

## Prenatal Testing/Screening for Down Syndrome & Other Chromosomal Abnormalities

**Summary** The policy is directed to clinical and care providers involved in prenatal care. It provides direction on access to and provision of prenatal testing including prenatal screening, so women are informed about screening options and are appropriately directed to services. In recent years an increasing number of non-invasive biochemical screening tests and ultrasound techniques have been developed which can significantly increase the identification of pregnancies at risk for Down syndrome and other chromosomal abnormalities in women of all ages. Each test has advantages, disadvantages and limitations. Offers of screening need to be accompanied by sufficient information and support.

**Document type** Policy Directive

**Document number** PD2007\_067

**Publication date** 08 August 2007

**Author branch** Agency for Clinical Innovation

**Branch contact** 02 9464 4711

**Review date** 15 December 2017

**Policy manual** Not applicable

**File number** 06/2925

**Previous reference** N/A

**Status** Review

**Functional group** Clinical/Patient Services - Maternity, Medical Treatment

**Applies to** Area Health Services/Chief Executive Governed Statutory Health Corporation, Affiliated Health Organisations, Affiliated Health Organisations - Declared, Public Hospitals

**Distributed to** Public Health System, Divisions of General Practice, NSW Ambulance Service, Ministry of Health, Public Health Units, Public Hospitals, Private Hospitals and Day Procedure Centres, Tertiary Education Institutes

**Audience** Clinical;nursing;maternity staff;fetal medicine staff

## Prenatal Testing/Screening for Down Syndrome & Other Chromosomal Abnormalities

**Document Number** PD2007\_067

**Publication date** 08-Aug-2007

**Functional Sub group** Clinical/ Patient Services - Maternity  
Clinical/ Patient Services - Medical Treatment

**Summary** The policy is directed to clinical and care providers involved in prenatal care. It provides direction on access to and provision of prenatal testing including prenatal screening, so women are informed about screening options and are appropriately directed to services. In recent years an increasing number of non-invasive biochemical screening tests and ultrasound techniques have been developed which can significantly increase the identification of pregnancies at risk for Down syndrome and other chromosomal abnormalities in women of all ages. Each test has advantages, disadvantages and limitations. Offers of screening need to be accompanied by sufficient information and support.

**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, nursing, maternity staff, fetal medicine staff

**Distributed to** Public Health System, Divisions of General Practice, NSW Ambulance Service, Ministry of Health, Public Health Units, Public Hospitals, Private Hospitals and Day Procedure Centres, Tertiary Education Institutes

**Review date** 08-Mar-2013

**Policy Manual** Not applicable

**File No.** 06/2925

**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.

---

## **PRENATAL TESTING** **including prenatal screening for Down syndrome** **and other chromosomal abnormalities**

Guidelines for Testing for Genetic Disorders (Circular 97/48) (GL2005\_012) has been replaced by two policy directives:

1. Prenatal Testing - including prenatal screening for Down syndrome and other chromosomal abnormalities
2. Genetic Testing – including DNA diagnostic testing, DNA testing for mutation carriers and DNA predictive and presymptomatic testing

---

## **PRENATAL TESTING** **including prenatal screening for Down syndrome** **and other chromosomal abnormalities**

This policy directive addresses:

**Prenatal screening tests** – these tests may identify a baby as being at an increased risk of having a particular problem and include:

- First trimester screening for women presenting between 10 to 14 weeks of pregnancy using a combination of maternal age, ultrasound nuchal translucency measurement (NTS) and serum screening tests (free  $\beta$ -hCG and PAPP-A).
- Second trimester screening for women presenting between 15 to 18 weeks of pregnancy, using maternal age, maternal serum screening (free  $\beta$ hCG, AFP and unconjugated estriol).
- Ultrasound

**Prenatal diagnostic tests** – these tests may be used following an increased risk result on prenatal screening or independently:

- Ultrasound
- Chorionic villus sampling
- Amniocentesis
- Fetal blood sampling

This policy is directed to NSW Health clinical and care providers involved in prenatal care. It provides direction on access to and provision of prenatal screening and diagnostic tests so pregnant women are informed about screening options and are appropriately directed to services.

See also:

Genetic Testing – including DNA diagnostic testing, DNA testing for mutation carriers and DNA predictive and presymptomatic testing  
PD2007\_066

Professor Debora Picone AM  
**Director-General**

## Table of Contents

1	Introduction	
2	Down syndrome and other chromosomal abnormalities	
3	Consent and provision of information on prenatal screening and testing	
4	<u>Prenatal screening</u> for Down syndrome and other chromosomal abnormalities	
4.1	Women at increased risk	
4.2	Women not in the increased risk category	
5	<u>Prenatal diagnosis</u> using amniocentesis, chorionic villus sampling and fetal blood sampling to determine the fetal karyotype	
6	Serum alpha fetoprotein testing for neural tube defects	
7	Prenatal screening and diagnosis by fetal imaging	
8	Professional experience, audit, quality assurance and monitoring	
	General information for testing for all genetic disorders.....	Appendix 1
	Resources .....	Appendix 2
	Consent Form .....	Appendix 3
	Clinical and genetic counselling service locations .....	Appendix 4

## **1. Introduction**

In recent years, an increasing number of non-invasive biochemical screening tests and ultrasound techniques have been developed which can significantly increase the identification of pregnancies at risk for Down syndrome and other chromosomal abnormalities in women of all ages.

The use of prenatal screening tests has added to the complexity of prenatal care. These screening tests give a risk indication only and are not definitive tests. Use and interpretation are dependent on a number of factors including accurate gestational age, the stage of pregnancy, maternal age, and in the case of ultrasound, operator expertise. They are associated with varying levels of false positive and false negative results, depending on different combinations of tests offered. Women indicated to be at high risk on screening tests should be offered follow-up definitive testing by amniocentesis or chorionic villus sampling.

Each screening test has advantages, disadvantages and limitations. Offers of screening need to be accompanied by sufficient information and counselling, with professional interpreter services if necessary, to help women choose screening on an informed basis. This includes accurate information about the health and development issues for children with Down syndrome and the potential ramifications for women entering into the screening process.

NSW Health's policy of offering diagnostic testing by chorionic villus sampling and amniocentesis to women at increased risk for chromosome errors through family history or advanced maternal age (35 years and older at estimated date of confinement) remains unchanged (see Section 5).

## **2. Down syndrome and other chromosomal abnormalities**

In NSW, the incidence of Down syndrome in pregnancy is approximately 2.5 per 1000. In 2004, the NSW Birth Defects Register<sup>1</sup> reported 98 births and 132 terminations of pregnancy for Down syndrome, ie a total of 230. The number of livebirths and stillbirths for all chromosomal abnormalities (including Down syndrome) in 2004 was 202 with 243 reported terminations, ie a total of 445.

The risk of having a baby with Down syndrome increases with advancing maternal age. Since 1990 confinements to women 35 years and over have risen from 10.4% to 20.7% in 2005, increasing the potential for more Down syndrome affected pregnancies in this age group.

*Title: Prenatal Testing*

**Table 1 Risk by age of Down syndrome and other chromosomal abnormalities**

maternal age at delivery	* chance of having a live-born baby with Down syndrome	** chance of having a live-born baby with a chromosomal abnormality
20-24 years	1 in 1411	1 in 506
25 years	1 in 1383	1 in 476
26 years	1 in 1187	1 in 476
27 years	1 in 1235	1 in 455
28 years	1 in 1147	1 in 435
29 years	1 in 1002	1 in 417
30 years	1 in 959	1 in 385
31 years	1 in 837	1 in 385
32 years	1 in 695	1 in 323
33 years	1 in 589	1 in 286
34 years	1 in 430	1 in 244
35 years	1 in 338	1 in 179
36 years	1 in 259	1 in 149
37 years	1 in 201	1 in 124
38 years	1 in 162	1 in 105
39 years	1 in 113	1 in 81
40 years	1 in 84	1 in 64
41 years	1 in 69	1 in 49
42 years	1 in 52	1 in 39
43 years	1 in 37	1 in 31
44 years	1 in 38	1 in 24
45 years	1 in 32	1 in 19

\* Morris JK, Mutton DE, and Alberman E (2002). Revised estimates of the maternal age specific live birth prevalence of Down's syndrome. *J Med Screen*, 9, 2-6.

\*\* Hook EB (1981) Rates of chromosomal abnormalities. *Obs Gyn* 58 282-285

Although the risk to an individual pregnancy increases with age, about half of the occurrences of Down syndrome and other chromosomal abnormalities are in babies of women under the age of 35 who comprise a much greater proportion (80%) of women giving birth.

**Table 2 Chromosomal abnormalities among birth defect cases by maternal age, NSW 2002**

Maternal age	No.	%
<25	32	7.4
25-29	42	9.7
30-34	79	18.2
35-39	87	20.0
40+	60	13.8
Not Stated	135	31.0
Total	435	100

Source: NSW Birth Defects Register, Centre for Epidemiology and Research, NSW Department of Health<sup>1</sup>

Note: Includes terminations of pregnancy, livebirths and stillbirths

### 3. Consent and provision of information on prenatal screening and testing

It is recognised that not all women will want to use prenatal screening or diagnostic tests.

Any test undertaken should be consented to on the basis of provision of full relevant information. It is important that women/couples considering prenatal screening or diagnostic testing make an informed choice appropriate to them and free from coercion. They should have access to written information on the tests, their implications and their risks (see resources listed in Appendix 2). The relative advantages, disadvantages and limitations of the available tests should be discussed with the woman or couple prior to testing as part of general counselling and support. Discussion should include:

- information on the conditions being screened for
- the timing of testing
- the importance of accurate gestation dates
- the risk to the woman of having an affected offspring
- test performance information based on the audit figures of the local providers of prenatal screening services
- that a screening test alone does not identify any birth defect but indicates a risk only
- interpretation of both **increased and low risk** screening results. Women must be informed about their residual risk.
- the impact of an **increased risk** result and the options available for definitive diagnosis, such as amniocentesis.
- the impact on a woman and her family of a false negative or false positive result
- the time frame for receiving results and making further decisions if necessary
- long term implications for the person and their family of having an affected baby and the health and development issues for children with the condition.

Women at increased risk should have access to consultation with an individual or centre able to provide both counselling and prenatal diagnosis procedures. (See Appendix 4 for contact details). Full information about the risks of amniocentesis and chorionic villus sampling should be provided (See Section 5).

Appendix 1 provides general information for testing for all genetic disorders including:

- Professional experience
- Duty to inform
- Educational resources (listed in Appendix 2)
- Pre-test counselling
- Consent
- Individuals and families from culturally and linguistically diverse backgrounds
- Post-test counselling

## 4. **Prenatal screening for Down syndrome and other chromosomal abnormalities**

The following policy has been developed to inform health care providers about the tests available and their effectiveness, so that women can be helped to make informed decisions and be given appropriate care.

The policy is based on the data currently available on the performance of screening tests<sup>2,3,4,5,6</sup>. However, prenatal screening tests continue to evolve and providers need to ensure their information is up-to-date.

### 4.1 **Women at increased risk**

Women considered at increased risk for chromosome errors, ie those with an indicative previous history or who will be 35 years or older at estimated date of confinement; should be offered prenatal screening or diagnostic testing as an integral part of management. Offers of screening or testing should be accompanied by sufficient information and counselling to help women choose and consent to testing on an informed basis and to be aware of the potential ramifications of entering into the screening process (see Section 3).

#### **First trimester screening tests for women presenting between 10 to 14 weeks of pregnancy:**

- First trimester screening using combination of maternal age, ultrasound nuchal translucency measurement (NTS) and serum screening tests (free  $\beta$ -hCG and PAPP-A).
- A detection rate of up to 90% with up to 5% false positive results should be achieved<sup>7</sup>.
- Women should be sure of their gestational age based on a known LMP and regular pre-conception menstrual cycle. In the event of any uncertainty about gestational age a pre-test ultrasound examination is optimal.
- Only accredited operators - Fetal Medicine Foundation (FMF) or Nuchal Translucency – Ultrasound, Education and Monitoring Project (NT-UEMP) should perform nuchal translucency (NT) screening (see Section 8).
- The first trimester combined screening risk provided to women is calculated automatically 'at the gestation at screening', not 'at term', since up to a third of pregnancies affected by Down syndrome spontaneously abort between 10-40 weeks gestation.



*Title: Prenatal Testing*

## **Second trimester screening tests for women presenting between 15 to 18 weeks of pregnancy:**

- Second trimester triple screening using maternal age, maternal serum screening (free  $\beta$ hCG, AFP and unconjugated estriol).
- A detection rate of at least 60% with up to 5% false positive results should be achieved. About 40% of babies with Down syndrome will therefore be missed using this test.

## **Management of high-risk screening results**

All women receiving a high-risk test result should be provided with adequate post-test counselling. The level of counselling support needed may vary with the type of result and the resources of the referring practitioner to deal with the issues surrounding an abnormal result. Abnormal screening test results should be dealt with as urgent clinical problems requiring early referral to an individual or centre able to provide both counselling and diagnostic procedures for prenatal diagnosis (see Appendix 3 for a list of services). Full information about the risks of amniocentesis and chorionic villus sampling should be provided (see Section 5). Referral to family doctor, the Association for Genetic Support of Australasia (AGSA) and support networks should be included.

## **Collection and transport of maternal serum specimens for 1<sup>st</sup> and 2<sup>nd</sup> trimester screening**

A minimum of 3 mls of serum is required. The blood sample can be drawn at a collecting pathology service and sent immediately to the laboratory. It may be refrigerated for up to 24 hours or frozen if longer.

Laboratories offering maternal serum testing should provide request forms detailing the information that must be provided for accurate interpretation of the test result. This will include at least:

- Gestational age by dating scan
- Patient's age – it is most important that this be clearly and correctly stated on the referral form
- Patient's weight
- Patient's height
- Whether the patient is an insulin dependent diabetic
- Previous history of Down syndrome and other chromosomal abnormalities
- Patient's suburb and postcode
- A signed referral form that specifies the test(s) to be performed

*Title: Prenatal Testing*

## 4.2 Women not in the increased risk category (above)

Women not in the increased risk category, ie no previous history and under the age of 35 years at estimated date of confinement should be informed about screening tests and given appropriate risk information by their doctor or midwife (see Section 2, Table 1 - Risk by age of Down syndrome and other chromosomal abnormalities risks) and informed of the availability and potential cost of screening tests.

For further information contact:

**Information on prenatal screening for all women is available from:**

Centre for Genetics Education  
PO Box 317  
ST LEONARDS NSW 1590

Tel: 02 9926 7324

Fax: 02 9906 7529

Website: <http://www.genetics.com.au>

**Prenatal Testing - Special tests for your baby during pregnancy**

<http://www.genetics.com.au/pdf/pubs/prenatal.pdf>

## A note on second trimester ultrasound at 18 – 20 weeks gestation

For all women, ultrasound assessment of markers of chromosomal abnormality in the second trimester is not recommended as a primary screening test for Down syndrome (see section 7).

## 5. **Prenatal diagnosis using amniocentesis, chorionic villus sampling and fetal blood sampling to determine the fetal karyotype.**

These are invasive tests which pose a risk of miscarriage (of less than 1%) in addition to the “background risk” of miscarriage due to natural causes. It is important to discuss this. After the age of 35 the risk of Down syndrome rises rapidly so the risk of a Down syndrome affected pregnancy becomes greater than the risk of miscarriage associated with amniocentesis and chorionic villus sampling (CVS) procedures.

**Prenatal diagnostic services should be made available to:**

- Women of 35 years and over at date of confinement

---

*Title: Prenatal Testing*

- Women who have a screening test for Down syndrome which suggests increased risk
- Women who have had a child with a neural tube defect
- Women determined to be at high risk for a neural tube defect in light of first or second trimester screening
- Other women who have a high risk of a fetus with a genetic or chromosomal disorder which may be detectable by prenatal diagnosis.

## **Additional clinical and counselling issues**

It is recommended that where practical patients are counselled face to face at least one day before the procedure with the opportunity to clarify information and options. This may not be possible in some situations. Telephone counselling through the local genetics service may provide an alternative. Counselling should address:

- criteria for access to procedures and choice between CVS and amniocentesis
- a clear and simple explanation of the probability of an affected fetus
- stage of pregnancy when the procedure should be undertaken
- explanation of the process of the procedure
  - amniocentesis
  - trans-abdominal CVS
  - trans-vaginal CVS
  - fetal blood sampling
- the risk of pregnancy complications including a risk of miscarriage (of less than 1%) in addition to the “background risk” of miscarriage due to natural causes.
- waiting time for results and how they will be conveyed
- the possibility that the procedure may not be successful
- the possibility that laboratory testing of specimens obtained may fail
- the laboratory analysis may not accurately reflect the fetal status and in rare cases this can lead to incorrect interpretation of results
- options to be considered if the result is abnormal
- acknowledgement of the individual nature of decisions about continuing or terminating the pregnancy
- methods of termination of pregnancy
- a normal result on amniocentesis, chorionic villus sampling or fetal blood sampling means that within the diagnostic limitations of the test, the fetus is not affected for the disorder being tested. It does not exclude the possibility that the child may have birth defects and/or mental retardation due to other causes. It is important that health professionals ensure that people are fully informed about their residual risk.
- the implications of a multiple pregnancy
- the possibility of unexpected results which are difficult to interpret eg a chromosome marker
- costs involved and how they are to be met

*Title: Prenatal Testing*

**NB Abnormal results should be reported to the NSW Birth Defects Register (see Appendix 1-1.8).**

## Consent

A template consent form is attached at Appendix 3.

## Collection and transport of specimens:

- Specimens should be collected under optimum conditions including type of specimen tube and conditions for sample storage during transport.
- Specimen tubes are to be labelled with the **full name** and **date of birth** of the person being tested. It would be preferable if the patient could sign the tube containing their 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.
- **DNA testing** - A copy of the completed consent form should be forwarded to the testing laboratory with the specimen.

## Quality Assurance

- Effective communication between the clinician and the testing laboratory regarding requirements is essential to achieving optimum specimen quality.
- Laboratories should be NATA accredited or should participate in an appropriate quality assurance program performing sufficient numbers of tests relevant to the area of investigation.
- Where prenatal testing is to be performed on DNA from chorionic villus tissue on cells, it would be advisable to also test the sample and parental DNA samples to rule out maternal contamination.

## 6. Serum alpha fetoprotein testing for neural tube defects

### Neural Tube Defects

Neural tube defects (NTD) include anencephaly, spina bifida and encephalocele and occur in about 1 in 800 births. The incidence is increased among women who:

- have a neural tube defect
- have had a previous child or pregnancy with NTD
- have a close family history of NTD
- have insulin-dependent diabetes

---

*Title: Prenatal Testing*

- are taking specific anticonvulsant medications
- are obese

These women should be offered genetic counselling.

### **Note on Folate**

Approximately 70% of cases of neural tube defects can be prevented by increased folate intake at least one month before and continuing for the first three months of pregnancy. See Appendix 2 for further information on pamphlets.

### **Serum Alpha Fetoprotein (AFP) Testing**

AFP testing is a voluntary and optional prenatal test which gives a risk assessment for **neural tube defects. It is not a diagnostic test.** An elevated level of serum AFP signifies an increased risk for neural tube defects. Elevated AFP is also associated with other causes such as multiple gestation or threatened miscarriage and indicates the need for follow up procedures such as ultrasound or an amniocentesis.

### **Timing**

The optimal time for AFP testing is between 15 and 18 weeks of gestation.

### **Sensitivity and specificity**

The high risk cut off point is normally set at 2.5 MOM so that for every 10 pregnancies identified at high risk, one will have spina bifida.

### **Collection and transport of specimens**

A minimum of 3 mls of serum is required. The blood sample can be drawn at a collecting pathology service and sent immediately to the laboratory. It may be refrigerated for up to 24 hours or frozen if longer.

Laboratories offering serum alpha fetoprotein testing should provide request forms detailing the information that must be provided for accurate interpretation of the test result. This will include at least:

- Gestational age by dating scan
- Patient's age – it is most important that this be clearly and correctly stated on the referral form
- Patient's weight
- Patient's height
- Whether the patient is an insulin dependent diabetic
- Whether the patient is taking anticonvulsant medication
- Multiple pregnancy (if known)
- Previous history of neural tube defects
- Patient's suburb and postcode
- A signed referral form that specifies the test(s) to be performed

## **7. Prenatal screening and diagnosis by fetal imaging**

Ultrasound has become a routine part of prenatal care and may be done at any stage during the pregnancy. It may be used as either a screening or a diagnostic test.

Parents often view the ultrasound as an opportunity to bond with the fetus and may not have given consideration to the prospect of an adverse result. When an abnormality is detected, care should be taken to provide counselling and emotional support to minimise the impact of the result on the woman and her family.

It is recommended that providers of fetal imaging services develop and implement protocols for managing counselling issues associated with the detection of abnormalities. Important elements would include:

- counselling by obstetricians, ultrasound staff, referring doctor and other health professionals prior to routine ultrasound, concerning possible outcomes and management strategies.
- training of ultrasound technologists in management strategies in the event of an abnormality being detected.
- ensuring abnormal results are conveyed in a supportive, informative and timely manner to reduce unnecessary anxiety associated with the diagnosis.
- ensuring appropriate tertiary referral for women when an abnormality is detected eg. referral to genetic counselling and fetal medicine units, and follow-up by family doctor and support networks such as the Association of Genetic Support of Australasia (AGSA).

***NB Abnormal results should be reported to the NSW Birth Defects Register (see Appendix 1-1.8).***

### **Second trimester ultrasound at 18 – 20 weeks gestation**

For all women, ultrasound assessment of markers of chromosomal abnormality in the second trimester is not recommended as a primary screening test for Down syndrome. However, if women have not had the opportunity to have a prior screening test, the second trimester ultrasound may be used to indicate an increased or decreased risk for Down syndrome based on the presence or absence of markers of aneuploidy. This scan has poor sensitivity and specificity for aneuploidy.

## **8. Professional experience, audit, quality assurance and monitoring**

Users of services should be provided with accurate and current information on the numbers of pregnancies screened, the detection rate, and the screen positive rate. Ideally, providers should contribute data to a central body to facilitate pooling of data and allow for external scrutiny of results.

Women's understanding of and satisfaction with the screening methods, counselling and associated procedures ideally should be assessed so that appropriate information and education can be offered.

### **Standards for laboratories providing Down Syndrome Screening services**

Laboratories should have internal audit and external QA through UK National External Quality Assessment Schemes (UK NEQAS) as well as accreditation with the National Association of Testing Authorities (NATA)

### **Ultrasound**

All procedures should be performed by experienced operators who have appropriate training. Nuchal translucency screening should only be performed by trained operators, using a risk-assessment program that incorporates NT, crown-rump length (CRL) and maternal age. Accreditation should be either through the Fetal Medicine Foundation UK or through the Nuchal Translucency – Ultrasound, Education and Monitoring Project (NT-UEMP) administered through the Royal Australian and New Zealand College of Obstetricians and Gynaecologists.

All ultrasound providers have to be audited at least six monthly by the NT-UEMP.

### **Providers of prenatal diagnosis by amniocentesis, chorionic villus sampling or fetal blood sampling**

Amniocentesis, chorionic villus sampling and fetal blood sampling should be performed in a prenatal diagnosis service where the operator(s) have sufficient training and annual experience of the procedure to keep the complication rate as low as possible. There is evidence that fetal loss rate is multifactorial eg maternal age, gestation and operator experience.

**Providers should have an established relationship with a fetal medicine unit**, and have ready access to appropriate genetic counselling in the fields of cytogenetics, molecular genetics and biochemical genetics. This association does not necessarily imply that an individual practitioner is geographically located at a hospital containing a fetal medicine unit.

**Providers should participate in quality assurance activities** and contribute to the statistics of a fetal medicine unit with regard to sampling success rate,

---

*Title: Prenatal Testing*

proportion of abnormalities detected, fetal loss rate and other complications. They should participate in clinical audit procedures of the Royal Australian and New Zealand College of Obstetricians and Gynaecologists.

**Providers should have demonstrated adequate training and experience in prenatal diagnostic procedures.** A minimum level of experience for amniocentesis would be at least 100 procedures in training and 50 per year. A minimum for chorionic villus sampling is at least 100 procedures in training and 50 per year. Fetal blood samplings should only be performed in fetal medicine units.

**Minimum qualifications** would be FRANZCOG, FRANZCR, DDU or equivalent, with appropriate invasive prenatal diagnosis training.



## Appendix 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, 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 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.

#### 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:

*Title: Prenatal Testing*

- 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.

### **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](http://www.health.nsw.gov.au/policies/PD/2005/PD2005_217.html)

---

*Title: Prenatal Testing*

## **1.9 Quality Assurance**

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

### **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.

## Appendix 2

### Resources

#### 1. Centre for Genetics Education

PO Box 317  
ST LEONARDS NSW 1590

Tel: 02 9926 7324

Fax: 02 9906 7529

<http://www.genetics.com.au>

The following publications are available:

- Prenatal Testing: Special tests for your baby during pregnancy
- Screening Tests for Your Baby in Early Pregnancy (Consumer and Professional versions available)
- The Maternal Serum Test
- Genetic Services and counselling: Why knowing about your genes is important to your future
- Ultrasound - Obstetric care for pregnant women
- Prenatal Diagnosis and Counselling – Importance of checking your baby's health before birth
- The Importance of Your Family Health Information (Consumer and Professional versions available)
- When your baby has a problem: How to manage the weeks ahead
- Diagnosis Of Abnormality In An Unborn Baby, The Impact, Options and Afterwards
- Folate pamphlet
- Tests to protect your baby – newborn screening
- The Genetics Resource Book

#### 2. Multicultural Resources Available from Multicultural Health Communication Services

<http://www.health.nsw.gov.au/health-public-affairs/mhcs/index.html>

The following publications available in a variety of languages other than English

- Prenatal Testing - Special tests for your baby during pregnancy  
<http://www.health.nsw.gov.au/health-public-affairs/mhcs/publications/7750.html>
- Ultrasound Examination Preparation  
<http://www.health.nsw.gov.au/health-public-affairs/mhcs/publications/864.html>

*Title: Prenatal Testing*

---

- How can Genetic Counselling Help?  
<http://www.health.nsw.gov.au/health-public-affairs/mhcs/publications/3070.html>
  - Bringing up children with Down Syndrome  
<http://www.mhcs.health.nsw.gov.au/health-public-affairs/mhcs/publications/3370.html>
  - Questions Women ask about Abortion  
<http://www.health.nsw.gov.au/health-public-affairs/mhcs/publications/5915.html>
3. **The NSW Department of Health**
    - Having a baby - free to all women at their first antenatal booking appointment at a public hospital. Also available on the NSW Health website at [http://www.health.nsw.gov.au/pubs/2006/having\\_a\\_baby.html](http://www.health.nsw.gov.au/pubs/2006/having_a_baby.html)
  4. **The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG)**  
<http://www.ranzcog.edu.au>
    - Amniocentesis and Chorionic Villus Sampling (CVS) – a Guide on Prenatal Diagnostic Procedures
    - Antenatal Care and Routine Tests During Pregnancy – a Guide for Women
    - Prenatal Screening Tests for Down Syndrome and Other Conditions
    - Why Aren't All Babies Perfect?
  5. **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](mailto:agsa@ozemail.com.au)  
Web: <http://www.agsa-geneticsupport.org.au>
  6. **The Down Syndrome Association of NSW**  
PO Box 2356,  
North Parramatta NSW 1750  
Tel: 02 9683 4333  
Fax: 02 9683 4020  
<http://www.dsansw.org.au/index.html>



# Consent Form for Prenatal Diagnosis

# Appendix 3

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

(Name of Hospital)

Genetic File No.		MRN
Surname		Given Name(s)
Address		
		Postcode
Date of Birth		Telephone

## PART A

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

### 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 of the prenatal diagnosis procedure.**

- 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

Interpreter present Yes/No

\_\_\_\_\_  
Signature of Interpreter

\_\_\_\_\_  
Signature of Health Professional

\_\_\_\_\_  
Date

## PART B

### PATIENT CONSENT To be completed by Patient

\_\_\_\_\_ and I have discussed the consequences and  
Insert name of Proceduralist  
**procedures involved in prenatal diagnosis.**

- 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.

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

\_\_\_\_\_  
Signature of Patient

\_\_\_\_\_  
Print name of Patient

\_\_\_\_\_  
Date

## Appendix 4

### Fetal Medicine Services in Public Hospitals Associated with Clinical Genetics Services

Camperdown	Royal Prince Alfred Hospital, Department of Molecular and Clinical Genetics, 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

### 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

*Title: Prenatal Testing*

## 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



*Title: Prenatal Testing*

## 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

## Genetics Education Services

<b>Centre for Genetics Education</b>	PO Box 317, St Leonards NSW 1590 Ph: (02) 9926 7324, Tax: (02) 9906 7529 Web: <a href="http://www.genetics.com.au">http://www.genetics.com.au</a>
--------------------------------------	---

## Association for Genetic Support of Australasia (AGSA)

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

## 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
------	---

---

## References

---

<sup>1</sup> *New South Wales Mothers and Babies 2005*, NSW Public Health Bulletin Supplement, Vol 18, Number S-1. February 2007.

<sup>2</sup> *'Best Practice' Guidelines on antenatal screening for Down syndrome and other fetal aneuploidy* prepared by the Joint Human Genetics Society of Australasia and Royal Australian and New Zealand College of Obstetricians and Gynaecologists

<sup>3</sup> *First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS)*, Wald NJ, Rodeck C, Hackshaw AK, Walters J, Chitty L, Makinson A, Health Technology Assessment NHS R&D HTA Programme, Vol 7, No 11, 2003.

<sup>5</sup> *Nuchal Translucency Measurement in the First Trimester of Pregnancy for Screening of Trisomy 21 and other Autosomal Trisomies*, Medical Services Advisory Committee, Department of Health and Ageing May 2002

<sup>6</sup> *Joint estimation of Down syndrome risk and ascertainment rates: a meta-analysis of nine published data sets*. *Prenatal Diagnosis* 1998;18(1):9-20) Bray I, Wright DE, Davies C, Hook EB

<sup>7</sup> *Maternal serum biochemistry at 11-13+6 weeks in relation to the presence or absence of the fetal nasal bone on ultrasonography in chromosomally abnormal fetuses: an updated analysis of integrated ultrasound and biochemical screening*. *Prenatal Diagnosis* 2005; 25:977-983, Cicero S, Spencer K, Avgidou K, Faiola S, Nicolaides K.