



Genetics Society of America
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Medical Genetics Ethics Cases Resource Justification

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Resource Description

As our understanding of the genetic basis of human disease has grown, genetic testing applications for these conditions have grown in parallel. Discussion of the nuances of these testing strategies in the context of direct-to-consumer genetic testing, identification of incidental findings in genomic strategies, and other ethically complex scenarios is imperative for trainees to fully consider responsible use of genomic technologies.

The case collection was developed for use in a first year Genomic Medicine course for medical students at the Boston University School of Medicine. Although this was initially piloted with medical students, it would be appropriate to use with advanced undergraduate students as well as graduate students with an interest in the clinical applications of the science they are studying.

Resource Justification

This resource is aligned primarily with the GSA's core concepts in genetic variation and core competencies in communication and critique of scientific issues relating to society or ethics.

Under the umbrella of the GSA genetic variation core concept, we pose a variety of cases to the students where the identification of genetic variation that can cause human disease is the central genetic finding. In terms of the GSA competency relating to critique of scientific issues relating to society or ethics, we challenge students to consider the ramifications of identification of human genetic variation when it is conducted in a direct-to-consumer approach. Likewise, we have the students consider the implications of uncovering disease-causing mutations incidentally while actually searching for genetic variation to explain a different clinical presentation. This case collection also allows the students to explore their beliefs on appropriate use of genome manipulation technologies, disclosure of testing results for adult onset conditions to children, and privacy of genetic information. Because all of this is done in a discussion setting, students will also be developing their proficiency in communicating genetics concepts.

Students gain experience applying these concepts to a translational context through the in-class cases, grappling with both scientific and ethical issues. The cases themselves are brief, frequently do not have a single best answer, and therefore maximize opportunity for productive classroom debate. This is a unique pairing that encourages students to think critically and translationally.

Session Learning Objectives

At the end of this section, the student should 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 of genetic test results and genetic databases to safeguard their impact on a patient's family relationships, their employment status, and their ability to secure health insurance.
3. Appraise the nuances and consequences of the current recommendations around 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.

Keywords

genomic medicine, ethics, medical genetics, whole genome sequencing, whole exome sequencing, direct-to-consumer genetic testing, stem cells, regenerative medicine, pharmacogenetics, GINA, precision medicine, incidental finding, secondary finding.