBS, NBSTRN developed an interactive data tool called the Newborn Screening Conditions Resource (NBS-CR, <https://nbstrn.org/tools/nbs-cr>). The NBS-CR provides a centralized resource of facts and statistics on both screened and candidate conditions. For NBS researchers, the NBS-CR offers clinical characteristics, diagnosis, management, clinical trial, and suggested reading resources through links to MedGen, an online portal developed and maintained by the National Library of Medicine (NLM) National Center for Biotechnology Information (NCBI).

Lastly, to facilitate comprehensive NBS long-term follow-up care and data collection efforts, the NBSTRN team at ACMG created the Longitudinal Pediatric Data Resource (LPDR, <https://nbstrn.org/tools/lpdr>). The LPDR was designed by leading geneticists who care for newborns with an NBS condition and enables the secure collection, aggregation, analysis, sharing and visualization of health information. The LPDR provides electronic case report forms that utilize question and answer sets, called common data elements (CDEs), developed by an expert group of researchers, clinicians, ethicists and newborn screening teams. The LPDR helps accelerate NBS-related research and foster the secondary use of the original data sets.

NBSTRN educational resources and interactive data tools are designed to shape the future NBS research. Visit NBSTRN.org to plan your next discovery!



Summer Genetics Scholars Program



The ACMG Foundation for Genetic and Genomic Medicine (ACMGF) is pleased to announce that the 2021 Summer Genetics Scholars Program (SGSP) will support ten students this summer, with two being supported by their host institution. This is a popular Foundation program that allows students who have completed their first year of medical school to work directly with a medical geneticist mentor. The six-week program educates future physicians about medical genetics and genomics in a “real world” setting and thanks to support from BioMarin, participating students receive a stipend.

SGSP was started in 2011 and has sponsored over 150 scholars at more than 30 different institutions around the country since then. This program has a lasting impact on participants. As one scholar wrote, “I feel so lucky to have been exposed to medical genetics at such a renowned institution, and to feel welcomed by such distinguished leaders in the field. I look back on that summer very fondly and still reference my notebook frequently!”

The ACMG Foundation thanks BioMarin and all past corporate partners who supported students and our ongoing efforts to expand the genetics workforce pipeline. Below is a list of the 2021 participating institutions and mentors.



Institution	Principal Mentor/ Faculty Member	Summer Scholars
Nationwide Children's Hospital Columbus, OH	Kim McBride, MD, FACMG	Hannah Layey
University of Alabama* Birmingham, AL	Nathaniel Robin, MD, FACMG	Meg Hayslip Mahnoor Asif
University of California, San Francisco San Francisco, CA	Anne Slavotinek, MD, PhD, FACMG	Rommell Noche
University of Iowa Iowa City, IA	John A. Bernat, MD, PhD, FACMG	Kristofer May
University of Maryland School of Medicine* Baltimore, MD	Carol Greene, MD, FACMG	Hannah Palmer Matthew Boden
University of North Carolina, Chapel Hill Chapel Hill, NC	William Hannah, MD	Amelia Loydpierson
University of Texas Health Science Center at Houston Houston, TX	Hope Northrup, MD, FACMG	Ariana Flores
University of Washington Seattle, WA	Fuki M. Hisama, MD, FACMG	Génesis Serrano-Rodríguez

*Host institution provided supported for a 2nd Summer Scholar

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