



February 1, 2011

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 2010 and January 2011.

ACMG Celebrates Twenty Years of Translating Genes into Health!

In 2011 we celebrate the achievements of two decades since a group of visionary physician geneticists and doctoral level laboratory practitioners founded the American College of Medical Genetics. ACMG has grown tremendously in both size and scope since 1991. We speak for our community in the specialty of medical genetics with organizations and agencies concerned with medical research and service, certification, and regulatory issues. Along with a significant growth in membership, our annual conference has grown since its inception from 600 attendees at our first meeting in 1992 to just over 1,800 at our last conference!

As one of the 20th anniversary events planned throughout the year, ACMG will publish a special issue of its journal, *Genetics in Medicine*, to coincide with our Annual Clinical Genetics Meeting and even more importantly, to recognize and give thanks to individuals and organizations that have contributed to our success throughout the years. We also look forward to the exciting future of our profession.

The ACMG Foundation will also celebrate its 20th Anniversary throughout 2011. The ACMG Foundation is a community of friends and supporters who understand the importance of medical genetics in healthcare. To support the future of medical genetics and to accomplish our important mission—better health through genetics—the Foundation has launched a “20 More...” campaign to help the ACMG flourish for another 20 years.

2011 Annual Clinical Genetics Meeting

The 2011 Clinical Genetics Meeting will be held in Vancouver, BC, March 16-21, 2011, and it promises to be our biggest meeting to date. The meeting will include the March of Dimes Clinical Genetics Conference, featuring new understandings of connective tissue disease. There will also be two pre-meeting short courses, “New Insights into the Evaluation of Individuals with Intellectual Disability” and “Preparing for an Expanding Range of Adult Genetic Consultations,” both coordinated and leading experts. In addition, the National Coordinating Center for the Genetic and Newborn Screening Services Collaboratives, (a MCHB/HRSA, Genetic Services Branch-funded cooperative agreement, housed at ACMG) will be sponsoring a Community Conversation, “Screening for CPT1 in First Nations’ Populations.” Further details are available at www.acmgmeeting.net.

New ACMG Foundation Summer Genetics Scholars Program Promises to Grow Medical Genetics Pipeline

The 2011 Summer Genetics Scholars Program is an exciting new opportunity for rising second-year medical students, sponsored by ACMG and its Foundation. The purpose of the

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program is to identify and encourage highly qualified medical students who are early in their training to consider careers in medical genetics by introducing them to the breadth of day-to-day opportunities that comprise the professional lives of medical geneticists. This will occur through hands-on experiences in clinical and laboratory genetics, research, and services across the United States. Interested students will choose leading participating medical genetics institutions, approved by ACMG, for a 6-8 week period during the summer between their first and second years of medical school, and they will work under the direct supervision of experienced medical geneticists. Applicants must be enrolled in a US medical school and in good standing at the time of the application; all Principal Mentors must be ACMG members. Stipends will be available to students accepted into the program. Applications are due February 4, 2011, with final selection to be made by mid-late February.

ACMG Comments on NIH Genetic Testing Registry (GTR) at November Meeting

In September we reported that ACMG responded by letter to the NIH regarding the Genetic Testing Registry (GTR) it is developing as an online resource that would provide a “centralized location for test developers and manufacturers to voluntarily submit test information such as indications for use, validity data and evidence of the test’s usefulness.” While ACMG and its members recognize the importance of having accurate information on genetic testing readily available to health professionals and lay public, ACMG outlined several concerns with regard to the GTR as proposed in its original letter. On November 2, ACMG President, Bruce Korf, MD, PhD, FACMG spoke at a public meeting on the GTR, held in Washington DC. His remarks can be found at <http://www.acmg.net/StaticContent/Foundation/GTR%20Remarks.pdf>.

Cutting-Edge NBSTRN-CC Activities Continue

In September we reported that during the second year of the Newborn Screening Translational Research Network-Coordinating Center (NBSTRN-CC) contract between ACMG and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NIH), its list of accomplishments grew exponentially. As we continue in year three of the contract each of the committees and workgroups remains very active.

- **The NBSTRN Standing Committee** is responsible for oversight of workgroup activities and charges, and for collaborations and relationships between NBSTRN and key partners such as the RCs, NICHD, HRSA, APHL, Genetic Alliance, etc. The workgroup is assisting in planning a Coordinating Meeting, June 15-17, 2011, to ensure that no duplication of effort occurs in the Newborn Screening Community, report on the progress made by the NBSTRN, and share our plans for the rest of the contract.
- **The Laboratory Network Workgroup** is dealing with issues related to dried blood spot retention, storage, and use in the NBSTRN Virtual Repository; developing a laboratory network for new test and technology pilots; and planning for the implementation of nursery-based newborn screening. This workgroup has regular conference calls and two face-to-face meetings per year.
- **The Clinical Centers Workgroup** has been developing the uniform and disease specific long-term follow-up (LTFU) data sets and addressing ownership of data, data sharing/storage, IT issues, consumer representation, increasing patient involvement, and development of a clinical survey. This workgroup has regular conference calls to finalize the LTFU data sets and has two face-to-face meetings for the year.
- **The Bioethics & Legal Issues Workgroup** is developing a process to assess NBSTRN services and resources, identifying policies and procedures required for participation within the network, outlining policies for data sharing and access, and discussing ethical and legal implications of NBS research on a national level. The workgroup has created a series of FAQs aimed at new investigators, answering questions about newborn screening-related research. The FAQs will be available on the NBSTRN website. This workgroup has also drafted User Agreements for accessing the NBSTRN website and the Virtual Repository, and is working on an IRB survey aimed at gathering state IRB information related to newborn screening research. This workgroup has monthly conference calls and two in-person meetings per year.
- **The Information Technology Workgroup** has focused its activities on the elements of the Virtual Repository and the infrastructure necessary to support the other workgroups’ activities. Two tools are being developed: One is for a bedside LTFU data capture tool and the other is for a registry system. This workgroup has begun a survey of state newborn screening IT systems that will continue throughout this year. The workgroup has regular conference calls and meets in-person twice a year.

- **LSD and SCID Pilot Projects.** The NBSTRN-CC has been contracted to assist with the development of meetings related to the LSD (lysosomal storage disease) and SCID (severe combined immunodeficiency) Pilot Projects. The LSD workgroup met twice last year and is meeting again in Las Vegas in February 2011. Many workgroup members will also attend the LSD World Meeting. The agenda will include establishing provider networks, discussions of informed consent issues, developing diagnostic and long-term follow-up criteria, and IT/Informatics development issues. The SCID working group has been having monthly conference calls since June 2010; they met in-person in September 2010 and will meet again at the end of March 2011 to ensure all goals of the contract have been met. This workgroup has established screening algorithms and data is being collected in California, New York, Louisiana, and Puerto Rico.

The NBSTRN-CC currently administrates two subcontracts:

Development of the Virtual Repository of Dried Blood Spots

Through a subcontract with 5AM Solutions, Inc., the NBSTRN is developing a centralized, web-based source for access to samples for newborn screening related research. This virtual repository links information from participating states and provides a de-identified view of stored dried blood spots, allowing researchers to browse and query for samples. For public health departments, the virtual repository provides the ability to review and manage sample requests, configure approved orders, track shipments, and view researcher responses to the samples they have received. The demo for the Virtual Repository is complete and the NBSTRN team will begin piloting the Virtual Repository within the next few months.

Adaptation of the R4S Laboratory Performance Database

Through a subcontract with the Mayo Clinic, the NBSTRN is adapting the Region 4 Stork Laboratory Performance Database to collect information from the SCID and LSD Pilot Projects.

Hearing Loss, Genetics & Your Child Brochures Now Available in English and Spanish

ACMG and the National Coordinating Center for the Regional Genetic and Newborn Screening Services Collaboratives partnered to develop a free brochure to increase genetics referrals as part of comprehensive follow-up to a diagnosis of hearing loss. The goal is to have the brochures available at the point of care—waiting rooms in primary care settings, ENT and Audiology centers—and other settings where infants and children with hearing loss are most often evaluated. The brochure was designed to both assist medical professionals in conveying information about the purpose and process of genetic evaluation and to increase parents’ comfort with genetic services by empowering them with information about the importance of genetic services in the etiologic diagnosis of hearing loss, thus complementing the referral. Developed by a team of medical geneticists, genetic counselors, audiologists, parent advocates, and health science information specialists led by Kathleen Arnos, PhD, FACMG of Gallaudet University, the brochure is written at a low-literacy level and is available in both English and Spanish. Downloadable brochures are available online at www.nccrcg.org (under the Resources tab) at no cost and contain modifiable space for adding local resource information. Print copies may be ordered by contacting Matthew Tranter (mtranter@acmg.net).

Nature Publishing Group to Begin Publication of Genetics in Medicine (GIM) in 2012

One more piece of evidence that the work of the College during the last 20 years has taken us to new heights occurred when it was time to renew our journal publication contract. The search committee declared that it was a “remarkable experience to see the value that several publishers recognized in *GIM*.” The credit for this accomplishment belongs to the founding editor, Dr. Richard King, and to the current editor, Dr. Jim Evans, as well as the associate editors and many contributors to the journal. The Nature Publishing Group was interested in *Genetics in Medicine* as a sibling publication to Nature Genetics, focusing on medical genetic issues.

New Practice Guideline and Special Newborn Screening Supplement Published by Genetics in Medicine

Between September 2010 and January 2011, the following ACMG practice guideline was published in the College’s monthly journal, *Genetics in Medicine (GIM)*:

Manning M, Hudgins L: for the Professional Practice and Guidelines Committee. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. *Genetics in Medicine*. 12(11):742-745 (November 2010)

In addition, the December 2010 issue of *Genetics in Medicine* was accompanied by a substantial Supplement devoted exclusively to **Newborn Screening Long-Term Follow-Up**.

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.

ACMG in the Media

ACMG leaders continued their prominent presence in both the print media and on the airwaves during the past months. Additionally, articles published in *Genetics in Medicine* are increasingly being quoted. Examples of ACMG in the news include: A *Self* magazine story, *Who Owns Your Genes?*, a *Men’s Health* piece on DTC genetic testing; a feature on family health history as the gold standard for predicting disease, on **ABCNews.com**; an article on gene patents for **Newsweek International**; and a touching story about living with SMA and SMA carrier screening, in the *Omaha World-Herald*. Finally, ACMG’s Executive Director, Michael Watson, PhD, FACMG, discussed issues and risks related to proposed regulatory oversight and reimbursement of diagnostic genetic tests on the television program “**BioCentury This Week**.” He was accompanied by a co-panelist from the American Clinical Laboratory Association. ACMG has also been interviewed on background and had its messages conveyed to the public by *Women’s Health*, *ABC News*, *Martha Stewart Whole Living* magazine, and **MSN Health**. Links to all of these items can be found in the Newsroom section of the ACMG website at http://www.acmg.net/AM/Template.cfm?Section=Press_Room.

2011 ACMG Genetics Review Course

The 2011 ACMG Genetics Review Course will be held June 2-5, 2011 at the Hilton DFW Lakes Executive Conference Center, Grapevine, Texas. Detailed program information, online registration and hotel information will be available at www.acmg.net in February 2011.

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