Genetic Testing

Summary This policy sets out NSW Department of Health requirements for testing for genetic disorders and particularly addresses counselling issues and laboratory requirements associated with genetic testing.

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Audience Clinical

Secretary, NSW Health

Policy Directive

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http://www.health.nsw.gov.au/policies/

Genetic Testing

Document Number	PD2007_066
Publication date	08-Aug-2007
Functional Sub group	Clinical/ Patient Services - Medical Treatment
Summary	This policy sets out NSW Department of Health requirements for testing for genetic disorders and particularly addresses counselling issues and laboratory requirements associated with genetic testing.
Replaces Doc. No.	Genetic Disorders (Guidelines for Testing of) [GL2005_012]
Author Branch	NSW Kids and Families
Branch contact	NSW Kids & Families 9391 9503
Applies to	Area Health Services/Chief Executive Governed Statutory Health Corporation, Affiliated Health Organisations, Affiliated Health Organisations - Declared, Public Hospitals
Audience	Clinical
Distributed to	Public Health System, Divisions of General Practice, NSW Ambulance Service, Ministry of Health, Public Health Units, Public Hospitals, Tertiary Education Institutes
Review date	08-Mar-2013
Policy Manual	Not applicable
File No.	06/3554
Status	Active

Director-General

This Policy Directive may be varied, withdrawn or replaced at any time. Compliance with this directive is **mandatory** for NSW Health and is a condition of subsidy for public health organisations.

GENETIC TESTING
including DNA diagnostic testing, DNA testing for mutation carriers and DNA predictive and presymptomatic testing

Guidelines for Testing for Genetic Disorders (Circular 97/48) (GL2005_012) has been replaced by two policy directives:

1. Genetic Testing – including DNA Diagnostic Testing, DNA Testing for mutation carriers and DNA Predictive and Presymptomatic Testing
2. Prenatal Testing - including prenatal screening for Down syndrome and other chromosomal abnormalities

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GENETIC TESTING

including DNA diagnostic testing, DNA testing for mutation carriers and DNA predictive and presymptomatic testing

This policy sets out NSW Department of Health requirements for testing for genetic disorders and particularly addresses counselling issues and laboratory requirements associated with genetic testing.

Genetic tests and procedures are available for individuals at high risk for certain genetic disorders and birth defects. Testing may benefit individuals and families in a number of ways but it may also create dilemmas which need sensitive management. Counselling is an essential element of genetic testing. Each test has distinct advantages, disadvantages and limitations and should only be used after the individual being tested has given full consideration to these issues. All testing should be carried out with the informed consent of the person being tested. Health professionals and potential test users need to become familiar with the context in which the tests are used.

See also:

- Prenatal testing - including prenatal screening for Down syndrome and other chromosomal abnormalities - PD2007_067

- Guidelines for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders with predictive implications for other family members and which are likely to reduce normal life expectancy – (PD2005_303) http://www.health.nsw.gov.au/policies/PD/2005/PD2005_303.html

Professor Debora Picone AM
Director-General

1 General Information for testing for all genetic disorders

1.1 Professional experience

It is important that health professionals involved with the use of genetic tests and procedures have adequate knowledge and experience to achieve a high standard of service. Health professionals need to be aware of their own professional limitations and of the availability of others with specific expertise. It will sometimes be necessary to transfer responsibility to, or consult with clinical geneticists, cancer geneticists, fetal medicine specialists, obstetricians trained in prenatal diagnosis procedures, genetic counsellors or other appropriate specialists. (See Appendix 1 for Genetics Services contact details)

1.2 Duty to inform

The outcome of genetic testing can have a significant impact not only on the individual being tested but also on other members of their families. Testing must only be undertaken when the individual has been fully informed about the purpose of the test or the procedure and the possible implications of the results.

1.3 Consent

The person being tested must be legally competent to give consent; must consent freely without coercion by professional staff, family members, employers, insurers or others; and must be adequately informed about all relevant issues including available future options. The person may withdraw consent at any time. (See 2.2 and Appendix 3 for template consent forms)

1.4 Educational resources

A variety of resources is available to assist with patient education (See Appendix 2 for details).

1.5 Pre-test counselling

Testing should be accompanied by pre and post test counselling carried out by a health professional, knowledgeable about:

- the genetic disorder being tested
- genetic risk assessment and pre-test counselling
- the features or limitations of the laboratory test
- interpretation of results and post-test counselling
- implications of positive and negative results, and
- options available on the outcome of testing.

The way the health professional gives information should help a patient understand the testing process and purpose. The health professional should:

- communicate information and opinions in a form that the patient can understand.
- counsel without coercion; the patient is free to accept or reject the advice or the test.
- allow the patient sufficient time to make a decision, reflect on opinions, ask more questions and consult with the family, within the time constraints of the test.

- encourage the patients to make their own decisions.

1.6 Post-test counselling

Careful consideration should be given to the way results are conveyed. The health professional should take this opportunity to explain again the implications of the result. (See also Section 2.1)

1.6.1 Normal result:

Where the sensitivity of a test is less than 100%, a low risk result will not indicate the absence of a genetic disorder. It is therefore important that health professionals ensure that people are fully informed about their residual risk.

1.6.2 Abnormal result:

Notification of an abnormal result may precipitate a crisis and the person may for some time be unable to absorb any information. Appropriate pre-test counselling will help to reduce post-test anxiety. Post-test counselling must be offered and follow up support may require several consultations. Counselling should be sensitive to the nature of decisions to be taken, should respect individual decisions and allow time to reach decisions. Appropriate follow-up when an abnormality is detected may require referral to genetic counselling services, other professional services or support networks.

When an abnormality is detected women should be offered appropriate follow-up eg. referral to genetic counselling, family doctor and support networks such as the Association of Genetic Support of Australasia (AGSA).

1.7 Individuals and families from culturally and linguistically diverse backgrounds

Professional interpreter services should be used. The interpreter should not be a member of the family.

1.8 NSW Birth Defects Register

All abnormal results identified by prenatal testing and postnatal testing in the first year of life should be notified to the NSW Birth Defects Register of the NSW Health Department. For further information see http://www.health.nsw.gov.au/policies/PD/2005/PD2005_217.html

1.9 Quality assurance

Quality assurance should be undertaken to achieve optimum results and quality care. (See Section 2.3 and 2.4 for further details)

1.10 Exception to pre-test counselling requirements

Pre-test counselling requirements are not usually applicable to certain routine haematology, biochemistry, biochemical genetic tests, although testing may lead to diagnosis of a genetic condition. Information should be made available prior to

newborn screening and other population screening tests. Counselling should be offered if a result is abnormal.

2 Additional information for DNA diagnostic testing, DNA testing for mutation carriers and DNA predictive and presymptomatic testing

2.1 Clinical and counselling issues in DNA predictive testing

In addition to the general information for testing for all genetic disorders outlined in section 1, the following apply specifically to counselling about DNA predictive testing:

- An abnormal result will indicate the presence of a particular mutation, but the presence of a mutation may not necessarily define the presence or severity of disease.
- Implications for other members of the family including information which changes the risk of other family members who have not requested testing.
- Implications for future reproductive options.
- Availability of treatment.
- Clinical examination by an experienced specialist prior to a test result is encouraged, as knowledge of a normal recent examination in the event of an abnormal DNA test result will be reassuring. If signs of the disorder are present, appropriate further assistance can be obtained.

See also:
Guidelines for predictive and diagnostic DNA testing for serious adult onset neurogenetic disorders with predictive implications for other family members and which are likely to reduce normal life expectancy – (PD2005_303)
http://www.health.nsw.gov.au/policies/PD/2005/PD2005_303.html

2.2 Consent

Different types of genetic testing raise specific issues that need to be discussed as part of the consent process. Template consent forms (Appendix 3) provide direction on particular considerations to be addressed.

- Request Form for Specialised Molecular Genetic/DNA Testing for Genetic Conditions
- Consent Form for Specialised/DNA Diagnostic Testing/Storage
- Consent Form for Collection, Testing and Storage of Human Tissue for Research
- Consent Form for Analysis of Genes Associated with Cancer
- Consent Form for Pre-symptomatic, Predictive and Diagnostic DNA Testing for Serious Adult Onset Neurogenetic Disorders with Predictive Implications for other Family Members

2.3 Collection and transport of specimens

- Specimens should be collected under optimum conditions including type of specimen tube, conditions for sample storage during transport, etc.
- DNA predictive testing optimally requires 2 samples from separate blood draws at separate times, with each time recorded on the tube.

- Specimen tubes are to be labelled with the full name and date of birth of the person being tested. The person being tested should sign the specimen tube at the time of collection.
- A copy of the consent form should be forwarded to the testing laboratory with the specimen.
- Patient's suburb and postcode should be included on the test request form.
- The specimen must be accompanied by a signed referral form that specifies the test(s) to be performed.
- The transport of specimens is to occur at times agreed to by the testing laboratory.
- The time frame for receiving results should be estimated with advice from the testing laboratory.

2.4 Quality assurance

All laboratories providing human diagnostic test results (including both diagnostic and research laboratories) must comply with relevant requirements including

- Therapeutic Goods Act of 1989, its regulations and subsequent amendments, particularly with regard to IVDs
- NATA/RCPA

All laboratories should participate in an appropriate quality assurance program (where available) and perform sufficient numbers of tests relevant to the area of investigation in order to maintain reliability and expertise.

Effective communication between the clinician and the testing laboratory regarding requirements is essential to achieving optimum specimen quality.

Appendix 1

General Clinical Genetics and Genetic Counselling Services

Metropolitan Centres

Camperdown	Royal Prince Alfred Hospital, Department of Molecular and Clinical Genetics, Missenden Road, Camperdown NSW 2050 Ph: (02) 9515 5080 Fax: (02) 9550 5389
Kogarah	St George Hospital, Kogarah NSW 2217 Ph: (02) 9113 3635 Fax: (02) 9113 3694
Liverpool	Liverpool Health Services, Clinical Genetics Department, Locked Bag 7103, Liverpool BC 1871 Ph: (02) 9828 4665 Fax: (02) 9828 4650
Newcastle	Newcastle Western Suburbs Hospital, Hunter Genetics, PO Box 84, Waratah NSW 2298 Ph: (02) 4985 3100 Fax: (02) 4985 3105
Penrith	Nepean Hospital Clinical Genetics Department, Penrith NSW 2750 Ph: (02) 4734 3362 Fax: (02) 4734 2561
Randwick	The Sydney Children's Hospital Department of Medical Genetics, High St, Randwick NSW 2031 Ph: (02) 9382 1704 Fax: (02) 9382 1711
St Leonards	Royal North Shore Hospital St Leonards NSW 2065 Ph: (02) 9926 6478 Fax: (02) 9926 7880
Westmead	The Children's Hospital Department of Clinical Genetics, Westmead NSW 2145 Ph: (02) 9845 3273 Fax: (02) 9845 3204

Regional Centres

Bathurst	Community Health Centre PO Box 1479 Bathurst NSW 2795 Ph: (02) 6339 5677 Fax: (02) 6339 5655
Broken Hill	Greater Western Area Health Service Community Health Centre, PO Box 457, Broken Hill NSW 2880 Ph: (02) 8080 1554 Fax: (02) 8080 1611
Coffs Harbour	Primary Health Service Coffs Harbour Health Campus Locked Mail Bag 812, Cnr High & Boambee Sts, Coffs Harbour NSW 2450 Ph: (02) 6656 7200 Fax: (02) 6656 7203
Forster	Forster Community Health Centre Breeze Pde, Forster NSW 2428 Ph: (02) 6555 6822 Fax: (02) 6554 8874
Gosford	Child And Family Health Gateway Centre, PO Box 361, Gosford NSW 2250 Ph: (02) 4328 7994 Fax: (02) 4328 7925
Goulburn	CIFTS, Locked Bag 15, Goulburn NSW 2580, Ph: (02) 4827 3950, Fax: (02) 4827 3958
Kempsey	C/- North Coast Area Health Service Community Health Centre, Morton Street, Port Macquarie NSW 2444 Ph: (02) 6588 2882 Fax: (02) 6588 2800
Mudgee	Macquarie Area Health Service PO Box 29, Mudgee NSW 2850 Ph: (02) 6378 6236 Fax: (02) 6372 7341
Muswellbrook	Community Health Centre Brentwood Street, Muswellbrook NSW 2333 Ph: (02) 6542 2050 Fax: (02) 6542 2005
North Coast	Lismore Base Hospital PO Box 419, Lismore NSW 2480 Ph: (02) 66250 111 Fax: (02) 66250 102
Port Macquarie	North Coast Area Health Service Community Health Centre, Morton Street, Port Macquarie NSW 2444 Ph: (02) 6588 2882 Fax: (02) 6588 2800
Tamworth	Community Health Centre 180 Peel Street,Tamworth NSW 2340 Ph: (02) 6767 8100 Fax: (02) 6766 3967
Taree	Community Health Centre 22 York Street, Taree, NSW 2430 Ph: (02) 6592 9703 Fax: (02) 6592 9607
Wagga Wagga	Wagga Wagga Base Hospital, Cnr Edward and Docker Sts, Wagga Wagga NSW 2650 Ph: (02) 6938 6666 Fax: (02) 6921 5632

Familial Cancer Services

Camperdown	Royal Prince Alfred Hospital, Department of Molecular and Clinical Genetics, Missenden Rd, Camperdown NSW 2050 Ph: (02) 9515 5080 Fax: (02) 9550 5389
Darlinghurst	St Vincent's Hospital, Family Cancer Clinic, Victoria Rd, Darlinghurst NSW 2011 Ph: (02) 8382 3395 Fax: (02) 8382 3386
Kogarah	St George Hospital, Hereditary Cancer Clinic, Cancer Care Centre, Gray St, Kogarah, NSW 2217 Ph: (02) 9350 3815 Fax: (02) 9350 3958
Westmead	Westmead Hospital, Familial Cancer Service, Department of Medicine, Westmead NSW 2145 Ph: (02) 9845 6947 Fax: (02) 9687 2331
Newcastle	Hunter Family Cancer Service, PO Box 84, Waratah NSW 2298 Ph: (02) 4985 3132 Fax: (02) 4985 3133
Penrith	Nepean Hospital, Clinical Genetics Department, Level 5 South Block, PO Box 63, Penrith NSW 2750 Tel: (02) 4734 3362 Fax: (02) 4734 2567
Randwick	Prince of Wales Hospital, Hereditary Cancer Clinic, High St, Randwick NSW 2031 Ph: (02) 9382 2551 Fax: (02) 9382 2588
St Leonards	Royal North Shore Hospital, Family Cancer Service, Level 2, Vindin House, St Leonards NSW 2065 Ph: (02) 9926 5665

Fetal Medicine Services in Public Hospitals Associated with Clinical Genetics Services

Camperdown	Royal Prince Alfred Hospital, Department of Molecular and Clinical Genetic, Building 65, Level 6 Missenden Road, Camperdown NSW 2050 Ph: (02) 9515 5080, Fax: (02) 9550 5389
Kogarah	St George Hospital, Women and Children's Health Gray Street, Kogarah NSW 2217 Ph: (02) 9350 3635,Fax: (02) 9350 3694
Liverpool	Liverpool Hospital, Fetal Medicine Unit, Locked Bag 7103 Liverpool BC NSW 1871 Ph: (02) 9828 5631, Fax: (02) 9828 5570
Newcastle	John Hunter Hospital, Maternal and Fetal Medicine, Locked Bag 1, Hunter Region Mail Centre Newcastle, NSW 2310 Ph: (02) 4921 4694, Fax: (02) 4921 3133
Penrith	Nepean Hospital, Perinatal Ultrasound, Level 3 South Block, Derby Street Penrith NSW 2751 Ph: (02) 4734 2578, Fax: (02) 4737 3206
Randwick	Royal Hospital for Women, Maternal/Fetal Medicine, Barker Street, Randwick, NSW 2031 Ph: (02) 9382 6098, Fax: (02) 9382 6706
St Leonards	Royal North Shore Hospital, Fetal Medicine Unit, Pacific Highway, St Leonards NSW 2065 Ph: (02) 9926 6478, Fax: (02) 9926 7880
Westmead	The Children's Hospital, Department of Clinical Genetics, Locked Bag 4001, Westmead NSW 2145 Ph: (02) 9845 3273, Fax: (02) 9845 3204

Genetics Education Services

Centre for Genetics Education	PO Box 317, St Leonards NSW 1590 Ph: (02) 9926 7324, Tax: (02) 9906 7529 Web: http://www.genetics.com.au

Association for Genetic Support of Australasia (AGSA)

AGSA	66 Albion Street, SURRY HILLS NSW 2010 Ph: (02) 9211 1462, Fax: (02) 9211 8077 Email: agsa@ozemail.com.au Web: http://www.agsa-geneticsupport.org.au

Medications in pregnancy and lactation service (NSW)

Mothersafe	Medications in Pregnancy and Lactation Service, Royal Hospital for Women High St, Randwick, NSW 2031 Ph: (02) 9382 6539 or 1800 647 848

Birth Defects Register (NSW)

NSW Birth Defects Register	Centre for Epidemiology and Research, NSW Health Department Locked Mail Bag 961, North Sydney NSW 2061 Ph: (02) 9424 5829 Fax: (02) 9391 9232

Genetics of Learning Disability Service (GOLD)

GOLD	Hunter Genetics, PO Box 84, WARATAH NSW 2298 Ph: (02) 4985 3131, Fax: (02) 4985 3133

Appendix 2

Resources

Centre for Genetics Education
PO Box 317
ST LEONARDS NSW 1590

Tel: 02 9926 7324
Fax: 02 9906 7529
http://www.genetics.com.au

AGSA
Association of Genetic Support of Australasia Inc.
66 Albion Street
SURRY HILLS NSW 2010

Tel: 02 9211 1462
Fax: 02 9211 8077
Email: agsa@ozemail.com.au
Web: http://www.agsa-geneticsupport.org.au

Appendix 3

Template consent forms

- Request Form for Specialised Molecular Genetic/DNA Testing for Genetic Conditions
- Consent Form for Specialised/DNA Diagnostic Testing/Storage
- Consent Form for Collection, Testing and Storage of Human Tissue for Research
- Consent Form for analysis of Genes Associated with Cancer
- Consent Form for Pre-symptomatic, Predictive and Diagnostic DNA Testing for Serious Adult Onset Neurogenetic Disorders with Predictive Implications for other Family Members

Request Form for Specialised Molecular Genetic/DNA Testing for Genetic Conditions

- Must be used for **non-Medical Benefits Schedule items**
- Before testing is commenced, the laboratory may require the following details (see ***Guidelines for Specialised DNA Testing for Genetic Disorders*** http://www.health.nsw.gov.au/health-public-affairs/publications/gentest/)

Send by courier/express post to:

Send samples at room temperature
Same day OR overnight

Patient ID MRN

Last name ……

First name ……

Address ……

…… Postcode ……

Date of birth ……../…… /…… Sex M F
(dd/mm/yyyy)

Sample Date Drawn ……../……./……..
(dd/mm/yyyy)

Blood

☐ EDTA …….. mL (room temp)
☐ Lithium heparin…….. mL (room temp)

Prenatal

☐ amniotic fluid …….. mL (room temp)
☐ cultured amniocytes xT25 Flask(s) (room temp)
☐ CVS sample ……. mg ☐ on ice
☐ cleaned
☐ uncleaned

Other

☐ DNA ……..µg
Other, specify:

Test requested

PLEASE ATTACH FAMILY/PEDIGREE INFORMATION

Purpose of test

☐ Confirm clinical diagnosis
☐ Predictive/presymptomatic testing
☐ Carrier Status
☐ Prenatal Diagnosis - complete box below
☐ Determine feasibility of prenatal Dx
☐ Family study (no report for this individual)
☐ For research (no report for this individual)
☐ Bank DNA until further notice
☐ Other……..

Pregnancy Information (if applicable)

Is this individual or the partner of this individual currently pregnant

L.M.P. (dd/mm/yyyy) ……

Amnio (dd/mm/yyyy) ……

CVS (dd/mm/yyyy) ……

Family Information

Have samples from this family been sent to a DNA lab before? ☐ Yes ☐ No

If Yes, specify ……

Date of birth or age ……..

Ethnic background ……

Genetic Counselling

Has the individual been offered counselling consistent with ***Specialised/DNA Testing for Genetic Disorders***? http://www.health.nsw.gov.au/health-public-affairs/publications/gentest/

☐ Yes ☐ No ☐ Refused

Consent to Testing

Has a ***Consent Form for Specialised/DNA Testing*** been completed?

☐ Yes ☐ No

Consent to payment

☐ **Public patient, or**
☐ **Privately referred non-inpatient**
Payment to be made by Area Health Service by arrangement

Authorised by……

☐ **Private patient** - Payment to be made by patient
Consent to payment……

Send Account to:

Name……

Address……

……Postcode……..

Test requested by:

Name……Initials……

Address……

…… Postcode……..

Telephone No……

Signature……Date……

Specialty/Appointment……

Copy of report to:

Name……Initials……

Address……

……Postcode……

Telephone No……

Consent Form for Specialised/DNA Diagnostic Testing/Storage

This form has been designed to ensure that your consent is on an informed basis. Please read and consider each section.

Genetic File No	MRN

Patient

Surname	Given Name(s)
Address	
	Postcode
Date of Birth	Telephone

Parent or Guardian
(Patient under age for or unable to consent)

Surname	Given Name(s)
Address	
	Postcode
Date of Birth	Telephone

PROVISION OF INFORMATION TO PATIENT — To be completed by Health Professional

I, _____ **have informed this patient/guardian as detailed**
Insert name of Health Professional and designation

below including the nature, likely results, and material risks of DNA diagnostic testing.

Interpreter present Yes/No

Signature of Interpreter	Signature of Health Professional	Date

PATIENT CONSENT — To be completed by Patient

_____ **and I have discussed the consequences and procedures**
Insert name of Health Professional

involved in testing and storage of my tissue/blood/DNA. I have been told that:

- Testing may reveal non-paternity or non-maternity of a presumed natural parent
- Testing may not be informative for some families or family members
- Tissue/blood/DNA will be stored in good faith but may not remain in a suitable state for testing
- The collection of samples of blood/muscle/skin/_____from me/ _____
  will be used for (tick where applicable):

❑ **direct testing**

❑ **testing in family studies** (indirect testing)

❑ storage of **cell lines** from the sample for _____
insert period of time

❑ **storage** of the tissue/blood/**DNA** for _____
insert period of time

Explanation of terms
1 direct testing: testing of the gene for the disorder to determine whether a mutation is present.
2 **indirect testing (family studies)**: the tracking through a family of a mutation in a gene using 'markers' to identify the mutation.
3 **cell lines**: cells from blood or other tissues kept alive in the laboratory.
4 **DNA (deoxyribonucleic acid)** The chemical compound which the genes are made of.

- The information gained from testing may be used to assist the health care of other family members Yes ❑ No ❑
  - ❑ other family members
  - ❑ only the following individual(s)
- I have been advised to inform other adult family members who may be at risk
- I request that the sample be stored and retested if testing is inconclusive and future testing may be more informative
- I understand the potential benefits and adverse consequences involved in testing and storage of this sample
- I have had the opportunity to ask questions and am satisfied with the explanation and the answers to my questions
- I understand that consent may be withdrawn

I request and consent to the above

Signature of Patient/Guardian	Print name of Patient	Date

Consent Form for Collection, Testing and Storage of Human Tissue for Research

This form has been designed to ensure that your consent is on an informed basis. Please read and consider each section.

Genetic File No	MRN

Patient

Surname	Given Name(s)
Address	
	Postcode
Date of Birth	Telephone

Parent or Guardian
(Patient under age for or unable to consent)

Surname	Given Name(s)
Address	
	Postcode
Date of Birth	Telephone

PROVISION OF INFORMATION TO PATIENT — To be completed by Health Professional

I, _____ have informed this patient as detailed below
Insert name of Health Professional and designation

including the nature, likely results, and material risks of testing and storage of tissue/blood/DNA.

Interpreter present Yes/No

Signature of Interpreter	Signature of Health Professional	Date

PATIENT CONSENT — To be completed by Patient

_____ **and I have discussed the consequences and procedures**
Insert name of Health Professional

involved in testing and storage of my tissue/blood/DNA. I have been told that:

- ☐ The tissue/blood/DNA will be used in a research study entitled _____
- ☐ The study has been approved by the Institutional Ethics Committee of _____
- ☐ My tissue/blood/DNA (*cross out two)
  - ❖ will be destroyed at the completion of the project
  - ❖ will be stored for _____ years after completion of the project
  - ❖ may be stored indefinitely
- ☐ I will not necessarily receive a report on the outcome of the project
- ☐ I or my attending doctor will be advised if the project produces information which could be of value to me or my family
- ☐ Testing may reveal non-paternity or non-maternity of a presumed natural parent
- ☐ If tissue/blood/DNA is stored it may not remain in a suitable state for testing

DNA is an abbreviation for deoxyribonucleic acid, the name of the chemical compound of which genes and chromosomes are made

I have had the opportunity to ask questions and am satisfied with the explanation and the answers to my questions.

I understand that I may withdraw my consent.

After testing has been completed:

- ☐ **I consent to my de-identified DNA sample being used for future Institutional Ethics Committee approved research**

OR

- ☐ **My DNA sample may not be used for research without my written consent**

I request and consent to the above

Signature of Patient/Guardian	Print name of Patient	Date

(Name of Hospital)

Consent Form for analysis of Genes Associated with Cancer

This form has been designed to ensure that your consent is on an informed basis. Please read and consider each section.

Title	Family Names		MRN		
Given Name			VMO		
Address	Street		DOB	Sex	HIS
Suburb		Postcode	Admission Date		

PROVISION OF INFORMATION TO PATIENT **To be completed by Health Professional**

I, _____ have informed this patient
Insert name of Medical Practitioner/Health Professional and designation

as detailed below including the nature, likely results, and risks associated with gene testing.

_____ _____ _____

Signature of Medical Practitioner/Health Professional | Signature of Interpreter (if present) | Date

PATIENT CONSENT **To be completed by Patient/Guardian**

_____ **and I have discussed diagnostic testing for the**
Insert name of Health Professional

analysis of genes associated with cancer. He/she has told me that:

TESTING

- The collection of blood/…… will be used for testing of genes involved in:
  (tick the appropriate box)

☐ hereditary breast/ovarian cancer

☐ hereditary bowel cancer

☐ hereditary cancer predisposition (specify)

……

- The sample will be stored by the laboratory according to regulations.
- The sample will not be used for any purpose other than that agreed upon in this consent.
- Testing is voluntary and it is possible to withdraw from the testing process at any stage.

RESULTS

A Mutation Screen-when a gene change <u>has not</u> been found in any other family member

- A **<u>positive</u>** test result means that I carry a gene change (mutation) that gives me an increased risk for cancer. Each of my children have a 50% chance of inheriting the same gene change.
- A **<u>negative</u>** result is uninformative. This may be because
  - We have not been able to find a gene change using current technology or
  - It is possible that changes in other genes may be responsible for the increased risk of cancer in the family.
  - A negative result **<u>does not</u>** exclude an inherited predisposition in the family.
- Results of **<u>unknown significance</u>** - Sometimes a gene change is found and we are not sure whether it has caused the increased risk of cancer in the family. This is because the exact effect of this change on the gene is, as yet unknown.
- Other relevant information:

  ……

  ……

  Further testing may be performed in the future as our knowledge of cancer genetics improves.

B. Predictive Test-when a gene change has already been found in another family member

- A **positive** test result means that I carry the gene change that causes an increased risk of cancer in my family. Each of my children have a 50% chance of inheriting the same gene change.
- A **negative** result means that I have **not** inherited the gene change that has caused an increased risk of cancer in my family. As I do not carry this gene change, I cannot pass it on to my children.
- Other relevant information……

  ……

  ……

The test result:

- cannot predict whether a cancer will occur.
- cannot predict the age of onset or type of cancer that may develop.
- of one individual can change the estimation of risk for other family members.
- may affect the ability to obtain some types of insurance.
- may reveal non-maternity or non-paternity of a presumed parent.

CONFIDENTIALITY

- The test result will be held by this centre and will be known by those involved in the testing process.
- My test result will be given to me first in person. Other arrangements please specify -

  ……

  ……
- In the event of my death, the test results may be made known to:

  Name:……Relationship……Contact details……..

  ……

  .

  Name:……Relationship……Contact details……..

  ……

  .
- The fact that I have had a genetic test will not be revealed to any other person or organisation without my written consent except in situations where disclosure is legally required.
- My test result may be revealed to my Doctor(s) Yes ☐ No ☐

  specify ……
- The information gained from the testing may be used to assist the health care of other family members Yes ☐ No ☐
- Other relevant information

  ……

AFTER TESTING IS COMPLETED:

☐ I consent to my de-identified DNA sample being used for future ethics approved research

☐ I do not consent to my DNA sample being used for research without my written consent

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I request and consent to the test described above.

I understand the potential benefits, potential consequences and limitations involved in testing and the storage of this sample. I have had an opportunity to ask questions and I am satisfied with the explanations and answers to my questions. I understand that genetic counselling will be available for myself and my family.

Signature of person being tested	Print name of person being tested	Date

or

Signature of guardian	Print name of guardian	Date
Signature of guardian	Print name of guardian	Date

Explanation of terms used in this consent form

- **Genes associated with cancer:** Specific genes in which changes (mutations) are associated with an increased risk of cancer.
- A **gene test** involves analysis of one or more of those genes to determine whether a mutation is present
- **Cancer predisposition gene mutation:** Changed DNA code which gives rise to an increased risk of certain cancers
- **DNA** (Deoxyribonucleic acid): The chemical compound of which the genes are made

Consent Form for Pre-symptomatic, Predictive and Diagnostic DNA Testing for Serious Adult Onset Neurogenetic Disorders with Predictive Implications for other Family Members

This form has been designed to ensure that your consent is on an informed basis. Please read and consider each section.

(Name of Hospital)

Title Family Names		MRN		
Given Name		VMO		
Address Street		DOB	Sex	HIS
Suburb	Postcode	Admission Date		

PROVISION OF INFORMATION TO PATIENT **To be completed by Health Professional**

I, _____ have informed this patient as detailed below
Insert name of Health Professional and designation

the nature, likely results, and risks associated with gene testing for _____
name of disorder

Interpreter present Yes/No

_____ _____ _____
Signature of Interpreter Signature of Health Professional Date

PATIENT CONSENT **To be completed by Patient/Guardian**

_____ **and I have discussed predictive testing**
Insert name of Health Professional

testing for the analysis of the gene fault (mutation) for _____
name of disorder

He/she has told me that:

- The collection of blood will be used to examine my DNA and tested for the gene involved in _____
name of disorder
- A **positive test result** indicates that I have inherited a faulty gene (mutation). This means that I am at high risk of developing/will develop _____ and my children and siblings have a _____ % chance
name of disorder
percentage
- A positive test result cannot accurately predict the age of onset of the disorder.
- A **negative test result** means that I have not inherited the faulty gene (mutation). I will not develop _____ and cannot pass the faulty gene involved on to my children
name of disorder
- An intermediate result means that I may or may not develop _____
name of disorder
- In some instances this may have implications for my siblings and children and their descendents

- Test results of one individual can change the estimation of risk for other family members and I have been advised to inform other adult family members who may be at risk.
- The test result may impact on obtaining some types of insurance or employment.
- Testing may reveal non-paternity or non-maternity of a presumed natural parent
- Genetic counselling will be available for myself and other family members during the testing process and after the test result has been given.

I have been told about storage of the test results and the DNA sample. I understand the following:

- The test result will be held by this centre and will only be known by those involved in the testing process.
- My own test result, the fact that I have had a test, and my DNA sample will not be revealed or made available to any other person or organisation outside of the testing process, except with my written consent (as detailed below), or in situations where disclosure is required by law.
- The test results will be given to me first.
- The DNA sample will remain the property of the laboratory. It will be stored in good faith, but its suitability for future use cannot be guaranteed. It will be disposed of at a time determined by standard laboratory practices or regulatory requirements.
- My identified DNA sample will not be used for any other purpose except in accordance with my written consent (as detailed below).

I request and consent to the test described above.

I understand the potential benefits of testing and storing this sample and I accept the risks involved. I have had the chance to ask questions and am satisfied with the explanations and the answers to my questions.

I understand that I may withdraw my consent for this test to be processed.

I consent to my test results being revealed at any time to the following people:

- ❑ Any family member
- ❑ Only to the following individuals (specify) _____
- ❑ My doctor(s) *(specify)* _____
- ❑ No other individual
- ❑ In the event of my death **test results** may be made known to: _____

After testing has been completed:

- ❑ **I consent to my de-identified DNA sample being used for future Institutional Ethics Committee approved research**

OR

- ❑ **My DNA sample may not be used for research without my written consent**

Signature of Patient/t/Guardian	Print name of Patient	Date

Explanation of terms used in this consent form

- A **gene test** involves analysis of one or more of those genes to determine whether a mutation is present
- **Mutation:** Change in the normal DNA code which may cause or increase risk for a condition
- **DNA** (Deoxyribonucleic acid): The chemical compound of which the genes are made