

# Virtual Mentor

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## HEALTH LAW

### Genetic Diseases and the Duty to Disclose

Shawneequa Callier, JD, MA, and Rachel Simpson

Amy, a long-term patient of yours, has been diagnosed with a bipolar disorder that has a hereditary component likely to manifest itself in some of her relatives. While efforts to identify specific susceptibility genes are still underway, disclosure of her test results to siblings and children, followed by careful monitoring, could improve the future health of her family. Worried about the possibility of estrangement, however, Amy says that she is unwilling to warn her at-risk relatives of the genetic link to bipolar disorder that they may share. As a clinician, what are your duties to Amy and her family, and how are they affected by patient confidentiality requirements?

With increased use of personalized genomic medicine (PGM)—individualized care that incorporates patients’ genetic profiles for treatment and diagnosis purposes—scenarios like the one described will be more common [1]. Using genetics to diagnose and treat medical conditions raises significant privacy and genetic discrimination concerns because diagnoses of gene-related health conditions may have implications for those related to the patient. The law, however, has been inconsistent in its guidance to physicians regarding their duties to nonpatient family members, especially when the implications of patients’ genetic test results are unclear.

Generally, physicians only have duties to their patients, and, unless a patient expressly consents to disclosure or a law requires it, they are obliged to hold patients’ medical information in the strictest confidence [2]. This professional obligation is intended to encourage patients to communicate fully and candidly with their doctors [3]. If they can trust that their communications will remain confidential, the argument goes, patients will be more forthcoming about behaviors and history that might influence treatment strategies [3]. Exceptions to confidentiality exist, primarily to prevent a contagious threat to the public’s health from communicable disease [3], to prevent foreseeable, serious risk to an identifiable victim [3], and when violence or abuse is the suspected cause of a patient’s injury. The Health Insurance Portability and Accountability Act requires potential danger or imminent threat for disclosure of medical information to third parties [4].

With personalized genomic medicine, the threat to family members is rarely imminent and the level of foreseeable harm is often difficult to predict. Further, PGM complicates what it means to act in the best interest of the patient. Variations in family dynamics, for instance, can quickly and dramatically transform the

fulfillment of professional duty in one situation to a questionable act in another. Unlike traditional medical test results, genetic test results often provide only probabilistic information rather than a clear diagnosis or definite prediction of disease. Whether relatives should be warned of hereditary conditions when there are no means of prevention, treatment, or cure is unclear, and there is little support for warning underage family members of adult-onset conditions [5]. Further, patients' relatives have a "right *not* to know" about their genetic makeup, so informing them might interfere with their autonomy, in addition to breaching the patient's confidentiality [6].

### **Case Law**

In considering physicians' duty to warn at-risk family members of possible harm from genetic variations, the courts provide limited and conflicting guidance.

In a 1995 case, *Pate v. Threlkel*, the plaintiff, Mrs. Pate, inherited medullary thyroid carcinoma from her mother and sued her mother's physician for negligent failure to warn the mother that her children might inherit the cancer risk [7]. Mrs. Pate alleged that the physician "knew or should have known" of the risk to his patient's children and had an affirmative duty to recommend immediate testing for the patient's children. Had she been warned, Mrs. Pate argued, she would have sought preventive treatment for the disease at an early stage in its development. The court ruled in favor of the physician that "in any circumstances in which the physician has a duty to warn of a genetically transferable disease, that duty will be satisfied by warning the patient" [7]. In this instance, the court did not impose a duty upon the doctor to warn a third party, but merely to encourage the patient to warn her at-risk relatives.

One year later (1996), a New Jersey appellate court came to a different decision. In *Safer v. Estate of Pack*, the plaintiff's father had died of multiple polyposis, an inherited condition that can develop into cancer if it is left untreated [8]. Because the plaintiff, Mrs. Safer, was a child when her father died, she only learned of her predisposition to developing the disease when she was diagnosed with multiple polyposis herself in adulthood. The plaintiff sued her father's physician, alleging a duty on the part of the doctor to warn at-risk relatives of the possibility that they might develop this treatable condition. The *Safer* court ruled that "the duty to warn might not be satisfied in all cases by informing the patient." Sometimes, the decision went on to say, the physician might have to resolve the "broader duty to warn and...fidelity to the expressed preference of the patient that nothing be said to family members" about the disease [8]. These holdings and case law in general are inadequate to apply to the gamut of scenarios in which physicians could apply PGM routinely.

Returning to Amy's condition, we have little knowledge about her relatives' interest in or understanding of genetics. With a specific genetic susceptibility test still in development, warning Amy's relatives could potentially *cause* "avoidable harm," especially if the clinician is ill-equipped to properly advise the family. Encouraging Amy to inform her at-risk relatives that she has an inherited bipolar disorder (as

urged in the *Pate* decision) may be appropriate, but creative and practical solutions for the family may be needed, such as directing them to resources and people who can explain the health, emotional, and discrimination risks for those who may wish to seek confirmatory testing.

### **An Ethical Approach**

While some scholars have called for the adoption of a system in which genetic information is shared among family members by default, others prefer to quantify the levels of genetic risk and probability of harm on a case-by-case basis [1]. Until legal agreement is achieved on when and how to make sensitive disclosures to at-risk family members, clinicians can at least fulfill their duty to fully educate their patients about the meaning and scope of their diagnosis [9].

For instance, patients should understand that guidelines on the duty to inform at-risk relatives of possible genetic conditions differs among professional organizations. For potentially life-threatening genetic mutations, the Institute of Medicine recommends disclosure when the following conditions are met: (1) irreversible or fatal harm of the relative is highly likely, (2) attempts to elicit voluntary disclosure fail, (3) disclosure will prevent harm, (4) the harm resulting from the disclosure is less than the harm that may result from failure to disclose, and (5) there is no other way to avert the harm [10]. In such cases, the disclosure should be limited to the information necessary for diagnosis or treatment of the relative [10]. In nonfatal cases, a clinician's duty may be fulfilled by encouraging patients to communicate with relatives [11] or providing the name of a counselor who specializes in such discussions. The *AMA Code of Medical Ethics*, however, does not construe finding and notifying family members as a physician's duty, though it does recommend that physicians inform patients in advance of what they expect them to disclose to their families and be available to assist in this communication [9].

### **Conclusion**

Medical care tailored to the genomic makeup of an individual can reduce adverse drug reactions, improve the efficacy of treatment, and help patients better comprehend gene-environment reactions that influence individual health. Because an important genetic mutation can affect family members, however, concerns about confidentiality are likely to increase as personalized medicine becomes a more widely used tool in clinical management, and clinicians' duties may widen to include at-risk family members. Medical staff should be conscientious about their patients' potential needs for genetic counseling (given by the clinician or a qualified genetic counselor) and be ready to advise patients on communicating their diagnoses to family members.

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Shawneequa Callier, JD, MA, is an assistant professor in the Department of Clinical Research and Leadership at the George Washington University School of Medicine and Health Sciences in Washington, D.C.

Rachel Simpson is a graduate student pursuing a master of public health degree in epidemiology at the George Washington University School of Public Health and Health Sciences in Washington, D.C. She received a bachelor of science degree in cognitive science with a specialization in neuroscience from the University of California, San Diego.

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