Patient Confidentiality vs Disclosure of Inheritable Risk: A Survey-Based Study

Edmund L. Erde, PhD; Michael K. McCormack, PhD; Robert A. Steer, EdD; Carman A. Ciervo, Jr, DO; and Gary N. McAbee, DO, JD

Background: As the field of genetic medicine advances and more tests for genetic diseases become available, a dilemma of legal and ethical importance will be increasingly encountered by family physicians. Protecting the confidentiality of a patient with a genetic disease when the patient’s family is at risk for inheriting the disease is a conflict that more and more physicians will be forced to address.

Methods: In March 2003, osteopathic family physicians in New Jersey were given a vignette in which a patient reveals that he has a genetic disease and demands that the information be kept confidential. The physicians were then given a 33-item questionnaire asking their opinions about disclosing an untreatable and a treatable disease to each of the patient’s children and their mother, a former spouse. Also, physicians’ opinions on larger issues were gathered, such as potential legal consequences of their actions and state laws in this area. Correlations between physicians’ opinions and their demographic characteristics were also explored. Data were analyzed using Pearson product moment correlations and repeated-measures analyses of variance.

Results: Physicians tended to agree that adult children should be told if the disease were treatable, and these physicians were willing to accept responsibility for deciding whom to tell. With regard to the characters in the vignette, physicians felt comfortable telling the 22-year-old daughter, were unsure about telling the 17-year-old son, and would not tell either a former spouse or the 12-year-old son. The respondents agreed that state laws should permit disclosure rather than require it, and they did not think state laws should forbid it.

Conclusion: The authors recommend that states draft a definitive public policy about when, how, and whether to disclose pertinent medical information to those at risk for inheriting a genetic disease.

Patients’ ethical and legal rights to confidentiality may at times interfere with the ethical responsibilities and legal duties of physicians. Advancements in genetic medicine and the increasing availability and sensitivity of genetic testing continue to provide a dilemma to physicians when diagnosed patients decline physician disclosure to relatives with an inherited predisposition to the disease. Surveys have studied how medical geneticists and genetic counselors view this conflict. Findings show that more than two thirds of medical geneticists think they have a duty to warn, a quarter of practitioners seriously considered overriding the patient’s refusal to notify at-risk relatives, and four of 206 actually did. The views of family physicians have not been studied to our knowledge.

The conflict is complex. Statutes, case laws, regulations, and policy statements from professional organizations contradict one another on this issue. For example, two appellate courts in different jurisdictions ruled that physicians have a duty to warn their patients’ family members about their risks of inheriting a genetic disease. However, definitions of how that duty is to be discharged vary greatly. In one Florida case, the physician must instruct the patient to warn family members about their genetic risks. In New Jersey, as a result of Safer v Pack, physicians are required to notify those at risk even if they are young children at the time of discovery of the risk. Case law may differ from state to state and may conflict with established moral principles related to physician-patient confidentiality.

The present study examined physicians’ opinions with regard to violating physician-patient confidentiality to inform a patient’s children and former spouse about the children’s risks of having a treatable vs an untreatable genetic disease. In addition, their opinions on larger issues were gathered, such as legal consequences of their actions and state laws in this area. Relationships between physicians’ demographic characteristics (ie, sex, age, ethnicity, religious affiliation, parental status, and years in practice) and their opinions were investigated.
Methods
The study was approved by the institutional review board of the University of Medicine and Dentistry of New Jersey–School of Osteopathic Medicine in Stratford. In March 2003, a 33-item questionnaire was mailed to 840 family physicians who were members of the New Jersey Association of Osteopathic Physicians and Surgeons (Monmouth Junction) at that time. Their names and addresses were obtained from the membership list for. The questionnaire comprised a case vignette followed by a series of questions, as well as a request for demographic information. In one version of the vignette, the disease is not treatable, and in the other, the disease is treatable if detected early. All physicians were asked to respond to both versions of the questionnaire. Respondents were given the option to answer the questionnaire anonymously and were instructed to return it by mail within 1 month. Return postage was paid for by the investigators.

Case Vignette and Questionnaire
On a first visit to a new doctor, Mr C, a 43-year-old, divorced, white man presented to Dr S complaining of abdominal discomfort. He was diagnosed as having an ulcer and treated symptomatically. During the history and examination, Mr C reported having an inheritable disease for which there is a commercially available genetic test that can be administered via a blood sample (it has no obvious tie to the chief complaint). Dr S does not know or treat anyone else in Mr C’s family. When queried, Mr C informs Dr S that:
- He knows the disease is rare and each son (aged 12 and 17 years) and his daughter (aged 22 years) have a 50:50 chance of inheriting it (ie, it is “autosomal dominant”).
- He knows it is fatal: his father and aunt died of it at a young age.
- He is bitter about having it and resents his father for passing it to him.
- He is estranged from his ex-wife and children.
- None of them knows about his disease.
When Dr S expresses a desire to inform them, Mr C states that he does not want anyone informed and orders the physician to refrain from informing them.

Respondents were asked whether they believed that disclosure would be grounds for a lawsuit for violation of confidentiality, whether licensing board members should support disclosure if the patient files a complaint, and whether New Jersey law should either forbid or require disclosure in such cases. Physicians were also asked to reveal their sex, age, ethnicity, religion, parental status, and years in practice.

Statistical Analyses
Because the physicians were asked to answer the same questions with respect to whether a patient’s genetic disease was considered to be untreatable or treatable, paired t tests were first calculated to compare mean ratings, and standardized descriptive statistics for matched groups were used to represent the effect sizes of the mean differences. Pearson product moment correlations were then used to ascertain whether any of the physicians’ background characteristics were related to their average ratings. Repeated-measures analyses of variance (ANOVAs) were next used to determine whether physicians agreed or disagreed with informing each of the patient’s three children or his ex-wife, and effect sizes (η²) were calculated for one-way repeated-measures ANOVAs. Finally, post hoc analyses were calculated to compare physicians’ mean ratings for the children and former spouse in the significant one-way ANOVAs. To control for the family-wise error rate incurred in calculating the number of analyses described above, Bonferroni adjustments were used to control for finding spurious significant effects (minimum Bonferroni-adjusted level of significance was set at .05 [two-tailed test]).

Results
Of the 840 questionnaires mailed to osteopathic physicians in New Jersey, 29 (3%) were undeliverable. We received 192 (24%) responses; however, only 165 (86%) surveys were complete (ie, all questions were answered). Respondent characteristics are presented in Table 1.

One hundred sixty-five physicians answered both sets of identical questions regarding the treatable and untreatable disease vignettes. From these responses, we derived a composite score to estimate physicians’ overall willingness or unwillingness to inform the patient’s children and former spouse. We reversed the rating scales for questions about whether breaching confidentiality is grounds for a lawsuit and whether New Jersey should forbid physicians from contacting relatives, and the ratings were then totaled to represent physicians’ overall willingness or unwillingness to disclose the confidential information. The Cronbach α values for the untreatable and treatable composite scores were .92 and .91, respectively, indicating that both total scores had a high index of internal consistency and were psychometrically reliable. Furthermore, the item correlations of the ratings for the treatable and untreatable composite scores were greater than 0.45 and significant beyond the .001 level (two-tailed test), even after applying a Bonferroni adjustment of .001 for all 13 comparisons. To facilitate interpretation, we divided the total scores by 13 so that the average ratings for both scores would also range from −2 to 2 (−2, strongly disagree; −1, disagree; 0, unsure; 1, agree; and 2, strongly agree).

Correlations
To ascertain whether any physician characteristics were associated with the average scores, we calculated two sets of correlations. The average scores for both genetic diseases (ie, untreatable and treatable) were correlated with the following physician characteristics: sex (0, man; 1, woman), age

SPECIAL COMMUNICATION

616 • JAOA • Vol 106 • No 10 • October 2006

Erde et al • Special Communication
All of the correlations and $t$ statistics in Table 2 are significant beyond the .001 level (two-tailed test) after applying a Bonferroni adjustment of .001. The mean differences between the ratings for an untreatable and treatable disease are not trivial. The majority of the effect sizes for the mean differences presented in Table 2 were greater than 0.30, which is the lower limit of a medium effect size according to Cohen. The average overall effect size based on the 165 physicians who answered all 13 questions for both forms of genetic disease (untreatable vs treatable) was 0.56.

The mean values in Table 2 suggest that surveyed physicians were generally unsure about whether the children or former spouse should be informed about a life-threatening disease when a patient instructed them not to tell anyone (mean [SD], −0.16 [0.96]). Still, they tended to agree that the children or former spouse should be told if the disease is treatable (mean [SD], 0.36 [0.91]).

### Family Member

To determine whether the physicians agreed or disagreed with informing different family members, we performed four repeated-measures ANOVAs. Table 3 presents the means (SDs) for the different types of family members by whether the disease was untreatable or treatable along with $F$ statistics and effect sizes ($η^2$) for the one-way repeated ANOVAs. Responses to questions about whether informing the family members would be morally or legally justified for untreatable and treatable diseases are included. The post hoc contrasts were conducted as Bonferroni-adjusted paired $t$ tests in which we set the level of significance at .05.

The results of the one-way repeated ANOVAs in Table 3 are significant beyond the .001 level (two-tailed test), using a Bonferroni adjustment of .001. Regardless of whether the disease was treatable or whether informing a family member was rated as morally or legally justified, the physicians made distinctions about which types of family members should be informed. The post hoc contrasts for the “morally justified, untreatable disease” category in Table 3 indicate that the physicians tended to agree that the 22-year-old daughter should be told. However, they were unsure about whether the 17-year-old son should be told, agreed that the 12-year-old son should not be informed. With respect to a treatable disease, the respondents would be more inclined to tell the 22-year-old daughter than the 17-year-old son and would be least likely to tell the 12-year-old son.

The respondents agreed that there was no legal justification to inform anyone if the disease were untreatable. If the disease were treatable, respondents agreed that the 12-year-old son should not be informed and would be more likely to tell the 22-year-old daughter than the 17-year-old son. The physicians tended to agree that adult children should be told if the disease is treatable, and these physicians were willing to accept responsibility for deciding whom to tell. With regard to the characters in the vignette, physicians felt comfortable telling the 22-year-old daughter than the 17-year-old son and would be least likely to tell the 12-year-old son.

Table 1

<table>
<thead>
<tr>
<th>Variable</th>
<th>Results*</th>
</tr>
</thead>
<tbody>
<tr>
<td>![Sex (n=188)]</td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Men" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Women" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Mean (SD) age, y" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="European American" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="African American" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Asian American" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Native American" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Other" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td>![Religious affiliation (n=184)]</td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Catholic" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Jewish" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Protestant" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Other" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td>![Parental status (n=185)]</td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Is a parent" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="Is not a parent" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td>![Years in practice (n=187)]</td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="&lt; 16 y" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
<tr>
<td><img src="https://example.com" alt="&gt; 16 y" /></td>
<td><img src="https://example.com" alt="Results*" /></td>
</tr>
</tbody>
</table>

* Data are given as No. (%) unless otherwise indicated.
year-old daughter, were unsure about telling the 17-year-old son, and would not tell the 12-year-old son.

**Comment**

The conflict between duties of confidentiality and duties to warn those at risk for inheritable diseases arose about a quarter of a century ago. At least one state had for a time empowered ethics committees to authorize breaches of genetic confidentiality in some cases. Amended versions of this statute have dropped that authorization.13

Physicians’ opinions about their legal and moral duties are relevant to establishing laws that are effective for patients and physicians. The present study demonstrates that in the face of moral and legal obligations, physicians are generally unsure about whether to inform a patient’s relatives about a potentially life-threatening genetic disease when a patient requests that the information remain confidential. Respondents did not believe that they had a legal duty to inform anyone of the risk of inheriting an untreatable disease, or to inform the youngest child or ex-spouse even if the disease were treatable. Although the respondents’ views on disclosure to the 12-year-old child is inconsistent with the court’s decision in *Safer v Pack*,4 it is consistent with therapeutic privilege and with “a right not to know,” both of which might pertain to a minor.14

![Table 2](http://jaoa.org/pdfaccess.ashx?url=/data/journals/jaoa/932054/)
These views may differ from case laws about the duty to warn relatives, but they are in line with statutes that routinely and generically prohibit disclosure of genetic information for the sake of privacy.

Accepted general standards of patient-physician confidentiality, including the Osteopathic Oath and the Hippocratic Oath, prohibit physicians from disclosing patients’ medical information without consent. This rule has been underscored by state and federal confidentiality laws, such as the Health Insurance Portability and Accountability Act of 1996 and the Arizona code. Still, there are noteworthy exceptions to the general standard. For example, disclosure to third parties without the patient’s consent has public benefit (eg, notification of third parties at risk of harm or exposure to communicable disease or public registries of cancer).

The American Society of Human Genetics (ASHG) has also addressed this issue. Incorporating some of the concepts from the President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, the ASHG devised a recommendation that ordinary duties of medical confidentiality should be overridden if each item in the following set of conditions holds:

- The patient refuses the professional’s urging to warn the relatives
- The harm is very likely serious and foreseeable
- Individuals at risk can be identified
- Early monitoring will prevent or treat the medical harms
- The harm of the disease outweighs that from the disclosure
- Only the information necessary to prevent or treat the inheritable disease is disclosed

The respondents in the current study applied this approach to the adult child. Although the criteria seem reasonable, they have serious flaws. One issue is the urgency of the warning. Some inheritable diseases have a late onset (eg, Huntington disease and familial Alzheimer disease), and there may be an opportunity for the patient to change his or her mind about informing relatives of risks. Second, advancements in medical science, such as new treatments for previously untreatable diseases, may change whether notification would help prevent harm. Further, knowing when the harm of disclosure outweighs the harm of the disease is vague, speculative, and individualized. It has been argued that this position changes the focus of the patient-physician relationship from personal to familial. Such a change in relationship may be very costly in terms of the impact on family interactions and on a patient’s trust in his or her physician. Patients may have good reasons for not wanting the information to be disclosed, and some of these reasons may not be evident to the physician. Finally, the physician may not take such reasons as seriously as the patient does.

The results of the current study cannot be directly compared with findings from earlier studies because the questions, contexts, and methods vary from one study to the next. We used questionnaires to evaluate hypothetical situations, whereas earlier studies were retrospective and asked practitioners what they did and how they felt about what they did. As mentioned previously, 69% of 206 medical geneticists in one study reported feeling obliged to warn relatives, against the patient’s direction, in hypothetical situations. Of the 123 geneticists who had been in such a situation, 25% had considered violating confidentiality, and 3% actually did so. In another study, 21% of 119 genetic counselors had strongly considered warning the relatives at risk despite their patients’ demands to the contrary.

Our study has several limitations related to the sample’s demographic composition. All respondents were family physicians who were members of the New Jersey Association of Osteopathic Physicians and Surgeons. They were selected because they are likely to treat multiple family members across generations. Also, our physician sample was predominantly male and of European American ethnicity. It is unclear whether
There is no medicine like hope, no incentive so great, and no tonic so powerful as expectation of something better tomorrow.
—Orison Swett Marden