CHALLENGES TO GENETIC PRIVACY

The case of disclosure of genetic information to a patient's genetic relatives

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The aim of this paper is to briefly explore Australia's genetic privacy[1] regime as it currently stands within the context of the National Health and Medical Research Council's Public Consultation Draft, *Disclosure of Genetic Information to a Patient's Genetic Relatives Under Section 95AA of the Privacy Act 1988 (Cth): Guidelines to Health Practitioners in the Private Sector*. In doing so, this paper acknowledges the law seeks to balance two competing interests. On the one hand, patients have a right to expect that their medical information will be kept confidential by their health care providers and the law seeks to protect this right by placing strict disclosure requirements on those health care providers in dealing with the patient's health records. On the other hand, it can be argued that health care providers who want to disclose a patient's genetic information to the patient's genetic relatives should be allowed to do so where it would protect those genetic relatives by potentially preventing[2] those genetic relatives from developing the same acute or chronic hereditary illnesses. Proponents of the latter view not only recognise that a patient's right to confidentiality and privacy is not absolute; they seek to expand upon the lawful exceptions that exist so as to override any objections a patient may have in disclosing their health information to third parties. Notwithstanding these conflicting interests, this paper argues that a patient's right to genetic confidentiality and privacy should not be lightly overridden by health practitioners and that there are inherent problems with overruling a patient's requests for privacy by disclosing the patient's genetic status to the patient's at-risk relatives.

The centrality of genetic testing

The mapping of the human genome[3] has ushered in an era in which individuals can screen themselves for an ever-increasing[4] number of genetic or hereditary medical conditions. By choosing to subject themselves to genetic testing, individuals can learn whether they are prone to developing particular heritable medical conditions, long before any onset of those hereditary conditions.

The most important thing to understand with genetic testing is that an individual will not ultimately develop a particular heritable condition if that individual tests positive for a particular genetic fault. However, it does mean that individual is predisposed to developing that particular medical condition to a point where it can almost be said that it will be a certainty[5] that the individual will develop the hereditary condition in their life time. It is therefore important to realise that it is impossible to predict when a particular genetic medical condition will actually materialise in a person carrying a genetic fault, if at all. However, if an individual does have a particular genetic fault, then it should be remembered that those individuals do have, at least, a one-in-two chance of passing on their genetic fault to their future offspring. This process of passing on genetic faults can continue in a never-ending cycle until the faulty genetic link is severed and that only occurs when future offspring do not inherit the genetic problem from the parent or parents possessing the genetic fault.

Genetic privacy regulation in Australia The *Essentially Yours* Report

On 29 May 2003, the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee of the National Health and Medical Research Council (NHMRC) officially launched their monumental 2-year report, *Essentially Yours: The Protection of Human Genetic Information in Australia*. Prior to the launching of that report, Issues and Discussion Papers were released to stimulate debate among experts and interested parties about the protection of human genetic information in Australia. The subsequent inquiry involved 15 open forums taking place around Australia, 200 meetings with interested parties, both in Australia and overseas and ultimately elicited over 300 written submissions.

The inquiry, upon which the final report was ultimately written, directed attention in its Terms of Reference as to whether, and to what extent, a regulatory framework was required:

- to protect the privacy of human genetic samples and information;
- to provide protection from inappropriate discriminatory use of human genetic samples and information; and
- for any related matter.

In its totality, the *Essentially Yours* report is a two-volume, 1200-page text that makes 144 recommendations about the legal, ethical and social implications of the genetics revolution; for example, the use of genetic testing and information in employment and insurance as well as pertinent issues about genetic discrimination.

In her overview of the *Essentially Yours* report, Margaret Otlowski emphasises that a critical feature of the approach of the inquiry that led to the final report was a recognition 'that there are competing interests which

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1. Privacy and confidentiality are conceptually different notions; however, they are inseparable from one another. While the issue of confidentiality is largely concerned with the disclosure and communication of information, privacy 'relates less to interpersonal communications and more to *the right to control information about oneself and the right to exclude others from accessing it*' [Emphasis added]: Danuta Mendelson, 'Travels of a Medical Record and the Myth of Privacy' (2003) 11 *Journal of Law and Medicine* 136 at 140.

2. See Wylie Burke, 'Genetic Testing' (2002) 347 *New England Journal of Medicine* 1867 at 1872, in which it is argued that genetic testing is central to preventive medicine.

3. Further, see ALRC, *Essentially Yours: The Protection of Human Genetic Information in Australia* Report No 96 (2003), Chapter 2.

4. The genetic basis of almost 2000 hereditary conditions has been identified so far; however, screening tests have only been developed for a small number of those conditions: Laura Forrest et al, 'Communicating Genetic Information in Families – A Review of Guidelines and Position Papers' (2007) 15(6) *European Journal of Human Genetics* 612.

5. For example, in the case of Huntington's disease, where the genetic mutation is present, 'the person's risk of Huntington's disease is virtually 100 per cent, given a normal life span': Burke, above n 2 at 1868.

need to be balanced, *with the consequence that reliance on an 'individual rights' model is singularly inappropriate in this area*'[6]. Otlowski further stresses that:

> This is well illustrated by the familial nature of human genetic information; it is intensely personal but at the same time, of relevance to a person's blood relations, and circumstances may arise where the individual's rights and interests (for example, preserving the privacy of genetic information) come into conflict with those of a genetic relative who may need access to that information to advance their own health.

An individual rights approach, however, may sometimes be necessary in particular circumstances and this is recognised, in part, by the proposed draft guidelines to health practitioners in the private sector wishing to disclose a patient's genetic status to at-risk relatives. In meeting the individual patient's needs, a health practitioner is urged to 'understand (that) the family situation involves much more than taking a family (medical) history to identify genetic risks'[7]. Health practitioners are urged to realise there are myriad reasons that patients do not want to disclose their genetic status to at-risk relatives, no matter how serious that risk, and health practitioners are asked to appreciate where the patient is coming from based upon the particular family dynamics of their patient. Breakdown of relationships within the family and long-term familial estrangement, where patients have completely lost touch with genetic relatives either through choice or otherwise can mean patients will not want to re-establish contact with their former family members. Some patients may also fear the stigma of being the carrier of a genetic medical condition and this can have dire consequences on a relationship if a spouse or partner fears that they cannot burden the responsibility of being a carer if their spouse or partner does eventually develop their genetic medical condition. A health practitioner may fail to foresee such consequences, especially if the patient has not communicated such fears to their health care provider.

Dean Bell and Belinda Bennett rightly articulate that 'the interests of family members in not knowing such genetic information must be considered'[8] and this is quite lacking in the discourse in the area of genetic privacy since a large part of the existing academic discourse assumes that people naturally want to know if they carry a genetic problem. On the contrary, it appears that most people *do not* want to know if they are the carriers of a genetic fault because of fears of discrimination in areas such as insurance and employment[9]. There is also the overriding fear that is instilled in people who *know* they may develop the hereditary medical condition they are predisposed to developing because of a faulty genetic condition. It reduces their quality of life by making them nervous about what may happen in the future and thus cannot enjoy their present life. In order to allay their fears, carriers of genetic faults may also nervously resort to constantly monitoring their health and thus add a further strain on Medicare because of greater use of pathology and radiology services, a strain which is further heightened if patients are not privately insured.

Australian federal privacy legislation and genetic information

As reflected in the meaning of 'sensitive information' in section 6 of the *Privacy Act 1988* (Cth), the disclosure of health information about an individual is particularly deserving of legal protection so as to ensure that patients have a high degree of control over their own genetic material. 'Health information' is defined under the same section as including genetic information about an individual in a form that is, or could be, predictive of the health of the individual or a genetic relative of the individual.

The *Essentially Yours* report recommended that privacy legislation be amended to broaden the circumstances in which health care providers could use and disclose genetic information. The effect of this recommendation was the insertion of National Privacy Principle 2.1(ea) in the *Privacy Act*, insofar as it relates to the private sector, to allow health practitioners to disclose patients' genetic information, whether or not the patient gives consent. However, this amendment only catches genetic information collected after 21 December 2001 and prevails over inconsistent state or territory privacy laws.

The *Privacy Legislation Amendment Act 2006* (Cth) also requires the NHMRC, in consultation with the federal Privacy Commissioner, to develop guidelines and address the circumstances in which disclosure to genetic relatives by health care providers is legally justified. Specifically, these guidelines establish when, by whom and in what manner disclosure without the patient's consent may take place, with particular reference to two primary statutory tests: that the threat be a 'serious' one to life, health or safety, and the disclosure be necessary to 'lessen or prevent' that threat *whether or not the threat is imminent*. The fact that the threat does not need to be 'imminent' in NPP 2.1(ea) reflects the scientific fact that it is impossible to determine *when* a patient's genetic relatives will develop their inherited medical condition, if they have inherited the same genetic problem, thereby enabling health practitioners to disclose a patient's genetic information to at-risk relatives without the need to prove an imminent threat since the threat will almost never be an immediate one but may become one at some uncertain point in the future, if at all.

The public consultation draft and guidelines to health practitioners wishing to disclose a patient's genetic information to the patient's genetic relatives under s 95AA of the *Privacy Act*, extrapolates the process that would need to be followed if the guidelines are passed in their current draft format. Even though the genetic threat to at-risk relatives does not need to be imminent for private sector health practitioners to disclose the risk to the at-risk relatives, it cannot be said that the process adopted by the proposed guidelines imposes a low threshold on health practitioners.

It is particularly important to remember that, although the National Privacy Principles do not apply to the public health sector[10], federal privacy laws are currently

6. Margaret Otlowski, 'Essentially Yours: An Overview of the ALRC/AHEC Report on the Protection of Human Genetic Information in Australia' (2003) 12(2) *Australian Health Law Bulletin* 20 at 21. For a critique largely supporting this view, see Loane Skene, 'Genetic Secrets and the Family: A Response to Bell and Bennett' (2001) 9 *Medical Law Review* 162. For a critique asserting greater rights of the individual over the 'communitarian project in familial genetics', see Dean Bell and Belinda Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130.

7. NHMRC, *Disclosure of Genetic Information to a Patient's Genetic Relatives under Section 95AA of the* Privacy Act 1988 *(Cth): Guidelines to Health Practitioners in the Private Sector*, Consultation Draft (2008), 19–20.

8. Bell and Bennett, above n 6 at 156.

9. See, for example, Amy Harmon, 'Fear of Insurance Trouble Leads Many to Shun or Hide DNA Tests', *New York Times*, 24 February 2008, 1.

10. The Information Privacy Principles pursuant to s 14 of the *Privacy Act* do.

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being examined by the ALRC and, in the near future, a similar genetic privacy situation may also prevail in the federal public sector if further legislative privacy amendments are passed, as happened with the insertion of NPP 2.1(ea) with regard to the private sector. It is for this reason that it is particularly pertinent to briefly explore the legal obligations placed on private health practitioners under the draft guidelines.

Challenges to genetic privacy

A working committee convened by the CEO of the NHMRC, in consultation with targeted consultants, developed the consultation draft which was available for public comment from 11 February 2008 to 12 April 2008. The key point to remember is that there is no duty[11] on health care providers (whether in the private or public sector) to disclose a patient's genetic information to at-risk genetic relatives when the patient withholds consent. But if health care providers feel compelled to override the wishes of the patient, the draft guidelines state that *all* of the following legal obligations must be met by the health care professional in order for the disclosure to be lawful under s 95AA of the federal *Privacy Act* and for the professional to have discharged their obligations under NPP 2.1(ea).

Proposed guidelines to private-sector health practitioners

The NHMRC draft guidelines to private-sector health practitioners wishing to disclose a patient's genetic information to at-risk relatives imposes 10 legal obligations on these practitioners and further best practice points under those particular legal obligations. They can be summarised[12] as follows:

Legal Obligation 1: Ethical considerations must be taken into account when making a decision about whether or not to disclose genetic information without consent.[13]

Legal Obligation 2: Reasonable steps should be taken to obtain the patient's consent to disclose genetic information to at-risk genetic relatives.[14]

It is also best practice under this obligation to:

- explain to the patient the implications for genetic relatives when a heritable condition or genetic status as carrier is confirmed and why they should be informed of any risk to them and to advise the patient that, in certain circumstances, disclosure may be made without consent;
- consider referring patients to a health practitioner with expertise in conveying relevant genetic information or consult such an expert;
- consider arranging for genetic counselling for patients or referring them to an organisation that provides genetic counselling;
- take reasonable steps to enable patients who have impaired mental capacity or are children to be involved in decision-making about disclosure;
- seek independent advice, if consent to disclose genetic information concerning an adult with impaired mental capacity or a child is sought, to ensure that the person's best interests are respected.

Legal Obligation 3: The disclosing health practitioner should have a significant role in the care of the patient and the requisite knowledge of the patient's condition or genetics to take responsibility for decision-making and disclosure.[15]

Legal Obligation 4: Decision-making concerning disclosure without consent must involve health practitioners with appropriate expertise to assess the specific situation.

It is also best practice under this obligation to seek advice on the nature of the threat to genetic relatives and on the advisability of disclosure without consent from colleagues and relevant experts and/or committees, and document the outcomes of these discussions.[16]

Legal Obligation 5: Wherever possible, the identity of the patient should not be disclosed in the course of inter-professional discussions.[17]

Legal Obligation 6: Disclosure of genetic information without consent may proceed only if a serious threat to the life, health or safety of genetic relatives exists.[18]

Legal Obligation 7: Disclosure of genetic information without consent may proceed only when this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative.[19]

It is also best practice under this obligation to:

- allow time for review of the decision and access to genetic counselling before further discussion of disclosure when patients or surrogate decision-makers choose to withhold consent, unless the nature of the condition requires an urgent response;
- discuss the basis of the decision and the process of disclosure with the patient or surrogate decision maker if disclosure without consent is deemed necessary;

11. There is no general duty on a health practitioner to warn a person other than their patient of imminent or genetic harm, unless required by the law. Cf the situation in the United States where such a duty to warn exists: *Tarasoff v Regents of the University of California* 551 P 2d 334 (1976). For further discussion on the duty to warn of genetic threats, see Sharon Keeling, 'Duty to Warn of Genetic Harm in Breach of Patient Confidentiality' (2004) 12 *Journal of Law and Medicine* 235.

12. NHMRC, above n 7, 7–8

13. For further, ibid 16–23.

14. For further, ibid 25–29.

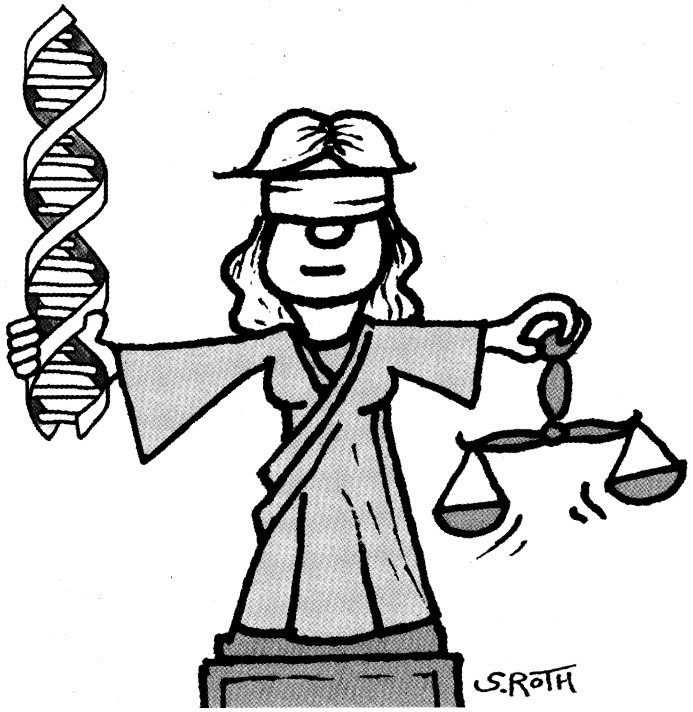
15. For further, ibid 29.

16. For further, ibid 30–31.

17. For further, ibid 30–31.

18. For further, ibid 31–33.

19. For further, ibid 33–38.



- contact genetic relatives of a patient using publicly available documents or information that has previously been provided by the patient;
- communicate in writing with the at-risk relatives, preferably by letter, as this gives the recipient relatives the opportunity to decide whether or not to seek further information.

Legal Obligation 8: Genetic information disclosed to genetic relatives should be limited to that which is necessary for communicating the increased risk and should avoid identifying the patient.[20]

Legal Obligation 9: Disclosure of genetic information without consent should generally be limited to blood relatives no further removed than grandparents or first cousins.[21]

Legal Obligation 10: All stages of the process of disclosure must be fully documented, including how the decision to disclose without consent was made.[22]

Conclusion

So what do we learn from the proposed guidelines for private-sector health practitioners who want to altruistically disclose a patient's genetic status to the patient's genetic relatives? We learn that, if they choose to do so, health practitioners would need to subject themselves to a vast array of legal obligations because the law does not treat the breaching of patient confidence and privacy lightly,[23] especially where a patient has expressly and persistently communicated to their health practitioner that they do not want their genetic information conveyed to their genetic relatives, for whatever reason.

Ultimately, because there is no legal obligation on health practitioners to disclose a patient's genetic information to at-risk relatives when the patient withholds consent, it would undoubtedly be safer and easier (legally speaking, at least) for the health professional to uphold the patient's requests for genetic privacy and for such information not to be conveyed to the patient's genetic family members. Undoubtedly, this can present a very difficult ethical dilemma for health care providers who feel they can avert a patient's at-risk relatives from the same health problems as that of the patient. However, it must also be borne in mind that those at-risk relatives may themselves not want to know about their own genetic status.

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20. For further, ibid 38–40. This is obviously impractical since information spreads quickly among families and it would eventually be discovered through gossip, especially where the family is not large or where suspicions already exists about the health of particular family members.

21. For further, ibid 40–41.

22. For further, ibid 41.

23. It is not difficult to see that a private sector health practitioner can likely be sued for breach of an implied term of contract, tortious breach of confidence and/or professional negligence by a patient if the health practitioner does not lawfully discharge their duties under NPP 2.1(ea) under the draft guidelines and unlawfully discloses their genetic information to at-risk relatives contrary to their wishes and no lawful justification exists warranting such disclosure. There could also be professional misconduct consequences for the health practitioner. For further, see Janine McIlwraith and Bill Madden, *Health Care and the Law* (4th ed, 2006).