Antenatal and newborn screening

The National Screening Unit (NSU) of the Ministry of Health is responsible for the development, implementation and management of three antenatal and newborn screening programmes:

- Universal Offer Antenatal HIV Screening Programme
- Newborn Metabolic Screening Programme
- Universal Newborn Hearing Screening and Early Intervention Programme.

The NSU is also responsible for the introduction of quality improvements to antenatal screening for Down syndrome and other conditions.

Screening for various conditions is a significant aspect of contemporary antenatal care and many of the procedures carried out during routine antenatal appointments are used as screening tools. Taking a woman’s blood pressure and reviewing a urinalysis are used to screen for pre-eclampsia, diabetes, or urinary infection. Abdominal palpation is used to screen for babies who are small for their gestational age, and pregnancies with polyhydramnios. Blood is tested to screen for hepatitis B, HIV, and rhesus antibodies.

The quality improvements to antenatal screening for Down syndrome and other conditions have been introduced to bring screening in New Zealand into line with international best practice, and to provide pregnant women and their families/whānau with information with which to make choices about the care and management of their pregnancy.

Acknowledgements

These Guidelines have been produced in consultation with a Technical Working Group. The National Screening Unit would like to thank the many individuals and groups who contributed to the development of the final version of this document.
## CONTENTS

Antenatal and newborn screening  
Acknowledgements  
List of acronyms  
Definitions  

<table>
<thead>
<tr>
<th>1</th>
<th>INTRODUCTION</th>
<th>5</th>
</tr>
</thead>
</table>

2 | BACKGROUND INFORMATION | 6 |
2.1 | Quality improvements to antenatal screening for Down syndrome and other conditions | 6 |
2.2 | Improving equity of access to antenatal screening for Down syndrome and other conditions | 7 |
2.3 | Conditions screened for by antenatal screening for Down syndrome and other conditions | 8 |
2.4 | Down syndrome  
2.4.1 | Common health issues associated with Down syndrome | 9 |
2.5 | Potential benefits and harms of antenatal screening for Down syndrome and other conditions | 9 |

3 | THE PRACTICALITIES | 11 |
3.1 | The screening pathways | 11 |
3.2 | Timing of screening tests | 13 |
3.3 | Informed consent | 14 |
3.4 | Documentation | 15 |
3.5 | Communication | 16 |
3.6 | Provision of information | 16 |
3.7 | Initial discussion | 18 |
3.8 | Offer of screening  
3.8.1 | First trimester combined screening | 21 |
3.8.2 | Second trimester maternal serum screening | 21 |
3.8.3 | Referring women for screening | 22 |
3.9 | Laboratory processes | 23 |
3.9.1 | Laboratory responsibilities | 24 |
3.9.2 | Exceptions | 25 |
3.10 | Results  
3.10.1 | Increased risk results | 28 |
3.11 | Genetic counselling and other referrals | 29 |
3.12 | Funding of diagnostic testing | 30 |
3.13 | Data collection, monitoring and reporting | 30 |

APPENDIX ONE: GLOSSARY OF TERMS  
APPENDIX TWO: RESOURCES AND CONTACTS  
APPENDIX THREE: BIBLIOGRAPHY
List of acronyms

<table>
<thead>
<tr>
<th>Acronym</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>AFP</td>
<td>Alpha-fetoprotein (a biochemical marker)</td>
</tr>
<tr>
<td>ßhCG</td>
<td>Beta-human chorionic gonadotrophin (a biochemical marker)</td>
</tr>
<tr>
<td>BPD</td>
<td>Biparietal diameter</td>
</tr>
<tr>
<td>CRL</td>
<td>Crown–rump length</td>
</tr>
<tr>
<td>CVS</td>
<td>Chorionic villus sampling</td>
</tr>
<tr>
<td>DHB</td>
<td>District Health Board</td>
</tr>
<tr>
<td>EDD</td>
<td>Estimated Date of Delivery</td>
</tr>
<tr>
<td>IVF</td>
<td>In vitro fertilisation</td>
</tr>
<tr>
<td>LMC</td>
<td>Lead Maternity Carer</td>
</tr>
<tr>
<td>LMP</td>
<td>Last Menstrual Period</td>
</tr>
<tr>
<td>NSU</td>
<td>National Screening Unit of the Ministry of Health</td>
</tr>
<tr>
<td>MoM</td>
<td>Multiple of the Median</td>
</tr>
<tr>
<td>NT</td>
<td>Nuchal translucency (an ultrasound marker)</td>
</tr>
<tr>
<td>PAPP-A</td>
<td>Pregnancy-associated plasma protein A (a biochemical marker)</td>
</tr>
<tr>
<td>µE₃</td>
<td>Unconjugated oestriol (a biochemical marker)</td>
</tr>
</tbody>
</table>

Definitions

In these Guidelines ‘maternity provider’ and ‘primary maternity services’ have the same meaning as in the Primary Maternity Services Notice 2007.

Maternity provider means an organisation or an individual that provides primary maternity services.

Primary maternity services means:
- lead maternity care
- maternity non-LMC services
- specialist medical maternity services.
1 INTRODUCTION

The purpose of these Guidelines is to identify best practice for maternity providers offering antenatal screening for Down syndrome and other conditions in New Zealand. The Guidelines are intended as a resource to inform practice and support maternity providers to improve the quality of screening.

The Guidelines promote women-centred care, and involve the co-ordination of activities of a range of maternity providers. A central concept of antenatal screening for Down syndrome and other conditions is unconditional acceptance of and support for choices made by women about screening.

Maternity providers have a contractual obligation under the Primary Maternity Services Notice 2007, issued pursuant to section 88 of the New Zealand Public Health and Disability Act 2000, to provide services within screening initiatives endorsed by the Ministry of Health, including antenatal screening for Down syndrome and other conditions.

Maternity providers are responsible for:

(a) providing information and education about screening
(b) offering referrals for screening tests
(c) communicating screening results
(d) offering specialist referrals
(e) ensuring compliance with the:
   • Privacy Act 1993 and Health Information Privacy Code 1994
   • New Zealand Public Health and Disability Act 2000
   • Code of Health and Disability Services Consumers’ Rights 1996
   • Health Act 1956
   • Health Practitioners Competence Assurance Act 2003
   • Public Records Act 2005.

These Guidelines should be read in conjunction with local District Health Board (DHB) policies and guidelines.
2 BACKGROUND INFORMATION

Antenatal screening for Down syndrome and other conditions has been available to pregnant women in New Zealand since 1968. In recent years concern has been expressed about the quality and safety of practice for antenatal screening for Down syndrome and other conditions. Mounting evidence has supported safer and higher-quality practice. In 2006, Professor Peter Stone identified that current practice for antenatal screening for Down syndrome and other conditions was "ad hoc, based mostly on ultrasound scanning or maternal age alone, and did not reflect international best practice".1

Some members of the 2007 Antenatal Down Syndrome Screening Advisory Group debated whether it was appropriate to screen for Down syndrome and other conditions at all. However, there was agreement that the status quo could not be continued when New Zealand practice was inconsistent with international understanding, technology and practice. In October 2007 Cabinet agreed to quality improvements to antenatal screening for Down syndrome and other conditions.

The quality improvements to antenatal screening for Down syndrome and other conditions are to improve the quality and safety of screening services for pregnant women in New Zealand who choose to have screening. These services will be consistent with international practice.

2.1 Quality improvements to antenatal screening for Down syndrome and other conditions

Antenatal screening for Down syndrome and other conditions is a way of assessing the likelihood that a fetus has Down syndrome or another specific condition. Screening is not diagnostic, and does not detect with certainty if a condition is present. Screening divides women into two groups: a positive result means there is an increased risk of a particular condition being present, while a negative result means there is a low risk.2 Women with an increased risk result may or may not choose to go on to diagnostic testing.

Screening for Down syndrome and other conditions involves a sequence of events referred to as the 'screening pathway'. The aim is to offer women information and choice in the care and management of their pregnancy.

Quality improvement measures for antenatal screening for Down syndrome and other conditions include:

[a] **First trimester combined screening**, which combines the results of a first trimester maternal serum test with a nuchal translucency (NT) scan result3 and other parameters such as crown–rump length, maternal age and weight, and gestation to give a single first trimester risk result. This combined result provides a significantly better risk assessment than NT scanning in isolation.

---


3Although the first trimester maternal serum screening is publicly funded, the woman is often required to make a co-payment for the NT scan component of first trimester combined screening.
(b) **Second trimester maternal serum screening**, which involves the addition of a fourth marker to the existing second trimester maternal serum screening (‘triple test’). The results of the serum tests are incorporated with other parameters such as maternal age and weight, and gestation, to give a single second trimester risk result.

(c) **Recommendations for practice**, including the discontinuation of the use of maternal age and NT scanning as screening tools in isolation.

(d) **Consumer resources**.

(e) **Education and training** for providers.

The goals of the implementation of quality improvements to antenatal screening for Down syndrome and other conditions are to:

- improve equity of access to screening
- support women to make an informed choice about whether or not to participate in screening
- improve the sensitivity and specificity rates of available screening options
- ensure maternity providers are appropriately educated about antenatal screening for Down syndrome and other conditions.

Desired outcomes of the quality improvements include:

- improved detection rates and reduced false positive rates in screening for Down syndrome and other conditions
- voluntary participation at each step of the screening pathway
- unconditional support for the choices made by women throughout their pregnancy
- the provision of accurate and non-directional information (both medical and non-medical) to support women in their decision-making
- the active involvement and support of family/whānau (if the woman wishes).

### 2.2 Improving equity of access to antenatal screening for Down syndrome and other conditions

Improving equity of access is a key goal of the quality improvements for antenatal screening for Down syndrome and other conditions. Data indicate that Māori and Pacific women are less likely to access early antenatal care than non-Māori, and that barriers to accessing aspects of antenatal care may be faced by new migrants and women from rural and lower socioeconomic environments.⁴

Lack of knowledge, transport, travel time and child care have all been identified as potential barriers to accessing screening, and women in rural areas may have difficulty accessing ultrasound and amniocentesis services.⁵

The quality improvements for antenatal screening for Down syndrome and other conditions aim to improve equity by providing two access points to screening, one in the first and one in the...

---


second trimester. The option of screening during the second trimester means screening can be offered to women who do not access maternity care early in their pregnancy.

The maternal serum screening component of first trimester combined screening is publicly funded. Women are usually required to make a co-payment for the NT scan. Second trimester maternal serum screening is fully publicly funded.

Women may only access one publicly funded screening option. However, second trimester maternal serum screening is funded for women who accepted but did not complete first trimester combined screening.

2.3 Conditions screened for by antenatal screening for Down syndrome and other conditions

Antenatal screening for Down syndrome and other conditions may indicate an increased risk for Down syndrome (Trisomy 21), Trisomy 18 (Edwards syndrome), Trisomy 13 (Patau syndrome), neural tube defects (e.g., spina bifida) and other rare metabolic or genetic disorders.

In addition, ultrasound scans undertaken as part of screening may detect some major fetal structural anomalies, such as skeletal anomalies, brain and neural tube defects, congenital heart defects, and abnormalities of the renal tract, gastrointestinal system, and abdominal wall.

2.4 Down syndrome

Down syndrome occurs in approximately 1 in 700 births (approximately 90 babies each year in New Zealand). Two-thirds of babies with Down syndrome are born to women under the age of 35, due to the higher birth rate in this age group.6

Down syndrome is caused by an extra copy of chromosome 21 inside each of the body’s cells. The chromosomes are located in the nucleus of each cell, and contain the genetic material that, in combination with environmental influences, determines a person’s individual characteristics. In Down syndrome, instead of a pair there are three copies of chromosome 21. The extra genetic material from the extra chromosome gives the characteristics of Down syndrome.

The New Zealand Down Syndrome Association advises that:

*People with Down syndrome are all unique individuals and vary in their abilities and achievements. They do have features in common, but they also closely resemble their parents and family. Many characteristics are associated with Down syndrome, but any one person will only have some of them. Thus each person is an individual, with a unique appearance, personality and set of abilities. The extent to which a child shows the physical characteristics of the syndrome is no indication of his or her abilities and achievements.*7

People with Down syndrome have varying degrees of disability. The New Zealand Disability Strategy states that:

*Disability has a lot to do with discrimination, and has a lot in common with other attitudes and behaviours such as racism and sexism that are not acceptable in our society. Disability is also closely linked to ideas about the human rights of people with impairments. Without human rights we cannot live as full human beings.*8

---

7NZDSA: www.nzdsa.org.nz, retrieved 09 September 2009
The average life expectancy of people with Down syndrome has increased with improved healthcare, better education, greater opportunities and a shift in societal attitudes during the past 20 to 30 years. Studies indicate that average life expectancy in the UK was estimated to be 9 years of age in 1929 and 12 years in 1949. Subsequent reports have shown a marked increase in life expectancy that began in the 1950s. By the year 2000 the median life expectancy for people with Down syndrome in Australia was 60 years. 9, 10

2.4.1 Common health issues associated with Down syndrome

People with Down syndrome experience varying degrees of delay in their learning and development, and may have additional health needs. Some of the health issues associated with Down syndrome include:

- hearing loss in up to 50 percent of people with Down syndrome
- congenital heart disease in up to 50 percent
- thyroid disorders, most commonly hypothyroidism, in up to 40 percent
- gastrointestinal tract congenital malformations, such as duodenal atresia and Hirschsprung’s disease
- cataracts and visual refractive errors
- childhood leukaemia in about 2 percent
- early onset Alzheimer’s disease.

2.5 Potential benefits and harms of antenatal screening for Down syndrome and other conditions

Screening poses different ethical considerations from those that arise when a person presents for medical care because they are unwell. Maternity providers have a special duty of care when referring healthy asymptomatic women for screening. All pregnant women must be given full information regarding antenatal screening for Down syndrome and other conditions, including the risks, benefits and harms of screening, so that they may make informed choices.

Antenatal screening for Down syndrome and other conditions has complex ethical and social implications, as well as technical considerations. Technical considerations involve a trade-off between the sensitivity (detection rate) and the specificity (false positive rate) of the screening tests. A valid, reliable and safe screening test is at the core of any organised population-based screening initiative. In the context of antenatal screening for Down syndrome and other conditions, it has been identified that combining ultrasound and maternal serum markers increases detection rates (improves sensitivity) and/or reduces the number of women considered to be at increased risk (improves specificity).

The potential benefits of antenatal screening for Down syndrome and other conditions include:

- access to information that may provide more choice in the care and management of a pregnancy
- a low risk result means a baby is unlikely to have Down syndrome or another condition screened for.

The potential harms of antenatal screening for Down syndrome and other conditions include that:

- an increased risk result may turn out to be a false positive result
- a low risk result may lead a woman to believe her baby will not have Down syndrome or another condition screened for, when the baby does have a condition
- an increased risk result may lead to a decision to have a diagnostic test that has an inherent risk of iatrogenic miscarriage.

While the purpose of these quality improvements is to offer women information and choice, there may be a perception that they will lead to more terminations of pregnancy and, ultimately, fewer people with Down syndrome in society. This is not the intention of the quality improvements for antenatal screening for Down syndrome and other conditions, which aim to improve current screening practice.
3 THE PRACTICALITIES

3.1 The screening pathways

**FIRST TRIMESTER COMBINED SCREENING**

- Provision of information about screening
  - Section 3.6
- Initial discussion
  - Section 3.7
- Offer of screening
  - Section 3.8
- Screening declined
- Results to maternity provider
  - Section 3.10
- First trimester combined screening
  - Blood test (2 maternal serum markers)
  - NT scan
  - Section 3.8.1
- Low risk
  - Section 3.10
- Increased risk
  - Section 3.10.1
- Offer of specialist referral
  - Section 3.10.1
- End of screening process

**SECOND TRIMESTER MATERNAL SERUM SCREENING**

- Provision of information about screening
  - Section 3.6
- Initial discussion
  - Section 3.7
- Offer of screening
  - Section 3.8
- Screening declined
- Results to maternity provider
  - Section 3.10
- Second trimester maternal serum screening
  - Blood test (4 maternal serum markers)
  - Section 3.8.2
- Low risk
  - Section 3.10
- Increased risk
  - Section 3.10.1
- Offer of specialist referral
  - Section 3.10.1
- End of screening process
The Ministry of Health recommends that all pregnant women are offered antenatal screening for Down syndrome and other conditions in either the first or second trimester of pregnancy. The exception is women who have previously been pregnant with or have had a child with a significant physical or learning disability, or have a family history of a genetic condition. These women have a different risk status. Screening may still be a good option for them, but before screening is offered they should be offered a referral for a discussion with a specialist obstetrician or geneticist to clarify their options and the appropriateness of screening.

Screening is not compulsory, so all pregnant women need to decide if they want to participate. Women who decide not to participate in screening may do so for many reasons. For example, they may perceive that they are at little or low risk, or be uncomfortable with the concept of screening. Their reasons are their own and their decision must be respected.

<table>
<thead>
<tr>
<th>First trimester combined screening to be offered to all women who present early in pregnancy</th>
<th>Second trimester maternal serum screening to be offered to all women who present later in pregnancy</th>
<th>Recommendations for practice</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Blood test that measures two maternal serum markers (PAPP-A and ßhCG) combined with NT scan results</td>
<td>• Blood test that measures four maternal serum markers (ßhCG, AFP, µE₃ and inhibin A)</td>
<td>• The discontinuation of the use of maternal age and nuchal translucency as screening tools in isolation</td>
</tr>
<tr>
<td>• Available to all women who present in the first trimester</td>
<td>• Available to women who present after the first trimester or who do not access first trimester combined screening</td>
<td></td>
</tr>
<tr>
<td>• The blood test is fully funded</td>
<td>• The blood test is fully funded</td>
<td></td>
</tr>
<tr>
<td>• Women are usually required to make a co-payment for the NT scan</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Provision of accurate and non-directive information (both medical and non-medical)

Unconditional support for decisions made by women throughout pregnancy, including the decision as to whether or not to participate in screening

Support if women want family/whānau to be actively involved
3.2 Timing of screening tests

The following diagram shows when the different antenatal screening tests for Down syndrome and other conditions may be undertaken. The timing of screening relative to the woman’s gestation may be important in relation to the choices available to her.

<table>
<thead>
<tr>
<th>First trimester combined screening</th>
<th>Second trimester maternal serum screening</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>9 weeks to 13 weeks 6 days</strong></td>
<td><strong>14 weeks to 20 weeks</strong></td>
</tr>
<tr>
<td>Blood collected 9W to 13W 6D</td>
<td>Blood collected 14W to 20W</td>
</tr>
<tr>
<td>NT 11W to 13W 6D</td>
<td>Optimal timing four maternal serum markers (ßhCG, AFP, µE3 and inhibin A) 14 - 18 weeks</td>
</tr>
<tr>
<td>Optimal timing two maternal serum markers (PAPP-A and ßhCG) 10 - 12 weeks</td>
<td>Optimal timing NT 11½ - 13½ weeks</td>
</tr>
</tbody>
</table>
3.3 Informed consent

An appropriate informed consent process is key to offering screening in a way that is socially and ethically responsible. Informed consent must be integrated throughout the screening pathway and include consent that anonymised information can be used for monitoring and reporting.

The Code of Health and Disability Services Consumers’ Rights provides that New Zealand health care consumers have a legal right to appropriate information to enable them to give informed consent. Information about the Code can be viewed on the website of the Health and Disability Commissioner (www.hdc.org.nz).

There is anecdotal evidence that some pregnant women have had antenatal screening for Down syndrome and other conditions without understanding the nature and implications of the screening. The quality improvements for antenatal screening for Down syndrome and other conditions will reduce the potential for screening to occur without women having given informed consent, by increasing knowledge and understanding among maternity providers, and by providing appropriate consumer information for women.

Maternity providers should be cognisant of the important implications for pregnant women and their families/whānau whenever any aspect of screening is discussed.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>PARTICIPATION IN SCREENING</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must establish whether the woman wishes to participate in screening or not.</td>
<td>While screening is to be routinely offered, participation in screening is not to be regarded as routine. All women should be offered the choice to participate in screening, fully informed of the decisions and options that are involved. The right to decline screening must be made clear and any such decision, including withdrawal of consent, must be respected.</td>
</tr>
<tr>
<td><strong>PRINCIPLES OF INFORMED CONSENT</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must apply the principles of informed consent.</td>
<td>The woman must have a clear understanding of the decisions she may need to make at each step in the screening pathway. Informed consent may include the use of interpreters and culturally appropriate counselling and support services.</td>
</tr>
</tbody>
</table>
### 3.4 Documentation

Written consent for antenatal screening for Down syndrome and other conditions is not required by the Code of Health and Disability Services Consumers’ Rights, if the process and consent decisions are clearly documented in the woman’s notes.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
</table>
| **DOCUMENTATION OF PROCESS AND DECISIONS** | Maternity providers should document information about:  
  - the initial discussion/s  
  - when and how the opportunity to ask questions was provided  
  - whether any further information was requested and/or issues raised, and the information provided and/or how the issues were addressed  
  - whether interpreters or other services were used  
  - whether the woman consented to or declined screening  
  - if the woman consented to screening, then that she consented to have her anonymised information included in monitoring and reporting. |
| Maternity providers must document information about the process and the woman’s decisions at each step of the screening pathway, in the woman’s notes. |  |

| **DOCUMENTATION OF ALL RESULTS** | Maternity providers must document that results have been given to the woman. |
| Maternity providers must document all results received from the laboratory in the woman’s notes. | If a woman has indicated that she only wants to receive some of the results, this must be documented and the results that were given must be clearly identified. |
|  | Maternity providers may also document details of when, where, and how results were given to the woman (and her family/whānau). |

| **DOCUMENTATION OF INCREASED RISK RESULTS** | Maternity providers must document decisions that were taken in relation to results and any follow-up actions. |
| Maternity providers must document the woman’s decisions following an increased risk result in her notes. | Where a screening result is ‘increased risk’, the woman’s notes should document that an informed consent process was followed for the offer of referral to a specialist obstetrician. This may include:  
  - that information about referral to a specialist obstetrician was given  
  - whether the offer was accepted or declined  
  - if accepted, the date the specialist obstetric referral was made, and the outcome of the referral  
  - if declined, any details about the decline  
  - other support, resources and information that were made available. |
| Maternity providers must document the consent process when offering specialist obstetric referral. |  |
3.5 Communication

The maternity provider referring the woman for screening is responsible for explaining the purpose of antenatal screening for Down syndrome and other conditions, including the potential benefits and harms, and for receiving and following up the results. Maternity providers should not allow screening to undermine the concept of pregnancy as a positive experience.

Women need to know what conditions screening might indicate as well as the implications of low risk and increased risk results.

3.6 Provision of information

Provision of information throughout the screening process is integral to informed consent. Information should be tailored to suit the needs of each woman throughout the screening process.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>APPROPRIATE PROVISION OF INFORMATION</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must provide accurate and non-directional information about antenatal screening for Down syndrome and other conditions to all pregnant women.</td>
<td>Women should have sufficient time to ask questions, consult others and fully consider their choices at each step in the screening pathway.</td>
</tr>
<tr>
<td>Maternity providers must provide each pregnant woman with information about screening in a manner that enables her to understand the nature and implications of screening.</td>
<td>Ministry of Health consumer resources have been developed to facilitate discussions about screening. These resources should be offered to all women.</td>
</tr>
<tr>
<td></td>
<td>Online education modules and other resources are available for maternity providers via <a href="http://www.nsu.govt.nz">www.nsu.govt.nz</a>.</td>
</tr>
</tbody>
</table>
WAYS OF PROVIDING INFORMATION

| Maternity providers should provide information about antenatal screening for Down syndrome and other conditions in a variety of ways. | Information should be provided in different ways, including:  
- discussions about antenatal screening for Down syndrome and other conditions  
- written resources  
- referral to other sources of information, such as specialist organisations and their websites  
- with the assistance of an interpreter if necessary  
- with family/whānau present [if the woman wishes]. |

FAMILY HISTORY

| Maternity providers must offer to refer women who have previously been pregnant with or have had a child with a significant physical or learning disability, or a family history of a genetic disorder, for a discussion with a specialist obstetrician or geneticist before offering screening. | Maternity providers should exercise particular sensitivity and be aware that these women may have already given careful thought to having or not having screening. |

TAILORING COMMUNICATION TO THE INDIVIDUAL

| Maternity providers should take into account that the woman’s sociocultural environment may impact on the way the message is communicated. | Maternity providers should consider and discuss the woman’s individual circumstances to enable and support informed decision-making. This may involve more than one discussion. |
3.7 Initial discussion

A discussion introducing antenatal screening for Down syndrome and other conditions should be initiated by the maternity provider as early as possible in the pregnancy, to allow women the opportunity to consider participation in screening, ask questions, and seek further information.

The discussion should include:

(a) That the purpose of screening is to provide pregnant women and their families/whānau with information with which to make choices.

(b) The screening options available are first trimester combined screening or second trimester maternal serum screening.

(c) The nature of screening, and the importance of timing:
   First trimester combined screening:
   • Combines a first trimester blood test result with a nuchal translucency scan measurement, and incorporates age, weight, gestation, and other information (as requested on the referral form) to give one result
   • Blood test can be taken between 9 weeks and 13 weeks 6 days, and the optimal timing is 10 to 12 weeks
   • NT scan can be undertaken between 11 weeks and 13 weeks 6 days
   • A co-payment for the NT scan is usually required.

OR

Second trimester maternal serum screening:
• Incorporates a second trimester blood test result with age, weight, gestation, and other information to give one result
• Blood test can be taken between 14 weeks and 20 weeks, and the optimal timing is 14 to 18 weeks.

(d) The conditions screened for:
• Down syndrome (Trisomy 21)
• Trisomy 18 (Edwards syndrome)
• Trisomy 13 (Patau syndrome)
• Neural tube defects
• Some rare metabolic and genetic disorders.

In addition, ultrasound scans can show major fetal structural anomalies, such as skeletal anomalies, brain and neural tube defects, congenital heart defects, and abnormalities of the renal tract, gastrointestinal system and abdominal wall.

(e) That screening is voluntary and the woman has the right to change her mind at any time.
The choices available to women:
- Whether or not to participate in screening
- Involving family/whānau members in the decision-making process
- Receiving selected screening results only.

The limitations of screening:
- Screening is not diagnostic and does not identify all babies who have a condition
- Screening does not cover every disorder
- Screening can give false positive and false negative results.

What screening results might mean:
- There is a low risk that the baby has one of the conditions screened for
- There is an increased risk that the baby has one of the conditions screened for
- That the woman may need to decide whether to proceed to diagnostic testing.

What the woman might do if screening indicates her baby has an increased risk of Down syndrome or another condition:
- Accept a referral to a specialist obstetrician to discuss her options
- Decide not to accept a referral to a specialist obstetrician.

The diagnostic testing options available to the woman in the region, and the risks inherent in these.

The potential benefits of screening:
- Access to information that may provide more choice in the care and management of a pregnancy
- A low risk result means a baby is unlikely to have Down syndrome or another condition screened for.

The potential harms of screening:
- An increased risk result may turn out to be a false positive result
- A low risk result may lead a woman to believe her baby does not have Down syndrome or another condition screened for when the baby does have a condition
- An increased risk result may lead to a decision to have a diagnostic test which has an inherent risk of iatrogenic miscarriage.

Where further information, resources, and support are available, including contact details and availability of support groups, counsellors and cultural advisors.

Information about data collection, monitoring, and reporting, including that the woman’s anonymised information will be used for monitoring and reporting.
3.8 Offer of screening

When offering antenatal screening for Down syndrome and other conditions, maternity providers must ensure that women have been given information that allows them to be aware of the limitations and uncertainties of screening, and in particular the risk of false positive and false negative results.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>OFFERING ACCESS TO SCREENING</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must offer women access to first trimester combined screening or second trimester maternal serum screening.</td>
<td>All pregnant women should be offered access to first trimester combined screening, unless some components of first trimester combined screening are not available in their area, or they have presented later in pregnancy, in which case they should be offered second trimester maternal serum screening.</td>
</tr>
<tr>
<td>Maternity providers must ensure that women are not made to feel that they must accept screening as part of their antenatal care.</td>
<td>Women should understand that only one screening option will be publicly funded in each pregnancy.</td>
</tr>
<tr>
<td><strong>ENSURING INFORMATION HAS BEEN PROVIDED AND DISCUSSED</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must not make an offer of screening without first providing women with information and engaging in a discussion about screening.</td>
<td>The offer should not be made until women have been given all the information listed under the ‘Provision of information’ and ‘Initial discussion’ sections in these Guidelines.</td>
</tr>
<tr>
<td>Provision of information about screening</td>
<td>Some women may wish to discuss their options with family/whānau.</td>
</tr>
<tr>
<td>↓</td>
<td>The overriding principle is the maintenance of the concept of pregnancy as a positive experience.</td>
</tr>
<tr>
<td>Initial discussion</td>
<td>Women are free to decline screening and must be given ongoing unconditional support, regardless of the choices they make during their pregnancy.</td>
</tr>
<tr>
<td>↓</td>
<td></td>
</tr>
<tr>
<td>Offer of screening</td>
<td></td>
</tr>
<tr>
<td><strong>OTHER CONDITIONS SCREENED FOR</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must discuss the other conditions that may be indicated as a result of participation in antenatal screening for Down syndrome and other conditions.</td>
<td>The woman may decide that there are some results that she does not want to be told. For example, a woman may decide that she wants results that show an increased risk of Trisomy 13 and 18 [which may be incompatible with life], but not results that show an increased risk for Down syndrome. However, women should also be advised that it may not be possible to give individual results for specific conditions, as results may indicate an increased risk for more than one condition.</td>
</tr>
</tbody>
</table>
# 3.8.1 First trimester combined screening

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>FIRST TRIMESTER COMBINED SCREENING COMPONENTS AND TIMEFRAMES</strong></td>
<td>For first trimester combined screening, maternity providers must ensure the woman knows there are two components, blood test and NT, and she needs to have each within certain timeframes.</td>
</tr>
<tr>
<td>Maternity providers must advise women where they can go for their blood test and NT scan and when they must have each of these tests.</td>
<td>First trimester maternal serum screening timing is 9 weeks to 13 weeks 6 days, and the optimal sensitivity is 10–12 weeks. The NT scan can be undertaken between 11 weeks and 13 weeks 6 days, and the woman is usually required to make a co-payment for the NT scan.</td>
</tr>
</tbody>
</table>

### COMBINED RESULT

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Maternity providers must advise women that they should expect to receive a single (combined) result from the blood test and the NT.</td>
<td>The results of the two screening tests are combined to provide a single assessment of the likelihood that the baby has Down syndrome or another condition. The laboratory will advise the result to the maternity provider who referred the woman for screening, and the maternity provider must communicate the result to the woman. Maternity providers should ensure that the woman knows that she can expect the combined results within a week to 10 days of her last test.</td>
</tr>
</tbody>
</table>

# 3.8.2 Second trimester maternal serum screening

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>SECOND TRIMESTER MATERNAL SERUM SCREENING TIMEFRAMES</strong></td>
<td>For second trimester maternal serum screening, maternity providers should ensure the woman knows that this does not include NT (although dating scan information may be used if available).</td>
</tr>
<tr>
<td>Maternity providers must advise women where they can go for their blood test and when they must have this test.</td>
<td>Second trimester serum timing is 14–20 weeks and the optimal sensitivity is 14–18 weeks. Maternity providers should ensure that the woman knows she can expect the results within a week to 10 days of the blood test.</td>
</tr>
</tbody>
</table>
3.8.3 Referring women for screening

Referrals for blood tests should be made on the approved referral (laboratory) form. Referrals for NT ultrasound should be made in accordance with the Primary Maternity Services Notice 2007, section DC4, code NT.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>COMPLETING THE LABORATORY REFERRAL FORM</td>
<td>To make an accurate assessment, the laboratory requires:</td>
</tr>
<tr>
<td>Maternity providers must fill in all sections of the laboratory referral form for the laboratory to be able to make an accurate assessment.</td>
<td>• LMP from certain dates</td>
</tr>
<tr>
<td>Contact details of the maternity provider referring the woman for screening must be legible and correct.</td>
<td>• EDD</td>
</tr>
<tr>
<td>Maternity providers must state on the referral form if the woman has seen genetic, specialist, or specialist obstetric services, due to a:</td>
<td>• gestational age according to scan, if available</td>
</tr>
<tr>
<td>• previous pregnancy with or child with a significant physical or learning disability</td>
<td>• maternal weight and date this was taken</td>
</tr>
<tr>
<td>• family history of a genetic condition.</td>
<td>• maternal age</td>
</tr>
<tr>
<td></td>
<td>• maternal smoking status</td>
</tr>
<tr>
<td></td>
<td>• presence of insulin dependent diabetes mellitus</td>
</tr>
<tr>
<td></td>
<td>• Whether this is a multiple pregnancy (if so, how many)</td>
</tr>
<tr>
<td></td>
<td>• whether this is an IVF pregnancy and, if so, the egg donor’s or mother’s age at time of donation or egg retrieval</td>
</tr>
<tr>
<td></td>
<td>• history of threatened miscarriage in this pregnancy</td>
</tr>
<tr>
<td></td>
<td>• any history of a previous pregnancy with Down syndrome or another condition screened for</td>
</tr>
<tr>
<td></td>
<td>• copies of any scans (for both trimesters) and details about those scans.</td>
</tr>
</tbody>
</table>
### 3.9 Laboratory processes

The laboratory will provide a report to the maternity provider who referred the woman for screening.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
</table>
| **LABORATORY SERVICES: REPORTING INFORMATION TO THE MATERNITY PROVIDER** | The laboratory report will include:  
- screening result (‘increased risk’ or ‘low risk’)  
- multiples of the median (MoMs) of each analyte (not raw numbers)  
- individual risk assessments for:  
  - Down syndrome (Trisomy 21)  
  - Trisomy 18 (Edwards syndrome)  
  - Trisomy 13 (Patau syndrome)  
  - Neural tube defects (for second trimester, but not for first trimester). |
| The laboratory is responsible for providing a first trimester combined screening or second trimester maternal serum screening result to the maternity provider who referred the woman for screening, by electronic means and/or hard copy reporting. | The risk result will be calculated for the pregnancy at term, rather than for the gestation at the time of screening, and reported as a proportion. |
| **LABORATORY SERVICES: PROVISION OF SPECIALIST LABORATORY ADVICE** | If specialist laboratory advice is indicated, the laboratory will contact the maternity provider. |
| The laboratory is responsible for providing specialist laboratory advice to the maternity provider. |  |
| **LABORATORY SERVICES: REPORTING INFORMATION TO THE NATIONAL SCREENING UNIT** | Reporting information required by the NSU from the laboratory includes:  
- separate data for first and second trimester screening  
- number of women screened  
- ethnicity of women screened  
- geographical location of women screened  
- referrer  
- number of business days between receipt of sample and provision of risk calculation  
- number of increased risk results  
- number of low risk results. |
| The laboratory is responsible for providing reporting information to the NSU. |  |
### 3.9.1 Laboratory responsibilities

<table>
<thead>
<tr>
<th>Responsibilities</th>
<th>First Trimester</th>
<th>Second Trimester</th>
</tr>
</thead>
<tbody>
<tr>
<td>Collection and transportation of all blood samples for the purpose of antenatal screening for Down syndrome and other conditions</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Supply and distribution of laboratory referral forms to maternity providers for the collection of blood specimens, pregnancy dating information and other required pregnancy and personal data</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>(Where available) the receipt of scan information from radiology providers of fetal measurements including NT, crown-rump length (CRL) or biparietal diameter (BPD) (if scanning is after 12 weeks)</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Analysis of the appropriate analytes [biochemical markers] in maternal serum in the first trimester: Pregnancy-associated plasma protein-A (PAPP-A) and free beta-human chorionic gonadotrophin (ßhCG)</td>
<td>✓</td>
<td></td>
</tr>
<tr>
<td>Analysis of the appropriate analytes [biochemical markers] in maternal serum in the second trimester: alpha-fetoprotein (AFP), ßhCG, unconjugated oestriol (µE₃) and inhibin A</td>
<td></td>
<td>✓</td>
</tr>
<tr>
<td>Application of an algorithm using the appropriate biochemical and ultrasound markers to calculate an assessment of risk that a pregnancy may be affected by Down syndrome or another condition screened for</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Provision of first trimester combined screening and second trimester maternal serum screening results and information to maternity providers who have referred women for screening by telephone (in the case of increased risk results) and by electronic means and/or hard copy</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Provision of specialist advice to and availability for consultation with maternity providers who have referred women for screening</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Provision of reporting information to the NSU</td>
<td>✓</td>
<td>✓</td>
</tr>
</tbody>
</table>
3.9.2 Exceptions
The laboratory will contact the maternity provider who referred the woman for screening in the following circumstances.

<table>
<thead>
<tr>
<th>LABORATORY SERVICES: EXCEPTIONS</th>
<th>Action</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Scenario</strong></td>
<td><strong>Action</strong></td>
</tr>
</tbody>
</table>
| The laboratory receives a blood sample for first trimester combined screening, but no NT. | The laboratory will advise the maternity provider to contact the woman about the need to have her NT performed before 13 weeks 6 days for a first trimester combined screening risk assessment to be possible.  
If the NT scan has not been performed by 13 weeks 6 days, the laboratory will:  
• issue a report to the maternity provider that provides the values of the analytes  
• advise that first trimester combined screening cannot be completed because the scan data was not available  
• recommend that the woman is offered second trimester maternal serum screening.  
The maternity provider should advise the woman that first trimester combined screening has not been completed and invite her to participate in second trimester maternal serum screening.  
However, if the NT scan has been done before 13 weeks 6 days, it can still be sent to the laboratory for first trimester combined screening to be completed. |
| The laboratory receives a scan result indicating the pregnancy is more advanced than 13 weeks 6 days. | The laboratory will advise the maternity provider that the woman’s LMP suggests she is within 13 weeks 6 days but the scan measurements indicate a more advanced pregnancy, and that first trimester combined screening will not be completed.  
The maternity provider should advise the woman that first trimester combined screening has not been completed and invite her to participate in second trimester maternal serum screening. |
| The laboratory receives a blood sample for second trimester maternal serum screening for a woman who has already completed first trimester combined screening. | The laboratory will advise the maternity provider that first trimester combined screening has already been accessed by the woman for this pregnancy.  
The maternity provider should advise the woman that second trimester maternal serum screening will not be publicly funded. |
| The laboratory receives a blood sample for second trimester maternal serum screening with an NT scan result. | The laboratory will provide a risk result calculation that does not incorporate the NT. The report will state that NT measurement has not been incorporated into the assessment. |
3.10 Results

The maternity provider who referred the woman for screening is responsible for communicating the screening results to the woman.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>RECEIVING SCREENING RESULTS</td>
<td>The laboratory must ensure that maternity providers have all the information required to inform women of their screening results. Screenings will be completed by the laboratory within three business days after the receipt of the blood sample and scan information. Sample transit times to the laboratory may vary between regions, but should generally be within two to three days. If the screening result is low risk the laboratory will dispatch the results to the maternity provider by mail or electronic means within 24 hours of the results being available. If the screening result is increased risk the laboratory will contact the maternity provider by telephone within 24 hours of the results being available. Results will also be dispatched to the maternity provider by mail or electronically. Maternity providers must check the woman’s notes to confirm if there are any results the woman did not want to be told, before communicating the results. If the radiologist finds a significant anomaly during the NT scan, they may inform the woman directly and/or advise the maternity provider that urgent referral is required.</td>
</tr>
</tbody>
</table>
### COMMUNICATING SCREENING RESULTS

<table>
<thead>
<tr>
<th>Maternity providers must understand what screening results mean and be able to present them in a clear and concise way to support women in their decision-making. This includes understanding statistical risk information.</th>
<th>All results, whether low risk or increased risk, should be given in person.</th>
</tr>
</thead>
<tbody>
<tr>
<td>An appropriate private and comfortable environment should be provided for giving results.</td>
<td>Giving results may include:</td>
</tr>
<tr>
<td>• discussing the limitations of screening</td>
<td></td>
</tr>
<tr>
<td>• discussing that a low risk result means that the baby is unlikely to be born with one of the conditions screened for, but it does not mean they will definitely not be born with one of these or another condition not indicated by screening</td>
<td></td>
</tr>
<tr>
<td>• providing an opportunity for the woman (and her family/whānau) to ask questions</td>
<td></td>
</tr>
<tr>
<td>• providing information about other services, including community support agencies the woman (and her family/whānau) can contact if they have specific concerns.</td>
<td></td>
</tr>
<tr>
<td>If a woman with a low risk result wants a referral to a specialist obstetrician, the maternity provider should make a referral.</td>
<td></td>
</tr>
</tbody>
</table>

### ADDITIONAL SUPPORT

<table>
<thead>
<tr>
<th>Maternity providers must offer additional support to women who may have difficulty understanding information because of language difficulties (e.g., English as a second language), hearing impairment or intellectual disability.</th>
<th>Using family members or friends as interpreters is not recommended practice. Use Language Line, a DHB interpreter, or a New Zealand sign language (NZSL) interpreter.</th>
</tr>
</thead>
<tbody>
<tr>
<td>The woman may wish to have family/whānau members present.</td>
<td></td>
</tr>
<tr>
<td>Women with diminished competence may wish to have an independent advocate present, to support their decision-making. Some women with diminished competence may have a welfare guardian, who should be present to assist their decision-making.</td>
<td></td>
</tr>
</tbody>
</table>
### 3.10.1 Increased risk results

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>COMMUNICATING INCREASED RISK RESULTS</strong></td>
<td>The Ministry of Health increased chance result consumer resource has been developed to facilitate the discussion of results.</td>
</tr>
<tr>
<td>Maternity providers must inform women of all screening results indicating an increased risk of Down syndrome or another condition, except where a woman has indicated that she does not want to be told certain results.</td>
<td>Consideration should be given to the timing of giving results, and whether access to support services or further information is available (e.g., on public holidays).</td>
</tr>
<tr>
<td>Maternity providers must provide accurate and balanced information to all women.</td>
<td></td>
</tr>
<tr>
<td>Maternity providers must discuss increased risk results with women as soon as possible.</td>
<td></td>
</tr>
<tr>
<td><strong>OFFER OF SPECIALIST OBSTETRIC REFERRAL</strong></td>
<td>Women with increased risk results should be offered a referral to discuss their results and options with a specialist obstetrician.</td>
</tr>
<tr>
<td>Maternity providers should offer all women with increased risk results a specialist obstetric referral.</td>
<td>Acceptance of referral does not mean acceptance of diagnostic testing.</td>
</tr>
<tr>
<td></td>
<td>The maternity provider should discuss what an appointment with a specialist obstetrician might involve and what the woman’s options might be.</td>
</tr>
<tr>
<td><strong>PROVISION OF INFORMATION ABOUT OPTIONS FOR SPECIALIST REFERRAL</strong></td>
<td>The woman should be given information about diagnostic testing including:</td>
</tr>
<tr>
<td>Maternity providers must provide information about the options available for specialist obstetric referral and diagnostic testing.</td>
<td>• Chorionic villus sampling (CVS) can be performed from 11–14 weeks of pregnancy but is typically performed between 10 and 13 weeks. CVS is presently restricted to a few sites. CVS results may take one to three weeks.</td>
</tr>
<tr>
<td></td>
<td>• Amniocentesis can be performed from 14 weeks of pregnancy and is typically performed between 15 and 20 weeks. Amniocentesis results may take one to three weeks.</td>
</tr>
<tr>
<td></td>
<td>• There are risks associated with both CVS and amniocentesis. Both procedures carry a miscarriage risk of approximately 1 percent above the spontaneous miscarriage rate. Because of this risk, some women may decide not to have these tests (or choose not to participate in screening).</td>
</tr>
<tr>
<td></td>
<td>A decision about diagnostic testing involves a separate informed consent process after specialist obstetric consultation.</td>
</tr>
<tr>
<td></td>
<td>Informed consent for diagnostic testing is the responsibility of the specialist who performs the procedure.</td>
</tr>
</tbody>
</table>
3.11 Genetic counselling and other referrals

Maternity providers should be able to provide women with information about other relevant support services in their area and how to access them, if requested.

<table>
<thead>
<tr>
<th>Guidelines</th>
<th>Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>GENETIC COUNSELLING SERVICES</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers should advise women with increased risk results about the availability of genetic counselling services.</td>
<td>Women with increased risk results will usually have an appointment with a specialist obstetrician as the first step about making an informed decision about what to do next. However, some women may also choose to talk to Genetic Services.</td>
</tr>
<tr>
<td></td>
<td>Genetic Services are physically located in Auckland, Wellington, and Christchurch. Telephone counselling is available to women who cannot access these centres.</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>OTHER REFERRALS</strong></td>
<td></td>
</tr>
<tr>
<td>Maternity providers must provide information about medical and non-medical services the woman may access to help her make decisions about the management of her pregnancy.</td>
<td>Maternity providers should find out about services in their area and how to access them. Services include:</td>
</tr>
<tr>
<td></td>
<td>• obstetricians</td>
</tr>
<tr>
<td></td>
<td>• paediatricians</td>
</tr>
<tr>
<td></td>
<td>• Genetic Services</td>
</tr>
<tr>
<td></td>
<td>• counsellors</td>
</tr>
<tr>
<td></td>
<td>• interpreters</td>
</tr>
<tr>
<td></td>
<td>• disability support services</td>
</tr>
<tr>
<td></td>
<td>• culturally appropriate voluntary agencies</td>
</tr>
<tr>
<td></td>
<td>• consumer support providers/associations</td>
</tr>
<tr>
<td></td>
<td>• ultrasound providers</td>
</tr>
<tr>
<td></td>
<td>Maternity providers should advise what options are available and the timeframes of these, and find ways to provide practical support and assistance.</td>
</tr>
</tbody>
</table>
3.12 Funding of diagnostic testing

Diagnostic testing is publicly funded for women who have:
- an increased risk result
- an abnormal ultrasound scan (structural abnormalities)
- previously had a baby with a congenital anomaly
- a family history of Down syndrome and/or other conditions, if recommended by Genetic Services
- a known carrier status, or a partner with a known carrier status, if recommended by Genetic Services.

In some areas, diagnostic testing is also funded for women who are aged over 35 years. However, international best practice does not support direct referral to diagnostic testing based on age.

3.13 Data collection, monitoring and reporting

The data reported by the laboratory to the NSU includes the:
- number of women screened
- ethnicity of women screened
- geographical location of women screened
- referrer
- number of business days between receipt of sample and provision of risk calculation
- number of increased risk results
- number of low risk results
- number of repeat sample requests.

Data is provided separately for first and second trimester screening.

While complete data on maternity services and pregnancy outcomes are not readily collated in New Zealand, this will improve over time. Monitoring and evaluation will follow the screening pathway and assess process and outcomes, with the aim of assisting the ongoing development of the quality improvements for antenatal screening for Down syndrome and other conditions and improving equity of access to screening.

Where feasible and appropriate, indicators will be calculated and reported by:
- DHB
- ethnicity
- deprivation status
- first and second trimester screening.
APPENDIX ONE: GLOSSARY OF TERMS

**Amniocentesis** is a procedure involving the withdrawal of a small amount of amniotic fluid which contains fetal cells. The sample is used to obtain fetal chromosomes for karyotype analysis.

**Alpha-fetoprotein (AFP)** is a biochemical marker used in second trimester maternal serum screening for neural tube defects and Down syndrome and other conditions.

**Beta-human chorionic gonadotropin (ßhCG)** is a biochemical marker used in first trimester combined and second trimester maternal serum screening for Down syndrome and other conditions.

**Biparietal Diameter (BPD)** is the distance between the parietal bones at their widest point as measured during a fetal ultrasound.

**Chorionic villus sampling (CVS)** is a procedure involving the withdrawal of a small amount of chorionic villi. The sample is used to obtain chromosomes for karyotype analysis to identify some genetic disorders.

**Crown-rump length (CRL)** means the distance from the top of the fetal skull to the bottom of the spine, measured during fetal ultrasound.

**Cut-off point** is the value of a screening variable at which an individual is more likely to be helped than harmed by the offer of a diagnostic test. This point forms the division between low risk and increased risk. In both screening options for Down syndrome and other conditions the cut-off point is 1:300.

A **false positive result** is a positive screening result for a condition in a person who does not have the condition.

A **false negative result** is a negative screening result for a condition in a person who has the condition.

**Inhibin A** is a biochemical marker used in second trimester maternal serum screening for Down syndrome and other conditions.

**Karyotype** is a depiction of all the chromosomes in an individual cell. Any chromosomal abnormality that can be viewed via a microscope will be visible in the karyotype. The most common of these include:

- Trisomies, triploidy and sex chromosome abnormalities
- Structural genetic abnormalities, such as inversions and translocations, deletions and duplications of chromosomes

**Last menstrual period (LMP)** is the estimated or actual date of the first day of a woman’s last menstrual period.

**Multiple of the Median (MoM)** is a measure which compares the values of a biochemical marker in an individual sample with the median value of that biochemical marker in other women at the same gestation.

**Nuchal translucency (NT)** is an ultrasound marker which measures the fluid-filled space in the tissue at the back of a fetus’ neck and is a marker for chromosomal and other anomalies.
Pregnancy-associated plasma protein A (PAPP-A) is a biochemical marker used in first trimester combined screening for Down syndrome and other conditions.

Radiologist is a health practitioner who is, or is deemed to be, registered by the Medical Council of New Zealand (established by the Health Practitioners Competence Assurance Act 2003) in the vocational scope of diagnostic and interventional radiology and holds an annual practicing certificate.

Screening is a way of identifying a group of people who are more likely than others to have a condition. The screening process involves testing people (who may not have symptoms) for the presence of the condition, and predicting the likelihood that they have the condition. Antenatal screening for Down syndrome and other conditions predicts the likelihood of the conditions being present in the fetus.

Sensitivity is the ability of screening to identify persons with the condition screened for. A test with high sensitivity will have few false negative results.

Specificity is the ability of screening to identify persons who do not have the condition screened for. A test with high specificity will have few false positive results.

Spina bifida is a neural tube defect marked by congenital cleft of the spinal column usually with hernial protrusion of the meninges and sometimes the spinal cord.

Trisomy 18 (Edwards syndrome) is a chromosomal condition associated with severe intellectual disability and abnormalities in many parts of the body. Trisomy 18 is characterised by a low birth weight, a small, abnormally shaped head, a small jaw and mouth, clenched fists with overlapping fingers, heart defects, and abnormalities of other organs. Due to the presence of several life-threatening medical problems, babies with Trisomy 18 may be stillborn, and almost all will die within their first month.

Trisomy 13 (Patau syndrome) is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Trisomy 13 is characterised by heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers and/or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate), and weak muscle tone (hypotonia). Due to the presence of several life-threatening medical problems, the majority of babies with Trisomy 13 are stillborn, or die within their first days or weeks of life.

Unconjugated oestriol (μE₃) is a biochemical marker used in second trimester maternal serum screening for Down syndrome and other conditions.

For further information refer to the NHS glossary of screening terminology at http://www.screening.nhs.uk/screening
APPENDIX TWO: RESOURCES AND CONTACTS

Contact details for support services and sources of further information are listed here. This list should be supplemented by the local or regional services within your own networks.

Auckland District Health Board
Management of Babies with Down Syndrome

Australian Centre for Genetics Education
Down Syndrome Fact Sheet
Changes to Chromosomes – Number, Size and Structure Fact sheet

Antenatal Results and Choices (UK)
http://www.arc-uk.org/

CCS Disability Action
http://www.ccs.org.nz
Tel. 0800 227 200

Disability Services
http://www.moh.govt.nz/disability

Down’s Syndrome Association (UK)
A New Parent’s Guide
http://www.downs-syndrome.org.uk

Down syndrome online
http://www.down-syndrome.org

Fetal Medicine Foundation, London
http://www.fetalmedicine.com/FMF/
The 11–13+6 Weeks Scan

Genetic Services, New Zealand
Northern and Midland Region
Tel: 0800 476 123

Central Region
Tel: 0508 364 436

Southern Region
Tel: 0508 364 436

Health and Disability Commissioner
http://www.hdc.org.nz/

Human Genetic Society of Australasia (HGSA)
http://www.hgsa.com.au

IHC
http://www.ihc.org.nz
Tel. (04) 472 2247

International Mosaic Down Syndrome Association
Booklet for professionals
http://www.imdsa.org/Information/professional.htm
APPENDIX THREE: BIBLIOGRAPHY

Chang Choong T. 2005. *Antenatal Screening for Down Syndrome in New Zealand: Time for a national screening policy?* Department of Obstetrics and Gynaecology, Faculty of Medical and Health Sciences, University of Auckland.


