

Letter from the President

September 30, 2005

Dear Members:

This letter is to update the membership on the activities of the ACMG over the past six months. The 2005 Annual Clinical Genetics Meeting in Grapevine, Texas (Elaine Spector, Program Committee Chair) was extremely successful. Over 1000 professionals participated in educational sessions, committee activities, and industry sponsored forums to assimilate information that might improve outcome for patients, identify new research initiatives, and enhance individual practice.

The Program Committee for 2006, chaired by Rick Martin, has digested the comments from last years' attendees and integrated the proposals from the membership to organize what looks to be an outstanding educational event in San Diego. I hope you have marked March 23-26, 2006 on your calendars and plan to attend the meeting. Speaking as a San Diegan, the Manchester Grand Hyatt is an outstanding downtown property within easy walking distance of the renovated Gas Lamp District (with *many* dining options). In addition to a spectacular bay front location it affords easy access to the airport.

The ACMG has benefited from increased member and non-member attendance as well as increased participation by exhibitors. Thanks to a generous grant from the ACMG Foundation, the 2005 meeting turned a small profit. Our meeting planners surveyed the needs/preferences of attendees of the annual meeting for the past three years. The membership clearly prefers meeting content to location. The Board is sensitive to the cost issues and will strive to avoid terrariums in the future. Some of the most preferred cities, however, are among the most expensive venues in the country. After San Diego, comes a moderately priced center in Nashville, Tennessee.

The Board of Directors met in Miami, Florida in July to review and update the strategic plan for the College. One of the major goals of that plan was the recruitment of an Executive Director. Michael Watson has provided outstanding national (and international) leadership for the College. Through his efforts in securing a range of grants and contracts as well as his tight fiscal management, the ACMG is now in a position to build back the reserves that were expended in the implementation of that plan. I am pleased to announce that his contract will be renewed. Moreover, the Washington office is at full staffing with Melissa Forburger replacing Gerry Gurvitch as Director of Administration, Judith Benkendorf as Project Manager, Lori Oxendine as Executive Assistant, and Kristin Harwood as Office Manager. Do visit if you are in the DC area.

Issues from the strategic plan that remain ongoing concerns include:

1. identity of the medical geneticist;
2. marketing of medical genetics both in terms of recruitment to the profession and recognition among other medical practitioners of our unique skills and expertise;
3. training of future clinical and laboratory geneticists;
4. advocacy for patients and the profession at the national and state level; and
5. financing medical genetics.

The issue of "identity" is central to the discussion. Most of you are aware that the ACMG (Charlie Epstein, Mike Watson and I) participated in a conference at Cold Spring Harbor (October 2004) organized by Bruce Korf (representing ACGME/Genetics Residency Review Committee), Georgia Wiesner (representing the American Board of Medical Genetics), and Jerry Feldman (representing Association of Professors of Human and Medical Genetics) to discuss physician training in medical genetics. Also in attendance were leaders of ASHG, NSGC, the Canadian College of Medical Genetics, AAP, NHGRI, and SIMD. Many of you may have seen the document that resulted from this conference, which included the affirmation that Medical Genetics is a primary specialty (It currently is; but many practitioners neither think nor function that way.).

As result of extensive discussion stemming from this Banbury Conference, I have appointed a work group, chaired by Bruce Korf (University of Alabama, Birmingham) and Michael Murray (Children's Hospital, Boston) to flesh out the issue of the identity of the medical geneticist in the early 21st century, not only with respect to training but with respect to the expected skill sets that such individuals will need to possess, the settings in which they will practice, and evolving overlap with other specialties. The committee will include individuals representing the perspectives of OB geneticists, dysmorphologists, metabolic geneticists, and adult geneticists. It has become clear from this endeavor that a similar effort will be necessary on the laboratory side so that pathologists do not swallow the field as the current ABMG certified practitioners retire.

With respect to recruitment to training, the ACMG now has a handsome brochure advertising Medical Genetics Residencies. We are most grateful to Genzyme Therapeutics for the grant in-kind to the Foundation to produce the

brochures. Copies have been distributed at the AMA Medical Specialty Showcase, at the ASHG Meeting and to residency advisors in US medical schools. The ACMG plans to host a session for training program directors in San Diego in an attempt to develop a forum in which common problems can be addressed.

The ACMG has received a great deal of national and some international publicity surrounding the HRSA/MCHB supported project to bring uniformity to newborn screening in the US. The panel of suggested disorders emanating from this project was discussed by the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, which has forwarded recommendation to the Secretary for Health and Human Services. Some of you are involved in the National Coordinating Center: Genetics and Newborn Screening Regional Collaboratives, which has begun the task of developing resources and approaches that will facilitate integration of genetics services into the main stream of health care delivery particularly, but not exclusively, related to newborn screening. In addition to management guidelines, part of this effort will be compiling lists of existing clinical and laboratory resources, defining what constitutes a "genetics center". A parallel but separate effort of the College is the National Collaborative Study for Rare Genetic Diseases, which is flushing out current obstacles (such as IRB issues) to this effort and exploring models that may facilitate such endeavors.

Leadership from the ACMG has discussed with counterparts on the ABMG ways in which the two organizations may collaborate on Maintenance of Certification, which is rapidly evolving. A joint work group, co-chaired by Mira Irons (the ACMG CMSS representative) and Karla Matteson (executive director, ABMG), will be addressing this.

Other committee activities of the College are as follows:

- 1) Standards and Guidelines (chair, Sue Richards) is developing standards for array-based comparative genomic hybridization;
- 2) A work group headed by David Flannery under Economics (chair, Marc Williams) will attempt to define RVU's for clinical genetics;
- 3) The Education Committee (chairs, Muffin Alford and Melissa Fries) will develop modules to be used as MOC activities;
- 4) The Social, Ethical and Legal Issues Committee (chair, Lynn Fleisher) continues to address issues related to a code of ethics and expert testimony. With respect to the latter, our AMA representative, Ray Lewandowski, supported a proposal on expert witness testimony brought before the AMA House of Delegates by the AAP.
- 5) Professional Practice and Guidelines Committee (co-chaired by Louanne Hudgins and Helga Toriello) is moving forward documents on preconception and prenatal testing of husbands or partners for carrier status, genetic evaluation of suspected osteogenesis imperfecta, and cytogenetics evaluation of the individual with developmental delay or mental retardation. A guideline on Charcot-Marie-Tooth disease and related hereditary polyneuropathies is in preparation.
- 6) The newly constituted Committee on Therapeutics (chaired by Bill Wilcox) is developing an agenda to address translation of new treatments into clinical practice.

The College has begun a search for a new Editor of *Genetics in Medicine*. Dick King who has shepherded the journal through its inception to a respected monthly publication will retire in 2006. A search committee is actively pursuing a successor. If any of you are interested or know of someone the committee should approach, please forward a CV or contact information to Mike Watson or me. The journal has become an important face for the College. The Editor sits on the Board as a non-voting member. This recruitment will be critical for the ongoing success of our organization.

Lastly, the Foundation, under the able leadership of Rodney Howell, has embarked on an aggressive fundraising campaign. The College is only as strong and as capable as our Foundation can make it. We hope that the membership will consider the ACMGF important among the organizations they choose to support financially. The \$100,000 TM Bioscience Award will again be presented at the annual meeting. Program announcements for the 2006 award are posted to the ACMG web site (acmg.net).

As always, the College's Board of Directors looks forward to hearing from members about issues that are of concern. Only in this way will we be able to represent your interests in an appropriate and responsible way.

With best wishes,



Marilyn C. Jones, MD
President

ACMGF/Genzyme Announce Biochemical Fellowships

The ACMG Foundation is pleased to announce the "Genzyme Clinical Genetics Fellowship in Biochemical Genetics" Award Program. The purposes of the program is to advance education, research and standards of practice in medical genetics; develop and expand clinical and laboratory expertise in medical genetics in the United States; initiate and develop a broad-based infrastructure for industry funding of high quality projects in the fields of medical genetics. Below is information on the Fellowships.

Background Newborn screening programs are rapidly expanding in the United States. However, there is a deficit of physicians specifically trained in the diagnosis, management and treatment of individuals with the metabolic diseases identified in the programs. It is expected that these programs will continue to expand as more conditions such as the lysosomal storage diseases are added into newborn screening. Clinical Geneticists, of whom there is a subset who focus on biochemical genetic disorders, are often the physicians involved in the coordination of care of these patients. The ability of medical genetics to adapt to the evolution of therapeutic approaches to genetic diseases will be critical to its sustained success. Genzyme/ACMGF seeks to support a robust national fellowship program to promote and encourage the recruitment and training of clinicians for this purpose. To provide an in-depth clinical experience at a premier medical center with expertise and significant clinical volume in the area of biochemical genetics, including lysosomal storage diseases, as well as in therapeutics and clinical trials involving patients with these and other metabolic diseases and, thereby, to increase the number of medical geneticists with interest, knowledge, and expertise in this field.

Award Program The Award Program is designed to support the sustainable development of clinically interested and qualified Clinical Geneticists with expertise in biochemical genetics. Each trainee-applicant and his/her sponsoring institution will submit a joint application to the ACMGF committee for the sponsorship of the trainee through the Award Program to focus the trainee-applicant's first year of fellowship following his/her residency in a genetics fellowship on biochemical genetic diseases.

Activities Fellowship activities of the Awardee will focus on significant clinical care of patients with biochemical genetic disorders. The Awardee will design a research project to be conducted over the course of the Fellowship Year, which will be clinical in nature, and may relate to any area of current or future therapy for metabolic diseases. This Research Project may include work with registries, clinical trials, investigator initiated work, or other areas of clinical investigation. The ACMGF will provide a platform for the presentation of the Awardee's work at the appropriate time in the Fellowship Year.

Award Process Trainee-applicants and institution-applicants will submit their joint applications at the beginning of the trainee-applicant's second year of residency, with selection occurring in October, notification of the Awardee in December, and announcement of the Awardee at the ACMG Annual meeting which will be held in March. The Fellowship Year will commence on July 1st.

For application and criteria information, please visit www.acmg.net

ACMGF/Tm Bioscience Award Request for Applications

Tm Bioscience and the American College of Medical Genetics Foundation (ACMGF) have entered into an agreement to make an award(s) to an ACMG member(s) to undertake activities aimed at promoting safe and effective genetic testing and services. The award will support one applicant for \$100,000 for one year beginning on or before September 1, 2006 and is aimed at promoting the safe and effective integration of genetic testing and services into health care such as the development of research guidelines.

Purpose of the Request for Applications

Medical geneticists should play a crucial role in translating genetic science into medical genetic services and in defining and promoting the integration of genetics into health care. This program will provide awards to medical genetics professionals to assist them in undertaking activities aimed at promoting safe and effective genetic testing and services.

Project Topics

Project topics may include, but shall not be limited to:

- a. Medical genetics research
- b. Research on healthcare economics related to medical genetics
- c. Medical education and training in medical genetics
- d. Development of medical genetics standards and guidelines

Award Deadline/Notification

For application information, visit www.acmg.net to download an application. Applications are due to ACMGF headquarters by December 1, 2005. The award recipient(s) will be announced during the 2006 Annual Clinical Genetics Meeting, March 23-26, 2006 in San Diego, California.