

## Summary of Recent Activities of The American College of Medical Genetics and Genomics

*The American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. ACMG represents over 2000 members, nearly 80% of which are board certified clinical and laboratory geneticists and genetic counselors. ACMG's mission, as redefined in the 2015 Strategic Plan, is to "develop and sustain genetic and genomic initiatives in clinical and laboratory practice, education and advocacy." Three guiding pillars underpin ACMG's activities: 1) Clinical and Laboratory Practice: Establish the paradigm of genomic medicine by issuing statements and evidence-based or expert clinical and laboratory practice guidelines and through descriptions of best practices for the delivery of genomic medicine. 2) Education: Provide education and tools for medical geneticists, other health professionals and the public and grow the genetics workforce. 3) Advocacy: Work with policymakers and payers to support the responsible application of genomics in medical practice. This report highlights key activities of the ACMG between mid-May and early-September 2016.*

### ACMG Moves to New Offices

- On August 15, ACMG opened for business in beautiful new offices located at 7101 Wisconsin Avenue, Suite 1101. Our new location is across the street and one block south of our home for the last seven years, in downtown Bethesda. Our phone, fax and email address have not changed.

### Advocacy, Policy and Practice Activities

- *ACMG Continues Activities Related to Educating Members and Policymakers About its Position on the Regulation and Oversight of Laboratory Developed Tests (LDTs):* On Tuesday September 20<sup>th</sup> ACMG will join the Infectious Diseases Society of America (IDSA) and the Association for Molecular Pathology (AMP) as co-sponsors of a *Congressional Briefing* on the basics of LDTs and the vital role they play in patient care in medical genetics, oncology and infectious disease. This briefing, "*What do Cancer Moonshot, PMI, Zika and the CARB National Action Plan All Have in Common?*" is one of many activities in which ACMG has participated since the Food and Drug Administration (FDA) released its 2014 draft guidance that sets forth a proposed framework to regulate LDTs. While FDA has stated its intent to finalize this guidance in 2016, legislative alternative regulations are also under consideration. Experts from the three sponsoring societies will highlight how LDTs are currently regulated, designed, validated, and used in a variety of settings, providing Congressional staff and other stakeholders a rich context for crafting policy around this issue. The program will be held from 3:00-4:00 PM in the Capitol Visitor Center, SVC 208-09. For more information, email Shion Chang at [schang@idsociety.org](mailto:schang@idsociety.org).

- *ACMG Efforts Lead to AMA's Support of Health Coverage for Medical Food Products:* At the June 2016 Annual Meeting of the American Medical Association (AMA) House of Delegates, ACMG submitted Resolution 122 (*Health Coverage For Nutritional Products For Inborn Errors Of Metabolism*), asking AMA to support legislation mandating insurance coverage for medical food products used to treat inborn errors of metabolism with minimal deductible or copays, and to advocate with the Department of Health and Human Services and members of Congress for the regulation of specialized nutritional products for the medical treatment of inborn errors of metabolism as drugs.

R. Rodney Howell, MD, FACMG, ACMG's Delegate to the AMA House of Delegates (HOD), "sponsored" the Resolution at the Annual Meeting of the HOD, and per the Reference Committee report, he eloquently persuaded the Delegates to support the Resolution: "The sponsor emphasized that health coverage for nutritional products for inborn errors of metabolism is essential as these medical foods can be expensive and failure to treat these conditions can result

in debilitating health conditions and even death. ...While the intent of Resolution 122 is consistent with existing policy, your Reference Committee heard compelling testimony from the sponsor of Resolution 122 to adopt more current, focused policy.”

This is the third AMA Resolution that ACMG has sponsored. Its adoption is very important to all stakeholders and can now be cited in communicating with insurers and policymakers. Notably, the AMA Resolution came just days before the US Senate authorized medical foods coverage for military families in the National Defense Authorization Act for Fiscal Year 2017 (S.2943), on Tuesday June 14; this bill must now be harmonized with the House version passed in May. ACMG will be working with the National Organization for Rare Disorders, the Society for Inherited Metabolic Disorders, the National PKU Alliance and other stakeholders who stand behind this important public health issue.

- *ACMG Tackles New Medicare NCD for Cytogenetic Tests*: Late last year, ACMG became aware that laboratories were receiving many denials from Medicare for cytogenetic tests that were previously paid by Medicare. It was determined that there had been a change in Medicare’s coverage policy. After obtaining a copy of the new Medicare National Coverage Determination (NCD), ACMG Medical Director, Dr. David Flannery, conducted a survey via the ACMG Economics ListServ to obtain data from laboratories to assess the volume of denials and their financial impact. Our findings demonstrated that labs across the country were experiencing a significant volume of denials for tests that were previously reimbursed, and that the financial impact could threaten the sustainability of these labs.

Prior to the new Medicare NCD, cytogenetic tests were covered and reimbursed under Local Coverage Determinations (LCDs) established by each of the regional Medicare Administrative Contractors (MACs). Comparison of the new NCD to the LCDs that we were able to obtain showed that the list of diagnoses covered under the NCD was much shorter than any prior LCDs for cytogenetics. It appeared likely that this truncation happened in Medicare’s process of converting ICD-9 codes from prior coverage policies into ICD-10 codes for the new NCD. A list of appropriate diagnoses that should be covered for cytogenetic testing was compiled, based on the prior LCDs. The ICD-9 codes for those diagnoses were then converted into ICD-10 codes. This list was over 450 ICD-10 codes.

Dr. Flannery and leaders from ACMG’s Economics of Genetic Services Committee wrote a detailed letter to Medicare regarding the discrepancy in the NCD versus the prior LCDs for cytogenetics and describing the impact the non-coverage was having on laboratories and ultimately on patients. The letter was accompanied by our list of diagnoses that would constitute medically appropriate indications for coverage of tests. On August 19, 2016, Medicare published “Transmittal 1708; Change Request 9751 (“Coding Revisions to National Coverage Determination (NCDs)””, which included an updated list of ICD-10 codes that would be covered for cytogenetic testing. The list was substantially longer than the original NCD list, but did not include many codes that ACMG had submitted. ACMG will continue its efforts to interact with Medicare on this important issue.

- *ACMG’s Secondary Findings Working Group*, co-chaired by Drs. David Miller and Christa Martin, has implemented an ongoing process for maintaining the list of mandatory genes that are recommended for testing when patients opt to learn about secondary findings following WES/WGS. Using a standardized, evidence-based vetting process genes will be continually added to (and perhaps removed from) the list originally dubbed “the ACMG 56”. The results of the first round of evaluating candidate genes were presented to the ACMG Board of Directors in July. One gene was removed from the original panel (*MYLK*, familial thoracic aortic aneurism and dissection), and four new genes were added (*BMPRIA* and *SMAD4*, both of which have been implicated in juvenile polyposis; *OTC*; and *ATP7B*, Wilson’s disease), leading to a new panel, “the ACMG 59”. A manuscript describing the gene nomination and vetting process and its outcome is in the final stages of preparation.

## Grant and Contract Updates

ACMG is increasingly integrating the activities of shared interest among the several national projects that it either leads or in which it participates.

- *The National Coordinating Center for the seven HRSA-funded Regional Genetic Service Collaboratives (NCC)* is developing a collaboration with the states and Regional Collaboratives to collect long-term follow-up data on their patients identified in newborn screening programs. Public health programs value data sharing that informs all such programs.

- *Curating the Clinical Genome*, the 2016 ClinGen/DECIPHER public meeting was held on the Wellcome Genome Campus, Hixon, Cambridge, UK, June 22-24, 2016 and will return to the U.S. in 2017. Collaborations between ClinGen and state newborn screening laboratories that generate and store genomic variant level and limited clinical data are also in development. States currently acquire their clinical and laboratory information on patients identified through

their public health programs, under their Public Health Authorities. Ascertainment of asymptomatic or presymptomatic individuals can provide a novel perspective on penetrance that is not available through clinically ascertained populations.

### **Genetics in Medicine Updates**

*Genetics in Medicine (GIM)*, ACMG's official journal, received a record high impact factor of 7.710 for 2015, placing it in the top 3% of all indexed journals. This is up from 7.329 in 2014, and places *GIM* thirteenth of 165 titles in the Genetics & Heredity category and at the top of genetics journals that have a primarily clinical focus.

The following ACMG documents have been published by *Genetics in Medicine* since our last report:

Mikhail FM, Heerema NA, Rao KW, Burnside RD, Cherry AM and Cooley LD; on behalf of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. **Section E6.1–6.4 of the ACMG technical standards and guidelines: chromosome studies of neoplastic blood and bone marrow–acquired chromosomal abnormalities.** *Genet Med* 18(6):635-642 (June 2016)

Cooley LD, Morton CC, Sanger WG, Saxe DF and Mikhail FM; on behalf of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. **Section E6.5–6.8 of the ACMG technical standards and guidelines: chromosome studies of lymph node and solid tumor–acquired chromosomal abnormalities.** *Genet Med* 18(6):643-648 (June 2016)

**ADDENDUM: Technical standards and guidelines for spinal muscular atrophy testing.** *Genet Med* 18(7):752 (July 2016)

**ERRATUM: Section E6.1–6.4 of the ACMG technical standards and guidelines: chromosome studies of neoplastic blood and bone marrow–acquired chromosomal abnormalities** *Genet Med* 18(8):859 (August 2016)

Gregg AR, Skotko BG, Benkendorf JL, Monaghan KG, Bajaj K, Best RG, Klugman S, and Watson MS; on behalf of the ACMG Noninvasive Prenatal Screening Work Group. **Noninvasive screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics.** *Genet Med* 18(10): (October 2016) <http://www.nature.com/gim/journal/vaop/ncurrent/pdf/gim201697a.pdf>

The following ACMG documents are in preparation:

- Laboratory and Clinical Genomic Data Sharing is Crucial to Improving Genetic Healthcare: An ACMG Position Statement
- Points to Consider in Genomic Screening of Asymptomatic Individuals
- Laboratory Guideline: Selection of Genes in a Gene Panel
- Laboratory Diagnosis of Creatine Deficiency Syndromes: A Technical Standard and Guideline of the ACMG

### **Meetings and Education Updates**

• Plans for the *2017 Annual Clinical Genetics Meeting* are progressing rapidly, with the program nearly complete. The meeting will be held March 21-25, 2017 in Phoenix, AZ, and it will include the 48th Annual March of Dimes Clinical Genetics Conference, “The Undiagnosed Diseases Network: Changing the Paradigm of Rare Disease Diagnosis, Treatment and Research”. Visit [www.acmgmeeting.net](http://www.acmgmeeting.net) for ongoing updates; meeting registration and hotel reservations open in October. The Abstract Submission site also opens in October with a submission deadline of December 2, 2016.

• *The 2017 ACMG Genetics and Genomics Review Course (GGRC)* will be held May 4-7, 2017 in Tampa, Florida. Details will be posted in October 2016 on the ACMG Genetics Education website. Designed for individuals preparing for the ABMGG certification examinations, the GGRC is also an excellent refresher course for practitioners looking to update their skills and knowledge, and those seeking medical genetics CME. Provided as a live face-to-face course and also offered in streaming live video, attendees have two ways to participate in this popular educational update.

• ACMG's live monthly *Case Conferences* resume this fall: There are two ongoing series, and all of the conferences are delivered via webinar and then archived in the On-Line E-Learning section of the ACMG Genetics Education website.

- Genomics Case Conferences will occur on the third Wednesday of each month at 2:00 PM ET (except in October due to the ASHG Meeting). Conferences for the remainder of 2016 are scheduled for October 12, November 16, and December 14.
- Adult Genomics Case Conferences occur on a quarterly basis (February, May, August and November), with the last session for 2016 currently slated for November 8. In 2017, they will move to the first Tuesday of the month.
- Plans are underway to begin a Carrier Screening Genomic Case Conference in 2017, with support from the Claire Altman Heine Foundation. Further details will be posted on the ACMG Genetics Education website as they become available.

*Further information about all ACMG activities and a full listing of our press releases and clinical genetics laboratory and practice guidelines can be found on our website at [www.acmg.net](http://www.acmg.net). The ACMG website now houses an Online Learning Center, as well. ACMG uses Facebook, LinkedIn, YouTube, and Twitter to augment its educational and advocacy missions, provide news and resources related to medical genetics, and improve communication with and among its members and stakeholders.*

*Submitted by Michael S. Watson, MS, PhD, FACMG  
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