

Original Publication

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Medical Genetics Ethics Case Collection: Discussion Materials for Medical Students in the Genomic Era

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Abstract

Introduction: This collection of cases in medical genetics focuses on ethical dimensions of genetic testing. Given the recent and continuing revolution in genetic testing technologies, understanding the nuances of genetic tests and the implications of their outcomes for patients is a critical learning goal for medical students. **Methods:** This case collection was developed for first-year medical students. The cases fall into two types: discussion cases that lend themselves to both small-group and lecture settings and brief audience-response clicker cases to be used in larger lecture settings. The cases span topics such as direct-to-consumer genetic testing, patient privacy, economic and legal issues of genetic testing, and secondary findings in whole exome/genome sequencing. The clicker cases can be used to punctuate class sessions on the related science, while the discussion cases can be deployed as a single 2-hour session focusing on ethics. The associated materials include teaching notes on the scientific and ethical dimensions of the cases, a slide presentation of the cases, and implementation advice. **Results:** Students found that engaging with these cases was very stimulating and eye-opening. Student comments indicated that they appreciated the opportunity to grapple with the ethical dimensions of the genetic testing technologies and that the challenges brought to light highlighted the complexity of medical practice in the genomic era. **Discussion:** Although these cases were originally developed for use with medical students, they could easily be adapted for use in postgraduate and CME settings to explore complex ethical scenarios on which even the experts disagree.

Keywords

Incidental Findings, Stem Cells, Pharmacogenetics, Genomic Medicine, Whole Genome Sequencing, Whole Exome Sequencing, Direct-to-Consumer Genetic Testing, Regenerative Medicine, GINA, Precision Medicine, Secondary Findings, Mitochondrial Manipulation, Mitochondrial Replacement Therapy

Educational Objectives

At the end of this session, learners will be able to:

1. Consider the potential advantages and disadvantages of widespread use of whole genome sequencing approaches and direct-to-consumer initiatives.
2. Identify the critical need to protect individual privacy as it relates to genetic test results and genetic databases in order to safeguard their impact on patients' family relationships, employment status, and ability to secure health insurance.
3. Appraise the nuances and consequences of the current recommendations around the reporting of genetic test results with respect to whole genome sequencing.
4. Recognize the economic ramifications of genetic technology for precision medicines and patented inventions.
5. Formulate a deeper understanding of the ethical dimensions of genetic testing technologies and the complexity of medical practice in the genomic era.

Appendices

- A. Medical Genetics Ethics Cases Presentation.pptx
- B. Medical Genetics Ethics Cases Handout.docx
- C. Medical Genetics Ethics Cases Facilitator Notes .docx
- D. Ethics Cases Audience-Response Results.pdf

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

Introduction

As understanding of the genetic basis of human disease has grown, genetic testing applications for these conditions have grown in parallel.^{1,2} Discussion of the nuances of these testing strategies in the context of direct-to-consumer genetic testing, identification of secondary findings in genomic strategies, and other ethically complex scenarios is imperative in order for trainees and physicians to fully consider responsible use of genomic technologies.³⁻⁵

Although general frameworks for ethics in the practice of medical genetics have been proposed,⁶⁻⁸ with the development of new genetic and genomic technologies have come new specialized cases requiring application-specific ethical analyses. For instance, the American College of Medical Genetics and Genomics has systematically provided clinical and ethical guidance on genetic testing in the context of direct-to-consumer genetic testing,⁹ noninvasive prenatal screening,¹⁰ and clinical exome and genome sequencing,¹¹ especially as this last point pertains to the reporting of secondary findings^{12,13} and informed consent.¹⁴ Notably, experts in the field do not uniformly support these recommendations,¹⁵ indicating that simply memorizing a group of practice guidelines is not sufficient. Rather, trainees need to be comfortable with contemplating the ethical implications of the genomic technologies they will be using in the clinic so as to adapt in this rapidly evolving field.

A small number of resources have been developed that can be used to introduce trainees to the ethics surrounding genetic testing for hereditary breast and ovarian cancer,¹⁶ noninvasive prenatal testing for single-gene disorders,¹⁷ preimplantation genetic diagnoses,¹⁸ the clinical utility of whole exome or genome sequencing,¹⁹ nondirective counseling,²⁰ and privacy of genetic test results.²¹ There is also a small series of online cases²² that touches upon a number of these topics but does not provide teaching notes or discussion prompts to make the cases usable for in-depth discussion in the classroom. This current collection of discussion and clicker/discussion cases aims to delve further into these emerging areas in genomic medicine and provide educational tools to encourage critical thinking and analysis of the scientific, ethical, legal, and social implications of these genomic technologies. This case collection also includes facilitator teaching notes, a student handout, and a slide presentation to aid in implementation in the classroom.

These cases were used at the Boston University School of Medicine beginning in 2004. The content was discussed annually in a Medical Genetics (2004-2015) or Principles Integrating Science and Medicine (2016-present) course for first-year medical students, with the addition of new cases as the field progressed. The cases were used both in ethics-focused educational settings and in sessions that focused mainly on clinical science. Thus, the cases can be used in the context of both traditional and integrated curricula. Furthermore, the cases are designed to work well both in small-group settings (discussion cases) and in large-format lecture halls (clicker questions and discussion cases). By implementing some or all of these cases, instructors in medical schools, residency programs, and CME settings can promote high-level discussion of the ethical complexities of practicing medicine in the genomic era.

Methods

Materials

The cases are provided in two groups: discussion cases and discussion cases with clicker questions (Appendices A & B). Instructor notes covering the scientific and ethical dimensions of the cases are also provided (Appendix C).

A classroom equipped with an LCD projector and an audience-response system should be utilized for the clicker case discussions. In this setting, the facilitator leads an interactive case discussion employing the PowerPoint slides (Appendix A) and audience-response technology when prompted in the presentation. Instructors can use electronic audience-response systems such as Turning Point or iClicker. This clicker-based audience-response technology lets students submit their responses to questions posed on the slides, thereby allowing for immediate polling of student opinions on topics. If the institution does not have this technology available, class polling of answers can be accomplished using sheets of colored paper that

students hold up in class to share their opinions. However, an important advantage of using the electronic clickers is that responses on these systems occur instantly in real time and are anonymous to peers. Given that the discussion involves nuanced and sometimes personal ethics topics, this anonymity can be an advantage. These clicker questions can be used in large-format classes, and students can report individual opinions before engaging in think-pair-share exercises within the larger room. Sample clicker data from use of these cases with first-year medical students are included in Appendix D. Notably, there is no general consensus of opinion on these cases. This fact lets the cases serve as the starting point for in-depth discussions that highlight differing perspectives and approaches in handling each case.

The discussion cases can be used with live discussion in a large lecture hall with similar techniques to those described above or in smaller discussion groups. They may even be used as writing prompts for essay-based assignments. The discussion cases are available in the PowerPoint collection (Appendix A), but handouts with the cases (Appendix B) can be distributed in advance of a small-group discussion, for which the facilitator has discussion prompts (Appendix C).

Length

If an instructor is planning to use the full set of discussion cases in one session, allotting 2 hours of class time is recommended. However, the cases are independent and can be distributed among multiple sessions where the relevant science is introduced. The cases in this collection may be used both throughout an integrated foundational sciences curriculum and in a special session focusing on ethics. The ease with which these cases can be adapted to various curricular models is a strength of this resource.

Room Setup and Facilitation

The following setup procedures have been found to be important when administering the session with a large group (approximately 180 students). With groups this size, a large auditorium or hall may be used. Ideally, students sit at round tables, with each table forming a group of six to eight students. Students can talk over the discussion questions within their groups, and the facilitator can ask for group opinions. The facilitator should also take care to repeat questions and comments from individual groups of students so that all participants in the large hall can hear the contributions of their peers. Alternatively, the room can be set up conference style, with microphones in the aisles that students step up to so as to be amplified for the discussion. To best facilitate this type of discussion, the facilitator should have the ability to welcome a range of responses from students, to subsequently invoke recommendations from professional societies, and to apply the material to patient scenarios.

Educational Context and Target Audience

In-class discussion does not necessarily need to be limited to the preclerkship years of medical school, but students should be experienced in the concepts of direct-to-consumer genetic testing, whole exome/genome sequencing, mitochondrial manipulation, privacy of genetic information, and patenting of genetic material. For example, background topics can be introduced in the preclerkship phase of the curriculum, and application of this knowledge to cases can occur at that time or within the context of clerkships in pediatrics, neurology, obstetrics and gynecology, hematology/oncology, or other relevant medical specialties. Importantly, the extent to which the technical science is the focus can vary without impacting the relevance of the ethical discussion.

Discussion of these cases does require some comfort on the part of the learners when discussing potentially controversial issues in a classroom setting. For this reason, use of anonymity-preserving techniques, such as audience-response systems or small groups, to create a safer space for discussion of potentially sensitive issues is recommended.

Results

The interactive cases were successfully implemented in a class of 180 first-year medical students. Audience-response data from in-class use of these cases are included in Appendix D. Notably, student opinions for these cases were wide ranging, which allowed for more in-depth discussion of contrasting

viewpoints and highlighted the need for discussion of these complex topics among both trainees and experts. These discussions served as an excellent launching point to allow students to break into smaller groups within the lecture hall, compare their perspectives on these issues, and then share their views with the larger student group.

Students' engagement during the sessions was high, and they raised many complex points. There was rich discussion on the influence of for-profit genetic testing companies, the nature of secondary findings, and the limited preventative options available in genetic conditions. Feedback from course evaluations was strongly positive. Examples of such feedback include the following statements:

- “[The professor] recognized the relevance of race, ethnicity, sexuality, and cultural difference on what she/he was teaching, and . . . addressed these topics in a respectful way that enhanced our overall understanding of the material and our ability to provide good care to diverse patient populations. This course should provide the standard for teaching diversity in our other courses.”
- “I really appreciated how [the professor] brought in real-world applications of the material and pushed us to think about the issues that we will face in our practice of medicine.”
- “Small group discussions between students were always engaging as they touched on ethics, future technology, and prompted us to reflect on our value systems and future practice.”
- “[The professor] has done a great job in organizing this course. The topics were relevant and the information was current and genetics as a social, political, ethical issue was also covered which was really informative.”
- “The repeated references to the socioeconomic considerations around testing, screening, and access were greatly appreciated as a way to tie the knowledge to the clinical and societal applications/implications of genetic medicine.”
- “The best section of the course for me was the lecture on the historical and emerging issues in ethics in genetics, and I think the points brought to light are very real challenges most of us will encounter in the future in practice.”
- “It’s been one of my favorite classes thus far. A lot of that has to do with how [the professor] emphasized the fact that medical genetics is fraught with ethical issues that force us to consider what it truly means to be human—one of the fundamental questions that got me interested in medicine in the first place.”

Discussion

This case collection is a versatile pedagogical resource as it is easily adaptable to multiple curriculum structures (traditional or integrated), settings (from large lecture to small-group discussions), and trainee levels (preclerkship, clerkship, residency, or CME). The trainees at the Boston University School of Medicine were very engaged with the complex concepts introduced by these cases, which reflected issues they would face in their future clinical practice.

This collection of cases has been successfully deployed over the past 13 years, with the content adapting over the years to developments in the field of genomic medicine. As technology has progressed, ethical issues have warranted further discussion, and the medical students have been highly engaged in these discussions. They have consistently provided positive feedback on the session. Students have also suggested future directions and future points of discussion to be incorporated in later iterations. An example of one proposed future direction is a discussion of the presence of genetics in popular media. However, it should be noted that despite this supportive feedback demonstrating student engagement with the content, a limitation of this resource is the lack of quantitative data to support the effectiveness of introducing ethics into a medical curriculum using these cases. This will be an important avenue for future inquiry.

Although this resource may be utilized in a large-group setting, small-group settings may a preferred pedagogical model for delving even deeper into the content. When considering use of these cases in a small-group setting, the limiting resources are likely to be discussion leaders, rooms, and curricular time. To address the limiting resource of discussion leaders, one potential solution could be to incorporate

interprofessional models of education. Clinical genetics is a specialty for which there is a shortage of practitioners; however, these individuals also work closely with genetic counselors.²³ For the purposes of educating trainees in genomic medicine, genetic counselors or genetic counseling students would be excellent small-group discussion leaders, would provide valuable clinical context, and would have opportunities to model the interprofessional nature of their practice.

This case collection, although covering a range of ethics topics relevant to genomic medicine, allows only for introductory analysis of these issues. If time permits, perhaps in the context of a specialized clinical elective on medical genetics, students could engage with the practice guidelines in these areas to develop a deeper understanding of the principles governing these guidelines' development. This is likely beyond the scope of what is appropriate for first-year medical students, but it would be an interesting exercise for trainees wishing to specialize in this area.

Medical schools are embracing more active forms of learning.²⁴ This type of activity offers students an in-depth and active experience regarding the content surrounding the rapidly evolving fields of precision and genomic medicine.

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Ethical Approval

Reported as not applicable.

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