

# ACMG In Action

Essential Updates: Member News You Can Use



August 2019

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## **Breaking News – ACMG Announces New Evidence-Based Guidelines Program: Will Accelerate Review of Old Guidelines and Development of New Ones**



ACMG Foundation for  
Genetic and Genomic Medicine

Better Health Through Genetics™

[www.acmgfoundation.org](http://www.acmgfoundation.org)



**ACMG establishes Evidence-Based Guidelines Program to develop unbiased guidelines in medical genetics & genomics.**

"Development of impartial evidence-based guidelines in genetic and genomic medicine is one of the most powerful ways in which ACMG can advance both the field of medical genetics and our members' ability to prevent, diagnose and treat disease. This new EBG initiative will greatly accelerate this important mission." - ACMG Foundation President Bruce R. Korf, MD, PhD, FACMG

Since 1991, ACMG has regularly set genetics and genomics standards for the medical field by publishing unbiased guidelines for laboratory and clinical geneticists around the world. On August 8, 2019, ACMG announced that it has established a new Evidence-Based Guidelines (EBG) Program that will develop new evidence-based guidelines and also formally review and update existing guidelines. This important new program, including a dedicated staff, addresses a critical need for unbiased evidence-based guidelines. The guidelines developed through this program will help clinicians make decisions regarding the use of genetic and genomic testing and help both government and private health insurers determine coverage options for tests and treatments. The ACMG's fundraising arm, the ACMG Foundation for Genetic and Genomic Medicine, has established a fund to support the new EBG Program. Illumina provided initial funding for the program and additional funding is being sought.

"Development of impartial evidence-based guidelines in genetic and genomic medicine is one of the most powerful ways in which ACMG can advance both the field of medical genetics and our members' ability to prevent, diagnose and treat disease. This new EBG initiative will greatly accelerate this important mission," said Bruce R. Korf, MD, PhD, FACMG, president of the ACMG Foundation.

## Upcoming Events

### 2020 ACMG Annual Clinical Genetics Meeting

March 17-21, 2020

Henry B. Gonzalez Convention Center

San Antonio, Texas

## Website Links

[ACMG](#)

[ACMG Foundation](#)

[ACMG Meetings Website](#)

[ACMG Education Center](#)

[NBSTRN](#)

[NCC](#)

[ClinGen](#)

[GIM](#)

Read the news release about the program [here](#).

## We've Streamlined Your Access to *Genetics in Medicine* Online



*Genetics in Medicine*, the official journal of the American College of Medical Genetics and Genomics, can now be accessed directly from the [ACMG home page](#) by clicking on the new “Access *Genetics in Medicine* Online” [button](#). Enter your ACMG website login credentials when prompted and you will automatically be redirected to the full access version of *GIM* online.

Can't remember your ACMG website login credentials? Your username name is preset to your ACMG preferred email. If you need to change your email address or password, select the Forgot Password link and follow the prompts to reset and receive an email confirming your changes.

## August Opportunity from the Lab QA Committee! Participate in the Review of Our New Peroxisomal Disorders Testing Standard

The ACMG Board of Directors requests your comments on a new draft document, developed under the auspices of the College's Laboratory Quality Assurance Committee. This is a members-only opportunity, which will end on **Friday August 30, 2019 at 11:59 PM PT**. Please join your colleagues in reviewing the following manuscript:

### Laboratory Diagnosis of Disorders of Peroxisomal Biogenesis and Function: A Technical Standard of the American College of Medical Genetics and Genomics (ACMG)

The addition of your 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!

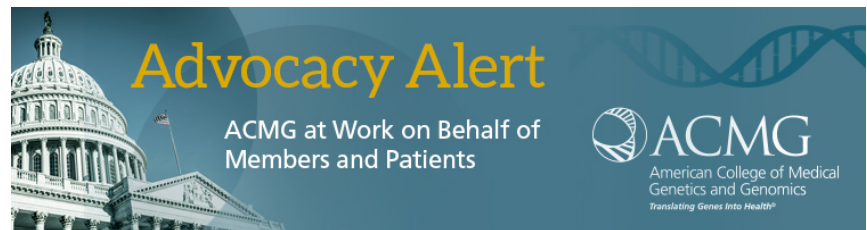
[Read more](#)

### August 2019 *GenePod*: Creating a Framework to Assess Resource Needs for Genetic Services



As the appetite for clinical genetic testing grows, it seems the demand for geneticists able to provide the needed services outpaces the supply. Yet health services do not have a way to measure that growth or the resources needed to provide those services. In a [recent study](#) published in *Genetics in Medicine* (*GIM*), researchers describe a new framework for evaluating the resource needs of clinical genetics services, including recent huge increases in genetic testing and the accompanying counseling. In the August episode of *GenePod*, *GIM*'s monthly podcast, host Cynthia Graber talks with lead study author Andrew Fennell about the new framework, its current limitations and how clinicians can use it now to plan for the future.

### ACMG and ASHG Presidents Ask Congress to Protect Existing Policies That Prevent Patenting of Genetic Information



In case you missed it, earlier this year, US Senators Coons (D-DE) and Tillis (R-NC) and Representatives Collins (R-GA), Johnson (D-GA) and Stivers (R-OH) released draft [legislation](#) to amend Section 101 of the Patent Act. The proposed amendments would reverse current policies that prevent patenting of genetic information and its association with disease. Allowing such patents to occur would create significant barriers to genetics and genomics research and its application to clinical care. Additionally, it would result in increased costs of and reduced access to genetic testing.

To express strong concern over the draft bill, the current and past presidents of ACMG and ASHG joined together to send an urgent letter to Congress, highlighting the importance of innovation in clinical care and research and asking that Congress affirm and protect existing policies that encourage sharing of emerging fundamental genetic knowledge. The full letter can be viewed [here](#).

ACMG, together with ASHG and other organizations with our shared concerns, will continue to work with Congress to encourage development

of policies that enhance patient care by advancing medical innovation and promoting sharing of scientific knowledge.

## ACMG Reaches Millions on Social Media

The communications team at ACMG uses a variety of communication strategies to share the latest advances in medical genetics and genomics and connect with individuals committed to translating genetic discoveries into better patient care and public health. Social media is one part of this overall communications plan.

In 2018, ACMG's posts on [Twitter](#), [Facebook](#) and [YouTube](#) earned nearly 2 million impressions. So far in 2019, posts about Medical Genetics Awareness Week have earned more than three million impressions and posts about the 2019 ACMG Annual Meeting have earned nearly 11 million impressions.

On [LinkedIn](#), ACMG also has an [annual meeting group](#) page for meeting attendees and a [private, members-only group](#) page through which we share content of special interest to members. This spring, we added [Instagram](#) to our social media portfolio, which will help ACMG reach a valuable younger demographic.

We encourage you to follow ACMG across our social media channels and join the conversation. Here are a few recent ACMG social posts showing the broad range of topics discussed on our social media channels, ranging from important advocacy news to educational offerings and ways we are working to attract students to medical genetics.



**Advocacy Alert**  
ACMG at Work on Behalf of Members and Patients  
ACMG  
American College of Medical Genetics and Genomics  
Preventing Disease Into Health

ACMG and ASHG presidents voice strong concern over draft bill to amend the Patent Act and permit patenting of genetic information.

"Congress should affirm and protect existing policies that encourage sharing of emerging fundamental genetic knowledge."

Follow the link in our bio to read the letter.

theacmg

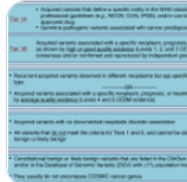
theacmg Current and past presidents of the ACMG and @geneticsociety have joined together to send an urgent letter to Congress voicing strong concern over proposed amendments to the Patent Act that would permit patenting of genetic information stating that the proposed amendments would increase the cost of genetic testing, reduce access to genetic testing, and stymie genetics and genomics research and the development of novel medical applications. "Congress should affirm and protect existing policies that encourage sharing of emerging fundamental genetic knowledge." Read the full letter at <http://bit.ly/2LOwsWy> #medicalgenetics #clinicalgenetics #precisionmedicine #nenenatents



ACMG  
@TheACMG



A new joint consensus recommendation from the ACMG and @CG\_Consortium addresses an urgent need to standardize the interpretation and reporting of acquired CNAs and CN-LOH in neoplastic disorders. Read this technical laboratory standard at



Technical laboratory standards for interpretation and reporting of acquired copy-number abnormalities and copy-neutral loss of heterozygosity (CN-LOH) in neoplastic disorders. [nature.com](https://doi.org/10.1038/ng.395)

9:18 AM · Jul 17, 2019 · Twitter Web App



ACMG Foundation for  
Genetic and Genomic Medicine  
Better Health Through Genetics™  
[www.acmgfoundation.org](http://www.acmgfoundation.org)



Each year, the **ACMG Foundation Summer Genetics Scholars Program** engages medical students in the exciting field of medical genetics & genomics.

In 2019, 10 students join geneticist mentors for six-week experiences in clinical & lab genetics.

Learn more at [www.acmgfoundation.org](http://www.acmgfoundation.org)



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theacmg Each year, the #ACMGFoundation Summer Genetics Scholars Program engages medical students in the exciting field of medical genetics & genomics. In 2019, 10 students join geneticist mentors for 6-week experiences in clinical & lab genetics <http://bit.ly/2OmXVAS> #medicaleducation #medicalstudent #medicalstudents #medicalgeneticsawareness #medicalgenetics







Studying for ABMGG or ABGC board exams? Or just looking for continuing education credits in medical [#genetics](#) & [#genomics](#)? The 2019 ACMG Genetics & Genomics Review Course OnDemand is now available for purchase in the ACMG Genetics Academy <http://bit.ly/2GgMqnc> [#medicaleducation](#)



The 2019 ACMG Genetics and Genomics Review Course OnDemand is now available at [www.acmgeducation.net](http://www.acmgeducation.net)

### ACMG Issues Addendum to Joint Practice Guideline on Genetic Counseling and Testing for Alzheimer Disease

The 2011 ACMG-National Society of Genetic Counselors (NSGC) joint practice guidelines entitled [“Genetic counseling and testing for Alzheimer disease: Joint practice guidelines of the American College of Medical Genetics and the National Society of Genetic Counselors”](#) was reaffirmed by the ACMG Board of Directors as of June 25, 2018 with the following suggested considerations:

1. To use the phrase “pathogenic variant” rather than the word “mutation” in discussing pathogenic variants related to autosomal dominant early-onset Alzheimer disease. This would be consistent with current ACMG/AMP Guidelines for Variant Interpretation and Reporting.
2. Because this document no longer meets the criteria for an evidence-based practice guideline by either the American College of Medical Genetics and Genomics (ACMG) or National Society of Genetic Counselors (NSGC), NSGC reclassified this document as a Practice Resource in 2016, and ACMG is also classifying it as a Practice Resource as of this reaffirmation.

An [addendum](#) was published online by *Genetics in Medicine* on June 19, 2019.

The College extends its appreciation to Jill S. Goldman MS, MPhil, Susan E. Hahn MS, Jennifer Williamson Catania MS, MPH, Susan LaRusse-Eckert MS, Melissa Barber Butson ScM, Malia Rumbaugh MS, Michelle N. Strecker MS, J. Scott Roberts PhD, Wylie Burke MD, PhD, Richard Mayeux MD, MSc and Thomas Bird MD.

## **The ACMG PP&G and NSGC Practice Guidelines Committees Issue Addendum to Guideline on Referral Indications for Cancer Predisposition Assessment**

The ACMG Professional Practice and Guidelines (PP&G) Committee and the National Society of Genetic Counselors (NSGC) Practice Guidelines Committee reviewed and reaffirmed the 2014 practice guideline entitled “[A practice guideline from the American College of Medical Genetics and Genomics and the National Society of Genetic Counselors: referral indications for cancer predisposition assessment](#)” with the following comment:

The committees feel that this document remains a useful guide in assessing indications for cancer referral; however, it should not be considered comprehensive nor utilized to limit access to genetics professionals. While the principles outlined for genetics referral for the specific tumors and syndromes listed remain valid, in many cases the indications for referral have expanded. The field of cancer genetics is rapidly evolving, including frequent discovery of additional genes and new clinical presentations, expanded gene panel testing, paired tumor and germline sequencing, and expanded utility of molecular testing in treatment planning. These changes have impacted referral considerations outlined in this document. We encourage clinicians to consult additional updated sources in making final decisions regarding referral. These include more recent versions of the [National Comprehensive Cancer Network guidelines](#) and [GeneReviews](#). Additionally, we recognize increasing need for all health care providers to collaborate to meet the clinical care needs of individuals known or suspected to be impacted by a hereditary cancer condition, and the evolving role of non-genetics professionals in this area.

In accordance with current policy, this document is now classified as a Practice Resource by the NSGC.

An [addendum](#) was published online by *Genetics in Medicine* on July 23, 2019.

The College extends its appreciation to Michael T. Bashford MD, Wendy Kohlman MS, Jessica Everett MS, Ashley Parrott MS, Toni I. Pollin MS, PhD for the Practice Guidelines Committee of the National Society of Genetic Counselors and the Professional Practice and Guidelines Committee of the American College of Medical Genetics and Genomics.

## **ACMG Welcomes Twelve New Members**

ACMG welcomes and congratulates its 12 new and newly certified members. There has never been a more exciting time to be 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.

**ACMG Fellows**

Yazmin Enchautegui Colon, MD, FACMG

Christina T. Lam, MD, FACMG

Pawel Pomianowski, MD, FACMG

**Corresponding Fellow**

Ruqaiyah S. Altassan, MD, FACMG

**Candidate Fellows**

Nicole Boczek, PhD

Ariel F. Martinez, PhD, MS

**Associate Members**

Michelle Q. Bosworth, MGC

Pamela Gerrol, MS

Brianna N. Gross, MGC

Meadow Heiman, MS

**Trainee Member**

Chuan Gao, PhD, MBA

**Student Member**

Yasemin Cole, MS

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

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



Nominations for the David L. Rimoin Lifetime Achievement Award in Medical Genetics are being accepted online and are due **Friday, October 25, 2019**. Please consider nominating your professional colleagues and friends for this distinguished honor. To learn more about Dr. Rimoin's remarkable legacy, [click here](#). For additional information about the award and selection criteria please review the [David L. Rimoin Lifetime Achievement Award in Medical Genetics application](#).

**Genetics in Medicine Partners with Preprint Servers, Enabling**



## Authors to Directly Transmit Preprint Files to *GIM*'s Editorial Manager

*Genetics in Medicine* (*GIM*), the official journal of the ACMG, has partnered with the preprint servers [bioRxiv](#) and [medRxiv](#) to provide authors the option to directly transfer manuscripts to the journal.

bioRxiv and medRxiv are repositories for preliminary research in the life sciences and the medical, clinical and related health sciences fields, respectively. These repositories allow authors to share their work with colleagues—to receive comments and feedback—prior to the essential peer review that is carried out by a journal such as *Genetics in Medicine*.

These partnerships will allow authors to directly transmit their preprint manuscript files and metadata, such as author information, to *GIM*'s submission system, Editorial Manager. If articles are ultimately published in the journal, the preprint manuscript will be linked to the published article.

## 2019 Meetings OnDemand Are Now Available in the ACMG Genetics Academy!



### 2019 ACMG Genetics and Genomics Review Course

Earn up to 21.5 CME, P.A.C.E.®, MOC Part II and NSGC Credits with the 2019 ACMG Genetics and Genomics Review Course OnDemand. This well-attended course covers a wide range of topics including genetic transmission, biochemical genetics, clinical genetics, neurogenetics, molecular genetics, genetic counseling, cancer genetics, cytogenetics, prenatal/reproductive genetics, Mendelian genetics, developmental genetics, and genomic medicine.

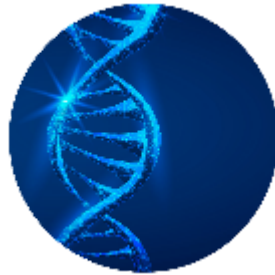
### Specialty Training Course in Cytogenetics and Molecular Genetics and Genomics OnDemand

This course features 19 presentations split between basic principles in cytogenetics and genomics and molecular genetics and genomics. The in-person course was held at the Center for Advanced Medical Learning and Simulation (CAMLS) in Tampa, Florida June 30–July 1, 2019. **This OnDemand course does not offer educational credits and does not meet the requirements associated with ABMGG LGG certification.** We will be launching this course in the future with CME, P.A.C.E.® credit offered.

## 2019 ACMG Annual Clinical Genetics Meeting OnDemand

*Earn up to 45.75 CME and P.A.C.E.® Credits, and 44 NSGC Credits with the ACMG Annual Clinical Genetics Meeting OnDemand. This OnDemand course, which includes scientific presentations from the popular ACMG Annual Meeting, offers learners the opportunity to experience the critical role of medical genetics and genomics in the practice of medicine and how genetics and genomics are being integrated into medical, clinical and laboratory practice.*

### Check Out the NEW Practice Improvement Modules for MOC Part IV



Practice Improvement Modules are derived from accepted practice guidelines related to genetics patient care. Modules are available for pediatric genetics, adult genetics and prenatal/obstetric genetic care providers.

The ACMG Maintenance of Certification Committee recently transitioned these modules from a system of scoring to one of self-reflection and peer comparison. The process for the newly designed modules is:

- I. Educational content: review module, literature and practice checklist
- II. Charting: review up to 10 patient charts and document certain findings according to the module questions
- III. Complete assessment and self-reflection questions
- IV. Evaluation
- V. You're done!

**Purchase these and other educational products in the ACMG Genetics Academy at [www.acmgeducation.net](http://www.acmgeducation.net).** If you have any questions about ACMG educational products, please contact [education@acmg.net](mailto:education@acmg.net).



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