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MANATŪ HAUORA

New Zealand Government

Antenatal Screening for Down Syndrome and Other Conditions

Monitoring Report

January 2011 to December 2014

National Screening Unit

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Executive summary

This report presents the data for the four calendar years from 1 January 2011 to 31 December 2014 and is based on screening that occurred during that time. Due to lack of data from one of the diagnostic laboratories, the indicators that involve diagnostic data are only reported for 17 DHBs (indicators 6 to 11).

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. This screening is optional for pregnant women. Women who are less than 20 weeks pregnant are 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 the Nuchal Translucency scan should be done 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 January 2011 to December 2014

- Screening was commenced for more than 75% of pregnancies [indicator 1].
- Screening uptake by Māori and Pacific women was less than half the rate of Other women in 2014 but has increased each year [indicators 1 and 2].
- The national screening completion rate exceeded two-thirds of births for the first time in 2013 (69%) and increased again in 2014 (71%). Trimester one screens made up 87% of all completed screens in 2014 [indicator 2].
- Most DHBs showed a trend of increasing rates of screening commencement and completion over the four years covered in this report [indicators 1 and 2].
- Nearly half of all completed trimester 2 screens were commenced in trimester 1 [indicator 3].
- Nine percent of screens commenced in 2014 were not completed and nearly all related to screens commenced in the first trimester. The rate of incomplete screens was higher for younger women, for Māori and Pacific women, and for women from areas of higher deprivation [indicator 4].
- The positive test rate (number of increased risk results per 100 screens) for trisomy 21, 18 and 13 was 2.8 in 2014, up from 2.7 in 2013. Positive test rate was higher for second trimester screens (5 per 100 screens) than for first trimester screens (2.4 per 100 screens) for 2014 [indicator 5].

- The false positive rate for trisomy 21, 18 and 13 was 2% in 2014, which was equal to 2013. The rate was higher for second trimester screens (5%) than for first trimester screens (2%) [indicator 10].
- The overall detection rate for trisomy 21, 18 and 13 was 80% in 2014, up from 76% in 2013. The detection rate was lower for first (79%) compared with second (86%) 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 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 weeks and 13 weeks and 6 days gestation and combined with an ultrasound scan to determine nuchal translucency (NT) and crown rump length (CRL) measurements between 11 weeks and 2 days and 13 weeks and 6 days, or
- 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
- Turner syndrome
- 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

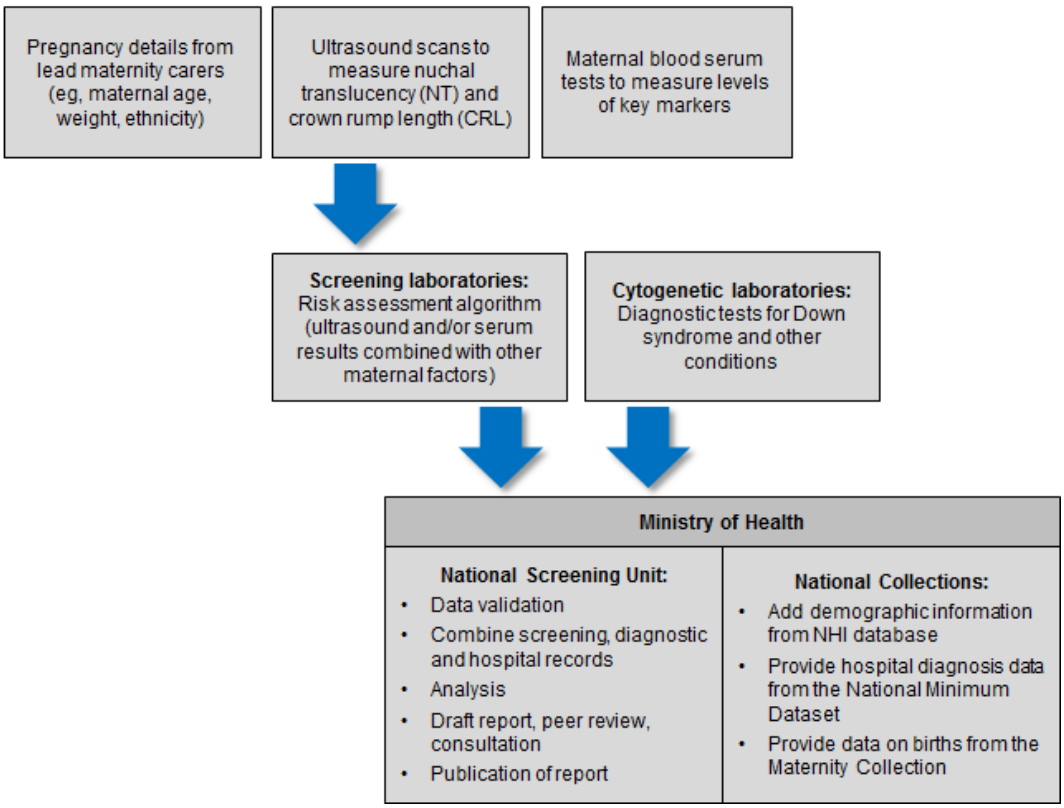
This report presents information on antenatal screening for Down syndrome and other conditions between 1 January 2011 and 31 December 2014.

The indicators in this report are taken from the 2014 Antenatal Screening for Down Syndrome and Other Conditions, Monitoring and Evaluation Framework. Appendix 1 contains definitions for these indicators. Figure 1 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 screening data in this report was sourced from LabPLUS and covers all of New Zealand. Cytogenetic testing data was received from LabPLUS, Waikato, and Capital and Coast laboratories but was not provided by Canterbury Health Laboratories (CHL). 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, cytogenetic laboratories, and the NMDS was undertaken to identify the outcome for all screened women.

Definitions

Commenced screening

At least one of the required components of the screening test was completed.

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 1:310 is a low risk.

Increased risk result

An increased risk result is defined as a risk higher than or equal to 1:300. For some indicators increased risk screening results are further stratified into:

- 1:5 to 1:20
- 1:25 to 1:50
- 1:55 to 1:300.[1]

[1] Risk ratio values increase in increments of 5 between 1:10 and 1:100, increments of 100 between 1:100 and 1:10,000, and then increments of 1000 to 1:100,000.

Inclusion criteria

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

- screening commencement date between 1 January 2011 and 31 December 2014 (ie, 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)
- single screening result per pregnancy.

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

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 79% of the *Other* ethnicity group. There were no women in the final dataset with ethnicity recorded as *Unknown*.

NZ Deprivation

The New Zealand deprivation index (NZ Dep) is the average level of deprivation of people living in an area at a particular point in time, relative to the whole of New Zealand. Deprivation refers to areas (based on New Zealand Census mesh blocks) rather than individuals. All reporting by NZ Dep is based on the 2013 New Zealand deprivation index decile associated with the residential address held in the NHI database for each woman at the time of data extraction.

This report presents results by 2013 NZ Dep quintiles. Each quintile groups two deciles together and contains about 20% of small areas in New Zealand. The two quintiles at opposite ends of the scale are quintile 1 (deciles 1 and 2), which represents children living in the least deprived 20% of small areas ('the least deprived areas'), and quintile 5 (deciles 9 and 10), which represents children living in the most deprived 20% of small areas ('the most deprived areas'). This is opposite to some other systems of classification, such as that used by education, where level 10 is the least disadvantaged and level 1 the most disadvantaged.

Births

Data on the number of live and still births[2] was obtained from the national Maternity Collection for each calendar year. Appendix 2 contains tables for the denominators used in this report.

Small numbers

Small numbers can affect the reliability of results. Where an indicator calculation involves small counts (less than 10) 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 (CVS) or amniocentesis (tests on products of conception are not included). 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 screen for the same woman within 112 days. Where this occurred, the first completed screen was retained for the analysis. The figure of 112 days was based on the timing of the screening test and considering how soon a woman may become pregnant again following a miscarriage.

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)
- 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.

These were based on the possible timing of the different screening and diagnostic tests.

Data limitations

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 (ie, all pregnant women that reach 9 weeks gestation) is likely to be larger than the denominator used (ie, all births reaching at least 20 weeks gestation or at least 400 g birth weight).

[2] Births reaching at least 20 weeks gestation or ≥400 g 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.
- 328 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 (48 records less than 12 years, 69 records 50 years old or greater) and three instances where date of death as recorded in the NHI database was prior to the date of screen. 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 includes screening records for women from Canterbury, South Canterbury and West Coast DHBs. Records for these women are included in the results for indicators 1, 2, 3, 4 and 5 but excluded from indicators 6 to 11.

Table 1: Screening dataset cleansing

	Number	Percentage
Records received for report period	189,965	100.0%
Final screening dataset for analysis	178,228	94.3%
Total excluded records[1]	10,737	5.7%
Private/overseas screens	4938	2.6%
Invalid NHI	134	0.1%
Unknown DHB	328	0.2%
Date of death prior to screen	3	<0.01%
Age at screen < 12	48	<0.01%
Age at screen > 49	69	<0.01%
Repeat screen[1]	5217	2.7%

1 For this report data on both complete and incomplete screens was received. Where a completed screen exists for a pregnancy any incomplete screens (those with no risk reported) are not considered true incompletes and have been excluded. This has led to a higher number of repeat screen exclusions when compared with the July 2010 to June 2013 report.

Indicator 1: Screens commenced

This indicator reports the number of screens commenced by trimester of screening (first or second), reported by DHB, age, ethnicity, and NZ deprivation quintile.

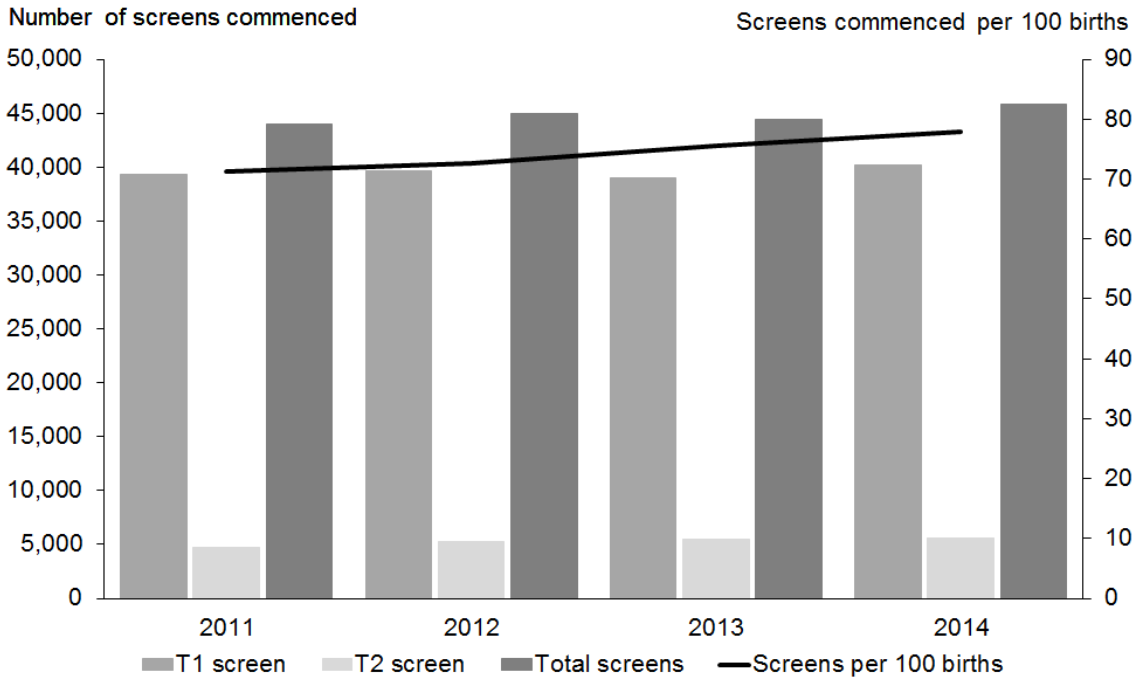
Total screens commenced by trimester

During 2014, a total of 45,840 screens were commenced, a rate of 78.0 per 100 births. Table 2 shows the total number of screens commenced by year and trimester of screen. Throughout the report T1 is used to refer to first trimester and T2 to second trimester. The vast majority of screens were T1 screens. The number of screens commenced per 100 births has increased over time from 71.2 in 2011 to 78.0 in 2014 (see Table 2 and Figure 2).

Table 2: Total screens commenced by trimester, January 2011 to December 2014

Trimester of screen	Number and rate of screens commenced			
	2011	2012	2013	2014
T1 screen	39,315	39,679	38,961	40,230
T2 screen	4698	5238	5497	5610
Total screens	**44,013**	**44,917**	**44,458**	**45,840**
Screens per 100 births	71.2	72.6	75.6	78.0

Figure 2: Count and rate of screens commenced, January 2011 to December 2014



Screens commenced by DHB

Figure 3 shows the screening commencement rates by DHB for 2014. There was a large variation in rates from 56 per 100 births in Northland to 97 per 100 births in Nelson Marlborough (see Figure 3). Most DHBs (12) had rates of 77 per 100 births or above. Table 3 gives a full breakdown by the trimester of the screen.

Figure 3: Screens commenced by DHB, January 2014 to December 2014

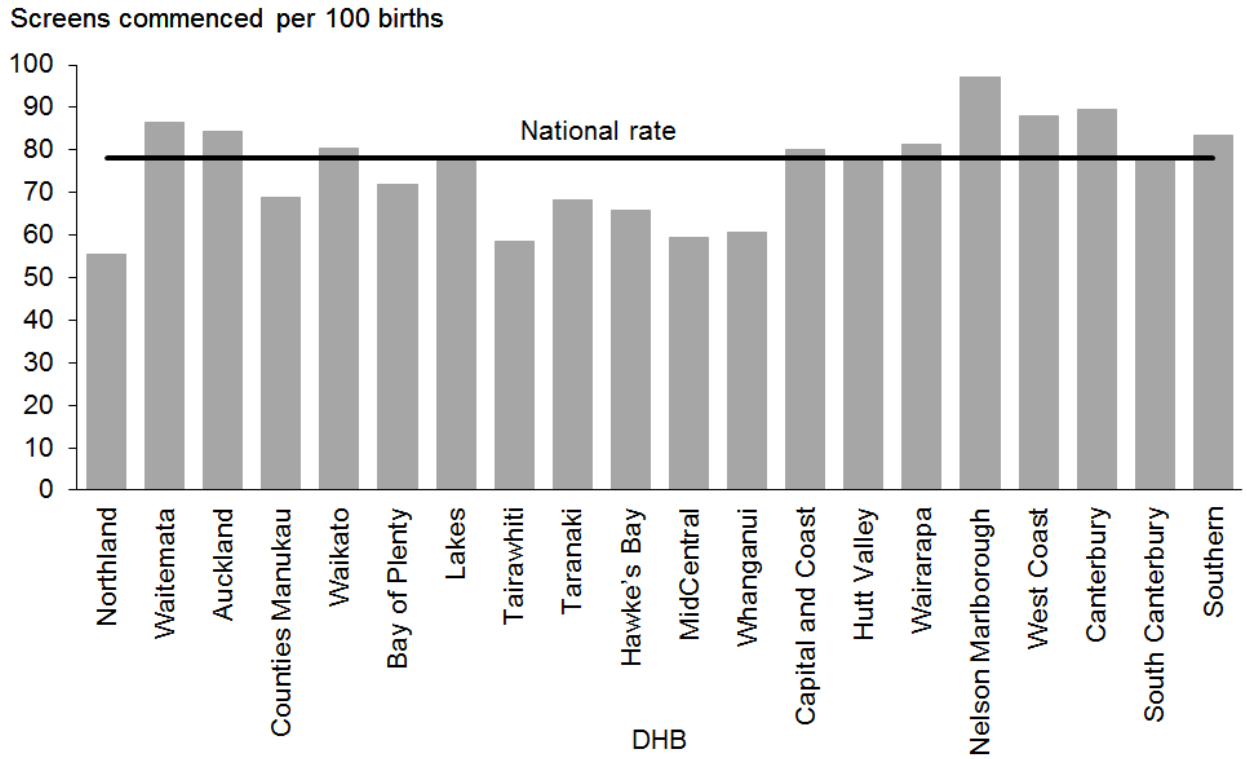


Table 3: Screens commenced by trimester and DHB, January 2014 to December 2014

DHB	Number of screens commenced			Screens commenced (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	1010	160	1170	48.0	7.6	55.6
Waitemata	6069	722	6791	77.3	9.2	86.5
Auckland	4673	643	5316	74.1	10.2	84.3
Counties Manukau	4587	1125	5712	55.3	13.6	68.9
Waikato	3768	455	4223	71.6	8.7	80.3
Bay of Plenty	1813	196	2009	65.0	7.0	72.0
Lakes	899	178	1077	64.5	12.8	77.3
Tairawhiti	343	64	407	49.3	9.2	58.5
Taranaki	827	210	1037	54.5	13.8	68.3
Hawke's Bay	1223	141	1364	58.9	6.8	65.7
MidCentral	1105	134	1239	52.9	6.4	59.3
Whanganui	418	77	495	51.1	9.4	60.5
Capital and Coast	2575	257	2832	72.9	7.3	80.2
Hutt Valley	1277	173	1450	68.8	9.3	78.1
Wairarapa	343	42	385	72.4	8.9	81.2
Nelson Marlborough	1238	144	1382	87.0	10.1	97.1
West Coast	270	38	308	77.1	10.9	88.0
Canterbury	4812	575	5387	80.0	9.6	89.6
South Canterbury	472	40	512	72.2	6.1	78.3
Southern	2508	236	2744	76.3	7.2	83.5
Total	**40,230**	**5610**	**45,840**	**68.4**	**9.5**	**78.0**

Most DHBs showed a trend of increasing rates of screening over the four years covered in this report. Exceptions to this were Waitemata and Canterbury, where the rate levelled off between 2013 and 2014, and South Canterbury, which had a decreasing trend over the four-year period (see Table 4).

Table 4: Screens commenced per 100 births by DHB, January 2011 to December 2014

DHB	Screens commenced (per 100 births)			
	2011	2012	2013	2014
Northland	47.0	50.0	53.3	55.6
Waitemata	84.2	83.1	86.5	86.5
Auckland	75.2	74.7	82.6	84.3
Counties Manukau	61.1	63.6	65.1	68.9
Waikato	73.2	72.5	76.7	80.3
Lakes	65.4	68.8	69.7	72.0
Bay of Plenty	60.9	67.9	70.2	77.3
Tairawhiti	44.5	49.5	53.0	58.5
Taranaki	63.3	60.5	61.6	68.3
Hawke's Bay	56.1	62.0	64.6	65.7
Whanganui	51.3	54.5	58.3	59.3
MidCentral	45.5	45.4	48.2	60.5
Hutt Valley	77.0	79.5	78.4	80.2
Capital and Coast	71.5	71.0	72.7	78.1
Wairarapa	73.0	69.4	76.5	81.2
Nelson Marlborough	88.5	91.1	87.6	97.1
West Coast	69.4	76.9	82.3	88.0
Canterbury	85.9	87.2	90.7	89.6
South Canterbury	92.0	86.0	88.8	78.3
Southern	76.0	80.2	81.7	83.5
Total	**71.2**	**72.6**	**75.6**	**78.0**

Screens commenced by age, ethnicity and deprivation

Table 5 provides an overall view of screens commenced by age, ethnicity and NZ deprivation quintile for January 2011 to December 2014. The 30–34 year age group had the highest rate of screens commenced with 83 women starting screening per 100 births in 2014. This was followed by the 25–39 years age group with 82 per 100 births (see Figure 4).

Screening commencement rates were highest among women of Other ethnicity at 96 per 100 births for 2014. This was followed closely by Asian women at 91. The rate of commenced screens for Pacific and Māori women was lower at 49 per 100 births and 44 per 100 births respectively (see Figure 5). However, both groups have shown significant increase in over the four years (see Table 5).

Screening commencement rates were highest among women in less deprived areas with 92 women per 100 per births starting screening for quintiles 1 and 2 in 2014 compared with 60 per 100 births for quintile 5 (see Figure 6). However, the rate decreased for quintile 1 between 2013 and 2014 (see Table 5).

Table 5: Screens commenced by age of mother, ethnicity and NZ deprivation quintile, January 2011 to December 2014

	Number of screens commenced				Screens commenced (per 100 births)[#]			
	2011	**2012**	**2013**	**2014**	**2011**	**2012**	**2013**	**2014**
Age at screen								
Under 20 years	2298	2136	1952	1989	56.7	54.7	58.6	66.3
20–24 years	6863	6998	6959	7066	58.6	61.0	64.4	68.6
25–29 years	11,584	12,131	12,066	12,804	74.5	76.1	79.0	81.5
30–34 years	13,506	13,792	13,971	14,641	78.4	79.1	83.3	83.2
35–39 years	8050	8063	7662	7631	75.0	77.5	76.3	78.7
40–44 years	1639	1720	1768	1628	68.2	66.7	72.6	69.4
45 years and over	73	77	80	81	58.4	63.6	55.9	61.4
Ethnicity								
Māori	5562	5903	5823	6294	35.2	37.8	40.2	44.4
Pacific	3068	3116	3012	3012	43.4	45.4	47.5	48.9
Asian	6515	7421	7491	8442	91.3	87.8	91.8	91.6
Other	28,868	28,477	28,132	28,092	90.7	92.1	94.4	96.1
NZ Deprivation Quintile								
Quintile 1	8176	8107	7692	7764	96.1	93.4	94.1	91.7
Quintile 2	8216	8425	8262	8415	86.4	87.6	89.3	91.7
Quintile 3	8575	8708	8757	8899	76.9	78.0	82.4	84.2
Quintile 4	9586	9859	9914	10,345	69.4	72.2	73.9	77.8
Quintile 5	9451	9814	9829	10,416	50.2	52.4	56.8	60.4
Unknown	9	4	4	1	–	–	–	–
Total	**44,013**	**44,917**	**44,458**	**45,840**	**71.2**	**72.6**	**75.6**	**78.0**

Rate suppressed if the number of screens was <10.

Figure 4: Screens commenced by age of mother at screen, January 2014 to December 2014

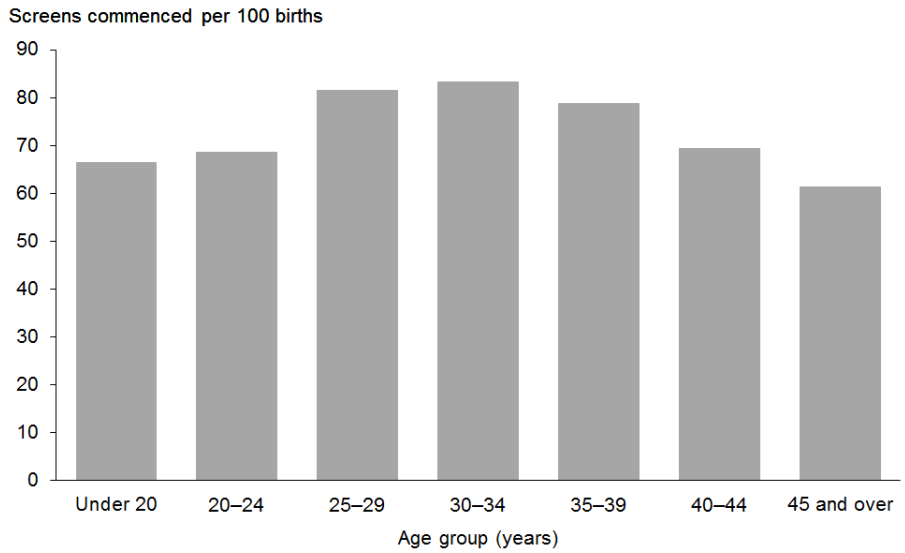


Figure 5: Screens commenced by ethnicity of mother, January 2014 to December 2014

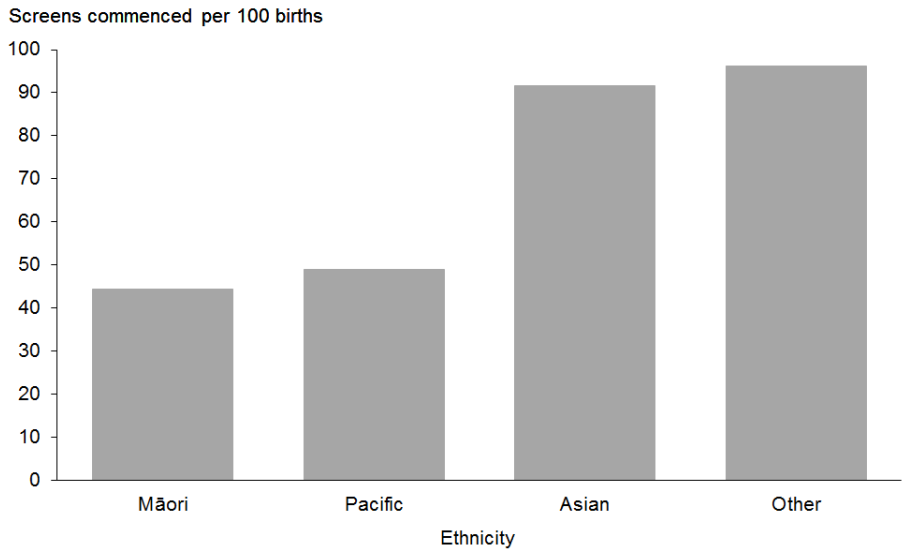
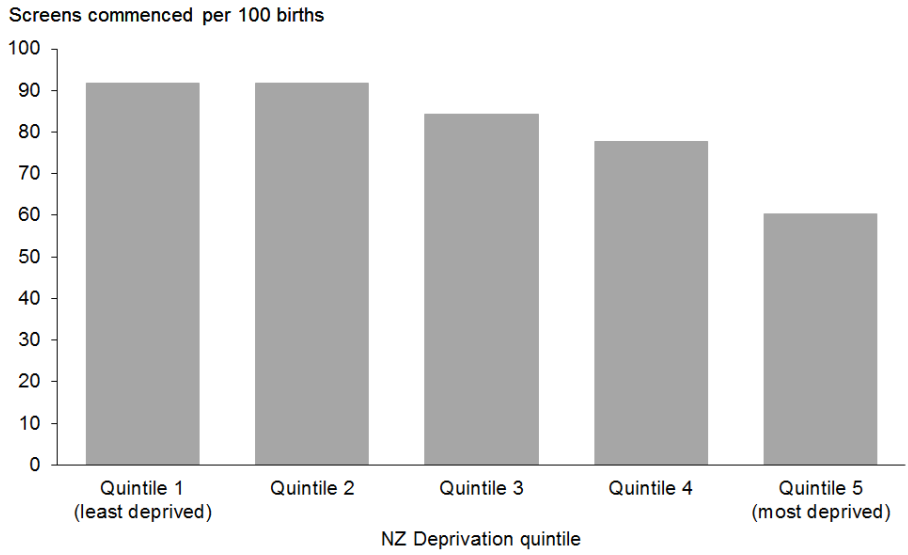


Figure 6: Screens commenced by NZ deprivation quintile, January 2014 to December 2014



Indicator 2: Screens completed

This indicator reports the number of screens completed by trimester of screening, DHB, age, ethnicity, and NZ deprivation quintile.

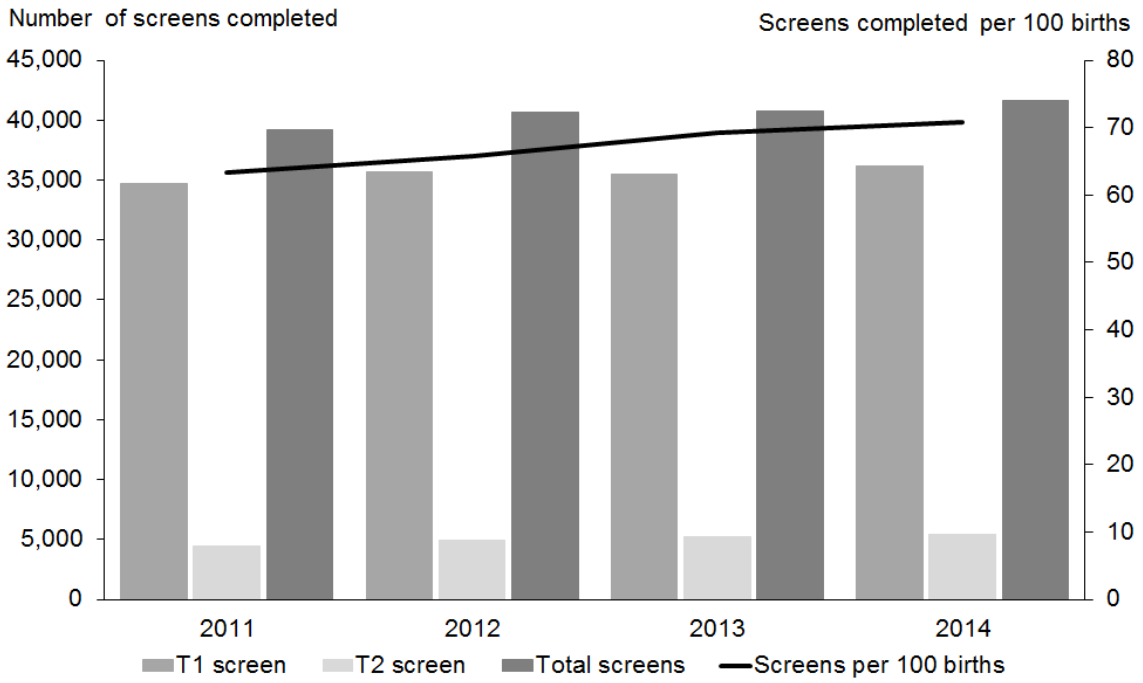
Total screens completed by trimester

During 2014, a total of 41,656 screens were completed, a rate of 71 per 100 births. Table 6 and Figure 7 show the total number of screens completed per year and trimester of screen. Across all years the majority of screens were completed in the first trimester. The total number of completed screens has increased annually since 2011. The trend for screens per 100 births was similar, with an increase of 7.5 per 100 births since 2011.

Table 6: Total screens completed by trimester, January 2011 to December 2014

Trimester of screen	Number and rate of screens completed			
	2011	2012	2013	2014
T1 screen	34,735	35,691	35,464	36,206
T2 screen	4446	4957	5269	5450
Total screens	**39,181**	**40,648**	**40,733**	**41,656**
Screens per 100 births	63.4	65.7	69.3	70.9

Figure 7: Count and rate of screens completed, January 2011 to December 2014



Screens completed by DHB

Screening completion rates for 2014 varied across DHBs from 87 per 100 births in Nelson Marlborough to 48 per 100 births in Northland (see Figure 8). Table 7 gives a full breakdown by the trimester of the screen.

Figure 8: Screens completed by DHB, January 2014 to December 2014

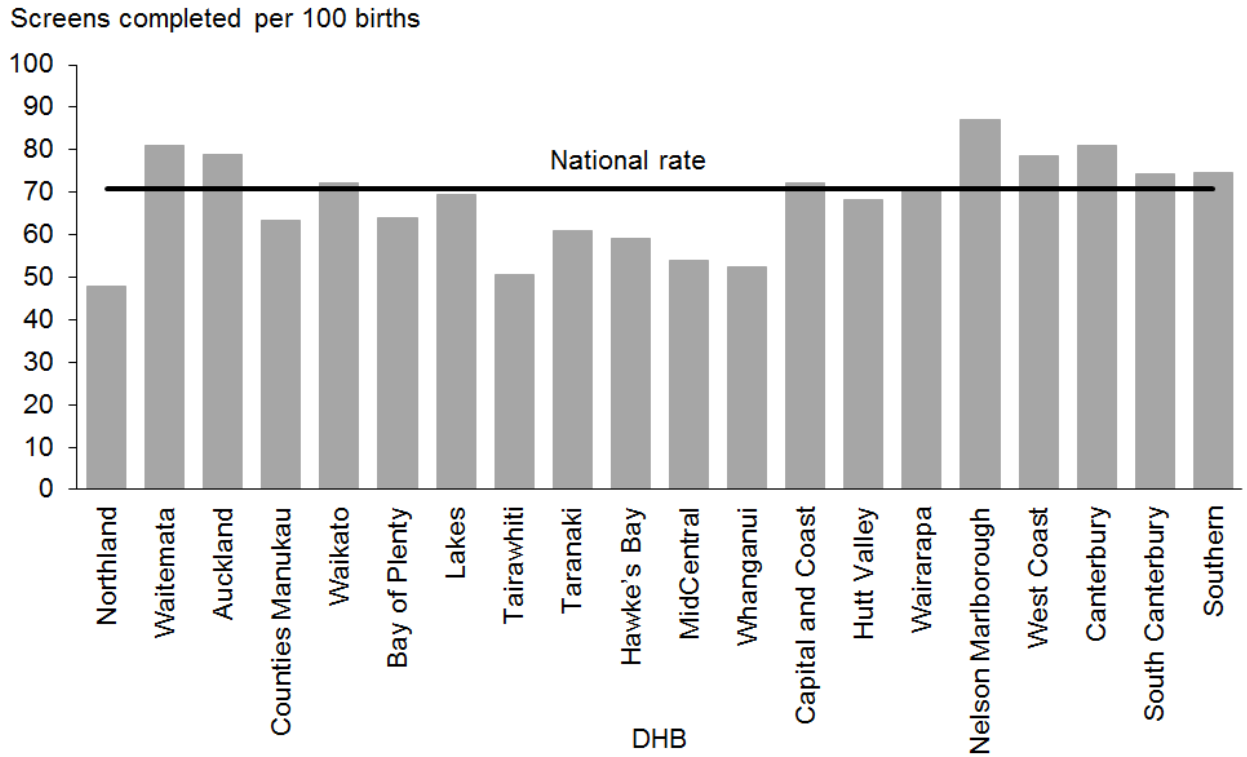


Table 7: Screening completion by trimester and DHB, January 2014 to December 2014

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	853	155	1008	40.5	7.4	47.9
Waitemata	5653	704	6357	72.0	9.0	81.0
Auckland	4344	625	4969	68.9	9.9	78.8
Counties Manukau	4162	1080	5242	50.2	13.0	63.2
Waikato	3354	440	3794	63.8	8.4	72.1
Lakes	1596	191	1787	57.2	6.8	64.1
Bay of Plenty	797	172	969	57.2	12.3	69.6
Tairawhiti	290	63	353	41.7	9.1	50.7
Taranaki	718	206	924	47.3	13.6	60.9
Hawke's Bay	1091	134	1225	52.6	6.5	59.0
Whanganui	992	133	1125	47.5	6.4	53.8
MidCentral	353	77	430	43.2	9.4	52.6
Hutt Valley	2300	253	2553	65.1	7.2	72.3
Capital and Coast	1095	173	1268	59.0	9.3	68.3
Wairarapa	293	40	333	61.8	8.4	70.3
Nelson Marlborough	1101	138	1239	77.4	9.7	87.1
West Coast	237	38	275	67.7	10.9	78.6
Canterbury	4307	558	4865	71.6	9.3	80.9
South Canterbury	447	40	487	68.3	6.1	74.5
Southern	2223	230	2453	67.7	7.0	74.7
Total	**36,206**	**5450**	**41,656**	**61.6**	**9.3**	**70.9**

As for screens commenced, most DHBs showed a trend of increasing rates of screening completion over the four years covered in this report. South Canterbury was an exception to this with decreased completion rates, particularly between 2013 and 2014. Several other DHBs (Northland, Waitemata, Hawke's Bay, Whanganui and Canterbury) showed a levelling off of completion rates between 2013 and 2014 (see Table 8).

Table 8: Screening completion by DHB, January 2011 to December 2014

DHB	Screens completed (per 100 births)			
	2011	2012	2013	2014
Northland	41.2	44.5	47.1	47.9
Waitemata	77.9	77.8	82.1	81.0
Auckland	70.4	69.4	77.6	78.8
Counties Manukau	53.8	57.3	59.7	63.2
Waikato	65.1	64.2	69.1	72.1
Lakes	58.2	61.7	61.9	64.1
Bay of Plenty	53.1	59.0	62.6	69.6
Tairawhiti	39.2	44.6	46.8	50.7
Taranaki	58.2	55.6	55.0	60.9
Hawke's Bay	50.2	55.8	59.7	59.0
Whanganui	45.3	49.5	53.8	53.8
MidCentral	40.2	41.8	45.1	52.6
Hutt Valley	67.8	71.8	70.9	72.3
Capital and Coast	59.1	62.6	64.6	68.3
Wairarapa	62.8	59.6	66.5	70.3
Nelson Marlborough	78.7	81.3	77.9	87.1
West Coast	55.6	68.8	73.1	78.6
Canterbury	72.3	75.8	81.9	80.9
South Canterbury	86.9	82.6	85.5	74.5
Southern	67.3	73.7	75.6	74.7
Total	**63.4**	**65.7**	**69.3**	**70.9**

Screens completed by age, ethnicity and deprivation

Table 9 provides an overall view of screens completed by age, ethnicity and NZ deprivation quintile for January 2011 to December 2014, with similar trends shown as for screening commencement. Screening completion rates were highest in the 30–34 year age group with 78 women completing screening per 100 births in 2014. This was followed by the 25–39 years age group with 74 per 100 births (see Figure 9).

Screening completion rates were highest among women of Other ethnicity at 89 per 100 births for 2014. This was followed closely by Asian women at 87. The rate of completed screens for Pacific and Māori women remains lower at 42 per 100 births and 37 per 100 births respectively (see Figure 10). However, both groups have shown significant increase in screening completion over the four years (see Table 9).

Screening completion rates were highest among women in less deprived areas with rates around 85 per 100 per births for quintiles 1 and 2 in 2014 compared with 53 per 100 births for quintile 5 (see Figure 11).

Table 9: Screens completed by age of mother, ethnicity and NZ deprivation quintile, January 2011 to December 2014

	Number of screens completed				Screens completed (per 100 births)[#]			
	2011	**2012**	**2013**	**2014**	**2011**	**2012**	**2013**	**2014**
Age at screen								
Under 20 years	1808	1699	1610	1600	44.6	43.5	48.4	53.4
20–24 years	5754	5890	6010	6059	49.2	51.4	55.6	58.8
25–29 years	10,276	10,997	11,097	11,665	66.1	69.0	72.6	74.3
30–34 years	12,353	12,859	13,089	13,645	71.7	73.7	78.1	77.5
35–39 years	7453	7543	7214	7132	69.5	72.5	71.8	73.6
40–44 years	1474	1588	1643	1483	61.3	61.6	67.5	63.2
45 years and over	63	72	70	72	50.4	59.5	49.0	54.5
Ethnicity								
Māori	4561	4880	4893	5170	28.9	31.2	33.8	36.5
Pacific	2479	2591	2606	2596	35.1	37.7	41.1	42.2
Asian	6024	6990	7091	8021	84.4	82.7	86.9	87.1
Other	26,117	26,187	26,143	25,869	82.1	84.7	87.7	88.5
NZ Deprivation Quintile								
Quintile 1	7519	7520	7255	7236	88.4	86.7	88.7	85.4
Quintile 2	7480	7805	7749	7850	78.6	81.2	83.7	85.6
Quintile 3	7748	8028	8102	8181	69.5	71.9	76.2	77.4
Quintile 4	8401	8851	9001	9299	60.8	64.8	67.1	69.9
Quintile 5	8027	8441	8622	9089	42.7	45.0	49.8	52.7
Unknown	6	3	4	1	–	–	–	–
Total	**39,181**	**40,648**	**40,733**	**41,656**	**63.4**	**65.7**	**69.3**	**70.9**

Rate suppressed if the number of screens was <10.

Figure 9: Screens completed by age of mother at screen, January 2014 to December 2014

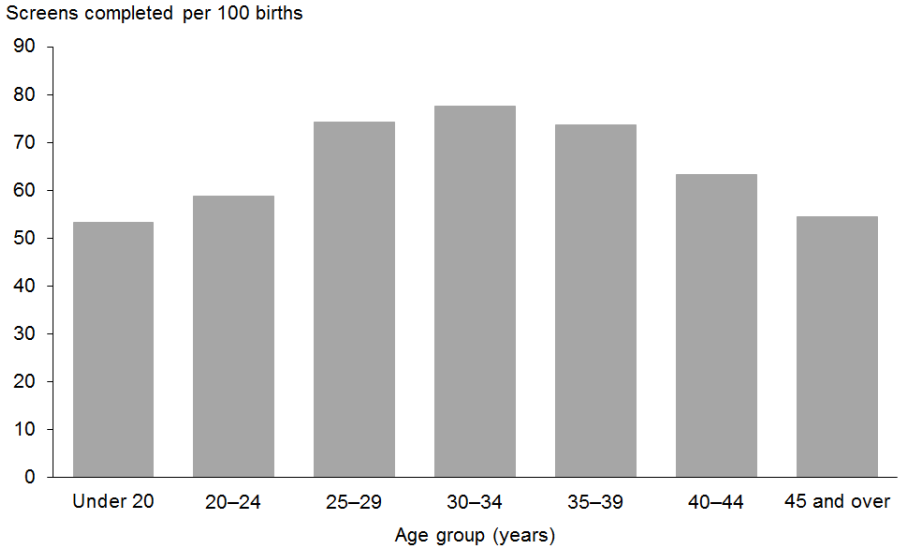


Figure 10: Screens completed by ethnicity of mother, January 2014 to December 2014

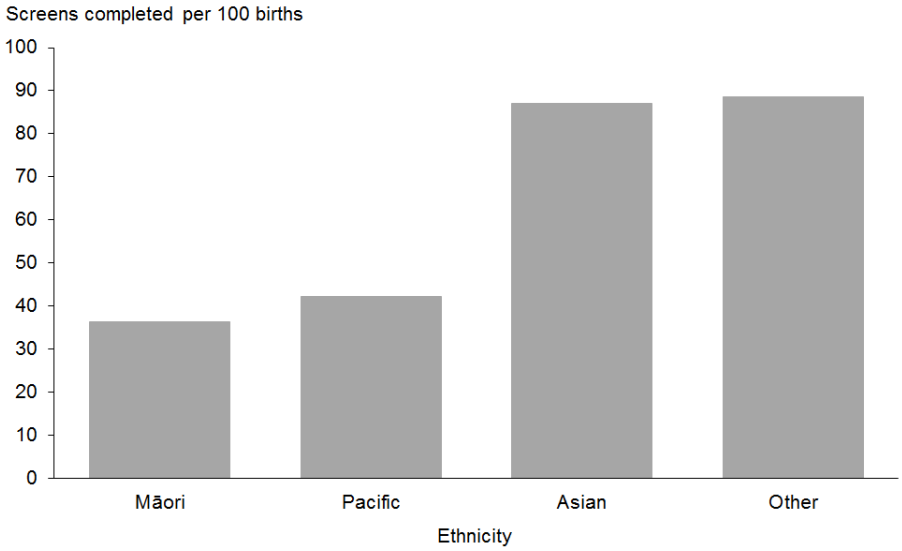
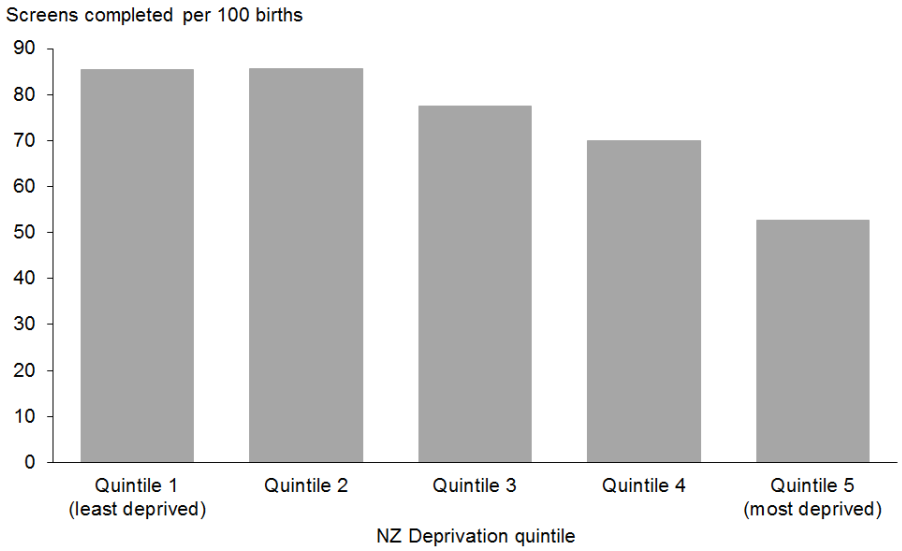


Figure 11: Screens completed by NZ deprivation quintile of mother, January 2014 to December 2014



Indicator 3: Screening pathway variance

This section reports on the number of screens completed in the second trimester which included first trimester screening inputs. First trimester combined screening requires a blood sample (PAPP-A and ßhCG) and ultrasound scan measurements of NT and CRL. Without both items a risk is not calculated and a second trimester blood sample is recommended. Information (NT or PAPP-A) from the first trimester will be included in the second trimester risk assessment.

Second trimester results with an NT measurement indicate that the screening laboratory did not receive a first trimester blood sample, or the blood sample was taken outside the accepted timeframe for first trimester screening. Second trimester results with PAPP-A indicate that the screening laboratory did not receive the NT scan report, or that the scan was performed outside the accepted timeframe for first trimester screening.

Screening pathway variance by year

Table 10 shows total number of second trimester screening results that included first trimester inputs over the period from 2011 to 2014. This has been broken down by the type of pathway variance.

The proportion of trimester 2 screens with an NT measurement has increased of the four year period from 41% to 44%. The proportion with PAPP-A increased slightly from 6% to 7% between 2012 and 2013.

Table 10: Screening pathway variance by type, January 2011 to 31 December 2014

Year	Second trimester screening results				
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
	Number			Percentage	
2011	4446	1811	264	40.7	5.9
2012	4957	2048	291	41.3	5.9
2013	5269	2219	361	42.1	6.9
2014	5450	2378	376	43.6	6.9

Screening pathway variance by DHB

Table 11 shows a breakdown of screening pathway variance by DHB and type of variance for the 2014 year. Many DHBs did not have sufficient numbers to calculate the proportion with PAPP-A. Generally, the overall results are reflected at DHB level with a far higher proportion of T2 screens with NT compared with those with PAPP-A. Taranaki was an exception to this, with a higher proportion of T2 screens with PAPP-A (23%) than with NT (17%).

Table 11: Screening pathway variance by DHB, January 2014 to December 2014

DHB	Second trimester screening results				
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A#
	Number			Percentage	
Northland	155	64	9	41.3	–
Waitemata	704	322	48	45.7	6.8
Auckland	625	241	55	38.6	8.8
Counties Manukau	1080	358	65	33.1	6.0
Waikato	440	197	26	44.8	5.9
Bay of Plenty	191	102	9	53.4	–
Lakes	172	75	7	43.6	–
Tairawhiti	63	28	4	44.4	–
Taranaki	206	34	48	16.5	23.3
Hawke's Bay	134	69	4	51.5	–
MidCentral	133	50	13	37.6	9.8
Whanganui	77	43	1	55.8	–
Capital and Coast	253	120	22	47.4	8.7
Hutt Valley	173	96	12	55.5	6.9
Wairarapa	40	24	–	60.0	–
Nelson Marlborough	138	88	2	63.8	–
West Coast	38	23	1	60.5	–
Canterbury	558	300	41	53.8	7.3
South Canterbury	40	15	1	37.5	–
Southern	230	129	8	56.1	–
Total	**5450**	**2378**	**376**	**43.6**	**6.9**

Rate suppressed if the number of screens was <10.

Screening pathway variance by age, ethnicity and deprivation

Table 12 shows a breakdown of screening pathway variance by age, ethnicity and NZ deprivation quintile for the 2014 year. The results show higher proportions for pathway variance for older age groups, for women of Other ethnicity, and women in areas of lower deprivation.

Table 12: Screening pathway variance by age, ethnicity and NZ deprivation quintile, January 2011 to December 2014

	Second trimester screening results				
	Total T2 screens	with NT	with PAPP-A	with NT#	with PAPP-A#
	Number			Percentage	
Age at screen					
Under 20 years	467	181	22	38.8	4.7
20–24 years	1185	509	52	43.0	4.4
25–29 years	1538	683	111	44.4	7.2
30–34 years	1398	659	104	47.1	7.4
35–39 years	681	283	69	41.6	10.1
40–44 years	175	62	18	35.4	10.3
45 years and over	6	1	-	–	–
Ethnicity					
Māori	1337	529	66	39.6	4.9
Pacific	905	275	61	30.4	6.7
Asian	1003	373	85	37.2	8.5
Other	2205	1201	164	54.5	7.4
NZ Deprivation quintile					
Quintile 1	553	319	46	57.7	8.3
Quintile 2	717	354	59	49.4	8.2
Quintile 3	888	423	66	47.6	7.4
Quintile 4	1305	564	97	43.2	7.4
Quintile 5	1987	718	108	36.1	5.4
Total	**5450**	**2378**	**376**	**43.6**	**6.9**

Rate suppressed if the number of screens was <10.

Indicator 4: Incomplete screens

This section reports on the number of women who commenced screening but were not issued with a risk result. Women that start screening in trimester 1 but complete screening in trimester 2 are not included in this indicator and are instead covered under indicator 3, pathway variances.

Total incomplete screens

Table 13 shows total number of incomplete screens by calendar year and trimester of screen. Nearly all incomplete screens related to the first trimester, which reflects the different components required to complete screening depending on trimester. First trimester screening requires a blood sample and an NT scan, whereas second trimester screening involves only a blood sample. The total number of incomplete screens for 2014 was 4184 which equates to 9% of screens commenced that year.

Table 13: Incomplete screens by trimester, January 2011 to 31 December 2014

Trimester of screen	Number of incomplete screens			
	2011	2012	2013	2014
T1 screen	4580	3988	3497	4024
T2 screen	252	281	228	160
Total screens	**4832**	**4269**	**3725**	**4184**

Incomplete T 1 screens by reason incomplete

Table 14 shows provides a breakdown of incomplete T1 screens according to which component of the screen was missing. Results have been reported as a percentage of all commenced screens, and then as a percentage of all incomplete screens.

The proportion of incomplete T1 screens out of all commenced T1 screens decreased from 12% in 2011 to 9% in 2013 before increasing slightly to 10% in 2014. This appears to be driven by an overall decrease in screens without blood samples (by 2% between 2011 and 2014) combined with fluctuation in the percentage without NT scans.

The split between the percentage of incompletes due to no blood or no NT scan has varied of the period covered in this report (see far right columns of Table 14), with an increasing proportion of incompletes being due to no NT scan (34% in 2014 compared with 26% in 2011).

Table 14: Incomplete T1 screens by reason incomplete, January 2011 to December 2014

Year	Commenced first trimester		Reason incomplete		Incomplete as percentage of commenced			Type as percentage of all T1 incomplete	
	Total commenced	Incomplete	No blood	No NT scan	T1 no blood	T1 no NT scan	Total T1 incompletes	T1 no blood	T1 no NT scan
2011	39,315	4580	3384	1196	8.6	3.0	11.6	73.9	26.1
2012	39,679	3,988	2892	1096	7.3	2.8	10.1	72.5	27.5
2013	38,961	3497	2368	1129	6.1	2.9	9.0	67.7	32.3
2014	40,230	4024	2657	1367	6.6	3.4	10.0	66.0	34.0

Incomplete T1 screens by reason and DHB

Table 15 provides the same breakdown by DHB. The lower numbers involved limit DHB comparisons. However, as with the pathway variance indicator, Taranaki DHB stands out with a much higher percentage of commenced screens being incomplete due to not having an NT scan (10%). Taranaki also stands out in the split of incomplete screens by type, with 73% due to no NT scan compared with the national average of 34%.

Table 15: Incomplete T1 screens by reason and DHB, 1 January 2014 to 31 December 2014

DHB	Commenced first trimester		Reason incomplete		Incomplete as percentage of commenced			Type as percentage of all T1 incomplete	
	Total commenced	Incomplete	No blood	No NT scan	T1 no blood	T1 no NT scan	Total T1 incomplete	T1 no blood	T1 no NT scan
Northland	1010	157	123	34	12.2	3.4	15.5	78.3	21.7
Waitemata	6069	416	267	149	4.4	2.5	6.9	64.2	35.8
Auckland	4673	329	182	147	3.9	3.1	7.0	55.3	44.7
Counties Manukau	4587	425	280	145	6.1	3.2	9.3	65.9	34.1
Waikato	3768	414	276	138	7.3	3.7	11.0	66.7	33.3
Bay of Plenty	1813	217	140	77	7.7	4.2	12.0	64.5	35.5
Lakes	899	102	76	26	8.5	2.9	11.3	74.5	25.5
Tairawhiti	343	53	34	19	9.9	5.5	15.5	64.2	35.8
Taranaki	827	109	29	80	3.5	9.7	13.2	26.6	73.4
Hawke's Bay	1223	132	98	34	8.0	2.8	10.8	74.2	25.8
MidCentral	1105	113	56	57	5.1	5.2	10.2	49.6	50.4
Whanganui	418	65	43	22	10.3	5.3	15.6	66.2	33.8
Capital and Coast	2575	275	196	79	7.6	3.1	10.7	71.3	28.7
Hutt Valley	1277	182	148	34	11.6	2.7	14.3	81.3	18.7
Wairarapa	343	50	36	14	10.5	4.1	14.6	72.0	28.0
Nelson Marlborough	1238	137	96	41	7.8	3.3	11.1	70.1	29.9
West Coast	270	33	24	9	8.9	3.3	12.2	72.7	27.3
Canterbury	4812	505	349	156	7.3	3.2	10.5	69.1	30.9
South Canterbury	472	25	13	12	2.8	2.5	5.3	52.0	48.0
Southern	2508	285	191	94	7.6	3.7	11.4	67.0	33.0
Total	**40,230**	**4024**	**2657**	**1367**	**6.6**	**3.4**	**10.0**	**66.0**	**34.0**

Incomplete T1 screens by age, ethnicity and deprivation

Table 16 shows a breakdown of incomplete screens by reason incomplete, age, ethnicity, and NZ deprivation quintile for the 2014 year. This shows higher rates of incomplete screens for younger women (25% for women up to 29 years of age). There were higher rates of incomplete screens for Māori (22%) and Pacific (18%) women when compared with Asian (5%) and Other (8%). The rate of incomplete screens also increased with increasing deprivation (15% for quintile 5 compared with 7% for quintile 1).

Table 16: Incomplete T1 screens by age, ethnicity and NZ deprivation quintile, 1 January 2014 to 31 December 2014

	Commenced T1 screens		Reason incomplete		Incomplete as percentage of commenced			Type as percentage of all T1 incomplete[#]	
	Total commenced	Incomplete	No blood	No NT scan	No blood	No NT scan	All T1 incomplete	No blood	No NT scan
Age at screen									
Under 20 years	1501	368	274	94	18.3	6.3	24.5	74.5	25.5
20 – 24 years	5851	977	731	246	12.5	4.2	16.7	74.8	25.2
25 – 29 years	11,221	1094	774	320	6.9	2.9	9.7	70.7	29.3
30 – 34 years	13,206	959	585	374	4.4	2.8	7.3	61.0	39.0
35 – 39 years	6931	480	242	238	3.5	3.4	6.9	50.4	49.6
40 – 44 years	1445	137	49	88	3.4	6.1	9.5	35.8	64.2
45 years and over	75	9	2	7	–	–	–	–	–
Ethnicity									
Māori	4905	1072	775	297	15.8	6.1	21.9	72.3	27.7
Pacific	2059	368	238	130	11.6	6.3	17.9	64.7	35.3
Asian	7419	401	212	189	2.9	2.5	5.4	52.9	47.1
Other	25,847	2183	1432	751	5.5	2.9	8.4	65.6	34.4
NZ Deprivation quintile									
Quintile 1	7199	516	322	194	4.5	2.7	7.2	62.4	37.6
Quintile 2	7687	554	335	219	4.4	2.8	7.2	60.5	39.5
Quintile 3	7986	693	437	256	5.5	3.2	8.7	63.1	36.9
Quintile 4	9000	1006	686	320	7.6	3.6	11.2	68.2	31.8
Quintile 5	8357	1255	877	378	10.5	4.5	15.0	69.9	30.1
Total	**40,230**	**4024**	**2657**	**1367**	**6.6**	**3.4**	**10.0**	**66.0**	**34.0**

Suppressed if the number of incomplete screens was <10.

Incomplete T2 screens

T2 screens do not require an NT scan, just a blood sample, making it less likely that a screen commenced in the second trimester will be incomplete. For the 2014 year 3% of T2 commenced screens were incomplete, compared with 10% of T2 commenced screens. As Table 17 shows, the percentage of incomplete T2 screens has decreased from 5% in 2011 to 3% in 2014.

Table 17: Incomplete T2 screens, 1 January 2011 to 31 December 2014

Year	Commenced second trimester	No result issued	Percentage incomplete
2011	4698	252	5.4
2012	4957	281	5.7
2013	5269	228	4.3
2014	5450	160	2.9

Incomplete T2 screens by DHB

Table 18 shows a breakdown of incomplete T2 screens by DHB for the 2014 year. The very low numbers involved limit meaningful percentage calculations and DHB comparisons.

Table 18: IncompleteT2 screens by DHB, 1 January 2011 to 31 December 2014

DHB	Commenced second trimester	No result issued	Percentage incomplete[#]
Northland	160	5	–
Waitemata	722	18	2.5
Auckland	643	18	2.8
Counties Manukau	1125	45	4.0
Waikato	455	15	3.3
Bay of Plenty	196	5	–
Lakes	178	6	–
Tairawhiti	64	1	–
Taranaki	210	4	–
Hawke's Bay	141	7	–
MidCentral	134	1	–
Whanganui	77	–	–
Capital and Coast	257	4	–
Hutt Valley	173	–	–
Wairarapa	42	2	–
Nelson Marlborough	144	6	–
West Coast	38	–	–
Canterbury	575	17	3.0
South Canterbury	40	–	–
Southern	236	6	–
Total	**5610**	**160**	**2.9**

Suppressed if the number of incomplete screens was <10.

Incomplete T2 screens by age, ethnicity and deprivation

Table 19 shows a breakdown of incomplete T2 screens by age, ethnicity and NZ deprivation quintile for 2014. Once again, the numbers involved are low. However, the percentage incomplete was higher for the youngest age group, and higher for Pacific compared with women of other ethnicities. There was no trend by NZ deprivation quintile.

Table 19: Incomplete T2 screens by age, ethnicity and NZ deprivation quintile, 1 January 2014 to 31 December 2014

	Commenced second trimester	No result issued	Percentage incomplete[#]
Age at screen			
Under 20 years	488	21	4.3
20–24 years	1215	30	2.5
25–29 years	1583	45	2.8
30–34 years	1435	37	2.6
35–39 years	700	19	2.7
40–44 years	183	8	–
45 years and over	6	-	–
Ethnicity			
Māori	1389	52	3.7
Pacific	953	48	5.0
Asian	1023	20	2.0
Other	2245	40	1.8
NZ Deprivation quintile			
Quintile 1	565	12	2.1
Quintile 2	728	11	1.5
Quintile 3	913	25	2.7
Quintile 4	1345	40	3.0
Quintile 5	2059	72	3.5
Total	**5610**	**160**	**2.9**

Suppressed if the number of incomplete screens was <10.

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

This indicator 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. This means that an individual woman may be at increased risk for more than one trisomy.

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

Table 20 shows total number of screening risk results that were classified as increased risk for one or more of trisomy 21, 18 or 13 by calendar year, together with the number of increased risk results per 100 screens (positive test rate). For the 2014 year, 2.8 increased risk results were issued for every 100 screens completed. This was slightly higher than 2013 but consistent with the rate for 2011 and 2012.

Table 20: Number and rate per 100 screens of increased risk screening results for trisomy 21, 18 or 13, January 2011 to 31 December 2014

	Number and rate of increased risk screens			
	2011	**2012**	**2013**	**2014**
Total increased risk results	1099	1156	1103	1155
Positive test rate per 100 screens	2.8	2.8	2.7	2.8

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

Table 21 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 2014 year.

Positive test rate increases markedly with increasing age and is also higher for Pacific and Asian women compared with other ethnicities. Older women are more likely to have a positive test and are also more likely to have a higher detection rate. This is in keeping with the inclusion of prior risk (age) as part of the risk calculation. Different levels of deprivation do not appear to affect the positive test rate.

Table 21: Increased risk screening results for trisomy 21, 18 or 13 by age, ethnicity and deprivation, January 2014 to December 2014

	Number of increased risks for trisomy 21, 18 or 13	Total number of completed screens	Positive test rate per 100 screens
Age at screen			
Under 20 years	18	1600	1.1
20–24 years	76	6059	1.3
25–29 years	149	11,665	1.3
30–34 years	245	13,645	1.8
35–39 years	385	7132	5.4
40–44 years	258	1483	17.4
45 years and over	24	72	33.3
Ethnicity			
Māori	137	5170	2.6
Pacific	97	2596	3.7
Asian	279	8021	3.5
Other	642	25,869	2.5
NZ Deprivation quintile			
Quintile 1	231	7236	3.2
Quintile 2	190	7850	2.4
Quintile 3	238	8181	2.9
Quintile 4	242	9299	2.6
Quintile 5	254	9089	2.8
Unknown	–	1	–

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

Table 22 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 calendar year.

Trisomy 18 and 13 each showed low positive test rates (from 0.3 per 100 screens) while the positive test rate for trisomy 21 was close to 3 per 100 screens for all years. The second trimester positive test rate for trisomy 21 was significantly higher than the first trimester positive test rate (approximately twice as high in all years). This may be due to variability in nuchal translucency scanning accuracy.

The positive test rate for any one or more of trisomy 21, 18 or 13 was similar to that of trisomy 21 alone. This reflects the far higher number of trisomy 21 increased risks compared with trisomy 18 and 13.

Table 22: Increased risk screening results for trisomy 21, 18 and 13 by trimester of screen, January 2011 to December 2014

Year	Total increased risks for specified trisomy	Positive test rate per 100 screens	T1 results with increased risk for specified trisomy	Positive test rate per 100 T1 screens	T2 results with increased risk for specified trisomy	Positive test rate per 100 T2 screens
Trisomy 21						
2011	1081	2.8	868	2.5	213	4.8
2012	1144	2.8	871	2.4	273	5.5
2013	1081	2.7	840	2.4	241	4.6
2014	1129	2.7	868	2.4	261	4.8
Trisomy 18						
2011	134	0.3	123	0.4	11	0.2
2012	161	0.4	149	0.4	12	0.2
2013	145	0.4	125	0.4	20	0.4
2014	135	0.3	119	0.3	16	0.3
Trisomy 13						
2011	143	0.4	140	0.4	3	0.1
2012	169	0.4	161	0.5	8	0.2
2013	158	0.4	144	0.4	14	0.3
2014	148	0.4	134	0.4	14	0.3
Any one or more of trisomy 21, 18 or 13[3]						
2011	1099	2.8	878	2.5	221	5.0
2012	1156	2.8	874	2.4	282	5.7
2013	1103	2.7	847	2.4	256	4.9
2014	1155	2.8	881	2.4	274	5.0

[3] The sum of the values for trisomy 21, 18 and 13 separately is greater than the value for the fourth grouping (any trisomy) because a result can be at increased risk for more than one trisomy.

Increased risk screening results stratified by risk level

Table 23 shows the number of increased risk results stratified by risk level for each of trisomy 21, 18 and 13 for the 2014 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 Table 23 will be greater than the total number of increased risk results for 2014.

Table 23: Increased risk screening results for trisomy 21, 18 and 13 by risk level, January 2014 to December 2014

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 – 1:20	247	44	51
1:25 to 1:50	179	14	25
1:55 to 1:300	703	77	72

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

This indicator reports information on the number and proportion of women who complete prenatal diagnostic testing (CVS or amniocentesis) 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 CVS) 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 24 shows the diagnostic testing rate from 2011–2014 by trimester of screen. In 2014, for every 100 women that received an increased risk result after a first trimester screen, 61 women had a diagnostic test. This is lower than previous years. The diagnostic testing rate was lower for women who received an increased risk after a second trimester screen (47 women per 100 increased risk screens) compared with first trimester screens. See Appendix 3 for a summary of diagnostic test results for women who had increased risk screen in 2014, as well as pregnancy outcomes (where known) for women that did not have a prenatal diagnostic.

Table 24: Diagnostic testing volumes for women with increased risk screens by trimester of screen, January 2011 to December 2014

Trimester of screen	Diagnostic tests per 100 increased risk screens			
	2011	2012	2013	2014
T1 screen	64.4	65.6	66.2	60.5
T2 screen	41.9	42.7	48.5	46.6
Total screens	**59.7**	**59.8**	**62.0**	**57.1**

Diagnostic testing volumes for women with increased risk screens by DHB

The rate of diagnostic testing for women with increased risk screens in 2014 varied across DHBs from 48 per 100 increased risk results in Taranaki, to 78.3 per 100 increased risk results in Nelson Marlborough. Bay of Plenty was next highest with 63.6 per 100 increased risks (see Table 25).

Table 25: Diagnostic testing volumes for women with increased risk screens by DHB, January 2011 to December 2014

DHB	Number of diagnostic tests				Tests per 100 increased risk screens[#]			
	2011	2012	2013	2014	2011	2012	2013	2014
Northland	24	13	28	26	49.0	38.2	56.0	59.1
Waitemata	136	137	140	115	67.0	67.2	72.9	61.2
Auckland	117	117	89	89	72.2	68.4	67.4	55.3
Counties Manukau	67	75	72	76	54.5	50.7	46.5	50.3
Waikato	15	26	40	40	20.5	38.2	57.1	63.5
Bay of Plenty	11	22	21	21	36.7	68.8	55.3	63.6
Lakes	15	23	21	21	55.6	69.7	67.7	53.8
Tairawhiti	5	5	2	–	–	–	–	–
Taranaki	14	18	18	12	63.6	75.0	66.7	48.0
Hawke's Bay	22	17	21	19	62.9	47.2	53.8	55.9
MidCentral	20	20	10	11	54.1	62.5	38.5	57.9
Whanganui	4	4	6	3	–	–	–	–
Capital and Coast	52	61	55	45	72.2	69.3	75.3	60.0
Hutt Valley	14	23	18	15	56.0	60.5	58.1	55.6
Wairarapa	5	7	9	1	–	–	–	–
Nelson Marlborough	23	11	17	18	67.6	47.8	89.5	78.3
West Coast	n/a	n/a	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	n/a	n/a
South Canterbury	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a
Southern	33	39	32	28	66.0	53.4	61.5	57.1
Total	**577**	**618**	**599**	**540**	**59.7**	**59.8**	**62.0**	**57.1**

Rate suppressed if the number of diagnostic tests was <10.

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

Table 26 shows the diagnostic testing rate for women with increased risk screens by age, ethnicity and NZ deprivation quintile for 2011 to 2014. The diagnostic testing rate ranged from 52 per 100 increased risk screens for women aged 20 to 24 years, to 66 per 100 for women aged 30–34 years.

Diagnostic testing rates were highest for women of Asian ethnicity (67 per 100 increased risks), followed by Other (61 per 100 increased risks). While diagnostic testing rates are generally and have historically been higher in less deprived areas, 2014 suggests a change in this trend with a smaller difference in rates between quintile 5 and quintile 1 when compared with previous years.

Table 26: Diagnostic testing volumes for women with increased risk screening results by age at screen, ethnicity and deprivation, January 2011 to December 2014

	Diagnostic tests per 100 increased risk screens[#]			
	2011	**2012**	**2013**	**2014**
Age at screen				
Under 20 years	–	–	–	–
20–24 years	56.1	53.1	64.8	51.6
25–29 years	60.4	61.9	62.1	61.8
30–34 years	64.6	68.4	69.4	65.7
35–39 years	65.1	59.9	62.0	54.9
40–44 years	48.9	55.6	57.8	54.8
45 years and over	–	–	44.0	–
Ethnicity				
Māori	40.4	43.2	52.6	38.1
Pacific	35.6	37.0	37.9	37.1
Asian	70.7	72.1	70.2	66.4
Other	64.2	63.7	66.3	61.0
NZ Deprivation quintile				
Quintile 1	71.6	67.5	72.6	62.4
Quintile 2	70.4	71.6	66.9	61.6
Quintile 3	60.2	63.2	62.0	55.1
Quintile 4	52.5	52.1	59.4	60.6
Quintile 5	47.1	47.6	53.7	49.4

Rate suppressed if the number of diagnostic tests was <10.

Diagnostic testing volumes for women with increased risk screening results stratified by risk level

Each 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 27 shows the number of diagnostic tests for women with increased risk screening results during 2014 for one or more of trisomy 21, 18 or 13, stratified by risk level. Uptake of diagnostic testing was higher in the very increased risk groupings. While 51% of women with a risk between 1:55 and 1:300 had a prenatal diagnostic test, this increased to 67–68% for women with risks of 1:50 or above.

Table 27: Diagnostic testing volumes for women with increased risk screens by risk level, January 2014 to December 2014

Risk level	Number of diagnostic tests	Number of increased risk screens	Tests per 100 increased risk screens
1:5 to 1:20	135	198	68.2
1:25 to 1:50	97	144	67.4
1:55 to 1:300	308	604	51.0

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 prenatal diagnostic testing (CVS or amniocentesis) 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 each of trisomy 21, 18 and 13 but increased risk for neural tube defects then the woman was categorised as at increased risk for the purposes of this indicator.

Some women with low risk screening results may have other indications for diagnostic testing, eg, 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 0.63 per 100 low risk screens in 2014. This was a decrease from the previous three years (see Table 28). This suggests that a diminishing number of women (now well under 1%) are having prenatal diagnostic tests after low risk screens.

Table 28: Diagnostic testing volumes for women with low risk screens by trimester of screen, January 2011 to December 2014

Trimester of screen	Diagnostic tests per 100 low risk screens			
	2011	2012	2013	2014
T1 screen	0.89	0.90	0.80	0.65
T2 screen	0.65	0.61	0.39	0.51
Total screens	**0.86**	**0.86**	**0.75**	**0.63**

Diagnostic testing volumes for women with low risk screens by DHB

The rate of diagnostic testing for women with low risk screens during 2014 varied across DHBs, as shown in Table 29. Given the low numbers involved, caution should be taken in making comparisons.

Table 29: Total diagnostic testing volumes for women with low risk screens by DHB January 2011 to December 2014

DHB	Number of diagnostic tests				Tests per 100 low risk screens[#]			
	2011	2012	2013	2014	2011	2012	2013	2014
Northland	5	2	7	–	–	–	–	–
Waitemata	62	60	55	34	1.04	1.00	0.90	0.55
Auckland	71	71	54	38	1.60	1.58	1.15	0.79
Counties Manukau	38	25	27	18	0.83	0.51	0.57	0.35
Waikato	5	18	18	28	–	0.52	0.51	0.75
Bay of Plenty	5	10	9	14	–	0.56	–	0.80
Lakes	3	3	3	5	–	–	–	–
Tairawhiti	–	3	–	1	–	–	–	–
Taranaki	6	11	9	3	–	1.31	–	–
Hawke's Bay	11	8	5	7	1.00	–	–	–
MidCentral	7	4	9	8	–	–	–	–
Whanganui	4	4	2	2	–	–	–	–
Capital and Coast	23	18	21	14	0.90	0.67	0.84	0.57
Hutt Valley	12	10	8	11	1.01	0.82	–	0.89
Wairarapa	1	–	–	–	–	–	–	–
Nelson Marlborough	9	14	12	6	–	1.15	1.01	–
West Coast	n/a	n/a	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	n/a	n/a
South Canterbury	n/a	n/a	n/a	n/a	n/a	n/a	n/a	n/a
Southern	24	35	16	32	0.99	1.36	0.63	1.33
Total	**286**	**296**	**255**	**221**	**0.86**	**0.86**	**0.75**	**0.63**

Rate suppressed if the number of diagnostic tests was <10.

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

Table 30 shows the rate of diagnostic testing for women with low risk screening results by age, ethnicity and NZ deprivation quintile. The rate of diagnostic testing was higher for older age groups, for women of Other ethnicity, and for women in the lowest deprivation quintiles.

Table 30: Diagnostic tests per 100 low risk screens by age, ethnicity and NZ deprivation quintile, January 2011 to December 2014

	Diagnostic tests per 100 low risk screens#			
	2011	**2012**	**2013**	**2014**
Age at screen				
Under 20 years	–	–	–	–
20–24 years	0.31	0.26	0.31	0.37
25–29 years	0.35	0.38	0.35	0.38
30–34 years	0.53	0.66	0.54	0.48
35–39 years	1.92	1.56	1.20	0.95
40–44 years	5.49	5.66	5.90	4.17
45 years and over	–	–	–	–
Ethnicity				
Māori	0.44	0.69	0.57	0.47
Pacific	0.35	0.21	0.30	0.25
Asian	0.89	0.79	0.67	0.55
Other	0.99	0.99	0.86	0.73
NZ Deprivation quintile				
Quintile 1	1.56	1.71	1.15	0.85
Quintile 2	1.05	0.98	0.77	0.76
Quintile 3	0.82	0.63	0.81	0.62
Quintile 4	0.67	0.79	0.62	0.56
Quintile 5	0.37	0.39	0.50	0.45

Rate suppressed if the number of diagnostic tests was <10.

Diagnostic testing volumes for women with low risk screening results stratified by risk

Table 31 shows the rate of diagnostic testing for women with low risk screening results, stratified by risk level. Given the low numbers involved for some risk categories, numbers have been aggregated for all years. The rate of diagnostic testing was more than 10 times higher for the highest category compared with the lowest category and the rate of diagnostic testing drops away as risk decreases below 1:1000.

Table 31: Diagnostic tests per 100 low risk screens stratified by risk level, January 2011–December 2014 aggregated

Risk level	Number of diagnostic tests	Number of low risk screens	Tests per 100 low risk screens
1:301 to 1:500	137	2180	6.28
1:501 to 1:1000	187	5648	3.31
1:1001 to 1:2000	171	9813	1.74
1:2001 to 1:3000	103	8441	1.22
1:3001 to 1:4000	60	7714	0.78
1:4001 to 1:5000	56	6834	0.82
1:5001 to 1:10,000	136	27,316	0.50
1:10,001 to 1:100,000	208	68,542	0.30

Indicator 8: Diagnostic testing for unscreened women

This section reports information on the number of women who complete prenatal diagnostic testing (CVS or amniocentesis) but were not screened in the 105 days prior to the diagnostic test. The indication for diagnostic testing is not reliably reported on laboratory request forms but it is likely that many of these women will have had a predetermined risk (eg, family history or previous child with Down syndrome) or an abnormal ultrasound finding.

Diagnostic volumes for unscreened women

During 2014, 221 diagnostic tests were completed for unscreened women. This is up from 208 in 2013 but similar to 2012. Table 32 shows the number of tests by DHB and Table 33 shows the breakdown by age, ethnicity and NZ deprivation quintile. Due to the low numbers involved, rates per 100 births are not shown.

Table 32: Diagnostic testing volumes for unscreened women by DHB, January 2012 to December 2014

DHB	Number of diagnostic tests		
	2012	2013	2014
Northland	10	7	8
Waitemata	37	25	27
Auckland	32	26	32
Counties Manukau	18	28	25
Waikato	16	24	22
Bay of Plenty	2	5	7
Lakes	10	19	15
Tairawhiti	5	-	2
Taranaki	13	12	5
Hawke's Bay	12	6	7
Mid Central	4	3	3
Whanganui	9	11	11
Capital and Coast	10	11	11
Hutt Valley	17	16	31
Wairarapa	5	1	1
Nelson Marlborough	6	1	4
West Coast	n/a	n/a	n/a
Canterbury	n/a	n/a	n/a
South Canterbury	n/a	n/a	n/a
Southern	13	13	10
Total	**219**	**208**	**221**

Table 33: Total diagnostic testing volumes for unscreened women by age, ethnicity and deprivation quintile, January 2012 to December 2014

	Number of diagnostic tests		
	2012	2013	2014
Age			
Under 20 years	13	11	13
20–24 years	27	34	30
25–29 years	37	33	36
30–34 years	56	49	57
35–39 years	49	40	52
40–44 years	36	37	31
45 years and over	1	4	2
Ethnicity			
Māori	28	47	31
Pacific	15	16	22
Asian	37	29	30
Other	139	116	138
NZ Deprivation quintile			
Quintile 1	52	31	41
Quintile 2	38	39	31
Quintile 3	39	34	49
Quintile 4	49	56	45
Quintile 5	41	48	55

Diagnostic results for unscreened women

A breakdown of prenatal diagnostic testing results for unscreened women for the 2014 year is given in Table 34. Of the 221 diagnostic tests in 2014 for unscreened women, 168 (76%) had a normal karyotype. There were 12 trisomy 21 diagnoses, nine trisomy 18 diagnoses and one diagnosis of trisomy 13.

Table 34: Total diagnostic testing results for unscreened women, January 2014 to December 2014

Karyotype result	Number	Percentage
Normal karyotype	168	76.0%
Trisomy 21	12	5.4%
Trisomy 18	9	4.1%
Trisomy 13	1	0.5%
Turner syndrome	3	1.4%
Triploidy	4	1.8%
Other chromosome abnormality	20	9.0%
Failed test	4	1.8%
Total	**221**	**100.0%**

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

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 2014 was 0.10, which was lower than previous years (see Table 35). A value of 0.10 means that if a woman receives an increased risk result for trisomy 21, 18 or 13 there is a 10% probability that she is carrying a fetus with one of these trisomies. When data was aggregated across all years the PPV value for second trimester screens was 0.04 compared with 0.14 for first trimester screens.

Table 35: Positive predictive value of screening for trisomy 21, 18 or 13, January 2011 to December 2014

Trimester of screen	True positives				False positives				Positive predictive value#			
	Positive diagnostic test/infant diagnosis after increased risk screen				Negative diagnostic test/infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	104	111	109	92	660	662	628	620	0.14	0.14	0.15	0.13
T2 screens	6	10	12	6	197	250	217	228	–	0.04	0.05	–
Total screens	**110**	**121**	**121**	**98**	**857**	**912**	**845**	**848**	**0.11**	**0.12**	**0.13**	**0.10**

Rate suppressed if the number of diagnostic tests was <10.

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21 the PPV for 2014 was lower than the combined PPV at 0.08 (see Table 36). 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.

Table 36: Positive predictive of screening for trisomy 21, January 2011 to December 2014

Trimester of screen	True positives				False positives				Positive predictive value#			
	Positive diagnostic test/infant diagnosis after increased risk screen				Negative diagnostic test/infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	70	76	82	68	687	695	650	634	0.09	0.10	0.11	0.10
T2 screens	3	7	12	5	193	244	202	218	–	–	0.06	–
Total screens	**73**	**83**	**94**	**73**	**880**	**939**	**852**	**852**	**0.08**	**0.08**	**0.10**	**0.08**

Rate suppressed if the number of positive diagnoses was <10.

Trisomies 13 and 18 involve small numbers and have similar risk profiles so combined results for PPV and remaining indicators have been calculated for these trisomies.

The combined PPV for trisomies 13 or 18 for 2014 was higher than the trisomy 21 PPV at 0.16 (see Table 37). However, the number of positive diagnoses for these two trisomies is low so caution should be taken when interpreting these results.

Table 37: Positive predictive of screening for trisomy 13 or 18, January 2011 to December 2014

Trimester of screen	True positives				False positives				Positive predictive value#			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Negative diagnostic test/ infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	33	29	25	22	98	118	101	97	0.25	0.20	0.20	0.18
T2 screens	1	2	–	–	12	15	24	19	–	–	–	–
Total screens	**34**	**31**	**25**	**22**	**110**	**133**	**125**	**116**	**0.24**	**0.19**	**0.17**	**0.16**

Rate suppressed if the number of positive diagnoses was <10.

Positive predictive value of screening for trisomy 21 stratified by risk level

Table 38 shows PPV stratified by the risk level indicated in the screening result. For 2014, women that received a very increased risk result of 1:5 to 1:20 for trisomy 21 had a 29% probability that they were carrying a fetus with trisomy 21. There were insufficient numbers to calculate PPV for the other two categories for 2014, but looking at previous years the PPV was lower for women with increased risks of 1:25 to 1:150, and lower again for women with increased risk results of 1:55 to 1:300.

Table 38: Positive predictive of screening for trisomy 21 stratified by risk level, January 2011 to December 2014

Risk level	True positives				False positives				Positive predictive value[#]			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Negative diagnostic test/ infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
1:5 to 1:20	48	58	61	56	155	166	140	139	0.24	0.26	0.30	0.29
1:25 to 1:50	15	15	14	8	137	126	95	132	0.10	0.11	0.13	–
1:55 to 1:300	10	10	19	9	588	647	617	581	0.02	0.02	0.03	–

Rate suppressed if the number of positive diagnoses was <10.

Positive predictive value of screening for trisomy 21 by age, ethnicity and deprivation

The PPV of screening for trisomy 21 also varied by age group, as shown in Table 39. For 2014 PPV was highest for the 40–44 years age group, with insufficient numbers to calculate a rate for the youngest and oldest age groups.

Table 39: Positive predictive of screening for trisomy 21 by age, January 2011 to December 2014

Age	True positives				False positives				Positive predictive value[#]			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Negative diagnostic test/ infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
Under 20 years	–	1	–	1	9	11	6	14	–	–	–	–
20–24 years	–	1	3	4	55	48	48	56	–	–	–	–
25–29 years	5	5	5	6	84	92	89	102	–	–	–	–
30–34 years	17	14	18	12	190	210	183	191	0.08	0.06	0.09	0.06
35–39 years	32	36	36	21	317	334	290	288	0.09	0.10	0.11	0.07
40–44 years	16	24	32	29	207	224	212	183	0.07	0.10	0.13	0.14
45 years and over	3	2	–	–	18	20	24	18	–	–	–	–

Rate suppressed if the number of positive diagnoses was <10.

The number of true and false positive results by ethnicity is shown in Table 40. Aggregating data across all four years gives a PPV of 0.06 (6%) for Māori, 0.02 (2%) for Pacific, 0.05 for Asian, and 0.12 (12%) for women of Other ethnicity.

Table 40: Positive predictive of screening for trisomy 21 by ethnicity, January 2011 to December 2014

Ethnicity	True positives				False positives				Positive predictive value#			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Negative diagnostic test/ infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
Māori	7	7	9	3	95	115	103	120	–	–	–	–
Pacific	1	1	6	2	100	114	108	85	–	–	–	–
Asian	6	9	11	10	161	199	175	228	–	–	0.06	0.04
Other	59	66	68	58	524	511	466	419	0.10	0.11	0.13	0.12

Rate suppressed if the number of positive diagnoses was <10.

Table 41 shows PPV by NZ deprivation quintile. There does not appear to be any relationship between PPV and NZ deprivation quintile.

Table 41: Positive predictive of screening for trisomy 21 by NZ deprivation quintile, January 2011 to December 2014

NZ Deprivation quintile	True positives				False positives				Positive predictive value#			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Negative diagnostic test/ infant without diagnosis after increased risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
Quintile 1	16	23	24	16	159	167	151	160	0.09	0.12	0.14	0.09
Quintile 2	23	14	19	13	161	183	139	133	0.13	0.07	0.12	0.09
Quintile 3	13	24	14	12	168	184	150	182	0.07	0.12	0.09	0.06
Quintile 4	12	12	16	19	183	175	181	156	0.06	0.06	0.08	0.11
Quintile 5	9	10	21	13	209	230	231	221	–	0.04	0.08	0.06

Rate suppressed if the number of positive diagnoses was <10.

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 a trisomy, or a baby born without a 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

The overall false positive rate for trisomy 21, 18 and 13 for 2014 was 0.02 (or 2%). This means that out of all women who have a negative diagnostic or a baby without a trisomy, 2% 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 42: False positive rate for trisomy 21, 18 or 13, January 2011 to December 2014

Trimester of screen	False positives				True negatives				False positive rate			
	Negative diagnostic tests/ infant without diagnosis after increased risk screen				Negative diagnostic tests/ infant without diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	660	662	628	620	29,330	30,075	29,777	30,479	0.02	0.02	0.02	0.02
T2 screens	197	250	217	228	3742	4152	4361	4579	0.05	0.06	0.05	0.05
Total screens	**857**	**912**	**845**	**848**	**33,072**	**34,227**	**34,138**	**35,058**	**0.03**	**0.03**	**0.02**	**0.02**

The false positive rate for trisomy 21 when considered alone was similar to the overall false positive rate (see Table 43). However, the combined false positive rate for trisomy 18 and trisomy 13 is much lower (0.003 for 2014 – see Table 44).

Table 43: False positive rate for trisomy 21, January 2011 to December 2014

Trimester of screen	False positives				True negatives				False positive rate			
	Negative diagnostic tests/ infant without diagnosis after increased risk screen				Negative diagnostic tests/ infant without diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	687	695	650	634	29,346	30,087	29,792	30,499	0.02	0.02	0.02	0.02
T2 screens	193	244	202	218	3750	4163	4378	4590	0.05	0.06	0.04	0.05
Total screens	**880**	**939**	**852**	**852**	**33,096**	**34,250**	**34,170**	**35,089**	**0.03**	**0.03**	**0.02**	**0.02**

Table 44: False positive rate for trisomy 13 or 18, January 2011 to December 2014

Trimester of screen	False positives				True negatives				False positive rate			
	Negative diagnostic tests/ infant without diagnosis after increased risk screen				Negative diagnostic tests/ infant without diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	98	118	101	97	29,984	30,713	30,407	31,084	0.003	0.004	0.003	0.003
T2 screens	12	15	24	19	3931	4398	4570	4794	0.003	0.003	0.005	0.004
Total screens	**110**	**133**	**125**	**116**	**33,915**	**35,111**	**34,977**	**35,878**	**0.003**	**0.004**	**0.004**	**0.003**

False positive rate for screening for trisomy 21 by age, ethnicity and deprivation

The false positive rate for trisomy 21 increased with age. For example, in 2014 the false positive rate for women under 20 years was 0.01 (1%) compared with 0.30 (30%) for women 45 years and older (see Table 45). This difference is due to the inclusion of prior risk (age) 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 45: False positive rate for trisomy 21 by age, January 2011 to December 2014

Age	False positives				True negatives				False positive rate#			
	Negative diagnostic tests/ infant without diagnosis after increased risk screen				Negative diagnostic tests/ infant without diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
Under 20 years	9	11	6	14	1586	1455	1392	1376	–	0.01	–	0.01
20–24 years	55	48	48	56	4917	5062	5127	5174	0.01	0.01	0.01	0.01
25–29 years	84	92	89	102	8818	9417	9450	9945	0.01	0.01	0.01	0.01
30–34 years	190	210	183	191	10,534	10,902	11,055	11,610	0.02	0.02	0.02	0.02
35–39 years	317	334	290	288	6134	6218	5933	5882	0.05	0.05	0.05	0.05
40–44 years	207	224	212	183	1074	1155	1178	1060	0.16	0.16	0.15	0.15
45 years and over	18	20	24	18	33	41	35	42	0.35	0.33	0.41	0.30

Rate suppressed if false positives <10.

The false positive rate for 2014 was relatively consistent across ethnic groups. The Pacific rate, which showed a higher rate for 2011, 2012 and 2013, was consistent with other ethnic groups in 2014.

Table 46: False positive rate for trisomy 21 by ethnicity, January 2011 to December 2014

Ethnicity	False positives				True negatives				False positive rate			
	Negative diagnostic tests/ infant without diagnosis after increased risk screen				Negative diagnostic tests/ infant without diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
Māori	95	115	103	120	4078	4392	4380	4670	0.02	0.03	0.02	0.03
Pacific	100	114	108	85	2273	2349	2357	2363	0.04	0.05	0.04	0.03
Asian	161	199	175	228	5377	6179	6262	7082	0.03	0.03	0.03	0.03
Other	524	511	466	419	21,368	21,330	21,171	20,974	0.02	0.02	0.02	0.02

False positive rate was also relatively consistent by deprivation with rates between 2% and 3% for 2014 (see Table 47).

Table 47: False positive rate for trisomy 21 by NZ deprivation quintile, January 2011 to December 2014

NZ Deprivation quintile	False positives				True negatives				False positive rate			
	Negative diagnostic tests/infant without diagnosis after increased risk screen				Negative diagnostic tests/infant without diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
Quintile 1	159	167	151	160	6031	6006	5726	5770	0.03	0.03	0.03	0.03
Quintile 2	161	183	139	133	6067	6355	6256	6441	0.03	0.03	0.02	0.02
Quintile 3	168	184	150	182	6584	6804	6908	6915	0.02	0.03	0.02	0.03
Quintile 4	183	175	181	156	6906	7201	7272	7500	0.03	0.02	0.02	0.02
Quintile 5	209	230	231	221	7502	7882	8005	8462	0.03	0.03	0.03	0.03
Unknown	–	–	–	–	6	2	3	1	–	–	–	–

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, and the receiver operating characteristic (ROC) curve of detection rate against false positive rate for trisomies 21, 18 and 13 combined is contained in Appendix 6.

Detection rate for screening

The overall detection rate for trisomy 21, 18 and 13 for 2014 was 0.80 (80%). This was higher than all previous years (see Table 48). A detection rate of 0.80 means that there is an 80% probability that a woman carrying a fetus with one of trisomy 21, 18 or 13 will have an increased risk screening result for trisomy 21, 18 or 13.

Table 48: Detection rate for trisomy 21, 18 or 13, January 2011 to December 2014

Trimester of screen	True positives				False negatives				Detection rate#			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Positive diagnostic test/ infant diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	104	111	109	92	31	28	31	24	0.77	0.80	0.78	0.79
T2 screens	6	10	12	6	2	6	6	1	–	0.63	0.67	–
Total screens	**110**	**121**	**121**	**98**	**33**	**34**	**37**	**25**	**0.77**	**0.78**	**0.77**	**0.80**

Rate suppressed if the number of positive diagnoses was <10.

The detection rate for trisomy 21 alone is shown in Table 49. The rate for 2014 was slightly higher (0.83) than the overall rate for trisomy 21, 18 and 13. The detection rate for trisomy 13 and 18 was lower at 0.63 for 2014 (see Table 50).

Table 49: Detection rate for trisomy 21, January 2011 to December 2014

Trimester of screen	True positives				False negatives				Detection rate#			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Positive diagnostic test/ infant diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	70	76	82	68	22	18	21	14	0.76	0.81	0.80	0.83
T2 screens	3	7	12	5	1	4	4	1	–	–	0.75	–
Total screens	**73**	**83**	**94**	**73**	**23**	**22**	**25**	**15**	**0.76**	**0.79**	**0.79**	**0.83**

Rate suppressed if the number of positive diagnoses was <10.

Table 50: Detection rate for trisomy 13 or 18, January 2011 to December 2014

Trimester of screen	True positives				False negatives				Detection rate#			
	Positive diagnostic test/ infant diagnosis after increased risk screen				Positive diagnostic test/ infant diagnosis after low risk screen							
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
T1 screens	33	29	25	22	10	16	12	12	0.77	0.64	0.68	0.65
T2 screens	1	2	–	–	3	3	2	1	–	–	–	–
Total screens	**34**	**31**	**25**	**22**	**13**	**19**	**14**	**13**	**0.72**	**0.62**	**0.64**	**0.63**

Rate suppressed if the number of positive diagnoses was <10.

Detection rate for screening for trisomy 21 by age, ethnicity and deprivation

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 four years in order to present more stable rates. Numbers for the youngest and oldest age groups were still too low after aggregation to present a rate. Across the other age groups the detection rate for trisomy 21 appears to increase with age from 0.68 for women 25–29 years to 0.94 for women 40-44 years (see Table 51).

Table 51: Detection rate for trisomy 21 by age, January 2011 to December 2014 (aggregated)

Age	True positives	False negatives	Detection rate#
	Positive diagnostic test/ infant diagnosis after increased risk screen	Positive diagnostic test/ infant diagnosis after low risk screen	
Under 20 years	2	4	–
20–24 years	8	7	–
25–29 years	21	10	0.68
30–34 years	61	31	0.66
35–39 years	125	26	0.83
40–44 years	101	7	0.94
45 years and over	5	–	–

Rate suppressed if the number of positive diagnoses was <10.

The aggregated detection rate for Pacific women appears to be lower than for other ethnicities (see Table 52). However, low numbers mean this difference should be interpreted with caution.

Table 52: Detection rate for trisomy 21 by ethnicity, January 2011 to December 2014 (aggregated)

Ethnicity	True positives	False negatives	Detection rate
	Positive diagnostic test/ infant diagnosis after increased risk screen	Positive diagnostic test/ infant diagnosis after low risk screen	
Māori	26	9	0.74
Pacific	10	5	0.67
Asian	36	11	0.77
Other	251	60	0.81

The aggregated detection rates by deprivation quintile ranged from 0.76 to 0.84 (see Table 53). There was no clear trend with increasing deprivation.

Table 53: Detection rate for trisomy 21 by NZ deprivation quintile, January 2011 to December 2014 (aggregated)

NZ Deprivation quintile	True positives	False negatives	Detection rate
	Positive diagnostic test/ infant diagnosis after increased risk screen	Positive diagnostic test/ infant diagnosis after low risk screen	
Quintile 1	79	20	0.80
Quintile 2	69	18	0.79
Quintile 3	63	12	0.84
Quintile 4	59	19	0.76
Quintile 5	53	16	0.77

Appendix 1: Indicator definitions

Table 54: Definitions used for monitoring indicators

Indicator	Methodology
Indicator 1: Screens commenced	Numerator: number of women who start screening Denominator: number of live births and stillbirths
Indicator 2: Screens completed	Numerator: number of women who have a risk result calculated Denominator: number of live births and stillbirths
Indicator 3: Pathway variances	Numerator: completed second trimester screens that have an ultrasound or PAPP-A reading recorded against them Denominator: number of completed second trimester screens
Indicator 4: Incomplete screens	Numerator: number of screens commenced that have no risk result reported against them Denominator: number of screens commenced
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.
- If a woman has more than one screen for the same pregnancy (defined as being within 112 days) then the first completed screen has been retained for the analysis and the others excluded.
- 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[4] was obtained from the national Maternity Collection for each financial year.

Table 55: Live births and still births by district health board 2011–2014

DHB	2011	2012	2013	2014
Northland	2299	2292	2121	2105
Waitemata	7883	7973	7655	7852
Auckland	6542	6703	6243	6307
Counties Manukau	8745	8768	8166	8288
Waikato	5390	5485	5223	5259
Lakes	1588	1559	1419	1393
Bay of Plenty	2862	2967	2758	2790
Tairawhiti	748	733	710	696
Taranaki	1566	1558	1523	1518
Hawke's Bay	2257	2260	2160	2076
Whanganui	830	874	827	818
MidCentral	2297	2150	2122	2090
Hutt Valley	2054	2006	1915	1856
Capital and Coast	3861	3871	3631	3531
Wairarapa	530	510	502	474
Nelson Marlborough	1650	1531	1551	1423
West Coast	405	407	372	350
Canterbury	6064	5987	5826	6013
South Canterbury	572	648	640	654
Southern	3672	3593	3446	3286
Total	**61,815**	**61,875**	**58,810**	**58,779**

4 Births reaching at least 20 weeks gestation or ≥400 g birth weight.

Table 56: Live births and still births by age group 2011–2014

Age group	2011	2012	2013	2014
Under 20	4053	3907	3329	2998
20–24	11,703	11,466	10,802	10,296
25–29	15,553	15,936	15,277	15,707
30–34	17,231	17,447	16,768	17,596
35–39	10,727	10,407	10,044	9691
40–44	2403	2579	2434	2346
45 and over	125	121	143	132
Unknown	20	12	13	13
Total	**61,815**	**61,875**	**58,810**	**58,779**

Table 57: Live births and still births by 2013 NZ deprivation quintile, 2011–2014

NZ Dep 2013 quintile	2011	2012	2013	2014
Quintile 1	8505	8677	8177	8471
Quintile 2	9512	9615	9256	9175
Quintile 3	11,154	11,165	10,628	10,570
Quintile 4	13,807	13,657	13,418	13,299
Quintile 5	18,814	18,743	17,299	17,239
Unknown	23	18	32	25
Total	**61,815**	**61,875**	**58,810**	**58,779**

Table 58: Live births and still births by ethnicity

Ethnicity	2011	2012	2013	2014
Māori	15,787	15,637	14,495	14,181
Pacific	7069	6870	6344	6157
Asian	7138	8455	8161	9213
Other	31,821	30,913	29,810	29,228
Total	**61,815**	**61,875**	**58,810**	**58,779**

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

Summary of prenatal diagnostic testing uptake for women with increased risks for trisomy 21, 18 or 13

Of the 1151 screens that had an increased risk for trisomy 21, 18 or 13 during 2014, 946 related to women in DHBs covered by a cytogenetic lab other than CHL. Of these 946 women, 540 had a prenatal diagnostic test (CVS or Amniocentesis) and 406 did not. Table 59 shows the diagnostic testing results for the 540 prenatal tests, of which 105 had an abnormal karyotype. Table 60 shows a breakdown of pregnancy outcomes for the 406 women that had an increased risk screen but did not have a prenatal diagnostic test.

Table 59: Diagnostic results for women that accessed a prenatal diagnostic test following an increased risk screen for trisomy 21, 18 or 13 during the 2014 year

Karyotype result	Number	Percentage
Normal karyotype	435	80.56%
Confirmed Down syndrome	63	11.67%
Other result*	42	7.78%
Total	**540**	**100%**

* The 42 'Other' results were made up of the following:

Result	Number
Trisomy 18	14
Trisomy 13	5
Turner syndrome	10
Triploidy	1
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)	2
Apparently balanced chromosome rearrangement	4
Total	**42**

Table 60: Pregnancy outcomes (where known) for women that did not have a prenatal diagnostic test following an increased risk screen for trisomy 21, 18 or 13 during the 2014 year

Result	Number
No abnormality detected on postnatal diagnostic test	16
Trisomy 21	10
Trisomy 18	8
Triploidy	5
Other aneuploidy	2
No diagnosis	365
Total	**406**

Appendix 4: Measuring screening performance

Figure 12 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 12: 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.

Detection rate = A/(A+C)

Data for women screened during 2014

Figure 13 shows the data break down in relation to trisomy 21 for women screened during 2014. This data focuses on trisomy 21 and excludes Canterbury, South Canterbury and West Coast (because pregnancy outcomes for women in these areas are unknown) so the totals will not be the same as indicators 2 and 5 in this report.

Figure 13: Categorisation of trisomy 21 screening results 2014

	Trisomy 21 diagnosis	No trisomy 21 diagnosis	Total
Screen result = Increased risk	A = 73	B = 852	A + B = 925
Screen result = Low risk	C = 15	D = 35,089	C + D = 35,104
	A + C = 88	B + D = 35,941	N = 36,029 (total screens)

Positive predictive value (indicator 9)

PPV = A/(A+B)
= 73 / 925
= 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)
= 852 / 35,941
= 0.02 (or 2%)

Out of all women that ultimately have a negative diagnostic test or a baby without trisomy 21, 2% will have received an increased risk screening result.

Detection rate (indicator 11)

Detection rate = A/(A+C)
= 73 / 88
= 0.83 (or 83%)

There is an 83% 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 130 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 61 shows the number of false negatives for each of the four calendar 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 made up 0.1% of all negative screens for each of the years from 2011 to 2013. The false negative rate for 2014 was lower at 0.07%.

Table 61: False negative screens for trisomy 21, 18 and 13 by risk level, January 2011 to December 2014

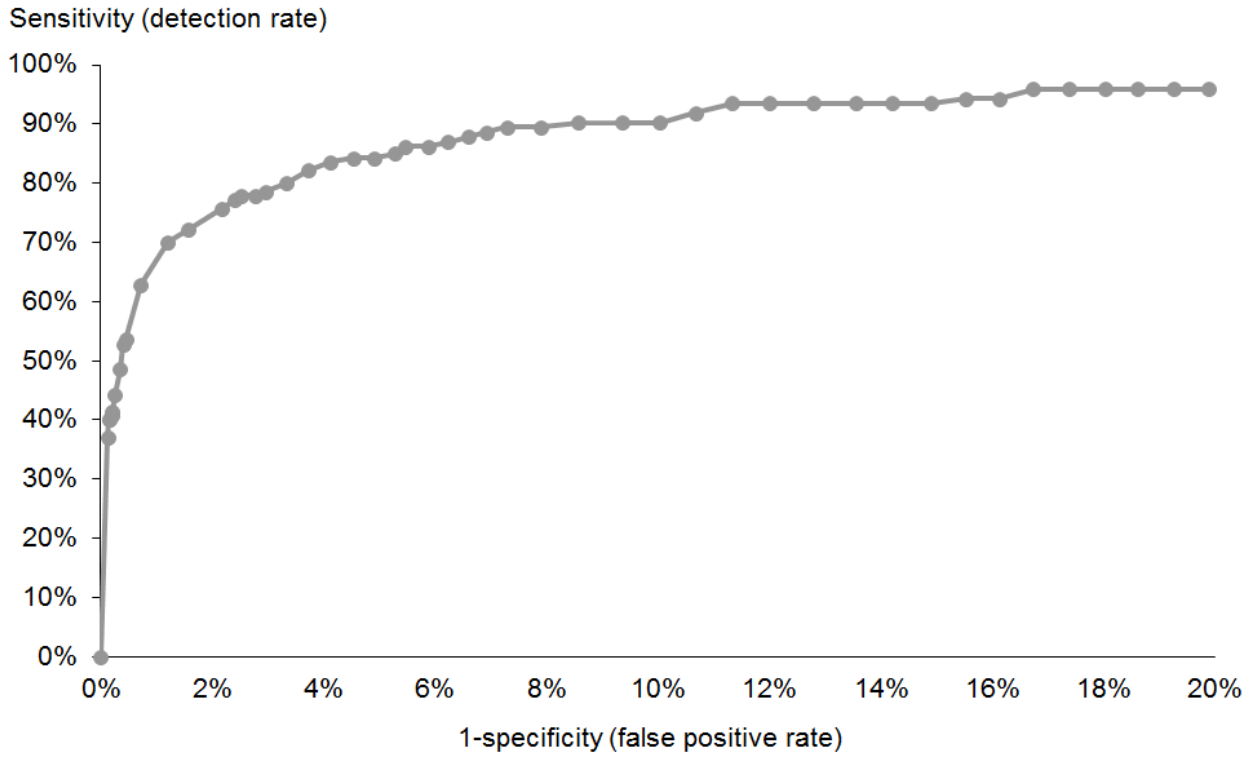
Risk level	False negatives				Total negative (low risk) screens				% of negative screens that are false negatives			
	2011	2012	2013	2014	2011	2012	2013	2014	2011	2012	2013	2014
1:301 to 1:500	9	7	8	6	482	554	571	580	1.87	1.26	1.40	1.03
1:510 to 1:1,000	6	5	7	6	1407	1439	1395	1423	0.43	0.35	0.50	0.42
1:1100 to 1:2000	7	7	6	5	2377	2441	2496	2512	0.29	0.29	0.24	0.20
1:2100 to 1:3000	3	4	4	3	2017	2139	2089	2208	0.15	0.19	0.19	0.14
1:3100 to 1:4000	–	3	2	–	1914	1942	1955	1913	–	0.15	0.10	–
1:4100 to 1:5000	4	2	–	1	1693	1741	1689	1713	0.24	0.11	–	0.06
1:5100 to 1:10,000	2	3	6	1	6699	6792	6880	6965	0.03	0.04	0.09	0.01
Less than 1:10,000	2	3	4	3	16,516	17,213	17,100	17,769	0.01	0.02	0.02	0.02
Total	**33**	**34**	**37**	**25**	**33,105**	**34,261**	**34,175**	**35,083**	**0.10**	**0.10**	**0.11**	**0.07**

Appendix 6: ROC curve

Figure 14 shows the false positive rate plotted against the detection rate in what is known as a 'receiver operating characteristic' (ROC) curve. 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 2014 was 80%, and the false positive rate was 2.4%. 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 if the cut off was lowered to increase the detection rate to 85.2%, the false positive rate would increase from 2.4% to 4.7%. This occurs at a risk cut off of 1:650.

Figure 14: ROC curve for trisomy 21, 18 and 13 screening 2014



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 69 rather than the normal 46 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