

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 1800 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. 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." Thus, three guiding pillars underpin ACMG 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 May and early September 2015.

Advocacy, Policy and Practice Activities

ACMG Leadership Releases New Scope of Practice Statement and Strategic Plan

This spring, the ACMG Board of Directors approved two important new ACMG documents: the [2015-2019 Strategic Plan](#) and a revised [Scope of Practice](#) (attached). These complimentary publications define how ACMG plans to position itself, as the national medical organization representing the dynamic specialty of medical genetics and genomics and its practitioners. This has never been more important than in these exciting (and challenging) times of rapid scientific discovery and subsequent translation into medical practice, public policy and public health. The ACMG has redefined its mission to "developing and sustaining genetic and genomic initiatives in clinical and laboratory practice, education and advocacy" with three guiding pillars related to clinical and laboratory practice, education, and advocacy. Each pillar is underpinned with a specific value proposition, followed by goals, tactical considerations and metrics.

The Strategic Plan provides ACMG with the infrastructure to maintain the highest possible standards in all facets of practice and in its relationships with all stakeholders. In a companion Commentary to the publication of ACMG's revised Scope of Practice in the September issue of *Genetics in Medicine*, ACMG President, Dr. Jerry Feldman, discusses the importance of ACMG in redefining the practice of medical genetics and genomics in a field that is increasingly technology-driven. The new Scope of Practice begins by clearly stating the value of board-certified Clinical Geneticists and Clinical Laboratory Geneticists, from their roles performing test interpretation in the diagnostic laboratory to the clinical genetics consultation. It also establishes how we relate to other members of the medical genetics healthcare team and where we find common ground with other medical specialists.

With a future that is ours to create, the ACMG Board of Directors believes these two new documents allow ACMG to take a major step in defining this future for the practice of medical genetics and genomics.

ACMG's Genetic Testing Recommendations Provided Through the *Choosing Wisely*® Campaign are Broadly Recognized

In mid-July, ACMG released a list of five items patients and providers should discuss regarding specific genetic tests as part of *Choosing Wisely*, an initiative of the American Board of Internal Medicine Foundation. ACMG's five evidence-based recommendations are aimed at improving genetic testing decisions made between patients and clinicians. To date, more than 100 national and state medical specialty societies, regional health collaboratives and consumer partners have joined the *Choosing Wisely* campaign about appropriate care.

ACMG's list, which has been widely cited, was developed over the past year with careful consideration of the latest evidence, expert opinions and research. Input was sought from ACMG's Laboratory Quality Assurance, Professional

Practice and Guidelines, and Therapeutics Committees. A list of 18 items was reviewed by the ACMG Board of Directors and the five items currently thought to most likely improve quality of care and reduce costs related to genetic testing were selected. The five recommendations identified by ACMG are:

1. Do not order a duplicate genetic test for an inherited condition unless there is uncertainty about the validity of the existing test result.
2. Do not order *APOE* genetic testing as a predictive test for Alzheimer disease.
3. Do not order *MTHFR* genetic testing for the risk assessment of hereditary thrombophilia.
4. Do not order *HFE* genetic testing for a patient without iron overload or a family history of *HFE*-associated hereditary hemochromatosis.
5. Do not order exome or genome sequencing before obtaining informed consent that includes the possibility of secondary findings.

As genetics and genomics continue to play an increasingly important role in all aspects of patient care, patients will have more questions about genetic and genomic testing. ACMG hopes its *Choosing Wisely* list will provide helpful direction for clinician-patient conversations about the genetic tests on this list and any related decision-making. All clinicians, not just medical geneticists, play a key role in making sure that the appropriate tests and treatments are prescribed.

Economics of Genetic Services Committee is Benchmarking Clinical Geneticist wRVUs and Monitoring Changes to CMS's Clinical Laboratory Fee Schedule

ACMG's Economics of Genetic Services Committee has developed a mechanism for collecting accurate benchmarking data for average work RVU's (wRVU's) for clinical geneticists. The clinical productivity of geneticists is often compared to other specialty benchmarks because the groups (Association of Administrators in Academic Pediatrics (AAAP), Medical Group Management Association (MGMA) and University HealthSystems Consortium (UHC)) that produce the averages for work RVU's do not have enough data for genetics. ACMG's Medical Director, Dr. David Flannery, has worked with UHC to combine the different titles for geneticists into a subspecialty of Clinical Genetics. ACMG members who practice as clinical geneticists have been asked to tell their administrators to classify them as Clinical Genetics, and this number has increased more than six-fold, from 37 to 235 clinical geneticists whose productivity is now being tracked in this new category! Moving forward, we are trying to obtain information from MGMA and AAAP to be able to do the same since ACMG members may work at institutions that use MGMA or AAAP.

The Economics Committee continues to monitor CMS's Clinical Laboratory Fee Schedule meetings. The most recent rate-setting meeting occurred in July, with comments presented regarding the molecular pathology codes. It was also announced that nothing would be discussed regarding the Protecting Access to Medicare Act of 2014 so we will have to await the completion of rulemaking before ACMG can make any public comments.

ACMG Acts on Belief that Education is Key to Effective Application of NIPS

In May 2013, ACMG published its "Statement on noninvasive prenatal screening (NIPS) for fetal aneuploidy", clearly delineating major points to consider with regard to test implementation. This statement is about to be updated to incorporate scientific advances related to testing technology, clinical utility and the breadth and limitations of anticipated results. The information in the updated clinical practice document will then be translated into a patient education brochure in order to provide prospective parents with more complete information (and questions to have answered) before and after consenting to NIPS, with its range of anticipated and unanticipated results.

ACMG's 2013 Statement came to the attention of Congresswoman Jaime Herrera-Beutler earlier this year, and our technical expertise was heavily sought in the drafting of HR 3441, the "Accurate Education for Prenatal Screenings Act." Introduced in August, this bill would amend the Public Health Service Act to establish pre- and posttest education programs for patients and providers regarding cell-free DNA prenatal screening. The CDC is listed as the HHS agency that would coordinate these efforts with broad input from stakeholder groups, including medical geneticists, obstetricians and other prenatal care providers, pediatricians, disability advocacy and parent groups, as well as industry. HR 3441 recognizes that NIPS is unique because it has rapidly become a high volume test, being offered to a large proportion of pregnant patients and thereby shifting the rendering of a prenatal testing modality away from the maternal-fetal-medicine experts into primary prenatal care. As such, pretest education and counseling, and some posttest education and genetic counseling, will be done by obstetricians, family physicians and other members of the prenatal healthcare team, creating a need for ongoing professional education to meet standards set forth in professional guidelines and to establish inter-professional consistency. ACMG and its members are ideally poised to tackle this issue because of our unique experience and expertise managing both the complexities and nuances of the conditions potentially detected by NIPS.

HR 3441 has been referred to the House Committee on Energy and Commerce. The updated ACMG statement on NIPS and the companion patient brochure will be shared with Council as they are completed.

ACMG Joins FDA's Network of Experts

ACMG has entered into an agreement with the US Food and Drug Administration's Center for Devices and Radiologic Health (CDRH) to be part of its Network of Experts. CDRH's mission, to protect and promote the public health by assuring that patients and providers have access to safe, effective and high quality medical devices, includes regulation and oversight of genetic and genomic testing. As CDRH staff members identify issues requiring external expertise, they routinely include a description of the types of experts they are seeking. Any time FDA determines that ACMG members are likely to have the necessary expertise, ACMG is now prepared to respond with a list of potential experts and their supporting information. ACMG joins forty-three other specialty societies in this Network, including the American Academy of Neurology, American Academy of Pediatrics, American Society of Human Genetics, and the Association for Molecular Pathology.

ClinGen featured in *NEJM*, Coinciding with Successful International Meeting with DECIPHER

ACMG received high visibility during the latter part of May with enhanced activity surrounding an international meeting and a landmark publication, all part of ACMG's role as one of the NIH-supported Clinical Genome Resource (ClinGen) grantees. (*The ClinGen project aims to build a genomic knowledgebase to improve patient care. Genetic clinicians and medical researchers hope to use this information about genetic variants in a variety of ways including making better predictions about an individual's risk of disease, developing tailored treatments and improving clinical decision making.*)

The ClinGen project was in the news with a comprehensive paper published in the *New England Journal of Medicine (NEJM)* on May 27, coinciding with a very successful meeting held in Washington, DC on May 27-28 in conjunction with DECIPHER (DatabasE of genomiC variation and Phenotype in Humans using Ensembl Resources). The topics of the meeting included: curating gene-disease relationships, initiatives facilitating clinical genomic IT development, integrated approaches to informed consent, lessons learned from aggregating population and patient data, and discussions related to the tools used in genomics. Information about this meeting, and other related meetings, can be found on the [ClinGen website](#). The 2016 ClinGen/DECIPHER meeting is slated for June 24-26, in Cambridge, England.

The *NEJM* article presented a detailed overview of the ClinGen program. It is expected to become a landmark paper because as genetic sequencing becomes more common, interpreting data in a meaningful way and standardizing practices is imperative. ClinGen aims to develop new standards for interpreting those many, many genetic variants.

Laboratory Developed Test (LDT) Regulation, Oversight and Reimbursement: National Initiatives

ACMG continues to address the issues presented by FDA's new guidance on the regulation of Laboratory Developed Tests (LDTs). Many in the laboratory community consider the LDTs to be locally developed and delivered clinical procedures rather than the types of classical laboratory tests to be regulated by FDA. There has been significant concern expressed as to whether FDA actually has the authority to oversee these or if it should be under the CLIA regulations that apply to laboratory practices. Multiple professional medical associations with interests in laboratory testing have been meeting to develop alternatives. Because both the House Energy and Commerce Committee and the Senate HELP Committee are in the process of developing bills that address FDA and CLIA oversight, including that of LDTs, groups continue to meet with staff and legislators to discuss how LDTs should be addressed in their legislation. Final resolution has not been reached yet.

We also continue to monitor the implications of the Protecting Access to Medicare Act for genetic testing laboratories. It is clear that in this area of testing, reducing reimbursement to the lowest level of any payer would have significant implications for the viability of academic laboratories and the innovation required to improve genetic and genomic testing, as well as the access of patients to services.

ACMG Continues to Strongly Advocate on Behalf of Members Regarding Regulation of LDTs

The FDA's proposed regulation of laboratory-developed tests (LDTs) would do more harm than good, argue ACMG's Executive Director Dr. Michael Watson and *Genetics in Medicine* Editor Dr. James Evans in "[The Case Against FDA Regulation of Laboratory-Developed Diagnostic Tests](#)". For LDTs to be under the FDA's strict regulations would be detrimental to genetic testing and the future of genomic medicine, impeding innovation and potentially leading to patients losing access to timely, life-saving testing.

ACMG has been vocal in publicly stating its position that the FDA should not regulate laboratory-developed diagnostic tests, with a highly visible Viewpoint article published in the *Journal of the American Medical Association*

(*JAMA*) in January 2015, "Genetic Testing and FDA Regulation – Overregulation Threatens the Emergence of Genomic Medicine."

Grant and Contract Updates

The Clinical Genome Resource Project (ClinGen)



See *ClinGen featured in NEJM, Coinciding with Successful International Meeting with DECIPHER* on the previous page of this report.

The Newborn Screening Translational Research Network (NBSTRN)



Now finishing its seventh year at ACMG, the mission of the NICHD-NIH funded Newborn Screening Translational Research Network (NBSTRN) is to improve the health outcomes of newborns with genetic or congenital disorders through an infrastructure that allows investigators access to robust resources for newborn screening research. The NBSTRN infrastructure includes three tools; the Virtual Repository of Dried Blood Spots (VRDBS), the Longitudinal Pediatric Data Resource (LPDR), and the Region 4 Stork Database (R4S). Recently, the NBSTRN launched the ELSI Advantage, a new resource for NBS researchers that addresses ethical, legal and social issues. This tool is comprised of an interactive website that contains information on IRB's, NBS related FAQ's, and templates to customize your own Consent Forms. Visit the NBSTRN.org for more information.

NBSTRN is now organizing to address the implications of proposed changes to the Common Rule that were recently released by the Office for Human Research Protections (OHRP). This was done in response to the Newborn Screening Saves Lives Reauthorization Act that has required consent for the use of newborn dried blood spots in federally-funded research until OHRP addresses the issue itself. The multistate prospective pilot studies of Pompe disease to assess its appropriateness for newborn screening have been taken off of a 'hold' because they weren't found by OHRP to meet their definition of "research". Multistate prospective pilots for mucopolysaccharidosis type 1 (MPS-1) will begin in October. These issues were highlighted during the very successful national NBSTRN Network Meeting, held September 10-11, 2015.

The National Coordinating Center for the Regional Genetic Service Collaboratives (NCC)



Since 2004, ACMG has served as the National Coordinating Center (NCC) for the seven Health Resources and Services Administration (HRSA) Genetics Collaboratives (RCs), through a cooperative agreement with HRSA. The current award continues through May 2017. Under the new award, NCC will: 1) develop a framework for regional genetic care centers; 2) provide an infrastructure that strengthens communication and collaboration between the RCs, offer technical and clinical expertise as needed, promote and disseminate outcomes of RC activities; and 3) implement the NGEEN.

During its current funding cycle the NCC has worked to address gaps in access to and financing of genetic services across the lifespan and across healthcare delivery systems. At the time of the rollout of the ACA and the state exchanges the NCC was funded to carry out several system-wide projects aimed at understanding the impact of the ACA on essential healthcare services and access for individuals with genetic conditions. On June 15, 2015 a (free) national meeting, *Genetic Considerations for Accountable Care Organizations and Integrated Care Delivery System*, of insurers, policymakers, healthcare providers, delivery systems, consumers, regional collaborative leadership, advocacy organizations and public health was sponsored by the NCC. The meeting participants identified gaps and feasible solutions that can be developed into a framework the NCC/RC system can use to collaborate with integrated delivery systems and payers to move towards solutions that will improve access to and availability of genetic services across the lifespan and across service delivery systems.

To begin thinking about new models for care delivery, the NCC is presently conducting a survey of a breadth of genetic service providers to ascertain practice characteristics, the capacity of the current genetics workforce, and to seek participants' visions of ways to enhance access to services and the potential role of regional genetic service support centers. Survey findings will be reported to HRSA and inform planning the work for the next funding cycle.

Genetics in Medicine Updates

Record High Impact Factor for 2014 Places *Genetics in Medicine* in the Top 4% of All Indexed Journals

The Thomson Reuters Impact Factor Journal Citation Reports recently announced that the impact factor of *Genetics in Medicine* (*GIM*), ACMG's peer-reviewed medical genetics and genomics journal, increased to 7.329 in 2014 from its value of 6.435 in 2013. *GIM* is currently ranked 15 of 167 titles in the Genetics and Heredity category and in the top tier of genetics journals that have a primarily clinical focus. These indices place *GIM* in the top 4% of all indexed journals. ACMG attributes this increase to the vibrancy of the field of Medical Genetics as a whole and its rising importance to clinical practice across all medical specialties, as well as its relevance to public health.

Recent ACMG Publications

The following ACMG publications appeared in the College's monthly journal between June and September 2015.

ACMG Board of Directors. **Clinical utility of genetic and genomic services: a position statement of the American College of Medical Genetics and Genomics.** *Genet Med* 17(6):505-507 (June 2015) PMID: 25764213

ACMG Board of Directors. **Scope of practice: a statement of the American College of Medical Genetics and Genomics (ACMG).** *Genet Med* 17(9): online only (September 2015)
PMID: 26204421 (https://www.acmg.net/docs/ACMG_SOP_GIM_AOP_072315.pdf)

Feldman G. **American College of Medical Genetics and Genomics: revised scope of practice and strategic plan.** *Genet Med* 17(9):713 (September 2015) PMID: 26204422

GIM Website Now Houses All ACMG Statements and Guidelines

Genetics in Medicine has posted all the published ACMG Statements and Guidelines from the last 17 years on one easily accessed webpage (http://www.nature.com/gim/statements_and_guidelines_by_date.html). Authored by the Board of Directors and Committees of the College, these recommendations are all Board-approved and were designed to serve as educational resources for medical geneticists and other healthcare professionals to help provide quality medical genetics and genomics services.

GIM Podcasts

Use the URL <http://feeds.nature.com/gim/podcast/current> to access *Genetics in Medicine*'s monthly Podcast, known as *GenePod*, to hear live a discussion of a timely (and often controversial) article from the most recent published journal.

Meetings and Education Updates



ACMG's Annual Clinical Genetics Meeting Returns to Tampa in March 2016

The 2016 ACMG Annual Meeting will be held March 8-12, 2016 in Tampa, FL. The Program is set, the meeting website is live at www.acmgmeeting.net, attendees can save \$200 on Early Bird registration through December 16, 2015 and abstract submission remains open through November 20, 2015, 11:59 PT. Recognized as one of Trade Show Executive Magazine's Fastest Growing Meetings for several years running, the ACMG Annual Meeting has become the place for professionals with an interest in clinical genetics and genomics research and practice to network with colleagues, meet new collaborators from related disciplines, and hear about the very latest developments. The 2016 Annual Meeting will be preceded by three short courses: "NAMA at the ACMG: The Best of the North American Metabolic Academy"; "Advanced Molecular Cancer Genetics: State of the Art Today and Beyond"; and "Tools and Approaches to Assess the Genetic Basis of Disease". The meeting will also include the March of Dimes Clinical Genetics Conference, "Prader-Willi Syndrome: New Insight Into A Classic Genetic Disorder".

ACMG's Genetics Academy Continues to Grow



With technical expertise provided by ACMG's Education and CME Committee, our Genetics Academy Learning Center continues to grow. Featuring both live programs and online content, there are genetics

and genomics curricula (from webinars to Annual meeting webcasts and MOC modules) to meet the needs of genetics and non-genetics health professionals at all practice levels.



Back Again! The very popular **ACMG Genomics Case Conferences** resumed on September 16th with a team from the University of Chicago presenting, “Clinical Exome Sequencing: MEDICAL Genetics is Required to Reach a Diagnosis.” This conference drew on cases from their own laboratory to demonstrate the utility of exome sequencing for rare and more common conditions with congenital to adult onset. Scheduled from 2:00 - 3:00 pm ET on the third Wednesday of the month, experts from a selected host institution present and discuss intriguing, complex or difficult patient cases in the area of genomics. The fall Case Conference Schedule continues with the Children’s Mercy Hospital, Kansas City, MO and the University of Missouri, Kansas City, MO (October 21); Geisinger Health System (November 18); and Baylor College of Medicine (December 16). *The Genomics Case Conferences are supported by an educational grant from QIAGEN Bioinformatics and the Ingenuity Clinical Decision Support Platform.*

Two new programs are coming soon: **Quarterly Adult Genomic Case Conferences** will begin in November, and a **Mouse Genome Recorded Webinar**, “Using Mouse Model Data to Prioritize and Characterize Genes with Unknown Clinical Significance” will be available late fall. This webinar will provide an overview on the Mouse Genome Project and its relevance to clinical genetics. The ACMG Genetics Academy can be accessed at ACMG.net/education.

ACMG Foundation for Genetic and Genomic Medicine Updates



The **2015 Summer Genetics Scholars Program (SGSP)** just completed another successful year, with a record number of 28 rising second-year medical students participating in 6-8 week hands’ on experiences with medical genetics and genomics mentors at twenty-two institutions across the United States. Continued support for the very popular SGSP is included in the ongoing fundraising efforts of the ACMG Foundation, with the intention of expanding the Program to include more deserving medical students in future years.

When the ACMG Foundation convenes for its Annual Board Meeting on September 29, **four newly elected Directors** will be in attendance. Just beginning their terms are: Arthur L. Beaudet, MD, FACMG, Henry and Emma Meyer Professor in the Department of Molecular and Human Genetics at the Baylor College of Medicine; Laurel Estabrooks, PhD, FACMG, Vice President of Genetics Business Development for SCC Soft Computers; Laura Furmanski, PhD, Senior Vice President, Bioinformatics at QIAGEN; and Anthony Romaine Gregg, MD, FACOG, FACMG, Professor and Chief, Division of Maternal-Fetal Medicine at the University of Florida Department of Obstetrics and Gynecology.

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

ACMG Liaison to the National Advisory Council for the National Human Genome Research Institute, NIH