

2003 ASHG AWARD FOR EXCELLENCE IN HUMAN GENETICS EDUCATION The Importance of Genetic Counseling*

Joan H. Marks

Sarah Lawrence College, Bronxville, NY



Joan H. Marks

I was deeply honored to receive the ASHG 2003 Award for Excellence in Medical Genetics Education. I am grateful to the committee and those who nominated me for this wonderful honor. This award means not only a great deal to me personally but also to all members of the new profession of genetic counseling.

It is a tradition to thank one's colleagues for their participation in a research project. In this instance, I believe that the success of master's-level genetic counselors, a program that was initiated at Sarah Lawrence College, is due to a major extent to those "pioneer" graduates from

Sarah Lawrence. These early graduates established the field by showing what they could accomplish in the practice of medical genetics as advocates for patients seeking understanding of their particular genetic risks. Several of the early graduates were hired by the geneticists with whom they did their student training. These centers, all in the New York area, included Albert Einstein College of Medicine, New York Hospital–Cornell College of Medicine, Creedmoor Psychiatric Hospital, Long Island Jewish Hospital, and, in the late 70s, Presbyterian Hospital–College of Physicians and Surgeons, Columbia University. By 1980, the Human Genetics Program at Sarah Lawrence was attracting students from all over the country, and these graduates were also recruited to join a genetic counseling service in many states across the nation.

When the first grants were awarded to states under the National Genetics Diseases Act of 1976, most of the states who applied for funds to develop a statewide genetic service program included salaries to recruit master's-level genetic counselors. The fact that an innovative, high-risk program in graduate education could be initiated and flourish at Sarah Lawrence—with a concentration in the humanities, not the sciences—owes a lot to the culture of the college, a culture that emphasizes initiative and the individual.

The hallmark of the training of genetic counselors is based on the premise that understanding that the emotional component of genetic risk is central to providing good genetic services. That integration of medical-genetics training with psychological-counseling skills has remained at the core of all graduate programs training genetic counselors now offered in the United States and abroad.

The curriculum at Sarah Lawrence originally concentrated on basic science courses, such as human physiology, medical genetics, probability and statistics, and cytogenetics. By the mid-70s, course work was developed in the psychological component of illness and loss. At the same time, a serious effort was made to recruit students who had previous experience working with patients who had undergone traumatic, stressful situations. As our contact with patients seeking genetic services increased, so did our appreciation that the emotional component of being at risk for passing on one's malfunctioning genes was a traumatic experience for most patients. Learning to provide

Received November 26, 2003; accepted for publication November 26, 2003; electronically published February 19, 2004.

Address for correspondence and reprints: Joan H. Marks, Sarah Lawrence College, Bronxville, NY 10708. E-mail:joanh-marks@mskcc.org

* Previously presented at the annual meeting of The American Society of Human Genetics, in Los Angeles, on November 8, 2003.

© 2004 by The American Society of Human Genetics. All rights reserved. 0002-9297/2004/7403-0007\$15.00

supportive care and sensitivity tailored to each patient's particular dynamic was something that needed to be taught. Selecting a student who had already shown an interest in providing this level of care seemed promising for future training. All applicants were required to have a personal interview with the director and assistant director of the training program.

The counseling component of the curriculum soon matched the number of hours devoted to the sciences. A two-semester seminar in genetic counseling was developed by a psychiatrist. I began to work individually with students in a weekly seminar, as well as one-on-one conferences, both devoted to helping students open up to their own life experiences and, as patient contacts developed, to their patients' emotional needs and problems in seeking genetic services. A one-semester course on interviewing, based on the concepts of Carl Rogers, was introduced in the early 1980s and remains part of the core curriculum today. The science curriculum expanded to include reproductive genetics, developmental biology, biochemistry, introduction to clinical medicine, and a thesis requirement. An oral exam based on developing an impromptu case plan for a typical genetics referral was also introduced.

Going back to the 1970s, the emphasis was on prenatal diagnosis and birth defects. Over the decades, as our understanding of the role of genetic risk for diseases has expanded, so has the role of the genetic counselor. In the next decades, genetic counseling will undoubtedly have an increasing and ever-more-challenging role in delivering health services. Alan Guttmacher has said, "Stunning scientific and technological advances in genetics will mean little if they do not benefit people." That is the challenge today for genetic counselors as they begin to apply the scientific knowledge that the Human Genome Project has made available. The process of genetic counseling also includes, to a large extent, the need to educate patients and the public in a nontechnological language that will be understandable to people at all intellectual levels. These same skills must—and will—be utilized to play an important role in helping the public become aware of the many facets of our new knowledge

and its implications for individual patients wanting to know about their genetic makeup.

I was first invited to speak before the American Society of Human Genetics when I had submitted an abstract about the Sarah Lawrence Human Genetics Program for the 1974 meeting in Portland, Oregon. On a hot July day, Fred Hecht, the program chair, called me to say my paper had been chosen for the plenary session! I was shocked! I assured him I had no intention of presenting this paper, but that I merely wanted to put the new profession on the map! He responded that I had no choice but to present the talk! That was a unique day for me, as I had never presented a talk to a large group, not even the PTA! As Victor McKusick introduced me to the audience, he referred to me as "Doctor Marks!" I had a few moments on the way to the podium to decide whether to accept my promotion to medical doctor or to concentrate on the job for the day: to convince a doubtful audience that master's-level-trained genetic counselors had a real contribution to make in the world of medical genetics and health care.

In the year 2003, there has been a remarkable acceptance of the importance of the role of the master's-level-trained genetic counselor. Traditionally, based in genetics clinics and medical centers, that role now encompasses all medical specialties, health-care management, health-professional education, public health, and the use of technology to deliver genetic services. In cancer genetics alone, the genetic counselor has become an integral member of the oncology team. Industry, too, continues to recruit many genetic counselors to both promote utilization of genetic screening tools, but also to educate management about medical genetics on a worldwide basis.

Today, I am still not an M.D.! But this award signifies that medical genetics has accepted the profoundly important work that genetic counselors perform as members of the genetics team. While I am enormously grateful for the recognition of my role in this development, I am even more appreciative of the fact that the committee's decision commends the field of genetic counseling itself for our role in helping so many patients at genetic risk for illness and disability and for improving the quality of care these patients receive.