The Current Status of Medical Genetics Instruction in U.S. and Canadian Medical Schools

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Abstract

Purpose
Relatively little is known about how medical genetics is being taught in the undergraduate medical curriculum and whether educators concur regarding topical priority. This study sought to document the current state of medical genetics education in U.S. and Canadian accredited medical schools.

Method
In August 2004, surveys were sent from the Indiana University School of Medicine to 149 U.S. and Canadian medical genetics course directors or curricular deans. Returned surveys were collected through June 2005. Participants were asked about material covered, number of contact hours, year in which the course was offered, and what department sponsored the course. Data were collated according to instructional method and course content.

Results
The response rate was 75.2%. Most respondents (77%) taught medical genetics in the first year of medical school; only half (47%) reported that medical genetics was incorporated into the third and fourth years. About two thirds of respondents (62%) devoted 20 to 40 hours to medical genetics instruction, which was largely concerned with general concepts (86%) rather than practical application (11%). Forty-six percent of respondents reported teaching a stand-alone course versus 54% who integrated medical genetics into another course. Topics most commonly taught were cancer genetics (94.2%), multifactorial inheritance (91.3%), Mendelian disorders (90.3%), clinical cytogenetics (89.3%), and patterns of inheritance (87.4%).

Conclusions
The findings provide important baseline data relative to guidelines recently established by the Association of American Medical Colleges. Ultimately, improved genetics curricula will help train physicians who are knowledgeable and comfortable discussing and answering questions about genetics with their patients.

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The recent mapping and sequencing of the human genome promises to revolutionize the field of biology and the practice of medicine. All aspects of medicine and all medical specialties will ultimately benefit from and be dramatically altered by changing genetic technology. In recognizing the emerging importance of genetics in medicine, the Association of American Medical Colleges (AAMC) issued a report in 2004 entitled "Contemporary Issues in Medicine: Genetics Education."[1] This report, part of the AAMC's Medical School Objectives Project, outlines the core competencies in genetics that all medical students and residents should attain by the end of their training. These core competencies provide specific recommendations regarding the attitudes, knowledge, and skills pertaining to genetics that all graduating medical students should possess, as well as effective educational strategies. Korf et al[2] have noted that the number of physicians entering the field of medical genetics is small and getting smaller. With rapid advances in medical genetics and with fewer physicians specializing in the field, all physicians must be better prepared to extend the revolution in genetic knowledge to patient care.

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Medical schools are responsible for imparting the next generation of physicians with an adequate knowledge of medical genetics so that they can take full advantage of the coming advances in genomic-based diagnosis and treatment. However, despite the emerging importance of medical genetics in health care, there has been no systematic appraisal of what and how medical students are being taught about medical genetics. The purpose of this study was to document the content and form of medical genetics education in U.S. and Canadian medical schools.

Method

In August 2004, we mailed questionnaires to 149 medical genetics course directors or other knowledgeable faculty, including curricular deans, at all 149 U.S. and Canadian medical schools accredited by the Liaison Committee on Medical Education. The names and addresses were drawn from individual medical schools' Web sites or the AAMC Curriculum Management and

Information Tool. Because our primary interest was in undergraduate medical education, we excluded individuals whose medical genetics instruction was limited to continuing medical education classes or postgraduate training programs. Follow-up mailings were sent to all nonresponders in January 2005. Those who failed to respond to this second mailing were contacted by phone or e-mail in April 2005 and were urged to return the survey. We collected questionnaires through June 2005 at Indiana University School of Medicine.

The two-page instrument consisted of 10 questions with a check-box or fill-in-the-blank design, with one space at the end for written comments or clarifications. Participants were asked about the year of medical school in which a course was taught (first, second, third, or fourth); whether the course was stand-alone or integrated into another course; whether it was taught by a single instructor or multiple instructors; the total hours scheduled for the course (<20, 20–40, 41–60, or >60); the name of the sponsoring unit; whether board-certified geneticists were involved in teaching; the instructional formats used (instructor lectures, guest seminars, group discussions, case studies, clerkships, Web-based, or other); the principal course objective (broad survey of medical genetics concepts, critical evaluation of the scientific literature regarding basic human genetics, practical training in the use of medical genetics in general practice, or other); and the specific topics covered and the time devoted to each (selected from a checklist of 30 medical genetics topics, with space for the respondent to provide additional topics if needed). An additional question asked whether medical genetics was incorporated into third- and fourth-year clinical rotations at the participant's institution. The questionnaire was designed to be completed in 5 to 10 minutes. IRB approval was obtained for this study.

Results

Of the 149 individuals we contacted, 114 returned completed questionnaires; of those, two replied that they were not currently teaching medical genetics topics to medical students. Because we were interested in the current state of medical genetics instruction, these two respondents were excluded from the analysis, yielding a response rate of 75.2% (112/149). As shown in Table 1, 46% (52/112) of the respondents indicated that medical genetics was taught as a stand-alone course at their medical schools, and 54% (60/112) indicated that it was integrated into other courses. Most medical genetics courses (88%, 99/112) were team taught in either the first (77%, 86/112) or second (31%, 35/112) year of medical school. These courses were of moderate duration, with nearly two thirds of the respondents (62%, 69/112) reporting 20 to 40 contact hours. However, almost one fifth (20/112) of the respondents taught courses with fewer than 20 contact hours. Clinical departments sponsored about half (55/112) of the courses taught by the respondents, with the majority of these (46/55) being sponsored by some type of genetics department or division. Twenty-nine percent (32/112) of courses were affiliated with basic science departments, and 17% (19/112) were multidisciplinary.

To determine how many of the lecturers had formal training in medical genetics, respondents were asked whether they or any of the course lecturers were certified by the American Board of Medical Genetics (ABMG) or the Canadian College of Medical Genetics. As shown in Table 2, approximately three fourths (83/112) of the courses were taught by or had lecturers who were certified medical geneticists. Nearly all (98%, 110/112) of the respondents used instructor-led lectures to convey relevant information. Case studies and group discussions were likewise heavily employed. Over one fourth (31/112) of the respondents used the Internet for teaching. The most frequently mentioned "other" instructional formats were patient presentations/clinical correlates, student presentations, and problem sets. Most of the respondents (86%, 96/112) taught courses whose primary objective was to

Table 1

General Characteristics of Courses in Medical Genetics Taught in U.S. and Canadian Medical Schools, 2004 to 2005

Characteristics	No. (%) respondents
Type of course	
Stand-alone	52/112 (46)
Integrated	60/112 (54)
Course taught with multiple instructors	
Yes	99/112 (88)
No	12/112 (11)
Unspecified	1/112 (1)
Year of curriculum in which course was taught*	
First	86/112 (77)
Second	35/112 (31)
Third	6/112 (5)
Fourth	1/112 (1)
Unspecified	0/112 (0)
Total hours taught in course	
<20	20/112 (18)
20–40	69/112 (62)
41–60	15/112 (13)
>60	5/112 (4)
Unspecified	3/112 (3)
Type of sponsoring unit	
Clinical sciences	55/112 (49)
Basic sciences	32/112 (29)
Multidisciplinary/integrated	19/112 (17)
Other/unspecified	6/112 (5)

* Column total exceeds 100% because some respondents reported teaching medical genetics in more than one year.

Table 2

Instructional Characteristics of Courses in Medical Genetics Taught in U.S. and Canadian Medical Schools, 2004 to 2005

Characteristics	No. (%) respondents
Course taught by an ABMG- or CCMG-certified instructor*	
Yes	83/112 (74)
No	26/112 (23)
Unspecified	3/112 (3)
Instructional formats used†	
Instructor lectures	110/112 (98)
Guest seminars	19/112 (17)
Group discussions	58/112 (52)
Case studies	83/112 (74)
Clerkships	6/112 (5)
Web based	31/112 (28)
Other	26/112 (23)
Principal course objective‡	
Provide a broad survey of medical genetics concepts	96/112 (86)
Critical evaluation of the scientific literature regarding basic human genetics	1/112 (1)
Practical training in the use of medical genetics in general practice	12/112 (11)
Other	8/112 (7)
Medical genetics incorporated into third- and fourth-year clinical teaching	
Yes	53/112 (47)
No	17/112 (15)
Unspecified	42/112 (38)

* ABMG, American Board of Medical Genetics; CCMG, Canadian College of Medical Genetics.
† Column total exceeds 100% because most respondents used more than one format.
‡ Column total exceeds 100% because some respondents indicated more than one main objective.

provide students with a broad survey of medical genetic concepts. Many of the "other" course objectives cited by the respondents emphasized a broad overview perspective. Examples are "integration of role of genetics in health and disease with other first- and second-year courses," "basic overview of medical genetics as part of a framework of biochemistry and cell biology," and "explore the activity of genes in development and disease." Eleven percent (12/112) of respondents offered practical training in the use of medical genetics in general practice. One of the respondents considered critical evaluation of the scientific literature regarding basic human genetics to be a principal course objective.

In response to the question, "Is medical genetics integrated into clinical teaching at your institution?" 47% (53/112) of the respondents indicated that medical genetics was currently being taught in the third and fourth years of medical school (Table 2). The remaining 53% (59/112) of respondents indicated that medical genetics was not being taught in the third and fourth years (17/59), did not know whether it was taught in the clinical years (27/59), or declined to answer (15/59), suggesting lack of knowledge about the placement of genetics in the clinical curriculum. In those instances where medical genetics was taught in the clinical years, the venues for instruction were as follows: pediatrics clerkship (51%, 27/53), obstetrics–gynecology clerkship (11%, 6/53), medicine clerkship (11%, 6/53), elective rotations (11%, 6/53), family medicine clerkship (8%, 4/53), and other clinical experiences (8%, 4/53).

Respondents were asked to indicate what topics in genetics were covered in their curricula (Table 3). Of the responses (92%, 103/112), the topics most commonly taught were cancer genetics (94%, 97/103), multifactorial inheritance (91%, 94/103), Mendelian disorders (90%, 93/103), clinical cytogenetics (89%, 92/103), and patterns of inheritance (87%, 90/103). Fewer than 30% (30/103) of respondents discussed immunogenetics, evolution, eugenics, or creationism in their programs. The instructional time devoted to a given topic ranged from 0.45 to 4.1 hours, with a mean of 1.5 hours per topic. Biochemical genetics, development, and Mendelian disorders received the most attention, with an average of 3.5 contact hours, whereas eugenics, uniparental disomy, and stem cells received an average of only 0.6 contact hours.

Discussion

To our knowledge, there have been no published reports looking at the status of genetics education in U.S. and Canadian medical schools. This is the first systematic assessment of how and what medical students are taught about medical genetics. Our findings show that approximately half of the responding schools have medical genetics as a stand-alone course and half have it integrated into other courses. Most courses are taught in the first two years of medical school, using instructor-led lectures, case studies, and group discussions as the predominant instructional formats. Half of the courses are sponsored by clinical departments or divisions, with the remaining being multidisciplinary or sponsored by basic science departments. Most courses present a broad survey of medical genetic concepts, with very few offering training in the use of medical genetics in general practice. When asked whether medical genetics was integrated into clinical teaching, fewer participants responded. Therefore, we can state only that at least half of the schools have integrated genetics into clinical teaching and the rest either do not teach it in the clinical years or the respondents lacked relevant information. In those instances where medical genetics was taught in the clinical years, most of the instruction occurred in the pediatrics clerkship.

To determine the relative importance of medical genetics topics in the curriculum, respondents were asked to indicate what topics were taught at their institutions and how much contact time was spent on each. Not surprisingly, Mendelian disorders, patterns of inheritance, and clinical cytogenetics were among the most commonly taught subjects, as well as cancer genetics and multifactorial inheritance. These subjects had

Table 3
Genetics Topics Taught in U.S. and Canadian Medical Schools, 2004 to 2005

Topic	Number of instructional hours	No. (%) of respondents teaching topic (n = 103)
Biochemical genetics	4.10	81 (78.6)
Development	3.19	57 (55.3)
Mendelian disorders	3.10	93 (90.3)
Cancer genetics	2.60	97 (94.2)
Clinical cytogenetics	2.40	92 (89.3)
Molecular pathogenesis	2.20	57 (55.3)
Molecular genetic techniques	1.90	80 (77.7)
Ethical issues	1.75	72 (69.9)
Patterns of inheritance	1.70	90 (87.4)
Genetic testing	1.70	87 (84.5)
Genetic counseling	1.50	85 (82.5)
Multifactorial inheritance	1.40	94 (91.3)
Dysmorphology	1.33	70 (67.9)
Gene therapy	1.28	78 (75.7)
Pharmacogenetics	1.28	59 (57.3)
Prenatal diagnosis	1.25	82 (79.6)
Immunogenetics	1.23	28 (27.2)
Linkage analysis	1.20	77 (74.7)
Population genetics	1.19	84 (81.6)
Cancer cytogenetics	1.07	72 (69.9)
Cloning	1.02	55 (53.4)
Trinucleotide repeat diseases	0.99	83 (80.6)
Evolution	0.93	20 (19.4)
Mitochondrial genetics	0.90	83 (80.6)
Teratogens	0.90	58 (56.3)
Imprinting	0.83	80 (77.7)
Creationism	0.75	2 (1.9)
Stem cells	0.74	46 (44.7)
Uniparental disomy	0.66	77 (74.8)
Eugenics	0.45	18 (17.5)

substantial contact time, averaging more than two hours each. The least commonly taught subjects were immunogenetics, evolution, eugenics, and creationism, with fewer than a third of respondents teaching these subjects. These subjects, along with uniparental disomy and stem cells, received the least amount of contact time, averaging less that 1 hour each. Although it is perhaps not surprising that evolution, eugenics, and creationism are not major topics in medical genetics courses, it is surprising how little attention is apparently being devoted to the topics of immunogenetics, uniparental disomy, and stem cells, all of which promise to have a significant impact on clinical medicine in the near future. This deficiency may be attributable to faculty's unfamiliarity with the subject, reservations in discussing controversial topics such as stem cells, the topics being taught in other courses, or simply a lack of time in the curriculum.

Why is this important?

Although genetics is becoming more integrated into all areas of medicine, the number of physicians choosing to practice the specialty of medical genetics is declining. There are fewer than 3,300 genetics professionals in the United States who are certified by the ABMG and/or the American Board of Genetic Counseling,[2] and they are unable to meet even the current demand for genetic services. Therefore, primary care providers will be increasingly relied on for genetic counseling and risk assessment.

A recent survey of family physicians asked how many times in the past year they had discussed genetic information with their patients.[3] Every respondent reported having addressed at least one condition from a genetics perspective in the last year, and the genetics of common cancers, cardiovascular disease, and Alzheimer disease had been discussed with two or more patients in the past year.[3] Genetics is becoming increasingly relevant to primary care physicians as the genetics of common disorders are better understood. Genetic risk assessment and testing are now standard for some forms of cancer, and genetic testing is available for a number of neurological and cardiovascular disorders. Other examples include testing for factor V Leiden, which is associated with an increased risk of venous thromboembolism. Pharmacogenetic testing is becoming routine in the treatment of some types of leukemia and will undoubtedly become widely used in routine medical decisions.

All of this serves to underscore the growing relevance of medical genetics to primary care, and yet the limited genetics knowledge of most primary care providers is well documented.[4] This lack of proficiency is attributable both to a rapid advancement in genetics knowledge and to limitations in genetics education in medical school curricula.[5] Recent surveys indicate poor skills in evaluating patient family history for the possibility of a genetics condition,[6–9] missed opportunities for genetic diagnoses,[7,10,11] and few referrals to genetic counseling services.[12]

Comparison with AAMC recommendations

A comparison between our findings and the AAMC's report "Contemporary Issues in Medicine: Genetics Education"[1] revealed areas of strength and weakness in the current status of genetics education in U.S. and Canadian medical schools. The AAMC's recommendations are divided into four main areas: attitudes, knowledge, skills, and educational strategies. Our survey did not directly address students' attitudes, but on the basis of the contact hours devoted to genetic counseling, it seems likely that most schools discuss the potential

psychological impact of genetic diagnoses on patients. Whether these schools also delve into privacy issues or the need to reduce public fear and misinformation about genetics is unknown. All schools seem to adequately cover the knowledge of genetic transmission, molecular biology, and population genetics, but only a small number have begun to integrate genetics into clinical training outside the pediatrics clerkship. In the area of skills, we can deduce from the contact hours devoted to teaching patterns of inheritance that many schools teach students how to take a multigenerational family history, determine the pattern inheritance, and evaluate risks associated for individual family members. Most schools also seem to instruct students on how to interpret genetic test results and deliver this information to the patient. However, from our findings, it is unclear whether students are taught the need for informed consent, how to obtain it, and how to access and evaluate scientific genetic literature.

Finally, in terms of educational strategies, the AAMC's report strongly encourages the use of standardized patients, case presentations, and integration of basic science and clinical medicine in genetics education. Most medical schools use case presentations in their curricula, but none of the respondents mentioned the use of standardized patients for genetics education, and few discussed the integration of basic science and clinical medicine. The report also recommends using common illustrations, developing "a culture where genetics is 'seen' on wards and in the clinics," and asking students to consider the question, "Why is this condition in this person being evidenced now?" None of the respondents described using these clinical applications; however, it is possible that many of the respondents were simply unaware of how genetics was taught outside the basic science years. In addition, the report describes several evaluation modalities including the use of online portfolios and a trackable curriculum map of genetics integration. Our survey did not address the issue of evaluations; however, the online portfolio is in use in at least one medical school.[13]

Our findings shed light on the current state of medical genetics education in U.S. and Canadian medical schools and suggest areas for improvement to ensure that all students achieve the core competencies in genetics and are prepared for its integration into their day-to-day medical practice. One apparent area of weakness is the teaching of medical genetics in the third and fourth years; however, this finding may be open to interpretation because several respondents indicated they did not know the status of genetics in the clinical years or chose not to respond. Further study is needed to better characterize how medical genetics is being incorporated into students' clinical training and how best to correct any deficiencies. The AAMC's report provides the appropriate guidance, and medical schools should strive to adopt as many of its recommendations as possible. Medical genetics will continue to exert a growing influence on the practice of medicine, and future physicians will need better training if they are to provide optimal health care for their patients.

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