

Specialised Testing for

Genetic Disorders

**Part 2 - Guidelines for Specialised
Testing for Genetic Disorders**

Better Health Good Health Care

NSW  HEALTH

NSW HEALTH DEPARTMENT

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Produced by:

The NSW Genetic Service Advisory Committee
Statewide Services Development Branch
NSW Health Department
Locked Mail Bag 961
North Sydney, NSW 2059
Ph: (02) 9391 9520

For further copies please contact:

Better Health Centre – Publications Warehouse
Locked Mail Bag 5003
Gladesville, NSW 2111
Ph: (02) 9816 0452
Fax: (02) 9816 0492

A full copy of this report and others in this series can be downloaded from the NSW HealthWeb site:
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1 Genetic counselling and risk assessment

Genetic tests and procedures are available for individuals at high risk for certain genetic disorders. Each test has distinct advantages, disadvantages and limitations and should only be used after the person being tested has given full consideration to the relevant issues. Genetic disorders by their nature run in families, so that a diagnosis in one member has implications for other family members. Testing may benefit individuals and families in a number of ways but it may also create dilemmas which need sensitive management, making genetic counselling an essential element of genetic testing (see 1.11 for exemptions).

It is important for health professionals requesting tests and potential test users to become familiar with the context in which the tests are used. All testing should be carried out with the informed consent of the person being tested.

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 genetics professionals, fetal medicine specialists, obstetricians trained in prenatal diagnosis procedures, genetic counsellors or other appropriate specialists.

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 should 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 Educational resources

A variety of resources is available to assist with patient education (Section 5).

1.4 Pre-test genetic counselling

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

- the genetic disorder being tested, including where appropriate, clinical features, age of onset, pattern of inheritance and availability of treatment
- genetic risk assessment and pre-test genetic counselling
- the features or limitations of the laboratory test
- details of the test, testing process, length of time to results

- interpretation of results and post-test genetic 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

1.5 Individuals and families from non-English speaking background

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

1.6 Consent

Consent must be given by a legally competent person. Consent must be given 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.

Sample consent forms for specialised genetic testing/storage and prenatal diagnosis are included in Section 2.

1.7 Confidentiality

Those being tested should be reassured that the results, and the fact that they have undergone testing, will be confidential between themselves and the testing team, and will not be revealed to others without specific agreement and written consent. The possibility that the information could be revealed by a family member should be mentioned.

1.8 Giving results and post-test genetic 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.

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 due to the complexity of inheritance or environmental factors.

Notification of a result may precipitate a crisis and the person may for some time be unable to absorb any information. Appropriate pre-test genetic counselling may help to reduce post-test anxiety. Post-test genetic counselling must be offered and follow up support may require several consultations. Genetic 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 other health professionals for treatment or management, genetic counselling services, family doctor or other professional services, or support networks.

1.9 NSW Birth Defects Register

All abnormal results as defined by the NSW Birth Defects Register, identified in the first year of life by prenatal or postnatal testing, should be notified to the NSW Birth Defects Register of the NSW Department of Health.

1.10 Quality assurance

Quality assurance should be undertaken to achieve optimum results and quality care.

1.11 Exception to pre-test genetic counselling requirements

Pre-test genetic counselling requirements are not applicable to certain routine tests, although testing may lead to diagnosis of a genetic condition. Information about newborn screening and other population screening tests should be made available prior to testing. Genetic counselling should be offered if a result has genetic implications.

2 Request and Consent Forms

- Request Form for Specialised/DNA Testing for Genetic Disorders
- Consent Form for Specialised/DNA Testing/Storage
- Consent Form for Collection, Storage and Testing of Human Tissue for Research
- Consent Form for Prenatal Diagnosis
- Consent Form for Diagnostic Testing/Analysis of Genes Associated with Cancer

Request Form for Specialised/DNA Testing for Genetic Disorders

<p>Send by express post to:</p> <p>Send samples at room temperature Same day OR overnight</p>	<p>Patient ID</p> <p>Last name</p> <p>First name</p> <p>Address</p> <p>Postcode</p> <p>Birthdate Sex M F</p> <p>Status <input type="checkbox"/> private patient in a private hospital/day facility</p>
<p>Test requested</p> <p>PLEASE PROVIDE FAMILY/PEDIGREE INFORMATION</p>	<p><input type="checkbox"/> public patient in hospital <input type="checkbox"/> private patient in hospital</p> <p><input type="checkbox"/> outpatient <input type="checkbox"/> privately referred outpatient</p> <p>Genetic Counselling (see below #) Has the individual been offered counselling consistent with Guidelines for Specialised/DNA Testing for Genetic Disorders?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Refused</p>
<p>Purpose of test</p> <p><input type="checkbox"/> Confirm clinical diagnosis</p> <p><input type="checkbox"/> Predictive/presymptomatic testing</p> <p><input type="checkbox"/> Carrier Status</p> <p><input type="checkbox"/> Prenatal Diagnosis</p> <p><input type="checkbox"/> Determine feasibility of parental Dx</p> <p><input type="checkbox"/> Family study (no report for this individual)</p> <p><input type="checkbox"/> For research only</p> <p><input type="checkbox"/> Bank DNA until further notice</p> <p><input type="checkbox"/> Other</p> <p>Have samples from this patient been sent to a DNA lab before?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No</p> <p><input type="checkbox"/> Don't know</p> <p>If yes, Where.....</p>	<p>Consent (see below #) Has a Consent Form for Specialised/DNA Testing been completed? <input type="checkbox"/> Yes <input type="checkbox"/> No</p> <p>Family Information Have samples from this family been sent to a DNA lab before?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No</p> <p>If Yes, specify</p> <p>Country of birth</p> <p>Ethnic background</p> <p>Is this patient the index case?</p> <p><input type="checkbox"/> Yes <input type="checkbox"/> No</p> <p>If no, specify</p> <p>Name of index case in the family</p> <p>D.O.B.</p> <p>Relationship to this patient</p>
<p>Sample Requisition Date Drawn / / (dd/mm/yyyy)</p> <p>Blood</p> <p><input type="checkbox"/> EDTA mL room temp</p> <p><input type="checkbox"/> Lithium heparin mL room temp</p> <p>Prenatal</p> <p><input type="checkbox"/> amniotic fluid mL room temp</p> <p><input type="checkbox"/> cultured amniocytes xT25 Flask(s) room temp</p> <p><input type="checkbox"/> CVS sample mg <input type="checkbox"/> on ice</p> <p><input type="checkbox"/> cleaned <input type="checkbox"/> uncleaned</p> <p><input type="checkbox"/> Other</p> <p><input type="checkbox"/> DNA mg Other (specify)</p>	<p>Pregnancy Information (if applicable) Is this individual or the partner of this individual is currently pregnant</p> <p>L.M.P. (dd/mm/yyyy)</p> <p>Amnio (dd/mm/yyyy)</p> <p>CVS (dd/mm/yyyy)</p> <p>Test requested by:</p> <p>Name Initials</p> <p>Address</p> <p>Phone</p> <p>Signature</p> <p>Copy of report to:</p> <p>Name Initials</p> <p>Address</p> <p>Phone</p>
<p>Comments</p>	

TESTING WILL ONLY COMMENCE ON RECEIPT OF COMPLETED REQUEST FORM
Consent Forms and Guidelines for Specialised/DNA Testing for Genetic Disorders are available from the laboratory



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.

Testing Centre	MRN
----------------	-----

Parent or Guardian

(Patient under age for or unable to consent)

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

Patient/Test Subject

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 DNA diagnostic testing.

Interpreter present: Yes / No

 Signature of Interpreter

 Signature of Health Professional

PATIENT CONSENT

To be completed by Patient/Test Subject

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

involved in storage and testing of my tissue/blood/DNA. The doctor has told me 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 for an indefinite time but may not remain in a suitable state for testing
- The collection of samples of blood/muscle/skin/ _____ from me/ _____ will be used for (delete where not applicable):

1. **direct testing**
2. **testing in family studies** (indirect testing)
3. storage of **cell lines** from the sample for an indefinite time
4. extraction of **DNA**
5. **storage** of the tissue/blood/DNA for an indefinite time

- The results of the test carried out on this sample of the stored material may be made known if reasonably indicated to:

- other family members
- only the following individuals(s) _____

- 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 I may withdraw my consent

I request and consent to the test described above

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.

 Signature of Test Subject/Guardian

 Print name of Test Subject

 Date



Consent Form for Collection, Storage and Testing 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.

Testing Centre	MRN
----------------	-----

Parent or Guardian

(Patient under age for or unable to consent)

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

Patient/Test Subject

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 storage and testing of tissue/blood/DNA.

Interpreter present: Yes / No

Signature of Interpreter

Signature of Health Professional

PATIENT CONSENT

To be completed by Patient/Test Subject

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

involved in storage and testing of my tissue/blood/DNA. The doctor has told me that:

Initials

- 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

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

I request and consent to the test described above

Signature of Test Subject/Guardian

Print name of Test Subject

Date



Consent Form for Prenatal Diagnosis

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

PART A of this form must be completed by all patients requesting prenatal diagnosis whether they have the procedure or not.

(Name of Hospital)

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

PART A

PROVISION OF INFORMATION TO PATIENT

To be completed by Medical Practitioner/
Genetic Counsellor

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

including the nature, likely results, and risks of prenatal diagnosis procedure.

Interpreter present: Yes / No

Signature of Interpreter

Signature of Health Professional

PATIENT CONSENT

To be completed by Patient

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

involved in prenatal diagnosis. The doctor/genetic counsellor has told me that:

- The procedure involves a risk of causing pregnancy complications including miscarriage
- There is a possibility that the procedure may not be successful
- There is a possibility that laboratory testing of specimens obtained may fail
- The laboratory analysis may not accurately reflect the fetal status and in some instances this can lead to incorrect interpretation of the results
- A normal result from this test means that, within the diagnostic limitations of the test, the fetus is not affected for the disorder being tested. It does not exclude other abnormalities
- No assurance has been given that any particular doctor will perform this procedure

I understand that undergoing the procedure carries risks.

I have had the opportunity to ask questions.

I am satisfied with the explanation and the answers to my questions

I understand that I may withdraw my consent.

PART B

I request and consent to _____ and accept the risks involved in the procedure.
Insert name of Procedure

Signature of Patient

Print name of Patient

Date



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.

(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

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

Interpreter present: Yes / No

Signature of Interpreter

Signature of Health Professional

PATIENT CONSENT To be completed by Patient/Test Subject/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:

- The collection of blood/ _____ will be used to examine my DNA and tested for one or more of the genes involved in predisposition to:
 - Hereditary breast/ovarian cancer
 - Hereditary Non Polyposis Colorectal Cancer (HNPCC)
 - Familial Adenomatous Polyposis (FAP)
 - Other hereditary cancers (specify) _____
- A **positive test result** indicates that I have inherited a gene fault (mutation). This means that I have an increased risk of cancer and my child has a 50% chance of inheriting the mutation from me.
- If a mutation **has** already been found in an affected family member, a **negative test result** means that I have not inherited the gene change and cannot pass it on to my children
- If a mutation **has not** been found in an affected family member a **negative result is not informative**. That means the test result does not tell us whether or not a gene fault (mutation) is present, because the test will not detect all possible mutations.
- The test result cannot accurately predict the age of onset or type of cancer that may develop.
- Test results of one individual can change the estimation of risk for other family members
- The test result may affect the ability to obtain some types of insurance.

- 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, and the reasons for which they may be disclosed or used. 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: _____

I consent to the details of the mutation causing cancer in the family being made available to laboratories which have been asked to test other family members, provided that to do so would not reveal any details of my identity or personal test result without 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

Signature of Patient/Test Subject/Guardian

Print name of Patient/Test Subject

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
- **Mutation:** Change in the normal DNA code which may cause disease
- **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

3 Collection and transport of specimens

3.1 Test request form and pedigree

A completed **test request form** (Section 2) should accompany all specimens, along with a family history presented as a **genetic pedigree**. It should be noted that the request form requires an indication that genetic counselling has occurred.

The laboratory may not commence testing until all the requirements have been met.

3.2 Specimens

- Specimens should be collected under optimum conditions
- Specimen tubes are to be labelled with two unique identifiers, eg the full name and date of birth or medical record no. of the person being tested. The person being tested or parent or guardian should sign the specimen tube at the time of collection or identify the specimen in some other way.
- Similar security procedures should be followed in anonymous testing, eg Tay Sachs testing.
- DNA testing optimally may require 2 samples, each taken by separate venepunctures or a single venepuncture separated into 2 samples. The second sample is used to confirm results.
- Sample requirements vary according to the type of DNA testing - refer to specific guidelines or contact the laboratory.
- Samples should be sent by express post as per IATA 650 regulations at room temperature, except between December and the end of February. During this period it may be necessary to cool the sample to 4°C. **DO NOT FREEZE.**
- 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.
- Requirements may vary for specific disorders. Further information is available from the testing laboratory.