MINISTRY OF HEALTH
MANATŪ HAUORA

newzealand.govt.nz

Antenatal Screening for Down Syndrome and Other Conditions

Monitoring Report

1 July 2010 to 30 June 2013

National Screening Unit

Contents

List of Tables

List of Figures

Executive summary

This report presents the data for the three financial years from 1 July 2010 to 30 June 2013. It is intended that future reports will be completed on an annual basis. The information in this report is based on screening that occurred from 1 July 2010 to 30 June 2013. Due to lack of data from one of the diagnostic laboratories, the indicators that involve diagnostic data are only reported for 17 DHBs.

Key points for Antenatal Screening for Down Syndrome and Other Conditions

- Antenatal screening for Down syndrome and other conditions provides a risk estimate for Down syndrome (trisomy 21), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome) and some other rare genetic disorders.
- Antenatal screening for Down syndrome and other conditions is optional for pregnant women. Women who are less than 20 weeks pregnant must be advised about the availability of screening and provided with up-to-date information to support the screening discussion, to enable women to make an informed decision about whether to participate.
- First trimester combined screening should be completed between 9 weeks and 13 weeks 6 days gestation. The recommended timing for the blood test is 9 to 10 weeks and for the Nuchal Translucency scan is at 12 weeks.
- Second trimester maternal serum screening should be completed between 14 weeks and 20 weeks gestation. The recommended timing for this test is 14 to 18 weeks.

Key points for July 2010 to June 2013

- National uptake of screening did not reach higher than two thirds of births. The highest rate of completed screens was 66.5% for 2012/13 [indicator 2]
- Screening uptake by Māori and Pacific women was less than half the rate of Other women [indicator 2]
- Trimester one screens made up 87% of all completed screens in 2012/13 [indicator 2]
- Most DHBs showed a trend of increasing rates of screening over the three years covered in this report [indicator 2]
- The positive test rate (number of increased risk results per 100 screens) was 2.8 in the 2012/13 year, down from 3.2 in 2010/11. Positive test rate was higher for second trimester screens (4.7 per 100 screens) than for first trimester screens (2.5 per 100 screens) for the 2012/13 year [indicator 5]
- The false positive rate for trisomy 21, trisomy 18 and trisomy 13 was 3% in 2012/13, which was equal to 2010/11. The rate was higher for second trimester screens (5%) than first trimester screens (2%) [indicator 10]
- The overall detection rate for trisomy 21, trisomy 18 and trisomy 13 was 78% in 2012/13, up from 73% in 2010/11. The detection rate was similar for first and second trimester screens [indicator 11]

Introduction

Background to screening for Down syndrome and other conditions in pregnancy in New Zealand

Antenatal screening for Down syndrome and other conditions has been available to pregnant women in New Zealand since 1968. In October 2007, the government agreed to implement quality improvements to ensure consistency with international best practice. The improvements were introduced in February 2010 and included incorporating maternal serum screening with ultrasound, providing practitioner guidelines and providing consumer resources.

Health practitioners providing maternity care are required to provide women with information about antenatal screening services for Down syndrome and other conditions. There are two screening options:

- first trimester combined screening, which includes a blood test that measures two maternal serum markers, pregnancy-associated protein A (PAPP-A) and free beta- human chorionic gonadotropin (ßhCG). The blood sample is collected between 9 and 13 weeks gestation and combined with an ultrasound scan to determine nuchal translucency (NT) and crown rump length (CRL) measurements between 11-13 weeks, and
- second trimester screening, which is a blood test that measures four maternal serum markers free beta-human chorionic gonadotropin (ßhCG), alpha-fetoprotein (AFP), unconjugated oestriol (uE3) and inhibin A taken between 14 and 20 weeks gestation.

The results of the ultrasound scan and/or serum are combined with other demographic and maternal factors to provide a risk result. For consistency all screening risk results are produced by the screening laboratories. The screening laboratories are LabPLUS at Auckland District Health Board (for samples from Taupo north) and Canterbury Health Laboratories at Canterbury District Health Board (for samples south of Taupo). A shared data repository (PerkinElmer LifeCycle) contains data on all screens. Ultrasound scanning is performed by private and public radiology practices around New Zealand and the ultrasound report is sent to the screening laboratories to include in the risk calculation algorithm.

The conditions covered by screening include:

- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Triploidy
- Neural tube defects
- Unusually high or low levels of the serum analytes

Antenatal screening involves many health professionals including radiology staff, Lead Maternity Carers (LMCs), General Practitioners (GPs) and laboratory personnel. The quality of the information provided by health professionals to the laboratories regarding the pregnancy

details (such as gestation, maternal age, weight, ethnicity and the ultrasound finding) is critical because these details have a significant impact on the risk calculation and report that is issued.

Programme monitoring and data collection

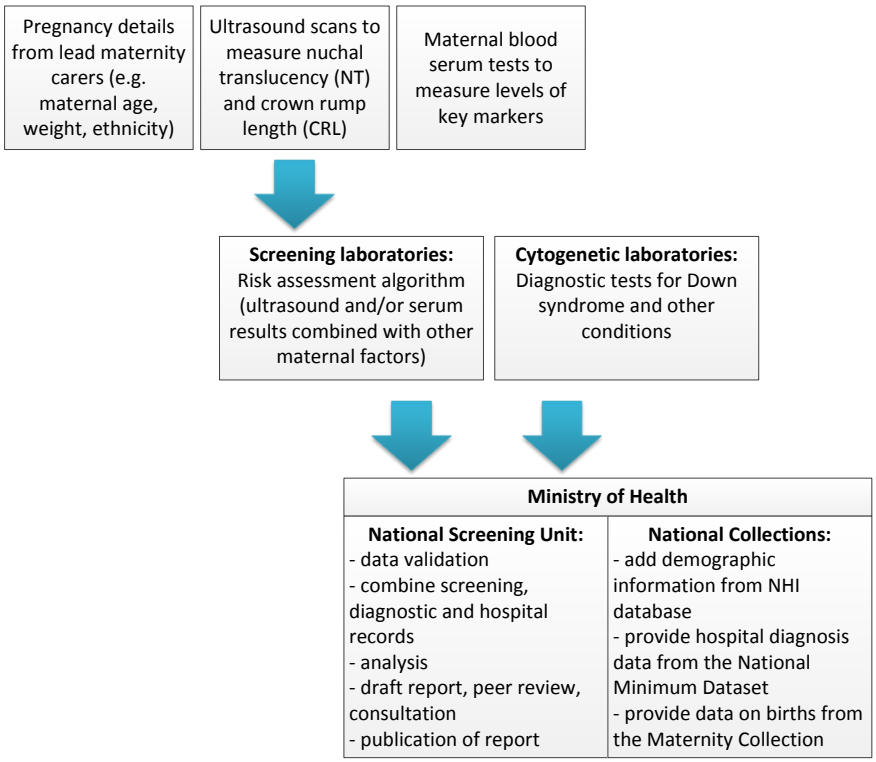
This report presents information on antenatal screening for Down syndrome and other conditions between 1 July 2010 and 30 June 2013. It covers a three year period with the intention that reports will in future be produced annually.

The indicators in this report are taken from the 2014 Antenatal Screening for Down Syndrome and Other Conditions Monitoring and Evaluation Framework. This report covers indicators 2, 5, 6, 7, 8, 9, 10, and 11. Appendix 1 contains definitions for these indicators. Figure 1, below, outlines the data collection process used to produce this report.

The indicators contained within this monitoring report form one part of the evaluation and audit of the quality improvements to antenatal screening for Down syndrome and other conditions. Other activities include:

- Yearly screening laboratory audits by IANZ
- Two yearly peer review of screening laboratories
- Contract monitoring and reporting on a monthly and quarterly basis
- Occasional studies and qualitative information

Figure 1: Data collection process



Information included in this report

The data in this report was sourced from all but one of the laboratories involved in screening and diagnosis of Down syndrome and other conditions from 1 July 2010 to 30 June 2013. Canterbury Health Laboratories screening data is included, however, cytogenetic data from CHL was not provided in time for inclusion in this report. As CHL provides cytogenetic testing for Canterbury, South Canterbury, and West Coast DHBs, women from those DHBs were excluded from the analysis for indicators that required diagnostic data (indicators 6, 7, 8, 9, 10, and 11).

The screening and cytogenetic data was combined with hospital discharge data, sourced from the National Minimum Data Set (NMDS), held by the Ministry of Health. This matching between data from screening laboratories, cytogenetics laboratories, and the NMDS was undertaken to identify the outcome for all screened women.

Definitions

Completed screening
All the required components of each screening test were complete and a risk result was calculated.

Required components of each screening test
First trimester screening comprises analysis of two serum analytes (βhCG, PAPP-A) and a NT measurement. *Second trimester screening* comprises analysis of four serum analytes (βhCG, AFP, uE3 and Inhibin A).

Low risk result
A low risk result is defined as a risk lower than 1:300. So a risk of 1:301 is a low risk.

Increased risk result
An increased risk result is defined as a risk higher than or equal to 1:300. For Indicator 9, positive predictive value, increased risk screening results are further stratified into:

- 1:5 to 1:20
- >1:20 to 1:50; and
- >1:50 to 1:300

Inclusion criteria

Women's screens were included in this analysis if the following criteria were met:

- screening commencement date between 1 July 2010 and 30 June 2013 (i.e. date of the first test the woman had as part of the screening pathway);
- valid National Health Index identifier (NHI);
- known District Health Board (DHB) of domicile;
- age at screen from 12 years to 49 years (calculated using the NHI database date of birth); and
- single screening result per pregnancy (screening laboratories allocate a Case Identification number to each screening episode, where there was more than one screening result for a given identification number the most recent one was used for the analysis).

Data calculations

DHB of domicile

Each woman was allocated to a DHB based on the residential address recorded in the National Health Index (NHI). Where the NHI database did not have a DHB recorded for an NHI, information from the LabPlus database was used to assign the DHB.

Ethnicity and NZ Deprivation decile

Ethnicity data in this report is grouped according to a prioritised system, which is commonly applied across the New Zealand health sector. Prioritisation involves allocating each person to a single ethnic group, based on the ethnicities that person has identified, in the prioritised order of Māori, Pacific, Asian and Other ethnicity. For example, if someone identifies as being New Zealand European and Māori, under the prioritised ethnicity method, they are classified as Māori for the purpose of the analysis. Under this method, the *Other* ethnicity group effectively refers to non-Māori, non-Pacific, non-Asian people. In this report, women identifying as New Zealand European/Pākehā made up approximately 77% of the *Other* ethnicity group. There were no women in the final dataset with ethnicity recorded as *Unknown*.

All reporting by NZ Deprivation decile is based on the 2006 New Zealand Deprivation index decile associated with the residential address held in the NHI database for each woman.

Births

Data on the number of live and still births[1] was obtained from the national Maternity Collection for each financial year. Data used in this report for the 2012/13 year is considered provisional. Appendix 2 contains tables for the denominators used in this report.

Small numbers

In keeping with rules used by Statistics NZ, where an indicator calculation involves small numbers (less than six) then those results have been suppressed as they are considered too unstable.

Prenatal cytogenetic test

The focus of indicators 6, 7, and 8 is on tests that women choose to have as part of managing their pregnancy. For these indicators prenatal tests are defined as chorionic villus sampling or amniocentesis (not products of conception). For indicators 9, 10 and 11 cytogenetic tests on products of conception are used in addition to CVS, amniocentesis and infant diagnoses to determine the outcome of the pregnancy.

Repeat screens

A repeat screen was defined as a second screening result recorded against a given Case Identification number. Where this occurred, only the most recent screening result was retained for the analysis.

Linking rules

When matching screening and diagnosis data the following rules were followed:

- for a birth to link to a commenced screen the screen date must be earlier than the birth date and the date difference must not be greater than 230 days (approximately 33 weeks); and

[1] births reaching at least 20 weeks gestation or >=400g birth weight

- for a prenatal cytogenetic test to link to a screen the cytogenetic sample date must be later than the screen date, but not more than 105 days (15 weeks) later

Data limitations

Provisional Maternity Collection data for 2012/13
As stated above, the births data for the 2012/13 year that was used for this report is considered provisional. Incomplete births data could have affected denominator calculations (see denominator underestimation) and also the linkage of infant diagnoses back to screened women as the Maternity Collection data was used for this.

Denominator underestimation
Screening completion rates derived using total births may overestimate the proportion of women participating in antenatal screening for Down syndrome and other conditions. This is because the true denominator (i.e. all pregnant women that reach 9 weeks gestation) is likely to be larger than the denominator used (i.e. all births reaching at least 20 weeks gestation or at least 400g birth weight).

Missing data
Missing or incorrect data for any screened woman will affect indicator calculations. Known data issues in this report relate to the following:

- Some women may have incomplete data if they were screened outside of Canterbury, South Canterbury and West Coast DHBs but had a cytogenetic test through Canterbury Health Laboratories. Given known laboratory catchment areas it is unlikely that this has occurred in enough cases to be significant.
- Ten records did not have DHB of domicile information recorded in either the NHI database or in the laboratory information system. These records were excluded from the analysis.

Inconsistent data

In some instances there was variation between the demographic information held in the NHI database and that held by LabPlus. The NHI database was used as the definitive source which led to instances where the age at screen calculated using the NHI date of birth was outside the range of 12 to 49 years (16 records less than 12 years, 20 records 50 years old or greater) and an instance where date of death as recorded in the NHI database was prior to the date of screen (1 record). The Ministry will work with laboratories to implement a process to resolve these variances for future reports. For this report, records where the age at screen was younger than 12 or older than 49 have been excluded.

Final dataset

Table 1 summarises the records received and excluded from the screening dataset. The final dataset of 119,120 includes screening records for women from Canterbury, South Canterbury and West Coast DHBs. Records for these women are included in the results for indicators 2 and 5 but excluded from indicators 6 to 11.

Table 1: Screening dataset cleansing

	Number	Percentage
Total screening records received	122,162	100.00%
Final screening dataset for analysis	119,120	97.51%
Total excluded records	3,042	2.49%
Private/overseas screens	2,764	2.26%
Invalid NHI	34	0.03%
Unknown DHB	10	0.01%
Date of death prior to screen	1	0.00%
Age at screen < 12	16	0.01%
Age at screen > 49	31	0.03%
Repeat screen	186	0.15%

Indicator 2: Screens completed

This indicator looks at screens completed by trimester of screening (first or second), reported by DHB, age, ethnicity, and NZ deprivation index.

Total screens completed by trimester

During the 2012/13 year 40,215 screens were completed in total, a rate of 66.5 per 100 births. Table 2 shows the total number of screens completed by financial year and trimester of the screen. T1 refers to the first trimester and T2 the second trimester. The vast majority of screens are T1 screens. The total number of completed screens increased from 2010/11 to 2011/12 but stayed relatively stable between 2011/12 and 2012/13. The trend for screens per 100 births was similar, with an increase of nearly 5 between the first two financial years before the rate levels off.

Table 2: Total screens completed by trimester, July 2010 to June 2013

	Number and rate of screens completed		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**
T1 screen	34,036	35,692	35,121
T2 screen	4,416	4,761	5,094
Total screens	**38,452**	**40,453**	**40,215**
Screens per 100 births	61.1	65.9	66.5

Screens completed by DHB

Screening completion rates for 2012/13 varied across DHBs from 82 per 100 births in Nelson Marlborough to 42 per 100 births in Whanganui (see figure 2). Table 3 gives a full breakdown by trimester of the screen.

Figure 2: Screens completed by DHB, 1 July 2012 to 30 June 2013

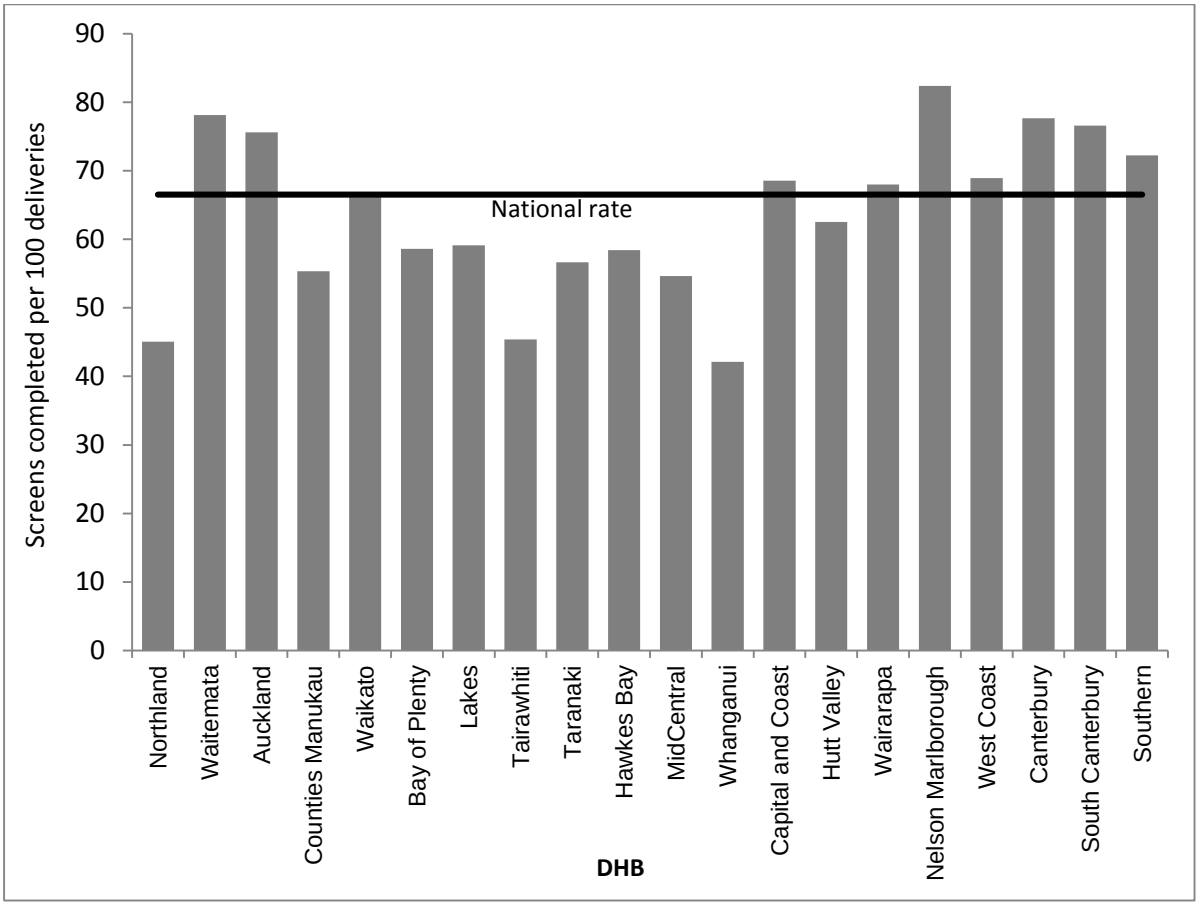


Table 3: Screening completion by DHB, 1 July 2012 to 30 June 2013

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	863	136	999	38.9	6.1	45.1
Waitemata	5,477	682	6,159	69.5	8.6	78.1
Auckland	4,207	664	4,871	65.3	10.3	75.6
Counties Manukau	3,730	1,023	4,753	43.4	11.9	55.4
Waikato	3,105	393	3,498	58.9	7.5	66.4
Bay of Plenty	1,543	144	1,687	53.6	5.0	58.6
Lakes	801	89	890	53.2	5.9	59.1
Tairawhiti	265	55	320	37.6	7.8	45.4
Taranaki	725	131	856	48.0	8.7	56.7
Hawke's Bay	1,115	154	1,269	51.3	7.1	58.4
MidCentral	994	142	1,136	47.8	6.8	54.6
Whanganui	280	73	353	33.4	8.7	42.1
Capital and Coast	2,356	280	2,636	61.3	7.3	68.6
Hutt Valley	1,039	172	1,211	53.6	8.9	62.5
Wairarapa	297	30	327	61.7	6.2	68.0
Nelson Marlborough	1,159	107	1,266	75.4	7.0	82.4
West Coast	256	26	282	62.6	6.4	68.9
Canterbury	4,127	498	4,625	69.3	8.4	77.7
South Canterbury	437	77	514	65.1	11.5	76.6
Southern	2,345	218	2,563	66.1	6.1	72.3
Total	**35,121**	**5,094**	**40,215**	**58.1**	**8.4**	**66.5**

Most DHBs showed a trend of increasing rates of screening over the three years covered in this report. Exceptions to this include Bay of Plenty, Capital and Coast and South Canterbury DHBs who each showed an increase from 2010/11 to 2011/12 but then a decrease for 2012/13 (see figure 3 and table 4).

Figure 3: Screens completed by DHB, 1 July 2010 to 30 June 2013

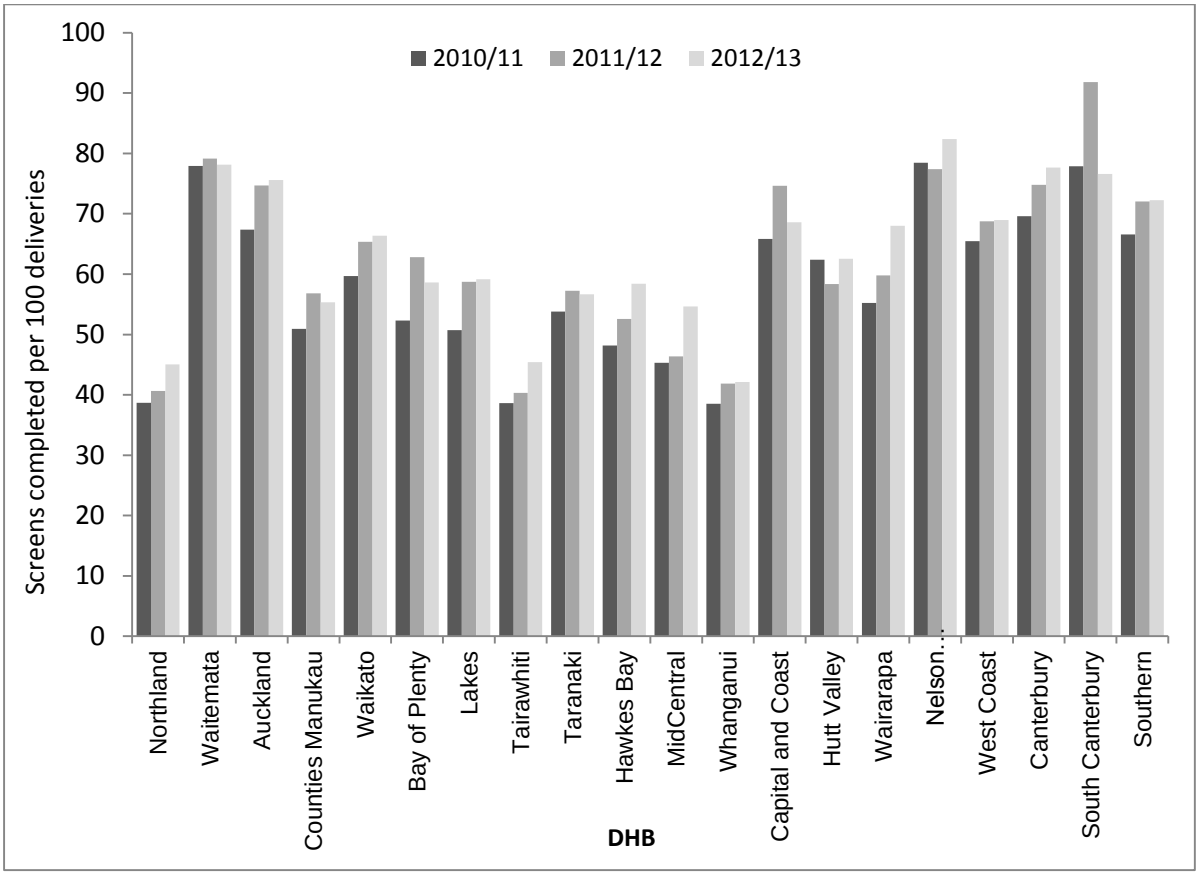


Table 4: Screening completion by DHB, 1 July 2010 to 30 June 2013

	Screens completed (per 100 births)		
DHB	**2010/11**	**2011/12**	**2012/13**
Northland	38.7	40.6	45.1
Waitemata	77.9	79.1	78.1
Auckland	67.4	74.7	75.6
Counties Manukau	50.9	56.8	55.4
Waikato	59.7	65.3	66.4
Bay of Plenty	52.3	62.8	58.6
Lakes	50.7	58.7	59.1
Tairawhiti	38.6	40.3	45.4
Taranaki	53.8	57.2	56.7
Hawke's Bay	48.2	52.6	58.4
MidCentral	45.3	46.4	54.6
Whanganui	38.5	41.9	42.1
Capital and Coast	65.8	74.6	68.6
Hutt Valley	62.4	58.4	62.5
Wairarapa	55.2	59.8	68.0
Nelson Marlborough	78.4	77.4	82.4
West Coast	65.5	68.8	68.9
Canterbury	69.6	74.8	77.7
South Canterbury	77.9	91.8	76.6
Southern	66.5	72.0	72.3
Total	**61.1**	**65.9**	**66.5**

Screens completed by age, ethnicity and NZ deprivation index

Table 5 provides an overall view of screens completed by age of mother at time of screen, ethnicity and NZ deprivation index for 1 July 2010 to 30 June 2013.

Screening completion rates were highest in the 30-34 years age group with 74 women completing screening per 100 births in 2012/13. This was followed closely by the 35 – 39 years age group with 73 per 100 births (see figure 5).

Screening completion rates were highest among women of Other ethnicity at 85 per 100 births for 2012/13. This was followed by Asian women at 80. The rate of completed screens for Pacific and Māori women was much lower at 39 per 100 births and 33 per 100 births respectively (see figure 6).

Screening completion rates were highest among women in less deprived areas with 77 women completing screening per 100 births for deciles 1-2 in 2012/13 and 49 women screened per 100 births for deciles 9-10 (see figure 7).

Table 5: Screens completed by age of mother, ethnicity and NZ deprivation index, 1 July 2010 to 30 June 2013

	Number of screens completed			Screens completed (per 100 births)		
	2010/11	2011/12	2012/13	2010/11	2011/12	2012/13
Age at screen						
Under 20 years	1,729	1,768	1,649	40.1	43.8	46.0
20 – 24 years	5,466	5,824	5,954	45.9	50.8	53.0
25 – 29 years	9,849	10,880	10,907	62.8	70.0	69.5
30 – 34 years	12,192	12,811	12,631	70.0	74.1	73.6
35 – 39 years	7,709	7,490	7,435	70.0	71.8	73.1
40 – 44 years	1,449	1,614	1,565	58.3	65.7	63.2
45 years and over	58	66	74	53.7	47.8	59.7
Ethnicity						
Māori	4,301	4,791	4,875	26.8	30.9	32.8
Pacific	2,327	2,584	2,543	32.3	37.4	38.6
Asian	5,588	6,810	6,821	78.5	90.5	79.8
Other	26,236	26,268	25,976	80.6	83.6	85.3
NZ Deprivation Index						
Decile 1 - 2	7,480	7,381	6,906	83.7	83.5	77.4
Decile 3 – 4	7,215	7,453	7,522	70.4	75.6	75.2
Decile 5 – 6	7,960	8,350	8,363	67.2	72.5	73.1
Decile 7 – 8	8,613	9,241	9,339	60.3	65.6	67.9
Decile 9 – 10	7,109	7,947	8,024	40.4	46.6	49.2
Total	**38,452**	**40,453**	**40,215**	**61.1**	**65.9**	**66.5**

Figure 4: Screens completed by age of mother at screen, 1 July 2012 to 30 June 2013

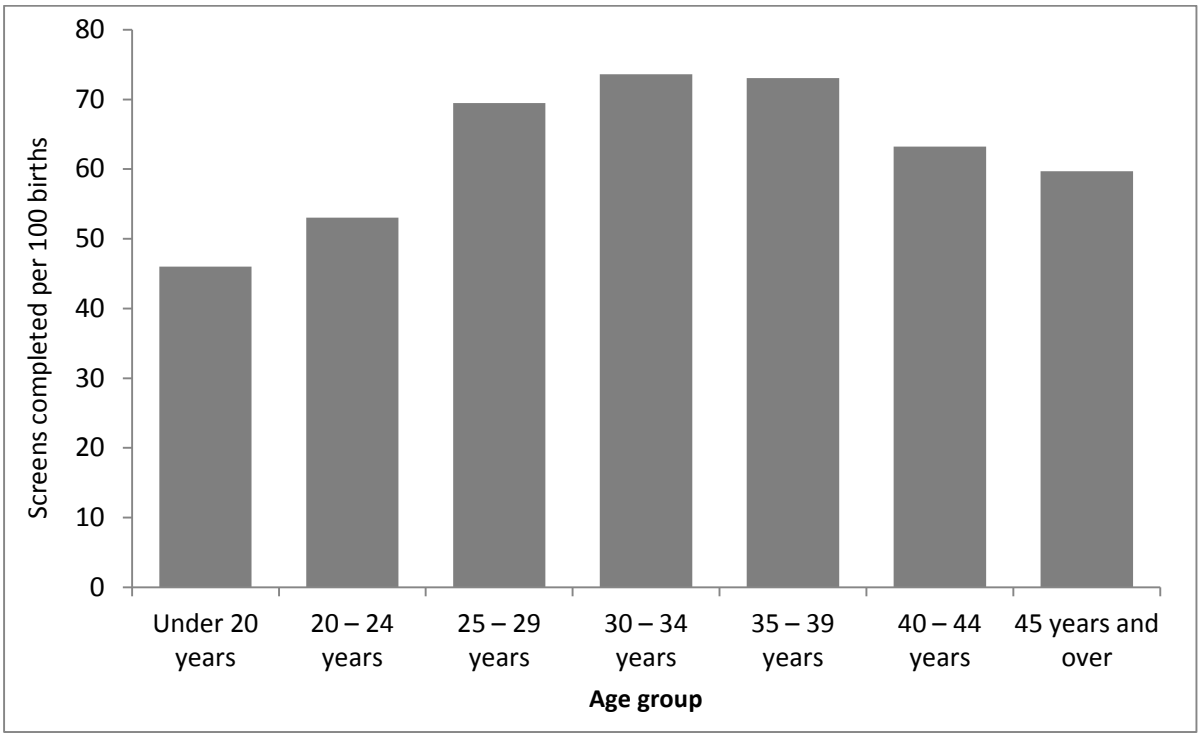


Figure5: Screens completed by ethnicity of mother, 1 July 2012 to 30 June 2013

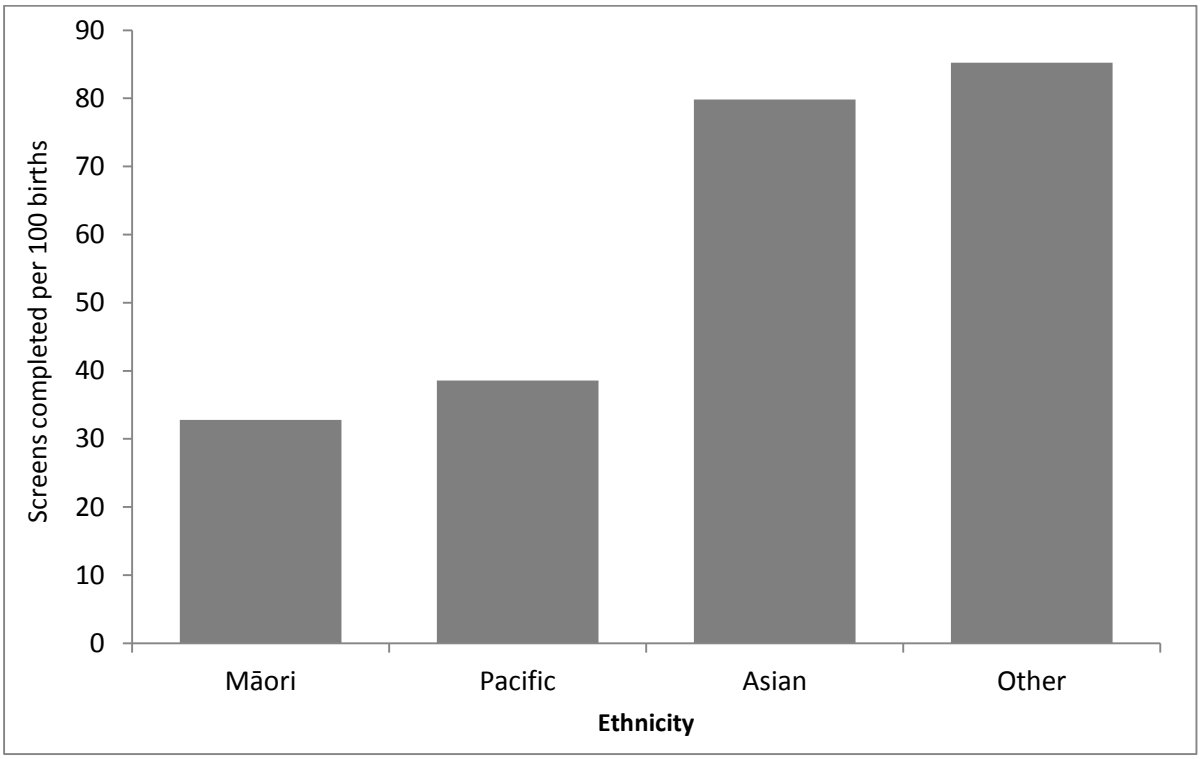
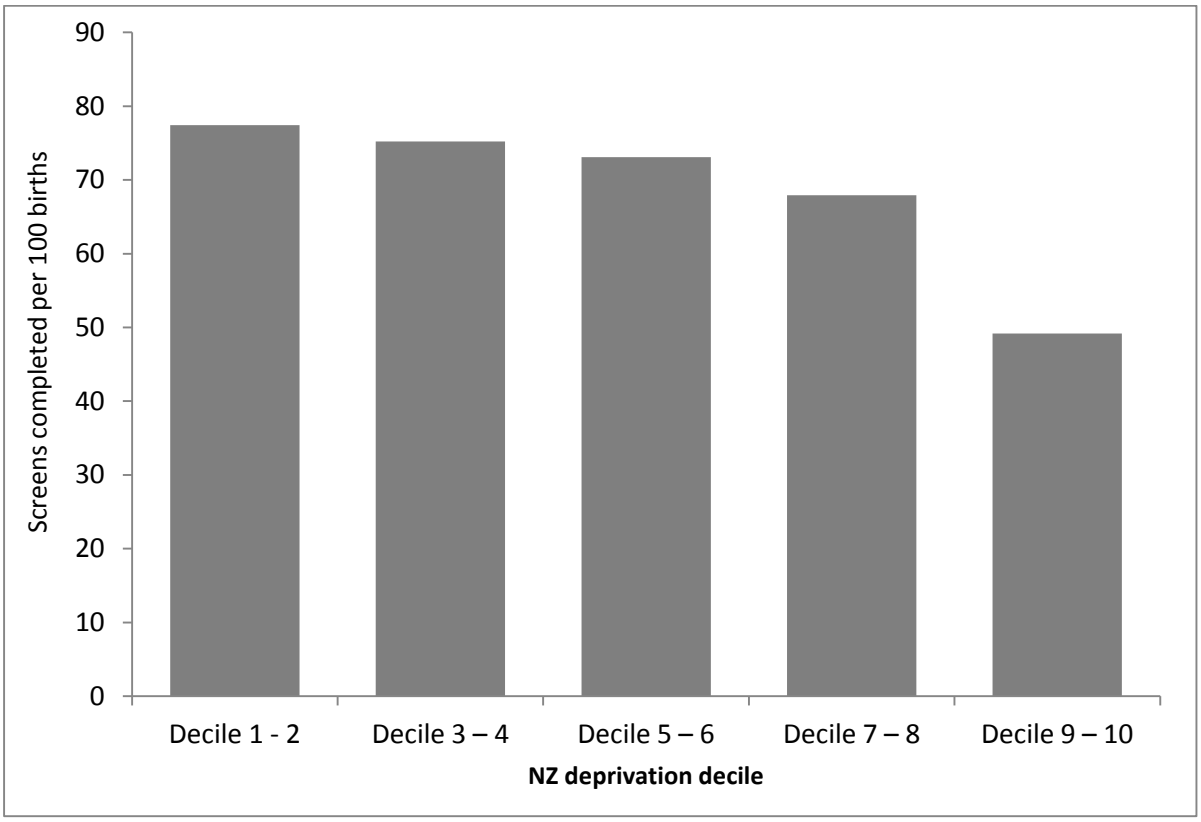


Figure 6: Screens completed by NZ deprivation decile of mother, 1 July 2012 to 30 June 2013



Indicator 5: Increased risk screening results for trisomy 21, trisomy 18 and trisomy 13

This section reports on the screening risk results issued for trisomy 21, trisomy 18 and trisomy 13. Women who complete screening receive a risk result, either low risk or increased risk for each trisomy. The results may show an increased risk for more than one of the three trisomies, in which case a woman's results may count in the numerator for more than one of the trisomy calculations presented here.

Total increased risk screening results for trisomy 21, 18 or 13

The table below shows total number of screening risk results that were classified as increased risk for one or more of trisomy 21, 18 or 13 by financial year, together with the number of increased risk results per 100 screens (positive test rate). In 2012/13, for every 100 screens completed 2.8 increased risk results were issued. This is a lower rate than 2010/11 but is slightly higher than 2011/12.

Table 6: Number and rate per 100 screens of increased risk screening results for trisomy 21, 18 or 13, 1 July 2010 to 30 June 2013

	Number and rate of increased risk screens		
	2010/11	2011/12	2012/13
Total increased risk results	1,247	1,099	1,126
Positive test rate per 100 screens	3.2	2.7	2.8

Increased risk screening results for trisomy 21, 18 or 13 by age, ethnicity and deprivation

The table below shows the number and proportion of screening risk results that were classified as increased risk for any one or more of trisomy 21, 18, or 13 by age at screen, ethnicity and deprivation for the 2012/13 year. Positive test rate increases markedly with increasing age and is also higher for Pacific women compared to other ethnicities. The prior risk (age) is included in the calculation. Older women are more likely to have a positive test and are also more likely to have a higher detection rate. Different levels of deprivation do not appear to affect the positive test rate.

Table 7: Increased risk screening results for trisomy 21, 18 or 13 by age, ethnicity and deprivation, 1 July 2012 to 30 June 2013

	Number of screens that include an increased risk for trisomy 21, 18 or 13	Total number of screens	Positive test rate per 100 screens
Age at screen			
Under 20 years	7	1,649	0.4
20 – 24 years	62	5,954	1.0
25 – 29 years	106	10,907	1.0
30 – 34 years	241	12,631	1.9
35 – 39 years	387	7,435	5.2
40 – 44 years	292	1,565	18.7
45 years and over	31	74	41.9
Ethnicity			
Māori	133	4,875	2.7
Pacific	114	2,543	4.5
Asian	212	6,821	3.1
Other	667	25,976	2.6
NZ Deprivation Index			
Decile 1 - 2	215	6,906	3.1
Decile 3 – 4	211	7,522	2.8
Decile 5 – 6	211	8,363	2.5
Decile 7 – 8	260	9,339	2.8
Decile 9 – 10	226	8,024	2.8
Unknown decile	3	-	-

Increased risk screening results for trisomy 21, 18 or 13 by trimester of screen

Table 8, over page, shows the positive test rate for each of trisomy 21, 18 and 13 as well as the positive test rate for the three trisomies together by trimester of screen and financial year. The low number of increased risk results for trisomy 18 and 13 correspond with low positive test rates (from 0.36 per 100 screens) while the positive test rate for trisomy 21 was close to 3 for all years (2.72 to 3.24). The second trimester positive test rate for trisomy 21 was significantly higher than the first trimester positive test rate in 2012/12 (and in 2011/12 it was twice as high). This may be due to variability in nuchal translucency scanning accuracy.

Table 8: Increased risk screening results for trisomy 21, 18 and 13 by trimester of screen, 1 July 2010 to 30 June 2013

Year	Total results that include an increased risk for specified trisomy	Positive test rate per 100 screens	T1 results that include an increased risk for specified trisomy	Positive test rate per 100 T1 screens	T2 results that include an increased risk for specified trisomy	Positive test rate per 100 T2 screens
Trisomy 21						
2010/11	1,222	3.2	980	2.9	242	5.5
2011/12	1,090	2.7	822	2.3	268	5.6
2012/13	1,110	2.8	870	2.5	240	4.7
Trisomy 18						
2010/11	150	0.4	132	0.4	18	0.4
2011/12	149	0.4	141	0.4	8	0.2
2012/13	143	0.4	125	0.4	18	0.4
Trisomy 13						
2010/11	158	0.4	151	0.4	7	0.2
2011/12	157	0.4	150	0.4	7	0.1
2012/13	151	0.4	142	0.4	9	0.2
Any one or more of trisomy 21, 18 or 13						
2010/11	1,247	3.2	992	2.9	255	5.8
2011/12	1,099	2.7	826	2.3	273	5.7
2012/13	1,126	2.8	873	2.5	253	5.0

Increased risk screening results stratified by risk level, 1 July 2012 to 30 June 2013

The table below shows the number of screening risk results stratified by risk level for each of trisomy 21, 18 and 13 for the 2012/13 year. A woman's screen result may indicate an increased risk for more than one of trisomy 21, 18 and 13 so the sum of the values in this table will be greater than the total number of increased risk results.

Table 9: Increased risk screening results for trisomy 21, 18 and 13 by risk level, 1 July 2012 to 30 June 2013

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 – 1:20	242	56	60
>1:20 to 1:50	138	24	28
>1:50 to 1:300	730	63	63

Indicator 6: Diagnostic testing volumes for women with increased risk screens

This indicator reports information on the number and proportion of women who complete diagnostic testing following an increased risk screening result for trisomy 21, trisomy 18 or trisomy 13. Following an increased risk result, women may choose to have diagnostic testing (either amniocentesis or chorionic villus sampling) to determine the absence or the presence of the condition.

Results for this indicator, and all remaining indicators, exclude screened women from Canterbury, South Canterbury and West Coast DHBs due to unavailability of diagnostic data.

Diagnostic testing volumes for women with increased risk screens by trimester of screen

Table 10 shows the diagnostic testing rate for 2012/13 by trimester of screen. During the 2012/13 year, for every 100 women that received an increased risk result after a first trimester screen, 66 women had a diagnostic test. The rate was lower for women who received an increased risk after a second trimester screen (48.5 women per 100 increased risk screens). See appendix 3 for a summary of diagnostic test results for women who had increased risk screens in 2012/13.

Table 10: Diagnostic testing volumes for women with increased risk screens by trimester of screen, 1 July 2012 to 30 June 2013

	Diagnostic tests per 100 increased risk screens		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**
T1 screen	63.9	64.4	65.9
T2 screen	42.6	40.6	48.5
Total screens	**59.4**	**58.3**	**61.9**

Diagnostic testing volumes for women with increased risk screens by DHB

The rate of diagnostic testing for women with increased risk screens in 2012/13 varied across DHBs from 46.3 per 100 increased risk results in Northland to 100 in Wairarapa (noting that the Wairarapa rate is based on only 9 diagnostic tests). Auckland was the next highest with 74.4 (see table 11).

Table 11: Diagnostic testing volumes for women with increased risk screens by DHB, 1 July 2010 to 30 June 2013

	Number of diagnostic tests			Tests per 100 increased risk screens#		
DHB	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
Northland	20	13	19	43.5	41.9	46.3
Waitemata	156	125	129	69.3	66.5	67.2
Auckland	127	121	116	71.8	66.9	74.4
Counties Manukau	73	70	72	51.4	48.3	51.1
Waikato	15	24	33	18.8	32.9	48.5
Bay of Plenty	15	19	16	62.5	54.3	57.1
Lakes	13	17	24	48.1	54.8	77.4
Tairawhiti	5	7	1	-	63.6	-
Taranaki	17	12	21	63.0	60.0	75.0
Hawke's Bay	28	20	22	65.1	57.1	55.0
MidCentral	27	17	18	51.9	65.4	58.1
Whanganui	1	6	3	-	54.5	-
Capital and Coast	66	55	58	76.7	67.1	69.9
Hutt Valley	17	19	20	54.8	67.9	62.5
Wairarapa	6	4	9	75.0	80.0	100.0
Nelson Marlborough	21	15	11	72.4	55.6	55.0
West Coast	n/a	n/a	n/a	n/a	n/a	n/a
Canterbury	n/a	n/a	n/a	n/a	n/a	n/a
South Canterbury	n/a	n/a	n/a	n/a	n/a	n/a
Southern	33	31	37	52.4	53.4	56.9
Total	**640**	**575**	**609**	**59.4**	**58.3**	**61.9**

rate suppressed if the number of diagnostic tests was <6

Diagnostic testing volumes for women with increased risk screens by age, ethnicity and deprivation

Table 12 shows the diagnostic testing rate for women with increased risk screens by age at screen, ethnicity and NZ deprivation index for 2010/11 to 2012/13. The diagnostic testing rate ranged from 57.9 per 100 increased risk screens to 67.1 per 100 for age groups from 20 to 44 years. The rates for women under 20 years and women 45 years and over were lower but based on low numbers. During the 2012/13 year 7 women under 20 received increased risk results and 3 had a diagnostic test. Diagnostic testing rates were higher for women of Asian ethnicity and for women from less deprived areas.

Table 12: Diagnostic testing volumes for women with increased risk screening results by age at screen, ethnicity and deprivation, 1 July 2010 to 30 June 2013

	Diagnostic tests per 100 increased risk screens#		
	2010/11	2011/12	2012/13
Age at screen			
Under 20 years	-	42.9	-
20 – 24 years	64.0	47.1	61.4
25 – 29 years	59.3	59.6	62.1
30 – 34 years	61.3	67.7	67.1
35 – 39 years	61.7	61.2	63.3
40 – 44 years	53.5	49.4	57.9
45 years and over	61.1	45.0	46.2
Ethnicity			
Māori	41.9	37.7	50.8
Pacific	34.5	33.1	39.4
Asian	73.2	68.4	73.8
Other	63.2	63.8	64.6
NZ Deprivation Index			
Decile 1 - 2	68.9	68.1	69.3
Decile 3 – 4	68.4	65.3	72.8
Decile 5 – 6	64.1	61.1	60.6
Decile 7 – 8	54.7	55.0	59.9
Decile 9 – 10	40.7	45.0	50.5

rate suppressed if the number of diagnostic tests was <6

Diagnostic testing volumes for women with increased risk screening results stratified by risk level, 1 July 2012 to 30 June 2013

The table below shows the number of diagnostic tests for women with increased risk screening results for one or more of trisomy 21, 18 or 13, stratified by risk level, for the 2012/13 year. A screening result includes a separate risk level for each of the three trisomies. Women were assigned a risk level based on the highest risk across the three trisomies. As diagnostic data was not available for women from Canterbury, South Canterbury and West Coast, screening volumes for women from these three DHBs are not included for this indicator. Subsequently, the increased risk screen values do not match with indicator 5.

Table 13: Diagnostic testing volumes for women with increased risk screens by risk level, 1 July 2012 to 30 June 2013

Risk level	Number of diagnostic tests	Number of increased risk screens	Tests per 100 increased risk screens
1:5 – 1:20	139	214	65.0
>1:20 to 1:50	87	121	71.9
>1:50 to 1:300	383	650	58.9

Indicator 7: Diagnostic testing volumes for women who receive a low risk screening result

This section reports information on the number and proportion of women who complete diagnostic testing following a low risk screening result. Following a low risk screen, women may still choose to have diagnostic testing to determine the absence or the presence of a condition. This indicator intends to capture only those that had a low risk in isolation so for this calculation a woman was only counted as having a low risk screen if there was no increased risk for any of the other conditions covered by the screening test in addition to trisomy 21, 18 and 13. So for example, if the result was low risk for trisomy 21, 18 and 13 but increased risk for neural tube defects then the woman was categorised as at increased risk for this indicator. Some women with low risk screening results may have other indications for diagnostic testing, e.g. family history of another condition that diagnostic testing can identify. Information on the indication for diagnostic testing is not reliably provided on laboratory forms so the calculations for this indicator cannot exclude these women.

Diagnostic testing volumes for women with low risk screens by trimester of screen

The national rate of diagnostic testing for women that received low risk screening results was 0.83 per 100 low risk screens in 2012/13. This was a decrease from the previous two years. This means that, for each of the years covered in this report, just under 1% of the women that received a low risk result proceeded on to a prenatal diagnostic test.

Table 14: Diagnostic testing volumes for women with low risk screens by trimester of screen, 1 July 2012 to 30 June 2013

	Diagnostic tests per 100 low risk screens		
Year	**T1 screen**	**T2 screen**	**Total screens**
2010/11	0.91	0.61	0.88
2011/12	0.91	0.66	0.88
2012/13	0.88	0.43	0.83

Diagnostic testing volumes for women with low risk screens by DHB

The rate of diagnostic testing for women with low risk screens during 2012/13 varied across DHBs from 0.5 per 100 in Counties Manukau, Waikato and Hawke's Bay to 1.4 per 100 in Auckland (see table 15).

Table 15: Total diagnostic testing volumes for women with low risk screens by DHB 2010/11 to 2012/13

	Number of diagnostic tests			Tests per 100 low risk screens#		
DHB	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
Northland	7	2	4	0.8	-	-
Waitemata	65	55	63	1.1	0.9	1.1
Auckland	60	83	68	1.4	1.7	1.4
Counties Manukau	27	28	21	0.6	0.6	0.5
Waikato	6	10	16	0.2	0.3	0.5
Bay of Plenty	5	9	10	-	0.5	0.6
Lakes	2	4	3	-	-	-
Tairawhiti	0	1	1	-	-	-
Taranaki	8	10	10	1.0	1.1	1.2
Hawke's Bay	12	9	6	1.1	0.8	0.5
Mid Central	9	5	7	0.9	-	0.6
Whanganui	2	5	1	-	-	-
Capital and Coast	27	19	25	1.1	0.7	1.0
Hutt Valley	11	13	9	0.9	1.1	0.8
Wairarapa	2	0	0	-	-	-
Nelson Marlborough	12	13	12	1.0	1.1	1.0
West Coast	n/a	n/a	n/a	n/a	n/a	n/a
Canterbury	n/a	n/a	n/a	n/a	n/a	n/a
South Canterbury	n/a	n/a	n/a	n/a	n/a	n/a
Southern	27	34	23	1.1	1.3	0.9
Total	**282**	**300**	**279**	**0.88**	**0.88**	**0.83**

rate suppressed if the number of diagnostic tests was <6

Diagnostic testing volumes for women with low risk screening results by age, ethnicity and NZ deprivation index

Table 16 shows the rate of diagnostic testing for women with low risk screening results by age, ethnicity and NZ deprivation index. The rate of diagnostic testing increased with increasing age. Asian and Other women were far more likely to have a diagnostic test following a low risk screen compared with Māori and Pacific women. Women from NZ deprivation deciles 1 and 2 were more likely to have a diagnostic test than women from deciles 9 and 10.

Table 16: Diagnostic tests per 100 low risk screens by age, ethnicity and NZ deprivation index 2010/11 to 2012/13

	Diagnostic tests per 100 low risk screens		
	2010/11	2011/12	2012/13
Age			
Under 20 years	0.20	0.52	0.28
20 – 24 years	0.28	0.24	0.33
25 – 29 years	0.24	0.45	0.33
30 – 34 years	0.60	0.50	0.71
35 – 39 years	2.04	1.82	1.31
40 – 44 years	5.28	5.66	6.10
45 years and over	9.38	8.11	8.11
Ethnicity			
Māori	0.31	0.60	0.46
Pacific	0.29	0.26	0.39
Asian	0.81	0.84	0.83
Other	1.05	1.01	0.95
NZ Deprivation Index			
Decile 1 - 2	1.54	1.32	1.43
Decile 3 – 4	1.24	1.10	0.93
Decile 5 – 6	0.74	0.81	0.78
Decile 7 – 8	0.63	0.80	0.76
Decile 9 – 10	0.30	0.47	0.35

Indicator 8: Diagnostic testing volumes for unscreened women

This section reports information on the number of women who complete diagnostic testing but who have not been screened in the 105 days prior to the diagnostic test. Reporting is limited to 2011/12 and 2012/13 because of the need to have screening data for the period prior to assessing whether unscreened.

Measuring the distribution of unscreened women having diagnostic testing by DHB, age ethnicity and NZ deprivation decile can highlight areas where there are barriers to access or other issues with antenatal screening for some population groups.

Diagnostic testing volumes for unscreened women

During the 2011/12 year 251 diagnostic tests were completed for unscreened women. In 2012/13 this figure was 208. Table 17 shows the number of tests by DHB and table 18 shows the breakdown by age, ethnicity and NZ deprivation index. Due to the low numbers involved rates per 100 births are not shown.

Table 17: Diagnostic testing volumes for unscreened women by DHB 1 July 2011 to 30 June 2013

	Number of diagnostic tests	
DHB	**2011/12**	**2012/13**
Northland	14	3
Waitemata	35	31
Auckland	48	29
Counties Manukau	33	18
Waikato	14	19
Bay of Plenty	13	10
Lakes	2	7
Tairawhiti	2	4
Taranaki	12	17
Hawke's Bay	10	10
Mid Central	12	10
Whanganui	4	4
Capital and Coast	19	18
Hutt Valley	6	11
Wairarapa	6	2
Nelson Marlborough	4	6
West Coast	n/a	n/a
Canterbury	n/a	n/a
South Canterbury	n/a	n/a
Southern	17	9
Total	**251**	**208**

Table 18: Total diagnostic testing volumes for unscreened women by age, ethnicity and deprivation, 1 July 2011 to 30 June 2013

	Number of diagnostic tests	
	2011/12	2012/13
Age		
Under 20 years	14	9
20 – 24 years	33	26
25 – 29 years	40	36
30 – 34 years	63	46
35 – 39 years	58	48
40 – 44 years	39	39
45 years and over	4	4
Ethnicity		
Māori	39	33
Pacific	23	13
Asian	46	34
Other	143	128
NZ Deprivation Index		
Decile 1 - 2	57	39
Decile 3 – 4	33	38
Decile 5 – 6	51	40
Decile 7 – 8	52	54
Decile 9 – 10	58	37

Diagnostic testing results for unscreened women

The results of prenatal diagnostic testing (CVS or amniocentesis) for unscreened women are summarised in the table below. Of the 208 diagnostic tests in 2012/13 for unscreened women, 162 (78%) had a normal karyotype. There were 13 trisomy 21 diagnoses, 11 trisomy 18 diagnoses and 1 diagnosis of trisomy 13. The remaining 21 'other' results included 2 failed tests, 4 diagnoses of triploidy, and 3 diagnoses of Turner syndrome.

Table 19: Total diagnostic testing results for unscreened women, 1 July 2012 to 30 June 2013

Karyotype result	Number	Percentage
Normal karyotype	162	77.9%
Confirmed trisomy 21	13	6.3%
Confirmed trisomy 18	11	5.3%
Confirmed trisomy 13	1	0.5%
Other result	21	10.1%
Total	**208**	**100%**

Indicator 9: Diagnostic testing outcomes for women with increased risk screening results

This section reports information on the positive predictive value of screening. Positive predictive value (PPV) is calculated by dividing the number of true positives (increased risk screening result and then a positive diagnostic test for trisomy, or a baby born with trisomy) by the number of true positive and false positives (increased risk screening result and then a negative diagnostic test for a trisomy, or a baby born without a trisomy). Appendix 4 contains a summary of how screening measures, such as PPV, are calculated.

Positive predictive value of screening 1 July 2010 to 30 June 2013

The combined PPV for trisomy 21, 18 or 13 was calculated by categorising any screening result that included an increased risk for any of trisomy 21, 18 or 13 as a positive screen. If there was a subsequent diagnosis of any of trisomy 21, 18 or 13 then it was classified as a true positive. If there was no diagnosis of any of these three trisomies it was a false positive.

It should be noted that there were a small number of screens where the trisomy with the increased risk screening result was not the trisomy that was ultimately diagnosed. For example, a screening result may have shown an increased risk for trisomy 21 and normal risk for trisomy 13 but the cytogenetic result or infant diagnosis was trisomy 13. For the indicator 9, 10 and 11 calculations that combine the three trisomies together this record was categorised as a true positive. For the calculations looking at trisomy 21 specifically it was a false positive and for the trisomy 13 calculations it was a false negative. Due to this conflict in categorisation, the breakdowns by screening risk level, age, ethnicity, and deprivation have only been reported for trisomy 21 rather than combining trisomy 21, 18 and 13.

The overall PPV for 2012/13 was 0.11. This means that if a woman receives an increased risk result for trisomy 21, 18 or 13 there is an 11% probability that she is carrying a fetus with trisomy 21, 18 or 13. The PPV for all years is higher for first trimester screens than second trimester screens.

Table 20: Positive predictive value of screening for trisomy 21, 18 or 13, 1 July 2010 to 30 June 2013

	Positive diagnostic tests or infant diagnoses for women screened as increased risk (True Positives)			Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	97	117	98	751	616	659	0.11	0.16	0.13
T2 screens	7	10	11	223	244	216	0.03	0.04	0.05
Total screens	104	127	109	974	860	875	0.10	0.13	0.11

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21 alone (see table 21), the PPV for 2012/13 was lower than the overall PPV at 0.08. This means that if a woman receives an increased risk result for trisomy 21 there is an 8% probability that she is carrying a fetus with trisomy 21. Once again, the PPV for all years is higher for first trimester screens than second trimester screens.

Table 21: Positive predictive of screening for trisomy 21, 1 July 2010 to 30 June 2013

	Positive diagnostic test or infant diagnosis for women screened as increased risk (True Positives)			Negative diagnostic test or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	73	74	72	766	656	684	0.09	0.10	0.10
T2 screens	5	9	8	214	240	206	0.02	0.04	0.04
Total screens	78	83	80	980	896	890	0.07	0.08	0.08

Trisomies 13 and 18 involve small numbers and have similar risk profiles so combined results for PPV, false positive rate, and detection rate have been calculated for these trisomies.

The combined PPV for trisomies 13 or 18 for 2012/13 is higher than the trisomy 21 PPV at 0.18 (see table 22). However the number of positive diagnoses for these two trisomies is low so caution should be taken when interpreting these rates.

Table 22: Positive predictive of screening for trisomy 13 or 18, 1 July 2010 to 30 June 2013

	Positive diagnostic test or infant diagnosis for women screened as increased risk (True Positives)			Negative diagnostic test or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value#		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	23	38	23	125	106	97	0.16	0.26	0.19
T2 screens	1	0	2	17	14	19	-	-	-
Total screens	24	38	25	142	120	116	0.14	0.24	0.18

rate suppressed if the number of positive diagnoses <6

Positive predictive value of screening for trisomy 21 stratified by risk level, 1 July 2010 to 30 June 2013

Table 23 shows PPV stratified by the risk level indicated in the screening result. For 2012/13, women that received a very increased risk result of 1:5 to 1:20 for trisomy 21 had a 24% probability that they were carrying a fetus with trisomy 21. The PPV remained at a higher level for women with increased risks of 1:21 to 1:50 (10% probability) but PPV decreased to a low value for women with increased risk results of 1:51 to 1:300 (3% probability).

Table 23: Positive predictive of screening for trisomy 21 stratified by risk level, 1 July 2010 to 30 June 2013

	Positive diagnostic test or infant diagnosis for women screened as increased risk (True Positives)			Negative diagnostic test or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value		
Risk level	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
1:5 to 1:20	56	53	50	170	159	157	0.25	0.25	0.24
1:21 to 1:50	12	18	12	158	130	106	0.07	0.12	0.10
1:51 to 1:300	10	12	18	652	607	627	0.02	0.02	0.03

Positive predictive value of screening for trisomy 21 by age, ethnicity and deprivation, 1 July 2010 to 30 June 2013

The PPV of screening for trisomy 21 also varied by age group, as shown in table 24. Low numbers for the two youngest and the oldest age groups mean that these have been suppressed. For the other age groups there is a trend of increased PPV with increasing age.

Table 24: Positive predictive of screening for trisomy 21 by age, 1 July 2010 to 30 June 2013

	Positive diagnostic test or infant diagnosis for women screened as increased risk (True Positives)			Negative diagnostic test or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value#		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
Age									
Under 20 years	1	0	1	9	13	5	-	-	-
20 – 24 years	1	1	3	47	49	52	-	-	-
25 – 29 years	5	5	6	99	89	81	0.05	0.05	0.07
30 – 34 years	17	18	15	204	199	192	0.08	0.08	0.07
35 – 39 years	33	32	32	385	312	308	0.08	0.09	0.09
40 – 44 years	19	25	22	220	216	227	0.08	0.10	0.09
45 years and over	2	2	1	16	18	25	-	-	-

rate suppressed if the number of positive diagnoses <6

PPV results by ethnicity are shown in table 25. The rate for Pacific is suppressed due to low numbers. Numbers for Māori and Asian groups are also low so caution should be taken when comparing rates.

Table 25: Positive predictive of screening for trisomy 21 by ethnicity, 1 July 2010 to 30 June 2013

	Positive diagnostic test or infant diagnosis for women screened as increased risk (True Positives)			Negative diagnostic test or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value#		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
Ethnicity									
Māori	8	4	12	106	101	110	0.07	0.04	0.10
Pacific	1	2	2	113	115	103	-	-	-
Asian	8	10	9	169	186	184	0.05	0.05	0.05
Other	61	67	57	592	494	493	0.09	0.12	0.10

rate suppressed if the number of positive diagnoses <6

Table 26 shows PPV by NZ deprivation index. There appears to be a trend of lower PPV with increasing deprivation but this should be interpreted with caution due to the low numbers for true positives.

Table 26: Positive predictive of screening for trisomy 21 by deprivation, 1 July 2010 to 30 June 2013

	Positive diagnostic test or infant diagnosis for women screened as increased risk (True Positives)			Negative diagnostic test or infant without diagnosis for women screened as increased risk (False Positives)			Positive predictive value		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
NZ Deprivation Index									
Decile 1 - 2	20	19	25	204	163	149	0.09	0.10	0.14
Decile 3 – 4	19	14	19	169	158	161	0.10	0.08	0.11
Decile 5 – 6	20	18	12	200	183	166	0.09	0.09	0.07
Decile 7 – 8	11	21	13	210	185	215	0.05	0.10	0.06
Decile 9 – 10	8	11	11	195	205	197	0.04	0.05	0.05
Unknown	0	0	0	2	2	2	0.00	0.00	0.00

Indicator 10: False positive rate

This section reports information on the false positive rate. The false positive rate is calculated by dividing the number of false positives (increased risk screening result and then a negative diagnostic test for trisomy, or a baby born without trisomy) by the number of false positive and true negatives (low risk screening result and then a negative diagnostic test for a trisomy, or a baby born without a trisomy).

False positive rate for screening 1 July 2010 to 30 June 2013

The overall false positive rate for trisomy 21, 18 or 13 for 2012/13 was 0.03 (or 3%). This means that out of all women who have a negative diagnostic or a baby without a trisomy, 3% will have received an increased risk result for trisomy 21, 18 or 13. The false positive rate was higher for second trimester screens than for first trimester screens.

Table 27: False positive rate for trisomy 21, 18 or 13, 1 July 2010 to 30 June 2013

	Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Negative diagnostic tests or infant without diagnosis for women screened as low risk (True Negatives)			False positive rate FP / (FP + TN)		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	751	616	659	28,470	30,215	29,516	0.03	0.02	0.02
T2 screens	223	244	216	3,649	3,983	4,263	0.06	0.06	0.05
Total screens	974	860	875	32,119	34,198	33,779	0.03	0.02	0.03

The false positive rate for trisomy 21 when considered alone was similar to the overall false positive rate (see table 28). However, the combined false positive rate for trisomy 18 and trisomy 13 is much lower (0.003 for 2012/13, see table 29).

Table 28: False positive rate for trisomy 21, 1 July 2010 to 30 June 2013

	Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Negative diagnostic tests or infant without diagnosis for women screened a low risk (True Negatives)			False positive rate FP / (FP + TN)		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	766	656	684	28,491	30,228	29,525	0.03	0.02	0.02
T2 screens	214	240	206	3,661	3,990	4,277	0.06	0.06	0.05
Total screens	980	896	890	32,152	34,218	33,802	0.03	0.03	0.03

Table 29: False positive rate for trisomy 13 or 18, 1 July 2010 to 30 June 2013

	Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Negative diagnostic tests or infant without diagnosis for women screened as low risk (True Negatives)			False positive rate FP / (FP + TN)		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	125	106	97	29,194	30,814	30,170	0.004	0.003	0.003
T2 screens	17	14	19	3,861	4,225	4,470	0.004	0.003	0.004
Total screens	142	120	116	33,055	35,039	34,640	0.004	0.003	0.003

False positive rate for screening for trisomy 21 by age, ethnicity and deprivation, 1 July 2010 to 30 June 2013

The false positive rate for trisomy 21 increased with age. For example, in 2012/13 the false positive rate for women under 20 years was 0.04 (0.4%) compared to 0.40 (40%) for women 45 years and over (see table 30). The reason being that prior risk (age) is included in the calculation. Older women are more likely to have a positive test and are also more likely to have a higher detection rate.

Table 30: False positive rate for trisomy 21 by age, 1 July 2010 to 30 June 2013

	Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Negative diagnostic tests or infant without diagnosis for women screened as low risk (True Negatives)			False positive rate FP / (FP + TN)		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
Age									
Under 20 years	9	13	5	1,489	1,549	1,414	0.01	0.01	0.004
20 – 24 years	47	49	52	4,664	4,972	5,110	0.01	0.01	0.01
25 – 29 years	99	89	81	8,435	9,358	9,279	0.01	0.01	0.01
30 – 34 years	204	199	192	10,268	10,900	10,734	0.02	0.02	0.02
35 – 39 years	385	312	308	6,240	6,214	6,108	0.06	0.05	0.05
40 – 44 years	220	216	227	1,024	1,188	1,120	0.18	0.15	0.17
45 years and over	16	18	25	32	37	37	0.33	0.33	0.40

False positive rate was relatively consistent across ethnic groups with the exception of Pacific, which showed a higher rate for all three financial years.

Table 31: False positive rate for trisomy 21 by ethnicity, 1 July 2010 to 30 June 2013

	Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Negative diagnostic tests or infant without diagnosis for women screened as low risk (True Negatives)			False positive rate FP / (FP + TN)		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
Ethnicity									
Māori	106	101	110	3,821	4,323	4,365	0.03	0.02	0.02
Pacific	113	115	103	2,112	2,342	2,307	0.05	0.05	0.04
Asian	169	186	184	4,931	6,065	6,014	0.03	0.03	0.03
Other	592	494	493	21,288	21,488	21,116	0.03	0.02	0.02

False positive rate was also relatively consistent by deprivation between 2.2% and 2.7% for 2012/13.

Table 32: False positive rate for trisomy 21 by deprivation, 1 July 2010 to 30 June 2013

	Negative diagnostic tests or infant without diagnosis for women screened as increased risk (False Positives)			Negative diagnostic tests or infant without diagnosis for women screened as low risk (True Negatives)			False positive rate FP / (FP + TN)		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
NZ Deprivation Index									
Decile 1 - 2	204	163	149	6,027	6,061	5,586	0.033	0.026	0.026
Decile 3 – 4	169	158	161	5,897	6,105	6,137	0.028	0.025	0.026
Decile 5 – 6	200	183	166	6,765	7,197	7,158	0.029	0.025	0.023
Decile 7 – 8	210	185	215	7,135	7,725	7,745	0.029	0.023	0.027
Decile 9 – 10	195	205	197	6,275	7,071	7,139	0.030	0.028	0.027

Indicator 11: Detection rate

This section reports information on the detection rate, or sensitivity, of screening. Detection rate is calculated by dividing the number of true positives (increased risk screening result for a specific trisomy and then a positive diagnostic test or a baby born with that specific trisomy) by the number of true positives and false negatives (low risk screening result for a specific trisomy and then a positive diagnostic test or a baby born with that specific trisomy).

Further information on the number of false negatives stratified by risk result is given in Appendix 5.

Detection rate for screening 1 July 2010 to 30 June 2013

The overall detection rate for trisomy 21, 18 or 13 for 2012/13 was 0.78 (or 78%). This was slightly down on 2011/12 (0.81) but higher than 2010/11 (0.73). A detection rate of 0.78 means that there is a 78% probability that a woman carrying a fetus with one of trisomy 21, 18 or 13 will receive an increased risk screening result for trisomy 21, 18 or 13.

Table 33: Detection rate for trisomy 21, 18 or 13, 1 July 2010 to 30 June 2013

	Positive diagnostic tests or infant with diagnosis for women screened as increased risk (true positives)			Positive diagnostic tests or infant with diagnosis for women screened as lowl risk (false negatives)			Detection rate TP / (TP + FN)		
	2010/11	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	97	117	98	37	25	28	0.72	0.82	0.78
T2 screens	7	10	11	2	5	3	0.78	0.67	0.79
Total screens	104	127	109	39	30	31	0.73	0.81	0.78

The detection rate for trisomy 21 alone is shown in table 34. The overall detection rate for 2012/13 is the same as the overall rate. The detection rate for trisomy 13 and 18 is lower at 0.66 for 2012/13 (see table 35).

Appendix 6 plots the receiver operating characteristic (ROC) curve for trisomies 21, 18 and 13 combined.

Table 34: Detection rate for trisomy 21, 1 July 2010 to 30 June 2013

	Positive diagnostic tests or infant with diagnosis for women screened as increased risk (true positives)			Positive diagnostic tests or infant with diagnosis for women screened as low risk (false negatives)			Detection rate TP / (TP + FN)		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	73	74	72	25	15	20	0.74	0.83	0.78
T2 screens	5	9	8	1	3	2	-	0.75	0.80
Total screens	78	83	80	26	18	22	0.75	0.82	0.78

rate suppressed if the number of positive diagnoses <6

Table 35: Detection rate for trisomy 13 or 18, 1 July 2010 to 30 June 2013

	Positive diagnostic tests or infant with diagnosis for women screened as increased risk (true positives)			Positive diagnostic tests or infant with diagnosis for women screened as lowl risk (false negatives)			Detection rate # TP / (TP + FN)		
Trimester of screen	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
T1 screens	23	38	23	13	15	11	0.64	0.72	0.68
T2 screens	1	0	2	2	3	2	-	-	-
Total screens	24	38	25	15	18	13	0.62	0.68	0.66

rate suppressed if the number of positive diagnoses <6

Detection rate for screening for trisomy 21 by age, ethnicity and deprivation, 1 July 2010 to 30 June 2013

Due to the low number of true positives and false negatives for some groups the detection rates for trisomy 21 have been calculated in aggregate across the three years in order to present more stable rates. Even after aggregating, rates for the youngest and oldest age groups have still been suppressed, as has the rate for Pacific.

The detection rate for trisomy 21 increased with age from 0.67 for women 25 to 29 years to 0.94 for women 40 to 44 years.

Table 36: Detection rate for trisomy 21 by age, 1 July 2010 to 30 June 2013 (aggregated)

	Positive diagnostic tests or infant with diagnosis for women screened as increased risk (true positives)	Positive diagnostic tests or infant with diagnosis for women screened as low risk (false negatives)	Detection rate#
Age			
Under 20 years	2	3	-
20 – 24 years	5	5	-
25 – 29 years	16	8	0.67
30 – 34 years	50	22	0.69
35 – 39 years	97	24	0.80
40 – 44 years	66	4	0.94
45 years and over	5	0	-

rate suppressed if the number of positive diagnoses <6

The aggregated detection rate for Asian women was 0.73 compared to 0.80 for Māori women and Other women (see table 37). However, low numbers mean this difference should be interpreted with caution.

Table 37: Detection rate for trisomy 21 by ethnicity, 1 July 2010 to 30 June 2013 (aggregated)

	Positive diagnostic tests or infant with diagnosis for women screened as increased risk (true positives)	Positive diagnostic tests or infant with diagnosis for women screened as low risk (false negatives)	Detection rate#
Ethnicity			
Māori	24	6	0.80
Pacific	5	5	-
Asian	27	10	0.73
Other	185	45	0.80

rate suppressed if the number of positive diagnoses <6

The aggregated detection rates by deprivation ranged from 0.73 to 0.83. There is no clear trend with increasing deprivation.

Table 38: Detection rate for trisomy 21 by deprivation, 1 July 2010 to 30 June 2013 (aggregated)

	Positive diagnostic tests or infant with diagnosis for women screened as increased risk (true positives)	Positive diagnostic tests or infant with diagnosis for women screened as low risk (false negatives)	Detection rate
NZ Deprivation Index			
Decile 1 - 2	64	13	0.83
Decile 3 – 4	52	12	0.81
Decile 5 – 6	50	15	0.77
Decile 7 – 8	45	17	0.73
Decile 9 – 10	30	9	0.77

References

National Screening Unit. 2013. Antenatal Screening for Down Syndrome and Other Conditions: *Guidelines for health practitioners*. Wellington: Ministry of Health.

Wald NJ, Rodeck C, Hackshaw AK, Walters J, Chitty L, Mackinson AM. 2003. First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS). Journal of Medical Screening. 10(2): 56–104.

Appendix 1: Indicator definitions

Table 39: definitions used for monitoring indicators

Indicator	Methodology
Indicator 2: Screens completed	Numerator: number of women who have a risk result calculated Denominator: number of live births and stillbirths
Indicator 5: Increased risk screening results	Numerator: number of women who receive an increased risk result Denominator: number of women who have a risk result calculated
Indicator 6: Diagnostic testing, increased risk screens	Numerator: number of women with an increased risk result that have a diagnostic test Denominator: number of women with increased risk results
Indicator 7: Diagnostic testing, low risk screens	Numerator: number of women with a low risk result that have a diagnostic test Denominator: number of women with low risk results
Indicator 8: Diagnostic testing, unscreened women	Number of women who have diagnostic test that have not participated in screening
Indicator 9: Positive predictive value	Numerator: number of women given an increased risk screen result who have a positive diagnostic test/baby with positive diagnosis Denominator: number of screened women with an increased risk result
Indicator 10: False positive rate	Numerator: number of women given an increased risk screen result who do not have a positive diagnostic test/baby with positive diagnosis Denominator: number of screened women who do not have a positive diagnostic test/baby with positive diagnosis
Indicator 11: Detection rate	Numerator: number of women given an increased risk screen result who have a positive diagnostic test/baby with positive diagnosis Denominator: number of screened women who have a positive diagnostic test/baby with positive diagnosis

Calculation rules

- Screen date is the date given as the 'Collected date' in the lab system
- Each screen is assigned a case identification number in the lab system. If a woman has a second screen for the same pregnancy it will have the same case number. For all indicator calculations the result of the latest screen is used and a given case number can only count once towards the total
- Denominator is live births and still births >20 weeks or > 400g
- Tests on products of conception are excluded from prenatal tests for the purposes of indicators 6, 7 and 8. However, they are included for indicators 9, 10 and 11
- For a prenatal cytogenetic test to link to a screen the cytogenetic sample date must be later than the screen date, but not more than 105 days (15 weeks) later
- For an infant diagnosis to link to a commenced screen the screen date must be earlier than the infant's birth date and the date difference must not be greater than 230 days (approximately 33 weeks)

Appendix 2: Birth denominator data

Data on the number of live and still births[2] was obtained from the national Maternity Collection for each financial year. Data used in this report for the 2012/13 year is considered provisional.

Table 40: Live births and still births by District Health Board

DHB	2010/11	2011/12	2012/13
Auckland	6,710	6,600	6,442
Bay of Plenty	3,012	2,865	2,879
Canterbury	6,412	5,917	5,954
Capital and Coast	3,941	3,805	3,844
Counties Manukau	8,676	8,681	8,586
Hawkes Bay	2,301	2,234	2,172
Hutt Valley	2,088	2,030	1,937
Lakes	1,591	1,555	1,505
MidCentral	2,359	2,212	2,079
Nelson Marlborough	1,638	1,603	1,537
Northland	2,393	2,294	2,217
South Canterbury	624	586	671
Southern	3,713	3,601	3,547
Tairawhiti	766	756	705
Taranaki	1,591	1,571	1,511
Waikato	5,525	5,413	5,269
Wairarapa	538	530	481
Waitemata	7,809	7,882	7,885
West Coast	411	400	409
Whanganui	859	838	838
Total	**62,957**	**61,373**	**60,468**

Table 41: Live births and still births by age group

Age group	2010/11	2011/12	2012/13
Under 20	4,314	4,034	3,583
20-24	11,914	11,456	11,229
25-29	15,691	15,539	15,703
30-34	17,413	17,297	17,162
35-39	11,007	10,438	10,176
40-44	2,485	2,456	2,476
45 and over	108	138	124
Unknown	25	15	15
Grand Total	**62,957**	**61,373**	**60,468**

[2] births reaching at least 20 weeks gestation or >=400g birth weight

Table 42: Live births and still births by 2006 deprivation decile

Deprivation decile 2006	2010/11	2011/12	2012/13
1	4,551	4,462	4,607
2	4,382	4,378	4,314
3	4,799	4,656	4,782
4	5,454	5,200	5,219
5	5,418	5,112	5,208
6	6,434	6,406	6,237
7	6,363	6,302	6,033
8	7,930	7,778	7,719
9	8,663	8,445	8,131
10	8,926	8,600	8,183
Unknown	37	34	35
Grand Total	**62,957**	**61,373**	**60,468**

Appendix 3: Summary of diagnostic testing uptake and results for women that had an increased risk screen

Summary of prenatal diagnostic testing uptake 1 July 2012 – 30 June 2013

Of the 984 increased risk screens during the 2012/13 year, 609 (61.9%) women had a prenatal diagnostic test (CVS or amniocentesis). Of the 609 diagnostic tests, 104 had an abnormal karyotype, 2 were failed tests.

Table 43: Diagnostic results for women that accessed a prenatal diagnostic test following an increased risk screen

Karyotype result		
	Number	Percentage
Normal karyotype	503	82.6%
Confirmed Down Syndrome	63	10.3%
Other result*	41	6.7%
Failed test	2	0.3%
Total	609	100%

*The 41 'Other' results were made up of the following:

Result	Number
Trisomy 18	16
Trisomy 13	7
Turner syndrome	3
Triploidy	2
Sex chromosome aneuploidy (other than non-mosaic 45,X)	4
Autosomal trisomy (other than 13, 18, 21) (including mosaic)	2
Partial aneuploidy (autosome) (including mosaic)	4
Apparently balanced chromosome rearrangement	2
Structural abnormality	1
Total	41

Appendix 4: Measuring screening performance

Figure 7 shows the categorisation of screening results used to calculate screening performance measures such as positive predictive value, false positive rate and detection rate. The examples given in this appendix focus on trisomy 21.

Figure 7: categorisation of screening results

	Trisomy 21 diagnosis	**No trisomy 21 diagnosis**	**Total**
Screen result = Increased risk	A (true positives)	B (false positives)	A+B
Screen result = Low risk	C (false negatives)	D (true negatives)	C+D
	A+C	B+D	N (total screens)

Positive predictive value and positive test rate

The positive test rate is the number of increased risk screens per 100 screens.

$$\text{Positive test rate} = ((A+B)/N)*100$$

Positive Predictive Value is the probability of having the condition given screen result was increased risk.

$$PPV = P(\text{Disease} \mid \text{Screen Positive}) = A/(A+B)$$

In order for PPV to increase, 'A' needs to be higher (more true positives) and/or 'B' needs to be lower (less false positives). However, an increase in positive test rate can come about when 'A' and/or 'B' increase. If the positive test rate increases due to higher true positives (A), then PPV will also increase. If instead the number of false positives increases, then the positive test rate will increase but PPV will decrease.

False positive rate

False positive rate is the number of false positives divided by false positives plus true negatives. It gives the proportion of women that did not have a baby or fetus with trisomy 21 that received an increased risk screening result.

$$FPR = B/(B+D)$$

Detection rate

Detection rate is the number of true positives divided by true positives plus false negatives. It gives the probability that a woman carrying a fetus with trisomy 21 will receive an increased risk screening result for trisomy 21.

$$\text{Detection rate} = A/(A+C)$$

Data for women screened during 2012/13

Figure 8, below, shows the data break down in relation to trisomy 21 for women screened during 2012/13. This data focuses on trisomy 21 and excludes Canterbury, South Canterbury and West Coast so the totals will not be the same as indicators 2 and 5 in the report.

Figure 8: categorisation of trisomy 21 screening results 2012/13

	Trisomy 21 diagnosis	**No trisomy 21 diagnosis**	**Total**
Screen result = Increased risk	A = 80	B = 890	A+B = 970
Screen result = Low risk	C = 22	D = 33,802	C+D = 33,824
	A+C = 102	B+D = 34,692	N = 34,794 (total screens)

Positive predictive value (indicator 9)

PPV = A/(A+B)
= 80 / 970
= 0.08 (or 8%)

If a woman receives an increased risk screening result for trisomy 21, there is an 8% probability that she is carrying a fetus with trisomy 21.

False positive rate (indicator 10)

FPR = B/(B+D)
= 890 / 34,692
= 0.03 (or 3%)

Out of all women that ultimately have a negative diagnostic test or a baby without trisomy 21, 3% will have received an increased risk screening result.

Detection rate (indicator 11)

Detection rate = A/(A+C)
= 80 / 102
= 0.78 (or 78%)

There is a 78% probability that a woman carrying a fetus with trisomy 21 will have received an increased risk screening result for trisomy 21.

Appendix 5: False negative screens by risk level

There were 100 false negative screens in total across the period covered by this report. A false negative means that the screen result was low risk for each of trisomy 21, 18 and 13 but there was then a positive diagnostic test or infant diagnosis for one of trisomy 21, 18 or 13.

Table 44 shows the number of false negatives for each of the three financial years broken down by the screening risk result in the first group of columns. The next group of columns gives the total numbers of negative (low risk) screens. Overall, false negative screens make up 0.1% of all negative screens.

Table 44: False negative screens for trisomy 21, 18 and 13 by risk level, 1 July 2010 to 30 June 2013

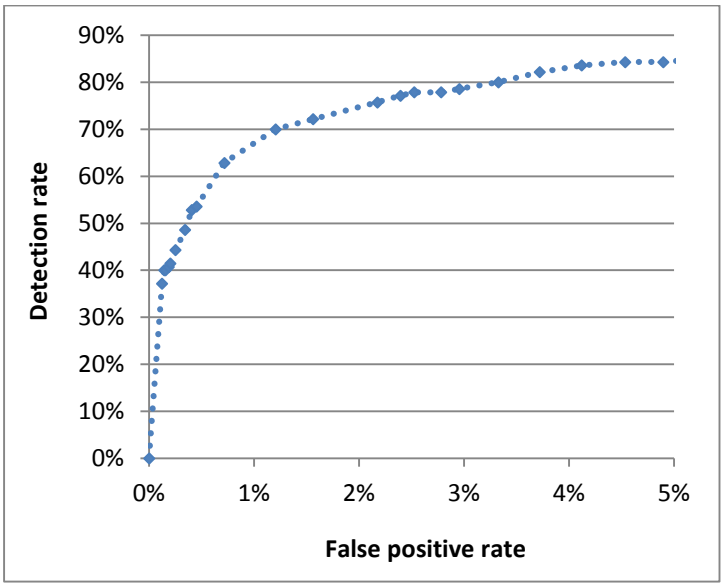
	False negatives			Total negative (low risk) screens			% negative screens that are false		
Risk level	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**	**2010/11**	**2011/12**	**2012/13**
1:301 to 1:500	8	5	8	544	518	561	1.5%	1.0%	1.4%
1:501 to 1:1,000	8	7	5	1,422	1,378	1,483	0.6%	0.5%	0.3%
1:1001 to 1:2000	6	6	8	2,372	2,380	2,489	0.3%	0.3%	0.3%
1:2001 to 1:3000	4	3	1	2,096	2,064	2,169	0.2%	0.1%	0.0%
1:3001 to 1:4000	4	0	3	1,883	1,960	1,961	0.2%	0.0%	0.2%
1:4001 to 1:5000	3	2	1	1,679	1,704	1,739	0.2%	0.1%	0.1%
1:5001 to 1:10,000	2	4	3	6,689	6,895	6,684	0.0%	0.1%	0.0%
1:10,001 to 1:100,000	4	3	2	15,473	17,329	16,724	0.0%	0.0%	0.0%
Total	**39**	**30**	**31**	**32,158**	**34,228**	**33,810**	**0.1%**	**0.1%**	**0.1%**

Appendix 6: ROC curve

Figure 9 shows the receiver operating characteristic (ROC) curve for the test used to screen for Down syndrome and other conditions. This plots the false positive rate on the horizontal x axis against detection rate on the vertical y axis for different possible cut off points of the screening test. The aim for a screening test is to maximise detection rate while minimising false positive rate.

In New Zealand the cut off used for screening is 1:300. With this cut off the overall detection rate for trisomy 21, trisomy 18 and trisomy 13 in 2012/13 was 78%, and the false positive rate was 3%. To create the graph the detection rate and false positive rate were calculated for a range of other cut off points in order to plot the curve. What the curve shows is that it if the cut off was lowered to increase the detection rate to 84%, the false positive rate would increase from 3% to 5%.

Figure 9: ROC curve for trisomy 21, 18 and 13 screening 2012/13



Appendix 7: Glossary

Alpha-fetoprotein (AFP) - a protein that is normally produced by the fetus. Maternal serum AFP levels can be used as a biochemical marker in the detection of certain fetal abnormalities including neural tube defects (NTDs) after 15 weeks of pregnancy.

Amniocentesis – a procedure involving the withdrawal of a small amount of amniotic fluid by needle and syringe through the abdomen guided by ultrasound performed at the same time. The tests performed on fetal cells in this sample can detect a range of chromosomal and genetic disorders.

Analyte - a substance that is undergoing analysis or being measured. Analytes measured in antenatal screening include: pregnancy associated plasma protein-A, beta human chorionic gonadotropin, unconjugated oestriol, alpha fetoprotein and inhibin A.

Beta-human chorionic gonadotropin (ßhCG) - a hormone produced during pregnancy and present in maternal blood and urine. It is used as a biochemical marker for Down syndrome and other conditions in first trimester combined and second trimester maternal serum screening.

Chorionic villus sampling (CVS) – a procedure involving the withdrawal of a small amount of placental tissue by needle and syringe through the abdomen guided by ultrasound performed at the same time. Tests performed on placental cells can detect a range of chromosomal and genetic disorders.

Crown rump length (CRL) - the measurement from the fetal crown to the prominence of the buttocks or breech. This is used for dating in the first trimester.

Detection rate - the ability of screening to identify individuals with the condition screened for. A test with a high detection rate will have few false negative results. Also referred to as sensitivity.
False negative result - when a woman receives a low risk screening result but the baby does have the condition screened for.

False positive result - when a woman receives an increased risk screening result but the baby does not have the condition screened for.

False positive rate – the false positive rate is the number of false positives divided by the number of false positives and true negatives. A low false positive rate corresponds with a high level of specificity, which refers to the ability of screening to identify individuals who do not have the condition screened for.

Inhibin A - a hormone secreted by the ovary that is used as a biochemical marker in second trimester maternal serum screening for Down syndrome and other conditions.

Neural tube defect (NTD) - a congenital anomaly involving the brain and spinal cord caused by failure of the neural tube to close properly during embryonic development. Open NTDs occur

when the brain and/or spinal cord are exposed at birth through a defect in the skull or vertebrae. Examples of open NTDs are spina bifida (myelomeningocele), anencephaly, and encephalocele.

Nuchal translucency (NT) - sonographic appearance of the collection of fluid under the skin at the back of the fetal neck. NT is a marker for chromosomal and other anomalies and can be measured in the first trimester of pregnancy.

Pregnancy-associated plasma protein A (PAPP-A) - a protein originating from the placenta used as a biochemical marker in first trimester combined screening for Down syndrome and other conditions.

Risk calculation algorithm - an explicit protocol (in this case computer based) that combines a number of factors in determining overall risk of a particular outcome or condition.

Screening - a way of identifying a group of people who are more likely than others to have a particular condition. The screening process involves testing people for the presence of the condition, and predicting the likelihood that they have the condition. Antenatal screening for Down syndrome and other conditions predicts the likelihood of the conditions being present in the fetus.

Triploidy – an extremely rare chromosomal disorder in which a baby has three of every chromosome making a total of sixty-nine rather than the normal forty-six chromosomes.

Trisomy - a group of chromosomal disorders in which there are three copies, instead of the normal two, of a particular chromosome present in the cell nuclei. The most common trisomies in newborns are trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome).

Unconjugated oestriol (uE3) - a hormone produced by the placenta and used as a biochemical marker in second trimester maternal serum screening for Down syndrome and other conditions.

Further terms can be found at www.nsu.govt.nz