Ethical and Legal Implications of Cancer Genetic Testing: Do Physicians Have a Duty to Warn Patients' Relatives About Possible Genetic Risks?

By Courtney Storm, JD, MBE, Rinki Agarwal, MD, and Kenneth Offit, MD, MPH

Introduction

The use of genetic screening as a new tool for early detection and cancer prevention is a significant medical advancement that can also result in ethical and legal challenges for physicians. Some of these challenges arise from tension between the principles of patient autonomy and confidentiality on one hand, and the duty to warn family members of known genetic risks on the other. This vignette raises questions about the extent of physicians' obligations to warn relatives of a patient about a hereditary cancer risk.

Vignette

A 34-year-old woman with uterine cancer comes to your office for treatment. She tells you that her father died of a transitional cell carcinoma of the ureter, and that there is also a history of a grandparent dying at an early age of a gastrointestinal cancer. Her 36-year-old brother is sitting in your waiting room. You notice that the pathology report from your patient's uterine cancer showed absence of expression of the protein encoded for by the gene *MSH2*. You recommend *MSH2* sequencing to confirm, but based on those data and the patient's family history, you make the preliminary diagnosis of hereditary nonpolyposis colon cancer syndrome, which carries an 80% lifetime risk of colon cancer, and is usually diagnosed in patients aged 40 to 50 years. You suggest that your patient inform her brother that he is at greatly increased risk for colon cancer and should consider genetic testing and screening. Your patient tells you that she will not inform her brother of his potential genetic risk because she is afraid of genetic discrimination. Under these circumstances, what are your obligations to your patient, and to your patient's brother sitting in the waiting room?

Discussion

It is not uncommon for physicians to encounter patients who do not want to notify a relative of potential genetic risk. In a survey of geneticists, 60% of respondents were involved in caring for a patient who did not want to inform relatives about their potential genetic risk for reasons including pre-existing estrangement, fear of blame, and fear of insurance discrimination.[1] When patients are reluctant to share relevant genetic information with family members, physicians may have to consider how to balance their patients' privacy interests with the interests of at-risk family members who could benefit from available screening and interventions.[2]

The concept of a duty to warn implies that physicians should take reasonable steps to warn an identifiable third party of a serious and imminent threat of harm.[3] The duty to warn is well established in cases where a patient threatens to cause serious bodily harm to a third party,[3] or has an infectious disease.[4-6]

Though case law applying the duty to warn to third-party relatives of patients with genetically inheritable diseases is sparse, two cases demonstrate distinct lines of thinking on this issue. First, in a 1995 case called *Pate v Threlkel*,[7] the Florida Supreme Court ruled that a physician's duty to warn about a cancer predisposition syndrome was satisfied by educating the patient about familial cancer risk. Genetic risks were distinguished from infectious diseases because a patient's genetic risk was already present, and disease onset was not imminent or preventable.

The following year, in *Safer v Estate of Pack*,[8] a New Jersey appellate court advocated a broader duty to warn. Here the court found that a physician's duty to warn extends to identifiable third parties known to be at risk of avoidable harm from a genetically transmissible condition, and that physicians should take "reasonable steps" to warn at-risk family members. In expanding the duty to warn the court found "no essential difference" between the type of genetic threat at issue in the case and "the menace of infection, contagion, or a threat of physical harm."[8]

The policies of both ASCO and the American Medical Association (AMA) are consistent with the Florida court's opinion in *Pate*. The AMA Code of Medical Ethics emphasizes the overriding importance of patient autonomy and confidentiality, but states that physicians have a clear responsibility to inform patients with genetically linked diseases with information about the mode of inheritance, associated risks, and appropriate screening and intervention recommendations for at-risk family members.[9,10] ASCO's 2003 policy statement notes that any obligations to relatives who may be at risk are best fulfilled by communication to the person undergoing testing that emphasizes the importance of sharing this information with family members so that they may also benefit.[11]

The American Society of Human Genetics policy statement on disclosure of genetic information is consistent with those of ASCO and the AMA, in that it does not extend the duty to warn to genetic risks. However, the American Society of Human Genetics statement recognizes that it may be acceptable for health professionals to breach genetic confidentiality under certain conditions, including if the harm is serious and foreseeable, the at-risk individuals can be identified, the disease is preventable or treatable, or early monitoring is medically accepted to reduce risk or avert harm.[9] Factors including variable penetrance levels, age of disease onset, disease severity, and

advances in screening and preventive medicine make these criteria difficult to apply to genetic risks.[12]

Given the ambiguity in existing case law and recommendations by professional organizations, physicians may wonder if they should warn at-risk family members to shield themselves from liability. However, this approach may not be consistent with physicians' ethical obligations to patients, and could be at odds with state and federal privacy laws.[13-15] Furthermore, forcing patients to notify at-risk relatives could make them more reluctant to obtain genetic counseling services, and delay or decrease the benefit of risk-reducing interventions.[16] Repeated reminders and creative outreach measures can result in successful notification of the patient's relatives regarding their inherited cancer risk. For example, in a case involving two estranged family members, the genetic counselor encouraged the patient to inform her family priest about her genetic risk. The priest was ultimately able to facilitate the sharing of genetic risk information with appropriate family members.

Generally, it is appropriate for physicians to encourage patients to share genetic information with relatives so that they can benefit from preventive interventions. Physicians can raise the issue of disclosure during pretest genetic counseling in a manner that is deliberate but not coercive.[9,10] Additional best practices include documenting the patient's willingness and ability to identify at-risk relatives and providing those relatives with specific referrals to cancer genetic resources.

Another concern raised in this vignette is the perceived risk of discrimination based on genetic information, which in this case served as a barrier to communication of genetic information. Though there are few documented cases of genetic discrimination in insurance or employment, the fear of genetic discrimination remains high.[17-20] If not assured of their protection, people may shy away from genetic testing, genetic-based clinical trials, and cutting-edge genomic treatments. To ensure that people take advantage of genetic technology, President Bush recently signed into law the Genetic Information Nondiscrimination Act of 2007 (Pub. L. No. 110-223), which protects against discrimination based on genetic information when it comes to health insurance and employment.[21,22] Genetic information is already protected by stringent Standards for Privacy of Individually Identifiable Health Information (Privacy Rule), promulgated under the Health Insurance Portability and Accountability Act of 1996,[13,14] as well as the Americans with Disabilities Act,[23] which provides significant privacy and nondiscrimination protections for people in the employment sector. In addition, many states have genetic nondiscrimination laws that address insurance and employment issues.[24,25]

Conclusion

As genetic technology becomes more advanced and more accessible, the challenges inherent in the identification of and communication regarding inherited cancer risk will become more apparent. Continued discussion by practitioners and consumers regarding the ethical and legal challenges posed by genetic and genomic testing will be necessary to inform the most responsible application of these technologies to the practice of oncology. Practitioners will need to closely monitor updated ethical guidelines and emerging case law in this area. Throughout this process it will be important to protect the framework of the physician-patient relationship, which is premised on trust, confidentiality, and a commitment to facilitating patient autonomy.

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