

Report to the

National Screening Unit

Assessment of Antenatal Screening for

Down Syndrome in New Zealand

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Executive Summary

The purpose of this report is to provide the National Screening Unit of the Ministry of Health information on the current state of the antenatal screening for Down syndrome in New Zealand and how a co-ordinated approach to such screening could be implemented. In achieving these aims the report can provide a basis on which to make improvements in a situation that currently all groups surveyed defined as needing urgent change.

- Currently, the antenatal screening for Down syndrome (Trisomy 21) is ad hoc, based mostly on ultrasound scanning and does not reflect the best evidence or best practice internationally.
- There is a lack of clarity regarding exactly what is being screened for, Down syndrome or other fetal abnormalities.
- This project identified that the antenatal screening for Down syndrome satisfies the criteria for a screening programme. Screening programmes have quality and safety requirements. These are not being met by the current approaches to Down syndrome screening in New Zealand.
- Current literature supports a coordinated, multi-test approach to Down syndrome screening.
- This project convened a working group from representative consumer and professional organisations to discuss:
 - the current situation regarding Down syndrome screening in New Zealand
 - issues pertaining to each organisation regarding antenatal screening
 - the design of the survey which produced the data for this report
 - issues about the implementation of a screening programme for Down syndrome.
- A postal survey of health practitioners (midwives, general practitioners and obstetric specialists) and providers (ultrasound practices and District health Boards) was developed, piloted through the working group and distributed by post with freepost return envelopes and e-mail via the following organisations;
 - Royal Australian and New Zealand College of Obstetricians and Gynaecologists
 - South Auckland Maternity Care Limited (SAMCL)
 - New Zealand Branch of the Royal Australian and New Zealand College of Radiologists.

General Practitioners were selected from a list provided by the Ministry of Health with every fifth doctor on the list being sent a survey.

Midwives were contacted from the internet based list of Lead Maternity Carer midwives.

The results were collated by the Research Assistant and entered onto a database.

- The response rate overall for the survey was 35% of those surveyed. The regional distribution of respondents reflected the regional distribution of births from the national total births.
- Key findings from the survey are as follows.
 - Unanimous view that current system of antenatal screening for Down syndrome is not best practice and is not sustainable.
 - Around 50% of practitioners would offer screening to all women.
 - Around 46% of practitioners offer screening on a selection basis, generally based on maternal age or previous abnormality.
 - GPs are less likely to offer screening than other practitioners (less than one third offer screening to all).
 - Responses to the survey have identified a clear need for education of practitioners about screening.
 - Accurate data collection and monitoring of both screening and diagnostic tests currently does not occur.
 - There is equal support for a screening programme with combined first trimester Nuchal Translucency (NT) scan and maternal serum testing and integrated testing (first trimester scanning and first and second trimester maternal serum testing).
 - When practitioners were presented with information on test performance there was little support for NT scanning alone as the screen leading to invasive testing.
 - The issue of screening specifically for Down syndrome or for other detectable anomalies in addition remains unresolved and requires wide debate.
- All of this report's recommendations, except developing and implementing a screening programme for Down syndrome, could be proceeded with now.
- Failure to fund second trimester screening is disadvantaging a group of women who book late (after the first trimester of pregnancy).
- The exact form of a national screening programme would need to be resolved by a working party with the likely outcome being a number of options based on at least the following considerations:
 - scope of screening programme
 - timing of presentation for screening in pregnancy
 - findings at the first test episode
 - consumer preference.

Recommendations

Based on all the information gathered during this project, the following recommendations are made.

It is recommended that:

1. the current situation of age and NT screening leading to invasive testing be urgently reviewed with the view to stopping this as a screening practice.
2. on the basis of this report the National Screening Unit recommend to the Ministry of Health that not only is current practice not best practice but that there is general support for change to achieve a 'best practice' for New Zealand.
3. second trimester maternal serum testing be funded, promoted and offered as well as NT scanning as an interim measure, whilst other components of a programme are being developed. This would be possible within the current infrastructure.
4. a working party be established to develop and implement a programme suitable for New Zealand.
5. a system of data monitoring be established now for both screening and diagnostic tests.
6. sonographers and sonography services be accredited, have New Zealand standards developed and individual MOMs for NT scanning be established.
7. a knowledge pack regarding antenatal screening for Down syndrome, with test characteristics clearly and simply displayed, be disseminated as a matter of urgency given the level of knowledge of the surveyed practitioners.

1. Introduction

The substance of this report is a detailed analysis of the current status of antenatal screening for Down syndrome (Trisomy 21) in New Zealand.

Clinicians have been aware for some years that current practice is neither particularly effective at detecting fetuses with Trisomy 21 nor is practice consistent with the best evidence available.

At present, Trisomy 21 cannot be diagnosed without resorting to invasive testing, which has many implications for the pregnant woman, fetus and health services. Therefore a number of approaches are used in attempts to screen for, or predict the likelihood that a given fetus has the chromosome abnormality. Those pregnancies determined to be “high risk” or high probability of Trisomy 21 or be screen positive are then offered invasive or diagnostic testing. For a predetermined risk cut off, each screening test will have a detection rate and a false positive rate (more pregnancies tested than actually have the condition) and a false negative rate (an affected pregnancy that is not tested because the screening test was screen negative). In New Zealand, as distinct from many other countries with screening available, modern screening began by using ultrasound techniques and has gradually been widely offered but not in the context of a coordinated screening programme. Previously maternal age had been used as the screen with women 35 years and older being able to have publicly funded invasive testing, usually amniocentesis and fetal karyotyping.

Mounting evidence in New Zealand has shown that not only is this becoming an increasingly expensive and labour intensive approach but has failed to reduce the numbers of babies born with undiagnosed Trisomy 21. This can be due both to failure to offer screening and/or the false negative rate of current screening processes. At the same time, best estimates would suggest that more normal pregnancies may be adversely affected by amniocentesis than the numbers of Down syndrome fetuses detected. Prior to the widespread use of ultrasound screening by NT scan, second trimester maternal serum screening had been piloted and introduced as a clinical service but subsequently public funding for this screening was withdrawn.

It is with this background of uncoordinated and relatively ineffective screening for Trisomy 21 that a number of clinicians raised concerns that the current situation in New Zealand was unsatisfactory. Following the publishing of the SURUSS report (Wald 2003) and a number of clinical meetings around the country, concern grew that some action was needed. After a meeting of interested parties hosted by the Ministry of Health in 2004, it became clear that all involved in the area believed that change was needed to the current screening approaches.

After discussions between the Ministry of Health (National Screening Unit) and the principal author of this report, a project was designed to:

- Review the literature and “best practice” of antenatal screening for Down syndrome
- Assess current practice in New Zealand
- Discuss issues of implementation of a New Zealand screening programme based on the research into current practice and the views of health practitioners and consumers
- Survey and collate information on screening for Down syndrome that could be used in a New Zealand context

After a scoping exercise developing the terms of reference and appointing a research assistant, the project commenced June 2005.

The results of the surveys and analysis of qualitative comments from two face to face meetings, which included health practitioners and representatives of consumer groups, form the basis of this report.

Information for women and health practitioners on screening has also been collated but will be included in a later document, which would be more relevant to an implementation phase. The results from the surveys described in this report do inform in detail our current practice, the understanding of screening and guide the development of effective screening approaches for New Zealand.

Inevitably a number of controversies have been found in approaches to screening with differing opinions on testing for what and when and during the implementation of a programme those controversies identified in the report and others that may arise will need to be addressed.

A formal cost analysis has not been done due to time and funding constraints. There are data available from the SURUSS study to guide the cost for each case of Down syndrome detected. Variations on the integrated screening programme as described in SURUSS could be modelled in a New Zealand context.

2. Literature review

2.1 Introduction

This literature review is not intended to be an exhaustive discussion of all the references relating to prenatal screening for Down syndrome (or other fetal anomalies detectable in the first part of pregnancy) but rather, an up to date summary of the key references, particularly of large well designed studies, that can inform the current debate in New Zealand about the way forward for aneuploidy screening in this country.

There are two recent trials which have produced remarkably similar results that enable us to describe “best practice” from at least a theoretical view. (SURUSS, Wald et al 2003; FASTER, Malone et al 2005). Smaller studies plus the research which forms the basis of this report provide information on issues of implementation and a pragmatic approach to the development and operation of a screening programme.

The laboratory science and ultrasound techniques are not discussed except in terms of test performance within a screening process. The analysis of serum markers is now largely performed using patented commercially available kits and the main issues are therefore quality control rather than the basic science of the testing. Similarly, the ultrasound markers of aneuploidy are reviewed on the basis of their performance as screening tools. There are a number of approaches to quality assurance in ultrasound, but the actual processes have not been subjected to trials of efficacy.

2.2 Screening for Down Syndrome (Trisomy 21)

2.2.1 Rationale

The majority of cases of Down syndrome are due to non dysjunction leading to 3 copies of chromosome 21 in the cells of the embryo-fetus. The event of non dysjunction is related principally to maternal age. With the increasing median maternal age for women having pregnancies in New Zealand it could be predicted that the background rate of Down syndrome births would increase. In addition, anecdotally at least, with smaller families and delay in childbearing, the expectation of women and partners regarding reproductive outcomes has also increased. In 4% of cases of Down syndrome a translocation has occurred and in 1% of cases are due to mosaicism. In this situation the extra chromosome usually is of maternal origin but may in around 9% of the time be paternal (Mutton 1996). The evidence of the effect of paternal age on the incidence of Down syndrome remains controversial.

2.2.2 Maternal age

It had been recognised since the 1930's that maternal age was related to the risk of having a pregnancy with Down syndrome (Penrose 1933) and in 1968 the first antenatal diagnosis was made. Maternal age became the first screen used with a typical cut off for “screen positive” being maternal age of 35 years or more at the expected date of delivery. Many studies (SURUSS, FASTER) and clinical experience have shown that this is a very crude form of

screening which leads to a very high false positive rate, that is, many invasive test procedures for cases detected.

It is now accepted that maternal age should not be used alone as the screening test to determine whether or not invasive testing is offered. Karyotyping by culturing fetal cells in the amniotic fluid obtained by amniocentesis has been available and funded for around 30 years. When the median age for women in pregnancy was 8-10 years younger than today, the workload involved in offering women over 35 years of age amniocentesis was much smaller than today and the pregnancy losses associated with the procedure seemed to be an acceptable cost for the diagnostic information that was achieved. That situation is no longer tenable for many reasons most of which are obvious. (Stone 2005).

2.2.3 Serum Biomarkers

In the 1980's the association of low maternal serum a-fetoprotein (AFP) levels with Down syndrome was noted (Merkatz 1984). The inclusion of two other biomarkers and maternal age were grouped into a single antenatal screening test which became known as the "triple test" with a detection rate of around 60% in the second trimester (Wald 1988). The detection rate increased to 70% where ultrasound was used to determine the gestational age. Later addition of a fourth analyte (inhibin-A) was shown to increase the detection rate to 76% for a 5% false positive rate and this test was termed the "quadruple test". (Wald 1996). *It was around this time that funding for second trimester maternal serum testing was withdrawn in New Zealand.*

This testing also served to screen for neural tube defects at a time when the ultrasound detection was less effective and widespread use of folate supplementation had not been instigated. Ultrasound has now superseded serum markers as the principal method of screening for and diagnosing neural tube defects. Whilst AFP may be raised in pregnancies that have adverse outcomes, currently there is no good evidence that the positive predictive value of AFP in determining likelihood of such an event is useful clinically.

First trimester serum markers, in particular pregnancy associated placental protein A (PAPP-A) and free B HCG (for review, Hyett 1999) were investigated in the 1990's. These 2 markers, between 8-14 weeks used with maternal age had a detection rate of 62% of T21 pregnancies for a 5% false positive rate.

Following the first description of nuchal fluid collection in fetuses with Down syndrome by vaginal ultrasound (Szabo 1990) this technique was also shown to be useful for detecting Trisomy 21 fetuses. (See below). In 1997 using the same methodology that was used to develop the triple test, maternal age, NT scan, PAPP-A and free B HCG were put together into a single first trimester screening test which has come to be called the "combined test" (Wald NJ 1997). This first trimester combined testing achieved an 85% detection rate for a 5% false positive rate. (For subsequent full analysis of this test refer to SURUSS and FASTER).

Finally in 1999, first and second trimester tests, "combined" and "quadruple" were integrated into a single test (noting that the results of the individual components were not revealed separately) and this new "integrated" test procedure achieved a high detection rate (85%) for a very low false positive rate (0.9%). (Wald 1999). Subsequently two trials have been undertaken one in Britain (SURUSS) the other in the United States of America

(FASTER) to prospectively assess the performance of these screening procedures in large populations.

2.2.4 Ultrasound

This discussion will be confined largely to first trimester or very early second trimester ultrasound scanning specifically for screening for aneuploidy.

One of the aspects of ultrasound, which differs from the serum markers, is that it may be both a screening tool and diagnostic for the obvious reasons that certain fetal abnormalities will be apparent when the scan is performed. However there are no absolutely diagnostic signs of Down syndrome on ultrasound, hence it is a screening modality for this abnormality and can be assessed as such in terms of its test performance. Snijders (Snijders 2002) reviewed Nuchal Translucency (NT) scanning and its role in screening for Down syndrome. The Fetal Medicine Foundation from which the work was published reported varying detection rates for this test. Apart from the technical issues related to operator performance many of the early reports on NT screening did not have complete case ascertainment.

A significant number of Down syndrome fetuses will spontaneously miscarry between the first and second trimester testing times and unless this is factored into the efficacy of the NT screening, the detection rate for a given false positive rate will be over estimated. Around 43% of Down's syndrome fetuses alive at 10 weeks will miscarry subsequently and 23% of those alive at 16 weeks will also miscarry (Morris 1999). A full review of NT based screening (Welch & Malone 2005) has discussed the many factors that influence the efficacy of NT scan alone as a screening test for Down syndrome. It is also unclear from the original studies whether fetuses with obvious severe abnormalities visible on the NT scan (but not necessarily part of the "ultrasound phenotype" of T21) were included in the figures of test performance. The FASTER trial took care to report fetuses with and without septated cystic hygroma. Fetuses which are clearly abnormal on the NT scan have in effect had a partial diagnosis made and it would be generally accepted in any screening process that when such an event occurs, the index case be offered appropriate diagnostic testing and be removed from the screening programme.

The NT measurement increases with gestational age. It is therefore most appropriate to express the measurements as multiples of the median for the given gestational age. It has also been found that the difference between unaffected and T21 fetuses with respect to NT is greater at 10-11 than 12-13 weeks and therefore the NT test will perform better at the earlier gestation periods because there is less "overlap" between normal and T21 fetuses in earlier pregnancy (Schuchter 1998). This has implications for the design and implementation of screening which includes ultrasound as a component.

Other ultrasound markers, in particular the fetal nasal bone remain to be fully evaluated and at present comparative data with other tests are not available to permit comment as to the effect of inclusion of this marker in a screening algorithm.

What will be important to demonstrate will be the independence of this newly proposed marker compared with other ultrasound markers. It has been shown that the ultrasound markers are independent from the serum analytes and therefore can be included in a screening process to improve the overall test performance

2.3 Consumer issues and approaches to prenatal testing

There is a considerable literature on differing approaches to offering testing to women. It can be difficult to assess this because of the bias of the authors, which is based on their views of the “best” approach and also by the study design. For example, it is not always clear whether in giving detection rates, both the false positive rate and the odds of actually being affected after being found screen positive are given. Also the loss rate from the invasive procedure in the study centre is not always given. A further consideration, which is very relevant to a regionally or nationally organised programme, is the cost per case detected and how this may alter with different screening regimens.

Before undergoing the screening process, parents require information. Testing for Down syndrome is not considered “routine” for the very reason that this is screening and the current forms of diagnostic testing have significant implications for the woman. The dissemination of the information and how this is achieved is a fundamental part of screening. A survey of what women over 37 years of age had expected prenatal testing for birth defects to tell them about their pregnancy showed that nearly 40% did not mention Down syndrome in the responses. This was more likely to be the case from a non-English speaking background and if diagnostic testing was not done (Jacques 2004).

An “opinion piece” discussing concerns about unwanted information concluded that “couples simply need an opportunity to inform professionals of their wishes prior to undergoing testing, and they should have this opportunity routinely”(Boyle 2003), though exactly how this is assessed is difficult to determine.

The UK National Screening Committee has also been concerned about a model of good practice and the issues of both introduction of screening and false positive rates. In addition to patient anxiety, false positive rates lead to the potential loss of unaffected fetuses as well as increasing the costs per affected case detected (<http://www.nelh.nhs.uk/screening/documentNSC/Downsyndrome/modelclarification/7.9.04> J A Muir Gray Programme Director). In that document figures for differing screening model outcomes are clearly listed by numbers of Down syndrome cases diagnosed, numbers of unaffected cases lost and numbers of Down syndrome cases diagnosed per unaffected fetuses lost. Importantly this was based on assuming an 80% acceptance rate for amniocentesis or chorionic villus sampling and a 0.9% procedure related loss rate. When all these factors are considered it was clear that NT scanning alone could not be recommended and serum integrated or integrated screening as defined in SURUSS were the preferred procedures.

Apart from the issues of the performance of various screening procedures, women’s choice is also important in any screening process. Whilst clinicians have tended to focus on detection rates, consideration of the screen positive rates is also very important for a number of reasons. For a given detection rate, minimising the screen positive rate will reduce the total number of diagnostic tests performed, the cost of the programme will be reduced, the number of normal pregnancies that miscarry as a result of diagnostic testing will be reduced and importantly there will be less maternal anxiety associated with the fewer false screen positive test results (Marteau 1992). There is other evidence that women prefer a low screen positive rate when assessing screening test performance (Mulvey 2003).

The development of first trimester screening approaches which have included combining NT scanning with availability of either immediate diagnostic or serum screening (with bench-top

analysers) has led to an increasing debate about offering women one stop clinics for the assessment of risk (OSCAR) or revealing the results at the first trimester screen even when a second sampling in the second trimester may have been planned. A questionnaire survey in a unit, which offers a first trimester approach, reported a majority of respondents preferring first trimester test with rapid reporting of results (Spencer 2004). A small study from the Netherlands where prenatal screening is not offered to women under 36 years of age also found a preference for screening in the first trimester (de Graaf 2002), but larger studies have found contradictory results.

In a study designed to assess the relative values respondents place on test attributes it was found that it was the health care professionals that placed a higher value on the earlier tests. Women would wait almost double the amount of additional time as health care professionals for a 1% decrease in risk of miscarriage. This has not been assessed in New Zealand but there have been similar findings from Australian research (Mulvey 2003).

As discussed previously, it is well recognised that around 20% of Trisomy 21 fetuses miscarry between the first and second trimesters and should all of these have been detected, then women would have had to go through a medical procedure when nature would have effected a spontaneous miscarriage. There is considerable evidence that at least in the first half of pregnancy, the decision to terminate a wanted but affected pregnancy is very traumatic regardless of gestation and it may be the doctors rather than the women who perceive advantages of ending the affected pregnancy as soon as possible. (Baram 1997, Moulder 1999, Bishop 2004).

Two other patient issues which affect the efficacy of any screening programme are the uptake of diagnostic testing in the screen positive group and the timing (gestational age) of presentation. The data being used in the UK by the National Screening Committee assumes an 80% uptake of invasive testing. In a country with ad hoc screening and no national programme, Ireland, a study showed only 1/3rd of screen positive women took up the offer of invasive testing though this may reflect other issues such as attitudes on termination of pregnancy. Even in the study by Wapner, generally supporting first trimester testing, (Wapner 2003) only half of the women who elected to terminate the pregnancy did so before 16 weeks.

A recent study of antenatal care attendance of Pacific Island women in South Auckland found that over 26% attended late, that is, after 15 weeks gestation (Low 2005) and other reports have found higher figures (Ekeroma 1999).

There will be a need to research issues of women's preferences within a New Zealand context, taking into account cultural and ethnic differences as screening becomes established in a coordinated way.

2.4 Comparing Screening tests

SURUSS and FASTER

It was not until the publication of the SURUSS report that it was possible to compare different screening tests for Down syndrome because of different study design. SURUSS and FASTER were large prospective studies.

Assessment of Antenatal Screening for Down Syndrome

In SURUSS, conducted between 1995 and 2002, women were seen in the first and second trimesters without intervention in the first. The details of the methodology are available elsewhere (Wald 2003). A summary, with the practical clinical implications, was published in 2004 (Wald 2004). The conclusion of SURUSS was that the fully “integrated “ test i.e. first trimester NT scan and serum plus second trimester serum with the results only revealed then was the test of choice based on safety, efficacy and cost. This is consistent with the findings of Wald in 1999 based on combining findings of different studies. Where quality ultrasound was not available, serum integrated, revealing the blood results in the second trimester was the next best choice. Other approaches are compromises due to either late testing or individual request.

Table 1: A summary of efficacy and safety of the recommended tests in SURUSS based on an 85% detection rate for Down syndrome is as follows:

Test	Efficacy		Safety
	False +ve %	OAPR*	Procedure related loss/100,000
Integrated	0.9	1:5	6
Serum Integrated (if no NT scan)	3.9 (2.7)**	1:20 (1:14)**	28 (19)**
Quad test (2 nd trimester only)	6.2	1:32	45
Combined (1 st trimester test)	4.3	1:22	35

* Odds of a positive result - odds of being affecting i.e. having Down syndrome if screen positive

** If PAPP-A measured at 10 weeks (as it has more discrimination at this time)

For an 85% detection rate the NT scan had a false positive rate of 15%

OAPR 1:94
losses 108

In addition, overall at one examination, NT was not obtained in 9% with the best time for success being 12 weeks when 93% of scans successfully obtained the NT measurement. There was also evidence presented to show that sonographer specific multiples of the median (MOMs) would improve the detection rate for a given false positive rate. This has implications for the assessment of sonographers and the case load that each should have. This will require the development of a process to do this should NT screening be adopted in a national programme. At detection rates of 85% or higher, integrated was the most efficient in terms of cost having the lowest cost per case of Down syndrome detected.

The FASTER trial (Malone 2005) 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, but women whose fetuses had a septated cystic hygroma (a clear abnormality) were offered diagnostic testing and these cases were analysed separately and did not contribute to the calculation of risk in the main part of the

study. Despite the differences in the two studies in terms of design and location the results have proven to be remarkably similar.

In FASTER, the failure to either obtain a NT image or have it scored by the investigators as satisfactory was 7% again similar to SURUSS. Both studies confirm that the earlier first trimester testing performs better than later i.e. 12-13 weeks and further research in a future programme would need to determine whether the differences are important in practice. As in SURUSS the fully integrated test performed best. Serum integrated i.e. in first and second trimesters, performed similarly to first trimester combined screening i.e. NT plus first trimester serum.

FASTER specifically addressed the issues of revealing results after first trimester testing and 2 approaches were investigated. The first, termed independent sequential screening revealed results and if screen positive, diagnostic testing could be considered. Screen negative women return for second trimester quadruple testing and these results are not combined with the previous first trimester screen. Adopting this would lead to an 11% false positive rate for an 85% detection rate and with reduced accuracy could not be supported.

The second approach termed stepwise sequential screening combines the results of the first and second trimester screening in the screen negative and if each component has a false positive rate set at 2.5% yields a high detection rate, similar to fully integrated screening but with a slightly higher overall false positive rate. In FASTER the use of the nasal bone was assessed and found to have little value. Other reports principally from Britain have had a differing view but the study design has been different from that of FASTER and the nasal bone has been suggested to be a component of further fetal assessment where risk is between 1:100 and 1:1000 (Avgidou 2005) but the evidence for the efficacy of this and other ultrasound markers individually has not been presented.

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 discussed above, in addition to the problem of having screening available to women who present at non optimal times with respect to test performance.

The issue of revealing results of first trimester testing in some way has been addressed very carefully in a paper, which modelled "contingent screening" (Wright 2004) and compared this to non-disclosure sequential screening as in SURUSS. Contingent screening is a strategy whereby second trimester marker determination is contingent upon the first trimester results. False positive rates are marginally higher than with the fully integrated approach but may allow 75% of women to complete screening at the end of the first trimester. This method requires more experience in practice but may offer an effective and safe way of screening a number of women who request early disclosure of results. Two risk cut-offs defined three types of first trimester result, namely, positive and refer for diagnostic testing, negative and screening complete and intermediate needing second trimester markers. This is different from simply using the combined first trimester test, which as SURUSS has shown has a significantly poorer performance than fully integrated screening.

Another issue is the late presenter who has missed the opportunity for first trimester screening. In a study of women over 35 years of age and with negative second trimester

serum screening, a detailed ultrasound (“genetic sonogram”) increased detection rates modestly (DeVore 2003).

Nicolaides et al. (2005) studied the effect of another model of screening, a risk-orientated two stage first trimester screening for Down syndrome. Maternal age, fetal nuchal translucency scan and maternal serum were used to identify risk in the first trimester. Women who were high risk (1 in 100 or more, 12% of population) were offered chorionic villus sampling. Those with an intermediate risk (1 in 101 to 1 in 1000, 16% of population) were offered further scanning for nasal bone, abnormal ductal flow or tricuspid regurgitation, if negative no further screening was required and if positive chorionic villus sampling was offered. Low risk women (1 in 1001 or less, 82% of population) received no further screening. This process was found to potentially identify more than 90% of affected fetuses for a false-positive rate of 2-3%. However, it is not entirely clear what the demographics of the population studied was and how this could be applied more widely.

2.4.1 Discussion of SURUSS and FASTER

It is apparent in reviewing much of the literature on antenatal screening and that of commentaries about SURUSS and FASTER, that epidemiologists and statisticians have varying views on differing methodologies of study design and data analysis and interpretation (Cuckle 2003; Krantz 2004). For example Cuckle (2003) presented a brief discussion of SURUSS in the Down’s Screening News. In that discussion, it is suggested that the effect of the serum markers to increasing detection rate by NT is exaggerated. The reanalysed SURUSS (Wald 2004) has agreed very closely with FASTER and addresses some of the issues identified by Cuckle (2003).

A review of screening options by Simpson (2005) highlights the issue of women failing to complete the protocol if a single risk calculation is given after second trimester testing. In the SURUSS trial one third of participants did not return in the second trimester, some of whom may have an increased risk score from the first trimester that they were not aware of. There is debate about the effects on the sensitivity of ‘integrated’ screening of disclosing first trimester results (Cuckle & Arbusova 2004).

The solution for New Zealand would seem to be prospective monitoring of the performance of a screening programme. The key issues, rather than the statistical debates, would really be the outcomes which require a high degree of consumer uptake of the screening and a low and defined intervention and iatrogenic pregnancy loss rate. These variables which are strictly independent of the actual screening tests, but fundamental to a programme will ultimately determine the efficacy and therefore the success or otherwise of screening strategies.

2.5 Establishment of a Prenatal Screening service

There are a number of components of a screening programme and in a generic way these have been detailed in documents published by the National Screening Unit of the Ministry of Health in New Zealand (MOH 2005). Prenatal screening has some unique features which are in part responsible for the discussion on issues such as timing of disclosure of results and acceptance rates.

Different countries have taken differing approaches to prenatal screening from no screening, age based (The Netherlands), state based (California), ad hoc availability of tests (Australia), to moving to a nationwide programme with a staged introduction in terms of programme outcomes, screening test availability and performance measures (<http://www.nelh.nhs.uk/screening/dssp/policy.htm>). In a country the size of New Zealand with an average of 55,000 births per annum the size of a national programme would be feasible and manageable and be an efficient use of laboratory services.

The results of the survey which is the subject of this report and other information from a centre with a similarly structured screening to that in New Zealand at present highlight the need for professional education about prenatal screening (Tyzack 2003) with less than 10% of clinicians being able to provide detection or screen positive rates. In the United Kingdom, a staged approach to the introduction of the screening programme has included professional education including developing counselling services for women and the production of patient information. Training of sonographers may be less of a problem in New Zealand where first trimester scanning has attracted some government funding for some time now, but the quality assurance issues and development of individual sonographer MOMs for NT discussed above would need to be considered.

This literature review has focussed on the assessment of screening for Down syndrome. Both lay and some professional groups argue in favour of screening for more than just Trisomy 21. The performance of the test procedures cannot be directly extrapolated to other aneuploidies or other fetal anomalies. To do so would reduce the efficacy of the programme and has the potential to mislead. On the other hand, it is suggested that the maximum information should be obtained from any test be it ultrasound or serum markers. The common assumption that detection of an abnormality per se is worthwhile can be challenged (for full review of ultrasound see Wald and Leck, 2000, Chapter 18). It will be a matter of debate whether New Zealand chooses introduce prenatal screening for Down syndrome or prenatal screening for fetal abnormalities

2.6 Conclusions

Current evidence allows some conclusions to be made to guide screening practice in New Zealand.

- NT screening alone is not supported by the evidence and excluding a few centres is not used in any large screening programmes.
- Addition of nasal bone to “intermediate risk” women requires validation in new studies.
- Sonographer based MOMs will improve efficacy of screening which includes NT.
- Evidence on timing of disclosure of results in the currently operational or proposed screening programmes shows a divergence of opinion dependent upon study type, location and views of consumer versus clinician.
- Initiation of a screening programme for the prenatal detection of Down syndrome requires a staged approach with outcomes being reached over an agreed timeline.
- Efficacy of programmes depends on many factors including time of presentation, type of screening protocol, and uptake of diagnostic tests.
- The efficacy of Down syndrome screening testing cannot be directly extrapolated to other fetal abnormalities.

3. Working group

3.1 Working group members

In August 2005, at the initiation of this project, a working group was convened by the authors to assist in providing information on the current situation in New Zealand and advise on if / how a screening programme for Down syndrome should be implemented. Assistance was also provided in developing the questionnaires and piloting them amongst professional colleagues. Ten people attended representing 9 organisations or groups. Four further members were unable to attend. The working group met for 3 hours and further discussion was undertaken via e-mail and phone. (Appendix 1)

The opportunity for a second meeting occurred in October 2005 following the screening symposium convened by the National Screening Unit of the Ministry of Health. Other relevant practitioner groups were also invited to this discussion group where Professor Nick Wald also attended. (Appendix 1)

3.2 Working group meeting

During the first meeting short presentations were given on the topics listed below.

- Purpose of meeting (Professor Peter Stone)
- Prenatal screening for Down syndrome (Diana Austin)
 - What is a screening programme
 - Current situation in New Zealand
 - Overseas literature and experience
 - Developing a quality programme
- Laboratory tests (Dr Di Webster)

Following the presentations each member of the group presented their views and comments from both the organisation they represented and individual experience. There was general discussion around each issue raised. Comments from this first meeting were collated by Diana Austin and sent to members for verification and circulation to the member organisations. A summary of the key issues raised is presented in the following section.

3.3 Issues raised

3.3.1 Definition of topic

The group members highlighted that currently the testing used to identify Down syndrome also detects other chromosomal abnormalities. However the efficacy of the tests in the literature relate to Down syndrome only. The key aspect is that women need to be accurately informed about what information can or will be given to them following screening tests. The National Committee meeting of the New Zealand Down Syndrome Association, following the project working group meeting, discussed the terminology regarding screening and the following statement was sent to us.

The NZDSA has requested that Professor Peter Stone include in his report that the NZDSA would prefer that in the future screening is no longer referred to as Screening for Down syndrome but rather Screening for Chromosomal Abnormalities.

3.3.2 Current situation

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.

Access to screening

Access to screening is not always available with many women not booking early enough with a LMC for screening to be offered. If another health practitioner has been seen by the women prior to booking screening may not have been discussed. Lack of time and funding for practitioners to provide all the health information and screening options within the booking visit was also raised.

Screening process

The consumer representatives present indicated that there was a trust in health professionals to offer the best options and to give detailed information about these. Currently the information resources to make an informed choice are not available. The current perception by some women is a feeling of "being put in a moving vehicle that has lost control and end up with a TOP". It was commented that on occasion the NZ Down Syndrome Association picks up families who have terminated a baby then later see families with a child with Down syndrome and decide they could have coped with the situation. They then regret what they have done.

NT scanning

It was commented that sometimes a scan of adequate quality is not available. Some practitioners stated they send women for a repeat scan if screens positive to check results. The method of scanning has been changed arbitrarily by Fetal Medicine Foundation. A woman who was screened last year would have a higher risk if screened again this year. This has a huge implication in cost and time for diagnostic tests.

Lack of consumer knowledge

Both practitioner and consumer representatives believed many women are not aware of the benefits of early booking with a LMC and require further education. The NT scan is seen by some consumers as providing the first picture of baby rather than providing health benefits.

3.3.3 Working group recommendations for a screening programme

Members of the working group suggested recommendations to improve the quality of screening in New Zealand. These are outlined below.

Appropriate screening options

The group was in agreement that the best screening programme needs to be available to reduce the trauma caused to families by having false positives. A National screening programme was considered by some as the best way to achieve this. Due to the varying

gestation at which women present for antenatal care a screening process would need to allow for multiple entry into the programme.

Screening process

A high standard of pre-screening counselling was expected by all to be an essential part of any screening programme. The screening process should be simple, supportive and relevant to the varying needs, particularly cultural, of consumers. Allowing time to understand and reflect on the information and options available to women is required. Following testing the current system of receiving a positive screen needs to be improved i.e. counselling, timeliness of diagnostic tests and getting results. Again time and support to consider the results was considered important.

Resources

Practitioner members of the group identified a current lack of appropriate resources (financial, personnel and time) to provide screening needs and that this lack needs to be addressed to ensure appropriate screening can occur. Increased first trimester funding in particular was considered of importance. Some attendees felt that the issue of resources should be addressed in relation to other screening in pregnancy.

Improved information for women

It was indicated that there is a need for education of consumers of the benefits of seeing a health practitioner early in pregnancy for general health reasons as well as options relating to Down syndrome screening. 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.

All participants agreed on the need to improve the current situation and provide practitioners with a national guideline on screening for Down syndrome.

4. Consumer viewpoint

The viewpoint of consumers was intended to be a major focus of this project. As ethics approval was required to undertake focus groups there was no available time or resources to implement this aspect of the project. Three consumer groups were represented on the working groups and a summary of their comments has been presented in the section pertaining to the working group. Further consumer input is required in developing screening for Down syndrome in New Zealand that meets the needs of women and their families.

5. Cultural implications of screening for Maori

The Treaty of Waitangi principles of partnership, protection and participation need to be applied to the practice of screening for Down syndrome to ensure equity and that the specific needs of Maori are met. A representative from Maori health at Auckland District Health Board (ADHB) working group and was able to provide input into the discussion. ADHB have a Tikanga Recommended Best Practice Policy which was believed to be relevant nationally (ADHB 2003).

6. Assessment of Prenatal Screening for Down Syndrome

Survey of the current situation in New Zealand

6.1 Aim

The aim of the survey was to gain information about the current practice of screening for Down syndrome offered to women, general knowledge about Down syndrome screening, further education needs and opinions about how a screening programme should occur in New Zealand. A survey of District Health Boards also aimed to find out if there was any directive by the organisations on what antenatal screening should take place and what they would consider workable within their service.

6.2 Method

6.2.1 Data collection tool

Three questionnaires were developed to survey the current practice of antenatal screening for Down syndrome in New Zealand (Appendix 2). The working group which represented the range of health practitioners and some consumer organisations was convened to provide the researchers with information and advice about their views on the current situation. The group that each party represented had been contacted by the researchers to explain the purpose of the project. The nominees were not chosen by the researchers. A second opportunity for all the members of the working group to meet occurred following the seminar in on screening convened by the National Screening Unit of the Ministry of Health in October 2005. Issues raised at that meeting were incorporated into the project's findings.

Survey one examined the current practice of screening among midwives, obstetricians and General Practitioners. A separate survey was sent to the managers and/ or clinical leaders of maternity services within the 21 District Health Boards. A third survey was developed for the providers of NT scans, namely public and private ultrasound / radiology practices.

The surveys were developed in collaboration with a working group (Appendix 1). Each member provided feedback on behalf of the organisation they represented. Feedback was incorporated and revised copies sent back to the working party members to provide further comment and to be piloted amongst professional colleagues where appropriate. Eight pilot

questionnaires were returned (Obstetrician 3, GP 1, DHB 2, NT scan provider 2) further changes were made. The three final questionnaires were posted in September and October 2005 and distributed to each practitioner group as outlined below.

6.2.2 Sample

A total of 1454 practitioners or practitioner groups were sent a questionnaire with the option of returning it in a Freepost envelope, fax or e-mail. The survey was anonymous unless the respondent chose to include their name. We were therefore unable to follow up those that did not respond apart from the DHB survey. A description of the sample groups is outlined below.

General Practitioners

Due to the larger number of General Practitioners and financial restraints of the project a sample of 691 General Practitioners from a total sample of 3457 on the register (obtained through the September 2005 Atlantis Mailing list received from the NSU) were posted a survey. Every fifth GP was systematically selected from the list which was ordered numerically by Medical Council Registration Number to ensure a good and even range of experience as the MCNZ registration number is correlated with GP registration date.

Obstetricians

The surveys to obstetricians were initially sent electronically by the Executive Officer of the New Zealand committee of the Royal Australian and New Zealand College of Obstetrics and Gynaecology (RANZCOG). A hard copy of the survey was then sent to 216 Fellows, Members and Trainees with their newsletter with a reminder in the newsletter to complete it.

Midwives

The New Zealand College of Midwives and Midwifery Council were unable to distribute the survey to its members. However the New Zealand College of Midwives provided a response statement to the questionnaire, which is included in Appendix 4. The names and addresses of 326 midwives were therefore obtained from the white and yellow pages of the telephone directory, and web pages via the internet. Thirty of these were returned to sender with address unknown.

South Auckland Maternity Care Limited (SAMCL) a private maternity service provider who was represented on the working group, provided support for the project by sending the questionnaire electronically to 63 of its midwifery members.

District Health Boards

Surveys were sent to each of the 21 District Health Boards in New Zealand addressed to the midwifery manager of antenatal clinic / community services. Where a response was not received on the first mailing a second questionnaire was addressed to the clinical leader of Obstetrics and Gynaecology of each DHB. The repeat questionnaire was sent to the 9 DHBs that did not respond initially.

NT providers (Radiologists / Sonologists / Sonographers)

Questionnaires were sent to 94 radiology facilities accessed via the web. The survey was also sent electronically to radiologists via the New Zealand Branch of the Royal Australian and New Zealand College of Radiologists.

6.2.3 Data analysis

A database was established using Access and Excel programmes (Microsoft). Each survey was assigned a number. Frequency and percentages for each question were calculated and compared between practitioner type, DHB area and years of experience. Themes were developed from qualitative data and each survey coded accordingly. Some of the qualitative responses have been copied verbatim and are included in Appendix 3. The number after the comment refers to the assigned number given to the response.

6.3 Results

The principle findings relating to each subject area in the questionnaire are presented in this section. The questionnaire (Appendix 2) and details of all responses (Appendix 3) are presented as an appendix. Each subject heading within this section corresponds to the equivalent numbering in Appendix 3, i.e. **6.3.2 Screening offered to women** corresponds to **A3.2 Screening offered to women** in the appendix.

6.3.1 Survey responses

A total of 1454 surveys were posted, and 499 completed surveys were returned. One was removed as it applied to practice in the UK. Thirty nine were returned to sender as the address was unknown. The overall response rate was 35.2 % (498/1415). The response rate, for individual practitioner groups, is shown in Table 2. The highest response rate was from midwives. At the beginning of the survey for midwives, obstetricians and GPs, practitioners were given the option of completing only questions 1 – 6 if they felt they were unable to complete all questions. Three midwives and twenty six GPs took this option. They have been removed from the analysis of questions 7 onwards.

The responses to the first questionnaire consisted of 34.6% midwives, 16.9% obstetricians and trainees, and 48.5% GPs. A breakdown of each practitioner group is provided in Appendix 3.

To assess whether the sample obtained was representative of all the areas within New Zealand the responses were compared to the proportion of women who had given birth within each DHB area according to the latest Report on Maternity 2002 (NHIS 2004). Overall the responses were similar to the birth number with a lower rate for the Capital & Coast, Taranaki and Tairāwhiti regions and a higher rate in the Auckland and Bay of Plenty regions (Table 3). This suggests that the survey does provide a representation of practitioners and practices around all regions in New Zealand.

A summary of the survey results was offered to health practitioners and 153 requested a copy.

Table 2: Summary of surveys received

	Postal surveys sent	Returned to sender	Completed surveys	Response Rate
Midwives - posted	326	30	128	43.2
Midwives –e-mail via SAMCL	63	0	21	33.3
Obstetricians*	199	0	62	31.2
Trainee registrar	60	0	11	18.3
GPs	691	7	209	30.6
Radiology practices **	94	2	47	
DHB	21	0	18	85.7
Not stated			2	
Total	1454	39	498	35.2

- * Not all Fellows are in active practice or active obstetric practice. For the remainder of the survey analysis where response rate is not required trainees and specialists are grouped together.
- ** Two questionnaires from radiology practices indicated that they included a response from more than one practitioner (10 and 20 practitioners). However each questionnaire has only been counted as one response to prevent skewing of the results.

Assessment of Antenatal Screening for Down Syndrome

Table 3: DHB area responses received from

DHB	Midwives	Obstetrician	GP	NT Provider	Total	%	2002 % women per DHB
Northland	3	2	8	2	15	3.1	3.4
Waitemata	27	3	31	0	61	12.7	12.1
Auckland	16	14	31	1	62 +2 ns	13.3	11.6
Counties Manukau	11	4	19	5	39	8.1	13.7
Auckland - all or not defined	8	9	1	14	32	6.7	-
Total Auckland	62	30	82	20	196	40.8	37.4
Waikato	17	2	10	1	30	6.3	8.2
Bay of Plenty	15	1	17	5	38	7.9	4.6
Taranaki	0	2	4	0	6	1.3	2.5
Lakes	6	1	5	3	15	3.1	2.8
Tairāwhiti	1	0	1	0	2	0.4	1.4
Wanganui	3	0	2	0	5	1.0	1.6
Mid Central	3	4	5	3	15	3.1	3.5
Hawkes Bay	8	3	8	0	19	4.0	3.9
Capital and Coast	3	2	7	2	14	2.9	6.7
Hutt	3	1	2	0	6	1.3	3.3
Wairarapa	3	0	1	0	4	0.8	0.9
Nelson marlborough	1	0	9	3	13	2.7	2.6
West Coast	1	0	1	0	2	0.4	0.6
Canterbury	10	9	23	1	43	9.0	9.6
South Canterbury	1	2	4	0	7	1.5	1.0
Otago	3	7	6	3	19	4.0	3.5
Southland	3	4	4	0	11	2.3	2.3
Not stated	3	3	10	4	20	4.2	0.1
	149	73	209	47	480	100.0	100.0

- One response was also received from an obstetrician working in the UK but has been removed from the analysis.

Most respondents in all practitioner groups had greater than 5 years experience with only 17.1% having 5 years or less experience. Over 50% received their practitioner training in New Zealand (Table 17, 18 in Appendix 3).

6.3.2 Screening offered to women

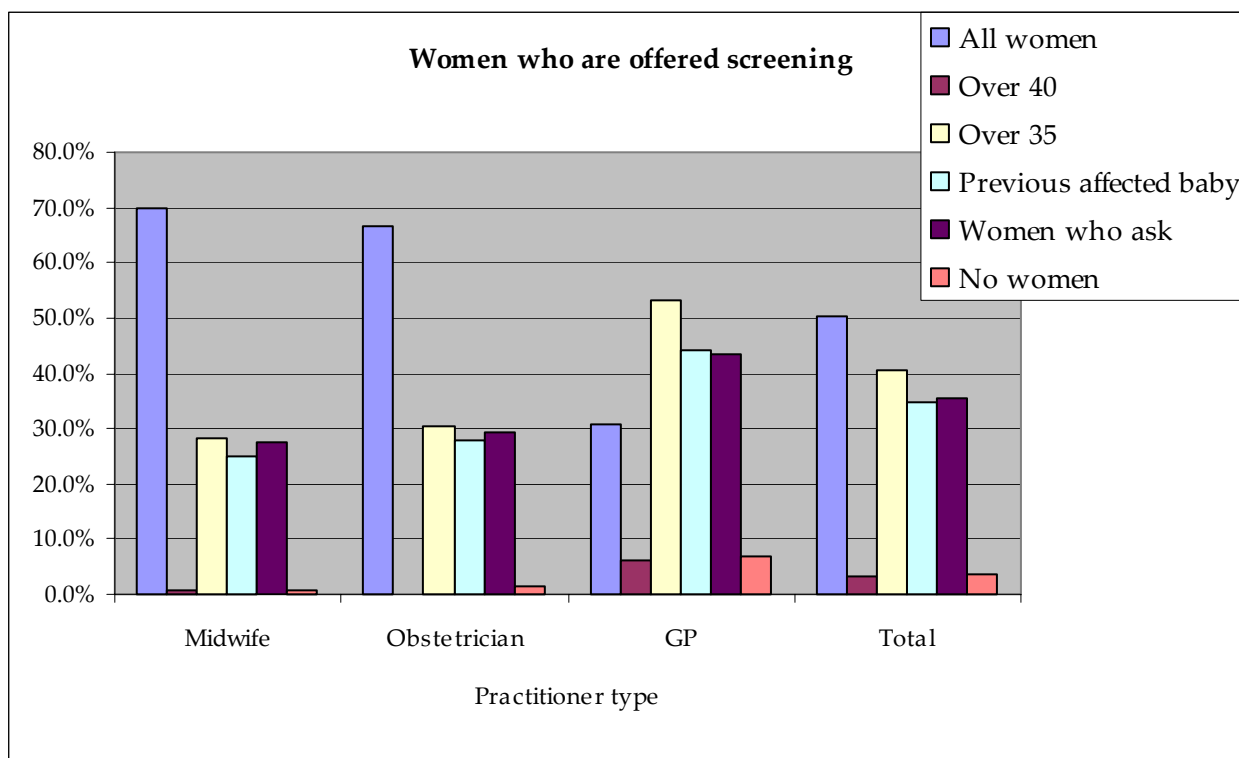
The results of questions about health practitioner's current screening practices and their own assessment of their knowledge of screening are presented in this section. In Table 4 and Figure 1, clear differences are seen between practitioner groups with midwives and specialists being more likely to offer screening all women. GPs are more likely to not to offer screening at all or only to women who ask for it.

From the analysis of the 15 responses where screening was not offered one midwife stated that screening was not believed in. The 14 GPs stated they were not involved in obstetric care. (Data not included).

Table 4: Women who are offered screening by practitioners

Offer screening to:	Midwife n=149		Obstetrician n=73		GP n=209		Total N=431	
	n	%	n	%	n	%	n	%
All women	104	69.8	48	65.8	64	30.6	216	50.1
Women over 40	1	0.7	0	0.0	13	6.2	14	3.2
Women over 35	43	28.9	22	30.1	111	53.1	175	40.6
Women with previous affected baby	38	25.5	20	27.4	92	44.0	149	34.6
Women who ask	42	28.2	21	28.8	91	43.5	153	35.5
No women	1	0.7	0	0.0	14	6.7	15	3.5
Not complete/ NA	0	0.0	2	2.7	0	0.0	2	0.5

Figure 1: Graph of women who are offered screening by different practitioner groups



Practitioners were asked to give their reason for offering screening to the group of women they selected. These reasons were grouped into common themes. (Some responses contained more than one theme). In table 5 and 6 these reasons are shown by practitioner type and whether offered screening to all or a selected group of women. A selection of practitioner comments is presented in Appendix 3 (section A3.2). Overall, the main reason for offering screening to all women was for women’s right to informed choice.

Table 5: Reasons for offering screening to all women

	Midwives n=104		Obstetricia n=48		GP n=64		Total N=216	
	n	%	n	%	n	%	n	%
All women at risk	21	20.2	17	35.4	22	34.4	60	27.8
Women’s right to informed choice	57	54.8	13	27.1	13	20.3	73	33.8
Awareness/request	9	8.7	0	0.0	5	7.8	14	6.5
Parental concern	2	1.9	1	2.1	3	4.7	6	2.8

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Tests available	6	5.8	7	14.6	6	9.4	19	8.8
Current practitioner knowledge	2	1.9	0	0.0	1	1.6	3	1.4
Other	3	2.9	6	12.5	1	1.6	10	4.6
NA / refer to other	0	0.0	2	4.2	1	1.6	3	1.4
Not stated	10	9.6	5	10.4	15	23.4	30	13.9

Other includes reasons indicated in comments in appendix.

Table 6: Reasons for offering screening to selected groups of women

	Midwives n=44		Obstetricia n=23		GP n=131		Total N=198	
	n	%	n	%	n	%	n	%
Women have increased risk factors	22	50.0	7	30.4	70	53.4	99	50.0
Women's right to informed choice	1	2.3	2	8.7	5	3.8	8	4.0
Awareness/request	8	18.2	2	8.7	9	6.9	19	9.6
Parental concern	2	4.5	0	0.0	6	4.6	8	4.0
Tests available	1	2.3	1	4.3	1	0.8	3	1.5
Current practitioner knowledge	3	6.8	1	4.3	11	8.4	15	7.6
Other	6	13.6	5	21.7	11	8.4	22	11.1
NA / refer to other	6	13.6	4	17.4	28	21.4	38	19.2
Not stated	9	20.5	5	21.7	17	13.0	31	15.7

Other includes reasons indicated in comments in appendix.

6.3.3 Knowledge levels of practitioners

The survey sought to investigate the practitioners own assessment of their knowledge of Down syndrome and screening for the condition.

a) Knowledge of Down syndrome

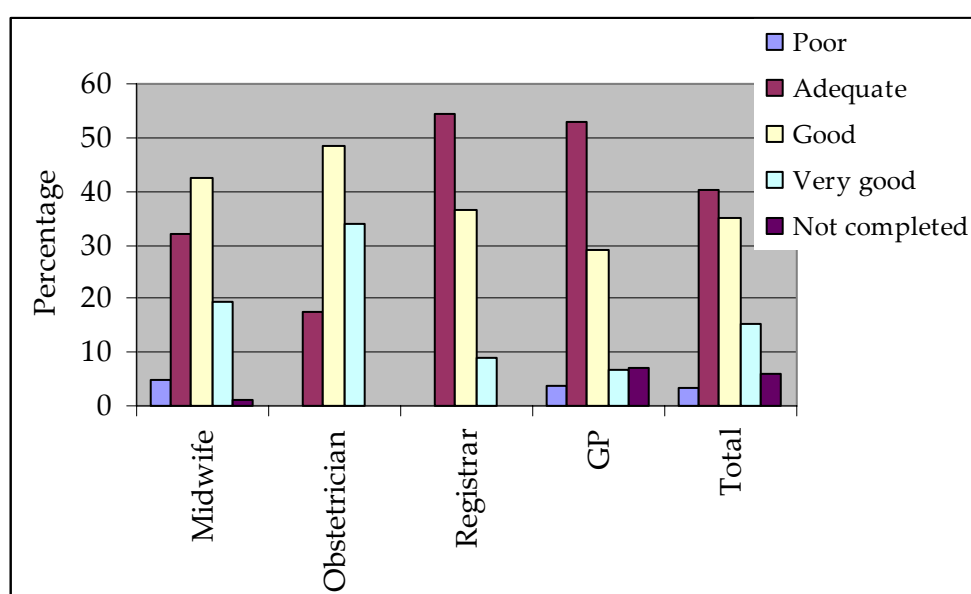
Most responses reported adequate or good knowledge (Table 7).

Table 7: Practitioners self assessment of knowledge of Down syndrome

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	Poor		Adequate		Good		Very Good		Not completed	
	n	%	n	%	n	%	n	%	n	%
Midwife	7	4.7	48	32.2	63	42.3	29	19.5	2	1.3
Obstetrician	0	0.0	11	17.7	30	48.4	21	33.9	0	0.0
Registrars	0	0.0	6	54.5	4	36.4	1	9.1	0	0.0
GP	8	3.8	111	53.1	61	29.2	14	6.7	15	7.2
Total	15	3.5	176	40.8	158	36.7	65	15.1	17	3.9

Figure 2: Graph of practitioners self assessment of knowledge of Down syndrome



b) Knowledge of Screening for Down syndrome

Similarly practitioners generally felt their knowledge of screening was at least adequate with GPs being least likely to report good or very good levels of knowledge (Table 8).

Table 8: Practitioners self assessment of knowledge of Down syndrome screening

	Poor		Adequate		Good		Very Good		Not completed	
	n	%	n	%	n	%	n	%	n	%
Midwife	2	1.3	41	27.5	76	51.0	27	18.1	3	2.0
Obstetrician	0	0.0	9	14.6	26	41.9	27	43.5	0	0.0
Registrars	1	9.1	5	45.4	4	36.4	1	9.1	0	0.0

Assessment of Antenatal Screening for Down Syndrome

GP	26	12.4	102	48.8	53	25.4	11	5.3	17	8.1
Total	29	6.7	157	36.5	159	36.9	66	15.3	20	4.6

6.3.4 Purpose of screening

Practitioners were asked to write what they believed to be the purpose of screening. Common themes were identified and each response coded according to these themes as shown in Table 9.

Despite the results regarding knowledge assessment, only 16.3% overall stated that the purpose of screening for Down syndrome was to assess the woman's risk of having a baby with Down syndrome thus allowing them to make an informed choice/give them options about going on to have a diagnostic test. Interestingly, while most practitioners mentioned both outcomes of screening, some practitioners focused only on the screen-positive woman (i.e. option to have amnio, option to terminate) and others focused only on the screen-negative woman (i.e. reassure and allay anxiety). Some practitioners stated that the purpose of screening was to "detect" or "diagnose" Down syndrome, or alternatively to "reassure parents," which underscores the need to further educate practitioners and their patients of the limitations of screening tests, i.e. false positives and false negatives. Some practitioners stated that screening allowed for earlier detection of Down syndrome, enabling parents more choices to act on test results. Lastly, a few practitioners commented that the purpose of screening was to reduce the number of babies born with Down syndrome at a community or society level.

Table 9: Purpose of screening

	Midwives		Obstetrician		GP		Total	
	n	%	n	%	n	%	n	%
Identify risk / Offer option of amnio	28	18.8	20	27.4	22	10.5	70	16.3
Choices - Continue or TOP	80	53.7	29	39.7	94	45.0	203	47.1
Provide information	2	1.3	4	5.5	4	1.9	10	2.3
Identify DS	3	2.0	7	9.6	4	1.9	14	3.2
Reassure women	3	2.0	0	0	3	1.4	6	1.4
Prepare to care for Disable child	2	1.3	0	0	3	1.4	5	1.2
TOP only	13	8.8	5	6.8	41	19.7	59	13.7
Decrease DS in society	2	1.3	6	8.2	7	3.3	15	3.5
No purpose	0	0	0	0	1	0.5	1	0.2
Other	1	0.7	1	1.4	2	1.0	4	0.9
Not completed	15	10.1	1	1.4	28	13.4	44	10.2

Total	149	100.0	73	100.0	209	100.0	431	100.0
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6.3.5 Screening tests offered to women

Practitioners were given a list of screening tests available in New Zealand and were asked which they recommend to women. Many practitioners ticked more than one option (Table 11).

Amniocentesis was not given as an option because it is not a screening test however 30 practitioners added it to the list. NT scan and maternal age were most commonly offered. A selection of the many comments received is shown in Appendix 3 (A3.5). The comments reflect a very large amount of inaccurate information and lack of ready availability of 2nd trimester serum screening.

Table 10: Screening tests offered to women

Screening tests recommended:	Midwife	Obstetrician	GP	Total N=402	Total (%)
Maternal age	78	43	90	211	52.5
NT scan	132	65	140	337	83.8
Second trimester blood	24	15	22	61	15.2
NT scan & second trimester blood test	16	13	33	62	15.4
Other - amnio	15	2	13	30	7.5
- 2 nd trimester US markers / anatomy scan	1	2	1	4	1.0
- Nasal scan	2	0	1	3	0.7
-Refer to LMC / obstetrician	0	0	6	6	1.5
Not complete/ NA	4	2	10	16	4.0

It was found that no District Health Boards had guidelines for antenatal screening for Down syndrome. Whilst there were information sheets on prenatal tests there were no policies or guidelines on an approach to screening.

Nuchal Translucency (NT) scanning

NT providers were asked specific questions relating to the practice of NT scanning such as waiting times and cost of scan and number of NT scans per month. These data is presented in Appendix 3 (Tables 36-42, A3.16). In addition to the government subsidy of \$80 per scan, NT cost women between \$0 - \$95 with most in the \$20 - \$40 range.

6.3.6 Practitioner knowledge about gestation at which screening tests undertaken

Practitioners were asked to state at what gestation the NT scan and second trimester blood test is undertaken. This was an open question with no options given. There were 75% (NT scan) and 58.5% (second trimester blood test) of practitioners who gave answers within a generally agreed acceptable range for the correct time for testing. Conversely 25% (NT scan) and 41.5% (second trimester blood test) did not respond correctly (Table 20 & 21 in Appendix 3).

6.3.7 Risk threshold used by practitioners

Overall 38% of practitioners answered within the accepted range of 1:250 – 1:300. In assessing risk range for screening 64.4% of obstetricians indicated an answer within a generally agreed acceptable range, whereas only 37% of midwives and 21.3% of GPs did so. Of note is that 66.6% of NT scan providers indicated an answer within the accepted range, i.e. one third of those providing NT screening were unclear of the risk threshold used in the computer algorithm.

6.3.8 Topics covered in discussion with women about screening

The survey provided a range of options for clinicians to choose about screening topics to be discussed with women. Nearly three quarters of clinicians overall discussed Down syndrome but one quarter did not despite this being the condition being screened for. (Table 23, Appendix 3). We were unable to assess the quality of the information giving from this type of survey. (Practitioner comments are in section A3.8 of Appendix 3).

6.3.9 Consent for screening

The vast majority of clinicians obtained oral consent for screening, with a few stating it was written when the second trimester blood test was requested (Table 24, Appendix 3).

6.3.10 Giving of risk estimates

Practitioners were asked to tick from a selection of options how they would give risk estimates to women, both positive and negative.

Positive

When a woman screened 'positive' 77.4% of practitioners indicated they would give the results face to face. However 18 (4.5%) practitioners said the positive result would have already been given by the NT scan provider (Table 25, Appendix 3).

Negative

About one half of practitioners would give a screen 'negative' result face to face with a quarter receiving them via the telephone. Again close to 5% would receive the result from the NT scan providers (Table 26, Appendix 3).

NT providers

Thirty two percent of NT scan providers stated they always gave women their risk estimate at the time of scan, with 45% sometimes giving a result. Only 19% stated they would never give a result to a woman (Table 27, Appendix 3).

6.3.11 Recommendations if screens 'positive'

This section deals with issues around giving results to the screen 'positive' women. Practitioners were asked to state what they would recommend to women if they screened 'positive'. Common themes were identified and responses coded (Table 11). Some responses contained more than one theme. Most practitioners (69.6%) suggested a diagnostic test or referral to an obstetrician for further discussion. Sixteen practitioners stated they had never had a women screen positive so had therefore not been in this situation. Eight practitioners said that any recommendations to be made would have already been discussed as part of informed consent prior to screening.

There were 17 comments stating that they (the practitioner) did not recommend anything to women rather they gave options for the women to make the decision. However it is possible women may not appreciate this difference.

e.g. "don't recommend anything. I can suggest an amnio" (373)

Twenty three (5.3%) responses indicated either a lack of understanding of the difference between a screening test and a diagnostic test or a misunderstanding of the question being asked (7 midwives, 3 obstetricians & 13 GPs.). These practitioners suggested a termination or support for continuing the pregnancy if they screened positive.

Table 11: Recommendations given to women if screened 'positive'

	Midwives n=146		Obstetrician n=73		GP n=183		Total N=402	
	n	%	n	%	n	%	n	%
Another screening test (2 nd tri bloods)	5	3.4	3	4.1	3	1.6	11	2.7
Diagnostic testing	63	43.2	52	71.2	54	29.5	169	42.0
Refer - obstetrician	67	45.9	na	0.0	43	23.5	110	27.6
- geneticist	12	8.2	2	2.7	2	1.1	16	4.0
- paediatrician	5	3.4	1	1.4	1	0.5	7	1.7
- Counselling/advice	1	0.7	1	1.4	8	4.4	10	2.5

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Support networks	16	11.0	1	1.4	9	4.9	26	6.5
Discussion with family	7	4.8	2	2.7	7	3.8	16	4.0
TOP or continue pregnancy	7	4.8	3	4.1	13	7.1	23	5.7
Discuss options (not specified)	22	15.1	6	8.2	25	13.7	53	13.2
Situation has not occurred	6	4.1	0	0.0	10	5.5	16	4.0
Will have been discussed prior to screening	4	2.7	3	4.1	1	0.5	8	2.0
Not completed	13	8.9	5	6.8	34	18.6	52	12.9

Around one fifth of clinicians would base definite advice (assumption of fetus affected) on the outcome of the screening.

As the NT screen is most commonly used test we assessed what NT scan providers recommended. Just over half would suggest discussion with LMC but over 40% of NT scan providers stated that they would recommend invasive testing based on their assessment of risk after the scan (Table 12).

Table 12: Recommendations given by NT scan provider to women who have a high risk result

Recommendations	Number N=47	%
Do not discuss	6	12.8
Discuss with LMC	25	53.2
Counselling	1	2.1
Referral to obstetrician	9	19.1
Speak to radiologist	1	2.1
Further prenatal testing	1	2.1
Recommend an amnio	19	40.4
Discuss with partner	1	2.1
Not completed	2	4.3

a) Information given about fetal loss rate with amniocentesis

Overall 48% of practitioners informed women that there was a 1% fetal loss rate with amniocentesis (Table 13). However between 13 and 40 % were informed that the rate was

Assessment of Antenatal Screening for Down Syndrome

0.5% especially among the obstetricians (39.7%). Practitioners who gave a reason for the lower rate said it was due to the experience of the clinicians in their area (Auckland, Tauranga, Nelson, and Christchurch).

Table 13: Fetal loss rate for amniocentesis given to women.

	Midwives		Obstetrician		GP		Total	
	n	%	n	%	n	%	n	%
0.5%	20	13.7	29	39.7	30	16.4	79	19.7
0.5 – 1%	11	7.5	6	8.2	3	1.6	20	5.0
1%	74	50.7	32	43.8	88	48.1	194	48.3
1-2%	13	8.9	0	0.0	9	4.9	22	5.5
2%	14	9.6	1	1.4	16	8.7	31	7.7
1:100 – 1:400	0	0.0	1	1.4	0	0.0	1	0.2
1:300	1	0.7	0	0.0	0	0.0	1	0.2
1:400	0	0.0	1	1.4	0	0.0	1	0.2
*Other	3	2.1	0	0.0	10	5.5	13	3.2
Not completed	10	6.8	3	4.1	27	14.8	40	10.0
Total	146	100.0	73	100.0	183	100.0	402	100.0

*Other included those who did not give a specific rate but rather said it depended on factors such as practitioners, gestation, placenta site or that they referred the women to another practitioner for this information. (Practitioner comments in Appendix 3).

6.3.12 Responsibility for Screening

Most respondents stated this was the responsibility of the LMC even when all groups gave comments indicating that at the timing of screening they would not be LMC (Table 28 & comments in Appendix 3).

6.3.13 Is it possible for a woman who screens 'negative' to have a baby with Down syndrome?

Ninety two percent of practitioners (368/402) indicated that it was possible to have a baby with Down syndrome if the woman screened 'negative'. However some practitioners' answers indicated a lack of understanding between screening and a diagnostic test. Several stated it was possible to have a baby with Down syndrome if screen 'negative' as there can be sampling errors with an amnio. The few who said no – added it was not possible with an amnio and one response stated it was "dependent on test used" (334).

6.3.14 Education for Down syndrome screening

Training health practitioners is an important part of screening. Having made an assessment of levels of current knowledge the survey sought to investigate how practitioners would view further education.

Some practitioners who stated they did not want further education answered the question relating to the most useful way to receive that education so all these responses were also included in the analysis (Table 29). Overall written guidelines (46.1%) or an information booklet (44.1%) were the most wanted method of further education with workshops (30.3%) and online education (27.8%) being the least wanted. There were differences within practitioner groups with workshops being one of the preferred methods for midwives but least preferred for the obstetrician and GP groups (Table 30, Appendix 3).

6.3.15 Future screening programme

During the workshop, all attendees stated the need for some form of screening programme. The survey gave options for respondents to choose about who should be offering screening and a question on payment.

a) Aspects of a good programme

Pre-test information and counselling were considered the most important factors (Table 32, Appendix 3). Important comments are included in Appendix 3. Health practitioners have commented on deficiencies they see in others' practices.

b) Who should be offered screening

Only 59% thought screening should be offered free to all women (Table 31, Appendix 3).

c) Funding of programme

We were interested in views on funding for participating in part of a programme or having diagnostic testing if screen negative. Overall the practitioners were fairly evenly divided in their responses (Table 33 & 34, Appendix 3).

d) Screening tests

The survey sought to determine views on "best practice" and favoured approach. We provided a simple description of tests and test performances. The two most favoured options, with similar numbers in both were either combined (first trimester NT scan and serum screening) or the integrated test (first trimester NT scan and first and second trimester serum testing). When given options in this way, very few chose maternal age alone, in contradiction to responses earlier in the survey. In Appendix 3 we have also listed a number of comments relating to why the choices for a particular option were made by practitioner groups.

6.4 Discussion

Within the limits of this type of survey the response rate is as expected. The geographic representation of responses does reflect birth numbers in the associated regions. The authors of the report are confident that this survey is representative and that the survey has confirmed the issues raised by the working group as being the key topics to be addressed in carrying out Down syndrome screening in New Zealand. The individual members of the working group had consulted or been briefed by the organisations they represented. As these members are individuals, the authors do not imply that every statement made represents the collective opinion of the parent organisations.

The one major topic that the survey did identify, not discussed by the working group, was the issue 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% of practitioners.

Only 17% of the respondents had been in practice less than 5 years, consistent with other workforce surveys suggesting an older age working group. Over 30% of the respondents were non New Zealand trained.

Training and service provision will both be key issues in the implementation of a screening programme in New Zealand. This is clearly apparent in the GP group. Responses in the appendix suggest that GPs are less involved and quote 'exclusion' from the antenatal screening process though this is done prior to confirmation of an LMC. It appears as though GP attitudes to antenatal screening for Down syndrome are different from the other health practitioners, with more GPs selectively offering screening or not offering screening at all. Should there be a change in the involvement of GPs in antenatal screening, it would seem reasonable to suggest that there be an education process developed.

Only 50% of respondents offer screening to all women. In New Zealand at present screening is offered on an ad hoc, inconsistent, or selective basis. It was found that 3.5% of health practitioners did not offer screening to any woman. This situation potentially deprives women of choices or information. The authors of this report would support the National Screening Unit bringing this data to the attention of the appropriate professional groups.

NT scans and maternal age were the screening tests most commonly offered. There are significant impediments to the use of second trimester maternal serum screening relating to poor knowledge and cost. Specific comments are in the appendix.

None of the District Health Boards had guidelines or policies on the actual offering of screening antenatally. There are some protocols for the performance of invasive testing but this is not relevant to screening.

The assessment of timing for various tests and information about these showed what appears to be confusion on the part of many clinicians. There was better knowledge demonstrated about the invasive tests. Whilst many practitioners replied that their

knowledge was adequate or good, the answers to the specific questions would not necessarily confirm this.

The majority of clinicians felt that the LMC should be responsible for initiating screening. As screening currently by age or NT scan is usually done prior to the confirmation of an LMC, their response is difficult to interpret. Either respondents believe the LMC should assume clinical responsibility earlier or that (as figures showed) the GP should, -though a number of individual comments highlighted the small role GPs have in this currently.

Both the working group and the survey respondents have indicated the need for and support of change in the way antenatal screening for Down syndrome is performed in this country. Those clinicians who did wish to have further education tended to favour being given guidelines rather than educative processes involving adult learning although there was some support for workshops, information booklets and online education. Consistent with evidence presented in the literature, when given a range of screening options with test performance, respondents chose very differently from the tests they currently do.

Combined 1st trimester (NT and bloods) and integrated screening were chosen equally as the preferred options for screening. This re affirms the importance of the way test information is presented to both the clinicians and of course women. There is also the potential for bias, which to some degree is inevitable, for example presentation of absolute risk versus likelihood ratio will elicit differing responses from women to the same clinical situation.

There was very little support for blood tests alone. Of the 285 replies selecting one best option, only 8% chose NT scan alone, this included NT scan providers. This suggests that ultrasound services recognise that NT scan alone is not necessarily best practice.

The lack of support for serum testing probably reflects the change in emphasis in the funding of antenatal procedures and the lack of co-ordinated screening in New Zealand. In most other countries it is the provision of sonography, not blood testing, that limits the widespread implementation of screening programmes. The survey of NT scan providers has shown that whilst some practices do large numbers of NT scans using Registered programmes with quality assurance processes, there are others doing few scans with little external audit. This type of situation does not arise with centralised laboratory services processing serum. There are special processes needed to ensure quality in ultrasound. The use of individual MOMs has been discussed in the literature review.

The involvement of the NT scan provider in counselling, risk estimation and recommendations for action with a 'high probability' result is an area of concern. There are a range of approaches taken and views expressed, detailed in Appendix 3. There is no co-ordinated or agreed method and advice from an NT scan provider (not clinician responsible) to proceed to invasive testing has the potential to place practitioners and the service at risk. Over 40% of NT scan providers would recommend an amnio for a 'high risk' result.

The respondents' views on funding screening were somewhat inconsistent or even parsimonious. Noting that many felt that even serum negative women should have invasive testing funded, only 59% of respondents felt that screening should be freely available to all women. This is also despite the fact that NT scans are available to most women funded at least in part.

The authors suggest that with education about the screening process, practitioners would be able to formulate a clearer view on an effective approach to screening and may then have a greater clarity about questions such as partial involvement in a screening programme.

7. The costs of a screening programme

It was not the brief of this project to assess the cost effectiveness of screening or screening options. Before the implementation of a screening programme this will be required. Variations in the screening programme would need to be costed to determine what screening process would be supported financially by the government.

Costing a programme can be somewhat limited or artificial depending upon the outcome measures that are chosen to be included in the exercise. For example, no recent economic modelling has included the costs to an economy of individuals born with disabilities that result in ongoing health care expenditure. It becomes a philosophical as well as a moral or ethical debate as to whether it should be done.

Generally programmes are costed on a basis of outcomes which include cost of cases of fetal abnormality detected, the total cost of screening strategy employed with comments or estimates about iatrogenic losses. 'Cost-benefit' analysis is a difficult approach to take in assessing screening strategies as it is difficult to assign an appropriate value to the results (information) obtained from screening or diagnostic test. These issues have been recently reviewed by Ritchie et al (2005).

It is probably more appropriate for budgeting purposes to cost a programme once the various options for screening strategies within the programme have been reviewed. SURUSS (Wald 2004) provides comparative information on different strategies, and given that in the United Kingdom ultrasound is not paid as a 'fee for service' is probably a reasonably accurate representation of costs as some of the SURUSS strategies did not involve ultrasound.

A working party charged with implementation of a screening programme for the antenatal detection of Down syndrome or other chromosomal abnormalities would usefully include a health economist.

8. Conclusions

This project, completed by the authors on behalf of the National Screening Unit of the Ministry of Health through a contract administered by Auckland Uniservices Limited, has endeavoured to describe the current state of antenatal screening for Down syndrome in New Zealand. Information was also sought on the possible shape of screening in the future.

Screening programmes are population health programmes and, as such, they are planned, funded, delivered and monitored from a population health perspective. Thus, screening programmes have resources committed to the development,

implementation, monitoring and evaluation of all aspects of the programme, from the identification of the population at risk, the diagnosis of the disease or its precursor in certain individuals, to the treatment of those individuals (Gray 2001 in MOH 2003).

The working group supported this statement and strongly believed that antenatal screening for Down syndrome satisfied the criteria for a screening programme.

The eight quality requirements of a screening programme have been defined as:

- Standard setting and monitoring
- Performance management
- Training and certification
- Opportunities for shared learning
- Effective information systems
- Appropriate resources
- Research and development
- Information for individuals and communities (MOH 2005).

All of these were deemed relevant and appropriate to antenatal screening for Down syndrome

Standard setting, monitoring and performance management can be established with reference to the literature and programmes already in existence (e.g. United Kingdom).

Training and certification has been identified in this project as a key issue and would require specific action within a New Zealand context.

Shared learning would occur both through training and by ongoing quality improvement activities.

Effective information systems would need to be established as part of the programme.

The clear message from the working group gives the National Screening Unit the necessary information to defend **resourcing** a programme adequately.

Research and development. There is sufficient expertise within New Zealand to provide ongoing enhancements of any programme as new knowledge and results become available.

Information for individuals and communities is essential. It would also include the reporting of the monitoring of the programme.

The working party, convened at the start of the project provided important information in addition to the survey. Due to time and financial constraints it was not possible to have more than one full meeting of the working party but we believe the results can represent the current situation.

It is important that the National Screening Unit be aware that some clear messages came from the working group. Most importantly the current situation of ad hoc screening, increasing NT scans and amnio rates in New Zealand is not leading to efficient detection of Down syndrome fetuses. There was a unanimous view that there was urgent need to change practices. This is consistent with the responses in the survey where, given the information a minority (including NT scan providers) selected NT scan alone as the best screening option.

Whilst recognising cultural issues the working party felt that all women had a right to access screening. Of more concern was the issue of late booking, a particular problem in Maori and Pacific Island groups. Availability of second trimester bloods remains the only option for later booking and the group concluded that this option should be available.

The most difficult area for the working group was the issue of screening for just Down syndrome or other aneuploidies and other abnormalities. The midwifery and consumer groups felt it was important to state very clearly exactly what it was that was being screened for. The New Zealand Down Syndrome Association was concerned that a screening programme which detected other problems apart from Trisomy 21 not be called a Down syndrome programme.

The NT scan provider groups have a professional tension between screening only for a particular condition and obtaining 'maximum' information from the screening. This affects the performance of any screening test procedure.

The survey has highlighted issues of knowledge about screening, inconsistent approaches to screening, the communication of results, women's choice and funding. There is much detail in the individual comments (Appendix 3) which shows a desire to see change.

It has been found impossible to accurately determine how many invasive diagnostic procedures are done in New Zealand after a screening episode. This is because the laboratories do not code the indications for karyotyping in a way which can accurately distinguish a test done purely for screening from one done where there was another indication such as a clear structural abnormality. Similarly separating maternal age, NT scan and NT scan plus age has not been possible. What is well reported elsewhere is the increasing number of invasive procedures and karyotyping and the overall aneuploidy rates, both per test and at birth. This is a further reason to ensure much better data collection, reporting and reduction in invasive procedures.

The authors have detailed recommendations based on the working party responses, the survey and literature review and concludes that change is needed, there are immediate ways forward and the implementation of a coordinated national screening programme is achievable, given the interest and goodwill on the part of all contributors to this project.

9. Acknowledgements

We would like to acknowledge the invaluable assistance of the working group in providing guidance on the key issues relating to current screening for Down syndrome in New Zealand and increasing the awareness of screening within their organisations.

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Appendix 1

Members of the working group

Working group members and meetings attended

1. Thursday, 11 August 2005

Auckland

1:00 – 4:00pm

Present

Zandra Vaccarino	<i>National co-ordinator; The New Zealand Down Syndrome Association</i>
Liz Berry	<i>New Zealand Council of Women</i>
Sharon James	<i>Auckland Regional Co-ordinator, Parents Centre</i>
Janie Lawakeli	<i>Women's health - Maori ADHB</i>
Jenny Woodley	<i>Committee member, NZ College of Midwives</i>
Emma Farmer	<i>Committee member, NZ College of Midwives</i>
Anne Whyte	<i>Director and Midwife, South Auckland Maternity Care Limited</i>
Margaret Shanks	<i>RNZ College of GPs</i>
Gill Gibson	<i>Royal Australian and NZ College of Obstetricians and Gynaecologists</i>
Dianne Webster	<i>ADHB Laboratory specialist</i>
Peter Stone	<i>Project Leader</i>
Diana Austin	<i>Research Assistant, Uniservices</i>

Apologies

Tish Taihia	<i>Pacific Island peoples representative</i>
Aumea Herman	<i>Pacific Island peoples representative</i>
Janet Chen	<i>Asian representative</i>
Alistair Woodward	<i>Biostatistician/epidemiologist, Population Health</i>

2. Wednesday 5 October 2005

Auckland

Presentation by Prof Nick Wald **1:30pm – 3:00pm**

Discussion and working group **3:30pm – 5:00pm**

Over 100 health practitioners attended the presentation by Prof Nick Wald. The following also attended a discussion group meeting with Prof Nick Wald.

Zandra Vaccarino	<i>National co-ordinator; The New Zealand Down Syndrome Association</i>
Alastair Haslam	<i>Director Obstetrics and Gynaecology, Waikato DHB, NZ vice president</i>
RANZCOG	
Ian Page	<i>Clinical Director of Obstetrics and Gynaecology, Northland DHB</i>
Anne Whyte	<i>Director & Midwife, SAMCL</i>
Tony Mansfield	<i>General Manager, SAMCL</i>
Mary Stuart	<i>Team Leader, National Testing Centre</i>
Dianne Webster	<i>Clinical Director, National Testing Centre</i>
Lesley Irvine	<i>Midwife</i>
Elizabeth Berry	<i>NZ Council of Women</i>
Dwayne Crombie	<i>CEO, Waitemata District Health Board</i>
Alistair Roberts	<i>Ultrasound subspecialist</i>
Kathy Bendikson	<i>National Screening Unit</i>

Angie Partridge
Sylvia Rosevear
Sue Fitzgerald
Nick Wald
Peter Stone
Diana Austin

National Screening Unit
Private Obstetrician
Midwife Manager Waitemata DHB, NZ College of Midwives
Director Wolfson Institute
Head of Department of O & G, University of Auckland
Research Assistant, University of Auckland

Appendix 2

Survey questionnaires

- **Health Practitioners**
- **District Health Boards**
- **NT scan providers**

1. Health practitioners (Midwife, Obstetrician and GP)



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Antenatal Screening for Down syndrome and other chromosomal conditions

Survey of screening practices in New Zealand

The National Screening unit is aiming to provide guidance to the Ministry of Health on screening for chromosomal abnormalities in pregnancy. We have been asked to find out what is currently occurring in practice.

We would appreciate you taking the time to complete the questions and provide any further feedback you feel may be useful in developing a complete screening programme in New Zealand. If you feel unable to complete the survey it would be invaluable if you could complete questions 1-6 and return to us in the freepost envelope.



If you have other comments to make please use the space provided at the end of the survey.

Please tick the box that best answers the question.

1. Type of practitioner

- | | | |
|--|--|---|
| <input type="checkbox"/> Independent midwife | <input type="checkbox"/> Obstetrician (private only) | <input type="checkbox"/> General Practitioner |
| <input type="checkbox"/> Hospital midwife (core staff) | <input type="checkbox"/> Obstetrician (public only) | <input type="checkbox"/> Registrar |
| <input type="checkbox"/> Hospital midwife (case loading) | <input type="checkbox"/> Obstetrician (private and public) | |

2. ...Number of years in practice

3. Did you train in New Zealand

- Yes No

4. Geographic location of practice Suburb Town/City

5. Do you discuss Down syndrome screening with (you may tick more than one)

- | | |
|--|--|
| <input type="checkbox"/> all women | <input type="checkbox"/> women over 35 |
| <input type="checkbox"/> women over 40 | <input type="checkbox"/> women with previous affected baby |
| <input type="checkbox"/> women who ask for screening | <input type="checkbox"/> no women |

6. What are the reasons for your answer in question 5?

.....

1. Health practitioners (Midwife, Obstetrician and GP)

7. How would you rate your knowledge of Down syndrome?

- Very good Good Adequate Poor

8. How would you rate your knowledge of **screening** for Down syndrome?

- Very good Good Adequate Poor

9. What do you believe is the purpose of screening for Down syndrome?

.....

10. Do women ask you about screening for Down syndrome

- Never Sometimes Often Always

11. The following are screening tests available in New Zealand. Which do you recommend to women who decide to have screening?

- Maternal age (women over a certain age are offered a diagnostic test) Nuchal translucency
- Second trimester blood test
- Nuchal translucency combined with 2nd trimester blood test (results not given separately)
- Other (describe)

12. At what gestation is nuchal translucency undertaken?

13. At what gestation is second trimester serum screening undertaken?

14. What is the risk threshold / level for offering diagnostic testing?

- 1:50 1:100 1:250 1:500 other don't know

15. If you do provide discussion on screening which of the following topics do you include in your discussion?

- Information about Down syndrome and other chromosomal conditions
- Types of screening options
- False positive and false negative results
- Need for a diagnostic test
- Support networks if positive diagnosis e.g. NZ Down syndrome association
- Termination of pregnancy
- Other

16. Is consent for screening oral or written?

- Oral Written

17. How are women given their risk estimate results if positive?

- Face to face Telephone Post Other (please state)

18. How are women given their risk estimate results if negative?

- Face to face Telephone Post Other (please state)

19. If a women screens 'positive' for Down syndrome what do you recommend to her?

20. In discussing amniocentesis what rate of fetal loss do you quote?

1. Health practitioners (Midwife, Obstetrician and GP)

0.5% 1% 1.5% 2% Other

21. Is it possible for a woman who screens 'negative' to have a baby with Down syndrome?

Yes No

22. Do you feel you are adequately trained to provide advice and counselling on screening for Down syndrome?

Yes No

23. Would you find further education on Down syndrome screening useful?

Yes No (please give reason)

24. If you answered yes above what would be the most useful way to receive this education?

Workshops Written guidelines

Information booklet Online education

25. Whose responsibility do you think it is to inform and offer screening **currently**?

LMC GP before booking District Health Board Ministry of Health

Other (Please state)

26. Whose responsibility do you think it **should** be to inform and offer screening?

LMC GP before booking District Health Board Ministry of Health

Trained midwife/counsellor Trained consumer Other (Please state)

27. What do you consider to be essential aspects of a good screening programme?

Pre-test written consumer information Informed consent

Pre-test counselling Laboratory quality audit process

Post-test counselling of all women Annual reporting

Post-test counselling of women who screen positive

Only offer best option of screening

Offer a range of options

Other

Comments

28. Do you think screening should be offered free to

All women Women over a certain age (state what age)

Women who ask No women

29. If a programme is funded, should women who want only parts of the programme, or invasive testing even if screen negative (that is low risk) have their tests paid for?

Yes No Don't Know Other

30. The following are possible **options for screening for Down syndrome**. What would you see as the best option for New Zealand? Please tick your preference in the **table** after reading the following explanations. The values are based on Serum Urine and Ultrasound Screening Study (SURUSS) report. [1]

1. Nuchal Translucency (NT) scan only

1. Health practitioners (Midwife, Obstetrician and GP)

2. Combined test

The first trimester (11 weeks) blood test and NT scan are combined to estimate the risk of Down syndrome.

1. Health practitioners (Midwife, Obstetrician and GP)

3. Second trimester blood test

A blood test is taken between 14 and 20 weeks gestation to test for substances produced by the placenta or fetus. They are alpha-fetoprotein (AFP), β -human chorionic gonadotrophin (β -hCG free and total), unconjugated oestriol and in the future inhibin-A.

4. Serum Integrated test

The first and second trimester blood tests are combined to give an integrated estimate of a woman's risk of Down syndrome. The integrated risk estimate is only available after the 2nd blood test.

5. Integrated test

The NT result is combined with both the first and second trimester blood tests to give an integrated estimate of a woman's risk of Down syndrome. The integrated risk estimate is only available after the 2nd blood test.

6. Nuchal translucency and second trimester blood test

Actual NT measurement and 2nd trimester blood levels are combined in a formula to give a risk assessment. Result is given after the 2nd trimester blood test.

7. Age alone

Women are offered a diagnostic test based on their age only.

Table 1: Performance of each screening test for Down syndrome

Screening test	Trimester	Detection rate	False positive rate	Odds of being affected given a positive result	Number of losses (of babies without DS) following amnio/ CVS <i>Per 100,000 screened</i>	Please tick preferred option/s
1. Nuchal translucency	<i>First trimester</i>	85%	15%	1:94	108	<input type="checkbox"/>
2. Combined		85%	4.3%	1:22	35	<input type="checkbox"/>
3. Second trimester blood	<i>Second trimester</i>	85%	6.2%	1:32	45	<input type="checkbox"/>
4. Serum Integrated test		85%	3.9%	1:20	28	<input type="checkbox"/>
5. Integrated test	<i>Both trimesters</i>	85%	0.9%	1:5	6	<input type="checkbox"/>
6. NT & 2 nd trimester blood		80%	5%	*	*	<input type="checkbox"/>
7. Age alone	0.8% of women referred for diagnostic testing based on age alone had a positive diagnosis of Down syndrome. [2]					<input type="checkbox"/>
Do not know						<input type="checkbox"/>

* There is little data on this option. Detection is increased by 25% (compare to each test on its own) [3]

1. Health practitioners (Midwife, Obstetrician and GP)

31. Please give the reason for your answer to Q. 30 on the previous page.

.....
.....

Additional comments

.....
.....
.....
.....

**FAX to 09 373 7900 or use the FREEPOST envelope provided.
(or e-mail to d.austin@auckland.ac.nz /post to address above)**

If you would like a summary of the results please include your name and address.

*Diana Austin
Research Assistant*

*Professor Peter Stone
Head of Department*



References:

1. Wald, N. J., et al., (2004). SURUSS in perspective, *BJOG : an International Journal of Obstetrics and Gynaecology*, 111, p. 521-531.
2. ADHB. (2004). Auckland: Unpublished.
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2. DHB Survey



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Antenatal Screening for Down syndrome and other chromosomal conditions

Survey of antenatal screening within District Health Boards in New Zealand

The National Screening unit is aiming to provide guidance to the Ministry of Health on a screening programme for chromosomal abnormalities in pregnancy. We have been asked to find out what is currently occurring in practice.

We would appreciate you taking the time to complete the questions and provide any further feedback you feel may be useful in developing a complete screening programme. Please ensure the questionnaire is completed in conjunction with those who are involved in screening in your facility.



If you have other comments to make please use the space provided at the end of the survey.

Please tick the box that best answers the question.

1. Name of DHB

.....

2. Does your facility provide primary, secondary or tertiary services?

Primary Secondary Tertiary

3. Number of births per year at your facility

.....

4. Number of births per year where a DHB employee is the Lead Maternity Carer?

.....

5. Does your facility have a guideline for antenatal screening for Down syndrome?

(If yes could you please enclose a copy of this guideline)

Yes No

2. DHB Survey

6. If no to Q.5 are the staff employed by your DHB directed in any way about how to undertake antenatal screening for Down syndrome?

Yes No

7. What tests are offered to women booked in your facility for antenatal screening for Down syndrome?

.....
.....

8. Does your facility ...

i, provide pre-test counselling Yes No

ii, provide post-test counselling Yes No

9. If you answered yes to the questions above please state who provides this counselling.

.....

10. How are women given their risk estimate results if positive?

Face to face Telephone Post Other (please state)

11. How are women given their risk estimate results if negative?

Face to face Telephone Post Other (please state)

.....

12. If a women screens 'positive' for Down syndrome what does your facility recommend to her?

.....
.....
.....

13. Whose responsibility do you think it is to inform women and offer screening **currently**?

LMC GP before booking District Health Board Ministry of Health
 Other (Please state).....

14. Whose responsibility do you think it **should** be to inform and offer screening?

LMC GP before booking District Health Board Ministry of Health
 Trained midwife/counsellor Trained consumer Other (Please state)

15. Are there any costs to the women for any of the tests mentioned above?

.....
.....

16. What do you consider to be essential aspects of a good screening programme?

Pre-test written consumer information Informed consent
 Pre-test counselling Laboratory quality audit process
 Post-test counselling of all women Annual reporting
 Post-test counselling of women who screen positive

2. DHB Survey

- Only offer best option of screening
 Offer a range of options
 Other

Comments

17. The following are possible options for screening. What would you see as the best option for New Zealand? Please tick your preference in the table below the explanations. The values are based on Serum Urine and Ultrasound Screening Study (SURUSS) report. [1]

1. NT scan only

2. Combined test

The first trimester (11 weeks) blood test and NT scan are combined to estimate the risk of Down syndrome.

3. Second trimester blood test

A blood test is taken between 14 and 20 weeks gestation to test for substances produced by the placenta or fetus. They are alpha-fetoprotein (AFP), β -human chorionic gonadotrophin (β -hCG free and total), unconjugated oestriol and in the future inhibin-A.

4. Serum Integrated test

The first and second trimester blood tests are combined to give an integrated estimate of a woman's risk of Down syndrome. The integrated risk estimate is only available after the 2nd blood test.

5. Integrated test

The NT result is combined with both the first and second trimester blood tests to give an integrated estimate of a woman's risk of Down syndrome. The integrated risk estimate is only available after the 2nd blood test.

6. Nuchal translucency and second trimester blood test

Actual NT measurement and 2nd trimester blood levels are combined in a formula to give a risk assessment. Result is given after the 2nd trimester blood test.

7. Age alone

Women are offered a diagnostic test based on their age only.

Table 1: Performance of each screening test for Down syndrome

Screening test	Trimester	Detection rate	False positive rate	Odds of being affected given a positive result	Number of losses (of babies without DS) following amnio/ CVS Per 100,000 screened	Please tick preferred option/s
1. Nuchal translucency	First trimester	85%	15%	1:94	108	<input type="checkbox"/>
2. Combined		85%	4.3%	1:22	35	<input type="checkbox"/>
3. Second trimester blood	Second trimester	85%	6.2%	1:32	45	<input type="checkbox"/>
4. Serum Integrated test		85%	3.9%	1:20	28	<input type="checkbox"/>
5. Integrated test	Both trimesters	85%	0.9%	1:5	6	<input type="checkbox"/>
6. NT & 2nd trimester blood		80%	5.0%	*	*	<input type="checkbox"/>
7. Age alone	0.8% of women referred for diagnostic testing based on age alone had a					<input type="checkbox"/>

2. DHB Survey

positive diagnosis of Down syndrome. [2]

Do not know

* There is little data on this option. Detection is increased by 25% (compare to each test on its own) [3]

18. What type of screening programme do you think would work best for women and would work within your facility?

.....
.....

19. If a programme is funded, should women who want only parts of the programme, or invasive testing even if screen negative (that is low risk) have their tests paid for?

Yes

No

Don't Know

Other

20. Please give the reason for your answer

.....
.....

Additional comments

.....
.....
.....
.....

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(or e-mail to d.austin@auckland.ac.nz /post to address above)**

*Diana Austin
Research Assistant*

*Professor Peter Stone
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3. NT Provider Survey



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Antenatal Screening for Down syndrome and other chromosomal conditions

Survey of Nuchal Translucency (NT) screening in New Zealand

The National Screening unit is aiming to provide guidance to the Ministry of Health on a screening programme for chromosomal abnormalities in pregnancy. We have been asked to find out what is currently occurring in practice.

We would appreciate you taking the time to complete the questions and provide any further feedback you feel may be useful in developing a complete screening programme.



If you have other comments to make please use the space provided at the end of the survey.

Please tick the box that best answers the question.

1. Do you work as a public or private provider?

Public Private Public and Private

2. What is your radiology / sonology / sonographer qualification?

.....

3. Number of years in practice

≤ 5 years > 5 years

4. Do you provide NT measurements for women?

No Yes (please state number per month)

5. Do you use a software programme to calculate the risk of Down syndrome?

Yes No

6. Have you undertaken training to use this package?

Yes No

7. Are you registered by the Fetal Maternal Foundation?

3. NT Provider Survey

Yes No

8. What threshold value or cut-off level is used for determining a high risk result?

.....

9. Do you have any ongoing quality assurance process for NT scanning?

Yes No

If yes please describe

10. Do you give a woman her level of risk at the time of scan?

Never Sometimes Always

11. What do you advise a woman if you are unable to measure the NT?

.....

12. Do you ...

i, provide pre-test counselling Yes No

ii, provide post-test counselling Yes No

iii, expect LMC to provide counselling Yes No

Comments

13. What cost is there to a woman who has a NT ultrasound at your facility?

.....

14. What is the usual length of time a woman will need wait to get an appointment for an early pregnancy scan?

.....

15. Explain how you get informed consent for the tests you discuss?

.....

.....

16. Is consent oral or written?

Oral Written

17. If a women screens 'positive' for Down syndrome what do you recommend to her?

.....

.....

.....

3. NT Provider Survey

18. Do you feel you are adequately trained to provide advice and counselling on screening for Down syndrome?

- Yes No

19. Would you find further education on Down syndrome screening useful?

- Yes No (please give reason)

20. If you answered yes above what would be the most useful way to receive this education?

- Workshops Written guidelines
 Information booklet Online education

21. Whose responsibility do you think it is to inform and offer screening currently?

- LMC GP before booking District Health Board Ministry of Health
 Other (Please state).....

22. Whose responsibility do you think it should be to inform and offer screening?

- LMC GP before booking District Health Board Ministry of Health
 Trained midwife/counsellor Trained consumer Other (Please state)

23. What do you consider to be essential aspects of a good screening programme? Please number in order of importance (1=most important).

- Pre-test written consumer information Informed consent
 Pre-test counselling Laboratory quality audit process
 Post-test counselling of all women Annual reporting
 Post-test counselling of women who screen positive
 Only offer best option of screening
 Offer a range of options
 Other

Comments

24. Do you think screening should be offered free to

- All women Women over a certain age (state what age)
 Women who ask No women

25. If a programme is funded, should women who want only parts of the programme, or invasive testing even if screen negative (that is low risk) have their tests paid for?

- Yes No Don't Know Other

3. NT Provider Survey

26. The following are possible options for screening for Down syndrome. What would you see as the best option for New Zealand? Please tick your preference in the table after reading the following explanations. The values are based on Serum Urine and Ultrasound Screening Study (SURUSS) report. [1]

1. Nuchal Translucency (NT) scan only

2. Combined test

The first trimester (11 weeks) blood test and NT scan are combined to estimate the risk of Down syndrome.

3. Second trimester blood test

A blood test is taken between 14 and 20 weeks gestation to test for substances produced by the placenta or fetus. They are alpha-fetoprotein (AFP), β -human chorionic gonadotrophin (β -hCG free and total), unconjugated oestriol and in the future inhibin-A.

4. Serum Integrated test

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5. Integrated test

The NT result is combined with both the first and second trimester blood tests to give an integrated estimate of a woman's risk of Down syndrome. The integrated risk estimate is only available after the 2nd blood test.

6. Nuchal translucency and second trimester blood test

Actual NT measurement and 2nd trimester blood levels are combined in a formula to give a risk assessment. Result is given after the 2nd trimester blood test.

7. Age alone

Women are offered a diagnostic test based on their age only.

Table 1: Performance of each screening test for Down syndrome

Screening test	Trimester	Detection rate	False positive rate	Odds of being affected given a positive result	Number of losses (of babies without DS) following amnio/ CVS Per 100,000 screened	Please tick preferred option/s
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2. Combined		85%	4.3%	1:22	35	<input type="checkbox"/>
3. Second trimester blood	Second trimester	85%	6.2%	1:32	45	<input type="checkbox"/>
4. Serum Integrated test		85%	3.9%	1:20	28	<input type="checkbox"/>
5. Integrated test	Both trimesters	85%	0.9%	1:5	6	<input type="checkbox"/>
6. NT & 2nd trimester blood		80%	5%	*	*	<input type="checkbox"/>
7. Age alone	0.8% of women referred for diagnostic testing based on age alone had a positive diagnosis of Down syndrome. [2]					<input type="checkbox"/>
Do not know						<input type="checkbox"/>

* There is little data on this option. Detection is increased by 25% (compare to each test on its own) [3]

3. NT Provider Survey

27. If a programme is funded, should women who want only parts of the programme, or invasive testing even if screen negative (that is low risk) have their tests paid for?

- Yes No Don't Know Other

28. Please give the reason for your answer

.....
.....

Additional comments

.....
.....
.....
.....

**FAX to 09 373 7900 or use the FREEPOST envelope provided.
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.....

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Appendix 3

Survey questionnaires results

A3. Results of Survey of Current Practice in New Zealand

A3.1 Type of practitioner and survey responses

Table 14: Type of midwifery practice obstetric practice

Midwives	No.
Independent	139
Hospital (core staff)	2
Hospital (case loading)	8
Total	149

Table 15: Type of

Obstetricians	No.
Private	10
Public	21
Private & Public	31
Registrar	11
Total	73

Table 16: NT scan practitioner

NT provider	No.
Radiologist	23
Sonologist	1
Sonographer	21
Not known	2
Total	47

Table 17: Training in New Zealand

	Midwives n=149		Obstetricia n=73		GP n=209		Total N=431	
	n	%	n	%	n	%	n	%
Yes	88	59.1	50	68.5	130	62.2	268	62.2

Table 18: Years of experience

	Midwives n=149		Obstetricia n=73		GP n=209		NT provider n=47		Total N=478	
	n	%	n	%	n	%	n	%	n	%
≤ 5 years	35	23.5	17	23.3	24	11.5	6	12.8	82	17.1
> 5 years	113	75.8	49	67.1	177	84.7	40	85.1	379	79.3

Not known	1	0.7	7	9.6	8	3.8	1	2.1	17	3.6
-----------	---	-----	---	-----	---	-----	---	-----	----	-----

Table 18a: Years of experience and screening offered to women

Midwives

Years experience	All women		Risk based screening		Total	
	n	%	n	%	n	%
≤5	25	71.4	10	28.6	35	100.0
>5	79	69.9	34	30.1	113	100.0

1 midwife did not offer screening and was removed from calculations

Obstetrician

Years experience	All women		Risk based screening		Total	
	n	%	n	%	n	%
≤5	11	64.7	6	35.3	17	100.0
>5	33	67.3	16	32.7	49	100.0

Years of experience unknown for 7 practitioners

General Practitioner

Years experience	All women		Risk based screening		Total	
	n	%	n	%	n	%
≤5	9	40.9	13	59.1	22	100.0
>5	52	31.0	116	69.0	168	100.0

Years of experience unknown for 5 practitioners

14 GPs did not offer screening, 11 had over 5 years experience and 3 did not complete the question.

A3.2 Screening offered to women

Comments relating to why they offer screening to all women or particular groups.

All women

Midwife

"The nuchal fold screening has become a fashionable scan and women have often had 2-3 scans before 12 wks by GPs. I feel this should be offered to women over 35

(esp. 1st baby and women with family history) government should not be funding these scans to everyone" (155)

"explain scan is funded by MOH for detection of abnormality primarily down's if termination not an option then why have a scan at all. Amazed how few people understand that scans are for detecting abnormalities" (296)

"used to be >35, previous baby & those that asked, now advise a 12 wk scan as an option for NT - strongly recommended >35 & other factors" (351)

"right for women to know. Would hate to not discuss & then woman angry because she could have found out." (401)

Obstetrician

"haphazard introduction of NT scanning in NZ without programme. Would be supportive of such & feel I disadvantage someone booking at our practice if I don't mention it as it is so widespread" (361)

"used to limit in early days to >30 but last few years become inclusive and since rate with the younger women has become more important and test more accepted." (109)

"Later onset of childbearing means more women at higher risk of DS i.e. older" (113)

GP

"Down's syndrome can occur at any age. Nuchal translucency is non-invasive" (158)

"Most 'ask' re it - explain re risk factors (age, previous history etc.) with all" (214)

"Because the technology is available." (53)

Risk based screening (i.e. not all women offered screening)

Midwife

"because at present the screening facilities in NZ do not generally allow for every women to be screened" (304)

"is free for 35 & over." (349)

"feel families focus on Down Syndrome screening & often see it as a panacea to ensure their baby will be 'normal'. Have personal conflict with selection of those babies worthy of continued life or not. Am aware of conflict & present info in unbiased way." (353)

"am not in this for other people / companies to make money" (415)

"My understanding is that screening should be offered to high risk women i.e. over 35 &/or previous affected baby" (133)

Obstetrician

"Limitations on screening options & availability (currently available where I practice)" (348)

"increased risk over 35. If women request info then obliged to give it." (377 Registrar)

GP

"...I mention nuchal thickness scanning but do not believe all women should have this. Some declined, sometimes hard to establish when 12-14 wks. Generally don't mention serum screening due to cost" (196)

"there is so much to organise that we target those illnesses with the highest risk incidence first. Frightening people with lower incidence illnesses for the sake of informed consent is cruel" (237)

"Screening involves risk to healthy baby so don't initiate discussion in younger women even though more DS babies born to younger women (because younger women have more babies) Increased risk increase age/previous Downs baby" (249)

"time constraint / cost" (391)

"Laziness, concern about testing for a condition that could only be "prevented" by abortion"(8)

"I think I actually discuss with most patients, but maybe less likely to in young girls (under 25)" (66)

A3.3 Knowledge levels of practitioners

No further data

A3.4 Purpose of screening

Selected practitioner comments.

Midwife

"Detect affected fetus & give women choices whether or not they would terminate. I strongly advise them not to opt for screening if they wouldn't terminate" (352)

Obstetrician

"Depends on your view 1. cost effective reduction of incidence or 2. patient service" (322)

GP

2 GPs stated they thought the answer to this question was obvious and provided not further info.

A3.5 Screening tests offered to women

Selected practitioner comments.

Midwife

"Don't recommend but offer all options & assist women to understand screening & choices & consequences" (198)

"Let women decide, discuss range of tests available" (208)

"NT is combined with blood test if results borderline or reluctant for amnio if at all possible." (351)

"Some women prefer to have CVS in spite of miscarriage risk" (358)

2nd trimester blood test

"2nd tri not available in Whangarei" (266)

"Don't offer as NT easily available & only \$25 but bloods \$80 & difficult to organise." (192)

"Can't sign 2nd trimester blood test form - would if could" (390)

"Didn't know there was 2nd trimester serum test in NZ. Asked recently -told them not available" (128)

"Results come too late for most patients" (117)

Obstetrician

2 Obstetricians did not complete this question – one stating that they "never recommend any type of screening - offer to couples"

"2nd trimester - not routinely due to cost \$75. NT & 2nd - did not think available in NZ (as in Australia) ? Only in private" (381)

"would like to offer NT & 2nd tri when available." (92)

"combined test not available" X 3

2nd trimester offered on request (310)

NT if under 35 (97).

"[Option offered] only because it is available" (385)

GP

"2nd trimester rarely. More often they ask for it even after negative NT" (53)

"Amniocentesis if NT positive" X2

"don't understand some options" (254)

"offer scan to all and amnio to those over 37yrs or other risk factors" (289)

"only suggest amniocentesis" (163)

"Triple test not available" (246)

"Age is not a screening test. 2nd tri & NT - this is very good but ?available in here." (7) X2

Table 19: Screening tests offered with District Health Boards

Screening tests recommended:	Total N=18	Total (%)
Maternal age	1	5.6
NT scan	14	77.8
Second trimester blood	5	27.8
NT scan & second trimester blood test	0	0.0
Other - amnio	10	55.6
- 2nd trimester US markers / anatomy scan	1	5.5
Not done at hospital -LMC / GP / obstetrician responsibility	3	16.7

No DHB had a guideline for antenatal screening for Down Syndrome. Four DHBs provided some direction to their staff on screening.

A3.6 Practitioner knowledge about gestation at which screening tests undertaken

The highlighted areas indicate the generally agreed acceptable range for the correct time for testing.

Table 20: Gestation at which NT scan undertaken

	Midwife	Obstetrician	GP	Total	%
6, 8	0	0	2	2	0.5
8-11	0	0	2	2	0.5
9-13	0	1	0	1	0.2
10	0	0	4	4	1.0
10-11	0	0	1	1	0.2
10-12	0	0	4	4	1.0
10 - 13	2	1	0	3	0.7
10-14	0	1	4	5	1.2
11	3	1	17	21	5.2
11-12	5	5	9	19	4.7
11-13	39	21	15	75	18.7
11-14	43	15	13	71	17.7
12	21	8	47	76	18.9
12-13	11	6	11	28	7.0
12-14	13	6	14	33	8.2
12-16	0	0	1	1	0.2
13	4	0	5	9	2.2
13-14	0	0	1	1	0.2
14	1	1	5	7	1.7
14-18	0	0	1	1	0.2
18	0	0	4	4	1.0
19 - 20	1	1	2	4	1.0
35	0	1	0	1	0.2
Don't know / Not completed	3	5	21	29	7.2
Total	146	73	183	402	100.0

Depends on scanner

11-14 (nasal 12-14)

Some practitioners put a range and an ideal gestation

Midwife: 3 stated 12 weeks was ideal and 1 stated 11 weeks

Obstetrician: 1 stated 12 weeks preferable and 1 stated 13 weeks

GP: 1 stated 12 weeks preferable

Table 21: Gestation for second trimester serum screening

	Midwife	Obstetrician	GP	Total	%
12	0	0	1	1	0.2
12-14	1	1	2	4	1.0
12-16	1	0	1	2	0.5
12-17	0	0	1	1	0.2
13	0	0	1	1	0.2
13-16	0	0	1	1	0.2
14	4	1	8	13	3.2
14-15	1	2	4	7	1.7
14-16	4	3	3	10	2.5
14-17	1	2	0	3	0.7
14-18	1	3	1	5	1.2
14-20	8	2	9	19	4.7
15	27	18	19	64	15.9
>15	1	0	1	2	0.5
15-16	8	9	2	19	4.7
15-17	13	3	2	18	4.5
15-18	0	3	4	7	1.7
15-20	3	2	2	7	1.7
<16	0	1	0	1	0.2
16	13	9	23	45	11.2
16-18	3	3	8	14	3.5
16-20	0	0	1	1	0.2
<17	1	0	0	1	0.2
18	0	0	7	7	1.7
18-19	1	0	0	1	0.2
18-20	0	0	1	1	0.2
19	0	0	3	3	0.7
19-22	1	0	0	1	0.2
14-22	0	1	0	1	0.2
20	2	0	2	4	1.0
24	1	0	0	1	0.2
24-26	1	0	0	1	0.2
28	1	0	1	2	0.5
Not used/don't offer	7	0	11	18	4.5
Didn't know was available	3	0	0	3	0.7
Don't know gestation	21	5	39	65	16.2
Not completed	18	5	25	48	11.9
Total	146	73	183	402	100.0

A3.7 Risk threshold used by practitioners

Table 22: Risk Threshold

Risk threshold given	NT provider	Midwife	Obstetrician	GP	Total	%
2mm	1	0	0	0	1	0.2
2.5mm	1	0	0	0	1	0.2
>2.5mm	1	1	0	0	2	0.4
>2.8mm	1	0	0	0	1	0.2
3mm	2	0	0	0	2	0.4
1:50	0	3	1	2	6	1.3
1:80	0	0	0	1	1	0.2
1:100	0	4	4	19	27	6.0
1:200	3	0	1	1	5	1.1
>1:250	1	0	0	0	1	0.2
1:250	5	38	43	39	125	27.9
1:250 – 1:300	1	0	0	0	1	0.2
>1:300	3	2	0	0	5	1.1
1:300	20	14	4	0	38	8.5
1:300 –depends on mat age and measurement	1	0	0	0	1	0.2
1:350	0	1	0	0	1	0.2
≥ 1:400	1	0	0	0	1	0.2
1:500	0	12	1	9	22	4.9
As per Fetal Medicine Foundation	2	0	0	0	2	0.4
Depends on maternal age & fetus	1	0	0	2	3	0.7
No threshold – use risk assessment from programme	1	0	0	0	1	0.2
No threshold - adjusted risk is given & compared to risk of misc. from amnio/CVS & level of 'risk' is a personal choice for parents	1	0	0	0	1	0.2
Women's choice	0	3	4	3	10	2.2
Based on radiologist	0	2	0	0	2	0.4
Other	0	1	5	3	9	2.0
Offer discussion	0	0	1	1	2	0.4
Offer to all	0	1	0	0	1	0.2
Do not know	0	48	5	77	130	29.0
Not completed	1	16	4	26	47	10.5
Total	47	146	73	183	449	100.0

Midwife

"1:250 cut-off or if higher than women's age" (284) "increased risk>than age to risk following screening" (198)

"1:50 mat serum, 1:300 for NT" (150) (not put in table above yet)

"1:500 If risk greater of miscarriage than risk of DS wouldn't do (416)

"Ambiguous question. If you mean offering amnio to women <35 then 1:300" (264)

"none offered to all. Depends on age - at age 46 1:6" (263)

"offer it to all." (204)

Obstetrician

"Depends on assessed risk of amnio." (322) (? What this means – have put in other)

"depends where you work gave both 1:100 & 1:250 - we risk losing a baby by amnio/CVS" (335)

GP

"1:250 for women over 35" (36)

"1:100 my reading, 1:250 recommended." (269)

"tend to go on age" (220)

A3.8 Topics covered in discussion with women about screening

Table 23: Topics included in discussion about DS screening

	Midwife n=146	Obstetrician n=73	GP n=183	Total N=402	%
Information about Ds and other chromosomal abnormalities	99	65	107	271	67.4
Types of screening options	130	69	128	327	81.3
False positive & negative results	114	64	109	287	71.4
Need for a diagnostic test	100	64	102	266	66.2
Support networks if positive diagnosis	70	27	62	159	39.6
Termination of pregnancy	100	67	120	287	71.4
Other					
- availability of genetic couns.	1	0	0	1	0.2
- availability of Obstetric spec.	3	0	0	3	0.7
- continue pregnancy if positive	2	0	1	3	0.7
- scanning providers & quality	2	0	0	2	0.5
- do what is right for family	2	0	0	2	0.5
- consider what would do with info	0	1	0	1	0.2
- could indicate other complications	1	0	0	1	0.2
- detailed anatomy scan at 18/40	0	2	0	2	0.5
- whether assisted fertility	0	1	0	1	0.2
- aims of screening	0	0	0	0	0.0
- fathers age	0	0	1	1	0.2
- option of not having a screening test	0	0	1	1	0.2
Not completed / NA	9	2	27	38	9.5

Midwife

“depends on age - which tests I discuss” (416)

“discuss diagnostic tests if in high risk group > screening test or if maternal choice >35 yrs or if family history of chromo disorder.” (162).

Other topics covered

“different scanning providers & their expertise doing the NT.” (143)

“other associations e.g. could indicate heart defect or kidney problem.” (330)

“reality of impact of FPR and risk of abortion after amnio - if elect to continue better to receive diagnosis at child's birth.” (344).

Obstetrician

"If don't want top not much point having test." (428)

"Support networks only when diagnosed (361)

GP

"in context of race & religious beliefs." (228)

"my own experience (elderly primip)" (220)

"need pamphlet , only 15mins & get paid \$32." (246)

"people usually know what is meant by Downs -I check they do." (312)

"re support networks - but should / will." (189)

"Top ? Would they - if not don't screen" (387)

"top only if brought up by women." (47)

A3.9 Consent for screening

Table 24: Consent for screening

	Midwives	Obstetrician	GP	NT provider	Total N=449	%
Oral	119	64	129	31	343	76.4
Written	13	7	14	0	34	7.6
Oral (NT) & bloods	2	0	2	0	4	1.3
written						
Not completed	12	2	38	1	53	11.8
NA	0	0	0	15	15	3.3

A3.10 Giving of risk estimates

Table 25: Method of giving risk estimates if screen 'positive'

	Midwives	Obstetrician n	GP	Total N=402	%
Face to face	89	55	120	264	65.7
Telephone	9	0	4	13	3.2
Face to face / telephone / post	31	12	4	47	11.7
Other	2	2	4	8	2.0
Given by radiologist	8	1	9	18	4.5
Not completed	7	3	42	52	12.9

- A total of 16 midwives said the NT provider was the first person to give women the results, 6 of these did not indicate results were given by themselves face to face/telephone or post.

Midwife

“Women read scan result & phone otherwise I ring women” (135)

Obstetrician

“Face to face if risk is >1:100” (396)

Table 26: Method of giving risk estimates if screen ‘negative’

	Midwives	Obstetricia n	GP	Total N=402	%
Face to face	61	31	67	159	39.6
Telephone	33	21	45	99	24.6
Face to face / telephone	30	10	9	49	12.2
Post	2	1	4	7	1.7
Other	0	3	7	10	2.5
Given by radiologist	11	1	9	21	5.2
Not completed	9	6	42	57	14.2

Midwife

“Usually told when having scan if results negative then post - occasionally telephone to answer questions” (210)

GP

“Can't be negative” (154)

“Left to radiology” (177)

Table 27: Risk level given by NT scan provider

Give woman risk level at time of NT scan	No.	%
Never	9	19.1
Sometimes	21	44.7
Always	15	31.9
Not completed	2	4.3
Total	47	100.0

A3.11 Recommendations if screens 'positive'

Obstetrician

"Invasive prenatal testing, 2nd trimester scan if patient refuses the above" (317)

"suggest diagnostic testing if TOP is to be considered" (396)

GP

"give her options to terminate pregnancy or continue with pregnancy, if wants to continue regardless then screening not offered in 1st place" (75)

NT provider

"Don't agree with this concept. There is only increasing risk. Results are sent to LMC or referrer. If greater than 1:250 referral to obstetrician advised." (6)

"Discuss with LMC. If they will terminate an abnormal result then have amnio, if not no amnio as risk of amnio about 1:100." (14)

"Should have amnio (100%) as NT isn't." (15)

"Needs to decide if want further testing i.e. CVS or amnio. Explain where & how procedures done ... Depending on risk will recommend early anatomy scan if negative for DS." (29)

"Feedback from LMC unless specifically ask." (20)

"Show her the results. Amnio to be considered & tell her to get in touch with her LMC to discuss options (after she has thought about it & personalised it with partner), pamphlet given." (75)

a) Information given about fetal loss rate with amniocentesis

Low fetal loss with amnio due to practitioner skill

"1% HVH stats 2% national stats" (122)

"Literature 1%, practice now 0.5%" (416)

"Depends on practitioner" (173) X2

"Our operators are highly exp. (234)

"NWH amnio clinic say 0.5% with their Drs" (182)

"0.5% at ACH" (120)

"radiologists in TGA claim their rate is well below 1% due to their experience" (263)

A3.12 Responsibility for screening

Table 28: Provider perceived to currently be responsible for screening

Thirty five practitioners stated that it was the first health professional that the woman visited that was responsible.

	Midwives n=146		Obstetricia n=73		GP n=183		NT provider n=47		Total N=449	
	n	%	n	%	n	%	n	%	n	%
LMC	127	86.9	64	87.7	145	79.2	43	89.4	379	84.4
GP before booking	60	41.1	21	28.8	87	47.5	26	53.2	194	43.2
District Health Board	12	8.2	9	12.3	8	4.4	4	8.5	34	7.6
Ministry of Health	12	8.2	11	15.1	7	3.8	4	8.5	34	7.6
Other Obstetrician	3	2.1	3	4.1	4	2.2	1	0.0	11	2.4
First practitioner	20	13.7	1	1.4	15	8.2	0	0.0	36	8.0
No-one	0	0.0	1	1.4	0	0.0	0	0.0	1	0.2
Random mixture	0	0.0	1	1.4	0	0.0	0	0.0	1	0.2
Not completed	5	3.4	1	1.4	7	3.8	0	2.1	13	2.9

- 15 practitioners selected all options

Comments relating to who is currently providing screening

Midwife

“screening NT takes place before LMC signed up - no guidelines.” (263)

“Booking cannot take place till 14 wks - too late for screening, this doesn't happen.

Women don't see LMC till too late for screening” (337)

Obstetrician`

“probably nobody as MOH not advised us” (389)

“if prepared to be LMC & paid to do so - should be able to inform & offer screening & refer appropriately” (381)

GP

GPs shut out of care

“all above as not registered with LMC till after initial scan” (172)

“With the government led demise of GP obstetrics GPs have little interest or input into ante-natal care, so the person best suited to provide good info on screening has been precluded, as a result of government strategy” (38)

“opportunistically now that women don't necessarily contact us” (87)

A3.13 Is it possible for a woman who screens 'negative' to have a baby with Down Syndrome?

No further data

A3.14 Education for Down syndrome screening

Table 29: Practitioners perception of current knowledge level and need for further education

Practitioners perception	Adequately educated N=449		Want further education N=449	
	No.	%	No.	%
Midwife	70	47.0	131	87.9
Obstetrician	65	89.0	57	78.1
GP	63	30.1	145	69.4
NT provider	22	46.8	34	72.3

Some practitioners qualified their response to this question by stating they only gave initial advice or they referred on to LMC or specialist.

GP

"initial advice only" (176)

"government severely reduced GP role in obstetrics" (3)

Education required - comments

Midwife

"but I don't see that independent MW should provide counselling" (123)

"able to give advice but not counselling" (129)

"updates on screening programme re pick up rates (my understanding is NT is 70% DS picked up & 30% missed. & depends on practitioner skill level" (162)

"education only useful if given in unbiased way i.e. not heavily laden with medical opinion" (167)

"if unbiased" (263)

"knowledge obtained from practice - not from training" (272)

"resistant unless paid more! Sick and tired of all the extra screening midwives are expected to do. Family violence, HIV, immunisation register" (296)

"believe however Down syndrome to be one of the least common abnormalities that can happen" (354)

Not required as:

"I don't need to be a specialist just need to know who to refer to" (121)

"already been practising 26yrs" (155)

"too busy" (166) – this LMC does not screen

"my role is awareness, support & referral" (193)

“have undertaken educations when NWH ran its trial on maternal serum. Take a leaf out of very successful serum study & how they educated care providers” (198)

“it’s the woman's choice, we just give advice” (284)

“there are experts already available to give advice” (403)

“am not a genetic counsellor” (415)

Obstetrician

“But would like to be able to offer a 12 wk combined nuchal / most like in Aussie” (306)

GP

Would like more info re combo test and bloods X 4

“more on nuchal screening risk/age tables” (19)

“not adequately trained therefore do not do it. Pamphlet for patient would be good” (81)

“questionnaire has led me to read MOH site - I do need an update. Are the guidelines such that we offer screening to all women now” (387)

Not required as:

There were many comments stating further education was not required as they referred women on to a LMC or specialist for this information.

“experienced GP” – (NB does not provide screening to all women)

Have already had training

“only providing info at pregnancy diagnosis. Inform as part of general education in case delay in finding a midwife. Not formal antenatal care.” (236)

“GPs are now excluded from maternity care so what is the point of continuing education . If this problem is addressed then of course” (312)

“probably wouldn't read it. Only want to have up to date info re local amnio skill re fetal loss (to give more accurate info)” (397)

Table 30: Method of further education

	Midwife n=146	Obstetrician n=73	GP n=183	NT provider n=47	Total N= 449	Total %
Workshops	84*	17	35	15	137	30.5
Written guidelines	74	32	101	16	207	46.1
Information booklet	95	22	68	13	198	44.1
Online education	49	28	31	17	125	27.8
None	8	14	21	9	43	9.6

A3.15 Future screening programme

Table 31: Provider who should provide screening

	Midwives n=146		Obstetricia n n=73		GP n=183		NT provider n=47		Total N=449	
	n	%	n	%	n	%	n	%	n	%
LMC	120	82.2	50	68.5	133	72.7	36	76.6	339	75.5
GP before booking	68	46.6	18	24.7	96	52.5	27	57.4	209	46.5
District Health Board	16	11.0	9	12.3	5	2.7	5	10.6	35	7.8
Ministry of Health	16	11.0	13	17.8	10	5.5	5	10.6	44	9.8
Other										
- Obstetrician	0	0.0	2	2.7	0	0.0	2	4.3	4	0.9
- PHO	0	0.0	0	0.0	1	0.5	0	0.0	1	0.2
First practitioner	3	2.1	1	1.4	2	1.0	0	0.0	6	1.3
Trained midwife / counsellor	27	18.5	11	15.1	19	10.4	14	29.8	61	13.6
Trained consumer	7	4.8	4	5.5	4	2.2	0	0.0	15	3.3
Not completed	7	4.8	3	4.1	5	2.7	0	0.0	15	3.3

“Currently no funding for providers. PHOs are only organisation who should be taking this responsibility - Mat services must be combined under PHOs & MW subcontracted to PHO not MOH” (9)

Midwife

“but LMC are not paid for that service” (123)

“risk needs to be assessed preconceptually”

“[Trained midwife / counsellor] or LMC once they are trained” (192)

“some women not put onto Mw till too late & miss opportunity for NT” (206)

“if screening means amnio then means DHB responsible” (272)

“trained midwife (not counsellor)” (330)

“LMC not signed till 14 wks so again need to be earlier contact – GP” (351)

“written info from MOH” (416)

a) Aspects of a good programme

Table 32: Aspects of a good screening programme as perceived by practitioners

	Midwives n=146		Obstetricia n=73		GP n=183		NT provider n=47		Total N=449	
	n	%	n	%	n	%	n	%	n	%
Pre-test written consumer information	106	72.6	60	82.2	116	64.4	39	83.0	282	62.8
Pre-test counselling	85	58.2	61	83.6	137	74.9	41	87.2	324	72.2
Post-test counselling of all women	43	29.5	44	60.3	79	43.2	25	53.2	191	42.5
Post-test counselling of women who screen positive	89	61.0	44	60.3	96	52.5	23	48.9	252	56.1
Only offer best option of screening	31	21.2	20	27.4	35	19.1	17	36.2	103	22.9
Offer a range of options	81	55.5	44	60.3	76	41.5	21	44.7	222	49.4
Informed consent	114	78.1	60	82.2	130	71.0	31	66.0	335	74.6
Laboratory quality audit process	74	50.7	61	83.6	104	56.8	33	70.2	272	60.6
Annual reporting	57	39.0	45	61.6	49	26.8	25	53.2	176	39.2
Not completed	6	4.1	1	1.4	14	7.7	3	6.4	24	5.3

Practitioners could select more than one response however some were intended to be incompatible such as offer the best option or offering a range of options. For this particular selection 20 practitioners selected both options and 112 practitioners did not select either but did complete other aspects of the question.

Midwife

Information sharing and counselling

“Essential women be pre test counselled. Had experience with high risk NT results devastating women till amnio results could be obtained” (133)

“written information is essential as different LMCs have differing opinions. Women should be asked to consider why they want the test, what they will do if they test positive” (278)

“LMC should be able to do pretest counselling & ? Post test counselling” (200)

“scanning quality audit. More specific counselling for women at greater risk i.e. from history & >35yrs” (264)

Concerns

“radiologists to be more informative instead of just scaring women” (142)

“because health dollars limited, need funding for best option supported by best evidence / research findings” (182)

“ensure adequately funded & doesn't deflect \$ from already under funded maternity services. Such as family violence which is currently under funded. HIV not funded. Any screening must be funded” (198)

Screening programme

“offer only best for high risk clients. Range for lower risk who are worriers” (178)

“good screening programme is essential” (192)

“should not be routine” (205)

“screening is a personal choice, not medical decision. Women should be offered choices, then left to make up their own minds” (364)

Obstetrician

Information sharing and counselling

“all these things good but? Practical to do pre-test counselling & post test counselling for everyone” (329)

Concerns

“clear knowledge of screening test is essential. Currently many LMC do not understand the distinction between screening & diagnostic test - women remain confused” (106)

Screening programme

“accessible to all, currently serum screening not used because of cost” (365)

“publicly funded. Radiology QA. Adequate training of person offering test - many LMCs are woefully ignorant of the implications of screening tests. Need best for pop purpose but range for individual purpose” (97)

“...Can offer a rang of options but best option should be highlighted” (104)

“best option if cost effective” (105)

“national programme” (260)

“need options as some women present later” (389)

“good population uptake is important” (429)

“test with reasonable S & S. No point screening those who would not take further action” (152)

GP

Information sharing and counselling

“If adequate pre-test counselling then women would be aware of false negatives” (56)

“most women grasp the issues pretty well” (57)

Concerns

“concerned that NT very operator dependent & am unsure if there is any auditing to test sensitivity & specificity in out town” (66)

“cost is huge issue in NZ” (226)

“not a perfect world, cost effectiveness needs to be considered.” (237)

Screening programme

“improved overall health outcome” (18)

“offer range if appropriate & valid & reliable” (87)

“to provide free or low cost screening, so that all women can participate” (197)

“needs to be simple, public sense should only fund best option, not whole range. If want this should pay” (255)

“Include GPs as often have to pick up emotional pieces without background info” (258)

“offer best option specific to each women's risk” (279)

NT provider

“proper train/funding of counselling is important” (34)

b) Who should be offered screening

Table 33: Should screening be provided free to all women?

	Midwives n=146		Obstetricia n=73		GP n=183		NT provider n=47		Total N=449	
	n	%	n	%	n	%	n	%	n	%
All women	89	61.0	62	84.9	85	46.4	28	59.6	264	58.8
Women who ask	21	14.4	1	1.4	22	12.0	2	4.3	46	10.2
Women over certain age (Total) ^a	50	34.2	11	15.1	80	43.7	16	34.0	157	35.0
- 30 years	3		2		4		5		14	
- 35 years	25		9		46		6		86	
- 38 years	2		0		2		0		4	
- 40 years	0		0		5		0		5	
- age not stated	20		0		23		5		48	
No women	4	2.7	0	0.0	0	0.0	0	0.0	4	0.9
Not completed	2	1.4	0	0.0	9	4.9	1	2.1	12	2.7

Eleven practitioners (3 midwives, 1 obstetrician, 7 GPs & 0 NT scan providers) stated screening should only be for women who ask. Practitioners who selected age also thought risk based screening should be added to the options).

c)Funding for programme

Table 34: Funding for programme

	Midwives n=146		Obstetricia n=73		GP n=183		NT provider n=47		Total N=449	
	n	%	n	%	n	%	n	%	n	%

Yes	45	30.8	28	38.4	55	30.0	16	34.0	144	32.1
No	53	36.3	29	39.7	46	25.1	17	36.2	145	32.3
Don't know	33	22.6	9	12.3	56	30.6	7	14.9	105	23.4
Other (total)	10	6.9	7	9.6	14	7.7	5	10.6	36	8.0
-yes if high risk (age / fam history)	4		4		1		2			
-yes if parts of programme but no to invasive testing if screen neg	4		3		3					
- other	2		0		10		3			
Not completed	5	3.4	0	0.0	12	6.6	2	4.3	19	4.2

Other includes responses that indicated it was up the woman, depended on the situation or no alternative given.

Midwife

"Is screening a NT or amnio - not clear to me." (212)

"NT best of poor choices - explaining risk can be tricky" (363)

"Yes if >35 as low risk may not be enough reassurance for older women - may need reassurance of funded invasive testing" (182)

Obstetrician

"If want invasive tests regardless should proceed directly to that if can't dissuade them" (105)

"screening tests not good enough" (355)

GP

"Yes if want parts, or if neg & want invasive testing (unless old or high risk & want to exclude false neg. These people should have invasive test & not NT or blood)" (56)

"Should be able to choose if they only want part of programme & still be eligible for funding. If screen neg & <35 should not automatically be able to have funding for invasive testing" (36)

449 responses

Table 35: Screening tests

ONE OPTION

TWO OPTIONS

Not completed	
40	
Midwife	9
Obstetrician	1
GP	

Do not know	43
Midwife	15
Obstetrician	3
GP	

Nuchal alone	
23	
Midwife	
16	
Obstetrician	2

Combined	
119	
Midwife	
22	
Obstetrician	21

2nd trimester blood	
2	
Midwife	

Serum integrated	5
Midwife	0
Obstetrician	2
GP	

Integrated	
115	
Midwife	
44	
Obstetrician	22

NT & 2nd trimester Blood	14
Midwife	10
Obstetrician	1
GP	

Age alone	
7	
Midwife	
2	

NT & combined	
14	
Midwife	
3	
Obstetrician	2

NT & 2nd trim blood	
3	
Midwife	1

NT & Integrated	6
Midwife	1
Obstetrician	1

NT & Age	
2	

Combined & Serum integrated	
2	

Combined & integrated	
26	
Midwife	8
Obstetrician	10

Combined & age	
1	

Serum integrated & Integrated	
6	
Midwife	3

Integrated & 6	
1	

Integrated & Age	6
Midwife	3
GP	

THREE
OPTIONS

**Combined,
integrated & age
1**

**Second tri bloods,
Integrated and Age
1**

**NT, 2nd tri & Age 1
Obstetrician 1**

**NT, combined &
integrated
1**

**Combined, serum
integrated &
integrated
2
Midwife**

**Combined,
integrated & 6
1**

**Combined, 6 & age
1
Obstetrician 1**

FOUR
OPTIONS

**1 & 2 & 4 & 73
Midwife
2**

One midwife ticked all options and stated, “women have right to make informed decisions based on sound diagnosis & professional responses to personal questions & fears” (139)

There were comments relating to the possible increase in cost to the health system and the lack of costs given in the survey. Several practitioners also commented that they were not aware of the results of the SURUSS study.

"Presume blood test detects chromosomal abnormality only. NT shows up any other gross abnormalities..." (191)

"Least false positive. Lot of women don't want testing of any sort, don't grasp 'false positive' or 'screening' idea. Think NT scan = yes/no. At 11-13/40 may only see women 1 or at most 2 time therefore a lot of information at early stage." (271)

"...Not enough reliable info for rural LMCs. Where are blood tests tested? What is time frame for getting to lab? Cost?" (370)

"tragedy of this type programme is underlying eugenic philosophy (called by Sir William Liley 'search & destroy') to rid our community of DS people. What does this say of our attitudes toward disabled? Should put time & energy into caring for disabled not search ..." (116)

"1. am concerned NT has become so fashionable 2. don't want a screening programme to assume there is no place in society/family of handicapped child" (385)

"...Best to ask consumers I think" (177)

"Older women often fearful of pregnancy loss. Integrated test would reduce no's having amnio/CVS. Most women whatever age socialised into scans, very eager to have early scan - hence NT" (295)

GPs feel excluded from maternity care

"less invasive. Hard for GP to answer - rarely seen, excluded from decision making and problems during pregnancy. E.g. polycose may reflect health problems later in life. Was a GPO now irritated by this. Not offered serum test for 2 babies in 40's. ?Available down south? (258)

"GPs have been removed from consideration of opinions or consultation in all this issue" (147)

"if widespread formal screening is going to be instituted would have to be most reliable testing we can afford. This could provide impetus for reengaging GP in 1st trimester care & building on this with other important screening like chlamydia, syphilis, substance abuse" (312)

Poor information provided in survey

"think table is very poor way of explaining SURUSS results. Most LMC's will not understand OR of false pos. Why not present it as the SURUSS study did in its summary? - too late now I suppose? i.e. DR for 3% FPR. What loss rates for amnio/CVS are used in table. costs i.e. should also be a factor in national decision making. (95)

"This is rubbish. You need to look at you 2x2 table again. Combined 1st trimester testing seems to be getting better sensitivities than you are quoting??" (322)

"Suruss is only 1 report avail. Others show improved results from combined 1st trim testing e.g. FMF. (389)

1. NT scan alone

Reasons

Midwife

"Because it gives an earlier result & can be counselled re further testing & not many false pos here [Christchurch] from NT" (117)

"In my 8 years of practice I haven't had one false pos NT. They are getting accurate" (159)

"Most reliable" (171)

"NT seems to be more accurate than maternal serum blood test" (274)

"Nuchal scans only will save NZ dollars and if needed amnio should be offered. Women today are fearful enough without being spoken to about tests that have a false/positive result." (284)

"For women the NT seems less invasive. Blood test is still only screening. If major concerns – amnio" (326)

"Most women want 1st trimester USS, it is therefore possibly more cost effective to do than blood test especially in 2nd trimester. Very difficult psychosocially /emotionally for families if late top. Also US socially acceptable & expected now (unfortunately)" (162)

"Subjecting women to extra testing in pregnancy can lead to medical style of care & detract from the pregnancy. Most women want screening to be non-invasive to her & baby. For some DS is part of life -would never consider TOP. Expectation baby normal after anatomy scan" (359)

Obstetrician

"NT is currently the best (in my opinion) available single screening test. Too many tests will be too time consuming and expensive" (106)

"2nd trimester blood test leaves little time for amnio & consider options. Think it is only useful to screen those who would go on to have amnio & top when DS confirmed" (152)

"If it is going to explore entire antenatal pop need a single episode test. Whilst multiple tests yield greater accuracy for see major difficulties implementing to wider AN pop esp. lower SE groups" (336)

GP

"NT followed by further testing if possible. Simple to counsel re results." (254)

"My practice is to offer NT as also benefits of twin diagnosis etc..." (269)

2. Combined

Most comments related to this test having lower false positive rates with results being available to women in the first trimester for earlier decision making.

Midwife

7 midwives commented that earlier test allowed for earlier TOP

"Good to get results early in pregnancy to allow for top if required. Seems better than NT alone" (168)

Obstetrician

3 obstetricians commented that earlier test allowed for earlier TOP

"Most women would prefer to know earlier rather than later the results if abnormal" (101)

"Screening is complete by end of 1st trimester. Perhaps the integrated test could be offered to women under 35 - will allow risk of T21 & 1st trimester screening for over 35" (165)

"Current practice" (201)

"little further gain from integrated test with increased cost" (396)

"both results available at the same time to allow decision making early in 1st trimester. My great concern re 1st trim is that some O&G specialists ignorant to talk about it but also refuse to refer patients for NT even if patient chosen to do so" (374)

"Women properly counselled relatively unlikely to proceed to amnio. Litigation risk from screen negative, women with DS unable to afford private amnio" (429)

GP

"think 1st trimester screening is preferable - could consider 2nd blood test if high risk after option 2 if prefer not to have an amnio but want more information" (16)

"NT early & high detection rate but if positive need further test to decrease number false positive before invasive test. Generally offer NT if positive give further options. If not top -discuss risks amnio & often women decline it." (434)

3. Second Trimester Blood test

Midwife

"NT waste of time - so many women misguided by scan - just depends on the ultrasonographer - there has been many mistakes. DS common in Middle East where 1st cousins marry but not ethical to screen Islamic. But think they should have option to be screened." (155)

"Is not diagnostic therefore information irrelevant & unreliable. High risk women should have definite tests with yes/no outcome or why bother? Offers false assurance. Also USS exposure not proven to be safe." (330)

4. Serum integrated test

Obstetrician

"Low false positive rate yet no loss of sensitivity. You will notice I have changed my mind since q.11" (109)

"Thanks for trying to provide some clarification in this confusing area" (335)

GP

2 comments relating to it being less expensive and less invasive.

5. Integrated

Most practitioners who selected this option commented on the low false positive rate and low amnio rate and fetal loss.

Midwife

"Clearly this option will result in the fewest healthy babies lost and reduce unnecessary amniocentesis. However I am unaware of the blood tests. If initial results from 1st blood test and NT are low risk, 2nd bloods should be omitted." (278)

"...Integrated test should be offered for all women >35yrs who request it." (308)

"Gives best DR with least FPR but is a lot of testing. Wonder if stress of waiting is worth it & why not enjoy the excitement of having a baby. Always start with question 'would you terminate if abnormality found? In practice many prefer NT, if positive have amnio" (349)

"...Do not want to see guidelines recommending all women have NT scans. Want to see respect for people who choose minimal intervention" (351)

"... If more funding available would be preferred option statistically. Would bypass if women over 38 or have family history as risk factors would indicate amnio as more reassuring" (379)

"Number of losses of babies without DS greatly reduced with good incidence of detection -85%. Number of false positives in my area is high. Does this reflect the skill of sonographer? Is it coincidental? Or is the formula for calculation wrong?" (394) – Taupo

Obstetrician

"All tests have same detection rate but no. 5 has lowest FPR & loss rate with highest OR of those being detected being affected. However need to evaluate cost & perhaps choose slightly less accurate test if more cost effective e.g. #4 or #2" (381)

GP

"depend on when 2nd trim bloods can be done & how quickly results obtained - if at same time as amnio seems best option. Despite protests of disability groups I think testing is appropriate for those who want to consider top" (7)

6. NT scan & 2nd trimester blood test

Midwife

"women feel reassured with both scan & blood test." (211)

"Through discussion regarding triple test was informed was the most appropriate test. If questionnaire finds my knowledge to be inadequate I would most definitely consider a workshop to be of benefit." (272)

7. Age

Midwife

"Need to provide confident screening/diagnosis, don't want to make low risk women fearful of what is normal. Also partial testing can give false sense security. Testing should only be offered to high risk i.e. older previous history" (208)

GP

"I think the other options are not specific enough" (23)

"higher rate of possibilities of complications & fetal abnormalities with age remains most important factor" (75)

"costly screening & high FP in many 1st trimester tests. Personally don't think late top based purely on chromosomal study is justified" (89)

"highest detection rate" (240)

1 & 2

GP

"women may not turn up for 2nd tri blood test" (62)

"thought pick up was higher (for NT & combined) NT alone - early screening, more options for Top if needed. Should offer additional 1st trimester bloods if results give better pick up." (84)

"got to keep it relatively simple to work. Multiple tests will have a lot of non compliance. Results seem good on this alone as well." (188)

1 & 3

Obstetrician

"SURUSS is only 1 report avail. Others show improved results from combined 1st trimester testing e.g. FMF. Tidy to organise screening together/ early on. Earlier diagnosis/more options for management. Many women prefer 1st trimester Top. Much easier to organise" (389)

1 & 5

Midwife

"If we are truly screening those with existing risk factors I prefer no 5. For many casual requesters I prefer no. 1" (185)

Obstetrician

"as it is non invasive. Integrated test looks good too. But we have not been doing - might start that" (378)

1 & 7

GP

"Don't believe blood test is useful" (234)

2 & 4

GP

"if positive result should be available early in pregnancy as possible so combined test quite good. If waiting for 2nd trimester bloods including NT does not increase specificity much & assume would be more open to operator error & subjective." (66)

“choice is essential in screening” (424)

2 & 5

Most comments stated integrated test performed better but women should have option of earlier testing if desired.

Midwife

“NT alone has high FPR, most women ask for NT & FPR causes undue stress & intervention. Have read Suruss -makes sense to offer comprehensive test rather than piecemeal approach that is currently on offer” (210)

3 & 5 & 7

“Important that women who book late have screening options & those who book early have the best.” (95) – does not agree with selected answers.

A3.16 NT scan providers

Table 36: Number of NT scans per month

Number of scans	Number of responses	
≤10	9	
11-20	11	
21-30	9	
31-40	5	• One practice with 20 practitioners included in response stated 135 NT scans were undertaken each month.
41-50	5	
>50	6	• One response indicated 80-100 NT scans were undertaken
Not stated	12	
Total	47	

Risk calculation programme and quality assurance

A computer software package can be used by NT scan providers to calculate the risk of Down syndrome. Practitioners were asked to indicate whether they had such a programme, had they undertaken training to use the programme and whether they were registered by the Fetal Medicine Foundation (Table 37). Practitioners were also asked to describe the quality assurance process used for NT scanning (Table 38)

Table 37: Risk calculation programme for Down syndrome

	Number N=47	%
Training to use programme	46	97.9
Registered with Fetal Medicine Foundation	35	74.5

Two practitioners stated they personally were not registered but the practice was.

- 92% indicated they had a quality assurance programme for NT scanning
- 3 practitioners indicated they did not have a QA process
- 2 practitioners did not complete the question

Table 38: Quality assurance process

Type of Quality Assurance	No. n=47	%
Fetal Medicine Foundation	36	
Internal audit	17	
Auditing (type not specified)	2	
Not completed	5	

*excludes those that indicated they did not have a QA process

Giving of risk level to women

Table 39: Giving of risk level to women by NT scan provider (copy)

Give woman risk level at time of NT scan	No.	%
Never	9	
Sometimes	21	
Always	15	
Not completed	2	

Table 40: Counselling provided grouped according to whether give risk levels (for those that completed the following questions)

	Never n=8	Sometimes n=20	Always n=14	Not completed n=1
Provide pre-test counselling	2	5*	8	1
Provide post-test counselling	0	4	10	1
Expect LMC to counsel	7	20	13	0

*One practitioner stated this counselling was limited and another that it was undertaken while scanning.

- Nine practitioners added comments emphasising that they felt the LMC should provide counselling and five also included that this frequently did not occur.

•
“Lot of patients have no idea what NT is - expect absolute assurance about Down syndrome not risk assessment.” (24)

“LMC should provide counselling but they don't often.” (9)

“Always ensure patient has been informed however due to differences between what has been said/heard/understood I have taken on an active role - should be from LMC!” (4)

Cost of NT scan

Table 41: Cost of NT scan

Cost (\$)	No.
0	2
10	1
0-20	2
20	8
25	8
30	10
35	6
0-35	2
40	1
50	2
95	2
Not completed or did not want to disclose	3

- The cost to women for a NT scan ranged from \$0 -\$95 with most costing between \$20 and \$40.
- Some practitioners stated this was the same for all pregnancy scans.
- Several also indicated there was no charge to Community Service Card holders.

	Number N=47	%	Waiting times for NT scan Table 42: Waiting time for women to have a NT scan
No wait	8	17.0	
1-5 days	23	48.9	
5- 7 days	2	4.3	
10 days	1	2.1	
1-7 days	3	6.4	
1-14 days	2	4.3	
7-14	3	6.4	
Other	2	4.3	
Not completed	3	6.4	

Other

- booked for 12-14 weeks even if same day
- varies

Four answers were qualified by saying that if it was urgent it would be done immediately.

Appendix 4

New Zealand College of Midwives statement



NEW ZEALAND COLLEGE OF MIDWIVES (INC)

20 September 2005

Diana Austin
University of Auckland
Department of Obstetrics and Gynaecology
Private Bag 92 019
AUCKLAND

Dear Diana

Re Down Syndrome Questionnaire

Further to a recent conversation with Peter Stone we have drafted up a response from New Zealand College of Midwives (NZCOM) to add to the views you are receiving from the focus groups you are holding on the issue of antenatal screening for Down syndrome and other chromosomal conditions. This response has been circulated around our regional contacts for not only additional comments but also to ensure the response reflects the current view of the NZCOM National Committee.

As a College we are unable to provide you with a mailing list for this questionnaire. As discussed with Peter the midwifery profession is frequently being approached to participate in research. As a result our membership has made clear the parameters within which we can access the database. Essentially the database is available for research that is commissioned by the profession and supported by its membership.

The New Zealand College of Midwives represents over 80% of the practising midwifery workforce in this country. Its members are both self employed and employed. The structure and processes of NZCOM uphold the principle of partnership between the profession and the public. There has been consumer representation as of right in NZCOM's committees since its foundation. This right exists at every level of governance, nationally within the National Committee and regionally within professional review and complaints committees. There are ten regional committees, and five sub committees in the smaller provincial centres.

Midwives in New Zealand have been educated here and/or in the United Kingdom and Australia. According to our latest data the average age of midwives is now around 50 years. We know that over 40% of midwives' clients have a rural coding. Midwives are involved in either LMC caseloading as employed or self employed, core midwifery employed by a maternity facility and there is a small number employed in education and policy/administration

There is an expectation, within the midwifery Handbook for Practice, that midwives discuss the possibility of chromosomal abnormality with women. The older the woman is the more pertinent the discussion is because the diagnostic endpoint of screening – amniocentesis – is only funded for women over 35 years.

We would estimate from discussions that the knowledge of most midwives would be good in relation to Down syndrome. The screening for Down syndrome has become more complex in the past five years, particularly as other screening methods have surreptitiously entered the screening area. We have determined that midwives knowledge of these tests is good to very good, but the public perception of the validity of some of this screening determines what is offered to/demanded by women.

Midwifery has a view that each woman is an individual. The purpose of discussing screening with women is to ascertain which women may have the risk factors or concerns that necessitate them being offered screening. Unfortunately the societal view is that the screening is the diagnosis and that having the screening confirms the baby does not have an aneuploidy. This is particularly so in the case of nuchal translucency.

It would appear from discussions nationally that nuchal translucency in isolation at 13 weeks followed by an amniocentesis at 16 – 18 weeks is currently the most common method of screening for Down syndrome. The nuchal translucency is generally done even in isolation of risk factors and maternal age and is only the thickness of the nuchal fold on scan. This then necessitates a further scan within the secondary service where the other factors are included. We note however, that even if this comes back within normal limits, the maternal anxiety by that time means women are wanting an amniocentesis for them to have 'peace of mind'.

It is of concern to NZCOM, and something we have debated at length both internally and more widely in the past years, that women require good information around which to base screening decisions. Unfortunately this is not the case currently. Many women see that an ultrasound is no longer a "screening tool" but rather an opportunity to "see their baby". The fact that

on viewing the baby an abnormality may be detected comes as a shock and surprise to women.

Even having an amniocentesis with a 1% chance of foetal loss become less important due to the “screening” that has occurred previously in such an ad hoc way that anxiety is raised.

In conclusion, midwives do feel confident having the initial conversation with women about screening whether it is Down syndrome or other chromosomal conditions. Our concerns at present are the ad hoc approach to screening that appears to have developed through the popular media advocating nuchal translucency scans and the increasing use of scans particularly during the first trimester. Currently the lack of an overview of this screening is contributing to an expensive ultrasound budget.

We consider that information needs to be improved for consumers to ensure they are informed when agreeing to be screened. We also consider that appropriate referral templates need to be available in DHBs for when questions occur, during the screening of a well woman, as a result of an unexpected discovery that needs to progress to a diagnostic test.

Down syndrome is only one of the many things screened for in pregnancy. Discussion between women and midwives determine that a positive result for Down syndrome does not always indicate a decision to terminate the pregnancy. Women sometimes want to know to prepare themselves whilst other will choose to terminate. NZCOM considers that the decision to screen, to progress to diagnostic and then the decision that follows are for each woman and her family/whanau to make. We would just like to see a more streamlined, evidence informed, cost effective screening introduced as a first point.

Thank you for the chance to contribute to this work.

Yours sincerely

Norma Campbell
Midwifery Advisor
New Zealand College of Midwives
On behalf of the NZCOM National Committee

down syndrome screening.september2005

Appendix 5

New Zealand Down Syndrome Association Position Statement

NZDSA position statement on pre-natal testing for Down syndrome

March 2004

The New Zealand Down syndrome Association believes that people with Down syndrome have a right to life.

- We welcome babies with Down syndrome
- We believe people with Down syndrome can lead full and satisfying lives and enrich the lives of those around them.
- We provide support and information to people whose lives have been changed by Down syndrome.

The New Zealand Down Syndrome Association acknowledges that people may choose to have prenatal testing for Down syndrome and that a complex interplay of factors influence this choice.

We believe that

- Counselling, support and up-to-date information about Down syndrome should be available to people considering having pre-natal testing.
- Information about the pre-natal testing. The information should include the test's risk, accuracy and waiting time for results.
- Up-to-date information about Down syndrome should be available to the professionals involved in pre-natal testing.
- People should not be pressured into having prenatal testing for Down syndrome.