ACMG In Action

Essential Updates: Member News You Can Use

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

July 2022

In This Issue...

ACMG-ASHG-NSGC Joint Response to Supreme Court Decision to Overrule *Roe v Wade*

On June 24th, ACMG, together with the American Society of Human Genetics (ASHG) and the National Society of Genetic Counselors (NSGC), issued a statement in response to the US Supreme Court decision to overrule *Roe v Wade*. Read the statement here.

See also the joint statement here from more than 75 healthcare organizations, including ACMG, opposing legislative interference in the patient-clinician relationship.

ACMG Seeks Editor-in-Chief for New Gold Open Access Journal

The ACMG is excited to announce the launch of a new gold open access journal titled *Genetics in Medicine Open*. ACMG CEO Max Muenke, MD, FACMG states: "*GIM Open* will be an international journal with a focus on medical genetics and genomic medicine, including all aspects of therapy. It will be a trailblazer for diversity, equity and inclusion and will have a double anonymous review process for submitted manuscripts."

The College is currently recruiting for an **Editor-in-Chief (EIC)** who will help determine the scope and direction of the journal and be responsible for its scientific content. Specifically, the new EIC will decide which manuscripts should be reviewed, evaluate reviewer's input and select the final manuscripts for publication. We are looking for someone who is passionate about genetics and genomics and who can help grow a brand new journal.

Visit here to learn more about this position or how to apply. Applications will be accepted through **July 31, 2022 at 11:59 PM EST**.

Sneak Peek: Summer Edition of Member Magazine Now Online

Upcoming Events

2023 ACMG Annual Clinical Genetics Meeting:
March 14-18, 2023

Website Links

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

The summer issue of *The ACMG Medical Geneticist* will be in your mailbox later this month, but you can read it online now in the members-only section of the ACMG website. The issue highlights the College's current education activities and takes a joyous look back at the 2022 ACMG Annual Clinical Genetics Meeting. In his quarterly message, President Marc S. Williams, MD, FACMG writes about two unique educational approaches – including one in which students spur learning in their mentors. The Q&A is with the ACMG Education and CME Committee Chair John Bernat, MD, PhD, FACMG, who elaborates on how the College is updating existing educational offerings while rolling out new ones. The issue will get you up to speed on the amazing number of educational resources offered by each of the College's grant-funded programs (ClinGen, NBSTRN and NCC). The ACMG Annual Meeting is all about education, and the 2022 edition is wrapped up including plenty of wonderful photos across several pages. Other highlights include profiles of the nine impressive recipients of the 2022 ACMG Foundation Awards, a feature on the children and families who participated in this year's Day of Caring, and a fun look at the successful 2022 Medical Genetics Awareness Week worldwide celebration.

Four Draft ACMG Documents Open for Member Comment

The ACMG Board of Directors routinely invites members to comment on draft ACMG documents, including new and updated laboratory technical standards, points to consider statements, clinical practice resources and more. The contribution of members' expertise to the review process for ACMG draft documents consistently leads to higher quality publications because members serve as expert peer reviewers—it is your valuable input that earns ACMG its continued reputation as the trusted experts in medical genetics and genomics practice.

Presently, the ACMG Board of Directors is requesting member comments on four draft documents:

• Phenylalanine Hydroxylase Deficiency Treatment and Management: A Systematic Evidence Review
• Solid Organ Transplantation in Methylmalonic Acidemia and Propionic Acidemia: A Points to Consider Statement of the American College of Medical Genetics and Genomics (ACMG)
• Noninvasive Prenatal Screening (NIPS) for Fetal Chromosome Abnormalities in a General-risk Population: An Evidence-based Clinical Guideline of the American College of Medical Genetics and Genomics (ACMG)
• Points to Consider in the Detection of Germline Structural Variants Using Next-generation Sequencing: A Statement of the American College of Medical Genetics and Genomics (ACMG)

For more information about each draft document, including instructions for accessing the document and deadlines for comments, please click on the document titles. Thank you for your participation in this important ACMG members-only activity.

Advocacy Alert: Ask Congress to Oppose Passage of the Current VALID Act

ACMG needs your help to prevent passage of the current version of the Verifying Accurate Leading-edge IVCT Development (VALID) Act. Congress is actively working to pass a legislative package that would reauthorize necessary user fee agreements for the Food and Drug Administration (FDA), and they are also considering inclusion of the VALID Act. **Click here to send a letter to your elected officials asking them to oppose the current VALID Act**.

As written in the version already passed by the Senate HELP Committee, the VALID Act would significantly hinder laboratories' ability to develop and use laboratory developed tests (LDTs). The bill would give FDA authority over all LDTs, and many would require premarket approval prior to use of a new or modified LDT. FDA premarket review takes many months per test and would be accompanied by costly fees for each review application. While Congress has included some exemptions for specific types of tests, the legislation is still very flawed and could have significant negative consequences for clinical testing laboratories, innovation in testing technology and patient access to clinical tests and timely results. The legislation has also been opposed by more than 130 organizations (see letter here).

READ MORE

GenePod: Next Generation Sequencing Data Reanalysis

Next generation sequencing has becoming increasingly powerful in diagnosing Mendelian disorders, yet typically more than 50 percent of cases remain unsolved after an initial clinical exome or clinical genome sequencing. So a team of researchers set out to review the literature to attempt to answer the question: Is it worth reanalyzing unsolved cases, and if so, when should such a reanalysis take place to reap the most possible benefit? Tri Phan, PhD, professor of medicine at the Garvan Institute and the University of New South Wales in Sydney, discusses the research on this month's GenePod.

ACMG Welcomes Seven New Members

ACMG welcomes and congratulates its seven 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.

Fellow Members
Marvin Natowicz, MD, PhD, FACMG
Tatiana Tvrdik, PhD, MGC, MS, FACMG

Associate Member
Brayden West, MGC, CGC

Affiliate Member
Jessica Cooke Bailey, PhD

Trainee Member
Lauren Choate, PhD

Student Members
Marija Debeljak
Emilie Chenda Ung

Thank you to all our members who make important contributions and the work of the College possible through your membership, support and involvement.

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

ICYMI: ACMG Releases Update to Secondary Findings List

On June 17, ACMG released "ACMG SF v3.1 List for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing: a Policy Statement of the ACMG," an update to the recommended minimum gene list for the reporting of secondary findings (SF). In 2021, the ACMG Board of Directors and Secondary Findings Working Group (SFWG) declared that the College would update the list (SF v3.0) annually. The recent update (SF v3.1) adds five new genes – four associated with dilated cardiomyopathy predisposition and one associated with hereditary transthyretin amyloidosis, a cause of heart failure.

"The v3.1 list is the first of our ongoing yearly updates and embodies our working group's goals of maintaining a minimum list of actionable results that will impact patients and their families in a positive way," said lead author and co-chair of the ACMG SFWG, David T. Miller, MD, PhD, FACMG.

New Knowledge Nugget: Mucopolysaccharidosis Type I

The National Coordinating Center for the Regional Genetics Networks (NCC) is proud to announce the third video of our Knowledge Nugget Series! The series, intended as a companion education module to the ACMG ACT Sheets, walks users through an individual ACT Sheet via a fun, short, animated video. The third NCC Knowledge Nugget is a companion to the Mucopolysaccharidosis Type I (MPS I) Newborn Screening ACT Sheet. Access the module and earn .25 *AMA PRA Category 1 Credit* ™ in the ACMG Genetics Academy.

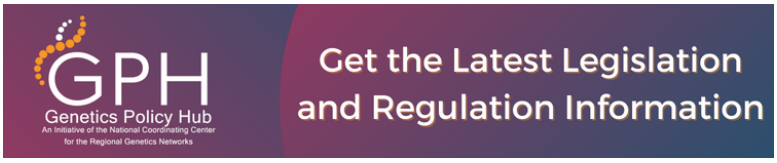
Explore all ACMG ACT Sheets and Algorithms at acmg.net/act. If you have any questions about the ACMG ACT Sheets, please contact Megan Lyon, NCC Associate Project Director.

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.

NCC Genetics Policy Hub Provides the Latest Information on Genetics Legislation



As the National Coordinating Center for the Regional Genetics Networks (NCC), we track proposed legislation and regulation at the state and federal level that could impact the delivery of genetic services. We update our tracking system with the latest information daily, which is then available on our policy website, Genetics Policy Hub, and on our GPH Twitter account. Access these or subscribe to our weekly newsletter to get the latest information in your state.

• **Genetics Policy Hub Website** - Filter proposed legislation and regulation by state or genetics policy topic.

• **Genetics Policy Hub Twitter – Learn about new proposed legislation or regulation, as well as any major actions taken on the policy. Get an overview of why the policy is related to the delivery of genetic services.**

• **Genetics Policy Hub Wrap-Up Newsletter** – Receive a quick overview each Friday of action taken at the state or federal level related to genetics policy. Access our wrap-up on the GPH Twitter account or receive the newsletter directly in your inbox.

The National Coordinating Center for the Regional Genetics Networks 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.

All Three Medical Necessity Webinars Now Available On-Demand

The National Coordinating Center for the Regional Genetics Networks (NCC) and the Catalyst Center, two programs supported by the Health Resources and Services Administration, are pleased to announce that all

three Medical Necessity webinars are available for *3 AMA PRA Category 1 Credits ™* in the ACMG Genetics Academy.

The three-part series includes the following information:

• **Part 1**-Define and Discuss the Use of Evidence to Form Medical Necessity Criteria and Policy
• **Part 2**-Medicaid and Early and Periodic Screening, Diagnosis, and Treatment (EPSDT)- The Title V and Medicaid Relationship
• **Part 3**-Practical Application of Medical Necessity: Understanding Prior Authorization Process, Requesting Authorizations, and Denials and Appeals

Have questions after watching one or more of the webinars? Please reach out to Megan Lyon, NCC Associate Project Director.

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2022 NBSTRN Network Meeting: Innovative Advocacy, Research, and Public Health Efforts to Advance Newborn Screening

NBSTRN | Newborn Screening Translational Research Network

Highlighting the efforts of national groups working to advance newborn screening through advocacy, research, and public health efforts, the 2022 NBSTRN Network Meeting will be held July 19-20. Please join us by registering for the event here!

Two presentations in particular showcase how NBSTRN helps to facilitate advancements in both policies and research to advance newborn screening. The first day will include a presentation by The EveryLife Foundation for Rare Diseases, a nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy. EveryLife presents their efforts in “RUSP Alignment Legislation” which implements a timeline for states to begin screening for conditions recommended for nationwide screening.

Dr. Ingrid Holm presents an overview of the BabySeq2 Project which is designed as an implementation study to expand and improve upon the BabySeq Project. The BabySeq project revealed unanticipated monogenic disease risks in 11% of newborn babies, and parent surveys using validated measures showed no evidence that newborn genomic sequencing caused increased psychological distress (i.e., no increased risk

of anxiety, depression, or family disruption compared to standard care) that persisted throughout the study, even if the baby had a disease risk identified.

READ MORE

Newly Available in the ACMG Genetics Academy

Gene Therapy: Ethical, Social and Economic Issues
Thursday, July 21 at 4:30 pm ET

The 2022 ACMG Summer Gene Therapy Education Series: Advancements in Gene Therapy Options for Rare Diseases is designed to provide a basic background on gene therapy for all healthcare professionals. This is the fourth session in a four-part series of live multidisciplinary team webinars. Experts will present new pathways in gene therapies and demonstrate best practices for patient care from existing guidelines from ACMG and ASGCT. The webinar will be 60 minutes with the last 10 minutes reserved for panel discussion/Q&A.

Learn more

2022 ACMG Summer Gene Therapy Education Series OnDemand

Did you miss one of the previous webinars in the Gene Therapy Series? Don't worry, you can find them here: The recorded series is free to ACMG Members and $50 for nonmembers.

- The Basics of Gene Therapy: Preclinical Development
- Gene Therapy Targets: Inborn Errors of Metabolism
- Gene Therapy Targets: Broadening the Scope of Therapy
- Gene Therapy: Ethical, Social and Economic Issues (coming July 27)

Evidence Based Guidelines Webinar 201: Exome and Genome Sequencing for Pediatric Patients with Congenital Anomalies or Intellectual Disability

Monday, July 25 at 4:00 pm ET

Register here

Miss an Evidence Based Guidelines webinar? *You can access recordings 24/7.*
Evidence Based Guidelines Webinar 101: Exome and Genome Sequencing for Pediatric Patients with Congenital Anomalies or Intellectual Disability - OnDemand!

Learn more

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

Tuesday, July 26 at 11:00 am – 12:00 pm ET

This monthly webinar series is for all medical and healthcare professionals and researchers interested in understanding cancer genomic testing and somatic and germline variant interpretation methods. It is presented as a collaboration between ClinGen Somatic and VICC consortia, and the ACMG.

Register now

ACMG Genetics101: A Course for Nongenetics Healthcare Professionals

Rapid advances in genetic and genomic knowledge have made it challenging for primary care and other nongenetics health care providers to stay current on recommendations and practices in clinical genetics. To address this gap, the ACMG has created **Genetics101 for Healthcare Providers**, an online accredited continuing education offering. In each module a board-certified medical genetics expert provides a case-based presentation. This activity has been approved for A*MA PRA Category 1 Credit*™.

Register here

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