National Screening Unit

Newborn Metabolic Screening Programme

Annual Report 2021

Released 2023

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This publication reports on information Te Toka Tumai Auckland has provided to the National screening Unit. 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 National Screening Unit.

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National Screening Unit

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Executive summary

The Newborn Metabolic Screening Programme (NMSP) screened 61,585 of the 62,623 babies born in 2021. This represents a national coverage rate of 98.3 percent, which is comparable with coverage rates since the programme began in 1969. Coverage rates at a district health board (DHB) level range from 97.0 percent to 99.5 percent. 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. In 2021, national coverage varied by ethnic group: 97.1 percent of Māori newborns, 96.8 percent of Pacific newborns, 99.2 percent of Asian and 98.9 percent of newborns of all other ethnicities were screened.

In 2021, 66 newborns were diagnosed with a screened disorder. This is in line with 2020 where 67 babies were diagnosed.

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

- Indicator 1: Coverage
- Indicator 2: Timing of sample taking
- Indicator 4: Sample dispatch and delivery
- Indicator 5: Receipt and follow-up of second samples
- Indicator 6: Laboratory turnaround time for positive results
- Indicator 7: Age of receipt into clinical care

Blood spot cards are expected to arrive at the laboratory within four days of sampling. In 2021 78.4 percent arrived in the indicator timeframe. The national standard is 95 percent.

Improving transit times has been the focus of process quality improvement since 2015. The result has seen significant increases, from 66 percent in 2014 to 88 percent in 2019. However, current trends indicate a reversal of these improvements, most likely due to the impact of COVID 19. Rates dropped 5 percent in 2020 and another 5 percent in 2021, resulting in a 10 percent drop since 2019.

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 lead maternity carers (LMCs). The rate of return within the expected 10-day timeframe has risen from 38 percent in 2014 to 76.8 percent in 2021 but is down from 83 percent in 2020.

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. 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.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 60 to 70 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 2021 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 were screened for the 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 Te Toka Tumai Auckland for all blood spot cards received in the 2021 calendar year.

Birth data from the 2021 calendar year is sourced from the Maternity data collection at the Te Whatu Ora. In this 2021 report, for the first time, 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 due to the fact that maternity is an incomplete data set and doing so prevents coverage rates exceeding 100 percent as sometimes was the case in previous years reports.

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

Ethnicity data is 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 at November 2022.

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 98.3 percent which is below target. Total coverage by DHB varied from 97.0 percent (Northland) to 99.5 percent (South Canterbury). Coverage by ethnicity varied: 96.8 percent for Pacific newborns, 97.1 percent for Māori newborns, 99.2 percent for Asian newborns and 98.9 percent for Other newborns.

Comment: All 20 DHBs achieved at least 97 percent coverage in total. Six DHBs made the 99 percent target for total coverage; however, no DHBs made 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 approximately 1038 newborns in 2021. It is not possible to distinguish between those unscreened who were offered screening and declined and those who were missed. Some DHBs 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 NSU if they would like to put the same processes in place.

Table 1: Coverage over time, 2011 – 2021.

Year	Births	Babies Screened	Coverage (%)
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
2019	59,733	59,315	99.3
2020	58,373	57,930	99.2
2021	**62,623**	**61,585**	**98.3**

Table 2: Coverage by ethnicity, January to December 2021

Ethnicity	Births	Babies Screened	Coverage (%)
Māori	16,078	15,618	97.1
Pacific	6,007	5,817	96.8
Asian	11,822	11,725	99.2
Other	28,716	28,401	98.9
Total	**62,623**	**61,585**	**98.3**

Table 3: Coverage by DHB of domicile and ethnicity, January to December 2021

	Coverage (%)					
DHB of domicile	**Maori**	**Pacific**	**Asian**	**Other**	**Total**	**Ratio†**
Northland	97.0	98.3	98.0	96.8	97.0	1.00
Waitematā	97.5	98.0	99.4	99.1	98.8	0.98
Auckland	99.0	98.4	99.8	99.5	99.4	1.00
Counties Manukau	95.9	95.0	98.9	99.0	97.2	0.98
Waikato	95.8	99.2	99.5	98.6	97.8	0.97
Lakes	96.0	98.1	98.4	98.6	97.2	0.98
Bay of Plenty	96.4	98.8	98.7	99.3	98.1	0.97
Tairāwhiti	98.1	100.0	97.0	98.0	98.0	1.00
Hawkes Bay	98.1	96.3	98.9	99.2	98.5	0.99
Taranaki	96.6	96.4	99.1	99.0	98.2	0.98
MidCentral	96.2	95.7	97.5	98.0	97.2	0.98
Whanganui	98.0	100.0	93.3	98.4	98.0	1.00
Capital and Coast	97.8	97.5	99.4	98.4	98.4	0.99
Hutt Valley	97.2	97.2	98.6	99.5	98.5	0.98
Wairarapa	98.8	96.2	97.1	100.0	99.3	0.99
Nelson Marlborough	97.1	100.0	99.3	98.3	98.2	0.99
West Coast	100.0	100.0	100.0	98.7	99.1	1.01
Canterbury	98.9	98.6	99.4	99.2	99.2	1.00
South Canterbury	98.1	100.0	100.0	99.8	99.5	0.98
Southern	98.9	99.1	99.2	99.0	99.0	1.00
Unknown	100.0	100.0	100.0	100.0	100.0	1.00
National	**97.1**	**96.8**	**99.2**	**98.9**	**98.3**	**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.*

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.

Note: The optimal time for collection was updated in December **2022** to between 24 and 48 hours.

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. The optimum window for sample collection in 2021 was 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 64.2 percent (Lakes DHB) to 88 percent (Canterbury District). The national average was 75.3 percent. Currently there are no DHBs meeting the target. Māori and Pacific ethnic groups have a higher proportion of samples taken after 72 hours than Asian and Other ethnic groups.

Comment: Canterbury has the highest proportion of samples taken between 48 and 72 hours after birth at 88 percent. Lakes and Waikato DHBs had over 25 percent of samples taken after 72 hours after birth (30.1 percent and 26.1 percent respectively).

From late 2019 samples collected from 24hrs of age were considered acceptable for screening completion and in mid 2020 the NSU communicated with LMCs and other health professionals to encourage samples being collected from 24 hours of age, so samples could be collected while baby was still in the birth facility, to help offset some of the delays being experienced by couriers as a result of COVID. This resulted in around 6.2 percent of samples being collected between 24 and 48 hours (most notably in Waitemata and Auckland DHBs), a significant increase from 1 percent in previous years.

Figure 1: Percentage of samples taken between 48 and 72 hours, January to December 2021

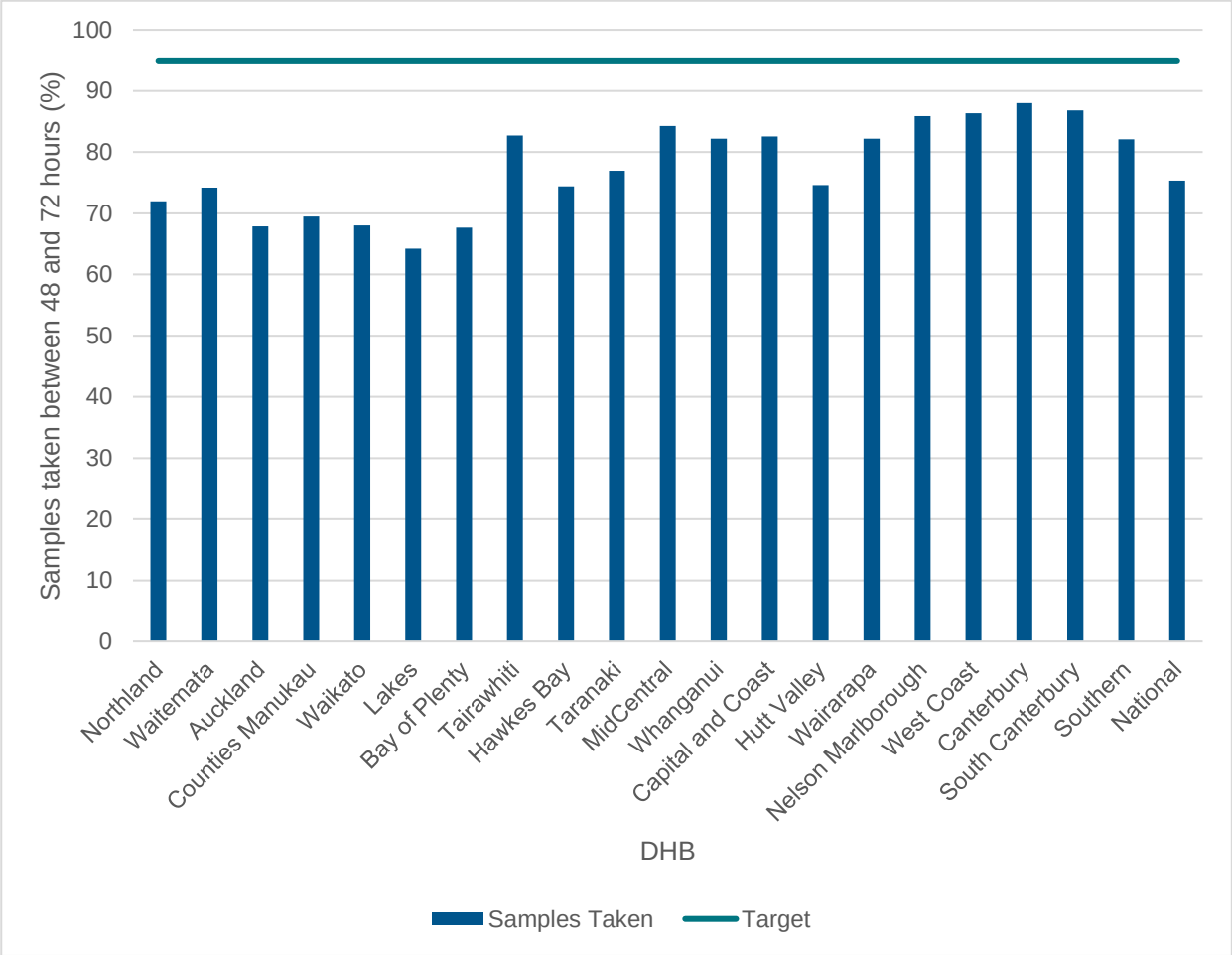


Table 4: Timing of sample taking by DHB of domicile, January to December 2021

DHB	Less than 24 Hours		Between 24 and 47 Hours		Between 48 and 72 Hours		Greater than 72 Hours		No Collection Time or Date	
	N	%	N	%	N	%	N	%	N	%
Northland	4	0.2	65	2.8	**1,692**	**72.0**	541	23.0	49	2.1
Waitemata	11	0.1	829	10.5	**5,844**	**74.2**	1,059	13.4	134	1.7
Auckland	14	0.3	1,030	18.8	**3,729**	**67.9**	575	10.5	145	2.6
Counties Manukau	16	0.2	687	7.9	**6,013**	**69.5**	1,725	19.9	216	2.5
Waikato	7	0.1	178	3.0	**3,999**	**68.0**	1,534	26.1	161	2.7
Lakes	0	0.0	41	2.6	**998**	**64.2**	467	30.1	48	3.1
Bay of Plenty	3	0.1	159	4.7	**2,276**	**67.7**	852	25.3	73	2.2
Tairawhiti	1	0.1	19	2.5	**623**	**82.7**	98	13.0	12	1.6
Hawkes Bay	6	0.3	75	3.4	**1,665**	**74.4**	444	19.8	47	2.1
Taranaki	2	0.1	68	4.3	**1,208**	**76.9**	264	16.8	28	1.8
MidCentral	6	0.3	57	2.5	**1,910**	**84.3**	244	10.8	49	2.2
Whanganui	2	0.3	30	3.8	**652**	**82.2**	96	12.1	13	1.6
Capital and Coast	8	0.2	90	2.7	**2,733**	**82.6**	403	12.2	75	2.3
Hutt Valley	5	0.2	82	4.0	**1,525**	**74.6**	401	19.6	31	1.5
Wairarapa	1	0.2	19	3.3	**476**	**82.2**	72	12.4	11	1.9
Nelson Marlborough	3	0.2	44	2.9	**1,293**	**85.9**	144	9.6	21	1.4
West Coast	0	0.0	11	3.3	**291**	**86.4**	30	8.9	5	1.5
Canterbury	4	0.1	227	3.3	**6,027**	**88.0**	457	6.7	133	1.9
South Canterbury	0	0.0	29	4.6	**547**	**86.8**	44	7.0	10	1.6
Southern	1	0.0	93	2.6	**2,882**	**82.1**	478	13.6	56	1.6
Unknown	0	0.0	2	6.7	**20**	**66.7**	1	3.3	7	23.3
National	**94**	**0.2**	**3,835**	**6.2**	**46,403**	**75.3**	**9,929**	**16.1**	**1,324**	**2.1**

Table 5: Timing of sample taken by Ethnicity January to December 2021

Ethnicity	Less than 24 Hours	Between 24 and 47 Hours	Between 48 and 72 Hours	Greater than 72 Hours	No Collection Time or Date
	%	%	%	%	%
Maori	0.2	5.1	71.3	21.2	2.4
Pacific	0.2	7.6	68.9	20.8	2.5
Asian	0.2	9.0	76.3	12.4	2.1
Other	0.1	5.5	78.5	13.9	2.0
Total	**0.2**	**6.2**	**75.4**	**16.1**	**2.2**

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 DHB) to 99.7 percent (West Coast DHB) across DHBs. The national average was 99 percent.

Comment: Overall sample quality improved slightly in 2021, with 1.0 percent (612) of all samples being unsatisfactory compared with 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. 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 6: Percentage of samples of a satisfactory quality, January to December 2021

DHB	Satisfactory		Unsatisfactory		Total
	No.	%	No.	%	No.
Northland	2,333	99.2	18	0.8	2,351
Waitemata	7,808	99.1	69	0.9	7,877
Auckland	5,440	99.0	53	1.0	5,493
Counties Manukau	8,550	98.8	107	1.2	8,657
Waikato	5,830	99.2	49	0.8	5,879
Lakes	1,546	99.5	8	0.5	1,554
Bay of Plenty	3,338	99.3	25	0.7	3,363
Tairawhiti	748	99.3	5	0.7	753
Hawkes Bay	2,210	98.8	27	1.2	2,237
Taranaki	1,547	98.5	23	1.5	1,570
MidCentral	2,239	98.8	27	1.2	2,266
Whanganui	782	98.6	11	1.4	793
Capital and Coast	3,275	99.0	34	1.0	3,309
Hutt Valley	2,019	98.8	25	1.2	2,044
Wairarapa	572	98.8	7	1.2	579
Nelson Marlborough	1,497	99.5	8	0.5	1,505
West Coast	336	99.7	1	0.3	337
Canterbury	6,771	98.9	77	1.1	6,848
South Canterbury	627	99.5	3	0.5	630
Southern	3,475	99.0	35	1.0	3,510
Unknown	30	100.0	0	0.0	30
National	**60,973**	**99.0**	**612**	**1.0**	**61,585**

Table 7: Reason for unsatisfactory samples, January to December 2021*

Reason	Number	Percentage
Collection	485	79.3
Timing	73**	11.9
Transport	51	8.3
Other	3	0.5
Total	**612**	**100**

*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
- **other:** any other reason for the sample being unsatisfactory.

** this number differs to the one in Table 4 as early samples taken from babies with low birth weight are not counted as unsatisfactory samples, and new samples requested, as second samples are routinely collected within a few days.

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 62.7 percent (South Canterbury DHB) to 91 percent (Auckland DHB) of samples received within four days. National timeliness has decreased from 83 percent in 2020 to 78.4 percent in 2021, continuing a downward trend since 2019.

Comment: Considerable quality improvement work has been undertaken for this indicator, since 2016, and this work is on-going. The national rate of samples reaching the laboratory within four calendar days of being taken increased from 66 percent in 2014 to 78.4 percent in 2021. However, it did drop from 83 percent in 2020. COVID-19 is the primary reason for this drop in dispatch and delivery timeliness as courier delivery times increased, particularly during October to December 2021. The courier delivery delays were closely monitored by LabPlus throughout 2021 and the NSU worked with some DHBs to review their processes for getting samples to the laboratory.

The NSU continues to provide DHBs with support to develop quality improvement actions, review processes and 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/**.

No courier pickups or deliveries on weekends and public holidays impacts on some samples reaching the laboratory within four calendar days of being taken.

Figure 2: Percentage of samples the laboratory received within four days of sample taking, January to December 2021

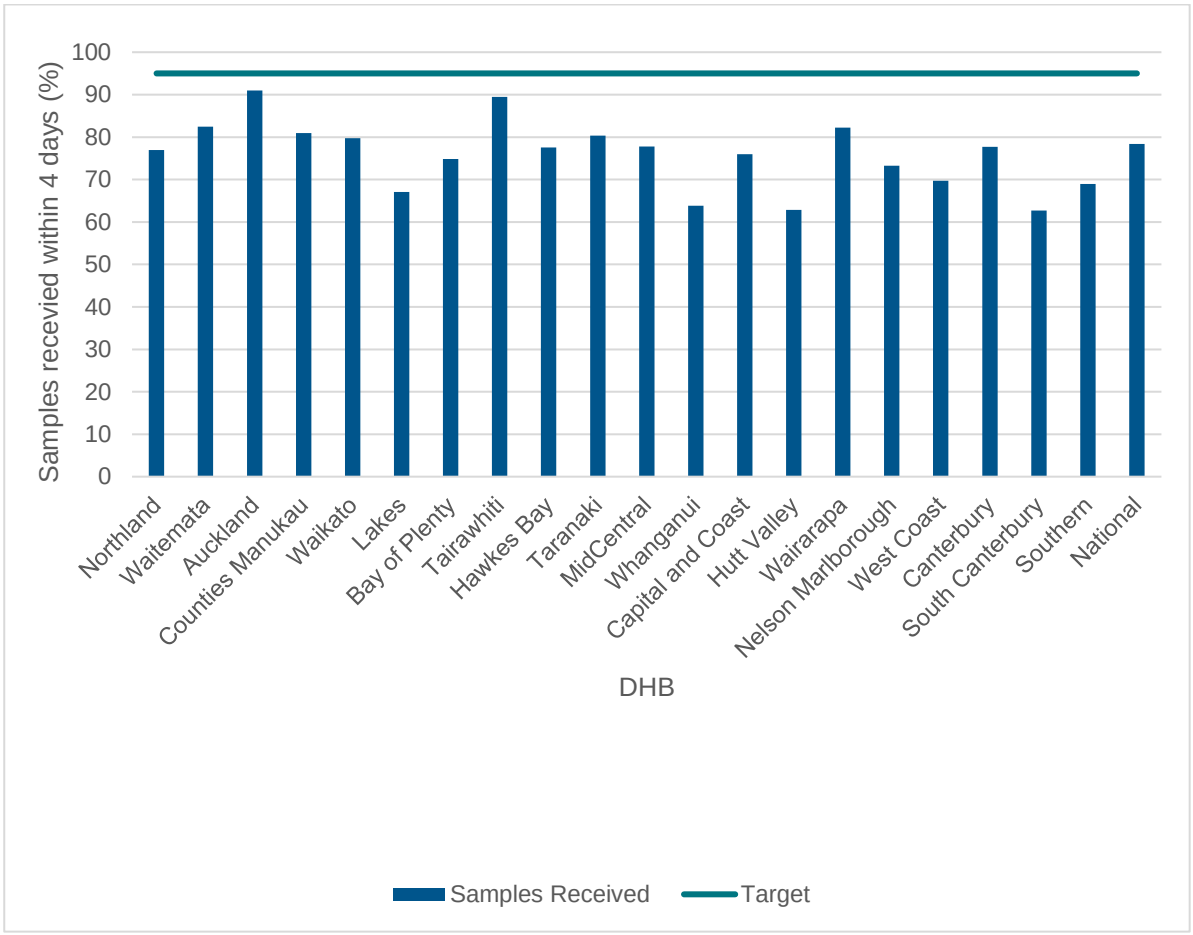


Table 8: Percentage of samples the laboratory received within four days of sample taking, January to December 2021

DHB of domicile	Within 4 days		Greater than 4 Days		Total
	No.	%	No.	%	No.
Northland	1810	77.0	511	21.7	2351
Waitemata	6497	82.5	1328	16.9	7877
Auckland	4999	91.0	444	8.1	5493
Counties Manukau	7010	81.0	1560	18.0	8657
Waikato	4688	79.7	1135	19.3	5879
Lakes	1042	67.1	494	31.8	1554
Bay of Plenty	2516	74.8	811	24.1	3363
Tairawhiti	674	89.5	79	10.5	753
Hawkes Bay	1735	77.6	482	21.5	2237
Taranaki	1262	80.4	297	18.9	1570
MidCentral	1762	77.8	486	21.4	2266
Whanganui	506	63.8	278	35.1	793
Capital and Coast	2513	75.9	770	23.3	3309
Hutt Valley	1284	62.8	747	36.5	2044
Wairarapa	476	82.2	95	16.4	579
Nelson Marlborough	1102	73.2	397	26.4	1505
West Coast	235	69.7	99	29.4	337
Canterbury	5319	77.7	1465	21.4	6848
South Canterbury	395	62.7	233	37.0	630
Southern	2420	68.9	1059	30.2	3510
Unknown	24	80.0	3	10.0	30
National	**48269**	**78.4**	**12773**	**20.7**	**6158**

*543 samples had no collection time

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 2021, 76.8 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, other appropriate follow-up had occurred, or that the newborn had died.

Comment: In the 2021 reporting period, 765 second samples were requested. A second sample was received, declined or had other follow-up in 98 percent of the instances when a second sample was requested.

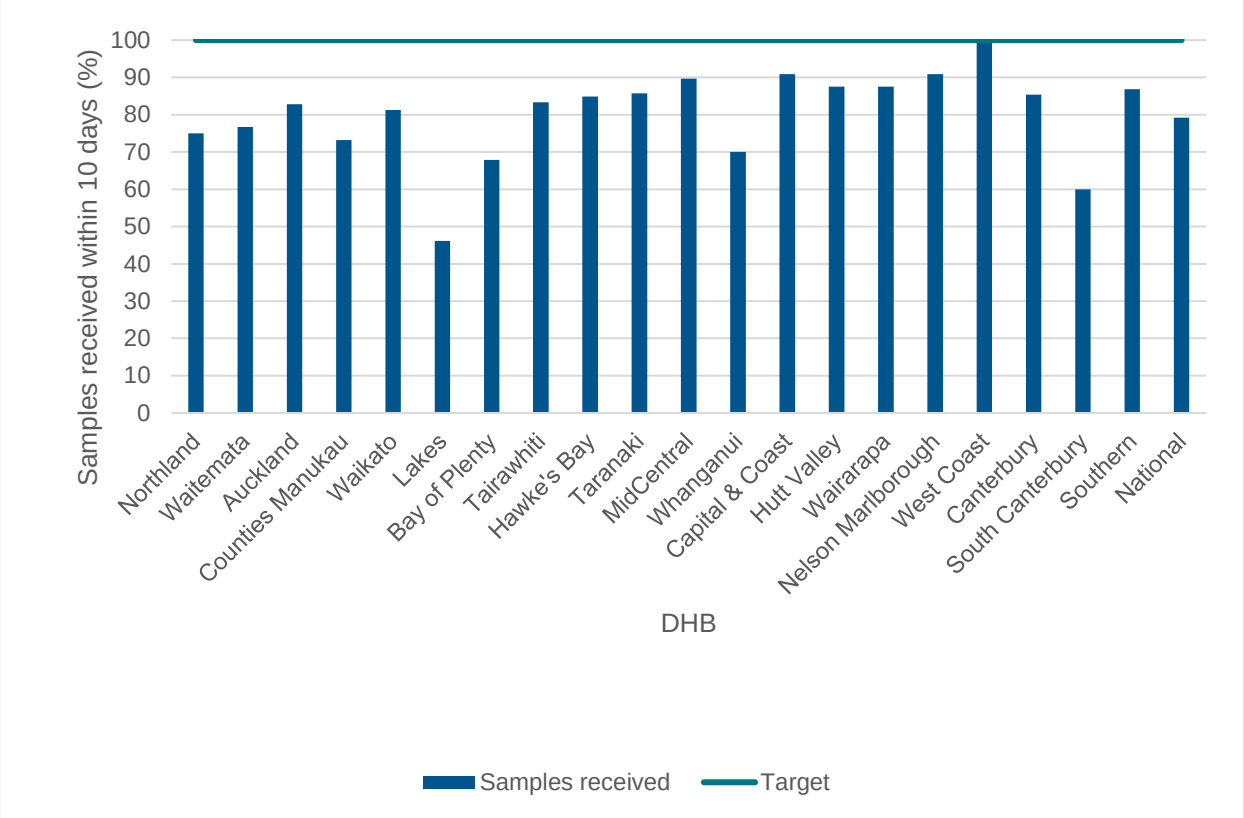
79.2 percent of second samples were received by the laboratory, had other appropriate follow-up, or were declined by parents/guardians, within 10 calendar days of the request. 20.8 percent of samples or declines were received after 10 days, this number includes 2 percent (15) that were lost to follow-up and the task closed at 28 days.

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 for the laboratory to receive it. During 2021 COVID levels 3 and 4, when the laboratory phoned LMCs to request repeat samples, they also asked the LMC to collect the next sample at their next scheduled visit instead of an extra earlier visit in order to reduce unnecessary face-to-face contact and ensure it was safe for the baby. During this time there were also delays due to pressure on courier services.

For all of the 15 cases that were lost to follow-up the laboratory has documented multiple messages between the screening lab and LMC and multiple LMC attempts to contact families to obtain follow-up samples.

Additionally, the total number of requests has declined significantly since 2014, when the laboratory made 1,352 second sample requests. This reduction is the result of stopping screening for 3MCC and carnitine uptake disorders and suspending screening for tyrosinemia; introducing second-tier tests in screening for Congenital Adrenal Hyperplasia; 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 2021



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

Table 9: Percentage of second samples the laboratory received (or when other appropriate follow-up occurred) within 10 days, January to December 2021

DHB	Within 10 Days		After 10 Days		Total
	No.	%	No.	%	No.
Northland	15	75.0	5	25.0	20
Waitemata	66	76.7	20	23.3	86
Auckland	53	82.8	11	17.2	64
Counties Manukau	90	73.2	33	26.8	123
Waikato	52	81.3	12	18.8	64
Lakes	6	46.2	7	53.8	13
Bay of Plenty	19	67.9	9	32.1	28
Tairawhiti	5	83.3	1	16.7	6
Hawke's Bay	28	84.8	5	15.2	33
Taranaki	24	85.7	4	14.3	28
MidCentral	26	89.7	3	10.3	29
Whanganui	7	70.0	3	30.0	10
Capital & Coast	40	90.9	4	9.1	44
Hutt Valley	28	87.5	4	12.5	32
Wairarapa	7	87.5	1	12.5	8
Nelson Marlborough	10	90.9	1	9.1	11
West Coast	1	100.0	0	0.0	1
Canterbury	76	85.4	13	14.6	89
South Canterbury	3	60.0	2	40.0	5
Southern	33	86.8	5	13.2	38
Unknown	17	51.5	16	48.5	33
National	**606**	**79.2**	**159**	**20.8**	**765**

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

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:

	Calendar days*	
Disorder	**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: Overall, 97.4 percent of clinical critical screen positives, and 95.2 percent of non-clinical critical screen positives, were notified within the expected timeframes in 2021. While there was an improvement from 86 percent of clinical critical screen positives, and 88 percent of non-clinical critical screen positives in 2020, both are below the target of 100 percent. The clinical critical figure has increased since 2020 by 11.4 percent and the proportion of non-clinical critical results meeting the target time-frame has increased since 2020 by 7.2 percent. 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 results being reported.

It should be noted that samples that arrive at the laboratory on a Friday afternoon are not tested until Monday morning, and this has had 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, and it was discontinued.

Comment: In 2021, 38 of 39 '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 clinical critical sample that did not meet the target turnaround time as it arrived at the laboratory on a Friday afternoon (outside of the trial period of Friday evening testing.

Non clinical-critical samples received outside of the target turnaround time were due to COVID-related stress on resources or occurred for clinical reasons. There were no adverse consequences from samples that did not meet target turnaround times.

The 'non-clinical critical' cases warrant different indicator timeframes. In 2021, 256 of 269 '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 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.

Figure 4: Percentage of screen positives the laboratory notified within the disorder-specific timeframe, January to December 2021

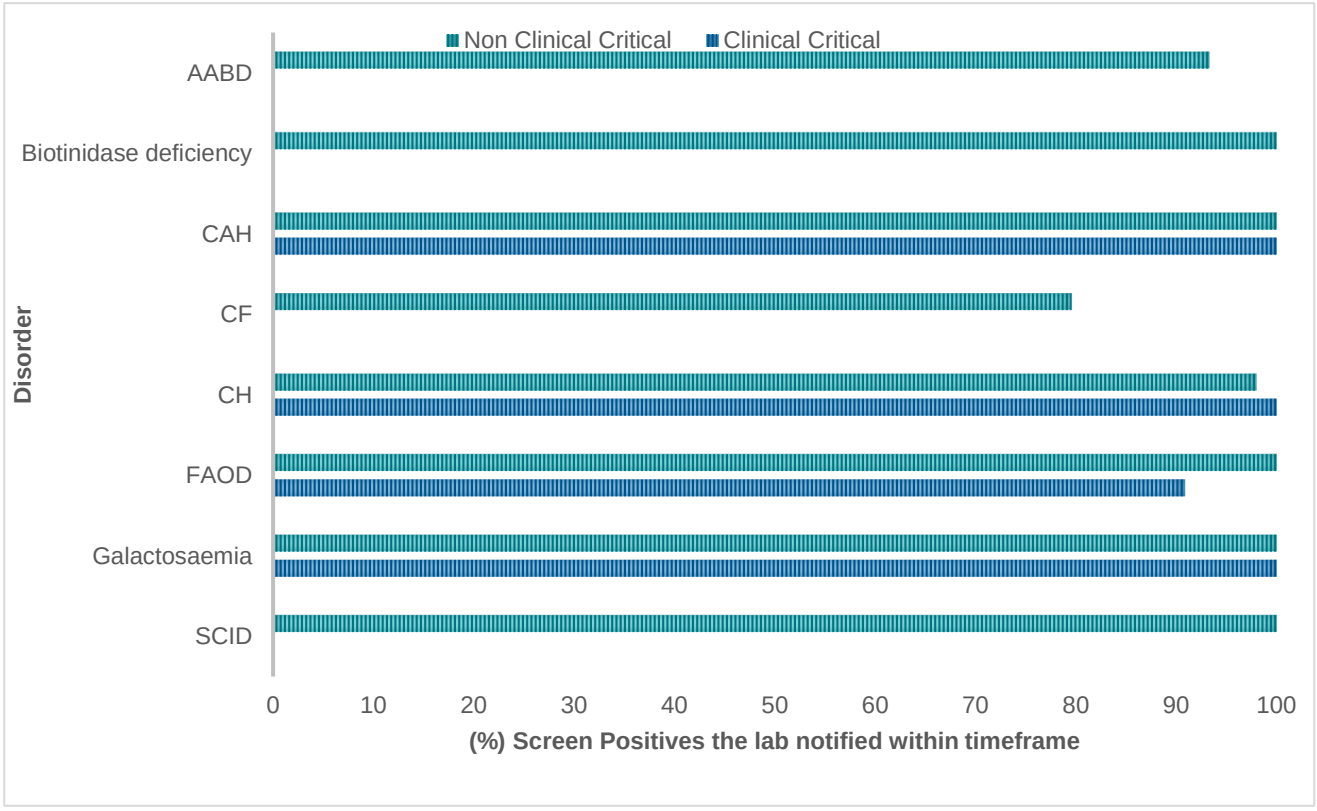


Table 10: Notification of screen positives, January to December 2021

	Timeframe		TAT met		TAT not met		Total number	
Disorder	**CC**	**NCC**	**CC**	**NCC**	**CC**	**NCC**	**CC**	**NCC**
Amino acid	2	7	0	14	0	1	0	15
Biotinidase	-	7	-	15	-	0	-	15
Congenital Adrenal Hyperplasia	2	7	2	30	0	0	2	30
Cystic Fibrosis	-	7	-	39	-	10	-	49
Congenital Hypothyroidism	4	7	22	96	0	2	22	98
Fatty acid oxidation disorders	2	7	10	3	1	0	11	3
Galactosaemia	2	7	4	14	0	0	4	14
SCID	-	7	-	45	-	0	-	45
Total	**-**	**-**	**38**	**256**	**1**	**13**	**39**	**269**
Percentage	**-**	**-**	**97.4**	**95.2**	**2.6**	**4.8**		

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

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 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:

	Age of baby in days	
Disorder	**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 24 out of 28 cases identified as clinical critical. 38 of 38 non-clinical critical cases were received into clinical care within the specified timeframe.

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

The four cases that were detected and referred into clinical care outside of timeframe primarily occurred due to courier transit delays. There were no adverse consequences from the delayed diagnosis of MCAD. The impact of delayed CH diagnosis is less clear. Optimal neuro-cognitive outcomes in CH 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 11: Timeframe met for receipt into clinical care after confirmed diagnosis, January to December 2021

	Timeframe (calendar days)		Timeframe met		Timeframe not met		Total number	
Disorder	**CC**	**NCC**	**CC**	**NCC**	**CC**	**NCC**	**CC**	**NCC**
Amino acid disorders	10	28	-	3	-	-	-	3
Biotinidase deficiency	-	28	-	1	-	-	-	1
Congenital Adrenal Hyperplasia	10	28	-	1	-	-	-	1
Cystic Fibrosis*	-	28	-	18	-	-	-	18
Congenital Hypothyroidism**	10	28	17	16	3	-	20	16
Fatty acid oxidation disorders	10	28	4	-	1	-	5	-
Galactosaemia	10	28	1	-	-	-	1	-
SCID	-	14	-	-	-	-	-	-
Total			**24**	**38**	**4**	**0**	**28**	**38**

CC: Clinical Critical; NCC: Not Clinical Critical

*Cystic Fibrosis (CF) count includes 10 cases labelled as CFSPID, ie. CF screen positive indeterminate diagnosis. These babies typically have a normal sweat test combined with 2 CFTR variants, of which at least 1 has unclear phenotypic consequences, or the combination of 1 pathogenic variant and a borderline sweat test. Children in this category are followed until 6 years of age in order 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 in LBW <1500g babies. LBW babies with CH often have delayed TSH rise and are undetectable on an initial (48 hour) sample. Of the 15 babies, none were detectable on a routine 48hour sample, 13 were detected from a scheduled 2 week sample and 2 from a scheduled 4 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 for individual disorders is presented as five-year rolling data because the number of cases varies significantly year on year. Overall, there is a 25.9% likelihood that a baby with a positive screen will be confirmed to have a target disorder.

Comment: Five-year rolling data is slow to show the benefits of adding second-tier testing for congenital adrenal hyperplasia and amino acid breakdown disorders and the improved protocols for some other disorders. Comparing the 5 year averages of 2013-2017 to 2017-2021 the sensitivity and specificity are nearly identical with sensitivity decreasing slightly from 98.9 to 98.4 percent and specificity increasing from 99.4 to 99.7 percent. Overall positive predictive value of the screening test has increased notably by over 10 percent from the 2017 annual report (14.7 percent) to 2021 annual report (25.9 percent).

Table 12: Positive predictive value of the screening test, January 2017 to December 2021

Disorder	Babies Screened	Positive tests	Cases		Missed cases		Sensitivity	Specificity	
			True Positive	False Positive	False Negative	True Negative	%	%	PPV (%)
Amino acid disorders	295369	161	13	148	0	295065	100	99.9	8.1
Biotinidase deficiency	295369	36	3	33	0	295333	100	100	8.3
Congenital Adrenal Hyperplasia	295369	126	11	115	0	295243	100	100	8.7
Cystic Fibrosis	295369	247	83	164	0	295102	100	99.9	33.6
Congenital Hypothyroidism	295369	412	167	245	2	295055	98.8	99.9	40.5
Fatty acid oxidation disorders	295369	79	32	47	0	295290	100	100	40.5
Galactosaemia	295369	37	2	35	3	295331	40	100	5.4
SCID*	240265	118	4	114	0	240146	100	100	3.4
Total	**295369**	**1216**	**315**	**901**	**5**	**294148**	**98.4**	**99.7**	**25.9**

*SCID screening started 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
Congenital adrenal hyperplasia
Cystic fibrosis
Biotinidase deficiency
Galactosaemia
Severe combined immunodeficiency (SCID)