Education Edition | Summer 2022

THE ACMG Medical Geneticist

The Newsmagazine of the American College of Medical Genetics and Genomics

Letter From the President

The focus of this edition of *The ACMG Medical Geneticist* is education. Providing education for medical professionals and the public through high quality programs and materials has been at the heart of the ACMG's mission since its inception. This issue will highlight many of our current educational activities and the people who lead them.

In my message, I wanted to highlight two ideas about learning that may be less familiar to you and fall outside traditional educational approaches. One of these is brand new to me, while the other is one I've been working on for close to 20 years.

The new idea is one that was recently introduced to me by the new Dean of the Geisinger School of Medicine, Dr. Julie Byerly. In a discussion of incorporating genomics into the medical school and resident curricula for the system, the issue of lack of mentors among practicing physicians was raised as a barrier to learning. Dr. Byerly responded by saying that medical students and residents can serve as change agents and spur learning among practicing physicians. While new to me, this approach has been studied for over 10 years in the area of quality improvement (much of this work was done in the UK). Medical students now have formal training in quality improvement methods as part of their curriculum. By embedding projects in their clinical experiences, not only do the students become facile in the application of quality improvement methods, but it also seems to positively impact the practices of their clinical mentors. It's an intriguing idea to think about for genetics and genomics. The ACMG, through its support of medical student interest groups, is well-positioned to explore ways in which students can raise the awareness of genetics and genomics in their institutions. I'm looking forward to exploring this idea with our education leaders.

The second idea has been part of my research for over 15 years. How can we provide the answers to clinician questions about genetics and genomics at the point of care, as soon as the question arises? Better yet, how can we provide an answer to a genetics question the clinician didn't even think of, but is important to the care of that patient? The answer, at least at a superficial level, is through the bane of our existence, the electronic health record (EHR). While much has been written about the contribution of EHRs to clinician stress and burnout, there is an alternative reality where EHRs can deliver

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From the **Editor-in-Chief**

The theme of this edition of your newsmagazine is education. In his presidential message, Dr. Marc S. Williams explores two innovative educational ideas. The first is making medical students agents for change in raising awareness of genetics and genomics in their institutions. The second is embedding answers to questions about genetics and genomics in electronic health records so clinicians have point- of-care access.

The educational efforts of the NCC and NBSTRN are summarized on pages 38 and 36, respectively. These are valuable resources for both members and non-members, with the added benefit that many provide the opportunity to earn CME credits. In keeping with the education theme, it is noteworthy that for the summer of 2022, the ACMG Foundation Summer Genetics Scholars Program has its largest cohort of students to date (page 39). Eighteen institutions will sponsor 25 medical students to work in the field of medical genetics and genomics. Attracting students to consider careers in medical genetics is a key part of the ACMG Strategic Plan.

ACMG's most prominent education offering is our Annual Meeting (page 12). The 2022 Annual Clinical Genetics Meeting in Nashville was our first in-person meeting since the 2019 meeting in Seattle. The attendees were grateful for the excellent meeting content and for the opportunity to reconnect face-to- face with friends and colleagues. During the meeting, the ACMG Foundation presented nine awards including the David L. Rimoin Lifetime Achievement Award, the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award, the David L Rimoin Inspiring Excellence Award, the Richard King Trainee Award for Best Publication, the Carolyn Mills Lovell Genetic Counselor Award and four Next Generation Fellowship & Training Program Awards. Award details and recipients can be found on page 18.

The IDEA Committee of *Genetics in Medicine* held a workshop to discuss inclusion, diversity, equity and anti-racism initiatives at the journal (page 24). Another session of note was the Diversity Breakfast (page 33) which was an overwhelming success. Speaker Dr. Ericka Boone described imposter syndrome and discussed tools to confront imposter fears and thoughts. At the ACMG Foundation Day of Caring, adaptive bicycles and helmets were given to 12 children from the Angelman Syndrome and Kennedy Ladd Foundations (page 16). This is truly the most heartwarming event of the Annual Meeting and is the day we missed the most during the pandemic.

All in all, the Annual Meeting was a resounding success, and we look forward to the 2023 meeting in Salt Lake City!

Until next time,
Katy

Katy Phelan, PhD, FACMG, Editor
kphelan@flcancer.com

information at the point of care, "just in time" to answer critical clinical questions. Through understanding the condition of the patient through clues in the EHR, coupled with an appreciation of clinician workflow based on the current function of the EHR (e.g., laboratory results review, medication ordering, etc.) an inference can be made about what information might be needed and how and when it could be presented. The ACMG has anticipated this process in the ACT sheets, which not only include a narrative description of conditions and recommended actions, but have a flow chart that could allow computerization that would facilitate ordering and coordination of care through the EHR. These functions have yet to be fully implemented, but there is increasing interest about exploring these opportunities, which could greatly enhance the impact of the educational materials the ACMG creates. I've had initial conversations with several Fellows who have training and interest in informatics and this will be an area we will explore in the next few years.

As with all we do, we are grateful for all the energy and effort from our Fellows and Affiliate Members who are engaged in education of colleagues and patients every day. If you have special interest in the ideas described above, please feel free to reach out for more information on how to get involved.

Marc S. Williams, MD, FACMG
President

Meet ACMG's Newest Staff Members

Olivia Demarest, MS, MPH
Associate Methodologist

Olivia Demarest, MS, MPH joined ACMG in January 2022 as Associate Methodologist. She graduated with an MPH from the University of Michigan, School of Public Health, Health Behavior and Health Education with a concentration in Health Communication and a Master of Science in Human Genetics from the University of Michigan. She was a teaching assistant in the School of Nursing at the University of Michigan and a Patient Education Intern at St. Jude Children's Research Hospital, Memphis, TN. Olivia will be involved in the development of evidence-based guidelines and systematic evidence-based reviews at ACMG. Since joining ACMG, Olivia says, "I am thrilled to begin my career with ACMG and continue to learn from the best in the field. I hope to use my background in public health and genetics to bring breakthroughs in medical genetics to the public in a meaningful way."

Caroline Lumpkins, MBE
ELSI Research Associate, NBSTRN

Caroline Lumpkins, MBE joined ACMG as ELSI Research Associate for NBSTRN in November 2021. In this role, Caroline serves as the staff lead for NBSTRN's Bioethics and Legal Workgroup and provides ELSI (Ethical, Legal and Social Issues) support and review for NBSTRN projects. Prior to joining ACMG, Caroline served as the Education Programs Manager for the National Organization for Rare Disorders (NORD), a patient advocacy non-profit. During her time at NORD, she supported the development and implementation of educational programs and resources for patients and caregivers, medical professionals, researchers and students. Caroline received her Masters of Bioethics from Johns Hopkins University and Bachelor of Science in Health Science from Boston University. "I am very excited to be part of the ACMG team and support newborn screening research by providing guidance and education on the ethical, legal and social considerations arising from advancements in genetics, genomics and emerging biotechnologies," she said.

Myy Nguyen
Administrative Assistant

Myy joined ACMG in March 2022 as an administrative assistant. She was born in Danang, Vietnam, before moving with her family to Houston, Texas. She earned a BSA in Biology and Nonprofit Management from the University of Texas at Austin and has resided in Washington, DC since 2018. Her recent nonprofit work experience includes brief stints with the National Geographic Society and the Association of American Medical Colleges. During her free time, Myy likes to hike, read, and spend time with family. "I love the field of genetic counseling. I am excited to join the team at ACMG at such an interesting time for genetics and genetic counseling. My goal is to create change through meaningful actions and to use my experience to help people who do not have access to healthcare and especially, families who need a dedicated advocate for their health journey."

Zohreh Talebizadeh, PhD
Translational Research Manager, NBSTRN

Zohreh Talebizadeh, PhD joined ACMG as Translational Research Manager in January 2022. Zohreh brings over two decades of research experience to the College, including holding a faculty appointment at the University of Missouri-Kansas City School of Medicine and serving as a director of the autism genetics research laboratory at Children's Mercy Hospital. She has led several projects with a focus on identifying genetic/epigenetic risk factors and developing integrated approaches to address heterogeneity in autism. Her accomplishments include numerous research publications and an initiative to promote connecting genetics to outcomes research. She says, "As a genetic researcher, I embrace diversity and found it necessary to integrate the knowledge and perspectives not only from multiple scientific silos, but also from the populations impacted by these conditions. Joining ACMG has given me the opportunity to work toward this goal. I am excited to be a part of the leading medical genetics organization."

ACMG Thanks Outgoing Committee Chairs

During the Membership Business Meeting at the 2022 ACMG Annual Clinical Genetics Meeting, the College recognized and thanked five committee chairs who completed their volunteer terms of service. Committees are vital to advancing the work of the College and we invite you to join us in honoring these outgoing committee chairs for their hard work and commitment:

- Jennifer Bassetti, MD, FACMG: Chair, Membership Committee
- Sheila M. Dobin, PhD, FACMG: Chair, Advocacy and Government Affairs Committee
- Theresa A. Grebe, MD, FACMG: Chair, Social, Ethical and Legal Issues Committee
- Katy Phelan, PhD, FACMG: Chair, Diversity, Equity, and Inclusion Committee
- Myra Wick, MD, PhD, FACMG: Chair, Program Committee for Annual Meeting

Q&A with ACMG Education & CME Committee Chair Dr. John Bernat, FACMG, on Updates and New Initiatives to Enhance Educational Offerings of the College

ACMG's Education & CME Committee is chaired by John A. Bernat, MD, PhD, FACMG, Clinical Associate Professor in the Stead Family Department of Pediatrics, Division of Medical Genetics and Genomics at the University of Iowa. In a recent interview with *The ACMG Medical Geneticist*, Dr. Bernat discussed how the College is updating current educational programs and unveiling new initiatives designed to meet members' needs this year and in the future.

ACMG Medical Geneticist (ACMG): What are some of the most exciting current initiatives of the Education & CME Committee?

John Bernat (JB): We have a great deal of exciting work going on in the ACMG Education & CME Committee, but there is one very exciting initiative I'd especially like to highlight. We have a new UME/GME workgroup, led by Tracey Weiler, PhD, who is an experienced genetics educator of medical students at Florida International University. This workgroup is looking at ways to further involve and serve trainees at all levels, including medical students, residents, and fellows, to get them really involved in and excited about the field of medical genetics and genomics. One of their projects, which has been live for a couple of years now, is the ACMG Student Challenge, available through the ACMG Genetics Academy. Anyone (not just students!) can enroll in this education tool, and once they do, they're emailed a monthly challenge question that works through a clinical case and reviews a number of genetics topics. It's proven to be quite popular, with 407 participants from July 2020 to July 2021 and nearly 300 currently.

Our committee was also very involved in the ACMG's Annual Clinical Genetics Meeting; this year we led a workshop for early career genetics professionals, a student workshop on career pathways in genetics, and the always popular early career genetic mentor luncheon. (Editor's note – see full article about the 2022 ACMG Annual Clinical Genetics Meeting starting on page 12 in this edition.)

ACMG: Please tell us about the Gene Therapy Education Series scheduled for summer 2022.

JB: I believe the working title is "Advancements in Gene Therapy Options for Rare Diseases." This will be a 4-session series that would include a grand rounds-style webinar with a case presentation and panel discussion covering basics of gene therapy, different disease targets, and ethical/legal implications of gene therapy. These would be webcast live over the summer and also recorded and made available later this fall, and the target audience is both genetics professionals and non-genetics healthcare professionals. We recognize that gene therapy is coming into the limelight now with a number of gene therapies receiving FDA approval over the last few years and a growing number of trials for other therapeutic products. So, we want to educate health professionals about the "ins and outs" of gene therapy.

ACMG: An important part of the ACMG Strategic Plan (https://www.acmg.net/PDFLibrary/Strategic-Plan-Board-Approved.pdf) is to: "Develop customized education and resources for non-geneticists such as collaborative short courses at other specialty meetings." Can you talk about the new Genetics101 for Healthcare Providers course available now in the AMA EdHub™?

JB: We're really excited about this as an opportunity to focus on the main genetic issues in a number of different specialties and primary care. I understand there's a separate article in this magazine issue that discusses Genetics101 in more detail, so I won't spend too much time talking about it here other than to give a broad overview and to acknowledge the Course Director Shweta Dhar, MD, MS, FACMG, FACP and Jane E. Radford, MHA, CHCP, ACMG's Director of Education, who worked very hard to make this program come to fruition. Essentially, we have genetics professionals in various medical specialties presenting the key genetic issues for their specialty, and it's all available online through the AMA Ed Hub™. In each of the 10 modules, a board-certified medical genetics expert will provide a case-based presentation, along with supporting materials. These webinars are free and we invite our members to share them with their nongenetics colleagues and students. We appreciate the patience and support by an independent medical education grant from Illumina, Inc.

> **"We have a new UME/GME workgroup, led by Tracey Weiler, PhD, who is an experienced genetics educator of medical students at Florida International University. This workgroup is looking at ways to further involve and serve trainees at all levels, including medical students, residents, and fellows, to get them really involved in and excited about the field of medical genetics and genomics."**

ACMG: What other new and innovative programs are under way in the Education Department at ACMG?

JB: One of the things that comes to mind, and I don't have a lot of details on it yet, but members may recall that we had the ACMG Genomics Case Conference Series until the end of 2020. It was a monthly webinar that looked at innovations in genomic medicine, particular in areas of genetic and genomic testing. Our

committee was responsible for programming the series. Now we are looking at reformatting it to address other areas of genetic information. We hope that by the end of the year we will have this education program back as a regular series, the way it was previously.

We also have a number of updated course offerings. "Improving Patient Safety: An Imperative in Medical Genetics and Genomics" is a key component of the certification program for our genetics professionals, and it was updated just this past fall by our committee members. This course looks at several topics that impact patient healthcare safety, including contributing factors for medical errors. It had been around for a number of years and it was great to get that refreshed, since most of our membership goes through this course as part of their maintenance of certification cycle. We have other course offerings for some of the other genetics specialty training programs, including laboratory genetics and genomics, as well as the comprehensive genetics and genomics review course that is held every 2 years and available online. All of these offerings are available through the ACMG Genetics Academy. It really is a one-stop-shop for genetics information our members can binge watch.

"We recognize that gene therapy is coming into the limelight now with a number of gene therapies receiving FDA approval over the last few years and a growing number of trials for other therapeutic products. So, we want to educate health professionals about the "ins and outs" of gene therapy."

ACMG: Is there anything else you would like ACMG members to know about the educational offerings at ACMG?

JB: We're always looking to serve our membership, for opportunities to develop new content and new programming. So, if members have feedback about content offerings or anything our committee can help with in terms of knowledge gaps or needs, we ask them to please reach out to us.

CEO CORNER

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

There are numerous definitions of lifelong learning. Here are just a few: the process of gaining knowledge and skills throughout your life; the ongoing, voluntary, and self-motivated pursuit of knowledge for either personal or professional reasons; the continuing development of knowledge and skills that people experience after formal education. In medical genetics and genomics continuing education of current and future healthcare providers serves ONE goal: optimal patient care.

Education is the focus of this entire issue of *The Medical Geneticist* and is at the heart of ACMG's mission. The College contributes to educating its members and non-members at various stages of their careers, including undergraduate, graduate, and as medical students, clinical residents, laboratory fellows and board-certified medical geneticists.

Education was the focus of our in-person ACMG Annual Clinical Genetics Meeting in Nashville, TN, the first since 2019 in Seattle, WA. There was an abundance of clinical and scientific sessions. The excitement was palpable and best summarized by an email from one of the participants: "Being able to interact in person with friends and colleagues after a long dry spell is wonderful… I feel my soul is being nourished at this meeting." The Annual Meeting also included the announcement of four Next Generation Residency and Fellowship Awards, including two new awards, one in Laboratory Genetics and Genomics and one in Ophthalmic Genetics, as well as five other ACMG Foundation awards.

As ACMG's CEO, I am proud of all of our educational offerings including, 1) our journal *Genetics in Medicine*, 2) our Annual Meeting and 3) our wide-ranging online ACMG Genetics Academy with the most recent addition being the outstanding Genetics101 series.

Enjoy reading this issue of *The ACMG Medical Geneticist*.

Max Muenke

Maximilian Muenke, MD, FACMG

ACMG Evidence-Based Guidelines Program Spurs Member Involvement in Guiding Patient Care

by **Jennifer Malinowski, MS, PhD, Senior Methodologist**

The Evidence-Based Guidelines (EBG) Program is housed within the newly-created Practice Research and Methodology Department at ACMG. In addition to the EBG Program, ACMG methodologists work across ACMG departments, committees and workgroups, providing assistance with data analysis, project and manuscript review and research support. The department is staffed by Associate Methodologist, Olivia Demarest, MS, MPH, Methodologist Gabrielle Jenkins, MSPH and Senior Methodologist Jennifer Malinowski, MS, PhD.

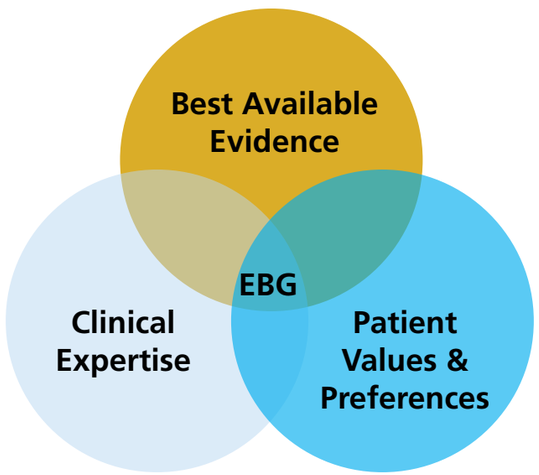
> **"Support from ACMG membership for the EBG program has been very enthusiastic. More than 80 individuals volunteered for SER and EBG workgroups for two projects: Autism Spectrum Disorder and Fatty Acid Oxidation Disorders, which both began in January 2022."**

Evidence-based guidelines are widely respected within the medical field and represent the intersection of clinical expertise, the best available evidence and patient-important outcomes (See Figure). Building on ACMG's strong legacy of guidelines created through expert consensus, the EBG program uses the Grading of Recommendations, Assessment, Development and Evaluation (GRADE) framework and best practices in guideline development to create recommendations that may be used in the United States and around the world.

Each ACMG EBG starts with the careful assessment of a topic by ACMG methodologists. Once the methodologists have completed their primary evaluation, a call for volunteers is sent out. For each new project, two workgroups are created: a Systematic Evidence Review (SER) Workgroup and an EBG Workgroup. Under the direction of ACMG methodologists, volunteers complete a SER in 9-15 months which results in a manuscript published in Genetics in Medicine. The EBG Workgroup receives all the training necessary to create a guideline using GRADE while the SER team is completing the SER. Once the SER results are finalized, the EBG Workgroup creates the recommendation statements and prepares a separate manuscript for publication. ACMG methodologists are trained to help workgroup members create EBGs, even in the absence of large cohort studies or randomized controlled trials. And all EBGs are informed by clinical expertise.

Support from ACMG membership for the EBG program has been very enthusiastic. More than 80 individuals volunteered for SER and EBG workgroups for two projects: Autism Spectrum Disorder and Fatty Acid Oxidation Disorders, which both began in January 2022. That response was exceeded during the next call for volunteers for projects scheduled to start in July 2022—with more than 100 applications from around the world. The EBG Program is an exciting opportunity for ACMG members to get involved with the College and help guide patient care.

For more information, please contact Jennifer Malinowski, Senior Methodologist at jmalinowski@acmg.net.



Current and Upcoming Educational Opportunities with the ClinGen Resource

by **Danielle Azzariti, MS, CGC; Jennifer Goldstein, PhD, CGC; Erin Riggs, MS, CGC; Courtney Thaxton, PhD**

ACMG is a proud partner of the National Institutes of Health (NIH)-funded Clinical Genome Resource (ClinGen), which is dedicated to building a central resource that defines the clinical relevance of genes and variants for use in precision medicine and research. The ClinGen Education, Coordination, and Training (ECT) Working Group would like to inform readers of their current educational opportunities and initiatives.

- During the 2022 summer, ClinGen hosted its second annual Clinical Genomics Career Panel Series. The multi-part, virtual series is available to undergraduate students, interns, and other interested learners to hear more about various careers in genomics.
    - See recordings from the 2021 panel: https://www.clinicalgenome.org/tools/genomics_careers_panel/
- Individuals that volunteer to participate and contribute to ClinGen are now issued training certificates of recognition for each completed curation activity training session through our Community Curation Database (CCDB: https://ccdb.clinicalgenome.org/login).
- Video recordings of presentations to the Biocurator Working Group (a forum for education and training for ClinGen biocurators) provide in-depth discussions from experts in the field on a diverse range of topics such as population databases, genome browsers, animal model databases, ClinGen frameworks and tools, and curation examples and challenges. These videos are available here: https://clinicalgenome.org/tools/educational-resources/.
- ClinGen Curation Modules are intended to provide learners with educational credit for participating in ClinGen curation activities and are available to individuals who are existing members of ClinGen Working Groups or Expert Panels and have had at least one curation previously approved within those groups. The current CEU/CME opportunities are:
    - ClinGen Gene-Disease Validity Curation Module: https://www.clinicalgenome.org/tools/clingen-gene-disease-validity-curation-module/clingen-gene-disease-validity-curation-module/
    - ClinGen Dosage Sensitivity Curation Module: https://www.clinicalgenome.org/tools/clingen-gene-disease-validity-curation-module/clingen-dosage-sensitivity-curation-module/
    - Continuing education opportunities for variant curation and actionability curation. (Currently in development)
- An upcoming webinar series (starting September 2022) will partner ACMG disease experts with ClinGen curation tool experts.

We look forward to maintaining these excellent educational resources and developing new and exciting tools.

Certification Matters! Upcoming Changes for ABMGG Diplomates

by **Carlos Bacino, MD, FACMG**
Professor and Vice Chair Clinical Affairs
Department of Molecular and Human Genetics,
Baylor College of Medicine
Secretary, ABMGG

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

In recent years, healthcare delivery and, concomitantly, medical genetics and genomics practice, have been profoundly impacted by rapid advances in technology and scientific knowledge. These advances have required healthcare providers to continue to find ways to stay current in their practice. To encourage this, modifications in the ABMGG continuing certification activities for diplomates are being implemented to facilitate ongoing learning and assessment.

For many years, CME credits were one of the few requirements to maintain board certification, but as rapid advances continue, the demand for activities to assure ongoing learning and proficiency have grown as well. CME may encourage learning, but additional elements are required to address key core competencies, including improvement in medical practice, patient care and procedural skills, systems-based practice, interpersonal and communication skills and professionalism. The goal of ABMGG's continuing certification program is to assess these components in a comprehensive fashion while allowing flexibility and integration of activities already part of a diplomate's day-to-day practice.

All ABMS member boards, including ABMGG, are revising their continuing certification programs. ABMGG is transitioning from a 10-year maintenance of certification cycle to a continuing certification program, with ongoing activities and a longitudinal assessment program (CertLink) allowing for formative and summative assessments. It allows for more flexibility, engagement and transparency. And in collaboration with ACMG, completion of Certlink assessment now grants up to 10 CME credits per year.

The new changes are intended to decrease the burden for practicing professionals, increase transparency by allowing them to assess how they are performing, and identify possible knowledge gaps to address. Navigating re-certification, continuing education and longitudinal assessments should be a simple, practical and meaningful process. These changes will require adjustments to be implemented by 2024, but the ABMGG is here to help you. We will provide webinars every 4 months to update our diplomates and respond to concerns. Contact us at any time at MOC@abmgg.org with any concerns or questions!

"The new changes are intended to decrease the burden for practicing professionals, increase transparency by allowing them to assess how they are performing, and identify possible knowledge gaps to address. Navigating re-certification, continuing education and longitudinal assessments should be a simple, practical and meaningful process."

2022 ACMG Annual Clinical Genetics Meeting Highlights

by **Myra Wick, MD, PhD, FACMG**
2022 Program Committee Co-Chair

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

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

Jane Radford, MHA, CHCP
Director of Education

The 2022 ACMG Annual Clinical Genetics Meeting was presented in hybrid format for the first time. The in-person meeting took place in Nashville, with online attendees joining sessions from 50 countries. A selection of 25 sessions, including all plenary sessions, concurrent scientific sessions and satellite symposia were live-streamed each day for real-time viewing. The remainder of the scientific sessions were captured in audio with a synch-to-slides format and was made available as on-demand content on the meeting platform shortly after the conclusion of each session. All of this content is now available in the 2022 Digital Edition available through the ACMG Genetics Academy.

With 3,131 participants comprised of 1,664 in-person (attendees, guests, and exhibit personnel) and 1,467 online, 2022 meeting attendance was close to pre-pandemic levels. The Exhibit Hall featured more than 100 exhibiting companies, close to 300 posters, plus Learning Lounges, Exhibit Theaters and a Wellness Pavilion.

Those participants who made the choice to participate remotely for reasons such as being located outside of the United States, institutional travel restrictions, work and family obligations or the continued uncertainty surrounding the state of the pandemic were able to access almost all sessions via the livestreams or on-demand.

The participants in Nashville were overjoyed to be meeting in person once again. The excitement of seeing and reconnecting with friends, colleagues and mentors was profound. With health and safety protocols in place – including proof of vaccination, adherence to the CDC Guidelines that were in place during the meeting dates, thorough cleaning and sanitation processes throughout the facility, and physical distancing in meeting room spaces whenever possible. There were zero reported positive Covid cases during or after the meeting.

Highlights from the 2022 Meeting

The meeting started on Tuesday with two Short Courses that were offered in person and to the online attendees. On Tuesday evening, the Public Health and the Medical Director's SIGs held interactive forums that included in-person and online attendees who joined in via Zoom.

The program featured several workshops, scientific plenary sessions each day and concurrent scientific sessions, specialty sessions and platform presentations for a total of 45 sessions throughout the meeting.

> **A highlight of the meeting, the Hackathon–style Session elicited feedback that included: "a fantastic workshop, with great discussion," "probably one of the most interesting sessions in the entire program" and "noteworthy, valuable and intelligent conversations among attendees and a good break from lecture-style sessions."**

The range of educational topics included both research and clinical topics that promote the science and the practice of clinical genetics and genomics. Sessions focused on the latest discoveries of the etiology and the pathogenesis of genetic disorders, the latest developments in genetic testing and screening, the laboratory's role in the diagnosis of genetic disorders, the treatment of genetic disorders in children and adults, the delivery of genetic services and more.

The 2022 Presidential Plenary Session, "From Exceptional to Routine: Transformation of Genomic Medicine in the 21st Century," moderated by ACMG President Marc S. Williams, MD, FAAP, FACMG, focused on the future of genomic medicine and the transformation of data into knowledge and, ultimately, clinical utility. The session also included discussion of increasing diversity in clinical genomics. Thursday morning's Plenary Session highlighted the

top four abstract submissions presented as Platform Presentations and was followed by three TED-style talks. Dr. Heidi Rehm presented on the story of ClinVar, Dr. Joseph McInerney on Mendel's legacy in recognition of his birth bicentennial and Dr. Ellen Clayton on the future of prenatal diagnosis. Other Plenary Sessions included: "Developing Recommendations for the Application of Direct-to-Consumer Genetic Testing in Clinical Care," "What are the Important Issues in the Arena of Public Policy and Legislation for Medical Genetics?," and the R. Rodney Howell Symposium, which focused on "Population Genomic Health: Expanding the Reach of Genomic Medicine to Diverse Populations." Many of the Plenary Sessions concluded with panel and audience discussions.

Scientific Concurrent Sessions included "Human Chromosomal Ring Disorders," "Hot Topics in Reproductive Diagnosis," "Health Disparities and Hereditary Cancer Syndromes," "Cardinal Signs" and "Down Syndrome Regression Disorder." The ACMG NBSTRN program was highlighted in "Exploring the Role of Medical Genetics and Genomics in Advancing Newborn Screening Research."

Specialty sessions included the annual Diversity, Equity, and Inclusion Breakfast planned by the ACMG DEI Committee (see separate article in this edition.) The session featuring a presentation by Dr. Ericka Boone on "Practical Tools on Confronting and Quieting Imposter Syndrome" is available on demand at no charge in the ACMG Genetics Academy at https://www.acmgeducation.net.

The ACMG Program Committee presented the Genetic Counselors Forum, which reviewed the genetic counseling, medical and ethical considerations as genome sequencing emerges as a clinical tool for rapid diagnosis. Clinical case examples provided the background for understanding the implications of this technology and the session concluded with a panel discussion.
In addition to the Plenary Presentations, an additional 60 abstracts were presented in the form of Platform Presentations and over 300 posters were on display in the exhibit hall with an additional 200 available as ePosters.

Several opportunities were offered for students and geneticists-in-training, including a lounge where early career professionals had the opportunity to rest and recharge, meet with College leadership and network with peers. A Welcome Reception held on Tuesday evening provided a relaxed atmosphere for students and trainees. The Early Career Workshop provided attendees with important tips on pursuing the next stage in their genetics career and focused on the application and interview process for fellowships and early career faculty positions. The Early Career Genetic Mentor Luncheon offered the opportunity for nearly 100 trainees to talk informally with senior members of the College about career options, goals and professional opportunities. The Special Student Session provided medical students, undergraduate & graduate students and genetic counseling students with the opportunity to learn more about careers in the field of medical genetics including various component training and career pathways.

ACMG is grateful to the industry partners that provided support for many of the services and events of the meeting and the eight companies that presented non-accredited Industry Workshops and accredited Satellite Symposia during the meeting.

> **The 2022 ACMG Annual Meeting was abuzz with enthusiasm and gratitude as many of us met 'in 3D' for the first time since the start of the pandemic. Nashville was a wonderful setting with the fantastic convention center, great accommodations and signs of spring for those of us from the 'northern climates.' We enjoyed the cutting-edge science and many new features including a Hackathon-style session and the Wellness Pavilion that featured therapy dogs for attendees to enjoy."**

Special thanks go to the 2022 Program and Education Committees for their significant contributions towards the success of this year's meeting.

Myra Wick, the 2022 Program Committee Chair, summed up her experience saying, "The 2022 ACMG Annual Meeting was abuzz with enthusiasm and gratitude as many of us met 'in 3D' for the first time since the start of the pandemic. Nashville was a wonderful setting with the fantastic convention center, great accommodations and signs of spring for those of us from the 'northern climates.' We enjoyed the cutting-edge science and many new features including a Hackathon- style session and the Wellness Pavilion that featured therapy dogs for attendees to enjoy. We are grateful to the ACMG staff for their tireless efforts in bringing this meeting to fruition."

Video highlights from the meeting and the Day of Caring can be viewed on the ACMG YouTube channel at https://www.youtube.com/user/TheACMGChannel.

Day of Caring

During the 2022 ACMG Annual Clinical Genetics Meeting in Nashville, the ACMG Foundation shared smiles, happy tears and bicycles with 12 families and kids at the Day of Caring – marking a heartwarming return to the full in-person event for the first time since 2019 due to the pandemic. Customized bicycles, as well as helmets, were presented to Nashville-area children with genetic conditions from the Angelman Syndrome Foundation and the Kennedy Ladd Foundation.

The ACMG Foundation Day of Caring is supported by PerkinElmer and the ACMG Foundation for Genetic and Genomic Medicine. Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation; Lora Bean, PhD, FACMG, senior director of quality assurance for PerkinElmer; as well as some parents of the children conducted media interviews during the event. Please enjoy the report filed by WSMV (NBC in Nashville) and our own Day of Caring highlight video on the ACMG YouTube channel at www.youtube.com/theacmgchannel.

Hackathon–style Session Recap

The 2022 meeting welcomed a new session style to the agenda and the first ACMG Hackathon-style session – "How Can We Maximize the Yield of Genetic Disease Diagnostic Evaluation and Testing?"

Led by Robert Wilden, MD, FACMG and Heidi Rehm, PhD, FACMG, and attended by approximately 60 participants, the session began with a 10-minute stage-setting lecture. Then the audience worked in teams of 4-6 to brainstorm innovations (novel, or combinatorial technological and operational) to meaningfully maximize and optimize clinical diagnostic yield. Each team selected an area of focus:

- Laboratory operations (sample collection, billing, prior authorization, reimbursement, lab reporting, data flow)
- Clinical workup (Physical workup; tools, ontologies and approaches for collecting and transmitting phenotypic data; role of clinician in making a molecular diagnosis; EHR)
- Variant and gene evidence collection and generation (segregation, population data, case observations, functional assays, in silico/ML/AI, data sharing approaches)
- Patient and physician education
- Genetic counseling and return of results

Factors taken into consideration included advances in technologies, informatics, clinical valuation, education, data sharing and stronger connections to research.

Next, a member of each team presented at least one bold idea for increasing genetic diagnostic yield and how it might be implemented. Most proposals spanned four major areas of focus including:

1) improved collection of phenotypic data
2) more advanced technologies to enhance the accuracy of genomic sequencing and complementing it with additional functional and other omic data
3) addressing patient engagement and equitable access to genomic sequencing tests, and
4) expanding data sharing to improve interpretation

These presentations were then followed by open discussion, thoughtfully contemplating the proposals as well as challenges and opportunities in addressing the topic. Attendees left inspired to take these new ideas back home and to think about the ways in which they could implement the concepts.

A highlight of the meeting, the Hackathon–style Session elicited feedback that included: "a fantastic workshop, with great discussion," "probably one of the most interesting sessions in the entire program" and "noteworthy, valuable and intelligent conversations among attendees and a good break from lecture-style sessions."

The ACMG Foundation for Genetic and Genomic Medicine Presents Nine Awards during the 2022 ACMG Annual Clinical Genetics Meeting

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

The ACMG Foundation for Genetic and Genomic Medicine was established in 1992 to support the ACMG mission to "translate genes into health." Through its work, the ACMG Foundation fosters charitable giving, promotes training opportunities to attract future medical geneticists and genetic counselors to the field, shares information about medical genetics and genomics, and sponsors important research.

Each year at the ACMG Annual Clinical Genetics Meeting, the ACMG Foundation presents an array of awards and scholarships. The College and Foundation are pleased to recognize here the nine individuals who received 2022 awards. To read the complete news announcements, visit www.acmgfoundation.org.

To learn more about the vital work of the ACMG Foundation and how you can support our mission, visit www.acmgfoundation.org or contact Karl Moeller at acmgfoundation@acmg.net.

Elaine H. Zackai, MD, FACMG addresses the audience at the 2022 ACMG Annual Clinical Genetics Meeting upon receiving the ACMG Foundation for Genetic and Genomic Medicine David L. Rimoin Lifetime Achievement Award.

The David L. Rimoin Lifetime Achievement Award in Medical Genetics

Revered clinical geneticist and pediatrician Elaine H. Zackai, MD, FACMG received the 2022 ACMG Foundation for Genetic and Genomic Medicine's David L. Rimoin Lifetime Achievement Award in Medical Genetics. Dr. Zackai, a fellow of the College of Physicians of Philadelphia and a founding fellow of ACMG, directs the Clinical Genetics Program at the Children's Hospital of Philadelphia (CHOP) and holds the Letitia B. and Alice Scott Endowed Chair in Human Genetics and Molecular Biology. She was chosen for this award to recognize her renowned expertise diagnosing birth defects and genetic disorders; her compassionate and resourceful care for patients and families; her prolific research collaboration with colleagues around the world; and her mentorship and teaching of clinicians, researchers, and genetics counselors who have in turn modeled their work on her standards for excellence.

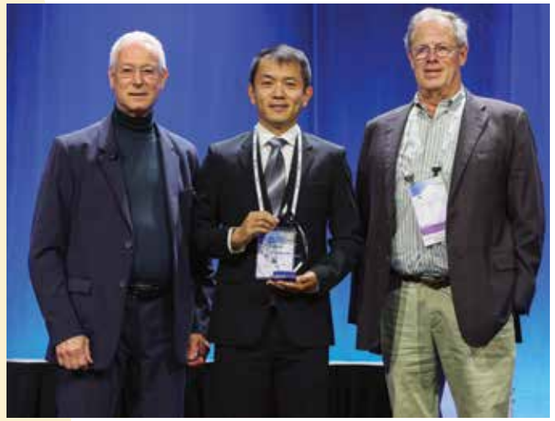
Touched and honored by the news that she was selected for the David L. Rimoin Lifetime Achievement Award, she explained the significance of Dr. Rimoin's role in her training, going back to when she was one of his first fellows. "I have modeled my whole career around what I learned from David Rimoin, always putting the patient first and treasuring the exceptions that don't fit the obvious diagnoses," Dr. Zackai said. "I spent hours xeroxing his entire set of journal articles before he left St. Louis to become an attending in California. This was back in the day before computers! They are still one of my most valuable tools and when I use them, I think of him and what he would have done for the patient I have at hand. I would not be where I am today without the essential foundation that he imparted to me."

President of the ACMG Foundation, Bruce R. Korf, MD, PhD, FACMG said, "Dr. Zackai has trained at least a generation of clinical geneticists and provided care to children and families dealing with genetic disorders for decades. Her commitment, compassion, and expertise are precisely in keeping with the values we seek to recognize in the David L. Rimoin Lifetime Achievement Award in Medical Genetics. Indeed Dr. Zackai began her genetics training with Dr. Rimoin, making this award all the more appropriate."

"Dr. Rimoin's widow, Dr. Ann Garber-Rimoin said, "It is with pleasure that the Rimoin family honors Elaine Zackai, MD, FACMG with the 2022 David L. Rimoin Lifetime Achievement Award in Medical Genetics. Dr. Zackai's outstanding career is aligned with the qualities that characterized David's career in medical genetics, including her excellence in teaching and mentoring, her ability to apply lab-based genomics into her clinical practice, and her gift for connecting with patients and their families. We congratulate Dr. Zakai as the recipient of the Rimoin Lifetime Achievement Award for 2022."

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

Pengfei Liu, PhD, FACMG is the recipient of the 2022 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award. He is an Assistant Professor at the



Pengfei Liu, PhD, FACMG (center), recipient of the ACMG Foundation's 2022 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award, is flanked by ACMG Foundation President Dr. Bruce R. Korf, FACMG (left) and Michael S. Watson, PhD, FACMG.

Molecular and Human Genetics Department at Baylor College of Medicine (BCM), the Director of the ACGME-accredited Laboratory Genetics and Genomics (LGG) Fellowship Training Program at BCM, and an Associate Clinical Director at the Baylor Genetics diagnostic laboratory. A board-certified laboratory geneticist who holds a PhD in molecular and human genetics from BCM, Dr. Liu's scientific contribution includes utilizing clinical diagnostic big data to generate knowledge that advances genomic science, as well as developing novel approaches to improve the implementation of genomic medicine. He is one of the recipients of the Genomic Innovator Award from the National Human Genome Research Institute and has been active in many NIH-funded team science projects.

"It is my greatest privilege and honor to receive the Michael S. Watson Genetic and Genomic Medicine Innovation Award from the ACMG Foundation. The last 10 years have been a period of dramatic innovation in our field of human and medical genetics," said Dr. Liu. "I feel so fortunate to have received my training and started my career during this time. I am excited to continue this journey to translate cutting-edge technologies to clinical implementation, and to improve the utilization of diagnostic big data."

"Dr. Liu has been at the forefront of developing approaches to harness large genomic data sets to improve our ability to diagnose genetic disorders. He is a true innovator in our field, and, as such, highly deserving of recognition as the recipient of this award," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation.

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

Boxun Zhao, PhD, received the 2022 ACMG Foundation/David L. Rimoin Inspiring Excellence Award during the 2022 ACMG Annual Clinical Genetics Meeting.

Boxun Zhao, PhD is the recipient of the 2022 ACMG Foundation/David L. Rimoin Inspiring Excellence Award. Dr. Boxun Zhao obtained a PhD in Genetics in 2017 from Peking Union Medical College and Tsinghua University in Beijing, China. He joined the labs of Drs. Timothy W. Yu and E. Alice Lee in the Division of Genetics and Genomics at Boston Children's Hospital in 2018 as a postdoctoral fellow. Dr. Zhao received the award for his featured platform presentation at the 2022 ACMG Annual Clinical Genetics Meeting, "Discovery and therapeutic implications of pathogenic retroelements in neurodegenerative diseases."

Dr. Zhao's research interests include: 1) basic research: somatic transposable element (TE) insertions in the human brain and neurological disorders 2) molecular diagnosis: developing a specialized pipeline to capture the full spectrum of genetic variation to resolve un- or half-diagnosed genetic cases, and 3) genomic medicine: developing individualized medicines for patients with diseases that currently are considered too rare for traditional commercial investment. His long-term goal is understanding the importance and functional impact of TE insertions in human diseases; and translating scientific discoveries into therapeutics.

"Congratulations to Dr. Boxun Zhao, the 2022 recipient of the David L. Rimoin Inspiring Excellence Award! Dr. Zhao's work on diagnosis and individualized treatment for rare genetic disorders is an exciting new direction in the study of rare disorders that were so important to Dr. Rimoin throughout his career," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation for Genetic and Genomic Medicine.

The Richard King Trainee Award

Kushani Jayasinghe, MBBS, is the recipient of the 2022 Richard King Trainee Award for Best Publication by a Trainee in *Genetics in Medicine*.

Kushani Jayasinghe, MBBS is the recipient of the 2022 Richard King Trainee Award for Best Publication by a Trainee in *Genetics in Medicine* (*GIM*). Dr. Jayasinghe is a nephrologist at Monash Health, Melbourne and is undertaking clinical genetics training at the Royal Melbourne Hospital in Melbourne, Australia. She received the award for her published article, "Clinical impact of genomic testing in patients with suspected monogenic kidney disease," which was published online in *GIM* in January 2021.

"I am delighted to receive the Richard King Award. Genomic testing in kidney disease is still not routinely implemented into clinical practice. This study shows that genomic testing is clinically useful in terms of providing a diagnosis and also has significant management implications for patients. I am very grateful for the many researchers and clinicians who contributed to this study, and my mentors who have supported me through my research and training," said Dr. Jayasinghe upon receiving the award.

"We had many outstanding articles published by trainees this year. It was a real pleasure going back to re-read those articles as part of the selection process. *GIM* is delighted to name Dr. Kushani Jayasinghe as this year's worthy recipient of the Richard King Award," 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* and serving as the founding editor-in-chief of the journal. The award was established by the ACMG Foundation to encourage ABMGG, international equivalents, or genetic counseling trainees in their careers and to foster publication of the highest quality research in *Genetics in Medicine*.

The ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award

Carly Peterson is the recipient of the 2022 ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award. Ms. Peterson received the Lovell award for her platform presentation at the 2022 ACMG Annual Clinical Genetics Meeting, "Parenting Stress Raising Children with Sex Chromosome Aneuploidies: First Year of Life Results from the eXtraordinarY Babies Study." A genetic counseling graduate student at the University of Colorado Anschutz Medical Campus who will earn her Master of Science degree in genetic counseling in May 2022, Ms. Peterson's specific interests lie in the psychosocial aspects of genetic counseling, and in providing support and resources for patients throughout the genetic testing process.

ACMG Foundation's Next Generation Fellowship & Training Program Awards

Carly Peterson receives the 2022 ACMG Foundation Carolyn Mills Lovell Genetic Counselor Award from ACMG Foundation President Dr. Bruce R. Korf, FACMG.

Upon receiving the award, Ms. Peterson said, "I am incredibly honored and grateful to receive the 2022 Carolyn Mills Lovell Genetic Counselor Award. This research would not have been possible without the insightful contributions and support of my research team: Talia Thompson, PhD, Nicole Tartaglia, MD, Shanlee Davis, MD, and Susan Howell, MBA, MS, CGC. Thank you to the ACMG Foundation for selecting our abstract to receive this award."

"Congratulations to Carly Peterson as the recipient of this year's award, for the highest scoring abstract by a genetic counselor. This year's award is especially unique, as Ms. Peterson is still a genetic counseling student. I am sure that this is just the first of many accomplishments in her career," 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. Their research demonstrates the important role genetic counselors play in clinical research in medical genetics and improving delivery of genetic services."

Ibrahim Elsharkawi, MD/MB BCh BAO (NUI); Jessica Priestley, MD, PhD; Nikhil Sahajpal, PhD; and Emile Vieta, MD each received 2022 Next Generation Fellowship & Training Program Awards. These "Next Gen Awards" will support these medical genetics professionals with one year of postgraduate training. Support for this year's class of Fellows was generously provided by Bionano Genomics, Spark Therapeutics, Takeda, and Sanofi-Genzyme.

Dr. Elsharkawi, recipient of a Medical Biochemical Genetics Subspecialty Fellowship, is currently completing a one-year fellowship in Mitochondrial Medicine at Children's Hospital of Philadelphia, to be followed by a one-year Medical Biochemical Genetics fellowship at Boston Children's Hospital. Upon receiving his award, Dr. Elsharkawi said, "I am absolutely thrilled to have been given this prestigious award by the ACMG Foundation, which will allow me the opportunity to build on what I am learning during my current Mitochondrial Medicine fellowship and to carry it forward into my Medical Biochemical Genetics fellowship. My dream of becoming a medical geneticist in the USA began when I first set foot in the USA in 2013 as a visiting medical student rotating at Massachusetts General Hospital."

Dr. Priestly also received a Medical Biochemical Genetics and is currently completing her combined Pediatrics/ Medical Genetics training at the Children's Hospital of Philadelphia, where she will remain for her fellowship next year. ""I am delighted and honored by the support the ACMG Foundation Next Generation Award provides for both my professional development and research interests in providing high quality genetic care to diverse populations," said Dr. Priestly. "I am grateful to the selection committee for this recognition, my team of mentors throughout my training who have served as role models and fostered my sense of curiosity, especially Dr. Rebecca Ganetzky, my inspiring patients and their families, and my own family, especially my daughter Rosalyn."

From left, Jessica Priestley, MD, PhD and Ibrahim Elsharkawi, MD/MB, BCh, BAO, (NUI), recipients of 2022 Next Generation Fellowship Awards, with ACMG Foundation President Dr. Bruce R. Korf, FACMG.

Dr. Sahajpal, recipient of the Laboratory Genetics and Genomics Subspecialty Fellowship Award, is a post-doctoral fellow in the Department of Pathology, Augusta University, GA and an ABMGG LGG fellow at the Greenwood Genetic Center (GGC) in Greenwood, SC. The Next Generation Award will further his career development at the GGC. "As an incumbent ABMGG LGG fellow at the Greenwood Genetic Center (GGC) in Greenwood, SC, I feel delighted and honored to receive this prestigious Next Generation Fellowship and Training Award from the ACMG Foundation. I am humbled by this amazing opportunity given to me by the selection committee of the ACMG Foundation to further my career development in the field of laboratory genetics and genomics at the GGC. I am proud to be part of the ACMG community as I start my fellowship at the GGC to become a laboratory Director to advance clinical diagnostic testing and support patients and families affected with genetic disorders," he said.

Dr. Vieta, who received the first-ever Ophthalmic Genetics Subspecialty Fellowship awarded by the ACMG Foundation, will undertake training in both Ophthalmology and Medical Genetics at the University of California at Los Angeles - Jules Stein Eye Institute. "It is truly an honor to receive this award," he remarked. "I would like to wholeheartedly thank my family, my co-workers, my mentors, the ACMG, and the ACMGF. It is a privilege to learn and work in this field as well as to have the support of such wonderful mentors. I look forward to the work ahead."

"There has never been a more exciting time in our field of genetic and genomic medicine, with new approaches to prevention, diagnosis, and treatment of rare and common disorders. The need for training the next generation of clinical and laboratory geneticists has never been greater, so we are especially grateful to Pfizer for their continuing support of the Next Generation Fellowship Award," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation.

From left, Emile Vieta, MD; ACMG Foundation President Dr. Bruce R. Korf, FACMG; Nikhil Sahajpal, PhD; and Bionano Genomic's Erik Holmlin, PhD. Drs. Vieta and Sahajpal received Next Generation Fellowship Awards.

Differing IDEAs: New Inclusion, Diversity, Equity and Anti-Racism Initiatives at *Genetics in Medicine*

by **Mildred K. Cho, PhD** and **Maria Laura Duque Lasio, MD, FACMG**

As many academic and medical institutions reckon with their part in the history and perpetuation of scientific racism, they have begun to address Diversity, Equity and Inclusion (DEI) issues. *Genetics in Medicine*, the official journal of the ACMG, has taken a leadership role among journals in tackling these issues on multiple fronts. In 2021, the journal published an article articulating principles of anti-racism in publishing of human genetics and genomics research.[1] Since then, the Inclusion, Diversity, Equity and Anti-Racism (IDEA) Committee of the journal has begun to implement these principles.

First, the journal is now tracking diversity by gender, race, ethnicity, disability and career stage for its editors, staff, peer reviewers and authors. Second, *GIM* revised its mission statement to specifically encourage research that combats racism, includes diverse study populations, and is written by authors from diverse and underrepresented backgrounds. The mission statement also clarified that articles reporting observations from a limited geographic region would not necessarily be viewed as lacking significance if they filled a knowledge gap about underrepresented populations or health disparities. In response to recent concerns about the reported use of genetic samples taken without consent or for surveillance purposes,[2] *GIM's* IDEA and Ethics Advisory Committees collaborated to expand the authors' declarations to state that samples are collected and used ethically.

At the 2022 ACMG Annual Meeting in Nashville, IDEA Committee members held a workshop, "An Action Plan for Inclusion, Diversity, Equity and Anti-Racism in Publishing: Putting IDEAs into Practice at *Genetics in Medicine*." This workshop discussed the DEI challenges faced by those involved in human research, especially in the field of genetics and genomics, and presented specific actions that authors, reviewers and editors could take to enhance IDEA in publishing. A recurring theme of the panel was the need for genetic researchers to acknowledge that commonly-used categories of race and ethnicity, especially those required by the U.S. federal government, "reflect a social definition of race/ethnicity recognized in this country and not an attempt to define it biologically, anthropologically, or genetically."[3] Furthermore, that the language and terminology used to describe race, ethnicity and ancestry are continuously evolving and differ around the world. Another recurring theme was that researchers should define the terms used to describe populations, being as specific as possible, and consulting communities about their preferred descriptors. Because some of these labels are assigned at the point of care, clinicians and health care institutions also need to be involved in IDEA-based changes. These recommendations apply to descriptors of race, ethnicity, gender, disability and other variables.

The workshop also provided examples of how to incorporate assessment of systemic racism and health disparities into study design. Panelists had recommendations for using conceptual models to select study variables that may elucidate mechanisms through which systemic racism operates. The panel also presented implementation frameworks and evaluation methods that include equity domains such as cultural factors and patient-provider interactions to better promote access to and utilization of genomic medicine. This workshop highlighted that enhancing inclusion, diversity, equity and anti-racism in publishing will require concerted efforts of the entire biomedical research and clinical enterprise.

[1]Brothers KB, Bennett RL, Cho MK. (2021) Taking an Anti-Racist Posture in Scientific Publications in Human Genetics and Genomics. *Genetics in Medicine*. https://doi.org/10.1038s/41436-021-01109-w
[2]https://theintercept.com/2021/08/04/dna-profiling-forensic-genetics-journal-resignations-china/
[3]US Census Bureau (2021) 2020 Census Frequently Asked Questions About Race and Ethnicity. US Department of Commerce. Washington, DC. https://www.census.gov/programs-surveys/decennial-census/decade/2020/planning-management/release/faqs-race-ethnicity.html

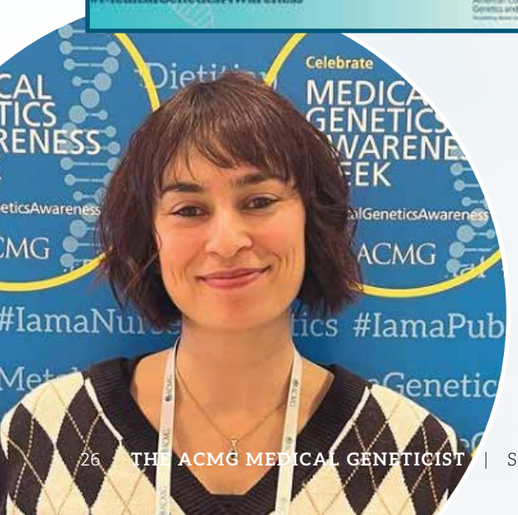


Members of the IDEA Committee present their workshop at the 2022 ACMG Annual Meeting in Nashville. Far left: Maria Laura Duque Lasio, MD, FACMG; far right: Robin L. Bennett, MS, CGC.

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

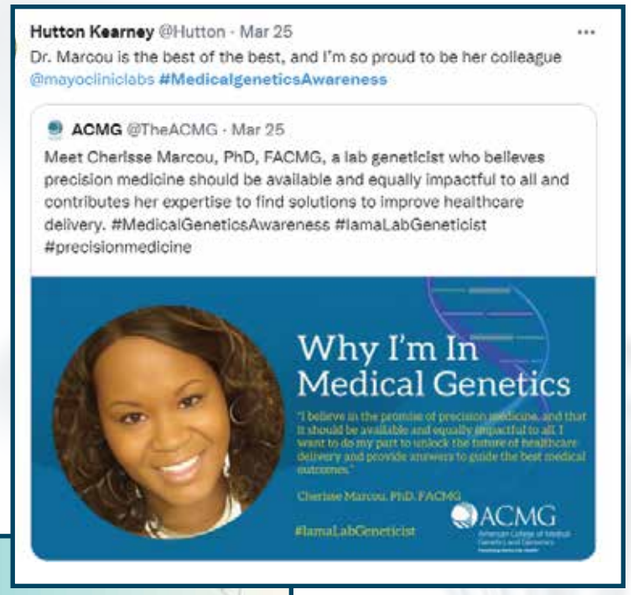
"Have you ever seen someone more excited to be a medical geneticist?!? I didn't know this specialty even existed until my fourth year of medical school! This is why #MedicalGeneticsAwareness is so important!" – Dr. Elle Geddes, associate professor of Clinical Medical & Molecular Genetics at the Indiana University of Medicine.

With that Twitter post, Dr. Elle Geddes crystallized the spirit and purpose of Medical Genetics Awareness Week, which took place March 22-25. Accompanied by a photo of herself posed in front of the Medical Genetics Awareness Week selfie wall at the ACMG Annual Clinical Genetics Meeting, her post conveys both pride and the need to shine a spotlight on our field. During the week, we take time to honor the skills and commitment of all of those who work to translate genetic and genomic discoveries into better patient care and public health.



"Medical Genetics – an extraordinary and complex specialty! I am so happy this specialty chose me" – Dana Schroeder, APRN, CNP, Genetics Specialist, Sanford Health, Sioux Falls, SD.

Thank you to everyone who participated in this year's celebration – the fourth Medical Genetics Awareness Week since 2019. We saw participation from a wide array of medical genetics professionals – from clinical and laboratory geneticists to genetic counselors, nurses, physician assistants and others. We distributed hundreds of temporary tattoos and more than one thousand hashtag buttons. Our Zoom virtual backgrounds were frequently seen before and during the ACMG Annual Meeting. In the month of March alone, our Medical Genetics Awareness Week hashtags garnered more than 3.2 million impressions on Twitter, Facebook and Instagram combined. On Twitter alone, 200 unique authors tweeted nearly 500 times using the #MedicalGeneticsAwareness hashtag,



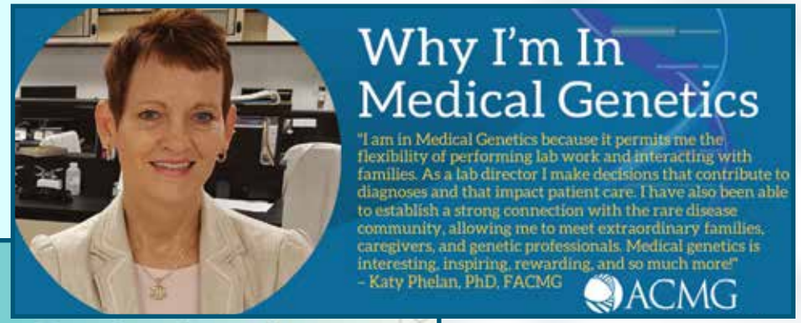
Our goal to increase engagement from students and generate participation from a diverse population of medical geneticists around the world was a success! Six of the top 10 Influencers on Twitter for the #MedicalGeneticsAwareness hashtag were from accounts representing student or minority organizations, including two medical schools

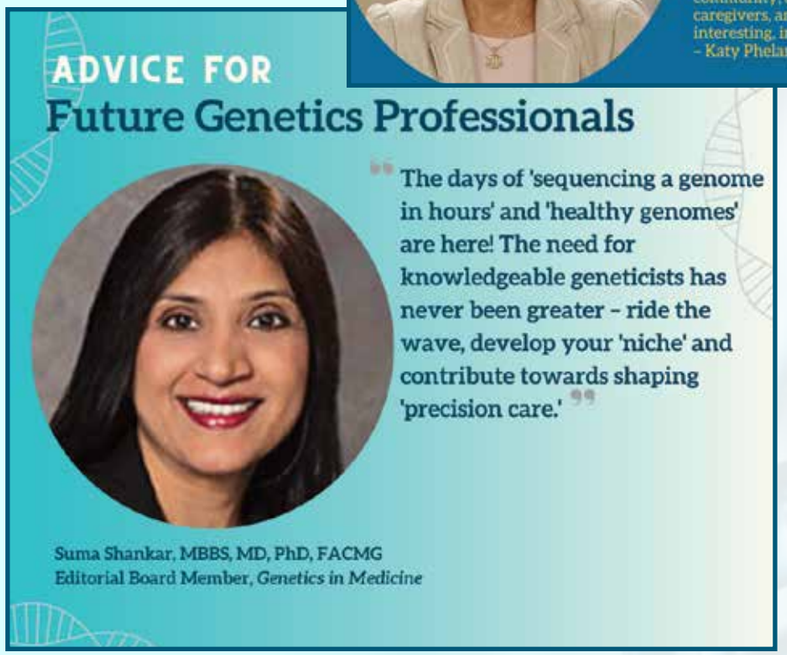
> "I am a rising 3rd year medical student and hopeful for a career in medical genetics as a #FutureGeneticsProfessional so that I can be the doctor for all of those who fall under the label of rare." – Francesco Sautto, president of the Medical Genetics Student Interest Group at the University of Central Florida College of Medicine.

at Historically Black Colleges and Universities. Of course, social media engagement from across the U.S. was prevalent, but we were thrilled to see #MedicalGeneticsAwareness posts from more than 20 countries.

There was participation from current and future genetics healthcare professionals, professional societies, commercial laboratories and others. Visitors to our #MedicalGeneticsAwareness selfie wall in the Exhibit Hall at the ACMG Annual Meeting displayed pride and had fun posing for photos with colleagues. The engagement around Diversity Day, Student and Trainee Day and Resource Day—inspired us and we hope it did the same for you.

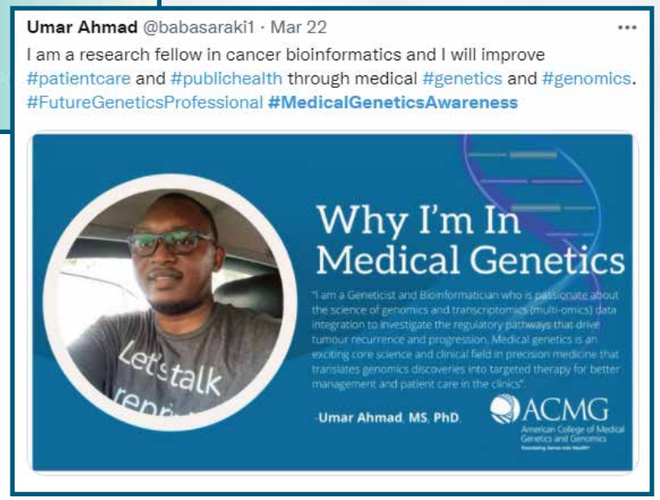
A special thank you goes out to the ACMG Board Members, the DEI Committee and the Genetics in Medicine Editorial Board, who contributed "Why I'm in Medical Genetics" and "Advice for Future Genetic Professionals" statements that got the ball rolling for social media testimonials by other participants. In addition, we thank ACMG CEO Dr. Max Muenke, FACMG and ACMG President Dr. Marc S. Williams, FACMG who contributed introductory and wrap-up videos, respectively. If you are on social media, we encourage you to search for these posts using #MedicalGeneticsAwareness.





As we look forward to next year's Medical Genetics Awareness Week, which will take place March 14-17, 2023, we invite you to visit our Medical Genetics Awareness Week web pages at www.acmg.net/MedicalGeneticsAwareness. On these pages, you will find tools and tips to help you promote awareness all year long. As President Williams noted in his closing video, "While we always enjoy celebrating Medical Genetics Awareness Week, it's important to recognize that Medical Genetics Awareness is a full-time job…we need to be thinking about it all the time."

> **"We say goodbye to Medical Genetics Awareness Week, inviting everyone to learn more about this discipline and the interdisciplinary work that is carried out to improve the health and well-being of human beings." (Translated from Spanish) – Dr. Oriana Batista, Director, Centro Gendiagnostik, Panama**



Top 10 Influencers on Twitter for #MedicalGeneticsAwareness

1. @theACMG
2. Minority Genetic Professionals Network
3. NCC for the Regional Genetics Networks
4. Howard University College of Medicine Dean
5. Charles R. Drew University of Medicine and Science
6. American Academy of Physician Associates.
7. American Medical Student Association
8. BlackInGenetics
9. Genetics In Medicine Journal Latino Medical Student Association
10. Latino Medical Student Association

Top 10 Countries Participating on Twitter for the #MedicalGeneticsAwaress

1. USA
2. Canada
3. Indonesia
4. United Kingdom
5. Brazil
6. France
7. Netherlands
8. Colombia
9. India
10. Italy

Source: Symplur Healthcare Hashtag Project

ACMG Members Meet with Students at Vanderbilt University

by **Fuki M. Hisama, MD, FACMG** and **Cynthia Morton, PhD, FACMG**

During the 2022 Annual Meeting, five leaders in the field of Medical Genetics visited Vanderbilt University in Nashville on Friday afternoon to share career advice and their enthusiasm for medical genetics with over 30 undergraduate students. Dr. Karina Gonzalez Herrera and Dr. Xavier du Maine from the Harvard Graduate School of Arts and Sciences Office of Diversity and Minority Affairs organized a hybrid in-person and virtual event and reached out to Dr. Cynthia Morton (Harvard) and Dr. Fuki M. Hisama (University of Washington) to lead the session. They invited colleagues with varied roles in clinical care, research, and education including: Dr. Bruce Korf, (Associate Dean for Genomic Medicine, University of Alabama); Dr. Katy Phelan, (Director of Genetics, Florida Cancer Specialists and Research Institute) and Dr. Gail P. Jarvik, (Division Head and Professor of Medical Genetics, and Genome Sciences at the University of Washington, and immediate past president of the ASHG) to participate in a panel discussion on careers in medical genetics. Dr. Kathy Friedman and Dr. Doug McMahon, the Co-Directors of the Maximizing Access to Research Careers (MARC) Scholars Program at Vanderbilt University hosted the event and invited undergraduate students from Vanderbilt, Fisk University, and Tennessee State University.

Panelists shared the need for more geneticists and genetic counselors, and particularly the current shortage of individuals from diverse backgrounds who are critical to ensuring that advances in genomic research and genomic medicine benefit everyone equally. Among the questions raised for discussion by the early career scientists included:

- How do you decide whether to pursue an MD, PhD, or MD-PhD?
- How do you prepare to be a successful applicant for these degrees?
- What can you do with these degrees and what do the career paths look like?
- What types of summer research rotations are available at your institutions?

The students learned that PhDs can participate in the care of patients as well as research and education, and that MDs in genetics may do research in addition to direct patient care and educating the next generation. After the panel discussion, the students had an opportunity to talk one-on-one with the visitors. One student commented "I never knew that you could have a career in Medical Genetics. How can I find out more?" Dr. Kathy Friedman sent the following message: "Thank YOU all very much for reaching out to us and suggesting this event and for helping to organize it. It was absolutely fantastic and the students were engaged and energized as well! A number of them stayed afterwards to talk with us and among themselves – they all expressed how much they learned and how helpful the session was."

During the ACMG Annual Clinical Genetics Meeting, five leaders in the field of Medical Genetics shared career advice and their enthusiasm for medical genetics with more than 30 undergraduate students in-person and with many more online at Vanderbilt University in Nashville, TN.

2022 ACMG Diversity, Equity, and Inclusion Breakfast: Tools to Confront Imposter Fears/ Thoughts

by **Laura Duque Lasio, MD, FACMG, Clinical Biochemical Genetics fellow at University of Utah,** and **Fabiola Quintero-Rivera, MD, FACMG Professor, Departments of Pathology, Laboratory Medicine, and Pediatrics, School of Medicine, University of California Irvine**

The ACMG DEI Committee's overarching goal is to celebrate and promote DEI in our membership. We know this will not only improve our sense of community and belonging but ultimately result in better science and improved care for the communities we serve. Among the Committee's programs is the annual Diversity, Equity, and Inclusion (DEI) Breakfast. Held at the 2022 ACMG Annual Clinical Genetics Meeting in Nashville, TN, this 1.5-hour session had 55 in-person attendees, among them were members of the ACMG DEI Committee and many members of the College's leadership, including ACMG President, Marc S. Williams MD, FACMG and ACMG CEO, Max Muenke, MD, MBA, FACMG. As organizers and moderators (Drs. Duque Lasio and Quintero-Rivera), had big shoes to fill, taking over the planning of this session from Dr. Fuki Hisama Professor of Medicine at University of Washington School of Medicine, who successfully organized it since its inception in 2017. Prior topics discussed in these sessions over the years have included implicit bias, microaggressions and "Inspiring people who look like us" featuring stories of accomplished Asian, Black, Hispanic, and female physicians and scientists.

Planning this session amid uncertainty was a challenge, but it paid off and the event was a success. Our speaker, Dr. Ericka Boone, Acting Director, Division of Biomedical Research Workforce, National Institutes of Health, joined us via zoom to discuss the important topic of imposter fears and how to confront them. Our initial concerns about having a virtual presenter were quelled as soon as Dr. Boone started her very engaging talk. Her remarks garnered everyone's attention and, on some occasions, resulted in bursts of laughter. Imposter fears/thoughts are described as "a pattern of thought where an individual doubts their own accomplishments and has a persistent internalized fear of being exposed as completely unprepared and unqualified." It is estimated that 70% of the population has battled the 'imposter monster' including highly successful individuals. While most attendees shared their prior experiences with imposter fears, some were learning about it for the first time. This session provided an excellent opportunity for our attendees to be vulnerable and know they are not alone.

While much of the work to confront imposter fears/ thoughts (a.k.a. imposter syndrome) is individual, institutions have a significant role in creating safe, collaborative and equitable work environments. As we work towards making positive changes, let's breathe, talk to each other, visualize our success, practice self-care and consider mentoring someone else. Not only will you empower your mentee, but you will also empower yourself at the same time!

Maria Laura Duque Lasio, MD, FACMG and Fabiola Quintero-Rivera, MD, FACMG at the DEI Breakfast during the ACMG Annual Clinical Genetics Meeting in Nashville, TN.

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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.

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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.

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NBSTRN Educational Resources for Medical Geneticists

Medical geneticists play key roles in diagnosing, caring for, and managing individuals identified with a condition through newborn screening (NBS). In addition, the increasing use of genomics in NBS translates to more involvement of medical geneticists in NBS. NBSTRN operates with the mission to facilitate discoveries in NBS research. With this mission in mind, NBSTRN provides educational tools and resources for the medical genetics and genomics community. These resources include:

Accredited Session at the ACMG Genetics Academy
Check out our accredited session from the 2022 ACMG Annual Clinical Genetics Meeting titled, "Exploring the Role of Medical Genetics and Genomics in Advancing Newborn Screening Research." This session features unique perspectives from three medical geneticists and highlights their role in NBS research. This session offers CME, P.A.C.E.®, or NSGC credits (www.acmgeducation.net/Public/Catalog/Main.aspx).

Educational NBS Research Events
The NBSTRN hosts educational events for medical geneticists and NBS researchers every year. These events feature presentations from innovators who are expanding the reach of newborn screening research. Save the date for the 2022 Network Meeting: July 19th-20th, 2022 (www.nbstrn.org/2022-Network-Meeting).

National NBS Pilot Monthly Webinar
The NBSTRN hosts a monthly webinar via zoom to facilitate information sharing between state newborn screening programs, researchers, clinicians, advocates and federal partners about conditions recently added to the Recommended Uniform Screening Panel (RUSP) or conditions currently part of pilots (www.nbstrn.org/resources/national-nbs-pilot-monthly-webinar).

NBS SPOTlight Podcast
This podcast is about advancements in rare disease research from the perspectives of health professionals, researchers, parents and advocates. With this podcast, the NBSTRN strives to educate listeners on the life-saving benefits of newborn screening research, new technologies and rare disease treatments (www.nbstrn.org/podcast).

Newsletter and Blogs
The NBSTRN newsletter and blog share the latest discoveries in newborn screening. View upcoming NBS research events, new NBSTRN resources and recent NBS research news in NBSTRN newsletters and blogs (www.nbstrn.org/latest-news).

Register as a member of the NBSTRN to receive the newsletter (www.nbstrn.org/registration)!

FIVE NBSTRN RESOURCES FOR MEDICAL GENETICISTS

Accredited Session at the ACMG Genetics Academy

Earn CME, P.A.C.E.®, or NSGC credits by completing our course titled, "Exploring the Role of Medical Genetics and Genomics in Advancing Newborn Screening Research."

NBS Research Events

Save the date for the 2022 Network Meeting: July 19th-20th, 2022.

National NBS Pilot Monthly Webinar

This webinar facilitates information sharing between state newborn screening programs, researchers, clinicians, advocates, and federal partners.

Newsletter and Blogs

View upcoming NBS research events, new NBSTRN data tools, and recent NBS research news in our newsletters and blogs. Register as a member of the NBSTRN to receive our newsletter!

NBS SPOTlight Podcast

Enjoy our podcast and learn about advancements in rare disease research through the voices of health professionals, researchers, parents, and advocates.

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ACMG ACT Sheets and Algorithms

In collaboration with ACMG, NCC develops and maintains the 100+ ACT Sheets and Algorithms to provide clinical decision support to non-genetic providers. Check them out, including the newly updated Metabolic Conditions ACT Sheets, at https://acmg.net/act.

Knowledge Nugget Series

As a companion educational offering to the ACMG ACT Sheets, NCC develops the Knowledge Nugget Series. These 5-7 minute videos walk you through a specific ACT Sheet. Earn CME credit by watching the series in the ACMG Genetics Academy.

Educational Efforts

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Medical Necessity Webinar Series

In collaboration with The Catalyst Center, the NCC held a three-part webinar series this past spring that helps providers navigate the complexities of medical necessity. View the enduring webinars and earn CME credit by visiting the ACMG Genetics Academy.

Evidence-Based Guideline Webinar Series

In partnership with ACMG, NCC is excited to announce a new two-part webinar series in which an ACMG Evidence-Based Guideline will be reviewed and discussed. Part 1, which is sponsored by NCC, will provide a general overview of the guideline. Register for upcoming webinars by visiting the ACMG Genetics Academy.

Find all of these educational resources, plus many, many more, in our Resource Repository or subscribe to our social media channels for the latest updates.

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Latest ACMG Foundation Summer Genetics Scholars Cohort is Largest Ever

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

The ACMG Foundation for Genetic and Genomic Medicine (ACMGF) is pleased to announce that the 2022 Summer Genetics Scholars Program (SGSP) will include its largest cohort to date. Thanks to grants from BioMarin and Ultragenyx, the Foundation will sponsor 18 students this summer, with seven additional students being supported by their host institution, for a total cohort of 25 students! Of the 18 institutions participating this year, six are new to the program.

> **"After my six weeks with the medical genetics department at UNC, I can see myself going into this field. I really love the science behind genetics and knowing that there will always be new discoveries and more to learn in the field."**

SGSP 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 while providing a modest stipend.

Started in 2011, SGSP has sponsored over 150 scholars at more than 50 different institutions around the country. This program influences the career paths of participants. As one scholar wrote, "After my six weeks with the medical genetics department at UNC, I can see myself going into this field. I really love the science behind genetics and knowing that there will always be new discoveries and more to learn in the field."

The ACMG Foundation thanks BioMarin, Ultragenyx and all past corporate partners who supported students and our ongoing efforts to expand the genetics workforce pipeline.

Below is a list of the 2022 participating institutions, mentors and scholars.

Institution	Principal Mentor/ Faculty Member	Summer Scholars
Baylor College of Medicine	Reza Bekheirnia, MD, FACMG	Ava Berrier
Case Western Reserve University/University Hospitals of Cleveland	Anna Mitchell, MD, PhD, FACMG	Tripti Rathi
Greenwood Genetic Center	Elliot Stolerman, MD, FACMG	Evdokia Angelidis
Hospital of the University of Pennsylvania	Staci Kallish, DO, FACMG	Yehuda Elkaim
Kaiser Permanente Bernard J. Tyson School of Medicine	Amanda Freed, MD	Kelly Wang
Loma Linda Medical School	Hua Wang, MD, PhD, FACMG	Curtis Grossheim
Mercy Health System/University of Illinois College of Medicine	Lea Parsley, MD, FACMG	Tiayrra Kirkwood
Norton Children's Medical Group	Joseph Hersh, MD, FACMG	Marina Sasnau
Penn State Health Children's Hospital	Patricia Gordon, MD, FACMG	Elizaveta Makarova
University of Alabama at Birmingham*	Nathaniel Robin, MD, FACMG	Morgan Jenkins Jacob Greenway Tanya Hsiung
University of Iowa*	Hatem El-Shanti, MD	Allyson Rose Noel Tolvanen
University of Maryland School of Medicine*	Carol Greene, MD, FACMG	Armella Diane Poggi Sachin Kuruvilla
University of Miami, Miller School of Medicine	Deborah Barbouth, MD, FACMG	Sofia Zoullas
University of Pittsburgh School of Medicine/UPMC Children's Hospital of Pittsburgh*	Jerry Vockley, MD, PhD, FACMG	Anisha Verma Peter Nelson
University of South Florida	Judith Ranells, MD, FACMG	Elizabeth Fletcher
University of Tennessee Health Science Center/St. Jude Children's Research Hospital	Henry Mroczkowski, MD, PhD, FACMG	Yanxin (Emily) Chen
University of Texas Health Science Center	Hope Northrup, MD, FACMG	Shelby Mills
University of Utah*	David Viskochil, MD, PhD, FACMG	Shelby Geilmann Priya Swaminathan

*Host institution provided supported for a 2nd Summer Scholar

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

To facilitate the delivery of quality clinical and laboratory medical genetics and genomics services, the ACMG—through its Board of Directors, committees and workgroups—publishes policy statements, evidence based or expert clinical and laboratory practice guidelines, and descriptions of best practices in genomic medicine. Additionally, the ACMG and the National Coordinating Center for the Regional Genetics Networks (NCC) publish ACTion (ACT) Sheets, which serve as clinical decision tools for healthcare providers without genetics expertise. ACT Sheets are categorized by subject areas including newborn screening, carrier, diagnostic test, family history, transition and secondary findings. The following is a list of the College's recent publications since the Winter edition of *The ACMG Medical Geneticist*:

ACMG Statements, Guidelines, Technical Standards and Practice Resources:

David T. Miller, Kristy Lee, Noura S. Abul-Husn, Laura M. Amendola, Kyle Brothers, Wendy K. Chung, Michael H. Gollob, Adam S. Gordon, Steven M. Harrison, Ray E. Hershberger, Teri E. Klein, C. Sue Richards, Douglas R. Stewart, Christa Lese Martin and the ACMG Secondary Findings Working Group. **ACMG SF v3.1 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 17 June 2022; doi: https://doi.org/10.1016/j.gim.2022.04.006.

Marilyn M. Li, Ahmad Abou Tayoun, Marina DiStefano, Arti Pandya, Heidi L. Rehm, Nathaniel H. Robin, Amanda M. Schaefer, Christine Yoshinaga-Itano and the ACMG Professional Practice and Guidelines Committee. **Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 10 May 2022; doi: https://doi.org/10.1016/j.gim.2022.03.018.

Nancy C. Rose, Elizabeth S. Barrie, Jennifer Malinowski, Gabrielle P. Jenkins, Monica R. McClain, Danielle LaGrave, Marco L. Leung and the ACMG Professional Practice and Guidelines Committee. **Systematic evidence-based review: The application of noninvasive prenatal screening using cell-free DNA in general-risk pregnancies.** *Genet Med* published online May 24, 2022; doi: https://doi.org/10.1016/j.gim.2022.03.019.

Marwan K. Tayeh, Andrea Gaedigk, Matthew P. Goetz, Teri E. Klein, Elaine Lyon, Gwendolyn A. McMillin, Stefan Rentas, Marwan Shinawi, Victoria M. Pratt, Stuart A. Scott and the ACMG Laboratory Quality Assurance Committee. **Clinical pharmacogenomic testing and reporting: A technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 10 February 2022; doi: https://doi.org/10.1016/j.gim.2021.12.009.

Erin T. Strovel, Kristina Cusmano-Ozog, Tim Wood, Chunli Yu and the ACMG Laboratory Quality Assurance Committee. **Measurement of lysosomal enzyme activities: A technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 10 February 2022; doi: https://doi.org/10.1016/j.gim.2021.12.013.

Laurie H. Seaver, George Khushf, Nancy M.P. King, Dena R. Matalon, Kunal Sanghavi, Matteo Vatta, Kristi Wees and the ACMG Social, Ethical and Legal Issues Committee. **Points to consider to avoid unfair discrimination and the misuse of genetic information: A statement of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 16 December 2021; doi: https://doi.org/10.1016/j.gim.2021.11.002.

Robert G. Best, George Khushf, Sara Schonfeld Rabin-Havt, Ellen Wright Clayton, Theresa A. Grebe, Jill Hagenkord, Scott Topper, Jaime Fivecoat, Margaret Chen, Wayne W. Grody and the ACMG Social, Ethical and Legal Issues Committee. **Stewardship of patient genomic data: A policy statement of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 16 December 2021; doi: https://doi.org/10.1016/j.gim.2021.11.001.

Patrick R. Gonzales, Erica F. Andersen, Teneille R. Brown, Vanessa L. Horner, Juli Horwitz, Catherine W. Rehder, Natasha L. Rudy, Nathaniel H. Robin, Erik C. Thorland and the ACMG Laboratory Quality Assurance Committee. **Interpretation and reporting of large regions of homozygosity and suspected consanguinity/uniparental disomy, 2021 revision: A technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* published online 03 December 2021; doi: https://doi.org/10.1016/j.gim.2021.10.004.

Focused Revisions and Addenda:

Caroline Astbury, Judith Benkendorf and the ACMG Laboratory Quality Assurance Committee. **Addendum: Technical standards and guidelines: Molecular genetic testing for ultra-rare disorders.** *Genet Med* published online 30 November 2021; doi: https://doi.org/10.1016/j.gim.2021.10.006.

ACMG ACT Sheets and Algorithms:

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Argininemia Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Arginine.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Argininemia Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/Arginine-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Biotinidase Deficiency Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Biotinidase-Deficiency-ACT-Sheet.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Biotinidase Deficiency Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/Biotinidase-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Carnitine Palmitoyltransferase I (CPT I) Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/C0_C16+C18.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Carnitine Palmitoyltransferase I (CPT I) Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/C0-C16-C18-Elevated-Algorithm.pdf.

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American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Carnitine Uptake Defect Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Carnitine-Uptake-Defect-ACT-Sheet.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Carnitine Uptake Defect Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/C0-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Citrullinemia Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Citrullinemia.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Citrullinemia Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/Citrulline-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Classic Galactosemia Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Classical-Galactosemia-ACT-Sheet.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Classic Galactosemia Newborn Screening Algorithm.** Available at https://www.acmg.net//PDFLibrary/GALT-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **DMD Elevated Creatine Kinase Muscle Isoform Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/DMD-Elevated-CKMM-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Fabry Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Fabry.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). **Fabry Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/Fabry-Algorithm.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the

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C. Thomas Caskey, MD, FACMG

by **Robert L. Nussbaum, MD, FACMG, Invitae** and **James R. Lupski, MD, PhD, D.Sc (hon), Baylor College of Medicine**

It is with great sadness that we note the death of our mentor C. Thomas Caskey, MD, FACMG, one of the most influential and outstanding human and medical geneticists of his generation. He died 13 Jan 2022, age 83, after a brief illness (1). His engaging acceptance speech for the 2021 Allan Award from the American Society of Human Genetics described his education at the University of South Carolina, Duke, and NIH, and his move to Houston, TX. There, with his colleague, Arthur L. Beaudet MD, he founded research and clinical programs in genetics at Baylor College of Medicine (BCM) (2).

Tom was a highly successful human genetics investigator. He was quick to recognize the impact that new technologies would have on medical genetics research and practice. However, his career achievements went far beyond the research he directed and the advances in human genetics they represent. He revolutionized medical genetics, molecular diagnostics, and the law through forensic DNA technologies. Furthermore, as part of the Human Genome Project, he helped industrialize genomics and made important contributions in human genome analyses.

But this brief overview of Tom's career fails to capture what was really special about him. Tom was a bold visionary. He understood "the big picture" and saw the direction in which human genetics needed to go, both in research and the practice of medicine. His vision for genetics research, teaching and practice, essentially melding together the 'trifecta of medicine' in a 'bridging department', that would become the world-renowned BCM Department of Human and Molecular Genetics. Students embedded in this community of geneticists, could thrive in a fertile environment for learning, teaching, exploring and applying human and medical genetics. We all, including patients and families worldwide, owe a debt of gratitude to Tom.

Finally, a word about Tom as a human being. He was serious, but never pompous, bold in his thinking, enthusiastic about his science, and generous with his knowledge and in his dealings with colleagues and students. His keen sense of humor was legendary and was often manifested by an irrepressible twinkle in his eyes accompanied by loud guffaws and exclamations delivered in his distinctive southern accent. He was a successful husband and partner with his wife Peggy, a proud father to his children Caroline (Goodner) and Clifton, and an enthusiastic sailor. We join with the entire world-wide genetics community in mourning the loss of a remarkable physician-scientist, visionary leader, mentor, and colleague.

1. https://www.bcm.edu/news/genetics-pioneer-dr-c-thomas-caskey-dies
2. https://www.youtube.com/watch?v=3xDKuzMa4OU

Haig H. Kazazian Jr, MD, FACMG

by **Ada Hamosh, MD, MPH, FACMG; Gregg Semenza, MD PhD, FACMG** and **David Valle, MD, FACMG, Johns Hopkins University School of Medicine**

Haig H. Kazazian Jr, MD, FACMG was a leader in the field of human molecular genetics for five decades. At a time when techniques were primitive by modern standards, he first demonstrated that disease-causing mutations in patients with beta-thalassemia occur on haplotypes defined by linked DNA polymorphisms at the beta-globin locus. Haig then focused on the gene encoding factor VIII (F8) in boys with hemophilia A and found that a different spectrum of molecular mechanisms was involved in mutagenesis of this large X-linked gene compared to the much smaller autosomal beta-globin gene. Taken together, his thalassemia and hemophilia studies developed a paradigm that was subsequently utilized by many other investigators to perform detailed molecular characterization of scores of genetic disorders as well as somatic mutations in human cancer and was a scientific progenitor of current day GWAS (genome-wide association studies). He recognized the clinical significance of this paradigm and opened the first DNA diagnostic laboratory in the US. He also led human genetics centers/departments at both Johns Hopkins and the University of Pennsylvania. During the hemophilia study, Haig identified an allele produced by de novo insertion of a partial L1 long interspersed nucleotide element (LINE) into the F8 gene. Fascinated by this observation, he went on to identify and isolate a full-length L1 element, to demonstrate that it functioned as a retrotransposon in a mouse model and to show that retrotransposition occurs early in human embryonic development generating somatic mosaicism. The importance of Haig's discovery of mobile L1 elements in the human genome and their role in disease pathogenesis was underscored in 2001 (13 years after he began his pioneering studies) when determination of our genome sequence revealed that L1 elements comprise a staggering 16% of the human genome. Haig's contributions were recognized by numerous awards including election to the National Academy of Science and the ASHG's William Allan award for "far-reaching scientific contributions to human genetics." Beyond these scientific accomplishments, Haig was a good-humored role model and enthusiastic mentor par excellence, who generously and continuously supported the career development of his many trainees, present and past, and enriched the lives of all of us.

Continued from page 41

Regional Genetics Networks (NCC). **Gaucher Disease Newborn Screening ACT Sheet.** Available at https://www.acmg.net/PDFLibrary/Gaucher.pdf.

American College of Medical Genetics and Genomics (ACMG) and the National Coordinating Center for the Regional Genetics Networks (NCC). G**aucher Disease Newborn Screening Algorithm.** Available at https://www.acmg.net/PDFLibrary/Gaucher-Algorithm.pdf.

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Contact Information

American College of Medical Genetics and Genomics
7101 Wisconsin Avenue, Suite 1101
Bethesda, MD 20814
Tel: 301-718-9603
Fax: 301-718-9604
acmg@acmg.net
www.acmg.net

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