

ASSOCIATION OF PROFESSORS OF HUMAN OR MEDICAL GENETICS
Minutes from the Business Session of the 2000 Workshop

SIXTH ANNUAL WORKSHOP
SHERATON SAND KEY RESORT
CLEARWATER BEACH, FLORIDA
APRIL 14-17, 2000

Session I: APHMG Activities and Interactions

The session was called to order by Dr. Skip Elsas, President.

Minutes of October 22 Meeting in San Francisco were approved. The Treasurer*s Report was delivered by Dr. Mary Kay Pelias. A copy of the report is attached to these minutes. Dr. Gerald Feldman was approved as the incoming Secretary-Treasurer; he will assume these responsibilities July 1, 2000.

Progress Reports:

Genetic Issues from the AAMC/CAS Meeting were discussed by Dr. Desnick. CAS and Mandal Groups are contacts to advance the genetic agenda, while AAMC addresses, for example, issues of salaries.

Liaison Committee on Medical Education (LCME), discussed by Dr. Korf, published "Functions and Structures of A Medical School" as a document for serious study. The leadership of LCME is planning to recognize genetics as a fundamental component of instruction in medical school. This is in recognition of requests by students for more instruction in genetics. These requests are included in "graduating questionnaires" filled out by medical students.

National Coalition for Health Professional Education in Genetics (NCHPEG) and USLME were presented by Dr. Mimi Blitzer. Dr. Desnick emphasized that we should use test scores to bring genetics to the attention of medical education faculty so that more genetics questions will be included in Parts I, II, and II of the exam. APHMG should appoint 4 or 5 representatives to read the 600 questions for the genetics tests. Dr. Blitzer noted that NCHPEG is hiring an executive director, coordinates teaching in more than 100 organizations, provides an "information highway," has a committee on licensing and certification, and concentrates on core competencies in genetics for health care professionals. "Genetics in Primary Care" is the product of a contract to the Society of Teachers in Family Medicine, with RFP*s for primary care in family medicine, internal medicine, and pediatrics, for training professionals in genetics.

American College of Medical Genetics, represented by Dr. Reed Pyeritz, has a membership of about 1100, with a \$1m reserve in its financial balance sheet but income weak and likely to worsen. The College publishes "Genetics in Medicine." Current issues and interests include clinical practice guidelines, patent controversies, strategic planning, advocacy roles, standards of care, and translating the results of the Human Genome Project into medical benefits.

APHMG Question Database, reported by Dr. Peggy McGovern, is ready for updating and possible presentation on CD ROM. Collecting small group problems will be emphasized in the next round.

Development of Canadian Institutes for Health Research, reported by Dr. Jan Friedman, is the replacement for the MRC, with a broader research mandate, including clinical and social interests. Canada will have 5 new Genome Canada Centres, with matching funds from the private sector, and generously funded research chairs.

Review and Vote on Amended Guidelines for Medical Genetics Training Program was led by Dr. Claire Leonard, who noted that subspecialties in genetics should have a voice in their own areas on this first pass. Dr. Herb Lubs and others noted that the guidelines should be consistent in language throughout, particularly with regard to "recommend" and "require."

Other Business included a call for nominations for President and one Councillor, with introduction of Drs. Desnick, Pyeritz, and Drew Carroll as the Nominating Committee.

Session II: Recent Advances in Neurogenetics

Dr. Bruce Korf introduced this scientific session as a response to the question of how geneticists will be able to work with colleagues in other fields to effectuate clinical applications.

Dr. Joseph Martin, from Harvard University, delivered an "Overview of the Molecular Bases of Neurodegenerative Disorders." His discussion included the molecular genetics, clinical manifestations, pathological correlations and treatments of Huntington disease, spinal cerebellar atrophy, spinal muscle atrophy, Machado-Josef disease, dentatorubropallidoluysian atrophy, and Alzheimer disease. Also discussed were the quantitative phenomena of loss-of-function and gain-of-function as well as the qualitative phenomenon of change-of-function in these diseases.

Dr. Rudy Tanzi, also from Harvard, discussed "Recent Advances in Understanding and Intervention for Alzheimer Disease," the most common form of dementia. Research now focuses on how to lengthen the life of neurons and prevent or inhibit the formation of amyloid plaques. Genetic research aims at locating and defining the genes that cause Alzheimer disease and using this information for diagnosis and treatment modalities.

Session III: Bioinformatics

Dr. Jan Friedman chaired this session.

Dr. Bonnie Pagon addressed the question, "Is Bioinformatics of Any Use to Clinical Geneticists?" Two sources of information are (1) GeneTests, open to the public with lab consent, a clinic and lab directory, with an education component, funded by MCHB, and (2) GeneClinics, open to the public, with disease profiles and overviews of 75 monogenic and common disorders, funded by NIH. Dr. Korf noted that the computer-based genetics course at Harvard requires students to use these two sources and OMIM for instruction.

Dr. Muin Khoury's presentation on "Bioinformatics for Genetics and Public Health: The Human Genome Epidemiology Network (HuGENet)" delineated the types of data collected. Categories of data include prevalence of variants, burden of disease and disability, disease risks associated with gene-gene and gene-environment interactions, evaluation of clinical validity and utility of genetic tests, evaluation of determinants and impact of genetic services and testing in different populations, and types of information needed by public health officials and policy makers, by researchers, health care providers, and consumers.

Dr. Alan Guttmacher discussed "GROWing a New Breed of Geneticists: Genetic Resources on the Web." The number of organizations with a "presence" on the Web now stands at about 36, but this presence is hampered by the limited pool of writers of quality documents. These Web Sites anticipate use by non-professionals. Entry is at <http://search.info.nih.gov/grow/>.

Dr. Jan Friedman queried, "Should Bioinformatics Replace Genetics?" and noted a broad array of functions for bioinformatics in genetics. Bioinformatics is essential to most research in molecular biology and molecular genetics, in genetic medicine, and in teaching genetics. However, we do not "own" bioinformatics, and we should use this source of knowledge as an encyclopedia. Bioinformatics will not replace humans as integrators of biology and medicine. Nor will it translate science into patient care or serve as a conscience in the applications of genetic information.

The ensuing panel discussion noted that computers do not talk to patients or students. Further, physicians who use the web may impart incorrect information in an effort to avoid expensive referrals. The consensus is that computers will not replace human geneticists.

Session IV: Integrating Medical Genetics Into Clinical Years of Medical School

This session was chaired by Dr. Art Beaudet.

Dr. Priscilla Short discussed "Current Status of Education Resources in the U.S.A." These resources include the AMA (mostly for CME credits), ASHG Information and Education Committee Core Curriculum, clinical clerkships, AAMC exit interviews, and LCME surveys.

Dr. Friedman discussed Canadian Resources, noting that Canada has nothing that is strictly Canadian in the 3rd and 4th year medical curriculum and that the Canadian board exams are abysmal in genetics.

Dr. Beaudet discussed "Electronic Methods at Baylor," including a shift toward small group learning and a move to make genetics a "theme" in the curriculum, with computer modules created in the 3rd and 4th years. Whether students "like" the courses nearly always depends on whether the students like the teacher (i.e. personality) and how much effort is put into preparing lectures.

Dr. Peggy McGovern discussed "Simulated Clinical Education," with "standardized patients" as actors who "teach" students how to take histories and physicals, how to convey information, and how to interview patients. These methods have several advantages, including eliminating risks to patients.

Dr. Mimi Blitzer discussed the "Examination Process (USMLE)," noting that NBME governs parts I, II, and III, on basic science, clinical diagnosis and disease pathogenesis, and clinical management respectively. Genetics is still underrepresented in part III. We must continue to push genetics and provide persons to write genetics questions.

Dr. Suzanne Cassidy discussed "Clinical Education at Case Western Reserve University," noting 1 hour of genetics in each pediatrics rotation, none in ob/gyn, but a fair amount in 2nd year clinical correlations - in all, not a very good situation.

The ensuing panel proposed an "Action Plan," to include (1) 10-15 one hour computer modules for 3rd and 4th year students in all major blocks; (2) 2-3 modules for residents in various subspecialties; (3) 200 assignment sheets for

exercises on GeneClinic, OMIM, and PubMed; (4) counselors of faculty to elicit family history confidentially on students; and (5) use of standardized patients. APHMG might consider forming committees for (1), (2), and (3) above.

Family History Problems at the Medical College of Virginia and elsewhere were discussed by Dr. Walter Nance, with a query that APHMG should respond to OPRR on the issue of informed consent for family histories and the concept of waiver of consent in low-risk situations. A letter written by Dr. Rich Spritz, at the University of Colorado, will be circulated by e-mail for endorsement.

Business Meeting included ratification of Dr. Bruce Korf as President Elect and Dr. Bronya Keats as Councillor.

Social Events included several excellent meals, a "Marine Life Adventure" tour with biologists from the Clearwater Marine Aquarium, and an evening of cruising, dining, and dancing on the chartered yacht "Starlight Majesty." APHMG is grateful for the many contributions of Nancy Elsas, Rhonda Heermans, and Gayle Pullen, to the success and pleasure of this meeting.

The next annual workshop will be held in Tucson, Arizona, at the Westward Look Resort.

Respectfully submitted,

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Interim Secretary Treasurer

Louis J. Elsas, II, M.D.
President