MINISTRY OF HEALTH
MANATŪ HAUORA

New Zealand Government

Newborn Metabolic Screening Programme

Annual Report

January to December 2017

National Screening Unit

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Disclaimer

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Contents

List of Figures

List of Tables

Introduction

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

This is the first annual report of the NMSP after the release of the new monitoring indicators document in February 2018 and the seventh annual report following the development of national indicators in 2010. The NMSP Monitoring Framework and monitoring reports are published on the National Screening Unit (NSU) website: www.nsu.govt.nz/health-professionals/newborn-metabolic-screening-programme/procedures-guidelines-and-reports-2

Background to the Programme

The aim of the NMSP is to reduce morbidity and mortality associated with specific congenital metabolic disorders by screening newborns to detect the conditions before life-threatening illness or developmental delays occur. Since 1969 almost all newborns in New Zealand have been screened by the programme. Currently the NMSP identifies about 50 newborns a year with a metabolic disorder and treatment is commenced.

A midwife, nurse, phlebotomist or doctor collects a blood sample from the newborn's heel onto a blood spot card (a Guthrie card). Samples must be collected between 48 and 72 hours of age for optimal testing. Cards are sent urgently to LabPlus at Auckland District Health Board (ADHB) for analysis and reporting of results to appropriate clinicians. Blood spot samples are screened for the 25 metabolic disorders listed in Appendix A.

Since 2005, the NMSP has been overseen nationally by the NSU at the Ministry. A significant milestone for the programme was the introduction in 2006 of expanded newborn screening, adding fatty acid oxidation and more amino acid breakdown disorders to the screening panel. Screening for Severe Combined Immuno-deficiency (SCID) was added in December 2017.

Data summary

Screening data is sourced from LabPlus at ADHB for all blood spot cards received in the 2017 calendar year. Birth data in the 2017 calendar year is sourced from the National Maternity Collection at the Ministry. Ethnicity data is prioritised in accordance with Statistics New Zealand's prioritised ethnicity model which is standard across the health sector. When a newborn's District Health Board (DHB) of domicile is unknown, it is set to 'Unknown'.

Executive summary

1. Of the 59,517 babies born in 2017, 58,935 were screened by the NMSP; a national coverage rate of 99.0%, which is in line with coverage rates since the programme began in 1969. However, there was variance at a local DHB level, with coverage rates ranging from 94.3% (Tairāwhiti) to 101% (Nelson Marlborough).

2. In 2017 coverage varied by ethnic group, with 98.0% of Māori, 98.1% of Pacific, and 99.6% of newborns of all other ethnicities screened. From 2017 DHBs have been increasingly encouraged to match their birth data and babies screened data to ensure all consented babies are screened.

3. The congenital disorders screened for by the NMSP are rare. In 2017 41 newborns were diagnosed with a screened disorder compared to 48 in 2016.

4. The NMSP monitors timeframes along the screening pathway, from collection of blood spot samples through to clinical handover for care if needed, to ensure that newborns diagnosed with a screened condition are treated as soon as possible. While laboratory testing timeframes were uniformly high, as in previous years few of the general timeframe standards were met in 2017.

5. Blood spot cards are expected to arrive at the laboratory within four days of sampling. In 2017 79% arrived in the timeframe. The national standard is 95%. This shortfall is a known and longstanding issue that, since 2015, has been the focus of quarterly 'transit time' reports to DHBs, to prompt a process quality improvement focus. The result has been a 13% lift in the four day transit rate, from 66% to 79% over the two years between 2014 and 2017. Also, higher volume maternity units are now shifting to using courier services, which is expected to improve transit rates further.

6. A phone and text service between LabPlus and Lead Maternity Carers (LMCs), aimed at improving the turnaround time of requests for second samples was introduced in 2016. The rate of return within the expected 10-day timeframe has risen 33% over two years, from 38% in 2014 to 71% in 2017. There has been a 2% drop in the return rate from 73% in 2016. It is planned to systematically follow-up non responses from LMCs in 2017/18.

7. In 2016 the NSU, in conjunction with the programme's lead paediatricians and laboratory scientists, started a review of the monitoring indicators. The revision was completed in February 2018, and this annual report will use the updated indicators.

Indicator 1: Coverage

Description: Monitoring the proportion of newborns in New Zealand who complete newborn metabolic screening.

Rationale: Newborn screening must be offered for all newborns. All newborns whose parent/guardians consent to screening should be screened.

Standard: 99% of babies born nationally and within each of Maori, Pacific, Asian and Other population groups are screened.

Interpretation: Coverage at 99.0% is in line with an average of 99.0% between 2007 and 2017. Coverage by DHB varied from 94.3% upward. Coverage by ethnicity varied from 98.0% for Māori and Pacific (98.1%), to 99.6% for Other.

Comment: Overall programme coverage remained high, with one large DHB (Nelson Marlborough) achieving more than 100% coverage. Tairāwhiti DHB had the lowest coverage rate of 94.3%.

In 2017, 582 newborns were not screened by the NMSP. Of those, 310 (54%) of those were from four DHBs (Counties Manukau, Bay of Plenty, Canterbury and Waitemata DHBs), with 127 from Counties Manukau alone. It is not yet possible to distinguish between the few newborns who are unscreened due to parents/guardians withholding consent and those not screened because they are missed altogether. Some DHBs have begun to actively identify and follow up on their unscreened newborns. National Women's Health at Auckland DHB now regularly matches birth and screened data. Waikato, Tairawhiti, and Taranaki DHBs have begun using the National Child Information Platform (NCHIP) application for the same purpose.

Coverage rates for Māori are lower than the general population at 15 DHBs, particularly so at Tairāwhiti DHB. This ought to improve with increased matching of birth and screening data to identify unscreened newborns.

As in previous years, there was some non-alignment of denominator data (birth volumes) with numerator data (newborns screened). Reasons include: the indicator reports DHB of domicile when many newborns (particularly in Auckland) are born and/or screened at a different DHB to where they live; and birth year and screened year can be different. Cross-matching and data cleansing to overcome these problems continues to improve, meaning that DHB coverage rates are in 2017 are likely to be more accurate than in the past.

Figure 1: Coverage over time

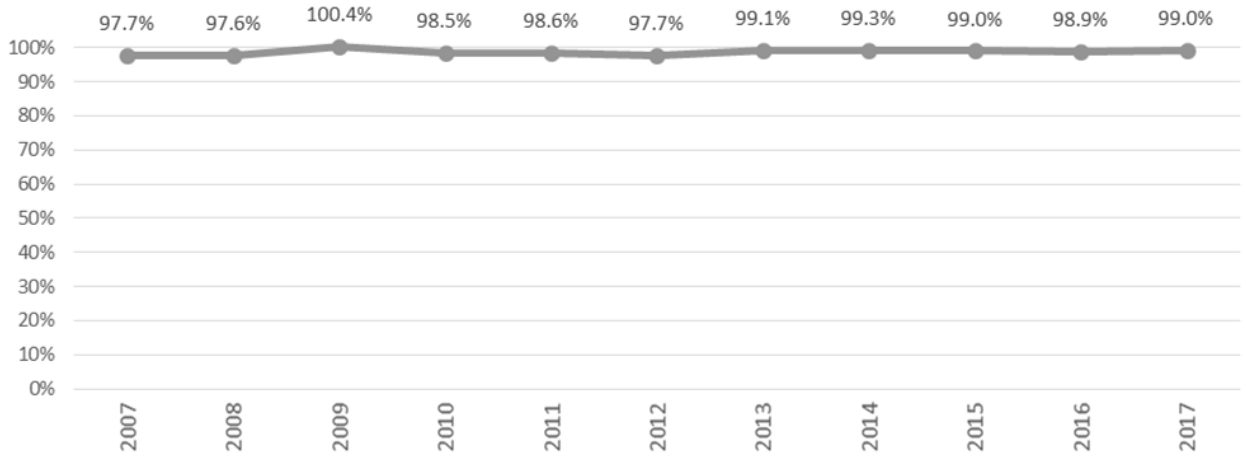


Table 1: Coverage over time

Year	Births	Babies screened	Coverage
2007	64,040	65,121	97.7%
2008	65,333	63,794	97.6%
2009	63,285	63,516	100.4%
2010	64,699	63,727	98.5%
2011	62,733	61,859	98.6%
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%

Figure 2: Coverage by DHB of domicile, January to December 2017

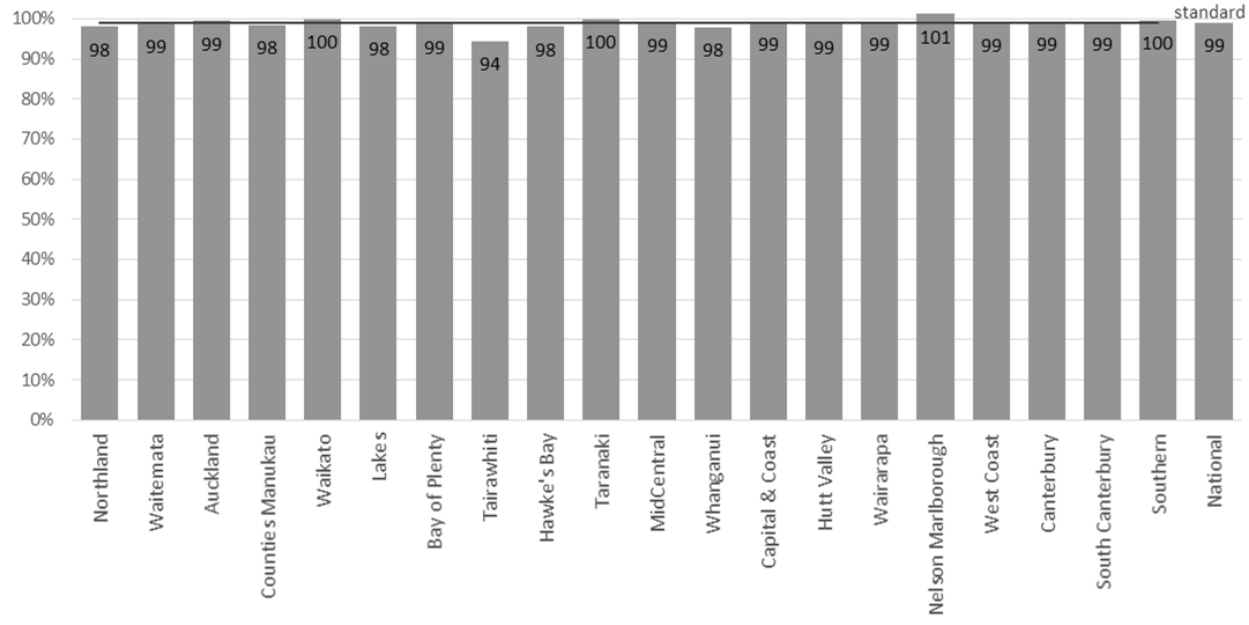


Table 2: Coverage by DHB of domicile, January to December 2017

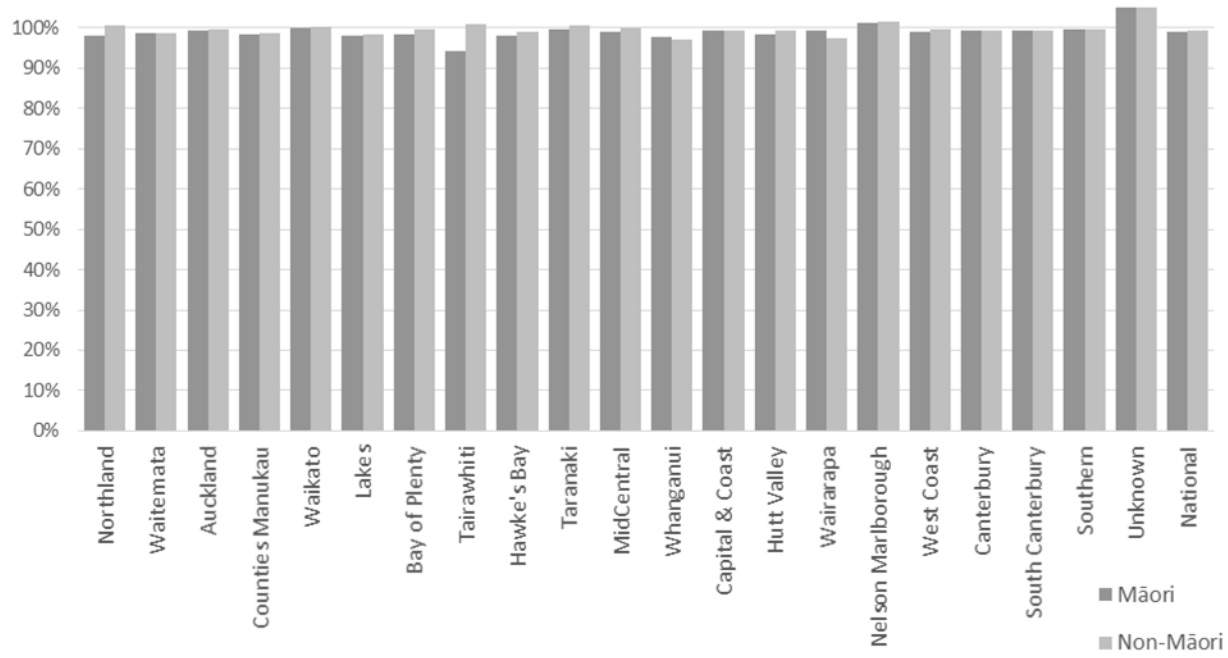
DHB of domicile	Births	Newborns screened	Newborns unscreened	Coverage
Northland	2,221	2,177	44	98.0%
Waitemata	7,738	7,647	91	98.8%
Auckland	5,671	5,636	35	99.4%
Counties Manukau	8,340	8,213	127	98.5%
Waikato	5,354	5,350	4	99.9%
Lakes	1,552	1,523	29	98.1%
Bay of Plenty	3,088	3,043	45	98.5%
Tairawhiti	706	666	40	94.3%
Hawke's Bay	2,134	2,091	43	98.0%
Taranaki	1,419	1,416	3	99.8%
MidCentral	2,136	2,116	20	99.1%
Whanganui	847	829	18	97.9%
Capital & Coast	3,496	3,472	24	99.3%
Hutt Valley	1,957	1,928	29	98.5%
Wairarapa	510	506	4	99.2%
Nelson Marlborough	1,418	1,434		*
West Coast	354	351	3	99.2%
Canterbury	6,421	6,374	47	99.3%
South Canterbury	633	628	5	99.2%
Southern	3,445	3,430	15	99.6%
Unknown	77	105		*
National	**59,517**	**58,935**	**582**	**99.0%**

* Percentages greater than 100% are suppressed because of a mismatch between numerator and denominator data due to such things as: newborns are not always born or screened in their DHB of domicile, year of birth and year of screening are not always the same.

Table 3: Coverage by ethnicity, January to December 2017

Ethnicity	Births	Babies screened	Coverage
Māori	16,284	15,966	98.0%
Pacific	6,002	5,886	98.1%
Other	37,231	37,083	99.6%
Total	**59,517**	**58,935**	**99.0%**

Figure 3: Coverage rate ratio* by DHB of domicile and ethnicity Māori / non-Māori, January to December 2017



* 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 to non-Māori.

Table 4: Coverage by DHB of domicile and ethnicity

DHB of domicile	Māori		Non-Māori		Total		Ratio
Northland	1,239	98%	938	101%	2,177	98%	0.98
Waitemata	1,295	99%	6,352	99%	7,647	99%	0.98
Auckland	632	99%	5,004	100%	5,636	99%	0.98
Counties Manukau	1,876	98%	6,337	99%	8,213	98%	0.98
Waikato	2,083	100%	3,267	100%	5,350	100%	0.97
Lakes	846	98%	677	98%	1,523	98%	0.99
Bay of Plenty	1,285	99%	1,758	100%	3,043	99%	0.99
Tairawhiti	458	94%	208	101%	666	94%	0.93
Hawkes Bay	936	98%	1,155	99%	2,091	98%	0.94
Taranaki	483	100%	933	101%	1,416	100%	0.97
MidCentral	747	99%	1,369	100%	2,116	99%	0.96
Whanganui	396	98%	433	97%	829	98%	1.01
Capital and Coast	641	99%	2,831	99%	3,472	99%	0.97
Hutt Valley	522	99%	1,406	99%	1,928	99%	1.01
Wairarapa	180	99%	326	98%	506	99%	1.00
Nelson Marlborough	350	101%	1,084	102%	1,434	101%	1.02
West Coast	81	99%	270	100%	351	99%	1.01
Canterbury	1,106	99%	5,268	99%	6,374	99%	0.97
South Canterbury	129	99%	499	99%	628	99%	1.01
Southern	660	100%	2,770	100%	3,430	100%	1.00
Unknown	21	136%	84	153%	105	136%	0.69
National	**15,966**	**99%**	**42,969**	**99%**	**58,935**	**99%**	**0.98**

Indicator 2:
Timing of sample taking

Description: Monitoring 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 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. The optimum window for sample collection is between 48 and 72 hours after birth.

Standard: 95% of first samples are taken between 48 and 72 hours after birth.

Interpretation: Timeliness of sample taking varied from 65% (Waikato) to 90% (Canterbury) between DHBs, with a national average of 79%, compared to 78% in 2016. 17% of samples were taken too late, and 1% too early.

Comment: Canterbury DHB continues to perform best. Counties Manukau, Waikato, Bay of Plenty and Lakes DHBs lag in meeting the standard due to the number of their samples being taken late. It is expected that this will progressively improve when DHBs review all their internal blood spot card processes and timeframes, including sample taking time, as is expected as part of the current roll-out of courier services to higher-volume maternity units.

Figure 4: Percentage of samples taken between 48 and 72 hours, January to December 2017

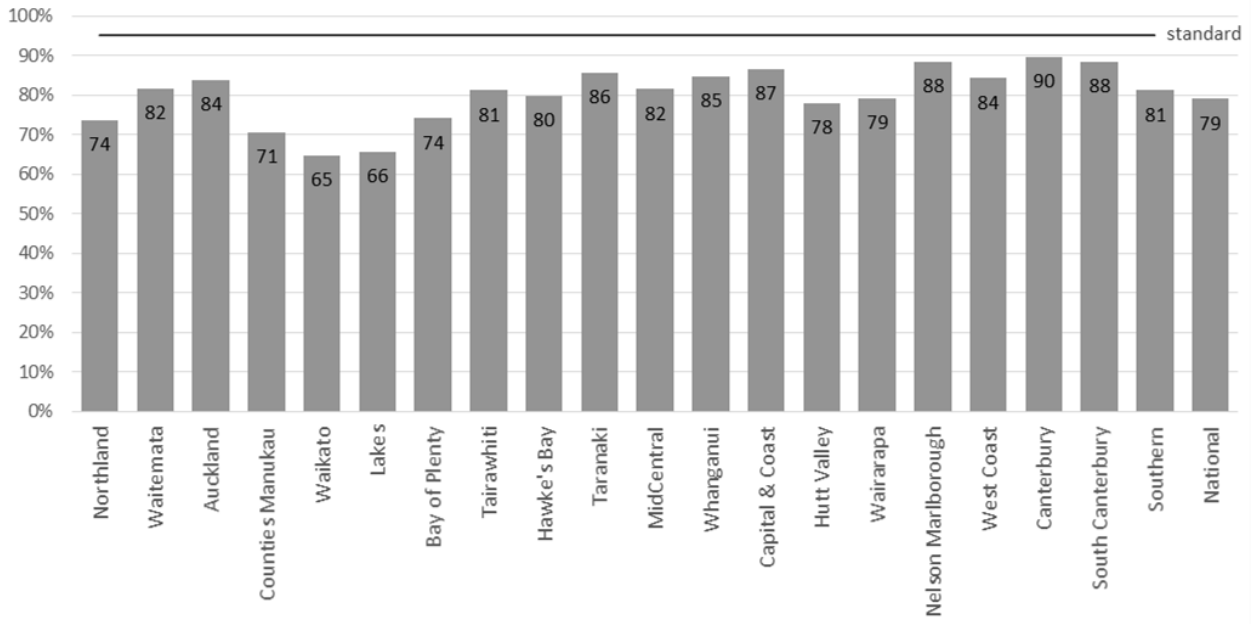


Table 5: Timing of sample taking, January to December 2017

DHB of domicile	Less than 48 hours		48 to 72 hours		More than 72 hours		Unknown		Total
	no.	%	no.	%	no.	%	no.	%	no.
Northland	30	1%	1,603	74%	492	23%	52	2%	2,177
Waitemata	83	1%	6,240	82%	1,175	15%	149	2%	7,647
Auckland	61	1%	4,722	84%	653	12%	200	4%	5,636
Counties Manukau	62	1%	5,799	71%	2,101	26%	251	3%	8,213
Waikato	44	1%	3,469	65%	1,645	31%	192	4%	5,350
Lakes	11	1%	1,002	66%	473	31%	37	2%	1,523
Bay of Plenty	27	1%	2,258	74%	678	22%	80	3%	3,043
Tairawhiti	4	1%	541	81%	112	17%	9	1%	666
Hawke's Bay	21	1%	1,669	80%	369	18%	32	2%	2,091
Taranaki	16	1%	1,214	86%	153	11%	33	2%	1,416
MidCentral	36	2%	1,726	82%	300	14%	54	3%	2,116
Whanganui	9	1%	702	85%	103	12%	15	2%	829
Capital & Coast	37	1%	3,010	87%	351	10%	74	2%	3,472
Hutt Valley	11	1%	1,505	78%	367	19%	45	2%	1,928
Wairarapa	6	1%	401	79%	83	16%	16	3%	506
Nelson Marlborough	18	1%	1,268	88%	126	9%	22	2%	1,434
West Coast	4	1%	296	84%	46	13%	5	1%	351
Canterbury	87	1%	5,721	90%	396	6%	170	3%	6,374
South Canterbury	7	1%	555	88%	55	9%	11	2%	628
Southern	36	1%	2,788	81%	544	16%	62	2%	3,430
Unknown	1	1%	80	76%	13	12%	11	10%	105
National	**611**	**1%**	**46,569**	**79%**	**10,235**	**17%**	**1,520**	**3%**	**58,935**

Indicator 3: Quality of blood samples

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

Rationale: Accurate testing of newborn metabolic screening samples is reliant on the quality of the sample. Unsatisfactory samples require a repeat sample which could have been avoided.

Standard: 99% of samples are of satisfactory quality.

Interpretation: The proportion of blood samples that were satisfactory ranged from 98.0% to 99.6% across DHBs, with a national average of 98.7%.

Comment: While only three DHBs met the standard (Auckland, Tairāwhiti and Wairarapa), overall sample quality improved nationally in 2017, with 1.3% (743) of all samples being unsatisfactory as against 1.5% (892) in 2016. In 2017/18 DHBs with unusually high volumes of unsatisfactory samples will be asked to identify and address the causes.

Sample collection quality, such as insufficient blood on the card, remains the main reason for unsatisfactory samples. There was a 1% decrease in transport related unsatisfactory samples between 2016 (9%) and 2017 (8%). Each unsatisfactory sample is followed up with a request for a second sample (Indicator 5) to reduce the risk to the babies affected.

Table 6: Percentage of samples of a satisfactory quality, January to December 2017

DHB of domicile	Satisfactory		Unsatisfactory		Total
	no.	%	no.	%	no.
Northland	2,134	98.0%	43	2.0%	2,177
Waitemata	7,543	98.6%	104	1.4%	7,647
Auckland	5,579	99.0%	57	1.0%	5,636
Counties Manukau	8,082	98.4%	131	1.6%	8,213
Waikato	5,291	98.9%	59	1.1%	5,350
Lakes	1,507	98.9%	16	1.1%	1,523
Bay of Plenty	3,011	98.9%	32	1.1%	3,043
Tairawhiti	661	99.2%	5	0.8%	666
Hawke's Bay	2,061	98.6%	30	1.4%	2,091
Taranaki	1,401	98.9%	15	1.1%	1,416
MidCentral	2,080	98.3%	36	1.7%	2,116
Whanganui	819	98.8%	10	1.2%	829
Capital & Coast	3,430	98.8%	42	1.2%	3,472
Hutt Valley	1,906	98.9%	22	1.1%	1,928
Wairarapa	504	99.6%	2	0.4%	506
Nelson Marlborough	1,417	98.8%	17	1.2%	1,434
West Coast	347	98.9%	4	1.1%	351
Canterbury	6,304	98.9%	70	1.1%	6,374
South Canterbury	619	98.6%	9	1.4%	628
Southern	3,392	98.9%	38	1.1%	3,430
Unknown	104	99.0%	1	1.0%	105
National	**58,192**	**98.7%**	**743**	**1.3%**	**58,935**

Collection: insufficient blood, incomplete demographics on the card, or the sample was contaminated.

Timing: samples were collected too early (before 48 hours of age).

Transport: took more than one month to arrive, blood was wet when folded, damaged in transit, or put wet into a plastic bag.

Table 6: Reason for unsatisfactory samples, January to December 2017

Reason	no.	%
Collection	508	68.4%
Timing	175	23.6%
Transport	58	7.8%
Error	2	0.3%
Total	**743**	**100.0%**

Indicator 4: Sample dispatch and delivery

Description: Monitoring the time between the sample being taken and receipt by the laboratory.

Rationale: To ensure early diagnosis and treatment, samples must be received by the laboratory as soon as possible after being taken.

Standard: 95% of samples are received at the laboratory within four (calendar) days of being taken.

Interpretation: Timeliness of sample dispatch and delivery varied widely between DHBs, ranging from 58% to 91% meeting the standard. While the national average of 78% has slightly increased from the 76% in 2016, there was significant improvement in rates at Tairāwhiti and Hawke's Bay (13%) and Capital and Coast (16%) DHBs, offset by decreases at South Canterbury (-11%) and Bay of Plenty (-5%) DHBs.

Comment: As in 2016, this indicator remained the focus of considerable quality improvement work. The NSU continued to provide DHBs with quarterly 'transit' reports, for feedback on transit time turnaround. Variances in postal service provision remained an issue, compounded by unexpected natural events such as the Kaikoura earthquake in November 2016. These variables impact on DHBs' ability to achieve the 95% standard, and the impacts vary significantly across the country.

Improving blood spot card transit times by taking a dedicated process improvement approach can make a real positive difference, as has been illustrated over recent years by improved transit times from National Women's Health and Birthcare Auckland (ADHB) and Botany Downs Primary Birthing Unit (Counties Manukau DHB). Promotion of this approach, together with the progressive roll out of courier to replace FastPost of blood spot cards from main maternity units nationwide (commenced in late 2016), is expected to lead to improvement across all DHBs.

Figure 5: Percentage of samples received by the laboratory within four days of being taken, January to December 2017

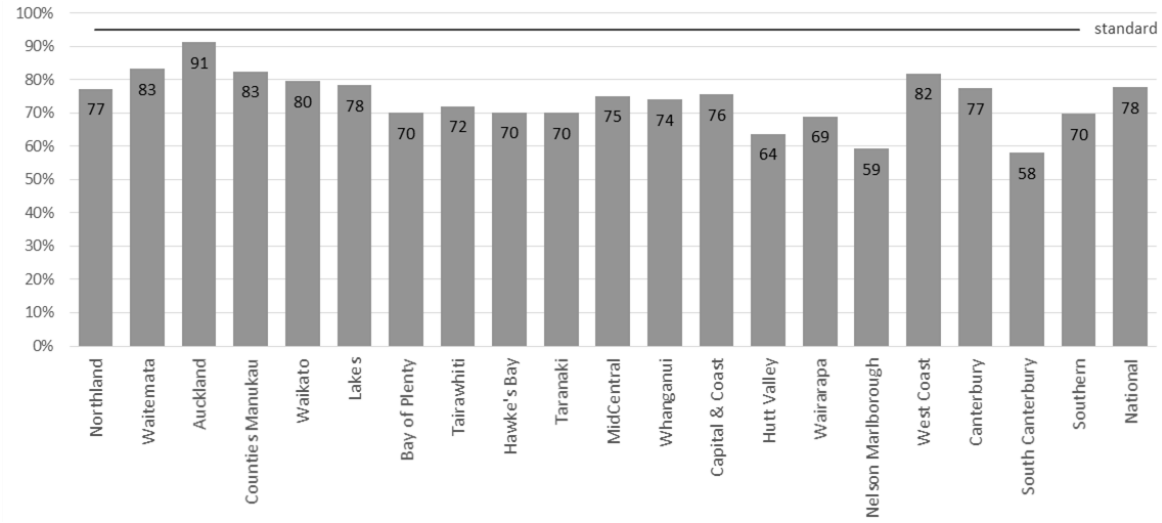


Table 7: Percentage of samples received by the laboratory within four days of being taken, January to December 2017

DHB of domicile	Within 4 days		More than 4 days		Unknown		Total
	no.	%	no.	%	no.	%	no.
Northland	1,683	77%	477	22%	17	1%	2,177
Waitemata	6,364	83%	1,215	16%	68	1%	7,647
Auckland	5,141	91%	418	7%	77	1%	5,636
Counties Manukau	6,776	83%	1,353	16%	84	1%	8,213
Waikato	4,257	80%	1,025	19%	68	1%	5,350
Lakes	1,192	78%	317	21%	14	1%	1,523
Bay of Plenty	2,132	70%	875	29%	36	1%	3,043
Tairawhiti	479	72%	184	28%	3	0%	666
Hawke's Bay	1,468	70%	604	29%	19	1%	2,091
Taranaki	992	70%	413	29%	11	1%	1,416
MidCentral	1,590	75%	498	24%	28	1%	2,116
Whanganui	613	74%	209	25%	7	1%	829
Capital & Coast	2,624	76%	815	23%	33	1%	3,472
Hutt Valley	1,229	64%	680	35%	19	1%	1,928
Wairarapa	349	69%	150	30%	7	1%	506
Nelson Marlborough	850	59%	577	40%	7	0%	1,434
West Coast	287	82%	64	18%	0	0%	351
Canterbury	4,934	77%	1,341	21%	99	2%	6,374
South Canterbury	364	58%	261	42%	3	0%	628
Southern	2,392	70%	1,004	29%	34	1%	3,430
Unknown	84	80%	18	17%	3	3%	105
National	**45,800**	**78%**	**12,498**	**21%**	**637**	**1%**	**58,935**

Indicator 5: Collection and follow-up of second samples

Description: Monitoring the follow-up of requests for second blood spot samples when the original sample is either unsuitable for testing or gives a borderline result.

Rationale: If a second sample is required it means that a sample was not adequate, or results were borderline. Second samples should be taken as soon as possible so that the newborn can be treated early if they have a disorder.

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

Interpretation: In 2017 71% of requests for second samples resulted in either a second sample arriving at the laboratory, or notification that the parents/guardians had declined the request, or that the newborn had been referred to a specialist, or had died. In the reporting period, a second sample was received, declined, or had other follow-up at some stage in 97% of the instances when a second sample was requested.

Comment: The time taken to receive a follow-up sample is influenced by: the time it takes to generate, send and receive the request; and the time it takes for the second sample to be collected (usually at the next scheduled LMC visit), sent to and received by the laboratory.

In line with the improvement in the quality of blood spot samples received at the laboratory (Indicator 3), there was a decline in the need to request second samples. In 2014 there were 1,352 requests, with 1,171 in 2015, 988 in 2016, and 998 in 2017. Also, in May 2015 a new protocol for follow-up samples was introduced along with phone and text requests from LabPlus to LMCs to supplement the usual paper reports per request, and regular reminders. Between 2014 and 2017 this resulted in a 33% improvement, from 38% to 71%, in the 10 day turnaround time of second samples. The LabPlus staff's initiative with this quality improvement was recognised with an Auckland DHB Excellence Award in 2016.

There has been a 2% drop in the return rate from 73% in 2016. Waitemata, Counties Manukau and Waikato DHBs had more than half (23) of the 41 requests for second samples that drew no response in 2017. It is planned to systematically follow-up non-responses from LMCs in 2017/18.

Figure 6: Percentage of second samples received (or other appropriate follow-up occurred) within 10 days, January to December 2017

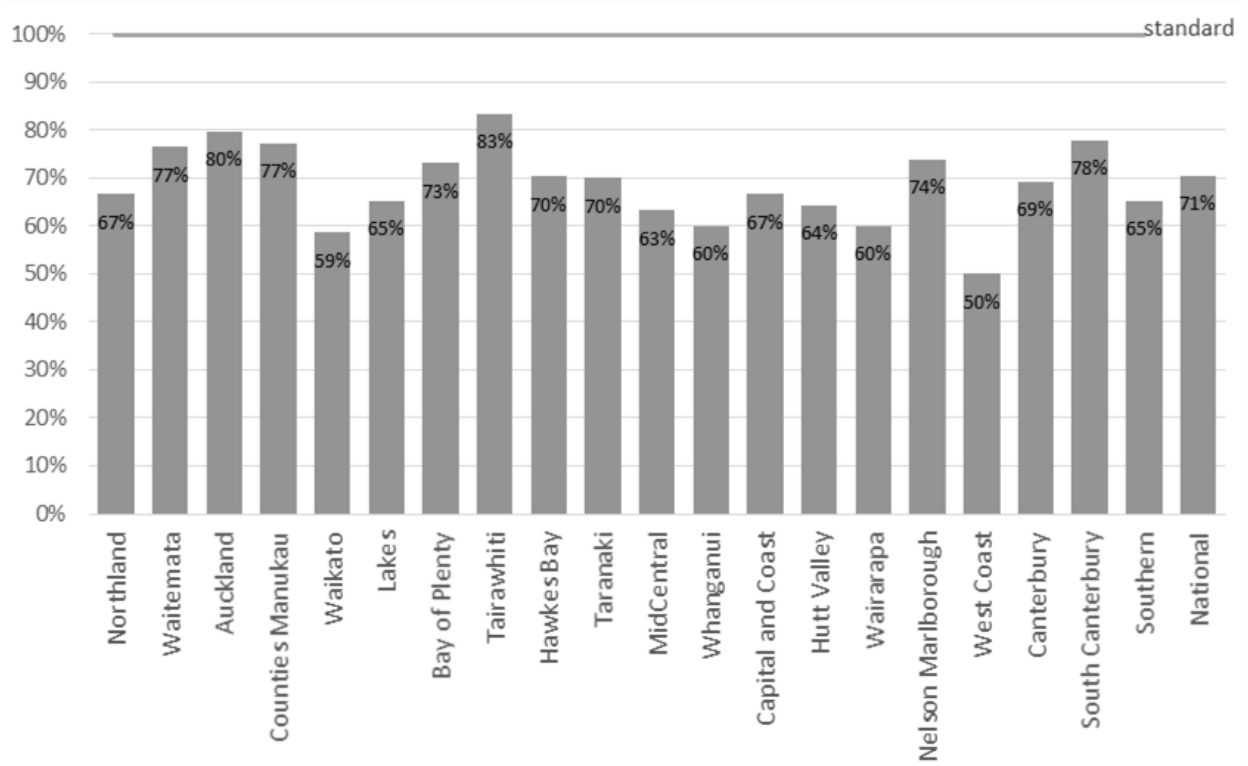


Table 8: Percentage of second samples received (or other appropriate follow-up occurred) within 10 days, January to December 2017

DHB of domicile	Within 10 days		Other follow up*		Follow up complete		No follow up		Total
	no.	%	no.	%	no.	%	no.	%	no.
Northland	34	67%	14	27%	48	94%	3	6%	51
Waitemata	101	77%	22	17%	123	93%	9	7%	132
Auckland	67	80%	14	17%	81	96%	3	4%	84
Counties Manukau	129	77%	34	20%	163	98%	4	2%	167
Waikato	44	59%	21	28%	65	87%	10	13%	75
Lakes	15	65%	5	22%	20	87%	3	13%	23
Bay of Plenty	38	73%	13	25%	51	98%	1	2%	52
Tairawhiti	5	83%	0	0%	5	83%	1	17%	6
Hawke's Bay	26	70%	11	30%	37	100%	0	0%	37
Taranaki	14	70%	6	30%	20	100%	0	0%	20
MidCentral	31	63%	17	35%	48	98%	1	2%	49
Whanganui	6	60%	4	40%	10	100%	0	0%	10
Capital & Coast	36	67%	18	33%	54	100%	0	0%	54
Hutt Valley	18	64%	8	29%	26	93%	2	7%	28
Wairarapa	3	60%	2	40%	5	100%	0	0%	5
Nelson Marlborough	17	74%	6	26%	23	100%	0	0%	23
West Coast	4	50%	4	50%	8	100%	0	0%	8
Canterbury	74	69%	31	29%	105	98%	2	2%	107
South Canterbury	7	78%	2	22%	9	100%	0	0%	9
Southern	32	65%	15	31%	47	96%	2	4%	49
Unknown	3	33%	6	67%	9	100%	0	0%	9
National	**704**	**71%**	**253**	**25%**	**957**	**96%**	**41**	**4%**	**998**

Indicator 6: Laboratory turnaround time positive results

Description: This indicator monitors the timeliness of reporting of newborns with screen positive results by the laboratory.

Rationale: Early detection of screened disorders is dependent on timely referral of newborns with positive screening results for diagnostic testing.

Standard: 100% of screen positive results are notified to the newborn's referring practitioner within the disorder specific number of calendar days.

Interpretation: Overall 82% of screen positives were notified in 2017 within the standard timeframes; an 23% increase on 2016 (59%). There was wide variation in the timeliness of notification of screen positive results across the screened disorders, with disorder specific timeframes being met for all of the 8 disorder groups.

Comment: This indicator is being reviewed to improve accuracy and clinical utility. In 2016 all 'clinical critical' results were reported within the timeframes. A 'clinical critical' screening result is one which 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 1-2 days can affect the outcome.

The 'non clinical critical' cases warrant different indicator timeframes. Also, 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 immune-reactive trypsin in the cystic fibrosis screen, it is sent for mutation analysis. The request for a second sample to confirm the thyroid result will be made after the cystic fibrosis mutation result is available.

Figure 7: Percentage of screen positives notified within the disorder specific timeframe, January to December 2017

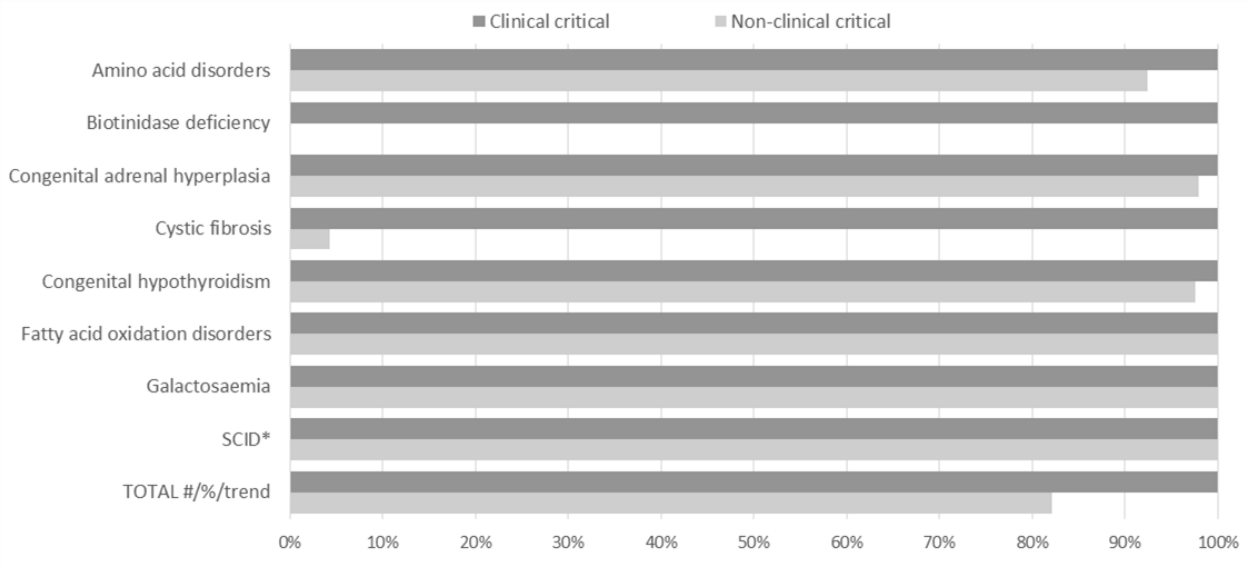


Table 9: Notification of screen positives, January to December 2017

Disorder	Timeframe (calendar days)		Timeframe met				Timeframe not met				Total no.	
	Clinical critical	Non-clinical	Clinical critical		Non-clinical critical		Clinical critical		Non-clinical critical		Clinical critical	Non-clinical
			no.	%	no.	%	no.	%	no.	%		
Amino acid disorders	2	7	2	100%	73	92%	0	0%	6	8%	2	79
Biotinidase deficiency	–	7	0	100%	0	0%	0	0%	0	0%	0	0
Congenital adrenal hyperplasia	2	7	1	100%	95	98%	0	0%	2	2%	1	97
Cystic fibrosis	–	7	0	100%	2	4%	0	0%	44	96%	0	46
Congenital hypothyroidism	4	7	9	100%	41	98%	0	0%	1	2%	9	42
Fatty acid oxidation disorders	2	7	8	100%	28	100%	0	0%	0	0%	8	28
Galactosaemia	2	7	0	100%	4	100%	0	0%	0	0%	0	4
SCID*	–	7	0	100%	1	100%	0	0%	0	0%	0	1
Total # / % / trend	**20/20**	**(100%, no change)**	**20**	**100%**	**244**	**82%**	**0**	**0%**	**53**	**18%**	**20**	**297**

Note: SCID (Severe Combined Immuno-deficiency) testing was introduced in December 2017.

* The validity of these timeframes are being reviewed to more accurately reflect clinical utility, for example not all screen positive cases were 'clinical critical'.

Indicator 7: Age of receipt into clinical care

Description: Monitoring the commencement of treatment for newborns diagnosed with a screened condition.

Rationale: The NMSP aims for early confirmed diagnosis and timely treatment to ensure that newborns with metabolic conditions have their development potential impacted as little as possible.

Standard: 100% of newborns who have a screen positive result and confirmed diagnosis have treatment commenced within the disorder specific time frame (age of newborn in days).

Interpretation: There was wide variation in timeliness of commencement of treatment for newborns diagnosed with a screened disorder. The disorder specific timeframe was met for all of the eight disorders with cases.

Comment: Delays in treatment are caused by a combination of: later diagnosis of mild disease, difficulties obtaining diagnostic tests, or difficulty making a definitive diagnosis. Delayed diagnosis is far more likely when the disease is mild, for example where the initial test is marginally abnormal and confirmed with a second dried blood spot. Diagnosis may also be delayed due to diagnostic test processes, for example some laboratories do not do sweat tests for possible cystic fibrosis until the newborn is a month old.

Table 10: Confirmed diagnosis commencement of treatment, January to December 2017

Disorder	Timeframe (calendar days)		Timeframe met				Timeframe not met				Total no.	
	Clinical critical	Non-clinical	Clinical critical		Non-clinical critical		Clinical critical		Non-clinical critical		Clinical critical	Non-clinical
			no.	%	no.	%	no.	%	no.	%		
Amino acid disorders	10	28	1	100%	0		0	0%	0		1	0
Biotinidase deficiency	–	28	0		0		0	0%	0		0	0
Congenital adrenal hyperplasia	10	28	1	100%	0		0	0%	0		1	0
Cystic fibrosis	–	28	12	100%	0		0	0%	0		12	0
Congenital hypothyroidism	10	28	9	100%	11	100%	0	0%	0		9	11
Fatty acid oxidation disorders	10	28	6	100%	0		0	0%	0		6	0
Galactosaemia	10	28	0		0		0	0%	0		0	0
SCID*	–	14	1	100%	0		0	0%	0		1	0
Total			**30**	**100%**	**11**		**0**	**0%**	**0**		**30**	**11**

* The validity of these timeframes are being reviewed to more accurately reflect clinical utility. There were no known clinical consequences of delayed treatment.

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, with associated costs and anxiety for families. Reporting of PPV helps to monitor potential harm of the programme due to identification of false positives through screening.

Standard: None.

Interpretation:

Comment:

Table 11: Positive predictive value of the screening test, 2013–2017

	Babies screened	Positive tests	Cases		Missed cases		Sensitivity %	Specificity %	PPV %
			True positive A	False positive B	False negative C	True negative D			
AABD	294,293	748	19	729	2	293,544	90.5	99.8	2.5
Galactosemia	294,293	15	1	14	0	294,278	100	100	6.7
Biotinidase def	294,293	8	1	7	0	294,285	100	100	12.5
CH	294,293	276	154	122	1	294,016	99.4	100	55.8
CF	294,293	258	60	198	0	294,035	100	99.9	23.3
CAH	294,293	253	7	246	0	294,040	100	99.9	2.8
FAOD	294,293	342	38	304	0	293,951	100	99.9	11.1
SCID	3,843	0	0	0	0	3,843		100	
Total		**1,900**	**280**	**1,620**	**3**	**290,490**	**98.9**	**99.4**	**14.7**

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)
Tyrosinaemia (fumaryl acetoacetase deficiency, tyrosine aminotransferase deficiency)

Fatty acid oxidation disorders
CACT (carnitine acylcarnitine translocase deficiency
Carnitine transporter defect
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 (CH)
Congenital adrenal hyperplasia (CAH)
Cystic fibrosis (CF)
Biotinidase deficiency
Galactosaemia
Severe Combined Immuno-deficiency (SCID)