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

May 4, 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 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 February and April 2011.

Annual Meeting Updates

The ACMG Annual Clinical Genetics Meeting, held in Vancouver, BC in March, broke all previous records. Professional attendance was up by 8%, as was overall attendance, including exhibitors and guests. Attendees came from 47 states, Puerto Rico and Guam, and 16% of attendees came from 27 countries outside the United States. Abstract submissions, poster presentations and exhibitor space also showed significant increases. The March of Dimes portion of the program featured an update on Hereditary Disorders of Connective Tissue, and preceding the conference there were two very popular short courses, "Preparing for an Expanding Range of Adult Genetic Consultations," and "New Insights into the Evaluation of Individuals with Intellectual Disabilities." Finally, the National Coordinating Center for the Genetic and Newborn Screening Service Collaboratives, (a MCHB/HRSA, Genetic Services Branch-funded cooperative agreement, housed at ACMG) co-sponsored a Community Conversation, "Screening for Carnitine-Palmitoyl Transferase, Type 1A (CPT1A) in the First Nations Populations of Alaska and British Columbia," in partnership with 'Kloshe Tillicum' British Columbia and Yukon Territories Network Environment for Aboriginal Health Research.

ACMG Annual Clinical Genetics Meeting Listed by The Trade Show Executives Among "The Fastest 50"

The unprecedented growth experienced by the ACMG's Annual Meeting (a 19% increase in booth sales in 2010) led to its inclusion on the list of the Trade Show Fastest 50, by the industry's executive magazine. ACMG's selection is not only a great honor, but made ACMG eligible for one of the Grand Prize Awards in 13 categories, or to be among 7 organizations spotlighted for growth that exceeded 2010 industry benchmarks in terms of increases in exhibit space sold, number of exhibits or attendees. Selections were made based on a rate of growth for meetings held in 2010 that exceeded at least 14.7 percent, and the awards were presented in March. ACMG's overall ranking was 39.

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, and will include the March of Dimes Clinical Genetics Conference as well as short courses. Further details, including abstract submission information, will be available in the early fall at www.acmgmeeting.net.

New Officers and Directors Begin Terms

Four new Directors, elected by the ACMG membership, began their terms at the close of the

Annual Meeting. **Gail E. Herman, MD, PhD** is the new President-Elect. Dr. Herman is a Principal Investigator, Center for Molecular and Human Genetics, The Research Institute at Nationwide Children's Hospital and Professor, Department of Pediatrics, The Ohio State University, Columbus, Ohio. Three additional new Directors elected to the Board include: **Mira Irons, MD**, Children's Hospital of Boston as a Clinical Genetics Director; **Christa Lese Martin, PhD**, of Emory University School of Medicine, Atlanta, GA as a Cytogenetics Director; and **Soma Das, PhD**, University of Chicago as a Molecular Genetics Director. All newly elected Directors will serve 6-year terms from 2011 – 2017. Directors completing their terms with the ACMG Board in 2011 include Past President **Joe Leigh Simpson, MD**; Secretary and Molecular Genetics Director **Elaine B. Spector, PhD**; Treasurer and Cytogenetics Director **Daynna J. Wolff, PhD**; and Clinical Genetics Director **Gerald Feldman, MD PhD.**

In addition, **Bruce Korf, MD, PhD**, Wayne H. and Sara Crews Finley Professor of Medical Genetics and Chairman, Department of Genetics and Director, Heflin Center for Human Genetics at the University of Alabama at Birmingham completed his 2-year term as President and passed the gavel to physician-scientist. **Wayne W. Grody, MD, PhD**. Dr. Grody is a Professor in the Departments of Pathology & Laboratory Medicine, Pediatrics, and Human Genetics at the UCLA School of Medicine, where he directs the Diagnostic Molecular Pathology Laboratory within the UCLA Medical Center, one of the first such facilities in the country to offer DNA-based tests for diagnosis of a wide variety of genetic, infectious, and neoplastic diseases, as well as bone marrow engraftment, patient specimen identification and paternity testing by DNA finger-printing. Dr. Grody is also an attending physician in the Department of Pediatrics, specializing in the care of patients with or at risk for genetic disorders. In addition, to being an active basic molecular genetics researcher, Dr. Grody has been one of the primary developers of quality assurance and ethical guidelines for DNA-based genetic testing for a number of governmental and professional agencies including the FDA, AMA, CAP, ACMG, ASHG, AMP, CDC, and the NIH-DOE Human Genome Project (ELSI program). He served as a member of the NIH-DOE Task Force on Genetic Testing, and was Working Group chair for development of ACMG's national guidelines for cystic fibrosis and factor V-Leiden mutation screening. Most recently, he was appointed chair of an Advisory Committee on Genomic Medicine for the entire VA healthcare system.

American College of Medical Genetics Foundation Awardees Honored

ACMG's Foundation continued with its tradition of presenting a large number of awards during the Annual Meeting. The late **Dr. Charles J. Epstein, MD**, Founding Fellow and Past President of ACMG was the 2011 recipient of the **American College of Medical Genetics Foundation Lifetime Achievement Award,** and only the third such honoree. Dr. Epstein, who lost his battle with pancreatic cancer on February 15th, was notified of this award before his death. Dr. Epstein was honored for his extensive research on Down syndrome and other genetic disorders and for his major role in gaining recognition for medical genetics by the American Board of Medical Specialists, which eventually led to the founding of the ACMG.

After graduating first in his class at Harvard Medical School in 1959, Dr. Epstein began a long career in medical genetics. He completed his internship and residency at Peter Bent Brigham Hospital in Boston and then held positions at the National Heart Institute and a fellowship at the University of Washington School of Medicine in 1963 with Arno Motulsky. In 1967, he began his long tenure at the Department of Pediatrics at University of California San Francisco. During that time he trained many medical geneticists, including many recognized leaders in the field today. He was named Professor in 1972 and was appointed head of human genetics in the Pediatrics Department, a position he held until 2005. Dr. Epstein was Board Certified in Clinical Genetics in 1982.

Among his most notable accomplishments was the work he did with the help of his wife, Dr. Lois Epstein, on the study of Down syndrome. Together with postdoctoral fellow David Cox, MD, PhD, they developed a mouse model for the disorder. The model led researchers to begin identifying specific genes and proteins that play a role in the development of the Down syndrome phenotype. Dr. Epstein also did extensive research on cellular aging and how various enzymes affected the aging process. Because of these interests Dr. Epstein was appointed chairman of the scientific advisory board and board of trusties of the Buck Institute for Research on Aging.

Dr. Epstein was the recipient of multiple awards, including the William Allan Memorial Award and the 2010 American Society of Human Genetics' Victor McKusick Leadership Award. In addition to serving as president of the ACMG, Dr. Epstein also served as president and board director of the American Society of Human Genetics, and president and board director of the American Board of Medical Genetics. He was also editor of many professional journals and a member of a several government panels. A prolific writer, Dr. Epstein produced more than 500 scholarly papers, authored and edited several books and served as a highly successful editor of *The American Journal of Human Genetics* for seven years. A beautiful tribute video was played at the award presentation, and may be viewed on The ACMG Channel on Youtube.

ACMG's Executive Director, **Michael S. Watson, PhD,** was surprised to find himself the inaugural recipient of **ACMG and ACMG Foundation Presidents' Award.** This award was established in 2011 to thank and honor the valuable contributions made by an ACMG member. Dr. Watson received his BS from American University and his MS in Medical Genetics and PhD in Physiology and Biophysics from the University of Alabama at Birmingham. After completing his postdoctoral fellowship in the Medical Genetics Training Program at Yale University School of Medicine, he was certified by the American Board of Medical Genetics in Clinical Cytogenetics and PhD Medical Genetics. He remains an Adjunct Professor of Pediatrics at Washington University in St. Louis, where he was Director of Clinical and Molecular Cytogenetics from 1986-2000. Dr. Watson became ACMG's first Executive Director 10 years ago, and during that time he has been responsible for the dramatic expansion of the ACMG staff and the remarkable involvement of the organization in so many areas of genetics in the practice of medicine.

Dr. Watson has a long-standing interest in the translation of genetic information into health care use. He was a founder of the American College of Medical Genetics, for which he has served as Vice President for Laboratory Affairs and has chaired its Test and Technology Transfer Committee, Economics Committee, Laboratory Practices Committee and Intellectual Property Committee. He co-chaired the NIH-ELSI/HHS Task Force on Genetics Testing from 1995-1997. Most recently, he was the project director for a MCHB/HRSA-funded project to bring uniformity to newborn screening in the United States. He is currently the Director of Administration MCHB/HRSA-funded National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development's Newborn Screening Translational Research Network (NBSTRN) Coordinating Center. In accepting his award, Dr. Watson thanked those who have allowed ACMG's success to be realized including the many Boards of Directors and Committees of ACMG, the marvelous staff of the College and ACMG Foundation, and the members of the ACMG community who work daily to deliver genetic services to patients.

The **ACMG 2011 Foundation's Richard King Trainee Award** for the best research publication in *Genetics in Medicine* in a given year was presented to **Rebekah Stackpole Zimerman, PhD** who is only the third winner of this $1500 prize, which was awarded for her manuscript titled "A Novel Custom Resequencing Array for Dilated Cardiomyopthy," which was published in the May 2010 issue of *Genetics in Medicine*. Dr. Zimmerman, a laboratory director at BioReference Laboratories, Inc., in Elmwood Park, New Jersey, was a Clinical Molecular Genetics Fellow at Harvard University Medical School at the time her manuscript was published. She and her colleagues addressed the challenge of designing an assay to improve the clinical sensitivity of genetic testing for Dilated Cardiomyopathy (DCM). Creating a test that increased the number of genes sequenced from 10 to 19 required using a sequencing technology other than the gold standard of Sanger sequencing. This manuscript presented the validation of the use of a resequencing array, the DCM CardioChip, to sequence 19 genes associated with DCM. The study showed that, in comparison to Sanger sequencing, the DCM CardioChip reduced test cost and turn around time by approximately 50%, while also predicting to increase the clinical sensitivity. This award was established by the ACMG Foundation to encourage ABMG trainees in 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.

The 2011-2012 **Genzyme/ACMG Foundation Clinical Genetics Fellowship in Biochemical Genetics** was awarded to **Anna-Kaisa Niemi, MD, PhD**, a resident at Stanford University Medical Center. This award encourages the recruitment and training of physicians in clinical biochemical genetics, with emphasis on the diagnosis, management and treatment of individuals with metabolic disorders. The award grants $75,000 to sponsor a trainee's first year of fellowship following residency.

Adam H. Buchanan, MS, MPH, a board-certified genetic counselor and research scientist at Duke University was honored as the 2011 recipient of the **PerkinElmer Signature Genomic Laboratories Travel Award**. Mr. Buchanan was selected for his platform presentation, "Telemedicine vs. in-person cancer genetic counseling in rural oncology clinics: a randomized controlled trial of cost and patient satisfaction," which reported that cancer genetic counseling provided via telemedicine was as well accepted as in-person counseling at less than half the cost. These findings are promising for improving access to cancer genetic counseling in underserved areas. 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.

New Alice and YT Chen Scholarships Allow Trainees to Attend Annual Meeting
Each year the demand for travel assistance and scholarships to attend the ACMG Annual Clinical Genetics Meeting continues to grow. For the first time in 2011, the ACMG Foundation offered scholarships to promising graduate and medical students and medical genetics trainees (residents and fellows in either an ACGME or ABMG accredited program) whose young careers and future goals would be enhanced by attending the Annual Meeting. These scholarships were made possible through generous donations from ACMG members and from Dr. and Mrs. YT Chen (in honor of Dr. R. Rodney Howell, President of the ACMG Foundation). Interest in the 2011 Alice and YT Chen Scholarships was extraordinary, with over 70 applications received. Ten scholarships were awarded to: James T. Bennett MD, PhD (University of Washington); Kerry K. Brown, PhD (Harvard Medical School); Aaron B. Chance (Johns Hopkins Bloomberg School of Public Health); Jill Farhner, MD, PhD (Johns Hopkins Hospital, McKusick-Nathans Institute of Genetic Medicine); Natalie Gallant, MD (UCLA); Christie Maier, MS (Northwestern University); Elaine M. Pereira, MD (Children's Hospital of Montefiore) Erica F. Sanford, (UCSF); Angela Sun, MD, (UCLA); and Xinjie Xu, PhD (University of Wisconsin).

ACMG Foundation Hosts First-Annual Day of Caring with Free Bicycles for Children with Genetic Diseases
The ACMG Foundation added a new event to the Annual Meeting with its first **Day of Caring** on Saturday, March 19th. This special event set a precedent for the Foundation's commitment to also serving in a charitable capacity, as the ACMG community worked with the Rare Disease Foundation in Vancouver Foundation to identify 20 children with genetic diseases who would benefit from a new bicycle and helmet. "Children with rare diseases are so often left to feel like they are 'different' or 'unique' and all they want to be is like every other child, to be able to do and experience the same things as their peers. Having a bicycle like the 'other kids' means a lot to these children and to their families. As one mom said to me, 'Anything that brings normality to my child's world makes a huge difference, not just for him but also for us as parents.' Programs like this one are as important for ill children as are the programs that support the research that will one day hopefully find cures," said Christele du Souich, M.Sc., CCGC, CGC, Genetic Counselor, Researcher, The Rare Disease Foundation. The ACMG Foundation's 2011 Day of Caring was supported by PerkinElmer, Agilent Technologies, members of the American College of Medical Genetics, and the ACMG Foundation.

ACMG congratulates all of its 2011 awardees. More information on the ACMG Foundation and applications for these and other awards can be found on ***www.ACMGFoundation.org***

Regional Collaboratives Bring Emergency Preparedness and Planning to Local Communities
Providing seamless follow-up for the four million infants who receive newborn screening (NBS) tests and services in the United States each year is one of public health's greatest successes, especially when one considers the many steps and providers involved in the process. However, any glitch in the continuity of services in this complex system—from a local laboratory power outage to a disaster of the magnitude of hurricane Katrina—may mean delayed treatment, and devastating consequences on the lives of babies with positive newborn screening results, particularly those with metabolic disorders. For this reason, the HRSA Genetics Collaboratives (RCs) and their National Coordinating Center (NCC) have been addressing NBS emergency preparedness in a variety of ways since 2006. Go to the NCC website at www.nccrcg.org to access the December 2010 issue of the *NCC Collaborator*, which brings together reports on the activities occurring across the seven RCs, which collectively include all of the elements of a national emergency preparedness plan for any medically fragile patient. Also in this issue is a must read article by William Perry, NCC Emergency Preparedness Consultant, "Emergency Preparedness in Ten Easy Steps." Together, these efforts are building national preparedness systems that will benefit genetic centers, primary care, state public health programs, and families so that unforeseen circumstances will not disrupt newborn screening and genetic services and follow-up care.

NBSTRN-CC Activities Continue to Break New Ground in Developing a National NBS Collaborative Research Infrastructure
In February we delineated many of the year three activities 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). Each of the committees and workgroups, comprised of numerous national experts, continues to meet regularly. A list of the workgroups follows, with several salient updates highlighted. In recent months several NBSTRN-CC staff members have presented their activities and accomplishments both nationally and internationally.

- **The NBSTRN Standing Committee**

- **The Laboratory Network Workgroup**
- **Clinical Centers Workgroup,** whose **Disease Specific Sub-Workgroups** are identifying the disease specific elements of the long-term follow-up data sets (LTFU). The Sub-Workgroups meet regularly via conference call and efforts are coordinated with the team at the National Library of Medicine (NLM) in preparation for the creation of standardized language and HL7 messaging of the LTFU data sets.
- **The Bioethics & Legal Issues Workgroup**
- **The Information Technology Workgroup**
- **LSD Pilot Project Workgroup**: The LSD workgroup has been developing the infrastructure to support the pilot testing of these new conditions in NBS and to reach consensus on the clinical parameters of diagnosis and follow-up of patients identified with lysosomal storage disorders by:
  - Developing the data systems and the data analysis from the newborn screening programs; and
  - Developing the protocols that define the datasets for the diagnosis and follow-up of these patients.

  By bringing together a group of clinical experts as well as individuals from the states that are or will be screening for LSDs, the NBSTRN assists in creating standardization and consensus at the outset of screening for these disorders. Ongoing discussions have centered around the development of a clinical centers network, updates on national efforts, entry of screening data into the R4S database, and the collection of long-term follow-up data. Posters and platform presentations on the LSD Pilot Project were presented at the SIMD and the WORLD Symposiums.
- **SCID Pilot Project Workgroup:** The SCID group has had monthly conference calls since June and met in September 2010 and March 2011, with representatives from states not yet screening for SCID joining the regular conference calls in increasing numbers. Their work has culminated in report that is being presented Secretary's Advisory Committee on Heritable Disorders in Newborns and Children on May 6th. Through this pilot (and during the past eight months), over 521,000 babies have been screened and seven cases of classic SCID and eight cases of SCID variant have been identified, diagnosed and treated. In addition, almost 98,000 babies have been screened in the ongoing Massachusetts and Wisconsin pilot and four classic SCID infants have been identified. The incidence of SCID and related T-cell lymphocyte deficiencies appears to be higher than previously thought, and new information about the molecular basis and sub-population variability of cases is emerging.

The NBSTRN-CC currently administrates two subcontracts, with a third program about to begin:

1. **Development of the Virtual Repository of Dried Blood Spots**
2. **Adaptation of the R4S Laboratory Performance Database**

3. **Long-Term Follow-Up Data Collection Initiative Planned**
   The Center for Biomedical Informatics (CBMi) at the Children's Hospital of Philadelphia (CHOP) and the NBSTRN are partnering to present a solution for collecting, managing, and delivering long-term care data for individuals identified with conditions through newborn screening. Effective capture of long-term clinical data for these individuals represents an exciting opportunity to establish a longitudinal data collective, which would produce a transformative resource for biomedical research. Delivery of a mechanism to collect, manage, and disseminate long-term clinical data in the context of newborn screening information and biomaterials is a key outcome for the remaining 3 years of the initial NBSTRN project period.

   This project aims to establish a network of centralized and institutionally-enabled infrastructure to support the capture and managed storage of longitudinal clinical data from individuals following newborn screening. Data governance, security, and workflow processes will be developed and implemented to ensure that accrued data is standardized, secure, and of the highest possible quality for conducting research. These data will be made available to the research community using procedures that comply with established legislative and data practice guidelines. We plan to construct the NBSTRN data network as a highly scalable and extensible project that is compatible with other emerging national networks and data grids. We also plan to develop and contribute tools and procedures to the open-source community without restrictions to the greatest extent achievable.

***Competencies for the Physician Medical Geneticist in the 21st Century* Slated for Summer Publication**

Since 2004, ACMG has been actively examining the needs of the medical genetics workforce in light of the very rapid scientific progress that is occurring. A 2005 report proposed expanding the scope of medical genetics training to include new areas of practice. This was followed by a set of recommendations in 2008, defining a scope of practice and agreed upon principles given the direction of genetic and genomic medicine. It was also suggested that medical genetics curricula for training and maintenance of certification be updated to align with new scientific

trends. A group of invited experts met at Stone Mountain Conference Center in Atlanta, GA in February 2010 to tackle this challenge. The result is a set of competencies and learning objectives that describe not only what the medical genetics physicians should *know*, but also what s/he should be able to *do*. The document contains overarching competencies as well as discipline–specific competencies covering seventeen areas of practice (biochemical-metabolic genetics, cancer genetics, connective tissue disorders, cardiovascular genetics, dermatologic genetics, dysmorphology, endocrine genetics, gastrointestinal genetics, hematology genetics, immunological disorders, nephrologic disorders, ophthalmologic genetics, pulmonary genetics, prenatal and reproductive genetics, psychiatric genetics, and skeletal genetics). The document is currently being finalized, with publication expected early this summer.

New ACMG Student Interest Groups to Aid in Recruiting Medical Genetics Trainees
The College has 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 student interest groups around the country. Beyond providing an institution's Student Interest Group with recognition by the College, establishing this connection will optimize two-way communication between the College and students enthusiastic about the field of medical genetics. This new program will also offer targeted resources, educational materials, and opportunities for networking. More information will be posted on the ACMG website in the coming months. Please help feel free to identify medical genetics student interest groups at your institution to us (or consider starting a group), and direct your students, or the faculty advisors of your existing Medical Genetics Interest Groups, to Denise Calvert (dcalvert@acmg.net), ACMG's Membership Services Coordinator, for more information and an application.

ACMG Launches New Employment Resource Center
This online resource was designed to help medical genetics professionals find the right position, and to assure that employers can find the right medical genetics professionals to fill their needs. ACMG's new on-line Employment Resource Center is open to anyone with an interest in the medical genetics profession and will generate more opportunities for both job seekers and employers. The new site is fully automated, easy to use, and makes automatic posting and renewal possible. Users can even sign up to receive automatic email notifications regarding positions available and qualified candidates that meet their criteria. For more information go to the Resources tab on the ACMG website at www.acmg.net.

Two Special Issues Published by *Genetics in Medicine*
The March issue of *Genetics in Medicine* celebrates ACMG at 20 years with a look back and an exploration of where genetics is taking medicine—from whole genome sequencing of patients to educating the next generation. The April issue is dedicated to the ethical, legal and social implications of genetics and genomics.

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 a "mobile view" feature.

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 are now available at http://www.acmg.net/AM/Template.cfm?Section=2011_Genetics_Review_Course&Template=/CM/HTMLDisplay.cfm&ContentID=6344.

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