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## Recent Activities of The American College of Medical Genetics

The American College of Medical Genetics (ACMG) is the professional home to more than 1500 board certified clinical and laboratory genetics professionals. ACMG's myriad activities advance the practice of medical genetics and strengthen its workforce through educational programs, the development of practice guidelines, advocating for genetic services in health care and in public health, and by promoting the development of methods to diagnose, treat and prevent genetic disease. This report summarizes key activities of the ACMG between May and September 2006.

The Fellows of the ACMG recently elected a new slate of Officers, who will begin their 6-year terms on April 1, 2007.

#### **President Elect**

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ACMG has begun the third year of its cooperative agreement with GSB/MCHB/HRSA, to serve as the National Coordinating Center (NCC) for the nation's seven Regional Genetic Service and Newborn Screening Collaborative Groups (RCs). This program continues to work towards improving the health of children and their families by promoting the translation of genetic medicine into public health and health care services. The RCs strengthen and support the genetics and newborn screening capacity of the States, and therefore the nation, using a regional approach to addressing maldistribution of genetics providers and bring services closer to local communities. The NCC uses ACMG's unique expertise to enhance the activities of the RCs by providing the infrastructure, coordination, technical assistance and resources necessary to address issues of universal importance. On-going initiatives focus on telegenetics capacity building; development of a network of genetics service providers that includes a framework for the designation of clinical genetics centers; an assessment of the business

case for medical genetics services and service providers; creation of disaster preparedness systems to support newborn screening and the care of metabolic patients; the development of additional management guidelines for genetic disorders, written for primary care providers; and establishing a national collaborative data collection mechanism for the disorders tested in newborn screening. The ACMG and the NCC are cosponsoring a meeting developed by NICHD/NIH on “Strengthening Newborn Screening in North Africa and the Middle East” on November 13-15, 2006 in Marrakech, Morocco.

National workgroups on Telegenetics and Newborn Screening Emergency Preparedness have been active since this past summer. The Telegenetics workgroup is preparing to conduct a national survey of existing telegenetics programs, which will allow us to build a directory of local experts and identify barriers to expanding these programs. Addressing issues such as reimbursement and interstate licensing of providers will be a priority in order to build national capacity. The Newborn Screening Emergency Preparedness workgroup will be conducting a needs assessment that will fuel the development of a robust and deliberately redundant national system involving federal, state and local experts in genetics, public health, telecommunications and emergency management. A meeting of the leaders of the Regional Collaborative Groups was convened on September 7<sup>th</sup> to focus on using the RCs to design and implement a cohesive collaborative system for collecting follow-up data on newborn screening patients that can be used for research purposes—and particularly for identifying participants for clinical trials for therapeutics for these rare conditions. This latter activity is a critical step in ensuring that the scientific breakthroughs from the human genome project can move into clinical care safely and effectively.

The NCC was recently awarded several supplemental grants from MCHB/HRSA to:

- 1) Develop point-of-care management guidelines for newborn screening and genetics. This work will expand the development of ACT sheets and similar materials for genetic conditions that are not part of newborn screening programs (such as cystic fibrosis and fragile X). The materials are intended to be disseminated by genetic testing laboratories and will accompany positive test results in order to enhance the capabilities of primary care providers to interact with patients and families. In addition, the ACMG ACT Sheet for identifying genetic etiologies in the work-up of newborns with hearing loss will be adapted into a brochure that is tailored to parents of infants who screen positive in state-based Early Hearing Detection and Intervention programs. Finally, we will develop model management guidelines for primary care providers to address issues in the transition of patients with genetic conditions, many of whom have been identified through newborn screening, from pediatric to adult care, using hemoglobinopathies as the pilot.
- 2) Enhance, improve, expand and increase access to counseling and health care services to newborns and children having or at risk for genetic disorders by focusing on genetics education at the medical school level and for practicing physicians. This will involve reviewing Steps 2 and 3 of the USMLE and doing a case study of genetics teaching in one US medical school in order to drive curricular updates that will promote the application of genetics knowledge. We will also be developing of point-of-care educational strategies and systems for practicing physicians, including working with manufacturers of electronic medical records systems to promote inclusion of genetics patient information and related prompts, ACT sheets and management guidelines for health professionals.
- 3) Consider how to integrate the Regional Collaboratives and the NCC into data collection efforts that include the development of registries of patients from newborn screening and genetics clinics. This would provide a resource to improve public health quality assurance of NBS programs and to allow for the identification of large numbers of patients for studies requiring those. Further, it will allow for more in depth clinical research of the nature of the diseases and the available management options within the RCs.

For further information about the National Coordinating Center, visit the new NCC website at [www.nccrcg.org](http://www.nccrcg.org).

To date, little to no attention has been directed towards quality improvement in clinical genetics. Few studies have been designed and conducted, and key medical outcomes have not been defined. Yet, this is both an area of national interest (as evidenced by two recent IOM reports) and evidence of quality performance in practice is a part of Expanded Maintenance of Certification for medical geneticists. To address this issue, the ACMG has established a Clinical Genetics Quality Special Interest Group, which will be chaired by Marc Williams, MD, FACMG. Members of this group will participate in a two-day training workshop at Intermountain Healthcare, Salt Lake City, UT in December 2006. The training will include didactic instruction as well as strategic planning activities to determine and prioritize key processes of clinical genetics, and discuss the design of projects that assess these key processes. Expected benefits will include robust, multi-center data collection, and the ability to translate these findings into quality improvement projects. The data will also inform and underpin activities to build the business case for genetic services, develop best practice models, and conduct cost and cost-benefit analysis. On-going member education about quality improvement will occur through workshops at the Annual Clinical Genetics conferences.

On Monday November 20<sup>th</sup> the ACMG, together with Montefiore Medical Center/Albert Einstein College of Medicine will hold an open forum on carrier screening for genetic conditions, in New York City. This meeting is the first in a series of activities to examine and address population-based carrier screening—ultimately leading to the development of clinical practice guidelines. Community leaders will be presenting the views of ethnically diverse populations, and Dr. Mary-Claire King will discuss the pitfalls of using race as a biological marker. It is the belief of the organizers of this conference that the preparation of panethnic screening recommendations, as published for cystic fibrosis carrier screening, would be a difficult and extensive undertaking. Hence, a multi-step approach is being employed here as we move towards developing broader guidelines for carrier screening in specific populations or the entire population. Written proceedings will be one end product of this meeting.

ACMG will hold its Annual Clinical Genetics Conference from March 21-25, 2007 at the Nashville Convention Center and Nashville Renaissance Hotel, Nashville, TN. The meeting will include the 38<sup>th</sup> March of Dimes Clinical Genetics Conference on the Genetics of Skeletal Dysplasias, and a joint symposium with the Society for Inherited Metabolic Disease on Expanded Newborn Screening. Details can be found on the ACMG website at [www.acmg.net](http://www.acmg.net).

The following new ACMG Guidelines have been published in *Genetics in Medicine*:

Pompe disease diagnosis and management guideline. (*Genet Med* 2006;8:267)

Newborn Screening: Toward a Uniform Screening Panel and System (*Genet Med* 2006;8:1S)

ACMG Practice Guideline: Genetic evaluation of suspected osteogenesis imperfecta (OI) (*Genet Med* 2006;8:383)

The September issue of our newsletter, *The Medical Geneticist* will be posted on our website at [www.acmg.net](http://www.acmg.net), later this month.

Further information about all ACMG activities, the 2007 Annual Clinical Genetics Conference, and a full listing of our clinical genetics laboratory and practice guidelines, can be found on our website at [www.acmg.net](http://www.acmg.net).

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