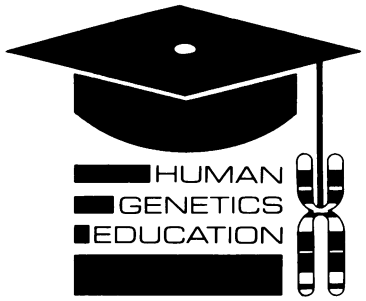
ASHG ACTIVITIES RELATIVE TO EDUCATION

Human Genetics as a Component of Medical School Curricula: A Report to The American Society of Human Genetics



Vincent M. Riccardi* and Roy D. Schmickel†

*Neurofibromatosis Program, Baylor College of Medicine, Houston; and
†Department of Human Genetics, University of Pennsylvania Medical School, Philadelphia

Summary

In recent years, there has been a remarkable increase in both the rate of acquiring new information about human genetics and the importance of human genetics for modern health care. As a result, human genetics educators have queried whether the teaching of human genetics in North-American medical schools has kept pace with these increases. To address this question, a survey of these medical schools was undertaken to assess how human geneticists perceive the teaching of human genetics in their respective institutions. The results of the survey, begun and completed in 1985, indicate the following: (1) the teaching of human genetics in medical schools is extremely variable from one institution to another, with some schools having no identifiable human genetics teaching at all; (2) the relevance of human genetics to other basic science and clinical disciplines apparently leads to noncategorical or fragmented teaching of human genetics, which may also contribute to the absence of a specific medical school course in the subject; and (3) there is a need for closer collaboration between human genetics educators and their respective medical school administrators and curriculum committees.

Introduction

It is widely accepted that human genetics is central to the bodies of knowledge that are the responsibilities of modern medical schools. However, human genetics educators have become increasingly concerned about the adequacy of human genetics components of medical school curricula (Childs et al. 1981; Childs 1982). One step in dealing with this concern is to determine whether there is a real discrepancy between what is taught and what could and should be taught. To identify discrepancies between one medical school and another in the teaching of human genetics, a survey of each medical school in Canada and the United States, including Puerto Rico, as listed in the 34th edition of Medical School Admission Requirements (1985), was undertaken.

The present survey was not intended to duplicate a similar, but more elaborate, survey of medical school human genetics teaching published six years before by Childs, Huether, and Murphy (Childs et al. 1981), although the conclusions from the present study are not unlike those reported previously. Likewise, the present survey was not intended to replicate the major review of the human genetics teaching efforts made by American dental schools during the late 1970s and early 1980s, the publication of which

Received October 19, 1987; revision received November 4, 1987.

Address for correspondence: Vincent M. Riccardi, Neurofibromatosis Program, Baylor College of Medicine, One Baylor Plaza, Houston, TX 77030.

0002-9297/88/4204-0018$02.00

(Farrington et al. 1982) has contributed to improvements of human genetics teaching in many dental schools. Rather, *the purpose of the present study is to assess how human genetics educators perceived the situation in 1985.*

Material and Methods

Before we proceed to the survey itself, it may be useful to clarify several terms and phrases. "Genetics" in the broadest sense is the science of variability, accounting for differences and similarities from one individual to another, from one species to another. "Human genetics" is the science of human variability, dealing with variation in terms of specific human genes and the factors that influence the frequency, nature, and consequences of those genes. "Medical genetics" is the medical discipline that concerns itself with the contribution of the human genome (and potential and proved influences on it) to compromised health and well-being. "Clinical genetics" is the medical subspecialty that addresses the application of medical genetics principles in day-to-day clinical practice. We emphasize that *the primary subject of this report is the science of human genetics as it is taught in medical schools*—and not the day-to-day clinical application of human genetics in terms of medical genetics or clinical genetics.

"Human genetics teaching" may take the form of either isolated lectures on human genetics topics *or* a structured human genetics course comprising a coordinated series of lectures on human genetics topics delivered in an ordered sequence over a circumscribed time period. The primary concern of this report is the latter, the teaching of human genetics through a structured course devoted to the subject. Our viewpoint is that, because human genetics is central to a modern understanding of human health and disease and because the concepts of human genetics as a discipline are unique, the teaching of the principles involved is probably best done by utilizing the strategies and impact of a structured course.

The teaching of human genetics in the clinical years of medical school is not a focus of this survey. While it would be safe to say that all medical students are exposed to clinical genetics problems in their clinical rotations, evaluation of the teaching elements of these experiences would be exceedingly difficult and, in any case, beyond the original intentions of our survey.

The Survey

The *survey letter* was addressed preferentially to a geneticist known or thought to be responsible for teaching human genetics in one capacity or another. If the identity of the presumed human genetics course teacher/organizer was unknown, the letter was addressed to the dean of the medical school. The survey instrument itself was addressed "Dear Colleague" and asked for the following information and materials: the name of the human genetics course and its teacher/organizer, a course schedule, a course syllabus, sample examination questions, an itemization of the course's strengths, and an itemization of the course's weaknesses or of problems in conducting or providing the course. Approximately three months after the initial inquiry, for each medical school that had not responded, a reminder and copy of the original request was sent, either to the original contact or an alternative person. After another two months, telephone inquiries were attempted for each of the schools that had not responded to either inquiry by mail.

Responses to the survey were considered from three vantage points: (1) quantitative analysis of data (e.g., determining the average number of hours in medical school human genetics courses), (2) qualitative analysis of (*a*) the nature and character of the overall human genetics teaching effort and (*b*) selected components thereof, and (3) itemization of problems cited by respondents.

Results

Quantitative Data

By mail and/or by telephone conversations, information was received from 119 of the 140 medical schools surveyed; the respondents themselves had various levels of responsibility, ranging from upper-echelon school administrators to faculty members responsible for the teaching. From these 119 schools, seventy-nine course schedules, forty-one syllabuses, and forty sets of examination questions were received. There were sixty-nine independent human genetics courses, as well as twenty-nine courses that were integrated into another course, typically "pathophysiology." Among these ninety-eight human genetics courses, fifty-one were taught in the first year, thirty-nine in the second year, and eight in both the first and second years.

Table 1

Numbers of Hours Assigned to Human Genetics Lectures and Discussion

Hours	No.
0	21
1–10	10
11–20	30
21–30	30
31–40	9
>40	3
	103

For 103 of the 119 responding medical schools, it was possible to tally the number of hours devoted to lectures and discussions. The number of hours for human genetics lectures was extremely variable (see table 1). The mean average allotment for teaching human genetics was eighteen hours. If only schools with one or more hours were included, the average was 21.6 hours, less than the 24.3 hours reported six years previously by Childs et al. (1981); although the methods of data collection in the two studies were different, the results presented here suggest that there has been no significant progress in expanding medical school human genetics teaching efforts.

As shown in table 2, the responsibility for teaching human genetics was assumed by departments of pediatrics more often than by all other departments combined: fifty-five pediatrics departments (52 percent of the sample) were responsible for the human genetics course. For the remainder of the medical schools surveyed, the departments responsible for teaching human genetics also are listed in table 2. As we expected, if the school had a department of genetics, it was responsible for the human genetics course.

Table 2

Departments Responsible for Teaching Human Genetics

Department	No.
Pediatrics	55
Genetics	20
Medicine	6
Anatomy	5
Biochemistry	5
Microbiology	5
Obstetrics/gynecology	5
Pathology	2
Biomedical science	2
Community medicine	2

Qualitative Assessment

The authors evaluated the content of the human genetics courses on the basis of the course schedules and the syllabuses. There was a remarkable consistency in the designated topics; cytogenetics, Mendelian principles, and prenatal diagnosis were covered by every school. In general, at least six hours were devoted to these topics. In addition, most schools taught something about multifactorial/polygenic inheritance, and several included elements of the subject of inborn errors of metabolism. A twenty-four-hour human genetics course was likely to devote the hours as follows: cytogenetics, five hours; Mendelian disorders and inheritance patterns, six hours; inborn errors of metabolism, six hours; multifactorial/polygenic inheritance, three hours; genetic counseling, one hour; prenatal diagnosis, one hour; cancer genetics, one hour; and social/ethical/legal issues, one hour.

All human genetics courses had some clinical content, but only a portion had elements of normal biology. We were thus able to distinguish those courses with only a clinical approach from those with an additional, basic or biological approach. One way of making this distinction was to determine whether the gene was treated merely as a heritable unit in a Mendelian sense or as a complex informational molecule. For example, courses considered to have a biological approach made reference not only to the triplet codon but also to promoter sequences, consensus sequences for processing, enhancers, or polyadenylation signals. In so-called clinical courses, variation appeared to be treated as a theoretical "mutation" or, sometimes, as a base substitution; but apparently it was not considered, for example, in terms of transposition, amplification by unequal homologous exchange, or alternative splice junctions, and so on. Of the seventy-nine course schedules examined, only twenty appeared to designate discussions about gene structure and the molecular basis of gene expression. The lack of basic science information about genes was striking. While this might reflect merely organizational fragmentation, there is concern that the students are left without an appreciation of human genetics as a *distinct entity* with both clinical *and* basic science components. Although most of the twenty courses that included the biology of the

gene were taught during the first year of medical school, the majority of first-year courses were purely clinical medical genetics. As a strictly clinical course, the genetics teaching effort was said to be both well received by the students and often the first one in which a student was presented with a clinical problem.

We also attempted to gauge the apparent overall adequacy of the human genetics teaching effort for each medical school. Our primary goal here was to estimate what proportion of medical schools had good or excellent human genetics teaching efforts as indicated by their response to the survey, the quality of the materials provided (e.g., course syllabus), and the topics covered in the course outline. "Good" was applied to a human genetics course with modest emphasis on basic biology and taught by an identifiable human genetics faculty. "Excellent" was applied if there were twenty-five to forty or more hours of human genetics lectures in a course taught by a strong, diverse faculty who utilized recent information derived from basic biology to exemplify and clarify the salient principles of human genetics in the medical setting. "Poor" was applied to schools with apparently no human genetics teaching. A final group, whose human genetics teaching efforts fell between the two extremes, was designated to be "fair."

Among the 119 respondents to the survey, at least twenty-one medical schools had neither an identifiable series of lectures in human genetics nor a human genetics course (table 1). For the twenty-one medical schools that did not respond, additional efforts were made to identify each school's human genetics course and faculty. Despite intense efforts, no such course or faculty could be identified for these schools, and they were classified to have a "poor" human genetics teaching commitment.

Thus, on the basis of these considerations, each institution was scored as to whether its human genetics teaching was nonexistent/poor, fair, or good/excellent. The number of schools in each category is given in table 3. Even when the likelihood of some inaccuracy in this assessment is discounted, the results are noteworthy: 47 percent of the medical schools were considered to have nonexistent or poor human genetics teaching and only 21 percent appeared to provide good or excellent human genetics teaching. That is, the teaching of human genetics in medical schools is very uneven, with many graduates apparently entering the practice of medicine without an adequate understanding of the role of genetics in the cause and pathogenesis of human disease.

Table 3

Assessments of 140 Canadian and American Medical Schools in Terms of Human Genetics Teaching

Nonexistent/poor	47%
Fair	33%
Good/excellent	21%

Respondents' Notations of Problems

The most frequently cited problems noted by medical school human genetics course organizers included the following paraphrased quotations:

1. "The school (i.e., administrators, curriculum committees, chairmen) does not appreciate or understand human genetics and its role in medical education."
2. "Students enroll in medical schools with an enormous range of prior experience and familiarity with genetics, human and otherwise." For example, the University of Pennsylvania Medical School noted that for one of its recent entering classes, 25 percent of the students had never had a genetics course at all.
3. "There is no suitable genetics textbook for teaching human genetics in medical schools." Thompson and Thompson (1983) was cited as the most popular and most frequently used text.
4. "The students do not take the subject seriously because it is a very small component of the overall curriculum." It was often repeated that students were under pressure to do well in their "important" (i.e., larger) courses.
5. "There are not enough teachers of human genetics." It was often considered impossible to have discussion groups or meaningful contact with students.
6. "There is not enough time allotted to teach human genetics." This was a nearly universal response, even from the schools that had courses with more than forty hours of student contact.

Discussion

The information presented here is only a portion of the total picture. However, the picture is a rather consistent one. There has been little progress during the past six years to improve the position of human genetics in the medical school curriculum. It is rarely

a major subject, and it is not usually taught as a basic science, although components are taught in other basic science courses. With few exceptions, there has not been an effective voice for presenting genetics as a discipline, as a specified and coherent subject. It appears that medical schools may have a problem acknowledging and identifying human genetics as a specific entity. As a result, the medical student is not always being prepared to understand the revolution in human genetics that seems to be changing everything except itself as a subject.

Acknowledgments

We wish to express special thanks to other members of the 1985 Information and Education Committee for their help in collecting and analyzing the survey data: Joann Boughman, Ph.D; Judith L. Benkendorf, M.S.; Carl A. Huether, Ph.D.; Richard L. Ingraham, Ph.D.; Judith H. Miles, M.D., Ph.D.; Jerome I. Rotter, M.D.; Ann C. M. Smith, M.A.; and Helga Toriello, Ph.D.

References

Association of American Medical Colleges (AAMC). 1985. Medical school admission requirements 1984–86: United States and Canada. 34th ed. AAMC, Washington, DC.

Childs, B. 1982. Genetics in the medical curriculum. Am. J. Med. Genet. 13:319–324.

Childs, B., C. A. Huether, and E. A. Murphy. 1981. Human genetics teaching in U.S. medical schools. Am. J. Hum. Genet. 33:1–10.

Farrington, F. H., R. G. Sanger, R. E. Stewart, R. J. Jorgensen, and P. Domoto. 1982. Curricular guidelines in human genetics. J. Dent. Ed. 46:184–187.

Thompson, J. S., and M. W. Thompson. 1983. Genetics in medicine. W. B. Saunders, Toronto.