



May 10, 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 mid-January and mid-May, 2010.*

#### Annual Meeting Updates

ACMG held its 2010 Annual Clinical Genetics Meeting in March in Albuquerque, New Mexico, with unexpectedly high growth in all areas. Professional attendance showed a 5% increase over 2009; attendees came from 46 states and Puerto Rico, with 8% of attendees from 25 countries outside the United States. The March of Dimes portion of the program featured the Genetics of Microcephaly, Megalencephaly and Cortical Dysplasia, and there was a popular half-day joint plenary session with the Society for Inherited Metabolic Disorders discussing Metabolic Causes of Autism and Neurodevelopmental Disability. A free webcast from our 2010 Annual Clinical Genetics Meeting, from the Presidential Plenary session on the "Intersection of Medical Genetics and Informatics in the Genomics Era" is available on the ACMG website at [www.acmg.net](http://www.acmg.net).

#### American College of Medical Genetics Foundation Awardees Honored

It has become a tradition for the ACMG Foundation to present a large number of awards during the Annual Meeting. **David L. Rimoin, MD, PhD** of Cedars-Sinai Medical Center in Los Angeles was presented the Foundation's second annual **Lifetime Achievement Award**, which is its highest honor and recognizes an individual who has demonstrated outstanding leadership in the field of medical genetics and who exemplifies the highest standards of the profession over a lifetime. A native of Montreal Canada, Dr. Rimoin was recognized for his decades of contributions to genetic medicine; for his research into short stature, skeletal dysplasias and heritable disorders of connective tissue; and for his leadership in organizing the field of medical genetics, including establishing the American Board of Medical Genetics, and the College and its Foundation. Dr. Rimoin was applauded by colleagues, former students, friends and family for his role as one of the true giants of medical genetics whose enormous contributions in every facet of the field, including teaching, have inspired geneticists around the world. Dr. Rimoin is presently Professor of Pediatrics, Medicine, and Human Genetics at the David Geffen School of Medicine at UCLA and Steven Spielberg Chair and Director of the Medical Genetics Institute at Cedars-Sinai Medical Center.

Another award only in its second year is the **ACMG Foundation's Richard King Trainee Award**, for the best research publication in *Genetics in Medicine* in a given year. The 2010 recipient, **Marwan Khaled Tayeh, PhD** was awarded \$1,500 for his manuscript "Targeted comparative genomic hybridization (CGH) array for the detection of single- and multi-exon gene deletions and duplications," which was published in the April 2009 issue of *Genetics in Medicine*. This award was instituted by the ACMG Foundation to encourage ABMG trainees in

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their careers and to foster the publication of quality research in *Genetics in Medicine*. It is named for Dr. Richard King in recognition of his instrumental role in creating *Genetics in Medicine* and serving as the journal's first Editor-in-Chief.

**Dr. Marilyn Li, MD, PhD**, Associate Professor of Pediatrics at the Tulane Medical Center, in New Orleans, Associate Director of the Hayward Genetics Center, Director of the Cytogenetics and Laboratory and Molecular Diagnosis Laboratory at the Tulane University School of Medicine and the Director of the Genetics/Genomics Core Laboratory of the Louisiana Cancer Research Consortium was the sixth recipient of the \$100,000 **Luminex/ACMGF Award**, established to promote safe and effective genetic testing and services, including the development of research guidelines. In response to the growing interest of applying microarray technology in cancer diagnosis, Dr. Li initiated and organized, along with Dr. Anwar Iqbal of University of Rochester Medical Center (URMC) and Dr. Charles Lee of Harvard Medical School, the Cancer Cytogenomics Microarray Consortium (CCMC). Dr. Li and her co-PI Dr. Iqbal will study the efficacy of microarray-based testing for detection of genetic aberrations in hematologic malignancies.

The 2010-2011 **Genzyme/ACMG Foundation Clinical Genetics Fellowship in Biochemical Genetics** was awarded to **Ayman W. El-Hattab, MD**, a Medical Genetics fellow at Baylor College of Medicine in Houston, Texas. Also in its sixth year, this generous award encourages the recruitment and training of physicians in clinical biochemical genetics by granting \$75,000 to sponsor a trainee's first year of fellowship following residency. Dr. El-Hattab will enter the Medical Biochemical Genetics fellowship program at Baylor College of Medicine in 2010-2011; the award will also support Dr. El-Hattab's study of glucose kinetics in patients with MELAS syndrome via the stable isotope technique.

**Alecia Willis, PhD** was honored as the 2010 recipient of the **Signature Genomic Laboratories Travel Award** for her platform presentation, "Examination of Data from the Personal Medical Genomic Profile in a Control Population." This \$1500 award is given to a selected student, trainee or junior faculty ACMG member whose abstract submission is chosen as a platform presentation during the ACMG Annual Clinical Genetics Meeting. Dr. Willis is an Assistant Professor, Department of Molecular and Human Genetics, and Assistant Laboratory Director, Medical Genetics Laboratories, Baylor College of Medicine.

ACMG congratulates all of our 2010 awardees. More information on the ACMG Foundation and applications for these and other awards can be found on [www.ACMGFoundation.org](http://www.ACMGFoundation.org)

#### ***ACMG Foundation Development Committee Names C. Scott Palubiak, Chair***

During this important time of rapid growth, the ACMG Foundation Development Committee invited Mr. Scott Palubiak, ACMG Foundation Board Member since 2007 and Vice President at PerkinElmer, Inc. to serve as its new Chairman. In this position, Mr. Palubiak follows in the footsteps of Dr. Richard King, the Development Committee's first chair, who recently retired.

This is an exciting time for both the Development Committee and the ACMG Foundation because they are writing their first comprehensive fundraising strategic plan and rewriting the Foundation's mission and vision statements. Mr. Palubiak's business, marketing, and management expertise provides the insight and foresight necessary to propel the Foundation and our fundraising efforts for the next half-decade. As Vice President of Newborn Screening and Child Health for the genetic screening business unit of PerkinElmer, Inc., he joined the company in 1990 after holding several senior level sales and marketing positions with multinational corporations and venture capital backed start-ups. Among his most notable achievements, Mr. Palubiak is recognized for leading a highly effective lobbying campaign at both the state and federal level for the expansion of newborn screening. This effort also contributed to the successful passage of the Children's Healthcare Act of 2000, signed by President Clinton.

#### ***2011 Annual Clinical Genetics Meeting***

The 2011 Clinical Genetics Meeting will be held in Vancouver, BC, March 16-21, 2011 and will include the March of Dimes Clinical Genetics Conference. Further details, including abstract submission information, will be available in the early fall at [www.acmgmeeting.net](http://www.acmgmeeting.net).

#### ***Another "Date to Save" in 2011***

The 2011 ACMG Genetics Review Course will be held June 2-5, 2011 at the Hilton DFW Lakes Executive Conference Center, Grapevine, Texas. Registration and hotel information will be available January 2011 at [www.acmg.net](http://www.acmg.net).

#### **ACMG's Media Outreach Continues to Offer the Public Valuable Medical Genetics Information**

- ACMG Executive Director Michael Watson, PhD and Board of Directors Past President Joe Leigh Simpson, MD were featured in *The New York Times* story, "Firm Brings Gene Tests to Masses," on January 28.

- Also on January 28, Dr. Watson was featured in the ABC News article, “Born Without Eyes and Ultrasound Couldn’t See: Ultrasounds Can Detect Hundreds of Deformities, But Not Babies Missing Eyes.”
- Dr. Watson and ACMG Foundation President R. Rodney Howell, MD, are quoted in a *Wall Street Journal* article on carrier screening that appeared on February 14, “Discovering the Unseen Risks of Genetic Diseases.”
- ACMG’s newborn screening recommendations were featured in the March issue of *Parents* magazine, “The Test That Saved My Baby’s Life: Screening every newborn for rare genetic diseases can seem unnecessary and invasive -- unless your child is the one who is diagnosed.” ACMG Board Vice President for Laboratory Affairs, Piero Rinaldo, MD, PhD, also was featured in the article. *Parents* magazine is the largest parenting magazine in the US with 15.3 million adult readers month, and its website has 2.9 million unique visitors each month.
- ACMG also submitted a firm response to a CNN.com story about Newborn Screening Residual Dried Bloodspots.

### **ACMG Lauded Recent Federal Court Ruling of Gene Patents as Invalid**

As one of the original plaintiffs in the anti-gene-patenting lawsuit filed by ACLU and other in 2009, ACMG is pleased with the US District Court’s ruling that genes are “unpatentable.” ACMG believes that the outcome of this case is likely to have far-reaching positive implications for physicians, researchers and patients. The invalidation of gene patents will allow patients to get second opinions on test results, encourage quality improvement of current testing, allow researchers to develop new and better methods of testing and decrease costs of laboratory testing, representing a huge victory for patient care.

ACMG was the first professional medical association to establish a position against gene patenting. In its 1999 *ACMG Position Statement on Gene Patents and Accessibility of Gene Testing*, which was reaffirmed in 2005, ACMG stated that, “It is the American College of Medical Genetics’ position that genes and their mutations are naturally occurring substances that should not be patented.” The decision by the Federal District Court for the Southern District of New York was a direct response to response ACMG’s and the other plaintiffs’ request for summary judgment on the question of whether seven patents relating to the BRCA1 and BRCA2 genes are naturally occurring substances and, therefore, not patentable. The basic legal question was whether “isolated” DNA is “markedly different” from naturally occurring DNA. The court concluded that, even if there are differences in structure and function, it is not. The court found that the patented genes were merely purified and not changed in a way that would make the patentable subject matter rather than naturally occurring substances.

### **ACMG Recently Weighed In Strongly and Effectively on DTC Genetic Testing**

With this past week’s announcement of another set of over-the-counter DNA tests being offered, this time at a national drugstore chain, ACMG reinforced its recommendations about Direct-to-Consumer Genetic Testing through a FOX TV television interview, featuring Medical Director Barry Thompson, MD and Judith Benkendorf, MS CGC, as well as written statements. Many ACMG members were called upon to comment on their concerns about this test by national and local media. In keeping with ACMG’s mission to advocate for the safe and effective use of genetic tests, our efforts contributed to the FDA’s issuing an immediate cautionary letter to the test manufacturer and the drug store chain severing ties with the testing company until regulatory issues are resolved. ACMG’s 2008 Statement on Direct-to-Consumer Genetic Testing, with its five critical points to consider, can be found on the ACMG website under Policy Statements.

### **ACMG Partners in Third Annual Global Rare Disease Day**

ACMG served as a partner for 2010 Global Rare Disease Day. The first Global Rare Disease Day was observed in Europe in 2008 and was organized by EURORDIS, the European Rare Disease Organization, and the national alliances of rare disease patient organizations in each country. The objective of this day is to recognize that rare diseases are a public health issue, affecting millions of people around the world. The goal of Global Rare Disease Day is to increase awareness of rare diseases, the special challenges encountered by those affected, and the need for research to develop safe, effective treatments or cures.

### **2009 ACMG Salary Survey Report Now Available**

The long-awaited 2009 ACMG Salary Survey Report is now available on the Members’ Only section of the ACMG website. ACMG’s biannual Salary Survey is a highly valued Member Benefit. Results of the 2007 survey were used by members to negotiate raises in their current positions, negotiate compensation and benefits packages when changing jobs, and in recruiting new medical geneticists to organizations and institutions. This new Report, which is almost twice as long as the 2007 Report, contains the findings of the Salary Survey conducted in October 2009 of American Board of Medical Genetics (ABMG)/Molecular Genetic Pathology (MGP) certified MD and PhD ACMG members who practice in the United States. In 2009, the survey tool and data analyses were expanded to include many new sections, such as

information regarding gender; benefits; the extent to which salary increases and decreases were awarded during both the current and prior fiscal year; MD/DO salaries based on relative value units (RVUs) generated as a medical geneticist; as well as an analysis of both MD/DO and PhD salaries as a function of time spent performing various responsibilities including laboratory direction, direct patient care, administration, consulting and research.

At a time when the medical genetics profession is working to increase the pipeline and assure access to services to all who need them, the survey results highlight the need to improve recognition and compensation to those clinicians and laboratory directors whose duties are primarily patient care. Non-members may contact the ACMG Office at [acmg@acmg.net](mailto:acmg@acmg.net) for information on how to purchase a copy of the Survey Report.

### **ACMG Continues Multifaceted Approach to Improve Reimbursement for Genetic Services: Data for Work-Unit Study Still Needed**

Reimbursement for genetic services remains nuanced and complex. ACMG's 2009 Salary Survey data underscore our need to attend to this issue. Yet, the ability of medical geneticists to provide quality genetic services is dependent on adequate reimbursement. ACMG continues to be engaged in several activities related to improved CPT coding for genetic laboratory tests. The time-work study is still collecting data from across the United States to use 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.

Clinical genetics programs that would like to contribute data are urged to do so; ACMG's goal is to be able to develop a data set that represents the breadth of services ACMG members provide as well as geographic practice differences. Participants are being asked to document the components of five patient care encounters, as well as the time spent providing genetic services, on forms provided by ACMG. Necessary forms and instructions can be obtained by contacting [acmg@acmg.net](mailto:acmg@acmg.net).

### **ACMG Supports the Recommendations to Include SCID in NBS Panel**

In late January, the Secretary's Advisory Committee for Heritable Disorders in Newborns and Children (SACHDNC) unanimously agreed to recommend that Severe Combined Immunodeficiency (SCID) be added to the universal newborn screening programs conducted by state public health agencies, a decision wholeheartedly supported by ACMG. SCID represents the first disorder to be added to the uniform newborn screening panel, developed by ACMG in 2005. It is also one of the best new examples of a condition for which early identification and treatment can totally transform the outcome of those who are affected since these infants can be treated and their lives both greatly improved and prolonged.

The SACHDNC recommendation highlighted three areas that need to be addressed to initiate screening: 1) educational materials for the providers, the public, and those who screen positively; 2) quality assurance materials for the screening laboratories; and 3) diagnosis and follow-up protocols and a network of providers who can deliver these services. ACMG will play a major role in each of these three areas, with the infrastructure and resources for pilot programs and both short- and long-term follow-up data collection being coordinated through ACMG's contract with the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) of the National Institutes of Health (NIH) to serve as the National Newborn Screening Translational Research Network (NBSTRN) Coordinating Center, now in its second year.

### **NBSTRN-CC Workgroup Activities Intensify**

Year two of the NBSTRN-CC contract between ACMG and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development has been marked by tireless workgroup activities. All workgroups are well on the way to fulfilling their goals, with frequent face-to-face meetings and conference calls. These include a Bioethics Workgroup, IT Workgroup, Clinical Centers Workgroup (which recently met with the Long-term Follow-up Workgroup of the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives), Laboratory Workgroup, Disease-Specific Subgroup, Lysosomal Storage Disease (LSD) Workgroup, Policy Workgroup, and the Standing Committee. A fulltime Director of Information Management Services will be joining the NBSTRN team on June 1.

### **Updated ACMG/NCC Newborn Screening ACT(ion) Sheets on ACMG Website with New Format and URL**

The NBS ACT(ion) Sheets and accompanying algorithms are valuable resources to primary care providers and medical geneticists needing critical clinical decision support at the point-of-care upon receipt of a "screen positive" result in one of their patients. ACT Sheets for all 29 disorders in the NBS panel have been updated and new ACT Sheets are available for all hemoglobin disorders, adult PKU, fragile X syndrome, cystic fibrosis and family history of colon

cancer. The ACT Sheets now have a new format and appearance, reducing them to two pages that are capable of being reproduced on a single, double-sided page. Additionally, they now are located on a new webpage; all who have linked directly to the ACT Sheets on the ACMG website should go to [www.acmg.net](http://www.acmg.net) and update their links.

#### **NCC Emergency Preparedness Workgroup Meeting Held in April**

The Emergency Preparedness Workgroup of the National Coordinating Center (NCC) for the Regional Genetic and Newborn Screening Collaboratives (RCs) met on April 6. RC representatives, members of the Genetic Services Branch, Maternal Child Health Bureau, HRSA, and the CDC's National Center for Birth Defects and Developmental Disabilities, responsible for framing the agency's draft Contingency Plan (CONPlan), joined emergency preparedness experts at the table. The task-oriented conversation included a review of the RC site visits and an overview of RC emergency preparedness activities either planned or in place; these collectively address State Public Health and Newborn Screening (NBS) Laboratory needs, clinical and information systems issues (including EHR models), and family preparedness mechanisms. General gaps in RC NBS emergency coverage are being assessed for follow up during the funding cycle commencing June 1, 2010. The role of medical genetics professionals in DNA identification following disasters was brought into the conversation, and background information necessary to develop and recruit a cadre of qualified volunteers will be integrated into the Emergency Preparedness section of the NCC website ([www.nccrcg.org](http://www.nccrcg.org)). Next steps include working to fill noted gaps in current activities, better integrate telemedicine technologies in NBS emergency preparedness efforts, and working in the future to implement the CDC's CONPlan.

#### **Genetics in Medicine Special Newborn Screening Issue**

In March 2010 *Genetics in Medicine* published a special issue on newborn screening that features a number of Reports either made to or promulgated by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (HHS). These include commentaries on evidence development process for newborn screening; perspectives on adding tests to the universal newborn screening platform; and an evaluation process for assessing nominated conditions. Newborn screening leaders from the ACMG membership contributed to this issue, and much of it is underpinned by earlier HRSA-commissioned work carried out by ACMG that led to the recommended universal panel.

#### **Genetics in Medicine Special Electronic Supplement on Gene Patents**

In April *Genetics in Medicine* issued a special electronic supplement, "Patently Complicated: Case Studies on the Impact of Patenting and Licensing on Clinical Access to Genetic Testing in the United States," which has received much acclaim by the media. It can be accessed at <http://journals.lww.com/geneticsinmedicine/toc/2010/04001>.

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](http://www.acmg.net). ACMG has also moved into the world of social media with a presence on Facebook, LinkedIn, YouTube, and Twitter. ACMG uses 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 its members and stakeholders.

*Submitted by R. Rodney Howell, MD, FACMG*

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