The virtual diagnostic laboratory: A new way of teaching undergraduate medical students about genetic testing

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Purpose: Medical students often perceive genetics as a discipline focused on rare diseases with relevance only to genetics specialists. Because genetic testing has now infiltrated most if not all medical disciplines, we need new teaching approaches to help trainees incorporate emerging genetic testing strategies appropriately into medical practice. With the ever-increasing number of known disease-associated genes, it is also important to shift from a paradigm of memorization to one of critical evaluation and an awareness of available resources. **Methods:** We designed case-based virtual laboratory sessions for first-year medical students at Emory University. These sessions emphasize both rare and common health issues and allow the students to practice applying their fundamental genetics knowledge in the diagnostic setting. **Results:** These sessions proved a valuable approach to presenting the intricacies of diagnostic genetic testing. Students rate the sessions very highly, with 92% of them agreeing or strongly agreeing that the sessions had educational value. The students commented that ours was an effective approach to teaching the material that illustrates well the impact of genetics on patient care. **Conclusions:** The virtual diagnostic laboratory approach is an effective, nonlecture-based method of teaching medical students about genetic testing strategies and their application in the clinical setting. ***Genet Med*** **2011:13(11):973–977.**

Key Words: *curriculum, medical students, genetic testing*

Although the importance of genetics and genomics to modern medicine continues to grow, surveys of medical students and medical professionals in the United States, Canada, and other countries suggest that many of them feel ill prepared to use genetics in their practice.[1–3] Perhaps even more troubling is the perception that genetics is the study of rare disorders and is only relevant to certain specialties.[3] In their 2011 report on genetics education and training,[4] the Secretary's Advisory Committee on Genetics, Health, and Society found that inadequate genetics education limits integration of genetics into clinical care. As part of this report, a 2008 survey of organizations involved in health professional education indicated that to address gaps in genetics education, the clinical relevance of genetics must be demonstrated in educational programs. Groups within the United States and Europe, including the National Coalition for Health Professional Education in Genetics (www.nchpeg.org), the Association of Professors of Human and Medical Genetics (www.aphmg.org), the American Association of Medical Colleges (www.aamc.org), the European Society of Human Genetics,[5] and the National Genetics Education and Development Centre (www.geneticseducation.nhs.uk), have proposed lists of genetics learning objectives and competencies for medical students and other health professionals. Although it is up to each individual medical school to determine how these recommendations should be incorporated into their curricula, the approaches should provide tools for lifelong learning[6,7]; they should also be case-based, focused on common conditions,[3,8] and they should emphasize that genetics is not an isolated discipline in medicine.[6] Particular stress has been placed on the importance of learning objectives related to the appropriate interpretation of genetic test results and their communication to patients and families.[2,5,7–11] Indeed, two studies on the provision of genetic testing for hereditary cancer syndromes suggest that a significant fraction of nongenetic specialists currently practicing medicine are unprepared to discuss these tests with their patients, order the tests appropriately, or interpret the results of these tests accurately.[12,13]

At Emory University School of Medicine, the genetics curriculum was formerly taught in a traditional lecture-based format during a nonintegrated basic science curriculum. This course emphasized Mendelian and non-Mendelian disease and was organized by genetic mechanism and disease classification. In 2007, Emory introduced a new, integrated undergraduate medical curriculum that is competency-based and stresses active learning. Course directors were charged with reducing lecture time, integrating basic and clinical sciences, and downplaying rote memorization. The faculty was also encouraged to incorporate more clinical simulation into their pedagogical approach. In this new curriculum, genetics and genomics is integrated throughout the 4-year curriculum but is anchored by a 2-week module focused on genetics and human evolution that is taught during the first semester. The sessions we describe in this study are presented twice per week during this 2-week introductory module.

Given the opportunity to revamp the full genetics curriculum, we incorporated the core genetics competencies into the goals of the new curriculum and developed several new active learning approaches to teaching this material. Of particular importance in the development of the genetics module was the desire to emphasize genetics and genomics as disciplines that impact all fields of medicine. To that end, we incorporated content related to the genetics of common diseases and focused on scenarios that might be encountered in a medical specialty other than genetics, especially a primary care specialty.

In previous years, we had included sessions designed to teach students about genetic testing techniques; however, we received some negative comments then, such as "I left with little understanding of how the technologies worked, why I should even know how they worked, and which were in use for what." Based

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on this experience, which told us our prior approach was not always effective at teaching medical students the relevance or complexities of genetic testing, and based on the curriculum requirements mentioned earlier, we developed a set of virtual genetics laboratory sessions. As described later, we took a case-based approach to discuss the use of genetic testing in common situations, and to make the sessions realistic, we use deidentified test results and laboratory report formats from our own clinical diagnostic laboratories.

METHODS

Faculty preparation

The virtual laboratory sessions were planned and facilitated by faculty from the Emory University Department of Human Genetics. The session learning objectives for the laboratories are listed in Box 1. The course director worked with an American Board of Medical Genetics (ABMG)-certified individual from the appropriate genetics discipline (cytogenetics, biochemical genetics, or molecular genetics) to develop the narrative for each case and to collect and modify the illustrative materials, deidentified laboratory reports, and genetic counseling letters. The materials were provided to the session facilitators in advance of a training session that was attended by all the facilitators and the course director. Attendance of all facilitators at this session ensured that each understood the intended goals and expected scope of presentation of the exercise. This session allowed the facilitators to discuss complex or ambiguous aspects of the cases and ensured consistency between the groups. Approximately 90 minutes were needed to discuss the materials for all four of the cases. Training sessions have been held each year by the course director for new facilitators, and, when possible, the course director or an experienced facilitator has visited a classroom session led by the new facilitators to provide feedback and guidance.

Cases were designed to have multiple testing options and possible outcomes. Thus, the facilitators were provided with a varied collection of laboratory results for each case. The facilitators discussed the different testing options and came to an agreement on what they felt was the optimal testing strategy for the patient based on techniques that had already been discussed in class. This strategy was then developed into a testing flowchart that could be used to keep the discussion on track during the session (Figs., Supplemental Digital Contents 1 and 2, http://links.lww.com/GIM/A188 and http://links.lww.com/GIM/A189). These flowcharts also helped to organize the set of test results and associated laboratory reports to be used with each case.

We selected case topics to illustrate the relevance of genetic testing to common or representative patient situations; among these was a positive newborn screening result, a child with motor delays, an adult concerned about a family history of colon cancer, and a prenatal diagnosis. A particular genetic discipline was the focus of at least one case, including biochemical genetics, cytogenetics, molecular genetics, and cancer genetics, and this exposed the students to a range of genetic testing methodologies, their usefulness, and their limitations. To instigate discussion, we purposely selected topics with more than one potential testing strategy, so that the pros and cons of each could be compared. When possible, we incorporated guideline statements from professional organizations into the development of the case. The four relevant diagnoses for the cases were Duchenne muscular dystrophy (OMIM# 310200) in a child with motor delay, classic and Duarte galactosemia (OMIM# 230400) in a set of fraternal twins, Lynch syndrome (OMIM# 120435 and 609310) in a family with a history of colon cancer, and mosaic trisomy 20 in a prenatal sample. Cases were developed to touch on a wide range of learning objectives, but they represented realistic scenarios. To protect patient anonymity, the case descriptions were fabricated, rather than derived from patient files, birth dates and genotypes were changed, and reference numbers were fabricated. Cases were organized to allow the facilitators to guide students through a series of topics and alternate testing strategies, several of which are listed in Table 1. To further illustrate our approach, we have provided the materials for the Duchenne muscular dystrophy and the Lynch syndrome cases in the Figures, Supplemental Digital Contents 1–3, http://links.lww.com/GIM/A188, http://links.lww.com/GIM/A189, http://links.lww.com/GIM/A190 and the Appendix, http://links.lww.com/GIM/A203. Further information and the other case materials are available from the authors on request.

BOX 1: Session learning objectives

At the end of these sessions, students should be able to:

1. Identify valid resources for up-to-date information on genetic testing decisions.
2. Propose appropriate genetic testing strategies for simple cases.
3. Interpret reports from a clinical genetics laboratory.
4. Communicate laboratory results to patients and answer basic questions about these results.

Student preparation

Before the virtual laboratory sessions, the students attended lectures that gave them relevant background, including a discussion of the types of genetic variation that are found in humans and an overview of the laboratory techniques used to detect this variation. Students were exposed to the concept of the mutation spectrum of a disease and the idea that knowledge of the mutation spectrum of a particular phenotype is a critical piece of the puzzle when deciding on a genetic testing strategy. Students also attended a workshop, led by genetic counselors, which addressed learning objectives related to collecting family histories, drawing pedigrees, and discussing genetic information with patients and families.

In preparation for each session, the students were given two case studies to research (see Appendix, Supplemental Digital Content 4, http://links.lww.com/GIM/A203 for preparatory materials). In addition to a short description of each patient's clinical features and concerns, the students were given a set of learning issues and resources to guide their preparation. The list of resources included several of the publicly available genetics databases with which we feel students should become familiar, including GeneReviews, OMIM, and Genetics Home Reference. Students were told to use this framework to research each case and to come to the session prepared to discuss the case, including such topics as the information they will need to gather from the patient and family, the types of disorders that are relevant to the case and the mutations that might cause them, and an appropriate testing strategy to reach a diagnosis and/or perform a risk assessment. In the classroom sessions, the students were asked to lead the discussion of each case, while the facilitator provided guidance. In response to student feedback, we expanded the instructions for the laboratory sessions after the first year. We found that providing a fairly structured set of guidelines for the students focused their preparatory research and led to more fruitful discussions in class.

Table 1 Discussion topics for each laboratory case

DMD	Galactosemia	Lynch syndrome	Prenatal testing
Blood sample vs. muscle biopsy	Newborn screening vs. diagnostic testing	Sporadic vs. familial vs. hereditary cancer	Chorionic villus sampling vs. amniocentesis
Protein-based vs. DNA-based test	Biochemical genetic testing vs. molecular genetic testing	Blood sample vs. tumor sample	Interphase FISH vs. karyotype for prenatal testing
Full gene sequencing vs. targeted sequencing	Mutation panel-based testing and its limitations	Protein-based vs. microsatellite instability testing to assess risk	Maternal cell contamination
DNA sequence vs. deletion/duplication analysis for DMD	Duarte galactosemia vs. classic galactosemia	Genetic heterogeneity	The use of published literature for interpretation of genetic test results
Mutation-specific testing in family members		The limitations of DNA sequence-based testing	Tissue-specific mosaicism

FISH, fluorescence in situ hybridization; DMD, Duchenne muscular dystrophy.

In-class discussions

For the virtual laboratory sessions, each group of 15–16 medical students was paired with one facilitator. The class size is approximately 140 students. Eight of nine facilitators for the fall 2009 semester were certified by the ABMG in at least one medical genetics discipline. In 2010, we had the same group of facilitators with the addition of some cofacilitators who were either certified genetic counselors or fellows in our ABMG-laboratory fellowship program. Although we realize this number of ABMG-certified faculty would not be available to all medical schools, the unique perspective and expertise they brought added greatly to the initial design and success of our sessions. For an overview of the stages of these in-class discussions see Box 2. As each group worked through a case, facilitators gave the students additional information about the patient, such as patient and family histories, that would help them refine their differential diagnosis. This information was developed as part of the preparatory materials for the facilitators. After this initial stage, the students presented and justified their suspected diagnoses and explained which testing strategies they would recommend to confirm or refute the diagnosis. This allowed a comparison of different approaches to the same problem. Facilitators were instructed to avoid answering "What would you do?" types of questions until the students had discussed the case from all angles.

Once consensus among the students was achieved and a test agreed on, the facilitator would provide a laboratory result for the requested test. By this, we mean a graphic or tabular representation of assay data. As a group, students interpreted the results of the test and discussed potential limitations to the interpretation. If the students ultimately recommended a suboptimal testing strategy here, the facilitator would use a discussion of the caveats of the chosen test strategy to guide the discussion back to the most appropriate options. For example, for our hereditary colon cancer case, several groups found it beneficial to discuss the inherent difficulties of interpreting a negative genetic testing result when the person tested does not have cancer and the familial mutation is not known. Even when students opted for what was deemed the optimal testing strategy, they and the facilitators also sometimes found it helpful to talk through the alternative strategies and potential results to stimulate further discussion of test limitations and the types of scenarios that would favor use of one test option over another.

To show how test results and interpretation are communicated to the referring physician, the facilitators presented laboratory reports and highlighted the utility of each piece of information on the report. Students then discussed the clinical report interpretation in the context of the in-class discussion. Facilitators stressed the fact that different testing laboratories may use different assay methodologies and that the assay methodology is a critical factor in the interpretation of results and in understanding a test's utility and limitations. Next, the students talked through the implications of each test result for patient diagnosis and management, as well as risk assessment for specific family members. At this point in the class, students were asked to design a testing scheme for other at-risk family members and to explain how the testing strategy and interpretation might differ for these family members compared with the proband. The students were then shown a fictionalized letter from a genetic counselor to the patient explaining their test results and the implications of those results for them and their family members; the point was to make students aware of the need for written communication with patients and to allow them to compare the information and language used in this type of communication with the wording of the actual laboratory report. Students practiced explaining the laboratory report and letter to their "patient" in lay terms. Herein, we emphasized the need to be respectful and nondirective at all times, while providing the key information in a way that would be accessible to the patient and their family.

One goal of the genetics and evolution module at Emory is for students to gain practice incorporating information from primary research articles into medical decision making. For the prenatal

BOX 2: Stages for each case discussion

What information will help us limit the differential?
What testing approaches are available?
Which approach would be best in this case? In what order should the tests be used?
What do these results mean?
What are the caveats to interpreting these results?
What do the results mean for patient management and for risk assessment in the family?
How would we approach testing in at-risk family members?
How do we communicate this information to the patient and his/her family?

testing case, the cytogenetic test result the students received indicated that the fetus was mosaic for trisomy 20. For the students to appreciate how published literature can help guide interpretation of clinical genetic test results, they were assigned a research article that compiled data on karotype-phenotype correlations for particular mosaic trisomies,[14] allowing them to predict with greater confidence the potential outcome for the virtual patient.

We discussed two cases in each 2-hour session. We found that pacing each session proved to be very important for keeping the discussion on target and the students engaged. Although the faculty development that we did in advance of the sessions ensured that there was consistency in terms of the preparation of the faculty, the student-led format of the discussions did mean there was variability between groups. To reduce between-group differences in the material presented, an overview of key learning points from each case was posted electronically for all students after the session (see Figures, Supplemental Digital Contents 1–3, http://links.lww.com/GIM/A188, http://links.lww.com/GIM/A189, http://links.lww.com/GIM/A190 and Appendix, http://links.lww.com/GIM/A203.

Student evaluations

Anonymous student evaluations of each educational session of the Foundations Phase of the Emory undergraduate medical curriculum are managed through the Office of Medical Education and Student Affairs. A randomized sample of approximately 25 students is asked to evaluate each session, and their grades are held until these evaluations are completed. The evaluation is administered electronically through the One45 system (www.One45.com) and includes a series of six statements about each session for which the respondents have to indicate their preference on a Likert scale that ranges from "Strongly Disagree" to "Strongly Agree." In addition, the students are given extra space for freeform comments and suggestions for improvement. In our analysis, we combined the scores for the two laboratory sessions because each had independent evaluators.

RESULTS

The laboratory sessions were very well received, with students finding them "a good way for us to understand the intricacies of what we're learning," "effective for demonstrating how to understand the genetic testing and when and who to test," and "effective at introducing pedigrees and launching really interactive discussion of genetics, family involvement, and the long list of specialties exhausted before patients reach a genetic diagnosis. The preparation that went into the lab made it a great learning experience." The session design was also praised, because "we could take (the discussion) where we wanted to go. We always have pretty good ethics discussions in our group."

Most students responded positively to the virtual laboratory sessions. When asked to respond to the statement: "There was educational value to this session," 20 of 52 (38%) students strongly agreed, and 28 of 52 (54%) agreed, for an overall 92% approval rating. Furthermore, 88% of surveyed students either strongly agreed or agreed with the statement that "the teaching format was suitable for the objectives," whereas 94% strongly agreed or agreed that "the teaching materials were useful and made the presentation more effective." Although we realize that student satisfaction is not the only or ultimate assessment for a teaching strategy, in the past, we have found that medical students often do not appreciate the relevance of genetics to medicine overall, and the student feedback on these laboratory sessions does indicate that our approach has helped us remedy this to some extent.

DISCUSSION

As a result of the Human Genome Project and subsequent genomics initiatives, there has been an explosion in our understanding of the genes that impact human health and disease. Given the quantity of information now available, rather than sticking with older educational formats that stress categorization of genetic diseases and memorization of the genes involved, we felt that it was crucial to switch to a pedagogical approach that conveys a framework for understanding medical genetics and focuses on critical thinking, as others have also argued for.[15] In particular for these sessions, we wanted our students to become familiar with relevant databases to find genetic information[16,17] and to be able to evaluate genetic test results critically.[2,5,7–11] We believe the virtual laboratory approach proved to be an effective way to show the utility of genetics knowledge in real-world applications and to have the students practice using their genetics knowledge in a set of case-based scenarios.

In addition to the short list of learning objectives for these sessions (Box 1), we feel this session design allowed us to address several of the core competencies proposed by the Association of Professors of Human and Medical Genetics (http://www.aphmg.org/pdf/Med%20Competencies.pdf), among them that students should be able to:

> Recognize the indications for a genetics evaluation.
> Take a family history and draw a pedigree.
> Describe the role of somatic and germline mosaicism in assessing recurrence risk.
> Explain the role of genetic testing for diagnostic purposes in the evaluation of a patient and in predictive and presymptomatic testing.
> Differentiate sporadic versus familial versus hereditary cancers.
> Describe the role of genetic testing, including the benefits, limitations, and ethical implications for cancer patients and their unaffected family members.
> Demonstrate knowledge and appropriate use of electronic resources for clinical diagnoses, testing, and understanding of genetic conditions.

We found that the approach we took, in which students had to choose among several ostensibly valid testing approaches, gave the students more insight into the appropriate application of genetic testing strategies and test interpretation. In previous attempts at teaching similar concepts, we gave students a list of genetic testing techniques, one of which was appropriate for each particular type of mutation. The students then had to pick the best technique from the list for a particular type of mutation. This taught them to match techniques with mutation types, but this approach did not emphasize the intricacies of how to interpret genetic test results or the fact that multiple genetic tests might be required for a single patient to rule in or rule out just one genetic disease. In the format we describe herein, there is more of an opportunity to weigh the pros and cons of different approaches and also to discuss how the testing strategy might differ for other members of the same family. By including cases that involve topics such as prenatal testing and presymptomatic testing, students had a chance to separate their personal perception of risk from the need to offer a nondirective presentation of risks.

In our experience, we found group size to be an important factor in the success of the sessions. Too large a group stifles the discussion in class, as we realized after trying the laboratory sessions with group sizes of approximately 30 students to one facilitator. Ideally, we would prefer to reduce each group to 8–10 students, if we can recruit additional qualified facilitators and secure appropriate classrooms.

Overall, we feel this educational strategy has been successful at connecting basic science with clinical science in an approachable way for first-year medical students. Beyond the fact that active learning sessions such as these force the students to practice "thinking genetically," an added value to the sessions is that they open a dialog with the facilitators about the work they do and the types of genetics professionals who make up the extended healthcare team with whom the students will interact as they enter medical practice.

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