

ASHG STATEMENT Professional Disclosure of Familial Genetic Information

The American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure

Genetic conditions, defined as changes in a gene or in a group of genes, are often caused by inheritance of a familial disease gene. Accordingly, genetic information about an individual reveals genetic-risk information about both the individual and other family members. This personal—yet simultaneously familial—information raises new and profound questions about health-care professionals' legal and moral obligations to disclose genetic information to at-risk relatives (Andrews 1997a).

The present statement focuses on the potential for conflict to arise within the health-care professional–patient relationship if the patient refuses to warn at-risk relatives about relevant genetic information. It maintains that genetic information should be considered as medical information, albeit with special concerns and implications. As such, the legal and ethical norm of patient confidentiality should, as a general rule, be respected. Only in exceptional cases is a health-care professional ethically permitted to breach confidentiality. However, a health-care professional ought to be legally privileged—that is, given a discretionary right to disclose genetic information to at-risk relatives, without incurring liability—provided that certain conditions are met. Finally, and at a minimum, health-care professionals have an ethical duty to inform patients, both prior to testing and on receipt of results, that the information obtained may have familial implications. The health-care professional (family physician, geneticist, genetic counselor, or nurse) should consider the points outlined below when deciding whether to disclose genetic information to a patient's at-risk relatives.

I. Points to Consider

A. The General Rule of Confidentiality

Genetic information, like all medical information, should be protected by the legal and ethical principle of

confidentiality. As a general rule, confidentiality should be respected. In the context of medical information, privacy rights translate into protection of personal data, affirmation of confidentiality, and freedom of choice. However, the principle of confidentiality is not absolute, and, in exceptional cases, ethical, legal, and statutory obligations may permit health-care professionals to disclose otherwise confidential information.

B. Exceptional Circumstances That Permit Disclosure

1. *Disclosure should be permissible where attempts to encourage disclosure on the part of the patient have failed; where the harm is highly likely to occur and is serious and foreseeable; where the at-risk relative(s) is identifiable; and where either the disease is preventable/treatable or medically accepted standards indicate that early monitoring will reduce the genetic risk.* If these conditions are met—that is, if the genetic information reveals that family members are at a substantially higher risk of suffering from a serious and otherwise undetected genetic disorder, and if prevention or treatment is available—the health-care professional may warn the at-risk family members. For diseases that are neither treatable nor preventable, disclosure would not be permissible. At-risk relatives possess genetic relatedness, are identifiable, include siblings and their children, and may also extend to identifiable parents, cousins, aunts, and uncles whom the health-care professional can reasonably contact.

2. *The harm that may result from failure to disclose should outweigh the harm that may result from disclosure.* Failure to warn may lead to irreparable harm if opportunities for avoidance, treatment, or prevention of the genetic condition are thereby limited. The harm that may arise from nondisclosure should outweigh the potential psychological, social, financial, and discriminatory harm that may arise from disclosure.

C. Ethical Duty to Inform Patients about Familial Implications

At a minimum, health-care professionals should be obliged to inform patients about the implications of their genetic test results and about the potential risks to their family members. This duty to inform the patient about

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familial implications, both prior to genetic testing and again if the patient refuses to communicate results, is paramount. It is presumed that most patients, provided with the proper information, will inform their relatives of potential risks so that early monitoring, detection, and treatment are available to them.

II. Background

A. Ethical Frameworks for Disclosure of Otherwise Confidential Information

There are four ethical positions regarding a health-care professional's duty or privilege to warn at-risk relatives about genetic information. In the first paradigm, confidentiality is absolute, and all medical information is strictly private. Although the health-care professional may inform the patient about implications for at-risk relatives, confidentiality prevents the health-care professional from disclosing any genetic information to relatives; the health-care professional has a duty *not* to breach confidentiality. From this viewpoint, it would be unethical to do so.

Alternatively, the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research (1983) proposed that health-care-professional disclosure to at-risk family members should take place only when (1) reasonable efforts to elicit voluntary consent to disclosure have failed; (2) there is a high probability that harm will occur if the information is withheld, and the disclosed information will actually be used to avert harm; (3) the harm that would result to identifiable individuals 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. These ethical guidelines do not imply a legal duty to warn; they simply set out circumstances in which the commission believes it would be permissible to do so. The Institute of Medicine Committee (1994), in its report on the assessment of genetic risks, adopted similar language.

A third possible approach involves warning the patient, before genetic testing, about the circumstances that would result in disclosure of genetic information to other family members, regardless of the patient's intentions to disclose (Macklin 1992). In this way, arguably, the health-care professional-patient relationship would not be jeopardized, as long as the health-care professional assures the patient of the presumption of confidentiality and outlines the exceptions to that presumption, prior to testing.

Finally, the duty to warn could be considered an ethical duty that might eventually become obligatory rather than permissive. Indeed, absence of explicit legal regu-

lations does not translate into an absence of duty. An ethical duty can become a professional norm of practice, and it may become the legal standard, as long as there are no important countervailing policy issues.

B. The Duty to Warn under Law

Statutes and legislation, which protect the confidentiality of medical/genetic information in general, permit health-care professionals to disclose, in exceptional cases, otherwise confidential information, without incurring liability. Physicians are required, on the basis of public-policy interests, to report, to the appropriate authorities, communicable diseases, gunshot and other wounds, and evidence of child abuse and neglect.

U.S. case law dealing with the general duty to warn identifiable third parties of a threat of violence (*Tarasoff v. Regents of the University of California*, 551 P. 2d 334 [CA 1976] [en banc]) found that a duty to warn is likely to exist if (1) the physician has a *special relationship* with either the person who may cause the harm or the potential victim, (2) the potential victim or person at risk is *identifiable*, and (3) the harm to the victim is *foreseeable* and *serious*. Such a duty has been distinguished from a possible duty to warn at-risk relatives about potential genetic risk, insofar as it is the patient's actions that are likely to harm others in the former case, whereas in the latter case the patient is not putting relatives at risk by simply carrying the gene mutation—the relatives already either have or do not have the mutation (Suter 1993; Park and Dickens 1995)).

More specifically, a health-care professional's duty to warn in the context of genetic information was recently considered by two U.S. courts. These cases may indicate an increasing trend toward disclosure: physicians were held to a duty to warn patients about familial implications (*Pate v. Threkel*, 661 So. 2d 278 [FL 1995]), and, further, they were held to a duty to warn relatives known to be at risk (*Safer v. Estate of Pack*, 677 A 2d 1188 [NJ Super A.D. 1996]), irrespective of potential conflicts between the duty to warn and the obligation to protect confidentiality.

C. International Trends and Positions

Although the majority of foreign jurisdictions (e.g., World Medical Association, World Health Organization, Council of Europe, Nuffield Council on Bioethics, Health Council of the Netherlands, and Privacy Commissioner of Australia) maintain that confidentiality must be ensured and protected, the majority are also in favor of limited disclosure of genetic test results (without the consent of the patient) in cases where the harm to at-risk relatives is grave and imminent and where the disclosure of information could result in effective inter-

vention. Only a few jurisdictions (Norway, Swiss Academy of Medical Sciences, and possibly France's National Ethics Committee) maintain that confidentiality is absolute and that the patient's wishes with regard to non-disclosure must be respected at all times.

III. Summary

Genetic information should be considered as medical information. However, genetic information is both individual and familial in nature. This raises conflicts between the duty to protect confidentiality and the duty to warn. Yet ethical, legal, and statutory exceptions limit the principle of confidentiality and, in specific and very limited circumstances, permit disclosure. It is clear that a health-care professional has a positive duty to inform the patient about the potential genetic risks to the patient's relatives. Moreover, where the harm is serious (a concept that defies exact definition and must be determined on an ad hoc basis [Knoppers et al. 1995]) and likely, and where prevention or treatment is available, the health-care professional may have a privilege to warn at-risk relatives, irrespective of the patient's wishes. Although this position is in line with the emerging international trend, the ethical duty of health-care professionals to warn patients' at-risk relatives will undoubtedly be the topic of future debate.

IV. Discussion

Genetic conditions are often caused by inheritance of a familial disease gene. Accordingly, genetic information about an individual reveals genetic-risk information about both the individual and other family members. This personal—yet simultaneously familial—information raises new and profound questions about health-care professionals' legal and moral obligations to disclose genetic information to at-risk relatives (Andrews 1997a, 1997b).

Given the complex, nonindividualistic nature of genetic information, some ethicists have maintained that it is "vital to recognize that hereditary information is a family possession rather than simply a personal one."¹ Yet if genetic information is treated as "family property," the traditional boundaries, definitions, and obligations of the health-care professional–patient relationship would be extended to include family members, leaving the health-care professional to deal with the potential conflict between the best interests of the traditionally defined patient and the best interests of the newly defined

"patients."² The inherent limitations of test results to predict the onset, severity, or complexity of a disorder complicate even further the medical, legal, and ethical issues that surround disclosure of genetic information to at-risk relatives. Like all medical conditions, genetic conditions rarely exhibit homogeneity in terms of how the disease process manifests itself among affected individuals. Since some genetic conditions are caused not by one but by several genes, the combination of individual variations produces an even more complex set of potential clinical outcomes and often leads to more unknowns than definitive predictions (Abbott 1996; Beardsley 1996).

Must the uniqueness of genetic information be established to justify a breach of confidentiality in the health-care professional–patient relationship and, thereby, to allow disclosure of genetic-risk information to relatives? Genetic information has been described as unique not only because it is both individually identifying and trans-generationally familial, but also because it implies probabilistic risk information. Advocates of such genetic exceptionalism³ have urged that genetic testing warrants additional protections because the information it yields can result in social discrimination and stigmatization (particularly in the areas of insurance and employment). While this genetic-exceptionalism argument maintains that genetic information is sufficiently distinct from other health-related information to warrant special privacy protection, the ASHG subcommittee maintains the more commonly held view that genetic information should be considered part of mainstream medical infor-

² Gevers (1988) presents the notion of genetic information as family property as a possible alternative to health-care professional–patient confidentiality but ultimately rejects this approach, arguing that "[t]he infringements of the principle of confidentiality would be potentially unlimited, partly because it may be impossible to draw the line between medical information that is relevant to genetic counselling and information that is not relevant, and partly because in the future ever more diseases will be found to contain hereditary components." Contra: Wertz et al. (1995), who adopt this family-property concept; see also Wachbroit (1989, 1993), who suggests a family-health model that contemplates the physician's patient as the entire family; "family" is understood to refer to a genetic network rather than a social institution. Therefore, the physician's duties pertain to the genetic family as a whole. de Wachter (1997) notes that "privacy seen as a family matter should be considered." He further notes the attention paid by the United Nations Educational, Scientific and Cultural Organization (Unesco) to various concepts of privacy among societies, and he points out that "the duty of the health professional is to secure the appropriate privacy for genetic information that is laid down by the norms of a particular society."

³ See, for example, Annas et al. (1995), who believe that genetic information should be accorded special status and who note that "genetic information is uniquely powerful and uniquely personal, and thus merits unique privacy protection."

¹ Wertz et al. (1995) suggest that, at the level of the person, genetic information, although individual, should "be shared among family members" as a form of shared familial property.

mation, albeit with special concerns and implications.⁴ Even those who believe that genetic information cannot be easily distinguished from other medical information note that “genetic information does not have to be completely unique in order to warrant special protection”; it simply has to be treated as “distinctive and especially sensitive.”⁵ Moreover, genetic exceptionalism only strengthens the notions of genetic determinism and genetic reductionism (Murray 1997).

The contagious-disease model, often cited in discussions of potential duties to warn at-risk relatives, is not an ideal paradigm for the disclosure of genetic information. First, genetic conditions are inherited—transmitted vertically through succeeding generations—and the connections are solely dependent on biological relations. Conversely, contagious disease is generally transmitted horizontally (save for parent-offspring transmission), and its impact on others occurs through some form of contact. Finally, contagious disease is controlled by isolation of affected people, by avoidance of whatever contact causes infection, or by cure. Genetic conditions, on the other hand, are controlled not only through prevention or palliative treatment but also through reproductive decisions and choices (adoption, reproductive technologies, etc.).

We will now examine, in brief, the various ethical, legal, and international contexts with regard to the issue of disclosure of genetic information to at-risk family members. We will not, however, consider the particulars of disclosure to minors; to spouses, for the purposes of

making informed reproductive choices⁶; or to at-risk relatives who have the same family physician as the patient.⁷

V. Contexts

A. Ethical Considerations

The concept of privacy has evolved, from a right of privacy, to a personal right to be left alone and, ultimately, to a fundamental right that is based on human dignity and respect for the individual—the latter notion being understood in terms of self-determination (Knoppers et al. 1995; LeBris and Knoppers 1997). Privacy rights, in the context of medical information, translate into protection of personal data and affirmation of confidentiality. As such, genetic information is protected by the legal and ethical principle of confidentiality that exists within the health-care professional–patient relationship. There is a commonly held view that, without an expectation of confidentiality, patients will be less forthcoming in disclosing sensitive personal information.

Confidentiality, however, is not absolute. Codes of medical ethics permit physicians, in exceptional cases, to disclose otherwise confidential information.⁸ Four ethical positions regarding a health-care professional’s duty or privilege to warn at-risk relatives about genetic information are found in the literature.

In the traditional health-care professional–patient relationship, confidentiality is absolute. All medical information is strictly private. Health-care professionals are obliged to inform patients about the implications of their genetic test results and about the potential risks to family members. However, confidentiality prevents the health-care professional from disclosing any genetic information to relatives; the health-care professional has a duty *not* to breach confidentiality. From this viewpoint, it would be unethical to do so.

Alternatives to this strict view of health-care profes-

⁴ This issue has been addressed specifically in the literature, with regard to legislation concerning genetic information. See Reilly (1997), who ultimately determines that, since genetic information will (in the not-too-distant future) become a standard part of the general medical record, laws that protect the privacy of all individual medical records, as opposed to laws that regulate genetic information specifically, are required. He notes that the Medical Records Confidentiality Act of 1995 (s. 1360, 104th Cong., 1st sess., sec. 2[1]), for example, presents a reasonable general approach to medical information and that it would be comprehensive if “genetic information” were added to its list of protected entities. See also Gostin (1995), who notes that the enactment of genetic-specific privacy legislation could create inconsistencies in the rules that govern the dissemination of health information and that, since “the flow of medical information is rarely restricted to particular diseases or conditions,” “comprehensive legislation on health information privacy, with explicit language applying privacy and security standards to genomic information” would be a better alternative. See also Knoppers (1997).

⁵ See Murray (1997, pp. 60–73). Murray rejects unique distinctions between genetic information and other medical information and notes, *inter alia*, that cholesterol levels are probabilistic information, that other medical information affects the family (although sensitivity may be amplified with genetic information), and that discrimination occurs with nongenetic information—as, for example, in the insurance industry.

⁶ The issue of disclosure to spouses is controversial; although there may be no personal risk to the spouse, genetic test results are pertinent for the purposes of making informed reproductive choices. The Institute of Medicine (1994) recommended that health-care professionals not reveal genetic information about a patient’s carrier status to the patient’s spouse, without the patient’s permission, since a spouse’s claim of harm—the possibility of bearing a child with a genetic disorder—could not be considered substantial and imminent.

⁷ This issue raises the question of whether the family physician’s obligation to disclose is greater given that the physician may already be treating relatives of the patient.

⁸ Note that the code of ethics of the Canadian Medical Association (1996) permits a breach of a patient’s right to confidentiality, *inter alia*, “when the maintenance of confidentiality would result in a significant risk of harm to others” (article 22). In contrast, the code of ethics of the American Medical Association (1996) does not refer to risk of harm but restricts itself to “disclosure need[ed] to protect the welfare of the individual or the public interest.”

sional-patient confidentiality have been proposed by the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research (1983). The President's commission states that if a patient refuses to inform at-risk family members, disclosure by a health-care professional would take place only when (1) reasonable efforts to elicit voluntary consent to disclosure have failed; (2) there is a high probability that harm will occur if the information is withheld, and the disclosed information will actually be used to avert harm; (3) the harm to identifiable individuals that would result from nondisclosure 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.⁹

These ethical guidelines do not imply a legal duty to warn; they simply set out circumstances in which the commission believes it would be permissible to do so. Accordingly, health-care professionals, in addition to informing patients that genetic information may affect the patients' relatives, may warn at-risk family members, if the above criteria are satisfied. Such criteria have been translated into a consideration of the following factors: the seriousness of the defect, the likelihood that the relative has the genetic defect, the likelihood that the defect may be detected by other means, the availability of treatment, and the seriousness of the harm to be suffered by third parties (Macklin 1992). Disclosure without consent is therefore justified if the information reveals that the relative is at a substantially higher risk of suffering from a serious and otherwise undetected genetic disorder and if treatment or prevention is available.

The Institute of Medicine Committee (1994), in its report on assessing genetic risks, recommended that genetic information be considered confidential. With regard to possible disclosure, the committee adopted language similar to that of the President's commission.¹⁰ The report indicated that, if the genetic disorder is highly likely to be present and is treatable or preventable, many health-care professionals would overrule a patient's refusal to disclose and would inform a relative.

A third possible ethical position involves warning the patient in advance about circumstances that would result in disclosure of genetic information to other family members. This warning would be provided before testing,

regardless of the patient's intentions to disclose. In this way the health-care professional-patient relationship would not be jeopardized, as long as the health-care professional both assures the patient of the presumption of confidentiality and outlines the exceptions to that presumption (Macklin 1992). Such a forewarning suggests that, if the appropriate factors are satisfied, the health-care professional, more than just encouraging the patient to disclose, *will* warn at-risk relatives about genetic information that involves a serious and treatable or preventable condition.

Finally, the duty to warn could be considered an ethical imperative. Eventually, the duty to warn could become obligatory rather than merely permissive. Indeed, absence of explicit legal regulations does not translate into an absence of duty. An ethical duty can become a professional norm of practice, and it may become the legal standard, as long as there are no other important countervailing policy issues.

Choosing between these ethical positions also requires further consideration of the notion of harm. It is important to consider the very real possibility that, within a family group, knowledge may constitute a greater harm than nondisclosure, particularly in the case of family members who do not want to know. Harm from disclosure may include psychological, social, and financial harm; the possibility of stigmatization, discrimination, and labeling; and the potential either to lose or to encounter difficulty in obtaining employment or insurance.

Yet failure to disclose may also lead to harm. In terms of reproductive choices, children who might otherwise have been spared the effects of a genetic condition will have to endure them, and couples who would otherwise choose not to conceive would be denied such an option.¹¹ Failure to warn may also lead to irreparable harm by limiting opportunities for treatment or prevention of the genetic condition. If the genetic condition is serious and preventable or treatable—that is, if the harm from nondisclosure outweighs the harm from disclosure—health-care professionals may have an ethical duty, depending on the circumstances, to warn family members, irrespective of their patients' wishes. We will now examine the issue of professional disclosure within the legal context.

B. Legal Considerations

The duty to maintain confidentiality extends as far back as the Hippocratic oath and, in the absence of statute, is based on theories of contract and on the fi-

⁹ Similar positions have been adopted by the Institute of Medicine (1994), the Science Council of Canada (Genetics in Health Care 1992), Canada (Medical Research Council of Canada et al. 1996), and the United Kingdom (Nuffield Council on Bioethics 1993).

¹⁰ Institute of Medicine (1994). The committee noted that the strongest case for warning by a health-care professional would exist where there is a high likelihood that the relative has the genetic defect, where the defect presents a serious risk to the relative, and where there is reason to believe that the disclosure is necessary to prevent serious harm.

¹¹ One author argues that warning relatives about the risk of conceiving a child with a deleterious gene does not pose the type of serious, imminent harm that would generally require disclosure (Andrews 1997a).

duciary nature of the health-care professional–patient relationship. Courts have distinguished between secrets and confidentiality; the former result in no liability for disclosure, whereas the latter impose a duty to protect information obtained through the relationship.¹² Despite its ability to impose liability for breach, the legal principle of confidentiality, as mentioned, is not absolute. As a result, the health-care professional may be faced with conflicting ethical, legal, and statutory obligations.

Although specific statutes protect the confidentiality of genetic information, they also set out situations in which such information may be disclosed without liability. For example, in the case of adoption, a genetic history and a history of hereditary conditions must be compiled and disclosed, if known, by either the child-placing agency or the biological parents, to the adoptee and the adoptive parents.¹³ Moreover, at least one state provides that, where a child-placing agency receives information that a birth parent or a subsequent child of a birth parent has or may have a genetically transmissible disease, the agency must notify either the adoptee (if the adoptee is ≥ 18 years of age) or a custodian, guardian, or adoptive parent (if the adoptee is < 18 years of age).¹⁴

Legislatures have required that other types of medical information be reported as well. There are statutes that require physicians to report, to the proper authorities, communicable diseases, gunshot and other wounds, and evidence of child abuse and neglect. The permission or the requirement to breach confidentiality in the case of contagious disease is based on public-policy interests in preventing the spread of disease. However, many contagious-disease statutes make breach of confidentiality a last-resort measure. Universal precautions against the transmission of the acquired immunodeficiency syndrome and hepatitis provide for precaution without requiring breach of confidentiality (unless the precautions fail, in which case a limited right to disclosure would exist). Finally, notification is warranted only where (1) the disease is easily transmitted, (2) it poses serious harm, and (3) treatment can lead to medical benefit.¹⁵

Some proposed legislation¹⁶ would make *no exception*

¹² See *Humphers v. First Interstate Bank of Oregon* (696 P. 2d 527 [OR 1985]), in which a physician revealed the identity of a former patient to the daughter she had given up for adoption. The physician was held to a nonconsensual duty of confidentiality imposed by virtue of his profession and “determined by standards outside the tort claim for its breach.”

¹³ For an extensive review of genetic disclosure in relation to adoption laws, see Andrews (1997b) and The American Society of Human Genetics (1991), who take the position that genetic history should be included in an adoptee’s record and who recommend that, where appropriate, genetic data should be shared among adoptive parents, biological parents, and adoptees.

¹⁴ Wisconsin Statute, sec. 48.432 (1994).

¹⁵ Andrews (1997b), citing Steele (1990, p. 451).

¹⁶ See, for example, Annas et al. (1995).

for a health-care professional to warn relatives who may be at risk for developing a genetic condition. This “no-exception rule” is based on the belief that it is difficult, if not impossible, to set boundaries on such an exception. Further, such a rule is seen to maximize the privacy between the patients who receive private genetic information and their health-care providers (Annas et al. 1995). The responsibility to inform family members of their potential genetic risks thus rests on the individual who has such knowledge. Others have also argued that family members should receive genetic information only when the family member, at the behest of the person tested, consents to or initiates such an inquiry (Pelias 1991, 1992).

The leading case on the general duty to warn is *Tarasoff v. Regents of the University of California* (551 P. 2d 334 [CA 1976] [en banc]).¹⁷ In this case, a psychiatrist was held responsible for not warning his patient’s intended victim of the patient’s murderous intentions. The court held that a duty to warn is likely to exist if (1) the physician has a *special relationship* with either the person who may cause the harm or the potential victim, (2) the potential victim or person at risk is *identifiable*, and (3) the harm to the victim is *foreseeable* and *serious*. Thus, a duty to warn could override the health-care professional’s duty to maintain confidentiality, if disclosure of genetic information could foreseeably prevent serious harm.

Responding to the logic of the *Tarasoff* case, many commentaries have distinguished between a duty to warn identifiable third parties of a threat of violence and a possible duty to warn at-risk relatives about potential genetic risk.¹⁸ They note that the crucial difference lies in the nature of the harm for which they are at risk. In the case of a threat of violence, the patient’s actions are likely to harm others; in the case of genetic conditions, the patient does not put relatives at risk simply by car-

¹⁷ See 551 P. 2d 334 (CA 1976) (en banc). Relevant to the issue of duty to warn and disclosure, see also (1) *Berry v. Moench* (331 P. 2d 814 [UT 1958]), in which a physician who disclosed a patient’s depression and legal and financial problems to a friend of the patient’s fiancée was granted a conditional privilege to reveal such information because a sufficiently important interest needed to be protected; and (2) *Simonsen v. Swenson* (104 NE 224, 177 N.W. 831 [NE 1920]), in which a physician was granted a privilege to disclose a patient’s infectious syphilitic state to the proprietor of the hotel in which the patient was staying. The physician was found to be immune from liability, because he acted in good faith, without malice, and because he did not disclose more than was necessary (at 177 N.W. 833).

¹⁸ Suter (1993); Park and Dickens (1995); and Miller (1994), who points out that there are a number of similarities between the two cases: a special relationship exists between the physician and the patient, the third party is identifiable, there is no special relationship between the physician and the third party, the information to be disclosed is confidential, and there is an opportunity to prevent harm.

rying the gene mutation—the relatives already either have or do not have the mutation.

Recent case law has dealt specifically with the issue of a physician's duty to warn in the context of genetic information. In the Pate case,¹⁹ a daughter suffering from medullary thyroid carcinoma sued her mother's treating physician on the grounds that (1) he knew or should have known that the mother's previously diagnosed cancer was hereditary; (2) this knowledge gave rise to a duty to warn the mother that her children might be at risk and that they should be tested; (3) had she been tested, she would have taken preventive measures; and (4) her condition, in all likelihood, would have been preventable. The court held, on the basis of state law protecting confidentiality and pursuant to the prevailing standard of care, that the physician had a duty to warn the mother—but not the daughter. The court noted that "[t]o require the physician to seek out and warn various members of the patient's family would often be difficult or impractical and would place too heavy a burden upon the physician."²⁰

However, the New Jersey court, in the Safer case, extended the physician's duty to warn to those "known to be at risk of avoidable harm from a genetically transmissible condition," irrespective of potential conflicts between the duty to warn and the obligation of confidentiality.²¹ In this case, a daughter diagnosed with colon cancer and multiple polyposis sued the estate of her father's treating physician 26 years after her father's death. The daughter alleged that the disease was hereditary and that the physician breached his duty to inform her, thus depriving her of the chance for monitoring, early detection, and early treatment. The appellate court, even while considering the Pate case, overruled the trial court's protection of confidentiality within the doctor-patient relationship and held that there can be a duty to warn relatives.²² The court applied the infectious-dis-

ease model²³ and noted that genetic risks are as foreseeable as infectious ones. "[T]he individual or group at risk is easily identified, and substantial future harm is easily identified or minimized by a timely and effective warning" (Safer case, p. 1192).

These cases may indicate an increasing trend toward disclosure, irrespective of the potential impact of such disclosure. Yet finally, in the United States, there is no general legal duty to rescue.²⁴ This means that, in the absence of other statutory provisions obliging disclosure, and without new definitions of the patient that include the family, the health-care professional is under no legal obligation to warn at-risk relatives, particularly since there is no existing relationship with the relative. Furthermore, neither the patient nor the health-care professional causes the potential risk to the relative. However, the health-care professional may have a privilege—that is, a discretionary right to act in a manner that would otherwise give rise to legal liability.²⁵ As some commentators have noted, "[t]he courts have recognized that if disclosure is performed to protect a person perceived to be in imminent peril of harm and is the minimum disclosure necessary to serve that purpose, it may be considered legally excusable" (Park and Dickens 1995, unpublished data). A consideration of the international positions on disclosure to at-risk relatives may be helpful in determining the appropriate paradigm.

C. International Positions

Many foreign jurisdictions and international organizations have examined questions related to genetic privacy and confidentiality and have formulated recom-

¹⁹ See also *Dimarco v. Lynch Homes—Chester County* (583 A. 2d 422 [PA 1990]), in which the court held that a physician has a duty of care to tell the patient about the risk of exposing third parties to a communicable disease (hepatitis B). The court considered, in *dicta*, that the physician should recognize that services rendered for the patient are necessary for the protection of third parties likely to be affected by the patient's disease.

²⁰ *Pate v. Threkel*, 661 So. 2d 278 (1995), at 282.

²¹ See 677 A. 2d 1188 (NJ Sup. 1996).

²² The case was remanded for trial to determine whether the duty to warn was breached in this case. The court noted that the trial court would have to consider the extent of the daughter's risk, compare the costs of monitoring versus the expenses associated with the breach of duty, and determine whether, if the father had instructed the doctor not to disclose his condition to his daughter, that request should have been honored.

²³ For a duty to warn in the contagious-disease cases, see, for example, *Skilling v. Allen*, (143 323, 173 N.W. 663 [MN 1919]), in which a physician was found to have a duty to use due care in advising a patient's parents about the possible transmission of infection; and *Gammill v. United States* (727 F. 2d 950, 954 [10th Cir. 1984]), in which it was held that a physician may be found liable for failing to warn a patient's family, a patient's treating attendants, or other persons likely to be exposed to the patient about the nature of the disease and the danger of exposure. Interestingly, a duty to warn was also found recently in a noncontagious-disease case: see *Bradshaw v. Daniel* (845 S.W. 2d Sup. Ct. 865 [TN 1993]), in which a physician was found to have a legal duty to warn a spouse of the risk of exposure to the source of the patient's noncontagious disease—in this case, Rocky Mountain spotted fever (RMSF)—because the patient and the spouse had the same epidemiological risk and because knowledge of that risk could have saved the spouse's life (notwithstanding that the patient could not give RMSF to his spouse).

²⁴ In Canada, there is generally no duty, in the absence of statute, to render assistance to individuals in danger (Linden 1993). However, Quebec (in the civil-law tradition) does recognize a general duty to rescue, in its Charter of Rights (L.R.Q., chap. C-12, 1978).

²⁵ *Black's Law Dictionary*, defines a privilege as "[t]hat which releases one from the performance of a duty or obligation, or exempts one from a liability" (p. 1197).

mentations that attempt to balance patients' expectations of privacy with other parties' claims to sensitive genetic information. The majority of foreign jurisdictions are in favor of permitting limited disclosure of genetic test results (without the consent of the patient) if the potential harm to at-risk relatives is grave and imminent. Only a few jurisdictions maintain that confidentiality and the patient's wishes with regard to non-disclosure must be respected without exception.

Limited disclosure has been recognized at international, regional, and national levels. Internationally, both the World Medical Association (in its "Declaration on the Human Genome Project" [44th World Medical Assembly 1992]) and experts advising the World Health Organization (regarding proposed guidelines on medical genetics and genetic services [Wertz et al. 1995]) recommend that confidentiality of genetic information be maintained except where family members are at high risk of serious harm and where disclosure could avert this harm.

At the regional level, the Council of Europe (1992), which maintained that confidentiality of genetic information must be ensured at all times and must be protected by the rules governing medical data, did make an allowance for disclosure in the case of severe genetic risks that affect the health of family members or their future children. However, the genetic data of one member of a couple cannot be communicated without the free and informed consent of the other member (Council of Europe 1990). The 1997 Convention on Human Rights and Biomedicine allows for the communication of genetic test results when necessary, *inter alia*, for the interest of public safety, the protection of public health, or the protection of the rights and freedoms of others (Council of Europe 1997).

Nationally, in the United Kingdom, the Nuffield Council on Bioethics (1993) recommended that accepted standards of confidentiality of medical information be followed as much as possible. The council also recommended that, if a patient refuses to disclose test results to family members, and if the physician has stressed the importance of sharing such information and has attempted to persuade the individual to allow disclosure, the patient's desire for confidentiality may be overridden, in exceptional circumstances only. Such decisions would be made on a case-by-case basis.

The Committee of the Health Council of the Netherlands (1989) holds the view that unauthorized disclosure may be permissible, under limited circumstances, when serious harm can be avoided. The council has noted that relatives' right to privacy should be considered in decisions as to whether disclosure should be made.

A recent report of the Privacy Commissioner of Aus-

tralia (1996) recommends that an individual's right to privacy give way to the imperative to prevent harm, where the risk is serious, real, and imminent and "where there is a possibility of effective intervention and the consequences of nonintervention are serious for affected relatives."²⁶

The Japan Society of Human Genetics (1996), in its "Guidelines for Genetic Testing," also stipulates that, where necessary to avoid serious injury, confidentiality can be broken, even in the absence of (subject) consent. However, such an exception must be made by a responsible ethics committee.

Other jurisdictions are explicit in making no allowances for unauthorized disclosure. Norway, for instance, has taken a clear and firm legislative stand on protecting personal privacy, with no exceptions made for disclosure, even under extreme circumstances.²⁷

The Swiss Academy of Medical Sciences (1993) and France's National Ethics Committee (1991) have established guidelines that do not permit disclosure without the patient's consent.²⁸ However, in its reiteration of this principle in 1995, the French National Ethics Committee noted that, given the principle of assistance to persons in danger, in the event of a research subject's refusal, the physician is confronted with an ethical dilemma that must be resolved, particularly where children are involved.²⁹

In 1995, the members of the House of Commons Science and Technology Committee (1995) disagreed with the Nuffield Council's suggestion that confidentiality should be less than absolute. The committee holds the

²⁶ Privacy Commissioner of Australia (1996, p. 33-35. See also Medical Research Ethics Committee of the National Health and Medical Research Council (NHMRC) (1991), who noted that their recommendation to respect the wishes of the individual tested was still contentious when the guidelines were written. They noted that it could well be argued that the right of the person at risk should outweigh privacy when there is a possibility either that a life could be saved or that a condition could be effectively treated, if the information were disclosed to the at-risk relative (p. 4).

²⁷ Norway Ministry of Health and Social Affairs (1993); Norway (1994). The Ministry of Health and Social Affairs determined "that information on the genes of people regarded as healthy must remain strictly confidential. Such information must not be stored, not even by health institutions in patient records" (p. 64-65).

²⁸ Comité Consultatif National d'Éthique pour les Sciences de la Vie et de la Santé (1991); Swiss Academy of Medical Sciences (1993, guideline 3.7); Law 94-654 (29 July 1994) On the donation and use of elements and products of the human body, medically assisted procreation, and prenatal diagnosis (1994) 45:4 *Int Digest Health Legis* 473, Title VI, A.L. 145-15.

²⁹ Comité Consultatif National d'Éthique pour les Sciences de la Vie et de la Santé, Avis 46 (7 novembre 1995) *Génétique et médecine: de la prédiction à la prévention*, rec. 2. The ethics report accompanying this opinion noted both the importance of genetic information for subsequent generations and the possible duty of the physician to disclose genetic information, when useful, to persons at risk (R. 3.3).

opinion that, if an individual cannot be persuaded through counseling to share information with relatives, then the individual's decision to withhold the information should be paramount.³⁰

VI. Conclusion

It is clear that genetic information is both individual and familial. This raises conflicts between the duty to maintain confidentiality and the duty to warn. Yet ethical, legal, and statutory exceptions limit the principle of confidentiality and, in specific and very limited circumstances, may permit disclosure. At the very least, it is clear that a health-care professional has a positive duty to inform a patient about potential genetic risks to the patient's relatives. Then, depending on the circumstances, the health-care professional may have a privilege to warn at-risk relatives if the harm is serious, imminent, and likely; if prevention or treatment is available; and if the health-care professional, if she or he were in similar circumstances, would disclose. "Seriousness" defies exact definition and must be determined on an ad hoc basis.³¹ While overly broad definitions may leave a health-care professional confused or in conflict, such ambiguity is often the hallmark of professional judgement. The ethical duty of health-care professionals to warn at-risk relatives will undoubtedly be the topic of future debate.³²

³⁰ House of Commons Science and Technology Committee (1995). The report maintained that the failure to inform relatives places them "at no worse position than if no test has been performed. To fail to respect the privacy of genetic information in this way could discourage couples from participating in research for the common good or from seeking information which could help them safeguard their health" (par. 227-228).

³¹ The notion of serious harm is highly subjective. See Knoppers et al. (1995), in which geneticists certified by American Board of Medical Genetics (ABMG) or American Board of Genetic Counselors (ABGC) and members of the Canadian College of Medical Geneticists, the European Society of Human Genetics, and the Ibero-American Society of Human Genetics were surveyed on how they viewed the term "serious." No consensus was found. Furthermore, of 947 responses received, 24% of U.S., 18% of Canadian, 37% of European, and 65% of Latin American respondents thought that professional associations should develop lists of serious disorders. Eight percent of respondents (4% of U.S. respondents) favored a legal definition of "serious." Some thought a national ethics committee (16%), hospital ethics committees (14%), or individual practitioners (28%) should define the term. Seventy-two percent of U.S.-Canadian respondents, 54% of European respondents, and 31% of Latin American respondents thought that individual patients should define "serious."

³² See Safer case, in which the court noted that "the duty to warn of avertible risk from genetic causes, by definition a matter of familial concern, is sufficiently narrow to serve the interests of justice."

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