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

August 31, 2011

Recent Activities of
The American College of Medical Genetics

The American College of Medical Genetics (ACMG) is the professional home to nearly 1,500 board certified clinical and laboratory genetics professionals and is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetic and genomics. 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 May and August 2011.

ACMG Takes the Lead in Addressing Clinical Laboratory Issues, Including Next Generation Sequencing

ACMG Weighs in at FDA Hearing on Next Generation Sequencing

ACMG's Executive Director, Dr. Michael S. Watson represented the College at the US Food and Drug Administration's (FDA) June meeting on next generation sequencing, "Ultra High Throughput Sequencing for Clinical Diagnostic Applications: Approaches to Assess Analytical Validity." In his public testimony, Dr. Watson acknowledged how hard it has been for regulatory agencies to keep up with the rapid pace of development of technology and genetic testing and offered input, recommendations, and an overview of actions that ACMG is taking in the area of whole genome analysis and next generation sequencing including:

1. An official Task Force on Whole Genome Analysis (WGA) and Next Generation Sequencing (NGS), led by Sherri Bale, PhD, FACMG, has been asked to broadly examine the types of actions that will be needed as these technologies move forward, and to make recommendations to ACMG and its committees;
2. A short course on NGS will be offered just prior to the 2012 Annual Meeting;
3. Recommendations addressing how families and patients could be informed and consented for testing in ways that are understandable to them will be developed;
4. Two practice guidelines were published in *Genetics in Medicine* in July (see *Genetics in Medicine* section of this report); one focuses on technical standards for interpreting and reporting whole genome copy number studies and the other makes recommendations for the design and performance of these whole genome copy number analyses. The latter guideline was prepared in response to a very successful stakeholders' meeting, hosted and funded by the ACMG Foundation, that brought together state and national regulators, industry, and the medical genetics community in the summer of 2010 for a frank dialogue on the future of regulation of genetic testing. The other guideline is a product of ACMG's Laboratory Quality Assurance (Lab QA) Committee.
5. ACMG's Lab QA Committee is developing a technical standard and guideline on NGS;
6. The College's CAP/ACMG Resource Committee is developing proficiency testing programs for NGS; and
7. Medical genetics training programs are being adapted to include these new technologies, with other educational offerings are forthcoming.

ACMG also suggested that the FDA will need to find a reasonable balance between what manufacturers can validate and what the laboratory must validate related to the reportable targets, and that we must put more weight on post-market surveillance, which is a common

feature of orphan drug clinical trials. This recognizes that some diseases are so rare that products are made available to patients with less robust data but with systems in place to capture data so we know they continue to do what they appeared to be doing when first approved.

aCGH Team Receives FDA Commissioner's Award

ACMG's Executive Director, Dr. Michael S. Watson, along with members of the FDA's Array CGH Review Team recently received the FDA Commissioner's Special Citation award recently. The award was given for "Outstanding initiative in identifying and addressing complex regulatory and scientific issues in DNA microarray-based cytogenetic devices for postnatal diagnosis of developmental delay." The award recognized the team for organizing FDA's 2010 public hearing on the use of cytogenetic array technologies and its coordination of a series of subsequent meetings that contributed to the FDA's planning of regulatory approaches to these new technologies.

Insurance Coverage for Cytogenomic Microarray in Jeopardy

ACMG is working with the International Standards for Cytogenomic Arrays (ISCA) Consortium to improve coverage for postnatal cytogenomic microarray (CMA) testing by payers. Both organizations have issued recommendations stating that CMA testing is standard of care for the evaluation of unexplained developmental delay, autism, and/or multiple congenital anomalies. The ISCA Consortium has developed a survey to formally document the frequency of denials for testing and is also collecting information to document the real-world clinical impact of microarray testing on medical management. ACMG stands ready to work with ISCA on next steps, as appropriate.

ACMG's New Online Live Learning Center Brings Genetics and Genomics Education to Health Professionals

ACMG has just launched an Online Live Learning Center, which will bring genetics and genomics education to a variety of healthcare professionals. This on-demand resource features educational programs from ACMG's live learning activities, such as the Genetics Review Course and Annual Meeting Short Courses, to health professionals who are practicing in, or who are interested in, genetics and genomics. The Learning Center content will be expanded throughout the year, but particularly after each Annual Meeting and other live ACMG programs.

Presently, recorded content from the 2011 ACMG Annual Clinical Genetics Meeting Short Courses, a selection of sessions from the 2011 Annual Meeting, and the complete 2011 ACMG Genetics Review Course in audio, fully synchronized to the onsite slide presentations are available in streaming media format or as downloads to a portable media player. There is also the ability to download the sessions in MP3 format to a mobile device for education-on-the-go. Go to the ACMG Live Learning Center Website (www.acmg.net, under the "Resources" tab) to access current offerings and learn about purchase options.

ACMG Offers Searchable Online Library of Annual Meeting Abstracts

The ACMG Online Abstract Library houses all abstracts presented at the 2006, 2007, 2008, 2009, 2010, and 2011 ACMG Annual Clinical Genetics Meetings. It is searchable by title, author, keywords, topics, and abstract numbers. The ACMG Abstract Library can be accessed at www.acmg.net, under the "ACMG Events" tab.

AMA Adds ACMG/Genetics to "Choosing A Medical Specialty" Webpage

The American Medical Association (AMA) has added the ACMG to its "Choosing A Medical Specialty" webpage. This page is designed to help medical students explore various specialty options. The link to ACMG mentions the new ACMG Foundation Summer Genetics Scholars Program and the new ACMG Medical Student Interest Groups. Finally, a newly revised edition of ACMG's brochure, *Graduate Medical Education in Medical Genetics* has just been released. It is available on our website (www.acmg.net, under the "Education" tab) and will be distributed to academic deans and medical genetics course directors in US medical schools. All of these initiatives are part of ACMG's on-going efforts to inform students about, and attract them to, a career in medical genetics.

ACMG Student Interest Groups Forming in Medical Schools Across the USA

Earlier this year, the College recently formed a Student Interest Groups Program as an organized way to reach and attract the interest of potential medical genetics trainees. The ACMG Student Interest Groups Program is open to medical, graduate, and genetic counseling students. Beyond providing an institution's Student Interest Group with recognition by the College, establishing this connection optimizes two-way communication between the College and students enthusiastic about the field of medical genetics. The program offers targeted resources, educational materials, and opportunities for networking; further information can be obtained by contacting Denise Calvert

(dcalvert@acmg.net), ACMG's Membership Services Coordinator. To date, we have welcomed the following ACMG Student Interest Groups:

- ACMG Student Interest Group of Tufts University Medical School
- ACMG Student Interest Group of University of Kansas Medical Center
- ACMG Student Interest Group of David Geffen School of Medicine (UCLA)
- ACMG Student Interest Group of Wayne State University School of Medicine

Inaugural ACMG Foundation Summer Genetics Scholars Program (SGSP) is an Enormous Success

ACMG and the ACMG Foundation joined forces this summer to implement an exciting one-of-a-kind program to introduce rising 2nd year medical students to the field of medical genetics. Twenty students from 19 institutions participated in the 6-8 week program, which placed each student with a medical geneticist mentor to experience the gamut of activities that comprise the field— from clinics to diagnostic laboratories, research, and collaborative activities with other departments, where applicable. The overall goal of the program is to introduce students to hands' on medical genetics activities earlier in their medical education and attract them to the field for residency training and beyond.

Response to the SGSP far exceeded all expectations and we are working to double the number of students who will be able to enroll in 2012. Applications for summer 2012 are available on the ACMG Foundation website (www.acmgfoundation.org, under the "ACMGF Programs" tab) and are due on September 15th. There are plans to keep in touch with all program participants via social media and the ACMG Foundation will host the 2011 Summer Scholars and their mentors in a culminating event at the 2012 ACMG Annual Meeting.

The 2011 participants represented the following institutions:

Emory University School of Medicine
University of Maryland School of Medicine
University of Alabama Birmingham
Tufts Medical Center
Children's Hospital Boston, Division of Genetics
University of North Carolina at Chapel Hill
Nationwide Children's Hospital Research Institute
Greenwood Genetic Center
Loma Linda University Medical Center
UCLA Department of Human Genetics
Cedars-Sinai Medical Center
University of Miami, Miller School of Medicine
Vanderbilt Clinical Genetics Program
Mount Sinai School of Medicine
University of Oklahoma College of Medicine
UC San Francisco
George Washington University School of Medicine and Health Services
DuPont Hospital for Children
Case Western Center for Human Genetics

All SGSP students have completed their experiences, and sample feedback highlights just how meaningful the program was to two participants:

"The inpatient consultations allowed me to experience the whole process a geneticist would go through with a patient, from identification and initial assessment of symptoms to a diagnosis and long-term care plan. ... I had the opportunity to work with a variety of medical geneticists who were at varying stages of their training and from different medical backgrounds. This wide exposure allowed me to gain an in depth understanding of what a career in medical genetics is like." (Jessica)

"There was not a dull day in the past 6 weeks; I learned something new with each and every patient. The incredible amount of knowledge and wisdom of the geneticists I have worked with has been motivating and inspiring. ... Getting this experience early in my medical education will give me confidence and experience that many others won't have when I begin rotations in my third year of medical school. I learned a lot from interacting with each patient that I wouldn't have necessarily learned by just watching." (Emily)

ACMG Foundation Updates

Expanded Genzyme Clinical Genetics Fellowship in Biochemical/Metabolic Genetics Announced for 2012-2013

For the first time since its inception, the Genzyme Clinical Genetics Fellowship in Biochemical/Metabolic Genetics will be supporting two fellows a year, beginning with the 2012-2013 cycle. This doubles their commitment to: advancing education, research and standards of practice in medical genetics; developing and expanding clinical and laboratory expertise in medical genetics in the United States; and initiating and sustaining a broad-based infrastructure for industry funding of high quality projects in the fields of medical genetics. A record number of applications were received for $75,000 awards, with the deadline just closing on August 15th.

ACMG Foundation Board Undergoes Strategic Planning and Announces Task Force on Genetics Education

The ACMG Foundation Board of Directors travelled to Dallas, TX in May for a two-day, professionally facilitated, strategic planning initiative. While the entire process will be implemented over time, goals were identified and are now being operationalized by the Foundation's Board and staff. These include defining specific fundraising strategies, as well as key programs, with genetics education and workforce issues taking priority.

For over three decades, federal advisory bodies have reported on the inadequacies in genetics knowledge among non-genetics trained health care providers, as well as the precarious state of the current workforce of medical genetics trained providers given the rapid growth of the field. Likewise, the public's level of genetic literacy is similarly poor, yet people will increasingly be asked to make decisions about the types of genetic information they might want and how they might act on it. Furthermore, we expect that it will become even more difficult for those without a knowledge base on which to build to keep up with the field. Given the breadth and depth of this problem, ACMG has formed a Task Force on Genetic Education and Training, which Dr. Bruce Korf has agreed to chair. It has already begun to identify all activities within ACMG and the ACMGF that are aimed at providing medical genetics education from medical school to residency training and practice for geneticists and non-genetics trained providers in other specialties or in primary care health care delivery. The Task Force had its first meeting in late July and began identifying major gaps in the educational resources available and considering some strategies that might be pursued in both long-distance learning and point-of-care education that could begin to fill these gaps.

With funding from one of our ACMGF affiliated industry partners, the Abbott Corporation by way of the Abbott Fund, we've had the resources to bring together experts across many areas of genetics education (e.g., cancer genetics, neurogenetics, cardiogenomics, etc.) develop the competencies that will be required of medical geneticists and other health care providers; these will be published in *Genetics in Medicine* before the end of the year. Furthermore, the Accreditation Counsel for Graduate Medical Education (ACGME) that oversees all medical residency training in the United States has made these competencies one of the models for their Milestones Project, an effort aimed at creating the next system by which Residency training will be assessed in the future. Next steps include identifying those longer-term needs and translating them into a focused fund-raising campaign. We hope that these efforts provide the basis for the ACMG Foundation Board to develop a plan on which we are able to realize our expanding goals for fund-raising.

ACMG Foundation Board Names Six New Directors

The ACMG Foundation recently welcomed six new directors. The Foundation's Directors are active participants in our fund-raising activities and serve as advocates for the Foundation by advancing its policies and programs. The new Directors are: **Thomas Erickson, CFA**, Co-founder and Partner, BlueStream Ventures; **Rick Martin, MD, FACMG,** Medical Director of Shire Human Genetic Therapies; **Lisa G. Shaffer, PhD, FACMG,** President of PerkinElmer's Signature Genomics; **Brian E. Ward, PhD, FACMG,** past COO of On-Q-ity Inc.; **Judy Yu, PhD,** Associate Director, Scientific Affairs, Global Marketing of Abbott Molecular; and **Gail E. Herman, MD, PhD, FACMG** is an *ex-officio* director by virtue of being the new President-Elect of the ACMG Board.

Genetics in Medicine Updates

***Genetics in Medicine* Announces 35% Increase in Impact Factor**

The Thomson Reuters Impact Factor Journal Citation Reports has increased the impact factor of the ACMG's peer-reviewed medical genetics and genomics journal, *Genetics in Medicine* (GIM) from 3.922 to 5.280. This places *GIM* in the top 15% of all Genetics journals. (A journal's impact factor is an objective measure of the world's leading journals based on articles' cited references and is often considered a measure of a journal's overall successful performance and relevance of a journal to its field.) Since its inception, *GIM's* goal has been to be the leading journal in the field of Clinical Medical Genetics, focusing on the wide dissemination of evidence-based articles that address a broad range of issues. Increasingly, genetics and genomics are permeating medicine; thus, we strive to publish material that matters not only to those in clinical genetics but also to non-genetics specialists who

need to understand how to incorporate genetic advances into their fields. *GIM,* which has been published since 1998, also recently announced that beginning in January 2012, it would be published by the prestigious Nature Publishing Group.

New Clinical and Laboratory Practice Guidelines

The following ACMG clinical and laboratory practice guidelines were published in the College's monthly journal, *Genetics in Medicine (GIM)* between May and August 2011:

Wang RY, Bodamer OA, Watson MS, Wilcox WR, on behalf of the ACMG Work Group on Diagnostic Confirmation of Lysosomal Storage Diseases. **Lysosomal storage diseases: Diagnostic confirmation and management of presymptomatic individuals.** *Genet Med* 13(5):457-484 (May 2011)

Goldman JS, Hahn, SE, Catania JW, LaRusse-Eckert S, Butson, MB, Rumbaugh M, Strecker MN, Roberts JS, Burke W, Mayeux R, Bird T. **Genetic counseling and testing for Alzheimer disease: Joint practice guidelines of the American College of Medical Genetics and the National Society of Genetic Counselors** *Genet Med* 13(6):597-605 (June 2011)

Toriello HV, for the Policy and Practice Guideline Committee of the American College of Medical Genetics. **Policy statement on folic acid and neural tube defects** *Genet Med* 13(6):593-596 (June 2011)

Kearney HM, South ST, Wolff DJ, Lamb A, Hamosh A, Rao KW: A Working Group of the American College of Medical Genetics. **American College of Medical Genetics recommendations for the design and performance expectations for clinical genomic copy number microarrays intended for use in the postnatal setting for detection of constitutional abnormalities**. *Genet Med* 13(7):676-679 (July 2011)

Kearney HM, Thorland EC, Brown KK, Quintero-Rivera F, South ST: A Working Group of the American College of Medical Genetics (ACMG) Laboratory Quality Assurance Committee. **American College of Medical Genetics standards and guidelines for interpretation and reporting of postnatal constitutional copy number variants**. *Genet Med* 13(7):680-685 (July 2011)

Mascarello JT, Hirsch B, Kearney HM, Ketterling RP, Olson SB, Quigley DI, Rao KW, Tepperberg JH, Tsuchiya KD, Wiktor AE: A Working Group of the American College of Medical Genetics (ACMG) Laboratory Quality Assurance Committee. **Section E9 of the American College of Medical Genetics technical standards and guidelines: Fluorescence in situ hybridization.** *Genet Med* 13(7):667-675 (July 2011)

Prior TW, Nagan N, Sugarman EA, Batish SD, Braastad C. **Technical standards and guidelines for spinal muscular atrophy testing.** *Genet Med* 13(7):686-694 (July 2011)

ACMG in the News…Again

ACMG continues to enjoy increasing prominence as a source of trusted expert opinion by print and non-print news media, covering such topics as newborn screening (NBS), gene patents, direct-to-consumer genetic testing, testing minors for adult onset conditions and much more.

Of particular note is a one-hour radio, syndicated national program, "The Remarkable Public Health Program of Newborn Screening," co-sponsored with the MCHB/HRSA-funded National Coordinating Center for the Genetic and Newborn Screening Service Collaboratives (NCC). NBS is a vital public health program and was just named one of the most significant, lifesaving and important public health programs in the United States of the past 50 years. On the broadcast radio program (aired originally on *Life, Love & Health*, America's most listened-to daily health, prevention and wellness program) ACMG and NCC experts and a parent discuss how it is now possible to diagnose, treat and actually prevent the negative outcomes of death and disability of these diseases. The conversation stressed the importance of all expectant parents becoming versed in the nuances and lifesaving benefits of NBS, including proactively asking about NBS and their baby's test results. The program is available on the ACMG website at http://www.acmg.net/LLH-AFN_052711.mp3.

The NBSTRN has a New Website

The URL remains the same (www.nbstrn.org), but the Newborn Screening Translational Research Network (NBSTRN), which is a contract between ACMG and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NIH) has a new state-of-the-art website. (The NBSTRN seeks to improve the health outcomes of newborns with genetic or congenital disorders through an infrastructure that provides the research community access to robust newborn screening resources.) The site has separate sections for Researchers, NBSTRN Workgroups, and Parents/Families, but features such research tools as a Virtual Repository of Dried Blood Spots, a Laboratory Performances Database, a Long-Term Follow-Up Data Collection module and Disease Registries.

Annual Meeting Updates

Mark Your Calendar Now for the 2012 Annual Clinical Genetics Meeting

The 2012 Clinical Genetics Meeting will be held in Charlotte, NC from March 27-31, 2012. Each year the breadth and number of program offerings increase, and 2012 will continue that tradition. Two short courses will be offered on Tuesday March 27—Next Generation Sequencing: Clinical Utility, Laboratory Implementation and Bioinformatics Analysis; and Cancer Genetics. The March of Dimes Clinical Genetics Conference will once again be incorporated into the meeting, and the last day will feature a joint plenary session with the Society of Inherited Metabolic Disorders (SIMD), whose conference will follow the College meeting. Finally, this year's meeting will incorporate a record number of events for students, residents and fellows at various stages of pursuing careers in medical genetics. Further details, including abstract submission information, will be available in the early fall at www.acmgmeeting.net.

2013 and 2014 ACMG Annual Meeting Dates and Locations Announced

For those planning ahead, the 2013 ACMG Annual Clinical Genetics Meeting will be held March 19 - 23 in Phoenix, Arizona and the 2014 ACMG Annual Clinical Genetics Meeting will be held March 25-29 in Nashville, Tennessee. Both cities have previously hosted ACMG Annual Meetings and both have expanded or new convention centers that will provide outstanding facilities for our growing meeting.

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