Antenatal Screening for Down Syndrome and Other Conditions

Monitoring Report January 2011 to December 2015

National Screening Unit

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Executive summary

This report presents data on antenatal screening for Down syndrome and other conditions for the five calendar years from 1 January 2011 to 31 December 2015, and is based on screens that commenced during that time. For the first time a complete data set, with all cytogenetic testing data, has been used.

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 2015

- Screening was commenced for 80% of pregnancies [indicator 1].
- Screening uptake by Māori and Pacific women was half or less the rate of Other women in 2015. Pacific rates have increased each year since 2011, but the rate for Māori reduced slightly for 2015 after increases in previous years [indicators 1 and 2].
- The national screening completion rate has increased each year with 72% of births being screened in 2015. Trimester one screens made up 87% of all completed screens in 2015 [indicator 2].
- Most DHBs showed a trend of increasing rates of screening commencement and completion [indicators 1 and 2].
- Just over half of all completed trimester 2 screens were commenced in trimester 1 [indicator 3].
- Eleven percent of screens commenced in 2015 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 2015, consistent with 2014. The positive test rate was higher for second trimester screens (4 per 100 screens) than for first trimester screens (2.6 per 100 screens) for 2015, but the difference in rates was smaller than in 2014 due to a lower T2 positive test rate [indicator 5].

- The false positive rate for trisomy 21, 18 and 13 was 2% in 2015, consistent with previous years. The rate was higher for second trimester screens (4%) than for first trimester screens (2%) [indicator 10].
- The overall detection rate for trisomy 21, 18 and 13 increased to 87% in 2015, up from 81% in 2014 [indicator 11].
- The radiology quality improvement project has produced positive results with a higher proportion of ultrasound operators completing sufficient scans to enable valid statistical monitoring and feedback. There has also been improvement in the proportion of ultrasound operators whose nuchal translucency measurements are within accepted levels of variance relative to the Fetal Medicine Foundation reference curve [indicators 12, 13 and 14].

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 (and nasal bone assessment if provided) 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:

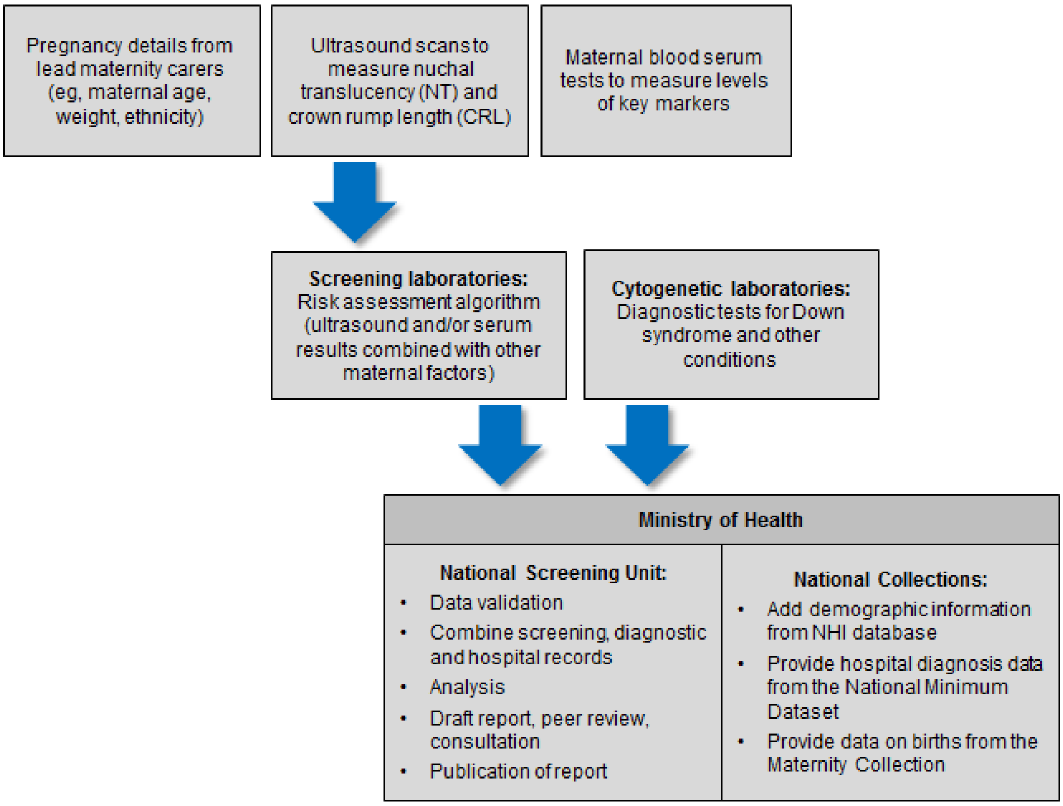
- trisomy 21 (Down Syndrome)
- trisomy 18 (Edwards syndrome)
- trisomy 13 (Patau syndrome)
- triploidy
- Turner syndrome
- neural tube defects

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 monitoring results for antenatal screening for Down syndrome and other conditions for the period 1 January 2011 to 31 December 2015. The definitions for the 14 indicators in this report are contained in Appendix 1. Figure 1 outlines the data collection process the National Screening Unit used to produce indicators 1 to 11. Indicators 12 to 14 relate to separate, independent analysis of NT measurements from ultrasound scans that was completed for 2014 and 2015 screens.

Figure 1: Data collection process



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.

Information included in this report

The screening data in this report was sourced from LabPLUS and covers all of New Zealand. For the first time diagnostic testing data was received from all cytogenetic laboratories (LabPLUS, Waikato, Capital and Coast, and Canterbury Health Laboratories). This has enabled complete results to be calculated for all indicators for the full period. This has led to adjustment in historical results for indicators 6 to 11, which in previous reports excluded women screened from Canterbury, West Coast and South Canterbury DHBs.

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.

This report also includes radiology monitoring indicators (indicators 12 – 14) for the first time. The ultrasound scan data used for these indicators was sourced from LabPlus. These data were forwarded to an independent analytical service in the United Kingdom (Statistical Solutions Limited (SSL)) where it was analysed, and returned, presented in graphical format which was sent to each radiology practice, radiologist and ultrasound operator as part of the Feedback to Radiology project, an initiative to improve the quality of NT and CRL measurements when assessed against the Fetal Medicine Foundation (FMF) reference curve. The same data have been used as a basis for indicators 12 to 14 in this monitoring report.

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]

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 2015 (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. Less than 1% of records related to women with unknown/not stated ethnicity. These were grouped with *Other* for this report.

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

[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.

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 (denominator 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 a karyotype or array by chorionic villus sampling (CVS) or amniocentesis procedures (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.

- 93 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 (2 records less than 12 years, 3 records 50 years old or greater). These records were excluded from the analysis.

Indicator 1: Screens commenced

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

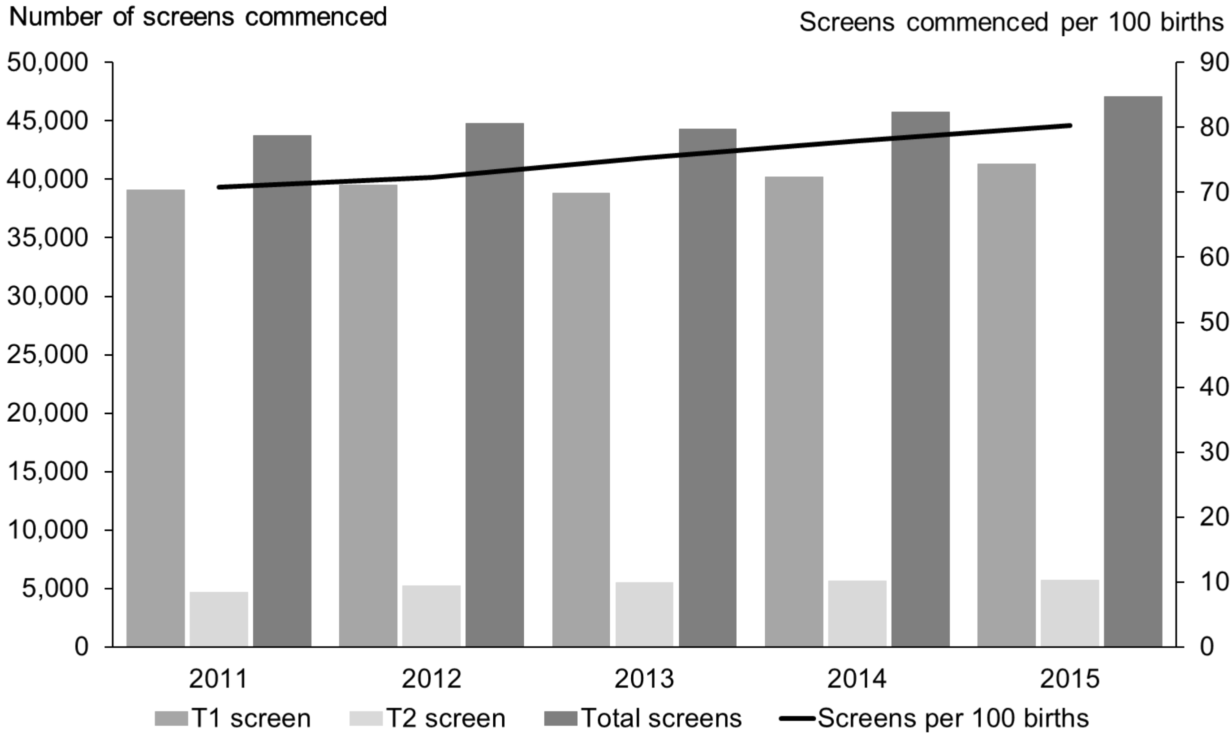
Total screens commenced by trimester

During 2015, a total of 47,064 screens were commenced, a rate of 80 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 in 2011 to 80 in 2015 (see Table 2 and Figure 2).

Table 2: Total screens commenced by trimester, January 2011 to December 2015

Trimester of screen	Number and rate of screens commenced				
	2011	2012	2013	2014	2015
T1 screen	39,087	39,526	38,803	40,172	41,332
T2 screen	4,690	5,230	5,487	5,613	5,732
Total screens	**43,777**	**44,756**	**44,290**	**45,785**	**47,064**
Screens per 100 births	70.8	72.3	75.3	78.0	80.3

Figure 2: Count and rate of screens commenced, January 2011 to December 2015



Screens commenced by DHB

Figure 3 shows the screening commencement rates by DHB for 2015. There was a large variation in rates from 60 per 100 births in Northland to 96 per 100 births in Nelson Marlborough (see Figure 3). Half of all DHBs had rates of above 80 per 100 births. Table 3 gives a full breakdown by the trimester of the screen.

Figure 3: Screens commenced by DHB, January 2015 to December 2015

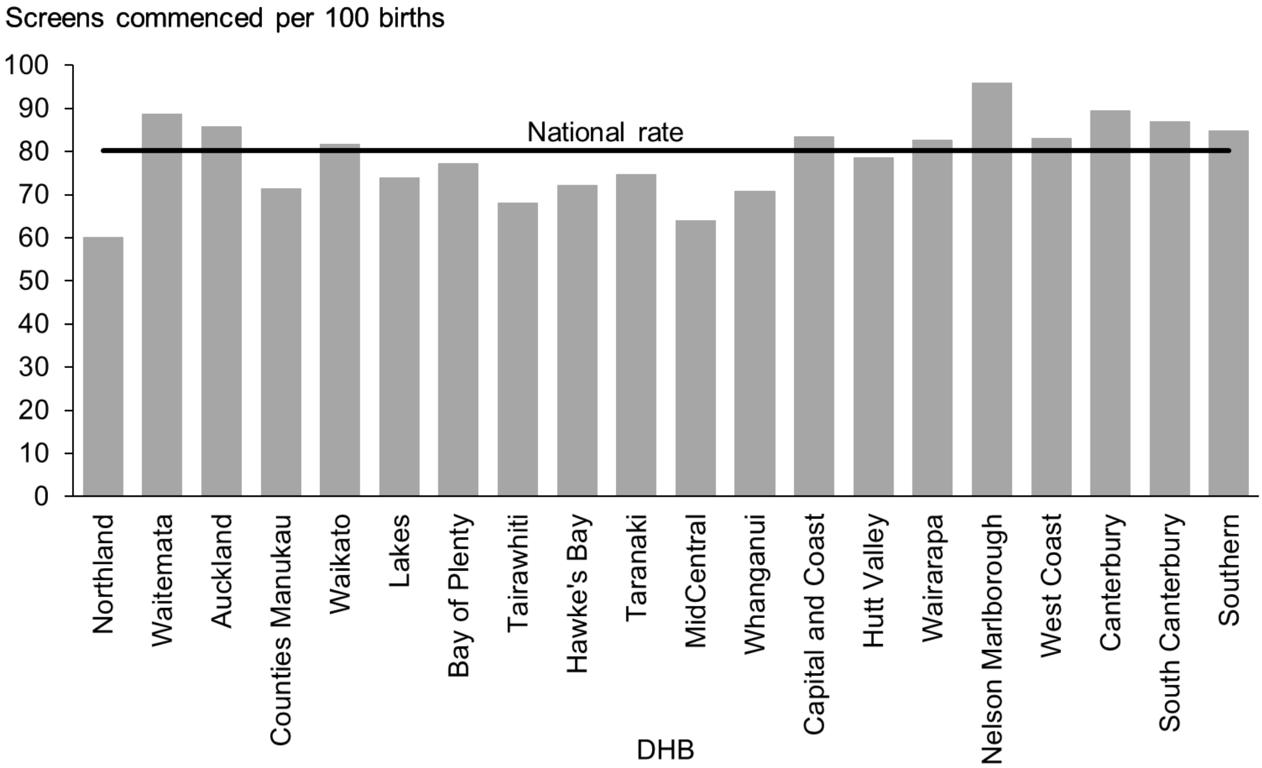


Table 3: Screens commenced by trimester and DHB, January 2015 to December 2015

DHB	Number of screens commenced			Screens commenced (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	1,120	164	1,284	52.5	7.7	60.1
Waitemata	5,960	737	6,697	78.9	9.8	88.7
Auckland	4,452	615	5,067	75.4	10.4	85.9
Counties Manukau	4,677	1,178	5,855	57.0	14.4	71.4
Waikato	3,855	459	4,314	72.9	8.7	81.6
Lakes	1,002	113	1,115	66.4	7.5	73.9
Bay of Plenty	1,985	176	2,161	70.9	6.3	77.2
Tairawhiti	435	70	505	58.6	9.4	68.1
Hawke's Bay	1,292	153	1,445	64.5	7.6	72.2
Taranaki	942	189	1,131	62.2	12.5	74.7
MidCentral	1,201	149	1,350	56.9	7.1	63.9
Whanganui	492	85	577	60.3	10.4	70.7
Capital and Coast	2,658	291	2,949	75.2	8.2	83.4
Hutt Valley	1,348	197	1,545	68.5	10.0	78.5
Wairarapa	339	44	383	73.2	9.5	82.7
Nelson Marlborough	1,248	111	1,359	88.1	7.8	95.9
West Coast	260	36	296	73.0	10.1	83.1
Canterbury	4,953	609	5,562	79.8	9.8	89.6
South Canterbury	485	88	573	73.6	13.4	86.9
Southern	2,628	268	2,896	77.0	7.9	84.8
Total	**41,332**	**5,732**	**47,064**	**70.5**	**9.8**	**80.3**

Most DHBs showed an increase in their rate of screens commenced between 2014 and 2015, or had fairly stable rates. Exceptions to this were Lakes, Nelson Marlborough and West Coast where rates decreased between 1.5 and 5% between 2014 and 2015 (see Table 4).

Table 4: Screens commenced per 100 births by DHB, January 2011 to December 2015

DHB	Screens commenced (per 100 births)				
	2011	2012	2013	2014	2015
Northland	46.5	49.7	52.8	55.6	60.1
Waitemata	83.9	82.9	86.3	86.2	88.7
Auckland	75.0	74.4	82.4	84.0	85.9
Counties Manukau	60.9	63.4	64.9	68.8	71.4
Waikato	72.8	72.1	76.2	80.4	81.6
Lakes	60.5	67.8	70.2	77.3	73.9
Bay of Plenty	65.3	68.6	69.6	72.5	77.2
Tairawhiti	44.2	49.1	52.7	58.5	68.1
Hawke's Bay	55.8	61.8	64.3	66.1	72.2
Taranaki	62.6	60.2	61.3	68.2	74.7
MidCentral	51.0	54.4	58.3	59.2	63.9
Whanganui	45.1	44.9	48.1	61.1	70.7
Capital and Coast	76.4	79.3	78.2	80.3	83.4
Hutt Valley	70.9	70.7	72.6	78.6	78.5
Wairarapa	72.8	69.2	76.6	81.6	82.7
Nelson Marlborough	87.9	90.8	87.2	97.6	95.9
West Coast	68.9	76.5	81.3	88.0	83.1
Canterbury	85.4	86.8	90.3	89.4	89.6
South Canterbury	92.1	85.5	88.1	78.7	86.9
Southern	75.3	80.0	81.4	83.3	84.8
Total	**70.8**	**72.3**	**75.3**	**78.0**	**80.3**

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 2015. The 30–34 and 25-29 year age groups had the highest rate of screens commenced for 2015 with 84 women starting screening per 100 births. This was closely followed by the 35-39 year age group with 82 per 100 births. Rates dropped sharply for other age groups to 72% or below (see Figure 4). Rates appear to be increasing for all age groups, except for 40-44 years and 45 years plus, which do not show a clear trend.

Differences in screening commencement rates by ethnicity remained consistent for 2015. Women of Other ethnicity had the highest rate (100%) followed by Asian women (95%). The rate slightly above 100% for Other women is due to the current denominator limitations as discussed under the Data Limitations section. The rate of commenced screens for Pacific and Māori women was lower at 52 per 100 births and 43 per 100 births respectively (see Figure 5). All groups have shown increasing rates over the five years, except for Māori which decreased 1% in 2015 (see Table 5).

A trend of higher screening commencement rates for women in less deprived areas was evident, with 96 women per 100 per births starting screening for quintile 1 in 2015 compared with 64 per

100 births for quintile 5 (see Figure 6). All quintiles showed a rate increase between 2014 and 2015 (see Table 5).

Table 5: Screens commenced by age of mother, ethnicity and NZ deprivation quintile, January 2011 to December 2015

	Number of screens commenced					Screens commenced (per 100 births)#				
	2011	2012	2013	2014	2015	2011	2012	2013	2014	2015
Age at screen										
Under 20 years	2,282	2,128	1,947	1,990	1,928	56.3	54.5	58.6	66.4	69.2
20–24 years	6,817	6,966	6,932	7,055	7,129	58.2	60.8	64.2	68.6	71.6
25–29 years	11,509	12,078	12,022	12,800	13,206	74.0	75.8	78.7	81.5	83.9
30–34 years	13,433	13,751	13,914	14,623	15,124	78.0	78.8	83.0	83.2	84.4
35–39 years	8,027	8,040	7,628	7,610	8,004	74.8	77.3	76.0	78.6	82.0
40–44 years	1,636	1,716	1,767	1,626	1,595	68.0	66.6	72.5	69.4	69.3
45 years and over	73	77	80	81	78	57.9	64.2	55.9	61.4	56.1
Ethnicity										
Māori	5,540	5,881	5,805	6,284	6,269	35.0	37.5	39.9	44.2	43.1
Pacific	3,055	3,102	2,999	3,005	3,130	43.2	45.1	47.3	48.8	51.7
Asian	6,484	7,405	7,474	8,438	8,714	90.9	87.6	91.6	91.7	94.6
Other	28,698	28,368	28,012	28,058	28,951	90.3	91.9	94.2	96.3	100.6
NZ Deprivation Quintile										
Quintile 1	8,130	8,073	7,654	7,732	7,896	95.5	93.0	93.6	91.3	95.8
Quintile 2	8,174	8,395	8,231	8,413	8,660	86.0	87.4	89.0	91.8	92.7
Quintile 3	8,529	8,685	8,730	8,878	9,135	76.5	77.7	82.1	84.1	86.2
Quintile 4	9,526	9,822	9,882	10,353	10,482	69.0	71.9	73.6	77.9	79.1
Quintile 5	9,409	9,777	9,789	10,408	10,885	50.0	52.1	56.6	60.4	63.8
Unknown	9	4	4	1	6	-	-	-	-	-
Total	**43,777**	**44,756**	**44,290**	**45,785**	**47,064**	**63.4**	**65.7**	**69.3**	**71.1**	**72.0**

Rate suppressed if the number of screens was <10.

Figure 4: Screens commenced by age of mother at screen, January 2015 to December 2015

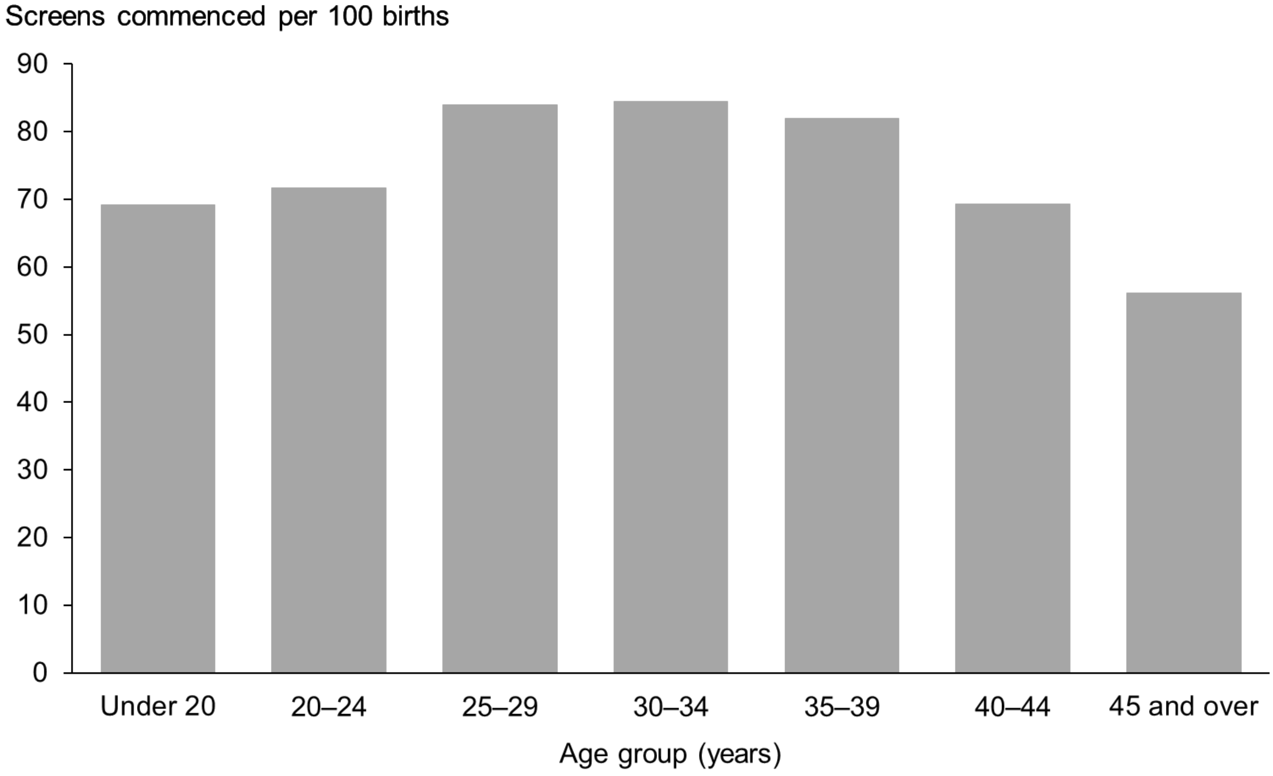


Figure 5: Screens commenced by ethnicity of mother, January 2015 to December 2015

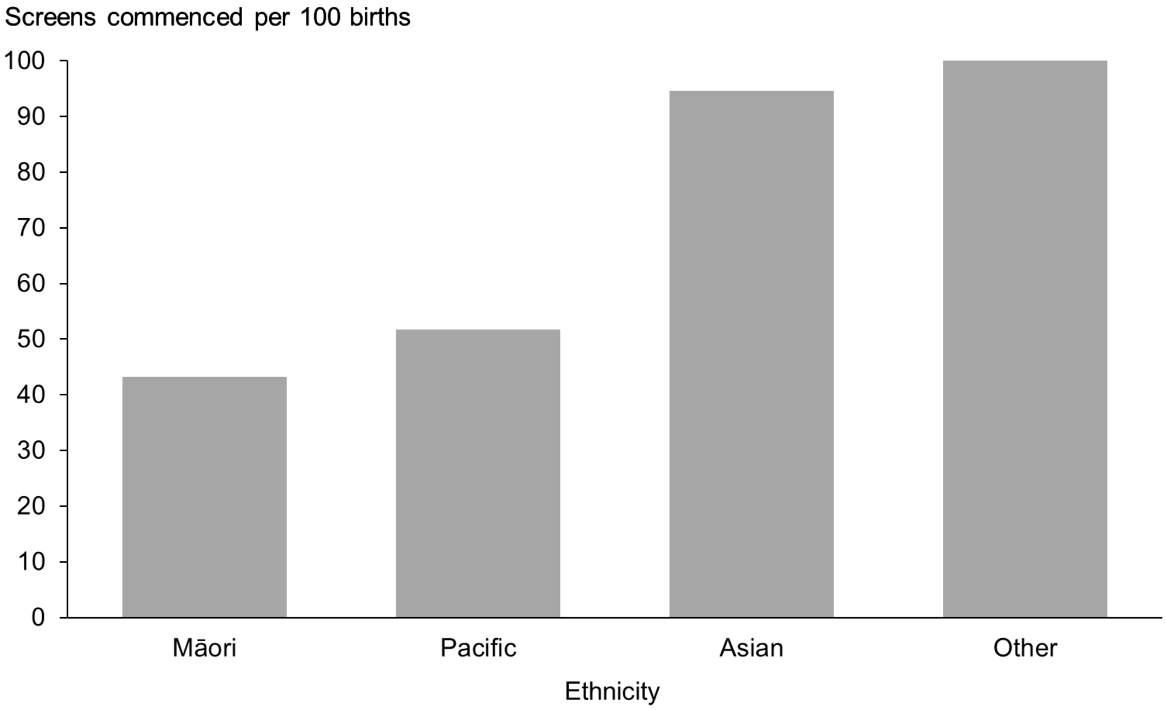
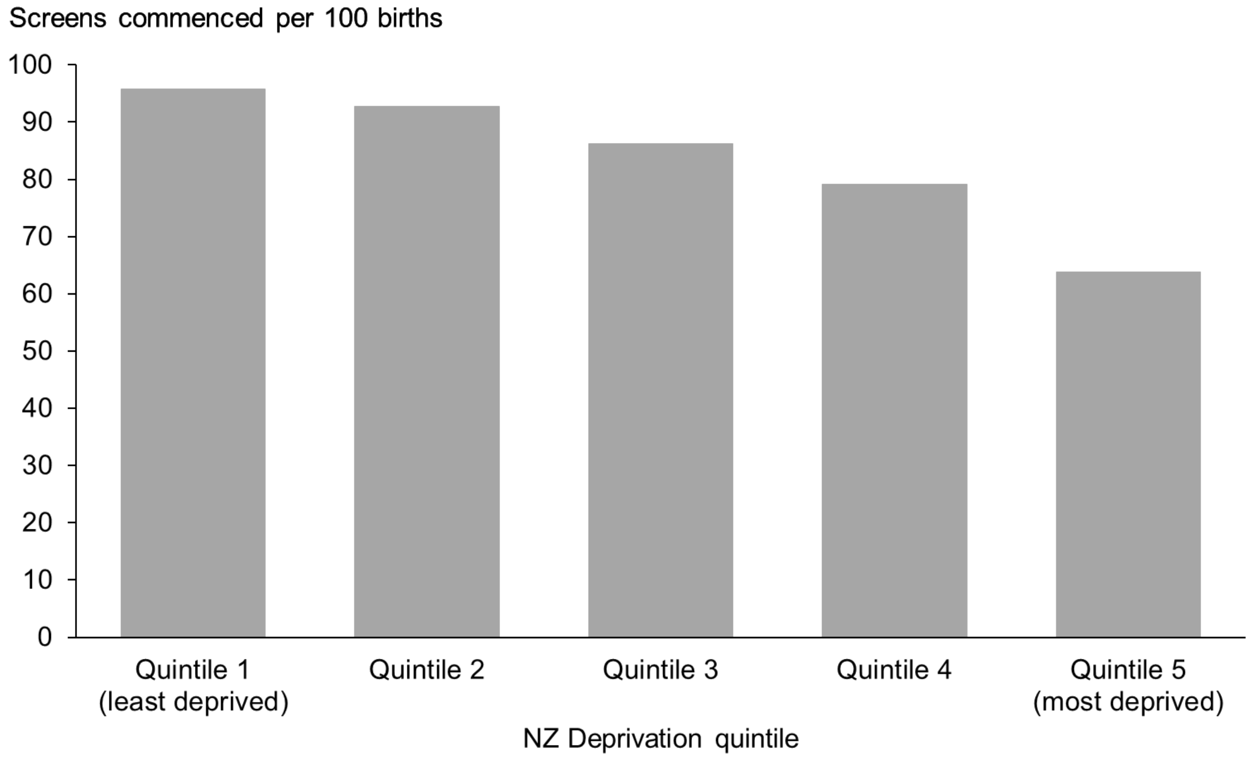


Figure 6: Screens commenced by NZ deprivation quintile, January 2015 to December 2015



Indicator 2: Screens completed

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

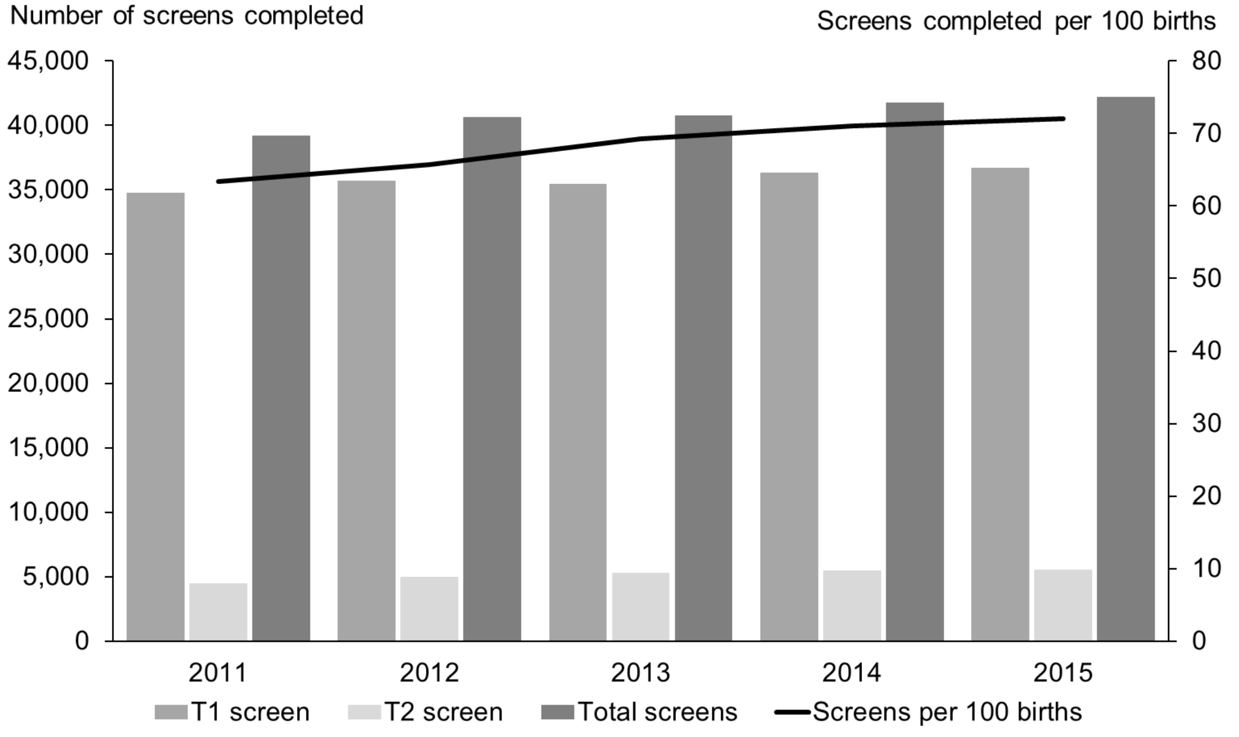
Total screens completed by trimester

During 2015, a total of 42,212 screens were completed, a rate of 72 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 and rate of completed screens has increased annually since 2011.

Table 6: Total screens completed by trimester, January 2011 to December 2015

Trimester of screen	Number and rate of screens completed				
	2011	2012	2013	2014	2015
T1 screen	34,735	35,691	35,464	36,280	36,704
T2 screen	4,446	4,957	5,269	5,456	5,508
Total screens	**39,181**	**40,648**	**40,733**	**41,736**	**42,212**
Screens per 100 births	63.4	65.7	69.3	71.1	72.0

Figure 7: Count and rate of screens completed, January 2011 to December 2015



Screens completed by DHB

Screening completion rates for 2015 varied across DHBs from 51 per 100 births in Northland to 84 per 100 births in Nelson Marlborough (see Figure 8). Table 7 gives a full breakdown by the trimester of the screen.

Figure 8: Screens completed by DHB, January 2015 to December 2015

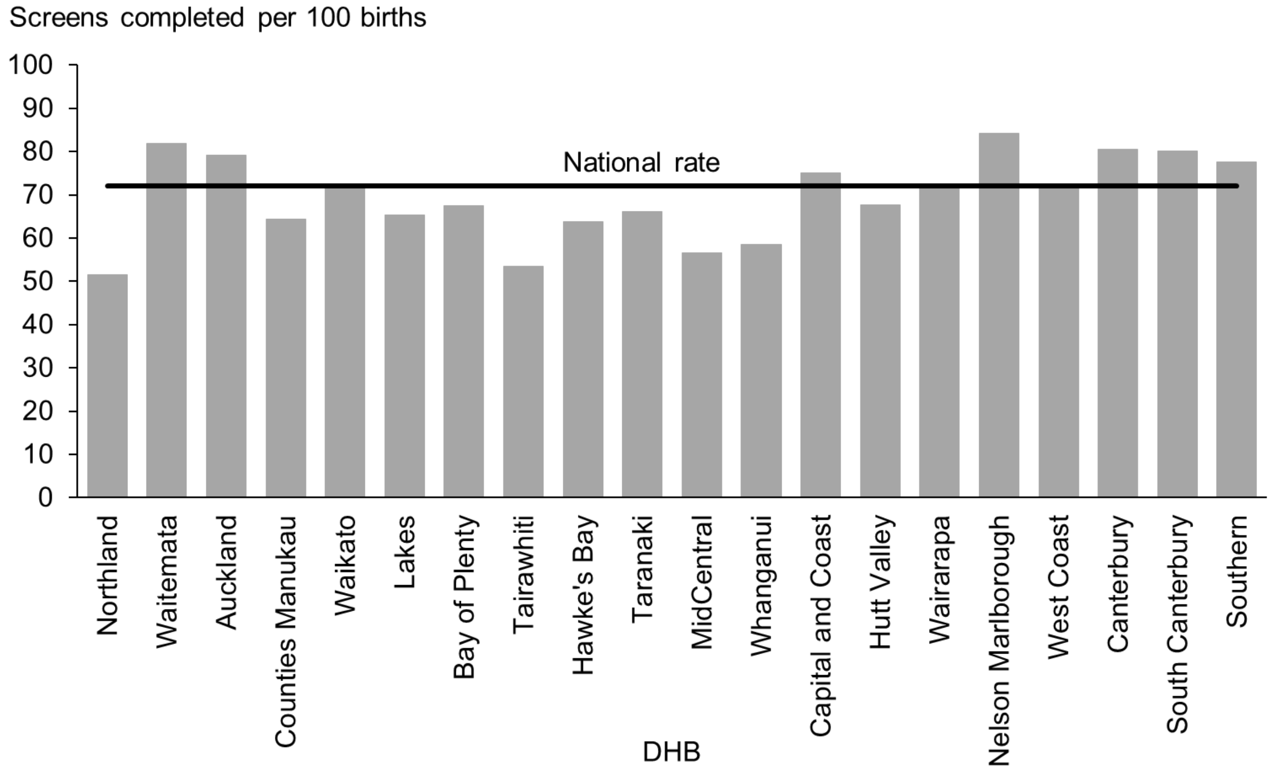


Table 7: Screening completion by trimester and DHB, January 2015 to December 2015

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	941	159	1,100	44.1	7.4	51.5
Waitemata	5,473	710	6,183	72.5	9.4	81.9
Auckland	4,081	587	4,668	69.1	9.9	79.1
Counties Manukau	4,153	1,132	5,285	50.6	13.8	64.4
Waikato	3,369	440	3,809	63.7	8.3	72.0
Lakes	875	111	986	58.0	7.4	65.3
Bay of Plenty	1,718	169	1,887	61.4	6.0	67.4
Tairawhiti	329	68	397	44.3	9.2	53.5
Hawke's Bay	1,133	144	1,277	56.6	7.2	63.8
Taranaki	819	182	1,001	54.1	12.0	66.1
MidCentral	1,053	144	1,197	49.9	6.8	56.7
Whanganui	396	82	478	48.5	10.0	58.6
Capital and Coast	2,373	277	2,650	67.1	7.8	75.0
Hutt Valley	1,140	190	1,330	58.0	9.7	67.6
Wairarapa	291	41	332	62.9	8.9	71.7
Nelson Marlborough	1,087	106	1,193	76.7	7.5	84.2
West Coast	224	34	258	62.9	9.6	72.5
Canterbury	4,421	583	5,004	71.2	9.4	80.6
South Canterbury	440	88	528	66.8	13.4	80.1
Southern	2,388	261	2,649	69.9	7.6	77.6
Total	**36,704**	**5,508**	**42,212**	**62.6**	**9.4**	**72.0**

Similar to screens commenced, most DHBs showed a trend of increasing rates of screening completion over the five years covered in this report. West Coast was an exception to this with a decrease in completion rates 2015. South Canterbury's rate showed an increase in 2015 after a decrease in 2014 (see Table 8).

Table 8: Screening completion by DHB, January 2011 to December 2015

DHB	Screens completed (per 100 births)				
	2011	2012	2013	2014	2015
Northland	41.1	44.4	47.0	48.0	51.5
Waitemata	78.0	77.9	82.1	81.0	81.9
Auckland	70.4	69.4	77.6	78.9	79.1
Counties Manukau	53.8	57.3	59.7	63.3	64.4
Waikato	65.1	64.2	69.0	72.5	72.0
Lakes	53.1	59.1	62.7	69.8	65.3
Bay of Plenty	58.3	61.7	62.1	64.5	67.4
Tairawhiti	39.4	44.3	46.7	50.7	53.5
Hawke's Bay	50.2	55.9	59.7	59.5	63.8
Taranaki	58.2	55.5	55.0	61.3	66.1
MidCentral	45.2	49.5	53.8	53.9	56.7
Whanganui	40.3	41.8	45.2	53.1	58.6
Capital and Coast	67.8	71.9	70.9	72.6	75.0
Hutt Valley	59.0	62.6	64.6	68.9	67.6
Wairarapa	62.8	59.6	66.7	70.6	71.7
Nelson Marlborough	78.7	81.4	78.0	87.6	84.2
West Coast	55.6	68.6	72.5	78.6	72.5
Canterbury	72.3	75.8	81.9	81.1	80.6
South Canterbury	87.0	82.6	85.6	75.2	80.1
Southern	67.3	73.6	75.6	74.9	77.6
Total	**63.4**	**65.7**	**69.3**	**71.1**	**72.0**

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 2015, 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 2015. This was followed by the 35–39 years and 25-29 years age groups with 76 per 100 births and 75 per 100 births respectively (see Figure 9).

Screening completion rates were highest among women of Other ethnicity at 92 per 100 births for 2015. This was followed closely by Asian women at 88 per 100 births. The rate of completed screens for Pacific and Māori women remains lower at 43 per 100 births and 34 per 100 births respectively (see Figure 10). The rate for Māori is a decrease on the previous year (see Table 9).

Screening completion rates were highest among women in less deprived areas with a rate of 89 per 100 per births for quintile 1 in 2015 compared with 54 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 2015

	Number of screens completed					Screens completed (per 100 births)[#]				
	2011	2012	2013	2014	2015	2011	2012	2013	2014	2015
Age at screen										
Under 20 years	1,808	1,699	1,610	1,604	1,507	44.6	43.5	48.4	53.5	54.1
20–24 years	5,754	5,890	6,010	6,070	5,988	49.2	51.4	55.6	59.1	60.2
25–29 years	10,276	10,997	11,097	11,685	11,811	66.1	69.0	72.6	74.4	75.1
30–34 years	12,353	12,859	13,089	13,675	14,018	71.7	73.6	78.1	77.8	78.3
35–39 years	7,453	7,543	7,214	7,144	7,418	69.5	72.5	71.9	73.8	76.0
40–44 years	1,474	1,588	1,643	1,486	1,406	61.3	61.6	67.4	63.4	61.1
45 years and over	63	72	70	72	64	50.0	60.0	49.0	54.5	46.0
Ethnicity										
Māori	4,561	4,880	4,893	5,178	4,902	28.8	31.1	33.6	36.4	33.7
Pacific	2,479	2,591	2,606	2,598	2,623	35.1	37.7	41.1	42.2	43.3
Asian	6,024	6,990	7,091	8,034	8,114	84.4	82.7	87.0	87.3	88.1
Other	26,117	26,187	26,143	25,926	26,573	82.2	84.8	87.9	89.0	92.3
NZ Deprivation Quintile										
Quintile 1	7,519	7,520	7,255	7,242	7,329	88.4	86.6	88.7	85.5	88.9
Quintile 2	7,480	7,805	7,749	7,867	8,025	78.7	81.3	83.8	85.9	85.9
Quintile 3	7,748	8,028	8,102	8,195	8,318	69.5	71.9	76.2	77.6	78.5
Quintile 4	8,401	8,851	9,001	9,325	9,293	60.8	64.8	67.1	70.2	70.2
Quintile 5	8,027	8,441	8,622	9,106	9,241	42.7	45.0	49.8	52.9	54.2
Unknown	6	3	4	1	6					
Total	**39,181**	**40,648**	**40,733**	**41,736**	**42,212**	**63.4**	**65.7**	**69.3**	**71.1**	**72.0**

Rate suppressed if the number of screens was <10.

Figure 9: Screens completed by age of mother at screen, January 2015 to December 2015

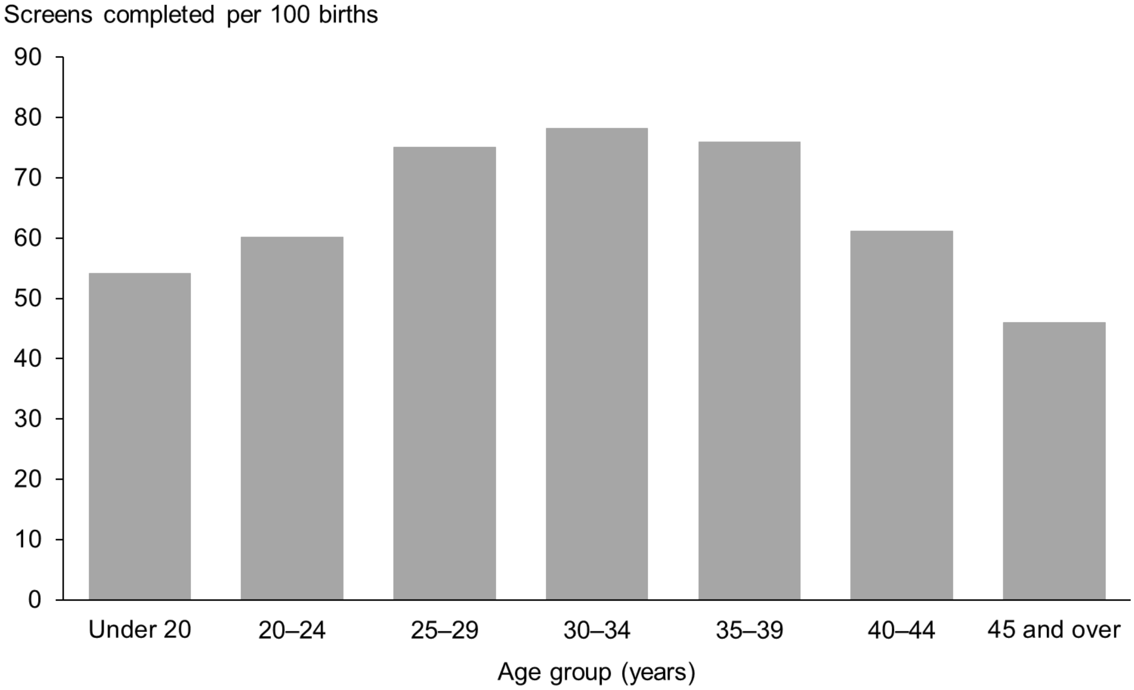


Figure 10: Screens completed by ethnicity of mother, January 2015 to December 2015

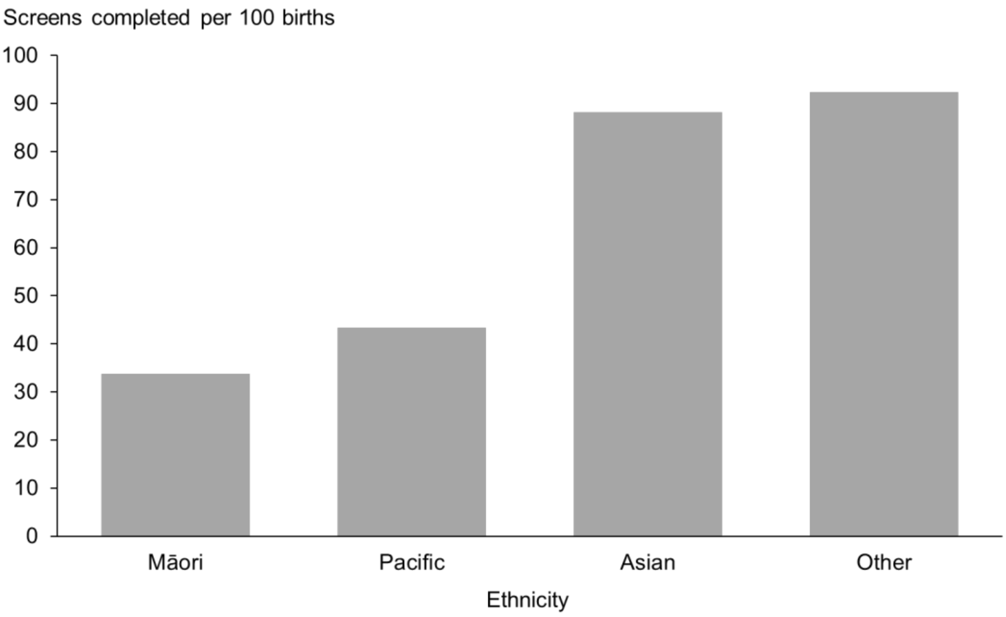
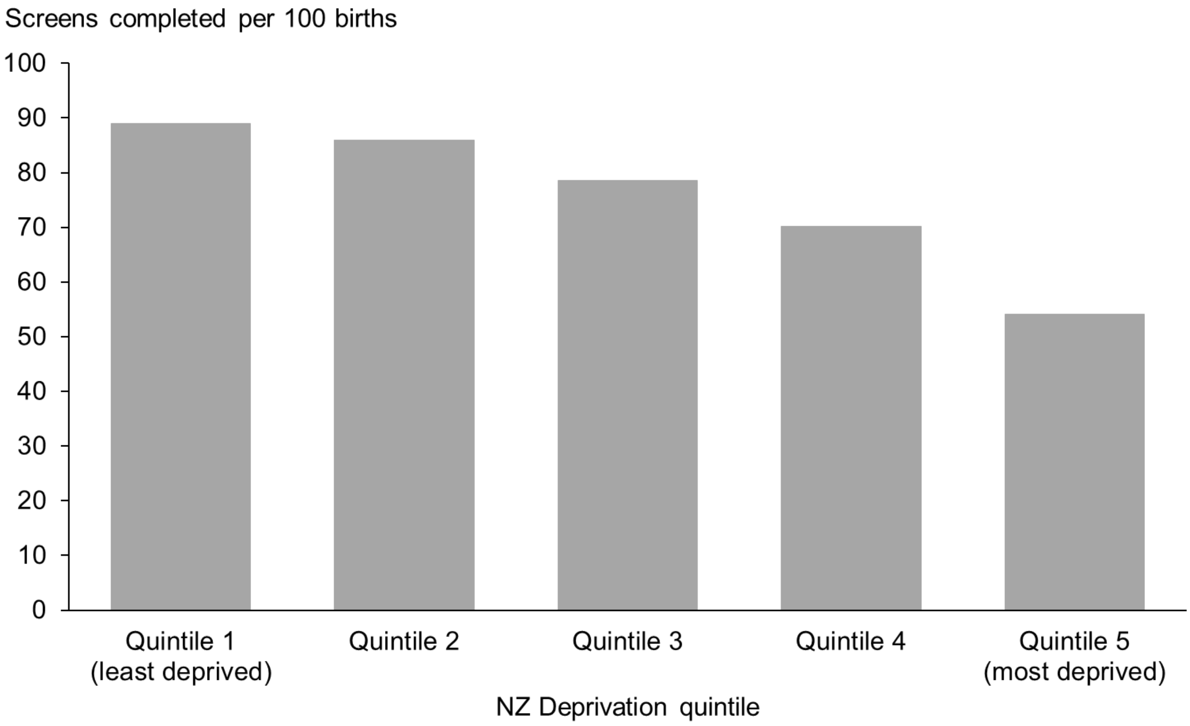


Figure 11: Screens completed by NZ deprivation quintile of mother, January 2015 to December 2015



Indicator 3: Screening pathway variance

This section reports on the number of screens completed in the second trimester which included first trimester screening components. 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. Any information available from the first trimester (NT or PAPP-A) 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 suitable first trimester blood sample. Second trimester results with PAPP-A indicate that the screening laboratory did not receive an 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 the number and proportion of second trimester screening results that included first trimester inputs over the period from 2011 to 2015. This has been broken down by the type of pathway variance.

The proportion of trimester 2 screens with an NT measurement has increased over the four year period from 41% to 45%. The proportion with PAPP-A increased slightly from 6% to 7% between 2012 and 2013 before returning to 6% for 2015.

Table 10: Screening pathway variance by type, January 2011 to December 2015

Year	Second trimester screening results				
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
	Number			Percentage	
2011	4,446	1,811	264	40.7	5.9
2012	4,957	2,048	291	41.3	5.9
2013	5,269	2,219	361	42.1	6.9
2014	5,456	2,379	376	43.6	6.9
2015	5,508	2,466	343	44.8	6.2

Screening pathway variance by DHB

Table 11 shows a breakdown of screening pathway variance by DHB and type of variance for the 2015 year. Care should be taken with interpretation given the low number of T2 screens for many DHBs. In general, the national result is reflected at DHB level with a far higher number of women having an NT scan and a T2 screen than those having a T2 screen with PAPP-A.
The crown rump length (CRL) measured by ultrasound is used by the screening laboratory to calculate gestation (may be different from the clinically gestation) leading to women being assessed in a different trimester.

Table 11: Screening pathway variance by DHB, January 2015 to December 2015

DHB	Second trimester screening results				
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
	Number			Percentage	
Northland	159	66	18	41.5	11.3
Waitemata	710	355	35	50.0	4.9
Auckland	587	221	33	37.6	5.6
Counties Manukau	1,132	397	54	35.1	4.8
Waikato	440	195	18	44.3	4.1
Lakes	111	56	7	50.5	6.3
Bay of Plenty	169	84	9	49.7	5.3
Tairawhiti	68	32	9	47.1	13.2
Hawke’s Bay	144	55	8	38.2	5.6
Taranaki	182	62	22	34.1	12.1
MidCentral	144	55	12	38.2	8.3
Whanganui	82	51	2	62.2	2.4
Capital and Coast	277	121	30	43.7	10.8
Hutt Valley	190	98	18	51.6	9.5
Wairarapa	41	23	3	56.1	7.3
Nelson Marlborough	106	76	3	71.7	2.8
West Coast	34	19	3	55.9	8.8
Canterbury	583	294	45	50.4	7.7
South Canterbury	88	56	2	63.6	2.3
Southern	261	150	12	57.5	4.6
Total	**5,508**	**2,466**	**343**	**44.8**	**6.2**

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 2015 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 2015 to December 2015

	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	420	164	19	39.0	4.5
20–24 years	1,328	566	67	42.6	5.0
25–29 years	1,549	702	102	45.3	6.6
30–34 years	1,359	659	91	48.5	6.7
35–39 years	711	316	50	44.4	7.0
40–44 years	135	57	14	42.2	10.4
45 years and over	6	2	0	-	-
Ethnicity					
Māori	1,262	491	78	38.9	6.2
Pacific	1,024	301	50	29.4	4.9
Asian	980	410	69	41.8	7.0
Other	2,242	1,264	146	56.4	6.5
NZ Deprivation Quintile					
Quintile 1	564	321	33	56.9	5.9
Quintile 2	695	385	44	55.4	6.3
Quintile 3	933	450	65	48.2	7.0
Quintile 4	1,304	589	89	45.2	6.8
Quintile 5	2,011	721	112	35.9	5.6
Unknown	1	0	0	-	-
Total	**5,508**	**2,466**	**343**	**44.8**	**6.2**

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 2015 was 4,852, which equates to 10% of screens commenced that year.

Table 13: Incomplete screens by trimester, January 2011 to December 2015

Trimester of screen	Number of incomplete screens				
	2011	2012	2013	2014	2015
T1 screen	4,352	3,835	3,339	3,892	4,628
T2 screen	244	273	218	157	224
Total screens	**4,596**	**4,108**	**3,557**	**4,049**	**4,852**

Incomplete T1 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 in 2015 was 11%, up slightly from 2014. This was the result of increases in both screens without blood samples and screens without NT scans. The split between the percentage of incompletes due to no blood or no NT scan has changed over the 5 years covered in this report (see far right columns of Table 14), with an increasing proportion of incompletes being due to no NT scan (36% in 2015 compared with 24% in 2011).

During 2015 there was one further incomplete T1 screen that had both an NT scan and a blood sample but no weight was provided. In situations of missing maternal weight the screening laboratory follows up but in this case no weight was supplied. This means that the sum of the ‘Reason Incomplete’ columns of tables 14, 15, and 16 is one short of the total number of incomplete T1 screens given in table 13. Inclusion of actual weight in the risk algorithm, as opposed to entering a default weight, leads to a far more accurate risk result.

Table 14: Incomplete T1 screens by reason incomplete, January 2011 to December 2015

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	39,087	4,352	3,294	1,058	8.4	2.7	11.1	75.7	24.3
2012	39,526	3,835	2,844	991	7.2	2.5	9.7	74.2	25.8
2013	38,803	3,339	2,318	1,021	6.0	2.6	8.6	69.4	30.6
2014	40,172	3,892	2,630	1,262	6.5	3.1	9.7	67.6	32.4
2015	41,332	4,628	2,974	1,653	7.2	4.0	11.2	64.3	35.7

Incomplete T1 screens by reason and DHB

Table 15 provides the same breakdown by DHB. The lower numbers involved limit DHB comparisons. The range in the percentage of screens incomplete due to no blood sample was from 48 % (at Taranaki) to 84% (at South Canterbury). For screens incomplete due to no NT scan the range was from 16% (at South Canterbury) to 52% (at Taranaki). As these range values indicate, Taranaki DHB had the most even split for reason incomplete, while other DHBs had a higher proportion with no blood sample.

Table 15: Incomplete T1 screens by reason and DHB, January 2015 to December 2015

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	1,120	179	117	62	10.4	5.5	16.0	65.4	34.6
Waitemata	5,960	487	294	193	4.9	3.2	8.2	60.4	39.6
Auckland	4,452	371	214	157	4.8	3.5	8.3	57.7	42.3
Counties Manukau	4,677	524	315	209	6.7	4.5	11.2	60.1	39.9
Waikato	3,855	486	326	160	8.5	4.2	12.6	67.1	32.9
Lakes	1,002	127	90	37	9.0	3.7	12.7	70.9	29.1
Bay of Plenty	1,985	267	178	89	9.0	4.5	13.5	66.7	33.3
Tairawhiti	435	106	80	26	18.4	6.0	24.4	75.5	24.5
Hawke's Bay	1,292	159	111	48	8.6	3.7	12.3	69.8	30.2
Taranaki	942	123	59	64	6.3	6.8	13.1	48.0	52.0
MidCentral	1,201	148	82	66	6.8	5.5	12.3	55.4	44.6
Whanganui	492	96	76	20	15.4	4.1	19.5	79.2	20.8
Capital and Coast	2,658	285	180	105	6.8	4.0	10.7	63.2	36.8
Hutt Valley	1,348	208	145	63	10.8	4.7	15.4	69.7	30.3
Wairarapa	339	48	33	15	9.7	4.4	14.2	68.8	31.3
Nelson Marlborough	1,248	161	113	47	9.1	3.8	12.9	70.2	29.2
West Coast	260	36	27	9	10.4	3.5	13.8	75.0	25.0
Canterbury	4,953	532	346	186	7.0	3.8	10.7	65.0	35.0
South Canterbury	485	45	38	7	7.8	1.4	9.3	84.4	15.6
Southern	2,628	240	150	90	5.7	3.4	9.1	62.5	37.5
Total	**41,332**	**4,628**	**2,974**	**1,653**	**7.2**	**4.0**	**11.2**	**64.3**	**35.7**

Incomplete T1 screens by age, ethnicity and deprivation

Table 16 shows a breakdown of incomplete screens with reason incomplete, by age, ethnicity, and NZ deprivation quintile for the 2015 year. This shows higher rates of incomplete T1 screens for younger women (73-74% for women up to 24 years of age). There were higher rates of incomplete screens for Māori (26%) and Pacific (22%) women when compared with Asian (7%) and Other (9%). The rate of incomplete screens also increased with increasing deprivation (18% for quintile 5 compared with 8% for quintile 1).

Table 16: Incomplete T1 screens by age, ethnicity and NZ deprivation quintile, January 2015 to December 2015

	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	Total T1 incomplete	No blood	No NT scan
Age at screen									
Under 20 years	1,492	405	299	106	20.0	7.1	27.1	73.8	26.2
20 – 24 years	5,754	1,094	797	296	13.9	5.1	19.0	72.9	27.1
25 – 29 years	11,597	1,335	891	444	7.7	3.8	11.5	66.7	33.3
30 – 34 years	13,710	1,051	625	426	4.6	3.1	7.7	59.5	40.5
35 – 39 years	7,257	550	281	269	3.9	3.7	7.6	51.1	48.9
40 – 44 years	1,451	180	77	103	5.3	7.1	12.4	42.8	57.2
45 years and over	71	13	4	9	5.6	12.7	18.3	30.8	69.2
Ethnicity									
Māori	4,947	1,307	935	372	18.9	7.5	26.4	71.5	28.5
Pacific	2,058	459	287	172	13.9	8.4	22.3	62.5	37.5
Asian	7,683	549	260	288	3.4	3.7	7.1	47.4	52.5
Other	26,644	2,313	1,492	821	5.6	3.1	8.7	64.5	35.5
NZ Deprivation Quintile									
Quintile 1	7,316	551	332	219	4.5	3.0	7.5	60.3	39.7
Quintile 2	7,941	611	370	241	4.7	3.0	7.7	60.6	39.4
Quintile 3	8,165	780	490	290	6.0	3.6	9.6	62.8	37.2
Quintile 4	9,121	1,132	748	383	8.2	4.2	12.4	66.1	33.8
Quintile 5	8,784	1,554	1,034	520	11.8	5.9	17.7	66.5	33.5
Total	**46,839**	**4,628**	**2,974**	**1,653**	**6.3**	**3.5**	**9.9**	**64.3**	**35.7**

Incomplete T2 screens

T2 screens do not require an NT scan, just a blood sample, but may be incomplete if missing dating information or weight, if the sample is taken later than 20 weeks of pregnancy, or if the sample is damaged and not repeated. For 2015, 4% of T2 commenced screens were incomplete, compared with 10% of T1 commenced screens. As Table 17 shows, the percentage of incomplete T2 screens decreased from 5% in 2011 to 3% in 2014, before increasing to 4% in 2015.

Table 17: Incomplete T2 screens, January 2011 to December 2015

Year	Commenced second trimester	No result issued	Percentage incomplete
2011	4,690	244	5.2
2012	5,230	273	5.5
2013	5,487	218	4.1
2014	5,613	157	2.9
2015	5,732	224	4.1

Incomplete T2 screens by DHB

Table 18 shows a breakdown of incomplete T2 screens by DHB for the 2015 year. The very low numbers involved limit meaningful DHB comparisons.

Table 18: IncompleteT2 screens by DHB, January 2015 to December 2015

DHB	Commenced second trimester	No result issued	Percentage incomplete
Northland	164	5	3.0
Waitemata	737	27	3.7
Auckland	615	28	4.6
Counties Manukau	1,178	46	3.9
Waikato	459	19	4.1
Lakes	113	2	1.8
Bay of Plenty	176	7	4.0
Tairawhiti	70	2	2.9
Hawke's Bay	153	9	5.9
Taranaki	189	7	3.7
MidCentral	149	5	3.4
Whanganui	85	3	3.5
Capital and Coast	291	14	4.8
Hutt Valley	197	7	3.6
Wairarapa	44	3	6.8
Nelson Marlborough	111	5	4.5
West Coast	36	2	5.6
Canterbury	609	26	4.3
South Canterbury	88	-	0.0
Southern	268	7	2.6
Total	**5,732**	**224**	**3.9**

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 2015. The percentage incomplete was higher for older age groups, lower for women of Other ethnicity, and lower for women in the least deprived quintile.

Table 19: Incomplete T2 screens by age, ethnicity and NZ deprivation quintile, January 2015 to December 2015

	Commenced second trimester	No result issued	Percentage incomplete
Age at screen			
Under 20 years	436	16	3.7
20–24 years	1,375	47	3.4
25–29 years	1,609	60	3.7
30–34 years	1,414	55	3.9
35–39 years	747	36	4.8
40–44 years	144	9	6.3
45 years and over	7	1	14.3
Ethnicity			
Māori	1,322	60	4.5
Pacific	1,072	48	4.5
Asian	1,031	51	4.9
Other	2,307	65	2.8
NZ Deprivation Quintile			
Quintile 1	580	16	2.8
Quintile 2	719	24	3.3
Quintile 3	970	37	3.8
Quintile 4	1,361	57	4.2
Quintile 5	2,101	90	4.3
Unknown	1	-	-
Total	**5,732**	**224**	**3.9**

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 2015 year, 2.8 increased risk results were issued for every 100 screens completed. This was consistent with the rates for previous years.

Table 20: Number and rate per 100 screens of increased risk screening results for trisomy 21, 18 or 13, January 2011 to December 2015

	Number and rate of increased risk screens				
	2011	**2012**	**2013**	**2014**	**2015**
Total increased risk results	1,099	1,156	1,103	1,157	1,163
Positive test rate per 100 screens	2.8	2.8	2.7	2.8	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 2015 year.

Positive test rate increased markedly with increasing age and was 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 have a relationship with the positive test rate.

Table 21: Increased risk screening results for trisomy 21, 18 or 13 by age, ethnicity and deprivation, January 2015 to December 2015

	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	13	1,507	0.9
20–24 years	60	5,988	1.0
25–29 years	129	11,811	1.1
30–34 years	282	14,018	2.0
35–39 years	378	7,418	5.1
40–44 years	284	1,406	20.2
45 years and over	17	64	26.6
Ethnicity			
Māori	132	4,902	2.7
Pacific	103	2,623	3.9
Asian	278	8,114	3.4
Other	650	26,573	2.4
NZ Deprivation Quintile			
Quintile 1	211	7,329	2.9
Quintile 2	248	8,025	3.1
Quintile 3	202	8,318	2.4
Quintile 4	253	9,293	2.7
Quintile 5	249	9,241	2.7
Unknown	0	6	0.0

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 individually as well as the positive test rate for the three trisomies together by trimester of screen and calendar year. The sum of the individual values for trisomy 21, 18 and 13 is greater than the value for the fourth grouping (any of the three trisomies) because a result can be at increased risk for more than one trisomy.

Trisomy 18 and 13 each showed low positive test rates (0.3 and 0.4 per 100 screens respectively) while the positive test rate for trisomy 21 was just below 3 per 100 screens for all years. The second trimester positive test rate for trisomy 21 was higher than the first trimester positive test rate but the difference was not as large for 2015 as it was in previous years. This difference in rates may be due to variability in nuchal translucency, nasal bone and crown rump length assessments. 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 2015

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	1,081	2.8	868	2.5	213	4.8
2012	1,144	2.8	871	2.4	273	5.5
2013	1,081	2.7	840	2.4	241	4.6
2014	1,131	2.7	870	2.4	261	4.8
2015	1,140	2.7	937	2.6	203	3.7
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	136	0.3	120	0.3	16	0.3
2015	145	0.3	127	0.3	18	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	149	0.4	135	0.4	14	0.3
2015	159	0.4	147	0.4	12	0.2
Any one or more of trisomy 21, 18 or 13						
2011	1,099	2.8	878	2.5	221	5.0
2012	1,156	2.8	874	2.4	282	5.7
2013	1,103	2.7	847	2.4	256	4.9
2014	1,157	2.8	883	2.4	274	5.0
2015	1,163	2.8	942	2.6	221	4.0

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 2015 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 2015.

Table 23: Increased risk screening results for trisomy 21, 18 and 13 by risk level, January 2015 to December 2015

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 – 1:20	257	57	69
1:25 to 1:50	168	26	20
1:55 to 1:300	715	62	70

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, include screened women from Canterbury, South Canterbury and West Coast DHBs for the first time. Screening and outcome data was completely re-matched for all years from 2011 to 2015 for this report.

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

Table 24 shows the diagnostic testing rate from 2011 to 2015 by trimester of screen. In 2015, for every 100 women that received an increased risk result after a first trimester screen, 56 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 (45 women per 100 increased risk screens) compared with first trimester screens (59 per 100 increased risk screens). See Appendix 3 for a summary of diagnostic test results for women who had increased risk screen in 2015, as well as pregnancy outcomes (where known) for women who did not have a prenatal diagnostic test.

Table 24: Diagnostic testing volumes for women with increased risk screens by trimester of screen, January 2011 to December 2015

Trimester of screen	Diagnostic tests per 100 increased risk screens				
	2011	**2012**	**2013**	**2014**	**2015**
T1 screen	65.0	66.1	66.2	62.3	59.0
T2 screen	43.4	42.6	46.5	47.4	44.8
Total screens	**60.7**	**60.4**	**61.7**	**58.8**	**56.3**

Diagnostic testing volumes for women with increased risk screens by DHB

The number of diagnostic tests and rate per 100 increased risk screens by DHB is given in Table 25. Many DHBs have low numbers and care should be taken with comparisons. The rate of diagnostic testing for women with increased risk screens in 2015 varied across DHBs from 44 per 100 increased risk results in Taranaki, to 75 per 100 increased risk results in South Canterbury.

Table 25: Diagnostic testing volumes for women with increased risk screens by DHB, January 2011 to December 2015

DHB	Number of diagnostic tests					Tests per 100 increased risk screens				
	2011	2012	2013	2014	2015	2011	2012	2013	2014	2015
Northland	24	13	28	26	21	49.0	38.2	56.0	59.1	48.8
Waitemata	137	138	140	116	107	67.5	67.6	72.9	61.7	57.5
Auckland	117	118	89	89	76	72.2	69.0	67.4	55.3	54.3
Counties Manukau	67	76	73	76	86	54.5	51.4	47.1	50.3	53.8
Waikato	15	26	40	41	42	20.5	38.2	57.1	64.1	60.0
Lakes	15	23	21	21	28	55.6	69.7	67.7	53.8	71.8
Bay of Plenty	11	22	21	21	20	36.7	68.8	55.3	63.6	66.7
Tairawhiti	5	5	2	2	4	83.3	50.0	28.6	33.3	57.1
Hawke's Bay	22	18	21	20	15	62.9	50.0	53.8	58.8	51.7
Taranaki	14	18	18	12	10	63.6	75.0	66.7	48.0	43.5
MidCentral	20	20	10	11	8	54.1	62.5	38.5	57.9	44.4
Whanganui	4	4	6	3	4	33.3	33.3	46.2	60.0	66.7
Capital and Coast	52	61	55	45	66	72.2	69.3	75.3	60.0	61.7
Hutt Valley	14	24	18	15	18	56.0	63.2	58.1	53.6	66.7
Wairarapa	5	7	9	1	2	71.4	100.0	81.8	25.0	40.0
Nelson Marlborough	23	11	17	18	15	67.6	47.8	89.5	78.3	57.7
West Coast	3	2	2	8	3	50.0	50.0	40.0	42.1	50.0
Canterbury	76	66	73	119	82	66.1	60.6	59.8	64.7	50.3
South Canterbury	6	4	4	3	9	54.5	40.0	40.0	50.0	75.0
Southern	37	42	33	33	39	74.0	57.5	63.5	67.3	59.1
Total	**667**	**698**	**680**	**680**	**655**	**60.7**	**60.4**	**61.7**	**58.8**	**56.3**

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 2015. The diagnostic testing rate ranged from 52 per 100 increased risk screens for women aged 20 to 24 years, to 62 per 100 for women aged 30–34 years, with no clear trend with increasing age.

Diagnostic testing rates were highest for women of Asian ethnicity (63 per 100 increased risks), followed by Other (59 per 100 increased risks), with much lower rates for Māori (45 per 100 increased risks) and Pacific (37 per 100 increased risks). Diagnostic testing rates by deprivation have fluctuated over time but there appears to be a consistent difference in rates between least deprived (Quintile 1) and most deprived (Quintile 5) areas, with women in the most deprived areas less likely to have a diagnostic test.

Table 26: Diagnostic testing volumes for women with increased risk screening results by age at screen, ethnicity and deprivation, January 2011 to December 2015

	Diagnostic tests per 100 increased risk screens				
	2011	**2012**	**2013**	**2014**	**2015**
Age at screen					
Under 20 years	50.0	35.7	28.6	50.0	53.8
20–24 years	60.0	56.4	64.5	53.9	51.7
25–29 years	65.4	60.7	60.5	61.7	58.1
30–34 years	65.3	69.6	68.4	65.4	61.7
35–39 years	64.6	60.3	62.4	56.7	56.6
40–44 years	50.2	55.7	57.3	58.1	51.8
45 years and over	43.5	40.0	44.4	33.3	41.2
Ethnicity					
Māori	42.2	43.9	52.5	38.7	44.7
Pacific	35.5	36.4	38.2	39.2	36.9
Asian	70.7	71.0	69.6	67.0	63.3
Other	64.9	64.4	65.2	62.4	58.8
NZ Deprivation Quintile					
Quintile 1	70.2	66.7	71.4	65.8	62.1
Quintile 2	71.2	69.5	65.3	63.9	62.9
Quintile 3	60.7	65.2	62.6	57.6	58.4
Quintile 4	54.2	52.3	58.5	59.7	57.7
Quintile 5	48.0	48.8	53.2	48.8	41.8

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

Each screening result includes a separate risk for each of trisomy 21, 18 and 13. For the analysis in this report women were assigned a combined trisomy risk level based on the highest risk score they received across the three trisomies. Table 27 shows the number of diagnostic tests for women that received an increased risk result during 2015 for one or more of trisomy 21, 18 or 13, stratified by risk level. As this shows, uptake of diagnostic testing was higher at higher risk levels. While 7% of women with a risk between 1:55 and 1:300 had a prenatal diagnostic test, this increased to 76% for women with risks of 1:20 or above.

Table 27: Diagnostic testing volumes for women with increased risk screens by risk level, January 2015 to December 2015

Risk level	Number of diagnostic tests	Number of increased risk screens	Tests per 100 increased risk screens
1:5 to 1:20	201	265	75.8
1:25 to 1:50	112	169	66.3
1:55 to 1:300	342	729	46.9

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

This section reports information on the number and proportion of women who complete prenatal diagnostic testing (CVS or amniocentesis procedures) 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 or an abnormal ultrasound finding. 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 0.69 per 100 low risk screens in 2015. The rate has been consistently below 1 across all years.

Table 28: Diagnostic testing volumes for women with low risk screens by trimester of screen, January 2011 to December 2015

Trimester of screen	Diagnostic tests per 100 low risk screens				
	2011	2012	2013	2014	2015
T1 screen	0.89	0.92	0.77	0.68	0.74
T2 screen	0.81	0.67	0.48	0.56	0.34
Total screens	**0.88**	**0.89**	**0.74**	**0.67**	**0.69**

Diagnostic testing volumes for women with low risk screens by DHB

The rate of diagnostic testing for women with low risk screens has varied each year from 2011 to 2015, as shown in Table 29. Given the low numbers involved, caution should be taken in making comparisons, however rates appear to have decreased over time for Waitemata, Auckland and Counties Manukau DHBs.

Table 29: Total diagnostic testing volumes for women with low risk screens by DHB January 2011 to December 2015

DHB	Number of diagnostic tests					Tests per 100 low risk screens				
	2011	2012	2013	2014	2015	2011	2012	2013	2014	2015
Northland	5	2	7	-	7	0.56	0.20	0.74	-	0.66
Waitemata	64	61	55	34	33	1.08	1.02	0.90	0.55	0.55
Auckland	71	73	54	38	36	1.60	1.63	1.15	0.79	0.80
Counties Manukau	39	25	27	18	22	0.85	0.51	0.57	0.35	0.43
Waikato	6	18	19	30	21	0.17	0.52	0.54	0.80	0.56
Lakes	3	3	3	5	7	0.37	0.34	0.35	0.54	0.74
Bay of Plenty	5	10	9	14	7	0.31	0.56	0.54	0.80	0.38
Tairawhiti	-	3	-	1	-	-	0.95	-	0.29	0.00
Hawke's Bay	11	8	6	7	8	1.00	0.65	0.48	0.59	0.64
Taranaki	6	11	9	3	1	0.67	1.31	1.11	0.33	0.10
MidCentral	7	4	9	8	11	0.70	0.39	0.81	0.72	0.93
Whanganui	4	4	2	2	2	1.24	1.14	0.56	0.47	0.42
Capital and Coast	23	18	21	16	22	0.90	0.67	0.84	0.64	0.87
Hutt Valley	12	10	8	11	9	1.01	0.82	0.66	0.88	0.69
Wairarapa	1	-	-	-	1	0.31	-	-	-	0.31
Nelson Marlborough	9	14	12	6	9	0.71	1.15	1.01	0.49	0.77
West Coast	-	-	1	1	2	-	-	0.37	0.39	0.79
Canterbury	40	46	31	45	52	0.94	1.04	0.67	0.96	1.07
South Canterbury	2	3	1	-	2	0.41	0.57	0.19	-	0.39
Southern	26	37	18	32	30	1.07	1.44	0.71	1.33	1.16
Total	**334**	**350**	**292**	**271**	**282**	**0.88**	**0.89**	**0.74**	**0.67**	**0.69**

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, and for women of Asian or Other ethnicity. Women in the most deprived Quintile appear to have a lower rate of diagnostic testing compared to less deprived areas, but the trend across quintiles is not clear.

Table 30: Diagnostic tests per 100 low risk screens by age, ethnicity and NZ deprivation quintile, January 2011 to December 2015

	Diagnostic tests per 100 low risk screens				
	2011	**2012**	**2013**	**2014**	**2015**
Age at screen					
Under 20 years	0.39	0.71	0.38	0.44	0.33
20–24 years	0.37	0.34	0.32	0.37	0.35
25–29 years	0.38	0.45	0.37	0.49	0.51
30–34 years	0.57	0.64	0.53	0.53	0.60
35–39 years	1.83	1.54	1.21	0.98	1.12
40–44 years	5.40	5.59	5.30	3.92	2.94
45 years and over	7.50	10.64	6.98	2.08	2.13
Ethnicity					
Māori	0.45	0.70	0.57	0.46	0.46
Pacific	0.51	0.33	0.28	0.28	0.48
Asian	0.89	0.87	0.65	0.58	0.80
Other	0.98	0.98	0.84	0.78	0.71
NZ Deprivation Quintile					
Quintile 1	1.45	1.66	1.11	0.91	0.77
Quintile 2	1.10	1.03	0.77	0.73	0.93
Quintile 3	0.81	0.60	0.73	0.65	0.63
Quintile 4	0.70	0.80	0.63	0.62	0.72
Quintile 5	0.39	0.43	0.52	0.49	0.43

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 aggregated rate of diagnostic testing is more than 15 times higher for the highest category compared with the lowest category and the rate drops away rapidly as risk decreases below 1:1000.

Table 31: Diagnostic tests per 100 low risk screens stratified by risk level, January 2011–December 2015 aggregated

Risk level	Number of diagnostic tests	Number of low risk screens	Tests per 100 low risk screens
1:310 to 1:500	185	3,094	5.98
1:510 to 1:1000	253	8,081	3.13
1:1100 to 1:2000	221	14,044	1.57
1:2100 to 1:3000	149	12,156	1.23
1:3100 to 1:4000	79	11,146	0.71
1:4100 to 1:5000	77	9,999	0.77
1:5100 to 1:10,000	222	40,382	0.55
1:11,000 to 1:100,000	351	99,767	0.35

Indicator 8: Diagnostic testing for unscreened women

This section reports information on the number of women who completed prenatal diagnostic testing 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 an increased prior risk (eg, family history, previous child with Down syndrome, late maternal age) or a diagnostic test done for another reason and the karyotype reported or an abnormal ultrasound finding.

Diagnostic volumes for unscreened women

During the 2015 year, 252 diagnostic tests were completed for unscreened women. This is similar to the number undertaken in previous years. Table 32 shows the number of tests by DHB and Table 33 shows the breakdown by age, ethnicity and NZ deprivation quintile.

Table 32: Diagnostic testing volumes for unscreened women by DHB, January 2012 to December 2015

DHB	Number of diagnostic tests			
	2012	2013	2014	2015
Northland	10	6	7	8
Waitemata	37	24	22	22
Auckland	31	23	25	18
Counties Manukau	19	27	21	18
Waikato	16	24	14	15
Lakes	2	5	6	8
Bay of Plenty	10	18	12	14
Tairawhiti	5	-	1	3
Hawke's Bay	11	6	7	7
Taranaki	13	11	5	11
MidCentral	9	11	11	8
Whanganui	4	2	3	2
Capital and Coast	17	16	30	36
Hutt Valley	9	11	11	22
Wairarapa	5	1	1	3
Nelson Marlborough	7	1	4	6
West Coast	-	1	1	-
Canterbury	27	23	37	30
South Canterbury	-	2	4	2
Southern	17	18	13	19
Total	**249**	**203**	**235**	**252**

Table 33: Total diagnostic testing volumes for unscreened women by age, ethnicity and deprivation quintile, January 2012 to December 2015

	Number of diagnostic tests			
	2012	**2013**	**2014**	**2015**
Age				
Under 20 years	15	13	10	16
20–24 years	32	33	29	19
25–29 years	43	35	39	53
30–34 years	62	56	66	70
35–39 years	55	50	54	54
40–44 years	41	39	34	35
45 years and over	1	4	3	5
Ethnicity				
Māori	33	49	31	44
Pacific	17	14	20	21
Asian	39	31	29	33
Other	160	136	155	154
NZ Deprivation Quintile				
Quintile 1	62	36	55	48
Quintile 2	45	47	39	48
Quintile 3	40	40	49	51
Quintile 4	58	59	46	52
Quintile 5	44	48	46	53

Diagnostic results for unscreened women

A breakdown of prenatal diagnostic testing results for unscreened women for the 2015 year is given in Table 34. Of the 252 diagnostic tests in 2015 for unscreened women, 191 (76%) had a normal karyotype. There were thirteen trisomy 21 diagnoses, six trisomy 18 diagnoses and five diagnoses of trisomy 13.

Table 34: Total diagnostic testing results for unscreened women, January 2015 to December 2015

Karyotype result	Number	Percentage
Normal karyotype	191	75.8%
Trisomy 21	13	5.2%
Trisomy 18	6	2.4%
Trisomy 13	5	2.0%
Turner syndrome	6	2.4%
Triploidy	8	3.2%
Other chromosome abnormality	18	7.1%
Failed test	5	2.0%
Total	**252**	**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 positives 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 for any of these three trisomies it was classified as 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 2015 was 0.11, slightly higher than the 2014 result, but lower than the highest PPV result of 0.13 in 2013 (see Table 35). A value of 0.11 means that if a woman receives an increased risk result for trisomy 21, 18 or 13 there is an 11% probability that she is carrying a fetus with 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 2015

Year	True positives	False positives	PPV
2011	135	964	0.12
2012	141	1,015	0.12
2013	139	964	0.13
2014	121	1,036	0.10
2015	126	1,037	0.11

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21 the PPV for 2015 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 2015

Year	True positives	False positives	PPV
2011	88	993	0.08
2012	95	1,049	0.08
2013	109	972	0.10
2014	89	1,042	0.08
2015	95	1,045	0.08

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

The combined PPV for trisomies 13 or 18 for 2015 was higher than the trisomy 21 PPV at 0.17 (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 2015

Year	True positives	False positives	PPV
2011	43	127	0.25
2012	38	148	0.20
2013	28	150	0.16
2014	27	144	0.16
2015	30	149	0.17

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. Data have been aggregated across the 2011 to 2015 period. Women that received a very increased risk result of 1:5 to 1:20 for trisomy 21 had a 27% probability that they were carrying a fetus with trisomy 21. 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.
There is discordance between the PPV and the reported risk estimates, i.e. the reported risk is lower than the observed risk (see ratios added next to the PPV values in table 38). The reason for this will be explored in future reports.

Table 38: Positive predictive of screening for trisomy 21 stratified by risk level, aggregated 2011 – 2015

Risk level	True positives	False positives	PPV	PPV as a ratio
1:5 to 1:20	341	903	0.27	1:3.7
1:25 to 1:50	65	747	0.08	1:125
1:55 to 1:300	70	3,451	0.02	1:200

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. The aggregated PPV for 2011 to 2015 was highest for the 35-39 and 40–44 years age groups.

Table 39: Positive predictive of screening for trisomy 21 by age, aggregated 2011 – 2015

Age group	True positives	False positives	PPV
Under 20 years	3	57	0.05
20 – 24 years	13	294	0.04
25 – 29 years	33	565	0.06
30 – 34 years	91	1,149	0.07
35 – 39 years	195	1,736	0.10
40 – 44 years	135	1,192	0.10
45 years and over	6	108	0.05

The number of true and false positive results by ethnicity is shown in Table 40. Aggregating data across all years gives a PPV of 0.06 (6%) for Māori, 0.03 (3%) for Pacific, 0.05 (5%) for Asian, and 0.11 (11%) for women of Other ethnicity.

Table 40: Positive predictive of screening for trisomy 21 by ethnicity, aggregated 2011 – 2015

Ethnicity	True positives	False positives	PPV
Māori	34	576	0.06
Pacific	15	523	0.03
Asian	53	1,102	0.05
Other	374	2,900	0.11

Table 41 shows PPV by NZ deprivation quintile. There appears to be a relationship between PPV and deprivation with higher PPV values for women in areas of lower deprivation.

Table 41: Positive predictive of screening for trisomy 21 by NZ deprivation quintile, aggregated 2011 – 2015

NZ Deprivation Quintile	True positives	False positives	PPV
Quintile 1	129	965	0.12
Quintile 2	107	957	0.10
Quintile 3	86	975	0.08
Quintile 4	89	1,053	0.08
Quintile 5	65	1,151	0.05

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 2015 was 0.02 (or 2%). This means that out of all women who had a negative diagnostic or a baby without a trisomy, 2% received an increased risk result for trisomy 21, 18 or 13.

Table 42: False positive rate for trisomy 21, 18 or 13, January 2011 to December 2015

Year	False positives	True negatives	False positive rate
2011	964	38,045	0.02
2012	1,015	39,452	0.03
2013	964	39,589	0.02
2014	1,036	40,551	0.02
2015	1,037	41,030	0.02

The false positive rate was higher for second trimester screens than for first trimester screens, consistent with previous years.

Table 43: False positive rate for trisomy 21, 18 or 13 by trimester of screen, January to December 2015

Trimester	False positives	True negatives	False positive rate
T1 screens	826	35,744	0.02
T2 screens	211	5,286	0.04
Total screens	**1,037**	**41,030**	**0.02**

The false positive rate for trisomy 21 when considered alone was similar to the overall false positive rate (see Table 44). However, the combined false positive rate for trisomy 18 and trisomy 13 is much lower (0.004 for 2015, see Table 45).

Table 44: False positive rate for trisomy 21, January 2011 to December 2015

Year	False positives	True negatives	False positive rate
2011	993	38,075	0.03
2012	1,049	39,477	0.03
2013	972	39,626	0.02
2014	1,042	40,587	0.03
2015	1,045	41,060	0.02

Table 45: False positive rate for trisomy 18 and 13, January 2011 to December 2015

Year	False positives	True negatives	False positive rate
2011	127	38,995	0.003
2012	148	40,441	0.004
2013	150	40,538	0.004
2014	144	41,550	0.003
2015	149	42,025	0.004

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

False positive rates by age, ethnicity and NZ deprivation quintile are shown in Table 46. The false positive rate for trisomy 21 increases with age. For example, the false positive rate for women under 20 years in 2015 was 0.01 (1%) compared with 0.32 (32%) for women 45 years and older. 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. This difference has been consistent over time.

Table 46: False positive rate for trisomy 21 by age, aggregated January 2011 to December 2015

Age group	2011	2012	2013	2014	2015
Under 20 years	0.01	0.01	0.00	0.01	0.01
20 – 24 years	0.01	0.01	0.01	0.01	0.01
25 – 29 years	0.01	0.01	0.01	0.01	0.01
30 – 34 years	0.02	0.02	0.02	0.02	0.02
35 – 39 years	0.05	0.05	0.05	0.05	0.05
40 – 44 years	0.16	0.16	0.15	0.15	0.18
45 years and over	0.33	0.33	0.37	0.31	0.27

The false positive rate for 2015 varied across ethnic groups from 0.02 (2%) for Māori and Other to 0.04 (4%) for Pacific. These rates are consistent with previous years.

Table 47: False positive rate for trisomy 21 by ethnicity, January 2011 to December 2015

Ethnicity	2011	2012	2013	2014	2015
Māori	0.02	0.02	0.02	0.02	0.02
Pacific	0.04	0.04	0.04	0.04	0.04
Asian	0.03	0.03	0.03	0.03	0.03
Other	0.02	0.02	0.02	0.02	0.02

False positive rate was relatively consistent across deprivation levels with rates between 2% and 3% for 2015 and previous years (see Table 48).

Table 48: False positive rate for trisomy 21 by NZ deprivation quintile, January 2011 to December 2015

NZ Deprivation Quintile	2011	2012	2013	2014	2015
Quintile 1	0.03	0.03	0.03	0.03	0.02
Quintile 2	0.03	0.03	0.02	0.02	0.03
Quintile 3	0.02	0.03	0.02	0.03	0.02
Quintile 4	0.02	0.02	0.02	0.02	0.02
Quintile 5	0.03	0.03	0.03	0.03	0.02

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 positive results (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 positive and false negative results (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 negative results stratified by risk is given in Appendix 5, and the receiver operating characteristic (ROC) curve of detection rate against the 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 the 5 years ending 2015 is given in table 49. Rates for trisomy 21 alone, and for trisomies 18 and 13 together are given in tables 50 and 51 respectively. As each of these tables show, detection rates increased between 2014 and 2015. These changes may be related to quality improvements undertaken with radiology practices from 2015, but may also be related to other factors, such as improved completion of screening lab forms (e.g. inclusion of mother's weight on a greater proportion of forms), or could be partially due to random fluctuation, given the relatively low numbers involved in the calculation of detection rates. The addition of a further data point in next year's report will give a clearer trend.

The overall detection rate for trisomy 21, 18 and 13 for 2015 was 0.87 (87%). This was higher than all previous years (see Table 49). A detection rate of 0.87 means that there is an 87% 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 49: Detection rate for trisomy 21, 18 or 13, January 2011 to December 2015

Year	True positives	False negatives	Detection rate
2011	135	37	0.78
2012	141	40	0.78
2013	139	41	0.77
2014	121	28	0.81
2015	126	19	0.87

The detection rate for trisomy 21 alone is shown in Table 50. The rate for 2015 was slightly higher (0.89) than the overall rate for trisomy 21, 18 and 13. The detection rate for trisomy 13 and 18 was lower at 0.79 for 2015 (see Table 51, over page).

Table 50: Detection rate for trisomy 21, January 2011 to December 2015

Year	True positives	False negatives	Detection rate
2011	88	25	0.78
2012	95	27	0.78
2013	109	26	0.81
2014	89	18	0.83
2015	95	12	0.89

Table 51: Detection rate for trisomy 13 or 18, January 2011 to December 2015

Year	True positives	False negatives	Detection rate
2011	43	16	0.73
2012	38	21	0.64
2013	28	17	0.62
2014	27	15	0.64
2015	30	8	0.79

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

Due to the low number of true positives and false negative results for some groups the detection rates for trisomy 21 have been calculated in aggregate across the five years in order to present more stable rates. Numbers for the youngest and oldest age groups are still very low after aggregation so care should be taken with interpretation of these. Across the other age groups the detection rate for trisomy 21 appears to increase with age from 0.59 (59%) for women 20–24 years to 0.95 (95%) for women 40-44 years (see Table 52).

Table 52: Detection rate for trisomy 21 by age, aggregated 2011 – 2015

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	3	5	-
20–24 years	13	9	0.59
25–29 years	33	15	0.69
30–34 years	91	40	0.69
35–39 years	195	32	0.86
40–44 years	135	7	0.95
45 years and over	6	0	-

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

The aggregated detection rates by ethnicity ranged from 0.75 (75%) for Pacific to 0.83 (83%) for women of Other ethnicity (see Table 53). Low numbers mean these rates should be interpreted with caution.

Table 53: Detection rate for trisomy 21 by ethnicity, aggregated 2011 – 2015

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	34	10	0.77
Pacific	15	5	0.75
Asian	53	16	0.77
Other	374	77	0.83

The aggregated detection rates by deprivation quintile ranged from 0.78 to 0.84 (see Table 54). There was no clear trend with increasing deprivation.

Table 54: Detection rate for trisomy 21 by NZ deprivation quintile, aggregated 2011 – 2015

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	129	24	0.84
Quintile 2	107	25	0.81
Quintile 3	86	18	0.83
Quintile 4	89	25	0.78
Quintile 5	65	16	0.80

Indicators 12, 13 & 14: Radiology monitoring Nuchal Translucency (NT) ultrasound volumes by NT operator

In 2015, the NSU introduced a quality improvement initiative for radiology. This included a statistical service to provide radiology practices, reporting radiologists and NT operators with feedback on the quality of their paired NT and CRL measurements provided as part of antenatal screening for DSOC. Individual ultrasound operators, reporting radiologists and practices received reports for 2014 and 2015 on their NT and CRL measurements during the period.

The reports showed the quality of their measurements in terms of bias, spread and trend when compared against the Fetal Medicine Foundation (FMF) reference curve. Results were assigned either a Green Flag (where the results are good), or an Amber or Red Flag (where action is required to improve the quality of their scans). The reports, which will be sent out for every 6 months from January 2016, included a plotted graph of paired NT and CRL measurements against the FMF reference curve[3] as well as summary and explanation of the data.

The data within each graph was assessed to indicate performance in three key areas:

1. Bias – The difference between the observed NT measurements and those we would expect from the FMF curve.
2. Spread – The way most measurements cluster along the FMF curve.
3. Trend – The shape and direction of the curve of observed NT and CRL measurements relative to that of the FMF reference curve.

Further detailed definitions for summary measures of bias, spread and trend are presented in appendix 7.

Table 55 shows the number of ultrasound scans received for radiology monitoring for 2014 and 2015. A large proportion of scans (56%) did not have the individual operator identified which limited the analysis that could be completed. Radiology practices were reminded late in 2015 of the requirement to include the name of the NT operator on ultrasound scan reports and it is expected that the proportion of unknown operators will decrease for future reporting rounds.

[3] More information about the FMF can be found at: https://fetalmedicine.org/

Table 55: Ultrasound scan data received for radiology monitoring, January 2014 to December 2015

	2014	2015
Total NT scan results received	39,563	39,703
Total records by unidentified NT operators	10,346 (26%)	10,468 (26%)
Number of radiologists reported	232	248
Number of NT operators reported	346	407

Indicator 12: Nuchal Translucency (NT) ultrasound volumes by NT operator

This indicator looks at the number of ultrasound scans for nuchal translucency (NT) performed by each ultrasound operator in a period. This is important because a minimum number of scans are required in order for valid statistical analysis to be undertaken to assess the quality of the ultrasound operator's performance, and because it is assumed that the proficiency is likely to be linked to the volume of scans performed. For 2014 and 2015, the number of scans performed by each operator was assessed over 12 month period, and the threshold used for the analysis was 26 scans. As table 56 shows, 51% of operators met this threshold in 2014, and this increased to 56% for the 2015 year.

Table 56: NT volumes by operator, January 2014 to December 2015

Year	0-10 scans		11-25 scans		26+ scans	
2014	138	38%	38	11%	183	51%
2015	143	35%	38	9%	226	56%

Those operators that did not perform 26 or more scans during the calendar year still had their data analysed but were assigned a White Flag to indicate that the results were not deemed statistically significant. For the 2016 year onwards the threshold for coloured flag status will move to 25 or more scans per 6 month period.

Indicator 13: Distribution of bias by NT operator

This indicator reports information on the number of nuchal translucency (NT) ultrasound operators whose measurements were within specific ranges. Quality can be determined by assessing NT measurements by individual operators over time by bias, deviation of multiple of the median (MoM) and spread against the Fetal Medicine Foundation (FMF) reference curve. Over or under measuring of NT impacts the positive test rate and detection rate for women screened as outlined in table 57.

Table 57: Impact of measurement bias on screening risk result

Bias	Description	Effect on risk result
Negative	Points tend to lie below the FMF curve	Risk estimate is lower
Positive	Points tend to lie above the FMF curve	Risk estimate is higher

The most common variance from the FMF reference curve is a tendency to under or over measure the NT. The effect of this on the ultrasound practitioners report is to shift NT measurements downwards or upwards relative to the FMF reference curve. An estimate of the overall bias relative to the FMF reference curve was given in each operator's report. This was accompanied by a flag categorising the bias as Red, Amber or Green as defined in Table 58 below. An example report is given in Appendix 8.

Table 58: Flag status definitions

Flag type	Flag	Bias
Green flag		Assigned when NT Bias relative to FMF reference curve is less than or equal to 0.10mm
Amber flag		Assigned when NT Bias relative to FMF reference curve is between 0.1 and 0.30mm
Red flag		Assigned when NT Bias relative to FMF reference curve is greater than 0.30mm

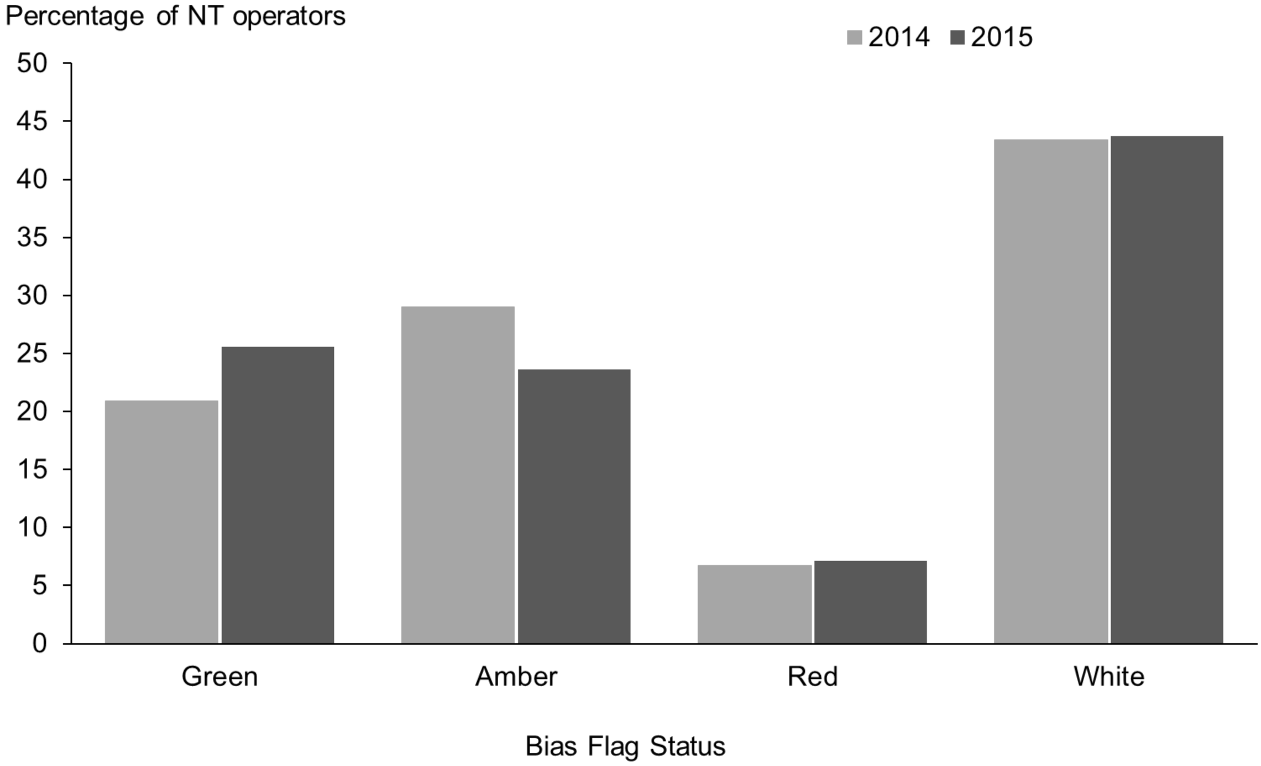
Table 59 shows a breakdown of NT bias by operator for 2014 and 2015. A coloured flag bias status was only assigned were sufficient scans were performed. Percentages refer to the proportion of all operators that scans were received for. There was an improvement in the proportion of operators with a green flag for bias from 2014 (21%) to 2015 (26%). This corresponded with a decrease in amber flagged operators, while the proportion with a red flag for bias stayed constant at 7%.

Table 59: Distribution of bias by NT operator, January 2014 to December 2015

Year	Green		Amber		Red	
2014	75	21%	104	29%	24	7%
2015	104	26%	96	24%	29	7%

The improvement in the proportion of NT operators performing at an acceptable level of bias (green flag status) can be seen in figure 12, below. It should be noted that both sets of results were largely generated before the first round of feedback was distributed to radiology practices. Further improvement is therefore expected for 2016.

Figure 12: Distribution of bias by NT operator, January 2014 to December 2015



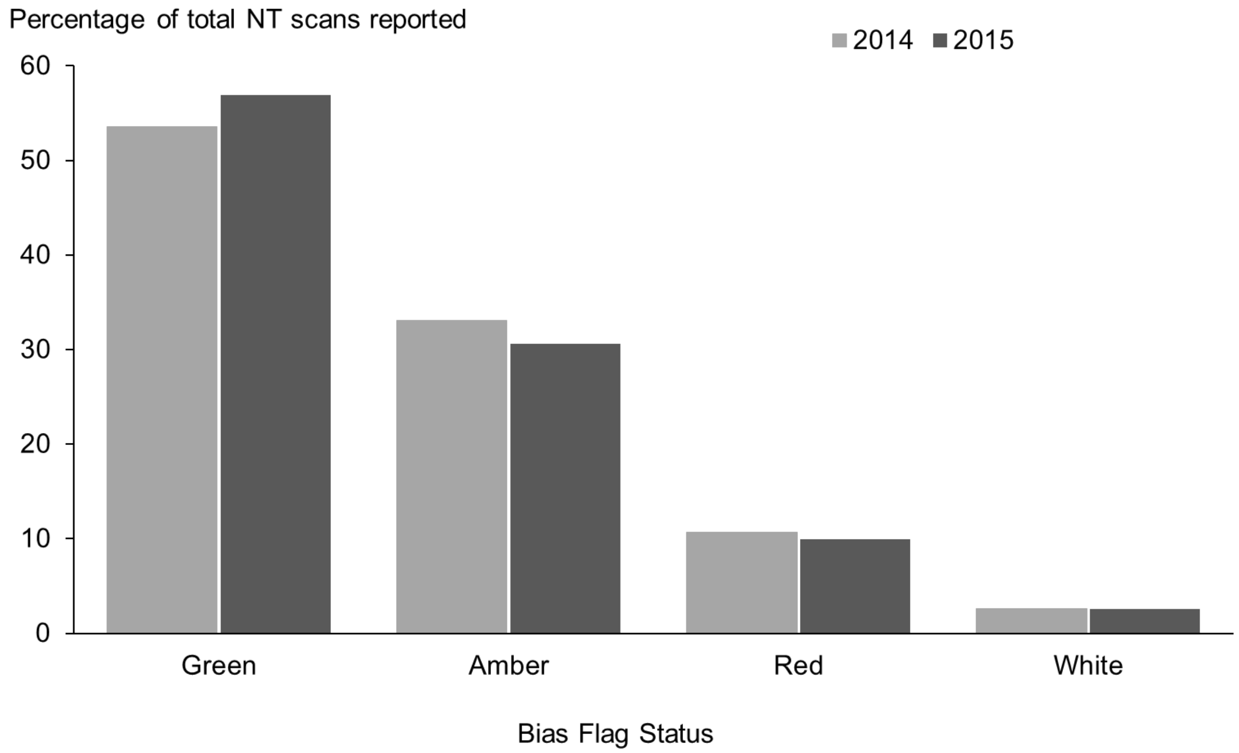
Indicator 14: Overall distribution of bias

The final radiology monitoring indicator reports on the overall distribution of bias in NT scans that were reported as part of antenatal screening. Table 60 gives a breakdown of the proportion of all NT scans undertaken in 2014 and 2015 that were assigned to each bias category. For 2015 57% of women received a scan from a green flagged practitioner, up 3% from 2014.

Table 60: Distribution of bias as a proportion of total scans reported, January 2014 to December 2015

Year	Green		Amber		Red	
2014	21,207	54%	13,092	33%	4,216	11%
2015	22,594	57%	12,166	31%	3,949	10%

Figure 13: Distribution of bias as a proportion of total NT scans reported, January 2014 to December 2015



Appendix 1: Indicator definitions

Table 61: 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
Indicator 12: NT volumes by operator	Number of ultrasound scans for nuchal translucency (NT) performed by each NT operator within the period

Indicator 13: Distribution of bias by NT operator	Distribution of NT Bias relative to FMF reference curve by operator. Report presents number and proportion of operators in bias categories of Green (bias relative to FMF reference curve less than or equal to 0.10mm), Amber (bias between 0.10mm and 0.30mm) and Red (bias greater than 0.30mm)
Indicator 14: Overall distribution of bias	Distribution of NT Bias relative to FMF reference curve for each NT scan. Number and proportion of NT scans in bias categories of Green (bias relative to FMF reference curve less than or equal to 0.10mm), Amber (bias between 0.10mm and 0.30mm) and Red (bias greater than 0.30mm)

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 in the outcome set 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 62: Live births and still births by district health board 2011–2015

DHB	2011	2012	2013	2014	2015
Northland	2,302	2,300	2,129	2,099	2,135
Waitemata	7,881	7,969	7,653	7,850	7,554
Auckland	6,540	6,704	6,244	6,302	5,902
Counties Manukau	8,740	8,767	8,168	8,283	8,206
Waikato	5,390	5,483	5,227	5,252	5,287
Lakes	1,589	1,558	1,417	1,393	1,509
Bay of Plenty	2,859	2,968	2,752	2,782	2,798
Tairawhiti	744	738	711	698	742
Hawkes Bay	2,259	2,259	2,161	2,068	2,002
Taranaki	1,566	1,559	1,524	1,518	1,514
MidCentral	2,300	2,152	2,120	2,094	2,112
Whanganui	829	874	826	817	816
Capital and Coast	3,860	3,869	3,627	3,528	3,534
Hutt Valley	2,056	2,006	1,914	1,853	1,967
Wairarapa	530	510	501	473	463
Nelson Marlborough	1,650	1,529	1,549	1,419	1,417
West Coast	405	408	375	351	356
Canterbury	6,064	5,987	5,825	6,004	6,210
South Canterbury	571	648	639	653	659
Southern	3,673	3,597	3,446	3,285	3,414
Total	**61,808**	**61,885**	**58,808**	**58,722**	**58,597**

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

Table 63: Live births and still births by age group, 2011–2015

Age group	2011	2012	2013	2014	2015
Under 20	4,055	3,908	3,324	2,997	2,786
20–24	11,704	11,465	10,801	10,279	9,952
25–29	15,548	15,936	15,282	15,700	15,732
30–34	17,223	17,460	16,768	17,574	17,913
35–39	10,728	10,406	10,040	9,683	9,762
40–44	2,405	2,578	2,436	2,344	2,300
45 and over	126	120	143	132	139
Unknown	19	12	14	13	13
Total	**61,808**	**61,885**	**58,808**	**58,722**	**58,597**

Table 64: Live births and still births by 2013 NZ deprivation quintile, 2011–2015

NZ Deprivation Quintile	2011	2012	2013	2014	2015
Quintile 1	8,510	8,680	8,177	8,471	8,241
Quintile 2	9,505	9,606	9,248	9,160	9,342
Quintile 3	11,147	11,173	10,627	10,557	10,592
Quintile 4	13,809	13,658	13,423	13,285	13,244
Quintile 5	18,813	18,750	17,301	17,224	17,063
Unknown	24	18	32	25	115
Total	**61,808**	**61,885**	**58,808**	**58,722**	**58,597**

Table 65: Live births and still births by ethnicity, 2011-2015

Ethnicity	2011	2012	2013	2014	2015
Māori	15,829	15,694	14,560	14,232	14,543
Pacific	7,067	6,872	6,342	6,154	6,056
Asian	7,134	8,450	8,155	9,199	9,210
Other	31,778	30,869	29,751	29,137	28,788
Total	**61,808**	**61,885**	**58,808**	**58,722**	**58,597**

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 1,163 screens that had an increased risk for trisomy 21, 18 or 13 during 2015, 655 (56%) had a prenatal diagnostic test (CVS or Amniocentesis) and 508 (44%) did not. Table 66 shows the diagnostic testing results for the 655 prenatal tests, of which 164 had an abnormal karyotype, including 89 confirmed with Down syndrome. Table 67 shows a breakdown of pregnancy outcomes for the 508 women that had an increased risk screen but did not have a prenatal diagnostic test.

Table 66: Diagnostic results for women that accessed a prenatal diagnostic test following an increased risk screen for trisomy 21, 18 or 13 during the 2015 year

Karyotype result	Number	Percentage
Normal karyotype	491	75.0%
Confirmed Down syndrome	89	13.6%
Other result*	75	11.5%
Total	**655**	**100.0%**

* The 75 'Other' results were made up of the following:

Result	Number
Trisomy 18	19
Trisomy 13	5
Turner syndrome	10
Triploidy	4
Sex chromosome aneuploidy (other than non-mosaic 45, X)	3
Partial aneuploidy (autosome) (including mosaic)	7
Uniparental disomy	1
Structural abnormality	2
Apparently balanced chromosome rearrangement	24
Total	**75**

Table 67: 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 2015 year

Result	Number
No abnormality detected on postnatal diagnostic test	13
Trisomy 21	6
Trisomy 18	5
Trisomy 13	2
Turner syndrome	1
Triploidy	2
Sex chromosome aneuploidy (other than non-mosaic 45, X)	1
Autosomal trisomy (other than 13, 18, 21) (including mosaic)	1
Uniparental disomy	8
No link to a diagnosis	469
Total	**508**

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 14: 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.

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 (Disease | 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 2015

Figure 15 shows the data break down in relation to trisomy 21 for women screened during 2015.

Figure 15: Categorisation of trisomy 21 screening results 2015

	Trisomy 21 diagnosed	No trisomy 21 diagnosis	Total
Screen result = Increased risk	A = 95	B = 1,045	A + B = 1,140
Screen result = Low risk	C = 12	D = 41,060	C + D = 41,072
	A + C = 107	B + D = 42,105	N = 42,212 (total screens)

Positive predictive value (indicator 9)

PPV = A/(A+B)
= 95 / 1,140
= 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)
= 1,045 / 42,105
= 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)
= 95 / 107
= 0.89 (or 89%)

There is an 89% 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 165 false negative screens in total across the 5 year 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 68 shows the number of false negatives for each of the five calendar years broken down by the screening risk result in the first group of columns. The next group of columns gives the number of false negatives as a percentage of all negative (low risk) screens. Overall, false negative screens made up less than 0.1% of all negative screens for each of the years from 2011 to 2015.

Table 68: False negative screens for trisomy 21, 18 and 13 by risk level, January 2011 to December 2015

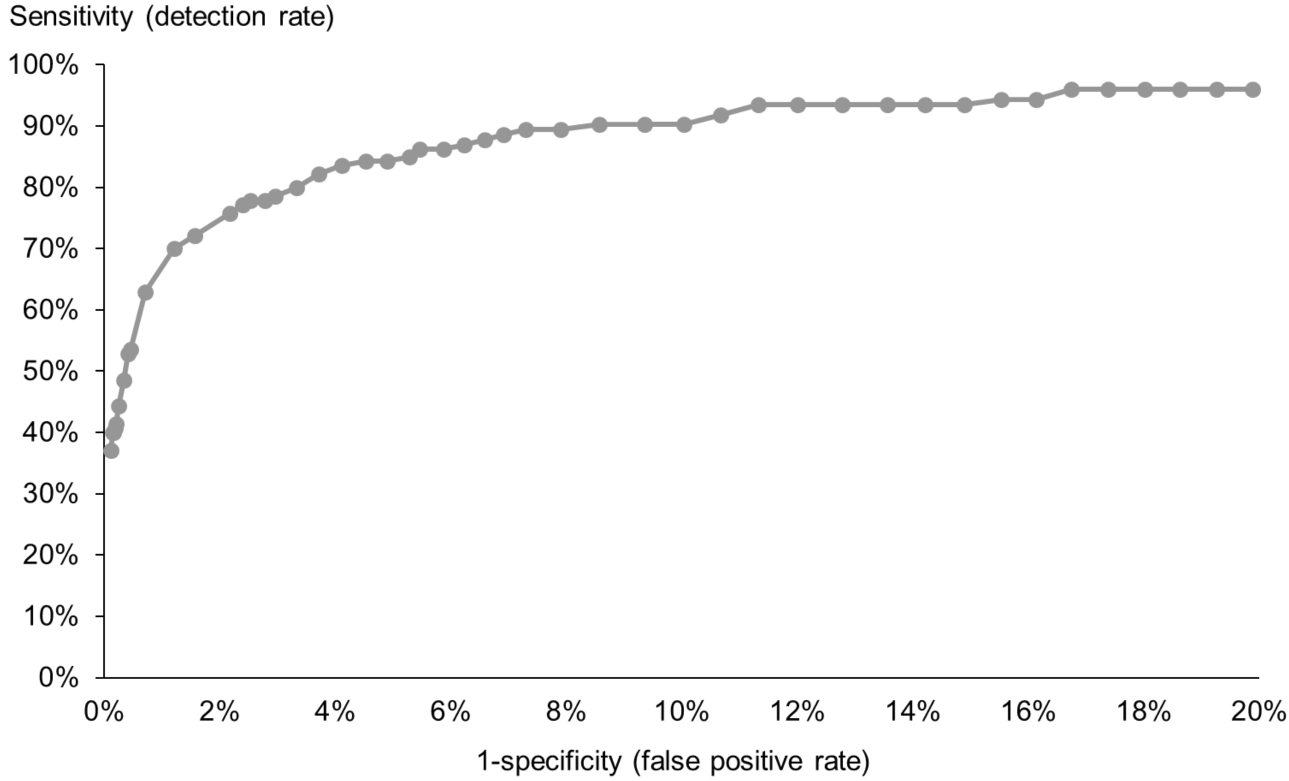
Risk level	False negatives					% of negative screens that are false negatives				
	2011	2012	2013	2014	2015	2011	2012	2013	2014	2015
1:301 to 1:500	10	7	8	7	4	1.83	1.13	1.26	1.07	0.62
1:510 to 1:1,000	7	6	7	6	4	0.44	0.38	0.45	0.37	0.23
1:1100 to 1:2000	7	8	7	5	2	0.26	0.29	0.25	0.17	0.07
1:2100 to 1:3000	3	4	4	3	3	0.13	0.16	0.17	0.12	0.12
1:3100 to 1:4000	0	3	4	0	1	-	0.14	0.18	-	0.04
1:4100 to 1:5000	4	2	0	2	1	0.20	0.10	-	0.10	0.05
1:5100 to 1:10,000	4	5	6	1	2	0.05	0.06	0.08	0.01	0.02
Less than 1:10,000	2	5	5	4	2	0.01	0.03	0.02	0.02	0.01
Total	**37**	**40**	**41**	**28**	**19**	**0.10**	**0.10**	**0.10**	**0.07**	**0.05**

Appendix 6: ROC curve

Figure 16 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 2015 was 87%, 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 89.6%, the false positive rate would increase from 2.4% to 4.7%. This occurs at a risk cut off of 1:600.

Figure 16: ROC curve for trisomy 21, 18 and 13 screening 2015



Appendix 7: Radiology indicator summary measures

Bias – the bias is the difference between observed NT measurements and those we would expect from the FMF curve. For example, the expected NT measurement for a CRL of 60mm is 1.65mm. If a fetus with a CRL measurement of 60mm has an NT measurement of 2mm, the difference is 0.35mm

2mm – 1.65mm = 0.35mm

Or if the measured NT is 1mm, the difference is -0.65mm

1mm – 1.65mm = -0.65mm

The figures shown in the distribution plot are the number and percentage of measurements above and below the FMF curve.

Spread – the spread of NT measurements is the way most measurements will cluster along the FMF curve. The number shown is the factor by which the spread is increased (the measurements vary more greatly than would be expected given the CRL) or decreased (the measurements cluster very tightly around the curve, without the expected normal variance).

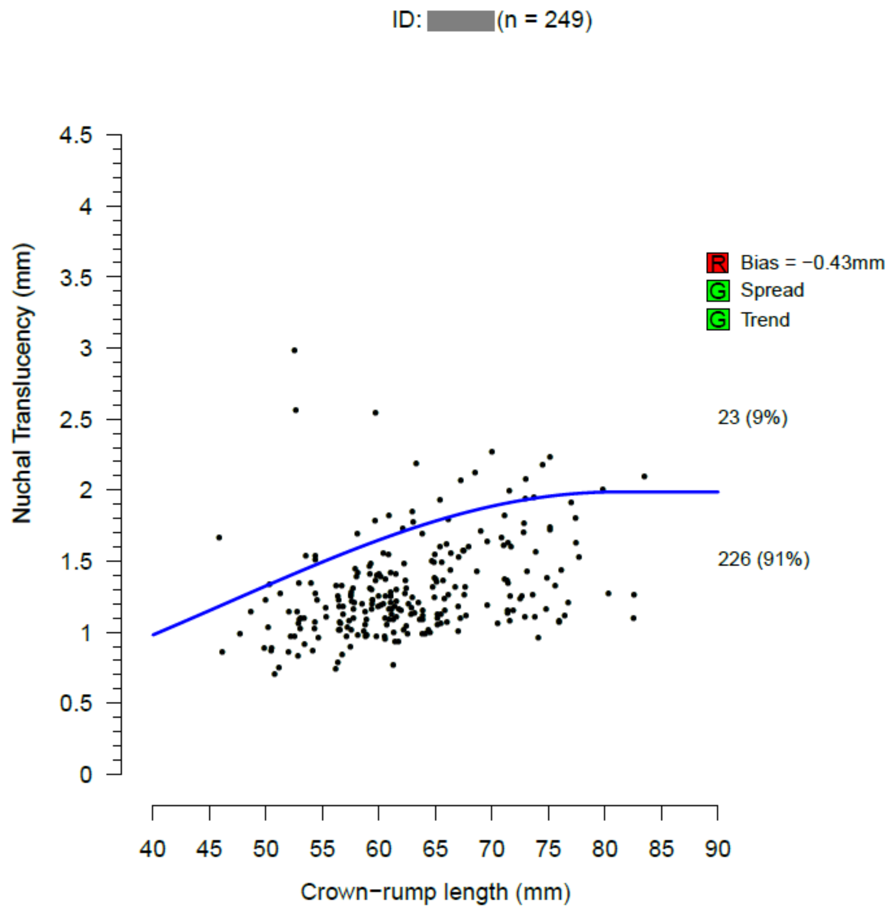
Trend – the curve of observed NT /CRL values should mimic the FMF curve in shape and direction. The value displayed shows the degree of discrepancy between the expected trend and the observed measurements.

Appendix 8: Example NT Operator Report

Figure 17 shows an example NT operator report with identifying codes removed. The report states the time period it relates to (6 January 2014 to 31 December 2014) and the number of NT reports completed by the operator (n = 249). The graph is a scatter plot of nuchal translucency measurement versus crown rump length measurement. The blue line in the graph is the Fetal Medicine Foundation reference curve and the black dots plot where each of the operator's ultrasounds sit in relation to the curve. Each operator is assessed on the amount of bias, spread, and trend. In this example, the operator has a red flag for bias (with a bias value of negative 0.43mm), a green flag for spread, and a green flag for trend. The comment below the graph provides interpretation of these results.

Figure 17: Example NT operator report

Report date:	27/10/15	Practice ID	[redacted]
From:	06/01/14	Frequency	246 1 2
To:	31/12/14		



Overall, there is evidence of a substantial negative bias relative to the FMF reference curve; NT measurements are lower than expected.

Appendix 9: 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.

Fetal Medicine Foundation (FMF) –a Registered Charity that aims to improve the health of pregnant women and their babies through research and training in fetal medicine. Further information can be found at: https://fetalmedicine.org

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.

Multiple of the median (MoM) – a measure of how far an individual result deviates from the median. MoM is commonly used to report the results of medical screening tests, particularly where the results of the individual tests are highly variable.

Nasal bone (NB)- an assessment of nasal bone will be included in the risk calculation if it is reported at the same time as the NT measurement.

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