MINISTRY OF HEALTH
MANATŪ HAUORA

New Zealand Government

National Screening Unit

Newborn Metabolic Screening Programme

Annual Report

January to December 2018

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Disclaimer

This publication reports on information Auckland District Health Board has provided to the Ministry of Health. 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 the Ministry of Health.

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Executive summary

1. The Newborn Metabolic Screening Programme (NMSP) screened 57,880 of the 58,163 babies born in 2018. This represents a national coverage rate of 99.5 percent, which is comparable with coverage rates since the programme began in 1969. Coverage rates at a district health board (DHB) level range from 97.3 percent to 100 percent.

2. In 2018, coverage varied by ethnic group: 98.1 percent of Māori newborns, 98.6 percent of Pacific newborns and 100 percent of newborns of all other ethnicities were screened. Since 2017, DHBs 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.

3. In 2018, 67 newborns were diagnosed with a screened disorder. This is comparable with previous years.

4. Blood spot cards are expected to arrive at the laboratory within four days of sampling. In 2018, 85 percent arrived in the indicator timeframe. The national standard is 95 percent. 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 focus on process quality improvement. The result has been a 19 percent increase in the four-day transit rate, from 66 percent in 2014 to 85 percent in 2018.

5. A phone and text service between LabPlus and lead maternity carers, aimed at improving the turnaround time of requests for second samples, was introduced in 2015. The rate of return within the expected 10-day timeframe has risen from 38 percent in 2014 to 77 percent in 2018.

6. In 2016, the National Screening Unit, together with the programme's lead paediatricians and laboratory scientists, started a review of the monitoring indicators. This review was completed in February 2018 and this report contains the updated indicators.

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.

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**. 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.

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 to 60 newborns a year with a metabolic 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'). Samples must be collected when the newborn is between 48 and 72 hours of age for optimal testing. Cards are sent urgently to LabPlus at Auckland District Health Board (DHB), which analyses the samples and reports the results to appropriate clinicians. Blood spot samples are screened for the 23 conditions listed in Appendix 1.

Since 2005, the NSU at the Ministry of Health has overseen 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.

Data summary

Screening data is sourced from LabPlus at Auckland DHB for all blood spot cards received in the 2018 calendar year. Birth data in the 2018 calendar year is sourced from the National Maternity Collection at the Ministry of Health. Ethnicity data is prioritised following Statistics New Zealand's prioritised ethnicity model, which is the standard approach across the health sector. When a newborn's DHB of domicile is unknown, it is set to 'Unknown'.

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 is at 99.5 percent which is above target. Coverage by DHB varied from 97.3 percent upward. Coverage by ethnicity varied from 98.1 percent for Māori newborns, to 98.6 percent for Pacific newborns and 100 percent for Other newborns.

Comment: All DHBs achieved at least 97 percent coverage. Seven DHBs have not made the 99 percent target.

It is estimated that the NMSP did not screen between 250 and 350 newborns in 2018. It is not yet possible to distinguish between the few newborns who are unscreened because parents/guardians withhold consent and those not screened because they are missed altogether. Some DHBs have begun to actively identify and follow up on their unscreened newborns, with the support of LabPlus.

Coverage rates for Māori are lower than for the general population at 14 DHBs, as measured using a rate ratio. These rates are expected to improve with increased matching of birth and screening data.

Note: Due to a mismatch between denominator data (babies born in the calendar year) and numerator data (screening performed in the calendar year) the percentages calculation may vary by ~0.2 percent at a national level.

Table 1: Coverage over time

Year	Births	Babies screened	Coverage (%)
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
2018	58,163	57,880	99.5

Table 2: Coverage by ethnicity, January to December 2018

Ethnicity	Births	Babies screened	Coverage (%)
Māori	14,175	13,910	98.1
Pacific	5,886	5,803	98.6
Other	38,102	38,167	100*
Total	**58,163**	**57,880**	**99.5**

* Percentages greater than 100 percent (due to a mismatch between numerator and denominator data) are capped at 100 percent.

Table 3: Coverage by DHB of domicile and ethnicity, January to December 2018

DHB of domicile	Māori (%)	Non-Māori (%)	Total (%)	Ratio†
Northland	97.4	98.7	97.9	0.99
Waitematā	97.5	100*	100.0	0.97
Auckland	99.2	100.0	99.9	0.99
Counties Manukau	97.7	99.1	98.9	0.99
Waikato	97.5	100*	99.5	0.97
Lakes	97.3	97.7	97.5	1.00
Bay of Plenty	100.0	100*	100*	0.99
Tairāwhiti	96.7	98.2	97.3	0.99
Hawke's Bay	98.0	98.8	98.5	0.99
Taranaki	99.1	100*	99.8	0.99
MidCentral	96.4	100*	100*	0.94
Whanganui	100*	100*	100*	1.00
Capital & Coast	97.3	99.3	99.0	0.98
Hutt Valley	98.5	99.0	98.9	1.00
Wairarapa	95.3	100*	98.8	0.95
Nelson Marlborough	100.0	100.0	100.0	1.00
West Coast	100.0	98.9	99.1	1.01
Canterbury	99.7	99.8	99.8	1.00
South Canterbury	100.0	100*	100*	0.99
Southern	98.5	100*	99.8	0.98
National	**98.1**	**100.0**	**99.5**	**0.98**

* Percentages greater than 100 percent (due to a mismatch between numerator and denominator data) are capped at 100 percent.

† 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.

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 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.

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

Interpretation: Timeliness of sample taking varied between DHBs from 66 percent (Lakes) to 90 percent (South Canterbury). The national average was 79 percent, which is the same as 2017. Currently no DHBs are meeting the standard.

Comment: Canterbury and South Canterbury DHBs have the highest proportion of samples taken between 48 and 72 hours after birth (89 percent and 90 percent respectively). Over a third of samples from Waikato and Lakes DHBs were taken outside the standard period (33 percent and 34 percent respectively).

Figure 1: Percentage of samples taken between 48 and 72 hours, January to December 2018

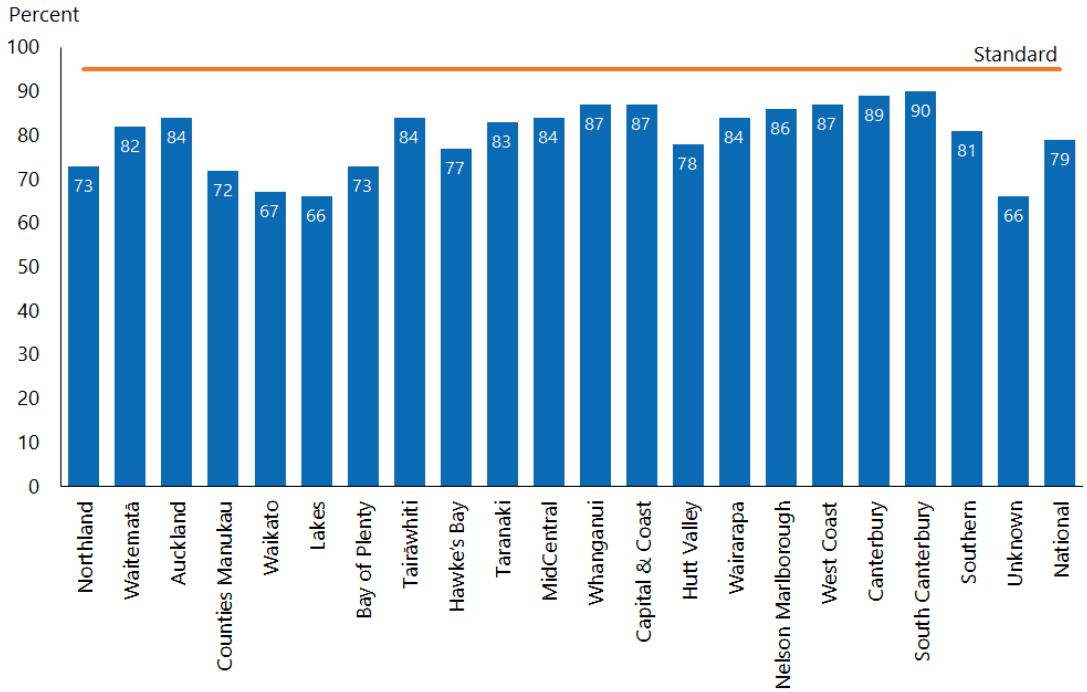


Table 4: Timing of sample taking, January to December 2018

DHB of domicile	Less than 48 hours		48 to 72 hours		More than 72 hours		Unknown		Total
	No.	%	No.	%	No.	%	No.	%	No.
Northland	15	1	1,565	73	505	24	60	3	2,145
Waitematā	60	1	6,064	82	1,179	16	120	2	7,423
Auckland	69	1	4,606	84	609	11	179	3	5,463
Counties Manukau	78	1	5,831	72	2,006	25	225	3	8,140
Waikato	54	1	3,554	67	1,564	29	165	3	5,337
Lakes	5	0	969	66	466	32	38	3	1,478
Bay of Plenty	15	0	2,198	73	726	24	84	3	3,023
Tairāwhiti	5	1	574	84	92	13	11	2	682
Hawke's Bay	17	1	1,613	77	391	19	64	3	2,085
Taranaki	11	1	1,296	83	228	15	28	2	1,563
MidCentral	14	1	1,789	84	274	13	63	3	2,140
Whanganui	4	0	700	87	90	11	13	2	807
Capital & Coast	33	1	2,788	87	287	9	83	3	3,191
Hutt Valley	17	1	1,498	78	361	19	43	2	1,919
Wairarapa	10	2	404	84	51	11	17	4	482
Nelson Marlborough	9	1	1,273	86	168	11	26	2	1,476
West Coast	0	0	279	87	35	11	8	2	322
Canterbury	71	1	5,579	89	447	7	159	3	6,256
South Canterbury	4	1	548	90	51	8	6	1	609
Southern	25	1	2,649	81	530	16	58	2	3,262
Unknown	2	3	51	66	14	18	10	13	77
National	**518**	**1**	**45,828**	**79**	**10,074**	**17**	**1,460**	**3**	**57,880**

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 98.4 percent to 99.4 percent across DHBs. The national average was 98.8 percent.

Comment: Overall sample quality improved slightly in 2018, with 1.2 percent (698) of all samples being unsatisfactory compared with 1.3 percent (743) in 2017.

Sample collection quality, such as insufficient blood on the card, remains the main reason why samples were unsatisfactory. Each unsatisfactory sample is followed up with a request for a second sample (Indicator 5) to reduce the risk to the babies affected.

Table 5: Percentage of samples of a satisfactory quality, January to December 2018

DHB of domicile	Satisfactory		Unsatisfactory		Total
	No.	%	No.	%	No.
Northland	2,117	98.7	28	1.3	2,145
Waitematā	7,355	99.1	68	0.9	7,423
Auckland	5,405	98.9	58	1.1	5,463
Counties Manukau	8,022	98.6	118	1.4	8,140
Waikato	5,262	98.6	75	1.4	5,337
Lakes	1,457	98.6	21	1.4	1,478
Bay of Plenty	2,990	98.9	33	1.1	3,023
Tairāwhiti	677	99.3	5	0.7	682
Hawke's Bay	2,052	98.4	33	1.6	2,085
Taranaki	1,550	99.2	13	0.8	1,563
MidCentral	2,105	98.4	35	1.6	2,140
Whanganui	802	99.4	5	0.6	807
Capital & Coast	3,165	99.2	26	0.8	3,191
Hutt Valley	1,896	98.8	23	1.2	1,919
Wairarapa	479	99.4	3	0.6	482
Nelson Marlborough	1,461	99.0	15	1.0	1,476
West Coast	318	98.8	4	1.2	322
Canterbury	6,171	98.6	85	1.4	6,256
South Canterbury	605	99.3	4	0.7	609
Southern	3,222	98.8	40	1.2	3,262
Unknown	71	92.2	6	7.8	77
National	**57,182**	**98.8**	**698**	**1.2**	**57,880**

Table 6: Reason for unsatisfactory samples, January to December 2018

Reason*	Number	Percentage
Collection	501	71.8
Timing	161	23.1
Transport	33	4.7
Other	3	0.4
Total	**698**	**100.0**

* Summary of main reasons:

- **Collection:** insufficient blood or the sample was contaminated.
- **Timing:** sample was collected too early (before 48 hours of age).
- **Transport:** sample took more than one month to arrive, blood was wet when folded, damaged in transit or put wet into a plastic bag.
- **Other:** any other reason for the sample being unsatisfactory.

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 DHBs, ranging from 72 percent to 92 percent of samples received within four days. National timeliness has improved from 78 percent in 2017 to 85 percent in 2018.

Comment: As in 2016 and 2017, this indicator remained the focus of considerable quality improvement work in 2018. The NSU continue to provide DHBs with quarterly 'transit time' reports as feedback on transit time turnaround. To access the transit time reports, go to: **https://minhealthnz.shinyapps.io/nsu-nmsp-transittime/**.

Figure 2: Percentage of samples the laboratory received within four days of sample taking, January to December 2018

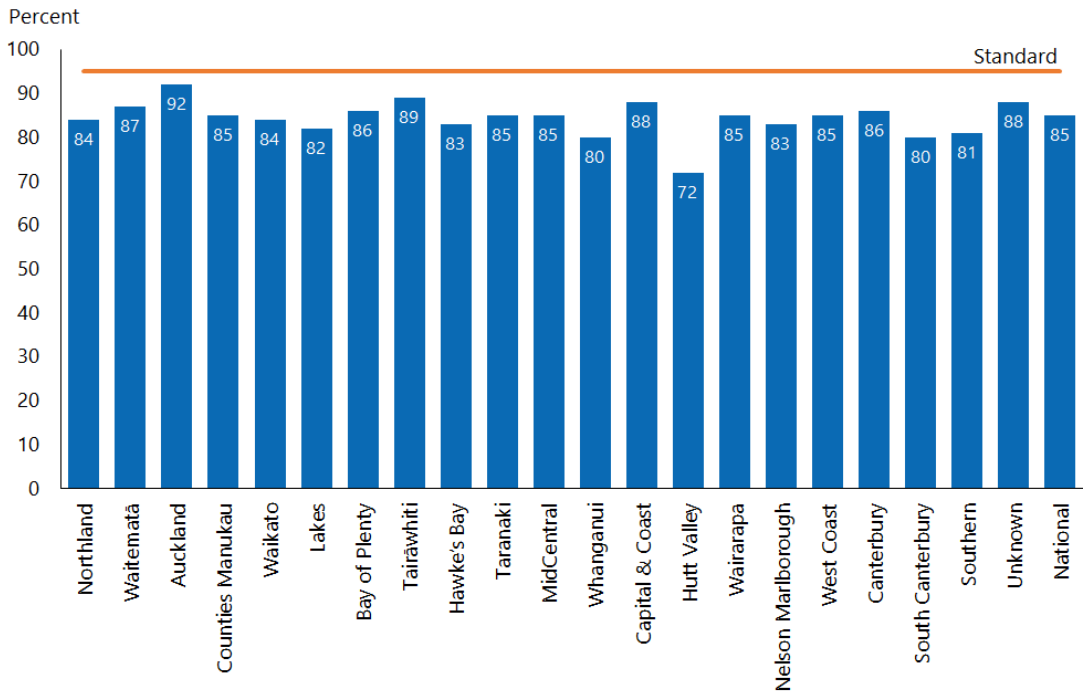


Table 7: Percentage of samples the laboratory received within four days of sample taking, January to December 2018

DHB of domicile	Within 4 days		Total
	No.	%	No.
Northland	1,793	84	2,145
Waitematā	6,473	87	7,423
Auckland	5,047	92	5,463
Counties Manukau	6,925	85	8,140
Waikato	4,491	84	5,337
Lakes	1,206	82	1,478
Bay of Plenty	2,585	86	3,023
Tairāwhiti	607	89	682
Hawke's Bay	1,740	83	2,085
Taranaki	1,321	85	1,563
MidCentral	1,825	85	2,140
Whanganui	643	80	807
Capital & Coast	2,800	88	3,191
Hutt Valley	1,384	72	1,919
Wairarapa	412	85	482
Nelson Marlborough	1,219	83	1,476
West Coast	273	85	322
Canterbury	5,406	86	6,256
South Canterbury	490	80	609
Southern	2,642	81	3,262
Unknown	68	88	77
National	**49,350**	**85**	**57,880**

Indicator 5: Receipt and follow-up of second samples

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 2018, 77 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 the newborn had been referred to a specialist, or had died.

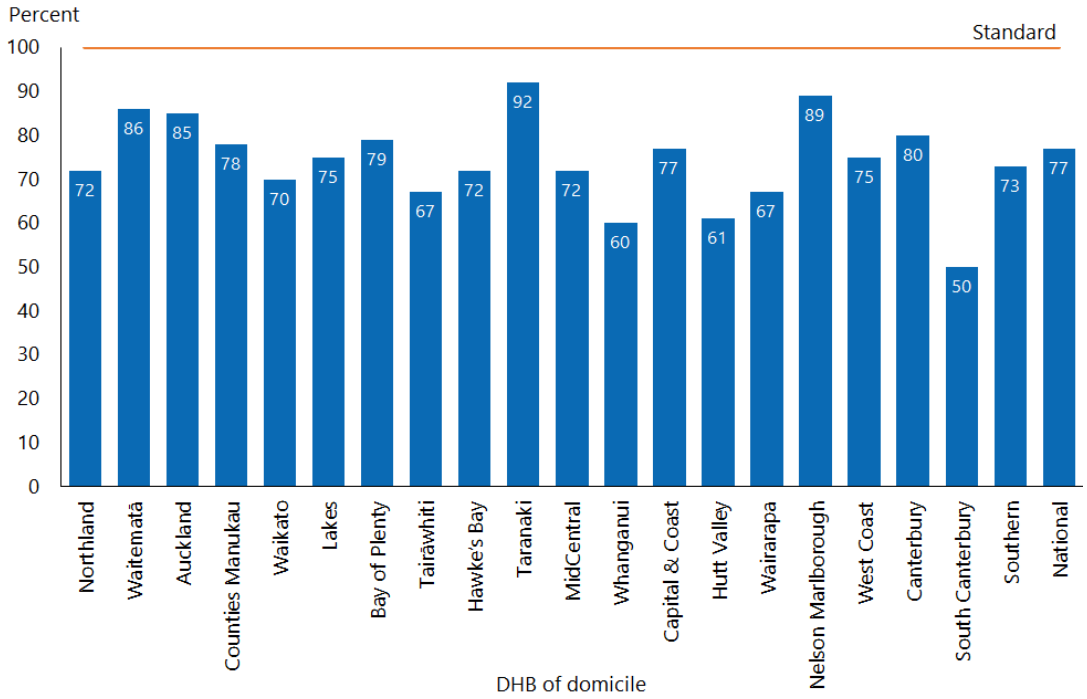
Comment: The time taken to receive a follow-up sample is influenced by: the time it takes to generate, send and receive the request; and the time it takes to collect the second sample (usually at the next scheduled visit of the lead maternity carer) and send it to the laboratory and for the laboratory to receive it.

May 2015 saw the introduction of a new protocol (which included sending text messages, making extra phone calls and providing written reports) for reminding lead maternity carers when the laboratory did not receive follow-up samples. Between 2014 and 2017, the percentage of second samples received in 10 days or fewer increased from 38 percent to 71 percent; it rose further in 2018 to 77 percent.

In the reporting period, a second sample was received, declined or had other follow-up at some stage in 97 percent of the instances when a second sample was requested.

Since 2014, when the laboratory made 1,352 second sample requests, the number of requests has declined: it requested 1,171 in 2015, 988 in 2016, 998 in 2017 and 755 in 2018. This reduction is the result of: stopping screening for two conditions with a high positive test rate (3MCC and tyrosinemia); introducing second-tier tests in screening for some amino acid breakdown disorders; and improving sample quality.

Figure 3: Percentage of second samples the laboratory received (or when other appropriate follow-up occurred) within 10 days, January to December 2018



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 lead maternity carer / specialist paediatrician by the laboratory within the following timeframes:

Reason for report	Calendar days (from receipt in lab to notification of screen positives)	
	Clinical critical	Non-clinical critical
Amino acid disorders	2	7
Biotinidase deficiency	–	7
Congenital adrenal hyperplasia (CAH)	2	7
Cystic fibrosis (CF)	–	7
Congenital hypothyroidism (CH)	4	7
Fatty acid oxidation disorders	2	7
Galactosaemia	2	7
SCID	–	7

Interpretation: Overall, 83 percent of clinical critical screen positives, and 71 percent of non-clinical critical screen positives, were notified within the expected timeframes in 2018. Both are below target of 100 percent and both are lower than 2017 figures. The timeliness of notification of screen positive results varied widely across the screened disorders, and caution should be used due to the relatively low numbers of disorders being reported.

Comment: In 2018, 24 of 29 'clinical critical' results were reported within the timeframes. 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.

The ‘non-clinical critical’ cases warrant different indicator timeframes. In 2018, 113 of 160 ‘non-clinical critical’ cases were reported within the timeframes. 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 laboratory will request a second sample to confirm the thyroid result after the cystic fibrosis mutation result is available.

Notably, the change to counting transit times from the ‘date of receipt’ in the laboratory (rather than from the date of registration – that is, the start of the test process) has impacted this indicator as it adds two more days to the laboratory testing timeframe for 25 percent of samples. For most conditions, the number of cases involved is small.

Figure 4: Percentage of screen positives the laboratory notified within the disorder-specific timeframe, January to December 2018

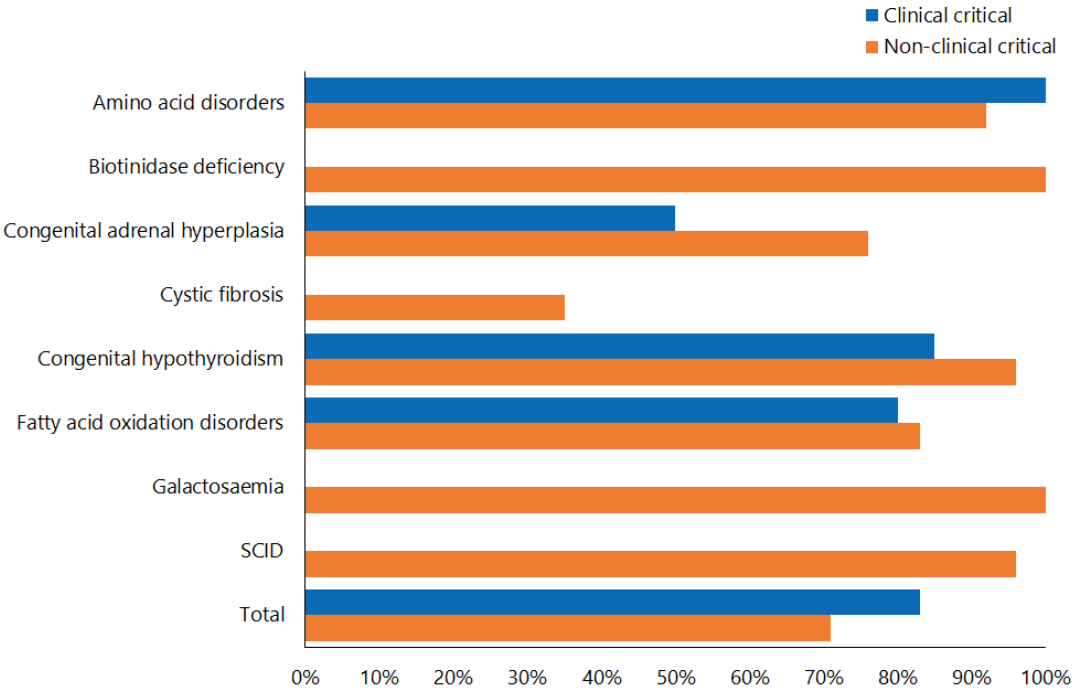


Table 8: Notification of screen positives, January to December 2018

Disorder	Timeframe		Timeframe met				Total	
	Clinical critical	Non-clinical critical	Clinical critical		Non-clinical critical		Clinical critical	Non-clinical critical
	Calendar days		No.	%	No.	%	No.	No.
Amino acid disorders	2	7	4	100	11	92	4	12
Biotinidase deficiency	–	7	0	–	6	100	0	6
Congenital adrenal hyperplasia	2	7	1	50	19	76	2	25
Cystic fibrosis	–	7	0	–	20	35	0	57
Congenital hypothyroidism	4	7	11	85	27	96	13	28
Fatty acid oxidation disorders	2	7	8	80	5	83	10	6
Galactosaemia	2	7	0	–	1	100	0	1
SCID	–	7	0	–	24	96	0	25
Total			**24**	**83**	**113**	**71**	**29**	**160**

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 metabolic disorders have their development potential impacted 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 conditions	Age of baby in days – 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 all 26 cases identified as clinical critical. Most non-clinical critical cases were received into clinical care within the specified timeframe; however, three of the 41 cases did not meet this timeframe.

Comment: Two of the three cases identified as not meeting the timeframe were very low birthweight babies, who were screened appropriately.

Table 9: Timeframe met for starting treatment after confirmed diagnosis, January to December 2018

Disorder	Timeframe*		Timeframe met				Total		Total
	Clinical critical	Non-clinical critical	Clinical critical		Non-clinical critical		Clinical critical	Non-clinical critical	
	Timeframe		No.	%	No.	%	No.	No.	No.
Amino acid disorders	10	28	0	–	4	100	0	4	4
Biotinidase deficiency	–	28	0	–	1	100	0	1	1
Congenital adrenal hyperplasia	10	28	2	100	0	–	2	0	2
Cystic fibrosis	–	28	0	–	19	100	0	19	19
Congenital hypothyroidism	10	28	14	100	12	80	14	15	29
Fatty acid oxidation disorders	10	28	10	100	0	–	10	0	10
Galactosaemia	10	28	0	–	0	–	0	0	0
SCID	–	14	0	–	2	100	0	2	2
Total			**26**	**100**	**38**	**93**	**26**	**41**	**67**

* The validity of these timeframes is being reviewed to more accurately reflect their 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.

Target: None.

Interpretation: The PPV for individual disorders is presented as five-year rolling data because the number of cases varies significantly year on year. Over all the tests, a baby with a positive screen is 17 percent likely to be affected with the screened disorder.

Comment: Five-year rolling data is slow to show the benefits of adding second-tier testing to the amino acid breakdown disorders and the improved protocols for some other disorders. The benefits should become evident in future reports. The overall PPV for 2018 is 35 percent.

Table 10: Positive predictive value of the screening test, 2014–2018

2014–2018	Babies screened	Positive tests	True positive	False positive	False negative	True negative	Sensitivity %	Specificity %	PPV %
Amino acid disorders	293,002	564	16	548	2	292,437	88.9	99.8	2.8
Biotinidase deficiency	293,002	14	2	12	0	292,988	100.0	100.0	14.3
CAH	293,002	224	6	218	0	292,778	100.0	99.9	2.7
CF	293,002	269	70	199	0	292,724	100.0	99.9	26.0
CH	293,002	257	147	110	2	292,822	98.7	100.0	57.2
Fatty acid oxidation disorders	293,002	266	40	226	0	292,738	100.0	99.9	15.0
Galactosaemia	293,002	14	1	13	0	292,988	100.0	100.0	7.1
SCID*	61,744	27	3	24	0	61,717	100.0	100.0	11.1
Total	**293,002**	**1,635**	**285**	**1,350**	**4**	**291,363**	**98.6**	**99.5**	**17.4**

* SCID screening started in December 2017.

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 (CH)
Congenital adrenal hyperplasia (CAH)
Cystic fibrosis (CF)
Biotinidase deficiency
Galactosaemia
Severe combined immunodeficiency (SCID)