ACMG In Action

Essential Updates: Member News You Can Use



July 2019

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Upcoming Events2020 ACMG Annual Clinical Genetics Meeting

March 17-21, 2020 Henry B. Gonzalez Convention Center San Antonio, Texas

The Spring/Summer 2019 Issue of *The ACMG Medical Geneticist* Is Now Available Online

By now, you should have received in the mail your copy of the Spring/Summer 2019 issue of *The ACMG Medical Geneticist* member newsmagazine. The Spring/Summer issue is packed with 36 pages of announcements and information including an introduction of ACMG's new president, Dr. Tony Gregg, MD, MBA, FACOG, FACMG, and six new board members, an overview of ACMG's new strategic plan, a summary of the highlights of the 2019 ACMG Annual Clinical Genetics Meeting, articles honoring the recipients of the 2019 ACMG Foundation awards, and much more.

An electronic copy of the newsmagazine is now available for download from the Members Only section of the ACMG website. To download your copy, <u>click here</u>, log in to your member account, and click on "The Medical Geneticist" in the gold menu bar.

Genetics in Medicine Receives 2018 Impact Factor of 8.683

Genetics in Medicine, the official journal of the ACMG, received an Impact Factor of 8.683 for 2018: second highest in the journal's history. Genetics in Medicine (GIM) is currently ranked 12th of 174 titles in the Genetics & Heredity category.

ACMG President Tony Gregg, MD, MBA, FACOG, FACMG said, "Genetics in Medicine is our journal. We should all be proud that GIM has achieved an impact factor rating of 8.683. We can attribute our successful impact factor to our editor-in-chief, Bob Steiner, and before him, Jim Evans. Now is the time to also acknowledge the excellent editorial board assembled by these leaders. The outstanding articles submitted by our members and colleagues help garner ACMG recognition as a worldwide source of genetics and genomics information."

GIM's Editor-in-Chief Robert D. Steiner, MD, FAAP, FACMG stated, "GIM's prominence in the field is a clear reflection of the importance of the American College of Medical Genetics and Genomics, the organization leading the medical community in incorporating genetics and genomics into patient care. Genomics and precision medicine are

Website Links

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

changing how medicine is practiced today, and ACMG along with *GIM* are at the forefront of this remarkable transformation."

The Impact Factor is an objective measure of the world's leading journals, based on articles' cited references and is oft considered a measure of a journal's impact, overall successful performance and relevance to its field.

<u>Click here</u> to read the full ACMG news release about *GIM*'s 2018 Impact Factor.

ACMG July Advocacy Updates



Update on the Newborn Screening Saves Lives Reauthorization Act

On June 25th, the House Energy & Commerce Subcommittee on Health held a hearing on four public health reauthorization bills, including H.R. 2507, the Newborn Screening Saves Lives Reauthorization Act (NBSSLRA) of 2019. Joseph Bocchini, MD, the immediate past chair of the Advisory Committee on Heritable Disorders in Newborns and Children, testified on the importance of this legislation, which was well received by the US representatives present at the hearing. The legislation has cleared the subcommittee and is scheduled for markup by the full Energy and Commerce Committee on July 17th 2019. We anticipate introduction of a similar bill in the Senate in the near future, possibly this week. ACMG continues to work with Congress to advocate for the NBSSLRA as it moves through the legislative process.

ACMG's members have also been supporting the NBSSLRA by submitting letters to their elected officials requesting that they cosponsor the bill. To date, your letters have reached 71 different members of the US House of Representatives! Thank you to everyone who has submitted a letter. Your letters are incredibly important and help encourage representatives to support this bill as it moves forward. For those who have not submitted a letter yet, it's not too late. Simply follow this link, enter in your name and address, and a letter will be automatically sent to your representative.

ACMG Presents Recommendations at the CMS CLFS Annual Public Meeting

Earlier this year, ACMG was successful in getting two new Tier 1 Current Procedural Terminology (CPT®) codes for *PALB2* accepted by the CPT® Editorial Panel (for more information on this effort, <u>click here</u>). The two new codes, 813X1 (full gene sequence) and 813X2 (known familial variants), have now been placed on the Clinical Laboratory Fee Schedule (CLFS) for payment amount determination by the Centers for Medicare &

Medicaid Services (CMS) using either the gapfill or crosswalk method. On June 24th, ACMG participated in the CMS CLFS Annual Public Meeting in Baltimore, MD. With the help of ACMG's Economics Committee and other ACMG members, public recommendations were made to CMS on the payment methodology for the two new codes. CMS is expected to release their preliminary payment determinations in September and final determinations in November. The new codes and associated CMS payment amounts will go into effect beginning January 1, 2020. Thank you to ACMG member, Kathleen Hruska, PhD, FACMG, who presented on behalf of ACMG at this important meeting.

July 2019 *GenePod*: Testing Guidelines for Neurodevelopmental Disorders—Time for an Update?



Nearly a third of neurodevelopmental disorders are thought to have a genetic basis and the most recent ACMG guidelines, from 2010, recommend chromosomal microarray (CMA) and fragile X testing as first-tier genetic tests. In a <u>recent study</u> published in *Genetics in Medicine* (*GIM*), Srivastava et al. carried out a meta-analysis of 30

papers published since those guidelines were issued and determined exome sequencing had a significantly higher diagnostic yield than these previous testing standards. In this month's *GenePod*, *GIM's* monthly podcast, host Cynthia Graber talks to Mustafa Sahin, senior author of the study, about the authors' hopes that the increased diagnostic yield will lead to exome sequencing being more widely accepted as the first-line testing. The authors are also hopeful that having a molecular diagnosis might result in potentially actionable treatments for some patients.

Jonathan Sheldon, PhD Is Elected to the Board of Directors of the ACMG Foundation for Genetic and Genomic Medicine



In case you missed it, Jonathan Sheldon, PhD, senior vice president of the Bioinformatics Business Area at QIAGEN, has been elected to the board of directors of the ACMG Foundation for Genetic and Genomic Medicine. The ACMG Foundation is a national nonprofit foundation dedicated to facilitating the integration of genetics and genomics into medical practice. The board members are active participants, serving as advocates for the Foundation and for advancing its policies and programs. Dr. Sheldon was elected to a six-year term starting immediately.

"It is an honor to be elected to this prestigious board at this critical time wherein biological and translational research has enabled us to be truly close to the bedside. The ACMG organization has been at the forefront of driving efforts towards responsible, consistent and reproducible reporting of variant information. By establishing clear guidance based on their

collective expertise, they aim to provide actionable and meaningful information to physicians and patients, providing clarity that is so much needed," said Dr. Sheldon.

Read More

ACMG Welcomes Thirty-Six New Members

ACMG welcomes and congratulates its 36 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.

ACMG Fellows

Jillian G. Buchan, PhD, MS, FACMG Kathryn C. Chatfield, MD, PhD, FACMG Amanda L. Fortier, PhD, MS, FACMG Naga M. Guruju, PhD, FACMG Debra G. Leonard, MD, PhD, FACMG Robert S. Wildin, MD, FACMG Lawrence Wong, MD, FACMG

Candidate Fellows

Joseph Alaimo, PhD Bharatendu Chandra, MBBS Ryan N. Doan, PhD Jingyu Huang, PhD

Associate Members

Kari E. Branham, MGC, CGC Sandra M. Brown, MS, CGC Natalie Burrill, MGC, CGC Rebecca Crosetto, MGC, CGC Pamela Gerrol, MS, CGC Anastasia R. Richardson, MGC, CGC Natasha Rudy, ScM, CGC

Affiliate Members

Tero-Pekka Alastalo, MD, PhD Brian C. Mansfield, PhD Jesse Walsh, PhD Stephan Zuchner, MD

Trainee Members

Allison J. Cox, PhD, MS Avinash V. Dharmadhikari, PhD, MS Lauren Pronman, DO Joy Tanaka, PhD

Students

Esteban Astiazaran Symonds, MD Ana C. Jara Ettinger, MD Kevin S. Landau, MS Kaitlin Winston O'Connell, BS Maggie Ruderman, MGC Sadaf Saba, MS Jason Allen Shandler, AS Jenna Faith Stoltzfus, BS Erin Hannah Sybouts, BS Giavanna Verdi, BS

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

ACMG Call for Volunteers

Are you interested in serving on an ACMG Committee, helping with public policy initiatives or serving as a mentor? Committee volunteers are needed for the two-year term April 2020-March 2022, and volunteers for special initiatives are needed on an ad hoc basis. To respond to our Call for Volunteers, please use your member credentials to log onto the <u>ACMG Members Only webpage</u> and select the "Volunteer" option at the bottom of the benefits blocks to access our volunteer form.

Qualifications for committee service:

- Student and trainee members are encouraged to apply for service on ACMG Committees. Committee service terms are determined by the length of eligibility for these membership categories.
- Due to the technical nature of some committee projects, board certification in clinical genetics, cytogenetics, biochemical genetics, molecular genetics, medical genetics or genetic counseling may be considered in some committee appointment decisions.
- Current membership in the ACMG is required.
- Because some of the activities of committees involve working with the AMA and associated organizations, AMA membership may be considered in some committee appointment decisions.

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Mentoring in Action: ACMGF Welcomes Its Eighth Year of Summer Genetics Scholars



The ACMG Foundation for Genetic and Genomic Medicine (ACMGF) Summer Genetics Scholars Program (SGSP) welcomes its 2019 class of medical student trainees. Now in its eighth year, the SGSP continues to attract medical students interested in learning more about genetics and genomics as a possible career specialty.

This summer, the ACMGF placed 10 first-year medical students at universities and institutions across the country to participate in the competitive SGSP program. The students will spend the next six to eight weeks working closely with geneticist mentors who will share their knowledge and daily interactions with patients or in a medical laboratory as well as introduce students to various employment and networking opportunities within the medical genetics field.

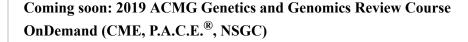
To learn more about the ACMGF Summer Genetics Scholars Program, the awards the ACMGF offers and donating to sustain these programs, please visit the ACMGF website at www.acmgfoundation.org. Questions should be directed to ACMG Foundation Manager Nicole Bell at nbell@acmg.net.

The ACMG Genetics Academy: New Content Available Now, More Coming Soon

Available Now: Printed Syllabus from the 2019 ACMG Genetics and Genomics Review Course

Preparing for the ABMGG or ABGC
Boards? The printed syllabus from the 2019
ACMG Genetics and Genomics Review Course is now available for sale in the <u>ACMG</u>
Genetics Academy. This black and white-

printed, spiral-bound syllabus consists of approximately 900 pages and includes a practice exam and content covering a broad range of genetic and genomic topics presented by recognized experts in the field. Order your copy today at www.acmgeducation.net.



The course syllabus and recorded lecture content from the 2019 ACMG Genetics and Genomics Review Course will be available for purchase soon in the <u>ACMG Genetics Academy</u>. This OnDemand format will include unlimited online and mobile access to the course's audio and video content, a practice exam and much more.

Coming soon: 2019 ACMG Foundational Specialty Training Course OnDemand (CME, P.A.C.E.®)

Did you miss the 2019 ACMG Foundational Specialty Training Course? The recorded lecture content from the course will be available for purchase soon in the <u>ACMG Genetics Academy</u>. The OnDemand format

will include unlimited online and mobile access to the course's audio and video content. *Note: the OnDemand version does not qualify as a component of formal education in the Alternative Pathway to LGG Certification.*

Available Now: 2019 ACMG Annual Meeting OnDemand (CME, P.A.C.E.[®], NSGC)

ACMG Annual Clinical Genetics Meeting
APRIL 2-6 | EXHIBIT DATES: APRIL 3-5

OnDemand Education

WASHINGTON STATE CONVENTION CENTER | SEATTLE, WA

Select sessions and short courses from the 2019 ACMG Annual Clinical Genetics Meeting are now available for purchase in the <u>ACMG Genetics Academy</u>. Your OnDemand purchase includes unlimited online and mobile access to great content such as this TED-style talk by <u>Patricia Furlong</u>. To purchase your copy of the 2019 ACMG Annual Clinical Genetics Meeting OnDemand, visit the <u>ACMG Genetics Academy</u>.











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