Retention of medical genetics knowledge and skills by medical students

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Purpose: To determine whether specific knowledge and skills medical students acquire after completing a Year 1 genetics course are retained at the end of Year 3. **Methods:** A genetics case was developed for an observed structured clinical exam at the end of Year 3. The case involved a pregnant patient who underwent population screening for cystic fibrosis and is identified as a carrier of a common mutation. Student's performance in completing eight essential genetic tasks taught in Year 1 was assessed by their ability to apply these concepts in the Year 3 observed structured clinical exam. **Results:** A total of 212 students were included in the study. Performance on the essential tasks revealed that students were better able to discuss inheritance pattern (73.1%). Students were less likely to calculate and discuss fetal risk (25%), discuss the option of prenatal diagnosis if the father is a carrier (25%), and ask about a family history of cystic fibrosis (36.8%). Only half (50%) explained the test result and implications to the patient. There was no correlation between individual student exam scores in Year 1 and the eight essential genetics tasks scores assessed in the observed structured clinical exam ($r = 0.003$, $P \leq 0.67$). **Conclusion:** Third year medical students do not retain medical genetics knowledge and skills learned in the first year of medical school. Medical schools need to integrate genetics curriculum through the continuum of the 4 years of medical school. ***Genet Med*** **2009:11(5):365–370.**

Key Words: *medical genetics, retention, medical students, knowledge and skills, observed structured clinical exam*

The practice of medicine is changing as clinical applications based on genetic technologies continue to emerge as a result of the Human Genome Project. Physicians are addressing patients' questions about familial diseases, assessing the appropriateness of genetic testing, facilitating informed decision making, and promoting preventative health measures. This personalized approach to patient care allows physicians to focus on prevention and earlier diagnosis. Targeted therapies are beginning to provide a more effective and efficient method to treatment. For genomic medicine to reach its full potential, health care providers need a sound genetics knowledge base and practical skills to clinically apply this knowledge in a competent and responsible way.

Medical schools are responsible for teaching medical genetics to each new generation of physicians. In 2004, the Association of American Medical Colleges (AAMC) issued a report, *Contemporary Issues in Medicine: Genetics Education,* delineating the learning objectives related to medical genetics that students should acquire before graduation from medical school.[1] In addition to a knowledge base in fundamental genetic principles, the report lists specific skills and attitudes that students need to responsibly utilize genetic information and technologies in clinical practice. Educational strategies are also suggested, which include the use of standardized patients, case presentations, and the integration of basic science and clinical medicine in genetics education.

A recent study provided baseline information on the genetics curriculum in 112 US and Canadian accredited medical schools.[2] Information collected included material covered, number of contact hours, year in which the course was offered, and what department sponsored the course. The study found that half of the schools have a standalone genetic course in the preclinical years. Eighty percent of schools have 40 or fewer contact hours. Topics commonly taught were cancer genetics, multifactorial inheritance, Mendelian disorders, cytogenetics, and patterns of inheritance. Forty-seven percent of schools reported some medical genetics instruction in the third and fourth years, typically a lecture during the pediatric or obstetrics/gynecology (OB/GYN) rotation. In comparison with the AAMCs' genetics objectives, the authors conclude that schools cover a selection of genetic principles but do not teach students how to apply them in clinical situations.

Numerous studies have explored practicing physicians' knowledge and practices regarding medical genetics and their confidence in addressing patients' questions. A systematic review of 68 peer-reviewed articles pertaining to the delivery of genetic health care services for common adult-onset conditions was recently published.[3] The review focused on outcomes and delivery of genomic medicine, barriers to genomic medicine as well as consumer information needs. The most consistent finding was that the primary care workforce is unprepared to integrate genetics into the regular practice of medicine. The authors conclude that remediation of this deficiency should be a top priority and that more studies are needed to determine how this can best be accomplished.

Although deficiencies in physicians' genetic knowledge have been documented, it is unclear where in the different stages of medical education the gaps exist. How effective is the current approach in undergraduate medical education? Should the genetics curriculum be expanded? Should more efforts be focused at the level of graduate or continuing medical education? These questions should be assessed to best utilize resources to develop targeted genetics educational tools and programs. The purpose of this study was to evaluate the effectiveness of the genetics curriculum at Wayne State University School of Medicine (WSU SOM) by determining whether specific knowledge and skills medical students acquire after completion of a first year medical genetics course are retained at the end of the third year of medical school.

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MATERIALS AND METHODS

Curriculum overview

The genetics curriculum at WSU SOM follows current standards for medical genetics training established by the AAMC and the Association of Professors of Human and Medical Genetics.[4] The curriculum consists of a standalone course at the end of the first year of medical school, which covers basic genetic principles with applications to various clinical specialties. Table 1 provides a course overview, demonstrating how the course builds on different cognitive levels and introduces issues to develop student attitudes. The approach to learning in genetics is to provide students with knowledge, skills, and attitudes so they can apply genetic principles to clinical problems involving a wide variety of diseases and family situations. This approach includes an analysis of clinical features and disease expression, etiology, pathophysiology, molecular genetics, population genetics and risk assessment, approach to genetics testing and test interpretation, public health genomics and population screening, prenatal diagnosis, and ethical and psychosocial issues.

The 5-week course contains 30 contact hours and is a requirement for all medical students. It is mostly lecture based but does include five small group discussion sessions (three problem solving and two case studies) and four patient panels. A documentary about families' stories is shown the first day of class to expose students to the ethical dilemmas and privacy issues uniquely raised by genetic information. The lecture and small group faculty consists almost entirely of clinical genetics professionals. Student competency in medical genetics is assessed on passing a 75-question examination at the end of the course. Examination questions are in United States Medical Licensing Examination format and measure specific learning objectives. Student pass rate on the exam is determined by using the 99.6% confidence interval around the mean.

The curriculum content that is being assessed for student retention in this study relates to cystic fibrosis (CF) (Table 2). CF is a common genetic condition and used throughout the course to illustrate many genetic principles and clinical situations. Students' understanding of the issues specific to CF is accomplished through lectures, small group discussion sessions, a documentary, and a patient panel. The documentary is about a family who discovers through population screening and subsequent prenatal diagnosis that their twin daughters both have CF. The story deals with the twins' medical problems and decisions that the family makes during two subsequent pregnancies. A man in his 20s with CF is a member of one of the course's four patient panels. He discusses his initial diagnosis, medical problems, and recent lung transplant. He shares with the students the relationship challenges he faces as someone with a chronic and life-threatening condition.

Students work together during two case studies with a faculty facilitator through modeling and student role plays to obtain hands on experience applying genetic principles to specific clinical situations. The first case study is a fragile X case and the second a Duchenne muscular dystrophy case. The importance of diagnostic information, collection and interpretation of family history information, agenda setting to address patients' questions, risk assessment, explaining inheritance patterns, interpreting molecular tests, discussing options of genetic testing, and prenatal diagnosis are all discussed and practiced during these sessions. The students are engaged in a discussion about the

Table 1 First year medical genetics course overview — 5 weeks

General content and learning activity	Cognitive level	Specific content areas
Set the stage	Attitudes	Eugenics, history, genetics in the context of values, beliefs, and society
Genetic principles Clinical features Technology	Knowledge	Mitosis and meiosis, chromosome structure and syndromes, cytogenetic techniques
Problem set Family perspective	Application attitudes	Cytogenetics problem set Down syndrome patient panel
Genetic principles Clinical features Technology resources	Knowledge	Single gene disorders, inheritance patterns, population genetics, multifactorial inheritance, imprinting, uniparental disomy, mitochondrial inheritance, molecular genetic diagnosis, self-study on resources
Two problem sets Family perspective	Application attitudes	Risk assessment and "other" problem sets neurogenetics patient panel
Clinical genetic settings	Knowledge Application skills	Metabolic conditions, clinical genetics, prenatal diagnosis, cancer genetics, genetic family history
Family perspective	Attitudes	Metabolic patient panel
Two case studies	Problem solving skills Counseling Attitudes	Fragile X Duchenne muscular dystrophy
Population medicine	Application	Public health genomics
Family perspective	Attitudes	Final patient panel

Table 2 Application of genetic principles to clinical problems involving cystic fibrosis (CF)

Genetic principle	Learning objectives
Clinical features and disease expression	Describe pancreatic insufficiency and pulmonary obstruction as main clinical features of CF Explain the average life expectancy of individuals with CF is 25 Describe the variability in CF clinical features, including isolated male infertility
Etiology	Identify CF as inherited in an autosomal recessive manner Discuss the characteristics of autosomal recessive inheritance
Pathophysiology	Describe the CF gene as one that transports chloride across the cell membrane Discuss that mutations in CF gene result in abnormal chloride ion transport
Molecular genetics	Appreciate that more than 1000 CF causing mutations have been identified Distinguish that the most common CF mutation is [Delta]F508 Be aware that some genotype/phenotype correlations exist
Population genetics and risk assessment	Differentiate that CF is more common in Northern European ancestries, but occurs in all ethnic groups Perform a risk assessment using probability if there is a family history of CF Perform a risk assessment using Hardy-Weinberg if there is not a family history of CF
Approach to genetic testing and test interpretation	Order a genetic test because of a family history of CF by identifying mutation in family and instructing laboratory to look for that mutation in at risk family members Order a genetic test because of CF carrier screening by instructing laboratory to test for common mutations recommended by national groups
Public health genomics and population screening	Recognize that offering CF carrier screening to all couples planning a pregnancy or seeking prenatal care is standard of care
Prenatal diagnosis	Describe two prenatal diagnostic techniques available to test fetus for CF including gestational timing, accuracy, and risks and benefits
Ethical and psychosocial issues	Realize families make decisions about prenatal testing based on their own personal values and beliefs Understand physicians' role is to facilitate informed decision making and support their patients' decisions

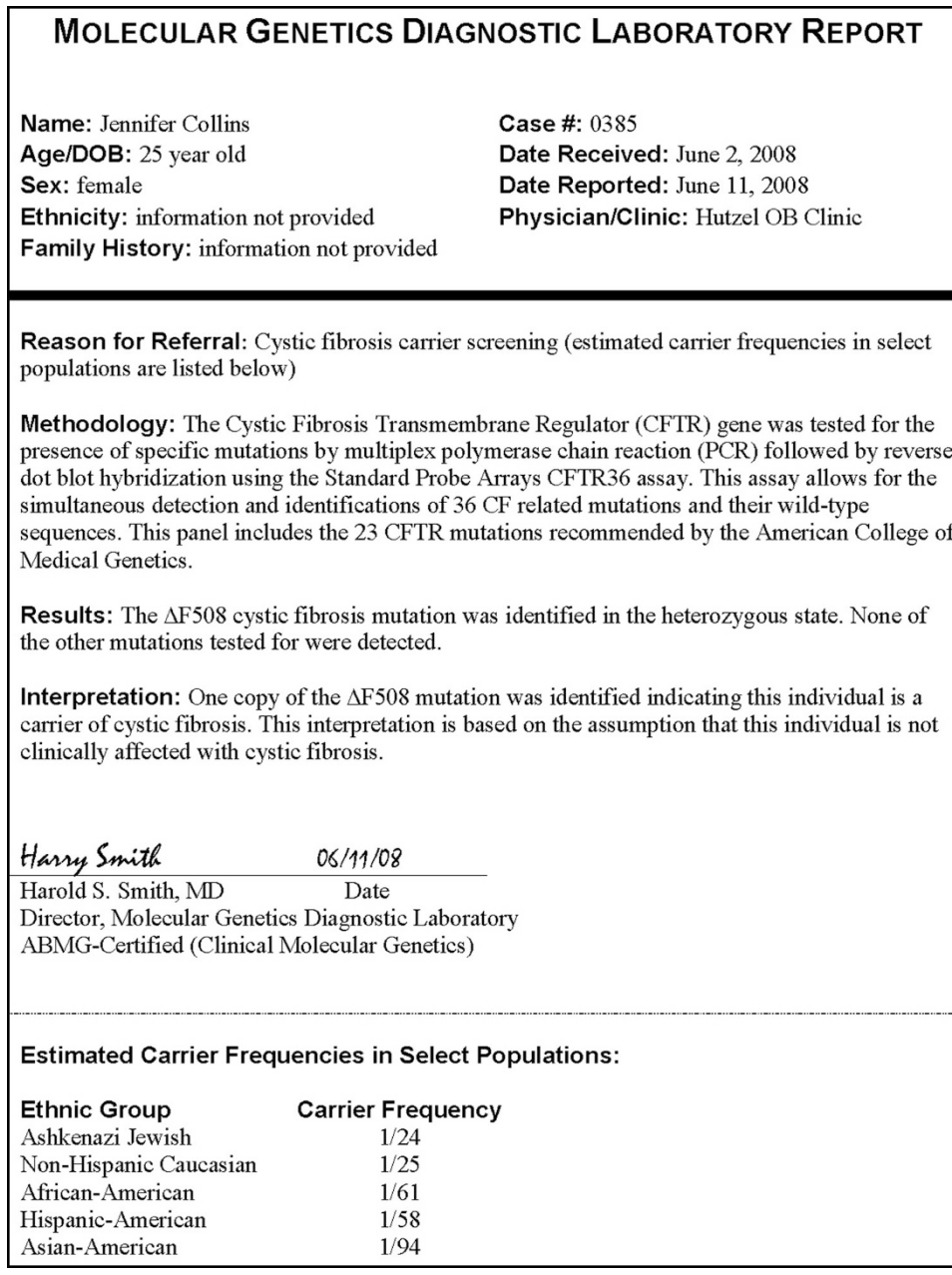
potential psychosocial impact of genetic information on patients, their family, and their community.

Observed structured clinical exam

All medical students at WSU SOM are required to take an observed structured clinical examination (OSCE) after the successful completion of the third year of medical school. The OSCE is a clinical skills competency examination consisting of 10 structured clinical encounters and two note documentation stations. Cases are developed using a standard protocol and standardized patients are trained to portray the clinical scenarios in a consistent and structured manner. Stations are designed to test basic knowledge of history taking, physical examination, communication skills, and data synthesis. Students are told that there are 10 patient encounter stations with standardized patients and two note documentation stations. Students are not given information about the nature of the encounters or which specialty areas they represent. A checklist specific to the encounter is used to evaluate student performance. Students are evaluated and given scores in the areas of history taking, communication, physical exam, and note documentation. Students falling two standard deviations below the mean fail the OSCE and undergo remediation to correct their deficiencies.

A genetics case was developed for the OB/GYN station to prospectively measure student retention of medical genetics knowledge and skills from the first year of medical school. The case was structured in a way to assess knowledge and skills identified as important medical genetics learning objectives by national bodies and coincided with learning objectives established for the first year medical genetics course at WSU SOM.[1,4] A CF case in an OB/GYN setting was selected because offering CF screening to all reproductive patients has been standard of care since 2001. The clinical scenario involved a pregnant patient who underwent population screening for CF and is identified as a carrier of a common mutation. The students were asked to interpret the test result and counsel the patient appropriately. Standardized patients were trained to play the role of the patient and rate student performance using an evaluation tool. Two standardized patients were involved in each student OSCE session and alternated between functioning as the patient and the rater. Students were given 2 minutes before the session to review the patient problem, laboratory report, and the station's instructions (Fig. 1). The patient's ethnicity and family history information was not provided. If the students asked for this information during the session they were told the patient's ethnic background was German and English, the father of the baby's ethnicity was African American, and that there was not a family history of CF. The report also contains information about CF carrier frequencies in different ethnic groups, including the African American population. Students were given 8 minutes to complete the patient encounter.

Given the foundation in medical genetics in the first year medical genetics course, the authors felt it was reasonable to ask students to address a common clinical genetics problem during the OSCE. Additionally, all students attend a 1-hour didactic session on reproductive genetics during their OB/GYN clerkship. Students were expected to ask questions related to routine



MOLECULAR GENETICS DIAGNOSTIC LABORATORY REPORT

Name: Jennifer Collins
Age/DOB: 25 year old
Sex: female
Ethnicity: information not provided
Family History: information not provided

Case #: 0385
Date Received: June 2, 2008
Date Reported: June 11, 2008
Physician/Clinic: Hutzel OB Clinic

Reason for Referral: Cystic fibrosis carrier screening (estimated carrier frequencies in select populations are listed below)

Methodology: The Cystic Fibrosis Transmembrane Regulator (CFTR) gene was tested for the presence of specific mutations by multiplex polymerase chain reaction (PCR) followed by reverse dot blot hybridization using the Standard Probe Arrays CFTR36 assay. This assay allows for the simultaneous detection and identifications of 36 CF related mutations and their wild-type sequences. This panel includes the 23 CFTR mutations recommended by the American College of Medical Genetics.

Results: The ΔF508 cystic fibrosis mutation was identified in the heterozygous state. None of the other mutations tested for were detected.

Interpretation: One copy of the ΔF508 mutation was identified indicating this individual is a carrier of cystic fibrosis. This interpretation is based on the assumption that this individual is not clinically affected with cystic fibrosis.

Harry Smith 06/11/08
Harold S. Smith, MD Date
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Estimated Carrier Frequencies in Select Populations:

Ethnic Group	Carrier Frequency
Ashkenazi Jewish	1/24
Non-Hispanic Caucasian	1/25
African-American	1/61
Hispanic-American	1/58
Asian-American	1/94

Fig. 1. Genetics OSCE patient laboratory report.

prenatal care, and use proper interviewing techniques and attending skills. In terms of the genetic issues, students were expected to perform the following essential tasks to be considered minimally competent in addressing the clinical problem: (1) ask about the ethnicity of the father of the baby, (2) ask whether there is a family history of CF, (3) explain the test results to the patient, (4) describe the main clinical features of CF, (5) explain how CF is inherited, (6) determine and explain the risk of CF to the fetus, (7) explain the next step is to test the father of the baby, and (8) explain the next step if the baby's father is a carrier. The first two tasks are in the area of history taking, and the last six are in the area of counseling.

Analysis

A dichotomas scale was used to assess students performance on the OSCE. The student was given a score of 1 for performing the task and a score of 0 for failure to perform the task. Individual student performance on the OSCE was compared to his/her performance on the first year medical genetics course examination. Descriptive statistics were used and are presented as percentages, means, and standard deviation. A Pearson's correlation was conducted to determine whether there was a relationship between students' scores on the Year 1 genetics exam and their performance on the Year 3 OSCE. A $P \leq 0.05$ was used to determine statistical significance.

RESULTS

The study participants included all third year medical students who matriculated in 2005, and took the medical genetics course during their first year of medical school and the OSCE at the end of Year 3. A total of 259 students took the OSCE over a 2-day period in June 2008. These students had also completed the core obstetrics and gynecology clerkship. Two students declined to participate in the study and three students were transfers who did not take the genetics course. Forty-two students were not on a full-time academic track for various reasons and did not take the genetics course in 2006. These 47 students were excluded from the study. A total of 212 students met the inclusion criteria and participated in the study.

The Year 1 genetics exam had a total of 75 items. The average score for the group was 81%. A total of five (2.4%) students failed the exam. The OSCE station had a total of 37 items. Of these there were eight essential genetics tasks (Table 3). Mastery of these tasks was determined by a score of 75% or higher. The average score for completing these tasks was 46.9%. A total of 186 (88%) students failed the genetics section of the OSCE exam.

Student performance on each of the eight essential tasks was also analyzed and is shown in Table 3. Fifty-three (25%) students accurately calculated the risk of CF in the fetus. Of these, 52 (98%) also asked about the father of the baby's ethnic background, 12 (23%) asked about a family history of CF, and 11 (21%) asked about both ethnicity and family history. One student was given credit for calculating the risk but was not given credit for asking about ethnicity. Fifty-two (24.5%) students asked about the father's ethnic background but did not calculate the correct risk of CF in the fetus.

The highest student scores $n = 155$ (73.1%) were related to discussing the inheritance pattern and that the next step was to test the father of the baby 133 (62.7%). Only half of the students explained the test result to the patient. Student performance was poorest in determining and explaining the risk of CF in the fetus (25%), discussing options of prenatal diagnosis if the father is found to be a carrier (25%), and asking whether there was a family history of CF (36.8%).

Individual student genetic exam scores were correlated with the eight essential genetics OSCE scores. The Pearson's correlation (r) was 0.003 with a $P \leq 0.67$ showing no significant relationship between the student's percent score on the genetics exam and their percent score on the OSCE.

Table 3 Student performance on the eight essential genetics OSCE tasks

Task	Number correct	Percent correct (%)
1. Asks the ethnicity of the father of the baby	104	49.1
2. Asks whether there is a family history of cystic fibrosis	78	36.8
3. Explains the test results to the patient	106	50.0
4. Describes the main clinical features of cystic fibrosis	115	54.2
5. Explains how cystic fibrosis is inherited	155	73.1
6. Determines and explains the risk of cystic fibrosis to the fetus using the carrier frequency in the African American population	53	25.0
7. Explains the next step to refine the risk is to test the father of the baby	133	62.7
8. Explains next step if the baby's father is a carrier	53	25.0
Total	797	46.9

DISCUSSION

Research generated by the Human Genome Project has already impacted the practice of medicine. The ability to identify the genetic basis of disease sheds light on disease etiology and pathophysiology, and it gives clinicians more targeted diagnostic and prognostic tools. High-risk individuals can be identified and tailored surveillance and treatment plans developed. For this approach to be effective, primary care providers will need to make initial assessments about patients' risks. Risk assessments involve the ability to gather family history information and construct a pedigree documenting not just medical information but family relationships and information such as ethnicity and consanguinity.

Physicians also need a "genetic skill set" to assess the utility of genetic testing for genetic disorders when a diagnosis is either suspected or known. A recent study conducted to assess OB/GYN familiarity, knowledge, and application of practice guidelines associated with carrier screening for CF, found that only 67% of obstetricians/gynecologists and 42% of gynecologists had either read or skimmed the guidelines.[5] They established that correctly responding to basic questions regarding CF was associated with having read the guidelines, although responding to questions about a more complex, but common clinical CF scenario was not illustrating deficiencies in physician understanding about genetic test sensitivity.

How well are our medical schools preparing future physicians to address genetic issues and properly utilize genetic technologies? Learning objectives have been established at the undergraduate level, but has this translated into improved student learning in genetics? Based on our literature review there are limited published reports looking at the retention of medical genetics knowledge and skills from the basic science into the clinical years of medical education.[6]

The goal of the current study was to evaluate whether students can apply basic genetic concepts to a common and routine clinical problem 2 years after the completion of a medical genetics course. Unlike other studies that have used comparisons of examination scores as measures of student retention,[7–11] this study sought to measure retention by evaluating student ability to utilize previously obtained knowledge and skills in a structured clinical manner, which is more representative of future practice. Our study found that the class average went from 81% on the first year genetics exam to 47% on the essential genetics tasks from the third year OSCE, showing a substantial loss of knowledge and skills during the 2-year time period. If the essential tasks were considered those that a minimally competent student should be able to perform, only one student who took the OSCE successfully completed the patient encounter.

Students performed the poorest in the area of risk assessment, anticipating the next step if the father is found to be a carrier, and asking whether there is a family history of CF. The poor performance in the first two of these three tasks may be explained because they require a higher level of cognitive ability. To correctly ascertain the fetal risk, the student needed to ascertain the father's ethnicity and then factor this information

into the risk calculation. Discussing the availability of prenatal diagnosis if the father is found to be a carrier requires the student to recognize the different outcomes and anticipate a course of action. It is unclear, however, why students performed so poorly in asking about a family history of CF. CF is an inherited disorder and family history information is important to assess risks and understand the patient's experiences with the disorder. Both family history information and ethnicity were information fields on the laboratory report and both fields indicated that this information was missing, serving as a trigger for students to ask the question. It is concerning that only half of the students explained the test results to the patient given that the test interpretation was on the laboratory report, and the student instructions were to interpret the report and counsel the patient appropriately.

Lastly, our analysis showed that there was no correlation of individual student performance on the genetics exam and essential OSCE tasks. Even those students who demonstrated mastery of the material after the first year medical genetics course were unable to show competence 2 years later using their previously obtained knowledge and skills.

From a methodological standpoint, there are limitations to this study. Utilizing the written examination and a clinical competency exam are appropriate for measuring and tracking performance at a didactic and at a clinical point in training, respectively. However, directly comparing outcome data from these two assessment methods has limitations, yet does provide some educational insight into students' ability to retain knowledge and apply the genetics skills learned during the Year 1 genetics course. The true goal of training medical students to be physicians is to challenge them to integrate knowledge and then provide it to patients.

We were not able to determine at which cognitive level the deficits exist. Is knowledge retained, but students were not able to analyze and apply the knowledge? Finding the answer to this question will be important in developing better educational strategies in medical genetics and in various other fields. Also, unknown is how much modeling in genomic medicine do students observe by residents and faculty in clinical settings. Studies that have looked at physician knowledge and practices about medical genetics suggest that medical students are not observing the utilization of genetic information or technologies in their clinical training.

Only half of the medical schools have a standalone genetics course in the preclinical years and the majority of those have $<$40 contact hours according to Thurston et al.[2] study. Formal genetics training is virtually nonexistent in the clinical years. Our study shows that what little genetics knowledge and skills students obtain early in their medical training is not retained. To prepare future physicians for practice in this era of genomic medicine, medical schools must promptly respond to this deficiency. The AAMC has outlined medical genetics learning objectives and suggests educational strategies to obtain these objectives.[1] It is time for schools to seriously review these objectives and investigate mechanisms to formally incorporate them into their curricula and evaluate the effectiveness of the curriculum.

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