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Should Physicians Inform Relatives of Genetic Risks, Without the Consent of the Patient?

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March 5, 2009 — Do physicians have an obligation to disclose genetic-risk information to relatives, even without the consent of the patient?

This is the question posed — using *BRCA1/2* mutation in breast cancer as an example — in a report published in the March issue of the *Lancet Oncology,* and explored here in a couple of in-depth interviews with *Medscape Oncology .*

Protecting patient confidentiality and protecting family members from potential harm presents a dilemma for healthcare professionals. The results of genetic testing reveal information not only about the patient, but about their relatives as well, and the information could be of great value to them.

For instance, if a women with breast cancer has the *BRCA1/2* mutation, her siblings might also carry it. Although the patient can certainly inform her siblings that she is carrying the *BRCA1/2* mutation, current laws in the United Kingdom prevent her physician from doing so without the consent of the patient. The report questions whether breaking confidentiality outweighs the good that could be done by a doctor informing a patient's relatives, and whether the time has come to change patient confidentiality laws.

"There are, at times, ethical hierarchies based on critical values," said Ross McKinney, Jr, MD, professor of pediatrics, molecular genetics, and microbiology, and director of the Trent Center for Bioethics, Humanities and History of Medicine at Duke University School of Medicine, in Durham, North Carolina. "In this case, the 2 values in question are autonomy — the right to personally control your own destiny, which in this case includes privacy — and the right to optimal health and safety."

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"The problems develop when the decisions affected by those values overlap and the results of the decisions do not align," he told *Medscape Oncology*.

In the case of the *BRCA* mutation, what happens when a patient's right to privacy (not wanting people to know about an increased risk for breast cancer) imposes itself on her sister's right to optimize her healthcare (breast cancer can be treated more effectively if caught early, or even prophylactically by mastectomies) and know her risk? "As a society, we are in the uncomfortable position of ordering these values hierarchically, which is a problem when we don't have a consensus," said Dr. McKinney.

"In truth, I think most people in the United States would place a higher value on the person's right to critical information that can keep them healthier," he added. "However, we're unsure enough that we'd like to find a win–win solution, like 'let someone else — a third party — notify my sister so that I don't have to reveal my own risk.' But sooner or later, that discussion between sisters will probably happen, no matter who hears it first."

Breaching the Law

Although current laws in the United Kingdom are designed to protect patient confidentiality, there are well-recognized justifications for breaching it. According to Richard Ashcroft, PhD, professor of biomedical ethics at Queen Mary, University of London, in the United Kingdom, the 3 main reasons for breaching confidentiality are the consent of the patient, the necessity of preventing a significant risk for serious harm to a third party, and promotion of a significant public interest.

In the case of HIV infection, the situation is more clear cut, because notification of sexual partners is necessary to prevent a significant risk for serious harm and further spread of the virus. The situation of genetic mutations, such as *BRCA1/2*, is far more ambiguous. In the *Lancet Oncology* report, Dr. Ashcroft points out that although the risk posed by an adverse *BRCA1/2* mutation is serious, it is not usually imminent, so there is time to discuss disclosure with the patient. There is also the complex issue of the right not to know one's genetic risk.

HIV infection is an example of a "safe-harbor entity," explained Dr. McKinney. "If I have a newly identified HIV-infected patient, I am required to notify the state, who then tracks their sexual contacts."

With HIV cases, it has been determined that the health of others and their right to treatment circumvents the right of the patient to privacy concerning his or her HIV diagnosis. "However, our attempt at a win–win [situation] means that the doctor doesn't do the notification of partners, thus protecting the physician–patient relationship," said Dr. McKinney. "In reality, we've codified a system that values the at-risk person's right to receive treatment over the privacy of the infected person."

In the United States, patient confidentiality is already riddled with exceptions, explained Hank Greely, JD, professor of law at Stanford University, in Palo Alto, California, and an expert on the legal, ethical, and social issues surrounding health law and the biosciences. American doctors "are obligated to report a number of otherwise confidential facts about patients to the state, from the existence of some infectious diseases, to evidence of child or elder abuse, to pesticide poisoning, to the presence of gunshot or knife wounds," he told *Medscape Oncology*. "They have also been obligated to report to third parties facts learned from a patient that a reasonable doctor would realize would put a third party at grave risk. These cases are mainly about family members and contagious diseases."

Court Cases Leave Issue Open

A number of cases involving confidentiality issues have been heard in courts around the United States that illustrate some of the complexities involved, Mr. Greely said in an interview. In 1 court case in Tennessee, a man died from Rocky Mountain spotted fever, which is tick borne and not spread by human-to-human contact. His wife also contracted it and died, and his son sued the doctor for not warning his mother. "He won the case. The Court held that the doctor should have warned the wife that, as the husband had suffered a bite from an infectious tick, she might have been bitten, too, and should watch out for symptoms," said Mr. Greely.

Court cases specifically involving genetic disclosure are uncommon, Mr. Greely said, and 2 cases related to cancer reached somewhat different conclusions. The first case, heard by the Florida Supreme Court, involved a woman diagnosed with medullary thyroid carcinoma whose mother had been diagnosed with the same disease 3 years earlier. The patient claimed the doctor should have known it was highly genetic and warned her. She then might have discovered the cancer earlier when it was more treatable.

"The lower court threw out her claim on the ground that the doctor owed her no duty," said Mr. Greely. "The Florida Supreme Court reversed it, saying he could owe her a duty, but that whether he did or not was determined by the relevant standard of care."

But the Florida case, without expressly addressing confidentiality, put in a strong limitation on the claim, stating that "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."

The second case, explained Mr. Greely, was heard by the New Jersey Appellate Division and involved a woman with colon cancer who learned that she had familial adenamotous polyposis. Her father had been treated many years earlier for colon cancer and she sued the estate of the physician (now deceased), claiming that he should have informed the family of the genetic risk.

The court rejected the limitation set previously by the Florida case, which said that "in all circumstances, the duty to warn will be satisfied by informing the patient. It may be necessary, at some stage, 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 about the details of the disease."

However, the Court went on to note that if evidence was produced to show that the physician had received instructions from his patient not to disclose details of the illness or of genetic risk, "the Court will be required to determine whether, as a matter

of law, there are or ought to be any limits on physician–patient confidentiality, especially after the patient's death, where a risk of harm survives the patient, as in the case of genetic consequences."

Although it might be surprising to many physicians in the United States, patient confidentiality laws probably do not bar them from disclosing genetic risks to family members, explained Mr. Greely. "Whether it is just the patient or whether the duty goes further remains to be determined."

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Dr. McKinney agrees. "There are no 'right' answers," he said, and this is the reason that these issues are controversial. "But I tend to prefer something like the HIV notification model. It's clumsy, but orders the values in the same way I would."

The Exception Rather Than the Rule

The American Society of Human Genetics has issued a policy statement on this topic, which Wylie Burke, MD, PhD, professor and chair of the Department of Bioethics and Humanities, University of Washington, in Seattle, feels provides a very balanced review.

"This statement emphasizes that every effort should be made first to encourage the patient to disclose risk to relatives," she said. "Only if that effort has failed and the benefits of disclosure outweigh the harms should the doctor inform relatives directly. This judgment has to be made on a case-by-case basis."

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But instances where patients are unwilling to share information with relatives seem to be the exception rather than rule. "My impression from my own practice and from talking to colleagues is that family members rarely refuse to disclose information about shared risk to their relatives," said Dr. Burke. "Certainly, they may wish sometimes to keep their own results private, but as a general rule, family members are willing and often eager to let relatives know about a genetic risk running in the family or an opportunity to have genetic testing."

Genetic information should be disclosed if a clear health benefit can be achieved, but physicians need to be rigorous in justifying their action, explained Dr. Burke. "The patient should be told that the disclosure will occur, and why — and should be offered as much support and sympathy as possible."

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