ACMG In Action

Essential Updates: Member News You Can Use

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

June 2022

In This Issue...

Upcoming Events

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

Website Links

ACMG

Two Draft ACMG Documents Now 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 two draft documents:

• 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.

Call for ACMG Board of Directors Nominations: Deadline is June 15

The ACMG will hold elections this year for ACMG Board officers and directors. The following seats will be open in 2023 and the Nominations Committee is looking for your input on suggested replacements. If you have not done so already, please submit your nominations by tomorrow — June 15, 2022. Please see submission requirements below.

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

NOTE: A director of the College may not concurrently serve as a College employee, or as an officer, director or employee of the American Society of Human Genetics, the American Board of Medical Genetics and Genomics, the Society for Inherited Metabolic Disorders, the Association for Molecular Pathology or any other national or international professional organization that the Board of Directors deems to present a potential conflict with the Director's service to the College.

One (1) President-Elect Seat

Eligibility: Any Fellow in good standing (MD/DO or PhD) of the College is eligible to run for President-Elect. Physician Fellows must also be AMA members.

Term: Six years total: 2023 - 2029 (as President from 2025 - 2027)

Number of Candidates Needed:

• One (1) President-Elect to fill the seat that will be vacated by Susan D. Klugman, MD, FACMG.

Three (3) Director Seats

Eligibility: Any Fellow in good standing (MD/DO or PhD) of the College is eligible to run for the Board of Directors. Physician Fellows must also be AMA members.

Term: Six years: 2023 - 2029

Number of Candidates Needed:

• Two (2) Candidates - BOD Director Clinical Genetics to fill the seat vacated by Laurie Demmer, MD, FACMG
• Two (2) Candidates - BOD Director Clinical Molecular Genetics to fill the seat vacated by Elaine Lyon, PhD, FACMG
• Two (2) Candidates - BOD Director Clinical Cytogenetics to fill the seat vacated by Catherine Rehder, PhD, FACMG

Please submit the names of candidates for the Board of Directors to the ACMG Nominations Committee by **Wednesday**, **June 15, 2022**. **A current CV must also accompany each nomination submitted.**

The Nominations Committee will develop a slate of candidates from the suggestions provided by the membership. The Nominations Committee considers the following criteria for candidate selection: the maintaining of a diverse representation of certified geneticists on the ACMG Board, professional experience, previous service to the College, and the absence of a significant conflict of interest. An email notification with election participation instructions will be sent to members in late September. While all members may submit nominations, ACMG bylaws limit the right to vote to ACMG Fellow members.

Please contact the ACMG Nominations Committee with any questions about the election or call 301-718-9603.

GenePod: Harmonizing Gene–Disease Evidence Resources Globally

As more genes are implicated in disease, one of the challenges in implementing genomics in medical practice has been the lack of a single, standardized and shared genomics database, for both labs and clinicians to access. But a team of scientists have joined together to create the Gene Curation Coalition (GenCC), which includes participation from the developers of the leading publicly available genetics and genomics databases, such as OMIM, Orphanet, the Gene2Phenotype Database and ClinGen, as well as those involved in curating data at commercial laboratories (which has not until now generally been publicly available).

Marina DiStefano, PhD, FACMG, Assistant Professor at Geisinger in the Precision Health Program, and Heidi Rehm, PhD, FACMG, Chief Genomics Officer at Massachusetts General Hospital and Co-Director of the Medical and Population Genetics Program at the Broad Institute, and Medical Director of the Broad's clinical lab, join this month's GenePod to discuss the standardization process and the resulting database.

ACMG Welcomes 25 New Members

ACMG welcomes and congratulates its 25 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.

Fellow Members

Rebecca Ahrens-Nicklas, MD, PhD, FACMG

Jessica Gold, MD, PhD, FACMG
Nichole M. Owen, PhD, FACMG
Jennifer Schymick, MD, PhD, FACMG
Siby Sebastian, PhD, MS, FACMG
Richard Sidlow, MD, FACMG
Brittany Simpson, MD, FACMG
Agshin F. Taghiyev, PhD, MS, FACMG
Jariya Upadia, MD, FACMG

Candidate Fellow
Cedrik Ngongang, MD

Associate Member
Cristi Radford, MGC, CGC

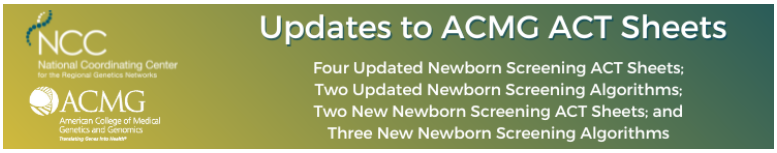
Affiliate Members
Melissa S. Cline, PhD
Cathleen Marshall, MD
Marcy Richardson, PhD
Hadley S. Smith, PhD, MPP
Sean V. Tavtigian, PhD
Joowon Yi, MD, MMedSc

Trainee Members
Yunjia Chen, PhD, MBA
Michael H. Duyzend, MD, PhD

Student Members
Brenna M. Boyd, MA
Sudipta Chakraborty
Kelsey Ferguson, BS
Zachary Thomas Sentell, BS
Sara Elizabeth Tsimerman, BS
Ugne Zekonyte, PhD

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

Now Available: Updated ACMG Newborn Screening ACT Sheets and Algorithms



The National Coordinating Center for the Regional Genetics Networks (NCC), in partnership with ACMG, is pleased to announce new and updated ACMG Newborn Screening ACT Sheets and Algorithms. Direct links to the ACMG ACT Sheets and Algorithms can be found below.

• Acid Sphingomyelinase Deficiency (ASMD) Newborn Screening ACT Sheet and Algorithm *formerly listed as Niemann Pick*
• Decreased Citrulline Newborn Screening ACT Sheet and Algorithm
• Isovaleric Acidemia Newborn Screening ACT Sheet and Algorithm
• Krabbe Newborn Screening ACT Sheets (Infantile Form) and (Late-Onset) and Algorithm *formerly one ACT Sheet; now split into two ACT Sheets*
• Short Chain Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency Newborn Screening ACT Sheet and Algorithm *formerly listed as M/SCHAD*

NCC thanks the NCC ACT Sheet Advisory Group, chaired by Dr. Dietrich Matern, NCC Medical Consultant, Dr. Nancy Rose, and the experts on the NCC Metabolic Conditions ACT Sheet Small Group for their leadership and expertise that allowed these Newborn Screening ACT Sheets and Algorithms to be updated.

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.

Register for the Last of Three Medical Necessity Webinars

The National Coordinating Center for the Regional Genetics Networks (NCC) and the Catalyst Center, two programs supported by the Health Resources and Services Administration, recently announced a three-part webinar series to help you navigate medical necessity. Each webinar focuses on various aspects of medical necessity through the lens of genetic cases.

Listed below is more information about the last webinar, to be held on June 29 at 3 PM ET, in the ACMG Genetics Academy.

• **Part 3 -** Practical Application of Medical Necessity: Understanding Prior Authorization Process, Requesting Authorizations, and Denials and Appeals

Miss the first two webinars in the series? Watch them on-demand in the ACMG Genetics Academy.

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.

The Catalyst Center (Grant U1TMC31757) is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling $500,000, with no financing by nongovernmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS or the U.S. Government.

X-ALD Knowledge Nugget Video Now Available

The National Coordinating Center for the Regional Genetics Networks (NCC) is proud to announce the second 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 using a fun, short, animated video. The second video in the series is a companion to the X-linked adrenoleukodystrophy (X-ALD) Newborn Screening ACT Sheet. You can access the module and earn .25 *AMA PRA Category 1 Credit* ™ in the ACMG Genetics Academy.

Explore all of the ACMG ACT Sheets and Algorithms at www.acmg.net/act. If you have questions about the ACMG ACT Sheets or the NCC Knowledge Nugget Series, 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.

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NBSTRN Recognizes Cytomegalovirus (CMV) Awareness Month

NBSTRN | Newborn Screening Translational Research Network

In 2011, Congress passed a resolution naming June as National Cytomegalovirus (CMV) Awareness Month and NBSTRN joins other rare disease stakeholders in efforts to increase awareness of the virus in the United States during this month. CMV is the most common infectious cause of birth defects in the country. CMV infection involves brain abnormalities leading to neurological problems that become apparent during infancy, affecting intellectual ability and the development of motor skills such as sitting and crawling or other health problems like hearing loss. Approximately 1 of 5 babies diagnosed with a CMV infection will develop birth defects.

The National CMV Foundation recently submitted a nomination for CMV to be added to the Advisory Committee on Heritable Disorders in Newborns and Children's (ACHDNC) Recommended Uniform Screening Panel (RUSP). The ACHDNC recommends that every NBS program in the nation include a standardized panel consisting of 35 core disorders and 26 secondary disorders. The ACHDNC provides the HHS Secretary with insight into grant and award allocation, technical information and ability to reduce mortality and morbidity for the screening of heritable disorders. The CMV nomination was an integral first step for CMV newborn screening to be implemented in state NBS programs. It is part of NBSTRN's mission to engage rare disease stakeholders and to assist with research, review, and gathering evidence to support the nomination of NBS-qualifying conditions to expand the RUSP. Click here to learn more about this nomination.

Check out the CDC's CMV Resource Center for fact sheets, videos, and other CMV resources for pregnant women, parents, and healthcare providers.

Visit the NBS Conditions Resource (NBS-CR) to learn more about CMV. Register as a member of the NBSTRN to become more involved in the NBS community.

ICYMI: ACMG Publishes SER on Cell-Free DNA Screening in General Risk Pregnancies

On May 24, ACMG released its second systematic evidence-based review (SER): "The Application of Noninvasive Prenatal Screening Using Cell-free DNA in General Risk Pregnancies." Noninvasive prenatal screening

with cell-free DNA (cfDNA), or NIPS, is used worldwide but, in the United States, there are insurance coverage limitations on who is eligible to receive this test as a covered benefit. Many insurers cover only those at higher risk, such as patients of advanced maternal age.

"We are pleased to report this systematic evidence review on cell-free DNA screening in pregnancy in a predominantly general risk population to consolidate the available recent data on various types of aneuploidies, twin gestations, maternal conditions and cost considerations regarding this test," said co-author Nancy C. Rose, MD, FACMG. "We hope that this report will inform public health policies in this area of care."

Now Available in the ACMG Genetics Academy

ACMG Summer Gene Therapy Education Series Provides Background for All Healthcare Professionals

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. There are four live multidisciplinary team webinars – the first was held June 9th. Each webinar is 60 minutes with the last 10 minutes reserved for panel discussion/Q&A.

Don't miss our next webinar: **June 23, 2022, at 4:30 pm ET**. "**Gene Therapy Targets: Inborn Errors of Metabolism**."

Learn more

Purchase the 2022 ACMG Short Courses Digital Edition

Did you miss the 2022 ACMG Annual Clinical Genetics Meeting? Are there sessions you were unable to participate in during the meeting or want to experience again? The 2022 ACMG Digital Edition offers on-demand access to select sessions in video or synchronized slides and audio with unlimited online and mobile access. The **2022 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.

Purchase the 2022 ACMG Short Courses Digital Edition

In addition to all of the other sessions, the content capture recordings of the two Short Courses offered at the 2022 Annual Meeting are each

available for purchase at the cost of $100 (member); $120 (nonmember).

• **Episodic Movement Disorder Phenotype in Children: Approach to Diagnosis, Review, and Updates of Selected Conditions**
Purchase Now

• **Clinical Applications of Long-Read Sequencing: Ending the Diagnostic Odyssey and Increasing Diagnostic Yield**
Purchase Now

Free Content from the 2022 ACMG Annual Meeting:

•Abstracts and Poster Gallery

•2022 Satellite Symposia, Industry Workshop Digital Edition

•2022 Exhibit Theaters Digital Edition

•2022 Digital Edition - Student Sessions

•2022 Diversity, Equity, & Inclusion - Imposter Syndrome: Confronting the Career Development Monster Hiding Under the Bed

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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