ACMG In Action

Essential Updates: Member News You Can Use

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

August 2020

In This Issue...

ACMG Advocacy Updates

GAO Releases Report on the Medical Genetics Workforce

In response to a request from Congress, the US Government Accountability Office (GAO) recently performed a nationwide assessment of the clinical geneticist and genetic counselor workforce. Their final report, "GENETIC SERVICES: Information on Genetic Counselor and Medical Geneticist Workforces (GAO-20-593)," is publicly available here. The GAO assessment request was presented to the House Appropriations Committee by Representative Herrera Beutler (R-WA) and ultimately included in report language for the 2019 Departments of Labor, Health and Human Services (LHHS) appropriations legislation. Recognition of the need for such an assessment was a direct result of ACMG advocacy outreach with members of Congress.

To complete their assessment, the GAO reviewed data available from multiple sources such as the relevant certifying and accreditation organizations, the Bureau of Labor Statistics, and recent workforce surveys. This included a recent survey performed by ACMG and the National Coordinating Center for the Regional Genetics Networks (NCC) to assess clinical genetics practices. The NCC, funded through a cooperative agreement between the Health Resources and Services Administration (HRSA) and ACMG, organized a Medical Genetics Workforce workgroup to develop the survey and analyze the results. A small subgroup is currently finalizing a manuscript that will provide an updated estimate of the number of clinical geneticists in the workforce. The workgroup included representation from ACMG, the Regional Genetics Networks, the American Board of Medical Genetics and Genomics (ABMGG), the National Society for Genetic Counselors (NSGC), and the Society for Inherited Metabolic Disorders (SIMD), among others.

- Save the Dates: 2021 ACMG Annual Clinical Genetics Meeting

Upcoming Events

2020 ACMG Annual Clinical Genetics Meeting – Digital Edition

2021 ACMG Annual Clinical Genetics Meeting
April 13-17, 2021
Los Angeles Convention Center
Los Angeles, California

Website Links

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

READ MORE

COVID-19 Provider Relief Extended

In June, the US Department of Health and Human Services (HHS) announced phase 2 of its COVID-19 Provider Relief Fund distribution plan in which eligible Medicaid, Medicaid managed care, Children's Health Insurance Program (CHIP), and dental providers could apply for funding of up to 2% of reported revenue from patient care. To allow more providers time to complete their application, HHS had extended the deadline to August 28, 2020. The goal is to reach providers who were not covered by the phase 1 general distribution that automatically went out to Medicare providers.

HHS also announced that it is allowing Medicare providers who missed the opportunity to apply for additional funding under the phase 1 distribution a second chance to apply. This largely applies to providers who do not submit comprehensive cost reports with the Centers for Medicare and Medicaid Services (CMS) and were asked to submit revenue information to receive their phase 1 payment. The deadline for these applications is also August 28, 2020. See the complete HHS announcement here and additional information about applying for the Provider Relief Fund here.

ACMG Publishes New Practice Resource on Treatment of Mucopolysaccharidosis Type II (Hunter Syndrome)

Last week, ACMG announced the publication of a new practice resource entitled "Treatment of Mucopolysaccharidosis Type II (Hunter Syndrome): A Delphi Derived Practice Resource of the American College of Medical Genetics and Genomics (ACMG)." This practice resource was developed by the ACMG Therapeutics Committee.

Mucopolysaccharidosis type II (MPS II, MIM 309900) is a severe lysosomal storage disease with multisystem involvement. Based on an evidence-based review and subsequent literature, a Delphi study was conducted to generate an MPS II clinical practice resource. Recommendations from the Delphi process were generated, and areas were highlighted that warrant further study to help guide clinical care. Some recommendations from the study include: all individuals with severe MPS II or predicted to have severe MPS II based on genotype warrant starting ERT (enzyme replacement therapy), prior to showing signs or symptoms; individuals with signs or symptoms with either attenuated or severe MPS II warrant ERT; individuals with attenuated MPS II who are not showing signs or symptoms of disease do not warrant ERT; clinical evaluation of liver and spleen size are recommended for judging clinical effectiveness of treatment, with optional use of imaging modalities (ultrasound or MRI of the abdomen) to follow organ size.

The College extends its appreciation to Kim L. McBride, MD, MS, FACMG (workgroup chair); Susan A. Berry, MD, FACMG; and Nancy

Braverman, MD, MS, FACMG for their work on this practice resource.

Applications Now Being Accepted for the 2021 ACMG Foundation Next Generation Fellowship and Residency Awards

The ACMG Foundation for Genetic and Genomic Medicine is now accepting applications for the 2021 ACMG Foundation Next Generation Fellowship and Residency Awards. The purpose of these awards is to:

• advance education, research and standards of practice in medical genetics;
• develop and expand clinical and laboratory expertise in medical genetics in the United States; and
• initiate and develop a broad-based infrastructure for industry funding of high-quality projects in the field of medical genetics.

For more information and to apply for the ACMG Foundation Next Generation Clinical Genetics and Genomics Residency Award or the Clinical Laboratory Biochemical Genetics Fellowship Award, click here. The deadline for applications is August 28, 2020.

For more information and to apply for the ACMG Foundation Next Generation Medical Biochemical Genetics Subspecialty Fellowship Award, click here. The deadline for applications is October 16, 2020.

Reviews will be conducted this summer and fall. Awardees will be notified in the fall. Residency/Fellowships will begin by July 1, 2021.

August 2020 GenePod: A Discussion of ACMG's Recent Guidance on the Integration of Genomic Information into the EHR

Genetics inMedicine GENEPOD

Genetic and genomic information is a powerful tool in personalized medical care. It is essential for both diagnostic purposes and medical management. But when it comes to genetic test results, electronic health records (EHRs) are generally not searchable or standardized. Tune in to this month's GenePod, *Genetics in Medicine*'s monthly podcast, to hear co-authors Dr. Theresa Grebe, a clinical geneticist at Phoenix Children's Hospital, and Dr. Marc Williams, director emeritus of the Genomic Medicine Institute at Geisinger, discuss ACMG's recent points to consider statement on the interface of genomic information with the EHR and how to optimize EHRs for genomic medicine.

Have You Seen *Genetics in Medicine*'s SARS-CoV-2/COVID-19 Collection?

Genetics in Medicine (*GIM*) is excited to announce our SARS-CoV-2/COVID-19 collection of articles, podcasts and resources.

GIM's goal has been to ensure timely distribution of important SARS-CoV-2/COVID-19 research and commentaries by expediting review and publication of relevant articles submitted to the journal. All articles related to COVID-19 published in the journal are freely available.

Featured in this collection are podcasts in which you will hear experts discuss international collaborations aimed at finding genetic clues to COVID-19's variable disease path, as well as challenges faced by geneticists and metabolic specialists in care of patients during this pandemic. The collection also provides access to resources from our publisher, Springer Nature, such as free data deposition services or help finding suitable repositories for COVID-19 data. We are also proud to highlight our editors who are contributing to the fight against COVID-19 every day in their work in the medical genetics and genomics community. Take a look at *GIM*'s collection of COVID-19 resources!

Nasha Fitter Elected to ACMG Foundation Board of Directors

Nasha Fitter, director of Rare and Neurological Diseases at Ciitizen and co-founder, CEO and head of research at FOXG1 Research Foundation, was recently elected to the board of directors of the ACMG Foundation for Genetic and Genomic Medicine. The ACMG Foundation is a national nonprofit foundation dedicated to facilitating the integration of genetics and genomics into medical practice. The board members are active participants, serving as advocates for the ACMG Foundation and for advancing its policies and programs. Ms. Fitter was elected to a two-year term starting immediately.

ACMG Foundation President Bruce R. Korf, MD, PhD, FACMG said, "I am delighted to welcome Nasha Fitter to the ACMG Foundation board as a public member. Nasha has a passion for improving the lives of individuals who are affected with genetic conditions, and also has extraordinary skills in business, education and technology. She is superbly qualified to represent the interests of the public on the ACMG Foundation board."

About being elected to the ACMG Foundation Board of Directors, Ms. Fitter said, "In the next few years we will see the immense power of genetic medicine in saving and transforming people's lives. I am thrilled

to be joining an organization at the forefront of this incredible science and look forward to working with such a diverse and experienced board."

To learn more about Ms. Fitter and the ACMG Foundation board, click here. To view a complete roster of the ACMG Foundation Board of Directors, click here.

ACMG Welcomes 15 New Members

ACMG welcomes and congratulates 15 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
Dwight Koeberl, MD, PhD, FACMG

Corresponding Fellow
Karin Weiss, MD, FACMG

Associate Members
Amy Dobson, MS, CGC
Luanne M. Fraer, MS, CGC

Affiliate Members
Suwicha T. Chitpatima, PhD
Willow Sheehan, DNP
Lisa Anne Shephard, BS
Holly Tabor, PhD
Huntington F. Willard, PhD
Masaaki Yamada, MD

Student Members
Mianne Lee, MS
Chelsea Lowther, PhD
AnnaLisa V. Wilson, MPA
Christina M. Wright, PhD
Changrui Xiao, MD

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

It's a Wrap! ACMG Completes the 2020 Educational Sessions of the Alternative Pathway to Board Certification in LGG Training Program

ACMG congratulates the more than 120 participants who completed the 2020 ACMG live/hybrid educational sessions of the

Alternative Pathway to Board Certification in Laboratory Genetics and Genomics (LGG) Training Program. Individuals enrolled in the program are cross-training to sit for the American Board of Medical Genetics and Genomics (ABMGG) LGG Board Exam. ACMG is pleased to provide the educational components of the training.

In June, ACMG held the live Zoom meetings for each track's (Cytogenetics and Genomics, and Molecular Genetics and Genomics) "Mentored Cases," with 15 ACMG Expert Fellows. In July, ACMG held the Foundational Specialty Training hybrid events, including live Q&A, with seven ACMG Expert Fellows.

ACMG in the News

On July 23, 2020, the American Heart Association (AHA) published a new scientific statement entitled "Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association." In this statement, the AHA outlined current best practices for genetic testing of patients with confirmed or suspected inherited cardiovascular disease and offered guidance for disease-specific genetic testing. Citing ACMG's "Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics," the AHA also summarized current best practices for the evaluation of individuals with secondary findings, noting that of the 59 genes deemed medically actionable by the ACMG, 30 are related to cardiovascular diseases. Both *Modern Healthcare* and GenomeWeb reported on AHA's statement, noting ACMG's guidance on the reporting of secondary findings from clinical exome and genome sequencing.

NBSTRN Webinar Focuses on SMA Pilot Studies

Spinal muscular atrophy (SMA) is a genetic disease affecting the central nervous system, peripheral nervous system and voluntary muscle movement (skeletal muscle). Research has shown that early detection and treatment of the most severe types of SMA is most effective when started in the first few months of life. Although there is currently no cure for SMA, treatments such as enzyme replacement and gene therapy can be used to manage symptoms and improve quality of life. The availability of these treatments led to early pilot studies of newborn screening for SMA. Based on data from these studies, SMA was recommended for nationwide newborn screening in 2018. Today, 31 states screen for SMA, covering about 65% of all newborn babies in the US.

The Newborn Screening Translational Research Network (NBSTRN) hosts a monthly webinar on Newborn Screening Pilot Studies in which researchers, state program representatives, advocacy organizers,

clinicians, the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories (APHL) present up-to-date findings from newly implemented newborn screening initiatives. Webinars are recorded and posted for public viewing on the NBSTRN YouTube channel. In the July 2020 webinar, representatives from several state newborn screening laboratories presented data on the number of cases screened positive for SMA and other candidate conditions. Sharing these updates allows state laboratories to learn from each other about best practices for implementation of newborn screening for new conditions.

ACMG Social Media Highlights

Do you tweet, post and share? ACMG does. The College actively participates on Twitter, Facebook, Instagram, YouTube, and LinkedIn. ACMG also hosts a private, members-only group on LinkedIn and an ACMG Annual Clinical Genetics Meeting group on LinkedIn. Through these pages, ACMG provides followers with updates and news about the various events and programs of the College and the ACMG Foundation, earning nearly 125,000 impressions each month. The College invites you to follow ACMG on social media and join the conversation. We are pleased to share here a few of our posts from the past month.

ACMG - American College of Medical Genetics and Genomics

Citing ACMG statements and guidelines, a new scientific statement from the American Heart Association outlines best practices for #genetictesting of patients with confirmed or suspected inherited #cardiovascular disease, and for evaluation of individuals with #secondaryfindings from #exomesequencing or #genomesequencing #cardiogenetics

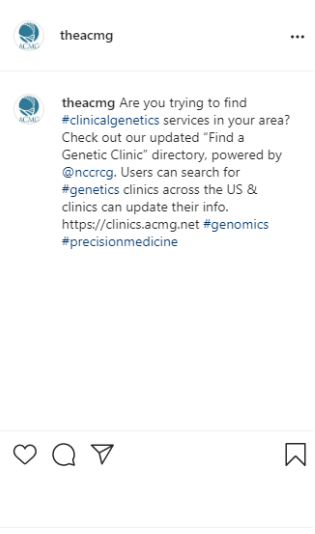
American Heart Association Like Page

Some cardiovascular diseases may be inherited, and a new American Heart Association scientific statement says genetic testing can help patients and families identify risks. Have you ever thought about genetic testing?

About this website

HEART.ORG

Genetic testing a tool for families dealing with certain heart diseases

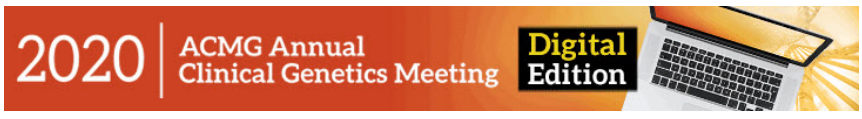


NBSTRN to Host NBS Virtual Summit during Newborn Screening Awareness Month

Join the Newborn Screening Translational Research Network (NBSTRN) September 8-10, 2020, from 9 AM to noon PST, for the three-day NBSTRN Newborn Screening (NBS) Virtual Summit. This summit offers participants the opportunity to learn the latest newborn screening research from leading experts, state programs, industry, and parent advocates; connect and expand their research networks; and support newborn screening programs. Register today on the NBSTRN website. Registration is free!

The NBSTRN is funded by a contract from the Eunice Kennedy Shriver *National Institute of Child Health and Human Development (NICHD), National Institutes of Health (NIH) to the ACMG with the goal of advancing newborn screening related research.*

View the 2020 ACMG Annual Meeting – Digital Edition Anytime, Anywhere



The 2020 ACMG Annual Clinical Genetics Meeting – Digital Edition is an online blended learning format available through the meeting's website

and the ACMG Genetics Academy. The Digital Edition provides access to the educational content, posters and abstracts that were planned for the 2020 ACMG Annual Meeting, and offers more than 50 hours of continuing education credit (*AMA PRA Category 1 Credits*™, NSGC CEUs, and P.A.C.E.® CEUs).

Access the Recorded 2020 Satellite Symposia Sessions and Exhibit Theaters for Free!

The 2020 ACMG Annual Meeting recorded Satellite Symposia sessions are now available for free to all. Simply go to the ACMG Genetics Academy and log in with your ACMG account to access the symposia. *AMA PRA Category 1 Credits*™ are available, but NSGC CEUs and P.A.C.E.® CEUs are not available with the free version.

In addition, many of the Exhibit Theater sponsors recorded their presentations. These presentations are now available to anyone—purchase of the 2020 ACMG Annual Meeting – Digital Edition is not required. To see a list of the recordings, click here.

Highlights of the ACMG Annual Meeting – Digital Edition Include:

Scientific Session Recordings
Platform Presentations
Abstracts and Poster Gallery
Exhibit Theaters
Satellite Symposia
Exhibit Hall

Access to the complete Digital Edition, including scientific session recordings, is available for purchase in the ACMG Genetics Academy.

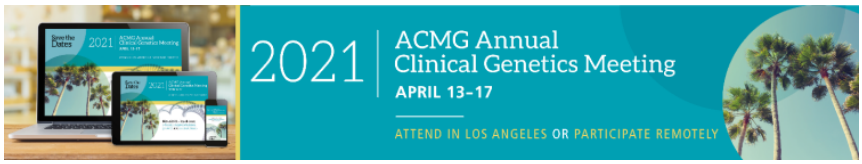
New ACMG Student Challenge Question Released

The ACMG Education and CME Committee is pleased to provide medical and medical genetics students and trainees, and genetic counseling students with a monthly **Student Challenge question**. Each month, starting in July 2020 and continuing through June 2021, a new student challenge question will be released. These questions are great opportunities to learn more about clinical genetics and inborn errors of metabolism.

Participation in the ACMG Student Challenge is **free** and open to all medical, medical genetics, and genetic counseling students and trainees.

To sign up for the challenge, log in and register in the ACMG Genetics Academy. Miss the first month's question? No problem, past months' questions will remain available for catch up learning.

Save the Dates: 2021 ACMG Annual Clinical Genetics Meeting



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

Telephone: (301) 718-9603 | Fax: (301) 718-9604 | Privacy Policy | Feedback