The Ministry of Health
New Zealand Government

National Screening Unit

Antenatal Screening for Down Syndrome and Other Conditions

2020 Monitoring Report

Released 2023

New Zealand Government

Citation: National Screening Unit. 2023. *Antenatal Screening for Down Syndrome and Other Conditions: 2020 Monitoring Report.* Wellington: National Screening Unit.

Published in November 2023 by the National Screening Unit
PO Box 5013, Wellington 6140, New Zealand

ISBN 978-1-99-106771-5 (online)

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 2015 to 31 December 2020 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 2020

- Screening was commenced for 86 percent of women who gave birth in 2020.
- There has been a steady increase in trimester two screens (both commenced and completed) since 2015.
- The national screening completion rate was 75 percent in 2020 (range 71–75 percent between 2015 and 2020). First trimester screens made up around 85 percent of all completed screens in 2020.
- Completion rates for Māori and Pacific have increased since 2015 but remain much lower in 2020 compared to Asian and Other women. Screening completion decreased with increasing deprivation in 2020.
- In 2020, while the number of women screened did not appear to be affected by COVID-19 restrictions, the reduced availability of nuchal translucency (NT) scans

during the national lockdowns may have caused a slightly higher number of incomplete screens in March and April.[1]

- Thirteen percent of screens commenced in 2020 were not completed and nearly all were screens commenced in the first trimester. This is an increase from 10 percent in 2015.
- The overall positive test rate (number of increased-risk results per 100 screens) for trisomy 21, 18 and 13 was 4.2 in 2020, which is the same as 2019 but an increase from 2.8 in 2015. The positive test rate was higher for second trimester screens (5.3 per 100 screens) than for first trimester screens (4.0 per 100 screens) for 2020.
- Diagnostic testing volumes following an increased-risk screen decreased from 56 percent in 2015 to 30 percent in 2020 (diagnostic tests per 100 increased-risk screens).
- The overall false positive rate for trisomy 21, 18 and 13 was 4 percent in 2020, the same as in 2018 and 2019 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 82 percent in 2020 (range 75–84 percent between 2015 and 2020).
- 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 non-invasive prenatal screening (NIPS). The information presented in this report will have been influenced by use of NIPS, but the impact cannot be quantified.

[1] Antenatal Screening for Down Syndrome and Other Conditions in Covid-19 time January to June 2020: report from LabPLUS and CHL

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 Te Toka Tumai Auckland (for samples from Taupō and north of Taupō) and Canterbury Health Laboratories at Waitaha Canterbury (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.

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. In 2020, some women who received an increased-risk screening result were offered NIPS from Maternal Fetal Medicine services. NIPS was offered, where possible, as an alternative to invasive diagnostic testing (COVID-19 initiative). Use of NIPS is increasing in New Zealand but as the tests are mostly done privately, there is limited 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.

During 2020, antenatal screening was considered an essential service and continued through the Level 3 and 4 national lockdowns. The data suggests that screening volumes were not affected by COVID-19 restrictions, but the reduced availability of NT scans may have caused a slightly higher number of incomplete screens in March and April 2020.[2]

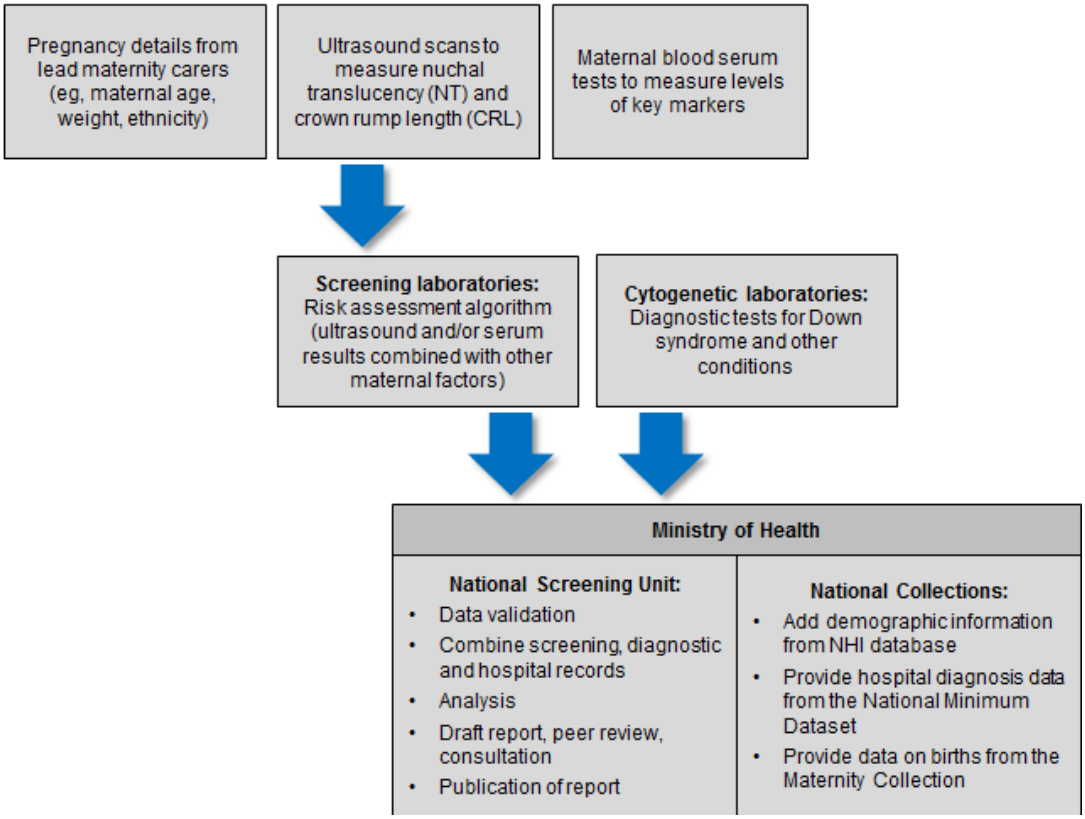
Programme monitoring and data collection

This report presents data on antenatal screening for Down syndrome and other conditions for the six calendar years from 1 January 2015 to 31 December 2020 and is based on screens that commenced during that time. The definitions for the 11 indicators in this

[2] Antenatal Screening for Down Syndrome and Other Conditions in Covid-19 time January to June 2020: report from LabPLUS and CHL

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

The screening and diagnostic data was matched with hospital discharge data, sourced from the National Minimum Data Set (NMDS), held by Te Whatu Ora. 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.[3]

Inclusion criteria

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

- Screening commencement date between 1 January 2015 and 31 December 2020 (ie, date of the first test the woman had as part of the screening pathway).

[3] 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

Figures have been broken down by NZ Deprivation Index Quintiles. Quintiles are aggregations of two NZ deprivation deciles. Deprivation is derived by obtaining the domicile code of the mother at time of screen and matching that to the NZ Deprivation Decile 2018 index.

Births

Data on the number of live and still births[4] was obtained from the National Maternity Collection for each calendar year. Appendix 2 contains tables for the denominators used in this report.

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

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.

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 2017–2020 data but not for years prior to this. Caution is therefore required when comparing data for 2015–2016 with 2017–2020. Where a six-year rate would ordinarily have been applied, a decision has been made to supply a four-year rate (2017–2020) where this does not compromise privacy. An additional improvement was made for the 2020 report to better identify women who had a diagnostic test but no prenatal screening test (indicator 8).

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 2020, 27 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, ethnicity and deprivation.

Total screens commenced by trimester

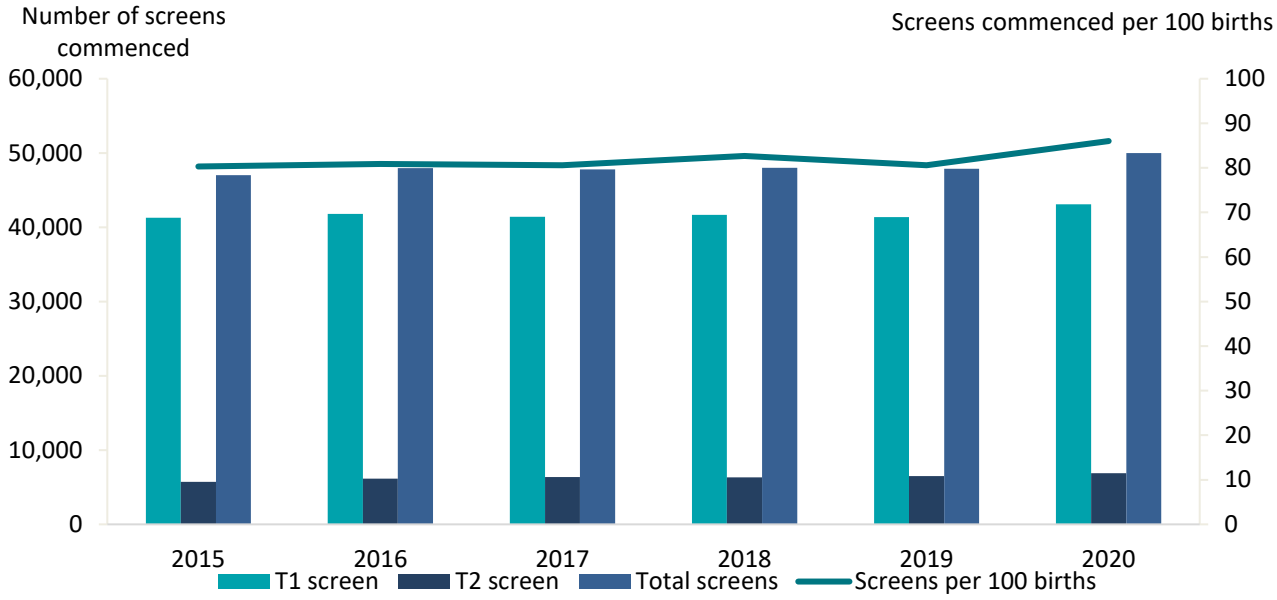
During 2020, a total of 49,990 screens were commenced, a rate of 86 per 100 births. Table 1 shows the total number of screens commenced by year and trimester of screen. Throughout the report, T1 is used to refer to the first trimester and T2 to the second trimester.

The majority of screens were T1 screens. The rate of screens commenced per 100 births has stayed largely flat over time, from 80.3 in 2015 to 80.6 in 2019, but has noticeably jumped to 86 in 2020 (see Table 1 and Figure 2).

Table 1: Total screens commenced by trimester, January 2015 to December 2020

Trimester of screen	Number and rate of screens commenced					
	2015	2016	2017	2018	2019	2020
T1 screen	41,283	41,816	41,403	41,681	41,365	43,102
T2 screen	5,742	6,152	6,369	6,330	6,503	6,888
Total screens	**47,025**	**47,968**	**47,772**	**48,011**	**47,868**	**49,990**
Screens per 100 births	80.3	80.9	80.6	82.7	80.6	86.0

Figure 2: Number and rate of screens commenced, January 2015 to December 2020



Screens commenced by DHB

Figure 3 shows the screening commencement rates by DHB for 2020. There was a large variation in rates, from 64 screens commenced per 100 births in Northland to over 100 screens commenced per 100 births in Canterbury and Nelson Marlborough. Three-quarters (75%) of all DHBs had rates of above 80 per 100 births in 2020, compared to only half of DHBs in 2019. Table 2 gives a full breakdown by the trimester of the screen.

Figure 3: Screens commenced by DHB, January to December 2020

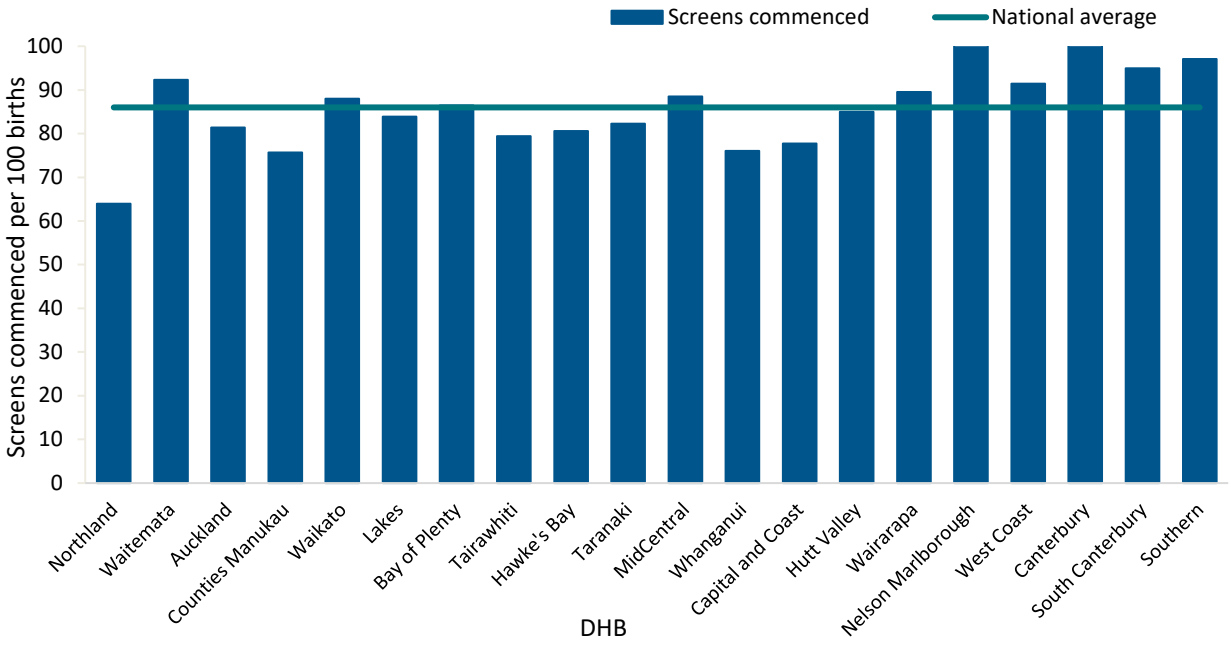


Table 2: Screens commenced by trimester and DHB, January to December 2020

DHB	Number of screens commenced			Screens commenced (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total**
Northland	1,257	265	1,522	52.8	11.1	63.9
Waitematā	6,109	788	6,897	81.8	10.5	92.3
Auckland	3,598	599	4,197	69.8	11.6	81.4
Counties Manukau	4,909	1,448	6,357	58.4	17.2	75.7
Waikato	4,320	583	4,903	77.6	10.5	88.0
Lakes	1,027	176	1,203	71.6	12.3	83.9
Bay of Plenty	2,440	266	2,706	78.0	8.5	86.5
Tairāwhiti	470	93	563	66.2	13.1	79.4
Hawke's Bay	1,425	246	1,671	68.7	11.9	80.6
Taranaki	1,061	137	1,198	72.9	9.4	82.3
MidCentral	1,685	216	1,901	78.4	10.0	88.5
Whanganui	467	155	622	57.0	18.9	76.0
Capital & Coast	2,150	254	2,404	69.6	8.2	77.7
Hutt Valley	1,438	265	1,703	71.7	13.2	85.0
Wairarapa	397	75	472	75.6	14.3	89.6
Nelson Marlborough	1,306	144	1,450	92.3	10.2	102.3
West Coast	231	37	268	78.3	12.5	91.5
Canterbury	5,455	731	6,186	88.3	11.8	100.2
South Canterbury	465	96	561	78.8	16.3	94.9
Southern	2,870	309	3,179	87.6	9.4	97.1
National*	**43,102**	**6,888**	**49,990**	**74.2**	**11.9**	**86.0**

*DHB counts do not sum to National total and **screen rates may exceed 100% due to a lag in maternity data collection.

Most DHBs showed an increase in their rate of screens commenced between 2015 and 2020. All but one DHB showed an increase in the rate of screens commenced between 2019 and 2020 (see Table 3).

Table 3: Screens commenced per 100 births by DHB, January 2015 to December 2020

DHB	Screens commenced (per 100 births)*					
	2015	2016	2017	2018	2019	2020
Northland	60.1	58.6	64.2	61.7	62.1	63.9
Waitematā	88.4	87.1	86.7	91.4	86.0	92.3
Auckland	85.7	82.0	75.8	82.2	78.3	81.4
Counties Manukau	71.1	71.0	70.6	71.1	70.0	75.7
Waikato	81.8	83.7	85.5	84.0	85.1	88.0
Lakes	74.3	76.7	73.6	80.9	74.7	83.9
Bay of Plenty	77.6	81.1	82.2	82.9	84.1	86.5
Tairāwhiti	68.3	63.6	70.2	78.1	70.6	79.4
Hawke's Bay	72.6	76.2	71.8	75.6	76.6	80.6
Taranaki	74.9	67.8	72.7	74.7	71.0	82.3
MidCentral	63.9	73.1	79.9	74.7	78.3	88.5
Whanganui	70.5	74.1	71.8	77.8	75.0	76.0
Capital & Coast	83.4	86.3	76.1	81.4	77.6	77.7
Hutt Valley	78.7	82.2	76.3	84.0	80.6	85.0
Wairarapa	83.8	89.0	90.1	92.7	94.2	89.6
Nelson Marlborough	96.0	85.1	98.6	91.5	95.5	102.3
West Coast	82.4	86.5	84.4	84.6	84.5	91.5
Canterbury	89.4	91.5	92.4	94.3	91.3	100.2
South Canterbury	86.4	87.5	94.0	94.7	88.4	94.9
Southern	85.1	87.8	89.0	90.2	87.0	97.1
National average	**80.3**	**80.9**	**80.6**	**82.7**	**80.6**	**86.0**

*Screen rates may exceed 100% due to a lag in maternity data collection.

Screens commenced by age, ethnicity and deprivation

Table 4 provides an overall view of screens commenced by age and ethnicity for the period from January 2015 to December 2020.

The rate of screens commenced for age groups up to 34 years have increased since 2015 while conversely the rate of screens commenced for age groups of 35 and over have decreased since 2015. The 25–29 years age group had the highest rate of screens commenced for 2020, with a rate of 93 women commencing screening per 100 births (see Figure 4). Screening commencement rates for women aged 45 and over dropped from 62 screens commenced per 100 births in 2019 to 41 per 100 births in 2020, the lowest for that age group in any year from 2015 to 2020. Low volumes in this age group may be contributing to the variation in rates.

Differences in screening commencement rates by ethnicity have continued in 2020. Women of Other ethnicity had the highest rate (100 of 100 births), followed by Asian women (97 of 100 births). The rate of commenced screens for Pacific and Māori women was lower at 58 per 100 births and 56 per 100 births respectively (see Figure 5). All groups have shown increasing rates over the reporting period, particularly for Māori, with an increase of 13 percentage points from 43 percent in 2015 to 56 percent in 2020. However, this rate is still well below the national rate of 86 per 100 births in 2020.

Table 4: Screens commenced by age and ethnicity of mother, January 2015 to December 2020

	Number of screens commenced						Screens commenced (per 100 births)**					
	2015	**2016**	**2017**	**2018**	**2019**	**2020**	**2015**	**2016**	**2017**	**2018**	**2019**	**2020**
Age at screen (years)												
Under 20	1,925	1,829	1,683	1,546	1,565	1,441	69.1	74.9	73.3	72.7	74.9	73.0
20–24	7,109	7,000	6,899	6,475	6,341	6,407	71.5	73.0	74.0	74.5	74.3	77.7
25–29	13,189	13,943	14,037	14,162	13,882	14,681	84.0	84.3	84.4	87.1	84.7	93.3
30–34	15,124	15,732	15,804	16,171	16,605	17,855	84.5	85.6	84.5	86.4	85.0	90.8
35–39	8,007	7,781	7,659	8,091	7,973	8,169	82.0	78.1	77.5	80.8	76.6	79.6
40–44	1,593	1,574	1,587	1,476	1,416	1,372	69.3	69.2	68.6	70.5	62.5	65.7
45 and over	78	109	103	90	86	65	56.1	86.5	67.8	55.6	62.3	41.4
Ethnicity												
Māori	6,256	7,176	7,754	7,675	7,844	8,388	42.9	48.7	52.0	52.7	52.9	55.9
Pacific	3,120	3,089	3,284	3,206	3,380	3,494	51.5	52.9	55.0	53.7	55.0	57.9
Asian	8,695	9,851	9,720	10,330	10,554	11,039	94.4	93.6	92.0	97.5	92.0	97.3
Other	28,954	27,852	27,005	26,796	26,090	27,069	100.9	98.7	97.0	99.5	96.9	105.2
National*	**47,025**	**47,968**	**47,772**	**48,011**	**47,868**	**49,990**	**80.3**	**80.9**	**80.6**	**82.7**	**80.6**	**86.0**

*Ethnic group counts do not sum to National total.

**Screen rates may exceed 100% due to a lag in maternity data collection.

Figure 4: Screens commenced by age of mother at screen, January to December 2020

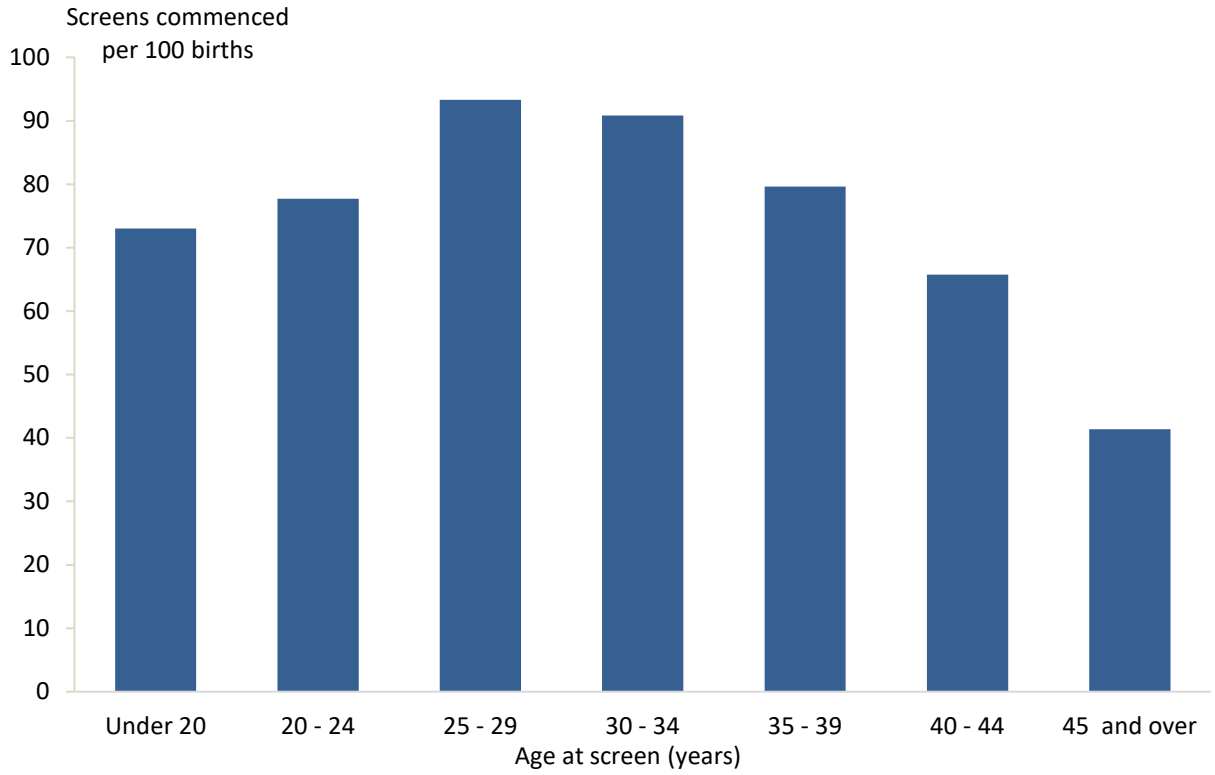
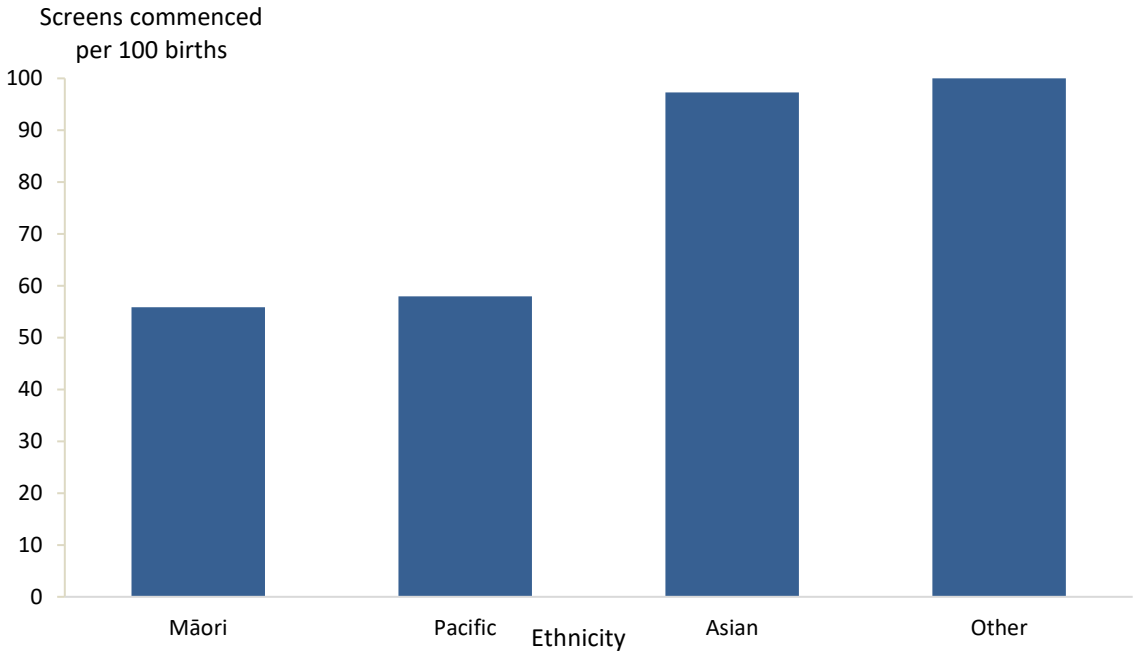


Figure 5: Screens commenced by ethnicity of mother, January to December 2020

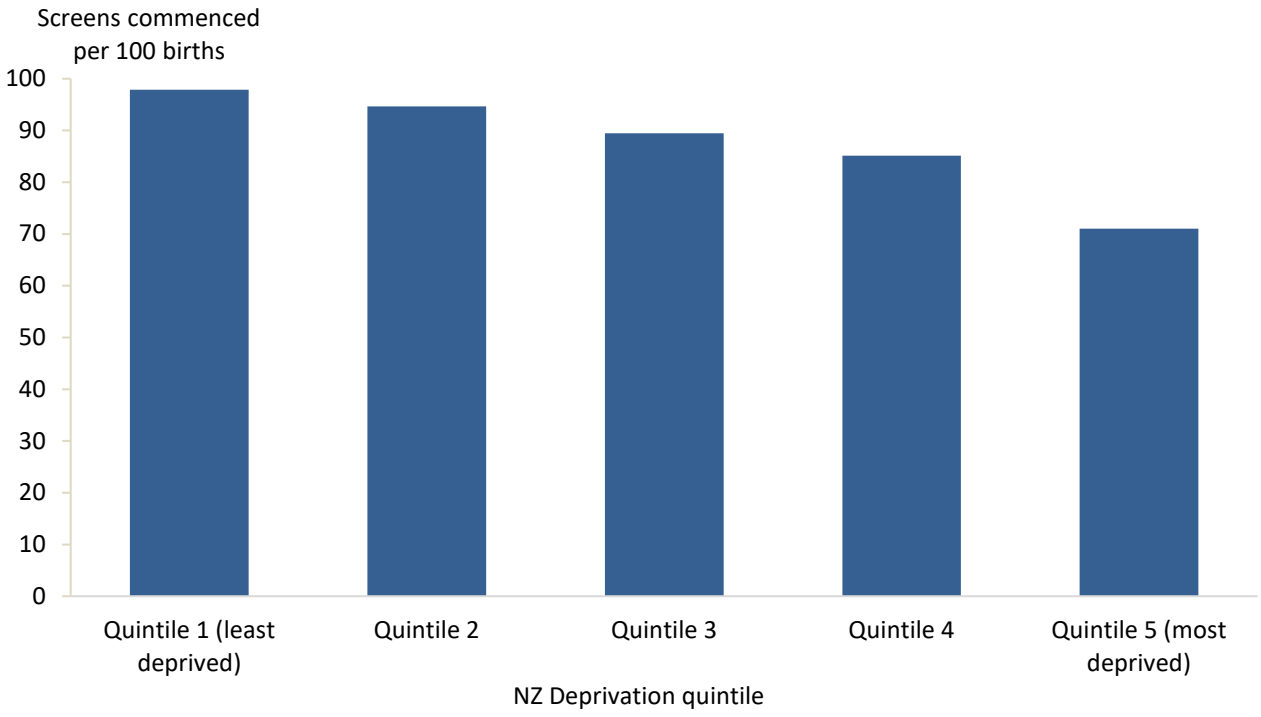


A trend of higher screening commencement rates for women in less deprived areas was evident, with 98 women per 100 per births starting screening for quintile 1 women in 2020 compared with 71 per 100 births for quintile 5 (see Table 5 and Figure 6).

Table 5: Screens commenced by deprivation quintile of mother, January to December 2020

NZ Deprivation quintile	Number of screens commenced	Screens commenced (per 100 births)
Quintile 1 (least deprived)	8,941	97.9
Quintile 2	9,475	94.7
Quintile 3	9,637	89.5
Quintile 4	11,398	85.2
Quintile 5 (most deprived)	10,510	71.0
Unknown	29	-
National	**49,990**	**86.0**

Figure 6: Screens commenced by deprivation quintile of mother, January to December 2020



Indicator 2: Screens completed

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

Total screens completed by trimester

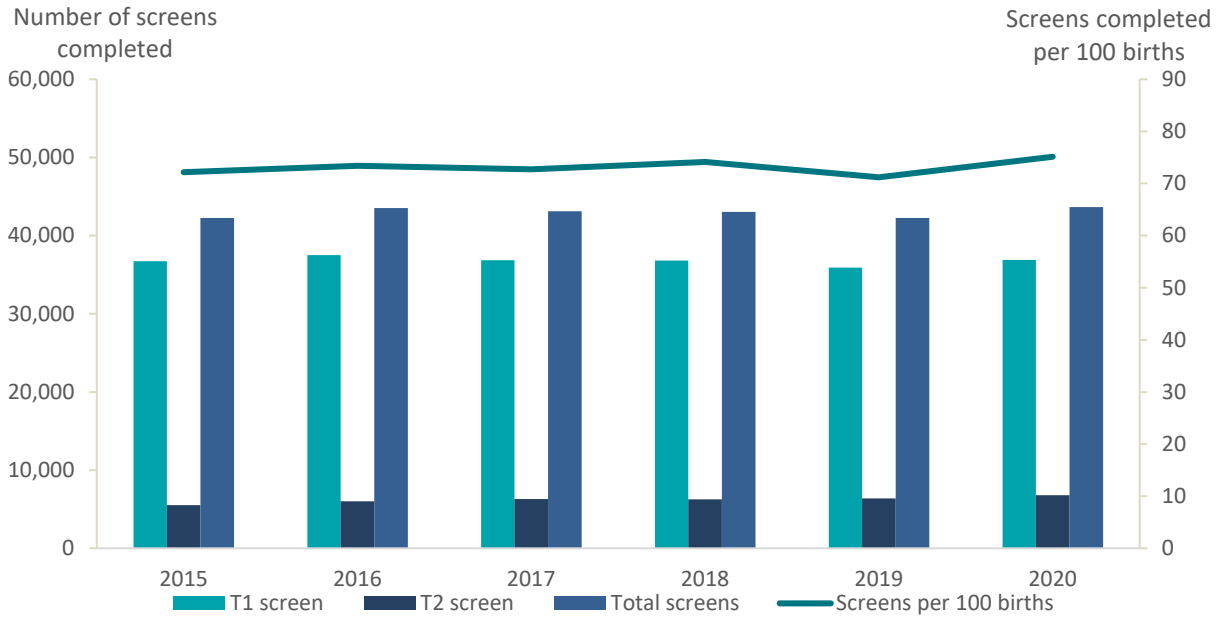
During 2020, a total of 43,669 screens were completed, at a rate of 75 screens per 100 births.

Table 6 and Figure 7 show the total number of screens completed per year and trimester of screen. Across all years, the majority of screens were completed in the first trimester. The rate of completed screens increased from 72 per 100 births in 2015 to 75 per 100 births in 2020.

Table 6: Total screens completed by trimester, January 2015 to December 2020

Trimester of screen	Number and rate of screens completed					
	2015	**2016**	**2017**	**2018**	**2019**	**2020**
T1 screen	36,739	37,511	36,836	36,810	35,900	36,893
T2 screen	5,517	6,008	6,284	6,242	6,377	6,776
Total screens	42,256	43,519	43,120	43,052	42,277	43,669
Screens per 100 births	**72.2**	**73.4**	**72.7**	**74.2**	**71.2**	**75.1**

Figure 7: Number and rate of screens completed, January 2015 to December 2020



Screens completed by DHB

Screening completion rates for 2020 varied across DHBs, from 55 completed screens per 100 births in Northland to 92 per 100 births in Nelson Marlborough (see Figure 8). Table 7 gives a full breakdown by the trimester of screen.

Figure 8: Screens completed by DHB, January to December 2020

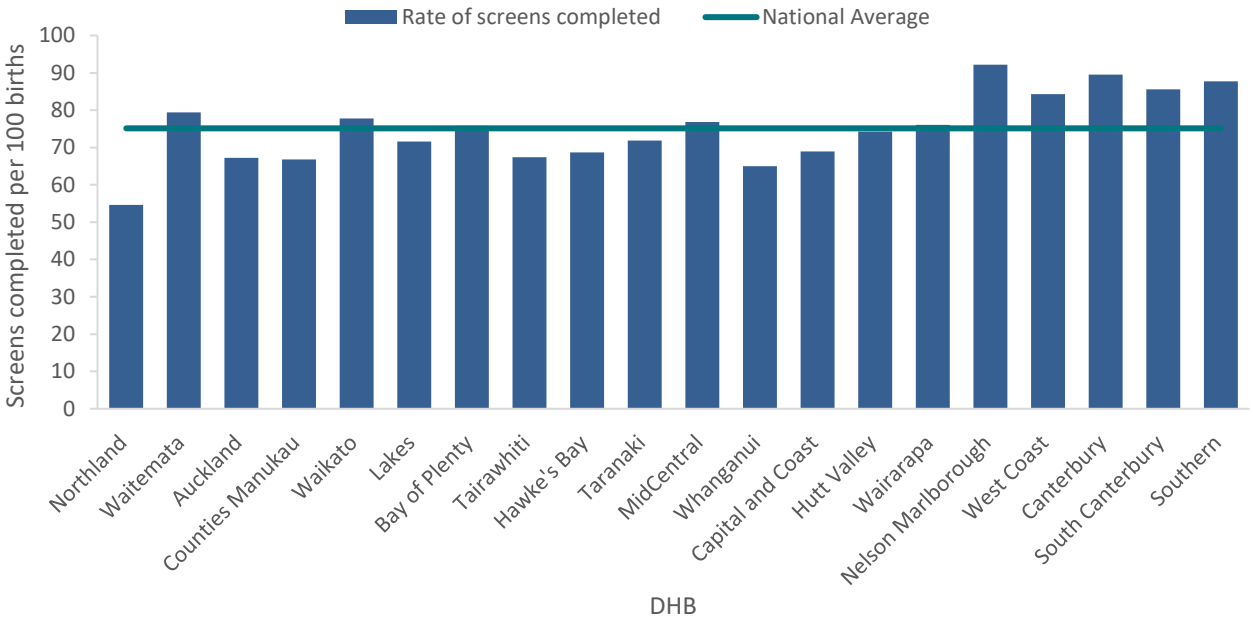


Table 7: Screening completion by trimester and DHB, January to December 2020

DHB	Number of screens completed			Screens completed (per 100 births)		
	First trimester	Second trimester	Total	First trimester	Second trimester	Total
Northland	1,041	260	1,301	43.7	10.9	54.6
Waitematā	5,155	775	5,930	69.0	10.4	79.4
Auckland	2,874	591	3,465	55.8	11.5	67.2
Counties Manukau	4,186	1,426	5,612	49.8	17.0	66.8
Waikato	3,764	568	4,332	67.6	10.2	77.7
Lakes	853	174	1,027	59.5	12.1	71.6
Bay of Plenty	2,110	263	2,373	67.4	8.4	75.8
Tairāwhiti	387	91	478	54.6	12.8	67.4
Hawke's Bay	1,180	244	1,424	56.9	11.8	68.7
Taranaki	912	134	1,046	62.6	9.2	71.8
MidCentral	1,437	214	1,651	66.9	10.0	76.9
Whanganui	381	151	532	46.6	18.5	65.0
Capital & Coast	1,884	247	2,131	60.9	8.0	68.9
Hutt Valley	1,229	259	1,488	61.3	12.9	74.3
Wairarapa	326	75	401	61.9	14.2	76.1
Nelson Marlborough	1,164	142	1,306	82.1	10.0	92.2
West Coast	211	36	247	72.0	12.3	84.3
Canterbury	4,800	726	5,526	77.7	11.8	89.5
South Canterbury	413	93	506	69.9	15.7	85.6
Southern	2,570	302	2,872	78.5	9.2	87.7
National*	**36,893**	**6,776**	**43,669**	**63.5**	**11.7**	**75.1**

*DHB counts do not sum to National total.

As shown in Table 8, many DHBs showed a trend of increasing rates of screening completion over the five years from 2015 to 2020. Furthermore, for the majority (85%) of DHBs, screening completion rates increased from 2019 to 2020.

Table 8: Screening completion by DHB, January 2015 to December 2020

DHB	Screens completed (per 100 births)					
	2015	2016	2017	2018	2019	2020
Northland	51.6	50.9	56.2	53.6	54.2	54.6
Waitematā	81.8	81.4	79.8	82.9	76.1	79.4
Auckland	79.1	75.6	68.6	72.2	66.1	67.2
Counties Manukau	64.5	65.5	64.4	64.9	62.7	66.8
Waikato	72.4	74.6	76.3	74.8	76.2	77.7
Lakes	65.7	67.8	65.7	71.2	67.4	71.6
Bay of Plenty	67.8	71.8	73.6	74.6	75.4	75.8
Tairāwhiti	53.8	51.1	59.1	65.1	59.8	67.4
Hawke's Bay	64.2	68.6	63.7	67.6	66.9	68.7
Taranaki	66.3	62.1	66.4	68.3	61.7	71.8
MidCentral	56.9	66.1	72.3	66.3	68.4	76.9
Whanganui	58.5	65.8	63.6	67.6	65.6	65.0
Capital & Coast	75.1	77.8	67.8	73.3	69.2	68.9
Hutt Valley	68.0	71.6	67.3	74.4	70.2	74.3
Wairarapa	72.8	77.9	80.6	81.0	81.1	76.1
Nelson Marlborough	84.7	77.4	90.1	84.6	86.8	92.2
West Coast	72.3	77.7	76.8	72.6	74.6	84.3
Canterbury	80.6	82.5	83.0	84.2	80.3	89.5
South Canterbury	79.8	81.5	85.4	88.2	80.1	85.6
Southern	77.9	81.1	81.7	82.5	78.1	87.7
National average	**72.2**	**73.4**	**72.7**	**74.2**	**71.2**	**75.1**

Screens completed by age, ethnicity and deprivation

Table 9 provides an overall view of screens completed by age and ethnicity for January 2015 to December 2020, with similar trends to screening commencement.

In 2020, screening completion rates were highest in the 25–29 age group (see Figure 9), with 83 women completing screening per 100 births, an increase of 6 screens completed per 100 births compared to 2019. The completion rate for women aged 45 and over fell to 29 completed screens per 100 births in 2020, mirroring the drop in screening commencement for this age group. Low volumes in this age group may be contributing to the variation in rates.

Screening completion rates were highest among women of Other ethnicity, at 93 per 100 births in 2020 (see Figure 10). This was followed by women of Asian ethnicity at 88 per 100 births. Screening completion rates have increased over time for Māori and Pacific women; however the rates remain lower than other groups at 45 per 100 births and 50 per 100 births respectively.

Factors that may contribute to the differences in screening completion rates between ethnic groups include inequitable offer of screening and barriers such as the cost and accessibility of the first trimester ultrasound scan. However, the lower rates for Māori and Pacific women could also be partly due to personal choice and cultural or religious views.[5],[6]

[5] Implementing NIPT into publicly funded antenatal screening services for Down syndrome and other conditions in Aoteaora New Zealand. BMC Pregnancy and Childbirth (2017) 17:344

[6] Inequity in timing of prenatal screening in New Zealand: Who are our most vulnerable? Aust NZ J Obstet Gynaecol (2017) 1-8

Table 9: Screens completed by age and ethnicity of mother, January 2015 to December 2020

	Number of screens completed						Screens completed (per 100 births)					
	2015	**2016**	**2017**	**2018**	**2019**	**2020**	**2015**	**2016**	**2017**	**2018**	**2019**	**2020**
Age at screen (years)												
Under 20	1,510	1,474	1,376	1,243	1,282	1,129	54.2	60.3	59.9	58.4	61.3	57.2
20–24	5,992	6,079	5,948	5,588	5,426	5,400	60.3	63.4	63.8	64.3	63.6	65.5
25–29	11,824	12,675	12,779	12,898	12,554	13,056	75.3	76.6	76.9	79.4	76.6	83.0
30–34	14,030	14,709	14,651	14,823	14,940	15,913	78.3	80.1	78.4	79.2	76.5	81.0
35–39	7,430	7,137	6,959	7,205	6,897	7,046	76.1	71.6	70.4	71.9	66.3	68.7
40–44	1,406	1,366	1,328	1,225	1,119	1,080	61.2	60.0	57.4	58.5	49.4	51.7
45 and over	64	79	79	70	59	45	46.0	62.7	52.0	43.2	42.8	28.7
Ethnicity												
Māori	4,911	5,924	6,442	6,387	6,513	6,804	33.7	40.2	43.2	43.8	44.0	45.3
Pacific	2,626	2,673	2,876	2,782	2,927	2,988	43.3	45.8	48.2	46.6	47.6	49.5
Asian	8,114	9,304	9,093	9,594	9,649	9,973	88.1	88.4	86.1	90.6	84.1	87.9
Other	26,605	25,618	24,701	24,287	23,188	23,904	92.7	90.8	88.7	90.2	86.1	92.9
National*	**42,256**	**43,519**	**43,120**	**43,052**	**42,277**	**43,669**	**72.2**	**73.4**	**72.7**	**74.2**	**71.2**	**75.1**

*Ethnic group counts do not sum to National total.

Figure 9: Screens completed by age of mother at screen, January to December 2020

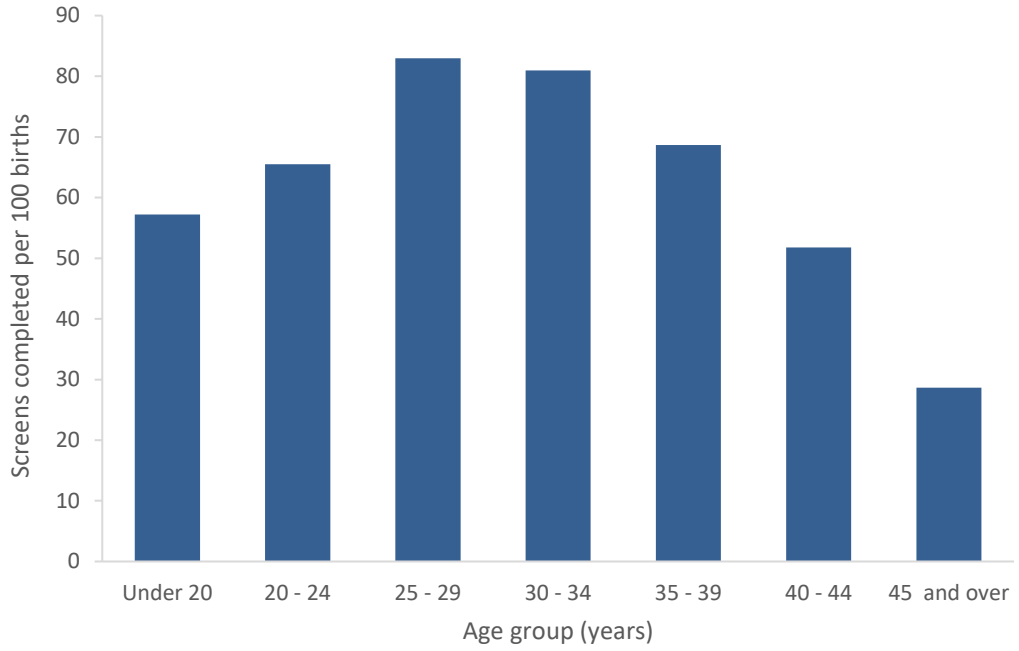
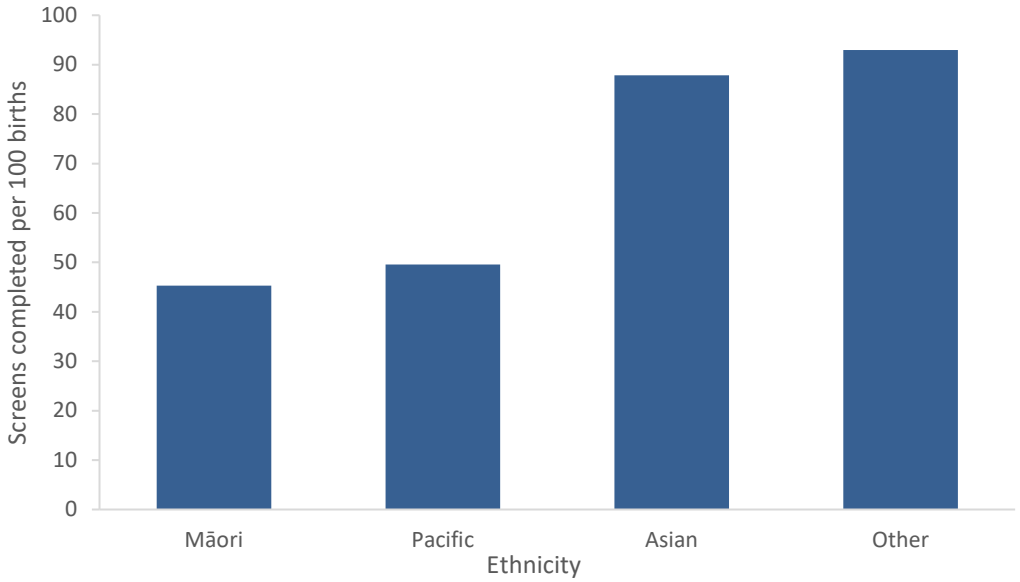


Figure 10: Screens completed by ethnicity of mother, January to December 2020

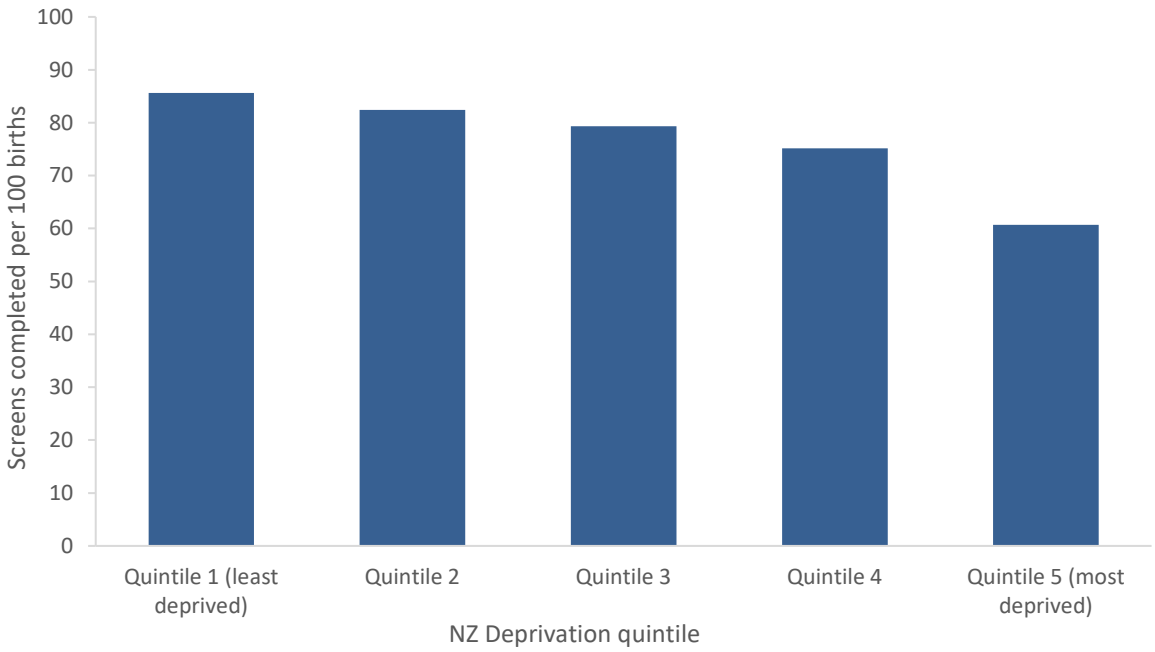


As shown in Table 10 and Figure 11, screening completion rates in 2020 were highest among women in less deprived areas with a rate of 86 per 100 births for quintile 1, compared with 61 per 100 births for quintile 5.

Table 10: Screens completed by deprivation quintile of mother, January to December 2020

NZ Deprivation quintile	Number of screens completed	Screens completed (per 100 births)
Quintile 1 (least deprived)	7,820	85.6
Quintile 2	8,248	82.4
Quintile 3	8,541	79.3
Quintile 4	10,058	75.1
Quintile 5 (most deprived)	8,979	60.7
Unknown	23	-
National	**43,669**	**75.1**

Figure 11: Screens completed by deprivation quintile of mother, January to December 2020



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 11 shows the number and proportion of second trimester screening results that included first trimester inputs over the period from 2015 to 2020. 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 (45.7% in 2020). PAPP-A was included in 13 percent of second trimester screens in 2020, up from 11.5 percent in 2019.

Table 11: Screening pathway variance by type, January 2015 to December 2020

Year	Second trimester screening results				
	Number			Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
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
2020	6,776	3,095	883	45.7	13.0

Screening pathway variance by DHB

Table 12 shows a breakdown of screening pathway variance by DHB and type of variance for the 2020 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 12: Screening pathway variance by DHB, January to December 2020

DHB	Second trimester screening results				
	Number			Percentage	
	Total T2 screens	with NT	with PAPP-A	with NT	with PAPP-A
Northland	260	125	29	48.1	11.2
Waitematā	775	376	113	48.5	14.6
Auckland	591	191	103	32.3	17.4
Counties Manukau	1,426	428	202	30.0	14.2
Waikato	568	334	31	58.8	5.5
Lakes	174	89	15	51.1	8.6
Bay of Plenty	263	153	22	58.2	8.4
Tairāwhiti	91	49	S	53.8	S
Hawke's Bay	244	99	47	40.6	19.3
Taranaki	134	73	18	54.5	13.4
MidCentral	214	123	22	57.5	10.3
Whanganui	151	58	17	38.4	11.3
Capital & Coast	247	135	12	54.7	4.9
Hutt Valley	259	137	24	52.9	9.3
Wairarapa	75	44	S	58.7	S

Nelson Marlborough	142	92	15	64.8	10.6
West Coast	36	15	10	41.7	27.8
Canterbury	726	356	145	49.0	20.0
South Canterbury	93	50	17	53.8	18.3
Southern	302	166	33	55.0	10.9
National*	**6,776**	**3,095**	**883**	**45.7**	**13.0**

*DHB counts do not sum to National total.
(S) Suppressed if the number of screens was < 6.

Screening pathway variance by age, ethnicity and deprivation

Table 13 shows a breakdown of screening pathway variance by age, ethnicity, and deprivation for the 2020 year. The results show higher proportions for pathway variance for women in the 25–29 age group (48.7%), women of Other ethnicity (55.1%) and NZ deprivation quintile 1 (54.2%).

Table 13: Screening pathway variance by age, ethnicity and deprivation, January to December 2020

	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	379	173	20	45.6	5.3
20–24	1,316	604	121	45.9	9.2
25–29	2,056	1,002	259	48.7	12.6
30–34	1,994	859	331	43.1	16.6
35–39	849	379	128	44.6	15.1
40–44	174	75	22	43.1	12.6
45 and over	8	3	2	37.5	25.0

Ethnicity					
Māori	1,775	831	146	46.8	8.2
Pacific	1,105	364	95	32.9	8.6
Asian	1,460	557	261	38.2	17.9
Other	2,436	1,343	381	55.1	15.6
NZ Deprivation quintile					
Quintile 1 (least deprived)	744	403	106	54.2	14.2
Quintile 2	1004	520	156	51.8	15.5
Quintile 3	1072	508	157	47.4	14.6
Quintile 4	1715	757	233	44.1	13.6
Quintile 5 (most deprived)	2235	905	230	40.5	10.3
National*	**6,776**	**3,095**	**883**	**45.7**	**13.0**

*Deprivation counts do not sum to National total.

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 14 shows the total number of incomplete screens by calendar year and trimester of screen. Nearly all incomplete screens are 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 2020 was 6,321, which equates to 12.6 percent of screens commenced that year and demonstrates an overall increase in incomplete screens over the past six years.

Table 14: Incomplete screens by trimester, January 2015 to December 2020

Trimester of screen	Number and percentage of incomplete screens					
	2015	2016	2017	2018	2019	2020
T1 screens	4,544	4,305	4,567	4,871	5,465	6,209
T2 screens	225	144	85	88	126	112
Total screens	**4,769**	**4,449**	**4,652**	**4,959**	**5,591**	**6,321**
Percentage incomplete	10.1	9.3	9.7	10.3	11.7	12.6

Incomplete T1 screens by reason incomplete

Table 15 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.

In 2020, the proportion of incomplete T1 screens out of all commenced T1 screens was 14 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.

During 2020, antenatal screening was considered an essential service and continued through COVID-19 restrictions. Further analysis (not shown here) suggests that while the number of women screened was not impacted, the reduced availability of NT scans during the national lockdowns may have caused a slightly higher number of incomplete screens in March and April 2020.[7]

Table 15: Incomplete T1 screens by reason incomplete, January 2015 to December 2020

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
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
2020	6,209	36,893	43,102	4,703	1,504	7	10.9	3.5	14.4	75.7	24.2

[7] Antenatal Screening for Down Syndrome and Other Conditions in Covid-19 time January to June 2020: report from LabPLUS and CHL

Incomplete T1 screens by reason and DHB

Table 16 provides a breakdown of incomplete T1 screens by DHB and reason for the 2020 year. The lower numbers involved limit the comparisons that can be made between DHBs. The percentage of T1 screens that were incomplete due to no blood sample ranged from 61 percent (Nelson Marlborough) to 86 percent (Wairarapa).

Table 16: Incomplete T1 screens by reason and DHB, January to December 2020

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	216	1,041	1,257	163	53	S	13.0	4.2	17.2	75.5	24.5
Waitematā	954	5,155	6,109	759	195	S	12.4	3.2	15.6	79.6	20.4
Auckland	724	2,874	3,598	591	133	S	16.4	3.7	20.1	81.6	18.4
Counties Manukau	723	4,186	4,909	523	200	S	10.7	4.1	14.7	72.3	27.7
Waikato	556	3,764	4,320	449	106	S	10.4	2.5	12.9	80.8	19.1
Lakes	174	853	1,027	144	30	S	14.0	2.9	16.9	82.8	17.2
Bay of Plenty	330	2,110	2,440	253	77	S	10.4	3.2	13.5	76.7	23.3
Tairāwhiti	83	387	470	66	17	S	14.0	3.6	17.7	79.5	20.5

Hawke's Bay	245	1,180	1,425	182	63	S	12.8	4.4	17.2	74.3	25.7
Taranaki	149	912	1,061	99	50	S	9.3	4.7	14.0	66.4	33.6
MidCentral	248	1,437	1,685	198	50	S	11.8	3.0	14.7	79.8	20.2
Whanganui	86	381	467	63	23	S	13.5	4.9	18.4	73.3	26.7
Capital & Coast	266	1,884	2,150	202	64	S	9.4	3.0	12.4	75.9	24.1
Hutt Valley	209	1,229	1,438	162	47	S	11.3	3.3	14.5	77.5	22.5
Wairarapa	71	326	397	61	10	S	15.4	2.5	17.9	85.9	14.1
Nelson Marlborough	142	1,164	1,306	86	56	S	6.6	4.3	10.9	60.6	39.4
West Coast	20	211	231	14	6	S	6.1	2.6	8.7	70.0	30.0
Canterbury	655	4,800	5,455	424	231	S	7.8	4.2	12.0	64.7	35.3
South Canterbury	52	413	465	38	14	S	8.2	3.0	11.2	73.1	26.9
Southern	300	2,570	2,870	221	78	S	7.7	2.7	10.5	73.7	26.0
National*	**6,209**	**36,893**	**43,102**	**4,703**	**1,504**	**7**	**10.9**	**3.5**	**14.4**	**75.7**	**24.2**

*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 2020, 1.6 percent of T2 commenced screens were incomplete, compared with 14.4 percent of T1 commenced screens. As Table 17 shows, the percentage of incomplete T2 screens decreased from 3.9 percent in 2015 to 1.6 percent in 2020.

Table 17: Incomplete T2 screens, January 2015 to December 2020

Year	Commenced second trimester	No result issued	Percentage incomplete
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
2020	6,888	112	1.6
Total	**37,984**	**780**	**2.1**

Incomplete T2 screens by DHB

Table 18 shows a breakdown of incomplete T2 screens by DHB for the 2020 year. The low volumes involved limit meaningful DHB comparisons.

Table 18: Incomplete T2 screens by DHB, January to December 2020

DHB	Commenced second trimester	No result issued	Percentage incomplete
Northland	265	S	S
Waitematā	788	13	1.6
Auckland	599	8	1.3
Counties Manukau	1,448	22	1.5
Waikato	583	15	2.6
Lakes	176	S	S
Bay of Plenty	266	S	S
Tairāwhiti	93	S	S
Hawke's Bay	246	S	S
Taranaki	137	S	S
MidCentral	216	S	S
Whanganui	155	S	S
Capital & Coast	254	7	2.8
Hutt Valley	265	6	2.3
Wairarapa	75	S	S
Nelson Marlborough	144	S	S
West Coast	37	S	S
Canterbury	731	S	S
South Canterbury	96	S	S
Southern	309	7	2.3
National*	**6,888**	**112**	**1.6**

*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 19 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 2020, 4.2 increased-risk results were issued for every 100 screens completed. This is the same as the rate reported in 2019 and similar to the rate reported in 2018.

Table 19: Number and rate per 100 screens of increased-risk screening results for trisomy 21, 18 or 13, January 2015 to December 2020

	Number and rate of increased-risk screens					
	2015	2016	2017	2018	2019	2020
Total increased-risk results	1,168	1,189	1,318	1,764	1,764	1,844
Positive test rate per 100 completed screens	2.8	2.7	3.1	4.1	4.2	4.2

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

Table 20 shows the number and proportion of screening risk results that were classified as increased risk for any one or more of trisomy 21, 18, or 13 by age at screen, ethnicity and deprivation for the 2020 year.

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. Positive test rate was higher for Pacific and Asian women compared with other ethnicities. Women in deprivation quintiles 4 and 5 had a higher positive test rate than women in less deprived areas.

Table 20: Increased-risk screening results for trisomy 21, 18 or 13 by age, ethnicity and deprivation, January to December 2020

	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	12	1,129	1.1
20–24	76	5,400	1.4
25–29	213	13,056	1.6
30–34	498	15,913	3.1
35–39	737	7,046	10.5
40–44	288	1,080	26.7
45 and over	20	45	44.4
Ethnicity			
Māori	233	6,804	3.4
Pacific	197	2,988	6.6
Asian	522	9,973	5.2
Other	892	23,904	3.7

NZ Deprivation quintile			
Quintile 1 (least deprived)	313	7,820	4.0
Quintile 2	322	8,248	3.9
Quintile 3	340	8,541	4.0
Quintile 4	457	10,058	4.5
Quintile 5 (most deprived)	412	8,979	4.6
Unknown	0	23	0.0
National	**1,844**	**43,669**	**4.2**

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

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

Trisomy 18 and 13 each had low positivity rates of 0.4 per 100 screens, while the positive test rate for trisomy 21 was 4.1 per 100 screens which is unchanged since 2019. The second trimester positive test rate for trisomy 21 was higher than the first trimester positive test rate (5.0 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 21: Increased-risk screening results for trisomy 21, 18 and 13 by trimester of screen, January 2015 to December 2020

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						
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
2020	1,793	4.1	1,452	3.9	341	5.0
Trisomy 18						
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
2020	188	0.4	160	0.4	28	0.4
Trisomy 13						
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
2020	159	0.4	140	0.4	19	0.3

Any one or more of trisomy 21, 18 or 13						
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
2020	1,844	4.2	1,483	4.0	361	5.3

Increased-risk screening results stratified by risk level

Table 22 shows the number of increased-risk results stratified by risk level for each of trisomy 21, 18 and 13 for the 2020 year. A woman's screen result may indicate an increased-risk for more than one of trisomy 21, 18 and 13 so the sum of the values in Table 22 will be greater than the total number of increased-risk results for 2020.

Table 22: Increased-risk screening results for trisomy 21, 18 and 13 by risk level, January to December 2020

Risk level	Trisomy 21	Trisomy 18	Trisomy 13
1:5 to 1:20	230	64	46
1:21 to 1:50	179	32	27
1:51 to 1:300	1,384	92	86

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

This indicator reports information on the number and proportion of women who complete prenatal diagnostic testing (CVS or amniocentesis) following an increased-risk screening result for trisomy 21, trisomy 18 or trisomy 13. Following an increased-risk result, women may choose to have diagnostic testing (either amniocentesis or CVS) to determine the absence or the presence of the condition.

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

Table 23 shows the diagnostic testing rate by trimester of screen from 2015 to 2020. In 2020, for every 100 women that received an increased-risk result after a first or second trimester screen, 30 women had a diagnostic test. There is an overall downward trend in diagnostic testing from 2015 to 2020, which may be partly due to increasing availability and uptake of non-invasive prenatal screening (NIPS). For the second consecutive year, the second trimester diagnostic testing rate (32.1) was slightly higher than the first trimester diagnostic testing rate (29.3) in 2020. See Appendix 3 for a summary of diagnostic test results for women who had an increased-risk screen in 2020.

Table 23: Diagnostic testing volumes for women with increased-risk screens by trimester of screen, January 2015 to December 2020

Trimester of screen	Diagnostic tests per 100 increased-risk screens					
	2015	2016	2017	2018	2019	2020
T1 screen	59.0	46.9	36.7	38.4	38.7	29.3
T2 screen	44.3	40.5	29.0	35.3	39.4	32.1
Total screens	**56.3**	**45.7**	**35.1**	**37.7**	**38.8**	**29.9**

Diagnostic testing volumes for women with increased-risk screens by DHB

The number of diagnostic tests and rate per 100 increased-risk screens by DHB is given in Table 24. Many DHBs have low numbers and care should be taken with comparisons.

In 2020, many DHBs saw a significant drop in the rate of diagnostic testing for women with increased-risk screens.

Table 24: Diagnostic testing volumes for women with increased-risk screens by DHB, January 2015 to December 2020

DHB	Number of diagnostic tests						Diagnostic tests per 100 increased-risk screens					
	2015	2016	2017	2018	2019	2020	2015	2016	2017	2018	2019	2020
Northland	21	12	12	18	25	19	48.8	40.0	34.3	38.3	44.6	39.6
Waitematā	107	82	78	102	97	66	57.5	44.6	37.5	37.2	37.2	25.1
Auckland	76	72	49	63	70	41	53.5	45.0	30.6	31.8	34.7	23.7
Counties Manukau	86	78	55	99	107	95	53.8	54.9	31.4	39.6	45.5	33.9
Waikato	42	45	29	56	66	56	60.0	52.9	30.2	39.4	40.2	32.4
Lakes	28	16	14	19	14	16	71.8	59.3	46.7	46.3	41.2	35.6
Bay of Plenty	20	17	18	26	24	25	66.7	44.7	40.0	39.4	38.1	35.2
Tairāwhiti	S	S	S	7	S	7	S	S	S	43.8	S	30.4
Hawke's Bay	15	8	7	15	13	17	51.7	28.6	26.9	33.3	40.6	35.4
Taranaki	10	8	S	10	17	14	43.5	36.4	S	35.7	51.5	41.2
MidCentral	8	15	20	19	25	19	44.4	46.9	50.0	52.8	44.6	33.3
Whanganui	S	6	S	9	11	S	S	66.7	S	52.9	57.9	S
Capital & Coast	65	41	30	34	37	23	60.7	60.3	32.6	37.0	36.3	20.9
Hutt Valley	18	15	15	18	19	24	64.3	45.5	45.5	34.0	28.4	28.6
Wairarapa	S	S	S	S	6	S	S	S	S	S	50.0	S

Nelson Marlborough	15	14	13	20	29	28	57.7	51.9	48.1	44.4	56.9	58.3
West Coast	S	6	S	S	S	S	S	85.7	S	S	S	S
Canterbury	83	80	70	95	90	65	50.6	36.7	32.0	34.2	34.5	27.4
South Canterbury	9	S	7	S	S	S	75.0	S	36.8	S	S	S
Southern	40	20	31	44	24	23	60.6	37.0	44.9	48.4	32.0	24.2
National*	**657**	**543**	**463**	**665**	**685**	**551**	**56.3**	**45.7**	**35.1**	**37.7**	**38.8**	**29.9**

*DHB counts do not sum to National total.
(S) Suppressed if the number of diagnostic tests was < 6.

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

Table 25 shows the diagnostic testing rates for women with increased-risk screens by age and ethnicity for 2015 to 2020.

For 2020, women aged 20–24 had the highest rate of diagnostic testing compared to the other age groups. Diagnostic testing rates were highest for Asian and Māori women (32 tests per 100 increased-risk screens), followed by women of Other ethnicity (30 per 100 increased-risk screens). Pacific women had the lowest rate of diagnostic testing (23 per 100 increased-risk screens).

Table 25: Diagnostic testing volumes for women with increased-risk screens by age and ethnicity, January 2015 to December 2020

	Diagnostic tests per 100 increased-risk screens					
	2015	**2016**	**2017**	**2018**	**2019**	**2020**
Age at screen (years)						
Under 20	53.8	45.5	17.4	28.6	64.3	33.3
20–24	51.7	55.6	43.5	50.0	48.6	42.1
25–29	58.1	49.4	38.2	44.7	43.1	35.7
30–34	61.8	47.7	38.8	41.3	42.7	33.3
35–39	57.0	46.0	32.9	35.3	36.0	27.5
40–44	50.9	39.0	29.8	32.5	33.1	21.9
45 and over	41.2	27.8	35.3	13.6	43.8	35.0
Ethnicity						
Māori	45.1	46.7	30.1	37.3	40.9	31.8
Pacific	36.2	34.3	31.0	33.1	36.1	23.4
Asian	63.3	56.3	37.7	37.9	39.5	32.0
Other	58.7	42.1	35.9	38.5	38.3	29.6
National	**56.3**	**45.7**	**35.1**	**37.7**	**38.8**	**29.9**

Table 26 provides diagnostic testing rates by deprivation quintile. Women in the least deprived quintile had the lowest rate of diagnostic testing in 2020 (25 tests per 100 increased-risk screens).

Table 26: Diagnostic testing volumes for women with increased-risk screens by deprivation, January to December 2020

NZ Deprivation quintile	Diagnostic tests	Diagnostic tests per 100 increased-risk screens
Quintile 1 (least deprived)	77	24.6
Quintile 2	93	28.9
Quintile 3	107	31.5
Quintile 4	150	32.8
Quintile 5 (most deprived)	124	30.1
National	**551**	**29.9**

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

Each screening result includes a separate risk for each of trisomy 21, 18 and 13. For the analysis in this report, women were assigned a combined trisomy risk level based on the highest risk score they received across the three trisomies. Table 27 shows the number of diagnostic tests for women that received an increased-risk result during 2020 for one or more of trisomy 21, 18 or 13, stratified by risk level. As expected, diagnostic testing increased with increasing risk level, going from 23 tests per 100 women with a risk of 1:51 to 1:300 to 58 tests per 100 women with a risk of 1:5 to 1:20.

Table 27: Diagnostic testing volumes for women with increased-risk screens by risk level, January to December 2020

Risk level	Number of diagnostic tests	Number of increased-risk screens	Tests per 100 increased-risk screens
1:5 to 1:20	149	259	57.5
1:21 to 1:50	77	188	41.0
1:51 to 1:300	325	1,397	23.3

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 0.45 per 100 low-risk screens in 2020, the lowest it has been in the reporting period.

Table 28: Diagnostic testing volumes for women with low-risk screens by trimester of screen, January 2015 to December 2020

Trimester of screen	Diagnostic tests per 100 low-risk screens					
	2015	2016	2017	2018	2019	2020
T1 screen	0.74	0.53	0.75	0.80	0.84	0.48
T2 screen	0.36	0.69	0.70	0.74	0.64	0.28
Total screens	**0.69**	**0.55**	**0.75**	**0.79**	**0.81**	**0.45**

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 2015 to 2020, as shown in Table 29. Given the low numbers involved, caution should be taken in making comparisons between DHBs.

Table 29: Diagnostic testing volumes for women with low-risk screens by DHB, January 2015 to December 2020

DHB	Number of diagnostic tests						Diagnostic tests per 100 low-risk screens					
	2015	2016	2017	2018	2019	2020	2015	2016	2017	2018	2019	2020
Northland	7	S	S	11	S	S	0.66	S	S	0.98	S	S
Waitematā	33	37	43	52	53	41	0.55	0.59	0.72	0.88	0.94	0.72
Auckland	36	20	29	33	32	18	0.80	0.46	0.78	0.89	0.92	0.55
Counties Manukau	23	28	45	29	53	24	0.45	0.53	0.87	0.57	1.05	0.45
Waikato	21	16	33	34	30	18	0.56	0.41	0.83	0.88	0.75	0.43
Lakes	8	S	6	7	11	8	0.84	S	0.60	0.67	1.10	0.81
Bay of Plenty	7	12	13	20	17	12	0.38	0.59	0.58	0.92	0.75	0.52
Tairāwhiti	S	S	S	S	S	S	S	S	S	S	S	S
Hawke's Bay	8	S	6	14	7	S	0.64	S	0.45	1.01	0.53	S
Taranaki	S	S	S	S	S	S	S	S	S	S	S	S
MidCentral	11	S	11	S	6	8	0.93	S	0.73	S	0.42	0.50
Whanganui	S	S	S	S	S	S	S	S	S	S	S	S
Capital & Coast	22	19	15	18	17	8	0.86	0.72	0.66	0.80	0.81	0.40
Hutt Valley	9	6	10	6	7	S	0.69	0.44	0.78	0.43	0.53	S
Wairarapa	S	S	6	S	S	S	S	S	1.41	S	S	S

Nelson Marlborough	9	9	7	10	13	S	0.77	0.77	0.56	0.82	1.08	S
West Coast	S	S	S	S	S	S	S	S	S	S	S	S
Canterbury	52	37	47	44	47	20	1.08	0.74	0.92	0.88	0.96	0.38
South Canterbury	S	7	7	S	S	S	S	1.35	1.35	S	S	S
Southern	29	23	22	23	19	9	1.12	0.87	0.80	0.88	0.73	0.32
National*	**283**	**233**	**312**	**325**	**330**	**188**	**0.69**	**0.55**	**0.75**	**0.79**	**0.81**	**0.45**

*DHB counts do not sum to National total.
(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 30 shows the rate of diagnostic testing for women with low-risk screening results by age and ethnicity for 2015 to 2020.

For 2020, the rate of diagnostic testing was highest for women aged 40–44 years. Asian women were the most likely to have a diagnostic test after a low-risk screen (0.7 tests per 100 low-risk screens) and Pacific women the least likely (0.1 tests per 100 low-risk screens).

Table 30: Diagnostic testing volumes for women with low-risk screens by age and ethnicity, January 2015 to December 2020

	Diagnostic tests per 100 low-risk screens					
	2015	**2016**	**2017**	**2018**	**2019**	**2020**
Age at screen (years)						
Under 20	0.33	0.34	0.81	0.81	0.71	0.09
20–24	0.35	0.43	0.68	0.71	0.67	0.51
25–29	0.52	0.50	0.65	0.60	0.66	0.36
30–34	0.60	0.54	0.67	0.84	0.81	0.36
35–39	1.11	0.66	0.99	0.96	1.17	0.79
40–44	3.04	1.33	1.67	1.70	1.83	1.01
45 and over	2.13	3.28	1.61	2.08	0.00	0.00
Ethnicity						
Māori	0.46	0.50	0.65	0.74	0.68	0.30
Pacific	0.48	0.35	0.75	0.79	0.83	0.11
Asian	0.80	0.54	0.89	0.76	0.86	0.71
Other	0.72	0.58	0.73	0.80	0.83	0.43
National	**0.69**	**0.55**	**0.75**	**0.79**	**0.81**	**0.45**

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

Table 31 shows the rate of diagnostic testing for women with low-risk screening results, stratified by risk level. Given the low numbers involved for some risk categories, numbers have been aggregated for 2017–2020.

Table 31: Diagnostic testing volumes for women with low-risk screens by risk level, aggregated 2017–2020

Risk level	Number of diagnostic tests	Number of low-risk screens	Tests per 100 low-risk screens
1:301 to 1:500	79	3,486	2.27
1:501 to 1:1,000	131	9,077	1.44
1:1,001 to 1:2,000	115	13,952	0.82
1:2,001 to 1:3,000	109	11,330	0.96
1:3,001 to 1:4,000	66	9,805	0.67
1:4,001 to 1:5,000	53	8,274	0.64
1:5,001 to 1:10,000	173	31,052	0.56
1:10,001 to 1:100,000	429	78,436	0.55

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, advanced maternal age), a diagnostic test done for another reason and the karyotype reported, or an abnormal ultrasound finding.

The methodology for calculating unscreened[8] women has been updated for the 2020 report, improving identification of unscreened women. The improved identification means that figures for 2020 are higher than in recent years.

Diagnostic volumes for unscreened women

During 2020, 247 diagnostic tests were completed for unscreened women. This is higher than 2018 and 2019 (156 and 174 tests respectively) but similar to the number of tests undertaken in 2015 (252). Part of this increase in 2020 is due to better data linkage to identify those women who have had a diagnostic test but have not had a prenatal screen. In addition, the increase may be partly due to COVID-19 isolation requirements and people choosing to stay home, despite screening services being available as they were considered essential services.

Table 32 shows the number of diagnostic tests by DHB for 2015–2020, and Table 33 shows the breakdown by age and ethnicity. Table 34 shows the breakdown by NZ deprivation quintile for 2020 only.

[8] Unscreened = no prenatal screen result so either didn't start screening or started but didn't complete screening.

Table 32: Diagnostic testing volumes for unscreened women by DHB, January 2015 to December 2020

DHB	Number of diagnostic tests					
	2015	2016	2017	2018	2019	2020
Northland	8	6	S	S	S	9
Waitematā	22	19	14	24	23	45
Auckland	18	23	10	13	26	27
Counties Manukau	18	21	11	10	23	25
Waikato	15	16	6	12	12	23
Lakes	8	S	S	7	S	6
Bay of Plenty	14	10	S	S	6	7
Tairāwhiti	S	S	S	S	S	S
Hawke's Bay	7	8	S	S	S	S
Taranaki	11	S	S	7	S	9
MidCentral	8	9	S	6	S	12
Whanganui	S	S	S	S	S	S
Capital & Coast	36	25	12	8	16	13
Hutt Valley	22	10	6	6	8	7
Wairarapa	S	S	S	S	S	S
Nelson Marlborough	6	S	S	S	S	S
West Coast	S	S	S	S	S	S
Canterbury	30	30	18	31	25	30
South Canterbury	S	S	S	S	S	S
Southern	19	14	S	11	7	16
National	**252**	**212**	**107**	**156**	**174**	**247***

*DHB counts do not sum to National total.
(S) Suppressed if the number of diagnostic tests was < 6.

Table 33: Diagnostic testing volumes for unscreened women by age and ethnicity, January 2015 to December 2020

	Number of diagnostic tests					
	2015	**2016**	**2017**	**2018**	**2019**	**2020**
Age at screen (years)						
Under 20	16	12	4	4	4	4
20–24	19	17	12	18	19	24
25–29	53	36	27	29	30	46
30–34	70	60	26	47	56	77
35–39	54	56	22	45	48	61
40–44	35	28	15	13	15	29
45 and over	5	3	1	0	2	6
Ethnicity						
Māori	44	32	14	32	18	34
Pacific	21	11	11	7	11	11
Asian	33	36	17	19	35	48
Other	154	133	65	98	110	154
National	**252**	**212**	**107**	**156**	**174**	**247**

Table 34: Diagnostic testing volumes for unscreened women by deprivation quintile, January to December 2020

NZ Deprivation quintile	Number of diagnostic tests	Percentage
Quintile 1 (least deprived)	51	20.6
Quintile 2	58	23.5
Quintile 3	48	19.4
Quintile 4	45	18.2
Quintile 5 (most deprived)	44	17.8
Unknown	1	0.4
National	**247**	**100.0**

Diagnostic results for unscreened women

A breakdown of prenatal diagnostic testing results for unscreened women for the 2020 year is given in Table 35. Of the 247 diagnostic tests in 2020 for unscreened women, 194 fetuses (78.5%) had a normal karyotype.

Table 35: Diagnostic testing results for unscreened women, January to December 2020

Karyotype result	Number	Percentage
Normal karyotype	194	78.5
Trisomy 21	23	9.3
Trisomy 18	13	5.3
Trisomy 13	5	2.0
Turner syndrome	5	2.0
Triploidy	3	1.2
Other chromosomal abnormality	4	1.6
Total	**247**	**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 2020 was 0.061, continuing a declining trend over the previous years (see Table 36). A value of 0.061 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 36: Positive predictive value of screening for trisomy 21, 18 or 13, January 2015 to December 2020

Year	True positives	False positives	PPV	95% confidence interval
2015	132	1035	0.113	(0.095, 0.131)
2016	110	1079	0.093	(0.076, 0.109)
2017	107	1211	0.081	(0.066, 0.096)
2018	118	1646	0.067	(0.055, 0.079)
2019	113	1651	0.064	(0.053, 0.075)
2020	113	1731	0.061	(0.050, 0.072)

The PPV changes when calculated for a specific trisomy. When looking at trisomy 21, the PPV for 2020 is the lowest it has been in the six-year reporting period (0.047). This means that if a woman receives an increased-risk result for trisomy 21 there is a 4.7 percent probability that she is carrying a fetus with trisomy 21.

Table 37: Positive predictive value of screening for trisomy 21, January 2015 to December 2020

Year	True positives	False positives	PPV	95% confidence interval
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)
2020	85	1,708	0.047	(0.038, 0.057)

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

In 2020, the combined PPV for trisomies 18 or 13 was 0.104 (see Table 38) which is higher than the PPV for trisomy 21. However, the number of positive diagnoses for these two trisomies is low, so caution should be taken when interpreting these results.

Table 38: Positive predictive value of screening for trisomy 18 or 13, January 2015 to December 2020

Year	True positives	False positives	PPV	95% confidence interval
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)
2020	27	233	0.104	(0.067, 0.141)

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

Table 39 shows the PPV stratified by the risk level indicated in the screening result. Data has been aggregated for 2017–2020. Women that received an increased-risk result of 1:5 to 1:20 for trisomy 21 had a 27 percent probability of carrying a fetus with trisomy 21. As expected, the PPV was lower for women with increased risks of 1:21 to 1:50 at 4 percent probability, and lower again for women with increased-risk results of 1:51 to 1:300 at 1 percent probability.

Table 39: Positive predictive value of screening for trisomy 21 by risk level, aggregated 2017–2020

Risk level	True positives	False positives	PPV
1:5 to 1:20	237	639	0.27
1:21 to 1:50	30	653	0.04
1:51 to 1:300	69	4,861	0.01

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

Table 40 shows true positives, false positives and PPV aggregated for 2017–2020 by age and ethnicity.

The PPV of screening for trisomy 21 varied by age group. Women aged 40–44 had the highest PPV (0.06 or 6%) and women under 20 had the lowest PPV (0.03 or 3%). The PPV also 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%).

Table 40: Positive predictive value of screening for trisomy 21 by age and ethnicity, aggregated 2017–2020

	True positives	False positives	PPV
Age at screen (years)			
Under 20	2	58	0.03
20–24	16	277	0.05
25–29	34	733	0.04
30–34	80	1,642	0.05
35–39	127	2,245	0.05
40–44	73	1,129	0.06
45 and over	4	69	0.05
Ethnicity			
Māori	36	784	0.04
Pacific	11	599	0.02
Asian	60	1,775	0.03
Other	229	2,995	0.07
Total	**336**	**6,153**	**0.05**

Table 41 shows PPV by deprivation quintile for 2020. While there is little variation between quintiles, the PPV is lower for women from areas of higher deprivation (quintiles 4 and 5).

Table 41: Positive predictive value of screening for trisomy 21 by deprivation, January to December 2020

NZ Deprivation quintile	True positives	False positives	PPV
Quintile 1 (least deprived)	15	292	0.05
Quintile 2	17	295	0.05
Quintile 3	16	310	0.05
Quintile 4	20	427	0.04
Quintile 5 (most deprived)	17	384	0.04
Total	**85**	**1708**	**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 2020 was 0.04 (or 4%), which is unchanged since 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 42: False positive rate for trisomy 21, 18 or 13, January 2015 to December 2020

Year	False positives	True negatives	False positive rate	95% confidence interval
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, 0.040)
2020	1,731	41,801	0.04	(0.038, 0.042)

As shown in Table 43, the false positive rate was higher for second trimester screens (5.2%) than for first trimester screens (3.8%), consistent with previous years.

Table 43: False positive rate for trisomy 21, 18 or 13 by trimester of screen, January to December 2020

Trimester	False positives	True negatives	False positive rate	95% confidence interval
T1 screens	1,382	35,388	0.038	(0.036, 0.040)
T2 screens	349	6,413	0.052	(0.046, 0.057)
Total	**1,731**	**41,801**	**0.040**	**(0.038, 0.042)**

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 44). However, the combined false positive rate for trisomy 18 and trisomy 13 is much lower (0.005 or 0.5% for 2020, see Table 45).

Table 44: False positive rate for trisomy 21, January 2015 to December 2020

Year	False positives	True negatives	False positive rate	95% confidence interval
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)
2020	1,708	41,860	0.04	(0.037, 0.041)

Table 45: False positive rate for trisomy 18 and 13, January 2015 to December 2020

Year	False positives	True negatives	False positive rate	95% confidence interval
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)
2020	233	43,366	0.005	(0.005, 0.006)

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

False positive rates by age and ethnicity are shown in Table 46. The false positive rate for trisomy 21 increases with age. For example, the false positive rate for women under 20 years of age in 2020 was 0.01 (1%) compared with 0.44 (44%) for women 45 years and over. 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 2020 varied across ethnic groups from 0.03 (3%) for Māori and Other to 0.06 (6%) for Pacific.

Table 46: False positive rate for trisomy 21 by age and ethnicity, January 2015 to December 2020

	2015	2016	2017	2018	2019	2020
Age at screen (years)						
Under 20	0.01	0.01	0.02	0.01	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	0.02
30–34	0.02	0.02	0.02	0.03	0.03	0.03
35–39	0.05	0.05	0.05	0.08	0.09	0.10
40–44	0.19	0.15	0.17	0.26	0.30	0.26
45 and over	0.27	0.21	0.17	0.31	0.25	0.44
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	0.06
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

In 2020, there appears to be little difference across deprivation quintiles (see Table 47). The false positive rate for trisomy 21 is slightly higher for women in the most deprived quintiles with Quintiles 4 and 5 having a false positive rate of 0.043 (4.3%) while Quintiles 2 and 3 have the lowest rate (0.036 or 3.6%).

Table 47: False positive rate for trisomy 21 by deprivation, January to December 2020

NZ Deprivation quintile	False positives	True negatives	False positive rate
Quintile 1 (least deprived)	292	7,507	0.037
Quintile 2	295	7,933	0.036
Quintile 3	310	8,212	0.036
Quintile 4	427	9,609	0.043
Quintile 5 (most deprived)	384	8,576	0.043
Unknown	-	23	
Total	**1,708**	**41,860**	**0.039**

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 2020 is given in Table 48. Rates for trisomy 21 alone, and for trisomies 18 and 13 together are given in tables 49 and 50 respectively. As each of these tables show, detection rates fluctuated over this period.

The overall detection rate for trisomy 21, 18 and 13 for 2020 was 0.82 (82%) (see Table 48). A detection rate of 0.82 means that there is an 82 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 48: Detection rate for trisomy 21, 18 or 13, January 2015 to December 2020

Year	True positives	False negatives	Detection rate	95% confidence interval
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)
2020	113	24	0.82	(0.761, 0.888)

The detection rate for trisomy 21 alone is shown in Table 49. The rate for 2020 (0.84) was slightly higher than the overall rate for trisomy 21, 18 and 13 (0.82). The detection rate for trisomy 18 and 13 was lower, at 0.71 (Table 50).

Table 49: Detection rate for trisomy 21, January 2015 to December 2020

Year	True positives	False negatives	Detection rate	95% confidence interval
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)
2020	85	16	0.84	(0.770, 0.913)

Table 50: Detection rate for trisomy 18 or 13, January 2015 to December 2020

Year	True positives	False negatives	Detection rate	95% confidence interval
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)
2020	27	11	0.71	(0.566, 0.855)

Appendix 1: Indicator definitions

Table 51: 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 (no prenatal screen result)
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[9] was obtained from the National Maternity Collection for each year.

Table 52: Live births and still births by DHB, 2015–2020

DHB	2015	2016	2017	2018	2019	2020
Northland	2,135	2,265	2,235	2,190	2,310	2,381
Waitematā	7,560	7,930	7,720	7,425	7,780	7,470
Auckland	5,900	5,905	5,625	5,430	5,590	5,155
Counties Manukau	8,190	8,240	8,280	8,160	8,400	8,399
Waikato	5,275	5,355	5,320	5,380	5,450	5,572
Lakes	1,510	1,550	1,555	1,525	1,535	1,434
Bay of Plenty	2,790	2,900	3,105	3,005	3,105	3,130
Tairāwhiti	735	775	705	700	685	709
Hawke's Bay	1,995	2,055	2,125	2,110	2,020	2,073
Taranaki	1,515	1,435	1,400	1,565	1,515	1,456
MidCentral	2,110	2,080	2,135	2,160	2,165	2,148
Whanganui	815	800	845	810	865	818
Capital & Coast	3,535	3,455	3,500	3,200	3,185	3,092
Hutt Valley	1,965	1,970	1,950	1,940	1,965	2,004
Wairarapa	465	465	535	495	515	527
Nelson Marlborough	1,415	1,545	1,425	1,500	1,450	1,417
West Coast	355	320	355	325	345	293
Canterbury	6,215	6,310	6,395	6,250	6,440	6,174
South Canterbury	660	650	635	610	625	591

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

Southern	3,415	3,310	3,435	3,270	3,440	3,275
Total	**58,560**	**59,310**	**59,290**	**58,050**	**59,375**	**58,118**

Table 53: Live births and still births by age group, 2015–2020

Age group (years)	2015	2016	2017	2018	2019	2020
<20	2,780	2,445	2,290	2,130	2,090	1,973
20–24	9,945	9,585	9,325	8,685	8,535	8,246
25–29	15,705	16,540	16,630	16,250	16,395	15,737
30–34	17,910	18,370	18,695	18,705	19,535	19,654
35–39	9,770	9,965	9,875	10,020	10,415	10,261
40–44	2,295	2,275	2,310	2,095	2,265	2,087
45+	140	125	155	160	140	157
Unknown	15	15	10	5	5	3
Total	**58,560**	**59,310**	**59,290**	**58,050**	**59,375**	**58,118**

Table 54: Live births and still births by ethnicity, 2015–2020

Ethnicity	2015	2016	2017	2018	2019	2020
Māori	14,805	15,000	14,955	14,595	14,865	15,015
Pacific	6,075	5,855	5,965	5,970	6,160	6,031
Asian	9,210	10,515	10,560	10,585	11,470	11,350
Other	28,475	27,950	27,810	26,895	26,885	25,722
Total	**58,560**	**59,310**	**59,290**	**58,050**	**59,375**	**58,118**

Table 55: Live births and still births by NZDEP 18, 2015–2020

NZDEP Quintile	2015	2016	2017	2018	2019	2020
1	8,680	8,940	9,160	8,850	9,295	9,134
2	9,635	9,990	9,955	9,820	10,155	10,008
3	11,370	11,420	11,390	10,965	11,365	10,768
4	13,175	13,395	13,485	13,470	13,410	13,387
5	15,565	15,530	15,245	14,900	15,105	14,801
Unknown	135	35	55	40	45	20
Total	**58,560**	**59,310**	**59,290**	**58,050**	**59,375**	**58,118**

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 1,844 women that had an increased risk for trisomy 21, 18 or 13 during 2020, 551 (30%) had a prenatal diagnostic test (CVS or amniocentesis) and 1,293 (70%) did not. Table 56 shows the diagnostic testing results for the 551 prenatal tests, of which 96 had an abnormal karyotype, including 63 confirmed with Down syndrome.

Table 56: Diagnostic results for women who accessed a prenatal diagnostic test following an increased-risk screen for trisomy 21, 18 or 13 during the 2020 year

Karyotype result	Number	Percentage
Normal karyotype	455	82.6
Confirmed Down syndrome	63	11.4
Other result	33	6.0
Total	**551**	**100.0**

Appendix 4: Measuring screening performance

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

Figure 12: Categorisation of screening results

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

Positive predictive value and positive test rate

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

Positive test rate = ((A+B)/N)*100

Positive Predictive Value is the probability of having the condition given 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 170 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 57 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 0.08 percent or less of all negative screens for each of the years from 2015 to 2020.

Table 57: False negative screens for trisomy 21, 18 and 13 by risk level, January 2015 to December 2020

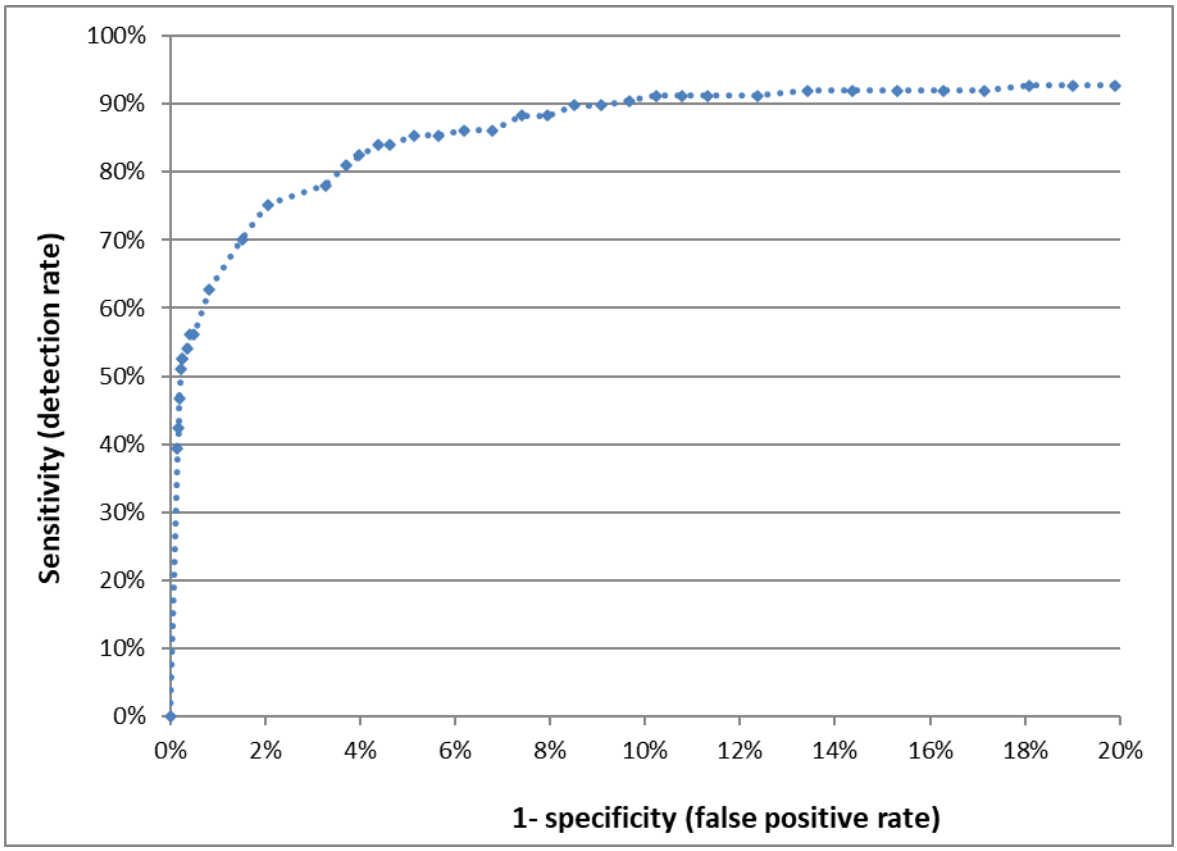
Risk level	False negatives						% of negative screens that are false negatives					
	2015	2016	2017	2018	2019	2020	2015	2016	2017	2018	2019	2020
1:301 to 1:500	4	8	7	5	5	5	0.63	1.25	1.21	0.51	0.51	0.52
1:501 to 1:1,000	10	7	8	12	7	7	0.58	0.46	0.52	0.51	0.28	0.26
1:1,001 to 1:2,000	4	3	8	2	1	2	0.14	0.11	0.33	0.05	0.03	0.05
1:2,001 to 1:3,000	2	6	3	4	6	3	0.08	0.25	0.14	0.14	0.19	0.09
1:3,001 to 1:4,000	1	0	2	0	1	3	0.04	0.00	0.11	0.00	0.04	0.11
1:4,001 to 1:5,000	0	0	0	2	0	1	0.00	0.00	0.00	0.09	0.00	0.04
1:5,001 to 1:10,000	3	2	2	3	2	1	0.03	0.02	0.03	0.04	0.02	0.01
Less than 1:10,000	1	4	5	5	1	2	0.00	0.02	0.02	0.03	0.01	0.01
Total	**25**	**30**	**35**	**33**	**23**	**24**	**0.06**	**0.07**	**0.08**	**0.08**	**0.06**	**0.06**

Appendix 6: ROC curve

Figure 13 shows the false positive rate plotted against the detection rate in what is known as a 'receiver operating characteristic' (ROC) curve. This plots the false positive rate on the horizontal x axis against detection rate on the vertical y axis for different possible cut-off points of the screening test. The aim for a screening test is to maximise detection rate while minimising false positive rate.

In New Zealand the cut-off used for screening is 1:300. With this cut-off, the overall detection rate for trisomy 21, trisomy 18 and trisomy 13 in 2020 was 82.5 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 13: ROC curve for trisomy 21, 18 and 13 screening, 2020



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.

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.

Chromosome – an organised structure of DNA and protein found in all living cells that carries the genes determining heredity.

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

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 (or chance) of a particular outcome or condition.

Screening – a way of identifying 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 negative – when a woman receives a low-risk screening result, and the baby does not have the condition screened for.

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.