



National Screening Unit

Newborn Metabolic Screening Programme Monitoring Framework

November 2010

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Newborn Metabolic Screening Programme Monitoring Framework
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Executive Summary

Monitoring is the routine analysis of service based information to ensure funded services are being delivered. This Monitoring Framework links to the *Antenatal and Newborn Screening Programmes Monitoring and Evaluation Framework 2010*, and supports the routine, systematic collection and recording of information about aspects of the Newborn Metabolic Screening Programme (NMSP) over time.

The purpose of this NMSP Monitoring Framework is to provide a set of national indicators to routinely assess the performance of specific components of the NMSP and the programme overall. This Monitoring Framework covers the screening pathway from informed consent to take screening samples from newborns through to diagnostic testing and treatment.

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 is a priority for the NMSP. Annual monitoring reports will be published on the NSU website.

The NMSP is an organised screening programme. An organised screening programme is characterised by planning, co-ordination, monitoring and evaluation of all activities along the screening pathway to ensure quality in all parts of the programme. Quality is also addressed through the NMSP Policy and Quality Standards, programme evaluation, regular audits and contract management.

National indicators for routine monitoring of the NMSP are:

1. Newborn Metabolic Screening Coverage
2. Timing of sample-taking
3. Quality of blood samples
4. Sample dispatch and delivery
5. Laboratory testing timeframes
6. Timeliness of reporting – notification of screen positives
7. Collection and receipt of second samples
8. Diagnosis and commencement of treatment by disorder
 - Number of cases not found by screening (through paediatric surveillance)
9. Blood spot card storage and return.

Indicators will be developed further as data is collected over time and will be subject to regular review. Data for key indicators is planned to be developed and reported by DHB, ethnicity and deprivation status as appropriate, for review and advice from the expert NMSP Advisory Group.

Introduction

The purpose of this Monitoring Framework is to provide a set of national indicators to assess the performance of specific components of the NMSP and the programme overall.

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 is a priority for the NMSP. Annual reports will be published on the NSU website at www.nsu.govt.nz/health-professionals/1012.asp

Background

The NMSP is overseen nationally by the National Screening Unit (NSU) of the Ministry of Health. Almost all babies born in New Zealand have been screened since the NMSP began in 1969, and as a result, approximately 45 babies are identified with and treated for a metabolic disorder each year. When a baby is diagnosed with a metabolic disorder in early infancy, treatment can commence immediately, preventing life-threatening illness and limiting the impact on the baby's development potential.

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

There are hundreds of metabolic disorders. The NMSP screens for a small percentage of these disorders, focusing on those for which appropriate testing is available and that can be successfully treated in the early newborn period. Disorders are subject to a process to determine their appropriateness for screening before being added to the programme, and once in the programme disorders are monitored to ensure that they continue to be suitable candidates for screening. The programme also monitors new technologies to maintain high quality in the programme.

Tested blood spot cards are stored, with consent from parents/guardians, for quality assurance purposes and possible future uses. Stored cards may be used, with informed consent of parents/guardians or individuals, for research that complies with programme policy and quality standards. Informed consent for research using de-identified cards, and with ethics committee approval, may be provided at the time of sample-taking. Other research requires specific consent.

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. Advice from the NMSP Advisory Group has informed the NMSP published *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand* (Programme Guidelines). Programme Guidelines are referred to in this Monitoring Framework and provide further information around key indicators. The NMSP reports publicly on its uptake rates and outcomes.

Aim and Objectives

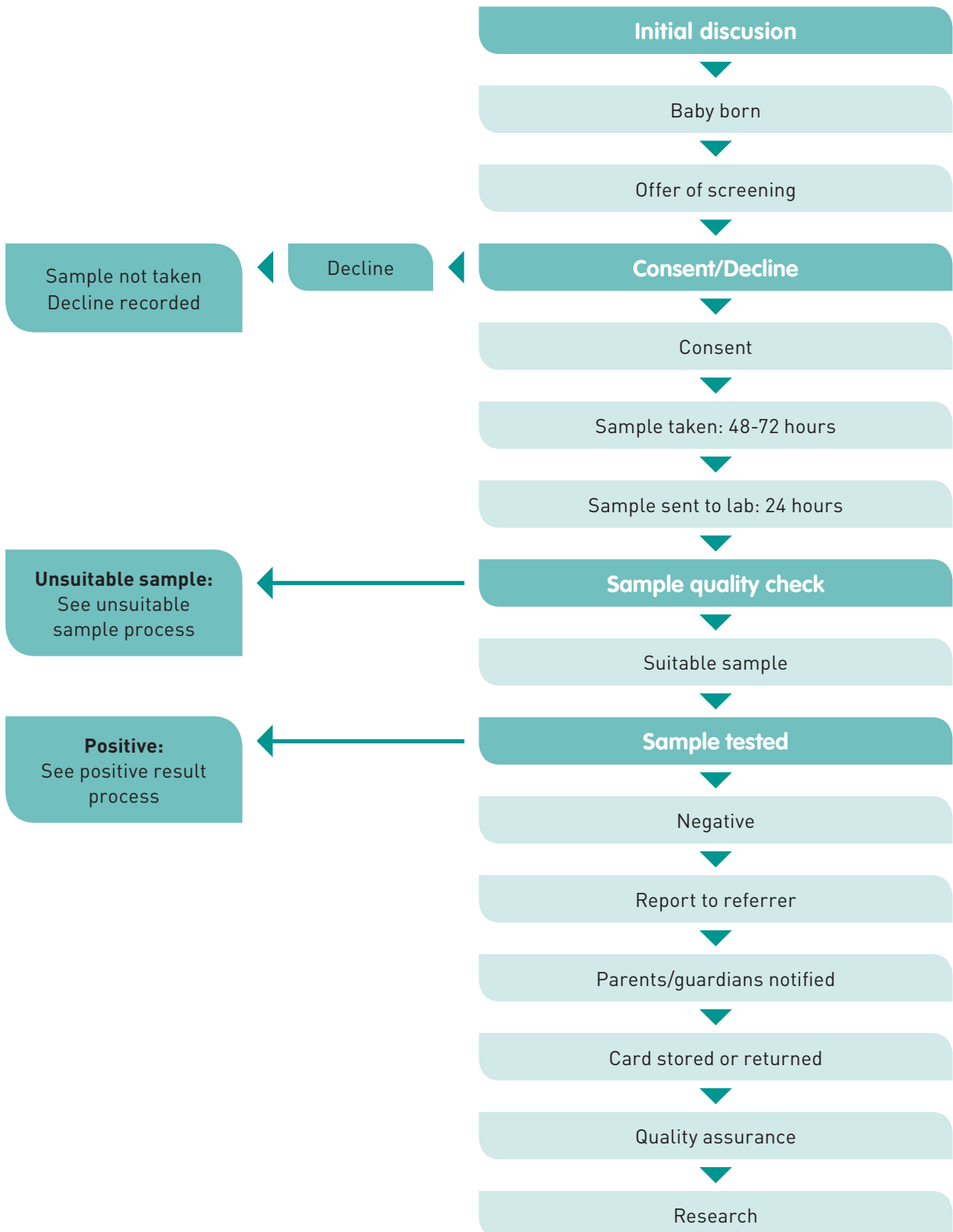
The aim of the NMSP is to reduce newborn morbidity and mortality through high-quality screening that facilitates early detection and treatment of specific metabolic disorders in pre-symptomatic babies.

The objectives of the programme are to:

- enable early detection of pre-symptomatic newborns
- ensure appropriate early 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
- facilitate continuous quality improvement through the development of quality assurance, reporting and a strategic planning framework
- inform the community of all aspects of newborn screening.

Figure 1 outlines the NMSP Screening Pathway.



Newborn Metabolic Screening Programme Logic Model and Stakeholder Map

The NMSP Logic Model

A programme logic model is a planning tool to identify the programme’s key objectives, outcomes and outputs. It describes the purpose and the what, when, and how of the programme.

Figure 2 demonstrates the relationships between inputs, activities, outputs and outcomes and shows the generic structure of programme logic.

Figure 2: A diagram of the relationships between inputs, activities, outputs, outcomes, indicators, and targets of a generic programme logic.

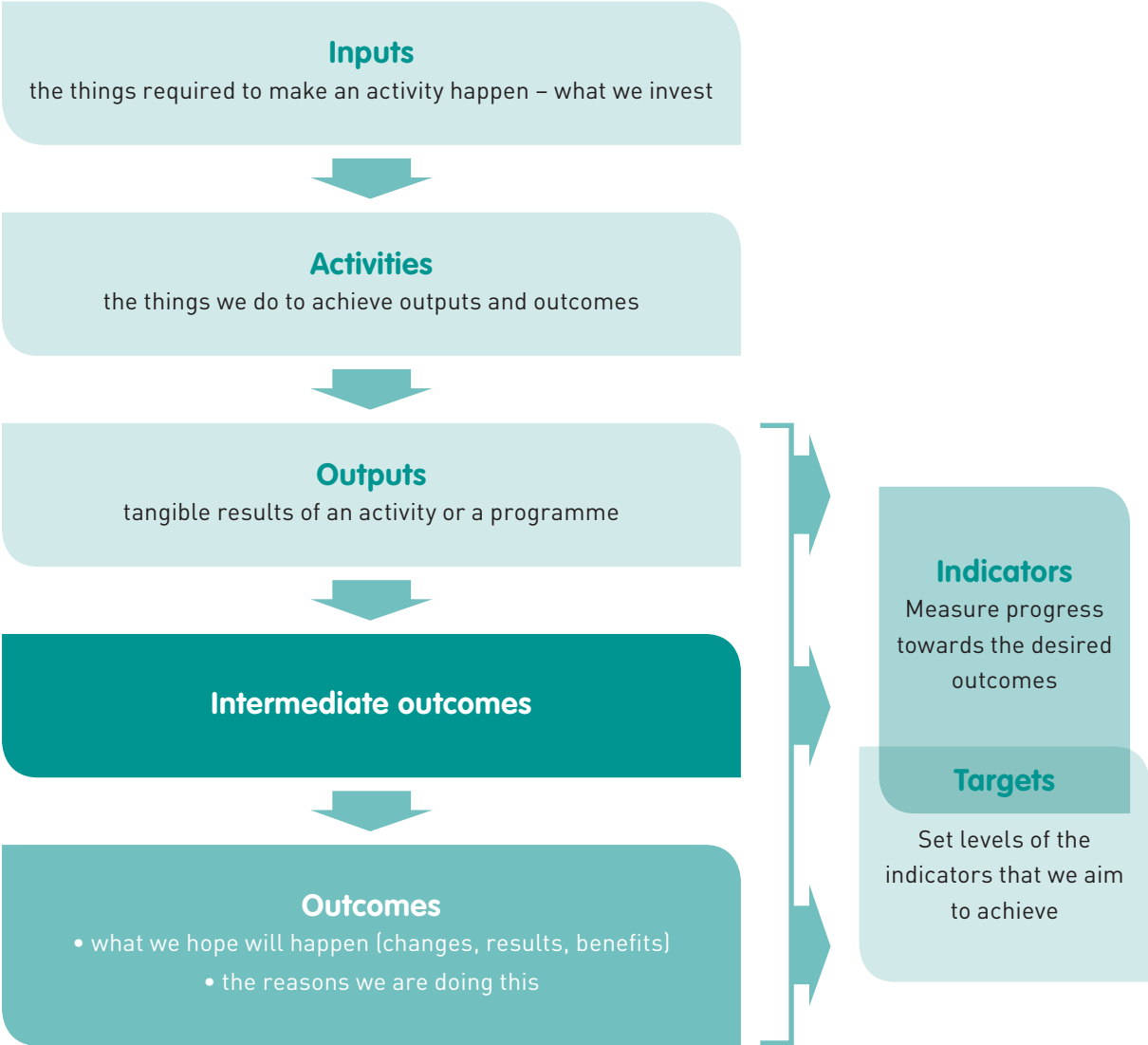
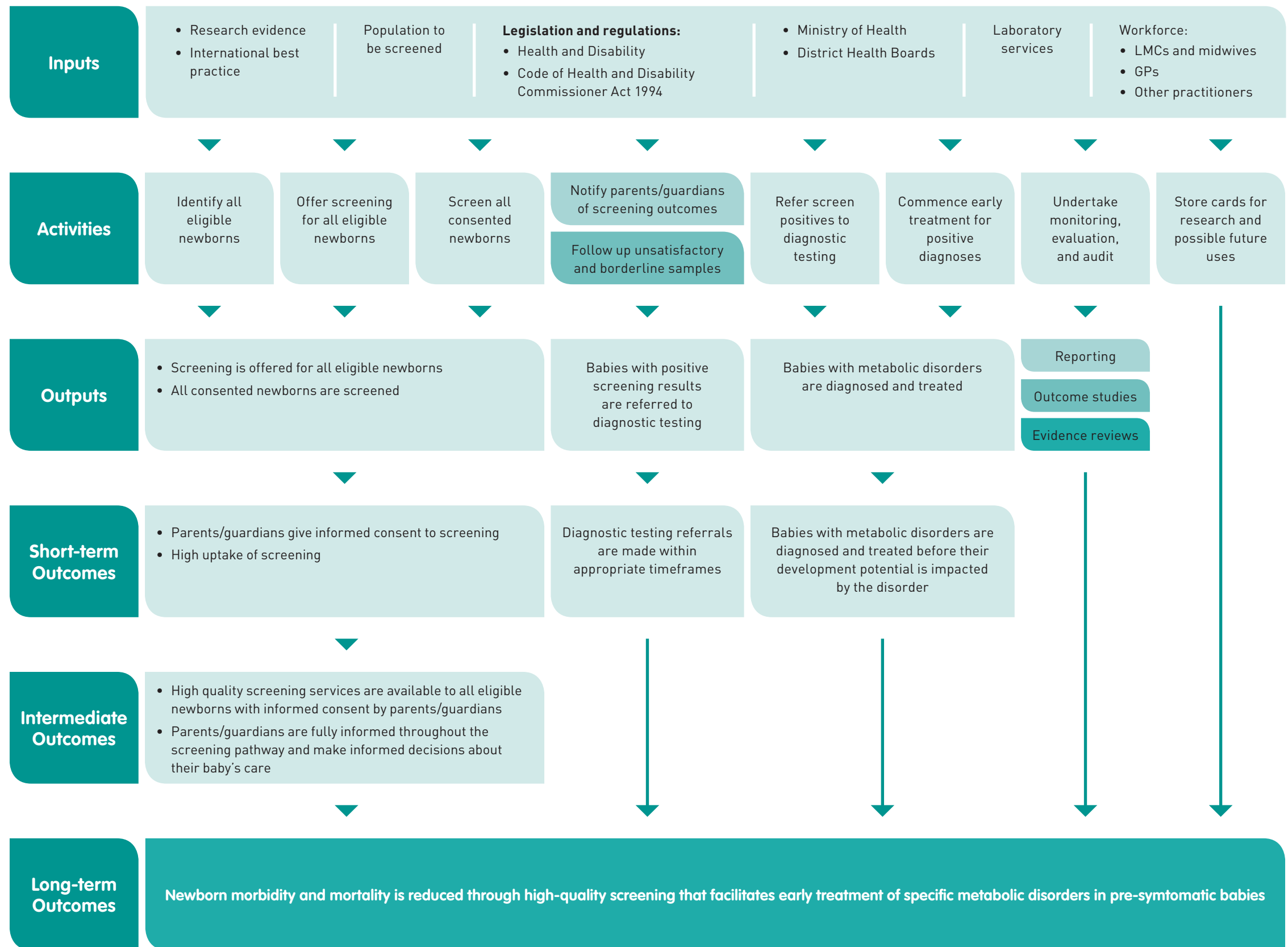


Figure 3 is the NMSP logic model. The NMSP logic model was developed in accordance with Ministry of Health policy as set out in *A Guide to Developing Public Health Programmes: a Generic Programme Logic Model* (Ministry of Health, 2006).

Figure 3: The NMSP logic model



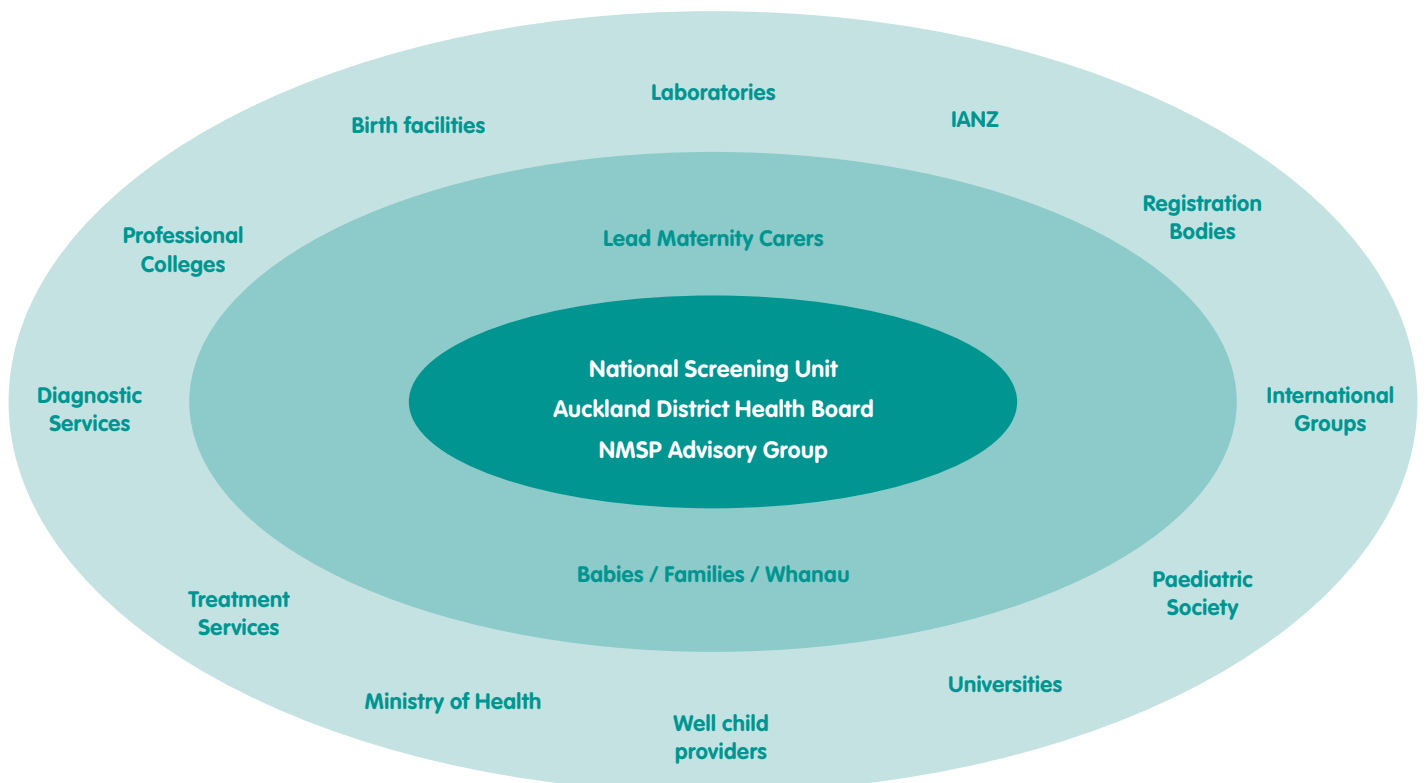
The NMSP Stakeholder Map

Figure 4 is the stakeholder map for the NMSP. The programme's stakeholders include:

- Babies and their families/whanau
- The screening laboratory
- Lead Maternity Carers (LMCs) including obstetricians and midwives
- The NMSP Advisory Group
- District Health Boards, including hospitals, birthing units, and specialist care units
- Diagnostic specialists and paediatricians
- Well child providers including primary health practitioners
- Professional colleges and registration bodies
- Parent support organisations
- Universities and academic representatives
- Ministry of Health: National Screening Unit and Child Youth and Maternity Services (Well Child/Tamariki Ora)
- International Accreditation New Zealand (IANZ)
- New Zealand Health Information Services (NZHIS).

The programme's stakeholders have a key role in the ongoing development of the NMSP. The NSU consults and liaises with key stakeholders on programme documentation and changes to the NMSP.

Figure 4: The NMSP Stakeholder Map



Development of the NMSP Monitoring Framework

Determining Indicators for Monitoring

Screening occurs either opportunistically or within an organised screening programme. Organised screening programmes are distinguished from opportunistic screening by the use of centralised quality management processes, which balance the achievable benefits of screening with the potential harms. Organised screening is delivered through a screening programme with planning, co-ordination, monitoring and evaluation of all activities along the screening pathway.

Selection of appropriate outcomes to monitor is a complex process. The *How to Monitor for Population Health Outcomes: Guidelines for Developing a Monitoring Framework* (Ministry of Health, 2007) has been used to inform the development of indicators to monitor for the NMSP.

It is not always feasible, or necessary, to measure everything. The process of selecting outcomes for monitoring involves identifying what could and should be measured. The steps involved are to:

- identify what *could* be monitored
- prioritise these into those that *should* be monitored if resources allow and the method and frequency is defined
- identify the essential few that *must* be monitored in quarterly, six monthly and annual monitoring reports.

As the NSMP has been operating in New Zealand since 1969, the programme is well established. The development of the Monitoring Framework is an opportunity to review the programme's progress and consider how quality can be improved.

The standards for the NMSP have been set based on evaluation and audit of the programme achievements to date, and through international benchmarking. The first priority was to highlight those areas where gaps have been identified and that can be quickly improved.

The Indicators

What must be monitored:

- Coverage – uptake of metabolic screening (consent/decline)
- Timing of sample-taking (between 48 and 72 hours of birth for maximum utility)
- Quality of blood samples
- Sample dispatch and delivery
- Laboratory testing timeframes
- Timeliness of reporting – notification of screen positives
- Collection and receipt of second samples
- Diagnosis and commencement of treatment
- Number of cases not found by screening (through paediatric surveillance)
- Disorder incidences
- Blood spot card storage and return.

Indicators will be developed further as data is collected over time. Data for key indicators should be reported as indicated /appropriate¹ by:

- DHB
- ethnicity
- deprivation status
- birth location (DHB birthing facility, private birthing facility and home births).

For preterm infants, “corrected age²” should be used for the calculation of all indicators.

What should be evaluated by an audit programme or case review to ensure safety and quality:

- Offer of screening
- Timing of sample despatch and delivery to laboratory
- Whether blood spot cards are fully and correctly completed
- Timing of referrals for diagnostic treatment
- Timing and appropriateness of LMC follow-up where follow-up is required
- Timing of diagnosis and commencement of treatment
- Age at treatment
- Identification of at-risk siblings

What could be monitored or evaluated:

- Provision of information and advice to parents/guardians (to allow informed consent)
- Timing of initial discussion with parents/guardians
- Timing of notification of results to parents/guardians
- Age at negative diagnosis

Reducing Inequalities

A guiding principle underpinning all health targets is the reduction of inequalities for those who have difficulty in accessing services or who currently have worse health status than other New Zealanders, particularly Māori and Pacific peoples, the most deprived and people living with disabilities. Reducing inequalities and improving Māori health are health and disability sector goals, and efforts to achieve these goals are mandated by the New Zealand Health and Disability strategies (Minister of Health, 2000; Minister for Disability Issues, 2001) and the *He Korowai Oranga: Māori Health Strategy* (Minister of Health & Associate Minister of Health, 2002).

The high uptake of screening in the NMSP (close to 100%) indicates that the programme is successful in achieving equity of access. Data on follow-up and treatment outcomes from periodic audit could be reported by ethnicity to determine if inequalities exist in access to treatment.

¹ Note that due to small numbers and need for privacy some data will not be published when there is a risk of identification.

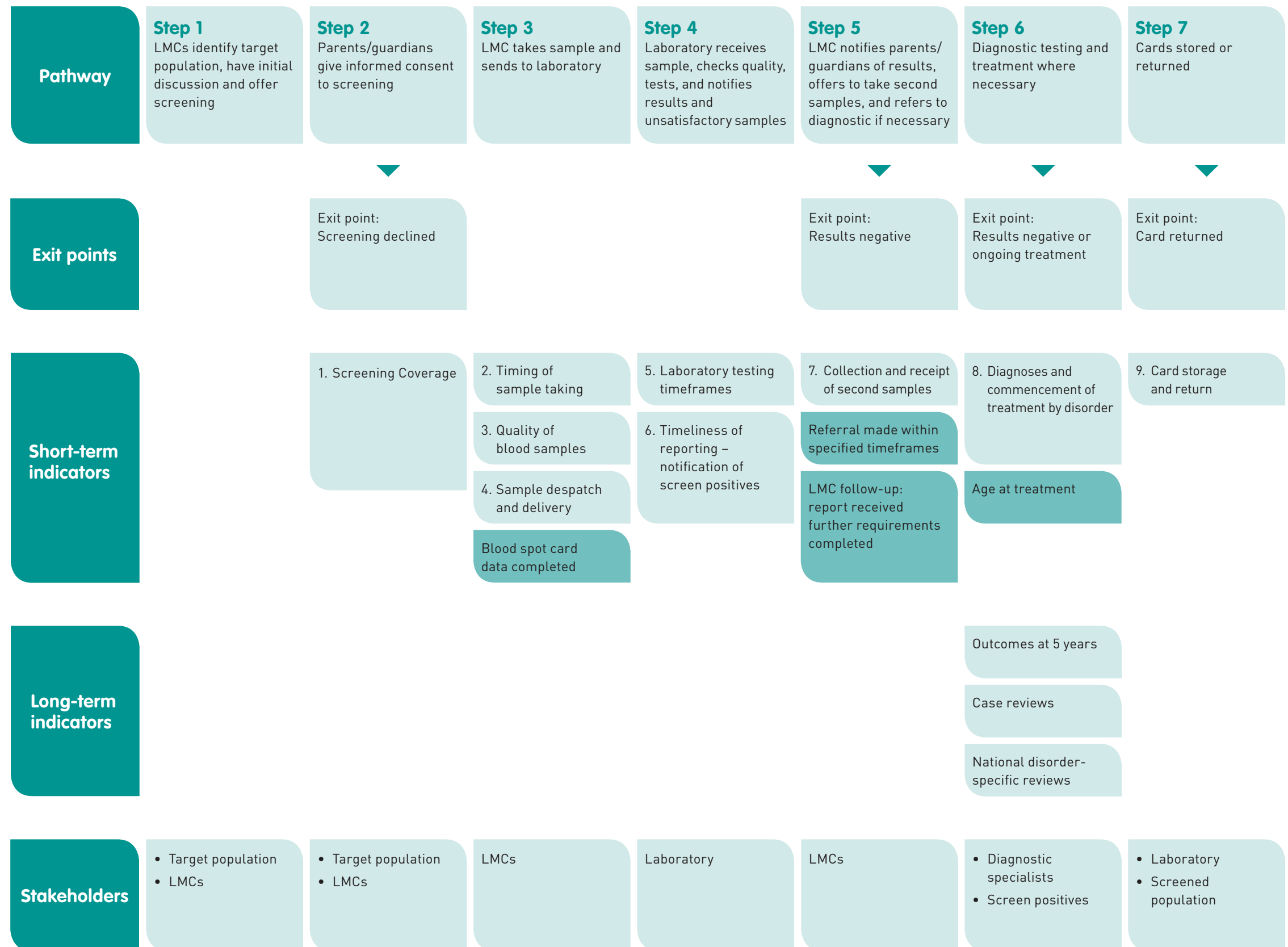
² “Corrected age” is the age a premature baby would be if he/she had been born on their due date. Using a babies’ corrected age allows for the physical and brain development to occur that would have occurred by this time had they been delivered full-term.

Figure 5: summarises all NMSPI indicators used in regular monitoring with reporting frequency and detail.

INDICATORS	QUARTERLY	BIANNUALLY	ANNUALLY	DETAIL
1. Newborn Metabolic Screening Coverage			X	<ul style="list-style-type: none"> • DHB • Ethnicity • Deprivation status
2. Timing of sample taking	X	X	X	<ul style="list-style-type: none"> • DHB • Ethnicity • Deprivation status
Laboratory Reporting				
3. Quality of Blood Samples	X	X	X	<ul style="list-style-type: none"> • DHB
4. Sample dispatch and delivery	X	X	X	<ul style="list-style-type: none"> • 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	<ul style="list-style-type: none"> • DHB
Incidence			X	
8. Diagnosis and commencement of treatment by disorder: <ul style="list-style-type: none"> • 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	

The Screening Pathway and Indicators

Figure 6: is the NMSP screening pathway and monitoring outcomes, showing where each of the indicators sits on the pathway.



The following tables detail how each indicator will be calculated and the rationale behind the indicator

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

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.

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

Notes

- Samples for screening must be taken in accordance with Programme Guidelines (Chapter 7) 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 outcome

All samples are tested within the specified timeframes.

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

Preliminary results for all samples are available by day 2.

Standard

100% of samples meet the following laboratory turnaround times:

Disorder	Working days (from receipt by laboratory)
CAH	2
Galactosaemia	2
Amino acid disorders	2
Fatty acid oxidation disorders	2
Biotinidase deficiency	5
Cystic fibrosis	5
CH	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.

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Appendix A:

Key Terms and Definitions

The table below defines the terms frequently used in this document.

TERM	DEFINITION
Outcomes	Outcomes are specific statements about the intended change in public health related attitudes, knowledge, behaviours, or physical (including mental) health status in the target population(s) sought by undertaking the planned public health activity.
Objectives	Objectives are statements about the results a programme seeks to achieve. Objectives can usually be translated directly into “outcomes”, and can be hierarchical.
Outputs	Outputs are things (e.g. goods) produced, services delivered, events held, or participation generated resulting from the activities undertaken.
Indicators	Indicators are either quantitative or qualitative measures that assess the direction and size of change in the thing being measured.
Targets	A target is a level of performance that we aim to achieve against a specific “indicator”, within a health area.
Programme logic model	A programme logic model graphically identifies and links programme outcomes with interventions and processes and the theory and assumptions.
Monitoring Framework (or plan)	<p>A plan for the routine, systematic collection and recording of framework information about aspects of a programme over time.</p> <p>The purpose is to assess whether progress is being made on achieving the programme objectives.</p>
Policy and Quality Standards	<p>Policy and Quality Standards are an important aspect of ensuring programme quality. The NMSP has a Policy Framework to provide guidance for all programme providers, particularly Lead Maternity Carers (LMCs), maternity managers and laboratories.</p> <p>Audits will be conducted against Policy and Quality Standards.</p>

Source: These definitions are taken from:

- *How to Monitor for Population Health Outcomes: Guidelines for Developing a Monitoring Framework (Ministry of Health, 2007, pg 5 & 42);*
- *A Guide to Developing Public Health Programmes: a Generic Programme Logic Model (Ministry of Health, 2006, pg 3).*

