

Ethical Code

Governing the Provision of

Genetics Services

Produced by:

**The NSW Genetics Service Advisory Committee
Statewide Services Development Branch
NSW Health Department
Locked Bag 961
NORTH SYDNEY NSW 2059**

Ph: 02 9391 9520

For further copies contact:

Better Health Centre
Ph: 02 9954 1193
Fax: 02 9955 5196

State Health Publication No: (SWS) 980068
ISBN 0 7313 4036 1

June 1998

Contents

1 Introduction	1
2 Case Study Scenarios	3
Family matters - the shared implications of genetic information	
Scenario A Implications of a high risk result for breast cancer	3
Scenario B Insurance Issues	4
Scenario C A breach of confidentiality	4
Scenario D When should the children be told?	5
Predictive/presymptomatic testing for genetic disorders	
Scenario E Predictive testing for Huntington disease	6
Reproductive choices/prenatal testing	
Scenario F Prenatal diagnosis for an adult onset disorder	7
Scenario G Fetal sexing	7
Scenario H Cultural and religious beliefs and emotional issues in genetic diagnosis	8
Storage of DNA	
Scenario I Research on stored samples	9
Screening for carrier mutation status	
Scenario J Testing for cystic fibrosis	10
Newborn screening	
Scenario K Retention of samples	11
3 Ethical Framework	12
Justice	12
Autonomy	13
Beneficence	14
Non-Maleficence	15
4 Genetic Testing Procedures and Practices with Specific Ethical Issues	16
Family matters - the shared implications of genetic information	16
Predictive/presymptomatic testing	17
Reproductive choices/prenatal testing	17
Storage of DNA	18
Screening for carrier mutation status	19
Newborn screening	19
Genetic registers	20

1 Introduction

"Genetics and biomedical technology open up vast avenues for research and can provide humankind with much needed therapeutic tools. But, where human life and dignity are at stake, technology cannot be left to govern ethics...."

WHO (1995)¹

"The search for meaningful ethical principles to guide us through difficult personal and collective decisions arising from the applications of modern genetics will be an endless process".

David Suzuki (1989)²

New genetics and biomedical technology can bring to society enormous benefits, but at the same time has potential for harm. To enable the achievement of its goals and values within a framework of ethical principles and to ensure the interests and well-being of the public are served, the NSW Genetics Service Advisory Committee has developed a code which addresses the responsibilities of health professionals and limitations of testing.

The NSW Genetics Service

The service is a network of agencies (laboratory, clinical, educational and administrative) and professional disciplines which provide genetics services to the people of New South Wales in a coordinated and integrated way. The service is organised on a statewide basis in order to achieve equitable access to high quality care and to use resources efficiently and economically.

Genetics Services

Genetics services are available to all, and provide information and education, counselling, risk assessment, diagnosis, laboratory testing and clinical management where appropriate for genetic disorders and birth defects.

Genetics Service Providers

Specialised services are provided by professionals such as clinical geneticists, genetic counsellors, fetal medicine specialists, genetic social workers, biochemical, molecular and cytogenetic scientists and specialists in genetic education.

Other health professionals such as specialist clinicians, general practitioners and allied health personnel as well as community support groups are partners in the provision of this service.

Genetic Disorders

Genetic disorders result usually but not always, from a change in the hereditary (genetic) material contributed by the parents at the time of conception. Symptoms may be apparent at birth or may occur later in life. Genetic disorders can affect growth, development and all aspects of health. They include for example: Down syndrome, spina bifida, some muscle diseases, cystic fibrosis, haemophilia, thalassaemia, Huntington disease; and some forms of cancer, osteoporosis, heart disease, intellectual disability, psychiatric and behavioural disorders.

Goals of the NSW Genetics Service

Goal 1

To ensure provision of, and access to, genetics services appropriate to the needs of the people of NSW

Goal 2

To optimise utilisation of information and services relating to genetic disorders and birth defects

Goal 3

To reduce the impact of genetic disorders and birth defects on affected individuals, individuals at risk and their families

More explicit details of the goals of the NSW Genetics Services can be found in:

- *The Goal and Objectives of the NSW Genetics Service*
- *Genetics Services in NSW Five Year Plan 1993-1998*

available from NSW Department of Health.

The shared implications of genetic inheritance

Because genetic disorders are family health problems, a diagnosis in one member has implications for other family members.

The shared implications of genetic inheritance and some aspects of genetic testing procedures and practices raise specific ethical issues. The purpose of this document is to identify and elaborate on ethical issues which are special to the practice of genetics and different from those associated with other medical interventions.

Report Format

Case Study Scenarios

The following section contains scenarios drawn from real life experiences to illustrate the ethical issues which arise in genetics consultations, practices and testing procedures and to provide the context for the application of ethical principles.

The two subsequent sections contain references back to the scenarios to bridge the gap between theory and practice.

Ethical Framework

The principles of justice, autonomy, beneficence and non-maleficence form the code of ethical conduct in the practice of medical genetics.

Genetic Testing Procedures and Practices

Key ethical issues, capabilities and limitations of genetic testing procedures and practices are identified, and the responsibilities of health professionals are addressed.

2 Case Study Scenarios

The following scenarios are intended to illustrate the dilemmas which are faced by individuals, families and genetics health professionals. They exemplify the importance of accurate information, counselling, consent, privacy, confidentiality, a full understanding of the implications of a genetic diagnosis, limitations of genetic testing, and the responsibilities of those involved.

Family Matters - The Shared Implications of Genetic Information

Implications of a high risk result for Breast Cancer

Scenario A:

Susan, a 40 year old woman, embarked on a family research project. During the course of the research, she discovered other family members hitherto unknown to her, who live abroad. While she knew that her mother had been diagnosed with breast cancer at aged 43, she was unaware that her mother's 5 sisters and 3 cousins had also been diagnosed with breast cancer, some at relatively early ages.

Given this history, she consulted her GP who advised her about the availability of predictive genetic testing through a mail order system in the USA. She could utilise this without her mother or other relatives having to know.

The result indicated that Susan had inherited a predisposition to develop breast cancer which meant that she had an 80% chance of developing breast cancer during her lifetime. Being a private person and because there was some discord within the family, Susan did not wish to discuss this result with other family members. However, she did tell her 18 year old daughter who now has a 50% chance of having inherited the same faulty gene. She also requested that her GP not communicate this information to her 3 sisters or 3 brothers, although her GP did suggest she inform other family members. Two years later, her older sister, Barbara, who lived interstate, was diagnosed with advanced breast cancer. She was unaware of her risk for developing breast cancer.

Issues:

- **duty of care**
- **privacy and confidentiality**
- **obligations to other family members**

Dilemmas:

This scenario highlights the importance of health professionals providing full information prior to testing concerning the possible results and their consequences.

- ? **What obligation does the GP have toward his/her patient versus obligations to other family members, if they are not also patients of the GP?**
- ? **What is the situation if Susan's sisters and brother were also the GP's patients?**
- ? **Should the GP have made stronger efforts to encourage Susan to share the information with her family to minimise the harm which followed?**

Insurance Issues

Scenario B:

Just after Susan (Scenario A) had received her genetic test result, her younger sister, Mary, applied for life insurance. Susan and Mary are both patients of the local GP. The GP was asked to provide relevant medical information for the insurance policy. When asked about family history of cancer, Mary answered honestly that her mother had breast cancer but that there was no other family history. She was not aware of the research undertaken by Susan. Having only one other family member with breast cancer would not be considered significant in determining if there was an inherited predisposition to breast cancer running in the family. She is also of course unaware of the result of Susan's genetic test.

A Breach of Confidentiality

Scenario C:

Questions then arise about the doctor's and the patient's responsibility towards relatives. In a different scenario, Susan, after careful presentation of the information, may agree to an approach being made to the relatives for the purpose of alerting them to their potential risk. Susan's name or test results need not be made known to the relatives, and these details should not be released without the patient's permission. However, sometimes relatives will be able to infer the identity of the family member from whom the inquiry originated, and sometimes they will be able to infer his or her test result. In these cases, approaching the relatives will in practice involve breaching confidentiality with the patient.

Issues:

- **duty of care**
- **privacy and confidentiality**
- **access to information by third parties**

Dilemmas:

- ? **Is the GP obliged to reveal to the insurance company his knowledge of the family history?**
- ? **If he does reveal the information about Susan to the insurance company, what guarantee is there that the company will not use this information in the assessment of risk for Mary in underwriting any policies issued to her?**

Dilemmas:

- ? **An individual's right to privacy versus responsibilities to others**

When should the Children be told?

Scenario D:

A 45 year old male develops symptoms and is diagnosed with polycystic kidney disease. He will need to have his blood purified periodically by an artificial kidney machine (haemodialysis). People with the disease usually die a few years after symptoms appear, although dialysis and transplants can prolong life for up to ten years. He and his wife have two children aged 16 and 18. Both parents know that the condition is genetically determined and may have been passed on to their children and may be passed to their children's offspring. The parents insist that hospital staff not tell the children the nature of the disease because it would frighten them unnecessarily, inhibit their social life and make them feel hopeless about the future. The staff are concerned that the children may innocently involve their future spouses and children.

Issues:

- **privacy**
- **obligations to other family members**

Dilemmas:

- ? **Should the children be told?**
- ? **To what extent would the following factors be relevant to this decision:**
 - **if the condition were less severe in its effects**
 - **if effective treatment existing or seemed likely to be found shortly**
 - **if the chances of the children manifesting the illness were low**
 - **if the condition were likely to affect the children, if at all, only very late in life**
 - **if the children were much younger?**³

Predictive/Presymptomatic Testing for Genetic Disorders

Predictive testing for Huntington Disease

Scenario E:

Huntington disease (HD) is a neurological degenerative disease which has its onset in most people between the ages of 35 and 60 (approx.) There is no cure for this disorder which is slowly progressive over around 10 years and involves a deterioration in movement, cognition and generalised functioning. Death usually results from respiratory illness.

HD is an autosomally dominant inherited disorder. A child of an affected person has a 50% chance of inheriting the faulty gene which causes the disorder. Genetic predictive testing is now available for persons over the age of 18 who have an affected parent or relative, which will tell them in almost all cases whether they will develop the disease at some stage in their life. Worldwide, of those eligible for the test, only around 15% of people have taken up the option of testing.

Harry is a 25 year old man whose maternal grandfather died some 10 years ago from Huntington disease. Harry's mother has therefore a 50% chance of developing HD. She decided to have the predictive genetic test and has been shown to have the faulty gene. She will definitely develop HD at some time and Harry and his two sisters are now at 50% risk of developing HD.

Harry is an air traffic controller. He loves his job and feels he could perform his duties most adequately for many years, irrespective of whether he carries the faulty gene for HD or not. He does not wish to have the genetic test. His employer is unaware of his family history

Issues:

- **privacy**
- **employment issues**
- **ownership of information**
- **access to information by third parties**
- **rights of the individual versus public good**

Dilemmas:

- ? **Do employers in industries involving public safety have the right to demand family health history information?**
- ? **In cases where genetic predictive testing is available for disorders which may impact on public safety, do employers have a right to predictive testing information about an individual whose current health status is excellent?**
- ? **Who should decide who can access it?**

Reproductive Choices/Prenatal Diagnosis

Prenatal diagnosis for an Adult Onset Disorder

Scenario F:

Patricia is married to Harry (Scenario E) and is now 12 weeks pregnant, but has not told Harry. She is in a state of anxiety and is concerned that they might have a baby who will later develop HD, although she is not sure that she would terminate the pregnancy if the fetus was shown to have the faulty gene for HD.

She requests prenatal diagnosis and DNA testing of the baby from her fetal medicine specialist, but does not want Harry to know. If this is undertaken, a result which shows that the baby has the faulty gene means that Harry must also have the faulty gene and will develop HD. A negative test result on the baby will relieve Patricia's anxiety about having a child who will develop HD. However it does not indicate whether Harry has the HD faulty gene as there is only a 50/50 chance that he could pass on the faulty gene, even if he had it.

Issues:

- **privacy**
- **rights of the father not to know his genetic status in relation to HD versus the right of the mother in choosing prenatal diagnosis**
- **confidentiality**
- **responsibilities and obligations to relatives**

Dilemmas:

If the baby is shown to have the HD faulty gene, which must have been inherited from Harry, Patricia and the fetal medicine specialist now have information about Harry's HD status which is not known to Harry himself.

- ? **What responsibilities do Patricia and the fetal medicine specialist have in relation to his occupation?**

Fetal Sexing

Scenario G

A 21 year old woman requested prenatal testing in her first pregnancy because her spouse wants a "boy". She also informed her doctor that a second cousin had Duchenne muscular dystrophy (DMD), a genetic disorder which is carried by females and expressed in males. She wanted prenatal testing to exclude this as well.

Dilemmas:

- ? **What are the implications of prenatal testing in this situation?**

Cultural and Religious Beliefs and Emotional Issues in Genetic Diagnosis

Scenario H:

A couple who did not support termination of a pregnancy due to their particular religious beliefs came to genetic counselling regarding the diagnosis of thalassaemia in their first child. She had died at 6 months of age from severe anaemia which did not respond to treatment. The couple were told that a baby from any future pregnancy had a 1 in 4 chance of being affected with the condition.

The mother, who had been the chief care giver during the short life of her daughter felt that she could not cope with the same demands of caring for another child with the same condition nor of watching another baby die. She felt that she would want prenatal diagnosis in a second pregnancy and, despite her religion, would terminate the fetus if it was affected.

On the other hand, the father was adamant that there was to be no prenatal testing and no termination as this was against their religious beliefs. He maintained that they would pray for an unaffected baby (for which they had 3 out of 4 chances) and have faith in the outcome.

Issues:

- **reproductive choices**
- **personal beliefs**

Dilemmas:

- ? **If these opposing positions remain, does one individual have the right to impose his/her view on the other?**
- ? **Is there any common ground which could be explored for these two people?**

Storage of DNA

Research on Stored Samples

Scenario I:

Anne, now 30 years old, has a rare genetic disorder, affecting only about 25 people in Australia. In the course of the diagnosis of the disorder when she was a child, samples of skin and bone were taken for testing and subsequently stored in the pathology laboratory, as is common practice for such tissues. Consent for the tests was obtained from Anne's parents. At that time it was not envisaged that these stored samples could be a source of DNA.

Research on the disorder has been undertaken over the last 5 years and DNA has been extracted from the stored samples. Consent was not sought from Anne, or her parents, to conduct research on the samples and on the extracted DNA. The research results in the discovery of an important gene and the gene is patented, providing the researcher and the Institution where the research was undertaken, with considerable funding for on-going work.

Issues:

- consent
- ownership of DNA
- patenting of DNA
- public good

Dilemmas:

- ? **Who has ownership of the stored sample, and the DNA extracted from it?**
- ? **What are the researcher's obligations in relation to the obtaining of consent from persons whose DNA they want to use?**
- ? **Should the rarity of the disorder or the benefits of research to others in the community have any impact on Anne's right to refuse consent for the research on her DNA?**

Screening for Carrier Status

Testing for Cystic Fibrosis

Scenario J:

A couple are told by their GP that their baby son has had a positive screening test for cystic fibrosis (CF). CF is one of the inherited disorders for which newborn babies are routinely screened. It is an autosomal recessive disorder causing respiratory and digestive problems. When both parents are carriers of the mutation, there is a 1 in 4 chance with each pregnancy that they will have a child affected by CF. Carriers of the mutation do not develop CF.

A follow up diagnostic test shows that the baby is not affected by CF but that he is a carrier of the faulty gene. He will not develop CF but he may have children with CF if his partner is also a carrier. The parents of the child are tested and only the father is shown to carry the CF mutation.

There is a 50% chance that their children will be unaffected carriers of the CF mutation, just like the father. The parents are very anxious to learn if their other two children aged 5 and 7 are also carriers of this faulty gene. It is not the policy of the genetics unit to test children under the age of 16 for carrier status which has no impact on their subsequent health. The parents are insistent and believe that they, as guardians of the children, should be able to decide what medical tests and procedures are performed.

Issues:

- **testing of minors**

Dilemmas:

- ? **Should the young boy be told he is a carrier of a genetic disorder before he turns 16?**
- ? **Should the other children in the family be tested for their carrier status?**

Newborn Screening Program

Retention of Samples

Scenario K:

Agnes and Robert have a daughter Sarah aged 12 years. Their older daughter Margaret had cystic fibrosis and died in 1985.

Agnes and Robert wish to start another pregnancy and ask about the possibility of prenatal diagnosis. They undergo DNA testing and the CF mutation is found in Robert but not in Agnes. (She must have a mutation but it is not one that the laboratory can detect). They are advised that prenatal testing might be possible if there was DNA or tissue stored from Margaret, for example from an autopsy. They don't know of any such stored DNA or tissue.

Agnes and Robert are surprised to learn that there is probably a blood spot from Margaret, retained after routine newborn screening tests, which could be used. Agnes vaguely recalls that Margaret had a newborn blood test but she does not recall whether anyone told her what it was for or asked for her consent to proceed with it.

The blood spot is obtained and prenatal diagnostic testing is successful.

Issues:

- consent for testing
- storage of DNA
- testing of minors

Dilemmas:

- ? **Did the circumstances by which the laboratory obtained Margaret's blood sample meet good ethical standards? If not, how could the process be improved?**
- ? **Was the laboratory ethically justified in retaining the sample without the knowledge of Margaret's family?**
- ? **Agnes and Robert want Sarah's carrier status determined, which is possible now that the blood spot has been obtained. From an ethical viewpoint, should that proceed?**

3 Ethical Framework

Ethical conduct will be discussed under the principles of:

- **Justice**
- **Autonomy**
- **Beneficence and Non-Maleficence**

which are directed towards the general public, the patient or consulting family, other family members and the unborn.

Justice

Scenarios
Refer to Section 2

Justice can be interpreted as fair, equitable and appropriate treatment. Concerning genetics services, it includes respect for human diversity and avoidance of discrimination. In health care, justice addresses two main themes:

1 In the Australian context, there is a right to a minimum standard of health care which encompasses values concerning public good and social justice. This implies that services should be delivered efficiently in order not to deny opportunities for other people to use services which might be of benefit to them. The interests of one individual or group of individuals should not disadvantage others.

- **Respect for people with disabilities**
 - recognises the intrinsic value of individuals regardless of their disorder
 - acknowledges that provision of testing to enable the avoidance of a particular disorder does not devalue persons with that disorder
 - acknowledges the need for support for people with disorders and for those making difficult decisions concerning the avoidance of a particular disorder
- **Respect for the patient’s or family’s decisions**
 - recognises that informed understanding and autonomy should underpin a patient’s decisions
 - accepts that individuals are free to decide whether or not to make use of facilities
 - accepts that options should be put in a non-judgmental way and choices supported

A - K
E
E

2 The allocation and distribution of limited resources to enable:

- **Equity of access to:**
 - services) regardless of place of residence,
 - information) ethnicity, gender, religion, age or disability

Autonomy

Scenarios
Refer to Section 2

Autonomy is the principle of self-determination. As an individual right it may have to be balanced against the rights of others and is therefore not absolute.

Essential to the maintenance and achievement of autonomy for individuals using the services are:

Information and counselling:

- providing accurate information on the implications of testing **A**
- providing information about the shared nature of genetic information and that the patient's problem is potentially a problem for other family members **A**
- recognition of the potential for conflicting interests among family members **A,H**
- respect for the individual's right to decline to receive information that his/her own body can yield **E**
- working with families and individuals to balance the individual's right to privacy with the duty of care to other family members as in the case when information would allow effective treatment of the condition **A**
- provision of accurate and comprehensive information for rare and complex disorders: to help the individual to understand the medical facts, to appreciate the concepts of inheritance and risk, to consider options, understand the consequences, choose a course of action free from coercion and make the best possible adjustments to each stage of the situation as it unfolds **E**
- awareness that the nature of genetic testing may involve clashes of cultural or religious beliefs, particularly in prenatal diagnosis and termination of pregnancy **H**
- support for the individual and family members to cope with the emotional repercussions of a genetic diagnosis or procedure, particularly concerning issues such as shame and grief **H**

Consent:

- assuring that consent is obtained on the basis of provision of all relevant information **I,K**
- obtaining and recording of consent for genetic tests and related procedures taking into account the complexity of the issues and the emotional context in which decisions are made. **A**
- obtaining and recording of consent to retain samples, specifying the purpose for which storage is permitted **I,K**
- assurance that tissue samples acquired for one purpose will not be used for another without consent **I**
- obtaining and recording consent for participation in research except as otherwise permitted by an Institutional Ethics Committee **I**

Duty of confidentiality:

- assurance that health professionals do not reveal genetic information without the consent of the person
- the duty of confidentiality may be overridden only in special circumstances which relate to the public good. If, in a developing doctor-patient relationship, the doctor foresees a situation in which there may be a need to set confidentiality aside, he or she should signal this to the patient.

Beneficence and Non-Maleficence

Beneficence is the obligation to ensure that the policies and practices of genetics professionals are to the good of patients.

Essential to the promotion of beneficence is:

- allowing people to make choices about reproductive options **E,H**
- permitting the early detection of treatable disease **J**
- permitting the identification of risk for a treatable disease or preventable disease eg breast cancer **A**
- permitting the identification of risk for non-treatable diseases, thereby enhancing the ability of people to be responsible for their own lives by enabling informed choices **A,E**
- reducing the likelihood of decisions being imposed on individuals or families by others **E**
- reducing hardship and suffering caused by the impact of genetic disorders on individuals and families **D,H**
- providing support and advocacy for individuals and families affected by genetic disorders
- encouraging people to accept their own responsibilities with regard to the needs and rights of other family members **A,C**

Ethical Framework

Non-maleficence - *Primum non nocere* - above all, do no harm

Scenarios
Refer to Section 2

Beneficence and maleficence are subjective terms. Given the shared implications of genetic inheritance and disorders, what is to the good of one individual may result in harm or disadvantage to another. Inevitably, there must be a balance of responsibility to the individual, the family and the public good.

Practically all interventions by health care professionals have a capacity to cause harm. Health care professionals have to exercise judgement as to whether any particular intervention is justified by the balance of potential benefit and potential harm. They must maintain and employ high levels of professional skills, knowledge and attitudes.

Some harmful outcomes arise from obvious causes such as failure to perform a surgical procedure with due skill and care, prescribing the wrong medication for an illness or conveying incorrect information. Some causes of harmful outcomes may be less obvious.

Actions which may avoid harm include the following:

- gauging or attempting to gauge the person's prior level of understanding of a subject being discussed A
- providing correct information in comprehensible language A-K
- providing verbal and written information in word and manner and in a culturally sensitive way to minimise creating fear and anxiety beyond that which is unavoidable H
- providing information or services in a way which takes account of the ethnic, cultural, moral, religious or other relevant personal background and views of the recipient E
- providing information or services in a way which does not diminish the autonomy nor invade the privacy of the individual A-K
- intervening only with consent A
- acting only on consent which is based on adequate information A,C
- reinforcing the idea that disclosure of genetic risk to a person raises the issue of intra-familial duties to warn and protect family members from harm.

4 Genetic Testing Procedures and Practices with Specific Ethical Issues

Family Matters - The Shared Implications of Genetic Information

Scenarios
Refer to Section 2

The implications of a genetic disorder potentially involve a number of family members who relate to each other in various ways. Because genetic disorders are family health problems, a diagnosis in one member has implications for other family members. By the same token, ownership of information should be considered in the family context.

Issues:

- A genetic consultation or test may elicit information about not only the individual but also other blood relatives **A,C**
- Recording a patient's information in a pedigree or medical record may also be a record of the health status of relatives **A**
- Just as there are ethical responsibilities for the doctor concerning privacy and confidentiality of the patient so there are obligations to prevent harm or avoid jeopardising the health of others **A**
- The patient too has responsibilities and duties. Ethically speaking, patients need to appreciate that the communal context of their medical interaction may involve them in considering not only what will contribute to their own health and well-being but also their responsibility towards their relatives **A,C**

Responsibilities of health professionals:

- Work with families to balance the rights and needs of other family members with the privacy rights and needs of the individual regarding the information generated from genetic testing **A,C, I**
- Undertake testing of minors, in general only when there is a health benefit. If benefits will not accrue until adulthood, genetic testing should be deferred until the individual is able to make an informed decision **D,J,K**
- Ensure genetic testing is undertaken only for medical or health benefit **G**

Predictive/Presymptomatic Testing

Scenarios
Refer to Section 2

Issues:

- Pre and post test information and counselling **A,B,C**
- Consent **A,B,C**
- Confidentiality **A,B,C**
- Obligations to family members **E**
- Third party access (eg insurance companies, employers) to information **B,E**

Responsibilities of health professionals:

- Acknowledge and inform that detection of a mutation is not necessarily predictive of severity of the disorder, age of onset or likelihood of disease **A,E**
- Work with individuals and families undertaking testing to ensure that they are aware of the purpose and implications of genetic testing **A,E**
- Ensure procedures and policies are in place to prevent disclosure of the information generated from genetic testing to third parties without consent **B**
- Use consent forms to ensure that access to tissue, DNA or results of research is limited to those who have been designated by the individual tested or their guardian **I**

Reproductive Choices/Prenatal Testing

Issues:

- Pre and post test information and counselling **F**
- Consent **F**
- The choice of reproductive options alters the probability that an individual (eg fetus) or potential individual will be born or conceived **F**
- The value of a life, especially the life of a person with a disability **F**

Genetic Testing Procedures and Practices with Specific Ethical Issues

Scenarios
Refer to Section 2

Responsibilities of health professionals:

- Acknowledge and inform that the technology may not detect all possible genetic abnormalities J
- Acknowledge and inform that detection of a genetic mutation or chromosomal abnormality may not provide all the information about the quality of life of that fetus or person or the severity of a particular disorder F
- Provide prenatal diagnostic testing only where there is a medical or health benefit G
- Acknowledge and inform that the ability to detect a genetic mutation may not be sufficient reason for prenatal testing G
Recognise cultural influences in perception of disability, and work with families to ensure genetic testing is undertaken only for disorders which are detrimental to health G

Storage of DNA

Other medical services store blood or tissue but not in the same context as storage of DNA

Issues:

- Ownership of DNA - guardianship
- Ownership of the information/result I
- Special aspects of consent - DNA consent forms I

Responsibilities of health professionals:

- Store DNA in good faith but provide no guarantees of its future viability for testing I,J
- Use stored DNA for genetic testing or research only with permission of the individual, guardian or next of kin I
- Acknowledge the contribution of families who have enabled research on stored DNA samples I
- Inform families of the progress of research into their stored DNA I
- Destroy tissue or DNA taken for genetic testing after testing is concluded unless specific consent has been obtained for its storage I

Screening for Carrier Mutation Status

Scenarios
Refer to Section 2

This refers to carriers of a genetic mutation which confers no health risk to the individual but a possible risk to their offspring

Issues:

- Information
Consent
Confidentiality
Obligations to family members

J
J
J
J

Responsibilities of health professionals:

- See under family matters (page 16)
Acknowledge and inform that testing may not detect all mutations

J,K

Newborn Screening

Newborn screening tests for hypothyroidism, phenylketonuria, cystic fibrosis and galactosaemia are part of the normal care of newborn babies. Early diagnosis means that treatment can be started quickly before the baby becomes sick.

After testing the dried blood sample is stored in the laboratory indefinitely. The stored sample may be used for research (without identifying the baby). Rarely it may be used at a later time to provide new medical information for the benefit of the family. No further tests which could identify a particular disorder in a particular baby will be performed on the stored blood sample without written consent from the parent/guardian.

Issues:

- Consent
Autonomy (including privacy)

K

Responsibilities of health professionals:

- Ensure supplies of pamphlets are available to provide every parent with information about the Newborn Screening Program
Inform parents about newborn screening tests including limitations
Ensure as far as possible that newborn screening will be undertaken only with the consent of the parent or guardian
Where genetic testing reveals genetic carrier status, ensure that parents or guardians are fully informed of the meaning of that carrier status
Inform parents that dried samples are stored indefinitely after testing and inform them about the implications of storage and that they may dissent

K
J,K

Genetic Registers

Issues, responsibilities and limitations:

Ensure that data are collected, held and released in accordance with the Guidelines for the Use of Genetic Registers in Medical Research - Report to the National Health and Medical Research Council by the Medical Research Ethics Committee of the NH&MRC.

References

1. WHO, Summary statement on ethical issues in medical genetics. Geneva, February 1995.
2. Genethics - the Ethics of Engineering Life: Suzuki D, Knustson P. Allen & Unwin, Sydney 1989.
3. The Privacy Implications of Genetic Testing, Information Paper Number Five, Privacy Commissioner, September 1996.
4. WHO Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetics Services, 1995.

Genetics Service Locations

CLINICAL AND COUNSELLING

CAMPERDOWN

Department of
Molecular and Clinical
Genetics, Royal Prince
Alfred Hospital
CAMPERDOWN 2050
Tel: 02 9515 5080
Fax: 02 9515 7595

LIVERPOOL

Liverpool Hospital
Elizabeth Drive
LIVERPOOL 2170
Tel: 02 9828 4665
Fax: 02 9828 4650

RANDWICK

Department of Medical
Genetics, Sydney
Children's Hospital
RANDWICK 2031
Tel: 02 9382 1708
Fax: 02 9382 1711

WESTMEAD

Department of Genetics
The New Children's
Hospital
WESTMEAD 2145
Tel: 02 9845 3273
Fax: 02 9845 3204

NEWCASTLE

Hunter Genetics
Cnr Turton & Tinonee
Sts, WARATAH 2298
Tel: 02 4985 3100
Fax: 02 4985 3105

PENRITH

Nepean Hospital
PENRITH 2750
Tel: 02 4724 2000
Fax: 02 4724 3206

GENETIC COUNSELLING

Services in conjunction
with visiting clinical
genetics services:

ST LEONARDS

Fetal Medicine Unit
Royal North Shore
Hospital
ST LEONARDS 2065
Tel: 02 9926 7280
Fax: 02 9906 1872

BATHURST

Community Health
Centre
158 William Street
BATHURST 2795
Tel: 02 6331 5533
Fax: 02 6332 2039

BROKEN HILL

Community Health
Centre
BROKEN HILL 2880
Tel: 08 8088 5800
Fax: 08 8088 2926

CESSNOCK

Community Health
Centre
View Street
CESSNOCK 2325
Tel: 02 4990 4444
Fax: 02 4990 4228

COFFS HARBOUR

Community Health
Centre, Cnr High and
Boambee Streets
COFFS HARBOUR
2450
Tel: 02 6659 9722
Fax: 02 6659 9703

GOSFORD

"Birralee" Cnr Pacific
Highway & Kinarra Ave
WYOMING 2250
Tel: 02 4320 4534
Fax: 02 4320 4566

GOULBURN

Child Development Unit
Cnr Goldsmith &
Faithful Sts
GOULBURN 2580
Tel: 02 4827 3950
Fax: 02 4827 3958

LISMORE

37 Oliver Avenue
GOONELLABAH
2480
Tel: 02 6625 0111
Fax: 02 6625 0102

MUDGEE/DUBBO

Mudgee Community
Health Centre
MUDGEE 2850
Tel: 02 6372 6455

MUSWELLBROOK

Community Health
Centre
Brentwood Street
MUSWELLBROOK
2332
Tel: 02 6543 1777
Fax: 02 6542 5008

PORT MACQUARIE

Hastings Community
Health Centre
Morton Street
PORT MACQUARIE
2444
Tel: 02 6583 3944
Fax: 02 6584 2459

TAMWORTH

Community Health
Centre
180 Peel Street
TAMWORTH 2340
Tel: 02 6766 2555
Fax: 02 6766 3967

TAREE/FORSTER

Community Health
Centre
22 York Street
TAREE 2430
Tel: 02 6551 1315
Fax: 02 6554 8874

WAGGA WAGGA

Wagga Base Hospital
WAGGA WAGGA
2650
Tel: 02 6938 6393
Fax: 02 6921 5632

WOLLONGONG

PO Box 1798
WOLLONGONG 2500
Tel: 02 4222 5216
Fax: 02 4222 5313

PRENATAL DIAGNOSIS & COUNSELLING

Specialised services:

CAMPERDOWN

Fetal Medicine Unit
King George V Hospital
CAMPERDOWN
2050
Tel: 02 9515 8258
Fax: 02 9515 6579

LIVERPOOL

Fetal Medicine Unit
Liverpool Hospital
Elizabeth Drive
LIVERPOOL 2170
Tel: 02 9828 5679
Fax: 02 9828 5672

NEWCASTLE

Prenatal Diagnosis Unit
John Hunter Hospital
NEWCASTLE 2310
Tel: 02 4921 4694
Fax: 02 4921 4394

RANDWICK

Centre for Fetal
Medicine, Royal
Hospital for Women
RANDWICK 2031
Tel: 02 9382 6099
Fax: 02 9382 6098

PENRITH

Fetal Medicine Unit
Nepean Hospital
PENRITH 2750
Tel: 02 4724 3163
Fax: 02 4724 3206

ST LEONARDS

Fetal Medicine Unit
Royal North Shore
Hospital
ST LEONARDS 2065
Tel: 02 9926 7280
Fax: 02 9906 1872

WESTMEAD

Fetal Medicine Unit
Westmead Centre
WESTMEAD 2145
Tel: 02 9845 6802
Fax: 02 9845 7793

CANCER GENETICS

Specialised services:

LIVERPOOL

Liverpool Hospital
Elizabeth Drive
LIVERPOOL 2170
Tel: 02 9828 4665
Fax: 02 9828 4650

RANDWICK

Hereditary Cancer Clinic
Prince of Wales Hospital
RANDWICK 2031
Tel: 02 9382 2577
Fax: 02 9382 2588

WESTMEAD

Familial Cancer Services
Westmead Hospital
WESTMEAD 2145
Tel: 02 9845 5079
Fax: 02 9687 2311

NEWCASTLE

Hunter Genetics
Cnr Turton & Tinonee
Sts
WARATAH 2298
Tel: 02 4985 3100
Fax: 02 4985 3105

FURTHER INFORMATION

On services in other
areas and newly
developed services:

NSW Genetic Education
Program
PO Box 317,
ST LEONARDS 2065
Tel: 02 9926 7324
Fax: 02 9906 7529