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

February 7, 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 September 2013 and January 2014.

ACMG Continues its Involvement in the Current Crisis in Molecular Pathology (MoPath) Test Coverage and Reimbursement

ACMG continues its activities to address the impact of the 2014 CPT codes for molecular genetic tests that were announced following the Center for Medicare and Medicaid Services' (CMS) "gap-filling" process. Reimbursement rates for many tests have been decreased as much as 25-35%, and in some cases are lower than actual costs. Aggressive stances in this area are based on projections at CMS that the current cost of laboratory testing in health care in the US is $70 - $80 billion per year with an anticipated doubling over the next four years due to molecular and genetic/genomic testing.

On October 30, ACMG joined CAP, AMP, and several other professional groups representing more than 150,000 medical and laboratory professionals engaged in molecular diagnostic testing, in sending a 100-page document to CMS. The strongly-worded message requested a meeting with CMS to discuss concerns and recommendations for changes to the Molecular Diagnostic Services Program ('the MolDX Program') including the assignment of payment rates for molecular diagnostic tests via the "gap-filling" process, and the changes to coverage policy for previously recognized standard of care service. The letter stated, "We have serious concerns with the MolDX program and the inconsistencies of the program with the established Local Coverage Decision (LCD) process." It also declared, "These issues are creating serious confusion and opacity to a process that has functionally worked for determining appropriate coverage for Medicare beneficiaries. We bring this to your attention along with the Regional Offices that oversee the Palmetto and Noridian contracts so that these issues can be rectified and meet the instructions as set by CMS and the statutory requirements. Letters and other information about this are freely available on the ACMG website under the Education tab (Molecular Coding and Reimbursement 2013).

The AMA is also following the issue closely, with specific attention to situations in which patient care and access are compromised either because labs have had to close (others are simply absorbing greater losses), limiting access to necessary standard of care testing, or because tests (such as chromosomal micro-arrays and exome sequencing, in some cases) are no longer covered. The AMA is collecting examples of cases in which individuals did not have access to testing because of CMS's new coverage decisions. These issues overlap with those of interest to the Rare Diseases community, so in addition to working with the professional organizations already mentioned, ACMG is in communication with a larger

7220 Wisconsin Avenue, Suite 300, Bethesda, MD 20814 Telephone 301-718-9603 Fax 301-718-9604 www.acmg.net

coalition looking at rare disease policies regarding access to care in several contexts—with our emphasis being on orphan disease tests.

ACMG Intensifies Focus on Educational Needs in Genetic and Genomic Medicine

In the years since the human genome sequence was first reported, clinical applications have been entering the patient care arena at an unprecedented pace. This has exacerbated the need for more physician and non-physician healthcare professions with a solid, basic understanding of genetics and genomics, and who are well versed in its implications and applications to patient care—from selecting the right modalities for the patient or family to eliciting informed consent, facilitating decision making, interpreting test results, overseeing care coordination, collecting outcomes data and implementing clinical trials. ACMG has recognized many other gaps to achieving optimal care delivery, from the lack of manpower, to needs for curriculum reform and health professional education. In response to these deficiencies several new and exciting initiatives are underway. These include Banbury III, "Genetics Training in the Genomics Era," an invited workshop to be held February 24-26 at the Cold Springs Harbor Banbury Center to consider how the rapid integration of genomics into healthcare impacts our service delivery models and the training/education of both medical geneticists and non-genetics trained providers.

We recently established an ACMG Education Department, which will seamlessly merge our educational activities with our Meetings Department and allow us to broaden the scope of our Educational activities. An Associate Director of Education, with expertise in the administration of CME, will begin in a newly created position later this month. Finally, targeted, intensified fundraising by the ACMG's Foundation for Genetic and Genomic Medicine is allowing ACMG to truly take a leadership role in providing continuing education to healthcare professionals about genetics and genomics in medicine by implementing innovative new programs, as well as continuing current successful activities, such as the Summer Genetics Scholars Program.

One such new program is a CME webinar series for physicians, *ACMG Genomics in Clinical Practice.* The goal of this program is to help non-geneticist physicians and other healthcare professionals understand genomics in the clinical setting. The first four webinars are being offered for free, with a modest CME certificate fee for those who wish to earn CME credits. These cover: Preconception and Prenatal Cystic Fibrosis Carrier Screening; Noninvasive Prenatal Screening; Genomics in Clinical Practice; and Preimplantation Genetic Diagnosis and Genomic Testing. The *ACMG Genomics in Clinical Practice* webinar series can be found on the ACMG website under the Education tab. (ACMG's *Genomics in Clinical Practice* is supported by an unrestricted educational grant from Illumina, Inc.)

Dr. David B. Flannery Named ACMG's Next Medical Director

David B. Flannery, MD, FACMG, FAAP begins his position as ACMG's new Medical Director later this month. Dr. Flannery is a Founding Fellow of the College and has served on the ACMG's Economics of Genetic Services Committee (member and co-chair), Professional Practice and Guidelines Committee (member), and CPT and Reimbursement Committee (member). He was on the Editorial Panel for the ACMG's *Genetics Billing and Reimbursement Manual* and he is currently the ACMG representative to the American Medical Association's CPT Advisory Committee. He has also served as a member of the Southeast Regional Newborn Screening & Genetics Collaboratives (SERC). SERC is one of the seven HRSA Genetics Collaboratives that is under the National Coordinating Center, housed at ACMG through its cooperative agreement with MCHB/HRSA.

Dr. Flannery is presently employed by the Medical College of Georgia where he serves as Professor of Pediatrics, Vice Chair for Administration, Medical Director of Pediatric Ambulatory Care and Chief of the Genetics Division. He received his BA in Psychology from Columbia University and his Doctorate of Medicine from Emory University. Dr. Flannery succeeds Dr. Barry Thompson, who recently retired after five years of service to ACMG as its first Medical Director.

Genetics in Medicine Updates

The October 2013 issue of *Genetic in Medicine* is solely dedicated to Genomics in Electronic Health Records (EHR). Co-edited by Joseph Kannry, MD, a board-certified internist and Lead Technical Informaticist of the Epic Clinical Transformation Group, Mount Sinai Medical Center (New York) and Marc Williams, MD, FACMG, director of the Genomic Medicine Institute, Geisinger Health System (Danville, PA), this special issue contains a collection of research articles addressing the challenges of, and solutions for, the integration of genomic data into EHRs. A number of insights are offered from research teams actively engaged in integrating genomic medicine into clinical care. Most of the contributions derive from the experiences of the individual sites that comprise the Electronic Medical Records and Genomics (eMERGE) Network, a national consortium funded by the National Institutes of Health. However, additional perspectives are provided by a commercial EHR vendor and by the

Clinical Sequencing Exploratory Research (CSER) consortium, a cooperative group exploring applications of genomic sequencing. In a co-authored lead editorial Drs. Kannry and Williams articulate their vision that the volume should serve as a valuable reference and jumping off point for moving this nascent field into the future. The included case studies address such questions as: How can genomics be meaningfully incorporated into routine healthcare? How will genomic data be stored, processed, updated and retrieved? How will genomic data first be used to help patients? How will patients be involved in decisions about their genomic information? The journal can be accessed at http://www.nature.com/gim/journal/v15/n10/index.html.

Between September 2013 and January 2014 the following Clinical and Laboratory Practice Guidelines and ACMG Policy Statements were also published in *Genetics in Medicine*:

ACMG Board of Directors. **Points to consider for informed consent for genome/exome sequencing.** *Genet Med* 15(9):748-749 (September 2013)

Rehm HL, Bale SJ, Bayrak-Toydemir P, Berg JS, Brown KK, Deignan JL, Friez MJ, Funke BH, Hegde MR, and Lyon E; for the Working Group of the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee. **ACMG clinical laboratory standards for next-generation sequencing.** *Genet Med* 15(9):733-747 (September 2013)

ACMG Board of Directors. **ACMG statement on access to reproductive options after prenatal diagnosis.** *Genet Med* 15(11):900 (November 2013)

South ST, Lee C, Lamb AN, Higgins AW and Kearney HM; for the Working Group for the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. **ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013.** *Genet Med* 15(11):901-909 (November 2013)

Hegde M, Ferber M, Rong M, Samowitz, W and Ganguly, A; a Working Group of the American College of Medical Genetics and Genomics (ACMG) Laboratory Quality Assurance Committee. **ACMG technical standards and guidelines for genetic testing for inherited colorectal cancer (Lynch syndrome, familial adenomatous polyposis, and MYH-associated polyposis)** *Genet Med* 16(1):101-116 (January 2014)

New Electronic Resources Now Available From ACMG Grants and Contracts

Two new websites were launched in November. The NICHD/NIH-funded **Newborn Screening Translational Research Network's** (NBSTRN) redesigned website has a number of new features for researchers, including a dedicated "Planning your Research" section that includes such helpful tools as state-specific profiles, interactive consent modules and information regarding return of results. Central to the new website is the unified registration page, an easy to use registration portal which, upon approval, grants access to the Virtual Repository of Dried Blood Spots (VRDBS), the Longitudinal Pediatric Data Resource (LPDR), and the Region 4 Stork-Laboratory Performance Database (R4S). The website can be accessed at www.NBSTRN.org.

The MCHB/HRSA-funded **National Coordinating Center for the Regional Genetic Service Collaboratives** (NCC) has updated its website with a number of interactive tools and resources for families, providers and the Regional Collaboratives (RC). There are also new features, including an interactive RC map, a central Newsroom, and an up-to-date calendar with all NCC/RC activities. Visit this website at www.nccrcg.org.

The long-awaited **ACMG ACT Sheet Mobile App** is now available, with more information on the NCC website. For nearly a decade, the ACMG ACT Sheets and their accompanying algorithms have been "go to" resources for clinicians seeking information on genetic conditions (often those identified through newborn screening), to help inform clinical decision-making. Developed by the NCC and the ACMG, the ACT Sheets are approved by the ACMG Board of Directors and revised every five years. The ACMG ACT Sheets will remain on the NCC and ACMG websites. However, given the rarity of many genetic conditions, these resources are now accessible on a smartphone or tablet with the ACT Sheet Mobile App, downloadable for free at the Google Play or the Apple App Store.

As NACHGR knows well, ACMG is also partnered with other grantees and in the NHGRI sponsored ClinGen Resource Project.

March 2014 Annual Clinical Genetics Meeting is Predicted to Break All Previous Records

Participant registration, exhibit booth sales and abstract submissions are all at record highs for the 2014 ACMG Annual Clinical Genetics Meeting, to be held in Nashville, TN, March 25-29. Program highlights include: Two short courses, "Interpretation and Reporting of Sequence Variants" and "Recent Advances in Clinical Neurogenetics." A workshop on "Transition to Evidence-Based Clinical Guidelines: Understanding Systematic Review and Translation of Evidence to Recommendations" also precedes the meeting. The annual R. Rodney Howell Symposium in Public Health Genomics, "Care Models for the Delivery of Clinical Genetics and Genomics Services" and the 45th Annual March of Dimes Clinical Genetics Conference, "Vascular Anomalies: Classifications, Etiologies and Therapies" also merit recognition, along with two other special sessions, a panel discussion, "Point-Counterpoint: One Year Later, the Influence of the ACMG Recommendations for Reporting of Incidental Findings in WES/WGS," moderated by NPR science correspondent Joe Palca, and the closing plenary session: "Revisiting 'Duty to Recontact' in the Genomics Era: Interdisciplinary Perspectives and An Open Forum," funded by the ACMG Foundation's Father Robert C. Baumiller Fund for Genetics and Society. This year's meeting will also feature more activities designed for students, residents, fellows and other trainees. Detailed information and a complete schedule of events, can be found by visiting the meeting website at www.acmgmeeting.net.

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