Submission to the Public Consultation on the Draft Guidelines for Disclosure of Genetic Information to a Patient's Genetic Relatives under section 95AA of the Privacy Act 1988 (Cth) - Guidelines for health practitioners in the private sector

1. Introduction

1.1 Privacy law needs a major overhaul. The ALRC is recommending significant changes. The proposed guidelines illustrate some of the problems.

1.2 It is worth noting that the genome project has not led to any cures or treatments only diagnostic procedures. The genetic defects in Huntington's disease, cystic fibrosis, muscular dystrophy for example were identified 30 years ago. No treatments or cures resulted from that genetic (and gene product) information. A DNA sequence does not help the emergency doctor faced with a young patient presenting in the final stages of cystic fibrosis.

1.3 It is a moot point but it is at least arguable that the information that a family group harbors a genetic defect, as opposed to the information that a particular individual has a genetic disease, is joint personal information or sensitive information. It should be treated similarly to the information that a joint bank account is overdrawn or a caveat has been registered over jointly owned property. The reason is that it carries consequences for all partners. (I am aware that property law is not a preferred jurisdictional basis for breach of confidential information.)

1.4 It is also arguable that the primary purpose of genetic tests includes, by its nature, sharing that information with relatives likely to be affected. That is to say that disclosure to relatives is a primary or closely related secondary purpose of genetic tests.

1.5 Private sector medical practitioners, particularly those in the large clinics, are running businesses and are interested in short consultations. They may not be prepared to take the time required to comply with aspects of the guidelines but may unnecessarily send the patient to a genetics specialist or, more likely, to a public hospital emergency which is not subject to the guidelines.

1.6 There are significant legal consequences in knowing this type of genetic information. Most notably with insurance. Once advised then health and life insurers think they are entitled to know under the
concept of utmost good faith (and via questions in the proposal). They then may increase premiums or refuse to insure. In my view that includes being advised that a close relative has a genetic disorder.

1.7 My own view is that insurers have already allowed for these conditions in population data used to calculate premiums, and would be receiving an unwarranted bonus, but that is peripheral to the issues here.

2. Draft Guidelines are not Rigorous Enough

2.1 The draft guidelines can be summarized simply as suggesting to private sector health practitioners that they assess the situation for themselves after consultation with other practitioners.

2.2 With respect to the authors, parliament, the Privacy Commissioner and practitioners are entitled to more. Genetic conditions are quantifiable. It is entirely appropriate and in fact expected that the guidelines should include a table of the major, if not all, conditions that can be readily referred to by the practitioner.

2.3 It could detail probabilities of relatives carrying the defective gene, ethnic variations, prognoses, probabilities for present or future children, available treatments or preventative measures. The table should be detailed enough to identify different forms of conditions such as cystic fibrosis or muscular dystrophy.

2.4 Most importantly, the table should guide the practitioner whether or not particular relatives should be advised when consent is withheld. The practitioner can produce the table to the patient.

2.5 Such a table (or tables) could already substantially exist in the medical genetics literature or internet.

2.6 Failing a comprehensive table, a list of genetic diseases indicating which, if any, relatives should normally be informed if consent is withheld. It should be based on a calculated, mathematical risk to particular classes of relatives.

2.7 The claim that there are 2000 identified genetic conditions is misleading. On one view everything has a genetic component. Even if all 2000 were included in the suggested table, with small fonts and shorthand notations it would be shorter than the draft guidelines.
2.8 It is not clear to me why a table has not been included as an appendix. The Committee would be expected to have access to the required expertise but really it is only a compilation from the literature. It is obviously the best way to guide practitioners.

2.9 It is not for me to say but I would presume that the Privacy Commissioner who needs to approve the guidelines, would be more happy with such a table rather than having individual medical practitioners making random decisions on privacy on a case by case basis. That is what the current draft comes down to.

2.10 NHMRC or Committee members might fear litigation if they are too definite but, even failing legislative public servant protection, they are acting under a legislative directive and with ultimate approval of the Privacy Commissioner who administers federal privacy law. There is as yet no breach of privacy cause of action anyway. The Commissioner has the final word, subject to appeals.

3. Practitioners Remain Exposed to Litigation

3.1 The distinction between privacy and confidentiality needs to be addressed. The legislation and the guidelines do not, in my view, protect the medical practitioner against an action for breach of fiduciary duty or breach of confidence. The legislation would need to be very specific if it was intended to do so.

3.2 Where a patient specifically refuses consent to divulge information about their health, undoubtedly confidential information, because of both its nature and the fiduciary relationship, it is unlikely that a privacy exception would protect against a breach of confidence action (assuming loss).

3.3 All such cases will turn on their facts and particularly just what is disclosed. I will be interested to see the proposed form disclosure letters.

3.4 Breach of confidence may occur in discussions with other health professionals, as specifically required under the draft guidelines, where no or limited consent was given.

3.5 Further, the medical practitioner is still at risk from action by relatives who develop a condition when consent was withheld and the medical practitioner chose not to inform genetic relatives. The claim
might be negligence. The leading cases would be where the spouses or partners of an HIV positive individuals were not informed of the risk.

3.6 Those cases might assist the drafters. Even though legislative protection for people with that condition is unusual and specific it is relevant here. Care needs to be taken not to treat serious genetic disease in a substantially different manner than HIV infection. The Committee should have addressed that.

3.7 Scenario 10 is relevant. The medical practitioner attempted to avoid the privacy consequences of conveying the information by doing it anonymously through a third party. That was somewhat futile where the relatives could reasonably ascertain the actual source (see definition of "personal information"). The Privacy Act does address limited disclosure of confidential information to third parties. I, apparently alone, wanted that provision expanded to include private health services, but the ALRC will recommend its abolition.

4. Misuse of the Guidelines for Business Reasons

4.1 A major concern with the guidelines, as drafted, is that they provide a means for private medical practitioners, particularly GP clinics, in the business of providing medical services, to generate work for themselves.

4.2 A single patient can give rise to healthy family members becoming patients for initial screening, consultations, referrals, ongoing life-time monitoring, preventative therapy (if available) and ultimately diagnosis and treatment (if available). Life to death patients who might be entirely healthy for much of their lives. A new and potentially less honorable kind of family doctor.

4.3 Under the guidelines it is entirely within the hands of the medical practitioner. It is lucrative, legal, appears in the interests of the family members. For diseases with no real treatment or preventative procedures, and for diseases of small, ill defined or spurious genetic associations, it is unjustifiable from both public and private health perspectives.

4.4 These are premium patients. The disease is know and they will be healthy for most visits.

4.5 Much better to allow people to live normal lives until they present with early symptoms of the disease.
4.6 If the NHMRC and HREC want to assist private medical practitioners financially they could look at the contracts offered to junior and overseas trained doctors in some clinics, for example. Or grade clinics on percentage of well patients leaving rather than numbers of patients seen or pseudo patients entering.

5. Guidelines should Specifically Exclude Certain Genetic Associations

5.1 Most clearly polygenetic disorders or disorders which have a significant environmental or lifestyle components, such as heart disease or obesity, should be designated not for informing relatives. They are more properly dealt with under public health regimes.

5.2 Scenario 4 is on point. Informing relatives without consent would be wrong. Type I diabetes affects about 1% of population; about 2% first degree relatives; and probably about 2-3% aboriginal population (that is a guess). It is a recognized public health issue more properly handled in a public health manner. The aboriginal population frequency is only peripherally genetic (ethnicity or racial) but primarily the result of environment especially the change from hunter/gatherer diet to our western diet. My point is that the real issue was missed.

5.3 It is another moot point but the information intended to be conveyed here might be classed as very sensitive because it is health information, genetic information, racial information and about children (possibly; juvenile diabetes). Further, disclosure to relatives will have no more benefit than public health "healthy diet" advertising would.

6. Guidelines should Provide Scientific Basis for Arguing that Relatives would Wish to Know

6.1 A more detailed study of people's attitudes towards being informed is warranted. It is clear that the Committee favors disclosure but they should pursue the majority public view. Data on point must be available from numbers of informed relatives who chose to be tested.

6.2 Advising people that they may have a genetic disorder that will likely progress to a serious disease is psychologically debilitating to them. Many will become psychological invalids. I would be certain that studies on HIV positive individuals advised of their status whilst healthy would show that to be the case. I recently heard of a teenage "problem patient" attending frequently at local emergency departments with no real clinical problems but obviously stemming...
from diagnosis some years ago of a genetic (I think degenerative and untreatable) condition. He would have been better off at the footy rather than emergency, in my opinion.

6.3 Anecdotally there is a male female divide. Most young males do not wish to know where it is in the future and there is nothing that can realistically be done. Many women enjoy the short lived notoriety and sympathy value. Compare with suicides: Fewer males attempt but with much higher "success" rates; Females fail but repeat for the recognition and status. If that is so then, without proper guidance, whether or not relatives find out might depend on the medical practitioner's sex.

6.4 Possibly the draft guidelines reflect the number of females involved in their preparation. Presumably the guidelines are for everyone. Again data on male to female respondents to being advised of a genetic disorder would be available and informative.

7. Are Health Practitioners the best placed to judge Who Would Want to Know?

7.1 I found scenario 2 disturbing because it clearly showed an attitude and intention in the guidelines to take the decision making power from a competent parent and place it with the medical practitioner. In my view the mother had every right to withhold consent. She could be deemed to act in the best interests of her children as she saw it, as their mother, as the person most likely to know what the incompetent father would have wanted and as the primary carer of her demented spouse. She knew the terrible outcome and still refused consent. Excepting family conflict she should not be overruled. The children had no doubt wondered if some day they will end up like their father. Everyone knows basic genetics.

7.2 Whilst accepting the difficulties in dealing with the perceived uneducated public, people should be presumed to know what their immediate genetic relatives would prefer. After all they have the same genetic background. It is wrong for medical practitioners to assume that they know more about people they may not even know than a close genetic relative and to assume that they know what is best. Rather too paternalistic, altruistic and questionable where potential financial benefit flows from that position.

7.3 It is generally accepted that medicine is not an exact science but genetics and epidemiology are open to quantification at least in the
statistical sense. It would be helpful for any medical practitioner dealing with a patient refusing consent to disclosure to genetic relatives to have access to comparative statistics such as, for example: The risk of being seriously injured or killed in accidents; The risks of contracting, being invalidated by or killed by non-genetic diseases; The age, sex, lifestyle and ethnic comparisons for these.

8. Increased Risk has a Quantifiable Value

8.1 Most scenarios in the guidelines refer to an "increased risk" to relatives of patients of developing dementia, cancer, heart disease or other serious conditions. These risks have statistical meaning and can be quantified. The probability values are crucial, first for patients in deciding whether or not to consent to disclosure and secondly for the relevant medical practitioner in deciding whether or not to disclose the information without consent. Again I am at a loss to understand why relevant probabilities and statistics are not provided as part of the draft guidelines.

8.2 For example, an increased risk from 0.5% to 2% , although four times is still very low (1 out of 50 family members; but since the patient is 1 then, arguably, 1 out of 99). This has to be ignored by the practitioner where no reliable preventative measures are available. On the other hand, an increased risk from 0.5% to 20% (1 in 5 less the patient giving 1 in 9) is still an acceptable risk to many (particularly if they think a small family appears to reduce that risk). It could support disclosure if a dietary preventative treatment to a serious condition was available.

8.3 The draft guidelines, in my opinion, contain too much retoric and speculation without providing the available scientific information to back medical practitioners in their decisions.

8.4 It is a bit like the weather forcaster saying it will be hot when they could say it will be 26oC. To many people that is only warm.

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