Newborn Metabolic Screening Programme

Annual Report 2022

October 2024

Te Kāwanatanga o Aotearoa
New Zealand Government

Health New Zealand
Te Whatu Ora

Acknowledgements

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This publication reports on information Te Toka Tumai Auckland has provided to the Health New Zealand | Te Whatu Ora. The purpose of this publication is to inform discussion and assist the ongoing development of the Newborn Metabolic Screening Programme. All care has been taken in the production of this report, and the data was deemed to be accurate at the time of publication. However, the data may be subject to slight changes over time as further information is received. Before quoting or using this information, it is advisable to check the current status with Health New Zealand | Te Whatu Ora.

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Introduction

This annual report provides information on the performance of the Newborn Metabolic Screening Programme (NMSP) against the agreed set of national indicators in 2022. Regular analysis and reporting of NMSP data are key tools in enabling continuous quality improvement of the programme.

The NMSP Monitoring Framework and monitoring reports are published at Quality standards and monitoring reports – Health New Zealand | Te Whatu Ora. The Newborn Metabolic Screening Programme monitoring indicators dated February 2018, updates and replaces the indicators in the Newborn Metabolic Screening Programme Monitoring Framework, November 2010.

The reports and monitoring framework are available at:
https://www.tewhatuora.govt.nz/health-services-and-programmes/newborn-metabolic-screening-programme/quality-standards/

Background to the programme

The aim of the NMSP is to reduce morbidity and mortality associated with specific congenital disorders by screening newborns to detect the conditions before life-threatening illness or developmental delays occur. Since 1969, almost all newborns in Aotearoa New Zealand have been screened by the programme. Currently, the NMSP identifies about 60 newborns a year with a screened disorder.

To conduct the screening, a midwife, nurse, phlebotomist, or doctor collects a blood sample from the newborn's heel onto a blood spot card (a 'Guthrie card'). The recommended collection time for samples in 2022 was when the newborn was between 48 and 72 hours of age, although samples collected from 24 hours of age were considered acceptable for screening completion. The optimal time for collection was updated in December 2022 to between 24 and 48 hours. Cards are sent urgently to the laboratory, LabPLUS at Auckland City Hospital, which analyses the samples and reports the results to appropriate clinicians. These blood spot samples are screened for the conditions listed in Appendix 1.

In 2005, the National Screening Unit (NSU) at the Ministry of Health - Manatū Hauora began overseeing the NMSP nationally. A significant milestone for the programme came in 2006 when newborn screening was expanded to include fatty acid oxidation disorders and more amino acid breakdown disorders in the screening panel. Screening for severe combined immunodeficiency (SCID) was added in December 2017. In 2023 the NSU was disestablished, and its functions incorporated into the National Public Health Service, Health New Zealand |Te Whatu Ora.

Data summary

Screening data are sourced from LabPLUS at Te Toka Tumai Auckland for all blood spot cards received in the 2022 calendar year.

Birth data from the 2022 calendar year are sourced from the Maternity data collection at Health New Zealand | Te Whatu Ora. In this 2022 report, as occurred for the 2021 report, when a baby is present in screening data but is not in maternity data, that baby is added into the denominator (births). This supplementation is because maternity is an incomplete data set and doing so prevents coverage rates exceeding 100 percent as sometimes was the case in previous years' reports.

District of domicile is taken from the National Enrolment System (NES) data-warehouse. The NES has historical address data so the district of domicile for a baby during the reporting year can be derived. In cases where a baby is not in the NES, the District is then taken from the Health Care User (HCU) data-warehouse. When a newborn's district of domicile is unknown it is set to 'Unknown'.

Ethnicity data are prioritised following Statistics New Zealand's HISO 10001:2017 ethnicity data protocol, which is the standard approach across the health sector.

This report uses data as of November 2023.

Executive summary

The Newborn Metabolic Screening Programme (NMSP) screened 56,875 of the 58,113 babies born in 2022. This represents a national coverage rate of 97.9 percent, which is slightly lower than previous years' coverage rates.

Coverage rates at a District level range from 95.7 percent to 99.5 percent. Since 2017, Districts have been increasingly encouraged to match their birth data with their data on babies screened to ensure all babies whose parents/guardians have given consent are screened. In 2022, national coverage varied by ethnic group: 96.3 percent of Māori newborns, 96.6 percent of Pacific newborns, 98.9 percent of Asian and 98.6 percent of newborns of all other ethnicities were screened.

In 2022, 52 newborns were diagnosed with a screened disorder.

Of the seven indicators with a national target, six were not met:

Indicator	Target	Commentary
Indicator 1: Coverage	**≥ 99**% of babies born nationally and within each of Māori, Pacific, Asian and Other population groups are screened.	The national coverage rate was **97.9%.**
Indicator 2: Timing of sample taking	**≥ 95**% of first samples are taken between 24 and 72 hours after birth.	The national average of first samples taken between 24 and 72 hours after birth was **83.4%.**
Indicator 3: Quality of blood samples	**≥ 99%** of blood spot samples received are of satisfactory quality.	**99.1%** of all samples were of satisfactory quality.
Indicator 4: Sample dispatch and delivery	**≥ 95%** of samples are received by the laboratory within four calendar days of being taken.	**79.8%** of the blood spot cards arrived at the laboratory within four days of sampling.
Indicator 5: Receipt and follow-up of second samples	**100%** of second samples requested are received by the laboratory, had other appropriate follow-up, or were declined by parents/guardians, within ten calendar days of request.	**82.7%** of requests for second samples had appropriate follow up within ten days.
Indicator 6: Laboratory turnaround time for positive results	**100%** of babies with positive results are notified to their Lead Maternity Carer (LMC) / specialist paediatrician by the laboratory within the set timeframes.	**100% of clinical critical** screen positives, and **98% of non-clinical critical** screen positives, were notified within the expected timeframes.
Indicator 7: Age of receipt into clinical care	**100%** of babies who receive a screen positive result and are diagnosed with a screened condition receive active clinical management by the set time frames.	**95%** of babies with disease detected following a positive newborn screen were received into clinical care within an acceptable timeframe.

Indicator 1: Coverage

Description: The proportion of babies born who complete newborn metabolic screening.

Rationale: Newborn screening must be offered for all babies. All babies whose parents/guardians have consented to screening should have completed screening.

Target: ≥99 percent of babies born nationally and within each of Māori, Pacific, Asian and Other population groups are screened.

Interpretation: National coverage was at 97.9 percent, which is below target. Total coverage by District varied from 95.7 percent (MidCentral) to 99.5 percent (Southern). Coverage by ethnicity varied: 96.3 percent for Māori newborns, 96.6 percent for Pacific newborns, 98.9 percent for Asian newborns and 98.6 percent for Other newborns.

Comment: All 20 Districts achieved at least 95 percent coverage in total. Only one District reached the 99 percent target for total coverage; however, no Districts achieved the 99 percent target within each of the Māori, Pacific, Asian, and Other population groups. The slight drop in the coverage rate is likely due to the introduction of the supplementation of data as outlined in the Data Summary.

It is estimated that the NMSP did not screen 1,238 newborns in 2022. It is not possible to distinguish between those unscreened who were offered screening and declined and those who were missed.

Some districts are actively identifying and following up on unscreened newborns, with the support of LabPLUS, to ensure that an offer of screening has been made. Maternity services should contact the Antenatal and Childhood Screening Programme if they would like to put the same processes in place. Improving data quality initiatives are underway to ensure future reporting uses the most accurate data possible.

Table 1: Coverage by Ethnicity, January to December 2022

Ethnicity	Babies screened	Births	Coverage (%)
Māori	14857	15425	96.3
Pacific	5653	5854	96.6
Asian	10816	10932	98.9
Other	25519	25889	98.6
Total*	**56875**	**58113**	**97.9**

**Sub-counts do not add up to the total due to unknowns not being shown.*

Table 2: Coverage by District, January to December 2022

District	Babies screened	Births	Coverage (%)
Northland	2334	2415	96.6
Waitemata	7341	7419	98.9
Auckland	4964	5020	98.9
Counties Manukau	7916	8165	97.0
Waikato	5325	5521	96.4
Lakes	1511	1561	96.8
Bay of Plenty	3108	3182	97.7
Tairawhiti	698	723	96.5
Hawkes Bay	2025	2077	97.5
Taranaki	1367	1402	97.5
MidCentral	2113	2208	95.7
Whanganui	800	825	97.0
Capital and Coast	2894	2931	98.7
Hutt Valley	1814	1866	97.2
Wairarapa	529	538	98.3
Nelson Marlborough	1390	1415	98.2
West Coast	291	297	98.0
Canterbury	6498	6579	98.8
South Canterbury	588	601	97.8
Southern	3348	3365	99.5
National*	**56875**	**58113**	**97.9**

**Sub-counts do not add up to the total due to unknowns not being shown.*

Table 3: Coverage by district and ethnicity, January to December 2022

	Coverage (%)					
District	Māori	Pacific	Asian	Other	Total	Ratio†
Northland	95.6	97.4	100.0	97.7	96.6	0.98
Waitemata	97.7	99.0	99.3	99.1	98.9	0.99
Auckland	97.1	97.2	99.9	99.4	98.9	0.98
Counties Manukau	95.7	95.1	98.6	98.5	97.0	0.98
Waikato	93.6	94.1	98.7	98.0	96.4	0.96
Lakes	95.4	96.4	98.1	98.9	96.8	0.97
Bay of Plenty	97.8	92.8	97.0	98.0	97.7	1.00
Tairawhiti	96.2	100*	90.3	97.6	96.5	0.99
Hawkes Bay	96.5	92.5	100.0	98.6	97.5	0.98
Taranaki	94.1	100.0	99.0	98.9	97.5	0.95
MidCentral	93.2	93.7	100.0	96.7	95.7	0.96
Whanganui	96.5	94.7	97.6	97.6	97.0	0.99
Capital and Coast	98.2	98.1	98.8	99.1	98.7	0.99
Hutt Valley	96.8	99.5	96.2	97.5	97.2	0.99
Wairarapa	98.9	100.0	95.8	98.1	98.3	1.01
Nelson Marlborough	99.3	97.9	100*	97.5	98.2	1.01
West Coast	94.8	100.0	100.0	99.0	98.0	0.96
Canterbury	98.1	99.1	99.0	98.9	98.8	0.99
South Canterbury	98.0	93.3	100.0	97.8	97.8	1.00
Southern	99.7	98.2	98.9	99.6	99.5	1.00
National	**96.3**	**96.6**	**98.9**	**98.6**	**97.9**	**0.98**

† *A rate ratio is used here to focus on equity. It is calculated by dividing Māori coverage by non-Māori coverage. A ratio over 1 means higher coverage for Māori compared with non-Māori.*

** Due to a data mismatch between the denominator and numerator, percentages in this table may exceed 100%. Values greater than 100% are capped at 100*

Table 4: Coverage by year, 2012 to 2022

Year	Births	Babies Screened	Coverage (%)
2012	62,842	61,422	97.7
2013	59,707	59,192	99.1
2014	59,097	58,673	99.3
2015	59,058	58,463	99.0
2016	59,640	59,010	98.9
2017	59,517	58,935	99.0
2018	58,163	57,880	99.5
2019	59,733	59,315	99.3
2020	58,373	57,930	99.2
2021	62,623	61,585	98.3
2022	58,113	56,875	97.9

Indicator 2: Timing of sample taking

Description: The proportion of babies screened who have a newborn metabolic screening sample taken between 48 and 72 hours of age.

Rationale: Prompt sample collection leads to the best possible chance of a baby with a screened condition receiving early diagnosis and treatment. Severe forms of some of the disorders can be fatal within seven to ten days, and many babies may not show any signs or symptoms of disease until irreversible damage has occurred. However, the baby must have been independent of their mother long enough for some biochemical markers to show an abnormality.

Target: ≥95 percent of first samples are taken between 48 and 72 hours after birth.

Note: Since mid-2020 LMCs and other health professionals have been encouraged to collect samples from 24 hours of age to help offset some of the delays being experienced by couriers because of COVID. In December 2022 the optimal time for collection was officially updated to between 24 and 48 hours.

This report discusses the 2022 data using the target ≥95 percent of first samples, taken between 24 and 72 hours after birth, which was officially amended in December 2022.

Interpretation: The national average of first samples taken between 24 and 72 hours after birth was 83.4 percent. Timeliness of sample taking varied between Districts, from 69.4 percent (Lakes District) to 92.2 percent (South Canterbury District) of first samples taken between 24 and 72 hours after birth.

Currently, there are no Districts meeting the target. Māori and Pacific ethnic groups have a higher proportion of samples taken after 72 hours when compared to Asian and Other ethnic groups.

Comment: South Canterbury District has the highest proportion of samples taken between 24 and 72 hours after birth at 92.2 percent. Lakes and Northland Districts had over 24 percent of samples taken after 72 hours after birth (27.6 percent and 24 percent respectively).

The numbers of samples collected between 24 and 48 hours has increased significantly since 2021, from 6.2 to 22.5 percent. However, the percentage of samples collected after 72 hours has not seen corresponding large reductions. In 2020, 17.0 percent of samples were collected after 72 hours and 14.3 percent in 2022.

The national average percentage of samples collected between 24 and 72 hours did see a slight improvement from 2021 to 2022 (from 81.5 to 83.4 percent).

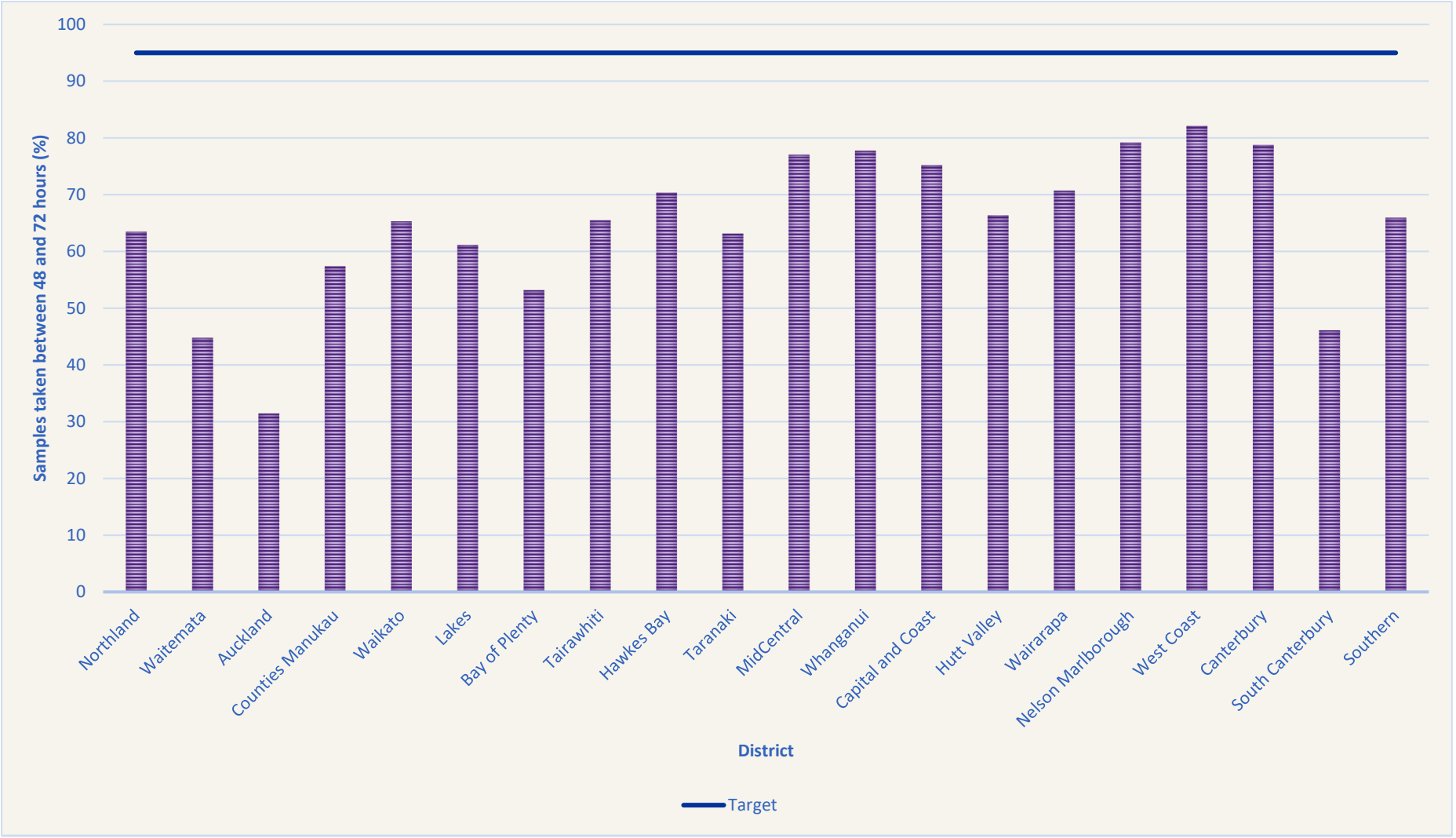
Table 5: Timing of samples taken by ethnicity, January to December 2022

Ethnicity	Less than 24 hours %	Between 24 and 47 hours %	Between 48 and 72 hours %	Greater than 72 hours %	No collection date or time %
Māori	0.2	17.4	61.0	19.2	2.2
Pacific	0.2	23.5	55.8	18.2	2.3
Asian	0.1	30.4	56.9	10.5	2.1
Other	0.2	21.9	63.6	12.2	2.0
Total	**0.2**	**22.5**	**60.9**	**14.3**	**2.1**

Table 6: Timing of samples taken by district, January to December 2022

District	Less than 24 hours		Between 24 and 47 hours		Between 48 and 72 hours		Greater than 72 hours		No collection date or time	
	N	%	N	%	N	%	N	%	N	%
Northland	1	0.0	250	10.7	1,482	63.5	560	24.0	41	1.8
Waitemata	13	0.2	3,247	44.2	3,288	44.8	680	9.3	113	1.5
Auckland	13	0.3	2,875	57.9	1,562	31.5	365	7.4	149	3.0
Counties Manukau	17	0.2	1,657	20.9	4,543	57.4	1,508	19.1	191	2.4
Waikato	4	0.1	502	9.4	3,479	65.3	1,201	22.6	139	2.6
Lakes	1	0.1	125	8.3	924	61.2	417	27.6	44	2.9
Bay of Plenty	6	0.2	660	21.2	1,653	53.2	713	22.9	76	2.4
Tairawhiti	0	0.0	138	19.8	457	65.5	91	13.0	12	1.7
Hawkes Bay	1	0.0	181	8.9	1,425	70.4	384	19.0	34	1.7
Taranaki	1	0.1	263	19.2	863	63.1	214	15.7	26	1.9
MidCentral	5	0.2	244	11.5	1,628	77.0	179	8.5	57	2.7
Whanganui	0	0.0	91	11.4	622	77.8	80	10.0	7	0.9
Capital and Coast	13	0.4	272	9.4	2,177	75.2	360	12.4	72	2.5
Hutt Valley	5	0.3	295	16.3	1,204	66.4	288	15.9	22	1.2
Wairarapa	0	0.0	93	17.6	374	70.7	57	10.8	5	0.9
Nelson Marlborough	5	0.4	131	9.4	1,101	79.2	138	9.9	15	1.1
West Coast	0	0.0	26	8.9	239	82.1	21	7.2	5	1.7
Canterbury	7	0.1	809	12.4	5,119	78.8	443	6.8	120	1.8
South Canterbury	0	0.0	271	46.1	271	46.1	37	6.3	9	1.5
Southern	3	0.1	676	20.2	2,207	65.9	406	12.1	56	1.7
National	**95**	**0.2**	**12,809**	**22.5**	**34,625**	**60.9**	**8,143**	**14.3**	**1,203**	**2.1**

Figure 1: Percent of samples taken between 48 and 72 hours, January to December 2022



Indicator 3: Quality of blood samples

Description: The proportion of samples received by the laboratory that are of satisfactory quality.

Rationale: Accurate testing is reliant on a good quality blood spot sample. Unsatisfactory samples require a repeat sample which could have been avoided. This indicator measures the proportion of blood spot samples that require repeating due to a quality issue.

Target: ≥99 percent of blood spot samples received are of satisfactory quality.

Interpretation: The proportion of satisfactory blood samples ranged from 98.5 percent (Taranaki and Whanganui Districts) to 100 percent (West Coast District) across Districts. The national average was 99.1 percent.

Comment: Overall sample quality improved slightly in 2022, with 0.9 percent (500) of all samples being unsatisfactory compared with 1.0 percent (612) in 2021 and 1.1 percent (636) in 2020.

Sample collection quality, for example, due to insufficient blood on the card, remains the main reason why samples were unsatisfactory, although the number decreased in 2022. The percentage of samples that were unsatisfactory due to being collected early has gone down (from 23.1 percent in 2018) predominantly due to samples collected from 24 hours now being acceptable samples. Each unsatisfactory sample is followed up with a request for a second sample (Indicator 5) to ensure that all babies have been adequately screened.

Table 7: Reason for unsatisfactory sample, January to December 2022

Reason	Number	Percentage
Collection	377	75.4
Timing	74*	14.8
Transport	49	9.8
Total	**500**	**100**

** This number differs to the one in Table 6 as samples collected early from babies with low birth weight are not counted as unsatisfactory samples with repeat samples requested, as second samples are routinely collected within a few days.*

Summary of main reasons:

- **collection:** insufficient blood or the sample was contaminated.
- **timing:** sample was collected too early (before 24 hours of age).
- **transport:** sample took more than one month to arrive; blood was wet when sample card was folded, damaged in transit; or sample was put wet into a plastic bag.

Table 8: Percent of samples of a satisfactory quality, January to December 2022

District	Satisfactory		Unsatisfactory		Total
	N	%	N	%	N
Northland	2,320	99.4	14	0.6	2,334
Waitemata	7,287	99.3	54	0.7	7,341
Auckland	4,926	99.2	38	0.8	4,964
Counties Manukau	7,830	98.9	86	1.1	7,916
Waikato	5,284	99.2	41	0.8	5,325
Lakes	1,493	98.8	18	1.2	1,511
Bay of Plenty	3,078	99.0	30	1.0	3,108
Tairawhiti	691	99.0	7	1.0	698
Hawkes Bay	2,011	99.3	14	0.7	2,025
Taranaki	1,347	98.5	20	1.5	1,367
MidCentral	2,085	98.7	28	1.3	2,113
Whanganui	788	98.5	12	1.5	800
Capital and Coast	2,867	99.1	27	0.9	2,894
Hutt Valley	1,804	99.4	10	0.6	1,814
Wairarapa	524	99.1	5	0.9	529
Nelson Marlborough	1,378	99.1	12	0.9	1,390
West Coast	291	100.0	0	0.0	291
Canterbury	6,451	99.3	47	0.7	6,498
South Canterbury	582	99.0	6	1.0	588
Southern	3,317	99.1	31	0.9	3,348
National	**56,375**	**99.1**	**500**	**0.9**	**56,875**

Indicator 4: Sample dispatch and delivery

Description: The time taken for the sample to be received by the laboratory after being taken.

Rationale: Samples must be received by the laboratory as soon as possible after they are taken.

Target: ≥95 percent of samples are received by the laboratory within four calendar days of being taken.

Interpretation: Timeliness of sample dispatch and delivery varied widely between Districts, ranging from 65.3 percent (West Coast District) to 90.7 percent (Auckland District) of samples received within four days. National timeliness has slightly increased to 79.8 percent of samples received within four days from 78.4 percent in 2021.

Comment: Considerable quality improvement work has been undertaken for this indicator since 2016 and this work is on-going. The result has seen significant increases from the 66 percent achieved in 2014.

The national rate of samples reaching the laboratory within four calendar days of being taken was 79.8 percent in 2022. However, it remains lower than the 83 percent achieved in 2020. COVID-19 was the primary reason for drop in dispatch and delivery timeliness prior to 2022 as courier delivery times increased.

Throughout 2022 there were several severe weather events that caused disruption to roads leading to access issues to a number of regions. It is acknowledged that these events may have had an impact on the timeliness of samples reaching the laboratory. Having no courier pickups or deliveries available on weekends and public holidays also impacts on some samples reaching the laboratory within four calendar days of being taken.

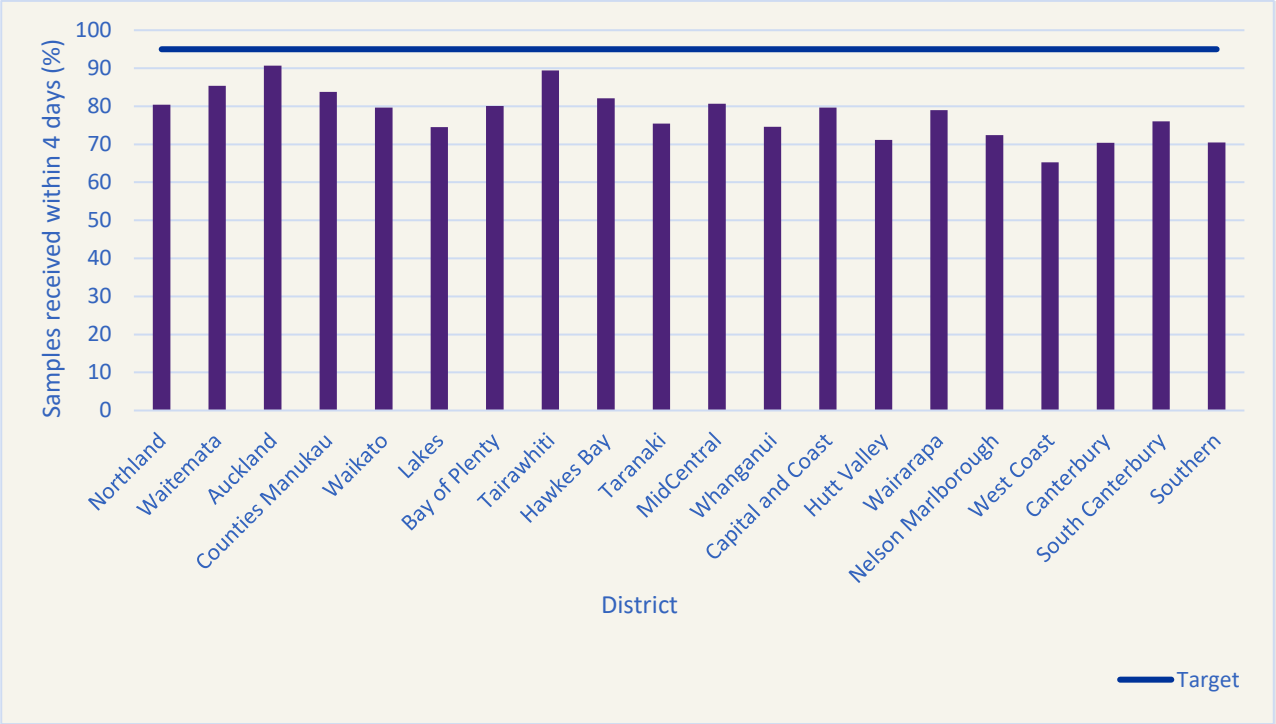
The Antenatal and Childhood Screening team continues to work with Districts to review their processes for getting samples to the laboratory, to develop quality improvement actions, and provide quarterly ‘transit time’ reports as feedback on transit times. To access the transit time reports, go to: https://tewhatuora.shinyapps.io/nsu-nmsp-transittime/

Table 9: Percent of samples received by the laboratory within four days of being taken, January to December 2022

District	Within 4 days		Greater than 4 days		Total
	N	%	N	%	N
Northland	1876	80.4	439	18.8	2334
Waitemata	6267	85.4	1020	13.9	7341
Auckland	4503	90.7	373	7.5	4964
Counties Manukau	6635	83.8	1202	15.2	7916
Waikato	4241	79.6	1026	19.3	5325
Lakes	1126	74.5	366	24.2	1511
Bay of Plenty	2488	80.1	584	18.8	3108
Tairawhiti	624	89.4	72	10.3	698
Hawkes Bay	1663	82.1	349	17.2	2025
Taranaki	1031	75.4	321	23.5	1367
MidCentral	1705	80.7	385	18.2	2113
Whanganui	597	74.6	200	25.0	800
Capital and Coast	2305	79.6	566	19.6	2894
Hutt Valley	1291	71.2	520	28.7	1814
Wairarapa	418	79.0	109	20.6	529
Nelson Marlborough	1006	72.4	376	27.1	1390
West Coast	190	65.3	99	34.0	291
Canterbury	4574	70.4	1865	28.7	6498
South Canterbury	447	76.0	138	23.5	588
Southern	2361	70.5	955	28.5	3348
National	**45363**	**79.8**	**10968**	**19.3**	**56875***

**Totals of columns do not add up as 544 samples had no collection date*

Figure 2: Percent of samples received by the laboratory within four days of being taken by district, January to December 2022



Indicator 5: Receipt and follow-up of second sample

Description: The proportion of second sample requests that had appropriate follow-up (timely receipt of second sample, decline notified or other appropriate follow-up).

Rationale: Second samples are requested if first samples give borderline results or are inadequate. Where requested, second samples should be taken as soon as possible.

Target: 100 percent of second samples requested are received by the laboratory, had other appropriate follow-up, or were declined by parents/guardians, within 10 calendar days of the request.

Interpretation: In 2022, 82.7 percent of requests for second samples resulted in one of the following within 10 days: a second sample arrived at the laboratory; or the laboratory received notification that the parents/guardians had declined the request; or that the newborn had been referred to a specialist, or other appropriate follow-up had occurred; or that the newborn had passed away.

Comment: In 2015 a new protocol was introduced by LabPLUS, which aimed to improve the time second samples were received at LabPLUS. This included sending text messages, making extra phone calls, and providing additional written reports to LMCs. The rate of return within the expected 10-day timeframe has risen from 38 percent in 2014 to 82.7 percent in 2022. There were no adverse clinical consequences from second samples not being received within the 10-day timeframe.

In the 2022 reporting period, 619 second samples were requested. A second sample was received, declined (by the family), or other follow-up occurred in 98.9 percent of the instances when a second sample was requested. For the remaining seven (1.1 percent) second sample requests, multiple attempts were made by the screening lab to contact the LMC or by the LMC to contact families to obtain follow-up samples, without success and the task closed at 28 days. These seven samples were requested as the initial sample was considered an inadequate sample rather than borderline results.

The time taken to receive a follow-up sample is influenced by:

- the time it takes to generate, send, and receive the request (usually reported via text with a request to recollect ASAP);
- the time it takes to collect the second sample (usually at the next scheduled visit of the LMC) and send it to the laboratory; and
- the transit time of the sample to reach the laboratory.

Table 10: Percentage of second samples the laboratory received (or when other appropriate follow-up occurred) within 10 days, January to December 2022

District	Within 10 Days		After 10 Days		Total
	N	%	N	%	N
Northland	10	66.7	5	33.3	15
Waitematā	53	82.8	11	17.2	64
Auckland	44	84.6	8	15.4	52
Counties Manukau	80	81.6	18	18.4	98
Waikato	30	63.8	17	36.2	47
Lakes	13	68.4	6	31.6	19
Bay of Plenty	29	93.5	2	6.5	31
Tairāwhiti	7	77.8	2	22.2	9
Hawkes Bay	15	88.2	2	11.8	17
Taranaki	23	95.8	1	4.2	24
MidCentral	26	83.9	5	16.1	31
Whanganui	9	64.3	5	35.7	14
Capital and Coast	31	91.2	3	8.8	34
Hutt Valley	12	100.0	0	0.0	12
Wairarapa	4	80.0	1	20.0	5
Nelson Marlborough	14	82.4	3	17.6	17
Canterbury	48	85.7	8	14.3	56
South Canterbury	6	85.7	1	14.3	7
Southern	41	95.3	2	4.7	43
Unknown	17	70.8	7	29.2	24
National	**512**	**82.7**	**107**	**17.3**	**619**

Note: small numbers of second samples will have a large effect on percentages

Figure 3: Percentage of second samples the laboratory received (or when other appropriate follow-up occurred) within 10 days, January to December 2022

Indicator 6: Laboratory turnaround time for positive results

Description: The time from receipt of the sample in the laboratory to notification of the referring practitioner or specialist paediatrician of a screen positive result.

Rationale: Timely processing and notification of screen positive samples is essential to ensure early detection and treatment. This indicator is a measure of laboratory performance.

Target: 100 percent of babies with positive results are notified to their LMC / specialist paediatrician by the laboratory within the following timeframes:

Disorder	Calendar days* Clinical critical	Non-clinical critical
Amino acid disorders	2	7
Biotinidase deficiency	-	7
Congenital adrenal hyperplasia	2	7
Cystic fibrosis	-	7
Congenital hypothyroidism	4	7
Fatty acid oxidation disorders	2	7
Galactosaemia	2	7
SCID	-	7

** From receipt in laboratory to notification of screen positives*

Interpretation: A 'clinical critical' screening result indicates a reasonable or high probability of a disorder that can present with severe illness in the early neonatal period, and where a delay of one to two days can affect the outcome.

Overall, 100 percent of clinical critical screen positives, and 98 percent of non-clinical critical screen positives, were notified within the expected timeframes in 2022. This was an improvement from 97.4 percent of clinical critical screen positives, and 95.2 percent of non-clinical critical screen positives in 2021.

Caution should be used due to the relatively low numbers of results being reported. It should be noted that samples that arrive at the laboratory on a Friday afternoon are not tested until Monday morning which has an impact on meeting critical clinical timeframes. A Quality Improvement project to trial undertaking additional testing on a Friday evening was undertaken for three months in 2021. The results were inconclusive as no critical clinical samples were received in the trial time period and it was discontinued.

Comment: In 2022, **all** 'clinical critical' results were reported within the timeframes and 249 of 254 'non-clinical critical' cases were reported within the timeframes.

Non-clinical critical samples received outside of the target turnaround time were due to needing clarification of out-of-range results with second-tier testing and clinical reasons (i.e. borderline results). Borderline newborn screening results are not reported until all results are available on the sample so the notification can include all results in one contact. For example, a borderline hypothyroid result may be available in two days, but if the sample also has a raised immunoreactive trypsin in the cystic fibrosis screen, it is sent for mutation analysis. The laboratory will request a second sample to confirm the thyroid result after the cystic fibrosis mutation result is available.

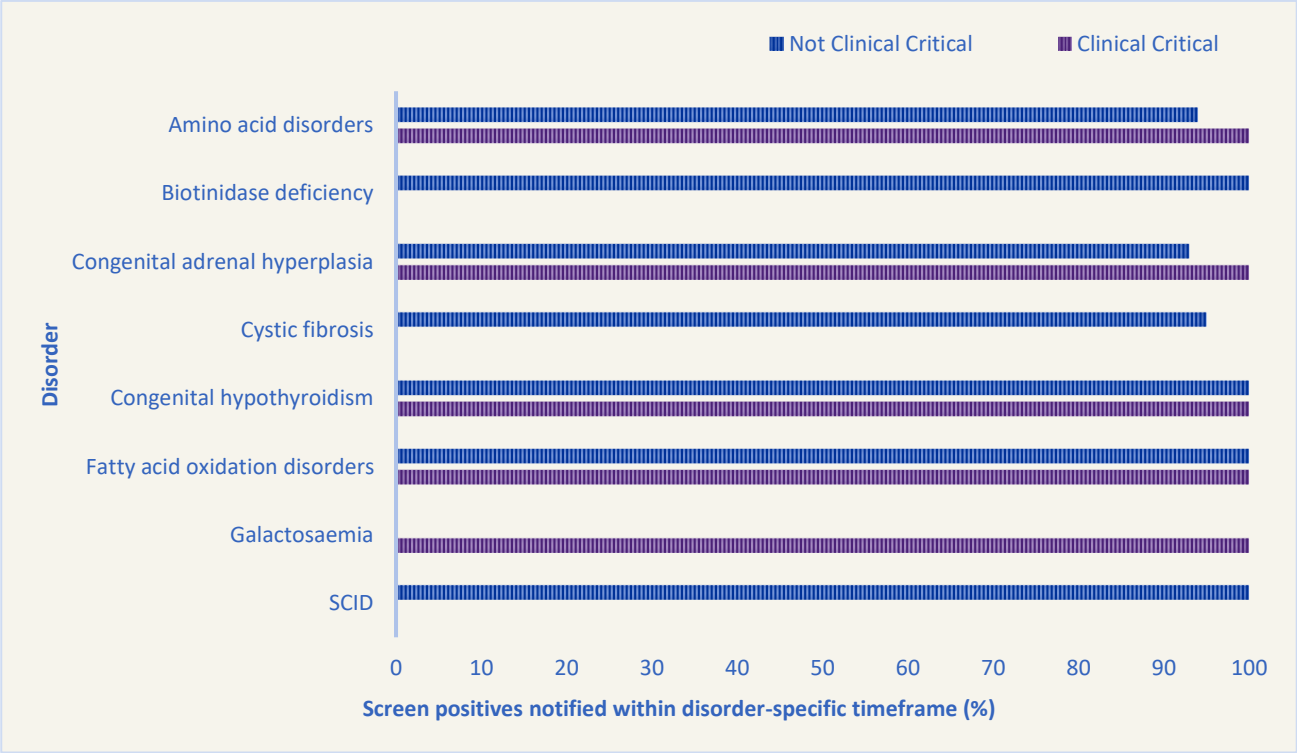
There were no adverse clinical consequences from positive screens reported outside the timeframes.

Table 11: Notification of screen positives by disorder, January to December 2022

Disorder	Timeframe (calendar days)		Total number		TAT met		TAT not met	
	CC	NCC	CC	NCC	CC	NCC	CC	NCC
Amino acid disorders	2	7	**2**	**31**	2	29	0	2
Biotinidase deficiency	-	7	**-**	**18**	-	18	-	0
Congenital adrenal hyperplasia	2	7	**4**	**14**	4	13	0	1
Cystic fibrosis	-	7	**-**	**39**	-	37	-	2
Congenital hypothyroidism	4	7	**12**	**115**	12	115	0	0
Fatty acid oxidation disorders	2	7	**10**	**2**	10	2	0	0
Galactosaemia	2	7	**1**	**0**	1	0	0	0
SCID	-	7	**-**	**35**	-	35	-	0
		Total	**29**	**254**	**29**	**249**	**0**	**5**
		Percentage			**100%**	**98%**	**0%**	**2%**

TAT: Turn Around Time; CC: Clinical Critical; NCC: Not Clinical Critical

Figure 4: Percentage of screen positives the laboratory notified within the disorder-specific timeframe, January to December 2022



Indicator 7: Age of receipt into clinical care

Description: For babies with screened conditions, the age of the baby at transfer into clinical care.

Rationale: To ensure babies with congenital disorders have their development potential affected as little as possible, all babies with a screened condition must receive a confirmed diagnosis and timely commencement of treatment/active clinical management.

Target: 100 percent of babies who receive a screen positive result and are diagnosed with a screened condition receive active clinical management by the following timeframes:

Disorder	Age of baby in days	
	Clinical critical	Non-clinical critical
Amino acid disorders	10	28
Biotinidase deficiency	-	28
Congenital adrenal hyperplasia	10	28
Cystic fibrosis	-	28
Congenital hypothyroidism	10	28
Fatty acid oxidation disorders	10	28
Galactosaemia	10	28
SCID	-	14

Interpretation: The disorder-specific timeframe was met for 20 out of 21 cases identified as clinical critical: 30 of 31 non-clinical critical cases were received into clinical care within the specified timeframe.

Comment: 95 percent of babies with disease detected following a positive newborn screen were received into clinical care within an acceptable timeframe, compared to 94 percent in 2021 and 100 percent in 2019 and 2020.

The two cases that were detected and referred into clinical care outside of timeframe primarily occurred due to late sample collection and delayed transit times. Optimal neuro-cognitive outcomes in Congenital Hypothyroidism are achieved through early detection and treatment. The potential impacts of delayed diagnoses depend on both disease severity and age at detection and are likely to be minimal for these cases.

Table 12: Timeframe met for receipt into clinical care after confirmed diagnosis, January to December 2022

Disorder	Timeframe (calendar days)		Total number		Timeframe met		Timeframe not met	
	CC	NCC	CC	NCC	CC	NCC	CC	NCC
Amino acid disorders	10	28	**0**	**6**	0	6	0	0
Biotinidase deficiency	-	28	**-**	**0**	-	0	-	0
Congenital adrenal hyperplasia	10	28	**3**	**0**	3	0	0	0
Cystic fibrosis*	-	28	**-**	**8**	-	8	-	-
Congenital hypothyroidism**	10	28	**14**	**14**	13	13	1	1
Fatty acid oxidation disorders	10	28	**3**	**2**	3	2	0	0
Galactosaemia	10	28	**1**	**0**	1	0	0	0
SCID	-	14	**-**	**1**	-	1	-	0
		Total	**21**	**31**	**20**	**30**	**1**	**1**
		Percentage			**95%**	**97%**	**5%**	**3%**

CC: Clinical Critical; NCC: Not Clinical Critical

** The Cystic Fibrosis (CF) case count includes 2 cases labelled Cystic Fibrosis Screen Positive, Inconclusive Diagnosis (CFSPID), ie an asymptomatic infant with a positive newborn screen result for CF but an indeterminate diagnosis. Children in this category are followed until 6 years of age to determine a final outcome, ie CF or not CF.*

***The Congenital Hypothyroidism (CH) count excludes an additional 15 babies with CH detected through routine repeat card collection as per the Low Birth Weight (LBW) protocol. Preterm babies with CH are frequently not detected through screening samples collected at 48 hours, so additional scheduled samples are collected. Of the 15 babies, none were detectable on a routine 48 hour sample, 13 were detected from a scheduled two week sample and two from a scheduled four week sample.*

Indicator 8: Positive predictive value of the screening test

Description: The probability of a baby having a positive diagnosis for a screened condition given a positive screening result for that condition.

Rationale: Positive predictive value (PPV) is a measure of the performance of the screening test. A low PPV means many babies without a screened condition will be referred for diagnostic testing, this brings associated costs and anxiety for families. Reporting of PPV helps to monitor the potential harm of the programme due to identification of false positives through screening.

Target: None.

Interpretation: The PPV is affected by the sensitivity and specificity of a test, as well as the prevalence of a condition, and as a result the PPV is likely to vary by condition tested for in newborn blood screening.

The PPV for individual disorders is presented as five-year rolling data because the number of cases varies significantly year on year. Overall, there is a 25.7% likelihood that a baby with a positive screen will be confirmed to have a target disorder. However, there is variation depending on the specific condition and associated test (as seen in Table 13).

Comment: Overall, the PPV of the screening test has increased notably by over 10 percent from the 2017 annual report (14.7 percent) to 2022 annual report (25.7 percent). PPV improvement has occurred due to introduction of second-tier tandem tests and the improved protocols for some other disorders. Comparing the 5-year averages of 2013-2017 to 2018-2022 the sensitivity and specificity are nearly identical with sensitivity decreasing slightly from 98.9 to 98.6 percent and specificity increasing from 99.4 to 99.7 percent.

Table 13: Positive predictive value of the screening test, January 2018 to December 2022

Condition	Babies Screened	Positive tests	True Positive	False Positive	False Negative	True Negative	Sensitivity (%)	Specificity (%)	PPV (%)
Amino acid disorders	293306	149	18	131	0	293031	100	99.96	12.1
Biotinidase deficiency	293306	54	3	51	0	293252	100	99.98	5.6
Congenital Adrenal Hyperplasia	293306	132	13	119	0	293174	100	99.96	9.8
Cystic Fibrosis	293306	248	79	169	0	293048	100	99.94	31.9
Congenital Hypothyroidism	293306	502	190	312	2	292902	98.96	99.89	37.8
Fatty acid oxidation disorders	293306	62	31	31	0	293244	100	99.99	50.0
Galactosaemia	293306	34	3	31	3	293271	50	99.99	8.8
SCID	293306	152	5	147	0	293154	100	99.95	3.3
Total	**293306**	**1333**	**342**	**991**	**5**	**291968**	**98.56**	**99.66**	**25.7**

Appendix 1: List of screened conditions

Amino acid disorders
Phenylketonuria
Maple syrup urine disease
Argininosuccinic aciduria (argininosuccinate lyase deficiency)
Citrullinaemia (argininosuccinate synthetase deficiency)
Glutaric acidaemia type I (glutaryl-CoA dehydrogenase deficiency)
Homocystinuria (cystathionine beta-synthase deficiency)
Isovaleric acidaemia (isovaleryl-CoA dehydrogenase deficiency)
Methylmalonic acidurias (mutase deficiency, CblA, CblB, CblC, CblD defects)
Propionic acidaemia (propionyl-CoA carboxylase deficiency)
Fatty acid oxidation disorders
CACT (carnitine acylcarnitine translocase deficiency)
CPT-I (carnitine palmitoyltransferase-I deficiency)
CPT-II (carnitine palmitoyltransferase-II deficiency)
LCHAD (3-hydroxy long-chain acyl-CoA dehydrogenase deficiency)
TFP (trifunctional protein deficiency)
MADD (multiple acyl-CoA dehydrogenase deficiency)
MCAD (medium-chain acyl-CoA dehydrogenase deficiency)
VLCAD (very-long-chain acyl-CoA dehydrogenase deficiency)
Additional disorders
Congenital hypothyroidism
Congenital adrenal hyperplasia
Cystic fibrosis
Biotinidase deficiency
Galactosaemia
Severe combined immunodeficiency (SCID)