GENETICS

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

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Medical uses of genetic information have multiplied over the last several years. When an individual is a carrier of a clinically important allele, their kindred are at increased risk of carrying the same allele and of sharing the consequent risk of disease. If there were an intervention that could modify the risk of progression to disease, then there would be a clear advantage to kindred to be so informed. However, some probands may resist divulging that information to kindred for any of a variety of reasons, including the potential for discrimination. In this article we will review the manner in which the courts and professional organizations have viewed the conflict between 1 individual's right to privacy and another's right to information that could potentially be life saving or life prolonging. We will then consider the ethics of this issue and suggest an approach that physicians should take when confronting it.

Key words: confidentiality, genetic information, kindred, proband

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The Human Genome Project has transformed what had only recently seemed scientific fantasy into a realistic expectation for 21st century medicine. The advent of an office accessible genome seems inevitable. Direct-to-consumer genetic tests are now being marketed[1] and the $1000 genome has already been heralded in the press.[2] At the same time, voices of caution have suggested that attaining genetic transparency (ready access to all genes of all people) will be a Faustian bargain. They have, for example, detailed the not inconsequential iatrogenic morbidity that may arise in the wake of genetic fingerprinting, focusing on the extremely large false-positive rates with which genetic prediction of disease will be saddled.[3] Similarly, ethical dilemmas that seemed purely hypothetical in the recent past, choosing offspring with a particular set of traits, for example, will undoubtedly confront physicians in the near future.

Genetic information can inform care in several ways. It may provide a diagnosis or signal predisposition to disease. An individual identified with a predisposing gene also has kindred who are at increased risk (relative to the general population) of carrying that same gene. If there were an opportunity to reduce the proband's likelihood of progression to disease with appropriate interventions, then it would seem proper medical and ethical practice to provide kindred with the chance to learn their own genetic status. However, a proband might not want the fact that she or he carries a deleterious gene to be known by kindred or may, for other reasons, be hesitant to share genetic information. As Hudson[4] recently noted, "Without comprehensive legal protections, the public fears genetic discrimination." That legal protection does not yet exist.

There may be particular reasons for concern. The proband's relative may also be his or her employer or have other reasons and opportunities to limit the prospects of the proband were she or he to learn that the proband was at risk for disease. Surveys have shown that more than 80% of respondents would not trust their employers with access to their genetic information.[4]

Whereas the question of whether a patient's right to confidentiality should ever be tempered by concerns about another individual or community has been addressed by professional societies and in the medical literature,[5] the question has not received similar attention in obstetrical journals, whose readership is often called on to provide genetic counseling. In this paper we address legal, practical, and ethical considerations that should inform physicians' responsibility in such circumstances.

Legal Precedents and Organizational Guidelines

Although the medical provenance of certain core ethical principles (eg, informed consent) is of relatively recent vintage,[6] the physician's duty to protect patient's confidences was present at the birth of medicine. The Hippocratic Oath contains the words, "Whatever I may see or learn about people in the course of my work or in my private life which should not be disclosed I will keep to myself and treat in complete confidence."[7] Many consider this sentence to be the heart of this oath, and a commitment that has been passed down through the centuries as an unadulterated obligation of physicians. Indeed, recent American Medical Association policy statements would, with a few specific exceptions, reinforce that belief, noting specifically that "a

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physician shall respect the rights of patients, colleagues, and other health professionals, and shall safeguard patient confidences and privacy within the constraints of the law."[8]

However, there are circumstances when the physician's duty to maintain a confidence has to be balanced against competing obligations, at least as adjudged by courts, legislation, and medical organizations. Several court cases have already commented on a physician's duty to inform kindred about genetic information, although their holdings have not been entirely consistent. In Pate v Threkel, the Supreme Court of Florida found that even if information might be vital to a family member, it would be untenable to establish a requirement for a physician to seek out and warn those at peril.[9] Rather, the court held that a physician could fulfill his or her duty to warn by working through the proband (ie, telling his or her patients of the need to inform their biologic kin).

Other courts have placed a heavier onus on the physician. In Safer v Estate of Pack, an appellate court in New Jersey recognized a "physician's duty to warn those known to be at risk from a genetically transmissible condition."[10] In that case the daughter of a man who had been diagnosed with multiple polyposis (a disease that is notable for being linked to a gene with 100% penetrance) developed that same condition, which then progressed to metastatic colorectal cancer. She alleged that the physician knew the hereditary nature of the condition yet failed to warn her. It is important to note that the decision in this case did not argue that a family member's right to genetic information always supersedes the proband's right to confidentiality. Rather, the court's ruling reinforces the importance of discussing with the proband their wishes with regard to data because the opinion found that, in the absence of specific guidance from that individual (ie, a refusal to give permission to divulge), it cannot be assumed that an obligation to kindred does not have a legal and/or moral base. In the end, both the Pate and Safer courts seemingly 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 also proposed guidelines for the disclosure of genetic information to relatives of those tested. The approach suggested by the American Society of Human Genetics (ASHG) varied, depending on the degree and immediacy of risk faced by kindred.[11] Although the guidelines encouraged voluntary disclosure, they also described circumstances in which providers should not accede to a proband's request to withhold information from a relative. Specifically, these guidelines stated that disclosure would be acceptable if "the harm is likely to occur and is serious, immediate, and foreseeable." The guidelines add that the at-risk relative must be identifiable and that there must be some existing intervention that could have a beneficial effect on the course or outcome of the genetic disease.

The President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research also proposed criteria that would make disclosure of genetic information, in the absence of the proband's consent, appropriate.[12] The specific criteria they cited were: (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

In acknowledging a set of conditions, as uncommon as they may be, under which confidentiality should be breached, 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." The enforced quarantine of Typhoid Mary, mandatory testing of pregnant women for syphilis, notification of potential crime victims,[13] and notification of partners of individuals with sexually transmitted diseases (STDs) are all precedents that recognize a community's right to be protected from hazard and that place responsibility for the execution of that right on the physician.

Among recent court cases that have been identified as supporting such precedents is Tarasoff v the Regents of California.[13] 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 the public peril begins. The Tarasoff case gained currency in medical circles during the early acquired immunodeficiency syndrome era when clinicians were concerned about their obligation to warn partners of individuals found to be human immunodeficiency virus (HIV) infected. Physicians, in the context of HIV, interpreted this ruling as a legal precedent that, de minimus allowed, and more strictly interpreted required, providers to warn sexual partners of HIV-infected patients of their potential risk from sexual exposure to the virus, even if it meant breaching confidentiality.

But genes are not infectious, and the analogy with HIV or other STDs may seem fallacious on face. However, a person's genome, like a person's HIV status, can speak to risks that others face, risks that physicians can help to mitigate. In the case of an STD, if the provider knows a specific sexual partner, then the Tarasoff decision might suggest that there is an obligation to give that individual the tools with which to avoid a potentially lethal disease (eg, to discontinue the relationship or to adopt safer sex practices).

Whereas in the case of a carrier of a dangerous gene the opportunity to avoid contagion by a gene is illusory, other important aspects of the construct hold. There still may be an identifiable individual who could be given the tools with which to avoid a potentially lethal disease. For example, the child of someone with polyposis has a 50% chance of inheriting that gene and would thereby be at high risk of developing a fatal disease (colorectal cancer). There are tools that can be used by that individual that might

avoid the inevitability of progression from genotype to metastatic phenotype. In this case, the fact that the risk of transmission of the relevant risk-vector is not "immediate" seems wholly irrelevant.

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 ethics or communitarian ethics.[14] 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 first among the 4 principles.[15] In vacuo this approach would seem to have its thumb squarely on the confidentiality side of the balance. The bias in favor of confidentiality would reflect a belief that it most directly protects the autonomy of the patient with whom the provider has an established fiduciary relationship.

In contrast, a communitarian approach would look beyond the singular relationship between the provider and patient to the many other relationships and shared values that are the basis of family and community.[14] In so doing, a greater deference might be ceded to the rights of kindred.

However, the choice is not merely between the individual and the community but also between community writ small (kindred) and community writ large (the public good from having a confidential medical system). In other words, even if the physical harm to the proband from a breach of confidentiality were inconsequential, the harm to the medical "system" could be substantial.

Confidentiality is a core component of the trust that is at the heart of the doctor-patient relationship, allowing patients to share facts and concerns that are concealed from all others. In protecting this relationship and in writing laws that recognize its sanctity, society has acknowledged its importance to the pubic health. To undermine that principle for the benefit of 1 or a small group of individuals, relatives of the proband, and, in many cases, for only a possible benefit at that, would risk weakening this vital aspect of medicine.

What is to be Done?

Thus, in theory due deference to autonomy as well as respect for the importance of confidentiality as a community virtue would argue persuasively for nondisclosure of genetic information to kindred without the express consent of the proband. In reality however, when the provider has identified or even has a relationship with the potentially affected kindred, adherence to this doctrine can be difficult. Balancing confidentiality against the concept of potential harm is one thing, balancing it against the health of a particular, known human being quite another. Some states have addressed this conundrum in the context of HIV status through legislation that is both permissive and protective to the physician who is contemplating a warning. For example, in New York if a physician chooses to notify an infected individual's sexual partner, he will be protected from prosecution, although he or she is permitted to nondisclose if circumstances (such as the probability of intimate partner violence), as perceived through the provider's own virtue-based ethics, do not justify disclosure.[16]

Practical Considerations

As described in previous text, professional organizations have promulgated recommendations for the handling of genetic information. However, although these recommendations can serve as ethical guideposts, in practice they are unlikely to have substantive effects. For example, the ASHG advocated disclosure when "harm is likely to occur, and is serious, immediate, and foreseeable."[11] Although this recommendation to disclose seems unequivocal, it also posits circumstances for its exercise that are rare at the current time: There are few genetic diagnoses that pose an immediate risk, and fewer still that can be substantively modified with an intervention.

However, it is becoming increasingly common for interventions to be available that can mitigate risk in probands and affected kindred. Those at risk for hereditary cancers may choose screening or prophylactic surgery to remove at risk tissues. Individuals with thrombophilias may be candidates for specific thromboprophylaxis if undergoing surgery. Diet, exercise, and cholesterol-lowering drugs may reduce morbidity in individuals at genetic risk for cardiovascular disease. Finally, couples at risk for transmitting a condition to their offspring may, if informed of the risk, choose to avoid reproduction.

Many of these ethical dilemmas may be obviated by engaging the proband in the disclosure process whenever possible. Many, if they understand the importance of disclosure, will do so voluntarily. The consenting patient should understand the possible consequences of carrying a particular gene for both themselves and their kindred. It is during this process that the physician can articulate the values that should animate a person's responsibility to others who might benefit from knowledge of some aspect of a relative's genome.[17] In fact, studies of individuals who carry genes that might have consequence for their kindred suggest that concern for family members is a salient feature of their decisions about being tested in the first instance.[18]

Some ethicists have suggested that counseling given to patients about their obligations to kindred should be directive. Macklin[19] discussed a genetic Miranda warning that would serve to inform patients of the provider's intention to notify kindred of relevant results should the proband decline to do so themselves. This seems appropriate if, regardless of the patient's wishes, the provider feels that he or she will be compelled to inform potentially affected relatives of the results. Theoretically such advance warning of how the practitioner perceives his responsibilities would allow the proband the opportunity to seek genetic testing elsewhere, in a setting in which anonymous testing is possible or in which other providers might have reached a different conclusion about the need to inform.

Although this approach would avoid the discomfort providers might feel if their patients refused disclosure after their results became known, it still leaves several ethical points to be considered. In the first instance there is no a priori reason to assume that genetic testing is

more deserving of Miranda warnings than any other type of testing that might yield information of health import to others. Thus, psychiatrists, physicians testing for STDs, and many other providers might be under an equal compulsion to share with their patients the limits they set to their oath of confidentiality. Second, there will be instances in which the physician confronts results before the Miranda had been proffered. For example, the genome may yield results coincident to those primarily sought but results that still have relevance. Or the provider may be called on to see a patient based on results of tests ordered by other providers who do not adhere to the same tenets of ethical genetics.

Perhaps of greatest concern with regard to the Miranda approach is what will happen to the individual who refuses to agree, before the fact, to disclose. That patient must then seek care elsewhere and will either find themselves again in the care of a provider who demands an acceptance of the duty to disclose or in the care of a provider who does not agree with the Miranda approach and who will not breach confidentiality under any circumstance. In the former case, the primary physician, in failing to provide an alternative source of care, has in essence obviated their patient's autonomy and their ability to avail themselves of important health information. In the latter, they have enabled the patient to pursue what the provider has determined to be an unethical course. In that case the patient's blood relatives might still be put at risk. Although the provider can comfort himself or herself in knowing that she or he was not the individual who failed to warn, he or she still must accept his or her role in helping the proband to find a way to endanger that relative.

Conclusion

The future will undoubtedly reveal an increasing number of instances in which the genetic make-up of a proband will be of health interest to others. Whereas the ethical physician will wrestle with the management of these occurrences, some things already seem clear. Consideration of disclosure should not be an afterthought in the process of genetic counseling and consent for testing. 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 directive 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, their 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 (a high likelihood of progression to serious illness and a high likelihood that interventions can modify the probability of progression or magnitude of disease) and the at-risk individual is known and contactable, legislation and medical guidelines should permit providers to consider the competing interests discussed in previous text and decide about disclosure accordingly.[16] ■

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