ACMG In Action

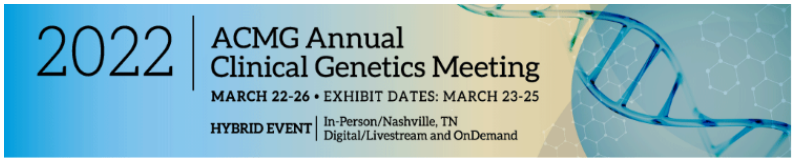
Essential Updates: Member News You Can Use

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

March 2022

In This Issue...

2022 ACMG Annual Clinical Genetics Meeting Updates and Reminders



The ACMG Annual Clinical Genetics Meeting is next week! We are excited to report that there will be more than 1,600 people participating in-person and 1,200 participating remotely.

Have you registered yet? There is still time! Visit the ACMG Meeting website to register now!

For those attending in-person, the following are important reminders regarding on-site health and safety protocols:

- ACMG is working diligently to provide as safe an environment as possible during the 2022 Annual Clinical Genetics Meeting. Please visit the Health and Safety page on the ACMG Meeting website to review the Health and Safety Protocols and download a copy if needed.
- Proof of vaccination is required for entry to the meeting. If you are already registered, you should have received a text from HealthShield, ACMG's vaccination verification partner, with instructions to upload your proof of vaccination against COVID-19. If the text was not received, US attendees may text "upload" to (866) 413-3462 to receive a text with the link to upload their proof of vaccination. International attendees need to go to the International Document Upload Site to upload their proof of vaccination.

Nashville!
- NBSTRN Session at ACMG Annual Clinical Genetics Meeting
- Now Available in the ACMG Genetics Academy

Upcoming Events

2022 ACMG Annual Clinical Genetics Meeting
March 22-26, 2022

4th Annual Medical Genetics Awareness Week, March 22-25

RENEW MEMBERSHIP

Website Links

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

Participating in the Meeting – In-person or Remote

This year, ACMG will be using an Online Event Platform in place of a mobile app. No matter where you are or how you are participating – from your office, your home or at the meeting in Nashville – this will be your key tool for the meeting. The platform is browser-based, can be accessed on all devices and there is no need to download an app. It is scheduled to go live on March 18 so that attendees can view the features and become familiar with it before the meeting starts. The link will be provided to all registered attendees in an email the day before it goes live.

Features include:

- Participate in chat for the session you are attending (in-person or remote)
- Send a question to the presenters in Q&A (in-person or remote)
- See the schedule
- Build your personal schedule
- View abstracts
- View ePosters
- Join a livestreamed session
- Watch sessions on demand
- Network with other attendees, including video chats for up to 15 people

Several sessions per day will be livestreamed and available to watch in the platform on demand shortly after each session ends. If a session is not livestreamed, the content (audio with a synch to the slides) will also be loaded in the platform as on-demand sessions shortly after each session ends.

Program

Be sure to review the full Program on the ACMG Meeting website to start planning the sessions you will attend. If the session is being livestreamed it will be indicated on the schedule.

Exhibit and Poster Hall

Visit the Exhibit Hall pages on the ACMG Meeting website to view participating exhibitors, the Exhibit Theater and the Learning Lounge session schedules.

Over 250 posters will be displayed in the Exhibit Hall with additional posters available as ePosters in the Online Platform. Posters will be available for viewing online beginning at 4:00 pm CT on Wednesday,

March 23 and in the Exhibit Hall beginning with the Opening Reception on Wednesday evening.

ACMG Liaison Opportunity to the American Academy of Pediatrics

The ACMG maintains liaison relationships with various organizations that work in the field of genetics and genomics. Liaisons represent the ACMG at external meetings and share updates of relevant College initiatives. Following external meetings, liaisons are requested to provide a brief report to the Board of Directors within 30 days of the meeting. Meetings may be held virtually or in-person and the ACMG will cover approved costs associated with travel to an approved meeting.

The ACMG Board of Directors is searching for a new liaison for the Council on Genetics of the American Academy of Pediatrics (AAP). This will be a three-year term and requires participation in two Council meetings that are held in the spring and fall of each year. If you or someone you know is a pediatrician, a Fellow of the AAP and the ACMG, please send a letter of interest and CV by March 24, 2022 to administration@acmg.net with the subject line: AAP Liaison. The new liaison will be appointed in April.

Advocacy Alert

Reducing Hereditary Cancer Act Introduced in Senate

On February 16, Senators Lisa Murkowski (R-AK) and Ben Cardin (D-MD) introduced S. 3656, the Reducing Hereditary Cancer Act, legislation that would allow Medicare to cover germline testing for individuals with a known or suspected personal or family history of a hereditary cancer. The bill would also allow Medicare to cover certain preventive surgeries and screenings as recommended by National Comprehensive Cancer Network guidelines or other nationally recognized professional organizations. This follows introduction of a companion bill, H.R. 4110, in the House of Representatives last year by Representatives Debbie Wasserman Shultz (D-FL), Mariannette Miller-Meeks (R-IA), Elissa Slotkin (D-MI) and Rodney Davis (R-IL).

ACMG joins approximately 100 other professional and patient organizations in support of the legislation.

Congress Introduces Bill on Pharmacogenomic Test Education

On February 28, Representatives Eric Swalwell (D-CA) and Tom Emmer (R-MN) introduced H.R. 6875, the Right Drug Dose Now Act, which would support education about pharmacogenomic (PGx) testing for the general public and healthcare professionals and incentivize improved incorporation of PGx information into electronic health records. ACMG joins a growing list of organizations in support of this bill. See ACMG's letter of support here.

ACMG Responds to RFI on NIH Genomic Data Sharing Policy

ACMG recently responded to a request for information (RFI) on potential updates to the National Institutes of Health (NIH) Genomic Data Sharing (GDS) Policy. The GDS policy, originally finalized in 2014, describes expectations for responsible sharing of genomic data generated from NIH-funded research as well as procedures for individuals wanting to access genomic information stored in NIH databases. The NIH is now considering updating the policy to ensure that it keeps pace with advances in technology and use of genomic data in research. Thank you to ACMG's Advocacy and Government Affairs committee and Social, Ethical, and Legal Issues committee for assisting with ACMG's response, available here.

Updated ACMG Newborn Screening ACT Sheets and Algorithms Now Available

The National Coordinating Center for the Regional Genetics Networks (NCC), in partnership with ACMG, is pleased to announce one new algorithm (Fabry) and five updated ACMG Newborn Screening ACT Sheets and Algorithms (Argininemia, Tyrosinemia, CPT II, Fabry ACT Sheet, and Biotinidase Deficiency). Direct links to the ACMG ACT Sheets and Algorithms can be found below.

• Argininemia Newborn Screening ACT Sheet and Algorithm

• **Tyrosinemia Newborn Screening** ACT Sheet **and SUAC Normal** Algorithm **Normal/Elevated and SUAC Elevated** Algorithm

• Carnitine Palmitoyltransferase II (CPT II) Newborn Screening ACT Sheet and Algorithm
• Fabry Newborn Screening ACT Sheet and Algorithm
• Biotinidase Deficiency Newborn Screening ACT Sheet and Algorithm

NCC sincerely thanks the NCC ACT Sheet Advisory Group, chaired by Dr. Dietrich Matern, Dr. Nancy Rose, NCC Medical Consultant 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, 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.

ACMG Publishes Technical Standard for Clinical Pharmacogenomic Testing

ACMG is pleased to announce the publication of the technical standard, "**Clinical pharmacogenomic testing and reporting: A technical standard of the American College of Medical Genetics and Genomics (ACMG).**" This technical standard was undertaken by the Laboratory Quality Assurance Committee's Molecular Genetics Subcommittee and supersedes the original laboratory standard on this topic (Lyon E, et al, *Genet Med*. 14(12):990-1000).

In a commonly used pharmacogenomic nomenclature system, combinations of sequence variants (i.e., haplotypes) are often designated by star (*) alleles. The *1 allele is often assigned as a "default" if none of the tested variants are detected. In addition, some pharmacogenes are affected by copy number variants, including gene deletions, duplications, multiplications and gene rearrangements. The analytical strategy used in pharmacogenomic testing depends on a variety of factors, such as the

complexity of the gene; the extent, frequency, and type of genetic variation; and the time needed for return of the results.

READ MORE

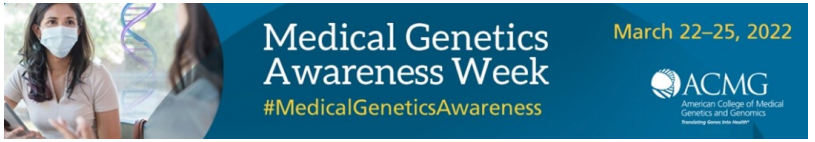
ACMG Publishes Technical Standard for Measurement of Lysosomal Enzyme Activities

ACMG is pleased to announce the publication of the technical standard, **"Measurement of lysosomal enzyme activities: A technical standard of the American College of Medical Genetics and Genomics (ACMG)."** This technical standard was undertaken by the Laboratory Quality Assurance Committee's Biochemical Genetics Subcommittee.

Lysosomal storage disorders (LSD) comprise a group of over 70 genetically distinct conditions. While most LSDs result from pathogenic variants in genes encoding specific lysosomal hydrolases, others arise from pathogenic variants in genes encoding activator proteins, transport proteins or proteins that post-translationally modify lysosomal enzymes.

READ MORE

Medical Genetics Awareness Week Is Fast Approaching!



This year's Medical Genetics Awareness Week is just six days away – March 22-25! How will you celebrate the contributions of the medical genetics team to patient care and public health? We look forward to seeing many of you in Nashville to celebrate with us during the 2022 ACMG Annual Clinical Genetics Meeting. If you cannot be there, however, you still will be an important part of Medical Genetics Awareness Week on social media and where you live, work and study.

Here are two great ways for you to join the conversation: Tell everyone why you love your work in medical genetics by posting a short quote explaining "**Why I'm in Medical Genetics**." We have a downloadable template for that. Or send pearls of wisdom to students and trainees with our "**Advice for Future Genetics Professionals**" template! Both ready-made templates are in our Resource Tool Kit. Simply download the template, add your headshot, professional title/credentials, and a few brief

sentences. Add the #MedicalGeneticsAwareness hashtag and share it on social media.

To support your participation, we have prepared an online Resource Tool Kit that offers resources you can use to raise awareness. The Tool Kit includes printable graphics to use on social media or to decorate your lab, office or classroom; draft social posts; a set of Medical Genetics Awareness Week hashtags; virtual Zoom backgrounds to use for meetings; and much more.

"Be a Champion for Medical Genetics" and join ACMG in celebrating Medical Genetics Awareness Week, March 22-25. Stay up to date on all the latest activity by following @TheACMG on Twitter, Facebook and Instagram.

GenePod: Genome Sequencing as a First-line Diagnostic Test in Newborns: Does It Serve the Underserved and Low-income Communities?

Genome sequencing holds great potential to diagnose newborns with phenotypes suggestive of a genetic disorder. However, this technology has not been widely adopted for this population, and particularly not in newborns from underserved and low-income communities.

To provide data as to the utility of this approach, a team of researchers in the South conducted a clinical study that aimed to detect genetic disorders via genome sequencing in underserved African-American and low-income communities and compared the results to a more typical diagnostic odyssey. On this month's GenePod, Greg Cooper, PhD, faculty investigator at the HudsonAlpha Institute for Biotechnology in Huntsville, Alabama discusses the results.

ACMG Welcomes 90 New Members

ACMG welcomes and congratulates its 90 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
Aya Abu-El-Haija, MD, MPH, FACMG
Neena S. Agrawal, MD, FACMG
Sara Akhavanfard, MD, PhD, FACMG
Najla A. Al-Sweel, PhD, MS, FACMG
Katherine Anderson, MD, MA, FACMG
Jorune Balciuniene, PhD, FACMG
Subit Barua, PhD, FACMG
Stela Berisha, PhD, FACMG
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Christie M. Buchovecky, PhD, FACMG
Rebecca J. Burke, MD, PhD, FACMG
Kameryn M. Butler, PhD, FACMG
Chun-An Chen, PhD, MS, FACMG
Andrew P. Dervan, MD, MBA, FACMG
Laura Dillon, PhD, FACMG
Marina T. DiStefano, PhD, FACMG
Xiaoli Du, PhD, FACMG
Maria L. Duque Lasio, MD, FACMG

Debbie M. Figueroa, PhD, FACMG

RaeLynn Forsyth, MD, FACMG
Jennifer M. Gass, PhD, FACMG
Aixa M. Gonzalez Garcia, MD, FACMG
Bin Guan, PhD, FACMG
Fen Guo, PhD, FACMG
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Lori Ramkissoon, PhD, FACMG
Linda Z. Rossetti, MD, FACMG
Jacquelyn J. Roth, PhD, FACMG
Inderneel Sahai, MD, FACMG
Kathleen M. Schieffer, PhD, FACMG
Ana Sequerra Amram Cohen, PhD, MS, FACMG
Nidhi D. Shah, MD, FACMG
Rebecca B. Smith, PhD, FACMG

Scott C. Smith, PhD, MS, FACMG

Andrew B. Stergachis, MD, PhD, FACMG
Thelma Alessandra A. Sugrañes, MD, FACMG

Panieh Terraf, PhD, FACMG

Fumihiko Urano, MD, FACMG
Francesco Vetrini, PhD, FACMG
Hongcheng Wang, PhD, FACMG
Heather E. Williams, PhD, MS, FACMG
Chung Wah Wu, PhD, FACMG
Catherine A. Ziats, MD, FACMG

Candidate Fellows
Theresa L. Kowalski, MD
Tamara S. Roman, PhD
Jing Xiao, PhD

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Natalie Beck, MGC, CGC
Christine Bergeon Burns, PhD, MS, CGC
Heidi Cope, MS, CGC
Jessica Kenney, MS, CGC
Sheetal Parmar, MS, CGC
Summer Pierson, MGC, CGC

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Angella Charnot-Katsikas, MD
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Nicole Tartaglia, MD, MS

Chelsea Zimmerman, PhD

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Erika Meaddough
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Anna N. Noreuil, MS
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Harpreet-Kaur Satnam-Singh, PhD, MS
Leila K. Schwanemann, BS
Amanda Marie Shrewsbury, MGC
Abigail Turnwald, BS
Laura Voss, BA

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

Don't Let Your Membership Benefits and Discounts Expire!

The 2022 membership renewal deadline was January 31, 2022, but ACMG is extending the renewal window to ensure all members have ample time to complete this process in view of the COVID pandemic.

If you have a medical or financial hardship prohibiting you from renewing, ACMG may be able to offer a reduction or waiver of dues for 2022. If you would like more information about hardship requests or need assistance with renewing membership, please email the Membership department or call the ACMG membership department at 240-858-8022.

ACMG's online renewal system is a fast, easy and secure way to renew your ACMG membership instantly and receive immediate payment confirmation. The online system accepts Visa, MasterCard, Discover and American Express payments. If you prefer to pay by check or by fax, download a 2022 Dues Renewal Form here. Renewal by phone is also available by calling 301-718-9603.

Renew today to avoid interruption of membership services and benefits. Membership payment must be current to register at the member rate for the ACMG Annual Clinical Genetics Meeting, receive member discounts in the Genetics Academy and access the ACMG Members Only sections of the ACMG website.

Any questions regarding membership renewal can be emailed to the ACMG Membership Department or call 301-718-9603.

ACMG Young Professional Fellow Membership

If you passed your initial medical genetics and genomics certification exam(s) this August, congratulations! You are now eligible for the discounted ACMG Young Professional Fellow Year One membership ($160). Please visit our Join ACMG webpage and select the Status Change option to submit your application and payment.

If you are completing an additional training/fellowship program, and want to remain a Trainee member, you will need to submit a completed Student/Trainee Verification Form via email to the ACMG Membership Department or fax to 301-718-9604. (Forms must be signed by the program director or by an authorized representative.)

Learn More about NCC in Nashville!

NCC is excited to be back in-person for this year's ACMG Annual Clinical Genetics Meeting! Throughout the week we will be participating in a platform presentation, poster presentations and be in the Exhibit Hall! Stop by our booth (**#732**) and play a life-size version of *Regionopoly* to learn about the efforts of the Regional Genetics Networks, NCC and Family Center to improve access to genetic services for underserved populations and win a set of travel playing cards!

Platform Presentation

Are you interested in learning more about how Medicaid provides coverage for genetic services? NCC will be presenting data from our State

Genetics Policies Medicaid Database and share trends we saw in 2021. Join us for this presentation in Davidson A from **3:30-3:45 PM CST** on **Wednesday, March 23**.

Poster Presentations

NCC has many different projects and initiatives that help improve access to genetic services. Learn about some of these initiatives by attending one of the posters.

Thursday, March 24 from 10:30-12:00 PM CST: Location: Exhibit Hall AB
•Disseminating Genetics Policy Information on Social Media
•Interpreting for Genetics: Crossing the Language Barrier in Communities
•Performance Measures for Evaluating Access to Genetic Services

Friday, March 25 from 10:45-12:00 PM CST: Location: Exhibit Hall AB
•Utilization of the ACMG ACT Sheets and Algorithms on the ACMG Website

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 Session at ACMG Annual Clinical Genetics Meeting Explores the Role of Medical Geneticists in Newborn Screening Research

NBSTRN | Newborn Screening Translational Research Network

Each year in the United States, more than 12,500 newborns are diagnosed with a condition through newborn screening. Rapid developments in new technologies to screen and treat newborns is expected to expand the number of conditions that are candidates for screening. For more than 14 years, ACMG has led the development of NBSTRN, a key component of the Hunter Kelly Newborn Screening Research Program at NICHD. NBSTRN will be presenting an Accredited Scientific Session titled, "Exploring the Role of Medical Genetics and Genomics in Advancing Newborn Screening Research" at the 2022 ACMG Annual Meeting on

Saturday, March 26th, 8:30 AM – 10 AM CST, in Music City Center room 106ABC. This session will feature unique perspectives from three highly accomplished medical geneticists: Cynthia M. Powell, MD, FACMG; Debra Freedenberg, MD, PhD, FACMG; and Beth A. Pletcher, MD, FACMG. Read more about them below.

Also, please visit the NBSTRN Exhibitor Booth #736 to meet the NBSTRN staff and discuss how NBSTRN can support your newborn screening research.

READ MORE

Now Available in the ACMG Genetics Academy

It's Here! ACMG Genetics101 for Healthcare Providers

The College is excited to announce the launch of "ACMG Genetics101 for Healthcare Providers," a new series which seeks to provide educational content on clinical genetics for the non-genetics health professional and which will be featured in the widely popular American Medical Association's AMA Ed Hub™. In each module of this free, accredited course, a board-certified medical genetics expert will provide a case-based presentation. This activity has been approved for AMA PRA Category 1 Credit™. This course is supported by an independent medical education grant from Illumina, Inc.

Topics include:

1. General Overview of Genetics
a. Utility of a Genetic Evaluation

b. Interpretation of Genetic Testing

2. Online Genetics Resources
3. Key Principles in Pharmacogenomics
4. Inherited Cancer Syndromes
5. Genetics for Cardiologists
6. Genetics for Neurologists

7. Genetics for Endocrinologists
8. Prenatal Genetics
9. Genetics for the Primary Care Provider (Adult)
10. Genetics Workup for the Pediatrician

To register and read about the series and faculty, visit here. Note – watch for a formal public announcement about the new Genetics101 series once it launches in the AMA Ed Hub™ in the coming weeks.

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

Tuesday, March 29, 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 ACMG.

Register Now

American College of Medical Genetics and Genomics | 7101 Wisconsin Avenue, Suite 1101, Bethesda, MD 20814

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