LETTER TO JMG

Should healthcare providers have a duty to warn family members of individuals with an HNPCC-causing mutation? A survey of patients from the Ontario Familial Colon Cancer Registry

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Background: As genetic testing becomes more common and increasingly intertwined with medical care, the issues of genetic privacy and doctor–patient confidentiality are being examined. Hereditary non-polyposis colorectal cancer (HNPCC) is a genetic predisposition to colorectal and certain other cancers. Effective screening that can prevent colorectal cancer is an important incentive for genetic testing.

Methods: A survey regarding the duty to warn family members of the risks associated with an HNPCC-causing mutation was mailed to 227 participants in the Ontario Familial Colon Cancer Registry (OFCCR). To our knowledge, the opinions of patients on this subject have not been reported previously in the literature. Responses were analysed quantitatively using the SAS system and qualitatively by the review of written comments.

Results: Completed surveys were returned by 105 participants, with a response rate of 46.3%. The majority felt a personal responsibility to warn relatives, but there was no significant agreement that doctors or genetic counsellors should have a duty to warn relatives without a patient's permission.

Conclusions: Patients undergoing genetic testing for HNPCC generally understand that relatives could benefit from being informed of genetic risk, but may not be willing or able to inform each family member. Healthcare professionals should engage patients in a discussion of familial implications before genetic testing. An agreement should be formulated regarding which of the relatives should be informed. Patients should be encouraged to personally disseminate the information, given the unrealistic burden on practitioners to perform this task and patients' preference for control over the information.

Genetic information is distinct from much medical information because of its familial nature¹ and its applicability to predicting future health.² It has been suggested that with increased availability of health-related genetic information, individuals should be aware of and accept responsibility to share genetic test results with their family members.^{3–5} If a patient does not inform at-risk relatives about a genetic condition that could confer serious harm, the duty to warn may potentially be extended to the healthcare provider.^{4–7} More clarity is needed to define if and when there is a legal and/or ethical duty to warn family members at risk, especially since this violates autonomy and confidentiality and would probably present an impracticable burden to health professionals.

Genetic testing should be performed with informed consent after meeting with a genetics professional to discuss risks,

benefits and limitations. Certain laws have been enacted to protect genetic privacy.^{2–9} The Health Insurance Portability and Accountability Act¹⁰ states that “genetic information” cannot be used by health plans for discrimination. As genetic testing continues to provide information relevant to medical care, it will become increasingly difficult, and perhaps less beneficial to patients, to keep the results confidential.

Healthcare providers have been encouraged to discuss with patients the implications of disclosing test results to family members during pretest counselling.¹¹ Acceptance of personal responsibility should reduce the risk of disagreements once results are available. Also, previous research has shown that patients prefer to have control over the process of disclosing genetic information to their relatives.⁹

The aim of this study was to survey participants of the Ontario Familial Colon Cancer Registry (OFCCR) regarding their duty to warn family members about a hereditary non-polyposis colorectal cancer (HNPCC)-causing mutation. The survey was designed to determine opinions about personal responsibility, barriers to communication and the responsibility of health professionals, particularly if the participant was unwilling or unable to inform relatives.

METHODS

A one-page survey was mailed to 227 participants in the OFCCR, a population-based registry of colorectal cancer patients in Ontario, Canada. As described previously in detail,¹² individuals diagnosed between 1997 and 2000, aged 20–74 years, were recruited. Participants received genetic counselling and were offered genetic testing. At the time of mailing the survey, 19 were identified as carriers of an HNPCC-causing mutation and 21 were classified as “high risk” based on meeting Amsterdam or modified Amsterdam criteria, but had test results suggesting the absence of a mutation or had not yet received results. The survey was mailed to an 197 additional probands classified as “intermediate risk” based on family history of HNPCC-related cancers, personal history of colorectal cancer at or before age 35 years, certain tumour pathology or Ashkenazi Jewish heritage.

Participants were asked to indicate their agreement to statements using a five-point Likert scale and to provide written comments. The SAS system was used to calculate frequency distributions for Likert-scale responses, and the proportion agreed was calculated with 95% CIs. Finally, a content analysis was completed from open-ended statements or comments provided by participants.

Abbreviations: HNPCC, hereditary non-polyposis colorectal cancer; OFCCR, Ontario Familial Colon Cancer Registry

RESULTS

A total of 105 OFCCR participants returned the survey, with a response rate of 46.3%. Five surveys were returned incomplete because the participants were no longer at that address and five other participants were reported deceased. Of the high-risk probands with no mutation identified, 52.4% responded to the mailing. Of the mutation carriers, 50% responded.

The proportion of respondents who agreed with the statement "If I am told I have an HNPCC mutation, it would mean that my family members could also have an increased chance to develop colon cancer" was 94.3% (95% CI 89.5% to 98.7%, $p < 0.001$). Of all survey respondents, 93.3% (95% CI 88.5% to 98.1%, $p < 0.001$) thought that it was their duty to tell all at-risk family members and 95.2% (95% CI 91.1% to 99.3%, $p < 0.001$) thought that it was their duty to encourage regular screening examinations (eg, colonoscopy). All nine mutation carriers who responded agreed that they had a duty to tell family members about the mutation and about their possible risks of cancer.

The vast majority, 96.2% (95% CI 92.5% to 99.9%, $p < 0.001$), indicated that they would tell their family if they were diagnosed as having cancer. The total number of respondents who would tell their family members about a negative HNPCC result was 87.6% (95% CI 81.3% to 93.9%, $p < 0.001$). The proportion of respondents who thought that difficult relationships would prevent them from informing family members about an HNPCC-causing mutation was 18.5% (95% CI 11.0% to 25.9%, $p < 0.001$); 28.9% (95% CI 20.1% to 37.6%, $p < 0.001$) indicated that lack of contact would prevent disclosure. Most participants (84.3%) denied having issues telling family members. All 15 respondents identified particular relatives whom they would not be able to tell and specified the reasons.

The proportion of participants agreeing that they would give permission for a genetic counsellor or doctor to warn relatives was 73.5% (95% CI 65.0% to 82.1%, $p < 0.001$). There was no significant agreement with the statement "I would want my physician or genetic counselor to tell certain family members (whom I did not feel comfortable telling) that I had an HNPCC mutation, even without my permission." The proportion agreeing was 55.8% (95% CI 46.2% to 65.3%, $p = 0.1197$), with 44.2% selecting "strongly agree". Of the nine mutation carriers who responded, six agreed, two disagreed and two were neutral. When asked whether they would have sought genetic counselling knowing that their provider could inform their family members of a mutation without permission, 67.0% (95% CI 57.9% to 76.0%, $p = 0.0003$) agreed, with 55.3% strongly agreeing.

Content analysis

A total of 40 participants (38.1%) provided written comments, revealing three major themes. The majority ($n = 27$) strongly believed that disclosure of test results to family members is important and is a personal responsibility. Some respondents indicated that they would want their relatives to be informed, even if directly through their doctor without their permission. Two believed strongly that disclosure should only be through the patient who is tested. Seventeen additional respondents were hesitant to contact family members owing to communication barriers or lack of contact. The strongest responsibility was felt towards immediate family or relatives with whom they were in contact.

DISCUSSION

Almost all respondents (94.3%) demonstrated an understanding of the implications of a positive genetic test result. This suggests that participants gained knowledge from genetic counselling sessions, as supported by other studies.¹³ Participants thought that it was a personal duty to warn family

members about the risks of cancer and screening. A common reason cited by OFCCR participants for having genetic testing was to protect their family members.¹⁴

In this study, 18.5% of participants identified barriers to warning at-risk family members. Content analysis identified various reasons for non-communication, including "no contact", "relative being elderly", "living at a distance" and "family doesn't want to discuss issue". These barriers are similar to those found among individuals with *BRCA* results.¹⁵ One proband indicated reluctance to tell parents so as "not to worry them" and children because they were "too young to understand". An interesting dynamic emerges between protecting loved ones' physical health and choosing to protect emotional or mental health by not disclosing information of concern.¹⁵

Most participants (87.6%) would tell family members about a negative genetic test result. A "negative" result is not completely informative. A mutation could be missed by the testing methodology or could be present in another gene. Genetic counsellors stress these limitations during pre- and post-test counselling. Studies should continue to investigate whether individuals inform their relatives of the residual population risk for colon cancer, and/or increased risk based on the family history, and whether they encourage appropriate cancer screening.¹⁶

If there were certain relatives whom participants could not or would not tell about an HNPCC-causing mutation, 73.5% would give permission for the healthcare provider to disclose the information. In this case, the duty to warn family members is recognised, but all the responsibility does not necessarily fall on the proband. At a familial cancer clinic in Australia, the proportion of relatives who received predictive testing increased from 23% to 40% when providers sent letters to at-risk relatives with permission from the proband.¹⁷

In a survey of medical geneticists, 60% reported patients who refused to warn an at-risk family member.¹⁸ Of 259 genetic counsellors surveyed, 119 reported having patients who would not inform an at-risk family member. Twenty-five counsellors "seriously considered" warning the relative without consent, and one actually disclosed the information, although the relative was also a patient of the counsellor.¹⁹ In this study, 55.8% of respondents indicated that they would want their doctor or genetic counsellor to warn family members; however, most were willing to warn their own relatives. If healthcare providers assumed a duty to warn family members without a patient's permission, 15.3% of respondents were unsure whether they would have sought genetic counselling.

In written comments, 27 respondents suggested a strong belief that family members should be informed of the risks associated with an HNPCC-causing mutation. One respondent, whose family was classified at intermediate risk, wrote, "Cancer or its possibility is nothing to hide. If there is any chance that I have a HNPCC mutation then I feel it is my responsibility to inform all my family members. Recognizing that there are some families where certain people do not correspond in any fashion puts the onus on medical professionals to communicate with such members ... To save lives all family members of someone having the HNPCC mutation must be informed by whoever is able to do so (family member, physician, or genetic counselor) ... Being notified of the possibility TWICE, in lieu of not being notified at all, is a much better solution to this problem."

Other respondents indicated that "the matter becomes a toss up between my security and someone else's threat to life, and someone else's threat to life weighs more heavily than my security" and "My cancer should help others and I have no right to stop this." These instances may relate to the population

surveyed. Participants in cancer registries often cite a duty to help others as their incentive to donate time and energy to participate, even in the absence of a clear personal benefit.

Seventeen respondents suggested hesitance to tell family members or barriers to communication. Statements included "The one who is 'telling' has invaded the 'hearer's' privacy" and "In this technological age who can be assured that the information would remain confidential?" There were no obvious differences between responses of mutation carriers and those who tested negative, although the sample size of mutation carriers was small ($n = 9$).

Two respondents specifically indicated that they were opposed to disclosure of genetic test results to family members without their consent. One wrote "it would do damage to the doctor/patient relationship". Another indicated "The privacy of information protection act does not allow physicians to overstep confidentiality."

It has been proposed that the following four conditions be met before breaching genetic confidentiality: (1) the condition is serious; (2) has a high recurrence risk; (3) occurs in identifiable family members; and (4) effective treatment options are possible.^{4, 20} According to the 1998 American Society of Human Genetics statement, there should be no legal duty to violate confidentiality to warn family members, but the practice is ethically acceptable as a last resort.

What is a family?

If there is to be a duty to warn family members at risk, the scope of family must be defined. Some have never met half-siblings, and extended family members often reside in distant locations.² This is especially relevant for those caring for multiple members of the same family.²¹

The right not to know

Some individuals choose to decline or postpone testing on the basis of extraneous life stressors²² or fear of insurance discrimination.²³ The receipt of information will not necessarily be life saving. A genetic mutation is not always predictive of disease and treatment or screening is not always proven. Also, the proband does not actively create risk to relatives since the genetic mutation, if present, was inherited at conception.²⁴ Ethically, the difficulty with informing family members about a mutation is that their right to have access to informed consent is taken away.¹

Pertinent court decisions

In *Pate v Threlkel*,⁶ the plaintiff was diagnosed with medullary thyroid cancer and filed charges against her mother's doctor, who previously treated her mother for the same condition. Citing state confidentiality laws, the Supreme Court of Florida emphasised that the verdict "should not be read to require the physician to warn the patient's children of the disease". Duty would be satisfied by warning the patient herself.

In *Safer v Estate of Pack*,⁷ a New Jersey court ruled that a doctor had a more direct duty to warn. The plaintiff's father was treated for colon polyposis, a condition that clearly suggests a hereditary causation and is life threatening if left untreated. Thirty years later she was diagnosed with the same condition. The court ruled that the doctor's duty to warn was extended beyond his patient to immediate family members.

Defining obligations

A review of the *Safer v Estate of Pack* case²⁵ revealed that the court did not characterise the "immediate family" to which the doctor held a duty to warn or define what "reasonable measures" the doctor needed to employ. Other authors have

added that the court did not consider the possible impractical burden to the practitioner.¹¹

Communication about HNPCC

Although individuals at risk for HNPCC may decline testing, they should receive counselling to review cancer risks and screening. In a prospective study, no harmful emotional consequences were identified following HNPCC genetic testing.²⁶ However, long-term follow-up was not performed and little was asked about familial interaction, an important factor for disclosure.²⁷ For women tested for mutations in the *BRCA* genes, disclosure to children depended on the age of the child and the communication style of the mother.²⁸ In another study, sisters who did not disclose *BRCA* results cited emotionally distant relationships as the major barrier.²⁹

Importance of genetic counselling

It is well recognised that genetic testing should occur within the context of a comprehensive counselling protocol.^{3, 26, 30} Individuals may feel guilty or want to avoid hurting family members or increasing anxiety, and therefore dread the act of disclosure.¹⁵ Some individuals may wait to pass along genetic knowledge until a young adult has made plans for life.¹⁵ Genetic counselling can provide information and guidance before, during and after testing, as well as provide perspectives on the importance of genetic information to a family. One study showed that counselees' concern about close relatives decreased from 81% to 70% after counselling.³⁰

Study limitations

The results of this survey may not be generalisable to other patients undergoing genetic testing. All OFCCR participants have been diagnosed as having colorectal cancer, are of a specified age category and are mostly of similar socioeconomic status. Also, most participants have not been identified as HNPCC mutation carriers. Patients in Ontario with universal healthcare may be less concerned about insurance than American patients. Individuals who are not part of a registry may have different views about the duty to warn family members; however, our findings are similar to those reported among clinic samples. Further research should examine the views of more mutation carriers and unaffected individuals who are not in a registry.

Most participants from a colorectal cancer registry in Ontario felt personal responsibility to warn relatives of the risks associated with an HNPCC-causing mutation. There was no significant agreement that doctors or genetic counsellors should have a duty to warn family members without a patient's

Key points

- This study is a survey of patients from the Ontario Familial Colon Cancer Registry regarding the duty to warn family members about the risks associated with a hereditary non-polyposis colorectal cancer-causing mutation.
- To our knowledge, the opinions of patients from a survey such as this one have not been published previously.
- Patients with colorectal cancer participating in a registry recognise the importance of warning their family members, and prefer to have control over disclosure to family members, which is also preferable to practitioners given the staggering responsibility and resources that would be required for healthcare providers to inform patients' relatives about genetic test results.

permission. The best course of action is for health professionals to engage probands in a discussion of familial implications before genetic testing. An agreement can be formulated between provider and patient as to which relatives will need to know the information and how they should be informed. Owing to the likely unrealistic resources required for provider notification, patients should be counselled and encouraged about the personal duty to warn family members.

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CORRECTION

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The journal apologises for an error that has occurred within this paper. In the abstract on page 64 (under Results, line 3) and in the key points box on page 67 (line 5) the term "limit of detection" should read as "LOD".