



**Australian Government**  
**Department of Health and Ageing**

Ms Cheryl Cooke  
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Dear Ms Cooke

**Public consultation on the NHMRC draft genetic privacy guidelines**

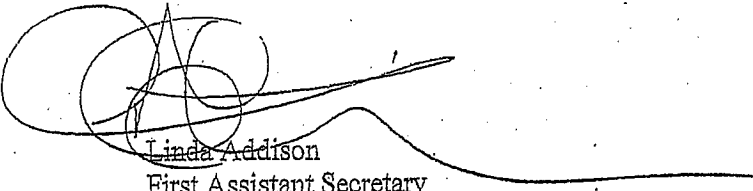
Thank you for the opportunity to comment on the *Disclosure of genetic information to patient's genetic relatives under section 95AA of the Privacy Act 1988 (Cth) - Guidelines for health practitioners in the private sector*, as part of the public consultation. I am responding on behalf of the Department of Health and Ageing.

I understand that the requirement for these guidelines arose as a result of amendments to the *Privacy Act 1988 (Cth)* (amended 2006), which allow for personal genetic information to be disclosed to genetic relatives, without the consent of the patient, under certain circumstances. I note that these guidelines will assist health professionals to decide whether they will disclose personal patient genetic information to the patient's relatives, without the consent of the patient.

The Department previously provided general comments on an earlier draft of these guidelines in October 2007. Enclosed are the Department's comments on the current draft of these guidelines.

If you require any further information, please contact [REDACTED] Assistant Secretary, Research Policy and Biotechnology Branch, Regulatory Policy and Governance Division on [REDACTED]

Yours sincerely



Linda Addison  
First Assistant Secretary  
Regulatory Policy and Governance Division

11 April 2008

enc.



**Australian Government**  
**Department of Health and Ageing**

**Submission to the**  
**National Health and Medical Research Council draft guidelines on**  
***Disclosure of genetic information to a patient's genetic relatives -***  
***Guidelines for health practitioners in the private sector***

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**General Comments**

**1. Inclusion of a Foreword**

A foreword could explain that the guidelines are to assist health professionals disclosing genetic information to family members, both with and without patient consent.

**2. Interim Disclosure of Genetic Information**

The draft text suggests that health practitioners cannot disclose genetic information until the Guidelines are completed. The Department received advice prior to the Privacy Act amendments being introduced into Parliament that health practitioners could disclose the information in the interim. NHMRC should consider taking legal advice on this issue.

**3. Language used in the Guidelines**

While Guidelines must be in plain English, they are intended to provide healthcare providers with legal protection. For example, Obligation 2 (Seeking Consent) says "Every attempt should be made to avoid ...". The first sentence of the explanation then starts "All reasonable steps should be taken ...". There is a difference between the two phrases. The whole text would benefit from editing by an experienced (legal) drafter before further circulation.

**4. Inclusion of a Reference to the NPAAC Guidelines**

The National Pathology Accreditation Advisory Committee (NPAAC) document *Classification of Human Genetic Testing (A supplementary Guide to Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection and Analysis)* provides guidance about the need for professional pre-test genetic counselling for certain genetic tests. The NPAAC Guidelines may further help health professionals consider whether they need to consult with other specialists about their decision to disclose without consent.

**5. Provision for Deceased Persons**

Disclosure without consent could also be applied to deceased persons where a known genetic condition is present, and it may be useful to clarify this in the guidelines. A surrogate decision maker should still be determined in this case - provisions exist to allow for medical records being released and so a similar principle should apply. Cascade contact and consent should also be employed if consent is refused.

## 6. Ethical Considerations

Chapter 2 Ethical Considerations - The Guidelines appear to create the ethical obligation to inform a genetic relative.

Obligation 1 requires "Ethical considerations must be taken into account when making a decision about whether or not to disclose genetic information without consent." Chapter 2 then sets out a range of ethical considerations. It concludes with the boxed text "Health practitioners are ethically obliged to ... encourage [their patients] to inform at-risk relatives or give consent for the health practitioner to do so." (Section 2.3.2)

Health care practitioners are professionally obliged to conduct themselves ethically. Creating an ethical obligation for health practitioners to encourage disclosure is not the intent of the amendments to the Privacy Act. For genetic counsellors, an 'obligation' to encourage disclosure is contrary to the principle of non-directive counselling and would create an ethical dilemma.

## 7. Pharmacists as Disclosing Health Practitioners

The NHMRC draft guidelines on *Disclosure of genetic information to a patient's genetic relatives - Guidelines for health practitioners in the private sector* (the Guidelines) will have little or no immediate effect for community pharmacists or the operation of programs under the Fourth Community Pharmacy Agreement (Fourth Agreement). In the longer term community pharmacists may find themselves involved in multi-disciplinary discussions relating to disclosure of genetic information however, in all but the most rare of circumstances the pharmacist will not be the Disclosing Health Practitioner. Therefore, while community pharmacists should be aware of their obligations under these Guidelines, I foresee very little impact on community pharmacy practice or Fourth Agreement programs and activities.

The introduction, and expected expansion, of genetic testing as a prerequisite for access to identified funded medicines and as a predictor of response to medication therapy or adverse effects will introduce pharmacists to patients' genetic information. Community pharmacists currently practice under the obligations of the *Privacy Act 1988* and the ethical constraints of the *Code of Professional Conduct* published by the Pharmaceutical Society of Australia. It will be necessary for community pharmacists to be made aware of the amendment to the Act allowing disclosure of genetic information to primary relatives under specific circumstances, and the revised obligations to which they, as health practitioners, must adhere.

The Guidelines require that the Disclosing Health Practitioner "should have a significant role in the care of the patient and the requisite knowledge to the patient's condition or genetics to take responsibility for decision-making and disclosure". At this time I cannot foresee a situation where the community pharmacist would fulfil the criteria to take the role of the Disclosing Health Practitioner. In the future, and if pharmacists take on a role of comprehensively managing medication therapy, then it is possible community pharmacists may become more involved in disclosure discussions.

## 8. Inclusion of Case Study on Creutzfeldt-Jakob Disease (CJD)

*The following scenario on CJD has been prepared in consultation with the Australian National CJD Registry and would be suitably placed in Section 3.2.3: Determining whether a serious threat to life, health or safety exists.*

### **Scenario:**

A patient has died from a rapid and progressive dementia illness at the age of 60, his illness is later confirmed as Creutzfeldt-Jakob disease following an autopsy. As the surrogate decision maker, the patient's wife consents to have diagnostic genetic testing on her husband to exclude a genetic basis of the CJD illness. The two adult children are keen to exclude genetic CJD as a basis of their father's illness, as they are planning to start their own families soon. Testing of the father reveals he has a genetic form of CJD. His wife is informed of the final diagnosis by the primary clinician involved in her husband's care. Despite her adult children wanting the information and being a part of the pre-test discussions about the small risk for genetic CJD and the implications of a diagnosis of genetic CJD, the wife is now scared by the reality of the real implications for her adult children. Despite discussions with the clinician about these implications which include public health concerns, she decides to tell her children the results were negative for genetic CJD.

The managing clinician discusses the case with colleagues and a geneticist, and refers the wife to a local genetic counseling service. After counseling, emotional support and discussion of the public health implications, the wife remains unwilling to disclose the risk of genetic CJD being passed through her husband's family. Her aim is to protect her adult children from the emotional burden this diagnosis may bring.

CJD is a fatal and usually rapidly progressive dementia illness. It is the most commonly seen disease from a disease group referred to as prion diseases, or transmissible spongiform encephalopathies. Genetic forms of prion diseases are classed as autosomal dominant. One unique feature of prion diseases is it is transmissible, which is a practical problem in the health care setting when patients with CJD or those at risk of developing a CJD illness are having certain surgical procedures. Patients at risk of inheriting a CJD associated mutation are included in this patient group, requiring extra infection control measures for some surgeries. Infection control measures require non-routine sterilization techniques for some surgeries, consequently health care providers routinely perform pre-surgery patient screening to identify patients who may have a risk of genetic CJD.

### **Points for consideration:**

*What factors support disclosure in these circumstances?*

The adult children were informed and discussed implications for genetic testing, and supported having genetic testing performed. Disclosure of the genetic results to the adult children and siblings is appropriate to inform their own key life-decisions, including their own family planning. They also need to be informed of public health implications when managing their own health care and making blood donations. Infection control precautions are taken to minimize the potential for iatrogenic transmission of CJD between patients. The adult children and siblings of the gentleman may elect to have predictive testing themselves, to determine if they have inherited the disease associated mutation. The unusual public health concerns in genetic CJD also need to form a part of the decision making process for clinicians and the surrogate decision maker, when deciding if the genetic CJD diagnosis needs to be disclosed without the surrogate decision maker's consent.

*What information could be given to the surrogate decision maker?*

In this situation, the surrogate decision maker needs supportive counseling services organized as soon as practicable, after being informed of the genetic results. Discussions need to

address that her adult children have the right to make their own autonomous life decisions, and also to understand public health precautions are taken to minimise the risks of transmission of CJD to others in the health care setting. Information to improve her understanding of CJD may also be needed. Discussion of the public health issues need to address that the clinical team may need to inform the adult children without her consent, accompanied by an explanation why and how this would occur.

*Who might be involved in the decision making?*

The treating doctor, often a neurologist, in consultation with colleagues is best placed to make decisions on whether the adult children need to be informed despite the surrogate decision makers choosing not to disclose the information. A geneticist and a counselor can assist the managing clinician decide on how best to proceed, to assist the surrogate decision maker and the adult children.

*How might the disclosure take place?*

A Genetic counseling service or the family's clinician (if the family is located in a regional setting and no genetics service is readily available) assisted by a local counselor, may need to offer family assistance in a timely manner as there is a public health risk posed to others if the information is not disclosed to family members at risk of inheritance. There could also be a significant burden for individuals after being informed of these issues. Careful planning, consideration of the accuracy of the information being provided and a plan to approach family members at risk is needed. The genetic counseling service is best place to send out letters alerting the adult children to a risk for genetic disease and inviting the adult children to call to organize for a meeting.

**Comments on Definitions**

(Page 3 of NHMRC draft genetic privacy guidelines)

<b>Definition</b>	<b>Comments from the Department of Health and Ageing</b>
<b>'serious' threat</b>	This is still open to personal interpretation by the disclosing health professional. The definition could be improved by noting that professional expertise eg by a genetic counsellor, clinician, neurologist may assist in deciding whether it is a 'serious' threat.
<b>'genetic relative'</b>	It may be more helpful to say 'no further removed than a third degree relative – that is a grandparent, grandchild or cousin of the first individual.'
<b>'genetic information'</b>	This does not provide any additional information / clarity. Presumably this applies to the genetic information itself (eg gene sequence, mutations, chromosome arrangements), as well as the information about what it means (diagnosis, predisposition etc)
<b>'disclosing health practitioner'</b>	The last sentence should come first, as this sentence defines the disclosing health practitioner.

Specific Comments

Reference to the Guidelines	Comments from the Department of Health and Ageing
<p>Section 1.2</p> <p><i>Privacy Legislation Amendment Act 2006</i></p> <p>Page 14-15</p>	<p>The final paragraph on page 14 refers to the NPP 2.1(ea)(i) requirement that disclosure be necessary to lessen or prevent a threat. The actual requirement in the legislation is that 'the organisation reasonably believes' that the use or disclosure is necessary. In other words, the test is not whether it is necessary but whether the organisation has a reasonable belief.</p> <p>Suggest re-wording the second sentence on page 15 so that it reads as follows:</p> <p align="center">'Use or disclosure of genetic information by a health practitioner without the consent of the patient is required by NPP 2.1(ea) to be conducted in accordance with these guidelines.'</p>
<p>Section 2.1.2</p> <p><i>Cultural Competency</i></p> <p>Page 17</p>	<p>Incorporate the following changes highlighted in brackets to last paragraph in Section 2.1.2, concerning Indigenous patients:</p> <p align="center">"As with members of the community, Indigenous patients should be given [information and] advice in a respectful and non-coercive fashion. [The option of i]nvolving an Indigenous Hospital Liaison Officer and/or Indigenous Health Worker[, with the patient's agreement,] will help to ensure that communication takes place in a culturally appropriate way."</p> <p>The discussion of the effects of these Guidelines on Aboriginal and Torres Strait Islander people should be strengthened, as follows:</p> <p>Practitioners should be aware that in some cases Aboriginal and Torres Strait Islander people may consider it appropriate that genetic information should be given by the practitioner to people who are outside the definition of 'genetic relative' used in these Guidelines.</p> <p>Conversely, practitioners should be aware that in other cases, Aboriginal and Torres Strait Islander people concerned with the individual may consider it undesirable that genetic information should be given by the practitioner to people who are within the definition of 'genetic relative' used in these Guidelines.</p> <p>In all such cases advice should be sought from senior community members as to the appropriate course of action.</p>
<p>Section 2.2.2</p> <p><i>Predictive versus Diagnostic Testing</i></p> <p>Page 18</p>	<p>There are 3 types of genetic tests: Diagnostic, presymptomatic and predictive which are defined in <i>Genetics in Family Medicine: The Australian handbook for general practitioners</i>. It is recommended that the two documents use consistent definitions:</p> <ol style="list-style-type: none"> <li>1. <i>Diagnostic testing</i> is done to make or confirm a diagnosis of a specific condition in a person who generally already has signs or symptoms of that condition. This may involve molecular, cytogenetic or biochemical genetic testing.</li> </ol>

Reference to the Guidelines	Comments from the Department of Health and Ageing
<p>Section 2.2.2 <i>[continued]</i></p>	<p>2. <i>Predictive testing</i> is done to determine if a person, who generally has no signs or symptoms of a specific condition at the time of testing, has the specific genetic mutations that increase the likelihood that he or she may, or will, develop the condition in the future. Predictive testing is often performed in relation to genetic conditions that are not evident at birth but have their onset during adulthood, such as some cancers. Predictive genetic testing in conditions such as familial cancer can only be done when the family-specific genetic mutation is known. Hence, genetic testing must first be done on a family member affected with cancer, as they are the most likely to carry the genetic mutation.</p> <p>3. <i>Pre-symptomatic testing</i> is done to determine if a person will develop the condition if they live long enough but symptoms of the condition have not yet manifested, eg Huntington disease.</p>
<p>Section 2.2.5 <i>Special Situations</i> Page 20</p>	<p>Dementia may be present but un-associated with the genetic test being considered. It would be better to say:</p> <p><b>[Dementia may also be the result of a heritable genetic condition involving neurological degeneration]</b></p>
<p>Section 2.3.1 <i>Ethical Issues Raised by Sharing Genetic</i></p>	<p><i>Under the heading "The preference not to know":</i></p> <p>If a patient indicates that they do not want to be informed of their genetic risk, this should be respected – this is the ethical principle of respect for autonomy. It is not necessary for the guidelines to say ‘the standpoint</p>
<p>Page 22</p>	<p>to not be informed, and not try to defend wanting to tell them.</p>
<p>Section 3.1.3 <i>Seeking consent for disclosure</i> Page 25-28</p>	<p>Health practitioners should be encouraged to consider the pros and the cons of disclosure. Scenarios would be improved by also considering what factors support not disclosing the information. For instance, <i>Scenario 2: Seeking consent when the patient has mental incapacity</i> (Page 27), disclosure about risk of Huntington’s disease may not be supported, because the condition is usually late onset, with variable severity, and there is no current effective intervention or treatment. Disclosure may increase anxiety in the relatives about whether they will be affected.</p>
<p>Section 3.2 <i>Following appropriate processes when consent to disclose is withheld</i> Page 29-36</p>	<p>The language used in the text dealing with processes to follow when consent is withheld indicates that the NHMRC considers the NPP 2.1(ea) exception to be an alternative where the consent exception in NPP 2.1(b) has been expressly withheld. As a matter of statutory construction, NPP 2.1(b) and NPP 2.1(ea) are alternatives.</p> <p>There is however, a difference between absence of consent for the reason that it has not been sought and absence of consent for the reason that it has been sought but refused. The practitioner would need to be clear in his or her own mind that there was, in fact, a serious threat to life, health or safety and would be well advised to ensure that there was credible evidence of such a risk.</p>

Reference to the Guidelines	Comments from the Department of Health and Ageing
<p>Section 3.3.1</p> <p><i>How does disclosure take place?</i></p> <p>Page 38</p>	<p><i>Scenario 9: When disclosure may not be possible</i></p> <p>It should be made clear in the example whether the daughter is already pregnant when the mother attended the clinic, or subsequently becomes pregnant. Disclosure of a serious genetic condition to an already pregnant woman may pose additional difficult questions for her about continuing the pregnancy. If she was not already pregnant, but known to be considering becoming pregnant, then the urgency of disclosure may be greater as this knowledge may affect her reproductive decisions.</p>
<p>Section 3.3.2</p> <p><i>What information should be provided?</i></p> <p>Page 39</p>	<p><i>Scenario 10: When it is difficult to protect the patient's identity</i></p> <p>This poses a case where family members may receive a letter advising them that they may be at risk of a heritable condition. It would be useful to include an example of the procedure the genetic relative's GP would follow. For example:</p> <ol style="list-style-type: none"> <li>1. Contact the clinical geneticist for the first doctor's contact details</li> <li>2. Contact the first doctor for information about the genetic condition</li> <li>3. Provide information to the relatives, whilst protecting the identity of the first relative.</li> </ol> <p>Secondary doctors / health care providers who are acting on behalf of extended family members in the disclosure event also need information about how to behave in order to protect the first patient's confidentiality.</p>
<p>Section 3.3.3</p> <p><i>Process of cascade contact</i></p> <p>Page 40</p>	<p><i>Scenario 11: Using cascade contact to disclose to more family members</i></p> <p>Prophylactic surgery is an option for very high risk breast cancer families, but more commonly annual mammograms are advised. Many genetic conditions can be managed by lifestyle modification and regular screening programs. Disclosure may still be warranted in lower risk breast cancer families for whom surgery would not normally be the preventative option.</p>