“Serious” but not “imminent”: genetics and the disclosure of health information to at-risk relatives

Joanne Lee

Abstract

The most recent amendment to the Health Information Privacy Code (HIPC) permits disclosure of health information to third parties where there is “serious” risk to the life and safety of an individual or to others. In this article, I discuss the complexities that arise in the application of the new test for “seriousness” to genetic information and why this standard is subjective and a threat to patient privacy.

Health care practitioners are obliged to maintain the confidentiality of a patient’s health information with very few exceptions. There are strong public health and personal interests in maintaining health privacy and unauthorised disclosure carries serious consequences.

In New Zealand, information privacy principles of the Privacy Act 1993 previously permitted the disclosure of information held by an agency to third parties where there was a “serious and imminent” threat to a person’s life or health or to public health or public safety. This standard was recently amended by the Privacy (Information Sharing) Bill 2011 which has modified the threshold for the disclosure of information to third parties to occasions where there is a “serious” threat to a person’s life or health or to public health or public safety.\(^1\)

This change to the Privacy Act 1993 was similarly recommended by the Privacy Commissioner for the Health Information Privacy Code (HIPC) in which the information privacy principles are mirrored.\(^2\) Health care practitioners now have a wider statutory exception to the common law obligation of medical confidentiality. This amendment has significantly broadened the exceptions where a threat to public safety or an individual justifies disclosure and may override an individual’s privacy interests.

This change was supported by many in New Zealand, particularly those with concerns for the safety of vulnerable children. Previously, information regarding children whose long-term safety was a concern could not be disclosed unless the threat of harm was both “serious” and “imminent”. Removing the need for imminence has broadened the scope of permissible disclosure and grants health practitioners and other agencies the discretion to disclose information where there is serious concern for the safety of children but no urgent threat.

Another argument supporting this new threshold is the disclosure of genetic information to at-risk relatives where there is risk of “serious” harm but where harm may not eventuate for many years. Many feel that the requirement for imminence is too strict, especially since symptoms of genetic disorders sometimes take years to manifest.
Removing imminence from the standard has significantly altered the moment at which health practitioners may be exempted from the obligation of absolute medical confidentiality and may legally disclose sensitive health information. This change to the HIPC now grants health professionals the discretion to make disclosures to third parties, without the consent of a person, where necessary. However, as discussed by Richmond Wee, medical professionals remain bound by professional standards and guidelines that place responsibility on practitioners to seek guidance and exercise due care before making disclosures.3

Although many believe that this amendment will create more benefit than harm, I argue that this new standard is too simplistic and subjective when it comes to complex genetic conditions. This new threshold grants health practitioners too much discretion and too little guidance in their assessment of “seriousness” and increases the risk of unjustified breaches of patient privacy.

The test for “seriousness”

Currently, disclosure to at-risk relatives is permissible if it is one of the purposes for which the information was collected in the first place.4 Thus, provided the practitioner informs the patient prior to testing, disclosure to at-risk relatives may take place if the need ever arises. This is often the case in genetic counselling. However, complications arise when an incidental result is produced and a patient refuses to inform at-risk relatives, despite the potential for serious harm.

Rule 11(2)(d) of the HIPC now permits the disclosure of health information where it is necessary to prevent or lessen a “serious” threat to the life or health of another individual. A “serious” threat is one where an agency—or in this case, a health practitioner—“reasonably believes to be a serious threat having regard to all of the following:

(a) the likelihood of the threat being realised; and
(b) the severity of the consequences if the threat is realised; and
(c) the time at which the threat may be realised.”5

Proponents of the amendment argue that the third limb of this test retains the need to consider the imminence of threat. While this may be true, this change merely requires a practitioner to have regard for the time of the threat (or its imminence).

Provided a practitioner has reasonable belief that the threat is severe and likely enough to occur, disclosure may occur and the time at which the threat may occur is not an absolute requirement. There is no guidance on how to judge “likelihood”, “severity” and the timing of the threat and I discuss (using conditions like familial adenomatous polyposis, Alzheimer’s disease and familial breast/ovarian cancer) how this may become problematic for genetic conditions where there are often multiple variables at play.

A genetic condition is a very broad term. It can refer to an FAP mutation that almost certainly guarantees cancer for carriers of the mutation and it can also refer to a slightly higher apolipoprotein E level that has been linked to an increased risk of late-onset Alzheimer’s disease. It is widely accepted that with the exception of a handful of high penetrance diseases, the likelihood of carriers of genetic mutations developing
a disease is dependent on many other non-genetic factors and that a carrier status carries with it more probable risk than certain risk.

As our understanding of genetics and disease advances, it is likely that more links between genetics, disease and other factors will be uncovered. The development of a genetic disease can rightfully be regarded as a serious threat to one’s overall health and wellbeing. If disease progresses, it may also become a threat to life. However, the multifactorial nature of these diseases, the diversity of harms, its likelihood, as well as the time at which the disease occurs (if at all) all contribute to a very complicated (and uncertain) picture — one that I argue is not easily assessed by the test for “serious” risk.

Likelihood of threat being realised—“imminent” versus “lifetime” risk

Familial Adenomatous Polyposis (FAP) is an inherited disorder caused by a genetic mutation in the APC gene. Without early intervention, FAP eventually causes cancer of the large intestine and rectum and the child of an FAP sufferer caused by mutation of the APC gene has a 1:2 chance of inheriting the disorder. This is similarly the case for Huntington Disease. In these high penetrance diseases with quantifiable risks, at-risk relatives of a sufferer have a very high likelihood of being a carrier and carriers are at imminent risk of disease.

Besides high penetrance diseases, other genetic mutations arguably carry a much more variable risk of developing into a genetic condition. Examples include BRCA mutations that predispose carriers to breast, ovarian and other cancers. BRCA1- and BRCA2-mutation carriers have a 45-80% lifetime risk of breast cancer; and either a 56-60% risk of ovarian cancer for BRCA1-mutation carriers or 11–35% risk of ovarian cancer for BRCA2-mutation carriers.6 7

While BRCA mutation carriers have a significant lifetime risk of cancer, whether or not a relative is at risk of being a carrier and whether genetic screening is a cost-effective means of determining this is dependent on a number of factors.8 This precise assessment of genetic risk in a patient and in his or her relatives is a skill that requires the expertise of a geneticist (or similarly qualified specialist) who is abreast with the latest evidence.

Another example of a risk not fully quantifiable is the apolipoprotein E (APOE) gene that has been linked to an increase in an individual’s risk for developing late-onset Alzheimer disease. Although various hypotheses have been discussed, it is still uncertain how the gene contributes to this increased risk of disease. However, what is known is that the APOE gene is a major genetic risk factor for the disease and sufferers who carry this allele are associated with an increased number of amyloid plaques in their brain tissue.9

Many genetic mutations have been associated with an increased risk of disease. In a small number of cases, risks are nearly half-certain. However, in most of them, the risks are variable with risks ratios that can indicate “lifetime risks” — anything from a strongly confirmed but little understood association to a stronger predisposition towards disease. My examples of genetic mutations that are linked to debilitating
conditions highlight the vast difference in risk and likelihood that different mutations have.

Even though certain genetic mutations may predispose a person to certain conditions, at which point does a predisposition or “lifetime risk” turn into a “likely” threat? Right or not, the removal of the requirement for imminence suddenly permits genetic conditions with very small “lifetime” risk to enter into consideration in the determination of a “serious” threat to the life and/or health of others.

**Severity of the consequences if the threat is realised**

Most genetic conditions carry with it symptoms or a knowledge that goes on to affect the quality of a person’s personal and family life. With knowledge of a person’s status come decisions on lifestyle, available interventions, family planning and the impact of disclosure on family dynamics. In addition, the severity of the consequences depends on the availability and realistic possibility of interventions.

Some genetic conditions do not have a cure yet. Currently, sufferers of Alzheimer disease seek treatment to manage their symptoms although progression of the disease is inevitable. The consequences of AD are severe both on a patient and the family and the disease can sometimes instil a sense of fear and hopelessness of the inevitable. Furthermore, its hereditary pattern dramatically impacts the lives of a patient’s children. For some children of AD sufferers, knowledge of whether one is a carrier can be empowering. However, some children of sufferers, knowing their potential of carrying the mutation and its consequent inevitability, prefer not to undergo testing and would rather live ignorant of their status.

In FAP, people with known mutations for the disease can be closely observed so that immediate action can be taken when polyps begin to develop in the colon. Eventually, removing the colon is necessary and this significantly reduces a sufferer’s overall risk of cancer. The severity of the effects of FAP can be dramatically reduced by early aggressive intervention. However, even intervention involves major surgery that is known to potentially trigger desmoid tumours developing in the surrounding region. The consequences of FAP are severe but interventions are available that reduce its severity.

Similarly, BRCA mutation sufferers are offered both prophylactic intervention options and treatment options where cancer has developed. Interventions range from regular mammography and prophylactic mastectomy to chemotherapy and radiation for cancer patients. The severity of consequences for carriers of BRCA mutations are diverse and the risk of harm to a person depends significantly on how much intervention a person is willing to take on (for example, mammography versus the more invasive preventative mastectomy) as well as how advanced the disease is.

In genetic conditions, assessing the severity of consequences is complicated and dependent on numerous external and personal factors. It is diverse and not fixed and the potential for a threat to severely impact a person depends on a permutation of factors that vary with each disease. In some cases—especially where the outlook is grim—choosing to remain ignorant of one’s own genetic status may even be the less severe option.
The time at which the threat may be realised

This is perhaps the most controversial limb of the test in the assessment of a “serious” threat. In high-penetrance genetic conditions, carriers have a high likelihood of developing a condition at a certain time of their lives (in FAP, the onset of polyps occur in a person’s 30s; in early-onset HD, the disease can begin in childhood or adolescence). However, in the majority of conditions linked to genetic mutations, knowledge of a predisposition to disease generally offers only a vague indication of risk and makes it very hard to assess when a threat may actually be realised. A “lifetime risk” is vastly different from an “imminent risk”, the latter reflecting the near certainty in which something will arise in the future.

However, it is for the conditions in which there may be a “lifetime risk” (such as BRCA mutations) but no “imminent risk” that concern arises. At which point of “lifetime risk” is the legal threshold met thus permitting unauthorised disclosure to take place? Should the existence of a predisposition to a disease like cancer justify informing at-risk relatives even in the absence of consent? Furthermore, does the importance of a person having knowledge about a potential condition justify the stress and anxiety that is attached to that knowledge?

A subjective standard

This change to the HIPC broadens the scope of permissible disclosure of health information to third parties where there is “reasonable belief” of a “serious threat”. With respect to the disclosure of genetic information, it is argued that there are very few occasions where a condition is so certain and severe enough to justify a breach of patient privacy in favour of informing at-risk relatives. Furthermore, such a situation is unlikely as informed consent is normally sought during genetic counselling and before testing begins.

However, as our understanding of genetics and disease advances, it is likely that new situations will arise that the law may be insufficiently prepared for. At present, a health practitioner with knowledge of a person’s genetic risk is left with the responsibility of assessing the seriousness of the condition and whether it justifies a disclosure against the wishes of the patient. The field of genetics is fast-paced and changing and evidence is constantly being updated.

Making an assessment of the seriousness of a genetic condition and whether it justifies unauthorised disclosure to at-risk relatives requires special expertise and should be a role for specialists with full knowledge of genetics. Assessing the genetic risk of a person and his or her extended family requires knowledge of the condition and the latest evidence, its risk factors and the available intervention. Furthermore, an assessment also requires a consideration of the personal factors of each patient and the impact (or how “serious” a threat may be) on an at-risk relative.

When a practitioner considers whether to alert at-risk relatives without a patient’s consent, he faces the dilemma of balancing the privacy interests of a patient, the public interest in maintaining patient confidentiality and the health interest of a third party. This is a heavy burden on the practitioner and an equation I argue only gets more complicated with our increased understanding of genetics.
Conclusion

This change to the HIPC has created a new standard that removes the requirement for imminent danger and instead replaces it with a test that requires an assessment on the basis of severity, likelihood and time of danger. I argue that this test is subjective and too simple for the assessment of genetic conditions and its risk to relatives, especially when this standard marks the threshold in law where a person’s privacy may legally be set aside for the sake of another person’s health.

Besides the guidelines and standards of the Medical Council of New Zealand that apply to good medical practice, there is little else to guide practitioners on this highly consequential decision. In the care of vulnerable children, the amendment to the HIPC is helpful and a reasonable measure. However, in the area of genetics, this blows open the possibilities of practitioners legally revealing genetic information to third parties against the wishes of a patient and the impact this may have on our wider understanding of privacy.

Competing interests: Nil.

Author information: Joanne Lee, LLM graduate, Faculty of Law, University of Otago, Dunedin

Correspondence: Joanne Lee, LLM graduate, Faculty of Law, University of Otago, PO Box 56, Dunedin 9016, New Zealand. Email: joanne.ytlee@gmail.com

References: