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Genetic Testing and Counseling in Metabolic Liver Disease: An Interactive Lecture for Medical Students

Molly A. McPheron, MD*, Hannah J. Craven, MLIS, Jean P. Molleston, MD, Christen K. Dilly, MD, MEHP

*Corresponding Author: mmcphero@iupui.edu

Abstract

Introduction: Medical students have limited opportunities to learn about current genetic testing. This session provided exposure to different types of testing and the complex issues that physicians may encounter when counseling patients on proper testing and interpreting results. **Methods:** We designed a 1-hour interactive lecture for second-year medical students. We presented an overview of the topic, then applied the concepts to specific disorders and cases. Students were asked to answer questions regarding cases using an audience response system, and we used their responses as the basis for our in-class discussion. This session has been held twice, with 25 students attending in 2018 and 31 students in 2019. The session was also recorded so that additional students not in attendance could watch, and was available to 151 students in 2018 and 333 students in 2019. **Results:** Students answered questions via audience response system. There was a range of 47%-100% of students giving the correct answers in 2018, and 55%-93% in 2019. Exam questions covering genetic counseling issues were answered correctly by 66% and 77% of students in 2018, and 70% and 68% of students in 2019. **Discussion:** This session provided an opportunity for medical students to be exposed to some of the complex ethical and psychosocial issues that may arise with genetic testing for liver disease and to consider how to navigate them. Using an audience response system during the lecture made the session more interactive and allowed the teacher to correct errors and teach based on the responses.

Keywords

Genetic Testing, Metabolic Liver Disease, Biochemistry & Cell Biology, Ethics/Bioethics, Genetic Counseling, Gastroenterology, Medical Genetics, Pediatric Gastroenterology, Case-Based Learning

Educational Objectives

By the end of this activity, learners will be able to:

1. Identify ethical and psychosocial issues that may arise when performing genetic counseling.
2. Describe types of clinical genetic testing currently available, including each test's purpose and some of its limitations.
3. Given a clinical scenario, choose the appropriate genetic test or recommend against testing.
4. Determine whether testing of asymptomatic minors and/or prenatal testing is appropriate for patients of families concerned about the following liver diseases: Alagille syndrome, alpha-1 antitrypsin deficiency, ornithine transcarbamoylase (OTC) deficiency, Wilson disease, Niemann-Pick type C, and hemochromatosis.

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Introduction

The use and scope of clinical genetic testing is rapidly expanding. Newer technologies such as exome- and genome-based sequencing are changing the amount and type of information that patients can receive, and the expansion of direct-to-consumer testing is altering how that information is delivered.[1] Doctors will increasingly receive questions from their patients regarding genetic testing.[2,3] However, medical students often get minimal instruction in genetics, particularly the practical aspects of testing and ethical issues that may arise.[4] Many physicians report that they feel underqualified in several aspects of providing genetic services including counseling on risk factors, identifying the correct test to send, interpreting results, and navigating ethical and social issues related to testing.[5,6]

New ways of delivering this information such as online modules and simulations are being investigated.[7,8] Previous *MedEdPORTAL* publications addressed several topics in genetics

such as risk assessment[9,10] and ethics,[11] but none covered the intricacies of providing genetic testing. One source provided an excellent overview of several common genetic conditions including testing and ethical issues[12] but was geared towards pediatric residents rather than learners earlier in their career. We did not find any similar resources that provided this type of clinical genetics education for preclinical medical students. We chose to integrate the information into the basic science curriculum for medical students in their preclerkship years through an interactive lecture. We preferred this format to a standard lecture or module because we were able to provide specific expertise, engage students and encourage discussion, and address any misconceptions in real time. Our goals were to provide background knowledge on types of genetic testing and on some of the unique ethical and social issues surrounding testing,[13] then to help students apply these concepts to patient cases. Because the session was embedded within a gastroenterology course, we centered our discussion on liver diseases with a genetic component.

Methods

This session was presented in December 2018 and again in December 2019 to second-year medical students during their course on gastroenterology and liver disease. The students had previously learned about general liver pathophysiology and the specific liver diseases that we discussed.

This 1-hour session was designed as an interactive lecture incorporating large-group discussion using a PowerPoint presentation (Appendix A). The session was facilitated by a gastroenterologist and a geneticist. We first discussed general information about genetic counseling and testing, then moved on to discuss specific liver diseases and genetic counseling issues that may apply to each. We designed at least one discussion question per case.

Students were encouraged to attend the lecture in person so that they could participate. However, the session was also recorded and offered online for students to view later if they did not attend. For those students in-person, we used Top Hat to administer the questions. Top Hat is a polling software that allowed students to log in and give answers on their personal computers; however, any audience response system could be used. During the session students were asked to answer the questions through Top Hat and then the results were displayed on the projection screen. Before revealing the correct answer, we paused and asked students to share their answers and their reasoning behind them. This encouraged discussion and debate within the group. We then revealed the correct answer and discussed why each option was correct or incorrect, emphasizing some of the concepts that we had covered at the beginning of the session. Important points for discussion can be found in the discussion guide (Appendix B).

Student questions and responses were listed in Table 1 and Table 2. The information in Table 1 represented the questions that were asked during the first version of this session in 2018. After recognizing that the questions were too complex for standard *yes* or *no* answers, we changed the questions to better assess students' understanding. Answer choices regarding genetic testing were broadened to offer three possible choices: *yes, strongly recommend*; *yes, after counseling/consideration of several issues*; and *no, decline testing*. This allowed for more robust discussion and allowed us to more accurately assess our effectiveness in achieving our objectives. In 2019 we also added three general knowledge questions addressing basic principles of genetic testing (Table 2, Q1-Q3). These revised questions are included in Appendix A. We also reviewed the students' performance on the questions and on relevant questions on the final exam to analyze the effectiveness of our session.

Results

When this session was first administered in December 2018, enrollment in the course was 151 students. Twenty-five students

Table 1. Audience Response System Questions and Answers—2018 ($N = 25$)

Question	Correct Answer	Responses		Correct Responses (%)
		Yes	No	
1. Is it appropriate to test a child for alpha-1 antitrypsin deficiency?	Yes	17[a]	8[a]	68
2. Would you advise parents to have prenatal testing for Alagille syndrome?	Yes or No	18[b]	4[b]	
3. Would you counsel a carrier of ornithine transcarbamoylase deficiency to have prenatal testing?	Yes	23[c]	0[c]	100
4. Would you test the siblings of an individual with Wilson disease?	Yes	17[b]	5[b]	77
5. Would you recommend prenatal testing for Wilson disease?	Yes or No	8[d]	11[d]	
6. Would you recommend prenatal testing for Niemann-Pick type C?	Yes or No	15[c]	8[c]	
7. Would you test the minor children of known carriers of hemochromatosis?	No	9[d]	10[d]	47

[a] $n = 25$
[b] $n = 22$
[c] $n = 23$
[d] $n = 19$

Table 2. Audience Response System Questions and Answers—2019 ($N = 31$)

Question	Correct Answer	Responses				Correct Responses (%)
		A	B	C	D	
1. Would you order gene testing for the minor daughters of a woman with a breast cancer type 1 mutation?	C	0[a]	14[a]	17[a]		55
2. What is the most appropriate next step in the genetic testing of a child with autism and speech delays?	A	19[b]	2[b]	0[b]	9[b]	63
3. What is the most appropriate test to send in an infant with liver disease and distant family history of a heart defect?	C	0[b]	2[b]	28[b]	0[b]	93
4. Is it appropriate to test a child for alpha-1 antitrypsin deficiency?	B	4[c]	25[c]	0[c]		86
5. Is prenatal testing for Alagille syndrome appropriate?	B	3[d]	22[d]	2[d]		81
6. Would you recommend that a mother who is a carrier of ornithine transcarbamoylase deficiency have prenatal diagnostic testing?.	A	22[d]	5[d]	0[d]		81
7. Would you recommend genetic testing for the siblings of a child with Wilson disease?	A	18[e]	6[e]	1[e]		72
8. Is prenatal testing for Wilson disease appropriate?	B	0[f]	17[f]	6[f]		74
9. Would you offer prenatal testing to parents of a previous child with Niemann-Pick C disease?	B	2[f]	21[f]	0[f]		91
10. Would you send genetic testing for hemochromatosis in the child of an affected individual?	C	1[g]	4[g]	16[g]		76

[a] $n = 31$
[b] $n = 30$
[c] $n = 29$
[d] $n = 27$
[e] $n = 25$
[f] $n = 23$
[g] $n = 21$

came in person and participated via audience response system, and the session was recorded so that the rest of the students could view it online later. Based on the audience response system, correct responses, for those that had one correct response (Q1, Q3, Q4, Q7), ranged from 47% to 100% (Table 1).

Students' answers tended to be more consistent when the case was more clear-cut, and they varied more widely on complicated questions. For example, question three (Q3) asked about a condition where prenatal testing is clearly indicated, and 100% (23/23) of students correctly answered that *yes*, they would recommend prenatal testing. In contrast, for a more complex condition (5), 42% (8/19) answered *yes* (Table 1). We felt that this reflected that the students were appreciating that this type of testing can be complicated and ambiguous, based on the information that we discussed.

Students performed well on the 2018 end-of-course exam, with a mean score of 86% and every student passing the course. There were two specific exam questions regarding the appropriateness of familial genetic testing. One of the questions asked students whether the adult children of a man with symptomatic hemochromatosis should have genetic testing, and 66% of students correctly answered that he should. The other question asked about prenatal testing for a mother who had a previous child with ornithine transcarbamoylase (OTC) deficiency, and 77% correctly answered that treatment would need to begin within the first few hours of life.

In 2019, a total of 31 students participated in the course in person. It was recorded and online access was expanded to include all of the second-year medical students at our satellite campuses, a total of 333 students. The percentage of correct answers ranged from 55%-93% with an average of 70% on general knowledge questions (Q1-Q3), and 72%-91% with an average of 80% on questions relating to specific cases and scenarios later in the session (Q7-Q10). The improvement in scores from the general questions to the specific cases demonstrated that students learned important concepts throughout the lesson. On the exam at the end of the course, 70% of students answered the question about OTC deficiency correctly and 68% of students answered the question about hemochromatosis correctly.

Discussion

This session provided the opportunity for students to learn about the complexities of genetic testing and apply that knowledge to clinical cases. Using the Top Hat audience response system, knowledge gaps were addressed interactively during the session and important concepts were emphasized. While most medical students receive some genetics instruction, the clinical relevance of this instruction was variable and may be outdated, as genetic testing and the issues that it can produce are changing over time. Lack of knowledge about genetic testing can lead to poor outcomes including inappropriate testing, poor communication of results, and emotional harm to patients. The specifics of genetic testing will continue to change, but our goal was to provide students with a framework for approaching future problems.

We acknowledged several limitations. Our main difficulty was in assessing the effectiveness of reaching our objectives. We

had not performed any baseline assessment prior to adding this session to the curriculum, so we were unable to determine whether students' knowledge of this topic improved as a result of the session. Students' responses through the audience response system gave us a way to determine whether they were learning the main concepts that we wanted to communicate. However, this was only helpful for the students who attended the session and used the audience response system. It may be helpful to give the session via a virtual learning platform so that students could choose to watch the session from home, answer questions via audience response system, and participate in the discussion remotely. The poor attendance was comparable to what is seen in most lectures at our institution, and we were also limited by the fact that we have multiple satellite campuses where students are not able to attend in person. Having mandatory attendance or remote access may allow more students to participate. Our analysis of the sessions' effectiveness was also limited by the fact that there were only two end-of-course exam questions covering this topic, and we noticed that students did not perform particularly well on these questions, with average scores of 66% and 77% in 2018 and 70% and 68% in 2019. This was lower than what we expected, particularly in 2019 given that students scored an average of 80% on similar case-based questions during the session. It is possible that this was due to poor attendance in the session. However, we were not able to distinguish whether students who attended the session had a better performance on these test questions than students who did not. We also found that we were limited by time. This was a large amount of material to cover in a 60-minute session. In the future, we would encourage facilitators to schedule a 90-minute session to allow more time for discussion and participation.

We expect this session to work well at most medical schools. We do think that having students attend class in person is important to the success of this session. This session was given after other lectures describing the metabolic liver diseases addressed in the cases, so it is important that the session is given at the appropriate time so that students have a basic understanding of liver diseases. We had both a gastroenterologist and a geneticist available for this session. Having both experts present broadened clinical insights that could be shared during robust discussion with the students after each question. We provided a discussion guide, hoping to partially fill the potential gap in expertise if both a geneticist and a gastroenterology clinician cannot be present.

We hope to continue to expand knowledge regarding the appropriate use and interpretation of genetic testing. In the future, it would be helpful to try to reach a broader audience with this information. It may also be applicable for medical students on a gastroenterology or genetics rotation during their clerkship years, medicine and pediatric residents, and gastroenterology fellows. The group of participants may be smaller in these environments, but it should be effective as long as the learners are encouraged to participate. The case-based part of our discussion could be transformed into an interactive module for students who cannot attend an in-person discussion, although the timely feedback implemented in the audience response system would have to be replicated in the module. While it was a great opportunity to engage with medical students early in their career and explore approaches to genetic counseling, we would like to expand the effort to practicing physicians in the future.

Appendices

A. Genetic Testing & Counseling in Metabolic Liver Disease.pptx

B. Discussion Guide.docx

All appendices are peer reviewed as integral parts of the Original Publication.

Molly A. McPheron, MD: Assistant Professor, Medical and Molecular Genetics, Indiana University School of Medicine; ORCID: https://orcid.org/0000-0003-2133-1987

Hannah J. Craven, MLIS: Assistant Librarian, Ruth Lilly Medical Library, Indiana University School of Medicine

Jean P. Molleston, MD: Professor of Clinical Pediatrics, Department of Pediatrics, Indiana University School of Medicine

Christen K. Dilly, MD, MEHP: Assistant Professor, Department of Medicine, Indiana University School of Medicine; Roudebush VA Medical Center

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