

Legal considerations in clinical cancer genetics

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In the context of cancer genetics, familiar legal concepts such as due care, informed consent, and confidentiality become far more complex. Continuing education, a collaborative approach with colleagues and patients, and careful documentation will reduce the likelihood of an adverse encounter with the courts.

Diagnostic procedures, therapies, and basic biologic understanding in oncology evolve with disconcerting speed. As the medical landscape changes, the legal landscape changes with it. Physicians often worry that, with perfect hindsight, courts are likely to place the blame on them when the outcome of a case is not as desired. The actual measure is the state of medical knowledge and practice at the time of the encounter. Typically, this measure is established by expert testimony and written evidence such as professional journals.

When a question of negligence arises, courts generally apply a “reasonable physician” test to judge

whether the care provided was consonant with accepted practice. In essence, this standard asks whether a reasonable and prudent physician, practicing in the same specialty and using ordinary skill and care, might adopt a similar course under similar circumstances. Patients are entitled to expect that their physicians read the current literature, are skilled in their area of expertise, and use their skills to benefit their patients. Moreover, consumer activism and the emergence of patient autonomy as a central tenet of medical care have prompted the development of a “reasonable patient” standard by which physician-patient communication and informed consent may be measured. In the context of clinical oncology, these standards build on the traditional professional duties to assess the patient’s medical and family histories, conduct an appropriate physical examination, order tests as needed, and make a timely, accurate diagnosis. Next, the contemporary physician communicates the diagnostic findings and their implications, outlines the risks and benefits of medically acceptable responses, and collaborates with the patient in developing a care plan.

Expanding duties in diagnosis, treatment, and consent

As the hereditary potential of various cancers comes to light, physicians treating them must be prepared to identify patients who are candidates for further study. Accurate diagnosis is essential in determining which

KEY POINTS

Oncologists should be alert to the possibility that hereditary syndromes can be associated with the cancers they treat.

Medical and family histories should be sufficiently detailed to identify patients who could benefit from genetic investigation.

Informed consent for cancer susceptibility testing should include a discussion of the potential importance of test results to the health care planning of other family members.

Although a physician clearly has a duty to inform the patient about the hereditary potential of a diagnosis, the extent of the physician’s duty to warn at-risk relatives is not settled.

At all stages of care, candor and good faith can help deflect lawsuits.

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therapeutic options should be on the menu and which surveillance schedules should be observed thereafter. Failure to recognize a reasonably discoverable hereditary risk may subject the patient to inappropriate or harmful interventions, undermine the consent process, and compromise the care of other family members.

Genuine informed consent is predicated on an understanding of the diagnosis (causation, manifestations, likely course, inheritance patterns, if relevant) and the medically acceptable interventions (risks, benefits, and likely sequelae of each option, including the option of no treatment). The physician need not provide information on all possible aspects of the diagnosis or the proposed intervention but must provide information material to the deliberations of the patient, who must, in turn, make a considered judgment based on his or her goals and values.

Ironically, genetic testing itself often requires elaborate informed consent. Oncologists who are not well versed in the nuances of genetic testing and counseling will want to develop working relationships with genetic specialists to whom they can refer. Oncologists who directly provide cancer genetics services should look for

guidance to the literature, professional organizations, and possibly state statutes¹ concerning the appropriate elements of the consent process. For instance, the American Society of Clinical Oncology offers basic elements of informed consent for cancer susceptibility testing (Table 1).²

Genetic discrimination

Genetic discrimination is hardly new to people with obvious syndromes. This population can also testify that legislative attempts to curb discrimination, such as the Americans with Disabilities Act (ADA)³ and the Health Insurance Portability and Accountability Act (HIPAA),⁴ are at best partially successful. Indeed, the ADA does not specifically address genetic conditions, although the Equal Employment Opportunity Commission, which enforces the ADA, interprets the definition of disability to include them. The advent of presymptomatic genetic testing was closely followed by predictions of loss of employment, loss of insurability, and social stigma based on genetic test results. The paucity of documented instances of discrimination against asymptomatic individuals has not quieted the uproar. Cancer patients themselves may not readily ap-

preciate that once cancer has been diagnosed, subsequent genetic testing is unlikely to alter their employment or insurance prospects.

Since many individuals believe that genetic test results have greater potency than other medical information and are subject to greater abuse, patients may resist genetic testing despite its medical utility. They may request anonymous testing or ask that their genetic test results be withheld from their charts. Sequestering test results is inadvisable for several reasons. First, it is likely to compromise patient care. Memory is a notoriously unreliable source of medical information; patients are famous for getting it wrong and physicians come and go. Second, creative editing of a medical record invites fraud against insurers. Moreover, there are no legitimate public policy reasons to single out hereditary disorders as deserving of special protection. Better solutions include developing a universal-participation, community-rated system of health insurance and a requirement that rating for other forms of insurance be based on actuarially sound principles.⁵ Although it is legitimate to withhold investigational test results from a patient's chart, results of tests that have gained clinical acceptance should become part of the record.

Genetics: a family affair

Courts have long recognized that the professional obligations engendered by the physician-patient relationship can extend beyond the patient. The earliest of these cases concerned timely diagnosis of contagious diseases and warnings issued to third parties at risk. Next came the problem of dangerous patients, most famously addressed in *Tarasoff v. Regents of the University of California*.⁶ With the genetic era came cases establishing that the physician's obligation to recognize a heritable childhood disorder extended beyond the child to the parents, whose reproductive decisions might hinge on that knowledge.

TABLE 1

Basic elements of informed consent for cancer susceptibility testing

- Information on the specific test being performed
- Implications of positive and negative results
- Possibility that the test will not be informative
- Options for risk estimation without genetic testing
- Risk of passing a mutation to children
- Technical accuracy of the test
- Fees involved in testing and counseling
- Psychologic implications of test results (benefits and risks)
- Risks of insurance or employer discrimination
- Confidentiality issues
- Options and limitations of medical surveillance and strategies for prevention following testing
- Importance of sharing genetic test results with at-risk relatives so they may benefit from this information

Adapted from American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility.²

Inevitably, the logic of these cases has been brought to the question of what duty, if any, a physician owes to the relatives of an adult patient with a genetic disorder.

In the case of Mr. J, whose story is presented in the adjacent box, the physician has a duty to recognize and disclose to Mr. J the possibility that his cancer may have a hereditary component. Mr. J's coloring, his numerous irregular moles, and his early-onset malignancy are suggestive of familial atypical mole malignant melanoma syndrome (FAMMM) or dysplastic nevus syndrome. Mr. J's constellation of signs and symptoms is distinctive, and the similar physical appearance of his daughter suggests she too may be at risk. Once they know this to be the case, Mr. J and his physician should evaluate the option of molecular analysis, based on its possible utility (or lack thereof) for Mr. J, and its possible importance for his daughter. They should consider whether, in addition to existing pathology specimens, Mr. J wishes to bank DNA for his daughter's possible future use. Finally, Mr. J and his physician must have a frank discussion about Molly's situation. Even if FAMMM has not been genetically confirmed, Molly has a foreseeable risk due to her father's history and her own phenotype.

Upon Mr. J's death, his physician did not believe Mr. J's daughter had been informed of her risk. At this point, disclosure would not harm Mr. J, who, in any event, may eventually have agreed to tell Molly of her risk. Molly can take immediate action to protect herself from excessive sun exposure and can arrange to have regular evaluation of her moles. Moreover, Molly, as Mr. J's presumptive heir, has inherited his medical information as well as any tissue samples he has banked. The physician would serve the best interests of both Mr. J and Molly by disclosing this information.

Real-life dilemmas are rarely this simple. Families may be deeply divid-

Case study in cancer genetics: duty to disclose?

MR. J, A 40-YEAR-OLD WRITER, is undergoing treatment for malignant melanoma. He has a fair complexion with blue-green eyes; red hair; abundant freckles; and many moles of irregular size, shape, and color. Mr. J, a single parent, has raised a now 17-year-old daughter, Molly. He has not told his daughter about his diagnosis, not wanting to burden her at this stage of her life. He maintains he'll tell her "when she's older." Despite aggressive therapy, Mr. J's condition worsens, and he is hospitalized. Molly often visits him, eventually meeting her father's physician. The physician notices that Molly has her father's coloring and a similar pattern of moles. Mr. J says he has told Molly he has cancer but has not told her what type. A few months later, Mr. J dies suddenly and unexpectedly. The doctor does not believe Mr. J told his daughter about his diagnosis or her potential risk.

ed, patients may be hostile, and physicians rarely know the identity and location of relatives. In most cases, informing the patient (and creating an adequate record of the conversation) will satisfy a physician's obligation to other family members. In those rare instances in which physicians wonder whether disclosure without permission may be warranted, it can be useful to ask which course of action they would rather explain to a jury.

Consensus of the courts

To date, only two appellate courts have directly addressed the issue of such a professional obligation. In *Pate v. Threlkel*,⁷ a daughter sued her mother's physician when the daughter was diagnosed with advanced medullary thyroid cancer (MTC), 3 years after her mother had been treated for MTC. The daughter alleged that had her mother been warned about the hereditary nature of this cancer, she would have told her daughter, who, in turn, would have been diagnosed earlier. The lower courts rejected the case on two grounds: the daughter had no professional relationship with her mother's physician, and since the physician had not created the risk to the daughter, he had no duty to mitigate it.

On appeal, the Florida Supreme Court considered whether the physician's treatment of the patient satis-

fied the standard of care at the time treatment was given. If the prevailing standard was to warn the patient (in this case, of the hereditary nature of her disease), that standard logically must be for the benefit of the patient's children as well as the patient. Accordingly, the intended beneficiary of that warning, the daughter, would have a cause of action against a physician who failed to warn.

The court then concluded:

*To require the physician to seek out and warn various family members of the patient's family would often be difficult or impractical and would place too heavy a burden upon the physician. Thus, we emphasize that in any circumstances in which the physician has a duty to warn of a genetically transferable disease, that duty will be satisfied by warning the patient.*⁸

Very shortly thereafter, an appeals court in New Jersey dealt with a similar, but much more attenuated, situation. In *Safer v. Estate of Pack*,⁹ a 36-year-old woman diagnosed with metastatic familial adenomatous polyposis (FAP) sued the estate of her father's doctor for his alleged failure to disclose during a long course of treatment that her father had FAP. As in the Florida case, the trial court rejected the suit on two grounds: lack of a professional relationship and lack of a public health risk giving rise to a duty to warn. The appeals court overturned the lower court,

agreeing with the *Pate* court that the standard of care at the time services were rendered should govern. However, the appeals court went on to say:

*We decline to hold...that in all circumstances the duty to warn will be satisfied by informing the patient. It may be necessary...to resolve a conflict between the physician's broader duty to warn and his fidelity to an expressed preference of the patient that nothing be said to family members.*¹⁰

Clearly, both courts reaffirmed that the prevailing practice at the time of the encounter was the important measure of conduct. Prevailing practice will govern such elements as the recognition that the disorder may be hereditary, the measures taken to confirm or rule out the diagnosis, the management alternatives proposed, and the scope of the conversations held with the patient. However, these opinions are (potentially) split on whether a physician might in some instances have a duty to warn relatives directly if a patient refuses to do so. Indeed, "prevailing practice" on this point has not been established; medical authorities are similarly divided.

According to the *American Medical Association Code of Medical Ethics* opinion on disclosure of familial risk in genetic testing, "Physicians should identify circumstances under which they would expect patients to notify biological relatives of the availability of information related to risk of disease. In this regard, physicians should make themselves available to assist patients in communicating with relatives to discuss opportunities for counseling and testing, as appropriate."¹¹ *The American Society of Human Genetics* statement on *Professional Disclosure of Familial Genetic Information*¹² suggests disclosure should be made over the objections of a patient only when the harm of silence outweighs the harm of disclosure, whereas the *American Society of Clinical Oncology Policy Statement Update: Genetic Testing for Cancer Susceptibility* maintains that the ob-

ligation to relatives at risk, if any, is "best fulfilled by communication of familial risk to the person undergoing testing."²

Additional considerations

The HIPAA Privacy Rule permits physicians, nurses, and other health care providers to use or disclose protected patient health information for treatment purposes without patient authorization. Currently, the US Department of Health and Human Services interprets this clause as permitting use or disclosure "to treat a different patient."¹³ Although this startling interpretation may not prevail, the Privacy Rule also includes a proviso that confidentiality may be breached to avert a serious threat to the health or safety of third parties.¹⁴

After a physician has broached the topic of a possible hereditary cancer, it is often helpful to dictate a letter to the patient, setting forth the basic information covered in the conversation. This letter might include the reasons hereditary cancer is suspected, the possible diagnostic alternatives, the ways in which a firmer diagnosis could aid in managing the patient's care, and the potential importance of this information to relatives at risk. This measure serves two important purposes: it documents the transmission of the information by the physician and provides the patient an opportunity to evaluate the information under quieter circumstances.

Ordinarily, a physician is not expected to follow-up with a patient to determine whether or not relatives have been notified. However, if the patient's response suggests that notification is in doubt, the physician may have an ethical, if not a legal, obligation to talk again about its importance. The physician might emphasize the generous nature of the act, the opportunity to enhance family unity and support, or the patient's moral obligation. The physician might offer to help the patient to explain or provide

literature to be forwarded to relatives.

Occasionally, a physician will learn that a diagnosis has been deliberately misrepresented to other members of the family. If this misrepresentation can cause harm to others, firmer measures may be indicated. Consultation with an ethicist and/or an attorney may be necessary to determine the proper approach. In such an instance, it is particularly important to emphasize one's commitment to the welfare of the patient and maintain open lines of communication.

Conclusion

The challenges posed by clinical cancer genetics are permutations of the traditional legal duties of due care, informed consent, and confidentiality. Oncologists must be alert to the possibility of hereditary cancer and must apprise their patient—ideally in writing—of the implications for themselves and their relatives and themselves if a hereditary cancer is suspected. Therapies and surveillance should reflect genetic potential. Careful genetic counseling will assist patients in determining whether genetic testing is in their best interest. Physicians may exercise professional judgment in the matter of warning relatives at risk for genetic disease. At all stages of care, candor and good faith are potent deflectors of lawsuits.

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Commentary

Protecting your patient and yourself

By Judith Mackarey, JD

THE ABOVE PAPER by Anderson and Haidle, "Legal Considerations in Clinical Cancer Genetics," discusses the legal issues related to cancer genetics and the complexities of hereditary forms of cancer. Complex moral and legal quandaries can conflict with physician-patient confidentiality when relatives are at risk of genetic disease and the patient resists disclosure of that information.

The article explains the potential liability with respect to a duty to warn relatives who may be genetically at risk from hereditary cancers. Contrasted with this, and of utmost importance, is ensuring the care and treatment of the patient by the physician and preserving the doctor-patient privilege.

From a legal perspective, this is the proverbial "rock and a hard place," since liability may also result from disclosure of confidential information against the direct wishes of the patient. Courts have recognized the right of patients to sue physicians for unauthorized disclosure of information based on invasion of privacy or breach of the confidential relationship.

As the authors note, although confidentiality of patient information is not absolute, any potential disclosure of information should be

narrowly tailored to avoid violation of protected privacy rights. The American Society of Human Genetics recommends that before disclosing a patient's information to an at-risk relative there should be evidence that:

- attempts to convince the patient to disclose the information have failed;
- the at-risk relative is identifiable;
- the harm to the relative is highly likely to occur;
- the disease is preventable, treatable, or early intervention will substantially reduce the genetic risk.

A high degree of harm must seem likely to occur *before* the legal obligation to disclose is triggered, which may be difficult to show. That is, when assessing potential harm to an unknown relative based on results of genetic testing, it's not clear whether a sufficient level of injury can be determined in advance. In light of this, the physician certainly should discuss the risks to the patient and the patient's family and encourage disclosure to potentially affected family members. The process of educating the patient and letting the patient make the final decision whether to tell family members may be sufficient and may avoid violating patient confidentiality.

To the extent that disclosure by the physician is a possibility in any situa-

tion, I would recommend that the patient be informed prior to pre-test counseling of the circumstances under which genetic information may be disclosed to relatives without the consent or approval of the patient. With notice in advance, patients would at least be informed about the potential for disclosure against their wishes. This also protects the physician, since consent by the patient constitutes a waiver of the privilege and preserves the doctor-patient relationship. This may not be the perfect solution, since it could result in a patient's reluctance to undergo genetic testing, knowing that the information could be revealed to family members. On balance, however, it communicates the important facts to the patient, who is able to make an informed decision. Absent this, the patient's care and treatment may be compromised and the relationship with the physician destroyed.

Judith Mackarey, a partner with Mackarey & Davidson, PC, is a health law attorney who practices in the greater Philadelphia area. Her practice encompasses a range of legal issues that confront the healthcare professional, from general corporate and employment law matters to health care issues. Ms. Mackarey is also an Associate Professor at Widener University, where she teaches a course entitled "Healthcare and the Law."