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Antenatal Screening for Down Syndrome and Other Conditions

2018 Monitoring Report

National Screening Unit

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Executive summary

This report presents data on antenatal screening for Down syndrome and other conditions for the six calendar years from 1 January 2013 to 31 December 2018 and is based on screens that commenced during that time.

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), Edwards syndrome (trisomy 18), Patau syndrome (trisomy 13) 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 ultrasound scan is ideally performed around 12 weeks. Second trimester maternal serum screening should be completed between 14 weeks and 20 weeks gestation. The recommended timing for this test is 14 to 18 weeks.

Key points for 2018

- Screening was commenced for 83 percent of women who gave birth in 2018.
- 2018 saw the lowest number of births for the six-year reporting period (approximately 58,000 births).
- There has been a steady increase in trimester two screens (both commenced and completed) since 2013.
- Māori screening completion rates have increased since 2013.
- The national screening completion rate has increased from 69 percent in 2013 to 74 percent of births being screened in 2018. First trimester screens made up 86 percent of all completed screens in 2018.
- Most district health boards (DHBs) showed a trend of increasing rates of screening commencement and completion.
- Ten percent of screens commenced in 2018 were not completed and nearly all related to screens commenced in the first trimester.
- The overall positive test rate (number of increased risk results per 100 screens) for trisomy 21, 18 and 13 was 4.1 in 2018, higher than 2016 (2.7).
- The positive test rate was higher for second trimester screens (6.3 per 100 screens) than for first trimester screens (3.7 per 100 screens) for 2018.
- The overall false positive rate for trisomy 21, 18 and 13 was 4 percent in 2018, higher than previous years (2–3%). The rate was higher for second trimester screens (6%) than for first trimester screens (3%).
- The overall detection rate for trisomy 21, 18 and 13 was 78 percent in 2018, compared to 79 percent in 2016.
- Over this reporting period several changes have occurred that may have impacted on the programme indicators, for example, nasal bone measurement has been excluded since March 2018.
- Despite the increasing availability of non-invasive prenatal screening (NIPS) to pregnant women in New Zealand, screening completion rates have increased over the reporting period, except for Auckland DHB.
- Changes to data linkage processes were implemented from 2017. Caution is required when comparing 2017 and 2018 data with previous years.

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 at the time. 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 and an ultrasound scan. The blood sample is collected between 9 weeks and 13 weeks 6 days gestation and measures two maternal serum markers: pregnancy-associated protein A (PAPP-A) and free beta-human chorionic gonadotropin (ßhCG). The ultrasound scan determines nuchal translucency (NT) and crown rump length (CRL) measurements and is performed between 11 weeks 2 days and 13 weeks 6 days gestation.
- Second trimester screening, which is a blood test taken between 14- and 20-weeks' gestation that measures four maternal serum markers: ßhCG, alpha-fetoprotein (AFP), unconjugated oestriol (uE3) and inhibin A.

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 DHB (for samples from Taupo and north of Taupo) and Canterbury Health Laboratories at Canterbury DHB (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

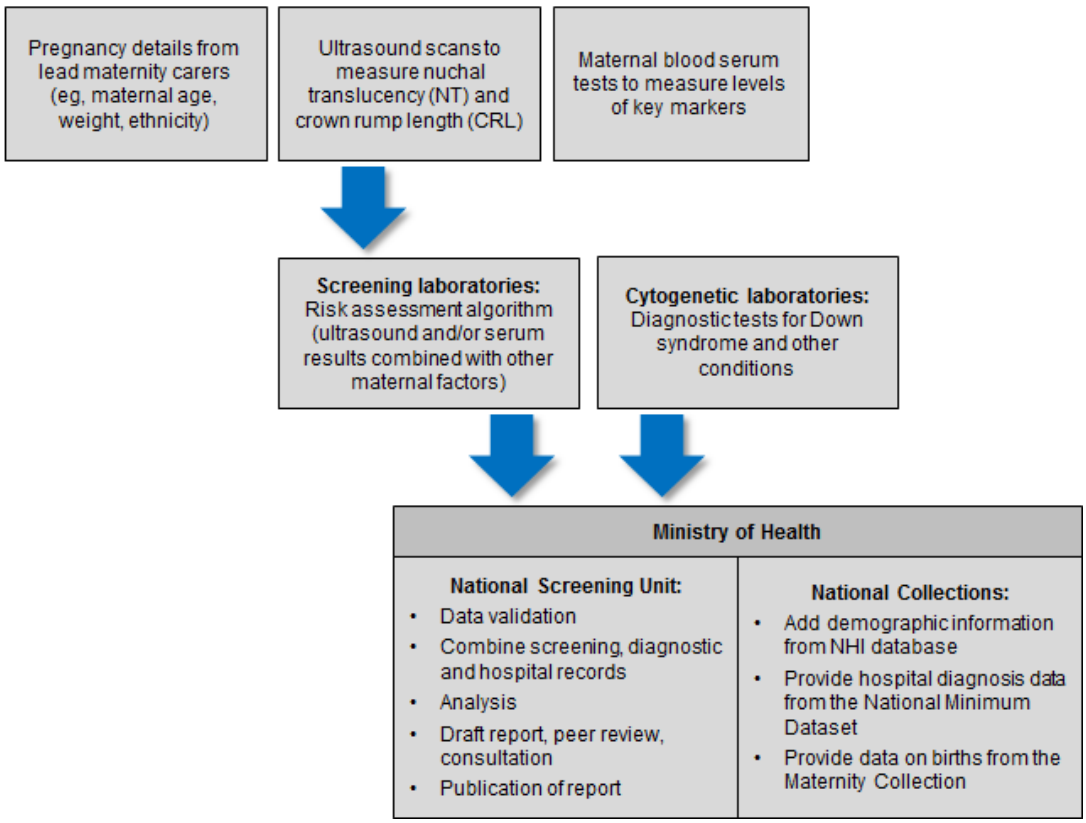
- neural tube defects.

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 that is produced by the laboratories.

Programme monitoring and data collection

This report presents monitoring results for antenatal screening for Down syndrome and other conditions for the period 1 January 2013 to 31 December 2018. 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.

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:

- IANZ accreditation assessment

- 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 2016, diagnostic testing data was received from all cytogenetic laboratories (LabPLUS, Waikato, Capital & Coast, and Canterbury Health Laboratories).

The screening and cytogenetic data was matched 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

Required components of each screening test

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

Demographic and maternal factors are also required (eg, date of birth, weight).

Commenced screening

At least one of the required components of the screening test was completed (NT measurement or serum analytes).

Completed screening

All the required components of each screening test were completed, and a risk result was reported.

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]

Inclusion criteria

Screens were included in this analysis if the following criteria were met.

- Screening commencement date between 1 January 2013 and 31 December 2018 (ie, date of the first test the woman had as part of the screening pathway).
- Valid National Health Index (NHI) identifier.
- Age at screen from 12 years to 49 years (date of birth as supplied by the requestor).
- 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 and non-Asian people.

[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.

NZ Deprivation

Due to issues with NZ Deprivation Index (NZ Dep), breakdown by deprivation has not been included in this report.

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 (numerator less than six) then those results have been suppressed as they are considered too unstable.

Prenatal cytogenetic test

The focus of indicators 6, 7, and 8 is on tests that women choose to have as part of managing their pregnancy. For these indicators prenatal tests are 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.

- Joining Births: Births are joined where they match the mothers NHI and are between 0- and 230-days post screen (approximately 33 weeks).
- Joining NMDS Outcomes: Outcomes are joined where they match the babies NHI.

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

- Joining Cytogenetics Data: Cytogenetics data is joined where 1: they are from the mother and between 0- and 105-days post screen (15 weeks), or 2: are from the baby and are between 0- and 230-days post screen.

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

A project reviewing the end-to-end data analysis process for the Down syndrome and other conditions report was started in 2018 and has resulted in changes to data linking rules. These changes have been applied to 2017 and 2018 data but not for years prior to this. Caution is therefore required when comparing data for 2013–2016 with 2017–2018. Where a five-year rate would ordinarily have been applied, a decision has been made to supply a two-year rate (2017 and 2018) where this does not compromise privacy.

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).

Incomplete data

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

- Thirteen women had no DHB of domicile or ethnicity information recorded in either the NHI database or in the laboratory information system. These women are included in the national total but not in DHB or ethnicity breakdowns.

Indicator 1: Screens commenced

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

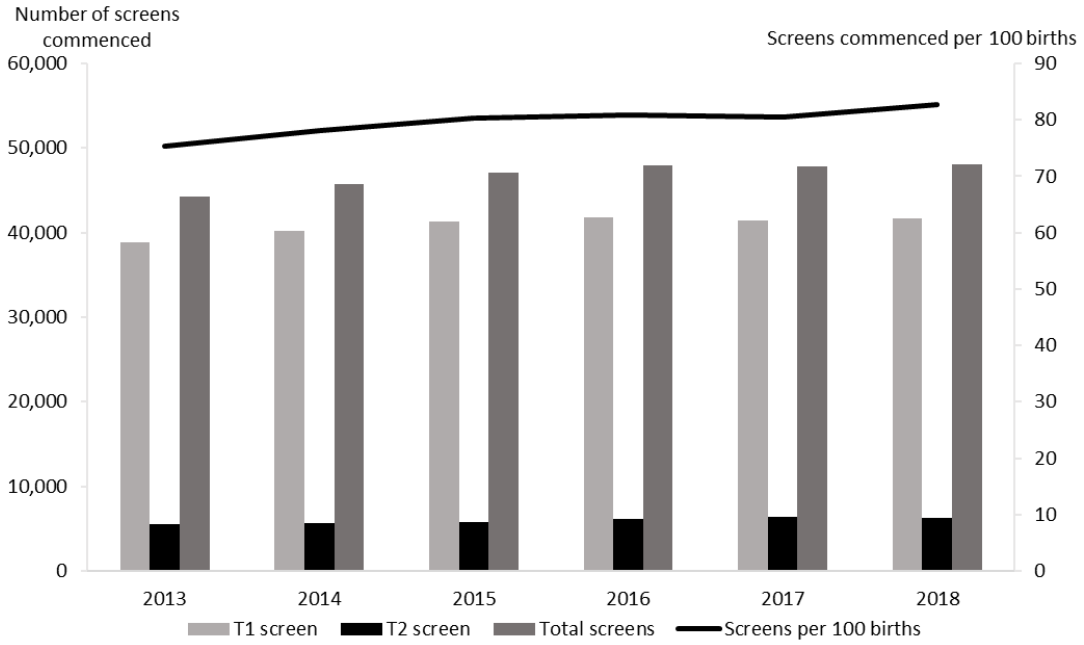
Total screens commenced by trimester

During 2018, a total of 48,011 screens were commenced, a rate of 83 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 rate of screens commenced per 100 births has increased over time from 75 in 2013 to 83 in 2018 (see Table 1 and Figure 2).

Table 1: Total screens commenced by trimester, January 2013 to December 2018

Trimester of screen	Number and rate of screens commenced					
	2013	2014	2015	2016	2017	2018
T1 screen	38,803	40,172	41,283	41,816	41,403	41,681
T2 screen	5,487	5,613	5,742	6,152	6,369	6,330
Total screens	**44,290**	**45,785**	**47,025**	**47,968**	**47,772**	**48,011**
Screens per 100 births	75.3	78.0	80.3	80.9	80.6	82.7

Figure 2: Count and rate of screens commenced, January 2013 to December 2018



Screens commenced by DHB

Figure 3 shows the screening commencement rates by DHB for 2018. There was a large variation in rates from 62 per 100 births in Northland to 95 per 100 births in South Canterbury. Two-thirds (65%) 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 to December 2018

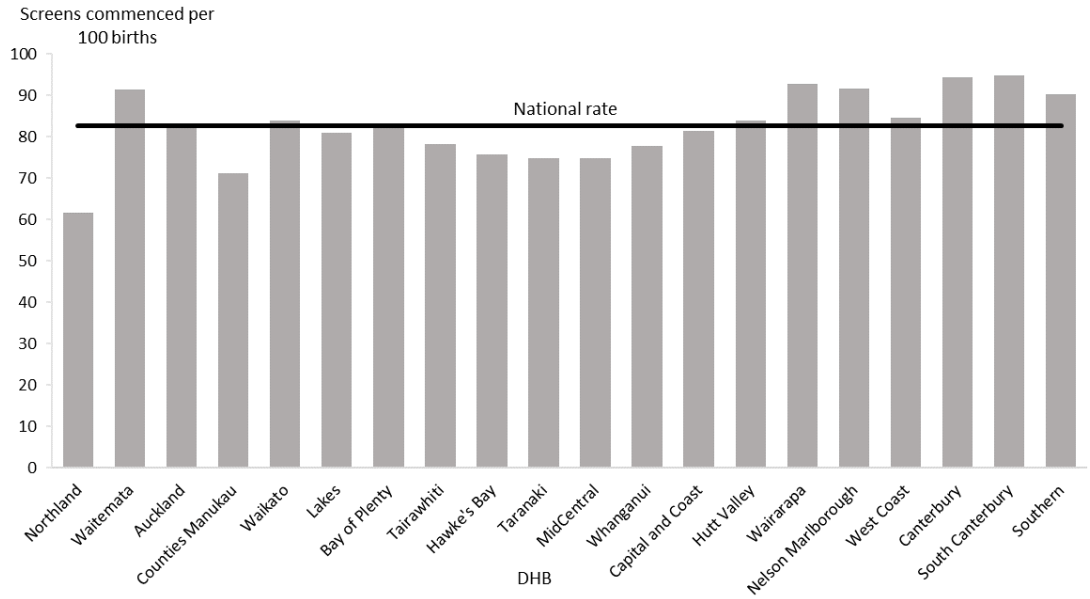


Table 2: Screens commenced by trimester and DHB, January to December 2018

DHB	Number of screens commenced			Screens commenced (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	1,156	197	1,353	52.7	9.0	61.7
Waitematā	6,007	781	6,788	80.9	10.5	91.4
Auckland	3,873	591	4,464	71.3	10.9	82.2
Counties Manukau	4,473	1,333	5,806	54.8	16.3	71.1
Waikato	4,036	484	4,520	75.0	9.0	84.0
Lakes	1,042	193	1,235	68.3	12.6	80.9
Bay of Plenty	2,270	227	2,497	75.4	7.5	82.9
Tairāwhiti	469	77	546	67.1	11.0	78.1
Hawke's Bay	1,426	168	1,594	67.6	8.0	75.6
Taranaki	874	293	1,167	56.0	18.8	74.7
MidCentral	1,471	144	1,615	68.0	6.7	74.7
Whanganui	534	95	629	66.1	11.8	77.8
Capital & Coast	2,337	271	2,608	73.0	8.5	81.4
Hutt Valley	1,408	219	1,627	72.7	11.3	84.0
Wairarapa	417	43	460	84.1	8.7	92.7
Nelson Marlborough	1,235	136	1,371	82.4	9.1	91.5
West Coast	239	36	275	73.5	11.1	84.6
Canterbury	5,230	671	5,901	83.6	10.7	94.3
South Canterbury	495	76	571	82.1	12.6	94.7
Southern	2,666	290	2,956	81.4	8.9	90.2
National	**41,681**	**6,330**	**48,011**	**71.8**	**10.9**	**82.7**

Note: DHB counts do not sum to National total.

Most DHBs showed an increase in their rate of screens commenced between 2013 and 2018 or had fairly stable rates (see Table 3).

Table 3: Screens commenced per 100 births by DHB, January 2013 to December 2018

DHB	Screens commenced (per 100 births)					
	2013	2014	2015	2016	2017	2018
Northland	52.9	55.6	60.1	58.6	64.2	61.7
Waitematā	86.3	86.3	88.4	87.1	86.7	91.4
Auckland	82.4	84.0	85.7	82.0	75.8	82.2
Counties Manukau	64.8	68.7	71.1	71.0	70.6	71.1
Waikato	76.4	80.4	81.8	83.7	85.5	84.0
Lakes	70.1	77.4	74.3	76.7	73.6	80.9
Bay of Plenty	69.6	72.4	77.6	81.1	82.2	82.9
Tairāwhiti	53.2	59.3	68.3	63.6	70.2	78.1
Hawke's Bay	64.6	66.0	72.6	76.2	71.8	75.6
Taranaki	61.4	68.2	74.9	67.8	72.7	74.7
MidCentral	58.3	59.3	63.9	73.1	79.9	74.7
Whanganui	47.9	61.0	70.5	74.1	71.8	77.8
Capital & Coast	78.1	80.3	83.4	86.3	76.1	81.4
Hutt Valley	72.7	78.6	78.7	82.2	76.3	84.0
Wairarapa	76.6	81.6	83.8	89.0	90.1	92.7
Nelson Marlborough	87.4	97.6	96.0	85.1	98.6	91.5
West Coast	81.1	88.3	82.4	86.5	84.4	84.6
Canterbury	90.3	89.5	89.4	91.5	92.4	94.3
South Canterbury	88.1	78.8	86.4	87.5	94.0	94.7
Southern	81.4	83.3	85.1	87.8	89.0	90.2
National average	**75.3**	**78.0**	**80.3**	**80.9**	**80.6**	**82.7**

Screens commenced by age and ethnicity

Table 4 provides an overall view of screens commenced by age and ethnicity for January 2013 to December 2018. During this reporting period the overall rate of screens commenced has increased and though variation between age and ethnicity is still evident these differences have become less marked.

Table 4: Screens commenced by age and ethnicity of mother, January 2013 to December 2018

	Number of screens commenced						Screens commenced (per 100 births)					
	2013	**2014**	**2015**	**2016**	**2017**	**2018**	**2013**	**2014**	**2015**	**2016**	**2017**	**2018**
Age at screen (years)												
Under 20	1,947	1,990	1,925	1,829	1,683	1,546	58.5	66.6	69.1	74.9	73.3	72.7
20–24	6,932	7,055	7,109	7,000	6,899	6,475	64.2	68.7	71.5	73.0	74.0	74.5
25–29	12,022	12,800	13,189	13,943	14,037	14,162	78.8	81.5	84.0	84.3	84.4	87.1
30–34	13,914	14,623	15,124	15,732	15,804	16,171	83.0	83.2	84.5	85.6	84.5	86.4
35–39	7,628	7,610	8,007	7,781	7,659	8,091	76.0	78.6	82.0	78.1	77.5	80.8
40–44	1,767	1,626	1,593	1,574	1,587	1,476	72.6	69.3	69.3	69.2	68.6	70.5
45 and over	80	81	78	109	103	90	55.9	61.4	56.1	86.5	67.8	55.6
Ethnicity												
Māori	5,805	6,284	6,256	7,176	7,754	7,675	39.6	43.9	42.9	48.7	52.0	52.7
Pacific	2,999	3,005	3,120	3,089	3,284	3,206	47.2	48.7	51.5	52.9	55.0	53.7
Asian	7,474	4,835	8,695	9,851	9,720	10,330	91.7	91.8	94.4	93.6	92.0	97.5
Other	28,012	28,058	28,954	27,852	27,005	26,796	94.5	96.6	100.9	98.7	97.0	99.5
National	**44,290**	**45,785**	**47,025**	**47,968**	**47,772**	**48,011**	**75.3**	**78.0**	**80.3**	**80.9**	**80.6**	**82.7**

Note: Ethnic group counts do not sum to National total.

The 25–29 and 30–34 age groups had the highest rate of screens commenced for 2018 with 87 and 86 women commencing screening per 100 births respectively. From 2013 to 2018 rates have increased overall for most age groups, particularly the younger age groups. The only age groups that did not increase were 40–44 years and 45 years and over.

Figure 4: Screens commenced by age of mother at screen, January to December 2018

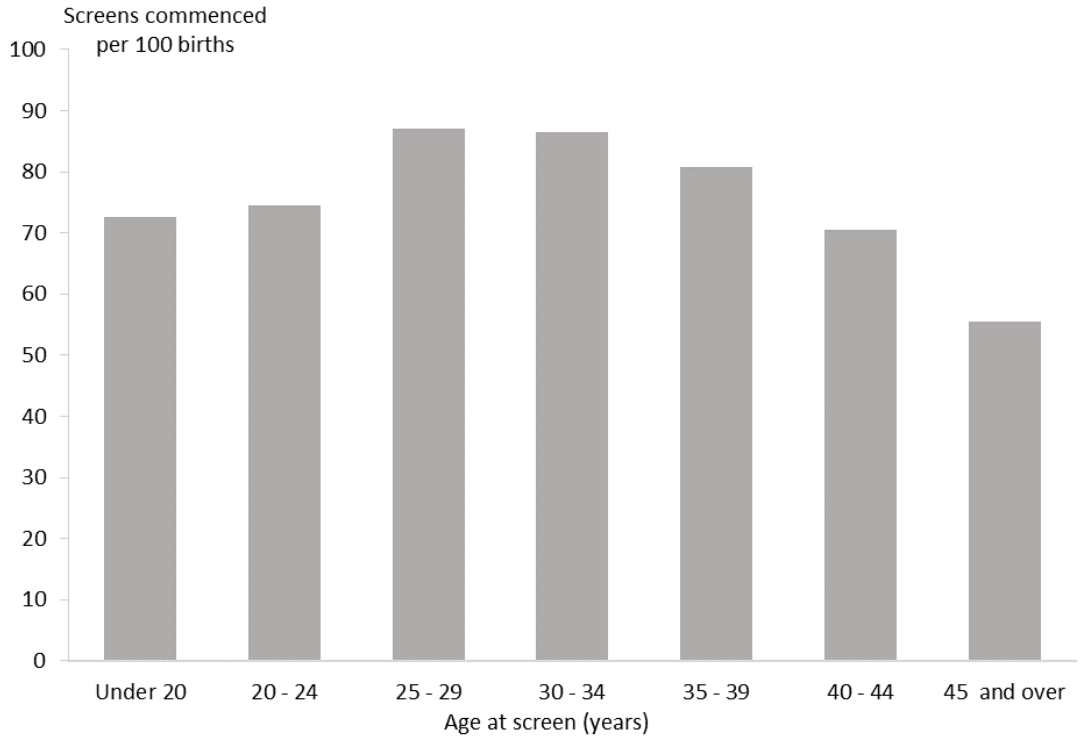
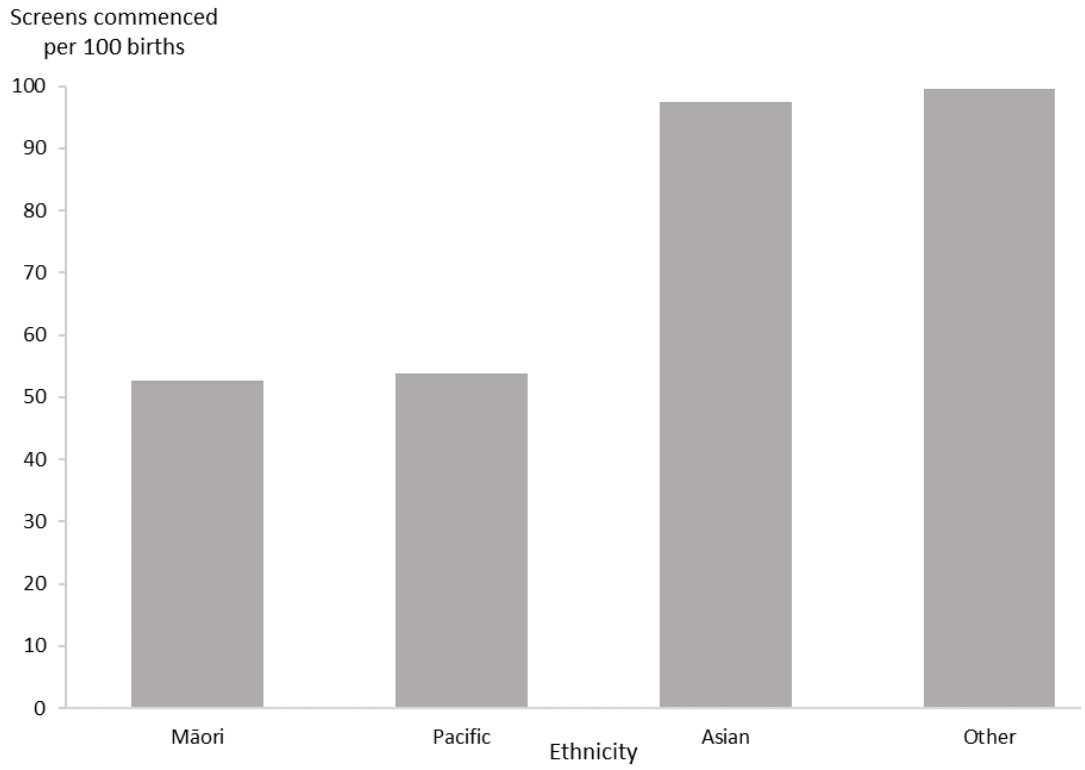


Figure 5: Screens commenced by ethnicity of mother, January to December 2018



Differences in screening commencement rates by ethnicity remained consistent for 2018. Women of Other ethnicity had the highest rate (100 of 100 births) followed by Asian women (98 of 100 births). The rate of commenced screens for Pacific and Māori women was lower at 54 per 100 births and 53 per 100 births respectively (see Figure 5). All groups have shown increasing rates over the six years, particularly for Māori with an absolute increase of 13 percentage points from 40 percent in 2013 to 53 percent in 2018 (see Table 4). This rate is however well below the national rate of 83 per 100 births in 2018.

Indicator 2: Screens completed

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

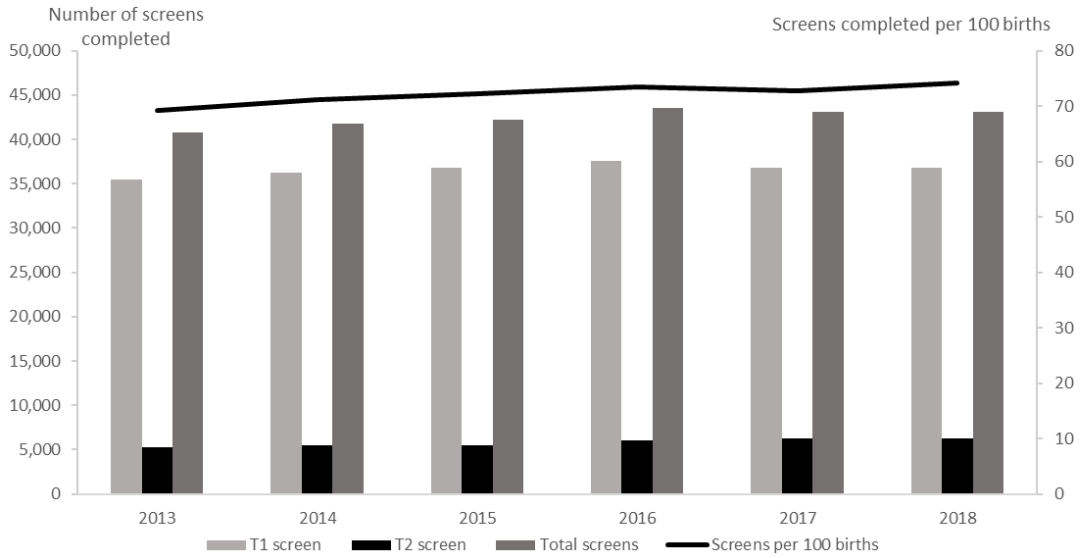
Total screens completed by trimester

During 2018, a total of 43,052 screens were completed, a rate of 74 screens per 100 births. Table 5 and Figure 6 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 since 2013 (from 69 to 74 in 2018).

Table 5: Total screens completed by trimester, January 2013 to December 2018

Trimester of screen	Number and rate of screens completed					
	2013	2014	2015	2016	2017	2018
T1 screen	35,464	36,280	36,739	37,511	36,836	36,810
T2 screen	5,269	5,456	5,517	6,008	6,284	6,242
Total screens	**40,733**	**41,736**	**42,256**	**43,519**	**43,120**	**43,052**
Screens per 100 births	69.3	71.1	72.2	73.4	72.7	74.2

Figure 6: Count and rate of screens completed, January 2013 to December 2018



Screens completed by DHB

Screening completion rates for 2018 varied across DHBs from 54 per 100 births in Northland to 88 per 100 births in South Canterbury (see Figure 7). Table 6 gives a full breakdown by the trimester of screen.

Figure 7: Screens completed by DHB, January to December 2018

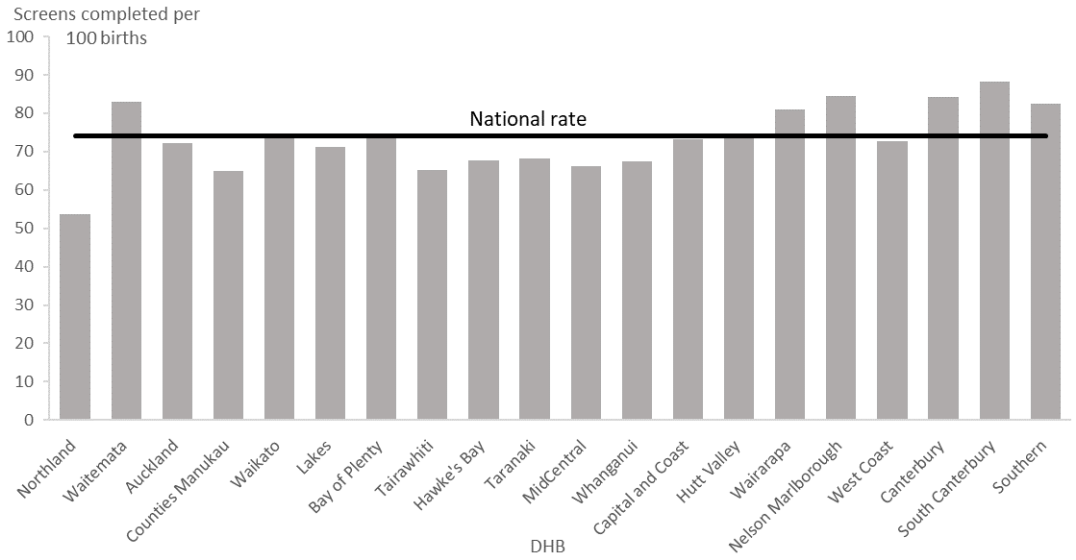


Table 6: Screening completion by trimester and DHB, January to December 2018

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	980	195	1,175	44.7	8.9	53.6
Waitematā	5,384	769	6,153	72.5	10.4	82.9
Auckland	3,335	585	3,920	61.4	10.8	72.2
Counties Manukau	3,986	1,311	5,297	48.8	16.1	64.9
Waikato	3,548	479	4,027	65.9	8.9	74.8
Lakes	896	190	1,086	58.7	12.5	71.2
Bay of Plenty	2,020	226	2,246	67.1	7.5	74.6
Tairāwhiti	382	73	455	54.6	10.4	65.1
Hawke's Bay	1,260	166	1,426	59.8	7.9	67.6
Taranaki	777	290	1,067	49.7	18.6	68.3
MidCentral	1,292	141	1,433	59.8	6.5	66.3
Whanganui	452	94	546	55.9	11.6	67.6
Capital & Coast	2,084	264	2,348	65.1	8.2	73.3
Hutt Valley	1,224	217	1,441	63.2	11.2	74.4
Wairarapa	359	43	402	72.4	8.7	81.0
Nelson Marlborough	1,133	134	1,267	75.6	8.9	84.6
West Coast	200	36	236	61.5	11.1	72.6
Canterbury	4,608	661	5,269	73.6	10.6	84.2
South Canterbury	456	76	532	75.6	12.6	88.2
Southern	2,415	287	2,702	73.7	8.8	82.5
National	**36,810**	**6,242**	**43,052**	**63.4**	**10.8**	**74.2**

Note: DHB counts do not sum to National total.

Table 7: Screening completion by DHB, January 2013 to December 2018

DHB	Screens completed (per 100 births)					
	2013	2014	2015	2016	2017	2018
Northland	47.1	48.0	51.6	50.9	56.2	53.6
Waitematā	82.1	81.0	81.8	81.4	79.8	82.9
Auckland	77.7	78.8	79.1	75.6	68.6	72.2
Counties Manukau	59.6	63.2	64.5	65.5	64.4	64.9
Waikato	69.2	72.5	72.4	74.6	76.3	74.8
Lakes	62.6	69.9	65.7	67.8	65.7	71.2
Bay of Plenty	62.0	64.5	67.8	71.8	73.6	74.6
Tairāwhiti	47.1	51.5	53.8	51.1	59.1	65.1
Hawke's Bay	59.9	59.4	64.2	68.6	63.7	67.6
Taranaki	55.1	61.2	66.3	62.1	66.4	68.3
MidCentral	53.8	54.0	56.9	66.1	72.3	66.3
Whanganui	45.0	53.1	58.5	65.8	63.6	67.6
Capital & Coast	70.9	72.6	75.1	77.8	67.8	73.3
Hutt Valley	64.7	68.9	68.0	71.6	67.3	74.4
Wairarapa	66.7	70.6	72.8	77.9	80.6	81.0
Nelson Marlborough	78.1	87.6	84.7	77.4	90.1	84.6
West Coast	72.3	78.9	72.3	77.7	76.8	72.6
Canterbury	81.9	81.2	80.6	82.5	83.0	84.2
South Canterbury	85.6	75.3	79.8	81.5	85.4	88.2
Southern	75.5	74.8	77.9	81.1	81.7	82.5
National average	**69.3**	**71.1**	**72.2**	**73.4**	**72.7**	**74.2**

Similar to screens commenced, most DHBs showed a trend of increasing rates of screening completion over the six years covered in this report, with the exception of Auckland DHB, which showed a decreasing trend from 2016 to 2018 (the rate for 2013 was 77.7 compared with 72.2 per 100 births in 2018).

Screens completed by age and ethnicity

Table 8 provides an overall view of screens completed by age and ethnicity for January 2013 to December 2018, with similar trends to screening commencement. Screening completion rates were highest in the 25–29 years age group with 79 women completing screening per 100 births in 2018.

Screening completion rates were highest among women of Asian ethnicity at 91 per 100 births for 2018. This was followed closely by women of Other ethnicity at 90 per 100 births. The rate of completed screens for Pacific and Māori women remains lower at 47 per 100 births and 44 per 100 births respectively (see Figure 9).

Table 8: Screens completed by age and ethnicity of mother, January 2013 to December 2018

	Number of screens completed						Screens completed (per 100 births)					
	2013	2014	2015	2016	2017	2018	2013	2014	2015	2016	2017	2018
Age at screen (years)												
Under 20	1,610	1,604	1,510	1,474	1,376	1,243	48.4	53.6	54.2	60.3	59.9	58.4
20–24	6,010	6,070	5,992	6,079	5,948	5,588	55.6	59.1	60.3	63.4	63.8	64.3
25–29	11,097	11,685	11,824	12,675	12,779	12,898	72.7	74.4	75.3	76.6	76.9	79.4
30–34	13,089	13,675	14,030	14,709	14,651	14,823	78.0	77.8	78.3	80.1	78.4	79.2
35–39	7,214	7,144	7,430	7,137	6,959	7,205	71.9	73.9	76.1	71.6	70.4	71.9
40–44	1,643	1,486	1,406	1,366	1,328	1,225	67.5	63.3	61.2	60.0	57.4	58.5
45 and over	70	72	64	79	79	70	49.0	54.5	46.0	62.7	52.0	43.2
Ethnicity												
Māori	4,893	5,178	4,911	5,924	6,442	6,387	33.4	36.2	33.7	40.2	43.2	43.8
Pacific	2,606	2,598	2,626	2,673	2,876	2,782	41.0	42.1	43.3	45.8	48.2	46.6
Asian	7,091	8,034	8,114	9,304	9,093	9,594	87.0	87.4	88.1	88.4	86.1	90.6
Other	26,143	25,926	26,605	25,618	24,701	24,287	88.2	89.2	92.7	90.8	88.7	90.2
National	**40,733**	**41,736**	**42,256**	**43,519**	**43,120**	**43,052**	**69.3**	**71.1**	**72.2**	**73.4**	**72.7**	**74.2**

Note: Ethnic group counts do not sum to National total.

As seen in screening commencement rates, most groups showed an overall increase in completion rates over the six-year period, with the biggest increases seen in Māori and Pacific ethnicities and younger women. The only groups to show a decrease in screening completion rates were women aged 40–44 years and 45 years and over.

Figure 8: Screens completed by age of mother at screen, January to December 2018

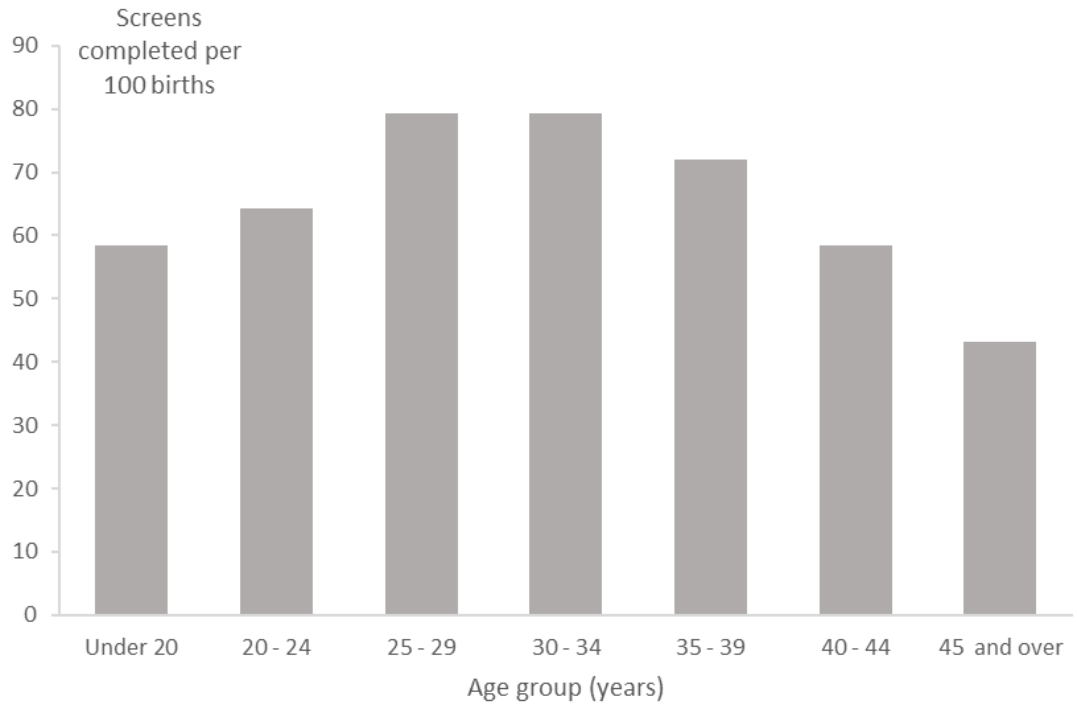
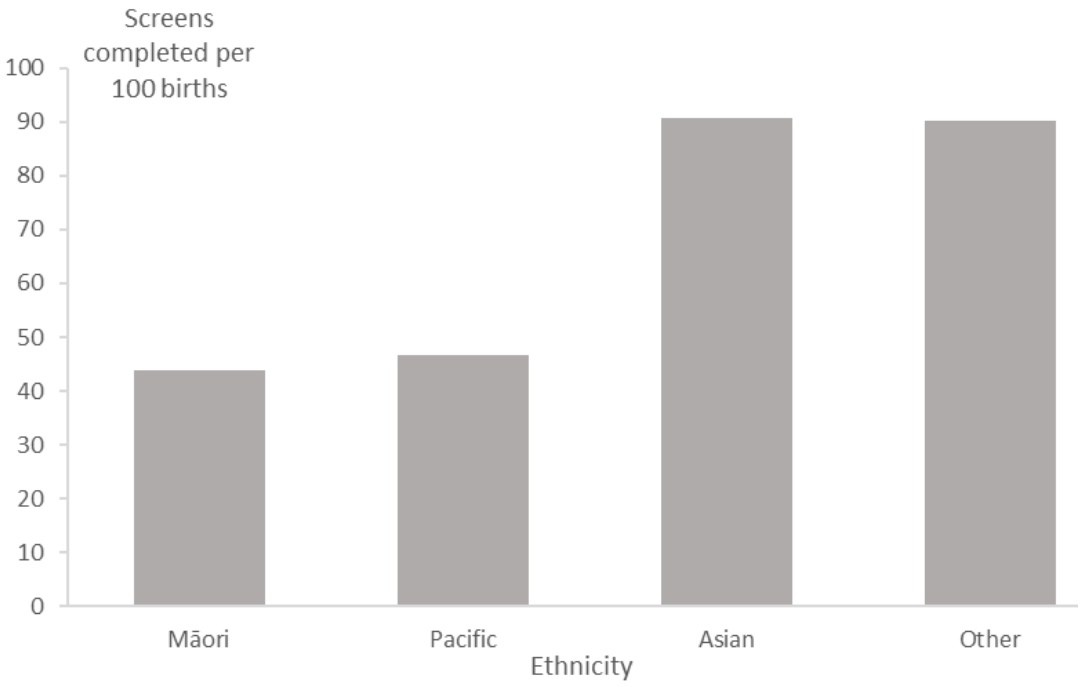


Figure 9: Screens completed by ethnicity of mother, January to December 2018



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 2013 to 2018. 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 (41 percent in 2018). PAPP-A was included in 12 percent of second trimester screens in 2018, higher than previous years.

Table 9: Screening pathway variance by type, January 2013 to December 2018

Year	Second trimester screening results				
		Number		Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
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
2017	6,284	2,561	656	40.8	10.4
2018	6,242	2,563	735	41.1	11.8

Screening pathway variance by DHB

Table 10 shows a breakdown of screening pathway variance by DHB and type of variance for the 2018 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 to December 2018

DHB	Second trimester screening results				
	Number			Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
Northland	195	66	22	33.8	11.3
Waitematā	769	316	107	41.1	13.9
Auckland	585	199	101	34.0	17.3
Counties Manukau	1,311	369	157	28.1	12.0
Waikato	479	249	33	52.0	6.9
Lakes	190	70	18	36.8	9.5
Bay of Plenty	226	111	15	49.1	6.6
Tairāwhiti	73	27	9	37.0	12.3
Hawke's Bay	166	62	24	37.3	14.5
Taranaki	290	57	45	19.7	15.5
MidCentral	141	78	12	55.3	8.5
Whanganui	94	43	9	45.7	9.6
Capital & Coast	264	129	16	48.9	6.1
Hutt Valley	217	107	22	49.3	10.1
Wairarapa	43	29	S	67.4	S
Nelson Marlborough	134	78	13	58.2	9.7
West Coast	36	19	S	52.8	S
Canterbury	661	351	93	53.1	14.1
South Canterbury	76	29	17	38.2	22.4
Southern	287	172	17	59.9	5.9
National	**6,242**	**2,563**	**735**	**41.1**	**11.8**

Note: DHB counts do not sum to National total.

(S) Suppressed if the number of screens was < 6.

Screening pathway variance by age and ethnicity

Table 11 shows a breakdown of screening pathway variance by age and ethnicity for the 2018 year. The results show higher proportions for pathway variance for women in the 20–24 and 30–34 age groups (43%) and women of Other ethnicity (51%).

Table 11: Screening pathway variance by age and ethnicity, January to December 2018

	Second trimester screening results				
		Number		Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
Age at screen (years)					
Under 20	389	144	27	37.0	6.9
20–24	1,269	548	104	43.2	8.2
25–29	1,867	761	226	40.8	12.1
30–34	1,675	719	221	42.9	13.2
35–39	853	328	133	38.5	15.6
40–44	177	59	24	33.3	13.6
45 and over	12	4	0	33.3	0
Ethnicity					
Māori	1,551	613	129	39.5	8.3
Pacific	1,071	299	104	27.9	9.7
Asian	1,364	492	202	36.1	14.8
Other	2,256	1,159	300	51.4	13.3
National	**6,242**	**2,563**	**735**	**41.1**	**11.8**

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 the 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 2018 was 4,959, which equates to 10 percent of screens commenced that year.

Table 12: Incomplete screens by trimester, January 2013 to December 2018

Trimester of screen	Number of incomplete screens					
	2013	2014	2015	2016	2017	2018
T1 screens	3,339	3,892	4,544	4,305	4,567	4,871
T2 screens	218	157	225	144	85	88
Total screens	**3,557**	**4,049**	**4,769**	**4,449**	**4,652**	**4,959**

Incomplete T1 screens by reason incomplete

Table 13 shows provides a breakdown of incomplete T1 screens according to which component of the screen was missing. Results have been reported as a percentage of all commenced screens, and then as a percentage of all incomplete screens.

The proportion of incomplete T1 screens out of all commenced T1 screens in 2018 was 12 percent. 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 2013 to December 2018

Year	Commenced first trimester			Reason incomplete			Incomplete as percentage of commenced			Type as percentage of all incomplete T1 screens	
	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
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
2017	4,567	36,836	41,403	3,275	1,286	12	7.9	3.1	11.0	71.7	28.2
2018	4,871	36,810	41,681	3,530	1,334	13	8.5	3.2	11.7	72.5	27.4

Incomplete T1 screens by reason and DHB

Table 14 provides the same breakdown by DHB for the 2018 year. The lower numbers involved limit DHB comparisons. The range in the percentage of screens incomplete due to no blood sample was from 45 percent (at Taranaki) to 90 percent (at South Canterbury).

Table 14: Incomplete T1 screens by reason and DHB, January to December 2018

DHB	Commenced first trimester			Reason incomplete			Incomplete as percentage of commenced			Type as percentage of all incomplete T1 screens	
	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
Northland	176	980	1,156	132	44	S	11.4	3.8	15.2	75.0	25.0
Waitematā	623	5,384	6,007	443	180	S	7.4	3.0	10.4	71.1	28.9
Auckland	538	3,335	3,873	409	129	S	10.6	3.3	13.9	76.0	24.0
Counties Manukau	487	3,986	4,473	334	153	S	7.5	3.4	10.9	68.6	31.4
Waikato	488	3,548	4,036	374	113	S	9.3	2.8	12.1	76.6	23.2
Lakes	146	896	1,042	107	39	S	10.3	3.7	14.0	73.3	26.7
Bay of Plenty	250	2,020	2,270	182	68	S	8.0	3.0	11.0	72.8	27.2
Tairāwhiti	87	382	469	66	21	S	14.1	4.5	18.6	75.9	24.1
Hawke's Bay	166	1,260	1,426	106	60	S	7.4	4.2	11.6	63.9	36.1
Taranaki	97	777	874	44	53	S	5.0	6.1	11.1	45.4	54.6
MidCentral	179	1,292	1,471	135	41	S	9.2	2.8	12.2	75.4	22.9
Whanganui	82	452	534	61	21	S	11.4	3.9	15.4	74.4	25.6
Capital & Coast	253	2,084	2,337	184	67	S	7.9	2.9	10.8	72.7	26.5
Hutt Valley	184	1,224	1,408	134	50	S	9.5	3.6	13.1	72.8	27.2
Wairarapa	58	359	417	47	11	S	11.3	2.6	13.9	81.0	19.0
Nelson Marlborough	102	1,133	1,235	72	29	S	5.8	2.3	8.3	70.6	28.4
West Coast	39	200	239	29	10	S	12.1	4.2	16.3	74.4	25.6
Canterbury	622	4,608	5,230	466	156	S	8.9	3.0	11.9	74.9	25.1
South Canterbury	39	456	495	35	S	S	7.1	S	7.9	89.7	S
Southern	251	2,415	2,666	168	83	S	6.3	3.1	9.4	66.9	33.1
National	**4,871**	**36,810**	**41,681**	**3,530**	**1,334**	**13**	**8.5**	**3.2**	**11.7**	**72.5**	**27.4**

(S) Suppressed if the number of screens was < 6.

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 2018, 1 percent of T2 commenced screens were incomplete, compared with 12 percent of T1 commenced screens. As Table 15 shows, the percentage of incomplete T2 screens decreased from 4 percent in 2013 to 1 percent in 2018.

Table 15: Incomplete T2 screens, January 2013 to December 2018

Year	Commenced second trimester	No result issued	Percentage incomplete
2013	5,487	218	4.0
2014	5,613	157	2.8
2015	5,742	225	3.9
2016	6,152	144	2.3
2017	6,369	85	1.3
2018	6,330	88	1.4
Total	**35,693**	**917**	**Ave: 2.6**

Incomplete T2 screens by DHB

Table 16 shows a breakdown of incomplete T2 screens by DHB for the 2018 year. The very low numbers involved limit meaningful DHB comparisons.

Table 16: Incomplete T2 screens by DHB, January to December 2018

DHB	Commenced second trimester	No result issued	% incomplete
Northland	197	S	S
Waitematā	781	12	1.5
Auckland	591	6	1.0
Counties Manukau	1,333	22	1.7
Waikato	484	S	S
Lakes	193	S	S
Bay of Plenty	227	S	S
Tairāwhiti	77	S	S
Hawke's Bay	168	S	S
Taranaki	293	S	S
MidCentral	144	S	S
Whanganui	95	S	S
Capital & Coast	271	7	2.6
Hutt Valley	219	S	S
Wairarapa	43	S	S
Nelson Marlborough	136	S	S
West Coast	36	S	S
Canterbury	671	10	1.5
South Canterbury	76	S	S
Southern	290	S	S
National	**6,330**	**88**	**1.4**

Note: DHB counts do not sum to National total.

(S) Suppressed if the number of screens was < 6.

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 17 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 2018 year, 4.1 increased risk results were issued for every 100 screens completed, which is higher than previous years.

Table 17: Number and rate per 100 screens of increased risk screening results for trisomy 21, 18 or 13, January 2013 to December 2018

	Number and rate of increased risk screens					
	2013	2014	2015	2016	2017	2018
Total increased risk results	1,111	1,162	1,168	1,189	1,318	1,764
Positive test rate per 100 completed screens	2.7	2.8	2.8	2.7	3.1	4.1

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

Table 18 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 and ethnicity for the 2018 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.

Table 18: Increased risk screening results for trisomy 21, 18 or 13 by age and ethnicity, January to December 2018

	Number of screens that include an increased risk for trisomy 21, 18 or 13	Total no. of completed screens	Positive test rate per 100 screens
Age at screen (years)			
Under 20	14	1,243	1.1
20–24	74	5,588	1.3
25–29	208	12,898	1.6
30–34	453	14,823	3.1
35–39	648	7,205	9.0
40–44	345	1,225	28.2
45 and over	22	70	31.4
Ethnicity			
Māori	209	6,387	3.3
Pacific	151	2,782	5.4
Asian	486	9,594	5.1
Other	918	24,287	3.8
National	**1,764**	**43,052**	**4.1**

Note: Ethnic group counts do not sum to National total (for total number of completed screens).

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

Table 19 shows the positive test rate for each of trisomy 21, 18 and 13 individually as well as the positive test rate for the three trisomies together by trimester of screen and calendar year. The sum of the individual values for trisomy 21, 18 and 13 is greater than the value for the fourth grouping (any of the three trisomies) because a result can be at increased risk for more than one trisomy.

Trisomy 18 and 13 each had low positivity rates (0.4 per 100 screens) while the positive test rate for trisomy 21 has increased to 4 per 100 screens. The second trimester positive test rate for trisomy 21 was higher than the first trimester positive test rate (6.1 and 3.7 respectively). This difference was the same in 2014 (4.8 and 2.4) but has generally been a smaller difference in previous years. The difference in rates may be due to variability in nuchal translucency and crown rump length assessments and the removal of nasal bone from the risk calculation algorithm. The positive test rate for any one or more of trisomy 21, 18 or 13 was similar to that of trisomy 21 alone. This reflects the far higher number of increased risk screening results for trisomy 21 compared with trisomy 18 and 13.

Table 19: Increased risk screening results for trisomy 21, 18 and 13 by trimester of screen, January 2013 to December 2018

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						
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
2017	1,287	3.0	1,033	2.8	254	4.0
2018	1,740	4.0	1,361	3.7	379	6.1
Trisomy 18						
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
2017	140	0.3	123	0.3	17	0.3
2018	161	0.4	143	0.4	18	0.3
Trisomy 13						
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
2017	161	0.4	143	0.4	18	0.3
2018	167	0.4	155	0.4	12	0.2
Any one or more of trisomy 21, 18 or 13						
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
2017	1,318	3.1	1,046	2.8	272	4.3
2018	1,764	4.1	1,373	3.7	391	6.3

Increased risk screening results stratified by risk level

Table 20 shows the number of increased risk results stratified by risk level for each of trisomy 21, 18 and 13 for the 2018 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 20 will be greater than the total number of increased risk results for 2018.

Table 20: Increased risk screening results for trisomy 21, 18 and 13 by risk level, January to December 2018

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 to 1:20	245	50	62
>1:20 to 1:50	187	27	27
>1:50 to 1:300	1,308	84	78

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 21 shows the diagnostic testing rate from 2013 to 2018 by trimester of screen. In 2018, for every 100 women that received an increased risk result after a first or second trimester screen, 38 women had a diagnostic test. There has been a downward trend since 2013, although 2018 has seen a slight increase in the number of women having a diagnostic test compared to 2017. The diagnostic testing rate was lower for women who received an increased risk after a second trimester screen (35 women per 100 increased risk screens) compared with first trimester screens (38 per 100 increased risk screens). 2018 sees the smallest difference between the first and second trimester diagnostic testing rate and this has steadily dropped since 2013. See Appendix 3 for a summary of diagnostic test results for women who had an increased risk screen in 2018.

Table 21: Diagnostic testing volumes for women with increased risk screens by trimester of screen, January 2013 to December 2018

Trimester of screen	Diagnostic tests per 100 increased risk screens					
	2013	2014	2015	2016	2017	2018
T1 screen	66.0	62.5	59.0	46.9	36.7	38.4
T2 screen	46.9	47.4	44.3	40.5	29.0	35.3
Total screens	**61.6**	**59.0**	**56.3**	**45.7**	**35.1**	**37.7**

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

Table 22: Diagnostic testing volumes for women with increased risk screens by DHB, January 2013 to December 2018

DHB	Number of diagnostic tests						Diagnostic tests per 100 increased risk screens					
	2013	2014	2015	2016	2017	2018	2013	2014	2015	2016	2017	2018
Northland	28	26	21	12	12	18	56.0	59.1	48.8	40.0	34.3	38.3
Waitematā	141	116	107	82	78	102	72.7	61.7	57.5	44.6	37.5	37.2
Auckland	89	89	76	72	49	63	67.4	55.3	53.5	45.0	30.6	31.8
Counties Manukau	73	76	86	78	55	99	47.1	50.3	53.8	54.9	31.4	39.6
Waikato	41	41	42	45	29	56	57.7	64.1	60.0	52.9	30.2	39.4
Lakes	21	21	28	16	14	19	67.7	53.8	71.8	59.3	46.7	46.3
Bay of Plenty	21	21	20	17	18	26	53.8	63.6	66.7	44.7	40.0	39.4
Tairāwhiti	S	S	S	S	S	7	S	S	S	S	S	43.8
Hawke's Bay	21	20	15	8	7	15	53.8	58.8	51.7	28.6	26.9	33.3
Taranaki	18	12	10	8	S	10	66.7	48.0	43.5	36.4	S	35.7
MidCentral	10	11	8	15	20	19	38.5	57.9	44.4	46.9	50.0	52.8
Whanganui	6	S	S	6	S	9	46.2	S	S	66.7	S	52.9
Capital & Coast	55	46	65	41	30	34	74.3	59.7	60.7	60.3	32.6	37.0
Hutt Valley	18	15	18	15	15	18	58.1	53.6	64.3	45.5	45.5	34.0
Wairarapa	9	S	S	S	S	S	81.8	S	S	S	S	S
Nelson Marlborough	17	19	15	14	13	20	89.5	79.2	57.7	51.9	48.1	44.4
West Coast	S	8	S	6	S	S	S	42.1	S	85.7	S	S
Canterbury	74	122	83	80	70	95	60.2	65.6	50.6	36.7	32.0	34.2
South Canterbury	S	S	9	S	7	S	S	S	75.0	S	36.8	S
Southern	34	33	40	20	31	44	64.2	67.3	60.6	37.0	44.9	48.4
National	**684**	**685**	**657**	**543**	**463**	**665**	**61.6**	**59.0**	**56.3**	**45.7**	**35.1**	**37.7**

(S) Suppressed if the number of diagnostic tests was < 6.

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

Table 23 shows the diagnostic testing rate for women with increased risk screens by age and ethnicity for 2013 to 2018.

For 2018, diagnostic testing rates were highest for women of Other ethnicity (39 per 100 increased risk screens), followed by Asian and Māori women (38 and 37 per 100 increased risk screens respectively), and then Pacific women (33 per 100 increased risk screens). From age 25 years, diagnostic testing rates reduce with each age grouping.

Table 23: Diagnostic testing volumes for women with increased risk screens by age and ethnicity, January 2013 to December 2018

	Diagnostic tests per 100 increased risk screens					
	2013	**2014**	**2015**	**2016**	**2017**	**2018**
Age at screen (years)						
Under 20	28.6	50.0	53.8	45.5	17.4	28.6
20–24	62.5	53.9	51.7	55.6	43.5	50.0
25–29	60.5	62.7	58.1	49.4	38.2	44.7
30–34	68.1	64.9	61.8	47.7	38.8	41.3
35–39	62.6	57.1	57.0	46.0	32.9	35.3
40–44	57.4	58.1	50.9	39.0	29.8	32.5
45 and over	44.4	36.0	41.2	27.8	35.3	13.6
Ethnicity						
Māori	52.5	38.4	45.1	46.7	30.1	37.3
Pacific	38.2	39.2	36.2	34.3	31.0	33.1
Asian	69.2	67.0	63.3	56.3	37.7	37.9
Other	65.2	62.8	58.7	42.1	35.9	38.5
National	**61.6**	**59.0**	**56.3**	**45.7**	**35.1**	**37.7**

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 24 shows the number of diagnostic tests for women that received an increased risk result during 2018 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 30 to 70 tests per 100 women with an increased risk.

Table 24: Diagnostic testing volumes for women with increased risk screens by risk level, January to December 2018

Risk level	Number of diagnostic tests	Number of increased risk screens	Tests per 100 increased risk screens
1:5 to 1:20	178	255	69.8
>1:20 to 1:50	96	185	51.9
>1:50 to 1:300	391	1,324	29.5

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 screening result 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, for example, 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 0.79 per 100 low risk screens in 2018, which was the highest rate for the reporting period.

Table 25: Diagnostic testing volumes for women with low risk screens by trimester of screen, January 2013 to December 2018

Trimester of screen	Diagnostic tests per 100 low risk screens					
	2013	2014	2015	2016	2017	2018
T1 screen	0.77	0.68	0.74	0.53	0.75	0.80
T2 screen	0.48	0.56	0.36	0.69	0.70	0.74
Total screens	**0.73**	**0.67**	**0.69**	**0.55**	**0.75**	**0.79**

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 2013 to 2018, as shown in Table 26. Given the low numbers involved, caution should be taken in making comparisons and some numbers have been withheld where the numerator is lower than six.

Table 26: Diagnostic testing volumes for women with low risk screens by DHB, January 2013 to December 2018

DHB	Number of diagnostic tests						Diagnostic tests per 100 low risk screens					
	2013	2014	2015	2016	2017	2018	2013	2014	2015	2016	2017	2018
Northland	7	S	7	S	S	11	0.74	S	0.66	S	S	0.98
Waitematā	54	35	33	37	43	52	0.89	0.57	0.55	0.59	0.72	0.88
Auckland	55	38	36	20	29	33	1.17	0.79	0.80	0.46	0.78	0.89
Counties Manukau	27	18	23	28	45	29	0.57	0.35	0.45	0.53	0.87	0.57
Waikato	18	30	21	16	33	34	0.51	0.80	0.56	0.41	0.83	0.88
Lakes	S	S	8	S	6	7	S	S	0.84	S	0.60	0.67
Bay of Plenty	9	14	7	12	13	20	0.54	0.80	0.38	0.59	0.58	0.92
Tairāwhiti	S	S	S	S	S	S	S	S	S	S	S	S
Hawke's Bay	6	7	8	S	6	14	0.48	0.59	0.64	S	0.45	1.01
Taranaki	9	S	S	S	S	S	1.11	S	S	S	S	S
MidCentral	9	8	11	S	11	S	0.81	0.72	0.93	S	0.73	S
Whanganui	S	S	S	S	S	S	S	S	S	S	S	S
Capital & Coast	21	15	22	19	15	18	0.84	0.60	0.86	0.72	0.66	0.80
Hutt Valley	8	11	9	6	10	6	0.66	0.88	0.69	0.44	0.78	0.43
Wairarapa	S	S	S	S	6	S	S	S	S	S	1.41	S
Nelson Marlborough	12	S	9	9	7	10	1.01	S	0.77	0.77	0.56	0.82
West Coast	S	S	S	S	S	S	S	S	S	S	S	S
Canterbury	31	45	52	37	47	44	0.67	0.96	1.08	0.74	0.92	0.88
South Canterbury	S	S	S	7	7	S	S	S	S	1.35	1.35	S
Southern	17	33	29	23	22	23	0.67	1.37	1.12	0.87	0.80	0.88
National	**290**	**271**	**283**	**233**	**312**	**325**	**0.73**	**0.67**	**0.69**	**0.55**	**0.75**	**0.79**

(S) Suppressed if the number of diagnostic tests was < 6.

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

Table 27 shows the rate of diagnostic testing for women with low risk screening results by age and ethnicity. The rate of diagnostic testing was higher for women in the older age groups. Māori women were the least likely to have a diagnostic test after a low risk screen.

Table 27: Diagnostic testing volumes for women with low risk screens by age and ethnicity, January 2013 to December 2018

	Diagnostic tests per 100 low risk screens					
	2013	**2014**	**2015**	**2016**	**2017**	**2018**
Age at screen (years)						
Under 20	0.38	0.44	0.33	0.34	0.81	0.81
20–24	0.32	0.37	0.35	0.43	0.68	0.71
25–29	0.37	0.49	0.52	0.50	0.65	0.60
30–34	0.53	0.53	0.60	0.54	0.67	0.84
35–39	1.19	0.98	1.11	0.66	0.99	0.96
40–44	5.30	3.92	3.04	1.33	1.67	1.70
45 and over	6.98	0.00	2.13	3.28	1.61	2.08
Ethnicity						
Māori	0.57	0.46	0.46	0.50	0.65	0.74
Pacific	0.28	0.28	0.48	0.35	0.75	0.79
Asian	0.65	0.58	0.80	0.54	0.89	0.76
Other	0.83	0.78	0.72	0.58	0.73	0.80
National	**0.73**	**0.67**	**0.69**	**0.55**	**0.75**	**0.79**

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

Table 28 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 2017 and 2018.

Table 28: Diagnostic testing volumes for women with low risk screens by risk level, aggregated 2017–2018

Risk level	Number of diagnostic tests	Number of low risk screens	Tests per 100 low risk screens
1:301 to 1:500	43	1,550	2.77
1:501 to 1:1,000	65	3,882	1.67
1:1,001 to 1:2,000	58	6,144	0.94
1:2,001 to 1:3,000	54	4,997	1.08
1:3,001 to 1:4,000	35	4,392	0.80
1:4,001 to 1:5,000	23	3,783	0.61
1:5,001 to 1:10,000	93	14,523	0.64
1:10,001 to 1:100,000	266	43,819	0.61

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 2018 year, 156 diagnostic tests were completed for unscreened women. This is generally lower than the number undertaken in previous years, with 2017 being the exception with only 107 diagnostic tests. Table 29 shows the number of tests by DHB and Table 30 shows the breakdown by age and ethnicity.

Table 29: Diagnostic testing volumes for unscreened women by DHB, January 2013 to December 2018

DHB	Number of diagnostic tests					
	2013	2014	2015	2016	2017	2018
Northland	6	7	8	6	S	S
Waitematā	24	22	22	19	14	24
Auckland	23	25	18	23	10	13
Counties Manukau	27	21	18	21	11	10
Waikato	24	14	15	16	6	12
Lakes	S	6	8	S	S	7
Bay of Plenty	18	12	14	10	S	S
Tairāwhiti	S	S	S	S	S	S
Hawke's Bay	6	7	7	8	S	S
Taranaki	11	S	11	S	S	7
MidCentral	11	11	8	9	S	6
Whanganui	S	S	S	S	S	S
Capital & Coast	16	30	36	25	12	8
Hutt Valley	11	11	22	10	6	6
Wairarapa	S	S	S	S	S	S
Nelson Marlborough	S	S	6	S	S	S
West Coast	S	S	S	S	S	S
Canterbury	23	37	30	30	18	31
South Canterbury	S	S	S	S	S	11
Southern	18	13	19	14	6	6
National	**230**	**235**	**252**	**212**	**107**	**156**

(S) Suppressed if the number of diagnostic tests was < 6.

Table 30: Diagnostic testing volumes for unscreened women by age and ethnicity, January 2013 to December 2018

	Number of diagnostic tests					
	2013	**2014**	**2015**	**2016**	**2017**	**2018**
Age at screen (years)						
Under 20	13	10	16	12	4	4
20–24	33	29	19	17	12	18
25–29	35	39	53	36	27	29
30–34	56	66	70	60	26	47
35–39	50	54	54	56	22	45
40–44	39	34	35	28	15	13
45 and over	4	3	5	3	1	0
Ethnicity						
Māori	49	31	44	32	14	32
Pacific	14	20	21	11	11	7
Asian	31	29	33	36	17	19
Other	136	155	154	133	65	98
National	**230**	**235**	**252**	**212**	**107**	**156**

Diagnostic results for unscreened women

A breakdown of prenatal diagnostic testing results for unscreened women for the 2018 year is given in Table 31. Of the 156 diagnostic tests in 2018 for unscreened women, 110 (71%) had a normal karyotype.

Table 31: Diagnostic testing results for unscreened women, January to December 2018

Karyotype result	Number	Percentage
Normal karyotype	110	70.5
Trisomy 21	17	10.9
Trisomy 18	10	6.4
Trisomy 13	4	2.6
Turner Syndrome	3	1.9
Triploidy	5	3.2
Other chromosomal abnormality	7	4.5
Total	**156**	**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 indicators 9, 10 and 11, the 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 and ethnicity have only been reported for trisomy 21 rather than combining trisomy 21, 18 and 13.

The overall PPV for 2018 was 0.07, lower than previous years (see Table 32). A value of 0.07 means that if a woman receives an increased risk result for trisomy 21, 18 or 13 there is a 7 percent probability that she is carrying a fetus with one of these trisomies.

Table 32: Positive predictive value of screening for trisomy 21, 18 or 13, January 2013 to December 2018

Year	True positives	False positives	PPV	95% confidence interval
2013	142	969	0.128	(0.108, 0.147)
2014	122	1,040	0.105	(0.087, 0.123)
2015	132	1,035	0.113	(0.095, 0.131)
2016	110	1,079	0.093	(0.076, 0.109)
2017	107	1,211	0.081	(0.066, 0.096)
2018	118	1,646	0.067	(0.055, 0.079)

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21, the PPV for 2018 was lower than previous years at 0.05 (see Table 33). This means that if a woman receives an increased risk result for trisomy 21 there is a 5 percent probability that she is carrying a fetus with trisomy 21.

Table 33: Positive predictive value of screening for trisomy 21, January 2013 to December 2018

Year	True positives	False positives	PPV	95% confidence interval
2013	109	980	0.100	(0.082, 0.118)
2014	90	1,046	0.080	(0.064, 0.095)
2015	99	1,046	0.090	(0.070, 0.103)
2016	74	1,072	0.060	(0.050, 0.079)
2017	79	1,184	0.063	(0.049, 0.076)
2018	86	1,629	0.050	(0.040, 0.060)

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 2018 was higher than the PPV for trisomy 21 at 0.14 and 0.05 respectively (see Table 34). However, the number of positive diagnoses for these two trisomies is low so caution should be taken when interpreting these results.

Table 34: Positive predictive value of screening for trisomy 13 or 18, January 2013 to December 2018

Year	True positives	False positives	PPV	95% confidence interval
2013	30	153	0.160	(0.110, 0.218)
2014	27	147	0.160	(0.101, 0.209)
2015	33	148	0.180	(0.126, 0.239)
2016	32	181	0.150	(0.102, 0.198)
2017	25	183	0.120	(0.076, 0.164)
2018	31	199	0.135	(0.091, 0.179)

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

Table 35 shows PPV stratified by the risk level indicated in the screening result. Data have been aggregated for 2017 and 2018. Women that received an increased risk result of 1:5 to 1:20 for trisomy 21 had a 27 percent probability that they were carrying a fetus with trisomy 21. As expected, the PPV was lower for women with increased risks of >1:20 to 1:50 at 6 percent probability, and lower again for women with increased risk results of >1:50 to 1:300 at 1 percent probability.

Table 35: Positive predictive value of screening for trisomy 21 by risk level, aggregated 2017–2018

Risk level	True positives	False positives	PPV
1:5 to 1:20	120	320	0.27
>1:20 to 1:50	18	305	0.06
>1:50 to 1:300	27	2,188	0.01

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

Table 36 shows true positives, false positives and PPV aggregated for 2017–2018 by age and ethnicity. The PPV of screening for trisomy 21 varied by age group. Pacific

women had the lowest PPV (0.02 or 2%) and women in the Other ethnicity had the highest PPV (0.07 or 7%).

Table 36: Positive predictive value of screening for trisomy 21 by age and ethnicity, aggregated 2017–2018

	True positives	False positives	PPV
Age at screen (years)			
Under 20	0	34	0.00
20–24	7	142	0.05
25–29	20	359	0.05
30–34	34	758	0.04
35–39	62	954	0.06
40–44	40	531	0.07
45 and over	2	35	0.05
Ethnicity			
Māori	19	351	0.05
Pacific	6	258	0.02
Asian	27	774	0.03
Other	113	1430	0.07
Total	**165**	**2,813**	**0.06**

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 2018 was 0.04 (or 4%), the highest in the reporting period. This means that out of all women who had a negative diagnostic test or a baby without a trisomy, 4 percent had received an increased risk result for trisomy 21, 18 or 13.

Table 37: False positive rate for trisomy 21, 18 or 13, January 2013 to December 2018

Year	False positives	True negatives	False positive rate	95% confidence interval
2013	969	39,584	0.02	(0.022, 0.025)
2014	1,040	40,547	0.03	(0.024, 0.027)
2015	1,035	41,063	0.02	(0.023, 0.026)
2016	1,079	42,300	0.02	(0.023, 0.026)
2017	1,211	41,767	0.03	(0.027, 0.030)
2018	1,646	41,255	0.04	(0.037, 0.040)

The false positive rate was higher for second trimester screens (6%) than for first trimester screens (3%), consistent with previous years.

Table 38: False positive rate for trisomy 21, 18 or 13 by trimester of screen, January to December 2018

Trimester	False positives	True negatives	False positive rate	95% confidence interval
T1 screens	1,262	35,406	0.034	(0.033, 0.036)
T2 screens	384	5,849	0.062	(0.056, 0.068)
Total	**1,646**	**41,255**	**0.038**	**(0.037, 0.040)**

In 2018, the false positive rate for trisomy 21 when considered alone (0.04 or 4%) was the same as the overall false positive rate (see Table 39). However, the combined false positive rate for trisomy 18 and trisomy 13 is much lower (0.005 or 0.5% for 2018, see Table 40).

Table 39: False positive rate for trisomy 21, January 2013 to December 2018

Year	False positives	True negatives	False positive rate	95% confidence interval
2013	980	39,618	0.02	(0.023, 0.026)
2014	1,046	40,583	0.03	(0.024, 0.027)
2015	1,046	41,093	0.02	(0.023, 0.026)
2016	1,072	42,352	0.02	(0.023, 0.026)
2017	1,184	41,794	0.03	(0.026, 0.029)
2018	1,629	41,272	0.04	(0.036, 0.040)

Table 40: False positive rate for trisomy 18 and 13, January 2013 to December 2018

Year	False positives	True negatives	False positive rate	95% confidence interval
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)
2017	183	42,862	0.004	(0.004, 0.005)
2018	199	42,781	0.005	(0.004, 0.005)

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

False positive rates by age and ethnicity are shown in Table 41. The false positive rate for trisomy 21 increases with age. For example, the false positive rate for women under 20 years in 2018 was 0.01 (1%) compared with 0.31 (31%) 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 41: False positive rate for trisomy 21 by age and ethnicity, January 2013 to December 2018

	2013	2014	2015	2016	2017	2018
Age at screen (years)						
Under 20	0.00	0.01	0.01	0.01	0.02	0.01
20–24	0.01	0.01	0.01	0.01	0.01	0.01
25–29	0.01	0.01	0.01	0.01	0.01	0.01
30–34	0.02	0.02	0.02	0.02	0.02	0.03
35–39	0.05	0.05	0.05	0.05	0.05	0.08
40–44	0.15	0.15	0.19	0.15	0.17	0.26
45 and over	0.37	0.32	0.27	0.21	0.17	0.31
Ethnicity						
Māori	0.02	0.03	0.02	0.02	0.02	0.03
Pacific	0.04	0.04	0.04	0.04	0.04	0.05
Asian	0.03	0.03	0.03	0.03	0.03	0.05
Other	0.02	0.02	0.02	0.02	0.02	0.03

The false positive rate for 2018 varied across ethnic groups from 0.03 (3%) for Māori and Other to 0.05 (5%) for Pacific and Asian.

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 of screening

The overall detection rate for trisomy 21, 18 and 13 for the six years ending 2018 is given in Table 42. Rates for trisomy 21 alone, and for trisomies 18 and 13 together are given in Table 43 and Table 44 respectively. As each of these tables show, detection rates fluctuated over this period.

The overall detection rate for trisomy 21, 18 and 13 for 2018 was 0.78 (78%) (see Table 42). A detection rate of 0.78 means that there is a 78 percent 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 42: Detection rate for trisomy 21, 18 or 13, January 2013 to December 2018

Year	True positives	False negatives	Detection rate	95% confidence interval
2013	142	38	0.79	(0.729, 0.849)
2014	122	27	0.82	(0.757, 0.881)
2015	132	25	0.84	(0.784, 0.898)
2016	110	30	0.79	(0.718, 0.854)
2017	107	35	0.75	(0.683, 0.824)
2018	118	33	0.78	(0.716, 0.847)

The detection rate for trisomy 21 alone is shown in Table 43. The rate for 2018 was higher (0.82) than the overall rate for trisomy 21, 18 and 13 (0.78). The detection rate for trisomy 13 and 18 was lower at 0.65 (Table 44).

Table 43: Detection rate for trisomy 21, January 2013 to December 2018

Year	True positives	False negatives	Detection rate	95% confidence interval
2013	109	26	0.81	(0.741, 0.874)
2014	90	17	0.84	(0.772, 0.910)
2015	99	18	0.85	(0.781, 0.912)
2016	74	21	0.78	(0.696, 0.862)
2017	79	24	0.77	(0.685, 0.849)
2018	86	19	0.82	(0.745, 0.893)

Table 44: Detection rate for trisomy 13 or 18, January 2013 to December 2018

Year	True positives	False negatives	Detection rate	95% confidence interval
2013	30	15	0.67	(0.529, 0.804)
2014	27	15	0.64	(0.498, 0.788)
2015	33	8	0.80	(0.684, 0.926)
2016	32	13	0.71	(0.579, 0.844)
2017	25	14	0.64	(0.490, 0.792)
2018	31	17	0.65	(0.511, 0.781)

Appendix 1: Indicator definitions

Table 45: 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 46: Live births and still births by DHB, 2013–2018

DHB	2013	2014	2015	2016	2017	2018
Northland	2,126	2,099	2,136	2,267	2,236	2,192
Waitematā	7,651	7,850	7,561	7,940	7,725	7,423
Auckland	6,239	6,303	5,896	5,903	5,628	5,431
Counties Manukau	8,179	8,286	8,199	8,239	8,283	8,161
Waikato	5,215	5,244	5,274	5,357	5,318	5,381
Lakes	1,421	1,392	1,506	1,548	1,556	1,526
Bay of Plenty	2,753	2,787	2,789	2,896	3,107	3,012
Tairāwhiti	704	683	734	775	704	699
Hawke's Bay	2,155	2,062	2,000	2,056	2,131	2,108
Taranaki	1,527	1,517	1,516	1,437	1,401	1,562
MidCentral	2,123	2,088	2,112	2,078	2,133	2,162
Whanganui	826	819	815	801	843	808
Capital & Coast	3,630	3,529	3,536	3,461	3,496	3,202
Hutt Valley	1,915	1,853	1,966	1,967	1,948	1,937
Wairarapa	500	473	463	462	536	496
Nelson Marlborough	1,548	1,421	1,417	1,550	1,424	1,498
West Coast	375	352	357	314	358	325
Canterbury	5,822	5,994	6,207	6,306	6,395	6,257
South Canterbury	640	652	660	651	631	603
Southern	3,448	3,284	3,412	3,314	3,439	3,276
Total	**58,797**	**58,688**	**58,556**	**59,322**	**59,292**	**58,059**

Note that 2018 has the lowest number of births recorded over the six-year reporting period.

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

Table 47: Live births and still births by age group, 2013–2018

Age group (years)	2013	2014	2015	2016	2017	2018
<20	3,326	2,991	2,782	2,442	2,296	2,128
20–24	10,810	10,265	9,944	9,580	9,319	8,692
25–29	15,268	15,697	15,719	16,542	16,626	16,251
30–34	16,765	17,568	17,897	18,383	18,692	18,708
35–39	10,036	9,682	9,764	9,961	9,883	10,016
40–44	2,436	2,342	2,297	2,274	2,314	2,095
45+	143	130	140	127	152	162
Unknown	13	13	13	13	10	7
Total	**58,797**	**58,688**	**58,556**	**59,322**	**59,292**	**58,059**

Table 48: Live births and still births by ethnicity, 2013–2018

Ethnicity	2013	2014	2015	2016	2017	2018
Asian	8,122	9,189	9,209	10,516	10,564	10,590
Māori	14,885	14,497	14,785	14,969	14,918	14,569
Other	29,403	28,812	28,486	27,983	27,837	26,934
Pacific	6,387	6,190	6,076	5,854	5,973	5,966
Total	**58,797**	**58,688**	**58,556**	**59,322**	**59,292**	**58,059**

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

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

Of the 1,764 women that had an increased risk for trisomy 21, 18 or 13 during 2018, 665 (38%) had a prenatal diagnostic test (CVS or amniocentesis) and 1,099 (62%) did not. Table 49 shows the diagnostic testing results for the 665 prenatal tests, of which 156 had an abnormal karyotype, including 86 confirmed with Down syndrome.

Table 49: Diagnostic results for women who accessed a prenatal diagnostic test following an increased risk screen for trisomy 21, 18 or 13 during the 2018 year

Karyotype result	Number	Percentage
Normal karyotype	509	76.5
Confirmed Down syndrome	86	12.9
Other result	70	10.5
Total	**665**	**100.0**

Appendix 4: Measuring screening performance

Figure 10 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 10: 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 68 false negative screens in total across the 2017–2018 period. 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 50 shows the number of false negatives aggregated for 2017 and 2018 broken down by the screening risk result in the first column. The second column gives the number of false negatives as a percentage of all negative (low risk) screens. Overall, false negative screens made up 0.08 percent of all negative screens for 2017 to 2018.

Table 50: False negative screens for trisomy 21, 18 and 13 by risk level, January 2017 to December 2018 aggregated

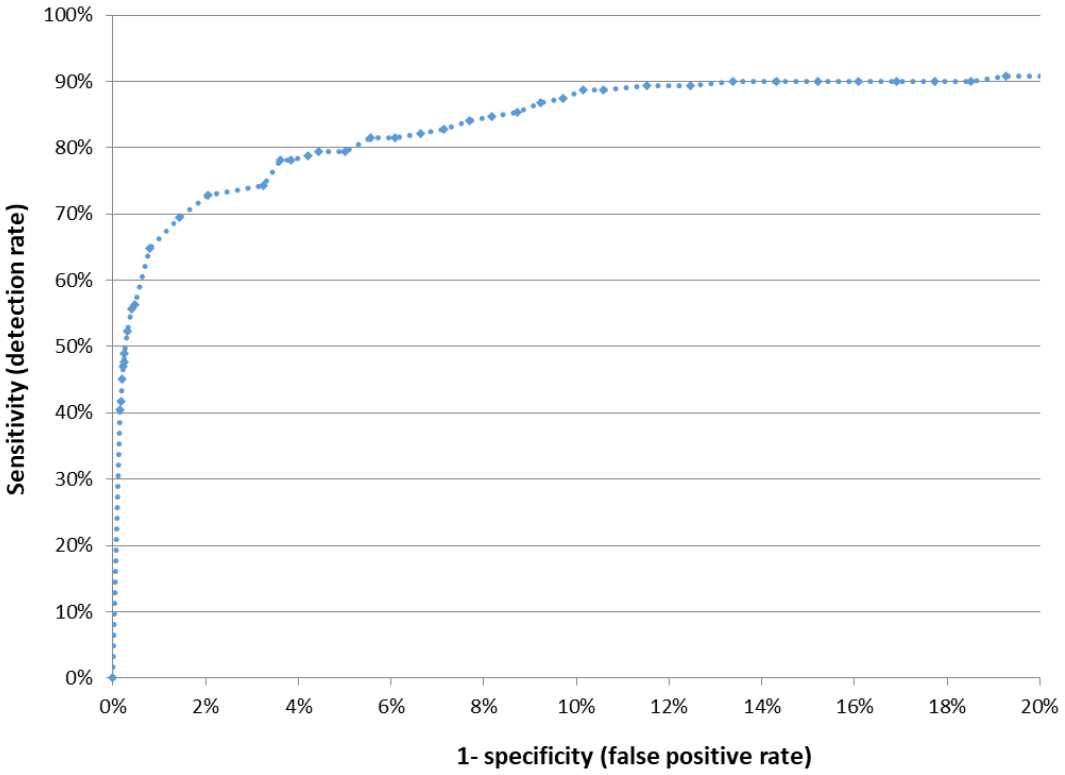
	False negatives	% of negative screens that are false negatives
Risk level	**2017–2018**	**2017–2018**
1:301 to 1:500	12	0.77
1:501 to 1:1000	20	0.52
1:1001 to 1:2000	10	0.16
1:2001 to 1:3000	7	0.14
1:3001 to 1:4000	2	0.05
1:4001 to 1:5000	2	0.05
1:5001 to 1:10,000	5	0.03
Less than 1:10,000	10	0.02
Total	**68**	**0.08**

Appendix 6: ROC curve

Figure 11 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 2018 was 78 percent, and the false positive rate was 3.8%. 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 percent, the false positive rate would increase from 3.8 percent to 8.1 percent. This occurs at a risk cut off of 1:700.

Figure 11: ROC curve for trisomy 21, 18 and 13 screening 2018



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) from 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 compares to the median. MoM is commonly used to report the results of medical screening tests, particularly where the normal range varies according to parameters.

Nasal bone (NB) – an assessment of nasal bone was included in the risk calculation if it was reported at the same time as the NT measurement. Note that since March 2018 the nasal bone measurement is no longer included.

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**.