MedEdPORTAL® The AAMC Journal of Teaching and Learning Resources

Original Publication

Open Access

Complexities of Clinical Genetics Consultation: An Interprofessional Clinical Skills Workshop

Jodi D. Hoffman, MD, Rachel Thompson, MD, Kathleen B. Swenson, MS, MPH, Shoumita Dasgupta, PhD*

*Corresponding author: dasgupta@bu.edu

Abstract

Introduction: Advances in genomic medicine contribute to increased demand for clinical genetics services and require physicians to understand the interprofessional practice of this field. Medical students receive a foundation in genetics during preclinical studies, but variability in clinical experience may limit knowledge of and recruitment into this clinical specialty. In this resource, we describe an approach for simulating exposure to the practice of clinical genetics during the core pediatrics clerkship. **Methods:** Prior to class, students researched and considered a mock genetics case. In class, each of four small groups discussed two cases demonstrating varied presentations, with facilitation by genetic counseling students. Each case highlighted the variability in presentation, testing, management strategies, and psychosocial issues of a genetics case. Groups reported out to the class, and individuals completed an anonymous evaluation survey. **Results:** Surveys were distributed to nine of 10 pilot sessions (210 of 235 students) with a response rate of 48%. Students frequently reported no previous exposure to seeing patients with genetics professionals, indicated a preference for learning in case discussion format over traditional lectures, and felt the format helped them apply clinical skills and reasoning. Medical students appreciated the opportunity to interact with genetic counseling students in an interdisciplinary setting and desired further educational opportunities regarding delivering complex information to patients and their families. **Discussion:** This session expanded exposure to clinical genetics content and professionals, serving as an important foundation for further development of genetic knowledge during clinical training.

Keywords

Medical Genetics, Clinical Genetics, Interprofessional, Interdisciplinary, Genetic Counseling, Pediatrics, 22q, Duchenne Muscular Dystrophy, Velocardiofacial Syndrome, DiGeorge Syndrome

Educational Objectives

By the end of the in-class session, the student will be able to:

1. Integrate physical examination findings and presenting features to propose a differential diagnosis.
2. Analyze a family history and create a pedigree to identify genetic transmission patterns and familial risks.
3. Formulate a clinical plan involving genetic testing to refine the differential diagnoses.
4. Translate clinical findings into patient-centered language to communicate genetic diagnoses, testing options, and patient/family support resources.
5. Synthesize the findings that would be presented when seeking a genetics consultation.
6. Initiate a partnership between physicians and genetic counselors.

Citation:
Hoffman JD, Thompson R, Swenson KB, Dasgupta S. Complexities of clinical genetics consultation: an interprofessional clinical skills workshop. *MedEdPORTAL*. 2020;16:10869.
https://doi.org/10.15766/mep_2374-8265.10869

Introduction

As most chromosomal and Mendelian genetic conditions are rare, few physicians frequently encounter these conditions during their professional career. However, collectively, these rare Mendelian disorders are estimated to impact nearly one in 300 live births in North America.[1] With the rapid pace of discovery, increased exposure to clinical genetics education alone is inadequate without introducing the importance of collaboration with genetics professionals. Generalists and nongenetics professionals regularly partner with medical geneticists and genetic counselors for evaluation and support. As the practice of genetics broadens to include common diseases with multifactorial inheritance, the need for partnership with genetics is ever expanding. Without

exposure to clinical collaboration with genetics professionals during the clinical years, this opportunity for improving patient care may be underrecognized.

Currently, there are approximately 1,500 medical geneticists in the US, and job postings in a recent single year exceeded 180 positions. However, of the approximately 83 slots available for graduate medical training in genetics, less than half are filled annually, translating to an enduring deficit of supply relative to demand for patient care and education.[2] The field of genetic counseling has developed over the past 50 years, which greatly expands the availability of genetics services. Recent estimates identified 3,100 genetic counselors employed in the US.[3] There are currently just over 450 clinical training positions within 49 genetic counseling master's degree training programs in North America approved by the Accreditation Council for Genetic Counseling.[4,5] Less than half of the US training programs are located within a school of medicine, whereas all four Canadian training programs are located within a school of medicine.[4] However, integration with other health care professionals in training, specifically, medical students, is not standard in all genetic counseling training programs, and therefore, physicians are often unaware of the opportunity for interprofessional collaboration. Increased exposure to the practice of clinical genetics for both medical and genetic counseling students during training is therefore critical both for recruitment of future physicians to the specialty and for collaboration between nongenetics physicians and genetic counselors.

Medical school curricula are frequently chronologically organized into two primary segments: basic or foundational sciences and clinical sciences. Because of this structure, medical students often compartmentalize their learning into these two categories, challenging their ability to apply foundational knowledge in the clinical setting without reinforcement. The observation that genetics instruction (reported by 75% of North American institutional representatives) is primarily taught in the first year of medical school,[6] the year with the highest proportion of foundational science learning, may lead students to view genetics as a basic rather than a clinical science. As the practice of medical genetics is dynamic, exposure to medical genetics during the clinical years is critical to conveying the true scope of this specialty, yet only 26% of respondents reported any inclusion of genetics content during years 3 and 4, years typically focused on clinical education. Medical students seldom observe the detailed history and physical or the intimate interaction between a clinician and a patient/family performed in a genetics consultation. These curricular trends not only limit exposure to role models in the field of clinical genetics but may also inadvertently minimize the field itself and hamper recruitment of future physicians into this specialty.[7]

This workshop has been designed to provide advantages in clinical genetics education over a standard case-based lecture. Model cases reflect what might be seen by neonatologists, pediatricians, obstetricians, internal medicine physicians, and other doctors, demonstrating the diverse presentation of genetics cases and relevance to various clinicians. Also presented are multiple psychosocial scenarios, consent issues, and relevant sensitive matters to model the complex nature of genetics consultations and the value of partnering with a genetics team to provide optimal care to families. The workshop prioritizes use of evidence-based online texts routinely consulted by genetics professionals, including Online Mendelian Inheritance in Man (OMIM)[8] and *GeneReviews*,[9] to demonstrate their utility to nongenetics professionals. OMIM allows users to identify genetic causes of conditions, mechanism of disease, and models of inheritance. *GeneReviews* provides actionable information for diagnosis, management, and counseling of patients and families. Although OMIM is used in medical training, it has been emphasized in the preclerkship curriculum,[10] and therefore, confidence with use of OMIM declines over time.[11] This loss of confidence is compounded by the limitation that medical students organize and store their learned medical knowledge in a pattern that mirrors the structure of the curriculum in which it has been learned, which can make recall and application of that knowledge (or the resources used) challenging[12] in the multifactorial and multisystem clinical environment. Clinical encounters are the scaffolding on which learners begin to bridge this gap between stored and applied knowledge, generating associations and cognitive connections between clinical presentations and their preclinical education.[13] This collaborative educational activity presents an opportunity to advance the vertical integration of the curriculum by the reintroduction of or novel exposure to online texts, knowledge of modes of inheritance and genetic diseases, and strategies of clinical diagnostic testing. The simulated patient cases push students to integrate their developing clinical reasoning skills and augmented understanding of patient care with their textbook knowledge of genetic disease and testing. Finally, using online texts and consultation with the genetic counseling student facilitator, medical students are pushed to choose diagnostic testing wisely in the cases in an evidence-based fashion, as well as to appreciate the potential psychosocial aspects of case management.

Active learning methods are employed in this educational session because they serve as the "essential ingredient of effective instruction"[14] and allow facilitators to focus on activities such as collaborative problem solving and completion of related practice activities to support learning. With a comprehensive facilitator guide and appropriate question prompts, medical students participating in this classroom workshop think through and discuss both medical and psychosocial aspects of these cases. Furthermore, through the use of this interdisciplinary model, there are opportunities to incorporate role-plays to simulate patient encounters.

Although there are examples of published genetics curricula that focus on the same diagnoses represented in these cases, they are targeted primarily toward foundational science rather than clinical practice considerations.[15-17] In terms of interprofessional curricula, there are similarly few resources appropriate for other allied health professions.[18,19] This resource fills a unique niche in uniting clinical genetics education and interprofessional practice.

Methods

Boston University School of Medicine (BUSM) currently has a traditional curriculum with 2 years of foundational science education followed by 2 years of clinical training. This session was piloted during the required, third-year, 6-week pediatrics clerkship at BUSM beginning in the 2017-2018 academic year. Each clerkship block ranged in size from 17 to 26 students distributed across a variety of clinical sites. In conducting this pilot, most students were present in the classroom, whereas several distance learners participated by video conference as described below.

To build on BUSM's first-year foundational sciences curriculum, we developed an active learning, interprofessional session through two rounds of pediatric genetics cases (Appendices A-C). In this workshop, students encountered the principles of the genetic basis of disease, diagnostic and predictive genetic testing and screening, treatment strategies, and the ethical issues and communication skills surrounding these topics. To further these goals, medical students engaged in role-play to practice delivery of the information and diagnosis. Electronic surveys were distributed to medical students (Appendix D) and genetic counseling students (Appendix E) after completion of the educational session to gauge their perceived success in achieving the learning objectives and their satisfaction with the format.

Case Design and Class Format

The content of this educational session was subdivided into preclass content review accompanied by a written assignment (Appendix B) and the in-class active learning session (Appendix C). In preparation for class, students reviewed content on Mendelian genetics and pedigree analysis, after which they completed and submitted the assignment, which served to familiarize them with content and functionality of the online textbooks to be used during the in-class session.[20-22] The preclass homework was designed to take less than 1 hour to complete. The assignment materials were hosted on Blackboard Learn, a learning management system. Students uploaded their completed homework to Blackboard Learn for review and comments. Although the assignment was ungraded in the pilot, submission was required. An answer key was provided for the grader (see Appendix A).

The content of the in-class portion focused around the diagnoses 22q11.21 and Duchenne muscular dystrophy (cases A and B, respectively, in Appendix C), with each case broken down into four subcases distributed to four randomly assigned small groups. These subcases represented variable presentations of the selected diagnoses across the life cycle and from different patient vantage points (neonatal, pediatric, adult, pregnant, and fetal), allowing future doctors of all interests to appreciate the potential yield of genetics consultation. Within the small group, students collaboratively responded to and discussed the prompts for their case using the online texts OMIM[8] and *GeneReviews*[9] as resources,[23-30] followed by a role-play of their scenario. During the report-out after groups finished case A (and subsequently case B), students benefited from peer teaching on the unique aspects of the individual subcases. In this way, students reviewed different modes of inheritance (including cases of dominant, recessive, and X-linked inheritance patterns in males and females), addressed strategies for risk assessment and psychosocial complexities, and shared their discussions around genetic and nongenetic testing options.

In the course of the role-play scenarios, facilitators encouraged medical students to practice empathic and appropriate communication skills specific to the clinical scenario and patient circumstances. Each case included complex psychosocial concerns, such as custody status, neglect, substance use, mental health issues, length of diagnostic odyssey, lack of family support, consanguinity, and/or reason for pursuing genetic testing. Prompts provided in the case served to reinforce consideration of the impact of receiving a genetic diagnosis from the patient perspective, including guilt, anger, shame, the feeling of being

overwhelmed, grief, a sense of loss, or perhaps a sense of relief and empowerment. In addition, prompts instructed students to provide patient education resources, to highlight the importance of identifying materials that are accurate and written in patient-friendly language. Facilitators skilled in addressing the genetic issues at hand and challenging social dynamics supported and advised students during the role-play.

Setup and Management of Small Groups

At the start of the workshop session, faculty provided a brief introduction to the learning goals and facilitators. Immediately afterward, the class was divided into groups of five to seven learners using a random distribution of printed handouts of the cases (Appendix C). When remote learners were participating in the session, cases 4A and 4B were held aside and assigned to these students. The course was designed for a possible maximum of four small groups, but a minimum of three is suggested to accomplish the goal of providing adequate exposure to a variety of presentations of the genetic condition in each case. Each small group on campus was assigned a genetic counselor facilitator (as described below in the Facilitator Requirements section) and designated one of its members to be responsible for providing the report-out in front of the class.

The course was modified for remote learners. On average, there were an additional two to four remote learners at separate sites who participated in the educational session using either Skype for Business or Zoom Video Communications for large-group participation and collaborated with one another by phone conference. These students conducted their own small-group sessions for both cases, and although they did not have a facilitator, they received feedback and facilitation during their report-out.

Facilitator Requirements

The overall facilitator for this workshop required prior experience and knowledge about the diagnosis, care, and counseling of patients and families with possible or known genetic conditions; for the purposes of the learning goal of interprofessional education, the small-group facilitators needed to be genetic counseling graduate students or genetic counselors. In our pilot, the overall facilitator was a board-certified clinical geneticist, and the small-group facilitators typically were genetic counseling students. When the number of genetic counseling students available was inadequate, a genetic counselor or clinical geneticist facilitated the group. In settings where genetic counselors or genetic counseling students are not available to participate, the facilitator guide (Appendix A), which provides detailed answers and guidance, can be used to train other medical educators.

Small-group facilitator preparation: Genetic counseling students enrolled in an accredited training program receive didactic training throughout their first year of graduate school education, which lays the foundation for their clinical knowledge. Additional coursework, complemented by clinical training and experiences, further hones their understanding of genetic syndromes, communication with patients, and scope of practice. In this setting, first- or second-year genetic counseling students took the role of the expert in each group—a role they often fill in clinical practice with nongeneticist physicians as well.

In addition, for preparation of the workshop, (a) all facilitators read and reviewed the facilitator guide (Appendix A), and (b) a board-certified genetic counselor or clinical geneticist with experience in teaching, as well as the medical school genetics professor, met with genetic counseling students before each session to review content and techniques of small-group facilitation, including the following:

- Details of each case.
- Group problem solving and collaborative learning.
- Modeling lifelong learning (i.e., expressing when their knowledge had been exceeded and showing how to use online or more experienced consultants to find answers).
- Role-play with active listening.
- Peer teaching.

Preclass assignment: The clerkship director reviewed the preclass assignment and provided individual feedback as needed, requiring approximately 1-3 minutes of review per student.

Student facilitator observation: The overall facilitator observed the genetic counseling students throughout the workshop with regard to small-group management skills; ability to encourage student participation versus answering questions directly, allowing time for the medical students to process and investigate while moving the case forward; and ability to present the appropriate level of confidence as a subject matter expert while encouraging collegiality with medical students. At the end of the session, genetic counseling students were allowed time to discuss this teaching experience and receive verbal feedback.

Surveys

At the close of the workshop session, medical students were emailed a link to an anonymous survey (Appendix D) hosted

on Qualtrics (Qualtrics, Provo, Utah) via the Boston University subscription. The survey asked for information about the students' perceived improvement in clinical skills surrounding the care of patients with genetic syndromes; opinions on the format of the workshop; and feedback on the preclass assignment, facilitators, and course overall. A related survey (Appendix E) was circulated to the genetic counseling students who served as facilitators. Reminders for survey completion were sent in follow-up emails to improve the response rate, but no incentives were offered.

The data analysis was generated using Qualtrics software. The free-text responses from these data sets were manually coded and thematically analyzed by two independent coders (Kathleen B. Swenson and Jodi D. Hoffman) until saturation was met.

Results

A total of 235 medical students participated in the workshop, with 30 (12%) participating as remote learners. Surveys were distributed to 210 of the 235 students, and responses were received from 100 of the 210 participants to whom the surveys had been distributed (48%). Most (87%) respondents had not previously seen a patient with a medical geneticist or genetic counselor.

Students also provided their perspectives on elements of the session and whether session learning objectives had been successfully achieved (Table 1). Most of the criteria, including ability to document a pedigree based on family history, to utilize online tools to propose clinical diagnoses, and to recognize the value added by genetic counselors and clinical geneticists in these interprofessional teams, were rated in the agree to strongly agree range.

Students had the opportunity to provide free-text responses in the Qualtrics study survey, as well as on an electronic, medical-school-designed, required session evaluation that students completed at the close of each lecture. The top three themes to emerge from analysis of the responses are the benefit of an interprofessional educational experience, the overall positive experience on the small-group facilitation, and appreciation for the opportunity to discuss how to communicate information to patients about a genetic diagnosis. Representative comments focusing on these themes follow:

- "I personally love multidisciplinary learning, and wish we had more of it throughout medical school. It was nice to have the counselors driving the conversation and bringing up salient points in our role plays, since genetic syndromes are something we do not see much of."
- "Very effective teaching. I really enjoyed the case format, especially that each of the groups had variations on the same case. I think that the genetic counseling students also added a lot to simulate how we would work on cases like those presented in real practice."
- "I thought this was a great format! I appreciated the emphasis on finding reliable resources and communicating a difficult diagnosis to patients, since these are the skills most of us will be using in practice. Much more helpful than a traditional lecture on rare genetic diseases."

Most open-ended student feedback was positive; however, some students reported discomfort with the role-play experiences in simulating delivery of difficult news or diagnoses. Many students suggested that observing a model role-play between the genetic counseling students or faculty present at the time of the workshop would be a preferred introductory or alternative learning experience.

Table 1. Medical Student Self-Assessment Responses to the Prompt "Please Rate Your Agreement With the Following Statements" ($N = 114$)

Statement	*M*[a]	*SD*
I feel prepared to analyze a family history to identify inheritance pattern and familial risks.	4.0	0.6
I feel able to document a pedigree based on a family history that I have obtained.	4.2	0.6
I feel prepared to integrate physical exam findings and presenting features using online resources such as OMIM and *GeneReviews* to propose a diagnosis in future clinical settings.	4.1	0.6
I can identify reliable resource references for suggesting genetic and laboratory testing to confirm or exclude suspected diagnoses.	4.2	0.7
I have an improved understanding of the range of clinical presentations for 22q and DMD.	4.1	0.8
I have an improved understanding of how to translate complex clinical features into family- and patient-centered language.	3.8	0.8
I feel that the class format for observing/participating in a role-play conversation around delivering a difficult diagnosis was helpful to my training.	3.4	1.2
I know where to refer families for reliable online information and support resources about a diagnosis.	4.0	0.7
I have a better understanding of the role of a genetic counselor within an interprofessional team as a result of this didactic session.	4.0	0.8
Genetic counselors and clinical geneticists should be involved in guiding the ordering and interpretation of diagnostic labs for a patient being evaluated for a genetic condition.	4.4	0.6

Abbreviations: DMD, Duchenne muscular dystrophy; OMIM, Online Mendelian Inheritance in Man.
[a]Rated on a 5-point scale (1 = *strongly disagree*, 2 = *disagree*, 3 = *neither agree nor disagree*, 4 = *agree*, 5 = *strongly agree*).

Likewise, genetic counseling student facilitators provided feedback through a postteaching study survey, their required Student Experience logbooks, and additional follow-up communication requesting their feedback. Data collected from 100% of the 18 unique genetic counseling students were also analyzed as described previously. Although more than half of the genetic counseling student facilitators reported previous experience with teaching, such as peer tutoring or providing an informational lecture, none of the genetic counseling student facilitators reported previous experience teaching medical students or facilitating small groups. Most who participated as a facilitator found the shared learning environment to be a positive experience, appreciated the chance to work with students outside of their training focus, and enjoyed sharing their clinical knowledge with other learners. Analysis of the collected survey data demonstrated that the genetic counseling students felt particularly confident about their preparation to highlight the psychosocial aspects of the presented cases, to guide medical students to appropriate resources, and to understand the range of clinical presentations for the conditions (Table 2). Furthermore, several genetic counseling student facilitators felt strongly about the importance of collaboration with other health care professionals due to their desire to raise awareness of the genetic counseling profession and the benefits of this specialized area of medical practice. Students reported the following insights, reflecting the themes that emerged from analysis of the qualitative data:

- "It was good to begin to foster a relationship between future physicians and future genetic counselors; as the designated leaders in this scenario, I believe that this could build credibility for the genetic counseling profession."
- "I think it's important as a (future) genetics provider that I educate and work with other (future) healthcare professionals because genetics is only becoming a larger and larger part of medicine. I did this as a way to help educate medical students so that in the future they're more knowledgeable about genetics AND the role of genetic counselors."

In reflecting on the format of the class, the medical students (Table 3) responded in the range of neutral to agree with regard to the format's encouraging learning application of clinical skills and its invoking clinical reasoning skills. Medical students were neutral in their preference for this format over a more traditional lecture, and they supported that the preclass assignment was helpful in preparation for the session. Genetic counseling students agreed that the format was an effective manner in which to learn the application of clinical skills and that it called on clinical reasoning skills, and they preferred facilitation of a small group over lecture for teaching style (Table 4).

Discussion

We developed an educational activity to address key gaps in medical education, including the dearth of genetics in the clinical phase of training and limited interprofessional opportunities between medical students and allied health professionals. The use of two sets of cases, each set with a shared diagnosis but varied scenario, allowed exploration of many issues that arise in genetics visits in a relatively short period of time. As only 13% of the medical students reported prior exposure to seeing a patient with a genetics professional, this activity provided students with a novel opportunity to interact closely with genetics trainees and professionals for a 1.5-hour period. Expanding the time available for such cases may be of additional value to students. This experience increased medical student awareness of the skill set that genetics specialists bring to the health care setting and the breadth of their expertise and may allow for improved utilization of resources and collaboration once in practice.

The educational goals of this session additionally focused on the development of pertinent clinical reasoning and medical

Table 2. Genetic Counseling Student Self-Assessment Responses to the Prompt "Please Rate Your Agreement With the Following Statements" ($N = 18$)

Statement	*M*[a]	*SD*
I feel prepared to facilitate small-group learning.	3.6	0.7
I feel able to provide additional clinical insight to the cases being discussed.	3.9	0.8
I feel prepared to integrate my counseling training to highlight the psychosocial aspects of the cases presented.	4.6	0.6
I can confidently guide the medical students to appropriate resources (OMIM, etc.).	4.2	0.7
I have an improved understanding of the range of clinical presentations for 22q and DMD having prepared for this session.	4.1	0.8
I have an improved understanding of how to translate complex clinical features into family- and patient-centered language due to this experience.	3.9	0.8
I have a better understanding of the training received in medical school due to this interprofessional teaching experience.	3.9	0.6

Abbreviations: DMD, Duchenne muscular dystrophy; OMIM, Online Mendelian Inheritance in Man.
[a]Rated on a 5-point scale (1 = *strongly disagree*, 2 = *disagree*, 3 = *neither agree nor disagree*, 4 = *agree*, 5 = *strongly agree*).

Table 3. Medical Student Self-Assessment Responses Regarding Class Format to the Prompt "Please Rate Your Agreement With the Following Statements" ($N = 114$)

Statement	*M*[a]	*SD*
The format of this class was helpful for learning application of clinical skills.	3.6	1.1
The format of this class called on my clinical reasoning skills.	3.6	1.1
I would prefer to learn clinical genetics in a more traditional lecture format.	3.0	1.1
The preclass assignment was helpful preparation for the class session.	3.3	1.2

[a]Rated on a 5-point scale (1 = *strongly disagree*, 2 = *disagree*, 3 = *neither agree nor disagree*, 4 = *agree*, 5 = *strongly agree*).

decision-making skills essential to caring for patients with suspected genetic disorder. After the session, students reflected positively on their perceived readiness to use skills practiced during the session, including preparedness to analyze a family history to identify inheritance pattern and familial risks, ability to document a pedigree based on a family history, and capacity to use recommended enduring online resources such as OMIM[8] and *GeneReviews*[9] to integrate physical examination findings and presenting features to propose a diagnosis and laboratory testing. Although not specifically designed to deepen knowledge of particular diagnoses, students additionally endorsed that this session provided an improved understanding of the range of clinical presentations for 22q11.21 deletion syndrome and Duchenne muscular dystrophy. Overall, students expressed significant interest in observing clinical genetics sessions, and bridging classroom to clinical learning would be an excellent avenue to pursue in the future.

In general, both medical and genetic counseling students responded favorably to the instructional methodology of this interactive learning session. One aspect that generated variable feedback was the role-play integrated into each set of cases. Our goal was to provide a low-stakes environment in which to actively practice communication of a difficult and/or emotionally fraught diagnosis with the benefit of coaching from a trained counselor or clinician. Prompts in the guide for teaching faculty

Table 4. Genetic Counseling Student Self-Assessment Responses Regarding Class Format to the Prompt "Please Rate Your Agreement With the Following Statements" ($N = 18$)

Statement	*M*[a]	*SD*
The format of this class was an effective manner in which to learn the application of clinical skills.	4.1	0.5
The format of this class called on my clinical reasoning skills.	4.1	0.5
I would prefer to prepare a lecture on this material as opposed to facilitate a small-group discussion.	2.2	0.6

[a]Rated on a 5-point scale (1 = *strongly disagree*, 2 = *disagree*, 3 = *neither agree nor disagree*, 4 = *agree*, 5 = *strongly agree*).

are designed to increase student awareness of the broad impact a diagnosis may have on the family and help support the novice role-play facilitator. A representative example of student narrative feedback supporting the role-play was that "[medical students] were used to thinking about patients more or less in a silo, solely treating the person in front of them as opposed to considering the ramifications for the family as a whole." In the future, continued faculty development around facilitating the role-play could help students with less enthusiasm for the experience engage and potentially increase the efficacy of this component of the session. Alternatively, the role-play portion of the case exercises could be adapted to have students observe a simulated session including the teaching faculty for the first set of cases, with active student participation in the second set.

Finally, we had the unique opportunity to pilot this educational session with a cohort of remote learners. These students completed their questions and role-play independently, then received support from a geneticist or genetic counselor during their report-out. Their ability to answer questions in the cases did not appear to have been negatively impacted by the absence of real-time facilitation, as this group of students provided appropriate and thorough comments when presenting their cases to the group. One student provided the following narrative comment about the experience:

> I appreciated that the remote participants were given a specific role and think of all of the various ways the workshop/flipped classrooms tried to include us, this might have worked the best in terms of being engaging and easier to follow along.

Limitations

Our project analysis has several limitations. First, as is common in survey analysis, the lack of a presession survey for comparison and lack of a robust response to the survey have the potential to introduce bias into the analysis. Second, there is the lack of objective outcome measures. In the current study, we used students' perceived readiness to use the skills outlined in the educational objectives as a proxy for observed implementation. Given the relative infrequency of genetics consultations on pediatric wards and the brief nature of the pediatrics clerkship, there is no reliable mechanism for studying whether knowledge or skills gained during the workshop are applied in a real clinical setting. Alternatively, an objective structured clinical examination to confirm short-term skill mastery might be designed for future study but was outside the scope of the current project. However,

because this workshop is designed to reintroduce genetics in the clinical environment and provide skills useful across numerous medical fields, we could create a subsequent study utilizing a new cohort of students who engage in the exercise as third-year students and complete a follow-up survey as fourth-year students to assess the longevity of the information addressed in the workshop. Finally, a limitation in the generalizability of this model to other medical schools is that some institutions may have more limited access to an affiliated genetic counseling training program to provide small-group facilitators for this interdisciplinary session. Our intent in providing the detailed facilitator guide is to offer a resource that can be used to train nongeneticists and enable faculty familiar with the guide to lead the session.

Lessons Learned and Future Directions

The Qualtrics survey data and the free-text evaluations completed by the students have been beneficial to the continued refinement of the format of the cases for the in-class portion of the workshop, the approach to the role-plays, and the methods to train facilitators for the small groups. In terms of format, initially only the facilitators were provided with the question prompts, and they used these as scaffolding for structuring the discussions with students. In time, we adjusted the format and provided prompts in the student handouts, noting that the momentum of the groups improved and students seemed to have a better grasp of the learning points when presenting to the class. In addition, we recognized from the comments that we needed to improve the messaging at the start of the workshop to clearly emphasize that the educational goals concerned interprofessional teamwork, teaching of clinical skills, resource utilization, and clinical reasoning rather than a broader review of various genetic disorders.

We were struck by the variable reception to the role-plays. Several cohorts of students requested that difficult conversations around providing a diagnosis be modeled before they were asked to role-play. Other students suggested that additional reading be added to the preclass assignment on this topic. Although role-plays are used routinely in the genetic counseling training space to provide students a safe place to explain challenging and difficult concepts, this technique is not as frequently used in the medical school curriculum, which may have contributed to student discomfort. We also hypothesize that early in the course of clinical training, students may find role-play a difficult skill and feel vulnerable when asked to participate. The student suggestions may be valuable additions to consider for a future iteration of the curriculum. We are considering moving to a hybrid model in which genetic counseling students participate in the first role-play as the provider delivering the news to help offer a framework for the second case. In the second set of cases, the medical students could work in pairs within their groups to try applying these skills to a different scenario. Alternatively, students still hesitant to role-play could think-pair-share about strategies learned from the first role-play and how to apply them in the second scenario.

Finally, it was observed that some genetic counseling students lacked confidence in answering questions posed by the medical students and/or had time management difficulty. We anticipated that this could be a risk of a structure where students facilitated an interprofessional group of peers, and therefore, genetic counseling students were provided with the case material and facilitator guide in advance of the session, accompanied by a brief facilitator training session. However, we agree that augmenting the presession preparation to reinforce teaching skills and include instruction specifically on time management with the cases, methods for handling uncertainty, and techniques for exhibiting more confidence when leading a small group could improve this concern. In addition, we may consider allowing genetic counseling students to run a mock discussion as part of their preparation. Having the clinical geneticist circulate more actively between groups to field questions and provide valuable additional knowledge to the discussions could provide additional needed support as well.

In closing, we believe that this interactive workshop is a unique resource for integrating the practice of clinical genetics and preclinical knowledge into the core curriculum for pediatric and other clerkships. The interprofessional structure mirrors and reinforces best practices in the collaborative care of patients with suspected underlying genetic disease while simultaneously enhancing the knowledge and skill sets of both medical and genetic counseling student participants. Although the cases focus on only two underlying diagnoses, students complete the workshop with an appreciation for the broad variability possible in presentation and a core clinical skill for the use of reliable online tools to augment their clinical reasoning and management planning in an evidence-based and cost-effective fashion. It is our hope that with the material presented in an intellectually challenging yet engaging format, some students may have a sustained interest and consider pursuing a career in this exciting and stimulating field.

Appendices

A. Facilitator Guide.docx

B. Student Homework Assignment.docx

C. Student Case Handouts.docx

D. Medical Student Survey.docx

E. Genetic Counseling Student Survey.docx

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

Jodi D. Hoffman, MD: Associate Professor, Department of Pediatrics, Boston University School of Medicine

Rachel Thompson, MD: Assistant Professor, Department of Pediatrics, Boston University School of Medicine

Kathleen B. Swenson, MS, MPH: Assistant Professor, Department of Medical Sciences and Education, Boston University School of Medicine

Shoumita Dasgupta, PhD: Professor, Department of Medicine, Biomedical Genetics Section, Boston University School of Medicine

Disclosures
None to report.

Funding/Support
None to report.

Ethical Approval
Reported as not applicable.

References

1. Srinivasan BS, Evans EA, Flannick J, et al. A universal carrier test for the long tail of Mendelian disease. *Reprod Biomed Online*. 2010;21(4):537-551. https://doi.org/10.1016/j.rbmo.2010.05.012
2. Campion M, Goldgar C, Hopkin RJ, Prows CA, Dasgupta S. Genomic education for the next generation of health-care providers. *Genet Med*. 2019;21(11):2422-2430. https://doi.org/10.1038/s41436-019-0548-4
3. Genetic counselors. U.S. Bureau of Labor Statistics website. https://www.bls.gov/ooh/healthcare/genetic-counselors.htm. Accessed August 13, 2018.
4. Program directory. Accreditation Council for Genetic Counseling website. http://gceducation.org/Pages/Accredited-Programs.aspx. Accessed March 12, 2019.
5. Riconda D, Grubs RE, Campion MW, Cragun D. Genetic counselor training for the next generation: where do we go from here? *Am J Med Genet C Semin Med Genet*. 2018;178(1):38-45. https://doi.org/10.1002/ajmg.c.31598
6. Plunkett-Rondeau J, Hyland K, Dasgupta S. Training future physicians in the era of genomic medicine: trends in undergraduate medical genetics education. *Genet Med*. 2015;17(11):927-934. https://doi.org/10.1038/gim.2014.208
7. Hyland K, Dasgupta S. Medical genetics and genomics education and its impact on genomic literacy of the clinical workforce. *Genet Med*. 2019;21(5):1259-1260. https://doi.org/10.1038/gim.2017.127
8. OMIM website. https://www.omim.org/. Accessed August 14, 2018.
9. Adam MP, Ardinger HH, Pagon RA, et al., eds. *GeneReviews*. NCBI website. http://www.ncbi.nlm.nih.gov/pubmed/20301295. Accessed August 14, 2018.
10. Diehl AC, Reader L, Hamosh A, Bodurtha JN. Horizontal integration of OMIM across the medical school preclinical curriculum for early reinforcement of clinical genetics principles. *Genet Med*. 2015;17(2):158-163. https://doi.org/10.1038/gim.2014.84
11. Lee-Barber J, Kulo V, Lehmann H, Hamosh A, Bodurtha J. Bioinformatics for medical students: a 5-year experience using OMIM in medical student education. *Genet Med*. 2019;21(2): 493-497. https://doi.org/10.1038/s41436-018-0076-7
12. Bowen JL. Educational strategies to promote clinical diagnostic reasoning. *N Engl J Med*. 2006;355(21):2217-2225. https://doi.org/10.1056/NEJMra054782
13. Custers EJFM, Regehr G, Norman GR. Mental representations of medical diagnostic knowledge: a review. *Acad Med*. 1996; 71(10)(suppl):S55-S61. https://doi.org/10.1097/00001888-199610000-00044
14. Clark RC. *Evidence-Based Training Methods: A Guide for Training Professionals*. 2nd ed. Alexandria, VA: ATD Press; 2015.
15. Brescia WF, Park VM, Ward JC. Introductory TBL exercise on genetic risk. *MedEdPORTAL*. 2016;12:10381. https://doi.org/10.15766/mep_2374-8265.10381
16. Whitfield C, Schwentker E. On tiptoes. *MedEdPORTAL*. 2009;5:463. https://doi.org/10.15766/mep_2374-8265.463
17. Shanower G. Pediatric genetics in the clinic. *MedEdPORTAL*. 2013;9:9636. https://doi.org/10.15766/mep_2374-8265.9636
18. De Biase I, Diaz-Ochu M, Rindler M, Hobson-Rohrer WL. Introduction to biochemical genetics from the clinical laboratory prospective: a case-based discussion. *MedEdPORTAL*. 2017; 13:10586. https://doi.org/10.15766/mep_2374-8265.10586
19. Dasgupta S. Medical genetics ethics case collection: discussion materials for medical students in the genomic era. *MedEdPORTAL*. 2017;13:10562. https://doi.org/10.15766/mep_2374-8265.10562

20. Hartz PA, Hamosh A, Kniffin CL, et al. Williams-Beuren syndrome; WBS. OMIM website. https://www.omim.org/entry/194050. Published June 2, 1986. Updated September 18, 2017.

21. Kniffin CL, O'Neill MJF, Wright MJ, Hamosh A, Jackson JF. Williams-Beuren syndrome; WBS [synopsis]. OMIM website. https://www.omim.org/clinicalSynopsis/194050. Published June 15, 1995. Updated March 23, 2015.

22. Morris CA. Williams syndrome. *GeneReviews*. NCBI website. https://www.ncbi.nlm.nih.gov/books/NBK1249/. Published April 9, 1999. Updated March 23, 2017.

23. Hamosh A, Kniffin CL, Przylepa KA, et al. Velocardiofacial syndrome. OMIM website. https://www.omim.org/entry/192430. Published June 2, 1986. Updated October 14, 2013.

24. Kniffin CL, Wright MJ, Hamosh A, Jackson JF. Velocardiofacial syndrome [synopsis]. OMIM website. https://www.omim.org/clinicalSynopsis/192430. Published June 15, 1995. Updated November 29, 2010.

25. Hamosh A, Kniffin CL, O'Neill MJF, et al. DiGeorge syndrome; DGS. OMIM website. https://www.omim.org/entry/188400. Published June 2, 1986. Updated January 31, 2014.

26. Kniffin CL, O'Neill MJF, Kelly J, Przylepa KA. DiGeorge syndrome; DGS [synopsis]. OMIM website. https://www.omim.org/clinicalSynopsis/188400. Updated November 29, 2010.

27. McDonald-McGinn DM, Emanuel BS, Zackai EH. 22q11.2 deletion syndrome. *GeneReviews*. NCBI website. https://www.ncbi.nlm.nih.gov/books/NBK1523/. Published September 23, 1999. Updated February 28, 2013.

28. Hamosh A, Tiller GE, Converse PJ, et al. Muscular dystrophy, Duchenne type; DMD. OMIM website. https://www.omim.org/entry/310200. Published June 4, 1986. Updated November 26, 2018.

29. Kniffin CL, Amberger JS, Wright MJ, Hamosh A, Jackson JF. Muscular dystrophy, Duchenne type; DMD [synopsis]. OMIM website. https://www.omim.org/clinicalSynopsis/310200. Published June 15, 1995. Updated May 21, 2010.

30. Darras BT, Urion DK, Ghosh PS. Dystrophinopathies. *GeneReviews*. NCBI website. https://www.ncbi.nlm.nih.gov/books/NBK1119/. Published September 5, 2000. Updated April 26, 2018.

Received: March 29, 2019
Accepted: September 4, 2019
Published: January 24, 2020