

## 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 nearly 1,600 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 genetics services and to promote the integration of genetics into all of medicine; and 4) to serve as advocates for providers of medical genetics services and their patients. This report summarizes key activities of the ACMG between September 2011 and April 2012.*

### **ACMG is now the American College of Medical Genetics and Genomics**

During its March 2012 Annual Meeting in Charlotte, NC, the American College of Medical Genetics officially became the American College of Medical Genetics and Genomics. In addition, a stylized new logo was unveiled that uses a double helix in forward motion, set inside a sphere to represent the global impact of genetic and genomic medicine. The ACMG's Board of Directors voted on this name change several months earlier to reflect the increasingly central role of medical genomics and its importance alongside genetics in fulfilling the mission of the College. Although medical geneticists' use of genomic tools is not new, the vastly increased power of the genomic approach has made it more and more vital to the practice of medical genetics. And, it was felt that adding the term "Genomics" to ACMG's name at this time recognizes the current importance of genomics as well as its future roles in both the clinical and laboratory practices of the medical geneticist. This name change will also broadcast to current and future trainees the extraordinary opportunities that lie before us. The College will continue to use the "ACMG" acronym, and members' FACMG appellation, will remain the same.

### **ACMG Sets New Records in 2012**

- The 2012 ACMG Annual Clinical Genetics Meeting, held March 27-31 in Charlotte, NC, broke all attendance records with nearly 1800 professional attendees, and an overall attendance of 2244, reflecting a 20% increase in registration over 2011.
- ACMG membership is at an all-time high, with approximately 1600 members and increasing numbers of students and medical genetics trainees joining the College.
- Eleven US Medical Schools now have official ACMG Student Interest Groups.

### **ACMG Releases Policy Statement on Genomic Sequencing**

In late March, ACMG released an important new Policy Statement on genomic sequencing, titled *Points to Consider in the Clinical Application of Genomic Sequencing*. The [Policy Statement](#) represents the first professional guideline for appropriate integration of genomic sequencing for the detection of germ-line mutations into clinical practice, and it outlines recommended general principles and points to consider when applying genome-scale DNA sequencing to the care of patients, whether associated with a phenotype or secondary to the phenotype that drove the need for testing. The Statement will help clinicians and laboratories understand:

- Diagnostic and screening applications for which genomic sequencing will likely be the most helpful;
- What patients need to know about genomic sequencing before it is performed; and
- How to use the information from genomic sequencing to improve patient care and medical decision-making.

One of the main conclusions of the Policy Statement is that genomic sequencing data that is not related to the clinical indications for testing must be integrated in the context of the medical and family history of the individual patient to guide medical decision-making. This should be done by a health provider who is familiar with the enormously complex and voluminous data that is obtained from genomic sequencing. By clearly articulating these recommendations, ACMG hopes that its Statement will help set a framework for clinical genomic sequencing that insures maximum value and quality for patients and the public. ACMG also expects that this document will require revision as this rapidly changing field evolves.

This Policy Statement represents the first in a series of initiatives planned by ACMG to prepare the health workforce for the integration of genomic sequencing of germ-line mutations into clinical medicine. A consensus statement on the reporting of incidental findings in whole genome/exome clinical testing, and several other clinical and laboratory practice guidelines are currently being developed.

### **ACMG-Led Inter-Organization Task Force on Medical Genetics Training Tackles Education and Workforce**

Under the auspices of the ACMG and the ACMG Foundation, a Task Force has been convened to: 1) assess current ACMG activities with regard to medical genetics and genomics training and the activities of other groups or organizations (e.g., American Board of Medical Genetics [ABMG], Association of Professors of Human and Medical Genetics/Organization of Program Directors [APHMG/OPD], American Society of Human Genetics [ASHG], and the Residency Review Committee [RRC]); 2) identify major gaps in availability of educational resources for medical geneticists and non-genetics trained providers; 3) make recommendations to the ACMG Board of Directors about activities in which ACMG should engage; and 4) work with the ACMG Foundation on fundraising needs to accomplish its goals. The eleven-member Task Force is chaired by Bruce Korf, MD, PhD, and has representatives from ASHG, ABMG, APHMG/OPD, the RRC, NHGRI/NIH, ACMGF, as well as a trainee member and a genetics laboratory director.

To date, the Task Force has identified programmatic strategies that address: 1) improving medical school visibility of medical genetics and student engagement; 2) enhancing medical genetics residency training to meet contemporary needs; and 3) developing the training needs for the next generation of medical genetics and genomics service providers involved in the use and interpretation of whole genome data. Initiatives in each of these areas in which ACMG is taking the lead include:

#### *Improving Medical School Visibility of Medical Genetics and Student Engagement*

- Summer Genetics Scholars Program (see below)
- ACMG Medical Student Interest Groups
- Increased student involvement in ACMG Annual Meeting
- Medical Genetics GME marketing brochure
- Career recruitment video

#### *Enhancing Medical Genetics Residency Training to Meet Contemporary Needs*

- Development of core educational materials
- Infusing medical genetics and genomics teaching into other residency programs, including information on genomic sequencing
- Development of educational resources for medical geneticists in training and in practice on rapidly evolving and new areas of practice

ACMG is collaborating with the RRC, ABMG and APHMG on several other residency-based initiatives. Finally, in the near future ACMG's Foundation will be launching a fundraising campaign to sponsor a 2-3 day, multifaceted CME-based Academy on Genomic Sequencing that will target medical genetics practitioners.

### **ACMG Grants and Contracts**

#### ***Newborn Screening Translational Research Network (NBSTRN) Coordinating Center***

- Part of a program authorized and appropriated under the Newborn Screening Saves Lives Act; operates under a contract from NICHD/NIH.
- Includes a Standing Committee and working groups in areas of Bioethics and Legal Issues, Newborn Screening Laboratories and Programs, Clinical Centers, and IT and Informatics.
- Provides a virtual repository for dried blood spots held by State Newborn Screening Programs and the only large biospecimen repository of the general population in the United States.
- Provides the tools for longitudinal point-of-care data capture related to diagnosis, evaluation and management with a centralized data warehouse.

Currently involves grantees studying more than 50 conditions that are identified by newborn screening programs or other conditions that are candidates for newborn screening.

#### ***National Coordinating Center for Regional Genetics and Newborn Screening Service Collaboratives (NCC)***

- Part of a program funded by the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA).
- Facilitates interactions among the seven regional collaboratives, public health and primary care providers, and specialists (including medical geneticists).
- Develops clinical decision support tools for primary care providers preparing to meet with a patient/family that has screened positively in newborn screening.
- Coordinates regional activities in telemedicine, patient transition from pediatric to adult care, roles of primary care medical homes in collaborative management of patients, relationships with other HRSA and federal resources.

### **ACMG Foundation's Summer Genetics Scholars Program (SGSP) Embarks on Second Year**

In the coming weeks, the ACMG and the ACMG Foundation will embark on the second round of their highly successful and sought-after Summer Genetics Scholars Program (SGSP). This program is designed to identify and encourage qualified medical students to consider careers in medical genetics by providing hands' on experience in the many exciting opportunities, from clinical and laboratory medicine and research to service delivery across the lifespan, available in medical genetics. This year, students at twenty-one institutions—eight of which are participating for the first time—will be placed with medical geneticist mentors under whose supervision they will work for a period of six to eight weeks. All principal mentors are ACMG Fellows, and students will receive a weekly stipend as part of the program. The intended outcome of the program is that by immersing students in hands' on medical genetics activities and fostering relationships with medical geneticist mentors earlier in their medical education, more young physicians will choose the field of medical genetics and genomics for residency training and beyond.

The following institutions were selected for participation in the 2012 SGSP:

Cedars-Sinai Medical Center, Los Angeles  
Children's Hospital Boston  
Children's National Medical Center, DC  
Emory University School of Medicine  
George Washington University School of  
Medicine and Health Services  
Greenwood Genetic Center  
Oregon Health and Science University  
Phoenix Children's Hospital  
Tufts University Medical Center  
Tulane University School of Medicine  
University of Alabama Birmingham  
School of Medicine

UCSF School of Medicine  
UCLA, David Geffen School of Medicine  
University of Maryland School of Medicine  
University of Miami, Miller School of Medicine  
University of Michigan Medical School  
UNC School of Medicine, Chapel Hill  
University of Oklahoma Health Sciences Center  
University of Pittsburgh School of Medicine  
University of Washington School of Medicine  
Wayne State University/Detroit Medical Center

### **ACMG Foundation Updates**

#### ***ACMG Foundation Changes its Name to the "ACMG Foundation for Genetic and Genomic Medicine" and Elects Dr. Bruce R. Korf as its New President***

During its March 2012 Board meeting the former American College of Medical Genetics Foundation changed its name to the **ACMG Foundation for Genetic and Genomic Medicine**. This name change realigns the Foundation with the new name of the College, and also recognizes its ongoing mission to raise funds that will help attain better health through genetics for all by addressing education, medical research, public health and medical practice needs in the genetics and genomics era.

**Bruce R. Korf, MD, PhD, FACMG**, was elected President of the ACMG Foundation for Genetic and Genomic Medicine. Dr. Korf is currently the Wayne H. and Sara Crews Finley Chair in Medical Genetics and Professor and Chair, Department of Genetics and Director, Heflin Center for Genomic Sciences at University of Alabama at Birmingham. He is Immediate Past President of the American College of Medical Genetics and Genomics, and has completed terms as president of the Association of Professors of Human and Medical Genetics, and as a member of the boards of directors of the American College of Medical Genetics and Genomics and the American Society of Human Genetics. He is presently a member of the Board of Scientific Counselors of the National Human Genome Research Institute.

Dr. Korf is known for his national leadership and landmark contributions to medical genetics education, and states that he looks forward to "working with the Foundation and with the College in educating both our members and other health providers on the application of genetics and genomics in this extraordinary and exciting era." Dr. Korf assumes the leadership of the ACMG Foundation at a time of almost unfathomable growth and change and of increasing need for education and training of healthcare providers and the public to deal with the remarkable opportunities that medical genetics and genomics has to improve health and prevent disease. There is unanimous agreement that he is the right person for this position at this time!

#### ***Dr. R. Rodney Howell Receives ACMG Foundation's 2012 Lifetime Achievement Award***

R. Rodney Howell, MD, FACMG, was awarded the ACMG Foundation's highest honor at our Annual Meeting in Charlotte, NC in March—the 2012 American College of Medical Genetics (ACMG) Foundation Lifetime Achievement Award. Dr. Howell is Professor of Pediatrics and Chairman Emeritus at the Miller School of Medicine at the University of Miami. He was is also Past President of both the College and its Foundation, as well as just completing a long tenure as ACMG's liaison to the National Advisory Council for Human Genome Research. Dr. Howell was honored for his life-long commitment and work in the field of pediatrics and genetic research and for his role and leadership in the development and advancement of newborn screening.

Duane Alexander, MD, the former Director of the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, where Dr. Howell served as the Senior Advisor to the Director from 2004 to 2011, a time when the Institute's research was focused on newborn screening, stated, "Dr. Howell really is a champion of Newborn Screening. He has been the leader in that area of pediatrics ever since the 1960s. He was an outstanding pediatrician and advocate for children as well."

Pursuing a career path as both a physician and a geneticist, Dr. Howell also became a teacher and mentor to many young medical students and a skilled administrator who could meld the views of many into an accepted consensus. He has been a leader of many causes, most notably as the Chairman of the Board of Directors of the Muscular Dystrophy Association, and as Chair of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (reporting to the Secretary of HHS).

In 2007, Dr. Howell was recognized with the Lifetime Achievement Award from the Duke University Medical Alumni Association. He is the fourth recipient of The ACMG Foundation for Genetic and Genomic Medicine's Lifetime Achievement Award, and was selected by a committee of ACMG's past presidents.

Other awards presented by the ACMG Foundation for Genetic and Genomic Medicine include: *The Richard King Trainee Award*, presented to an ABMG trainee who has published the best research paper in ACMG's peer-reviewed journal, *Genetics in Medicine*, during the past year; *The Genzyme/ACMG Foundation for Genetic and Genomic Medicine Medical Genetics Training Award in Clinical Biochemical Genetics*, presented for the first time to **two awardees**, to support a year of fellowship training for clinicians planning to devote their careers to the field of clinical biochemical genetics and especially the diagnosis, management and treatment of individuals with metabolic diseases; and the *ACMG Foundation for Genetic and Genomic Medicine/Signature Genomic Laboratories, PerkinElmer Inc., Travel Award*, presented to a selected student, trainee or junior faculty ACMG member whose abstract submission was chosen as a platform presentation during the Annual Clinical Genetics Meeting. For more information about the ACMG Foundation's Awards Programs, visit its website at [www.acmgfoundation.org](http://www.acmgfoundation.org).

### **Genetics in Medicine Updates**

A seamless transition occurred on January 1, 2012, when the prestigious Nature Publishing Group took over publication of ACMG's monthly journal, *Genetics in Medicine*. Listed below are the ACMG clinical and laboratory practice guidelines published in *Genetics in Medicine* between September 2011 and April 2012:

Pyeritz RE for the Professional Practice and Guidelines Committee of the American College of Medical Genetics. **Evaluation of the adolescent or adult with some features of Marfan Syndrome.** *Genet Med* 14(1):171-177 (January 2012)

**\*CORRIGENDUM to Laboratory Guideline for Turner Syndrome** (Wolff DJ, Van Dyke DL, Powell CM; a Working Group of the ACMG Laboratory Quality Assurance Committee. *Genet Med* 2010; 12(1):52-55). *Genet Med* 14(2):281 (February 2012)

\*Note that this correction corresponds to a change to **Section E5.1.2.2** of the *Standards and Guidelines for Clinical Genetics Laboratories*, referring to the detection of sex chromosome mosaicism, which has been changed to read, "Cases being studied for possible sex chromosome abnormalities, in which mosaicism is common, should include the standard 20-cell assessment. If mosaicism is confirmed, the analysis is complete. A minimum of 10 additional metaphase cells should be evaluated when one cell with a sex chromosome loss, gain or rearrangement is observed within the first 20 cells analyzed." This change is based on new evidence in the scientific literature and will be reflected in the CAP Proficiency Testing Checklist, as well.

Additionally, in April 2012 *Genetics in Medicine* published a special issue solely devoted to "Managing Incidental Findings and Research Results in Genomic Research Involving Biobanks and Archived Data Sets."

### **2013 ACMG Annual Meeting**

The 2013 ACMG Annual Clinical Genetics Meeting will be held March 19-23, 2013 in Phoenix, Arizona. Further details, including abstract submission information, will be available in the early fall at [www.acmgmeeting.net](http://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](http://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.*