Conference Report

Summary of the Association of Professors of Human and Medical Genetics Fourth Annual Workshop

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INTRODUCTION

The Association of Professors of Human and Medical Genetics (Association of Professors) is the organization of Chairs, Directors, and Chiefs of Human or Medical Genetics units in universities throughout North America. The Association of Professors meets annually to discuss issues and concerns related to academic genetics in medical and graduate schools. Officers for 1998 included President Robert Desnick (Mount Sinai), President-Elect Louis Elsas II (Emory University), Past President Jan Friedman (University of British Columbia), and Secretary-Treasurer Emmanuel Shapira (Tulane University). Councilors were Uta Francke (Stanford University, 1995–1998), Huntington Willard (Case Western Reserve, 1996-1999), and Miriam Blitzer (University of Maryland, 1997-2000). Emmanuel Shapira unexpectedly and sadly passed away in May 1998, and Mary Kay Pelias has served as Interim Secretary-Treasurer. Bruce Korf (Harvard) was elected as a councilor for the 1998–2001 term, replacing Dr. Francke.

The Fourth Annual Workshop was held on 13–15 March 1998 at the Asilomar Conference Center, Pacific Grove, California. More than 100 participants representing more than 75 universities throughout North America attended. The workshop included four plenary sessions entitled (1) Information and Updates, (2) Training Program Directors Meeting, (3) Genetics in Primary Care Specialties, and (4) Guidelines for 3rd and 4th Year Medical School Students. Highlights of each session are summarized below.

SESSION I: INFORMATION AND UPDATES (R.J. Desnick and Louis Elsas, Co-Chairs)

This session was devoted to updates and the latest information relevant to academic human/medical genetics from related organizations. The following individuals presented:

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- American College of Medical Genetics (ACMG): Reed Pyeritz, M.D., Ph.D., President and Michael Watson, Ph.D., Vice President for Laboratory Ser-
- American Board of Medical Genetics (ABMG): Daniel Van Dyke, Ph.D., President
- American Academy of Pediatrics (AAP): Beth A. Pletcher, M.D., Liaison, Section on Genetics and Birth Defects
- American College of Obstetricians and Gynecologists (ACOG): Sherman Elias, M.D., Genetics Committee
- Council of Regional Genetics Networks (CORN): Louis J. Elsas, M.D., President
- American Society of Human Genetics (ASHG): Miriam Blitzer, Ph.D., Chair, Information and Education Committee
- APHMG Question Database: Margaret McGovern (Mount Sinai) reported on the status of the APHMG Question Database. The Database containing over 1,200 questions categorized by topic was distributed in PC or Mac formats to requesting universities. Dr. McGovern discussed the need for additional high-quality questions in certain areas. Overall, the database has been well received, and the membership congratulated Dr. McGovern on having produced and maintained an excellent resource for exam questions.
- USMLE Medical Genetics: On behalf of the APHMG-USMLE Liaison Committee, Dr. Blitzer was pleased to report that the National Board of Medical Examiners (NBME) accepted our recommendation to establish a Step 1 Exam "Discipline Score" for Genetics that would be reported to the Deans of all Medical Schools. In fact, this was accomplished for the 1997–1998 Step 1 Examination and will be repeated annually.

This year the committee did not make a USMLE site visit, because the NBME requested that we review these exams every other year. Also, based on our recommendation, a geneticist (Dr. Barry Wolf, Virginia Medical College) was recruited as a question writer for the USMLE Step 1 exam, and he will be responsible for reviewing and identifying the genetics questions for future Discipline Scores.

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Dr. Blitzer then introduced Dr. David Swanson, Director of the USMLE Step 1 Exam, who provided an overview of the content and recent trends in the USMLE Steps 1, 2, and 3 examinations. He also discussed the "Discipline Score" in Genetics, and the potential inclusion of more questions relevant to "genetics and primary care." In the discussion that followed his presentation, the membership indicated its appreciation of the USMLE's responsiveness to the importance of genetics as a discipline and its role in basic biomedical science and the practice of primary care medicine.

SESSION II: TRAINING PROGRAM DIRECTORS MEETING (John Carey and Robert Saul, Co-Chairs)

This session was the first annual Training Program Directors Meeting at the APHMG. At the 1997 APHMG meeting, it was decided to devote a session at our annual meeting for the Training Directors to discuss common opportunities, issues, policies, and problems. This year Dr. Robert Saul (Greenwood Genetic Center) organized a session designed to establish training program guidelines for residents and fellows in each of the ABMG/RRC training programs. Prior to the meeting, committees were organized for each of the training areas, and the committee chairs presented their initial drafts of the training guidelines for each discipline for discussion and debate. It was the goal of these guidelines to establish a knowledge/competency set for residents and fellows in each training area so that they would know what they were expected to achieve in their training program.

This session reviewed preliminary drafts of guidelines for trainees in medical genetics training programs (clinical genetics residency, Ph.D. medical genetics, clinical biochemical genetics, clinical cytogenetics, and clinical molecular genetics). These drafts represented the initial efforts of an ad hoc group of medical genetics program directors. The importance of developing these guidelines (identifying issues in training in competencies above and beyond the mandates of the RRC in clinical genetics and the ABMG) was recognized and will be developed further over the next year.

Dr. Saul reviewed the process leading to this focused workshop on training program guidelines. In response to changes occurring in genetics training programs accredited by the ABMG (Ph.D.) and the Accreditation Council of Graduate Medical Education (M.D.), medical genetics training program directors have explored the need to establish a working group to address the needs of this group that are separate and distinct from any other existing group. The ABMG sponsors a program directors meeting each year, but the focus of that meeting is to discuss items that the Board wants to bring to the attention of the Program Directors. The APHMG has encouraged the Program Directors to come together to address issues of common interest. The initial project of the working group will be to develop guidelines for trainees in medical genetics training pro-

Initial drafts were presented in the following areas

(the chairs of these committees indicated in parenthesis): Clinical Genetics (Robert Saul), Ph.D. Medical Genetics (Joann Boughman), General Laboratory (Mike Watson), Clinical Biochemical Genetics (Steve Goodman), Clinical Molecular Genetics (Charles Schwartz, presented by Robert Saul), and Clinical Cytogenetics (Mary C. Phelan, presented by Dan Van Dyke).

The lively discussion that followed the presentation of the drafts yielded several points that will guide this ongoing drafting process through the next year.

- These guidelines should not be limited to RRC or ABMG mandated guidelines. At the same time, it would be helpful to have RRC and ABMG representatives collaborate with this process.
- 2. We are attempting to define "uniform skills" we expect trainees to obtain during training and to possess after the completion of training. We must recognize that Board certification does not ensure clinical competency. Clinical competency comes from training, not by examination.
- We are looking for methods of teaching clinical genetics skills and the methods of evaluating these skills.
- 4. We need to have a list of expectations for residents/ fellows <u>and</u> program directors to ensure a clear understanding before, during, and after training.
- 5. We have the unique opportunity to establish a significant degree of uniformity in medical genetics training without sacrificing the unique aspects of training that individual training sites might possess.
- 6. We are looking to preserve the uniqueness of medical genetics professionals, a broad spectrum of professionals from physician-scientists to private-practice clinical geneticists.
- 7. Medical geneticists are leaders in multidisciplinary medical care yet often are not viewed in those terms. This process of drafting, adopting, and maintaining guidelines for medical genetics training programs should have the effect of bringing medical genetics into the mainstream of medical education, medical training, and clinical practice.

The drafts will, over the next year, be revised and presented in preliminary form at the APHMG-Program Directors meeting in October 1998 (in conjunction with the 1998 ASHG meeting) and formally adopted at the APHMG-Program Directors meeting in April 1999.

SESSION III: GENETICS IN PRIMARY CARE SPECIALTIES (Miriam Blitzer and R.J. Desnick, Co-Chairs)

The objective of this session was to review the current status of clinical genetics education in primary care residencies and to develop liaisons to establish or increase the residency guidelines for genetics in each. Our goal is to assist each primary care specialty in providing an optimal, up-to-date educational and training experience in clinical genetics.

As a keynote speaker, Dr. Susan J. Hayflick (Univer-

sity of Oregon), spoke on the findings of her survey of genetics in primary care specialties and residencies. The clear message was that the level of knowledge in genetic medicine was low, particularly for physicians in family medicine and internal medicine. After her excellent review of the current state of clinical genetics education in residency programs, the following speakers discussed the specific situations in their respective residency programs.

- Family Practice: Dr. Perry Pugno, Director of the Family Practice Program at Mercy Healthcare/ Methodist Hospital in Sacramento, CA, and a member of the American Academy of Family Physicians (AAFP) Commission on Education, discussed the training of family practice physicians. He stated that the AAFP appreciated the importance of genetics in family practice and the fact that currently there are limited curricula in clinical genetics, but no core educational guidelines in genetics. On behalf of the AAFP, he requested that the Professors assist the AAFP Commission on Education in developing core educational guidelines in genetics. Subsequently, an AAFP committee has been formed, and an APHMG representative will serve on the steering committee overseeing the development of the core educational guidelines in genetics.
- Pediatrics: Dr. Carolyn Schanen (UCLA) pointed out that there are specific genetic guidelines for residency training in pediatrics and that genetic components are intertwined throughout the professional competencies and RRC guidelines. A large number of questions on the Pediatrics Board examination already encompass clinical genetic topics.
- Obstetrics and Gynecology: Dr. Sherman Elias (Baylor) was invited to review the residency guidelines in genetics for residents in obstetrics and gynecology. He emphasized that the Council on Resident Education in Obstetrics and Gynecology had established a core curriculum and educational objectives in genetics. He also pointed out that genetics was prominent in written and oral Board examinations as well as in the recertification and subspecialty Board exams. In addition, he noted that ACOG had developed a variety of educational materials in the various areas of clinical genetics. Most geneticists were impressed with the extensive residency guidelines and educational resources, but wondered to what extent these guidelines were actually pursued because of the limited time and residency contact with local genetic units.
- Internal Medicine: Dr. Reed Pyeritz (Allegheny-Singer Research Institute) discussed the current lack of genetics education in internal medicine residency training and the fact that internists in general did not recognize genetics as a specialty. Internal Medicine Program Directors rarely include genetics in residency training. He suggested several steps that would be required to attempt to include genetics in medicine. Clearly, the need exists to increase genetics education in this field, but the competition for time and the complex process involved in establishing guidelines create a challenging opportunity.

A lively panel discussion ensued with questions from the participants to the expert speakers. In general, the discussion focused on how genetics could be integrated into the primary care specialty residency requirements and CME programs. A variety of suggestions were made, including having the APHMG establish relationships with Program Director organizations in the other primary specialties.

SESSION IV: GUIDELINES FOR 3RD AND 4TH YEAR MEDICAL SCHOOL STUDENTS (Jan Friedman and Bruce Korf, Co-Chairs)

At the 1997 meeting of the APHMG a committee was assembled to formulate a set of objectives in medical genetics for third and fourth year medical students. The committee is co-chaired by Drs. Friedman and Korf. Other members are Dr. Louis J. Elsas, Miriam Blitzer, Rachel Fisher, Joann Bodurtha, Margretta Seashore, G.S. Pai, Eva Sujansky, and Linda Adkison.

Dr. Bruce Korf presented background information about the context in which third and fourth year medical school teaching occurs. With the shift of medical care increasingly from an inpatient to an outpatient focus, there is a necessary change in the organization of clinical teaching that affects all specialty areas. Genetics faces a particular challenge in that it is not only a recognized specialty itself, but also contributes to all other areas of medicine. Incorporating these objectives into medical school teaching will require active participation by medical geneticists in the teaching program and a high level of visibility of geneticists as providers of care at their institutions. There will also be a need for development of new approaches to teaching, including problem-based exercises and possibly computer- or web-based exercises, to supplement the material that can be presented at the bedside. Finally, methods of evaluating the effectiveness of teaching in genetics, both through Step 2 of the USMLE and evaluative tools used locally, will be needed.

Dr. Jan Friedman then presented the draft of the objectives. He noted that the committee made a set of assumptions, including the following: 1) Medical students already possess an understanding of the basic principles of human genetics; 2) Learning in medical genetics will take place over the career of each physician; 3) Many of the skills and attitudes that are integral to the practice of genetics are also important in all areas of medicine, and teaching of genetics can help to reinforce the development of these skills and attitudes; 4) A qualified medical geneticist would be given the authority and responsibility to implement this curriculum at each medical school. The objectives have been published recently [Friedman et al., Genetics in Medicine 1:54–55, 1998]

Discussion of the objectives was followed by a series of brief talks outlining the approach to teaching medical genetics in the clinical years at different medical schools.

Dr. Linda Adkison (Mercer University) reported on small group teaching in genetics in the context of the Ob/Gyn clerkship. She also described efforts to identify the genetics issues that arise in other clerkships and to

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work with clerkship directors to enhance the teaching of these issues within those clerkships.

Dr. Debra Day-Salvatore (UMDNJ) described efforts to incorporate genetics teaching into a third year Ob/Gyn clerkship. Each student attends one genetic counseling session, and there is an option to spend half a day in a pediatric genetics clinic. There are also monthly pediatric genetics conferences, weekly genetics/maternal fetal medicine conferences, and semimonthly neonatology/genetics conferences, all of which are attended by medical students.

Dr. Charleen Moore (University of Texas, San Antonio) described a medical genetics course that is presented to fourth year students as part of a one-month course. There are four lectures on genetics as well as small group discussions. The latter are led by clinical geneticists, postdoctoral fellows, genetic counselors, and researchers.

Dr. John Mulvihill (University of Oklahoma) noted that genetics is represented in a course on the molecular basis of disease. Students may also elect to participate in a genetics elective.

Dr. Greta Seashore (Yale) noted that curriculum committees for years I/II and III/IV have recognized the possibility of organizing integrated clerkships for specialties that cut across classical specialty lines. Genetics is ideally suited to be taught in such an integrated clerkship, which is being explored.

Dr. Louis Elsas (Emory) described the structure of genetics teaching at Emory, noting the important contribution made by nonphysicians, including genetic counselors and researchers. This not only is a necessity given the large staff resources required by small-group teaching, but also introduces the students to various types of professionals involved in genetics clinical practice and research.