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ACMG In Action

Essential Updates: Member News You Can Use

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

September 2021

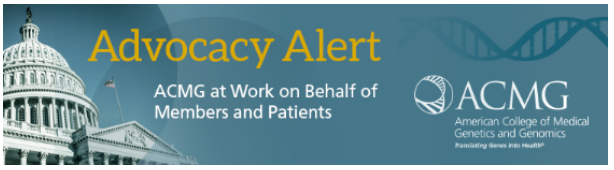
In This Issue...

Upcoming Events

Sneak Peek: Fall Edition of Member Magazine Now Online

The fall issue of *The ACMG Medical Geneticist* will be coming to your mailbox in late September, but you can read it online now in the members-only section of the ACMG website. The issue shines a spotlight on the College's advocacy efforts on behalf of medical genetics. Our collaboration with other organizations to impact federal policy is highlighted in a Q&A with ACMG President-Elect Susan Klugman, MD, FACMG, about how the ACMG collaborates with the AMA to advocate for medical genetics and how our participation in the AMA House of Delegates advances causes that are important to the field. How you can become an effective advocate at the local level or within your institution is also covered. The issue also includes two articles on diversity – one on efforts to increase diversity in the clinical genetics workforce and another that provides a summary of the 2021 Updates in Health Disparities in Medical Genetics Symposium. Other highlights include a legislative update in the 117th Congress and much more.

ICYMI: ACMG Responds to State Laws Threatening Access to Safe Reproductive Options



ACMG recently responded to the US Supreme Court's 5 to 4 decision to allow a radical Texas abortion law (2021 Senate Bill 8) to go into effect despite standing legal challenges. The law severely restricts patients' access to safe reproductive healthcare options and subjects healthcare providers, their staff, and other citizens to punitive citizen lawsuits. Several months earlier ACMG responded to a decision of the US Circuit Court of Appeals for the Sixth Circuit upholding an Ohio law (2017 House Bill 214) that discourages patients from conveying health information to their physicians and penalizes physicians for providing abortion services if a patient informs them of a prenatal diagnosis or suspected diagnosis of Down syndrome.

Over the past few years numerous states have passed restrictive laws targeting patient access to the full spectrum of reproductive healthcare options. Some of the laws include broad abortion restrictions while others specifically target genetic conditions, and many of these focus on criminalizing healthcare providers and/or discouraging patients from sharing relevant health information with their physicians. These laws continue to be challenged as violations of the constitutional rights established under *Roe v. Wade*. With the exception of the Texas law, enactment of the majority of these laws has been placed on hold until the legal challenges are settled.

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DEI Committee Announces DEI Definitions – A Starting Point for Further Conversations

The vision of the American College of Medical Genetics and Genomics is that of empowering its members to be leaders in the integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. The assurance and celebration of diversity, equity and inclusion (DEI) is central to this vision, and to the commitment that all members of our community can thrive. As multiple

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

Website Links
ACMG
ACMG Foundation
ACMG Meetings Website
ACMG Education Center
NBSTRN
NCC
ClinGen
GIM

national efforts advance DEI in various sectors, definitions of DEI may differ between institutions; for this reason, the ACMG DEI Committee has provided the following definitions for the DEI terms. These definitions were compiled and expanded using statements, glossaries and other resources from various academic institutions. These terms are a starting point to engage in DEI conversations at the ACMG; however, we recognize they will likely evolve over time, and we are open to your feedback as we build honest conversations around the roles of DEI in medical genetics and genomics.

Education Committee Forms UME/GME Work Group

The ACMG Education Committee recently created a Medical Genetics in Undergraduate Medical Education (UME)/Graduate Medical Education (GME) Working Group to coordinate educational offerings from ACMG targeted to medical students, genetic counseling students and residents. In addition to a focus on students and trainees, the UME/GME Working Group will also consider how to support medical genetics educators across the country. Dr. Tracey Weiler, associate professor and academic director of the Graduate Certificate in Molecular and Biomedical Sciences at Florida International University, will lead the working group.

The group is working on or considering the following initiatives:

• Completion of the 2nd iteration of questions for the ACMG Student Challenge, created by Dr. Lauren Pronman —now available in the ACMG Genetics Academy.

• Development of ACMG Annual Meeting activities including the Trainee-Mentor Luncheon, Young Professional Trainee Workshop, and Careers in Medical Genetics.

• Creation of monthly/quarterly "Meet the Patient" sessions for students, focused on common genetic conditions that will be encountered by primary care practitioners (Dr. Pronman and Dr. Weiler).
• Development of a series of short videos about common medical genetics conditions that can be used in UME/GME curricula.
• Launch of a newsletter targeted to medical students, genetic counseling students and residents interested in genetics.
• Creation of a medical genetics educator mentoring program.

To learn more and get involved, please contact ACMG Education.

ACMG Forms New Special Interest Group for Medical Directors

The College is delighted to announce the formation of the new ACMG Medical Director's Special Interest Group (MD-SIG). The group will provide a discussion forum for physicians in a leadership position for a clinical genetics service to discuss topics related to clinical service. Medical directors face challenges when setting expectations for clinical productivity, or when asking for additional resources. This SIG will offer an avenue for leaders of clinical programs to share experiences, best practices and discuss topics such as faculty productivity expectations (e.g., patients per clinic, clinics per month, wRVU) and workflow (e.g., do you work with a genetic counselor? what does the genetic counselor do for a patient encounter vs. what does the physician do?) and even generate clinical and research collaborations.

The chair of the group is Nathaniel Robin, MD, FACMG, Medical Director for Genetics at the University of Alabama at Birmingham, and the co-chair is Rizwan Hamid, MD, Director for Pediatric Medical Genetics and Genomic Medicine at Vanderbilt University. To join the discussion listserv, please contact Dr. Robin.

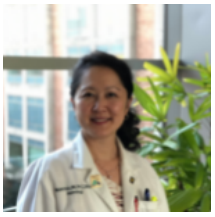
Introducing the New NIH-ACMG Fellows: Deepika Burkardt and Nguyen Park

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

NIH-ACMG fellows participate in rotations at the ACMG and the four participating components of the NIH including the National Human Genome Research Institute (NHGRI), National Heart, Lung, and Blood Institute (NHLBI), National Institute on Minority Health and Health

Disparities (NIMHD), and the *All of Us* Research Program. Fellows also design an elective rotation that comprises the final six months of their second fellowship year.

Deepika Burkardt, DO, BChE, BS is a clinical and biochemical geneticist in her first year of fellowship training. During her current ACMG rotation, Dr. Burkardt is working with the NBSTRN to understand opportunities for screening for treatable causes of abnormal growth among newborns, and ClinGen to learn about curating gene-disease associations based on gene dosage and somatic variation. “I am grateful for this opportunity to learn about a variety of efforts to continue to improve care across the lifespan,” she said about the fellowship program. “It helps me gain perspective on strategically planning and implementing successful genomic initiatives beyond fellowship. It also gives me the chance to continue to work on understanding disorders of growth and development, learn from our amazing patients and their families, and teach future geneticists.”



During her rotation at ACMG, **Nguyen Park, MS, PA-C, DFAAPA** has worked with both NCC and NBSTRN. With NCC she has helped on the Genetics Policy Hub, connecting stakeholders across the PA profession, as well as on developing preliminary surveys of both non-genetics/genetics PAs and PA students prior to the 2023 ACMG Workforce Study. With NBSTRN she is working to increase awareness of NBS programs among clinicians through a presentation to the American Academy of Physician Assistants. In addition, she is providing clinical input on provider-targeted information on the NBSTRN website. She is looking forward to working with Dr. Paul Liu, MD, PhD at NHGRI on his RUNX1 natural history study. “I am so honored to be the first PA chosen for the fellowship,” said Ms. Park, “and to be able to participate with so many of ACMG and NHGRI’s cutting edge programs - bringing my unique perspective as a clinician and highlighting the role PAs can play in genetics.”

ACMG Foundation Now Accepting Nominations for Rimoin and Watson Awards

Nominations are due November 15, 2021 for the 2022 ACMG Foundation for Genetic and Genomic Medicine's (ACMGF) David L. Rimoin Lifetime Achievement Award in Medical Genetics. This award honors the legacy of Dr. Rimoin, ACMG’s founding president. The most distinguished of the ACMG Foundation's professional awards, it recognizes individuals whose careers exemplify a lifetime of achievements and the personal qualities embodied by Dr. Rimoin. Visit our website to learn more about the Rimoin Lifetime Achievement Award. Click here to nominate a colleague today.

Nominations are due November 15, 2021 for the 2022 Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award. Do you know an early career genetics and genomics professional who has demonstrated innovation in their work through the development or implementation of a new concept, method or idea that has had significant impact on genetic and genomic medicine? As an innovation award, this honor is open only to nominees who have made contributions to the field of genetics and genomics for less than 10 years. To nominate a colleague or yourself visit here.

To learn more about ACMG professional awards visit our website.

Now Available: ACMG Foundation Summer Scholars Program Institutional Applications

Institutions can apply to host a scholar for the 6-week 2022 ACMG Foundation Summer Genetics Scholars Program until October 29, 2021. This innovative ACMG Foundation Program is designed to identify and encourage highly qualified medical students to consider careers in medical genetics by providing a variety of hands-on experiences in clinical and laboratory medicine, research, and services. To learn more, visit our website.

It's Almost Time for the 2021 ACMG Salary Survey of Board-Certified Medical Geneticists

ACMG will soon launch our biannual salary survey of board-certified medical geneticists. This year the survey will be administered by Infosurv Research. All ACMG fellows and candidate fellows are urged to participate. Your participation in the survey is critical to the continued growth and development of medical genetics and its subspecialties. Such surveys are being used by individual ACMG members as well as department heads, chairs, and deans to influence salaries and remain competitive. This survey has direct and immediate impact on the ability to grow the field and on your ability to have data to support discussions within your institutions.

To be sure you receive your survey invitation from Infosurv Research, **please add survey@infosurvfeedback.com to your list of approved senders**.

ACMG Seeks Volunteers for New International Outreach Taskforce

ACMG is organizing a new taskforce on international outreach whose purpose will be to determine if globalization is appropriate for the College and to develop a proposal regarding how ACMG could expand its international reach. The long-term goal of the taskforce will be to increase the number of ACMG members from diverse backgrounds, raise awareness of ACMG activities (and therefore become a global leader in clinical genomics) and increase revenue generated by offering educational products for international institutions that lack clinical and/or laboratory genetics training programs. The taskforce is seeking members with diverse perspectives and innovative ideas. Madhuri Hegde, PhD, FACMG, Sr. Vice President and Chief Scientific Officer, Global Lab Services at PerkinElmer, Inc. will be leading the taskforce. If you are interested in being considered for membership on the taskforce, please email a letter of interest and CV with the subject line: Taskforce on International Outreach and Engagement.

ACMG In the News: New Clinical Practice Resource Cited in *New York Times*

On August 17, 2021, *The New York Times* published an article entitled "This Breast Cancer Gene is Less Well Known, but Nearly as Dangerous," which cited ACMG's new clinical practice resource "Management of individuals with germline variants in PALB2" and quotes Douglas R. Stewart, MD, FACMG, one of the authors of the ACMG paper. The resource, which appeared in *Genetics in Medicine*, was developed under the auspices of the ACMG Professional Practice and Guidelines Committee. *The New York Times* reporter focused on how relatively unknown *PALB2* is among the lay public compared to *BRCA1* and *BRCA2* and emphasized that it is almost as important in terms of screening for breast, ovarian and pancreatic cancer risk.

September GenePod: How Experts Convened to Provide ACMG's First Evidence-based Clinical Guideline

Genetics inMedicine GENEPOD

Congenital anomalies (CA), developmental delay (DD), and intellectual disability (ID) are among the most common indicators in children that lead to genetic testing. Identification of an underlying diagnosis for CA or DD/ID can be consequential to care management and long-term prognosis for the child. But there has been no evidence-based guideline for clinicians to refer to

that supports the use of exome or genome sequencing as a first-line or second-line test for the evaluation of pediatric patients with CA or DD/ID.

On this month's GenePod, Fuki Hisama, MD, FACMG, FAAN and Murugu Manickam, MD, FACMG, who co-chaired the ACMG's evidence-based work group, discuss how a team of experts was brought together to provide the ACMG's first-ever evidence-based clinical guideline. The guideline lays out clear recommendations for use of exome or genome sequencing in clinical care to optimize outcomes for pediatric patients with CA or DD/ID. "In a way, this model of an evidence-based guideline is creating the standard and a template for future studies," says Dr. Manickam.

ACMG Welcomes 13 New Members

ACMG welcomes and congratulates its 13 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 of our members who make important contributions and the work of the College possible through your membership, support, and involvement.

Candidate Fellow
Rizwan Yousaf, PhD, MS

Affiliate Member
Sandra T. Cooper, PhD
Geoffrey Ginsburg, MD, PhD
Julie C. Moreau, MSN
Laura B. Ramsey, PhD
Naw Wah Wah, PhD

Trainee Members
Jennifer Cassady, MD
Benjamin E. Kang, PhD
Christian Parobek, MD, PhD
Tam P. Sneddon, PhD
Katarzyna Thompson, D_Sci

Student Members
Ashley E. Blau, MA
Taylor Nicole Young

Do you know someone who should join ACMG? Please invite them to visit our Join ACMG webpage.

ACMG Member Comment: Clinical Pharmacogenomic Testing Technical Standard

The ACMG Board of Directors requests your comments on a new laboratory testing standard, developed under the auspices of the Molecular Genetics Subcommittee of the Laboratory Quality Assurance Committee:

Clinical Pharmacogenomic Testing and Reporting: A Technical Standard of the American College of Medical Genetics and Genomics (ACMG)

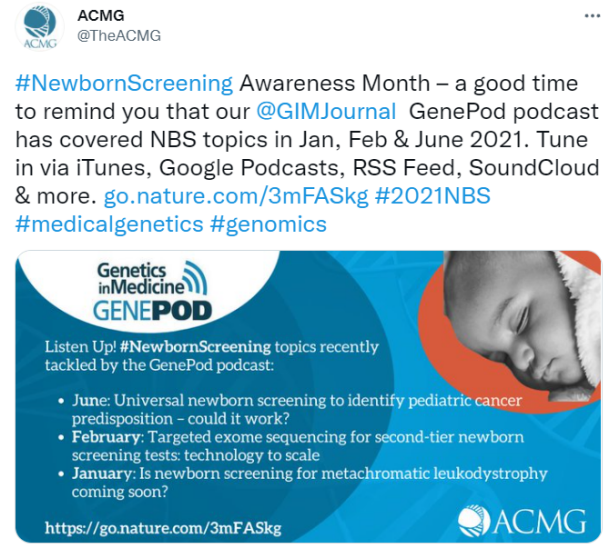
When submitting comments for review, please provide your full name, degrees, affiliation and contact information. All comments should reference the precise line numbers to which they pertain. Comments should be forwarded, via email, to Documents@acmg.net by Friday October 22, 2021 at 7:00 AM ET. The subject line of your emailed comments for this document should read: "Clinical Pharmacogenomic Testing Technical Standard".

READ MORE

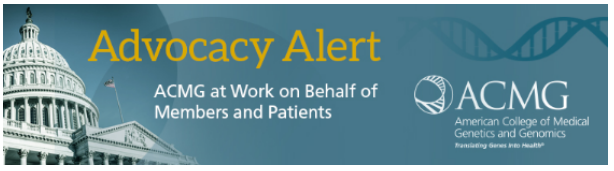
ACMG Promotes Newborn Screening Awareness Month

ACMG is promoting Newborn Screening Awareness Month throughout September across multiple social media platforms including Twitter,

Facebook, Instagram and LinkedIn. Joining other organizations in using the hashtag #2021NBS, the College will highlight several of its NBS resources including recent NBS topics covered by *GenePod*, the podcast from our official journal *Genetics in Medicine*; advocacy for the bipartisan Newborn Screening Saves Lives Reauthorization Act; and the collection of ACMG and NCCRCG ACT Sheets concerning NBS. We invite you to follow the the College's #2021NBS posts on social media and join the conversation. Here are some examples of recent posts:



Update on Congressional Newborn Screening Reauthorization Legislation



Since September is Newborn Screening Awareness Month, we wanted to provide an update on the Newborn Screening Saves Lives Reauthorization Act (NBSSLRA), legislation that would reauthorize and fund important newborn screening programs operated by the CDC, the NIH and the HRSA. Newborn screening is one of the most successful public health programs in the country, and funding these programs is critical for their continued success.

Earlier this year, the U.S. House of Representatives passed the NBSSLRA (H.R. 482), but action is still needed in the Senate. ACMG

has been working closely with other provider and patient organizations to encourage the Senate to pass the NBSSLRA. Earlier this year ACMG members helped by sending more than 200 letters to 58 Senators. During Newborn Screening Awareness Month, help raise awareness about the NBSSLRA and our call for the Senate to pass this legislation. You can share social media posts about the NBSSLRA (#NewbornScreeningSavesLives), tag your Senators on Twitter, or even reach out to them directly by phone or email to ask them to co-sponsor the NBSSLRA.

Innovations from Technology, Advocacy and Clinical Care: NBSTRN 2021 NBS Research Summit

NBSTRN | Newborn Screening Translational Research Network

In celebration of Newborn Screening Awareness Month, NBSTRN hosted its 2021 Newborn Screening Research Summit on September 1-3. The three-day summit featured nine sessions from leaders in genomics, ethics, advocacy, and clinical research. Hundreds of viewers from across the United States and other countries discussed emerging findings and practices. The NBS Research Summit directly advances NBSTRN's vision to facilitate and support ground-breaking research to accelerate understanding of genetic disease and foster collaborations with clinicians, families and health professionals. A program, complete with biographies for each speaker and abstracts for their presentations can be found on the summit's webpage.

Several talks referenced NBSTRN initiatives, including Dr. Aaron Goldenberg, Case Western Reserve University, whose presentation highlighted an effort by one of the five NBSTRN expert workgroups, the NBSTRN Bioethics & Legal Workgroup, to improve the design of NBS pilots: "Including ELSI Research Questions in Newborn Screening Pilot Studies."

Recordings of 2021 NBS Research Summit presentations can be found on the summit's webpage or NBSTRN's YouTube channel.

READ MORE

200th Birthday Celebration for Founder of Genetics Gregor Mendel

Gregor Mendel (born Johann Mendel on July 22, 1822), whose scientific work was largely not understood during his own time, became recognized as the founder of genetics 35 years after his death in 1884. Over the next year, marking Mendel's 200th birthday, many in the medical genetics community will celebrate him in order to foster more awareness of his life, achievements, and his laws of genetics.

Various activities over the next year, coordinated in part by John J. Mulvihill, MD, FACMG, will emphasize Mendel's lessons for today. A friar and abbot of St. Thomas' Abbey in Brno (located in today's Czech Republic), Mendel had the ability to apply quantitative tools from physics and mathematics to qualitative biologic data, to anticipate the mechanisms of Darwinian evolution, and to improve horticulture and animal breeding – all while being a faithful practicing priest and leader of his religious community. An exemplar of mentoring, Mendel thrived at the interdisciplinary interfaces of botany, evolution, mathematics, and physics, while being a scientist-citizen, a community and social activist against inequality, and bank director and president. He illustrates the parallel and compatible realms of science and religion and the pros and cons of solitary science and team science.

Watch for an update on the Mendel celebration in the Winter edition of *The ACMG Medical Geneticist* member magazine and primarily on this international website. Your own ideas for local activities are welcome and can be sent to Dr. Mulvihill.

Back to School: ACMG Student and Trainee Opportunities

As medical and genetic counseling students and trainees settle into fall, it is an excellent time for you to remind those at your institution that ACMG offers a free student membership as well as an array of resources tailored to their needs, including:

• The College's Student Interest Group (SIG) Program provides access to unique educational resources and opportunities for networking and is currently available at 36 medical schools or teaching institutions across the country. The opportunity is always open for other institutions to form a SIG.
• The ACMG Foundation sponsors the Summer Genetics Scholars Program each year. Ten highly qualified students were selected for a six-week mentored experience in 2021.
• The ACMG Genetics Academy contains a free bundle of outstanding recorded workshops and TED-style talks geared to students and trainees and delivered by medical genetics experts.

Please check out the College's full array of opportunities available to students and trainees on the ACMG wesbsite.

Now Available in the ACMG Genetics Academy

2021 ACMG Student Challenge

ACMG invites all medical genetic students, residents, and genetic counseling trainees to participate in our 2021 Student Challenge. Each month, we will release a student challenge question, starting July 1, 2021 through June 30, 2022. These questions are great opportunities to learn more about clinical genetics and gain knowledge about clinical genetics and inborn errors of metabolism.

To sign up for the challenge, log in and register here.

New Part IV CCP Modules

• **Evaluation of the individual with Suspected Marfan Syndrome (version 2021):** For clinical geneticists who are involved in the diagnosis and counseling of patients with suspected Marfan syndrome.
Purchase Now

• **Neurofibromatosis-Type 1 (NF1) (version 2021):** For geneticists who are involved in the initial and ongoing care of patients with Neurofibromatosis type 1. This does NOT include the evaluation of patients for possible Neurofibromatosis but the ongoing care of diagnosed patients.
Purchase Now

• ***BRCA1* and *BRCA2*-Associated Hereditary Breast and Ovarian Cancer:** This module evaluates clinical practice in the care of patients who are either affected and/or have a family history of breast and ovarian cancer.
Purchase Now

• **Chromosome and FISH Analyses of Patients with Suspicion of Turner Syndrome:** This module provides an overview of recommendations for metaphase chromosome analysis of patients with suspicion of Turner syndrome. Chromosome analysis, with adjunct fluorescence in situ hybridization (FISH) and/or chromosomal microarray, can diagnose Turner syndrome and identify the multiple different abnormal karyotypes that may be present. The aim of this module is to encourage appropriate work-up that will detect mosaicism and other karyotypic variations that may impact clinical outcome.
Purchase Now

ABMGG Longitudinal Assessment Program CertLink

ABMGG CertLink meets ACCME requirements for continuing medical education with assessment. Through our partnership with ABMGG, ACMG will award ten *AMA PRA Category* 1 Credits™ each year (5 credits/period) to diplomates participating in ABMGG CertLink.

For more information visit the ABMGG Certilink Pilot Program website.

There's Still Time to Purchase the 2021 ACMG Digital Edition

The **2021 ACMG Digital Edition** is available for purchase in the ACMG

Genetics Academy with content available to view immediately after purchase. The Digital Edition includes access to videos of course presentations, slides, and the ability to claim CME, P.A.C.E.®, and NSGC credits.

If you purchased the extended access when registering for the April virtual meeting, you should have received instructions by email from education@acmg.net on July 16th to access the content. If you did not receive the instructions, please email education@acmg.net.

Purchase the 2021 ACMG Short Courses Digital Edition

• A Clinician's Perspective on Obesity: Syndromic and Non-Syndromic Causes, Treatment and Challenges: Purchase Now
• Integration of Functional Genomics to Improve Variant Interpretation and Diagnosis: Purchase Now

Free Resources Now Available

• **2021 Abstracts and Poster Gallery**
• **2021 Satellite Symposia Digital Edition**

ClinGen Somatic Cancer and VICC Virtual Molecular Tumor Board Case Series

September 28 (4th Tuesday), 11:00 am – 12:00 PM ET
All medical and healthcare professionals and researchers interested in understanding cancer genomic testing and somatic and germline variant interpretation methods. This series is presented as a collaboration between ClinGen Somatic and VICC consortia, and ACMG.

Register Now!

2022 ACMG Annual Clinical Genetics Meeting Updates

Call for Abstracts – Deadline November 19

ACMG invites the submission of abstracts to be considered for poster and selected platform presentations at the 2022 ACMG Annual Clinical Genetics Meeting. The online submission site will open October 1, 2021. The submission deadline is November 19, 2021, 11:59 PM PST.

All abstracts must be submitted online at www.acmgmeeting.net.

Abstracts will be posted online on the ACMG Meeting Website and published as a supplement of the March issue of *Genetics in Medicine*, the College's official journal. Posters will be available online via an ePoster Gallery and onsite in the Exhibit Hall in March. Platform presentations will be presented during the in-person event – some will be live streamed for remote attendees, but all will be available on demand shortly after the live presentation.

Research and/or studies must be in one of the following ACMG 2022 Abstract Categories:

• Clinical genetics and therapeutics
• Cancer genetics and therapeutics
• Biochemical and metabolic genetics
• Laboratory genetics and genomics
• Genetic counseling
• Prenatal genetics
• Public health, health services and implementation

• Social, ethical and legal issues, education

Registration and Housing to Open in October

Watch your email for the meeting registration and hotel reservation announcement. Be sure to register early and book your hotel at the same time. Registration for the meeting is required prior to making hotel

reservations. Reservations must be booked through ACMG's Housing Partner – CMR.

Important Dates and Deadlines

• Registration, Housing and Abstract Submission Opens: October 2021
• Online Abstract Submission Deadline: November 19, 2021, 11:59 PM PST
• Early Bird Registration Deadline: Wednesday, December 22, 2021
• Advance Registration Deadline: February 9, 2022
• Late Registration Fees Apply: February 10, 2022
• Hotel Reservation Deadline: March 2, 2022

Detailed information will be updated on a regular basis on the ACMG Meeting website, so please check often for exhibitor, speaker and session updates.

American College of Medical Genetics and Genomics | 7101 Wisconsin Avenue, Suite 1101, Bethesda, MD 20814

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