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

September 3, 2014

Recent Activities of The American College of Medical Genetics and Genomics

The American College of Medical Genetic and Genomics (ACMG) is the professional home to over 1700 members, nearly 80% of whom are board certified clinical and laboratory geneticists and genetic counselors; it is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. The College's mission includes the following major goals: 1) to define and promote excellence in the practice of medical genetics and genomics and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics and genomics education to fellow professionals, other healthcare providers, and the public; 3) to improve access to medical genetics and genomics services and to promote their integration into all of medicine; and 4) to serve as advocates for providers of medical genetics and genomics services and their patients. This report summarizes key activities of the ACMG between February and September 2014.

Leadership and Advocacy Updates

The Board of Directors of the College and the ACMG Foundation held their first joint meeting in July to undertake a rigorous strategic planning exercise that will eventually shape the thrust of both organizations for the coming decade. The rapid integration of genomic tools into clinical practice—and their subsequent demands on geneticist and non-geneticist practitioners, as well as the College—gave impetus to this activity. It also allowed for the two organizations to better define where their missions and goals are complementary and overlapping. Moving forward, ACMG activities not related to our grants and contracts will focus on advancing our educational mission, promoting advocacy (particularly in the areas of billing and reimbursement) and accelerating the development of timely, evidence-based clinical and laboratory practice guidelines.

Secondary Findings and Clinical Genome and Exome Sequencing: In early 2014 a survey was conducted of all ACMG members, assessing opinions about the March 2013 "ACMG Recommendations for Reporting Incidental Findings in Clinical Exome and Genome Sequencing". Following preliminary analysis of the survey results and a year of formal and informal conversations with a variety of stakeholders within and beyond the medical genetics community the ACMG Board of Directors re-evaluated this heavily debated issue and modified its original recommendation as follows on March 27, 2014:

> "There appears to be consensus among ACMG members that patients should have an opportunity to opt out of the analysis of medically actionable genes when undergoing whole genome or exome sequencing. While the Board still considers [secondary findings] to be important medical information that is of great value to families, it has voted to recommend that such an "opt-out" option be offered to families who are considered candidates for clinical genome-scale sequencing." … This discussion should take place at the time the sample is being sent to the lab, rather than when the ordering clinician receives the results (as stated in the 2013 recommendation). Explanations of the "opt-out" option and its implications should be part of the standard pre-test education and counseling process, undertaken before the test is ordered. (The full press release can be found at https://www.acmg.net/docs/Release_ACMGUpdatesRecommendations_final.pdf)

A workgroup has been formed, co-chaired by Drs. Christa Lese Martin and David Miller, to

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develop a process to update—on a continual basis—the list of actionable genes to be reviewed as secondary findings in clinical exome/genome sequencing. Representation on the workgroup includes clinical and laboratory geneticists, an ethicist, a genetic counselor, a consumer and those with expertise in oncology, neurogenetics and cardiovascular genetics.

Medicare Coverage for Molecular Genetic Tests: Under the leadership of ACMG's new Medical Director, Dr. David Flannery, we are engaged in many activities to address the issue of reimbursement for molecular genetic tests. These efforts are critical to ensuring that patients have appropriate access to the tests they need for diagnostic confirmation and treatment, and that the clinical testing labs will be paid. Additionally, access to clinical trails is predicated on the confirmation and molecular delineation of a genetic diagnosis, which requires the use of genetic tests; thus, limited access to testing has chilling implications for clinical investigators as well as patients and families.

- ACMG participated in the CMS Annual Meeting on the Clinical Laboratory Fee Schedule, which included the presentation of proposed pricing for the new Molecular Pathology CPT Codes for 2015.
- ACMG followed up with a letter to CMS outlining concerns as well as recommendations. For all new Molecular Pathology CPT codes established in 2014 and beyond, ACMG recommends that CMS establish the National Limitation Amounts (NLAs) using cross-walking to existing codes rather than gap-filling. ACMG also supported the methodology for pricing multi-gene panels set forth by the Association for Molecular Pathology (AMP).
- ACMG has just completed a formal review with comments and recommendations to National Government Services (NGS) (the Medicare Administrative Contractor for WI, IL, ME, NH, NY, CT, VT, MA, RI) with regard to local coverage decisions for several molecular tests. This included a request from ACMG that NGS develop policies in a transparent manner and include "dossiers" for each of the molecular tests that explain why a Medicare patient would need to have the test performed as well as explaining the clinical utility of the test and results in order to justify coverage.
- ACMG is responding to recent draft FDA guidance on oversight of laboratory-developed tests (LDTs). There is significant concern about impact on germ-line genetic testing since the overwhelming majority of such tests are for the several thousand rare genetic diseases. Conflation of the LDT policy with differential reimbursement exacerbates the negative impacts of such a policy.

To engage the ACMG membership in these important activities:

- The Economics of Genetic Services Committee has created a listserv to facilitate communication regarding coverage, billing, and reimbursement problems; disseminate relevant materials; and keep ACMG members up-to-date on this critical and rapidly evolving issue.
- Medicaid and private payers are setting local coverage decisions and reimbursement rates for those CPT codes that have no established NLAs. Because regional payers respond better to local constituents than to national organizations, a providers' network is being formed to communicate with the regional payers. This also provides an opportunity to share information and examples of "best practices" among peers.
- ACMG is engaging CMS and the MACs to develop appropriate coverage policies and NLAs for Tier 1 codes currently considered statutorily excluded. However, we have also stressed to our members that it is vital for every lab to appeal both coverage denials and reimbursement rates that are below cost.

ACMG Participation at the AMA House of Delegates Results in Amended Language in Resolution Opposing Genetic Test Ordering Restriction Based on Specialty: In late spring, the American Society for Clinical Oncology (ASCO) submitted a proposed Resolution for the American Medical Association (AMA) House of Delegates (HOD) June meeting regarding a payer's policy that they believed to restrict a physician's ability to order genetic testing for patients. The payer's policy required that an independent board-certified Clinical Geneticist or a Certified Genetic Counselor assess patients before the payer would cover genetic testing for hereditary breast, ovarian and colorectal cancers. The American Congress of Obstetricians and Gynecologists (ACOG) joined ASCO in submitting the Resolution, and expressed the opposition to this payer policy because "the ultimate decision on the medical necessity for testing should be within the realm of the treating physician and his or her patient". The resolution also proposed that the AMA oppose limiting the ordering of genetic testing based solely on physician specialty or other non-medical based criteria. Further the original resolution asked the AMA to oppose requirements for utilization of non-affiliated medical specialists or non-physicians prior to ordering genetic testing. The resolution would then require the AMA, working with other interested specialty and component societies, to communicate general opposition to non-medical restrictions to genetic testing to relevant health insurers.

The ACMG Board of Directors reviewed the ASCO/ACOG resolution, and ACMG reached out to ASCO to discuss our concerns. This led to our developing amended language to the original resolution, which changed its tone, re-affirmed the AMA's own policy recognizing that not all physicians are knowledgeable about genetics, and affirmed the

importance of genetic evaluation, and pre- and post-test counseling by a qualified health professional when a patient is considered to be at risk for a hereditary susceptibility for cancer. Dr. Reed Pyeritz represented ACMG at the AMA meeting in Chicago and presented ACMG's concerns. The majority of the proposed amended language was adopted into the resolution, which then was passed by the AMA House of Delegates. The Resolution is now official AMA policy and will guide AMA staff in all matters. A copy of the final Resolution follows.

115. OPPOSITION TO GENETIC TESTING RESTRICTIONS BASED ON SPECIALTY

Introduced by American Society of Clinical Oncology, American Congress of Obstetricians and Gynecologists *Reference committee hearing: see report of Reference Committee A.*

HOUSE ACTION: ADOPTED AS FOLLOWS *See Policy TBD.*

RESOLVED, That our American Medical Association oppose limiting the ordering of genetic testing based solely on physician specialty or other non-medical care based criteria; and be it further

RESOLVED, That our AMA oppose public and private payers imposing a standard of practice with requirements for utilization of non-affiliated medical specialists or non-physicians prior to ordering genetic testing; and be it further

RESOLVED, That our AMA, working with other interested specialty and component societies, communicate our opposition to non-medical restrictions to genetic testing to relevant health insurers; and be it further

RESOLVED, That our AMA continue to support the importance of pre- and post-testing counseling when a patient is considered to be at risk for a hereditary susceptibility for cancer and other diseases by a qualified health professional so that patients have the benefit of informed decision-making regarding genetic testing.

Grant and Contract Updates
The MCHB/HRSA funded **National Coordinating Center for the Regional Genetic Service Collaboratives** continues to: 1) assess the extent to which genetic testing and services are integrated into the Affordable Care Act's Essential Health Benefits, 2) develop clinical decision support tools for non-genetics trained providers, and 3) improve access to genetic services through telegenetics, and other modalities.

The NHGRI/NIH funded **ClinGen Resource Project** held a very successful conference in the DC area in June, and planning is underway for its May/June 2015 meeting, to focus on improving genomic testing through collaboration. Work continues with development of standards, and data collection activities are ramping up.

The NICHD/NIH funded **Newborn Screening Translational Research Network (NBSTRN)** continues with data collection related to diagnosis and longitudinal evaluation and management of infants identified in newborn screening programs. We are now working directly with the grantees of the NHGRI/NICHD Newborn Screening and Sequencing projects.

***Genetics in Medicine* Updates**
ACMG's monthly journal, *Genetics in Medicine,* received a record high impact factor of 6.435 for 2013, up from 5.56 in the previous year. This ranks the journal at 17th out of 164 titles in the Genetics and Heredity category, and in the top tier of those with a primary clinical focus.

Between February and August 2014, the following Practice Guidelines were published in *Genetics in Medicine*:

Vockley J, Andersson HC, Antshel KM, Braverman NE, Burton BK, Frazier DM, Mitchell J, Smith WE, Thompson BH, and Berry SA; for the American College of Medical Genetics and Genomics Therapeutics Committee. **Phenylalanine hydroxylase deficiency: diagnosis and management guideline.** *Genet Med* 16(2):188-200 (February 2014) PMID: 24385074

CORRIGENDUM for the Phenylalanine hydroxylase deficiency: diagnosis and management guideline. *Genet Med* 16(4):356 (April 2014)

Alford RL, Arnos KS, Fox M, Lin JW, Palmer GC, Pandya A, Rehm HL, Robin NH, Scott DA, and Yoshinaga-Itano C; ACMG Working Group on Update of Genetics Evaluation Guidelines for the Etiologic Diagnosis of Congenital Hearing Loss; for the Professional Practice and Guidelines Committee. **American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss.** *Genet Med* 16(4):346-355 (April 2014) PMID: 24651602

Meetings and Education Update

ACMG held its most successful Annual Clinical Genetics Meeting in Nashville, TN, March 25-29, 2014. Increased numbers in all areas (abstract submissions, attendees, exhibitors, etc.) resulted in our meeting being named one of the 50 fastest growing meetings of the year.

Plans are well underway for the 2015 Annual Clinical Genetics Meeting, to be held March 24-28 in Salt Lake City, UT. Meeting highlights include:

- The 46th Annual March of Dimes Clinical Genetics Conference, "Interdisciplinary Approaches to Disorders of Sex Development: From Genes to Quality of Life"
- Two Short Courses: Cancer Genetics and Clinical Exome Sequencing
- New Sessions on Billing and Reimbursement in the Genomic Era and Prenatal/Perinatal Diagnostic Dilemmas
- Joint Plenary Session with the Society for Inherited Metabolic Disorders (SIMD)
- Abstract submission opens in October 2014; meeting details can be found at www.acmgmeeting.net.

The 2015 Genetics and Genomics Review Course will be held in Tampa, FL, June 18-21, 2015. On-site and online options will be available.

ACMG's Learning Center, a new website dedicated exclusively to genetics and genomic education for medical geneticists and non-geneticist providers is ready to launch. The site will contain interactive and multi-media course offerings, online events, opportunities for paced learning, discussions and more. It will also be searchable for course content, including PDF files and the Knowledge Direct Direct-to-WEB content for just-in-time learning support. Users will be able to track their CME credits and view personalized reports on course activity and test scores. The URL is ACMG.net/education.

ACMG Foundation Updates

• The ACMG Foundation awarded three Fellowship Training Grants and two awards to Trainees for research accomplishments during the 2014 Annual Clinical Genetics Meeting.

• The 2014 Summer Genetics Scholars Program enrolled its largest class to date this past summer. Twenty-five rising second-year medical students had hands' on experiences in medical genetics, with direct mentor-mentee relationships, at twenty-one institutions.

• Fundraising efforts continue in order to support new educational initiatives in genomic medicine and expand ongoing programs, especially those that target opportunities for trainees and growing the pipeline.

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. 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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