MINISTRY OF HEALTH
MANATŪ HAUORA

New Zealand Government

Antenatal Screening for Down Syndrome and Other Conditions

2016 Monitoring Report

National Screening Unit

Released 2018 **health.govt.nz**

Citation: Ministry of Health. 2018. *Antenatal Screening for Down Syndrome and Other Conditions: 2016 Monitoring Report*. Wellington: Ministry of Health.

Published in September 2018
by the Ministry of Health
PO Box 5013, Wellington 6140, New Zealand

ISBN 978-1-98-853982-9 (online)
HP 6912

This document is available at health.govt.nz

Contents

Appendices

List of Tables

List of Figures

Executive summary

This report presents data on antenatal screening for Down syndrome and other conditions for the six calendar years from 1 January 2011 to 31 December 2016, and is based on screens that commenced during that time. This is the second year 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 between 12–13 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 2016

- Screening was commenced for 81% of pregnancies [indicator 1].
- Screening uptake by Māori and Pacific women was half or less the rate of Other women in 2016. Pacific and Māori rates have increased each year since 2011 [indicators 1 and 2].
- The national screening completion rate has increased each year with 73% of births being screened in 2016. First trimester screens made up 86% of all completed screens in 2016 [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].
- Nine percent of screens commenced in 2016 were not completed and nearly all related to screens commenced in the first trimester. The rate of incomplete screens was higher for Māori and Pacific women, and for women from areas of higher deprivation [indicator 4].
- The overall positive test rate (number of increased risk results per 100 screens) for trisomy 21, 18 and 13 was 2.7 in 2016, similar to 2015 (2.8). The positive test rate was higher for second trimester screens (3.7 per 100 screens) than for first trimester screens (2.6 per 100 screens) for 2016 [indicator 5].

- The overall false positive rate for trisomy 21, 18 and 13 was 2.0% in 2016, consistent with previous years. The rate was higher for second trimester screens (4.0%) than for first trimester screens (2%) [indicator 10].
- The overall detection rate for trisomy 21, 18 and 13 was 79% in 2016, compared to 84% in 2015 [indicator 11].

Introduction

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

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

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

- first trimester combined screening, which includes a blood test that measures two maternal serum markers, pregnancy-associated protein A (PAPP-A) and free beta- human chorionic gonadotropin (ßhCG). The blood sample is collected between 9 weeks and 13 weeks and 6 days gestation and combined with an ultrasound scan to determine nuchal translucency (NT) and crown rump length (CRL) measurements (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 north of Taupo) and Canterbury Health Laboratories at Canterbury District Health Board (for samples from 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:

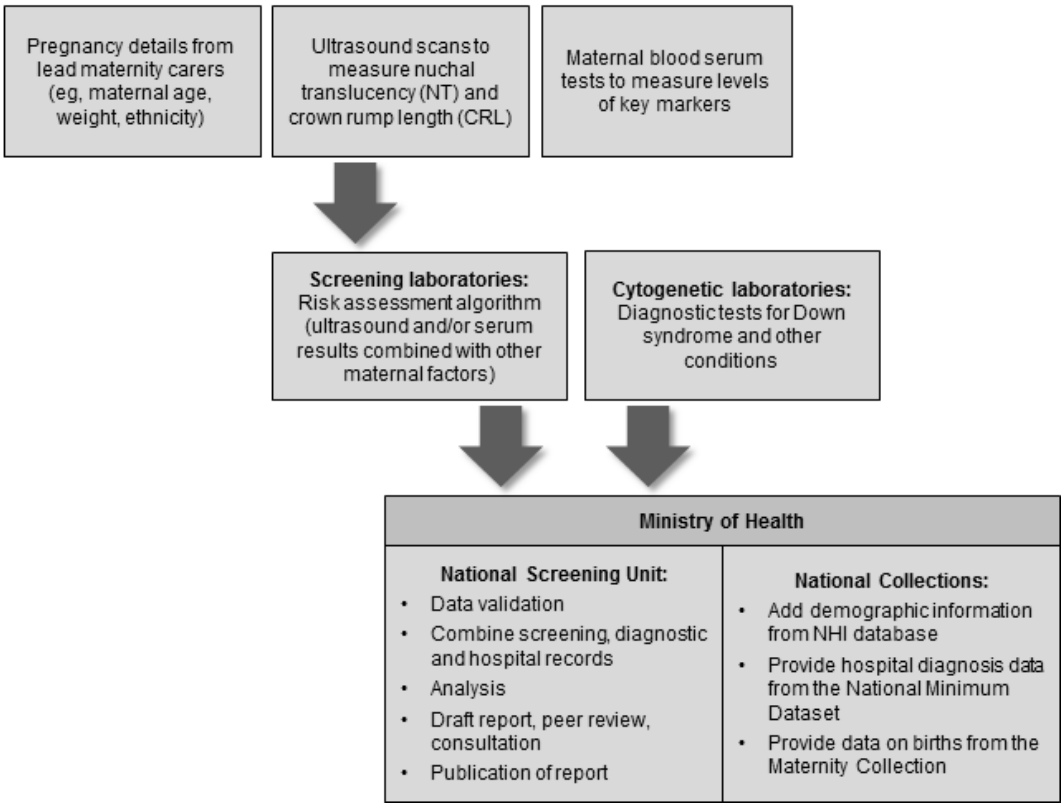
- trisomy 21 (Down Syndrome)
- trisomy 18 (Edwards syndrome)
- trisomy 13 (Patau syndrome)
- triploidy
- Turner syndrome
- neural tube defects.

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 2016. The definitions for the 11 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 are not available in 2016.

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
- four-yearly peer review of screening laboratories
- contract monitoring and reporting on a six-monthly 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. As in 2015, diagnostic testing data was received from all cytogenetic laboratories (LabPLUS, Waikato, Capital and Coast, and Canterbury Health Laboratories).

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

Definitions

Commenced screening

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

Completed screening

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

Required components of each screening test

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

Low risk result

A low risk result is defined as a risk lower than 1:300. So a risk of 1:310 is a low risk.

Increased risk result

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

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

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

Inclusion criteria

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

- screening commencement date between 1 January 2011 and 31 December 2016 (ie, date of the first test the woman had as part of the screening pathway)
- valid National Health Index (NHI) identifier
- 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.

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 women living in the least deprived 20% of small areas ('the least deprived areas'), and quintile 5 (deciles 9 and 10), which represents women living in the most deprived 20% of small areas ('the most deprived areas'). This is opposite to some other systems of classification, such as that used by education, where level 10 is the least disadvantaged and level 1 the most disadvantaged.

Births

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

Small numbers

Small numbers can affect the reliability of results. Where an indicator calculation involves small counts (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:

- women with no DHB of domicile information recorded in either the NHI database or in the laboratory information system were excluded from the analysis
- three babies identified with a positive diagnostic test could not be matched to a mother in the National Maternity Collection (MAT). This could be for a number of reasons including that the mothers of these babies may not have accessed publicly funded maternity care, their data may have been delayed, incorrectly entered or the babies may have been stillborn. While mothers with a delivery outcome of stillbirth are recorded in MAT (provided the gestation period was 20+ weeks or the birth weight was 400+ grams), details for the baby are usually not available. These babies may have been either a true positive, a false negative or have an unscreened mother. Due to this indicator 9 (Positive predictive value) and indicator 11 (Detection rate) should be interpreted with caution.

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.

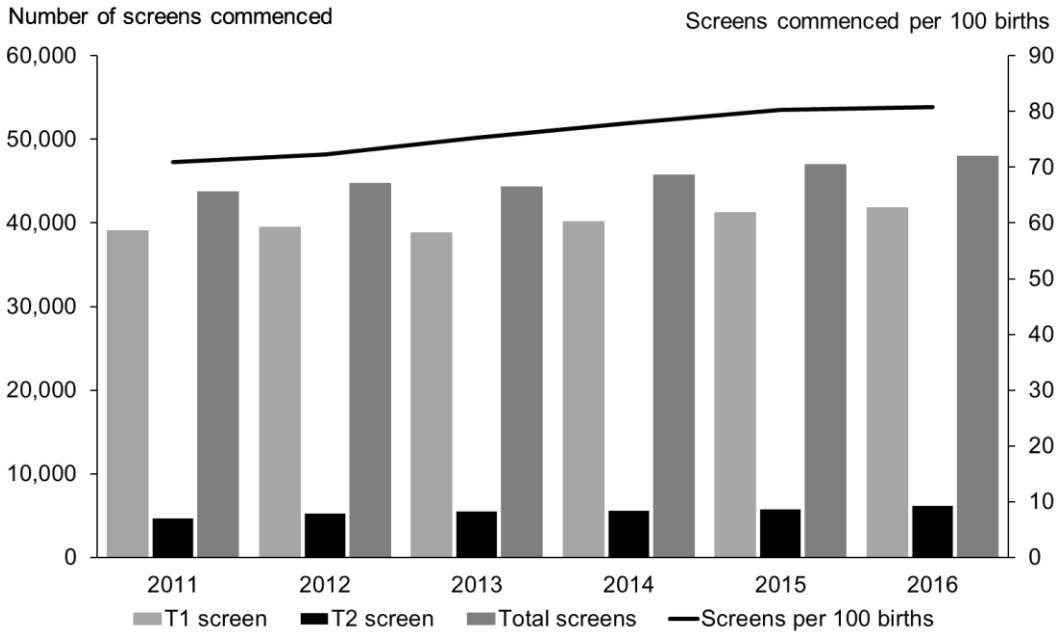
Total screens commenced by trimester

During 2016, a total of 47,968 screens were commenced, a rate of 81 per 100 births. Table 1 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 81 in 2016 (see Table 1 and Figure 2).

Table 1: Total screens commenced by trimester, January 2011 to December 2016

	Number and rate of screens commenced					
Trimester of screen	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
T1 screen	39,087	39,526	38,803	40,172	41,283	41,816
T2 screen	4,690	5,230	5,487	5,613	5,742	6,152
Total screens	**43,777**	**44,756**	**44,290**	**45,785**	**47,025**	**47,968**
Screens per 100 births	70.9	72.3	75.3	78.0	80.3	80.9

Figure 2: Count and rate of screens commenced, January 2011 to December 2016



Screens commenced by DHB

Figure 3 shows the screening commencement rates by DHB for 2016. There was a large variation in rates from 59 per 100 births in Northland to 92 per 100 births in Canterbury (see Figure 3). Over half of all DHBs had rates of above 80 per 100 births. Table 2 gives a full breakdown by the trimester of the screen.

Figure 3: Screens commenced by DHB, January 2016 to December 2016

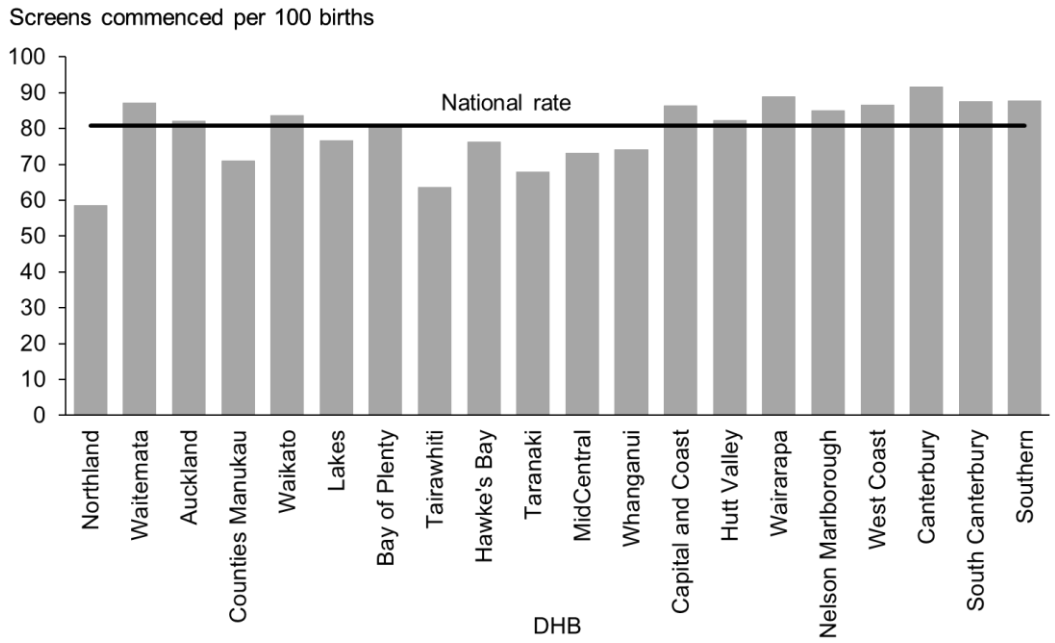


Table 2: Screens commenced by trimester and DHB, January 2016 to December 2016

DHB	Number of screens commenced			Screens commenced (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	1,113	214	1,327	49.1	9.4	58.6
Waitemata	6,152	759	6,911	77.5	9.6	87.1
Auckland	4,264	578	4,842	72.2	9.8	82.0
Counties Manukau	4,600	1,255	5,855	55.8	15.2	71.0
Waikato	4,023	464	4,487	75.1	8.7	83.7
Lakes	1,054	131	1,185	68.2	8.5	76.7
Bay of Plenty	2,151	200	2,351	74.2	6.9	81.1
Tairawhiti	428	65	493	55.2	8.4	63.6
Hawke's Bay	1,369	200	1,569	66.5	9.7	76.2
Taranaki	805	167	972	56.1	11.6	67.8
MidCentral	1,355	167	1,522	65.1	8.0	73.1
Whanganui	505	88	593	63.1	11.0	74.1
Capital and Coast	2,680	303	2,983	77.5	8.8	86.3
Hutt Valley	1,370	247	1,617	69.7	12.6	82.2
Wairarapa	351	60	411	76.0	13.0	89.0
Nelson Marlborough	1,184	133	1,317	76.5	8.6	85.1
West Coast	238	37	275	74.8	11.6	86.5
Canterbury	5,089	685	5,774	80.7	10.9	91.5
South Canterbury	470	99	569	72.3	15.2	87.5
Southern	2,615	300	2,915	78.8	9.0	87.8
Total	41,816	6,152	47,968	Av.70.5	10.4	80.9

Most DHBs showed an increase in their rate of screens commenced between 2011 and 2016, or had fairly stable rates (see Table 3).

Table 3: Screens commenced per 100 births by DHB, January 2011 to December 2016

DHB	Screens commenced (per 100 births)					
	2011	2012	2013	2014	2015	2016
Northland	46.5	49.7	52.9	55.6	60.1	58.6
Waitemata	84.0	82.9	86.3	86.3	88.4	87.1
Auckland	75.1	74.5	82.4	84.0	85.7	82.0
Counties Manukau	60.9	63.4	64.8	68.7	71.1	71.0
Waikato	73.1	72.1	76.4	80.4	81.8	83.7
Lakes	60.5	67.8	70.1	77.4	74.3	76.7
Bay of Plenty	65.3	68.5	69.6	72.4	77.6	81.1
Tairawhiti	44.5	49.2	53.2	59.3	68.3	63.6
Hawke's Bay	55.8	61.9	64.6	66.0	72.6	76.2
Taranaki	62.6	60.2	61.4	68.2	74.9	67.8
MidCentral	51.1	54.4	58.3	59.3	63.9	73.1
Whanganui	45.0	44.9	47.9	61.0	70.5	74.1
Capital and Coast	76.4	79.4	78.1	80.3	83.4	86.3
Hutt Valley	71.0	70.7	72.7	78.6	78.7	82.2
Wairarapa	72.8	69.1	76.6	81.6	83.8	89.0
Nelson Marlborough	87.8	90.8	87.4	97.6	96.0	85.1
West Coast	68.9	76.3	81.1	88.3	82.4	86.5
Canterbury	85.4	86.8	90.3	89.5	89.4	91.5
South Canterbury	92.3	85.5	88.1	78.8	86.4	87.5
Southern	75.3	80.0	81.4	83.3	85.1	87.8
National average	**70.9**	**72.3**	**75.3**	**78.0**	**80.3**	**80.9**

Screens commenced by age, ethnicity and deprivation

Table 4 provides an overall view of screens commenced by age, ethnicity and NZ deprivation quintile for January 2011 to December 2016. During this reporting period the overall rate of screens commenced has increased and though variation between age, ethnicity and deprivation is still evident these differences have become less marked.

Table 4: Screens commenced by age of mother, ethnicity and NZ deprivation quintile, January 2011 to December 2016

	Number of screens commenced						Screens commenced (per 100 births)					
	2011	**2012**	**2013**	**2014**	**2015**	**2016**	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
Age at screen												
Under 20 years	2,282	2,128	1,947	1,990	1,925	1,829	56.4	54.5	58.5	66.6	69.1	74.9
20–24 years	6,817	6,966	6,932	7,055	7,109	7,000	58.3	60.8	64.2	68.7	71.5	73.0
25–29 years	11,509	12,078	12,022	12,800	13,189	13,943	74.1	75.8	78.8	81.5	84.0	84.3
30–34 years	13,433	13,751	13,914	14,623	15,124	15,732	78.0	78.8	83.0	83.2	84.5	85.6
35–39 years	8,027	8,040	7,628	7,610	8,007	7,781	74.9	77.2	76.0	78.6	82.0	78.1
40–44 years	1,636	1,716	1,767	1,626	1,593	1,574	68.1	66.5	72.6	69.3	69.3	69.2
45 years and over	73	77	80	81	78	109	57.9	64.2	55.9	61.4	56.1	86.5
Ethnicity												
Māori	5,540	5,881	5,805	6,284	6,256	7,176	34.9	37.3	39.6	43.9	42.9	48.7
Pacific	3,055	3,102	2,999	3,005	3,120	3,089	43.2	45.1	47.2	48.7	51.5	52.9
Asian	6,484	7,405	7,474	8,438	8,695	9,851	91.0	87.7	91.7	91.8	94.4	93.6
Other	28,698	28,368	28,012	28,058	28,954	27,852	90.6	92.2	94.5	96.6	100.9	98.7
NZ deprivation quintile												
Quintile 1	8,130	8,073	7,654	7,732	7,898	8,509	95.6	93.1	93.6	91.3	95.8	98.2
Quintile 2	8,174	8,395	8,231	8,413	8,652	8,780	86.0	87.3	89.0	91.7	92.7	90.7
Quintile 3	8,529	8,685	8,730	8,878	9,130	9,278	76.5	77.8	82.2	84.1	86.3	86.6
Quintile 4	9,526	9,822	9,882	10,353	10,475	10,584	69.1	71.9	73.7	78.0	79.1	79.6
Quintile 5	9,409	9,777	9,789	10,408	10,864	10,805	50.0	52.1	56.6	60.5	63.8	63.7
Unknown	9	4	4	1	6	12						
National	**43,777**	**44,756**	**44,290**	**45,785**	**47,025**	**47,968**	**70.9**	**72.3**	**75.3**	**78.0**	**80.3**	**80.9**

Rate suppressed if the number of screens was <10.

Figure 4: Screens commenced by age of mother at screen, January 2016 to December 2016

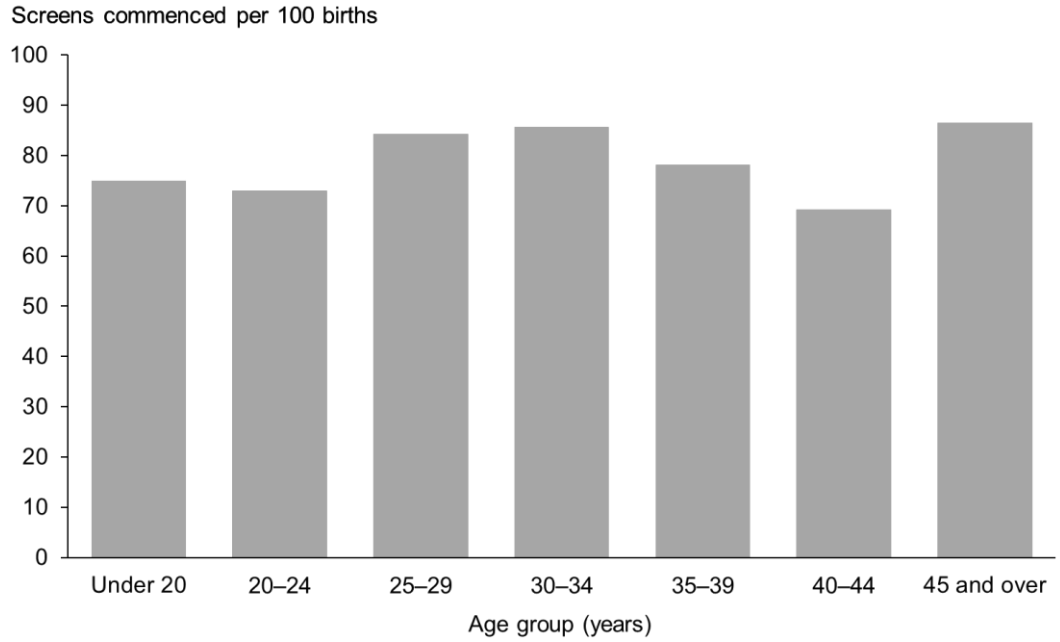
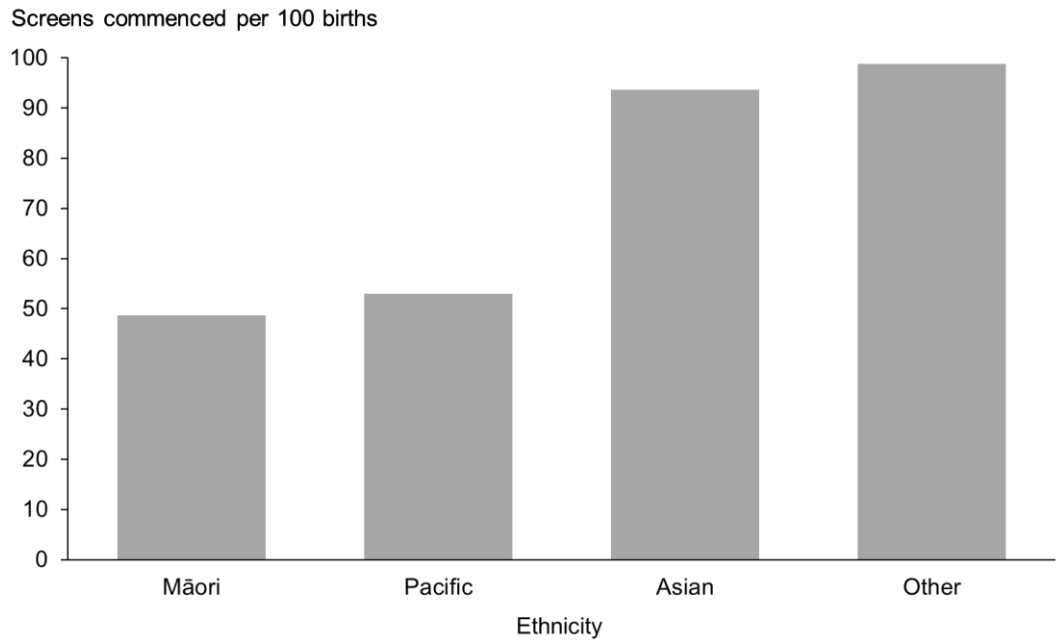
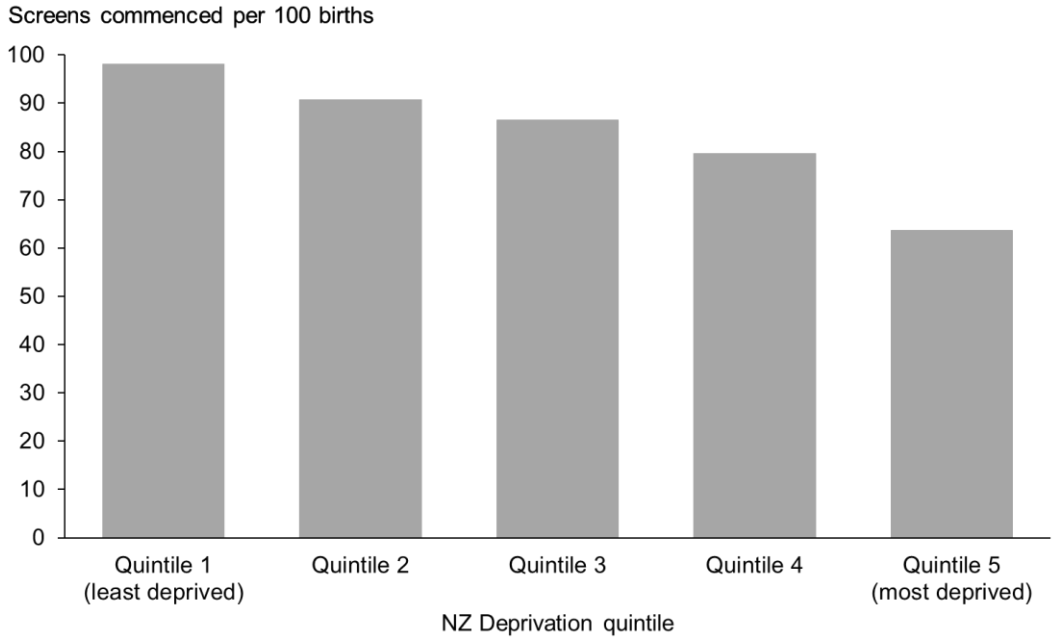


Figure 5: Screens commenced by ethnicity of mother, January 2016 to December 2016



Differences in screening commencement rates by ethnicity remained consistent for 2016. Women of Other ethnicity had the highest rate (99 of 100 births) followed by Asian women (94 of 100 births). The rate of commenced screens for Pacific and Māori women was lower at 53 per 100 births and 49 per 100 births respectively (see Figure 5). All groups have shown increasing rates over the five years, particularly for Māori with an absolute increase of almost 14 percentage points from 35% in 2011 to 49% in 2016 (see Table 4). This rate is however well below the national average.

Figure 6: Screens commenced by NZ deprivation quintile, January 2016 to December 2016



A trend of higher screening commencement rates for women in less deprived areas was evident, with 98 women per 100 per births starting screening for quintile 1 women in 2016 compared with 64 per 100 births for quintile 5 (see Figure 6). All quintiles showed a rate increase between 2011 and 2016, particularly women in more deprived regions (see Table 4).

Indicator 2: Screens completed

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

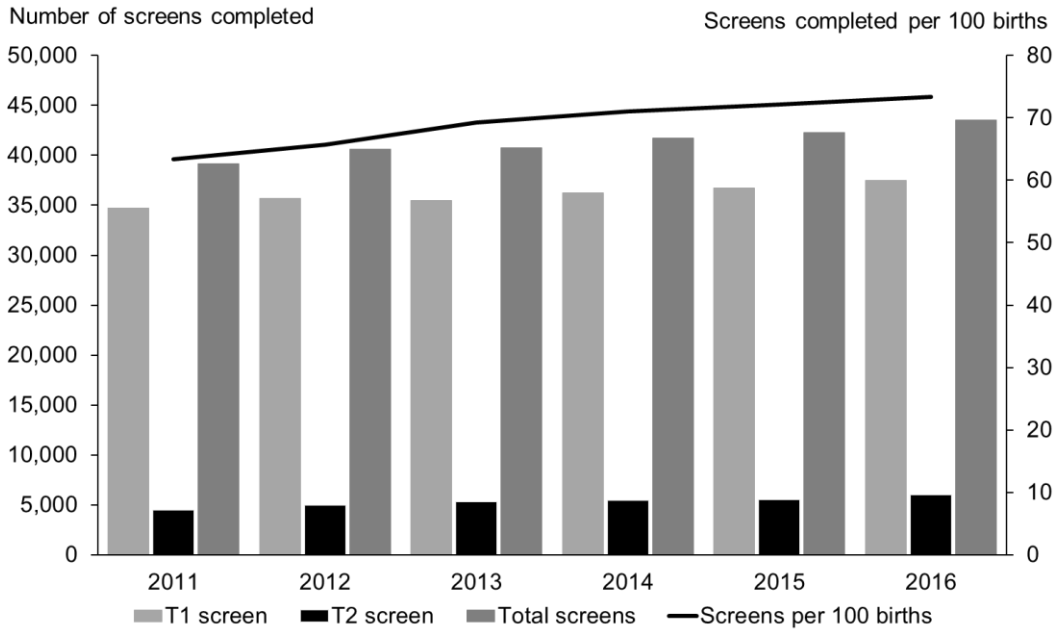
Total screens completed by trimester

During 2016, a total of 43,519 screens were completed, a rate of 73 per 100 births. Table 5 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 (from 63% to 73% in 2016).

Table 5: Total screens completed by trimester, January 2011 to December 2016

	Number and rate of screens completed					
Trimester of screen	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
T1 screen	34,735	35,691	35,464	36,280	36,739	37,511
T2 screen	4,446	4,957	5,269	5,456	5,517	6,008
Total screens	**39,181**	**40,648**	**40,733**	**41,736**	**42,256**	**43,519**
Screens per 100 births	63.4	65.7	69.3	71.1	72.2	73.4

Figure 7: Count and rate of screens completed, January 2011 to December 2016



Screens completed by DHB

Screening completion rates for 2016 varied across DHBs from 51 per 100 births in Northland to 82 per 100 births in Canterbury (see Figure 8). Table 7 gives a full breakdown by the trimester of the screen.

Figure 8: Screens completed by DHB, January 2016 to December 2016

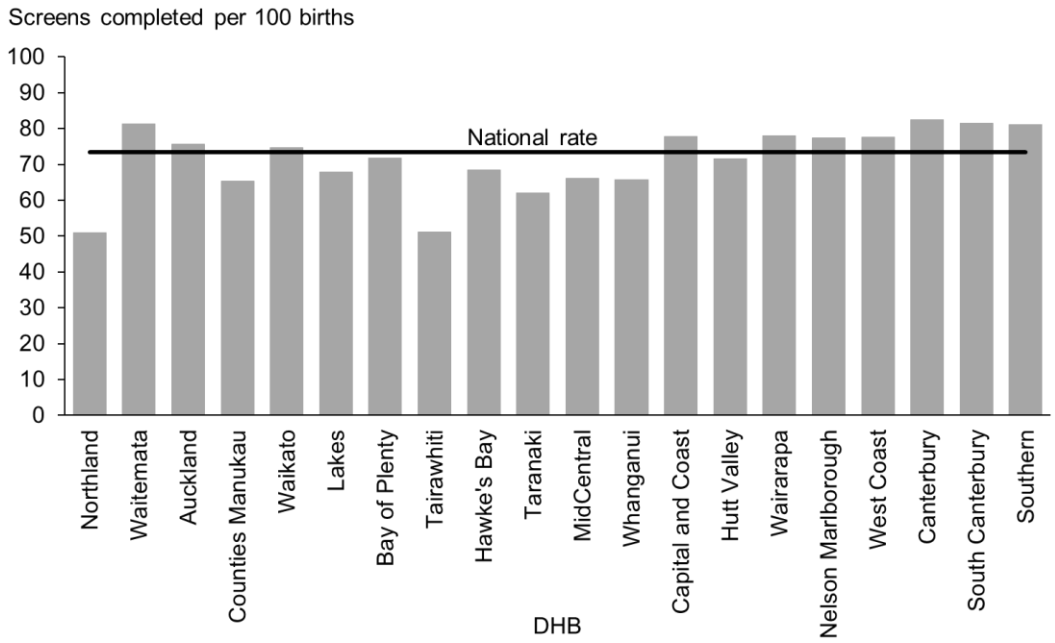


Table 6: Screening completion by trimester and DHB, January 2016 to December 2016

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	941	212	1,153	41.5	9.4	50.9
Waitemata	5,715	741	6,456	72.0	9.3	81.4
Auckland	3,899	565	4,464	66.0	9.6	75.6
Counties Manukau	4,173	1,221	5,394	50.6	14.8	65.4
Waikato	3,556	445	4,001	66.4	8.3	74.7
Lakes	922	126	1,048	59.7	8.2	67.8
Bay of Plenty	1,887	192	2,079	65.1	6.6	71.7
Tairawhiti	333	63	396	43.0	8.1	51.1
Hawke's Bay	1,216	196	1,412	59.0	9.5	68.5
Taranaki	725	165	890	50.6	11.5	62.1
MidCentral	1,212	165	1,377	58.2	7.9	66.1
Whanganui	441	85	526	55.1	10.6	65.8
Capital and Coast	2,393	296	2,689	69.2	8.6	77.8
Hutt Valley	1,167	240	1,407	59.4	12.2	71.6
Wairarapa	301	59	360	65.2	12.8	77.9
Nelson Marlborough	1,065	133	1,198	68.8	8.6	77.4
West Coast	210	37	247	66.0	11.6	77.7
Canterbury	4,525	674	5,199	71.7	10.7	82.4
South Canterbury	433	97	530	66.6	14.9	81.5
Southern	2,397	296	2,693	72.2	8.9	81.1
Total	**37,511**	**6,008**	**43,519**	**Av. 63.2**	**10.1**	**73.4**

Similar to screens commenced, most DHBs showed a trend of increasing rates of screening completion over the six years covered in this report.

Table 7: Screening completion by DHB, January 2011 to December 2016

	Screens completed (per 100 births)					
DHB	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
Northland	41.1	44.4	47.1	48.0	51.6	50.9
Waitemata	78.0	77.9	82.1	81.0	81.7	81.4
Auckland	70.5	69.5	77.7	78.8	79.1	75.6
Counties Manukau	53.9	57.3	59.6	63.2	64.4	65.4
Waikato	65.3	64.2	69.2	72.5	72.4	74.7
Lakes	53.1	59.1	62.6	69.9	65.7	67.8
Bay of Plenty	58.3	61.7	62.0	64.5	67.8	71.7
Tairawhiti	39.6	44.4	47.1	51.5	53.8	51.1
Hawke's Bay	50.2	55.9	59.9	59.4	64.2	68.5
Taranaki	58.2	55.6	55.1	61.2	66.3	62.1
MidCentral	45.3	49.5	53.8	54.0	56.9	66.1
Whanganui	40.2	41.8	45.0	53.1	58.5	65.8
Capital and Coast	67.9	71.9	70.9	72.6	75.1	77.8
Hutt Valley	59.1	62.5	64.7	68.9	68.0	71.6
Wairarapa	62.8	59.5	66.7	70.6	72.8	77.9
Nelson Marlborough	78.6	81.3	78.1	87.6	84.7	77.4
West Coast	55.6	68.5	72.3	78.9	72.3	77.7
Canterbury	72.4	75.8	81.9	81.2	80.6	82.4
South Canterbury	87.2	82.6	85.6	75.3	79.8	81.5
Southern	67.3	73.7	75.5	74.8	77.9	81.1
National average	**63.4**	**65.7**	**69.3**	**71.1**	**72.2**	**73.4**

Screens completed by age, ethnicity and deprivation

Table 8 provides an overall view of screens completed by age, ethnicity and NZ deprivation quintile for January 2011 to December 2016, with similar trends to screening commencement. In the six years of reporting, screening completion rates were highest in the 30–34 year age group with 80 women completing screening per 100 births in 2016.

Screening completion rates were highest among women of Other ethnicity at 91 per 100 births for 2016. 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 46 per 100 births and 40 per 100 births respectively (see Figure 10). Completion rates improved in all ethnic groups with the greatest improvement seen in Māori and Pacific women (see Table 8).

Screening completion rates were highest among women in less deprived areas with a rate of 91 per 100 per births for quintile 1 in 2016 compared with 56 per 100 births for quintile 5 (see Figure 11).

Table 8: Screens completed by age of mother, ethnicity and NZ deprivation quintile, January 2011 to December 2016

	Number of screens completed						Screens completed (per 100 births)					
	2011	**2012**	**2013**	**2014**	**2015**	**2016**	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
Age at screen												
Under 20 years	1,808	1,699	1,610	1,604	1,510	1,474	44.7	43.5	48.4	53.6	54.2	60.3
20–24 years	5,754	5,890	6,010	6,070	5,992	6,079	49.2	51.4	55.6	59.1	60.3	63.4
25–29 years	10,276	10,997	11,097	11,685	11,824	12,675	66.1	69.0	72.7	74.4	75.3	76.6
30–34 years	12,353	12,859	13,089	13,675	14,030	14,709	71.7	73.7	78.0	77.8	78.3	80.1
35–39 years	7,453	7,543	7,214	7,144	7,430	7,137	69.6	72.5	71.9	73.8	76.1	71.6
40–44 years	1,474	1,588	1,643	1,486	1,406	1,366	61.3	61.6	67.5	63.3	61.2	60.0
45 years and over	63	72	70	72	64	79	50.0	60.0	49.0	54.5	46.0	62.7
Ethnicity												
Māori	4,561	4,880	4,893	5,178	4,911	5,924	28.7	30.9	33.4	36.2	33.7	40.2
Pacific	2,479	2,591	2,606	2,598	2,626	2,673	35.1	37.7	41.0	42.1	43.3	45.8
Asian	6,024	6,990	7,091	8,034	8,114	9,304	84.5	82.7	87.0	87.4	88.1	88.4
Other	26,117	26,187	26,143	25,926	26,605	25,618	82.4	85.1	88.2	89.2	92.7	90.8
NZ deprivation quintile												
Quintile 1	7,519	7,520	7,255	7,242	7,335	7,847	88.5	86.7	88.7	85.5	89.0	90.5
Quintile 2	7,480	7,805	7,749	7,867	8,028	8,126	78.7	81.2	83.8	85.8	86.0	84.0
Quintile 3	7,748	8,028	8,102	8,195	8,323	8,541	69.5	71.9	76.3	77.6	78.6	79.7
Quintile 4	8,401	8,851	9,001	9,325	9,307	9,554	60.9	64.8	67.1	70.3	70.3	71.9
Quintile 5	8,027	8,441	8,622	9,106	9,257	9,440	42.7	45.0	49.8	52.9	54.3	55.6
Unknown	6	3	4	1	6	11						
National	**39,181**	**40,648**	**40,733**	**41,736**	**42,256**	**43,519**	**63.4**	**65.7**	**69.3**	**71.1**	**72.2**	**73.4**

Figure 9: Screens completed by age of mother at screen, January 2016 to December 2016

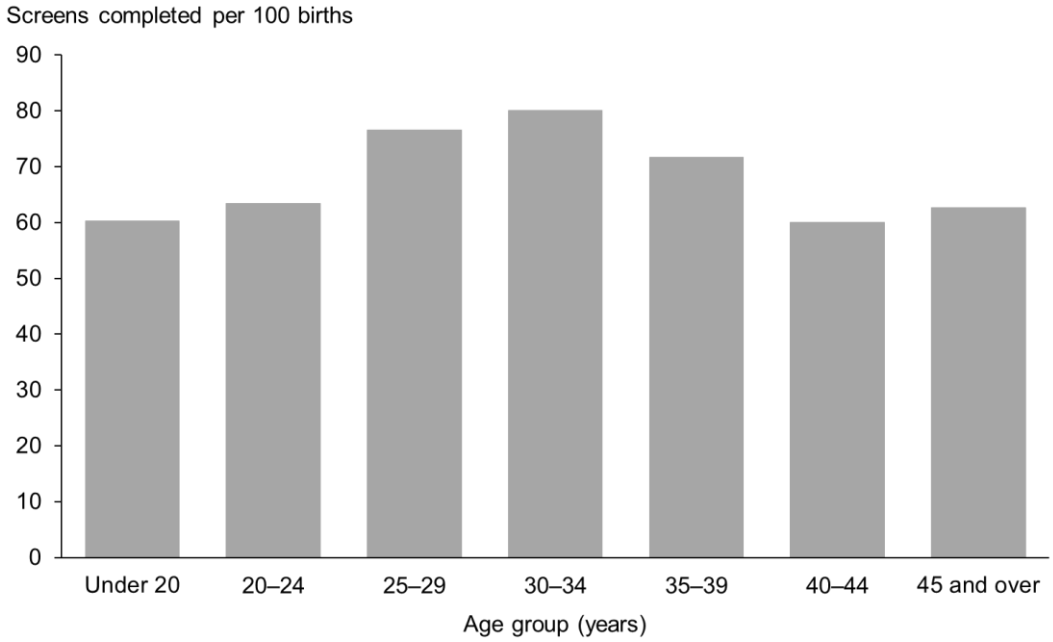


Figure 10: Screens completed by ethnicity of mother, January 2016 to December 2016

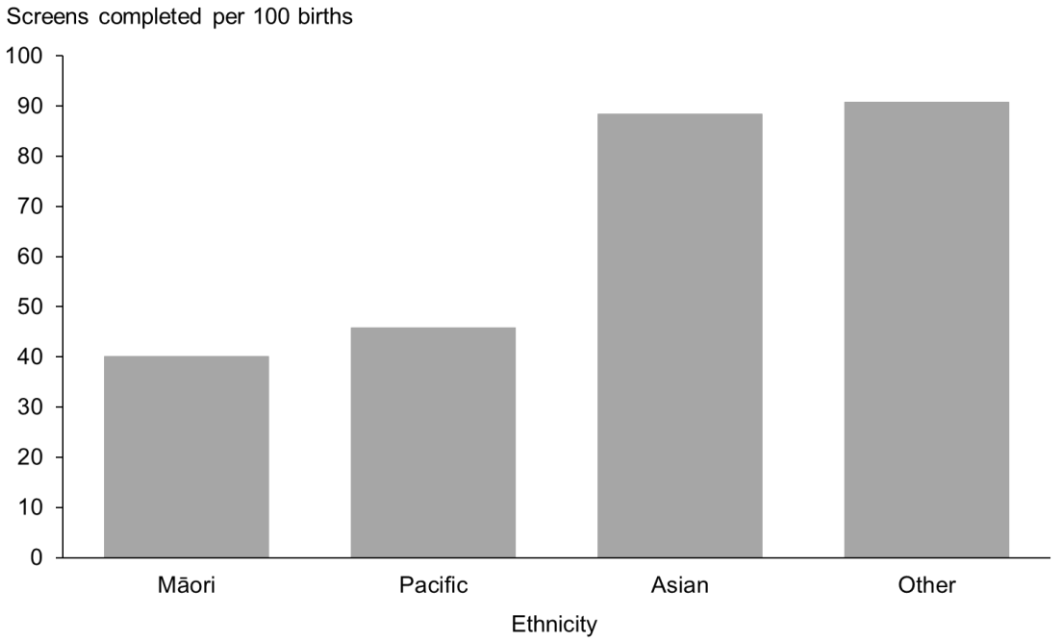
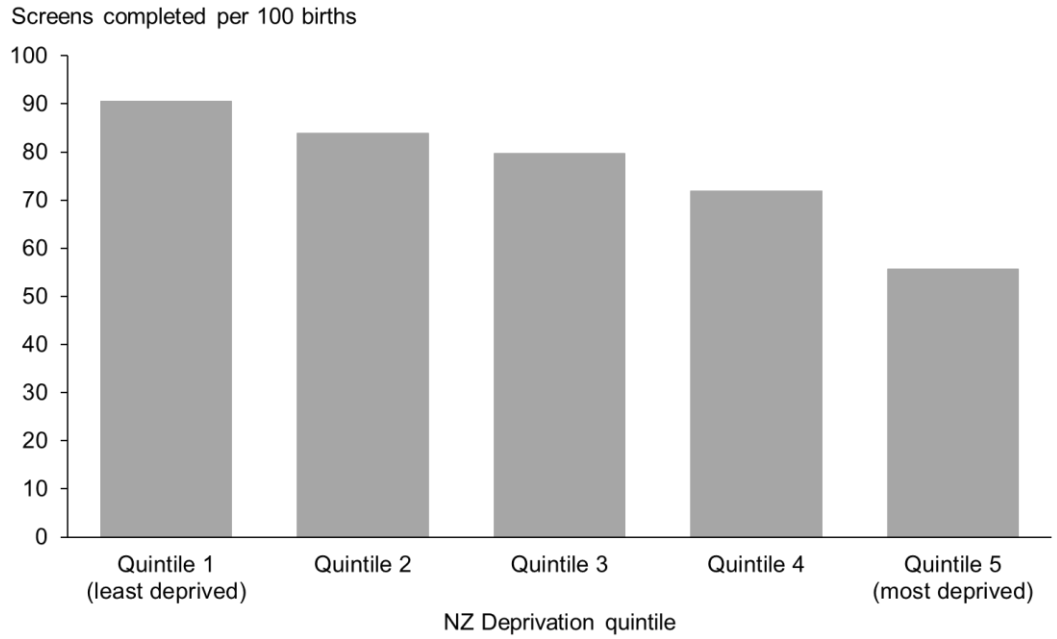


Figure 11: Screens completed by NZ deprivation quintile of mother, January 2016 to December 2016



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 9 shows the number and proportion of second trimester screening results that included first trimester inputs over the period from 2011 to 2016. This has been broken down by the type of pathway variance.

The largest pathway variance was due to second trimester screens with an NT measurement (44% in 2016). PAPP-A was included in 8% of second trimester screens in 2016, higher than previous years.

Table 9: Screening pathway variance by type, January 2011 to December 2016

		Second trimester screening results			
		Number		Percentage	
Year	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
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	5,517	2,466	344	44.7	6.2
2016	6,008	2,670	500	44.4	8.3

Screening pathway variance by DHB

Table 10 shows a breakdown of screening pathway variance by DHB and type of variance for the 2016 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 clinical gestation) leading to women being assessed in a different trimester.

Table 10: Screening pathway variance by DHB, January 2016 to December 2016

		Second trimester screening results			
		Number		Percentage	
DHB	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
Northland	212	74	19	34.9	9.0
Waitemata	741	343	61	46.3	8.2
Auckland	565	216	53	38.2	9.4
Counties Manukau	1,221	377	113	30.9	9.3
Waikato	445	223	31	50.1	7.0
Lakes	126	55	10	43.7	7.9
Bay of Plenty	192	105	9	54.7	4.7
Tairawhiti	63	25	12	39.7	19.0
Hawke's Bay	196	74	9	37.8	4.6
Taranaki	165	55	19	33.3	11.5
MidCentral	165	77	11	46.7	6.7
Whanganui	85	44	4	51.8	4.7
Capital and Coast	296	166	22	56.1	7.4
Hutt Valley	240	116	21	48.3	8.8
Wairarapa	59	33	5	55.9	8.5
Nelson Marlborough	133	75	5	56.4	3.8
West Coast	37	18	6	48.6	16.2
Canterbury	674	357	69	53.0	10.2
South Canterbury	97	59	13	60.8	13.4
Southern	296	178	8	60.1	2.7
Total	**6,008**	**2,670**	**500**	**Av. 44.4**	**8.3**

Screening pathway variance by age, ethnicity and deprivation

Table 11 shows a breakdown of screening pathway variance by age, ethnicity and NZ deprivation quintile for the 2016 year. The results show higher proportions for pathway variance for women of Other ethnicity, and women in areas of lower deprivation.

Table 11: Screening pathway variance by age, ethnicity and NZ deprivation quintile, January 2016 to December 2016

	Second trimester screening results				
		Number		Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
Age at screen					
Under 20 years	423	149	19	35.2	4.5
20–24 years	1,270	560	80	44.1	6.3
25–29 years	1,808	833	152	46.1	8.4
30–34 years	1,551	718	145	46.3	9.3
35–39 years	766	332	89	43.3	11.6
40–44 years	178	71	14	39.9	7.9
45 years and over	12	7	1	58.3	8.3
Ethnicity					
Māori	1,552	632	87	40.7	5.6
Pacific	982	276	76	28.1	7.7
Asian	1,224	500	116	40.8	9.5
Other	2,250	1,262	221	56.1	9.8
NZ deprivation quintile					
Quintile 1	657	367	51	55.9	7.8
Quintile 2	822	428	82	52.1	10.0
Quintile 3	915	470	79	51.4	8.6
Quintile 4	1,420	633	123	44.6	8.7
Quintile 5	2,189	770	165	35.2	7.5
Unknown	5	2	0	40.0	0.0
Total	**6,008**	**2,670**	**500**	**Av 44.4**	**8.3**

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 12 shows the 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 2016 was 4,449, which equates to 9% of screens commenced that year and demonstrates an improvement from 2015.

Table 12: Incomplete screens by trimester, January 2011 to December 2016

Trimester of screen	Number of incomplete screens					
	2011	2012	2013	2014	2015	2016
T1 screen	4,352	3,835	3,339	3,892	4,544	4,305
T2 screen	244	273	218	157	225	144
Total screens	**4,596**	**4,108**	**3,557**	**4,049**	**4,769**	**4,449**

Incomplete T1 screens by reason incomplete

Table 13 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 2016 was 10%. This was the result of both screens without blood samples and screens without NT scans. The majority of incomplete screens in T1 were due to a missing blood sample.

Table 13: Incomplete T1 screens by reason incomplete, January 2011 to December 2016

	Commenced first trimester			Reason incomplete			Incomplete as percentage of commenced			Type as percentage of all incomplete trimester 1 screens	
Year	No result issued	Result issued	Total	No blood	No NT scan	No weight	T1 no blood	T1 no NT scan	Total T1 incompletes	T1 no blood	T1 no NT scan
2011	4,352	34,735	39,087	3,294	1,058	–	8.4	2.7	11.1	75.7	24.3
2012	3,835	35,691	39,526	2,844	991	–	7.2	2.5	9.7	74.2	25.8
2013	3,339	35,464	38,803	2,318	1,021	–	6.0	2.6	8.6	69.4	30.6
2014	3,892	36,280	40,172	2,630	1,262	–	6.5	3.1	9.7	67.6	32.4
2015	4,544	36,739	41,283	2,925	1,619	–	7.1	3.9	11.0	64.4	35.6
2016	4,305	37,511	41,816	2,946	1,335	24	7.0	3.2	10.3	68.4	31.0

Incomplete T1 screens by reason and DHB

Table 14 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 45% (at Taranaki) to 80% (at Whanganui).

Table 14: Incomplete T1 screens by reason and DHB, January 2016 to December 2016

	Commenced first trimester			Reason incomplete			Incomplete as percentage of commenced			Type as percentage of all incomplete	
DHB	No result issued	Result issued	Total	No blood	No NT scan	No weight	T1 no blood	T1 no NT scan	Total T1	T1 no blood	T1 no NT scan
Northland	172	941	1,113	118	54	0	10.6	4.9	15.5	68.6	31.4
Waitemata	437	5,715	6,152	275	162	0	4.5	2.6	7.1	62.9	37.1
Auckland	365	3,899	4,264	254	111	0	6.0	2.6	8.6	69.6	30.4
Counties Manukau	427	4,173	4,600	252	175	0	5.5	3.8	9.3	59.0	41.0
Waikato	467	3,556	4,023	346	121	0	8.6	3.0	11.6	74.1	25.9
Lakes	132	922	1,054	100	32	0	9.5	3.0	12.5	75.8	24.2
Bay of Plenty	264	1,887	2,151	188	76	0	8.7	3.5	12.3	71.2	28.8
Tairawhiti	95	333	428	59	34	2	13.8	7.9	22.2	62.1	35.8
Hawke's Bay	153	1,216	1,369	112	39	2	8.2	2.8	11.2	73.2	25.5
Taranaki	80	725	805	36	44	0	4.5	5.5	9.9	45.0	55.0
MidCentral	143	1,212	1,355	107	35	1	7.9	2.6	10.6	74.8	24.5
Whanganui	64	441	505	51	10	3	10.1	2.0	12.7	79.7	15.6
Capital and Coast	287	2,393	2,680	203	83	1	7.6	3.1	10.7	70.7	28.9
Hutt Valley	203	1,167	1,370	159	44	0	11.6	3.2	14.8	78.3	21.7
Wairarapa	50	301	351	37	12	1	10.5	3.4	14.2	74.0	24.0
Nelson Marlborough	119	1,065	1,184	85	34	0	7.2	2.9	10.1	71.4	28.6
West Coast	28	210	238	21	7	0	8.8	2.9	11.8	75.0	25.0
Canterbury	564	4,525	5,089	382	175	7	7.5	3.4	11.1	67.7	31.0
South Canterbury	37	433	470	28	9	0	6.0	1.9	7.9	75.7	24.3
Southern	218	2,397	2,615	133	78	7	5.1	3.0	8.3	61.0	35.8
National	**4,305**	**37,511**	**41,816**	**2,946**	**1,335**	**24**	**7.0**	**3.2**	**10.3**	**68.4**	**31.0**

Incomplete T1 screens by age, ethnicity and deprivation

Table 15 shows a breakdown of incomplete screens with reason incomplete, by age, ethnicity, and NZ deprivation quintile for the 2016 year. There were higher rates of incomplete screens for Māori (22%) and Pacific (18%) women when compared with Asian (6%) and Other (9%). The rate of incomplete screens also increased with increasing deprivation (15% for quintile 5 compared with 8% for quintile 1).

Table 15: Incomplete T1 screens by age, ethnicity and NZ deprivation quintile, January 2016 to December 2016

	Commenced first trimester			Reason incomplete			Incomplete as percentage of commenced			Type as percentage of all incomplete	
	No result issued	Result issued	Total commenced	No blood	No NT scan	No weight	T1 no blood	T1 no NT scan	All T1 incomplete	T1 no blood	T1 no NT scan
Age at screen											
Under 20 years	344	1,051	1,395	257	85	2	18.4	6.1	24.7	74.7	24.7
20–24 years	886	4,809	5,695	653	229	4	11.5	4.0	15.6	73.7	25.8
25–29 years	1,230	10,867	12,097	869	349	12	7.2	2.9	10.2	70.7	28.4
30–34 years	989	13,158	14,147	630	356	3	4.5	2.5	7.0	63.7	36.0
35–39 years	626	6,371	6,997	387	237	2	5.5	3.4	8.9	61.8	37.9
40–44 years	201	1,188	1,389	128	73	–	9.2	5.3	14.5	63.7	36.3
45 years and over	29	67	96	22	6	1	22.9	6.3	30.2	75.9	20.7
Ethnicity											
Māori	1,199	4,372	5,571	907	287	5	16.3	5.2	21.5	75.6	23.9
Pacific	376	1,691	2,067	232	143	1	11.2	6.9	18.2	61.7	38.0
Asian	531	8,080	8,611	298	230	3	3.5	2.7	6.2	56.1	43.3
Other	2,199	23,368	25,567	1,509	675	15	5.9	2.6	8.6	68.6	30.7
NZ deprivation quintile											
Quintile 1	648	7,190	7,838	459	186	3	5.9	2.4	8.27	70.8	28.7
Quintile 2	643	7,304	7,947	438	200	5	5.5	2.5	8.1	68.1	31.1
Quintile 3	720	7,626	8,346	486	231	3	5.8	2.8	8.6	67.5	32.1
Quintile 4	992	8,134	9,126	677	309	6	7.4	3.4	10.9	68.2	31.1
Quintile 5	1,301	7,251	8,552	885	409	7	10.3	4.8	15.2	68.0	31.4
Unknown	1	6		1	–	–					
National	**4,305**	**37,511**	**41,816**	**2,946**	**1,335**	**24**	**7.0**	**3.2**	**10.3**	**68.4**	**31.0**

Incomplete T2 screens

T2 screens do not require an NT scan, just a blood sample, but may be incomplete if missing dating information or no weight, if the sample is taken later than 20 weeks of pregnancy, or if the sample is damaged and not repeated. For 2016, 2% of T2 commenced screens were incomplete, compared with 10% of T1 commenced screens. As Table 16 shows, the percentage of incomplete T2 screens decreased from 5% in 2011 to 2% in 2016.

Table 16: Incomplete T2 screens, January 2011 to December 2016

Year	Commenced second trimester	No result issued	Percentage incomplete
2011	4,690	244	5.2
2012	5,230	273	5.2
2013	5,487	218	4.0
2014	5,613	157	2.8
2015	5,742	225	3.9
2016	6,152	144	2.3
Total	**32,914**	**1,261**	**Av of last six years: 3.8**

Incomplete T2 screens by DHB

Table 17 shows a breakdown of incomplete T2 screens by DHB for the 2016 year. The very low numbers involved limit meaningful DHB comparisons.

Table 17: Incomplete T2 screens by DHB, January 2016 to December 2016

DHB	Commenced second trimester	No result issued	Percentage incomplete
Northland	214	2	0.9
Waitemata	759	18	2.4
Auckland	578	13	2.2
Counties Manukau	1,255	34	2.7
Waikato	464	19	4.1
Lakes	131	5	3.8
Bay of Plenty	200	8	4.0
Tairawhiti	65	2	3.1
Hawke's Bay	200	4	2.0
Taranaki	167	2	1.2
MidCentral	167	2	1.2
Whanganui	88	3	3.4
Capital and Coast	303	7	2.3
Hutt Valley	247	7	2.8
Wairarapa	60	1	1.7
Nelson Marlborough	133	0	0.0
West Coast	37	0	0.0
Canterbury	685	11	1.6
South Canterbury	99	2	2.0
Southern	300	4	1.3
Total	**6,152**	**144**	**Av.2.3**

Incomplete T2 screens by age, ethnicity and deprivation

Table 18 shows a breakdown of incomplete T2 screens by age, ethnicity and NZ deprivation quintile for 2016. The percentage incomplete was higher for Māori and Pacific women with no clear trends by age and deprivation.

Table 18: Incomplete T2 screens by age, ethnicity and NZ deprivation quintile, January 2016 to December 2016

	Commenced second trimester	No result issued	Percentage incomplete
Age at screen			
Under 20 years	434	11	2.5
20–24 years	1,305	35	2.7
25–29 years	1,846	38	2.1
30–34 years	1,585	34	2.1
35–39 years	784	18	2.3
40–44 years	185	7	3.8
45 years and over	13	1	7.7
Ethnicity			
Māori	1,605	53	3.3
Pacific	1,022	40	3.9
Asian	1,240	16	1.3
Other	2,285	35	1.5
NZ deprivation quintile			
Quintile 1	671	14	2.1
Quintile 2	833	11	1.3
Quintile 3	932	17	1.8
Quintile 4	1,458	38	2.6
Quintile 5	2,253	64	2.8
Unknown	5		
National	**6,152**	**144**	**2.3**

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 19 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 2016 year, 2.7 increased risk results were issued for every 100 screens completed. This was consistent with the rates for previous years.

Table 19: Number and rate per 100 screens of increased risk screening results for trisomy 21, 18 or 13, January 2011 to December 2016

	Number and rate of increased risk screens					
	2011	2012	2013	2014	2015	2016
Total increased risk results	1,104	1,160	1,111	1,162	1,168	1,189
Positive test rate per 100 completed screens	2.8	2.9	2.7	2.8	2.8	2.7

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

Table 20 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 2016 year.

Positive test rate was 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 because of 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 20: Increased risk screening results for trisomy 21, 18 or 13 by age, ethnicity and deprivation, January 2016 to December 2016

	Number of screens that include an increased risk for trisomy 21, 18 or 13	Total number of completed screens	Positive test rate per 100 screens
Age at screen			
Under 20 years	22	1,474	1.5
20–24 years	63	6,079	1.0
25–29 years	164	12,675	1.3
30–34 years	323	14,709	2.2
35–39 years	363	7,137	5.1
40–44 years	236	1,366	17.3
45 years and over	18	79	22.8
Ethnicity			
Māori	135	5,924	2.3
Pacific	102	2,673	3.8
Asian	311	9,304	3.3
Other	641	25,618	2.5
NZ deprivation quintile			
Quintile 1	218	7,847	2.8
Quintile 2	230	8,126	2.8
Quintile 3	244	8,541	2.9
Quintile 4	231	9,554	2.4
Quintile 5	266	9,440	2.8
Unknown		11	0.0

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

Table 21 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.

In 2016, trisomy 18 and 13 each had low positivity rates (0.4 per 100 screens) 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 slightly higher than the first trimester positive test rate (3.3 and 2.5 respectively). This difference was more marked in previous years (4.8 and 2.5 in 2011). The 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 in 2016. This reflects the far higher number of trisomy 21 increased risks compared with trisomy 18 and 13.

Table 21: Increased risk screening results for trisomy 21, 18 and 13 by trimester of screen, January 2011 to December 2016

Year	Total results that include an increased risk for specified trisomy	Positive test rate per 100 screens	T1 results that include an increased risk for specified trisomy	Positive test rate per 100 T1 screens	T2 results that include an increased risk for specified trisomy	Positive test rate per 100 T2 screens
Trisomy 21						
2011	1,086	2.8	873	2.5	213	4.8
2012	1,148	2.8	874	2.4	274	5.5
2013	1,089	2.7	848	2.4	241	4.6
2014	1,136	2.7	875	2.4	261	4.8
2015	1,145	2.7	942	2.6	203	3.7
2016	1,146	2.6	950	2.5	196	3.3
Trisomy 18						
2011	136	0.3	125	0.4	11	0.2
2012	162	0.4	150	0.4	12	0.2
2013	150	0.4	130	0.4	20	0.4
2014	139	0.3	123	0.3	16	0.3
2015	147	0.3	129	0.4	18	0.3
2016	171	0.4	142	0.4	29	0.5
Trisomy 13						
2011	145	0.4	142	0.4	3	0.1
2012	170	0.4	162	0.5	8	0.2
2013	162	0.4	148	0.4	14	0.3
2014	152	0.4	138	0.4	14	0.3
2015	161	0.4	149	0.4	12	0.2
2016	174	0.4	161	0.4	13	0.2
Any one or more of trisomy 21, 18 or 13						
2011	1,104	2.8	883	2.5	221	5.0
2012	1,160	2.9	877	2.5	283	5.7
2013	1,111	2.7	855	2.4	256	4.9
2014	1,162	2.8	888	2.4	274	5.0
2015	1,168	2.8	947	2.6	221	4.0
2016	1,189	2.7	969	2.6	220	3.7

Increased risk screening results stratified by risk level

Table 22 shows the number of increased risk results stratified by risk level for each of trisomy 21, 18 and 13 for the 2016 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 22 will be greater than the total number of increased risk results for 2016.

Table 22: Increased risk screening results for trisomy 21, 18 and 13 by risk level, January 2016 to December 2016

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 to 1:20	286	61	61
>1:20 to 1:50	184	22	33
>1:50 to 1:300	676	88	80

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.

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

Table 23 shows the diagnostic testing rate from 2011 to 2016 by trimester of screen. In 2016, for every 100 women that received an increased risk result after a first or second trimester screen, 46 women had a diagnostic test. This is lower than previous years and there has been a downward trend since 2013. The diagnostic testing rate was lower for women who received an increased risk after a second trimester screen (41 women per 100 increased risk screens) compared with first trimester screens (47 per 100 increased risk screens). See Appendix 3 for a summary of diagnostic test results for women who had increased risk screen in 2016, as well as pregnancy outcomes (where known) for women who did not have a prenatal diagnostic test.

Table 23: Diagnostic testing volumes for women with increased risk screens by trimester of screen, January 2011 to December 2016

	Diagnostic tests per 100 increased risk screens					
Trimester of screen	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
T1 screen	65.2	66.2	66.0	62.5	59.0	46.9
T2 screen	43.4	42.8	46.9	47.4	44.3	40.5
Total screens	**60.9**	**60.5**	**61.6**	**59.0**	**56.3**	**45.7**

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 24. Many DHBs have low numbers and care should be taken with comparisons.

Table 24: Diagnostic testing volumes for women with increased risk screens by DHB, January 2011 to December 2016

	Number of diagnostic tests						Tests per 100 increased risk screens					
DHB	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
Northland	24	13	28	26	21	12	49.0	38.2	56.0	59.1	48.8	40.0
Waitemata	140	138	141	116	107	82	68.0	67.6	72.7	61.7	57.5	44.6
Auckland	117	119	89	89	76	72	71.3	69.2	67.4	55.3	53.5	45.0
Counties Manukau	67	77	73	76	86	78	54.5	51.7	47.1	50.3	53.8	54.9
Waikato	15	26	41	41	42	45	20.5	38.2	57.7	64.1	60.0	52.9
Lakes	15	23	21	21	28	16	55.6	69.7	67.7	53.8	71.8	59.3
Bay of Plenty	11	22	21	21	20	17	36.7	68.8	53.8	63.6	66.7	44.7
Tairawhiti	5	5	2	2	4	1	83.3	50.0	25.0	33.3	57.1	14.3
Hawke's Bay	22	18	21	20	15	8	62.9	50.0	53.8	58.8	51.7	28.6
Taranaki	14	18	18	12	10	8	63.6	75.0	66.7	48.0	43.5	36.4
MidCentral	20	20	10	11	8	15	54.1	62.5	38.5	57.9	44.4	46.9
Whanganui	4	4	6	3	4	6	33.3	33.3	46.2	60.0	66.7	66.7
Capital and Coast	53	61	55	46	65	41	73.6	69.3	74.3	59.7	60.7	60.3
Hutt Valley	14	24	18	15	18	15	56.0	63.2	58.1	53.6	64.3	45.5
Wairarapa	5	7	9	1	3	3	71.4	100.0	81.8	25.0	50.0	60.0
Nelson Marlborough	23	11	17	19	15	14	67.6	47.8	89.5	79.2	57.7	51.9
West Coast	3	2	2	8	3	6	50.0	50.0	40.0	42.1	50.0	85.7
Canterbury	77	67	74	122	83	80	67.0	60.4	60.2	65.6	50.6	36.7
South Canterbury	6	4	4	3	9	4	54.5	40.0	40.0	50.0	75.0	30.8
Southern	37	43	34	33	40	20	74.0	58.9	64.2	67.3	60.6	37.0
Total	**672**	**702**	**684**	**685**	**657**	**543**	**Av. 60.9**	**60.5**	**61.6**	**59.0**	**56.3**	**45.7**

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

Table 25 shows the diagnostic testing rate for women with increased risk screens by age, ethnicity and NZ deprivation quintile for 2011 to 2016.

In 2016, diagnostic testing rates were highest for women of Asian ethnicity (56 per 100 increased risks), followed by Māori (47 per 100 increased risks), with much lower rates for Pacific women (34 per 100 increased risks).

Table 25: Diagnostic testing volumes for women with increased risk screening results by age at screen, ethnicity and deprivation, January 2011 to December 2016

	Diagnostic tests per 100 increased risk screens					
	2011	**2012**	**2013**	**2014**	**2015**	**2016**
Age at screen						
Under 20 years	50.0	35.7	28.6	50.0	53.8	45.5
20–24 years	60.0	55.4	62.5	53.9	51.7	55.6
25–29 years	65.7	60.2	60.5	62.7	58.1	49.4
30–34 years	65.8	70.5	68.1	64.9	61.8	47.4
35–39 years	64.5	60.1	62.6	57.1	57.0	46.0
40–44 years	50.6	56.1	57.4	58.1	50.9	39.0
45 years and over	43.5	40.0	44.4	36.0	41.2	27.8
Ethnicity						
Māori	42.7	43.9	52.5	38.4	45.1	46.7
Pacific	35.5	36.4	38.2	39.2	36.2	34.3
Asian	70.7	70.7	69.2	67.0	63.3	56.3
Other	65.1	64.7	65.2	62.8	58.7	42.1
NZ deprivation quintile						
Quintile 1	70.5	68.0	71.4	65.8	62.1	43.1
Quintile 2	71.0	69.3	64.8	64.2	63.1	49.1
Quintile 3	60.4	65.0	62.4	57.6	58.6	43.4
Quintile 4	55.1	52.1	58.6	60.0	57.3	47.2
Quintile 5	48.0	48.8	53.2	49.0	41.6	45.5

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 26 shows the number of diagnostic tests for women that received an increased risk result during 2016 for one or more of trisomy 21, 18 or 13, stratified by risk level. As expected the number of women having a diagnostic test increased with increasing risk level, increasing from 40 to 60 tests per 100 women with an increased risk.

Table 26: Diagnostic testing volumes for women with increased risk screens by risk level, January 2016 to December 2016

Risk level	Number of diagnostic tests	Number of increased risk screens	Tests per 100 increased risk screens
1:5 to 1:20	180	300	60.0
>1:20 to 1:50	88	188	46.8
>1:50 to 1:300	275	701	39.2

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. 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 0.55 per 100 low risk screens in 2016. This rate has decreased over the reporting period.

Table 27: Diagnostic testing volumes for women with low risk screens by trimester of screen, January 2011 to December 2016

	Diagnostic tests per 100 low risk screens					
Trimester of screen	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
T1 screen	0.89	0.92	0.77	0.68	0.74	0.53
T2 screen	0.83	0.67	0.48	0.56	0.36	0.69
Total screens	0.89	0.89	0.73	0.67	0.69	0.55

Diagnostic testing volumes for women with low risk screens by DHB

The rate of diagnostic testing by DHB for women with low risk screens has varied each year from 2011 to 2016, as shown in Table 28. Given the low numbers involved, caution should be taken in making comparisons, some numbers have been withheld where denominators are lower than 10.

Table 28: Total diagnostic testing volumes for women with low risk screens by DHB January 2011 to December 2016

	Number of diagnostic tests						Tests per 100 low risk screens					
DHB	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
Northland	5	2	7	0	7	5	0.56	0.20	0.74	0.00	0.66	0.45
Waitemata	62	61	54	35	33	37	1.04	1.02	0.89	0.57	0.55	0.59
Auckland	72	73	55	38	36	20	1.62	1.63	1.17	0.79	0.80	0.46
Counties Manukau	40	25	27	18	23	28	0.87	0.51	0.57	0.35	0.45	0.53
Waikato	6	18	18	30	21	16	0.17	0.52	0.51	0.80	0.56	0.41
Lakes	3	3	3	5	8	0	0.37	0.34	0.35	0.54	0.84	0.00
Bay of Plenty	5	10	9	14	7	12	0.31	0.56	0.54	0.80	0.38	0.59
Tairawhiti	0	3	0	1	0	0	0.00	0.95	0.00	0.29	0.00	0.00
Hawke's Bay	11	8	6	7	8	4	1.00	0.65	0.48	0.59	0.64	0.29
Taranaki	6	11	9	3	1	1	0.67	1.31	1.11	0.33	0.10	0.12
MidCentral	7	4	9	8	11	4	0.70	0.39	0.81	0.72	0.93	0.30
Whanganui	4	4	2	2	2	2	1.24	1.14	0.56	0.47	0.42	0.39
Capital and Coast	24	18	21	15	22	19	0.94	0.67	0.84	0.60	0.86	0.72
Hutt Valley	12	10	8	11	9	6	1.01	0.82	0.66	0.88	0.69	0.44
Wairarapa	1	0	0	0	1	1	0.31	0.00	0.00	0.00	0.30	0.28
Nelson Marlborough	9	14	12	5	9	9	0.71	1.15	1.01	0.41	0.77	0.77
West Coast	0	0	1	1	2	2	0.00	0.00	0.37	0.39	0.79	0.83
Canterbury	41	46	31	45	52	37	0.96	1.04	0.67	0.96	1.08	0.74
South Canterbury	2	3	1	0	2	7	0.41	0.57	0.19	0.00	0.39	1.35
Southern	27	38	17	33	29	23	1.11	1.48	0.67	1.37	1.12	0.87
Total	**337**	**351**	**290**	**271**	**283**	**233**	**Av. 0.89**	**0.89**	**0.73**	**0.67**	**0.69**	**0.55**

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

Table 29 shows the rate of diagnostic testing for women with low risk screening results by age, ethnicity and NZ deprivation quintile. In 2016, the rate of diagnostic testing was higher for women in the older age groups and women in less deprived regions. Pacific women were the least likely to have a diagnostic test after a low risk screen.

Table 29: Diagnostic tests per 100 low risk screens by age, ethnicity and NZ deprivation quintile, January 2011 to December 2016

	Diagnostic tests per 100 low risk screens					
	2011	**2012**	**2013**	**2014**	**2015**	**2016**
Age at screen						
Under 20 years	0.39	0.71	0.38	0.44	0.33	0.34
20–24 years	0.37	0.34	0.32	0.37	0.35	0.43
25–29 years	0.39	0.45	0.37	0.49	0.52	0.50
30–34 years	0.57	0.64	0.53	0.53	0.60	0.54
35–39 years	1.88	1.56	1.19	0.98	1.11	0.66
40–44 years	5.32	5.59	5.30	3.92	3.04	1.33
45 years and over	7.50	10.64	6.98	0.00	2.13	3.28
Ethnicity						
Māori	0.45	0.70	0.57	0.46	0.46	0.50
Pacific	0.51	0.33	0.28	0.28	0.48	0.35
Asian	0.89	0.87	0.65	0.58	0.80	0.54
Other	1.00	0.98	0.83	0.78	0.72	0.58
NZ deprivation quintile						
Quintile 1	1.45	1.66	1.12	0.91	0.80	0.76
Quintile 2	1.14	1.03	0.76	0.70	0.91	0.60
Quintile 3	0.81	0.62	0.72	0.68	0.63	0.57
Quintile 4	0.70	0.80	0.62	0.62	0.72	0.54
Quintile 5	0.39	0.43	0.52	0.49	0.43	0.33

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

Table 30 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 (2011–2016). The aggregated rate of diagnostic testing is more than 14 times higher for the highest category compared with the lowest category and the rate drops away rapidly as risk decreases below 1:1000.

Table 30: Diagnostic tests per 100 low risk screens stratified by risk level, January 2011–December 2016 aggregated

Risk level	Number of diagnostic tests	Number of low risk screens	Tests per 100 low risk screens
1:301 to 1:500	194	3,702	5.24
1:501 to 1:1000	271	9,514	2.85
1:1001 to 1:2000	252	16,604	1.52
1:2001 to 1:3000	165	14,442	1.14
1:3001 to 1:4000	90	13,178	0.68
1:4001 to 1:5000	87	11,930	0.73
1:5001 to 1:10,000	251	48,309	0.52
1:10,001 to 1:100,000	455	123,339	0.37

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 2016 year, 212 diagnostic tests were completed for unscreened women. This is lower than the number undertaken in previous years. Table 31 shows the number of tests by DHB and Table 32 shows the breakdown by age, ethnicity and NZ deprivation quintile.

Table 31: Diagnostic testing volumes for unscreened women by DHB, January 2012 to December 2016

	Number of diagnostic tests				
DHB	**2012**	**2013**	**2014**	**2015**	**2016**
Northland	10	6	7	8	6
Waitemata	37	24	22	22	19
Auckland	31	23	25	18	23
Counties Manukau	19	27	21	18	21
Waikato	16	24	14	15	16
Lakes	2	5	6	8	3
Bay of Plenty	10	18	12	14	10
Tairawhiti	5	0	1	3	2
Hawke's Bay	11	6	7	7	8
Taranaki	13	11	5	11	4
MidCentral	9	11	11	8	9
Whanganui	4	2	3	2	2
Capital and Coast	17	16	30	36	25
Hutt Valley	9	11	11	22	10
Wairarapa	5	1	1	3	3
Nelson Marlborough	7	1	4	6	5
West Coast	0	1	1	0	0
Canterbury	27	23	37	30	30
South Canterbury	0	2	4	2	2
Southern	17	18	13	19	14
Total	**249**	**230**	**235**	**252**	**212**

Table 32: Total diagnostic testing volumes for unscreened women by age, ethnicity and deprivation quintile, January 2012 to December 2016

	2012	2013	2014	2015	2016
Age					
Under 20 years	15	13	10	16	12
20–24 years	32	33	29	19	17
25–29 years	43	35	39	53	36
30–34 years	62	56	66	70	60
35–39 years	55	50	54	54	56
40–44 years	41	39	34	35	28
45 years and over	1	4	3	5	3
Ethnicity					
Māori	33	49	31	44	32
Pacific	17	14	20	21	11
Asian	39	31	29	33	36
Other	160	136	155	154	133
NZ deprivation quintile					
Quintile 1	62	36	55	48	45
Quintile 2	45	47	39	48	46
Quintile 3	40	40	49	51	45
Quintile 4	58	59	46	52	42
Quintile 5	44	48	46	53	33
Unknown					1

Diagnostic results for unscreened women

A breakdown of prenatal diagnostic testing results for unscreened women for the 2016 year is given in Table 33. Of the 212 diagnostic tests in 2016 for unscreened women, 57 (74%) had a normal karyotype.

Table 33: Total diagnostic testing results for unscreened women, January 2016 to December 2016

Karyotype result	Number	Percentage
Normal karyotype	157	74.1%
Trisomy 21	17	8.0%
Trisomy 18	6	2.8%
Trisomy 13	4	1.9%
Turner syndrome	5	2.4%
Triploidy	2	0.9%
Other chromosome abnormality	21	9.9%
Total	**212**	**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.

Also in 2016, 3 babies with a positive test for one of the trisomies could not be matched to a maternal screen. These babies may have been either a true positive, a false negative or have an unscreened mother.

The overall PPV for 2016 was 0.09, slightly lower than previous years (see Table 35). A value of 0.09 means that if a woman receives an increased risk result for trisomy 21, 18 or 13 there is a 9% 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 0.13 for first trimester screens.

Table 34: Positive predictive value of screening for trisomy 21, 18 or 13, January 2011 to December 2016

Year	True positives	False positives	PPV	95% confidence interval
2011	136	968	0.123	(0.105, 0.144)
2012	144	1,016	0.124	(0.106, 0.144)
2013	142	969	0.128	(0.109, 0.149)
2014	122	1,040	0.105	(0.089, 0.124)
2015	133	1,035	0.114	(0.097, 0.133)
2016	110	1,079	0.093	(0.077, 0.110)

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21 the PPV for 2016 was lower than previous years at 0.06 (see Table 35). This means that if a woman receives an increased risk result for trisomy 21 there is a 6% probability that she is carrying a fetus with trisomy 21.

Table 35: Positive predictive of screening for trisomy 21, January 2011 to December 2016

Year	True positives	False positives	PPV	95% confidence interval
2011	88	998	0.08	(0.066, 0.099)
2012	97	1,051	0.08	(0.07, 0.102)
2013	109	980	0.10	(0.084, 0.119)
2014	90	1,046	0.08	(0.065, 0.096)
2015	99	1,046	0.09	(0.072, 0.104)
2016	74	1,072	0.06	(0.052, 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 2016 was higher than the PPV for trisomy 21 at 0.15 and 0.06 respectively (see Table 36). However, the number of positive diagnoses for these two trisomies is low so caution should be taken when interpreting these results.

Table 36: Positive predictive of screening for trisomy 13 or 18, January 2011 to December 2016

Year	True positives	False positives	PPV	95% confidence interval
2011	44	128	0.26	(0.196, 0.326)
2012	39	148	0.21	(0.156, 0.272)
2013	30	153	0.16	(0.117, 0.224)
2014	27	147	0.16	(0.109, 0.216)
2015	33	148	0.18	(0.133, 0.245)
2016	32	181	0.15	(0.108, 0.204)

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

Table 37 shows PPV stratified by the risk level indicated in the screening result. Data have been aggregated across the 2011 to 2016 period. Women that received an increased risk result of 1:5 to 1:20 for trisomy 21 had a 26% probability that they were carrying a fetus with trisomy 21. As expected the PPV was lower for women with increased risks of 1:21 to 1:50, and lower again for women with increased risk results of 1:51 to 1:300.

Table 37: Positive predictive of screening for trisomy 21 stratified by risk level, aggregated 2011–2016

Risk level	True positives	False positives	PPV
1:5 to 1:20	406	1,135	0.26
1:21 to 1:50	70	932	0.07
1:51 to 1:300	81	4,126	0.02
Total	557	6,193	0.08

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

The following tables show true positives, false positives and PPV aggregated for 2011–2016 by age, ethnicity and deprivation. The PPV of screening for trisomy 21 also varied by age group, as shown in Table 38. The aggregated PPV for 2011 to 2016 was highest for women 30 years and over (0.09) compared to women under 30 (0.05).

Table 38: Positive predictive of screening for trisomy 21 by age, aggregated 2011–2016

Age	True positives	False positives	PPV
Under 20 years	5	75	0.06
20–24 years	17	353	0.05
25–29 years	40	718	0.05
30–34 years	108	1,447	0.07
35–39 years	222	2,073	0.10
40–44 years	158	1,402	0.10
45 years and over	7	125	0.05

Table 39 shows aggregated PPV data across all years by ethnicity. Pacific women had the lowest PPV (0.03 or 3%) and women in the Other ethnicity had the highest at (0.11 or 11%).

Table 39: Positive predictive of screening for trisomy 21 by ethnicity, aggregated 2011–2016

Ethnicity	True positives	False positives	PPV
Māori	39	702	0.05
Pacific	17	623	0.03
Asian	70	1,385	0.05
Other	431	3,483	0.11

Table 40 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 40: Positive predictive of screening for trisomy 21 by NZ deprivation quintile, aggregated 2011–2016

NZ dep quintile	True positives	False positives	PPV
Quintile 1	148	1,161	0.11
Quintile 2	123	1,175	0.09
Quintile 3	107	1,193	0.08
Quintile 4	106	1,268	0.08
Quintile 5	73	1,396	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 positives 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 2016 was 0.02 (or 2%) similar to previous years. This means that out of all women who had a negative diagnostic test or a baby without a trisomy, 2% had received an increased risk result for trisomy 21, 18 or 13.

Table 41: False positive rate for trisomy 21, 18 or 13, January 2011 to December 2016

Year	False positives	True negatives	False positive rate	95% confidence interval
2011	968	38,039	0.02	(0.02, 0.03)
2012	1,016	39,451	0.03	(0.02, 0.03)
2013	969	39,584	0.02	(0.02, 0.03)
2014	1,040	40,547	0.03	(0.02, 0.03)
2015	1,035	41,063	0.02	(0.02, 0.03)
2016	1,079	42,300	0.02	(0.02, 0.03)

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

Table 42: False positive rate for trisomy 21, 18 or 13 by trimester of screen, January to December 2016

Trimester	False positives	True negatives	False positive rate
T1 screens	865	36,519	0.023
T2 screens	214	5,781	0.036
Total screens	**1,079**	**42,300**	**0.025**

The false positive rate for trisomy 21 when considered alone was similar to the overall false positive rate (see Table 43). However, the combined false positive rate for trisomy 18 and trisomy 13 is much lower (0.004 for 2016, see Table 44).

Table 43: False positive rate for trisomy 21, January 2011 to December 2016

Year	False positives	True negatives	False positive rate	95% confidence interval
2011	998	38,069	0.03	(0.02, 0.03)
2012	1,051	39,475	0.03	(0.02, 0.03)
2013	980	39,618	0.02	(0.02, 0.03)
2014	1,046	40,583	0.03	(0.02, 0.03)
2015	1,046	41,093	0.02	(0.02, 0.03)
2016	1,072	42,352	0.02	(0.02, 0.03)

Table 44: False positive rate for trisomy 18 and 13, January 2011 to December 2016

Year	False positives	True negatives	False positive rate	95% confidence interval
2011	128	38,993	0.003	(0.003, 0.004)
2012	148	40,441	0.004	(0.003, 0.004)
2013	153	40,535	0.004	(0.003, 0.004)
2014	147	41,547	0.004	(0.003, 0.004)
2015	148	42,067	0.004	(0.003, 0.004)
2016	181	43,293	0.004	(0.004, 0.005)

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

The false positive rate for trisomy 21 increases with age (see Table 45). For example, the false positive rate for women under 20 years in 2016 was 0.01 (1%) compared with 0.21 (21%) 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 45: False positive rate for trisomy 21 by age, aggregated January 2011 to December 2016

Age	2011	2012	2013	2014	2015	2016
Under 20 years	0.01	0.01	0.00	0.01	0.01	0.01
20–24 years	0.01	0.01	0.01	0.01	0.01	0.01
25–29 years	0.01	0.01	0.01	0.01	0.01	0.01
30–34 years	0.02	0.02	0.02	0.02	0.02	0.02
35–39 years	0.05	0.05	0.05	0.05	0.05	0.05
40–44 years	0.16	0.16	0.15	0.15	0.19	0.15
45 years and over	0.33	0.33	0.37	0.32	0.27	0.21

The false positive rate for 2016 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 (see Table 46).

Table 46: False positive rate for trisomy 21 by ethnicity, January 2011 to December 2016

Ethnicity	2011	2012	2013	2014	2015	2016
Māori	0.02	0.02	0.02	0.03	0.02	0.02
Pacific	0.04	0.04	0.04	0.04	0.04	0.04
Asian	0.03	0.03	0.03	0.03	0.03	0.03
Other	0.02	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 2016 and previous years (see Table 47).

Table 47: False positive rate for trisomy 21 by NZ deprivation quintile, January 2011 to December 2016

NZ dep quintile	2011	2012	2013	2014	2015	2016
Quintile 1	0.03	0.03	0.03	0.03	0.02	0.03
Quintile 2	0.03	0.03	0.02	0.02	0.03	0.03
Quintile 3	0.02	0.03	0.02	0.03	0.02	0.03
Quintile 4	0.02	0.02	0.02	0.02	0.02	0.02
Quintile 5	0.03	0.03	0.03	0.03	0.02	0.03

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.

Detection rate for screening

The overall detection rate for trisomy 21, 18 and 13 for the six years ending 2016 is given in Table 48. Rates for trisomy 21 alone, and for trisomies 18 and 13 together are given in Tables 49 and 50 respectively. As each of these tables show, detection rates fluctuated over this period.

The overall detection rate for trisomy 21, 18 and 13 for 2016 was 0.79 (79%) (see Table 48). A detection rate of 0.79 means that there is a 79% probability that a woman carrying a fetus with one of trisomy 21, 18 or 13 will have an increased risk screening result for trisomy 21, 18 or 13.

Table 48: Detection rate for trisomy 21, 18 or 13, January 2011 to December 2016

Year	True positives	False negatives	Detection rate	95% confidence interval
2011	136	38	0.78	(0.71, 0.84)
2012	143	37	0.79	(0.73, 0.85)
2013	142	38	0.79	(0.72, 0.84)
2014	122	27	0.82	(0.75, 0.87)
2015	132	25	0.84	(0.78, 0.89)
2016	110	30	0.79	(0.71, 0.85)

The detection rate for trisomy 21 alone is shown in Table 49. The rate for 2016 was similar (0.78) to the overall rate for trisomy 21, 18 and 13. The detection rate for trisomy 13 and 18 was lower at 0.71.

Table 49: Detection rate for trisomy 21, January 2011 to December 2016

Year	True positives	False negatives	Detection rate	95% confidence interval
2011	88	26	0.77	(0.69, 0.84)
2012	97	25	0.80	(0.71, 0.86)
2013	109	26	0.81	(0.73, 0.87)
2014	90	17	0.84	(0.76, 0.9)
2015	99	18	0.85	(0.77, 0.9)
2016	74	21	0.78	(0.69, 0.85)

Table 50: Detection rate for trisomy 13 or 18, January 2011 to December 2016

Year	True positives	False negatives	Detection rate	95% confidence interval
2011	44	16	0.73	(0.61, 0.83)
2012	39	20	0.66	(0.53, 0.77)
2013	30	15	0.67	(0.52, 0.79)
2014	27	15	0.64	(0.49, 0.77)
2015	33	8	0.80	(0.66, 0.9)
2016	32	13	0.71	(0.57, 0.82)

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 six 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 0.61 (61%) for women 20–24 years to 0.93 (93%) for women 40–44 years (see Table 51).

Table 51: Detection rate for trisomy 21 by age, aggregated 2011–2016

	True positives	False negatives	Detection rate#
Age	Positive diagnostic test/ infant diagnosis after increased risk screen	Positive diagnostic test/ infant diagnosis after low risk screen	
Under 20 years	5	5	0.50
20–24 years	17	11	0.61
25–29 years	40	17	0.70
30–34 years	108	48	0.69
35–39 years	222	40	0.85
40–44 years	158	12	0.93
45 years and over	7	0	

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

The aggregated detection rates by ethnicity ranged from 0.71 (71%) for Pacific to 0.82 (82%) for women of Other ethnicity (see Table 52). Low numbers mean these rates should be interpreted with caution.

Table 52: Detection rate for trisomy 21 by ethnicity, aggregated 2011–2016

	True positives	False negatives	Detection rate
Ethnicity	Positive diagnostic test/ infant diagnosis after increased risk screen	Positive diagnostic test/ infant diagnosis after low risk screen	
Māori	39	11	0.78
Pacific	17	7	0.71
Asian	70	22	0.76
Other	431	93	0.82

The aggregated detection rates by deprivation quintile ranged from 0.78 to 0.83 (see Table 53). There was no clear trend with increasing deprivation.

Table 53: Detection rate for trisomy 21 by NZ deprivation quintile, aggregated 2011–2016

	True positives	False negatives	Detection rate
NZ deprivation quintile	Positive diagnostic test/ infant diagnosis after increased risk screen	Positive diagnostic test/ infant diagnosis after low risk screen	
Quintile 1	148	30	0.83
Quintile 2	123	32	0.79
Quintile 3	107	25	0.81
Quintile 4	106	26	0.80
Quintile 5	73	20	0.78

Appendix 1: Indicator definitions

Table 54: Definitions used for monitoring indicators

Indicator	Methodology
Indicator 1: Screens commenced	Numerator: number of women who start screening Denominator: number of live births and stillbirths
Indicator 2: Screens completed	Numerator: number of women who have a risk result calculated Denominator: number of live births and stillbirths
Indicator 3: Pathway variances	Numerator: completed second trimester screens that have an ultrasound or PAPP-A reading recorded against them Denominator: number of completed second trimester screens
Indicator 4: Incomplete screens	Numerator: number of screens commenced that have no risk result reported against them Denominator: number of screens commenced
Indicator 5: Increased risk screening results	Numerator: number of women who receive an increased risk result Denominator: number of women who have a risk result calculated
Indicator 6: Diagnostic testing, increased risk screens	Numerator: number of women with an increased risk result that have a diagnostic test Denominator: number of women with increased risk results
Indicator 7: Diagnostic testing, low risk screens	Numerator: number of women with a low risk result that have a diagnostic test Denominator: number of women with low risk results
Indicator 8: Diagnostic testing, unscreened women	Number of women who have diagnostic test that have not participated in screening
Indicator 9: Positive predictive value	Numerator: number of women given an increased risk screen result who have a positive diagnostic test/baby with positive diagnosis Denominator: number of screened women with an increased risk result
Indicator 10: False positive rate	Numerator: number of women given an increased risk screen result who do not have a positive diagnostic test/baby with positive diagnosis Denominator: number of screened women who do not have a positive diagnostic test/baby with positive diagnosis
Indicator 11: Detection rate	Numerator: number of women given an increased risk screen result who have a positive diagnostic test/baby with positive diagnosis Denominator: number of screened women who have a positive diagnostic test/baby with positive diagnosis

Calculation rules

- Screen date is the date given as the 'Collected date' in the lab system.
- If a woman has more than one screen for the same pregnancy (defined as being within 112 days) then the first completed screen has been retained for the analysis and the others excluded.
- Denominator is live births and still births >20 weeks or >400g.
- Tests on products of conception are excluded from prenatal tests for the purposes of indicators 6, 7 and 8. However, they are included 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[3] was obtained from the national Maternity Collection for each year.

Table 55: Live births and still births by district health board 2011–2016

DHB	2011	2012	2013	2014	2015	2016
Northland	2,302	2,300	2,124	2,099	2,135	2,265
Waitemata	7,878	7,970	7,654	7,845	7,555	7,934
Auckland	6,535	6,697	6,242	6,305	5,900	5,905
Counties Manukau	8,732	8,768	8,181	8,291	8,197	8,242
Waikato	5,372	5,485	5,216	5,250	5,274	5,359
Lakes	1,590	1,558	1,420	1,392	1,509	1,545
Bay of Plenty	2,859	2,969	2,753	2,784	2,791	2,898
Tairawhiti	739	736	705	688	738	775
Hawke's Bay	2,259	2,256	2,153	2,072	1,994	2,060
Taranaki	1,566	1,558	1,521	1,519	1,515	1,434
MidCentral	2,298	2,151	2,120	2,090	2,111	2,082
Whanganui	831	874	828	818	816	800
Capital and Coast	3,858	3,866	3,631	3,528	3,537	3,456
Hutt Valley	2,053	2,008	1,911	1,854	1,967	1,966
Wairarapa	530	511	501	473	463	462
Nelson Marlborough	1,652	1,530	1,546	1,419	1,417	1,548
West Coast	405	409	376	350	357	318
Canterbury	6,062	5,985	5,825	5,997	6,205	6,308
South Canterbury	570	648	639	652	660	650
Southern	3,675	3,594	3,448	3,287	3,411	3,320
Total	**61,766**	**61,873**	**58,794**	**58,713**	**58,552**	**59,327**

[3] Births reaching at least 20 weeks gestation or ≥400 g birth weight.

Table 56: Live births and still births by age group, 2011–2016

Age group	2011	2012	2013	2014	2015	2016
Under 20	4,049	3,906	3,327	2,990	2,784	2,443
20–24	11,690	11,461	10,803	10,275	9,941	9,584
25–29	15,542	15,933	15,262	15,697	15,708	16,546
30–34	17,222	17,451	16,771	17,578	17,908	18,374
35–39	10,716	10,409	10,039	9,681	9,761	9,964
40–44	2,403	2,580	2,435	2,347	2,298	2,276
45 and over	126	120	143	132	139	126
Unknown	18	13	14	13	13	14
Total	**61,766**	**61,873**	**58,794**	**58,713**	**58,552**	**59,327**

Table 57: Live births and still births by 2013 NZ deprivation quintile, 2011–2016

NZ deprivation quintile	2011	2012	2013	2014	2015	2016
Quintile 1	8,500	8,672	8,175	8,468	8,242	8,669
Quintile 2	9,502	9,614	9,244	9,171	9,332	9,675
Quintile 3	11,151	11,163	10,623	10,562	10,584	10,716
Quintile 4	13,789	13,657	13,417	13,273	13,243	13,289
Quintile 5	18,800	18,749	17,303	17,214	17,036	16,965
Unknown	24	18	32	25	115	13
Total	**61,766**	**61,873**	**58,794**	**58,713**	**58,552**	**59,327**

Table 58: Live births and still births by ethnicity, 2011-2016

Ethnicity	2011	2012	2013	2014	2015	2016
Māori	15,892	15,783	14,649	14,299	14,579	14,749
Pacific	7,064	6,880	6,355	6,166	6,063	5,838
Asian	7,127	8,448	8,147	9,188	9,212	10,523
Other	31,683	30,762	29,643	29,060	28,698	28,217
Total	**61,766**	**61,873**	**58,794**	**58,713**	**58,552**	**59,327**

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 1,189 screens that had an increased risk for trisomy 21, 18 or 13 during 2016, 543 (46%) had a prenatal diagnostic test (CVS or Amniocentesis) and 646 (54%) did not. Table 59 shows the diagnostic testing results for the 543 prenatal tests, of which 108 had an abnormal karyotype, including 57 confirmed with Down syndrome. Table 60 shows a breakdown of pregnancy outcomes for the 646 women that had an increased risk screen but did not have a prenatal diagnostic test.

Table 59: Diagnostic results for women that accessed a prenatal diagnostic test following an increased risk screen for trisomy 21, 18 or 13 during the 2016 year

Karyotype result	Number	Percentage
Normal karyotype	435	80.1%
Confirmed Down syndrome	57	10.5%
Other result*	51	9.4%
Total	**543**	**100.0%**

Table 60: Pregnancy outcomes (where known) for women that did not have a prenatal diagnostic test following an increased risk screen for trisomy 21, 18 or 13 during the 2016 year

Result	Number
No abnormality detected on postnatal diagnostic test	7
Trisomy 21	17
Trisomy 18	8
Trisomy 13	1
Turner syndrome	4
Triploidy	1
Sex chromosome aneuploidy (other than non-mosaic 45, X)	1
Autosomal trisomy (other than 13, 18, 21) (including mosaic)	1
Other	24
No link to a diagnosis	582
Total	**646**

Appendix 4: Measuring screening performance

Figure 12 shows the categorisation of screening results used to calculate screening performance measures such as positive predictive value, false positive rate and detection rate. The examples given in this appendix focus on trisomy 21.

Figure 12: Categorisation of screening results

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

Positive predictive value and positive test rate

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

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)

Appendix 5: False negative screens by risk level

There were 195 false negative screens in total across the six-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 61 shows the number of false negatives for each of the six 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 2016.

Table 61: False negative screens for trisomy 21, 18 and 13 by risk level, January 2011 to December 2016

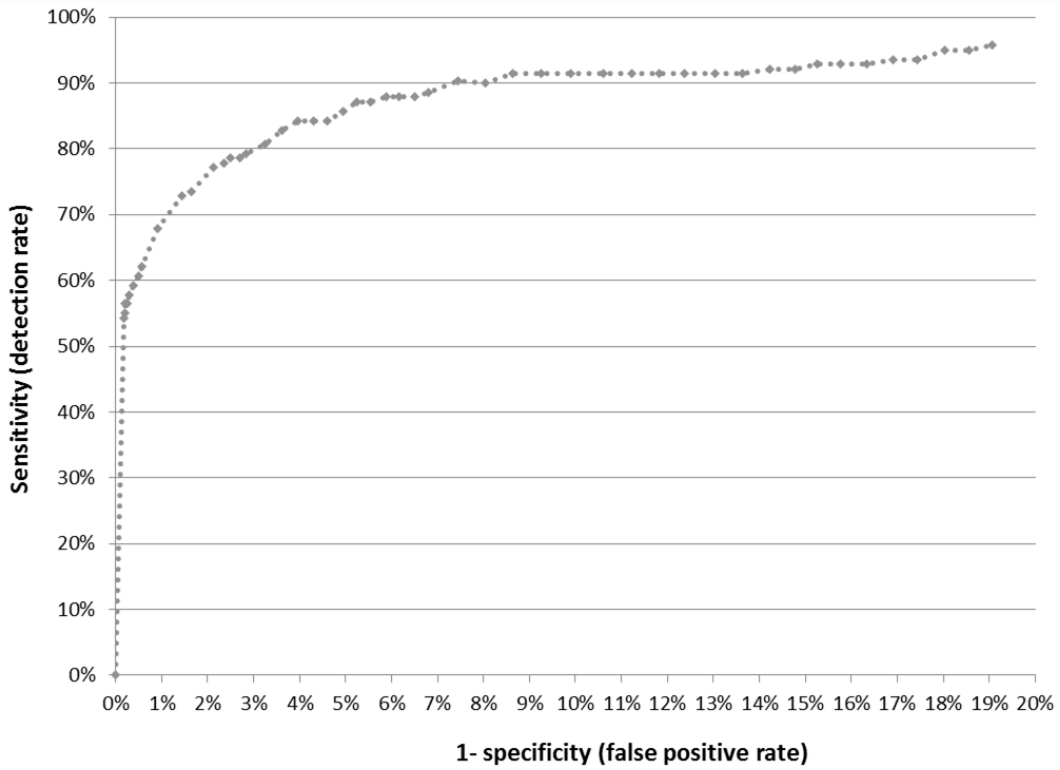
	False negatives						% of negative screens that are false negatives					
Risk level	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**
1:301 to 1:500	9	5	7	6	4	8	1.69	0.83	1.14	0.94	0.63	1.25
1:510 to 1:1,000	9	4	8	5	10	7	0.58	0.26	0.52	0.31	0.58	0.46
1:1,100 to 1:2,000	8	6	6	4	4	3	0.30	0.22	0.21	0.14	0.14	0.11
1:2,100 to 1:3,000	2	5	2	5	2	6	0.09	0.21	0.09	0.20	0.08	0.25
1:3,100 to 1:4,000	0	3	3	0	1	0	–	0.14	0.13	–	0.04	–
1:4,100 to 1:5,000	3	3	1	2	0	0	0.16	0.15	0.05	0.10	–	–
1:5,100 to 1:10,000	5	6	5	2	3	2	0.06	0.08	0.06	0.02	0.03	0.02
Less than 1:10,000	2	5	6	3	1	4	0.01	0.02	0.03	0.01	0.00	0.02
Total	**38**	**37**	**38**	**27**	**25**	**30**	**0.10**	**0.09**	**0.10**	**0.07**	**0.06**	**0.07**

Appendix 6: ROC curve

Figure 13 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 2016 was 79%, and the false positive rate was 2.5%. To create the graph the detection rate and false positive rate were calculated for a range of other cut off points in order to plot the curve. What the curve shows is that if the cut off was lowered to increase the detection rate to 85%, the false positive rate would increase from 2.5% to 4.6%. This occurs at a risk cut off of 1:600.

Figure 13: ROC curve for trisomy 21, 18 and 13 screening 2016



Appendix 7: Glossary

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

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

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

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

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

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

Detection rate – the ability of screening to identify individuals with the condition screened for. A test with a high detection rate will have few false negative results. Also referred to as sensitivity.

False negative result – when a woman receives a low risk screening result but the baby does have the condition screened for.

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

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

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).

True positive – when a woman receives an increased risk screening result and the baby does have the condition screened for.

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