Genetic testing and breach of patient confidentiality: law, ethics, and pragmatics

Howard Minkoff, MD; Jeffrey Ecker, MD

BACKGROUND AND OBJECTIVE

The Human Genome Project has transformed what had once seemed scientific fantasy into a realistic expectation for 21st-century medicine. However, voices of caution have suggested that attaining genetic transparency (ready access to all genes of all people) will be a Faustian bargain. Physicians will soon confront ethical dilemmas that recently seemed hypothetical, such as choosing offspring with a particular set of traits.

The confidentiality of genetic information can also create an ethical dilemma. An individual identified with a predisposing gene has kindred who are at increased risk (relative to the general population) of carrying that same gene. If appropriate interventions could reduce the proband’s likelihood of progression to disease, it would seem proper medical and ethical practice to inform kindred of their genetic status. Yet a proband might not want kindred to know that she or he carries a deleterious gene. We address legal, practical, and ethical considerations that should inform physicians’ responsibility in such circumstances.

Legal precedents and organizational guidelines

There are circumstances in which the physician’s duty to maintain a confidence must be balanced against competing obligations. In Pate v Threkel, the Supreme Court of Florida found that even if genetic information might be vital to a family member, it would be untenable to establish a requirement that the physician seek out and warn those at peril. Rather the court held that a physician could fulfill his or her duty to warn by telling his or her patients of the need to inform their biologic kin.

In Safer v Estate of Pack, the daughter of a man with multiple polyposis, a disease notable for being linked to a gene with 100% penetrance, developed the same condition, which progressed to metastatic colorectal cancer. She alleged that the physician had known the hereditary nature of the condition yet failed to warn her. In their ruling, a New Jersey appellate court recognized a “physician’s duty to warn those known to be at risk from a genetically transmissible condition.” Both the Pate and Safer courts seem to agree on a duty to warn, although they differ markedly in defining the standard for considering the obligation fulfilled.

Organizations involved in genetic care and counseling have proposed guidelines for the disclosure of genetic information to the relatives of those tested. The approach suggested by the American Society of Human Genetics varies, depending on the degree and immediacy of risk faced by kindred. According to the society, disclosure would be acceptable if “the harm is likely to occur, and is serious, immediate and foreseeable.”

The President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research proposed criteria that would make disclosure of genetic information appropriate in the absence of the proband’s consent. The criteria were that: (1) efforts to elicit voluntary disclosure by the proband have failed, (2) there is a high probability that harm will occur if disclosure is not made and intervention can avert that harm, (3) the harm will be serious, and (4) efforts are made to limit disclosed information to genetic information needed for diagnosis and treatment.

Ethical considerations

These court rulings and organization guidelines echo past efforts to enlist physicians in efforts to protect the public and to fulfill a perceived “duty to warn.” Among recent court cases that have been identified as supporting such precedents is Tarasoff v the Regents of California. The case involved a campus psychologist who failed to warn a woman of a threat to her life made by his patient (her boyfriend). The court, which addressed the case after her murder, held that the protective privilege ends where public peril begins.

Ultimately how one works through these issues from an ethical perspective may depend on whether the problem is viewed through the prism of principle-based or communitarian ethics. In principle-based ethics, respect for autonomy, beneficence, nonmaleficence, and justice are considered and balanced. Although the balance is a complex calculus, respect for autonomy is usually considered the first among the 4 principles. This approach seems to weigh on the confidentiality side of the balance. The bias in fa-
...the practitioner has an affirmative obligation to raise this issue and to go beyond the traditional nondirective model of genetic counseling in leading the proband to optimal health values with regard to disclosure. If despite direct counseling the patient refuses to divulge information that could be of vital interest to kindred, practitioners must consider their own obligations to the proband, to kindred, and to society.

In almost all circumstances, the practitioner’s autonomy-based obligations to the proband, his or her recognition of confidentiality as a pillar of medicine, and the practical impediment of needing to contact individuals who can generally be known only through the graces of the proband should preclude nonconsented disclosure. In the rare instance in which the data are vital, when a high likelihood of progression to serious illness and a high likelihood that interventions can modify the probability of progression or magnitude of disease are noted, and the “at-risk” individual is known and can be contacted, legislation and medical guidelines should permit providers to consider the competing interests discussed in earlier text and decide whether to disclose the information accordingly.

**Clinical Implications**

- Genetic information pertaining to a given individual often has implications for kindred.
- Physicians should discuss those implications and the proband’s own “duty to warn” at the time of initial genetic counseling.
- In the rare circumstance in which an individual refuses to disclose information that would allow a useful intervention, physicians must balance patient confidentiality against the potential harm that would befall kindred who were left unaware.

**Conclusion**

Consideration of disclosure should not be an afterthought in the process of genetic counseling and consent for testing.