ACMG In Action

Essential Updates: Member News You Can Use

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

May 2022

In This Issue...

New ACMG Clinical Practice Resource Assists Treatment of Patients with Hearing Loss

ACMG has released a new Clinical Practice Resource that will help direct the treatment of patients with hearing loss: "Clinical Evaluation and Etiologic Diagnosis of Hearing Loss: A Clinical Practice Resource of the American College of Medical Genetics and Genomics." This ACMG Clinical Practice Resource offers information about the frequency, causes and presentations of hearing loss, and suggests approaches to the clinical and genetic evaluation of deaf and hard-of-hearing individuals aimed at identifying an etiologic diagnosis and providing informative and effective patient education and genetic counseling.

Lead author Marilyn M. Li, MD, FACMG said, "This clinical practice resource has integrated the current knowledge into clinical practice and has proposed a new diagnostic algorithm to ensure early detection and intervention for deaf and hard-of-hearing individuals to maximize language development and quality of life. Healthcare providers should pay attention to the performance characteristics of HL tests when selecting a test for HL, including test design, genomic regions covered, technologies used, analytic sensitivity and limitations of the test. As the knowledge regarding the genetic etiology of all childhood diseases, including HL, improves, genetic screening is likely to become part of more comprehensive universal newborn screening in the near future."

ACOG Shows Support for ACMG Document

ACOG has officially announced support of an important 2021 ACMG document: "Screening for Autosomal Recessive and X-Linked Conditions During Pregnancy and Preconception: A Practice Resource of the ACMG."

See ACOG's support of ACMG by clicking here and scrolling down to the 2022 documents. ACOG stated, "The American College of Obstetricians and Gynecologists supports the value of this clinical document as an educational tool." ACOG support denotes a lower level of approval than endorsement and means that ACOG considers the clinical

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

document to be of educational value to its members, although ACOG may not agree with every recommendation or statement in the document.

Call for ACMG Board of Directors Nominations

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 June 15, 2022**. Please see submission requirements below.

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.

ACMG in the News: ACMG Leaders Add Expertise to Medical Association Articles

ACMG President Marc S. Williams, MD, FACMG provided an interview for an article on genetic testing in the May issue of *The Nation's Health*, the official newspaper of the American Public Health Association (APHA) that goes to all APHA members and other subscribers. In the article, "When's it best to get a test?" Dr. Williams explains that because science is always rapidly changing "a negative test today will not necessarily be a negative test tomorrow. The advantage of genetic testing as opposed to almost any other type of testing that's done in medicine is that once we do the test, we can go back to that information and we can reinterpret it using the newest knowledge." The article is available for download here.

The American Association of Medical Colleges (AAMC) recently interviewed ACMG President-Elect Susan Klugman, MD, FACMG for an article on NIPS entitled "Prenatal screenings can lead to false positives, heightened anxiety" that was published online April 14. The piece was prompted by an article published weeks earlier by *The New York Times*, which Dr. Klugman responded to with a Letter to the Editor. "There's a big difference between a screening test and a diagnostic test," Dr. Klugman told AAMC. "The screening test is offered to detect potential disease. If the screen is positive, it is appropriate to consider a diagnostic test."

GenePod: Clinical Variant Analysis Tool Provides Systematic Method to Assess Genomic Testing Results

When a clinician receives the results of genomic testing, there are several tools that can help the clinician interpret those results: guidelines from ACMG/AMP, the Quest Diagnostic Laboratory scoring system, and the ClinGen gene-disease association framework to name a few. The challenge is that there's no single tool that can synthesize all the information in a way that helps the clinician – who may be unsophisticated in genomic test interpretation – understand how to use it with their patient. Indeed, clinicians might spend hours attempting to interpret the report and still not end up with something useful for their patient.

To tackle this challenge, a team of researchers headed by Hui-Lin Chin and Nour Gazzaz, under the mentorship of medical geneticist Cornelius Boerkoel, MD, PhD and a medical geneticist at the University of British Columbia, developed a new tool called the Clinical Variant Analysis Tool. Dr. Boerkoel and Dr. Chin, a pediatrician in the division of genetics and metabolism at the National University Hospital's Khoo Teck Puat National University Children's Medical Institute in Singapore, discuss the tool on this month's GenePod.

ACMG Welcomes 30 New Members

ACMG welcomes and congratulates its 30 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

April Adams, MD, MS, FACMG

Stephanie M. Baskin, MD, FACMG
Zhenbin Chen, MS, DSci, FACMG
Cheryl Garganta, MD, FACMG
Ayuko Iverson, MD, FACMG
Kaitlin C. Lenhart, PhD, FACMG
Ryan H. Peretz, MD, FACMG
Priya Prasad, MBBS, MD, FACMG
Sara Procknow, MD, PhD, FACMG
Kristen L. Sund, PhD, MGC, FACMG
Liesbeth Vossaert, PhD, ScM, FACMG
Stephen J. Wicks, PhD, FACMG
Fang Xu, PhD, FACMG
Chen Yang, PhD, MS, FACMG

Associate Member
Afifa Hamilton, MS, MPH, CGC

Affiliate Members
Rhonda Anderson, MSN
Dylan C. Brock, MD, MA
Kimberly Ezell, MSN
Luis E. Figuera, MBBS, DSci
Taylor Kerrins, MD
Katherine Maria Lindsay
Jennifer Stefanich, MSN

Trainee Members
Casey Brewer, PhD
Michael A. Finkel, DO, MPH
Madhulatha Pantrangi, PhD
John Wang, MD, PhD

Student Members
Julie Boyd, MSN
Anna L. Capria, MS
Caroline Taylor Mayk
Badí Israel Quinteros Espinoza

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

ICYMI: ACMG Foundation Clinical Genetics and Medical Biochemical Genetics Applications Now Available

All members should have received an eblast message last week announcing that the ACMG Foundation for Genetic and Genomic Medicine's (ACMGF) Next Generation Fellowship and Residency Training Program is now accepting institutional applications for **Clinical Genetics** and **Medical Biochemical Genetics** residencies. Only applications for those two specialties are currently being accepted. For a detailed timeline for all Next Generation Fellowship awards click here. Based on feedback from last year, and discussions with several program directors, the ACMG Foundation will not require applying institutions to name a trainee as part of their application. Institutions can apply until **June 17, 2022**.

Each year, the ACMG Foundation grants its Next Generation fellowship awards to promising early career professionals in a range of medical genetics and genomics specialties. We are strongly encouraging applications from **all** institutions, including those who have not participated in the past. If you have any questions, please contact Nataly Schwartz.

Public Health Genetics Week is Around the Corner! May 23-27, 2022

Join the National Coordinating Center for the Regional Genetics Networks (NCC) in celebrating the third annual Public Health Genetics Week (PHGW) from May 23-27! The purpose of the week is to raise awareness and celebrate the field of public health genetics. During the week, each day will have the following themes:

• May 23 - What is Public Health Genetics?
• May 24 - Who is Involved in Public Health Genetics?
• May 25 - Public Health Genetics Programs
• May 26 - Public Health Screening
• May 27 - Public Health Genetics Resources

Find information about each day, along with fact sheets and social media images, at phgw.org.

READ MORE

Request for Volunteers: NCC Hemoglobinopathies ACT Sheet Small Group

The National Coordinating Center for the Regional Genetics Networks (NCC) is seeking providers who have expertise in working with infants who have hemoglobinopathies to serve on the Hemoglobinopathies ACT Sheet Small Group. Experts on this group will be charged with reviewing and editing the existing Hemoglobinopathies Newborn Screening ACT Sheets and Algorithms. Individuals who serve on the group will be expected to spend approximately two hours per month on activities related to the group: a one-hour monthly call and an estimated hour reviewing and editing materials prior to the call. The group will be convened for about six months (mid-summer to winter 2022).

If you are interested in serving on the group or know someone who may be interested, please send your CV to Molly Caisse, NCC Project Coordinator.

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 Second of Three Webinars about Medical Necessity

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 excited to announce a three-part webinar series to help you navigate medical necessity. Each webinar will focus on various aspects of medical necessity through the lens of genetic cases.

Listed below is more information about the remaining two webinars. Be sure to register for Part 2, which will be held online here on May 20 at 1 PM ET in the ACMG Genetics Academy.

• **Part 2** - Medicaid and Early and Periodic Screening, Diagnosis, and Treatment (EPSDT)- The Title V and Medicaid Relationship - May 20 at 1 PM ET
• **Part 3** - Practical Application of Medical Necessity: Understanding Prior Authorization Process, Requesting Authorizations, and Denials and Appeals - June 2022

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.

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NBSTRN 2022 Network Meeting to Commemorate Decade of Advances in Newborn Screening Research

NBSTRN | Newborn Screening Translational Research Network

Join us to celebrate advances in newborn screening research that have helped to improve health outcomes for babies with rare genetic diseases. On July 19th and 20th, the Newborn Screening Translational Research Network (NBSTRN) will host the 2022 Network Meeting from 12 PM to 4 PM ET. NBSTRN was founded in 2008 as a key component of the Hunter Kelly Newborn Screening Program at the Eunice Kennedy Shriver National Institutes of Child Health and Human Development (NICHD) through a contract with ACMG. NBSTRN has now grown into an international network involved in supporting cutting-edge research, population-based pilots and ethical, legal and social (ELSI) discussions. Our annual Network Meeting draws upon this growing network of research, clinical care and advocacy to present innovations in newborn screening research.

Each day will feature three hour-long sessions organized into a 30-minute presentation followed by a live discussion. Presentations will describe innovative efforts to incorporate whole-genome sequencing into healthy newborn screening, initiatives to modernize newborn screening and snapshots of ongoing projects using NBSTRN data tools and resources. The NBSTRN team will provide training on the use of the tools and resources that support this important work including the Longitudinal Pediatric Data Resource (LPDR), the NBS-C Condition Resource (NBS-CR) and the NBS Virtual Repository of States, Subjects & Samples (NBS-VR).

Sign up for this event to learn about the latest developments in newborn screening research. The 2022 Network Meeting will be archived.

200th Birthday Celebrations Continue for Gregor Mendel, Founder of Genetics

The founder of genetics, Johann Mendel, was born on July 22, 1822. As we approach what would have been his 200th birthday, the medical genetics community has been celebrating him in order to foster more awareness of his life, achievements and contributions to genetics.

Far from being a lonely monk (as he is sometimes described), Mendel was well mentored and had mentees, was a polymath scientist, social activist, and bank director, and helped nephews get to medical school. A pioneer in big data science (28,000 plants), he applied quantitative tools from physics and mathematics learned at the University of Vienna to qualitative biologic data.

At the 2022 ACMG Annual Clinical Genetics Meeting in March, Joseph McInerney, executive vice president of ASHG from 2013-2017, delivered

a TED-style talk entitled "Honoring Mendel and Exploring Some Educational Boxes We've Built from His Work," as well as an update of an international program of education, celebration, and commemoration. *Nature Genetics* opened 2022 with an editorial and the January 2022 *American Biology Teacher* had a Mendel cover and theme issue. *Plant Cell* had a special Mendel issue and soon, *Heredity* and *National Geographic* will as well. A new book, *Gregor Mendel: His Life and Legacy* by Daniel J. Fairbanks is due to be out in August, and an expanded compilation of images is expected from the Mendelianum Museum.

READ MORE

Now Available in the ACMG Genetics Academy

ClinGen Somatic Cancer and VICC Virtual Molecular Tumor Board Case Series - May Webinar

Tuesday, May 24, 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 here

Medical Necessity Webinar Series OnDemand
Did you miss a webinar? Don't worry, you can find them here.

New Part IV CCP Modules

Diagnosis and Management of Adult Patients with Cowden Syndrome (version 2022):

This module is for geneticists who are involved in the diagnosis and management of an adult patient with a diagnosis of Cowden syndrome or PTEN hamartoma tumor syndrome.

Purchase Now

Counseling for Inherited Cancer Risk (version 2022):

This module concerns counseling for individuals presenting for evaluation of a possible hereditary risk for cancer based on personal or family history.

Purchase Now

Save the Dates: 2023 ACMG Clinical Genetics Meeting Set for Salt Lake City

ACMG returns to Salt Lake City in March 2023 and is planning for an in-person meeting. We will offer a diverse educational program and the meeting will include a joint session with the Society for Inherited Metabolic Disorders (SIMD) on Saturday. The meeting will feature "tracks" that will aid the attendee in selecting sessions.

Salt Lake City is one of the most popular destinations for past ACMG meetings. The city and surrounding area offer a one-of-a-kind combination of metro and mountain - an urban oasis with a breathtaking, majestic alpine backyard. Plan to explore the walkable, vibrant, artistic downtown with plenty of shopping and entertainment options, or take advantage of the multitude of opportunities for outdoor recreation.

Miss the 2022 ACMG Annual Meeting? No Worries – Get the Digital Edition(s)

Did you miss the 2022 ACMG Annual Clinical Genetics Meeting in Nashville or online? Are there sessions you were unable to participate in during the meeting? 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. If you have not yet purchased the Digital Edition, you can purchase it here. The cost is $349 (member) and $399 (nonmember) and includes the ability to claim CME, P.A.C.E.® and NSGC credits until April 30, 2024.

If you purchased the extended access when registering for the March meeting, you should have received instructions to access the content from education@acmg.net on May 2nd. If you did not receive the instructions, please email education@acmg.net.

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**

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