ACMG In Action

Essential Updates: Member News You Can Use

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

July 2021

In This Issue...

Sneak Preview – Spring/Summer ACMG Member Magazine is Now Available Online

The spring/summer edition of *The ACMG Medical Geneticist* member magazine is a record-breaker at 40 pages! Watch for your copy in the mail or you can read it online now. Highlights include an in-depth Q&A with ACMG's new president Dr. Marc S. Williams, FACMG on priorities, opportunities and challenges; the 2021 ACMG Foundation Award recipients; welcome to new board members; a recap of both the 2021 Medical Genetics Awareness Week and the fantastic 2021 ACMG Annual Clinical Genetics Meeting – *a virtual experience*; participants of the 2021 ACMG Foundation Summer Genetics Scholars Program; ACMG's #IGottheShot PR campaign and so much more.

ACMG Releases First Evidence-based Clinical Guideline: Exome and Genome Sequencing for Pediatric Patients with Congenital Anomalies or Intellectual Disability

In case you missed the exciting news, ACMG has released its first Evidence-based Clinical Practice Guideline (EBG), "Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the ACMG." The EBG was published in ACMG's official journal, *Genetics in Medicine*.

"The publication of this guideline is a landmark achievement for the ACMG," said ACMG President Marc S. Williams, MD, FACMG in ACMG's news release. "It is the culmination of years of foundational work and a commitment to implementing the best practices for the development of evidence-based guidelines."

"We are excited to announce the publication of the first ACMG-endorsed evidence-based guideline on the use of exome and genome sequencing for evaluation of pediatric intellectual disability or multiple congenital anomalies," said Fuki Marie Hisama, MD, FACMG, FAAN and Murugu Manickam, MD, FACMG, who co-chaired the ACMG EBG working group. "Our committee included 10 experts in clinical genetics, neurogenetics, genetic counseling, a parent and advocate and methodologists. We expect this EBG will help to raise the quality and

- Get Your Copy of the 2021 ACMG Digital Edition – It's Not Too Late!
- 2022 Annual Clinical Genetics Meeting - Important Dates

Upcoming Events

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

consistency of health care and improve outcomes for patients with rare genetic disorders."

The paper concludes that based on the Systematic Evidence Review there is a strong recommendation to support the use of Exome/Genome Sequencing (ES/GS) as either a first- or second-line test in patients with CA/DD/ID.

The ACMG EBG has already garnered worldwide attention with close to 400,000 impressions on Twitter alone, significant media coverage, and is currently ranked in the 99th percentile (ranked 1,720th) of the 192,431 tracked articles of a similar age in all journals according to Altmetric.

ACMG Publishes Revised Technical Standard for Chromosomal Microarray Analysis

ACMG is pleased to announce the publication of an update to a pair of technical standards, "Chromosomal Microarray Analysis, Including Constitutional and Neoplastic Disease Applications, 2021 Revision: A Technical Standard of the American College of Medical Genetics and Genomics (ACMG)". The revision of these technical standards was undertaken by the Laboratory Quality Assurance Committee's Cytogenetics Subcommittee; it serves as a combined update of the two ACMG technical standards and guidelines for chromosomal microarray analysis that were published in 2013 (South, et al, *Genet Med*.15(11):901-9 [pre- and postnatal constitutional applications]; Cooley, et al, *Genet Med*.15(6):484-94 [neoplastic disorder applications]).

Chromosomal microarray (CMA) technologies have broad applications in the evaluation of constitutional and neoplastic disorders. The ACMG recommends CMA as the first-tier test for the evaluation of chromosomal imbalances associated with intellectual disability, autism, and/or multiple congenital anomalies. Furthermore, CMA analysis is recommended by the American College of Obstetricians and Gynecologists and the Society for Maternal-Fetal Medicine as the first-tier prenatal test for patients undergoing invasive prenatal diagnosis with one or more major fetal structural abnormalities identified by ultrasonographic examination, and for the evaluation of intrauterine fetal demise or stillbirth when further cytogenetic analysis is desired. More recently, CMA is being used as a follow-up test for small copy number changes that are reported by noninvasive prenatal screening. In the cancer genetics arena, CMA provides genomic data important in elucidating the diagnosis, prognosis, and therapy for neoplastic disorders, including both hematologic malignancies and solid tumors.

READ MORE

Now Available: Revised Technical Standard for Inherited Colorectal Cancer and Polyposis Testing

ACMG is pleased to announce the publication of an update to its 2013 technical standard, "Genetic Testing for Inherited Colorectal Cancer and

Polyposis, 2021 Revision: A Technical Standard of the American College of Medical Genetics and Genomics (ACMG)". The revision of this technical standard was undertaken by the Laboratory Quality Assurance Committee's Molecular Genetics Subcommittee and supersedes the original laboratory standard on this topic (Hegde M, et al, *Genet Med*.16(1):101-16).

Colorectal cancer (CRC) is the fourth most frequently diagnosed cancer and the second leading cause of cancer death in the United States. Thirty percent of all cases are thought to have a hereditary component, and up to one-third of these (10%) are hereditary. These latter cases are associated with pathogenic germline variants in multiple genes. With the adoption of massively parallel sequencing (also known as next-generation sequencing, NGS) by many clinical laboratories, it is now possible for timely, cost-effective analysis of multiple genes associated with inherited polyposis and/or CRC. Given the overlapping phenotypes of variable penetrance cancer syndromes along with the limited sensitivity of using clinical criteria alone, a multi-gene panel testing approach for the diagnosis of these conditions is effective and efficient.

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July GenePod: Artificial Intelligence May Provide a Timely Diagnosis for Fragile X Syndrome

While Fragile X syndrome is the most common cause of inherited intellectual disability, it is still underdiagnosed in the general population. As the phenotype may be subtle, the diagnostic pathway can take years. In addition to this, the syndrome is accompanied by many secondary health conditions — the full spectrum of which are not entirely understood by medical practitioners — adding to the burden of care for patients and families.

On this month's GenePod, Arezoo Movaghar, PhD, a post-doc in the Waisman Center at the University of Wisconsin-Madison, and Marsha Mailick, PhD, emeritus vice chancellor for research and graduate education at the University of Wisconsin-Madison, discuss the use of artificial intelligence to both identify the prevalence and severity of secondary medical conditions and to accurately diagnose patients years in advance of a typical clinical diagnosis.

Have you Read ACMG's Statement About Direct-to-Consumer Prenatal Testing?

ACMG recently announced the publication of "Direct-to-consumer prenatal testing for multigenic or polygenic disorders: a position statement of the American College of Medical Genetics and Genomics (ACMG)" in our official journal, *Genetics in Medicine*. Issues surrounding direct-to-consumer testing for monogenic disease or disease caused by copy-number variants are complex for disorders that exhibit multigenic or

polygenic inheritance. These disorders have been shown to be controlled in part by multiple genetic loci. Examples include psychiatric disease (schizophrenia), cardiovascular disease, complex heart disease, diabetes mellitus (type 2), and Alzheimer disease.

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Legislative Updates

Recently, there has been a considerable amount of federal legislative activity of importance to medical geneticists and their patients. Here are some of the updates:

Newborn Screening Saves Lives Reauthorization Act Passes the House!

Last month, the Newborn Screening Saves Lives Reauthorization Act (HR 482) passed in the House with broad bipartisan support. Now the legislation must pass the Senate where it has previously encountered challenges. While the bill has always had very strong bipartisan support, it was held up in the last Congress due to a senator's desire to add concerning language about consent requirements for federally funded research using deidentified residual dried blood spots. The language would override existing protections in the Common Rule and create additional barriers for research on deidentified residual dried blood spots.

Reducing Hereditary Cancer Act Introduced in House

The Reducing Hereditary Cancer Act (HR 4110) would allow Medicare to cover germline testing, screenings, and preventive surgeries recommended by National Comprehensive Cancer Network guidelines. ACMG applauds the sponsors for introducing this new legislation and is supportive of its passage.

Medical Nutrition Equity Act Reintroduced

The Medical Nutrition Equity Act (MNEA) was recently introduced in the House (HR 3783) and Senate (S 2013). The bill would require public and private payers to cover medically necessary foods for patients with inherited metabolic conditions and certain other conditions of malabsorption. ACMG continues to support this legislation and work with members of Congress to encourage its passage. For more information, see the Patients and Providers of Medical Nutrition sign-on letter, here.

VALID and VITAL Acts Reintroduced

The Verifying Accurate, Lead-edge IVCT Development (VALID) Act has been introduced in the House (HR 4128) and Senate (S 2209). Similar to the previous version, the legislation would create a new regulatory pathway at the Food and Drug Administration (FDA) for in vitro clinical tests (IVCTs). The IVCT definition includes both laboratory-developed tests (LDTs) and tests that are manufactured and distributed, both of which would be subject to the same premarket review requirements under the VALID Act. The Verified Innovative Testing in American Laboratories (VITAL) Act has also been introduced in the Senate (S 1666). This legislation would clarify that all LDTs, referred to as laboratory-developed testing procedures, would be regulated solely under the Clinical Laboratory Improvement Amendments (CLIA) and that no aspect would be regulated by FDA. The legislation would also require the Centers for Medicare & Medicaid Services (CMS), the federal agency responsible for enforcing CLIA, to hold a public meeting to solicit recommendations on updating the CLIA regulations. ACMG has a long history of calling for CLIA modernization and continues to support regulation of LDTs through CLIA.

Improving Seniors' Timely Access to Care Act Reintroduced

The Improving Seniors' Timely Access to Care Act was recently introduced in the House (HR 3173). The bill would require Medicare Advantage (MA) plans to implement a streamlined electronic prior authorization process; require increased transparency for beneficiaries and providers; enhance CMS oversight on processes used for prior authorization; provide for real-time decisions by an MA plan for certain prior authorization requests; and require MA plans to meet beneficiary protection standards, such as ensuring continuity of care when patients change plans. The legislation already has significant bipartisan support in the House and is well supported by medical associations, including ACMG.

Supreme Court Rejects ACA Challenge

The US Supreme Court recently rejected a challenge to the Affordable Care Act (ACA), thereby upholding the ACA once again. When the ACA was initially passed it included a financial penalty associated with the ACA's individual mandate, but that financial penalty was eliminated by another law passed in 2017. The plaintiffs in the lawsuit argued that removal of the penalty made individual mandate unconstitutional and that the mandate could not be severed from the law thereby making the entire ACA unconstitutional. In May 2020, ACMG joined an amicus brief led by the American Medical Association (AMA) that urged the Court to reject the challenge. In a 7-2 decision, the Court decided that the challengers did not have a legal right to sue because they could not prove that harm had occurred and thus rejected the case.

ACMG Welcomes 19 New Members

ACMG welcomes and congratulates its 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

Toni R. Prezant, PhD, FACMG

Candidate Fellow Members
Jennifer M. Gass, PhD
Meng Liu, MD, PhD, MBA
Gang Zhou, PhD

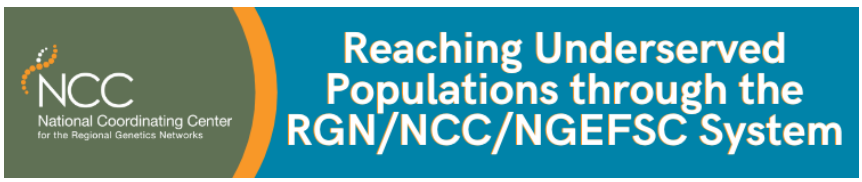
Affiliate Members
Sanjay Gupte, MD
Hyunjoo Lee
Mary C. Maj, PhD
Carrie A. Thompson, MSN

Trainee Members
Angella Charnot-Katsikas, MD
Brendan J. Floyd, MD, PhD
Bin Guan, PhD
Erica Lay, MD
Kelly A. Rafferty, PhD, MS
Laura Thompson, DSci

Student Members
Veronica A. Abraham, MD, MPH
Jennifer D. Cotter, MS
Sharanya Jayachandran, BS
Pinar Ozmizrak, MS
Samantha Ratner

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

Reaching Underserved Populations: Update from NCC



The mission of the National Coordinating Center for the Regional Genetics Networks (NCC), a cooperative agreement between the Health Resources and Services Administration (HRSA) and ACMG, is to improve access to genetic services for underserved populations. NCC works with the seven Regional Genetics Networks (RGNs) and the

National Genetics Education and Family Support Center (Family Center) to accomplish this mission.

In order to evaluate how well we are achieving our mission of reaching underserved populations, NCC collects data from each RGN and reports the data from the RGN/NCC/NGEFSC system. Access our newly updated report on NCC's website to learn more about how many providers and individuals/families come from underserved areas our system reaches through education/training, telemedicine assistance, and/or by facilitating connection between individuals and providers.

If you have any questions about the mission of the RGN/NCC/NGEFSC system or who our system assists, please contact Megan Lyon, NCC Senior Program Manager.

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #UH9MC30770 from 6/2020-5/2024 for $800,000 per award year.

This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

NBSTRN Publication Describes Tool to Advance Rare Genetic Disease Research

The goal of newborn screening (NBS) is to improve health outcomes by identifying and treating newborns with rare genetic diseases. The long-term follow-up of newborns diagnosed with a condition through newborn screening is important to ensure that we achieve the best possible health outcomes for these infants. The collection of longitudinal health information on affected newborns helps to document the benefit of early identification and treatment through NBS. Long-term data collection also helps to advance understanding of the diseases that are part of, or candidates for, newborn screening. To facilitate the collection, analysis, visualization, and sharing of longitudinal data, the NBSTRN team developed the Longitudinal Pediatric Data Resource (LPDR). The LPDR is described in a special issue of the *International Journal of Neonatal Screening* that is now available.

The ability to capture clinical information early in the clinical course of a disease, even before clinical symptoms appear, advances disease understanding, helps to establish the efficacy of new treatments and management approaches, informs the community at large about the value of early identification and treatment through newborn screening, and identifies areas for improvement in disease management throughout the

lifespan. From coast to coast over 100 researchers and NBS state programs have used the LPDR in over thirty basic, translational, public health, and clinical research projects. The LPDR is designed to share these team's new findings and to foster the secondary use of the original data sets. To learn more about the LPDR please visit NBSTRN Tools webpage.

ACMG Member Comment: Laboratory Standard for Lysosomal Enzyme Analysis

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

Measurement of Lysosomal Enzyme Activities: A Technical Standard of the American College of Medical Genetics and Genomics (ACMG).

To access the document, go to the ACMG Website and log on using your username and password. From the homepage, click on the Membership header (second from the right) and scroll down to Members Only (item four in the menu). Once in the Members Only section scroll down to the Benefits header, and on the left will be a column of gold tabs. Click on the seventh gold tab down, "Documents for Comment". By selecting this tab, you will find a list of all College documents open for member comment. Click on the document title to download the relevant document to your computer.

Upon downloading, a single Word file will appear.

Please note that all ACMG documents for member comment carry a strict embargo and are not to be shared with non-ACMG member colleagues.

READ MORE

Spread the Word: ACMG Launches Careers in Medical Genetics Series

Did you know that ACMG recently launched an updated "Careers in Medical Genetics and Genomics" video series on our ACMG YouTube Channel? Please share these videos with your network as we all work together to attract students to all areas of medical genetics and genomics.

These videos provide undergraduates, graduate, and medical students the opportunity to learn more about the various career paths in Medical Genetics and Genomics. Career perspectives are provided by a clinical/research academic geneticist, a clinical laboratory-based geneticist, a genetic counselor, a recent medical genetics resident graduate, and a representative from the American Board of Medical Genetics and Genomics who furnishes details on medical genetics training programs and certification.

- A Day in the Life of a Clinical Geneticist
  - Natario Couser, MD, MS, FACMG
- Clinical Laboratory-based Career Paths
  - Ruben Bonilla Guerrero, MD, FACMG
- A Career Path in Genetic Counseling
  - Andrea Schelhaas, MS, CGC
- Tips and Perspectives from a Recent Training Program Graduate
  - Cinthya Zepeda Mendoza, PhD
- All About the ABMGG
  - Miriam Blitzer, PhD, FACMG

Now Available in the ACMG Genetics Academy

ABMGG Longitudinal Assessment Program CertLink

ABMGG CertLink meets the ACCME requirements for continuing medical education with assessment. Through our partnership with ABMGG, ACMG will award 10 AMA PRA Category 1 Credits™ each year (5 credits/period) to diplomates participating in ABMGG CertLink.

For more information visit: CertLink Pilot Program | ABMGG

ClinGen Somatic Cancer and VICC Virtual Molecular Tumor Board Case Series
July 27 (4th Tuesday), 11:00 am – 12:00 pm ET

This series is a collaboration between ClinGen Somatic and VICC consortia and the ACMG and is appropriate for all medical and healthcare professionals and researchers interested in understanding cancer genomic testing and somatic and germline variant interpretation methods. Registration Coming Soon!

Printed Syllabus from the 2021 ACMG Genetics and Genomics Review Course

The 2021 Printed Syllabus from the highly rated ACMG Genetics and Genomics Review Course is the perfect add-on to help prepare yourself for the board exams or to refresh your knowledge. Printed syllabus is approximately 900+ pages, printed in black & white and spiral bound.

• $280 ACMG Members | $300 Non-Members

More information and purchase here.

2021 ACMG Foundational Specialty Training in Cytogenetics and Molecular Genetics and Genomics (CME, P.A.C.E.®)

This course meets one criterion of the ABMGG requirements for training in the LGG Alternative Pathway and includes 19 Recorded Presentations, 4 Live Q&A Sessions from July 19 – 22, 4:00 pm – 5:30 pm ET. More information here.

2021 - 2022 ACMG Student Challenge-Now Available!

ACMG invites all medical genetic students, residents and genetic counseling trainees to participate in our 2021-2022 Student Challenge. Each month, we will release a student challenge question, starting July 1, 2021 thru June 30, 2022. These questions are great opportunities to 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 (New Module):** 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.

The **2021 ACMG Digital Edition** is now available for purchase in the ACMG Genetics Academy.

Content will be available on July 19, 2021. If you purchased the extended access when registering for the April virtual meeting, you will automatically be given access, and you will receive an email when the content is ready to view. If you have not already purchased the Digital Edition, you may do so in the ACMG Genetics Academy. Cost to purchase is $349 for members; $399 for nonmembers.

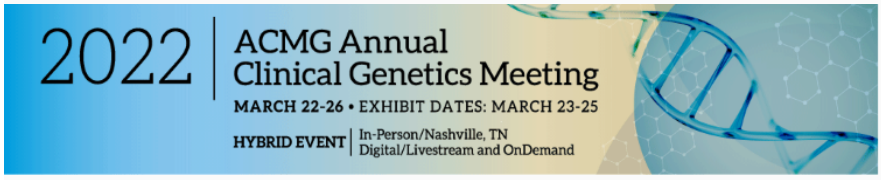
The **2021 Digital Edition** offers the convenience of learning on your own time from the comfort of your home or office. It's easier than ever before to watch – and re-watch – your favorite sessions from the 2021 virtual meeting.

Registration includes access to videos of course presentations, slides, and **the ability to claim CME, General CEU's, and P.A.C.E.® credits through July 19, 2023**.

This activity has been approved for *AMA PRA* Category 1 Credit™.

This event has been submitted to the National Society of Genetic Counselors (NSGC) for approval of Category 1 CEUs. The American Board of Genetic Counseling (ABGC) accepts CEUs approved by NSGC for purposes of recertification. Approval for the requested CEUs and Contact Hours is currently pending.

2022 Annual Clinical Genetics Meeting - Important Dates



Save the Dates – Attend in Nashville or Participate Online

The 2022 ACMG Annual Clinical Genetics Meeting will be delivered in hybrid format, offering the opportunity to gather in Nashville for those who are ready to travel again. Attendees who wish to participate remotely can join in-person attendees for the livestreamed content, connect with peers through the platform and view recorded sessions on demand.

Detailed information, registration, housing and abstract submission will be available at www.acmgmeeting.net in October.

2022 Proposal Submission Now Open – Deadline for Submission is July 30, 2021

Plan now to submit a session proposal for the 2022 ACMG Annual Clinical Genetics Meeting. Proposal submission opened June 15 and proposals must be submitted through the ACMG submission website by Friday, July 30, 2021.

Visit the ACMG meeting website for more information and to submit a proposal.

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