American College of Medical Genetics
Medical Genetics: Translating Genes Into Health®

January 29, 2010

Recent Activities of
The American College of Medical Genetics

The American College of Medical Genetics (ACMG) is the professional home to more than 1,400 board certified clinical and laboratory genetics professionals and is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics. The College's mission includes four major goals: 1) to define and promote excellence in medical genetics practice and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics education to fellow professionals, other healthcare providers, and the public; 3) to improve access to medical genetic services and to promote the integration of genetics into all of medicine; and 4) to serve as advocates for providers of medical genetic services and their patients. This report summarizes key activities of the ACMG between September 2009 and January 2010.

Newborn Screening Translational Research Network Coordinating Center (NBSTRN-CC) Leads ACMG's NBS Activities

As noted previously, in late 2008 ACMG was awarded a $13.5 million, 5-year contract from the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) of the National Institutes of Health (NIH) to establish a National Newborn Screening Translational Research Network (NBSTRN) Coordinating Center. During Year 1, the Standing Committee and each of the NBSTRN Workgroups established regular meeting schedules, using conference calls and face-to-face meetings to identify, prioritize, and begin accomplishing their goals. Efforts in the area of long-term follow-up continue to expand and remain a focus of the joint activities of the NBSTRN-CC and the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC). With the very recent approval by the Advisory Committee on Heritable Disorders in Newborns and Children of the addition of the severe combined immunodeficiencies (SCID) to the uniform NBS panel, the NBSTRN-CC will be taking the lead in the coordination of the use of its infrastructure and resources for pilot programs and both short- and long-term follow-up data collection. Additionally, ACMG members' expertise and the relationships of the NCC and the RCs with local primary care providers, State NBS Programs, and health departments will all come together with the preparation of educational materials for laboratories, healthcare providers, and families, related to newborn screening for SCID.

ACMG Continues to Participate in Litigation Related to Intellectual Property and the Patentability of Genes, and Supports Position of the Secretary's Advisory Committee on Genetics Health and Society (SACGHS)

The ACMG has a long-standing position that genes are products of nature, and as such should not be patentable. ACMG recently provided comments to the Secretary's Advisory Committee on Genetics Health and Society (SACGHS) on the issue of gene patents. Recommendations of the SACGHS recognize that gene patenting is creating problems both in access to genetic testing and in insuring the quality of genetic testing. SACGHS also noted the lack of evidence that gene patents have contributed to the development of products for the diagnostic laboratory marketplace, a view shared by ACMG.

Consistent with its position on gene patents and its mission to advocate for broad public access to quality genetic tests and services, ACMG joined with the College of American Pathologists (CAP), the Association of Molecular Pathologists (AMP), the American Society for Clinical Pathology (ASCP), Breast Cancer Action, the Boston Women's Health Book Collective and several individuals in a lawsuit against Myriad Genetics. The case, in which

the American Civil Liberties Union (ACLU) is representing the College and the other plaintiffs, challenges the legality and constitutionality of granting patents on the human genes associated with breast and ovarian cancer because these patents interfere with diagnostic testing, stifle research and limit women's options regarding their health care. In early November 2009, a New York District Court ruled against motions to dismiss the case, in an 88-page court decision. Further action will be forthcoming.

In another intellectual property case, *Prometheus Laboratories, Inc. v. Mayo Collaborative Services,* ACMG filed an *amicus curiae* brief in support of a petition to the US Supreme Court to grant certiorari in the case. Although the Court has considered the petition, it appears that a decision will await their decision in the related *Bilski* case.

ACMG/NCC's Newborn Screening ACT(ion) Sheet Library Updated and Expanded

Since 2006, the NBS ACT(ion) Sheets and accompanying algorithms have become valuable resources to primary care providers and medical geneticists by offering critical decision support at the point-of-care. The NCC/ACMG ACT Sheet Workgroup was tireless in 2009. They reviewed and updated all ACT Sheets and algorithms for the 29 conditions in the uniform NBS panel, with new versions available on the ACMG and NCC websites as of February 1, 2010. In the near future, new ACT Sheets will roll out for all hemoglobin disorders, adult PKU, fragile X syndrome, cystic fibrosis and family history of colon cancer. These, too, have been designed for primary care providers to use at the point-of-care; a formal evaluation of these will be conducted through the American Academy of Pediatrics' Quality Improvement Innovation Network (QuIIN) program in the first half of 2010. ACT Sheet development will continue in 2010, under the leadership of workgroup chairs Dr. Harvey Levy and Dr. Richard King, with an emphasis on ACT Sheets to facilitate seamless medical transition from pediatric to adult care for those with genetic conditions. All ACT Sheets and algorithms receive the approval of the ACMG Board of Directors prior to their dissemination.

ACMG and NCC Join Forces to Improve Access to Genetic Services Using Telemedicine Technologies

In November 2009, the ACMG and its National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC) co-sponsored a small meeting of subject matter experts to tackle the policy issues that have posed barriers for medical geneticists who wish to improve access to services—particularly in underserved areas—using telemedicine technologies. Meeting participants included national leaders in telehealth law and representatives of the American Telemedicine Association, the Federation of State Medical Licensing Boards, Regional Collaboratives, medical geneticists, nursing organizations, and HRSA's Office for the Advancement for Telehealth. A white paper is being prepared that will set forth recommendations addressing interstate physician licensing, hospital credentialing, reimbursement, and program sustainability issues.

ACMG Engages in Multifaceted Approach to Improve Reimbursement for Genetic Services

Reimbursement for genetic services is both nuanced and complex. Yet, the ability of all labs and providers to continue providing quality genetic services is dependent on adequate reimbursement. ACMG is currently engaged in several activities related to improved CPT coding for genetic laboratory tests. Additionally, a timework study is currently being conducted across the United States in order to gather the necessary data to make a case to the AMA CPT coding panel that existing CPT codes do not reflect that the work of clinical geneticists is often different from the standard medical consult. Making changes to the existing CPT and reimbursement processes is a lengthy process with multiple steps. ACMG remains optimistic that with the appropriate data changes that will benefit its members can be effected.

"Banbury 3" Curriculum Meeting set for Mid-February

In October 2004, and again in February 2006, thought leaders and representatives of the major human genetics professional organizations met at the Banbury Conference Center in Cold Spring Harbor, NY, to discuss the recruitment, training and scope of practice of medical genetics physicians. The goal of the first meeting was to identify strategies to increase the number of medical genetics physician trainees. A critical issue discussed at the 2004 meeting was defining the scope of medical genetics practice; this is essential to efforts to attract additional trainees. Yet, the issue was of such complexity that it became the focus of the 2006 meeting, with attendees again representing a breadth of perspectives and stakeholder constituencies. This subsequent meeting developed a set of agreed upon Principles related to the practice of medical genetics and attendees identified a set of Points for Consideration to facilitate the provision of high quality medical genetic services, optimize the skills of medical geneticists, and position medical genetics for the future. "Banbury 3," to occur in Atlanta in late February, will reconvene the former participants and use the Principles and Points to Consider developed in 2006 as a jumping off point for developing a forward-looking core curriculum for the training of medical genetics physicians. (Reports of the 2004 and 2006 Banbury conferences can be found in *Genetics in Medicine*.)

ACMG Foundation Ramps Up Its Activities

In mid-September 2009, the ACMG Foundation— our not-for-profit arm that raises money to fund the initiatives of the College as well as a myriad of educational activities related to medical genetic services—hired J. David Cotter as its full-time Director of Development. Overall donations in 2009 topped 2008 efforts, with a 50% increase in individual giving. The ACMG Foundation is currently raising money for several initiatives that will roll out in 2010 and 2011. The first of these will be a meeting with manufacturers and providers of array-CGH testing and representatives of the FDA and New York State regulatory programs, organized by ACMGF.

For the fifth consecutive year, the Foundation is seeking applicants for its two largest awards to ACMG members: The *Genzyme/ACMGF Clinical Genetics Fellowship in Biochemical Genetics* funds a biochemical genetics fellow for a year of training, with the goal of producing a physician-scientist with cutting-edge expertise in the treatment and management of patents with inborn errors of metabolism, and specifically lysosomal storage diseases. The *Luminex/ACMGF Award* funds a one-year, member-initiated research project with the outcome of promoting safe and effective genetic testing and services. Winners of both of these awards will be announced at the ACMG Clinical Genetics Meeting in March 2010.

ACMG Continues in the News and Now Can Be Found Using Social Media

ACMG continues its outreach to the media and has enjoyed increasing exposure in the news and a growing amount of media-initiated contact. We have also moved into the world of social media with a presence on Facebook, LinkedIn, YouTube, and Twitter. ACMG will use these venues to augment its educational and advocacy missions, provide news and resources related to medical genetics and the ACMG, improve communication with and among ACMG members, and enhance the Annual Clinical Genetics Meeting experience for attendees and exhibitors.

2010 Annual Clinical Genetics Meeting

The ACMG staff, directors and members are busy preparing for the 2010 Clinical Genetics Meeting, to be held in Albuquerque, NM, March 24-28, 2010. Registration has broken all past records, with an outstanding program planned. Highlights include:

- The 41st March of Dimes Clinical Genetics Conference, "Microcephaly, Megalencephaly and Cortical Dysplasia," featuring the Annual Pruzansky Address, presented by Christopher A. Walsh MD, PhD;
- Two pre-meeting short courses: "Array Technology: What Do Results Mean in the Clinic?" and "Metabolism and Newborn Screening for the Non-Metabolic Geneticist;"
- A session co-sponsored with the Society for Inherited Metabolic Disorders, "Metabolic Causes of Autism and Neurodevelopmental Disability;"
- Presidential Plenary: "The Intersection of Medical Genetics and Informatics in the Genomics Era," to include keynote addresses on telemedicine and medical genetics, and informed consent and genetics research; and
- Taking advantage of our location in America's Southwest, two special sessions focusing on genetics, research and the Navajo Nation: 1) A pre-meeting special satellite session, "Genetics and Genetic Research: Native American Perspectives," sponsored by the Regional Genetic and Newborn Screening Service Collaboratives and their National Coordinating Center, will include leaders and representatives of the Navajo (Diné) Nation (a Navajo Medicine Man; the head of the Navajo Nation IRB; representatives of the Navajo Nation Health Department, the Navajo Supreme Court, the Diné Policy Institute, and consumers) engaging in a discussion with the medical genetics community. There is also a scientific session, "Navajos and Genetic Disease: Genetic, Medical, and Educational Issues," sponsored by the Mountain States Regional Genetics Collaborative Center.

All information about the ACMG Annual Meeting can be found on the meeting website, www.acmgmeeting.net.

New Practice Guidelines Published in *Genetics in Medicine*

Between September 2009 and January 2010 the following ACMG practice guidelines were published in the ACMG's monthly journal, *Genetics in Medicine (GIM):*

Palomaki GE, Lee JES, Canik JA, McDowell GA, Donnenfeld AE for the American College of Medical Genetics (ACMG) Laboratory Quality Assurance Committee. Technical standards and guidelines: Prenatal

Screening for Down syndrome that includes first-trimester biochemistry and/or ultrasound measurements. *Genetics in Medicine* 11(9):669-681 (September 2009)

Driscoll DA, Gross SJ for the American College of Medical Genetics (ACMG) Professional Practice and Guidelines Committee. Screening for fetal anueploidy and neural tube defects. *Genetics in Medicine* 11(11):818-821 (November 2009)

Cooley LD, Mascarello JT, Hirsch B, Jacky PB, Rao PN, Saxe D, Rao KW; A Working Group of the American College of Medical Genetics (ACMG) Laboratory Quality Assurance Committee. Section E6.5 of the ACMG technical standards and guidelines: Chromosome studies for solid tumor abnormalities. *Genetics in Medicine* 11(12):890-897 (December 2009)

Wolff DJ, Van Dyke DL, Powell CM; A Working Group of the American College of Medical Genetics (ACMG) Laboratory Quality Assurance Committee. Laboratory Guideline for Turner syndrome. *Genetics in Medicine* 12(1):52-55 (January 2010)

Information about receiving *Genetics in Medicine* podcasts can be found at http://journals.lww.com/geneticsinmedicine/Pages/podcastepisodes.aspx?podcastid=1. *GIM* can also be read on a PDA with the new "mobile view" feature.

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.

Submitted by R. Rodney Howell, MD, FACMG
ACMG Liaison to the National Advisory Council for the National Human Genome Research Institute, NIH