ACMG In Action

Essential Updates: Member News You Can Use

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

November 2021

In This Issue...

Upcoming Events

2022 ACMG Annual Clinical Genetics Meeting
March 22-26, 2022

RENEW MEMBERSHIP

Website Links

ACMG
ACMG Foundation

Advocacy Update

Webinar on Regulatory Landscape of Laboratory Developed Testing Procedures

ACMG, together with the American Association for Clinical Chemistry (AACC), the Association for Molecular Pathology (AMP) and the Association of Pathology Chairs (APC), recently hosted a webinar on "The Regulatory Landscape of Laboratory Developed Testing Procedures: How We Got Here, Why It's Important, Where We're Going and How You Can Get Involved." The highly attended webinar covered the history of regulation of laboratory developed tests and the potential impact that current federal legislative proposals could have on clinical care. Recordings and slides from each of the four presentations, as well as other related resources, are publicly available here. We encourage you to share the presentations and keep an eye out for additional resources to help you get involved. For questions, please contact ACMG's public policy team at advocacy@acmg.net.

AMA Telehealth Survey: Share Your Knowledge

The American Medical Association (AMA) is conducting a survey to assess the current landscape and use of telehealth among physicians and other healthcare practitioners. Their goal is to learn more about physicians' experience with telehealth, including ongoing challenges, benefits and opportunities. Survey results will help inform future telehealth research and advocacy, resource development, and continued support for physicians, practices and health systems.

Since the COVID-19 pandemic, the US has witnessed a surge in the use of telemedicine. However, ACMG's members have been leaders in telemedicine since well before the pandemic, and this is an opportunity to share that knowledge with the broader medical community. Please consider participating in the AMA's telehealth survey, available here, which will close on **December 31, 2021 at 11:59 pm ET**.

Remember the ACMG Foundation on Giving Tuesday

[ACMG Meetings Website]
[ACMG Education Center]
[NBSTRN]
[NCC]
[ClinGen]
[GIM]

November 30th marks Giving Tuesday, the annual "day of generosity" created in 2012 as a way for people to give back to nonprofit organizations after the consumeristic post-Thanksgiving days of Black Friday and, more recently, Cyber Monday. During this year's Giving Tuesday, the ACMG Foundation for Genetic and Genomic Medicine (ACMGF) asks for your support as we continue to grow and bolster the mission of ACMG.

Your individual contributions will help support our programs and ensure that federal and state governments, healthcare professionals and key partners are educated on the scientific and medical issues related to the increasingly complex field of medical genetics. Gifts from many individual donors also helps us to secure even larger external grants because individual contributions show funders and corporate partners that we have strong support from our incredible membership.

Over the past 15 years, the ACMGF has raised nearly $15 million to lead educational efforts, support young health care providers and scientists, as well as further sustain the science, staff and programs of the College.

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ACMG In the News

Healthy Women Article Highlights NBSTRN

Healthy Women, a consumer media outlet that reaches more than 6 million readers each month, interviewed Amy Brower, PhD, associate project director of NBSTRN (an NICHD-funded project at ACMG) and ACMG's CEO, Max Muenke, MD, FACMG, for an article entitled "Newborn Screening Saves Lives: What Parents Need to Know." The article, which was published online November 9, 2021, conveys the importance of newborn screening, the testing discrepancies between states and how parents can become better educated and serve as advocates for newborn screening. A link to some of NBSTRN's online data tools is provided in the article. Dr. Brower tells *Healthy Women*, "The hallmark of newborn screening is that we can detect something in newborns that's unobservable, but by looking at either physiological or blood-based tests, we can determine that a disease is there and intervene."

ASCO Post Cites ACMG PALB2 Clinical Practice Resource

The ASCO Post recently published an article entitled "Enhanced Surveillance and Risk-Reducing Intervention Options for Individuals With PALB2 Variants" that includes interviews with some of the authors of "Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG)" which appeared in the August 2021 issue of *Genetics in Medicine*, ACMG's official journal.

"Our primary goal in writing the paper was to develop advice on the clinical management of people who have a pathogenic or likely pathogenic *PALB2* variant," Douglas R. Stewart, MD, FACMG, one of the authors, told *The ASCO Post*, which is sent to ASCO's 45,000 members. Two other authors of ACMG's *PALB2* clinical practice resource, Tuya Pal, MD, FACMG and Marc Tischkowitz, MD, PhD, also appear in *The ASCO Post* article.

November GenePod: The Potential Impact of a PRS-based Breast Cancer Risk Assessment

Polygenic risk scores (PRS) can be an important tool in breast cancer patients to help stratify individuals into levels of disease risk. The clinical utility of PRS is still being evaluated, but what hasn't yet been evaluated is how to communicate such results to patients, and how they respond to their PRS scores.

On this month's GenePod, Tatiane Yanes, a post-doctoral researcher at the University of Queensland and a genetic counselor at the Queensland Children's Hospital, discusses how a team of researchers surveyed an existing pool of patients that had undergone genetic testing for breast cancer. "We're really just trying to get an understanding of how someone might respond to receiving this information, and what sort of decisions they might make around their breast cancer risk management," said Yanes.

ACMG Member Comment: Application of Noninvasive Prenatal Screening

The ACMG Board of Directors requests your comments on a new draft systematic evidence-based review, developed under the auspices of the College's Evidence-Based Guidelines Program. This members' only opportunity consistently leads to higher quality publications, which benefit from the incorporation of the collective expertise of the ACMG membership in the peer review process. Please join your colleagues in reviewing the following manuscript:

Systematic Evidence-based Review: The Application of Noninvasive Prenatal Screening Using Cell-free DNA in Average Risk Pregnancies.

When submitting comments for review, please provide your full name, degrees, affiliation and contact information. **All comments should reference the precise line numbers to which they pertain**. Comments should be forwarded, via email, to Documents@acmg.net by **Friday December 17, 2021 at 7:00 am ET**. The subject line of your emailed comments should read: "**Application of Noninvasive Prenatal Screening**".

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ACMG Welcomes 24 New Members

ACMG welcomes and congratulates 24 new and newly certified members. There has never been a more exciting time to be a part of this field and we encourage current members of the College to invite friends

and colleagues to join and help grow the medical genetics and genomics field. Thank you to all our members who make important contributions and the work of the College possible through your membership, support and involvement.

Fellow Member
Rebecca D. Ganetzky, MD, FACMG

Candidate Fellow
Kiely James, PhD

Associate Member
Tanner Coleman, MS, CGC

Affiliate Members
Elizabeth B. Barnby, MSN, DNP
Mohammad A. Faghihi, MD
Cristiane M. Ida, MD
Brock Schroeder, PhD

Trainee Members
Zenobia Gonsalves, MD
Monique Morrison, PhD

Student Members
Michael Angerbauer, BS
Sasha Atira Bauer, BA
Francesca Caravano, BS
Eucaris C. Feliciano Morales, BS
Christina A. Hansen, BA
Arusa Khan, BA
Yasas D. Kolambage, MBBS
Alexis Martin, BS
Sarnaver Muroki, BS
David Niemynski, BS
Ayorinde Olowoyeye, BS
Mark David Orland, BS
Sheridan Schwartz, BS
Kimberly A. Toth, BS
Sofia Zoullas, BA

Know someone who should join ACMG? Please tell them to visit the Join ACMG webpage.

ACMG Young Professional Fellow Membership

If you passed your initial medical genetics and genomics certification exam(s) this August, congratulations! You are now eligible for the discounted ACMG Young Professional Fellow Year 1 membership ($160). Please visit our Join ACMG webpage and select the Status Change option to submit your application and payment.

If you are completing an additional training/fellowship program, and want to remain a Trainee member, please submit a completed Student/Trainee Verification Form via email to the ACMG Membership Department or fax to 301-718-9604. (Forms must be signed by the program director or by an authorized representative).

2021 ACMG Salary Survey Now Closed; Report Available

Spring 2022

Thank you to everyone who participated in the 2021 Medical Geneticist Salary Survey. The survey is now closed, and the resulting report will be made available for free to ACMG members in spring 2022. Such surveys are used by individual ACMG members as well as department heads, chairs, and deans to influence salaries and remain competitive, and we are pleased to announce a 9% increase in participation compared to 2019. We also want to congratulate Elizabeth Chao, MD, FACMG, who is this year's drawing winner and will be receiving a free iPad Mini! Thank you to Dr. Chao and so many others for supporting the ACMG Salary Survey.

ClinGen Seeks Workshop Proposal Submissions for June Conference

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. ClinGen and DECIPHER are co-hosting the Curating the Clinical Genome (CCG) 2022 conference virtually June 6-8, 2022. Workshop proposal submission for CCG 2022 is now open here.

Workshops may consist of educational events or discussion panels that relate to scientific scholarship, research tools, new technologies, skill development or public information related to science. Workshops will be streamed live and are expected to include interactive components.

Submissions are due January 31, 2022. Acceptance notifications will be sent by February 25, 2022. Accepted proposals will receive up to two complimentary registrations to CCG 2022. If you have questions about the workshop application, please email clingen@clinicalgenome.org.

NBSTRN Update: Federal Advisory Committee Considers Screening for MPS II and *GAMT* Deficiency

More than 12,500 newborns are diagnosed with a genetic disease through newborn screening (NBS) annually in the United States. A federal advisory committee, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), advises the Secretary of Health and Human Services (HHS) about aspects of newborn and childhood screening, including recommending which conditions to screen. The ACHDNC meets four times a year and meetings are open to all NBS stakeholders. The final meeting of 2021 was held on November 9th and 10th and included a discussion of the evidence supporting screening for two conditions, Mucopolysaccharidosis Type II (MPS II) and Guanidinoacetate Methyltransferase (GAMT) Deficiency. Although some states screen for these conditions, they are currently not a part of the Recommended Uniform Screening Panel (RUSP).

NBSTRN, an NICHD-funded project at ACMG, creates tools and resources to support research and pilots that help develop evidence that screening is feasible and beneficial. MPS II and *GAMT* Deficiency are featured in the Newborn Screening Conditions Resource (NBS-CR), a

tool that highlights essential information on disorders that are part of, or candidates for, the RUSP. NBS-CR provides a centralized resource of facts and statistics for both screened and candidate conditions and is designed to be an interactive resource for researchers, clinicians and families to learn more about these conditions. The NBS-CR provides condition-specific clinical characteristics, diagnosis information, clinical trial information and suggested readings through links to MedGen, a portal developed and maintained by the National Library of Medicine (NLM) National Center for Biotechnology Information (NCBI). In addition, resources for families and advocacy groups are included with links to Genetics Home Reference (GHR) and Genetic Alliance's Disease InfoSearch.

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Now Available in the ACMG Genetics Academy

December's ClinGen Somatic Cancer and VICC Virtual Molecular Tumor Board Case Series Webinar

Tuesday, December 14, 11:00 am – 12:00 pm ET
This monthly webinar series is for all medical and healthcare professionals and researchers interested in understanding cancer genomic testing and somatic and germline variant interpretation methods. It is presented as a collaboration between ClinGen Somatic and VICC consortia, and ACMG.

Registration coming soon! Check the ACMG Genetics Academy website.

NEW! Improving Patient Safety: An Imperative in Medical Genetics and Genomics Healthcare

This course for clinical geneticists and clinical laboratory specialists is comprised of an in-depth dialogue of key critical topics identified as impacting patient healthcare safety. In this activity, a participant discovers the burden of medical errors on the patient and healthcare system, contributing factors for medical errors, myths regarding patient safety, pathways to reducing errors, communication techniques to improve patient safety, the importance of teamwork in the safety of the patient, identifying adverse events and how to disclose adverse events. Cases cover instructive yet challenging situations that allow the participant to assess their comprehension and application of the material presented in the module. This course is offered free of charge to members and non-members.

More information.

New Part IV CCP Modules

- **PPM - Analysis of Urine Organic Acids:** For clinical biochemical geneticists who analyze, interpret, and report urine organic acids. Purchase Now
- **PPM - Cytogenomic Analysis of Newly Diagnosed Plasma Cell Disorders (Multiple Myeloma, MGUS, Plasmacytoma,**

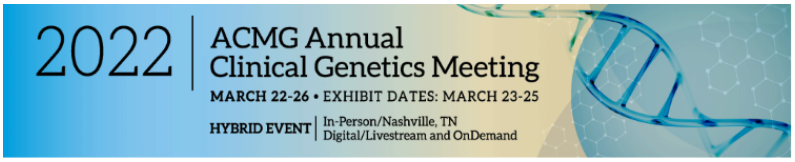
Smoldering Myeloma): This module is an overview of recommendations for cytogenomic laboratory work-up of patients with newly diagnosed plasma cell disorders.
Purchase Now

Claim Your Credits for the 2021 ACMG Annual Clinical Genetics Meeting

Participants in the 2021 ACMG Annual Clinical Genetics Meeting have until December 1st to claim their CME and P.A.C.E.® Credits. Go to the 2021 ACMG Meeting platform and log in using your registration confirmation number.

Questions? Email the ACMG Education team.

2022 ACMG Annual Clinical Genetics Meeting Update



Abstract Submission Deadline: Friday, November 19, 2021, at 11:59 pm PT

Gain exposure for your work and advance your career. The presentation of the most advanced research is an essential component of the ACMG Annual Clinical Genetics Meeting. All genetics professionals (ACMG members and non-members) are encouraged to submit abstracts for presentation. Submitted abstracts may be selected for poster displays or oral platform presentations. These presentations provide another opportunity to collaborate and network with your peers.

Abstract submission guidelines and a link to the online submission site is located on the ACMG Meeting website.

2022 ACMG Annual Meeting – Register Early to Save!

The Early Bird discount registration deadline is Wednesday, December 22, 2021 – register by the deadline and save up to $200 on registration fees! View fees and register.

ACMG has detailed plans in place to protect your health and safety at the meeting, which will be regularly assessed and updated as needed in response to pandemic-related changes. All in-person participants must provide proof of full vaccination against COVID-19 to gain entry to the Meeting. More information will be provided to registered attendees and exhibit booth personnel in February.

Hotel Reservations

You must be registered to make hotel reservations. A link to the hotel reservation site will be provided at the conclusion of the registration process. The ACMG hotel reservation deadline is March 3, 2022. Reserve early to secure one of your top hotel choices.
View Nashville Hotels

Please Note: CMR is ACMG's Official Housing Partner – they are the only company authorized to provide hotel reservation services for the

ACMG meeting. ACMG has not authorized any other company to contact meeting attendees or exhibitors. If contacted by any company other than CMR, even if they appear to be affiliated with the 2022 ACMG Annual Clinical Genetics Meeting and the ACMG hotel block, they are not. Neither ACMG nor CMR can assist in resolving any disputes. **Please do not answer these solicitations.**

Call for Cases – Diagnostic Challenges and Pediatric Diagnostic Dilemmas (Rare Knowns and Unknowns)

Do you have a puzzling case? Submit it as a Diagnostic Challenge! Diagnostic Dilemmas / Challenges are interactive sessions which allow genetics professionals to present cases of rare knowns and unknowns as well as share best practices. **Deadline for submission is January 14, 2022**. We are particularly interested in cases that fall into the following categories:

• Adult & Cancer Diagnostic Dilemmas
• Prenatal Diagnostic Challenges
• Laboratory Diagnostic Challenges (Constitutional and Neoplastic Cases in Molecular, Cytogenomic, and Biochemical Genetics Specialties)
• Pediatric Diagnostic Dilemmas (Rare Knowns and Unknowns)

Learn more and submit a case.

Program Highlights

Below are just a few of the sessions taking place in Nashville. View the full program.

Plenary Sessions:

- 2022 Presidential Plenary Session and ACMG Foundation Awards Presentation - From Exceptional to Routine: Transformation of Genomic Medicine in the 21st Century
- Four Featured Platform Presentations
- TED - Style Talks
  o Incentivizing Knowledge Sharing: The Story of ClinVar - Heidi Rehm
  o Honoring Mendel and Exploring Some Educational Boxes We've Built from His Work - Joseph McInerney
  o Whither Prenatal Diagnosis? - Ellen Clayton
- What are the Important Issues in the Arena of Public Policy and Legislation for Medical Genetics?
- Developing Recommendations for the Application of Direct-to Consumer Genetic Testing in Clinical Care
- R. Rodney Howell Symposium - Population Genomic Health: Expanding the Reach of Genomic Medicine to Diverse Populations

Workshops and Interactive Sessions:

- Student Workshop: Career Pathways in Genetics: What Do I Do and How Did I Get Here?
- Early Career Genetic Professionals Workshop – Pursuing a Career in Medical Genetics
- The ACMG Evidence-Based Guideline Program: Our Roadmap from Evidence to Recommendations and the Impact on Patient Care

- An Action Plan for Inclusion, Diversity, Equity and Anti-Racism in Publishing: Putting IDEAs into Practice at Genetics in Medicine
- How Can We Maximize the Yield of Genetic Disease Diagnostic Evaluation and Testing? An Interactive Workshop

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