Newborn Metabolic Screening Programme monitoring indicators

This document updates and replaces the indicators in the Newborn Metabolic Screening Programme Monitoring Framework, November 2010.

Regular analysis and reporting of Newborn Metabolic Screening Programme (NMSP) data is a key tool in enabling continuous quality improvement of the programme. The following indicators provide a set of measures that assess the performance of specific components of the programme as well as the programme overall. They cover various points on the screening pathway from taking the sample through to receipt into clinical care of those babies who are subsequently identified with a screened disorder.

Timely reporting against the indicators enables programme providers and the National Screening Unit and to identify where improvements are needed. Reporting on relevant indicators will be quarterly to district health boards (DHBs), while other indicators will form part of an annual report on programme performance and developments. Where appropriate, indicators will be reported by national population, ethnicity and deprivation status.

1: NEWBORN METABOLIC SCREENING COVERAGE

DESCRIPTION

The proportion of babies born who complete newborn metabolic screening.

RATIONALE

Newborn screening must be offered for all babies. All babies whose parents/guardians have consented to screening should have completed screening.

TARGET

≥ 99% of babies born nationally and within each of Maori, Pacific, Asian and Other population groups are screened.

METHOD

Numerator: Number of live births with a first card test result recorded

Denominator: Number of live births within the reporting period

Exclude perinatal deaths.

NOTES

- 'Completed' means a result on first blood spot cards received by the laboratory has been recorded.

2: TIMING OF SAMPLE -TAKING

DESCRIPTION

The proportion of babies screened who have a newborn metabolic screening sample taken between 48 and 72 hours of age.

RATIONALE

Prompt sample collection leads to the best possible chance of a baby with a screened condition receiving early diagnosis and treatment. Severe forms of some of the disorders can be fatal within seven to ten days, and many may not show any signs or symptoms of disease until irreversible damage has occurred. However, the baby must have been independent of their mother long enough for some biochemical markers to show an abnormality. The optimum window for sample collection is between 48 and 72 hours after birth.

TARGET

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

METHOD

Numerator: Number of first blood spot cards received by the laboratory that have a sampling time 48 - 72 hours post birth.

Denominator: Number of first blood spot cards received by the laboratory

Exclude NICU babies.

NOTES

3: QUALITY OF BLOOD SAMPLES

DESCRIPTION

The proportion of samples received by the laboratory that are of satisfactory quality.

RATIONALE

Accurate testing is reliant on a good quality blood spot sample. Unsatisfactory samples require a repeat sample which could have been avoided. This indicator measures the proportion of blood spot samples that require repeating due to a quality issue.

TARGET

≥ 99% of blood spot samples received are of satisfactory quality.

METHOD

Numerator: Number of satisfactory samples received by the laboratory.

Denominator: Number of samples received by the laboratory.

Exclude NICU samples.

NOTES

Reasons for samples being unsatisfactory will be reported according to the following categories:

- timing (taken when the baby was too young)
- collection (insufficient blood, contaminated)
- transport (≥1 month from sampling, damaged in transit).

4: SAMPLE DISPATCH AND DELIVERY

DESCRIPTION

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

RATIONALE

Samples must be received by the laboratory as soon as possible after they are taken.

TARGET

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

METHOD

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

Denominator: Number of samples received by laboratory.

Include NICU samples.

NOTES

Days not hours are used as the basis of calculation.

5: RECEIPT AND FOLLOW-UP OF SECOND SAMPLES

DESCRIPTION

The proportion of second sample requests that had appropriate follow-up (timely receipt of second sample, decline notified or other appropriate follow-up).

RATIONALE

Second samples are requested if first samples give borderline results or are inadequate. Where requested, second samples should be taken as soon as possible.

TARGET

100% of second samples requested are received by the laboratory, had other appropriate follow-up, or were declined by parents/guardians, within ten calendar days of request.

METHOD

Numerator: Number of second samples

- received
- declined or
- had other appropriate follow-up*

within ten calendar days of the request.

Denominator: Total number of second samples requested.

Exclude NICU babies.

Exclude neonatal deaths.

Reported for previous quarter.

NOTES

* 'Follow-up' includes a specialist referral or where testing (for example thyroid tests) was done in a community laboratory.

Follow up after 10 days will also be reported.

6: LABORATORY TURNAROUND TIME POSITIVE RESULTS

DESCRIPTION

The time from receipt of the sample in the laboratory to notification of the referring practitioner or specialist paediatrician of a screen positive result[1].

RATIONALE

Timely processing and notification of screen positive samples is essential to ensure early detection and treatment. This indicator is a measure of laboratory performance.

TARGET

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

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

METHOD

Numerator: Number of samples with screen positive results that are notified to their LMC / specialist paediatrician within the specified number of calendar days for the disorder /group of disorders (from receipt of sample in lab).

Denominator: Number of samples with a screen positive result for a particular disorder/group of disorders

Reported separately for clinical critical and non- clinical critical cases.

NICU babies included.

NOTES

1. 'Screening positive results' are where further action is required on the baby, including paediatrician referral, a second sample, or other test.
2. 'Clinical critical' refers to urgent results. Non clinical critical results may be non-urgent, borderline/mild, many of which require a second sample.

7: AGE OF RECEIPT INTO CLINICAL CARE

DESCRIPTION

For babies with screened conditions, the age of the baby age at transfer into clinical care.

RATIONALE

To ensure babies with congenital metabolic disorders have their development potential impacted as little as possible, all babies with a screened condition must receive a confirmed diagnosis and timely commencement of treatment/active clinical management.

TARGET

100% of babies who receive a screen positive result and are diagnosed with a screened condition receive active clinical management by the following time frames.

Disorder	Age of baby in days – clinical critical conditions	Age of baby in days – non-clinical critical
Amino acid disorders	10	28
Biotinidase deficiency	–	28
Congenital adrenal hyperplasia	10	28
Congenital hypothyroidism	10	28
Cystic fibrosis	–	28
Fatty acid oxidation disorders	10	28
Galactosaemia	10	28
SCID	–	14

METHOD

Numerator: Number of babies who receive a screen positive result, are diagnosed with a screened condition and are received into clinical care within the timeframes specified.

Denominator: Number of babies who receive a screen positive result and are diagnosed with a screened condition.

Reported separately for clinical critical cases and non- clinical critical cases.

Reported by***:***

- Disorder group
- Total cases.

Reported for previous quarter.

NOTES

- All cases are followed up individually to see if programme performance measures across the screening pathway were met and whether newborn screening was beneficial for the baby (refer Appendix 1: *Screen detected and missed cases*).

8: POSITIVE PREDICTIVE VALUE OF THE SCREENING TEST

DESCRIPTION

The probability of a baby having a positive diagnosis for a screened condition given a positive screening result for that condition.

RATIONALE

Positive predictive value (PPV) is a measure of the performance of the screening test. A low PPV means many babies without a screened condition will be referred for diagnostic testing, with associated costs and anxiety for families. Reporting of PPV helps to monitor potential harm of the programme due to identification of false positives through screening.

TARGET

None

METHOD

Numerator: Number of babies with a positive screening result and who subsequently have are diagnosed with a screened disorder.

Denominator: Number of babies with a positive screening result

Exclude NICU babies.

Reported by aggregated data for past five years for each category of conditions.

NOTES

Other measures

The following will also be reported annually:

- incidence of screened disorders over 10 years
- reasons for secondary use of blood spot samples.

Appendix 1: Template for screen detected and missed cases

SCREEN DETECTED AND MISSED CASES													
Diagnosis	**DOB**	**Date confirmed case**	**NICU Y/N**	**Age at sample collection (hours)**	**Transit time (calendar days)**	**Lab TAT to notification (calendar days)**	**Time to receipt 2nd sample (calendar days)**	**Age at transfer to clinical care (days)**	**Age of transfer to clinical care within target?**	**Symptomatic? no/mild /yes**	**Clinical suspicion prior to screen**	**If yes, appropriate test done/not done prior to screen result being available?**	**Clinical utility of screening**
Targets				48-72 - refers to 1st suitable sample	4 calendar d	2days cc 7 days non cc	10 days		10 days clinical critical, 14- 28 days non-clinical critical				

The purpose of this template is to help determine:

- the usefulness of screening for particular cases
- how well the programme performed
- where it did not perform well, what can be learnt
- what cases are missed / diagnosis delayed.

It forms a record of known cases of screened disorder.