Who's in Your Genes: A Physician’s “Duty to Warn” Patients’ Relatives about Genetic Risk

Colin Meyer-MacAulay (Meds 2012)
Faculty Reviewer: Dr. Victoria Siu

The genetic information of a patient as it relates to disease risk may prove invaluable to blood relatives who may wish to use it to make informed decisions about health care or reproduction. On the other hand, diagnosis of a genetic disorder may carry with it a certain social stigma, as well as concerns about discrimination with regards to employment and life insurance. Furthermore, genetic risk cannot be altered nor is it caused by the actions of the patient. A physician’s “duty to warn” individuals who may be put at risk by the actions of a patient has been recognized in the context of both infectious disease and psychiatric illness. However, it is unclear whether these same precedents may apply in the context of genetic information. Nonetheless, a number of lawsuits in the United States have been brought against physicians for their alleged failure to warn relatives of genetic risk. Canadian Policy decisions on this issue must be made based on an understanding the benefits and limitations of genetic medicine, as well as a clear appreciation of the physician’s dual duties to maintain confidentiality and prevent harm. In light of this, this article reviews the available literature to address legal and ethical issues as well as potential policy directions surrounding a physician’s “duty to warn” patients’ relatives about genetic risk.

Introduction

Recently, well-known Harvard professor and bestselling author of *The Stuff of Thought*, Steven Pinker, allowed his entire genome not only to be sequenced, but also to be posted on the Internet for the world to see. In so doing he has given celebrity status to key questions in the era of personalized genomics. These include whether one has a responsibility to disclose the results of genetic tests to family members who might likewise be affected, and who should protect the consumers of the health care system from what has become known as “genetic discrimination.”

To date, instances of so-called genetic discrimination have been scarce in the United States and nearly unheard of in Canada, aside from the odd account of discrimination with respect to life insurance. Furthermore, this topic has been well covered by this publication in a previous article and will not be expanded upon further. However, an issue that does warrant further investigation is whether physicians have an ethical and legal obligation to warn the relatives of patients with a positive genetic test if that information may benefit them. A thorough understanding of the theoretical benefits and limitations of genetic testing, as well as how these translate to our patients in practice, will ultimately have to guide policy in this matter.

Genetic Testing

According to Knoppers et al. (2004), genetic tests can be defined in a legal sense as either those tests that are based on “the presence or absence of specific genetic abnormalities” or more broadly, those that include “tests based on the end products of most genes.” Knoppers et al. argues that in the latter case, genetic risk factors amount to nothing more than probabilities, and thus are difficult to distinguish from other routine predictive tests performed by a family physician. Some diseases can be conferred by a single gene mutation, or the inheritance of two mutated genes at the same genetic locus. In contrast, many
common diseases such as diabetes, cancer, and heart disease have a multifactorial etiology, involving the interaction between genes and the environment. Up to now, most genetic tests have been aimed at detecting single gene disorders, and often provided definitive diagnoses when the clinical picture was suggestive of genetic disease. With the advent of whole genome sequencing, tests that enable clinicians to assess genetic risk in much the same way as a family history are increasingly available.6

Genetic Medicine

Medical uses of genetic information are nearly as diverse as the definition of genetic testing is broad. Physicians can use genetic information to make diagnoses in newborns (e.g. phenylketonuria, PKU), to identify future health risks (e.g. BRCA1 and 2 mutations), and to predict drug responses or even to predict health risks to children not yet born.6 Most pertinent to the current discussion are those genetic diagnoses or predictive genetic tests that may indicate a familial risk of disease. Often some of this risk can be mitigated through early recognition of a genetic risk factor. Conversely however, there are genetic diseases where pre-symptomatic diagnosis confers no prognostic advantage, but which may carry a significant associated burden of disease, such as Huntington disease (HD).6

Central to the ethical dilemma of predictive genetic testing is what the information will actually mean to the patient in terms of health care options. In fact, according to Wylie Burke only about 20% of those at risk for development of HD in the UK had opted for predictive testing as of 2002.6 This reluctance to pursue predictive testing for an incurable disease may lie in the psychosocial consequences of an unfavorable result. One study by Giargiulo et al. found that of 119 patients who underwent predictive testing for HD, a significantly greater number of pre-symptomatic carriers experienced depressive episodes than those found to be non-carriers.7 Those people who do choose to pursue predictive testing often do so in order to make informed decisions about reproduction, rather than for the hope of treatment.7,8 In this sense, predictive genetic testing can be used to make decisions that limit the possibility of birthing a child with significant risk of disease development. On the other hand, many genetic disorders show incomplete penetrance, with some individuals having an altered genotype but showing no evidence of disease.6

Confidentiality and the Duty to Warn

Physicians have a duty to protect the genetic information of a patient from unauthorized disclosure to any third party. However, under certain circumstances confidentiality may be justifiably breached, as is the case when legislation mandates it, or when it is breached with the aim of preventing harm to a third party.9 Numerous legal precedents, including Tarasoff v. Regents of the University of California10 and Pitman Estate vs. Bain11 recognize a physician’s “duty to warn” under circumstances where “disclosure is essential to avert danger to others”.9 Furthermore, the Personal Health Information Protection Act provides that physicians or other health care workers “may disclose personal health information if there are reasonable grounds to believe that the disclosure is necessary to eliminate or reduce a significant risk of serious bodily harm to a person or a group of persons.”12

It is important to understand this distinction when considering whether or not a family member may be entitled to know about a patient’s genetic information. An excusable breach of confidentiality is technically illegal, though the courts may choose not to mandate compensation or punishment based on well-intentioned motives.9 Thus, a patient’s right to confidentiality is legally protected in general, though the courts recognize that under certain circumstances it may be ethically necessary for a physician to breach this confidentiality. By contrast, a physician’s “duty to warn” refers to a legal obligation to inform a third party of imminent risk to their well-being, regardless of a patient’s right to confidentiality.9,13 Whether or not such a breach of confidentiality might be justified in the case where disclosure of genetic information to family members may enable them to decrease their risk of disease development remains to be determined.
by the courts.2,9 On the other hand where genetics is concerned, physicians become privy to information that has implications to both the patient and their relatives. In this sense, the calculation of genetic risk in one person may violate another’s right to personal autonomy where health care decisions are concerned.9 Thus Dickens et al. argue that health care professionals may take on a new and very real set of duties as it pertains to sensitive information that may be very beneficial to individuals who may in fact not be their actual patients.9

While a “duty to warn” has been recognized in the context of both psychiatric illness10 and infectious disease,11 Canadian courts have yet to set similar legal precedents where genetic information is concerned.2 Whether or not the aforementioned precedents might apply to cases where disclosure of genetic information may benefit a third party is questionable. Disease risk conferred by your genotype is not preventable, nor can relatives be viewed as potential victims of the patient (i.e. the patient does not directly cause the risk).2,9 Finally, less than 1% of physicians surveyed in the US believed a “duty to warn” was reasonable in instances where no valid medical therapy exists.13

Notwithstanding, as of 2004 failure to warn a relative of genetic risk had already resulted in 3 lawsuits against physicians in the United States.13 In all three of these cases, State Appellate courts ruled in favor of the plaintiffs, asserting that physicians must ensure that immediate family members are warned of impending genetic risk (though in one case the actual jury eventually decided in favor of the physician).13 Currently in Canada there is no obligation to warn family members of genetic risk, and disclosure could even potentially be punishable by courts or professional regulatory bodies.2 Some strategies aimed at resolving this issue include: strict confidentiality, a universal duty to warn, and informed consent or as Offit et al. would say a “genetic Miranda warning.”2,13 This third strategy involves physicians informing their patients of the circumstances under which they would be obliged to disclose information prior to genetic testing. While the first strategy does not adequately protect the interests of the family members, the latter two strategies are coercive and do not respect the patient’s right to make autonomous decisions about health care. In addition, it is likely that they would adversely affect patient utilization of predictive testing because of perceived genetic discrimination.2,13 Thus, the currently accepted strategy is to adopt an “intermediate position” whereby confidentiality is respected as a rule, but it is recognized that circumstances exist in which disclosure of genetic information may be justifiable.2,9

Conclusion

While no formal legal duty to warn currently exists in Canada, there is a general consensus that physicians should make a valiant effort to inform their patients that genetic information may prove invaluable or even life saving to their relatives. In cases where penetrance is high, risk of imminent and severe harm is great, and viable treatment options do exist, the benefits of disclosure to a third party may so far outweigh the potential harm to the patient as to make it justifiable in a court of law.2,9,13 Thus the physician’s dual duties to maintain confidentiality while simultaneously acting in a spirit of beneficence continue to be at odds in this matter. Unfortunately it is likely that they will remain so until more is known about the circumstances under which knowledge of their relative’s genetic risk might actually prove beneficial to an individual.

References