ACMG In Action

Essential Updates: Member News You Can Use

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

June 2021

In This Issue...

ACMG Publishes Revised Technical Standards for Clinical Genetics Laboratories

ACMG is pleased to announce the online publication of the *American College of Medical Genetics and Genomics (ACMG) Technical Standards for Clinical Genetics Laboratories (2021 Revision)*. These technical standards were developed by the Laboratory Quality Assurance Committee and contain significant updates.

The *ACMG Technical Standards for Clinical Genetics Laboratories*—one of ACMG's oldest and most popular publications—is a virtual textbook housed on the ACMG website. This textbook is comprised of seven sections and an introduction. As stated in the Introduction, these technical standards are developed primarily as an educational resource for clinical laboratory geneticists, to assist them in providing accurate and reliable genetic testing consistent with currently available technologies and procedures in the areas of clinical cytogenetics and genomics, clinical biochemical genetics, and clinical molecular genetics and genomics. They represent voluntary standards that establish criteria for clinical genetics laboratories to provide accurate and reliable diagnostic testing that is consistent with current technologies, procedures, and regulatory bodies.

Section A provides an overview of the standards; Section B discusses personnel policies; Section C details general policies; and Section D touches on shared methodologies. Sections E (Clinical Cytogenetics), F (Clinical Biochemical Genetics) and G (Clinical Molecular Genetics) have also been revised to best reflect current practices. Please note that these sections will be updated on a periodic basis to reflect newly published ACMG technical standards.

READ MORE

"The 2019 US Medical Genetics Workforce: A Focus on Clinical Genetics" Is Now Available in *Genetics in Medicine*

Privacy Preferences
- Now Available in the ACMG Genetics Academy
- 2022 ACMG Annual Clinical Genetics Meeting

Upcoming Events

ClinGen Somatic Cancer and VICC Virtual Molecular Tumor Board Case Series
June 29, 2021
11:00 AM–12:00 PM ET

2021 ACMG Genetics and Genomics Review Course – Online

Website Links

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

In the fall of 2019, the National Coordinating Center for the Regional Genetics Networks (NCC), developed and disseminated a survey to all board-certified medical geneticists in the United States to understand the characteristics of the workforce to inform workforce planning and public policy development. The survey, distributed in partnership with the American Board of Medical Genetics and Genomics (ABMGG), was completed by 984 medical geneticists. We are pleased to announce that the results of the survey are now available open access in *Genetics in Medicine*. To explore the manuscript and learn more about the demographics, practice characteristics and practice capacity of clinical geneticists in the United States, click here.

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ACMG Updates Recommendations for Secondary Findings Reporting, Releases SF v3.0 List

In case you missed it, ACMG has released an updated policy statement and gene list for the reporting of secondary findings (SF). Together, these two documents update the recommendations for SF reporting and unveil the highly anticipated recommended minimum gene list (SF v3.0) for reporting SF in clinical exome and genome sequencing. The ACMG also declares its intent to update the SF gene list now annually, with a goal of publishing the updated list each January.

The two papers, "Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing, 2021 Update: a Policy Statement of the American College of Medical Genetics and Genomics" and "ACMG SF v3.0 List for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing: a Policy Statement of the American College of Medical Genetics and Genomics," are the culmination of several years of work by the ACMG Secondary Findings Working Group (SFWG) and are published in ACMG's official journal, *Genetics in Medicine*.

"Our group worked very hard on this update, taking a thoughtful and careful approach that balances the goals of keeping it as a minimum list while also providing results that will impact patients and their families in a positive way," said co-lead author and co-chair of the ACMG SFWG, David T. Miller, MD, PhD, FACMG.

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ACMG Seeking Nominations for Systematic Evidence Reviews, Evidence-Based Guidelines

ACMG is seeking member input for new systematic evidence review (SER) topics as part of the College's Evidence-Based Guidelines (EBG) Program. The purpose of ACMG's EBG Program is to develop unbiased guidelines in medical genetics and genomics and provide a solid evidence base to demonstrate clinical utility, to help clinicians make informed decisions regarding the use of genetic and genomic testing and to help both government and private health insurers determine coverage options for new tests and treatments. An SER will be performed for new ACMG guidelines when appropriate and after approval of the topic by the Topic Selection Committee and the ACMG Board of Directors.

Don't miss this important and timely opportunity to help shape the future of the ACMG EBG Program! Topics for SERs can be submitted to the ACMG Topic Selection Committee online, using the committee's SER Topic Nomination Form. The form provides detailed instructions on formulating a question for submission using the PICOTS definitions for patient population (P), intervention (I), comparator (C), outcome (O), timing (T) and setting (S).

We look forward to receiving your nominations for topics **by Friday, June 18, 2021**. Topics submitted after June 18 will be considered for new projects on an ongoing basis.

Draft ACMG Document 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 the following document:

• Interpretation and Reporting of Large Regions of Homozygosity and Suspected Consanguinity/Uniparental Disomy, 2021 Revision: A Technical Standard of the ACMG

For more information, including instructions for accessing the document and the deadline for comments, please click on the document title above. Thank you for your participation in this important ACMG members-only activity.

June 2021 GenePod: Universal Newborn Screening to Identify Pediatric Cancer Predisposition — Could It Work?

Universal newborn screening has been successful at improving treatment and decreasing morbidity and mortality for a number of childhood diseases. Recently, a team

of researchers investigated the utility of newborn screening for rare genetic pediatric cancer syndromes. Knowing whether a newborn has a genetic variant strongly associated with pediatric cancer predisposition syndromes can potentially lead to focused surveillance of these infants, improved management, better health outcomes, and may even be cost-effective.

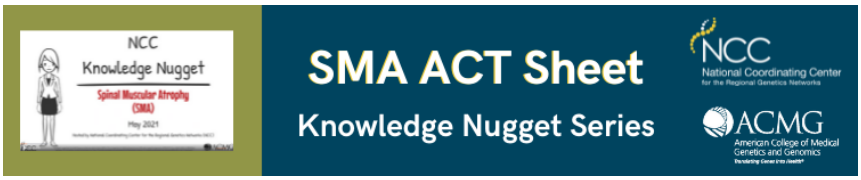
On this month's episode of GenePod, *Genetics in Medicine*'s monthly podcast, Lisa Diller, MD, professor of pediatrics at Harvard Medical School and vice chair of pediatric oncology at the Dana Farber Cancer Institute, and Jennifer Yeh, PhD, assistant professor of pediatrics at Harvard Medical School, discuss their model-based universal screening program to answer questions about potential benefits, costs and risks of universal newborn screening for pediatric cancer predisposition syndromes.

ACMG Annual Membership Business Meeting Planned for August

All ACMG members are cordially invited to attend the 2021 ACMG Virtual Annual Membership Business Meeting on Thursday, August 19, 2021 from 3:00 PM–4:30 PM ET. This virtual meeting will include reports from various departments as well as an update on *Genetics in Medicine* and the ACMG Foundation for Genetic and Genomic Medicine. Please plan on participating in the forum at the conclusion of the meeting, when members will have the opportunity to ask questions and make recommendations of initiatives that the College should pursue.

All ACMG members who wish to attend must register in advance in the ACMG Genetics Academy. There will be an option to submit any questions you have when you register for the meeting. Upon completion of registration for the meeting, an email will be sent with instructions on how to log in and participate. For questions about registration for the ACMG Virtual Annual Membership Business Meeting, please email Nataly Schwartz. Thank you and we look forward to seeing you on August 19 from 3:00 PM–4:30 PM ET.

NCC Knowledge Nugget Series: SMA Knowledge Nugget Available Now in the ACMG Genetics Academy



As you may have seen, the National Coordinating Center for the Regional Genetics Networks (NCC) launched their Knowledge Nugget Series last

month. The series, intended as a companion education module series to the ACMG ACT Sheets, walks users through an individual ACT Sheet by way of a fun, short, animated video. The first NCC Knowledge Nugget Series video is about the Newborn Screening (NBS) ACT Sheet on spinal muscular atrophy (SMA). You can access the module and earn .25 *AMA PRA Category 1 Credit™* in the ACMG Genetics Academy by clicking here.

To explore the ACMG ACT Sheets and Algorithms, click here. If you have any questions about the ACMG ACT Sheets or the new NCC Knowledge Nugget Series, 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: Accelerating the Pace of Newborn Screening Research

Learn How to Use NBSTRN Tools and Resources during the NBSTRN Annual Network Meeting, June 14–16, 2021

The NBSTRN team will conduct training sessions on NBSTRN data tools and resources from 3 PM to 4 PM ET on Tuesday, June 15 and Wednesday, June 16, during the Newborn Screening Translational Research Network (NBSTRN) annual Network Meeting, which will be held June 14–16, 2021. These interactive demonstrations will give attendees an inside look at NBSTRN's data tools and resources and the many ways they can be used to design and conduct innovative rare disease research studies. During these training sessions, we will discuss a use case and explore research questions for secondary data analysis using the available data sets on the NBSTRN website.

Are you registered for the NBSTRN annual Network Meeting yet? 2021 Network Meeting speakers include:

Monday, June 14, 2021:
• Mollie Minear, PhD and Melissa Parisi, MD, PhD – The National Institute of Child Health and Human Development (NICHD)
• Sarah Viall, PNP – Oregon Health & Science University (OHSU)
• Dorota Gruber, DHSc, MS, CGC – NewYork-Presbyterian Hospital

• Kathy Swoboda, MD – Massachusetts General Hospital

Tuesday, June 15, 2021:
• Ingrid Holm, MD, MPH – Boston Children's Hospital
• Chenelle Norman, MPH; Kayana Walters, MPH; and Sikha Singh, MHS, PMP - Association of Public Health Laboratories (APHL)
• William Wilcox, MD, PhD and Angela Wittenauer, MSN, FNPC, RN – Emory University

Wednesday, June 16, 2021:
• Sarah Bradley, MS, CGC, and Kathy Chou, PhD – New York State Department of Health
• Nicole Tartaglia, MD – Colorado Children's Hospital

• Ronald Wapner, MD; Jessica Giordano, MS, CGC; and Peter Robinson, MD, MS – Columbia University and The Jackson Laboratory

READ MORE

ACMG Welcomes 28 New Members

ACMG welcomes and congratulates 28 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

Irene J. Chang, MD, FACMG
Andrea Liliam Gomez Correedor, MS, DSci, FACMG
Samuel P. Yang, MD, FACMG

Candidate Fellow Members
Celeste C. Eno, PhD, MS

Rojeen Niazi, PhD

Louise C. Pyle, MD, PhD

Associate Member
Elena Ashkinadze, MGC, CGC

Affiliate Members
Kyle B. Brothers, MD, PhD
Amina Kozaric, PhD
Mauricio R. Murillo Vilches, MD

Victor B. Pastor Loyola, PhD, MS
Christine Yoshinaga-Itano, MA

Trainee Members
Katherine L. Mascia, MD
Nifang Niu, PhD
Rashedat Boluwatife Oshodi, MD
Hila Romi, MD, MS, DSci
Christina G. Tise, MD, PhD
Michael Volodarsky, PhD

Student Members
Cyrus Buckman, BA
Jordi Ysmael Camara, BS
Mallory A. DeCampos-Stairiker, BS

George Douganiotis, BS

Sahba Eskandari, BS
Sarah Lyon, BS
Tatiana Orlowski, BS
James Lewis Shepherdson, BS
Brady Slater, BS
Annika Lai-Mun Sundlof, BS

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

Verify Your ACMG Membership Directory Privacy Preferences

To protect your data and data preferences, ACMG only displays your information in our online membership directory if you have specifically opted in to appear. You also have options to specify which data to show or hide in your membership directory listing. Please verify your membership directory preferences today by clicking here.

Your data privacy and your communications preferences are important to us. At the ACMG, we are committed to both transparency and to protecting your privacy. In order to respect the privacy interests of all ACMG users (whether you are an ACMG member or not), the College is taking steps to determine your privacy wishes and attempt to manage your data accordingly. We want ACMG users to understand why their data is collected, how their data is used, and how ACMG protects their data; and we want to provide you with the flexibility to decide what information you want to receive, have shared or have removed from our systems. For information on how we protect your personal information, please see our ACMG Privacy Policy/Website Terms and Conditions of Use Notice.

Now Available in the ACMG Genetics Academy

ACMG Genetics Academy
acmgeducation.net

2021 Genetics and Genomics Review Course – Online

The 2021 ACMG Genetics and Genomics Review Course takes place throughout June and it's not too late to register for this important learning activity. Participants will take a 55-question practice exam as a baseline for discussion, attend watch parties or watch the 20 pre-recorded one-hour lectures on demand, and attend live Q&A webinars with the faculty. For more information and to register, click here.

ACMG Board Review Qbank 2021

Included with the 2021 Genetics and Genomics Review Course or available for separate purchase, the Board Review Qbank includes 636 practice test questions that you can draw from to find areas of strength and weakness in your genetics and genomics knowledge.
• When purchased separately from the 2021 Genetics and Genomics Review Course, $445 ACMG members; $545 for nonmembers.
• Two-year access
• 55 unique board-style practice questions with remediation
• Create your own practice exams by topic
• Ability to bookmark questions
• Ability to take notes

• **Peer comparison graphics**

• Can access on multiple devices

For more information and to purchase, click here.

Printed Syllabus from the 2021 ACMG Genetics and Genomics Review Course

The 2021 Printed Syllabus is the perfect add-on to help prepare yourself for the board exams or to refresh your knowledge. The printed syllabus is approximately 900 pages, printed in black & white and spiral bound. $280 ACMG members; $300 nonmembers. For more information and to purchase, click here.

2021 ACMG Annual Clinical Genetics Meeting – Digital Edition

2021 ACMG Annual Clinical Genetics Meeting DIGITAL EDITION

The 2021 ACMG Annual Clinical Genetics Meeting – Digital Edition is now available for purchase in the ACMG Genetics Academy. The Digital Edition gives you the full conference experience—with added convenience. Watch slides with video or view PDFs of presentation slide decks. We make it easy for you to access your content from any device!

Content will be available on July 19, 2021. For those who purchased the extended access when registering for the April virtual meeting, you will automatically be given access, and we will email you when the content is ready to view. If you have not already purchased the Digital Edition, you may do so now in the ACMG Genetics Academy by clicking here.

Registration Coming Soon for the Next ClinGen Somatic Cancer and VICC Virtual Molecular Tumor Board Case Series

June 29, 2021, 11:00 AM–12:00 PM ET

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

2021 ACMG Foundational Specialty Training in Cytogenetics and Molecular Genetics and Genomics: CME, P.A.C.E.® Credits Available

July 19–22, 2021, 4:00 PM–5:30 PM ET

With 19 recorded presentations and four live Q&A sessions, this course meets one criterion of the ABMGG requirements for training in the LGG Alternative Pathway. For more information, click here.

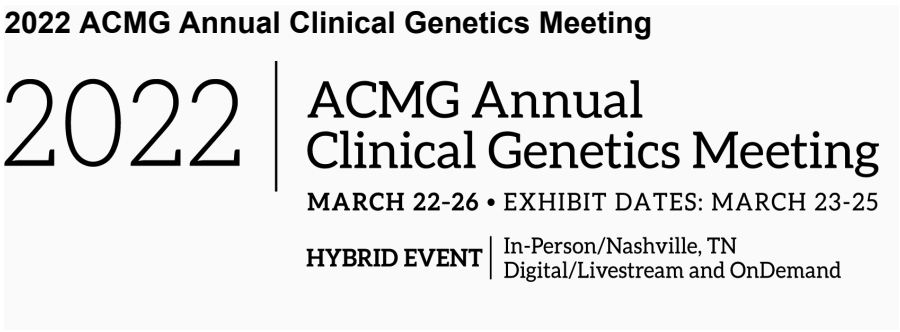
Coming Soon: New Part IV CCP Modules

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.

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.

***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.**

2022 ACMG Annual Clinical Genetics Meeting



2022 Proposal Submission Opens June 15

Plan now to submit a session proposal for the 2022 ACMG Annual Clinical Genetics Meeting, to be held March 22–26, 2022. The 2022 Annual Meeting will be presented as a hybrid event with an in-person component (in Nashville, Tennessee) and a digital component (online).

Proposal submission will open in June and proposals must be submitted through the ACMG submission website by Friday, July 30, 2021. More information and the link to the online submission site will be available on the ACMG Meeting website on June 15.

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