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Committee on Genetics
This Committee Opinion was developed by the American College of Obstetricians and Gynecologists’ Committee on Genetics in collaboration with committee members Steven J. Ralston, MD, MPH, and Susan D. Klugman, MD, with consultation from Judith Daar, JD.

Legal Considerations in Genetic Screening and Testing: Three Case Studies

ABSTRACT: The rapidly evolving genetic technologies that are available to patients and obstetrician–gynecologists have transformed the practice of clinical medicine. From cell-free DNA screening technologies in pregnancy to expanded carrier screening and hereditary cancer gene panels, obstetrician–gynecologists often are faced with questions about their legal responsibilities regarding genetic information as well as the legal ramifications of this information for their patients. The Committee on Genetics has constructed the following case studies to highlight some of the legal issues an obstetrician–gynecologist may encounter when performing genetic testing. These cases do not cover the breadth of legal issues affecting clinical genetics, but rather they illustrate certain legal concepts and principles as well as key pieces of legislation that are pertinent to clinical care. These case descriptions are not intended to serve as legal advice. Obstetrician–gynecologists are strongly encouraged to seek expert legal assistance to resolve questions involving legal rights or responsibilities.

Recommendations and Conclusions

- Legal and ethical requirements protect the identity of a patient’s confidential information. Under these requirements, a patient’s health information (including genetic diagnoses) should be kept confidential; however, if a patient gives you permission to reveal this information, you may do so to counsel the relevant family members.
- Obstetrician–gynecologists should not withhold information from a patient regarding her health, including test results, and have a responsibility to disclose such information, especially when it is of potential relevance to decision making.
- Obstetrician–gynecologists are encouraged to seek guidance from an ethics committee, an attorney, or both, before deciding whether to reveal genetic information to or withhold it from family members who may be at risk.
- Many prenatal genetic tests have the potential to reveal information about the patient and her family that is unexpected or unsuspected and that was not part of the original intent for testing. Such results often raise complex ethical and legal issues and may require consultation with a genetics professional, ethicist, or legal counsel.

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Case Number 1. Genetic Testing and Its Effect on Future Insurability

A 24-year-old patient sees you for BRCA1 testing because her mother has breast cancer and was found to be BRCA1 positive. The patient is worried that if she tests positive she may have difficulty obtaining health, life, or disability insurance. She also is concerned that her employer may fire her because of concerns that if she develops breast cancer, premiums will increase for all employees.

Some of this patient’s concerns are well founded, although others are not. The Genetic Information and Nondiscrimination Act (known as GINA) of 2008 is a federal law that prevents health insurance carriers from using or requesting genetic information to make decisions about coverage, premiums, or eligibility. The Patient Protection and Affordable Care Act of 2012 went further and protected individuals with preexisting conditions (a BRCA1 pathogenic variant, a new term for what was formerly referred to as a “mutation,” would fall into this category) from losing their health insurance or facing exorbitant premiums. This patient’s ability to obtain health insurance would not be affected by a positive BRCA1 test because of GINA, and the Patient Protection and Affordable Care Act would protect her eligibility (and premiums) if she developed cancer. Nevertheless, this mandated coverage of preexisting conditions is dependent on the survival of the Patient Protection and Affordable Care Act or its replacement with legislation or regulations that include similar protections.

It is critical, however, for her to understand that these protections do not extend to other forms of insurance, such as life, disability, and long-term care insurance. The Genetic Information and Nondiscrimination Act only will protect her from employment discrimination if her employer discovers the results of her genetic testing if she works for an organization of more than 15 employees. Smaller companies are excluded from this prohibition.

These insurance and employment issues, along with the medical complexities of genetic testing, highlight the role and value of trained genetic counselors and genetics practitioners in helping patients make these difficult screening and testing decisions.

Case Number 2. Familial Cancer Syndrome and Privacy

Patient A is a 34-year-old new patient in your practice. Her sister, patient B, also is in your practice. In taking patient A’s medical history, she tells you that she recently had a colectomy because of extensive polyposis. She was found to have the APC gene associated with autosomal dominant familial adenomatous polyposis syndrome, one of several hereditary cancer syndromes. When taking the family history, she tells you that her father died of colon cancer at 48 years of age. You think you should reveal the information about her APC pathogenic variant to her sister because she has a 50% chance of being a carrier as well and may want to have genetic screening for this colon cancer syndrome.

Legal and ethical requirements protect the identity of a patient’s confidential information. Under these requirements, a patient’s health information (including genetic diagnoses) should be kept confidential; however, if a patient gives you permission to reveal this information, you may do so to counsel the relevant family members. The case becomes challenging when the affected patient does not give permission to reveal her test result. Thus, if patient A did not give you permission to disclose, you could discuss the family history and its implications with patient B, but you could not discuss patient A’s APC pathogenic variant.

The Health Insurance Portability and Accountability Act of 1996, (known as HIPAA), is relevant in this situation. The rules set out in HIPAA and associated regulations protect the privacy of individually identifiable health information, including the genetic test result discussed in this case. Physicians and others who are covered by HIPAA are prohibited from disclosing protected health information to third parties (in this case the sister) without written authorization from the patient (1). The Health Insurance Portability and Accountability Act contains some limited exceptions permitting disclosure when the covered entity has a good faith belief that the disclosure 1) is necessary to prevent or lessen a serious and imminent threat to the health or safety of a person or the public, and 2) is to a person reasonably able to prevent or lessen that threat. Whether this exception to the privacy rule would include disclosure of genetic test results to a potentially affected family member is uncertain but seems unlikely except in very limited circumstances in which the threat is both imminent and preventable. To date, there are no published judicial opinions addressing the application of the HIPAA privacy exception to unauthorized disclosure of an inheritable risk of disease.

To be clear, before the enactment of HIPAA, disclosing patients’ personal health information did have legal ramifications; however, these ramifications usually were addressed by litigation brought by the patient, including negligence claims for a breach of confidentiality. Although HIPAA did not make these torts disappear, it added federal civil monetary and criminal penalties to the consequences of inappropriate disclosure of patients’ confidential information.

In addition to HIPAA, there are some relevant court cases dealing with personal health information and a so-called “duty to warn.” For example, in
In Pate v. Threlkel (3), the court found that a physician has the duty to warn a patient regarding the risk to her children from her inheritable form of cancer, and that the physician owes that duty both to the patient and to her children. Similarly, Safer v. Estate of Pack (4) found that a physician has a duty to a patient and to family members to warn of “avertible risk from genetic causes,” however, unlike in the Pate case, the court declined to state whether the physician can satisfy this duty by warning the patient or whether the physician must warn the family members. Obstetrician–gynecologists are encouraged to seek guidance from an ethics committee, an attorney, or both, before deciding whether to reveal genetic information to or withhold it from family members who may be at risk. In this case, however, disclosure may be unnecessary and can be avoided as the physician could easily offer genetic testing to patient B in light of a family history of colon cancer at a young age.

Case Number 3. Findings of Consanguinity on Prenatal Testing Results

A 35-year-old patient undergoes amniocentesis and the karyotype is reported as 46, XY, but the chromosomal microarray shows areas of homozygosity, which strongly suggests parental consanguinity. Are you legally obligated to reveal this information to the patient and what should be discussed?

Obstetrician–gynecologists should not withhold information from a patient regarding her health, including test results, and they have a responsibility to disclose such information, especially when it is of potential relevance to decision making. However, many prenatal genetic tests have the potential to reveal information about the patient and her family that is unexpected or unsuspected and was not part of the original intent for testing. Such results often raise complex ethical and legal issues and may require consultation with a genetics professional, ethicist, or legal counsel. Often, the clinical report or a discussion with the laboratory can provide more information to help in counseling the patient. In this case, the degree of homozygosity could reveal how closely the parents are related by blood.

In this case, the test result may have important implications regarding the health of the fetus and may have implications for future pregnancies. It is necessary to inform the patient of this finding and to consider the specific areas of homozygosity and the potential genes that may be involved. The possible risk of autosomal recessive conditions in the fetus should be discussed with the patient. In addition, if large areas of homozygosity are found, this test result may imply a close blood relationship, such as that seen with incestuous relationships. It may be necessary to have a discussion with the patient if you suspect that she is a survivor of incestuous rape. Obstetrician–gynecologists are encouraged to use other American College of Obstetricians and Gynecologists’ documents, including Committee Opinion 518 Intimate Partner Violence (6) for further resources.

Adequate pretest counseling and screening, including an extensive family history, may help to avoid discovering information unexpectedly. Patients should be made aware that information about blood relatedness may be revealed by some prenatal genetic tests. Because of the complex genetic, ethical, and legal implications of discovering such information, it will often be prudent to involve a genetic counselor, genetics professional, ethics consultant, or legal expert in caring for these patients.

Conclusion

The choice to perform genetic testing can have legal ramifications for patients and physicians. Patients require informed consent before these tests, and obstetrician–gynecologists could consider using genetic counselors or other practitioners trained in genetics when performing tests that are not routine or that require complex pretest or posttest counseling. Legal requirements may differ across the jurisdictions in which health care providers practice, and obstetrician–gynecologists should reach out to local ethics committees or to attorneys familiar with these issues when questions arise or counsel is necessary.

References

3. Pate v. Threlkel. 661 So.2d 278 (Fla. 1995).


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