National Screening Unit

Antenatal Screening for Down Syndrome and Other Conditions

2019 Monitoring Report

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National Screening Unit

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Contents

Executive summary

This report presents data on antenatal screening for Down syndrome and other conditions for the six calendar years from 1 January 2014 to 31 December 2019 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.

Non-invasive prenatal screening (NIPS) is a genetic blood test that can be used to identify pregnancies with a higher risk of trisomy 21, 18 and 13. This blood test is not routinely accessible in New Zealand as it is not included in the screening programme and must be self-funded. Use of NIPS is increasing in New Zealand but as the tests are done privately, there is no available data on how widespread use is, including which population groups are accessing it and at what stage of their pregnancy. The information presented in this report will have been influenced by use of NIPS, but the impact cannot be quantified.

Key points for 2019

- Screening was commenced for 81 percent of women who gave birth in 2019.
- There were approximately 59,300 births during 2019, slighter higher than the number of births in 2018 (58,000), but similar to 2016 and 2017.
- There has been a steady increase in trimester two screens (both commenced and completed) since 2014.
- Māori screening completion rates have increased since 2014/15.

- The national screening completion rate has ranged from 71 to 74 percent across the six years from 2014 to 2019 and was 71 percent in 2019. First trimester screens made up 85 percent of all completed screens in 2019.
- Twelve percent of screens commenced in 2019 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 4.2 in 2019, similar to 2018 (4.1).
- The positive test rate was higher for second trimester screens (5.0 per 100 screens) than for first trimester screens (4.0 per 100 screens) for 2019.
- The overall false positive rate for trisomy 21, 18 and 13 was 4 percent in 2019, the same as in 2018 but higher than previous years (2–3%). The rate was higher for second trimester screens (5%) than for first trimester screens (4%).
- The overall detection rate for trisomy 21, 18 and 13 was 83 percent in 2019, compared to 78 percent in 2018.
- Over this reporting period several changes have occurred that may have impacted on the programme indicators, for example, nasal bone assessment has been excluded since March 2018 and there is increasing use of NIPS.
- Changes to data linkage processes were implemented from 2017. Caution is required when comparing 2017 to 2019 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 plasma 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 and 2 days and 13 weeks and 6 days.
- Second trimester screening, which is a blood test taken between 14 and 20 weeks gestation that measures four maternal serum markers: free beta-human chorionic gonadotropin (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 District Health Board (for samples from Taupō and north of Taupō) and Canterbury Health Laboratories at Canterbury District Health Board (for samples from south of Taupō). 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:

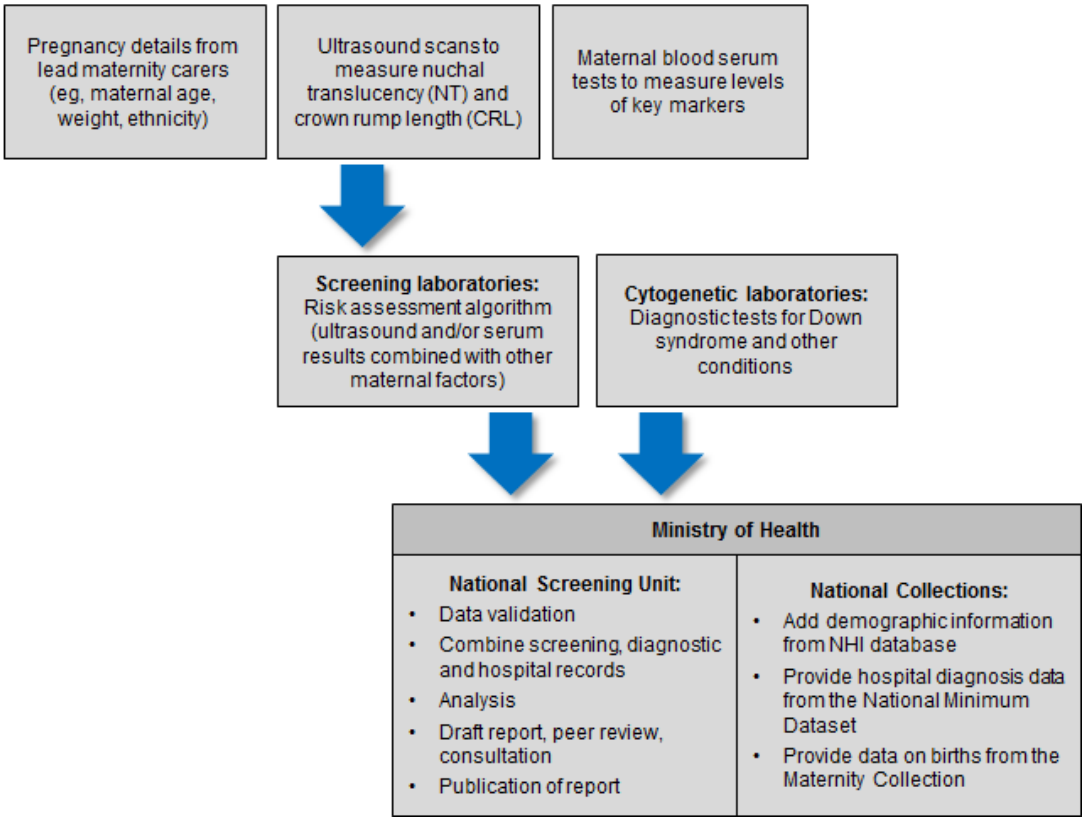
- trisomy 21 (Down Syndrome)
- trisomy 18 (Edwards syndrome)
- trisomy 13 (Patau syndrome)
- triploidy
- Turner syndrome.

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 2014 to 31 December 2019. 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 2018, 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
- 1:21 to 1:50
- 1:51 to 1:300.[1]

Inclusion criteria

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

- Screening commencement date between 1 January 2014 and 31 December 2019 (ie, date of the first test the woman had as part of the screening pathway).

[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.

- 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.

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, or privacy could be comprised.

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

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.
- 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 2017–2019 data but not for years prior to this. Caution is therefore required when comparing data for 2014–2016 with 2017–2019. Where a six-year rate would ordinarily have been applied, a decision has been made to supply a three-year rate (2017–2019) 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.

- In 2019, 17 women had no DHB of domicile 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 breakdowns.

Indicator 1: Screens commenced

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

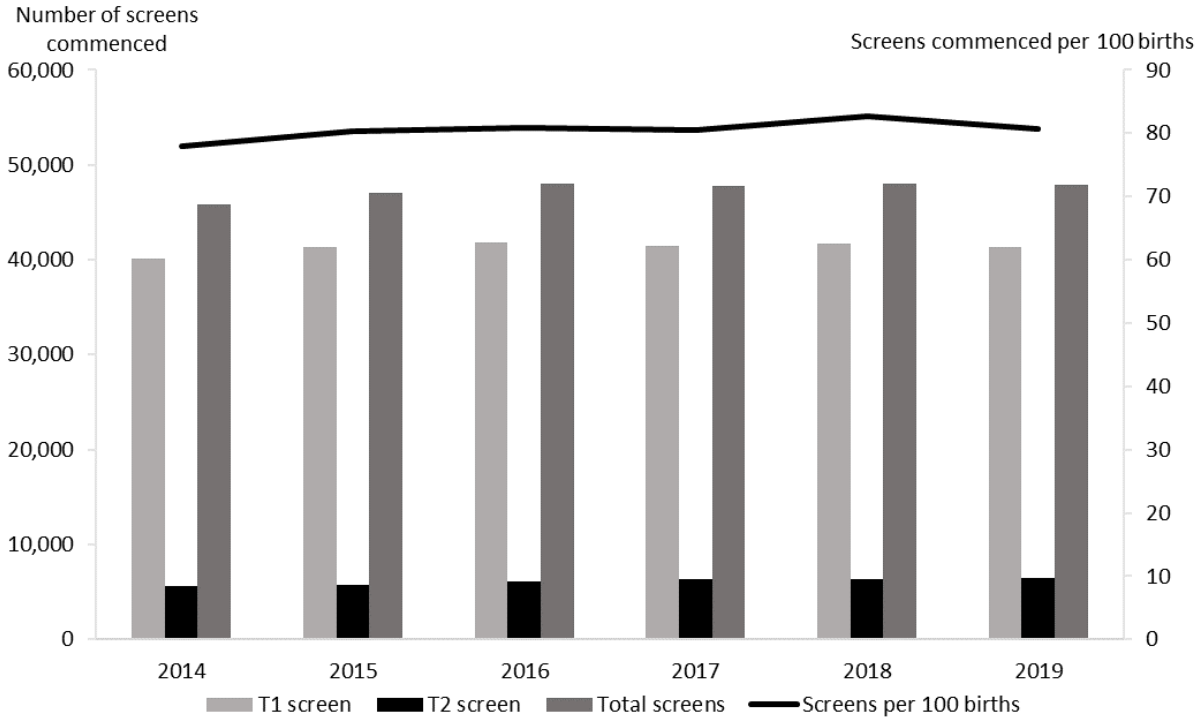
Total screens commenced by trimester

During 2019, a total of 47,868 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 majority of screens were T1 screens. The rate of screens commenced per 100 births increased over time from 78 in 2014 to 83 in 2018 then decreased to 81 in 2019 (see Table 1 and Figure 2).

Table 1: Total screens commenced by trimester, January 2014 to December 2019

Trimester of screen	Number and rate of screens commenced					
	2014	2015	2016	2017	2018	2019
T1 screen	40,172	41,283	41,816	41,403	41,681	41,365
T2 screen	5,613	5,742	6,152	6,369	6,330	6,503
Total screens	**45,785**	**47,025**	**47,968**	**47,772**	**48,011**	**47,868**
Screens per 100 births	78.0	80.3	80.9	80.6	82.7	80.6

Figure 2: Number and rate of screens commenced, January 2014 to December 2019



Screens commenced by DHB

Figure 3 shows the screening commencement rates by DHB for 2019. There was a large variation in rates from 62 per 100 births in Northland to 96 per 100 births in Nelson Marlborough. Only half (50%) 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 2019

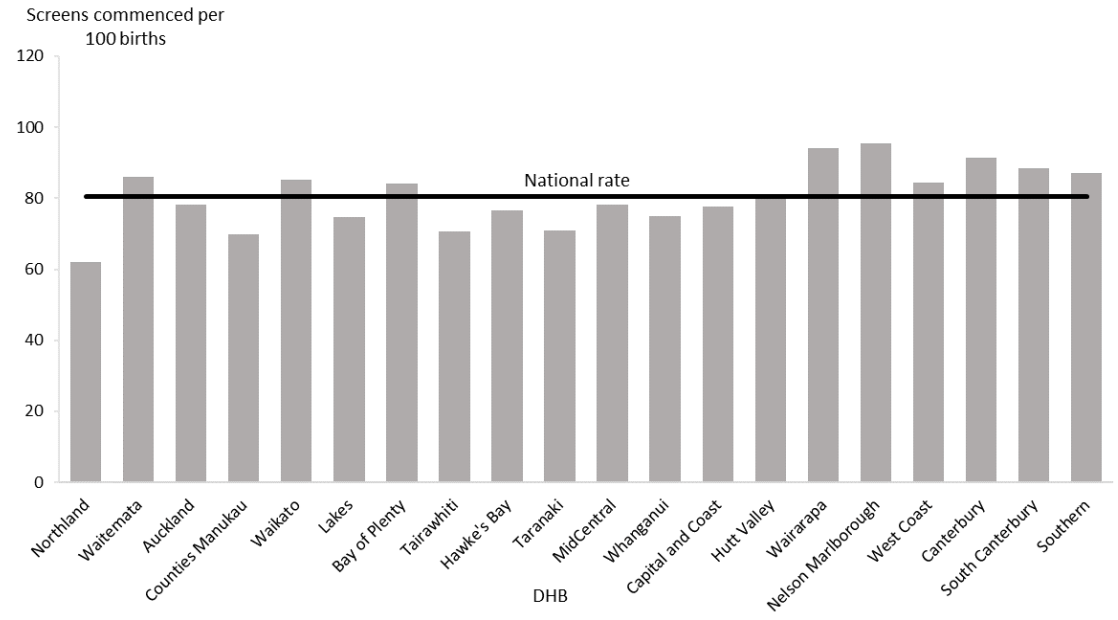


Table 2: Screens commenced by trimester and DHB, January to December 2019

DHB	Number of screens commenced			Screens commenced (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	1,170	264	1,434	50.6	11.4	62.1
Waitematā	5,851	839	6,690	75.2	10.8	86.0
Auckland	3,772	608	4,380	67.5	10.9	78.3
Counties Manukau	4,583	1,289	5,872	54.6	15.4	70.0
Waikato	4,068	571	4,639	74.6	10.5	85.1
Lakes	957	190	1,147	62.3	12.4	74.7
Bay of Plenty	2,385	229	2,614	76.8	7.4	84.1
Tairāwhiti	430	51	481	63.1	7.5	70.6
Hawke's Bay	1,373	173	1,546	68.0	8.6	76.6
Taranaki	911	163	1,074	60.3	10.8	71.0
MidCentral	1,508	186	1,694	69.7	8.6	78.3
Whanganui	537	111	648	62.2	12.8	75.0
Capital & Coast	2,199	269	2,468	69.2	8.5	77.6
Hutt Valley	1,370	210	1,580	69.9	10.7	80.6
Wairarapa	422	66	488	81.5	12.7	94.2
Nelson Marlborough	1,232	153	1,385	84.9	10.5	95.5
West Coast	255	34	289	74.6	9.9	84.5
Canterbury	5,178	700	5,878	80.4	10.9	91.3
South Canterbury	467	84	551	75.0	13.5	88.4
Southern	2,681	312	2,993	77.9	9.1	87.0
National	**41,365**	**6,503**	**47,868**	**69.7**	**11.0**	**80.6**

Note: DHB counts do not sum to National total.

Most DHBs showed an increase in their rate of screens commenced between 2014 and 2019 or had fairly stable rates (see Table 3).

Table 3: Screens commenced per 100 births by DHB, January 2014 to December 2019

DHB	Screens commenced (per 100 births)					
	2014	2015	2016	2017	2018	2019
Northland	55.6	60.1	58.6	64.2	61.7	62.1
Waitematā	86.3	88.4	87.1	86.7	91.4	86.0
Auckland	84.0	85.7	82.0	75.8	82.2	78.3
Counties Manukau	68.7	71.1	71.0	70.6	71.1	70.0
Waikato	80.4	81.8	83.7	85.5	84.0	85.1
Lakes	77.4	74.3	76.7	73.6	80.9	74.7
Bay of Plenty	72.4	77.6	81.1	82.2	82.9	84.1
Tairāwhiti	59.3	68.3	63.6	70.2	78.1	70.6
Hawke's Bay	66.0	72.6	76.2	71.8	75.6	76.6
Taranaki	68.2	74.9	67.8	72.7	74.7	71.0
MidCentral	59.3	63.9	73.1	79.9	74.7	78.3
Whanganui	61.0	70.5	74.1	71.8	77.8	75.0
Capital & Coast	80.3	83.4	86.3	76.1	81.4	77.6
Hutt Valley	78.6	78.7	82.2	76.3	84.0	80.6
Wairarapa	81.6	83.8	89.0	90.1	92.7	94.2
Nelson Marlborough	97.6	96.0	85.1	98.6	91.5	95.5
West Coast	88.3	82.4	86.5	84.4	84.6	84.5
Canterbury	89.5	89.4	91.5	92.4	94.3	91.3
South Canterbury	78.8	86.4	87.5	94.0	94.7	88.4
Southern	83.3	85.1	87.8	89.0	90.2	87.0
National average	**78.0**	**80.3**	**80.9**	**80.6**	**82.7**	**80.6**

Screens commenced by age and ethnicity

Table 4 provides an overall view of screens commenced by age and ethnicity for January 2014 to December 2019. 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.

The 25–29 and 30–34 years age groups had the highest rate of screens commenced for 2019 with a rate of 85 women commencing screening per 100 births in both these groups (Figure 4). From 2014 to 2019 rates have increased overall for most age groups, particularly the younger age groups.

Differences in screening commencement rates by ethnicity remained consistent for 2019. Women of Other ethnicity had the highest rate (97 of 100 births) followed by Asian women (92 of 100 births). The rate of commenced screens for Pacific and Māori women was lower at 55 per 100 births and 53 per 100 births respectively (Figure 5). All groups have shown increasing rates over the reporting period, particularly for Māori with an increase of 9 percentage points from 44 percent in 2014 to 53 percent in 2019. This rate is however well below the national rate of 81 per 100 births in 2019.

Table 4: Screens commenced by age and ethnicity of mother, January 2014 to December 2019

	Number of screens commenced						Screens commenced (per 100 births)					
	2014	**2015**	**2016**	**2017**	**2018**	**2019**	**2014**	**2015**	**2016**	**2017**	**2018**	**2019**
Age at screen (years)												
Under 20	1,990	1,925	1,829	1,683	1,546	1,565	66.6	69.1	74.9	73.3	72.7	74.9
20–24	7,055	7,109	7,000	6,899	6,475	6,341	68.7	71.5	73.0	74.0	74.5	74.3
25–29	12,800	13,189	13,943	14,037	14,162	13,882	81.5	84.0	84.3	84.4	87.1	84.7
30–34	14,623	15,124	15,732	15,804	16,171	16,605	83.2	84.5	85.6	84.5	86.4	85.0
35–39	7,610	8,007	7,781	7,659	8,091	7,973	78.6	82.0	78.1	77.5	80.8	76.6
40–44	1,626	1,593	1,574	1,587	1,476	1,416	69.3	69.3	69.2	68.6	70.5	62.5
45 and over	81	78	109	103	90	86	61.4	56.1	86.5	67.8	55.6	62.3
Ethnicity												
Māori	6,284	6,256	7,176	7,754	7,675	7,844	43.9	42.9	48.7	52.0	52.7	52.9
Pacific	3,005	3,120	3,089	3,284	3,206	3,380	48.7	51.5	52.9	55.0	53.7	55.0
Asian	4,835	8,695	9,851	9,720	10,330	10,554	91.8	94.4	93.6	92.0	97.5	92.0
Other	28,058	28,954	27,852	27,005	26,796	26,090	96.6	100.9	98.7	97.0	99.5	96.9
National	**45,785**	**47,025**	**47,968**	**47,772**	**48,011**	**47,868**	**78.0**	**80.3**	**80.9**	**80.6**	**82.7**	**80.6**

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

Figure 4: Screens commenced by age of mother at screen, January to December 2019

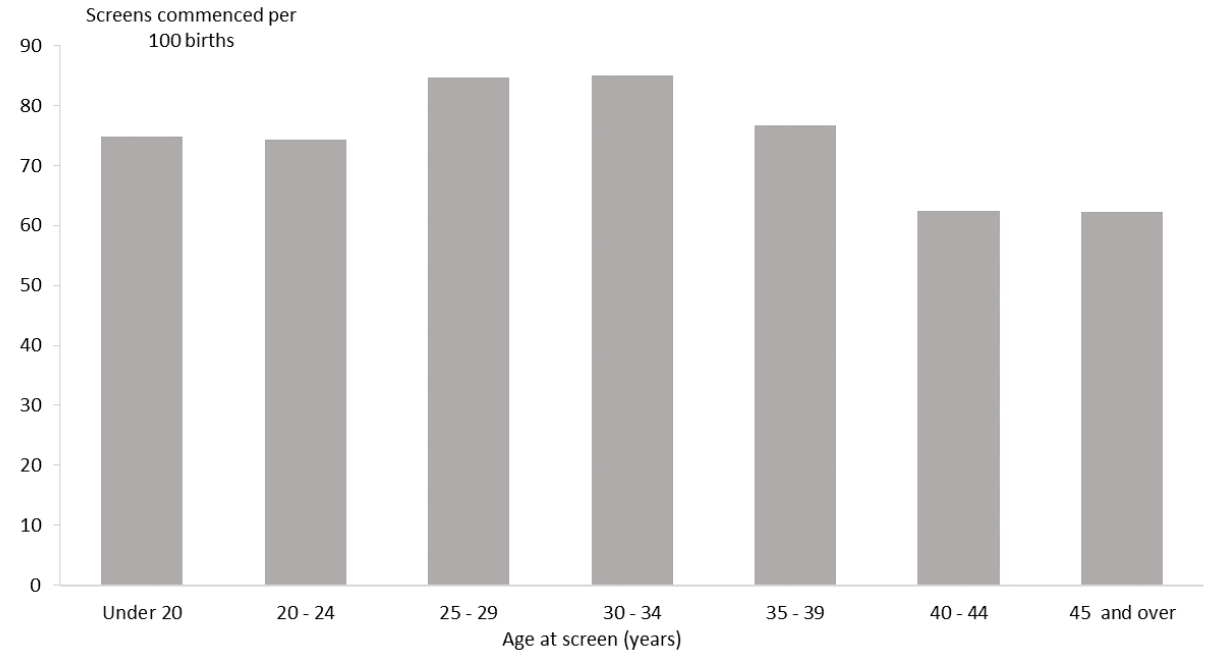
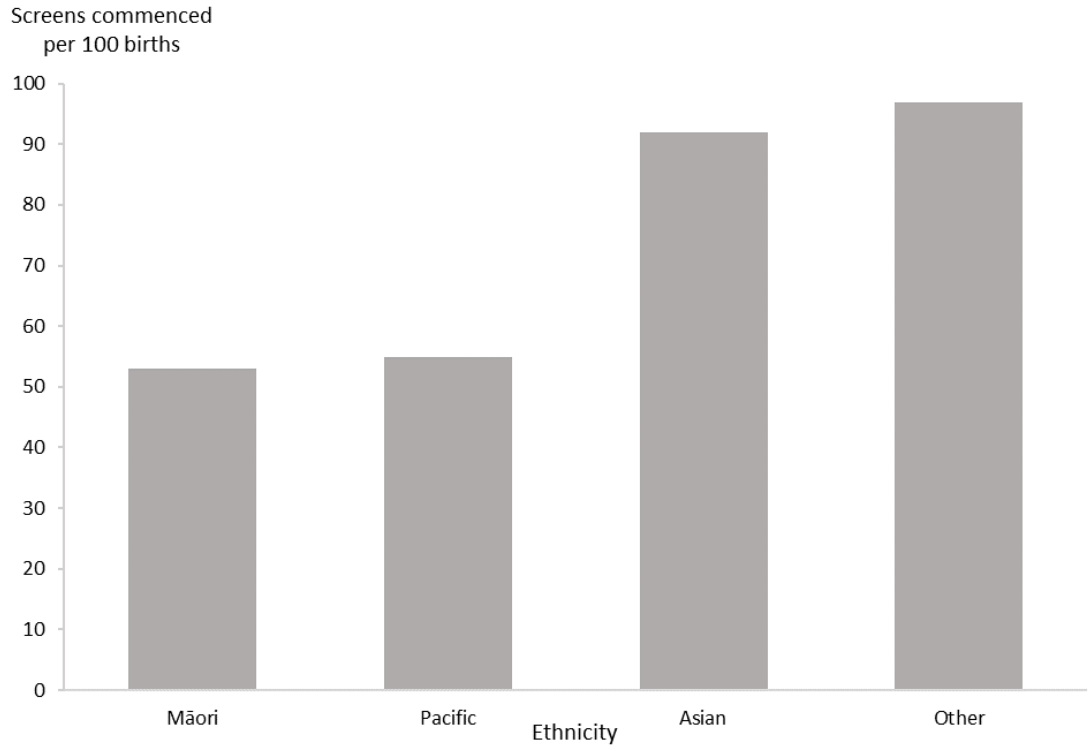


Figure 5: Screens commenced by ethnicity of mother, January to December 2019



Indicator 2: Screens completed

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

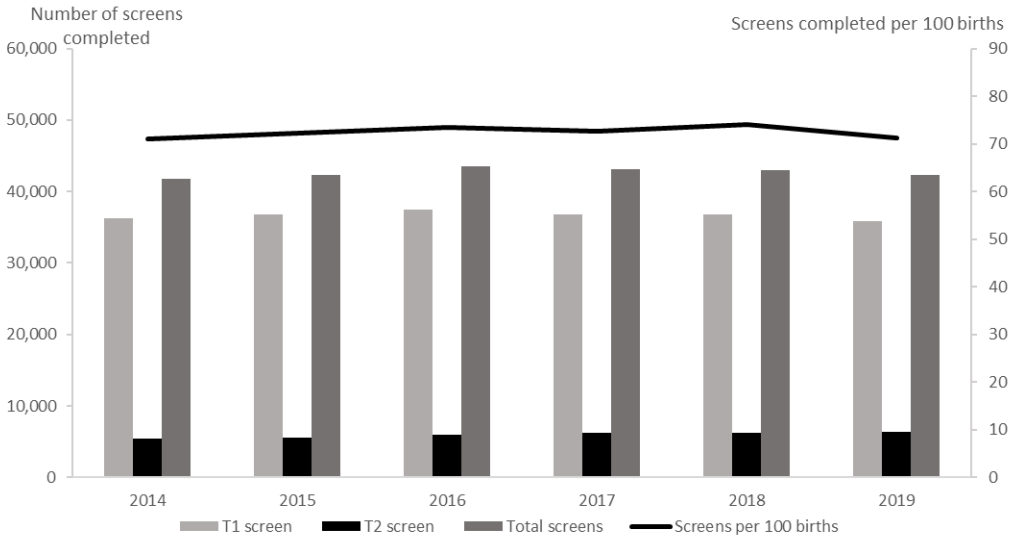
Total screens completed by trimester

During 2019, a total of 42,277 screens were completed, a rate of 71 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 rate of completed screens increased from 71 in 2014 to 74 in 2018, however decreased to 71 completed screens per 100 births in 2019.

Table 5: Total screens completed by trimester, January 2014 to December 2019

Trimester of screen	Number and rate of screens completed					
	2014	**2015**	**2016**	**2017**	**2018**	**2019**
T1 screen	36,280	36,739	37,511	36,836	36,810	35,900
T2 screen	5,456	5,517	6,008	6,284	6,242	6,377
Total screens	**41,736**	**42,256**	**43,519**	**43,120**	**43,052**	**42,277**
Screens per 100 births	71.1	72.2	73.4	72.7	74.2	71.2

Figure 6: Number and rate of screens completed, January 2014 to December 2019



Screens completed by DHB

Screening completion rates for 2019 varied across DHBs from 54 completed screens per 100 births in Northland to 87 per 100 births in Nelson Marlborough (see Figure 7). Table 6 gives a full breakdown by the trimester of screen.

Figure 7: Screens completed by DHB, January to December 2019

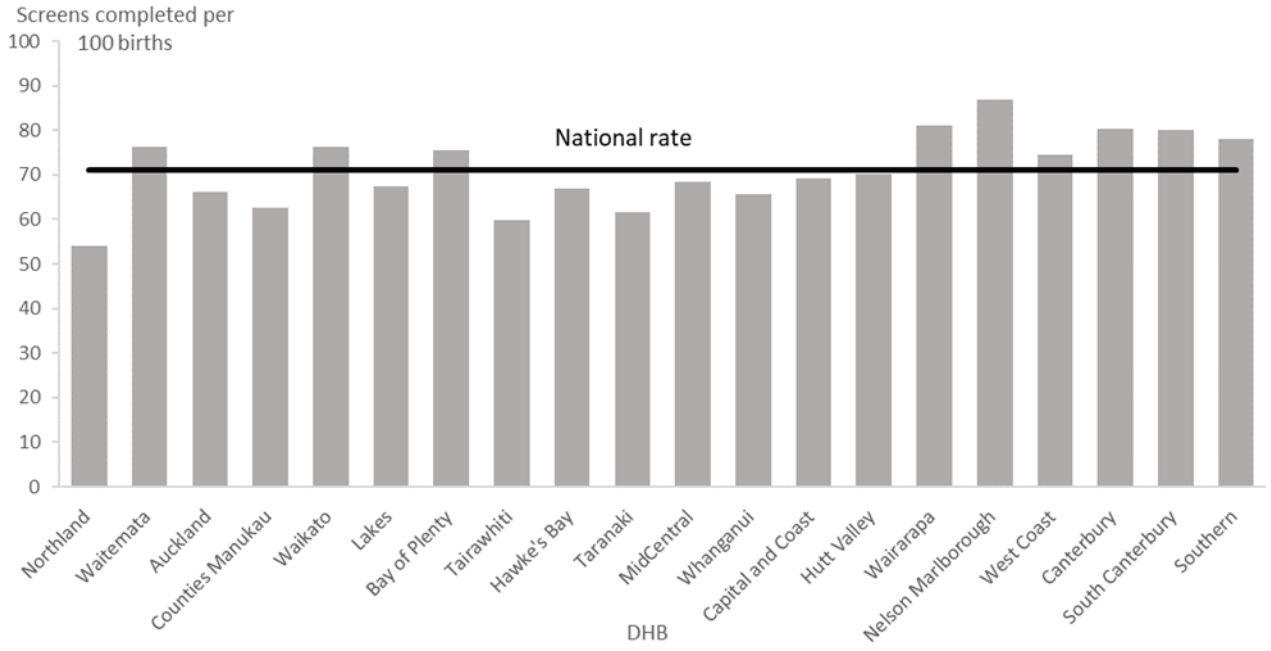


Table 6: Screening completion by trimester and DHB, January to December 2019

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	991	260	1,251	42.9	11.3	54.2
Waitematā	5,093	828	5,921	65.5	10.6	76.1
Auckland	3,104	594	3,698	55.5	10.6	66.1
Counties Manukau	4,010	1,253	5,263	47.8	14.9	62.7
Waikato	3,591	560	4,151	65.9	10.3	76.2
Lakes	848	187	1,035	55.2	12.2	67.4
Bay of Plenty	2,116	226	2,342	68.1	7.3	75.4
Tairāwhiti	357	50	407	52.4	7.3	59.8
Hawke's Bay	1,184	167	1,351	58.7	8.3	66.9
Taranaki	772	161	933	51.1	10.6	61.7
MidCentral	1,295	185	1,480	59.8	8.5	68.4
Whanganui	456	111	567	52.8	12.8	65.6
Capital & Coast	1,937	263	2,200	60.9	8.3	69.2
Hutt Valley	1,171	206	1,377	59.7	10.5	70.2
Wairarapa	354	66	420	68.3	12.7	81.1
Nelson Marlborough	1,107	152	1,259	76.3	10.5	86.8
West Coast	222	33	255	64.9	9.6	74.6
Canterbury	4,489	683	5,172	69.7	10.6	80.3
South Canterbury	415	84	499	66.6	13.5	80.1
Southern	2,378	307	2,685	69.1	8.9	78.1
National	**35,900**	**6,377**	**42,277**	**60.5**	**10.7**	**71.2**

Note: DHB counts do not sum to National total.

As shown in Table 7, many DHBs showed a trend of increasing rates of screening completion over the five years from 2014 to 2018, however for the majority (65%) of DHBs, screening completion rates decreased from 2018 to 2019.

Table 7: Screening completion by DHB, January 2014 to December 2019

DHB	Screens completed (per 100 births)					
	2014	2015	2016	2017	2018	2019
Northland	48.0	51.6	50.9	56.2	53.6	54.2
Waitematā	81.0	81.8	81.4	79.8	82.9	76.1
Auckland	78.8	79.1	75.6	68.6	72.2	66.1
Counties Manukau	63.2	64.5	65.5	64.4	64.9	62.7
Waikato	72.5	72.4	74.6	76.3	74.8	76.2
Lakes	69.9	65.7	67.8	65.7	71.2	67.4
Bay of Plenty	64.5	67.8	71.8	73.6	74.6	75.4
Tairāwhiti	51.5	53.8	51.1	59.1	65.1	59.8
Hawke's Bay	59.4	64.2	68.6	63.7	67.6	66.9
Taranaki	61.2	66.3	62.1	66.4	68.3	61.7
MidCentral	54.0	56.9	66.1	72.3	66.3	68.4
Whanganui	53.1	58.5	65.8	63.6	67.6	65.6
Capital & Coast	72.6	75.1	77.8	67.8	73.3	69.2
Hutt Valley	68.9	68.0	71.6	67.3	74.4	70.2
Wairarapa	70.6	72.8	77.9	80.6	81.0	81.1
Nelson Marlborough	87.6	84.7	77.4	90.1	84.6	86.8
West Coast	78.9	72.3	77.7	76.8	72.6	74.6
Canterbury	81.2	80.6	82.5	83.0	84.2	80.3
South Canterbury	75.3	79.8	81.5	85.4	88.2	80.1
Southern	74.8	77.9	81.1	81.7	82.5	78.1
National average	**71.1**	**72.2**	**73.4**	**72.7**	**74.2**	**71.2**

Screens completed by age and ethnicity

Table 8 provides an overall view of screens completed by age and ethnicity for January 2014 to December 2019, with similar trends to screening commencement.

Most groups showed an overall increase in completion rates over the five-year period from 2014 to 2018, with the biggest increases seen in Māori and Pacific ethnicities and younger women. However, screening completion rates decreased from 2018 to 2019 for most age groups and for Asian and Other ethnic groups.

In 2019, screening completion rates were highest for women aged 25–29 and 30–34 compared to other age groups, with 77 women completing screening per 100 births in these two age groups (Figure 8).

Completion rates were highest among women of Other ethnicity at 86 per 100 births in 2019. This was followed closely by women of Asian ethnicity at 84 per 100 births. The rate of completed screens for Pacific and Māori women remains lower at 48 per 100 births and 44 per 100 births respectively (Figure 9).

Table 8: Screens completed by age and ethnicity of mother, January 2014 to December 2019

	Number of screens completed						Screens completed (per 100 births)					
	2014	**2015**	**2016**	**2017**	**2018**	**2019**	**2014**	**2015**	**2016**	**2017**	**2018**	**2019**
Age at screen (years)												
Under 20	1,604	1,510	1,474	1,376	1,243	1,282	53.6	54.2	60.3	59.9	58.4	61.3
20–24	6,070	5,992	6,079	5,948	5,588	5,426	59.1	60.3	63.4	63.8	64.3	63.6
25–29	11,685	11,824	12,675	12,779	12,898	12,554	74.4	75.3	76.6	76.9	79.4	76.6
30–34	13,675	14,030	14,709	14,651	14,823	14,940	77.8	78.3	80.1	78.4	79.2	76.5
35–39	7,144	7,430	7,137	6,959	7,205	6,897	73.9	76.1	71.6	70.4	71.9	66.3
40–44	1,486	1,406	1,366	1,328	1,225	1,119	63.3	61.2	60.0	57.4	58.5	49.4
45 and over	72	64	79	79	70	59	54.5	46.0	62.7	52.0	43.2	42.8
Ethnicity												
Māori	5,178	4,911	5,924	6,442	6,387	6,513	36.2	33.7	40.2	43.2	43.8	44.0
Pacific	2,598	2,626	2,673	2,876	2,782	2,927	42.1	43.3	45.8	48.2	46.6	47.6
Asian	8,034	8,114	9,304	9,093	9,594	9,649	87.4	88.1	88.4	86.1	90.6	84.1
Other	25,926	26,605	25,618	24,701	24,287	23,188	89.2	92.7	90.8	88.7	90.2	86.1
National	**41,736**	**42,256**	**43,519**	**43,120**	**43,052**	**42,277**	**71.1**	**72.2**	**73.4**	**72.7**	**74.2**	**71.2**

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

Figure 8: Screens completed by age of mother at screen, January to December 2019

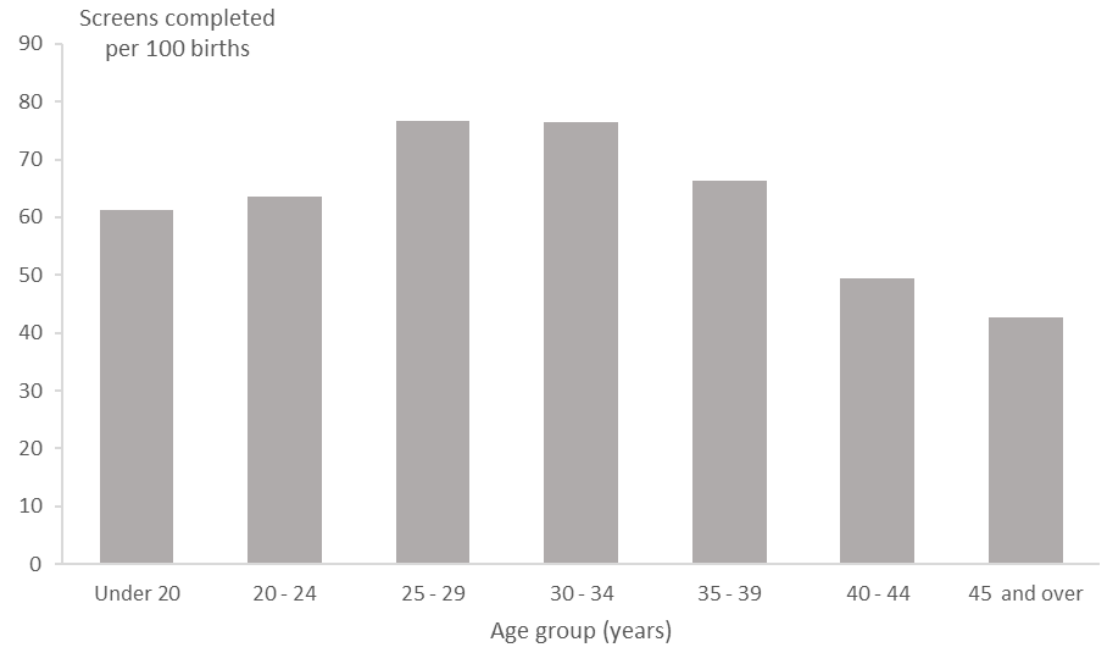
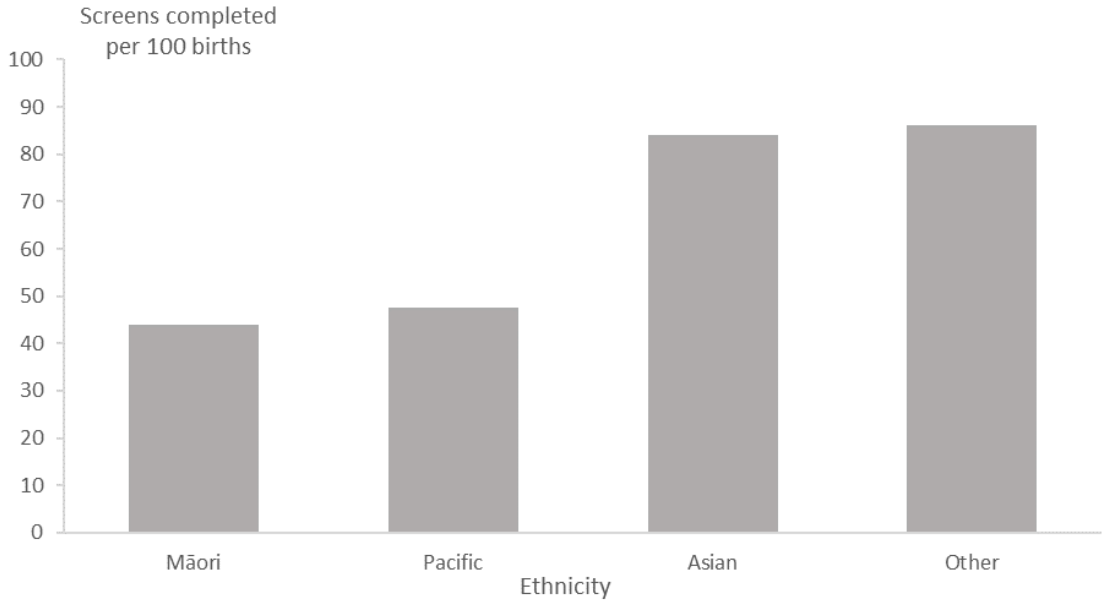


Figure 9: Screens completed by ethnicity of mother, January to December 2019



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 2014 to 2019. 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 (43% in 2019). PAPP-A was included in 11.5 percent of second trimester screens in 2019, similar to 2018.

Table 9: Screening pathway variance by type, January 2014 to December 2019

Year	Second trimester screening results				
	Number			Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
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
2019	6,377	2,743	732	43.0	11.5

Screening pathway variance by DHB

Table 10 shows a breakdown of screening pathway variance by DHB and type of variance for the 2019 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 2019

DHB	Second trimester screening results				
	Number			Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
Northland	260	94	34	36.2	13.1
Waitematā	828	351	95	42.4	11.5
Auckland	594	199	105	33.5	17.7
Counties Manukau	1,253	334	165	26.7	13.2
Waikato	560	298	36	53.2	6.4
Lakes	187	88	11	47.1	5.9
Bay of Plenty	226	111	20	49.1	8.8
Tairāwhiti	50	23	10	46.0	20.0
Hawke's Bay	167	77	23	46.1	13.8
Taranaki	161	62	18	38.5	11.2
MidCentral	185	105	14	56.8	7.6
Whanganui	111	62	S	55.9	S
Capital & Coast	263	134	14	51.0	5.3
Hutt Valley	206	111	14	53.9	6.8
Wairarapa	66	39	S	59.1	S

Nelson Marlborough	152	94	16	61.8	10.5
West Coast	33	16	8	48.5	24.2
Canterbury	683	328	110	48.0	16.1
South Canterbury	84	46	8	54.8	9.5
Southern	307	171	25	55.7	8.1
National	**6,377**	**2,743**	**732**	**43.0**	**11.5**

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 2019 year. The results show higher proportions for pathway variance for women in the 20–24 and 25–29 age groups (44–45%) and women of Other ethnicity (55%).

Table 11: Screening pathway variance by age and ethnicity, January to December 2019

	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	394	149	29	37.8	7.4
20–24	1,278	567	108	44.4	8.5
25–29	1,875	837	211	44.6	11.3
30–34	1,764	756	241	42.9	13.7
35–39	892	371	127	41.6	14.2
40–44	169	62	16	36.7	9.5
45 and over	5	1	0	20.0	0.0
Ethnicity					
Māori	1,691	719	131	42.5	7.7
Pacific	1,127	317	111	28.1	9.8
Asian	1,337	491	200	36.7	15.0
Other	2,222	1,216	290	54.7	13.1
National	**6,377**	**2,743**	**732**	**43.0**	**11.5**

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 2019 was 5,591, which equates to 12 percent of screens commenced that year and demonstrates an overall increase in incomplete screens over the past six years.

Table 12: Incomplete screens by trimester, January 2014 to December 2019

Trimester of screen	Number of incomplete screens					
	2014	2015	2016	2017	2018	2019
T1 screens	3,892	4,544	4,305	4,567	4,871	5,465
T2 screens	157	225	144	85	88	126
Total screens	**4,049**	**4,769**	**4,449**	**4,652**	**4,959**	**5,591**

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 2019 was 13 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 2014 to December 2019

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
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
2019	5,465	35,900	41,365	4,063	1,398	17	9.8	3.4	13.2	74.3	25.6

Incomplete T1 screens by reason and DHB

Table 14 provides the same breakdown by DHB for the 2019 year. The lower numbers involved limit DHB comparisons. The range in the percentage of screens incomplete due to no blood sample was from 36 percent (West Coast) to 85 percent (Tairāwhiti and MidCentral).

Table 14: Incomplete T1 screens by reason and DHB, January to December 2019

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	179	991	1,170	119	60	S	10.2	5.1	15.3	66.5	33.5
Waitematā	758	5,093	5,851	573	185	S	9.8	3.2	13.0	75.6	24.4
Auckland	668	3,104	3,772	551	117	S	14.6	3.1	17.7	82.5	17.5
Counties Manukau	573	4,010	4,583	403	170	S	8.8	3.7	12.5	70.3	29.7
Waikato	477	3,591	4,068	384	93	S	9.4	2.3	11.7	80.5	19.5
Lakes	109	848	957	82	27	S	8.6	2.8	11.4	75.2	24.8
Bay of Plenty	269	2,116	2,385	203	66	S	8.5	2.8	11.3	75.5	24.5
Tairāwhiti	73	357	430	62	11	S	14.4	2.6	17.0	84.9	15.1
Hawke's Bay	189	1,184	1,373	130	59	S	9.5	4.3	13.8	68.8	31.2

Taranaki	139	772	911	88	51	S	9.7	5.6	15.3	63.3	36.7
MidCentral	213	1,295	1,508	180	32	S	11.9	2.1	14.1	84.5	15.0
Whanganui	81	456	537	56	24	S	10.4	4.5	15.1	69.1	29.6
Capital & Coast	262	1,937	2,199	193	68	S	8.8	3.1	11.9	73.7	26.0
Hutt Valley	199	1,171	1,370	157	41	S	11.5	3.0	14.5	78.9	20.6
Wairarapa	68	354	422	52	16	S	12.3	3.8	16.1	76.5	23.5
Nelson Marlborough	125	1,107	1,232	82	43	S	6.7	3.5	10.1	65.6	34.4
West Coast	33	222	255	12	21	S	4.7	8.2	12.9	36.4	63.6
Canterbury	689	4,489	5,178	474	215	S	9.2	4.2	13.3	68.8	31.2
South Canterbury	52	415	467	38	14	S	8.1	3.0	11.1	73.1	26.9
Southern	303	2,378	2,681	220	83	S	8.2	3.1	11.3	72.6	27.4
National	**5,465**	**35,900**	**41,365**	**4,063**	**1,398**	**17**	**9.8**	**3.4**	**13.2**	**74.3**	**25.6**

Note: DHB counts do not sum to National total.

(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 they are missing dating information or weight, if the sample is taken later than 20 weeks of pregnancy, or if the sample is damaged and not repeated. In 2019, 2 percent of T2 commenced screens were incomplete, compared with 13 percent of T1 commenced screens. As Table 15 shows, the percentage of incomplete T2 screens decreased from 3.9 percent in 2015 to 1.9 percent in 2019.

Table 15: Incomplete T2 screens, January 2014 to December 2019

Year	Commenced second trimester	No result issued	Percentage incomplete
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
2019	6,503	126	1.9
Total	**36,709**	**825**	**Ave: 2.2**

Incomplete T2 screens by DHB

Table 16 shows a breakdown of incomplete T2 screens by DHB for the 2019 year. The very low numbers involved limit meaningful DHB comparisons.

Table 16: Incomplete T2 screens by DHB, January to December 2019

DHB	Commenced second trimester	No result issued	% incomplete
Northland	264	S	S
Waitematā	839	11	1.3
Auckland	608	14	2.3
Counties Manukau	1,289	36	2.8
Waikato	571	11	1.9
Lakes	190	S	S
Bay of Plenty	229	S	S
Tairāwhiti	51	S	S
Hawke's Bay	173	6	3.5
Taranaki	163	S	S
MidCentral	186	S	S
Whanganui	111	S	S
Capital & Coast	269	6	2.2
Hutt Valley	210	S	S
Wairarapa	66	S	S
Nelson Marlborough	153	S	S
West Coast	34	S	S
Canterbury	700	17	2.4
South Canterbury	84	S	S
Southern	312	S	S
National	**6,503**	**126**	**1.9**

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 the 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 2019-year, 4.2 increased-risk results were issued for every 100 screens completed. This is similar to the rate reported in 2018.

Table 17: Number and rate per 100 screens of increased-risk screening results for trisomy 21, 18 or 13, January 2014 to December 2019

	Number and rate of increased-risk screens					
	2014	2015	2016	2017	2018	2019
Total increased-risk results	1,162	1,168	1,189	1,318	1,764	1,764
Positive test rate per 100 completed screens	2.8	2.8	2.7	3.1	4.1	4.2

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 2019 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 2019

	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 (years)			
Under 20	14	1,282	1.1
20–24	74	5,426	1.4
25–29	197	12,554	1.6
30–34	466	14,940	3.1
35–39	644	6,897	9.3
40–44	353	1,119	31.5
45 and over	16	59	27.1
Ethnicity			
Māori	230	6,513	3.5
Pacific	155	2,927	5.3
Asian	539	9,649	5.6
Other	840	23,188	3.6
National	**1,764**	**42,277**	**4.2**

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 4.1 per 100 screens. The second trimester positive test rate for trisomy 21 was higher than the first trimester positive test rate (4.7 and 3.9 respectively). 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 2014 to December 2019

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						
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
2019	1,718	4.1	1,416	3.9	302	4.7

Trisomy 18						
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
2019	170	0.4	142	0.4	28	0.4
Trisomy 13						
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
2019	151	0.4	136	0.4	15	0.2
Any one or more of trisomy 21, 18 or 13						
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
2019	1,764	4.2	1,442	4.0	322	5.0

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 2019 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 2019.

Table 20: Increased-risk screening results for trisomy 21, 18 and 13 by risk level, January to December 2019

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 to 1:20	206	46	44
1:21 to 1:50	181	22	20
1:51 to 1:300	1,331	102	87

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 by trimester of screen from 2014 to 2019. In 2019, for every 100 women that received an increased-risk result after a first or second trimester screen, 39 women had a diagnostic test. There was a decreasing trend in the diagnostic testing rate from 2014 to 2017, followed by a slight increase in 2018 and 2019. The second trimester diagnostic testing rate (39.4) was slightly higher than the first trimester diagnostic testing rate (38.7) in 2019, for the first time. See Appendix 3 for a summary of diagnostic test results for women who had an increased-risk screen in 2019.

Table 21: Diagnostic testing volumes for women with increased-risk screens by trimester of screen, January 2014 to December 2019

Trimester of screen	Diagnostic tests per 100 increased-risk screens					
	2014	2015	2016	2017	2018	2019
T1 screen	62.5	59.0	46.9	36.7	38.4	38.7
T2 screen	47.4	44.3	40.5	29.0	35.3	39.4
Total screens	**59.0**	**56.3**	**45.7**	**35.1**	**37.7**	**38.8**

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 2014 to December 2019

DHB	Number of diagnostic tests						Diagnostic tests per 100 increased-risk screens					
	2014	2015	2016	2017	2018	2019	2014	2015	2016	2017	2018	2019
Northland	26	21	12	12	18	25	59.1	48.8	40.0	34.3	38.3	44.6
Waitematā	116	107	82	78	102	97	61.7	57.5	44.6	37.5	37.2	37.2
Auckland	89	76	72	49	63	70	55.3	53.5	45.0	30.6	31.8	34.7
Counties Manukau	76	86	78	55	99	107	50.3	53.8	54.9	31.4	39.6	45.5
Waikato	41	42	45	29	56	66	64.1	60.0	52.9	30.2	39.4	40.2
Lakes	21	28	16	14	19	14	53.8	71.8	59.3	46.7	46.3	41.2
Bay of Plenty	21	20	17	18	26	24	63.6	66.7	44.7	40.0	39.4	38.1
Tairāwhiti	S	S	S	S	7	S	S	S	S	S	43.8	S
Hawke's Bay	20	15	8	7	15	13	58.8	51.7	28.6	26.9	33.3	40.6
Taranaki	12	10	8	S	10	17	48.0	43.5	36.4	S	35.7	51.5
MidCentral	11	8	15	20	19	25	57.9	44.4	46.9	50.0	52.8	44.6
Whanganui	S	S	6	S	9	11	S	S	66.7	S	52.9	57.9
Capital & Coast	46	65	41	30	34	37	59.7	60.7	60.3	32.6	37.0	36.3
Hutt Valley	15	18	15	15	18	19	53.6	64.3	45.5	45.5	34.0	28.4
Wairarapa	S	S	S	S	S	6	S	S	S	S	S	50.0

Nelson Marlborough	19	15	14	13	20	29	79.2	57.7	51.9	48.1	44.4	56.9
West Coast	8	S	6	S	S	S	42.1	S	85.7	S	S	S
Canterbury	122	83	80	70	95	90	65.6	50.6	36.7	32.0	34.2	34.5
South Canterbury	S	9	S	7	S	S	S	75.0	S	36.8	S	S
Southern	33	40	20	31	44	24	67.3	60.6	37.0	44.9	48.4	32.0
National	**685**	**657**	**543**	**463**	**665**	**685**	**59.0**	**56.3**	**45.7**	**35.1**	**37.7**	**38.8**

(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 2014 to 2019.

For 2019, diagnostic testing rates were highest for Māori women (41 per 100 increased-risk screens), followed by Asian women and women of Other ethnicity (40 and 38 per 100 increased-risk screens respectively), and then Pacific women (36 per 100 increased-risk screens). From age 20 years, diagnostic testing rates reduce with each age grouping, except for women aged 45 and over.

Table 23: Diagnostic testing volumes for women with increased-risk screens by age and ethnicity, January 2014 to December 2019

	Diagnostic tests per 100 increased-risk screens					
	2014	**2015**	**2016**	**2017**	**2018**	**2019**
Age at screen (years)						
Under 20	50.0	53.8	45.5	17.4	28.6	64.3
20–24	53.9	51.7	55.6	43.5	50.0	48.6
25–29	62.7	58.1	49.4	38.2	44.7	43.1
30–34	64.9	61.8	47.7	38.8	41.3	42.7
35–39	57.1	57.0	46.0	32.9	35.3	36.0
40–44	58.1	50.9	39.0	29.8	32.5	33.1
45 and over	36.0	41.2	27.8	35.3	13.6	43.8
Ethnicity						
Māori	38.4	45.1	46.7	30.1	37.3	40.9
Pacific	39.2	36.2	34.3	31.0	33.1	36.1
Asian	67.0	63.3	56.3	37.7	37.9	39.5
Other	62.8	58.7	42.1	35.9	38.5	38.3
National	**59.0**	**56.3**	**45.7**	**35.1**	**37.7**	**38.8**

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

Each screening result includes a separate risk for each of trisomy 21, 18 and 13. For the analysis in this report, women were assigned a combined trisomy risk level based on the highest risk score they received across the three trisomies. Table 24 shows the number of diagnostic tests for women that received an increased-risk result during 2019 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 75 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 2019

Risk level	Number of diagnostic tests	Number of increased-risk screens	Tests per 100 increased-risk screens
1:5 to 1:20	164	219	74.9
1:21 to 1:50	109	184	59.2
1:51 to 1:300	412	1,361	30.3

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 Turner syndrome 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 0.81 per 100 low-risk screens in 2019, which is the highest rate for the reporting period.

Table 25: Diagnostic testing volumes for women with low-risk screens by trimester of screen, January 2014 to December 2019

Trimester of screen	Diagnostic tests per 100 low-risk screens					
	2014	2015	2016	2017	2018	2019
T1 screen	0.68	0.74	0.53	0.75	0.80	0.84
T2 screen	0.56	0.36	0.69	0.70	0.74	0.64
Total screens	**0.67**	**0.69**	**0.55**	**0.75**	**0.79**	**0.81**

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 2014 to 2019, as shown in Table 26. Given the low numbers involved, caution should be taken in making comparisons between DHBs.

Table 26: Diagnostic testing volumes for women with low-risk screens by DHB, January 2014 to December 2019

DHB	Number of diagnostic tests						Diagnostic tests per 100 low-risk screens					
	2014	2015	2016	2017	2018	2019	2014	2015	2016	2017	2018	2019
Northland	S	7	S	S	11	S	S	0.66	S	S	0.98	S
Waitematā	35	33	37	43	52	53	0.57	0.55	0.59	0.72	0.88	0.94
Auckland	38	36	20	29	33	32	0.79	0.80	0.46	0.78	0.89	0.92
Counties Manukau	18	23	28	45	29	53	0.35	0.45	0.53	0.87	0.57	1.05
Waikato	30	21	16	33	34	30	0.80	0.56	0.41	0.83	0.88	0.75
Lakes	S	8	S	6	7	11	S	0.84	S	0.60	0.67	1.10
Bay of Plenty	14	7	12	13	20	17	0.80	0.38	0.59	0.58	0.92	0.75
Tairāwhiti	S	S	S	S	S	S	S	S	S	S	S	S
Hawke's Bay	7	8	S	6	14	7	0.59	0.64	S	0.45	1.01	0.53
Taranaki	S	S	S	S	S	S	S	S	S	S	S	S
MidCentral	8	11	S	11	S	6	0.72	0.93	S	0.73	S	0.42
Whanganui	S	S	S	S	S	S	S	S	S	S	S	S
Capital & Coast	15	22	19	15	18	17	0.60	0.86	0.72	0.66	0.80	0.81
Hutt Valley	11	9	6	10	6	7	0.88	0.69	0.44	0.78	0.43	0.53
Wairarapa	S	S	S	6	S	S	S	S	S	1.41	S	S

Nelson Marlborough	S	9	9	7	10	13	S	0.77	0.77	0.56	0.82	1.08
West Coast	S	S	S	S	S	S	S	S	S	S	S	S
Canterbury	45	52	37	47	44	47	0.96	1.08	0.74	0.92	0.88	0.96
South Canterbury	S	S	7	7	S	S	S	S	1.35	1.35	S	S
Southern	33	29	23	22	23	19	1.37	1.12	0.87	0.80	0.88	0.73
National	**271**	**283**	**233**	**312**	**325**	**330**	**0.67**	**0.69**	**0.55**	**0.75**	**0.79**	**0.81**

(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 (35–39 and 40–44). 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 2014 to December 2019

	Diagnostic tests per 100 low-risk screens					
	2014	**2015**	**2016**	**2017**	**2018**	**2019**
Age at screen (years)						
Under 20	0.44	0.33	0.34	0.81	0.81	0.71
20–24	0.37	0.35	0.43	0.68	0.71	0.67
25–29	0.49	0.52	0.50	0.65	0.60	0.66
30–34	0.53	0.60	0.54	0.67	0.84	0.81
35–39	0.98	1.11	0.66	0.99	0.96	1.17
40–44	3.92	3.04	1.33	1.67	1.70	1.83
45 and over	0.00	2.13	3.28	1.61	2.08	0.00
Ethnicity						
Māori	0.46	0.46	0.50	0.65	0.74	0.68
Pacific	0.28	0.48	0.35	0.75	0.79	0.83
Asian	0.58	0.80	0.54	0.89	0.76	0.86
Other	0.78	0.72	0.58	0.73	0.80	0.83
National	**0.67**	**0.69**	**0.55**	**0.75**	**0.79**	**0.81**

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 2017–2019.

Table 28: Diagnostic testing volumes for women with low-risk screens by risk level, aggregated 2017–2019

Risk level	Number of diagnostic tests	Number of low-risk screens	Tests per 100 low-risk screens
1:301 to 1:500	63	2,521	2.50
1:501 to 1:1,000	106	6,388	1.66
1:1,001 to 1:2,000	95	9,917	0.96
1:2,001 to 1:3,000	95	8,109	1.17
1:3,001 to 1:4,000	50	7,046	0.71
1:4,001 to 1:5,000	41	6,043	0.68
1:5,001 to 1:10,000	142	22,631	0.63
1:10,001 to 1:100,000	375	60,941	0.62

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 2019 year, 174 diagnostic tests were completed for unscreened women. This is slightly higher than 2017 and 2018 (107 and 156 tests respectively) but lower than the number of tests undertaken in previous years (2014–2016). 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 2014 to December 2019

DHB	Number of diagnostic tests					
	2014	2015	2016	2017	2018	2019
Northland	7	8	6	S	S	S
Waitematā	22	22	19	14	24	23
Auckland	25	18	23	10	13	26
Counties Manukau	21	18	21	11	10	23
Waikato	14	15	16	6	12	12
Lakes	6	8	S	S	7	S
Bay of Plenty	12	14	10	S	S	6
Tairāwhiti	S	S	S	S	S	S
Hawke's Bay	7	7	8	S	S	S
Taranaki	S	11	S	S	7	S
MidCentral	11	8	9	S	6	S
Whanganui	S	S	S	S	S	S
Capital & Coast	30	36	25	12	8	16
Hutt Valley	11	22	10	6	6	8
Wairarapa	S	S	S	S	S	S
Nelson Marlborough	S	6	S	S	S	S
West Coast	S	S	S	S	S	S
Canterbury	37	30	30	18	31	25
South Canterbury	S	S	S	S	S	S
Southern	13	19	14	S	11	7
National	**235**	**252**	**212**	**107**	**156**	**174**

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

Table 30: Diagnostic testing volumes for unscreened women by age and ethnicity, January 2014 to December 2019

	Number of diagnostic tests					
	2014	**2015**	**2016**	**2017**	**2018**	**2019**
Age at screen (years)						
Under 20	10	16	12	4	4	4
20–24	29	19	17	12	18	19
25–29	39	53	36	27	29	30
30–34	66	70	60	26	47	56
35–39	54	54	56	22	45	48
40–44	34	35	28	15	13	15
45 and over	3	5	3	1	0	2
Ethnicity						
Māori	31	44	32	14	32	18
Pacific	20	21	11	11	7	11
Asian	29	33	36	17	19	35
Other	155	154	133	65	98	110
National	**235**	**252**	**212**	**107**	**156**	**174**

Diagnostic results for unscreened women

A breakdown of prenatal diagnostic testing results for unscreened women for the 2019 year is given in Table 31. Of the 174 diagnostic tests in 2019 for unscreened women, 128 (74%) had a normal karyotype.

Table 31: Diagnostic testing results for unscreened women, January to December 2019

Karyotype result	Number	Percentage
Normal karyotype	128	73.6
Trisomy 21	9	5.2
Trisomy 18	7	4.0
Trisomy 13	2	1.1
Turner syndrome	5	2.9
Triploidy	17	9.8
Other chromosomal abnormality	6	3.4
Total	**174**	**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, for 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 2019 was 0.06, lower than previous years (see Table 32). A value of 0.06 means that if a woman receives an increased-risk result for trisomy 21, 18 or 13, there is a 6 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 2014 to December 2019

Year	True positives	False positives	PPV	95% confidence interval
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)
2019	113	1,651	0.064	(0.053, 0.075)

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21, the PPV for 2019 was the same as 2018 at 0.05 (see Table 33), however this is lower than in the previous years (2014–2017). 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 2014 to December 2019

Year	True positives	False positives	PPV	95% confidence interval
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)
2019	86	1,632	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 2019 was higher than the PPV for trisomy 21 at 0.10 (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 2014 to December 2019

Year	True positives	False positives	PPV	95% confidence interval
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)
2019	23	207	0.100	(0.061, 0.139)

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 2017–2019. Women that received an increased-risk result of 1:5 to 1:20 for trisomy 21 had a 28 percent 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 at 5 percent probability, and lower again for women with increased-risk results of 1:51 to 1:300 at 1 percent probability.

Table 35: Positive predictive value of screening for trisomy 21 by risk level, aggregated 2017–2019

Risk level	True positives	False positives	PPV
1:5 to 1:20	183	463	0.28
1:21 to 1:50	24	480	0.05
1:51 to 1:300	44	3,502	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 2017–2019 by age and ethnicity.

The PPV of screening for trisomy 21 varied by ethnicity. Women of Other ethnicity had the highest PPV (0.07 or 7%), and Pacific women had the lowest PPV (0.02 or 2%). The PPV also varied by age group.

Table 36: Positive predictive value of screening for trisomy 21 by age and ethnicity, aggregated 2017–2019

	True positives	False positives	PPV
Age at screen (years)			
Under 20	1	47	0.02
20–24	12	209	0.05
25–29	26	537	0.05
30–34	56	1,190	0.04
35–39	92	1,557	0.06
40–44	60	856	0.07
45 and over	4	49	0.08
Ethnicity			
Māori	31	560	0.05
Pacific	7	409	0.02
Asian	45	1,283	0.03
Other	168	2,193	0.07
Total	**251**	**4,445**	**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 2019 was 0.04 (or 4%), which is the same as 2018. 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 2014 to December 2019

Year	False positives	True negatives	False positive rate	95% confidence interval
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)
2019	1,651	40,490	0.04	(0.037, 0.041)

As shown in Table 38, the false positive rate was higher for second trimester screens (5%) than for first trimester screens (4%), consistent with previous years.

Table 38: False positive rate for trisomy 21, 18 or 13 by trimester of screen, January to December 2019

Trimester	False positives	True negatives	False positive rate	95% confidence interval
T1 screens	1,339	34,436	0.037	(0.035, 0.039)
T2 screens	312	6,054	0.049	(0.044, 0.054)
Total	**1,651**	**40,490**	**0.039**	**(0.037, 0.041)**

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 2019, see Table 40).

Table 39: False positive rate for trisomy 21, January 2014 to December 2019

Year	False positives	True negatives	False positive rate	95% confidence interval
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)
2019	1,632	40,548	0.04	(0.037, 0.041)

Table 40: False positive rate for trisomy 18 and 13, January 2014 to December 2019

Year	False positives	True negatives	False positive rate	95% confidence interval
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)
2019	207	41,993	0.005	(0.004, 0.006)

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 2019 was 0.01 (1%) compared with 0.30 (30%) for women 40-44 years. 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.

The false positive rate for 2019 varied across ethnic groups from 0.03 (3%) for Māori and Other to 0.05 (5%) for Pacific and Asian.

Table 41: False positive rate for trisomy 21 by age and ethnicity, January 2014 to December 2019

	2014	2015	2016	2017	2018	2019
Age at screen (years)						
Under 20	0.01	0.01	0.01	0.02	0.01	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	0.03	0.03
35–39	0.05	0.05	0.05	0.05	0.08	0.09
40–44	0.15	0.19	0.15	0.17	0.26	0.30
45 and over	0.32	0.27	0.21	0.17	0.31	0.25
Ethnicity						
Māori	0.03	0.02	0.02	0.02	0.03	0.03
Pacific	0.04	0.04	0.04	0.04	0.05	0.05
Asian	0.03	0.03	0.03	0.03	0.05	0.05
Other	0.02	0.02	0.02	0.02	0.03	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 of screening

The overall detection rate for trisomy 21, 18 and 13 for the six years ending 2019 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 2019 was 0.83 (83%). A detection rate of 0.83 means that there is an 83 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 2014 to December 2019

Year	True positives	False negatives	Detection rate	95% confidence interval
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)
2019	113	23	0.83	(0.768, 0.894)

The detection rate for trisomy 21 alone is shown in Table 43. The rate for 2019 was higher (0.89) than the overall rate for trisomy 21, 18 and 13 (0.83). The detection rate for trisomy 13 and 18 was lower at 0.59 (Table 44).

Table 43: Detection rate for trisomy 21, January 2014 to December 2019

Year	True positives	False negatives	Detection rate	95% confidence interval
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)
2019	86	11	0.89	(0.823, 0.950)

Table 44: Detection rate for trisomy 13 or 18, January 2014 to December 2019

Year	True positives	False negatives	Detection rate	95% confidence interval
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)
2019	23	16	0.59	(0.435, 0.744)

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 a 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, 2014–2019

DHB	2014	2015	2016	2017	2018	2019
Northland	2,097	2,136	2,267	2,238	2,198	2,310
Waitematā	7,842	7,562	7,935	7,723	7,428	7,777
Auckland	6,301	5,898	5,902	5,626	5,429	5,592
Counties Manukau	8,282	8,195	8,235	8,276	8,154	8,393
Waikato	5,245	5,272	5,357	5,317	5,382	5,450
Lakes	1,391	1,511	1,548	1,555	1,525	1,535
Bay of Plenty	2,783	2,791	2,898	3,102	3,009	3,107
Tairāwhiti	686	737	776	704	702	681
Hawke's Bay	2,066	2,000	2,060	2,129	2,112	2,018
Taranaki	1,518	1,516	1,434	1,403	1,565	1,512
MidCentral	2,091	2,111	2,080	2,130	2,156	2,164
Whanganui	819	814	801	845	810	864
Capital & Coast	3,528	3,533	3,458	3,499	3,201	3,180
Hutt Valley	1,852	1,966	1,969	1,948	1,941	1,961
Wairarapa	473	463	462	536	497	518
Nelson Marlborough	1,420	1,417	1,547	1,426	1,501	1,451
West Coast	350	357	320	359	323	342
Canterbury	5,991	6,210	6,301	6,397	6,256	6,438
South Canterbury	650	659	651	634	601	623

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

Southern	3,285	3,414	3,316	3,435	3,271	3,440
Total	**58,670**	**58,562**	**59,318**	**59,282**	**58,061**	**59,356**

Table 47: Live births and still births by age group, 2014–2019

Age group (years)	2014	2015	2016	2017	2018	2019
<20	2,992	2,783	2,443	2,297	2,130	2,090
20–24	10,268	9,943	9,586	9,319	8,685	8,534
25–29	15,688	15,717	16,540	16,622	16,261	16,391
30–34	17,555	17,904	18,374	18,693	18,702	19,529
35–39	9,680	9,767	9,961	9,876	10,020	10,404
40–44	2,343	2,295	2,274	2,312	2,094	2,266
45+	131	140	127	153	162	138
Unknown	13	13	13	10	7	4
Total	**58,670**	**58,562**	**59,318**	**59,282**	**58,061**	**59,356**

Table 48: Live births and still births by ethnicity, 2014–2019

Ethnicity	2014	2015	2016	2017	2018	2019
Māori	14,500	14,791	14,983	14,927	14,578	14,814
Pacific	6,188	6,072	5,856	5,967	5,969	6,149
Asian	9,189	9,205	10,511	10,556	10,584,	11,470
Other	28,793	28,494	27,968	27,832	26,930	26,923
Total	**58,670**	**58,562**	**59,318**	**59,282**	**58,061**	**59,356**

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

Summary of prenatal diagnostic testing uptake and results for 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 2019, 685 (39%) had a prenatal diagnostic test (CVS or amniocentesis) and 1,079 (61%) did not. Table 49 shows the diagnostic testing results for the 685 prenatal tests, of which 130 had an abnormal karyotype, including 84 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 2019 year

Karyotype result	Number	Percentage
Normal karyotype	555	81.0
Confirmed Down syndrome	84	12.3
Other result	46	6.7
Total	**685**	**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 the 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 173 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 50 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 0.1 percent or less of all negative screens for each of the years from 2014 to 2019.

Table 50: False negative screens for trisomy 21, 18 and 13 by risk level, January 2014 to December 2019

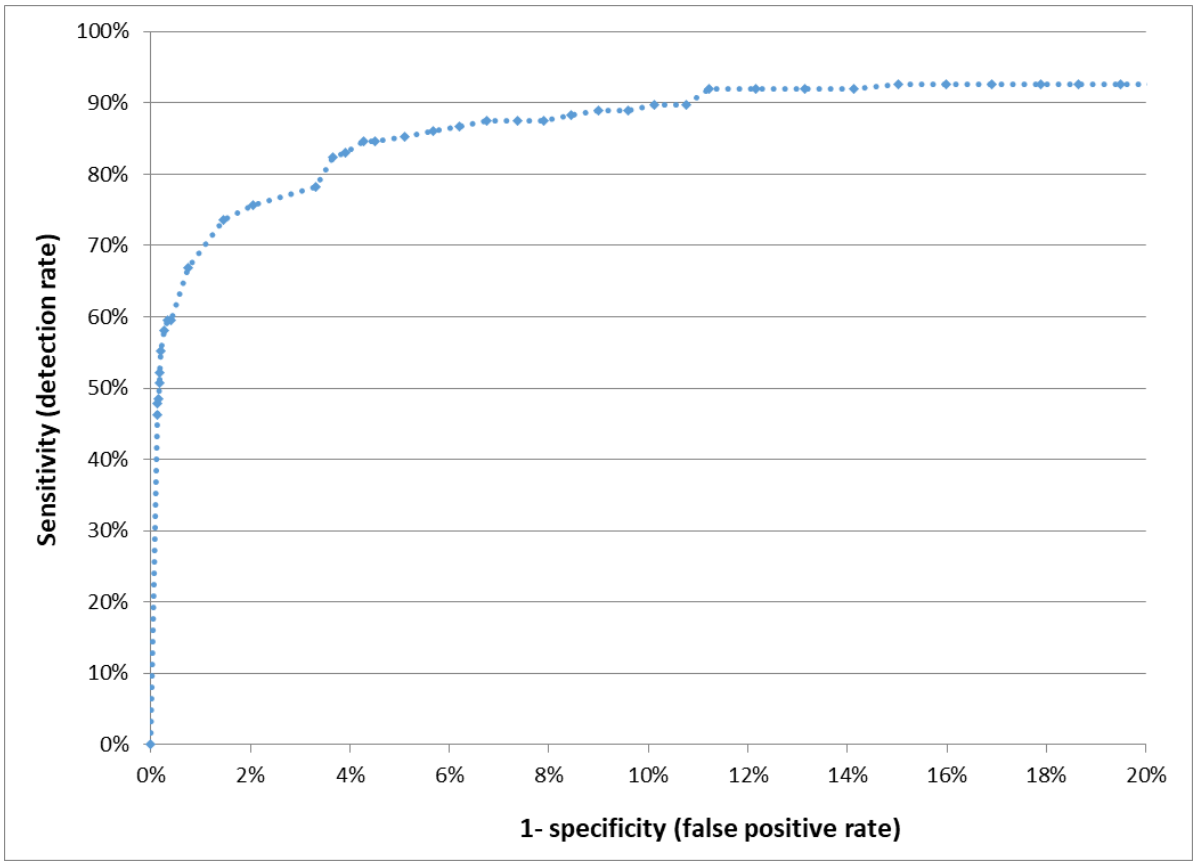
Risk level	False negatives						% of negative screens that are false negatives					
	2014	2015	2016	2017	2018	2019	2014	2015	2016	2017	2018	2019
1:301 to 1:500	6	4	8	7	5	5	0.94	0.63	1.25	1.21	0.51	0.51
1:501 to 1:1,000	5	10	7	8	12	7	0.31	0.58	0.46	0.52	0.51	0.28
1:1,001 to 1:2,000	4	4	3	8	2	1	0.14	0.14	0.11	0.33	0.05	0.03
1:2,001 to 1:3,000	5	2	6	3	4	6	0.20	0.08	0.25	0.14	0.14	0.19
1:3,001 to 1:4,000	0	1	0	2	0	1	0.00	0.04	0.00	0.11	0.00	0.04
1:4,001 to 1:5,000	2	0	0	0	2	0	0.10	0.00	0.00	0.00	0.09	0.00
1:5,001 to 1:10,000	2	3	2	2	3	2	0.02	0.03	0.02	0.03	0.04	0.02
Less than 1:10,000	3	1	4	5	5	1	0.01	0.00	0.02	0.02	0.03	0.01
Total	**27**	**25**	**30**	**35**	**33**	**23**	**0.07**	**0.06**	**0.07**	**0.08**	**0.08**	**0.06**

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 2019 was 83 percent, and the false positive rate was 3.9 percent. 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 3.9 percent to 5.0 percent. This occurs at a risk cut-off of 1:400.

Figure 11: ROC curve for trisomy 21, 18 and 13 screening 2019



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)[4] 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.

[4] Chance of neural tube defects is no longer reported by the screening laboratory (from March 2023).

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 – an assessment of nasal bone was included in the risk calculation algorithm if it was reported at the same time as the NT assessment. Note that since March 2018 nasal bone assessment 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**