

ACMG In Action

Essential Updates: Member News You Can Use

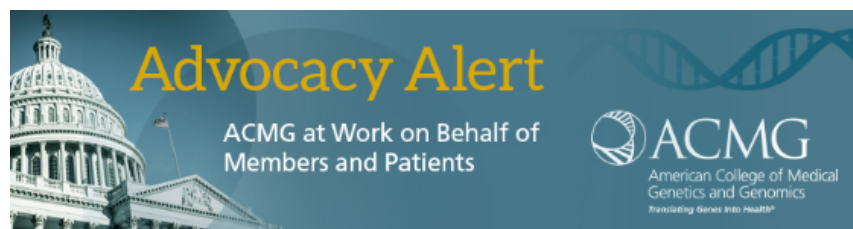


October 2019

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Advocacy Updates



Regulation and Oversight of Laboratory-Developed Tests

Although it's been quiet recently, Congressional efforts to reform regulation of in vitro clinical tests (IVCTs), including laboratory-developed tests (LDTs), are still underway. The most recent draft bill, the Verifying Accurate, Leading-edge IVCT Development (VALID) Act, was released in December 2018. If enacted, the draft bill would create a risk-based framework for regulation of LDTs by the Food and Drug Administration (FDA). While the current draft includes significant improvements over past proposals, we are still concerned about redundancy with the Clinical Laboratory Improvement Amendments (CLIA), unnecessary regulatory burdens for clinical testing laboratories and interference with the practice of medicine. Keep an eye out for future updates from ACMG as Congress continues to pursue legislation to reform regulation and oversight of IVCTs. Note: A joint working group of the Laboratory Quality Assurance and Professional Practice and Guidelines Committees is currently seeking member comment on the revised "Risk Categorization for Oversight of Laboratory-Developed Tests for Inherited Conditions: A Position Statement of the American College of Medical Genetics and Genomics (ACMG)." See the article "If the Regulation of LDTs Is on Your Mind, We Need Your Feedback" below.

Newborn Screening Saves Lives Reauthorization Act

Urgent action is needed by Congress to pass the Newborn Screening Saves Lives Reauthorization Act (NBSSLRA) of 2019. Newborn screening is one of the most successful public health programs in the United States and the federal funding authorized by this legislation is critical for the continued success of newborn screening. Unfortunately, the

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Upcoming Events

2020 ACMG Annual Clinical Genetics Meeting

March 17–21, 2020

Henry B. González Convention Center

San Antonio, Texas

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MEMBERSHIP**

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current legislation expired on September 30, 2019. Although the House of Representatives passed the bill in July, it remains in process in the Senate. ACMG is working closely with Congress to encourage the swift passage of the NBSSLRA.

Patenting of Genetic Information

Earlier this year, ACMG notified members about a draft bill to reform section 101 of the Patent Act. Section 101 currently prohibits patenting of laws of nature, natural phenomena and abstract ideas. The draft bill, released by bipartisan members of the Senate and House, sought to remove those prohibitions and abrogate several landmark court cases, including *Association for Molecular Pathology v. Myriad Genetics, Inc.*, which clarified a prohibition on gene patenting. ACMG quickly took action to help Congress understand the harms that would arise if patenting of genetic information were permitted. The bill sponsors have continued to meet with interested stakeholders over the past few months to improve the language, and we anticipate that a revised draft bill will be released before the end of the year. ACMG is prepared to continue working with Congress to ensure that any patent reform efforts do not lead to barriers in genetics and genomics research and clinical care by permitting patenting of genetic information.

ACMG Thanks Dr. Michael S. Watson for 19 Years of Service to ACMG and the ACMG Foundation for Genetic and Genomic Medicine



When Michael S. Watson, MS, PhD, FACMG became the first executive director of the ACMG in 2000, it would have been impossible to imagine the field of medical genetics today and the pivotal role ACMG would play in the evolution of the field. Now, as Mike prepares to depart ACMG later this year, the College conveys its enduring gratitude for his immense contributions to the ACMG, the ACMG Foundation for Genetic and Genomic Medicine and the grants he has managed while with ACMG. Mike has devoted himself to advancing the missions of the College

and Foundation and it is no hyperbole to say that his hard work and commitment to the growth of ACMG have made the organization what it is today and that he leaves the College in a strong position to face the exciting challenges and growth of the field ahead.

ACMG President Anthony R. Gregg, MD, MBA, FACOG, FACMG said, “In this age of genomic medicine, it may be hard for some to remember

when newborn screening was fragmented at a national level. Some states had no screening program or screened a minimum number of conditions. The same period in time was marked by prenatal screening that was at best targeted to one or a few ethnic groups. Medical foods were considered too expensive and were a luxury item for children with PKU and other metabolic conditions. Dr. Mike Watson was a key leader in the development of a nationally adopted newborn screening program. He guided one of the most important and first-ever collaborations between professional organizations. ACMG, the American College of Obstetricians and Gynecologists and the American Academy of Pediatrics collaborated to establish laboratory standards when reporting cystic fibrosis variants and there was agreement on a uniform approach to panethnic prenatal carrier screening. He was a vocal advocate for the diagnosis and treatment of children with metabolic conditions. Mike did all of this and much more under the ACMG moniker. His collaborative leadership, tenacious spirit, focus and high intellect allows ACMG the worldwide status it enjoys today. We are the “go to” source for genetics and genomics information; the “go to” source for policy makers, patients and providers, because of Mike’s leadership. We are proud members of ACMG. We can be proud to know that Mike leaves children with rare diseases, families seeking more information about their health risks and the nation positioned and equipped to take advantage of everything genomic technologies and their applications have to offer.”

[READ MORE](#)

ACMG and ASHG Work Together to Make a Difference, Host Joint Congressional Briefing on Capitol Hill



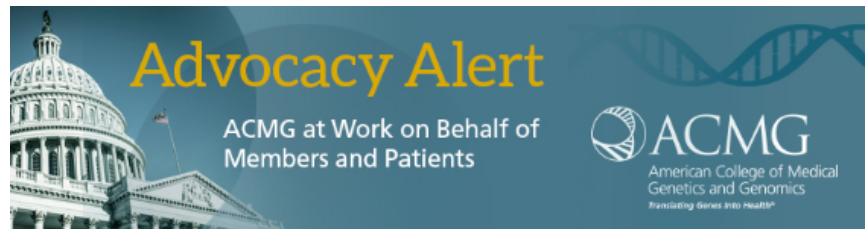
From left: Katie Murray, Health Policy Advisor for Rep. Loeb sack; John Phillips, III, MD, FACMG, Co-Principal Investigator at Vanderbilt UDN; Bill Gahl, MD, PhD, FACMG, Director of the Undiagnosed Diseases Program at NHGRI; Gail Jarvik, MD, PhD, FACMG, Co-Principal Investigator at University of Washington UDN; and Danny Miller,

ACMG and the American Society of Human Genetics (ASHG) are partnering on efforts to help members of Congress understand the value of genetics and genomics research, its translation to clinical applications that benefit patients, and how clinical findings inform further research. On September 26, 2019, ACMG and ASHG hosted their first joint Congressional briefing, *The Undiagnosed Diseases Network (UDN): The Interface of Research and Clinical Care to Solve Medical Mysteries*. Speakers Gail Jarvik, MD, PhD, FACMG; Bill Gahl, MD, PhD, FACMG; John Phillips, III, MD, FACMG; and Danny Miller, father of two children diagnosed through the UDN, spoke about the UDN to help Congress appreciate:

- how federally funded research is using genomic sequencing technology to help diagnose patients with ultra-rare and difficult to diagnose diseases;
- how the information learned from genetics research and clinical investigation is leading to improvements in the broader healthcare system, including through the use of federally funded data-sharing systems;
- the growing importance of genomic sequencing in establishing a diagnosis and improving health outcomes, and the potential for the UDN-like approach to minimize an expensive diagnostic odyssey; and
- the patient perspective regarding the value of the UDN and its impact on their health.

ACMG thanks Representative Dave Loebsack (Iowa) and his staff for making this event possible and for their continued support of genetics and genomics in healthcare. We look forward to continuing to partner with ASHG and the opportunity for future Congressional briefings.

ACMG Publishes Statement on Access to Reproductive Options after Prenatal Diagnosis—Patient Access and Physician Responsibilities



The ACMG is concerned with the enactment of laws in some states that prevent or restrict access to termination of pregnancy after prenatal diagnosis of genetic disorders or congenital anomalies and, as a result, recently made a strong public statement against these current or proposed laws. This new statement is an update to ACMG’s previous statement on access to reproductive options, which was released in 2013.

The 2019 “Access to reproductive options after prenatal diagnosis—patient access and physician responsibilities: an updated position statement of the American College of Medical Genetics and Genomics (ACMG)” states, “The practice of medical genetics is predicated on the principle of providing patients with complete and accurate information on

the condition that affects them, a member of their family, or an unborn fetus, and then discussing the management options that are available. The ACMG believes strongly that a balanced discussion of reproductive options is required. In some cases, patients will elect to alter the course of their pregnancy or pregnancy care. Patients may decide to investigate adoption or choose to learn about the expected outcome, neonatal care, and long-term care of a child with disabilities. They may also choose termination of pregnancy. Pregnancy termination should be available to pregnant women when their fetus has been diagnosed with a genetic disorder or congenital anomaly. Ideally, this is considered after discussions between the patient's healthcare provider(s) and the pregnant woman or couple with the goal of facilitating their choice of a safe and acceptable management plan. Access to safe and legal termination of pregnancy for genetic disorders or congenital anomalies that are diagnosed prenatally is an important option for some patients, and the ACMG strongly opposes any legislation that places limits on this access.

Further, it is the legal and ethical responsibility of healthcare professionals to provide complete and accurate information including all management options to patients. Laws that criminalize healthcare professionals for informing patients of their options, including termination of pregnancy, are contrary to the sanctity of the doctor-patient relationship and principles of autonomy. Accordingly, ACMG strongly opposes any legislation that prohibits healthcare professionals from providing complete and accurate medical care, or in any manner penalizes the provision of such care.”

The updated statement was published online in *Genetics in Medicine* on October 3, 2019 and is available by [clicking here](#).

If the Regulation of LDTs Is on Your Mind, We Need Your Feedback

Do you stay up at night worrying about regulatory changes to the oversight of Laboratory-Developed Tests (LDTs) and how these might affect your practice? Or perhaps you are comforted by the fact that ACMG has your back on this issue. Either way, WE NEED YOUR INPUT!

A joint working group of the Laboratory Quality Assurance and Professional Practice and Guidelines Committees recently reconvened to update [ACMG's 2012 statement](#) on the regulatory oversight of LDTs. This revision addresses advances in testing technologies and the current regulatory climate.

The revised document **Risk Categorization for Oversight of Laboratory-Developed Tests for Inherited Conditions: A Position Statement of the American College of Medical Genetics and Genomics (ACMG)**, one of ACMG's most widely used position statements in the public policy arena, is **open for member comment through Wednesday, November 6 at 11:59 PM PT**.

The addition of our members' collective expertise in the review process for ACMG's draft documents consistently leads to higher quality publications, as you are the peer reviewers. It is your valuable input that earns us our continued reputation as the trusted experts in medical genetics and genomics practice.

[READ MORE](#)

Malignant Hyperthermia ACT Sheet Now Available



The [National Coordinating Center for the Regional Genetics Networks \(NCC\)](#), in partnership with ACMG, is excited to announce a new Secondary Findings ACT Sheet: [Malignant Hyperthermia](#). You can find

this and all the other ACT Sheets at www.acmg.net/act.

NCC sincerely thanks the NCC ACT Sheet Workgroup, led by Dr. Dietrich Matern and Dr. Harvey Levy, as well as the reviewers of these ACT Sheets for their work. If you have any questions about the ACMG ACT Sheets, please contact NCC Associate Project Director Alisha Keehn (akeehn@acmg.net) or NCC Project Manager Megan Lyon (mlyon@acmg.net).

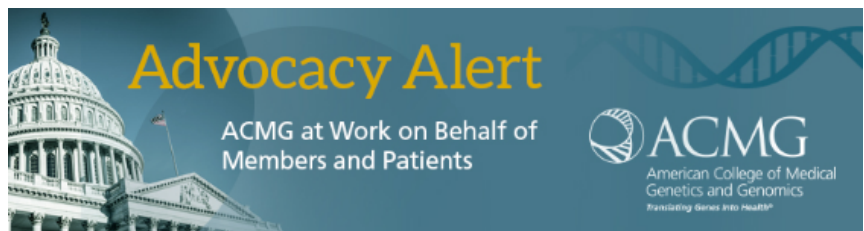
October 2019 *GenePod*: Variants on the Corresponding Allele May Explain Atypical Clinical Features in Patients with 22q Deletion Syndrome



One of the most common chromosomal deletions in humans happens on chromosome 22. It's known as 22q and affects as many as one in 2,000 live births. People with the deletion can present with a wide variety of traits. Some have heart defects. Others have defects in the palate, while still others have developmental delays. But in 2005, Donna

McDonald-McGinn, director of the 22Q and You Center at the Children's Hospital of Philadelphia and clinical professor at the University of Pennsylvania, noticed some patients had rare, atypical clinical features such as limb differences and rectal malformations. Now, in a [recent publication](#) in *Genetics in Medicine (GIM)*, she and an international team of scientists reveal variants in a gene called *CDC45* on the non-deleted chromosome are responsible for the unusual traits. The 22q deletion had unmasked the effects of the *CDC45* variants. On this month's [GenePod](#) podcast, clinician-researcher Donna McDonald-McGinn and host Cynthia Graber chat about how this unmasking phenomenon may be responsible for other conditions as well.

The 2019 ACMG Salary Survey Is Live



On October 3rd, all active fellows, candidate fellows and trainees should have received an invitation from survey@infosurveyfeedback.com to participate in the 2019 ACMG Salary Survey of Board-Certified Medical Geneticists. This survey is conducted every other year as an ACMG member benefit. The resulting Salary Survey Report is made available for free to all ACMG members and is available for purchase by nonmembers. Past surveys can be accessed in the Members Only section of the [ACMG website](#).

Increasingly, such survey reports are being used by individual ACMG members as well as department heads, chairs and deans to adjust, standardize and equalize salaries and remain competitive. Therefore, this survey has direct and immediate impact on the ability to grow the field and on your ability to have data to support discussions within your institutions. Please help us achieve a record participation rate this year so we can maximize the analyzability and statistical significance of the data. **The survey will remain open until Friday, November 15th.** For any questions about the salary survey, please contact Michelle McClure at mmclure@acmg.net.

ACMG Board Member Dr. Susan Klugman and NBSTRN Associate Project Director Dr. Amy Brower Are Featured in Mediaplanet's *Rare Diseases* Campaign



On September 27, 2019, *USA Today* published the printed supplement “[Future of Personal Health](#)” as part of Mediaplanet’s *Rare Diseases* campaign. The campaign invites readers to discover how professionals across a variety of fields and individuals living with rare diseases are working to raise public awareness about rare diseases and usher in a new wave of innovation in treatment and prevention.

ACMG board member and Vice President of Clinical Genetics Susan Klugman, MD, FACMG, FACOG and

Newborn Screening Translational Research Network (NBSTRN) Associate Project Director Amy Brower, PhD contributed articles to the campaign. In the article “[Prenatal Genetic Tests Offer New Noninvasive](#)

[Options](#),” Dr. Klugman describes the types of genetic analyses that can be done during pregnancy including noninvasive genetic screening, amniocentesis and chorionic villus sampling. In the article “[Fifty Years of Precision Medicine: Newborn Screening in the United States](#),” Dr. Brower discusses newborn screening in the US, the role of genetics and genomics in the development of new treatments and the importance of translational research.

The printed supplement was included in 500,000 copies of *USA Today*. The online supplement is expected to reach 600,000 people.

Drs. Brynn Levy and David Tilstra Are Elected to the Board of Directors of the ACMG Foundation for Genetic and Genomic Medicine



In September, the ACMG Foundation for Genetic and Genomic Medicine announced the election of Brynn Levy, MSc. (Med), PhD, FACMG and David Tilstra, MD, MBA, CPE to its board of directors. The ACMG Foundation is a national nonprofit foundation dedicated to facilitating the integration of genetics and genomics into medical practice.

“I am very pleased to welcome Drs. Levy and Tilstra to the ACMG Foundation Board of Directors. Both are seasoned and highly experienced geneticists who will bring important perspectives to help the Foundation fulfill its mission in support of activities of the College,” said ACMG Foundation President Bruce R. Korf, MD, PhD, FACMG.

[READ MORE](#)

We Need You! Complete the Medical Geneticist Workforce Survey Today

The banner features a yellow and black diagonal striped border. On the left, there are three logos: ABMGG (American Board of Medical Genetics and Genomics), ACMG (American College of Medical Genetics and Genomics), and NCC (National Coordinating Center for the Regional Genetics Networks). To the right of the logos, the text reads: "ARE YOU A BOARD CERTIFIED MEDICAL GENETICIST IN THE US?" followed by "Complete the 'Current Conditions in Medical Genetics Practice' Survey distributed by ABMGG today!"

ACMG, via the [National Coordinating Center for the Regional Genetics Networks \(NCC\)](#), in partnership with the American Board of Medical Genetics and Genomics (ABMGG), has developed a Current Practice in Medical Genetics Survey to assess the clinical and broader medical

genetics workforce in the United States. The information gathered from the survey will help provide information to the U.S. Government Accountability Office (GAO), which is [tasked](#) by the U.S. House of Representatives to conduct “a nationwide analysis of the medical genetics workforce.”

The survey, which is being distributed by the ABMGG, is seeking responses from board-certified, US-based medical geneticists. The genetic counselors workforce is being assessed via the National Society of Genetic Counselors. ACMG and NCC are currently working with metabolic dietitians and genetic nurses to obtain workforce information from these providers.

If you believe you should have received the survey but did not, please contact abmagg@abmagg.org. If you have questions about the development of the survey or how the data will be handled, please contact NCC Associate Program Director Alisha Keehn (akeehn@acmg.net) or NCC Project Manager Megan Lyon (mlyon@acmg.net).

Visit the ACMG and NCC Booths at the ASHG Meeting

Are you planning to attend the 2019 ASHG Annual Meeting in Houston, Texas next week? Stop by the ACMG booth (#322) to learn about ACMG activities, resources, education, events and membership discounts. Enter our drawings for an Apple iPad Mini and 2020 ACMG Annual Meeting registration.

After you visit the ACMG booth, be sure to stop by the NCC booth (#320), which will be right next door. The National Coordinating Center for the Regional Genetics Networks (NCC) will be sharing tools and resources developed by NCC, the RGNs, and the Family Center that help improve access to genetic services for medically underserved populations. Some highlighted resources will include the ACMG ACT Sheets, telegenetics training offered by each RGN, the MSRGN Development Delay Algorithm, the NCC State Medicaid Genetics Policies Database, and much more!

ACMG Welcomes 13 New Members

ACMG welcomes and congratulates its 13 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.

Associate Member

Aparna Murali, MS, MA, CGC

Affiliate Members

Karen Cadoo, MD
Lori S. Farmer, MS, MSN
Stacy W. Gray, MD
Tina Han, PhD, MGC
Elizabeth L. Lucas, MD, DMD
Mohammed A. Mohammed, MS

Trainee Members

Ari Morgenthau, MD, MS
Bianca Zapanta, DO

Student Members

Michael A. Finkel, MPH
Michael Mackley, PhD
Anna Nagy, BA
Genesys A. Santana, MA

Do you know someone who should join ACMG? Please let them know they can join online at: <https://www.acmg.net/join>.

It's ACMG Membership Renewal Time: 2020 Membership Renewal Deadline Is January 31, 2020



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, you can download a 2020 Dues Renewal Form [here](#). Renewal by phone is also available at 301-718-9603.

In order to avoid interruption of membership services and benefits, renew your membership **by January 31, 2020**. Membership payment must be current in order 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.

Don't Forget! *Genetics in Medicine*, ACMG's official journal, is going green. Online access and free downloads will remain the same for members, but if you wish to continue receiving a print copy of the journal you will need to add the print subscription cost when submitting your renewal payment.

Questions regarding membership renewal can be sent to membership@acmg.net.

Call for Nominations: ACMG Foundation David L. Rimoin Lifetime Achievement Award in Medical Genetics



In case you missed it, the ACMG Foundation for Genetic and Genomic Medicine is accepting nominations for its most prestigious award, the **David L. Rimoin Lifetime Achievement Award in Medical Genetics**. This award is given in memory of genetics pioneer, founding president of the ACMG and past president of the ACMG Foundation Dr. David L. Rimoin.

The award is presented to an individual in the medical genetics profession who exemplifies a lifetime of achievement and the personal qualities embodied by Dr. Rimoin including his passion for teaching, his care for patients and their families and his enthusiasm for integrating medical genetics and genomics into mainstream healthcare. As a skilled physician, admired teacher and mentor, prolific author, accomplished researcher, beloved friend and visionary, Dr. Rimoin touched the lives of generations of patients, trainees and colleagues.

For more information about Dr. Rimoin and his legacy, [click here](#). To nominate someone for the Rimoin Lifetime Achievement Award, [click here](#). **The deadline to submit nominations is Friday, October 25, 2019.**

To learn more about the ACMG Foundation for Genetic and Genomic Medicine, visit www.acmgfoundation.org.

The ACMG Foundation for Genetic and Genomic Medicine to Open Nominations for the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award



The ACMG Foundation for Genetic and Genomic Medicine will begin accepting nominations for a new award, the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award, on **Friday, October 11, 2019**. This award honors Dr. Watson for his nearly 20 years of transformational leadership of ACMG, the ACMG Foundation and the field of medical genetics and genomics.

The Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award was established to acknowledge individuals who have demonstrated innovation in their work and developed or implemented a new concept, method or idea that has had significant impact in genetic and genomic medicine. The deadline to submit nominations is **Friday, November 22, 2019**. To learn more about the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award, [click here](#).

The ACMG Foundation for Genetic and Genomic Medicine has established a fund to support this award. To donate to the fund, [click here](#) and be sure to designate your charitable gift to the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award.

Join the Conversation: Follow ACMG on Social Media

ACMG enjoys an active following on social media. With ACMG pages on [Twitter](#), [Facebook](#), [Instagram](#) and [YouTube](#), and an ACMG Annual Clinical Genetics Meeting page on [LinkedIn](#), social media users are kept up to date on ACMG's work to support members in their practices and lead the integration of genetics and genomics into all of medicine. In addition, our private, members-only group on [LinkedIn](#) affords ACMG the opportunity to share member-exclusive news including the availability of ACMG documents for member comment, early announcements of new ACMG statements and guidelines and opportunities for member engagement. We encourage you to follow ACMG on social media and join the conversation. Here are a few recent ACMG social posts showing the wide variety of news and information we share on social media.



ACMG - American College of Medical Genetics and Genomics

Working together, advocating for medical genetics: ACMG and ASHG are partnering on efforts to help members of the 116th Congress understand the value of genetics and genomics research, its translation to clinical applications that benefit patients, and how clinical findings inform further research.

Today, ACMG and @GeneticsSociety held a joint ACMG/ASHG Congressional Briefing on "The Undiagnosed Diseases Network (UDN): The Interface of Research and Clinical Care to Solve Medical Mysteries." Speakers Gail Jarvik, MD, PhD, FACMG; Bill Gahl, MD, PhD, FACMG; John Phillips, III, MD, FACMG; and Danny Miller, father of two children diagnosed through the UDN, described @udnconnect, how it uses genomic sequencing to solve medical mysteries, and what @udnconnect means to the healthcare system. Special thanks to Representative @DaveLoebsack for assisting with the briefing. #genetics #genomics #medicalgenetics #medicalgenomics #clinicalgenetics #clinicalgenomics #precisionmedicine #medicaleducation #GCCat



In appreciation of

Michael S. Watson, MS, PhD, FACMG,
for your immeasurable contributions
and dedication to ACMG since 2000.



Thank you
for your hard work and
extraordinary commitment
to building and growing
ACMG into what it is today.

www.acmg.net



theacmg

theacmg Thank you to Michael S. Watson, PhD, FACMG, ACMG's longstanding executive director, who will depart ACMG at the end of 2019.

The College conveys enduring gratitude for Mike's immeasurable contributions to the ACMG, the ACMG Foundation for Genetic and Genomic Medicine and the grants he has managed while with ACMG. His hard work and commitment have made ACMG the organization that it is today and he leaves the College in a strong position to face the exciting challenges and growth of the field ahead.

ACMG President Anthony R. Gregg.



Add a comment...

Post



ACMG is proud to join other provider and patient organizations in urging Congress to pass the Newborn Screening Saves Lives Reauthorization Act. [#nbsslra](#)
Current authorizations expire on Sept 30, and the time for Congress to act is now! [#nbs2019](#)

**MORE THAN 12,000 BABIES ARE
COUNTING ON YOU.**

Lifesaving newborn screening programs expire
September 30. Now is the time for Congress to act.



THE NEWBORN SCREENING SAVES LIVES REAUTHORIZATION ACT (H.R. 2607/S. 2168) will renew critical federal programs that support our nation's newborn screening system. The bill is sponsored by Reps. Lucille Roybal-Allard, Mike Simpson, Katherine Clark and Jaime Herrera Beutler and Sens. Maggie Hassan and Cory Gardner.

[#NewbornScreeningSavesLives](#)





ACMG

Are you a student interested in medical genetics? Start the school year right —join ACMG, the nation’s only membership organization representing the entire medical genetics team. Student membership is free! bit.ly/2MwQi9c #genetics #genomics #medicaleducation #GCChat

Are you a grad, med, genetic counseling, allied health or undergrad student interested in medical genetics?



Join ACMG today!
Student membership is free.
www.acmg.net



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The ACMG Foundation has established the Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award in honor of Dr. Watson’s nearly 20 years of transformational leadership of ACMG, the #ACMGFoundation and the field of medical genetics and genomics. bit.ly/2kt9TZ



ACMG Foundation for Genetic and Genomic Medicine
Better Health Through Genetics™

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The ACMG Foundation establishes the **Dr. Michael S. Watson Genetic and Genomic Medicine Innovation Award** in honor of Dr. Watson’s transformational leadership of ACMG, the ACMG Foundation and the field of medical genetics and genomics.

Learn more about this award at www.acmgfoundation.org



Artificial Intelligence: NBSTRN Facilitates Research in Rare Genetic Disease and Genomics



NBSTRN
Newborn Screening
Translational Research
Network

A five-year effort to explore the use of genomic sequencing in the neonatal period concluded this fall. As findings emerge from the Newborn Sequencing In Genomic medicine and public Health (NSIGHT) cohorts, we are learning that the incidence of

rare genetic disorders among critically ill infants may be greater than previously believed. To capitalize on these findings, ACMG’s Newborn Screening Translational Research Network (NBSTRN) Researcher Needs Workgroup brought together 80 experts to discuss the use of genomics to speed the diagnosis and inform the care of newborns in high-acuity intensive care units. Although the cost of genome sequencing has significantly decreased and the speed of sequencing analysis has exponentially increased in recent years, reservations are still evident for the implementation of sequencing into screening and genetics-based clinical diagnoses. The concerns stem from the overwhelming volume of genomic data for the clinical decision-making process that may exhaust

the current limitations of the healthcare system, unless an automated, streamlined system approach can be implemented. Thus, the focus of the meeting was to outline a research study exploring the use of next generation sequencing (NGS) in the neonatal intensive care unit (NICU) and to explore the use of NGS data across the lifespan. A key component highlighted by attendees was the incorporation of artificial intelligence (AI) to build a comprehensive knowledge base of rare disease using genomic and phenotypic data. AI could reduce costs and expedite genome analysis, while also applying variant discovery techniques uniformly across the patient population. Current challenges in the implementation of AI systems include the interoperability and governance policies of genomic data, scalability of large computational processes and natural language processing abilities. AI researchers from [Rady Children's Institute for Genomic Medicine](#), the [USTAR Center for Genetic Discovery](#) and [MarthLab](#) at the University of Utah, and [Foundation Twenty-nine](#) presented current efforts to overcome these challenges. NBSTRN will continue to facilitate these discussions and welcomes your involvement. Visit www.nbstrn.org to learn more.

NBSTRN is a key component of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) Hunter Kelly Newborn Screening Research Program and develops tools and resources to support investigators engaged in newborn screening-related research. This project has been funded in whole or in part with Federal funds from the NICHD, National Institutes of Health, Department of Health and Human Services, under Contract No. HHSN275201800005C.

Genetics in Medicine Is Going Green: Sign Up for Free E-TOC Alerts Today

In case you missed the recent announcement from *Genetics in Medicine (GIM)*, the official journal of the ACMG will be moving to an online, digital environment in 2020. As a result of this transition, ACMG members will only receive an annual print subscription to *GIM* if they opt in to do so. **An online *GIM* subscription will continue to be included with your ACMG membership.**

For those members who still prefer a print copy of the journal, *GIM* will offer an inexpensive printed hard copy subscription for just \$72 annually. The option for a monthly print copy will be included on your 2020 ACMG membership renewal notice. For those wishing to continue to receive a print journal, we encourage membership renewal by January 31, 2020 to allow uninterrupted delivery of your print copies.

The full text of all articles and free downloads will continue to be available to all members through the "Access *Genetics in Medicine Online*" button on the [ACMG home page](#).

Members are encouraged to sign up to receive monthly electronic Table of Contents (e-TOC) notifications, which alert you when the latest issue of *GIM* has been published. Your e-TOC email will include the full Table of Contents of the latest issue, complete with links to every article in the issue.

To set up your e-TOC delivery, [click here](#).

For further instructions on how to sign up, [read more](#).

Mark Your Calendar for the Next ACMG Genomics Case Conference



Wednesday, October 23, 2019, 2:00 PM–3:00 PM EST

This month's engaging topic is "Comprehensive Genetic Testing Informs Diagnosis and Facilitates Management in Patients with Kidney Diseases" and is hosted by experts from the Carver College of Medicine and Iowa Institute of Human Genetics.

Registration opening soon!

Call for Cases: ACMG Genomics Case Conferences



Do you have a challenging and interesting case that you would like to present at an ACMG Genomics Case Conference?

The ACMG Education and CME Committee invites you to present at our Genomics Case Conference webinars held every month from 2:00 PM to 3:00 PM EST.

We are particularly interested in cases that fall into the following categories:

- Reclassification of variants of uncertain significance
- Long-read sequencing and analysis
- Whole genome sequencing
- Microarray regions of homozygosity
- New intellectual disability syndromes
- Solid tumor next generation sequencing
- Pharmacogenomics
- Transcriptome and epigenome testing
- NGS in newborn screening
- Mitochondrial DNA variant interpretation and classification

Submission details:

- The primary focus of these live case conferences is the adaptation of exome or genome sequencing technology in clinical care.
- All cases must have a laboratorian, clinician and genetic counselor.
- Format: 60-minute presentation.

Send inquiries to Dr. John Bernat at john-bernat@uiowa.edu or to education@acmg.net.

2020 ACMG Annual Clinical Genetics Meeting Update



Plan now to attend the 2020 ACMG Annual Clinical Genetics Meeting March 17–21 in San Antonio—a new destination for the ACMG Annual Meeting. A vibrant city with a lot of local history, unique food, global culture and amazing entertainment around every corner, there's always something worth celebrating in San Antonio! Visit www.acmgmeeting.net for more information about what to do in San Antonio.

REGISTRATION, HOUSING AND ONLINE ABSTRACT SUBMISSION NOW OPEN

Visit www.acmgmeeting.net to register for the meeting and to submit your abstract. Be sure to register early and book your hotel at the same time.

Important Dates:

Online Abstract Submission Deadline: November 5, 2019, 11:59 PM PST

Early Bird Registration Deadline: December 13, 2019

Advance Registration Deadline: February 14, 2020

Late Registration Fees Apply: February 15, 2020

Hotel Reservation Deadline: February 20, 2020

Call for 2020 Mentors

The Trainee/Mentor Luncheon held during the ACMG Annual Meeting provides an opportunity for trainees (graduate students, postdoctoral fellows and clinical and laboratory trainees) in the field of medical genetics to meet and talk informally with junior and senior members of the College about career options, goals and professional opportunities. If you are an active member of the College, plan to attend the Annual Meeting in 2020 and are interested in being a mentor, we would like to hear from you. There are a limited number of spots available and we are looking for both junior and senior level members. Compensation includes our gratitude and a free lunch.

If you are interested, please provide information by [clicking here](#). **The deadline to submit your name is January 27, 2020.** We will have a final selection of volunteers on February 7, 2020.

Do You Have a Puzzling Case? Submit It as a Diagnostic Challenge

The Diagnostic Dilemmas and Challenges sessions at the ACMG Annual Clinical Genetics Meeting are interactive sessions that allow genetics professionals to present cases of rare knowns and unknowns and to share best practices. If you have a puzzling case, we invite you to submit it as a Diagnostic Dilemma or Challenge for the 2020 Annual Meeting.

We are particularly interested in cases that fall into the following categories:

- Prenatal Diagnostic Challenges
- Laboratory Diagnostic Challenges (Constitutional and Neoplastic Cases in Molecular, Cytogenomic, and Biochemical Genetics Specialties)
- Pediatric Diagnostic Dilemmas (Rare Knowns and Unknowns)
- Adult & Cancer Diagnostic Dilemmas

To learn more or to submit a case, [click here](#).

NEW FOR 2020 Sustainability Efforts

ACMG staff, vendors and the meeting facilities have been working toward making the ACMG Annual Meeting a sustainable meeting each year for the last several years. Environmental considerations are incorporated throughout all stages of the meeting to minimize a negative impact on the environment. We will move closer to becoming a green meeting in 2020 with the addition of reusable water bottles, fewer printed materials and the elimination of tote bags.

Startup Pavilion in the Exhibit Hall

The all-new Startup Pavilion will be a dedicated area for new market entrants looking to take their businesses to the next level at the ACMG Meeting. This will be a top destination for attendees interested in discovering the latest and newest products and services in the genetics and genomics space.



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