National Screening Unit

Antenatal Down Syndrome Screening in New Zealand 2007

A Report of the Antenatal Down Syndrome Screening Advisory Group to the National Screening Unit

A Report of the Antenatal Down Syndrome Screening Advisory Group
to the National Screening Unit, January 2007

Published in March 2007 by the National Screening Unit
PO Box 5013, Wellington, New Zealand

ISBN HP4394

This document is available on the Ministry of Health website:
http://www.moh.govt.nz

Contents

Foreword		v
Executive summary		1
Recommendations		3
1.0	**Background**	**6**
	Establishment of the Antenatal Down Syndrome Screening Advisory Group	6
	Process for developing report	7
2.0	**What is down syndrome?**	**8**
	Characteristics of Down syndrome	8
	Why is it called Down syndrome?	9
	Epidemiology	9
	Other conditions identified through screening	9
3.0	**What is screening?**	**10**
	The difference between screening and diagnosis	10
	The limitations of screening	10
	The screening pathway	10
	Opportunistic screening versus screening programmes	11
4.0	**What are the social and ethical issues in screening for Down syndrome?**	**12**
	Ethical principles	12
	The pivotal role of ethical debate	14
	Inequalities in screening	15
5.0	**Current methods of screening for Down syndrome in New Zealand**	**17**
	Purpose of screening for Down syndrome and other fetal anomalies	17
	Methods of screening currently used in New Zealand	17
	Diagnostic tests currently used in New Zealand	19
	New Zealand practice compared with international best practice	20
6.0	**Problems with current Down syndrome screening in New Zealand**	**22**
	Assessment of antenatal screening for Down syndrome	22
	The offer of screening	22
	Information to support decision-making	23
	Informed consent to screening	24
	Problems with the types of screening currently performed	24

7.0	Improving the quality of screening	29
	Balancing sensitivity and specificity	29
	Options for better tests	29
8.0	Options for action	33
	Option A: Do nothing	33
	Option B: Stop some, or all, elements of antenatal screening for Down syndrome	34
	Option C: Improve the current situation	34
9.0	Assessment of antenatal screening for down syndrome against criteria to assess screening programmes	49
	National Health Committee criteria for assessing screening programmes	49
	Are these appropriate criteria to assess antenatal screening programmes?	52
10.0	Preferred options	54
	Majority preference: continue to offer screening and to improve the quality and safety of screening tests	54
	Minority preference	55
References		56
Glossary		58
Appendices		62
Appendix 1: Membership of the Antenatal Down Syndrome Screening Advisory Group		64
Appendix 2: Social and Ethical Subgroup: Report to the Antenatal Down Syndrome Screening Advisory Group		64
Appendix 3: Nuchal Translucency Subgroup: Report to the Antenatal Down Syndrome Screening Advisory Group		73
Appendix 4: Maternal Serum Screening Subgroup: Report to Antenatal Down Syndrome Screening Advisory Group		87

Foreword

Down syndrome is a congenital disorder that causes severe learning disability. It occurs in approximately one in seven hundred pregnancies with about seventy affected children born in New Zealand each year. There is a wide variation in the degree of disability between individuals with Down syndrome. Some adults lead independent lives as paid members of the work force while many require significant support throughout their lives. Down syndrome is often associated with significant health issues including vision and hearing problems and major cardiac anomalies.

It has been known for many years that the incidence of Down syndrome increases with maternal age. This observation led to the earliest form of screening with some women over 35 being offered amniocentesis to identify affected fetuses. More recently nuchal fold thickness, measured on ultrasound, and a number of biochemical tests have emerged as additional methods of screening for Down syndrome. These have been tested in international clinical trials confirming that a combination of tests used in a formal screening programme improves the quality and safety of this type of screening. However, in New Zealand these screening tests have crept into practice in an ad hoc manner with little consideration about the most appropriate tests to use or the ethical and social consequences of this type of screening.

The Antenatal Down Syndrome Screening Advisory Group was established in response to a report commissioned by the National Screening Unit to review the existing problems with Down syndrome screening practice. The Stone and Austin Report highlighted many deficiencies including the use of outdated testing, inequity of access, lack of knowledge amongst health care professionals, no process to audit practice, and a lack of adequate information and counselling for parents. The major concern identified was that the existing practice is putting women through undue stress and results in unnecessary pregnancy loss.

The Advisory Group was asked to examine interim measures to improve access, safety and quality of the existing screening and to examine the need for and acceptability of a nationally organised Down syndrome screening programme.

The Advisory Group recognises that antenatal screening for Down syndrome is highly complex and involves many contentious issues. In order to encompass the full range of views and to ensure that all opinion was heard, discussion was made as broad as possible. It was not the intention of the Advisory Group to reach consensus on all issues as the members acknowledge that various interest groups hold a diverse range of views, often founded on fundamentally different values. The aim was rather to highlight significant issues so that the government recognises the complexity of this type of screening. The social and ethical considerations were particularly controversial and were debated in more depth by a subcommittee with broad representation including health care professionals, women's health advocates, disability advocates, and people with experience in health law and ethics. The debate touched on a broad range of topics including parental choice, termination of pregnancy, the rights of the fetus, issues of informed consent and the impact screening has on people with disabilities.

Separate subcommittees reviewed the technical aspects of screening, including methods to improve the quality of nuchal translucency scanning and discussion around the potential introduction of maternal serum testing in pregnancy and which combination of tests provides the best risk assessment. This is a rapidly changing field and will require ongoing review.

This report identifies the main issues relating to antenatal screening for Down syndrome and presents a number of options for consideration. The advantages and disadvantages of each option are considered in detail and a number of recommendations formulated. The Advisory Group were unanimous that the present screening practice is unsafe and should not continue. However, we were unable to reach unanimous agreement on the best way forward. The lack of consensus could be seen as a weakness of the report, but I believe it is one of its main strengths as it more truly reflects the diversity of views in society in general. Many of the recommendations in this document have evolved from careful consideration, balancing a combination of ethical, social and scientific factors, a process that some of the Advisory Group found challenging. The outcome was that the majority recommended that high-quality screening should be offered but this must be supported with adequate counselling services and education for health care professionals to ensure that prospective parents can make a truly informed choice. However a minority were opposed to a national screening programme; a view that should not be ignored. It is important to reflect on how a screening programme would be perceived by people with disabilities and to consider what it is saying about people with Down syndrome.

The Advisory Group considers that this report should be made widely available. We expect that the contents will stimulate wider public debate. The Advisory Group acknowledge that some change has to be implemented to modify existing practice and recognise that whatever decision is made will not be acceptable to everybody. It is important to note that a screening programme will subtly alter the make-up of society, something we must only embark upon after very careful consideration

Dr Paul Harper
Chair

Executive summary

This review of antenatal screening for Down syndrome and other fetal anomalies has been carried out because New Zealand has not kept pace with significant international developments in best practice, and there is widespread concern about the safety of current screening practices.

At present, screening for Down syndrome and other fetal anomalies in New Zealand is opportunistic. The most commonly used method is a risk assessment based on nuchal translucency (NT) and/or maternal age. International research indicates that these measures are the least safe and most inaccurate means of antenatal screening for Down syndrome. Using these methods results in women being unnecessarily referred for an invasive diagnostic procedure, which can result in miscarriage. These safety concerns are exacerbated by the lack of standards, monitoring, evaluation or quality assurance processes.

The purpose of antenatal screening is to provide pregnant women, who choose to have the screening tests, with information about whether the fetus is likely to have Down syndrome or some other fetal anomaly. This information may help women to make informed decisions and plans about their pregnancy, including:

- whether to have diagnostic testing
- whether to continue with or terminate the pregnancy
- preparing for the birth and life of a child with Down syndrome or other condition
- planning to give birth at a specialist centre that can provide any immediate neonatal treatment, if necessary.

The role of the Antenatal Down Syndrome Screening Advisory Group (the Advisory Group) included consideration of the broader aspects of screening, including the potential to identify other conditions and the social and ethical issues relating to screening. Most of the Advisory Group agreed that other fetal conditions that may be identified during screening for Down syndrome should also be considered. It was also agreed that screening should be considered in the context of pregnancy and other tests (mostly screening) offered to pregnant women.

The social and ethical considerations have proven to be particularly complex and difficult. This is largely due to the links between screening and other complex issues, including termination of pregnancy, social and medical models of disability, and whether there should be limits to reproductive autonomy.

Options for action

Taking the social, ethical and technical considerations into account, the Advisory Group developed three broad options for action in New Zealand.

1. Do nothing and allow the currently unsafe, opportunistic screening to continue.
2. Stop some, or all, elements of screening (either by prohibiting screening completely or by prohibiting public funding being used to support screening).
3. Continue to offer screening, and take steps to improve the quality and safety of screening tests (including the potential for a nationally organised screening programme).

A number of sub-options sit under these three options, providing further choices for action. These options and sub-options are discussed in greater detail in section 8.

Regardless of which of these options is chosen, consideration should be given to directing additional funding to disability support services. Relevant examples of ways additional funding could be used in the disability sector can be found in a report titled *To Have an Ordinary Life: Community membership for adults with an intellectual disability* (National Health Committee 2003b).

The Advisory Group was unable to reach a unanimous position on the best way forward for antenatal screening for Down syndrome in New Zealand. It is expected that due to the complex and difficult nature of the issues, the general public in New Zealand will have the same difficulty in reaching agreement. Despite this, there was one point on which the entire Advisory Group could agree: the Ministry of Health must not 'do nothing' about the current situation of antenatal screening for Down syndrome. The Advisory Group agreed that the current practice of screening by NT and/or maternal age alone is unacceptable and should not continue.

Without a unanimous position on the best option for action, the Advisory Group can only describe its members' preferences for action with the aim of providing some guidance to the Ministry of Health.

Preferred options

A large majority of Advisory Group members consider the best option is to continue to offer screening and to improve the quality and safety of screening tests using the structures of a nationally organised screening programme to facilitate improvements in quality and safety. This is discussed in further detail in section 10.

A minority of Advisory Group members do not support a national screening programme. International literature suggests that a national screening programme will lead to reduced incidence of Down syndrome and other fetal anomalies, which is a cause of concern in the disability community. The minority believes, regardless of what is decided in relation to screening, that the best option is to consider directing additional resource to disability support services. These issues are discussed in further detail in section 10.

If screening is to continue

Most members of the Advisory Group consider that if a woman is fully informed and chooses to have screening, then there is a responsibility to provide access to the safest and most effective test. There was general consensus that if screening is to continue to be offered in New Zealand it should meet a set of minimum conditions. These are described in Recommendation 3 (below).

Should there be a national screening programme?

It is untenable to continue to offer the current unsafe and unreliable forms of opportunistic screening. If screening is to continue, it must be nationally consistent and monitored to ensure it is safe and effective. A nationally organised screening programme can provide the structure to:

- monitor quality and safety
- strengthen processes for informed consent
- provide high-quality information for consumers and practitioners
- evaluate the programme at a national level, which is particularly important when the screening pathway is complex and involves a large number of health practitioners and providers.

It needs to be acknowledged that some groups are concerned about the potential for a national screening programme for Down syndrome. In particular, there are concerns about the messages this may send to the general public about disability and the undesirability of having a child with Down syndrome.

As part of its deliberations, the Advisory Group evaluated the criteria to assess national screening programmes (National Health Committee 2003a). These criteria have previously been used to assess whether national screening programmes should be implemented for prostate cancer screening, colorectal cancer screening and antenatal HIV screening. However, they have never before been used to assess antenatal screening for a condition associated with disability, which requires a number of different considerations.

Many members of the Advisory Group consider that some of the wording of the criteria to assess screening programmes is unhelpful when applied to antenatal screening for disability, and that more appropriate wording could be used. As a result the Advisory Group considered the relevance of criteria developed in the Netherlands to assess 'genetic screening' (Netherlands Health Council 1994).

Recommendations

After considering the social, ethical and technical implications, and with due regard to the complexities of the issue, the Advisory Group makes the following recommendations.

1. **The current practice of screening using only nuchal translucency and/or maternal age, without biochemical markers, is unsafe and inequitable, and should not continue.**

The purpose of screening

2. **If antenatal screening for Down syndrome is to continue in New Zealand it should be:**
   a. **explicitly stated that the purpose of screening is to provide pregnant women, who choose to have the tests, with information about whether the fetus is likely to have Down syndrome or other fetal anomaly**
   b. **explicitly stated that the purpose of screening is not to reduce the incidence of Down syndrome or any other condition**
   c. **explicitly affirmed that women choosing to continue their pregnancy, and their families, will be eligible for ongoing relevant support services.**

Social and ethical considerations in screening

3. **If antenatal screening for Down syndrome is to continue in New Zealand it should be:**
   a. **universally offered to all pregnant women**
   b. **accompanied by high-quality, balanced, easily understood information in an accessible range of formats for practitioners and consumers**
   c. **conditional on informed consent**
   d. **based on voluntary participation at each stage of the screening and diagnostic pathway**
   e. **based on unconditional acceptance of, and support for, the choices made by women as a result of screening and diagnostic tests**
   f. **delivered within a framework that is responsive to Māori and improves opportunities for Māori to make informed decisions**
   g. **integrated with other tests offered in pregnancy**
   h. **the safest and most effective tests, based on evolving international evidence**
   i. **focused on reducing inequalities in access to antenatal screening for Down syndrome**
   j. **fully funded so that any woman who chooses to have screening is able to access it**
   k. **monitored and evaluated at a national level.**

Technical considerations in screening

4. If antenatal screening for Down syndrome is to continue in New Zealand:
   a. the screening test should consist of the best combination of ultrasound and biochemical markers, based on evolving international evidence
   b. second trimester maternal serum screening (the quadruple test) with a dating scan should be offered to all women presenting for the first time in the second trimester of pregnancy
   c. serum integrated screening, with a dating scan, should be offered to all women who are not able to access high-quality nuchal translucency performed by an accredited practitioner
   d. practitioners performing screening tests should have to comply with best practice standards
   e. practitioners performing diagnostic tests should have to comply with best practice standards.

Should there be a national screening programme?

5. If it is decided to implement a national antenatal Down syndrome screening programme, the programme should:
   a. meet all elements outlined in recommendations 2, 3 and 4
   b. not be expected to meet targets in relation to participation or coverage, but have a focus on reducing inequalities in access to antenatal screening for Down syndrome
   c. be supported by an advisory group established to provide ethical and technical advice on the best form of screening for New Zealand, including issues relating to safety, consumer preferences, data collection and privacy
   d. be subject to routine monitoring and evaluation, and modified as necessary and appropriate.

6. To encourage wider societal debate, the Government should consider carrying out public consultation to determine the public's views on whether or not there should be an antenatal Down syndrome screening programme.

7. If public consultation does occur, then it should:
   a. include information on all options being considered for an antenatal Down syndrome screening programme, and this information should be provided in accessible formats and in a way that is easily understood by a wide range of audiences
   b. include information on how an antenatal Down syndrome screening programme might be implemented.

8. The criteria to assess screening programmes should be reviewed by the appropriate committee so that they provide greater guidance for the assessment of a wider range of screening, including antenatal screening and screening for genetic conditions.

9. A Ministerial committee should be established to approve the introduction of new technologies into New Zealand. The committee should be required to consider the social, ethical, technical and cost implications relating to the introduction of all new health technologies, in both the public and private sector.

Abbreviations

AFP	Alphafetoprotein (a biochemical marker)
CRL	Crown-rump length
CVS	Chorionic villus sampling
DHB	District Health Board
FASTER	First-trimester or second-trimester screening (a study)
FISH	Fluorescent in situ hybridisation
FMF	Fetal Medicine Foundation
hCG	human chorionic gonadatrophin (a biochemical marker)
IANZ	International Accreditation New Zealand
ICF	International Classification of Functioning
LMC	Lead Maternity Carer
MRT	Medical Radiation Technologist
NSU	National Screening Unit of the Ministry of Health
NT	Nuchal Translucency (an ultrasound marker)
PAPP-A	Pregnancy associated plasma protein A (a biochemical marker)
RANZCR	Royal Australian and New Zealand College of Radiologists
RANZCOG	Royal Australian and New Zealand College of Obstetricians and Gynaecologists
SURUSS	The Serum, Urine and Ultrasound Study
uE3	Unconjugated oestriol (a biochemical marker)

1.0 Background

There has been growing concern that the current practice of antenatal screening for Down syndrome in New Zealand is not in accordance with best practice. These concerns have intensified over the last five years as evidence supporting improved screening practices has grown. Concerns about the quality and safety of current practice have been raised by the New Zealand College of Midwives, the Royal Australian and New Zealand College of Obstetricians and Gynaecologists, the Royal Australian and New Zealand College of Radiologists, women's health organisations, health practitioners, academics, members of the National Screening Advisory Committee, and within the Ministry of Health.

Establishment of the Antenatal Down Syndrome Screening Advisory Group

In June 2005 the National Screening Unit (NSU) commissioned Professor Peter Stone and Diana Austin to assess antenatal screening for Down syndrome in New Zealand. As part of their work, Stone and Austin:

- reviewed the literature and best practice for antenatal screening for Down syndrome
- assessed current practice in New Zealand
- discussed issues for implementing a New Zealand screening programme, based on the research into current practice and the views of health practitioners and consumers
- surveyed and collated information on screening for Down syndrome that could be used in a New Zealand context (Stone and Austin 2006).

One of the key findings of the Stone and Austin report was that 'the current screening process in New Zealand was identified as being a physical, emotional and social risk to families with the worst of all options being available and not in line with international research' (Stone and Austin 2006). Stone and Austin presented their report to the NSU in February 2006. Following publication of the report, the Minister of Health agreed to the NSU developing policy on antenatal screening for Down syndrome. As part of this process the NSU established the Antenatal Down Syndrome Screening Advisory Group (the Advisory Group).

The NSU established the Advisory Group to provide technical and strategic advice on the appropriateness and feasibility of a national antenatal Down syndrome screening programme in New Zealand. In particular, the Advisory Group was asked to provide advice on:

- interim steps to improve the access, safety and quality of existing antenatal screening for Down syndrome
- the applicability of the National Health Committee's criteria to assess an antenatal Down syndrome screening programme
- the need for and acceptability of a nationally organised antenatal Down syndrome screening programme.

The Advisory Group comprised members appointed for their particular expertise and/or experience in the following areas:

- biochemical genetics
- consumers of maternity services
- consumers of disability services
- consumers of services for disabled people
- clinical genetics
- cytogenetics
- disabled people
- general medical practice
- health law
- Màori health
- midwifery
- obstetrics and fetal medicine
- paediatrics
- radiology
- National Screening Unit within the Ministry of Health
- Office for Disability Issues within the Ministry of Social Development.

In addition to regular monthly meetings, three subgroups were established to consider specific issues relating to social and ethical considerations, maternal serum screening and nuchal translucency.

Process for developing report

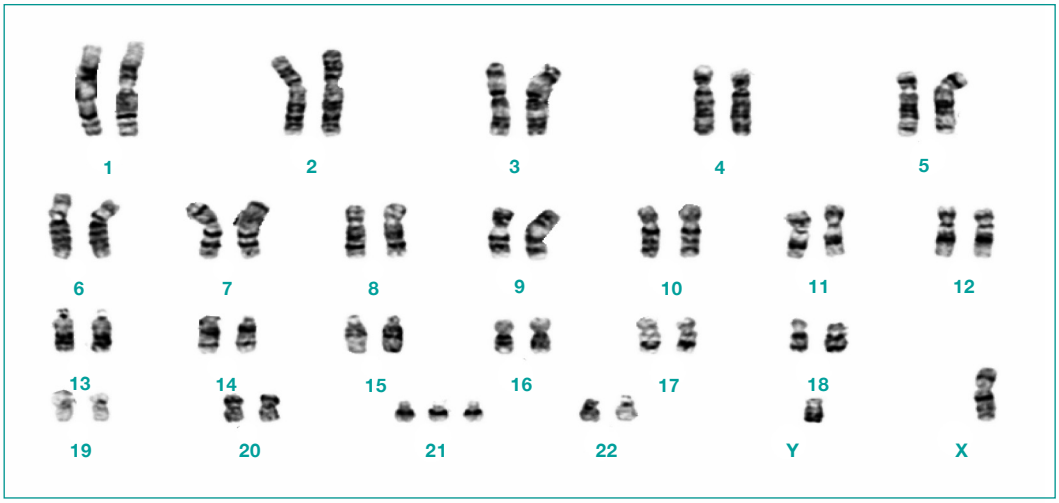
This report is based on the discussions and deliberations of the Advisory Group over an intensive six-month period, which involved significant amounts of reading material and full-day meetings on a monthly basis, as well as subgroup meetings and teleconferences.

In developing its advice, the Advisory Group has considered the medical, technical, social and ethical aspects of antenatal screening for Down syndrome. The diversity of opinion expressed by the Group reflects the wide range of knowledge and experience. Discussion was often challenging due to the close links with other complex issues, including maternity services, screening, and attitudes towards disability. The report reflects the fact that there was not always consensus among members of the Advisory Group and notes where consensus could not be reached.

2.0 What is Down syndrome?

Most human cells have 46 chromosomes (23 pairs).[1] The chromosomes are located in the nucleus of each cell, and contain the genetic material that, in combination with environmental influences, determines an individual person's characteristics. Down syndrome is caused by an extra copy of chromosome 21 inside each of the body's cells (see Figure 1). Instead of a pair there are three chromosomes. The extra genetic material from the extra chromosome gives the characteristics of Down syndrome.[2]

FIGURE 1: KARYOTYPE WITH AN EXTRA CHROMOSOME 21



Characteristics of Down syndrome

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 intellectual capacity (New Zealand Down Syndrome Association, no date).

As a congenital condition, Down syndrome has life-long implications that include intellectual and growth impairment of varying severity and a range of other possible medical complications. For example, around 40 percent of children experience a congenital heart abnormality. The most common problem is an atrial septal defect (hole in the heart). This may be detected on an ultrasound scan later in pregnancy, but is not always able to be seen.[3] Problems with the thyroid gland, which can produce over-activity or under-activity, are found in approximately 20 percent of children with Down syndrome. Problems with the intestinal tract are also common and can lead to surgery after birth. Approximately 6 percent of children will experience digestive problems. People with Down syndrome are now beginning to live longer, with appropriate support in the community and improved medical treatment. About 44 percent will live until the age of 60 years.[4]

[1] Sex cells (sperm and eggs) have half the number of chromosomes, because the chromosomes of the mother and father combine to produce a new individual.
[2] See www.screening.nhs.uk/downs/what.htm.
[3] See www.screening.nhs.uk/downs/what.htm.
[4] See www.screening.nhs.uk/downs/what.htm.

Why is it called Down syndrome?

An English doctor, John Langdon Down, first described Down syndrome in detail in 1866. He noticed the shape of the face, the tendency to get infections and that people with Down syndrome are able to learn and are good at copying others. It took nearly another hundred years to determine the cause of Down syndrome. In 1959 Jerome Lejeune in Paris and Patricia Jacobs in Scotland were studying chromosomes, and they discovered that individuals with Down syndrome have an extra chromosome 21. It was the first time that a disability and a chromosome disorder had been linked (New Zealand Down Syndrome Association no date).

Epidemiology

Down syndrome occurs in approximately 1 in 700 births. This figure is an overall population risk. The likelihood of having a child with Down syndrome increases as a woman gets older, and increases quite steeply for an individual woman from around 37 years of age onwards. Consequently, the older the population, the more prevalent the condition. However, most children with Down syndrome are born to women under 37 years of age, simply because this is when most women have children.

After conceiving a fetus with Down syndrome, women under 39 years have a slightly higher chance of conceiving another fetus with Down syndrome in a future pregnancy compared with other women of the same age. The exact figures vary between different studies, but are usually summarised as being approximately 0.8 percent for women less than 39 years (Warburton et al 2004; Morris and Wald 2005).

Other conditions identified through screening

Antenatal screening for Down syndrome has the potential to identify conditions other than Down syndrome. Some of these conditions are identified by ultrasound scans, while others can be identified using biochemistry or the diagnostic test. In particular:

- biochemistry may indicate trisomy 18, trisomy 13 and open neural tube defects, and some rare metabolic disorders
- first trimester ultrasound can demonstrate gross fetal structural anomalies (eg, skeletal anomalies, brain and neural tube defects, congenital heart defects, and abnormalities of the renal tract, abdominal wall and gastrointestinal system)
- karyotyping can diagnose chromosomal numerical anomalies (trisomies, triploidy, and sex chromosome abnormalities), as well as structural abnormalities, such as inversions and translocations, deletions and duplications (Wald and Leck 2000).

The wide range of conditions potentially able to be identified through screening creates significant implications for the development of educational resources and procedures to ensure informed consent. The Advisory Group considered the possibility of reporting only the results that relate to Down syndrome. However, this would mean that testing would either produce no information about the presence of other conditions and/or women would not be informed of other conditions identified as a result of screening. Following discussions with the Office of the Health and Disability Commissioner, the Advisory Group agreed that this approach is not acceptable.

The Advisory Group recommends that women be informed of the potential for screening to identify conditions other than Down syndrome. It is recommended that women be informed of all test results indicating the presence of a fetal anomaly, except where a woman has indicated that she does not wish to be given the information.

3.0 What is screening?

There are a large number of definitions of screening. The National Health

Committee has developed the following definition of screening:

> *Screening is a health service in which members of a defined population, who either do not necessarily perceive they are at risk of, or are already affected by, a disease or its complications, are asked a question or offered a test, to identify those individuals who are more likely to be helped than harmed by further tests or treatment to reduce the risk of a disease or its complications.* (National Health Committee 2003a: 29)

A definition used in the United Kingdom defines screening as:

> *The systematic application of a test or inquiry, to identify individuals at sufficient risk of a specific disorder to benefit from further investigation or direct preventive action, among persons who have not sought medical attention on account of symptoms of that disorder.* (Wald and Leck 2000: 572)

The difference between screening and diagnosis

There are important differences between screening and diagnosis. Screening does not indicate whether the condition is present. Instead, screening divides people into two groups: a positive result means there is an increased chance of a particular condition being present, while a negative result means there is a decreased chance. The group of people with screen-positive results may benefit from further investigation and may be offered diagnostic tests that can give a definite answer about whether or not the condition is present.

The limitations of screening

Because screening does not give a definite answer it has the potential to provide false-positive results and false-negative results. A false-positive is when an individual is informed they have an increased chance that the condition is present but the condition is not present. A false-negative is when an individual is informed they do not have an increased chance that the condition is present but the condition is present.

The screening pathway

Screening is more than just a test. It is a sequence of events, which is often referred to as the 'screening pathway'. All steps in the screening pathway must be of a high standard to ensure that the screening is safe, and that the benefits outweigh the harms (National Health Committee 2003a). The screening pathway should include:

- provision of information about the condition(s) being screened for
- provision of information about screening
- offer of tests
- screening test(s) performed
- offer of support and counselling to individuals with high-risk results
- offer of a diagnostic test to individuals with high-risk results
- diagnostic tests performed
- offer of support and counselling following the diagnostic test result
- intervention options.

Opportunistic screening versus screening programmes

Screening occurs in one of two ways: either opportunistically or as part of an organised screening programme. The National Health Committee defines opportunistic screening as follows:

> *The key feature that distinguishes opportunistic screening from screening programmes is the lack of a quality process, including routine monitoring and evaluation. Opportunistic screening usually occurs when a person who is presenting to the health system for another reason is asked a question or offered a test in order to detect the presence or confirm the absence of a specific condition. Opportunistic screening may be organised to a greater or lesser degree. However, because there are no attendant quality processes, its safety, effectiveness and cost-effectiveness cannot be assessed or guaranteed.* (National Health Committee 2003a: 7)

Where the screening pathway is complex, a nationally organised screening programme can provide a co-ordinated approach, including:

- clear lines of accountability
- high-quality service provision
- effective monitoring of defined policy and quality standards
- the timely availability and appropriate integration of screening services with diagnostic, support and intervention services
- monitoring and evaluation of the entire screening pathway.

> *In screening programmes, all activities along the screening pathway are planned and co-ordinated. Thus screening programmes have resources committed to the development, implementation, monitoring and evaluation of all aspects of the programme…*
> (National Health Committee 2003a: 7)

4.0 What are the social and ethical issues in screening for Down syndrome?

This section sets out some of the key ethical and social issues. More detailed discussion is provided in the Social and Ethical and Subgroup report (Appendix 2).

The National Health Committee's (2003a) criteria to assess screening programmes require that there be consideration of social and ethical issues.

> *There should be evidence that the complete screening programme (identification and invitation, test, diagnostic procedures, treatment/intervention) is clinically, socially and ethically understood and acceptable to health professionals and the wider public.* (National Health Committee 2003a: 26)

This provides very little guidance as to what is meant by ethical considerations, which will vary depending on the type of screening. For example, ethical considerations involved in offering cancer screening to adults, when the disease may be treatable, are quite different to the ethical considerations involved in offering pregnant women a screening test for disability.

Many of the key ethical and social issues the Advisory Group has identified centre on decisions that must be made. Decisions in relation to such matters as the moral status of the fetus are not easy to make because they raise fundamental questions about what kind of life is worth living. Discussion also included the ethical acceptability of terminating pregnancy, whether there are, or should be, limits to reproductive autonomy, and the subjectivity of any assessment of disability.

In addition, the following decisions need to be made.

- Individuals and families must decide whether to participate in testing, with whom to share the results, and how to act on them.
- Health professionals must decide when to offer testing, how to ensure its quality, how to interpret the results, and to whom they disclose information.
- Society must decide the relative social implications of individual choice and whether it supports participation in a government-funded screening programme to detect Down syndrome.
- The Government must decide how to ensure safe practice occurs, what antenatal tests to fund, and what information and counselling services to provide.

Ethical principles

The most commonly used approach to ethical analysis within medicine (and that used by the Advisory Group when considering ethical issues) is

the Four Principles Approach, which comprises:

- autonomy (self-determination)
- beneficence (doing good)
- non-maleficence (not doing harm)
- justice (fairness).

These principles do not provide easy answers to complex ethical questions, but they are a useful starting point for identifying and articulating ethical issues. Following are some of the fundamental issues discussed by the subgroup, categorised under the four principles.

Autonomy

Reproductive autonomy

Modern maternity care involves allowing women and their partners considerable freedom to make their own choices and decisions with regard to pregnancy and childbirth. Within the Advisory Group there is consensus that reproductive autonomy should not be without limit, however.

Most members believe that reproductive autonomy should extend to the ability to choose whether or not to have antenatal screening for Down syndrome and make decisions concerning continuation or termination of pregnancy when the fetus is affected. Others believe that this is beyond the limits of reproductive autonomy and should not be permitted. A large part of this disagreement centres on the issue of whether Down syndrome constitutes a 'harm' (see below). However, it should be noted that it is possible to hold personal views concerning the nature of Down syndrome but still believe that the reproductive autonomy of others in society should be respected.

A woman's reproductive autonomy can best be facilitated through an antenatal Down syndrome screening programme if:

- screening complies with international best practice
- practitioners are well educated on all aspects of screening
- complete high-quality information on screening and Down syndrome is provided to women
- all aspects of screening and testing are voluntary
- women's choices are unconditionally accepted and supported.

Autonomy of the child

The moral status of the fetus is an ongoing source of debate. There is a wide range of views, from those who consider full moral status is acquired at conception, to those who consider that a newborn baby does not have full moral status. Perhaps the most prevalent view is the gradualist approach, whereby a fetus gradually acquires moral status as pregnancy proceeds. The legal status of the fetus is more clear-cut, with the fetus possessing few, if any, legal rights.

Non-maleficence and beneficence

The most contentious issue in this section relates to whether or not Down syndrome should be considered 'harm'. This issue is complex on a number of levels.

- The clinical features of Down syndrome are variable.
- Assessments of the quality of life of people with impairments are subjective.
- Prospective parents have difficulties imagining/anticipating their reaction to prenatal diagnosis or birth of a child with Down syndrome.
- Different ethnic groups have varied perspectives on fundamental issues such as disability and termination of pregnancy.
- The society into which children with Down syndrome are born now can be considered more positive and accepting of disability than was previously true.

- The debate around the 'lived experience' of people with Down syndrome is closely related to discussion of the two predominant models of disability.
  - The 'social model' considers the lived experience of people with Down syndrome to be the product of societal arrangements that may be more or less disabling. From this point of view, the harms potentially experienced by people with Down syndrome do not arise necessarily from their condition but from the various attitudinal and institutional barriers they encounter.
  - The 'medical model' focuses more on the individual, their bodily impairments and limitations.
- An additional perspective expressed by some members is that both the medical and social models, taken in isolation, are inadequate ways to assess the impact of disability on the lives of individuals and families within society, or to inform public policy.

Justice

The concept of justice involves consideration of issues of equity and fairness, and of the societal impact of decisions. The most significant issues in relation to this principle are the potential for perpetuating or worsening negative attitudes towards people with disabilities, and equity of access to screening services.

- Antenatal screening is viewed by some members as being underpinned by an assumption that the life of a person with a disability has less value than that of an able-bodied person. This raises fundamental questions about what kind of life is worth living, equality of respect, and who has the right to decide. It may also reinforce the stigma associated with such conditions as Down syndrome and devalue the lives of people living with disabilities.
- There is general agreement that the aim of any proposed antenatal Down syndrome screening programme should not be to decrease the incidence of Down syndrome. However, the effect of any screening programme would almost inevitably be to decrease the incidence of Down syndrome. This will effectively mean there is less opportunity for interaction with children and adults who have Down syndrome (although there would be little effect on the overall prevalence of disability), which may influence society's views.
- Equity of access to services is not achieved with the current ad hoc screening, and could only be achieved through introducing a population screening programme.

The pivotal role of ethical debate

Any antenatal screening programme for Down syndrome must be founded on sound ethical premises. The Social and Ethical Subgroup's report (see Appendix 2) highlights some of the areas of ethical controversy underlying antenatal screening. These ethical considerations are fundamental and should be considered before any other requirement in planning a screening programme.

If the Ministry of Health recommends to the Minister of Health that a nationally organised screening programme should be put in place, there will need to be further ethical analysis of implementation issues. Many of the ethical issues associated with antenatal screening relate to how it would be implemented, and the contingencies that would be required. For instance, a government-funded population-screening programme would enhance reproductive autonomy only if prerequisites such as balanced information and unconditional support for all choices were met.

Members of the Social and Ethical Subgroup were unanimous on only one issue: current practice is untenable because it is unsafe. Because the Subgroup was not able to resolve some of the key issues, some members considered that wider societal debate should occur. Others felt that there was little chance of the issues being resolved this way, and that the unsatisfactory state of current screening practices means there is an urgency to improve current practices by introducing an antenatal Down syndrome screening programme.

Inequalities in screening

Inequalities in access to antenatal screening are likely to be determined by existing inequalities in access to maternity care. For example, socioeconomic differences in the use of screening might reflect limits to the ability to access information rather than an exercise of informed choice.

Inequalities are defined as differences in health that are unnecessary, avoidable and unjust. Health inequalities are observed in a number of dimensions, including ethnicity, socioeconomic position, gender and geography. Although these dimensions appear diverse, the existence of inequalities within them all reflects a fundamentally similar process: the unjust distribution of the social determinants of health (eg, education, income and access to health care). These have an impact on health status and health outcomes. Put simply, it is often easiest to reach those who are easy to reach (ie, non-Māori, non-Pacific, non-rural and well-educated people), thereby perpetuating and embedding inequalities.

All forms of screening have the potential to exacerbate inequalities. This is because well-informed, educated individuals who have the financial and social ability to access health services participate in screening, while individuals who already experience financial and social inequalities are less able to participate in screening. This increases existing inequalities.

To assess whether there will be social inequalities in the offer of antenatal Down syndrome screening it is important to assess the potential inequalities that may occur. For example, screening pathways based on women presenting in the first trimester of pregnancy, travel, multiple appointments, and tests that are complex to understand may all work to increase inequalities.

Current practice for antenatal Down syndrome screening contributes to increasing inequalities because some women are offered poor-quality screening tests, while others are not offered any form of screening at all. More women from lower socioeconomic groups do not have an opportunity to participate in screening because they do not register with a lead maternity carer (LMC) until the second trimester of pregnancy, and the Ministry currently only funds a screening test that cannot be performed after 13 weeks six days of pregnancy. Funding second trimester screening tests is one option that could help to address this inequality.

Inequalities of access to antenatal screening for Down syndrome may arise from a number of factors, including:

- age at pregnancy (which is related to the availability of support networks, level of education, and other social factors that may be more problematic than screening)
- timing of diagnosis of pregnancy and first contact with a health practitioner
- access to a health practitioner (level of knowledge)
- provision of information that is able to be easily understood (which is related to language proficiency, ethnicity and level of education)
- ability to access screening services (eg, transport, travel distance, ability to pay for tests)
- partner status
- employment status
- low socioeconomic status
- whether the pregnancy was planned
- the woman's reaction to the pregnancy
- contemplation of abortion
- availability of social support.

Age at pregnancy

Age at pregnancy may influence the level of social support that a woman can access. It may also indicate the woman's level of education and ability to access and understand information on screening options. Māori women have their children much younger than other ethnic groups. While this does not appear to affect their reproductive outcomes, the impact that young motherhood has on their education and social development may have significant social consequences that require separate interventions. Within this context, supportive antenatal care earlier might need to be less pregnancy focused and more geared to the wider health and social needs of Māori women. Also, competing social factors may make antenatal screening tests less important for Māori than for non-Māori.

Timing of confirmation of pregnancy and first contact with a health practitioner

Women who first consult a health practitioner after the first trimester of pregnancy may not have the same opportunity to access screening as women who consult a health practitioner earlier in pregnancy. The number of antenatal visits a woman attends may not necessarily alter the outcome of the pregnancy, but it will determine the antenatal screening pathways available to her.

Access to a health practitioner (level of knowledge)

Many women rely on a health practitioner to provide relevant information, present opportunities for informed decision-making, and offer referral for appropriate tests. Women who have difficulty consulting a health practitioner may find it more difficult to navigate the screening pathway, particularly if it is their first pregnancy.

Provision of information

It is important to recognise the complexity that underlies and will influence the decision-making process. The decisions women make about antenatal screening are often based in the context of their own cultural identities, their family/whānau and their communities.

Many factors can work against informed choice in antenatal screening. These include persuasive pressures on choice through the routine offer of screening tests, lack of balanced information about 'screened for' conditions, lack of health practitioners' time to present information and options, and the difficulties of declining tests.

The decision about whether to participate in antenatal screening should be made on the basis of good information. To make informed decisions, women need to know what conditions the test might detect as well as the implications of negative and positive results. Such information may also be of benefit in preparing women for possibly unexpected outcomes of screening.

All pregnant women should receive information written in plain language, describing available forms of antenatal screening, their advantages and limitations, together with possible risks to the pregnancy and the right to accept or reject what is being offered. It should also include information on the lived experience of Down syndrome and the range of supports available for people with Down syndrome. This information should be available in a range of accessible formats.
All resources for consumers and practitioners about antenatal screening for Down syndrome should be evaluated for their effectiveness in assisting informed decision-making.

Ability to access screening services (eg, transport, travel distance, ability to pay)

If a nationally organised programme for antenatal Down syndrome screening is recommended, it should be designed so that it reduces existing inequalities in access to screening for Māori, Pacific peoples, low socioeconomic groups, and women living rurally.

5.0 Current methods of screening for Down syndrome in New Zealand

Purpose of screening for Down syndrome and other fetal anomalies

The purpose of screening is to provide pregnant women who choose to have the tests with information about whether the fetus is likely to have Down syndrome or other fetal anomaly. This information may help women to make informed decisions and plans about their pregnancy, including:

- whether to have diagnostic testing
- whether to continue with or terminate the pregnancy
- preparing for the birth and life of a child with Down syndrome, or other condition
- planning to give birth at a specialist centre that can provide any immediate neonatal treatment required.

Although this is the purpose of screening, current practice does not achieve this well.

Methods of screening currently used in New Zealand

There is no organised antenatal screening programme for Down syndrome and other anomalies in New Zealand. However, some antenatal screening for Down syndrome and other anomalies does already occur as part of maternity care.

Screening uses 'markers' to calculate a pregnant woman's chance of having a child with Down syndrome. Commonly used markers for Down syndrome and other anomalies include:

- maternal age
- measurable structural anomalies that can be viewed using ultrasound (eg, nuchal translucency)
- measurable biochemical markers in the pregnant woman's blood (eg, alpha-fetoprotein) in the second trimester.

These markers, together with gestational age and maternal age, are used to calculate a pregnant woman's risk of having a child with Down syndrome. They are discussed in greater detail below and are summarised in Table 1.

Maternal age as a screening tool

> *It had been recognised since the 1930s that maternal age was related to the risk of having a pregnancy with Down syndrome … and in 1968 the first antenatal diagnosis was made.* (Stone and Austin 2006: 7)

The use of maternal age as a screening test for Down syndrome is likely to have been used in New Zealand since the 1970s. It involves offering women aged over a particular number of years a diagnostic test, although the age at which women are offered the diagnostic test may depend on the availability of resources. Currently, diagnostic testing in New Zealand is publicly funded for women 35 years or older at the date of conception.

Maternal-age screening is a crude screening test. For one thing, the sensitivity and specificity of the test vary according to the population being screened. Populations with more older women giving birth have a higher sensitivity but lower specificity.

Most international literature quotes a sensitivity of 30 percent for a 5 percent false-positive rate. This means that using maternal age alone will only identify 30 percent of fetuses with Down syndrome. The odds of being affected given a positive result are 1 in 130 (ie, 129 women given a screen-positive result will not be carrying a fetus with Down syndrome) (Wald and Leck 2000).

In New Zealand in 2003, 19.8 percent of women giving birth were aged 35 years or older (Ministry of Health 2006), compared to 11.2 percent in the United Kingdom. (Morris et al 2005). This means that in New Zealand screening on the basis of advanced maternal age gives a high detection rate (closer to 60 percent) and a higher false positive rate (closer to 20 percent).

Internationally, methods of antenatal screening for Down syndrome have been enhanced significantly over the last 15 years, with the development of biochemical and ultrasound markers.

The development of ultrasound screening

Ultrasound has been shown to be useful as a screening tool in the first trimester of pregnancy. Between 11 weeks and 13 weeks six days of pregnancy an increase in the fluid-filled area at the back of the fetal neck, between the spine and the skin, has been shown to be associated with Down syndrome. The depth of fluid in this space can be measured during an ultrasound scan and is referred to as an 'NT measurement' (nuchal translucency measurement). Computer software can provide an individualised assessment for each woman of the chance that the fetus has Down syndrome, based on the NT measurement and the woman's age.

Many women have ultrasound screening because it provides them with an early 'picture' of their 'baby'. Anecdotal evidence suggests that many of these women are not aware that this ultrasound is actually a screening test that may identify fetal anomalies. This raises significant issues relating to informed consent to screening.

The accuracy of the NT measurement is dependent on the skill of the operator and the suitability of the ultrasound machine used. When used by a skilled operator, NT has a sensitivity of 77 percent (ie, 77 percent of women who are carrying a fetus with Down syndrome will be given a screen-positive result) and a false-positive rate of 5 percent (Snijders et al 1998); this report focuses on well-trained operators. Two large prospective studies (SURUSS and FASTER) both showed NT to have a sensitivity of about 60 percent for a 5 percent false-positive rate. The odds of being affected given a screen-positive result are likely to be between 1 in 52 and 1 in 67 for a 5 percent false-positive rate (Wald et al 2003; Malone et al 2005).

Ultrasound is also commonly used between 18 and 20 weeks of pregnancy to screen for obvious structural fetal anomalies. This is known as the 'fetal anomaly scan' or 'fetal morphology scan'. Although this scan may identify fetal anomalies not previously found, it is not an effective antenatal screening tool for Down syndrome (Wald and Leck 2000). International evidence in this area continues to evolve.

The development of biochemical screening

Biochemical screening involves taking a sample of blood from pregnant women at specific gestation periods and analysing the levels of particular markers in the serum. This is also referred to as maternal serum screening.

Initially, the level of one serum marker, alpha-fetoprotein (AFP) was associated with the presence of open neural tube defects. In 1984 Merkatz and his colleagues reported an association between pregnancies and aneuploidies (including Down syndrome) and low serum alpha-fetoprotein levels (Wald and Leck 2000). Since then, other markers found in maternal serum have been shown to indicate the presence of Down syndrome. In 1986 levels of maternal serum human chorionic gonadatrophin (hCG) were shown to be, on average, higher in Down syndrome pregnancies than in unaffected pregnancies. At about the same time the levels of unconjugated oestriol were found to be about 25 percent lower in affected pregnancies (Wald and Leck 2000).

In 1988 a new method of screening was reported in which these three biochemical markers (AFP, hCG and unconjugated oestriol) were used, together with maternal age, in a single antenatal test for Down syndrome. This became known as the triple test (Wald and Leck 2000). Biochemical screening has developed significantly since the introduction of the triple test. In particular, additional maternal serum markers have been developed, and it is now possible to offer a first trimester maternal serum screening test.

In New Zealand a quadruple test is available, using four maternal serum markers (AFP, free beta hCG, free alpha hCG and unconjugated oestriol). In the SURUSS study the quadruple test used for comparison included inhibin A rather than free alpha hCG as the fourth analyte. Using these analytes, the researchers demonstrated that the quadruple tests had a sensitivity of about 83 percent for a 5 percent false-positive rate. This means that about 83 percent of fetuses with Down syndrome will be identified. The odds of being affected given a positive result are 1 in 48 (ie, 47 of the women given a screen-positive result will not be carrying a fetus with Down syndrome) (Wald et al 2004).

Combining ultrasound and biochemical markers

Combining ultrasound and biochemical markers has been shown to improve the sensitivity and specificity of the test (ie, the test will identify more cases of Down syndrome and is better at accurately identifying women who are not carrying a fetus with Down syndrome) (Wald et al 2003; Malone et al 2005). However, the technique of combining ultrasound and biochemical markers has not been systematically introduced into New Zealand.

A very small number of combined tests are performed each year for women who specifically request it and pay the relevant costs. The only form of combined testing currently performed in New Zealand includes an NT measurement and a quadruple test to provide a single assessment of the chance the fetus has Down syndrome. International literature shows that there are better combinations of markers (such as the integrated test) that provide higher quality results. This is discussed further in section 7.

Diagnostic tests currently used in New Zealand

Regardless of the type of screening used (maternal age, NT or biochemistry), women with a screen-positive result are offered a diagnostic test. Diagnostic testing involves taking a sample by an invasive procedure.

- Chorionic villus sampling (CVS) can be performed from 11 weeks of pregnancy. Under ultrasound guidance a fine needle is inserted through the abdomen into the uterus and a small sample of tissue is taken from the placenta. Very occasionally this procedure is performed by inserting a fine needle through the cervix under ultrasound guidance.
- Amniocentesis can be performed from 15 weeks of pregnancy. Under ultrasound guidance a fine needle is inserted through the abdomen and uterus into the amniotic cavity and a 10–20 ml sample of amniotic fluid (the fluid surrounding the fetus) is taken, depending on the gestation.

Samples taken via CVS or amniocentesis are cultured in the laboratory for 10–14 days. Chromosome preparations are then obtained and analysis performed using a microscope. This includes an examination of chromosome number and structure (karyotyping). Results obtained provide a definite diagnosis of Down syndrome. However, this analysis may also find other chromosomal anomalies, such as trisomy 13 or trisomy 18. Although these invasive diagnostic tests can confirm if the fetus has Down syndrome, both procedures carry a risk of spontaneous fetal loss (0.5 to 2 percent).

Very rarely, a second diagnostic test is requested (0.01 percent of cases), usually following culture failure or to confirm an unusual result.

New Zealand practice compared with international best practice

Antenatal screening for Down syndrome in New Zealand has not kept pace with international best practice. Both the United Kingdom (UK) and the United States of America (USA) have undertaken large trials that show that:

- screening by maternal age alone is the least safe method of screening
- screening by maternal age combined with NT scanning is the second-least safe method, especially in the absence of standards and monitoring (Wald et al 2003; Malone et al 2005).

The safest and most effective combinations of markers are not available, funded or provided in New Zealand. Studies have shown that screening tests, which involve combinations of first and second trimester markers, can reduce the number of women assessed to have a fetus with an increased chance of Down syndrome (Wald et al 2003; Malone, Canick et al 2005).
This is discussed further in the next section.

At the same time, the screening tests that are currently available are not always offered, or provided in a way that meets best practice, particularly in relation to practitioner competence, quality assurance, monitoring and evaluation. There is significant variation in practice across New Zealand, especially in relation to ultrasound screening. The types of tests offered or available to women vary, depending on what is available locally, the knowledge of the practitioner, the knowledge of the woman, and the woman's ability to pay for some tests. As a result, some women are:

- not offered any form of screening or diagnostic test at all
- offered invasive diagnostic procedures on the basis of their age alone
- offered an NT ultrasound scan, with the chance that it will be provided by an unaccredited practitioner using unsuitable equipment.

Table 1: Summary of methods of antenatal screening for Down syndrome currently used in New Zealand

METHOD	GESTATION PERFORMED	SENSITIVITY (DETECTION RATE)	SPECIFICITY (FALSE-POSITIVE RATE) (SURUSS)	AVAILABILITY	FUNDING
Maternal age, followed by an offer of diagnostic procedure	Up until 20 weeks of pregnancy	30% sensitivity, depending on the population. This is likely to be closer to 60% for New Zealand.	5–15% and higher depending on the population. This is likely to be closer to 20% for New Zealand.	No limit on availability. Diagnostic procedures are offered to women over the age of 35.	GP and LMC consultations are funded as part of general maternity care, under the Section 88 Maternity Services Notice. Diagnostic procedures are publicly funded, where they are utilised, for women over 35 years of age under the Secondary Maternity Service Specification (see also diagnostic tests).
NT	Between 11 weeks and 13 weeks 6 days of pregnancy	85%	20–25%	Approximately 115 practitioners are accredited by the Fetal Medicine Foundation. There are likely to be unaccredited practitioners performing the scan. It is unclear which areas of New Zealand are not able to access NT performed by an accredited practitioner. There are likely to be difficulties in rural areas. There are also likely to be some access issues as a result of co-payments.	Ultrasound scans, including NT, are publicly funded at $80 per scan under the Section 88 Maternity Services Notice. Most women are required to make a co-payment, which can range from $20 to - $95.
Biochemistry (using AFP, free beta hCG, free alpha hCG and unconjugated oestriol)	Between 14 and 18 weeks of pregnancy	85%	9.3%	Only LabPlus, at Auckland DHB, currently provides this test. It is available to any woman in New Zealand who is aware that a second trimester serum test is available, who chooses to have it, and who is able to pay the charges.	There is no public funding. It costs $75 per test, privately funded. Collection and shipping charges are often added.
NT + biochemistry using AFP, free beta hCG, free alpha hCG and unconjugated oestriol	NT: between 11 weeks and 13 weeks 6 days of pregnancy, combined with the results of maternal serum screening performed between 14 and 16 weeks of pregnancy.	85%	6.1%	Approximately 1100 combined tests are performed each year. They are available to any woman in New Zealand who is aware that a combined test is available, and who chooses to have their NT result combined with the results of the serum test, and who is able to pay the charges.	Ultrasound scans, including NT, are publicly funded at $80 per scan under the Section 88 maternity Services Notice. Most women are required to make a co payment, which can range from $20 to $95. Maternal serum screening receives no public funding. It costs $75 per test, privately funded. Collection and shipping charges are often added
DIAGNOSTIC TESTS					
Sample taken by either CVS or amniocentesis and analysed by karyotyping	CVS is performed between 11 and 14 weeks of pregnancy. Amniocentesis is performed typically between 15 and 17 weeks of pregnancy.	Karyotyping has a 99.9% detection rate. Rarely a second diagnostic test is requested (0.01%), usually following culture failure or to confirm an unusual result.		Amniocentesis is available in most places where there is access to ultrasound and an obstetrician. However, CVS is presently restricted to only 6 specialist centres in New Zealand. Analysis of samples by karyotyping is performed by four laboratories across New Zealand.	Procedures receive public funding if the woman is over 35 years of age or there are clinical indications. Procedures are currently publicly funded, per procedure, at: • CVS: $368 • amniocentesis: $368 • laboratory analysis: $295. This funding does not reflect the true costs of these services. The costs of laboratory analysis are closer to $500 per procedure.

6.0 Problems with current Down syndrome screening in New Zealand

As discussed in section 3 of this report, screening is more than just a test. Screening involves a pathway of care and intervention, including diagnostic tests, counselling and support. At present in New Zealand there are difficulties along the entire screening pathway. Some of these difficulties are technical, while others are social and ethical.

Assessment of antenatal screening for Down syndrome

The Stone and Austin Report (2006) identified the following problems.

- Currently, antenatal screening for Down syndrome is ad hoc, based mostly on ultrasound scanning or maternal age alone, and does not reflect the best evidence or best practice internationally.
- There is a lack of clarity regarding exactly what is being screened for.
- Quality and safety requirements are not being met by the current approaches to Down syndrome screening in New Zealand.
- Health practitioners surveyed hold a unanimous view that the current system of antenatal screening for Down syndrome is not best practice and is not sustainable.
- Almost half of practitioners currently offer screening to women either based on their age or on having a previous pregnancy with an abnormality.
- There is little support for NT screening alone.
- Health practitioners need to have increased knowledge about the purposes and methods of screening (Stone and Austin 2006).

The offer of screening

The offer of antenatal screening for Down syndrome involves a number of social and ethical difficulties, including:

- whether offering antenatal screening suggests supporting or recommending that women have the tests
- whether antenatal screening for Down syndrome should be offered to all pregnant women
- whether the same form of screening test should be offered to all pregnant women across New Zealand
- whether the offer of screening is made with an assumption that a women will terminate an affected pregnancy.

Currently, the offer of screening is inconsistent across New Zealand. Some health practitioners offer screening to all pregnant women, some offer it selectively and others do not offer it at all. It is not known how many women are offered antenatal screening for Down syndrome each year because the data is not collected or monitored. Stone and Austin (2006) found that:

- around 50 percent of practitioners surveyed offer screening to all women and 'around 46 percent of practitioners offer screening on a selection basis, generally based on maternal age or previous abnormality'
- GPs surveyed were less likely to offer screening than other practitioners (less than one-third offer screening to all).

Information to support decision-making

The decision about whether to participate in screening should be an informed one, based on relevant information and supported by effective communication. Stone and Austin (2006) found that the practitioners surveyed lacked an understanding of antenatal screening for Down syndrome and identified the need for educating practitioners.

> *The one major topic that the survey did identify … was the issues about health practitioner knowledge about screening. It is clear from the results and individual responses in the appendices that there is a need for increased knowledge about the purposes and processes of screening as much as the actual tests. Even the most widely used test (NT) was timed wrongly by 25 percent of practitioners.* (page 24)

> *High quality information about Down syndrome and screening for Down syndrome and other chromosomal abnormalities is required for women and health professionals. It was highlighted that this information needs to be complete, covering all options including support for women who choose to keep a child or consider adoption.* (page 11)

Information to support informed decision-making should be provided to women and their families by health practitioners and supported by written information. Some women and their families may need to have discussions with their health practitioner spanning a number of consultations and meetings. It is likely that many practitioners will require further support and training so that they are better able to communicate effectively about Down syndrome, other fetal anomaly and antenatal screening.

To facilitate this, one organisation needs to be given a clear mandate to provide easily written information, in a range of accessible formats, to practitioners and consumers. It should include information about:

- Down syndrome
- the right to refuse screening
- the potential for other conditions to be found during screening
- any significant medical, social or financial implications of screening
- the difference between a screening test and a diagnostic test
- the screening tests, including risks and benefits
- a description of the entire screening pathway, including the offer of counselling and support
- where to go for further information.

Informed consent to screening

New Zealanders have a legal right to give informed consent to health care services, including antenatal screening for Down syndrome. Right 7(1) of the Code of Health and Disability Services Consumers' Rights (the Code) states:

> *Services may be provided to a consumer only if that consumer makes an informed choice and gives informed consent, except where any enactment, or the common law, or any other provision of the Code provides otherwise.*

There is anecdotal evidence that currently some pregnant women have antenatal screening for Down syndrome without understanding the nature and implications of the tests. The potential for screening to occur without women having given informed consent is exacerbated by the lack of understanding among practitioners, and the absence of appropriate information for consumers.

Problems with the types of screening currently performed

As discussed on section 5, the safest and most effective tests are not currently available in New Zealand. Many studies have shown that there are screening tests, using combinations of biochemical and ultrasound markers in the first and second trimesters of pregnancy, that reduce the number of women considered to be high risk. At the same time, the tests that are available are not always provided in a way that meets best practice, particularly in relation to practitioner competence, quality assurance, monitoring and evaluation (Chang 2006).

It appears that the type of screening women are offered depends on the knowledge and attitudes of the individual practitioner, modified by the knowledge and attitudes of the individual woman. There is no national policy on antenatal screening or guidelines for the types of tests that should be offered and performed.

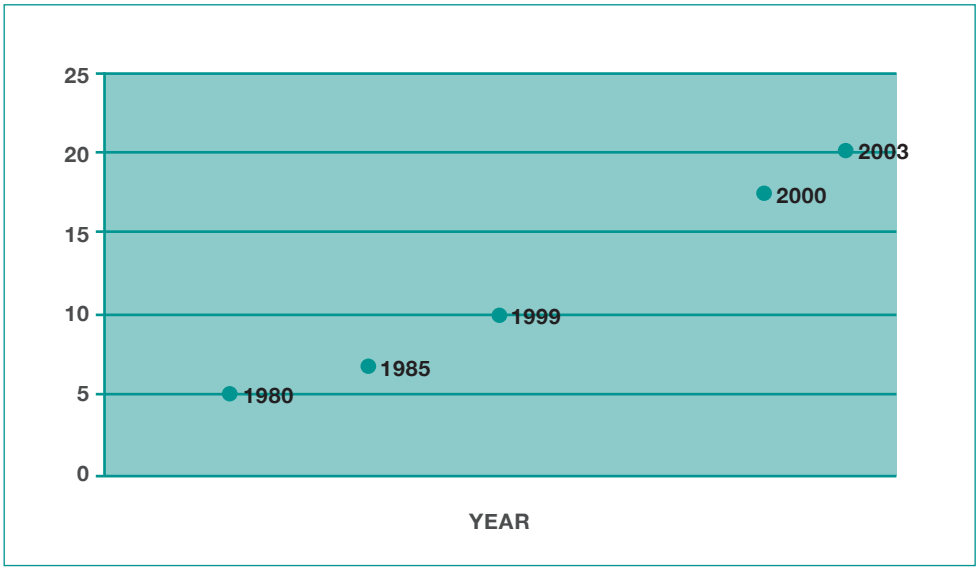
Problems with maternal age as a screening tool

Maternal age as a screening test (without additional markers) is problematic for the following reasons.

- Maternal age is a crude screening test, with poor sensitivity and specificity. Screening based on maternal age alone has a sensitivity of 30 percent for a 5 percent false-positive rate. This means it will find 30 percent of fetuses with Down syndrome. The odds of being affected given a positive result are 1 in 130 (ie, 129 women given a screen-positive result will not be carrying a fetus with Down syndrome) (Wald and Leck 2000).
- There is better technology available for risk assessment than maternal age. Women offered diagnostic testing on the basis of their age may be assessed as having a lower risk of carrying a fetus with Down syndrome than their age-related risk. Screening on the basis of maternal age results in women being referred to invasive procedures for diagnostic tests, which may be unnecessary; unnecessary referral for invasive procedures increases the risk of fetal loss (Berkowitz et al 2006).
- Younger women are automatically screen-negative and are denied access to diagnostic tests, but the use of additional markers may reveal they are at significantly increased risk. Maternal age screening means that women who are less than 35 years of age are advised they have a low risk, and are not offered other forms of screening or diagnostic tests. These women are denied an opportunity to receive more accurate information about their pregnancy.
- Although older women are more likely to have a child with Down syndrome, more children with Down syndrome are born to younger women due to the fact that there are more births in this age bracket.

- Patterns in New Zealand suggest that women are increasingly likely to have children after the age of 35 years (see Figure 2), so an increasing number of women are being offered diagnostic testing on the basis of their age (Ministry of Health 2006). In New Zealand in 2003, 19.8 percent of women giving birth were aged 35 years or older (Ministry of Health 2006), compared to 11.2 percent in the United Kingdom.

FIGURE 2: PERCENTAGE OF WOMEN OVER 35 GIVING BIRTH, 1980–2003



Source: New Zealand Health Information Service maternity data

Problems with ultrasounds markers (NT)

A significant number of women (approximately 25,000 each year) have NT measurement performed as part of their first trimester ultrasound scans (11 weeks to 13 weeks 6 days gestation). The current use of NT is problematic for the following reasons.

- There is no requirement for practitioners performing NT scans to meet specific standards, perform a minimum number of scans, be accredited, or use suitable equipment (Snijders et al 2002). Highly trained and experienced sonographers are more accurate at measuring the NT and produce a more accurate assessment. It is likely that many of the NT measurements performed in New Zealand are not of sufficient quality.
- Data on the quality of NT in New Zealand is not collected and evaluated at either a regional or a national level.
- There are likely to be some areas in New Zealand where women are either not able to access an NT ultrasound scan, or are not able to access one performed by an accredited practitioner.
- The funding for NT ultrasound scans is provided as part of funding for general ultrasound scans during pregnancy. The funding does not cover the costs involved in providing high-quality screening by accredited practitioners. As a result, many services charge a co-payment, which is paid directly to the service by the consumer. The existence of co-payments makes it difficult for some women to access NT ultrasound scans.
- Anecdotal evidence suggests that some women are having an NT measurement performed when they have not given informed consent and have not been informed it would be done.

There are also concerns that NT scanning alone is not the most accurate method of screening for Down syndrome, even when performed by highly trained and experienced practitioners. Even when used by the most competent practitioners, NT has a sensitivity of 85 percent for a 25 percent false-positive rate. The odds of being affected given a screen-positive result are 1 in 67 (ie, 66 women given a screen-positive result will not be carrying a fetus with Down syndrome) (Wald et al 2003; Malone et al 2005). Stone and Austin (2006) note that:

> *When practitioners were presented with information on test performance there was little support for NT scanning alone as the screen leading to invasive testing.*

Large trials carried out in the UK and the USA show the combination of NT scanning followed by amniocentesis to be one of the least safe methods (after maternal age alone), especially in the absence of standards and monitoring. Other countries combine NT scans with maternal serum markers to provide more accurate assessments. Combining NT scans with serum screening to provide better assessments is further discussed in section 7.

Problems with biochemical screening (maternal serum screening)

A small number of women (approximately 20 each week) pay privately for second

trimester maternal serum screening. Current maternal serum screening is

problematic because:

- very little information is provided to practitioners and consumers about the availability of second trimester maternal serum screening in New Zealand
- many practitioners and consumers are unaware that it is available
- some women do not have a dating scan before serum screening, so the assessments are not necessarily based on the correct gestation
- the most effective markers are not included in the test (ie, inhibin A), and one marker that is no longer used in other countries is still included (free alpha hCG)
- there is no public funding for maternal serum screening: 'Failure to fund second trimester screening is disadvantaging a group of women who book late (after the first trimester of pregnancy)' (Stone and Austin 2006).

Stone and Austin (2006) recommend that second trimester maternal serum markers be funded, promoted and offered as well as NT scanning as an interim measure while other components of a programme are being developed.

Absence of the most effective combinations of screening tests

Studies have shown that the use of combined ultrasound markers and maternal serum screening markers can substantially increase the sensitivity of screening for Down syndrome, and further reduce the false-positive rates by combining first and second trimester screening in a variety of different ways (Wald et al 2003; Malone et al 2005). These options for screening tests are discussed further in section 7.

Access to suitable counselling and support services

Some specialised counselling and support services are available in New Zealand. However, it is likely that a significant number of women are not offered a referral for suitable counselling and support services after receiving a positive result from a screening test and/or a positive result from a diagnostic test. This is due to limited clinical genetic services and a lack of awareness of the services.

As with all screening during pregnancy, most women participate in order to be reassured about their pregnancies, rather than with any expectation that a fetal anomaly will be identified. The diagnostic tests present potential risks for mother and fetus, requiring women to weigh up the different risks associated with different decisions.

Health practitioners who are not specialised in counselling can provide initial support and discussion. While some practitioners have experience in discussing positive results with women, it is likely that many practitioners in New Zealand would require additional support and training so that they feel comfortable providing this kind of support and discussion. Best practice would require health practitioners to offer referral for non-directive counselling services to ensure that women with positive results are supported in their decision-making.

More specialised counselling by genetic specialists or counsellors should focus on helping the parents to interpret the screening results if a high risk is indicated, and helping them to decide whether to proceed to prenatal diagnosis (Agence d'Evaluation des Technologies et des Modes d'Intervention en Sante 2001). Following a prenatal diagnosis, the counsellor should provide non-directive support and information to assist the woman and her family to make the decision about whether to continue the pregnancy. After an appropriate period of time, there should also be discussion about the risks associated with future pregnancies.

Lack of monitoring and evaluation

There is currently no monitoring or evaluation of antenatal screening for Down syndrome or other fetal anomaly on a regional or national basis. It is currently difficult to monitor and evaluate antenatal screening for Down syndrome due to:

- the large number of practitioners and services involved in offering and providing screening (ie, general practitioners, midwives, sonographers, radiologists, obstetricians, biochemists, clinical geneticists and genetic counsellors)
- the lack of a robust system of data collected to support monitoring and evaluation
- the lack of a clear set of standards as a benchmark against which to evaluate the screening.

The Advisory Group found the lack of access to accurate and reliable maternity data to be a significant obstacle to its work. It is highly likely that practitioners offering and/or performing screening are similarly unable to access accurate and reliable data about the use and outcomes of screening and diagnostic tests in New Zealand.

7.0 Improving the quality of screening

International studies have shown that there is no justification for using NT alone (with or without maternal age) as an antenatal screening test for Down syndrome and other fetal anomaly. NT should be combined with maternal serum screening to identify the highest number of fetuses with Down syndrome for the lowest false-positive rate, either as first trimester combined screening or as integrated testing (Wald et al 2003; Malone et al 2005).

Many studies have shown that screening tests that involve combinations of ultrasound and biochemical markers can reduce the number of women considered to be high-risk and/or improve sensitivity. Combining the results of different tests to provide a single assessment can:

- make the tests more sensitive (better detection rates)
- make the tests more specific (fewer false-positive results).

Balancing sensitivity and specificity

The question of the best form of screening, if any, for New Zealand has ethical and social implications, as well as technical considerations. There is always a trade-off between the sensitivity (detection rate) and the specificity (false-positive rate). A test with greater sensitivity may result in a greater number of false-positive results. At the same time, choosing a test with lower sensitivity in order to reduce the number of false-positive results may result in fewer fetuses with Down syndrome being identified.

When it is very important not to miss a diagnosis, a test with high sensitivity is chosen. When it is very important not to create false-positives, because of the risks associated with the diagnostic tests, greater emphasis should be placed on a test with higher specificity. Clearly, the balancing of these two outcomes requires consideration of the views of consumers, and wider social and ethical perspectives (Marteau et al 1992).

Options for better tests

The combinations of tests shown to have the highest sensitivity and specificity are described below. These tests are considered to be safer than the tests currently performed in New Zealand, because they provide more accurate risk assessment. These more accurate assessments can help to reduce the proportion of screened women offered a diagnostic test, which thereby reduces the risk of fetal loss.

For each of these combinations of screening tests, the individual results of each of the markers will not be provided to the women (ie, the woman will not receive a result from the serum test and a separate result from the NT). The results of the tests are combined to provide a single assessment of the likelihood that the fetus has Down syndrome. Most of these options span two trimesters of pregnancy, and require women to wait for a result following the second trimester maternal serum test.

International evidence

Two recent trials (SURUSS and FASTER) have compared the performance of different screening tests, and have provided modelled data on the sensitivity and specificity of tests. The trials produced remarkably similar results, describing best practice at least from a theoretical view (Stone and Austin 2006).

The UK trial (SURUSS) was a prospective study of 47,053 singleton pregnancies (including 101 pregnancies with Down syndrome) based on women attending 25 maternity centres (24 in the UK and one in Austria). It provides the largest data set on women seen in both the first and second trimester of pregnancy, without planned intervention in the first trimester. The SURUSS results show that the performance of screening tests in the first or second trimesters of pregnancy alone is much less effective than integrating screening measurements from both trimesters into a single test (Wald et al 2003).

The US trial (FASTER) was a study of 33,557 pregnancies in the USA. The researchers compared combinations of NT with first and second trimester maternal serum screening. The FASTER trial was similar to SURUSS in examining women prospectively after recruitment in the first trimester. First trimester results were not revealed until completion of the second trimester screening.

However, women whose fetuses had a clear abnormality were offered diagnostic testing and exited from the screening pathway. As with the SURUSS trial, the fully integrated test performed best (Malone et al 2005).

Table 2: A comparison of screening tests, derived from the main results from SURUSS

TEST*	MEASUREMENTS	NUMBER OF MARKERS PLUS MATERNAL AGE	FALSE POSITIVE RATE FOR 85% DETECTION RATE	UNAFFECTED WOMEN FOR CVS OR AMNIO PER 100,000 WOMEN SCREENED	NO. OF DOWN SYNDROME DIAGNOSED PER UNAFFECTED FETUSES LOST**
Integrated test	• First trimester serum (PAPP-A) at 10 weeks • Nuchal translucency • Second trimester serum (AFP, uE3, free ß-hCG, and inhibin A) between 14 and 20 weeks	6	1.2%	1200	19.2
Serum integrated test	• First trimester serum (PAPP-A) at 10 weeks • Second trimester serum (AFP, uE3, free ß-hCG, and inhibin A) between 14 and 20 weeks	5	2.7%	2700	9.1
Second trimester serum (quadruple test) with a dating scan	• Maternal serum screening at 14–20 weeks (AFP, uE3, free ß-hCG, inhibin-A)	4	6.2%	6200	3.8
First trimester combined test	• First trimester maternal serum test (free ß-hCG and PAPP-A) at 10 weeks combined with nuchal translucency	3	6.1%	6100	3.9
Nuchal translucency	• Nuchal translucency at 11–13 weeks +6 days	1	20–25%	20,000– 25,000	0.07

* All tests include maternal age.
** Assuming an 80 percent acceptance rate of amniocentesis or chorionic villus sampling and 0.9 percent loss rate. Unfortunately SURUSS does not provide data on the number of Down syndrome diagnoses made per fetus lost (both affected and unaffected).

Reasons to use tests other than integrated screening

Although SURUSS and FASTER recommend the integrated test as being the most specific and sensitive, they acknowledge that there are situations where other forms of combined screening may be more appropriate, as follows.

- Not all women would have contacted a heath practitioner in the first trimester of pregnancy. Women who do not contact a practitioner until the second trimester of pregnancy should be offered second trimester maternal serum screening (quadruple test) with a dating scan.
- High-quality NT will not be available in all areas. Where women are unable to access high-quality NT screening they should be offered serum integrated screening with a dating scan.
- Some women will want to complete screening in the first trimester (Spencer and Aitken 2004; de Graaf et al 2002). Women who do not choose to have the fully integrated screening test should be offered the first trimester combined test, with the dating of the pregnancy occurring at the same time as the NT screening.

The studies suggest that the following combinations of tests are the most effective and safest methods of screening for particular situations.

Table 3: Best practice screening methods

SITUATION	BEST TEST
Overall most effective test	Integrated
If NT measurements are not available	Serum integrated
For women who want to finish screening in the first trimester	Combined first trimester
For women who do not consult a practitioner until the second trimester of pregnancy	Maternal serum screening (quadruple test)

Stone and Austin (2006) note that:

> *Choosing the best prenatal screening protocol is clearly not a simple matter. Simply put, there is no right answer and it will remain important to continue to carry out intervention studies and assess programmes repeatedly. The appeal of early provision of screening results and consequent offer of diagnostic testing needs to be tempered by the patient considerations … in addition to the problem of having screening available to women who present at non-optimal times with respect to test performance.* (page 7)

8.0 Options for action

The Advisory Group discussed a wide range of options for action. This section of the report includes all options that were considered and debated. Some of these options have been dismissed by the Group, due either to not being feasible and/or not addressing the safety and quality issues relating to current screening practice. They are included in the report for the sake of completeness.

Taking the social, ethical, and technical considerations into account, the Advisory Group developed three broad options for action in New Zealand:

1. Do nothing and allow the currently unsafe, opportunistic screening to continue.
2. Stop some, or all, elements of screening.
3. Continue to offer screening, and take steps to improve the quality and safety of screening tests, including within a national screening programme.

Within each of these options there is a range of sub-options. A number of the sub- options involve only minimal intervention, which would address a number of the current problems with screening for anomalies including Down syndrome but would not address the entire problem. Other sub-options would require significant intervention and would address a number of the current problems.

All options, and the potential sub-options, were discussed by the Advisory Group and the impact of each is outlined in Table 4.

Option A: Do nothing

One option is to 'do nothing" to address the current problems relating to antenatal screening for fetal anomalies, including Down syndrome. The current opportunistic screening would continue. A recommendation to do nothing suggests that the current problem is either not significant enough to warrant intervention, or that the costs of intervening outweigh the benefits of intervening. This could be combined with an action to direct additional resources to disability support services. In summary, the two sub-options are:

A1. Do nothing to address the current problems relating to antenatal screening for Down syndrome and other fetal anomalies

A2. Do nothing to address the current problems relating to antenatal screening for Down syndrome and other fetal anomalies, but direct additional resources to disability support services and measures to ensure social inclusion.

Option B: Stop some, or all, elements of antenatal screening for Down syndrome

An alternative option is to stop some, or all, elements of antenatal screening for Down syndrome. There are a number of sub-options here, including:

B1. Prohibit public funding being used to support antenatal screening for Down syndrome and other fetal anomalies

B2. Prohibit public funding being used to support antenatal screening for Down syndrome and other fetal anomalies, and direct additional resources to disability support services and measures to increase social inclusion

B3. Prohibit all forms of antenatal screening for Down syndrome (whether publicly or privately funded)

B4 Prohibit all forms of antenatal screening for Down syndrome (whether publicly or privately funded), and direct additional resources to disability support services and measures to ensure social inclusion.

Option C: Improve the current situation

There are a number of sub-options for improving the quality and safety of antenatal screening for Down syndrome. A number of these sub-options would require only a minimal level of intervention, while others would require significant intervention.

Options for minimal intervention, which could address some of the problems, include:

C1. Provide education for practitioners about antenatal screening for Down syndrome

C2. Develop information resources for practitioners and consumers so that high-quality, consistent information is available

C3. Direct additional resources to disability support services and measures to ensure social inclusion.

C4. Improve the quality of screening using ultrasound markers (ie, NT)

C5. Introduce public funding for a second trimester maternal serum screening test

C6. Introduce a publicly funded first trimester maternal serum screening test.

The sub-options for minimal intervention could be combined to create additional sub- options. For example, all five of the options described above could be combined to provide a fully integrated screening pathway that includes education for health practitioners and information resources for consumers. While these interventions are minimal when introduced individually, if implemented together the total intervention is likely to be significant.

Options for significant intervention include:

C7. Implement all of the sub-options outlined above in C1 to C6, and implement these in such a way that screening is still provided opportunistically rather than as part of a nationally organised screening programme

C8. Implement all of the sub-options outlined above in C1 to C6, and implement these in the form of a nationally organised screening programme.

These options are discussed below in Table 4.

Table 4: Analysis of options for screening

A: DO NOTHING	
	Option A1: Do nothing to address the current problems relating to antenatal screening for Down syndrome and other fetal anomalies
Level of intervention	No intervention
Extent to which it will address the current problems	This would not address any of the current problems with antenatal Down syndrome screening.
Technical considerations	does not meet best practice and does not make the best use of available technology to provide high-quality and safe screening.
Ethical principles	*Reproductive autonomy* Current screening: • is not offered to all pregnant women • can be offered by practitioners who lack understanding of the screening tests and their implications • is not accompanied by high-quality written information • is not always based on informed consent • is not always followed by support and counselling, where necessary. *Non-maleficence* Current screening: • is not the safest screening test available • results in a significant number of women being given false-positive results • results in unnecessary referral for invasive procedures • increases the risk of fetal loss. *Justice* Current screening: • is not offered to all pregnant women • is not able to be accessed by all pregnant women (for geographic or financial reasons).
Risks	• Pregnant women may be unnecessarily harmed as a result of poor-quality screening. • Practitioners may be subject to disciplinary action as a result of providing poor-quality screening that is known not to meet best practice. • Providers of screening services (such as laboratories, radiology services, District Health Boards) may be similarly held to account as a result of providing poor-quality screening that is known not to meet best practice. • The Ministry of Health may be held to account for publicly funding a screening test that is known not to meet best practice. There are also risks of increased promotion of poor-quality screening in the private sector.

A: DO NOTHING	
	Option A2: Do nothing to address the current problems relating to antenatal screening for Down syndrome and other fetal anomalies, but direct additional resources to disability support services and measures to increase social inclusion.
Level of intervention	Minimal
Extent to which it will address the current problems	This would not address any of the current problems with antenatal Down syndrome screening. However, additional funding for disability support services may address concerns about existing support services for people with Down syndrome and their families being inadequately resourced. The lived experience of people with Down syndrome is the product of societal arrangements that may be more or less disabling. From this point of view, the harms potentially experienced by the person with Down syndrome do not necessarily arise from their condition, but from the various attitudinal and institutional barriers they encounter.
Technical considerations	As for A1. The identification of appropriate disability support services and measures, and their resource implications, should be covered by the review of long-term disability supports, which is currently being conducted by the Office for Disability Issues.[5]
Ethical principles	Same as for A1 with the following additional points. *Reproductive autonomy* The autonomy of pregnant women would be enhanced because decisions may be based on better information about the availability of appropriate support services for people with Down syndrome and their families, and the lived experience of Down syndrome. For children born with Down syndrome, autonomy is enhanced, by facilitating independence and social participation. *Beneficence* There is potential to provide benefit to pregnant women, their families and children born with Down syndrome by providing greater access to support services, which facilitates independence and community membership and removes barriers to participation. *Justice* Additional funding for disability support services would promote justice, to the extent that it would help make disability supports equitably accessible.
Risks	Same as for A1.

[5]See www.odi.govt.nz/what-we-do/review-dss.html

B: STOP SOME, OR ALL, ELEMENTS OF ANTENATAL DOWN SYNDROME SCREENING	
	Option B1: Prohibit public funding being used to support/provide antenatal screening for Down syndrome and other fetal anomalies
Level of intervention	Medium
Extent to which it will address the current problems	This addresses concerns relating to public funding being used to provide a test that is of poor quality. However, it would not address other elements of the problem, as described in the risks section (below).
Technical considerations	The only form of screening currently supported by public funding is NT. While NT is not specifically funded, it is often performed as part of a publicly funded first trimester ultrasound scan. The first trimester scan receives public funding under the Section 88 Maternity Services Notice, and is an important tool in screening of the fetus. It has a number of benefits, including: • confirmation of viability • confirmation of gestational age. Increased NT is also a marker for other conditions: • trisomy 18, trisomy 13, Turners syndrome and triploidy • structural heart defects • other structural abnormalities • some rare genetic syndromes. Prohibiting the use of public funding would essentially amount to prohibiting NT measurements being performed as part of a publicly funded first trimester ultrasound. Screening would still be able to be performed as long as a woman was able to pay the full price of the ultrasound scan with NT. This option is expected to result in increased inequalities in access to screening, without any guarantee that screening would be safer or of improved quality.
Application of ethical principles	*Reproductive autonomy* An increased number of women would be denied an opportunity to decide whether to have screening. Screening could continue to be performed without consumers having received full information and giving informed consent. *Non-maleficence* There is still potential for harm as a result of privately funded ultrasound screening. NT is still not the safest screening test available. It results in a significant number of women being given false-positive results and being offered unnecessary referral for invasive procedures. This increases the risk of fetal loss. In addition, there is potential to cause harm to women, who may be unnecessarily harmed as a result of being unable to access screening and make choices about their pregnancies. *Justice* Prohibiting the use of public funding to support/provide screening may result in: • a reduced perception that having a child with Down syndrome leads to suffering or disadvantage and is best avoided • a reduction in negative attitudes • increased inequalities in access – women who are able to afford screening may choose to have it, while women who are not able to afford screening are denied the choice.
Risks	Withdrawing public funding for screening would not address issues of safety and quality, including: privately funded NT screening could still continue, but by itself is not recommended best practice internationally there is increased potential for women to be referred directly to invasive diagnostic testing based on their age (because they cannot access screening) no provision of specific training and education for health practitioners a lack of high-quality information for practitioners and consumers the absence of national data collection the absence of clear responsibility for monitoring and evaluation at a national level

B: STOP SOME, OR ALL, ELEMENTS OF ANTENATAL DOWN SYNDROME SCREENING	
	Option B2: Prohibit public funding being used to support antenatal screening for Down syndrome and other fetal anomalies, and direct additional resources to disability support services and measures to increase social inclusion
Level of intervention	Medium
Extent to which it will address the current problems	This addresses concerns relating to public funding being used to provide a test that is of poor quality. It may also address concerns about existing support services for people with Down syndrome and their families being inadequately resourced. The lived experience of people with Down syndrome is the product of societal arrangements that may be more or less disabling. From this point of view, the harms potentially experienced by the person with Down syndrome do not necessarily arise from their condition, but from the various attitudinal and institutional barriers they encounter.
Technical considerations	As for option B1. Identification of appropriate disability support services and measures, and resource implications, should be covered by the review of long-term disability supports, which is currently being conducted by the Office for Disability Issues.[6]
Application of ethical principles	Same as for option B1 with some additional points. *Autonomy* For children born with Down syndrome, autonomy is enhanced by facilitating independence and social participation. *Beneficence* There is potential to provide benefit to pregnant women, their families and children born with Down syndrome by providing greater access to support services, which facilitates independence and community membership and removes barriers to participation. *Justice* Additional funding for disability support services would promote justice to the extent that respective disability supports are equitably accessible.
Risks	Same as for B1

[6]See: http://www.odi.govt.nz/what-we-do/review-dss.html

B: STOP SOME, OR ALL, ELEMENTS OF ANTENATAL DOWN SYNDROME SCREENING	
	Option B3: Prohibit all forms of antenatal screening for Down syndrome (whether publicly or privately funded)
Level of intervention	Significant
Extent to which it will address the current problems	This option addresses the concerns relating to: public funding being used to provide a test that is of poor quality a perception that having a child with Down syndrome leads to suffering or disadvantage and is best avoided (and suggests that children with Down syndrome 'should' be born).
Technical considerations	This option requires Parliament to pass legislation specifically prohibiting all forms of antenatal screening for Down syndrome and other fetal anomalies. The following technical issues are likely to arise: How would this affect tests for other conditions (eg, neural tube defects) or marker/structural anomalies that may also indicate a possibility of Down syndrome? How would this affect the second trimester scan, which also has the ability to identify fetal anomalies?
Application of ethical principles	*Reproductive autonomy* Prohibiting screening will result in women being denied an opportunity to make informed decisions about their pregnancy. *Non-maleficence* Prohibiting screening is likely to result in: • increased referral for invasive procedures, with the associated increased risk of fetal loss • avoidable harm for some women as a result of being unable to access screening and make choices about their pregnancies • difficult ethical dilemmas for practitioners where they can identify anomalies but are unable to report them. *Justice* Prohibiting screening suggests that children with Down syndrome 'should' be born. As a result it may result in a reduced perception that having a child with Down syndrome leads to suffering or disadvantage and is best avoided, and a reduction in negative attitudes. However, there are also justice implications for women who want to have screening but are not able to access it.
Risks	Prohibiting all forms of antenatal screening for Down syndrome (both publicly and privately funded) has the potential to create the following risks: significantly increased potential for women to be offered invasive diagnostic testing based on their age (because they cannot access screening) the potential for an increase in the number of late-term terminations of pregnancy, as an increased number of women will not discover the fetus has Down syndrome until diagnosis following the 18–20 weeks ultrasound scan a lack of high-quality information for practitioners and consumers the absence of clear responsibility for monitoring and evaluation at a national level.

B: STOP SOME, OR ALL, ELEMENTS OF ANTENATAL DOWN SYNDROME SCREENING	
	Option B4: Prohibit all forms of antenatal screening for Down syndrome (whether publicly or privately funded) and direct additional resources to disability support services and measures to increase social inclusion
Level of intervention	Significant
Extent to which it will address the current problems	This option addresses the concerns relating to: public funding being used to provide a test that is of poor quality a perception that having a child with Down syndrome leads to suffering or disadvantage and is best avoided (and suggests that children with Down syndrome 'should' be born) existing support services for people with Down syndrome and their families being inadequately resourced.
Technical considerations	As for B3. Identification of appropriate services and measures, and their resource implications, should be covered by the review of long-term disability supports, which is currently being conducted by the Office for Disability Issues.[7]
Application of ethical principles	As for B3, with the following additional points. *Beneficence* There is potential to provide benefit to pregnant women, their families and children born with Down syndrome by providing greater access to support services that facilitate independence and community membership and remove barriers to participation. *Justice* Additional funding for disability support services would promote justice, to the extent that respective disability supports are equitably accessible.
Risks	As for B3.

[7]See www.odi.govt.nz/what-we-do/review-dss.html

C: IMPROVE THE CURRENT SITUATION	
	Option C1: Provide education for practitioners about antenatal screening for Down syndrome
Level of intervention	Minimal
Extent to which it will address the current problems	This would assist practitioners to provide up-to-date information to consumers, and would support consumers to make informed decisions. It would not address other elements of the problem, as described under 'Risks' below.
Technical considerations	Professional education would need to be developed at a national level in conjunction with: • consumer and disability advocacy groups • colleges, professional bodies and practitioners. Education and training for health practitioners should include both medical and non-medical perspectives. Where possible, education should be delivered through existing mechanisms for practitioner education.
Application of ethical principles	*Reproductive autonomy and justice* Screening should be voluntary and based on a principle of informed consent. Enhanced education for health practitioners is necessary to ensure that screening is communicated in an appropriate way to consumers, and that consumers are supported to make informed decisions.
Risks	Education for practitioners, would not, by itself, address issues of safety and quality, including: • whether women are giving informed consent to screening • current practice does not meet best practice • the potential for women to continue to be referred directly to invasive diagnostic testing based on their age • women who first consult a health practitioner in the second trimester of pregnancy are not able to access a publicly funded screening test, and are not likely to be offered any form of screening • the absence of national data collection • the absence of clear responsibility for monitoring and evaluation at a national level.

C: IMPROVE THE CURRENT SITUATION	
	Option C2: Develop information resources for practitioners and consumers so that high-quality, consistent information is available
Level of intervention	Minimal
Extent to which it will address the current problems	This would assist practitioners to provide up-to-date information to consumers, and would support consumers to make informed decisions. It would not address other elements of the problem, as described in the 'Risks' section (below).
Technical considerations	Resources would need to be developed at a national level in conjunction with: • consumer and disability advocacy groups • colleges, professional bodies and practitioners. Information resources should include both medical and non-medical perspectives, and information about eligibility for support services. Information resources would need to be made available, free of charge, to all relevant practitioners and consumers, and be a in a range of accessible formats.
Application of ethical principles	*Reproductive autonomy and justice* Screening should be voluntary and based on a principle of informed consent. Information resources for health practitioners and consumers are necessary to ensure that consumers have access to relevant information communicated in an appropriate way, to support informed decision-making. Consumer information would need to be available in a range of languages commonly used in New Zealand.
Risks	Information resources for practitioners and consumers would not, by themselves, address issues of safety and quality, including: • whether women are giving informed consent to screening • current practice does not meet best practice • the potential for women to continue to be referred directly to invasive diagnostic testing based on their age • women who first consult a health practitioner in the second trimester of pregnancy are not able to access a publicly funded screening test, and are not likely to be offered any form of screening • the absence of national data collection • the absence of clear responsibility for monitoring and evaluation at a national level.

C: IMPROVE THE CURRENT SITUATION	
	Option C3: Direct additional resources to disability support services and measures to increase social inclusion
Level of intervention	Moderate
Extent to which it will address the current problems	This option addresses concerns about existing support services for people with Down syndrome and their families being inadequately resourced. The lived experience of people with Down syndrome is the product of societal arrangements that may be more or less disabling. From this point of view, the harms potentially experienced by the person with Down syndrome do not necessarily arise from their condition, but from the various attitudinal and institutional barriers they encounter.
Technical considerations	Identification of appropriate services and measures, and their resource implications, should be covered by the review of long-term disability supports, which is currently being conducted by the Office for Disability Issues.[8]
Application of ethical principles	*Autonomy* The autonomy of pregnant women would be enhanced because decisions may be based on better information about the availability of appropriate support services for people with Down syndrome and their families, and the lived experience of Down syndrome. For children born with Down syndrome, autonomy is enhanced by facilitating independence and social participation. *Beneficence* There is potential to provide benefit to pregnant women, their families and children born with Down syndrome by providing greater access to support services that facilitate independence and community membership and remove barriers to participation. *Justice* This option would promote justice, to the extent that respective disability supports would be more equitably accessible.
Risks	Providing additional resources for disability support services would not, by itself, address issues of safety and quality, including: • whether women are giving informed consent to screening • current practice does not meet best practice • the potential for women to continue to be referred directly to invasive diagnostic testing based on their age • women who first consult a health practitioner in the second trimester of pregnancy are not able to access a publicly funded screening test, and are not likely to be offered any form of screening • the absence of national data collection • the absence of clear responsibility for monitoring and evaluation at a national level.

[8]See www.odi.govt.nz/what-we-do/review-dss.html

C: IMPROVE THE CURRENT SITUATION	
	Option C4: Improve the quality of screening using ultrasound markers (NT)
Level of intervention	Minimal
Extent to which it will address the current problems	This option has the potential to improve the quality and safety of NT scanning, but it fails to address the remainder of the issues, as discussed in the section on 'Risks' below.
Technical considerations	High-quality NT has a false-positive rate of 25% for a sensitivity of 85% (SURUSS). However, if appropriate accreditation, audit and monitoring are in place, some practitioners consider that NT can have a false-positive rate of 5–8% for a sensitivity of 75–82%. Improvements to NT could include: • compulsory accreditation for practitioners carrying out NT assessments • compulsory IANZ accreditation for instruments and systems used to perform NT assessments. Some of the quality assurance and accreditation issues are likely to be addressed by the proposed revisions to the Section 88 Maternity Services Notice (see the report of the NT Technical Subgroup for additional detail, Appendix 3).
Application of ethical principles	*Non-maleficence* Offering a test that does not meet international best practice when used by itself (although of improved quality) creates the potential for anxiety as a result of high levels of false-positive results, and fetal loss as a result of diagnostic procedures. *Justice* Unless improvements for NT also include increased public funding and the ability for women in all regions to access it, there will still be ethical issues relating to equality of access to screening.
Risks	Improving the quality of NT, by itself, would not address other current problems, including: • NT scanning by itself is not recommended best practice internationally • whether women are giving informed consent to screening • there is potential for women to still be referred directly to invasive diagnostic testing based on their age • inequity of access (co-payments and location of services) remains unchanged • women who first consult a health practitioner in the second trimester of pregnancy are not able to access a publicly funded screening test, and are not likely to be offered any form of screening • the absence of national data collection • the absence of clear responsibility for monitoring and evaluation at a national level.

C: IMPROVE THE CURRENT SITUATION	
	Option C5: Introduce public funding for a second trimester maternal serum screening test
Level of intervention	Minimal
Extent to which it will address the current problems	If introduced as a stand-alone test for women who first consult a health practitioner in the second trimester of screening, this option may reduce the number of women offered diagnostic procedures without screening. If combined with option C4 (improve the quality of NT scanning), this option will improve the overall quality and safety of the screening. However, it fails to address the remainder of the issues, as discussed in the section on 'Risks' (below).
Technical considerations	By itself, second trimester maternal serum screening using the quadruple test (with AFP, uE3, free ß-hCG, inhibin-A) has a false-positive rate of 6.2% for a sensitivity of 85% (SURUSS). Second trimester maternal serum screening using the quadruple test, when combined with NT, has a false positive rate of 1.6% for a sensitivity of 85% (SURUSS).
Application of ethical principles	*Reproductive autonomy and justice* This option may reduce the potential for women who present for the first time in the second trimester to be offered diagnostic procedures without first having screening, and may improve equality of access to screening. *Non-maleficence* When combined with option C4, this has the potential to reduce potential harm (both in terms of anxiety and in potential for fetal loss) as a result of reducing the number of false-positive results.
Risks	This option, either by itself or combined with option C4, would fail to address the: • potential for some women to still be referred directly to invasive diagnostic testing based on their age • potential for some women to continue to have screening based only on NT • issue of whether women are giving informed consent to screening • absence of national data collection • absence of clear responsibility for monitoring and evaluation at a national level.

C: IMPROVE THE CURRENT SITUATION	
	Option C6: Introduce a publicly funded first trimester maternal serum screening test
Level of intervention	Minimal
Extent to which it will address the current problems	This option only addresses the problem if it is combined with options C4 and/or C5. First trimester maternal serum screening by itself is not best practice, but combined with other tests is more likely to result in high-quality and safe screening. However, it fails to address the remainder of the issues, as discussed in the section on 'Risks' (below).
Technical considerations	First trimester maternal serum screening (free ß-hCG, PAPP-A) combined with NT has a false-positive rate of 6.1% for a sensitivity of 85% (SURUSS). First trimester maternal serum screening integrated with second trimester maternal serum screening has a false-positive rate of 2.7% for a sensitivity of 85% (SURUSS). First trimester maternal serum screening integrated with both NT and second trimester maternal serum screening has a false-positive rate of 1.2% for a sensitivity of 85% (SURUSS).
Application of ethical principles	*Non-maleficence* When combined with the results of NT and/or second trimester serum screening, this option has the potential to reduce harm (both in terms of anxiety and fetal loss) as a result of reducing the number of false-positive results.
Risks	This option, either by itself or combined with options C1 would fail to address: • the potential for some women to still have screening based only on NT or maternal age • whether women are giving informed consent to screening • the potential for some women to still have screening without being fully informed and giving informed consent • the absence of national data collection • the absence of clear responsibility for monitoring and evaluation at a national level. Unless this option is accompanied by serum screening for women who first consult a health practitioner in the second trimester of screening, it has the potential for some women to be referred to diagnostic procedures without an opportunity to have screening.

C: IMPROVE THE CURRENT SITUATION	
	Option C7: Implement all of the sub-options outlined above in C1 to C6, and implement these in such a way that the screening is still provided opportunistically rather than as part of a nationally organised programme
Level of intervention	Significant
Extent to which it will address the current problems	This option addresses almost all elements of the problem, except for the issues that relate to co-ordination and accountability. However, this option has the potential to exacerbate negative attitudes towards disability. It is also likely to result in a reduced incidence of Down syndrome, despite this not being one of the purposes of screening. This is discussed in the section on 'Risks' below.
Technical considerations	Sensitivity and false-positive rates are as for option C6. Technical issues for this option would largely relate to the different ways in which test results could be combined to provide single assessments (including contingent screening). There needs to be further consideration of which combination of tests is preferable.
Application of ethical principles	*Reproductive autonomy* This option is expected to provide better access to safer and more effective screening. It is likely to reduce the potential for women to be offered diagnostic procedures without first having screening. *Non-maleficence* It may reduce potential harm (both in terms of anxiety and fetal loss) as a result of reducing the number of false-positive results. It should also provide more equitable access to screening. However, there is the potential to increase harm for people with disabilities, and their families, by increasing the perception that Down syndrome is something to be avoided. *Justice* An unintended consequence of this option is the likely decrease in the incidence of Down syndrome.
Risks	This option would fail to address the absence of national data collection, and the absence of clear responsibility for monitoring and evaluation at a national level. There is potential for public perception to be that the purpose of screening is to reduce the incidence of Down syndrome, or other conditions, and to undermine the value of people living with disabilities.

C: IMPROVE THE CURRENT SITUATION	
	Option C8: Implement all of the sub-options outlined above in C1 to C6, and implement these in the form of a nationally organised screening programme
Level of intervention	Significant
Extent to which it will address the current problems	This option addresses all elements of the problem. In addition to implementing safer screening tests, a screening programme should include: • development of education for practitioners • development of information resources for practitioners and consumers • standards that practitioners and services are required to meet • monitoring and evaluation of the screening programme at local and national levels • clear accountability for the screening programme. However, this option has the potential to exacerbate negative attitudes towards disability. It is also likely to result in a reduced incidence of Down syndrome, despite this not being one of the purposes of screening. This is discussed under 'Risks' (below).
Technical considerations	Sensitivity and false-positive rates are as for option C6. There would need to be further consideration of the combination of tests. A nationally organised screening programme requires a clear screening and diagnostic pathwa
Application of ethical principles	*Reproductive autonomy* This option is expected to ensure that consumers are supported to make informed decisions by providing clear requirements for health practitioner education and providing nationally consistent information resources. It should also provide better access to safer and more effective screening. It is likely to reduce the potential for women to be offered diagnostic procedures without first having screening. *Non-maleficence* It may reduce potential harm (both in terms of anxiety and fetal loss) as a result of reducing the number of false-positive results. It should also provide more equitable access to screening. However, it also has the potential to increase harm for people with disabilities, and their families, by increasing the perception that Down syndrome is something to be avoided. *Justice* Unintended consequences of this option are: the likely decrease in the incidence of Down syndrome the perception that having a child with Down syndrome or other condition leads to suffering or disadvantage and is best avoided negative attitudes towards people with disabilities, and their families.
Risks	There is potential for public perception to be that the purpose of the programme is to reduce the number of babies born with Down syndrome, or other conditions, and to undermine the value of people living with disabilities.

9.0 Assessment of antenatal screening for Down syndrome against criteria to assess screening programmes

National Health Committee criteria for assessing screening programmes

The Advisory Group considered antenatal screening for Down syndrome against eight criteria to assess screening programmes developed by the National Health Committee. These criteria are designed to aid the assessment of screening programmes in contrast to screening performed opportunistically. In the foreword to the criteria the National Health Committee Chair states:

> *… screening is a complex process that requires careful consideration of clinical, social, ethical and economic issues. Screening programmes should be based on good quality evidence that they do more good than harm, at reasonable costs, and they should be delivered within the context of an effective quality assurance programme. (National Health Committee 2003a).*

The criteria are summarised in the box below, and then discussed in more detail.

Summary of criteria

1. **The condition is a suitable candidate for screening.**
2. **There is a suitable test.**
3. **There is an effective and accessible treatment or intervention for the condition identified through early detection.**
4. **There is high-quality evidence, ideally from randomised controlled trials, that a screening programme is effective in reducing mortality or morbidity**
5. **The potential benefit from the screening programme should outweigh the potential physical and psychological harm (caused by the test, diagnostic procedures and treatment**
6. **The health care system will be capable of supporting all necessary elements of the screening pathway, including diagnosis, follow-up and programme evaluation.**
7. **There is consideration of social and ethical issues.**
8. **There is consideration of cost–benefit issues.**

The National Health Committee document provides the following guidance on applying the criteria:

Assessing a potential or existing screening programme will require balancing these criteria in the context of an overall programme. Their consideration will provide the necessary information to ensure an informed decision is made about whether to introduce, maintain or modify a screening programme. The criteria are not intended to be absolute, as no existing or potential screening programme fulfils every criterion entirely. (National Health Committee 2003a: 23)

Table 5: National Health Committee screening assessment criteria applied to antenatal screening for Down syndrome and other fetal anomalies

NATIONAL HEALTH COMMITTEE CRITERION	APPLIED TO ANTENATAL SCREENING PROGRAMME
Criterion 1 The condition is a suitable candidate for screening	Down syndrome is the single most common chromosomal disorder, with a population incidence of about 1 in 700 births in New Zealand. Down syndrome screening is already offered in New Zealand in the form of publicly funded NT measurement and assessments. At least 25,000 pregnant women choose to have NT screening each year. Antenatal screening can provide women with an individualised assessment of their chance that their fetus has Down syndrome. Screening also gives information about structural problems, other aneuploidies and biochemical conditions. This information can assist women to make decisions, including whether to have diagnostic testing (if they are screen positive). If the diagnostic testing gives a diagnosis of Down syndrome, or other fetal anomaly, it provides an opportunity to plan and make decisions about the pregnancy.
Criterion 2 There is a suitable test Tests should have the following characteristics: • safe, simple, reliable, accurate/valid • highly sensitive • highly specific.	Current screening practice is unacceptable – the tests used are unsafe, not considered sufficiently specific or sensitive, and do not meet best practice. International evidence has identified that the most suitable (sensitive and specific) tests are: • NT combined with first trimester serum screening • NT combined with second trimester serum screening • integrated screening (NT combined with both first and second trimester serum screening) • serum integrated (first and second trimester maternal serum screening). The methods noted above have a lower false-positive rate, and can minimise harm by reducing the number of invasive diagnostic procedures performed. Diagnostic procedures carry an additional risk of fetal loss of around 0.5–2%. All tests require a degree of co-ordination to ensure that the: • stage of pregnancy is accurately known • tests are performed at the correct stage of pregnancy • results are appropriately transferred and combined to arrive at a risk calculation.
Criterion 3 There is an effective and accessible treatment or intervention for the condition identified through early detection	(This criterion is difficult to apply to the concept of antenatal screening for Down syndrome and is more relevant for conditions where there is a means of directly treating the condition.) Antenatal screening for Down syndrome is not offered so that Down syndrome can be 'treated'. The purpose of screening for Down syndrome is to enable women who choose to have screening to make informed decisions about their pregnancy, with appropriate support and counselling. Informed decision-making could include: • whether to have a diagnostic test • considering continuation or termination of pregnancy • preparing for the birth and life of a child with Down syndrome, or another fetal anomaly • planning to give birth at a specialist centre, if necessary, that can provide any immediate neonatal treatment required. Some models of health care would include these purposes as 'interventions'.

Criterion 4 There is high-quality evidence, ideally from randomised controlled trials, that a screening programme is effective in reducing morbidity or mortality	(This criterion is difficult to apply to the concept of antenatal screening for Down syndrome, and is more relevant for conditions where the aim of the programme is a reduction in mortality and morbidity.) The purposes of screening for Down syndrome are discussed above, and do not include the reduction of morbidity or mortality. Outcomes of pregnancy after antenatal screening depend on the quality of the screening and diagnostic tests, and on parental choices. Focusing on mortality and morbidity implies that the purpose of screening is to reduce the number of children born with Down syndrome or other conditions. However, the aim of a screening programme would not be to reduce morbidity and mortality related to Down syndrome but to improve the quality (and therefore a reduction in false-positive rates) of screening tests and to reduce harm relating to diagnostic procedures.
Criterion 5 The potential benefit from the screening programme should outweigh the potential physical and psychological harm (caused by the test, diagnostic procedures and treatment)	Harms and benefits of screening are subjective and depend on the attitudes and beliefs of the prospective parent(s). For some, a diagnosis of Down syndrome provides an opportunity to plan and prepare. For others, not having a child with Down syndrome would be a positive outcome. Potential harms may include: • false-positive screening results and referral for an unnecessary and invasive diagnostic procedure • a miscarriage as a result of the diagnostic procedure • false-negative results and a missed opportunity to be referred to diagnostic testing • poorly informed decision-making • anxiety and stress • pressure to agree to screening and/or diagnostic tests • pressure to make a decision regarding the outcome of the pregnancy. Potential benefits may include equal: • opportunities to make informed decisions regarding screening, diagnosis and the outcome of the pregnancy • opportunities to access high-quality, safe screening and diagnostic services • access to high-quality information • access to high-quality support services • access to non-directive counselling services.
Criterion 6 The health care system will be capable of supporting all necessary elements of the screening pathway, including diagnosis follow-up and programme evaluation	Further investment would be necessary to support better-quality antenatal screening in New Zealand. This would include investing in: • education and information for health practitioners and consumers • better-quality screening tests • a system to co-ordinate services, screening results and referrals • a system of monitoring and evaluation, based on high-quality data collection • workforce development.

Criterion 7 There is consideration of social and ethical issues	Down syndrome screening is already widely available in New Zealand. However, the manner in which screening is offered and provided raises significant social and ethical concerns, with the potential for screening to result in more harm than benefit. For an antenatal Down syndrome screening programme to be socially and ethically acceptable it would need to be: • universally offered to all pregnant women • accompanied by high-quality, balanced, easily understood information in an accessible range of formats for practitioners and consumers • conditional on informed consent • based on voluntary participation at each stage of the screening and diagnostic pathway • based on unconditional acceptance of, and support for, the choices made by women as a result of screening and diagnostic tests • delivered within a framework that is responsive to Māori and improves opportunities for Màori to make informed decisions • integrated with other tests offered in pregnancy • the safest and most effective tests, based on evolving international evidence • focused on reducing inequalities in access to antenatal screening for Down syndrome • fully funded so that any woman who chooses to have screening is able to access it • monitored and evaluated at a national level.
Criterion 8 There is consideration of cost–benefit issues	(This criterion is difficult to apply to the concept of antenatal screening for Down syndrome.) While we agree that the costs and consequences of screening should be analysed, it is not always possible to perform a cost–benefit analysis. A cost–benefit analysis for Down syndrome screening would require us to place a dollar value on the outcomes of the screening. This would be very difficult to do and is likely to result in additional social and ethical issues. We do not think that a cost–benefit analysis is always appropriate. The National Screening Unit is carrying out an analysis of the costs and consequences of screening methods as part of its policy process. Due to timing constraints, however, the Advisory Group has not had an opportunity to consider this analysis.

Are these appropriate criteria to assess antenatal screening programmes?

Some members of the Advisory Group have questioned whether the criteria developed by the National Health Committee are the right ones to use when assessing an antenatal screening programme, including one for Down syndrome. Some members of the Advisory Group believe that the National Health Committee criteria were developed when the concept of screening programmes focused largely on cancer screening, and that the potential to apply these criteria to screening for conditions such as Down syndrome was not considered.

In addition, the criteria were developed for a context in which one screening test is able to detect one condition. For many of the antenatal and genetic screening tests, a single test has the potential to detect many conditions.

The Advisory Group has written to the National Screening Advisory Committee to suggest that the criteria to assess screening programmes be reviewed, with the goal of developing a useful set of criteria that are applicable to screening for a wide range of conditions. This would support a more robust assessment of potential screening programmes, not envisaged when the National Health Committee criteria were developed.

There are a number of additional criteria which some members of the Advisory Group feel would more usefully guide the assessment of a potential antenatal screening programme for Down syndrome. For example, the Netherlands Health Council (1994) has developed a set of criteria for genetic screening. While many of the criteria are similar to the National Health Committee's, there were three additional ones that the Advisory Group thought could be usefully applied to Down syndrome. These are:

A. Practical courses of action must be open to the participants

B. Participation in a genetic screening programme should be completely voluntary and should be conditional on consent based on good information

C. The target group should be supplied with good-quality, comprehensible information.

The following table summarises the application of these additional criteria to antenatal screening for Down syndrome.

Table 6: Additional criteria to assess screening programmes

Additional criterion A Practical courses of action must be open to the participants	Antenatal screening for Down syndrome, and other fetal anomalies, provides women with opportunities to make decisions. A number of practical courses of action should be offered to participants, including: • diagnostic testing • access to counselling and support services • considering continuation or termination of pregnancy • preparing for the birth and life of a child with Down syndrome, or another condition • planning to give birth at a specialist centre, if necessary, that can provide any immediate neonatal treatment required.
Additional criterion B Participation in a genetic screening programme should be completely voluntary and should be conditional on consent based on good information	A screening programme, if established, would be completely voluntary. Women, and their families, would have the right to make informed decisions to accept or decline an offer of screening. This is supported by New Zealand's legal framework, including the Code of Health and Disability Services Consumers' Rights.
Additional criterion C The target group should be supplied with good-quality, easily understood information	The 'target group' would be all pregnant women. All pregnant women, and their families, should be supplied with good-quality, comprehensible information including information about: • Down syndrome, and the experience of living with Down syndrome • the right to refuse screening • the potential for other conditions to be found during screening • any significant medical, social or financial side- effects/implications of screening • the difference between a screening test and a diagnostic test • the screening tests, including the risks and benefits • a description of the entire screening pathway, including the potential for counselling and support • where to go for further information.

10.0 Preferred options

The Advisory Group was not able to reach a unanimous view on the best way forward for antenatal screening for Down syndrome in New Zealand. It is expected that due to the complex and difficult nature of the issues, the general public in New Zealand will have the same difficulty in reaching a consensus.

However, while the Advisory Group was not able to reach a unanimous view on what should happen, it was able to reach consensus on what should not happen. The Advisory Group unanimously agreed that the National Screening Unit and the Ministry of Health must not 'do nothing' about the current situation of antenatal screening for Down syndrome.

The Advisory Group agreed that the current practice of screening using only NT and/or maternal age, without biochemical markers, is unsafe, inequitable and untenable, and should not continue.

Two options were preferred by Members of the Advisory Group. A large majority of the Advisory Group preferred Option C8: Implement all of the sub-options outlined above in C1 to C6, and implement these in the form of a nationally organised screening programme

A minority of Advisory Group members consider the best option is to consider directing additional resources to disability support services, regardless of which option is chosen in relation to screening.

Majority preference: continue to offer screening and to improve the quality and safety of screening tests

A significant majority of the Advisory Group considers that antenatal screening for Down syndrome should continue to be offered to women so that women can make informed choices about their pregnancies. Significant steps should be taken to improve the quality and safety of screening, in the form of a nationally organised screening programme. Members of the Advisory Group who support this option recognise that better-quality screening does not necessitate the introduction of a nationally organised screening programme, but the majority of members support the introduction of a nationally organised screening programme as the best way of ensuring consistently safe and effective screening that meets agreed standards, and is monitored and evaluated.

In addition, members acknowledge the concerns relating to a nationally organised screening programme, particularly in relation to public perceptions of the desirability of screening, negative attitudes towards disability and the potential impact on the number of children born with Down syndrome and other conditions. If a national screening programme is implemented it should aim to reduce these potential effects.

The Advisory Group's support for a national screening programme is conditional on screening being:

a. universally offered to all pregnant women

b. accompanied by high-quality, balanced, easily understood information in an accessible range of formats for practitioners and consumers

c. conditional on informed consent

d. based on voluntary participation at each stage of the screening and diagnostic pathway

e. based on unconditional acceptance of, and support for, the choices made by women as a result of screening and diagnostic tests

f. delivered within a framework that is responsive to Māori and improves opportunities for Māori to make informed decisions

g. integrated with other tests offered in pregnancy

h. the safest and most effective tests, based on evolving international evidence

i. focused on reducing inequalities in access to antenatal screening for Down syndrome

j. fully funded so that any woman who chooses to have screening is able to access it

k. monitored and evaluated at a national level.

The Advisory Group had limited opportunity to discuss the best screening pathway for a national screening programme, but consider that the best option is likely to be a combination of first trimester and second trimester biochemical and ultrasound screening to provide a single assessment. It is recommended that, if screening is to continue to be offered in New Zealand, an advisory group be established to provide ethical and technical advice on the best form of screening for New Zealand, including issues relating to safety, consumer preferences, data collection and privacy.

Minority preference

A minority of Advisory Group members do not support a national screening programme. International literature suggests that a national screening programme will lead to a reduced incidence of Down syndrome, and other fetal anomalies. There is concern that the establishment of a national screening programme will imply that the aim is to reduce the incidence of Down syndrome. This may cause fear that women who choose to continue a pregnancy when the fetus has been diagnosed as having Down syndrome, will not be supported in their decisions or be eligible for appropriate disability support services.

The minority considers the best option is to consider directing additional resource to disability support services, regardless of the decision made in relation to screening. The perceived availability of relevant support services for people with Down syndrome and their families is directly relevant to choices parents might make about whether or not to continue with the pregnancy when the fetus has been diagnosed as having Down syndrome. Strengthening disability support services accompanied by information about, and an assurance of eligibility for, services is crucial as it has the ability to increase the potential for women, and their families, to feel supported to raise a child with Down syndrome.

Additional funding for disability support services would improve the quality of life for people with Down syndrome, by increasing opportunities for community membership and independence.

Relevant examples of ways additional funding could be used in the disability sector can be found in a report titled ‘To have an Ordinary Life: Community membership for adults with an intellectual disability’ (National Health Committee 2003a).

References

1. Agence d'Evaluatiorides Technologies ct-des Modes d'Intervention en Sante. 2001. *Issues Concerning Prenatal Screening and Diagnosis of Down Syndrome.* Montreal: AETMIS, 2001, xix-90 p. (original French version published in September 1999.)

2. Avgidou K, Papageorghiou A, Bindra R, et al. 2005. *Prospective first-trimester screening for trisomy 21 in 30,564 pregnancies.* American Journal of Obstetrics and Gynecology 192: 1761–7.

3. Berkowitz R, Robters J, Minkoff H. 2006. *Challenging the strategy of age-based prenatal genetic counseling.* Journal of the American Medical Association 295: 12.

4. Boyle RJ, de Crespigny L, Savulescu J. 2003. *An ethical approach to giving couples information about their fetus.* Human Reproduction 18: 2253–6.

5. Chang T. 2006. *Antenatal screening for Down syndrome in New Zealand: time for a national screening policy?* Australian and New Zealand Journal of Obstetrics and Gynaecology 46: 92–6.

6. Cuckle H, Arbuzova S. 2005. *Down syndrome screening in the first and/or second trimester: model predicted performance using meta-analysis parameters.* Seminars in Perinatology 29(4): 252.

7. de Graaf IM, Tijmstra T, Bleker OP, et al. 2002. *Women's preference in Down syndrome screening.* Prenatal Diagnosis 22: 624–9.

8. Malone FD, Canick JA, Ball RH, et al 2005. *First-trimester or second-trimester screening, or both, for Down's syndrome.* New England Journal of Medicine 353: 2001–11.

9. Marteau TM, Cook R, Kidd J, et al. 1992. T*he psychological effects of false positive results in prenatal screening for fetal abnormality: a prospective study.* Prenatal Diagnosis 12: 205–14.

10. Minister for Disability Issues. 2001. *The New Zealand Disability Strategy: Making a world of difference – Whakanui Oranga.* Wellington: Ministry of Health.

11. Ministry of Health. 2006. ***Maternity Report 2003***. Wellington: Ministry of Health.

12. Morris J, Wald N. 2005. *Graphical presentations of distributions of risk in screening.* Journal of Medical Screening 12: 155–60.

13. Mulvey S, Zachariah R, McIlwaine K, et al. 2003. *Do women prefer to have screening tests for Down syndrome that have the lowest screen-positive rate or the highest detection rate?* Prenatal Diagnosis 23: 828–32.

14. National Health Committee. 2003a. *Screening to Improve Health in New Zealand: Criteria to assess screening programmes.* Wellington: Ministry of Health.

15. National Health Committee. 2003b. *To Have an 'Ordinary' Life: Community membership for adults with an intellectual disability.* Wellington: Ministry of Health.

16. Netherlands Health Council. 1994. *Genetic Screening.* The Hague: Health Council of the Netherlands.

17. New Zealand Down Syndrome Association [no date]. *Living with Down Syndrome.* New Zealand: New Zealand Down Syndrome Association.

18. Nicolaides KH, Spencer K, Avgidou K, et al. 2005. *Multicenter study of first-trimester screening for trisomy 21 in 75,821 pregnancies: results and estimation of the potential impact of individual risk-orientated two-stage first-trimester screening.* Ultrasound in Obstetrics and Gynecology 25: 221–6.

19. Penrose l. 1933. *The relative effects of paternal age and maternal age in mongolism.* Journal of Genetics 27: 219.

20. Simpson JL. 2005. *Choosing the best prenatal screening protocol.* New England Journal of Medicine 353(19): 2068–70.

21. Snijders R, Noble P, Sebire N, et al. 1998. *UK Multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10–14 weeks of gestation.* Lancet 352: 343–6.

22. Snijders R, Thorm E, Zachary J, et al. 2002. *First trimester trisomy screening: nuchal translucency measurement training and quality assurance to correct and unify technique.* Ultrasound in Obstetrics and Gynaecology 19: 353–9.

23. Spencer K, Aitken D. 2004. *Factors affecting women's preference for type of prenatal screening test for chromosomal anomalies*. Ultrasound Obstetrics and Gynaecology 24: 735–9.

24. Stone P, Austin D. 2006. *Report to the National Screening Unit: Assessment of antenatal screening for Down syndrome in New Zealand.* Wellington: Ministry of Health.

25. Stone PR. 2005. *Prenatal testing for aneuploidy in New Zealand: time for action.* New Zealand Medical Journal 118: 1217.

26. Wald NJ, Cuckle HS, Densem JW, et al. 1988. *Maternal serum screening for Down's syndrome in early pregnancy.* British Medical Journal 297: 883–7.

27. Wald NJ, Hackshaw AK. 1997. *Combining ultrasound and biochemistry in first trimester screening for Down's syndrome.* Prenatal Diagnosis 17: 821–9.

28. Wald N, Leck I. 2000. *Antenatal and Neonatal Screening (2nd edn).* Oxford: Oxford University Press.

29. Wald NJ, Rodeck C, Hackshaw AK, et al. 2003. *First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS).* Journal of Medical Screening 10: 56–104. [Fully reported in Health Technology Assessment 2003; 7: 1–77.]

30. Wald NJ, Rodeck C, Hackshaw AK, et al. 2004. *SURUSS in perspective.* British Journal of Obstetrics and Gynaecology 111: 521–531.57.

Glossary

Amniocentesis	An invasive procedure, performed trans-abdominally under ultrasound guidance, whereby a sample of amniotic fluid is removed and sent for analysis.
Aneuploidy	Having a chromosome number that is either more than or less than the standard number (eg, Down syndrome, which is characterised by having three copies of chromosome 21).
Chorionic villus sampling (CVS)	An invasive procedure, performed trans-abdominally or trans-cervically, whereby placental tissue is removed and sent for analysis.
Chromosome	A rod-like structure present in the nucleus of all body cells which contains genetic information. Normally humans have 23 pairs, with the unfertilised ova and each sperm carrying a set of 23 chromosomes. On fertilisation the ova and sperm combine to give a total of 46 (23 pairs).
Combined test	A screening test, comprising a first trimester test based on combining nuchal translucency measurement with free beta human chorionic gonadatrophin (hCG), pregnancy-associated plasma protein A (PAPP-A) and the woman's age.
Congenital	A condition that is present at birth, although it can be recognised antenatally, at birth or many years later. A congenital disorder can be genetic or acquired at any time during fetal development.
Cut-off threshold	The value of a screening variable at which the individual is more likely to be helped than harmed by the offer of a diagnostic test; the division between screen-negative / low-risk and screen-positive / high-risk.
Dating scan	A means of confirming gestational age (the length of pregnancy in weeks and days) using fetal measurements obtained during an ultrasound scan.
Detection rate (DR) sensitivity	The proportion of fetuses affected by the screened disorder that will be or identified by a screening test.
Diagnosis	Identification of a disease or condition.
Diagnostic test	A test that provides a diagnosis (ie, states if the condition definitely is present or absent).
Disability	According to the New Zealand Disability Strategy, disability is not something that individuals have. Individuals have impairments. These may be physical, sensory, neurological, psychiatric, intellectual or other impairments. Disability results from the interaction between people with impairments and the environmental and attitudinal barriers they face. See Impairment.
Down syndrome	A disorder caused by the presence of an extra chromosome 21 (ie, three instead of two)> This is also called trisomy 21.
False-negative	A fetus affected by the screened condition that was classified by screening as low-risk / screen-negative.

False-positive	A fetus unaffected by the screened condition that was classified by screening as high-risk / screen positive.
False-positive rate or specificity	The proportion of women screened whose test result describes a level of risk high enough to warrant an invasive diagnostic procedure but whose fetus is unaffected. Usually expressed as a percentage. The proportion of women screened whose test result describes a level of risk high enough to warrant an invasive diagnostic procedure but whose fetus is unaffected. Usually expressed as a percentage.
Fetal anomaly	A fetal malformation or variation from normal development, commonly identified by ultrasound scan.
First-trimester or second-trimester screening, or both, for Down's Syndrome FASTER)	A study published in the New England Journal of Medicine in November 2005, which concluded that 'first-trimester combined screening at 11 weeks of gestation is better than second-trimester quadruple screening but at 13 weeks has results similar to second -trimester quadruple screening. Both stepwise sequential screening and fully integrated screening have high rates of detection of Down's syndrome, with low false positive rates'.
Fluorescent in situ hybridisation (FISH)	A diagnostic test in which a chromosome-specific DNA probe is used on interphase cells from chorionic villi or amniotic fluid to demonstrate the presence or absence of the specified chromosomes.
Gestational age	The duration of an ongoing or completed pregnancy, measured from the first day of the last menstrual period (usually about two weeks longer than that measured from conception). Gestational age is usually measured in completed weeks (eg, a pregnancy between 16 weeks and 16 weeks 6 days counts as a 16-week pregnancy).
Impairment	As defined by the World Health Organization, impairment is any loss or abnormality of psychological, physiological or anatomical structure or function. Impairments do not necessarily result in disability. See Disability.
Incidence	One of the main ways to measure the frequency of a disease in a particular population. The incidence of a disease is the number of new cases that occur during a particular time.
Integrated test	A screening test involving the integration of measurements performed at different times of pregnancy into a single test result. Unless otherwise qualified, 'integrated test' refers to the integration of nuchal translucency measurement and PAPP-A in the first trimester with the quadruple test in the second trimester
Invasive diagnostic tests	A variety of invasive techniques used in pregnancy to obtain samples to determine the chromosomal or genetic constitution of the fetus by karyotyping or other means (eg, amniocentesis, chorionic villus sampling).
Karyotyping	Microscopic analysis of the number and type of chromosomes after blood sampling, amniocentesis or CVS.

Lead maternity carer (LMC)	A lead maternity carer is responsible for providing and co-ordinating a woman's maternity care, including developing a care plan and attending labour and birth. A lead maternity carer can be a midwife, a general practitioner with a Diploma in Obstetrics (or equivalent, as determined by the New Zealand College of General Practitioners), an obstetrician or a hospital team.
Maternal serum screening	A screening test whereby blood is taken from the woman at predefined times during the pregnancy and the concentration of various biochemical markers in the serum is measured. These are used as part of a calculation, including maternal age, to give a risk estimation for Down syndrome.
Multiple of the median	The serum marker concentration for a pregnant woman divided by the median concentration value for unaffected pregnancies of the same gestational age.
National Screening Unit (NSU)	The National Screening Unit (NSU) is part of the Ministry of Health. It has responsibility for the national operational functioning and strategic management of national screening programmes.
Nuchal translucency (NT) measurement	A screening test involving measurement of the depth of fluid at the nape of the fetal neck taken during an early ultrasound scan. An increased amount of fluid may indicate that the fetus has Down syndrome or another chromosomal, structural or genetic anomaly.
Odds of being affected given a positive result	This is the ratio of the number of affected individuals with positive screening test results to the number of unaffected individuals with positive screening test results.
Opportunistic screening	Opportunistic screening occurs for a wide range of conditions with varying degrees of organisation, but there is no formal co-ordination, monitoring or evaluation of the process. Such screening may be widely undertaken (eg, antenatal screening for a range of conditions), but not necessarily as part of a screening programme.
Prevalence	An epidemiological term describing the proportion of a defined group in the population having a condition at one point in time.
Quadruple test	A screening test, involving risk estimation in the second trimester based on the measurement of AFP, uE3, free beta-hCG (or total hCG) and inhibin-A, combined with the prior risk due to the woman's age.
Screening	The systematic application of a test or inquiry to identify individuals at specific risk of a specific condition who might benefit from further investigation.
Screening programme	Organised screening is delivered through a screening programme, with planning, co-ordination, monitoring and evaluation of all activities along the screening pathway. Quality assurance and improvement processes distinguish screening programmes from opportunistic screening, balancing the achievable benefits of screening with the potential harms.

Screening test	A test performed on people who do not have any symptoms. It predicts the likelihood of a person having or developing a particular condition.
Section 88 Maternity Services Notice	A notice under section 88 of the New Zealand Public Health and Disability Act 2000 which sets out the terms and conditions for the provision of maternity services.
Sensitivity	See Detection rate.
Serum markers	Biochemical substances found in a pregnant woman's serum (eg, alpha-fetoprotein, which have been proven to be at a statistically different level in normal fetuses compared to that in affected fetuses. Also known as analytes.
Specificity	See False-positive rate.
SURUSS	Refers to the results of the Serum, Urine and Ultrasound Study: a study into first and second trimester antenatal screening for Down syndrome:
Termination of pregnancy	The medical expulsion or surgical extraction from the uterus of a fetus in the first, second or third trimester of pregnancy.
(TOP) Trimester	For convenience the nine-month gestation period is divided into three three-month trimesters, as follows: • first trimester: from conception to the end of the 12th week • second trimester: from the beginning of the 13th week to the end of the 28th • third trimester: from the beginning of the 29th week until established labour.
Triple test	A second trimester screening test based on the measurement of AFP, unconjugated oestriol (uE3), and hCG (either total hCG or free beta-hCG), together with the woman's age.
Trisomy	The addition of a complete extra chromosome to a pair.
Ultrasound	Sound with frequencies above the range of human hearing. Diagnostically useable ultrasound is usually in the range of 2 to 10 MHz.
Ultrasound scan	High frequency echoes are turned into electrical signals and real-time images are produced on a screen, as the ultrasound transducer is moved across the skin.
Uptake rate	The proportion of women from the pregnant population who have screening.

Appendices

Appendix 1: Membership of the Antenatal Down Syndrome Screening Advisory Group

NAME	EXPERTISE
Paul Harper (Chair)	Down syndrome and a disability perspective
Norma Campbell	midwifery
Sharron Cole	women's health
Rea Daellenbach	women's health
Clive Felix	cytogenetics
John Forman	rare disorders and a disability perspective
Owen Hughes	government policy relating to disability
Nikki Kerruish	paediatrics and health ethics
Pippa Kyle	obstetrics and fetal medicine
Fiona McCrimmon	health law
Rachael McEwing	radiology/sonography
Katherine Neas	clinical genetics
Anne O'Connor	Down syndrome and a disability perspective
Peter Stone	obstetrics and fetal medicine
Mere Wallace	social work and a Maori health perspective
Dianne Webster	biochemical genetics
Wendi Wicks	disability perspectives
Jonathon Wilcox	general practice
Lynda Williams	women's health
Supported by	
Karen Mitchell	Group Manager, National Screening Unit (NSU)
Angie Partridge	Antenatal Screening Co-ordinator, NSU
Caroline Greaney	Senior Policy Analyst, NSU
Nicola Chapple	Senior Policy Analyst, NSU
Yvonne Walmsley	Technical Advisor, NSU
Andreya Brown	Policy Analyst, NSU

Social and Ethical Subgroup

The Social and Ethical Subgroup considered antenatal screening for Down syndrome from ethical, social and disability perspectives.

Membership

Paul Harper (Chair)

Sharron Cole

Rea Dallenbach

John Forman

Owen Hughes

Nikki Kerruish

Anne O'Connor

Fiona McCrimmon

Mere Wallace

Wendi Wicks

Lynda Williams

Serum Screening Technical Subgroup

The Serum Screening Technical Subgroup considered the feasibility of offering publicly funded second trimester maternal serum screening to women unable to access first trimester NT screening.

Membership

Dianne Webster (Chair)

Norma Campbell

Clive Felix

Pippa Kyle

Kate Neas

Anne O'Connor

Peter Stone

Jonathan Wilcox

Nuchal Translucency Technical Subgroup

The Nuchal Translucency Technical Subgroup considered the key issues relating to NT practice in New Zealand.

Membership

Rachael McEwing (Chair)

Pippa Kyle

Peter Stone

The Serum Screening Technical Subgroup and the Nuchal Translucency Technical Subgroup also considered technical issues relating to the various programme options.

Appendix 2: Social and Ethical Subgroup: Report to the Antenatal Down Syndrome Screening Advisory Group

Purpose

The purpose of this report is to:

a. ensure that the National Screening Unit (NSU) and the Government recognise the pivotal role ethical and social issues play in decisions relating to antenatal screening for Down syndrome

b. ensure that the NSU and the Government have a clear picture of the concerns and complexity of the ethical and social issues relating to antenatal screening for Down syndrome

c. identify and explore the fundamental ethical and social issues relating to antenatal screening for Down syndrome

d. explore the ethical and social issues relating to the current practices for screening for antenatal Down syndrome

e. explore the ethical and social aspects specific to the implementation of an antenatal screening programme for Down syndrome if a decision is made to introduce a publicly funded screening programme in New Zealand.

Recommendations

The recommendations of the Social and Ethical Subgroup of the Antenatal Down Syndrome Screening Advisory Group emphasise the need for:

- **further collaborative debate and discussion around antenatal screening that draws on the knowledge and experience of different stakeholders and takes into consideration wider societal views**
- **certain prerequisites that are essential to be met if an antenatal Down syndrome programme is recommended, regardless of the screening method/intervention selected.**

Recommendations

The following recommendations are made to the Antenatal Down Syndrome Advisory Group.

1. **Note** that the complexity of the ethical and social issues underlying antenatal screening for Down syndrome should be acknowledged.
2. **Note** that there was a lack of consensus concerning fundamental ethical and social issues within the Social and Ethical Subgroup. This plurality of views is likely to reflect the situation in society as a whole.
3. **Agree** that serious consideration should be given to establishing a permanent Ministerial Advisory Committee to assist with the monitoring of broader ethical and social issues related to antenatal population screening programmes, and to consider new technologies or strategies in a timely fashion.
4. **Agree** that the current situation for Down syndrome screening is untenable because it is unsafe, inequitable, and should not continue
5. **Agree** that to encourage wider societal debate, the Government should consider whether public consultation should be carried out to determine the public's views on whether or not there should be a Down syndrome screening programme. If consultation does occur, then it should:
   a. include information on all options being considered in a way that is easily understood by a wide range of audiences and in accessible formats
   b. include information on how an antenatal Down screening programme might be implemented.

If an antenatal Down syndrome screening programme is recommended, regardless of the screening method/intervention selected, the following prerequisites should be met.

6. **Agree** that the Ministry of Health should explicitly state that the aim of antenatal screening is not to decrease the incidence of Down syndrome.
7. **Agree** that the aim of a programme would be to ensure that those women who choose to know about a risk of Down syndrome in their pregnancy have access to a programme with high standards of quality and safety that can provide them with an appropriate range of information and support them in their decision-making.
8. **Note** that, though the aim may not be to decrease the incidence of Down syndrome, the effect of any screening programme would inevitably be to decrease the incidence of Down syndrome, which is likely to cause concern in the disability community.
9. **Agee** that if an antenatal Down syndrome screening programme is introduced, all those directly involved in the provision of antenatal screening should meet national quality and safety standards and that these should be audited and monitored.
10. **Agree** that the Government should aim to reduce the potential for adverse effects related to the screening programme. It is noted that many of these adverse effects are present in the current screening practice. In particular:
    a. generation of anxiety during pregnancy
    b. fetal loss related to invasive procedures
    c. the potential for a negative impact on societal views of disability.

11. **Agree** participation in a screening programme should be voluntary. In order for this to be the case there needs to be consideration of:
    a. provision of information that is consistent with Right 7(1) of the Code of Health and Disability Services Consumer's Rights and that is available in a range of accessible formats
    b. comprehensibility of information
    c. free choice.
12. **Agree** that complete, high-quality information should be provided by first-line health care professionals familiar with antenatal screening. This should include information on the range of supports available for people with Down syndrome and their families. This may be challenging to achieve and will require considerable input in terms of developing resources and educating health professionals.
13. **Note** there may be difficulties in communicating complex information concerning risks and benefits in a short time period. These should be anticipated as far as possible and resources developed to assist consumers.
14. **Agree** individual choices should be unconditionally accepted and supported. This should include:
    a. provision of non-directive counselling services
    b. support for continuing a pregnancy
    c. support services around termination of pregnancy
    d. information about support and the range of services available to people with impairments and their families, including eligibility for support.

Introduction

This report is divided into the ethical and social considerations relating to:

- antenatal screening for Down syndrome
- the options for action in New Zealand, including whether a screening programme should be established.

In preparing this report, the Antenatal Down Syndrome Advisory Group Ethical and Social Subgroup (the Subgroup) were unable to reach consensus regarding a number of fundamental issues. This plurality of views is likely to reflect the range of opinion in society as a whole.

Applying an ethical framework

The Four Principles approach to ethics is the most commonly utilised method in health care ethics. It does not provide easy answers to complex ethical questions, but is a useful starting point for identifying and articulating ethical issues. The four principles are:

- beneficence (doing good)
- non-maleficence (not doing harm)
- autonomy (self-determination)
- justice (fairness).

The following section discusses issues relating to the ethical principles underlying antenatal screening. This overview is not exhaustive, but does highlight areas where views differ.

Autonomy

Reproductive autonomy

Modern maternity care involves allowing women and their partners considerable freedom to make their own choices and decisions with regard to pregnancy and childbirth. This commitment to reproductive autonomy has, in part, come about because of the wish to distance modern services from historical eugenic practices. However, because technology has advanced rapidly over the last few years, technical abilities for detecting Down syndrome and other fetal abnormalities have outpaced societal consideration of the extent to which parents should be able to make decisions concerning which children should be born. The fact that many women in New Zealand currently access Down syndrome screening suggests (but does not in itself justify) that this is a choice that many women consider they should be free to make.

Within the Subgroup there is consensus that reproductive autonomy should not be without limit. Most members believe that reproductive autonomy should extend to the ability to make decisions concerning continuation or termination of pregnancy when the fetus has Down syndrome. Others believe that this is beyond the limits of reproductive autonomy and should not be permitted. A large part of this disagreement centres on the issue of whether Down syndrome constitutes a 'harm' (see below). However, it should be noted that it is possible to hold personal views concerning Down syndrome but still believe that the reproductive autonomy of others in society should be respected.

Some women will choose to terminate their pregnancy because it is the best choice for them. Their decision around their eligibility to cope is recognised in the United Nations Convention on the Rights of the Child 1989 (CRC), sections 183 and 186, which refers to 'serious impact on the physical and mental health … or serious risk of physical or mental health of the fetus'.

Autonomy of the child

The moral status of the fetus is an ongoing source of debate and it is beyond the scope of this report to provide a detailed overview. There is a wide range of views, ranging from those who consider full moral status is acquired at conception to those who consider that a newborn baby does not have full moral status. Perhaps the most prevalent view is the gradualist approach, such that a fetus gradually acquires moral status as pregnancy proceeds. The legal status of the fetus is more clear-cut, with the fetus possessing few, if any, legal rights.

Non-maleficence and beneficence

The ethical principle of beneficence in essence describes the ethical obligation to do good. The ethical principle of non-maleficence in essence describes the ethical obligation to prevent or minimise harm. The most contentious issue in this section relates to whether or not Down syndrome should be considered a harm. Consideration of this issue is complex on a number of levels.

- The clinical features of Down syndrome are very variable, with some children being significantly more affected or impaired than others. This, of course, is something that is not known at the time of antenatal diagnosis.
- Consideration must also be given to the different experiences of children with Down syndrome in our society by comparison with the negative experiences of children born in the 1960s. That is, the society into which these children are born can be considered more positive and accepting of disability than was previously true.

- Cultural issues also need to be considered in this regard. For some ethnic groups within New Zealand society, the birth of a child with Down syndrome is considered a significantly more adverse event than within other cultural and ethnic groups. Cultural differences also need to be considered when taking account of the harm of termination of pregnancy. There was discussion that a Māori woman who chooses termination of pregnancy on the basis of a diagnosis of Down syndrome may experience shame.
- The issue that prompted most debate was the subjectivity of any assessment of disability. Several people within the Subgroup did not consider that the birth of a child with Down syndrome necessarily constituted a significant harm. While agreeing that any child has an impact on family life, they did not find the negative characterisation of Down syndrome appropriate as they consider the lived experience of people with Down syndrome to be the product of societal arrangements that may be more or less disabling. From this point of view, the harms potentially experienced by people with Down syndrome do not arise necessarily from their condition but from the various attitudinal and institutional barriers they encounter.
- Others in the Subgroup clearly considered that the birth of a child with Down syndrome did constitute a harm, while some members held their own views of the nature of disability associated with Down syndrome but accepted the fact that the views of other people may differ but should be respected. The autonomy of the child can be considered in terms of the child's potential independence and participation in society, and the potential supports to facilitate this. This is relevant to the perceived future quality of life of children born with Down syndrome. Such perceptions may influence parents' decisions about whether or not to continue with the pregnancy.
- The debate around the 'lived experience' of people with Down syndrome is closely related to discussion of the two predominant models of disability presented in the literature.
  - The 'social model' considers the lived experience of people with Down syndrome to be the product of societal arrangements that may be more or less disabling. From this point of view, the harms potentially experienced by people with Down syndrome do not arise necessarily from their condition but from the various attitudinal and institutional barriers they encounter.
  - The 'medical model' focuses more on the individual, their bodily impairments and limitations.

The distinction between the social and medical models is summarised in the International Classification of Functioning, Disability and Health (ICF):

> *The medical model views disability as a problem of the person, directly caused by disease, trauma or other health condition, which requires medical care provided in the form of individual treatment by professionals. Management of the disability is aimed at cure or the individual's adjustment and behaviour change. Medical care is viewed as the main issue, and at the political level the principal response is that of modifying or reforming health care policy.*
>
> *The social model of disability, on the other hand, sees the issue mainly as a socially created problem, and basically as a matter of the full integration of individuals into society. Disability is not an attribute of an individual, but rather a complex collection of conditions, many of which are created by the social environment. Hence the management of the problem requires social action, and it is the collective responsibility of society at large to make the environmental modifications necessary for the full participation of people with disabilities in all areas of social life. The issue is therefore an attitudinal or ideological one requiring social change, which at the political level becomes a question of human rights. For this model disability is a political issue*[9]

[9]http://www3.who.int/icf/intros/ICF-Eng-Intro.pdf

- The consequences of medical models may include information provision that is biased (in relation to parental decision-making), as described by the Nuffield Council on Bioethics thus: 'Parents may be presented with overly negative images of the future lives … which are not balanced by more positive information about the day-to-day lives of disabled people'[10]
- An additional perspective expressed by some members is that both the medical and social models, taken in isolation, are inadequate ways to assess the impact of disability on the lives of individuals and families within society, or to inform public policy.

Justice

The concept of justice involves consideration of issues of equity and fairness, and of the societal impact of decisions. Consideration of the application of this ethical principle requires us to look at the impact of antenatal Down syndrome screening on society rather than on the individuals involved.

The most significant issue in relation to this principle is the potential for the perpetuation or worsening of negative attitudes towards people with disabilities.

- Some members considered an antenatal Down syndrome screening programme would serve to reinforce the stigma around such conditions as Down syndrome and devalue the lives of people. Members who held this view considered that a screening programme could be ethically defensible only if it addressed the societally engendered harms experienced by people with Down syndrome and their families. Such a programme would not be biased towards termination of pregnancies when Down syndrome is diagnosed. It would allow for the preparation of the birth, and life, of children with Down syndrome. Prospective parents would be informed about the likely and/or potential experience of people with Down syndrome (tragedy not assumed) and about available or potential supports for people with Down syndrome and their families.
- There is general agreement that the aim of any proposed antenatal Down syndrome screening programme should not be to decrease the incidence of Down syndrome. However, it is noted that the effect of any screening programme would almost inevitably be to decrease the incidence of Down syndrome. This potential effect is likely to cause well-founded alarm among the disability community and to raise issues such as eugenics. This debate is also complex and the negative associations of these terms have sometimes obscured the underlying issues. Even defining what is meant by eugenics is difficult, although most people like to refer to some sort of coercive, government-imposed or supported programme to affect the type of people who are born. The term has been applied to antenatal screening programmes, particularly where there are concerns regarding the nature of information given to pregnant women and their ability to make a free choice. Some members are of the view that it is possible to apply the term eugenics to antenatal Down syndrome screening.
- Equity of access to services is also an important component of justice. This is not achieved with the current ad hoc screening, and could only be achieved through introduction of a screening programme.
- Equity of access to the range of disability support services needs to be considered.

[10]Nuffield Council on Bioethics. 2006. Critical Care Decisions in Fetal and Neonatal Medicine.

Costs

Funding of a government screening programme implies implicit support for screening. This report does not explore the ethical considerations of funding any potential screening programme options. However, we acknowledge that if a nationally organised antenatal screening programme is introduced, ethical consideration of the most practical, technical and biotechnical options will need to occur alongside cost considerations.

There are significant cost implications that overlap with ethical issues. For example, a cheaper programme option may not be able to be justified on the best medical evidence and could do more harm. A government-funded programme could be seen as a routine part of the antenatal care leading to an ongoing funding commitment. Some members are also concerned that funding a programme could potentially mean that funding is diverted from other maternity or disability services.

The Subgroup did not have available to it any cost estimates of a screening programme. It is anticipated that the implementation of a nationally organised screening programme would require training health practitioners, and providing information and access to counselling support. This was a particularly poignant issue for some members of the Subgroup.

If a screening programme is implemented, consideration has to be given to data collection, carrying out the functions of an auditing programme will involve costs.[11]

Language

Differences in values and attitudes can be illustrated in the language that is used to discuss antenatal screening for Down syndrome. This is particularly highlighted when contentious issues such as termination of pregnancy are involved. The way information is presented influences both how professionals introduce it and how patients use it. Shakespeare has described how medical discourse around disability utilises narratives of tragedy, in which antenatal diagnosis is constructed as a way of avoiding disaster, with value-laden terms such as 'burden, severe handicap, suffering, abnormality, and disorders' being used .

Options being considered

The full range of options is set out in the main report. The following section highlights some of the key ethical and social issues.

Option A: Do nothing

Continuation of the status quo, or the 'do nothing' approach, suggests that the current problem is either not significant enough to warrant intervention, or that the costs of intervening, or the risks of intervening, outweigh the risks of not intervening. A do nothing approach will result in the current problems continuing.

Autonomy

Under this option, autonomy is not well supported because the choice to participate in screening is not offered to all pregnant women. Where choice is offered, practitioners may not have a detailed understanding of the tests, verbal and written information for consumers may be poor quality, and the informed consent process may be compromised.

[11]Shakespeare, T. (1999) Losing the plot? Medical and activist discourses of contemporary genetics and disability. Sociology of Health and Illness 21, pp 669-688.

Non-maleficence

Current screening for Down syndrome is not best practice and is not the safest or most effective form of screening. This may lead to:

- a significant number of women being given false-positive results
- unnecessary referral for invasive diagnostic tests
- increased risk of fetal loss.

Current screening for Down syndrome may not always be accompanied by the offer of high-quality, non-directive counselling and support. This may increase the potential for adverse psychosocial effects.

Justice

One of the difficulties with the status quo is inequality of access.

Option B: Stop antenatal screening for Down syndrome

Reproductive autonomy

An increased number of women would be denied the opportunity to decide whether or not to participate in screening.

Non-maleficence

Private screening would continue with similar (if not greater) potential for the harms discussed in the section relating to continuing current screening (above). Women who could not access screening and subsequently had a child with Down syndrome may view this as harm.

Justice

Prohibiting the use of public funding to provide screening may lead to:

- a reduced perception that having a child with Down syndrome leads to suffering or disadvantage and is best avoided
- a reduction in negative attitudes.

If the option of redirecting resources to the provision of disability support services is considered, and this option involves not funding a screening programme, then the following would need to be considered:

- harms discussed above would also be present but there would be benefit to families who chose to continue a pregnancy with Down syndrome (as potentially better support for people with Down syndrome)
- prohibiting the use of public funding to support/provide screening and redirecting any savings to disability support services may result in:
  - inequality of access to screening
  - a reduced perception that having a child with Down syndrome leads to suffering or disadvantage and is best avoided
  - a reduction in negative attitudes
  - greater range of choices for parents, and prospective parents, in relation to raising a child with Down syndrome, or other condition
  - the provision of more adequate funding for medical and social needs to people living with disabilities.

Option C: Improve the quality and safety of antenatal screening for Down syndrome

Reproductive autonomy

This option is expected to ensure that consumers are supported to make informed decisions by providing clear requirements for health practitioner education and nationally consistent information resources. It should also provide better access to safer and more effective screening and is likely to reduce the potential for women to be offered diagnostic procedures without first having screening.

Beneficence

If benefit is construed as best possible detection of a fetus with Down syndrome to enable a pregnant women to be in a position to make a choice about whether or not to continue the pregnancy, then benefit is provided by this option. This would require a significant intervention to introduce a comprehensive, robust screening pathway.

Non-maleficence

This option has the potential to reduce potential harms (through fewer false-positive results, fewer referrals for diagnostic procedures and less fetal loss). Psychosocial harms may be reduced by the provision of high-quality counselling and support.

However, it does have the potential to increase harm for people with disabilities, and their families, by increasing the perception that Down syndrome is something to be avoided.

Justice

Equity of access to screening would be improved. The unintended consequences of this option are:

- the likely decrease in the incidence of Down syndrome – it is likely that if there is a reduction in the number of children born with Down syndrome this will have an impact on society's view and understanding of Down syndrome, which may lead to changes in society's willingness to support children with Down syndrome
- promotion of the perception that having a child with Down syndrome or other condition leads to suffering or disadvantage and is best avoided.

Any screening programme should acknowledge these potential adverse effects and where possible be aligned with the New Zealand Disability Strategy.

Conclusion

Any antenatal screening programme for Down syndrome must be founded on sound ethical premises. This summary has highlighted some of the areas of ethical controversy underlying the issue of antenatal screening. These ethical considerations are fundamental and should be considered before any other requirement in planning a screening programme.

If the Ministry of Health recommends to the Minister of Health that a nationally organised screening programme should be put in place, further ethical analysis of implementation issues will need to occur, especially because many of the ethical issues associated with antenatal screening relate to how it would be implemented. For instance, a government-funded screening programme would enhance reproductive autonomy only if prerequisites such as balanced information and unconditional support for all choices were met.

Members of the Subgroup were unanimous on one issue: current practice is untenable because it is unsafe. Because the Subgroup was not able to resolve some of the key issues identified, some members considered that wider societal debate should occur. Others felt that there was little chance of the issues being resolved society-wide, and that coupled with the unsatisfactory state of current screening practices this means there is urgency to improve current practices by introducing a programme.

Appendix 3: Nuchal Translucency Subgroup: Report to the Antenatal Down Syndrome Screening Advisory Group

Purpose

The purpose of this report from the Nuchal Translucency Subgroup (the NT Subgroup) is to provide a summary of the key issues and recommendations relating to current NT practice in New Zealand and in the event of a national antenatal Down syndrome screening programme being implemented.

Recommendations

The following recommendations are made to the Antenatal Down Syndrome Advisory Group.

a) **Agree** that comprehensive information needs to be obtained on NT practitioners in New Zealand before a programme can be instituted. This information should include:

- the number of sonographers and sonologists in each practice performing NT
- the number of these individuals who are accredited, and by which accreditation body
- the approximate number of NT exams performed per practitioner per year
- the geographic area covered by their practice, including the maximum distance traveled by practitioners and consumers
- the type and age of equipment
- the frequency of equipment upgrade
- quality issues (eg, IANZ accreditation of NT practices).

b) **Note** that the NT subgroup recommends that the Fetal Medicine Foundation (FMF) or the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) accreditation programmes are the most appropriate option for New Zealand at this stage.

c) **Agree** that an ongoing quality assurance programme needs to be instated if a national programme is established.

d) **Note** that the NT Subgroup does not support the practice of sonographers performing NT measurements using another practitioner's accreditation number.

e) **Agree** that further work needs to be carried out to determine:

- who will decide which accreditation programme is appropriate for New Zealand (ie, who will 'recognise' the accreditation)
- what criteria will be used to guide this decision.

f) **Agree** that the situation in smaller centres needs to be evaluated to address issues of access and quality.

g) **Note** that the Subgroup recommends the use of NT alone as the ultrasound marker for risk assessment. Newer and experimental techniques, such as nasal bone and tricuspid regurgitation, should not be included. However, we accept that NT in combination with serum screening will be a more efficient screening test and could be offered in association with the NT process.

h) **Agree** that the issue of co-payments will require further examination. To ensure equitable access to all women under a screening programme, the Ministry will have to consider increasing payments for NT scans so that co-payments are no longer required to cover costs.

i) **Agree** that guidelines will need to be developed for:

- referrals for the screening test
- sharing NT measurements with laboratories, where the NT forms part of a combined or integrated test, or vice versa if NT is performed after the maternal serum (blood) is taken
- reporting results to the woman and her LMC, GP or obstetrician – the format of the report is essential for clarity of the information, and needs to be consistent nationally.

j) **Agree** that further work will need to be carried out to determine:

- how screening data should be matched/connected to data relating to diagnostic tests and outcomes of pregnancy
- which data is collected at a national (programme) level and which data is collected locally at a provider or DHB level
- the requirements of information systems to record, share, match and transmit data
- the workforce/staffing requirements in relation to data management.

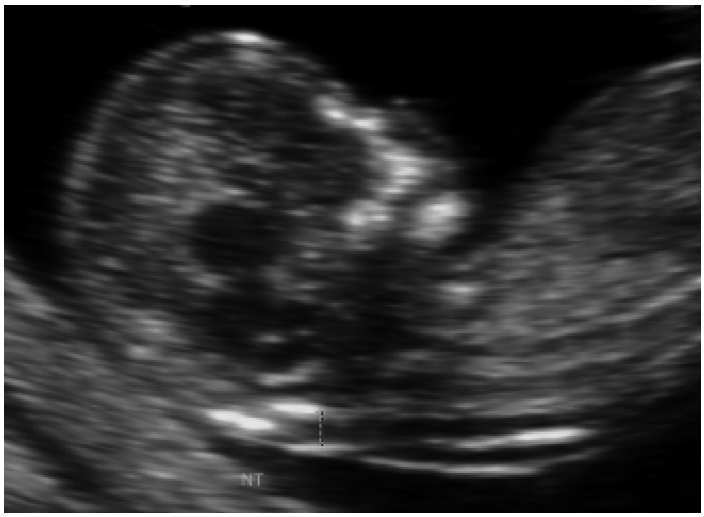
k) **Agree** that further work will need to be carried out to determine the elements of NT screening that require responsibility to be taken at a national level, at a DHB level, at a provider level and at an individual practitioner level.

The nuchal translucency scan

The scan is performed between 11 and 13 weeks 6 days of gestation, when the crown rump length (CRL) is 45 to 84 mm, and involves measurement of the fluid at the back of the fetal neck. The scan also enables:

- confirmation of viability
- confirmation of gestational age
- diagnosis of twins and multiple pregnancy, with chorionicity determination
- fetal anatomy, including skull and brain, spine, stomach, limbs, hands and feet, abdomen and bladder
- identification of up to 70 percent of lethal abnormalities and 38 percent of fetal major abnormalities
- detection of up to 77 percent of cases of Down syndrome with NT alone, and up to 85 percent combined with first trimester serum screening.

FIGURE A1: IMAGE OF THE FETUS AND THE NT MEASUREMENT



Increased nuchal translucency:

- is a strong marker for trisomy 18, trisomy 13, Turner syndrome and triploidy
- may also be associated with rarer chromosomal abnormalities
- is a marker for structural heart defects (which can be difficult to detect even at the 18–20 weeks scan), other structural abnormalities, and some rare genetic syndromes.

The current situation in New Zealand

Many practitioners performing NT in New Zealand (obstetricians, radiologists and sonographers) have been trained to perform NT measurements and are accredited by the Fetal Medicine Foundation (FMF) in London. Accreditation allows the practitioner to use the FMF software programme for the calculation of risk, using maternal age and NT measurement.

Information supplied by the FMF has identified in New Zealand:

- 115 individuals certified to perform NT
- 173 people either in the process of obtaining accreditation or who have previously been accredited but have failed audits and need to resubmit images
- 40 registered centres licensed to use FMF software.
- six registered practical trainers:
  - Deborah Andrews (Tauranga)
  - Robert Sim (Auckland)
  - Richard Davis (Auckland)
  - Pippa Kyle (Christchurch)
  - Rachael McEwing (Christchurch)
  - Jill Muirhead(Dunedin).

The FMF has expressed an interest in accrediting more trainers. A list of appropriate people has been recently forwarded to them for consideration.

It is important to realise that the above information appears to be incomplete, and probably under-represents the current level of accreditation in New Zealand. For example, the information provided by FMF does not align with data from HealthPac, whose claims data suggests there are approximately 200 practitioners making claims for NT.

Before we can address issues of access it is vital to establish who and where accredited individuals are so that an impression of geographic coverage and the number of operating NT practitioners can be identified. It was anticipated that some of this work would be covered in the workforce surveys, but these did not go ahead.

As a result, comprehensive information about current services is lacking, and it is recommended that this be obtained before a programme is instituted. This should include:

- the number of sonographers in each practice performing NT measurements
- the number of these individuals who are accredited, and by whom (FMF or similar)
- the approximate number of NT exams performed per practitioner per year
- the geographic area covered by their practice
- the type and age of equipment
- the frequency of equipment upgrade
- quality issues (eg, IANZ accreditation).

Practitioner qualifications and ongoing competency

It is likely that practitioners performing NT are registered by either the Medical Radiation Technologists (MRT) Board (in the sonographer scope of practice) or by the Medical Council (in either the diagnostic imaging scope of practice or in the obstetrician and gynaecology scope of practice). Although a significant number of practitioners are already accredited to perform NT, accreditation is not currently required and neither the Medical Council nor the MRT Board keeps a record of practitioners accredited to perform NT.

Under the proposed Section 88 Maternity Services Notice, accreditation to perform publicly funded NT will be compulsory as of July 2007. However, a roll-out period will be necessary as it will take some time to accredit all individuals and/or practices.

Currently FMF-accredited NT practitioners self-select images for annual assessment. These are submitted to the FMF and individual audit data is produced. Additionally, spot audits may be performed of individual practitioners by the FMF. The NT subgroup recommends that the FMF and/or RANZCOG accreditation programmes are the most appropriate option for New Zealand. Currently, to our knowledge, all credentialled sonographers and sonologists are accredited by FMF. Practitioners need to perform at least 30 examinations per year and provide their NT data and five images for annual audit.

For details of existing accreditation agencies, see Table A1.

A New Zealand-specific programme for NT accreditation could potentially be introduced in the future, as is the situation in the UK, where credentialling is currently managed by the FMF but with a view to central accreditation in the future. However, with our much smaller population and relative inexperience at nuchal accreditation, this step may be some years away from being feasible. At present all or most New Zealand practitioners are accredited by the FMF (although data is not readily available), and the process is easy to understand and compare from an audit point of view.

A New Zealand programme would probably still need to be run in collaboration with the FMF, as is the case in Melbourne (RANZCOG), at least in the medium term. It may be worth contacting the UK co-ordinators to discuss issues with implementation of quality assurance, etc. Ongoing quality assurance would eventually have to be done by whichever body takes ownership of the programme.

If an arrangement with the FMF were not developed, another approach could be the use of the scanning part of Alpha software.

The NT Subgroup does not support the practice of sonographers performing NT measurements using another practitioner's accreditation number.

Access to accreditation

There are a number of issues relating to access to accreditation training and funding for training. Because NT accreditation is now mandatory, under the revised Section 88 Notice, the Ministry may wish to consider meeting costs for training, which should not be substantial. Access issues are likely to improve if the FMF allows more trainers, which appears likely (see above).

Criteria to decide who will recognise/own accreditation

Further work needs to be carried out to determine:

- who will decide which accreditation programme is appropriate for New Zealand (ie, who will 'recognise' the accreditation)
- what criteria will be used to guide this decision.

Some of the accreditation programmes available internationally do not align with the standards and practice that are required for NT in New Zealand. An organisation in New Zealand needs to be charged with deciding which forms of accreditation are suitable and which should be 'recognised' in New Zealand. The following table sets out some of the options for the organisation charged with deciding which forms of accreditation are appropriate to New Zealand.

Table A1: Options for recognising NT accreditation in New Zealand

WHO COULD RECOGNISE NT ACCREDITATION?	WHY WOULD THIS BE A RELEVANT ORGANISATION TO RECOGNISE NT ACCREDITATION	BENEFITS/ ADVANTAGES OF THIS ORGANISATION RECOGNISING NT ACCREDITATION	COSTS/ DISADVANTAGES OF THIS ORGANISATION RECOGNISING NT ACCREDITATION
RANZCOG	RANZCOG is responsible for the standards of practice for obstetrics and gynaecology (O&G). A number of practitioners registered in the O&G scope of practice perform NT.	RANZCOG already has a role in setting professional standards.	A significant number of practitioners are registered with other professions / scopes of practice, such as sonography and diagnostic and interventional radiology. RANZCOG's role does not include setting standards for practitioners working outside of O&G.
The Royal Australian and New Zealand College of Radiologists (RANZCR)	RANZCR has a role in promoting standards of practice and in settling disputed points of practice and questions of professional usage.	RANZCR already has a role in setting professional standards.	A significant number of practitioners come under different colleges and professions (ie, RANZCOG, MRT Board, sonography etc). RANZCR's role does not include setting standards for practitioners working outside of radiology.
Joint colleges (RANZCOG and RANZCR)	RANZCR and RANZCOG could jointly be responsible for recognising accreditation as being appropriate for New Zealand.	Both colleges already have a role in setting professional standards. Both colleges represent practitioners who perform NT.	This may not include NT practitioners who are not medical practitioners (ie, sonographers), but at present all sonographers should be working under medical supervision (whether radiological or obstetric). Technical expertise regarding accreditation systems for NT would need to be developed, possibly with the assistance of an external advisory group.
Ministry of Health	The Ministry of Health's role includes monitoring the quality of publicly funded services (including NT) and facilitating collaboration and co-ordination within and across the sectors.	The Ministry of Health is not associated with any particular profession and could set standards for NT practitioners from a range of professions. The Ministry currently funds NT and should set minimum quality and safety standards for publicly funded services.	A system for considering the appropriateness/ suitability of particular accreditation systems would need to be established. Technical expertise regarding accreditation systems for NT would need to be developed, possibly with the assistance of an external advisory group.

WHO COULD RECOGNISE NT ACCREDITATION?	WHY WOULD THIS BE A RELEVANT ORGANISATION TO RECOGNISE NT ACCREDITATION	BENEFITS/ ADVANTAGES OF THIS ORGANISATION RECOGNISING NT ACCREDITATION	COSTS/ DISADVANTAGES OF THIS ORGANISATION RECOGNISING NT ACCREDITATION
NSU	If it is decided that a national screening programme will be implemented, the NSU has a role in providing national oversight, setting standards for the programme and carrying out monitoring and evaluation activities to ensure the programme is safe and of high quality.	The NSU could have broad oversight of the programme and is not associated with any particular profession or interests. It could be a neutral decision-maker. The NSU is already experienced at setting standards for national programmes, working with practitioners and colleges.	A system for considering the appropriateness/ suitability of particular accreditation systems would need to be established. Technical expertise regarding accreditation systems for NT would need to be developed, possibly with the assistance of an external advisory group.
A university or academic	Universities are not seen as having a role in making decisions relating to the operation of a programme or practice. It is not likely that a university or academic will be seen as the appropriate body to make decisions regarding the acceptance of various forms of accreditation.	Universities are a necessary link to ensure that NT accreditation is discussed at undergraduate level; however, this does not mean they are well placed to make decisions regarding the appropriateness of an accreditation system to NZ.	A significant number Universities have no authority to make these decisions, and are unlikely to be accepted by professionals and statutory authorities (Ministry of Health, NSU and registering authorities).
An accreditation body (like the FMF).	Accreditation bodies are likely to be seen as having a conflict of interest.	They will have a detailed knowledge of accreditation programmes, plus a wealth of data, and audit from practitioners already accredited by them.	Their role is not to determine which accreditation programmes are acceptable in NZ.

A New Zealand-specific accreditation programme for NT is unlikely to do away with the need to have a body responsible for deciding which forms of NT accreditation are accepted in New Zealand. Clearly the FMF already accredits a significant number of practitioners in New Zealand. There would need to be clear reasons why these forms of accreditation would no longer be acceptable. This is not to suggest that there should not be a New Zealand-specific programme; just that a New Zealand-specific programme could not be the only form of accreditation accepted. Please note, however, that the current FMF reaccredidation would not meet quality standards, and therefore the programme, once established, would be best placed to determine what these quality standards were and therefore the best processes to achieve them.

In order to determine whether an accreditation programme should be recognised for New Zealand, there will need to be a set of criteria to judge it against. One example of this type of criteria has been developed by the Society of Diagnostic Medical Sonography (SDMS) in Texas. SDMS is a non-profit association for ultrasound professionals. Criteria for New Zealand would have to be carefully considered, and include mechanisms for audit and monitoring.

The SDMS criteria for recognition of nuchal translucency credentialing organisations are as follows:

- a history of serving as a nuchal translucency credentialling agency for a minimum of one year
- a history of having credentialled a minimum of 1000 sonographers
- maintenance of an open dialogue with the sonography professional community
- recognition of the importance of operator qualifications in first trimester screening by requirement and provision of sonographer credentialling
- the requirement that non-physician operators credentialled in nuchal translucency first obtain a sonography credential by prerequisite education or experience and comprehensive examination, including obstetrical sonography content given by an SDMS-recognised credentialling organisation
- incorporation of sonographer member input into policies, procedures, organisational development, and testing and image review activities
- a standardised, criterion-referenced review of images with regular audit of consistency
- a standardised appeal and remediation process available for sonographer candidates
- regional accessibility of educational content and testing
- quality monitoring and regular feedback to participating sonographers.

Minimum volumes

The minimum number of NT examinations performed per practitioner per year will need to be established. Thirty studies per year is the minimum for FMF accreditation, which would seem reasonable, given our population. A smaller number of practitioners performing a larger number of studies would help to address concerns of quality. The situation in smaller centres needs to be evaluated to address issues of access. It may be preferable in future to ensure better-quality services in fewer locations.

Environmental, facility and equipment standards and maintenance

Further work will need to be carried out to determine suitable standards for:

- ultrasound machines used for NT, including the age of machines and the need to replace machines every three to five years
- quality assessment of ultrasound providers/practices (IANZ or similar)
- software used to calculate risk
- Multiples of the Median (MoMs). Given the relatively small number of NT examinations performed in New Zealand, this may be problematic, although a minimum number of measures per year (eg, 30 NT) should be achievable.

Other countries may already have developed standards for facilities and equipment that we can adopt as interim standards in the short term. Any standards developed will need to have the support of the colleges and practitioners.

New (experimental) ultrasound techniques

Decisions will need to be made regarding the techniques that are used to calculate an individual's risk, including whether only the NT and crown rump length (CRL) measurements are used, or whether other 'soft markers' are also used, such as the nasal bone measurement. Fetal nasal bone and tricuspid regurgitation work still appears experimental and varies by ethnicity. We are not aware of any studies done in the New Zealand context (this may be worth addressing in the future). FMF requires accreditation for nasal bone assessment in much the same manner as NT measurement, and this part of the software programme cannot be accessed if the sonographer/sonologist is not credentialled. In the meantime, the NT subgroup recommends the use of NT for risk assessment alone.

Information for consumers and communities

Under the Code of Health and Disability Services Consumers' Rights, consumers have a legal right to be fully informed. This requires providing clearly written and easy-to-understand information to health practitioners and consumers (pre-test counselling, post-test counselling and educational resources). An information sheet for consumers will need to be developed at a national level so that consumers are provided with consistent information. This could provide links to other sources of accurate information, such as the FMF website and brochures.

Written information will need to include:

- details of Down syndrome, including background information (eg incidence, potential associated health problems in people with Down syndrome)
- potential harms and benefits of screening, NT and diagnostic tests
- the role of NT in the screening pathway (as a stand-alone screening test or as part of combined or integrated screening)
- an explanation of the NT screening test, what it is, how it is done, the percentage of cases that are detected (detection rate) and the percentage of unaffected individuals who will have positive results.
- what happens if the screening test is positive, options for diagnostic testing and options if there is a positive diagnosis of Down syndrome
- contact details of available support (Wald 2006).

Written information/education will need to be developed for health professionals. This information will need to include:

- how to manage pre- and post-test discussion and the informed consent process
- the role of the LMC
- an explanation of the screening pathway
- how results will be provided, and the report format
- detailed information about the risk assessments and what they mean.

Timely availability of NT

There needs to be consideration given to issues relating to access to NT, including:

- identifying parts of New Zealand where there is a significant travel distance/time in order to access NT
- populations in New Zealand who may have difficulties accessing NT due to cost (travel, co-payments, etc)
- the issue of co-payments, which will require further examination – to ensure equitable access to all women under a screening programme, the Ministry will have to consider increasing payments for NT scans so that co-payments are no longer required to cover costs
- lack of access to an accredited practitioner.

NT in relation to the rest of a screening pathway

Guidelines will need to be developed for:

- referrals for the screening test
- sharing NT measurements with laboratories, where the NT forms part of a combined or integrated test, or vice versa if NT is performed after the maternal serum (blood) test.
  Note: one-stop shops (the OSCAR model adopted in the UK), where the woman has NT and first trimester bloods performed and receives risk assessment at the same visit, could be considered (unless an integrated model is adopted in the final programme and a further visit is required). Optimally, a reduction in the number of required visits would go some way to alleviating issues around access and affordability.
- reporting results to the woman and her LMC, GP or obstetrician.

In order for a screening programme to be successful, a co-ordinated approach is required, including:

- high-quality service provision
- timely availability and appropriate integration of screening services within diagnostic and follow-up services
- effective monitoring against defined policies and standards
- clear lines of accountability
- outcome information stored centrally.

Effective monitoring

A formal monitoring and evaluation plan will need to be developed for the proposed programme, including monitoring and evaluation of NT. If the Minister of Health decides not to implement a nationally organised antenatal Down syndrome screening programme, there is still a need for monitoring and evaluating NT practice in New Zealand.

Outcome data collection and auditing are essential, and ideally would be centralised.

The data required to monitor and evaluate NT will need to be identified. This could include, for example:

- the number of women referred for NT screening
- the number of women accepting/choosing NT screening
- the number of women receiving a low-risk NT result

- the number of women receiving a high-risk NT result
- the number of women receiving a high-risk result who are offered a diagnostic test
- the number of women accepting the offer of a diagnostic test
- the number of women receiving a low-risk result who choose to have a diagnostic test
- the outcome of pregnancies with a high-risk result
- all cases of chromosomal abnormality in women who are screened (ie, either high-risk or low-risk NT).

Further work will need to be carried out to determine:

- how screening data should be matched/connected to data relating to diagnostic tests and outcomes of pregnancy
- which data is collected at a national (programme) level and which data is collected locally at a provider or DHB level
- the requirements of information systems to record, share, match and transmit data
- the workforce/staffing requirements in relation to data management.

Clear lines of accountability

Screening programmes require clear national direction, effective planning and organising, and a good understanding of the nature of the work being undertaken. While some aspects of screening are best managed at a local or regional level, some high-level screening functions are best delivered nationally in order to prevent fragmentation of services and to ensure there is a cohesive overall approach to screening in New Zealand.

Further work will need to be carried out to determine the elements of the NT screening that require responsibility to be taken at a national level, at a DHB level, at a provider level and at an individual practitioner level. It has been suggested by some members of the Subgroup that New Zealand may be too small to have its own accreditation system and therefore may need to establish a list of approved accreditation bodies (ie, whoever is responsible for establishing a screening programme in New Zealand would have to decide which accreditation bodies they would recognise). Note that not all of the Subgroup agree that this is the case and some members would support a New Zealand accreditation system.

Funding

Funding arrangements for NT need to be investigated, and it needs to be determined whether funding arrangements under the Section 88 Maternity Services Notice will be adequate.

Table A2: Existing accreditation agencies

NAME	WHERE	WHAT IT SAYS IT DOES
Fetal Medicine Foundation	UK	The Fetal Medicine Foundation (FMF) is a registered charity that promotes research and training in fetal medicine. It promotes screening for Down syndrome at 11–13 weeks 6 days by nuchal translucency (NT) or a combination of NT and maternal serum biochemistry. It has set up a process for certification in the 11–13+6 weeks scan to ensure that those performing this ultrasound examination have been adequately trained to do so and that high standards of performance are maintained by continuous education and audit. Certification involves a day-long course, incorporating a written exam, a practical exam, and the provision of 10 satisfactory images to be sent to the FMF. Once certified, the doctor/sonographer is entitled to receive the FMF software for the calculation of risk of Down syndrome by a combination of maternal age, NT measurement and first-trimester maternal serum free ß-hCG and PAPP-A. A risk assessment can be calculated with or without these analytes. The accredited individual will be included in the FMF list of 'accredited sonographers' and the hospital/clinic of the doctor/sonographer will be included in the FMF list of 'registered centres'. Ongoing certification and use of the software is contingent on the provision of NT data and images (currently 5) by each sonographer for the purposes of annual audit. Spot audits are also randomly performed. The FMF also offers Certification of Biochemical Laboratories. http://www.fetalmedicine.com/f-fmf.htm
FMF with RANZCOG	Australia	The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) mission statement states dedication to maintaining the highest possible standards in obstetrics and gynaecology in Australia and New Zealand. RANZCOG, in conjunction with the FMF, has set up a process for certification of the 11–14 week scan. Use of FMF software and ongoing certification is conditional on candidates providing their measurements for the purpose of audit to RANZCOG, and via them to the FMF. Initial certification requires 50 consecutive images to be provided within a 3-month period, which may be difficult to achieve for many New Zealand practitioners. http://www.ranzcog.edu.au/nt/regulations.shtml
Nuchal Translucency Quality Review as part of the Society for Maternal-Fetal Medicine	United States of America	This group runs credentialling courses and is supported by the American Institute of Ultrasound in Medicine. They offer three programme track options: A: standard – educational course, exam and image submission, for providers who have had little or no prior NT training B: previously trained – exam and image submission only for providers who have prior NT training but have not participated in an ongoing quality review programme C: currently credentialled – exam and image submission is waived for providers who are previously credentialled by a number of other organisations Each participating laboratory plays a role in the monitoring process by providing sonographer-specific NT measurement data to Nuchal Translucency Quality Review. These data are used for ongoing epidemiologic monitoring to ensure that the NT measurements submitted by providers to laboratories are accurate. https://www.ntqr.org/SM/default.aspx

Appendix 3 References and bibliography

Carroll S, Soothill P, Abdel-Fattah SA, et al. 2002. Prediction of chorionicity in twin pregnancies at 10–14 weeks of gestation. *BJOG* 109: 182–6.

Carvalho M, Brizot M, Lopes L, et al. 2002. Detection of fetal structural abnormalities at the 11–14 week ultrasound scan. *Prenatal Diagnosis* 22: 1–4.

Cedergren M, Selbing A. 2006. Detection of fetal structural abnormalities by an 11–14 week ultrasound dating scan in an unselected Swedish population. *Acta Obstetricia et Gynecologica Scandinavica 85: 912–15.*

Makrydimas G, Sotiriadis A, Ioannidis JP. 2003. Screening performance of first-trimester nuchal translucency for major cardiac defects: a meta-analysis. *American Journal of Obstetrics and Gynecology* 189: 1330–5.

Saltvedt S, Almstrom H, Kublickas M, et al. 2006. Detection of malformations in chromosomally normal fetuses by routine ultrasound at 12 or 18 weeks of gestation: a randomised controlled trail in 39,572 pregnancies. *BJOG* 113: 664–74.

Snijders R, Noble P, Sebire N, et al. 1998. UK multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10–14 weeks of gestation. *Lancet* 351: 343–6.

Souka AP, Pilalis A, Kavalakis Y, et al. 2004. Assessment of fetal anatomy at the 11–14 week ultrasound examination. *Ultrasound in Obstetrics and Gynecology* 24: 730–4.

Wald N. 2006. Information leaflets in medical screening. *Journal of Medical Screening* 13: 109.

Appendix 4: Maternal Serum Screening Subgroup: Report to Antenatal Down Syndrome Screening Advisory Group

Executive summary

Considerable concern was raised in the sector following the letter from the Ministry of Health to health care providers discouraging the use of age screening for Down syndrome and other conditions.

The Maternal Serum Screening Subgroup of the Antenatal Down Syndrome Screening Advisory Group endorses the Ministry of Health's conclusion that screening on the basis of age is not acceptable in 2006 and that there are significant inequalities and risks associated with current screening.

The Subgroup acknowledges that the screening with the best sensitivity and specificity is a combination of nuchal translucency (NT) and biochemical markers in the first and second trimesters, but that consideration of the evidence for the best method of screening and the introduction of a screening programme is likely to take considerable time.

NT is a better screen than age and is currently available to some women. Second trimester serum screening is also a better screen than age and offers an alternative to those women for whom NT is not accessible. The Subgroup considers the introduction of second trimester biochemical screening (MSS2) to be feasible and recommends the introduction of this screening initially to those women unable to access NT screening.

There are risks associated with introducing an interim step, however, including:

- the lack of information about the number of women eligible and likely to accept screening;
- the ability of the workforce to cope with: the additional workload of explaining tests, diagnostic testing (cytogenetics) and post-test advice and counselling
- the potential for confusion, both in the community and among maternity health care professionals.

The benefits associated with introducing an interim step are:

- the concerns raised in the Ministry of Health's letter will be addressed
- some of the inequalities of NT-based screening will be addressed
- it will inform the implementation of a complete programme.

Background

In May 2006 Dr Don Matheson (Deputy Director-General Public Health) wrote to the maternity sector advising that maternal age alone is not a sufficient screening mechanism for Down syndrome. The letter suggested offering screening by nuchal translucency (NT) to women who present in the first trimester and serum screening to those who present in the second trimester. The former test is government-funded (although there is frequently a co-payment); the latter is not.

The Maternal Serum Screening Subgroup of the Antenatal Down Syndrome Screening Advisory Group was asked to consider as an interim step the feasibility of offering funded second trimester maternal serum screening (MSS2) to women for whom NT screening is unavailable.

The Subgroup acknowledges that the screening with the best sensitivity and specificity is a combination of age, NT and biochemical markers in the first and second trimesters, but that consideration of the evidence for the best screening and introduction of a screening programme is likely to take considerable time (years rather than months).

The Maternal Serum Screening Subgroup comprised Ms Norma Campbell, Mr Clive Felix, Prof Pippa Kyle, Dr Rachael McEwing, Dr Kate Neas, Ms Anne O'Connor, Prof Peter Stone, Dr Dianne Webster and Dr Jonathan Wilcox, supported by Ms Angie Partridge from the NSU.

Considerations

The Maternal Serum Screening group supports the Ministry of Health's conclusion that present screening in New Zealand is highly inequitable (due to differences in test availability and affordability) insensitive and non-specific (and therefore unsafe). The group further supports the Ministry's recommendation that (until the Advisory Group reports its conclusions) NT be offered to all women, as this screening is more sensitive (at the same specificity) than the currently most used screening (maternal age alone), and MSS2 to women who present too late for NT.

However, the group has reservations about this recommendation for the following reasons.

- NT is not available in all regions of New Zealand.
- There is frequently a co-payment on NT, and some women cannot afford this.
- The charge (currently $75) for MSS2 is not affordable for some women.
- There are currently some quality assurance concerns relating to nuchal scans. These are being addressed and will be implemented with the Section 88 Maternity Notice in July 2007.

An interim step additional to that recommended by the Ministry of Health may cause confusion if the final programme does not incorporate the interim arrangements. Mitigating this concern is the likelihood that both MSS2 and NT would be included as part of a complete screening programme (based on published data and practice in other jurisdictions).

The group gave consideration to the following issues in determining the feasibility of offering paid MSS2 to women who present too late for NT.

1. Is an interim solution practicable?
2. Who should offer the screening, and who should it be offered to?
3. When taking a blood sample, there is a need to consider:
    - who it is taken by
    - gestation limit
    - cost
    - location
    - shipping.
4. With respect to laboratory testing, there is a need to consider:
    - capacity
    - analytes
    - reporting.
5. Will there be capacity issues for diagnostic testing?
6. Will there be equity of access?

Practicality of an additional interim solution

The Subgroup considered the option of continuing the status quo (NT where this is accessible and age screening and/or self-funded MSS2 otherwise) until such time as a full screening programme could be implemented. Superficially, continuing with the status quo seems attractive, but there is already considerable concern (among specialists, LMCs and GPs) generated by the current situation and by the Ministry of Health's correspondence with the health sector.

Offer of screen

The group recommends that MSS2 be offered to women who have been unable to access NT screening due to one of the following reasons:

- NT services are not available in her region
- NT services available charge a co-payment which she is not able to pay
- she presents after the gestational cut-off (13 weeks 6 days) for NT screening.

Ideally, the blood should be drawn by 17 weeks 6 days, which is optimal for the timing of accessing diagnostic tests, receiving the results of diagnostic tests and making decisions about the pregnancy. There will be some women who are screened later than this, however.

A dating scan improves the precision of MSS2 and eliminates a small number of false-positive tests in women who have misdated pregnancies. Screening will be offered by the health care professional to whom the woman presents. This may be a midwife, GP or obstetrician.
The information offered to women at this stage should be simple, because this is an interim step. New single-page sheets are required for both women and caregivers pre-screening.

Taking blood samples

Women accepting screening will have their blood taken by hospital and community laboratories. The laboratories have well-established processes for sending samples to LabPlus. Some community laboratories currently charge for blood collection. This is the case in Auckland, where women are charged $15. A courier charge is sometimes also levied.

Screening laboratory

Screening should be by a four-analyte test (free ß-hCG, unconjugated oestriol, alphafetoprotein and inhibin-A). Providing an automated method is used, singlet screening will provide sufficient precision. Risk calculation will be done by the Alpha programme, using a cut-off of 1:300. Although the cut-off may be different in a final programme, the use of 1:300 (as used for NT screening) will minimise confusion. Individual risks and prior risk (age-related risk) will be provided. Other abnormal findings will be reported. A turnaround time for results in-laboratory must be specified to ensure timely reporting. Three days is suggested to make 90 percent of all results available to women in less than one week. There is sufficient capacity at LabPlus to accommodate any increases that may arise.

It should be noted that risk estimates are a continuum, and the decision about which level of risk is sufficiently high to merit the offer of a diagnostic test is influenced by a number of factors including:

- the acceptable sensitivity and specificity of the test
- possible confusion with the cut-off used for other tests also in use
- capacity of diagnostic testing.

Diagnostic testing

Currently, diagnostic testing is offered to women who have positive screening test results (older than 35 years or NT risk assessment $\geq$ 1:300). The gold standard test is a karyotype on tissue obtained by amniocentesis or chorionic villus sampling, although sometimes faster tests are used. The faster tests (eg, FISH) are funded by some DHBs in some circumstances, otherwise the women pay. The more rapid tests detect fewer abnormalities and are currently always used in conjunction with a karyotype.

The group is agreed that the only diagnostic test to be offered following a positive maternal serum screen should be a karyotype.

There are diagnostic testing laboratories in Auckland, Hamilton, Wellington and Christchurch. Presently three of the four centres have no capacity for increased workload, although this situation may improve over the next few months. The remaining laboratory could temporarily absorb some increase in workload, assuming staffing levels remain unchanged. There are well-established mechanisms for moving samples, results and payments between laboratories.

Reporting

Laboratory results will be reported to the requesting clinician. New single-page sheets for women and their caregivers giving the options for further action following positive test results are required.

The laboratory risk cut-off (the risk above which a diagnostic test will be offered) will be 1:300 as currently reported with NT, because there is concern that different cut-offs would be confusing for both consumers and health care professionals (see point 5 above). Any screening programme will consider risk cut-offs for each test and whether all options should be harmonised by cut-off (giving differing sensitivities) or by sensitivity (when each test will have a different risk cut-off).

A consideration for a full programme is whether results are reported with individual risk estimates or simply 'screen positive' or 'screen negative', or similar terminology. The arguments for the former are that this is the status quo (women know their age risk and NT screens are reported this way), and women are said to prefer a specific number.

Arguments for the latter are that different tests can have different cut-offs to give them the same sensitivity without creating confusion, and NT and biochemical tests give risk estimates which have inherent inaccuracy, so that giving numbers (eg 1:367) implies a precision the tests don't have.

A further consideration of reporting is whether individual risks given should be estimated at term or at the gestation of testing (abnormal pregnancies have a higher miscarriage rate, and hence there will be a higher risk estimate given if estimated at an earlier gestation).

Guidance is sought for the terminology to be used in reporting. A risk will be given, and diagnostic testing will be offered above the risk considered to be the screening cut-off (for NT this is 1:300). How should results above the cut-off (i.e. positive screening tests be reported? Suggestions include:

- higher/lower risk
- higher/lower chance
- diagnostic test recommended/not recommended
- positive/negative risk
- screen positive/negative
- increased/decreased risk
- high/low risk.

Women who have an increased risk result need timely access to high-quality counselling services in order to discuss the results and plan further diagnostic testing if the woman desires. In the interim this support will be provided by genetic services and fetal maternal medicine services. Due to the centralised nature of these services, telephone counselling may be needed for those women who cannot access any of the four centres. Information about how to contact these services will be provided on the information sheets for LMCs. The impact of the change in practice on these services can not easily be quantified, and extra services may be required if there is a significant increase in demand.

Monitoring

There are significant unknowns in the introduction of interim second trimester screening for women unable to access NT screening, so there should be frequent careful monitoring of the screening system in order to adjust the system if problems arise. The group suggests monthly reporting of the number of:

- women screened, by region
- women screened, by age
- women screened, by gestational age
- women screened who have had a dating scan, and who have not had a dating scan
- positive tests.

Continuing work

The group suggests the NSU provide the following information in order to help assess the impact of an interim stage in relation to funding and workforce implications:

Work relating to the size of the programme

This includes:

- the number of women presenting for the first time at 14–18 weeks
- NT not being accessible due to distance – the number of women in highly rural areas or areas where no NT screening is currently available
- NT not being accessible due to cost – the number of women who have a Community Services Cards, by age
- co-payments, by region and locality.

An estimate of the number of women will be done based on uptake rates of between 55 and 85 percent. Data will be used to inform test costing and pricing and the capacity for diagnostic testing.

Work relating to ancillary costs

- Provide an estimate of the number of women who do not reliably know the date of their last menstrual period and therefore need a dating scan additional to other scheduled scans, and the cost of these.
- Are karyotypes on the laboratory tests schedule?
- Which community labs might charge for taking tests?

Work relating to the introduction of the programme

- Information sheets for women (pre-test, post-positive test) must include information about the scan and that an additional scan to obtain gestational age for the screen should not replace the routine 18–20-week anomaly scan.
- Information needs to be provided for those people who request tests.
- A monitoring system needs to be established.

Risks

Sector concerns

Conversations with the sector have highlighted the following concerns:

- covering costs associated with the programme aside from the actual test (eg, diagnostic testing, post-test advice and counselling)
- the potential for confusion in having an interim step and then introducing a complete programme (this concern applies also to the current situation with NT).

Concerns of the Subgroup

- It is anticipated that all women who have not decided to terminate their pregnancy, or who have not miscarried by eight weeks gestation, would be offered a government-funded screen (either NT if before 13 weeks 6 days or MSS2 if after this and before 18 completed weeks, as per the criteria above). The exact number of women in either category (NT or MSS2) is unknown, and it is difficult to assess how affordability or accessibility of NT will affect numbers. It will ultimately be the responsibility of the LMC or GP to identify such women and offer them the appropriate funded screen.
- The number of women likely to accept screening is unknown – if the Subgroup's recommendation about including women who cannot afford the co-payment for NT in the cohort is accepted, this will increase the number of eligible women by an unknown figure.
- Availability of diagnostic testing (the capacity of cytogenetics laboratory services) – the risk may depend on payment. If laboratories are claiming under the laboratory test schedule for samples taken in the community, revenue will follow increased test numbers; if the tests are being done under the bulk non-scheduled testing, no additional revenue will be available to laboratories to compensate for the additional testing.
- The availability of tertiary support for health care professionals and counselling following complex positive diagnostic test results (ie, clinical genetics services) is a concern.
- If NT and serum screening are aligned by cut-off, there is disquiet over the better detection rate of serum screening. If they are aligned by detection rate, there may be confusion over the different cut-offs.

Risk mitigation

The Subgroup considers that risks could be mitigated by:

- close monitoring
- regular communication with the sector.
- an enhanced education programme incorporating new, clear information sheets
- timely development of a quality national programme that would address these issues.

Benefits

The Ministry of Health's letter raised concerns about the availability of screening to all women, especially given that age screening on its own is not recommended. The introduction of MSS2 will address some of those concerns, because it reduces inequalities by ensuring screening is available to women for whom NT is not currently available. This includes those presenting too late for NT, those for whom NT is not available in their region, and those for whom the co-payment presents a significant financial barrier.

The introduction of MSS2 in a limited way, with close monitoring, will inform the introduction of first and/or second trimester screening as constituents of a complete screening programme. The laboratory aspects of MSS2 will be the same as for a complete programme and so they can be a forerunner for laboratory aspects of a future programme. Each step that is introduced will provide better education for the community and providers about screening.

Conclusions and recommendations

1. The Serum Screening Technical Subgroup concludes that introduction of MSS2 to complement NT as an interim measure is feasible.
2. The Serum Screening Technical Subgroup recommends that fully funded MSS2 be introduced for those women unable to access NT screening.
3. The Serum Screening Technical Subgroup notes that some information about the number of eligible women and uptake cannot be known at the commencement of this interim step towards a programme, and so close monitoring to quickly identify points of stress in the system and provision for appropriate response to the stresses is essential.


MINISTRY OF
HEALTH
MANATŪ HAUORA