



# Guidelines for Genetic Registers and Associated Genetic Material

**NHMRC**

National Health and Medical Research Council

# **Guidelines for Genetic Registers and Associated Genetic Material**

Issued by the National Health and Medical Research Council in accordance with the  
*National Health and Medical Research Act, 1992 (Cth)*.

PLEASE NOTE

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ISBN 1 86496 103 1

Electronic ISBN 0 64242 235 4

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## PREAMBLE

The NHMRC published *Guidelines for the Use of Genetic Registers in Medical Research* in 1991. Rather than providing guidelines for the gathering, use and release of data related to registers, the *Guidelines* described the conventions which applied to these aspects of register function, and addressed only one area, namely, the use of register data for research. It has become clear subsequently that there is a need for guidelines to cover all aspects of register operation, including the storage of genetic material. These revised *Guidelines*, which have increased scope, replace *Guidelines for the Use of Genetic Registers in Medical Research*, issued in 1991. They should be read in conjunction with the NHMRC *National Statement on Ethical Conduct in Research Involving Humans* (1999) and the NHMRC publication *Ethical Aspects of Human Genetic Testing – An Information Paper* (2000).

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# CHAPTER 1

## INTRODUCTION

### **1.1 Background to the establishment of genetic registers**

In 1972, a World Health Organization Scientific Group recommended that medical genetics centres should set up registers of genetically determined disorders specifically for the purpose of prevention. It was recognised that registers are the most effective way of identifying members of families who are at significantly increased risk of developing an inherited disorder or of having affected children, and that performance of such activities goes beyond the responsibilities and resources of individual doctors. Subsequently, genetic registers were established in many countries, including Australia.

Genetic registers will have the greatest impact on the prevalence and burden of disorders that are serious and relatively common, for which the risk to relatives is high, for which prevention and/or improved outcome are possible as a result of surveillance, and for which there are reproductive choices which will enable couples to avoid the occurrence of severe genetic disorders in their children.

In general, genetic registers have focussed on disorders caused by mutations in a single gene (monogenic, Mendelian disorders). These include dominant disorders such as Huntington disease or familial adenomatous polyposis and X-linked disorders such as Duchenne muscular dystrophy or fragile X mental retardation. Registers have also addressed heritable chromosome abnormalities.

The focus is broadening to meet the needs of families in which certain disorders occur with high frequency, though not as a result of mutation in just one gene eg familial bowel cancer. These disorders have a multifactorial basis and result from interaction between variations in more than one gene and environmental factors. The potential value of developing registers for these disorders will become clearer as the genes which underlie predisposition to them are identified, as the interaction between genes and environmental factors becomes better understood, if risks to relatives are shown to be substantial, if effective prevention strategies become available and if the prevention strategies are acceptable to, and implemented by, those at increased risk.

### **1.2 Characteristics of a genetic register**

- (a) The following are the key characteristics of genetic registers:
  - (i) Genetic registers address heritable disorders. Each genetic register usually addresses one disorder, or a closely related group of disorders.
  - (ii) Genetic register staff contribute to the provision of care to family members, both affected and unaffected, either directly or through health professionals. They do so by:

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- Undertaking the systematic collection of accurate and up-to-date information over a long period, aiming at complete ascertainment of family members at risk and the collection of all relevant information about them.
  - Ensuring that family members have an opportunity to become aware of their risk, and of any genetic testing, prevention, surveillance, treatment or reproductive options that may be available to them.
  - Working closely with health professionals to facilitate, coordinate and, in some cases, to provide aspects of care.
  - Bringing together pedigree and medical information relating to individuals, nuclear families and branches of the family in order to construct a single large pedigree. This aggregation of family information may improve assessments of risk, assist with identifying and contacting those at risk, provide information about disorder severity and manifestations in the particular family, prevent duplication of genetic testing, help validate genetic test results and facilitate research.

(b) Genetic register staff may also undertake other tasks including:

- (i) Disseminating clinically relevant new information to registered families;
- (ii) Conducting and/or facilitating research;
- (iii) Carrying out public and professional education; and
- (iv) Analysing the data received to permit evaluation of service provision and outcomes.

(c) Other collections that contain genetic information but which are not considered to be genetic registers:

The following are not considered to be genetic registers as they do not have all the characteristics listed in 1.2 (a) above:

- medical records kept by health professionals in private practice and by hospitals (including the files of clinical genetics units);
- records of research studies (including those retained in accordance with good research practice for a period after the research has been completed);
- databases established purely for research purposes;
- public health surveillance data sets established to monitor and study the epidemiology of disorders in the community;<sup>1</sup>
- registers which do not deal with inherited disorders;

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<sup>1</sup> The National Health Information Management Group is developing guidelines for the operation of statute-based health data collections.

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- the results of genetic tests, or collections of tissue such as histopathology samples or blood spots used for newborn screening (some such collections are mandated) held by laboratories and blood banks; and
  - data sets created to monitor the utilisation and impact of health services in relation to inherited disorders.

(d) Settings in which genetic registers may be established

The setting in which a genetic register is established will influence how it functions. Thus, staff of a hospital-based register are more likely to include health professionals who will participate in the provision of health care to registrants and have personal contact with them. By contrast, staff of a community-based register are more likely to concentrate on data collection and management and to serve registrants through health professionals rather than directly. Also, registers based, for example, in a clinical genetics unit, surgical unit or outside a health care setting are likely to have different strategies for accessing information and may focus on the collection of different types of information. Staff of a register based in a clinical genetics unit are more likely to take responsibility for contacting relevant family members than are staff of a community-based register, who will rely on health professionals to perform this function.

### **1.3 Genetic register structure**

In addition to maintaining the main database, register staff may work collaboratively, and sometimes have administrative links, with:

- A laboratory, its staff and its associated genetic material. The genetic material will have laboratory information associated with it and this will be often stored in a laboratory database.
- A team of health professionals that is involved in register operation and/or provides services to registrants.

Register function will depend on the flow of information between these parties, some of which may be in different institutions. A body, which could be the advisory committee [see 2.1 (e) (ii)], with representation from all involved institutions, may be necessary to meet the accountability and information transfer elements of register function. Transfer of information between parts of a register should only include items which are required according to written protocols and for which consent has been obtained.

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#### **1.4 Genetic registers and links to others involved in the health care of registrants**

Staff of genetic registers perform specific functions in relation to the health care of registrants but cannot provide total care of a person with the disorder addressed by the register. If registers are to be effective, register staff will need to develop good working relationships with others, including those relevant clinicians and organisations who will deliver the health care services coordinated or facilitated by staff of the register.

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## CHAPTER 2

### ESTABLISHMENT OF A GENETIC REGISTER

#### **2.1 Requirements for establishing a genetic register:**

- (a) A rationale for establishing the register, including how it may benefit registrants, how it may reduce the burden of the disorder on individuals, families and the community, and consideration of the financial costs and benefits.
  - (b) Approval for the establishment of, and acceptance of responsibility for, the register by the organisation in which it is to be based. This will include the need to establish an organisational complaints procedure.
  - (c) Resources to allow the register to function for sufficient time to achieve its aims.
  - (d) A secure space to house the register.
  - (e) A nominated keeper/custodian who will have responsibility for the register's day-to-day operation and compliance with guidelines.
    - (i) The keeper/custodian should have a clear line of accountability within the organisation that is responsible for the register.
    - (ii) The keeper/custodian should be assisted by an advisory committee, with appropriate membership, which can provide advice in regard to the development and application of the register's ethical guidelines and operating procedures. Among its members, the committee should include:
      - the keeper/custodian;
      - at least one health professional with experience of caring for persons with the disorder(s) in question;
      - at least one member of a registered family (who might be a representative of a community group which provides information/support/services to affected families);
      - a representative of the institution which takes responsibility for the register;
      - a representative of the users of register information; and
      - if large amounts of data are to be stored and analysed and/or used for research, inclusion on the advisory committee of a person with epidemiological/statistical/data management expertise and/or a person with relevant research expertise, respectively, should be considered.
- It is envisaged that each register will have only one advisory committee, even when parts of the register are in several institutions.

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- In the future, registers may be established which address several unrelated disorders or an institution may find that it is responsible for several registers and wish to amalgamate them or their advisory committees. If this occurs, the membership of the advisory committee(s) will need careful consideration. Clinical expertise and registrant representation relevant to each disorder covered by the amalgamated register should be retained.
  - (iii) It is desirable that the keeper/custodian be a health professional with expert knowledge of the disorder(s) in question. If this is not to be the case, such a health professional should be a member of the advisory committee.
  - (iv) It is the responsibility of the administering institution to appoint a replacement if the keeper/custodian is unable, for any reason, to continue in the role.
- (f) Appropriately qualified, skilled and experienced staff to carry out the functions of the register. Particular care should be taken to appoint staff with appropriate clinical training, skills and experience if the register is one whose staff interact directly with families.
  - (g) Written ethical guidelines that address privacy/confidentiality, cultural sensitivity and register procedures. These guidelines should define the consents to be sought from registrants, procedures for collection and storage of genetic information and material, and procedures for the use of, access to and disclosure of genetic information and genetic material. When members of specific ethnic/cultural groups are likely to form a large proportion of the potential registrants, the guidelines should be developed in consultation with the relevant group(s).
  - (h) Definition of the number and roles of the staff of the register. These individuals will be in a privileged position with regard to access to information held by the register.
  - (i) Compliance with relevant state and/or national legislative requirements. For example, persons wishing to establish a genetic register should seek advice as to statutory confidentiality provisions which apply in their State or Territory prior to establishing the register.
  - (j) Facilities for the storage of genetic material, if the register is one which is associated with the collection and storage of such material. The storage facility may be a laboratory that is an administrative part of the register or an independent laboratory. In either case, the laboratory should be accredited by the National Association of Testing Authorities, Australia.
  - (k) Review and approval of the register's ethical guidelines and procedures by an ethics committee, which will commonly be a Human Research Ethics Committee (HREC), of the institution in which the register is to be based.

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## CHAPTER 3

### RECRUITMENT OF REGISTRANTS

#### 3.1 Identification of potential registrants

In these guidelines, a registrant is a person who has given consent for identified information about him/her to be collected, stored and used in specific ways by genetic register staff.

- (a) A person may be invited to become a registrant because:
- (i) he/she has the heritable disorder which is the focus of the register; or
  - (ii) he/she has a blood relative with the heritable disorder that is the focus of the register. The potential registrant may have inherited the mutant gene responsible for causing, or creating a predisposition to, the disorder. Staff of each register will define the level of risk that will be used to decide whether to approach a family member regarding inclusion in the register. The level of risk depends on the closeness of the relationship between an individual and the closest affected family member. A relative who is initially considered at too low a risk to be approached may become eligible if his/her risk increases. This may occur if a close family member develops symptoms of the disorder or is shown by genetic testing to have inherited the mutant gene; or
  - (iii) he/she is the current or previous spouse/partner of a person with the heritable disorder which is the focus of the register. While not at increased risk of developing the disorder in question, his/her genetic information may be needed to assess the risk that a child of the relationship has inherited the disorder. He/she may also be a person with whom register staff communicates, if he/she is the legal guardian of a child of the relationship.
- (b) Potential registrants may be identified in various ways.
- (i) They may be identified by register staff from the information provided by a registrant who identifies other family members (see Sections 4.2.2(a), 4.2.2 (b), 4.4, and Chapter 6).
  - (ii) Register staff may notify relevant health professionals and community-based support groups that the register exists, and ask them to discuss the register with their patients and members respectively and, provided that the potential registrant gives consent, to refer him/her to register staff.

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- (iii) Register staff may search medical records, after appropriate ethical review and approval by an ethics committee, which will commonly be an HREC, followed by an approach to potential registrants through their health professionals as in (ii) above.
  - (iv) The register's existence may be advertised in the media, followed by self-referral. In cases of self-referral, and provided that the registrant consents, his/her doctor, if one exists, should be identified and made aware that the registrant has come forward.
  - (v) Individuals may come to learn of the register's existence through other sources and may refer themselves. Again, provided that the registrant consents, his/her doctor should be identified, if one exists, and made aware that the registrant has come forward.
  - (vi) Consenting registrants may be referred by staff of a register based in a different region.
  - (vii) It is possible that in the future, individuals with genetic susceptibility to certain disorders who are identified by population screening may, after giving consent, be referred to register staff by the health professionals advising them about the consequences of the screening test result.

Potential registrants, described in 3.1(b)(i), (ii), (iii) and (vii) above, may not consent to participation in a register because they do not wish to know more about the disorder, do not wish to have contact with another group of health professionals, do not wish to confront the emotional consequences of the disorder, or may perceive little benefit from participation. Such wishes should be respected.

(c) Concern for those approached as potential registrants

If the register is one where its staff approach potential registrants (having received consent from an existing registrant to do so), the staff should be sensitive to the physical and emotional state of the persons concerned and consider carefully the timing of any approach and the demands which will be placed on the persons by the registration process.

### **3.2 The first contact with the genetic register**

Register structure and protocols will determine the nature of the first contact. For some registers, health professionals refer potential registrants but do not take part in the process of explaining the register in detail or obtaining consent. Thus, the first contact is with register staff and an ongoing relationship is established. For other registers, the potential registrant's health professional explains the register, obtains consent and remains the registrant's main link to the register. In this case registrants may never have direct contact with register staff.

(a) Explanation of how the potential registrant was identified

Before seeking a person's consent to be registered, the referring health professional or register staff should explain to the person how he/she was identified as a potential registrant and the register's aims, structure and way

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of working, and seek consent for registration and specific aspects of register function (see Chapter 4).

(b) Provision of information about the register and obtaining consent

The registrant should receive written or videotaped information about the register at this time. An interpreter should be present if the potential registrant has difficulty communicating in English. A potential registrant may wish to bring to this meeting a supporter/adviser, for example a family member, a friend or a representative of his/her ethnic or cultural group, such as a cultural liaison officer. The person's consent can then be sought for registration and, if consent is given, the various additional consents set out in Section 4.2 can also be sought, if relevant to the function of the register.

(c) Gathering information for the register

At this or other early meetings, personal and family information will be obtained, a pedigree will be constructed, and risk will be assessed for register purposes. It should be recognised that construction of a pedigree involves obtaining and recording information about family members without consent. Such information should be handled with particular care until consent can be sought (see Section 4.4).

(d) Identification of relevant health professionals

The professionals involved in the registrant's health care should be identified so that communication can be established.

(e) Provision of counselling

Counselling is primarily the role of the managing health professional. However, when first contact is with register staff, some general counselling may be offered if requested or if register staff detect that it would assist the registrant, provided that register staff have the necessary training, knowledge and skills. The registrant's managing health professional will usually undertake or arrange more detailed counselling and advice, individualised risk assessment, diagnostic investigation and/or management at another time. Managing health professionals may ask specific register staff to provide some of these services if the register has staff members with appropriate training, knowledge and skills.

(f) Referral to health professionals

Register staff may refer the registrant to a health professional if none has been involved so far and it is appropriate to do so.

(g) Explanation of registrant's role in involving other family members

It will be mentioned early on that other family members may benefit from participation in the register, and that the registrant may be asked to introduce register staff to his/her relatives (see Section 4.2.2(a) and Chapter 6).

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## CHAPTER 4

### CONSENT

#### **4.1 Obtaining consent from the registrant for inclusion in the register**

In general, consent should be obtained before identified information about a person is included in a register. If for any reason that person is unable to give consent eg intellectual disability, mental disorder or health disorder, consent should be obtained from a person or organisation who can legally give consent. Consent should usually be in writing. However, depending upon the circumstances of the person eg if a person is illiterate or cannot write, verbal consent, appropriately recorded, is acceptable. The process by which each non-written consent is obtained should be recorded. The following information should be provided in an appropriate form before consent is sought:

- (a) Participation in the register is voluntary. At any time, the registrant can request that identified information and genetic material relating to him/her be removed from the register (or that identifiers be removed from such information and material) and can withdraw the consents outlined in Section 4.2.
- (b) The aims of the register and how participation may help the individual and his/her relatives. In some cases there may be no immediate benefit from participation. Circumstances that could potentially disadvantage individuals should also be mentioned eg disclosure of information under subpoena.
- (c) An outline of the structure of the register and how it operates, including any computer linkages to laboratories or clinics which may form part of the register or work with register staff.
- (d) The relationship of register staff to the registrant and to the health professionals involved in his/her health care. This should include how register staff propose to communicate with registrants and their health professionals. Registrants should be asked to notify register staff of change of address and, in case this is inadvertently overlooked, to identify another family member as a point of contact.
- (e) The register's policy on providing feedback about the progress of the register and the communication of new research findings which may be of interest or relevance to registrants as a group, or to registrants as individuals. The registrant's views on feedback should be sought. Registrants should be informed if the register's policy is to not provide feedback.
- (f) The role the registrant may have in introducing register staff to other family members.
- (g) Who is responsible for the register, that is, the names and contact details of the keeper/custodian and the organisation which has taken responsibility for the register.

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- (h) The nature of the information and genetic material to be collected and stored.
- (i) The proposed duration of storage of information and genetic material.
- (j) The sources of register information eg the individual, the family, health professionals, medical records, laboratories, death records, and other registers, and the sources from which register staff may obtain genetic material eg laboratories. Explanation of information sources should include how information is collected initially and how it is updated.
- (k) The procedures which are in place to ensure the confidentiality of information and to protect the privacy of registrants. Registrants should be informed that they will determine to whom information about them may, and may not, be given.
- The registrant may be asked to consent to disclosure of his/her information to, and access to his/her genetic material by, others including family members and researchers whose research has been approved by an HREC.
  - Registrants should be made aware that register staff may be required to disclose information about them, and/or release their genetic material, to a court or persons/organisations to whom disclosure is authorised or required by law.
  - Register staff will not disclose identified information or the fact that the person is a registrant, or release genetic material, without the registrant's written informed consent. Situations requiring specific consent include disclosure to insurers, employers, family members who do not have the registrant's prior permission, and researchers if the registrant has not previously consented to use of his/her information or genetic material for research. Registrants should be informed about the possible consequences of disclosing genetic information to third parties, for example that such information might be sought and used in determining eligibility for life insurance.
  - Registrants should be informed of the register's policy on disclosure of information and the giving of genetic material in potentially identifiable (coded) or de-identified form. For example, requests for disclosure could come from family members wishing to know whether a mutant gene has been identified in the family and the nature of the mutation, and from researchers having HREC approval.
  - Collation of the results of genetic tests performed on multiple family members can potentially demonstrate non-paternity and non-maternity. Non-paternity or non-maternity should be disclosed in only the most exceptional circumstances as to do so may cause serious and permanent social and emotional harm to individuals and families. Registrants should be informed of the register's policy on disclosure of such information.

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- l) Whether the registrant can access the information that the register has about him/her and the procedure for doing so. Access will allow registrants to check, and if necessary to arrange correction of, the information.
  - (m) The uses of register data, including whether information is routinely transferred to another body, for example if the register has components in more than one institution, or if certain information is required by law.
  - (n) The uses of any stored genetic material eg for confirmation of diagnosis, for the study of genes involved in the process which results in the disorder with a view to identifying clinically relevant information, and for research. The *National Statement on Ethical Conduct in Research Involving Humans* (1999) provides guidelines for access procedures for the research use of tissue samples (Section 15) and genetic material (Section 16).
  - (o) Register information and associated genetic material may be used, after HREC approval, for research which, in time, may result in the development of a product of benefit to those with the disorder in question, or other related disorders. For example, the register's genetic material and information may contribute to discovery of the gene responsible for the disorder, followed by the patenting of a genetic test based on that discovery. The development of such a product is likely to occur in the private sector and may have commercial potential, but it is unlikely that registrants will receive any financial benefit from the use of their information and material for the research.

## **4.2 Obtaining other consents**

From those who consent to inclusion in the register, the following additional consents/instructions should be obtained, if relevant to the work of the register:

### ***4.2.1 Consents related to the registrant***

- (a) Consent for register staff to obtain additional information about the registrant, such as confirmation of diagnosis and additional information from medical records, health professionals, laboratories, databases that hold relevant information or other specified persons.
- (b) Consent for register staff to obtain a sample of the registrant's previously collected and stored genetic material from laboratories and to store it as part of the register. The material may be as extracted DNA or RNA, or as tissue samples. The intended duration of storage should be specified. For example, some registers will wish to store genetic material indefinitely because of its research potential while others will wish to dispose of it once any proposed genetic testing has been completed. It should be noted that current National Pathology Accreditation Advisory Council guidelines require indefinite storage of genetic material by diagnostic laboratories. Register staff should reassess the situation before disposing of genetic material. For example, new tests may have been developed with potential to provide the registrant with useful

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information, or the registrant may have died leaving his/her genetic material as an irreplaceable resource for the family.

- (c) Consent for register staff to arrange collection of a blood sample so that genetic material (DNA, RNA) can be obtained and stored. The material may be used:
  - (i) to confirm a clinical diagnosis made in a symptomatic registrant;
  - (ii) to try to identify the specific gene mutation responsible for the disorder in the registrant's family; and/or
  - (iii) to provide a source of genetic material for future presymptomatic, predictive or carrier testing, if requested by the registrant. As a rule, such testing will be arranged by the registrant's managing health professional with appropriate counselling and specific consent.<sup>2</sup>
- (d) If relevant, consent for register staff to approach the registrant if new knowledge makes it possible to provide genetic testing not available at the time of registration.
- (e) The registrant should be asked to specify what may be done with his/her identified information and stored genetic material after death, and his/her wishes should be recorded.
- (f) Consent to be approached by register staff with an invitation to participate in future research studies.
- (g) Consent for register information and genetic material to be used by register staff, and other researchers, in coded or de-identified form for HREC-approved research.

#### **4.2.2 Consents related to others**

- (a) Consent to initiate an agreed process by which nominated relatives will be invited to participate in the register. The person making the initial contact will usually be the registrant who will provide his/her relatives with preliminary information about the register (ideally in writing). He/she will request permission from the relatives for register staff to make contact, in order to explain the aims and procedures of the register in detail and to seek consent (see Chapter 6). The relatives could be blood relatives or relatives by marriage.
- (b) Consent for the registrant's children to be included. In general, consent should be obtained from both parents. While parents or a legal guardian may give consent for a child to be included, new consent should be sought from the child once he/she reaches the age at which he/she is legally able to give

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<sup>2</sup> The reader is referred to the NHMRC publication *Ethical Aspects of Human Genetic Testing – An Information Paper* (2000) for a more detailed discussion of genetic testing.

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his/her own consent. If the child is a mature minor at the time of registration, register staff should seek the child's consent in addition to that of the parents or legal guardian.

- (c) If the registrant has the disorder, and the gene mutation causing or predisposing to the disorder has been identified, consent to describe the mutation to other health professionals/laboratories so that the information can be used to provide genetic testing to other family members without the need to duplicate the work involved in finding the mutation. The consent should also cover other tests which may provide diagnostic information about the disorder. If the registrant consents, he/she should specify whether the information may be identified or must be de-identified.

### **4.3 Consent related to deceased persons**

Consent to include information about, and genetic material from, deceased individuals should be sought from their legal/authorised representative, usually their senior available next-of-kin, unless it is impossible or intrusive to do so. The obtaining of consent from a person would be 'intrusive' if approaching that person caused distress, either because of intrusion into his/her private life or because the request for consent awakened painful memories eg the loss of a loved-one, or raised a topic that the person did not wish to consider. Only the information set out in Section 4.4 paragraph 5 may be retained, without identifiers, if such consent cannot be, or is not, obtained.

### **4.4 Information which register staff obtain without consent**

The effectiveness of a genetic register requires the taking of a family history to determine who else in the family had the disorder in the past, who is known to have it now, who might unknowingly have it now (that is, have manifestations but are symptom free at present) and who might be at risk of developing it in the future.

When the taking of a family history occurs in the context of the doctor-patient relationship it is a routine and necessary part of the medical consultation. It may identify risk for the development of a disorder in the future in the patient and his/her offspring and contribute to diagnosis and treatment of an existing disorder. The fact that identified relatives are spoken about in this context is generally considered acceptable because the information is given and used only to provide health care to the patient and is treated confidentially within the doctor-patient relationship.

The collection and recording of the same information by a genetic register is different because the information is given and used not only for the registrant's health care but also for other purposes. Although the registrant can consent to providing his/her information and to its use by register staff and others, he/she cannot give that consent for information about other family members. For this reason, some consider provision of family information to involve a breach of the privacy of those family members who are identified and about whom personal information is given. Registers should distinguish information that identifies people who are not registrants and not disclose it in identified form without the consent of

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the identified person. The source, date of acquisition, absence of consent and reasons for that absence should be recorded. In general, the persons identified should be approached, in due course, so that their consent can be sought for inclusion of their information in the register.

Prior to obtaining that consent, register staff may use the identified family information, obtained from the registrant, to search the register's database. This is done to determine whether the registrant is a blood relative of registrants already known to register staff. If a link is identified, there may be immediate benefits to the registrant and his/her close relatives. For example, if the mutant gene responsible for the disorder in the family has already been identified in another branch of the family, predictive testing can be offered immediately, and without duplicating the process of mutant gene identification.

Sometimes it will not be possible for register staff to obtain consent to retain identified information provided by a registrant about a relative. For example, the registrant may provide his/her relative with information about the register but the relative declines contact. Or a registrant may be unwilling to contact his/her relative or, having agreed, does not do so. In these circumstances, register staff should remove identifiers from the information provided earlier by the registrant. The information which remains in the register, usually represented by a symbol in the pedigree, will be the position of the individual in the pedigree, the individual's gender, whether or not the individual was considered to have the genetic disorder and whether the individual is alive or dead. Even though de-identified, information of this type has the potential to improve the effectiveness of the register by facilitating recognition that branches of a family are part of the one kindred [see Section 1.2(a)].

At the time of constructing the pedigree there will be relatives of the registrant in whom register staff have no professional interest other than to confirm that the diagnosis provided by the registrant is correct. If the relative is deceased or if there are circumstances in which it would be intrusive or potentially upsetting to approach a living relative for consent, confirmation of diagnosis may be obtained without consent provided that identifiers are removed once the confirmed diagnosis has been recorded in the register.

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## CHAPTER 5

### CONFIDENTIALITY

#### 5.1 Confidentiality guidelines

- (a) The organisation responsible for a register should ensure the confidentiality of the information for which it is responsible and the privacy of registrants.
- (b) Written procedures should be developed that address the confidentiality of information held in a register and the privacy of its registrants.

These should address disclosure of information to the following:

- (i) Staff of the register

The keeper/custodian should ensure that staff understand their obligations in regard to privacy and confidentiality, and establish policies and procedures to protect the privacy and confidentiality of registrants. These policies should extend to those working closely with register staff, such as laboratory staff and health professionals who provide services to registrants in association with register staff. Each institution housing a register should have a process for dealing with unauthorised disclosure of register information, and all those who use register information should be made aware of the consequences of unauthorised disclosure.

- (ii) The registrant

If asked, register staff should provide a registrant with access to register information that relates to him/her [see Section 4.1(l)]. Before doing so, the reasons for the request should be assessed so that the information sought by the registrant can be provided in the most helpful way. Sometimes the registrant will simply wish to see, or to check the accuracy of, the information held by the register. If so, access should be provided, preferably in the presence of a register staff member who can answer questions and can assess if the registrant needs advice from the health professional responsible for his/her care. At other times the registrant will be hoping to find information to gain a better understanding of his/her medical situation. If this is recognised beforehand, register staff can suggest that the information be sought from the health professional responsible for his/her care so that it is up-to-date, complete and relevant to the current clinical situation of the registrant. Sometimes, however, register staff will have appropriate training, skills and experience and, if present at the time the information is accessed, can assist the registrant in coming to a correct understanding of his/her situation.

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(iii) Blood relatives and spouse of the registrant

Blood relatives are persons who share genes as a result of inheritance from a common ancestor. The more general term 'relatives' is used in this document to include both blood relatives and spouses/partners of blood relatives.

Information that relates to a registrant may only be provided to his/her blood relatives or spouse with his/her consent. Care should be taken not to inadvertently disclose registrants' confidential information to other family members, even if they are also registrants.

With regard to deceased registrants, such information may be provided without consent to a spouse or relative who is the registrant's senior available next-of-kin.

Register staff should be aware that legislation about adoption and infertility treatment may exist which requires an administrative procedure to be followed prior to the release of information to children who have been adopted out and children conceived by gamete donation.

Information of significance to health, such as the risk of developing a heritable disorder, will usually be communicated by a registrant to relevant blood relatives and his/her spouse. However, the sharing of important health information may not occur. Knowing of the existence of the register and of the disorder in the family, blood relatives or the spouse of a registrant may approach register staff directly for information. Blood relatives will be seeking information of relevance to their own health, while spouses will usually be seeking information because of concern about the well being of the children of the relationship. If such a situation arises, register staff should not release identifying information without the consent of the registrant. However, it may be possible to address the concerns of relatives without releasing identifying information. For example, it may be possible to reveal the mutation present in the family without revealing the identity of the individual who was tested to identify the mutation.

A registrant (or senior available next-of-kin of a deceased registrant) may refuse to allow disclosure to relatives of information of significance to the health of those relatives. If this happens, register staff should ensure, by involving the registrant's treating practitioner or general practitioner as a trusted counsellor, that the registrant understands the potential consequences of such a decision for the health of family members and, indirectly, for the registrant him/herself in terms of relationships within the family if a preventable adverse health outcome occurs. Ultimately, the registrant's decision should bind register staff. However, there may be rare circumstances in which a keeper/custodian considers that the risk to the health of relatives is sufficiently large, serious, imminent and potentially preventable that consideration should be given to breaching the registrant's

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confidentiality, by disclosing identifying information about the registrant (see Section 6.1(c), paragraphs 2 and 3 regarding disclosure of non-identifying information in the absence of consent). Before doing so, the keeper/custodian should consider the potential for professional censure or legal action if confidentiality is breached, and should take appropriate advice, including that of an experienced clinician with a sound understanding of the disorder and its impact on individuals and families, the advisory committee, the HREC of the institution which is responsible for the register, and the administration of that institution. If the keeper/custodian ultimately decides to inform family members against the wishes of the registrant, the registrant should be informed of the decision and the reasons for it.

(iv) Health professionals and other registers

Information may only be provided to health professionals and other registers with the consent of the registrant. At the time of registration, each registrant will identify those health professionals and organisations to whom information may be given. With the passage of time, these may change and the registrant should inform register staff of the changes and update his/her consent. Encryption should be used if identified information is to be transferred in electronic form to another register.

(v) Those with access to laboratory databases

Genetic registers will often arrange the collection, storage and testing of genetic material. This, with its associated genetic information (diagnosis and any test results) will be stored by the laboratory/ies that work with register staff. The information will be stored in the laboratory's database, some parts of which may be accessible through an organisational information system network. The person with administrative responsibility for the laboratory should ensure that access to the genetic information is restricted to authorised persons, who will be those persons/organisations who/which have the registrant's consent to access the information.

(vi) Researchers, including register staff

Research using information and genetic material held by a register should comply with the NHMRC *National Statement on Ethical Conduct in Research Involving Humans*, in particular the sections addressing Epidemiological Research, Use of Human Tissue Samples, and Human Genetic Research, and be approved by an HREC. In addition, the research should be approved by the register's advisory committee.

Research using register information should be conducted using the minimum level of personal identification needed to carry out the research. Sometimes it will be necessary to carry out the research using identified information, for example when the research requires contact with the individual, and in this circumstance, consent for the specific research study will have been obtained. Sometimes it will be sufficient

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to use potentially identifiable (coded) information, as for example when the research involves the pedigree and associated clinical and laboratory data. Sometimes it will be appropriate to use de-identified information, as for example when DNA and associated clinical information is provided to an external researcher who is interested in the genetic basis of the disorder in the group of registrants as a whole rather than in its genetic basis in an individual. Subject to HREC approval, it may be permissible to undertake record linkage (the combining of information from two or more data sets) using identified information initially to bring the two or more sets of data together and then to work with the combined data set in potentially identifiable (coded) or de-identified form.

HREC approval should require researchers to undertake to keep confidential any information provided to them by register staff, to keep the information and genetic material secure, to return any identified information and remaining genetic material to register staff on completion of the research, and not to use the information and genetic material for any purpose other than that specified in the research proposal.

It is the responsibility of the keeper/custodian to ensure that, if it is proposed to provide genetic material to a researcher, sufficient material is retained to meet the future clinical needs of registrants. For example, some DNA from a registrant with familial adenomatous polyposis should be retained until such time as the mutation in the APC gene has been defined in one affected family member and confirmed in another.

Research results using register information or genetic material should not be published in a form that permits identification of individuals.

- (vii) Others, including insurance companies, employers and financial organisations (See also (viii) below)

Information about a registrant may only be released to such entities with the written informed consent of the registrant, or other person (eg legal representative or senior available next-of-kin) or organisation (eg guardianship board) able to give legally valid consent.

Requests for information from such entities will often comprise a letter, not seen by the registrant, which details the information sought and which is accompanied by a general release signed by the registrant. If in doubt about disclosing the information, register staff should consider contacting the registrant to ensure that he/she knows what information has been requested, is aware of the consequences of providing it, and approves its disclosure.

Rarely, register staff may be approached for information by such an entity without the knowledge or written consent of the registrant. In this event, register staff could decide not to respond to the inquiry or could respond that they cannot confirm or deny knowledge of the person. Sometimes, even though consent has been given, register staff

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will consider that the information requested would be more appropriately provided by the managing health professional, and this option should be discussed with the registrant.

(viii) Such persons or organisations to which disclosure is required by law

Unless a register is established under statutory protection, access to the information it holds can potentially be sought under subpoena, and register staff could be compelled to provide it. This could occur, for example, if a registrant is involved in a court case to which the information is relevant eg in relation to a life insurance claim, workers compensation case, family law or paternity matter. However, a subpoena only requires production of documents to the court, which must then decide, after argument to it, whether all or some of the documents will be released and to which of the parties to the case.

Further, there are organisations that have a statutory right to information if it is relevant to their functions and some of these could conceivably have an interest in genetic information. These include the Department of Social Security which may access information relevant to payment of a benefit and the Police who may access information through a search warrant.

Freedom of Information (FOI) laws may allow access to documents held by a register, but only where the register is held by a government agency. It is unlikely that a registrant would seek information about him/herself under FOI as register staff would usually provide such information to the registrant freely. An applicant (eg a relative of a registrant) could seek disclosure of information under FOI claiming that it is "joint", as could be the case with some genetic information. All legislation, except that in the ACT, provides that access to information relating to another person will not be granted unless the other person has been given a reasonable opportunity to make submissions that the information is exempt, and those submissions have been considered. The common category of exempt documents that is most relevant to registers is the category which comprises documents, the disclosure of which would involve unreasonable disclosure of information relating to the personal affairs of any person, including a deceased person.

The registrant should be informed immediately if access to his/her information or genetic material is sought under subpoena or other legislative requirement so that, if he/she so desires, he/she has the opportunity to challenge the request.

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## CHAPTER 6

### CONTACTING FAMILY MEMBERS

#### **6.1 The value of contacting family members**

The effectiveness of a register is increased if it is able to identify all those family members who may have inherited the disorder or who may pass it on to their children.

Genetic registers, as defined in these guidelines (see Section 1.2), address heritable disorders and are established with the specific aim of providing relatives of registrants with the opportunity to obtain information about the disorder in question. This implies that experienced clinicians with a sound understanding of the disorder and its impact on individuals and families will have judged the disorder serious enough, and the potential benefits to registrants great enough, to warrant establishing the register. The implications of that judgement are that maintaining an effective register will require that relatives of registrants be contacted.

Experience of clinicians involved in establishing and maintaining registers will be a valuable guide to the issues and sensitivities involved in contacting relatives. A consideration of relevant ethical obligations can also be useful.

(a) Some ethical considerations

The process of contacting relatives, in order to inform them about a register, can involve tension between relevant ethical obligations.

Respecting registrants involves having their consent before disclosing the fact of their registration and possibly other personal information, such as whether or not they have the disorder in question, to relatives. Equally, it should involve accepting registrants' refusal to disclose information about them. Where registrants agree to disclosure, they should be involved in determining to which relatives the information will be offered. In order to participate in this process, they should be informed about the information that needs to be disclosed and given an understanding of what the information could mean for the relatives eg the emotional and social consequences.

Respecting those relatives who are to be approached involves either having their consent to be informed or respecting their refusal to be informed. In addition, avoiding harm to those relatives who are to be approached involves deciding whether informing them could be beneficial (and not solely harmful) to them. Factors to be taken into account in reaching that ethical judgement include the probability of their having inherited the disorder, the morbidity associated with the disorder, whether preventative or preparation strategies are available, whether early diagnosis and treatment can lead to improved prognosis, and the age, marital status, and physical and mental health of the relative. What cannot be known by those making the ethical

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judgement is whether or not these relatives would in fact value being told the information, either because they would expect to be made aware of their situation or because they would use the information when planning for the future. Showing them how the information can help such decisions would be an important part of the contact. Another way to consider this question is whether it is fair for relatives, who may already be, or may become, affected or whose children may already be, or may become, affected to bear the preventable burdens of the genetic disorder for which they are at risk.

(b) Where registrants agree to inform their relatives

The process for making contact usually begins with referral of the first family member to the register who, if he/she consents to be on the register, is then asked to introduce register staff to other family members.

This may be done by providing the registrant with the following documents for forwarding to relatives: a letter of invitation to participate, a description of the register and a consent form that, when signed and returned, provides register staff with permission to make contact with the relative. Letters should be in the relatives' preferred language. When a relative is illiterate, the information can be conveyed orally, and the relative can contact register staff by phone. It is left up to relatives to contact register staff if they wish to participate.

(c) Where registrants agree to inform their relatives but do not do so, or agree that relatives should be informed but do not wish to carry out the task personally

Most registrants will support informing their relatives about the register and will agree to make contact with them. Having agreed, registrants may, for one reason or another, not do so. Consequently, after an appropriate interval, it should be determined whether or not the letter of invitation has been passed on to relevant relatives. This should be done by the person who arranged with the registrant for contact to be made and, depending on the way in which the register operates, may be the managing health professional or a member of the register staff. If the letter of invitation has not been passed on, and if the registrant no longer wishes to contact relatives, the managing health professional or register staff should try to identify another registrant from the same family who is willing to do so. The same process should be followed when a registrant states at the outset that he/she does not wish personally to undertake the task of informing relatives.

When no such intermediary can be identified, and when family members are at risk of having children with a serious disorder, or are themselves at high risk of a serious disorder, especially when the disorder can be prevented or interventions exist to reduce its burden, it should be considered whether, in the specific situation, informing such a relative will be ethically justified. The justification for doing so should rest on possible benefit for the family member and not on the value of that family member to the register. A decision to inform a relative under these circumstances should involve an

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experienced health professional, who may be the managing health professional, a member of the register's staff or someone advising register staff.

The consent of a registrant should be sought. However, if the consent of a registrant cannot be obtained, it may still be considered that disclosure may be justified ethically. Again, the decision to inform a relative under these circumstances should involve an experienced health professional, who may be the registrant's managing health professional, a member of the register's staff or someone advising register staff. In disclosing information to a relative, especially without a registrant's consent, respecting the registrant(s) would involve keeping the confidential information to a minimum. For example, family members could be informed that the disorder is present in the family, that there is a possibility they might have inherited it and that more information can be obtained from their doctor, the registrant's managing health professional or from register staff. See also Section 5.1 (b) (iii) paragraph 6 for discussion of the situation in which consent is not given, or is refused, and it would be impossible to inform relatives without revealing confidential information which identifies a registrant.

- (d) Where relatives do not wish to receive information about the register

Sometimes, a registrant will be aware, from what one of his/her relatives has said in the past, that the relative would clearly not wish to be informed of genetic information derived from the registrant. Others, when first approached by the registrant, may be unwilling to be informed. In both these situations, the wishes of the relatives should be respected.

It may be difficult for a registrant's managing health professional or register staff to be sure that a family member does not wish to receive genetic information or information about a register. For example, forgetfulness or loss of the form may explain non-receipt of a signed consent form. A formal refusal could be based on inadequate understanding of information provided to the family member by a registrant. Under these circumstances, it should be considered whether there is justification for devising a process to assess such family members' awareness of their situations. A family member or a health professional may, with the consent of a registrant, be a suitable person to determine that refusal to receive information is based on a sound understanding of the issues.

- (e) Where relatives could be harmed by information about a register

It should also be considered whether receiving information about a register could harm relatives. This could occur, for example, when a person is unaware of the family history, the disorder is serious and not preventable or treatable eg Huntington disease, and the person could be vulnerable eg has depression or is stressed by some aspect(s) of his/her life. In some such circumstances, it will clearly be inappropriate to attempt to inform the relative. In others, the situation will not be clear and the risk to the relative may be high and imminent and the disorder serious and preventable. Here,

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coming to a decision about whether or not to disclose information must be made by persons with expert knowledge of the disorder and detailed current knowledge of the person who is to receive the information. They should include the health professional(s) caring for the person and the health professionals working with, or on the staff of, the register. A close family member, who knows the person well and will act in his/her best interests, should be involved in decision making.

## **6.2 Setting limits to family contact**

While staff of a genetic register will endeavour to identify and contact all family members at increased risk, they will need to define how far the pedigree will be extended. Each register will determine a cut-off level of risk below which a family member will not be contacted [see Section 3.1(a)(ii)]. Register staff may also need to set limits to the resources that will be expended to trace and to make contact with an at-risk family member.

## **6.3 Recognition of the role of registrants' health professionals**

A genetic register is only one way by which family members are made aware of their risk and of the options for addressing it. Health professionals caring for them in the community and in hospitals can, and do, undertake this task. Their role should be recognised by register staff who should endeavour to establish and maintain communication with relevant individuals and organisations eg general practitioners and specialists caring for family members, clinical genetics units and interstate and international registers. Collaboration avoids unnecessary duplication of work, the provision of consistent information to family members and enables register staff to gather together information about all branches of the family.

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## CHAPTER 7

### SECURITY OF A GENETIC REGISTER

#### **7.1 Facilities to ensure the security of register information and genetic material**

The keeper/custodian should ensure that register information and any associated genetic material are protected by such security safeguards as are reasonable in the circumstances to take against loss and against unauthorised access, use, modification or disclosure. The keeper/custodian should be aware of relevant standards such as Australian Standard AS 4400-1995, *Personal privacy protection in health care information systems*. A register should be housed in a secure location, with access only possible for authorised staff, and information and genetic material should be securely stored when not in use.

#### **7.2 Facilities to ensure integrity of register information and genetic material**

A register should have appropriate facilities, equipment and qualified staff to ensure the integrity of stored register information and any associated genetic material.

#### **7.3 Working with and transferring information**

When carrying out their role, register staff receive and release information in identified form eg when communicating with doctors caring for the registrant. In general, when working with registrants' information in-house, it is appropriate for register staff to use identified information provided that the register is secure and its staff has been properly educated about the importance of confidentiality and privacy. The use of identified information in-house reduces the chance of transcription errors as data are handled, aids linkage of branches of families and allows staff to respond more quickly to queries from registrants. Each individual will also have a code, often a number, which register staff can use to uniquely identify that individual when his/her name is not needed for register function. Information that is to be transferred electronically to others should be encrypted and transferred in potentially identifiable (coded) or de-identified form.

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## CHAPTER 8

### AMALGAMATION AND WINDING UP OF GENETIC REGISTERS

From time to time, there may be reason to amalgamate two or more registers. For example, a breast cancer register and a colon cancer register may join to form a familial cancer register. Similarly, one familial adenomatous polyposis register may take on the responsibilities of another such register that is being wound up.

The needs of registrants should be paramount in such circumstances. In the case of amalgamation, the institution(s) responsible for the new entity should be defined and institutional support obtained, HREC approval should be sought, a new keeper/custodian should be nominated and new procedural guidelines written.

Prior to amalgamation of registers, or the winding up of a register, register staff should communicate with registrants explaining how the change will affect them and seeking their views. In the case of amalgamation they should have the opportunity to withdraw from the register. In both cases, they should have the opportunity to ask that their information be returned to them or forwarded to the health professional of their choice, or disposed of. However, when a register is wound up, it is likely to be in the best interests of registrants if a copy of the register's data is retained by the responsible institution. With regard to genetic material at the time of winding up of a register, registrants' views should be sought as to whether the material can be disposed of or, if it remains of potential use to the registrants or their families, whether they would like it stored in some other repository. In such cases, register staff have an obligation to try to find a suitable repository. Alternatively, consent can be sought for the use of the material in de-identified form for HREC-approved research, and the material transferred with HREC approval to a research group with an interest in the disorder in question.

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## APPENDIX 1

### REFERENCES

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## APPENDIX 2

### GLOSSARY

**De-identified (not identifiable, anonymous) genetic information or genetic material**

The process of de-identification can be irreversible if the identifiers have been removed permanently or the genetic information or material has never been identified. This genetic information or material is referred to as “de-identified”. It should be recognised that the term “de-identified” is used frequently in documents other than the *National Statement on Ethical Conduct in Research Involving Humans* (1999) to refer to sets of data from which only names have been removed. Such data may remain “potentially identifiable”.

**Genetic material**

Any source of DNA or RNA which can be tested to obtain genetic information. It thus includes cells, whether as single cells or as part of tissues, and extracted DNA or RNA.

**Health professional**

The term “health professional” is used frequently in this document. When the reference is to a person undertaking the medical management of a registrant, “health professional” will almost always mean “medical practitioner”. In some instances, however, the term could mean medical practitioner and/or other professional, such as a genetic counsellor, social worker or nurse, who provides services to members of registrants’ families.

**Identified genetic information or genetic material**

Genetic information or material that allows the identification of a specific individual is referred to as “identified” information or material. Examples of identifiers may include the individual’s name, date of birth or address. In particularly small sets of data even information such as a postcode may be an identifier.

**Potentially identifiable (coded, re-identifiable) genetic information or material**

Genetic information or material may have identifiers removed and replaced by a code. In such cases it is possible to use the code to re-identify the person to whom the genetic information or material relate, that is the process of de-identification is reversible. In these cases the data are referred to as “potentially identifiable”.

# **The National Health and Medical Research Council**

The National Health and Medical Research Council (NHMRC) is a statutory authority within the portfolio of the Commonwealth Minister for Health and Aged care, established by the *National Health and Medical Research Council Act 1992*. The NHMRC advises the Australian community and Commonwealth; State and Territory Governments on standards of individual and public health, and supports research to improve those standards.

The NHMRC advises the Commonwealth Government on the funding of medical and public health research and training in Australia and supports many of the medical advances made by Australians.

The NHMRC also develops guidelines and standards for the ethical conduct of health and medical research.

The Council comprises nominees of Commonwealth, State and Territory health authorities, professional and scientific colleges and associations, unions, universities, business, consumer groups, welfare organisations, conservation groups and the Aboriginal and Torres Strait Islander Commission.

The Council meets four times a year to consider and make decisions on reports prepared by committees and working parties following wide consultation on the issue under consideration.

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