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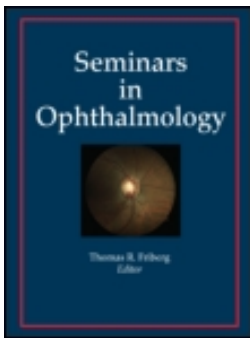
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# Preventive Ethics in the Management of Ophthalmic Genetic Disorders

Lisa S. Parker and Michael B. Gorin

**M**OST OF MODERN medical practice consists of identifying disease, deterioration, and disability and then attempting to ameliorate these ill effects. Only recently has medicine been equipped to turn greater attention to the prevention of debilitating conditions. Among the most powerful advances in prevention are genetic tools that permit the prediction of heritable and late-onset conditions (eg, macular degeneration and glaucoma). Some parents may engage in reproductive planning to prevent passing an increased risk of disease to their offspring; some patients may use information about their genetically based increased risk to engage in preventive behaviors, for example, being especially vigilant to detect early symptoms or avoiding environmental factors that may exacerbate or trigger the particular condition for which they are at risk. Gene therapy may soon offer an alternative approach to prevent or reduce the risk of many diseases.

Ophthalmologists will increasingly be trained in and held responsible for understanding and educating patients about the genetics of eye diseases. They will be accountable for the diagnostic and predictive aspects of the genetics of eye conditions and related systemic manifestations as they pertain not only to their patients but to other family members. As concern about the genetics of disease becomes more thoroughly integrated into traditional and preventive medicine, ophthalmologists will increasingly face ethical concerns for which their current training may not have prepared them.

Fortunately, the field of bioethics, like medicine, has taken a preventive turn.<sup>1,2</sup> Instead of merely responding to ethical conflicts as they arise in the ophthalmologist's office or during the course of a research protocol, preventive bioethics attempts to identify recurrent patterns of ethically problematic interaction (sometimes in quite different contexts) and to formulate procedures, policies, and protocols designed to attempt to avoid such difficulties in the future. Additionally, instead of examining an ethical problem in relative isolation, a preventive ethics strategy examines its cultural, political, and

economic contexts to determine how institutions and structures create or contribute to the problem. Not every problem can be prevented or anticipated; nevertheless, preventive ethics attempts to equip professionals and patients with the skills, resources, and models for considering problems as they arise.

The following discussion addresses a variety of ethical and legal issues that ophthalmologists are likely to confront in their practices and research. We consider not only the ethical, social, and economic considerations, but also the contexts in which political and economic institutions affect these ethical problems. We consider the resources available to help guide ophthalmologists' considerations and present preventive ethics models for reasoning about such situations.

## PREVENTIVE ETHICS, POLICIES, AND INFORMED CONSENT

Professional societies have, to some degree, adopted a preventive stance with respect to ethical issues by issuing policy statements or codes of ethics that provide frameworks to guide ophthalmic clinical practice and research. These documents seldom offer more than points to consider or general guidelines, and they do not provide guidance about their application to particular cases. At best, they provide an ethical context and illuminate values to be considered by ophthalmologists when formulating more specific policies and protocols.

The American Academy of Ophthalmology Policy Statement on the Unique Competence of the Ophthalmologist, for example, recognizes that the qualified ophthalmologist is competent to diagnose and appropriately manage a wide

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spectrum of ophthalmic disease including genetic syndromes and systemic diseases (eg, hypertension and diabetes mellitus) that are often revealed in the eye.<sup>3</sup> Yet it is not clear what constitutes the type of genetic syndrome an ophthalmologist should be competent to treat. Should the ophthalmologist only treat those syndromes that fall within the spectrum of ophthalmic disease? What if the genetic syndrome is detectable in the eye but causes no significant ocular or visual morbidity, such as Gardner's syndrome, Carney syndrome, or Wilson's disease? Does appropriate management include making the diagnosis, treating the underlying genetic condition, or referring the patient to a medical geneticist? Does the competent ophthalmologist also have to be a competent genetic counselor or, for example, a competent general internist?<sup>4</sup>

On this point, drawing analogies from the guidelines of the American Academy of Pediatrics may be instructive. Its Committee on Genetics states

Pediatricians may be called upon to counsel a family in which prenatal diagnosis is being considered or in which there is a fetus with a genetic disorder. In some settings, the pediatrician may be the primary resource for counseling the family. More frequently, counseling may already have been provided by a clinical geneticist and/or obstetrician. However, because of a previous relationship with the family, the pediatrician may be called on to review the information and to assist the family in the decision-making process. The pediatrician should be familiar with the principles of prenatal genetic diagnosis and know how to apply them to specific problems in genetic counseling, diagnosis, and management in clinical practice. At the same time, pediatricians should be familiar with resources available in their region for obtaining information about whether and how a specific disorder can be diagnosed and when and where to refer patients for prenatal genetic diagnosis. The technology of prenatal diagnosis is changing rapidly, and genetic consultants can assist pediatricians in the appropriate utilization and interpretation of the available diagnostic tests.<sup>5</sup>

Under certain circumstances, a pediatrician must be prepared to play the role of the genetic counselor and must know when to refer the patient to another physician.

Can we extrapolate from these comments the duties of an ophthalmologist? Pediatricians, like ophthalmologists, are specialists but they also serve as primary care providers to the

young. Patients (or their parents) often self-refer to both pediatricians and ophthalmologists without the advice or consent of another physician with whom they might have a greater rapport or a more ongoing relationship. On the other hand, pediatricians treat the whole body, and the province of the pediatrician's responsibilities might be broader than an ophthalmologist's and might, therefore, justify an active role in providing counseling about genetic-related conditions. However, this difference is not clear-cut; pediatricians may find themselves just as ill equipped to counsel families about the genetics of diseases whose symptoms first appear in adulthood as ophthalmologists would be to counsel about the genetics of hypertension. Nevertheless, both may bear responsibilities to provide such counseling or at least to see that such counseling is available to their patients.

Thus, responsible ophthalmologists need to consider the ethical concerns raised by discovering conditions with genetic and systemic implications for the health of their patients and their patients' families. They should develop policies within their practices or research protocols to govern appropriate referrals for genetic counseling and follow-up care for patients with increased genetic risks or risks for systemic disease related to ophthalmic findings, as well as appropriate disclosure of such findings to patients and the documentation of such findings and referrals. Such policies must take into account a variety of ethical considerations, precedents, and existing guidelines. The task is not easy because some of these considerations conflict. Risks and benefits to patients, their relatives, and professionals must be weighed.

### *Informed Consent*

Consider, for example, the Code of Ethics of the American Academy of Ophthalmology, which states that it is the ophthalmologist's responsibility to act in the patient's best interest.<sup>6</sup> Despite initial appearances, it is unclear whether informing patients of genetic conditions or risks is always in their best interests, because the psychological, social, and economic consequences of learning about their increased risks (and those of their relatives) may outweigh the benefits that preventive medicine may have to offer for the condition.<sup>7</sup> This dilemma has

been well recognized for individuals at risk for sickle cell anemia,<sup>8</sup> Huntington disease,<sup>9-11</sup> or familial Alzheimer disease.<sup>12,13</sup> Moreover, although respect for patients' rights of self-determination would argue in favor of allowing them to weigh these risks and benefits themselves, it is a challenge to provide patients with enough information to enable them to make these assessments. Finally, professional standards of care, concern to avoid future legal liability for failure to disclose information material to patients' health, and various reporting requirements (eg, to insurance companies, employers, or state agencies) all argue in favor of informing patients of their genetically based increased risks, although doing so may not be either in their overall self interests or what they themselves would choose.

As genetic counselors are well aware, the provision of genetic information itself constitutes a type of medical intervention. According to one of the fundamental doctrines of medical ethics and tort law concerning medical practice, patients must give their voluntary informed consent to medical interventions. A strange "catch 22" arises. To receive sufficient information to give fully informed consent, patients must often be provided with the relevant genetic information before their consent. At that point, however, they cannot refuse the information, so its receipt may not be voluntary, and consent certainly has not taken place. The best practical solution may be for a clinician or researcher to explain in general terms that genetic information may be available and that learning genetic information imposes various psychosocial and economic risks, as well as potential benefits. The clinician or researcher would then obtain informed consent to continue with more detailed discussion of the type of information that is and is not available and would obtain consent to continue at each stage where the information becomes more specific or more specifically pertinent to the patient or research subject. This may seem an elaborate or complicated process, but it can be clarified with a specific example. Informing a patient that there is a genetic test for early-onset Alzheimer's disease or Huntingtons disease imposes minimal psychological or social risk to that individual. However obtaining a family history

that might indicate that the person is at an increased risk for one of these diseases would be associated with an additional risk of psychological stress and economic concerns. Proceeding to the next level of evaluation, which might include diagnostic testing, imposes additional psychological, social, and economic risks to the patient. At each stage informed consent must be obtained and the patient must be an active participant in deciding what the appropriate risks and benefits are and whether to incur them. In the case of providing genetic information, informed consent is indeed an ongoing process and dialogue, not a single event like the signing of a form.<sup>14</sup>

It is unusual for an ophthalmologist to be able to make a conclusive genetic diagnosis without additional investigation of family history, clinical studies, or both. In most cases, recognition by the ophthalmologist of a genetic condition that has serious systemic manifestations is not pathognomonic or conclusive. Many conditions, such as retinitis pigmentosa, ocular albinism, Stargardt's disease, cone dystrophies, and optic atrophies, have several modes of inheritance. Determining the mode of inheritance to provide adequate genetic counseling for a given individual would require the ophthalmologist to investigate other family members. Like performing other diagnostic evaluations, the evaluation of additional family members to establish a genetic diagnosis requires informed consent including the disclosure of indications, potential complications, risks, and benefits. In family studies, the economic and psychosocial risks pertain not only to the patient (or proband) but also to the other participating family members and their relatives. Special care must be taken to ensure that all participating parties are adequately informed of the risks and benefits of their participation and that the family members are participating in the diagnostic or research study voluntarily.

Indeed in the research context, particular concern has been paid to whether family members who decide to participate in genetic family studies do so voluntarily. There are at least two possible sources of coercion: researchers themselves and other family members. These sources of pressure may also be relevant concerns in the nonresearch clinical environment. It has been

argued that genetic family studies do not present novel or increased risks of familial pressure as compared with other types of medical research or treatment.<sup>15</sup> Various types of pressures are present in most decisions, but the voluntariness of consent is only impugned if the pressure itself is illegitimate or if it comes from an inappropriate source.<sup>14</sup> Threats of physical force, be they from family members, physicians, counselors, or researchers, are an example of illegitimate pressures. On the other hand, family members might legitimately threaten to withdraw from contact with a prospective participant without impugning the voluntariness of the participant's consent. However, a physician or researcher who made a similar threat or encouraged this intrafamilial pressure would be coercing or putting illegitimate pressure on the prospective participant. In general, it is difficult to regulate familial interactions, therefore, researchers (and by analogy, physicians) should be most concerned that their protocols governing interactions with prospective participants ensure that information about potential risks and benefits is accurately disclosed and that researchers do not unduly influence decisions regarding participation.<sup>15</sup>

Because of these complications presented by family dynamics and by the psychosocial and economic nature of the risks to be disclosed to patients and their family members, clinical ophthalmologists may choose to refer to trained genetic counselors those patients who are (or are thought to be) at genetically based increased risk for disease, as well as family members seeking to learn their risk status. Clinical researchers may benefit from collaborating with genetic counselors to develop and implement appropriate protocols for informed consent and genetic counseling.

The ophthalmologist who chooses to pursue the confirmation of a diagnosis of a systemic genetic disease, first suspected on initial clinical findings during an eye examination, should engage in the informed consent process with his patient before initiating the medical follow-up necessary to establish or to confirm the diagnosis. For example, consider an individual with recurrent iritis. If the ophthalmologist suspects that the iritis is caused by ankylosing spondylitis, then there is an obligation to inform the

patient of the potential medical, economic, and psychosocial consequences of that diagnosis before initiating a diagnostic work-up. Should the patient be reluctant to have the systemic diagnosis established, the patient and the ophthalmologist may elect to have HLA-B27 testing done as part of the work-up for recurrent iritis, but not pursue radiological studies to detect subtle manifestations of arthritis. (This aspect of informed consent would also apply to discussions with the same iritis patient regarding the ordering of a blood test for syphilis, which is often included in a uveitis work-up. Most ophthalmologists recognize that the patient must be informed and must consent to this test being ordered, particularly because of the legal requirements for reporting positive test results to public health authorities.) The physician's records might indicate that the patient is HLA-B27 positive and at risk for ankylosing spondylitis but without systemic symptoms. This approach would adequately document the association for liability, diagnostic, and management purposes, without labeling a patient with the systemic diagnosis. Similarly, although an ophthalmologist may identify Lisch nodules as incidental findings in a patient who may or may not have a family history of neurofibromatosis, the diagnosis of neurofibromatosis 1 would, at a *minimum*, require confirmation of the family history or the identification of other dermatologic or systemic lesions. By informing their patients before additional evaluations about the possible psychosocial and economic risks that attend discovery of either disease or increased risk for disease, ophthalmologists can avoid unnecessary paternalism and permit patients to decide whether they want to assume those risks in light of both the risks and benefits of early diagnosis.

#### DISCLOSURE OF GENETIC INFORMATION TO THIRD PARTIES

##### *Confidentiality and Third Parties' Interests*

Patients' confidentiality must be maintained so far as is allowed given legal requirements.<sup>6</sup> Yet family members, employers, and insurance companies all have interests in obtaining genetic information about individuals, and each group has different motivations for wanting this information. Family members desire genetic

information to make reproductive decisions and plans concerning their own health care and health insurance. Notwithstanding the Americans with Disabilities Act (ADA) of 1990,<sup>16</sup> employers may want to use genetic information to eliminate employees or not hire persons who might present a safety risk to others, increase group health insurance premiums, or result in a loss on a training investment.<sup>17,18</sup> Insurance companies want to use genetic information, like other medical and actuarial information, in their risk assessments about individuals seeking insurance.<sup>19-21</sup> However, providing family members, employers, and insurance companies with genetic information presents serious ethical conflicts and difficulties, including the problem of preserving patient confidentiality within and outside their own families.

Diagnosis or discovery of a person's increased risk for a disease or disability has immediate implications for other individuals in his family and the individual's own insurability and employability. Discovery of a genetically based increased risk, or diagnosis of a condition with genetic components, can be made without performing any genetic tests. Ophthalmologists, for example, can identify lesions that are suspicious for von Hippel Lindau disease, Gardner's syndrome, neurofibromatosis 1, myotonic dystrophy, Marfan's syndrome, Stickler's syndrome, and many other conditions in the course of a thorough eye examination. These lesions are predictors of increased risk for systemic morbidity in the individual patient and for that person's relatives. The patient's family members may be unaware of their vulnerability to vision- or life-threatening abnormalities that are presently undetected and unaware that their offspring may be at risk for a potentially serious genetic disorder. Therefore, it would seem that the responsible ophthalmologist must consider the potential genetic risks for the patient's family members and may incur an obligation to inform family members of these possible genetic risks. However, this is not such an obvious obligation, nor is it clear how this apparent responsibility could best be fulfilled.

### *Informing Relatives*

Determining how to discharge an apparent obligation to inform family members about

potential genetic risks may be quite problematic. Can the ophthalmologist legitimately shift the responsibility of notifying other at-risk family members to the patient? Is it sufficient for the ophthalmologist to document that the patient has been informed and that the patient will assume responsibility for contacting family members? Regardless of the intervention that the ophthalmologist decides to pursue, it is essential that the physician's records clearly state the issues that were discussed with the patient, including the potential beneficial and harmful impact of this genetic condition on other family members.

In the case of a patient with a newly discovered genetic condition, the ophthalmologist has no physician-patient relationship with the other members of the family; there is no obligation based on the fiduciary nature of the traditional physician-patient relationship. If the ophthalmologist assumes this obligation, how should it be discharged without violating the confidentiality of the patient? How can the ophthalmologist obtain the information necessary to contact people who are not his patients? How can the ophthalmologist approach family members while preserving their rights of privacy, confidentiality, and informed consent, including the right not to learn unsought and undesired medical information (especially because learning of their increased risk would obligate them to divulge it were an insurance company to request it, which could lead to the cancellation of health or disability insurance)?

Ethical complications attend warning family members of genetic risks, especially if the patient whose genetic condition has been discovered does not wish to inform his relatives about his condition and its implications. A patient may prefer to keep a potentially stigmatizing disease secret or may have personal reasons for avoiding contact with specific family members. In some families and societies, loss of eyesight is itself stigmatizing, because it is linked to aging and loss of physical and mental prowess. Thus in the clinical setting, the ophthalmologist must grapple with the dilemma of preserving or violating patient confidentiality. Even in situations where the patient is willing to share the information with other family members, difficulties of respecting privacy can arise. Relatives

may resent being contacted, because they will consequently incur the psychosocial and economic risks of being at risk for disease.

There is little guidance in the literature for the practicing physician with regard to contacting relatives at risk for a genetic condition. However, these issues have been considered in the context of genetic research. We can, thus, turn to the clinical research environment to obtain insights that can guide better clinical practice. According to guidelines from the Office of Protection from Research Risks (OPRR), institutional review boards (IRBs) must bear in mind that within families, each person is an individual and as such deserves to have information about himself kept confidential. Family members are, therefore, not entitled to be informed of each other's diagnoses except in rare circumstances.<sup>22</sup> According to the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, confidentiality can be overridden only if the following conditions are satisfied: first, reasonable efforts to elicit voluntary consent to disclosure have failed; second, 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; third, the harm that identifiable individuals would suffer is serious; and finally, if third parties are warned, appropriate precautions are taken to ensure that only the genetic information needed for diagnosis and treatment of the disease in question is disclosed.<sup>22,23</sup>

It is difficult to imagine that the circumstances that ophthalmologists will face can truly fulfill these conditions because of the difficulty of predicting how others will use genetic and other medical information. Thus, the magnitude of possible benefit to others from such disclosure and the difficulty of predicting the magnitude of risks of disclosure, including deterioration among family members' relationships, anxiety, stigma, and loss of insurability or employability cannot be accurately assessed by most ophthalmologists. Informed of their increased risk of breast, cervical and colorectal cancer, for example, some people do not engage in preventive medical interventions.<sup>24-29</sup> Similarly, those with increased risks of hypertension and heart disease do not uniformly engage in

preventive strategies, including smoking cessation, exercise, and dietary modifications.<sup>30,31</sup> These people would incur all of the risks of knowing their increased risk without enjoying the benefits.

Although it could be argued that the ophthalmologist who discloses an increased risk for a genetic condition at least gives family members the opportunity to benefit from avoiding an obvious harm, the physician cannot be reasonably certain that the actual benefits will outweigh the harms of disclosing the patient's private information without his consent, as well as the psychosocial and economic risks to the individuals who are contacted and informed. Among the harms to the initial patient (or proband) would be the physician's breach of confidentiality and the patient's attendant loss of confidence in the medical profession, at a time when, because of his own diagnosis of disease or increased risk, such confidence and a good physician-patient relationship may be especially important.

In a clinical research environment, ophthalmologists should take two preventive ethics steps. First, they should specify in advance of a clinical examination (or examination for research purposes) what medical information will be disclosed to whom and under what circumstances, and they should disclose the risks attending such a disclosure policy. Patients and research subjects may then give their informed consent to the examination and may be better prepared to deal with the consequences of anticipated and unexpected findings. This strategy is especially important because most people do not anticipate the scope of possible medical findings; most think of an eye examination as having limited health consequences and do not contemplate psychosocial or economic risks at all. Indeed, OPRR guidelines suggest that IRBs require investigators to establish in advance what information will be revealed to whom and under what circumstances, and to communicate these conditions to subjects in a clear manner.<sup>22</sup> Furthermore, subjects should know, and agree ahead of time to what kinds of information they might or might not learn both about themselves and others and what others might learn about them. Second, if an ophthalmologist discovers a condition or finds himself in a circumstance



radically different from those contemplated by the preventive ethics policy that has been devised and discussed with the patient, the ophthalmologist should endeavor to have the patient make the decision about disclosure to relatives.

Complicating decisions about informing relatives is the fact that the potential benefits of making early clinical or molecular ophthalmic genetic diagnoses in at-risk family members are changing. As new therapies emerge, conditions that have been considered untreatable may be amenable to therapy that can slow progressive visual loss. One example is the use of Vitamin A palmitate supplementation to slow the progression of retinitis pigmentosa.<sup>32</sup> A national, multicenter study is currently underway to investigate the protective role of vitamin and mineral supplementation for age-related macular degeneration.<sup>33</sup> Identification of a fetus at risk for retinoblastoma allows for early screening and tumor management. The recognition of pigment dispersion syndrome can lead to earlier, and potentially more successful, treatment for secondary glaucoma. Even for those conditions that remain untreatable, there are potential benefits in establishing diagnoses in other family members. Children can, for example, experience considerable psychological and scholastic injury from vision difficulties that are unrecognized until their conditions become advanced. Finally, individuals may be relatively asymptomatic, but have sufficient visual deficits that create a personal and public hazard if they continue to drive.

Nevertheless, ophthalmologists should be wary of becoming too enamored of these potential benefits and acting paternalistically on the basis of their promise. Indeed, although ophthalmologists may be expert about the disease, its genetic implications, and the psychosocial and economic risks that generally accompany discovery of the condition, patients are likely to be more authoritative about their own and family members' circumstances. In all but the cases of most serious potential harm, conflicts about disclosure may be most appropriately resolved, from an ethical perspective, in the patient's favor, not the ophthalmologist's.

A problem, however, remains. Although people often agree about what constitutes a harm or a benefit, sometimes they do not,

especially if their cultural, personal values, or life circumstances differ. For example, Hispanic parents in New York tend to regard their children's diagnosis of mental retardation as not too serious in relation to other difficulties they face.<sup>34,35</sup> An ophthalmologist cannot be sure that patients or their relatives will share his view of a particular condition as a harm of sufficient magnitude to warrant warning them. Yet, because medical treatment strives to protect the interests and promote the values of patients, not of clinicians and researchers, the patient's conception of a serious harm is really the salient one. Perhaps the best that a clinician or researcher can offer in anticipation of these rare situations, when disclosure to relatives without a patient's consent may be warranted, is to inform patients, before the completion of their examinations and studies, of the professional standards concerning what constitutes a serious, preventable harm that would justify breaching a patient's confidence. Such a policy would contemplate not only genetic issues, but also infectious diseases that must be reported to health authorities or vision loss that would violate the legal standards for driving. In this latter instance, ophthalmologists are already confronted by legal obligations in many states that create conflicts with the patient's interests and confidentiality.

#### *Informing Employers*

Employers may have a variety of reasons for wanting genetic information about their employees. Especially when employers engage in self-insurance schemes, rather than participating in regulated group health insurance plans, the incentive for and possibility of using medical and genetic information to avoid employing, and thus insuring, "expensive" employees who are at increased risk for health problems increases dramatically. Employers could learn of their employees' health risk information in a variety of ways: medical screening before or during employment, company doctors' health records, and employees' self-reports. The ADA does afford employees some measure of protection, but it is unclear precisely how genetically based increased risks will be interpreted under the Act.<sup>36-39</sup> The ADA prohibits discrimination on the basis of disease or disability that is not

relevant to job performance and prohibits employers from conducting pre-employment medical examinations except to test for illegal drug use or physical agility.<sup>16,40</sup> Offers of employment may be conditioned on the results of medical examinations, which are permitted under the ADA as long as an offer of employment has been extended and employment has not commenced, and only if all employees are subjected to such examinations. The ADA does provide that separate health and employment records be kept and that decisions about hiring, retention, and promotion cannot consider health information.<sup>41</sup> Nevertheless, because of the subjective nature of such decisions, health information that becomes public knowledge or that leaks into personnel records may subtly affect such employment decisions with relative impunity.

A preventive ethics approach stresses “mirandizing” patients about the immediate and long-term psychosocial and economic risks of learning medical information, just as typical informed consent requires informing them of the (usually physical) risks and benefits of any medical intervention. Individuals who are seen by a company physician often realize that certain pieces of medical information, including substance abuse, disabilities likely to affect job performance, or deterioration of eye sight, will be shared with their employer. They are not likely, however, to realize that an eye examination can reveal information about systemic conditions, future conditions, or conditions of their offspring (who may be insured as dependents on a company group plan and who, thus, may be viewed as potentially expensive insurance liabilities). This information, as well as any conflicting responsibilities to report findings about the individual and his family’s health, needs to be disclosed before an examination. Although employees may have little choice but to submit to examinations required as terms of their employment, they can at least take steps to minimize the magnitude of possible harms if they are informed of the risks.

Company doctors’ reporting of examination findings to an employer or potential employer is not the most likely path for employees’ medical information to travel to employers. Employees who discover that they have a genetic condition,

for example by seeing their family ophthalmologist, may be compelled to give their employer this information, particularly if their employer requests it for insurance purposes or in the interests of public safety (eg, in the airline industry or other public transportation).<sup>36,42,43</sup> Under the ADA, self-insuring group health plans can be structured to anticipate and limit coverage for particular conditions such as expensive genetic disorders and high risk conditions.<sup>19</sup> Indeed, in *McGann v H & H Music Co* the court held that the self-insuring Texas music store was permitted to reduce coverage for HIV-related illnesses from \$1 million to \$5,000<sup>44,45</sup>; similarly, in *Owens v Storehouse, Inc* the court upheld the employer’s right to reduce another HIV-infected employee’s insurance coverage from \$1 million to \$25,000.<sup>46</sup>

#### *Social and Economic Institutions*

Throughout the preceding discussion, concern about the loss of insurance, particularly health and disability insurance, received much attention because loss of insurance (either directly, or by losing one’s employment) constitutes one of the major risks of diagnosing a condition that may be presymptomatic or that may have findings that are not yet evident. The situation becomes even more complicated when one realizes that one cannot always predict whether an individual will develop none, some, or all of the complications of a specific genetic disorder. It is not clear whether insurers would pay for medical services or deny coverage to an individual who developed a genetic disease (or was found to carry a gene that put him at higher risk for developing a particular genetic condition) by classifying him as having a preexisting condition.<sup>19</sup> Provision of universal health insurance coverage, for example by government mandate, would largely eliminate this risk, but only if it provided for an adequate minimum of health care and catastrophic coverage.

Whether or not such health care reform is forthcoming, some risks will remain. First, mandated coverage may not be sufficient to provide what many would deem an adequate level of care or quality of life, and private market insurance companies will still have the same incentives to avoid insuring (or to insure at much higher rates) those at increased risk.

Second, disability insurance may be necessary to ensure a minimally adequate quality of life, and this is not typically included in proposals for insurance reform. Companies that provide disability insurance typically have even stricter standards for underwriting disability insurance than those for life or health insurance because of the large number of potentially disabling conditions and the large payout; they may also demand more extensive information and information about applicants' medical status.<sup>19</sup>

Third, obtaining life insurance is not only important to protect one's dependents, but it is also required by some lending institutions to purchase property or obtain capital for business ventures.<sup>47</sup> Although group policies do not require much medical information, approximately 75% of life insurance policies are purchased individually.<sup>19</sup> Discovery of increased risk for serious disease has provided grounds to refuse to underwrite those at risk altogether.

Fourth, even if all of these insurance problems were resolved, the anxiety and social stigma that accompany being at increased risk for disease would not be eliminated. Although one might not worry about loss of insurance or employment, health worries, concerns about being a desirable domestic or business partner, and concerns about being able to accomplish one's life plans will still attend discovery of one's risk. Thus, even with proposed health care and insurance reforms, discovery of one's genetically related risk for a disease is not without psychosocial and economic risks.

In the absence of health care and insurance reforms, these risks are very real, albeit not new with the advent of genetic technologies. Discovery of high cholesterol with a family history of heart disease or increased intraocular pressures with a family history of glaucoma—findings that would constitute the first stage of formulating a preventive medical strategy and that do not use genetic technologies—could also place an individual at risk for paying higher insurance rates or being uninsurable. Insurance companies use medical information of all sorts to project claims, to deny or limit coverage, and to prevent applicants from concealing known risk factors. Nevertheless, genetic information is sometimes more quantifiable and, thus, seems to provide a sense of certainty or predictability, although usually

falsely based.<sup>48</sup> Moreover, genetic information may be more far reaching than some other health information; genetic information about one person that affects his access to health care could also affect access to health care for that person's family members.

Nevertheless, an additional problem might arise if physicians counseled patients about or assisted them in avoiding incurring higher premiums or losing insurance policies on the basis of genetic testing results. From the point of view of the insurance industry, they may then be contributing to the problem of "adverse selection," in which individuals who have more information about their medical risks than insurance companies engaged in underwriting are able to purchase coverage at lower rates. It has also been argued that instead of helping their patients skirt insurance company practices, health care practitioners should cooperate with insurers to achieve fair and reasonable policies for everyone.<sup>19,49</sup>

#### LEGAL CONSIDERATIONS

There is a growing awareness of the importance of genetics in general medical and ophthalmic care. There is also rapid development in the molecular diagnostics of genetic conditions and recognition that individuals have the right to use genetic information for the promotion of the health of themselves and their offspring. Despite proposal of various legislative initiatives to protect individuals' privacy with respect to genetic information and moves to regulate use of banked DNA materials, no significant federal legislation or regulation has addressed the responsibilities of clinicians and researchers specifically with respect to genetic information. Therefore, attention should be paid to relevant analogies and possible precedent legal cases, especially in tort law.

##### *Duty to Warn*

Malpractice liability for the failure to warn of genetic risks is likely to be of greatest concern to clinicians and researchers who are caught between the ethical (and often legal) duty to protect patient confidentiality and perceived responsibilities to warn of preventable harms. In *Simonsen v Swenson*, for example, despite the court's recognition that physicians have a posi-

tive duty to protect privacy, the physician was not liable for informing a hotel owner of the infectious syphilitic condition of one of her guests.<sup>50</sup> Disclosure was justified on the grounds of prevention of the spread of disease; immunity from liability was provided because the physician acted in good faith, without malice, and did not disclose more information than necessary.

It is important to distinguish, however, between the imposition of a duty to warn and provision of immunity from liability. In some cases, physicians are required to report their patients' conditions, eg, public health reporting requirements for particular infectious or sexually transmitted diseases and state requirements that vision loss be reported to authorities governing drivers' licensing. In other contexts, common law duties have evolved that place a duty on clinicians to warn third parties. The most famous "duty to warn" case in the case history of bioethics and law is *Tarasoff v Regents of University of California*, which established that psychiatrists had a duty to warn identifiable third parties of credible threats of serious harm by their patients.<sup>51</sup> Although *Tarasoff* is of limited scope and jurisdiction (California), it has served as a paradigm case for reasoning analogically about clinicians' and researchers' duties to warn in other contexts, most recently perhaps with respect to warning the sex and intravenous drug using partners of HIV-infected patients, as well as family members of patients with other infectious diseases. *Bradshaw v Daniel*, for example, held a physician liable who failed to warn the spouse of his patient who was infected with Rocky Mountain Spotted Fever; both the patient and spouse died.<sup>52</sup>

Thus, some cases that involve permissible breach of patient confidentiality are justified on the grounds of preventing further contagion or in the interest of the public's health. Cases modeled on *Tarasoff* ground the breach of confidentiality on the basis of preventing harms within private relationships. This increased risk of harm within a private relationship suggests that cases like *Tarasoff* and *Bradshaw* might provide useful insights for considering possible duties to warn family members of patients with particular genetic risks that they may share these risks. Such analogical reasoning has been

used to support physicians' informing the partners of HIV-infected patients of their possibly increased risk for contracting HIV (depending on their actual behaviors and precautions); similarly, the family members of a patient with a genetic condition may be at an increased risk for developing or passing on to offspring particular genetically based risks (again, depending on a variety of factors such as inheritance patterns and actual paternity). Disclosure of various genetic risks or diseases and disclosure of one's HIV infection may have similar social, economic, and psychological consequences.

Yet there are difficulties in determining whether there is any legal duty to warn of genetic risks. First, commentators and legislatures have failed to reach consensus about how to apply the common law to cases about duty to warn regarding HIV. It is not clear whether there should be a duty to warn or just immunity from liability for physicians who warn partners of a patient infected with HIV; "state legislatures have addressed this issue variously by providing immunity without imposing a duty to warn, by imposing a duty to warn, or just imposing a duty to maintain confidentiality unless a court finds there is a compelling need for disclosure."<sup>53</sup>

Second, as seen above, informing family members of one risk, the genetically based health risk, imposes a variety of countervailing psychosocial and economic risks on them. The law provides no guidance to clinicians and researchers regarding their liability for exposing relatives to these risks. Third, clinicians may often overestimate their role in family members (or, for example, partners of HIV-infected individuals) becoming informed about relevant health risks. Depending on a patient's personal and cultural circumstances and the disease at issue, the patient's family members (or partners) may have other avenues to learn of their risks. Just as within gay communities it would be inappropriate to assume that sexual partners of HIV-infected gay men remain unaware of their own risks of contracting HIV unless their infected partners' physicians informed them; within some families there are other means of members' learning of heritable diseases. On the other hand, information about the genetics of eye diseases and diseases detected in the eye is only

emerging, so physicians have a greater responsibility to educate their patients who are willing to learn about these genetic risks and inheritance patterns. It is unclear in most cases, however, that if patients do not wish to learn about these risks or to share their own risk information with family members that physicians are ethically or legally justified in breaching patient confidentiality to warn family members. Again, development and advance promulgation of a clinician's (or researcher's) policy on disclosing information to identifiable third parties may make prospective patients (or research subjects) aware of the clinician's or researcher's conception of his duty to warn.

#### *Wrongful Birth and Wrongful Life*

Wrongful birth and wrongful life cases concern the failure to disclose information that would have been material to parents' reproductive decision-making and the prevention of serious harm. Indeed, the reluctance of courts to recognize wrongful life suits has largely rested on the question of whether a person who would not exist, but for the physician's failure to disclose information, should be permitted to sue for damages. Wrongful birth suits, brought by allegedly wronged parents who are denied material information, have generally been more successful.<sup>54-56</sup>

The first wrongful life claim was brought in the case of *Gleitman v Cosgrove* before *Roe v Wade*<sup>57,58</sup>; because the mother's pregnancy could not have been legally terminated had her physician informed her of the likely consequences of her rubella attack, her claim was denied. With *Roe*, however, the crucial link was made between physicians' provision of information and the possibility of preventing what may be viewed by some as harm. The possibility of physicians' liability for failure to disclose material information, including genetic information, was established. Indeed, wrongful birth cases like *Jacobs v Theimer*,<sup>59</sup> *Becker v Schwartz*,<sup>60</sup> and *Park v Chesin*<sup>61</sup> have served as the main source of guidance concerning the duties of physicians and clinical geneticists with respect to disclosure.<sup>56</sup> These precedents address concerns of the ophthalmologist who hesitates to tell his patients about an incidental finding of a genetic condition for fear of placing him in a difficult

position (eg, in which the patient may be required to share the information, contrary to his interests, with his employer or insurance company). Again, establishing and informing patients of the policy concerning disclosure of information to them and to others may mitigate risks of necessary disclosures and maximize beneficial use of disclosed information.

#### CONCLUSION

To serve the patient's best interests and to protect themselves from suit, physicians should practice a preventive ethics strategy and first inform the patient of what genetic information is likely to be discovered and revealed to the patient himself and to potentially at-risk relatives under particular circumstances.<sup>62</sup> The same is also true regarding disclosure of genetic information to institutional or commercial third parties.<sup>63</sup> Moreover, in accordance with a preventive ethics approach, it is worth noting that these genetics-related concerns are not novel; ophthalmologists have, for example, faced the conflict between their patients' interests in maintaining their licenses to operate motor vehicles and a state's requirement that doctors notify motor vehicle licensing authorities about patients whose medical condition may make their driving dangerous to themselves and others. Although patients' self-determined best interests and the preservation of the confidentiality of their medical information would point toward not reporting advanced visual loss, such as that caused by glaucoma, macular degeneration, or retinitis pigmentosa, these interests are here superseded by the interests of third parties in promoting public safety and professionals' interests in complying with the law.<sup>64</sup>

Ophthalmologists employed by companies to ensure a standard of adequate sight among their employees often find themselves in conflict between the interests and confidentiality of their patients and the obligations imposed on them by the terms of their own employment. Nongenetic information disclosed to a patient, or notes recorded on the medical chart and subsequently released to an insurance company, has led to the denial of health and life insurance.<sup>65</sup> Finally, with the emergence of preventive medical strategies, patients have been placed in positions in which their personal interests

conflict; a patient usually has an interest in making full disclosure to his health care providers so as to receive the best and most appropriately integrated care, but each self-authorized breach of privacy exposes the patient (and his relatives) to psychosocial and economic risks. Only reform of existing economic, political, and

social institutions that create these risks can fully eliminate them. Until that reform occurs, ophthalmologists, like all physicians and researchers, bear a responsibility to inform their patients and research subjects of these risks so that individuals can best protect themselves within existing institutional structures.

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