

Association of Professors of Human and Medical Genetics
10th Annual Workshop
Thursday, January 22, 2004
Business Meeting

Following a wonderful opening reception, the meeting was called to order at 6:30 PM by the president of the APHMG, Dr. Bruce Korf.

Secretary-Treasurer's Report (Jerry Feldman)

There is currently \$79,363.35 in the APHMG treasury (\$27,065.62 in a CD note at Paramount Bank and \$52,287.73 in a no-service fee checking account at Comerica Bank, both in Detroit, MI). At the last APHMG meeting there was \$80,990.57 in the 2 accounts. Due to the fact that this year's meeting is being held in January, dues notices have not been sent out, so there is no 2004 dues revenue at this time. In addition, most of the upfront hotel charges have already been paid.

The meeting this year is estimated to cost approximately \$24,000 with revenues of \$22,500 for a net deficit of ~\$1,500. There were a total of 51 registrants for the meeting.

There were 83 paid members in the APHMG in 2003, which is a decrease of 9 compared to 2002.

American Board of Medical Genetics (Mimi Blitzer):

Dr. Blitzer discussed a change in the ABMG policy – if an examinee takes a second specialty exam in consecutive cycles, then the examinee does not have to retake the general exam.

Dr. Blitzer also gave some information on current trainees – there are 235 trainees (7 programs did not respond to the survey). They are beginning to gather information about the number of examinees for the 2005 examination. They expect ~177 individuals, some of whom will sit for more than 1 specialty.

There are currently 9 combined Pediatric-Medical Genetics residencies, 1 Internal Medicine-Medical Genetics residency and 10 Molecular Genetic Pathology fellowship programs. Dr. Rimoin raised a concern that some Pediatric-Medical Genetics residents do not like the rotating back and forth between pediatrics and genetics and that some genetics programs are not used to training residents so early in their career (i.e. after only 1 year of internship).

Maintenance of Certification requirements will be in place by 2009.

AAMC/CAS (Bruce Korf)

Dr. Robert Desnick is our representative to the Council of Academic Specialties (CAS); he will be attending this meeting later and will present his report at that time. Dr. Korf is our representative on the LCME (Liason Council on Medical Education), which accredits medical schools.

Residency Review Committee for Medical Genetics

Dr. Feldman, Chair of the RRC Medical Genetics Committee, discussed that Molecular Genetic Pathology fellowship program now has 10 accredited programs and they primarily have been developed by Molecular Pathologists, as would be expected. There has been good input from Medical Genetics at most of the programs. A workshop on Saturday will discuss the possibilities for an additional training mechanism in Clinical Biochemical Genetics.

American College of Medical Genetics (Mike Watson, Executive Director)

Dr. Watson discussed a newborn screening contract that ACMG is involved in. He also noted that Mira Irons (APHMG representative from Harvard) is our representative to CMSS, which is focusing on Maintenance of Certification issues. The ACMG is also involved in discussions to determine the needs of Medical Geneticists in 5-10 years – scope of practice, involvement in pharmacogenomics, treatment of genetic diseases, etc. The upcoming ACMG annual meeting has sold out its block of hotel rooms and the ACMG Foundation has developed relationships with commercial companies to support scholarship activities.

American Society of Human Genetics (Joann Boughman, Executive Director)

Dr. Boughman discussed that ASHG is focusing on public/privacy issues in genetics, especially related to current or pending Federal legislation to prevent genetic discrimination. Senate Bill 1053 passed with a vote of 95-0, whereas House Bill 1910 was referred to committee and is likely not going to come up for a vote. She also discussed that April 30th will be designated as DNA Day in 2004

The Nominating Committee (Peter O'Connell, Lori Potocki, Feige Kaplan and Bruce Korf)

The Committee previously had a conference call and selected the following slate of candidates:

Councilor (to replace Feige Kaplan): Diane Cox. A motion was made and seconded to appoint Dr. Cox as a Councilor (2004-2007). The vote was unanimous in favor.

President-elect: Gerald Feldman. A motion was made and seconded to appoint Dr. Feldman as President-elect (2006-2008). The vote was unanimous in favor. Because Dr. Feldman is currently Secretary-Treasurer, the Bylaws were reviewed by Lynn Fleisher, council for APHMG. There is no regulation preventing an individual from holding more than 1 office, but the Council and Membership felt it would be appropriate for a new Secretary-Treasurer to be elected within the next year so that a smooth financial transition can be made. The Council will solicit interested candidates.

A discussion of future APHMG meeting venues took place. There was a consensus that meeting in a locale such as Puerto Rico with a schedule that allows for time to network was beneficial. Though the number of registrants was somewhat down this year, the Membership felt it important to maintain its own identity and to not meet in conjunction with the ACMG. Dr. Keats will investigate West Coast meeting sites, such as Palm Springs, CA, Tucson, AZ, or Baja for 2005 and will consider returning to Puerto Rico in 2006. There was discussion that additional effort to invite the genetic counselor program directors will be made for future meetings. All felt it would be beneficial to have additional dialogue and input from those involved in genetics education at all levels.

The meeting was adjourned at 9PM.

Respectfully Submitted,

Gerald L. Feldman, MD, PhD, FACMG
Secretary-Treasurer, APHMG

Association of Professors of Human and Medical Genetics

10th Annual Workshop

January 22-24, 2004

Plenary Workshop Summary

Friday, January 23, 2004

Plenary Session

The Role of the GCRC in Genetic Research and Education

Feige Kaplan, Bronya Keats, Tony Wynshaw-Boris, moderators

The session on Medical Education focused on the role of General Clinical Research Centers (GCRCs) in clinical training in genetics. Anthony Hayward (Director of the Division of Clinical Research at the National Center for Research Resources [NCRR] at the NIH) began the session by discussing the organization of the NCRR, which has supported high-throughput genotyping through 12 GCRCs that have these resources available at core labs.

The enhancement of clinical trials is confronted by two other obstacles in academic medical centers. One is the growing bureaucracy of Institutional Review Boards (IRBs). An informed consent form has grown from one page of standard English to nine pages, much in legal jargon. The possibility of a uniform consent form will be engaged in a meeting organized by Dr. Hayward at NIH on March 10, 2004. Indeed, NIH has crafted uniform material transfer agreements (MTAs) for its cooperative groups; this form of consolidation is necessary if one is to follow Dr. Zerhouni's NIH "road map" toward an increase in collaborative, translational research. The impact of the diversity of IRBs on clinical research in this country and between countries is the subject of an upcoming article by Greg Koski in the *New England Journal of Medicine*.

Dr. Giovanna Spinella, Director of the Extramural Research Program at the Office of Rare Diseases (ORD) discussed the development of a cooperative rare diseases clinical research network utilizing institutional GCRCs in rare diseases research and education. The website is <http://rarediseasesinfo.nih.gov>. The goal is to promote scientific workshops and intramural research. The K08 award permits an MD to acquire laboratory competence in 3 to 5 years, while the K23 awards train for clinical competence in Medical and Clinical Genetics, Molecular Biology, and Biochemistry over the same timespan. Despite these programs, the numbers of new individuals being trained in Clinical Genetics has fallen to 69 in 2002, compared to 283 in 1982 and the average age of a Clinical Geneticist is now 51 years.¹

The second half of this workshop was devoted to a discussion of the development of clinical research in academic medical centers. Two medical schools summarized their recent experiences. Cincinnati (Greg Grabowski, MD) has combined support from the State of Ohio together with General Clinical Research Center (GCRC) funds from NCRR to develop a Center for Computational Medicine. This Center is able to perform between 20 and 40 x 10⁶ SNP-

genotyping assays per year on DNA samples from patients at Cincinnati. Cincinnati's GCRC grant is controlled by the Dean of the Medical School.

Baylor (Lori Potocki, MD) also depends strongly on its GCRC grant to support clinical research, but supplements this in several necessary ways:

- A cohort of Clinical Genetics trainees, 3 to 4 per year. These trainees are trained in clinical trial design, scientific writing, and biostatistics/epidemiology.
- Professional colleagues. This includes clinical research coordinators, collaborators from basic sciences, and senior mentors.
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Financial aid for trainees. The K23 awards from NIH are important, but private funding is also needed.

Feige Kaplan discussed the preliminary results of the Genetics Education Survey – 73 surveys have been completed. The surveys were mailed to the Deans at each medical school to collect information on medical genetics curriculums, such as the number of contact hours, what year genetics is taught and the importance of medical genetics in medical school.

Plenary Session

Mentoring and the Role of the Physician-Scientist

Reed Pyeritz and Robb Moses, moderators

This session was devoted to mechanisms for mentoring scientists in training and junior faculty. Interesting, the ABMG does not list mentoring in its program requirements, while the ACGME speaks to “supervision” to ensure that residents have the opportunities to develop a personal program. Dr. Pyeritz discussed the U Penn academic progression, including the importance of mentoring because of rigid requirements for appointment as an assistant professor.

Dr. Moses discussed the mentoring program at Oregon Health Sciences University and Dr. Korf discussed challenges and opportunities for mentoring the physician scientist. Both discussed methods of evaluating and gauging physician scientists in different stages of development, standards for selecting and evaluating junior faculty, and mechanisms of mentoring junior faculty in the academic environment and preparing the faculty for promotion and tenure judgments.

Saturday, January 24th, 2004

Plenary Session

The Question of Expanded Pathways for Biochemical Training

Jerry Feldman, moderator

There is a significant shortage of Clinical Biochemical Geneticists in the United States to manage patients with inborn errors of metabolism. Many university medical centers lack boarded biochemical geneticists, and newly trained biochemical geneticists are highly sought. Furthermore, there are very few young physicians currently enrolled in ABMG-accredited Clinical Biochemical Genetics Fellowship programs. This session served as a forum for discussing alternative training options for physicians that are currently being formulated by the ABMG, ACMG, APHMG and the Medical Genetics Residency Review Committee (RRC). Participants included Dr. John Belmont from Baylor College of Medicine, Dr. Mimi Blitzer from University of Maryland (who presented the proposal from the ABMG) and Mike Watson, the

Executive Director of the ACMG. Dr. Belmont, who is a member of the Medical Genetics RRC, and Dr. Feldman, who is the chair of that committee, discussed the involvement of the RRC in developing a new training track for physicians to be certified/boarded in managing patients with inborn errors of metabolism. Dr. Blitzer discussed a proposal from the ABMG, in which either the ABMG and/or the RRC would develop a subspecialty certification for a physician. There was a consensus that such a track was needed, though it was still unclear as to whether that should be developed through the RRC or the ABMG. **BRUCE – CAN YOU SUMMARIZE THE DISCUSSION AT THE FOLLOWUP BREAKFAST MEETING, WHICH I WAS UNABLE TO ATTEND.**

Plenary Session

Genetics and Clinical Research: Relationships with Industry

Bruce Korf, moderator

Access to new, powerful approaches in genetics and genomics presents major opportunities for the clinical investigator. These approaches enable studies of the natural history, etiology, and management of both rare and common disorders. In some instances, large cohorts can be assembled to identify genetic factors that contribute to disease risk. The genetics community is well positioned to participate in this effort, but this will require training and mentorship in the approach to clinical investigation. There are also chances to establish collaborations with industry, where there are major interests in the study of both rare and common disorders and where there are substantial resources to apply new technologies to large populations. This session explored the opportunities and challenges for genetics and genomics in clinical investigation.

Dr. David Rimoin (Cedars Sinai Medical Center) presented a talk titled “Jobs, Problems and Potential Therapy” in which he discussed the crisis developing with the lack of clinical researchers. He has previously presented this talk at the Institute of Medicine.

The keynote speaker was Dr. William Crowley, the Director of Clinical Research and the Director of the Harvard Reproductive Endocrine Science Center at Partners Health Care and Massachusetts General Hospital. Dr. Crowley discussed the ways in which medical centers can partner with private funding to develop the framework for clinical trials in the post-genomic era. He noted that there were 4 components necessary for success – core laboratories, a platform to support clinical research (i.e. adequate databases) proteomics facility (clinical and industrial) and repositories (tissue and DNA).

Respectfully submitted,

Gerald L. Feldman, M.D., Ph.D., FACMG
Secretary-Treasurer, APHMG