

Newborn Metabolic Screening Programme Policy Framework

June 2011



National Screening Unit

New Zealand Government

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Section One: Background

1. Introduction

- 1.1 This document sets out the policy framework for the Newborn Metabolic Screening Programme (NMSP). The policy framework provides guidance for all programme providers.
- 1.2 While this policy document has specific sections relating to particular providers, programme providers should ensure that they are familiar with the entire document in order to meet their obligations in regard to the programme.

2. Programme overview

- 2.1 The blood samples are tested for over 20 metabolic disorders. The National Screening Unit (NSU) has responsibility for the Newborn Metabolic Screening Programme (NMSP). The NSU contracts with one District Health Board laboratory to provide screening services.
- 2.2 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, which can assist with preventing irreversible damage and life-threatening illness.
- 2.3 Newborn metabolic screening involves collecting blood samples from babies' heels (the 'heel prick test') onto a blood spot card (sometimes known as a 'Guthrie card'). The optimal time for collection is between 48 and 72 hours of baby's age.
- 2.4 There are hundreds of metabolic disorders. The NMSP screens for a small number of these disorders, focusing on those for which appropriate testing is available and that can be treated in the early newborn period.
- 2.5 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 considers new technologies for possible inclusion in the programme.
- 2.6 Tested blood spot cards are either returned to individuals/parents/guardians or stored indefinitely in secure storage by the NMSP.
- 2.7 Primary uses of blood spots include:
 - the initial screening test
 - repeat confirmatory testing
 - investigation of initial screening test results that may have been a false positive or false negative
 - quality assurance and audit
 - assay improvement and validation of tests for disorders currently in the programme
 - validation of assays for potential new disorders to be added to the newborn screening panel.

- 2.8 Secondary uses of blood spots are:
 - those that benefit the individual and his/her family/whānau
 - forensic/police investigations
 - Coroner investigations
 - mortality review
 - research
 - other requests.
- 2.9 The NMSP is monitored and evaluated by the NSU to ensure it continuously meets high standards. A multi-disciplinary Governance Team and Technical Group provide expert advice for the programme. The programme reports publicly on its uptake rates and results.
- 2.10 Significant changes to this policy document require further stakeholder consultation.

3. The screening pathway

- 3.1 The screening pathway starts with the initial discussion with parents/ guardians and the provision of information and advice to allow them to make informed choices about screening, storage and possible uses of residual blood spots.
- 3.2 Figure 1 shows the screening pathway of the NMSP from the time of initial discussion, consent or decline for screening, consent or decline for storage, laboratory testing and reporting and result notification. Figures 2 and 3 show the pathways (processes) for unsuitable samples and positive results.

Figure 1: The NMSP screening pathway

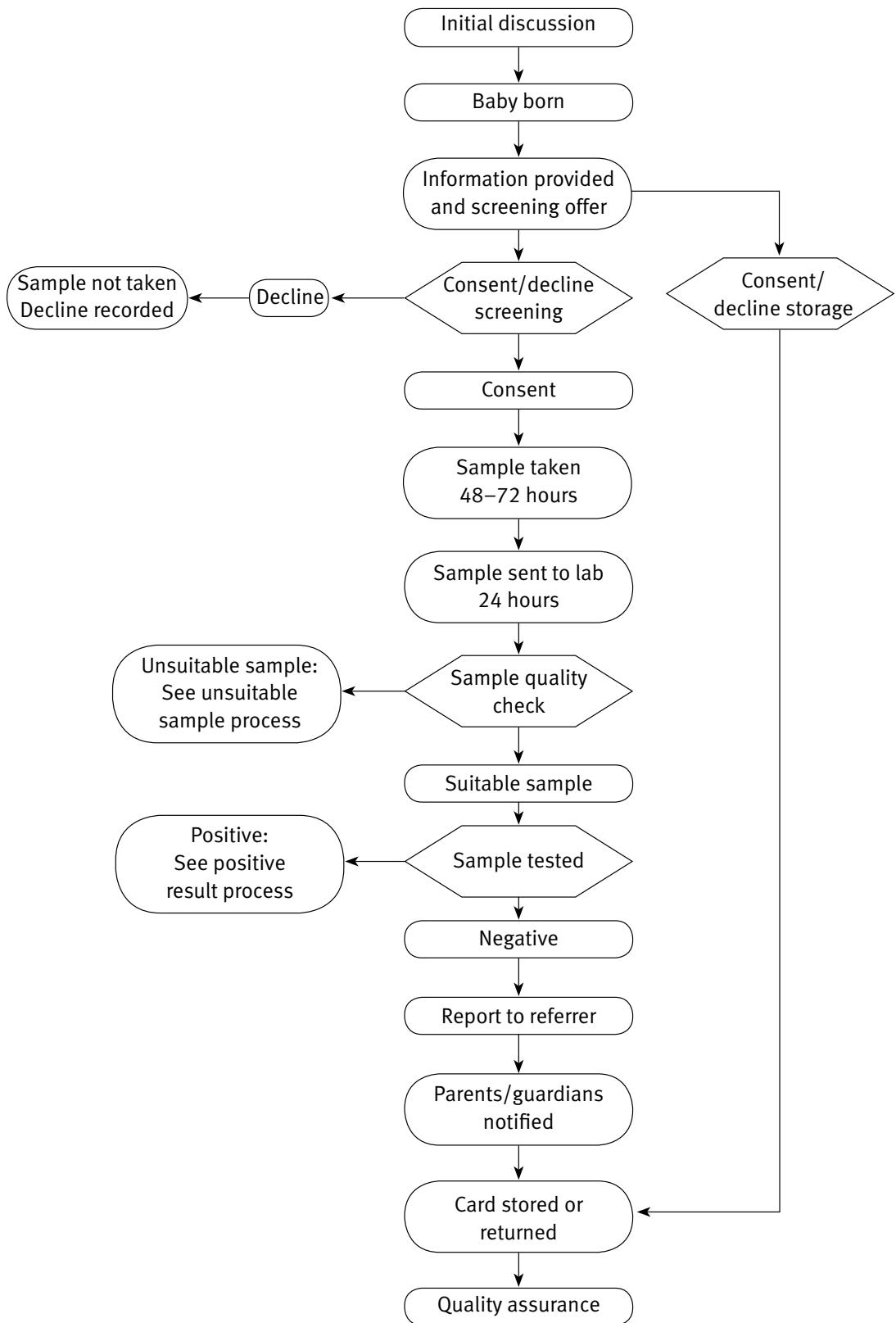


Figure 2: Unsuitable sample process

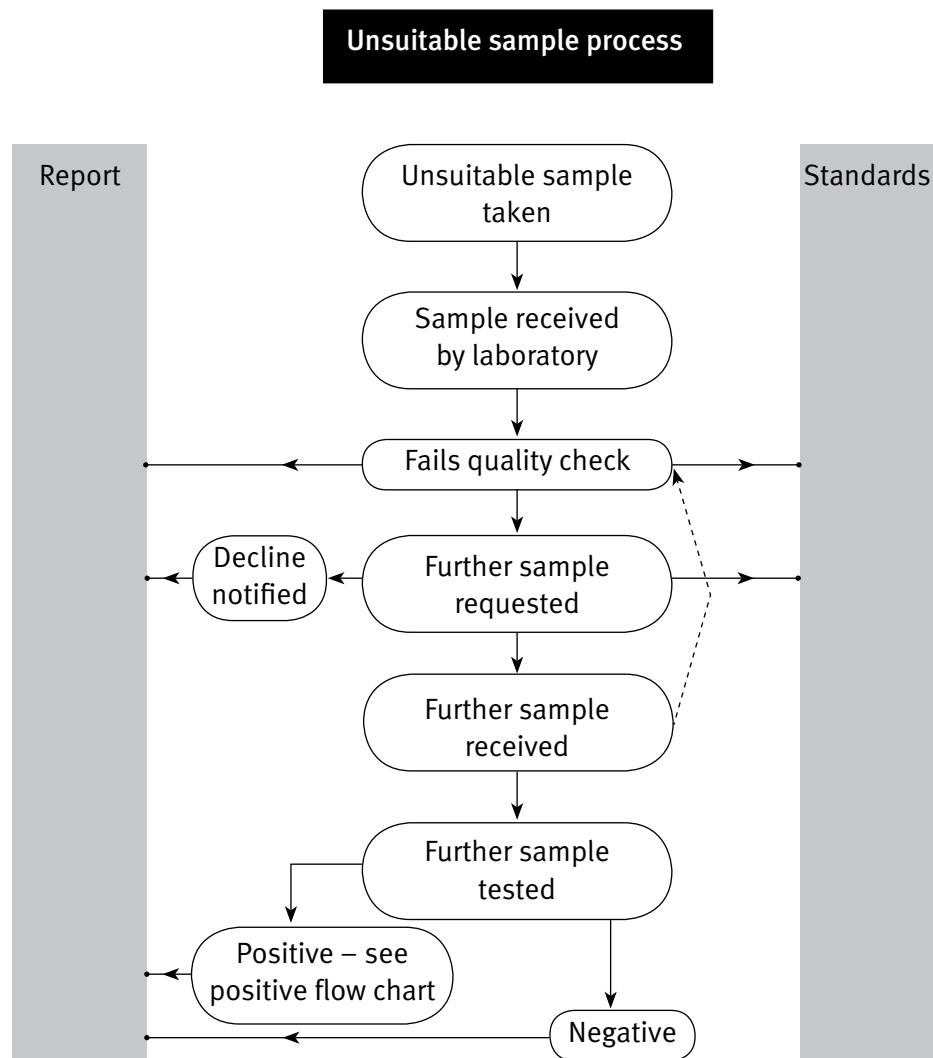
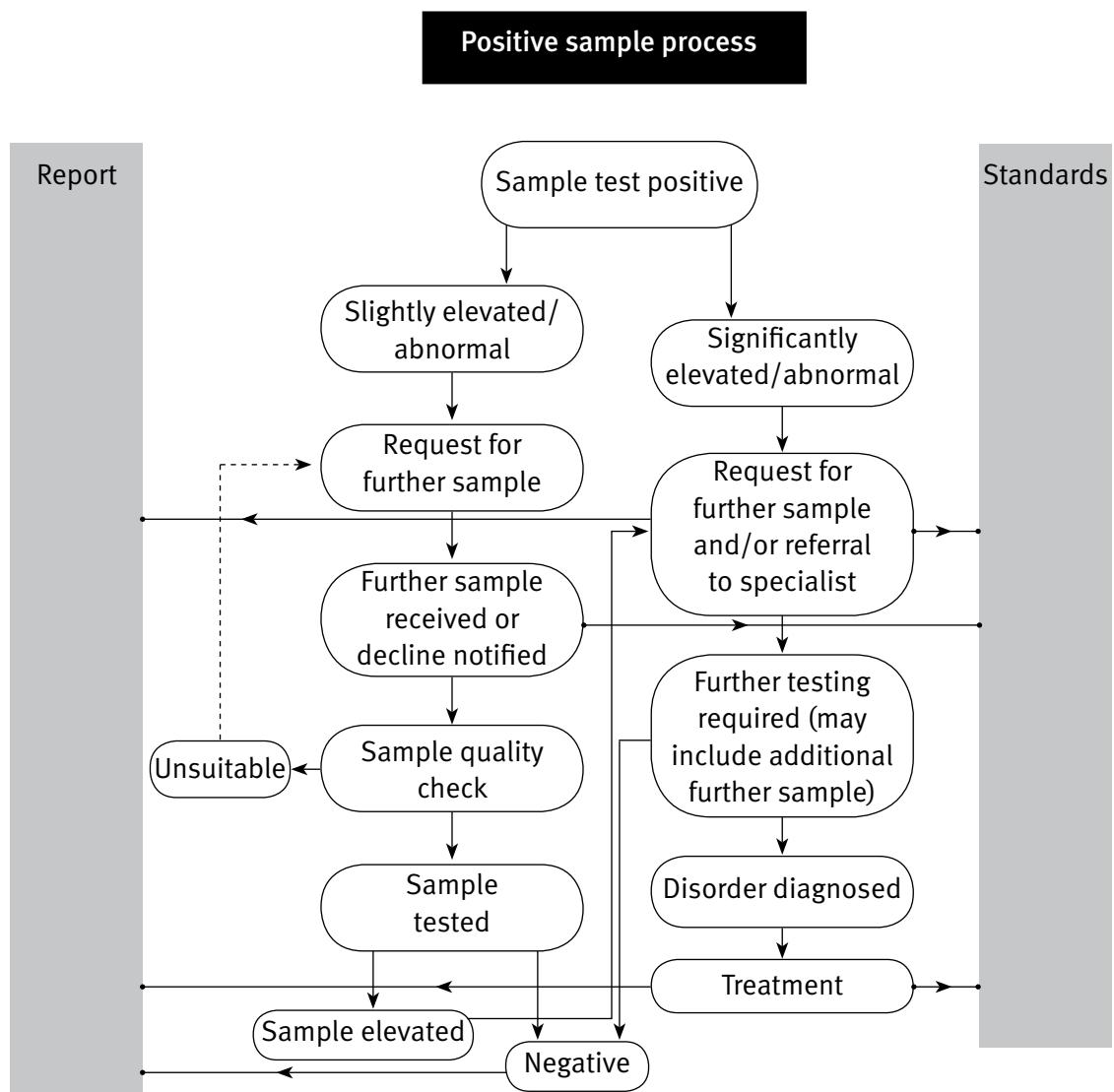


Figure 3: Positive sample process



4. Aim and objectives

- 4.1 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.
- 4.2 The objectives of the programme are to:
 - (a) enable early detection of pre-symptomatic newborns
 - (b) ensure appropriate early referral to treatment of newborns
 - (c) ensure babies born with congenital metabolic disorders have their development potential impacted as little as possible from the disorder
 - (d) facilitate early diagnosis, appropriate treatment and continuous monitoring of specific metabolic disorders
 - (e) maintain high uptake of screening, community participation and trust
 - (f) facilitate continuous quality improvement through the development of quality assurance, reporting, education and the strategic planning framework
 - (g) inform the community of all aspects of newborn screening including the storage and use of blood spot cards.

5. Ministry of Health recommendation

Newborn metabolic screening is strongly recommended by the Ministry of Health.

6. Screening programme requirements

- 6.1 The NMSP is an organised screening programme. An organised screening programme is characterised by planning, coordination, monitoring, and evaluation of all activities along the screening pathway to ensure quality in all parts of the programme.
- 6.2 The key elements for an organised screening programme are¹:
 - (a) an identified target population
 - (b) facilities for screening and interpretation of the screened material
 - (c) quality control both within and between centres for the screening procedure and its interpretation
 - (d) an agreed referral system
 - (e) a reliable fail-safe procedure to ensure that action is taken on all positive results
 - (f) facilities for the diagnosis and appropriate treatment of screening detected disease, and for the follow-up of treated individuals
 - (g) systematic evaluation and monitoring of the whole programme
 - (h) training for all key personnel.

¹ Fowler and Austoker.1997. Screening. In Detels R, Holland W, McEwan J and Omenn G (eds). *Oxford Textbook of Public Health*. Third edition Vol 1. Oxford: Oxford University Press.

7. Quality framework

- 7.1 The document *Improving Quality: A Framework for Screening Programmes in New Zealand* (National Screening Unit, 2005) underpins quality improvement initiatives for all screening programmes.
- 7.2 Other key documents are the *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand*, February 2010 and the *NMSP Monitoring Framework*, January 2011. These documents are available on www.nsu.govt.nz

Section Two – Programme Policy

1. Programme responsibilities

1.1 Purpose

This policy sets out the responsibilities of the National Screening Unit, Ministry of Health, with regard to the NMSP.

1.2 Background

The NSU of the Ministry of Health is responsible for the safety, effectiveness, and quality of health and disability screening programmes. The NSU leads, oversees and coordinates organised screening programmes in New Zealand.

1.3 The core functions of the NSU include:

- (a) national coordination, leadership, and advice to government regarding screening
- (b) research and development, including evaluation of new evidence related to screening, and evidence-based appraisal of technological advances in screening
- (c) developing frameworks, standards, and policy, monitoring performance, and evaluating screening services
- (d) coordination, leadership, and development of the screening workforce
- (e) administering legislation related to screening programmes
- (f) identifying under-screened groups and developing effective strategies to improve participation.

1.4 Requirements

The NSU's coordination and leadership of the NMSP includes:

- (a) management, oversight, and strategic direction
- (b) contracting laboratory and programme coordination services (currently from Auckland District Health Board (ADHB))
- (c) working closely with programme providers and District Health Boards (DHBs) to improve coverage and quality
- (d) provision of documentation to support the programme, including:
 - (i) policy documentation
 - (ii) programme standards
 - (iii) practitioner guidelines
 - (iv) electronic and hard copy information for consumers
 - (v) audit requirements
 - (vi) referral advice for practitioners in the case of positive screening results
- (e) practitioner education, including:
 - (i) continuing education modules
 - (ii) web-based information

- (f) appointment and leadership of the multi-disciplinary NMSP Governance Team and Technical Group
- (g) monitoring
- (h) evaluation
- (i) audit
- (j) public consultation and reporting
- (k) memoranda and/or contractual obligations with other parties, including with the New Zealand Police regarding access to blood spots.²

2. Participation in and access to screening

2.1 Purpose

This policy describes the criteria for participation in and access to screening.

2.2 Background

- (a) Newborn metabolic screening must be offered to all eligible babies born in New Zealand. Informed consent is a process that must be integrated throughout the screening pathway.
- (b) The *Health and Disability Services Eligibility Direction 2011* sets out the eligibility criteria for publicly funded health and disability services in New Zealand. A *Guide to Eligibility* is available on the Ministry of Health website www.moh.govt.nz/eligibility

2.3 Requirements

- (a) Newborn metabolic screening should be offered to all parents/guardians during pregnancy with sufficient information to allow informed consent.
- (b) Participation requires verbal informed consent from parents/guardians.
- (c) Consent and discussion must be recorded in the clinical notes.
- (d) Consent must be obtained both for screening, and for storage and possible future uses of blood spot.
- (e) For the return of blood spots to parents/guardians a written request is required.

3. Health professional stakeholders

3.1 Purpose

This policy lists the health professional stakeholders for the NMSP and describes NMSP interaction.

3.2 Background

The NSU consults colleges, consumers, organisations and groups on programme documentation and changes to the programme.

The NMSP, through the NSU and the laboratory, maintains current contact details for endocrinologists, metabolic physicians, dieticians, paediatricians, nurse educators, maternity managers, Lead Maternity Carers (LMCs), and other stakeholders, and liaises with these stakeholders regularly.

² The current Memorandum of Understanding with the New Zealand Police is available at www.nsu.govt.nz

3.3 Requirements

The NMSP liaises with the following health professional stakeholders.

Stakeholder	Nature of Linkage
College of Midwives Midwifery Council College of General Practitioners College of Obstetricians and Gynaecologists	<ul style="list-style-type: none"> Notification of updates to the programme, practitioner responsibilities, education resources CME certification requirements Education of practitioners
Dieticians	<ul style="list-style-type: none"> Liaison with the treatment team
District Health Boards	<ul style="list-style-type: none"> Education of practitioners Communication with individual practitioners Communication with maternity managers and other key staff
International Accreditation New Zealand (IANZ)	<ul style="list-style-type: none"> Audit and monitoring Programme peer review
Lead Maternity Carers	<ul style="list-style-type: none"> Liaise and work with LMCs. Information for families is provided through the LMC
Ministry of Health national systems and collections	<ul style="list-style-type: none"> Data collection/comparison
Paediatric Society	<ul style="list-style-type: none"> Consultation on practitioner resources, amendments to the programme
Paediatric Surveillance Unit	<ul style="list-style-type: none"> Data collection of rare disorders
Paediatricians Endocrinologists Metabolic physicians	<ul style="list-style-type: none"> Notification of positive results
University liaison	<ul style="list-style-type: none"> Midwifery education resources and training Outcome studies
Well child providers Primary health carers	<ul style="list-style-type: none"> Follow-up of babies who have consented but not completed screening or have positive test results
Australian programme links	<ul style="list-style-type: none"> Benchmarking of data, sharing issues, and quality initiatives

4. NMSP Governance Team and Technical Group

4.1 Purpose

This policy describes the roles of the NMSP Governance Team and Technical Group.

4.2 Background

The NMSP has a multi-disciplinary governance team and technical group appointed and led by the NSU.

The NMSP Governance Team comprises members who collectively have a wide knowledge and experience of newborn metabolic screening, including health practitioners, representatives of professional organisations, and consumers.

The NMSP Technical Group is a subset of the NMSP Governance Team.

The NSU Clinical Governance Group provides clinical, public health and strategic advice to support the delivery of the NSU's screening programmes.

4.3 Requirements

- (a) The role of the NMSP Governance Team is to:
 - (i) review, critique and interpret annual programme reports and make recommendations to the NSU
 - (ii) provide recommendations to the NSU Clinical Governance Group on changes to screened disorders or the introduction of new technology
 - (iii) provide recommendations to the NSU Clinical Governance Group on research proposals for blood spot cards
 - (iv) provide advice on the strategic direction of the NMSP
 - (v) provide advice from time to time on other areas of the NMSP as agreed by the Group and the NSU
 - (vi) share responsibility with the NSU for providing liaison back to their respective formal bodies and constituencies.³
- (b) The role of the NMSP Technical Group is to:
 - (i) review, critique and interpret NMSP monitoring and evaluation reports and make recommendations to the NSU
 - (ii) provide advice regarding technical and clinical aspects of the NMSP
 - (iii) provide advice from time to time on other areas of the NMSP as agreed by the Group and the NSU
 - (iv) share responsibility with the NSU, for providing liaison back to their respective formal bodies and constituencies.⁴

5. Programme reporting

5.1 Purpose

This policy describes the NMSP approach to programme reporting.

5.2 Background

The types of reporting include:

- (a) quarterly reporting on specified programme indicators
- (b) quarterly reporting on operational requirements
- (c) annual reporting on programme performance
- (d) reporting on the storage and use of blood spot cards
- (e) reporting on specific projects or events
- (f) scientific reporting at conferences or in written literature
- (g) annual public reporting which may combine a number of the above.

³ For those members who are endorsed by their college/body.

⁴ For those members who are endorsed by their college/body.

5.3 Requirements

- (a) Quarterly reporting on specified programme indicators will be provided by the laboratory to the NSU and the NMSP Technical Group for evaluation and critique.
- (b) Quarterly operational reporting will be provided by the laboratory to the NSU for evaluation and critique.
- (c) Annual reporting on programme performance will be provided by the laboratory to the NSU and NMSP Governance Team.
- (d) From time to time the NSU will report on specific projects or events. The types of events reported are significant changes to the programme, such as addition of new disorders, utilisation of new technology, changes to storage requirements or changes to possible future uses of blood spot cards. All of these events are subject to approval processes.
- (e) Scientific reporting is expected at regular intervals.
- (f) Public reporting will include a combination of the above reports.

6. Monitoring and evaluation

6.1 Purpose

This policy sets out the criteria for monitoring and evaluation of the programme.

6.2 Background

Monitoring and evaluation of the NMSP follows the screening pathway.

6.3 Requirements

- (a) The NMSP is monitored and evaluated through the following activities:
 - (i) regular analysis of data against programme indicators in accordance with the NMSP Monitoring Framework
 - (ii) IANZ accreditation for laboratory services
 - (iii) peer review audits
 - (iv) participation in the Centre for Disease Control (CDC) quality control programme
 - (v) collection of routine surveillance data
 - (vi) internal laboratory audits
 - (vii) regular liaison with Australian metabolic screening programmes
 - (viii) measurement against Australian cut-off levels and disorder definitions
 - (ix) specific quality improvement projects (for example, audit of transit times)
 - (x) benchmarking against other countries, specifically Australia
 - (xi) outcome studies
 - (xii) surveys, which include practitioner evaluations.
- (b) National indicators for reporting and routine monitoring of the NMSP are:
 - (i) newborn metabolic screening coverage
 - (ii) timing of sample-taking
 - (iii) quality of blood samples
 - (iv) sample dispatch and delivery

- (v) laboratory testing timeframes
 - (vi) timeliness of reporting – notification of screen positives
 - (vii) collection and receipt of second samples
 - (viii) diagnosis and commencement of treatment by disorder
 - (ix) blood spot card storage and return.
- 6.4 The national indicators will be developed further as data is collected over time and will be subject to regular review. The NMSP Monitoring Framework is available on www.nsu.govt.nz

7. Branding

7.1 Purpose

This policy sets out the criteria for branding the materials, resources and publications for the programme.

7.2 Background

The NSU has developed branding for the NMSP, including graphics, descriptor and colour sets. This branding should be utilised in all programme communications and publications to enhance the programme’s recognition and profile and to allow practitioners and parents/guardians to easily identify official programme communications.

7.3 Requirements

All materials, resources, and publications issued by the programme, including all results, reports, and communications issued by the provider must:

- (a) have the words ‘Newborn Metabolic Screening Programme’ clearly set out at the top of each document in a minimum font size of 12 pt Arial, above the document’s title or heading
- (b) where appropriate and possible, use the graphics, descriptor, and colour sets for the NMSP in accordance with the National Screening Unit Visual Standards Guide
- (c) comply with the current:
 - (i) Ministry of Health Communication Standards
 - (ii) Antenatal and Newborn Screening Programme ‘Mood Board’.

8. Improving Māori health

8.1 Purpose

This policy describes the NMSP obligations with regard to improving Māori health.

8.2 Background

- (a) The New Zealand Public Health and Disability Act 2000 established statutory obligations for DHBs to reduce inequalities by improving the health status of Māori, and increase Māori participation in the health and disability sector.
- (b) He Korowai Oranga (the Māori Health Strategy) sets out a framework to improve the health status of Māori. Core factors for improving health services for Māori include:

- (i) ensuring systems/processes are in place to facilitate the routine involvement of Māori in service development, planning and delivery
- (ii) addressing access to health care barriers, such as financial and geographical barriers
- (iii) providing health care services in a culturally appropriate manner.

8.3 Requirements

- (a) NMSP providers have obligations to:
 - (i) develop and implement Māori health plans that identify the specific ways in which they will contribute to improving outcomes for Māori
 - (ii) reduce access barriers to NMSP for Māori
 - (iii) facilitate the involvement of Māori throughout service design, development and delivery
 - (iv) develop relationships with Māori health providers
 - (v) develop staff competencies to meet the specific needs of Māori, including cultural sensitivities regarding human tissue.
- (b) The NSU will evaluate NMSP providers in accordance with these criteria, and monitor service provision and outcome data for Māori newborns.

9. Reducing inequalities

9.1 Purpose

This policy describes the NMSP responsibilities for reducing inequalities in programme delivery.

9.2 Background

- (a) Inequalities in health are differences in health status that are unnecessary, avoidable and unjust (Braveman and Gruskin 2003). Reducing inequalities is a health and disability sector goal. Efforts to reduce inequalities are mandated by a number of instruments including the New Zealand Health Strategy, and in the case of District Health Boards, the New Zealand Public Health and Disability Act 2000.
- (b) Coverage rates for the NMSP are estimated to be around 99% of the newborn population.
- (c) There are some disorders that are more prevalent in certain populations, such as cystic fibrosis in the European population.

9.3 Requirements

- (a) The NMSP monitors the screened disorders for any ethnic variations.
- (b) In the future the NMSP may gather data on the approximately 1% of newborns who are not screened, to analyse whether there are any trends in ethnicity or district.

Section Three – Programme Provider Responsibilities

1. Programme provider responsibilities

- 1.1 This section applies to all programme providers for the NMSP, including:
- (a) the programme’s contracted laboratory (currently the Auckland District Health Board (ADHB) laboratory)
 - (b) Lead Maternity Carers (LMCs)
 - (c) hospital midwives, phlebotomists, and nurses who provide screening services.

2. Legislative compliance

2.1 Purpose

This policy sets out the legislative provisions that NMSP programme providers must comply with.

2.2 Background

There is no overarching single piece of legislation governing the NMSP. The legislative framework is formed by a number of different pieces of legislation which cover different parts of the programme.

2.3 Requirements

Programme providers must meet professional and ethical standards and comply with legislation, including the:

- (a) Care of Children Act 2004
- (b) Code of Health and Disability Services Consumers’ Rights Regulation 1996
- (c) Health Act 1956
- (d) Health Information Privacy Code 1994
- (e) Health Practitioners Competence Assurance Act 2003
- (f) Human Rights Act 1993
- (g) Medical Practitioners Act 1995
- (h) New Zealand Bill of Rights 1990
- (i) New Zealand Public Health and Disability Act 2000
- (j) Official Information Act 1982
- (k) Human Tissue Act 2008
- (l) Standard NZS 8135:2009
- (m) Privacy Act 1993
- (n) Public Records Act 2005.

3. Use of health information

3.1 Purpose

This policy describes the criteria for managing health information collected, created and stored as part of the NMSP.

3.2 Background

Pursuant to section 4(1) of the Health Information Privacy Code 1994, health information is:

- (a) information about the health of an individual, including his or her medical history
- (b) information about any disabilities that individual has, or has had
- (c) information about any health services or disability services that are being provided, or have been provided, to that individual
- (d) information provided by that individual in connection with the donation, by that individual, of any body part or any bodily substance of that individual or derived from the testing or examination of any body part, or any bodily substance of that individual
- (e) information about that individual which is collected before or in the course of, and incidental to, the provision of any health service or disability service to that individual.

3.3 Requirements

- (a) Programme providers must ensure that:
 - (i) health information and data are collected, used, stored, accessed and destroyed to a standard that complies with the Health Information Privacy Code 1994
 - (ii) individual clinical records are unique to women and their babies
 - (iii) information is protected from unauthorised use or disclosure
 - (iv) parents/guardians are fully informed of the purpose, use, and recipients of information that is collected, and any consequences of not supplying the information.
- (b) Health information and data are used only:
 - (i) to interpret screening results
 - (ii) to report screening results
 - (iii) to monitor and evaluate the screening programme
 - (iv) in association with primary and secondary uses outlined in this policy.
- (c) Data is provided to the Ministry of Health for monitoring, evaluation and reporting.

4. Data disclosure

4.1 Purpose

This policy describes the criteria for access to de-identified NMSP laboratory data by practitioners outside of the NMSP laboratory, NMSP clinical staff and the Ministry of Health. This is for the purpose of monitoring/evaluating and reporting on the performance of the NMSP. This includes access for conferences, studies, projects, journal articles or other uses. This policy covers data disclosure only and not access or use of residual blood spot samples.

4.2 Background

The NMSP collects data for the primary purpose of screening newborns for specific metabolic disorders. This data collected includes general information about the mother, baby and LMC, the screening results and data associated with positive cases.

4.3 Applications

- (a) Applications for access to the data must be in writing on the Antenatal and Newborn Screening Programmes application form. This form is available on the NSU website www.nsu.govt.nz
- (b) Applications will then be considered by the Programme Director, NSU Programme Leader and the Chair of the NMSP Governance Team. The programme reserves the right to request additional information to support the application.
- (c) The NSU will inform the applicant of any costs associated with data access. All requests will be recorded and documented by the programme. This documentation will be held and managed by the Auckland District Health Board.
- (d) The NSU will inform the applicant of the outcome of their request.
- (e) Data will only be provided for the time period agreed to between the requestor and the NSU and must only be used for the purpose identified and agreed to in the application in accordance with the Data Access Agreement (Appendix F). Data must not be copied or forwarded to any other individuals or organisations other than co-requestors identified and agreed to by the NSU.

4.4 Security of data

All data must be held securely in compliance with legislation and guidance specified under data management in Section 5 in this document. The NSU is unable to provide direct access to raw data. No data will be released that could lead to the identification of individuals.

4.5 Reports and articles

Practitioners are asked to declare that they will acknowledge the assistance of the NSU and the programme in any manuscripts submitted for publication or conferences. However, the NSU does not exercise any control over where results are published, nor should the NSU be included in authorship. The NSU does require a copy of articles or abstracts for review prior to submission for publication or presentation at meetings. The NSU also requests a final copy of all articles, abstracts or publications.

5. Media communication

5.1 Purpose

This policy describes the criteria for communicating with the media about the NMSP. The policy applies to all programme providers.

5.2 Background

- (a) The NSU is responsible for all media communication regarding the NMSP. All media communication must comply with the Ministry of Health's media policy.
- (b) Any communication with the media initiated by the contracted laboratory about the NMSP requires prior approval from the NSU. The NSU will review and have editing rights to media material relating to the NMSP and return it to contracted laboratory within 48 hours, where possible.
- (c) The NSU contact for all media enquiries and referrals is the NSU NMSP Programme Leader.

5.3 Requirements

Programme providers must:

- (a) refer all media enquiries to the NSU NMSP Programme Leader for comment prior to responding to such enquiries
- (b) seek prior written approval from the NSU to respond to any enquiries or to initiate communication with the media
- (c) forward media material relating to the NMSP to the NSU for review prior to any public release.

6. Complaint management

6.1 Purpose

This policy describes the criteria for managing complaints from families/whānau, consumers, and the public about the NMSP.

6.2 Background

The NSU has a process to manage all complaints and queries from practitioners, programme providers or consumers. All programme providers are expected to have a process for managing complaints.

6.3 Requirements

Programme providers must:

- (a) provide families/whānau with a procedure for making complaints
- (b) maintain clearly defined processes for identifying, managing and resolving complaints that:
 - (i) are understood and implemented by all staff
 - (ii) have the underlying principle of being resolved at the lowest possible level
 - (iii) comply with legislative and contractual requirements
 - (iv) comply with the Code of Health and Disability Services Consumers' Rights Regulation 1996
- (c) record all complaints, comments and suggestions in a specific service logbook, file, and/or database
- (d) identify specific personnel with responsibility for ensuring that the complaint management process is effective and efficient
- (e) treat every complaint with anonymity and confidentiality
- (f) inform the NMSP Programme Leader of any serious complaint at the earliest opportunity.

7. Adverse and sentinel event management

7.1 Purpose

This policy describes the requirements programme providers must follow in the event of an adverse or sentinel event.

7.2 Background

- (a) An adverse event is one that may significantly compromise programme activities, and/or an event for which a facility fails to take corrective actions in a timely manner.
- (b) A sentinel event is an event that signals something serious has occurred and warrants in-depth investigation.

7.3 Requirements

Programme providers must:

- (a) have robust written processes that are followed in the event of an adverse and/or sentinel event
- (b) record all adverse and/or sentinel events using the template in Appendix C, including:
 - (i) false negative/missed cases
 - (ii) failure to give results in a timely manner
 - (iii) accidents, incidents, near misses and clinical events
 - (iv) complaints and suggestions
 - (v) other reportable events as indicated by legislation, regulation, professional practice standards and contracts
- (c) ensure records are available for review during audit and assessment visits
- (d) report adverse and/or sentinel events verbally and in writing at the earliest opportunity to the N MSP Programme Leader
- (e) agree to a review by a party designated by the NSU if the NSU deems that programme quality has been compromised by an adverse and/or sentinel event.

Section Four – Lead Maternity Carer Responsibilities

1. Lead Maternity Carer responsibilities

- 1.1 This section outlines specific provisions for LMCs. Other parts of this document are also applicable to practitioners including LMCs who provide newborn metabolic screening.
- 1.2 If care has been transferred and the woman is in the care of the secondary/tertiary service, that service is responsible for the screening process.
- 1.3 If there is no LMC assigned for maternity care, the primary maternity carer or health provider or the secondary/tertiary service is responsible for the screening process.

2. Lead Maternity Carer programme responsibilities

2.1 Purpose

This policy describes LMC responsibilities in respect of the NMSP.

2.2 Background

LMCs have responsibilities for provision of maternity services under the notice pursuant to section 88 of the New Zealand Public Health and Disability Act 2000 which includes provision of services within the Newborn Metabolic Screening Programme.

2.3 Requirements

- (a) LMCs and others providing NMSP services must comply with all legislative and government requirements including but not limited to the:
 - (i) Primary Maternity Services Notice 2007 (the Notice) issued pursuant to Section 88 of the New Zealand Public Health and Disability Act 2000
 - (ii) relevant indicators within the *NMSP Monitoring Framework*
 - (iii) Privacy Act 1993 and the Health Information Privacy Code 1994
 - (iv) Health and Disability Commissioner's Act 1994.
- (b) Within the NMSP, LMCs are responsible for:
 - (i) providing information and education to parents regarding the NMSP including the storage and possible uses of blood spot samples
 - (ii) offering newborn metabolic screening to the parents/guardians of all babies in their care
 - (iii) discussing the return or storage of left over blood spots including possible uses
 - (iv) obtaining informed consent for newborn metabolic screening
 - (v) obtaining informed consent for either return or storage of leftover blood spots
 - (vi) ensuring that where parents request the return of blood spots, written information is provided with the blood spot card when it is sent for testing
 - (vii) ensuring that the screening procedure is carried out according to section 7 of the *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme* (2010) either by themselves, or via referral to another trained provider

- (viii) ensuring that maternal, newborn and practitioner information which is required on the newborn metabolic screening blood spot card is complete and correct for all babies having screening
- (ix) ensuring that all samples collected are complete, satisfactory for testing and posted to the laboratory within 24 hours of collection
- (x) ensuring the receipt of a screening result from the laboratory and communication of results to parents/guardians for each screened baby under their care
- (xi) ensuring that all babies whose parents/guardians have consented to their screening have completed all aspects of metabolic screening in a timely manner which meets the standards within the NMSP Monitoring Framework and with the goal of completion of screening by four weeks of age.⁵ This includes practitioners responding appropriately and as requested to all abnormal results or requests for re-sampling, notified by the laboratory
- (xii) communicating with the laboratory as required regarding requests for second samples, positive results or other follow-up requirements to ensure completion of screening
- (xiii) ensuring documentation in the clinical notes, well child book and handover notes are completed. This includes the details of NMSP consent/decline, sampling, results and follow-up.

3. Guidelines

- 3.1 The *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme* (2010)⁶ identify best practice in newborn metabolic screening and provide supporting information regarding conditions screened for. LMCs and all other providers offering services within this programme should be familiar with the provisions contained within this document and carry out services as recommended.

4. Resources

- 4.1 LMCs will be provided with the following resources:
- (a) blood spot cards
 - (b) DVDs
 - (c) parent pamphlets
 - (d) disorder specific resources
 - (e) envelopes and postage
 - (f) lancets
 - (g) where to source additional material or information.

⁵ It is recognised that small numbers of newborns may not have completed screening by four weeks of age. These instances should be documented as to reasons why. An appropriate reason may be low birth weight babies who require more than one sample over a period of time.

⁶ Found at www.nsu.govt.nz

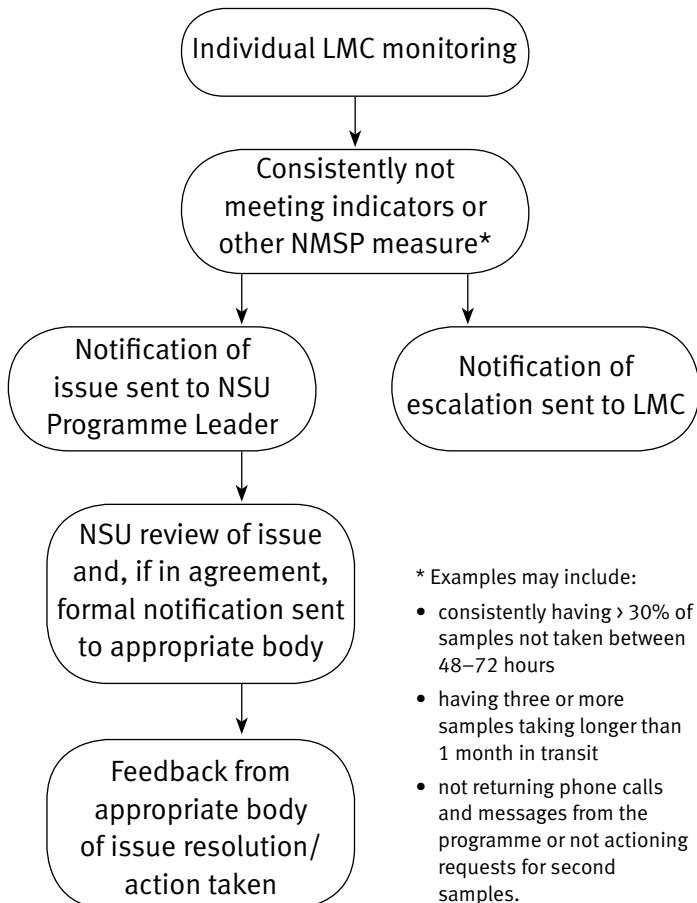
5. Continuing education

- 5.1 Online resources and educational material have been developed to support best practice of NMSP practitioners. These resources can be found at www.nsu.govt.nz
- 5.2 LMCs and all providers offering services within this programme are encouraged to familiarise themselves with these documents and attain the professional development/CME points attached to the e-Learning modules.

6. Monitoring

- 6.1 All LMC service delivery for the NMSP will be monitored. This includes monitoring against the indicators included in the NMSP Monitoring Framework. LMCs are expected to meet the appropriate requirements within the NMSP Monitoring Framework including indicators 2, 3, 4 and 7. LMCs that are continuously not meeting the indicators will be contacted and referred to the relevant professional regulatory body as per the escalation pathway in Figure 4 below.

Figure 4: Escalation pathway for LMC issues



* Examples may include:

- consistently having > 30% of samples not taken between 48–72 hours
- having three or more samples taking longer than 1 month in transit
- not returning phone calls and messages from the programme or not actioning requests for second samples.

Section Five – Laboratory Responsibilities

1. Laboratory responsibilities

1.1 This section describes specific requirements for laboratory practitioners. Laboratory practitioners must also comply with all other applicable provisions in this policy document.

1.2 Background

The NSU contracts with a DHB to provide laboratory screening services. This includes the resources required for blood sampling, the receipt, data entry, testing and reporting of samples and informing practitioners of results and follow-up requirements. The DHB also has responsibilities for data collection, monitoring and reporting of the NMSP.

2. Laboratory service responsibilities

2.1 Purpose

This policy describes the services contracted by the NSU from the laboratory.

2.2 Background

The Ministry of Health currently funds Auckland DHB to provide laboratory services for the NMSP through a contract managed by the NSU.

2.3 Requirements

Screening services include:

- (a) laboratory blood testing services to screen for specified metabolic disorders:
 - (i) receiving and registering all newborn blood spot samples
 - (ii) providing an information system to support the screening programme
 - (iii) testing and analysis of the blood samples for specific metabolic disorders using the presence and levels of different biochemical markers, using the cutoffs specified in Appendix B
- (b) supply and distribution of blood spot cards, envelopes and lancets to referring practitioners
- (c) liaison with referring practitioners
 - (i) reporting results to referring practitioners
 - (ii) assisting with referrals and advice for positive results
 - (iii) provision of specialist advice and consultation services
- (d) management of blood spot cards
 - (i) retention, secure storage, monitored use, release and tracking of tested blood spot cards
 - (ii) returning blood spot cards to parents/guardians or individuals by tracked courier within 28 days of request
- (e) provision of administration, quality assurance, and management services to support the effective, efficient and timely delivery of the contracted laboratory services
- (f) documentation of each screening event, including follow-up if required and completion of case audit forms in a timely manner

- (g) quality assurance, data provision, and reporting to the NSU as prescribed in the contract
- (h) development and documentation of protocols and processes relating to screening services
- (i) supply and distribution of resources to referring practitioners
- (j) provision of laboratory blood testing services for post-diagnosis monitoring of metabolic disorders
- (k) development and maintenance of a formal working relationship with the clinical metabolic treatment services, including communication of results, protocol development and test analysis
- (l) maintenance of an up to date database of LMC and specialist contact details.

3. Accreditation, peer review and quality assurance

3.1 Purpose

This policy sets out the requirements for accreditation, peer review, and quality assurance of the contracted laboratory.

3.2 Requirements

The laboratory must be appropriately accredited and peer-reviewed, including:

- (a) holding current accreditation against ISO 15189:2007 (Medical Laboratories – Particular Requirements for Quality and Competence)
- (b) being peer-reviewed by external assessors every two years as part of ISO 15189:2007 accreditation
- (c) complying with:
 - (i) all legislative and regulatory requirements relating to quality and service standards in respect of the provision of laboratory services for newborn metabolic screening
 - (ii) this policy framework
 - (iii) the NZS/ISO 15189:2007 and AS/NZS 2243
 - (iv) the IANZ Specific Criteria for Accreditation – Medical Testing – Draft 9, including having IANZ accreditation and registration
 - (v) the Health Records Standard NZS 8153:2002
 - (vi) the Code of Practice for Information Security Management AS/NZS ISO/IEC 17799:2001
 - (vii) the Health Network Code of Practice (SNZ HB 8169:2002)
 - (viii) the Health and Disability Sector Standards (NZS 8134:2001)
 - (ix) any other policy, quality and service standards or other requirements that apply to the provision of NMSP services.
- (d) The laboratory must have policies and practices that ensure the quality of NMSP testing and reporting. Policies must define staff responsibilities and laboratory procedures.

- (e) The laboratory must ensure that:
 - (i) an annual internal review of the service is completed by an appropriately qualified person
 - (ii) regular internal assessment of laboratory functions, including test sensitivity, specificity, positive predictive value and timeliness of reporting are undertaken
 - (iii) staff achieve and maintain competency in the tasks they perform
 - iv) satisfactory internal systems for internal control and quality improvement are in place, including monitoring of data entry.
- (f) The laboratory is expected to participate in external quality assurance including the Centre for Disease Control (CDC) quality control programme and use reports to improve their own quality control processes.

4. Staffing requirements

4.1 Purpose

This policy describes the general staffing requirements for the laboratory providing the contracted newborn metabolic screening services.

4.2 Requirements

- (a) The laboratory service must be staffed by suitably qualified scientists and technicians and professionally led by a suitably qualified director. Identified individuals must fill the mandatory leadership positions outlined in this section and be allocated sufficient time to fulfil the roles.
- (b) The laboratory must:
 - (i) ensure all staff are competent pursuant to the Health Practitioners Competency Assurance Act 2003
 - (ii) have a documented management structure with clear delineation of responsibilities of medical and non-medical staff
 - (iii) have sufficient levels of staffing to provide a high-quality newborn metabolic screening service
 - (iv) have a succession plan for all mandatory positions
 - (v) have appropriately qualified staff available to advise referring practitioners and clinicians on screening results, including advice on the quality of sample taking, referral recommendations, and result interpretation.

4.3 Mandatory positions

- (a) Newborn metabolic screening is a team activity and success depends on leadership and teamwork for all participants. It also relies on clear and accountable responsibilities for all aspects of the screening pathway. The programme relies on the support and administration of the laboratory and on the organisation's contractual obligations. Identified individuals must fill the mandatory positions outlined in this section. The positions identified here will require a clear allocation of time to fulfil their roles and have a specific job description that describes their role and individual responsibilities. All individuals must fulfil the qualifications and continuing professional development requirements of the relevant profession and/or position within the programme.

- (a) Mandatory positions are:
- (i) NMSP director
 - (ii) metabolic physician
 - (iii) LMC educator
 - (iv) lead scientist
 - (v) tandem mass spectrometer scientist
 - (vi) data manager
 - (vii) quality coordinator
 - (viii) NMSP follow-up nurse.

4.4 NMSP director requirements

- (a) The laboratory must employ an NMSP director.
- (b) This person is responsible for leadership of the services provided in respect of the NMSP.
- (c) The laboratory must ensure that the director is suitably qualified, competent, and capable of meeting the requirements of the role.
- (d) The laboratory must ensure that the director:
 - (i) is clinically qualified (MBChB or equivalent, American Board Certified in Biochemical Genetics, HGSA fellowship in Biochemical Genetics, or equivalent)
 - (ii) holds a current annual practising certificate issued by the Medical Council of New Zealand (or equivalent), with a scope of practice in clinical genetics or equivalent
 - (iii) has specialist knowledge of congenital metabolic diseases
 - (iv) has an in-depth understanding of screening and screening programmes including uptake, testing, audit, monitoring, evaluation, quality assurance, follow-up and reporting
 - (v) is registered to practise in New Zealand
 - (vi) is a regular attendee at appropriate internal and external meetings
 - (vii) monitors international developments in newborn screening and communicates these to other NMSP staff
 - (viii) shows evidence that they have continuing professional development in the area of newborn metabolic screening
 - (ix) has regular communication with the NSU Programme Leader
 - (x) builds relationships with other health professionals both national and international.

4.5 Metabolic physician requirements

- (a) The laboratory must employ a metabolic physician to provide interpretation, advice, treatment and documentation development for the programme.
- (b) The laboratory must ensure that the metabolic physician is suitably qualified, competent, and capable of meeting the requirements of the role.

- (c) The laboratory must ensure that the metabolic physician:
- (i) is a point of contact for referring practitioners
 - (ii) is clinically qualified (MBChB or equivalent, American Board Certified in Biochemical Genetics, HGSA fellowship in Biochemical Genetics or equivalent)
 - (iii) has specialist knowledge of congenital metabolic diseases
 - (iv) is registered to practise in New Zealand
 - (v) holds a current annual practising certificate issued by the Medical Council of New Zealand, with a scope of practice in clinical genetics/metabolic diseases
 - (vi) is a regular attendee at appropriate internal and external meetings
 - (vii) shows evidence that they have continuing professional development in the area of newborn metabolic screening
 - (viii) builds relationships with other health professionals both national and international.

4.6 LMC educator requirements

- (a) The laboratory must employ an LMC educator to provide programme guidance and leadership to LMCs providing services under the N MSP.
- (b) The laboratory must ensure that the LMC educator is suitably qualified, competent, and capable of meeting the requirements of the role.
- (c) The laboratory must ensure that the LMC educator:
- (i) is clinically qualified with an appropriate midwifery qualification
 - (ii) is registered to practise in New Zealand
 - (iii) holds a current annual practising certificate issued by the Midwifery Council of New Zealand, with a scope of practice in midwifery
 - (iv) has participated in newborn metabolic screening for a minimum of three years
 - (v) demonstrates an understanding of screening and screening programmes
 - (vi) shows evidence that they have continuing professional development in the area of newborn metabolic screening
 - (vii) has regular contact with NSU personnel, specifically the Education and Training Leader and N MSP Programme Leader.

4.7 Lead scientist requirements

- (a) The laboratory must employ a lead scientist to ensure scientific leadership and responsibility within the laboratory.
- (b) The laboratory must ensure that the lead scientist is suitably qualified, competent, and capable of meeting the requirements of the role.
- (c) The laboratory must ensure that the lead scientist:
- (i) is scientifically qualified
 - (ii) is registered to practise in New Zealand

- (iii) holds a current annual practising certificate issued by the New Zealand Medical Laboratory Science Board, with a scope of practice of medical laboratory scientist with subspecialty training in newborn metabolic screening
- (iv) has participated in newborn screening for a minimum of three years
- (v) demonstrates an understanding of screening and screening programmes
- (vi) shows evidence that they have continuing professional development in the area of newborn metabolic screening.

4.8 Tandem mass spectrometer scientist requirements

- (a) The laboratory must employ a scientist with appropriate experience and expertise to run the tandem mass spectrometer.
- (b) The laboratory must ensure that the tandem mass spectrometer scientist is suitably qualified, competent, and capable of meeting the requirements of the role.
- (c) The laboratory must ensure that the tandem mass spectrometer scientist:
 - (i) is scientifically qualified
 - (ii) is registered to practise in New Zealand
 - (iii) holds a current annual practising certificate issued by the New Zealand Medical Laboratory Science Board, with a scope of practice of medical laboratory scientist with subspecialty training in tandem mass spectrometry
 - (iv) has participated in newborn screening for a minimum of three years
 - (v) shows evidence that they have continuing professional development in the area of newborn metabolic screening.

4.9 Data manager requirements

- (a) The laboratory must employ a NMSP data manager to be responsible for the overall management, quality, security, integrity, and availability of data collected on behalf of the Programme.
- (b) The laboratory must ensure that the data manager is suitably qualified, competent, and capable of meeting the requirements of the role.
- (c) The laboratory must ensure that the data manager:
 - (i) has appropriate qualifications and experience in data management, including report generation
 - (ii) has the appropriate knowledge and adequate training in the information system/ database which captures and stores the information
 - (iii) has a minimum of two years experience managing a ‘business critical’ information system
 - (iv) demonstrates an understanding of audit, monitoring and evaluation requirements for the programme
 - (v) demonstrates an understanding of data backup and retrieval
 - (vi) has experience in managing data quality.

4.10 Quality coordinator requirements

- (a) The laboratory must employ a NMSP quality coordinator to be responsible for the overall coordination of quality activities of the Programme.
- (b) The laboratory must ensure that the quality coordinator is suitably qualified, competent, and capable of meeting the requirements of the role.
- (c) The laboratory must ensure that the quality coordinator:
 - (i) has appropriate qualifications, training and experience in laboratory quality assurance, including internal and external quality assurance activities
 - (ii) has a minimum of two years experience working in a laboratory
 - (iii) has appropriate knowledge of quality control and quality improvement including assay development and technical testing and reporting requirements
 - (iv) demonstrates an understanding of quality standards, audit and the monitoring and evaluation requirements of the NMSP
 - (v) demonstrates an understanding of data quality requirements for the NMSP.

4.11 NMSP follow-up nurse requirements

- (a) The laboratory must employ an NMSP follow-up nurse to provide administrative support for the leadership team to support case follow-up and review.
- (b) The laboratory must ensure that the NMSP follow-up nurse is suitably qualified, competent, and capable of meeting the requirements of the role.
- (c) The laboratory must ensure that the NMSP follow-up nurse:
 - (i) is clinically qualified with an appropriate nursing qualification
 - (ii) is registered to practise in New Zealand
 - (iii) holds a current annual practising certificate issued by the Nursing Council of New Zealand, with a scope of practice as a registered nurse
 - (iv) has received appropriate orientation and training to meet the requirements of the role
 - (v) demonstrates an understanding of screening and the NMSP
 - (vi) shows evidence that they have continuing professional development in the area of newborn metabolic screening.

5. Staff continuing education

5.1 Purpose

This policy describes the requirements for continuing education to be provided to staff by the laboratory.

5.2 Requirements

- (a) The laboratory must provide continuing education for all laboratory staff involved in the newborn metabolic screening programme, including:
 - (i) documented attendance at an internal and/or external teaching programme in newborn metabolic screening

- (ii) formal update courses every three years
 - (iii) provision of current editions of major standard texts and current issues of journals relevant to newborn metabolic screening within the laboratory
 - (iv) access to relevant local and international professional meetings regularly.
- (b) The laboratory must maintain an up-to-date record of individual staff members' participation in continuing education.

6. Staff communications

6.1 Purpose

This policy describes the requirements for communication with referring practitioners and parents/guardians/individuals who contact the laboratory.

6.2 Requirements

The laboratory must:

- (a) ensure that during laboratory hours, identified and appropriately qualified staff are available to provide advice to referring practitioners regarding test results, follow-up and early intervention
- (b) have a documented policy that is promulgated to staff on how to manage electronic, telephone, and personal contact with parents/guardians/individuals who contact the laboratory. This policy must include:
 - (i) the principles of respect, sensitivity and cultural appropriateness
 - (ii) the types of information that can be provided to parents/guardians and by whom
- (c) provide a private area for staff to make and receive telephone calls to and from parents/guardians, LMCs and other practitioners.

7. Workloads

7.1 Purpose

This policy describes the requirements to ensure that there is sufficient staff to handle the volume of samples referred to the laboratory for testing and reporting.

7.2 Requirements

The laboratory must:

- (a) ensure that individual scientific and technical staff's workload is appropriate to their level of skill and experience
- (b) monitor the workload of all staff to prevent work overload while ensuring competency and maintenance of skill
- (c) benchmark staff numbers and volumes with Australian and international laboratories who deliver newborn metabolic screening services
- (d) meet the requirements within the NMSP Monitoring Framework.

8. Equipment

8.1 Purpose

This policy describes the criteria for review of testing capabilities of laboratory equipment.

8.2 Requirements

- (a) The laboratory must review testing capabilities at regular intervals, including:
 - (i) reviewing cut-off levels for screened disorders a minimum of every five years, and more regularly if evidence indicates that optimum cut-off levels may have changed
 - (ii) reviewing testing equipment for screened disorders a minimum of every five years as part of a maintenance programme, and more regularly if evidence indicates that optimum testing is not being achieved
 - (iii) reviewing equipment and information technology to inform future recommendations
- (b) The laboratory must have:
 - (i) a detailed capital plan for replacement of equipment
 - (ii) a detailed contingency plan for all screening equipment that ensures that screening can continue uninterrupted in the event of any equipment failure.

8.3 Process

Following any review the laboratory may submit a written request to the NSU to make a change to the laboratory processes utilised in the NMSP. Requests will be reviewed by the NMSP Governance Team and Technical Group and are subject to confirmation by the NSU.

9. Receipt and coding of blood spot samples

9.1 Purpose

This policy describes the criteria for receipt and coding of blood spot samples by the laboratory.

9.2 Requirements

The laboratory must:

- (a) have detailed protocols regarding sample receipt, registration and coding of samples
- (b) collect samples from its mail box every day at 0730 hours and ensure delivery of the samples directly to the laboratory
- (c) ensure sample registration is performed on the day of receipt of the sample by appropriately trained individuals who are within restricted public access
- (d) provide a fail-safe process for correct identification of samples, bar coding, validation and transcription into information systems
- (e) manage specimens which are inadequately labelled, have incorrect data or are lacking information or blood
- (f) manage exceptions (repeat samples, diagnostic and monitoring specimens or any other specimen that is not a first sample) to ensure that exceptions are identified, processed, and tested according to clinical urgency
- (g) fast-track urgent specimens

- (h) ensure samples collected from the mailbox at 0730 are tested on the same day
- (i) ensure that second and subsequent specimens are linked within the information system with the first sample.

10. Testing of blood spot samples

10.1 Purpose

This policy describes the laboratory requirements for receipt and testing blood spot samples.

10.2 Requirements

- (a) The laboratory must have detailed written protocols in place for all testing requirements including relating to reagents used, filter paper specifications, blood spot punching, suitability of samples, analysis requirements and report generation.
- (b) The laboratory must test all blood spot samples received for the disorders specified in Appendix B:
- (i) within the timeframes in Appendix B, as per Indicator 5 of the NMSM Monitoring Framework
 - (ii) using the cut-off levels set out in Appendix B
 - (iii) including all inadequate or unsuitable samples to the best of its ability
- (c) The laboratory must have failsafe mechanisms that ensure repeat samples are requested and monitored.

11. Reporting results

11.1 Purpose

This policy describes how blood spot sample testing results must be reported.

11.2 Background

There are three possible screening results.

- (a) A negative screening result means that biochemical results are within the normal range and the baby has a very low risk of having one of the disorders screened for.
- (b) An unsuitable sample result means that the laboratory was unable to accurately test the sample. A repeat sample must be provided so that the baby can be screened.
- (c) A positive screening result includes slightly abnormal or significantly abnormal results, both of which will require active follow-up as per the *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand (2010)*.

11.3 Requirements

The laboratory must:

- (a) have a detailed written process for reporting results including sign out of reports by appropriately qualified personnel
- (b) provide a report for each sample tested⁷

⁷ Currently a hard copy report is provided to the LMC although over time this may change to electronic.

- (c) directly report all results to referring practitioners within the timeframes specified in the Newborn Metabolic Monitoring Framework
- (d) report on disorders based on the disorder definitions in Appendix B
- (e) ensure that reports to referring practitioners include
 - (i) the programme name clearly visible on the top of the first page of the report
 - (ii) the timing of the tests
 - (iii) transit times
 - (iv) the outcomes of testing
 - (v) any repeat sample requirements
 - (vi) any follow-up requirements
 - (vii) audit requirements, including data collection from referring practitioners on the outcome of diagnostic testing
- (f) advise referring practitioners and clinicians on screening results, including advice on the quality of sample taking, need for a repeat or further sample, result interpretation and referral recommendations
- (g) follow the referral pathway specific to each disorder or group of disorders
- (h) document all screening results and follow-up
- (i) assess, monitor and report false positive screening results to improve screening efficiency.

12. Follow-up of results

12.1 Purpose

This policy describes the requirements for follow-up through further testing and other actions where samples are unsuitable or have produced a positive result. The goal is to ensure completion of screening for all babies within four weeks of birth.⁸

12.2 Background

- (a) The laboratory must design procedures to achieve completion of follow-up and begin intervention prior to the occurrence of morbidity or mortality associated with the disorder.
- (b) Follow-up depends on the level of the result for a specific disorder. Each disorder has a defined process that must be followed.
- (c) The details of each disorder can be found in the *Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand* (2010).

⁸ It is recognised that small numbers of newborns may not have completed screening by four weeks of age. These instances should be documented as to reasons why. An appropriate reason may be low birth weight babies who require more than one sample over a period of time.

12.3 Follow-up requirements

The laboratory must:

- (a) have a written process for the follow-up of unsuitable and positive results showing clear, concise actions and contain timed follow-up which meet the requirements of this policy, including the goal above
- (b) have separate processes for unsuitable samples, slightly positive and significantly positive samples. These processes must reflect the clinical requirements (ie significantly positive results must be phoned to the LMC or, if required, contact made with another practitioner the same day the result is reported)
- (c) actively monitor and document all follow-up activities taken until case resolution
- (d) have a process for escalation of result notification if no response from the LMC or other practitioner
- (e) have a process for second sample requirements with the aim of completing screening in a timely manner
- (f) develop and maintain written procedures that define reasonable efforts to locate newborns who have not completed screening
- (g) provide education materials and appropriate advice to LMCs
- (h) send audit forms as required to practitioners, ensure they are completed and entered into the appropriate information system
- (i) have a protocol for cases lost to follow-up which includes case review to determine factors contributing to the loss and whether changes in follow-up practices are necessary
- (j) develop and maintain a quality assurance plan that assesses follow-up processes and procedures using performance indicators
- (k) maintain an information system which assists with identifying those cases requiring follow-up
- (l) report to the NSU as per contract requirements regarding follow-up monitoring and evaluation.

13. Data management information systems

13.1 Purpose

This policy sets out the requirements for the NMSP laboratory information systems.

13.2 Background

The laboratory is responsible for maintaining efficient and effective information systems and process/procedures for the management of patient information and the management of the Programme as a whole. This includes the areas of data collection, data storage, query, reporting and budgeting.

13.3 Data collection requirements

The laboratory must ensure that:

- (a) appropriate data is captured and maintained to support the clinical and operational aspects of the screening programme covering all aspects of the pathway and to meet monitoring, evaluation and audit requirements
- (b) data is captured in a complete, timely and accurate manner

- (c) checks are implemented for errors that may arise during data entry
- (d) data is validated against business rules through the application of edit criteria
- (e) data is accurate and complete prior to use for reporting
- (f) definitions and edit rules are understood and are being followed
- (g) inconsistencies are followed up and rectified
- (h) the information systems uses the National Health Index (NHI) as the unique identifier for newborns
- (i) the systems used for recall and follow-up are fail-safe
- (j) there are robust data backup procedures for all NMSP information systems
- (k) the transfer of data must be as secure as possible
- (l) all information updates have an audit trail which identifies the change, author and date
- (m) data relating to screening tests is held indefinitely
- (n) data in information systems is held separately from blood spot card storage.

13.4 Protocols

The laboratory is required to have clear data protocols that cover data set requirements, data entry, modification, validation, access, data backup and quality control.

13.5 Compliance with policy and legislation

- (a) All NMSP information systems must comply with legislative requirements, government policy and new policy principles where appropriate.
- (b) Of particular note, the laboratory will ensure that staff are familiar with the requirements of:
 - (i) The Privacy Act 1993
 - (ii) The Health Information Privacy Code 1994
 - (iii) Health and Disability Commissioner's Act 1994; and that the requirements of these documents are complied with in every respect in the collection and transfer of health information.
- (c) The laboratory must comply with retention and disposal requirements of the Public Records Act 2005.
- (d) The laboratory must also conform to the guiding principles of data collection and management as found in the 'Guide to NZHIS National Collections'. The guiding principles include:
 - (i) the need to protect patient confidentiality and privacy
 - (ii) the need to collect data once, as close to the source as possible, and use it as many times as required to meet different information requirements, in keeping with the purpose for which it was collected
 - (iii) the need for standard data definitions, classifications and coding systems
 - (iv) the requirement for national health data to include only that data which is used, valued and validated at the local level

- (v) the need for connectivity between health information systems to promote communication and integrity
- (vi) the need to address Māori health disparities.

13.6 Training

The laboratory must ensure:

- (a) that all staff with access to the information system have adequate training and appropriate documentation to allow them to correctly use the information systems
- (b) all staff with access to the information systems receive adequate training on their obligations in relation to privacy, accessibility and confidentiality of data.

13.7 Security and protection

The laboratory is responsible for implementing reasonable and sufficient security measures to ensure:

- (a) the maintenance of an access register which details job titles and access provisions
- (b) participant privacy and confidentiality
- (c) data availability
- (d) data integrity
- (e) data is not subject to loss, corruption, destruction, inappropriate alteration or miscalculation
- (f) ongoing security and protection provisions are effective and continue to be adequate in the context of possible threats
- (g) only appropriately authorised and trained individuals have access to systems accepting data entry and data update.

13.8 Data monitoring

- (a) The programme will be monitored to ensure it meets the programme aim and objectives.
- (a) The laboratory must ensure the:
 - (i) timely provision of quarterly reports to the NSU as specified within the NMSP Monitoring Framework
 - (ii) timely provision of quarterly and annual data and reports to the NSU as specified within the NMSP contract
 - (iii) timely provision of data to the NSU required for national monitoring and audit and evaluation purposes
 - (iv) generation and analysis of reports necessary to ensure ongoing internal audit and quality improvement of operational and business practices.

Section Six – Return of Residual Blood Spots

1. Return of residual blood spot samples to parents/guardians and individuals

- 1.1 This section describes specific requirements for requesting and returning residual blood spot samples to parents/guardians and individuals. The policy covers the return of residual blood spot samples:
 - (a) immediately after screening
 - (b) held in secure storage.

- 1.2 **Background**
 - (a) At the time of screening, parents/guardians consent to either return of the residual blood spot sample or for it to be held in indefinite storage by the programme for possible future uses. The Ministry of Health is the guardian of those blood spot samples held by the NMSP and consider protection of the cards from unauthorised access to be paramount. The Ministry of Health will ensure that all due care is taken in the handling and storage of the samples.
 - (b) The laboratory is responsible for returning residual blood spot samples when parents/guardians or individuals request them (representative). The representative may therefore include the individual, parent, guardian or legal representative. Evidence of appropriate authority may be required.
 - (c) For samples taken after 1998, the laboratory will return the residual blood spot sample which is separated from the demographic information at the time of screening. For samples taken prior to 1998, the residual blood spot sample is not separated from the demographic information and the whole card will be returned.

2. Release of residual blood spot samples to parents/guardians and individuals

2.1 Background

- (a) Retention of newborn blood spots is governed by the Code of Health and Disability Services Consumers' Rights Regulation 1996.
- (b) Right 7(9) states consumers have the right to make a decision about the return or disposal of body parts or substances removed in the course of a health care procedure.
- (c) Parents/guardians or individuals are entitled to request the return of residual blood spot samples at any time after screening.

3. Requirements – residual blood spot samples requested for return to parents/guardians at time of screening

- 3.1 At the time of screening, parents/guardians consent to either return of the residual blood spot sample or for it to be held in indefinite storage by the programme for possible future uses. At this time, the LMC is authenticating the representative entitled to the return of the residual blood spot sample (parent/guardian).

3.2 Process

- (a) When completing the blood spot card information, the LMC must attach a written request for the residual blood spot sample to be returned to parents/guardians when testing is complete.
- (b) While it is preferred that the written request be on the ‘Return of Newborn Metabolic Screening Samples’ form (the ‘NMSP return form’) a written communication may be used but must include the name, signature, address and the relationship of the person requesting the residual blood spot sample. If the person requesting the residual blood spot sample is not the mother or father, evidence of identity and guardianship is required.
- (c) The laboratory must assess and authorise each request for return of residual blood spot samples to ensure all requirements in 1 and 2 above are met.
- (d) If information is not complete the laboratory must inform the LMC.
- (e) The laboratory must only return residual blood spot samples to authorised parents or guardians.
- (f) The laboratory must send blood spot samples by tracked courier to the parent or guardian within 20 working days of completion of screening.

4. Requirements – residual blood spot samples requested from storage by a parent/guardian or individual

- 4.1 At any time, residual blood spot samples may be requested from storage by using the NMSP return form. The processes below must be followed.

4.2 Process

- (a) An individual, parent or guardian must complete the NMSP return form and comply with the requirements of evidence as stated on the form.
- (b) If an individual is requesting the return of their own blood spot sample and he/she is aged 16 years and over, only that individual (or their legal guardian) can request the return of their blood spot sample.
- (c) The laboratory must ensure all required sections on the form are completed. If all required information is not completed, the laboratory must inform the requestor.
- (d) The laboratory must assess and authorise each request for return and only return blood spot samples to authorised parents/guardians or individuals.
- (e) The laboratory must send blood spot samples by tracked courier to the individual/parent/guardian within 20 working days of a completed and authorised sample request.

5. Requirements – residual blood spot samples requested from storage for a deceased person

5.1 At any time, residual blood spot samples may be requested from storage by using the NMSP return form. The processes below must be followed.

5.2 Process

- (a) A person who has authority (close available relative) under the Human Tissue Act 2008 may request the return of a deceased person's blood spot sample.
- (b) The person who has authority must complete the NMSP return form and comply with the requirements of evidence as stated on the form.
- (c) The laboratory must ensure all required sections on the form are completed. If all required information is not completed, the laboratory must inform the requestor.
- (d) The laboratory must assess and authorise each request for return and only return blood spot samples to authorised persons.
- (e) The laboratory must send blood spot samples by tracked courier to the requestor within 20 working days of a completed and authorised sample request.

Section Seven – Storage and Uses of Residual Blood Spot Samples

1. Storage and uses of residual blood spot samples

- 1.1 This section covers the storage and uses of residual blood spot samples and includes specific requirements for storing, requesting and returning residual blood spot samples from storage.
- 1.2 **Background**
- (a) The Newborn Metabolic Screening Programme (NMSP) has been in operation since 1969. Since this time, blood spot samples have been either returned to parents/guardians or individuals or held indefinitely in secure storage by the programme.
 - (b) Because residual newborn blood spot samples contain only a limited amount of blood, their further use after screening has to be prioritised to ensure that enough blood is left to serve the most important purposes of the NMSP and for the benefit of babies and their family/whānau.
 - (c) Following screening, some blood spot samples may have residual material. Given that the residual material is limited, this policy outlines the priority for use (primary and secondary uses).

2. Governing provisions

- 2.1 The governing provisions for the NMSP and the blood spot samples are as follows.
- (a) The NMSP requires consent for both taking a heel prick sample to test for specific disorders and for either returning or storing the blood spot sample. If consent has been given for storage, blood spot samples are stored in a secure facility with restricted access. Further information and requirements for storage are detailed in Section 7.
 - (b) Residual blood spot samples are subject to protection as per Table 1 below.
 - (c) Legislative/regulatory protection and guidance for storage and use of residual blood spot samples is provided by the:
 - (i) Code of Health and Disability Services Consumers' Rights Regulation 1996 (Code of Rights)
 - (ii) Care of Children Act 2004
 - (iii) Health Information Privacy Code 1994
 - (iv) Health (Retention of Health Information) Regulations 1996
 - (v) New Zealand Public Health and Disability Act 2000
 - (vi) Public Records Act 2005
 - (vii) Human Tissue Act 2008
 - (viii) New Zealand Standard NZS 8135:2009 Non-therapeutic use of human tissue
 - (ix) Operational Standards for Ethics Committees
 - (x) *Guidelines for the Use of Human Tissue for Future Unspecified Research Purposes*

- (xi) Memorandum of Understanding between Ministry of Health and New Zealand Police
- (xii) Coroners Act 2006.

Table 1: Governing provisions

Blood spot sample/card storage and use	Relevant legislation/regulations and other requirements
Consent for storage or use of blood spot samples	<p>Covered by:</p> <ol style="list-style-type: none"> 1. The Code of Health and Disability Services Consumers' Rights (Right 6 and 7) requires that information must be provided and consent must be obtained. 2. Care of Children Act 2004 (Section 36(3)) provides that a parent/guardian may give consent to screening on behalf of their baby. 3. Right 7(10) of the Code of Rights which requires providers to obtain consent for the storage or use of newborn blood spots. Providers must therefore inform consumers what the blood spots may be used for as per the: <ul style="list-style-type: none"> • Guidelines for practitioners providing services within the Newborn Metabolic Screening Programme in New Zealand • Consumer pamphlet: <i>Your newborn baby's blood test</i> • <i>Newborn Metabolic Screening Programme Policy Framework 2011</i> (this document).
Protection of information held on blood spot sample cards	<p>Health Information Privacy Code 1994 and Health (Retention of Health Information) Regulations 1996.</p> <p>The Public Records Act 2005 has precedence over the regulations – Section 21, 26.</p>
The right to have the blood spot sample returned	Right 7(9) of the Code of Rights: consumers have the right to make a decision about the return or disposal of body parts or substances removed in the course of a health care procedure.
Long-term storage	<p>Public Records Act 2005 (requirements over 25 years)</p> <p>Human Tissue Act 2008</p> <p>New Zealand Standard NZS 8135:2009 Non-therapeutic use of human tissue</p>
Secondary uses of residual blood spots	<p>Code of Rights 7(10) (a) and (b)</p> <p>Guidelines for the Use of Human Tissue for Future Unspecified Research Purposes</p> <p>Memorandum of Understanding between Ministry of Health and New Zealand Police</p> <p>Coroners Act 2006</p> <p>New Zealand Public Health and Disability Act 2000 (Schedule 5 Mortality review committee)</p>

3. Primary uses

3.1 Purpose

The consent for performing a heel prick test and testing the blood spot sample for specific diseases covers the requirements for primary uses. Primary uses include ensuring a high quality screening programme.

3.2 Requirements

- (a) Blood spot samples are collected for the primary purpose of screening newborns for specific metabolic disorders. The primary uses encompass:
 - (i) the initial screening test
 - (ii) repeat confirmatory testing (including testing missed cases)
 - (iii) investigation of initial screening test results that may have been a false positive or false negative
 - (iv) quality assurance and audit of the programme
 - (v) assay improvement and validation of tests for disorders currently in the programme
 - (vi) validation of assays for potential new disorders to be added to the newborn screening panel.
- (b) Use for assay improvement and validation of tests for disorders currently in the NMSP must be presented to the NMSP Technical Group and/or NMSP Governance Team and authorised by the Ministry of Health (Ministry).
- (c) Use for assay validation of tests for potential new disorders must be presented to the NMSP Technical Group and/or NMSP Governance Team and authorised by the Ministry.
- (d) The laboratory must report on residual blood spot samples used for primary uses as per contractual requirements.
- (e) Residual blood spot samples retrieved from storage for primary use are subject to ‘Storage Requirements’ in Clause 13.
- (f) Residual blood spot samples must be placed in or returned to storage after primary use unless requested to be returned to parents/guardians.

4. Secondary uses

4.1 Purpose

- (a) Following screening, parents/guardians either consent to return of the residual blood spot sample or consent to storage by the NMSP. Those held in secure storage may have a number of authorised secondary uses. This policy details the authorised secondary uses, access and security requirements for blood spot samples and associated information.
- (b) Authorised secondary uses (with the requirements and limitations set out in this policy) are:
 - (i) those that benefit the individual and his/her family/whānau (for instance further blood testing not covered by the original consent) (Page 43)
 - (ii) forensic/police investigations (Page 43)
 - (iii) coroner investigations (Page 44)

- (iv) mortality review (Page 45)
 - (v) research (Page 46)
 - (vi) other requests (Page 49).
- (c) Where blood spot samples are sent to other laboratories for testing, any residual blood spot sample remaining after testing is complete must be returned to the NMSP.

5. Release of residual blood spot samples for the benefit of the individual and/or family/whānau

5.1 Background

Releases for the benefit of the individual and/or family/whānau may include further testing or repeat testing on residual blood spot samples not covered by the original consent. This may include testing the residual blood for disorders/diseases that may be a cause of death or disease. Requests for further testing are usually communicated to the laboratory by a medical practitioner.

5.2 Requirements

- (a) All requests for further testing must have appropriate consent from the individual or those authorised to give consent on their behalf.
- (b) All requests must have the reason for the request.
- (c) The laboratory must ensure that each request, authorisation and access for this use is documented, reported and available for internal and external audit and accreditation assessment visits.

6. Release of residual blood spot samples and related information to the New Zealand Police

6.1 Background

- (a) The Ministry of Health and the New Zealand Police entered into an agreement (Memorandum of Understanding relating to the disclosure of newborn blood spot samples and related information) in 2005 regarding the release of residual blood spot samples. The overriding principle in the Memorandum of Understanding (MoU) between the parties states that the blood spot card and information associated with it is collected for health purposes only. Any use of the blood spot card for any non-health related purpose is exceptional. The Police should have recourse to the blood spot cards and associated information only rarely, and as a last resort.
- (b) Residual blood spot samples may be requested in accordance with the MoU where:
 - (i) a body or body part is found and all other avenues of identifying the person are either not practicable or have failed
 - (ii) biological material is available and requires a match to identify a specific person who is deceased or missing, and there is no practical means of making the identification
 - (iii) coronial inquiries which require analysis of samples; or
 - (iv) where (i), (ii) or (iii) above do not apply and the Police have obtained a search warrant in accordance with clause 3.1.2 of the MoU.
- (c) The MoU is available on www.nsu.govt.nz

6.2 Requirements

- (a) Requests from the Police for residual blood spot samples must be in writing to the Ministry for consideration and approval.
- (b) Only the NMSP Director or person acting in this position may authorise and release residual blood spot samples to the Police following approval in writing from the Ministry.
- (c) The laboratory must ensure that each request, authorisation and access is documented and available for internal and external audit and accreditation assessment visits.
- (d) The laboratory must agree arrangements with the Police for the secure collection or transport of residual blood spot samples and information.
- (e) All requests and releases must be reported as per contractual obligations.

7. Release of residual blood spot samples and related information to a New Zealand Coroner

7.1 Background

- (a) Coroner releases are governed by the Coroners Act 2006 (the Act).
- (b) Coroners work closely with the New Zealand Police for criminal and non-criminal inquiries. All Police requests for blood spot samples for criminal inquiries are subject to the Memorandum of Understanding relating to the disclosure of newborn blood spot samples and related information.
- (c) A Coroner, who considers it necessary for the purposes of an inquiry the Coroner has opened under the Act, may request an individual's blood spot sample to be released and/or to be tested for the purposes of that inquiry.
- (d) All requests for an individual's blood spot sample must be authorised by the Chief Coroner.

7.2 For operational purposes, the NMSP Director will authorise the release of residual blood spot samples if the Chief Coroner's request meets with the following requirements.

- (a) The request must be from the Chief Coroner in writing, and include:
 - (i) identification of the inquiry for which the sample is required
 - (ii) the purpose for the request.
- (b) Any requests not meeting these requirements require further communication with the Chief Coroner.
- (c) Any issues or questions regarding requests must be communicated with the legal counsel for the District Health Board and/or the Ministry. Legal sign off to approve the request is then required.
- (d) When the request is approved, the NMSP Director will release the blood spot sample to the Chief Coroner, and either perform the requested test(s) or send the residual blood spot sample to the appropriate facility for testing/evaluation, as directed by the Chief Coroner.
- (e) The laboratory must ensure that each request, authorisation and access is documented and available for internal and external audit and accreditation assessment visits.
- (f) All requests and releases must be reported as per contractual obligations.

8. Release of residual blood spot sample information to mortality review committees

8.1 Background

From time to time, mortality review committees request information on screening results. Release of information about samples to mortality review committees is governed by the New Zealand Public Health and Disability Act 2000: Schedule 5. Provisions applying to mortality review committees include:

‘Chairperson may require person to give information

- (1) If a mortality review committee gives its chairperson, or an agent the committee appoints for the purpose, authority in writing to do so, the chairperson or agent may, by notice in writing to any person, require the person to give the committee information in the person’s possession, or under the person’s control, and relevant to the performance by the committee of any of its functions.
- (2) Examples of the information the chairperson or agent may require are:
 - (a) patient records, clinical advice, and related information
 - (b) answers to questions posed by the chairperson in the notice, and that the person is able to answer
 - (c) information that became known solely as a result of a declared quality assurance activity, within the meaning of Part 6 of the Medical Practitioners Act 1995, or a protected quality assurance activity within the meaning of section 53(1) of the Health Practitioners Competence Assurance Act 2003.
- (3) The person must take all reasonable steps to comply with the notice.’

8.2 Requirements

- (a) The NMSP Director can authorise the release of blood spot sample information to a mortality review committee with the following requirements:
 - (i) the request must be in writing
 - (ii) the request must include the mortality review committee authority provisions
 - (iii) the request must be for existing information.
- (b) Any request by a mortality review committee for the creation of new information (ie, further testing) must be accompanied by appropriate written consent from the person authorised to consent.
- (c) Any issues or questions regarding requests must be communicated with the legal counsel for the District Health Board and/or the Ministry. Legal sign off to approve the request is then required.
- (d) The laboratory must ensure that each request, authorisation and access for this use is documented, reported and available for internal and external audit and accreditation assessment visits.
- (e) All requests and releases must be reported as per contractual obligations.

9. Release of residual blood spot sample and information for research

9.1 Background

- (a) From time to time, there may be requests received by the NMSP for residual blood spots to be included in population research studies. The use of residual blood spots in research is covered by Right 7(10) of the Code of Rights which allows for research that has received the approval of an ethics committee. Any research must also comply with the following provisions.
- (b) The use of residual blood spot samples is subject to the *New Zealand Standard NZS 8135:2009 Non-therapeutic use of human tissue* and the *Guidelines for the Use of Human Tissue for Future Unspecified Research Purposes* (Ministry of Health, 2007). Part 1 of the Guidelines:
 - (1) provides that consent may be given for the use of human tissue in future unspecified research, and part 1 (2) provides that that consent must be distinct from consent to collect the sample.

9.2 General requirements for research application requests

- (a) The proposed research:
 - (i) must be considered an appropriate use of residual blood spot samples and contributes to the public good through increased scientific knowledge
 - (ii) will not use all residual blood from an individual blood spot card.
- (b) Ethics committee approval must be obtained.
- (c) All requests must be reviewed by the NMSP Governance Team.
- (d) Ministry approval must be obtained.
- (e) All applications must follow the research applications pathway (Figure 5).

9.3 Specific requirements

- (a) Residual blood spots collected prior to June 2011 (the formal introduction of this policy) require written consent for research use for each individual blood spot sample from the person authorised to give consent. Ethics committee approval is also required for use in research prior to consideration by the NMSP Governance Team and the Ministry.
- (b) Residual blood spot samples collected from July 2011 require (as a minimum) approval from an ethics committee prior to consideration by the NMSP Governance Team and the Ministry.
- (c) Use of residual blood spot samples and information for research may only occur with the Ministry's written permission.

10. Process to request information and/or residual blood spot samples for research

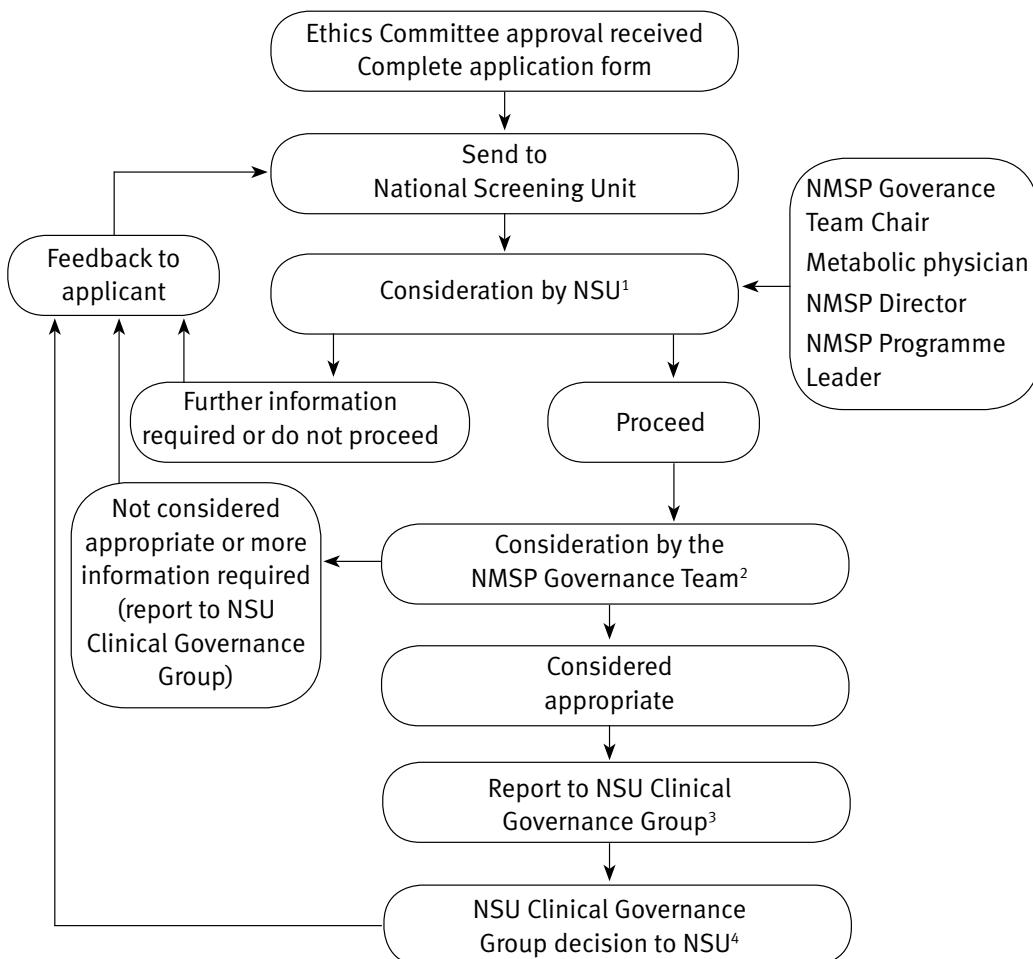
- (a) The Antenatal and Newborn Screening Programmes Application Form must be completed. This form is available on the NSU website www.nsu.govt.nz
- (b) Research requests will be considered as per the research request pathway diagram (Figure 5) and templates below.

- (c) All applications for research purposes must be accompanied by a copy of the ethics committee application and approval.
- (d) Inquiries regarding applications should go to the NMSP Programme Leader, Ministry of Health, as per the application form.

11. Reporting and publications

- (a) A copy of any report prepared using NMSP residual blood spot samples and information is required to be provided in confidence to the NMSP Programme Leader, Ministry of Health, prior to publication.
- (b) Any report or publication using residual blood spot samples and information must acknowledge the Ministry and the NMSP as the source of the data.

Figure 5: Research applications pathway



Notes:

1 Consideration by the NSU

- Completeness of information.
- Considered an appropriate request to continue through to the NMSP Governance Team.

2 Consideration by the NMSP Governance Team

- The NMSP Governance Team will consider applications yearly in the November meeting.
- All applications are to be received by the end of June of each year.

3 Template for NMSP Governance Team discussion

Date:	Comments
Research request	
Members present for discussion	
Conflicts of interest identified and managed (show details)	
Initial discussion	
Research request is robust	
Programme implications (eg resource)	
Risks Benefits Trade offs Public health value (economic, medical)	
Cultural considerations	
Retaining aims and objectives of the programme including community participation and trust	
Logistics	
Consideration of the <i>Ethical Guidelines for Observational Studies</i> (NEAC 2006)	
Operational considerations	
Discussions with other stakeholders if required	
Additional decision-making criteria: <ul style="list-style-type: none"> • Does the request comply with relevant legislation? • Have all appropriate ethical approvals been obtained? • Is the release authorised by individuals/parents/guardians/representatives or does it comply with Right 7(10) of the Code of Consumers Rights? • Does the request concern cards collected after this policy or before? • If prior to this policy, has specific renewed consent been given by the source subject or representative? • Will the research use compromise the primary use of the cards? • Can the costs of retrieval be met by the requestor? 	
Further discussion (show any changes to NMSP Governance Team membership, conflicts and date)	

Recommendation	
Date	
Letter sent to NSU Clinical Governance Group	
Letter sent to applicant (if appropriate)	

4 Recommendations to the NSU Clinical Governance Group are:

1. Endorse research proposal.
2. Require further information.
3. Decline research proposal.

Decision made by NSU Clinical Governance Group	
Date	
Letter sent to NMSP Goverance Team	
Letter sent to applicant	

12. Other requests for release of residual blood spot samples or information about blood spot samples

12.1 Background

From time to time requests for residual blood spot samples and/or related information are received by the NMSP. These requests may include requests for other testing and come from third parties including:

- (i) health providers
- (ii) insurance companies
- (iii) legal representatives
- (iv) other persons.

12.2 Policy

Access to samples by third parties including employers, insurers, legal representatives, other relatives or medical practitioners may only be granted with the written consent of the person authorised to consent, or by a court order.

12.3 Requirements

- (a) All requests for residual blood spot samples and/or related information by third parties must be notified to the NMSP Programme Leader.
- (b) All third party requests must be presented to and handled by the Ministry of Health Legal Team and/or ADHB Legal Team.
- (c) All requests must be approved by the Ministry of Health.
- (d) Any requests accepted, must include appropriate written consent from the individual or those authorised to give consent on their behalf or a court order.
- (e) When the request is approved, the NMSP Director may either perform the requested test(s) or send the residual blood spot sample to the appropriate facility for testing/evaluation.
- (f) The release must be reported in the annual programme report as per contractual obligations.
- (g) The laboratory must ensure that each request, authorisation and access for this use is documented, reported and available for internal and external audit and accreditation assessment visits.

13. Storage of residual blood spot samples

13.1 Purpose

This policy outlines the requirements for the secure retention of residual blood spot samples after screening. Secure retention requires that access to stored blood spot samples is monitored and controlled for authorised personnel only and all access is logged.

13.2 Background

Residual blood spot samples are stored indefinitely in secure storage for possible future uses, unless parents/guardians or individuals do not consent to storage and request their return.

- 13.3 The Ministry of Health is the custodian of residual blood spot samples. Storage is currently contracted to the Auckland District Health Board.

13.4 Storage requirements

Residual blood spot samples must be stored:

- (a) in an ambient temperature environment
- (b) in a secured locked area with safeguards to prevent unauthorised use, disclosure, loss or other misuse
- (c) to allow and facilitate authorised retrieval.

13.5 Security and access requirements

The laboratory must ensure the following.

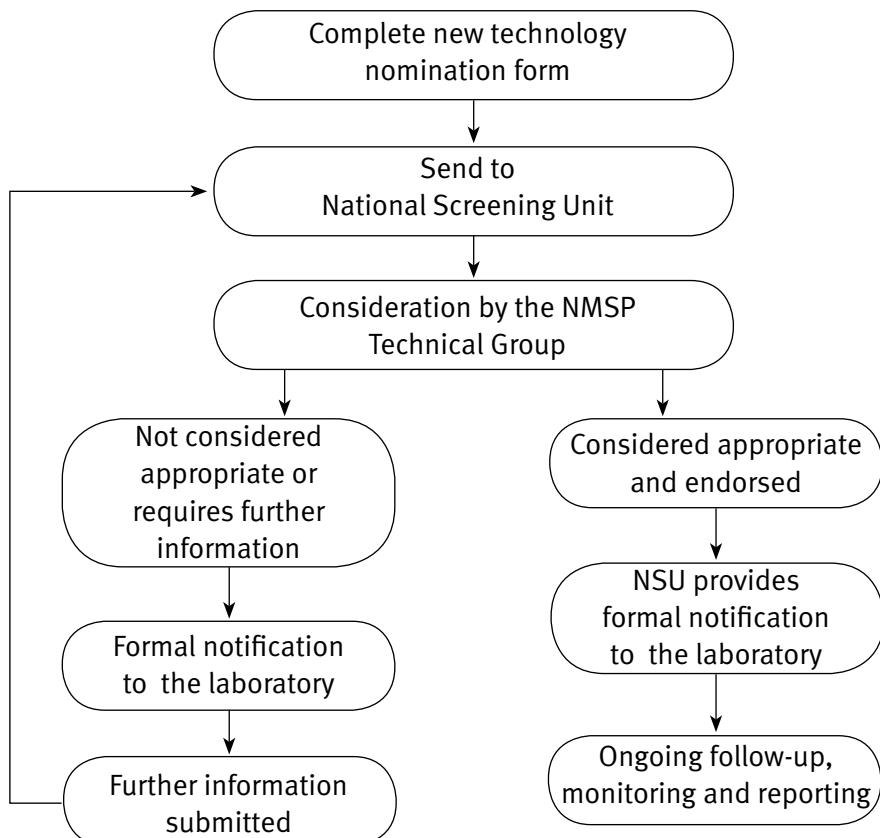
- (a) Access to stored residual blood spots and related information is limited to the:
 - (i) NMSP director
 - (ii) NMSP LMC educator
 - (iii) NMSP lead scientist.
- (b) One of the above personnel approves a request for release of a residual blood spot sample.
- (c) Release of a residual blood spot sample is for an authorised primary or secondary use as detailed in the above sections.
- (d) Each request, authorisation and access to the secure storage facility is documented, reported and available for internal and external audit and accreditation assessment visits.
- (e) The written agreement with the storage facility must be agreed with the Ministry and detail:
 - (i) storage requirements
 - (ii) safety and security of records including fire and flood protection
 - (iii) access restrictions
 - (iv) access process
 - (v) auditable record requirements
 - (vi) timeliness of authorised access
 - (vii) monitoring and reporting requirements which include a signout and tracking protocol.

Section Eight – New Technologies

1. New technologies

- 1.1 Ongoing evaluation of the NMSP should occur on a regular basis. This includes new technologies that have been developed for screening. They may include new equipment, new methods, new assays, significant changes to information systems or new ways of testing.
- 1.2 New technology will be considered as per the following pathway. The new technology nomination form can be found at Appendix D.

Figure 6: New technology pathway



Section Nine – Changes to the Disorder Panel

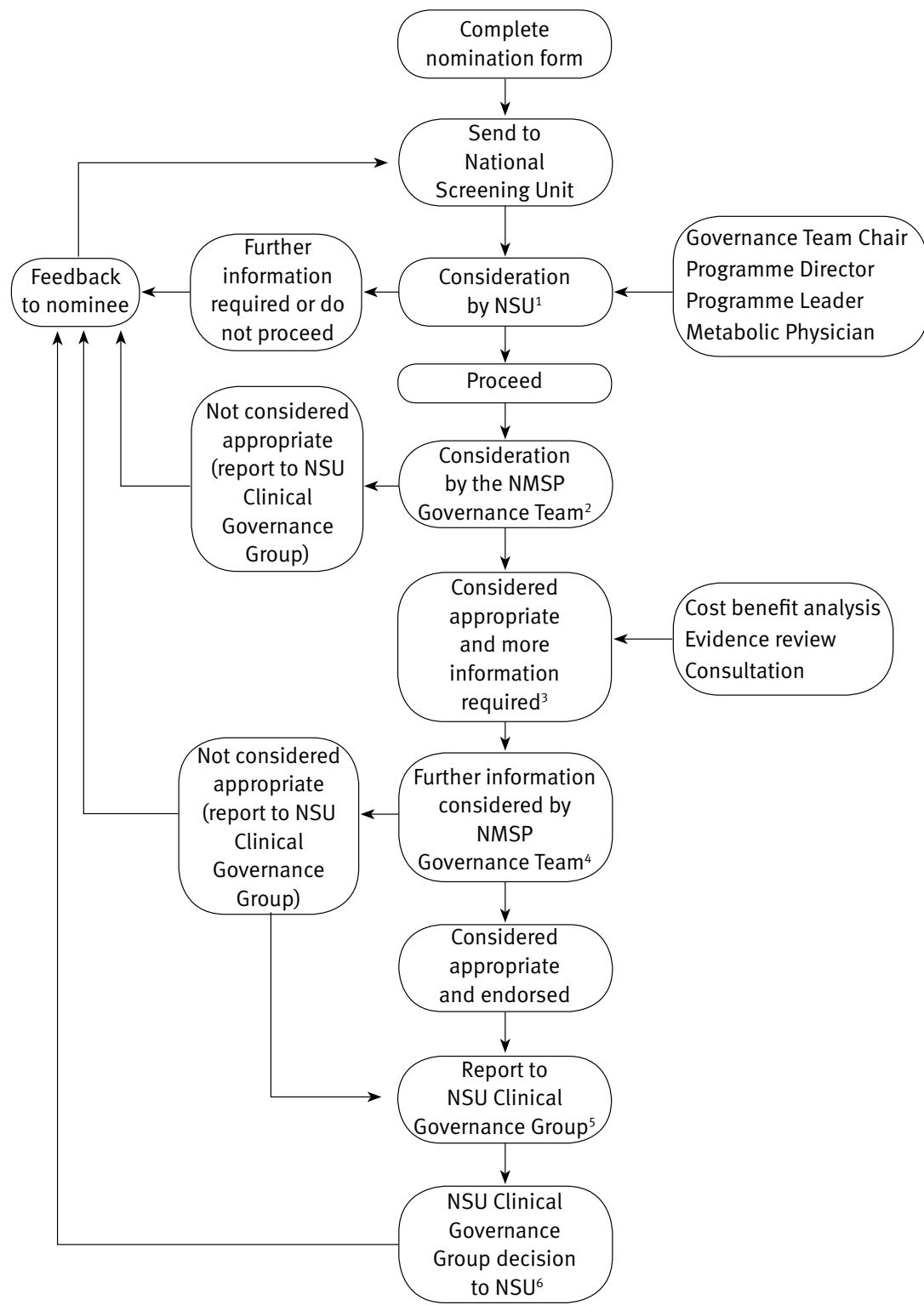
1. Changes to the disorder panel

- 1.1 From time to time, new disorders will be considered for inclusion in the screening programme and, through evaluation, current disorders may be considered for removal.

2. New disorder consideration

- 2.1 New disorders will be considered as per the following pathway using the evaluation template as below. The nomination form can be found at Appendix E.

Figure 7: Consideration for screening of a disorder in the newborn period as part of the Newborn Metabolic Screening Programme



1 Consideration by the NSU

- Completeness of information.
- Considered an appropriate request to continue through to the NMSP Governance Team.

2 Consideration by the NMSP Governance Team

- The NMSP Governance Team will consider applications yearly in the November meeting.
- All applications are to be received by the end of June of each year.

3 Further information potentially required as part of the NMSP Governance Team consideration

- Cost benefit analysis.
- Evidence review.
- Stakeholder consultation including Māori.
- Literature review.
- International review.

4 Template for NMSP Governance Team discussion

Date	Comments	Grade of evidence
<p>The condition should be an important health problem potentially leading to significant morbidity or mortality, and for which early identification appears likely to be of benefit to the infant. In some disorders, a benefit for the family may be important, where the condition is untreatable and may lead to early mortality, but where a definitive diagnosis might be aided by the performance of the screening test.</p>		
Disorder nominated		
NMSP Governance Team members present for discussion		
Conflicts of interest identified and managed (show details)		
Initial discussion		
Requests for further information May include: Cost benefit analysis Evidence review Surveys/consultations		
Discussions/reports from other stakeholders (eg Paediatric Society, practitioners)		
Further discussion (show any changes to NMSP Governance Team membership, conflicts and date)		

	Comments	Grade of evidence
Clinical / population characteristics <ul style="list-style-type: none">• incidence• presentation in newborn period• burden of disease• natural history• equity issues• cultural issues		
The proposed test <i>should be simple, safe, reliable, validated</i>		
Screening test <ul style="list-style-type: none">• sensitivity• specificity• costs• ? multiplex• what else might be detected• laboratory capability• false positive rate• false negative rate		
Trade-offs		
Integration into existing programme		
Diagnosis and follow-up <ul style="list-style-type: none">• availability and cost of diagnostics• reliability• efficacy• types of diagnostics• ? part of screening		

The treatment: *there should be established treatment or intervention which has the potential to prevent or ameliorate the clinical consequences of the disease.*

	Comments	Grade
Treatment: • interventions • which patients need treatment • efficacy • urgency – evidence for benefit or likely benefit • availability • costs (direct and infrastructure) • possible harms • prevention of mortality		
What else will be found?		
Ethics		
Harms/benefits • false positives • other		
Screening criteria <i>See other documentation</i>		
Test availability		
Impact on the programme		
National considerations		
International considerations		
Literature reviews		
Other ways of spending \$		

Grades of evidence:

A: At least one high-quality meta-analysis, systematic review, or RCT directly applicable to the target population *or* a systematic review of RCTs *or* a body of evidence consisting principally of well-conducted studies directly applicable to the target population and demonstrating overall consistency of results.

B: A body of evidence including high-quality case-control or cohort studies with a very low risk of confounding, bias or chance, a high probability that the relationship is causal, directly applicable to the target population and demonstrating overall consistency of results.

C: A body of evidence including well-conducted case-control or cohort studies with a low risk of confounding, bias, or chance and a moderate probability that the relationship is causal, directly applicable to the target population and demonstrating overall consistency of results.

D: Non-analytical studies, cohort or case-control studies with a significant risk of confounding, bias or chance, case reports, or case series. Expert opinion.

E: No evidence available.

Adapted from: Harbour R, Miller J. A new system for grading recommendations in evidence based guidelines *British Medical Journal*. 2001;323(7308):334–6.

Recommendation	
Date	
Letter sent to NSU Clinical Board	
Letter sent to nominee (if appropriate)	

5 Recommendations to the NSU Clinical Governance Group

- Recommend universal screening
- Recommend targeted screening
- Recommend pilot study
- Recommend against screening

Decision made by clinical board	
Date	
Letter sent to NMSP Governance Team	
Letter sent to nominee	

6 Clinical Governance Group decision

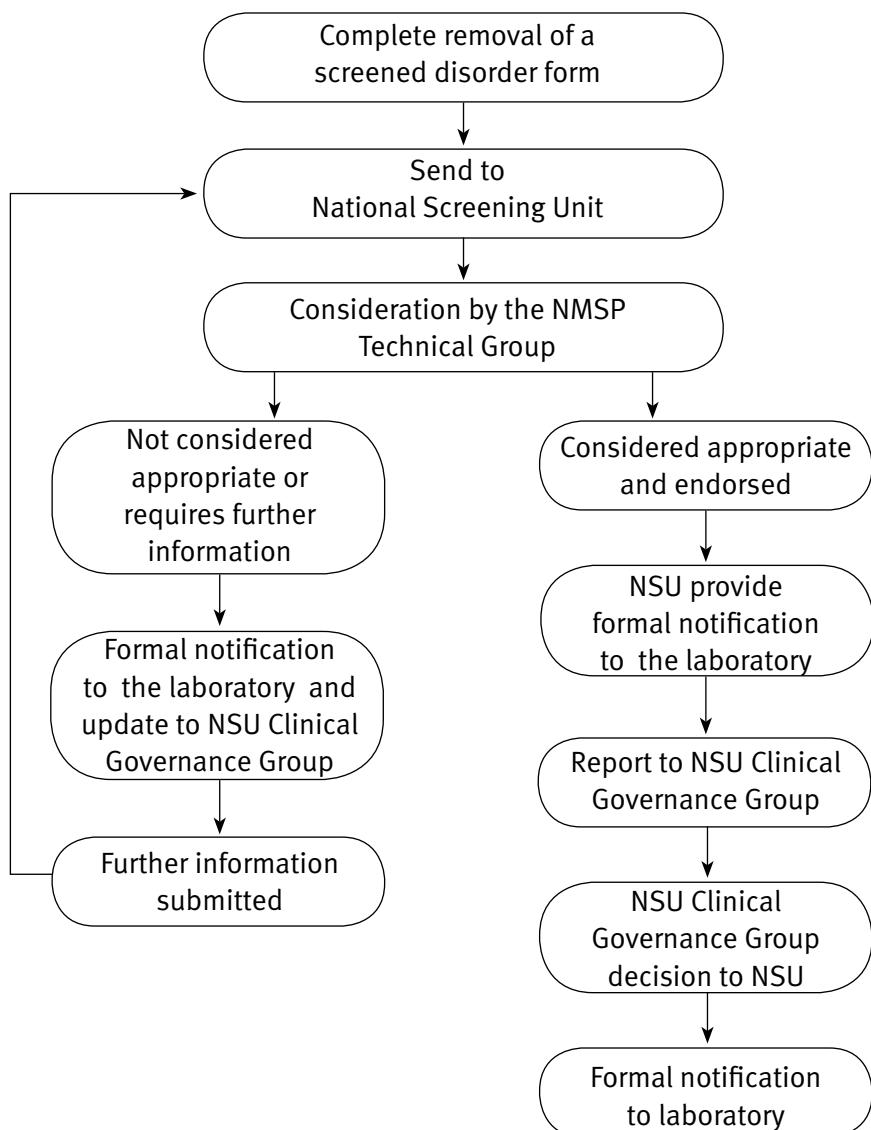
If the clinical governance group endorses the introduction of a new disorder the project moves into the next phase. This may include:

- pilot studies
- development of treatment protocols
- documentation of laboratory requirements
- documentation of treatment options and funding
- consultation with treatment providers
- consultation with other providers
- development of referral pathways.

3. Removal of a screened disorder

Removal of a screened disorder will be considered as per the following pathway.

Figure 8: Removal of a screened disorder



References

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Appendix A: Disorders in the Newborn Metabolic Screening Programme

1. Disorders in the Newborn Metabolic Screening Programme

- (a) Amino acid disorders:
 - (i) 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
 - (ii) 3-Methylcrotonyl-CoA carboxylase deficiency
 - (iii) Argininosuccinic aciduria (argininosuccinate lyase deficiency)
 - (iv) Beta-ketothiolase deficiency (mitochondrial acetoacetyl-CoA thiolase deficiency)
 - (v) Citrullinaemia (argininosuccinate synthetase deficiency, citrin deficiency)
 - (vi) Glutaric aciduria type I (glutaryl-CoA dehydrogenase deficiency)
 - (vii) Homocystinuria (cystathione beta-synthase deficiency)
 - (viii) Isovaleric aciduria (isovaleryl-CoA dehydrogenase deficiency)
 - (ix) Maple Syrup Urine Disease (MSUD)
 - (x) Methylmalonic acidurias (mutase deficiency, CblA, CblB, CblC, CblD defects)
 - (xi) Multiple carboxylase deficiency (holocarboxylase synthetase deficiency)
 - (xii) Phenylketonuria (PKU)
 - (xiii) Propionic aciduria (propionyl-CoA carboxylase deficiency)
 - (xiv) Tyrosinaemia (fumaryl acetoacetate deficiency, tyrosine aminotransferase deficiency).
- (b) Fatty acid oxidation disorders:
 - (i) CACT (carnitine acylcarnitine translocase deficiency)
 - (ii) Carnitine transporter defect
 - (iii) CPT-I (carnitine palmitoyltransferase-I deficiency)
 - (iv) CPT-II (carnitine palmitoyltransferase-II deficiency)
 - (v) LCHAD (3-hydroxy long-chain acyl-CoA dehydrogenase deficiency)
 - (vi) TFP (trifunctional protein deficiency)
 - (vii) MADD (multiple acyl-CoA dehydrogenase deficiency)
 - (viii) MCAD (medium-chain acyl-CoA dehydrogenase deficiency)
 - (ix) VLCAD (very-long-chain acyl-CoA dehydrogenase deficiency).
- (c) Other disorders:
 - (i) Congenital hypothyroidism
 - (ii) Congenital adrenal hyperplasia
 - (iii) Cystic fibrosis
 - (iv) Biotinidase deficiency
 - (v) Galactosaemia.

Appendix B: Testing timeframes and cut-offs

Condition	Disorder definition	Non-urgent recall	Urgent recall
Galactosemia	<p>Transferase deficiency: undetectable galactose-1-phosphate uridyl transferase (Gal-1-PUT) activity in red blood cells and elevated galactose-1-phosphate + galactose.</p> <p>Galactokinase deficiency: elevated galactose and normal galactose-1-phosphate and Gal-1-PUT activity.</p> <p>Urindine diphosphate-galactose 4 epimerase deficiency: galactose-1-phosphate is significantly elevated but Gal-1-PUT is normal.</p>	<p>Gal 1.5 mmol/L blood For NICU babies Gal 0.80 mmol/L blood</p>	<p>Gal 2.0 mmol/L blood For NICU babies Gal 1.0 mmol/L blood Lower limits may be used if gal-1-PUT is absent</p>
Congenital hypothyroidism	Elevation of thyroid stimulating hormone (TSH) and evidence of ectopic thyroid tissue or a hemithyroid or athyreosis or a patient with a documented persistent elevation of TSH and the institution of treatment with thyroxine.	TSH 15–49 mIU/L blood	TSH 50 mIU/L blood or higher
Congenital adrenal hyperplasia	Persistent elevation of 17-hydroxyprogesterone associated with confirmatory clinical features (eg abnormal electrolytes).	17 OHP 23–50 mmol/L blood for term babies	17 OHP 50 mmol/L blood or higher
Biotinidase deficiency	Confirmed deficiency of biotinidase enzyme.	One action only – written request for further sample when no enzyme present.	
Cystic fibrosis	Either two mutations associated with classical CF or a positive sweat test with sweat chloride of equal to or greater than 35 mmol/L	One action only – referral for specialist assessment when HRM indicates one or two CF mutations present.	
Aminoacid breakdown and fatty acid oxidation disorders	Enzyme deficiency as on website list of screened conditions as indicated by confirmatory biochemical and clinical observations.	Determination of urgent vs non-urgent recall is by use of individual marker analytes and ratios, and by clinical assessment of individual cases.	

Appendix C: Adverse Event Template

Adverse event/incident investigation and notification plan:

(State issue/complaint)

1. Description of incident or area of concern (include dates)
2. How was incident discovered/notified?
3. Which other organisations are involved?
4. Why does incident warrant further investigation?
5. Who is involved in investigation?
6. Who else needs to be notified of incident?¹⁰ (by whom and what/why?)
7. Investigation Plan (update ongoing – who is doing what?)

Date	Action	By who	Status

¹⁰ Notification required to NMSP Programme Leader

Appendix D: Nomination Form for New Technology

Nomination form for new technology for existing screened disorder		
Name	Organisation	Date
Disorder/information system		
Current screening method/ information system		
Proposed new screening method/ information system		

Details of proposed technology including: • benefits • risks • harms • clinical validity • longevity • back-up/ contingency	
Costings	(Attach detailed costings as appendix.)
Programme impacts	
Other jurisdiction experiences	

Please include the following as appendices:

- a contingency plan
- options paper (consideration of other solutions)
- implementation plan
- detailed costings.

Key references (Specific citations – limit to 15)	
1	
2	
3	
4	
5	
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11	
12	
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14	
15	
Submission check list	Submit nominations to:
Cover letter by proponent	Programme Leader Newborn Metabolic Screening Programme National Screening Unit Ministry of Health Private Bag 92522 Wellesley Street Auckland
Nomination form	
Copy of references listed on this form	
Options paper	
Contingency plan	
Implementation plan	
Detailed costings	
Contact Information (proponent)	

Appendix E: Nomination Form for Proposed New Screened Disorder

Nomination form for proposed disorder							
Name of proponent		(Organisation, if relevant)			Date		
Disorder							
Type of disorder							
Screening method							
Treatment strategy or intervention							
Disorder	Comment		Gene		Locus		OMIM or other names for disorder

*Note: Please reference each statement, listing references below (p.2)

	(Determined by what method(s): pilot screening or clinical identification?)
	(Relevance of the timing of newborn screening to onset of clinical manifestations)
	(Morbidity, disability, mortality, what spectrum of severity)
	(Cultural issues identified)
Comment	
	(High volume method, platform)
	(Dried blood spot, physical or physiologic assessment, other)
	(Location, duration, size, preliminary results of past/ongoing pilot study for clinical validation)
	(Sensitivity, specificity, detection rate, positive predictive value, false positive rate)
	(Reliability, availability, DNA analysis, cost)
	(False positives, carrier detection, invasiveness of method, other. Detection or suggestion of other disorders)

Treatment	Comment
Modality	(Drug(s), diet, replacement therapy, transplant, other)
Urgency	(How soon after birth treatment needs to be initiated to be effective)
Efficacy (Benefits)	(Extent of prevention of mortality, morbidity, disability. Treatment limitations, such as difficulty with acceptance or adherence)
Availability	(Any limits of availability)
Potential harms of treatment	(Potential medical or other ill effects from treatment)

Key references (Specific citations – limit to 15)		
1		
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Submission check list		Submit nominations to:
	Cover letter by proponent	Programme Leader Newborn Metabolic Screening Programme Ministry of Health Private Bag 92522 Wellesley Street Auckland
	Nomination form	
	Copy of references listed on this form	
	Formal conflict of interest statement by proponent	
Contact information (proponent)		

Appendix F: Data Access Agreement

1. NMSP data access agreement form

Name: _____

Address: _____

Institution: _____

I agree to the following conditions of access to the Newborn Metabolic Screening Programme (NMSP) data.

- (a) The data will be kept/stored for a minimum necessary period.
- (b) Data provided will be kept and maintained in a secure manner. At a minimum this should include data being kept in an encrypted, password protected directory on a computer that itself has password protected access.
- (c) Information will not be given/sold or transmitted to anyone else not party to this agreement, other than those under direct supervision from myself for example a biostatistician or lead clinician.
- (d) The data will not be linked with other data in any way that would make individuals identifiable.
- (e) Before public release or publication of data I agree to provide the NMSP with a copy of articles and abstracts for review prior to submission for publication or presentation at meetings.

Applicant signature and date: _____

Witnessed by and date: _____