MINISTRY OF HEALTH
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Newborn Metabolic Screening Programme

Annual Report

January to December 2013

National Screening Unit

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Contents

Glossary

CAH	Congenital Adrenal Hyperplasia
CF	Cystic Fibrosis
CH	Congenital Hypothyroidism
FAOD	Fatty Acid Oxidation Disorder
GP	General Practitioner
HIPC	Health Information Privacy Code
LMC	Lead Maternity Carer
MCAD	Medium Chain Acyl-CoA Dehydrogenase
MSUD	Maple Syrup Urine Disease
NICU	Neonatal Intensive Care Unit
NMSP	Newborn Metabolic Screening Programme
NSU	National Screening Unit
PKU	Phenylketonuria
SCBU	Special Care Baby Unit

Executive summary

Almost all babies born in New Zealand have been screened since the NMSP began in 1969, and as a result, over 45 babies are identified with and treated for a metabolic disorder each year. When a baby is diagnosed with a metabolic disorder in early infancy, treatment can commence immediately, preventing life-threatening illness and limiting the impact on the baby's development potential.

Key points for January to December 2013

- 59,192 babies were tested and 59,707 born, giving a screening coverage of 99%.
- 71% of samples were collected within the 48-72 hours timeframe. No DHB and no ethnic group met the standard of 95%. Babies of lower NZDep and European ethnicity are more likely to have the sample collected in the appropriate timeframe.
- Overall 99% of samples were suitable for testing. Eight DHBs met the standard of 99% suitable. The main reasons samples were unsuitable were 'taken too early' or 'not sufficient/contaminated sample card'. Follow-up of unsuitable samples was 91%.
- 72% of samples were received by the laboratory in four days or less. No DHB met the standard of 95%.
- The laboratory testing standard of 100% was not met for any disorder however was 99% except for fatty acid oxidation/amino acid breakdown disorders (98%).
- Of 48 clinical critical results 40 were notified within the timeframe, none were more than three days late; five babies were under paediatric care in neonatal units.
- Although only 35% of second samples were received in ten days or less (the standard is 100%), 96% of follow-up was completed appropriately.
- Of babies with diagnosed disorders, treatment was commenced by the specified age for between 33-100%, however most of the remainder were just outside the range and there were no clinical ill-effects from the delays.
- 99% of 584 requests for card returns were made in 28 days or less (the standard is 100%).
- 60 cards were used for additional testing for 'family health reasons', mostly for CMV testing.
- Follow-up of positive tests was between 99-100%. All clinical critical results had appropriate follow-up.
- 51 cases of screened disorders were detected by the programme and in 38 of these there was no clinical suspicion of the disorder before the screening test result.

Introduction

The purpose of this annual report is to provide information on the Newborn Metabolic Screening Programme (NMSP) and the performance of the NMSP against the agreed set of national indicators.

Regular analysis of data against programme indicators is a key monitoring and evaluation tool of the NMSP. The development of quarterly, biannual and annual reports has been a priority for the NMSP during 2013. Reports are published on the NSU website.

This is the third annual report of the NMSP following the development of national indicators and completion of the NMSP Monitoring Framework in November 2010.

Appendix 1 outlines the NMSP standards and indicators.

Further information on the NMSP Monitoring Framework can be found at:
http://www.nsu.govt.nz/files/NSU_Screening_Programme_2_0.pdf

Background to the Newborn Metabolic Screening Programme

Newborn babies in New Zealand have been screened for congenital metabolic disorders in a national screening programme since 1969. New Zealand was one of the first countries in the world (with Ireland), to have a national screening programme. The National Testing Centre (the laboratory arm of the programme) was established by the late Professor Arthur Veale working with the late Professor Bob Guthrie in the Human Genetics Research Unit at the School of Medicine in Dunedin. The laboratory moved to Auckland in 1973 when Professor Veale became the foundation professor of Community Health and Human Genetics in what was then the new medical school.

Since 2005, the NMSP has been overseen nationally by the National Screening Unit (NSU) of the Ministry of Health. Significant milestones for the programme include the introduction of expanded newborn screening (adding fatty acid oxidation and more amino acid breakdown disorders) in 2006. In 2009 educational and training resources (DVDs and videos) about newborn screening and best practice for lead maternity carers (LMCs) were produced and distributed.

Almost all babies born in New Zealand have been screened since the NMSP began and approximately 45 babies are identified with and treated for a metabolic disorder each year. When a baby is diagnosed with a metabolic disorder in early infancy, treatment can commence immediately, preventing life-threatening illness and limiting the impact on the baby's development potential.

Newborn metabolic screening involves collecting blood samples from babies' heels (the 'heel prick test') onto a blood spot card (a 'Guthrie card'). Blood samples must be collected between 48 and 72 hours of baby's age for maximum utility. The blood samples are screened for over 20 metabolic disorders.

The NMSP is monitored and evaluated by the NSU to ensure it continuously meets high standards. A multi-disciplinary advisory group provides expert leadership and advice for the programme. The NMSP Technical Group reviews monitoring reports and makes recommendations.

Timing of sample taking (Indicator 2) is now reported in hours unless the date and time of birth and sample collection are not provided. The improvement in the quality of data in 2012 to monitor this indicator is a significant achievement for the NMSP.

The aim of the Newborn Metabolic Screening Programme

The aim of the NMSP is to reduce morbidity and mortality through high-quality screening that facilitates early detection and treatment of specific metabolic disorders in pre-symptomatic babies.

The objectives of the programme are to:

- enable early detection of pre-symptomatic newborns
- ensure appropriate early referral to treatment of newborns
- ensure babies born with congenital metabolic disorders have their development potential impacted as little as possible from the disorder
- facilitate early diagnosis, appropriate treatment and continuous monitoring of specific metabolic disorders
- maintain high uptake of screening, community participation and trust
- facilitate continuous quality improvement through the development of quality assurance, reporting, education and the strategic planning framework
- inform the community of all aspects of newborn screening including the storage and use of blood spot cards.

Data included in this report

Data is obtained from the LabPLUS Delphic laboratory information system (Delphic). The extracted data is then placed in a temporary table on the Delphic Data Warehouse and imported into a MS Access database for analysis.

Data on DHB, ethnicity and NZ Deprivation Index (NZDep) is obtained from the Ministry of Health National Collections and merged with the LabPLUS data based on each individual's national health index (NHI).

Samples selected for inclusion in this report are based on the date they are received at the laboratory. For this reporting period, only valid samples received from 1 January to 31 December 2013 are included. For coverage and timing, samples are only included if they are a first sample received from a baby. Follow-up samples are excluded, because if a baby is screened in one reporting period, and has follow-up in the next period, they would be counted twice or age at sampling would likely fall outside the standard.

Ethnicity is prioritised based on the NHI ethnicity information. All reporting by NZDep is based on the extraction against the NHI associated with residential addresses. Decile 1 has the least deprivation and decile 10 the most deprivation.

While many LMCs are not directly responsible to a particular DHB, data is reported by DHB region, as this is the most usual way of comparing health information across New Zealand.

National monitoring indicators

Table 1 summarises all the NMSP indicators used in 2013 for regular monitoring with their reporting frequency and detail included in Appendix 2. This report, as an annual report, provides information on all nine indicators in addition to detected disorders. These indicators have been developed following consultation with key NMSP stakeholders. Indicators will be further refined as data is collected over time, and will be subject to regular review by the NMSP Advisory Group.

Table 1 NMSP indicators and monitoring frequency

Indicators	Biannually	Annually	Detail
1. Newborn Metabolic Screening Coverage		X	• DHB • Ethnicity • Deprivation status
2. Timing of sample taking	X	X	• DHB • Ethnicity • Deprivation status
Laboratory reporting			
3. Quality of Blood Samples	X	X	• DHB
4. Sample dispatch and delivery	X	X	• DHB
5. Laboratory testing timeframes	X	X	
6. Timeliness of reporting - notification of screen positives	X	X	
7. Collection and receipt of second samples		X	• DHB
Incidence		X	
8. Diagnosis and commencement of treatment by disorder: • Biotinidase deficiency • Cystic fibrosis • Congenital hypothyroidism • Congenital adrenal hyperplasia • Galactosaemia • Amino acid disorders • Fatty acid oxidation disorders		X	
9. Blood spot card storage and return	X	X	

Indicator 1: Screening coverage

Overall, samples were received from 59,192 newborns between January and December 2013. The number of newborns screened is determined by the number of unique NHI numbers for each DHB. Some instances of the same NHI number used for more than one infant in a DHB were found which explains why the total of infants counted in this way is slightly less than when counted by other parameters.

Data from National Maternity Collection of the Ministry of Health shows 59,707 babies were born in 2013. For screening, the numbers counted are babies screened within the calendar year. This might vary slightly from the number of babies in the calendar year.

Denominator data is sourced from the National Maternity Collection.

In 2013 approximately 99% of babies were screened. Table 2 outlines the numbers of babies screened and annual coverage from 2007 to 2013.

Table 2 Number of babies screened and coverage 2007 – 2013

Year	Births	Babies screened	Coverage %
2007	64,040	65,121	101.7
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

Table 3 outlines babies screened by DHB. Coverage by DHB ranges from 96 to 107%, indicating a mismatch of DHB data between the Ministry of Health maternity data and the NHI data.

Table 3 Number of babies screened by DHB, January to December 2013

DHB Region	Births	Babies screened	Coverage %
Northland	2,118	2,184	103.1
Waitemata	7,714	7,664	99.4
Auckland	6,321	6,168	97.6
Counties Manukau	8,238	8,204	99.6
Waikato	5,252	5,211	99.2
Lakes	1,423	1,382	97.1
Bay of Plenty	2,779	2,851	102.6
Tairawhiti	716	666	93.0
Taranaki	1,536	1,559	101.5
Hawkes Bay	2,170	2,184	100.6
Whanganui	831	862	103.7
Mid Central	2,109	2,155	102.2
Hutt Valley	1,930	1,867	96.7
Capital and Coast	3,665	3,580	97.7
Wairarapa	476	495	104.0
Nelson Marlborough	1,564	1,566	100.1
West Coast	377	381	101.1
Canterbury	5,890	5,851	99.3
South Canterbury	646	635	98.3
Southern	3,488	3,578	102.6
Not recorded*	464	149	32.1
Total	**59,707**	**59,192**	**99.1**

*includes babies born to mothers who usually reside overseas

Indicator 2: Timing of sample taking

This indicator is monitored by the number of screens performed. It is noted that some infants have more than one screen. In 2013, 59,192 babies had a screen and 59,356 screens were done. This includes 172 babies that had more than one screen.

The standard for this indicator is 95% of first samples are taken between 48 and 72 hours after birth. No DHB met the standard of 95%.

Table 4 identifies the percentage of samples taken by DHB. Nationally 71.4% of samples were collected at 48 to 72 hours, and 24.2% at greater than 72 hours.

Table 4 Timing of sample taking by DHB, January to December 2013

DHB region	Sampled at 48-72 hours		Sampled less than 48 hours		Sampled greater than 72 hours		No collection date and/or date of birth		Total number of screens*
	No.	%	No.	%	No.	%	No.	%	No.
Northland	1,475	67.4	11	0.5	627	28.6	76	3.5	2,189
Waitemata	5,709	74.3	43	0.6	1,732	22.5	199	2.6	7,683
Auckland	5,021	81.2	54	0.9	861	13.9	250	4.0	6,186
Counties Manukau	5,105	62.0	44	0.5	2,533	30.8	550	6.7	8,232
Waikato	2,950	56.4	37	0.7	2,022	38.7	217	4.2	5,226
Lakes	914	65.8	11	0.8	410	29.5	54	3.9	1,389
Bay of Plenty	1,477	51.7	15	0.5	1,253	43.8	114	4.0	2,859
Tairawhiti	529	79.0	0	0.0	125	18.7	16	2.4	670
Taranaki	1,208	77.2	10	0.6	306	19.6	40	2.6	1,564
Hawkes Bay	1,612	73.4	16	0.7	525	23.9	42	1.9	2,195
Whanganui	585	67.6	7	0.8	247	28.6	26	3.0	865
Mid Central	1,606	74.2	16	0.7	458	21.2	85	3.9	2,165
Hutt Valley	1,175	62.8	10	0.5	618	33.0	68	3.6	1,871
Capital and Coast	2,700	75.2	22	0.6	723	20.1	145	4.0	3,590
Wairarapa	341	68.8	1	0.2	137	27.6	17	3.4	496
Nelson Marlborough	1,215	77.5	7	0.4	305	19.5	40	2.6	1,567
West Coast	314	82.4	5	1.3	53	13.9	9	2.4	381
Canterbury	5,211	88.9	25	0.4	489	8.3	134	2.3	5,859
South Canterbury	525	82.5	2	0.3	101	15.9	8	1.3	636
Southern	2,621	73.1	25	0.7	812	22.7	126	3.5	3,584
Not recorded	94	63.1	5	3.4	27	18.1	23	15.4	149
Total	42,387	71.4	366	0.6	14,364	24.2	2,239	3.8	59,356*

*Total includes 172 babies who have had more than one screen

Figure 1 outlines the percentage of samples taken at 48 to 72 hours and shows that in 2013 no DHB met the 95% standard.

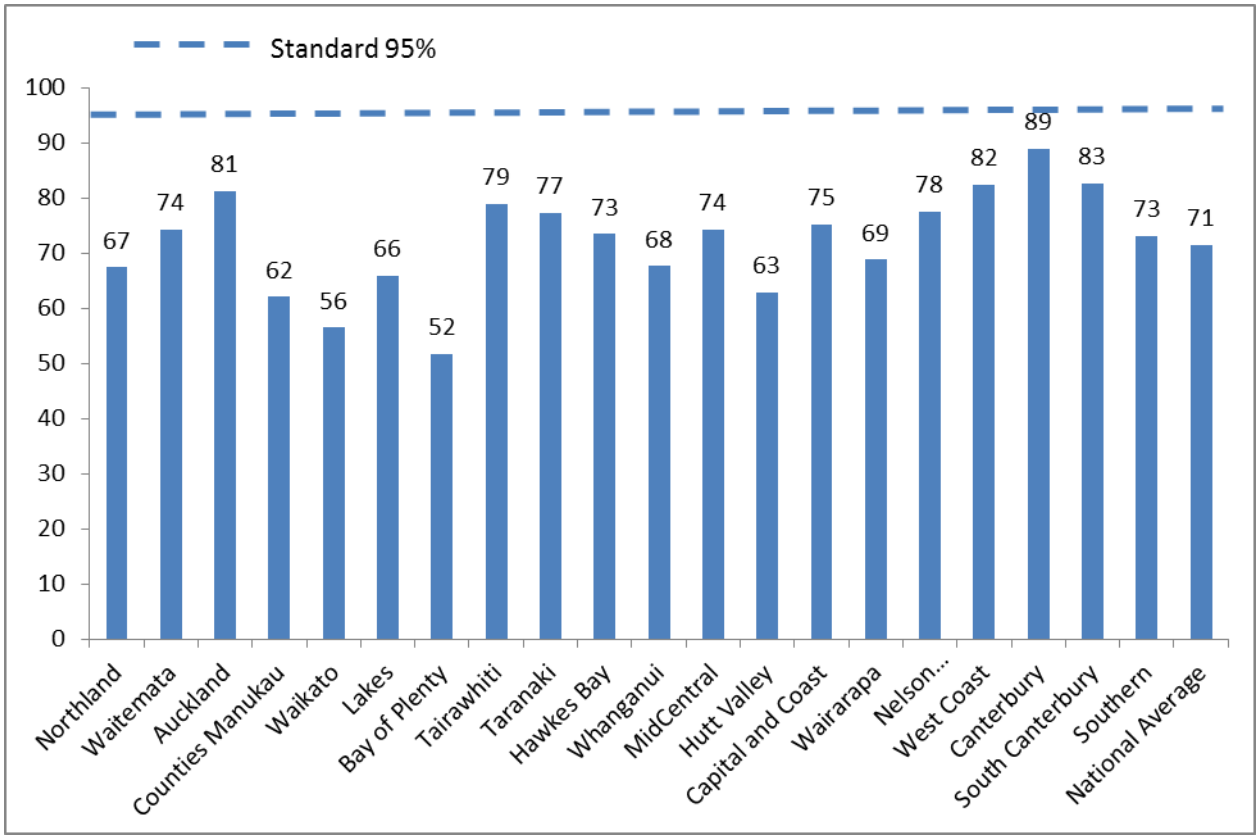

Figure 1 Percentage of samples taken at 48 to 72 hours, by DHB, January to December 2013

Table 5 shows detailed information by ethnicity. Māori (65%) and Pacific (64%) babies appear to be less likely than European (74%) and Asian (76%) babies to have a sample collected at two days. It is noted that the 95% standard was not met by any ethnic group.

Table 5 Timing of sample taking by Group 1 and Group 2 ethnicity, January to December 2013

Ethnicity (*Group 1 Group 2)*	**Sampled at 48-72 hours**		**Sampled less than 48 hours**		**Sampled greater than 72 hours**		**No collection date and/or date of birth**		**Total number of screens***
	No.	%	No.	%	No.	%	No.	%	No.
Māori	8,458	65.1	77	0.6	3,954	30.4	499	3.8	12,988
Pacific	3,936	63.8	46	0.7	1,871	30.3	314	5.1	6167
Cook Island Māori	562	60.6	11	1.2	303	32.7	51	5.5	927
Fijian	295	65.0	5	1.1	126	27.8	28	6.2	454
Niuean	219	65.6	3	0.9	98	29.3	14	4.2	334
Samoan	1,582	63.3	15	0.6	776	31.1	125	5.0	2,498
Tokelauan	75	60.5	2	1.6	42	33.9	5	4.0	124
Tongan	988	66.2	8	0.5	420	28.2	76	5.1	1492
Other Pacific	215	63.6	2	0.6	106	31.4	15	4.4	338
Asian	6,351	76.1	55	0.7	1,621	19.4	319	3.8	8,346
Chinese	2,400	80.2	17	0.6	484	16.2	90	3.0	2991
Indian	1,873	71.0	22	0.8	613	23.2	129	4.9	2,637
Southeast Asian	762	77.5	12	1.2	175	17.8	34	3.5	983
Other Asian	1,316	75.9	4	0.2	349	20.1	66	3.8	1,735
European	22,841	74.3	182	0.6	6,675	21.7	1,056	3.4	30,754
NZ European	19,635	74.2	164	0.6	5,766	21.8	899	3.4	26,464
Latin American / Hispanic	232	73.2	2	0.6	72	22.7	11	3.5	317
Other European	2,974	74.9	16	0.4	837	21.1	146	3.7	3,973
Other	801	72.8	6	0.5	243	22.1	51	4.6	1,101
African	262	73.8	2	0.6	80	22.5	11	3.1	355
Middle Eastern	344	74.0	2	0.4	102	21.9	17	3.7	465
Other/not known	195	69.4	2	0.7	61	21.7	23	8.2	281
Total	42,387	71.4	366	0.6	14,364	24.2	2,239	3.8	59,356*

*Includes 172 babies who had more than one first screen

Figure 2 outlines the percentage of samples taken at 48 to 72 hours by ethnicity. In 2013 the standard of 95% was not met for any ethnic group.

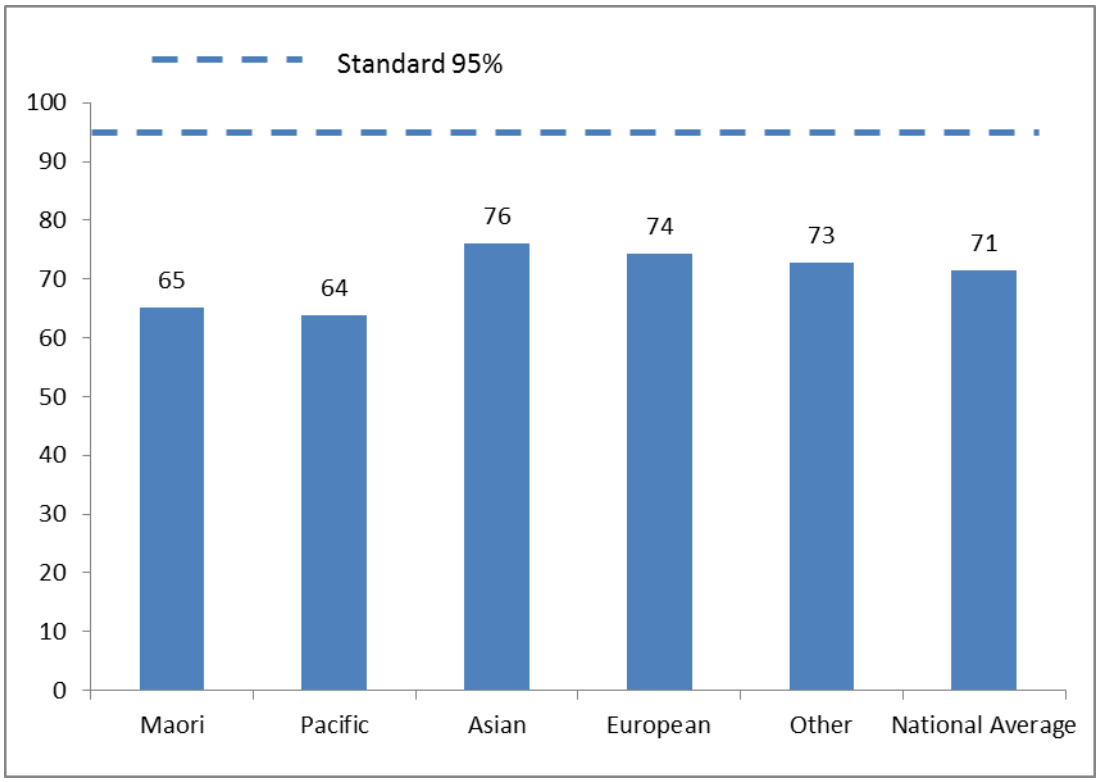

Figure 2 Percentage of samples taken at 48 to 72 hours, by ethnicity, January to December 2013

Table 6 shows the number of samples taken at two days by NZDep. The data indicates a slightly lower percentage of samples taken by the recommended time for babies in the five groups with the highest levels of deprivation (NZDep 6 to 10).

Table 6 Timing of sample taking by NZDep, January to December 2013

NZDep	Sampled at 48-72 hours		Sampled less than 48 hours		Sampled greater than 72 hours		No collection date and/or date of birth		Total number of screens*
	No.	%	No.	%	No.	%	No.	%	No.
1	2,985	77.9	16	0.4	709	18.5	123	3.2	3,833
2	3,483	76.0	23	0.5	906	19.8	170	3.7	4,582
3	3,604	75.5	32	0.7	975	20.4	162	3.4	4,773
4	3,579	76.6	30	0.6	903	19.3	162	3.5	4,674
5	4,069	72.9	38	0.7	1,262	22.6	210	3.8	5,579
6	4,024	72.1	27	0.5	1,320	23.7	210	3.8	5,581
7	4,551	71.2	39	0.6	1,590	24.9	212	3.3	6,392
8	5,290	69.8	42	0.6	1,994	26.3	255	3.4	7,581
9	5,391	68.0	61	0.8	2,155	27.2	325	4.1	7,932
10	5,311	64.2	53	0.6	2,522	30.5	387	4.7	8,273
Not recorded	100	64.1	5	3.2	28	17.9	23	14.7	156
Total	42,387	71.4	366	0.6	14,364	24.2	2,239	3.8	59,356*

*Includes 172 babies who had more than one first screen

Figure 3 identifies the percentage of samples taken at 48 to 72 hours by NZDep. In 2013 no New Zealand deprivation level decile reached the 95% target.

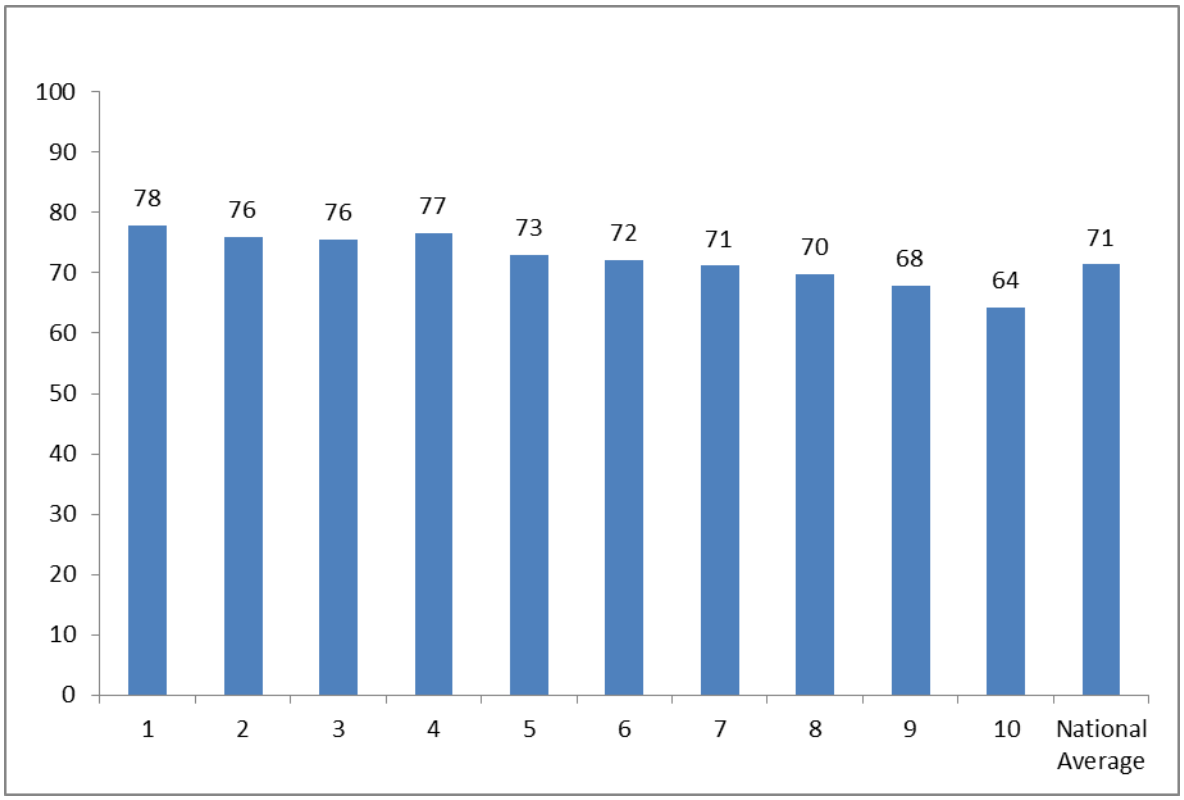

Figure 3 Percentage of samples taken at two days, by NZDep, January to December 2013

Indicator 3: Quality of blood samples

Accurate testing of blood spot samples is reliant on the quality of the blood spot sample. Unsatisfactory samples require a repeat sample which could have been avoided. Table 7 shows that eight DHBs met or exceeded the standard of 99% of blood spot samples suitable for testing.

Table 7 Percentage of blood samples that meet quality standards by DHB, January to December 2013

DHB region	Satisfactory		Unsatisfactory		Total samples
	No.	%	No.	%	No.
Northland	2,163	98.8	26	1.2	2,189
Waitemata	7,615	99.1	68	0.9	7,683
Auckland	6,119	98.9	67	1.1	6,186
Counties Manukau	8,107	98.5	125	1.5	8,232
Waikato	5,140	98.4	86	1.6	5,226
Lakes	1,372	98.8	17	1.2	1,389
Bay of Plenty	2,839	99.3	20	0.7	2,859
Tairawhiti	655	97.8	15	2.2	670
Taranaki	1,548	99.0	16	1.0	1,564
Hawkes Bay	2,166	98.7	29	1.3	2,195
Whanganui	850	98.3	15	1.7	865
Mid Central	2,143	99.0	22	1.0	2,165
Capital & Coast	1,858	99.3	13	0.7	1,871
Hutt Valley	3,533	98.4	57	1.6	3,590
Wairarapa	492	99.2	4	0.8	496
Nelson Marlborough	1,555	99.2	12	0.8	1,567
West Coast	372	97.6	9	2.4	381
Canterbury	5,814	99.2	45	0.8	5,859
South Canterbury	628	98.7	8	1.3	636
Southern	3,538	98.7	46	1.3	3,584
Not recorded	138	92.6	11	2.5	149
Total	58,645	98.8	711	1.2	59,356

*172 babies have two first samples.

Figure 5 outlines the reasons why samples were unsatisfactory. These included:

- 248 samples were collected too early (before 48 hours of age)
- 371 had a problem with the blood collection (such as insufficient blood, no demographics on the card or the sample was contaminated)

- 49 had a problem in transit (such as took longer than one month, flap folded onto wet blood causing significant loss onto the flap, damaged in transit or put wet into a plastic bag)

Second samples were requested from 711 babies and received from 650 (91.4%). The request was declined by nine families (1.3%); twelve babies died (1.7%) and the remaining 40 were lost to follow-up.

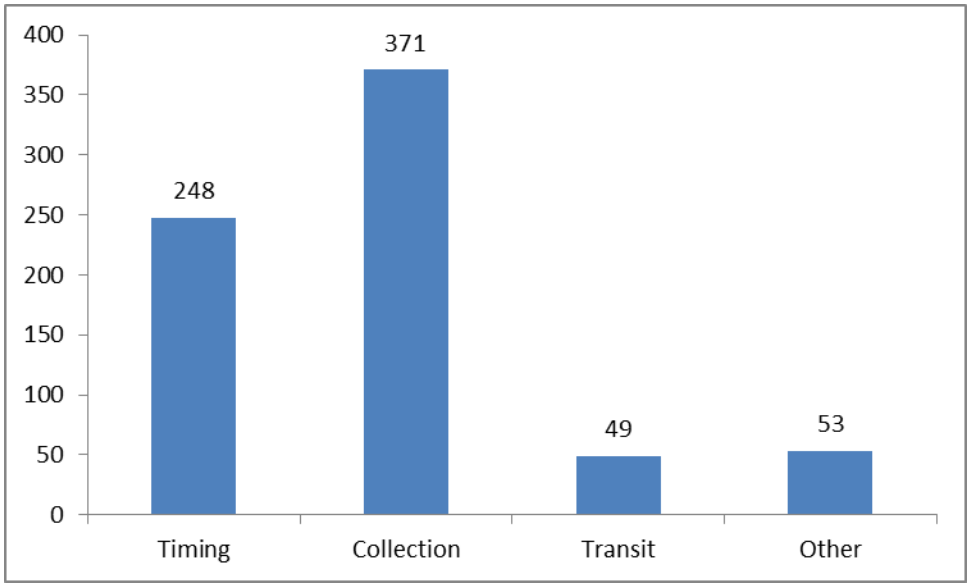

Figure 4 Reasons for unsatisfactory samples

Indicator 4: Sample dispatch and delivery

The NMSP relies on timeliness of sample dispatch and delivery. The standard is for 95% of samples to be received by the laboratory within four calendar days of being taken.

Table 8 shows that nationally 71.6% of samples were received within four days, and 27.0% received after four days. The range was 43.6 to 83.4%. Although postage paid envelopes have been supplied to specimen submitters and this has greatly improved transit times across all DHBs, no DHB met the standard of 95%. However there has been an improvement from 65.1% meeting the standard in 2011 to 68.8% in 2012 and further to 71.6% in 2013.

Table 8 Percentage of samples received by the laboratory within four days by DHB, January to December 2013

DHB region	Less than or equal to 4 days		Greater than 4 days		Unknown		Total samples
	No.	%	No.	%	No.	%	No.
Northland	1,556	71.1	596	27.2	37	1.7	2,189
Waitemata	5,884	76.6	1,726	22.5	73	1.0	7,683
Auckland	4,693	75.9	1,407	22.7	86	1.4	6,186
Counties Manukau	5,941	72.2	2151	26.1	140	1.7	8,232
Waikato	4,047	77.4	1,115	21.3	64	1.2	5,226
Lakes	1,094	78.8	262	18.9	33	2.4	1,389
Bay of Plenty	2,005	70.1	807	28.2	47	1.6	2,859
Tairawhiti	486	72.5	180	26.9	4	0.6	670
Taranaki	1,202	76.9	339	21.7	23	1.5	1,564
Hawkes Bay	958	43.6	1,219	55.5	18	0.8	2,195
Mid Central	721	83.4	131	15.1	13	1.5	865
Whanganui	1,685	77.8	442	20.4	38	1.8	2,165
Capital and Coast	1,225	65.5	619	33.1	27	1.4	1,871
Hutt Valley	2,509	69.9	1,034	28.8	47	1.3	3,590
Wairarapa	355	71.6	136	27.4	5	1.0	496
Nelson Marlborough	877	56.0	682	43.5	8	0.5	1,567
West Coast	273	71.7	106	27.8	2	0.5	381
Canterbury	3,898	66.5	1,887	32.2	74	1.3	5,859
South Canterbury	433	68.1	200	31.4	3	0.5	636
Southern	2,559	71.4	974	27.2	51	1.4	3,584
Not recorded	107	71.8	32	21.5	10	6.7	149
Total	**42,508**	**71.6**	**16,045**	**27.0**	**803**	**1.4**	**59,356**

Figure 5 details the percentage of samples received by the screening laboratory within four days or less from the date of sample taking. No DHB met the standard of 95% received within four days.

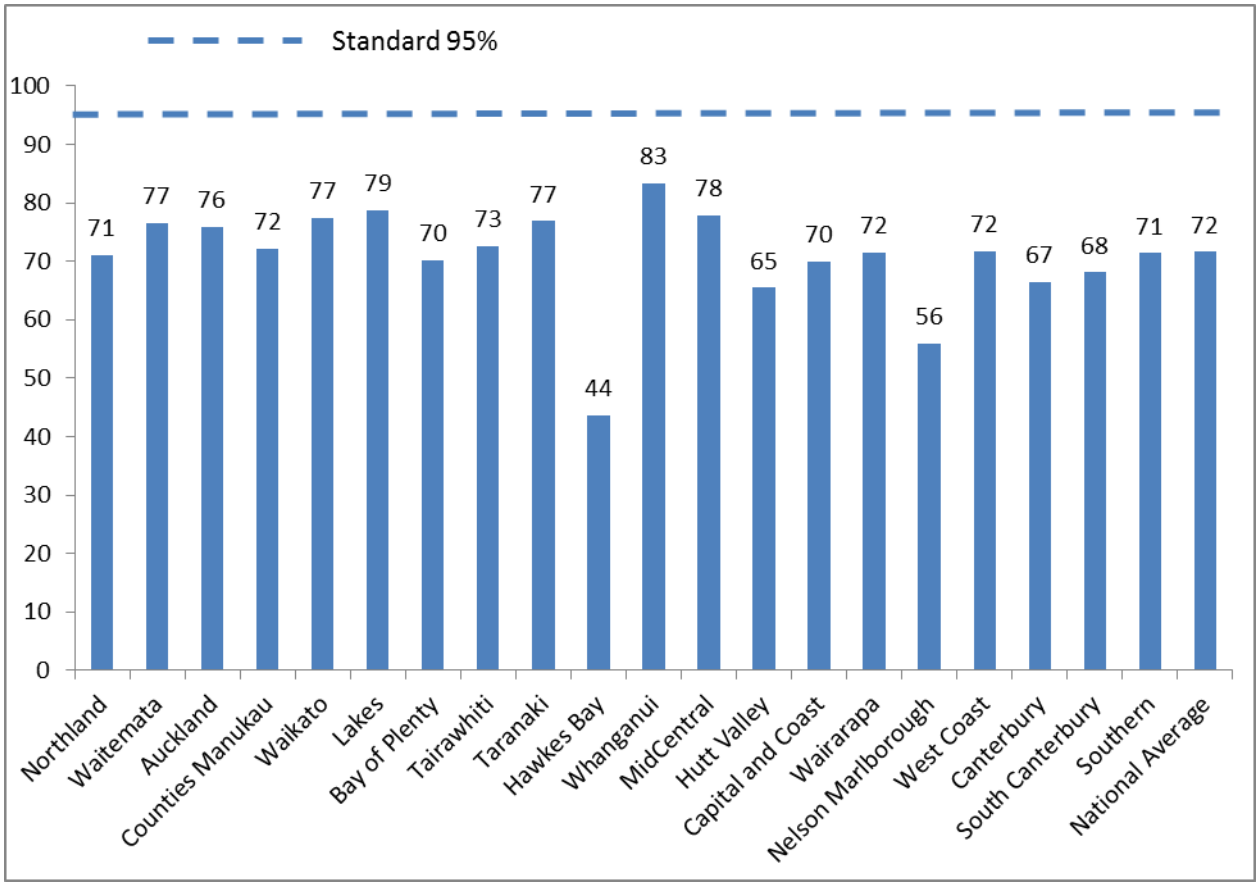

Figure 5 Percentage of samples received by the laboratory in four days or less, January to December 2013

Indicator 5: Laboratory testing timeframes

Table 9 identifies the percentage of samples that met the specified laboratory testing standard. The standard requires that 100% of samples meet the specified laboratory turnaround times. The range was 98.0% to 99.8%, and no disorder met the 100% standard. Delays in results for amino acid disorders and fatty acid oxidation disorders were due to instrument breakdowns as without a backup instrument analyses either wait for repair or for tests to be done by the New South Wales screening laboratory in Sydney. Delays in cystic fibrosis screening were due to delayed mutation analysis results.

Table 9 Percentage of results available within specified timeframes, by disorder, January to December 2013

Disorder	Standard for turnaround time (days)	Number met timeframe	% met timeframe
Congenital adrenal hyperplasia	2	59,169	99.7
Galactosaemia	2	59,238	99.8
Amino acid disorders	2	58,169	98.0
Fatty acid oxidation disorders	2	58,169	98.0
Biotinidase deficiency	5	59,288	99.9
Cystic fibrosis	5	58,820	99.1
Congenital hypothyroidism	5	59,204	99.7

(n= 59,356 samples)

Indicator 6: Timeliness of reporting – notification of screen positives

The NMSP relies on early detection and treatment. This ensures babies with congenital metabolic disorders have their development potential impacted as little as possible from the disorder. The standard for this indicator is that 100% of babies with positive results are notified to their LMC or referring practitioner by the timeframe specified for each disorder.

Most screening tests have a two-tier reporting system. Where results are highly likely to indicate the disorder is present, the results are telephoned to the LMC and referral made to an appropriate subspecialist paediatrician. All results in this category were reported inside the timeframes, as shown in Table 10. Marginal test results are reported by mail, and in this case the written report is not generated until all the screening test results are available. The results are available and will be phoned if there is a clinical reason to do so.

Table 10 Percentage of positive test results reported within specified timeframes, by disorder, January to December 2013

Reason for report	**Standard: Calendar days** (from receipt in lab to report)	**Number of positive test reports**	**Number met timeframe**	**% met timeframe**
Amino acid disorders	3	197	114	57.9
Fatty acid oxidation disorders	3	99	57	57.6
Congenital adrenal hyperplasia	3	78	46	59.0
Galactosaemia	3	4	2	50.0
Congenital hypothyroidism	4	60	46	76.7
Biotinidase deficiency	9	0	0	N/A
Cystic fibrosis	12	49	34	69.4

Of the reports which did not meet the turnaround time, the reasons include:

- waiting for cystic fibrosis gene testing or biotinidase deficiency screening results (all the delayed cystic fibrosis screen reporting was due to delayed gene results)
- waiting for amino acid and fatty acid oxidation screening results delayed due to breakdowns in the tandem mass spectrometer
- delay in sign-out
- a small number for other reasons.

Table 11 outlines the percentage of urgent clinical critical positive reports reporting by timeframes and disorder.
It is noted that the testing turnaround times are specified in working days but reported in calendar days. For example congenital adrenal hyperplasia (CAH) is two days for test result being available and three days for reporting. A sample which arrives on Friday and has a test result available and reported on Monday meets the testing timeframe but not the reporting timeframe.

Table 11 Percentage of urgent clinical critical positive results reported within specified timeframes, by disorder, January to December 2013

Reason for report	Standard: Calendar days (from receipt in laboratory to report)	Number of urgent critical positive test reports	Number met timeframe	% met timeframe
Amino acid disorders	3	17	15	88
Fatty acid oxidation disorders	3	10	9	90*
Congenital adrenal hyperplasia	3	5	3	60**
Galactosaemia	3	2	1	50**
Congenital hypothyroidism	4	14	12	86***
Biotinidase deficiency	9	0	0	N/A
Cystic fibrosis	12	0	0	0

*Outlier 6d.
**These babies in NICU
***Outliers at 5d and 6d.

Indicator 7: Collection and receipt of second samples

Second samples are requested when samples are not suitable for testing or there are minor elevations of screened metabolites. Table 12 outlines the follow-up of second samples requested by the screening laboratory by DHB. No DHB met the standard of 100% of second samples received by the laboratory, or declined, within ten calendar days of the request. The national average is 35.0%. Improvements have been made by the NMSP working closely with LMCs to ensure follow-up is completed before the baby is discharged from midwifery care at 4 to 6 weeks of age.

Table 12 Follow-up of requested second samples by DHB, January to December 2013

DHB region	Less than or equal to 10 days		Other follow up		No follow up		Follow up complete		Total samples
	No.	%	No.	%	No.	%	No.	%	No.
Northland	15	37.5	23	57.5	2	5.0	38	95.0	40
Waitemata	50	45.5	59	53.6	1	0.9	109	99.1	110
Auckland	44	45.4	49	50.5	4	4.1	93	95.9	97
Counties Manukau	66	37.9	102	58.6	6	3.4	168	96.6	174
Waikato	31	27.2	72	63.2	11	9.6	103	90.4	114
Lakes	7	25.0	20	71.4	1	3.6	27	96.4	28
Bay of Plenty	14	35.0	23	57.5	3	7.5	37	92.5	40
Tairawhiti	3	20.0	9	60.0	3	20.0	12	80.0	15
Taranaki	8	44.4	8	44.4	2	11.1	16	88.9	18
Hawkes Bay	11	28.9	27	71.1	0	0.0	38	100.0	38
Whanganui	7	36.8	12	63.2	0	0.0	19	100.0	19
Mid Central	18	45.0	20	50.0	2	5.0	38	95.0	40
Hutt Valley	6	28.6	13	61.9	2	9.5	19	90.5	21
Capital and Coast	21	26.3	59	73.8	0	0.0	80	100.0	80
Wairarapa	3	50.0	3	50.0	0	0.0	6	100.0	6
Nelson Marlborough	6	30.0	11	55.0	3	15.0	17	85.0	20
West Coast	1	10.0	9	90.0	0	0.0	10	100.0	10
Canterbury	25	32.9	50	65.8	1	1.3	75	98.7	76
South Canterbury	1	10.0	9	90.0	0	0.0	10	100.0	10
Southern	17	28.3	42	70.0	1	1.7	59	98.3	60
Not recorded	3	75.0	1	25.0	0	0.0	4	100.0	4
Total	357	35.0	621	60.9	42	4.1	978	95.9	1020

Note: follow-ups are not counted in the total number of screens. Follow-up samples include those that were unsuitable for testing or were suspected of a disorder.

Indicator 8: Diagnosis and commencement of treatment by disorder

The NMSP relies on confirmed detection and timely treatment to ensure babies with congenital metabolic disorders have their development potential impacted as little as possible from the disorder.

The standard is for 100% of babies who receive a screen positive result to be diagnosed and commence treatment by the time specified for each disorder.

The time to diagnosis and commencement of treatment is determined by the age of the baby when the specimen was collected, the transit time to the laboratory, the time to confirmation and reporting of test results and the time to make the diagnosis and commence treatment. The summarised numbers of detected cases of the screened disorders and the number treated by the specified age are given in Table 13.

Table 13 Age at treatment, January to December 2013

Disorder	Standard: Calendar days of age of baby at treatment commenced	Number of cases	Number treated by specified age
Biotinidase deficiency	14	0	N/A
Cystic fibrosis	28	9	3
Congenital hypothyroidism	10	22	12
Congenital adrenal hyperplasia	10	3	3
Galactosaemia	10	0	N/A
Amino acid disorders	10	7	3
Fatty acid oxidation disorders	10	10	8

Of the babies diagnosed with cystic fibrosis, the NMSP has data on six. Three were treated before 28 days, the others at 29.35 and 44 days of age.

Data is only available for one of the babies with CAH, baby was treated at 6 days. One baby had ambiguous genitalia and the screening sample was collected on the day of birth so this baby was most likely treated early. The other baby was seen by Paediatric Endocrinology at 6 days and had abnormal electrolytes so was probably treated at day 6 or 7.

There were six cases of CH treated outside the timeframe as their initial levels of TSH were between 15 and 29 mIU/L and notification was made following the results of a second sample. Of the remaining four, three were treated at 11 or 12 days and one not until 44 days (notified at 8 days).

Of the four patients with amino acid breakdown disorders not treated by the specified age, three had PKU treated at 11, 12 and 15 days. No information is available for one patient. The two patients with fatty acid oxidation disorders not treated by the specified age were treated at 11 days.

Indicator 9: Blood spot card storage and return

Where requested, blood spots are to be returned to parents/guardians/individuals within 28 days of the request.

Of 594 requests for blood spot returns, 586 (98.6%) were returned in the timeframe. Five cards were not returned as they had insufficient information. This was requested but not provided. The remaining three samples were returned in 34 or 35 days after the original request but within a day or two of receiving either a second sample or complete information for the return. In general samples are returned very quickly with a median time over this period of 2.1 days.

The NMSP Policy Framework lists possible secondary uses for residual screening cards (Section 4.1(b)).

Table 14 shows reasons for secondary use of cards. For the period 1 January 2013 to 31 December 2013, 60 cards were used for the benefit of the individual and family/whanau. 56 were for cytomegalovirus (CMV) testing to determine whether congenital CMV was the cause of symptoms in the baby or child. The remaining four were used for other viral studies (1), genetic studies (2) and investigation of late diagnosis of CAH (1).

Table 14 Reasons for secondary use, January to December 2013

Secondary use	Number
Benefit of the individual and family/whanau	60
Forensic/police/coroner investigations	0
Mortality review	0
Research	0
Other	0
Total	**60**

Screening performance and incidence

Follow-up of positive tests

Follow-up of positive tests is detailed in Table 15. Appropriate follow-up may be:

- a specialist paediatrician visit
- a further dried blood sample
- a test done elsewhere
- notification that baby has died.

Overall 411 babies were referred for paediatric examination or had a request for a second sample because of a positive screen result and 715 because of an unsuitable sample.

Table 15 Newborn screening follow-up by condition, 2013

Condition	**Number of positive tests**	**Follow-up by scheduled NICU or requested sample** (e.g. sample taken too early)	**Other follow-up** (paediatric referral, second test)	**Number with appropriate follow-up**	**% with appropriate follow-up**
Aminoacid breakdown and fatty acid oxidation disorders	220	0	218	218	99
Biotinidase deficiency	3	0	0	0	N/A
Congenital adrenal hyperplasia	78	0	78	78	100
Congenital hypothyroidism	60	2	58	60	100
Cystic fibrosis	46	0	46	46	100
Galactosaemia	4	0	4	4	100

NB: A borderline result when a further sample is scheduled is no longer counted as a positive screen as the screening consists of more than one sample.

Clinical utility

Newborn screening is justified for conditions in which there is clinical benefit from diagnosis made earlier by screening than it would be made by clinical presentation and diagnosis. Screening audit forms contain a question about whether the diagnosis was suspected before the screening test result is available. Reasons for clinical detection are:

- family history (FH)
- meconium ileus or other clinical signs.

The numbers are given in Table 16.

Table 16 Clinical utility of screening, 2013

Disorder	Number of cases	Diagnosis suspected before screen result	Reason for suspicion
Biotinidase deficiency	0	0	
Cystic fibrosis	9	4	3 FH, 1 meconium peritonitis
Congenital hypothyroidism	22	0	
Congenital adrenal hyperplasia	3	2	2 ambiguous genitalia
Galactosaemia	0	0	
Amino acid disorders	7	3	2 FH, 1 symptomatic
Fatty acid oxidation disorders	10	4	3 FH, 1 symptomatic
Total conditions	51	13	

One baby with PKU presented clinically (not included in screening statistics as family declined screening for baby). Overall 51 cases of screened disorders were detected by the programme and in 38 of these there was no clinical suspicion of the disorder before the screening test result.

Incidence of screened disorders

Amino acid breakdown disorders

Since screening started in 2006, 505,852 infants have been screened and 60 cases detected, giving an incidence of 1:8,400. This includes PKU, MSUD and hyperphenylalaninemia.

PKU

There were three cases of PKU found in 2013. Since screening started in 1969 2,584,585 infants have been screened for PKU and 121 cases found, none notified missed, to give an incidence of 1:21,360. Benign hyperphenylalaninemia is not counted in this incidence figure. It is problematic comparing PKU incidence as the definition of the disorder is 'a level of phenylalanine that requires treatment' and the level has varied time to time.

MSUD

No cases of MSUD were found in 2013. Since screening started in 1969, 2,584,585 infants have been screened for MSUD, ten classical cases found; none were notified missed, giving an incidence of 1:258,000.

Biotinidase deficiency

Since screening started in 1986, 1,651,291 infants have been screened and eight cases detected giving an incidence of 1:206,000.

Congenital adrenal hyperplasia

Since screening started in 1986, 1,698,240 infants have been screened and 73 cases detected giving an incidence of 1:23,200.

Congenital hypothyroidism

Since screening started in 1981, 1,918,656 infants have been screened and 508 cases detected giving an incidence of 1:3777. There is a trend to an increasing incidence of CH in New Zealand. The increase is in dyshormonogenesis. This condition is more common in people of Asian and Pacific origin and the increase coincides with an increase in Asian births as immigration changes the New Zealand demographic.

Cystic fibrosis

Since screening started in 1983, 2,633,872 infants have been screened and 385 cases detected giving an incidence of 1:6,841. There were 59,192 babies screened in 2013 (of whom approximately 47% were of European ethnicity) and 9 cases of CF detected. This gives an incidence in the European births of 1:3,103.

Fatty acid oxidation disorders

Since screening started in 2006, 505,852 infants have been screened and 61 cases detected giving an incidence of 1:.8,300.

Galactosaemia

Since screening started in 1973, 2,472,680 infants have been screened and 24 cases detected giving an incidence of 1:103,000.

Appendix 1: NMSP National Indicators

1: NEWBORN METABOLIC SCREENING COVERAGE
DESCRIPTION The proportion of babies who have had newborn metabolic screening.
RATIONALE All babies whose parents/guardians consent to screening should have screening.
RELEVANT OUTCOME All babies whose parents/guardians consent to newborn metabolic screening are screened.
STANDARD 100% of babies whose parents/guardians consent to screening are screened.
METHODOLOGY ***Indicator 1.1*** Numerator: Number of babies screened. Denominator: Number of live births.
NOTES • Denominator limitations to be explained in published reports • Reporting by: ➢ DHB ➢ Ethnicity ➢ Deprivation status

2. TIMING OF SAMPLE TAKING

DESCRIPTION

1. The proportion of eligible babies who have a newborn metabolic screening sample taken.
2. The proportion of eligible babies who have a newborn metabolic screening sample taken between 48 and 72 hours of birth.

RATIONALE

Timely sample collection leads to the best possible chance of a baby receiving early diagnosis and treatment where necessary. Severe forms of some of the disorders screened for can be fatal within seven to ten days. 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 their indicator biochemicals to show an abnormality. Therefore the optimum window for sample collection is between 48 and 72 hours of birth.

RELEVANT OUTCOME

Babies screened should have a newborn metabolic screening sample taken between 48 and 72 hours of birth.

STANDARD

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

METHODOLOGY

Indicator 2

Numerator: Number of babies who have a newborn metabolic screening sample taken between 48 and 72 hours of birth. (see data limitations above, the measure used in this report is the number of babies screened at 2 days)

Denominator: Number of babies who have a newborn metabolic screening sample taken.

NOTES

- Samples for screening must be taken in accordance with Programme Guidelines and Policy and Quality requirements.
- Reporting by:
  - DHB
  - Ethnicity
  - Deprivation status

3. QUALITY OF SAMPLE

DESCRIPTION

The quality of the blood spot sample.

RATIONALE

Accurate testing of blood spot samples is reliant on the quality of the sample. Unsatisfactory samples require a repeat sample which could have been avoided.

RELEVANT OUTCOME

Blood spot samples are of sufficient quality for laboratory testing for screened disorders.

STANDARD

99% of blood spot samples are of satisfactory quality.

METHODOLOGY

Indicator 3

Numerator: Number of samples of satisfactory quality as reported by the laboratory.

Denominator: Number of samples taken.

NOTES

- Requirements for a satisfactory sample are detailed in Chapter 7, page 21-22 of Programme Guidelines.
- Reporting by DHB

4: SAMPLE DISPATCH AND DELIVERY

DESCRIPTION

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

RATIONALE

The NMSP relies on timeliness. Samples must be sent to the laboratory as soon as they are dry. Samples must be received by the laboratory as soon as possible after they are taken.

RELEVANT OUTCOME

Samples are received by the laboratory within four days of being taken.

STANDARD

95% of samples are received by the laboratory within four calendar days of being taken.

METHODOLOGY

Indicator 4

Numerator:	Number of samples received by laboratory within four calendar days of being taken.
Denominator:	Number of samples received by laboratory.

NOTES

- Requirements for sending samples to the laboratory are detailed in Chapter 7, page 23 of Programme Guidelines
- Reporting by DHB

5: LAB TESTING TIMEFRAMES

DESCRIPTION

The time taken by the laboratory to test each sample for each of the specified disorders (turnaround time).

RATIONALE

Samples should be tested as soon as possible to ensure that screen positives can be acted on as quickly as possible to reduce / minimise avoidable harm.

RELEVANT OUTCOMES

All samples are tested within the specified timeframes.

Samples received before 07:30am are tested the same day.

STANDARD

100% of samples meet the following laboratory turnaround times:

Disorder	Working days (from receipt by laboratory)
Congenital Adrenal Hyperplasia	2
Galactosaemia	2
Amino acid disorders	2
Fatty acid oxidation disorders	2
Biotinidase deficiency	5
Cystic fibrosis	5
Congenital Hypothyrodism	5

METHODOLOGY

Indicator 5

Numerator: Number of samples tested and reported within specified timeframes.

Denominator: Number of samples tested.

6: TIMELINESS OF REPORTING – NOTIFICATION OF SCREEN POSITIVES

DESCRIPTION

The time taken for a baby with a positive screening result to be referred for diagnostic testing.

RATIONALE

The NMSP relies on early detection and treatment. This ensures babies with congenital metabolic disorders have their development potential impacted as little as possible from the disorder.

RELEVANT OUTCOME

All babies with positive screening results are referred for further testing within the specified timeframes after results become available.

STANDARD

100% of babies with positive results are notified to their LMC / referring practitioner by the laboratory within the following timeframes:

Reason for report	Calendar days (from receipt in lab test result)
Amino acid disorders	3
Fatty acid oxidation disorders	3
CAH	3
Galactosaemia	3
CH	4
Biotinidase deficiency	9
Cystic fibrosis	12

METHODOLOGY

Indicator 6

Numerator: Number of babies who are notified to their referrer for further testing for a particular disorder within the number of calendar days specified for that disorder.

Denominator: Number of babies who receive a positive screening result for a particular disorder.

7: COLLECTION AND REPEAT OF SECOND SAMPLES

DESCRIPTION

The number of babies that have had second samples taken, sent, and received by the laboratory. **Note**: this indicator does not cover highly positive samples. It is for those around the cut off who have letters sent to them.

RATIONALE

If a second sample is required it means that a baby has not been fully screened, or that his/her results were borderline. Second samples should be taken as soon as possible so that the baby can be treated early if he/she has a disorder.

RELEVANT OUTCOME

Second samples are taken, sent, and received by the laboratory as soon as possible.

STANDARD

100% of second samples are received by the laboratory, or declined, within ten calendar days of request.

METHODOLOGY

Indicator 7.1

Numerator: Total number of second samples collected, declined, or baby died.

Denominator: Number of second samples requested.

Indicator 7.2

Numerator: Number of second samples received within ten calendar days.

Denominator: Total number of second samples received and declined.

NOTES

- Requirements for repeat samples are detailed in Chapter 7, page 24-25 of Programme Guidelines.
- Reporting by DHB

8: DIAGNOSIS AND COMMENCEMENT OF TREATMENT BY DISORDER

DESCRIPTION

The number of babies with a positive screening result who receive a confirmed diagnosis and timely commencement of treatment.

RATIONALE

The NMSP relies on confirmed detection and timely treatment to ensure babies with congenital metabolic disorders have their development potential impacted as little as possible from the disorder.

RELEVANT OUTCOME

All babies with a metabolic disorder and a screen positive result receive a confirmed diagnosis and timely commencement of treatment.

STANDARD

100% of babies who receive a screen positive result are diagnosed and commence treatment by:

Disorder	Calendar days
Biotinidase deficiency	14
Cystic fibrosis	28
CH	10
CAH	10
Galactosaemia	10
Amino acid disorders	10
Fatty acid oxidation disorders	10

METHODOLOGY

Indicator 8

Numerator: Number of babies who are diagnosed and commence treatment within the timeframes specified.

Denominator: Number of babies who receive a screen positive result and are diagnosed with and treated for a metabolic disorder.

NOTES

- Clinically-diagnosed babies will be reported separately.
- Measurement may also be by case review or periodic audit / evaluation.

9: CARD STORAGE AND RETURN

DESCRIPTION

The time taken for the laboratory to return requested blood spot cards to parents/guardians/individuals.

RATIONALE

Where requested blood spot cards should be returned within:

- 28 days of completion of screening
- 28 days of valid (fully completed) request for return.

RELEVANT OUTCOME

All blood spot cards are returned to parents/guardians/individuals by tracked courier within 28 days.

STANDARD

1. Where requested, 100% of blood spot cards are returned to parents/guardians within 28 days of completion of screening.
2. 100% of blood spot cards are returned to the authorised person by tracked courier within 28 calendar days of valid request.

METHODOLOGY

Indicator 9

Numerator: Number of blood spot cards returned within 28 days.

Denominator: Number of blood spot cards requested by parents/guardians/individuals.

NOTES

- Complete information is required by the laboratory in order to process requests for return of blood spot cards, as per Programme Guidelines in Chapter 11.