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Association of Professors of Human or Medical Genetics: Summary of First Annual Workshop

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INTRODUCTION

The development of an organization for the heads of medical school genetics departments (and equivalent leaders from universities that do not have formal human or medical genetics departments) began in 1991 with a meeting arranged by Tom Caskey, who was then immediate past president of the American Society of Human Genetics (ASHG). The meeting was held during the 1991 International Congress of Human Genetics in Washington, D.C. About 50 invited participants attended. As a result of this meeting, a steering committee was struck that included Bob Sparkes (chair), Tom Caskey, Maimon Cohen, Tom Gelehter, and John Hamerton. Subsequent meetings of interested individuals were held at the ASHG annual meetings in 1992, 1993, and 1994. At the 1993 meeting, it was decided to proceed with the formal establishment of an Association of Professors of Human or Medical Genetics (APHMG). A constitution was adopted for the APHMG in 1994, and its first officers and council were elected.

The first workshop of the Association of Professors of Human or Medical Genetics was held in San Diego, California, April 22–24, 1995. There were 74 participants representing 61 medical genetics academic programs throughout the United States and Canada.

APHMG members are institutions, not individuals. Each university (or, in the case of universities with more than one campus, each campus) with a human or medical genetics program may become a member, but there may be only one member per university campus. Each member institution has only one official representative.

Current officers of the APHMG are Jan Friedman, M.D., Ph.D. (University of British Columbia), president; Robert Desnick, Ph.D., M.D. (Mount Sinai School of Medicine), president-elect; and Emmanuel Shapira, M.D., Ph.D. (Tulane University School of Medicine), secretary-treasurer. The president and president-elect each serve 1-year terms. The secretary-treasurer serves a 3-year term.

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The APHMG Council is composed of the officers and three councillors, each of whom serves a 3-year term. The current councillors are Jewell Ward, M.D., Ph.D. (University of Tennessee Medical School-Memphis), John Carey, M.D., M.P.H. (University of Utah School of Medicine), and Skip Elsas, M.D. (Emory University School of Medicine).

The Association meets twice yearly. A business meeting is held during the annual meeting of the American Society of Human Genetics. Issues facing academic medical genetics as well as the development and improvement of medical genetics teaching are discussed at the annual 3-day workshop. Highlights of the first annual workshop are summarized below.

SESSION 1: ROLE OF THE ASSOCIATION OF PROFESSORS OF HUMAN OR MEDICAL GENETICS (JAN FRIEDMAN, CHAIR)

Jan Friedman reviewed the purpose of the Association, which is the promotion and continuous development of human and medical genetics academic programs in North American medical and graduate schools. The emphasis on academic programs and the focus on medical and graduate schools distinguishes us from other genetics organizations.

Maimon Cohen discussed how the APHMG fits in with other genetics organizations and suggested that the APHMG expand its activities in three areas: education, administration, and lobbying.

With respect to education, the APHMG must work to improve undergraduate medical student education in genetics through implementation of the ASHG Model Curriculum, development of effective case-based problems in genetics, and establishment of clinical rotations in genetics.

The APHMG could work with the American College of Medical Genetics to improve postgraduate medical education in genetics. Establishment of continuing medical education courses in genetics for generalist physicians is another possible activity for the APHMG.

The APHMG can also provide administrative support to its members and help individual institutions adapt national standards to local educational programs. The collective expertise of APHMG members might be particularly important as a resource to uni-

versities with weak medical genetics programs or none at all.

The APHMG may have an important role in lobbying. APHMG representatives are distributed throughout the United States and Canada and are highly knowledgeable about both genetics and their local political situations. Face-to-face meetings with state and federal legislators and their staffs are an especially effective means of influencing them. APHMG members are often in a position to encourage the development of lay constituencies that can provide strong political support for genetics. Undertaking activities in support of maintaining federal discretionary funding for NIH and the training of health professionals would be an important first step in this regard.

Hunt Willard discussed his experience as an invited speaker at a recent meeting of the Association of Chairmen of Departments of Physiology. This organization, like other organizations of basic science department heads, does not concern itself with clinical matters, while organizations of clinical department heads deal much more with activities at hospitals and with residency training. Our group is concerned about both of these areas.

The physiology professors meet among themselves some years, and jointly with another basic science chairmen's organization other years. They spend much of their time on issues related to research funding, an area that we cannot afford to overlook. The physiology heads have developed important contacts by inviting to their annual workshops chairs of departments in other basic science disciplines, congressional leaders concerned with research funding, key congressional staff, and NIH directors.

One of the most important functions of the physiology heads' workshop is networking among participants. Maintaining the size of the meeting in the range of 50–100 participants is important to facilitate networking and to encourage effective deliberations.

A discussion among all participants raised many opportunities and difficulties for academic genetics programs. Genetics is currently in a paradoxical situation: most nongeneticists realize the importance of genetics in the curriculum, but few seem to understand the breadth of what geneticists do. Thus, many nongeneticists think that the essential part of genetics is rather simple and can easily be taught by nongeneticists.

Recommendations:

1) That the APHMG work to develop a United States Medical Licensing Examination (USMLE) Test Materials Development Committee in Genetics and to include information on student performance on genetics questions in USMLE reports to deans.

2) That the APHMG join the Council of Academic Societies as soon as possible, and work to include assessment of genetics teaching in the medical school accreditation process.

3) That the APHMG develop and maintain a data base of cases for problem-based teaching in genetics.

4) That the APHMG develop a data base of examination questions for medical genetics courses.

SESSION 2: DO MEDICAL SCHOOLS NEED DEPARTMENTS OF MEDICAL GENETICS? (EMMANUEL SHAPIRA AND ROBERT DESNICK, CHAIRS)

Bob Desnick presented the results of a survey that had been sent to 132 medical genetics units in the United States and Canada regarding their organization within the university. Responses were obtained from 91 units. Nineteen percent of the units were organized as full departments. Most (70%) were divisions or sections within departments, and the other 11% were centers, programs, or institutes.

There was a great deal of variability in the number of faculty in medical genetics units, but in general full departments were largest, averaging 15 faculty members each. Programs and centers averaged 8–9 members, and divisions and sections about 5 members each.

All units reported that they offered a Medical Genetics "Principles" course. Most offered the course in the first or second year of medical school. The course averaged about 32 hr.

About 90% of the units offered American Board of Medical Genetics (ABMG)-accredited training in clinical genetics, clinical electives for medical students, and rotations for residents in other specialties; 34 units offered Ph.D. programs in genetics; 47 units offered training for Ph.D. postdoctoral fellows.

The breadth of clinical services provided varied among units, but usually included clinical genetics services (86%), prenatal diagnosis (70%), metabolic disease services (60%), cancer genetics (58%), and newborn screening (55%). Most units included a general cytogenetics laboratory (74%), a general DNA diagnosis laboratory (63%), a cancer cytogenetics laboratory (59%), and a biochemical genetics laboratory (59%). The overall set of services provided was different in almost every unit.

Greg Grabowski described his division within the Department of Pediatrics at the University of Cincinnati and Cincinnati Children's Hospital. He felt that as division head he had freedom to structure his clinical, laboratory, research, and education programs as he wanted them. The biggest disadvantages he saw with his current structure were a lack of hard money and space dedicated to genetics, and an inability to hire and fire staff independently. He also considered it disadvantageous that most divisions cannot provide training to graduate students, that the parent department taxes the revenues of the divisions, and that genetics is considered to be a pediatric subspecialty and a clinical service rather than a basic science.

Jerry Schneider described the organization of medical genetics at the University of California at San Diego. There is a medical genetics division in the Medicine Department and five different genetics divisions in Pediatrics (Biochemical Genetics, Dysmorphology, Medical Genetics, Metabolic Diseases, and Molecular Genetics). He did not think that this provided an effective structure for enhancing medical genetics as a discipline.

Skip Elsas reviewed the development of medical genetics at Emory University. Recently, a new Depart-

ment of Human Genetics and Molecular Medicine was formed that incorporated Ph.D.s but not M.D.s from the Division of Medical Genetics in the Department of Pediatrics. Additional resources were provided for development of the new department, but some difficulties have occurred in changing the structure. The two units are located at different sites, and all University funding for genetics flows to the Department, although traditional genetic clinical and laboratory services remain in the Pediatric Division.

Emmanuel Shapira reported that medical genetics at Tulane University is organized as a center that has all of the advantages of a department, including its own defined space and faculty positions, its own endowments, a direct reporting relationship to the Dean, a seat on key administrative committees, and the ability to grant a Ph.D. degree in genetics. This structure emphasizes the position of genetics as a bridge discipline between the basic and clinical sciences. The biggest potential disadvantage is that faculty appointments, promotion, and tenure must be handled through arrangements with one of the formal academic departments.

Uta Francke described the Genetics Department at Stanford University. Genetics is one of 11 small, basic science departments. Clinical genetics services and training are provided in the Pediatrics and Obstetrics Departments, and the clinical genetics laboratories operate in the Department of Pathology. The Genetics Department is responsible for the medical student course in genetics and for a Ph.D. program. There is a weekly Human Genetics Journal Club in which faculty and students from many different departments participate. There is also an Interdepartmental Postdoctoral Training Program for both M.D.s and Ph.D.s. These joint undertakings work well because they are open, shared, and voluntary. However, medical genetics as a discipline is dependent on the good will of several different department chairs; there is no independent budget or space.

A new Department of Human Genetics was created at Mount Sinai School of Medicine in November 1993, Bob Desnick reported. The creation of this department was recommended after an in-depth review of the basic sciences there. Human Genetics is a "hybrid" department and includes clinical services as well as basic science. An important strength of the department is that it has clearly articulated research themes and budgeted responsibilities for research, clinical services, and education.

The transition from a division to department has provided several important advantages at Mount Sinai. These include direct reporting to the Dean, the ability to make primary faculty appointments, the ability as a hybrid department to balance clinical and basic science interests, access to dedicated space, the availability of a hard-money budget, better control of educational and clinical activities, and improved visibility and stature within the medical school and hospital.

Feige Kaplan described the evolution of the human genetics program at McGill University. In 1979, a Centre for Human Genetics was created that included faculty from clinical departments as well as from a previous Department of Genetics. The Centre's Director reported jointly to the Deans of Science, Medicine, and Graduate Studies. Faculty of the Centre held appointments and were given space in various departments and hospitals throughout the city. The Centre fostered research and clinical activities in medical genetics and provided recognition of the discipline within the university. However, the Centre did not have sufficient administrative, fiscal, and space resources, and it encountered particular difficulties in recruiting because of the need to obtain primary appointments (and funding) from various departments. In 1993, McGill created a new Department of Human Genetics. The key advantage of this new structure is its ability to control the destiny of medical genetics as a discipline at the university.

Richard King described the Institute of Human Genetics at the University of Minnesota. Before the creation of this Institute in 1985, genetics faculty were dispersed in seven different units in three colleges of the University and in the State Board of Health. The Institute incorporated both basic and clinical genetics. It has its own budget and space but cannot make primary faculty appointments. The director sits on key administrative committees within the University and the primary teaching hospital. Dr. King believes that the Institute structure is less rigid and permits more rapid response to changing circumstances than a department. The biggest potential disadvantage is the inability to make primary academic appointments, although this has not really caused problems so far.

Ed McCabe summarized the changes that have occurred in the organization of medical genetics at Baylor College of Medicine. Genetics divisions were established in the Departments of Medicine and Pediatrics in the early 1970s. In 1985, an Institute of Medical Genetics was formed which functioned much as a department with its own budget, research laboratories, and clinical services. The main problem with this arrangement was that the lack of formal departmental status left the Institute vulnerable to changes in funding and administration. In 1994, a Department of Medical Genetics was established that includes both basic science and clinical faculty and activities. This change has been beneficial, resulting in increased visibility, credibility, and stability for medical genetics.

Dr. McCabe, who recently became Chairman of Pediatrics at the University of California at Los Angeles (UCLA), reported that UCLA has a number of separate clinical divisions that work together through the Intercampus Genetics Training Program. Faculty are appointed in different departments. This makes it difficult to set priorities for medical genetics and puts the discipline in a vulnerable position. As Chairman of Pediatrics, he hopes to develop a program in medical genetics as a step toward establishing a freestanding department.

Maimon Cohen provided a perspective on this session from the point of view of a medical school dean. He concluded that there is no universal best answer as to how to organize a medical genetics unit. In order to improve the stature of medical genetics within any medical

school, it is necessary to develop a solution within the current climate of medical economics. It seems likely that reimbursement for medical genetics services will decrease as managed care increases. Clinical practice income has traditionally provided a substantial portion of the support for medical education and clinical research. Thus, many of medical genetics academic activities are also seriously threatened. Medical genetics units must deal with the changing market with speed and decisiveness. This may mean doing less, but doing it more efficiently. Medical genetics units must develop a change-oriented culture and a common vision with colleagues in the teaching hospitals.

Funding for research is also under pressure. For-profit companies are now receiving 30% of NIH investigator-initiated grants in genetics. It is increasingly expensive to set up laboratories for new faculty, even if funding can be found for their salaries and space for their research. Clinical faculty need to have protected research time despite their difficulty in maintaining clinical incomes. Genetics diagnostic laboratories are unlikely to provide the income that has been used to support academic activities in the past. In this climate, improving the structure and stature of medical genetics units within medical schools will be difficult. The process may take many years and proceed incrementally. In order to succeed medical genetics units must assure that the research, teaching, and health care they provide is (and is seen to be) an indispensable part of the academic medical center.

SESSION 3: GENETICS IN NEW MEDICAL CURRICULA (SKIP ELSAS AND JAN FRIEDMAN, CHAIRS)

Jan Friedman discussed the American Society of Human Genetics Model Curriculum in Genetics. Among the most important features of the Model Curriculum is its preamble which asserts that 1) genetics is both a basic medical science and a clinical specialty, 2) genetics needs to be taught during all phases of medical school, 3) genetics must be explicitly included in the curriculum, and 4) a person or committee should be given specific responsibility for all medical student teaching in genetics.

The USMLE is becoming the only route to medical licensure in the United States. The examination consists of three "step" examinations, each of which includes four sets of multiple-choice questions given over 2 days. Many genetics questions were recently written for the Step 1 and Step 2 examinations by an ad hoc Genetics Task Force which has now been disbanded. All genetics questions written by the Task Force must be selected for inclusion (or not) by a Test Materials Development Committee in a traditional basic science. There is no Test Materials Development Committee in genetics. At present, 10–15% of the Step 1 questions relate to genetics in some fashion, but there is much less genetics content on the Step 2 examination and almost none on the Step 3 examination.

The current status of genetic teaching in various medical schools was reviewed.

Skip Elsas described genetics teaching at Emory University School of Medicine. Medical genetics and molecular biology are taught in the spring semester of the first year in a course that consists of 47 lectures, 15 small group sessions, and three written examinations. Twenty to 25 faculty from various departments, and an equal number of teaching assistants, are involved in the course for 110–120 medical and graduate students. The small groups, each of which includes 5–7 students, have a wide range of content from hands-on karyotyping through problem-based learning sets to reading original literature on current topics. Each small group session is preceded by a breakfast meeting for the tutors in which the content and objectives of papers and handouts for the following week's sessions are discussed. Genetics teaching in the other 3 years of medical school at Emory consists of clinical electives and incidental involvement in problem-based learning sets.

Bruce Korf described Harvard's medical genetics teaching. There is a 6-week block devoted entirely to medical genetics, embryology, and reproductive biology, given to all students in the spring of the first year. During this block, students receive 61 hr of lectures (30 hr in genetics), nine patient presentations (all in genetics), one ethics panel, and 36 hr of case-based problem-oriented tutorials. The course is organized around 10 core concepts that emphasize the clinical importance of the material presented. Tutors are all faculty members, but most have primary appointments outside the Department of Genetics. Tutors are supplied with detailed guides to each case and meet together for review prior to presentation of the case to the students. The active participation of students in the learning process and their desire to solve the problems presented are important motivating factors. The use of patient presentations is especially effective in helping students understand the human context of genetic disease.

Hunt Willard described medical genetics teaching at Case-Western Reserve University, where a nondepartmental systems approach to teaching was adopted more than 30 years ago. Genetics is taught as part of the Cellular and Developmental Biology block given to first-year students. This is an 8-week block that also includes biochemistry, molecular biology, developmental biology, and microbiology. The course meets 4–5 hr a day, 6 days per week. All teaching is provided as lectures, and about 30 hr are devoted to human genetics.

SESSION 4: THE CASE FOR FREESTANDING MEDICAL GENETICS RESIDENCIES (JEWELL WARD, CHAIR)

By 1998, all clinical genetics residency training in the United States will require approval by the Accreditation Council for Graduate Medical Education (ACGME). The Council has established a Medical Genetics Residency Review Committee (RRC) which has assumed responsibility for all graduate medical education accreditation, including clinical fellowships. With RRC oversight, clinical genetics residencies can now be established and approved. ABMG will continue to ac-

credit training programs for Ph.D. medical geneticists as well as for clinical cytogeneticists, clinical molecular geneticists, and clinical biochemical geneticists.

Ross McLeod presented an overview of the development of Canadian genetics residency programs. The Canadian College of Medical Geneticists was formed in 1975. The College developed guidelines for 2-year fellowships in clinical genetics, cytogenetics, biochemical genetics, molecular genetics, and Ph.D. medical genetics. By 1987, the college included 100 fellows.

The Royal College of Physicians and Surgeons of Canada approved medical genetics as a freestanding specialty in 1989. There are currently five programs approved by the Royal College for medical genetics residency training. The Royal College residency lasts 5 years. Trainees are admitted right after graduation from medical school or after 1 year of internship.

Dr. McLeod described the University of Calgary program, which is typical of the Royal College medical genetics residencies. The first year is spent in either pediatrics or internal medicine. The second year includes 8 months of either internal medicine or pediatrics (whichever was not taken in the first year), 2 months of high-risk obstetrics, and a month of crisis counselling outside of a genetics center. The third year involves rotating through the cytogenetics, biochemical genetics, and molecular genetics laboratories, and years 4 and 5 consist of 18 months of clinical genetics and 6 months of research. The genetics program is "home" for all 5 years, with gradually increasing responsibility for genetics patient care, teaching, seminars, and rounds. The residency program is funded through the university. Candidates must successfully complete written and clinical examinations when their training is concluded to obtain Royal College certification.

Suzanne Cassidy described the development of medical genetics residency programs in the US. The purpose of the residency is to train physicians to *practice* clinical genetics. An alternative approach to the traditional 2- or 3-year clinical genetics fellowship, the 4-year freestanding clinical genetics residency includes 2 years of "pregenetics" training in other ACGME-accredited programs within the institution. A resident might add a fifth year for research. The institution must support all 4 core years.

The advantages of a freestanding program include better-rounded pregenetics training, longer exposure to genetics conferences and seminars, possible involvement in long-term clinical or laboratory research projects, and a shorter total training period than the current combination of specialty residencies and clinical genetics fellowships. The disadvantages include increased administration and possibly greater expense, difficulty in getting funding for long-term training slots, and little experience with the quality of training provided in this manner.

Steven Nestler, Secretary of the Medical Genetics RRC, summarized the current status of the ACGME, which reviews over 95,000 residency and fellowship positions throughout the US. There are now 26 separate residency review committees, medical genetics being the newest. Twelve institutions have returned completed applications for medical genetics residency programs which have been processed; 10 of these are 2-year (i.e., fellowship) training programs only. Ten programs have been granted provisional accreditation; none have been site-visited yet. Two 4-year residency programs have been approved.

The Medical Genetics RRC will maintain minimal requirements but will not be "cutting edge." A newsletter is planned for program directors, with proposals for changes in requirements. The RRCs are not charged with manpower needs assessments.

Bob Desnick summarized the Medical Genetics Residency Training Program that has been established at the Mount Sinai School of Medicine. The first year of training in clinical genetics (CGY-1) includes rotations in general pediatrics (6 months), pediatric and neonatal intensive-care units (3 months), term nursery (1 month), pediatric neurology (1 month), and pediatric emergency room (1 month). CGY-2 includes pediatric ward chief (3 months), pediatric and neonatal intensive care units (2 months), fetal assessment (1 month), and various pediatric and adult specialty rotations (6 months). CGY-3 and CGY-4 include clinical genetics (7 months), biochemical genetics (5 months), cytogenetics (2 months), molecular genetics (2 months), pathology (1 month), developmental pediatrics (1 month), and 6 months of electives or research. Trainees attend lectures, journal clubs, conferences, and seminars. The program will accept two CGY-1 residents per year; the faculty includes 12 M.D. and seven Ph.D. geneticists.

Jewell Ward summarized the effect that managed care has had on funding of graduate medical education in Tennessee. In the past, graduate medical education funding flowed from Medicare and Medicaid to hospitals. On January 1, 1994, Tennessee abruptly began a "TennCare" program through multiple managed-care organizations (MCOs) to replace Medicaid for the state. Continuance of training funding was not required of MCOs by the state. The major university medical centers formed their own MCOs, but due to inequality in assignment of patients and lack of state oversight of marketing practices, these MCOs have a minority of covered lives. On January 1, 1995, a shortfall in funds was announced, and it was stated that funds for training were no longer available outside of those allocated to MCOs. Every graduate medical education program in the state was affected by this decision, but the large programs with a disproportionate share of TennCare patients, i.e., the large metropolitan public-service hospitals, were hit hardest. As a result, some primary residency training positions were cut for 1995–1996. Future solutions may involve funding directly through medical training institutions.

The session provoked considerable discussion. Issues raised included:

1) Sources of funding for medical genetics residencies and fellowships.

2) The desirability of a needs assessment for clinical geneticists.

3) The ability of individuals in freestanding residency programs to obtain jobs and promotion in medical school or hospital departments other than medical genetics.

4) The need to compare the clinical competence of individuals trained through freestanding residencies with that of traditional trainees.

5) Competition within institutions for residency slots.

6) The need for trainees to make an earlier career decision in order to pursue a freestanding medical genetics residency.