

Managing Familial Risk in Genetic Testing

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ABSTRACT

Increasing numbers of people are seeking genetic testing and uncovering information that directly concerns their biological relatives as well as themselves. This familial quality of genetic information raises ethical quandaries for physicians, particularly related to their duty of confidentiality. In this article, the American Medical Association's Council on Ethical and Judicial Affairs examines the informed consent process in the specific context of genetic testing, giving particular consideration to the handling of information that has consequences for biological relatives. Furthermore, it addresses the question of whether physicians' obligation to warn biological relatives ever should override the obligation to protect patient confidentiality.

INTRODUCTION

IN THE PAST, GENETIC INFORMATION has been sought primarily in the context of reproductive counseling. It is now being sought by individuals who wish to learn whether they have a predisposition to an adult-onset genetic condition (Collins, 1999). At the same time, increased exposure to health information in general, through the media and especially through the Internet, has heightened public attention to medical disorders, including genetic conditions (American Medical Association, 2001). Consequently, more people are obtaining individual genetic information that, unlike other medical information, directly concerns not only the tested individuals (proband), but their biological relatives (kindred) as well. This familial aspect poses new ethical quandaries for physicians by challenging the limits of medical confidentiality.

According to many commentators, the role of genetic testing in medicine has not fundamentally changed the responsibility of physicians to respect patient autonomy and act in their patients' best interests. However, according to others, it has raised new questions regarding physicians' responsibilities when an individual patient's genetic information reveals serious information that could be directly relevant to kindred's health (Andrews, 1997; Hakimian, 2000).

In this report, we examine the informed consent process in the specific context of genetic testing; other aspects of medical genetics such as gene therapy fall outside the scope of this report. We consider whether there are circumstances in which the familial quality of genetic information justifies the compromise of physicians' duty of confidentiality, which is central to the patient-physician relationship. We also address how physicians should handle patients' genetic information when it could be relevant to their biological relatives.

GENETIC INFORMATION AND CONFIDENTIALITY

In general, American Medical Association (AMA) ethics policy includes and is derived from principles that recognize physicians' duty to safeguard the confidences of their patients (Principle IV) to whom their responsibilities are paramount (Principle VIII) (American Medical Association, 2004). Physicians have a general duty to treat information acquired from the patient in the context of the patient-physician relationship as confidential. As stated in Opinion 5.05, "Confidentiality," "the physician should not reveal confidential communications or

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information without the express consent of the patient, unless required to do so by the law” (Council on Ethical and Judicial Affairs, 2004).

In the medical ethics literature, there is a spectrum of opinions about the stringency of confidentiality regarding genetic information. At one end of the spectrum, commentators focus on the sensitivity of genetic information and call for more stringent confidentiality measures to protect it (Annas *et al.*, 1995). Genetic testing has the potential to reveal medical risks to an individual. Such risks usually are highly uncertain because of uneven scientific validity and reliability of genetic testing findings, variable penetrance of genes, and absence of recognized interventions for some identified predispositions. Regardless of whether genetic information signals risks that are likely to materialize into an illness or disability, such information may warrant special protection for fear of discrimination and stigmatization.

Commentators at the opposite end of the confidentiality spectrum focus on the nonindividualistic or biological cohort ownership of genetic information and question the practice of asking physicians to withhold this information from potentially affected kindred (Wachbroit, 1993). Some of these commentators would argue that the familial quality of genetic information requires extending the obligation of physicians beyond the patient, to the wider circle of immediate biological relatives (Hayes, 1992; Wachbroit, 1993). The rationale behind this position is that when a physician’s knowledge of information pertains as much to the family as to the patient, a professional obligation is created that extends to other affected parties, especially when the physician already has a professional relationship with the patient’s biological relatives. Under this view, confidentiality’s basis in the claim of individual privacy is significantly compromised in the family context, where the information is at once individual and familial (Safer v. Estate of Pack, 1996). Beyond the circle of affected relatives, however, the physician’s obligation toward the confidentiality of patient information is considered to remain unchanged.

Finally, an intermediate perspective exists among those who reject genetic exceptionalism (Gostin and Hodge, 1999). The limits of confidentiality are no more and no less stringent than those that already exist for other kinds of medical information. As such, confidentiality is near absolute: Physicians have a duty to maintain the confidentiality of genetic information about a patient, save “certain exceptions which are ethically and legally justified because of overriding social considerations” (Council on Ethical and Judicial Affairs, 2004).

Informed consent

Genetic information poses some special challenges, due to its inherited, and therefore shared, nature (Geller *et al.*, 1997). Challenges can arise when the need to maintain an individual patient’s confidentiality and autonomy conflicts with an obligation to inform kindred of information that may be directly relevant to their health. The pretesting period offers health-care professionals the opportunity to educate and counsel their patients in an effort to prepare for test results, including findings that could have significant implications for biological relatives.

Before they can arrive at a voluntary, informed decision about whether to undergo genetic testing, individuals need to receive information regarding the overall risks and benefits associated with the procedure, including potential implications of

test results for them and immediate biological relatives. Patients need to understand why relatives may have a substantial interest in this information, as it may influence relatives’ decisions to seek treatment, reproductive decisions, or lifestyle choices. Patients also need to understand that some relatives may prefer not to know whether they are affected by a genetic condition, for reasons such as fear of social stigmatization, loss of insurance, or work-related discrimination.

Once individuals understand the consequences for themselves and for others of obtaining genetic test results, they can address another important step of the pretesting phase. Guided by a professional with special genetic expertise, they can begin to contemplate, before any information is uncovered, whether to invite biological family members to participate in the testing process—directly by undergoing testing, or indirectly, by sharing in the findings.

For individuals who are comfortable with notifying immediate biological relatives that they intend to undergo genetic testing, the pretesting period is an opportune time to communicate their intention to family members. Genetic specialists can help patients inform their relatives by providing them with educational materials aimed at lay audiences and by offering themselves or another appropriate person as a resource to discuss opportunities for counseling and testing.

Just as families can benefit from discussing advance care planning or other health-care matters before there is reason for concern, so they can benefit from exploring considerations that surround genetic testing to clarify an individual’s preferences prior to obtaining test results. Relatives’ early involvement provides ample time to offer family members genetic counseling to help them make informed decisions about whether they would like to share in the findings once they become available. Knowing their preferences at this stage will shield physicians and patients from the awkward situation of trying to determine whether to involve family members after important information has been uncovered. It will also help prevent the unfortunate circumstance in which results for which kindred are unprepared are accidentally communicated to them by the proband before the relatives can receive counseling or arrive at an informed decision about the willingness to share in the results. Physicians cannot predict how individuals will react to genetic information, but they have the ability and the responsibility to encourage people to seek appropriate counseling to prepare them to receive results from testing.

Some individuals will opt not to tell their immediate biological relatives that they are contemplating testing. Even when this is clear from the start, there are still benefits to discussing the possibility of discovering familial genetic risk before the testing is done. Physicians can help their patients reach a decision about what they will do with information that could be important to immediate biological relatives, and, if they should decide to share genetic information with affected relatives, to encourage family members to seek genetic counseling before receiving the information. Whether genetic information is reassuring, neutral, or alarming, relatives may not want to know it, but if they do, they deserve to be prepared.

Finally, addressing the implications to biological relatives of genetic information gives professionals an opportunity to identify circumstances under which they would expect patients to notify biological relatives of the availability of information related to risk of disease (McKinnon, 1997). The use of what one

bioethics scholar coined the “genetic Miranda warning” gives the patient the opportunity to decline testing from a physician whose conditions seem unacceptable (Macklin, 1992).

An adequate informed consent is likely to avert most situations that could conflict the physician between the competing obligations to respect the patient’s confidentiality and to warn third parties of potential harm. When patients and physicians discuss the patient’s intended uses of genetic information during the pretesting phase, they will almost certainly identify any fundamental disagreements regarding the circumstances in which the physician would expect the patient to notify kindred that information related to risk for disease was available. Referral of the patient to another health professional might be indicated if such differences weaken the patient–physician relationship.

Disclosure of familial risk

After testing has occurred, there may be instances in which, despite a satisfactory informed consent process, patient and physician find themselves at odds regarding who should share in the information revealed by test results. Physicians should make themselves available to assist patients in communicating with relatives to discuss opportunities for counseling and testing. In breaching patient confidentiality against the patient’s will, the physician would be giving more weight to the health interests of a third party than to the patient’s interest, thus compromising a core constituent of the patient–physician relationship.

The American Society of Human Genetics (ASHG), in *Professional Disclosure of Familial Genetic Information*, warns that social, psychological, and financial harms as well as discrimination and stigmatization can accompany genetic findings. Failure to disclose to a patient’s affected family members the availability of genetic testing results can also lead to harm, particularly when knowledge could result in avoidance, treatment, or prevention of a genetic condition or in significant changes to reproductive choices or lifestyle (American Society of Human Genetics, 1998).

ASHG guidelines for familial disclosure are related to the magnitude of harm that may be incurred. Accordingly:

Disclosure should be permissible where: attempts to encourage disclosure on the part of the patient have failed; the harm is highly likely to occur and is serious, imminent, and foreseeable; the at-risk relative(s) is identifiable; and the disease is preventable, treatable, or medically accepted standards indicate that early monitoring will reduce the genetic risk. [. . .] The harm from failing to disclose should outweigh the harm from disclosure (American Society of Human Genetics, 1998).

The standard these guidelines establish with their requirement that harm be likely to occur, serious, imminent, and foreseeable is so stringent that it may only exist as a theoretical requirement that is unlikely to be met in practice. Indeed, in light of what is known of human genetics, no genetic test currently can result in the diagnosis of a condition with a high likelihood of causing imminent harm. For practical purposes, then, the ASHG’s guidelines would not allow a physician to breach patient confidentiality by notifying immediate biological relatives of genetic information that might impact their health. On balance, the guidelines suggest that the harm potentially caused by

the disclosure of information to relatives exceeds the harm potentially caused by withholding such information.

The President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research (1983) arrived at similar, but more measured, conclusions when it stated that disclosure without the patient’s consent is only justified if:

1. Reasonable efforts to elicit voluntary consent to disclosure have failed;
2. there is a high probability both that harm will occur if the information is withheld and that the disclosed information will actually be used to avert harm;
3. the harm the identifiable individuals would suffer would be serious; and
4. appropriate precautions are taken to ensure that only the genetic information needed for diagnosis and/or treatment of the disease in question is disclosed” (President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, 1983).

The Commission’s conclusions were less stringent than the ASHG’s in that they did not require the harm from withholding information to be imminent. Still, the requirements they established, especially those included in item 2, would justify breach of confidentiality only in very rare circumstances.

Case law

Case law also addresses physicians’ competing obligation to be mindful of the health-care interests of patients’ immediate biological relatives, when results reveal that family members are at risk for serious genetic conditions that could be avoided or treated with timely intervention. In *Pate v. Threkel* (Pate v. Threkel, 1995), the court held that in the exceptional circumstances that might warrant disclosure of genetic information to family members, it would often be too difficult and impractical for physicians to seek out and notify a patient’s immediate family relatives. The court found that a physician’s duty to warn about the transferability of a genetic condition would be satisfied by educating the patient about the implications of the information for affected relatives.

In the case of *Safer v. Estate of Pack* (Safer v. Estate of Pack, 1996), a woman brought suit against her deceased father’s physician for failing to warn her of her hereditary risk for multiple polyposis. The case was initially dismissed at the trial court level, on the basis of the judge’s conclusion that there was no patient–physician relationship between the woman and the physician. The appellate court, however, recognized a physician’s duty to warn immediate biological relatives who might be adversely affected by nondisclosure of avoidable risk from genetic causes. The appellate court emphasized that a physician could identify and warn at-risk relatives without too much difficulty, thereby helping them avoid substantial future harm.

Both cases recognized a physician’s duty to warn, although they arrived at very different conclusions about what warning might entail. One case took the approach that a physician’s duty to warn is limited to warning the patient of the familial implications of genetic testing—a step this Report recognizes as a requirement of the informed consent process. The other case demands more of the physician who, within reasonable limits, must identify affected family members to warn them of their at-risk status. The latter model is more problematic for both the patient and the physician, in that patient confidentiality is

breached. It also places a burden on the physician to try to locate immediate biological relatives.

CONCLUSION

Individuals who contemplate undergoing genetic testing must receive adequate education and counseling from a genetic specialist as part of the process of informed consent. Before deciding to have the test, individuals should understand the consequences of the information, both for themselves and for their biological relatives, and the possibility of inviting family members to be part of the testing process. Before they can communicate any of these details to patients accurately and thoroughly, many physicians will need to become more educated about the role of genetics in medicine and specific conditions for which they offer testing (National Coalition for Health Professional Education in Genetics, 2001).

Before testing occurs, individuals must be informed of circumstances under which their physician would expect them to communicate the availability of important test findings to biological relatives. In the spirit of respecting the kindred's right not to know or to be informed of results only after adequate counseling, physicians should make themselves or an appropriate professional available to assist in notifying relatives. If the patient and the physician identify fundamental disagreements prior to testing, it may be preferable for the physician to transfer the care of the patient to another health care professional.

RECOMMENDATIONS

1. Physicians have a professional duty to protect the confidentiality of their patients' information, including genetic information.
2. Pre- and post-test counseling of the patient must include implications of genetic information for patients' biological relatives. At the time patients are considering undergoing genetic testing, physicians should discuss with them whether to invite family members to participate in the testing process. Physicians also should identify circumstances under which they would expect patients to notify biological relatives of the availability of information related to risk of disease. In this regard, physicians should make themselves available to assist patients in communicating with relatives to discuss opportunities for counseling and testing, as appropriate.
3. Physicians who order genetic tests should have adequate knowledge to interpret information for patients. In the absence of expertise in pretest and post-test counseling, physicians should refer patients to an appropriate specialist.
4. Physicians should encourage genetic education throughout a medical career.

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