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Newborn Metabolic Screening Programme

Annual Report

January to December 2011

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 2011

- 61,859 babies were tested and 62,733 born giving coverage of 99%.
- 48% of samples were collected in the 48-72 hours timeframe. No DHB and no ethnic group met the 95% standard.
- Overall 99% of samples were suitable for testing. 14 DHBs were close or met the standard of 99% suitable.
- 65% 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 it was over 99% for all except for cystic fibrosis, fatty acid oxidation and amino acid disorders.
- Of the 63 clinical critical results, 62 were notified within the timeframe, and 1 was delayed.
- 37% of second samples were received in ten days or less (the standard is 100%), 98% of follow-up was completed appropriately.
- Of babies with diagnosed disorders, treatment was commenced by the specified age for between 29 to 62%. Most of the remainder were just outside the range and there were no reported clinical ill-effects from the delays.
- 98% of the 642 requests for card returns were made in 28 days or less (the standard is 100%).
- 46 cards were used for additional testing for family health reasons, mostly for CMV testing, and forensic/police/coroner investigations.
- During 2007 to 2011, screening sensitivities were 94.78 to 100%, with specificities 99.64 to 99.99% and positive predictive values 2.65 to 50.94.
- Follow-up of positive tests was between 50 to 100%. All clinical critical results had appropriate follow-up.

- 64 cases of screened disorders were detected by the programme and in 56 of these there was no clinical suspicion of the disorder before the screening test result.

- As a response to delayed transit times postage paid envelopes are being distributed to sample submitters.

- High quality lancets are now supplied to sample takers to reduce the number of unsuitable samples received.

1 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 2011. Reports are published on the National Screening Unit (NSU) website.

This is the first 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

1.1 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 Eire), 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.

Testing moved into the public healthcare system in 1991 and is now within LabPLUS at Auckland District Hospital. 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 aminoacid 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 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.

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 Governance Team and the Technical Group reviews Monitoring Reports and makes recommendations.

1.2 The aim of the Newborn Metabolic Screening Programme

The aim of the NMSP is to reduce newborn 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.

1.3 Data included in this report

Data is first 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 district health boards (DHBs), 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). This method follows a matching and data retrieval process that is defined within the business rules.

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 from 1 January to 31 December 2011 are included. 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.

Ethnicity is prioritised based on the NHI ethnicity information. All reporting by NZDep decile is based on the extraction against the NHI associated with residential addresses. Decile 1 is the highest and decile 10 is the lowest decile rating.

While many Lead Maternity Carers (LMCs) are not directly responsible to a particular DHB, data is reported by DHB region, as DHBs are responsible for the health of their population.

1.4 National Monitoring Indicators

Table 1 summarises all the NMSP indicators used in 2011 for regular monitoring with their reporting frequency and detail included in Appendix 2. This report, as an annual report, provides information on all 9 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	Quarterly	Biannually	Annually	Detail
1. Newborn Metabolic Screening Coverage			X	• DHB • Ethnicity • Deprivation status
2. Timing of sample taking	X	X	X	• DHB • Ethnicity • Deprivation status
Laboratory reporting				
3. Quality of Blood Samples	X	X	X	• DHB
4. Sample dispatch and delivery	X	X	X	• DHB
5. Laboratory testing timeframes	X	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	X	

2. Indicator 1: Screening coverage

Overall samples were received from 61,859 newborns between January and December 2011. 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 have been found which explains why the total number of infants counted in this way is slightly less than when counted by other parameters.

Data from National Collections of the Ministry of Health shows 62,733 babies were born in 2011. This includes 34 babies who were born to mothers that usually reside overseas. For screening, the numbers counted are babies screened within the calendar year. This might vary slightly from the number of babies born in the calendar year. Approximately 99% of babies were screened.

Table 2 outlines the numbers of babies' screened and annual coverage from 2007 to 2011.

Table 2 Number of babies screened and coverage 2007 – 2011

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

Table 3 outlines babies screened by DHB. Coverage by DHB ranges from 92.3% to 101.6%. The coverage rate for 17 DHBs was around or above the national average.

Table 3 Number of babies screened by DHB, January to December 2011

DHB Region	Births	Babies screened	Coverage %
Northland	2,320	2,357	101.6
Waitemata	7,917	7,800	98.5
Auckland	6,643	6,300	94.8
Counties Manukau	8,801	8,683	98.7
Waikato	5,438	5,466	100.5
Lakes	1,613	1,583	98.1
Bay of Plenty	2,897	2,936	101.3
Tairawhiti	771	712	92.3
Taranaki	1,586	1,605	101.2
Hawkes Bay	2,278	2,270	99.6
Whanganui	830	848	102.2
Mid Central	2,357	2,318	98.3
Hutt Valley	2,080	2,062	99.1
Capital and Coast	3,899	3,743	96.0
Wairarapa	538	549	102.0
Nelson Marlborough	1,677	1,690	100.8
West Coast	412	425	103.2
Canterbury	6,091	6,175	101.4
South Canterbury	575	594	103.3
Southern	3,702	3,679	99.4
Not recorded*	308	64	20.8
Total	**62,733**	**61,859**	**98.6**

*includes babies born to Mothers who usually reside overseas

Table 4 outlines the number of births and number of babies screened by ethnicity. In 2011 the coverage rate for Maori (85.2%) and MELAA (89.6%) was significantly lower than the national average (98.6%).

Table 4 Number of babies screened by ethnicity, January to December 2011

	Births	Babies screened	Coverage %
Maori	16,804	14,315	85.2
Pacific	7,127	6,937	97.3
Asian	7,326	7,174	97.9
European	30,092	32,061	106.5
MELAA*	1,202	1,077	89.6
Other	216	295	136.6
Total	**62,733**	**61,859**	**98.6**

* MELAA is Middle Eastern, Latin American and African

3 Indicator 2: Timing of sample taking

This indicator is monitored by the number of screens performed. It is noted that some infants had more than one screen. In 2011, 61,859 babies had a screen and 62,037 screens were done. This includes 178 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. For most of 2011 the screening laboratory system was unable to calculate the age in hours. The age in days has therefore been used and will underestimate the number of samples collected between 48 and 72 hours.

Table 5 identifies the percentage of samples taken by DHB. Nationally 48.4% of samples were collected at two days, and 49.6% of samples were taken at greater than two days.

Table 5 Percentage of samples taken at two days, by DHB, January to December 2011

DHB region	Sampled at 2 days		Sampled less than 2 days		Sampled greater than 2 days		No Collection Date and/or Date of Birth		Total number of screens*
	No.	%	No.	%	No.	%	No.	%	No.
Northland	1,020	*43.2*	13	*0.6*	1,288	*54.5*	42	*1.8*	2,363
Waitemata	3,807	*48.7*	56	*0.7*	3,876	*49.6*	76	*1.0*	7,815
Auckland	3,459	*54.8*	46	*0.7*	2,725	*43.1*	87	*1.4*	6,317
Counties Manukau	3,358	*38.5*	39	*0.4*	5,226	*60.0*	89	*1.0*	8,712
Waikato	1,946	*35.5*	35	*0.6*	3,414	*62.4*	79	*1.4*	5,474
Lakes	652	*40.9*	11	*0.7*	904	*56.7*	26	*1.6*	1,593
Bay of Plenty	875	*29.7*	9	*0.3*	1,999	*67.8*	64	*2.2*	2,947
Tairawhiti	242	*33.8*	4	*0.6*	456	*63.7*	14	*2.0*	716
Taranaki	991	*61.6*	11	*0.7*	592	*36.8*	15	*0.9*	1,609
Hawkes Bay	1,230	*53.9*	17	*0.7*	999	*43.8*	34	*1.5*	2,280
Whanganui	358	*42.0*	7	*0.8*	481	*56.4*	7	*0.8*	853
Mid Central	1,162	*50.0*	16	*0.7*	1,108	*47.7*	39	*1.7*	2,325
Hutt Valley	684	*32.9*	6	*0.3*	1,356	*65.3*	30	*1.4*	2,076
Capital and Coast	1,978	*52.8*	21	*0.6*	1,699	*45.3*	49	*1.3*	3,747
Wairarapa	271	*49.4*	3	*0.5*	268	*48.8*	7	*1.3*	549
Nelson Marlborough	947	*56.0*	6	*0.4*	717	*42.4*	22	*1.3*	1,692
West Coast	268	*62.9*	7	*1.6*	144	*33.8*	7	*1.6*	426
Canterbury	4,493	*72.6*	35	*0.6*	1,557	*25.2*	103	*1.7*	6,188
South Canterbury	374	*63.0*	2	*0.3*	216	*36.4*	2	*0.3*	594
Southern	1,883	*50.9*	33	*0.9*	1,730	*46.8*	52	*1.4*	3,698
Not recorded	31	*49.2*	0	*0.0*	26	*41.3*	6	*9.5*	63
Total	**30,029**	*48.4*	377	*0.6*	**30,781**	*49.6*	**850**	*1.4*	**62,037**

*Total includes babies who have had more than one screen

Figure 1 outlines the percentage of samples taken at two days and shows that no DHB met the 95% standard.

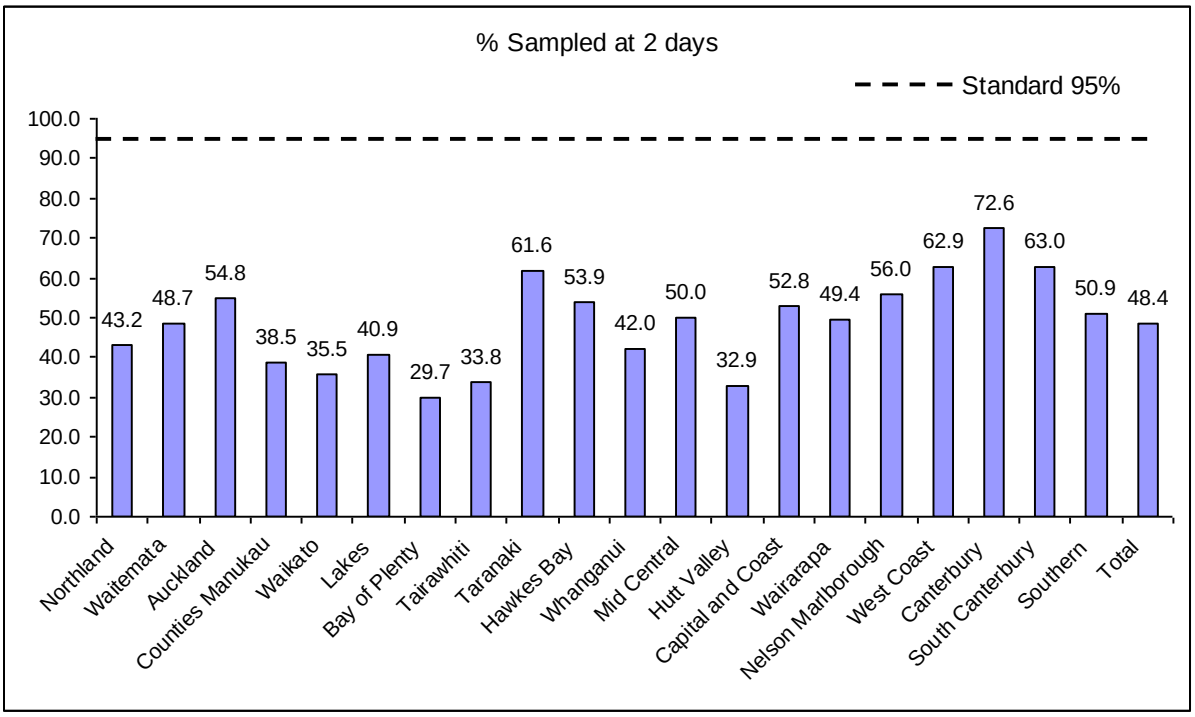

Figure 1 Percentage of samples taken at two days, by DHB, January to December 2011

Table 6 shows detailed information by ethnicity. Maori and Pacific babies appear to be less likely than European and Asian babies to have a sample collected at two days. It is noted that the 95% standard was not met by any ethnic group.

Table 6 Percentage of samples taken at two days, by Group 1 and Group 2 Ethnicity, January to December 2011

Ethnicity (*Group 1 Group 2)*	**Sampled at 2 days**		**Sampled less than 2 days**		**Sampled greater than 2 days**		**No Collection Date**		**Total number of screens**
	No.	%	No.	%	No.	%	No.	%	No.
Maori	5,994	41.7	94	0.7	8,055	56.1	221	1.5	14,364
Pacific	2,934	42.1	39	0.6	3,908	56.1	80	1.1	6,961
Cook Island Maori	412	39.7	8	0.8	602	58.0	16	1.5	1,038
Fijian	214	44.3	1	0.2	264	54.7	4	0.8	483
Niuean	172	44.0	2	0.5	213	54.5	4	1.0	391
Samoan	1,260	42.0	16	0.5	1,696	56.5	29	1.0	3,001
Tokelauan	58	44.6	1	0.8	71	54.6		0.0	130
Tongan	693	42.8	8	0.5	899	55.5	21	1.3	1,621
Other Pacific	125	42.1	3	1.0	163	54.9	6	2.0	297
Asian	3,594	49.9	46	0.6	3,487	48.4	75	1.0	7,202
Chinese	1,340	53.6	15	0.6	1,123	45.0	20	0.8	2,498
Indian	982	43.6	19	0.8	1,221	54.2	30	1.3	2,252
Southeast Asian	404	52.6	3	0.4	350	45.6	11	1.4	768
Other Asian	868	51.5	9	0.5	793	47.1	14	0.8	1,684
European	1,6908	52.2	190	0.6	1,4817	45.8	449	1.4	32,364
NZ European	1,4917	52.1	174	0.6	1,3142	45.9	379	1.3	28,612
Latin American / Hispanic	131	57.2	0	0.0	95	41.5	3	1.3	229
Other European	1,860	52.8	16	0.5	1,580	44.8	67	1.9	3,523
Other	599	52.3	8	0.7	514	44.9	25	2.2	1,146
African	213	51.2	4	1.0	190	45.7	9	2.2	416
Middle Eastern	219	50.6	2	0.5	205	47.3	7	1.6	433
Other/not known	167	56.2	2	0.7	119	40.1	9	3.0	297
Total	**30,029**	***48.4***	**377**	***0.6***	**30,781**	***49.6***	**850**	***1.4***	**62,037**

Figure 2 outlines the percentage of samples taken at two days by ethnicity. The standard of 95% was not met for any ethnic group.

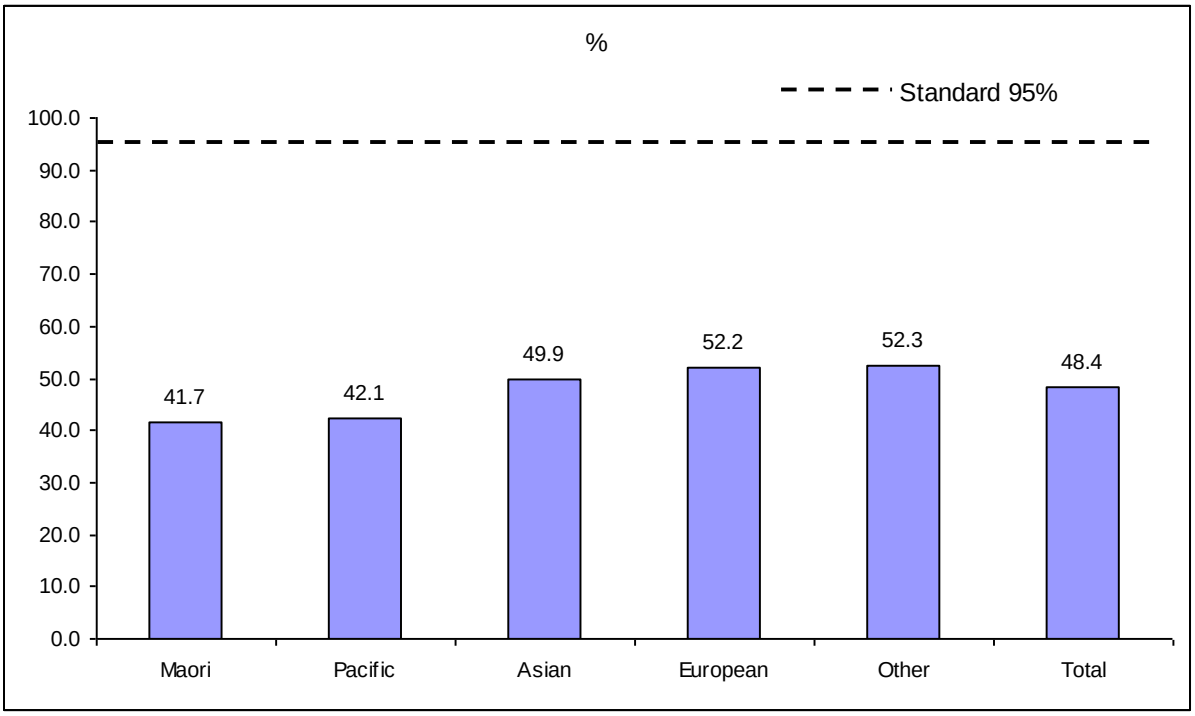

Figure 2 Percentage of samples taken at two days, by ethnicity, January to December 2011

Table 7 shows the number of samples taken at two days by NZDep index. The data does seem to indicate 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 7 Percentage of samples taken at two days by NZDep, January to December 2011

NZDep	Sampled at 2 days		Sampled less than 2 days		Sampled greater than 2 days		No Collection Date		Total number of screens
	No.	%	No.	%	No.	%	No.	%	No.
1	2,246	53.6	19	0.5	1,858	44.3	71	1.7	4,194
2	2,633	52.3	40	0.8	2,316	46.0	44	0.9	5,033
3	2,536	53.5	22	0.5	2,124	44.8	59	1.2	4,741
4	2,604	53.1	24	0.5	2,191	44.7	84	1.7	4,903
5	2,955	50.5	45	0.8	2,782	47.5	71	1.2	5,853
6	2,830	49.4	38	0.7	2,790	48.7	74	1.3	5,732
7	3,243	48.9	39	0.6	3,243	48.9	103	1.6	6,628
8	3,675	47.4	44	0.6	3,937	50.7	102	1.3	7,758
9	3,717	44.9	56	0.7	4,395	53.1	105	1.3	8,273
10	3,557	40.2	50	0.6	5,116	57.8	131	1.5	8,854
Not recorded	33	48.5	0	0.0	29	42.6	6	8.8	68
Total	30,029	48.4	377	0.6	30,781	49.6	850	1.4	62,037

Figure 3 identifies the percentage of samples taken at two days by NZDep. No NZ deprivation level that reached the 95% target.

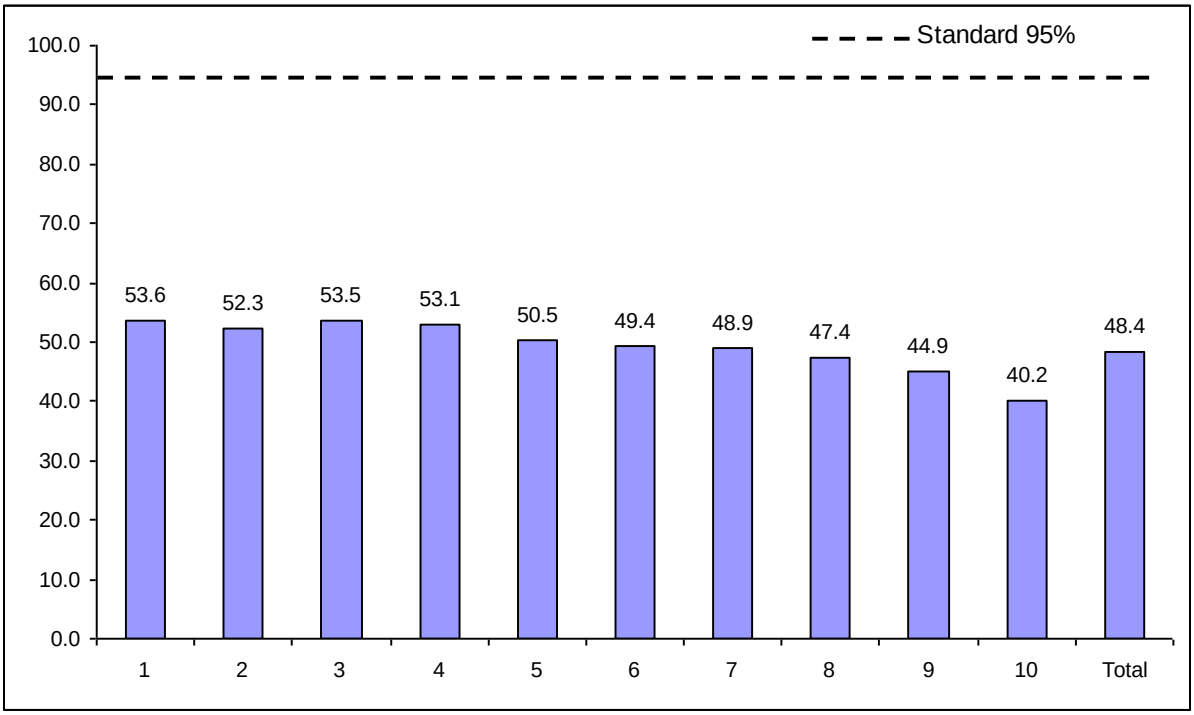

Figure 3 Percentage of samples taken at two days, by NZDep, January to December 2011

4 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 8 shows that 14 DHBs meet or are close to the standard of 99% of blood spot samples suitable for testing. Distribution of high quality lancets to specimen submitters has commenced and this indicator will show a significant improvement in 2012.

Table 8 Percentage of blood samples that meet quality standards by DHB, January to December 2011

DHB region	Satisfactory		Unsatisfactory		Total samples
	No.	%	No.	%	No.
Northland	2,317	98.1	46	1.9	2,363
Waitemata	7,717	98.7	98	1.3	7,815
Auckland	6,250	98.9	67	1.1	6,317
Counties Manukau	8,614	98.9	98	1.1	8,712
Waikato	5,392	98.5	82	1.5	5,474
Lakes	1,568	98.4	25	1.6	1,593
Bay of Plenty	2,910	98.7	37	1.3	2,947
Tairawhiti	701	97.9	15	2.1	716
Taranaki	1,588	98.7	21	1.3	1,609
Hawkes Bay	2,248	98.6	32	1.4	2,280
Whanganui	836	98	17	2	853
MidCentral	2,296	98.8	29	1.2	2,325
Capital & Coast	3,685	98.3	62	1.7	3,747
Hutt Valley	2,047	98.7	28	1.3	2,075
Wairarapa	542	98.7	7	1.3	549
Nelson Marlborough	1,677	99.1	15	0.9	1,692
West Coast	418	98.1	8	1.9	426
Canterbury	6,115	98.8	72	1.2	6,187
South Canterbury	587	98.7	8	1.3	595
Southern	3,644	98.5	54	1.5	3,698
Not recorded	58	90.6	6	9.4	64
Total	**61,210**	**98.7**	**827**	**1.3**	**62,037**

Figure 4 outlines the reasons why samples were unsatisfactory. These included:

- 377 samples were collected too early (before 48 hours of age)
- 350 had a problem with the blood collection (such as insufficient blood, no demographics on the card or the sample was contaminated)
- 98 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)
- 2 families declined testing so an empty card was sent.

Second samples were requested from 827 babies and received from 763 (92.3%). The request was declined by 26 families (3.4%); 2 babies died (0.3%) and the remaining 36 were lost to follow-up.

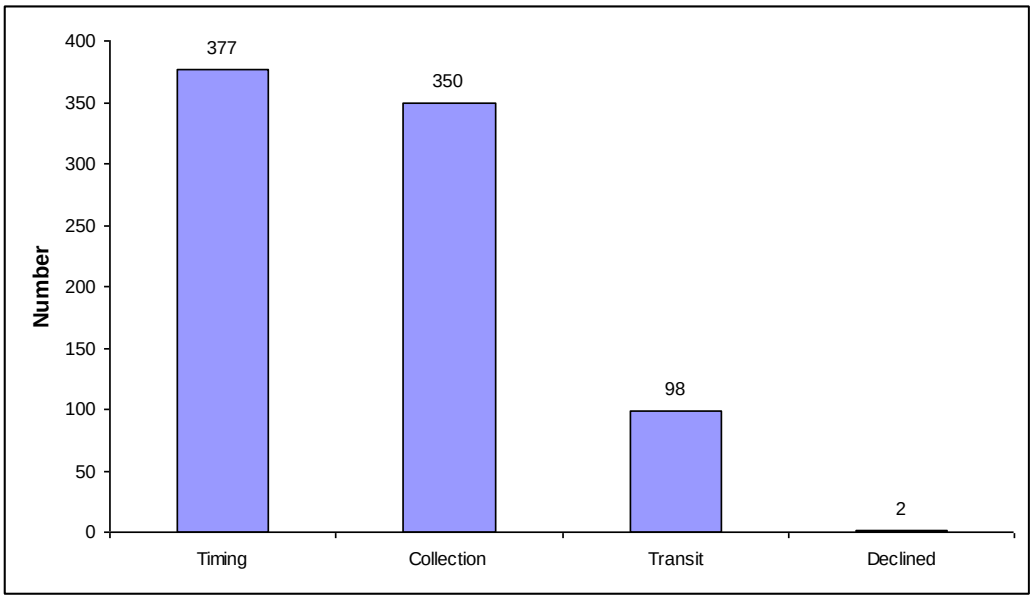

Figure 4 Reasons for unsatisfactory samples

5 Indicator 4: Sample dispatch and delivery

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

Table 9 shows that nationally 65.1% of samples were received with four days, and 33.5% received after four days. 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%.

Table 9 Percentage of samples received by the laboratory within four days by DHB, January to December 2011

DHB region	Less than or equal to 4 days		Greater than 4 days		Unknown		Total samples
	No.	%	No.	%	No.	%	No.
Northland	1,525	*64.5*	796	*33.7*	42	*1.8*	2,363
Waitemata	5,854	*74.9*	1,885	*24.1*	76	*1.0*	7,815
Auckland	4,989	*79.0*	1,241	*19.6*	87	*1.4*	6,317
Counties Manukau	6,230	*71.5*	2,393	*27.5*	89	*1.0*	8,712
Waikato	3,421	*62.5*	1,974	*36.1*	79	*1.4*	5,474
Lakes	1,050	*65.9*	517	*32.5*	26	*1.6*	1,593
Bay of Plenty	1,741	*59.1*	1,142	*38.8*	64	*2.2*	2,947
Tairawhiti	332	*46.4*	370	*51.7*	14	*2.0*	716
Taranaki	1,075	*66.8*	519	*32.3*	15	*0.9*	1,609
Hawkes Bay	1,354	*59.4*	892	*39.1*	34	*1.5*	2,280
Mid Central	523	*61.3*	323	*37.9*	7	*0.8*	853
Whanganui	1,390	*59.8*	896	*38.5*	39	*1.7*	2,325
Capital and Coast	2,471	*65.9*	1,227	*32.7*	49	*1.3*	3,747
Hutt Valley	1,149	*55.3*	897	*43.2*	30	*1.4*	2,076
Wairarapa	335	*61.0*	207	*37.7*	7	*1.3*	549
Nelson Marlborough	924	*54.6*	746	*44.1*	22	*1.3*	1,692
West Coast	208	*48.8*	211	*49.5*	7	*1.6*	426
Canterbury	3,562	*57.6*	2,523	*40.8*	103	*1.7*	6,188
South Canterbury	316	*53.2*	276	*46.5*	2	*0.3*	594
Southern	1,924	*52.0*	1,722	*46.6*	52	*1.4*	3,698
Not recorded	44	*69.8*	16	*25.4*	3	*4.8*	63
Total	40,417	*65.1*	20,773	*33.5*	847	*1.4*	62,037

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

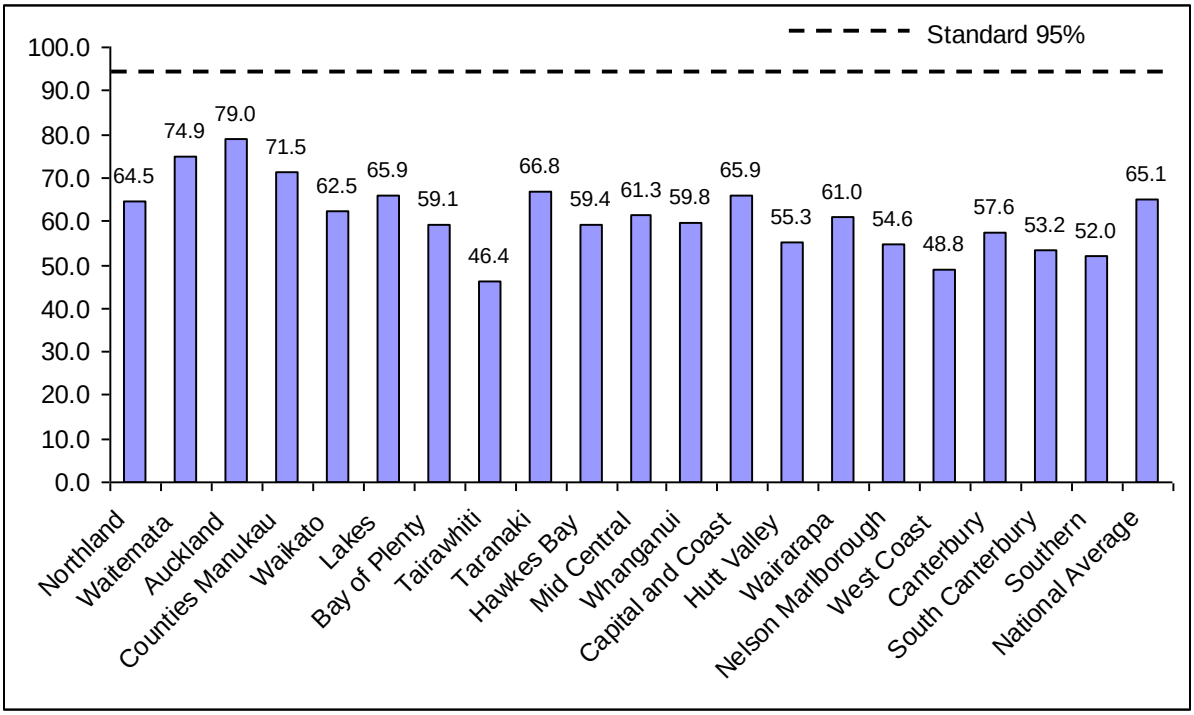

Figure 5 Percentage of samples received by the laboratory in four days or less, January to December 2011

6 Indicator 5: Laboratory testing timeframes

Table 10 identifies the percentage of samples that met the specified laboratory testing standard. The standard requires that 100% of samples met the specified laboratory turnaround times. 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 10 Percentage of results available within specified timeframes, by disorder, January to December 2011 (n= 62,037 samples)

Disorder	Standard for turnaround time (days)	Number met timeframe	% met timeframe
Congenital Adrenal Hyperplasia	2	61,842	99.7
Galactosaemia	2	61,874	99.7
Amino acid disorders	2	61,337	98.9
Fatty acid oxidation disorders	2	61,337	98.9
Biotinidase deficiency	5	61,928	99.8
Cystic fibrosis	5	61,222	98.7
Congenital hypothyroidism	5	61,927	99.8

7 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 11. 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 11 Percentage of positive test results reported within specified timeframes, by disorder, January to December 2011

Reason for report	Standard: Calendar days (from receipt in laboratory to report)	Number of positive test reports	Number met timeframe	% met timeframe
Amino acid & fatty acid oxidation disorders	3	392	265	67.6
CAH	3	111	74	66.7
Galactosaemia	3	6	5	83.3
CH	3	49	44	89.8
Biotinidase deficiency	4	7	7	100.0
Cystic fibrosis	9	62	30	48.4

Table 12 outlines the percentage of urgent clinical critical positive reports reporting by timeframes and disorder. 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.

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 12 Percentage of urgent clinical critical positive results reported within specified timeframes, by disorder, January to December 2011

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 and fatty acid oxidation disorders	3	36	35	97.8
CAH	3	2	2	100
Galactosaemia	3	0	0	0
CH	4	25	25	100
Biotinidase deficiency	9	0	0	0
Cystic fibrosis	12	0	0	0

8 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 13 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 36.8%. Improvements have been made by the NMSP working closely with lead maternity carers (LMCs) to ensure follow-up is completed before the baby is discharged from midwifery care at 4-6 weeks of age.

Table 13 Follow-up of requested second samples by DHB, January to December 2011

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	18	27.7	44	67.7	3	4.6	62	95.4	65
Waitemata	64	41.8	87	56.9	2	1.3	151	98.7	153
Auckland	64	55.2	48	41.4	4	3.4	112	96.6	116
Counties Manukau	83	41.1	113	55.9	6	3.0	196	97.0	202
Waikato	31	25.2	89	72.4	3	2.4	120	97.6	123
Lakes	16	42.1	21	55.3	1	2.6	37	97.4	38
Bay of Plenty	19	35.2	34	63.0	1	1.9	53	98.1	54
Tairawhiti	7	31.8	15	68.2	0	0.0	22	100.0	22
Taranaki	13	48.1	14	51.9	0	0.0	27	100.0	27
Hawkes Bay	9	19.6	35	76.1	2	4.3	44	95.7	46
Whanganui	10	47.6	11	52.4	0	0.0	21	100.0	21
Mid Central	18	36.7	28	57.1	3	6.1	46	93.9	49
Hutt Valley	13	34.2	25	65.8	0	0.0	38	100.0	38
Capital and Coast	25	33.8	46	62.2	3	4.1	71	95.9	74
Wairarapa	3	27.3	8	72.7	0	0.0	11	100.0	11
Nelson Marlborough	8	34.8	15	65.2	0	0.0	23	100.0	23
West Coast	3	25.0	9	75.0	0	0.0	12	100.0	12
Canterbury	30	26.1	84	73.0	1	0.9	114	99.1	115
South Canterbury	4	40.0	6	60.0	0	0.0	10	100.0	10
Southern	30	39.0	45	58.4	2	2.6	75	97.4	77
Not recorded	3	100.0	0	0.0	0	0.0	3	100.0	3
Total	**471**	**36.8**	777	**60.8**	**31**	**2.4**	**1,248**	**97.6**	**1,279**

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.

9 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 are 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 14.

Table 14 Age at treatment, January to December 2011

Disorder	Standard: Calendar days of age of baby at treatment commenced	Number of cases	Number treated by specified age
Biotinidase deficiency	14	1	0
Cystic fibrosis	28	14	4
CH	10	26	15
CAH	10	1	0
Galactosaemia	10	-	-
Amino acid disorders	10	9	4
Fatty acid oxidation disorders	10	13	8

The one baby with biotinidase deficiency was notified from a second sample at 19 days.

Of the babies diagnosed with cystic fibrosis, three were treated just outside the timeframe at 29 days. The delays in the remainder were in the time to diagnosis and treatment.

There were 5 cases of CH diagnosed outside the timeframe as their initial levels of TSH were between 15 and 50 mIU/L and notification was made following the results of a second sample. Of the remaining six, four had delayed transit times (5-10 days) and two delayed diagnosis and treatment (10 and 16 days after notification respectively).

The one case of CAH had treatment commenced just outside the timeframe at 11 days. The sample was received when baby was 10 days old (taken four days, transit six days) and treatment commenced at 11 days.

The five patients with amino acid breakdown disorders not treated by the specified age had mild disease diagnosed following a second blood sample.

Four of the five patients with fatty acid oxidation disorders not treated by the specified age had mild disease diagnosed following a second blood sample. The remaining case had delays in specimen collection (four days) and transit to the laboratory (seven days).

Some of the amino acid breakdown disorders and fatty acid oxidation disorders do not require urgent treatment (for example PKU). In all cases where a delay of diagnosis could have clinical consequences there was metabolic physician consultation with the LMC and a subsequent decision made by the metabolic physician about the urgency of follow-up, hence some patients diagnosed outside the timeframe on a second sample had earlier metabolic physician involvement.

10 Indicator 9: Blood spot card storage and return

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

Of 643 requests for blood spot returns, 633 (98.4%) were returned in the timeframe.
Two cards were not returned as they had insufficient information. This was requested but not provided. The delay in the other eight samples was due to a variety of reasons including:

- a delay retrieving card from storage
- an unclear information request
- a delay linking a second sample with a return request on the first sample.

In general samples are returned with a median time over this period of 1.3 days.

The NMSP Policy framework lists possible secondary uses for residual screening cards (section 4.1(b)). Table 15 outlines blood spots used for secondary purposes. This shows that of the 34 cards used for the benefit of the individual and family/whanau, 18 were for cytomegalovirus (CMV) testing to determine whether congenital CMV was the cause of symptoms in the baby or child. Sixteen were used for genetic studies, some related to newborn screening (e.g. expanded screening on a child born pre-screening) or testing of siblings of diagnosed cases.

Table 15 Reasons for secondary use, January to December 2011

Secondary use	Number
Benefit of the individual and family/whanau	34
Forensic/police/coroner investigations	12
Mortality review	0
Research	0
Other	0
Total	**46**

11 Screening performance and incidence

11.1 Screening performance

The screening performance from 2007 to 2011 for the NMSP is provided in Table 16. For this timeframe, 316,436 infants were screened. A longer time period is used to calculate this as these conditions are rare and it is important to have sufficient numbers for comparison, as this adds power to the data.

The table shows that during 2007 to 2011:

- screening sensitivities ranged from 94.78 to 100%
- screening specificities ranged from 99.64 to 99.99%
- positive predictive values ranged from 2.65 to 50.94.

Table 16 Newborn screening performance, 2007-2011

Condition	Sensitivity %	Specificity %	Positive Predictive Value %
Aminoacid breakdown disorders	100.00	99.64	3.30
Biotinidase deficiency	100.00	99.99	14.29
Congenital adrenal hyperplasia	100.00	99.77	0.69
Congenital hypothyroidism	94.78	99.96	50.94
Cystic fibrosis	97.01	99.92	22.22
Fatty acid oxidation disorders	100.00	99.84	11.50
Galactosaemia	100.00	99.99	2.65

11.2 Follow-up of positive tests

Follow-up of positive tests are detailed in Table 17. Appropriate follow-up may be:

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

Overall 1464 babies were referred for paediatric examination or had a request for a second sample (44% for disorders, 56% unsuitable samples).

Table 17 Newborn screening follow-up by condition, 2011

Condition	Number of positive tests	Number with appropriate follow-up	% with appropriate follow-up
Aminoacid breakdown disorders	301	295	98
Biotinidase deficiency	7	7	100
Congenital adrenal hyperplasia	111	111	100
Congenital hypothyroidism	49	49	100
Cystic fibrosis	62	62	100
Fatty acid oxidation disorders	91	89	98
Galactosaemia	6	3	50

11.3 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. Details are provided in Table 18.

Table 18 Clinical Utility of screening, 2011

Disorder	Number of cases	Diagnosis suspected before screen result	Reason for suspicion
Biotinidase deficiency	1	0	
Cystic fibrosis	14	5	1 FH, 3 MI, 1FTT
CH	26	0	
CAH	1	0	
Galactosaemia	0	0	
Amino acid disorders	9	1	HCS symptomatic
Fatty acid oxidation disorders	13	2	FH
Total conditions	**64**	**8**	

Reasons for clinical detection are:

- family history (FH)
- meconium ileus (MI)
- failure to thrive (FTT).

One baby with holocarboxylase synthase deficiency (HCS) was diagnosed before the screening test result following early symptomatic presentation. Overall 64 cases of screened disorders were detected by the programme and in 56 of these there was no clinical suspicion of the disorder before the screening test result.

11.4 Incidence of screened disorders

Aminoacid breakdown disorders

Since screening started in 2006, 385,238 infants have been screened and 43 cases detected, giving an incidence of 1:8,959. This includes PKU, MSUD and hyperphenylalaninemia.

PKU
There were four cases of PKU and one of hyperphenylalaninemia found in 2011. Since screening started in 1969, 2,463,777 infants have been screened for PKU and 116 found to give an incidence of 1:21,200. Benign hyperphenylalaninemia is not counted in this figure. It is problematic comparing PKU incidence as the definition of the disorder is a 'a level of phenylalanine that requires treatment' and the level has varied time to time.

MSUD
There were no new cases in 2011. Since screening started in 1969, 2,462,777 infants have been screened for MSUD, 9 classical cases found to given an incidence of 1:274,000.

Biotinidase deficiency

Since screening started in 1986, 1,530,513 infants have been screened and 8 cases detected giving an incidence of 1:191,314.

Congenital adrenal hyperplasia

Since screening started in 1986, 1,577,462 infants have been screened and 67 cases detected giving an incidence of 1:23,544. Most false positive results are in small babies and most are due another test scheduled according to the NICU protocol.

Congenital hypothyroidism

Since screening started in 1981, 1,798,042 infants have been screened and 467 cases detected giving an incidence of 1:3850. 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, 2,513,094 infants have been screened and 367 cases detected giving an incidence of 1:6,847. There were 61,859 babies screened in 2011 (of whom approximately 55% were of European ethnicity) and 14 cases of CF detected. This gives an incidence in the European births of 1:2,800.

Fatty acid oxidation disorders

Since screening started in 2006, 385,238 infants have been screened and 41 cases detected giving an incidence of 1:.9,396.

Galactosaemia

Since screening started in 1973, 2,351,902 infants have been screened and 24 cases detected giving an incidence of 1:97,995.

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 BLOOD SAMPLES

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 DESPATCH 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: LABORATORY 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 RECEIPT 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.