



Australian Government

National Health and Medical Research Council

N H M R C

# *Use and 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*

2014 (updated 2024)

BUILDING  
A HEALTHY  
AUSTRALIA

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E: [communications@nhmrc.gov.au](mailto:communications@nhmrc.gov.au)

P: (02) 6217 9000

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# INTRODUCTION

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The sequencing of the human genome has led to identification of the genetic basis of an ever-increasing number of conditions, including familial conditions. As a result, health practitioners will increasingly encounter people with genetic risk of disease and inherited disorders. Some of these conditions pose a **serious threat to life, health or safety**, and many are associated with dementia and impaired decision-making ability.

**Genetic information** resulting from assessment of an individual may be relevant not only to that person but also to genetic relatives, due to the shared genetic heritage within families. Depending on the nature and penetrance of the genetic condition, **genetic information** from one person can have consequences for the health of entire extended families. Because information gained through genetic testing can be seen as being relevant to a family rather than an individual alone (Davey et al 2006), people generally either notify family members themselves or give consent for health practitioners to do so. When this consent is not given, health practitioners may recognise the potential benefits of providing information to genetic relatives.

In 2003, a joint inquiry by the Australian Health Ethics Committee (AHEC) and Australian Law Reform Commission (ALRC) recommended that privacy legislation be amended to broaden the circumstances in which health practitioners may **use or disclose genetic information** without consent. The Privacy Legislation Amendment Act 2006 (Cth) (the Amendment Act) made changes to the Privacy Act 1988 (Cth) (the Privacy Act) to allow health practitioners to **use or disclose patients' genetic information, whether or not they give consent, in circumstances where there is reasonable belief that doing so is necessary to lessen or prevent a serious threat to the life, health or safety of their genetic relatives**. This is also reflected in Australian Privacy Principle (APP) 6.2(d) and section 16B(4) of the Privacy Act, which refer to permitted health situations concerning the use and disclosure by an organisation of genetic information.

Section 16A of the Privacy Act also provides an exception to using or disclosing patients' genetic information without consent where an agency or organisation reasonably believes that the use or disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of any individual, or to public health or safety. The inclusion of Section 95AA into the Privacy Act provides for guidelines that clarify circumstances in which **genetic information** may be **used or disclosed** without consent. The amendments **do not oblige disclosure** of information but provide the framework for this to occur under the appropriate circumstances.<sup>1</sup>

The Amendment Act also introduced a requirement in the Privacy Act for the NHMRC to develop and issue these Guidelines, which must be approved by the Australian Information Commissioner (the Commissioner). These Guidelines came into effect on the date nominated in the legislative instrument of the Commissioner in accordance with section 95AA(2) of the Privacy Act.

These 2024 Guidelines replace the 2014 guidelines, which were developed by an NHMRC Working Committee (see Appendix 1). The 2014 Working Committee reviewed the amendments to the legislation, considered the ethical issues involved and developed guidelines and practical guidance. These 2024 Guidelines have been remade in substantially the same terms as the 2014 guidelines; minor revisions were undertaken to maintain the document's currency. This action was taken to ensure the utility of these guidelines while the Australian Government implements its response to a substantial review of the Privacy Act that proposed significant changes to the legislation with implications for these guidelines. Once that work is complete, a substantial review of these guidelines is anticipated.

Breaches of the Guidelines may be pursued under the Complaints Procedures set out in the Privacy Act.

<sup>1</sup> The Privacy Act is available at [www.comlaw.gov.au](http://www.comlaw.gov.au)

## Purpose

The Guidelines specify the requirements that must be met by health practitioners in the private sector if they choose to **use or disclose genetic information without patient consent** under APP 6.2(d) and section 16B(4) of the Privacy Act. Disclosure of genetic information without consent must be in accordance with those provisions and these Guidelines. In contrast to other guidelines for clinical settings developed by the NHMRC, these Guidelines have been issued with the approval of the Commissioner as the means of implementing the amendment to the legislation.

## Application

These Guidelines apply to **organisations**, as defined in section 6C(1) of the Privacy Act that have obtained **genetic information** in the course of providing health services to individuals (including, for example, medical specialists and general practitioners [GPs] in private practice). APP 6.2(d) and section 16B(4) of the Privacy Act do not apply to State or Territory authorities (as defined in section 6C(3)) (or to **genetic information** stored in databases maintained by these authorities). Therefore, generally, these Guidelines **do not** apply to clinical genetics services or other medical practices that are in the State or Territory public health sector.<sup>2</sup>

Section 95AA, APP 6.2(d) and section 16B(4) of the Privacy Act only apply to **genetic information** collected on or after 21 December 2001. They are applicable to **genetic information** about a living person.<sup>3</sup>

## Scope

As well as reflecting the amendments to the Privacy Act, these Guidelines give general guidance that can be adapted to specific situations. These situations will differ depending on a range of factors including the genetic condition involved, relationships within the family, and the health care setting.

The scope of the Guidelines does **not** include:

- the **use or disclosure of genetic information** in the State/Territory public health sector<sup>4</sup> as such authorities are not subject to the Privacy Act<sup>5</sup>;
- the disclosure of **genetic information** to anyone other than the patient and genetic relatives;
- situations in which consent to **use or disclose genetic information** to relatives has been given — in these cases, the provisions under APP 6.2(d) and section 16B(4) of the Privacy Act are not applicable as they deal with disclosure without consent.<sup>6</sup> However, the APPs generally and the duty of confidentiality will still need to be considered before disclosing information even with the consent of the patient;
- situations concerning **genetic information** that present a serious threat to an unborn child, as these fall outside the intended scope of APP 6.2(d) and section 16B(4) of the Privacy Act;
- general information about genetic assessment, clinical information to support diagnosis, use of medical records, stored genetic samples or general consent issues;

2. Note though that some bodies established by or in connection with a State or Territory may still be organisations for the purposes of the Privacy Act (for example, incorporated State government-owned entities may come within the definition of an organisation). Organisations should take their own legal advice if they are in doubt about whether they are an organisation for the purposes of the Privacy Act, and these guidelines.

3. This does not necessarily mean that genetic information about a deceased person may be disclosed to any person without restrictions. The legal duty of confidentiality that exists outside the Privacy Act may also apply to this information.

4. Noting that there may be some exceptions – see footnote 3.

5. State or territory authorities may be subject to equivalent state or territory privacy legislation or guidelines, or Commonwealth legislation or guidelines if adopted by the state or territory.

6. Scenarios 1, 2 and 3 deal with circumstances where consent has been provided or the patient chooses to contact relatives. These are included to demonstrate good practice in these more usual circumstances.

- the health practitioner's professional obligation to seek, record, interpret and act on the patient's family history;
- more general issues relating to the application of the Privacy Act and the **duty of confidentiality** in health;
- genetic screening;
- **genetic information** that is stored in databases or registers maintained by State or Territory agencies, as this is outside the scope of APP 6.2(d) and section 16B(4) of the Privacy Act; or
- the use of genetic information in human research (this is discussed in Chapter 3.3 of the *National Statement on Ethical Conduct in Human Research* (the National Statement), as in force or existing from time to time).

## Structure of the Guidelines

The guidance in this document is intended to satisfy the purpose of Section 95AA of the Privacy Act. The document comprises four parts:

- Part A lists the nine Guidelines that specify the requirements that must be met for disclosure to take place and provides an explanation of the terms used in the Guidelines;
- Part B provides a summary of the Guidelines and key points for good practice;
- Part C includes discussion of:
  - the amendments to the Privacy Act introduced in the Amendment Act and the *Privacy Amendment (Enhancing Privacy Protection) Act 2012* (Chapter 1);
  - ethical considerations, including factors involved in understanding specific situations (Chapter 2);
  - requirements for **use or disclosure** without consent in accordance with APP 6.2(d) and section 16B(4) of the Privacy Act and good practice throughout the process of decision-making and, potentially, disclosure (Chapter 3); and
- Part D includes a number of scenarios, which provide general guidance on how **authorising medical practitioners** and **disclosing health practitioners** may meet the requirements under APP 6.2(d) and section 16B(4) of the Privacy Act and act in accordance with the Guidelines. However, it should be noted that the scenarios are to assist organisations to comply with APP 6.2(d) and section 16B(4) of the Privacy Act and the nine Guidelines. **Acting in accordance with the scenarios does not necessarily protect against a breach of the APPs or a breach of the duty of confidentiality (common law).**

The appendices provide:

- information about the development of the Guidelines (Appendix 1);
- sample materials that can be adapted for local use (Appendix 2); and
- answers to frequently asked questions (Appendix 3)

# A: THE GUIDELINES

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## Guidelines for the use or disclosure of genetic information without consent

The Guidelines are presented here for easy reference. The Guidelines provide a concise outline of the requirements for acting in accordance with APP 6.2(d) and section 16B(4) of the Privacy Act. They should be read in conjunction with the full explanation provided later in this document.

For the purposes of APP 6.2(d) and section 16B(4) of the Privacy Act:

Guideline	Purpose
Guideline 1	<b>Use or disclosure of genetic information</b> without consent may proceed only when the <b>authorising medical practitioner</b> has a reasonable belief that this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative.
Guideline 2	Specific ethical considerations must be taken into account when making a decision about whether or not to use or disclose genetic information without consent.
Guideline 3	Reasonable steps must be taken to obtain the consent of the patient or his or her authorised representative to use or disclose genetic information.
Guideline 4	The authorising medical practitioner should have a significant role in the care of the patient and sufficient knowledge of the patient's condition and its genetic basis to take responsibility for decision-making about use or disclosure.



Guideline	Purpose
Guideline 5	Prior to any decision concerning use or disclosure, the authorising medical practitioner must discuss the case with other health practitioners with appropriate expertise to assess fully the specific situation.
Guideline 6	Where practicable, the identity of the patient should not be apparent or readily ascertainable in the course of inter-professional communication.
Guideline 7	Disclosure to genetic relatives should be limited to genetic information that is necessary for communicating the increased risk and should avoid identifying the patient or conveying that there was no consent for the disclosure.
Guideline 8	Disclosure of genetic information without consent should generally be limited to relatives no further removed than third- degree relatives.
Guideline 9	All stages of the process must be fully documented, including how the decision to use or disclose without consent was made.

## Explanation of terms used in the Guidelines

A number of key terms are used in these Guidelines. Most of these are not defined in the Privacy Act. To aid readers, the way in which certain terms are used in these Guidelines is explained below. These explanations are included to assist clarity and do not constitute an interpretation of the legislation. Where a word or phrase is used in its defined sense, the word or phrase will appear in bold in these Guidelines. Otherwise the word or phrase should be interpreted according to its ordinary meaning.

### *Authorised representative*

In situations where a person is legally or physically incapable of making the relevant decisions, a representative of this person is generally involved (eg parent, guardian, or person who holds an enduring power of attorney). Legislation in each State and Territory authorises certain people to make decisions for those lacking the capacity to do so. The scope of this legislation varies between each State and Territory and there are differences regarding the powers, rights and responsibilities of people who are in a position to make decisions for those lacking the capacity to do so. The authorised representative may be someone other than a genetic relative of the individual (eg his or her spouse). However, being a close relative or spouse does not automatically convey status of authorised representative.

### *Authorising medical practitioner*

While a range of professionals may be involved in the care of a particular patient, final responsibility for decision-making on behalf of an organisation about use or disclosure should be taken by a person in the organisation who is a senior medical practitioner who has a significant role in the care of the patient, has sufficient knowledge of the patient's condition and of its genetic basis and has sought expert advice. This person may be a medical specialist or a general practitioner, as long as these criteria are met.

### *Cascade contact*

A step-by-step process that can provide access to genetic information for a wider cross-section of a family, in which each genetic relative who is notified about their increased risk and makes contact with the disclosing health practitioner, is asked for consent to contact his or her genetic relatives. When additional genetic relatives make contact, the process is repeated.

### *Confidentiality*

The general non-legal principle concerned with the obligation of people not to use private information — whether private because of its content or the context of its communication — for any purpose other than that for which it was given to them.

### *Disclosing health practitioner*

Once a decision has been made that disclosure without consent is necessary, the process of disclosure can be undertaken by the authorising medical practitioner.

In these circumstances, the authorising medical practitioner will be the disclosing health practitioner. Alternatively, the authorising medical practitioner can identify another suitably experienced and qualified professional to make the disclosure without consent (eg a genetic counsellor). In these circumstances, the person identified will be the disclosing health practitioner.

### *Duty of confidentiality*

It is common for a person to disclose information to another person with the intention that the information will only be used for a particular purpose, particularly in a health practitioner/patient relationship. In these circumstances the common law (the law developed through decision of courts rather than through legislation) recognises that an obligation or duty of confidence may arise and that the confidential information can only be used or disclosed with the consent of the party who communicated the information.

Note: The duty of confidentiality is in addition to the obligations set out in the Privacy Act.

### *Genetic information*

The amendments to the Privacy Act alter the definitions of “health information” and “sensitive information” to include “genetic information”, without expressly defining “genetic information”. In its 2003 report, the Australian Law Reform Commission did not apply a precise or exhaustive definition to “genetic information”, preferring to suggest consideration of the context to determine whether the use of genetic-related information requires any special handling or protection. It notes that genetic information is gained from a range of sources (eg clinical examination, DNA testing and chromosome studies, newborn screening, family history) and may confirm a condition that is clinically apparent, or be predictive of the likelihood of an individual developing or carrying a mutated gene causing a condition.

### *Genetic relative*

This term is defined as follows in Section 6 of the Privacy Act: “genetic relative of an individual (the first individual) means another individual who is related to the first individual by blood, including but not limited to a sibling, a parent or a descendant of the first individual”. In the context of these Guidelines, disclosure without consent is generally recommended to relatives no further removed than third-degree relatives,<sup>7</sup> as the process of cascade contact should facilitate access to information for the wider cross-section of a family.

### *Lessen*

The term “lessen”, as used in section 16B(4), requires an authorised person to form a reasonable belief that the contemplated use or disclosure of genetic information would reduce the serious threat that exists to an individual’s life, health or safety. In circumstances where a contemplated use or disclosure would not reduce a serious threat to life, health or safety, or assist in reducing that threat, the exception as described in section 16B(4) will not apply.

### *Life, health or safety*

The phrase “life, health or safety”, including as it is used in sections 16A and 16B(4), ordinarily refers to both physical or psychological/emotional health.

### *Necessary*

“Necessary” is defined by the Macquarie Dictionary to mean “something necessary, indispensable, or requisite”. Applying this ordinary meaning in the context of Section 16B(4) it can be said that use or disclosure of genetic information will be “necessary” when it is requisite to achieving the stated outcome. Deciding whether disclosure is “necessary” should therefore be based on whether it will lead to the intended outcome, that is, whether disclosure will lessen or prevent a serious threat to life, health or safety. See Section 3.3.3.

### *Organisation*

All health service providers in the private sector are “organisations” as defined in the Privacy Act, and are therefore covered by the APPs and the Privacy Act.

Note: Except where a health service provider is a sole trader, the obligations under the APPs are generally not imposed on individual health practitioners directly. Rather, the obligations are imposed on the health service provider for whom the health practitioner works. Nevertheless, the practical effect of the Privacy Act is that health practitioners working for a health service provider are required to act consistently with section 16B(4) when they seek to use or disclose genetic information on behalf of the health service provider. Accordingly, these Guidelines have been drafted in a way to guide health practitioners who work for health service providers.

### *Privacy*

While the Privacy Act regulates the collection, use and disclosure, quality and security of personal information, there is no general legal right to privacy in Australian law. Therefore, when the term “privacy” is bolded in these Guidelines the term is a reference to the general, non-legal, principle used to describe the domain within which individuals and groups are entitled to be free from the scrutiny of others.

7. Third-degree relative has been chosen for practical reasons eg for later onset/potentially fatal disorders like familial cancer it is possible that first and second-degree relatives are deceased and so specifying third-degree relatives gives health professionals the scope needed to reach other relevant family members.

### *Reasonable belief*

“Reasonable belief” is a belief that results from the exercise of sound judgement. If an organisation sought to rely on “reasonable belief” they would need to be able to explain, drawing on their experience, training and expertise, the basis on which they formed that belief.

### *Serious threat*

The Office of the Australian Information Commissioner (OAIC) guidance on health privacy<sup>8</sup> state that a “serious” threat “must reflect significant danger to the individual and could include a potentially life-threatening situation or one that might reasonably result in serious illness or injury”. In the context of these Guidelines, there must be a reasonable belief by experts in the field that the threat reflects a significant danger to the individual, which may or may not be imminent. This could include a potentially life-threatening situation, or one that might result in an illness or injury or the threat of a disease or psychological harm that may result in death or disability without timely decision or action.

### *Use or disclosure*

The “use” of genetic information refers to the sharing of genetic information within an organisation, and “disclosure” refers to the sharing of information outside an organisation (eg with the patient’s genetic relatives).

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<sup>8</sup> Available at: [www.oaic.gov.au](http://www.oaic.gov.au)

# PART B: SUMMARY AND PRACTICAL GUIDE

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## What are these guidelines for?

These Guidelines were developed in response to changes to the Privacy Act and to section 16B(4) that provide for disclosure of genetic information to genetic relatives without the consent of the patient in certain circumstances. Specifically, the exception in section 16B(4):

- allows use or disclosure of a patient's genetic information, without the patient's consent, in circumstances when there is reasonable belief that disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of his or her genetic relatives;
- applies to organisations that have obtained genetic information in the course of providing health services to individuals (these include private medical practices, pathology services, private hospitals) and their employees;
- applies only to genetic information concerning a living person that was collected by an organisation on or after 21 December 2001; and
- does not apply to situations concerning genetic information that presents a serious threat to an unborn child.

Disclosure without consent has the potential to cause distress. Appropriately managing the patient or authorised representative in such situations is considered an integral part of duty of care and good practice.

## When can disclosure without consent take place?

The Guidelines establish when, by whom and in what manner use or disclosure of genetic information may take place without patient consent, with particular reference to the statutory test set out in section 16B(4). That test provides for use or disclosure when:

- the organisation has obtained the information in the course of providing a health service;
- the organisation reasonably believes there is a serious threat to life, health or safety of a genetic relative of the individual to whom the genetic information relates;
- the use or disclosure is necessary to lessen or prevent that threat;
- in the case of disclosure, the recipient of the information is a genetic relative of the individual to whom the genetic information relates; and
- the use or disclosure is conducted in accordance with these Guidelines.

In the event that this statutory test is satisfied and the patient or his or her authorised representative has not given consent for use or disclosure, conveying this information to genetic relatives is permitted only if done in accordance with these Guidelines.

The obligations created by the other APPs and duty of confidentiality are other considerations. The application of the Guidelines is considered in more detail below.

Section 16B(4) does not create a legal obligation to use or disclose a patient's genetic information.

As disclosure without consent represents a significant departure from normal practice and is only permissible in certain circumstances, medical practitioners may wish to consult their medical defence organisation before authorising disclosure.

*To provide general guidance so that authorising medical practitioners may meet the requirements under section 16B(4) and act in accordance with the Guidelines, some scenarios are included in Part D. It should be noted that the scenarios are to assist compliance with section 16B(4) and the nine Guidelines given here. Acting in accordance with the scenarios does not necessarily protect against a breach of the APPs or a breach of the duty of confidentiality.*

## How are the guidelines applied?

Guideline 1	Use or disclosure of genetic information without consent may proceed only when the authorising medical practitioner has a reasonable belief that this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative
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When consent is withheld, the authorising medical practitioner will first need to determine whether there is a serious threat to genetic relatives, taking into consideration:

- the nature of the condition, its associated risks and treatment or care options; and
- the probability that a genetic relative may also have the condition or be a carrier of the relevant mutation.

If a serious threat to the life, health or safety of genetic relatives is identified, it should then be determined whether the potential to lessen or prevent the threat exists. Considerations include:

- whether the condition is preventable or manifestations treatable (eg whether the relatives can benefit from the information); and
- if the disease is incurable, whether knowledge of the condition would allow optimal management.

Before making a non-consensual use or disclosure, the authorising medical practitioner must form a reasonable belief that such an act is necessary to lessen or prevent the identified threat to genetic relatives. It must be determined whether a means other than use or disclosure exists to lessen or prevent the threat. The decision to use or disclose without consent must be made in good faith, with the health practitioners involved in the decision-making drawing on their experience, training and expertise.

Key points for good practice are to:

- hold further discussions with the patient and ask that they reconsider the refusal of consent if there is reasonable belief that there exists a serious threat to the life, health or safety of a genetic relative;
- allow time for review of the decision and consider arranging genetic counselling before further discussion of use or disclosure when patients or their authorised representatives choose to withhold consent – unless the nature of the condition requires an urgent response;
- discuss the basis of this decision and the process of disclosure with the patient or the authorised representative of the person if use or disclosure without consent is considered necessary; and
- be aware of the potential for patient distress and manage this appropriately.

The authorising medical practitioner may decide that disclosure should not proceed. This may be because:

- the requirements for disclosure without consent have not been met;
- all the requirements have been met but there are extenuating circumstances in the family to defer disclosure – in which case it may be appropriate not to proceed with disclosure without consent, or if appropriate in the clinical circumstances, to wait until the family's situation changes;
- all the requirements have been met but the medical practitioner is unwilling to disclose – in which case the practitioner should consider identifying another medical practitioner to review the circumstances.

If disclosure is permissible but the health practitioner is unwilling to disclose, he or she should consult another suitably qualified and experienced health practitioner and consider whether it would be more appropriate for the information to be disclosed by another health practitioner.

Health practitioners have an ethical obligation to advise the patient or the authorised representative to inform relatives of the diagnosis, but are under no legal obligation to disclose the information to genetic relatives themselves, whether consent is given or not. As the law currently stands, there is no valid basis to suggest that a doctor could be liable for non-disclosure.

- Whatever decision is made, the process of decision-making must be documented in writing, including details of the reasons for the decision.

## What ethical points need to be considered?

Guideline 1	Specific ethical considerations must be taken into account when making a decision about whether or not to use or disclose genetic information without consent
Guideline 2	Reasonable steps must be taken to obtain the consent of the patient or his or her authorised representative to use or disclose genetic information

In providing guidance on meeting the requirements of the APPs, the guidelines aim to ensure that ethical considerations are taken into account throughout the process of decision-making concerning the use or disclosure of genetic information without consent.

A health practitioner has an ethical obligation to maintain the confidentiality of information about his or her patient. With genetic conditions, an ethical responsibility can also be seen to extend to the wider family so that every effort is made to encourage sharing of information with relatives at risk. Only if these efforts are unsuccessful and the patient or his or her authorised representative continues to withhold consent should the authorising medical practitioner consider using or disclosing genetic information as outlined in these guidelines.

Whether or not the patient agrees that genetic relatives should be notified, the process of sharing genetic information should aim to maintain respect, as far as is possible, for the autonomy and confidentiality of the patient and the genetic relatives.

Key points for good practice are to:

- explain to the patient the implications for genetic relatives and why they should be informed of any risk to them;
- advise that in certain circumstances, use or 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 timely genetic counselling for patients or referring them to an organisation that provides genetic counselling;
- establish whether the patient is competent to make decisions concerning disclosure of his or her genetic information (an authorised representative can then be identified);
- take reasonable steps to enable patients who have impaired decision-making ability or are children or young people to be involved in decision-making);
- seek independent advice to ensure that the person's best interests are respected if consent to use or disclose genetic information concerning an adult with impaired decision-making ability or a child or young person is sought;
- ensure attempts are made to ascertain what the patient's wishes would likely have been before being affected by the disease if he or she is not competent to make decisions about disclosure of genetic information; and
- follow the principles and guidance given in the NHMRC guidelines on communicating with patients (NHMRC 2004a) and on providing patients with information (NHMRC 2004b) when communicating with patients.

If consent is provided, the provisions under section 16B(4) are not applicable.



## Who is responsible for decision-making and disclosure?

Guideline 4	The authorising medical practitioner should have a significant role in the care of the patient and sufficient knowledge of the patient's condition and its genetic basis to take responsibility for decision- making about use or disclosure
Guideline 5	Prior to any decision concerning use or disclosure, the authorising medical practitioner must discuss the case with other health practitioners with appropriate expertise to assess fully the specific situation
Guideline 6	Where practicable, the identity of the patient should not be apparent or readily ascertainable in the course of inter-professional communication

If a patient withholds consent to use or disclose genetic information, timely review of the situation by a health practitioner with relevant expertise is needed to determine the nature of any threat to relatives and the necessity for use or disclosure to lessen or prevent the threat. It is required that a medical practitioner takes responsibility for the process as authorising medical practitioner, even if another professional (eg a genetic counsellor with requisite knowledge of the particular condition) takes on the role of disclosing health practitioner.

It is important that the decision to proceed with use or disclosure of genetic information is made only after discussion with experienced colleagues, even when the medical practitioner involved is experienced in the field. In such discussions, wherever practicable, the authorising medical practitioner should not reveal the identity of the patient either verbally or in writing.

Key points for good practice are to:

- seek advice on the nature of the threat to genetic relatives and on the necessity for disclosure without consent, from colleagues and relevant experts and/or committees. Document the outcomes of these discussions;
- refer the patient to another medical practitioner with the appropriate expertise or consult colleagues and outside experts if not expert in the field yourself;
- organise discussion of the case so that all involved have time to prepare and document the outcomes of these discussions; and
- identify another medical practitioner who is able to fulfil the role if unwilling to undertake the role of authorising medical practitioner

## How does disclosure take place?

Guideline 7	Disclosure to genetic relatives should be limited to genetic information that is necessary for communicating the increased risk and should avoid identifying the patient or conveying that there was no consent for the disclosure
Guideline 8	Disclosure of genetic information without consent should generally be limited to relatives no further removed than third-degree relatives
Guideline 9	All stages of the process must be fully documented, including how the decision to use or disclose without consent was made

Many ethical concerns associated with disclosure can be mitigated through careful structuring of the way in which genetic relatives are contacted. Disclosure of genetic information needs to be sensitively handled with due consideration to the confidentiality of the patient, the preference of genetic relatives not to receive unsolicited information concerning their health, the autonomous right of genetic relatives to receive information affecting their future health, and the importance of offering genetic counselling.

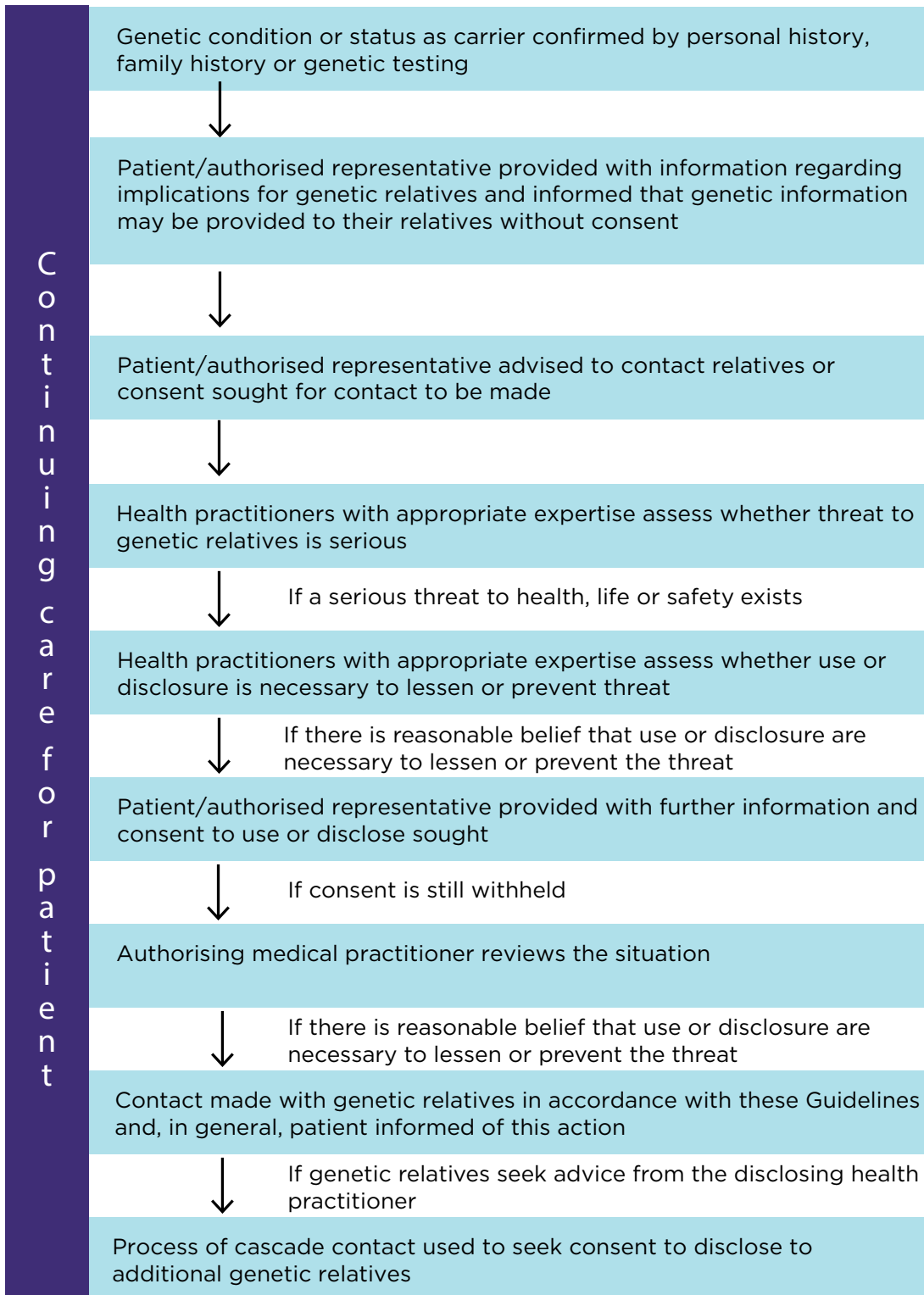
The collection of contact details of genetic relatives must accord with the Privacy Act, particularly APPs 3 and 4 and the related ssection 16A. Section 16A allows an APP entity to collect personal information if it is unreasonable or impracticable to obtain the individual's consent to the collection, use or disclosure and the entity reasonably believes that the collection, use or disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of any individual, or to public health or safety.

Key points for good practice are to:

- continue to exercise a professional duty of care to the patient whether or not consent for use or disclosure is given;
- notify the patient that a decision has been made to disclose without consent and advise them when disclosure has taken place unless there is a contraindication;
- provide written information, which gives the recipient the opportunity to decide whether or not to seek further information (in some circumstances telephone contact may be more appropriate);
- take steps to ensure that any information provided to genetic relatives does not directly identify the patient, the genetic condition or that consent was not given for the disclosure;
- consider using a step-by-step process of cascade contact if contemplating making contact beyond first-degree relatives; and
- fully document all stages of the process, including how decisions were made. It is also important to document situations where a decision is taken not to disclose.

A sample letter that may be used as a template for contact or as the starting point for cascade contact is included in Appendix 2.

# Framework for legal and ethical use or disclosure of genetic information



## Notes:

This framework is provided as a summary only and should be used in conjunction with the Guidelines.

- When a patient is assessed for a genetic condition that has the potential to have serious implications for genetic relatives, these should be discussed and the patient or the authorised representative of the person advised of the potential for genetic information to be used or disclosed without consent in certain circumstances.
- In situations where the patient's decision-making ability is limited (eg due to the impact of the disease process on memory or understanding), reasonable steps
- are required to ensure that the patient's understanding is as thorough as possible. It may be necessary to involve an authorised representative of the person. There are legislative differences between jurisdictions regarding the powers, rights and responsibilities of people in this role. Attempts should be made to ascertain what the patient's wishes would likely have been before he or she became affected by the disease.
- Throughout this process a medical practitioner with appropriate expertise and a significant role in the patient's care will take responsibility for decision-making. In seeking advice from colleagues, this professional should not reveal the identity of the patient.
- If disclosure without consent is to take place, the patient should be notified of this decision unless there is a contrary indication for doing so.
- All stages of the process should be documented, including reasons given if consent is withheld.
- If consent is provided, the provisions under section 16B(4) are not applicable. The APPs and common law apply.

# PART C: CONSIDERATIONS WHEN DECIDING WHETHER TO DISCLOSE WITHOUT CONSENT

Decisions regarding use or disclosure of genetic information without patient consent should be made after careful consideration of:

- Australian privacy legislation (see Chapter 1);
- context for decision making (see Chapter 2); and
- practical considerations (see Chapter 3).

## 1 Australian privacy legislation

The Privacy Act covers the collection, use and disclosure, access to, quality and security of personal information in Australia. As well as providing principles to regulate these areas (see below), the Privacy Act provides a framework for complaints about breaches of the Act, and defines the role of the Commissioner.

The Privacy Act regulates Australian Government and ACT agencies,<sup>9</sup> most private sector businesses with an annual turnover of more than \$3 million and all health service providers in the private sector. Section 6 of the Privacy Act defines “health service” as an activity performed in relation to an individual:

- to assess, record, maintain or improve the individual’s health;<sup>10</sup> or
- to diagnose the individual’s illness or disability; or
- to treat the individual’s illness or disability or suspected illness or disability; or
- the dispensing of a prescription drug or medicinal preparation by a pharmacist.

The Privacy Act applies to all organisations that deliver these types of services, including small health service providers. This includes private hospitals and day surgeries, health practitioners in private practice, private sector pharmacists, and allied health professionals such as counsellors.

### 1.1 Australian Privacy Principles

The Privacy Act contains the Australian Privacy Principles (APPs), which apply to organisations and Australian Government and ACT agencies.<sup>11</sup> The APPs aim to ensure that agencies and organisations that hold information about people handle that information responsibly. Of particular relevance to these Guidelines are APPs 3 and 4, which govern the collection of solicited and unsolicited personal information, and APP 6 (see below) and the Public Interest Determinations 12 and 12A<sup>12</sup> (2011), which establish exceptions from the usual requirements for collecting sensitive information to allow the collection of family history information in certain circumstances.

9. The ACT has its own privacy legislation for health information held by its agencies; therefore health information held by ACT agencies is not covered by the Privacy Act.

10. In this context, this includes matters relating to prevention and identification of illness.

11. These principles also govern most private sector organisations that earn more than \$3 million annually. The handling of personal information by Australian Government and ACT agencies is also governed by the Australian Privacy Principles.

12. See definitions.

The use or disclosure of personal information is governed by APP 6. Under APP 6.2, a health practitioner may only use or disclose health information for the purpose for which it was collected, unless:

- the individual concerned has consented to the particular use or disclosure for a secondary purpose, or
- prescribed exception applies, such as:
  - the individual would reasonably expect the organisation to use or disclose the information for a directly related secondary purpose;
  - where required or authorised by or under an Australian law;
  - a 'permitted general situation' under section 16A exists in relation to the use or disclosure;
  - a 'permitted health situation' under section 16B exists in relation to the use or disclosure.

APPs 3.3 and 3.4 are concerned with the collection of sensitive information, which is defined in section 6 of the Privacy Act and includes all health information. APP 3.3 prohibits the collection of sensitive information about individuals, unless an individual consents or an exception applies.

## 1.2 Privacy Legislation Amendment Act 2006

In 2006 the Privacy Act was amended to give effect to the Australian Government's decision to implement some of the recommendations of the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee in their report *Essentially Yours: the Protection of Human Genetic Information in Australia*. The *Privacy Legislation Amendment Act 2006* (the Amendment Act) amended the law relating to the protection of genetic information so that genetic information can be disclosed to genetic relatives in certain circumstances. The amendments apply to genetic information collected after 21 December 2001 and prevail over State or Territory privacy legislation, to the extent that these laws are inconsistent.

### *Health information and sensitive information*

The 2006 amendments to the Privacy Act aimed to safeguard the handling of genetic information by amending the definitions of "health information" and "sensitive information" to expressly include genetic information. Genetic information that is (or could be) predictive of the health of an individual is now treated as health information for the purposes of the Privacy Act. Genetic information that is not otherwise health information, such as the result of a parentage test, is treated as sensitive information for the purposes of the Privacy Act.

The Amendment Act also included a definition of a "genetic relative" of an individual (the first individual) as "another individual who is related to the first individual by blood, including but not limited to a sibling, a parent or a descendant of the first individual."

### *Changes to the Privacy Principles*

The Amendment Act amended the Privacy Act to include an additional exception to the general requirement that personal information must not be used or disclosed for any other purpose other than that for which it was collected. The inclusion of an additional exception allowed for the use or disclosure of genetic information to a patient's genetic relatives if the organisation reasonably believes that this is necessary to lessen or prevent a serious threat to life, health or safety of the relative, even if the threat is not imminent and consent has not been given.

### *Inclusion of Section 95AA*

Section 95AA was also added to the Privacy Act through the Amendment Act. It allows the Commissioner to approve guidelines developed by the NHMRC to clarify circumstances in which genetic information may be used or disclosed without consent.

Under section 16B(4), any use or disclosure of genetic information by a health practitioner without the consent of the patient must be conducted in accordance with these Guidelines (unless the use or disclosure may be permitted under section 16A (permitted general situations) – particularly item 1 – that is, where it is unreasonable or impracticable to obtain the individual's consent and the entity reasonably believes that the use or disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of any individual, or to public health or safety).

## 1.3 *Privacy Amendment (Enhancing privacy protection) Act 2012 (Cth)*

The *Privacy Amendment (Enhancing Privacy Protection) Act 2012 (Cth)* passed through Parliament on 29 November 2012 and received royal assent on 12 December 2012.

It also amended the Privacy Act, to implement the Government's first stage response to the Australian Law Reform Commission's 2008 report *For Your Information: Australian Privacy Law and Practice* (ALRC report). The most significant amendment is the implementation of a unified set of APPs that apply to both the public and private sector and replace the Information Privacy Principles and National Privacy Principles.

The amended Privacy Act commenced on 12 March 2014.

## 2 Context for decision-making

### 2.1 Meeting individual needs

#### 2.1.1 Communication

Effective communication from the first consultation may help the patient to fully understand the implications of the genetic information being discussed and avoid a situation where he or she refuses consent to disclose genetic information to genetic relatives. Good communication can also help the health practitioner to understand and respect the patient's decisions about disclosure.

NHMRC guidelines on communicating with and providing information to patients (NHMRC 2004a; 2004b) identify obstacles (eg anxiety about the condition, family discord) that may make the patient less able to take in or provide information and make decisions. They also identify obstacles that may prevent the health practitioner from fully appreciating the views of the patient.

Even where there are obstacles, better communication can be fostered through:

- establishing rapport and using active listening techniques;
- helping patients to express themselves and to understand and retain the information given;
- using plain language that is free of clinical terms and reinforcing discussions with written and other relevant materials and services (eg video, websites, advice on relevant support groups, interpreters); and

- considering the environment and length of consultation required before communicating potentially distressing news.

Patients will differ in the amount of information and support they require and there may be particular difficulties in communicating with patients with dementia or cognitive difficulties. The pace of information provision should be determined by each patient's needs and the particular situation.

## 2.1.2 Cultural and lifestyle factors

Health practitioners see patients from a range of ethnic, cultural and socio-economic backgrounds, and should aim to ensure good communication regardless of the social or cultural background of patients. As well as following general principles of good communication, additional strategies that may be helpful include (NHMRC 2004a):

- asking questions to appreciate the patient's understanding of health and disease;
- establishing an environment that welcomes and affirms each patient regardless of background;
- negotiating with the patient about using the assistance of agents such as patient advocates, family members, pastoral care workers or spiritual leaders; and
- seeking advice from community agencies that understand and advocate for patients.

Qualified interpreters and culturally appropriate materials should be available for people from culturally and linguistically diverse backgrounds. Where this is not possible, telephone interpreter agencies can provide relevant services. However, not all cultural groups welcome the involvement of non-family members in such circumstances and health practitioners need to be aware of and sensitive to this possibility (NHMRC 2004a).

Effective communication with Aboriginal and Torres Strait Islander patients requires consideration of cultural factors such as (NHMRC 2005):

- beliefs that the concept of a family differs from that of genetic connections;
- the recognition of both "blood" and "skin" relationships;
- the complexity of Aboriginal and Torres Strait Islander lore, which affects communication within families and communities;
- the importance of family and community involvement in decisions about health care;
- a holistic view of health that includes cause and effect arising from the body, the spiritual, the land and dreaming;
- the high degree of mobility among Aboriginal and Torres Strait Islander peoples; and
- the unique issues relevant to people that live in remote areas of Australia.

Involving an Aboriginal and Torres Strait Islander Hospital Liaison Officer and/or Health Worker, with the patient's agreement, can help to ensure that communication takes place in a culturally appropriate way and that the patient's understanding of the term genetic relative is clear. In cases where disclosure without consent is a possibility, advice from senior community members or Elders may assist in decision-making about the appropriate course of action after careful consideration. Initial contact with Elders should be made with discretion ensuring protection of the individual's privacy.



## 2.2 Understanding the situation

### 2.2.1 Settings

With the increasing use and utility of genetic testing, there is a widening range of settings in which genetic information is discussed. Settings relevant to these Guidelines include (but are not limited to) private hospitals, specialists' private rooms and general practice. Genetics services and familial cancer units are ideally positioned to deliver pre- and post-test counselling and involve at-risk family members as necessary. Outside these settings, the advice of other health practitioners may need to be sought.

### 2.2.2 Diagnostic and predictive testing

The results of genetic tests are not always straightforward, which can make them difficult to interpret and explain. The degree of uncertainty will affect discussion of the implications of the results for patients and their genetic relatives. Family history and experience are also important in determining how an individual will react to the results of genetic testing (Evans et al 2001).

- *Diagnostic testing* — This is done for patients who have clinical signs of disease to confirm or rule out a suspected genetic condition. While a positive result confirms a clinical diagnosis, it still cannot accurately predict the exact course of the disease, and in some situations, the exact phenotype.
- *Predictive testing* — While some heritable diseases are caused by changes in specific genes, most are caused by the interaction of multiple genes with each other and with environmental factors (Petrila 2001). Predictive or presymptomatic genetic testing is done in well individuals to predict future risk of disease.

In some cases, a single test can reveal both predictive and diagnostic information. For example, a diagnostic test for fragile X syndrome in a boy with intellectual disability may reveal a full mutation in the fragile X gene that explains his disability. This result would also have implications for genetic relatives who may carry a pre-mutation that puts them at increased risk of developing a neurodegenerative disorder in later life.

### 2.2.3 Nature of the genetic condition

The type of genetic information being discussed will vary widely, depending on the probability that someone with the mutation will develop the condition, whether the condition is serious or life threatening if it does develop, and whether it is preventable or treatable. The risk of a person developing the familial disease may vary for many reasons, including age, gender, and degree of relationship with an affected person. For example, genetic information may:

- be considered by the health practitioner or the patient as being straightforward;
- imply an increased risk but no certainty of developing a disease;
- have serious implications for present and future generations;
- concern a condition that is presently incurable but has serious manifestations of which the patient is unaware that can be ameliorated; or
- have the potential to cause significant psychological harm.

The nature of the genetic condition will influence decision-making on the benefits of informing the patient's genetic relatives because:

- the more serious the condition, the more important it is to consider the implications for genetic relatives and how they should be alerted to the option of genetic assessment; and
- the degree of risk for different genetic relatives will vary depending on the underlying

condition and its penetrance as well as on the closeness of the relationship with the patient.

#### 2.2.4 Family situation

Being aware of dynamics and pressures within a family can help health practitioners understand patients' reactions when they find out that they have a genetic condition. While many patients wish their information to be available to help their genetic relatives, there are a number of reasons why patients or their authorised representatives may choose not to provide this information to relatives (Clark et al 2005), including:

- cognitive change preventing the person from organising contact;
- shielding others from distress, particularly in the absence of effective therapy;
- breakdown of relationships within the family;
- denial about the condition leading to unwillingness to admit the situation to others;
- uncertainty about how or when they should share information with their genetic relatives;
- thinking that a genetic relative is too unwell or busy to hear the news;
- cultural, religious and spiritual factors;
- fear of the potential for discrimination or stigmatisation if anyone else is told;
- not understanding or acknowledging that others in the family may be at risk;
- having the perception that genetic relatives would prefer not to know;
- financial implications (eg information compromising subsequent applications for life or disability insurance, potential impact on superannuation); or
- fear of establishing or revealing non-paternity, or non-maternity.

If the patient's motivation is based on a lack of understanding or denial, exploring feelings and reactions may help him or her to reverse the decision not to share the information. However, when there is a long-term estrangement, patients may have completely lost touch with genetic relatives and they may be unable as well as unwilling to make contact.

#### 2.2.5 Special situations

For some patients there may be an additional level of complexity in the decision-making process. This may be due to their limited understanding and consequent inability to give informed consent (for example, due to the impact of the disease process on memory or understanding, or maturity levels). Decision-making in situations involving adults who have impaired decision-making or children and young people is discussed more fully in Section 3.1.3.

## 2.3 Ethics in decision-making

### 2.3.1 Ethical issues raised by sharing genetic information

In the context of these Guidelines, there are a number of ethical principles underpinning the practice of sharing genetic information, which are discussed briefly here and are the basis of the guidance given in Chapter 3. If a patient does not give consent for use or disclosure, there is likely to be conflict between the practitioner's ethical obligations to the patient and to his or her genetic relatives, which needs to be considered as part of the decision-making process.

#### *Justice*

Health practitioners may feel a responsibility not only towards their patients but also to the relatives that share their genetic heritage, as genetic information can be seen to be relevant to a family rather than to an individual alone (Davey et al 2006). Clinical genetics practice aims to make the family rather than an individual the unit of care and offers access to the benefits of genetic assessment to family members when the patient gives consent for them to be contacted. The ethical principle underpinning this sharing of information is justice, which may be breached if one member of a family benefits from genetic assessment and at the same time is allowed to exclude others in the family from access to such benefits (Parker & Lucassen 2004).

#### *Beneficence*

The likely benefits to genetic relatives must justify any risks of harm or discomfort to patients if information is used or disclosed without their consent. Most guidelines in this area agree that a health practitioner's minimum ethical responsibility is to tell patients about the implications of their genetic information for their family members, and to actively encourage patients to share this information with genetic relatives (Forrest et al 2007). Discussion about the condition may help patients to understand their genetic risks and those of their genetic relatives (Forrest et al 2007), and assist in avoiding a situation where consent to use or disclose is withheld.

#### *Respect*

Respect for human beings is a recognition of their intrinsic value. Respect also requires having due regard for the welfare, beliefs, perceptions, customs and cultural heritage of individuals (NHMRC, ARC, UA 2023). In this context, confidentiality, which has a long-established tradition in medicine, is relevant to respecting the patient's welfare. Ethically, this is based on the widely accepted view that competent patients should have control over decisions concerning their medical care, including the right to decide what happens to information about them. A further justification for respecting patient confidentiality arises out of concern that breaching confidentiality can undermine trust in the relationship between health practitioners and patients (Parker & Lucassen 2004). The potential for harm to individuals, and ultimately society, from breaching patient confidentiality contributes significantly to the ethical dilemma faced by health practitioners when patients withhold consent to disclose information to their genetic relatives.

### 2.3.2 Benefits and risks of sharing genetic information with genetic relatives

The process of informing genetic relatives about possible risk must be managed carefully. There are ethical issues to consider even when the patient gives consent.

The possibility that genetic relatives may not want to be informed about their risk is also a consideration. However, in most cases the health practitioner is unlikely to know the preferences of the genetic relatives.

However, once relatives have been informed of a genetic risk, they may prefer not to undergo further assessment to learn their own genetic status. There is a range of reasons for not wanting

to undertake such assessment, including fear of discrimination, anxiety, denial, lack of knowledge, perceptions of pressure (Swartling et al 2007) and autonomy (Malpas 2005; Wilson 2005).

The box below indicates some benefits and risks of informing genetic relatives about possible risk.

#### *Potential benefits of disclosing*

- Clarification of the risk status of clinically unaffected relatives so that they can consider predictive genetic testing (if available) and plan future medical and other life decisions.
- The possibility for steps to be taken to reduce the risk of disease or allow early diagnosis and management, including for the manifestations in as yet incurable conditions.
- The avoidance of the need for other investigative interventions if genetic testing identifies the relative as a non-carrier.
- The avoidance of mistaken diagnosis (of another condition) and inaccurate treatment.
- In some cases, shared knowledge of the genetic condition within the family may help to avoid family breakdown and anger.

#### *Potential risks of disclosing without consent*

- Possibility of the privacy of the patient being affected.
- Possibility of losing the patient's trust and confidence.
- Difficulties in the process of advising genetic relatives even if patient is willing to share genetic information.
- Potential for patient uncertainties about the practicalities of disclosure.
- Genetic relatives feeling that receiving unsolicited information about possible genetic risk is an invasion of privacy.
- Perceived pressure on genetic relatives to undertake genetic assessment.

Source: Adapted from Suthers et al 2006.

### 2.3.3 The ethics of disclosing without consent

Any departures from maintaining a patient's confidentiality must be taken very seriously (AMA 2006), and should be the exception rather than the rule. Accordingly, any decision to disclose genetic information to a patient's genetic relatives without the patient's consent must be made extremely carefully, weighing the patient's privacy and autonomy against the potential to lessen or prevent serious harm for genetic relatives (Falk et al 2003).

In addition to ensuring that use or disclosure meets the requirements of section 16B(4) and these Guidelines, the decision involves consideration of:

- the likely effect on the patient of breaching confidentiality; and
- the possible ambivalence of genetic relatives to receiving genetic information. The scenarios in Part D illustrate these considerations.

### 2.3.4 The ethics of non-disclosure

Even in circumstances where disclosure without consent would otherwise be permissible on the basis of the decision-making process outlined in these Guidelines, the treating health practitioner may be unwilling to disclose. This may be because of a belief that it is never acceptable for a clinician to breach a patient's confidentiality in the interests of others, or for other reasons.

The box below indicates some benefits and risks of not disclosing.

*Potential benefits of not disclosing*

- Avoids breaching confidentiality.
- Avoids potential for disruption to patient/doctor relationship.
- Avoids causing anxiety to relatives.
- Potential to reduce harm to family relationships.

*Potential risks of not disclosing*

- Relatives are left unaware of potential risk.
- Potentially preventable harm cannot be averted.
- Relationships within family may be damaged when relatives discover that information was not passed on to them.
- Relatives who were not informed due to a deliberate decision by a health practitioner not to disclose may be distressed and experience harm as a result.

Guidance on the practical and legal aspects of non-disclosure is given in Section 3.3.5

## 3 Practical considerations

### 3.1 Application of the guidelines

Guideline 1	Use or disclosure of genetic information without consent may proceed only when the authorising medical practitioner has a reasonable belief that this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative.
Guideline 2	Specific ethical considerations must be taken into account when making a decision about whether or not to use or disclose genetic information without consent.

These Guidelines establish when, by whom and in what manner use or disclosure of genetic information may take place without patient consent. They are underpinned by:

- Guideline 1, which outlines the requirements of the statutory test set out in section 16B(4); and
- Guideline 2, which concerns the ethical considerations outlined in Chapter 2.

In the event that the statutory test is satisfied and the patient or his or her authorised representative has not given consent for use or disclosure, conveying this information to genetic relatives is permitted only if done in accordance with all of the Guidelines. Other considerations are the obligations created by the other APPs and duty of confidentiality.

This chapter outlines the process of applying the Guidelines and good practice, including:

- providing relevant information to patients or authorised representatives, including referral for genetic counselling as appropriate (see Sections 3.2.1 and 3.2.2) and provision of

further information if consent is withheld (see Section 3.3.4);

- taking reasonable steps to obtain consent (see Sections 3.2.3 and 3.2.4);
- establishing who will take responsibility for the process if consent is withheld (see Section 3.3.1);
- involving other health practitioners with relevant experience in the decision-making process while maintaining patient confidentiality (see Section 3.3.2);
- determining whether the statutory test set out in section 16B(4) can be met and whether it is appropriate for disclosure to proceed (see Sections 3.3.3 and 3.3.5);
- providing only the necessary information and doing so in an appropriate manner, if disclosure is to proceed (see Sections 3.4.1 and 3.4.2);
- limiting disclosure to relatives generally no further removed than third-degree relatives and using a process of cascade contact to provide access to genetic information for a wider cross-section of the family (see Section 3.4.3); and
- accurately documenting the process (see Section 3.4.4).

## 3.2 Discussing use or disclosure of genetic information

### 3.2.1 Providing information about implications for genetic relatives

In any situation when confirmation of a genetic condition or predictive genetic information is likely, there should be discussion with the patient about:

- the implications for genetic relatives;
- the potential benefits of notifying genetic relatives or allowing the release of information; and
- the fact that there is legal provision for use or disclosure without consent in certain circumstances.

The fact that this advice has been given should be documented in the patient's record. Consent is a continuing process. These early discussions support patients in exercising their choice and form the basis for later discussions about consent to disclose the patient's genetic information to genetic relatives.

The Privacy Act requires that providers give notice to their patients about certain matters when they first collect health information. These matters include why the information is being collected, how it may be used and to whom it may be disclosed. The full notice requirements for the collection of personal information are set out in APPs 1 and 5. It is therefore important for patient information leaflets relating to the application of the Privacy Act to include possible use or disclosure of genetic information without consent. A sample patient privacy information leaflet is given in Appendix 2.

### 3.2.2 Genetic Counselling

In situations where genetic information has implications for individuals and their families, patients may be referred to a genetics service. If this is not possible within a reasonable timeframe, for example because of distance or waiting lists, the treating medical practitioner can seek advice from the genetics service about an appropriate course of action. If the patient

is distressed or the situation calls for immediate action, an urgent appointment or telephone counselling may be arranged.

### 3.2.3 Seeking consent for use or disclosure

Guideline 3	Reasonable steps must be taken to obtain the consent of the patient or his or her authorised representative to use or disclose genetic information.
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It is important that health practitioners seeking consent in these situations have the appropriate expertise to do so. Those who do not may elect to refer the patient to a colleague with such expertise. Where timely referral is not possible (eg in rural or remote areas), the treating medical practitioner should seek advice from suitably qualified professionals about the condition and its implications for genetic relatives, without revealing the identity of the patient.

Patients should be given the necessary information and assistance regarding use or disclosure that complements information they have already been given about their own condition and/or treatment, and allows them to make an informed decision. Such information will include:

- which genetic relatives are likely to be at risk;
- the likelihood of each relative developing the familial disease (relevant factors may include age, gender, and degree of relationship with the patient);
- the likely threat to those relatives if they are not advised of their risk and therefore do not seek health advice;
- potential preventive and early intervention measures and possible benefits of these to genetic relatives;
- the availability of genetic counselling for the patient and family members;
- the patient's involvement in the process; and
- the potential for information to be used or disclosed to genetic relatives without identifying the patient or condition.

In some circumstances, it may not be appropriate to seek consent from the patient, such as when:

- an individual has impaired decision-making ability (see below);
- seeking consent may itself cause a serious risk to the life, health or safety of the patient; or
- it is not possible to contact the patient.

Scenario 1 illustrates good practice in discussing use or disclosure in the more usual situation where consent is given.

#### *Seeking consent from adults with impaired decision-making ability*

Patients may have impaired decision-making ability due to a psychiatric illness or disability, intellectual disability, acquired brain injury, or some form of dementia. Impaired decision-making ability may be the result of the genetic condition of interest (eg dementia associated with the neurological degeneration of Huntington disease, Wilson disease, myotonic dystrophy).

Impairment to an individual's capacity to consent may be a permanent or temporary condition. In some cases, it may only affect decision-making ability some of the time, for example where a person has a psychological illness that is episodic in nature. In other cases, the impact on the

person's decision-making ability may be incremental, such as with dementias. However, it may be that the patient can make decisions about the handling of his or her genetic information, if they are provided with the necessary support.

#### *Establishing competency*

A first step would be to assess the patient's ability to give informed consent. Questions to consider include the following.

- Is the patient aware that he or she has the condition?
- Is the patient aware that his or her decision-making ability is impaired?
- Is the patient able to consent to inform genetic relatives?
- If not, would the patient have the capacity if enough time were spent explaining the issues in simple language?

It may also be useful to seek independent advice from a colleague or other relevant expert (eg psycho-geriatrician).

#### *Involving the patient in decision-making*

There may be difficulties in conveying the necessary information to patients with impaired decision-making ability, particularly if they attend consultations alone. The treating practitioner may encourage the patient to bring a spouse, relative, friend or advocate (such as a social worker or health worker with whom the patient is familiar) as a support person during consultations.

The Privacy Act adopts a common law approach to consent, whereby any individual with capacity may exercise choices over the handling of his or her personal information. Efforts are therefore required to ensure that every patient's understanding is as thorough as possible. For patients who are not competent to make decisions concerning disclosure of their genetic information, attempts should be made to ascertain what their wishes were before they became affected by the disease (Bernat 2008). Even in situations where individuals lack legal capacity, they should be involved as far as practicable in the decision-making process.

#### *Involving the authorised representative in decision-making*

If it is determined that a patient is not capable of understanding relevant matters, an authorised representative of the person<sup>13</sup> is generally involved. It is important that the authorised representative is provided with adequate advice, information and genetic counselling to assist them in understanding relevant matters and reaching an informed decision.

Scenario 4 describes a situation in which consent to disclose is sought from an authorised representative.

#### *Seeking consent for use or disclosure of genetic information concerning children and young people*

In situations where a genetic condition or genetic status is confirmed in a child or young person, the involvement of that child or young person in decision-making about sharing genetic information will depend on his or her age, maturity, emotional readiness and mental capacity. It is most likely that at least one parent will be involved in the consultation and will also be involved in decision-making.

The Privacy Act does not specify an age at which young people may make their own decisions about how personal information about them is handled. Under the Privacy Act, the capacity of children and young people to make a decision is assessed on a case-by-case basis rather than on attaining a prescribed age.

A child or young person can give or withhold consent if he or she has sufficient understanding and maturity to understand what is being proposed. The responsibility for exercising a child or young

<sup>13</sup> See definitions



person's rights under the Privacy Act falls to a parent or guardian, until the child reaches a level of maturity where they are able to make decisions independently.

Children may have a limited role in decision-making either because they are too young to understand or because they have mental impairment as a result of illness, injury or disability. Generally, older children should be encouraged to take a more active part in decision-making than younger children.

Involvement of a child or adolescent psychiatrist or psychologist may be of assistance, as these professionals are well placed to assist the parents and authorising medical practitioner by assessing the child's emotional maturity and perception of the situation. Resources are also available to assist in talking to children about illnesses (eg Hennig 2009).

When parents refuse consent to disclose information to genetic relatives because they wish to protect the child with the genetic condition, health practitioners may need to seek independent advice or refer the family to a genetics service.

The fact that the patient is a child does not reduce the requirement for a health practitioner to carefully consider the implications of use or disclosure without consent for the patient both now and in the future.

The situation can be more complicated if there are adopted children or children born through artificial reproductive technology (ART) where gametes have been donated. The implications of contacting unknown genetic relatives to inform them of possible genetic risk would need to be carefully considered.

Scenario 8 describes a situation where the patient is a child and a parent decides not to give consent to disclosure.

#### *When the genetic relative is a child*

Issues of competency are also relevant when determining whether genetic information should be disclosed to a child. In situations where a child does not have the maturity to make his or her own decisions under the Privacy Act, disclosure to the child's parent or guardian is permitted.

### 3.2.4 Documenting the process of consent

Documenting the process of consent should include notation in the patient's record of:

- when and by whom the patient or his or her authorised representative was informed of the implications of the identified condition or genetic status for the patient's genetic relatives;
- involvement of any other professionals (eg GPs, specialists, counsellors, ethicists) in the consent process;
- written consent if given by the patient;
- if consent is withheld, the reasons given by the patient or the authorised representative; and
- any particular issues that may have had an impact on the consent process such as the patient's language or capacity and what steps were taken to address these issues.

## 3.3 Following appropriate processes when consent is withheld

Most patients freely give consent for their genetic information to be disclosed to genetic relatives but, as discussed in Chapter 2, some may have reasons to withhold this information. In these situations, the patient's decision should be respected by allowing time for review of the decision and considering referral of the patient to a genetics service. In circumstances where an element of urgency exists, it may not be possible for expert counselling to be provided at a face-to-face appointment (particularly in rural and remote areas) but telephone counselling and support should be available.

### 3.3.1 Who is responsible for decision-making and disclosure?

Guideline 4	The authorising medical practitioner should have a significant role in the care of the patient and sufficient knowledge of the patient's condition and its genetic basis to take responsibility for decision-making about use or disclosure.
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A range of health practitioners may have a role in decision-making. However, throughout the process one senior medical practitioner will act as the authorising medical practitioner. The authorising medical practitioner should have a significant role in the care of the patient and will usually also take responsibility for disclosure. In some cases, he or she may choose to identify another professional to undertake the disclosure. In identifying professionals suitable for the role of authorising or disclosing practitioner, consideration should be given to whether they:

- have sufficient expertise in the relevant condition and its genetic basis to be able to determine whether a serious threat to the life, health or safety of genetic relatives exists and whether disclosure may lessen or prevent this threat;
- have legitimate access to this health information about the patient and family under the APPs; must access expert advice from colleagues who have specific expertise (see Section 3.3.2); and
- have an understanding of the patient's individual needs, the family situation and any factors contributing to the complexity of the situation (eg when a patient is a child or has impaired decision-making ability).

A medical practitioner must take responsibility for the process as authorising medical practitioner, even if another professional (eg a genetic counsellor with requisite knowledge of the particular condition) takes on the role of disclosing health practitioner.

It is essential that all health practitioners involved in the decision-making process have a clear understanding of their roles.

### 3.3.2 Taking a collaborative approach

Guideline 5	Prior to any decision concerning use or disclosure, the authorising medical practitioner must discuss the case with other health practitioners with appropriate expertise to assess fully the specific situation.
Guideline 6	Where practicable, the identity of the patient should not be apparent or readily ascertainable in the course of inter-professional communication.

Use or disclosure of genetic information without consent involves consciously acting against the patient's expressed wishes. It is therefore imperative that the decision to use or disclose is made only after discussion with experienced colleagues, even when the health practitioner involved is experienced in the field. In such discussions, wherever practicable, the authorising medical practitioner should not reveal the identity of the patient either verbally or in writing.

On each occasion, discussion of the case should be organised so that all involved have time to prepare. When a face-to-face meeting is not possible (eg for professionals in rural or remote areas consulting specialists in other areas), telephone conversations or conferences may be required. The outcomes of each discussion should be documented, signed, and retained in the patient's records.

In some circumstances it may also be advisable to seek ethical advice or consult a medical defence organisation.

Scenario 6 describes a complicated situation where considerable consultation between experts is needed to assess the necessity for disclosure without consent.

### 3.3.3 Decision-making about use OR disclosure without consent

#### *Is there a serious threat to life, health or safety of genetic relatives?*

When consent is withheld, the authorising medical practitioner will first need to determine whether there is a serious threat to genetic relatives. A serious threat reflects significant danger to the individual and could include a potentially life-threatening situation or one that might reasonably result in an illness or psychological harm without timely decision or action.

Consideration of the seriousness of a threat to the life, health or safety of genetic relatives will include identification of which relatives are at risk. A one in two risk of developing a serious disease in a close relative represents a serious threat but lower risk in a more distant relative is less serious, particularly as the risk starts to approach that in the general population. In some circumstances such a quantitative approach will be possible. In many situations the estimation of risk will rely on a number of factors and a range of expertise will need to be involved.

Issues for consideration when determining whether a threat is sufficiently serious to warrant use or disclosure without consent include:

- the nature of the condition, its associated risks and treatment or care options; and
- the probability that a genetic relative may also have the condition or be a carrier of the relevant mutation.

Scenario 9 describes a situation where the risk to genetic relatives is difficult to define and other measures are available to assess the risk to genetic relatives making disclosure without consent unnecessary.

#### *Psychological harm*

In some circumstances a serious threat to a genetic relative's psychological health could justify use or disclosure without consent. For example, it may be warranted to lessen or prevent a serious psychological threat to a woman associated with repeated miscarriage.

Scenario 7 describes a situation where there is a risk of psychological harm to genetic relatives from both disclosure and non-disclosure.

#### *Financial harm*

Generally, under the Privacy Act, a risk of financial harm is not considered to be a serious risk to life, health or safety. However, in some cases, a risk to an individual's financial status may result from psychiatric illness or dementia. Those psychiatric consequences may meet the test of a serious risk to life, health or safety.

### *Can the threat to genetic relatives be lessened or prevented?*

Many inherited conditions can be treated and symptoms lessened. If a serious threat to genetic relatives has been identified, the treating practitioner, in consultation with colleagues, needs to determine whether the potential to lessen or prevent the threat exists. Considerations include:

- whether the condition is preventable or manifestations treatable (eg whether the relatives can benefit from the information); and
- if the disease is incurable, whether knowledge of the condition would allow
- specific management, treatment of distressing manifestations (eg depression), and better understanding of the patient through recognition of cognitive and physical impairment (McCusker 2003).

### *Are use or disclosure without consent necessary?*

Before making a decision about non-consensual use or disclosure, the authorising medical practitioner must form a reasonable belief that such an act is necessary to lessen or prevent the identified threat to genetic relatives. Consideration should also be given to whether or not a means other than use or disclosure exists to lessen or prevent the threat (for example by including genetic relatives in a screening program).

The decision to use or disclose without consent must be made in good faith, with the practitioners involved in the decision-making on behalf of the organisation drawing on their experience, training and expertise.

### *Compliance with confidentiality requirements*

Health practitioners should be aware that information that can be disclosed consistent with the Privacy Act and these Guidelines may still be subject to the duty of confidentiality that exist outside the Privacy Act framework. Health practitioner may wish to seek legal advice from a medical defence organisation before making a disclosure.

## 3.3.4 Providing further information to a patient who has withheld consent

If the authorising health practitioner believes that the disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative, a further discussion should be held with the patient. By this stage, the patient will have had some time to come to terms with his or her own prognosis, may have attended genetic counselling and may have had a change of mind about contacting genetic relatives.

If not, discussion of the possible use or disclosure without consent should be initiated. This will include explanation of:

- the provision in legislation for health practitioners to provide information to
- genetic relatives in such circumstances;
- the basis of the authorising medical practitioner's belief that a serious threat exists and that the release of the information is necessary to lessen or prevent this threat, including the expert advice of relevant health practitioners;
- the fact that the information would be shared without directly identifying the patient or the condition or genetic status;
- the treating practitioner's continued duty of care towards the patient whether consent is given or not; and

- the continuing availability of genetic counselling for the patient, if desired.

The discussion should be factual and non-coercive, and can be reinforced with written information or other relevant materials. If consent is still not given, the patient's reasons for continuing to withhold consent should be documented. It may be advisable to request that the patient reads and signs a formal statement acknowledging that he or she:

- has been informed of the risk to genetic relatives;
- has chosen not to give consent for use or disclosure; and
- is aware that use or disclosure can take place without this consent. A sample statement is included in Appendix 2.

If use or disclosure without consent is to take place, the authorising medical practitioner should notify the patient of this decision unless there is a contrary indication for doing so. The authorising medical practitioner should explain to the patient that if the patient is unhappy with the health practitioner's decision to use or disclose their genetic information without consent, and this difficulty cannot be resolved between the patient and the health practitioner, that the patient can make a complaint to the OAIC.

The medical practitioner should be aware of the potential for patient distress and manage this appropriately.

Scenario 3 highlights the importance of ongoing provision of information, with the authorised representative of the person in the scenario deciding to pass on information to family members after several months of discussion and counselling.

### 3.3.5 Non-disclosure

In considering the details of a particular situation, the authorising medical practitioner may decide that disclosure without consent should not proceed. The decision not to proceed may be required under the Privacy Act, or may be an elective choice by the practitioner.

#### *Non-disclosure when the guidelines cannot be met*

- Disclosure without consent can only proceed if the Guidelines in this document are met. For example, the authorising medical practitioner may conclude that:
- there has been insufficient ethical consideration of the issues in this situation (see Section 2.3);
- there may be additional reasonable steps that could be taken to obtain the patient's consent (see Sections 3.2 and 3.3.4);
- he/she does not fulfill the essential criteria required of the authorising medical practitioner (see Section 3.3.1);
- there has been inadequate consultation with expert professionals (see Section 3.3.2);
- relatives are not at risk of a serious threat to life, health or safety (see Section 3.3.3);
- there are no interventions that could assist in the clinical care of the relative (see Section 3.3.3); or
- means other than disclosure are available to lessen or prevent the threat (see Section 3.3.3).

In any of these situations, the Privacy Act does not authorise disclosure without consent.

### *When the health practitioner chooses not to disclose or is unwilling to disclose*

There may be situations in which the requirements of these Guidelines are met, but the authorising medical practitioner chooses not to disclose, for example when:

- there are extenuating circumstances such that disclosure may be of little benefit to relatives;
- other services are already undertaking notification of relatives; or
- relatives have already stated that they do not wish to have this information.

Other considerations might include:

- the emotional impact of disclosure on the patient (eg the potential for suicide or violence);
- the potential negative impact on the patient's relationship with the family;
- the potential for disclosure without consent to erode the trust between the health practitioner and patient and the ramifications of this on ongoing treatment and counselling of the patient; and
- cultural factors (see Section 2.1.2).

In such situations, it may be appropriate not to proceed with disclosure without consent. Disclosure in the future may be possible if the patient changes his or her mind or the family situation changes.

It is important to remember that a health practitioner does have an ethical obligation to advise the patient to inform relatives of the diagnosis but is under no legal obligation to contact relatives about the diagnosis in the family.

Circumstances may arise in which disclosure of genetic information to genetic relatives without consent is permissible on the basis of the decision-making process and criteria included in the guidelines, but the health practitioner is unwilling to disclose (for example, because the health practitioner has a personal view that patient confidentiality should never be breached).

However, if disclosure is thought to be appropriate, the health practitioner should consult another suitably qualified and experienced health practitioner, keeping in mind the APPs and the duty of confidentiality, and consider whether it would be more appropriate for the information to be disclosed by another health practitioner.

As the legislation does not compel a health practitioner to disclose information to a genetic relative, the question may be asked whether an aggrieved relative, who has not been notified about a risk for a serious genetic condition, can take legal action against the organisation or health practitioner. As the law currently stands, there is no valid basis to suggest that the organisation or health practitioner could be liable for non-disclosure.

### *Documentation*

Irrespective of the decision made, the process of decision-making in relation to disclosure must be documented in writing. The reasons for the decision must be detailed. The fact that the patient has been advised to inform relatives must be documented.

## 3.4 The process of disclosure to genetic relatives

### 3.4.1 How does disclosure take place?

Even after a decision is made to disclose without consent, the practicalities of doing so can be complicated. It is not possible for health professionals to ascertain objectively the extent of a patient's knowledge about other family members, making it particularly important to manage disclosure very carefully.

If the patient has not given consent for disclosure, the authorising medical practitioner will usually not have access to contact details for genetic relatives. Section 16A allows an APP entity to collect personal information if it is unreasonable or impracticable to obtain the individual's consent to the collection, use or disclosure and the entity reasonably believes that the collection, use or disclosure is necessary to lessen or prevent a serious threat to the life, health or safety of any individual, or to public health or safety.

A variety of circumstances will influence how contact takes place. Written contact gives the recipient time to consider whether to seek further information, and in this sense can be perceived as non-coercive. It is suggested that a request for verification of receipt be included with written contact (see the sample form letter in Appendix 2). In some cases telephone contact may be suitable (eg when the recipient may know that the condition exists in the family and information concerning the potential risk to themselves is likely to cause distress). However, it is not appropriate to leave messages concerning private information on telephone answering machines or with someone who is not the intended recipient of the information.

Not all people who have been contacted will respond. Repeated attempts should not be made to contact non-responders because they may have made a choice not to seek further information.

If disclosure without consent is to take place, in general, the patient should be notified of this decision and advised when the disclosure has taken place.

### 3.4.2 What information should be provided?

Guideline 7	Disclosure to genetic relatives should be limited to genetic information that is necessary for communicating the increased risk and should avoid identifying the patient or conveying that there was no consent for the disclosure.
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Information provided to genetic relatives when first contacted should be worded in general terms but clearly indicate the importance of the communication. It should:

- not identify the patient or the genetic status or genetic condition that has been identified;
- simply state that a tendency to develop a potentially serious heritable disorder has been identified in the family;
- state that notification of relatives under such circumstances is permissible under the Privacy Act;
- suggest that the recipient use the contact details provided to receive further information (for example by taking the letter to their GP who could make contact for them);
- include details of the nearest genetic counselling services; and
- if possible, use a letterhead that does not identify the condition.

The information provided should not convey the fact that consent was not given for disclosure to genetic relatives.

A sample letter that may be used as a template is included in Appendix 2.

Scenario 5 describes a situation in which there is reasonable belief that disclosure is necessary to prevent harm to genetic relatives but difficulties arise in maintaining the confidentiality of the patient.

### 3.4.3 Process of cascade contact

Guideline 8	Disclosure of genetic information without consent should generally be limited to relatives no further removed than third-degree relatives.
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The Privacy Act defines “genetic relative” as including but not limited to sibling, parent or descendant. Many genetic conditions involve more than the immediate family. A step-by-step process of cascade contact allows more genetic relatives to receive information about a genetic condition. Each genetic relative who is notified about their increased risk and makes contact with the disclosing health practitioner is asked for consent to contact his or her genetic relatives. When additional genetic relatives make contact, the process is repeated. This process can provide access to genetic information for a wider cross-section of the family.

The sample letter in Appendix 2 can be used as the starting point for cascade contact. Scenario 2 describes a situation where the patient advises some

- family members to attend a genetics service and cascade contact is used to contact
- other genetic relatives.

### 3.4.4 Documenting the process

Guideline 9	All stages of the process must be fully documented, including how the decision to use or disclose without consent was made.
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The process of disclosure with or without consent should be documented, including details of:

- preliminary discussions with the patient or his or her authorised representative
- concerning the familial nature of the condition or genetic status;
- the recommendation to the patient or his or her authorised representative that
- genetic relatives be notified;
- request for consent to disclose to genetic relatives;
- refusal of consent and reasons for it;
- the identity of the genetic relatives contacted; and
- the process used to contact those genetic relatives (including a copy of any letter mailed to them).

If consent has been withheld and disclosure considered necessary, an accurate record of how the decision to disclose without consent was attained should be kept. This includes:

- the process of seeking advice from colleagues and the outcomes of these discussions;
- the basis for the belief that there is a serious threat to the life, health or safety of
- genetic relatives; and
- the basis for the belief that disclosure was necessary to lessen or prevent the threat to the genetic relative.

Situations where a decision is taken not to disclose should also be documented.



### 3.4.5 Continuing support for the patient and family

The decision of a patient to disclose, or not disclose, genetic information to relatives should have no bearing on the availability and quality of continuing care to the patient. However, if genetic information is disclosed to relatives without the patient's consent, the patient may prefer to have their continuing medical care provided by another health practitioner. The assurance of continuing care by either the same or a different health practitioner should be discussed with the patient.

# PART D SCENARIOS

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This section includes a number of scenarios that provide general guidance for authorising medical practitioners and disclosing health practitioners about meeting the requirements under section 16B(4) and acting in accordance with the Guidelines. While the scenarios centre on DNA-based testing, the Guidelines relate to genetic information irrespective of its source.

It should be noted that the scenarios are to assist compliance with section 16B(4) and the nine guidelines. Acting in accordance with the scenarios does not necessarily protect against a breach of the APPs or a breach of confidence (common law).

## When the patient chooses to contact relatives or provides consent

In the following scenarios the patient provides consent for relatives to be contacted. The process therefore falls beyond the scope of section 16B(4). Elements of standard good practice (such as documenting the process in patient records) are assumed and not highlighted in the scenarios.

### Scenario 1

This scenario describes a situation where consent is given and the provisions under section 16B(4) are not applicable. It is included here to illustrate good practice in discussing use or disclosure in the more usual situation where consent is given.

A patient who had recently been diagnosed with autosomal dominant polycystic kidney disease was referred by her nephrologist to a genetic counsellor. The patient had some knowledge of the pathophysiology of the condition and the counsellor was able to provide her with a clearer understanding of its heritability. The woman was concerned about the future health of her two children and future grandchildren. She was keen to pass any information on to her own children and was also interested in whether the condition was likely to affect her cousins and their children.

Points for consideration

- *What factors support disclosure in these circumstances?* — This is an autosomal dominant disorder with high penetrance, and each child is at 50 per cent risk of inheriting the causative mutation and of developing polycystic kidney disease. The disease can be life-threatening, often leading to chronic renal failure, and is associated with cerebral aneurysms.
- *What factors weigh against disclosure?* — Even though the patient is keen to pass on the information, she should be counselled to do so carefully and with due consideration to whether it is in her relatives' best interests (for example taking into account the age and maturity of her children and the views of their father about disclosing the information).
- *What information could be given to the patient?* — In this case, it is possible to quantify the risks to the woman's children and to give an indication of likely risks to the other relatives. Available preventive measures and treatment options could also be discussed.
- *Who might be involved in decision-making?* — In this case, a genetic counsellor has already become involved. This professional has the expertise to inform the woman of the genetic implications of the condition for herself and for her genetic relatives. The counsellor may also discuss the importance of informing genetic relatives of their increased risk in such a way

that they can choose whether or not to seek more information.

- *How might disclosure take place?* – Because the patient has consented to disclosure, the use of these Guidelines is not required. The patient could be supported with appropriate written materials, including information about support groups and counselling. The process of disclosure must be documented as part of the patient's medical record.

## Scenario 2

This scenario describes a situation where the patient advises some family members to attend a genetics service and cascade contact is used to contact other genetic relatives.

Suzanne, whose maternal grandmother died of breast cancer in her thirties, tested positive for a mutation in the BRCA2 gene. Suzanne was advised to contact genetic relatives and suggest that they make contact with the genetics service. She refused to contact her sisters or her mother, Margaret, for personal reasons, but advised her daughters to attend the service. One of the daughters came to the service for testing and readily agreed to advise Margaret. Margaret contacted the service and made an appointment. She too tested positive for the mutation. It was suggested that Margaret (now the patient) notify genetic relatives. Margaret contacted the two sisters. Both sisters attended the genetics service and Carol, the younger of the two, tested positive for the mutation. Carol gave consent for the genetics service to contact her children and grandchildren.

Points for consideration

- *Why might disclosure be advisable in these circumstances?* – The mutation identified increases the risk of breast cancer and ovarian cancer. Prophylactic mastectomy and/or oophorectomy can reduce the risk. Selective oestrogen receptor modulators (eg Tamoxifen) can also be used to reduce risk and early detection methods (eg mammography) can be used to detect the cancer when it is most treatable.
- *What factors weigh against disclosure?* – Suzanne did not consent to disclose to her mother or sisters. However, the fact that she did inform her daughters meant that a process of cascade contact could be used and there was no requirement for disclosure without consent.
- *What information could be given to the patient?* – In each case, the women need to be advised of the risks to themselves and the potential for disclosure to reduce the risk to genetic relatives. Genetic counselling should be offered to each patient.
- *How might disclosure take place?* – The process of cascade contact employed in this scenario allowed contact to be made with consent for a number of women who had the potential to carry the BRCA2 mutation, with only first or second-degree relatives being contacted in each instance. Any process of cascade contact needs to be carried out with due regard to the confidentiality of all patients involved.

## Scenario 3

This scenario highlights the importance of ongoing provision of information with the authorised representative of the person deciding to pass on information to family members after several months of discussion and counselling.

A patient with significant dementia and a history of psychosis of late onset had been diagnosed with Alzheimer disease before being seen by a neurologist in private practice.

The man had a movement disorder and progressively slurred speech consistent with Huntington disease. Genetic testing documented a pathogenic mutation in the Huntington disease gene. Initially, the patient's wife (as his authorised representative) decided not to pass on information to those at potential risk, as she was concerned about the impact of this information on her children. However, after several months of discussion and counselling, she decided to pass the information to other family members.

Points for consideration

- *What factors support disclosure in these circumstances?* — Even in incurable, slowly progressive illnesses, there is some urgency to inform genetic relatives. If they choose to have it, predictive testing for Huntington disease would allow the man's genetic relatives (siblings and their children and adult grandchildren) the potential to plan for the disease's onset and ability to make major life decisions.
- *What factors weigh against disclosure?* — The likely effect on relationships within the family and between the family and health practitioner are considerations, especially in this case where being informed of risk may cause great anxiety.
- *What information could be given to the patient or authorised representative?* Reasonable steps have been taken to determine whether the patient has the mental ability to understand this particular situation sufficiently to make an informed decision. As he has impaired decision-making ability, the information is given to his wife as authorised representative. This would include information about the course of the disease and treatment or care options.
- *Who might be involved in decision-making?* — Expert advice (eg from a psychiatrist, neurologist, geriatrician) may be required in this case to assess the mental ability of the patient and to assist in decision-making regarding the seriousness of the threat to genetic relatives and ways in which the threat could be lessened or prevented.
- *How might disclosure take place?* — In this case, the authorised representative changed her mind after being given more information and decided on his behalf that the information should be disclosed to genetic relatives. However, with or without consent, disclosure of information about progressive degenerative disorders such as Huntington disease should be done with great care and in a timely manner so that relatives can be informed of the possibility of being at risk but choose whether or not to undertake testing to find out their genetic status. Continuing family and genetic counselling may assist family members to understand the nature of the risk and come to terms with the situation.

## When consent is not given and disclosure without consent takes place

The following scenarios describe situations in which consent is not given and there is potential for disclosure to take place following the process outlined in these guidelines. The scenarios are provided to illustrate certain principles. They do not highlight every aspect of the process and cannot be used as templates.

### Scenario 4

This scenario describes a situation where an authorised representative of the patient does not give consent for disclosure. In the light of the serious threat to genetic relatives, a decision is taken to disclose to the relatives without the consent of the authorised representative.

A man with dementia came to a private clinic accompanied by his wife. In the past he had been shown to have a mutation for the Huntington disease gene. The husband was severely demented and could not communicate. Assessment confirmed that he was unable to understand his situation and give consent to inform genetic relatives of their risk and his wife was identified as his authorised representative. Information about the implications of the diagnosis for genetic relatives and consideration of disclosure that would have been given to the patient was then given to his wife. During the course of these discussions, the neurologist ascertained that the patient and his wife had not told their adult children or the patient's siblings of this risk. When the father was admitted to hospital, the three adult children supplied names and addresses for contact in the event of deterioration.

Despite careful explanation from the neurologist and the social worker on a number of occasions, as well as by other clinicians when the husband was admitted to hospital, the wife (as authorised representative) continued to refuse to notify her children of their risk.

Points for consideration

- *What factors support disclosure in these circumstances?* – The authorising medical practitioner has a reasonable belief that disclosure to the man's children is necessary to lessen or prevent a serious threat to the adult children's life, health or safety. The couple's adult children and other genetic relatives, if informed of their risk of inheriting the Huntington disease mutation, may wish to consider undertaking predictive testing. Knowledge of this risk would allow planning for the disease's onset. If a predictive test is taken, the risk of inheritance is further clarified and may influence major life decisions, as well as allowing early recognition of manifestations, such as treatable depression and cognitive changes.
- *What factors weigh against disclosure?* – Despite counselling, the children's mother, as authorised representative for her husband, is adamant that the children should not be informed of their risk. Disclosing without consent is likely to irrevocably change relationships within the family. There is the possibility that adult children could be unduly distressed, that they may already have the onset of illness or could have a prodromal psychiatric illness. It is also possible that the mother may be refusing to disclose in order to conceal non- paternity.
- *What information could be given to the patient?* – In this case, reasonable efforts have

been made to ensure that the patient's understanding is as thorough as possible. This included explaining the condition and the implications of disclosure using simple language. The neurologist then assessed the patient's ability to give informed consent. In this case the patient was severely impaired at presentation. When a person is judged as incompetent, reasonable efforts should be made to ensure that the person is, in fact, unable to understand this particular issue and its implications.

- *What information could be given to the authorised representative?* — As it has been determined that the patient lacks capacity to give informed consent, the wife as authorised representative should be given the necessary information and assistance regarding the disclosure to enable her to make an informed decision on the patient's behalf. Such information should include, for example, the likely threat to genetic relatives if they are not advised of their risk and therefore do not seek health advice, and the process for disclosure. It is important that the woman be asked to consider what her husband's wishes would have been. She could also be actively encouraged to seek further advice from a genetic counsellor.
- *Who might be involved in decision-making?* — The treating neurologist may elect to take this matter further by discussing with experienced colleagues whether or not to disclose in these circumstances. If there is reasonable belief that disclosure is necessary to lessen or prevent a serious threat, a decision may be taken to disclose without consent.
- *How might disclosure take place?* — In this case the couple's adult children could be contacted. Conditions such as Huntington disease are incurable and diagnosis can cause great anxiety. Before contacting the relatives, the disclosing health practitioner should be aware of interventions and actions that may help people who are dealing with the prodromal psychological consequences of being informed about the diagnosis, and of specific care for the relatives.

## Scenario 5

This scenario describes a situation in which there is reasonable belief that disclosure is necessary to prevent harm to genetic relatives but difficulties arise in maintaining the confidentiality of the patient.

A GP in private practice in a country town diagnosed haemochromatosis in a male patient in his late thirties. As far as the man knew, no other member of his family had been diagnosed with haemochromatosis but his mother had severe arthritis. The GP explained the likelihood of the man's parents and younger brother carrying the mutated gene for the potentially serious condition. The patient did not want to contact family members himself because he did not want his identity revealed but agreed to the GP contacting them. However, at the following consultation the patient withdrew consent, saying that he didn't want to worry his family, and he declined to be referred to a clinical genetics service.

After a telephone consultation with a clinical geneticist in private practice in the city, the GP believed that there was a serious threat to the health of the man's relatives that could be lessened and that he should disclose. However, he was concerned about protecting the patient's identity. The GP prepared a letter on his letterhead for genetic relatives. He was able to obtain

their contact details lawfully. He informed them that he had been advised that a member of the family had been diagnosed with a familial disorder. He explained that the serious nature of the condition provided exceptional circumstances in which they could be contacted without the consent of the patient concerned. He suggested that they attend their own GP or local health service, taking the letter with them.

Points for consideration

*What factors support disclosure in these circumstances?* — Hereditary haemochromatosis increases the amount of iron that the body absorbs, with excess iron being deposited in multiple organs of the body. Excess iron stores can result in cirrhosis, diabetes, cardiomyopathy, pigmentation of the skin, and arthritis. The condition is fatal if not treated and early intervention can prevent organ damage before it occurs. The GP thus has a reasonable belief that disclosure to the man's parents or younger brother is necessary to lessen or prevent a serious threat to life, health or safety.

*What factors weigh against disclosure?* — As this is a common disorder, and easy to screen for by measuring iron levels in the blood, it may be detected in relatives anyway, removing the necessity for disclosure without consent. However, early detection is preferable as late diagnosis is associated with poorer outcomes.

*What information could be given to the patient?* — In this case the patient has been provided with information about the condition and the benefits of informing genetic relatives. Disclosure to genetic relatives has also been discussed but the patient has withdrawn his consent.

*Who might be involved in decision-making?* — The GP provided the patient with sufficient information to make a decision about disclosure. He also consulted a clinical geneticist about the likely threat to the patient's relatives.

*How might disclosure take place?* — The approach taken considers the patient's privacy, the impact of the patient's diagnosis and the implications of this on genetic relatives. The GP should also notify the patient that disclosure to his relatives has taken place and that this was done in a way that reduced the likelihood of his identity and diagnosis being identifiable.

## When consent is not given and disclosure does not take place

In the following scenarios no decision is reached as to whether disclosure without consent is permissible and the circumstances remain under review.

### Scenario 6

This scenario describes a situation where the risk to genetic relatives is clear, with the potential for at least three people to benefit from the information. The scenario is provided as an illustration of how the nature of the situation and the potential for damage to relationships from non-consensual disclosure can complicate decision-making. A patient was referred to a gastroenterologist. His father had had a familial form of colorectal cancer, familial adenomatous polyposis, and had died when the patient was 12 years old. The patient had been found to carry a mutation in the APC gene. The patient had already been given an

explanation of the need to monitor people with this mutation and of the risks involved. This explanation was reinforced by the gastroenterologist, with discussion covering the importance of sharing the information with genetic relatives and the potential for disclosure to take place without consent in certain circumstances.

The patient refused to make contact with his estranged wife and their three sons. When the specialist suggested that she could contact them on his behalf, he said it was not his problem, that he did not know where they were and that he didn't want them to be contacted. However, the specialist realised that one of the sons (aged 13 years) had recently been referred to her complaining of abdominal pain. The gastroenterologist discussed the case with a senior colleague and also consulted a clinical geneticist.

Points for consideration

*What factors support use or disclosure in these circumstances?* — Because of the high risk and early onset of colon cancer (by age 40) in most individuals with an APC mutation, this situation represents a serious threat to the life, health or safety of genetic relatives that could be lessened by use or disclosure. Diagnosis before the development of cancer allows for preventive treatment eg colectomy during teenage years.

*What factors weigh against use or disclosure?* — Although a serious threat to genetic relatives exists, and this could be lessened or prevented by use or disclosure, consent has not been given and the case must be reviewed by experts in the area. Use or disclosure has the potential to compromise the relationship between the patient and the gastroenterologist, and to further compromise the relationship between the patient and his family.

*What information could be given to the patient?* — The patient has been given the information needed to understand the implications of the diagnosis for his genetic relatives. If he continues to withhold consent, discussion of the possible use or disclosure without consent should be initiated. This should cover the provision in legislation for non-consensual disclosure to genetic relatives, the basis of the belief that release of the information is necessary to lessen a serious threat and the fact that the information would not directly identify the patient or the condition. The practitioner's continued duty of care towards the patient and the continuing availability of genetic counselling for the patient should also be highlighted.

*Who might be involved in decision-making about use or disclosure?* — The gastroenterologist could make a decision regarding use or disclosure in consultation with her senior colleague and the clinical geneticist (ensuring the identity of the patient is not apparent or readily ascertainable where practicable). In doing so she would be acting in accordance with these guidelines and thus with the law. She may also choose to seek advice from

her medical defence organisation. If the gastroenterologist decides to use or disclose the information without consent, she should notify the patient of this decision.

*How might disclosure take place?* — In general clinical practice, the medical practitioner in this scenario would most likely disclose to the mother, explain the risk and provide the opportunity for DNA testing of the adolescent. If the gastroenterologist makes a decision to disclose, she could contact the mother in writing advising her to make an appointment to discuss the familial disorder. The opportunity for genetic counselling and DNA testing could then be provided. Cascade contact could then be used to reach other genetic relatives. All communications with genetic relatives would need to be undertaken with consideration of the privacy of the patient and other relatives.



## Scenario 7

This scenario describes a situation where there is a risk of psychological harm to genetic relatives. Further exploration of the situation would be required before non-consensual disclosure could proceed.

A 29 year-old man was found to have a balanced chromosome translocation during evaluation for his partner's history of recurrent miscarriages. When cells of people with balanced chromosomal rearrangement divide to create eggs or sperm for reproduction, some of the chromosomal material can be duplicated or missing. This leads to an unbalanced translocation, which often results in miscarriage or may result in a live-born child with major congenital malformations. The translocation diagnosed was considered to be unlikely to cause the man any medical problems but may have accounted for his partner's history of miscarriages. The man stated that there was no family history of children with major congenital malformations or disabilities.

Some of the man's relatives could carry the same balanced translocation, despite them being healthy. Disclosure of his genetic diagnosis could facilitate clarification of their risk of having miscarriages or may provide an explanation for miscarriages that have occurred. The man's sister was at risk of having the same translocation and potentially multiple miscarriages because of her carrier status. The man was provided with this information. He refused to advise her although she had recently miscarried and was known to be planning another pregnancy.

Points for consideration

*What factors support disclosure in these circumstances?* — Miscarriage and especially repeated miscarriages may result in high psychological burden for a woman. Even though the sister's physical health is not under any serious threat, her circumstances mean that she is at high risk of psychological damage in the event of repeated miscarriage. Disclosure would allow her to choose to clarify her carrier status by genetic testing. She could be prepared for the likelihood of further miscarriages and access support and counselling services if necessary. This would apply to other close relatives, male or female. While it may be usual for the couple's chromosomes to be checked after three miscarriages, prior knowledge of the woman's carrier status would avoid the many months or even years of delayed fertility and the psychological and physical impact that multiple miscarriages may have.

*What factors weigh against disclosure?* — Disclosure in this case may reinforce to the couple the man's perception that he is "responsible" for his partner's miscarriages. It may also affect relationships within the wider family.

*What information should be given to the patient?* — The implications for the man's genetic relatives and benefits of notifying his sister have been explained. It would be worth exploring the man's response to his new diagnosis that probably accounts for his partner's continued miscarriages and the impact that the experience is having on their relationship.

*Who might be involved in decision-making?* — The treating clinician should seek advice from other experts. Referral of the man for expert psychological counselling would also be advisable.

*How might the disclosure take place?* — The man's initial response to the possibility of disclosure may reflect an acute reaction to the diagnosis. With support, information, and the passage of time, he may subsequently agree to disclosure. On the other hand, his response could represent difficulties in the family dynamics. In a situation where the patient continues to

withhold consent and where there is reasonable belief that the threat to genetic relatives is of a serious nature and disclosure necessary to lessen the threat, informing the sister and other close relatives may be appropriate.

## Scenario 8

This scenario describes a situation where the patient is a child and a parent decides not to give consent to disclosure. The diagnosis is such that there is the possibility that a number of the genetic relatives could experience a late onset degenerative neurological condition, and female genetic relatives could experience premature ovarian failure. However, further consideration is required to determine whether disclosure would be necessary to lessen or prevent a serious threat.

The autistic symptoms of a five-year-old boy combined with family history suggested to his paediatrician a diagnosis of Fragile X syndrome. The mother's father had shown Parkinsonian symptoms and an aunt had been unable to have children. On DNA testing of the boy, a mutation of the FMR1 gene diagnostic of Fragile X syndrome was identified; a premutation of the same gene was identified in his mother. Following discussion of the diagnosis, the woman decided not to share this genetic information with genetic relatives. The woman had a number of siblings living in the same town. The paediatrician sought advice from practitioners with appropriate expertise on the seriousness of the threat to the life, health or safety of genetic relatives.

Points for consideration

- *What factors support disclosure in these circumstances?* — Fragile X syndrome is caused by a mutation of the FMR1 gene. When it occurs in a child, the mother usually carries a premutation of the gene. Males can also carry the premutation. Individuals with a premutation can experience early menopause and/or reduced fertility (women) and/or a late onset (over 50) degenerative neurological condition sometimes diagnosed as Parkinson's disease (FXTAS; more common in males). The diagnosis of Fragile X syndrome in the boy therefore has implications for a number of the woman's genetic relatives.
- *What factors weigh against disclosure?* The mother's lack of consent for disclosure is the main consideration in this case, as disclosure without consent is likely to affect family relationships and the doctor/patient relationship. The fact that the condition is not curable needs to be taken into account, although many of the symptoms can be treated.
- *What information could be given to the patient?* — The child in this case is very young and it is unlikely that he would play a role in discussion of the case. He is likely to have significant intellectual disability, and emotional and behavioural problems.
- *What information could be given to the authorised representative?* — Prior to DNA testing taking place, the mother of the child should have been given an explanation of the causes of Fragile X syndrome and the pattern of its inheritance, as well as an explanation of the significance of this diagnosis for genetic relatives, the need for them to be advised and the potential for them to be advised without her consent. She should also be aware that information allowing diagnosis in her relatives would prevent them having to undergo unnecessary interventions, some of which carry risk.
- *Who might be involved in decision-making?* — The seriousness of the threat to the life, health or safety of genetic relatives presented by the full mutation and the premutation would need to be considered by health practitioners with appropriate expertise (eg covering the areas of neurology and gynaecology as well as paediatrics and clinical genetics).

- *How might disclosure take place?* – It is possible that the paediatrician may identify another affected child in the family, thereby providing a second opportunity to notify the extended family and avoid invasive testing in other children in the family. If this does not occur, the paediatrician will need to make a decision about whether disclosure without consent is necessary to lessen or prevent a serious threat to the life, health or safety of genetic relatives.

### When consent is not given and use but not disclosure of the information takes place

In the following scenario disclosure of genetic information is not permissible under section 16B(4) but use of the genetic information within the organisation may be appropriate.

## Scenario 9

In this scenario the risk to genetic relatives is difficult to define and other measures (ie use rather than disclosure) are available to assess the risk to genetic relatives making disclosure without consent unnecessary.

A GP with considerable expertise in the management of diabetes diagnosed the condition in a middle-aged woman. The GP also treated most of the woman's immediate family, including her children and grandchildren. He explained the likelihood of other family members having a predisposition to the condition. The woman was adamant that no-one in her family should "know that she was sick or that there was a sickness in the family".

Points for consideration

- *What factors support disclosure in these circumstances?* – Diabetes is a common condition that can lead to a number of complications if left untreated. Early diagnosis and treatment can prevent many of these complications, and changes in lifestyle can delay onset of the condition.
- *What factors weigh against disclosure?* – The heritability of diabetes depends on multiple genes and their interactions with environmental factors, so the risks to genetic relatives are not clear. In addition, diabetes is easy to screen for during routine appointments, and is a common diagnosis. Given these factors, it is difficult to justify overriding the woman's refusal to consent to disclose.
- *What information could be given to the patient?* – The woman should be advised that although the risk to her genetic relatives is hard to define accurately, it would be preferable for them to know that they may be at increased risk so they can make lifestyle changes and have their glucose levels tested regularly.
- *Who might be involved in decision-making?* – In this case, the GP would not continue with consideration of disclosing genetic information without consent, as the risk to relatives is determined by a multitude of factors.
- *How might disclosure take place?* – Disclosure without consent would be inappropriate in this situation, as it would not lead to a lessening of the risk. As diabetes is a very prevalent disease, the condition is likely to be picked up by routine health screening. In dealing with other family members, the GP must ensure that his duty of confidentiality to the woman is not breached.

# ABBREVIATIONS AND ACRONYMS

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## Abbreviations

<b>AHEC</b>	Australian Health Ethics Committee
<b>ALRC</b>	Australian Law Reform Commission
<b>Amendment Act</b>	<i>Privacy Legislation Amendment Act 2006 (Cth)</i>
<b>APP</b>	Australian Privacy Principal
<b>ARC</b>	Australian Research Council
<b>GP</b>	general practitioner
<b>HGAC</b>	Human Genetics Advisory Committee
<b>NHMRC</b>	National Health and Medical Research Council
<b>OAIC</b>	Office of the Australian Information Commissioner
<b>Privacy Act</b>	<i>Privacy Act 1988 (Cth)</i>
<b>UA</b>	Universities Australia

# BIBLIOGRAPHY

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*NOTE: The bibliography was not reviewed in the 2024 minor review of this document. Revision of this information will form part of the next substantial review of these guidelines. If you require access to a document that is no longer in print or available online, please contact NHMRC Publications at communications @nhmrc.gov.au*

ALRC (2003) *Essentially Yours: The Protection of Human Genetic Information in Australia*. Australian Law Reform Commission.

AMA (2006) *AMA Code of Ethics*. Australian Medical Association.

Bernat JL (ed) (2008) *Ethical Issues in Neurology*. Third edition. Wolters Kluwer Lippincott Williams & Wilkins.

Clarke A, Richards M, Kerzin-Storarr L et al (2005) Genetic professionals' reports of nondisclosure of genetic risk information within families. *Eur J Human Gen* 13: 556–62.

Davey A, Newson A, O'Leary P (2006) Communication of genetic information within families: the case for familial comity. *J Bioethical Inquiry* 3: 160–66.

Dugan RB, Wiesner GL, Juengst ET et al (2003) Duty to warn at-risk relatives for genetic disease: genetic counselors' clinical experience. *Am J Med Gen* 119C: 27–34.

Evans JP, Skrzynia C, Burke W (2001) The complexities of predictive genetic testing *BMJ* 322:1052–1056.

Falk MJ, Dugan RB, O'Riordan MA et al (2003) Medical geneticists' duty to warn at-risk relatives for genetic disease. *Am J Med Gen* 120A: 374–80.

Forrest LE, Delatycki MB, Skene L et al (2007) Communicating genetic information in families — a review of guidelines and position papers. *Eur J Human Gen* 15: 612–18.

Gold JL (2004) To warn or not to warn? genetic information, families, and physician liability. *MJM* 8: 72–78.

Goddard B, Hurlimann T, Letendre M et al (2006) Guidelines for disclosing genetic information to family members: From development to use. *Familial Cancer* 5: 103–16

Hennig BL (2009) *Talking to Kids About Huntington Disease*. www.talkingtokidsabouthd.com. Accessed 11 February 2009.

Hudson KL (2007) Prohibiting genetic discrimination. *New Engl J Med* 356: 2021–23. Iles J (ed) (2006) *Neuroethics: Defining the Issues in Theory, Practice and Policy*. Oxford University Press, Stanford.

Kohut K, Manno M, Gallinger S et al (2007) Should healthcare providers have a duty to warn family members of individuals with an HNPCC-causing mutation? A survey of patients from the Ontario Familial Colon Cancer Registry. *J Med Genet* 44: 404–07.

Lucassen A & Parker M (2004) Confidentiality and serious harm in genetics - preserving the confidentiality of one patient and preventing harm to relatives. *Eur J Hum Genet* 12: 93–97.

Lucassen A, Parker M, Wheeler R (2006) Implications of data protection legislation for family history. *BMJ* 332: 299–301.

- Malpas P (2005) The right to remain in ignorance about genetic information — can such a right be defended in the name of autonomy? *NZ Med J* 118: U1611.
- McCusker EA (2003) The specialist neurologist and the “new genetics”. *MJA*: 179(3): 167–69.
- NHMRC (2018) *Ethical conduct in research with Aboriginal and Torres Strait Islander peoples and communities: Guidelines for researchers and stakeholders*. Commonwealth of Australia
- NHMRC (2004a) *Communicating with Patients. Advice for Medical Practitioners*. Commonwealth of Australia.
- NHMRC (2004b) *General Guidelines for Medical Practitioners on Providing Information to Patients*. Commonwealth of Australia.
- NHMRC (2005) *Strengthening Cardiac Rehabilitation and Secondary Prevention in Aboriginal and Torres Strait Islander Peoples*. Commonwealth of Australia.
- NHMRC (2006a) *Making Decisions about Test and Treatments: Principles for Better Communication between Healthcare Consumers and Healthcare Professionals*. Commonwealth of Australia.
- NHMRC (2006b) *Cultural Competency in Health: A Guide for Policy, Partnerships and Participation*. Commonwealth of Australia.
- NHMRC (2018) *Keeping Research on Track II*. Commonwealth of Australia.
- NHMRC (2007a) *Ethical Guidelines on the Use of Assisted Reproductive Technology in Clinical Practice and Research*. Commonwealth of Australia.
- NHMRC, ARC, Universities Australia (2023) *National Statement on Ethical Conduct in Human Research*. Developed jointly by the National Health and Medical Research Council, Australian Research Council and Universities Australia. Commonwealth of Australia.
- Otlowski MF (2007) Disclosure of genetic information to at-risk relatives: recent amendments to the Privacy Act 1988 (Cwlth). *MJA* 187: 398–99.
- Parker M & Lucassen AM (2004) Genetic information: a joint account? *BMJ* 329: 165–67.
- Petrila J (2001) genetic risk: the new frontier. *Behav Sci Law* 19: 405–21.
- Royal College of Physicians of London (2006) *Consent and Confidentiality in Genetic Practice. Guidance on Genetic Testing and Sharing Genetic information*. A Report of the Joint Committee on Medical Genetics. Royal College of Physicians of London, Royal College of Pathologists, The British Society for Human Genetics, London.
- Soleymani Lehmann L, Weeks JC, Klar N et al (2000) Disclosure of familial genetic information: perceptions of the duty to inform. *Am J Med* 109: 705–11.
- Suthers GK, Armstrong J, McCormack J et al (2006) Letting the family know: balancing ethics and effectiveness when notifying relatives about genetic testing for a familial disorder. *J Med Genet* 43: 665–70.
- Swartling U, Eriksson S, Ludvigsson J et al (2007) Concern, pressure and lack of knowledge affect choice of not wanting to know high-risk status. *Eur J Human Gen* 14: 556–62.
- Wagner Costalas J, Itzen M, Manick J et al (2003) Communication of BRCA1 and BRCA2 results to at-risk relatives: a cancer risk assessment program’s experience. *Am J Med Genet C Semin Med Genet* 119: 11–18.
- Wilson J (2005) To know or not to know? Genetic ignorance, autonomy and paternalism. *Bioethics* 19: 492–504

# APPENDICES

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## Appendix 1: 2014 working committee and terms of reference

### *Membership*

Chair – Dr Sandra Hacker AO, HGAC Ms Sharon Caris, AHEC

Dr Elizabeth McCusker  
Neurologist Westmead Hospital Huntington Disease Service

Dr Graeme Suthers  
Clinical geneticist  
Deputy Head, South Australian Clinical Genetics Service

Dr Samantha Wake  
Genetic Counsellor (FHGSA) Genetic Health Services Victoria

### *Ex officio members*

Professor Colin Thompson  
Chair, Australian Health Ethics Committee, NHMRC

Professor Ron Trent  
Chair, Human Genetics Advisory Committee, NHMRC

### *Observer*

Office of the Privacy Commissioner

### *NHMRC support*

Dr David Abbott

Ms Vesna Bencic

Ms Cheryl Cooke

Ms Vesna Cvjeticanin

Ms Jodie Francis

Dr Barbara Henderson-Smith

Ms Sharon Hill

Ms Cathy Mitchell

Ms Riki Porteous

Technical writers

Ms Elizabeth Hall & Ms Jenny Ramson,  
Ampersand Health Science Writing

### *Role*

The role of the Section 95AA Guidelines Working Committee was to advise the technical writer about the content of the previous version of these guidelines (which took effect on 15 December 2009), taking into account the requirements in the Privacy Act 1988 (Cth) (as amended 2006).

### *Line of reporting*

The working committee was required to:

- regularly report on its progress to the steering committee, and seek endorsement of its activities;
- through the steering committee, provide regular progress reports to the HGAC and AHEC; and
- through the Chairs of both Committees, report its progress to Council and the CEO on a regular basis.

## Appendix 2: Sample materials

### Sample Points for inclusion in a privacy leaflet

The following paragraphs related to the disclosure of genetic information could be included in privacy leaflets of health organisations in the private sector.

#### Privacy leaflet

This leaflet is about privacy, your personal information and our organisation.

##### *Your personal information is protected by law*

Our organisation handles your personal information, including your health information, in accordance with the *Privacy Act 1988* (Cth). The Privacy Act has rules about how your personal information is handled, including how it is given to others. There are special rules for health information. Generally, information about you and your health, such as test results and diagnoses, is confidential and will not be given to anyone else without your consent.

##### *Disclosing personal information without consent*

Sometimes, in special circumstances, your personal information must be given to someone else even if you do not consent. For example:

- a court may issue a subpoena requiring that we release this information to assist in resolving an investigation or a court case; or
- you might be diagnosed with a condition, such as a serious infection, that is an immediate threat to other members of the community.

In circumstances like this, we are required by law to release personal information about you. In such a situation, we will continue to provide you with ongoing care.

##### *It may be important to share personal information with relatives*

There is another situation in which personal information about you may be given to others.

People can develop a familial disease. A familial disease is one that can be inherited from one or both parents. Other people in your family may be affected, or this may be the first time that this disease has been diagnosed in your family. Being told that you have a familial disease is clearly very important for you as the affected person. This diagnosis is also very important for your genetic relatives because they may develop the disease in the future.

If you are diagnosed with a familial disease, we may recommend that you tell your relatives so that they can take action to reduce the risk, severity, or impact of the disease to themselves and their families.

Most people are willing to do this because it helps their relatives.



*The law may allow your doctor to give some information to relatives*

Sometimes, a patient may, for some reason, not want to tell relatives about the diagnosis, even though treatment and other help and support are available.

When this happens, privacy laws allow a doctor to inform genetic relatives that there is a genetic condition in the family without the patient's consent for this disclosure. This can only happen if the particular disease poses a serious threat to relatives and the information will be effective and necessary to prevent or lessen harm. The relatives would not be told what the genetic disease is or who in the family was found to have the disease. They would be advised to seek advice from a doctor.

If we decide to give information to your relatives, we will again advise you about the privacy law that allows this disclosure. Your ongoing care will not be affected by your decision.

*Do you have any questions?*

We would be pleased to give you further information and to answer any questions you may have. Please contact us.

## Statement of acknowledgement

In signing this form, I confirm that:

I (patient/authorised representative) have discussed the diagnosis of .....  
with my doctor, and

- understand that this condition is inherited and that my genetic relatives are at increased risk of developing this serious condition
- have been advised to disclose this information to my genetic family
- have been allowed time to discuss and ask questions about disclosing this information, and have been offered the opportunity to seek another medical opinion and genetic counselling
- give consent to have information regarding this diagnosis disclosed by my doctor to my genetic relatives.

OR

I (patient/authorised representative) do not give consent to have information regarding the diagnosis of a genetic disorder in my family disclosed by my doctor to my genetic relatives. I understand that disclosure without my consent may be allowed under federal privacy legislation, and that my identity would not be disclosed under this provision.

- I understand that any decision I make will not affect the care provided to me by my doctor.

Patient

Name..... Signature..... Date.....

or Authorised representative

.....

Name..... Signature..... Date.....

Doctor

Name..... Signature..... Date.....

## Form letter to relative

It is suggested that this letter be marked “private and confidential”.

Dear

Recently a genetic (blood) relative of yours was diagnosed with an inherited condition. This may mean that you and your genetic relatives (brothers and sisters and your children and other relatives) could also inherit this condition. Perhaps you are already aware of an inherited disease in the family.

While we want to respect your relative’s right to privacy, the condition has been judged to be serious enough for you to be contacted. In certain circumstances, the Privacy Act

1988 (Cth) allows for this information to be passed to genetic relatives like yourself.

This letter is not intended to create distress. Many inherited conditions can be treated and any symptoms lessened.

This letter does not give you any details of this disease, but allows you to decide for yourself whether you wish to have more information. If you do, I would be pleased to provide you with more details. Any information you give me will be treated confidentially. Please note that I cannot give you any information about other family members.

If you choose to make an inquiry, you are not committed to do anything more than receive more detailed information. Genetic testing can be done for some inherited conditions but that would only take place after allowing time to consider the full implications for you, at your request and with your consent.

Please take this letter to your GP if you would like him or her to make contact with me on your behalf. Or you may want to call the contact number listed below to arrange a meeting. Other genetic relatives may also wish to attend, or they can make individual appointments.

Appointments can be organised for discussions with suitable specialists and/or a genetic counsellor. If you live in remote or rural Australia, your GP or health worker is best placed to contact experts to advise and assist, and offer counselling if you would like more information.

It is important for us to know that you have received this letter. Even though you may not want to act on this information, please acknowledge receipt by telephoning the number below or returning the enclosed acknowledgement slip in the stamped addressed envelope provided.

I urge you to take this matter seriously as this information could be very important for the health of you and your close relatives.

Yours sincerely

Disclosing Health Practitioner

## Appendix 3: Further information

### *What is the role of the OAIC?*

The Office of the Australian Information Commissioner (OAIC) is an independent statutory office established under the *Australian Information Commissioner Act 2010*. The Australian Information Commissioner (Commissioner) has responsibilities under the Privacy Act and other federal legislation to regulate the way these agencies and organisations collect, use, store (to ensure accuracy and security of data records) and disclose people's personal information. The Commissioner investigates complaints from the public about the misuse of their personal information by agencies and organisations covered by the Privacy Act. More detailed information is available at [www.oaic.gov.au](http://www.oaic.gov.au).

### *Do these Guidelines apply to health practitioners in the public sector?*

In general, these Guidelines only apply to health service providers in the private sector).<sup>14</sup>

### *Do these Guidelines apply to research settings?*

These Guidelines do not apply to research settings. The use of genetic information in human research is discussed in Chapter 3.3 (Genomic research) of the *National Statement on Ethical Conduct in Human Research*, as in force or existing from time to time'.

### *Do these Guidelines apply to all genetics services?*

These Guidelines apply to private genetics services but not those in the State or Territory public sector.<sup>15</sup>

### *What happens if the patient with the genetic disorder is deceased?*

Currently the Privacy Act applies to living persons only. However, genetic information about a deceased person may be subject to legal duties of confidentiality. As well, information about a deceased person may have implications for the confidentiality and privacy of living genetic relatives and advice from a medical defence organisation should therefore be obtained.

The Royal College of Physicians of London publication, *Consent and Confidentiality in Genetic Practice. Guidance on Genetic Testing and Sharing Genetic information* (2006), provides a detailed discussion of this issue.

### *Where can further advice on genetics and genetic conditions be obtained?*

The Centre for Genetics Education develops resources specifically for health professionals. For more information, visit their website at: Welcome – Centre for Genetics Education - [Welcome - Centre for Genetics Education](#)

Support groups include [Genetic Alliance Australia](#) ([Genetic Support](#) | [Rare Diseases](#) | [Genetic Alliance Australia](#) | [Support for those affected directly or indirectly by genetic conditions so rare there is no support group](#))

### *Where can legal advice be obtained by health practitioners?*

Healthcare indemnity providers are generally available to provide medico-legal advice to members on a 24-hour 7-day basis.

### *Where can further advice on cultural issues be obtained?*

The NHMRC publications *Ethical conduct in research with Aboriginal and Torres Strait Islander peoples and communities: Guidelines for researchers and stakeholders 2018* and *Keeping Research on Track II 2018* provide advice about ethical conduct in research with Aboriginal and Torres Strait Islander communities. The guides are available at [www.nhmrc.gov.au](http://www.nhmrc.gov.au).

14. But note that there may be some bodies connected with States or Territories (such as a State government-owned company that provides genetic services) that fall under the definition of organisation and are therefore subject to the Privacy Act and these Guidelines.

15. See preceding footnote.

*What happens if a patient is unhappy with the medical practitioner's decision to use or disclose their genetic information without consent?*

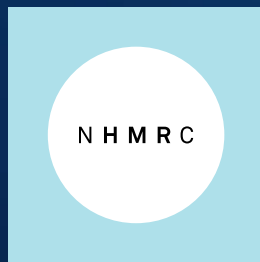
If an individual thinks a health service provider has interfered with their privacy, they can complain to the OAIC. In most cases, the Commissioner will not investigate the complaint unless the individual has first tried to resolve the complaint with the health service provider.

If the individual and the provider cannot resolve the complaint between themselves, the OAIC conciliates the complaint. As a last resort, the Commissioner can make a formal determination. The Commissioner can also investigate an act or practice that may be an interference with privacy even if there is no complaint.

Further information on the OAIC's complaint handling process is available at: [www.oaic.gov.au](http://www.oaic.gov.au).







[nhmrc.gov.au](http://nhmrc.gov.au)