ACMG In Action

Essential Updates: Member News You Can Use

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

July 2020

In This Issue...

Genetics in Medicine Receives Impact Factor of 8.904 for 2019 —Journal Is Ranked 13th of 177 Journals in Genetics & Heredity

ACMG is pleased to announce that *Genetics in Medicine* (*GIM*), the official journal of the ACMG, has received an Impact Factor of 8.904 for 2019. The 2019 Journal Impact Factors were published by Clarivate Analytics in the latest edition of *Journal Citation Reports*. This is the second highest Impact Factor in the journal's history. *GIM* ranks 13th of 177 titles in the Genetics & Heredity category.

The Impact Factor is an objective measure of the world's leading journals, based on articles' cited references, and is oft considered a measure of a journal's impact, overall successful performance and relevance to its field. The most highly cited article in *GIM* in 2019 was "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."

"*GIM*'s editors and editorial staff are delighted that our Impact Factor has increased from last year. This improvement in the Impact Factor once again demonstrates that the journal remains one of the most widely read and cited journals publishing clinically relevant research in the life sciences," said *GIM*'s Editor-in-Chief Robert D. Steiner, MD, FAAP, FACMG. "We are most thankful to the peer reviewers who put in countless hours to help maintain the outstanding quality of articles and the authors who trust us to disseminate their groundbreaking scholarly work. The Impact Factor is one of a number of metrics used to evaluate journals, and a journal should not be evaluated solely on that one metric. *Genetics*

Upcoming Events

ACMG Annual Clinical Genetics Meeting – Digital Edition Webinar Series
July 20, 2020, 3:30 PM EDT

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

in Medicine's continued success and relevance is also reflected in our very high overall downloads and reads as well as a prominent social media presence."

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Milestones 2.0 for Laboratory Specialties Announced by ABMGG and ACGME

Accompanying the recent and ongoing transition of laboratory training program accreditation from the American Board of Medical Genetics and Genomics (ABMGG) to the Accreditation Council for Graduate Medical Education (ACGME), revised milestones for the Laboratory Genetics and Genomics (LGG) and Clinical Biochemical Genetics (CBG) specialties have been developed and approved by the ACGME. These are effective July 1, 2020.

Milestones are integral to competency-based medical education (CBME), providing guidance for the expectations of a trainee's progress in the program. CBME curriculum design and assessment methods are based on the individual's skills and growth. Unlike traditional education, CBME is not time-oriented and allows for individualized feedback, coaching and reflection. In addition, the Supplemental Guides accompanying the specialty Milestones offer definitions, examples and references to facilitate their integration and use in the training process.

The Milestones 2.0 documents were developed by the LGG and CBG Milestones Working Groups, which were comprised of program directors, experts involved in training, and current fellows (listed below). While LGG and CBG are distinct specialties, there is an overlap in the developmental skills involved so these groups met separately and together, allowing for cross-pollination and depth of experience, and assuring consistency in the final documents.

The LGG and CBG Milestones 2.0, Supplemental Guides and other resources can be found at the ACGME website.

Members of the CBG and LGG Milestones 2.0 Working Groups are:

CBG: Miriam Blitzer, PhD, FACMG; Kerri Bosfield, MBBS (CBG fellow); Tina Cowan, PhD, FACMG; Kristina Cusmano-Ozog, MD, FACMG; Laura Edgar, EdD, CAE; Sarah H. Elsea, PhD, FACMG; Rebecca Ganetzky, MD; Alejandro Iglesias, MD; Marzia Pasquali, PhD, FACMG; and Daniel Sharer, PhD, FACMG.

LGG: Swaroop Aradhya, PhD, FACMG; Miriam Blitzer, PhD, FACMG; Theresa Brown, PhD, FACMG; Josh Deignan, PhD, FACMG; Laura Edgar, EdD, CAE; Celeste Eno, PhD (LGG fellow); Colleen Jackson-Cook, PhD, FACMG; Yuan Ji, PhD, FACMG; Azra Ligon, PhD, FACMG; and Stephen Moore, MBA, PhD, FACMG.

ACMG Advocacy Updates

Medical Nutrition Equity Act Update – Senate Companion Bill Introduced

Despite the ability to manage certain inherited metabolic disorders through medical foods, many patients struggle to get insurance to cover this critical nutrition. This past March, ACMG asked members to help ensure patient access to medical foods by sending letters to members of the US House of Representatives asking them to co-sponsor House bill HR 2501, the Medical Nutrition Equity Act of 2019. This bill would ensure patient access by requiring public and private health insurers to provide coverage for such medically necessary nutrition. Since our notification in March, 350 individuals have sent letters reaching 160 different representatives. Now, we need your help asking members of the US Senate to co-sponsor the Senate companion bill, S 3657, which was introduced in May 2020.

Click here to ask your US senators to co-sponsor S 3657, the Medical Nutrition Equity Act of 2020.

To make it very easy, ACMG has provided a template letter that can be sent electronically directly to your senators. All you have to do is fill in your name and address, then you have the option to send the letter as is or modify it to reflect your own experiences. When you hit send, the letter will automatically be sent to both of your US senators. You can also share this link with your colleagues. The letters are not linked to ACMG, and the letter can be modified to reflect other professionals' specialties.

Also, if you have not sent a letter to your House representative, please consider doing so now by clicking here (note that the links are different for the House and Senate letters).

Medical nutrition equity advocates have been working with Congress to identify a path forward for this bill, but this also requires that both the versions in the House and Senate have a strong list of congressional co-sponsors. Letters from constituents like you are often needed to convince senators and representatives to become co-sponsors. Your letters make a difference!

ACMG Economics Committee Presents at CMS CLFS Annual Public Meeting

On June 22, 2020, the Centers for Medicare and Medicaid Services (CMS) held their virtual Clinical Laboratory Fee Schedule (CLFS)

Annual Public Meeting. During this meeting, stakeholders were given the opportunity to provide recommendations on establishing payment amounts for new codes to be placed on the CLFS in CY 2021 and reconsidered codes that were new for CY 2020. The ACMG Economics of Genetic Services Committee presented recommendations on new germline test codes, including three *TP53* codes and a new code for an epilepsy gene panel. They also presented recommendations for the *PALB2* full gene sequence code from CY 2020 that ACMG had previously requested be reconsidered (see letter here). Copies of all stakeholders' presentations are available here.

So what happens now? On July 29-30, 2020, the Medicare Advisory Panel on Clinical Diagnostic Laboratory Tests (CDLTs) will meet virtually to review the stakeholder comments and develop their formal recommendations to CMS. Following review of both the stakeholder and advisory panel recommendations, CMS is expected to release their proposed determinations by early September. CMS is under no obligation to follow the recommendations from stakeholders or the advisory committee, and it is not uncommon for CMS to reject both and develop their own recommendations. Stakeholders will have the opportunity to provide written comments in response to CMS's proposed determinations, a process that the ACMG Economics Committee will participate in if warranted. The final determinations will be posted on CMS's website in November, at which point stakeholders have an additional opportunity to submit written feedback. If we strongly disagree with CMS's final determination for any new codes, we will request that those specific codes be reconsidered in 2021.

Special thanks to M. Laura Cremona, PhD, FACMG, ACMG Economics Committee vice chair, for presenting at the CLFS Annual Public Meeting on behalf of ACMG and the ACMG Economics of Genetic Services Committee.

Palmetto GBA to Offer Limited Coverage of Pharmacogenomic Tests

Medicare Administrative Contractor (MAC) Palmetto GBA recently finalized a MolDx local coverage determination (LCD) providing broad coverage for certain pharmacogenomic (PGx) tests, including single-gene tests and multi-gene panels. The LCD states that PGx tests are indicated when medications are being considered for use (or already being administered) that are medically necessary, appropriate, and approved for use in the patient's condition, and are known to have a gene(s)-drug interaction that has been demonstrated to be clinically actionable as defined by the Food and Drug Administration (FDA) or Clinical Pharmacogenetic Implementation Consortium (CPIC) guidelines (category A and B). Additional details about the PGx test coverage criteria are explained in the final LCD (L38294) available on the CMS website. The LCD does not address PGx testing for anticoagulation dosing as that is already addressed by the National Coverage Determination (NCD) 90.1.

Palmetto GBA's new policy will apply to services performed on or after July 26, 2020. While Palmetto GBA only covers Medicare beneficiaries in certain states in the southeast, this LCD was established as part of

Palmetto's MolDx program. It is anticipated that the other MACs that have implemented the MolDx program will finalize the same LCD for their jurisdiction(s) in the coming months resulting in coverage for a significant portion of Medicare beneficiaries in the US. One MAC, Noridian Healthcare Solutions, already finalized this same LCD (L38335 and L38337) for its two jurisdictions, which will be effective for services provided on or after August 17, 2020.

Understanding the Details: ACMG's Position on HR 3235

The following article is reproduced from the Spring/Summer 2020 issue of The ACMG Medical Geneticist *newsmagazine.*

Earlier this year, some people were surprised to learn that ACMG was requesting that additional language be added to HR 3235, the Access to Genetic Counselor Services Act, before we could fully support the bill. As the spread of misinformation has been an ongoing issue, we wanted you to hear directly from ACMG so you have accurate and current information. **ACMG resolutely supports reimbursement of genetic counselors for genetic counseling services but not for independent practice of medicine. Our proposed amendments to HR 3235 would not impede the direct reimbursement of genetic counselors, nor would it impede their ability to order genetic testing**. It would, however, encourage continuation of the collaborative environments that most currently work in and that benefit patients. We continue to strive for an agreement that we can fully support and to see this bill come to fruition with just the addition of brief language about a collaborative relationship. We know that we work best for patients and public health when we work together (not independently), and that is our goal.

For more detailed information, please read below:

• At the core of the issue is whether genetic counselors should engage in the unrestricted, independent ordering of genetic tests as well as other diagnostic studies, which the ACMG views as the practice of medicine.

- HR 3235, by providing for reimbursement, is inextricably linked to state legislation that expands scope of practice, and the detailed language of state and federal rules must be considered collectively. State licensure legislation currently in place or being pursued in numerous states expands genetic counselors' scope of practice to include independent ordering of genetic tests and other diagnostic studies (not just genetic) as well as interpretation and return of results to patients without any interaction with physicians. The ACMG believes that this is not the best model for patient care and that it is the practice of medicine.

- We recognize that some genetic tests, such as carrier screening tests to guide reproductive decision-making, generally do not need physician involvement. However, tests that confer a diagnosis or guide treatment decisions require physician involvement. Some tests may require a medical evaluation or other types of laboratory testing to identify genetic

testing needs. In certain cases, genetic testing should only be ordered after a patient has received a diagnosis from a physician. Others may require physician involvement to complete a diagnostic interpretation of results. While we believe that the majority of genetic counselors will continue to responsibly order only certain types of tests when appropriate criteria are met, precautions need to be added to legislation to ensure that optimal patient care is always provided in a healthcare team environment.

- For these reasons, ACMG is proposing a requirement for collaborative agreements (NOT supervision) between counselors and physicians. Each collaborative agreement could be tailored to allow genetic counselors and physicians to establish procedures and protocols that best fit their own institutions.

• ACMG supports reimbursement of genetic counselors for genetic counseling services, and our proposed amendments to HR 3235 would not impede their reimbursement.

- ACMG is unable to fully support HR 3235 in its current form because it would provide reimbursement for services that go beyond genetic counseling and constitute the independent practice of medicine. ACMG is not the only professional organization concerned about the current language. ACMG recognizes that reimbursement of genetic counselors for their services is **very** important. Therefore, rather than directly oppose HR 3235, we are working with Congress and other organizations to develop language that can be broadly supported and will not impede genetic counselors' ability to be directly reimbursed by Medicare.

- ACMG has offered a minor amendment to HR 3235 (shown underlined in bold just below), which would recognize the teamwork necessary to provide the best care to patients:

(kkk) (1) The term 'covered genetic counseling services' means genetic counseling services furnished by a genetic counselor (as defined in paragraph (2)) ***pursuant to a formally-documented collaborative agreement with a physician (as defined in subsection (r)(1))*** *(and such services and supplies furnished as an incident to the provision of such services) as would otherwise be covered under this title if furnished by a physician (or as incident to a physician's service),* ***but does not include the diagnosis or treatment of patients.***

• Rather than opposing HR 3235, ACMG has proposed a minor modification to its language focused on collaborative agreements (NOT supervision) to encourage continuation of team-based models for patient care.

- Collaborative agreements would not require supervision and would allow an enormous amount of flexibility. Such agreements would create an opportunity for healthcare professionals to ensure that protocols and procedures are in place to guide how patients will be managed in various situations. Medical geneticists and genetic counselors could work together to develop model collaborative agreements for their various practice scenarios.

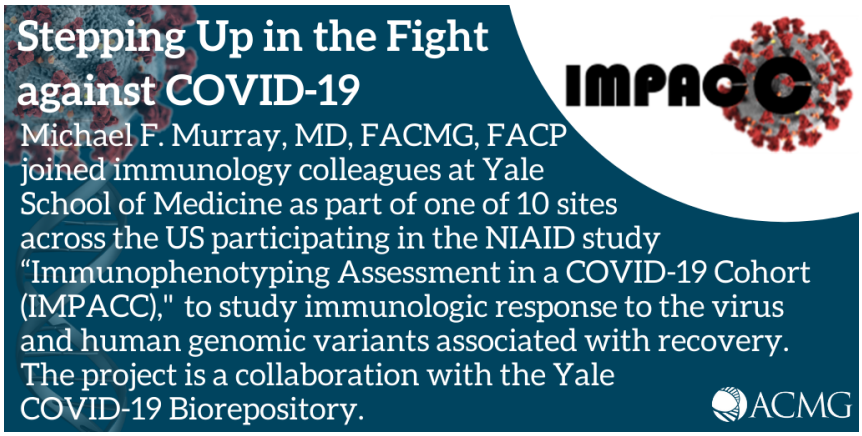
- While we understand the desire of some genetic counselors for independent practice, ACMG maintains that the most effective practice models are those in which genetic counselors work collaboratively in a team environment with physicians and laboratory geneticists. Bidirectional communication between genetic counselors and physicians helps both counselors and physicians, and in turn leads to optimal patient care. The majority of genetic counselors currently work in such collaborative environments and may be unaware of how such bills, without minor language modifications, could change this model practice.

Note: the above information was accurate to the best of our knowledge at the time the Spring/Summer 2020 issue of *The ACMG Medical Geneticist* went to press in May 2020.

ACMG Members Stepping Up in the Fight against COVID-19

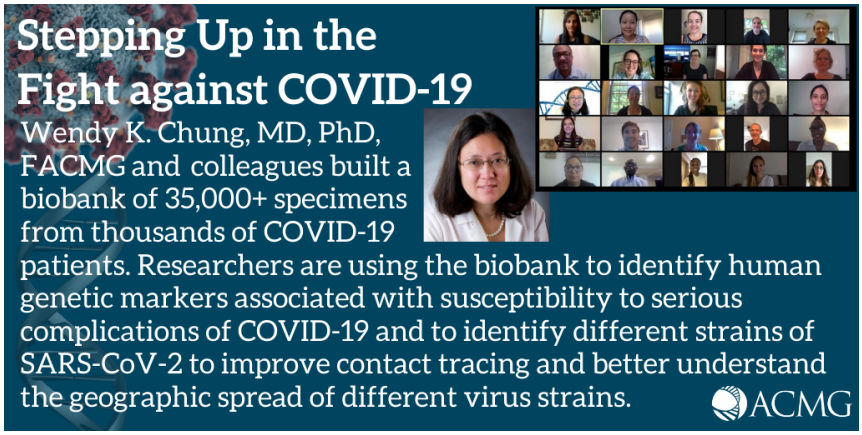
Over these last few months, ACMG has heard inspiring stories from members about how they have been adapting and innovating throughout the SARS-CoV-2 pandemic to provide essential services to patients (including telehealth services), conduct important research and COVID lab testing, and help and support colleagues. To thank our members for all they are doing, we have been sharing these stories on social media. We hope you will enjoy the most recent of these stories, which are reproduced here.

If you have, or know of, an inspiring story about ACMG members stepping up in the fight against COVID-19, please email Reymar Santos (rsantos@acmg.net) and tell us what you and your colleagues are doing so we can acknowledge you!



Michael F. Murray, MD, FACMG, FACP, professor of genetics and director for clinical operations in the Center for Genomic Health at Yale School of Medicine, joined with his immunology colleagues at Yale as part of one of 10 sites across the US participating in the National Institute of Allergy and Infectious Diseases (NIAID) study "Immunophenotyping Assessment in a COVID-19 Cohort (IMPACC)," as part of an in-depth investigation of immune responses to the SARS-CoV-2 virus. One

thousand study participants will be followed for a year post-hospitalization to gauge recovery and study both the immunologic response to the virus and the human genomic variants that are associated with recovery. The IMPACC project involves expertise across the Yale School of Medicine and is a collaboration with the Yale COVID-19 Biorepository.



Wendy K. Chung, MD, PhD, FACMG—Kennedy Family Professor of Pediatrics, chief of the Division of Clinical Genetics and medical director of the Columbia Genetic Counseling Graduate Program at Columbia University Irving Medical Center and NewYork-Presbyterian Hospital, and associate director for education at Columbia University Herbert Irving Comprehensive Cancer Center—worked with colleagues to build a biobank of more than 35,000 specimens from thousands of COVID-19 patients. Starting with nothing more than an empty lab, biobank coordinators and a team of genetic counselors quickly designed and implemented a remote consent system that enrolled 96% of patients and families contacted by utilizing recycled specimens from clinical tests, eliminating the need for patients to provide additional samples. In just 10 weeks, the team processed more than 20,000 serum, 11,000 plasma, 11,000 blood mononuclear cells, 3,000 nasopharyngeal, 500 cord blood, and 300 fecal samples. Researchers are using the biobank to identify human genetic markers associated with susceptibility to serious complications of COVID-19—including severe multisystem inflammatory syndrome in children (MIS-C)—and to identify different strains of SARS-CoV-2 to improve the accuracy of contact tracing and better understand the geographic spread of virus strains.

NCC Updates “Find a Genetic Service” Directory

Did you know that the National Coordinating Center for the Regional Genetics Networks (NCC), housed within ACMG, recently updated the “Find a Genetic Service” directory? On the directory web page, you can search for genetics clinics across the country, view clinics near you on a map, learn about specific services offered by a clinic, and much

more. Be sure to check out the directory today and submit any updates to your clinic's listing via the online form. If you have any questions about the directory, please contact NCC Associate Program Director Alisha Keehn (akeehn@acmg.net) and NCC Project Manager Megan Lyon (mlyon@acmg.net).

July 2020 GenePod: COVID-19 Presents Challenges for Care of Patients in Genetics and Metabolic Disease Clinics

The COVID-19 pandemic is unprecedented, and clinicians have faced correspondingly unprecedented challenges in caring for patients in genetics and metabolic disease clinics who require routine check-ups and ongoing treatments. Yet, in overcoming these challenges, doctors have identified a few boons to patient care that may continue to benefit patients in the long run. Listen in to this month's GenePod, *Genetics in Medicine's* monthly podcast, to hear how Dr. Nicola Brunetti-Pierri, a clinician at the Federico II University Hospital in Naples, Italy, and Dr. Elaine Pereira, a clinical geneticist at Columbia University in New York City, two of the areas hardest hit by the pandemic, adapted patient care amid government lockdowns and what silver linings they found.

ACMG Welcomes 19 New Members

ACMG welcomes and congratulates 19 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 Members
Areej Alhareeri, PhD, MS, FACMG
Angie C. Jelin, MD, FACMG
Shibani Kanungo, MD, MPH, FACMG
Veronica Mardo, MD, FACMG
Yaning Wu, PhD, FACMG

Associate Members
Priyanka R. Ahimaz, MGC, CGC
Colleen Dougherty, MS, CGC
Jessica L. Giordano, MS, CGC
Emily Qian, MS, CGC
Alanna Rahm, PhD, MS, CGC

Candidate Fellow
Ester Perales Clemente, DSci

Affiliate Members
Paul S. Appelbaum, MD

Carla L. Easter, PhD
Mohammad A. Hossain, MBBS, PhD
Berit Kerner, MD

Trainee Members
Andrew C. Edmondson, MD, PhD
Ria Garg, MBBS
Daniel J. Pomerantz, MBBS

Student Member
Savannah Jo Brennan

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

NBSTRN Joins with Partners to Host Quarterly Webinars to Facilitate SCID Newborn Screening

Severe Combined Immunodeficiency (SCID) is a devastating genetic condition and is uniformly fatal in the first years of life unless the immune system can be restored. Newborn screening saves the lives of newborns with SCID by alerting the family and healthcare providers of a suspected immune dysfunction before the onset of overwhelming infections. The development of a novel screening test led to recommended nationwide screening for SCID in 2010. The SCID newborn screening test is based on detection of T-cell receptor excision circles (TRECs)—DNA biomarkers of normal T lymphopoiesis that can be measured by a polymerase chain reaction test. Today, all states, territories and the District of Columbia screen for SCID and approximately 76 (1 in 50,000) newborns are ultimately diagnosed with SCID, and another 200 are diagnosed with non-SCID T-cell lymphopenia (TCL), each year.

To facilitate the implementation of SCID newborn screening, the Newborn Screening Translational Research Network (NBSTRN), housed at ACMG, has partnered with the Association of Public Health Laboratories' (APHL) Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) and the Immune Deficiency Foundation's SCID Compass Program to host a quarterly webinar for state programs, clinicians, parents, patients and advocates. In June of this year, Dr. Amy Brower, co-principal investigator of the NBSTRN, and Alissa Huston, from the SCID Compass Program, presented updates on recent efforts. Ms. Huston talked about resources for parents and providers found on their website, and announced a monthly virtual support group for parents. Dr. Brower discussed an effort by the Clinical Laboratory Standards Institute (CLSI) to update their guidance document on SCID newborn screening. The guidance document is designed to assist laboratories with TREC testing.

The Newborn Screening Translational Network (NBSTRN) is a network of clinicians, researchers, parents and advocates that supports the development of newborn screening research. To learn more about

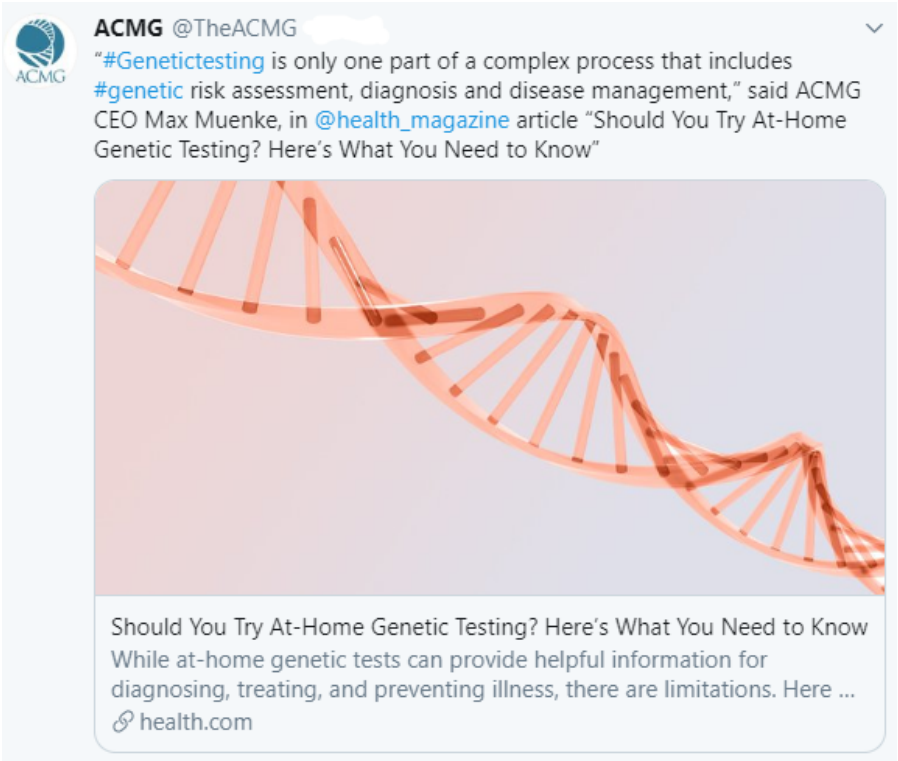
NBSTRN efforts and to become a member of the NBSTRN, visit the NBSTRN website and follow @NBSTRN on Facebook, Instagram, Twitter and LinkedIn.

ACMG Social Media Highlights

Do you follow ACMG on social media? ACMG actively participates on Twitter, Facebook, Instagram, YouTube, and LinkedIn. The College also hosts an ACMG Annual Clinical Genetics Meeting group on LinkedIn and a private, members-only group on LinkedIn.

Through these pages, the College earns nearly 125,000 impressions each month, providing followers with updates and news about educational programs and other activities of the College and the ACMG Foundation. The ACMG members-only group on LinkedIn is an important added benefit of membership in the ACMG. This exclusive group page provides news and opportunities directly to members, including invitations to comment on draft ACMG statements and guidelines, calls for participation in College and Foundation activities and programs, and advance notification of the publication of new ACMG statements and guidelines.

We hope you will follow these pages on social media and join the conversation. Please enjoy a sampling of a few of our recent social media posts.



Mark Your Calendar for the Final Live Webinar of the 2020 ACMG Annual Clinical Genetics Meeting – Digital Edition

The final live webinar of the 2020 ACMG Annual Clinical Genetics Meeting – Digital Edition Webinar Series is set for **Monday, July 20, 2020, from 3:30–5:00 PM EDT**. Join moderator Heather B. Radtke, MS and presenters Bruce R. Korf, MD, PhD, FACMG; David Viskochil, MD, PhD, FACMG; Ashley Cannon, PhD, MS; David T. Miller, MD, PhD, FACMG; and Scott R. Plotkin, MD, PhD for "Neurofibromatoses: New Frontiers in NF1, NF2, and Schwannomatosis."

To learn more about this webinar and to register, click here.

To learn about past webinars in this series, click here.

ACMG Education & CME Committee Launches Student Challenge

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 July 2020 and continuing through June 2021, we will release a new student challenge question. These questions are great opportunities to learn more about clinical genetics and gain knowledge 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.

Have You Logged In to the 2020 ACMG Annual Clinical Genetics Meeting – Digital Edition?

Check out what 1,800 of your colleagues have been doing since the May launch of the 2020 ACMG Annual Clinical Genetics Meeting – Digital Edition. View the Poster Gallery and Exhibits for free.

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

Access the outstanding educational content, posters and abstracts that were to be presented at the 2020 ACMG Annual Meeting. View the important research and cutting-edge developments shaping genetics and genomics and earn more than 50 hours of continuing education credit.

If you have not yet logged in or purchased access to the 2020 ACMG Annual Meeting – Digital Edition, don't miss the opportunity to participate in this blended learning activity. The Digital Edition features the following:

- **Abstracts and Poster Gallery**: Features abstracts and posters accepted for the 2020 ACMG Annual Meeting.

- **Webinar Series**: A series of webinars offered live and archived in the Digital Edition after the live presentations.

- **Recorded Scientific Sessions**: Recordings of sessions that were to be presented at the 2020 ACMG Annual Meeting.

- **Exhibit Theaters and Satellite Symposia**: Recordings of selected Exhibit Theaters and Satellite Symposia that were to be presented at the 2020 ACMG Annual Meeting.

- **Exhibit Hall**: View exhibitor eBooths showcasing state-of-the-art technologies, products, services and resources tailored to the needs of genetics professionals.

In addition to being offered as a complete package, there are several bundled opportunities to take advantage of such as Webinar and Short Course bundles. Visit the Digital Edition website for more information.

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