MINISTRY OF HEALTH
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Newborn Metabolic Screening Programme

Annual Report

January to December 2014

National Screening Unit

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Contents

Abbreviations

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 Newborn Metabolic Screening Programme (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 2014

- 58,673 babies were tested and 59,097 born, giving a screening coverage of 99%.
- 74% of samples were collected within the 48-72 hour timeframe. No DHB and no ethnic group met the standard of 95% within the timeframe. Babies of lower NZDep and European ethnicity are more likely to have the sample collected within the appropriate timeframe.
- Overall 98% of samples were suitable for testing. One DHB met the standard of 99% of samples suitable. The main reasons samples were unsuitable were 'taken too early' or 'not sufficient/contaminated sample card'. Follow-up of unsuitable samples was 92%.
- 66% of samples were received by the laboratory in four days or less. No DHB met the standard of 95% within four days.
- The laboratory testing timeframes standard of 100% was met for galactosemia and biotinidase deficiency screening and was 99% for the other disorders.
- Of 33 clinical critical results, 31 were notified within the timeframe, the remaining two were two days late.
- Although only 38% of second samples were received in ten days or less (the standard is 100%), 97% of follow-up was completed appropriately.
- Treatment was commenced by the specified age for 40% of babies with diagnosed disorders, for however most of the remainder were just outside the range, except for one patient with PKU treated at 25 days and one with MCAD at 50 days (both due to delayed transit time of the sample to the laboratory). There were no known clinical ill-effects from the delays.
- 99% of 666 requests for card returns were made in 28 days or less (the standard is 100%).
- 76 cards were used for additional testing for 'family health reasons', mostly for CMV testing.
- 150 cards were used for research in one study.
- Follow-up of positive tests was between 94-100%. All clinical critical results had appropriate follow-up.
- 60 cases of screened disorders were detected by the programme and in 52 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 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. Reports are published on the National Screening Unit (NSU) website. This is the fourth 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:
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 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. Post-paid envelopes and lancets are now supplied to LMCs. Parent information sheets about congenital hypothyroidism, cystic fibrosis and MCAD and a general sheet about autosomal recessive inheritance have been developed for use in the diagnostic process.

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 Technical Group reviews monitoring reports and makes recommendations.

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 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

Screening 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 2014 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 based on the prioritised 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.

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.

National monitoring indicators

Table 1 summarises the NMSP indicators used for regular monitoring with their reporting frequency and detail included in Appendix 2. 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 Technical Group.

Table 1 NMSP indicators

Indicators	Detail
1. Newborn Metabolic Screening Coverage	• DHB • Ethnicity • Deprivation status
2. Timing of sample taking	• DHB • Ethnicity • Deprivation status
Laboratory reporting	
3. Quality of Blood Samples	• DHB
4. Sample dispatch and delivery	• DHB
5. Laboratory testing timeframes	
6. Timeliness of reporting - notification of screen positives	
7. Collection and receipt of second samples	• DHB
Incidence	
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	
9. Blood spot card storage and return	

Indicator 1: Screening coverage

Overall samples were received from 58,673 newborns between January and December 2014. 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 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 59,097 babies were born in 2014. 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. Approximately 99% of babies were screened.

Denominator data is sourced from the National Maternity Collection.

Table 2 outlines the numbers of babies' screened and annual coverage from 2007 to 2014.

Table 2 Number of babies screened and coverage 2007 – 2013

Year	Births	Babies screened	Coverage %
2007	64,040	65,121	97.7
2008	65,333	63,794	97.6
2009	63,285	63,516	100.4
2010	64,699	63,727	98.5
2011	62,733	61,859	98.6
2012	62,842	61,422	97.7
2013	59,707	59,192	99.1
2014	59,097	58,673	99.3

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. As noted in previous reports there is no reliable screening denominator.

Table 3 Number of babies screened by DHB, January to December 2014

DHB Region	Births	Babies screened	Coverage %
Northland	2,097	2,146	102.3
Waitemata	7,910	7,740	97.9
Auckland	6,370	6,148	96.5
Counties Manukau	8,279	8,096	97.8
Waikato	5,325	5,274	99.0
Lakes	1,393	1,394	100.1
Bay of Plenty	2,785	2,809	100.9
Tairawhiti	722	695	96.3
Taranaki	1,529	1,548	101.2
Hawkes Bay	2,098	2,123	101.2
Whanganui	822	811	98.7
Mid Central	2,112	2,090	99.0
Hutt Valley	1,863	1,859	99.8
Capital and Coast	3,572	3,418	95.7
Wairarapa	443	472	106.5
Nelson Marlborough	1,438	1,468	102.1
West Coast	353	369	104.5
Canterbury	6,032	5,933	98.4
South Canterbury	658	649	98.6
Southern	3,296	3,277	99.4
Not recorded*		354	
Total	**59,097**	**58,673**	**99.3**

*does not include babies born to mothers who usually reside overseas

Indicator 2: Timing of sample taking

This indicator is monitored by the number of screens performed. In 2014, 58,747 babies had a screen and 58,944 screens were done. This includes 197 babies who 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 74.1% of samples were collected at 48 to 72 hours, and 21.9% at greater than 72 hours.

Table 4 Timing of sample taking by DHB, January to December 2014

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	1,447	67.1	13	0.6	621	28.8	74	3.4	2,155
Waitemata	5,938	76.5	61	0.8	1,601	20.6	158	2.0	7,758
Auckland	5,093	82.5	53	0.9	810	13.1	217	3.5	6,173
Counties Manukau	5,247	64.6	54	0.7	2,382	29.3	437	5.4	8,120
Waikato	3,174	60.0	47	0.9	1,885	35.6	183	3.5	5,289
Lakes	981	70.2	14	1.0	357	25.5	46	3.3	1,398
Bay of Plenty	1,740	61.8	17	0.6	968	34.4	92	3.3	2,817
Tairawhiti	532	76.2	1	0.1	147	21.1	18	2.6	698
Taranaki	1,241	80.0	10	0.6	259	16.7	42	2.7	1,552
Hawkes Bay	1,681	78.7	11	0.5	401	18.8	42	2.0	2,135
Whanganui	563	69.2	8	1.0	221	27.2	21	2.6	813
Mid Central	1,607	76.6	14	0.7	407	19.4	70	3.3	2,098
Hutt Valley	1,280	68.4	5	0.3	535	28.6	51	2.7	1,871
Capital and Coast	2,731	79.5	17	0.5	602	17.5	85	2.5	3,435
Wairarapa	351	74.2	4	0.8	105	22.2	13	2.7	473
Nelson Marlborough	1,192	80.8	14	0.9	227	15.4	42	2.8	1,475
West Coast	307	83.2	3	0.8	45	12.2	14	3.8	369
Canterbury	5,271	88.6	34	0.6	494	8.3	149	2.5	5,948
South Canterbury	534	82.2	3	0.5	101	15.5	12	1.8	650
Southern	2,509	76.4	9	0.3	667	20.3	101	3.1	3,286
Not recorded	269	75.4	7	2.0	63	17.6	18	5.0	357
Total	43,688	74.2	399	0.7	12,898	21.9	1,885	3.2	58,870*

*Total includes 197 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 2014 no DHB met the 95% standard.

Figure 1 Percentage of samples taken at 48 to 72 hours, by DHB, January to December 2014

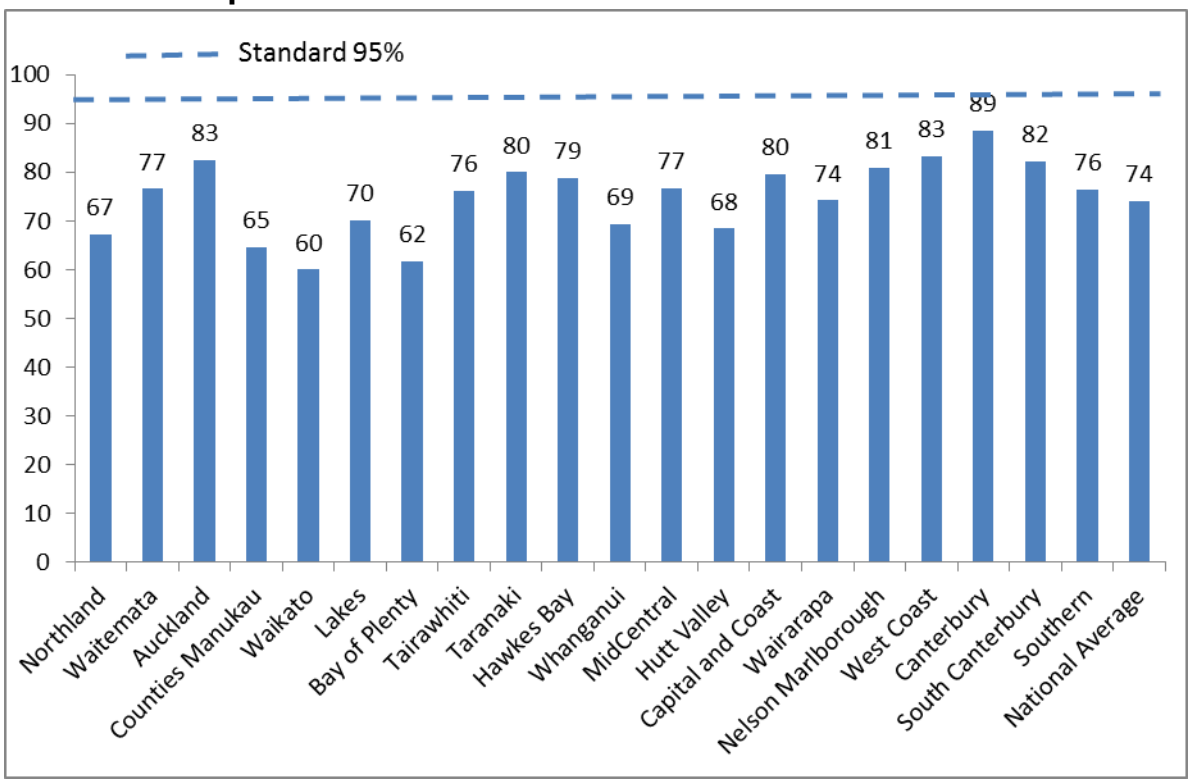


Table 5 shows detailed information by ethnicity. Māori (68%) and Pacific (66%) babies are less likely than European (77%) and Asian (78%) babies to have a sample collected at two days. It is noted that the 95% standard was not met by any ethnic group.

Table 5 Timing of sample taking by Group 1 and Group 2 ethnicity, January to December 2014

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	8,580	68.3	93	0.7	3,470	27.6	428	3.4	12,571
Pacific	3,933	65.9	48	0.8	1,725	28.9	262	4.4	5,968
Cook Island Māori	617	66.1	2	0.2	271	29.0	44	4.7	934
Fijian	322	68.4	0	0.0	128	27.2	21	4.5	471
Niuean	212	64.8	4	1.2	99	30.3	12	3.7	327
Samoan	1,577	65.7	25	1.0	698	29.1	100	4.2	2,400
Tokelauan	77	70.0	1	0.9	27	24.5	5	4.5	110
Tongan	919	65.2	13	0.9	415	29.4	63	4.5	1,410
Other Pacific	209	66.1	3	0.9	87	27.5	17	5.4	316
Asian	7,262	77.9	66	0.7	1,683	18.1	306	3.3	9,317
Chinese	2,925	82.0	17	0.5	540	15.1	83	2.3	3,565
Indian	2,079	73.3	27	1.0	604	21.3	128	4.5	2,838
Southeast Asian	827	77.8	8	0.8	199	18.7	29	2.7	1,063
Other Asian	1,431	77.3	14	0.8	340	18.4	66	3.6	1,851
European	23,106	77.2	182	0.6	5,793	19.4	854	2.9	29,935
NZ European	19,682	77.1	153	0.6	4,958	19.4	743	2.9	25,536
Latin American / Hispanic	265	78.9	2	0.6	62	18.5	7	2.1	336
Other European	3,159	77.8	27	0.7	773	19.0	104	2.6	4,063
Other	808	70.1	10	0.9	227	19.7	108	9.4	1,153
African	272	76.8	3	0.8	72	20.3	7	2.0	354
Middle Eastern	333	73.5	4	0.9	104	23.0	12	2.6	453
Other/not known	202	74.3	3	1.1	51	29.8	16	5.9	272
Total	43,688	74.2	399	0.7	12,898	21.9	1,885	3.2	58,870*

*Includes 197 babies who had more than one first screen

Figure 2 outlines the percentage of samples taken at 48 to 72 hours by ethnicity. In 2014 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 2014

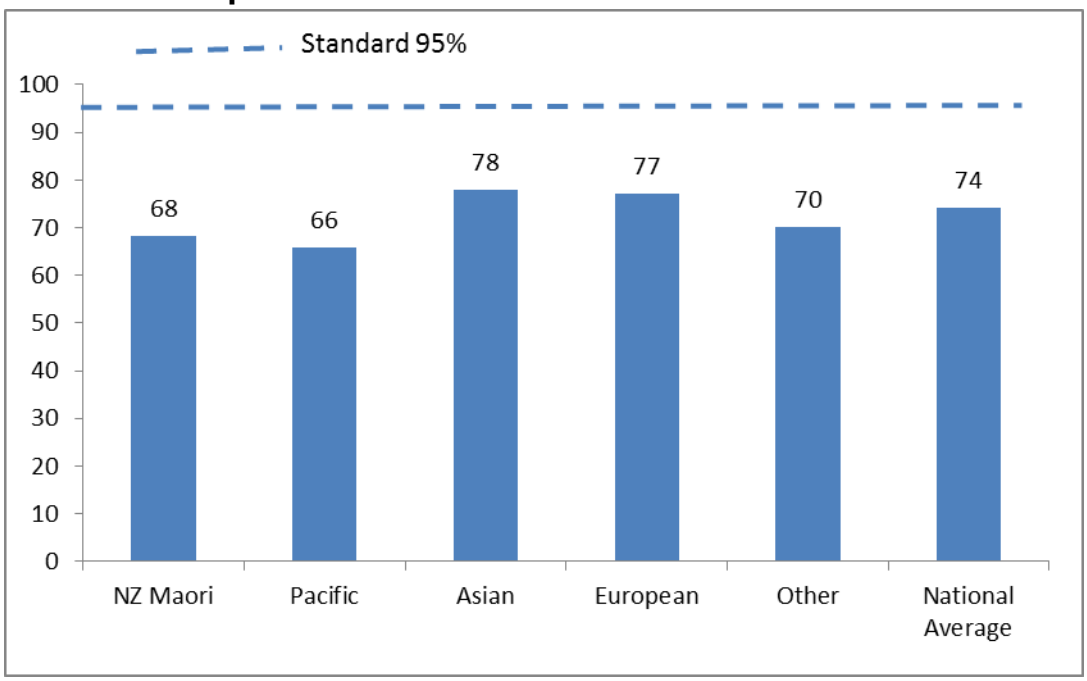


Table 6 shows the number of samples taken at two days by NZDep. The data indicates a slightly lower percentage of samples taken by the recommended time for babies in the five groups with the highest levels of deprivation (NZDep 6 to 10).

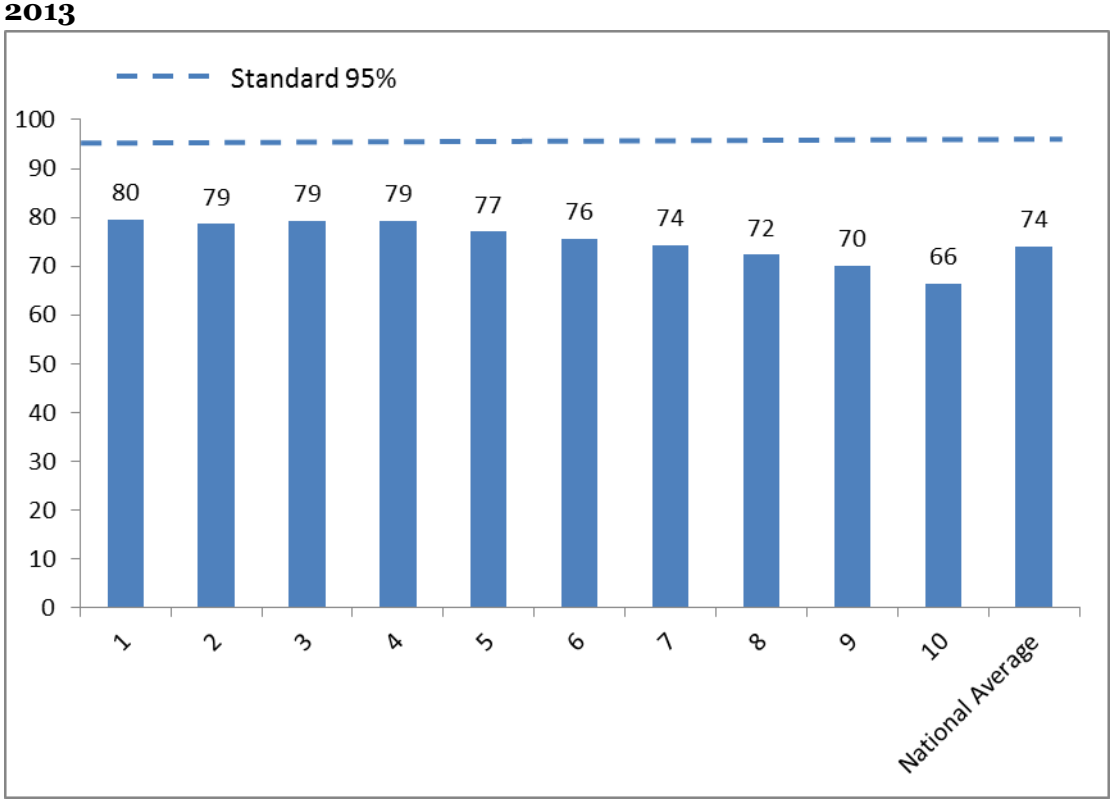
Table 6 Percentage of samples taken at 48 to 72 hours by NZDep, January to December 2014

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	3,131	79.6	19	0.5	690	17.5	92	2.3	3,932
2	3,718	78.6	30	0.6	836	17.7	146	3.1	4,730
3	3,689	79.2	28	0.6	833	17.9	109	2.3	4,659
4	3,562	79.2	35	0.8	769	17.1	131	2.9	4,497
5	4,110	77.1	31	0.6	1,035	19.4	158	3.0	5,334
6	4,266	75.6	39	0.7	1,177	20.9	159	2.8	5,641
7	4,713	74.2	45	0.7	1,402	22.1	193	3.0	6,353
8	5,341	72.3	45	0.6	1,757	23.8	242	3.3	7,385
9	5,379	70.0	55	0.7	1,945	25.3	300	3.9	7,679
10	5,508	66.4	65	0.8	2,391	28.8	337	4.1	8,301
Not recorded	271	75.5	7	1.9	63	17.5	18	5.0	359
Total	**43,688**	**74.2**	**399**	**0.7**	**12,898**	**21.9**	**1,885**	**3.2**	**58,870***

*Includes 197 babies who had more than one first screen

Figure 3 identifies the percentage of samples taken at 48 to 72 hours by NZDep. In 2014 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 only one DHB 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 2014

DHB region	Satisfactory		Unsatisfactory		Total samples
	No.	%	No.	%	No.
Northland	2,115	98.1	40	1.9	2,155
Waitemata	7,619	98.2	139	1.8	7,758
Auckland	6,090	98.7	83	1.3	6,173
Counties Manukau	7,934	97.7	186	2.3	8,120
Waikato	5,211	98.5	78	1.5	5,289
Lakes	1,361	97.4	37	2.6	1,398
Bay of Plenty	2,782	98.8	35	1.2	2,817
Tairawhiti	686	98.3	12	1.7	698
Taranaki	1,528	98.5	24	1.5	1,552
Hawkes Bay	2,098	98.3	37	1.7	2,135
Whanganui	796	97.9	17	2.1	813
Mid Central	2,054	97.9	44	2.1	2,098
Capital & Coast	1,837	98.2	34	1.8	1,871
Hutt Valley	3,371	98.1	64	1.9	3,435
Wairarapa	470	99.4	3	0.6	473
Nelson Marlborough	1,452	98.4	23	1.6	1,475
West Coast	365	98.9	4	1.1	369
Canterbury	5,878	98.8	70	1.2	5,948
South Canterbury	643	98.9	7	1.1	650
Southern	3,238	98.5	48	1.5	3,286
Not recorded	83	96.5	3	3.5	86
Total	57,875	98.3	995	1.7	58,870

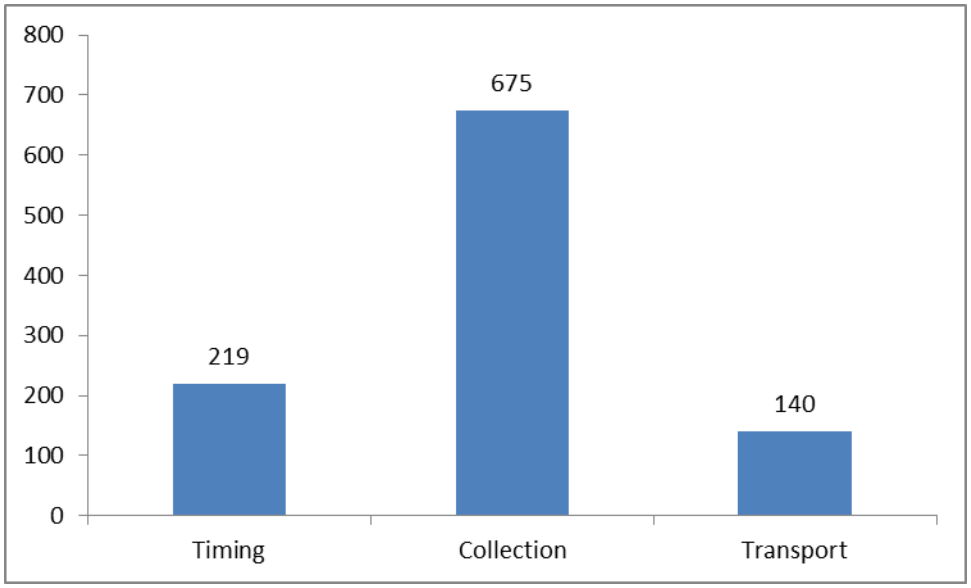
*197 babies have two first samples.

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

- 219 samples were collected too early (before 48 hours of age)
- 675 had a problem with the blood collection (such as insufficient blood, no demographics on the card or the sample was contaminated)
- 140 either took longer than one month, flap folded onto wet blood causing significant loss onto the flap, were damaged in transit or put wet into a plastic bag.

Second samples were requested from 1034 babies and received from 950 (91.9%). The request was declined by 42 families (4.1%); nine babies died (0.9%) and the remaining 33 (3.2%) were lost to follow-up.

Figure 4 Reasons for unsatisfactory samples, January to December 2014



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.

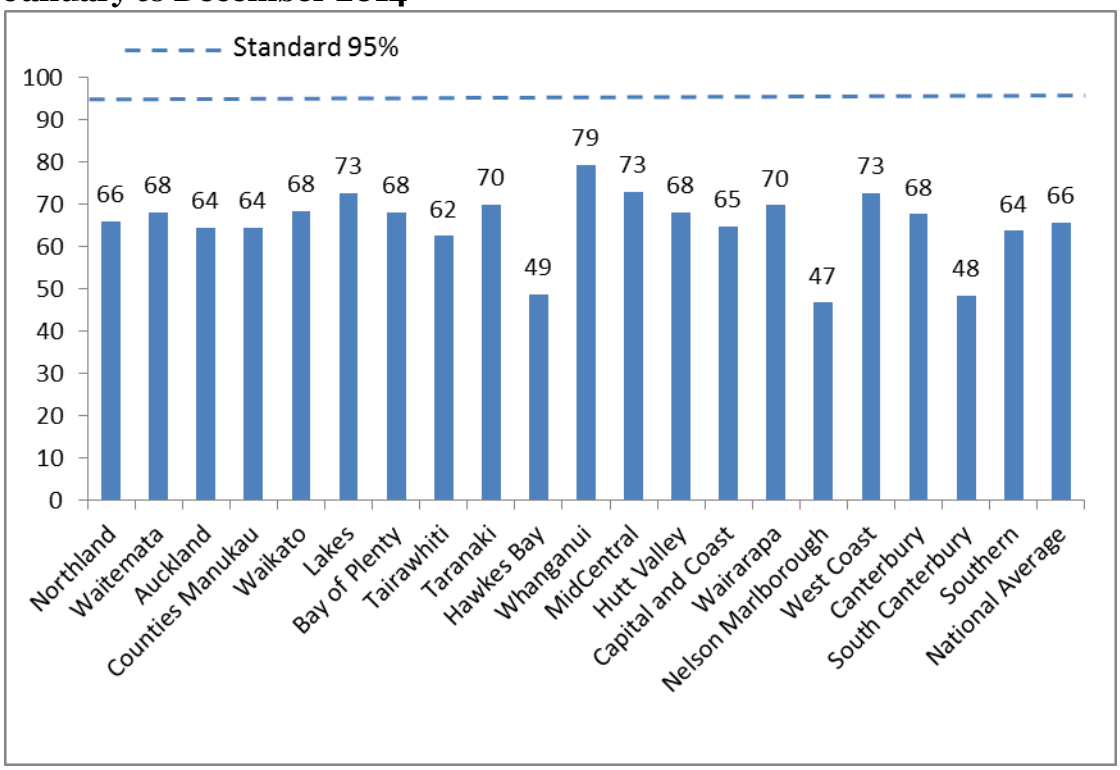
Table 8 shows that nationally 65.6% of samples were received within four days, and 34.4% received after four days. The range received in four days or less was 46.7 to 79.2%. Although postage paid envelopes have been supplied to specimen submitters and this greatly improved transit times across all DHBs, no DHB met the standard of 95%. Overall, fewer samples were received in time in 2014 compared to 2013 and this may be associated with a reduction in box clearance times by NZ Post.

Table 8 Percentage of samples received by the laboratory within four days by DHB, January to December 2014

DHB region	Less than or equal to 4 days		Greater than 4 days		Unknown		Total samples
	No.	%	No.	%	No.	%	No.
Northland	1,422	66.0	732	34.0	31	1.4	2,154
Waitemata	5,277	68.1	2,477	31.9	61	0.8	7,754
Auckland	3,965	64.3	2,200	35.7	74	1.2	6,165
Counties Manukau	5,215	64.3	2,894	35.7	133	1.6	8,109
Waikato	3,612	68.4	1,665	31.6	66	1.3	5,277
Lakes	1,012	72.5	383	27.5	26	1.9	1,395
Bay of Plenty	1,916	68.1	898	31.9	40	1.4	2,814
Tairawhiti	436	62.5	262	37.5	8	1.1	698
Taranaki	1,083	69.9	466	30.1	18	1.2	1,549
Hawkes Bay	1,037	48.7	1,094	51.3	19	0.9	2,131
Mid Central	644	79.2	169	20.8	12	1.5	813
Whanganui	1,524	72.9	567	27.1	37	1.8	2,091
Capital and Coast	1,273	68.0	598	32.0	24	1.3	1,871
Hutt Valley	2,216	64.6	1,213	35.4	32	0.9	3,429
Wairarapa	329	69.7	143	30.3	4	0.8	472
Nelson Marlborough	689	46.7	786	53.3	14	0.9	1,475
West Coast	268	72.6	101	27.4	3	0.8	369
Canterbury	4,013	67.6	1,925	32.4	77	1.3	5,938
South Canterbury	314	48.3	336	51.7	3	0.5	650
Southern	2,091	63.7	1,193	36.3	53	1.6	3,284
Not recorded	248	59.0	172	41.0	5	1.2	420
Total	38,584	65.6	20,274	34.4	740	1.3	58,858

Figure 5 shows 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 2014



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 98.7% to 100%. 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 2014

Disorder	Standard for turnaround time (days)	Number met standard	% met standard
Congenital adrenal hyperplasia	2	58,516	99.3
Galactosaemia	2	58,927	100
Amino acid disorders	2	58,198	98.7
Fatty acid oxidation disorders	2	58,198	98.7
Biotinidase deficiency	5	58,944	100
Cystic fibrosis	5	58,344	99.0
Congenital hypothyroidism	5	58,547	99.3

(n= 58,870 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 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 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 2014

Reason for report	Standard: Calendar days (from receipt in laboratory to report)	Number of positive test reports	Number met timeframe	% met timeframe
Amino acid disorders	3	205	113	55.1
Fatty acid oxidation disorders	3	99	57	57.6
Congenital adrenal hyperplasia	3	50	39	78.0
Galactosaemia	3	0	0	N/A
Congenital hypothyroidism	4	58	48	82.8
Biotinidase deficiency	9	0	0	N/A
Cystic fibrosis	12	57	30	52.6

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
- one positive cystic fibrosis test was notified after 36 days due to a laboratory error.

Table 11 outlines the percentage of urgent clinical critical positive results reported 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 2014

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	4	4	100
Fatty acid oxidation disorders	3	8	7	88*
Congenital adrenal hyperplasia	3	4	4	100
Galactosaemia	3	0	0	N/A
Congenital hypothyroidism	4	17	16	94*
Biotinidase deficiency	9	0	0	N/A
Cystic fibrosis	12	0	0	N/A

*Outlier at 5d (sample received 24/12/14). Outlier at 5d.

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 38.2%. Improvements compared to 2013 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 2014

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	32.1	36	64.3	2	3.6	54	96.4	56
Waitemata	88	48.4	92	50.5	2	1.1	180	98.9	182
Auckland	57	49.1	57	49.1	2	1.7	114	98.3	116
Counties Manukau	80	31.9	162	64.5	9	3.6	242	96.4	251
Waikato	44	38.3	70	60.9	1	0.9	114	99.1	115
Lakes	19	42.2	22	48.9	4	8.9	41	91.1	45
Bay of Plenty	22	43.1	27	52.9	2	3.9	49	96.1	51
Tairawhiti	5	27.8	13	72.2	0	0.0	18	100.0	18
Taranaki	12	34.3	22	62.9	1	2.9	34	97.1	35
Hawkes Bay	17	37.0	28	60.9	1	2.2	45	97.8	46
Whanganui	14	60.9	9	39.1	0	0.0	23	100.0	23
Mid Central	22	37.3	37	62.7	0	0.0	59	100.0	59
Hutt Valley	16	35.6	29	64.4	0	0.0	45	100.0	45
Capital and Coast	30	40.0	39	52.0	6	8.0	69	92.0	75
Wairarapa	1	14.3	6	85.7	0	0.0	7	100.0	7
Nelson Marlborough	10	31.3	22	68.8	0	0.0	32	100.0	32
West Coast	0	0.0	4	100.0	0	0.0	4	100.0	4
Canterbury	34	32.1	67	63.2	5	4.7	101	95.3	106
South Canterbury	1	12.5	7	87.5	0	0.0	8	100.0	8
Southern	23	37.1	39	62.9	0	0.0	62	100.0	62
Not recorded	4	25.0	12	75.0	0	0.0	16	100.0	16
Total	**517**	**38.2**	**800**	**59.2**	**35**	**2.6**	**1317**	**97.4**	**1352**

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 treatment commenced 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 2014

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	16	2
Congenital hypothyroidism	10	31	12
Congenital adrenal hyperplasia	10	2	2
Galactosaemia	10	0	N/A
Amino acid disorders	10	2	0
Fatty acid oxidation disorders	10	9	8

Of the 16 babies diagnosed with cystic fibrosis, the NMSP has data on 12. Two were treated before 28 days, the others at 29-63 days. Screening had been declined for one baby and a test was ordered by the paediatrician investigating the baby's failure to thrive. The test was positive and the baby treated at 160 days.

There were 10 cases of congenital hypothyroidism 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. No data is available for five. The remaining four were treated at 11, 14, 15 and 16 days.

The two patients with amino acid breakdown disorders both have PKU. One was treated at 11 days the other at 25 days (the sample took 20 days in transit).

One patient with MCAD treated at 50 days (sample took 48 days in transit).

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 671 requests for blood spot returns, 664 were returned within the timeframe. Five cards were not returned as they had insufficient information. This was requested but not provided. The remaining two samples were returned in 29 and 42 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 1 day.

The NMSP Policy Framework 2011 lists possible secondary uses for residual screening samples (section 4.1(b)). Table 14 shows that 76 cards were used for the benefit of the individual and family/whanau. 67 of these were used to determine whether congenital cytomegalovirus was the cause of symptoms in the baby or child.

The research study was to determine whether newborn screening for the fatty acid oxidation disorder VLCAD had missed any cases since the cut-off used in New Zealand is high relative to that in some other screening programmes; and whether these results could be due to a benign common mutation among some Pacific populations. Results are expected in 2015.

Table 14 Reasons for secondary use of blood spot samples, January to December 2014

Secondary use	Number
Benefit of the individual and family/whanau	76
Forensic/police/coroner investigations	0
Mortality review	0
Research	150
Other	0
Total	**226**

Screening performance and incidence

Follow-up of positive tests

Follow-up of positive tests are 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 436 babies were referred for paediatric examination or had a request for a second sample because of a positive screen result and 995 because of an unsuitable sample.

Table 15 Newborn screening follow-up by condition, 2014

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
Amino acid breakdown disorders	169	0	167	167	100
Fatty acid oxidation disorders	99	0	99	99	100
Biotinidase deficiency	3	1	0	1	100
Congenital adrenal hyperplasia	50	0	47	47	94
Congenital hypothyroidism	58	8	50	58	100
Cystic fibrosis	57	0	57	57	100
Galactosaemia	0	0	0	0	N/A

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.

The numbers are given in Table 16.

Table 16 Clinical utility of screening, 2014

Disorder	Number of cases	Diagnosis suspected before screen result	Reason for suspicion
Biotinidase deficiency	0	0	
Cystic fibrosis	16	6	1 FH, 2 meconium ileus, 3 failure to thrive
Congenital hypothyroidism	31	0	
Congenital adrenal hyperplasia	2	1	1 ambiguous genitalia
Galactosaemia	0	0	
Amino acid disorders	2	0	
Fatty acid oxidation disorders	9	1	1 FH
Total conditions	60	8	

Overall 60 cases of screened disorders were detected by the programme and in 52 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, 564,525 infants have been screened and 62 cases detected, giving an incidence of 1:9,100. This includes PKU and MSUD.

PKU

There were two cases of PKU found in 2014. Since screening started in 1969 2,643,258 infants have been screened for PKU and 123 cases found, none notified missed, to give an incidence of 1:21,500. Benign hyperphenylalaninemia is not counted in this 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 2014. Since screening started in 1969, 2,643,258 infants have been screened for MSUD, ten classical cases found; none were notified missed, giving an incidence of 1:264,000.

Biotinidase deficiency

Since screening started in 1986, 1,709.964 infants have been screened and eight cases detected giving an incidence of 1:214,000.

Congenital adrenal hyperplasia

Since screening started in 1986, 1,756,913 infants have been screened and 75 cases detected giving an incidence of 1:23,400.

Congenital hypothyroidism

Since screening started in 1981, 1,977,329 infants have been screened and 539 cases detected giving an incidence of 1:3668. There is a trend to an increasing incidence of congenital hypothyroidism 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, 2,692,545 infants have been screened and 400 cases detected giving an incidence of 1:6,700. There were 58,673 babies screened in 2014 (of whom approximately 46% were of European ethnicity) and 15 cases of CF detected. This gives an incidence in the European births of 1:1,800.

Fatty acid oxidation disorders

Since screening started in 2006, 564,525 infants have been screened and 70 cases detected giving an incidence of 1:.8,100.

Galactosaemia

Since screening started in 1973, 2,
515,353 infants have been screened and 24 cases detected giving an incidence of 1:105,000.

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 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 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: 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.